SWISS SOCIETY OF PAEDIATRICS

ABSTRACTS OF THE ANNUAL MEETING 2022

TABLE OF CONTENTS

ORAL COMMUNICATIONS

OC 1

RAFAEL: A useful chatbot to interact with children about post-covid (long covid) condition

Perrin A[1], Caflisch M[1], Schneider F[2], Yu WS[2], Guessous I[3], Nehme M[3], Posfay-Barbe KM[4]

[1]Adolescent clinic, Division of General Pediatrics, Department of Woman, Child, and Adolescent Medicine, Geneva University Hospitals, Geneva, Switzerland; [2]Division of Digital Communication, Departement of Communication, Geneva University Hospitals, Geneva, Switzerland; [3]Division and Department of Primary Care Medicine, Geneva University Hospitals, Geneva, Switzerland; [4]Division of General Pediatrics, Department of Woman, Child, and Adolescent Medicine, Geneva University Hospitals, Geneva, Switzerland

Post-Covid condition (or long covid) describes the persistence of symptoms several months to years after SARS-CoV-2 infection. It has been increasingly described in children as well, particularly in preteens and teenagers. Most commonly reported symptoms include fatigue, difficulty concentrating, sleeping disorders, shortness of breath, and mood disorders. Treatment of these symptoms is limited and consists mostly of adaptation of physical and mental activities, reconditioning and motivational support. Improving information and awareness of this condition are priorities to properly evaluate and treat these children. The Department of General Pediatrics in Geneva has set up a specific consultation for children and adolescents in spring 2021. In addition, in collaboration with the Department of Community Medicine and Primary Care and Emergency Medicine, we developed an academic digital citizen platform (RAFAEL), which combines an information site, a chatbot, and webinars/workshops for both children and adults.

We report here our experience with the use of the pediatric chatbot which was launched on February 1st 2022. The chatbot is a conversational agent that can answer questions in real-time and reorient people to a specialist/advisor if needed. Chatbot technology is useful in providing verified information and prompting communication and interaction with a large number of people simultaneously and can be adapted to all age groups. It is particularly appealing to teenagers who are very familiar with the concept of chatbot technology and may find this way to communicate more appealing than regular website information or traditional literature. We will report the initial frequently asked questions and their evolution over time within this chatbot. We will also report the number of users of the pediatric chatbot, the overall and individual match rates, and users' satisfaction.

A platform, such as RAFAEL, could become a new way to interact with young patients regarding non urgent subjects, prevention, chronic diseases, as well as a way to use their regular feed-back to be as close as possible to the patients' needs.

OC 2

Complementary and alternative medicine: Use in children and adolescents with inflammatory bowel disease in Switzerland

Guilcher K[1], Pittet V[2], Petit LM[3], Heyland K[4], Braegger C[5], Nydegger A[6], Garzoni L[7], Schibli S[1], Spalinger J[8], Righini-Grunder F[8], Sokollik C[1], Swiss paediatric IBD cohort study investigators

[1]Division of Pediatric Gastroenterology, Hepatology, and Nutrition, University Children's Hospital of Bern, Switzerland; [2]Institute of Social and Preventive Medicine, University Hospital Lausanne, CHUV, Lausanne, Switzerland; [3]Division of Pediatric Gastroenterology, University Hospital of Geneva, HUG, Geneva, Switzerland; [4]Department of Pediatrics, Cantonal Hospital Winterthur, Winterthur, Switzerland; [5]Division of Pediatric Gastroenterology and Nutrition, University Children's Hospital of Zurich,Switzerland; [6]Division of Pediatric Gastroenterology, University Hospital of Lausanne, CHUV, Switzerland; [7]Pediatric Gastroenterology, Clinique des Grangettes, Geneva, Switzerland; [8]Division of Pediatric Gastroenterology, Children's Hospital of Lucerne, Switzerland

Background

Complementary and alternative medicine (CAM) is considered any treatment beyond conventional medicine. Data about the use of CAM in children with inflammatory bowel disease (IBD) is lacking.

Aim

To evaluate the use of CAM in pediatric IBD patients in Switzerland and to identify associated factors.

Methods

Prospective cohort study employing a questionnaire about the detailed use of CAM in conjunction with clinical data from the Swiss pediatric IBD cohort study.

Results

We analysed data of 108 patients, 58 with Crohn's Disease (CD) and 50 with Ulcerative colitis (UC) or IBD- unclassified (IBD-U). The median age at diagnosis was 10.2 years. 42 (38.9%) patients reported to have used CAM during their disease course. Most used CAM was phytotherapy in 22%, followed by homeopathy (15%), massages (7%) and kinesiology (6%). Interestingly, 51% IBD patients followed a self-imposed restricted diet.

CAM users were younger at diagnosis (7.8 vs 10.1 years ($p = 0.01$)) and had a more extensive phenotype at diagnosis (for UC, $p = 0.002$). However, we did not find an association between CAM use and disease activity (PUCAI or PCDAI score) ($p = 0.7$ resp. $p = 0.8$) or use of TNF-alpha inhibitor ($p = 0.78$) as markers of a more severe disease course.

47% of caregivers attributed a positive impact to the use of CAM, this was in 80% in accordance with the view of the patient. There was no significant difference seen in overall quality of life ($p = 0.18$), bowel ($p = 0.77$) and systemic symptoms ($p = 0.92$), body image ($p = 0.12$) or emotional functioning ($p = 0.98$) between CAM and no-CAM users.

Conclusion

While there is a high demand by caregivers to use CAM and a positive attribution of effects to their use, our data assumes that CAM use does not make a relevant clinical difference. Clinical activity of disease do not seem to be an explanation of CAM use. Practitioners should be aware about CAM use in pediatric IBD patients to include CAM in their management strategies. Increasing knowledge about CAM, dietary restriction and supplement use will ensure a safer and more coordinated care for patient.

OC 3

Non-operative treatment versus suture refixation of the nail plate in paediatric fingernail avulsion injuries

Seiler M[1,2], Gerstenberg A[2,3], Kalisch M[4], Kennedy U[2,3], Scheer HS[3], Weber DM[2,3]

[1]Pediatric Emergency Department, University Children's Hospital Zurich, Zurich, Switzerland; [2]Children's Research Center, University Children's Hospital Zurich, Zurich, Switzerland; [3]Division of Hand Surgery, University Children's Hospital Zurich, Zurich, Switzerland; [4]Seminar for Statistics, ETH Zurich, Switzerland

Background

Fingertip injuries with avulsion of the nail plate are among the most common hand injuries treated in pediatric emergency departments (EDs). Many children sustain only isolated avulsion injuries with or without non-displaced fractures of the distal phalanx. The standard treatment recommendation for such injuries consists of exploration of of the nail bed, repositioning of the nail plate under the eponychial fold and suture fixation of the nail plate, which often requires general anaesthesia, particularly in young and uncooperative children. However, whether this nail refixation is necessary has not yet been studied. Therefore, the aim of this study was to assess whether non-operative treatment of fingernail avulsion injuries is not inferior to the standard operative treatment for the outcome regarding nail appearance and patient satisfaction.

Method

A non-inferiority hypothesis was tested in a single-center, prospective cohort study, comparing non-operative with operative treatment options in children 1-16 years with fingernail avulsion injures. Exclusion criteria were lacerations of the nail bed with a gap >1 mm and displaced fractures. Allocation to the treatment group was according to parental choice. The quality of the new nail was the primary outcome and was assessed with the Nail Appearance Score, the secondary outcome was patient and parental satisfaction, which was assessed with the Patients' and Parental Nail Satisfaction Score, a patient-reported outcome score. Both scores consist of six categories that are assessed with 4-point Likert scales with one being the lowest and four being the highest score.

Results

Fifty-one patients were enrolled with a median age of 3 years (IQR 5; range 1–14). Thirty-nine (76.5%) chose the non-operative treatment and 12 (23.5%) the standard operative therapy. The two groups did not differ in age, sex or presence of fractures on X-rays. No differences were found for the primary or secondary outcomes between the two groups.

Conclusion

The outcome was excellent in all fingers, and all patients and parents very satisfied. Comparison of the two groups confirmed the non-inferiority hypothesis for the non-operative treatment. In view of associated risks and costs for surgery, we recommend non-operative treatment for simple nail avulsion injuries in children.

OC 4

Multiple neonicotinoids in children's cerebro-spinal fluid, plasma, and urine

Laubscher B[1,2,3], Diezi M[1,3], Renella R[1,3], Mitchell EAD[4], Aebi A[5], Mulot M[4], Glauser G[6]

[1]Department of paediatrics, Lausanne University Hospital, Switzerland; [2]Department of paediatrics, Réseau hospitalier neuchâtelois, Neuchâtel, Switzerland; [3]Lausannne University, Lausanne, Switzerland; [4]Institute of biology, University of Neuchâtel, Switzerland; [5]Institutes of biology and anthropology, University of Neuchâtel, Switzerland; [6]Neuchâtel Platform of Analytical Chemistry, University of Neuchâtel, Switzerland

Background

Neonicotinoids (NN) are selective neurotoxic pesticides that bind to insect but also mammal nicotinic acetycholine receptors (nAChRs). As the most widely used class of insecticides worldwide, they are ubiquitously found in the environment, wildlife, and foods, and thus of special concern for their impacts on the environment and human health. nAChRs are vital to proper brain organization during the prenatal period and play important roles in various motor, emotional, and cognitive functions. Little is known on children's contamination by NN. In a pilot study we tested the hypothesis that children's cerebro-spinal fluid (CSF) can be contaminated by NN.

Methods

NN were analysed in leftover CSF, blood, and urine samples from children treated for leukaemias and lymphomas and undergoing therapeutic lumbar punctions. We monitored all neonicotinoids approved on the global market and some of their most common metabolites by ultra-high performance liquid chromatography-tandem mass spectrometry.

Results

From August to December 2020, 14 children were consecutively included in the study. Median age was 8 years (range 3-18). All CSF and plasma samples were positive for at least one NN. Nine (64%) CSF samples and 13 (93%) plasma samples contained more than one NN. Thirteen (93%) CSF samples had N-desmethyl-acetamiprid (median concentration 0.0123, range 0.0024-0.1068 ng/mL), the major metabolite of acetamiprid. Total NN CSF content varied from 0.005 to 0.107 ng/ml which, considering a mean molecular weight of 250 g/mol, represents 12-252x109 molecules per ml. All but one urine samples were positive for ≥ one NN. A statistically significant linear relationship was found between plasma/urine and CSF N-desmethyl-acetamiprid concentrations.

Conclusions

We have developed a reliable analytical method that revealed multiple NN and/or their metabolites in children's CSF, plasma, and urine. Our data suggest that contamination by multiple NN is not only an environmental hazard for non-target insects such as bees but also potentially for children.

OC 5

Pain concepts of Swiss Pediatricians: A qualitative study

Koechlin Helen[1,2], Locher Cosima[3,4], Carlander Maria[5], Kossowsky Joe[2], Woerner Andreas[7,9], Dratva Julia[6,8]

[1]Division of Psychosomatics and Psychiatry, University Children's Hospital Zurich, Switzerland; [2]Department of Anesthesiology, Critical Care and Pain Medicine, Boston Children's Hospital, Harvard Medical School, Boston, MA, USA; [3]Faculty of Health, University of Plymouth, UK; [4]Department of Consultation-Liaison Psychiatry and Psychosomatic Medicine, University Hospital Zurich, Switzerland; [5]UiT the Arctic University of Norway, Institute of Community Medicine, Tromso, Norway; [6]Zurich University of Applied Sciences, Institute of Health Economics, Winterthur, Switzerland; [7]Pediatric Rheumatoloty, University Children's Hospital Basel, Switzerland; [8]Zurich University of Applied Sciences, Institute of Health Sciences, Winterthur, Switzerland; [9]Medical Faculty, University of Basel, Switzerland

Chronic pain is a frequent complaint in children and adolescents. Pediatricians are usually the first line contact for children and adolescents with pain problems. However, in a recent Swiss survey, a large majority of pediatricians reported a lack of education and experience as well as not feeling comfortable treating children and adolescents with chronic pain. This study explored current experience of Swiss pediatricians to gain a better understanding of their pain concepts.

In a web-based survey sent to 1595 currently working pediatricians registered with the Swiss Society of Pediatrics, we collected demographic and professional characteristics of the sample and presented them with a case vignette. The vignette reported the (fictive) case of a 14-year old girl in good general health with musculoskeletal pain in the lower and upper extremities, with no evidence of harm found by an orthopedic surgeon and a pediatric rheumatologist. We then asked two open-ended questions about the case vignette: "What do you think is the etiology of the pain?" and "Please describe how you explain the cause of the pain to the parent and their child". Data was coded and analyzed by means of structuring content analysis, with the aim to subgroup categories of data into broader themes.

A total of 233 pediatricians (69% female, 31% male, mostly between 36-55 years of age) completed the survey and were included in this analysis.

Responses differed in form and length. Data showed that pediatricians explained the etiology of the girl's pain by biological factors, psychological factors, social context, multifactorial, disorder specific and unclear. Psychological factors had the highest number of quotations (i.e., 175), followed by biological factors (73 quotations), disorder specific (53 quotations), social context (21 quotations), and multifactorial (10 quotations). With regard to how participants would explain the pain to the family, a different order emerged: biological factors were mentioned most often (134 quotations), followed by psychological factors (106 quotations), and multifactorial (13 quotations).

Given the complex and biopsychosocial nature of pediatric chronic pain, debunking the misleading dichotomy of "biological" and "psychological" seems important. A clear explanation and understanding of the biopsychosocial model of chronic pain enables patients and families to potentially reconceptualize their pain, resulting in improved outcomes of multimodal interventions.

OC 6

Pediatric emergency room training: A novel, low-threshold approach

Nussberger E[1], Haberstich P[1], Semprebon M[1], Strebel R[1], Köhler H[1]

[1]Pediatric Emergency Division, Children's Hospital of Aarau, Switzerland

Background

Working as a physician or a nurse in a pediatric emergency room (ER) requires to be prepared to care for a seriously ill or injured child. ER training courses are often not easily accessible, expensive, and focus on complex clinical situations staged in an unfamiliar, technically demanding setting. Therefore, participants can easily feel overwhelmed, personally exposed, and subsequently might lack self-confidence in critical situations. We therefore sought to develop a more accessible approach also focusing on a positive training environment.

Methods

A senior emergency physician and an emergency nurse developed a novel local ER training approach. Since March 2020, all our pediatric residents and emergency care nurses were trained in groups of two, without any observers, in a supportive atmosphere and in our own ER. The training was split in small units and included the primary survey, the use of ER tools, and 22 selected pediatric and pediatric surgery ER scenarios, one of the trainees simulating a patient. Every participant also received a 28 page ER-knowledge folder, and had access to 15 self-designed card games to train the content on different occasions in a playful fashion. We evaluated our approach using standardized questionnaires.

Results

By the end of 2020, after 195 hours of training, 45 residents participated in the survey. All (100%) participants found the ER trainings delightful, 44 (98%) wanted to continue this kind of training. After the training the participants competence and quality of work self-assessment (1 = lowest grade, 6 = highest grade) in the pediatric ER settings improved from 3.3 to 4.6 and in surgical ER settings from 3.3 to 4.5. Eighty-four percent reported a major, and 16% a moderate benefit of the training concept in the daily work. By the end of 2021, after another 180 hours of training the overall rating of the training concept was 5.7. In a sub-analysis of 16 new residents, 15 (94%) reported to be less scared in ER situations because of the training.

Conclusion

Our approach to pediatric ER training is highly appreciated by our trainees, improving the sense of competence and the quality of work without the use of an expensive simulation equipment. The key seems to be a time intensive approach, caring for every single trainee, simple training units in the real-life setting, and we believe, above all, enthusiastic teaching in a delightful setting.

OC 7

Pediatric Tularemia in Central Europe - a Case Series

Schöbi N[1], Agyeman PKA[1], Duppenthaler A[1], Bartenstein A[2], Keller PM[3], Suter-Rhiniker F[3], Schmidt KM[4], Aebi C[1]

[1]Division of Pediatric Infectious Disease, Department of Pediatrics, Bern University Hospital, Inselspital, University of Bern, Switzerland; [2]Department of Pediatric Surgery, Bern University Hospital, Inselspital, University of Bern, Switzerland; [3]Institute for Infectious Diseases, University of Bern, Switzerland; [4]Swiss National Reference Center for Highly Pathogenic Bacteria, Laboratory Spiez, Switzerland

Background

Human tularemia, caused by Francisella tularensis, in Central Europe mainly transmitted by ticks, is an emerging disease. In children, the disease mostly manifests as glandular or ulceroglandular disease. To our knowledge, we describe the first larger case series on pediatric disease in Central Europe.

Methods

Retrospective single-center case series of patients <16 years of age diagnosed with tularemia at the University Children's Hospital Bern between January 1, 2010, and December 31, 2021. Case identification followed searching our electronic clinical microbiology database for specimens testing positive for F. tularensis by serology, culture, or PCR test.

Results

20 cases were identified with 55 % occurring between May and July. The time elapsed from day 1 of symptoms attributed to tularemia to both diagnostic and therapeutic intervention were 10 and 12 days, respectively. 14 patients presented with unilateral ulceroglandular compared to 5 with glandular disease (1 case of oropharyngeal tularemia). All entry site lesions showed multiple distinct vesicles, ulcers, or eschars. Affected lymph nodes were located on the head, neck, inguinal, and axillary areas. Systemic symptoms included fever during the first days of illness in 15 of 20 patients, which resolved in all patients before the initiation of targeted antimicrobial therapy against F. tularensis, i.e., ciprofloxacin (n = 15) or doxycycline (n = 5). Surgery was performed at a median of 13 days (range, -1 to 43) after starting targeted therapy. No patient was given additional antimicrobial therapy after the first targeted treatment and no secondary surgical intervention was performed.

Discussion

Causes for increasing case counts may include enhanced awareness, improved performance of diagnostic techniques, changes in leisure behavior, and a truly expanding animal reservoir. Reappraisal of the empirical management of pediatric acute lymphadenitis needs to be considered, as the currently used first-line treatment (betalactam antibiotics) for acute infectous lymphadenopathy is ineffective against F. tularensis and therefore resulting in therapeutic delays. Furthermore, this might cause an unsatisfactory outcome. The diagnostic process could be expedited by several approaches, e.g., increased awareness, not misinterpreting the resolution of influenza-like symptoms as treatment effect, performing a thorough search for scalp lesions, and performing serological diagnostics in suspected cases.

OC 8

New insights into acute hemorrhagic edema of young children

Bronz Gabriel[1], Rinoldi Pietro O.[1], Milani Gregorio P. [2, 3], Lava Sebastiano A. G.[4, 5], Kottanattu Lisa[1], Vanoni Federica[1], Zgraggen Lorenzo[1], Ferrarini Alessandra[6], Bianchetti Mario G.[7, 8]

[1]Pediatric Institute of Southern Switzerland, Ente Ospedaliero Cantonale, Bellinzona, Switzerland; [2]Pediatric Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy; [3]Department of Clinical Sciences and Community Health, Università degli Studi di Milano, Milan, Italy; [4]Pediatric Cardiology Unit, Department of Pediatrics, Centre Hospitalier Universitaire Vaudois and University of Lausanne, Lausanne, Switzerland; [5]Heart Failure and Transplantation, Department of Paediatric Cardiology, Great Ormond Street Hospital, London, United Kingdom; [6]Service of Medical Genetics, Ospedale Italiano di Lugano, Lugano, Switzerland; [7]Faculty of Biomedical Sciences, Università della Svizzera Italiana, Lugano, Switzerland; [8]Family medicine Institute, Faculty of Biomedical Sciences, Università della Svizzera Italiana, Lugano, Switzerland

Background

acute hemorrhagic edema of young children is a rare and poorly characterized vasculitis, which is sometimes considered the infantile variant of Henoch-Schönlein purpura. The majority of cases have been reported as case report or in small cases series. To provide more insight into the precursors, the clinical presentation, and the course of this condition, we created in 2019 the Acute Hemorrhagic Edema Bibliographic Database. The database included both the official and the grey literature on this vasculitis after 1969. The database currently contained 318 original reports, which documented 508 patients (351 males and 157 females, 11 [8-18] months of age, median and interquartile range).

Results

most cases were preceded by an infection (86%), by a vaccination (4%), or both an infection and a vaccination (4%). Annular (targetoid and not targetoid) or nummular purpuric eruptions and inflammatory skin edema characterized this condition. The following regions were affected, in decreas-ing order, by annular or nummular eruptions: legs, feet, face, arms, ears, trunk, and genitals. Apart from feet, which were very often affected, the same distribution was reported for edema. The initial eruption was often a wheal or a macule that evolved into a nummular or an annular eruption. In addition to the mentioned eruptions, the Köbner phenomenon (9.4%) and the Pastia sign (17.4%) were often observed. In contrast to Henoch-Schönlein purpura, the most common vasculitis in childhood, an extracutaneous (abdominal, articular or renal) involvement was rare and occurred in only 15% of cases. The disease spontaneously remitted within 3 weeks in 503 cases. The diagnosis of this vasculitis was supported by a biopsy in 50% of patients. Contrary to classical Henoch-Schönlein purpura, vascular immunoglobulin A deposits were observed in no more than one third of cases. Often, this vasculitis is confused with systemic bacterial infection, such as meningococcal sepsis, causing overuse of antibiotics and invasive diagnostic exams.

Conclusion

The creation of our database and the subsequent data analysis provided relevant new insights into this rather neglected vasculitis and confirmed the excellent prognosis of this benign skin-limited small vessel vasculitis: it does not need any therapies, normally it resolves spontaneously within 3 weeks without recurrences, and it is skin limited in about 85% of cases. These data may help physicians in rapidly making a clinical diagnosis.

OC 9

NEODOL intervention to improve interprofessional collaboration and involvement of parents in the management of painful procedures in newborns

Balice-Bourgois C[1], Zumstein-Shaha M[2], Simonetti GD[1, 4], Newman CJ[3]

[1]Pediatric Institute of Southern Switzerland, Ente Ospedaliero Cantonale, Bellinzona, Switzerland; [2]Department of Health, Bern University of Applied Sciences, Bern, Switzerland; [3]Woman-mother-child department, Lausanne University Hospital, Lausanne, Switzerland; [4]Faculty of Biomedical Sciences, University of Southern Switzerland, Lugano, Switzerland

Introduction

Hospitalized newborns are subject to many painful procedures. Pharmacological and non-pharmacological approaches alone are not enough, and it is necessary to consider other contributing elements such as interprofessional collaboration and parental involvement and the environment.

Aim of study: To explore interprofessionality and the role of parents to improve the management of painful procedures in newborns in relation to the implementation of NEODOL.

Methods

A pre-post feasibility study with a mixed method approach was used involving questionnaires, interviews and focus groups with the interprofessional team working and parents. Quantitative data was descriptively analysed. The qualitative data was treated with thematic analysis. This study was conducted in a level IIb neonatology unit in Switzerland.

Results

Interprofessional collaboration between physicians and nurses has improved as a result of NEODOL© implementation, a complex interprofessional intervention involving professionals, parents, and newborns. Nevertheless, parents remained in a passive role or were only marginally involved in the pain management of their infant. Parents stated that they wished for more information and inclusion in painful procedures performed on their infants.

Discussion

Management of painful procedures in neonates needs to change. Interprofessional collaboration contributes to improved procedural pain management in neonates. It is essential to include parents as active members in the interprofessional healthcare team. This research has implications for interprofessional education, practice, parent engagement, and research.

Relevance for research and practice

Pain management is an important issue in neonatology and it is necessary implement an interprofessional approach that involves all team members, including parents.

Parents want to be better informed about their baby's care and to be involved in the management of neonatal pain.

Partnerships between health care professionals, parents of infants, researchers and leaders are essential to improve the management of painful procedures.

OC 10

Comprehensive targeting of tumor metabolisms in pediatric high-grade gliomas

Petrovic A[1], Prasad R[1], Kritzer B[1], Laternser S[1], Stücklin A[1], Mueller S[1], Nazarian J[1]

[1]Department of Oncology, Children's Research Center, University Children's Hospital Zurich, Zurich, Switzerland

Pediatric diffuse midline gliomas (DMGs), including diffuse intrinsic pontine gliomas (DIPGs) are incurable childhood cancers. Less than 2% of patients survive 5 years after diagnosis. Over 80% of DMGs harbor histone mutations (H3K27M) and exhibits upregulated PI3K/mTOR pathway, increased aerobic glycolysis and energy production, contributing to oncogenic growth and chemoresistance. In order to target tumor metabolism, we have focused on systematic and effective co-targeting of mitochondrial (OXPHOS) and glycolysis pathways.

To target mitochondrial OXPHOs, we identified CNS-penetrant imipridones ONC201 and ONC206 as two promising therapies for children diagnosed with DMG. ONC201 has shown early clinical efficacy in a subset of DMGs by a near doubling of progression-free survival and sustained (>2 years) radiographic responses. We found that ONC201/6 functions by agonizing mitochondrial protease ClpP, which results in increased reactive oxygen species (ROS), OXPHOS impairment, and activation of integrated stress response. Our data enabled initiation of two clinical trials PNOC022 (ONC201), and PNOC023 (ONC206).

However, following OXPHOS impairment, DMG metabolism shifts to anaerobic glycolysis through ROS-induced PI3K/mTOR activation. Our preliminary data indicate that transcription factors FOXO1/3A play an important role in PI3K signaling. We hypothesize that effective inhibition of PI3K results in reduced FOXO phosphorylation and increased nuclear translocation. This in turn will result in reduced glycolysis, increased stress response and apoptosis.

As a proof of principal, we found that effective targeting of PI3K/mTOR using paxalisib in combination with ONC201/6 is synergistic, resulting in significant metabolic distress in DMGs. However, responses vary depending on model systems. To address this, we screened more than 300 clinically relevant/FDA approved drugs in four human DMG cell lines. We identified new PI3K inhibitors as potential combination therapy with ONC201/6. Ongoing in vivo studies are underway to further validate combinatorial synergy of our newly identified drugs

SWISSPEDNET

SPN 1

Systematic review of accelerated long-term forgetting in children and adolescents with neuropediatric diseases

Stähli Nadine [1], Bigi Sandra[1, 2, 3], Grunt Sebastian [1], Lidzba Karen[1], Studer Martina[1, 2]

[1]Division of Child Neurology, Development & Rehabilitation, University Children's Hospital, Inselspital, Bern University Hospital, and University of Bern, Switzerland; [2]Department of Neurology, Inselspital, Bern University Hospital, and University of Bern, Switzerland; [3]Institute of Social and Preventive Medicine, University of Bern, Bern, Switzerland

Background and objectives

Accelerated long-term forgetting (ALF) describes the phenomenon of normal learning and memory performance after short delays but greater forgetting after longer delays. First described in the 1990s, the topic is attracting increasing interest, particularly in adults with epilepsy. Studies investigating pediatric ALF remain rare and no systematic review has been published so far. Therefore, the aim of this systematic review was to assess published data on ALF in children and adolescents with neurological diseases.

Methods

A systematic literature search in Embase (via Ovid), APA PsycInfo (via Ovid), and Ovid MEDLINE® All was performed on 12/07/2020. Google Scholar, Web of Science, ProQuest and the World Health Organization International Clinical Trials Registry Platform were also consulted. After deduplication, we screened 601 articles. We included studies investigating episodic memory performance immediately, after 30 minutes, and after delays of 24 hours or longer. Studies investigating ALF in adults or with less time-points of memory assessment were excluded.

Results

Twelve studies were included, which discussed ALF in the following contexts: epilepsy (n = 9), traumatic brain injury (n = 1), 22q11.2 deletion syndrome (n = 1), and autism spectrum disorders (ASD, n = 1). All 12 studies investigated verbal memory performance and 5 of them tested visual memory as well. Evidence for ALF was more frequently found in verbal (8 studies) than in visual memory (1 study). In children with ASD, ALF was not significant. Furthermore, ALF was associated with earlier age of epilepsy manifestation and subcortical abnormalities as well as with problems in behavior and executive functioning.

Discussion

To date, only a handful of studies investigated ALF in children and adolescents with neuropediatric diseases. Although most studies have been conducted in children with epilepsy, there is first evidence of ALF in children after traumatic brain injury and 22q11.2 deletion syndrome, replicating the finding of the adult literature that ALF is not an epilepsy-specific disorder. Given that ALF as clinically relevant memory problem is missed using standardized memory assessments, we propose to add a delayed memory recall time-point in future studies to understand memory consolidation over time.

SPN 2

Paediatric transgender care from an endocrinologic point of view: experience over the past 9 years in a tertiary swiss center

Mazzi Sara[1], Nussbaum Marie-Lou[2], Wüthrich Christian[2], Flück Christa E.[1]

[1]Division of Pediatric Endocrinology, Diabetology and Metabolism, Bern University Hospital, University of Bern, Switzerland; [2]Division of Pediatric Mental Health Care, Department of Pediatrics, Bern University Hospital, University of Bern, Switzerland

Over the past 9 years, 57 children and adolescents with suspected gender dysphoria were referred to our multidisciplinary team in Inselspital Bern. Of these, 45 had a diagnosis of transgender, and among these 28 received a hormonal therapy in our center and were included in the study. There were no major side effects of the hormonal therapies and no reported pathological fractures. There were two regrets after short course of step 1 hormonal therapy; none of them reported irreversible physical changed. The data about bone health and fertility preservation were mostly lacking, partially due to the retrospective setting. However, the scarcity of these data stresses a crucial issue of the adolescent transgender medicine, which needs to be standardized and improved. Over all, we found a significant increase of the number of referred subjects with a suspected gender-dysphoria over the years (coeff. 1.5, $p < 0.001$), but not of subjects qualifying for the diagnosis transgender which were hormonal-treated (coeff. 0.55, $p = 0.15$). This result diverges from the literature and underscores the importance of a multidisciplinary approach for the care of pediatric and adolescent individuals with gender-dysphoria and the essential role of a professional assessment of the precise diagnosis before engaging in endocrine workup and hormonal treatments.

SPN 3

The Swiss Paediatric Inflammatory Brain Disease Cohort Study: Setting up a national registry for children and adolescents with paediatric onset MS and related disorders

Hulliger L[1], Bauder F[3], Capone Mori A[4], Dill P[5], Garcia-Tarodo S[6], Goeggel B[7], Hackenberg A[8], Kalser J[9], Kuehni C[1], Maier O[10], Schmid R[11], Strozzi S[12], Bigi S[1,2]

[1]Institute of Social and Preventive Medicine, University of Bern, Bern, Switzerland; [2]Department of Child Neurology, University Hospital Bern, University of Bern, Bern, Switzerland; [3]Department of Child Neurology, Luzerner Kantonsspital, Kinderspital Luzern, Luzern, Switzerland; [4]Department of Child Neurology, Kantonsspital Aarau, Clinic for Children and Adolescents, Aarau, Switzerland; [5]Department of Child Neurology, University Children's Hospital Basel, University of Basel, Basel, Switzerland; [6]Department of Child Neurology, University Hospitals of Geneva (HUG), University of Geneva, Geneva, Switzerland; [7]Department of Child Neurology, Paediatric Institute of Southern Switzerland, Ospedale San Giovanni, Bellinzona, Switzerland; [8]Department of Child Neurology, University Children's Hospital of Zurich, University of Zurich, Zurich, Switzerland; [9]Department of Child Neurology, University Children's Hospital Lausanne (CHUV), Lausanne, Switzerland; [10]Department of Child Neurology, Children's Hospital of Eastern Switzerland, St Gallen, Switzerland; [11]Department of Child Neurology, Kantonsspital Winterthur, Social Paediatric Centre, Winterthur, Switzerland; [12]Division of Neuropaediatrics and Neuropsychology, Kantonsspital Graubünden, Department of Child and Adolescent Medicine, Chur, Switzerland

Objectives

Paediatric onset multiple sclerosis (POMS) is a severe disease affecting children and adolescents in a period of essential brain development. Timely diagnosis and treatment initiation minimize neurological sequelae and improve patient outcomes. However, the diagnosis of POMS can be challenging, especially in young children. A systematic approach to the assessment of POMS patients versus patients with other inflammatory brain diseases (IBrainDs) will allow faster and more reliable diagnosis. A national registry will improve our understanding of POMS epidemiology, clinical presentation, and management.

Methods

Multicentre cohort study including prospective and retrospective data. Inclusion criteria: patients with POMS or another specified IBrainD with an onset before 18 years of age. Exclusion criteria: patients with 1) infectious diseases of the CNS; 2) genetic/metabolic causes of central demyelinating diseases; 3) neurological symptoms due to Guillain-Barré-Syndrome. Demographic and medical data are centrally collected.

Results

After the ethics committee approval at the end of 2020, all 11 participating centres have been initiated. We identified 195 potential participants with an IBrainD. Of those, 55 patients and/or families have consented to participate in the registry.

So far, we have collected the minimal dataset of 129 patients. their paediatric IBrainD diagnoses are distributed as follows: 60 (46%) POMS, 15 (12%) acute disseminated encephalomyelitis, 13 (10%) optic neuritis, 11

(9%) transverse myelitis, 9 (7%) different diagnoses (<5 patients per diagnosis), 8 (6%) antibody associated autoimmune encephalitis. The conclusive diagnoses of 13 (10%) patients are pending.

The identification of patients who do not regularly attend clinical follow-ups remains challenging due to lacking systematic and unified coding approaches.

Conclusion

The national registry will answer pressing questions about the epidemiology and clinical phenotypes of POMS and related diseases in Switzerland. It also offers the opportunity to assess treatment and outcomes of paediatric IBrainD patients in a longitudinal fashion. Furthermore, the registry facilitates the national and international collaboration by providing a research platform.

SPN 4

Best treatment option(s) for children and adolescents with migraine: A network meta-analytic approach

Locher Cosima[1], Koechlin Helen[2,3], Kossowsky Joe[3], Meissner Karin[4], Linde Klaus[5], Gaab Jens[6], Berde Charles B. [3]

[1]Department of Consultation-Liaison Psychiatry and Psychosomatic Medicine, University Hospital Zurich; [2]University Children's Hospital Zurich; [3]Department of Anesthesiology, Critical Care and Pain Medicine, Boston Children's Hospital, Harvard Medical School, Harvard University, Boston; [4]Institute of Medical Psychology, LMU Munich, Munich, Germany; [5]Institute of General Practice and Health Services Research, School of Medicine, Technical University Munich, Munich, Germany; [6]Division of Clinical Psychology and Psychotherapy, University of Basel

Background

Migraines are common in children and adolescents, with prevalence rates that increase with age. Migraine is a leading cause of disability across all age groups, and a major reason for outpatient and emergency department visits. A range of interventions exist for children and adolescents with migraine, both of pharmacological and non-pharmacological nature. Those interventions have been compared in pairwise meta-analyses in the past. However, interventions need to be systematically compared and ranked relative to each other which is possible with a network meta-analytic approach. In this talk/poster, I will present the results of two network meta-analyses (NMAs) that assessed pharmacological and non-pharmacological interventions for pediatric migraine.

Methods

We aimed to examine the efficacy and safety of pharmacological and non-pharmacological interventions in comparison to the control groups. We conducted a systematic review of studies in Medline, Cochrane, Embase, and PsycINFO. Randomized clinical trials in children and adolescents diagnosed as having episodic migraine were included.

Results

In our first analysis of pharmacological prophylactic treatments, we included 23 studies with a total of 2217 patients. Prophylactic pharmacological treatments included antiepileptics, antidepressants, calcium channel blockers, antihypertensive agents, and food supplement. In the short term (<5 months), propranolol (standard mean difference, 0.60; 95% CI, 0.03-1.17) and topiramate (standard mean difference, 0.59; 95% CI, 0.03-1.15) were significantly more effective than placebo. No significant long-term effects were found. In our second analysis of non-pharmacological treatment, we included 12 studies with a total of 576 patients. Non-pharmacological interventions were for example biofeedback, relaxation, and psychological treatments. Our findings revealed that components of non-pharmacological interventions are effective in treating pediatric migraine.

Discussion

There is only little evidence supporting efficacy in pediatric migraine for prophylactic pharmacologic treatments. Components of non-pharmacological interventions reveal meaningful effects. However, the results have to be interpreted carefully because they are based on small studies. Future researchers should identify factors associated with individual responses in large, multicentered studies.

SPN 5

Impact of the COVID-19 pandemic on children's and adolescents' quality of life and mental health

Richard Viviane[1], Dumont Roxane[1], Lorthe Elsa[1], Loizeau Andrea[1], Baysson Hélène[1,2], Barbe Rémy P.[3], Posfay-Barbe Klara[4], Guessous Idris[2,5], Stringhini Silvia[1,2,6], SEROCoV-KIDS group

[1]Unit of Population Epidemiology, Division of Primary Care Medicine, Geneva University Hospitals, Geneva, Switzerland; [2]Department of Health and Community Medicine, Faculty of Medicine, University of Geneva, Geneva, Switzerland; [3]Division of Child and Adolescent Psychiatry, Department of Woman, Child, and Adolescent Medicine, Geneva University Hospitals, Switzerland; [4]Division of General Pediatrics, Department of Woman, Child, and Adolescent Medicine, Geneva University Hospitals, Switzerland; [5]Division and Department of Primary Care Medicine, Geneva University Hospitals, Geneva, Switzerland; [6]University Center for General Medicine and Public Health, University of Lausanne, Switzerland

Background

The COVID-19 pandemic profoundly affected the life of children and adolescents, with potential negative repercussions on their well-being. However, this impact may have differed depending on context, household or individual level characteristics. We aimed to 1) identify risk factors for having been heavily impacted by the COVID-19 pandemic 2) evaluate the association between experiencing a heavy pandemic impact and the health-related quality of life (HRQoL) and mental health status of children and adolescents.

Methods

We invited children of adult participants in a Geneva population-based cohort (Specchio-COVID19), and from a random sample drawn from state registries to participate in the SEROCoV-KIDS cohort study. Children and adolescents aged 2-17 years old were included between December 2021 and February 2022. Parents were asked about the impact of the pandemic on their child(ren) with the Coronavirus impact scale, which covers health, financial and social aspects at the child and household level; a score higher than one standard deviation above the mean was deemed as a heavy impact. They also reported about their child(ren)'s HRQoL with the PedsQLTM, mental health with the Strengths and Difficulties Questionnaire, and on socio-demographic characteristics. Risk factors for having been heavily impacted by the COVID-19 were estimated with generalized estimating equations, as was the association between having experienced a heavy pandemic impact and having a poor HRQoL or mental health.

Results

Out of 1051 participants, 133 (12.7%) had experienced a heavy COVID-19 impact, 164 (15.6%) had a poor HRQoL, and 112 (12.7%) a poor mental health. Older age (aOR = 1.06; 95%CI = 1.01-1.11), average to poor parent-child relationship (aOR = 5.34; 95%CI = 2.39-11.94), average to poor parent's mood (aOR = 2.77; 95%CI = 1.63-4.71) and average to poor household financial situation (aOR = 2.68; 95%CI = 1.51-4.76) were associated with having been heavily impacted by the pandemic, after adjustment for age and sex. Participants who had experienced a heavy COVID-19 impact were more likely to present a poor HRQoL (aOR = 2.60; 95%CI = 1.64-4.13) and a poor mental health (aOR = 3.75; 95%CI = 2.15-6.55) compared to those with an average impact.

Discussion

Older children and adolescents, living in unfavourable family and financial conditions seemed at higher risk for being heavily impacted by the pandemic, which in turn was strongly associated with a decreased HRQoL and mental health.

SPN 6

Similarities and differences in the neurodevelopmental outcome of children with congenital heart disease and children born very preterm at preschool age

Wehrle F[1,2,3], Bartal T[1], Adams M[4], Bassler D[4,5], Hagmann C[2,3,5], Kretschmar O[3,5,6], Natalucci G[4,5,7], Latal B[1,3,5]

[1]Child Development Center, University Children´s Hospital, Zurich, Zurich, Switzerland; [2]Department of Neonatology and Intensive Care, University Children's Hospital Zurich, Zurich, Switzerland; [3]Children's Research Center, University Children´s Hospital Zurich, Zurich, Switzerland; [4]Newborn Research, Department of Neonatology, University Hospital Zurich, Zurich, Switzerland; [5]University of Zurich, Zurich, Switzerland; [6]Department of Cardiology, University Children´s Hospital, Zurich, Switzerland; [7]Family Larsson-Rosenquist Center for Neurodevelopment, Growth and Nutrition of the Newborn, Department of Neonatology, University Hospital Zurich, Zurich, Switzerland

Objective

To describe the similarities and differences in the neurodevelopmental outcome of children with congenital heart disease (CHD) undergoing cardiopulmonary bypass surgery compared to children born very preterm (VPT) at preschool age.

Study design: IQ, motor abilities, behaviour, and therapy utilization were assessed in 155 children with CHD as part of a prospective, single-center longitudinal study, and in 254 children born VPT as part of a national follow-up register at the same center. Group differences were tested using independent t-tests and X2-tests. Equivalence testing was used to investigate similarities between the groups, with equivalence bounds defined as d = 0.30 for continuous variables and 0.05 for proportions.

Results

Learning disabilities (i.e., 70 ≤IQ <85) and intellectual impairments (i.e., IQ <70) occurred in 17.4% and 4.5% of children with CHD compared to 23.1% and 5.5% in children born VPT, respectively. Motor and behavioral functions were impaired in 57.0% and 15.3% of children with CHD compared to 37.8% and 11.5% of children born VPT, respectively. Children with CHD had poorer global motor abilities (d = -0.26) and poorer dynamic balance (d = -0.62) than children born VPT, and children born VPT had poorer fine motor abilities than children with CHD (d = 0.34, all p <.023). Static balance and peer problems were statistically similar between the groups (both p <.049). Therapies were less frequent in children with CHD compared to children born VPT (23.4% versus 41.0%, p <.001).

Conclusion

Children with CHD undergoing cardiopulmonary bypass surgery and children born VPT share an overall risk for neurodevelopmental impairments at preschool age. Despite this, children with CHD receive fewer therapies, indicating a lack of awareness of the neurodevelopmental burden these children face. Long-term evaluation programs should be established for all high-risk children surviving neonatal critical illness to ensure adequate follow-up care and initiation of therapies.

SPN 8

White matter microstructure and executive functions in congenital heart disease from childhood to adulthood: A pooled case-control study

Ehrler Melanie[1,2,3], Brugger Peter[4,5], Greutmann Matthias[6], Schlosser Ladina[7], Wehrle Flavia M[1,2,8], Liamlahi Rabia[1,2,3,5], Naef Nadja[1], Kretschmar Oliver[2,3,9], Tuura O'Gorman Ruth[2,10], Latal Beatrice[1,2,3]

[1]Child Development Center, University Children's Hospital Zurich, Switzerland; [2]Children's Research Center, University Children's Hospital Zurich, Switzerland; [3]University of Zurich, Switzerland; [4]Department of Psychiatry, University Hospital Zurich, Switzerland; [5]Rehabilitation Center Valens, Switzerland; [6]Department of Cardiology, University Heart Center, University Hospital Zurich, Switzerland; [7]Department of Neurology, University Hospital Zurich, Switzerland; [8]Department of Neonatology and Intensive Care, University Children's Hospital Zurich, Switzerland; [9]Department Pediatric Cardiology, Pediatric Heart Center, University Children's Hospital Zurich, Switzerland; [10]Center for MR Research, University Children's Hospital Zurich, Switzerland

Background

Patients with congenital heart disease (CHD) are at risk for brain alterations. It is unclear whether the extent of alterations changes with age, especially during adolescence when the white matter microstructure (WMM) undergoes intense maturation processes. We investigated differences in WMM between CHD patients and healthy controls across age and associations with executive functions in a pooled sample of children, adolescents and young adults.

Methods

In total, 78 CHD patients and 137 controls between 9 and 32 years of age underwent diffusion tensor imaging and an executive function test-battery. Mean fractional anisotropy (FA) was calculated for each major white matter tract. Linear regression tested age and group effects and their interaction predicting FA. Relative variable importance (RI) estimated the independent contribution of the following factor to the variability in executive function: FA of major white matter tracts, group, CHD complexity, maternal education, sex, and age.

Results

Mean FA of almost all tracts was lower in patients compared to controls (p between 0.057 and <0.001). There was no significant interaction indicating that the extent of WMM alteration in patients did not change with age (all p>0.074). Strongest predictors for executive functions were group (RI: 53%), CHD complexity (RI: 14%), and FA of the hippocampal cingulum (9%) and the corticospinal tract (RI: 9%).

Conclusion

WMM remains altered in CHD patients across age and is associated with executive functions. There is no evidence for improvement after extensive brain maturation throughout adolescence. CHD is a chronic disease with cerebral and neurocognitive impairments persisting into adulthood and, thus, this population requires continuous attention when growing older.

SPN 9

Cerebral palsy in Switzerland - insights from a national clinical registry and cohort study

Hunziker S[1], Dreher T[2,3], Fluss J[4], Grunt S[5], Juenemann S[6], Kuehni CE[1,7], Kuenzle C[8], Meyer-Heim A[9], Newman CJ[10], Ramelli G[11], Tscherter A[1]

[1]Institute of Social and Preventive Medicine, University of Bern, Bern, Switzerland; [2]Paediatric Orthopaedics and Traumatology, University Children's Hospital Zurich, Zurich, Switzerland; [3]Paediatric Orthopaedics, University Hospital Balgrist, Zurich, Switzerland; [4]Paediatric Neurology Unit, University Children's Hospital Geneva, Geneva, Switzerland; [5]Division of Neuropediatrics, Development and Rehabilitation, Department of Pediatrics, Inselspital Bern, University Hospital, University of Bern, Switzerland; [6]Division of Neuropaediatric and Developmental Medicine, University Children's Hospital of Basel (UKBB), University of Basel, Basel, Switzerland; [7]Children's University Hospital, Inselspital, University of Bern, Bern, Switzerland.; [8]Children's Hospital of Eastern Switzerland, St Gallen, Switzerland.; [9]Swiss Children's Rehab, University Children's Hospital Zurich, Affoltern am Albis, Switzerland; [10]Paediatric Neurology and Neurorehabilitation Unit, Lausanne University Hospital (CHUV), Lausanne, Switzerland; [11]Neuropaediatric Unit, Paediatric Institute of Southern Switzerland, Ospedale San Giovanni, Bellinzona, Switzerland

Background

Cerebral palsy (CP), a group of chronic disorders of movement and posture, is the most common cause of motor disability in childhood. People with CP have a wide range of comorbidities, such as epilepsy, problems of speech, hearing, or vision, cognitive dysfunction, behavioral disorders, and secondary musculoskeletal problems. National data on people with CP in Switzerland was missing. Therefore, an interdisciplinary group of CP specialists founded the Swiss Cerebral Palsy Registry (Swiss-CP-Reg) in 2017. It aims to optimize treatment and improve the health and quality of life of people living with CP.

Methods

Swiss-CP-Reg includes individuals with CP who were born, are treated, or live in Switzerland. Data is collected from medical records and reports and from questionnaires sent to participants (people with CP) and their families. Swiss-CP-Reg contains harmonized data on diagnosis, clinical presentation, comorbidities, therapies, and quality of life. It serves as a platform for research and communication.

Results

By December 31st, 2021, 632 people with CP enrolled in Swiss-CP-Reg. Most individuals were diagnosed with spastic CP (77%), followed by ataxia (12%) and dyskinetic CP (11%). Motor impairment was mostly mild (Gross Motor Function Classification System [GMFCS] level I and II, 60%), but 13% live with a moderate (GMFCS level III) and 27% with a severe impairment (GMFCS level IV and V). Most participants had normal or only slightly impaired manual ability (Manual Ability Classification System level I and II, 60%). Intelligence quotient (IQ) was not impaired in the majority (IQ≥70, 55%), but was correlated with motor function. Several nested research projects aim to understand specific aspects of CP, and to standardize and improve CP therapies.

Conclusion

Swiss-CP-Reg is a clinical registry and cohort study with long-term follow-up, that collects data from people with CP and their physicians to describe the epidemiology of CP in Switzerland and to answer clinical questions. It follows a participatory approach by collaborating closely with patient representatives and serves as a research and communication platform to facilitate research on CP and to promote exchange between professionals involved in CP care.

SPN 10

Prevalence of SARS-CoV-2 positivity in infants with bronchiolitis: a multicentre international study

Cozzi G[1], Cortellazzo Wiel L[2], Amaddeo A[1], Gatto A[3], Giangreco M[1], Klein-Kremer A[4], Bosis S[5], Silvagni D[6], Debbia C[7], Nanni L[8], Chiappa S[9], Minute M[10], Morabito G[11], Gortan A[12], Colombo M[13], MArchetti F[14], Garelli D[15], Piffer A[16], Cardinale F[17], Levy N[18], Curatola A[3], Gojsina B[19], Basu S[20], Barbi E[1, 2], Sovtic A[19]

[1]IRCCS Materno Infantile Burlo Garofolo, Trieste, IT; [2]University of Trieste, Trieste, IT; [3]Policlinico Universitario Agostino Gemelli, Roma, IT; [4]Hillel Yaffe Medical Center, Hadera, IL; [5]Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, IT; [6]Azienda Ospedaliera Universitaria Integrata Verona, Verona, IT; [7]Istituto Giannina Gaslini Istituto Pediatrico di Ricovero e Cura a Carattere Scientifico, Genova, IT; [8]Azienda Ospedaliero Universitaria Meyer, Firenze, IT; [9]Ospedale dei Bambini di Brescia, Brescia, IT; [10]Ospedale Regionale Ca Foncello Treviso, Treviso, IT; [11]Ospedale Santa Maria degli Angeli di Pordenone, Pordenone, IT; [12]Azienda sanitaria universitaria Friuli Centrale, Udine, IT; [13]ASST Sette Laghi, Varese, IT; [14]Ospedale Santa Maria delle Croci, Ravenna, IT; [15]Regina Margherita Children's Hospital, Turin, IT; [16]Regional Hospital of Bellinzona and Valli, Bellinzona, CH; [17]Ospedale Pediatrico Giovanni XXIII, Bari, IT; [18]Ruth Rappaport Children's Hospital, Haifa, IL; [19]Institute for Health Protection of Mother and Child of Serbia 'Dr Vukan Cupic', Beograd, RS; [20]Bradford Royal Infirmary, Bradford, UK

Background

Bronchiolitis is the primary acute respiratory tract infection in infants during the winter season. Since the beginning of the Covid-19 pandemic, a reduction in the number of bronchiolitis diagnoses has been registered.

Objective

The present study aimed to describe the incidence and clinical features of bronchiolitis during the 2020-2021 winter season in a large cohort of children in Europe and Israel, and to clarify the role of Severe Acute Respiratory Syndrome Coronavirus 2 (SARS-CoV-2).

Setting, Patients, Interventions

We conducted a multicentre observational cross-sectional study in 23 paediatric emergency departments in Europe and Israel. Clinical and demographic data about all the cases of infants diagnosed with bronchiolitis from 1 October 2020 to 30 April 2021 were collected. For each enrolled patient, diagnostic tests, treatments, and outcomes were reported.

Main outcome measures

The main outcome was the prevalence of SARS-CoV-2 positive bronchiolitis.

Results

Three hundred and fourteen infants received a diagnosis of bronchiolitis during the study period. Among 535 infants who tested positive for SARS-CoV-2, 16 (3%) had bronchiolitis. Median age, male sex predominance, weight, history of prematurity, and presence of comorbidities did not differ between the SARS-CoV-2 positive and negative groups. Rhinovirus was the most common involved pathogen, while Respiratory Syncytial Virus (RSV) was detected in one case. SARS-CoV-2 bronchiolitis had a mild clinical course, with one patient receiving oxygen supplementation and none requiring paediatric or neonatal intensive care unit admission.

Methods

During the Covid-19 pandemic, a marked decrease in the number of bronchiolitis diagnoses and the disappearance of the RSV winter epidemic were observed. SARS-CoV-2 related bronchiolitis was rare and mostly displayed a mild clinical course.

SPN 11

Adherence to growth hormone - determined by a consensual, center-based supply - associates with height outcomes in youth

Madhavarapu A[1], Zacharin M[2], Flück C[1,3], Saner C[1,3]

[1]Division of Pediatric Endocrinology, Diabetology and Metabolism, Department of Pediatrics, Inselspital, Bern University Hospital, University of Bern, Switzerland; [2]Royal Children`s Hospital and Murdoch Children`s Research Institute, Department of Endocrinology and Centre for Hormone Research, Parkville, Victoria, Australia; [3]Department of Biomedical Research, University of Bern, Bern, Switzerland

Objective

This study investigated the association between adherence to rhGH-therapy with gain in height standard deviation (SD) in reference to mid-parental height (MPH), from treatment start to the most recent visit (deltaHSD).

Background

Recombinant human growth hormone (rhGH) therapy is proven safe and effective for children with short stature. Its efficacy depends on underlying cause, younger age and height at therapy onset, expected height related to mid-parental height (MPH), duration of treatment and optimal adherence.

Design/Patients/Measurements: Single-center, retrospective data analysis in patients on rhGH between 2006-2020. Anthropometric data were normalized using national representative growth charts. Adherence was calculated as the ratio (%) of rhGH distributed / rhGH needed (reported by specialized nursing staff and treating endocrinologist). Linear interpolation was used to calculate annualized follow-up data. Multiple linear regression modelling was applied.

Results

A total of 125 patients received rhGH, of whom 100 had longitudinal data (>12 months) over a mean of 5.4 years (SD 2.9) available for the analysis. In the first year of treatment, the mean (SD) growth velocity (GV) was 9.4cm/y (2.5), followed by measures in the upper normal range in subsequent years. Adjusted for duration of treatment, sex and MPH, adherence (per 10% increment) was associated with deltaHSD (β-coefficient 0.23, 95% confidence interval 0.03-0.42, p-value 0.022).

Conclusion

In the setting of a centre-based distribution of rhGH, the assessment of adherence as the ratio of rhGH distributed/needed can help to identify those who respond poorly and may benefit from further support to optimize treatment efficacy.

SPN 12

Evidence for protein leverage in children and adolescents in PANIC - Physical Activity and Nutritional Intervention in Childhood

Saner C[1,2,3], Senior A[4], Janner M[1], Raubenheimer D[4], Simpson S[4], Lakka T[5]

[1]Division of Pediatric Endocrinology, Diabetology and Metabolism, Department of Pediatrics, Inselspital, Bern University Hospital, University of Bern, Bern, Switzerland; [2]Department of Biomedical Research, University of Bern, Bern, Switzerland; [3]Murdoch Children's Research Institute, The Royal Children's Hospital, Parkville, Victoria, Australia; [4]Charles Perkins Centre and School of Life & Environmental Science, University of Sydney, NSW, Australia; [5]Department of Clinical Physiology and Nuclear Medicine, Kuopio University Hospital, University of Eastern Finland, Kuopio, Finland

Background

It is hypothesized that the intake of proteins is more strictly regulated than that of carbohydrates or fats. Consequently, a diet low in the proportion of energy from proteins is associated with i) higher total energy intake (TEI) (i.e., protein leverage, PL) and ii) increased risk of weight gain (i.e., protein leverage hypothesis, PLH). Here we aim to test for PL and the PLH in healthy children and adolescents.

Methods

Analysis of data from a nationally representative cohort of children and adolescents (PANIC) in Kuopio, Finland, investigated at age 8, 10 and 16 years. Data included: TEI (cal), the proportional energy intake of proteins (%EP), fats (%EF) and carbohydrates (%EC), and total fibre based on 4-day nutrient records; % body fat (%BF) and lean mass (%LM) based on dual-energy X-ray absorptiometry; body mass index (BMI) z-score; waist circumference (WC); accelerometery-data on physical activity; and estimated total energy expenditure (TEE). Power-function analyses between %E macronutrients and TEI, and compositional data analysis between macronutrient compositions and TEI, TEE, BMI z-score, %BF, %LM and WC were performed, and results illustrated in right-angled mixture triangles (RMTs) for fully adjusted models.

Results

A total of 422 individuals aged 8 years were followed-up at 10 years (N = 387) and 16 years (N = 229). Mean (SD) %EP, %EC and %EF was 17.0 (2.5), 17.0 (2.6), 17.8 (3.8); 52.6 (5.1), 51.1 (5.2), 47.6 (7.2); 30.4 (5.0), 31.8 (5.3) and 34.6 (6.6). BMI z-scores were -0.19 (1.08), -0.13 (1.07) and -0.03 (0.99). In fully adjusted models, %EP was inversely associated with TEI at all 3 timepoints following power functions at strength of leverage L = -0.36, L = -0.26, L = -0.25 (p-values<0.001). Across all three timepoints, TEI was positively associated with TEE (Pearson`s r = 0.35, 0.36 and 0.49, all p <0.001), but not with BMI z-scores (Pearson`s r = 0.07, p = 0.18; 0.06, p = 0.26; and -0.07, p = 0.31). RMTs revealed positive associations between %EP with weight, height, BMI z-score, waist circumference, %BF, and inverse associations with %LM.

Conclusion

This is the first study to provide evidence consistent with protein leverage in a large cohort of healthy children and adolescents in fully adjusted models. In this cohort, increased energy intake on lower %EP diets was counterbalanced by increased energy expenditure and therefore did not translate into increased adiposity.

POSTERS

P 1

Prophylactic Use of Levosimendan in Preoperative Setting for Surgical Repair of Congenital Heart Disease in Children

Wannaz L[1], Boillat L[2], Perez MH[2], Di Bernardo S[1]

[1]Pediatric Cardiology, Women-Mother-Child Department, Lausanne University Hospital and University of Lausanne, Lausanne, Switzerland; [2]Pediatric Intensive Care Unit, Women-Mother-Child Department, Lausanne University Hospital and University of Lausanne, Lausanne, Switzerland

Introduction

Low cardiac output syndrome (LCOS) is a significant cause of morbidity and the leading cause of mortality after pediatric cardiac surgery. Levosimendan has been demonstrated safe and effective in paediatrics to treat LCOS, and it has been used for years in this setting. In identified high-risk patients, its prophylactic administration has probably major advantages. We aimed to review our local strategy with preoperative prophylactic Levosimendan infusion in high-risk children to minimize LCOS after heart surgery.

Methods

We used hemodynamic parameters as surrogates of cardiac output, which is difficult to measured accurately in small children. These parameters were retrospectively collected through an electronic patient survey system (Metavision5®) after extracorporeal circulation.

Results

Seventy-two children received Levosimendan before surgery in our pediatric intensive care unit between 2010-2019. As expected, most patients were newborns and infants with long-lasting open-heart surgeries. The median cardiopulmonary bypass time was 182 [137-234] minutes, and aortic clamping time was 95 [64-126] minutes. The postoperative hemodynamic parameters remained stable throughout the first 48 hours, so as the vasoactive-inotropic score and urine output. Patients seems to remained out of a LCOS state, with a median arterial lactate of 2.8 [1.9-3.8] mmol/L during the first 6 postoperative hours and then decreased progressively. The median venous-to-arterial CO2 difference was the highest at 10 [6-11] mmHg between the 6-12 hours post-surgery. Median arteriovenous difference in oxygen saturation was 30% [23-37] between 6-12 hours post-surgery and decreased gradually thereafter. Nine patients (13%) required extracorporeal membrane oxygenation. No patient required dialysis or hemofiltration. Mortality was 0%.

Conclusion

Preoperatively prophylactic administration of Levosimendan is effective and safe to prevent LCOS in selected high-risk children before congenital heart surgery.

P 2

Incomplete Kawasaki disease in infancy: difficult to diagnose, dangerous to miss

Vismara SA[1], Saurenmann T[1], Hormann S[1]

[1]Departement of Pediatrics, Kantonsspital Winterthur, Zürich, Switzerland

Objective

To highlight the importance of early diagnosis of Kawasaki disease (KD) in infants under the age of 1 year. Kawasaki disease is an acute vasculitis of childhood causing coronary artery (CA) aneurysms in 25% of untreated cases. Incomplete KD, where the diagnostic criteria of KD are not fully met, is increasingly diagnosed. In a recent publication, incomplete KD accounted for approximately 20% of new cases, and was associated with a higher risk of CA lesions.

Case

A 2-month-old boy was referred to our emergency room because of unexplained fever and rash. He presented with 3 clinical KD criteria: non purulent bilateral conjunctivitis, palmar-plantar erythema and maculo-papular rash. On bloodwork he had elevated CRP, hypoalbuminaemia, thrombocytosis and sterile leukocyturia. Incomplete KD was suspected and treatment with immunoglobulins, aspirin and corticosteroids was started on day 2 of fever. Echocardiography performed within 12 hours of diagnosis revealed aneurysms of the right and left CA, which confirmed the diagnosis. Fever resolved within 12 hours after initiation of treatment and the boy remained afebrile thereafter. Despite appropriate treatment CRP levels remained slightly elevated and echocardiography showed progression of the CA aneurysms, reaching a maximum diameter of 4mm (z +9). ECG showed no signs of myocardial ischemia. Due to the size of the aneurysm, anticoagulation was started and the patient transferred to a tertiary center where treatment with the IL-1 inhibitor anakinra was initiated. On day 26 after onset, echocardiography showed stable to minimally decreasing size of aneurysms, so anticoagulation was stopped and cortisone gradually decreased. Cardiologic follow up showed slow healing of the CA lesions and by age 5 month the aneurysms had fully disappeared.

Discussion

This case highlights both the importance and the difficulty of early diagnosis of incomplete KD in infants. Diagnosis is difficult because the younger the patient the more frequent the incomplete presentation and the differential diagnosis of febrile diseases in infancy is large. On the other hand, it is of utmost importance to suspect incomplete KD in a febrile infant because early treatment can prevent irreversible CA lesions and resulting morbidity. In the case presented here, we were lucky to suspect the diagnosis at only 2 days of fever because of the unusual rash and the highly inflammatory laboratory results.

P 3

Multisystem Inflammatory Syndrome in Children (MIS-C)- A Case Report with severe Cardiac Involvement

Link H[1], Wildbolz M[1], Knirsch W[2]

[1]Department of Pediatrics, Stadtspital Triemli, Zurich, Switzerland; [2]Department of Pediatric Cardiology, University Childrens Hospital Zurich, Switzerland

We present the case and course of a 10-year-old girl with a multisystem inflammatory syndrome in children (MIS-C) following SARS CoV-2 infection.

The previously healthy girl was referred to our emergency department due to persistent fever, reduced general condition, thrombocytopenia and elevated CRP. Clinical exanimation confirmed a reduced condition and revealed bilateral conjunctivitis, palmar, facial and thoracal erythema, sinus tachycardia and fever. Diagnostics, including a negative SARS-CoV-2 test, showed pancytopenia, elevated liver enzymes and CRP. Initial echocardiography and ECG were normal. Based on history, clinical presentation and laboratory results reflecting a multisystem affection we considered sepsis, Kawasaki disease and as a result oft the SARS-CoV-2 pandemic MIS-C despite negative test. Treatment included antibiotics, intravenous immunoglobulins (IVIG) and systemic corticosteroids.

The condition worsened on the third day of hospitalization with clinical signs of heart failure due to new-onset myocarditis. Echocardiography showed a reduced biventricular function. There were repolarization abnormalities in ECG and elevated cardiac enzymes. The patient was then referred to ICU of a tertiary hospital for further treatment. Meanwhile SARS-CoV-2 infection was proven via positive antibodies. The girl was stabilized with high-flow therapy, catecholamines, diuretics, low-dose acetylsalicyl acid (ASA) and high-dose corticosteroids. Due to persistent fever, antibiotic therapy was extended and immune-modulating medication (Anakinra) was prescribed. Within a few days, cardiac function improved, cardiac enzymes normalized and there were no signs of coronary dilation.

After 6 days and a second course of IVIG, the patient was transferred from ICU to the hospital ward.

The patient was discharged in good general condition after 13 days of hospitalization with low dose ASA for 8 weeks and Prednisolon for 2 weeks in total.

Regular outpatient controls showed normalized cardiac function. After 6 months, a cardiac MRI was performed showing no signs of myocardial damage and normal coronary arteries. The patient was suffering from breathlessness and general weakness for several months. Six months after primary diagnosis of MIS-C she was allowed to participate in normal physical exercise again.

Summarized, pediatricians need to be aware of MIS-C as a serious condition whenever children present with persistent fever, rash and clinical signs of multiorgan affection.

P 4

Parental views on neurodevelopmental follow-up visits and early interventions in children after open-heart surgery during infancy

Ursprung Pascale[1,3], Knirsch Walter[2,3], Kretschmar Oliver[2,3], Latal Beatrice[1,3,4], von Rhein Michael[1,3]

[1]University Children's Hospital Zurich, Child Development Center, Zurich, Switzerland; [2]University Children's Hospital Zurich, Pediatric Cardiology, Pediatric Heart Center, Zurich, Switzerland; [3]University Children's Hospital Zurich, Children's Research Center, Zurich Switzerland; [4]University of Zurich, Switzerland

Background and aim

Congenital heart disease (CHD) is the most common birth defect affecting approximately 6-8:1`000 life born children. In Switzerland, developmental follow-up (FU) assessments at 1,2 and 5 years of age are recommended for children with CHD and open-heart surgery within the first year of life. We investigated the compliance, and parental views on FU and early interventions (EI).

Methods

Out of 201 children with CHD (born in 2015/2016) in whom cardiopulmonary bypass surgery was performed atour institution in the first year of life, 26 were excluded due to death (n = 21), and residence abroad (n = 5). 175 were contacted, and 129 answered a semi-structured telephone interview regarding the participation, timing, and place of their child`s developmental FU, and 47 additionally participated in an online-survey on parental views on FU and early interventions.

Results

Of the 129 participating children (51% male) 58% had a cyanotic CHD. In the telephone interview, parents reported, that 89 (69%) children received at least one FU assessment at a mean age of 18 months, with 79% receiving their first assessment during within the first two years of life. Of those children who received a FU, 48% showed a developmental delay in at least one domain (cognitive, language, motor) and for 58% at least one type of early intervention was initiated. In the online survey, 81% of parents rated the FU assessment very positive. Also, early interventions were highly appreciated. From the 25 children without FU, 16 parents were not aware of the necessity of a FU, 11 of them agreed to come to a FU subsequently.

Conclusions

The FU-rate in our study population was lower than we would have expected. Adaptions of our FU program were introduced, accordingly. Furthermore, the nationwide register (implemented in 2019) additionally aims at ensuring good FU rates. The parental satisfaction with the FU's and early interventions was high. However, some parents were not aware of the potential risk of developmental delays and the possibility of specific FU's. Ways to address the parents are to repeatedly include this information in routine consultations and by contacting them by phone.

P 5

Disregarded ambiguous external genitalia at birth - possible consequences of delayed diagnosis of a 46XY DSD difference of sex development with HSD3B2 mutation

l'Allemand Dagmar[1], Friedrich Christine[1], Eugster Florence[1], Goldberger Schlomit [1], Heldt Katrin[1], Flueck Christa[2], Schaller André[3]

[1]Pediatric Endocrinology and Diabetology, Children's Hospital of Eastern Switzerland, St. Gallen, Switzerland; [2]Pediatric Endocrinology and Diabetology, Department of Pediatrics and Clinical Research, University of Berne; [3]Clinics of Human Genetics, Inselspital, University of Berne, Switzerland

Introduction

Any child born with ambiguous genitalia needs immediate and specific interprofessional care, as recommended by international guideline (Cools M 2018) and the Swiss National Advisory Commission on Biomedical Ethics (NEK 2012).

Case report

A term-born girl of African / South American parents was referred by the pediatrician on the 10th day of life for ambiguous genitalia with normal 17-OH-Prog. newborn screening to our multiprofessional DSD team. At birth, the midwife and paediatrician had assigned a female sex. However, clinically the External Genital Score was 6.5/12 points with a 2 cm genital tubercle, perineal meatus urethrae and labioscrotal descended testes. Sonographically there were normally structured testes, no uterus, the karyotype was 46XY, and a specific steroid synthesis disorder was not detectable by serum immunoassays. Psychosocially, the family had a background of multiple strain; in addition, the mother experienced posttraumatic stress disorder and was skeptical about further diagnostics. In the course of the first 5 years the phallus grew to 4.5 cm, there was a premature adrenarche with non-familial tall stature and bone age advanced by 1.8 years, but the cognitive development was retarded. Gender reassignment is being carefully considered with the child and parents.

Results

On the 10th day of life, Anti-Müllerian hormone, testosterone, dihydrotestosterone, cortisol, ACTH and electrolytes were normal. After 4 months, HCG test showed decreased testosterone and increased DHEA and 17-hydroxy-pregneolone stimulation, but the GCMS urinary steroid metabolites showed no evidence of a steroid synthesis defect. At age 3, inhibin B was normal with 30.6 ng/ml, as was cortisol after ACTH stimulation, but baseline and stimulated DHEA(S) 10-fold increased. Molecular genetic analysis revealed pathogenic HSD3B2 mutations: in intron 3 a splice variant c.307+1G>A and in intron 4 a missense variant c.779C>T.

Discussion

This child has a specific form of 3-beta-hydroxysteroid dehydrogenase defect with slightly reduced androgen production in the testes and adrenal glands, but increasing virilisation due to peripheral testosterone production by the intact HSD3B1 isoenzyme. Affected adults can be sexually active and subfertile as men, so far without increased tumour risk. Postpartum appropriate care without immediate sex assignment but with full diagnostics up to molecular genetics could have avoided uncertainties and unnecessary diagnostics.

P 6

Singleton-Merten-Syndrome – an interdisciplinary approach from diagnosis to targeted therapy

Heldt K[1], Broser P[2]

[1]Department of pediatric endocrinology, Eastern Switzerland Children's Hospital, Sankt Gallen, Switzerland; [2]Department of pediatric neurology, Eastern Switzerland Children's Hospital, Sankt Gallen, Switzerland

Background

Singleton-Merten syndrome is rare type I interferonopathy caused by mutations in the IFIH1 gene. IFIH1 encodes the MDA5-receptor which senses

viral dsRNA and activates type I interferon signaling. A gain-of-function mutation of IFIH1 causes overactivation of type I interferon signaling leading to autoinflammation.

Presenting problem

The boy presented initially at the age of 3 years because of gross motor developmental delay, muscular weakness, short stature with bone age retardation of 1 ½ years and osteopenia. Endocrinological testing could exclude growth hormone deficiency. In the following years, he showed age-appropriate cognitive development, but short stature, muscular weakness and sarcopenia worsened. At the age of eight years he developed a severe treatment resistant psoriasis, dental dysplasia and x-ray showed expansion of the medullary cavities in the bones of the hand accompanied by severe osteoporosis in hands and feet and akroosteolysis. General osteoporosis could be excluded. Due to the clinical findings of neuromuscular, endocrinology and inflammatory symptoms we early chose an interdisciplinary approach. Since the different symptoms were highly suspicious of an overriding genetic disease, whole exome sequencing revealed a pathogenic de novo variant in the IFIH1 gene. Consistent with constitutive type I interferon activation, patient blood cells exhibited a strong IFN signature as shown by marked up-regulation of IFN-stimulated genes.

Clinical management

Based on genetic, clinical and laboratory findings, the decision was made to treat the child with Ruxolitinib, a Januskinase 1/2 inhibitor, which inhibits signaling at the IFN-α/β receptors. The patient responded with significant improvement within the first weeks. Thus, psoriatic lesions vanished within days, the muscle weakness and bone mineralization improved and the patient showed a significant weight gain. However, treatment period is too short to determine the expected effect on growth.

Clinical improvement was accompanied by a marked reduction of the interferon signature in blood, indicating that inhibition of overactive type I IFN signaling was effective.

Discussion

This case report highlights the potential of a patient-tailored therapeutic approach by silencing the metabolic pathway responsible for the clinical symptoms. He also emphasizes the importance of an interdisciplinary approach, especially in the presence of multisystem symptoms.

P 7

Not All That Shivers Is Epilepsy or How Can Eating a Sausage Cause Impaired Consciousness?

Stasinaki A[1], Heldt K[1], Hasselmann O[2], Schaller A[3], l'Allemand D[1]

[1]Pediatric Endocrinology and Diabetology, Children's Hospital of Eastern Switzerland, St.Gallen, Switzerland; [2]Center for Child Neurology, Development and Rehabilitation, Children's Hospital of Eastern Switzerland, St.Gallen, Switzerland; [3]Department of Human Genetics, Inselspital, Bern University Hospital, University of Berne, Switzerland

Introduction

Clarifying the causes of both hypoglycaemia and epilepsy is a major challenge in paediatrics.

Case report

A 2.7-year-old boy presented to the neurology department due to recurrent episodes of consciousness impairment and intermittent leg weakness without EEG abnormalities. After excluding epilepsy, hypoglyceamia was suspected as a possible cause.

The boy was born at term to non-consanguineous Swiss parents. Apart from borderline primordial short stature, personal history, clinical examination and psychomotor development were unremarkable. Continuous glucose monitoring revealed low mean glucose levels of 3.8 mmol/L with hypoglycaemic episodes (nighttime nadir of 2.2 mmol/L). Protein-rich meals, like sausages or salmon, seemed to provoke hypoglycemia. Nocturnal hypoglycaemia could be prevented by providing 3-4gr uncooked starch before sleep. Following a fasting test, severe hypoglycaemia of 1.5 mmol/L appeared after 21 hours of fasting. The critical sampling investigations revealed an inappropriately detectable insulin level of 3.5 mU/L.

Results

Genetic analysis revealed a de novo heterozygous gain of function GLUD1 variant c.820C>T (p.(Arg274Cys), rs56275071) compatible with Hyperinsulinism/Hyperammonemia Syndrome (HI/HA,1:200'000). GLUD1 encodes glutamate dehydrogenase (GDH), a mitochondrial enzyme catalyzing the deamination of glutamate to a-ketoglutarate and ammonia. Activating mutations in GDH lead to leucine-stimulated secretion of insulin from pancreatic b-cells as well as mild hyperammonemia (91 µmol/L in our patient).

The boy was started on oral diazoxide and diet was recommended with carbohydrates prior to any proteins in order to suppress leucine-induced insulin secretion. This treatment regime led to a better glucose profile without any further seizures. Diazoxide dose could rapidly be reduced with lessening of side effects (transient water retention, headaches, photosensitivity).

Discussion

This rare form of hyperinsulinism with hyperammonemia, is associated to neurodevelopmental problems in 70% of patients, most probably related to increased intracerebral breakdown of glutamate, an important neurotransmitter. Although HI/HA syndrome presents with milder hypoglycaemia and without macrosomia, diagnosis should not be delayed until late infancy in order to avoid negative effects on the developing brain. We recommend for any child with protein-induced or fasting hypoglycaemia, the investigation of ammonia to detect this rare disease.

P 8

Esophageal involvement in paediatric Crohn's disease

Ott A[1], Spalinger J[1,2], Righini-Grunder F[1,2]

[1]Department of Paediatrics, Children's Hospital Lucerne, Switzerland; [2]Division of Paediatric Gastroenterology, Hepatology and Nutrition, Children's Hospital Lucerne, Switzerland

Introduction

Paediatric Crohn's (CD) disease is a rare, inflammatory bowel disease characterized by severe, chronic inflammation of the intestinal wall in any part of the gastrointestinal tract. Involvement of the oesophagus is rare, making diagnosis more difficult. Since IBD can have far-reaching consequences, early diagnosis and initiation of appropriate therapy is important.

We present 2 adolescents with oesophageal involvement as a rare initial manifestation of CD.

Results

A 13-year-old boy presented with odynophagia, retrosternal pain between meals, oral lesions and involuntary weight loss. No abdominal pain or vomiting was reported. On endoscopy and histology, the diagnosis of CD with pronounced oesophageal involvement was confirmed. Induction therapy was started with steroids, followed by a maintenance treatment with methotrexate (MTX) with good clinical response. Within two months, the patient relapsed and a treatment with infliximab was started. Subsequent remission of disease could be achieved.

A 14-year-old girl presented with odynophagia, retrosternal pain for 4 weeks, subfebrile temperatures and involuntary weight loss. She showed oral lesions, perianal marisks and rectal bleeding. PPI treatment was unsuccessful, as it was vitamin C therapy (suspected, but not confirmed scurvy) and antimicrobial treatment (oral purulent erosions). After endoscopic and histological confirmation of the diagnosis of CD with pronounced oesophageal involvement, an exclusive enteral therapy with Modulen IBD® was started, followed by maintenance treatment with azathioprine (AZA). Because of a relapse on AZA, a switch to biologics early in the course of disease (infliximab, adalimumab) was necessary. Subsequent remission of disease could be achieved.

Conclusion

When odynophagia, retrosternal pain and involuntary weight loss occur in childhood and adolescence, CD should be considered as a differential diagnosis. In the absence of therapy response of first line treatment (AZA,

MTX) a switch to biologics such as infliximab or adalimumab should not be delayed.

P 9

A rare cause of vitamin B12 deficiency in infancy

Wenk Leyla[1], Gautschi Matthias[2], Sokollik Christiane[1], Schibli Susanne[1]

[1]Pediatric Gastroenterology, Hepatology, and Nutrition, Department of Pediatrics, Inselspital, University of Bern, Switzerland; [2]Paediatric Endocrinology, Diabetology and Metabolism, Department of Paediatrics, Inselspital, University of Bern, Switzerland

Introduction

Vitamin B12 deficiency is uncommon in infants as Vitamin B12 is widely available in animal foods and there are sufficient levels in breast milk. Deficiency is most often seen in healthy exclusively breastfed infants of B12-deficient mothers, in undernourished infants or in families following a vegan diet. It presents with nonspecific symptoms like weakness, failure to thrive and variable neurologic problems. Supplementation leads to resolution of the deficiency and associated symptoms. If left untreated, irreversible cognitive deficits may occur.

Case

A 1-year-old girl presented with failure to thrive and gross motor delay. Metabolic screening revealed elevated urinary methylmalonic acid leading to the diagnosis of vitamin B12 deficiency. A maternal B12 deficiency was excluded and the girl had a regular diet with sufficient amount of vitamin B12. The parenteral supplementation of vitamin B12 improved the neurological symptoms. However, failure to thrive and chronic diarrhea persisted. Further workup revealed exocrine pancreatic insufficiency. Neonatal screening for cystic fibrosis and sweat test were negative. Genetic testing confirmed suspected Shwachman-Diamond Syndrome. Under supplementation with pancreatic enzymes, vitamin B12 status remained sufficient without further need of vitamin B12 supplementation

Discussion

Exocrine pancreatic insufficiency is a rare, but acknowledged cause of severe vitamin B12 deficiency. The mechanism of vitamin B12 absorption is complex. The cleavage of haptocorrin by pancreatic proteases is a crucial step. This releases vitamin B12 and allows its binding to intrinsic factor, which is essential to its absorption in the terminal ileum. Since not all patients with exocrine pancreatic insufficiency develop vitamin B12 deficiency, there are other, still unknown mechanisms involved. Nevertheless, exocrine pancreatic insufficiency should be considered as a differential diagnosis in vitamin B12 deficiency, and vice versa, children with pancreatic insufficiency should be screened for vitamin B12 deficiency.

P 10

Severe symptomatic hypoglycemia under Trikafta® Therapy: adverse effect or coincidence?

Kunz S[1], Willi B[1], Steigert M[1], Casaulta C[1,2]

[1]Department of Pediatrics, Graubunden Central Hospital Chur, Switzerland; [2]Department of Pediatrics, University Children's Hospital Bern, Switzerland

Background

Trikafta® (Ivacaftor / Elexacaftor / Tezacaftor) is a new combined CFTR modulator and potentiator for the treatment of cystic fibrosis (CF), approved by Swissmedic since December 2020. It has shown to improve lung function in CF-patients. Trikafta® also influences pancreatic function and glucose homeostasis, whereby hypoglycemic episodes have been described. The underlying mechanisms are not yet fully understood.

Case report

We present a 16-year-old adolescent with CF with a first tonic-clonic self-limiting seizure while being hypoglycemic two weeks after therapy-onset with Trikafta®. Diagnostic work-up found no other underlying cause for the seizure-episode.

Discussion

Literature shows a complex influence of pancreatic CFTR channels on glucose homeostasis. CFTR channels have an important role in the first phase of insulin secretion as well as regulation of glucagon release. Hence, some effect on glucose homeostasis is expected from the treatment with CFTR modulators. Due to the timely connection in our patient, the hypoglycemic state could have been provoked by a combination of changing eating habits and the direct influence of Trikafta® on the regulatory mechanism of the pancreatic CFTR channels.

Conclusion

Medical caregivers should be aware of potential hypoglycemia risk in CF-patients put on Trikafta®. Based on the literature CF-pathophysiology and the pharmacologic effect of Trikafta® we assume that the medication may have triggered this critical event. We thus conclude that before initiating therapy, a) patients should be evaluated for prediabetic state; b) patients should be instructed regarding hypoglycemia-risk and c) the possibility of continuous-glucose-monitoring should be considered and discussed with the patients.

P 11

Evaluating the extent of pain in adolescents: preliminary results on test-retest reliability

Folli Anna[1,2], Falla Deborah[1], Cescon Corrado[2], Vanoni Federica[3,4], Soldini Emiliano[5], Barbero Marco[2]

[1]Centre of Precision Rehabilitation for Spinal Pain (CPR Spine), School of Sport, Exercise and Rehabilitation Sciences, College of Life and Environmental Sciences, University of Birmingham, Birmingham, UK; [2]Rehabilitation Research Laboratory 2rLab, Department of Business Economics, Health and Social Care, University of Applied Sciences and Arts of Southern Switzerland, Manno, Switzerland; [3]Clinic of pediatrics, Pediatric Institute of Southern Switzerland, EOC, Bellinzona, Switzerland; [4]Università della Svizzera Italiana, Lugano, Switzerland; [5]Competence Centre for Healthcare Practices and Policies, Department of Business Economics, Health and Social Care, University of Applied Sciences and Arts of Southern Switzerland, Manno, Switzerland

Background

A Pain Drawing (PD) is a self-administered pain assessment that requires the person who completes it to colour in on a body chart the areas in which he or she experiences pain, regardless of the intensity. PDs can be analysed by digitally extracting the coloured area and assessing the spatial spread of pain (pain extent) and localising it with respect to an anatomical grid (pain location). Clinicians can use these data to document the patient's pain history, to assess the evolution of a patient's symptoms or to evaluate their response to provocative tests.

Although body charts are included in several questionnaires for pain assessment of children and adolescents, no one has investigated the reliability of adolescents in reporting the extent and location of pain via a PD. Therefore, the aim of this study is to establish the test-retest reliability of adolescents in reporting the extent of their pain using a PD.

Methods

Adolescents with pain of musculoskeletal origin and aged 11-16 years were included. All participants were asked to colour the areas where they had had pain in the last week. After the administration of a questionnaire and the acquisition of further personal data (requiring at least 10 minutes), the PD was administered again. The PDs were then scanned and analysed using a digital platform, which allowed the extraction of pain extent values on which the test-retest reliability was calculated. Intraclass Correlation Coefficient (ICC2,1) and Bland Altman analysis was used to assess respectively relative and absolute test-retest reliability.

Results

Currently, 17 adolescents out of the 40 required by the sample size calculation were included. The analysis of the 17 included participants resulted in excellent ICC2,1: 0.99 (95% CI: 0.98- 1.00). The Bland Altman analysis showed a mean difference close to zero: -0.01 (limits of agreements -0.44 to 0.43).

Conclusions

Preliminary analysis of the data extracted from the first 17 participants of this ongoing study shows that adolescents with pain of musculoskeletal origin are reliable in reporting the extent of their pain with PDs.

P 12

Improving multidisciplinary case discussions in humanitarian settings: the MSF - HUG collaborative model

Ballester M[1], Chauvet Piat E[1], Salomon C[1], Jorgenssen C[1], Bottineau MC[2], Müller M[1], Vocat A[1], Alcoba G[3,4], Pinösch S[1], Peyraud N[1], Galetto-Lacour A[1], Martinez Garcia D[1,2], Wagner N[1]

[1]1Department of Women-Children-Teenagers, University Hospitals of Geneva, Geneva, Switzerland; [2]2 Women & Child Health & Nutrition Unit, Medical Department, Médecins Sans Frontières (MSF), Operational Centre Geneva, Geneva, Switzerland; [3]3 Division of Tropical and Humanitarian Medicine, University Hospitals of Geneva, Geneva, Switzerland; [4]4Tropical diseases and epidemics unit, Medical Department, Médecins Sans Frontières (MSF), Operational Centre Geneva, Geneva, Switzerland

Background and aims:

In humanitarian settings, access to specialists is limited or absent. The MSF telemedicine store and forward network provides a space for online discussion with different specialists. Complemented with specific planned sessions, real-time discussion can occur among all parties involved, facilitating the resolution of complex cases.

Method

Since 2018, case discussion sessions for paediatric cases have been established between MSF and the paediatric hospital at the University Hospitals of Geneva (HUG). Cases are sent in advance via the telemedicine platform to the fellows who collect information, contact relevant specialists and prepare the summary.

Results and discussion

Close to 40 sessions have been held. Most cases have been resolved positively for the patient despite their complexity and the lack of diagnostic/therapeutic means.

The focus of the sessions remains to improve the patient condition. Nonetheless, they allow senior and junior doctors and medical students to be confronted with new or rare medical situations. They learn skills to provide adapted medical advice for resource-limited and humanitarian settings. For some of them, it is a source of motivation to work in humanitarian contexts. From the project medical team's perspective: they become more efficient presenting and discussing clinical cases, which in turn helps their critical thinking during their clinical activities. A summary of recommendations is sent back to the project medical team after each discussion.

Due to COVID-19, the sessions take place virtually. This change has allowed much wider and easier access to MSF medical staff in HQ, and in particular the project medical teams, but also to specialists from all over the world.

Conclusion

This model allows for the focused management of complex cases and has multiple benefits for patients, project medical teams, specialists and medical trainees. This model could be replicated in other areas of the HUG for other types of patients depending on project needs and requests.

P 13

A new challenge: post-COVID syndrome in adolescents

Perrin A[1], Posfay-Barbe KM[2], Caflisch M[1]

[1]Adolescent clinic, Division of General Pediatrics, Department of Woman, Child, and Adolescent Medicine, Geneva University Hospitals, Geneva, Switzerland; [2]Division of General Pediatrics, Department of Woman, Child, and Adolescent Medicine, Geneva University Hospitals, Geneva, Switzerland

Post-COVID syndrome (or long COVID) is a set of multiple symptoms occurring after a documented SARS-CoV-2 infection and persisting for more than 2 months. The pediatric population is also affected, especially preadolescents and adolescents, even if data about this age group are still scarce. Persistent symptoms can have a strong impact on quality of life and schooling, school absenteeism and social withdrawal being of major concern. For this reason, the Division of General Pediatrics at the University Hospital of Geneva has set up in May 2021 a specific consultation for adolescents with post-COVID syndrome, offering global and multidisciplinary care.

To date 50 patients have been addressed to our consultation by their general practitioner. The mean age is 14 years, two thirds are girls. The symptoms are multiple and non-specific, and are similar to those described in adults. The most frequent ones are fatigue, dizziness, headaches, dyspnea, loss of smell, brain fog, sleep disorders, mood disorders.

The Peds-QL questionnaire (assessing 4 aspects of teenagers' daily life), shows an impact of these symptoms on the quality of life, schooling and daily activities being the most affected. Reassuringly, peer relationships seem preserved. Impact on schooling is important, with two thirds of patients reporting an impact on school performance, and one fourth having extended school absenteeism. The Adolescent Depression Rating Scale shows that 44% of our patients are at risk for depression. One third needs a psychological support.

If necessary, patients can be referred to specialized consultations in our multidisciplinary group (ENT, pulmonology, neurology, cardiology, etc.) or to complementary examinations (Tilt-Test, stress test). Patients having symptoms due to physical activity (fatigability, shortness of breath, dysautonomia with standing position intolerance) can benefit from a progressive and individualized reconditioning program with an adapted sport coach.

We offer a global follow-up to patients and families. School attendance is supported by making individual arrangements if required, through close collaboration with the education system.

Repetition of the questionnaires 3-6 months after the beginning of the follow-up shows a trend towards clear improvement, however a longer follow-up period would be necessary to confirm these observations.

(To allow fully up-to-date informations, numbers are susceptible to change until June)

P 14

SARS-CoV-2 seroprevalence, variability and clustering of seropositive cases in primary and secondary schools and classes of the canton Zurich: Ciao Corona study

Haile SR[1], Rainieri A[1], Rüegg S[1], Radtke T[1], Puhan MA[1], Kriemler S[1]

[1]Epidemiology, Biostatistics and Prevention Institute, University of Zurich, Switzerland

Background

Much remains unknown regarding the evolution of SARS-CoV-2 seroprevalence in children, variability and clustering of seropositive children in schools and classes as only a few school-based cohort studies exist.

Methods

SARS-CoV-2 antibodies were measured in 1854 to 2585 primary and secondary school children within 275-288 classes from 43-55 randomly selected schools in the canton of Zurich in June/July (R1) and October/November 2020 (R2), in March/April (R3) and November/December 2021 (R4). Seroprevalence was estimated using Bayesian hierarchical modelling. Variability in schools was expressed as maximum seroprevalence in a class minus minimum seroprevalence, and summarized as median (IQR).

Results

Median age at R1 was 11 yrs (min 6 to max 16), 47% of participants were male. At R4, 49% of participants at least 12 years old were vaccinated. Seroprevalence from R1 to R4 increased from 3% to 46.4% (95% credible interval [CrI] 42.6 to 50.9) including vaccinated children, or 24.7% (21.1 to 28.8) counting only recovered children.

While in R1-R3 seropositivity rates were always higher in primary than secondary schools by 0.2-4%, at R4 much higher seropositivity rates were observed in secondary schools, 75.8% (69.6% to 82.4%), than in primary schools, 31.3% (27.1% to 36.1%). This difference was also the case if only

recovered, unvaccinated children were considered. In R3 however children in primary school showed higher seroprevalence, 19.5% (16.0 to 23.7), than those in secondary school, 15.1% (10.7 to 19.6).

Variability of class seroprevalence rates within schools increased steadily over time: At R2, median variability was 11% (IQR 7-17%), in R3, 24% (17-37%). At R4, median variability had increased to 40% (22-49%), and all but one of the primary schools had lower seropositivity than all the secondary schools

Conclusion

We observed a large increase in seroprevalence from R1 to R4, especially from R3 to R4 following introduction of the vaccine for children 12 yrs and older. Up to R3, primary school children had higher seroprevalence, however at R4, secondary school children were more likely to be seropositive. This shift was in part due to introduction of the COVID-19 vaccine, but possibly also due to different behavior with more social contacts of older versus younger children outside school. Variability in seroprevalence among schools and classes was high and increased over time, even between different classes in the same school.

P 15

Low Prevalence of symptoms compatible with Long COVID in Children and Adolescents after infection with different SARS-CoV-2 variants: Results from the Ciao Corona Study

Raineri Alessia[1], Rueegg Sonja[1], Radtke Thomas[1], Ulyte Agne[1], Haile Sarah R[1], Puhan Milo A[1], Kriemler Susi[1]

[1]Epidemiology, Biostatistics and Prevention Institute, University of Zurich, Zurich, Switzerland

Introduction

Children generally have a mild course of SARS-CoV-2 infection and rarely suffer from a severe course of disease. Our previous findings showed a low prevalence (2%) of symptoms compatible with long COVID in a population-based school cohort while the wildtype variant was dominant. Here, we follow this existing cohort up for >1 yr and further assess the prevalence of long COVID with the alpha VOC in a new cohort.

Method

Our new cohort included children with serology testing in Mar/Apr 2021 and Nov/Dec 2021. Children who tested negative at both time-points served as control group. Symptoms lasting longer than 4 or 12 weeks were assessed via online questionnaires within 6 to 9 months after serology testing in Mar/Apr 2021. Additionally, children from the previous cohort reporting long-term symptoms in Mar/Apr 2021 were followed up for >1 yr (serology in Oct/Nov 2020; questionnaires in Mar, Sept and Dec 2021).

Result

Of 2450 eligible children, 561 children (8-16 yrs; 53% female; new cohort) had serology result in Mar/Apr 2021 and information on symptoms. Between Mar/Apr 2021 and Nov/Dec 2021, 7 of 230 seropositive and 6 of 331 seronegative children (3% vs 2%) reported at least 1 symptom lasting >12 weeks. Among seropositive children, headache (2/230, 1%), tiredness (4/230, 2%) and stomachache (2/230, 1%) were the most frequently reported symptoms. Within the same time-period, one seropositive and one seronegative child were hospitalized and 6 versus 11, respectively, children had to stay away from school. Self-reported health was similar in seropositive and seronegative children.

Among children in the previously established cohort (131 children reporting symptoms lasting >4 week), 38 were still eligible for re-assessment in Nov/Dec 2021 (i.e., serology and symptoms information available, and excluding seroconverted). Two of 9 seropositive and 1 of 28 seronegative children reported symptoms lasting >12 weeks. These individual symptoms were similar in Mar 2021 and Dec 2021.

Conclusion

In the new cohort, we observed a low prevalence of symptoms (1%) compatible with long COVID reproducing and strengthening the findings from our previous report (2%). Although much larger studies would be needed for precise estimates, our analysis implies no difference in risks of long COVID between VOCs (alpha vs wildtype) in children. For most children, post-acute symptoms never occurred or subsided (<12 weeks) and those with longer lasting symptoms recovered within 1 yr.

P 16

Post-COVID among children and adolescents, a population-based serological study in Geneva

Dumont Roxane[1], Richard Viviane[1], Lorthe Elsa[1], Baysson Hélène [1,2], Nehme Mayssam[2], Perrin Anne[3], L'Huillier Arnaud[3], Barbe Rémy P. Barbe[4], Posfay-Barbe Klara M. [3], Guessous Idris[2,5], Stringhini Silvia[1,5,6], SEROCoV-KIDS study group [1]

[1]Unit of Population Epidemiology, Division of Primary Care Medicine, Geneva University Hospitals, Geneva, Switzerland; [2]Department of Health and Community Medicine, Faculty of Medicine, University of Geneva, Geneva, Switzerland; [3]Division of General Pediatrics, Department of Woman, Child, and Adolescent Medicine, Geneva University Hospitals, Switzerland; [4]Division of Child and Adolescent Psychiatry, Department of Woman, Child, and Adolescent Medicine, Geneva University Hospitals, Switzerland; [5]Division and Department of Primary Care Medicine, Geneva University Hospitals, Geneva, Switzerland; [6]University Center for General Medicine and Public Health, University of Lausanne, Switzerland

Background and Objectives

Evidence to date indicates that children, like adults, can present post-COVID syndrome. However, data on paediatric post-COVID syndrome remains unclear. In this study we present preliminary results regarding the duration and characteristics of paediatric persistent symptoms, stratifying by serological status, confirmed infection and age groups.

Methods

Children aged 6 months to 17 years of adults participating in a Geneva population-based cohort (Specchio-COVID19) and from a random sample drawn from state registries were invited to participate in the SEROCoV-KIDS longitudinal study, from December 1st 2021 to February 18th 2022. Children were tested for anti-SARS-CoV-2 antibodies targeting the nucleocapsid (N) protein which is only detected after SARS-CoV-2 infection. Parents filled-in a questionnaire on persistent symptoms (lasting over 12 weeks), compatible with post-COVID syndrome. We estimated sex-and age-adjusted prevalence using marginal prediction after logistic regression.

Findings

Of 1015 children from 602 households, 555 (54.6%) were seropositive and 247 (24%) had a confirmed SARS-CoV-2 infection. Overall, 78 (7.7%) children had experienced persistent symptoms lasting over 12 weeks. The most frequent persistent symptoms declared among seropositives were fatigue, lower mood, insomnia and loss of smell.

The adjusted prevalence of persistent symptoms among seropositive children was 9.0% (95%CI: 6.7-11.6) and of 5.0% (95%CI: 3.1-7.2) among seronegatives with an adjusted prevalence difference (ΔaPrev) of 4.0% (95%CI: 1.0-7.3). Stratifying by age group, we observed that the prevalence of post-COVID was higher among adolescents (ΔaPrev = 8.3%; 95%CI: 3.6-13.5) than among younger children (ΔaPrev = 2.1% (95%CI: -4.0-3.6) among 6-11 years old and ΔaPrev = 5.1% (95%CI:-2.4-12.8) among 0-5 years old).

Interpretation

A significant proportion of children, especially adolescents, experience symptoms lasting over 12 weeks after a SARS-CoV-2 infection. However, many parents declared persistent symptoms compatible with post-COVID in seronegative children, which highlights the complexity of identifying post-COVID, clinically and methodologically. We need further investigation on timing of seroconversion and persistent symptoms, and potential other diagnoses. Nevertheless, the growing evidence of paediatric post-COVID indicates that more awareness in early detection and care management are needed.

P 17

Early support for pre-school children with visual impairments – a population-based survey in the canton of Zurich

von Rhein Michael[1,2], Jenni Oskar[1,2,3]

[1]University Children's Hospital Zurich, Child Development Center, Zurich, Switzerland; [2]University Children's Hospital Zurich, Children's Research Center, Zurich Switzerland; [3]University of Zurich, Switzerland

Background

In Switzerland, about 377`000 people live with a visual impairment. Many suffer from low vision or blindness from birth, or early childhood on, and need early intensive support. However, the exact number of children affected by severe visual impairment or blindness varies widely in the international literature, and there are no precise figures for Switzerland. In the canton of Zurich, early support is usually provided by low-vision specialists. The Units of Special Needs Education (USNE) evaluate all preschool children in need of this support, grant support measures, and allocate them to these specialists.

Methods

To assess the number of children affected, the average age of referral, developmental profiles, and additional diagnoses or impairments (such as combined hearing/visual impairments), and meassures granted to the affected children by the USNE, we analyzed examine USNE data of all referrals between 2015-2017.

Results

50 to 70 preschool-aged children in Zurich were referred for specialized low-vision support between 2015-2017. This corresponds to a reporting rate of approximately 0.35% of the children in a year group in the canton. In average, children are just over two years old (range: 7 months to 5.5 years) when they are being referred , which is about 1/2 year lower than the average age of registration for other types of measures, such as speech therapy or special needs education. 40% of the children suffered from mild, 45% from moderate, and 15% from severe visual impairments. The mean age of first presentation did not differ significantly between these groups. The children were granted an average of 85 hours of support for the 12-month period - despite a maximal possible number of 135 hours. Furthermore, on average, only about 53% of the hours granted were used.

Discussion

This is the first survey on early interventions for preschool children with visual impairments in Switzerland. The age differences between children who were early, or later referred were large, and can not be exclusively explained by the severity of the impairments. A relevant diffrence between granted and served hours was observed, which could be due to several reasons (e.g. a lack of therapeutic capacity, low success, or families being organizationally overwhelmed).

P 18

Has Telemedicine come to Fruition? Parents' and Physicians' Perceptions and Preferences Regarding Telemedicine

Bajwa Nadia[1,3], Braillard Olivia[2], Leuchinger Robin[3], Achab Sophia[4], Dominicé Dao Melissa[2], Hudelson Perneger Patricia[2], Mazouri Sanae[2], Junod Perron Noëlle[3,5]

[1]Geneva University Hospitals, Service of General Pediatrics; [2]Geneva University Hospitals, Division of Primary Care; [3]University of Geneva, Unit for Development and Research in Medical Education; [4]Geneva University Hospitals, Department of Psychiatry; [5]Geneva University Hospitals, Institute for Primary Care

Introduction

The SARS-Cov-2 pandemic boosted the use of telemedicine for both COVID-infected patients as well as for patients with acute or chronic disease. The aim of our study was to evaluate both patient and physician perceptions, preferences, and acceptability regarding the use of the different modalities of telemedicine for various health problems.

Methods

We conducted a cross-sectional survey in Geneva in 2021. Parents in waiting rooms of a paediatric emergency room were invited to answer an online questionnaire, while physicians working in private and public settings were asked to answer a similar questionnaire by email. The questionnaire focused on digital literacy, acceptability, preferences, as well as barriers and facilitators concerning a variety of telemedicine modalities for different health concerns.

Results

222 parents and 45 pediatricians participated (78% women, 42% physicians working in private practice and 58% physicians employed by public institutions). After face-to-face consultations, most parents preferred the telephone to other modalities for health issues such as simple medical advice (64%), discussion of parameters (64%), acute or chronic problems (64% and 57%), and psychological support (62%). They valued emails for communication of blood tests (62%) and renewal of medication (57%). A large majority of pediatricians considered the phone and e-mail to be an acceptable modality for all the issues mentioned above. Video was considered acceptable for follow-up of patients with both acute and chronic diseases (30%). Parents' main reasons for using telemedicine were avoiding travel (67%) and saving time (59%). Disadvantages were lack of physical examination (68%), technical problems (38%), and unsuitability (44%). Physicians feared the potential negative impact of telemedicine on the therapeutic relationship and insisted on the need for a facilitated access and a user-friendly format of online platforms.

Conclusion

The use of telemedicine has increased since the pandemic but both doctors and parents continue to prefer face-to-face consultations. Telephone remains more acceptable than video in most medical situations.

P 20

Cyanosis and "chocolate brown" colored blood; case report

Waldvogel C[1], Jacottin C[1], Martinez M[1], Kayemba-Kay's S[1]

[1]Department of Pediatrics, Riviera-Chablais Hospital, Rennaz, Switzerland

Methemoglobinemia is a rare cause of cyanosis that clinicians should be aware of. It affects preferentially infants and toddlers. Early recognition of this specific cyanosis with typical "chocolate brown" colored blood is crucial, as the outcome may be fatal when the disorder isn't identified on time and prompt treatment isn't initiated.

We report the case of two young children with the same diagnosis but different presentation and outcome.

P 21

When facing abdominal pain, don't forget ACNES!

Daeniker C[1], Caflisch M[1], Soroken C[1]

[1]Consultation médecine des adolescent, Service de Pédiatrie Générale, Département de la femme, de l'enfant et de l'adolescent, Hôpitaux Universitaires de Genève, Suisse

Introduction

Abdominal pain is a frequent complaint in adolescence, often diagnosed as functional disorder. We report two female teenagers with an anterior cutaneous nerve entrapment syndrome (ACNES) followed in our adolescent medicine consultation.

Case report

A 15 year-old brought for abdominal pain with a diminished appetite and a loss of one kilo in a week. The physical examination identified a very circumscribe pain in the lower right quadrant. The pinching of the abdominal skin caused disproportionate pain compared to the contralateral side. The contraction of the abdominal wall's muscle exacerbated the pain

(Carnett's sign). The blood, urine tests and abdominal ultrasound returned normal. A local Lidocaine infiltration relieved the pain. Despite two infiltrations, the pain reoccurred, therefore, she was referred for neurectomy which constitutes the ultimate treatment.

A 12 year-old brought for abdominal pain with nocturnal awakening without any other symptoms. The physical examination showed a localized pain in the upper left quadrant. The blood and urine tests returned normal. Overall, she consulted several times the emergency department and had three abdominal ultrasound; she was twice hospitalized and had multiple visits with different specialists (gastroenterologist, gynecologist, infectiologist and nephrologist). She was treated with laxatives, paracetamol, ibuprofen and tramadol without any pinpoint relief. ACNES was finally diagnosed given the persistent pain with typical physical signs. A local infiltration of lidocaine alleviated the pain.

Discussion

ACNES is insufficiently known and therefore underdiagnosed facing abdominal pain. It is caused by an entrapment of the terminal thoracic nerve branches, T7-T12, which pass through the anterior rectus muscle fascia and is responsible for chronic neuropathic pain. The incidence is of 1/1800 patients with a female predominance. The pain is mostly found in the right lower quadrant but can be all over the abdomen. The physical examination typically shows: a very well localized pain exacerbated by the pinching and a positive Carnett's sign. Other physical signs are allodynia, hyper/hypoesthesia or hyperalgia. The prognosis is favorable. The pain is immediately relieved following a local infiltration which strengthen the diagnosis.

Conclusion

ACNES is underdiagnosed because neurogenic abdominal wall pain is not thought of. The diagnosis is clinical! Therefore typical physical signs must be sought.

P 22

Lessons from the experience of adolescent migrants during the COVID-19 lockdowns in Switzerland, a qualitative study

Heiniger Caroline[1], Barrense-Dias Yara[2], Suris Joan-Carles[1,2]

[1]General Pediatrics, Lausanne University Hospital, Lausanne, Switzerland; [2]Research Group on Adolescent Health, DESS, Unisanté, Lausanne, Switzerland

Background

The Covid-19 pandemic and the implemented sanitary measures significantly impact adolescents in several essential areas of their lives. Young people with a migration background are a vulnerable population facing additional challenges compared to others of the same age, and little is known about their experience during this pandemic. The aim of the study is the describe their perspective.

Methods

Adolescents were recruited at the pediatric outpatient clinic at the University Hospital of Lausanne, in the French part of Switzerland. Thirteen interviews were conducted from November 2020 to January 2021. Inclusion criteria were: age between 14 and 19 years old, having lived in Switzerland for less than 10 years, and being fluent in French or English.A thematic content analysis was performed to extract themes and topics. We compared our results with other studies among adolescents.

Results

Thirteen adolescents (9 females and 4 males) from 4 countries participated in the study, with a median age of 16 years. Three of them were living in migrant reception centers. They had been living in Switzerland for an average of 2.3 years.

They encountered difficulties in understanding COVID-19 related information shared with them, whether it was about the virus or related to the lockdown. Sanitary measures were extremely well respected. Remote learning was described as stressful due to a lack of access to computers or other hardware, lack of understanding of homework, or limited space in the housing. For those living in migrant reception centers, sharing common areas and proximity with other people was very difficult. Adolescents were generally used to spending a lot of time with their families. Some youths were very isolate and did not notice any difference in their social lives during the lockdown. For example, communication using social networks with their friends abroad continued as usual. Several revealed difficulties in returning to school, notably challenges in learning French or finding an apprenticeship position.

Conclusions

Migrant adolescents share similar experiences to youth of the same age but present greater challenges in some areas like the understanding of information, or difficulties in their education and social life. In the event of further lockdown, special attention must be paid to this population to ensure their proper development and integration. A global follow up of this young population during and after the pandemic is needed

P 23

Mild symptomatology of SARS-CoV-2 infection in children born very preterm

Peralta GP[1], Adams M[2], Bassler D[2], Möller A[3], Natalucci G[2,4], Kriemler S[1]

[1]Epidemiology, Biostatistics and Prevention Institute (EBPI), University of Zurich, Zurich, Switzerland; [2]Newborn Research, Department of Neonatology, University and University Hospital Zurich, Zurich, Switzerland; [3]Division of Respiratory Medicine and Childhood Research Center, University Children's Hospital Zurich, Zurich, Switzerland; [4]Larsson-Rosenquist Centre for Neurodevelopment, Growth and Nutrition of the Newborn, University of Zurich and University Hospital Zurich, Zurich, Switzerland

Background

It is not known whether children born very preterm have an increased risk of severe symptoms following SARS-CoV-2 infection and whether a history of bronchopulmonary dysplasia (BPD) relates to more severe symptomatology. We aimed to describe the prevalence of SARS-CoV-2 infection and the severity of symptoms in a sample of children with and without BPD born at a gestational age below 32 weeks, between 2006 and 2019 in the Zurich area, in comparison to their siblings born at term (≥ 37 weeks).

Methods

Parents were invited to complete an online survey for their preterm child as well as for a term sibling of similar age, between May 2021 and January 2022. The survey included questions about SARS-CoV-2 confirmed infection, symptoms and treatment.

Results

The survey was completed for 654 preterm children (270 with prior BPD) and for 189 term children aged 2 to 15 years. 28 (7%) preterm children without BPD, 15 (6%) preterm children with BPD and 22 (12%) term children were infected by SARS-CoV-2. Out of the infected, the proportion of children with respiratory symptoms (cough, sore throat, shortness of breath) was higher in premature children with BPD (67%), than in preterm children without BPD (25%) and slightly higher than in those born at term (59%). In all groups, the majority of children had only mild symptoms. No child had to be hospitalised and only one preterm child with BPD required oxygen.

Conclusion

Very preterm children with BPD may be more likely to experience respiratory symptoms following SARS-CoV-2 infection. However, similar to children born at term, most very preterm children with and without BPD developed mild symptoms only.

Funding

Lunge Zürich (#2020-06) and ERS-LTRF-2020.

P 24

Dysfunctional Uterine bleeding and Sars-Cov-2 infection: a possible correlation.

Schillizzi M[1], Vandelli A C[1], Boffini B[1], Pasquale M F[1], Riavis M[1], Ferrucci E[1]

[1]Paediatric Institute of Southern Switzerland, Lugano Regional Hospital, Lugano, Switzerland.

Infection due to Sars-CoV-2 is known to present with a wide range of signs and symptoms, ranging from mild symptoms [1] to severe respiratory illness and inflammatory response, systemic dysfunctions even until death [2]. However, coagulation-related abnormalities are also observed.

So far, the scientific community has focused attention on hypercoagulability state causing thromboembolic events [3]. A tendency to bleed has been far less frequently investigated. Often, this tendency is referred to minor bleeding and seems to be explained by an immune thrombocytopenia [4].

In our clinical experience, however, a remarkably increasing number of cases of dysfunctional uterine bleeding has been observed in adolescents with ongoing or previous Sars-CoV-2 infection. As a consequence, in some cases a severe anaemia has been registered.

We report the case of a patient with severe anaemia associated with a sudden dysfunctional uterine bleeding started out after a Sars-CoV-2 infection. In our case, haemoglobin level was lower than 8 mg/dl (i.e., severe anaemia [5]) and the patient had to be treated with tranexamic acid, with a slow progressive decrease of bleeding. Other cases have been recorded, before realising of a likely association between bleeding and Sars-Covid infection.

Currently, a database has been created to study coagulation and hormonal status of the adolescents showing this clinical pattern. The aim is to explore possible relations between the bleeding condition and the Sars-CoV-2 infection and, eventually, to explain this clinical framework.

A multicentre trial involving international teams is going to be set up as far as to obtain more data, to have a clearer definition and to possibly confirm the suspicion of a correlation.

References

Gandhi R.T., Lynch J.B., del Rio C. Mild or moderate COVID-19. N. Engl. J. Med. 2020.

Wiersinga, W.J., Rhodes, A., Cheng, A.C., Peacock, S. J. & Prescott, H. C. Pathophysiology, transmission, diagnosis, and treatment of coronavirus disease 2019 (COVID-19): a review. JAMA 324, 782–793 (2020).

Emert, R., Shah, P., Zampella, J.G. COVID-19 and hypercoagulability in the outpatient setting (Open Access) (2020) Thrombosis Research, 192, pp. 122-123.

Bhattacharjee, S., Banerjee, M. Immune Thrombocytopenia Secondary to COVID-19: a Systematic Review. SN Compr. Clin. Med. 2, 2048–2058 (2020).

World Health Organization. Hemoglobin concentrations for the diagnosis of anaemia and assessment of severity (WHO/NMH/NHD/MNM/11.1).

P 25

Eating disorders: two cases of very rare complications.

Rinaldo C[1,3,4,5], Ragusi Guenet L[1,3,4,5], Queirolo S[1,3,4,5], Calciolari J[1,3,4,5], Stefani-Glücksberg AN[2,3,4,5], Ferrucci E[1,3,4,5]

[1]Department of General Pediatrics; [2]Department of Pediatric Cardiology; [3]Institute of Pediatrics of Southern Switzerland (IPSI); [4]Ente Ospedaliero Cantonale; [5]Switzerland

Among eating disorders, anorexia nervosa is one of the most burdened by serious physical consequences, linked to malnutrition and relative weight decrease, which complicate the psychiatric clinical picture and can also cause irreversible damage, unfortunately leading even to death.

We report on the cases of two adolescent female patients suffering from anorexia nervosa, both didactically relevant for their framework of presentation.

The first patient presented a 50% weight loss in 10 months, slowing of thinking, speech and general activity. In a serious context of general deterioration, a RMI was performed, finding an alarming radiological picture with a severe cerebral atrophy, visible in ventricles enlargement and diffuse cerebral convolution thickening, as well as a considerable reduction of adenohypophysis size, of course due to a deficient hormonal stimulation.

In the other patient, who had "only" a 20% weight loss - however starting from an already underweight condition of constitutional thinness, therefore not less serious – electrocardiography pointed out an atrial ectopic beat on a new-onset extreme bradycardia, as well as a non-specific repolarization disorder, with low voltages.

Obviously, the severity and the reversibility of the complications depend on chronicity and seriousness of eating disorders, although it has been shown that return to normal is almost never complete.

Of course, an adequate nutritional intake, with weight recovery, is the right way to reduce the majority of somatic complications for the patients; furthermore, an improvement in physical health can allow an improvement in the psychiatric situation.

A crucial prerequisite in approaching these patients, therefore, is setting-up a multidisciplinary team, with the aim to respond to their multiple needs: paediatrician, dietician, psychiatrist, psychologist and educator.

P 26

Use of tobacco, nicotine, and cannabis products among students in two Swiss cantons

Affolter J[1], Rohland E[1], Philippe M[2], Tal K[3], Auer R[3,4], Jakob J[3,5]

[1]Swiss Lung Association Aargau, Department of Health Promotion and Prevention; [2]Swiss Lung Association St.Gallen - Appenzell, Department of Health Promotion and Prevention; [3]Institute of Primary Health Care (BIHAM), University of Bern, Switzerland; [4]Center for Primary Care and Public Health (Unisanté), University of Lausanne, Lausanne, Switzerland; [5]Department of Paediatrics, University Hospital Bern, Inselspital, Bern, Switzerland

Introduction

Most of the people who smoke began in their teens and teens may be equally attracted to new tobacco, nicotine, and cannabis products. There is little data on how many teens use these new products or how often they consume them. We described use prevalence among students, including daily use, of tobacco, nicotine, and cannabis products, in two Swiss cantons.

Methods

We invited students at the upper secondary level in Aargau and St.Gallen to participate in an online survey between November 2021 and February 2022. The survey asked participants for basic demographic information and how often they used tobacco products (tobacco cigarettes in commercial packages, self-rolled tobacco cigarettes, hookahs, pipes, cigars and cigarillos, tobacco toasters, snus, snuff), nicotine products (nicotine pouches, e-cigarette with nicotine, e-cigarette without nicotine) and cannabis products (cannabis smoking with and without tobacco mulling, cannabis vaping). Answers were scored on a Likert scale (no use in past month, less than weekly, weekly but not daily, daily use, prefer not to say). We tabulated the responses and reported descriptive statistics.

Results

Of the 32,614 students who were invited to participate, 9,515 students from 23 schools completed the survey (29.2%); 4,712 identified as female (49.5%), 4,606 identified as male (48.4%), and 197 (2.1%) identified as another gender. In our age categories, 9.5% were under 16, 47% 16-17, 27.5% 18-19, and 16% were over 19. Figure 1 shows proportions and use frequency of all the products we monitored. Most participants (54.8%) reported they had used at least one product at least one time within the last month. Reported daily use was most frequent for tobacco cigarettes in commercial packages (14.2%), snus (4.1%) and cannabis smoking with tobacco mulling (3.6%).

Conclusion

Students who use tobacco are still most likely to smoke cigarettes, but many regularly use new tobacco, nicotine, and cannabis products, though

use frequency varies. We should repeat the survey regularly to track changes in the relative popularity of these products.

P 27

Swiss drugs authorised for use in children

Tilen R[1,2], Berger C[1], Allemann S[3], Meyer zu Schwabedissen H E [2]

[1]University Children's Hospital Zurich, Department of Infectious Diseases and Hospital Epidemiology, 8032 Zurich; [2]University Basel, Department of Pharmaceutical Science, Biopharmacy, 4056 Basel; [3]University Basel, Department of Pharmaceutical Science, Pharmaceutical Care, 4056 Basel

Introduction

Before the introduction of the European paediatric regulation (EC No 1901/2006), dosage-finding studies and drug approval were mostly limited to adults. As a result, a considerable proportion of drugs administered to children are not authorised for this purpose, and consequently off-label use in paediatrics belongs to standard of care. Studies in Switzerland have shown that in children's hospitals, about half of the prescriptions are not covered by the terms of the drugs marketing authorisation and are therefore not in accordance with the drug label, but it is unknown how many of the drugs on the Swiss market are actually not approved for use in children. Our aim is to quantify the number of drugs that have a dosage recommendation for children and to assess the quality of the available information.

Methods

We used natural language processing to screen all Swiss drug labels available in German for information about children and the presence of a specific dosage recommendation for this population. Based on a previous analysis of the labels of the most frequently used drugs in Swiss children's hospitals and the AmiKo Web, 10 different search terms were defined to retrieve this information.

Results

The analysis of the 4,452 drug labels describing 16,767 products on the Swiss market identified 29,653 sentences including the search terms. These sentences were in 4,330 different brand-name drugs corresponding to 1,579 chemical substances. The retrieved sentences were most often found in the sections "Posology and method of administration" (37.4%), "Other information" (13.8%), and "Warnings and Precautions" (11.0%).

Discussion & conclusions

The majority of Swiss drug labels contain information about children, but only a minority refer to a specific dosage recommendation for use in children. Information in drug labels about children frequently concerns warnings and precautions or other information, although this is often not evidence-based and has only legal meaning. Further analysis of the search results is required to quantify the number of drugs authorised for children in Switzerland.

P 28

Beckwith-Wiedemann Syndrome: from mosaicism to a unified management

Fernandez B[1], Perrin A[1], Ghinescu C[1], Papangelopoulou D[1], Bajwa N[1]

[1]Service de Pédiatrie – Hôpitaux Universitaire de Genève, HUG, Genève

Case report

A 4-week-old male infant was addressed to the emergency room by his pediatrician for a left arm cyanosis. Upon inspection he had macroglossia and left sided facial hypertrophy. He also presented with a left linear ear lobe crease and an umbilical hernia. Abdominal echography and MRI showed a left adrenal mass and left nephromegaly. Cardiac exam and echography were normal. Initial laboratory investigations showed an elevation of total plasmatic metanephrines and Neuron-Specific Enolase. He also developed episodes of hypoglycemia during hospitalization. Endocrinological consultation excluded hyperinsulinism. Genetic testing confirmed Beckwith-Wiedemann syndrome (BWS) due to paternal uniparental disomy of chromosome 11p15.5. The tumor board suspected a neuroblastoma of the left adrenal gland associated with BWS. The patient will be initially followed-up every month for the adrenal mass 3 months of age and, depending on the evolution, an MIBG scan will be realized. Because BWS patients are at risk for embryonal tumors, after the age of 3 months the patient will be followed-up with serial abdominal ultrasounds every 3 months and followed by an oncologist until 7 years of age.

Discussion

BWS is a paediatric overgrowth disorder involving a predisposition to solid tumor development. Its estimated population prevalence is about 1 in 10,300 but could be underestimate because of its milder phenotypes (due to mosaicism). It usually occurs sporadically (85%), but familial transmission occurs in approximately 15% of cases. Although the phenotype of BWS is variable, most patients have one or more of the following: macroglossia, macrosomia, asymmetric overgrowth of regions of the body (hemihyperplasia), omphalocele, visceromegaly and solid embryonal cell cancers during early childhood. The most common types of tumors are Wilms tumor (52%), hepatoblastoma (14%), neuroblastoma (10%), and adrenal carcinoma (3%). Cancer risk is the highest during the first 2 years of life and declines afterwards.

Conclusion

BWS is a genetic disorder that requires an intense oncology follow-up during the first years of life. Clinical manifestations may not be obvious at birth and patients may present with variable phenotypes due to mosaicism which may delay diagnosis and patient follow-up. Paediatricians should have a high index of suspicion for BWS in infants born with macroglossia. Awareness of this condition may enable prompt diagnostic and initiation of cancer screening.

P 29

The lysinuric protein intolerance : a case report

Tobler Caroline[1], Bajwa Nadia[1], Kern Ilse[1]

[1]Service de Pédiatrie Générale, Hôpitaux Universitaires de Genève, Suisse

A 6 ½ year-old female patient, originally from Macedonia, presented to the migrant consultation with failure to thrive (height and weight below the 3rd percentile), asthenia, muscular weakness, repeated episodes of diarrhea and vomiting, a pronounced distaste for meat as well as a medical history of impaired consciousness following meat consumption. The patient first presented with symptoms at the age of two months. Physical exam revealed hepatosplenomegaly as well as a growth and weight restriction.

A metabolic disease was strongly suspected, and further investigations were performed and revealed findings consistent with a lysinuric protein intolerance (high plasma glutamine, low plasma arginine-ornithine- lysine, and high urinary lysine). The blood tests also revealed signs of macrophagic activation syndrome (anemia, increased ferritin and LDH), one of the complications of lysinuric protein intolerance. Ammonia levels were within the normal range. The nutritional assessment revealed a prealbumin deficiency, compatible with insufficient protein intake.

The diagnosis of lysinuric protein intolerance was made based on the clinical and laboratory findings. A treatment consisting of a low-protein diet and citrulline therapy was initiated. With time and treatment, the goal will be to increase the protein content of her diet to 0.9g/kg/day (the minimum recommended daily allowance).

Lysinuric protein intolerance (LPI) is a rare metabolic autosomal recessive disease caused by mutations in the SLC7A7 gene encoding for the y+LAT1 protein, which normally allows for the transport of cationic amino acid (lysine, ornithine and arginine) at the basolateral membrane of renal tubular cells and intestinal cells. This defect causes arginine to become trapped in the cells which then leads to an overproduction of nitric oxide that leads to a pro-inflammatory state.

LPI is not only a urea cycle disorder but also a multi-systemic disease that can present with many complications, such as pulmonary alveolar proteinosis, macrophagic activation syndrome, hemophagocytic-lymphohistiocytosis, renal disease, some auto-immune disorders and even an immune deficiency. Mental development is usually normal. Although it is a rare disease with an extremely broad phenotype, the complications can be fatal if it is not diagnosed and treated properly. Recognition of this rare disease is important in order to prevent potentially lethal complications.

P 30

The use of professional interpreter services at a pediatric emergency department in Switzerland

Buser S[1], Gessler N[1], Gmuender M[1], Feuz U[1], Fayyyaz J[2], Jachmann A[3], Keitel K[1], Brandenberger J[1, 2]

[1]Pediatric emergency department, University Hospital of Bern, Bern, Switzerland; [2]Division of Pediatric Emergency Medicine, Hospital for Sick Children, Toronto, Ontario, Canada; [3]Emergency Department, University Hospital of Bern, Bern, Switzerland

The aim of our study was to analyze the use of interpreter services and improve communication during health encounters with families with limited language proficiency (LLP) at the pediatric emergency (ED) department of the University hospital of Bern.

This study is a pre and post interventional study analyzing the use of interpreter services for LLP families. All non-Swiss families presenting at the ED were eligible to participate in the study. If agreeing to participate, the language proficiency of the caregiver present during the health encounter was systematically assessed in a phone interview, using a standardized screening tool. Further variables were extracted using administrative health records. To improve the use of interpreter services, a package of interventions was implemented at the department during 3 months. It consisted of: i) in-person and online transcultural teaching ii) awareness raising through the regular information channels and iii) the introduction of a language proficiency pathway to identify and manage LLP families.

The proportion of LLP families who received an interpreter was 11.0% (14/127) in the pre intervention period compared to 14.8% (20/135) in the post intervention period. The interpreter use was therefore increased by 3.8%t (95% CI -0.43 to 0.21; p = 0.36). The assessed level of language proficiency of caregivers differed strongly from the self-reported level of language proficiency. Of the study participants whose language proficiency was screened as limited, 77,1% (81/105) estimated their language proficiency level as intermediate. More than half of the LLP families (54.0% (88/163)) reported, that they would have liked and interpreter during the consultation. Only a quarter of families (24.7% (22/178)) knew about their right to a free of charge interpreter service at the ED. 47.8% (22/46) of LLP families who did not receive an interpreter reported that a minor below 18 years of age translated during the consultation.

Conclusions

Systematic screening of language proficiency and management of families with LLP is feasible and needed at health care facilities to ensure safe and equitable health care. Relying on caregivers´ self-assessment in language proficiency and their active request for an interpreter is not sufficient to ensure safe communication during health encounters. Further interventional studies are needed to sustainably improve the use of interpreter services for families with LLP.

P 31

Burden of covid-19 pandemic on adolescents' mental health: eating disorders spreading

Beykirch J[1], Gamper L (shared first authorship)[2], Landolt M[1], Rutishauser C[2]

[1]Department of Psychosomatic Medicine, University Children's Hospital Zurich, Switzerland; [2]Department of Adolescent Medicine, University Children's Hospital Zurich, Switzerland

Adolescents are not an at risk population for severe COVID-19. However, an increase of mental health disorders in this age group were reported. The aim of this study was to compare rates of referred adolescents with eating disorders (ED) requiring inpatient treatment at the University Children's Hospital Zurich before and during the pandemic.

Methods

Retrospective data analysis of referred adolescents in need for inpatient treatment because of ED (anorexia nervosa, atypical anorexia nervosa, bulimia nervosa, eating disorders not otherwise specified) between 2019-2021. Inpatients on the pediatric ward (requiring urgent intervention) and those on the psychosomatic ward were evaluated separately.

Results

Pediatric ward: the rate of adolescent inpatients with eating disorders increased significantly in June 2020, shortly after the end of the lockdown: from June - December 2020, admission rates were 67% higher compared to the same time frame in 2019, and the number of treatment days increased 76%. Throughout 2021, the average treatment days per month remained high: they were only 6.4% lower (93.6 d/month) compared to June – December 2020 (100.1 d/month).

Psychosomatic ward:In 2020, there were 94% more patient referrals for inpatient treatment compared to 2019, and in 2021 115%. Only 38% of referred patients could be accepted for inpatient treatment in 2020, and 47% in 2021, in contrast to 94% in 2019.

Discussion

Our data show a sharp increase of adolescent inpatients treated for eating disorders shortly after the end of the pandemic lockdown and persisting throughout 2021. This could be a sign of increased vulnerability in this age group.

More than 80% of patients mentioned the lockdown with closures of schools, sport clubs and other adolescent meeting venues as inducing factors for developing an eating disorder, possibly associated with the disappearance of structures that provide support and regulate self-esteem. Increased conflicts between family members were also reported.

The figures also show an increased shortage of inpatient treatment capacity, with long waiting times even before the pandemic, and worsening after the lockdown. The burden of patient and their family was high during the waiting period leading to an excessive demand for outpatient triage clinics.

P 32

Persistence of respiratory symptoms in children born very preterm

Peralta GP[1], Adams M[2], Bassler D[2], Möeller A[3], Natalucci G[2,4], Kriemler S[1]

[1]Epidemiology, Biostatistics and Prevention Institute (EBPI), University of Zurich, Zurich, Switzerland; [2]Newborn Research, Department of Neonatology, University and University Hospital Zurich, Zurich, Switzerland; [3]Division of Respiratory Medicine and Childhood Research Center, University Children's Hospital Zurich, Zurich, Switzerland; [4]Larsson-Rosenquist Centre for Neurodevelopment, Growth and Nutrition of the Newborn, University of Zurich and University Hospital Zurich, Zurich, Switzerland

Background

The long-term respiratory consequences of very preterm birth are not yet well characterized. We aimed to describe the prevalence and severity of respiratory symptoms in a sample of children with and without bronchopulmonary dysplasia (BPD) born at a gestational age below 32 weeks, between 2006 and 2019 in the Zurich area, in comparison to their siblings born at term (≥ 37 weeks).

Methods

Parents were invited to complete an online survey for their preterm child as well as for a term sibling of similar age, between May 2021 and January 2022. The online survey included validated questions for cough (with and without cold), wheezing and breathing difficulties during physical exertion. It also included questions on absence from school and hospitalisation due to respiratory symptoms in the last 12 months.

Results

The survey was completed for 658 children born preterm (273 with prior BPD) and for 190 term children aged 2 to 15 years. A higher proportion of preterm children had cough (92% vs. 82%), wheezing (42% vs. 26%) and breathing difficulties during exertion (12% vs. 4%) than children born at term. Preterm children were also more likely to be absent from school for two weeks or more than term children (7% vs 3%). Sixteen children born preterm were hospitalised due to respiratory problems, while this was not the case for any of the children born at term. The prevalence of respiratory symptoms was higher in preterm children with BPD than in those preterm without BPD.

Conclusion

Children born very preterm remain at increased risk for respiratory morbidity during childhood and adolescence. Families of very preterm children may need additional support to cope with the burden of respiratory symptoms.

Funding

Lunge Zürich (#2020-06) and ERS-LTRF-2020.

P 33

Pediatric residents in Switzerland in 2021: training, opinions and concerns

Julian Jakob, MD[1,2], Office paediatrie schweiz[3], Christoph Aebi, MD[1]

[1] Department of Paediatrics, Bern University Hospital, Inselspital, Bern, Switzerland; [2] Institute of Primary Health Care (BIHAM), University of Bern, Switzerland; [3] Claudia Baeriswyl, Regina Balmer, Mélissande Imseng, paediatrie schweiz (Swiss Society of Paediatrics), Fribourg, Switzerland

Introduction

Medical residents are exposed to stress due to their situation as learning professionals. We know from studies on residents in general internal medicine in Switzerland that workload is high, and satisfaction mixed. We aim at describing training strategies, opinions, and concerns of residents in pediatrics in Switzerland.

Methods

In July 2021, we sent an invitation to participate in an anonymous online survey in German or French to all Swiss pediatric hospitals and asked the chairpersons to circulate the link among their resident staff. We also sent the invitation directly to all pediatric residents who were member of the Swiss Society of Pediatrics (N = 412). We collected basic demographic information and data on training strategies, working patterns, working experiences, and asked participants to rate possible development options (graded from 0 = not important to 100 = very important).

Results

In total, 212/529 (40%) residents filled in the survey, 147 (69%) in German, and 65 (31%) in French. Among the 212, 181 (85%) provided demographic information, and 85% reported being a women. The median age was 30 years. They worked exclusively in a hospital in 95% of cases, the remainder worked in a private practice or in research. They reported working a median of 55 hours for a 100% quota, with no change according to the year of residency or sex. Working hours were 4 hours per week lower in practice compared to university hospitals. Most worked full-time and 10% reported part-time employment. Overall, 55% of the participants felt they worked too much, and this was attributed to insufficient administrative support (case management missing in 47%) or dissatisfaction with the clinical software (39%). Training resources included textbooks (88%), online media and journals (83% each), congresses (61%) and the "Repetitorien/répétitoires" (58%).

Residents were satisfied with the training provided at their institution (84% satisfied or very satisfied). Suggestions for optimization were graded as follow (median/100): curriculum of ultrasound 95 (99.5 in German vs. 80 in French speaking participants), digital educative platform 90, more part time job opportunities 100, and a 42-hour workweek 100 (results similar in both language groups).

Conclusion

Pediatric residents in Switzerland are mostly female and were generally satisfied with their profession and training but wished for more administrative support and more part-time opportunities to reduce their high workload.

P 34

Plastic doesn't just cause global warming!

Testi C[1], Callias C[1], Glasson N[2], Laubscher B[1]

[1]Département de pédiatrie, Réseau hospitalier Neuchatelois-RHNE, Neuchâtel, Switzerland; [2]Service d'Oto-rhino-laryngologie, Centre hospitalier universitaire vaudois–CHUV, Lausanne, Switzerland

Case report

A healthy 5 month-old boy attended the ER in winter with light respiratory distress attributed to bronchiolitis without fever. He was discharged home. 24h later, he presented with hoarseness, barking cough and inspiratory stridor suggesting a viral croup. He improved with 2 epinephrine inhalations and oral dexamethasone, and was discharged home after 2 days. 48 hours later, he presented again with severe upper respiratory obstruction recurrence. He was hospitalized and treated again with epinephrine inhalations and oral dexamethasone. Over 6 days, recurring biphasic stridor led to repeated epinephrine inhalation (5 times) and a 7 days oral steroids treatment. Past history revealed full immunizations, a first bronchiolitis at 4 months, just after starting nursery, and a surprising persistent dry cough, noisy breathing and light chest retraction since then. He has no gastro-esophageal reflux, no history of foreign body inhalation, no intubation or cutaneous hemangioma on clinical examination. The chest X ray was normal. A naso-pharyngeal Adenovirus was found. At that time, the mother remembered that weird respiratory noises already started around the age of 3 months. Because of the atypical clinical history, an upper airway endoscopy was performed and revealed a subglottic plastic sheet of 1.5 cm maximal diameter in sagittal position with no inflammation (which explains the respiratory tolerance). After its extraction, the stridor and the respiratory distress immediately and completely disappeared.

Discussion

Croup is an upper respiratory tract infection leading from swelling especially from the subglottic space resulting in inspiratory stridor and barking cough. Main causes are viral infection (parainfluenza 1) affecting children between 6 months to 3 years. Before 6 months and after 6 years, viral croup is uncommon and it remains a self-limited disease. Symptoms usually resolve within 2 to 7 days. Persistent/recurrent stridor without fever should prompt consideration of other diagnosis such as laryngomalacia, vascular malformations, mass (hemangioma, papillomatosis), vocal cord palsy or subglottic stenosis. Foreign body inhalation is rare before 6 months of age but always has to be considered as a differential diagnosis, especially in cases with atypical history.

Conclusions

In an infant, even a very young infant, with an atypical croup syndrome, always consider a foreign body aspiration.

P 35

Correlating food allergic reactions with the consumed food (AAGE201 study)

Piletta-Zanin A[1], Scherl A[2], Harr T[3], Nigolian H[3], Manzano S[4], Grosgurin S[5], Tonson la Tour A[6], Brändle G[7], Soechbach H[5], caubet JC[1], Pahud JP[8], Ortelli D[2], EEdder P[2], Eigenmann P[1]

[1]Unité d'allergologie pédiatrique, Service des spécialités pédiatriques, Département de la femme, de l'enfant et de l'adolescent, HUG; [2]Service de la consommation et des affaires vétérinaires (SCAV), Direction Générale de la Santé, Etat de Genève; [3]Service d'allergologie et immunologie, Département de médecine, HUG; [4]Service d'accueil et d'urgences pédiatriques, Département de la femme, de l'enfant et de l'adolescent, HUG; [5]Service des urgences, Département de médecine aiguë, HUG; [6]Service des urgences pédiatriques, Hôpital de la Tour; [7]Urgences pédiatriques, Hirslanden Clinique des Grangettes; [8]Groupement intercommunal pour l'animation parascolaire, Association des communes genevoises

Introduction

5-8% of the population suffers from a food allergy that can potentially lead to a severe reaction. Different factors of severity have been studied, but the relationship between the amount of allergen consumed and the severity of the reaction has never been investigated. Therefore, there is uncertainty regarding the consumption of very small amounts of allergen (traces). The aim of our study was to analyze qualitatively and quantitatively the allergen content in foods that have caused a food allergy reaction.

Methods

We collected during one year (2021) medical history, food samples eliciting reactions and allergy diagnostic tests of patients presenting reactions suggestive with immediate food allergy and consulting in the emergency departments in the University Hospitals of Geneva and local hospitals or in the allergology outpatient consultation. The samples were analyzed by immunoassays and mass spectrometry.

Results

In total we recruited 145 patients, 105 (72.4%) were children. Patients were mostly reacting to milk, egg, tree and peanuts. 30% of the reactions were severe reactions needing epipen. When analyzing the foods, we found in the 115 food samples analyzed so far eliciting doses from 12.8 mg/kg to 1000g/kg of food.

Discussion

The analysis of the results is still in progress, but we can already conclude that in this non-exhaustive series of food allergic reaction over one year in the canton of Geneva, (1) children were over-represented among the reported reactions, (2) reactions were mostly mild and moderated, (3) most patients were reacting to significant amounts of foods and not traces. In conclusion, results for this study will help to provide better counselling for prevention of accidental reactions, in particular in children.

P 36

Incidental splenomegaly in a teenager

Baghin V[1], Prader S[1], Sirin S[2], Pachlopnik Schmid J[1], Trück J[1]

[1]Division of Immunology, University Children's Hospital Zurich, University of Zurich (UZH), Zurich, Switzerland; [2]Department of Diagnostic Imaging, Children's Hospital Zurich, University of Zurich (UZH), Zurich, Switzerland

We report the case of a healthy and asymptomatic male teenager who presented to his primary care pediatrician for a routine check-up. On clinical examination isolated splenomegaly was noted, later confirmed sonographically and by MRI (20x13x7cm, ca. 1000 ml). Laboratory investigations revealed bicytopenia (Hb 69 g/L, WBC 1.52 G/L, Tc 151 G/L), elevated double-negative T cells (14.7%), hypergammaglobulinaemia (IgG 19.3 g/l), vitamin B12 (1530 pg/ml) and soluble Fas-ligand (2428 pg/ml). Extensive investigations excluded an underlying oncologic, infectious or metabolic disease. Genetic investigation revealed a previously unknown heterozygous variant in FAS, which was functionally verified by defective in vitro lymphocyte apoptosis. According to current diagnostic criteria, autoimmune lymphoproliferative disease (ALPS) was diagnosed. Initiation of immunosuppressive therapy consisting of short term p.o. administration of prednisolone and long-term sirolimus treatment resulted in normalization of laboratory abnormalities and reduction of spleen size. ALPS is a rare primary immune dysregulation disorder caused by a defective FAS-mediated apoptotic pathway in lymphocytes. The clinical hallmarks are uncontrolled lymphocyte proliferation leading to lymphadenopathy, splenomegaly and autoimmune disease, mainly autoimmune cytopenia. The treatment focuses on symptom control and is centered on immunosuppressive drugs. This case highlights the importance of a thorough physical examination even in asymptomatic patients and presents a rare cause of incidental splenomegaly.

P 37

Recurrent vaccine injection site reactions during febrile PFAPA episodes

Görtz Sabrina[1], Bogatu Bettina[3], Trück Johannes[1]

[1]Division of Immunology, University Children's Hospital Zurich, University of Zurich (UZH), Switzerland; [2]Division of Allergology, University Children's Hospital Zurich, University of Zurich (UZH), Switzerland

We present the case of a 2.5-year-old girl with recurrent skin reactions at a vaccine injection site associated with PFAPA (Periodic Fever, Aphthous stomatitis, Pharyngitis, Adenitis) episodes. She had monthly fever episodes lasting 3-6 days since she was 7 months old, associated with pharyngitis, cervical lymphadenitis, vomiting and abdominal pain. In addition, aphthae appeared after about 1.5 years. The patient is otherwise healthy and shows normal growth and development.

Associated with febrile PFAPA episodes, a red non-pruritic macular skin lesion (diameter 4x2cm) occurred on the left anterolateral thigh at the exact site of a previous vaccine injection. These transient skin changes were first noted at the age of 16 months, approximately 7 weeks after vaccination with three routine vaccines in both thighs. There were no noticeable skin abnormalities between episodes of fever. Over time, the size and intensity of the skin eruption decreased with each episode of fever, reappearing again one day after administration of the quadrivalent meningococcal conjugate vaccine into the other thigh without accompanying symptoms.

To our knowledge, recurrent skin reaction at a vaccine injection site associated with PFAPA have not been reported previously. The pathomechanism remains unclear. Cases with self-limiting, delayed skin reactions at injection sites of vaccines or immunotherapies triggered by viral infections have been described. The persistence of adjuvants could trigger a local immune response during the general inflammatory response in a PFAPA fever episode. Other possible causes of the phenomenon are the presence of tissue-resident memory T cells triggering mild inflammation of the skin or a specific cross-reaction, similar to the known skin reaction at the BCG application site in Kawasaki patients.

This case demonstrates an expansion of the disease spectrum of PFAPA associated with vaccination. Mild and self-limiting skin reactions to vaccines are not a contraindication to future vaccinations with the same or similar vaccines. Clinicians treating PFAPA patients should be alert to the reported phenomenon to reassure parents and thus maintain confidence in continuing the vaccination schedule.

P 38

Transient synovitis, are you sure?

Mrabet-Deraoui I[1], Schmid H[2], Woerner A[3], Bruder E[4], Zoubir SA[1]

[1]Department of Pediatrics, Hospital of Jura, Delémont, Switzerland; [2]Pediatric Infectious Diseases and Vaccinology, University of Basel Children's Hospital Basel, Basel, Switzerland; [3]Division of Pediatric Rheumatology, University of Basel Children's Hospital Basel, Basel, Switzerland; [4]Pathology, Institute of Medical Genetics and Pathology, University Hospital Basel, University of Basel, Basel, Switzerland

Case report

A healthy 17-month-old boy presented with a 7-day history of intermittent limp, with pain and restricted movement of his left knee, worse in the morning and improving over the day, not responding to treatment with NSAID. He was overall in good condition, without associated fever. There was no history of preceding trauma nor viral illness. . Physical examination was unremarkable except discrete left-sided limping while walking, without any restriction of hip movement nor of other lower limb joints. Laboratory investigations showed moderate signs of inflammation with a CRP level of 28 mg/L and an ESR of 45 mm/h, with a normal white blood cell count. Ultrasound of the hips was normal. Due to persistence of symptoms as well as moderately raised inflammatory markers, differential diagnoses other than transient synovitis were considered, including osteoarticular infections (OAI).MRI showed an approximately 2 cm x 1.5 cm x 1 cm mass clearly delimited in the epiphyseal/metaphyseal growth plate with no joint effusion nor intra-articular involvement of the distal femur. CT-guided biopsy allowed to exclude malignancy and histiocytosis and confirmed chronic-florid osteomyelitis. Culture remained negative, but eubacterial PCR detected K. kingae. Along the course of the disease, the child remained in a good condition, afebrile, with no progress of focal symptoms and made a full recovery. Empiric antibiotic therapy using co-amoxicillin was started intravenously and switched to oral switch after 48 hours, in keeping with the pathogen identified. Repeat MRI 1 month into treatment showed improvement of the inflammatory changes, with however slightly progredient cystic changes in the femoral metaphysis, reason for which the antibiotic treatment course was extended to complete a total of 2 months.

Discussion

This case illustrates that K. kingae should be considered as a major bacterial cause of OAI in children less than 4 years. Culture is often negative. Eubacterial PCR and more accurately, PCR specific to K. kingae, are useful tools to confirm the diagnosis. The clinical presentation and biologic inflammatory response tends to be milder than in OAI caused by other bacterial pathogens. Radiologic findings are more subtle. Affected children tend to present fewer criteria evocative of OAI.

Conclusion

Don't forget, K.kingae OAI in the child less than 4 years old with mild, but persistent clinical symptoms. PCR testing is a major tool for the diagnosis.

P 39

H. influenzae meningitis in 20 month-old unvaccinated patient: case report

Leralta Anna[1], Martin Helena[1], Chrast Livia[1], Canciani Alberto[1], Cova Lorena[1], Sallin Christelle[1], Szita Gillian[1], Rochat Mascha[1]

[1]Department of Pediatrics, Hôpital Intercantonal de la Broye, Payerne, Switzerland

The ever-increasing number of cases of children not vaccinated against vaccine-preventable infections is forcing us to increasingly reconsider diseases we used to perceive as almost eradicated in the western world.

We report the case of an immunocompetent, unvaccinated patient aged 1 year and 8 months who was admitted to the emergency department with a reduced level of consciousness after 36 hours of drowsiness, fever and vomiting. He had been initially diagnosed with viral gastroenteritis and an otitis media which had not been treated with antibioticswas not treated. Physical examination revealed an axial atony and divergent eye fixation with mydriasis. The lumbar puncture showed high protein levels and 81% of polynucleated leukocytes. Empiric antibiotics (ceftriaxone) and corticotherapy (dexamethasone) were administered. A cerebral CT-scan showed an untreated otitis media and ethmoidal sinusitis. Serotyping of hemophilia found in the spinal fluid and blood revealed serotype type B. A follow-up with the pediatric otolaryngologist was fixed in order to evaluate possible sequelaes.

Efforts to keep the incidence of invasive Haemophilus type B meningitis low should be directed to maintaining a high level of vaccination coverage, especially among children under 2 years of age.

P 40

Hoagland's sign: when the eyes suggest an acute infectious mononucleosis

Bronz Gabriel[1,2], Kottanattu Lisa[1], Vanoni Federica[1], Bianchetti Mario[2]

[1]Pediatric Institute of Southern Switzerland, Ospedale San Giovanni, Bellinzona, Switzerland.; [2]Family Medicine Institute, Faculty of Biomedical Sciences, Università della Svizzera Italiana, Lugano, Switzerland.

Background: In subjects without immunodeficiency or autoimmunity, primary Epstein-Barr virus infectious mononucleosis typically affects children and young adults and presents with fatigue, poor appetite, fever, sore throat, and enlarged cervical lymph nodes, liver or spleen. Bilateral eyelid swelling, mostly referred to as Hoagland sign, occasionally occurs in infectious mononucleosis. Since reviews and textbooks do not or marginally refer to Hoagland sign in infectious mononucleosis, we present our prospective experience. Summary: Between 2019 and 2021, one of us managed 26 subjects with the characteristic clinical and laboratory features of primary Epstein-Barr virus infectious mononucleosis: 7 children (≤ 16 years of age) and 15 adults. A Hoagland sign was noted at presentation in 14 (54%) cases: 4 children and 10 adults. Patients with (4 males and 10 females, 18 [16-23] years of age; median and interquartile range) and without (8 males and 4 females, 18 [23 -25] years of age) Hoagland sign did not significantly differ with respect to sex and age. Take home message: Bilateral eyelid swelling, also called Hoagland sign, is a common but likely underrecognized finding in primary Epstein-Barr virus infectious mononucleosis.

P 41

Acute flaccid paralysis – fast action required

Bigi Sandra[1,2], Ramette Alban[3], Barbani Maria Teresa[3], Altpeter Ekkehardt[4], Bieri Andreas[5], Hoffmann Angelika[6], Aebi Christoph[7]

[1]Institute of Social and Preventive Medicine, University of Bern, Bern, Switzerland; [2]Department of Neurology, Bern University Hospital, Inselspital, University of Bern, Switzerland; [3]Institute for Infectious Diseases, University of Bern; [4]Federal Office of Public Health, Communicable Diseases Division, Bern, Switzerland; [5]Department of Pediatrics, Cantonal Hospital Aarau, Switzerland; [6]University Institute of Diagnostic and Interventional Neuroradiology, University Hospital Bern, Inselspital, University of Bern, Switzerland; [7]Division of Pediatric Infectious Disease, Department of Pediatrics, Bern University Hospital, Inselspital, University of Bern

Background

Acute flaccid paralysis (AFP) belongs to the rare diseases to be reported to the Swiss Pediatric Surveillance Unit (SPSU). Since 1998, a total of 268 cases were registered. The incidence of AFP ranges between 0.25-1.75/100'000. Poliomyelitis-like AFP associated with enterovirus D68 (EV-D68) has emerged globally during the past decade. Here we describe the first two cases reported in Switzerland.

Case presentations

A nine year-old boy and a seven year-old girl presented with AFP six weeks apart, five and two days after a febrile upper respiratory tract infection (URTI), respectively. The neurological examination revealed a near-total monoplegia of the right arm in the boy and a tetraparesis, the upper left arm being most affected, in the girl. MRI showed T2 hyperintense lesions in the cervical spine in the boy and multilevel T2 hyperintense myelitis with contrast enhancement in the girl. In both children, the anterior grey matter was predominantly affected. The girl additionally showed leptomeningeal and anterior nerve root enhancement. CSF demonstrated

pleocytosis of 186 (148 mononuclear cells) and 46 (45 mononuclear cells), respectively. Nasopharyngeal swabs were EV-positive by PCR in both children. Partial V2 sequencing followed by shotgun metatranscriptomics identified EV-D68 in the boy. In the girl, amplification-based sequencing and metatranscriptomics were both unsuccessful. Acute anti-inflammatory treatment using high-dose corticosteroids/plasma-exchange/IVIG in the boy and high-dose corticosteroids/IVIG in the girl led to minimal improvement. Both children still benefit from intense neurorehabilitation.

Discussion and conclusions

In any AFP it is of utmost importance to exclude Poliomyelitis by two consecutive stool samples which is reportable to the Federal Office of Public Health by the epidemic law. AFP occurring shortly after URTI is highly suspicious of an underlying EV-D68 infection. Identification of EV-D68 is difficult and requires early sampling of respiratory secretion and notification of the laboratory that EV-D68 is suspected. There is no established treatment and outcome is often dismal. EV-D68 is a communicable disease - awareness and immediate reporting to the SPSU is highly important for early identification of clustered cases. Establishing a fast-track diagnostic path similar to other countries would improve epidemiological management and avoid potentially unnecessary treatment escalation in the individual patient.

P 42

Malaria in Refugee Children Resettled to a Holoendemic Area of Sub-Saharan Africa

Hauser Manuela[1,2], Kabuya Jean-Bertin B.[3], Mantus Molly[4], Kamavu Luc K.[5], Sichivula James L.[3], Matende Wycliffe M.[6], Fritschi Nora[7,8], Shields Timothy[9], Curriero Frank[9], Kvit Anton[9], Chongwe Gershom[4], Moss William J.[9,10], Ritz Nicole[7,11,12,13]*, Ippolito Matthew M.[10,14]*, * Co-senior authors

[1]Faculty of Medicine, University of Basel, Switzerland; [2]Children's Research Center, University Children's Hospital Zurich, Switzerland; [3]Department of Clinical Sciences, Tropical Diseases Research Centre, Ndola, Zambia; [4]Johns Hopkins Bloomberg School of Public Health, Baltimore, Maryland, United States; [5]Office of Hospital Administration, Saint Paul's General Hospital, Nchelenge, Zambia; [6]United Nations High Commissioner for Refugees, Country Representation Office, Lusaka, Zambia; [7]Mycobacterial and Migrant Health Research Group, University of Basel Children's Hospital, Switzerland; [8]Department of Clinical Research, University of Basel, Switzerland; [9]Department of Epidemiology, Johns Hopkins Bloomberg School of Public Health, Baltimore, Maryland, United States; [10]Johns Hopkins Malaria Research Institute, Johns Hopkins Bloomberg School of Public Health, Baltimore, Maryland, United States; [11]Infectious Disease and Vaccinology Unit, University Children's Hospital Basel, University of Basel, Switzerland; [12]Department of Pediatrics, The Royal Children's Hospital Melbourne, The University of Melbourne, Australia; [13]Department of Paediatrics and Paediatric Infectious Diseases, Children's Hospital, Lucerne Cantonal Hospital, Lucerne Switzerland; [14]Division of Clinical Pharmacology and Division of Infectious Diseases, Department of Medicine, Johns Hopkins University School of Medicine, Baltimore, Maryland, United States

Background

Malaria is a leading cause of morbidity and mortality in refugee children in high transmission parts of Africa. Characterizing the clinical features of malaria in refugees can inform approaches to reduce its burden.

Methods

The study was conducted in a high-transmission region of northern Zambia hosting Congolese refugees. We analyzed surveillance data and hospital records of children with severe malaria from refugee and local sites using multivariable regression models and geospatial visualization.

Findings

Malaria prevalence in the refugee settlement was similar to the highest burden areas in the district, consistent with the local ecology and leading to frequent rapid diagnostic test (RDT) stockouts. We identified 2,197 children hospitalized for severe malaria during the refugee crisis in 2017 and 2018. Refugee children referred from a refugee transit center (n = 63) experienced similar in-hospital mortality to local children and presented with less advanced infection. However, refugee children from a permanent refugee settlement (n = 173) had more than double the mortality of local children ($p < 0.001$), had lower referral rates, and presented more frequently with advanced infection and malnutrition.

Interpretation

Malaria outcomes were more favorable in refugee children referred from a highly outfitted refugee transit center than those referred later from a permanent refugee settlement. Refugee children experienced higher in-hospital malaria mortality due in part to delayed presentation and higher rates of malnutrition. Interventions tailored to the refugee context in the early- and particularly late-term refugee setting are required to ensure capacity for rapid diagnosis and referral to reduce malaria mortality.

P 43

SARS-CoV-2 seroprevalence in subjects aged 0-16 years in Canton Ticino: preliminary results

Mazzara C[1], Bassi J[2], Simonetti GD[1,3], Vanoni F[1,3], Piccoli L[2], Kottanattu L[1,3]

[1]Pediatric Institute of Southern Switzerland, Ente Ospedaliero Cantonale, Ospedale San Giovanni, Bellinzona, Switzerland; [2]Humabs BioMed SA, a subsidiary of Vir Biotechnology, Bellinzona, Switzerland; [3]University of Southern Switzerland, Lugano, Switzerland

Introduction

During the last two years, the spreading of SARS-CoV-2 pandemic has represented the most important topic of public health worldwide. Although initial studies showed that children were marginally affected by SARS-CoV-2, the spreading of VOCs (variant of concern) is reshaping this observation, with an increasing number of children testing positive. The number of infected children remains unclear, also due to their asymptomatic and paucisymptomatic disease course, the variability of access to testing strategies and the changing of case definition.

Methods

Aim of this study is to define the prevalence of SARS-CoV-2 in the pediatric population (0-16 years of age) of Canton Ticino by seroconversion. The presence of highly specific antibodies reacting against SARS-CoV-2 was assessed using a high throughput, quantitative ELISA assay developed at Humabs BioMed. The study design includes a retrospective and a prospective part. After obtaining written consent, a retrospective analysis of stored blood samples was performed. Patients admitted from October 30, 2019 to outpatient clinics, and the emergency department and inpatient ward of the Pediatric Institute of Southern Switzerland of Bellinzona were enrolled. The reported data are the results of an interim analysis including samples collected until September 2021. Recruitment is ongoing and the final analysis is foreseen at the closure of the study planned in June 2022.

Results

A total of 666 samples from 506 patients were collected. Median age was 132 months (range 3-204). Male patients were 229 (45%). Prevalence of positive cases was 10.4% (53 patients). Only 2% of enrolled children resulted positive between November 2019 and April 2020, while incidence substantially increased between November 2020 and April 2021 (22%). Most positive cases occurred in females (64%) and the median age was 132 months (range 36-192). Symptomatic patients were 28 (52%), showing the typical manifestations of a viral infection: headache (18 pts), fever and cold (12 pts), sore throat (11 pts), asthenia (9 pts) and cough (8 pts). Additional information on symptoms, possible source of infection and public health measures were also collected.

Conclusion

Despite the high burden of COVID-19 disease experienced in Canton Ticino, our preliminary data show a low seroprevalence of SARS-CoV-2 in children, in line with the results from other published studies.

P 44

Lipshutz's acute vulvar ulceration and SARS-CoV-2: possible correlation?

Ridolfi A[1], Taverna A[1], Corigliano T[1], Ramelli GP[1], Ferrucci E[1]

[1]*Istituto Pediatrico della Svizzera Italiana, Ente Ospedaliero Cantonale, Switzerland*

Background

Vulvar ulcers are mostly caused by sexually transmitted microorganisms, like T. pallidum, HSV and, occasionally, HIV. When genital ulcers occur in not sexually active women and girls, Lipschutz's acute vulvar ulceration is the leading cause. This benign and self-remitting condition is a non-sexually acquired condition, generally related to flu-like infections or mononucleosis syndrome. A concurrent EBV infection occurs in nearly 50% of cases. Local hygiene, ulcers care and pain control are the mainstay of management of this condition.

Case study

A 14-year-old-girl, not sexually active, arrives to the emergency referring since four days severe pain in the genital area, enhanced during voiding and associated with vulvar ulcers. No other symptoms are referred. A two days therapy with acyclovir has shown to be ineffective. Local inspection of external genitalia shows 4 ulcerated lesions <5 mm, with no active bleeding and a slight oedema of the right labium minus. Hymen shows to be intact. A diagnosis of Lipschutz ulcers is hypothesize. Some tests are thus performed: microbiological cultures of the lesions are negative for HSV and VZV. Blood panel shows mild isolated lymphopenia and CRP is 2 mg/L. Serologies for CMV, HBV, HCV, HIV, Toxoplasmosis and T. pallidum are negative. Serologies for EBV turn out positive for past infection. PCR for SARS-Cov-2 is otherwise positive. We set home symptomatic therapy with ibuprofen alternating with co-paracetamol, and cold-water vulvar irrigation for voiding. A spontaneous resolution takes place in ten days. Due to her moving to Italy and to problems related to non-recognition of swiss immunity documents, the girl had to take the third dose of vaccine (BioNTech/Pfizer) one month after healing. Four days later the ulcers recur in the same place, although with eased symptoms.

Conclusion

According with other literature cases, Covid-19 infection could represent a likely explanation for this clinical situation. The recurrence after vaccine represents a strong evidence for this hypothesis. We therefore suggest to always include Covid-19 infection in the lab tests from now on, while screening for Lipschutz ulcers. Although benign, this clinical picture is confirmed to be highly invalidating and do not respond to any specific therapy. The treatment of pain is critical. It is always important to consider whether pain management can be carried out at home, if hospitalization and eventual catheterization can be avoided.

P 45

Parapharyngeal Abscess due to Gemella morbillorum - do not underestimate microorganism of low pathogenicity

Hersch Kristina[1], Willi Bettina[1], Milenovic Sinisa[2]

[1]*Kantonsspital Graubünden, Kinder- und Jugendklinik;* [2]*Kantonsspital Graubünden, Hals-Nasen-Ohrenklinik*

Introduction

We present a case of extensive pansinusitis with parapharyngeal - and peritonsillar abscess, diffuse inflammation reaching the epiglottis and bacteremia due to Gemella morbillorum. This is a microorganism generally known for low toxicity and pathogenicity.

Case presentation

A 10 -year-old boy presented to our emergency department with strong pain in the oropharyngeal area. Two days before palatal arch braces were installed but due to pain removed except the fixation attached to the teeth. Febrile temperature the day before emergency visit was recalled. Clinical findings were minimal infraorbital erythema and an oral ulcer. Infectious parameters were elevated. He was started on intravenous Amoxicillin/Clavulanic acid after drawing blood cultures. Only few hours later the inflammation progressed severely to the infraorbital area and left palate arch with uvula displacement. No neurological deficits were observed. CT scan showed sinusitis maxillaris and ethmoidialis and phlegmonous lesion periorbital with erosion of lamina papyracea, further tonsillitis palatina with extensive phlegmonous infiltration of the parapharyngeal- and retropharyngeal space as far as the epiglottis. Despite complementary treatment with Clindamycin and Metronidazol clinical worsening of the inflammatory process, as well as the patients general condition was observed. Surgical treatment was warrant. In the blood culture Gemella morbillorum was identified. By echocardiography endocarditis was excluded. A second blood culture drawn after surgery remained negative. After intravenous Co-Amoxicillin and Clindamycin for 14 days, followed by a 7 day course of oral Clindamycin the patient gradually recovered.

Discussion

Infections caused by Gemella morbillorum in the pediatric age group are rare, but retropharyngeal, peritonsillar abscess and sinusitis has been described. Dental procedures is one of the predisposing factor in otherwise immunocompetent infants. However this case underlines that also in a previously healthy child Gemella morbillorum, can cause severe infection and – as described in literature – even death.

Conclusion

The pathogenicity of Gemella morbillorum should not be underestimated and should prompt treatment of appropriate length to avoid life threatening complications.

P 46

Lemierre's syndrome in a 10-year-old boy with suspected immunodeficiency

Mrabet-Deraoui I[1], Stoelers T[1], Callias C[1], Plebani M[2], Arlabosse T[3], Guilcher P[4], Laubscher B[1]

[1]*Department of Paediatrics – Réseau hospitalier Neuchâtelois (RHNe), Neuchâtel, Switzerland.;* [2]*Paediatric Infectious Diseases Unit – Lausanne University Hospital, Lausanne, Switzerland.;* [3]*Paediatric Immunology, Allergology and Rheumatology Unit, Department of Paediatrics, Department Woman-Mother-Child – Lausanne University Hospital, Lausanne, Switzerland.;* [4]*Service of Otorhino-Laryngology – Lausanne University Hospital, Lausanne, Switzerland.*

Case

A 10-year-old boy presented unilateral right cervical adenitis with unfavourable evolution. In 2 days, he developed fever, painful growing right cervical swelling, trismus, torticollis and respiratory dependent chest pain. Vital signs and cardiorespiratory auscultation were unremarkable. An inflammatory response was detected (CRP 184 mg/L, leucocytosis 15.4 G/L, neutrophilia 10.6 G/L with left shift). A neck/chest CT scan was performed to search for suppurative and thrombotic complications. It revealed right lateral cervical adenopathies complicated with a multiloculated abcess, thrombophlebitis of the right internal jugular vein and pulmonary septic emboli affecting all lobes. With the triad of cervical bacterial adenitis, thrombophlebitis of the right internal jugular vein and pulmonary septic emboli, Lemierre's syndrome (LS) was diagnosed. Cervical abscess drainage was performed. The bacteria Aggregatibacter aphrophilus was identified on operative pus sample (bacterial culture). Next Generation Sequencing (NGS) did not reveal the presence of fusobacterium species. Blood cultures were sterile. He received co-amoxicilline for 6 weeks (first 2 weeks intravenously) and no anticoagulation therapy. Evolution was favourable. It was his 3rd severe bacterial infection (basal right necrotic pneumonia at 9yrs, right cervical abscess at 8yrs). An immune deficiency was suspected: IgA deficiency was found with abnormal antipneumococcal vaccines response.

Discussion

LS is a rare and severe infectious disease (2018 worldwide incidence 1/1'000'000) that mainly affects healthy adolescents and young adults. It is defined by a triad of acute neck anaerobic infection, thrombophlebitis

of the internal jugular vein and distant septic emboli. Fusobacterium necrophorum is the most common pathogen. LS is a life-threatening but treatable disease. Standard treatment consists of surgical drainage and antimicrobial treatment. Anticoagulation is debated.

This case is remarkable due to the presence of A. aphrophilus, a bacterium of the HACEK group that usually causes endocarditis, brain abscess and osteomyelitis. An immunodeficiency might explain why our patient presented LS due to an atypical bacterium.

Conclusion

Neck infection with signs of local complication, deteriorating condition and chest pain must raise suspicion of LS. In this case, the patient presented a LS by A. aphrophilus, a bacterium that to the best of our knowledge has not been described in association with LS.

P 47

A red herring in an unusual presentation of a rare paediatric neuroborreliosis syndrome

Kapp A[1], Giagkou E[2], Mitrovic I[3], Stahr N[4], Ritz N[5], Büttcher M[5]

[1]General Paediatrics, Lucerne Childrens Hospital, Switzerland; [2]Paediatric Neurology, Lucerne Childrens Hospital, Switzerland; [3]Department of Microbiology, Cantonal Hospital of Lucerne, Switzerland; [4]Paediatric Radiology, Lucerne Childrens Hospital, Switzerland; [5]Paediatric Infectious Diseases, Lucerne Childrens Hospital, Switzerland

Introduction

Lyme disease is a common tick-bite associated infection presenting with signs and symptoms of different organ systems. Early neuroborreliosis, particularly in children, includes facial nerve palsy and subacute meningitis. Work-up includes evaluation of cerebrospinal fluid (CSF) and serology.

Case

A 12-year-old female presented in January 2022 with progressive (2 week) muscular weakness of the legs noticed by gait and bike-riding difficulties. No current or previous episode of fever or infection. No tick bite in the past. Symmetrical hyporeflexia (patellar and achilles) and weakness of 4,5/5 (MRC) of both thighs were noted. Nerve conduction velocity testing (ulnar, tibial, peroneal, sural) were normal. Cerebrospinal fluid (CSF): lymphocytic pleocytosis (101/uL), mild hypoglycorrhachia (Glucose 2.9mmol/L) and elevated protein (0.99 g/L). Inflammatory markers were unremarkable. Initial CSF-multiplex-PCR was positive for H. influenzae. MRI (brain & spinal cord) revealed isolated inflammation of the cauda equina. Treatment with i.v. immunoglobulines (0,5 mg/kg/d for 4d) and ceftriaxone (100mg/kg/d) were started. Follow-up-MRI revealed persistent inflammation of the cauda fibres and medullary cone, albeit without intramedullary lesions. Second CSF (3d post treatment) demonstrated similar cytological and chemical analysis. However, single H. influenzae PCR was negative and results of intrathecal B. burgdorferi antibody (IgG and IgM) production and CSF-CXCL-13 (173pg/ml (<50 pg/ml)) were positive.

Discussion

This child presented with B. burgdorferi associated lymphocytic meningoradiculitis (Bannwarth syndrome), which is a rare manifestation of neuroborreliosis in children. The unusual symptom complex (progressive weakness, hyporeflexia, lymphocytic pleocytosis, inflammation of the cauda equina) open a broad differential diagnosis, generating several diagnostic and empirical treatment modalities. The positive H. influenzae CSF-multiplex-PCR was considered likely false positive, but, until an alternative diagnosis was established, ceftriaxone was initiated. Cases of false positive H. influenzae within a CSF-multiplex-PCR panel have been described previously.

Conclusion

Bannwarth syndrome with absence of pain of the lower extremities or cranial nerve palsy is a possible presentation in children. Despite advances in laboratory testing, clinicians should be aware of potential red herrings such as false positive tests in multiplex-PCRs in CSF.

P 48

Augmented reality ultrasound support for intravenous lines in paediatric emergency care Introduction

Hillenbrand N[1], Maas S[2], Quinones Lozada CA[1]

[1]Clinic for Children and Adolescents, University Hospital Frankfurt, Germany; [2]Research and Development, SomaView GmbH, Bochum, Germany

Introduction

Augmented reality (AR) systems are being investigated for their uses and feasibility in clinical practice. In paediatric emergency care establishing intravenous lines (IVs) are one of the most basic skills but can be challenging due to the particular situation of young patients: small vessels, agitated patients and nervous parents often complicate the situation. Ultrasound (US) support has proven to be very helpful in these challenging cases.

Farshad-Amacker et. al. (2020) showed a potential benefit for AR US systems against conventional US guided interventions. Based on these results our phantom study aimed to investigate the feasibility of AR systems in establishing IVs.

Material and methods

The setup for this study consists of:

- US device (Esaote MyLab X8)
- AR glasses (Microsoft Hololens 2)
- AR system (SomaView SonoXR)
- A self-constructed vascular phantom (180x120x26mm; surgical tubing with 3 mm inner and 6 mm outer diameter; GELITA® Gelatine Type Ballistic 3)
- IV catheter (B Braun Vasofix Safety, 18G x 1 ¾'')

15 subjects with different levels of experience (3 groups with each 5 operators: students, assistant physicians, intensive care practitioners) established each 6 conventional US-guided IVs and 6 IVs using the AR glasses and AR system at the vascular phantom. Time to successful establishment of the IVs and the number of head turns (between phantom and US-screen) were recorded for each puncture.

Results

Using the AR system reduced the time for establishing an IV for assistant physicians (median [range], 12 sec [3-70] vs. 18 sec [5-59]) and intensive care practitioners (median [range], 5 sec [2-36] vs. 8 sec [4-17]) compared to conventional US-guidance. No time gain was found in the student group.

Concerning all groups less head turns were needed (median [range], 0 [0-2] vs. 1 [1-16]) when using the AR system.

Conclusion and outlook

The study confirms the feasibility of AR assistance in establishing IVs. Intensive care practitioners and especially assistant physicians were faster when using the AR system. The fact that no time gain was observed in the student group may indicate that a minimum of experience in establishing IVs is necessary to benefit from the advantages of an AR system.

Future studies will include more complex phantoms and human subjects to identify the full potential of AR assistance in establishing IVs.

P 49

Major trauma in children - take a close look at the craniocervical junction

Bürgin A[1], Willi B[1], Stricker S[2,3]

[1]Department of Pediatrics, Kantonsspital Graubünden, Chur, Switzerland; [2]Neurosurgical Unit, Kantonsspital Graubünden, Chur, Switzerland; [3]Department of Neurosurgery, University Hospital Basel, Switzerland

Introduction

In children traumatic spine injuries are typically located in the upper cervical segment. Diagnosis of potentially fatal occipitocervical distraction injuries is important and can be challenging. We report a case of a boy with

an atlantooccipital dislocation after a high velocity accident who presented with bilateral abducens nerve palsy.

Case report

A 11-year-old boy was admitted to the emergency room after a high velocity sledding accident while wearing a helmet. At admission he had oral bleeding, was hemodynamically stable and had a normal neurological exam. The initial CT showed mandibular fractures, a fracture of the femur and a retroclival hematoma. The patient underwent surgical stabilization of the femur fracture. Secondary clinical survey revealed a complete abducens nerve palsy on the left, and an incomplete palsy on the right. MRI showed, in addition to the retroclival hematoma, a dislocation of the tectorial membrane from the clivus and an anterior atlantooccipital ligament injury with consequent partial atlantooccipital dislocation. He was treated with a soft collar for neck immobilization and alternate patching of the eyes to prevent diplopia. At 4 month follow-up the abducens palsy completely recovered on both sides, leaving the boy without any neurological sequelae.

Discussion

Occipitocervical distraction injuries are more often seen in children due to relatively smaller occipital condyles, a more horizontally orientated atlantooccipital articulation and generally greater ligamentous laxity. Diagnosis can be challenging because of wide clinical presentation and difficult detection on CT. The abducens nerve is especially vulnerable to trauma due to its anatomic course. In occipitocervical distraction injuries it can be compressed by an associated retroclival hematoma, damaged by accompanying petrous bone fractures as well as shearing and contusion of the nerve or brainstem. Although most of these palsies (73%) show spontaneous recovery after six months, bilateral and complete palsy are predictors of a poorer outcome.

Conclusion

In patients with severe trauma and abnormalities on initial CT or presence of neurological deficits cerebral MRI is important. To look for occipitocervical distraction injuries the craniocervical junction should be included on MR-imaging. Abducens nerve palsy can be a warning sign for these occipitocervical distraction injuries, and usually recovers within 6 months.

P 50

miniDNA - minimal African neonatal dataset - A tool to understand, intervene and follow neonatal mortality and morbidity in low income countries.

Zala Z[1,2], Pfister R[1,2]

[1]Neonatology and Paediatric Intensive Care Unit, Geneva University Hospitals, Switzerland; [2]Geneva University, Switzerland

Introduction

Of the 5.2 million deaths of children under 5 in 2019, 2.4 million died in their first month of life, accounting for about 47% of child mortality. The proportion of neonatal mortality among child mortality has been increasing since 1990, when it was 40%, reflecting a slower decline. One third of neonatal deaths occur on the first day of life and three quarters during the first week of life. Prematurity, low birth weight, asphyxia, and infections are the leading associations with neonatal mortality. The neonatal period is vulnerable, yet a compact time-specific target for action.

Sub-Saharan Africa and South Asia have the world's highest neonatal mortality rates. Indeed, a new-born is 10 times more likely to die there than in a high-income country. However, most low- and middle-income countries (LMIC) do not have reliable data on neonatal mortality and morbidity, which hinders effective targeted actions.

There is an urgent need to fill these data gaps. Existing neonatal databases in high-income countries have already proven to be effective. However, they are inadequate for low- and middle-income countries.

Methods

Our miniDNA is a minimal, standardized database adapted to LMIC. It consists of 100 clinical, administrative, and structural variables that are generally available and representative for neonatal health to allow effective and anonymous inter-hospital quality comparison. 2-weekly callings and two local field-trips during the one-year pilot-testing of the miniDNA in 3 Burkina Faso centres targeted variable improvements in definition and pertinence.

Results

We included >2000 new-borns, improved variable definitions for 20 variables and generated an automatic quality control report for neonatal care that was individually shared with participating centres. We also determined a workload of 25 minutes per case with the present manual data entry into a spreadsheet.

Discussion

Our miniDNA is a standardize dataset specific for LMIC for comparison within hospitals. The standardize data format, remaining equivalent for 60% of items with high-income neonatal databases, allows south-south comparison and gives leverage for quality improvement.

Our targets now will be a simplified and faster, root-structured IT-based data entry, and increased data security for a broader use in additional African centres. We hope that the miniDNA will not only identify levers for action through peer-comparison, but give follow up for interventions.

P 51

Liddle's syndrome – a genetic form of hypertension

Schüpbach-Bucher BS[1,2], Köhler H[2], Simonetti GD[3]

[1]Department of Pediatric nephrology, Children's Hospital of Aarau, Aarau, Switzerland; [2]Children's Hospital of Aarau, Aarau, Switzerland; [3]Pediatric Institute of Southern Switzerland EOC, Ospedale San Giovanni, Bellinzona, Switzerland

Introduction

Arterial hypertension in children is more often secondary than in adults and search for the underlying cause should always be performed. The most common causes are renal, cardiac or hormonal diseases; however, an extremely rare group of genetic forms of hypertension, called Low-Renin-Hypertension, should be considered in cases of severe hypertension of unknown etiology.

Case report

A 13 years old girl with arterial hypertension, known for several years and treated with the calcium channel blocker amlodipine, was referred to the pediatric nephrology clinic. Hypokalemia (2.8mmol/l) and extremely low values for renin and aldosterone were detected in the biochemical analyses. Furthermore, family history was positive for severe arterial hypertension in young age. The mother was treated for arterial hypertension with a double pharmacological antihypertensive therapy for many years and she also had hypokalemia.

The steroid profile in the urine was completely normal, excluding some genetic forms of Low-Renin-Hypertension (AME, CAH type IV ore V, GRA). Of the two remaining forms, Gordon's syndrome and Liddle's syndrome, only the latter is associated with hypokalemia. Accordingly, the suspicion of Liddle's syndrome was very high. Liddle's syndrome is an autosomal dominant disease, caused by gain of function mutations in either the SCNN1A, SCNN1B or SCNN1G genes. These mutations increase the activity of the sodium channel ENaC in the distal nephron of the kidney, leading to sodium retention and secondarily to fluid retention and potassium elimination. Liddle's syndrome mimics the symptoms of mineralocorticoid excess, although with suppressed aldosterone and renin levels, ore liquorice abuse.

Genetic testing revealed a heterozygous mutation in the SCNN1B gene (c.1854dupC) in the girl and in the mother.

The treatment of Liddle's syndrome is very effective with the use of a potassium-sparing diuretic, such as amiloride (blocking the activity of ENaC), and the efficacy can be further enhanced with dietary salt restriction. The

antihypertensive treatment was therefore modified from amlodipine to amiloride and a normalization of blood pressure and of potassium values were observed.

Conclusion

Exact evaluation of arterial hypertension in children is crucial. In patients with a positive family history of arterial hypertension in young relatives, genetic forms of hypertension must be considered in order to know the exact etiology and to adapt pharmacological therapy.

P 52

Acute kidney injury without liver involvement after Paracetamol intoxication

Ramelli V[1], Goischke A[2]

[1]University Children's Hospital, Basel, Switzerland; [2]University Children's Hospital, Department of Nephrology, Basel, Switzerland

Background

Renal insufficiency with paracetamol overdose occurs in approximately 1-2% of adult patients and limited data suggests that associated nephrotoxicity may be more common in the pediatric population (1,2). Usually, though not always acute kidney injury (AKI) is accompanied or preceded by significant hepatic dysfunction (3).

The histological substrate is acute tubular necrosis of the proximal and distal tubules (3).

Case report

A 15-year-old girl was admitted to our pediatric emergency department after taking 10 tablets of 1g paracetamol three days before. She complained about abdominal pain, nausea and vomited three times. The patient suffers from severe obesity (BMI 39.5kg/m2). Her past medical history reveals a Borderline personality disorder with depressive episodes.

Vital signs were in the normal range except an elevated blood pressure (max. 155/78 mmHg) . Laboratory studies showed an increased serum creatinine value of 251 µmol/l and urea of 9.8mmol/l without any hepatic involvement.

Other causes of AKI were excluded. Intravenous fluid administration was necessary for 4 days. Serum creatinine started to improve after day 2. Blood pressure normalized after 3 days. The patient could be discharged in good general condition on day 7.

Renal function had completely normalized at follow up one month later.

Conclusions

Acute liver failure in paracetamol intoxication is well known but monitoring of kidney function tends to be neglected. Renal failure can occur later than liver failure (mean time 2-5 days with creatinine peaks at day 7). Renal function often recovers within 1 month.

There is no clear dose-toxicity relationship for paracetamol and nephrotoxicity as in paracetamol and hepatotoxicity. Renal toxicity may occur at lower doses and almost 1/3 of patients develop renal insufficiency in the absence of significant hepatotoxicity as seen in our patient (3).

References

Prescott LF: Paracetamol overdosage: Pharmacological considerations and clinical management. Drugs 1983, 25:290-314

Boutis K: Nephrotoxicity after acute severe acetaminophen poisoning in adolescents. J Toxicol Clin Toxicol. 2001;39(5):441-5. doi: 10.1081/clt-100105413. PMID: 11545233.

Von Mach MA: Experiences of a poison centre network with renal insufficiency in acetaminophen overdose: an analysis of 17 cases. Clin. Toxicol. 2005; 43:31-37

P 53

Episodic encephalopathy caused by D-Lactate- acidosis – A case report

Zürcher L[1], Andersag J[2], Leiber A[2], Brühl F[1], Glock K[3]

[1]Department of Pediatrics, Children's Hospital of Eastern Switzerland, Switzerland; [2]Department of Pediatric Neurology, Children's Hospital of Eastern Switzerland, Switzerland; [3]Department of Pediatric Gastroenterology and Nutrition, Children's Hospital of Eastern Switzerland, Switzerland

Background

D-Lactate acidosis is a rare complication in patients with short bowel syndrome and intestinal failure. Carbohydrate concentration increases postprandial in the bowel. Bacterial overgrowth is common in short bowel patients. In this microbiome carbohydrate fermentation might produce D-Lactate. This metabolic product is physiologically not found in humans. Increased D-Lactate leads to reversible metabolic acidosis and variable neurological symptoms.

Case presentation

A 25-month-old male was seen in the emergency department with sudden impaired awareness (GCS 11-15) and gait disturbances. His medical history showed small intestine atresia with apple peel syndrome leading to short bowel syndrome. He was on partial parenteral nutrition via Broviac Catheter and in the process of parenteral weaning and an increasing oral intake. Blood gas analysis showed metabolic acidosis with pH 7.26, base excess -11.3mmol/l and elevated anion gap 25.4mmol/l, L-Lactate was normal (0.7mmol/l). Further blood- and urine-tests showed no signs of ketoacidosis, infection, uraemia or intoxication. X-ray and CCT were unremarkable, as was the EEG in the symptom-free phase. After intravenous rehydration, there was spontaneous improvement, but episodes of ataxia still occurred. An MRI of the neurocranium revealed no pathologies. After eating a piece of cake, the boy presented with rapid deterioration, including reduced GCS, ataxia and severe metabolic acidosis. Due to the episodic neurological impairment, short bowel syndrome, severe metabolic acidosis and the dependency on food intake, D-Lactate acidosis was suspected. Therapy with sodium bicarbonate and reduced intake of carbohydrates led to normalization of all symptoms, which confirmed the diagnosis.

Conclusion

Knowing the clinical presentation of D-Lactate-acidosis as a rare neurological complication in patients with short bowel syndrome assists in diagnosing quickly, avoids unnecessary diagnostics and prevents further crises.

P 54

Disease of a thousand faces – Chorea as presenting feature of systemic lupus erythematodes

Vomsattel S[1], Bolt I[2], Pospieszny K[3], Caccia J[4], Bigi S[5,6]

[1]Department of Pediatrics, Bern University Hospital, Inselspital, University of Bern, Switzerland; [2]Division of Pediatric rheumatology, Department of Pediatrics, Bern University Hospital, Inselspital, University of Bern, Switzerland; [3]University Institute of Diagnostic and Interventional Neuroradiology, University Hospital Bern, Inselspital, University of Bern, Switzerland; [4]Division of Pediatric nephrology, Department of Pediatrics, Bern University Hospital, Inselspital, University of Bern, Switzerland; [5]Institute of Social and Preventive Medicine, University of Bern, Bern, Switzerland; [6]Department of Neurology, Bern University Hospital, Inselspital, University of Bern, Switzerland

Background

Chorea is a rare extrapyramidal hyperkinetic movement disorder resulting from alterations in the basal ganglia circuit due to (neuro)vascular, metabolic-toxic, infectious or autoimmune aetiologies. Acute onset chorea is a symptom of potentially life-threatening, usually secondary inflammatory brain diseases (IBrainD) due to an underlying systemic disease, requiring immediate diagnosis and treatment. We report on a 14-year old girl with acute onset chorea as the presenting symptom of systemic lupus erythematodes (SLE).

Case presentation

A 14-year-old girl previously known for an idiopathic generalized epilepsy, learning disability and selective mutism presented to the emergency department with acute onset of involuntary movements in all four limbs. Clinical examination revealed severe choreatic movements, a skin rash and an altered level of consciousness (GCS 9) requiring admission to ICU. MRI of the brain showed multiple, bilateral punctiform ischemic lesions supra- and infratentorial, mainly in the subcortical white matter and the centrum semiovale. 4-channel EEG was normal. Extensive laboratory workup showed anemia (99 g/L), thrombocytopenia (80 G/L), elevated ESR (50 mm/h), antinuclear antibodies (ANAs) > 1:1280, dsDNA-antibodies > 379 U/mL, antiphospholipid-antibodies (anti-Cardiolipin-AK IgG 98.2 CU, anti-b2-Glycoprotein IgG 344.2 CU) and hypocomplementemia (C3 0.29 g/L, C4 0.01 g/L). Results from the CSF and coagulation assessment were normal. The diagnosis of a SLE was made. Clinically there were no signs for joint or other organ involvement. A therapy with high dose steroids for 5 days (1g/d) with tapering afterwards was initiated, leading to rapid improvement of choreatic movements. After a few days the girl developed macrohaematuria and proteinuria. A kidney biopsy was performed showing level III (A) lupus nephritis. Treatment was continued according to the Euro-Lupus Protocol, implementing cyclophosphamide (500 mg/dose every 2 weeks), hydroxychloroquine and prophylactic anticoagulation. Complete recovery of neurological and dermatological symptoms was documented within two weeks.

Conclusion

Autoimmune aetiologies are the most frequent cause of acute onset chorea in previously healthy children. Immediate neuroimaging is important to rule out structural causes and identify potentially treatable conditions in any child presenting with acute onset chorea.

P 55

Acute cerebellitis in an Adolescent after SARS-Cov-19 Infection: A case report

Schneider N[1], Stampfli M[1], Gessler P[1]

[1]*Department of Pediatrics, Cantonal Hospital Münsterlingen, Switzerland*

We report a 15-year-old boy presenting at our emergency department with acute onset of dysarthria and prior headache, rotary vertigo, nausea, vomiting and gait disturbance for 2 days. 18 days earlier he was tested positive for SARS CoV-19. Physical examination showed pronounced dysarthria, nystagmus, absent reflexes, dysmetria and ataxia. Laboratory findings showed relative lymphocytosis with a negative C-reactive protein, normal coagulation and elevated liver enzymes (AST, ALT, GGT). Drug screening was negative. First suspicion was postinfectious cerebellitis and he was admitted to IMC for observation. Subsequently, vomiting frequency rose, hence cMRI was performed, but showed no abnormalities. Miller-Fisher syndrome was suspected, however CSF examination showed no cytoalbumin dissociation, but discrete mononuclear cell count elevation. Ceftriaxone and acyclovir were administered empirically. In addition, intravenous immunoglobulin and corticosteroids were given to treat viral cerebellitis/encephalitis. Due to suggestive atypical lymphocytes in the blood count, EBV testing was performed and serologies were positive. Multiple CSF viral/bacterial testings (Borrelia burgdorferi, Enterovirus, VZV, HSV 1/2) showed slightly positive EBV (PCR), congruent with blood results. Additionally, SARS-CoV-19 IgG were positive. The patient's condition barely improved, a follow up cMRI showed no changes. Mobilization was fostered with a wheel chair and physical therapy. After two and a half weeks, he was discharged to pediatric neurorehabilitation in order to improve dysarthria, ataxia and address neuropsychological abnormalities. Two months after the diagnosis he is walking without aids but still shows trunk ataxia and dysarthria. He is still making constant progress and will hopefully recover completely.

Neurological manifestation of EBV infection is extremely rare with the majority of cases described in children. Nevertheless, some neurological manifestations have already been described including encephalitis, cerebellitis, meningitis, transverse myelitis, and Guillain-Barré syndrome. These manifestations can occur alone or in the setting of infectious mononucleosis.

In our case a co-infection of SARS-Cov-19 and EBV or a reactivation of EBV due to SARS-Cov-19 is possible as cause of the cerebellitis. Treatment options were fully employed, but the recovery presented a slow course.

P 56

Incontinentia pigmenti in a newborn girl with focus on potential neurologic complications and their management

Zimmermann M[1], Beiner M[1], Dysli C[2], Zandi S[2], Häfliger S[3], Bürgler C[3], Sanchez-Albisua I[1]

[1]*Department of Neuropediatrics, Inselspital University Hospital Bern, Switzerland;*
[2]*Department of Ophthalmology, Inselspital University Hospital Bern, Switzerland;*
[3]*Department of Dermatology, Inselspital University Hospital Bern, Switzerland*

Introduction

Incontinentia pigmenti is a rare x-linked multisystemic disorder. In addition to the characteristic skin findings, severe complications in eye and central nervous system may occur, predominantly in the first months to years of life. The authors report a rare case of Incontinentia pigmenti in a newborn girl, from the neuropediatric point of view.

Case description

We present the case of a newborn girl who arrived at the emergency department at the age of 7 days with a rapidly progressive skin rash. She was in good general condition, always afebrile and drinking well. Primarily antibiotic treatment was started on suspicion of impetigo due to the yellow crusts and vesicles. Maternal serology before birth, blood cultures as well as immunodeficiency tests showed no findings. Due to the typical distribution pattern of the skin findings along the lines of Blaschko, as well as the typical laboratory finding with peripheral eosinophilia, the presence of Incontinentia pigmenti was highly suspected. A skin biopsy revealed findings compatible with the suspected diagnosis. Ophthalmologic examination revealed spotty hemorrhages of the retina and avascular areas. The initial neurological examination was without any findings and genetic results are still pending at this time. Symptomatic treatment of skin lesions with Octenisan baths and Cicalfate was initiated as well as intensive follow-up in dermatology, ophthalmology and neuropediatrics.

Discussion

This case shows a classic cutaneous presentation of Incontinentia pigmenti in a newborn girl. One pathophysiological hypothesis is mutations in the NEMO/IKBKG gene leading to impaired endothelial apoptosis and altered angiogenesis resulting in vascular damage, stroke and further neurological complications, e.g. epilepsy, hemiparesis, mental retardation and microcephaly. These may occur within the first months of life and can be associated with ophthalmological findings. According to literature strokes usually present bilaterally and may be recurrent in some cases. Early diagnosis and establishment of frequent multidisciplinary follow-up are important for the prognosis. There is no causal therapy, although a recent study showed first successes of targeted gene therapy in mouse models. Fortunately, no neurological abnormalities have been detected in our patient so far.

P 57

Assessment of topical corticosteroid fear in parents of children with atopic dermatitis is a key element for successful therapy: An observational study

Brunner Corinne[1,5], Theiler Martin[2,5], Znoj Hansjoerg[3], Schwieger-Briel Agnes[2,5], Luchsinger Isabelle[2,5], Weibel Lisa[2,5], Seliner Brigitte[4,5]

[1]Pediatric Skin Center, Skin- and Wound management, University Children`s Hospital of Zurich, Zurich, Switzerland; [2]Pediatric Skin Center, Dermatology Department, University Children`s Hospital of Zurich, Zurich, Switzerland; [3]Department of Health Psychology and Behavioural Medicine, Institute of Psychology, University of Bern, Bern, Switzerland; [4]Department of Nursing science, University Children`s Hospital of Zurich, Zurich, Switzerland; [5]Children`s Research Center, University Children`s Hospital of Zurich, University of Zurich, Zurich, Switzerland

Objectives

Atopic Dermatitis (AD) is the most common chronic, pruritic inflammatory skin disease in children. Mainstay of treatment of AD are basic skin care and the use of topical corticosteroids (TCS). Despite their safety and effectiveness, the use of TCS is often met with worries by parents. Fear of TCS has implications for patient adherence and treatment outcomes in AD. Pediatricians as primary care providers are often the first physicians to diagnose and manage children affected by AD and play an important role in shaping parents' perceptions of TCS. Therefore, the aim of our study was to investigate parental TCS fear and disease severity of children affected by AD, who were referred from primary care providers in our Pediatric skin center.

Methods

We conducted an observational study with a cross-sectional design. Children affected by AD and their parents, who were referred to our department, were recruited consecutively for this study. Inclusion criteria were children aged 0 to 5 years, outpatient treatment, sufficient German language skills, and informed consent to the project. The primary outcome measure was the extent of fear of TCS evaluated with the "Topical Corticosteroid Phobia Score" (TOPICOP) with a score range from 0 to 36. A higher score means more TCS fear. Secondary outcome measure was disease severity assessed with the „Scoring atopic dermatitis" (SCORAD).

Results

A total of 40 patients was enrolled. Most children suffered from mild AD and were under 2 years of age. Daily skin care was well established. However, TCS fear among parents were present. The average mean score of the TOPICOP in our study was 18.08 ±7.06. Almost three quarters of the parents (72.5%) believed, that TCS could affect the future health of their child and stated that they waited as long as possible before applying TCS. 67.5% of parents stated that they would stop the treatment as soon as possible.

Discussion

Our results show that fear of TCS is common in parents of children with AD. We recommend to assess and discuss TCS fear at an early stage in the management of pediatric AD with the aim to improve outcomes and avoid undertreatment. Primary care providers play a central role in managing pediatric AD. They are in the best position to proactive address and discuss parental TCS fear and to provide TCS specific education. A recent study showed that the use of educational videos with the method of storytelling is highly beneficial and effective in reducing TCS fear.

P 58

SPARK: Supporting Parents of a Newborn with an Inborn Error of Immunity (IEI) by Raising their Illness Knowledge to Enhance Family Management

Exploring the experience of parents whose newborn is diagnosed with an inborn error of immunity (IEI) to develop an advanced practice nurse (APN) psycho-educational intervention based on support needs to enhance family management

Luck Patricia[1,3], Pachlopnik Schmid Jana[1,3], Schlüer Anna-Barbara[2,3]

[1]Department of Immunology, University Children's Hospital Zurich, Switzerland; [2]Department of Nursing Science, University Children's Hospital Zurich, Switzerland; [3]Children's Research Center CRC, University Children's Hospital Zurich, Switzerland

Background

Since 2019 inborn errors of immunity (IEI) are part of the Swiss newborn screening (NBS). The unexpected diagnosis is shocking for parents. They are confronted with a rare disease during the neonatal part and have to acquire skills in illness management. Chronic diseases diagnosed in NBS impact an increased need for information on the disease and its management, especially in the first weeks after diagnosing it. Information is helpful for a family management, which leads to better outcomes for patients. The impact of the diagnosis IEI on families and their family management is unclear. The goal of this research is to establish how families manage IEI within the Family Management Style Framework.

Methods

An exploratory sequential mixed method design is planned. This constitutes in five parts: a) an integrative literature review, b)couples Interviews of affected parents, which will be analysed by Constant Comparative Analysis to explore each couples experience and qualitative comparative analysis comparing the couples for factors influencing the family management style, c) developing a psychoeducational intervention based on the results from part one and two, using the Intervention Mapping Method, d) testing the intervention in a pilot feasibility study and comparing the outcomes to a historic control group, and e) integrating the qualitative and quantitative data in a joint display.

Significance

Parents of a newborn diagnosed with an IEI in NBS are in a highly vulnerable situation. A need-based psychoeducational intervention supporting them in the first few weeks after the diagnosis, should enhance their family management and therefore reduce parental stress and improve the child's therapy and clinical outcome.

P 59

Pediatric E.R.: from a quality survey to a new organization of the service

De Rosa v[1,4], Bassi d[4,4]

[1]Istituto pediatrico Svizzera Interna; [2]ente ospedaliero Cantonale; [3]ORBV; [4]Bellinzona

Introduction

The Pediatric Emergency Department of the San Giovanni Hospital in Bellinzona is a 24/7 service and hosts more than 10'000 patients per year. Our mission is to identify the severity of the patient's condition as early as triage and then to provide quality care to each patient. A quality survey based on E.R. user perceptions was conducted in January 2020. From the analysis of that survey, two critical issues emerged: the length of stay in the Emergency Department and the pediatric pain management. Our project now aims to improve processes, patient care and quality delivered to each child.

Methodology

To address these critical issues emerged from the survey a multidisciplinary workgroup was created to address four main processes: 1. Time to

Doctor according to patient severity during triage ; 2. Medical Team Evaluation 3. pediatric pain management; and 4. inpatient discharge by early afternoon. We made some changes in the working life of the pediatrics team, including Medical Team Evaluation, expedited admission to the Emergency Department, and design of increasingly simple and immediate electronic medical records. Interprofessional pain assessment training was implemented for the entire nursing and medical team. A multidisciplinary work has allowed to reorganize the rounds in the ward to allow the discharge of patients before 1 pm: in this way it becomes possible to admit in the ward as quickly as possible patients requiring hospitalization coming from the Emergency Department. Then, each week the medical and nursing teams meet to review the previous week and discuss results and possible improvements.

Results

Overall, the processes implemented have shown some improvements. The time spent in the emergency department has decreased: 65% of patients are discharged within 90 minutes of arrival. Pain management has significantly improved (from 0% to 100%). Currently 80% of admitted patients are discharged by 1 pm. As a result, the time spent in the emergency department before an inpatient stay has decreased to 130 minutes.

Conclusion

This project has immediately shown positive results, but it is necessary to continue along this path for a continuous improvement of the care of the pediatric patient. Our internal policy is to improve every day for the well-being of our young patients.

P 60

The landscape of pediatric Diamond Blackfan anemia in Switzerland: genotype and phenotype characteristics

Vogel N[1, 7], Schmugge M[2], Renella R[3], Waespe N[4, 5, 6], Hengartner H[7]

[1]Department of Pediatrics, Cantonal Hospital Graubünden, Chur, Switzerland; [2]University Children's Hospital, Zurich, Switzerland; [3]Department of Pediatrics, Lausanne University Hospital CHUV, Lausanne, Switzerland; [4]Division of Paediatric Oncology and Haematology, Department of Paediatrics, Inselspital, Bern University Hospital, Bern, Switzerland; [5]CANSEARCH Research Platform for Paediatric Oncology and Haematology, Department of Pediatrics, Gynecology and Obstetrics, Faculty of Medicine, University of Geneva, Geneva, Switzerland; [6]Childhood Cancer Research Group, Institute of Social and Preventive Medicine, University of Bern, Bern, Switzerland; [7]Hematology/Oncology Department, Pediatric Hospital of Eastern Switzerland, St. Gallen, Switzerland

Diamond Blackfan anemia (DBA) is a rare inherited bone marrow failure syndrome (IBMFS) characterized by a normochromic macrocytic anemia and congenital malformations. DBA is caused mainly by genetic mutations in large (RPL) or small ribosomal subunit genes (RPS). Clinical differences between genotypes are insufficiently understood and data from Switzerland is lacking. The aim of the study published in the European Journal of Pediatrics in June 2021 was to assess clinical features, treatment strategies, and genotypes in the Swiss DBA population.

We retrospectively reviewed medical charts of pediatric patients with DBA in Switzerland and stratified patients by RPL versus RPS mutations.

We report 17 DBA patients in Switzerland who were all genetically investigated. In our cohort, patients showed a wide spectrum of clinical presentations and treatment needs. We found a high proportion of physical malformations (77%) including lower limb (17%) and anorectal (12%) malformations. The two patients with anorectal malformations presented both with antepositioning of the anus needing surgery within the first 15 months of life. One of these patients had sphincteric dysfunction, the other coccygeal agenesis. Fourteen patients had a pathogenic mutation in a known DBA gene, two patients harbored a likely pathogenic mutation in the RPL17 gene, which is not commonly associated with DBA. We found that included patients with an RPL mutation had more frequently malformations but a milder form of anemia compared to patients with an RPS mutation.

We illustrate the wide clinical and genetic spectrum of DBA in Switzerland. Our findings highlight the need to take this diagnosis into consideration in patients with severe anemia but also in patients with mild anemia when malformations are present. Lower limb and anorectal malformation extend the spectrum of DBA-associated malformations. New genes should be explored to genetically classify more patients that were previously diagnosed only clinically as illustrated by the reported patients with possibly pathogenic RPL17 gene variants. Our report highlights the need for national and international collection of these patients with rare diseases to increase knowledge on long-term outcomes.

P 61

Risk factors for cancer-related fatigue in adult survivors of childhood cancer: A report from the CardioOnco Study

Sláma T[1,2], Belle FN[1,3], Christen S[4], Hägler-Laube ES[5], Kuehni CE[1,6], Rössler JK[6], Strebel S[1,7,8], von der Weid NX[9], Schindera C[1,9]

[1]Childhood Cancer Research Group, Institute of Social and Preventive Medicine, University of Bern, Switzerland; [2]Graduate School for Cellular and Biomedical Sciences, University of Bern, Switzerland; [3]Center for Primary Care and Public Health (Unisanté), University of Lausanne, Lausanne, Switzerland; [4]Department of Health Sciences and Medicine, University of Lucerne, Lucerne, Switzerland; [5]Department of Internal Medicine, Cantonal Hospital Baden, Baden, Switzerland; [6]Pediatric Oncology, Inselspital, Bern University Hospital, University of Bern, Switzerland; [7]Graduate School for Health Sciences, University of Bern, Switzerland; [8]CANSEARCH research platform in paediatric oncology and haematology, University of Geneva, Geneva, Switzerland; [9]Division of Pediatric Oncology/Hematology, University Children's Hospital Basel, University of Basel, Basel, Switzerland

Background

Cancer-related fatigue (CRF) is a distressing late effect in childhood cancer survivors (CCS) with a prevalence between 10-85%. There is little or conflicting evidence on its risk factors. We aimed to describe the prevalence of CRF in adult CCS and assess its risk factors.

Methods

As part of the CardioOnco study, we invited adult 5-year CCS treated at Inselspital Bern between 1976-2015 to a cardiooncological outpatient clinic and sent them questionnaires. We assessed fatigue with two validated instruments, the Checklist Individual Strength (CIS) subjective fatigue subscore (asking about fatigue during last 2 weeks; range 8-56) and the Visual Analog Scale (VAS; asking about fatigue at the current day; range 0-100). Increased fatigue was defined as CIS score 27-35 and VAS score ≥70. We collected information on previous cancer treatment and medical history and calculated β coefficients for the association between CRF scores and potential risk factors using multivariable linear regression adjusting for sex and age.

Results

We included 158 CCS (participation rate 29%) with a median age at study of 33 years (interquartile range 26-38). We found that 19% of CCS had increased fatigue with CIS and 11% with VAS. Increased fatigue based on the CIS subjective fatigue subscore was associated with female sex (β 3.0, 95% CI 1.0 - 4.9, $p = 0.003$), sleep disturbance (β 5.2, 95% CI 3.1 - 7.2, $p < 0.001$), endocrine abnormality (β 5.0, 95% CI 2.8 – 7.2, $p < 0.001$), history of both cranial and heart-directed radiotherapy (β 8.1, 95% CI 4.2 – 11.9, $p < 0.001$), cumulative dose of heart-directed radiotherapy ≥ 35Gy (β 5.7, 95% CI 2.6 – 8.8, $p = 0.002$), and cumulative dose of cranial radiotherapy ≥ 35Gy (β 8.0, 95% CI 4.5 – 11.4, $p < 0.001$). Same risk factors were identified based on VAS.

Conclusion

We found that one fifth of adult CCS experiences increased fatigue. Female CCS with history of cranial and heart-directed radiotherapy suffering from endocrine or sleep problems would profit from screening for CRF and further referral for counselling with a specialist.

P 62

Chronic microcytic anemia – not always caused by iron deficiency or a hemoglobinopathy. Diagnosis of two non-related children with congenital sideroblastic anemia (CSA)

Niedzwietzki K[1,2,3], Hengartner H[1,2,3]

[1]Department of Hematology/Oncology; [2]Ostschweizer Kinderspital St. Gallen; [3]Switzerland

The congenital sideroblastic anemias (CSAs) are a heterogeneous group of inherited disorders of erythropoiesis characterised by pathologic deposits of iron in the mitochondria of developing erythroblasts. Mutations of the mitochondrial glycine carrier SLC25A38 cause the most common autosomal recessive form of CSA, leading to an insufficient heme biosynthesis. However, the disease is still very rare, there being fewer than 70 reported families in the literature. Typical clinical presentation is a chronic microcytic anemia with reticulocytopenia with a first presentation mostly in the neonatal period or early infancy. In general, the patients do not respond to pyridoxine supplement and require transfusions chronically, necessitating iron chelation in due course. The only curative treatment so far is an allogeneic stem cell transplantation.

We report on the clinical course of two patients, both presenting at 6 months of age with persistent microcytic hyporegenerative anemia, refractory to iron substitution. After exclusion of other possible causes for this type of chronic anemia, like hemoglobinopathy/thalassemia and even less common causes like iron refractory iron deficiency anemia (IRIDA) or bone marrow failure syndrome, diagnosis of CSA was made by performing molecular genetic analysis. Both patients were diagnosed with a homozygote SLC25A38 mutation and now require regular blood transfusions.

These case reports confirm the importance of further diagnostic exams including genetic analysis when confronted with a chronic microcytic, hyporegenerative anemia without abnormalities in other organs. Especially when anemia presents at an early age (neonatal period, early childhood) a congenital disorder should be considered.

P 63

Treatment of NF1-associated Optic Pathway/Hypothalamic Gliomas in Patients with Diencephalic Syndrome

Weiser Annette[1], Hengartner Heinz[2], Kottke Raimund[3], Gerber Nicolas U[1], Grotzer Michael A[1], Guerreiro Stucklin Ana S[1]

[1]Department of Oncology, University Children's Hospital Zurich and Children's Research Center, Zurich, Switzerland; [2]Department of Oncology, Pediatric Hospital of Eastern Switzerland, St. Gallen, Switzerland; [3]Department of Diagnostic Imaging, University Children's Hospital Zurich, Zurich, Switzerland

Diencephalic syndrome (DS) is a rare symptom complex associated with tumors in the hypothalamic region mostly seen in children with optic pathway/hypothalamic gliomas (OPHG). Children with Neurofibromatosis type 1 (NF1) have predisposition to develop OPHG. As the predisposition for development of OPHG in NF1 patients is well known and 15-25 % of NF1 patients are diagnosed with OPHG in childhood or adolescence, these patients usually undergo regular ophthalmological assessments to identify changes in visual function as screening for OPHG.

We describe the clinical presentation and course in three girls with NF1 presenting with diencephalic syndrome as first symptom of OPHG. A seven year old girl with known familial NF1 was referred to pediatrics because of weight loss of 9 kg and emaciation. As part of the investigations a cranial MRI was done and showed a chiasmatic-hypothalamic tumor with mild hydrocephalus. A two year old girl with known familial NF1 and unexplained weight loss of 2 kg was referred for a cranial MRI which showed a large tumor in the chiasmatic-hypothalamic region without any signs of increased intracranial pressure. An infant was diagnosed with failure to thrive with weight below the third percentile at six months of age. She was followed by neurology because of NF1 and had cranial imaging at age two with finding of a hypothalamic-chiasmatic-optic pathway glioma. All three girls were started on chemotherapy with weekly vinblastine and showed normalization of weight percentiles after six months of treatment with decrease in tumor size. Yearly ophthalmological assessment is the primary screening tool for OPHG. But all of these patients didn't have any visual impairment at time of diagnosis of OPHG and the tumors were diagnosed because of the presentation with DS.

As DS is a potentially life-threatening symptom, a rapid response to treatment is highly desirable. Therefore, treatment decisions are even more challenging in patients with DS. Because of the rarity of DS in patients with NF1-associated OPHG and few reported cases, finding the right first-line treatment for these patients is challenging. Early recognition of the symptoms is key to make the diagnosis early and start treatment as soon as possible. It will be helpful to learn more about the treatment decisions by reporting more cases like those three.?

P 64

Hereditary thrombotic thrombocytopenic purpura presenting as atypical HUS (Hemolytic Uremic Syndrome) in identical twins

Bobot Nikola[1], Hengartner Heinz[2], Laux Regula[3], Tarasco Erika[4]

[1]Department of Adolescent Medicine, Children's Hospital of Eastern Switzerland St.Gallen; [2]Department of Hematology/Oncology, Children's Hospital of Eastern Switzerland St.Gallen; [3]Department of Nephrology, Children's Hospital of Eastern Switzerland St.Gallen; [4]Department of Hematology and Central Hematology Laboratory, Inselspital Bern

Hereditary thrombotic thrombocytopenic purpura (hTTP), also known as Upshaw-Schulman syndrome (USS) is a rare life-threatening disease, presenting with fever, mild gastrointestinal symptoms and sometimes with acute renal failure and cerebrovascular involvement. Cause for it is a severe congenital deficiency of ADAMTS13, a von Willebrand factor (VWF)-cleaving protease, resulting in the presence of large forms of VWF in the circulation, leading to intravascular platelet clumping and thrombotic microangiopathy. Usually there are environmental factors such as infections or medications which can trigger an episode.

We report on the clinical course of twin brothers with newly diagnosed hereditary TTP, twin A initially diagnosed with a pneumococcal associated haemolytic uremic syndrome (HUS) and twin B with haemolytic uremic syndrome. Both of them had a very similar clinical course with a HUS triad (non-immunological haemolytic anaemia with fragmentocytes, thrombocytopenia, acute renal failure), but in addition neurological symptoms, such as excessive sleepiness and confusion which were not fully explained with the above described diseases. Diagnosis of TTP has been made by measuring ADAMTS 13 activity, which in both boys was below 5%. There were no inhibitors against ADAMTS 13, leading to the suspicion of hereditary TTP, which finally has been proven performing molecular genetic analysis.

This case report confirms the importance of the analysis of the ADAMTS13 activity and its inhibitor in patients who have episodes of TTP, with a very low platelet count and sometimes without the classic biochemical signs of hemolysis.

P 65

No effect of lidocaine on ciliary beating frequency and pattern of human nasal epithelial cells in vitro

Savas Sibel T[1, 2], Tschanz Stefan A[3], Latzin Philipp[1, 2], Müller Loretta[1, 2], Casaulta Carmen[1, 2]

[1]Division of Paediatric Respiratory Medicine and Allergology, Department of Paediatrics, Inselspital, Bern University Hospital, University of Bern, Switzerland.; [2]Department of BioMedical Research (DBMR), University of Bern, Switzerland,; [3]Institute of Anatomy, University of Bern, Switzerland

Primary ciliary dyskinesia is a rare and underdiagnosed genetic disease affecting the ciliary motility, resulting in diverse symptoms such as chronic rhinitis, chronic otitis media, hearing impairment or situs inversus. Due to the heterogeneity of the disease and since there is no single gold standard diagnostic test, diagnosing PCD is be challenging. This challenge can be

helped by obtaining nasal epithelial cells via a nasal brushing and culturing them at the air-liquid interface.. These cells (fresh or cell culture) can then be analyzed for ciliary beating frequency (CBF) and the ciliary beating pattern (CBP) via high-speed-videomicroscopy. However, this nasal brushing can be uncomfortable and painful, especially for children. The use of lidocaine is proposed to mitigate pain during the brushing; but it is not clear whether the application of lidocaine changes CBF and the CBP in the subsequent analysis and thus would affect the diagnosis of PCD. The aim of this study was to analyze the effect of lidocaine on CBF and CBP of fully re-differentiated, air-liquid-interface cultured nasal epithelial cells (NECs) from healthy volunteers.

NECs from five healthy adult volunteers were obtained via brushing and cultured at the air-liquid interface. After re-differentiation, 1% lidocaine (lido) or isotonic saline (control, IS) were added to the apical side of the NECs for 1 or 5min each and ciliary motility were recorded for up to 150min after application. CBF was calculated by the Cilialyzer program; CBP, amplitude and coordination were analyzed with a semiquantitative approach.

Lido as well as IS increased the CBF significantly compared to baseline (1min IS: baseline 6.0Hz (range 5.1-8.2Hz), 15min 9.3Hz (6.8-12,1Hz), 150min 10.5Hz (8.7-12.5Hz); 1min lido: baseline 4.5Hz (3.9-5.1Hz), 15min 11.0Hz (9.5-12.3Hz), 150min 11.6Hz (10.1-14.6Hz); 5min IS: baseline 6.7Hz (4.2-10.1Hz), 15min 8.2Hz (6.5-10.1Hz), 150min 10.1Hz (8.5-12.0Hz); 5min lido: baseline 6.0Hz (5.5-6.5Hz), 15min 9.3Hz (8.2-10.4Hz), 150min 9.7Hz (7.0-11.1Hz)). However, there were no differences between the lido treatment and the controls. Additionally, no effect of lido compared to controls was observed for CBP, amplitude or coordination.

Therefore, we conclude that the effect in CBF increase relates to addition of fluid onto the mucus and periciliary layer and that 1% lido can be used in nasal analgesia without impact on the analysis of ciliary motility.

P 66

Short-term effects of elexacaftor/tezacaftor/ivacaftor – combination on glucose tolerance in young people with cystic fibrosis – an observational pilot study

Korten Insa[1], Kieninger Elisabeth[1], Krueger Linn[1], Bullo Marina[1], Flueck Christa[2, 3], Latzin Philipp[1], Casaulta Carmen[1], Boettcher Claudia[2]

[1]1Division of Paediatric Respiratory Medicine and Allergology, Department of Paediatrics, Inselspital, Bern University Hospital, University of Bern, Switzerland.; [2]2Department of Paediatric Endocrinology, Diabetology & Metabolism, Department of Paediatrics, Inselspital, Bern University Hospital, University of Bern, Switzerland.; [3]3Department of BioMedical Research, Bern University Hospital and University of Bern, Bern, Switzerland.

Background

The effect of elexacaftor/tezacaftor/ivacaftor (ELX/TEZ/IVA) on glucose tolerance and/or cystic-fibrosis-related diabetes (CFRD) is not well understood. We performed an observational study on the short-term effects of ELX/TEZ/IVA on glucose tolerance.

Methods

16 adolescents with CF performed oral glucose tolerance tests (OGTT) before and four to six weeks after initiating ELX/TEZ/IVA therapy. A continuous glucose monitoring (CGM) system was used three days before until seven days after starting ELX/TEZ/IVA treatment.

Results

OGTT categories improved after initiating ELX/TEZ/IVA therapy ($p = 0.02$). Glucose levels of OGTT improved at 60, 90 and 120 min ($p < 0.05$), whereas fasting glucose and CGM measures did not change.

Conclusions

Shortly after initiating ELX/TEZ/IVA therapy, glucose tolerance measured by OGTT improved in people with CF. This pilot study indicates that ELX/TEZ/IVA treatment has beneficial effects on the endocrine pancreatic function and might prevent or at least postpone future CFRD.

P 67

Is Artificial intelligence as accurate as spirometry in early diagnosis of asthma exacerbation in children ? A prospective study.

Cleverley-leblanc Heidi[1], Lacroix Laurence[2], Siebert Johan[2], Hartley Mary-Anne[3], Barazzone-Argiroffo Constance[1], Gervaix Alain[2], Ruchonnet-Metrailler Isabelle[1]

[1]Paediatric Pulmonology Unit, Department of Paediatrics, Gynaecology and Obstetrics, Geneva University Hospitals, Geneva, Switzerland; [2]Divison of Emergency Medicine, Department of Paediatrics, Gynaecology and Obstetrics, Geneva University Hospitals, Geneva, Switzerland; [3]Intelligent Global Health, Machine Learning and Optimization (MLO) Laboratory, Swiss Federal Institute of Technology (EPFL), Lausanne, Switzerland.

Background

Early recognition of asthma exacerbations is often unmanageable for children with chronic Asthma and their parents, leading to adverse outcomes, including school absenteeism, high hospitalisation rate and an increase morbi-mortality rate. While, these issues could widely be reduced by early detection and prompt treatment.

Aim

This study aims to evaluate the effectiveness of artificial intelligence (AI) powered analysis of lungs sound, in early diagnosis of asthma exacerbation in children, compared with spirometric values.

Method

A single center observational prospective double-blinded study conducted in the Paediatrics University hospital in Geneva to early detect acute asthma exacerbations thanks to artificial intelligence. Digitally registered lung sounds before and after beta 2 agonist (bronchodilator) administration, were recorded in patient aged ≥5 to ≤18 years old, attending their asthma follow-up consultation. These sounds were then analysed through AI program and compared with spirometry, wich is the gold standard exam. In total a 100 patients will be recruited between the 18th November 2020 and April 2022.

Results

59 out of 100 patients (n = 59/100) are included in the trial today. We are expecting to complete the enrolment in April 2022 and evaluate the results by May 2022.

Discussion

AI powered analysis of lungs sound could be a sustainable and effective method to early diagnosis of Asthma attacks. Besides being performant in early Asthma diagnosis and exacerbation's severity rating, it is a handy, uninvasif and inexpensive tool. It could foster and improve self-management of Asthma by patients and their family therefore reducing delayed care, hospitalization and ER visits.

Low and middle income countries could also benefit from this innovation, to counteract the scares medical expertise due to shortage of physicians, needed to recognize the specific acoustic signature of asthma

P 68

Neuroendocrine Cell Hyperplasia of Infancy: a form of childhood interstital lung disease

Kurian GS[1,2], Papangelopoulou D[1,2], Corbelli R[1,3,4], Ghinescu C[1,2]

[1]Département de la femme, de l'enfant et de l'adolescent; [2]Service de pédiatrie générale; [3]Service des spécialités pédiatrique; [4]Unité de pneumologie pédiatrique

A 5-month-old infant with recurrent episodes of sudden increased respiratory effort, presented to the emergency department because of a worsening of symptoms for 2 days. The onset of symptoms was at the age of 3 months. There is no history of respiratory infections. He was seen by his pediatrician 10 days prior to the current episode and was referred for further hospital assessment in another centre, where he was diagnosed as having episodes of gastro-esophageal reflux. The persistence of symptoms led to a second hospital stay in our centre. On clinical examination he had fluctuating respiratory crackles in the left lung, with persistent

tachypnea and reduced oxygen saturation, resolving with oxygen therapy. A radiograph of the chest showed hyperinflation of the lungs and ground glass opacitied in the right middle lobe and left lingular region. Cystic fibrosis was excluded, and both pulmonary hypertension and chronic aspiration caused by upper airway anomalies were excluded with echocardiography and videofluoroscopic swallow study respectively. Broncho-alveolar lavage further ruled out other causes of interstitial lung disease, and cellular and humoral immunodeficiency was excluded. High resolution computer tomography (HRCT) revealed a typical mosaic pattern appearance of ground glass opacity in the superior lobes, right middle lobe and left lingula, with the presence of air-trapping in the in the inferior lobes, as seen in Neuroendocrine Cell Hyperplasia of Infancy (NEHI). This form of interstitial lung disease was described in 2005 as persistent tachypnea of infancy associated with the proliferation of neuroendocrine cells in the distal airways on lung biopsy of unknown etiology. Lung biopsy was deemed unnecessary since HRCT has a 100% specificity for the diagnosis of NEHI. Children with NEHI often present with the typical symptoms as seen above, and failure to thrive, but do not classically develop respiratory failure. Our patient had a score of 8 out of 10 according to Liptzin score, helpful to diagnose NEHI. Genetic evaluation was carried out for specific genes related to interstitial lung disease. The patient was started on short courses of i.v corticosteroid treatment and supplemental oxygen therapy at home with a favorable response. Although neuroendocrine cell hyperplasia of infancy is rare in childhood, typical clinical presentation and radiological findings enable appropriate therapy, with a favorable long-term prognosis.

P 69

What respiratory pathology can be hidden behind a *situs inversus totalis*?

Rouge Elton Paola Andrea[1], Silke Grupe[2], Sylvain Blanchon[3], Anita C. Truttmann[4]

[1] Department of Women Mother Child, CHUV; [2] Clinics of Pediatrics, Hôpital d'Yverdon; [3] Unit t of Paediatrics Pneumology, CHUV; [4] Clinic of Neonatology, CHUV

We present a case of a 2-day-old newborn who developed respiratory distress with rapid worsening in the context of a *situs inversus totalis known antenatally.* We propose a review of the differential diagnoses of respiratory distress occurring rapidly after birth in a full-term newborn with situs inversus. Among the differential diagnoses, primary ciliary dyskinesia affects 25% of children with *situs inversus totalis,* an entity often ignored, occurring in 80% of cases at neonatal age. Antenatal consultation and anticipation of potential complications should be mandatory in this context.

P 70

THINK TWICE, THINK RIGHT

Anguissola G MD[1, 2], Vanoni F PD MD[1, 2], Leoni-Foglia C MD [1, 2], Cristallo Lacalamita M MD[1, 2], Prof. Simonetti GD MD[1, 2], Kottanattu L MD[1, 2]

[1]Clinic of pediatrics, Pediatric Institute of Southern Switzerland (IPSI), EOC, Bellinzona, Switzerland; [2]Università della Svizzera Italiana (USI), Via Buffi 13, 6900, Lugano, Switzerland

Background

Retropharyngeal abscess (RPA) in individuals <20 years has recently increased in incidence with 4.10 per 100'000 cases in 2012 and highest values among children <5 years. Patients clinically show cervical lymphadenopathy (77%), limitation of neck movements (64%), torticollis (54%), drooling (8%) and stridor (5%). Rarely, a retropharyngeal phlegmone, often difficult to distinguish from RPA, could be the first manifestation of Kawasaki disease (KD), due to the intense inflammatory response in the deep neck mucosa. In our case, the awareness of rare symptoms of KD prompted an early diagnosis and the start of the correct therapy, avoiding unnecessary surgery and cardiac complications.

Case report

A 4-year-old girl was referred to our hospital because of 3-day-history of fever and left neck adenopathy, recently complicated by a torticollis. The laboratory finding showed high inflammatory markers. The CT scan of the neck revealed a retropharyngeal abscess spreading from C2 and C3, 4 mm thick with associated cervical adenopathies and minimal mass effect on the left internal jugular vein. Parenteral antibiotics were started, with no clinical or inflammatory markers improvement over 48 hours. On clinical examination, the girl developed an exanthema, conjunctival hyperaemia, strawberry tongue and palmar and plantar swelling. The clinical suspicion quickly moved to KD, supported by blood tests showing cytolitic hepatopathy and elevated NT-proBNP and positive echocardiography with dilation of the left main coronary artery (Z-score 2.94). At day 5 of fever, we stopped antibiotics and started instantly intravenous immunoglobulin 2 g/Kg/dose over 12 hours, intravenous methylprednisolone 10 mg/Kg/dose for 3 days and oral ASA 80 g/Kg/day. The day after we appreciated a vanishing of the KD signs, a progress in laboratory tests and gradual improvement of the torticollis. She continued the therapy with ASA 5 mg/Kg/die, showing a perfect evolution with whole regression of coronary dilation (Z-score 1.36) at day 12.

Conclusion

Our case highlights the importance of considering KD in the differential diagnosis of children with ≥ 5 days of fever, cervical lymphadenopathies and torticollis unresponsive to antibiotic therapy. This report underlines the idea that is necessary to know the rarity and consider the entire clinical picture to achieve the right diagnosis and to avoid invasive measures and possible complications. If you think twice, you think right.

P 71

Rheumatology Transition of Young People in Switzerland – the HEROES study

Berben Lut[1], Daly Mary-Louise[1,2], Daikeler Thomas[2], Wörner Andreas[3]

[1]Department of Nursing Development, University Children's Hospital Basel, Switzerland; [2]Department of Rheumatology, University Hospital Basel, Switzerland; [3]Department of Pediatric Rheumatology, University Children's Hospital Basel, Switzerland

About half of all children with rheumatic diseases need continuous medical care during adolescence and adulthood. An effective and well-planned transition built on individual, structured management plans is crucial for successful transition into adult services. Although transition principles have been described and implemented, outcome data are still scarce. The overall objective of the HEROES study is to develop, implement and evaluate a transitional care (TC) program for adolescents and young adults (AYA) with a rheumatic disease moving from pediatric to adult settings in Switzerland.

Specific aims are:

1) assess AYAs' and parents' experiences and unmet needs related to current TC practices

2) assess stakeholder, healthcare professional, setting and system barriers and facilitators to implementing a TC program

3) develop and implement a TC program that includes a nurse TC coordinator

4) evaluate the effectiveness of the TC program in relation to disease-related outcomes

5) evaluate AYA-, parent- and healthcare professional-reported outcomes and care experiences in relation to the TC program

6) evaluate the implementation outcomes of the TC program in relation to adoption, implementation and sustainability

7) evaluate economic outcomes of the TC program

This study which includes all 10 Swiss pediatric rheumatology centers and their adult counterpart, will use a hybrid effectiveness-implementation type 2 design. For the different parts of the study, specific designs will vary.

For the qualitative data (i.e. AYA' and parents' experiences and unmet needs, acceptance and appropriateness of the intervention and assessment of contextual factors), an explanatory sequential mixed method design will be used.

The quantitative data, i.e. the intervention-effectiveness outcomes, in this study will be assessed using a multi-center quasi experimental pre-post design. To maximize the acceptability and sustainability, a participatory partnership approach will be used, i.e. people that the intervention aims to help and those who will implement it are involved throughout the process. Intervention outcomes will be measured at the AYA (e.g. quality-of-life, treatment adherence), parent (e.g. care satisfaction, giving responsibility for care to the AYA), healthcare professional (e.g. time for AYA, work satisfaction), setting (e.g. billing of consultations) and system (e.g. costs) levels. This innovation also has a potential to serve as a model for TC in other chronic diseases.

P 72

Lupus anticoagulant hypoprothrombinemia syndrome as first manifestation of pediatric systemic lupus erythematosus : a case report.

Mattesini BE[1,2], Vanoni F[1,2], Brazzola P[1,2], Carlomagno R[3], Gerber B[4], Prof. Simonetti GD[1,2]

[1]Clinic of pediatrics, Pediatric Institute of Southern Switzerland (IPSI), EOC, 6500, Bellinzona, Switzerland; [2]Università della Svizzera Italiana, 6900, Lugano, Switzerland; [3]Pediatric Immuno-Rheumatology of Western Switzerland, CHUV, University of Lousanne, 1011 Lousanne, Switzerland; [4]Division of Hematology, Oncology Institute of Southern Switzerland, 6500, Bellinzona, Switzerland

Background

Lupus anticoagulant hypoprothrombinemia syndrome (LAHPS) represents a rare manifestation of pediatric-onset systemic lupus erythematosus (pSLE). LAHPS is characterized by the coexistence of lupus anticoagulant (LAC) with anti-prothrombin antibodies, resulting in a state of acquired hypoprothrombinemia (low coagulation factor II, FII) and predisposing these children to bleed. We report a case of a girl that presents with spontaneous bleeds and LAHPS as first manifestation of pSLE.

Case report

A 10-year-old girl was referred to our paediatric department with widespread hematomas in the absence of trauma, musculoskeletal pain (calves, ankle and wrists) without evidence of arthritis, and severe mood disorder since several weeks. Complete blood cell count was normal, excluding an idiopathic thrombocytopenic purpura as cause of hematomas. Coagulation tests showed normal fibrinogen levels, but a prolonged prothrombin time (PT), a prolonged activated partial thromboplastin time (aPTT) with no correction in the aPTT mixing studies, a positive dilute Russell viper venom test confirming the presence of lupus anticoagulant, positive anti-cardiolipin IgG and IgM, anti-β2-glycoprotein IgG and IgM, as well as a low coagulation FII level. Complete immunologic workup showed positive ANA, anti-dsDNA and anti-C1q, positive Coombs test, and low C3 and C4. A cerebral angio-MRI did not show any abnormality. The patient was diagnosed with LAHPS in the context of pSLE, and treated with oral prednisone, hydroxychloroquine (HCQ) and vitamin D3. Her coagulation profile gradually normalized, her mood rapidly improved and, months later, she remains in clinical remission.

Discussion

LAHPS is a possible cause of bleeding in patients with overt pSLE. Early recognition of this rare disease and timely initiation of immunosuppressive therapy reduce the risk for major bleeding. Various immunosuppressive regimens are used, including corticosteroids alone or in combination with azathioprine, cyclophosphamide, mycophenolate, IVIG or rituximab. Management of LAHPS is delicate, but most children respond well to treatment with a rapid increase in FII levels. However, the normalization of the FII concentration may aggravate the thrombotic risk in pSLE due to the coexistence of LAC.

Conclusion

In patients with pSLE and positive LAC, LAHPS must be suspected in case of bleeding, and treatment promptly started. A close monitoring is crucial in the management of these vulnerable patients.

P 73

Incontinentia Pigmenti – a newborn rash following the Blaschko's lines

Muren S[1], Moser B[2], Bürgler C[3], Häfliger S[3], Fluri S[1]

[1]Klinik Frau & Kind, Abteilung für Pädiatrie/Neonatologie, Spitalzentrum Oberwallis, Hôpital du Valais, CH-3930 Visp; [2]Universitätsklinik für Kinderheilkunde, Inselspital, CH-3010 Bern; [3]Universitätsklinik für Dermatologie, Inselspital, CH-3010 Bern

Introduction

Neonatal erythematous vesicular rash is a diagnostic challenge. The differential diagnosis is wide and a prompt diagnosis can be crucial.

Methods

Case report and review of literature summarizing important clinical and diagnostic aspects.

Case report

A 2 weeks old female newborn was presented to our clinic with a generalized rash with erythematous crusted papules, plaques and blisters which started one week after birth on the trunk spreading to the extremities in a linear arrangement following the lines of Blaschko. The general condition was good. The baby was born spontaneously at term after an eventless pregnancy. The mother suffered from an unspecified eye condition. Maternal serology was negative for VDRL, HIV, HBV and HCV. An infectious etiology could be excluded and a routine blood testing was normal except for a leucocytosis (32 G/l) and eosinophilia (18%). A punch biopsy performed on a lesion showed histological findings consistent with the diagnosis of incontinentia pigmenti in the inflammatory stage. The dermatological management consisted in reducing the risk of infection of the blisters using topical desinfection and hygienic measures. A multisystem work-up and genetical investigation were started.

Discussion

Incontinentia pigmenti (Bloch-Sulzberger syndrome) is a rare X-linked dominant genodermatosis caused by a defect in the IKBKG gene affecting the skin, different organ systems, the central nervous system, eyes, teeth and skeleton with variable expression. There are approximately 25 new cases per year worldwide; 2/3 being sporadic mutations, 1/3 are familial. The disorder is typically identified by unique skin findings, presenting in a series of four stages (I. blistering, II. wart-like rash, III. whirling macular hyperpigmentation, IV. linear hypopigmentation) that emerge throughout the first year of life. The central nervous system manifestations in the eye and in the brain cause the most disability. The typical skin pattern with a linear arrangement which follows the lines of Blaschko is pathognomonic in the clinical setting. Blaschko's lines, named after the German dermatologist Alfred Blaschko, are lines of normal cell development in the skin. These lines are invisible under normal conditions. They become apparent when some diseases of the skin or mucosa manifest themselves according to these patterns. In the clinical examination the detection of a rash following the Blaschko's lines can be helpful to find the adequate diagnosis.

P74

Effects of elexacaftor/tezacaftor/ivacaftor therapy in children with cystic fibrosis – a comprehensive assessment using spirometry, lung clearance index and functional and structural lung MRI

Carmen Streibel, MD[1]*; Corin Willers, MD[1]*; Orso Pusterla, PhD[1,2,3]; Grzegorz Bauman, PhD[2,3]; Enno Stranzinger, MD[4]; Ben Brabandt, MD[4]; Oliver Bieri, PhD[2,3]; Marina Bullo, MD[1]; Insa Korten, MD[1]; Linn Krüger, MD[1]; Carmen Casaulta, MD PD[1]; Philipp Latzin, MD PhD[1]; Elisabeth Kieninger, MD PhD [1]

*[1] Division of Paediatric Respiratory Medicine and Allergology, Department of Paediatrics, Inselspital, Bern University Hospital, University of Bern, Switzerland; [2] Department of Radiology, Division of Radiological Physics, University of Basel Hospital, Basel, Switzerland; [3] Department of Biomedical Engineering, University of Basel, Allschwil, Switzerland; [4] Department of Interventional and Pediatric Radiology, Inselspital, Bern University Hospital, Bern, Switzerland; *contributed equally*

Background

With the introduction of cystic fibrosis transmembrane conductance regulator (CFTR)-modulator treatment in patients with cystic fibrosis (CF) a new era of milder lung disease started. Thus, sensitive parameters are needed to comprehensively monitor disease course and effects of CFTR-modulators in these patients. Functional lung matrix-pencil decomposition (MP)-MRI is a promising tool for assessing impaired ventilation and perfusion quantitatively.

Objective

To assess the treatment effect of ELX/TEZ/IVA on measures of structural and functional lung abnormalities.

Methods

24 children with CF underwent lung function tests (multiple breath washout, spirometry), functional and structural MRI twice (one year apart) before and once after at least two weeks (mean 4.7 ± 2.6 months) on ELX/TEZ/IVA. Main outcomes were changes in lung clearance index (LCI), forced expiratory volume in 1 second (FEV1), defect percentage of ventilation (VDP) and perfusion (QDP), defect distribution index (DDI) and Eichinger score.

Results

We observed a significant improvement in lung function and structural and functional MRI parameters upon ELX/TEZ/IVA treatment (mean; 95%-CI): ΔLCI (TO) -0.84 (-1.62 to -0.06), p = 0.04; ΔFEV1 (z-scores) 1.05 (0.56 to 1.55), p = 0.0002; ΔVDP (% of impairment) -6.00 (-8.44 to -3.55), p <0.0001; ΔQDP (% of impairment) -3.90 (-5.90 to -1.90), p = 0.0006; ΔDDIventilation -1.38 (-2.22 to -0.53), p = 0.003; ΔDDIperfusion -0.31 (-0.73 to 0.12) p = 0.15; ΔEichinger score -3.89 (-5.05 to -2.72), p <0.0001.

Conclusions

Besides lung function, functional and structural MRI is a suitable tool to monitor treatment response of ELX/TEZ/IVA therapy in patients with CF, and should thus be considered as potential outcome in intervention trials and clinical routine.

INDEX OF FIRST AUTHORS

The numbers refer to the numbers of the abstracts.

SWISS MEDICAL WEEKLY

Listed in: Index Medicus / MEDLINE; Web of science; Current Contents; Science Citation Index; EMBASE

EMH Swiss Medical Publishers Ltd.
Swiss Medical Weekly
Farnsburgerstrasse 8
CH-4132 Muttenz, Switzerland
office@smw.ch

ISSN online supplement: 2504-1622