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SWISS SOCIETY OF PAEDIATRICS

ABSTRACTS OF THE ANNUAL MEETING 2025 (BERN, MAY 22/23, 2025)

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ORAL COMMUNICATIONS

OC 1

The effects of Nirsevimab against respiratory syncytial virus on hospital admission numbers in Swiss Children's Hospitals, the RSVEpiCH study group

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Background: Since 2021, the RSV EpiCH project has aimed to provide representative nationwide surveillance of respiratory syncytial virus (RSV) infections in children in Switzerland. Before the COVID-19 pandemic, Switzerland experienced a stable biennial cycle of alternating strong and weak RSV winter epidemics in children. The RSV EpiCH project allowed to document the disruption of this consistent pattern during the COVID-19 pandemic with stringent implementation of non-pharmaceutical interventions. We have since observed two strong RSV seasons (2022/2023 and 2023/2024). Here, we present findings from the ongoing surveillance of RSV in children in Switzerland during the first winter season (2024/2025) in which Nirsevimab was generally available for children born after March 2024.

Methods: The RSV EpiCH project prospectively collects weekly aggregated data on the detection of RSV in children from 21 of 29 paediatric acute care hospitals in Switzerland. These hospitals represent >90% of available paediatric beds in Switzerland. All sites report the number of children with detection of RSV in three age groups, <1 year old, 1 to 2 years old, and older than 2 years. We compared total patient numbers and age distribution across four time periods: 1/2021–6/2022 (2021/2022 pandemic period), 7/2022–6/2023 (winter 2022/2023), 7/2023–6/2024 (winter 2023/2024), and 7/2024-1/2025 (winter 2024/2025). Incidence rates were calculated using resident population in Switzerland in the respective age-groups.

Results: From 4th January 2021 to 27th January 2025, RSV was detected in 16'058 children. 5'160 (incidence rate 2.3 per 1000 children-years) in the 2021/2022 pandemic period, 5'242 (3.5 per 1000 children-years) in the winter 2022/2023, 4'672 (3.1 per 1000 children-years) in winter 2023/2024, and 984 (1.1 per 1000 children-years) in winter 2024/2025. In infants incidence rates were 23 per 1000 infants in the pandemic period, 41 per 1000 infants in winter 2022/2023, 35 per 1000 infants in winter 2023/2024, and so far 9 per 1000 infants in winter 2024/2025. In parallel, the proportion of infants amongst all RSV cases decreased from 57–61% in previous seasons to 43%, so far, in winter 2024/2025.

Conclusion: In the first winter season with the general availability of Nirsevimab, preliminary data from paediatric acute care hospitals indicate that three to four times fewer infants with RSV infection are being seen in hospital. This has also led to a reduction in overall case numbers.

OC 2

Antibiotic exposure and adverse long-term health outcomes in children

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Background: Antibiotics are among the most commonly prescribed drugs during pregnancy and childhood. While essential in treating bacterial infections, their use has raised concerns about potential long-term health risks.

Methods: Two separate systematic reviews using PRISMA guidelines were done to identify original studies investigating the associations between antibiotic exposure exposure during pregnancy and early life with adverse long-term health outcomes in children.

Results: Prenatal antibiotic exposure: A total of 158 studies involving 21,943,763 children were analyzed, reporting 23 outcomes. Significant associations with prenatal antibiotic exposure were observed for atopic dermatitis (OR 1.27, 95% CI 1.06–1.52, p = 0.01), food allergies (OR 1.25, 95% CI 1.09–1.44, p <0.01), allergic rhinoconjunctivitis (OR 1.16, 95% CI 1.15–1.17, p <0.01), wheezing (OR 1.39, 95% CI 1.14–1.69, p <0.01), asthma (OR 1.36, 95% CI 1.24–1.50, p <0.01), obesity (OR 1.36, 95% CI 1.12–1.64, p <0.01), cerebral palsy (OR 1.25, 95% CI 1.10–1.43, p <0.01), epilepsy or febrile seizures (OR 1.16, 95% CI 1.08–1.24, p <0.01), and cancer (OR 1.13, 95% CI 1.01–1.26, p = 0.04). Antibiotic exposure during early-life: A total of 160 studies involving 22,103,129 children were analyzed, reporting 21 outcomes. Antibiotic exposure was significantly associated with atopic dermatitis (OR 1.40, 95% CI 1.30–1.52, p <0.01), allergic symptoms (OR 1.93, 95% CI 1.66–2.26, p <0.01), food allergies (OR 1.35, 95% CI 1.20–1.52, p <0.01), allergic rhinoconjunctivitis (OR 1.66, 95% CI 1.51–1.83, p <0.01), wheezing (OR 1.81, 95% CI 1.65–1.97, p <0.01), asthma (OR 1.96, 95% CI 1.76–2.17, p <0.01), obesity (OR 1.21, 95% CI 1.05–1.40, p <0.01), juvenile idiopathic arthritis (OR 1.74, 95% CI 1.21–2.52, p <0.01), psoriasis (OR 1.75, 95% CI 1.44–2.11, p <0.01), autism spectrum disorders (OR 1.19, 95% CI 1.04–1.36, p = 0.01), and neurodevelopmental disorders (OR 1.29, 95% CI 1.09–1.53, p <0.01). Dose-response relationships and stronger effects with broad-spectrum antibiotics were frequently observed. No associations were found for allergic sensitization, inflammatory bowel disease, type 1 diabetes, and attention deficit hyperactivity disorder.

Conclusion: While these studies cannot establish causality, the findings underscore the importance of antibiotic stewardship

during pregnancy and childhood to mitigate potential long-term health risks.

OC 3

Epidemiology, impact of early treatment and predictors of adverse outcomes in congenital CMV: a Swiss nationwide prospective study

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Background: Congenital cytomegalovirus (cCMV) is the most common congenital infection, with approximately 17-20% of infected neonates developing permanent sequelae. Early risk stratification remains challenging but critical for therapeutic decision-making. This study aimed to define epidemiology, identify early predictors of adverse neurological outcomes, and evaluate how early antiviral treatment impacts outcomes in a nationwide cohort.

Methods: We conducted a prospective multicenter observational study between 2017-2024, analyzing birth and one-year follow-up data of 209 neonates with confirmed cCMV in Switzerland.

Results: cCMV diagnosis was confirmed within three weeks of life in 87% of cases, primarily with urinary polymerase chain reaction (79%). Most common findings at birth included microcephaly (35%), intrauterine growth restriction (34%), and neuroimaging abnormalities (36%). Antiviral treatment was administered to 36% of patients; it was initiated early in 56 patients (within first month of life), and late in 19 patient (after 1 month of life). Multivariate analysis identified hepatosplenomegaly (OR 13.2, 95%CI 2.3-75.2) and hypotonia at birth (OR 6.6, 95%CI 1.1-41.1) as independent predictors of late-onset sensorineural hearing loss (SNHL). At one-year follow-up assessment, early treatment was associated with significantly lower rates of neurodevelopmental disorders compared to late treatment (23% vs 50% p = 0.04), while there was no significant difference compared to untreated patients (23% vs 10%, p = 0.2). SNHL at one-year follow up showed a similar trend, though not statistically significant (delayed vs early: 47% vs 32%, p = 0.5).

Conclusions: This first Swiss nationwide study confirms the previously reported association of hepatosplenomegaly with late-onset hearing loss and identifies hypotonia as a new independent predictor. The data also confirm the crucial role of early antiviral treatment, particularly for neurodevelopmental outcomes, emphasizing the importance of prompt cCMV diagnosis and treatment when indicated.

OC 4

Pediatric measles vaccine survey response and uptake by nationality – a Swiss national survey study

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Background: Accurate monitoring and uptake of measles vaccination is key as the disease continues to spread throughout the world due to inadequate immunization coverage.

Methods: We used the Swiss national vaccination survey to explore the association between nationality and a) survey response and b) measles vaccine uptake. The data were obtained from the Swiss National vaccination survey (SNVCS) which included 2020 – 2022 measles vaccination data for 24 - 35-month-old children selected via simple random sampling from all 26 cantons of Switzerland.

Results: Non-Swiss caregivers were significantly less likely to respond to the survey than Swiss nationals (aOR 0.51, 95%CI 0.47 – 0.57). This was specifically true for Eastern Europeans (aOR 0.46, 95%CI 0.39 – 0.55) and Africans (aOR 0.66, 95%CI 0.51 – 0.87). When aggregated, no clear association between having a non-Swiss nationality and MCV2 uptake was found (aOR 0.79, 95%CI 0.57 – 1.09; p 0.15). However when further disaggregated by nationality, Eastern European children had lower odds of MCV2 uptake than Swiss nationals (aOR 0.47, 95%CI 0.28 – 0.80).

Conclusions: Children of non-Swiss nationality are insufficiently represented by the SNVCS. Those of Eastern European nationalities were not only at particular risk of underrepresentation but also undervaccination. Strategies to better monitor vaccination coverage of non-Swiss nationals are needed. Vaccination campaigns tailored to specific, undervaccinated groups are required to reach herd immunity.

OC 5

Transfer of bacteria from mothers to infants through breast milk

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There is a critical early window during infancy for establishing the intestinal microbiota. Increasing evidence shows that breast milk (BM) harbours its own distinct microbiota. However, the extent of its contribution to the infant intestinal microbiota remains unclear. A systematic search was done to identify original studies investigating the transfer of bacteria from mothers to the infant's intestine through BM in the first two years of life. We identified 56 studies investigating a total of 2,509 children, 3,481 BM samples and 4,635 stool samples. Many studies reported a higher bacterial diversity of the BM microbiota compared with the infant stool microbiota. Most studies found a higher overlap of bacteria between BM and infant stool in mother-infant pairs compared with unrelated mother-infant pairs. It was estimated that BM bacteria contribute anywhere from 1 to 68% of the infant intestinal bacteria. This large variation is explained by a high degree of methodological heterogeneity between studies, both for microbiota analysis and estimation of overlapping bacteria, including different taxonomic levels analysed. Several studies reported that the overlap between bacteria in BM and infant stool was higher during the first week of life compared with later time points. Genera that were most frequently reported to overlap include Bifidobacterium, Streptococcus, Staphylococcus, Lactobacillus, Veillonella, Escherichia/Shigella and Bacteroides. In total, identical strains were isolated from BM and infant stool for 25 bacterial species. The first weeks of life are critical for shaping the infant intestinal microbiota, but the role of BM as a source of microbial transmission requires further investigation. The relationship between

BM and infant stool microbiota remains poorly understood, hindered by methodological variability and the risk of cross-contamination during BM collection. Future research should focus on clarifying the origins of the BM microbiota and quantifying its contribution to the infant intestinal microbiota.

OC 6

Assessing guideline adherence and child abuse evaluation in infants with fractures: a retrospective quality control study

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Aims: To improve child abuse detection and to standardize the assessment of infants with fractures, the University Children's Hospital Zurich implemented a guideline in February 2021. The guideline demands mandatory involvement of the child protection team (CPT) for infants with fractures, except in cases of birth traumas or specific fractures in infants older than six months with a compatible trauma history and no suspect of child abuse neither from the clinical examination in the hospital nor from the patient's pediatrician or family doctor. The aim of this study was to assess adherence to this guideline and to evaluate changes in patient management before and after the guideline implementation.

Methods: This retrospective single-center quality control study included infants younger than 12 months diagnosed with fractures at the emergency department (ED) between February 2021 and August 2022. Exclusions included children with a bone disease and families who declined data use. The primary outcome was the overall adherence to the guideline. Secondary outcomes focused on changes in the frequency of consultation of the CPT, skeletal survey rates, and child abuse detection, before and after guideline implementation.

Results: Among 9523 infant ED visits, 87 involved fractures (0.9%). After excluding 26 cases, 61 patients were included in the analysis. The median age was seven months, 46% were females, and 51% received outpatient care. Fracture types included skull (54%), lower leg (16%), femur (8%), clavicle (7%), forearm (7%), and humerus (7%). The overall guideline adherence rate was 39%, with the most common deviation (68% of cases) being the omission of telephone consultations with the patient's pediatrician. Adherence rates were similar across age groups but significantly higher in inpatient care (53%) compared to outpatient care (26%). CPT involvement increased to 54%, twice the rate observed before guideline implementation. However, skeletal survey rates decreased, and the child abuse detection rate remained unchanged.

Conclusions: Overall adherence to the guideline was poor, emphasizing the need for continuous training of clinicians to raise awareness regarding the differential diagnosis of child abuse. Although the guideline's implementation resulted in a doubled rate of CPT consultation, further improvements are still needed. In particular, outpatient care exhibited low guideline adherence, signaling an area that requires focused attention.

OC 7

Use of artificial intelligence to guide untrained individuals performing self-ultrasound scans of the knee joint.

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Background: Recurrent joint bleeds occur frequently in persons with haemophilia, often resulting in arthropathy. The value of ultrasonography (US) in detecting bleeds has been demonstrated; however, access to proficient US operators is limited. This innovative solution proposes the integration of real-time artificial intelligence (AI) assistance, which could empower less experienced physicians and even untrained patients to perform accurate US examinations to acquire US frames of diagnostic quality.

Aims: To investigate the performance of the AI solution in assisting untrained individuals with the scanning procedure, to acquire US frames suitable for synovial recess distension detection, indicating a joint bleed.

Methods: An AI algorithm was developed to provide real-time assessments of US frames collected during the scanning process. Two separate studies (Test I and Test II) were conducted to collect US data. We trained the algorithm with scans of 30 participants (Test I) and validated it on US scans of 6 participants (Test II); (Figure 1). Participants were non-haemophilic and untrained in performing US. Prior to the scan, participants were provided with a video on how to perform a self-scan of the knee. During the scan, they were provided with real-time feedback from the AI algorithm (via a mobile application), to assist finding the correct location and capturing diagnostic-quality US images. To enable algorithm training and validation, images were labelled by a medical expert, based on a verified labelling protocol.

Results: A to tal of 27,465 (Test I) and 3,105 (Test II) images were acquired via 30 and 6 scans, respectively. Of these, 5,493 and 621 images were labelled by a medical expert. The algorithm correctly classified 449/621 images (72%) from Test II as usable/not usable, demonstrating a sensitivity of 76%, and a specificity of 71%.

Conclusion(s): These data support the feasibility of using real-time AI-assisted software to assist untrained patients performing US self-examinations to acquire diagnostic-quality images.

OC 8

Estimating the Prevalence of Life-limiting Conditions in Children in Switzerland

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Background: Palliative care for children in Switzerland is underdeveloped, with only one of 26 cantons having a publicly funded paediatric palliative care program. Paediatric palliative care focuses on the needs of the child and family rather than specific diagnoses, but since policymakers require data to understand the demand for such services at any given time, the

Federal Office of Public Health funded this study. Previous studies using catalogues of life-limiting conditions (LLC) have indicated a rising prevalence of affected children, yet with notable variation in numbers.

Aim: This study estimates the prevalence of LLC in Swiss children to aid in planning future palliative care services.

Methods: We analysed hospital data (2018-2022) from four university children's hospitals and one regional hospital, covering five cantons and approximately 51% of the Swiss population, using a catalogue of LLC developed by Fraser et al. (2021) for the United Kingdom (UK). Epidemiological data from Switzerland and the UK were compared using national statistical tools.

Results: The prevalence of LLC in children increased from 33.5/10,000 in 2018 to 41.5/10,000 in 2022, equivalent to 7283 children with LLC in Switzerland in 2022. Children aged 0-1 had the highest prevalence, rising from 174.3/10000 in 2018 to 243.5/10000 in 2022. Congenital anomalies were the most prevalent diagnoses, followed by perinatal and neurological conditions. Although Switzerland and the UK share similarities in overall life expectancy, child mortality and fertility rates, ethnic subgroups are different, which may be important for the spectrum of diagnoses in paediatric palliative care.

Discussion: This is the first study estimating the prevalence of LLC in Switzerland and it confirms a rising prevalence, particularly among children aged 0-1. It highlights the need for prioritized development of paediatric palliative care services in this group emphasizing that these services should be a genuinely paediatric domain.

OC 9

Participation in Everyday Life – Experiences of Children with Cerebral Palsy and their Families

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Introduction: Children with cerebral palsy (CP) often experience restrictions in their participation in everyday life activities. Participation is influenced by environmental and personal factors and is associated with quality of life and general well-being. There is little knowledge about the participation of children with CP in Switzerland and their whole family. This qualitative study aimed to identify the key barriers and facilitators to participation experienced by children with CP living in Switzerland and their families.

Participants and methods: Semi-structured interviews were conducted with 16 families of children with CP from different language regions in Switzerland. The families included exhibited various characteristics (e.g., number and age of children, parents' work activities or socio-economic status). The children with CP were between 2 and 18 years old and showed a wide variation in CP severity. Family members participated in an interview with the whole family and, if they agreed, in an individual follow-up interview. We recorded and transcribed all interviews and translated those conducted in French or Italian into German. We used thematic and content analysis to evaluate the data from the family and follow-up interviews.

Results: Preliminary results show four main themes that influence the participation of the child with CP: (1) social support, (2) assistive devices, (3) behaviour of the child with CP, and (4) societal attitudes. The families identified various personal and environmental barriers and facilitators to the participation of the children with CP and shared insights into their daily lives. They report about different activities in their everyday life, such as at home, at school, in therapies, as well as holiday-specific activities or special events in the community. A similar pattern is described for the participation of the family as a whole. Important factors are the burden of care perceived and experienced by the parents, support from institutions, assistants, friends and family or the infrastructure provided. The analysis is still ongoing, and results may slightly change.

Conclusion: This study provides insights into the lived experiences of children with CP and their families, highlighting the barriers and facilitators to participation in everyday activities. The findings will facilitate understanding the perspective of children with CP and their families and foster their participation and engagement in everyday life.

OC 10

Bronchial carcinoid tumor in children: A case report

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Background/Introduction: Bronchial carcinoid tumor (BC) in children is very rare, with an incidence of 1.8/1'000'000 per year. Nevertheless, it represents the most common primary lung neoplasm in children. This case discusses the clinical presentation, diagnosis, and management of a 16-year-old girl diagnosed with an atypical carcinoid in the right upper lobe of the lung.

Case presentation: A 16-year-old girl presented with a 12-day history of cough and fever. Initial diagnosis by a pediatrician was an asthma attack, and treatment with Ventolin was started. On day 8, laboratory tests revealed a markedly elevated CRP (>200 mg/l), and chest X-ray showed signs of pneumonia, prompting the initiation of antibiotic therapy (Co-Amoxicillin 1g twice daily). Due to lack of improvement, additional Clarithromycin was prescribed, and a chest ultrasound revealed further abnormalities. The patient had a history of bronchial asthma and dust mite allergy. In the clinical presentation the patient had a normal oxygen saturation (SpO2 96%) and respiratory rate. Auscultation revealed amplified breath sounds over the right lung, but lymph node examination was unremarkable. Laboratory results showed CRP of 231 mg/l, a leukocyte count (Lc) of 8.12 G/l, and negative tumor markers (neuron-specific enolase, chromogranin A, serotonin, 5-HIES and calcitonin). A chest CT scan revealed a high suspicion of bronchial carcinoid with obstruction of the entire right upper lobe bronchus, post-stenotic atelectasis, and possible intrapulmonary abscesses/necrosis, as well as reactively enlarged ipsilateral lymph nodes. FDG-PET-CT showed no evidence of metastatic disease. A diagnostic bronchoscopy was performed, though no biopsy was obtained. The patient was treated with Co-Amoxicillin orally for pneumonia and underwent open adhesiolysis, upper lobe bronchus sleeve resection, and systematic right lymph node dissection. All dissected lymph nodes were tumor-free. The resected tissue confirmed the diagnosis of atypical carcinoid, with no lymph node metastasis.

Conclusion: Bronchial carcinoid tumors often present with nonspecific respiratory symptoms, including cough, recurrent pneumonia, and stridor. When patients experience repeated

episodes of pneumonia or asthma attacks that are refractory to treatment, bronchial carcinoid should be considered as a possible diagnosis.

OC 11

Echoes from the past #bariatric surgery

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A term (41+1) newborn girl was admitted on day 3 due to a >10% weight loss and persistent non-bilious vomiting. Blood gas analysis showed hypochloremic metabolic alkalosis. Newborn screening and other tests were normal. Abdominal and cranial ultrasounds, chest and abdominal X-rays and gastrointestinal passage examination were normal. The infant received oral and small portion tube feeding with maternal milk but vomiting persisted. A trial of esomeprazol for possible gastritis and atropin for possible pylorospasm remained unsuccessful. Vomiting resolved only at minimal enteral nutrition. Slow re-introduction of enteral nutrition with enriched maternal milk by continuous tube feeding was then tolerated and bolus feedings could be reintroduced. After switching completely to formula, the girl finally gained weight. Further metabolic investigations revealed elevated serum methylmalonic acid (MMA) levels (14,200 nmol/L). It turned out that the mother had gastric bypass surgery in 2012 and had discontinued B12 injections during the third trimester. At a check-up at 5 weeks, the infant showed improvement, normal vitamin B12 values and MMA levels decreased to 1,300 nmol/L.

Discussion: The newborn's symptoms suggest gastrointestinal disorders, with pyloric hypertrophy and pyloric spasm as differential diagnoses. In pyloric hypertrophy, vomiting would be projectile and ultrasound would prove diagnosis. Pyloric spasm, however, improves with atropine therapy. Elevated MMA levels point to vitamin B12 deficiency, in this case likely due to interruption of maternal substitution. Elevated MMA is linked to gastrointestinal symptoms, including vomiting, and has been reported as a symptom of B12 deficiency in neonates. Vitamin B12 deficiency is a concern for post-bariatric women due to altered absorption. These women are at higher risk for micronutrient deficiencies, affecting feto-neonatal health. Data show a higher risk of small for gestational age (SGA) infants in this population. Given that gastric bypass surgery can lead to micronutrient deficiencies, and these deficiencies are known to contribute to fetal growth restriction, it is likely that micronutrient deficiencies play a role in the increased risk of SGA in these women.

Conclusion: With increasing bariatric surgeries, micronutrient deficiencies in pregnant women pose significant risks. Tailored prenatal care, regular screening, and multidisciplinary support are crucial for optimizing maternal and fetal outcomes.

OC 12

PIA Bern: Enhancing Pediatric Training through Interprofessional Education in Switzerland

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Interprofessional education (IPE) is vital for healthcare safety and quality but remains underused. Pädiatrische Interprofessionelle Ausbildungsstation Bern (PIA Bern), Switzerland's first neonatal interprofessional learning unit, bridges this gap by enhancing healthcare students' confidence and competencies through work-based learning. It addresses the lack of neonatal care training and IPE in the Swiss curriculum, fostering collaboration in a high-stakes care setting.

Despite IPE's proven benefits in teamwork, error reduction, and patient safety, Switzerland yet lacks frameworks for integration. PIA provides a unique platform for interprofessional collaboration, emphasizing patient-centered care, empathy, and shared decision-making—critical for neonatal health.

Contributions from parents, students, and facilitators create an inclusive learning environment. Parents, often overlooked in medical training, help build trust and improve care. Students gain real-world experience, refining teamwork and communication in complex clinical settings, while facilitators guide diverse learners, reinforcing IPE's role in modern healthcare.

In May 2024 the pilot concluded successfully, proving its effectiveness. Continuation since October includes groups of five students from different medical professions rotating onto the unit for 4 to 8 weeks at a time before handing over to the following group, embedding IPE more profoundly into neonatal care education.

Despite progress, integrating IPE into the Swiss medical curriculum remains a challenge. PIA responds to the growing demand for IPE by improving the transition from academic learning to clinical practice. By assessing the impact of work-based learning on undergraduate health care professionals' confidence and teamwork PIA aims to create a sustainable, replicable model for interprofessional collaboration in neonatal care.

Using a mixed-methods approach, PIA measures its impact through focus groups, reflections, and surveys to assess changes in confidence, skills, communication, and teamwork while addressing patient safety—often overlooked in previous studies. So far 18 students and 8 facilitators have participated, first results of evaluation will be demonstrated.

By bridging gaps between education and practice, PIA fosters teamwork and collaboration, enhances neonatal care, and positions Inselspital as a leader in neonatal IPE, attracting future professionals and improving outcomes for vulnerable neonates and families.

OC 13

Finding the rare case "needle" in your routine data "haystack"

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Objectives: Patients with rare diseases pose a challenge to correctly identifying them in clinical practice. Using routine data may support this effort, but completeness and accuracy need to be assessed.

Materials and methods: As part of an initiative for rare diseases in Switzerland (KOSEK), the clinic for pediatric surgery in Lucerne identified a set of 23 different ORPHA codes, where all patients having this disease should be identified. The analysis consisted of these steps:

- Building a list of keywords relating to the above-mentioned codes. This list was constructed based on the Thesaurus provided by ID-Berlin and rather broad (on purpose, e.g. including terms like “congenital”).
- Matching these codes to diagnosis descriptions entered by providers during in- and outpatient encounters.
- Manual eliminating matches, from different medical contexts.
- Medical evaluation of the remaining cases for inclusion in cohort for this set of rare diseases.

Results: For the 23 ORPHA codes in our cohort, we identified 314 coded diagnosis descriptions from 136 patients in our database. Following the steps described above we found:

For 23 different Orpha Codes the thesaurus from ID Berlin provided 129 different words, from which 73 were selected for matching. Automated searching of these words in diagnostic texts contained in our Electronic Medical Record System (n = 1’363’738) resulted in 6’766 diagnosis descriptions for further evaluation. Manual elimination of cases with a different medical context resulted in 191 diagnostic descriptions for in depth medical evaluation. Medical evaluation identified 164 matching diagnosis texts, for which 88 have not been identified through normal coding previously. These 88 diagnosis descriptions relating to 45 patients represent an increase of 33% (patients) and 28% (diagnosis descriptions) respectively, compared with previously coded cases from our database.

Discussion: The combination of automated analysis of routine data with expert evaluation provides a viable approach to increase the number of identified patients with rare diseases as well as assess the quality of the coding process. We think that our approach could be replicated at other sites with similar effort.

Conclusion: With accompanying quality assessments routine diagnostic data may be used to improve identification of patients with rare diseases. Use of Large Language Models (LLM) may improve this approach even further.

OC 14

New structures for rare diseases

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In Switzerland, an estimated 500’000 people live with a rare disease. 50-75% of these are children. Not only the diseases are rare, but also the expert centres and the disease knowledge. The national concept for rare diseases aim is therefore to identify and recognise new clinical structures, build a national registry and critically review current laws.

Over the last years, 9 Centres for Rare Diseases for people living with a suspicion of a rare disease and 7 Swiss Reference Networks for people with a septic disease groups recognized. These new structures are helpful contacts for paediatricians in private practice either in the search of a diagnosis or for the interdisciplinary health care for these (mostly complex) diseases.

Other measures that are being implemented is the Swiss Rare Disease Registry, legal bases for mandatory reporting of rare diseases, Orphanet as a knowledge and information database for rare diseases and the strengthening of patients’ organisations as actors in this field.

The aim of this presentation is to give an overview of the existing national projects and useful contacts for paediatricians in their daily practice based on examples.

OC 15

Corail Center: Switzerland’s first complex care model for children with rare and/or complex diseases - 24-month update

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The prevalence of children requiring complex care is constantly increasing. Identifying medical complexity is an essential issue in terms of both health economics and quality of care. In Switzerland, there was no structure dedicated to supporting such children, despite a need clearly identified by families and healthcare teams. Following a report drawn up at the initiative of the HUG Private Foundation in 2019 requesting an inventory of the current state of care for children with rare and/or complex diseases in Geneva, the Corail Center (Centre for interdisciplinary coordination and care of complex rare diseases in children) based on "complex care" models already existing in the USA and Canada, opened its doors in January 2023 to meet family needs.

The center's main objective is to establish and coordinate a personalized care itinerary for each child, tailored not only to their illness, but also to the needs of their family, providing medico-psychosocial care and ensuring a link with the school network. This work is carried out in close collaboration with all those involved in the child's care, including the pediatrician, who remains the orchestrator of local care. In addition, the center offers families a support program and a social support for comprehensive care.

The team comprises 2 FTE medical staff, 2.9 FTE nursing staff, 1.6 FTE psychologist, 0.25 FTE social worker, 1 FTE secretary and 0.1 FTE project manager.

Twenty-four months after opening, the Corail Center is treating 157 patients with rare and/or complex diseases aged 0 to 18, 87% with an identified genetic diagnosis (e.g. NF1(9), STB(7), Smith-Magenis(5), Williams syndrome(4)). Patients may be referred by their pediatrician, through the hospital's internal or external network, or directly by the family. Among these families, 22% benefit from psychological support and 43% from social support within the Center.

The assessment after 24 months is very positive from the point of view of both families and professionals. The financial cost of a center that essentially provides coordination and support remains a major challenge for any healthcare system (86% non-billable in 2024). However, such a model of care meets a major public health need and could be beneficial in other cantons. This model of center, which brings together a large number of patients, could also provide better access to innovative treatments being developed for rare diseases.

OC 16

Choosing wisely in Pediatrics: Assessment of the implementation status and strategies for improving implementation in Switzerland

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Background: In 2021, the Swiss Society of Paediatrics published the Choosing Wisely (CW) initiative, featuring official recommendations aimed at reducing unnecessary medical interventions in children. In 2024, a second list was released, expanding the initiative with recommendations of five additional topics. Despite the list's publication, there is limited knowledge regarding how well general practitioners and pediatricians in Switzerland are aware of these guidelines, how consistently they apply them in practice, and what obstacles they face in implementation. Additionally, it is unclear how informed parents are about these recommendations and whether they are willing to follow them. Our study therefore aims to assess the familiarity of Swiss pediatricians and general practitioners with these CW guidelines and identify the factors that hinder their implementation.

Methods: The study will include a quantitative survey of pediatricians and general practitioners across Switzerland, as well as focus groups and qualitative interviews with a smaller group of healthcare providers. Also, parents and families attending clinics and pediatric emergency departments will be surveyed on these topics. Recruiting will start in summer 2025.

Discussion: The presentation will give examples of successful pediatric health research projects which served as blueprints for our collaboration and invite to participate and engage. Based on our findings, a strategy to improve implementation of the CW recommendations will be developed. This research initiative aims to bridge the gap between existing pediatric CW guidelines and their practical application in Swiss healthcare settings.

OC 17

KiKli Fit: Implementation of a physical activity program in pediatric oncology: A qualitative approach

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Introduction: Growing evidence in pediatric oncology highlights the benefits of physical activity (PA) during and after cancer treatment for physical and mental health. However, systematic promotion of PA during cancer therapy remains rare in Swiss pediatric oncology centers. To address this gap, the KiKli Fit project was developed, offering supervised PA therapy. It was implemented over the past 18 month in the department of pediatric hematology and oncology at the University Hospital of Bern. The aim of the research project was to evaluate the implementation into clinical practice, to assess its acceptance among stakeholders and to develop recommendations for the long-term implementation of PA in other centers.

Methods: The Consolidated Framework for Implementation Research (CFIR) was used to structure the study and to develop, implement, and evaluate the KiKli Fit project. A total of 23 qualitative interviews were conducted with children, parents, medical staff and implementation coordinators. These interviews explored attitudes, barriers, facilitators, and expectations regarding the implementation of the project in the pediatric oncology setting and will be analysed through qualitative content analysis.

Results: Preliminary findings indicate broad acceptance of the project among children, parents, and the medical staff. Children and parents reported positive experiences from participating in PA therapy, noting perceived benefits such as reduced treatment-related side effects, increased self-efficacy and improved well-being. Medical staff highlighted the successful integration of the KiKli Fit project into everyday clinical practice and emphasized the enriching aspects of PA therapy for patients' mental health. Key insights include overcoming limited resources and logistical barriers for successful implementation. Additionally, professionals from other units expressed interest in expanding PA therapy for hospitalized children with chronic illnesses.

Discussion: The high level of acceptance among all stakeholders and the reported benefits underscore the importance of PA therapy in improving treatment quality. The identified challenges and insights gained from the implementation process serve as guidance for a successful transfer to other pediatric oncology centers. The interest from other hospital units highlights the potential expansion to other units, i.e. pediatric cardiology, which should be further investigated and adapted as necessary.

OC 18

Transforming health care from day one: stakeholder analysis results for the development and implementation of an electronic health record for newborns in Central Switzerland

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Background: The establishment of an automatically opened electronic health record (EHR) for newborns represents a critical step in advancing pediatric healthcare. Currently, health data is managed in a decentralized manner with indiverse formats including analog data. The role of the existing national EPD is advancing slowly and not targeting newborns. Electronic health data collection right after birth is key for personalized healthcare. The aim of this survey was to assess the feasibility, functionality and implementation requirements for an EHR for newborns among stakeholders in Central Switzerland.

Methods: The stakeholders were selected from professional and personal networks as well as organizations with an interest in pediatric EHR development and aimed to represent healthcare professionals, parents and representatives from regulatory bodies. The survey was distributed via mail. The survey focused on three core topics: needed functionalities of the EHR post-birth, stakeholders' specific needs and expectations and perceived challenges and risks. Additional topics included preferences for platform solutions and the acceptability of a potential user fee.

Results: Of 51 stakeholders, 36 responded (rate of 71%). The three most often stated concerns about the implementation of the EHR after birth were lack of data maintenance (50%), data security (28%) and technical complexity (22%). Stakeholders expressed strong support in general with 30% indicating no general concerns regarding implementation. Stakeholders saw benefits in the areas of improved communication (84%), support for parents (61%) and availability of digital vaccination data (56%). Desired key functionalities of the EHR after birth were documentation (89%), sharing of data (86%), contact data of healthcare providers (70%) as well as information storage (70%) and appointment coordination (40%). The most often stated device for using the EHR was the smartphone (95). The use of a national EPD solution was the most popular option concerning platform use (78%).

Conclusion: This survey provides valuable insights into preferences of stakeholders and helps to guide the design and implementation of starting health data collection from day one in life. Addressing the identified challenges will be key to achieving stakeholder acceptance, overcoming organizational obstacles and ensuring the system's successful rollout in a pilot study in Central Switzerland for newborns born in the hospital.

OC 19

Bruising in pre mobile infants: child abuse or self induced sucking bruising? A case report

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We describe the case of a 23 day old infant, presenting with atypical linear ecchymosis diagnosed as sucking bruises. Given the scarcity of literature on the subject, we wish to report this case to raise awareness of sucking bruising and therefore limit the emotional and financial cost of unnecessary admissions and social investigations.

This previously healthy term infant was brought to the emergency department following a head trauma. The child fell out of the carrier whilst the father bent over, subsequently causing the infant to land on his occiput. There was no loss of consciousness nor symptoms of concussion in the following hours.

On clinical examination, the infant was well and neurologically reassuring. We however noted a linear ecchymosis on the proximal posterior-lateral side of his right forearm*. The rest of the examination was normal, with no other skin lesions nor apparent bone damage. Upon questioning, the parents noticed the lesion the day prior consulting and thought it was due to the infant bumping his arm against the chair but weren't sure. The infant had received prophylactic vitamine K at day 0 and day 4, was growing well and was exclusively breastfed. There were no known haematological diseases in the family.

Given the history of head trauma and bruising, the infant was admitted for surveillance and investigation of possible non-accidental injury. Blood tests showed no inflammation, no thrombopenia and a normal coagulation.

During his stay in hospital, two new similar but smaller ecchymosis appeared on the distal lateral side of the same forearm. His parents witnessed him suckling his own arm when hungry, subsequently causing the lesions. With no other concerns for child abuse and the discovery of similar cases in the literature, a diagnosis of sucking bruising was made allowing the infant to return home with his parents 24h after admission.

Bruising in pre-mobile infants is usually associated with non-accidental injuries or witnessed trauma. Self-inflicted bruising is rare but possible when hungry infants forcefully suckle their arms or hands. Although this phenomenon seems well described on parenting forums, the literature remains very scarce. To our best knowledge, there are only 5 other cases reported. We hope the publication of these cases will contribute in raising awareness amongst healthcare practitioners.

Parents have consented for the use of the photos and history.

OC 20

"Doctor, what would you do?": Effect of the Doctor's Personal Perspective on Parental Preparation for Shared Decision-Making

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Background: The question of whether physicians should disclose their personal perspective when asked by parents of seriously ill infants remains controversial. This study aimed to evaluate whether disclosing or withholding physicians' personal perspectives affects parental preparation for decision-making.

Methods: A web-based randomised clinical trial was conducted from February to April 2024. German-speaking parents of infants born before 37 weeks of gestation were recruited through parent support organisations. Participants were randomly assigned to view one of two animated video vignettes depicting a conversation between a neonatologist and the parents of a seriously ill infant considering tracheostomy placement. In the intervention group, the physician disclosed their personal perspective when asked, "Doctor, what would you do?", while in the control group, the physician withheld their perspective. Both scenarios adhered to the principles of the ANSWER framework (active listening, self-awareness, and eliciting needs and

values). The primary outcome was parental preparation for decision-making, measured using the Preparation for Decision Making Scale (PDMS), ranging from 0 (not at all prepared) to 100 (excellently prepared). The main secondary outcome was preference for or against disclosure.

Results: A total of 218 participants (mean [SD] age, 36.1 [5.5], 88.1% female) were included in the analysis (control group: n = 112; intervention group: n = 106). There was no difference in mean [SD] PDMS scores between the two groups (control group: 65.9 [19.0] vs. intervention group: 64.8 [18.9]; mean difference, 1.04 [95% CI, -4.03 to 6.10]; t(216) = 0.403; p = 0.687). Approximately half of the participants 76 (52.8%) expressed a preference for the physician to disclose their personal perspective, while the remaining 68 (47.2%) participants did not.

Conclusions: The present study found that participating parents did not report feeling better or worse prepared for decision-making as a result of physicians' disclosure of their personal perspective. This suggests that a response with the personal opinion to the question "Doctor, what would you do?" might be given, when paying attention to concepts like ANSWER.

OC 21

Parental views on involvement in neonatal research: a cross-sectional observational study

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Background: Research has been conducted to assess and improve neonatal healthcare, but little attention has been given to the parents' perspective. So far, only a few initiatives have been launched in neonatology to involve parents in research. The aim of this study was to explore the views of parents of preterm infants regarding their involvement in neonatal research.

Methods: This cross-sectional observational study included Swiss, Austrian, and German parents of preterm infants who had previously participated in a web-based randomized control trial. The self-developed questionnaire included nine questions about parental involvement in neonatal research in four areas: 1) research priorities, 2) research tasks, 3) availability of participants and 4) reimbursement of expenses. Data analysis was descriptive.

Results: In total, 187 parents participated. Suggestions for research priorities were primarily related to three areas: infant neurodevelopment, support for parents during their time in the neonatal unit, and parental mental health. In terms of research tasks, the most frequently mentioned were feedback on study material (91 [48.7%] parents) and support with data collection (43 [23.0%] parents). In total, 59 (31.6%) parents indicated their willingness to attend a minimum of two meetings per year. With regard to reimbursement of expenses, 117 (62.6%) parents indicated that covering of travel and parking costs was a priority.

Conclusions: Parents who have engaged in clinical trials represent a valuable resource for patient and public involvement. They are willing to participate and have specific ideas about neonatal research with a focus on neurodevelopment.

OC 22

Delta wave as an early sign of rhabdomyomas in Tuberous Sclerosis Complex

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Introduction: Tuberous sclerosis complex (TSC) is one of the Phacomatoses and is a genetic disorder attributed to mutations in the tumour suppressor genes TSC1 or TSC2. TSC manifests itself with many facets and is characterised by the development of benign tumours of ectodermal origin in various organ systems, such as rhabdomyomas of the heart. Although the majority are clinically silent, depending on their localization, they can have an influence on hemodynamics or cause arrhythmias.

Case report: A 7-month-old male infant presented to our emergency room due to recurrent seizures. The patient's personal history was otherwise unremarkable. The clinical examination showed 2 white spots in an otherwise healthy infant and especially no cardiac or neurologic abnormalities. An EEG demonstrated multifocal epileptic activity in part generalised classified as hypsarrhythmia. A cranial MRI revealed the presence of numerous subependymal nodules along the lateral ventricles, as well as a calcified subependymal nodule in the cella media. The clinical findings and the imaging were compatible with the diagnosis of tuberous sclerosis. Genetics confirmed the diagnosis showing a mutation in the TSC2 gene. Cardiac evaluation revealed the presence of 3 small rhabdomyomas. They were located within the atrioventricular canal, as well as in the left ventricular wall. No obstruction of the intracavitary inflow or outflow tract of either ventricle could be observed. A 12-lead ECG showed a clear delta wave with a short PQ interval of 74 ms at a heart rate of 126bpm. A 24-hour ECG revealed a continuous sinus rhythm without significant ectopy and no episodes of re-entry-tachycardia. The delta wave disappeared at a heart rate above 130 bpm. The patient was immediately seizure free with vigabatrin and continued to be seizure-free at two months follow-up. However, a slight motor developmental delay was observed. Cardiac investigations revealed no episodes of suprav-ventricular tachycardia. The echocardiography showed a regression in size of the rhabdomyomas with persistence of a Delta wave.

Conclusion: The delta wave is a classic ECG sign of the presence of an accessory pathway in the heart. In TSC it is rarely described, although electrocardiographic evidence of multiple atrioventricular accessory pathways has been described in cardiac rhabdomyomas. The presence of a delta wave may serve as an early detection sign of patients with TS and rhabdomyomas in patients with chilhood epilepsy.

OC 23

A randomized controlled trial comparing immersive virtual reality games versus nitrous oxide for pain reduction in common outpatient procedures in pediatric surgery

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Background: Children often experience anxiety and pain during minor surgical or interventional procedures, highlighting the need for effective, non-pharmaceutical pain management strategies. This study evaluates the efficacy of virtual reality

(VR) gaming compared to nitrous oxide in reducing pain during pediatric outpatient procedures and especially in minor surgical interventions.

Methods: In a single-centred randomized controlled trial in a university hospital in Switzerland, 100 children aged 6–15 undergoing minor surgical procedures were allocated to either a VR gaming or nitrous oxide group. Stratification was performed by age group (6–9 years, 10–15 years) and type of intervention. Pain levels were assessed using visual analog and face scales immediately after the procedure and two weeks later. Secondary outcomes included the enjoyment or fear experienced, willingness to repeat the procedure, and potential time limitations of VR compared to nitrous oxide. Adverse events were also monitored.The case report forms (CRFs) for this trial are implemented using the REDCap electronic data capturing system, which was also used for randomization. It is securely hosted and managed by the Clinical Trial Unit (CTU) of the University of Bern.

Results: We observed that pain levels were low in both groups, with slightly lower levels observed in the nitrous oxide group. The median pain scale in the VR group was 3/10 (mean = 3.48) and the median pain scale in NO group was 1/10 (mean = 2.16), p = 0.003. However, overall satisfaction, including enjoyment, fear and willingness to repeat, appeared higher in the VR group, despite the higher reported pain levels in this group. Subgroup analysis were performed additionally.

Conclusion: VR may be a promising alternative to nitrous oxide for managing pain and anxiety in pediatric procedures. While nitrous oxide demonstrated slightly better pain control, the higher satisfaction rates in the VR group highlight its potential as a versatile and engaging tool for personalized pain management. Further results will provide valuable insights to optimize pediatric pain management strategies, especially age-dependent differences in VR effects, influenced by higher gaming experience in older children.

OC 24

Prevention and management of sports-related mild traumatic brain injury in children and adolescents: A mixed-methods multi-stakeholder approach in Switzerland

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Children and adolescents in contact sports are highly susceptible to mild Traumatic Brain Injuries (mTBI), commonly known as concussions, necessitating effective prevention and management strategies. This project focuses on communication measures to achieve three objectives:

Systematic evidence collection on SRC incidence among Swiss adolescents and health education strategies promoting SRC awareness.

Establishment of an interdisciplinary network to discuss and evaluate communication strategies for SRC prevention.

Development of recommendations for communication measures aimed at preventing SRC among adolescents and their caregivers.

Using a participatory, mixed-methods approach, we contacted insurance companies, political entities, and institutions for data on SRC incidence. A systematic literature review identified 22 studies on SRC prevention, primarily from Canada and the US, showing short-term effectiveness in improving awareness, knowledge, and symptom reporting. Fifteen interviews with experts, coaches and parents revealed significant knowledge gaps and inconsistent communication strategies. Experts emphasized the need for sport-specific, sustainable communication strategies focusing on proper management to prevent SRC worsening.

At the conference, we will discuss recommendations for future SRC prevention strategies using communication measures and reflect on our participatory approach. These insights aim to help public health and health communication scholars translate research into effective policy and action.

PAEDIATRIC GRAND ROUNDS

PGR 1

Chronic cough – is it really asthma?

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Case report: A 14-year-old female adolescent was admitted to our pediatric pulmonology outpatient clinic for further investigation after a prolonged community acquired pneumonia with partial respiratory failure, recurrent febrile episodes, chronic cough, exertion-dependent breathing difficulties and physical limitation for the last year. She had been diagnosed with asthma three months ago. A pulmonary function test showed a severe obstructive ventilation pattern and reduced peak flow with improvement after bronchodilator inhalation (FEV1 60% predicted /+19%). A step-up in asthma therapy (OCS, high-doses ICS-LABA inhalation) showed no improvement of symptoms and lung function one week later. We planned a thoracic CT scan and bronchoscopy to evaluate the airways and lung structure. At this time, the patient first mentioned swallowing difficulties of dry food. Further, we excluded infectious causes of recurrent fever and defects of immunologic function. The CT showed findings compatible with an achalasia with dilation of the esophagus, compression of the trachea and signs of chronic aspiration. High-resolution manometry confirmed the diagnosis of achalasia type II (Chicago classification). The treatment consisted of dilation of the cardia with EsoFLIP. At the follow-up visit six weeks after the intervention, symptoms improved dramatically. The patient only reported mild dry cough and minimal dysphagia. She was able to fully resume her physical activities. Lung function test showed normal values (FEV1 110% predicted). A barium study revealed a decrease in esophagus width, while esophageal emptying was still slightly slower than normal.

Discussion and conclusion: The case demonstrates the importance of screening for esophageal disease in patients with unresolved chronic cough. Symptoms can be subtle and require active inquiry. Since undiagnosed achalasia increases the risks of complications during endoscopic procedures, it is important to establish the diagnosis prior to anesthesia.

PGR 2

Beware of hidden dangers: 1 patient, 2 rare diseases

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Background: Wilson's disease is a rare genetic disorder caused by a defect in copper excretion, resulting in its accumulation in the liver, eyes (forming Kayser-Fleischer rings), and brain (particularly in the basal ganglia). The clinical presentation can be either acute or chronic. Diagnosis is based on clinical evaluation, biochemical analyses, radiological, and confirmation through genetic testing.

Case report: A 10-year-old boy known for phenylketonuria (PKU) was followed elevated aminotransferases since the age of 3 with ultrasound findings compatible with steatosis, ascribed to his PKU. He was referred for acute liver failure. Following a viral infection with fever and diarrhea, he developed jaundice, dark urine, and abdominal distension two weeks later. At this time, PCR for SARS-COV2 was positive on the nasal swab and the laboratory investigations showed a mild elevation of liver tests with unremarkable serologies. The symptoms worsened with a more significant distension, a digestive discomfort, and a 3 kg weight gain, prompting a new consultation. Physical examination confirmed jaundice, spider naevi of the face, abdominal distension and splenomegaly. An extensive work up revealed the following:

- INR of 2.1 and V factor of 23% (N >70%)
- ASAT 95 U/l (N:0-33), ALAT 51 U/l (N:0-19), GGT 213 U/l (N 4-12), conjugated bilirubin 38 µmol/ (N 0.0 – 3.7)
- Pancytopenia (hemoglobin 102g/l, leukocytes 3.9G/l, platelets 92 G/l)
- Low ceruloplasmin levels: 0.12g/l (N 0.17-0.46)
- Elevated exchangeable copper: 2.44 µmol/L (N <1.15)
- Elevated 24-hour urinary copper 28.70 µmol/L (N <0.63)
- A1 anti-trypsine at phenotype MS
- Auto-immune hepatitis panel negative
- Parvovirus PCR + (trigger for the acute decompensation)
- Abdominal ultrasound: small cirrhotic liver, splenomegaly and significant ascites
- Brain magnetic resonance imaging normal
- Ophthalmological examination normal.

A liver biopsy confirmed cirrhosis stage, and the histological findings were compatible with Wilson's disease. All these results were suggestive of Wilson's disease. The patient was started on D-penicillamine, leading to a gradual improvement in laboratory results and overall condition. The genetic testing confirmed the diagnosis, with a homozygous variant identified in the ATP7B gene.

Conclusion: Abnormal aminotransferase levels need follow up and a full work up if persistently elevated. One rare disease does not rule out another, even in a non consanguinous family.

PGR 3

Diagnostic Challenges in neurodevelopmental disorders: ADHD? Epilepsy? Or Celiac disease? A case report.

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Background: Celiac disease (CD) is a multi-organ inflammatory disease. Associations between CD and neurological conditions such as epilepsy and behavioral symptoms or neurodevelopmental disorders, like attention-deficit hyperactivity disorder (ADHD), have been demonstrated in numerous studies.

Case description: A 9-year-old boy was referred for evaluation due to characteristic ADHD symptoms. A detailed assessment revealed gross and fine motor dysfunction, along with severe impaired spatial perception, without fulfilling diagnostic criteria for ADHD. An electroencephalogram (EEG) examination revealed "Occipital Intermittent Rhythmic Delta Activity (OIRDA)" with no clinical or EEG evidence of focal epilepsy at that time. Blood tests showed a lightly elevated tissue transglutaminase IgA, so that CD was suspected. Under a multimodal treatment approach, no significant progress was observed. During follow-up, unclear staring episodes and impaired consciousness were

reported and video documented. The EEG showed focal epileptic signals on the right temporal lobe side. An antiepileptic therapy for focal epilepsy was initiated. Under the antiepileptic therapy, the patient remained seizure free. Behavioral symptoms and visuospatial perception were significantly improved. A gastric biopsy for CD confirmed the suspected diagnosis of CD. The patient showed under the gluten-free diet a further marked improvement in his neurodevelopmental symptoms, previously mimicking ADHD. Antiepileptic treatment was stopped after 2 years. The gluten-free diet was continued. Finally, under continuation of the gluten-free diet, the behavioral and neurological situation remains stable.

Conclusion: In the differential diagnosis of neurodevelopmental disorders, such as ADHD, with atypical courses, lack of response to the multimodal therapy approach and with the occurrence of neurological manifestations, such as epilepsy, the possibility of CD must be considered. The association between CD and ADHD and/or neurological conditions like epilepsy is well established.

PGR 4

Congenital Diarrhea: A Case Series with Diverse Genetic Backgrounds

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Congenital diarrhea is a rare disorder with a broad spectrum of differential diagnoses. Early diagnosis and treatment are essential to prevent life-threatening complications. The aim of our case series is to demonstrate the value of early genetic testing.

We report four cases of congenital diarrhea, presenting with dehydration and metabolic decompensation. Three required parenteral nutrition (PN) for 4–26 weeks, one remains dependent on PN. The diagnostic work-up included tests for infectious, allergic, metabolic and immune diseases, with were insufficient to establish a diagnosis. Endoscopy with biopsies is typically the next step, but it is invasive, particularly in young infants. In the first case with watery diarrhea since birth and consanguinity, genetic analysis revealed a homozygous pathogenic variant in the STXBP2 gene leading to the diagnosis of presymptomatic hemophagocytic lymphohistiocytosis and microvillus inclusion disease, which was later confirmed by electron microscopy. In the second case, variants in the CLMP gene led to the diagnosis of congenital short gut syndrome. Targeted radiologic imaging demonstrated a significant shortening of the small and large intestine. In the third case, histology suggested autoimmune enteropathy with positive anti-enterocytes antibodies. Genetic testing identified a gain-of-function variant in the STAT3 gene, which is associated with early onset autoimmune disease, immunodeficiency and lymphoproliferation. The treatment strategy was adapted, including a potential stem cell transplantation to prevent the development of type 1 diabetes or lymphoproliferative diseases. In the last case, endoscopy and histology also revealed an autoimmune enteropathy. This child also had a juvenile myelomonocytic leukemia and polycystic kidney disease. Genetic testing allowed the diagnosis of a Noonan syndrome due to a heterozygous variant of the PTPN11 gene. All four patients underwent trio exome sequencing. Interestingly, targeted gene panels for congenital diarrhea do not include all the genes implicated in the patients of our series.

In congenital diarrhea underlying monogenetic diseases are common. Trio exome sequencing enables an early diagnosis and thus may avoid invasive investigations. Additionally, comorbidities may be detected early, even presymptomatic, early disease specific treatment may be established and further prognostic information may be provided.

PGR 5

Red scaly baby - a case report

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Background: Neonatal skin lesions are common and mostly harmless [1]. Extensive skin lesions at birth are rare and may indicate an underlying systemic disorder, demanding prompt diagnostics and treatment [2].

Case Report: A term male infant of a 22-year-old mother, born via spontaneous vaginal delivery, presented with erythema and superficial skin erosions in the centrofacial area, on the scalp, lateral forearms, thoracolumbar region, as well as in the scrotal and inguinal regions at birth. The lesions were partially exudative. Nikolsky's sign was negative. The mucous membranes and the remaining clinical examination were inconspicuous. There were no complications during pregnancy. Family history was unremarkable. The preliminary diagnosis of "red scaly baby" was made, and the dermatology department was consulted. A skin biopsy was performed. Local treatment was initiated, including disinfection measures and covering of the erosive lesions with absorbent foam dressing. Intact skin was regularly treated with Dexpanthenol. To reduce the risk of dehydration and hypothermia, incubator humidity was adjusted to 70%, and sodium levels were followed closely. Initially, the following differential diagnoses were the main focus: Netherton syndrome (or another form of ichthyosis), Langerhans cell histiocytosis, neonatal lupus, and immunodeficiency. Extensive laboratory diagnostics, including FACS, were initiated. Except for an elevated LDH, which we interpreted as being related to birth, and eosinophilia the tests came back negative. However, eosinophilia would have been consistent with a diagnosis of Netherton syndrome. Histology from the punch biopsy showed compact orthohyperkeratosis with an intact stratum granulosum and intraepithelial neutrophil granulocytes, thus excluding a diagnosis of Langerhans cell histiocytosis and making a treatment with topical steroids possible. Direct immunofluorescence was unremarkable. Unfortunately, the histology did not lead to a definite diagnosis. Given the clinical appearance, eosinophilia, and elevated sodium levels we suspected Netherton syndrome. LEKTI Immunohistochemistry was performed to confirm the suspected diagnosis. The result is still pending. Local treatment led to a rapid reduction of the skin lesions.

Conclusion: The diagnosis of a "red scaly baby" is a neonatal dermatological emergency with a broad differential diagnosis. However, initial work-up and treatment is similar regardless of the suspected diagnosis.

PGR 6

Doctor, my child stopped walking!

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Introduction: Chronic recurrent multifocal osteomyelitis (CRMO) is a rare inflammatory bone disease and a diagnosis of exclusion. It presents with recurrent bone pain and sterile inflammatory lesions at multiple bone sites.

Case description: We report the case of a 5-year-old child with autism spectrum disorder (ASD) and mild malnutrition who presented with acute limping and refusal to walk for 7 days, without fever. Physical examination, including neurological and osteoarticular assessments, was normal except for pain on palpation of both lower limbs. Laboratory tests revealed microcytic regenerative anemia and moderate inflammation (CRP 10 mg/L, ESR 50 mm/h). Intravenous antibiotics were started but showed no clinical improvement. Radiological investigations, including ultrasound and MRI, excluded osteoarticular infection. Whole-body MRI revealed symmetrical signal alterations, mainly in the metaphyseal regions of the lower limb long bones and sacrum, particularly the distal femur. The differential diagnosis was CRMO versus scurvy. Dietary history revealed severe food selectivity over two years, limited to white bread and chocolate milk. Although vitamin C levels were normal, a nutritional workup uncovered multivitamin deficiencies (A, E, D, B9, and zinc), hypoalbuminemia, and hypercalcemia due to high calcium intake. Rheumatological consultation led to anti-inflammatory treatment, with gradual recovery of walking ability within days. Enteral feeding corrected the nutritional deficits, yielding significant overall improvement. Follow-up confirmed CRMO as the primary diagnosis, and anti-inflammatory therapy was stopped after two months.

Discussion: This case highlights the diagnostic challenge of distinguishing CRMO from conditions linked to nutritional deficiencies in children with ASD and food selectivity. While no direct link between malnutrition and CRMO has been proven, severe deficiencies might influence disease onset or progression. Multidisciplinary management involving rheumatology, nutrition, and metabolism was critical in diagnosis and treatment.

Conclusion: CRMO should be considered in children with unexplained limping, especially in complex cases involving ASD and restrictive eating. Though a causative link between malnutrition and CRMO is unconfirmed, this case underscores the need for further research and a holistic, multidisciplinary approach to optimize care.

SWISSPEDNET

SPN 1

Association between hearing impairment and cognitive development in children with low birth weight

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Background: Low birth weight is a risk factor for various health outcomes, including deficient hearing and cognition. Hearing loss affects 2 in 1000 children and can impact cognitive development. Despite poor hearing and cognition being potential consequences of low birth weight, their association remains unexplored in this population.

Aim: This study investigated the hearing ability and the cognitive development of individuals with low birth weight at two time-points—in childhood and early adolescence—and how the two domains are associated over time.

Method: Data from the Zurich Longitudinal Studies were analyzed, including 376 children born between 1973 and 2002, categorized as either low birth weight (<2500g) or normal birth weight (≥ 2500g). Pure-tone audiometric assessments were conducted at 8 years, and cognitive testing was performed at 9 and 14 years at the University Children's Hospital Zurich. Mixed-effects models were used to examine the relationship between birth weight, hearing abilities and cognitive development over time, while controlling for socioeconomic status.

Results: Children born with low birth weight were 1.9 times more likely to have hearing impairment (≥ 26 dB) at a pure tone average (PTA) between 0.5 and 4kHz and 3.1 times at a PTA up to 6kHz than children with normal birth weight. A significant mean difference of 5.6 dB was observed for PTA up to 4kHz ($p < 0.001$), with low birth weight children showing on average poorer hearing compared to normal birth weight children (M = 21.04, SD = 5.99 vs. M = 15.43, SD = 6.72, respectively). Verbal IQ did not differ between groups, but full-scale and nonverbal IQ scores were significantly lower in the low birth weight group at both ages, controlling for hearing ability. Across the entire cohort, poorer hearing at 8 years significantly predicted lower full-scale IQ ($p < 0.01$) and lower verbal IQ scores ($p < 0.01$) at 9 years. This association diminished by the age of 14 years for both full-scale IQ ($p = 0.97$) and verbal IQ scores ($p = 0.91$). No significant association was evident between hearing and nonverbal IQ at either 9 years ($p = 0.07$) or 14 years ($p = 0.70$).

Conclusion: Low birth weight children were vulnerable to poorer hearing, which related to lower full-scale and verbal IQ scores at age 9 but not at age 14. Further research should explore the cause of this cognitive catch-up in individuals with poor hearing to support at-risk children.

SPN 2

Artificial Intelligence for Pediatric Fracture Detection: A Multivendor Comparison Study in Pediatric Emergency Medicine

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Background: Injured children with potential fractures frequently present to pediatric emergency departments (EDs), with diagnoses primarily reliant on radiographs. Relevant misinterpretations of these occur in up to 9% of cases in ED settings, due to insufficient knowledge or rapid decision-making. While artificial intelligence (AI) could improve radiograph interpretation accuracy, most AI tools are developed for adults and have limited evidence for pediatric fracture detection, which is particularly challenging due to growth-related variations and distinct pediatric fracture types. This study evaluated and compared the diagnostic accuracy of AI tools for detecting fractures and dislocations in forearm, elbow, and lower leg radiographs.

Method: Three CE-certified AI-software for pediatric fracture detection participated: Gleamer (BV, France), Radiobiotics (RB, Denmark), and Milvue (SU, France). Consecutive radiographs of approximately 1000 patients per anatomical site from children aged 2 to 18 years, who presented to the ED of the University Children's Hospital Zurich with suspected fractures, were included. Diagnostic accuracy was evaluated using consensus pediatric radiology diagnoses as reference standard. Assessment included fractures, elbow effusions as indirect fracture signs, and dislocations.

Results: In total, 3324 children were included with 1146 forearm radiographs (mean age 7.4 years), 1239 elbow radiographs (mean age 7.4 years), and 1070 lower leg radiographs (mean age 6.7 years). All AI tools showed high accuracy in detecting forearm fractures (sensitivity >91%, specificity >92%) and lower leg fractures (tibial sensitivity >91%, specificity >94%). However, sensitivity for fibular fractures was lower, ranging from 84% (RB) to 91% (BV), though specificity remained >97%. Accuracy for elbow radiographs was lower overall, mainly for detecting fractures and dislocations. Sensitivity/specificity for fractures: BV (89.4%/80.0%), RB (84.4%/86.1%), and SU (90.2%/83.9%); for effusion: BV (79%/96.2%), RB (88.8%/85.4%), SU (85.9%/99.1%). The most notable differences among AI tools were observed in sensitivity and specificity for dislocations: BV: 45.6%/100%, RB: 82.5%/96.3%, SU 53.6%/99.1%.

Conclusion: All AI tools show potential to reduce misdiagnoses in pediatric EDs, particularly for forearm and lower leg radiographs. However, the lower accuracy observed for elbow radiographs highlights the need for further improvement before reliable clinical implementation.

SPN 3

Can routine electronic healthcare records be used to collect and enhance data collection for cohort studies? The SPHN-SPAC demonstrator project.

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Background: The Swiss Paediatric Airway Cohort (SPAC) is a multi-centre study of children referred to Swiss respiratory outpatient clinics. Data is manually extracted from the medical records of over 4,500 participants, which is time- and resource-intensive. The Swiss Personalized Health Network (SPHN), a Swiss federal initiative to enable the use of routine healthcare data for research, could make data collection for clinical cohort studies more efficient.

Aims: The SPHN-SPAC demonstrator project aims to test how SPHN can be used to obtain data for SPAC participants.

Methods: We assessed concordance between manually collected SPAC data and automatically extracted SPHN data for 516 participants enrolled at Inselspital Bern (2017–2023). We obtained SPHN data on sex, clinical visit dates, anthropometric measures, and diagnoses. Concordance between SPAC and SPHN data was assessed using Cohen's kappa (κ) for categorical variables, Pearson correlation and paired t-tests for continuous variables. Diagnostic concordance was evaluated with Jaccard similarity. Discrepancies between self-reported and hospital-recorded hospitalisations were analysed using Intraclass correlation (ICC) and percentage agreement.

Results: SPHN data contained all respiratory clinical events from the hospital system, including hospitalisations and emergency visits, which are available only via self-reports in SPAC. We identified 2333 pneumology outpatient visits from SPHN compared to 1234 manually extracted in SPAC (median: 3 vs. 1). Over 90% of enrolment visit dates matched exactly, and sex showed high concordance (κ = 0.96). Anthropometric measures correlated strongly (>0.95) with no significant differences (p >0.55). Diagnostic concordance was high (Jaccard similarity >0.8 in 90% of cases), showing SPAC data accuracy and SPHN's reliability in identifying clinical data. Self-reported and hospital-recorded hospitalisations showed strong consistency (ICC 90–95%) and high agreement (>98%) on no hospitalisations. More self-reported hospitalisations were observed (24 vs. 17).

Discussion: These results show SPHN's potential to reliably enhance cohort studies, improving completeness by including all hospital-recorded visits. However, SPHN is restricted to individual hospital systems and does not yet capture external data, such as GP visits or events in other hospitals. SPHN data remains limited, requiring further development to enhance its utility for observational cohort studies.

SPN 4

Investigating the Role of Human Breast Milk Extracellular Vesicles (hMEVs) on the activation of Nasal Epithelial Cells in the Context of Maternal Asthma

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Background: Breastfeeding provides nutritional and immunological benefits, yet its role in allergy and asthma prevention remains debated. Human breast milk contains bioactive components, including extracellular vesicles (hMEVs), which influence infant immunity by modulating bronchial epithelial barrier integrity and inflammation.

Objective: This study investigates the immunomodulatory and protective potential of hMEVs on primary human nasal epithelial cells (NECs) in allergic asthma.

Methods: hMEVs were isolated from breast milk donated by one healthy and one asthmatic mother (both with 6-week-old newborns) via size exclusion chromatography. NECs were obtained from nasal brushings of healthy adults and cultured at the air-liquid interface for six weeks until full re-differentiation. To assess the short-term protective effects of hMEVs, cells were treated with hMEVs (1x10^10 or 5x10^10 particles) from either mother, followed by rhinovirus RV-16 infection (MOI = 4) after 24 hours. In a second experiment, cells received either a single or a 5-day hMEV treatment (only with 1x10^10 particles), then some were exposed to RV-16. Basolateral media were collected 24 hours post-infection to measure IL-6, CCL2, CCL5, and CXCL10 via ELISA.

Results: Short-term hMEV treatment did not alter cytokine production, suggesting no immediate immune modulation. However, long-term exposure revealed a significant increase in CCL5 production when cells were treated with hMEVs from asthmatic mothers compared to both single treatment (p = 0.0045) and treatment with hMEVs from healthy mothers in the presence of RV-16 (p = 0.0168).

Discussion: Previous research has indicated that maternal asthma could affect the outcomes of breastfeeding. Specifically, data show that infants born to mothers with asthma who are exclusively breastfed may experience higher allergen sensitization than those fed with formula. As the number of asthmatic women having children continues to rise, it is crucial to further examine this relationship. In our in-vitro study, we found no short-term immunological effects of hMEVs on NECs. However, hMEVs exposure over 5 days, which might closer reflect the physiological conditions in a breastfed child, yield to different CCL5 signals form healthy and asthmatic mothers in the presence of RV-16. Long-term treatment outcomes in our ongoing project may provide valuable insights into how breastfeeding impacts allergic sensitization and the development of asthma in children.

SPN 5

Prevalence and risk factors of right ventricular dysfunction in long-term survivors of childhood cancer - Results of the CardioOnco Study

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Background: Childhood cancer survivors are at risk of cardiac dysfunction many years after treatment with anthracyclines and heart-relevant radiotherapy (RT). While cardiotoxicity is known to impair left ventricular function, the effects on right ventricular (RV) function remain unknown. However, RV dysfunction is a strong predictor of all-cause mortality and cardiac death in other patient populations.

Objectives: We aimed to assess prevalence of RV dysfunction and risk factors for impaired RV function.

Methods: We invited childhood cancer survivors ≥18 years of age, diagnosed between ages 0–20, treated in one of five pediatric oncology centers across Switzerland from 1976–2017 who survived ≥5 years for an echocardiographic assessment of the RV including tricuspid annular plane systolic excursion (TAPSE, abnormal <17mm) and Doppler tissue imaging (DTI, abnormal <9.5cm/s).

Results: We included 432 survivors with a median age at study of 34 years (interquartile range [IQR] 26–40) and a median time since cancer diagnosis of 26 years (IQR 19–34). The prevalence of RV dysfunction overall was 4% (16/432) measured by TAPSE and DTI with a mean TAPSE of 22.5±3.8 mm and a mean DTI of 13.1±2.2 mm. Survivors exposed to heart-relevant RT showed the highest prevalence of RV dysfunction measured by TAPSE (13%) and DTI (9%). Risk factors for decreased RV function were cumulative heart-relevant RT dose [Beta coefficient (B) = -0.46; 95%CI -0.79– -0.20]; hematopoietic stem cell transplantation (HSCT) (B = -1.88; 95%CI -3.35– -0.40); chemotherapy with cisplatin (B = -1.29; 95%CI -2.38– -0.19); diabetes mellitus (B = -2.57; 95%CI -4.99– -0.15); and abdominal obesity (B = -0.79; 95%CI -1.58–0.00) when measured by TAPSE. For DTI, we identified cumulative heart-relevant RT dose (B = -0.19; 95%CI -0.37– -0.01); relapse (B = -0.94; 95%CI -1.74– -0.15); and abdominal obesity (B = -0.80; 95%CI -1.37– -0.24) as risk factors. Cumulative anthracycline dose was not associated with impaired RV function measured by TAPSE (B = -0.23; 95%CI -0.49 – 0.03) or DTI (B = -0.06; 95%CI -0.25 – 0.13).

Conclusions: Adult childhood cancer survivors are at risk of RV dysfunction after cardiotoxic treatment, especially when treated with heart-relevant RT, HSCT, or cisplatin, but also comorbidities such as diabetes mellitus and obesity play an important role. Given the prognostic value of RV function, we suggest including RV echocardiographic assessment to the routine follow-up care of adult CCS.

SPN 6

Enhancing Precision in Pediatric Brain Tumor MRI: Automated Segmentation Using Deep Learning

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Background: Pediatric brain tumors (pBTs) are the most frequent solid tumors and the leading cause of pediatric cancer-related deaths. Achieving accurate tumor detection on MRI is vital for planning both surgical and medical interventions, as well as for treatment responses and monitoring progress. However, manual segmentation is time-consuming and subject to inter-operators variability. These issues can be addressed through the use of Deep Learning (DL). In this study, we present DL-based automated segmentations across different pBTs.

Methods: Baseline MRI images of 176 pediatric patients with pBTs from the Children's Hospital of Zurich were analyzed. Multi-parametric MRI sequences (T1w, T1w-C, T2, and FLAIR) were pre-processed and manually segmented to delineate tumor subregions: T2 Hyperintensity (T2H), enhancing tumor (ET) and cystic component (CC). For ET and CC, only clinically significant subregions were considered, setting a 5% threshold of the segmented volume. The accuracy of the automated segmentation was assessed as volumetric overlap, expressed as Dice scores.

Results: Tumor types included low/high-grade gliomas (LGG/HGG), diffuse midline gliomas (DMG), ependymomas (EP), medulloblastomas (MB), and other rare tumors, in various regions (hemisphere, brainstem, optic pathway, cerebellum, posterior fossa). Overall Dice scores (median ± SD) were: T2H 0.85 ± 0.18, ET 0.75 ± 0.34, CC 0.52 ± 0.32.

Results by location

- Posterior Fossa: T2H 0.87 ± 0.1, ET 0.79 ± 0.4, CC 0
- Cerebellum: T2H 0.73 ± 0.3, ET 0.79 ± 0.4, CC 0.86 ± 0.01
- Hemispheric: T2H 0.75 ± 0.36, ET 0.55 ± 0.23, CC 0
- Brainstem: T2H 0.9 ± 0.05, ET 0.8 ± 0.4, CC 0
- Other: T2H 0.85 ± 0.09, ET 0.6 ± 0.2, CC 0.49 ± 0.3

Results by tumor type

- LGG: T2H 0.78 ± 0.3, ET 0.71 ± 0.3, CC 0.7 ± 0.4
- HGG/DMG: T2H 0.89 ± 0.1, ET 0.74 ± 0.4, CC 0
- EP: T2H 0.89 ± 0.27, ET 0.63 ± 0.44, CC 0
- MB: T2H 0.86 ± 0.06, ET 0.76 ± 0.4, CC 0
- Other: T2H 0.84 ± 0.07, ET 0.86 ± 0.4, CC 0.5 ± 0.3

Conclusion: The automated segmentation method showed high accuracy for T2H and ET, which are critical for surgical and radiotherapy planning, as precise delineation of these regions directly impacts pBTs' grading, treatment response prediction and prognosis. Lower accuracy for CC segmentation was due to limited representation in the dataset and variability in cystic component across tumor types. Expanding the dataset and addressing this variability could improve performance, while the current focus on T2H and ET ensures significant clinical utility.

SPN 7

Trajectories of Behavioural Difficulties in Children with Complex Congenital Heart Disease: A Longitudinal Study from 4 to 13 years

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Children with complex congenital heart disease (cCHD) are at risk for neurodevelopmental and behavioural difficulties, yet little is known about behavioural problems across developmental stages. This study aimed to investigate behaviour in children with cCHD from preschool to adolescence and to identify predictors for behavioural problems.

Children with cCHD who underwent infant cardiopulmonary bypass surgery were prospectively enrolled at the University Children's Hospital Zurich. Behaviour was assessed with the parent-reported Strengths and Difficulties questionnaire at 4, 6, 10, and 13 years. Age-adjusted total and domain scores were compared to normative means using Wilcoxon-rank-sum tests. Latent class growth analysis (LCGA) identified subgroups with distinct trajectories of total behavioural problems. Regression analysis examined demographic, cardiac, neurodevelopmental, and family predictors of behavioural problems. Self-reported quality of life (QoL) and special educational needs (SEN) in adolescence were compared between those with and without problematic behavioural trajectories using t-tests and Wilcoxon-rank-sum tests.

A total of 137 children with cCHD were included. Behavioural outcomes were normal on average at 4 and 10 years, but parents reported more emotional problems at 6 years ($p = 0.010$) and more emotional problems ($p < 0.001$), hyperactivity ($p = 0.045$), and total behavioural problems ($p = 0.040$) at 13 years. LCGA identified three behavioural trajectories: Class 1 demonstrated no behavioural problems across ages (n = 76, slope: $p = 0.28$); Class 2 showed early behavioral problems that improved with age (n = 25, slope: $p < 0.001$); Class 3 had increasing problems with age (n = 36, slope: $p < 0.001$). Lower child IQ (OR = 1.1, $p = 0.020$) and poorer maternal mental health (OR = 0.9, $p = 0.004$) at 4 years predicted behavioral problems. CHD severity and demographics were not significant predictors. Children with behavioural problems had more SEN ($p < 0.001$) but did not significantly differ in QoL ($p = 0.059$).

This study shows that behavioural problems in children with cCHD become more apparent during adolescence in a subgroup of patients and are associated with educational attainment. Higher IQ and better maternal mental health predict favourable behavioural outcomes, whereas CHD severity appears less relevant. These findings underscore the importance of neurodevelopmental follow-ups and family-centred care to identify at-risk individuals and promote long-term outcomes in cCHD.

SPN 8

Heart Rate Variability and Perioperative Care in Neonates with Transposition of Great Arteries: A Single-Centre Retrospective Analysis of 24h Holter Electrocardiogram Pre-Hospital Discharge

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Background: Transposition of the great arteries (TGA) represents 5-7% of all congenital heart diseases (CHDs). It predisposes neonates to cerebral hypoxemia, cyanosis, and altered autonomic nervous system (ANS). Despite improved survival after arterial switch operation (ASO), challenges such as reduced exercise tolerance and severe cardiac events persist. The association of perioperative factors with ANS function is still poorly investigated. The ANS function can be noninvasively assessed using heart rate variability (HRV), measured via Holter Electrocardiogram, as a proxy. HRV is a measure of the temporal fluctuation between successive heartbeats and provides information about the balance and adaptability of the ANS, which regulates stress, recovery and cardiovascular health. HRV includes metrics from time-domain and frequency-domain analyses, which offer detailed assessments of overall and short-term variability and nonlinear parameters.

Aims: This study investigates the HRV characteristics of neonates with TGA after ASO and evaluates the association of perioperative factors, such as bypass time and cumulative drug dosages, on HRV outcomes. We expect to identify key predictors of reduced HRV, shedding light on ANS dysregulation in this population. Hypothesis. It is hypothesized that perioperative factors such as prolonged bypass time and higher cumulative drug dosages will be associated with reduced HRV.

Methods: We analyzed pre-hospital discharge 24-hour Holter ECG recordings of neonates with TGA who underwent ASO (N = 83) between January 2015 and 2024 at the University Children's Hospital of Zurich. Patients were excluded if the gestational age was <30 days of age, died during hospitalization, received medications primarily influencing heart rate during ECG recording, or lacked consent for data use. ECG recordings were preprocessed and manually reviewed to remove artefacts, using only intervals between sinus beats to calculate HRV metrics. We used linear models to assess the association of perioperative factors and covariates on HRV parameters. Correlation analyses were performed to evaluate the relationship between HRV and clinical variables.

Results: Statistical analysis is ongoing, but preliminary analysis already suggests a significant correlation between certain perioperative factors and HRV. We are confident we can present our findings at the "Pädiatrie Schweiz" Congress 2025.

SPN 9

Parental smoking in children consulting for respiratory diseases in Switzerland

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Background: It is well known that exposure to environmental tobacco smoke (ETS) negatively affects children's health and worsens symptoms in children with respiratory diseases. We studied parental smoking prevalence, characteristics, and changes over one year among families in the Swiss Paediatric Airway Cohort (SPAC).

Methods: SPAC includes children aged 0–17 years referred to paediatric outpatient respiratory clinics between 2017 and 2024. Parents answered a questionnaire with questions on smoking at the initial clinic visit and again after one year. We analyzed parental smoking prevalence, socio-demographic characteristics associated with smoking, and changes in smoking behavior over one year using descriptive statistics and multivariable logistic regression.

Results: Among 4,199 children (median age 8.5 years [IQR 5–12], 60% male), 31% were exposed to parental smoking at baseline (paternal smoking: 16%; maternal smoking: 6%; both parents smoking: 9%). Factors associated with parental smoking included compulsory education only (OR 2.0, 95%CI 1.6–2.5 vs university education), non-Swiss nationality (OR 1.3, 1.0–1.6) and living in a socially disadvantaged neighborhood (OR 1.3, 1.0–1.7), with all given estimates for mothers and slightly higher values for fathers. The strongest association was observed for having a partner who smokes, with odds ratios above 6 for both parents. Additionally, fathers who smoked were more likely to be unemployed (OR 2.0, 1.3–3.2 vs full-time job). At one-year follow-up, smoking data from 2226 mothers and 1895 fathers showed that 225 (10%) of mothers and 382 (20%) of fathers continued smoking, 47 (2%) of mothers and 54 (3%) of fathers started, and only 63 (3%) of mothers and 90 (5%) of fathers quit.

Conclusions: One-third of children with respiratory diseases are exposed to ETS by their parents. Even after having visited a tertiary clinic for their children's respiratory problems, most parents continue to smoke. This highlights an urgent need for enhanced public education and stronger efforts to reduce smoking across the whole population. Additionally, we need to find effective approaches how to support parents of children with respiratory diseases to quit smoking and maintain a smoke-free lifestyle.

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SPN 10

Empagliflozin in paediatric heart failure: model-based optimization of a pharmacokinetic bridging study

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Introduction: Current therapy for paediatric heart failure is unsatisfactory, with 5-year mortality of 30-50%. Trials in this population have often failed, mainly because of sample size issues, suboptimal dose, inappropriate formulations, and issues in choosing the best-suited outcomes. We apply a model-based approach to optimize the study design and increase the probability of success of a prospective trial aimed at establishing the dose rationale for empagliflozin in children with heart failure.

Methods: A nonlinear mixed effects approach incorporating prior information from pharmacokinetics (PK) in adults was used to extrapolate empagliflozin disposition parameters to children and to optimize the study design. Protocol elements of interest were dose, sample size and sampling schedule. These features were explored using a combination of an optimization algorithm ($DESIGN) and a simulation re-estimation procedure (SSE) in a large virtual paediatric cohort, overcoming some of the difficulties associated with small populations. Comparable pharmacokinetic-pharmacodynamic relationship was assumed between adults and children.

Results: A two-compartment PK model with sequential zero- and first-order absorption, absorption lag-time and first-order elimination was identified. Clearance and distribution parameters were assumed to vary allometrically with body weight. We identified a lowest safe weight of 15kg as inclusion criterion for the prospective trial, achieving, with the lowest commercially available tablet of 10mg, a median AUC ratio of 1.03 (interquartile range 0.82-1.30) relative to a 50kg adult receiving the 25mg dose (median 7163, IQR 6115-8338 nmol*h/L). The optimized sampling scheme with 12 patients based on a sampling matrix with four different groups allowed precise estimates of primary and secondary PK parameters. These results justify the relatively small sample size in a prospective study aimed at characterizing empagliflozin exposure, and exploring safety and efficacy in the population of interest.

Conclusion: Repurposing of drugs for paediatric rare diseases is fraught with challenges. This simulation exercise identified the lowest safe weight for inclusion, and developed a sampling matrix able to maximize the reliability of the delivered PK information whilst minimizing the number of patients and samples needed. Extrapolation approaches allow to optimize evidence generation in clinical trials, whilst reducing patient burden.

SPN 11

Childhood cancer survivors' perceptions of a low-threshold screening program for hearing loss: a qualitative study

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Purpose: Childhood cancer survivors (CCS) are at risk for chronic health problems, including hearing loss after ototoxic treatments. Follow-up care clinics screen for incident health problems such as hearing loss. However, many adult CCS do not attend long term follow-up care due to barriers such as accessibility and emotional concerns associated with revisiting medical facilities. To address those barriers, the HEAR-study piloted a new low-threshold hearing screening program leveraging the extensive network of hearing aid shops across Switzerland. As part of this program, childhood cancer survivors completed audiometric screening at a hearing aid shop. In case of marginal or pathological results, the hearing aid shop advised them to consult with a medical doctor. We explored CCS' perceptions of this novel screening program.

Methods: We conducted semi-structured interviews with 29 participants who completed a hearing test at a hearing aid shop as part of the HEAR-study. We used thematic analysis of interview transcripts, supported by MAXQDA for data analysis.

Results: Thematic analysis revealed two key themes: First, participants appreciated the program's practicability, highlighting the efficiency and ease of integrating it into daily life. At the same time, some noted concerns about the hearing test being an additional appointment. Some survivors would prefer centrally organized follow-up care, where different examinations to screen for chronic health conditions are done at the same location within the same day, ideally consulting with one person regarding all results. Second, many participants valued the personal and approachable environment at the hearing aid shop as a relaxed alternative to medical facilities, while others would prefer hearing tests as part of an appointment with physicians, valuing immediate advice and contextual knowledge, especially in case of pathological findings.

Conclusions: From CCS' perspectives, this screening program shows promise as a practicable and accessible way to evaluate hearing after childhood cancer.

SPN 12

Registry-based frequency of molecularly confirmed Osteogenesis Imperfecta in a Swiss cohort of individuals with connective tissue disorders

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Introduction: This study evaluates the distribution, genotype-phenotype correlations, and intra-/interfamilial variability in Osteogenesis Imperfecta (OI) cases within a Swiss registry of connective tissue disorder (CTD) patients.

Patients/methods: We analyzed data from the first Swiss CTD registry, comprising 796 patients referred to our center between 1995 and 2022. The registry includes information on clinical history, anthropometric data, biochemical findings, histological results, and genetic analyses. We further analyzed a subcohort of patients with a molecularly confirmed diagnosis of OI to explore genotype-phenotype correlations, mutation severity, and inter-/intrafamiliar variability.

Results: Among 796 patients, the most frequent referrals were for Ehlers-Danlos syndrome (EDS) (n = 427) and OI (n = 224). Genetic testing confirmed diagnoses in 60 EDS and 98 OI cases. In-depth genetic analysis of 173 OI patients revealed that the majority carried dominant mutations in COL1A1 or COL1A2, whereas a minority presented with rarer recessive variants in genes affecting collagen synthesis and bone regulation. Of 98 genetically confirmed OI patients, 52 were followed longitudinally. The highest fracture incidence (≥20 fractures) was observed in rare OI types (27.3%), followed by COL1A1 cases (19.2%) and COL1A2 cases (13.3%). The mean age at diagnosis was lowest in rare OI types. Pathogenic COL1A1 variants causing haploinsufficiency were associated with mild phenotypes, while severe/lethal OI cases involved mutations in the COL1A1 signal peptide, C-terminal propeptide and distinct Glycine substitutions within the collagen triple-helical domain of COL1A1 and COL1A2.

Conclusion/Discussion: This registry-based study provides insights into the genetic and phenotypic variability of OI. COL1A1 and COL1A2 mutations accounted for the most frequent genetic cause of OI. Clinical severity among these cases correlated with mutations in critical structural regions, while haploinsufficiency in COL1A1 resulted in milder phenotypes. These findings underscore the importance of genetic testing for precise diagnosis, prognosis, and management of OI.

SPN 13

Survey on Cancer Predisposition Identification & Observation Practice in Europe (SCOPE)?

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Introduction: Cancer Predisposition Syndromes (CPSs) are heritable genetic conditions associated with an increased risk of various cancers throughout life. Early identification and tumor surveillance can improve outcomes, but they are often underdiagnosed in clinical practice. This study investigates current practices of CPS identification and management among pediatric oncologists across Europe.

Methods: We developed a series of electronic surveys targeting healthcare providers caring for pediatric cancer patients. The surveys included three parts: demographics and overview, screening and workup, and surveillance and education.

Results: We collected 185 unique responses across 21 countries. Of the respondents, 57% had more than 10 years of experience, and 84% reported working at institutions diagnosing >100 new childhood cancer cases annually and 16% <30 cases per year. Of the respondents, 50.3% reported access to a dedicated CPS clinic in their own institution. More than half of participants reported low or unsure confidence in counseling patients and families (64%), and in utilizing germline genetic data (61%). Specific CPS training increased awareness and usage of validated screening tools, but more than 70% of respondents were either unaware or did not regularly use common tools to identify patients at increased risk of CPS. A significant proportion of respondents identified the need for more regular training sessions for healthcare professionals (79%), dedicated genetic counsellors for patient support (70%), and accessible patient-friendly educational materials (63%).

Conclusion: We found a high variability and important gaps in CPS identification and management practices across Europe. Our findings underline the need for standardized practices, improved education, and targeted training to bridge knowledge gaps and promote the use of effective screening tools.

POSTERS

P 1

Minimal Handling during Extubation of Preterm Infants in Prone Position: A Prospective Observational Study

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Objective: To evaluate changes in end-expiratory lung impedance (EELI), a surrogate for end-expiratory lung volume (EELV), during a minimal-handling extubation protocol in very preterm infants in prone position.

Methods: Prospective observational study in preterm infants born before 32 weeks gestation. The protocol aimed to minimize interruptions in positive end-expiratory pressure (PEEP) delivery. During the final nursing care before extubation, the non-invasive ventilation (NIV) interface was partially secured, and the adhesive tapes securing the endotracheal tube were loosened. A manipulation-free 'recruitment period' was introduced to promote lung volume recovery. Changes in EELI and cardiorespiratory parameters were compared to baseline.

Results: Fifteen extubations were analyzed. At NIV initiation, median EELI decreased by -0.68 AU/kg (IQR: -0.99 to -0.21), reflecting a median EELV loss of 27.7 ml/kg (IQR: -40.6 to -8.6). The greatest EELI loss occurred during tape removal (-0.24 AU/kg, padj = 0.025), while the largest recovery was during recruitment (0.31 AU/kg, padj = 0.025). Extubation duration was 1.7 min (IQR: 1.3–2.0), with a 2-second median interval (IQR: 1–4) between tube removal and NIV. Oxygen saturation remained stable (padj = 0.32).

Conclusions: Minimal handling did not prevent lung volume loss during extubation, but it was regained during a recruitment period.

Impact: This study evaluated lung volume changes during extubation of very preterm infants following the introduction of a new minimal handling extubation protocol. While the new protocol did not prevent substantial lung volume losses overall, a manipulation-free recruitment period immediately prior to extubation allowed for partial lung volume recovery. The protocol was associated with shorter procedural times and more stable oxygen saturation levels. Further studies are needed to improve minimal-handling strategies during the extubation process and to assess their impact on relevant clinical outcomes.

P 2

A Twisted Tale: Midgut Volvulus Unmasked by Yellow Vomiting

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Introduction: Volvulus is rare in infants but has high morbidity and mortality. Since vomiting is a common reason for medical visits and volvulus has subtle early signs, diagnosing it in the initial stages is difficult. We describe two cases of young infants with yellow vomiting and midgut volvulus who presented to our emergency department.

Case 1: A 3-week-old male presented with a one-day history of vomiting, now yellow in colour, and bloody stools. He appeared critically ill with clear signs of an acute abdomen. Volvulus was immediately suspected and confirmed on ultrasound. Laparotomy revealed necrotic bowel, requiring the resection of 40 cm of the small intestine. The infant developed functional short bowel syndrome, necessitating parenteral nutrition

Case 2: A 5-week-old female was referred to the hospital by a medical hotline due to vomiting. She presented 4 hours after her first yellow vomit, appearing well with a normal abdominal examination. A review in the early morning revealed marked abdominal tenderness in the context of repeated yellow vomiting, raising suspicion of volvulus. The diagnosis was confirmed on ultrasound, and an emergency laparotomy was performed. All bowel was preserved, and the patient discharged on day 12.

Discussion: Volvulus results from arrested embryonic gut rotation (malrotation), causing the bowel to twist around the mesenteric root, leading to ischemia. The true incidence of malrotation is unknown, as many cases remain asymptomatic, but contrast studies show it in 2 out of 1000 infants. Volvulus occurs in one-third of children with malrotation, mostly before age five (75%), with peaks in the first year (58%) and first month (30%). For stable patients, ultrasound has high sensitivity and specificity, while upper GI series remain the gold standard. In unstable patients, an explorative laparotomy is required. While diagnosing volvulus in critically ill neonates (e.g., Case 1) is straightforward, early detection remains challenging. There is a misconception that bilious vomiting is always green. In young infants, any colour other than white – especially fluorescent yellow – is a red flag. Improved awareness of yellow vomiting in our team led to the early detection of a third case of midgut volvulus, which presented after only two yellow vomits. Laparotomy confirmed the diagnosis, showing venous congestion but no ischemia. Early recognition of yellow vomiting as a sign of volvulus can save bowel and potentially a child's life!

P 3

Title: Case report: Newborn with ptosis with confirmed neonatal myasthenia gravis

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Background: About 10 to 20% of infants born to mothers suffering from myasthenia gravis develop transient neonatal myasthenia gravis. There are some case reports of infants with asymptomatic mothers.

Case report: During the one-month preventive pediatric examination, a plagiocephaly with preferential posture to the right was initially noticed in an otherwise normal development, apart from uncertain slight ptosis. In the following check-up at the age of 2 months, the ptosis was then clearly pronounced, more on the left than on the right, with otherwise unremarkable neuromotor development. The patient was referred to the clinic for further examinations. Ophthalmologically, the ptosis on the left was confirmed with a lateral difference of 2 mm, which, however, did not currently require any treatment as the visual axis was free. A cranial sonogram performed as part of the neurological examination revealed unremarkable findings. However, the laboratory results revealed a slightly higher than normal auto-antibody level for anti-acetylcholine receptor antibodies of 0.7nmol/l (norm <0.5 nmol/l), so that an additional neurography examination was performed. This revealed a typical decrement in muscle action potentials during repetitive stimulation, confirming the diagnosis of transient neonatal myasthenia gravis. Typical symptoms are furthermore general weakness

and hypotonia. We therefore attribute the plagiocephaly to an initially present slight weakness. Unfortunately the plagiocephaly did not improve, even though physiotherapy was already started at the age of 1 month and was accompanied by additional osteopathic/chiropractic and pediatric orthopedic care. Helmet therapy is now initiated to correct the asymmetric head development. In all other areas, development continues to be unremarkable and age-appropriate and antibody levels already showed a slight decline to 0.63 nmol/l in a follow-up examination at the age of 4 months.

Discussion: Transient neonatal myasthenia gravis is caused by placentally transmitted antibodies from mothers with myasthenia gravis, leading to a temporary disruption of the neuromuscular end plate. To date, however, no myasthenia gravis is known for our patients mother, but there have been described cases of asymptomatic mothers in literature. We assume a spontaneous complete relief of symptoms with vanishing antibody levels in the upcoming follow up appointments.

P 4

Teaching of Neonatal Resuscitation and its Impact on Neonatal Mortality in the Outskirts of Greater Conakry, Guinea, Western Africa

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Background: High neonatal mortality remains a major health issue in low- and middle-income countries. Guinea indicates a neonatal mortality rate of 32 /1'000 live births, representing close to 15'000 neonatal deaths annually, with no improvement over the past decade.

Objectives: To evaluate the impact on early neonatal mortality (within 6 hours after birth, due to the outpatient delivery management of these obstetric units) and the need for neonatal transfer of a 2-days neonatal resuscitation training for health professionals working in private peri-urban health facilities in the outskirts of Conakry. Souffle2vie and its guinean partners focused on the private sector as it is particularly dominated by informal structures not accounted for in the national health information system. Some structures are run by insufficiently trained and poorly equipped paramedical staff.

Methods: The intervention consisted in a theoretical and practical training and was offered to health professionals in the predefined area. At the end of the training, all centers were equipped with basic resuscitation devices (resuscitation bag, mask, suction tool, training book). A comparison of obstetric and neonatal activity over a 6-month period before and after the intervention was performed. Theoretical knowledge was assessed by a pre- and post-training test with a 17-item questionnaire, followed by a third evaluation 6 months after the training.

Results: 27 nurses, midwives, and doctors (1 to 3 per facility) participated in the study. The impact assessment of the training is based on data from 13 private health facilities of various sizes. These health facilities conducted 589 deliveries during the two analysis periods. The early neonatal mortality rate (6h postpartal) dropped from 31.8 to 5.7 per 1'000 live births. The need for neonatal transfer to a referral hospital decreased from 27.3‰ to 11.3‰. The stillbirth rate remained very high and unchanged during the two periods, with 26.5‰ before and 27.5‰ after the training. Theoretical knowledge improved sustainably, with an average of correct answers increasing from 10.1/17 correct answers (59.3%) before the training to 13.9/17 (82.0%) directly after the training, and remained high at 14.6/17 (85.9%) six months later.

Conclusion: Thanks to a two-day training for health professionals, the early neonatal mortality rate could significantly be decreased in the outskirts of Conakry. Similar programs are now planned and performed in the whole country.

P 5

Congenital Nephrotic Syndrome: From Hospital Management to Promising Home Care Solutions

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Background: Congenital nephrotic syndrome (CNS) is a rare kidney disorder characterized by heavy proteinuria, hypoalbuminemia, and edema starting soon after birth, with an incidence of 1–3 per 100,000 live births, reaching 1 per 8,000 in Finland due to the Finnish Type variant. Most cases are caused by mutations in NPHS1, NPHS2, WT1, LAMB2, or PLCE1 genes (>80%). CNS requires a multidisciplinary approach involving pediatricians, nephrologists, nutritionists, hematologists, and other specialists.

Cases: Two clinical cases illustrate the complexity of CNS. The first involves an 18-month-old girl initially presenting with anasarca, an albumin level of 6 g/L, feeding difficulties, and coagulation issues, with challenging adaptation to vitamin K antagonist (no thrombosis), with a mutation in NPHS2 (podocin). The second case describes a 1-month-old boy diagnosed because of an initial facial edema with a finnish type CNS (NPHS1). He presented with a minimum albumin level of 11 g/L, in the follow-up he developed renal and jugular vein thrombosis related to a central venous catheter, despite prophylactic anticoagulation, alongside significant feeding difficulties and severe respiratory infections.

Management: Initial hospital care required close monitoring to stabilize the disease, assess renal function, and adjust treatments. Complications such as thrombosis necessitated careful anticoagulation management, with strong collaboration among multiple specialties. After stabilization, both patients transitioned to home care, which included daily monitoring of vital parameters, frequent blood draws, daily administration of IV treatment such as albumin infusions. Previous studies have shown that home-based albumin infusions are feasible for managing congenital nephrotic syndrome. This home care was supported by regular communication with the hospital's medical team, and regular in-hospital consultations, ensuring timely treatment adjustments.

Conclusion: While the management of congenital nephrotic syndrome remains complex, home care is feasible for the most complicated cases, provided there is close collaboration between hospital-based and home-care physicians and nurses. This coordination ensures rigorous monitoring, treatment adjustments, and offers families essential support for the daily management of this rare condition.

P 6

Impact of Intraventricular Hemorrhage on Cognitive and Motor Outcomes in Extremely Preterm Infants: up date from the Neonet

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Background: Premature neonates, particularly those born before 28 weeks of gestation, are at a high risk of intraventricular hemorrhage (IVH), a common complication associated with increased mortality and neurodevelopmental impairments. Advances in neonatal care have improved survival rates, necessitating updated insights into long-term outcomes.

Objective: To assess the impact of IVH on cognitive and motor outcomes, including cerebral palsy (CP), in extremely preterm infants at two years of corrected age.

Methods: This multicenter, prospective observational study analyzed data from the Swiss Neonatal Network & Follow-up Group (SwissNeoNet) for preterm infants (<28 weeks gestation) born between 2012 and 2019. The severity of IVH was classified using cranial ultrasound, and outcomes were assessed using the Bayley Scales of Infant Development, 3rd Edition (Bayley-III). CP incidence and severity were evaluated using the Gross Motor Function Classification System (GMFCS).

Results: Of 1,307 infants analyzed, 377 (28.8%) had IVH, with grades 1–4 distributed as 10%, 9.6%, 4.4%, and 4.5%, respectively. High-grade IVH (grades 3–4) was strongly associated with adverse cognitive, motor, and language outcomes, with motor function being most affected (16-point reduction in Bayley scores for grade 4 IVH). CP incidence increased with IVH severity, reaching 58.9% in grade 4 IVH (p <0.001). However, 41% of grade 4 IVH cases and 74% of grade 3 IVH cases did not develop CP. Among children with CP, cognitive outcomes were significantly worse than in those without CP.

Conclusion: High-grade IVH is a major risk factor for neurodevelopmental impairments, particularly motor dysfunction and CP. Despite this, a substantial proportion of infants with severe IVH achieve favorable outcomes, emphasizing the importance of individualized follow-up and targeted interventions.

P 7

Evaluation of nine population pharmacokinetic models for model-based vancomycin concentration monitoring in a local population of neonates

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Background and objective: To optimize vancomycin use and enhance therapeutic drug monitoring (TDM) in neonatology, model-informed precision dosing (MIPD) using suitable population pharmacokinetic (PopPK) models is proposed. We aimed to evaluate the potential suitability (predictive performance) of published PopPK models for vancomycin concentration prediction in our local population of neonates.

Methods: Retrospective study analysing vancomycin measurements from neonates treated between 2018-2023 at the University Children's Hospital Basel. Nine models predicting PopPK of vancomycin in neonates were evaluated using Tucuxi software. Initial vancomycin measurements were used in combination with PopPK models to estimate individual pharmacokinetic parameters. Those individual pharmacokinetic parameters were then used to predict initial concentrations and to forecast subsequent 2nd concentration measurements. Predictive performance was evaluated regarding bias (mean prediction error) and precision (root mean squared error). Secondary outcomes included target exposure achievement (nationally defined as trough concentration 10-20 mg/L), TDM timing and safety/effectiveness-related clinical outcomes.

Results: A total of 78 vancomycin level measurements of 32 neonates (median weight: 1834 g, post-menstrual age: 31.9 weeks) were collected. Lowest bias (<7%) and highest precision (20-32% for initial measurements, 42-44% for forecasted 2nd measurements) was obtained by PopPK models from Mehrotra 2012 and Frymoyer 2019. Nationally recommended target trough concentration was reached in 41/78 (52.6%) of measurements. Initial measurements were taken at steady state in 17/32 (53%) of cases. Acute kidney injury and infection-related mortality were observed in 2 neonates each (6.25%).

Conclusion: MIPD-guided vancomycin concentration monitoring requires careful local evaluation before implementation. We identified two potentially suitable PopPK models that may allow to handle non-steady-state measurements and predict area-under-the-curve as emerging preferred exposure metric. There is room for improvement in neonatal vancomycin use and monitoring, involving clarification of proposed targets in neonates.

P 8

Incidence, subtypes and severity of cerebral palsy in infants born extremely preterm in Switzerland: A retrospective study comparing two time periods

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Background: In 2011, the Swiss Society of Neonatology issued new guidelines for the care of preterm infants at the limit of viability leading to higher survival of infants born <28 weeks of gestation. It is unclear whether and how these recommendations affected the prevalence and severity of cerebral palsy (CP).

Objective: To investigate whether the prevalence, severity, and subtypes of CP in extremely preterm infants differ in two successive birth cohorts.

Methods: Retrospective, population-based analysis of prospectively collected data on infants born <28 weeks of gestation. CP prevalence, subtypes (Surveillance of cerebral palsy in Europe – SCPE classification), and severity (Gross Motor Function Classification System - GMFCS) assessed at 2 years corrected were compared between the two birth cohorts (2006-2011 and 2012-2017).

Results: Of 3244 registered infants, 2090 survived, of whom 1764 were followed up (84%). Mortality was 38% for the first period and 33% for the second and 112 were diagnosed with CP. CP prevalence was 34.5 per 1000 live births (37.0 for 2006-2011 and 32.4 for birth-years 2012-2017, p = 0.476). A trend towards more bilateral spastic CP (2006-2011: 32% and 2012-2017: 50%, p = 0.055) and more severe cases (2006-2011: 14.3% and 2012-2017: 24.9%, p = 0.154) was observed in the second period. CP severity was associated with cystic periventricular leukomalacia (PVL) (OR 3.4, 95%-CI 1.1-10.3, p = 0.033) and necrotizing enterocolitis (NEC) (OR 5.5, 95% CI 1.2-25.1, p = 0.028) but not with other neonatal morbidities.

Conclusion: These results suggest that the greater number of bilateral forms and severe cases of CP could be due to the higher number of surviving infants in the 2011-2017 cohort. PVL and NEC are the factors mostly associated with severe cases of CP in Switzerland.

P 9

A Bump In The Road

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Case report: A 45 day old infant was admitted to Geneva Children's Hospital following the spontaneous appearance of swelling of the occiput noticed 24 hours earlier by the mother. It was absent at the maternity ward and at prior pediatrician checkups. No accompanying symptoms present, notably no neurological symptoms, alteration of consciousness or signs of intracranial hypertension. The infant had normal growth metrics for age, was born full term by C-section delivery with a difficult fetal extraction without instrumentation. No history of traumatic brain injury, no family history of altered coagulation. She presented with stable vital signs, a normal neurological examination, a tender and fluctuating right parieto-occipital mass measuring 5x3 cm without skin discoloration. An ultrasonography showed a parietal paramedian anechoic fluid located in the subgaleal space, crossing suture lines. The coagulation workup was normal. Because of a hypothesis of subgaleal hematoma raising the differential diagnosis of non-accidental trauma, she was hospitalised for neurological and hemodynamic monitoring over 48 hours. She was treated conservatively. The swelling self-resolved after 3 weeks.

Discussion: A delayed subaponeurotic fluid collection of infancy is rare and occurs spontaneously between 3 weeks and 4 months. It is typically localized in the superior occiput, presenting as a fluctuating mass crossing suture lines without skin discoloration, bruising nor pain upon palpation. The diagnosis is primarily clinical. No additional workup is required. Imaging, if performed, reveals anechoic fluid in the subgaleal space, not likely compatible with acute hemorrhage. The underlying cause is idiopathic, most likely linked to minor obstetrical trauma leading to slow leakage of lymphatic, capillary or cerebrospinal fluid. The collection is self-limiting and resolves spontaneously within 1 to 2 months.

Conclusion: An unexplained tender occipital collection appearing after 3 weeks of age should raise the suspicion of head trauma and potential non-accidental injury. If the clinical presentation is typical however,the differential diagnosis of delayed subaponeurotic fluid collection of infancy must be considered. Given the clinical similarity to subgaleal hematomas, healthcare professionals must be cautious in distinguishing between these two conditions to avoid unnecessary work-ups, hospitalizations, and the emotional burden parents may face when suspected of child abuse.

P 10

Recurrent Pseudomonas aeruginosa Meningitis in an ELBW Infant: A Case Report on the Use of Intrathecal Colistin

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We report a rare and severe case of recurrent meningitis caused by Pseudomonas aeruginosa in an extremely low birth weight (ELBW) preterm infant. This case highlights the challenges of early detection, effective treatment strategies, and the management of complications, including cerebral venous sinus thrombosis (CVST), hydrocephalus and intraventricular hemorrhage (IVH). Despite the severity of the infection and associated complications, a favorable outcome was achieved, demonstrating the potential role of intrathecal Colistin as a safe and effective rescue therapy in recurrent neonatal meningitis.

P 11

Pediatric Respiratory Syncytial Virus rehospitalization rate – relevance for passive immunization strategies with monoclonal antibodies

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Background: Now that highly effective, long-acting monoclonal antibodies against Respiratory Syncytial Virus (RSV) are available for prevention of severe disease including RSV hospitalization in children below two years of age, data on the risk of RSV rehospitalization among children, who previously had a severe first RSV episode, remain important to inform the need for secondary prevention using (another) dose of such an antibody. We studied the risk of RSV rehospitalization in a large cohort of patients with a particular focus on same-season rehospitalizations.

Methods: Retrospective single-center study of all RSV rehospitalizations occurring in 13 RSV seasons between 2009 and 2023 based on an ongoing RSV surveillance program. RSV rehospitalization was defined as a virologically confirmed RSV hospitalization beginning >30 day after discharge from a previous RSV hospitalization. Overall and same-season rates of rehospitalizations for patients of any age below 16 years and for the first 5 years of life, respectively, were calculated. Clinical characteristics of primary hospitalizations and rehospitalizations were compared.

Results: In a cohort of 3'143 patients having had a primary RSV hospitalization, the overall risk of rehospitalization (69 cases) and the same-season risk of rehospitalization (2 cases) for a second RSV infection were 2.2% (95% confidence interval (CI), 1.73-2.79) and 0.06% (95% CI 0.02-0.23), respectively. The respective figures for RSV rehospitalization rates from birth until age 5 years of age were 2.3% (95% CI 1.76-3.07) and 0.04% (95% CI 0.01-0.25). The median length of stay (LoS) of rehospitalizations (4.0 days, interquartile range (IQR) 3.0-6.0) was significantly shorter than the LoS of first hospitalizations (6.0 days, IQR 4.0-9.0, p <0.0001). Children with a pre-existing health

condition (68%) and those born prematurely (40%) predominated among rehospitalized patients.

Conclusion: Same-season RSV rehospitalizations were exquisitely rare. Routine administration of a dose of a monoclonal antibody for protection against a same-season rehospitalization is not generally warranted. The majority of patients with subsequent-season rehospitalizations would be covered by the current Swiss recommendations for second-season RSV prophylaxis.

P 12

The effects of Nirsevimab on the number of children admitted with respiratory syncytial virus infection at the University Children's Hospital Bern

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Background: Historically, we have observed a biennial cycle of weak and strong respiratory syncytial virus (RSV) seasons at the University Children's Hospital Bern, based on hospital admission numbers. This cycle was disrupted by non-pharmaceutical interventions during the COVID-19 pandemic. After two unusually strong winter seasons in succession, winter 2024/2025 is the first season with general availability of Nirsevimab, a highly effective, long-acting monoclonal antibody against RSV, for infants in Switzerland. In this analysis, we compared hospital admission numbers during the 2024/2025 winter season to historical data prior to the COVID-19 pandemic.

Methods: Retrospective, single-center study of children admitted to the Inselspital Bern with a RSV infection during 12 winter seasons. We compared weekly admission numbers between 2008 and 2019 to the 2024/2025 winter season. RSV hospitalization was defined as hospital admission with virologically confirmed RSV infection. We used a Negative Binomial regression model to analyse weekly admission numbers with season strength and week of admission as independent variables.

Results: Prior to the COVID-19 pandemic on average 151 (IQR 131-163) infants were hospitalised in weak and 204 (185-217) in strong seasons. As of week 4 in 2025, 38 infants have been hospitalised in the 2024/2025 winter season. Compared to previous weak seasons, we observed an average reduction of weekly admission numbers in infants by 45% (95%CI 15 – 65%, p = 0.008) and by 60% (38 – 74%, p <0.001) compared to strong seasons. In contrast, we admitted more children between 1 and 2 years and children older than 2 years compared to previous weak seasons (+58% [6 – 129%, p = 0.02] and +73% [5 – 175%, p = 0.03], respectively), while there was no significant change compared to previous strong seasons in older children.

Conclusion: In the first winter season with general availability of Nirsevimab, we have, so far, seen a reduction in the number of infants admitted with RSV infection. Based on comparison with historical data, these preliminary data support that this reduction is due to the effects of general RSV prophylaxis and not only explained by alternating season strength.

P 13

Impact of Nirsevimab vaccination on hospitalization for Bronchiolitis: An Observational Study at Rennaz Hospital (December 2024–April 2025)

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Introduction: Bronchiolitis, an acute respiratory infection primarily caused by respiratory syncytial virus (RSV), leads to symptoms such as wheezing, coughing, and respiratory distress. It is a major cause of pediatric hospitalizations during the winter season. Nirsevimab is a monoclonal antibody recently introduced to prevent RSV infections. This study evaluates the impact of nirsevimab vaccination on the frequency and severity of bronchiolitis hospitalizations, while considering risk factors such as prematurity, exposure to tobacco smoke, and attendance in group childcare.

Methods: This prospective observational study includes pediatric patients hospitalized for bronchiolitis at Riviera-Chablais Hospital between December 2024 and April 2025. Each patient is tested to identify the viral cause of bronchiolitis, with a focus on RSV-related cases. The key variables analyzed include vaccination with nirsevimab, the time interval between vaccination and RSV-related bronchiolitis, and the severity of hospitalizations (e.g., hospital stay length, need for oxygen therapy). Risk factors such as tobacco smoke exposure, prematurity, and group childcare attendance are also evaluated.

Results: It is hypothesized that vaccination with nirsevimab will reduce hospitalizations due to bronchiolitis and the severity of the sickness, particularly by shortening hospital stays and decreasing the need for oxygen therapy. However, risk factors such as tobacco smoke exposure and prematurity may still significantly influence the occurrence and severity of bronchiolitis. As the study is ongoing, these results are projections.

Conclusion: This study aims to provide insights into the effectiveness of nirsevimab in reducing RSV-related hospitalizations and to highlight the impact of other risk factors in bronchiolitis management. It also seeks to explore the relationship between vaccination timing and the onset of bronchiolitis, offering valuable data for optimizing prevention strategies.

P 14

Tuberculosis in children – a diagnostic challenge

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Background: In Switzerland, approximately 550 individuals each year become infected by mycobacterium tuberculosis. According to WHO estimates, more than 50% of childhood tuberculosis cases are missed. Diagnosing tuberculosis in children remains a challenge due to the non-specific symptoms and the low concentration of bacilli. Often, bacteriological confirmation is complicated by the impossibility to collect sputum.

Case presentation: A 13-years old girl was admitted to our emergency department due to stomachache and elevated inflammation markers six days after she immigrated from Vietnam. One day later, community acquired pneumonia (CAP) was diagnosed. The initial x-ray showed no findings indicative for tuberculosis and no exposure to tuberculosis was reported, hence therapy with amoxicillin was initiated. Despite this therapy, inflammation markers increased further (CRP 156 to 209mg/l) and she developed pleural effusion. Quantiferon testing was positive. In the pleural effusion, as well as the gastric fluid, presence of mycobacteria was confirmed. Quadriple empirical tuberculostatic therapy was started. A 4-years old

asymptomatic boy from Eritrea was referred for tuberculosis diagnostic due to exposure to an index patient. The night before diagnostic sampling was planned, he developed coughing, fever, and tachypnea. He was admitted to the hospital for parenteral antibiotic treatment for the diagnosed CAP. Tuberculin skin testing was positive, so that by newly symptomatic child tuberculostatic therapy was initiated after asservation of gastric aspiration. Later, the presence of mycobacteria tuberculosis in the gastric fluid was confirmed.

Conclusion: Despite the low case numbers in Switzerland, tuberculosis needs to be considered as differential diagnosis, especially in children with known exposure or atypical clinical course of pneumonia as well as children from low income countries. Often, culture of expectorated or induced sputum is not possible in children, but detection in gastric fluid presents a valid alternative.

P 15

A rare complication of otitis media: Luc's abscess

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Introduction: Acute Otitis Media (AOM) is often a self-limited disease; however, there are significant possible complications due to its proximity to the intracranial and intratemporal compartments. In 1913, Henri Luc first described Luc's abscess as a rare extracranial complication of otitis media, caused by the spread of the middle ear infection, resulting in a subperiosteal collection beneath the temporalis muscle.

Case report: We report a case of a 4-year-old patient with a one-week history of bilateral AOM treated with oral Cefuroxime, who presented to the emergency department with complaints of worsening fever, otalgia and trismus. The clinical examination showed a swollen and tender lesion in the left temporal region, no signs of mastoiditis nor adenopathy were observed. Laboratory testing revealed increased C-reactive protein (CRP: 30 mg/L). In view of the clinical progression, a parenteral treatment with Amoxicillin-Clavulanate was started. Ultrasonography of the swollen region was uninformative, whereas MRI and subsequent CT-scan of the temporal region showed a purulent collection contiguous to the skull plate of the temporal bone, in the context of left otomastoiditis and reactive pachymeningeal tissue. Based on the radiological findings, we opted for a conservative approach by changing our choice of antibiotic to a dual therapy of Ceftriaxone and Metronidazole, without resorting to surgical drainage. The child showed rapid improvement, with gradual resolution of the preauricular swelling and stable defervescence. Follow up MRI after 22 days of treatment revealed a complete abcess resolution.

Conclusion: Luc's abscess is a possible complication of AOM. In contrast to other subperiosteal abscesses related to AOM, the infection may not be associated with mastoid bone involvement. It is classified as a benign complication; however, its infrequent occurrence may result in delayed diagnosis and treatment. Temporal bone CT is crucial to assess the extent of the abscess and guide management, avoiding unnecessary mastoid surgery. Management should be determined on a case-by-case basis, considering clinical assessment, radiological findings, and counseling, especially in pediatric patients.

P 16

Severe Mycoplasma-Induced Rash and Mucositis (MIRM): A Case Report

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Background: Mycoplasma pneumoniae is a common pathogen causing pneumonia, particularly in pediatric age group. Mucocutaneous manifestations occur in up to 25% of cases. Mycoplasma-Induced Rash and Mucositis (MIRM), first characterized as a distinct clinical entity in 2015, is marked by pronounced mucositis and milder skin lesions. Although 80% of patients experience full recovery, early recognition and interdisciplinary management are vital to prevent long-term complications. Prompt treatment with antibiotics and immunosuppressive agents can reduce morbidity.

Case presentation: An 11-year-old girl with no significant medical history presented with cough, fever, and mucositis. Painful lesions on the lips, tongue, and pharynx caused severe dysphagia, making oral intake impossible. Additional symptoms included conjunctivitis, a burning genital sensation, and a rash on her chest. Chest X-ray revealed signs of atypical pneumonia, and PCR of a nasopharyngeal swab confirmed M. pneumoniae infection. Clinical findings supported the diagnosis of MIRM.

Treatment: Treatment was initiated with intravenous doxycycline (100 mg twice daily for 5 days) and methylprednisolone (2 mg/kg once daily for 5 days). Topical steroids were applied to the affected skin, conjunctivae, and genital areas. Due to profound dysphagia, nasogastric tube feeding was initiated, resulting in refeeding syndrome, which was managed with daily electrolyte monitoring and supplementation. Oxygen therapy was administered for respiratory support. Despite this treatment, severe mucosal involvement persisted, necessitating escalation with a single subcutaneous dose of etanercept (50 mg), which led to gradual clinical improvement. With the aid of logopedic therapy, the patient regained the ability to eat and drink, and was discharged in stable condition

Discussion: This case illustrates the clinical severity of MIRM and emphasizes the importance of prompt diagnosis, appropriate antimicrobial and immunosuppressive therapies, and multidisciplinary care. Furthermore, it highlights the potential role of TNF inhibitors like etanercept in refractory cases, contributing to the growing evidence for targeted therapeutic strategies in MIRM.

P 17

Chronic non-bacterial osteomyelitis (CNO) in children: typical end atypical manifestation.

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Introduction: Chronic non-bacterial osteomyelitis (CNO) is a rare autoinflammatory bone disorder that usually affect metaphysis of long bones and axial skeleton. Inflammatory lesions usually appears as ill-defined bone marrow oedema at the MRI. In several cases, inflammatory lesion can show an atypical and more aggressive radiological pattern.

Aim: To describe clinical presentation, radiologic aspects and response to treatment of five patients with typical and atypical radiological manifestation, diagnosed with CNO at the Istituto Pediatrico della Svizzera Italiana.

Methods: Retrospective chart review of five patients diagnosed with CNO since 2016. Demographic data, clinical manifestation, laboratory values at onset, radiological features, treatment and response to treatment were reviewed. In addition, we wanted to address the possible differential diagnoses in respect to the particular localisation and the radiologic aspect for each case.

Results: Out of 5, 3 patients were females and 2 males. The median age at diagnosis was 10.3 years. The most frequent symptoms were pain and fatigue. The most frequent localisation were sacrum and tibia, the median number of affected bones per patient was 4. All five cases had an asymmetrical and multifocal disease course. Laboratory findings were nonspecific, with mild increase in inflammatory parameters. All patients underwent Whole Body MRI with asymptomatic inflammatory foci detected in all patients. Differential diagnoses evoked in respect of localisation and MRI appearance were: stress reaction, chronic regional pain syndrome, infectious osteomyelitis, leukaemia, lymphoma, Langerhans cell histiocytosis, tuberculous osteomyelitis, Ewing sarcoma, osteosarcoma. A bone biopsy was performed on four patients in order to exclude other conditions, and was negative. Six different medication classes have been used in five patients: NSAIDs, steroids, DMARDs, anti-TNF agents and bisphosphonates. Median follow up was of 5 years with resolution of symptoms in all cases.

Conclusions: CNO is a rare disease with still a long diagnostic delay, due to the low knowledge on this condition and the absence of specific clinical signs and pathognomonic diagnostic tests. WB-MRI is a crucial tool from a diagnostic point of view as well as for the follow up of CNO patients. In case of atypical radiological pattern it is important to exclude other benign or serious conditions, to institute proper therapy.

P 18

Paediatric Coxiella burnetii endocarditis

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Culture-negative endocarditis in children is a challenging condition seen predominantly in patients with congenital heart disease. We describe the complexity of diagnosis and management in a child with Coxiella burnetii endocarditis, which required prolonged antimicrobial therapy.

A 6-year-old boy with congenital heart disease presented with a six-week history of fever and cough. He had pulmonary atresia with VSD and aortopulmonary collateral arteries and a history of surgical corrections with VSD closure, unifocalization and a Contegra conduit in pulmonary position. Six months earlier, a Melody valve had been placed in pulmonary position. His private physician treated suspected pneumonia, but persistent fever led to referral. Initial investigations showed mildly elevated inflammation markers and a TTE showed no signs of endocarditis or Melody valve dysfunction.

While repeated blood cultures were sterile, C. burnetii serology detected Phase 1 IgG antibodies of 1:65'536. An 18F-FDG PET-CT showed metabolic activity surrounding the pulmonary conduit/Melody valve, confirming the diagnosis of C. burnetii endocarditis. We started treatment with doxycycline and hydroxychloroquine according to published recommendations. Despite increasing drug dosages to twice the recommended dose, we did not reach the target levels of 5-10mg/l for doxycycline (max. level measured 2.62mg/l) or 0.8-1.2mg/l for hydroxychloroquine (max. level measured 0.49mg/l). As the general condition quickly improved, and the patient experienced adverse effects of sunburn and nausea we did reduce drug doses to a tolerable level.

Over the 40 months of therapy a slow decline of serological markers was documented. A follow-up 18F-FDG PET-CT scan after 39 months of therapy demonstrated only minimal remaining metabolic activity at the conduit, allowing discontinuation of therapy. The Melody valve retained its function and valve replacement was not necessary. The patient was last seen 15 months after therapy end, with no clinical or serological signs of relapse.

There are only 41 case reports of paediatric C. burnetii endocarditis described in the literature and therefore the treatment guidelines are based on adult data. Our case highlights the challenges of treating C. burnetii endocarditis in children, given the lack of evidence for the optimal treatment strategy.

P 19

A rare pathogen to cause an osteoarticular infection

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Introduction: The causes of paediatric hip pain are extensive. One important differential is an infectious process, with staphylococcus aureus, group A streptococci and pneumococci being the most common pathogens. Typically, the patients have a rapid onset of fever and joint pain. In septic hip arthritis, however, the typical clinical signs swelling and redness are often absent.

Case presentation: A 12-year-old otherwise healthy male was referred by his general paediatrician with left-sided hip pain of 9 weeks' duration and weight-bearing intolerance, worsening over the past 3 days. No fever. Infection parameters were slightly elevated (ESR 36 mm/h, CrP 23 mg/dl). Radiographs were unremarkable except for the unexpected finding of a secondary bilateral hip dysplasia (neonatal hip ultrasound normal). Sonography revealed an effusion in the left hip joint and the MRI showed septic arthritis and acetabular osteomyelitis with a subperiosteal abscess. Joint lavage was performed, which revealed a serous-cloudy punctate. Microbiologic cultures of the punctate, the biopsy of the synovia and a wound swab showed growth of a fully sensitive strain of Fusobacterium nucleatum (F.N.). Blood cultures resulted negative. A calculated intravenous antibiotic therapy with Co-Amoxicillin was initiated after surgical joint lavage and continued for 8 days followed by oral administration up to a total of 6 weeks. After two weeks, the patient was symptom free, had returned to full participation in sports and his inflammation levels were low.

Discussion: In contrast to extensive abnormalities in the MRI, our patient had had a slow clinical course, which fits well to an anaerobic infection. F. n. is a gram-negative obligate anaerobic bacterium that colonizes the mucous membranes of the oral cavity and GI tract. It is best known to cause oropharyngeal infections, but seldomly causes osteoarticular infections. In literature, only a few cases have been reported. This case report impressively shows the importance of direct joint aspirate/ biopsy for diagnosis of paediatric septic arthritis, especially because blood cultures remain negative in most cases.

Conclusion: Our case underlines the importance of direct microbiologic diagnostics from operative/ puncture material in paediatric septic arthritis, as there is a broad spectrum of possible pathogens.

P 20

RSV Immunization Rate of Newborns and Hospitalization of Infants Due to RSV Infections in the Canton of Thurgau in the 2024/25 RSV-Season

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Objective: Since autumn 2024, the monoclonal antibody Nirsevimab (Beyfortus) has been available for infants in Switzerland. New recommendations of the Federal Office of Public Health (BAG) state that all infants born between October and the end of March should receive RSV immunization shortly after birth. The obstetric departments of the Spital Thurgau AG in Frauenfeld and Münsterlingen take care of all inpatient births of the Canton Thurgau resulting in over 2300 newborns per year in these two hospitals. RSV immunizations were initiated in both sites immediately after the vaccine became available in October 2024.

Method: This research involves a retrospective statistical analysis of RSV immunizations administered from October 2024 to March 2025 at Münsterlingen Hospital and Frauenfeld Hospital (Canton of Thurgau). Additionally, in infants younger than one year of age hospitalized with bronchiolitis due to RSV-infection, the immunization rate will be reported. We aim to report the total number of hospitalized infants in the Department of Pediatrics, Hospital Muensterlingen, and to compare them to previous years. Moreover, data of the length of stay and severity of illness e.g. respiratory support, will be provided.

Expected results: RSV immunization will be continued at both hospitals until March 31, 2025. Data analysis will be conducted in April 2025. The results of this study will be presented at the pediatric congress in Bern in May 2025. We hope to provide a comprehensive overview of the relevant numbers of RSV Infections in order to gain insight into epidemiology of this disease in a rural setting of a geographically widespread canton as well as to share the experience with the rapid implementation of this new prophylaxis.

P 21

A rare cause of acute respiratory distress syndrome presenting with biphasic stridor

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Introduction: Pulmonary manifestations, and in particular airway manifestations, in pediatric patients with Crohn's disease are rare. The diagnosis can be challenging.

Case presentation: An 11-year-old boy presented with a 10-day history of fever, cough, and erythema nodosum on both lower legs. Loose stools two times per day for three weeks with a weight loss of 1.5kg were additional clinical findings. Physical examination revealed the typical erythema nodosum skin lesions and a respiratory distress syndrome Laboratory examination showed Inflammatory syndrome. Intravenous antibiotics treatment with amoxicillin/clavulanic acid for initial suspicion of an invasive bacterial infection was begun without clinical response. Contrary, a deterioration was observed with progressive respiratory distress syndrome and biphasic stridor on day 4th of hospitalization Computed tomography and tracheobronchoscopy showed a picture of severe laryngotracheitis with subglottic fibrinous secretion and mucosal swelling with corresponding subglottic stenosis. In the lower part of the trachea, massive inflammatory changes with mucosal swelling and fibrinous endotracheal secretions were detected. The patient needed an invasive air way management with admission to the PICU. No pathogen could be detected in the tracheal secretion. Anti-inflammatory treatment with systemic corticosteroids was initiated with good response on the respiratory symptoms within 48 hours. Further investigations revealed high stool calprotectin level of 5041 ug/g stool. A manifestations of chronic inflammatory bowel disease, e.g. Crohn's disease (CD), was suspected. Accordingly, the subsequent gastroduodenoscopy and colonoscopy showed a picture of short-stretched Crohn's colitis and an involvement of the terminal ileum. Clinical remission of CD was obtained on induction therapy with systemic corticosteroids 1mg/kg with tapering down and normalization of calprotectin level was achieved within 3 months of subcutaneous weekly administration of Methotrexat. Airway problems did not flare up within follow up of 6 months. This patient was diagnosed with a severe laryngotracheitis as an unusual extraintestinal manifestation of Crohn's disease.

Conclusion: An acute severe laryngotracheitis may be a life threatening extraintestinal manifestation of CD. Therefore, Physicians should be alert to unusual extraintestinal manifestations of inflammatory bowel disease, especially in CD to confirm the right diagnosis timely.

P 22

Fulminant course of IgA vasculitis with pulmonary involvement

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Case report: A 4-year-old girl presented with generalized purpura on arms, legs and trunk as well as abdominal pain. Therefore clinical diagnosis of IgA vasculitis was made in an external hospital. Because of severe pain, recurrent bloody diarrhea and progressive skin lesions oral therapy with corticosteroids was implemented. Due to persisting abdominal pain patient was admitted to hospital after 1 week of disease course. Intussusception was excluded by ultrasound. Nevertheless abdominal complaints persisted during oral corticosteroid therapy and increasing signs of respiratory distress and hemoptysis were recognized. Patient therefore was transferred to our hospital for further diagnostics and observation. On admission intussusception was excluded again. The girl presented with reduced general status and increasing signs of respiratory deteriotation. Additional imaging was made and showed alveolar hemorrhage. Due to severe pulmonary hemorrhage leading to respiratory insufficiency and need for intubation, high-dose steroid therapy was started. As therapy was escalated (high dose corticosteroids and additional broad spectrum antibiotic therapy due to septical like image), the respiratory situation improved, so that patient could be extubated after 5 days. Intravenous corticosteroid therapy therefore could be stopped after 7 days and therapy was continued with oral steroids afterwards. In the following days general condition improved quickly, although recurrent microhematuria appeared as a potential sign of additional nephrological involvement. Due to fulminant course with pulmonary involvement and questionable renal involvement additional immunosuppressive therapy with mycophenolate mofetil was started. After hospital discharge, the patient was followed in our ambulance and showed further improvement. After 12 weeks oral corticosteroids could be stopped. Therapy with mycophenolate-mofetil will be continued until further notice.

Discussion: Pulmonary involvement in IgA vasculitis with alveolar hemorrhage occurs rarely but is a potentially life-threatening circumstance. The prevalence is given as 0.8-5% with a

mortality of 28%. Therapeutic basis is very thin due to its rare occurrence.

Conclusion: IgAV is mainly self-limiting and therefore often only requires symptom-oriented therapy. In rare case of organ- or life-threatening complications aggressive therapy with immunosuppressive drugs is required.

P 23

The effect of hypertonic saline inhalation on the upper airway microbiome in infants with cystic fibrosis

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Inhalation of nebulized hypertonic saline is widely used to improve mucociliary clearance in airway epithelia. While generally considered safe, its effect on early airway microbiome development remains unexplored. By building the first non-redundant gene catalog of the infant nasal microbiome with 706 longitudinal metagenomes from infants with and without cystic fibrosis (CF), we found an expansion of salt-related microbial transporter genes linked to increased salt exposure in early life. Notably, Haemophilus influenzae, Acinetobacter, Moraxella and Klebsiella spp., along with fungi such as Candida spp. and Aspergillus spp.— potential CF pathogens later in life—carry these genes with high sequence identity. Paired 16S rRNA amplicon sequencing revealed community shifts favoring Haemophilus, Staphylococcus, and Moraxella in nasal environments with higher salt concentrations. While the enrichment signature was also detected in an independent CF infant cohort with lower airway samples subjected to hypertonic saline treatment (N = 42), it was absent in age-matched healthy infants (N = 51), water controls (N = 10) and samples of a representative healthy cohort study (N = 1,218). In vitro experiments mimicking salt stress in Haemophilus influenzae showed increased biofilm formation and upregulation of salt-associated transporter systems. Thus, hypertonic saline inhalation may alter the early upper airway microbiome by selecting for salt-resistant opportunistic bacteria, potentially promoting virulent traits. In the era of novel modulator therapies, the clinical use of hypertonic saline inhalation in infants with early CF lung disease should be reconsidered because of questionable additional value.

P 24

E-learning course to teach spirometry and its quality control

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Background: Spirometry is the most commonly used method for structured assessment of lung function in pediatric pneumology. Only good quality spirometry can ensure accurate diagnosis. The patient's co-operation is therefore of great importance. This can be particularly difficult in pre-school children. Therefore, quality control of each measurement must be performed. So far, there is no cost-free and freely accessible platform for learning how to perform this quality control. Such a learning platform, making validated content freely available, could standardize knowledge about quality control and thus further improve the standard of spirometry measurements.

Aim: Providing a freely accessible and cost-free e-learning platform for learning the most important aspects of the indication, performance, evaluation and quality control of spirometry in the context of pediatric pneumology.

Methods: Creating an e-learning course on the MedSurf platform of the University of Bern, followed by an improvement process based on the results of cognitive interviews. [1] The target group consists primarily of students of human medicine, but also employees in pediatric pneumology. The process is supported by the Institute for Medical Education at the University of Bern.

Results: The e-learning course was created under https://elearning.medizin.unibe.ch/clinisurf/pediatric-spirometry-en. With the help of the results of cognitive interviews and continuous feedback from all users, the content and layout have been and will be further improved. The course is divided into three chapters: basics, clinical application and quality control. Each of these is divided into a theory and an exercise unit. Illustrations with dynamic explanations help to convey the knowledge. Practical examples allow learners to apply the knowledge and check their level of knowledge.

Outlook: The next step will be to test the learning effects of the e-learning course with medical students, junior doctors and pediatricians in training in pediatric pneumology, using questions about clinical cases. The results will be used to further improve the course before it is translated from English into German and made available to the public.

1. Hill, J., et al., Educator's blueprint: A how-to guide for collecting validity evidence in survey- based research. AEM Educ Train, 2022. 6(6): p. e10835.

P 25

Recruitment strategies for smoking parents and their children: The "Early Life Intervention in Pediatrics Supported by E-Health (ELIPSE) - Smoke" study

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Introduction: With around 24% of adults in Switzerland smoking, many children are exposed to second-hand smoke, resulting in increased health risks. Since recruiting smoking parents for clinical trials is difficult, we present our recruitment strategies and challenges we faced.

Methods: In the randomized controlled trial "Early Life Intervention in Pediatrics Supported by E-Health (ELIPSE) - Smoke", we are testing an app-based coaching intervention for German-speaking smoking parents to reduce exposure to second-hand smoke of children under six years. We developed three main recruitment strategies. First, we contacted parents in-house on the wards of the Bern University Hospital. Second, we provided primary care pediatricians and staff in our outpatient clinic with study information to encourage them to refer potential participants. Third, we advertised on social media, public transport, in-house monitors, and displayed flyers in healthcare-related institutions. We present the outcomes of these three strategies for the first nine months of recruitment (April-December 2024) in the Canton of Bern.

Results: We recruited 24 participants. First, we contacted 231 parents on the wards, 50 (22%) were eligible, of whom 12 (24%) joined the study. On the surgical ward, 23 of 91 (25%) parents were eligible, of whom five (22%) participated. On the general pediatric wards, 27 of 140 (19%) parents were eligible, of whom seven (26%) participated. Main reasons for refusal were no interest in the study and lack of time/being overwhelmed by the child's illness. Second, 22 of 28 pediatricians contacted were interested in the study and referred 17 parents. We enrolled eight (53%) of the 15 (88%) eligible parents, main reason for non-participation was declining after initial interest. The outpatient clinic referred eight parents, of the three (38 %) eligible parents, one (33%) was included, others declined or we lost contact after an initial contact. We could never reach two (25%) parents referred by the outpatient clinic. Third, seven people contacted us via our website or email. Of the three (43%) eligible, one person (33%) was included. Non-inclusion was due to declining and loss of contact. Two participants were recruited by personal contact.

Conclusion: Strategies yielding the highest number of enrolled participants were recruitment on hospital wards due to many contacted people, and referrals from primary care pediatricians due to high eligibility and enrollment rates.

P 26

Effect of vaping on mucociliary activity in nasal epithelial cell cultures of adolescents

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E-cigarette use, also known as vaping, is increasingly popular amongst adolescents, but we know little about the health effects in this population, especially about potential impacts on the mucociliary activity. We thus investigated associations between vaping and ciliary beating frequency (CBF) and mucociliary transport (MCT) in cultured nasal epithelial cells of vaping (at least weekly), vaping and smoking (both at least weekly, "mixed users") and neither vaping nor smoking control adolescents (no or less than monthly vaping or smoking). CBF and MCT was assessed 24h after infection of the cells with rhinovirus (RV-16) (CBF additionally at baseline) using an inverted transmission light microscope and the Cilialyzer software. We applied multivariable adjusted models accounting for sex, age, education level, exposure to second-hand smoke and ambient air pollution, and atopic disease.

Sixty adolescents agreed on a nasal brushing (mean age of 18 years, range of 15-22 years, 51% female) and the cells of 47 participants grew successfully at the air-liquid interface resulting in a respiratory epithelium with beating cilia and mucus-producing cells. The rate of successful cell cultivation was higher in controls (89%, N = 27, 17 (range 16-18) years) compared to vaping participants (72%, N = 25, 19 (15-22) years) and mixed users (67%, N = 6; 19 (16-22) years). At baseline, CBF did not differ between cells of vaping participants, mixed users and controls (median 12.2 Hz [95% confidence interval of 10.8 to 12.7 Hz], 12.4 Hz [9.5 to 17.3], 11.3 Hz [10.1 to 12.5 Hz], respectively). RV infection increased CBF in vaping (14.4 Hz [12.1 to 16.7]) and mixed users (13.6 Hz [9.7 to 23.5]), but not in control cell cultures (versus 11.0 Hz [9.7 to 12.0]). MCT speed of nasal epithelial cells in vaping participants was lower than in non-vaping controls (median speed of 5.7µm/s [95% CI of 0 to 15.0µm/s] versus 18.6µm/s [9.9 to 27.3µm/s], maximal speed of 15.7µm/s [0 to 34.8µm/s] versus 32.5µm/s [14.6 to 50.4µm/s]. Multivariable adjusted log-transformed median MCT speed was similar in vaping participants and controls (coefficient -0.4, 95% CI -2.3 to 1.5, p = 0.60), as was multivariable adjusted log-transformed maximal MCT speed (coefficient -0.1, 95% CI -2.3 to 2.1). We found no effect of RV infection on MCT.

In conclusion, vaping could affect mucociliary transport speed and CBF after viral infection. However, these results need to be further investigated for potential confounding factors.

P 27

Characteristics of pediatric parapneumonic effusion (PPE) caused by Streptococcus pneumoniae versus Streptococcus pyogenes

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Background: Streptococcus pneumoniae (Spn) is the most common agent responsible for pediatric parapneumonic effusion (PPE) complicating bacterial pneumonia. However, as a result of the 2022-2024 outbreak of invasive group A streptococcal infections, PPE caused by S. pyogenes (Spy) became more prevalent, allowing for sufficient case counts to compare the clinical characteristics of PPE associated with these two pathogens.

Methods: Retrospective single-center study from 2004 to 2024 of bacteriologically proven cases of PPE caused by Spn or Spy requiring pleural drainage. Study inclusion required pathogen identification by (1) culture of blood or pleural fluid, or (2) broad-spectrum PCR from pleural fluid. Thirty-four clinical variables were extracted from medical records. Radiologic evidence of acute lung structural damage was defined as the presence of ≥1 of the following three findings reported during the hospital stay: pneumatocele; seropneumothorax; bronchopleural fistula.

Results: The study included 173 patients with PPE with pleural drainage (female sex 45%; median age 4.4 years, interquartile range [IQR] 2.4-6.9). Spn was found in 68 (39%) and Spy in 31 (18%) cases, respectively. Septic and/or toxic shock on admission occurred in 4 (6%) Spn cases and 14 (45%) Spy cases (p <0.0001). While admission rates to the intensive care unit were similar (Spn 54%; Spy 66%), inotropic support was administered to 7 (10%) Spn cases and 12 (39%) Spy cases (p 0.002). The median duration of chest tube placement was 6 days [4-11] (Spn) vs. 4 days [2-6] (Spy) (p 0.004). Video-assisted thoracoscopic surgery was performed in 27 (38%) Spn cases and 10 (32%) Spy cases. However, thoracotomy was performed in 9 Spn cases (13%) as opposed to 0 Spy cases (p 0.054). Radiologic evidence for acute lung structural damage was found in 46 (71%) Spn cases and 6 (20%) Spy cases (p <0.0001). The median [IQR] length of the hospitalization stay was 17 days [14-25] (Spn) vs. 14 days [11-21] (Spy) (p 0.02).

Conclusion: We observed considerable clinical differences in patients with PPE caused by Spn vs. Spy. Spn was associated with substantially greater lung structural damage, while systemic illness with hemodynamic instability more commonly accompanied Spy. Although the inherent limitations of retrospective long-term studies apply, these findings suggest that studies addressing a differential management approach based on the bacterial etiology of PPE are warranted.

P 28

Case report: Mepolizumab-dependent hypereosinophilic syndrome

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Introduction: Hypereosinophilic syndrome is a rare disease, which can lead to severe morbidity. Mepolizumab is an anti-IL-5 therapy, which seems to be effective in patients with hypereosinophilic syndrome; however, it has not yet been approved for use in pediatric patients in Switzerland.

Methods: We report a case of a 13-year-old patient with hypereosinophilic syndrome with severe asthma, pericardial effusion due to eosinophilic peri- and myocarditis, and cardiorespiratory resuscitation due to anaphylactic shock. We treated the patient with mepolizumab for one year in intervals of consecutively 4-, 6-, and 8-weeks.

Results: After two months of treatment the pericardial effusion completely resolved and lung function improved from FEV1 42% to 110%, the total IgE levels decreased from 1424 to 58 kU/l, the oral corticosteroid treatment could be stopped while blood eosinophils levels remained low. Physical activity level continuously improved and was back to the initial level after 11 months.

Conclusion: We can show that Mepolizumab is effective in this patient with hypereosinophilic syndrom. Although improvement is variable and might differ between patients, Mepolizumab treatment should be considered for patients with hypereosinophilic syndrome.

P 29

Maternal mediterranean diet was associated with less atopic dermatitis development in offspring

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Background/objectives: Early-life nutrition has been linked to the development of atopic dermatitis later in life. Little is known about the effect of maternal diet on atopic dermatitis (AD) development in the offspring. Moreover, early onset of AD is frequently associated with the progression to other allergic diseases. Therefore, we evaluated associations of maternal nutrients and dietary patterns with AD development in the offspring up to two years of age.

Methods: A total of 116 mother–child dyads were prospectively recruited in the context of the Childhood, Allergy, Nutrition, and Environment (CARE) study. Information on maternal dietary intake was assessed with a validated self-administered 97-item food frequency questionnaire. AD was evaluated with questionnaires and clinical visits at the age of 4 months, 1 year and 2 years. The associations between the dietary patterns and AD were examined by logistic regression analysis.

Results: 27 children (23.3%) developed AD up to 2 years. Risk factors for development of AD up to 2 years were a family history of atopy in both parents (healthy 19.1%; AD 40.7%), maternal antibiotic therapy during pregnancy (healthy 4.5%; AD 29.6%), and maternal irregular vitamin supplementation (odds

ratio (OR) and 95% confidence interval (CI) 4.28; 1.18, 20.68), while adherence to the maternal mediterranean diet pattern showed a protective effect on AD (OR and 95% CI 0.23; 0.07, 0.72).

Conclusions: Maternal diet has the potential to influence AD development in the offspring.

P 30

Programmed Cell Death 1 Receptor Axis Blockade Enhances Antigen-Specific T Cell Proliferation in Pediatric Cystic Fibrosis

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Introduction: Cystic Fibrosis (CF) is associated with recurrent pulmonary infections leading to progressive lung tissue destruction and high morbidity. Chronic infections can produce CD4+ T-lymphocyte (CD4 T cell) exhaustion, characterized by overexpression of the Programmed Cell Death 1 Receptor (PD-1). Rare studies have assessed the role of the PD-1/PD-L1 (ligand) pathway in CF-associated immune dysregulation. In this study, we aimed to investigate CD4 T cell exhaustion in a local cohort of pediatric CF patients and to evaluate CD4 T cell rescue after blockade of the PD-1/PD-L1 axis.

Methods: Cross-sectional monocentric study including pediatric CF patients and controls. PD-1 expression was measured on total and memory CD4 T cells by mass cytometry. Antigen-specific CD4 T cells proliferation capacity in response to CF-relevant antigens stimulation (Staphylococcus aureus (Sa), or Pseudomonas aeruginosa (Pa)), was assessed using a CFSE-based flow cytometry assay in absence/presence of anti-PD-L1 blockade antibody.

Results: We included 25 pediatric CF patients and 7 controls (56% and 86% females, median age 11 and 9 years, respectively). No significant differences were observed between the two groups in terms of 1) PD-1 expression on total and memory CD4 T cells and 2) antigen-specific CD4 T cells proliferation capacity. However, PD-1/PD-L1 blockade significantly increased bacteria-specific CD4 T cell proliferation only in CF patients for Pa ($n = 20$, $p < 0.001$) and Sa ($n = 12$, $p = 0.02$). No correlation was found between PD-1 expression on total or memory CD4 T cells, nor with antigen-specific T cell proliferative capacity, with markers of disease severity (clinical, microbiological or radiological parameters) in CF patients.

Conclusion: To our knowledge, this is the first study in pediatric CF patients showing enhanced Sa- and Pa-specific CD4 T cells proliferation after PD-1/PD-L1 blockade, despite stable PD-1 levels on CD4 T cells. Further understanding of the precise mechanism is needed, as PD-1/PD-L1 immunotherapy may represent a future therapeutic option for selected CF patients.

P 31

The effect of antibiotics on the intestinal microbiota in children - a systematic review

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Background: Children are the age group with the highest exposure to antibiotics (ABX). ABX treatment changes the composition of the intestinal microbiota. The first few years of life are crucial for the establishment of a healthy microbiota and consequently, disturbance of the microbiota during this critical period may have far-reaching consequences. In this review, we summarise studies that have investigated the effect of ABX on the composition of the intestinal microbiota in children.

Methods: According to the PRISMA guidelines, a systematic search was done using MEDLINE and Embase to identify original studies that have investigated the effect of systemic ABX on the composition of the intestinal microbiota in children.

Results: We identified 89 studies investigating a total of 9,712 children (including 4,574 controls) and 14,845 samples. All ABX investigated resulted in a reduction in alpha diversity, either when comparing samples before and after ABX or children with ABX and controls. Following treatment with penicillins, the decrease in alpha diversity persisted for up to 6-12 months and with macrolides, up to the latest follow-up at 12-24 months. After ABX in the neonatal period, a decrease in alpha diversity was still found at 36 months. Treatment with penicillins, penicillins plus gentamicin, cephalosporins, carbapenems, macrolides, and aminoglycosides, but not trimethoprim/sulfamethoxazole, was associated with decreased abundances of beneficial bacteria including Actinobacteria, Bifidobacteriales, Bifidobacteriaceae, and/or Bifidobacterium, and Lactobacillus. The direction of change in the abundance of Enterobacteriaceae varied with ABX classes, but an increase in Enterobacteriaceae other than Escherichia coli was frequently observed.

Conclusion: ABX have profound effects on the intestinal microbiota of children, with notable differences between ABX classes. Macrolides have the most substantial impact while trimethoprim/sulfamethoxazole has the least pronounced effect.

P 32

Communication and education strategies for young athletes and their caregivers to prevent sports-related traumatic brain injury: a systematic review

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Introduction: Mild Traumatic Brain Injury (mTBI), commonly referred to as concussion, is one of the most frequent injuries in children and adolescents. Different strategies have been proposed to prevent concussions in contact sports. These include technical, policy, economic, and communication and education

strategies, which have been summarized in past systematic reviews, though either not focusing on sports or not considering children, adolescents, and their parents as targets. To close the gap, we conducted a pre-registered systematic review of past communication and education interventions targeting children and adolescents who practice contact sports and their caregivers to collect evidence on best practices to enhance awareness, knowledge, and prevention of sports-related concussions.

Methods: Following the PICOS search tool and PRISMA guidelines, in December 2023, we conducted a search in six academic databases (e.g., PubMed) and previous reviews on mTBI prevention strategies. After a stepwise screening process, we collected information on intervention and sample characteristics, study design and main outcomes for all included studies. We assessed the study quality using the CONSORT checklist.

Results: Our initial search resulted in 2974 articles. of which 22 were eventually included in the review. Twenty-one studies were conducted in North America, encompassing both online and in-person interventions. Ten were grounded in CDC's Heads-UP program. We found support for positive short-term effects on knowledge and less consistent effects on attitudes related to concussion. Positive long-term effects on both knowledge and attitudes were not always sustained. The overall study quality ranged between 10 and 25 out of maximum 34 points.

Conclusion: Public health experts need to take into consideration that the promising findings of previous communication and education strategies to prevent sport-related concussions are culturally bound, limited to knowledge change and short-term effects. Future strategies need to be targeted to the specific populations and go beyond mere information dissemination by focusing on contents and formats that promote positive attitudes, intentions, and behaviors.

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Association between Bisphenol A and Autism Spectrum Disorders (ASD): a systematic review and meta-analysis

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Endocrine-disrupting chemicals (EDCs), such as Bisphenol A (BPA), can interfere with hormonal systems, particularly during critical periods of development. BPA, widely used in the production of synthetic polymers and thermal paper[i], has been investigated for its potential link to neurodevelopmental disorders, including autism spectrum disorders (ASD). ASD is a complex condition characterized by deficits in social communication and repetitive behaviours. Despite growing interest, no definitive link between prenatal BPA exposure and ASD has been established, prompting the need for this systematic review.

Our study aims to evaluate whether prenatal BPA exposure is associated with the incidence of ASD. A comprehensive search was conducted in PubMed, Embase, APA PsycInfo, and Web of Science. Our inclusion criteria focused on human studies published between 1994 and 2023 that investigated the association between BPA exposure, mixtures of EDCs containing BPA, and ASD, as defined by DSM-IV and DSM-V criteria. Only studies with clear methodologies and statistical analyses were included, while reviews and animal studies were excluded. From 382 articles, 91 were duplicated and 272 were excluded during screening based on title and abstracts. Screening was performed in blind by three reviewers using Rayyan. The remaining 19 articles were evaluated for risk of bias using JBI guidelines for cohort and case studies. Discrepancies in the results were resolved through discussion.

Our preliminary meta-analysis shows that the risk ratio (RR) of the association between exposure to BPA and ASD ranges between 0 and 3.59 across cohort studies, displaying an important heterogeneity (I2 = 0.56). This heterogeneity is due to important methodological differences between the studies, such as differences in diagnosis criteria, statistical method, population evaluated and analytical BPA measurement.

Overall, we are synthesizing existing evidence on the potential link between prenatal BPA exposure and ASD, that will contribute to improving the understanding of the neurodevelopmental effects of EDCs and highlighting gaps for future research.

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Comparative efficacy of sepiapterin (SEP) vs. sapropterin (SAP) in phenylketonuria (PKU): a systematic literature review (SLR) and multi-level network meta-regression (ML-NMR)

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Objectives: Phenylketonuria (PKU) is caused by phenylalanine (Phe) hydroxylase deficiency, resulting in elevated Phe levels and ensuing neurological impairment. Despite management of Phe through diet, pharmacologic interventions are needed in PKU. This study aimed to compare the efficacy and safety of sepiapterin (SEP) vs. sapropterin (SAP) among PKU patients by systematic literature review (SLR) and data from placebo-controlled SEP trial (MD-003).

Methods: Aggregate data from this SLR, along with MD-003 data, were synthesized for change from baseline (CFB) Phe, percent (%) change in Phe, and adverse events (AEs). Bayesian network meta-analyses (NMA) using 2 approaches (pooling dosages for each treatment and separating different doses); ML-NMR with pooled dosages for SEP/SAP, adjusting for baseline Phe, age, weight; fixed-effect (FE); and random-effects (RE) models using medians for efficacy outcomes and odds ratios (OR) for AEs were run; 95% credible intervals (CrI) were computed.

Results: The SLR identified 9 randomized controlled trials; 5 were eligible for inclusion. With N = 98 MD-003 participants, the evidence base comprised 429 patients. Results from the FE NMA showed that SEP had statistically significant improvements over that of SAP in both CFB Phe (pooled: -222 [95% CrI: -269, -170]; SAP 10 mg/kg: -111 [-221, -5]; SAP 20 mg/kg: -181 [-258, -100]) and % change in Phe (pooled: -23 [95% CrI: -34, -13]; SAP 10 mg/kg: -31 [-45, -16]; SAP 20 mg/kg: -18 [-32, -5]). Findings from the ML-NMR FE model were comparable. Odds of any AE were comparable for SEP and SAP.

Conclusions: This study shows statistically significant Phe reduction with SEP over that of SAP among patients with PKU, with a similar safety profile. These findings align with results from the head-to-head trial PKU-002. Strengths included the use of multiple models, including ML-NMR, to validate findings from the NMA; results were consistent across models, which supported the robustness of these findings.

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Transient secondary pseudo-hypoaldosteronism in infants with urinary tract infections: systematic literature review

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Infants with a congenital anomaly of the kidney and urinary tract sometimes present with hyponatremia, hyperkalemia, and metabolic acidosis due to under-responsiveness to aldosterone, hereafter referred to as secondary pseudo-hypoaldosteronism. The purpose of this report is to investigate pseudo-hypoaldosteronism in infant urinary tract infection. A systematic review was conducted following PRISMA guidelines after PROSPERO (CRD42022364210) registration. The National Library of Medicine, Excerpta Medica, Web of Science, and Google Scholar without limitations were used. Inclusion criteria involved pediatric cases with documented overt pseudo-hypoaldosteronism linked to urinary tract infection. Data extraction included demographics, clinical features, laboratory parameters, management, and course. Fifty-seven reports were selected, detailing 124 cases: 95 boys and 29 girls, 10 months or less of age (80% of cases were 4 months or less of age). The cases exhibited hyponatremia, hyperkalemia, acidosis, and activated renin-angiotensin II-aldosterone system. An impaired kidney function was found in approximately every third case. Management included antibiotics, fluids, and, occasionally, emergency treatment of hyperkalemia, hyponatremia, or acidosis. The recovery time averaged 1 week for electrolyte, acid-base imbalance, and kidney function. Notably, anomalies of the kidney and urinary tract were identified in 105 (85%) cases.

Conclusions: This review expands the understanding of overt transient pseudo-hypoaldosteronism complicating urinary tract infection. Management involves antimicrobials, fluid replacement, and consideration of electrolyte imbalances. Raising awareness of this condition within pediatric hospitalists is desirable.

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Early Life Intervention in Paediatrics Supported by E-Health (ELIPSE I): Study Design and Preliminary Participant Characteristics of a Randomised Controlled Trial Targeting Childhood Obesity

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Background: Childhood obesity has dramatically risen in recent decades and is a major public health concern. In Switzerland, one of five school-aged children is overweight or obese. Like other non-communicable diseases, obesity often has its origin in early childhood driven by behavioural factors at home related to nutrition and physical activity. The aim of ELIPSE I is to develop, implement and evaluate a personalised, family-centred app-based intervention to coach parents to reduce their children's severity of obesity.

Methods: ELIPSE I is a randomised-controlled trial with two arms. We aim to recruit 148 dyads of a parent and a child, age 6-12 years with an body mass index (BMI) above the 97th percentile (according to Swiss national growth charts), that are randomly assigned to receive treatment as usual (TAU) at a tertiary weight-management clinic (control group) or a guided digital intervention in addition to TAU (intervention group) for a period of 20 weeks. Primary and secondary endpoints comprise the ratio of total energy intake and expenditure, severity of obesity and relevant cardiometabolic risk factors (e.g. blood pressure, lipid profiles). The ELIPSE app assesses nutritional and exercise habits and provides thematically tailored input and personalised feedback from a coach.

Results: From February 2024 to January 2025, we recruited 31 of 59 (52.5%) eligible parent-child dyads (total screened 131). Included children, 54.8% (n = 17) females, had a mean age of 10.1 (SD 2.1) years and a mean BMI of 27.1 (SD 3.9) kg/m2, corresponding to a mean z-score of 3.1 (SD 0.9). Screening for comorbidities revealed that 39.0% (n = 7/18) were non-dippers in the 24-hour ambulatory blood pressure monitoring and 66.7% (n = 12/18) showed hyperechogenicity on liver ultrasound, indicative of incipient hepatic steatosis. Self-reported screentime of children was 15.5 (SD 9.7) hours per week. A positive family history was most frequently reported for obesity (80%), arterial hypertension (66.7%) and dyslipidaemia (53.3%).

Conclusion: Based on the recruitment progress so far, we conclude that the ELIPSE app is perceived as welcome additional offering and parents are pleased to receive further support at home to help their children with weight reduction. Participating children clearly belong to a vulnerable group with high-risk for obesity-related morbidity and mortality later in life if left untreated. Outcome-related results of the trial are expected in the course of 2028.

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Participation of children and adolescents in outpatient health care settings: an invitation to a multidisciplinary nationwide study.

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The right to participation of children in healthcare is defined and anchored by a legal framework on various levels. To current, practice doesn't fully comply with these requirements in Switzerland. Barriers include legal, policy-based, and healthcare aspects.

Our study aims to describe and facilitate participation of children and adolescents in healthcare, looking at different ages, diagnoses and settings. We will apply a mixed method approach, including qualitative focus groups with 6-12 participants (young people, healthcare professionals and parents) to identify needs and barriers as well as field observations, and interviews with children and medical professionals to focus on children's perspectives on consultations. This will ensure a diverse representation across language regions, specialties, organizations, and age ranges. Further, an online qualitative and quantitative survey will be distributed to a wide range of healthcare professionals, administrators, and policy makers, which will cover various aspects of participation of children and adolescents, including legal awareness, barriers, motivations, challenges, collaboration, satisfaction, and improvement suggestions.

By adapting an interdisciplinary research approach, we will be able to combine an individual and an institutional perspective within Switzerland, as well as a cross-national legal perspective. With our approach, we also aim to ensure that perspectives of children and adolescents, their parents, and health care professionals are adequately and sensitively addressed.

Our aim is to promote a highly transdisciplinary model of participation, a solution that will reshape and modernize children's rights of participation in general, especially from the perspective of children's health services. The legal and practical recommendations and adaptations that we are aiming at will be developed through the identification of barriers and burdens that hinder and limit the participation of minors. For this reason, the role and perspectives of children and young people is of fundamental importance to our project.

The presentation is meant to inform about our project and to invite possible participants from hospitals and private practices. We want to integrate an active discourse and exchange of knowledge with all interested experts attending the congress. By reaching out to attendees, we look forward to receiving input based on their experiences and opinions to encourage their potential participation.

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Longitudinal risk and resilience factors in pediatric postoperative pain: Preliminary results from a prospective longitudinal study

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Introduction: Chronic postsurgical pain (CPSP) is defined as pain that develops or persists after a surgical procedure and has a significant impact on quality of life. Recent meta-analyses estimate the prevalence to be as high as 25% following orthopedic surgery. Previous studies point to the importance of psychological factors in the development of CPSP, yet the current understanding is limited, and hence prevention and treatment remain challenging. Therefore, the aim of this study is to prospectively observe a range of pain- and emotion-related risk and resilience factors in children and adolescents undergoing major orthopedic surgery.

Methods: This is an ongoing observational, multi-centric study that includes children and adolescents aged 8-18 years with a planned orthopedic surgery at one of the study centers (i.e., University Children's Hospital Zurich, University Children's Hospital Basel, and Balgrist University Hospital), and their parents. Participants complete five assessment time points: before surgery (baseline), 2 weeks after surgery (post), and 3 (follow-up [FU]1), 6 (FU2), and 12 (FU3) months after surgery. Children and parents complete questionnaires at each time point, including on emotional state, pain, symptoms of anxiety and depression, sleep, quality of life, and social support. Preliminary mean values of relevant psychological and pain-related variables will be compared before vs after surgery.

Results: The first 12 participants who completed the baseline and post measurements* showed differences with regard to functional disability, with significantly higher values after surgery, and regarding quality of life, with significantly lower values after surgery. Depression and anxiety did not change significantly.

Discussion: These preliminary results give first insights into the trajectories of emotional state and pain-related factors before and after surgery, pointing to potential targets for early intervention to prevent pain chronification after surgery. This is crucial, given the many negative consequences of pediatric chronic pain on school, sport, sleep, and social life of children, adolescents, and families. To the best of our knowledge, this study collects the first Swiss data on postsurgical pain in children and adolescents.

*at the time of abstract submission

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Interim Results From the APHENITY Extension Study: Sepiapterin Reduces Blood Phe With Improved Dietary Phe Tolerance in Participants With Phenylketonuria

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Introduction: Phenylketonuria (PKU) is an autosomal-recessive inborn error of metabolism caused by deficiency of the enzyme phenylalanine hydroxylase (PAH). Sepiapterin is being developed as a novel oral treatment for PKU. The Phase 3 APHENITY trial (NCT05099640) was a global, two-part, registration-directed study evaluating sepiapterin in a broad PKU population. Upon completion, participants could enroll directly into the APHENITY open-label extension study (NCT05166161). Here, we describe updated results from the extension study (30 June 2024 data cut), including those from participants on the extension study who were not involved in the APHENITY trial.

Results: As of the June 30, 2024 data cut, 169 participants (median age, 14.0 years [min, max: 0.2, 55.0]) were treated with sepiapterin. Dietary Phe Tolerance Assessments were performed in 100 participants; 27 of whom were non-feeder participants. The median (min, max) treatment exposure was 465.0 (26, 868) days for the participants who undertook Dietary Phe Tolerance Assessments: 497.0 (26, 868) days for feeder participants and 147.5 (26, 273) for non-feeder participants. For all participants undertaking the Dietary Phe Tolerance Assessment, the least-square mean change (95% confidence interval) for change in dietary Phe consumption from baseline to Week 26 was 38.0 mg/kg/day (32.1, 43.9) (protein: 0.76 g/kg/day [0.64, 0.88]). An approximately 2.3-fold increase from baseline (27.6 mg/kg/day [protein: 0.55 g/kg/day]) in mean daily Phe consumption was achieved at Week 26 (63.6 mg/kg/day [protein: 1.27 g/kg/day]) in the overall Dietary Phe Tolerance Assessments. Mean blood Phe remained within the recommended target of <360 µmol/L during the Dietary Phe Tolerance Assessment commensurate with increase in Phe consumption. Overall, sepiapterin showed a favorable safety profile and was well tolerated in the study. In the overall group (n = 169), treatment-related TEAEs reported in ≥2% of participants were diarrhea (13 participants, 7.7%), headache (11 participants, 6.5%), discolored feces (7 participants, 4.1%), vomiting (4 participants, 2.4 %), and fatigue (4 participants, 2.4%). There was a low rate of discontinuation due to TEAES of 1.8% (3 TEAEs in 2 patients); 1 patient discontinued due to increased bleeding tendency, and one patient discontinued due to nausea and constipation. All 3 TEAEs were resolved. There were no treatment-related serious TEAEs and no deaths during the study.

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Findings supporting neonatal screening for sickle cell disease: an observational study in Senegal

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Importance: Sickle cell disease (SCD) is a major public health challenge in Senegal. Neonatal screening offers a way to enhance early diagnosis and disease management, potentially reducing morbidity and mortality in affected children.

Objective: To assess the epidemiological situation, clinical evolution and outcomes of children diagnosed with SCD through neonatal screening (A) and those diagnosed during symptomatic episodes (B) in Senegal. To evaluate the feasibility and impact of systematic neonatal screening in Senegal.

Design: A retrospective cross-sectional study comparing two cohorts: children diagnosed through neonatal screening and those diagnosed during symptomatic episodes. Data spanned March 2012 to November 2020, according to cohorts, covering the five first years of the children.

Setting: The study was conducted in St. Louis, Senegal, involving the Center for Research and Ambulatory Management of Sickle Cell Disease and the Regional Hospital Center of St-Louis.

Participants: Cohort A included 44 children diagnosed through neonatal screening, while cohort B included 39 children diagnosed based on clinical symptoms.

Interventions/exposures: Neonatal screening involved blood sample analysis using techniques like isoelectric focusing, high-performance liquid chromatography or electrophoresis. Children diagnosed at birth were regularly monitored according to their age. Children diagnosed clinically had more irregular follow-up.

Main outcomes and measures: Primary outcomes included age at diagnosis, clinical complications such as vaso-occlusive crises, anemia, and hospitalization rates. Secondary outcomes included parental status, consanguinity rates and prescriptions rates.

Results: The mean age of diagnosis was 48.1 days for cohort A, and 21.9 months for cohort B. Complications were significantly more frequent in cohort B (vaso-occlusive crisis, acute anemia, transfusions and hospitalisations[LP1]). No deaths were reported in either cohort before age 5 years.

Conclusions and relevance: Neonatal screening for SCD in Senegal is feasible and could lead to earlier diagnosis and reduced morbidity compared to symptomatic diagnosis. Systematic screening could significantly reduce the disease burden, improve health outcomes, and enhance early intervention opportunities.

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Progressive Scoliosis And Areflexia – What Now?

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Background: Friedreich Ataxia (FA) is a rare progressive neurodegenerative disorder. It is the most common hereditary ataxia, with an autosomal recessive inheritance. FA is caused by loss-of-function mutations in the frataxin (FXN) gene, showing a GAA triplet expansion leading to a degeneration of the spinocerebellar pathway and the cerebellum. The clinical onset is between 8 to 14 years. The patients present with progressive ataxia of the limbs and gait, dysarthria, distal sensory loss, areflexia, oculomotor and auditory manifestations, progressive scoliosis and hypertrophic cardiomyopathy. There is no curative therapy for FA. Certain studies showed neurological improvement with disease modifying drugs as Omaveloxolone and Vatiquinone

Case report: A 13-year-old adolescent was referred by an orthopaedist to our department of pediatric neurology because of an unsteady gait pattern. Due to progressive scoliosis since the age of 9 he was under orthopaedic and physiotherapeutic care and had treatment with a corset. The patient himself described an increased unsteady gait, fatigue and a “burning” sensation in his chest during prolonged physical exercise. The examination showed an ataxia predominantly of the extremities, dysmetria, areflexia and an impaired depth sensitivity. An MRI of the neuroaxis and metabolic testing for ataxia revealed normal findings. Genetic testing showed GAA triplet expansions in both alleles of the FXN gene, securing the FA diagnosis. The cardiologic examination revealed a hypertrophic cardiomyopathy currently without restriction of cardiac function. Audiological and ophthalmological exams were normal. Despite intensified physiotherapy and the corset, the scoliosis showed further progression and surgical correction was performed. As it is not yet authorized by Swissmedic, Omaveloxolone cannot be administered at this point.

Conclusion: Friedreich Ataxia is a hereditary ataxia, mostly manifesting in childhood or adolescence and severely impairing quality of life. Progressive neurodegeneration leads to loss of various elementary functions of daily living. There exists a disease modifying therapy, which is not yet approved in Switzerland. Interdisciplinary care is necessary, including cardiological check ups, as cardiomyopathy is progressive. Further research is needed for drug therapy and its use in childhood - aiming to positively influence the course of FA.

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Type of gait pattern in children with cerebral palsy in relation to timing of lesion during brain development: A retrospective observational study

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Background: Cerebral palsy (CP) refers to a group of neurodevelopmental disorders caused by non-progressive brain injury, primarily affecting motor control. It is categorized into spastic, dyskinetic, and atactic types and encompasses a range of functional limitations. Gait abnormalities are common in children with CP, but no clear link has been found between gait patterns and the affected brain area. Recent data suggest lesion timing may be more critical than location. Abnormal prenatal ultrasound findings can reveal potential points of harm to the developing brain. Complications during pregnancy or birth may disrupt brain development, leading to neuromuscular deficits and gait abnormalities later in life.

Question: Can the timing of brain injury predict the severity and type of gait abnormalities in children with cerebral palsy?

Objective: This study explores correlations between perinatal data and gait analysis in CP patients, focusing on the impact of lesion timing on gait pattern. We categorize complications into three groups: 1) prenatal <32 weeks of gestation, 2) intranatal, and 3) postnatal. The goal is to identify gait pattern differences and predict severity of abnormalities in CP individuals.

Methods: We included children diagnosed with CP who are registered in the Swiss Cerebral Palsy Registry. Perinatal data was collected from archive reports. For medical reasons, patients had previously undergone 3D gait analysis, measuring parameters like joint angles, forces, and mobility across multiple gait cycles. The earliest gait analysis was used to capture the most representative state. Patients with significant orthopedic surgery before analysis were excluded.

Results: Preliminary findings suggest that perinatal diagnostics and postnatal gait analysis offer crucial insights into neonatal brain lesions. Data assessment is ongoing as the study is still in early stages. To date, 67 patients have been recruited, with detailed medical histories, MRI scans, and gait analyses collected. We are currently categorizing findings and determining injury timing using the dataset. Other factors such as lesion location, age at MRI, and individual growth and maturation also influence gait patterns, contributing to variability of gait.

Conclusions: It remains to be seen whether modern gait analysis can provide insights into the pathophysiology of cerebral palsy, particularly regarding the timing of brain damage. We expect to share more observations at the congress.

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Diagnosis and Management of Non-Convulsive Status Epilepticus (NCSE) in Children

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EEG evaluations are crucial in the medical management of epileptic patients, particularly in complex cases like Patient 1 and Patient 2. This abstract summarizes their clinical courses, EEG findings, and therapeutic interventions, emphasizing diagnostic challenges.

Patient 1, born on July 13, 2011, has severe symptomatic epilepsy and neurodevelopmental issues. Her EEG consistently shows intermittent high-amplitude rhythmic delta activity without clear epileptic potentials. Despite treatment with antiepileptics like Keppra and Urbanyl, she experienced recurrent status epilepticus, requiring emergency care and hospitalizations. During these admissions, she presented in a postictal state with tonic seizures. EEG changes persisted, although seizure activity was halted after propofol administration in 2024. However, clusters of tonic seizures continued, necessitating therapy adjustments. Her care requires an interdisciplinary team and regular follow-ups.

Patient 1’s EEG in September 2024 revealed rhythmic slow delta activity with spikes, indicating subclinical status epilepticus. This persisted despite treatment with Keppra, Rivotril, and Buc-

colam, but propofol administration significantly reduced epileptic activity, confirmed by improved EEG and clinical observations.

Patient 2, born on July 18, 2017, experienced severe pARDS after aspiration and developed epilepsy at four months. Her seizures were often triggered by infections and fever. Despite valproate, Rivotril, and Clobazam, she required ICU admissions for ventilation. Upon admission to Eastern Switzerland Children's Hospital, she had multiple infections and complications, requiring complex therapies. Continuous adjustments in sedation and antiepileptic medications were necessary. EEGs in October 2024 revealed theta-delta activity without clear epileptic potentials under sedation, requiring medication adjustments and careful sedation management.

In conclusion, complex epilepsy cases like those of Patient 1 and Patient 2 illustrate the challenges in diagnosing and managing recurrent status epilepticus. EEG evaluations are essential for detecting non-convulsive status epilepticus and guiding treatment. An interdisciplinary approach ensures comprehensive care and improves patient outcomes.

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Point-of-care EEG for seizure detection: comparison of electrode placement in two-channel EEG

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Background: The use of few-electrode electroencephalography (EEG) for seizure detection is an important point-of-care bedside tool and is being increasingly utilized in the pediatric emergency department for diagnosis of CNS disorders, including non-convulsive status epilepticus (NCSE) and seizure activity in children with impaired state of consciousness. The aim of this study was to compare the effectiveness of frontotemporal (FT) electrode placement (F7-T5 and F8-T6) with the usual centroparietal (CP) electrode placement (C3-P3 and C4-P4) in seizure detection, in a point-of-care two-channel EEG (tcEEG) setting.

Method: A sample of 38 prerecorded EEGs (gold standard, international 10-20 EEG system), with prior detected electrographic epileptic seizures in children (38 participants, median age 10.11 years), was reviewed. Samples were drawn from the database of the pediatric neurology department at the University Hospital in Bern, Switzerland. The EEG parameters were retrospectively converted into two tcEEGs (F7-T5 and F8-T6 versus C3-P3 and C4-P4) and were independently analyzed by two epileptologists, blinded to the gold standard EEG findings.

Results: No significant difference in seizure detection in FT electrode placement compared to CP placement (sensitivity 69.9% vs. 72.9%) was shown. In our secondary results, we found significantly higher false positive rates with FT electrode placement in samples with high interictal spike frequency. CP electrode placement showed significantly lower false negative rates for seizures with bilateral onset and seizures without side difference during propagation. No significant correlation of FT and CP electrode placement was found with other EEG parameters (age, seizure duration, presence of rhythmic spike wave activity, wakefulness during seizure).

Conclusion: FT and CP tcEEG electrode placements do not differ in seizure detection and can be used with similar efficacy (sensitivity and specificity) for point-of-care EEG bedside testing in pediatric emergency settings. The CP electrode placement shows only minor advantages compared to FT electrode placement.

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Efficacy of Nusinersen treatment over >4 years in Swiss patients with Spinal Muscular Atrophy: A multicenter observational study

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Background: Current disease-modifying treatments for spinal muscular atrophy have been shown to have a clear effect; however, data on long-term real-world outcomes remain scarce. In this study, we present outcome data of patients treated with nusinersen for at least 4 years.

Methods: This multicenter, observational study included 28 patients treated >4 years. Data were prospectively collected by the Swiss Registry for Neuromuscular Disorders. Treatment efficacy was evaluated using the motor function assessments CHOP-INTEND, HFMS/E, and RULM, depending on the patients' age and functional abilities. Additional analysis included respiratory and nutritional support and the PGI-I scale (Patient Global Impression of Improvement Scale).

Results: At treatment start, 7 patients were non-sitters, 14 sitters and 7 walkers. Treatment duration was between 4.1 – 6.3 years. Overall, 56% reached a clinically meaningful increase of ≥3 points in the HFMSE and 62% reached ≥2 points in the RULM. Most gains in motor function were achieved in the first year of treatment followed by reduced gain and stabilization. Eight patients were aged 0-2.5 years at treatment start and median gain of motor scores was 31.5 points (range -3 to 35) in the CHOP-INTEND and 10 points (range 6 to 32) in the HFMSE. Three patients started nutritional support and/or non-invasive ventilation during sleep throughout the treatment period. Fourteen patients were aged 3.5-16 years. They showed a more heterogeneous response to treatment, with a median gain of 1 point (range of -15 to 16) in the HFMSE. Four patients required nutritional support and/or non-invasive ventilation at treatment start, and three started support throughout the observation period. Six patients were aged >16 years. Of those, five patients achieved similar or higher motor scores (HFMSE/RULM) and none required nutritional support or chronic ventilation. After one year of treatment, 84% and after 4 years of treatment, 54% of all patients reported that their condition had improved in the last 6 months (PGI-I scale).

Conclusion: Our findings confirm sustained improvements of motor function in early-treated patients and disease stabilization in adults receiving nusinersen treatment. Additionally, patients consistently reported ongoing benefits over the treatment period.

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Guillain-Barré syndrom with surprisingly quick recovery after a 2-day course of IVIG

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Guillain-Barré syndrome (GBS) is an acute neuropathy often beginning after an infection. Therapy with intravenous immune globuline (IVIG) is a mainstay with recommended total dose but the dosage schedule varies. We report a short time course with favourable outcome.

Case report: The previously healthy 14-year-old adolescent presented to us with a history of progressive distal weakness over 3 days with inability to stand. She felt tingling paresthesias and a pronounced feeling of coldness in both feet up to the knee. On admission, several steps at a time were no longer possible. Examination revealed bilateral distal sensomotoric weakness of the lower extremity with extinguished motor reflexes. An MRI revealed poliradiculitis at the L5 level. The CSF showed significantly elevated proteins at 0.75 g/l with otherwise normal findings. An ENMG examination revealed a block of the right peroneal nerve as well as a prolonged distal motor latency of both tibial nerves. A suspected diagnosis of a sensorimotor variant of GBS was made and an immunomodulatory therapy with a total of 2g/kg IVIG was started. In an extensive literature search, we looked for data in favor of either the proposed short (2 days) or long (5 days) duration of therapy but found no advantages for either option. We decided to prefer the short course: The IVIGs could be administered on the first day of hospitalization, however, on the second day of treatment, headaches, nausea, and vomiting occurred and treatment was paused for one hour. Symptoms resolved and therapy could be completed. No anticoagulation was necessary due to ongoing mobilization. As a result, the patient recovered quickly in terms of her motor and sensory functions and was able to walk more than 10 steps at a time from the third day of hospitalization. Under physiotherapeutic care, she was able to reintegrate into her home after 10 days and did not require rehabilitation. From the 14th day after the start of therapy, she resumed school, initially partially via remote access from home. Within 3 months after the onset of the disease, the patient was able to resume her hobby of dancing.

We describe the case of an adolescent female patient with GBS with a surprisingly rapid response to a short 2-day cycle of immunoglobulins who subsequently showed an extraordinary good, complete and sustained remission of symptoms without complications.

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Advancing the Understanding of Dystrophinopathies in Switzerland: Disease Progression & Standards of Care

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Dystrophinopathies, including Duchenne (DMD), Becker (BMD) and intermediate (IMD) muscular dystrophy, are rare X-linked disorders caused by mutations in the dystrophin gene. These mutations lead to insufficient or absent production of dystrophin, a protein essential for muscle membrane stability. Corticosteroids (prednisolone and deflazacort) have been the cornerstone of DMD treatment, improving muscle function, delaying disease progression, and extending survival. However, there remains an urgent unmet medical need for more effective, disease modifying treatments. Managing patients with DMD requires a multidisciplinary approach and adherence to the Standards of Care (SOC) have a clear impact on outcome. While the SOC for DMD were updated in 2018, studies show variability in adherence to these guidelines based on treatment area, geographical region and age of patient. In Switzerland, it is unknown how well the SOC are adhered to. Since 2008, the Swiss Registry for Neuromuscular Disorders (Swiss-Reg-NMD) has collected comprehensive data on disease progression and treatments of patients with dystrophinopathies across all age groups and neuromuscular centres in Switzerland. Based on the data collected in the Swiss-Reg-NMD, this project aims to study the Swiss dystrophinopathy cohort in detail. The aim is to analyse the disease progression, to gain insights into the adherence to SOC in Switzerland and to identify barriers and facilitators to these. The disease progression will be assessed based on attributes such as age at loss of ambulation, age at spondylodesis, age at lung function decline <60% predicted, age at start of non-invasive ventilation, age at cardiac ejection fraction <50% and age at death. The adherence to SOC will be assessed by analysing the data collected in the Swiss-Reg-NMD and comparing them to the DMD SOC published in 2018. The results of this study will be crucial for advancing the understanding of dystrophinopathies in the Swiss cohort and for preparing to evaluate the effect of new disease modifying treatments in the real-world-setting. We will present the first results on disease progression.

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Fulminant cerebral cortical encephalitis (f-CCE) with anti-MOG IgG in a 3-Year-Old Child

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Background: Pediatric myelin oligodendrocyte glycoprotein antibody-associated disease (MOGAD) in children is typically related to central nervous system demyelinating disorders such as optic neuritis and acute disseminated encephalomyelitis (ADEM). Recently, non-ADEM cerebral phenotypes have been encountered. These cerebral cortical encephalitis (CCE) were first described in 2017 and are characterized by near-exclusive cortical gray matter inflammation. A subtype, FLAIR-hyperintense lesions in anti-MOG-associated encephalitis with seizures (FLAMES), presents with unilateral cortical lesions, nonrefractory seizures, treatment responsiveness, and minimal residual disability. More severe cases, known as fulminant-CCE

(f-CCE), involve bilateral, extensive cortical cytotoxic edema, carrying a much worse outcome. The risk factors for development and the underlying mechanisms remain unclear.

Case report: A 3-year-old boy with mild language delay was admitted with prodromal fever for 24 hours, followed by a first generalized seizure that resolved with midazolam. Two days later (day 0), he developed refractory status epilepticus, requiring intubation and prompt escalation to barbiturate coma therapy. Initial brain CT revealed no significant edema. Lumbar puncture showed mild pleocytosis (lymphocytic predominance) with positive oligoclonal bands (type 4). On day 3, a brain MRI revealed acute cytotoxic cortical edema with restricted diffusion affecting both hemispheres diffusely, causing uncal and tonsillar herniation. External ventricular drain was placed to reduce edema. An infectious workup was negative, and immunological testing revealed high anti-MOG antibodies (1:10240, norm <1:160), confirming MOGAD. Treatment with IV corticosteroids, intravenous immunoglobulin (IVIG), therapeutic plasma exchange (TPE), and rituximab led to clinical and radiological improvement, allowing cessation of the barbiturate coma on day 7, removal of the external drain on day 17, and extubation on day 20. Early physiotherapy, occupational therapy, and speech therapy were initiated. By day 60, he regained walking. By day 90, he has acquired basic social skills and progressed in non-verbal communication.

Conclusion: This case highlights the importance of MOG-antibody testing in children presenting with fulminant encephalitis. Pending further characterization of this MOGAD phenotype, aggressive immunotherapy and prompt neurorehabilitation are key to optimizing functional recovery.

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Photophobia, headache and vomiting: finding an etiology in the mist of symptoms

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Introduction: Primary Epstein-Barr virus (EBV) infections in children are common. Typical manifestation include sore throat and pharyngitis with enlarged tonsils. Less frequent manifestation of EBV infections can involve the central nervous system including meningitis and encephalitis. Therefore, diagnosis and treatment of EBV-related meningitis can be challenging.

Case presentation: A 5-year-old girl presented to the emergency department in December with severe recurrent vomiting, headache for 3 days and photophobia. She was in a poor general condition and had severe meningism. C-reactive protein and leukocytes in the blood were not elevated. A lumbar puncture revealed an elevated cerebrospinal fluid (CSF) pressure of 43 mmH2O. The CSF cell count (CC) was 190/µL (100% mononuclear cells), with elevated albumin levels (486 mg/L) and elevated total protein levels (0.67 g/L). Empiric intravenous treatment with ceftriaxone (100 mg/kg) and acyclovir (10 mg/kg) was started. After 2 days blood and CSF cultures as well as multiplex PCR and herpes simplex virus (HSV)-specific PCR from CSF remained negative. As there was no clinical improvement, magnetic resonance imaging (MRI) was performed which showed no intracranial abscess or signs of encephalitis. A second lumbar puncture revealed still elevated intracranial pressure of 24.5 mmH2O. The CSF CC was 89/µL (100% mononuclear cells), with a total protein of 0.63 g/L and CSF/serum glucose ratio of 0.43. EBV serology was suggestive of an acute infection with EBV (EBV VCA IgG 203 U/mL, EBV VCA IgM 34 U/mL and EBNA <3 U/mL). Additional PCR testing confirmed EBV in CSF. Treatment was initiated with ganciclovir (5 mg/kg intravenous) and prednisone (2 mg/kg orally). Neurological symptoms improved rapidly within 24–48 hours. Steroids were reduced after 3 days to 1 mg/kg for 2 days. The treatment with ganciclovir was switched to oral valganciclovir (25 mg/kg/day) after 5 days continued for another 2 days.

Conclusion: This case illustrates that an acute EBV infection may be suspected when CSF analysis shows exclusively mononuclear pleocytosis, elevated CSF pressure, and hypoglycorrhachia. To confirm the diagnosis EBV PCR testing in CSF is essential. The optimal treatment for EBV meningitis is unclear. In literature, the benefits of acyclovir, ganciclovir, and corticosteroids are debated. In our case, we chose ganciclovir combined with steroids for treatment as there was no initial improvement with acyclovir.

P 50

Diagnostic and therapeutic challenges in a child with headache, vomiting and coma after a minor head trauma – also consider the rare of the rare

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An 11-year-old girl presented with headache, vomiting and somnolence after a minor head trauma. Neuroimaging revealed a relevant left hemispheric cortical edema, EEG a severe slowing of the left hemisphere. Cerebrospinal fluid analysis was unremarkable. Six years earlier, the patient had suffered focal status epilepticus with Todd's paresis after a minor head trauma, with at that time right hemispheric cortical swelling, and complete recovery from it after several weeks. Suspecting hemiplegic migraine, treatment with methylprednisolone 100mg/d and levetiracetam was started, and targeted genetic testing initiated. Because of persistent coma after four days a follow-up imaging was performed. It showed new severe high-grade stenosis in the anterior circulation with vascular wall enhancement compatible with vasculitis, with perfusion delay and diffusion restriction. Hours later, the patient manifested hemiplegia of the left arm and focal seizures. Treatment with high-dose corticosteroids and acetylsalicylic acid was started to cover the neuroradiologically suspected vasculitis with consecutive stroke. As unilateral vasculitis seemed unlikely, literature search was performed, and revealed two publications (one case report, one case series including 8 patients) on vasospasms in children with familial hemiplegic migraine. Treatment with intravenous verapamil was added.

The vascular stenosis resolved within days, the suspected stroke was not confirmed in follow up imaging. Over the course of months, the girl showed complete normalization of clinical, neuroimaging and electroencephalographic findings. Genetic testing confirmed an ATP1A2 mutation, establishing the diagnosis of FHM type 2.

In familial hemiplegic migraine, severe vasospasms can occur and may rarely lead to dramatic clinical and neuroimaging findings, leading to confusion in diagnosis. The ideal therapy is not known. High-dose corticosteroids, intravenous verapamil (especially in CACNA1A mutation), or ketamine intranasal are the mainstay in acute treatment. Prophylactic treatment between attacks can be useful.

Unusual clinical presentation and evolutions warrant thinking of rare disorders. Neurologic deteriorations after minor head trauma is a typical presentation of familial hemiplegic migraine.

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A Rare Mutation in the Thyroglobulin Gene as a Cause of Congenital Goitrous Hypothyroidism: A Case Report

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Background: Congenital hypothyroidism (CH) is a leading cause of preventable intellectual disability, commonly due to thyroid dysgenesis or hormone synthesis defects. Rare mutations in the thyroglobulin (TG) gene, involved in thyroid hormone synthesis, can also cause CH. This case report highlights the diagnostic challenge of identifying a TG gene mutation as a cause of congenital goitrous hypothyroidism, particularly when thyroid function tests show low levels of thyroglobulin (TG) despite the presence of an enlarged thyroid gland.

Case presentation: A newborn was referred on day 6 of life due to elevated TSH levels in the newborn screening, with moderately elevated TSH (50mU/L). Upon presentation at our outpatient clinic, lab results confirmed the findings (TSH 74 mU/l, fT4 19 pmol/l) with slightly decreased fT4 and thyroglobulin in the lower normal range (40,1µg/L, Ref. >35). We initiated L-thyroxine substitution for neuroprotection at a dose of 13 µg/kg body weight. No signs of autoimmune disease were found in either the mother (who is euthyroid) or the child; iodine contamination was ruled out. Ultrasound showed an enlarged thyroid gland with total volume above 97th percentile. The bone age corresponded to the chronological age. Genetic testing identified a rare mutation in the TG gene, which has been described previously to cause congenital goitrous hypothyroidism due to TG deficiency (Clin Endocrinol Oxf 2011 PMID: 21128992).

Discussion: Relatively low TG levels in the presence of goiter should raise suspicion for a TG gene mutation. The TG gene (c.2206C>T; classified as a pathogen according to ACMG) is essential for thyroid hormone production, and mutations in this gene can impair hormone synthesis, resulting in hypothyroidism.

Conclusion: This case contributes to the limited number of reports describing TG gene mutations as a cause of congenital goitrous hypothyroidism and emphasizes the need for molecular genetic testing in such cases.

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Klinefelter Syndrome and type 1 Diabetes Mellitus: A Case Report

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Background: Klinefelter syndrome (KS), characterized by the presence of an additional X chromosome (47,XXY, classic form), is the most common sex chromosome abnormality in males, affecting 1 in 500–1,000 live births. The syndrome is associated with a spectrum of clinical features and the most frequent characteristics are small testes, azoospermia, hypergonadotropic hypogonadism and cognitive impairment. KS predisposes to increased risk of metabolic syndrome and a higher prevalence of type 2 diabetes mellitus (T2DM) is well-documented. The association between KS and autoimmune diseases, including type 1 diabetes mellitus (T1DM), remains less understood: various studies suggest that the hypogonadism due to KS creates insulin resistance that may promote beta cell destruction.

Case report: A 15-year-old male was admitted to the emergency room for polydipsia, nocturia and recent weight loss and upon further investigation was diagnosed with type 1 diabetes mellitus (T1DM). During the diagnostic evaluation, clinical examination revealed hypotrophic (4 ml) and spherical testes despite a nearly complete pubertal development. These findings, combined with biochemical evidence of high luteinizing hormone (LH) and follicle stimulating hormone (FSH) levels and low testosterone levels, prompted further genetic investigation, leading to the diagnosis of KS. The patient's medical history included delayed speech milestones, necessitating speech therapy during early childhood, which is a known neurodevelopmental manifestation in KS. Currently, the patient is under follow-up at our endocrinology clinic, receiving multidisciplinary management for both T1DM and KS.

Conclusion: This case highlights the concurrence of KS and autoimmune diseases such as T1DM, demonstrating the importance of considering KS in pediatric patients who present with diabetes alongside signs of pubertal abnormalities or developmental delays and a need for structured check-list for the follow-up in KS pediatric patients. Raising awareness among pediatricians is essential to ensure early detection and optimal multidisciplinary management of this patient population.

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Predicting Treatment Quality in Children and Adolescent with Congenital Adrenal Hyperplasia with Standard Lab Analyses and Clinical Parameters

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Introduction: Currently, assessing treatment quality in congenital adrenal hyperplasia (CAH) largely relies on bone age and steroids. It requires a lot of expertise and is unsatisfactory. A reliable biochemical indicator is still lacking. Most research on this topic has focused on steroids metabolites, which require highly specialized equipment. However, no studies have explored whether treatment quality can be predicted using more basic laboratory parameters, although many have investigated the long-term effects of over- or undertreatment on cardiovascular risk factors and metabolism.

Aim: To develop a score predicting the treatment quality in children and adolescent with CAH and to evaluate its performance.

Methods: We included patients with confirmed CAH due to mutations in CYP21A2 affecting 21-hydroxylase from four major paediatric endocrinology centres. Clinical parameters and standard lab analyses (including steroid and lipid profiles, glucose and insulin) were assessed at 3 outpatient visits. Patients were classified as under-, over- or well-treated at each visit. Based on univariant and then multivariant logistic regression results, we developed a predictive score for undertreatment.

Results: We included 233 visits from 81 patients in our analyses, 37 of which were female and 43 prepubertal. We excluded the 10 visits with overtreatment. HDL (p = .005), insulin

(p = .002), renin (p = .001), blood glucose (p = .047), and SDS of height (p = .012) were significantly different in the undertreated vs. well treated group. The final model, based on 60 visits, included insulin in mU/L (coefficient, 1.15) and renin in nG/L (1.038). With a threshold of >= 30-36, 19 visits (32%) were classified as undertreated. Specificity was 84% and sensitivity was 71%. Overall, 94% of visits were classified correctly. The AUC ROC was 0.785.

Conclusion: This score is able to predict undertreatment in children and adolescent with CAH using serum insulin and renin levels. Depending on clinical assessments or combination with other parameters, the threshold can be adjusted to optimize sensitivity or specificity. Although further studies are needed to confirm these findings, we recommend routinely measuring renin and insulin in individuals with CAH. Lipid status, renin and insulin may be particularly beneficial as indicators of treatment quality, as they are strongly associated with cardiovascular and metabolic risk, which are the main long-term complications of CAH.

P 54

Correlation Between Growth Hormone Therapy and Type 1 Diabetes

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Background: Growth hormone therapy is used to treat growth hormone deficiency, but it can affect carbohydrate metabolism and potentially increase the risk of developing diabetes. Growth hormone increases insulin resistance in the muscle and adipose tissues, and its interaction with insulin-like growth factor can worsen this effect. Even though there is no clear evidence in literature of a connection between Type 1 diabetes and growth hormone treatment, and the coexistence of Type 1 diabetes and growth hormone deficiency is rare, diabetes should be closely monitored in patients undergoing growth hormone therapy. We describe a case of a patient receiving growth hormone therapy who was diagnosed with Type 1 diabetes.

Case report: An 18-year-old male patient, with a history of growth hormone deficiency and a pituitary gland cyst, has been receiving somatropin treatment for 2 years and is responding well clinically. In July 2024, elevated HbA1c levels were reported on two separate occasions. Subsequently, diabetes-related antibodies were found to be positive, although he didn't show symptoms of diabetes. With these findings, the patient was diagnosed with subclinical type I diabetes. The patient has started therapy with long-acting insulin and dietary modification. He is currently monitored and short acting insulin is being considered as part of his treatment.

Conclusion: Although the relationship between GH therapy and type I diabetes is poorly understood, this case highlights the importance of glucose control in patients undergoing growth hormone therapy. Clinicians need to educate patients about early signs of diabetes and perform regular metabolic screenings. Prompt detection and intervention are critical for managing the complications and further providing the most appropriate care for patients receiving growth hormone therapy.

P 55

Unveiling Monogenic Diabetes in Pediatrics: The role of early genetic testing in autoantibody-negative cases

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Background: Monogenic diabetes is a rare form of diabetes caused by a single gene mutation, often misdiagnosed as type 1 or type 2 diabetes. Although it makes up less than 5% of diabetes cases, it carries profound implications for diagnosis and treatment. Unlike autoimmune or insulin-resistant diabetes, monogenic diabetes may exhibit specific features such as a family history, mild hyperglycemia, or a non-progressive course. The absence of autoantibodies and specific clinical characteristics can prompt genetic testing, which has significant implications for treatment strategies, family counseling and long-term management.

Aim: To highlight the importance of early genetic testing in pediatric patients with autoantibody-negative diabetes through two illustrative cases.

Methods: We analyzed the clinical presentation, diagnostic process, and management of two pediatric patients with autoantibody-negative diabetes, focusing on the role of genetic testing.

Results: Case 1: a 15-year-old boy presented with inaugural diabetes, significant hyperglycemia, and HbA1c >12%. Autoimmune markers were negative, leading to genetic testing, which identified a mutation in the HNF1A gene consistent with monogenic diabetes. A individualized treatment plan involving oral hypoglycemic agents replaced insulin therapy, resulting in improved metabolic control. Case 2: a 13-year-old English girl presented with inaugural diabetes, marked hyperglycemia (glucose >18 mmol/L) and HbA1c >14%. Autoantibodies were also negative, raising suspicion of monogenic diabetes. Although genetic testing is pending, the clinical presentation highlights the challenges of diagnosing non-autoimmune diabetes.

Conclusion: These cases illustrate the critical role and the importance of early genetic testing in guiding management of patients with autoantibody-negative diabetes. A large study conducted in the United Kingdom demonstrated an increased prevalence of monogenic diabetes, potentially attributed to superior diagnostic capabilities due to advanced genetic and biomarker testing infrastructure. This suggests that monogenic diabetes may be underdiagnosed in other countries. Enhanced access to genetic testing and targeted screening can refine the classification and management of diabetes, leading to more personalized and effective therapeutic strategies. A multidisciplinary approach remains essential to optimize care and outcomes for this unique patient subset.

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Spontaneous subdural haematoma in a child: Case report

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Background: Subdural haematoma (SDH) in a child is rare and can be caused by abusive head trauma (AHT), coagulopathy, accidents, metabolic disorders, infections of the central nervous system or prior neurosurgical procedures. AHT is a diagnosis of exclusion, considered when clinical presentation cannot be explained by natural diseases or accidental trauma. Cases of spontaneous SDH also have been described in current literature. With this case report we aim to highlight the challenges of diagnosing a spontaneous SDH.

Case presentation: We present a 2-year-old, former premature male with acute onset of lower back pain and refusal to walk. The patient had no previous history of trauma or medical issues. His gait appeared with a stiffness of the lower back, no signs of improvement with analgesic treatment. Repeated extensive laboratory findings showed no signs of inflammation, coagulopathies or metabolic disorders. Magnetic resonance imaging (MRI) performed 5 days after the appearance of symptoms revealed extensive, most likely subacute, subdural hematoma intracranial and spinal, especially lumbar. Eye fundoscopic exam and study of fractures as well as detailed social and third-party history remained without any signs of AHT. A genetic analysis resulted in a variant of unclear significance of the GCDH gene in heterozygous expression. The patient was discharged with spontaneous, complete remission of symptoms. One month after discharge the child was without complaints, physical examination showed no residues after SDH.

Conclusion: Identifying the source of a SDH can be challenging, especially when it could be attributed to abuse. Thus, a multidisciplinary approach is necessary to diagnose, investigate, and manage each case.

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Little patients, big cholesterol: Homozygous familial hypercholesterolemia is a challenging condition in pediatric patients - a case report.

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Background: Familial hypercholesterolemia (FH) is a genetic disorder characterized by elevated low-density lipoprotein cholesterol (LDL-C), leading to complications such as xanthelasma, xanthoma of tendons, early-onset atherosclerosis, and coronary heart disease in young age.

Case: An 8-year-old boy presented with multiple xanthomas on various body parts but was otherwise asymptomatic. Family history revealed high cholesterol requiring treatment in his father and paternal grandmother. Laboratory investigations showed significantly elevated blood lipids, prompting genetic analysis, which confirmed FH due to a compound heterozygous missense mutation in the LDLR gene, consistent with homozygous FH (HoFH). Baseline assessments revealed normal liver enzymes and creatine kinase levels, mild aortic insufficiency on echocardiography, and no evidence of calcifications or plaques on subsequently performed cardiac computed tomography (CT). Pravastatin therapy was initiated, but due to insufficient LDL-C reduction, ezetimibe was added after 8 weeks. Regular cardiology and endocrinology follow-ups are planned.

Discussion: HoFH is extremely rare in children (~1:1'000'000), often identified through familial screening or when children present with xanthelasma. Without treatment, most individuals experience cardiovascular events by adolescence, with survival rarely extending beyond the second decade. Routine cardiac imaging, including Doppler echocardiography and CT angiography, is crucial for detecting structural changes. A stepwise therapy plan is critical, starting with a strict diet and pharmaceutical lipid-lowering therapy. However, most medications are not approved for young children, and their efficacy depends on residual LDL-receptor activity. Conventional therapies are often insufficient for HoFH. While PCSK9 inhibitors are approved for individuals aged 10 years and older, other options, such as lipoprotein apheresis or newer agents like lomitapide, may need to be considered in severe cases.

Conclusion: Early detection and diagnosis of HoFH is critical for preventing severe cardiovascular complications in young patients. This case highlights the importance of family history assessment, genetic testing, and routine cardiovascular surveillance in children at risk. A tailored, stepwise approach, combining diet, pharmaceutical interventions, and advanced treatments when necessary, is essential for improving long-term outcomes.

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Dyslipidaemia in Children and Adolescents with Severe Obesity: The Bern Obesity in Childhood and Adolescence Biorepository (BOCAB)

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Background: Dyslipidaemia is causal to subclinical atherosclerosis in youth and cardiovascular morbidity and mortality in adulthood. However, data on dyslipidaemia in children and adolescents with severe obesity are sparse. With the Bern Obesity in Childhood and Adolescence Biorepository (BOCAB), we aimed to estimate proportions of dyslipidaemia, and cross-sectional associations between anthropometry and dyslipidaemia.

Methods: Anthropometric data (body mass index [BMI], percent of the 95th BMI percentile for age and sex [%BMIp95], waist circumference [WC], WC-to-height ratio [WHtR], percentage body fat [%BF] and muscle mass [%MM]), and fasted lipid profiles (low-density-cholesterol [LDL-C], high-density-C [HDL-C], non-HDL-C and triglycerides [TG]) were analysed using linear regression models adjusted for age, sex, self-reported screen time and physical activity.

Results: Data on 559 participants (52.2% males), aged 6-17 years (median [IQR] 11.8 years [9.7-14.1]), with a mean (SD) %BMIp95 of 125.0% (17.9), and WHtR of 0.6 (0.1) were analysed. Means and proportions (%) for dyslipidaemia (according to 2018 AHA guidelines) were: LDL-C 2.6 mmol/L (0.7), 12.9%; HDL-C 1.2 mmol/L (0.3), 21.8%; non-HDL-C 2.9 mmol/L (0.8), 14.0%; and TG 1.2 mmol/L (0.7), 25.4%. Overall, 249 (44.5%) participants had dyslipidaemia, including 121 (21.6%) with ³2 lipid measures affected. Mean estimates (95% confidence interval) from adjusted linear regressions between WC and WHtR with LDL-C were 0.01 (0.00-0.02) and 1.97 (0.82-3.12); with non-HDL-C 0.01 (0.01-0.02) and 2.33 (1.10-3.57); with TG 0.01 (0.00-0.02) and 1.37 (0.31-2.43); and with HDL-C -0.01 (-0.01 to -0.00) and -0.95 (-1.37 to -0.54). BMI, %BMIp95 and %BF were associated with TG and HDL-C.

Conclusion: Our results reveal high proportions of dyslipidaemia in children and adolescents with severe obesity. Other than measures of mass (BMI, %BMIp95) or body composition (%BF, %MM), measures for central adiposity (WC and WHtR) were consistently associated with all lipid profile measures in this high-risk population for premature cardiovascular disease.

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Congenital adrenal hyperplasia as a possible genetic etiology of peripheral precocious puberty in males

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Introduction: Precocious puberty is a commonly observed presentation in paediatric practice. While the majority of girls with precocious puberty exhibit no identifiable etiology, boys are more likely to have an underlying medical condition. One significant cause of peripheral precocious puberty in boys is congenital adrenal hyperplasia (CAH), resulting from a deficiency of 21-hydroxylase, an autosomal recessive disorder caused by mutations in the CYP21A2 gene. In contrast, central precocious puberty is more common in girls. The severity of this disease is related to the degree to which the mutations affect enzyme activity.

Case report: We present the case of a 12-year-old boy with a premature adrenarche and a rapid onset of puberty starting at the age of 11. At the endocrinological evaluation the patient presented: height 170 cm, weight 64.9 kg (5.4 SD), BMI 22.4 kg/m² (1.4 SD), Tanner stage A?,PP?,G?, testicles 3x2 cm bilaterally, bone age according to Greulich&Pyle method of 14 years and a predicted height of 188cm; hormonal exams showed a mature hypothalamus-pituitary-gonadal axis (HPG). At the clinical examination he presented with moderate acne on the forehead, greasy hair and corpulence diffuse adiposity. In view of the clinical picture, an adrenocorticotrophic hormone (ACTH) stimulation test was performed, which confirmed the diagnosis of late-onset androgenital syndrome. The patient was commenced on treatment with Hydrocortone 15mg/day. Genetic testing was then carried out, revealing the V281L mutation (c.841G>T) in heterozygosity. The patient was scheduled for a follow-up with laboratory analysis, hormone monitoring and left hand X-ray to assess growth and bone age until final height was reached. On replacement therapy with Hydrocortone, the clinical course was satisfactory, with regular bone maturation and puberty progression. In addition, a dietary consultation due to the increased BMI, and a future genetic consultation to inform about the increased risk in his offspring were recommended.

Conclusion: Non-classic CAH (NCCAH) is a less severe form of the disease, characterized by 20-50% of normal 21-hydroxylase enzyme activity, compared with 0-2% in the classic form. In view of the later clinical presentation of CAH in males, it is important to consider this disorder in cases of male precocious puberty by assaying 17-OHP and investigating genetics.

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Investigating the pharmacological chaperone effect of sepiapterin in phenylketonuria

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Background: Phenylketonuria (PKU) management primarily relies on dietary restrictions and tetrahydrobiopterin (BH4) supplementation, yet not all patients benefit, particularly those with BH4-unresponsive phenylalanine hydroxylase (PAH) variants. Sepiapterin, may act as a BH4 precursor and a pharmacological chaperone, and this dual-action offers a novel and potentially more efficient approach for stabilizing PAH enzyme variants.

Methods: We undertook a comparative analysis to elucidate sepiapterin effects versus BH4, including in silico docking and in vitro assays. Specifically, we assessed the enzymatic activity of PAH in COS-7 cells overexpressing PAH wild-type (WT) and protein variants R68S, R261Q, and Y417H, upon dose-dependent treatments. To further understand the role of sepiapterin as a pharmacological chaperone, we examined its binding and stabilization impact through isothermal titration calorimetry (ITC), and thermal-shift and enzymatic activity assays on PAH WT and variants.

Results: Docking analysis revealed sepiapterin's binding within the PAH enzyme, in the BH4 pocket with distinct interactions. Sepiapterin enhanced the activity of specific PAH variants dose-dependently more effectively than BH4. Sepiapterin direct binding was confirmed by ITC, with Kd (dissociation constant) values indicating a favorable affinity for both wild-type and variants (<20 µM), notably sepiapterin presented the highest affinity towards R68S (4.4 ± 3.7 µM). Sepiapterin induced thermal stabilization and by a dose-dependent direct inhibitory effect of PAH WT and variants. The inhibitory effect of sepiapterin further supports its binding to the catalytic center, similarly to the co-factor BH4.

Discussion/Conclusion: Our results support sepiapterin's independent effect as a pharmacological chaperone for PKU; it functions not only as a co-factor precursor but also binds and stabilizes PAH variants independently, underscoring its potential to improve treatment outcomes, particularly for those previously unresponsive to BH4.

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Protracted rotavirus infection following vaccination in an child with transient hypogammaglobulinemia of infancy

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Background: Rotavirus (RV) is a leading cause of gastroenteritis in young children responsible for 34.2% of hospitalizations among Swiss children under five (2015-2019). Since 2024, routine immunization with the oral live-attenuated rotavirus vaccine (RVV, Rotarix®) has been recommended in Switzerland. Severe RV infections, including vaccine-derived cases, have been reported in patients with inborn errors of immunity (IEI) (2-5). Here, we describe an infant with chronic diarrhea and protracted RV shedding following RVV, later diagnosed with transient hypogammaglobulinemia of infancy.

Case report: A 4-month-old infant presented with chronic diarrhea, feeding difficulties, and insufficient weight gain persisting for over 6 weeks. Symptoms begin 10 days after receiving the first RVV dose at 9 weeks of age. RV was detected in stool by PCR 55 days post-vaccination. Initial immunological work-up excluded severe IEI, with normal newborn screening for severe IEI (TREC/KREC assay) and unremarkable findings apart from slightly reduced CD8+ T lymphocytes[BC1] . Symptoms resolved by 5 months of age, with improved weight gain following nasogastric feeding, proton pump inhibitor therapy, and cow's milk protein-free diet. Immunological reassessment at 8-10 months revealed panhypogammaglobulinemia (IgA 0.22g/l, IgM 0.22g/l, IgG 1.93g/l), which normalized by 14 months, alongside adequate vaccine responses and CD8+ T-cell recovery.

Discussion: RVV is safe and effective in preventing severe rotavirus gastroenteritis, reducing hospitalizations by over 80% (6-7). Severe RVV-associated gastroenteritis, characterized by prolonged viral shedding (>7 weeks), has been documented in infants with severe combined immunodeficiency (2-5) (8). In this case, RV shedding over 6 weeks and associated symptoms resolved without long-term sequelae. The patient's transient immunological abnormalities suggest delayed maturation of adaptive immunity rather than a primary immundeficiency. While RV sequencing to confirm a vaccine-derived strain was not performed, this case underscores the need for thorough immunological evaluation in infants with protracted diarrhea and prolonged RV shedding after vaccination.

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Group A Streptococcal Arthritis of the Temporomandibular Joint in a 7-year-old Boy

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Background: Septic arthritis of the temporomandibular joint (TMJ) is a rare condition in children. The most commonly isolated organisms are Staphylococcus aureus and Group A Streptococcus (1,2). Bacterial spread to the TMJ typically occurs through hematogenous seeding or contiguous spread from otitis media or odontogenic infections (3). Prompt diagnosis using CT scan, drainage, and antibiotic therapy is crucial to prevent local complications, such as fibrosis and joint ankylosis (4).

Case presentation: A previously healthy, 7-year-old, non-vaccinated boy presented to the emergency department with a 3-day history of trismus, left-sided torticollis, fever of 38.5°C, left preauricular swelling and erythema, and pain in the left TMJ. The symptoms were preceded by viral rhinopharyngitis. Physical examination revealed no signs of otitis media or dental infection, but there was paronychia on his right middle finger. Blood tests showed a CRP of 170 mg/L and leukocytosis of 25 G/L. A CT scan revealed an effusion within the left TMJ, with expansion of the joint space to 6 mm on the left compared to 3mm on the right, without any locoregional complications. The patient was initially treated with intravenous ceftriaxone, followed by co-amoxicillin the next day, before being transferred to our tertiary care center. Under general anesthesia, ultrasound-guided arthrocentesis with washout of the left TMJ was performed, yielding 3 mL of hemorrhagic and purulent fluid. Culture of the joint fluid was positive for Streptococcus pyogenes. Blood cultures were negative. Intravenous antibiotics were continued for 24 hours post-intervention, followed by a 14-day course of oral co-amoxicillin. The patient became afebrile, and the swelling decreased rapidly. He then began mouth-opening physiotherapy to prevent joint ankylosis.

Discussion: Despite the rarity of this condition, prompt CT imaging guided by clinical signs allowed for a rapid diagnosis and timely management with targeted antibiotics and drainage. The microbiological findings were consistent with those reported in the literature. While the exact source of infection remains unclear, it is likely to be hematogenous, originating either from the paronychia or from the oropharyngeal mucosa in the context of a viral infection.

Conclusion: Trismus, fever, and preauricular swelling should raise suspicion for TMJ septic arthritis. Prompt treatment is essential to prevent complications.

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Mud- and water games

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Introduction: Fever in returning travellers is a major challenge. Although the presentation is usually non-specific and most of the infections are mild or self-limiting, we need to identify the relevant conditions.

Case: A 7-year-old boy was referred to our emergency room-with high fever and abdominal pain for three days with suspected acute appendicitis. Further anamnesis and examination revealed headache, pronounced limb, particularly calf pain, occasional vomiting, as well as a recent return from a 3 weeks trip to Costa Rica. 3 days later, the patient presented again with persistent fever and symptoms, now additionally accompanied by severe headaches, photophobia and conjunctival hyperemia. Clinically he showed meningismus. Subsequent lumbar puncture findings included 591/µL leukocytes and mildly elevated protein. A comprehensive PCR-based CSF panel was negative, as were cultures. The patient underwent symptomatic treatment, including rehydration and analgesia. The symptoms improved over the next few days. Based on the travel behaviour including playing in water and mud for many hours, clinical symptoms and aseptic meningitis, serology for leptospirosis revealed positive IgM with (yet) negative IgG.

Discussion: Finally our patient showed an unusual aseptic meningitis after a long run of high fever, limb and abdominal pain and conjunctivitis. A conclusive diagnosis cannot be made with the single testing of leptospirosis IgM. However, the symptoms, appropriate anamnesis and course would be consistent with a leptospirosis infection. Leptospirosis is a globally important zoonosis with a wide variation in clinical presentation. The transmission usually occurs through the skin in contact with water or mud, which is contaminated by urine of Leptospirosis-diseased animals. The initial phase is usually nonspecific, although conjunctival suffusions and pain in the calf and lumbar region are characteristic symptoms. In a second phase children may present with fever and aseptic meningitis with mostly self-limiting course. A feared severe course with liver-, renal failure and bleeding is fatal in about 20%.

Conclusion: A precise anamnesis and epidemiologic knowledge is important in returning travellers to initiate the right diagnostics in order to identify relevant conditions. A second look and additional symptoms may help to find the correct diagnosis.

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A summer dip with unexpected consequences: a case report

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Background: Leptospirosis, a zoonotic disease caused by the spirochetal Leptospira spp., is predominantly associated with human infection in tropical regions(1,2). A growing incidence of leptospirosis has been observed in non-tropical areas(3), underscoring the need to recognize this potentially life-threatening condition even in non-endemic regions. We report the case of a 7-year-old boy presenting to our emergency department (ED) with persistent fever, conjunctivitis, meningoencephalitis, and acute kidney injury due to leptospirosis. The infection was attributed to exposure to the Rhein River in Switzerland, a habitat of beavers—animals known to asymptomatically excrete

Leptospira spp. in their urine, facilitating pathogen transmission through contact with contaminated freshwater.

Case report: The patient presented to our ED with a one-week history of fever, headache, bilateral conjunctivitis, and significant weight loss (7 kg). Laboratory investigations revealed elevated inflammatory markers, bicytopenia, acute kidney injury, and cerebrospinal fluid (CSF) pleocytosis. Empiric antibiotic therapy with ceftriaxone was initiated, leading to rapid clinical improvement. CSF cultures were sterile, serological tests ruled out neuroborreliosis and tick-borne encephalitis. Further history-taking revealed that the patient had been swimming in shallow sections of the Rhein River prior to symptom onset—a habitat of beavers. Subsequent serological testing confirmed the diagnosis of leptospirosis with highly positive IgM antibodies. After three days of intravenous ceftriaxone, antibiotic therapy was switched to oral doxycycline for another 13 days, leading to complete clinical recovery.

Discussion: Leptospirosis in humans typically follows direct or indirect exposure to the urine of infected animals, frequently through recreational activities in contaminated water. While rodents such as rats are common vectors, other animals, including beavers, can also serve as reservoirs(4). Despite the lack of favorable environmental and socioeconomic conditions for Leptospira spp. proliferation in Switzerland(5,6), leptospirosis should be considered in patients with nonspecific febrile illnesses, particularly when the history reveals exposure to potentially contaminated freshwater sources inhabited by rodents. This case emphasizes the growing global reach of leptospirosis (3,7), even in regions not traditionally endemic, and the need for an increased awareness among healthcare providers.

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Diagnostic Value of Clinical Presentation in Pediatric Neuroborreliosis: A Retrospective Study from the Swiss Jura Region

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Background: Neuroborreliosis (NB) is a rare but significant neurological manifestation of Lyme disease, particularly in the pediatric population. Common presentations include headache and facial paralysis. Key diagnostic investigations consist of blood serology and lumbar puncture. Early diagnosis and subsequent antibiotic therapy are critical to prevent long-term complications.

Case study: In a retrospective analysis from the Jura region of Switzerland, we studied a cohort of 14 pediatric patients with suspected NB who presented with either headache or facial paralysis. Initial Borrelia serological testing was performed in all cases, revealing 3 positive, 2 inconclusive, and 9 negative results. Among these patients, 6 presented a stronger clinical suspicion based on specific presentations. Patients with headache exhibited characteristic red flags including nocturnal awakening, nocturnal or morning vomiting, and lack of response to analgesic therapy - all while maintaining normal clinical examination (including normal neurological and fundoscopic findings) and absence of fever. Similarly, patients with facial paralysis demonstrated distinct features from Bell's palsy, as it was never an isolated symptom but rather accompanied by headache, neck stiffness, profound fatigue, or loss of appetite. Those 6 patients underwent lumbar puncture (LP), all positive for Borrelia in cerebrospinal fluid (CSF). Cross-referencing these findings revealed varied patterns: in two cases, both serology and CSF were positive; in two cases, inconclusive serology was confirmed as positive in the CSF; and in two cases, serology was negative while the CSF was positive. Notably, these characteristic clinical features were absent in patients who did not have confirmed NB.

Conclusion: Our findings highlight that in endemic regions, NB should be strongly considered in pediatric patients presenting with specific clinical patterns. We recommend prioritizing LP over serology when additional testing is deemed necessary. The presence of red flags in headache cases and the multi-symptom nature of facial paralysis were found to have such a high positive predictive value, that it questions whether LP is indeed essential in cases with such highly suggestive clinical presentations. However, larger scale studies would be needed to confirm these findings and establish more definitive guidelines.

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Bridging the Past and Future: Transforming Analog Vaccination Records to Digital Data with Artificial Intelligence to improve Patient Care and Enable Research

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Background: Vaccinations are essential for preventing infectious diseases and protecting public health. In Switzerland, vaccination records remain fragmented and predominantly analog, stored as handwritten entries in paper vaccine cards or booklets. This impedes efficient data management and sharing, particularly as individuals navigate various points in the healthcare system. Consequently, incomplete information and inadequate vaccination coverage expose healthy individuals and high-risk patients to preventable risks. The national electronic patient dossier (EPD) initiative is progressing slowly, with less than 1% of the population enrolled. Historical vaccinations are recorded as PDFs/JPGs, lacking true integration and functionality.

Aim: We aim to develop an AI-powered solution to extract vaccination data from scanned vaccination cards formatted by the Federal Office of Public Health (FOPH) and integrate the digitalized data into the electronic health record (EHR) system "EPIC" at Lucerne Cantonal Hospital, ensuring future compatibility with the EPD and other EHRs.

Methods: Advanced Artificial Intelligence (AI) algorithms, including Optical Character Recognition (OCR), extract key details: vaccine names, lot numbers, and administration dates. Information Extraction (IE) further categorizes these entities and links vaccine names to corresponding administration dates. Challenges include variations in text, handwriting styles, and multilingual formats.

Results: A dataset of 300 vaccination records from children at KidZ was manually annotated for training. For testing, 40 samples were used, of which 33 were accurately digitized without corrections. The remaining samples contained minor, easily rectifiable errors. The AI model achieved an average accuracy of 83% in this pilot study.

Discussion: Additional training with larger data volumes will substantially enhance model accuracy. Digitized vaccination data will improve patient care by enabling quick access to comprehensive vaccination records and supporting personalized vaccination recommendations. It also paves the way for vaccine-related research, which is vital for individual and public health efforts to prevent infectious diseases.

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Infants aged 0-3 months with E. coli urinary infection: Is lumbar puncture necessary?

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Background: A 43-day-old infant was diagnosed with Escherichia coli meningitis concomitant with an urinary tract infection caused by the same pathogen. The infant initially presented with a febrile condition without a clear source. A diagnosis of acute pyelonephritis was subsequently made due to the presence of Escherichia coli in the urine analysis (catheterization). As the clinical condition worsened, suspicion of a central nervous system infection arose. Lumbar puncture results later confirmed this second diagnosis.

Objective: The objective of this study is to determine whether a lumbar puncture is necessary for young children (0-3 monhs old) presenting with an urinary tract infection caused by Escherichia coli.

Methodology: We conducted a systematic review across several databases (U.S. National Library of Medicine, Web of Science, Google Scholar). Eligible articles were those discussing the relationship between E. coli urinary tract infections and meningitis in children aged 0 to 3 months.

Results: Several studies have investigated the relationship between Escherichia coli urinary tract infections (UTIs), cerebrospinal fluid pleocytosis, and bacterial meningitis. The data suggest that, although rare, bacterial meningitis can coexist with a UTI in young infants, particularly those under 28 days of age. However, systematic lumbar punctures are not always necessary in this age group if the infant is considered to be at low risk of severe bacterial infection, based on clinical and biological criteria. It has been demonstrated that the prevalence of bacterial meningitis in infants with a positive urine analysis is not higher than that observed in infants with a negative urine analysis. Thus, it is shown that the coexistence of bacterial meningitis with an E. coli urinary tract infection in infants exists but is rare, suggesting the potential to reduce the number of routine lumbar punctures in these patients.

Conclusion: The necessity of performing a lumbar puncture in infants aged 0 to 3 months with E. coli urinary tract infection is a debated topic. Recommendations vary depending on the child's age and clinical condition. The authors suggest that the decision to proceed with a lumbar puncture should be based on additional clinical criteria rather than solely on the results of urine analysis.

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Acute aseptic meningitis temporally associated with trimethoprim and sulfamethoxazol: systematic review

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Introduction: Sulphonamides and trimethoprim are generally well-tolerated. However, they have sometimes been temporally associated with aseptic meningitis. To address its presentation and outcome, a literature search was performed.

Methods: A systematic review according to the Preferred Reporting Items for Systematic Reviews and Meta-Analysis (PRISMA) recommendations was performed. Articles reporting patients with features of acute aseptic meningitis following intake of trimethoprim, sulfamethoxazole, or sulfisoxazole were considered. A cerebrospinal fluid investigation in at least 1 episode was required for inclusion. Causality assessment was performed according to the World Health Organization-Uppsala Monitoring Centre (WHO-UMC) scale.

Results: Sixty articles, reporting on 74 patients experiencing a total of 155 episodes, were included. Fourty-five (61%) patients had at least one recurrence. Median age at first episode was 43 (interquartile range, IQR, 23-61) years, and 14 (19%) patients were children. Implication of the culprit drug was judged possible in 24 (32%), probable in 41 (55%) and certain in 9 (12%) cases. Symptoms presented within 48 (IQR 6-168) hours of intake at first episode, and within 1.3 (IQR 1-5) hours at recurrences ($p < 0.0001$). Most frequent presenting symptoms and signs were, in decreasing order of frequency, fever, headache, neck stiffness or other meningitis signs, abdominal symptoms, altered level of consciousness, nausea or vomiting, and ocular symptoms. Cerebrospinal fluid analysis revealed a predominantly neutrophilic (82%, IQR 65-94%) pleocytosis (180, IQR 38-507 106 cells/L), without low glucose or high proteins. There were no differences in culprit drug, number of episodes, causality assessment, presence and type of underlying immunological disease or biomarkers of altered immunity whilst comparing adults and children. Recovery took place within 2 (IQR 1-3) days after stopping the suspected agent. All but one patient completely recovered.

Conclusion: This systematic review suggests the existence of a causal relationship between trimethoprim, sulfamethoxazole or sulfisoxazole and the development of acute aseptic meningitis a few days thereafter. The reaction is generally mild and always self-limiting. Patients recover within one week upon discontinuation of the suspected agent.

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The lengthy path with various differential diagnoses in a patient with extensive splenomegaly and bicytoepenia leading to the rare diagnosis of visceral leishmaniosis

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We present the case of a 6-year-old girl who was referred to our hemato-oncological department for further evaluation of splenomegaly with bicytopenia. The patient has been experiencing progressive fatigue and asthenia for approximately 6 months, as well as leg pain and fever episodes. Suggesting a bone marrow disease, a bone marrow examination has been performed, showing no signs for leukemia or another primary bone marrow diseases.

Furthermore, a range of infectious diseases was screened: EBV, CMV, HAV, HBV, HCV, Toxoplasmosis, Brucellosis, Ricksettia and Bartonella. All of these were found to be negative.

Due to a mild hepatopathy (mild hepatomegaly and elevated liver enzymes) and increased immunoglobulins, further investigations were performed for autoimmune hepatitis and storage diseases, e.g. Gaucher`s disease, all of which were unremarkable.

Despite the absence of documented long-distance journeys in the preceding 18 months, an extensive serological diagnostic of leishmaniasis was conducted. Strikingly, a significant increase of anti leishmanial immunoglobulins showed evidence for leishmaniosis. Diagnosis was confirmed by PCR testing of the bone marrow.

Visceral leishmaniosis is a rare disease in our part of the world with only a limited number of cases reported from 15 European countries including Switzerland. It is caused by Leishmania donovani or Leishmania infantum, protozoa transmitted by sand flies. These protozoa typically inhabit tropical regions, but climate change has led to an increase in their numbers around the Mediterranean Sea. Our patient was in Liguria, Italy, 1.5 years ago, spending time on the beach. This was most likely the site of inoculation. The incubation period is usually 3-6 months. Symptoms are variable and often unspecific flu-like. Patients can also experience weight loss and lymphadenopathy. Oedematous organ swelling can lead to hepatosplenomegaly or, if affecting the bone marrow, to bleeding and infections. The diagnosis is confirmed by PCR testing of bone marrow. Visceral leishmaniosis is treated with systemic amphotericin B.

Our young patient responded quickly to treatment with improvement in general condition, normalisation of blood counts and disappearance of hepatosplenomegaly within a few weeks.

This case emphasizes the importance of considering unexpected diagnoses, even when the patient's history does not align perfectly.

P 70

Best picture: The crucial role of MRI on the diagnostic route to detect pyomyositis – a case report

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Background: Pyomyositis (PM) in children in non-tropical regions is rare and its diagnosis can be challenging. Caused by bacteraemia or as extension of an infection, PM challenges physicians with unspecific clinical and laboratory findings. PM is characterized by three clinical stages: (1) Inflammation and tenderness of infected muscles, (2) Muscular abscess developing in the suppurative stage with fever and pain, (3) Multifocal abscess formation associated with sepsis. Muscles of the thigh or pelvic girdle are mainly affected, and MRI (diffusion-weighted imaging and post-gadolinium enhancement) leads to the diagnosis.

Case report: An 11 year old boy without a history of recent infection presented with an antipyretic-sensitive fever for two days and pain reaching from his right groin to the tight and lower leg, aggravated while walking. Trauma or intense activity was denied. Physical examination revealed painful glutes on both sides, painful hip rotation and limping on the right.

Laboratory results: CRP 116 mg/l, ESR 26mm/h, WBC 5 Tsd/µl, Lymphocytes 0.7 Tsd/µl, CK 62 U/l. Based on normal hip X-ray and normal ultrasound (US) the boy was dismissed with suspected myalgia due to a common cold.

Two days later, he presented with analgetic-resistant pain in the same location, impaired range of motion and worsening of walking ability. CRP increased (230 mg/l). Blood culture was negative, and additional laboratory evaluation was uneventful (urine, C3, C4, ANA and ANCA). Suspecting PM, an MRI was performed and indicated isolated oedema in the right internal obturator muscle and the diagnosis of infect-associated PM was made. Intravenous antibiotic treatment with amoxicillin/clavulanic acid (40mg/kg bodyweight every 12h for a total of 10 days) was initiated and the boy could be dismissed after 5 days of hospitalisation.

Conclusion: Patients initially presenting with fever, myalgia, normal X-ray and normal US, showing persistent or worsening symptoms are challenging to diagnose and deep-muscle abscess should be kept in mind. In this case, an MRI-based diagnosis may lead to the diagnosis of pyomyositis. Even in early stages, the highly sensitive DWI-MRI demonstrates disease-extension and associated structures. CT rarely improves diagnostic yield.

P 71

Is there a shortage of pediatricians in primary care in Switzerland? A physician survey

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Introduction: Shortages of primary care pediatricians in Switzerland have been reported by media, parents, and physicians, with workforce estimates suggesting levels below recommended standards. Prior research indicates strong alignment between workforce data and perceived shortages. We surveyed pediatricians regarding their work setting, compared perceived pediatric primary care shortage across regions, and identified contributing factors.

Methods: We conducted an online survey among all board-certified members of the Swiss Society of Pediatrics (pädiatrie schweiz), excluding retired members. The questionnaire asked about medical training, work setting (workload, practice type), perceived shortage, contributing factors, acceptance of new patients in primary care practices, motivations for working in pediatric primary care, and number and duration of patients visits.

Results: Out of the 1970 members contacted, 1065 (54%) responded of whom 61% of worked in primary care practices, 28% in hospitals, 10% in both settings, and 2% were not clinically active. Most pediatricians worked part-time: 87% of primary care pediatricians and 65% of hospital pediatricians. This translated to a median of 35 hours per week (IQR: 26, 42) in practices and 40 hours per week (27, 54) in hospitals. Overall, 69% of pediatricians perceived a shortage of primary care pediatricians in their region, 19% perceived no shortage, and 12% were unsure. Perceived shortage was lowest in Ticino (16%) and the Lake Geneva region (47%) and highest in German-speaking regions (e.g. Eastern Switzerland: 90%, Central Switzerland: 85%). Among primary care pediatricians, 30% accepted new patients in their practice, 58% accepted only selected patients (e.g. siblings), and 12% did not accept new patients. Primary care pediatricians saw a median of 22 children per day (18, 26). Respondents suggested that the small number of physicians in Switzerland (69%), part-time work (65%), high administrative burdens (61%), and limited interest of young pediatricians for primary care (59%) contribute to the shortage.

Conclusion: Our study shows that two thirds of pediatricians in Switzerland perceive a shortage of primary care pediatricians, with large regional differences. German-speaking regions perceived the highest levels of shortage. An additional survey in

parents will broaden the understanding of primary healthcare for children in Switzerland.

P 73

The potential of APN in pediatric primary health care in Switzerland

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Background: Pediatricians in Switzerland are essential for primary care of children and adolescents. Due to the shortage of skilled professionals and other healthcare challenges, adjustments are needed. To ensure effective pediatric primary care, new models of healthcare delivery are necessary. The role of an APN in Swiss pediatric primary healthcare has not yet been implemented.

Objective: This study aimed to examine the organization and work content of Swiss primary care to derive recommendations for integrating an APN and to describe the process of introducing this role.

Method: Eight Swiss pediatric practices were examined using a qualitative multiple case design. Pediatricians in these practices were individually interviewed as part of a heterogeneous sample through semi-structured expert interviews. Inductive analysis was used to draw conclusions from specific observations obtained via cross-case analysis.

Results: The study involved six female and two male pediatricians (average age 46.5 years) with an average of 19 years of experience. Seven worked in group practices, three in rural areas, and five in urban settings. The study shows a predominantly positive attitude toward integrating the APN role, particularly to address the shortage of professionals in primary care, especially in rural regions. The APN is seen as a valuable resource for taking over certain medical tasks and improving care. A possible role profile includes triage, initial and follow-up treatment for minor acute illnesses, preventive screenings, nursing interventions, home visits, and case management for children with complex, chronic conditions. To establish effective primary pediatric health care the development of interprofessional commitment in such new health care delivery models are essential, this is a key factor to success. Hiring an APN is viewed as a valuable relief and enrichment for primary care, though regulations and financial challenges remain.

Conclusions: APNs can strengthen and ensure primary care. Their contribution demands a clear role definition. Successful implementation requires clear role profiles, comprehensive education on APN competencies, as well as funded pilot projects and legal clarifications to develop evidence-based care models.

P 74

Advanced Practice Nurses in Pediatric Primary Care: First Steps in German-Speaking Switzerland

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Background: The benefits of and the demand for Advanced Practice roles in pediatric nursing have been well-documented both nationally and internationally. However, in pediatric primary care in Switzerland, the role of the Advanced Practice Nurse (APN) remains underdeveloped. APN have the potential to make a significant contribution to improving care. Findings of a recent qualitative study conducted in eight Swiss pediatric practices in German-speaking Switzerland indicate a positive attitude among pediatricians towards the integration of APN.

Aim: To present preliminary insights into the practical implementation of two APN roles within a pediatric practice and a general practice serving children and adolescents.

Method: Description of initial experiences, opportunities and challenges of the two APN roles in pediatric primary care.

Results: In both the pediatric and general practices, the APN are responsible for triaging and managing cases of minor emergencies. Additionally, they contribute to the interprofessional management of children with school-related difficulties and ADHD, assuming case management responsibilities. Beyond that, the APN provides advice on skin and sleep issues, constipation, enuresis or obesity and common problems for which there is a lack of capacity. In the general practice setting, the APN conducts preventive health screenings and school health examinations. In both settings, the APN receive regular supervision and exchange sessions by the pediatrician or general practitioner, which are essential for fostering mutual trust. Several factors, such as the clarification of the role profile within the practice team, the lack of reimbursement mechanisms, and the uncertain legal framework, were identified as key challenges to the implementation of APN.

Conclusions: APN when incorporated into a structured task-sharing framework with clearly defined role specifications within the healthcare team, offer a viable strategy for ensuring the provision of high-quality pediatric care. Furthermore, they represent a promising strategy to mitigate the growing shortage of qualified healthcare professionals in primary care settings. Further research evaluating the benefits, costs, and the satisfaction levels of both families and healthcare professionals is necessary to substantiate this model. Additionally, the establishment of a specific tariff for APN and the clarification of regulatory frameworks for education, training, and registration as APN are crucial.

P 75

Rare disease: integrative and interprofessional approach: collaboration between advance practice nurse and medical pain and palliative management

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Rare paediatric diseases are often chronic, complex, and progressive, requiring specialised and personalised care approaches, with proactive and early integration of palliative care. Advanced practice nurses (APNs) play a central role in improving care for these patients by leveraging their clinical expertise, coordinating interdisciplinary teams, and integrating palliative care into the overall management plan. This presentation aims to highlight the essential of integrative and interprofessional mangement in the care of patients with rare diseases, based on a case study conducted in a paediatric neuromuscular disease centre in Ticino.

This case study illustrates how APNs optimise continuity of care, support interprofessional collaboration, and provide holistic support to patients and their families. Palliative care, in particular, is presented as a cornerstone for managing pain and complex symptoms, preventing complications, offering psychosocial and emotional support, and improving quality of life from the time of diagnosis. Their role extends beyond the hospital, linking social services, patient associations, schools and local resources to provide holistic support. Through concrete example, we demonstrate the impact of APN interventions, pain

and palliative care, which not only alleviate physical suffering but also address the psychosocial and spiritual needs of patients and their families throughout the disease trajectory.

Looking to the future, strengthening the integration of APNs into specialised teams is crucial to ensuring comprehensive follow-up, the early and tailored implementation of palliative care, and sustainable support for families. Expanding their presence in centres dedicated to rare diseases in Switzerland is essential to better address the complex needs of these patients and ensure equitable access to palliative care.

Through this case report, we would like to highlight the effectiveness and importance of integrated and inter-professional care in paediatric rare diseases from diagnosis. The expertise, combined with integrative ability to coordinate care, educate patients, and deliver comprehensive palliative support, enhances quality of life and addresses the challenges posed by these complex and demanding conditions.

P 76

Doing well with little means: an efficient pediatrician-nurse paired consultation in an asylum-seeking shelter in the Canton of Vaud

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Introduction: The growing number of vulnerable asylum-seeking children in Switzerland requires better access to healthcare. In 2024, a new consultation service was set up within the Temporary Accommodation Center (TAC) of the Etablissement Vaudois d'Accueil des Migrants for children seeking asylum, aligning with World Health Organization's recommendations.

Objectives: Using socio-demographic data and access to care data, the aim is to demonstrate the added value of this consultation for children seeking asylum in the canton of Vaud.

Method: A descriptive analysis of quantitative data on the number of consultations, their nationality and age. All migrant children were included, regardless of origin and residence permit. Consultations were held once a week and children were seen by a pediatrician-nursing pair and an interpreter. Data collection was anonymous.

Results: Of the 827 children housed in the TAC in 2024. Consultations occurred once a week for a total of 46.5 days and we carried out 505 consultations, 11 patients per day on average. 420 children (51%) attended general pediatric and/or urgent care consultation to assess each child's overall health, including psychosocial needs, and identifying any chronic conditions requiring specialist care. 78% for 0-4-year-olds, 60% for 5-8-year-olds, 35% for 9-12-year-olds, 39% for 13-15-year-olds, and 17% for 16-17-year-olds. The children consulted were from 24 different countries of origin, including 3 prominent ones: Ukraine (67%), Afghanistan (12%) and Turkey (8%).

Discussion: This analysis offers a valuable perspective on access to healthcare for migrant children in Swiss shelters. High demand for primary care was evident, with over half of the children requiring consultations. With just one day of presence per week, the on-site medical and nursing care model has improved access, reaching the majority of children aged 0 to 4 and two-thirds of children aged 5 to 8. One of the significant obstacles to access to care was the difficulty of accessing interpreters of all different languages.

Conclusion: The pediatrician-nurse paired model with interpreters significantly facilitates care access for vulnerable pediatric population. Data collected can inform targeted interventions and further research on adherence and health outcomes.

P 77

A novel way of safely discharging patients: criteria-led discharge - a study protocol.

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Background: In times of high patient loads and limited resources, paediatric hospitals are challenged to restructure their patient pathways. Internationally, criteria-led discharge (CLD) is promoted, aiming to standardize safe discharges, reduce delays and promote interprofessional responsibility. For CLD predefined discharge criteria serve to inform about discharge readiness and when fulfilled allow immediate discharge by nursing staff. At the Children's Hospital of Central Switzerland (KidZ) CLD was introduced in December 2024. Specifically, CLD can be prescribed to children hospitalized with either asthma exacerbation/obstructive bronchitis, bronchiolitis or uncomplicated pneumonia after assessment on the ward rounds and verbal informed consent of caregivers and patients. A disease specific checklist then serves to track their recovery and guide the discharge process. The pioneering of paediatric CLD in Switzerland is evaluated in a quality control study for its impact on patients, staff and the discharge process.

Methods: Patient outcomes are compared using retrospective data collected from the electronic health records and through a survey. The primary outcome is safety, measured as within 14-day-readmission and complication rates. These are compared in CLD versus non-CLD patients and in pre- and post-implementation cohorts. The secondary outcomes include efficacy and satisfaction. Efficacy is measured using length of stay and discharge delays pre- and post-CLD-implementation. Staff satisfaction is evaluated using a survey at baseline and 3 and 6 months after CLD initiation. For patients and parents, satisfaction is measured by continuously collected questionnaires shortly after discharge, both for CLD and non-CLD patients.

Discussion: CLD implementation at the KidZ was driven by its multiple positive effects proposed in literature. Those are to be verified by this quality control study. Successful and timely implementation of CLD, enabled through rapid recognition of discharge readiness, aims to standardize the discharge process and minimize discharge delays. Fast discharges free resources, which can be redirected towards more complex patients. Transparency regarding discharge criteria may increase health literacy and standardization leads to more evidence-based discharges. Among staff, it is expected that increased interprofessional responsibility between nurses and doctors and optimized workflows will improve satisfaction.

P 78

Improving early detection and support of preschool children with developmental delay: Combining the benefits of two different cantonal systems of care

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The 2007 Intercantonal Agreement on Collaboration in Special Education recommends harmonizing the care for children with special educational needs and implementing standardized tools

across cantons. In this context, our research compares two cantonal systems – Zurich and Geneva – focusing on data collection and the coordination of specialized services for preschool children at risk or with developmental delay requiring early intervention, with the goal of identifying areas for improvement. This poster will present the findings of this study collected from interviews with known healthcare and education institutions, as well as from available data and literature, revealing notable differences between Zurich and Geneva, particularly in the systematization of data and individualized support. These insights foster discussions on best practices that can be modeled in other cantons or nationwide. By optimizing the identification of children with developmental delays and addressing their specific needs, this research seeks to contribute to a more cohesive and effective approach to early detection and support of these children across Switzerland.

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Parental Perception of Treatment Options for Mucopolysaccharidosis: Bridging the Gap for Personalised Medicine

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Background: Mucopolysaccharidoses (MPS) are a group of autosomal-recessively inherited lysosomal storage disorders with substantial unmet medical needs affecting both patients and caregivers. Current therapeutic options are limited, and little is known about parental perceptions of investigational therapies.

Methods: We conducted a survey using the discrete choice experiment (DCE) method to assess parental preferences regarding approved and investigational therapies for both neuronopathic and non-neuronopathic MPS. The survey was distributed via patient organisations in Germany, Austria, and Switzerland. Scenarios included enzyme replacement therapy (ERT), haematopoietic stem cell transplantation (HSCT), gene therapy (GT), and individualised treatment trials (ITTs) utilizing repurposed drugs.

Results: Parents expressed the highest preference for ITTs with repurposed treatments (neuronopathic: 82%, 14/17; non-neuronopathic: 94%, 16/17), followed by ERT (88% in both groups, 15/17), HSCT (neuronopathic: 70%, 12/17; non-neuronopathic: 76%, 13/17). Gene therapy (GT), while seen as a promising investigational option, received lower preference scores (neuronopathic: 58%, 10/17; non-neuronopathic: 53%, 9/17). Positive attitudes toward ERT, HSCT, and ITTs strongly correlated with willingness to pursue these therapies (>80% in both patient scenarios), while favourable views of GT were less predictive (~50%). Mild side effects, including infections, injection site reactions, and treatment-related hospitalisations, were considered acceptable by most respondents. Patient organisations were identified as the primary source of information.

Conclusions: This study provides a novel framework for evaluating parental preferences for MPS treatments, emphasizing the importance of caregiver-centered approaches in risk–benefit modelling. The comparatively lower preference for GT likely reflects its status as an emerging therapy without regulatory approval for MPS at the time of the study. Limited access to information and a lack of long-term data on efficacy and safety may contribute to parental hesitancy. The findings highlight the need for tailored communication strategies, particularly for novel and emerging investigational therapies such as GT. An international follow-up project is underway to expand the dataset and explore parental perspectives across diverse settings.

P 80

How to Communicate Smarter Medicine by Choosing Wisely Your Words

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The pediatric group of Choosing Wisely Switzerland has published two Top 5 Smarter Medicine lists. The first in 2021 on unnecessary treatments and the second in 2024 on unnecessary tests in pediatrics.

In daily practice, we find it difficult to "sell" these evidence-based recommendations to parents, mainly due to historical and cultural expectations. Simply "regurgitating" scientific information is not enough.

We want to show how the use of argumentative strategies, like motivational interviewing techniques developed by

William R. Miller and Stephen Rollnick, among others, can help clinicians. We will use the example of recommending against the use of cough syrups for children's coughs.

We will begin with open-ended questions about their opinions about cough syrups, then practice reflective listening and provide scientific information with permission. We will affirm the strengths by acknowledging the parents' efforts and concerns, and when they express resistance, we will avoid arguing and instead work with them. We will encourage their belief in their ability to manage their child's symptoms and ultimately work together to develop a plan that is consistent with evidence-based recommendations.

Our final goal is to increase the acceptance of these 10 recommendations, as some of them go against long-held popular beliefs that are reinforced by manufacturer advertising and social media.

P 81

Not every effusion needs a puncture: Panner's disease, the Perthes of the elbow!

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Introduction: Panner's disease is a rare osteonecrosis of the capitellum of the humerus, like Perthes disease of the hip. In an 8-year-old boy with persistent elbow pain, restricted range of motion, and inconspicuous laboratory findings, the diagnosis was made through MRI imaging.

Case report: An 8-year-old active boy presented with two days of atraumatic elbow pain, mild swelling, warmth, and transient fever. Blood tests, including inflammatory markers and Borrelia serology, were unremarkable. Initial clinical suspicion included septic arthritis or Lyme arthritis. Examination revealed a swollen right elbow with limited flexion and extension (100-10-0) but no systemic illness or signs of infection. Imaging showed joint effusion with synovial thickening on ultrasound and a double contour sign on X-ray, suggesting intra-articular pathology. Joint aspiration was deferred due to the absence of infection markers, and the arm was immobilized with ibuprofen prescribed.

At follow-up 2 weeks after symptom onset MRI showed osteonecrotic changes in the capitellum. A multidisciplinary review concluded Panner's disease. Treatment focused on rest, avoiding high-impact stress, and physiotherapy without forced exercises. At six months, the patient was pain-free but showed

slight progression of range-of-motion limitations (100-20-0), confirmed by MRI. Orthopedic specialists recommended ongoing conservative management with regular follow-up.

Discussion: Panner's disease, first described in 1927 by H.J. Panner, is an osteochondrosis affecting mostly boys under 10 years, presenting with atraumatic elbow pain, swelling, and restricted motion. The pathogenesis is unclear but likely involves valgus stress, reduced blood supply, and ischemia. Diagnosis is based on characteristic radiographic and MRI findings.

The disease progresses through four stages: subchondral sclerosis, fragmentation, osteolysis, and repair. While conservative therapy (rest and symptomatic treatment) typically ensures recovery, prolonged courses lasting 1–3 years may occur. Differential diagnosis includes osteochondritis dissecans (OCD), which affects older children and involves intra-articular loose bodies.

Early recognition and appropriate management are key to preventing unnecessary interventions and ensuring complete recovery.

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Deep Venous Thrombosis and Consecutive Pulmonary Embolism as the Initial Presentation of Systemic Lupus Erythematosus in a 17-Year-Old Female

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Introduction: Systemic lupus erythematosus (SLE) is a rare and often overlooked diagnosis in the pediatric emergency department (ED). We present the case of a 17-year-old female whose initial manifestation of SLE was a significant deep venous thrombosis (DVT) and subsequent complications.

Case presentation: A 17-year-old female presented to the ED with acute, severe pain in the right leg, accompanied by discoloration. Her medical history was notable for rosacea and the use of oral contraceptives for the past five months. A family history of Factor V Leiden mutation and activated protein C (APC) resistance was reported, though the patient's inheritance of these conditions was not confirmed. On examination, her vital signs were stable. The right leg was markedly swollen, exhibited a red-violet discoloration and had strong palpable pulses. Clinical signs suggestive of DVT were positive. The remainder of the physical examination was unremarkable. Laboratory findings showed elevated D-dimer levels, and duplex ultrasonography confirmed thrombosis of the right external iliac vein. The patient was initiated on anticoagulation therapy and referred to the University Hospital of Zurich, where she underwent catheter-directed thrombectomy followed by oral anticoagulation with a factor Xa inhibitor. Two days later, the patient re-presented to the ED with severe chest pain, dyspnea, and tachypnea. Computed tomography revealed a paracentral pulmonary embolism. She was again transferred to the University Hospital of Zurich for further management. Subsequent laboratory evaluations identified a positive lupus anticoagulant. Further investigations confirmed a diagnosis of SLE, supported by the presence of anti-dsDNA antibodies. Genetic testing ruled out inherited thrombophilias. Her prior diagnosis of rosacea was retrospectively considered a potential cutaneous manifestation of SLE. Anticoagulation therapy was transitioned to phenprocoumon for long-term management.

Conclusion: This case initially presented as a classic DVT with apparent risk factors. However, further investigation revealed the underlying diagnosis of SLE. Retrospective analysis highlighted clinical features consistent with SLE, emphasizing the need for a broad differential diagnosis in the ED. While diagnosing rare conditions like SLE may fall outside the typical purview of emergency medicine, this case illustrates the importance of thorough evaluation and appropriate follow-up for atypical presentations.

P 83

Orbital floor fractures in children: why you should not blow your nose?

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Introduction: Orbital floor fractures are a significant concern in pediatric trauma, particularly in the context of high-impact sports. These injuries can lead to complex clinical presentations and require prompt management and diagnosis to prevent long-term complications.

Clinical case: A 14-year-old patient, with no notable medical or surgical history, was admitted after a facial impact against a tree while skiing. The patient reported ocular and periocular pain. Clinical examination revealed an important periorbital hematoma and pain upon palpation of the maxillary bone. No facial hypoesthesia was noted. The patient complained about diplopia with intense pain when moving the eye in all directions, but no limitation of oculomotricity. Visual acuity was preserved. A facial computed tomography demonstrated a right orbital floor fracture with fat incarceration and important periocular emphysema, due to nose blowing before imaging. The patient underwent orbital reconstruction using a resorbable plate by a maxillofacial surgeon. Postoperative course was uneventful, except de persistency of diplopia.

Conclusion: This clinical case highlights the diagnostic challenges associated with orbital fractures in pediatric patients, as the presentation may not consistently include all characteristic clinical signs. Computed tomography is discussed in pediatric population because of radioprotection. Prompt diagnosis is essential to prevent long-term functional and aesthetic complications. In pediatric patients, particularly due to trapdoor fractures, it is essential to avoid muscle ischemia and permanent impairment of ocular motility. The unique anatomy of the pediatric orbit, coupled with the potential for subtle clinical presentations, necessitates a high index of suspicion and timely intervention to ensure optimal outcomes.

P 84

What do siblings of children with disabilities need in daily life situations? Implications from a scoping review on childhood-onset disability research

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Introduction: Siblings of children with childhood-onset disabilities often face daily challenges such as disruptions of family routines and a lack of support in attending hobbies or doing homework. Some report major difficulties in their mental health,

quality of life, and psychosocial adjustment. Despite this, comprehensive research on their participation patterns, daily needs, and potential restrictions remains limited. Understanding these patterns and influencing factors is essential for enhancing family-centered care and developing effective support strategies. To address this gap, a scoping review of research on siblings' participation in everyday activities was conducted.

Methods: The review was conducted following the Joanna Briggs Institute methodology. A systematic search of MEDLINE, CINAHL, AMED, PsycINFO, and ERIC identified peer-reviewed studies published between 2001 and 2024. Inclusion criteria focused on siblings of children with childhood-onset disabilities or chronic conditions, exploring their participation across home, school, and community settings. Findings were collaboratively translated into a family-friendly factsheet with parents of children with disabilities and typically developing children. The factsheet will be piloted, refined, and provided in English, German, French, and Italian.

Results: The review included 62 studies: 45 qualitative (73%), seven quantitative (11%), seven reviews (11%), and three meta-studies (5%), covering a range of chronic conditions, with autism being the most frequently studied (22 studies). Key participation themes included family life, school involvement, leisure activities, peer relationships, information-seeking, and meaning-making. Factors such as advocacy, identity, age, gender, culture, and socioeconomic status intersected with siblings' participation experiences. The collaboratively developed factsheet highlights siblings' needs and was designed to facilitate family conversations focusing on the siblings.

Conclusion: This scoping review provides critical insights into the participation of siblings of children with disabilities, identifying key themes and influencing factors. The collaborative creation of a family-friendly factsheet demonstrates the value of translating research findings into actionable tools that support families. Its potential for distribution and practical use will be discussed. The findings emphasize the importance of addressing siblings' needs to strengthen family-centered care.

P 85

Over-the-counter poison - consequences of severe intoxication

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A 4-month-old boy presented to the emergency department in poor general condition and with signs of dehydration. He showed failure to thrive with a weight loss of 500 g in 10 days, diarrhea and vomiting. He had also had a fever for 2 days. The boy was born at term, otherwise healthy and unvaccinated. He was exclusively breastfed. Both parents were healthy, originally came from Albania and were not related.

The clinical examination revealed the typical signs of dehydration without any other abnormalities in the internal or neurological examination findings. In the first laboratory check, hypercalcemia with an ionized calcium of 3.12 mmol/l (N:1.15-1.29 mmol/l) was noted. In further diagnostics, the calcium/creatinine ratio in the urine was significantly elevated at 5.57 (N: <2.2). The 25-OH vitamin D3 was not measurably elevated at >240 mcg/l, nor was the 1.25-Di OH vitamin D at >1440 pmol/l. The PTH of 8.04 pg/ml (N:17.3-71.1 pg/ml) was suppressed. An ultrasound of the kidneys showed no nephrocalcinosis but clearly enlarged and hyperechogenic kidneys.

When asked again, the mother stated that she had only started vitamin D prophylaxis after 2 months postpartum with a self-purchased product from the Internet, the so-called "sun vitamin", which contained 5000 U/drops. This was administered over 8-10 weeks, whereby the mother administered generously and sometimes more than 1 drop per day. In total, the child received approx. 1.5 million IU during this time, which had led to a severe vitamin D intoxication.

The hypercalcemia was treated intensively with hyperhydration, administration of furosemide, cortisone and calcitonin (0.7 U/kg). This led to an initial stabilization with a secondary increase in calcium. Bisphosphonate (zoledronate 0.025 mg/kg) was therefore administered. This resulted in a prompt and long-term normocalcemia. Even after 4 months, the lipophilic vitamin D3 was elevated at >100 mcg/l (N: >30 mcg/l). Nephrocalcinosis has not yet occurred. As is often the case, the medical history led to the cause of the disease. In this case, precise inquiry revealed hypercalcemia caused by vitamin D intoxication. Normocalcemia could only be achieved with intensive therapy and the use of bisphosphonates. This case shows impressively how important it is to educate parents about vitamin D prophylaxis, as unfortunately there is no way to regulate over-the-counter supplements.

P 86

Consecutive clues to the diagnosis of a rare disease: H syndrome

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Background: H syndrome is a rare autosomal recessive genetic disorder caused by pathogenic variants in SLC29A3 gene, leading to abnormal histiocytic proliferation and dysregulated inflammatory response. Clinical symptoms are variable and may include recurrent fever, hyperpigmentation, hypertrichosis, hepatosplenomegaly, cardiac anomalies, hearing loss, hypogonadism, fixed flexion contractures of the toe or interphalangeal joints, and diabetes. We present an adolescent with diagnosis of H syndrome after a diagnostic delay of 15 years.

Case presentation: At the age of 4 the girl developed an antibody-negative diabetes. During school-age, a slowly progressive non-inflammatory camptodactyly of fingers and toes, hallux valgus, self-limited orbital tumor and mild hypertrichosis were noted. At the age of 16, she presented recurrent fever episodes, myalgia, urticarial exanthema, plantar panniculitis, and fatigue. Similar episodes were also described in early childhood. A sister had a history of diabetes, camptodactyly and bilateral orbital tumor. Laboratory tests showed increased erythrocyte sedimentation rate (ESR) up to 50 mm/h with increased CRP and serum amyloid A, hypergammaglobinemia, and increased interferon (IFN) signature. An organ screening revealed mild hepatomegaly and mild cardiac septal hypertrophy. First genetic testing for common monogenic fever syndromes was negative. Due to continued intermittent systemic inflammation the girl was treated with prednisone and methotrexate with discontinuation due to intolerance and anakinra with discontinuation due to ineffectiveness. Based on clinical signs, symptoms and laboratory findings, a therapy trial with the janus kinase inhibitor(JAKI) ruxolitinib was started, which showed rapid clinical improvement with resolution of fever and fatigue. Skin lesions improved. Laboratory showed normalization of IFN signature

and ESR. Trio-exome analysis revealed a compound heterozygous pathogenic variant of SLC29A3, compatible with the diagnosis of H syndrome.

Conclusion: H syndrome is an extremely rare disease with heterogenous presentation. In patients with consecutive multi-organ involvement over a prolonged time period, a complete and interdisciplinary re-evaluation, ideally in a medical specialised facility focused on rare clinical presentations, is essential in order to diagnose rare diseases. JAKI may represent a new treatment option for H syndrome.

P 87

Constipation, Meteorism, Positive Celiac Antibodies & Positive Family History for Celiac Disease: and the Diagnosis is ... yet something else (and potentially life-threatening!)

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Background: Gastrointestinal (GI) symptoms such as meteorism and constipation are non-specific and common in childhood. They usually have a harmless and/or self-limiting underlying cause. However, they can be a hallmark of a serious underlying disease.

Case study: 7-year-old girl referred to gastroenterology outpatient clinic for evaluation of possible celiac disease (CD). The general practitioner reported chronic obstipation, meteorism, with positive family history for CD and isolated slightly elevated Transglutaminase IgA (13kU/l; ref. 7-10kU/l).

Clinical findings at our clinic before endoscopy: Length 114.7cm -1.94SDS, weight 27kg +0.54SDS, BMI 20kg/m2 +1.93SDS. RR 79/59mmHg, HR 71/min with otherwise inconspicuous problem-oriented examination. With macroscopically normal endoscopy, extended lab evaluation on the same day revealed severe hypothyroidism (TSH >1000mU/l (0.6-4.9mU/l), fT4 0.65pmol/l (12.5-21.5pmol/l), fT3 1.18pmol/l (3.9-8.0pmol/l)) as the patients underlying disease. The patient was hospitalized. Detailed physical exam revealed cold extremities, pretibial edema, puffy face, enlarged tongue, lanugo hair, decreased muscle tone and reflexes. Bone age was significantly delayed. Ultrasound showed a hyperperfused yet hypoplastic thyroid, all fitting with the diagnosis. With two elevated antithyroid antibodies (TPO-AB 362 IU/ml (<20) and Tg-AB 312IU/ml (<46)) autoimmunity is the obvious cause for thyroid failure in our patient. After confirming adequate adrenal function, low-dose thyroid replacement therapy was established and gradually adjusted (lab-controlled). Within days, the girl's clinic improved dramatically and, remarkably, long before a euthyroid status was documented in the lab.

Summary: Initial presentation suggestive for celiac disease set the diagnostic course wrong and delayed the making of the diagnosis in our patient to an extent that could have been potentially life-threatening.

Conclusion: Even harmless symptoms require careful medical history and physical examination. This is crucial to refine the differential diagnosis in children and help to avoid unnecessary procedures and delay in diagnosis. In our patient on hearing the hoofbeats the many celiac horses made it difficult to think of the hypothyroid zebra in the herd.

P 88

Typical sympathomimetic toxidrome in an 11-month old infant after amphetamin intoxication

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Introduction: The number of prescribed Amphetamin-derived medication to treat attention deficit hyperactivity disorder (ADHD) in children is rising in Switzerland. Case reports concerning intoxications are rare. This case highlights the clinical manifestation and diagnostic challenge associated with accidental ingestion of Lisdexamfetamin (Elvanse ®) in an infant.

Case presentation: A previously healthy 11-month-old infant was admitted to the hospital with a two-day history of fever and cough, followed by a four-hour history of vomiting and new-onset of stereotypical movements. On examination, the infant was awake and alert but demonstrated unrelenting hand clenching and unclenching, persistent head nodding that later progressed to shaking, and constant leg movements. Additionally, she was crying out, almost every second, alongside an intense sucking urge and repeated episodes of vomiting. Vital signs showed her tachycardic and hypertensive. Toxicology testing revealed significantly elevated amphetamine levels in the urine. Further investigation suggested that the amphetamine exposure likely resulted from the unintentional ingestion of a sibling's Lisdexamfetamine, with one pill confirmed missing at home. Additionally, the infant was diagnosed with a febrile urinary tract infection, further complicating the clinical scenario. For urine catheterization, sedation with Diazepam was necessary. Interestingly, after sedation, the infant's symptoms improved briefly, including a reduction in the intensity of the rhythmic movements and crying. About 14 hours after onset of symptoms and the administration of overall 2x5mg Diazepam suppositories, they resolved completely. Child protection service was consulted and the family was counseled about safe storage of medication.

Conclusion: Accidental intoxication in infants may occur without caregiver awareness and is often difficult to diagnose based solely on clinical presentation. This case emphasizes the importance of urine toxicology screening in infants presenting with atypical or unexplained history or symptoms. It also underscores the need for secure medication storage in households with young children to prevent accidental ingestion.

P 89

Delayed bleeding in pediatric traumatic splenic injury

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Introduction: Management protocols for traumatic splenic injury changed over the last decade to much more liberal strategies regarding bedrest and blood transfusion management also because complications such as delayed splenic bleeding is rare.

Case report: We present a case of a 9-year-old boy transferred from a peripheral hospital after a mountain bike accident with traumatic splenic injury diagnosed by ultrasound. The patient presented hemodynamically stable. CT scan showed an isolated AAST Grade IV splenic injury with no signs of active hemorrhage. Non-operative management with ICU observation and transfer to the ward after 24h and bedrest for 72h was initiated. After mobilization he was complaining about increasing abdominal pain. Concomitant he became more tachycardic with initially stable blood pressure and hemoglobin. An abdominal ultrasound was repeated and showed a slight increase in free fluid. Further rapid deterioration to symptomatic shock with tachycardia, hypotension, decreased peripheral perfusion and

neurological impairment. Treatment of suspected hemorrhagic shock with initially fluid bolus of Ringer's followed by transfusion of PRBC's with stabilization of vital signs. Tranexamic acid was administered. A drop of hemoglobin to 69g/l (90g/l) during the secondary bleeding was seen. Repeated CT scan showed a progression of bleeding with suspected ongoing hemorrhage. Therapeutic angioembolization with successful coiling of a peripheral arterial branch of the spleen with afterwards no more detected perfusion in the affected area. Except a pleural effusion uneventful postinterventional course with normal follow up imaging and unremarkable mobilization 48 hours after intervention. The patient could be discharged home one week after embolization.

Discussion: The incidence of complications after traumatic splenic injuries are low. Delayed splenic bleeding is rare, reported with a less than 0.2% frequency. Nevertheless, it can lead to hemorrhagic shock and needs to be managed in a timely manner. The recent APSA guideline suggests therapeutic angioembolization as an alternative to an operative management in patients with ongoing hemorrhage, if hemodynamically stable enough to travel to the interventional suite. Our patient fulfilled the suggested criteria and could be coiled successfully.

Conclusion: Complications are rare but can be severe, therefore we suggest close monitoring after mobilisation especially in higher grade splenic injuries.

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Sodium glucose type 2 transporter inhibitors in Paediatrics: a systematic review

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Introduction: In adults, sodium glucose type 2 transporter inhibitors have revolutionized the treatment of type 2 diabetes mellitus, chronic kidney disease and heart failure. We aimed to review available literature on compassionate use, clinical pharmacology, efficacy, and safety of dapagliflozin and empagliflozin in children.

Methods: We performed a systematic review of clinical trials, case reports or observational studies in Medline, Excerpta Medica and Web of Science databases. For randomized-controlled trials, if possible, we implemented a meta-analysis.

Results: Thirty-five articles (nine case reports, 10 case series, two surveys, one prospective non-controlled trial, four controlled randomized trials, six pharmacokinetic studies, and three pharmacovigilance studies) were retained. 415 children, who received either dapagliflozin or empagliflozin, were reported: 189 diabetic patients (mean age 14.7±2.9 years), 32 children with glycogen storage disease type Ib (GSD Ib), G6PC3-deficiency or severe congenital neutropenia type 4 (8.5±5.1 years), 47 children with kidney disease or heart failure (11.2±6.1 years), 84 patients in pharmacokinetic studies (15.1±2.3 years) and 63 patients in toxicological series. In type 2 diabetes mellitus, the efficacy of dapagliflozin and empagliflozin was assessed by HbA1c reduction in two randomized trials including 177 adolescents, with a mean HbA1c difference of -0.82% (95%-CI -1.34% to -0.29%) as compared to placebo (I2 = 0%). Dose ranged between 5-20 (mean 11.4±3.7) mg for dapagliflozin and between 5-25 (mean 15.4±7.4) mg once daily for empagliflozin. In children with GSD Ib, empagliflozin 0.1-1.3 mg/kg/d improved neutropenia, infections, and gastrointestinal health. Dapagliflozin (mean dose 6.9±5.2 mg once daily) was well-tolerated in children with chronic kidney disease and heart failure. Side effects were globally mild, the most frequent being hypoglycaemia in children with GSD Ib (33% of patients) or type 2 diabetes mellitus (14% of patients) on concomitant hypoglycaemic drugs. Diabetic ketoacidosis is rare in children.

Conclusions: Available evidence suggests that dapagliflozin and empagliflozin are efficacious in adolescents with type 2 diabetes mellitus, and that they are well tolerated in children across indications. A clinical pharmacology rationale currently exists only for adolescents with diabetes mellitus.

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Severe acute kidney injury due to malaria in an adolescent patient

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Summary: Complications of severe malaria in children mainly include anemia, acidosis, hypoglycemia and cerebral malaria, whereas renal involvement is considered rare in Europe. To our best knowledge, this is the first case description in European literature of an adolescent with malaria who presented severe acute kidney injury (AKI) and also nephrotic range proteinuria.

Case report: A 15-year-old adolescent developed symptoms of fever, headache, body aches, vomiting, and diarrhea after visiting relatives in the Ivory Coast – recommended chemoprophylaxis was not fully implemented during travel. Laboratory tests confirmed malaria (P. falciparum) and revealed AKI (min. eGFR 12 ml/min./1.73m2) with nephrotic range proteinuria (max. P/C 685 g/mol). Treatment with intravenous artesunate was started immediately, along with a course of peroral acetaminophen for renal protection. Despite severe AKI, criteria to start renal replacement therapy were not fulfilled and renal function and proteinuria improved for the first time after 6 days. Intravenous therapy with artesunate could be switched to peroral arthemeter-lumefantrine and the patient discharged at home with outpatient follow-up. At 4 months, all clinical (blood pressure) and laboratory parameters (urinalysis, blood-tests) had normalized with eGFR 102 ml/min/1.73m2.

Discussion: Malaria is caused by various plasmodia, with P. falciparum being the main pathogen causing severe forms of the disease. Pediatric patients often develop severe anemia or cerebral malaria. In contrast with African studies, renal involvement in children with malaria has rarely been described in European literature. AKI in African children with severe malaria is associated with increased mortality and chronic renal failure. Renal involvement in malaria usually manifests as acute tubular necrosis, glomerulonephritis or interstitial nephritis. In addition to severe AKI, our patient also presented nephrotic range proteinuria with hypalbuminemia. The criteria for nephrotic syndrome were ultimately met with mild edema during the course of the disease.

Conclusion: The case demonstrates that severe kidney involvement can also occur in European children and adolescents with malaria. Therefore, it is important to test for kidney involvement in all children with severe malaria.

Disclosure: Full case report was accepted for publication in https://www.omnimedonline.de/paed/ (2025)

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Benefits of Ambulatory Blood Pressure Monitoring over Office Blood Pressure in Children and Adolescents with Severe Obesity: Findings from The Bern Obesity in Childhood and Adolescence Biorepository (BOCAB)

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Background: Prevalence of hypertension (HTN) among children and adolescents with severe obesity is up to five times higher compared to their normal-weight counterparts. HTN in youth is a strong predictor of cardiovascular (CV) morbidity and premature mortality already in young adulthood. Ambulatory blood pressure monitoring (ABPM) has proven superior to office blood pressure (oBP) for diagnosis and CV risk prediction in adults. Growing evidence supports its use in the paediatric population. However, data on children and adolescents with severe obesity in this context remain sparse.

Methods: Anthropometric data (percent of the 95th body mass index percentile for age and sex according to CDC [%BMIp95], waist circumference-to-height ratio [WHtR], percent of body fat [%BF] and muscle mass [%MM]) and oBP and 24-hour ABPM results (both graded according to ESH guidelines) and their outcomes (normal BP, ambulatory HTN, white coat HTN [WCH] and masked HTN [MH], non-dipping) were drawn from the Bern Obesity in Childhood and Adolescence Biorepository. Descriptive statistics were used for baseline characteristics and prevalence of oBP- and ABPM-related subgroups.

Results: A total of 352 individuals, 52.6% males, had a mean (SD) age of 12.2 (2.6) years and a %BMIp95 of 123.5 (17.8). WHtR (n = 318) was 0.59 (0.07), and bioimpedance-derived %BF and %MM (n = 242) was 41.9% (6.4) and 31.7% (3.7), respectively. Based on oBP measurements (n = 303), 83% had normal BP, 8% had Grade 1 HTN, 9% had Grade 2 HTN. Based on ABPM (n = 352), 15% had ambulatory HTN and 44% were non-dippers. In participants with both oBP and ABPM available (n = 303), abnormal oBP was revealed as WCH in 71%, normal oBP overturned in 11% of cases. ABPM disproved 18% of all oBP findings. Overall, when also accounting for the diagnosis of non-dipping patterns, ABPM added diagnostic value in 54% of cases.

Conclusion: Our results reveal clear benefits of applying ABPM in children and adolescents with severe obesity for discriminating BP subtypes and risk stratification. Identifying ambulatory HTN as well as MH allows early treatment, which increases the chances to revert to normal BP, while exposing WCH prevents overtreatment.

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Is nephrotic-range proteinuria in an adolescent directly nephrotic syndrome?

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Background: Idiopathic nephrotic syndrome is a familiar pediatric disease in the age range 1-12 years. It is defined by nephrotic proteinuria (Protein/creatinine ratio >200 g/mol), hypoalbuminemia (<25g/l), often accompanied by edema and hyperlipidemia. In contrast, nephritic syndrome means proteinuria (non-nephrotic or nephrotic-range), micro/macrohematuria and hypertension. Etiologies go from frequent postinfectious glomerulonephritis, IgA nephritis, Purpura Schönlein Henoch nephritis, Lupus nephritis to rarer diseases. General work-up basically comprises quantitative urinary analysis, laboratory exams, ultrasound, blood pressure measurement and clinical examination.

Case: A 14-year-old previously healthy girl suffered fatigue for several months. She received iron infusions without evidence of iron deficiency. Then, three weeks after a common cold she developed leg and lid edema and showed nephrotic range proteinuria and elevated creatinine. Her general practitioner suspected nephrotic syndrome and prescribed steroids and torasemide. After symptom worsening, dosages were doubled without any clinical effect. Back pain appeared which led to presentation at the pediatric emergency department. She was in reduced general state, very edematous, hypertensive, laboratory exams showed eGFR (Schwartz) at 57 ml/min/1.73m2, no anemia, leukocytosis, proteinuria (1012 g/mol) and glomerular microhematuria. Abdominal ultrasound was unremarkable. She was admitted to hospital with fluid -, salt restriction and amlodipine. Positive ANA and dsDNS results prompted a renal biopsy which revealed Lupus nephritis class IV + V. Treatment was initiated with steroid pulses followed by oral steroids in reduction, hydroxychloroquin, mycophenolate mofetil and several antihypertensive drugs. No other organ involvements or secondary antiphospholipid syndrome were detected and she was discharged after five days. She is now in regular rheumatologic-nephrologic follow-up and went back to her normal daily routines.

Conclusion: We would like to emphasize that nephrotic range proteinuria does not automatically imply idiopathic nephrotic syndrome. A thorough work-up and recognition of nephritic syndrome with its many differential diagnoses is crucial. Involvement of pediatric nephrologists is desirable in either syndrome. Lupus and lupus nephritis are certainly rare and need specialist (rheumatologist, nephrologist) diagnostics and treatment.

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Beyond proteinuria: status epilepticus in a child with nephrotic syndrome

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Background: Idiopathic nephrotic syndrome is the most common glomerular disease in the paediatric population (Veltkamp 2021). Patients carry an elevated risk of complications such as infections and thromboembolic events.

Case report: A 29 month old Caucasian boy presented to our emergency department with a prolonged status epilepticus of

over 90 minutes. Three weeks prior he had been diagnosed with nephrotic syndrome and had been started on an oral course of prednisolone (60 mg/m2 per day). After an initial good response, he had a relapse one week before the current admission. Additionally, the patient's history included a nonclassic congenital adrenal hyperplasia. Upon admission, the patient was intubated and sedated, precluding a comprehensive neurologic examination. Except for arterial hypertension his vital parameters were normal and he was normoglycemic. A stress dose of steroids was administered for potential adrenal insufficiency and intravenous antimicrobial therapy with ceftriaxone was initiated for possible meningoencephalitis after blood cultures were sampled. The patient urgently underwent non-contrasted cranial magnetic resonance imaging (MRI), which showed three small areas of focal hemorrhage and multiple micro bleeds. As increased intracranial pressure was ruled out, cerebrospinal fluid analysis was performed and revealed a normal cell count. Considering the elevated risk of thromboembolic events in nephrotic syndrome, a neuroradiologist reviewed the MRI images for cerebral venous thrombosis and identified thrombosis of the superior sagittal sinus. Serum fibrinogen, albumin and antithrombin were normal. Anticoagulation with unfractionated heparin was started and was switched to rivaroxaban over the course of the hospitalisation. The patient recovered without any neurological deficits.

Discussion: The imbalance caused by the urinary loss of anticoagulant proteins and the increased hepatic production of prothrombotic factors is believed to represent the main pathophysiological mechanism behind the hypercoagulable state in nephrotic syndrome (Ponticelli 2023). Around 4% of children with nephrotic syndrome suffer from thromboembolic complications—most frequently deep vein thrombosis, cerebral venous or arterial thrombosis, and pulmonary embolism (Dadgar 2023)—which usually occur during relapse (Suri 2014). Cerebral venous sinus thrombosis should be considered in patients with a history of nephrotic syndrome presenting with neurologic symptoms.

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Forehead edema as a rare clinical manifestation of Henoch-Schönlein purpura in paediatric patients.

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Background: Henoch-Schönlein purpura (HSP), also known as immunoglobulin A-associated vasculitis, is the most common vasculitis syndrome in childhood. Its clinical presentation often includes palpable purpura, abdominal pain, arthralgia, and, less frequently, renal involvement. Atypical manifestations, such as seizures, ophthalmic issues, or frontal edema, are rare and poorly described, but may represent a diagnostic challenge.

Case presentation: In mid-December 2024, a 6-year-old child with no relevant medical history presented to our emergency department (Istituto Pediatrico della Svizzera Italiana) with petechiae on the lower limbs, which had appeared the previous day. The child had developed flu-like symptoms a week before, which resolved four days later. Petechiae progressively worsened, extending to the lower limbs and buttocks. Physical examination confirmed the diagnosis of HSP, with palpable purpura, fever, and arthralgia. Biochemical investigations, including renal function tests and vital parameters, were normal except for slightly elevated blood pressure. During hospitalization, worsening petechiae, persistent fever, and intermittent abdominal pain (without abdominal complications on ultrasound) were observed. On day 23 of illness, a rare forehead non-pitting edema (approximately 3.5x3 cm, without erythema or tenderness) appeared. No recent trauma was reported. The edema was monitored and spontaneously resolved within 48 hours without specific treatment. We found fewer than 10 case reports describing this manifestation. In one case, severe facial edema led to disfigurement of the scalp. All cases resolved without complications. Subcutaneous edema in HSP is likely due to blood vessel inflammation, causing fluid leakage. Elevated blood pressure may also contribute by increasing intravascular pressure. However, there is no well-established explanation for why the forehead specifically is affected.

Conclusion: Forehead edema is a rare but possible manifestation of HSP in pediatric patients. This case emphasizes the importance of recognizing and monitoring atypical signs, which are usually benign but can cause concern.

Keywords: Henoch-Schönlein purpura (HSP); case report; forehead edema; atypical manifestations.

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Hypertensive crisis, emergency or urgency: Diagnostic challenges and multidisciplinary management in a case of renovascular hypertension due to unilateral poorly functioning kidney

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Case report: A healthy 5-year-old girl presented with a three-week history of fatigue, morning headaches, nausea and vomiting. Initial examination with blood pressure (BP) measurement was described as normal. Brain MRI ruled out a tumor and fundoscopy was negative. However, during the MRI, elevated BP (170/120 mmHg) was detected, leading to the diagnosis of a hypertensive crisis.

Work-up: Ultrasound revealed a hypoplastic right kidney and a normal left kidney, with no sign of renal artery stenosis. Blood tests showed impaired kidney function with an estimated glomerular filtration rate of 67 mL/min/1.73 m², without electrolyte imbalances. The urine protein/creatinine ratio was elevated at 472 g/mol. Echocardiography confirmed left ventricular hypertrophy (LVH).

Initial management: The patient was admitted to the PICU for a hypertensive urgency. BP reduction was targeted at 25% every 6-8 hours, aiming to reach the 90th percentile within 48-72 hours. Given a favorable response to oral antihypertensive medications, intravenous (IV) treatment was unnecessary. The patient was discharged with a BP stabilized between 90th and 95th percentiles, with maximum doses of three oral antihypertensive drugs (ACE inhibitor, calcium channel blocker, alpha and beta blocker).

Diagnosis and long-term management: Further investigations led to the diagnosis of secondary renal arterial hypertension due to undiagnosed pyelonephritis and high-grade bilateral vesicoureteral reflux with renal sequelae (right kidney function 7% at the DMSA scintigraphy, elevation of the blood renin and aldosterone). The patient underwent laparoscopic right nephrectomy and periurethral bulking agent injection. Nephrectomy enabled the discontinuation of one antihypertensive drug, improved BP control (<50th percentile), and led to the resolution of LVH.

Discussion: Hypertensive crises include: Hypertensive urgency: severe BP elevation with no or mild symptoms. Hypertensive emergency: severe BP elevation and symptoms suggesting end-organ damage. Symptoms are often non-specific, complicating diagnosis and the differentiation between urgency and emergency. Hypertensive crises are more frequent in secondary hypertension, necessitating a complete etiological

work-up. They should be considered life-threatening emergencies and immediate treatment -via oral or IV route- should be initiated. Management in the acute setting requires a delicate balance between gradual BP reduction and the prevention of further end-organ damage?

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Severe Hypertension in a Teenage Boy: A Rare Case of Renal Artery Branch Dissection Treated with Percutaneous Transluminal Angioplasty.

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Background: Severe hypertension in pediatric patients is rare and typically secondary to underlying pathologies, such as renovascular causes. This case highlights the challenges in diagnosing and treating renovascular hypertension caused by dissection of an anterior branch of the renal artery.

Case presentation: A 14-year-old male was admitted with persistent dizziness, headache, and presyncope during school sports. His pediatrician had previously noted elevated blood pressure, corroborated by home measurements. The personal medical history was unremarkable, the family history was positive for cardiovascular disease in the father at a young age, with normal coagulation studies. Despite Nifedipine treatment, systolic blood pressure remained at max 180 mmHg, even at night. The ECG and echocardiographic results were normal. Laboratory results, including renal function, metanephrine levels, vasculitis- and endocrinology panels revealed proteinuria and slightly elevated renin levels. Renal ultrasound showed subtle changes at the right kidney's lower pole, suggesting a renovascular cause. MR-angiography indicated reduced kidney size with hypoperfusion in the ventral renal parenchyma, but no renal artery stenosis was detectable. Finally, interventional angiography confirmed a dissection in an anterior branch of the right renal artery. Screening for fibromuscular dysplasia and connective tissue disorders was negative. Retrospective history revealed a blunt abdominal trauma from a ski accident several months earlier as a possible cause of the dissection.

Management and outcome: Percutaneous transluminal angioplasty (PTA) was performed, dilating the artery to 3mm and anticoagulation with ASA and Rivaroxaban was started. Initially blood pressure remained mildly elevated, requiring continued Nifedipine treatment, but normalized following discharge.

Discussion: This case highlights the importance of evaluating renovascular causes in pediatric hypertension, particularly in the context of a trauma history. Noninvasive imaging may be inconclusive, and interventional angiography could be crucial for the diagnosis. PTA is the method of choice and may provide a safe and effective treatment, although the risk of complications like renal artery damage must be considered.

Conclusion: Renal artery branch dissection is a rare but important cause of pediatric hypertension. A thorough history, advanced imaging, and timely intervention are critical for optimal management and outcome.

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Atrial Fibrillation and Pre-excitation: a deadly combination - also in children

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Background: Pre-excitation can be easily recognized as a delta wave in a 12-lead electrocardiogram (ECG). It is caused by an additional electrical connection from the atrium to the ventricle (i.e. accessory pathway (AP)). We would like to use a case study with instructing ECG traces to illustrate the differential diagnostic considerations when a pre-excitation is detected in a pediatric patient.

Case: A 14-year-old girl was referred to the emergency ward because of exercise-induced palpitations accompanied by weakness, nausea and vomiting. Similar, self-limited episodes had occurred for months. ECG showed a fast and irregular tachycardia with variable (narrow and broad) QRS complex morphology and 156-219 beats per minute measured the fastest. Of note, organized P waves or an isoelectric baseline were absent, as a hallmark of atrial fibrillation (AF). After spontaneous AF termination and conversion to sinus rhythm, ventricular pre-excitation with shortening of the PQ interval, slurring (delta wave) and prolongation of the QRS complex became evident. Hence, FBI tachycardia triggering the girl´s symptoms was diagnosed. She was referred for an electrophysiological study during which the accessory pathway was treated successfully by radiofrequency ablation. During the follow-up period of six months she remained asymptomatic and ECG demonstrated narrow QRS complexes without recurrence of ventricular pre-excitation, indicating complete AP elimination.

Conclusion: Patients with an atrioventricular AP leading to ventricular pre-excitation are at increased risk of developing AF. Studies found that AF is inducible in around 17% of asymptomatic patients with pre-excitation.1,2 In the presence of AF, the ECG of an AP patient shows a fast, broad and irregular tachycardia (FBI tachycardia). Further degeneration to ventricular fibrillation might potentially cause sudden cardiac death. This case report highlights a rare but potentially hazardous case of FBI tachycardia in childhood. Professionals taking care of pediatric patients should recognize that ventricular pre-excitation is not a benign condition but can pose life-threatening risks. This risk can be eliminated by radiofrequency catheter ablation.

P 99

Recurrent syncope with asystole: case report of neurocardiogenic syncope

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Background: Syncope can be defined as a transient loss of consciousness due to global cerebral hypoperfusion and is usually the result of hypotension or bradycardia or a combination of both. Neurocardiogenic syncope occurs with a lifetime incidence of over 35%. If it has a cardioinhibitory effect, it can lead to asystole or severe heart block.

Case study: We report a case of a 2.4-year-old girl who presented with increasing episodes of loss of consciousness. These occurred for up to 15 seconds and 3 times per day since the age of 6 months. The incidents were usually provoked by

crying, pain or getting frightened and presented with loss of tone, pallor, apnea and lack of response with subsequent fatigue for 30-60 minutes. The family history revealed a pacemaker and icd implantation in the maternal grandfather at the age of 64 and suspected childhood epilepsy in the mother. The patient initially presented in the pediatric neurology consultation with age-appropriate development and clinical status as well as inconspicuous findings in EEG and ECG. Subsequently, a 24-hour-ECG was performed for further evaluation. This showed a sinus rhythm most of the time, yet recorded one period of asystole for 7.7 seconds, which correlated clinically with a syncope. The following cardiological clinical assessment, echocardiography and another 24-hour-ECG showed no further pathological findings. Therefore, the diagnosis of neurocardiogenic syncope with distinct cardioinhibitory component was made. Given the increasing duration and long documented asystole, the IIb-indication criteria for pacemaker implantation were fulfilled. Due to the high burden of syncope, the parents agreed to the pacemaker implantation.

Discussion: In most cases, neurocardiogenic syncope proves to be self-limiting. Yet in a few cases, where the quality of life is diminished or traumatic injuries may occur and therefore criteria for pacemaker implantation are met, the procedure aids in preventing prolonged asystole or serious bradycardia with clinical correlation and should therefore be considered. Ideally, asystole or profound bradycardia should be observed during more than one episode of syncope for the diagnosis to be made, yet in this case, given the clinical findings and burden for the family, pacemaker implantation was justified with only one recorded asystole.

P 100

Lucky outcome after syncope during sport in a 14-year-old boy

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Background: Syncope is a common symptom leading to pediatric cardiologic evaluation. While vaso-vagal syncope is typically benign, cardiac causes—such as arrhythmias, cardiomyopathy, and coronary artery anomalies (CAAs)—must be ruled out due to their potentially fatal nature.

Case presentation: A 14-year-old male collapsed at school after 24 minutes of aerobic exercise, presenting with blurred vision, loss of consciousness, and subsequent dyspnea. Emergency evaluation on site revealed hypotension (74/45 mmHg), hypoxia (SpO2 76%), and ST-segment depression in inferior and left precordial ECG leads, raising concerns for myocardial ischemia. The patient had no significant personal or familial history of cardiac disease. After arrival at the emergency department, non-invasive ventilation improved oxygenation, and further investigations showed pulmonary edema on chest X-ray, elevated troponin levels (575 ng/L), and anomalous diastolic flow in the left coronary artery on echocardiography, despite normal cardiac anatomy, function, and no regional wall motion abnormalities. Advanced imaging confirmed an anomalous aortic origin of the left coronary artery (AAOCA) from the right ostium with an interarterial course—a rare congenital anomaly associated with exercise-induced ischemia and arrhythmias.

Management and outcome: The patient was referred to a pediatric tertiary center, where corrective surgery was performed. The procedure involved creating a new coronary ostium in the left coronary sinus using a glutaraldehyde-treated autologous pericardial patch, followed by a side-to-side anastomosis between the left coronary artery and the new ostium. Postoperative recovery was uneventful and the patient is currently in regular follow-up.

Discussion: AAOCA is a rare but critical diagnosis in pediatric patients with exertional syncope, requiring a systematic diagnostic approach and advanced imaging for accurate identification. Surgical correction is essential to prevent life-threatening outcomes in symptomatic patients.

Conclusion: This case underscores the importance of considering coronary anomalies in the differential diagnosis of syncope in pediatric patients, to prevent major adverse events and optimize outcomes.

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Exercise-induced symptoms in children after cancer treatment

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Background: Physical activity is important for childhood cancer survivors (CCS) to prevent chronic health issues, improve fitness, and mitigate side effects of cancer treatment, such as fatigue. However, CCS face potential pulmonary late effects that may impede physical activity. Limited knowledge exists regarding respiratory symptoms that CCS encounter during physical activity. We investigated the prevalence and risk factors of exercise-induced symptoms in young CCS.

Methods: We included children aged 6-20 years who had completed cancer treatment, were ≥1 year post-diagnosis, received regular pediatric oncology follow-up at three clinics in Switzerland, and were treated with any chemotherapy, chest surgery, radiotherapy, or hematopoietic stem cell transplantation. Participants completed a questionnaire on respiratory health and lifestyle. We further collected information about cancer history from medical records. We explored associations between exercise-induced respiratory symptoms and potential risk factors using multivariable logistic regression.

Results: The study included 196 CCS, with a median age of 14 years (interquartile range [IQR] 10-17) and a median time since diagnosis of 7 years (IQR 4-10). Sixty-seven participants (34%) reported exercise-induced symptoms, with most (n = 38, 57%) reporting multiple symptoms. Respiratory symptoms occurred in 46 (24%), including shortness of breath in 27 (14%), cough in 24 (12%), and wheeze in 13 (7%) participants. Participants further reported on side stitches (n = 40, 20%) and premature exhaustion (n = 32, 16%). Older CCS (odds ratio [OR] 1.1, 95%CI 1.0-1.3), obese (4.9, 1.4-16.7), and those with asthma (8.4, 1.8-40.5) were more likely to report respiratory symptoms during exercise. We found no signs of association between treatment-related exposures and respiratory symptoms during exercise.

Conclusions: Our study found that a third of CCS experienced exercise-induced symptoms with older, obese, and survivors with underlying asthma being at highest risk of respiratory

symptoms. Encouraging survivors to engage in physical activity, while addressing these symptoms with tailored support, can improve their exercise capacity and quality of life.

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Metacognitive Intervention in Youth with Oncological Disease - The Mio-App

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Background: Cancer and its treatment places children at increased risk for cognitive and motor long-term problems. Most cognitive and motor trainings currently available for children with cancer have limited efficacy and lack a transfer to non-trained tasks and to everyday functioning. We therefore developed an intervention at the intersection between motor and cognitive science with the aim to strengthen cognitive and motor development of patients with cancer in the long-term using a novel component in neurorehabilitation, namely metacognition. We expect that an increase in metacognition is associated with an increase in cognitive and motor performance.

Materials & Methods: The Mio study is a randomized controlled trial including 40 patients aged 8–16 years after cancer diagnosis shortly before the end of cancer treatment. Patients will be randomly assigned to the training or the waiting control group. We assess the training effect on the primary (metacognition) and secondary outcomes (cognitive and motor functions), before, immediately after the training and at a 3-months follow-up. The training is presented in an app, the Mio-App, with 38 digital games including the playful teaching of mnemonic strategies, intensive working memory training and metacognitive reflections.

Relevance: To reduce cognitive and motor long-term problems in young patients with cancer, it is necessary to address the current lack of effective treatment options. The combination of cognitive and motor training and the novel focus on metacognition will support the childrens' cognitive development and through this facilitate the transfer between cancer treatment and return to everyday and school life.

P 103

Dental Health after Childhood Cancer – A Report from the Swiss Childhood Cancer Survivor Study

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Background: Cancer treatments can affect dental health of childhood cancer survivors. We assessed prevalence of dental problems in survivors, compared them to their siblings, and investigated cancer-related risk factors.

Methods: As part of the population-based Swiss Childhood Cancer Survivors Study, we sent questionnaires to survivors aged 5-19 years and their siblings, inquiring about dental problems. We retrieved cancer-related characteristics from the Swiss Childhood Cancer Registry. We compared dental problems between survivors and siblings and investigated cancer-related risk factors.

Results: We included 735 survivors (median age = 12.8 years [IQR 10.2-15.4]; 46% girls; 70% German-speaking language region; 49% leukemias and lymphomas; 17% CNS tumors) and 144 siblings. Almost half of survivors and siblings reported at least one dental problem. Cavities were the most common problem among survivors (39%; 95%CI 35-43), followed by gum problems during (34%; 30-37) and after treatment (14%; 11-17), and microdontia (11%; 8-13). Compared to siblings, survivors had a greater risk for hypo- or microdontia (OR 1.7; 95%CI 0.9-3.2) and enamel hypoplasia (2.2; 0.8-6.0), but similar risk for cavities or cavity-related tooth loss (0.8; 0.6-1.3). Chemotherapy was associated with enamel hypoplasia (3.0; 1.2-10.4), cavities or cavity-related tooth loss (1.5; 1.0-2.3), and gum problems during (23.0; 9.4-76.2) and after (4.6; 2.0-13.5) treatment. Hematopoietic stem cell transplantation (HSCT) was related to hypo- or microdontia (5.4; 2.6-10.7), cavities or cavity-related tooth loss (2.1; 1.2-3.6), and gum problems during treatment (2.0; 1.2-3.6). Associations with hypo- or microdontia and cavities were most pronounced in patients diagnosed before the age of 5 years.

Conclusion: Childhood cancer patients treated with chemotherapy or HSCT, especially those at a young age, are at increased risk for dental problems. Regular dental check-ups guided by healthcare teams and dental hygiene practices can help mitigate these risks and promote long-term dental health among survivors.

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More than half of time to antibiotics passes before pediatric cancer patients with fever in neutropenia arrive at the hospital

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Background: For fever in neutropenia (FN) during chemotherapy timely start of antibiotics is recommended. It is still unclear if, how and in which patients time to antibiotics (TTA) influences FN outcome. Most studies measure and analyze TTA from arrival at the hospital to start of antibiotics. Here we aimed to analyze timespans between detection of fever and start of antibiotics in children undergoing chemotherapy with cancer and FN. Specifically we aimed to assess where delays happen and what variables influence TTA. We further aimed to see whether the order of certain process sequences are associated with TTA.

Methods: We analyzed data collected in the prospective multicenter Swiss Paediatric Oncology Group (SPOG) 2015 FN Definition study. We investigated 14 different timespans. Times analyzed were: first fever measurement, phone call, decision to treat FN, arrival at the hospital, complete blood count (CBC), prescription, start and end of antibiotics. We assessed nine variables (sex, age, location at diagnosis, center, weekend, office

hours, season, severity of disease at presentation, temperature limit defining fever), and two modifiable orders of process sequences (time point of decision to treat FN (before vs. after arrival) and time point of CBC (before vs. after recognition of fever)) for potential association with TTA.

Results: We analyzed 349 FN episodes in 155 patients from April 2016 to August 2018. In outpatients median TTA from fever to start of antibiotics was 165 min, with a median duration of 80 min from fever to arrival at the hospital. For inpatients median TTA from fever to start of antibiotics was 75 min. The longest delays were identified for the timespans phone call to arrival (median, 75 min) and arrival to decision on treatment (60 min). Known blood counts at recognition of fever, decision to treat FN before arrival at the emergency department and arrival during office hours (defined as daytime and Monday to Friday) contributed to shorter TTA.

Conclusion: Time passing before arrival at the hospital is relevant and should be considered when optimizing TTA and evaluating its influence on outcomes. Support for transportation may help to shorten time to arrival during the night, and the time passing from phone call to arrival should be used for preparations. Regular blood counts are indicated in children under chemotherapy, as knowledge of neutropenia and correspondingly early decision to treat FN, increases the chance for fast treatment.

P 105

Use of emergency departments for minor health problems among refugee and immigrant children and youth in Ontario, Canada – a population-based cohort study

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Importance: Emergency department (ED) use for minor health problems is an indicator of inadequate care navigation and management.

Objective: Our objective was to determine whether refugee and immigrant children visited the ED more frequently for all minor health problems early in the resettlement period.

Design: We conducted a population-based cohort study including immigrants (arriving 2008-2017) and matched Ontario-born minors. Participants were followed for 4 years.

Setting: Ontario, Canada

Participants: Children 0-14 years old at index.

Exposures: Refugees (government-assisted [GARs], privately sponsored [PSRs], successful asylum-seekers [SASs]), non-refugee immigrant [NRIs], Ontario-born children matched to each immigrant group on age, sex and residential geography at index.

Main outcomes/measures: The average proportion of all minor health visits (i.e., the sum of primary care sick visits plus ED visits for equivalent reasons) seen in the ED in the first and second two years after eligiblity for healthcare insurance. Using linear regression we modeled the difference in the average proportion of minor visits to the ED with 95% confidence intervals (CI), comparing each immigrant group to Ontario-born minors while adjusting for major morbidity, material resources quintile and primary care model.

Results: Overall, 737,695 minors were included (GARs N = 12,184; PSRs N = 9,402, SASs N = 14,255, NRIs N = 111,698; Ontario-born matches N = 590,156). In the first two years, all immigrant groups had significantly lower average adjusted proportions of minor visits to the ED compared to their Ontario-born matches (GARs -5.11% (-5.63% to -4.57%); PSRs -5.24% (-5.80% to -4.67%); SASs -3.37% (-3.86% to -2.87%); NRIs -4.24% (-4.40% to -4.09%). In the second two years, differences were slightly attenuated but remained significantly lower for all immigrant groups.

Conclusion: Contrary to our hypotheses, all refugee groups and non-refugee immigrant minors were less likely to use EDs for minor health problems than Ontario-born minors, suggesting they are appropriately navigating the healthcare system early in the resettlement period.

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"The Pill in the Diaper" - Malignant Bowel Obstruction in a 15-year-old Girl in Pediatric Palliative Home Care

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Background: Malignant bowel obstruction (MBO) is a serious complication in advanced cancer, primarily affecting adults but rarely seen in paediatric patients. When surgery is not feasible, management typically involves analgesics, antiemetics, and anti-secretory drugs. Octreotide has been a first-line treatment for over thirty years, yet paediatric dosing for MBO is underresearched. This study describes the management of MBO in a 15-year-old girl characterizing the care provided at home.

Case description: A fourteen-year-old girl with neurofibromatosis type 1 was diagnosed with an inoperable abdominal malignant peripheral nerve sheath tumour. After her condition progressed despite intensive treatment, she transitioned to paediatric palliative home care. Her abdominal tumour compressed her stomach, limiting oral intake and causing pain. After discovering an undissolved oxycodone pill in her diaper, we suspected that incomplete proximal bowel obstruction led to explosive transportation of intestinal content thereby hindering drug absorption. We initiated a hospital stay and rotated her pain medication to intravenous (IV) methadone, then hydromorphone. Symptoms were managed for five weeks until complete bowel obstruction signs emerged. She declined a nasogastric tube, so we started continuous IV octreotide at 1 mcg/kg/hour, escalating to 5 mcg/kg/hour within the following three days. This effectively controlled her symptoms for ten days until her passing at home. Her care involved a high intensity, well-coordinated, interdisciplinary care network, including specialized palliative care, responsive home nursing support, and local physician involvement.

Discussion: Optimal dosing of octreotide for paediatric MBO is unclear, though typical doses for other uses range from 0.5 mcg/kg/hour to a maximum of 10-20 mcg/kg/hour. Based on our findings and previous reports, starting at 3-5 mcg/kg/h with rapid escalation as needed may be effective. Home management of malignant bowel obstruction requires a coordinated care network, especially in Switzerland where paediatric palliative care is underdeveloped. This case highlights the need for improved collaboration among hospital teams, home care

nurses, and local physicians, serving as a potential model for integrated palliative care in paediatrics.

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PoCUS for pediatric shock assessment: the BePS-protocol (Berner Schockprotokoll)

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Point-of-Care Ultrasound (PoCUS) is an impactful diagnostic tool in the assessment of shock and its importance and evidence is progressively increasing in pediatrics. The recent EPALS guidelines released in 2021 by the European Resuscitation Council recommend and support the use of PoCUS in advanced life support by trained pediatric emergency physicians. In 2023 the American Society of Echocardiography released recommendations for pediatric cardiac PoCUS defining its use to evaluate physiologic causes and subsequent effects of hypotension, shock, and circulatory arrest in children.

We propose a simplified PoCUS protocol for shock assessment, called BePS (Berner Schockprotokoll), and illustrate by two clinical vignettes how the use of PoCUS during the primary survey of patients presenting with undifferentiated shock is determinant for diagnostic and therapeutic guidance.

Advances in pediatric PoCUS and the increasing awareness of its importance for pediatric shock assessment has led to the creation of a Swiss board-certified formal training. It was launched by the Swiss Society for Ultrasound Medicine (SGUM) in December 2024 and defined as "focused transthoracic cardiac ultrasound in pediatrics - PoCUS component no. 14" (Certificate of competence in Point-of-Care Ultrasound). The BePS is an integral part of the training and shall help facilitating early-use of PoCUS in pediatric shock assessment among Swiss pediatric emergency medicine departments and intensive care units.

P 108

The use of intestinal ultrasound in the care of pediatric patients with inflammatory bowel disease in Switzerland

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Introduction: Intestinal ultrasound (IUS) is emerging as a bedside examination tool in the care of patients with inflammatory bowel disease (IBD). IUS detects disease activity, therapy response, progression and complications. Being non-invasive and non-radiating it is the optimal tool for use in pediatric patients. The aim of this study is to define the use of IUS in pediatric IBD in Switzerland.

Methods: An anonymous online questionnaire was distributed to all pediatric gastroenterologists (PG) licensed in Switzerland.

Results: 18 out of 27 (67%) invited PG participated in the survey. Four PG had received formal training in IUS. Two perform IUS themselves, 16 PG refer their patients to the radiology department for IUS assessment. 15 PG agreed that IUS would provide high or very high benefits in the care of patients with IBD and three give it a low priority. PG valued IUS for additional information about response to current treatment (15), immediate result for clinical correlation (14) and revealing of possible complications (12). Additionally, five PG expressed the view that the use of IUS could reduce the need for invasive procedures such as endoscopy. Ten PG were convinced that IUS enables a better understanding of the patient's chronic illness. Various hurdles to implementing IUS into daily care were mentioned: lack of an available ultrasound machine (9), expensive training requirement (7), additional time needed during patients visits (8) and a missing reimbursement code (4).

Conclusion: Despite of its many advantages, IUS is not regularly used in the follow-up of pediatric IBD in Switzerland. The main hurdles are the high cost of training, time-consuming patient visits and lack of reimbursement. Therefore, superordinate concepts are needed to make this valuable tool accessible to vulnerable pediatric IBD patients.

P 109

When Bertolotti grooves with Castellvi: A rare cause of lower back pain in a young ambitious female dancer.

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Bertolotti's syndrome, a rare but significant cause of lower back pain (LBP) in paediatric populations, is often associated with lumbosacral transitional vertebrae (LSTV). We report a case of a 10-year-old girl presenting with acute LBP following a tumble during dance training. Physical examination revealed tenderness along the paravertebral regions and from thoracic-T8 to lumbar vertebra-L5 with sharp, non-radiating pain augmented by left lateral flexion. No neurological deficits were observed. Plain radiographs were unremarkable. An MRI scan identified a Castellvi Type IIa LSTV with pseudoarthrosis, microfractures, and degenerative changes in both sacroiliac joints. A diagnosis of Bertolotti's syndrome was established, likely exacerbated by repetitive mechanical stress from dance training. Symptoms resolved with conservative management of rest, activity modification, physiotherapy, and NSAIDs. At follow-up, the patient remained asymptomatic, with full functional recovery.

LSTVs, present in approximately 12% of the population, are classified using the Castellvi system, ranging from transverse process enlargement (Type Ia, Ib) to complete fusion of L5 and S1 uni- or bilateral (Type IIIa, IIIb resp.). Castellvi Type II lesions, particularly unilateral forms (Typ IIb), are prone to symptomatic presentations due to asymmetric spinal loading, leading to abnormal torque, scoliosis, and adjacent segment degeneration. The diagnosis of Bertolotti's syndrome is primarily image-based with pelvic radiographs providing initial assessment. Management of Bertolotti's syndrome involves conservative interventions such as physiotherapy and NSAIDs initially. In intractable cases, interventional approaches, including steroid injections or surgical resection of the pseudoarthrosis, may be warranted. Therapeutic guidelines or consensus recommendation in Bertolotti's syndrome are currently absent.

This case highlights the importance of considering Bertolotti's Syndrome in the differential diagnosis of LBP in paediatric patients, particularly in physically active individuals with a higher training load. Further common differentials of LBP are functional reasons, irritation of the facet joints, spondylolysis or Morbus Scheuermann. Prompt imaging and tailored conservative treatment can result in symptom resolution and a return to full activity, as demonstrated in this patient.

P 110

A rare case of pediatric microscopic polyangiitis with cardiac manifestation

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Background: Microscopic polyangiitis (MPA) is a necrotizing ANCA-associated vasculitis of the small vessels that frequently affects the respiratory tract and kidneys, but may involve other organ systems. Pediatric MPA is extremely rare and mostly occurs in early adolescence. Limited epidemiologic data and lack of diagnostic criteria may delay the diagnosis. We present a pre-school girl with MPA and cardiac arrhythmia.

Case: A previously healthy four-year-old girl presented in poor general condition with anuria for 24 hours, a seven-day history of dark urine, bilious vomiting, diarrhea, and a five-day history of high spiking fever at our emergency department. The girl had received ibuprofen (8 mg/kg/day) for both leg and abdominal pain for the past weeks. Clinical examination revealed mild dehydration without remarkable features (notably no fever, unremarkable abdominal and leg exam, or cardiopulmonary abnormalities). Laboratory testing confirmed severe acute kidney injury (peak serum creatinine 538 µmol/l, urea 40 mmol/l) with elevated inflammatory markers (CRP 171 mg/dl)), low-normal hemoglobin (102 g/l), normal leukocyte and eosinophile counts, no fragmented red blood cells, reduced C3, normal C4, ANA 1:80, pANCA 1:320, and negative anti-MPO and anti-PR3. Renal biopsy and capillary microscopy showed findings in accordance with MPA (capillary rarefication, active vasculitis of small vessels, tissue necrosis, negative immunohistochemistry). Interferon signature, cranial MRI, chest X-ray, ophthalmologic and neurological examination were unremarkable. Treatment with high-dose steroids and rituximab was initiated 26 and 53 days after symptom onset, respectively. On day 30 of hospitalization, the girl developed nocturnal drops in heart rate (25 bpm). An ECG showed a heterogeneous pattern of non-haemodynamically relevant cardiac arrhythmias (AV block II Wenckebach, extrasystoly, supraventricular couplets, and sinus tachycardia (130 bpm)) with unremarkable echocardiography. Laboratory evaluation showed elevated BNP (4410 ng/l) with normal troponin and electrolytes. Soon after treatment start, general condition improved, renal function stabilized with non-nephrotic proteinuria, and arrhythmia resolved.

Conclusion: Pediatric MPA with cardiac manifestation is extremely rare. Since MPA can affect blood vessels in any organ, a low-threshold workup of further symptoms is recommended.

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Don’t forget macrophagic activation!

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Case. R: An 11-year-old previously healthy girl presented with a decline in general health, characterized by persistent high fever(41°C), skin rash, abdominal pain and progressive joint pain preventing her from walking.Symptoms began in late July during a vacation in Spain.August, she sought medical attention for persistent symptoms.Clinical examination revealed a maculopapular rash that disappeared under pressure,pruritic areas on otherwise healthy skin,cold extremities during febrile peaks without signs of shock, and right knee swelling with severe pain preventing joint examination.The rapid streptococcal test was negative, and corticosteroids were initiated. Laboratory tests showed mild inflammation(CRP31.4 mg/L, WBC 11.8),and Azithromycin was prescribed for three days.Further investigations revealed no fractures or neoplastic lesions on the right knee X-ray and no effusion on knee ultrasound.Respiratory tests for Influenza, RSV, COVID, Mycoplasma, Chlamydia, Parvovirus,EBV, and Measles were negative.Stool tests for adenovirus, rotavirus, Campylobacter, Salmonella, Shigella, and E. coli were negative.Laboratory findings showed elevated liver enzymes(ALAT 45 U/L), hypertriglyceridemia(2.6 mmol/L),and high ferritin(1292 µg/L),suggesting macrophage activation syndrome(MAS).Blood cultures were negative. Elevated interleukins(IL-1,IL-18,IL-31,sCD25) confirmed MAS.Treatment with methylprednisolone was started and transitioned to oral prednisone.Infectious workup for Bartonella,Leishmania,Rickettsia,and Leptospira was negative.Abdominal ultrasound was normal.ECG showed no pericarditis despite suspicion of Still’s disease.Immuno-rheumatology specialists initiated ciclosporin therapy.Complications during the disease included left axillary deep vein thrombosis from a catheter,CMV reactivation with thrombocytopenia,mixed anemia,insulin-dependent diabetes mellitus,corticosteroid-induced systemic hypertension and intraocular hypertension,osteopenia,Candida albicans esophagitis, musculoskeletal deconditioning,and stage 1 sacral pressure ulcer, leading to prolonged hospitalization.

Discussion: This case highlights diagnostic challenges due to symptom onset within 6weeks and emphasizes the importance of early recognition of MAS criteria to anticipate complications.This case was refractory to treatments such as tocilizumab,ciclosporin, and anakinra, but responded to MAS-825.

Ccl: Early recognition of acquired MAS is essential as it can progress to a fatal form if untreated.

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When time doesn’t heal: a persistent case of FPIES to fish

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Food Protein-Induced Enterocolitis Syndrome (FPIES) is a non-IgE mediated food allergy that primarily affects the gastrointestinal system. It typically presents in infancy or early childhood. Acute FPIES is characterized by repetitive vomiting occurring 1 to 4 hours after ingestion of the triggering food. Other common symptoms include diarrhea, lethargy and pallor. In severe cases FPIES can lead to dehydration and shock. Common trigger foods are cow’s milk, egg, rice and fish. FPIES typically occurs once cow’s milk formulas or solid foods are introduced into the infant’s diet. The chronic form of FPIES develops over days to weeks with regular ingestion of the offending food, usually cow’s milk, and affects younger infants. It presents with frequent watery diarrhea, emesis and failure to thrive. Diagnosis is based on clinical history and confirmed by oral food challenge as there are no specific laboratory tests to confirm the condition. Treatment of acute episodes consist of ondansetron, rehydration and steroids. In general, strict avoidance of trigger foods is necessary.

Case presentation: A 3-year-old boy presented with repetitive vomiting, lethargy and pallor within two hours after the first

consumption of white fish. Due to the clinical presentation FPIES to fish was diagnosed. At the age of 9 years an oral food challenge (OFC) was initiated to confirm tolerance as expected by school age. Two hours after ingesting cod, he developed repetitive vomiting, pallor and lethargy. Treatment required ondansetron administration and intravenous rehydration. In consequence, strict avoidance of fish remains necessary.

Clinical presentation of FPIES can be severe and leads to shock. Despite that, awareness of FPIES is low. Once diagnosed, close follow-up with dietary advice to provide guidance during the introduction of complementary foods is necessary. Additionally, families need to know how to react in case of acute FPIES. Finally, OFCs are needed to reassess tolerance over time. The age of development of tolerance in patients with FPIES varies by food trigger and country of origin. For Europe, available data are limited. A recent study from Germany showed that time to proven tolerance was shortest in cow's milk and takes longer in fish, meat or vegetables. Here, we present a case of FPIES persisting beyond the age of 9 years. This should be considered in clinical practice and further studies are needed to enhance the understanding and management of FPIES.

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Effects and mechanisms of Sensory Afferent Electrostimulation (SAES) to improve Upper Limb Function in children with spastic Unilateral Cerebral Palsy: A Randomized Controlled Study – Study Protocol

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Background: Children with hemiparesis present with sensory and motor deficits, which negatively affect quality of life and decrease participation in everyday life. It is challenging to find the best individual training method for children with hemiparesis, as there is much variability in treatment response. In the present study, the effects and mechanisms of non-invasive electrostimulation of the hand, called Sensory Afferent Electrostimulation (SAES), will be investigated. SAES triggers action potentials in afferent nerve fibres leading to increased sensory afferent input in the sensorimotor regions of the brain. While proven effective in adults after stroke, SAES is safe with promising positive results in small studies in children with cerebral palsy.

Methods: The study design is a randomized controlled Bayesian phase II trial with a follow-up examination after 12 weeks. The study participants will be recruited via the University Children's Hospital Bern, the Swiss CP Register and the Swiss Neuropediatric Stroke Registry. The experimental intervention consists of a home-based SAES with a glove or adhesive electrodes (e.g. CefarTENS chattanooga) at 1-40Hz, 300µs, intensity between 2-10mA, during 30 minutes per day, 5x/week, for 5 weeks, combined with conventional occupation therapy. The subsequent therapy sessions will take place at home, with in-between telephone checks and communication via app. The control intervention (treatment as usual, TAU) consists of the prescribed conventional occupational therapy. The blinded evaluation comprises bimanual function and unimanual hand function, before and after therapy, with a follow up of 12 weeks. We will furthermore assess the efficacy of SAES using novel clinical assessment such as kinematic evaluations at the instrumented apartment (NeuroTec Loft). Magnetic Resonance Imaging will describe brain lesion characteristics, Resting-state functional MRI the topographically connectivity and Transcranial Magnetic Stimulation corticospinal projections and intracortical changes.

Conclusion: This project investigates a non-invasive stimulation method to improve hand function in children with hemiparesis. The results of the study thus fill an important knowledge gap in the medical literature and could provide the basis for implementing such stimulation in practice.

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Evaluation of the needs and satisfaction of families treated at the HUG's CORAIL Centre (Centre for Interdisciplinary Coordination and Care of Rare and Complex Diseases in Children and Adolescents) after two years of activity

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Complex rare paediatric diseases present considerable medical, psychosocial and educational challenges for families and carers. To provide the best possible response, in 2023 the HUG opened the Centre for interdisciplinary coordination and care of complex rare diseases in children (CORAIL). Its priority is to offer a personalised, coordinated care pathway for each child, with a multidisciplinary team that will support the child and his family through until the transition to adulthood. However, there is still a need to gain a better understanding of the impact of care by and on families in order to propose optimal support strategies. The aim of the study is to assess whether the care currently offered by CORAIL is in line with the expectations and needs of families.

An essentially quantitative questionnaire, based on data from the literature and including the validated ZARIT questionnaire, was sent to the 157 families followed at CORAIL in December 2024, with the aim of identifying needs in terms of medico-psycho-social support, exploring the impact of the child's condition on the families' daily lives and quality of life, and assessing satisfaction with the Centre's proposals.

47 families responded to the questionnaire. The responses concerning the impact on the parents' daily lives and quality of life revealed that the child's condition represented a moderate to severe "burden" in almost half the cases, and severe for 5 families according to the ZARIT grid. The two priority needs identified by the families are related to access and transmission of medical information, as well as listening to and including parents in decision-making. Centre CORAIL's activities met these needs for 80.5 % of the families. 85.1 % rated their satisfaction with the Centre >6 on a scale of up to 10, 40.4 % answered 10. 91.5 % of the parents would recommend a CORAIL follow-up to other families.

The results suggest that CORAIL's current care offer tends to be in line with the specific needs of families, although improvements should be considered in certain areas, in particular improving the circulation of information, the visibility of existing resources and links with other structures. A wider evaluation of the CORAIL Centre among families and professionals in the network is planned for 2025.

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Availability and quality of anthropometric data in hospitals: first results from the SwissPedGrowth project

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Introduction: Anthropometric data, such as body height and weight, are crucial in pediatric care and routinely assessed during clinical visits. The national data stream Pediatric Personalized Research Network Switzerland (SwissPedHealth) makes this data available and standardized for research by adhering to common data models. Hence, we describe the feasibility of retrieving height and weight from electronic health records (EHRs) in Swiss children's hospitals and evaluate the data quality.

Methods: SwissPedGrowth is a project nested within SwissPedHealth and analyses data from patients (<20 years) who visited hospitals in Basel, Bern, Geneva, Lausanne, Zurich, Luzern, or St. Gallen between 2017–2023. Clinical data warehouses transferred anonymous anthropometric measurements (height, weight, head and waist circumference), socio-demographic information (sex, age, nationality, socio-economic status), and clinical information (diagnoses, procedures, treatments) to BioMedIT. The national infrastructure BioMedIT enabled us to process the data in a secured way. We applied a weighted moving average of z-scores to identify and correct unit errors and swapped recordings, remove same-day and carried-forward duplicates, and flag implausible height and weight values (outliers) using the growthcleanr algorithm of Daymont et al.

Results: The first data deliveries from Basel, Geneva, and Lausanne included 46,585 children (54% boys) who, between 2017-2023, had 37,182 inpatient stays and 240,822 outpatient visits. The data contained 105,167 heights, 233,594 weights, 32,743 head, and 1,308 waist circumferences. Height was recorded during 53% of inpatient stays and 11% of outpatient visits, and weight during 87% of inpatient and 19% of outpatient visits. Many children had at least one height (n = 20,496, 44%) and one weight (n = 30,214, 65%) recorded. Among children with at least one measurement, a median of 2 (IQR: 1, 6) heights and 3 (2, 7) weights were available per child. We corrected 767 (0.2%) unit errors and 34 (<0.1%) swapped recordings and removed 37,506 (11%) same-day and 43,268 (13%) carried-forward duplicates. Of the remaining height and weight values, 3,174 (1%) were flagged as implausible.

Conclusion: SwissPedGrowth demonstrates the feasibility of retrieving high-quality anthropometric data from EHRs in Swiss children's hospitals for research. SwissPedGrowth provides a framework for future studies on growth and obesity in Swiss children.

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A Curious Case of Insulinoma with Symptomatic Hypoglycemia

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Background: Insulinomas are neuroendocrine tumors classified among rare diseases, with an incidence of about 0.4 per 100,000 people per year. These tumors are responsible for the hyperproduction of insulin regardless of the patient's blood glucose levels, causing neurohypoglycemic symptoms. The mechanism by which these tumors bypass the feedback mechanisms regulating insulin production is not fully understood yet. A recent study suggests that insulinomas may produce a splice variant of the mRNA responsible for insulin production, which is more abundant and effective than the native mRNA.

Case report: We present the case of a 6-year-old child referred by his family pediatrician to the pediatric endocrinologist due to recurrent episodes of vertigo, sleepiness, tremors, and confusion, without loss of consciousness. The episodes almost always occurred during the preprandial hours, around 11 AM, and each episode lasted about one minute. These episodes were often followed by fatigue and hunger, and while symptoms sometimes resolved after eating, this was not always the case. Blood tests evaluated by the specialist showed slightly elevated IGF levels, but all other results were within normal ranges, including normal blood glucose and insulin levels, with an HOMA index of 0.9. After excluding a neurological cause for the symptoms, a glucagon test was performed, revealing high insulin levels and low blood glucose in an abnormal pattern. An abdominal ultrasound showed a hyper-echoic lesion in the pancreas, which was subsequently confirmed with an MRI and PET scan as an insulinoma (T2 Nx). Taken into account the diagnosis, known the existence of genetic variances needing a specific therapeutic strategy a test aimed at finding specific genetic mutation has been carried out looking for the following mutations: GCK, GDH, HADH, HNF4A. HNF1A, UCP2, SLC16A1, PMM2, HK1, PGM1, FOXA2, CACNA1D, EIF2S3, ABCC8, KCNJ11, paternal mutation of the KATP channel and the heterozygous loss at the level of chromosome 11p15. The test came out negative. The condition required the combined efforts of the dietary specialist, endocrinology, and surgical teams for management, and the patient ultimately underwent enucleation of the lesion at the CHUV of Lausanne with positive results.

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