

SMW

Established in 1871

Swiss Medical Weekly

Formerly: Schweizerische Medizinische Wochenschrift

An open access, online journal • www.smw.ch

Supplementum 249

ad Swiss Med Wkly
2021;151

June 10, 2021

**Swiss Society of Paediatrics
Abstracts of the annual meeting 2021**

**pädiatrie
schweiz** Schweizerische Gesellschaft für Pädiatrie
Société Suisse de Pédiatrie
Società Svizzera di Pediatria

Jahrestagung SGP
Congrès annuel SSP

2021

10-11 Juni / Juin Forum Fribourg

SWISS SOCIETY OF PAEDIATRICS

ABSTRACTS OF THE ANNUAL MEETING 2021

TABLE OF CONTENTS

2 S	<u>Free communications: FM 1–18</u>
8 S	<u>SwissPedNet: SPN 2–18</u>
15 S	<u>Posters: P 1–47</u>
21 S	<u>Index of first authors</u>

FREE COMMUNICATIONS

FM 1

Children with an autism spectrum disorder in the canton of Zurich – Evaluation of daily life, early interventions, and need of support by professionalsSchneider N¹, Von Rhein M¹, Jenni O¹, Schaefer C¹¹Child Development Center, University Children's Hospital Zurich, Switzerland

In recent years, the topic of Autism Spectrum Disorder (ASD) received increasing attention in Switzerland and gained additional political relevance through the report on measures to improve the support of people with ASD published by the Federal Council (1). In Switzerland, the prevalence of ASD is estimated to be between 0.6 and 0.8 percent (with an increasing tendency) (2). ASD belongs to the pervasive developmental disorders (PDD) and parents of children with ASD are often greatly burdened by the intensive and challenging care of their child (3). Therefore, an early diagnosis associated and initiation of early therapeutic measures is crucial for the development and the quality of life of children with ASD and their families (4). However, little is known on the families' perspective regarding early interventions, so far: what specifically are the needs of affected families? Do early special education measures such as special education and speech therapy meet their needs, and how much experience with autistic children do therapists in these two disciplines have, considering the increased need for autism-specific interventions?

Based on a central register of all children registered for special educational measures before kindergarten entry, we are currently conducting a survey among 147 affected families to investigate daily life, early interventions, and need of further support. Based on the current response to recruitment, we assume a response rate of approximately 50 to 60 participants. In addition to the question of a possible imbalance between supply and demand for these therapies, the need and experiences of the parents are being assessed in a sequential-explanative mixed method design, and compared to the perspective of special education teachers working with affected children.

1. Bundesrat. Bericht Autismus-Spektrum-Störungen - Massnahmen für die Verbesserung der Diagnostik, Behandlung und Begleitung von Menschen mit Autismus-Spektrum-Störungen in der Schweiz. 2018.
2. Gundelfinger R. Autismus in der Schweiz. Pädiatrie; 2013.
3. Davis NO, Carter AS. Parenting stress in mothers and fathers of toddlers with autism spectrum disorders: Associations with child characteristics. *Journal of autism and developmental disorders*. 2008;38(7):1278.
4. Howlin P, Magiati I, Charman T. Systematic review of early intensive behavioral interventions for children with autism. *American journal on intellectual and developmental disabilities*. 2009;114(1):23-41.

FM 2

Utilization of medical cannabis in the treatment of autism spectrum disorder in children - current evidenz and treatment experienceOgal M¹, Seewald H²¹Paediatric Clinic, Brunnen, Switzerland; ²Department of Pediatrics, Triemli Municipal Hospital, Zurich, Switzerland**Background**

Autism spectrum disorder (ASD) is a complex neurodevelopmental disorder with a multifactorial etiology. The clinical presentation of children with ASD varies widely - patients commonly exhibit comorbid symptoms such as aggression, hyperactivity, and anxiety. Therefore, also the treatment is complex and needs a highly individual approach. There is growing evidence that the endocannabinoid system (ECS) plays a key role in neurodevelopment as well as in inflammatory processes. These findings are corroborated by various preclinical and clinical studies showing promising effects in the treatment of ASD in children with medical cannabis.

We here provide an introduction into the different cannabinoids with a special focus on CBD and THC, point out their medicinal effects and side effects, and show what needs to be considered when using medical cannabis preparations as a treatment option in children with ASD. Our aim is to give an example on how medical cannabis can be implemented in the pediatric clinic, to provide an overview of cannabis products that are available in Switzerland and to present a case report. The information provided is based on the current literature and gives an insight into our own treatment experience with cannabis.

FM 3

Parents' Vaccination Information Seeking Behaviors, Satisfaction with, and Trust in Providers in SwitzerlandDeml M^{1,2}, Ebi S^{3,2}, Jafflin K^{1,2}, Buhl A^{1,2}, Engel R^{3,2}, Picker J^{3,2}, Häusler J^{3,2}, Wingeier B⁴, Krüerke D⁴, Huber B⁵, Merten S^{1,2}, Tarr P^{3,2}¹Swiss Tropical and Public Health Institute, Socinstrasse 57, 4051 Basel, Switzerland; ²University of Basel, Petersplatz 1, 4051 Basel, Switzerland; ³University Department of Medicine, Kantonsspital Baselland, 4101 Bruderholz, Switzerland; ⁴Klinik Arlesheim, Pfeffingergweg 1, 4144 Arlesheim, Switzerland; ⁵Department of Pediatrics, HFR Fribourg Cantonal Hospital, Fribourg, Switzerland

Introduction. Previous research has shown how medical providers, individuals' social networks, and the media are among parents' most trusted information sources for vaccination decisions. In Switzerland, where complementary and alternative medicine (CAM) is popular, little research has examined parents' vaccination decision-making processes.

Methods

We employed a mixed-methods study design. We first conducted qualitative interviews with 30 parents and 37 providers (20 biomedical physicians, 17 CAM providers) and observed 34 vaccination consultations. These results informed quantitative data collection tools which we then used to conduct quantitative telephone interviews, including the validated Parent Attitudes about Childhood Vaccines (PACV) scale, with 130 providers and 1398 parents to collect data on parents' information sources and vaccination consultations with providers (both CAM- and biomedically-oriented). Our aims were to better understand parents' vaccination information seeking behaviors, vaccination information sources, and vaccination consultations with providers.

Results

Based on the PACV scale, we considered 893 parents as non-vaccine-hesitant (non-VH) and 505 parents as vaccine-hesitant (VH). Whereas both groups cited pediatricians as the most trusted information source, non-VH-parents were significantly ($p < 0.05$) more likely to cite pediatricians ($N = 759[85\%]$ vs $N = 359[71\%]$) and public health authorities ($N = 333[37\%]$ vs $N = 100[20\%]$) than VH-parents. VH-parents were significantly more likely to have consulted another provider ($N = 198[39\%]$ vs $N = 175[20\%]$) compared to non-VH-parents and to express less satisfaction with both their primary ($N = 346[82\%]$ vs $N = 588[91\%]$) and other providers ($N = 83[42\%]$ vs $N = 144[82\%]$) and less trust in their primary ($N = 372[88\%]$ vs $N = 635[98\%]$) and other providers ($N = 110[56\%]$ vs $N = 148[85\%]$). VH-parents were significantly less likely to be satisfied with their biomedically oriented primary provider than non-VH-parents ($102[69\%]$ vs $467[91\%]$). However, when the primary provider was CAM-oriented, we saw similar levels of satisfaction among both groups ($239[89\%]$ VH-parents vs $120[89\%]$ non-VH-parents).

Conclusions

Since physicians are parents' most trusted source of vaccination information, and dissatisfaction and distrust may push parents away from vaccination, it is important for physicians to create trusting environments where parents' questions about vaccination and vaccination concerns can be met with satisfaction.

FM 4

Health care professionals' needs around vaccination in Switzerland: A quantitative needs assessment surveyLucas Ramanathan Pia^{1,2,10}, Baldesberger Nadja^{1,2,10}, Lüthy Alyssa^{1,2}, Buhl Andrea^{2,3}, Gisin Martina⁸, Koch Roswitha⁹, Nicca Dunja¹¹, Suggs L. Suzanne^{4,5}, Huber Benedikt M.^{6,7}, Dietrich Léna G.^{1,2}, Deml Michael J.^{2,3}, Tarr Philip E.^{1,2}¹University Department of Medicine and Infectious Diseases Service, Kantonsspital Baselland, Bruderholz, Switzerland; ²University of Basel, Basel, Switzerland; ³Swiss Tropical and Public Health Institute (Swiss TPH), Basel, Switzerland; ⁴Institute for Public Health, and Institute for Public Communication, Università della Svizzera italiana, Lugano, Switzerland; ⁵Swiss School of Public Health, Zurich, Switzerland; ⁶Center for Integrative Pediatrics, Department of Pediatrics, Fribourg Hospital HFR, Fribourg, Switzerland; ⁷Faculty of Science and Medicine, University of Fribourg, Fribourg, Switzerland; ⁸Department of Obstetrics and Gynecology, University Hospital of Basel, Switzerland; ⁹Swiss Nurses Association, Bern, Switzerland; ¹⁰Pharmaceutical Care Research Group, Basel, Switzerland;

¹¹*Epidemiology, Biostatistics and Prevention Institute, University of Zurich, Zurich, Switzerland*

Background

As the most trusted advisors on vaccination decisions, health care professionals (HCPs) play an important role in making vaccine recommendations. In order to counsel vaccine hesitant (VH) patients and clients appropriately, HCPs need factual knowledge about vaccines (e.g. vaccine efficacy, vaccine safety, immunology) as well as skill in communication (e.g. shared decision making).

Methods

We conducted a vaccination knowledge and communication needs assessment survey among physicians, pharmacists, nurses and midwives in Switzerland. The online survey was conducted from 25.10.2020 to 15.03.2021 and contained 35 questions. The aim was to explore the following topics among respondents; (1) interest in obtaining additional training related to vaccination counseling and knowledge (vaccine safety, efficacy, side effects, vaccine-preventable diseases, vaccine ingredients, immunology, communication with VH persons, shared decision-making, the Swiss vaccination schedule); (2) vaccination recommendation practices; (3) experience with vaccination counseling and administration; (4) degree of ease when dealing with VH patients/clients; (5) own perspectives on VH, delays and refusals; (6) perspectives on individual, shared and mandated vaccination decisions, regarding children (measles vaccine) and HCPs (influenza vaccine).

Results

As of 16.02.2021, 491 physicians, 146 pharmacists, 599 midwives and 573 nurses took the survey. 92% of physicians, 98% of pharmacists, 90% of midwives and 83% of nurses were interested in obtaining additional training related to vaccination. Vaccine efficacy and side effects were mentioned most often by all 4 professional groups as key topics of interest. Interest in vaccine mandates was limited. More than 2/3 of respondents in each professional group were in favor of individual or shared decision making for both childhood vaccination and for influenza vaccination of HCP.

Conclusion

Preliminary results of our online needs assessment survey suggest a high interest among HCPs in Switzerland for additional training related to vaccination. Communication skills might be particularly important, given that the majority of respondents prefers a shared decision-making approach over mandates.

FM 5

Efficacy and safety of vaccines in children with rheumatic diseases on immunosuppressive therapy - a systematic review

Keller M¹, Zimmermann P^{1,2,3}

¹*Faculty of Science and Medicine, University of Fribourg, Fribourg, Switzerland;* ²*Department of Paediatrics, Fribourg Hospital HFR, Fribourg, Switzerland;* ³*Infectious Diseases Research Group, Murdoch Children's Research Institute, Parkville, Australia*

Background

The immunogenicity of vaccines in children with juvenile autoimmune rheumatic diseases (JARs) can be reduced, there are additional safety concerns around vaccination and there is a potential for worsening in disease activity. In this systematic review, we summarise studies that investigated the immunogenicity and safety of routine vaccines in children and adolescents with JARD on immunosuppressive treatment.

Methods

A systematic search was done using MEDLINE over the Ovid interface to identify original studies investigating vaccine antibody responses and safety of routine vaccines in children with JARD on immunosuppressive treatment.

Results

We identified 37 studies investigating 2,571 children and adolescents with JARD on immunosuppressive treatment and 4,895 control children. Overall, 56 geometric mean antibody titres were measured in children with JARD on immunosuppressive treatment of which 19 (34%) were lower, six (11%) higher and 31 (55%) similar compared with control children. Of the 39 seroprotection rates measured, 10 (26%) were lower, two (5%) higher and 27 (69%) similar in the two groups. Of the 27 seroconversion rates measured, nine (33%) were lower, two (8%) higher and 16 (59%) similar in children with JARD compared with control children. However, many of the studies were underpowered, and not designed to show non-inferiority between children with JARD and controls.

Severe adverse events were reported in 38 children (33 with juvenile idiopathic arthritis, four with systemic lupus erythematosus and one in a healthy child), most of them were likely not related to the vaccination (e.g. elective hospitalisations and surgeries). A worsening in disease activity was reported in 44 (2%) children with JARD on immunosuppressive treatment, again many of them were likely not related to the vaccination.

Conclusion

Vaccination in children with JARD on immunosuppressive treatment is safe and should be promoted, especially since these children are at increased risk for infection. The importance for the completion of vaccination schedules should be stressed. Strategies to compensate for the lower vaccine responses, which are found in approximately one third of these children, include measuring antibody levels to determine the optimal timing for the administration of additional booster doses.

FM 6

Management of acute demyelinating attacks in the pediatric population: a Swiss consensus statement

Hofer Seline¹, Bauder Florian², Capone Mori Andrea³, Chan Andrew⁴, Dill Patricia⁵, Garcia-Tarodo Stéphanie⁶, Boeggel Simonetti Barbara^{4,7}, Hackenberg Annette⁸, Kalsner Judith⁹, Maier Oliver¹⁰, Schmid Regula¹¹, Strozzi Susi¹², Bigi Sandra^{1,13}

¹*a Division of Child Neurology, Department of Pediatrics, Children's Hospital Bern, Inselspital, University of Bern, Switzerland;* ²*Children's Hospital, Lucerne, Switzerland;* ³*Children's Hospital, Aarau, Switzerland;* ⁴*Department of Neurology, University Hospital, Inselspital, University of Bern, Bern, Switzerland;* ⁵*University Children's Hospital Basel, Basel, Switzerland;* ⁶*University Children's Hospital, Geneva, Switzerland;* ⁷*Institute of Pediatrics of Southern Switzerland (IPSI EOC), Bellinzona, Switzerland;* ⁸*Department of Paediatric Neurology, University Children's Hospital, University of Zurich, Switzerland;* ⁹*Pediatric Neurology and Neurorehabilitation Unit, Lausanne University Hospital, Lausanne, Switzerland;* ¹⁰*Children's Hospital, St. Gallen, Switzerland;* ¹¹*Children's Hospital, Winterthur, Switzerland;* ¹²*Department of Pediatrics, Cantonal Hospital, Chur, Switzerland;* ¹³*Institute of Social and Preventive Medicine, University of Bern, Switzerland*

Background and Methods

Acquired demyelinating syndromes (ADS) encompass distinct entities and occur in approximately 1/100'000 children. While the use of high dose intravenous corticosteroids is well-established, agreement on steroid taper and type of second line therapy is lacking. A comprehensive, unified and standardized treatment approach is crucial in the management of patients with rare diseases. Therefore, this study performed from July 2018 to June 2020 aimed at developing a national consensus on the management of ADS in the pediatric population using the Delphi approach. Consensus was defined as agreement in >75%. Designated Neuropediatricians with an expertise in the management of pediatric neuroinflammatory diseases in all university and cantonal hospitals of Switzerland were included. The response rate was 100%.

Results

High-dose i.v. methylprednisolone (20-30 mg/kg/die for 5 days) is the first line treatment irrespective of the distinct entity of the ADS. An oral steroid taper is recommended in acute demyelinating encephalomyelitis (ADEM) and in neuromyelitis optica spectrum disorder (NMO-SD). However, in the latter more in the sense of bridging. The choice of second line treatment depends on the entity of ADS: in optic neuritis (ON) and ADS due to relapsing remitting multiple sclerosis, first line treatment should be repeated, whereas plasma exchange is recommended in NMO-SD, ADEM and transverse myelitis.

Conclusions

A national guideline allowing for a more unified approach in the management of pediatric ADS will enhance future research in this field, making data more comparable. The definition of inadequate treatment response to first line therapy remains a challenge and requires future research.

FM 7

A Whole Food Plant-based Diet parallel to treatment in Juvenile Idiopathic Arthritis – a pilot study on feasibility: The NutriJIA-Study.

Ngoumou G¹, Schroetter L¹, Klotsche J⁴, Kandil F^{1,3}, Stritter W¹, Kessler C^{3,5}, Michalsen A^{3,5}, Kallinich T^{2,4}, Minden K^{2,4}, Seifert G¹

¹Department of Pediatric Oncology and Hematology, Charité Universitätsmedizin, Berlin, Germany; ²Pediatric Pneumology, Immunology and Critical Care Medicine, Charité Universitätsmedizin, Berlin, Germany; ³Department of Internal and Complementary Medicine, Immanuel Krankenhaus Berlin, Germany; ⁴German Rheumatism Research Center, Leibniz Institute, Berlin, Germany; ⁵Institute of Social Medicine, Epidemiology and Health Economics, Charité Universitätsmedizin, Berlin, Germany

Juvenile Idiopathic Arthritis (JIA) is the most common rheumatologic condition in childhood and affects about 15.000 children in Germany, with an incidence of 1000 new cases per year. Little is known about the causes of the disease. In adults with Rheumatoid Arthritis (RA) dietary patterns with increased consumption of fruits and vegetables and reduction in animal products showed beneficial effects regarding pain, mobility limitations and overall inflammatory activity.

The NutriJIA-Study will examine the feasibility and provide first data on the effects of a whole food plant based diet in children with JIA.

Children and adolescents from 8 to 18 years from the pediatric rheumatology outpatient clinic of the Charité in Berlin with a diagnosis of JIA are randomly divided into a nutritional intervention group and a waiting list control group. The patients in the intervention group receive a diet change to a WFPB nutrition for 12 weeks in parallel with the ongoing therapy. Patients from the waiting list control group act as a control group for these 12 weeks after which they change their diet to a WFPB nutrition as well. Patients in the intervention group eat as desired from weeks 13 to 24. The change in general well-being is compared in the two groups. Additional parameters examined are clinical and anamnestic disease activity markers, quality of life, nutrient intake and supply, laboratory data and the feasibility of the change in diet assessed by parents and children.

The results of the study may provide first data that can be used in a broader follow up project with regard to suitable target values and sample size calculations.

FM 8

Keep calm, it's just about transition. Patients-based evaluation of a multidisciplinary structured group intervention for diabetic adolescents before transition from pediatric to adult care

Perrin A¹, Bussien C², Castellsague M³, Gastaldi G⁴, Perrenoud L³, Schwitzgebel V^{5,6}, Cafilish M⁷, Klee P^{5,6}

¹Service of General Pediatrics, Department of Women-Children-Teenagers, University Hospitals of Geneva, 1211 Geneva, Switzerland; ²Department of Community Medicine, Primary Care and Emergency Medicine, University Hospitals of Geneva, 1211 Geneva, Switzerland; ³Care directorate, University Hospitals of Geneva, 1211 Geneva, Switzerland; ⁴Division of Endocrinology, diabetology, hypertension and nutrition, department of Medical Specialities, University Hospitals of Geneva, 1211 Geneva, Switzerland; ⁵Pediatric Endocrine and Diabetes Unit, Department of Women-Children-Teenagers, University Hospitals of Geneva, 1211 Geneva, Switzerland; ⁶Diabetes Center of the Faculty of Medicine, University of Geneva; ⁷Adolescent Outpatient Clinic, Department of Women-Children-Teenagers, University Hospitals of Geneva, 1211 Geneva, Switzerland

Background and aims

Transition between pediatric and adult care is a crucial step for adolescents with chronic diseases like type 1 diabetes; many patients report having difficulties with this period. The change in the medical team occurs at a time when adolescents are furthermore confronted to physical and emotional changes and this period is associated with poorer metabolic control.

Since 2016, we offer a structured group intervention, delivered by a pediatric and adult multidisciplinary team, to all type 1 diabetic patients aged 16 to 18 preparing for their transition. This half-day meeting includes a playful introduction, three practical workshops about medical, technical and dietetic aspects of diabetes and a session of brainstorming about transition.

Our aim was to evaluate if the adolescents kept memories of this intervention, and to have their feedback and suggestions for improvements.

Methods

All participants who followed the program between 2016 and 2018 were called 1 - 3 years after the event and asked to answer a questionnaire about their memories, their satisfaction rate about the different parts of the intervention, and their suggestions for improvement. The results allowed a qualitative evaluation of the group intervention.

Results

Among the 21 adolescents, 17 remembered their participation. All of them gave a positive feedback, with a mean global mark of 8.3/10, and would recommend this program to other diabetic teenagers. The adolescents particularly enjoyed meeting other young diabetics, and sharing their experiences and feelings, as well as meeting the adult diabetologist before transitioning. Strikingly, the entertaining introduction and the structured workshops about technical and dietetic skills were best remembered, whereas informal, less structured discussions, had left fewer memories. The medical and technical skills workshops were best rated (8.3 and 8.6/10 respectively).

Conclusion

Our multidisciplinary group intervention delivered by pediatric and adult caregivers for diabetic adolescents preparing for transition is well-appreciated. The well-structured workshops give more persistent memories than less structured discussions. This project shows how important it is to ask for the teenagers' feedback, in order to provide the most appropriate group intervention. This qualitative evaluation will allow us to adapt our group intervention to better fit with the teenagers' expectations.

FM 9

Treatment of Functional Abdominal Pain Disorders (FAPDs) with Paediatric Acupuncture: A Case Series

König N¹, Seewald H², Ogal M³, Bildstein T⁴, Marx G^{2,4}

¹Center for Integrative Medicine, Cantonal Hospital St.Gallen, Switzerland; ²Department of Pediatrics, Triemli Municipal Hospital, Zurich, Switzerland; ³Paediatric Clinic, Brunnen, Switzerland; ⁴Department of Pediatric Gastroenterology, Hepatology and Nutrition, Children's Hospital of Eastern Switzerland, St.Gallen, Switzerland

Introduction

Chronic abdominal pain is one of the most common problems encountered in daily clinical paediatric practice, functional abdominal pain disorders (FAPDs) being the most common cause. FAPDs include functional dyspepsia, irritable bowel syndrome, abdominal migraine, and functional abdominal pain-not otherwise specified. However, pathophysiologic mechanisms underlying FAPDs remain largely unclear. Managing FAPDs is challenging as there is limited evidence for pharmacological therapies. Although various non-pharmacologic treatments are available, data on their efficacy and safety are scarce. Here we present 8 cases of children with functional abdominal pain treated with paediatric acupuncture.

Case series

8 Children (4 girls, 4 boys, age 4-11years) were referred for paediatric acupuncture treatment, having been diagnosed with functional abdominal pain by paediatric gastroenterologists. Some patients had underlying conditions such as coeliac disease (under strict gluten-free diet) or additional symptoms (headache, constipation, cyclic vomiting). Symptom frequency ranged from daily (6 patients) to several times a week (2 patients). 7 patients had school absences because of their symptoms. The number of treatments ranged from 5 to 22 during 3 to 24 months. In all patients, acupuncture treatments showed a positive effect already after 1-5 treatments. In 2 patients, symptoms resolved completely, while 4 patients continued to have symptoms very sporadically. 2 patients required supportive acupuncture therapy with greater intervals between treatments during a longer time. Additional symptoms resolved with acupuncture treatment in all patients.

Conclusions

Paediatric acupuncture did show a positive effect on chronic abdominal pain in 8 patients with FAPDs. While FAPDs are generally difficult to manage, paediatric acupuncture is a safe and effective treatment option. The effect of acupuncture in children with FAPDs remains understudied, highlighting the need for further investigation.

FM 10**Brain volume in patients with congenital heart disease from childhood to young adulthood**

Naef N.¹, Ehrler M.¹, Schlosser L.^{1,2}, Wehrle F.^{1,3}, Von Rhein M.^{1,3}, Liamlihi R.^{1,3}, Greutmann M.⁴, Kretschmar O.^{3,5}, Latal B.^{1,3}, Tuura O'Gorman R.^{3,5}

¹Child Development Center, University Children's Hospital Zurich, Switzerland; ²Department of Neurology, University Hospital Zurich, Switzerland; ³Children's Research Center, University Children's Hospital Zurich, Switzerland; ⁴Department of Cardiology, University Heart Center, University Hospital Zurich, Switzerland; ⁵Pediatric Cardiology, Pediatric Heart Center, University Children's Hospital Zurich, Switzerland; ⁶MR Research Center, University Children's Hospital Zurich, Switzerland

Background

Congenital heart disease (CHD) is associated with brain abnormalities. Smaller total brain volumes (TBV) are frequently described in neonates, yet little data exists beyond infancy. This study reports brain volumes in a large dataset of patients with CHD and controls across childhood, adolescence and early adulthood.

Methods

Data of 4 cross-sectional studies were pooled. Inclusion was restricted to CHD patients requiring cardiopulmonary bypass surgery before 6 years of age without genetic disorders. Patients and controls underwent 3T cerebral MRI and cognitive assessments. The dataset comprised 118 patients and 137 controls between 9 and 32 years of age. Linear Regression with an age-group interaction was calculated in all individuals.

Results

TBV was smaller and IQ was lower in CHD patients compared to controls (TBV: $P = 0.02$, IQ: $P < 0.001$). Across all individuals, gray matter volume was negatively associated with age ($\beta = -0.45$, $P < 0.001$), while white matter volume was positively associated with age ($\beta = 0.28$, $P < 0.001$). TBV was not associated with age ($\beta = -0.13$, $P = 0.06$). There was no interaction between age and group on either white or gray matter ($P > 0.1$). Cardiac risk factors for smaller TBV was the presence of a univentricular defect ($P = 0.01$). Smaller TBV was associated with lower IQ ($P = 0.04$).

Conclusion

CHD patients, particularly those with severe CHD, have smaller TBV from childhood to early adulthood. No evidence was found for a catch-up in brain growth compared to controls, suggesting a persistence of smaller TBV in CHD until young adulthood. Longitudinal studies are crucial to better understand brain-development in CHD.

FM 11**Children with Developmental Delay: Regional Distribution, Access to Care, and Utilization of Early Interventions in the Canton of Zurich.**

von Rhein Michael^{1,2}, Gruebler Etienne^{1,3}, Grübner Oliver^{3,4}, Jepathavakumar Daniela¹, Toumi Leila¹, Jenni Oskar^{1,2}

¹Child Development Center, University Children's Hospital Zurich, Switzerland; ²Children's Research Center, Zurich, Switzerland; ³Department of Geography, Geographic Information Visualisation and Analysis, University of Zurich, Switzerland; ⁴Epidemiology, Biostatistics and Prevention Institute, University of Zurich, Switzerland

Developmental delay (DD) with a prevalence of 15% of all children is one of the most frequent disorders in early childhood affecting thousands of individuals in Switzerland every year. Early identification of affected children is critical to ensure appropriate therapeutic interventions, to support the families, and finally to prevent life-long health, educational, and social consequences. It is widely accepted that early intervention programs are both ethically mandatory and cost-effective for the society on a long-term perspective. However, we note that there is a large paucity of information about supply, demand, and utilization of services for children, as well as about the regional distribution of children in need, referring doctors, and therapeutic places in Switzerland. Therefore, we do not know much about the spatial variation of health care need and corresponding services in Switzerland. Furthermore, regionally specific influencing factors for spatial variations in health care are not well understood.

In the Canton of Zurich, there is a centrally organized register of all children with DD in need of early interventions at the two Units of Special Needs Education (USNE). We have collected a comprehensive data set from all children (age 0-4) admitted to the USNE in 2017 ($n = 2033$) and analyzed it using the 2SFCA (Two-Step Floating Catchment Area Method) - a geospatial method to measure health care accessibility. The talk will present a complete description

of the cohort as well as the spectrum of the special needs support assigned by the USNE and contrast the findings to the eventually utilized interventions. Furthermore, the spatial distribution (and potential discrepancies) of needs and services, will be presented. Surprisingly, the number of hours used is by far lower than the assigned amount of support. We aim to explain this discrepancy between assigned therapeutic support and utilization rate of health and educational care for children with DD.

Key messages

It is challenging to organize a system to grant the children with developmental delay an easy and fair access to early intervention in Switzerland. Despite a sophisticated, interdisciplinary system to determine each child's individual needs, various reasons lead to a gap between recommended and utilized support. Pediatric health care research is crucial to assess and optimize pediatric health care in Switzerland. Geospatial methods may help to identify inequity in access to health care.

FM 12**Pathognomonic thyroid hormone constellation as clue to the diagnosis of MCT-8 deficiency in boys with global developmental delay**

Beiner M.¹, Schaller A.², Rusca N.², Grunt S.¹, Lämmle A.^{1,3}

¹Universitätsklinik für Kinderchirurgie und Kinderheilkunde, Inselspital Bern, CH-3010 Bern, Switzerland; ²Universitätsklinik für Humangenetik, Inselspital Bern, CH-3010 Bern, Switzerland; ³Universitäts-Institut für Klinische Chemie Inselspital, Bern, CH-3010 Bern, Switzerland

Allan-Herndon-Dudley syndrome is caused by pathogenic variants in the X-linked gene SLC16A2 encoding the monocarboxylate transporter 8 (MCT8). MCT8 is required for the transport of thyroid hormones (T3 and T4) into the brain. Male patients affected by MCT8 deficiency suffer from central hypothyroidism and peripheral hyperthyroidism. The broad clinical spectrum spans a neurological phenotype with a pronounced head lag and movement disorders and a phenotype associated with peripheral thyrotoxicosis. Recent clinical trials have demonstrated that TRIAC - an endogenous thyroid hormone analogue that enters the brain independent of MCT8 - effectively alleviates symptoms related to peripheral thyrotoxicosis. Currently a follow-up clinical trial investigates whether therapy with TRIAC ameliorates the neurological phenotype when therapy is commenced early (<36 months of age).

Here we present three patients which have been diagnosed with MCT8 deficiency by molecular genetic analysis within one year at our clinics. Patient 1 was diagnosed with 21 months. His first symptoms were a head lag and a generalized neurodevelopmental delay which began with a few weeks of life and got more pronounced with age. Patient 2 was diagnosed with 10 months. His first symptoms were a head lag, a generalized muscular hypotonia, a failure to thrive and episodes of inconsolable crying together with opisthotonic crisis when he was a few weeks old. Patient 3 was diagnosed with 23 months. His first symptoms were a head lag and a generalized muscular hypotonia and developmental delay which prompted first investigations at the age of 5 months. Retrospectively all three patients had the typical thyroid hormone constellation with elevated T3, decreased T4 and normal TSH levels. All three patients are now treated with high doses of TRIAC according to the protocol of the ongoing clinical trial (TRIAC II study).

Patients presenting with symptoms such as head lag, muscular hypotonia, developmental delay, failure to thrive, inconsolable crying and opisthotonic crisis should prompt determination of thyroid hormones. Elevated T3 levels in combination with decreased T4 and normal (to high) TSH levels are pathognomonic for MCT8 deficiency and requires molecular genetic confirmation of the disease. Timely diagnosis of MCT8 deficiency is of utmost importance in order to initiate early treatment with TRIAC, to avoid peripheral thyrotoxicosis and to improve the neurological phenotype.

FM 13**Scabies management in children in Switzerland**

Stebler A.¹, Pfister M.², Buettcher M.^{2,3}

¹Faculty of Medicine, University Basel, Switzerland; ²Paediatric Clinical Pharmacology, University Children's Hospital Basel, Switzerland; ³Paediatric Infectious Diseases, Children's Hospital Lucerne, Switzerland

Background

Scabies is a neglected disease with major global health concern, particularly in young children. Highest incidences occur in developing countries, however scabies is encountered globally. Management guidelines are rare and availa-

bility of therapeutic agents differ by region. Ivermectin is not licensed in children (<15kg). We reviewed management in Switzerland among different specialties (paediatricians (P), general practitioners (GP), dermatologists (D), paediatric dermatologists (PD), tropical medicine (TM), paediatric ID (PID)).

Methods

A total of 237 physicians including P (n = 141), GP (n = 28), D (n = 45), PD (n = 6), TM (n = 7), PID (n = 10) completed the national online survey (05 to 08/2020; 36 questions; 15 minutes) asking management, approximate cases seen and research priorities.

Results

Distribution Swiss vs. migrant were equal. Diagnostic criteria were diverse: 84% of P and 65% of GP rely on history and visual skin examination alone. 83% of PD use at least dermoscopy. D (47%) use laboratory diagnostics. 37%, 44%, 40% and 30% of P, D, PD and TM respectively will not use Ivermectin in <15kg as it is off-label. GP (53%) fear adverse reactions. Improved diagnostic tools are important research priorities for 68% and 47% of the P (incl. PID) and GP respectively. Reviewing Ivermectin in <15kg would be priority for 83% of PD, 64% of D and 42% of GPs. 54%, 58% of D (incl. PD) and P (incl. PID) respectively prioritize optimized dosing and treatment protocols.

Conclusion

Relevant scabies cases occur in migrant and Swiss populations equally. Management is heterogeneous as non-dermatologists use Ivermectin reluctantly. Research priorities include convenient diagnostic tools, a child-appropriate, oral Ivermectin formulation and optimized dosing and treatment protocols.

FM 14

Tularemia on the rise in Switzerland – children compared to adults

Imbimbo C¹, Friderich P², Buettcher M³

¹Medical student, Joint Medical Master Program, University Lucerne and Zurich; ²Department of Microbiology, Cantonal Hospital Lucerne; ³Department of Paediatric Infectious Diseases, Children's Hospital Lucerne, Cantonal Hospital Lucerne

Background

Tularemia, a rare zoonosis, is caused by *Francisella tularensis* (F.t.), a gram-negative intracellular bacterium transmitted by insects, contaminated environment and contact with infected animals. In Switzerland F.t. subsp. holarctica is the only causative subspecies. From 2010 to 2017 an increase (0.3 to 1.4 per 100'000 per year) of reported cases (24% in < 24 years) has been observed. Central Switzerland is particularly affected. Comparative clinical data from children and adults are rare.

Methods

Lucerne Cantonal Hospital serves a population of 700'000. Retrospective review of cases from 2010 to 2019 with positive F.t. serology or PCR (Serology data only from 2017-9) with 22 confirmed cases (8 children).

Results

Mean age in children and adults was 9.25 years (range 4-14) and 49.6 years (range 18- 84 years) respectively. Exposure to tick/insect bite was reported in 5 children and 8 adults. 8 cases occurred during the winter season. In adults it took longer (range: 11d – 2 months) to establish the diagnosis than in children (range: 3d – 1month). All children and 12 adults had ulcero-glandular and 3 adults had pulmonary tularemia. 7 children and 9 adults were treated with beta-lactam antibiotics for 1-3 weeks prior diagnosis. All adult patients were exposed to X-rays during medical work-up. In children US was the primary imaging modality. 6 children (mean 3d) and 10 adults (mean 8d) were hospitalized. All children received Ciprofloxacin as final treatment with neither adverse events nor recurrence whereas 3 adults had recurrent disease.

Conclusions

The disease was longer and more complicated in adults compared to children. For adult cases more diagnostics were used and it took longer to establish the diagnosis. Inadequate antimicrobial use was seen in both age groups. Tularemia should be part of differential diagnosis in children and adults with lymphadenopathy to avoid inadequate antibiotic treatment, minimize imaging and to reduce hospitalization duration and costs.

FM 15

Recent advances in meningococcal B disease prevention: real world evidence from 4CMenB vaccination

Schaffner TO¹, Martinon Torres F², Banzhoff A³, Azzari C⁴, De Wals P⁵, Marlow R⁶, Marshall H⁷, Pizza M⁸, Rappuoli R⁸, Bekkat Berkani R⁹

¹Presenter on behalf of the authors, Local Medical Affairs, GSK, Münchenbuchsee, Switzerland; ²Translational Pediatrics and Infectious Diseases, Hospital Clínico Universitario de Santiago de Compostela, Santiago de Compostela, Spain; ³Global Medical Affairs, GSK, Marburg, Germany; ⁴Department of Health, University of Florence, Florence, Italy; ⁵Department of Social and Preventive Medicine, Laval University, Quebec University, Quebec City, QC, Canada; ⁶Bristol Medical School, University of Bristol, Bristol, United Kingdom; ⁷Adelaide Medical School, University of Adelaide, Adelaide, Australia; ⁸R&D, GSK, Siena, Italy; ⁹Global Medical Affairs, GSK, Rockville, MD, United States

Background and objectives

The 4-component meningococcal serogroup B (MenB) vaccine (4CMenB) was licensed in 2013 based on immunogenicity and safety data. Considerable real-world data describing its impact, effectiveness and safety have only recently accumulated following 4CMenB program introduction.

Methods

Available evidence on vaccine impact (VI), effectiveness (VE) and safety of 4CMenB in routine use were reviewed.

Learning points / discussion

Estimates of VE are available from 5 countries obtained during funded routine use in the United Kingdom (UK) and Italy; a healthcare setting in Portugal; a prospective observational study in South Australia; and outbreak control in Saguenay-Lac-Saint-Jean, Canada. VE of at least 3 doses of 4CMenB administered to infants ranged from 59.1% to 93.6%, and estimates were usually higher than predicted strain coverage rates using the Meningococcal Antigen Typing System (MATS). VE in children and adolescents (including 2 months to 20-year-olds in Quebec), was 100% in the first 2-3 years after vaccination. Effectiveness was sustained for 4 years in Quebec and for 2 years after the booster dose in young children vaccinated in infancy in the UK.

The impact of 4CMenB on MenB invasive disease was demonstrated in infants in the UK, Italy, and Spain, and in children/adolescents/young adults in prolonged outbreaks in Saguenay-Lac-Saint-Jean and South Australia. VI on university/college outbreaks cannot be measured due to the small number of cases. However, the absence of breakthrough cases after vaccine implementation is suggestive of VI. The safety profile of 4CMenB administered in real-world settings appears to reflect that established in pre-licensure clinical trials. No safety concerns have been raised in post-marketing surveillance.

The substantial body of data demonstrating 4CMenB effectiveness and impact in real-world settings supports its use in IMD prevention.

FUNDING: GlaxoSmithKline Biologicals SA

ACKNOWLEDGEMENTS: Business & Decision Life Sciences (Writer: Joanne Wolker; Coordinator: Carole Desiron).

This is an ENCORE of an abstract submitted at ESPID2021.

FM 16

From SARS-CoV2 tests to vaccinations and beyond: establishing a collaborative research network for primary care Pediatricians in Switzerland

Trück Johannes^{2,4,6}, Seiler Michelle^{3,4,6}, Berger Christoph^{2,4,6}, von der Heiden Ralf^{5,6}, Laasner Ursula^{5,6}, Bewer Silvestri Anna^{5,6}, Sidler Marc^{5,6}, von Rhein Michael^{1,4,6}

¹Child Development Center, University Children's Hospital, Zurich, Switzerland; ²Pediatric Infectiology, University Children's Hospital, Zurich, Switzerland; ³Pediatric Emergency Department, University Children's Hospital, Zurich, Switzerland; ⁴Children's Research Center, Zurich, Switzerland; ⁵Kinderärzte Schweiz (KIS), Berufsverband der Kinder- und Jugendärzte/-innen in der Praxis; ⁶SentiPED study group

To date, there is no systematic collection of data on the care of children in pediatric primary care in Switzerland, whose perspective is thus dramatically underrepresented in pediatric research. Therefore, the establishment of a nationwide network of practices participating in collaborative pediatric care research is urgently needed to address research questions related to the routine business of frontline pediatricians.

As a first step on this path, we have initiated a joint pilot project with more than 40 pediatric practices in the Canton of Zürich to prospectively and longi-

tudinally collect data on SARS-CoV-2 testing and clinical information on positive cases in outpatient children. Based on this successful collaboration between the University Children's Hospital Zurich and pediatric practices, we plan to expand our existing research database and to open it to all pediatric practices in Switzerland. As a next step, research questions on vaccinations, vaccination counseling, and well child visits in pediatric practices are planned. These questions are of broad interest to the pediatric community and are therefore likely to attract many participants, which will aid the creation of structural and organizational conditions for further research projects on outpatient pediatric care. This approach also allows to evaluate the implementation of recommendations and guidelines, and thus helps to improve evidence-based pediatric primary care in Switzerland.

We will present epidemiological data on ambulatory SARS-CoV-2 testing in the Canton of Zurich from May 2020 to date. It also includes clinical and transmission data on SARS-CoV-2 positive children. We will also share our experiences on the collaboration we have built so far, our next planned steps and the vision we are pursuing.

FM 17

Child and parental well-being and concerns during the COVID-19 pandemic: A longitudinal cohort analysis

Ehrler M^{1,2,5}, Latal B^{1,2,5}, Hagmann C^{2,3,5}, Kretschmar O^{2,4,5}, Wehrle F M^{1,2,3,5}

¹Child Development Center, University Children's Hospital Zurich, ZH, Switzerland; ²Children's Research Center, University Children's Hospital Zurich, ZH, Switzerland; ³Department of Neonatology and Intensive Care, University Children's Hospital Zurich, ZH, Switzerland; ⁴Department of Pediatric Cardiology, University Children's Hospital Zurich, Switzerland; ⁵University of Zurich, Switzerland

The COVID-19 pandemic has substantial impact on society beyond its immediate medical consequences. This study examines child and parental well-being and concerns in the course of the ongoing pandemic.

Families of 73 typically-developing children, 54 children born very preterm, and 73 children with congenital heart disease (CHD) were assessed prior to (T0), and during the 1st (T1) and the 2nd wave (T2) of the pandemic. Child and parental psychological well-being were assessed with validated questionnaires and changes over time were tested with mixed-effect regression models. Parental concerns related to medical and academic implications of the pandemic were compared between groups with Mann-Whitney-U-tests.

For children, a u-shaped trajectory of well-being was observed: Compared to before the pandemic, child well-being dropped during the 1st and subsequently recovered during the 2nd wave (time effect: $p < 0.001$), while parental well-being dropped during the 1st wave without a subsequent sufficient recovery (time effect: $p = 0.001$; t-test T1 vs. T2: $p = 0.203$). Overall, well-being did not differ between the three groups (all $p > 0.08$). Parents of children with CHD were more concerned about their child becoming infected with SARS-CoV-2 than other parents ($p = 0.03$) and parents of both at-risk groups were more concerned about potential negative effects of the school-closure than parents of typically-developing children ($p < 0.001$).

While parental well-being remained low in the course of the ongoing COVID-19 pandemic, the well-being of all typically-developing and at-risk children recovered during the 2nd wave. Potentially, the re-opening of schools has benefited this process. The concerns of parents of children with CHD or born very preterm may require particular attention as this pandemic evolves further.

FM 18

PEPS - Pediatric EPA Project for Switzerland: designing a consistent curriculum from pregraduate education to the specialist exam

Schuler Barazzoni Mirjam¹, Pharis Cosette², Cont Micol³

¹Service de néonatalogie, DFME, CHUV, Lausanne; ²Pédiatrie, Krankenhaus Sterzing, Oesterreich; ³Pädiatrie, Krankenhaus Sterzing, Oesterreich

In 2018, Switzerland adopted the PROFILES document, which consists in a revision of the objectives of undergraduate medical education with a shift to outcome based education. By the end of 2021, first residents who have been trained according to PROFILES will begin their specialization, which is a good time to adjust also the postgraduate training program, specifically in general pediatrics. The objective of this paper is to describe the revision process as well as its main outcome.

Summary of work

To date, the learning objectives for general pediatrics consists of a 16-chapter curriculum, time based, describing theoretical knowledge, practical skills and expected attitudes responding to the needs of training in the hospital environment as well as to future outpatient practice. This revision shifts to a competency-based program, close to the structure and the educational strategies of the pregraduate program. Its focus is clinical practice and it aims to adjust the professional future: ambulatory practice, hospital practice and academic pediatrics. A 2-round Delphi method is used to build consensus with the directors of pediatric residency programs in Switzerland.

Summary of results

The outcome is a short, user-friendly, pediatric learning objective catalogue, based on the structure of PROFILES. The main chapter includes the generic Entrustable Professional Activities (EPAs) of the pregraduate education, a list of core pediatric EPAs as well as specific EPAs according to the future professional practice. The other chapters describe the main roles of the doctor and a reference of theoretical knowledge as a basis for the specialty examinations.

Discussion and conclusions

The main challenge of this reform is to provide a learning framework that fits the needs of both the trainers who monitor the autonomization of the learners and the residents who must have a clear description of what is expected of them. The Delphi methodology lives to facilitate the adoption of change.

The general pediatrics postgraduate learning objectives will be built on the same logic as the pregraduate training in order to facilitate a longitudinal curriculum, as the reform of pre-graduate learning objectives needs to be followed by an adjustment of postgraduate training to ensure educational consistency.

SWISSPEDNET

SPN 2

Six years of experience in testicular tissue cryopreservation: Example of a multi-center network approach in Switzerland

Moussaoui D¹, Adam C², Diesch T³, Girardin C⁴, Bénard J⁵, Vidal I⁶, Bernard F^{7,8}, Busiah K⁹, Bouthors T⁹, Primi M¹⁰, Ansari M^{7,8}, Vuillimoz N¹⁰, Gumy-Pause F^{7,8}

¹Division of General Pediatrics, Department of Woman, Child and Adolescent Medicine, Geneva University Hospitals, Geneva, Switzerland; ²Division of Pediatrics, Oncology and Hematology Unit, Woman-Mother-Child Department, Lausanne University Hospital, Lausanne, Switzerland; ³Division of Pediatric Oncology-Hematology, University Children's Hospital of Basel Basel, Switzerland; ⁴Pediatric Endocrine and Diabetes Unit, Department of Woman, Child and Adolescent Medicine, Geneva University Hospitals, Geneva, Switzerland; ⁵Unit for Reproductive Medicine and Gynecological Endocrinology, Department of Woman, Child and Adolescent Medicine, Geneva University Hospitals, Geneva, Switzerland; ⁶University Center of Pediatric Surgery of Western Switzerland, Division of Pediatric Surgery, Department of Woman, Child and Adolescent Medicine, Geneva University Hospitals, Geneva, Switzerland; ⁷Pediatric Onco-Hematology Unit, Department of Woman, Child and Adolescent Medicine, Geneva University Hospitals, Geneva, Switzerland; ⁸CANSEARCH research platform in oncology and hematology pediatric, Department of Woman, Child and Adolescent Medicine, Faculty of Medicine, University of Geneva, Geneva, Switzerland; ⁹Pediatric Endocrinology and Diabetology Unit, Division of Pediatrics, Woman-Mother-Child Department, Lausanne University Hospital, Lausanne, Switzerland; ¹⁰Fertility Medicine and Gynecologic Endocrinology Unit, Woman-Mother-Child Department, Lausanne University Hospital, Lausanne, Switzerland

Introduction

One of the major adverse effects of oncologic treatments and hematologic stem cell transplantation is the risk of infertility. According to the Childhood Cancer Survivor Study, prevalence of infertility in male is 46%. Testicular tissue cryopreservation (TTC) is an experimental procedure of fertility preservation for prepubertal boys aiming at preserving spermatogenic stem cells, which could be used for assisted reproduction in the future. We report our experience with a research protocol of TTC.

Method

Data were prospectively collected on patients who underwent TTC at the Lausanne University Hospital, Geneva University Hospitals and Basel University Children's Hospital between 2015 and 2020. Inclusion criteria were pre-pubertal boys aged more than 3 months or peri and post-pubertal boys with unsuccessful cryopreservation of sperm, who were scheduled to undergo high-risk gonadotoxic treatment (risk of infertility estimated to 80% or more). The Andrology and Reproductive Biology Laboratory at Lausanne University Hospital centralized all samples. This study was approved by the local ethics committee and registered with clinicaltrials.gov (NCT03180918).

Results

Testicular tissues from 35 patients were collected. Four families declined TTC. The mean age of patients was 8.5 years (range 0.5-18.5 years). Underlying diseases were a malignant disorder in 31 patients including 15 (42.9%) hematological malignancies (11 leukemias and 4 lymphomas) and 16 (45.7%) solid tumors (2 rhabdomyosarcomas, 2 osteosarcomas, 1 Ewing sarcoma, 4 medulloblastomas, 1 germ cells tumor, 5 neuroblastomas and 1 nephroblastoma). Four (11.4%) patients had a non-malignant disease (3 thalassemias and 1 sickle cell disease). The main indication for TTC was conditioning for hematologic stem cell transplantation (25 patients, 71.4%). A minor hematoma and a minor wound dehiscence were the only two reported adverse events. Tumoral cells were found in one testicular biopsy of a leukemic patient. Five patients died during the follow-up and their testicular tissue was destroyed according to the parents' wish.

Conclusion

Our data demonstrate the feasibility of a program of TTC coordinated by a multidisciplinary team of fertility preservation. Advances in assisted reproduction open great hopes to reduce adverse effects of gonadotoxic treatments.

The authors wish to thank the Zoé4life Association for its financial support.

SPN 3

Procedural Sedation and Analgesia in Switzerland: a National Survey of Tertiary Care Pediatric Emergency Departments

Sahyoun Cyril¹, Cantais Aymeric¹, Löllgen Ruth²

¹Service d'Accueil et d'Urgences Pédiatriques, Hôpitaux Universitaires de Genève, Geneva, Switzerland; ²Pediatric Emergency Department, Inselspital, University Hospital, University of Bern, Bern, Switzerland

Introduction

Procedural sedation and analgesia (PSA) have become standard in managing pain and anxiety in children undergoing procedures outside of the operating theatre. The objectives of this study are to describe the current PSA practice patterns in Swiss pediatric emergency departments (PEDs) and to perform a needs assessment-like analysis.

Materials and Methods

Using a survey methodology, we conducted an exploratory descriptive 30-question survey of tertiary care PEDs through the Pediatric Emergency Medicine Switzerland (PEMS) network. Study data were collected and managed using a REDCap electronic database. Descriptive statistics were performed. This study is a sub analysis of a larger, Europe-wide survey.

Results

Nine PEDs participated in this study, representing 9 cantons. The number of patients seen per year, per site, was 28,100 (95th CI 21,000 – 35,200).

The following medications were available for use in children: midazolam (9/9), ketamine (6/9), and propofol (4/9). Intranasal medications available for use in children included fentanyl (8/9 sites), and dexmedetomidine (1/9 sites). Nitrous oxide was available in all sites and chloral hydrate in 7/9.

Six out of 9 sites had specific PSA curriculum/courses required for the staff administering the sedation while 8/9 had general safety rules for administering sedation. In 7/9 sites, all physicians performing PSA were certified in an advanced life support course for children. Eight out of 9 sites reported having physician staffing issues at least sometimes.

All PEDs had a protocol which allows nurses to give analgesics from triage. All hospitals had LET/LAT gel and EMLA available, while 8/9 had skin glue available. Only 2/9 sites had accessed to Child Life Specialists and to hypnosis. When asked how satisfied with the management of pain and anxiety of children during procedures in their department, 6/9 reported being somewhat satisfied, 2/9 somewhat unsatisfied, and 1/9 very satisfied. In most domains, the surveyed sites had greater access to sedation medications and more safety guidelines in place than other European sites.

Conclusion

Procedural sedation and analgesia are prevalent in Swiss pediatric emergency departments, and sedation medications are widely available. Guidelines are common, as well as nurse-directed triage analgesia. Dedicated nonpharmacologic support to procedural sedation and analgesia is however lacking. Barriers to implementation include staff shortage, and lack of space.

SPN 4

Long-term follow-up for childhood cancer survivors: the Geneva experience

Babecoff* S¹, Mermillod* F¹, Fernandez E², Marino D³, Gayet-Ageron A⁴, Ansari M^{3,5}, Gumy-Pause F^{3,5}

¹Faculty of Medicine, University of Geneva, Geneva, Switzerland; ²Department of Oncology, University Hospitals of Geneva, Geneva, Switzerland; ³CANSEARCH research platform in pediatric oncology and haematology, University of Geneva, Geneva, Switzerland; ⁴Division of Clinical Epidemiology, University Hospitals of Geneva, Geneva, Switzerland; ⁵Department of Women, Children and Adolescents, Onco-Hematology Unit, University Hospitals of Geneva, Geneva, Switzerland

Aims of the study

Although the 5-year survival for pediatric cancer in Switzerland today is over 85%, two thirds of the survivors will develop chronic health conditions (CHCs) due to the disease or to the toxicity of treatments. In this context, a long-term personalized follow-up program (LTFU program) was set up at the University Hospitals of Geneva (HUG) since 2015. We aimed to describe this program, more particularly the cumulative burden of the CHCs, and assess the satisfaction of patients and/or their parents with it.

Methods

A monocentric retrospective study was performed where data on follow-ups and CHCs were collected from medical charts of people who had childhood cancer and who participated in the LTFU program. CHCs were classified and graded in severity with the Common Terminology Criteria of Adverse Events (CTCAE) classification, version 5.0. This study was completed by a satisfaction survey among patients and/or their parents.

Results

Out of 83 eligible patients, 51 (61.4%) accepted to participate, with an average age of 17.4 years (range, 10 to 35) at the time of study. Forty-two patients (82.3%) presented one or more CHCs and almost half of them (20/42) described 3 or more CHCs. The total number of Grade CTCAE 1-4 CHCs was 118 for the 51 participants, with a mean of 2.3 (range, 0 to 7) disorders per patient. The most frequently affected systems were neurological (14.4%), musculoskeletal (13.6%), endocrine (9.3%) and renal (9.3%) systems. Sarcoma, central nervous system tumors and neuroblastoma were the diagnoses associated with the highest average number of CHCs. Among the 118 questionnaires sent to patients and/or parents, we received 82 (69.5%) responses. The level of satisfaction was good to excellent for more than 90% of the participants, for all the items evaluated.

Conclusions

Childhood cancer survivors present a significant number of CHCs, confirming the need for appropriate long-term, multidisciplinary and patient-specific medical follow-up based on the primary diagnosis and therapies received. Moreover, the LTFU program at the HUG was highly appreciated by patients and/or their parents and this motivates its permanent conduct.

SPN 5

The Swiss Rare Disease Registry: Towards a nationwide platform for rare disease

Fux Michaela¹, Tschertner Ann¹, Spörri Adrian², Baumgartner Matthias³, Kuehni Claudia¹

¹Swiss Rare Disease Registry, Institute of Social and Preventive Medicine, University of Bern, Switzerland; ²SwissRDL, Medical Registries and Data Linkage, Institute of Social and Preventive Medicine, University of Bern, Switzerland; ³Division of Metabolism and Children's Research Center, University Children's Hospital, Zurich, Switzerland

Introduction

Over 7000 rare diseases affect 5-8% of people worldwide. In Switzerland, more than 100,000 children suffer from a rare disease. But their health-related data are either inaccessible or fragmented in disease-specific registries leading to inadequate management, diagnostic delay, and heterogenous treatment. With a national population-based registry for all rare diseases, the Swiss Rare Disease Registry (SRDR) aims to collect a core data set from all patients with rare diseases in Switzerland, approximately around 500 000 people, in a cost-efficient way.

Material and Methods

The SRDR core data set includes the common data elements for rare disease registration proposed by the European Commission's Joint Research Centre. Physicians in hospitals and private practice will identify patients with rare diseases during regular consultations and assign ORPHA Codes, a unique coding system for rare diseases. We offer automatic transfer of data to the server via a web service or a web client tool. The core data will be supplemented with data already available in other data sources, such as the Federal Statistical Office or the hospital warehouse data lakes from the Swiss Personalised Health Network (SPHN). Also existing disease-specific registries have the possibility to link their data to the SRDR database. Nested questionnaire surveys and clinical studies can be added as specific modules.

Results

In 2015-2017, an interdisciplinary working group of stakeholders developed the concept of the SRDR, which was commented in a formal consultation process and obtained ethical approval in 2018. In autumn 2020, the Federal Office

of Public Health (FOPH) approved funding under article 24 of the national law on cancer registration. We are currently engaging with University Hospitals, Children's Hospitals, and Centres for Rare Diseases to define processes of collection of informed consent and data transfer. Moreover, we develop strategies to link data from disease-specific registries.

Conclusion

The SRDR will constitute a national platform for rare diseases that will make people with rare diseases visible in health statistics and research. The registry will enable clinical and epidemiological studies on rare diseases, improve the diagnosis, treatment, and quality of life of people with rare diseases in Switzerland, and facilitate patients' participation in national and international studies.

SPN 6

Rotavirus disease and health care utilisation among children under 5 years of age in highly-developed countries

Ardura-Garcia C¹, Kreis C¹, Rakic M¹, Jaboyedoff M^{1,2}, Mallet MC^{1,3}, Low N¹, Kuehni CE^{1,4}

¹Institute of Social and Preventive Medicine, University of Bern, Bern, Switzerland; ²Department of Paediatrics, Vaud University Hospital, Lausanne, Switzerland; ³Graduate School for Health Sciences, University of Bern, Mittlestrasse 43, 3012 Bern, Switzerland; ⁴Division of Respiratory Medicine, Dept of Paediatrics, University Hospital of Bern, Switzerland.

Background

Rotavirus (RV) infection is the leading cause of diarrhoea-associated morbidity and mortality globally among children under 5 years. RV vaccination is available but has not been universally introduced in national immunisation plans, especially in highly developed countries such as Switzerland. We aimed to estimate the prevalence and incidence of RV gastroenteritis (RVGE) among children under 5 years in highly developed countries that have not introduced routine RV vaccination.

Methods

We searched PubMed and Embase from 2000 to 2018, using broad search terms with no language restrictions. We included publications reporting on incidence or prevalence of RVGE-related healthcare use in children under 5 years: primary care visits, emergency department consultations and hospitalisations. We included only studies with laboratory confirmed RV infection, undertaken in highly developed countries with no RV routine vaccination plans. We used random effects meta-analysis to generate summary estimates.

Results

We screened 4033 abstracts and included 75 studies from 20 countries. Summary estimates of the average proportion of RVGE among all episodes of acute gastroenteritis were: 21% (95% CI 16-26%) for primary care visits; 32% (25-38%) for emergency department consultations; and 41% (36-47%) for hospital admissions. The summary estimates of average incidence rates of RVGE related episodes per 100 000 person-years were: 2484 (95% CI 697-5366) primary care visits; 1890 (1597-2207) emergency department consultations; and 500 (422-584) hospitalisations. There were large variations between and within countries. Heterogeneity was high (>90%) in most models.

Conclusion

RV causes an important proportion of consultations and hospitalisations for gastroenteritis in highly developed countries without routine RV vaccination. Cross-country studies using standardised methods would allow more accurate estimates that could be used to model the burden of disease and cost-effectiveness of introducing RV vaccine in highly developed countries.

Funding: GlaxoSmithKline Biologicals SA (HO-18-19250) and University of Bern.

SPN 7

Effect of COVID19 preventive measures on respiratory infections and symptoms in children

Ardura-Garcia C¹, Mallet MC^{1,2}, Pedersen ESL¹, Berger D¹, Barben J³, Blanchon S⁴, Casaulta C⁵, Hoyler K⁶, Jochmann A⁷, Moeller A⁸, Mueller-Suter D⁹, Regamey N¹⁰, Latzin P¹¹, Kuehni CE^{1,12}

¹Institute of Social and Preventive Medicine, University of Bern, Switzerland.; ²Graduate School for Health Sciences, University of Bern, Mittlestrasse 43, 3012 Bern, Switzerland; ³Division of Paediatric Pulmonology, Children's Hospital St. Gallen, St. Gallen, Switzerland; ⁴Paediatric Pulmonology and Cystic Fibrosis Unit, Service of Paediatrics, Depart-

ment Woman?Mother?Child, Lausanne University Hospital, Lausanne, Switzerland; ⁵Division of Paediatric Pulmonology, Children's Hospital Chur, Chur, Switzerland; ⁶Division of Paediatric Pulmonology, Private Paediatric Practice Horgen, Horgen, Switzerland; ⁷Department of Paediatric Pulmonology, University Children's Hospital Basel, Switzerland; ⁸Division of Paediatric Pulmonology, University Children's Hospital Zurich, Zurich, Switzerland; ⁹Division of Paediatric Pulmonology, Kantonsspital Aarau, Aarau, Switzerland; ¹⁰Division of Paediatric Pulmonology, Children's Hospital Lucerne, Lucerne, Switzerland; ¹¹Paediatric Respiratory Medicine, Children's University Hospital of Bern, University of Bern, Bern, Switzerland; ¹²PedNet, Children's University Hospital of Bern, University of Bern, Bern, Switzerland

Background

Little is known on the effect of COVID19 preventive measures on respiratory symptoms in children. We studied the effect of these measures on reported respiratory symptoms in children previously followed by paediatric pulmonologists for respiratory problems.

Methods

The Swiss Paediatric Airway Cohort (SPAC) is a prospective cohort of children aged 0-16 years seen at respiratory outpatient clinics in Switzerland. Parents completed a monthly online questionnaire on respiratory symptoms and treatment for 1 year. We compared the questionnaires completed between October 2019-February 2020 (preCOVID-winter), March-April 2020 (lockdown with school closures) and October 2020-February 2021 (COVID-winter). We used panel data logistic regression to study the association between the period and respiratory infections, symptoms and treatment use. Results were adjusted for sex, age and previous severity of respiratory symptoms.

Results

Among the 392 questionnaires (204 children, mean age 9 years, 48% female), 83 were completed during preCOVID-winter, 91 during lockdown and 218 during COVID-winter. Compared to preCOVID-winter, questionnaires completed in lockdown and COVID-winter were less likely to report on respiratory infections (aOR 0.12, 95%CI 0.05-0.31; aOR 0.22, 0.10-0.46), cough (aOR 0.31, 0.14-0.67; aOR 0.17, 0.08-0.36), dyspnoea (aOR 0.30, 95%CI 0.10-0.96; aOR 0.34, 0.11-1.1), wheeze (aOR 0.15, 0.05-0.47; aOR 0.30, 0.12-0.71), treatment for respiratory problems (aOR 0.22, 0.06-0.85; aOR 0.06, 0.01-0.30), and asthma control-drugs use (aOR 0.65, 0.23-1.8; aOR 0.14, 0.04-0.49).

Conclusion

Children followed by paediatric pulmonologists for respiratory problems had less respiratory infections and symptoms during the COVID19 pandemic compared to a previous similar period, probably due to preventive measures.

Funding: SNSF (320030_162628)

SPN 8

Acute abdominal emergency due to infectious enteritis: a matched case-control study comparing Campylobacter spp. to other enteric pathogens

Papathanasiou Terzi M A¹, Prod'hom G², Croxatto A², Andrey V³, Crisinel P A⁴

¹Service of Pediatrics, Department Women-Mother-Child, Lausanne University Hospital, Lausanne, Switzerland; ²Institute of Microbiology, Department of Laboratory, University of Lausanne & Lausanne University Hospital, Lausanne, Switzerland; ³Service of Pediatric Surgery, Department Women-Mother-Child, Lausanne University Hospital, Lausanne, Switzerland; ⁴Unit of Pediatric Infectious Diseases and Vaccinology, Department Women-Mother-Child, Lausanne University Hospital, Lausanne, Switzerland

Background

Campylobacter spp. is a frequent cause of gastroenteritis, presenting in some patients as acute abdominal emergency. Here we describe the distinctive clinical characteristics of these patients.

Methods

We designed a retrospective single-center case-control study. Children and adolescents under 18 years of age who had positive stool cultures with Campylobacter spp. during the period from June 1, 2008 and May 31, 2016 were identified from our database. Hospitalized patients with Campylobacter spp. were then matched for age and gender with patients hospitalized for gastro-enteritis of other or unknown etiology. Patients who had undergone abdominal radiographic investigation or received a surgery consultation were included as "acute abdomen" (AA) cases. Demographics, clinical characteristics and management were compared between AA and non-AA cases.

Results

One hundred and forty-one patients with Campylobacter spp.-positive cultures were included in the analysis. Nineteen patients were identified as AA cases. They had less frequent diarrhea (14/19, 74% vs 117/121, 97, p = 0.02) and reported a lower sense of general wellbeing (8/18, 44% vs 8/108, 7%; p<0.001). Localized pain (9/18, 50% vs 20/115, 17%; p = 0.002) and abdominal tenderness (3/18, 11% vs 0/111; p = 0.02) were also more common among AA cases. Forty-four patients with Campylobacter spp. infection were hospitalized and matched with 44 patients with gastro-enteritis from other or unknown etiology. Campylobacter spp. infection (OR 5, 95% CI 1.4-18.4; p = 0.015) and localized pain (OR 10.1, 95% CI 2.6-39.1; p = 0.001) positively correlated with being seen by a surgeon and/or prescription for radiological examination.

Conclusions

We identified a subset of patients with Campylobacter spp. gastroenteritis who present as acute abdominal emergency. The presentation of these patients was characterized mainly by the nature of the associated abdominal pain.

SPN 9

Prevalence and Clinical Risk Factors of Severe Hearing Loss in Swiss Childhood Cancer Survivors

Strebel Sven^{1,2}, Mader Luzius¹, Luzi Fabienne¹, Waespe Nicolas^{1,2}, Weiss Annette¹, Parfitt Ross³, am Zehnhoff-Dinnesen Antoinette³, Kompis Martin⁴, von der Weid Nicolas⁵, Ansari Marc^{2,6}, Kuehni Claudia^{1,7}

¹Institute of Social and Preventive Medicine, University of Bern, Switzerland; ²Research Platform for Pediatric Onco-Hematology, Department of Pediatrics, Gynecology and Obstetrics, University of Geneva, Geneva, Switzerland; ³Department for Phoniatrics and Pedaudiology, Universitätsklinikum, Münster, Germany; ⁴Department of ENT, Head and Neck Surgery, University Hospital Bern, University of Bern, Bern, Switzerland; ⁵Department of Pediatric Oncology and Hematology, University Children's Hospital Basel (UKBB), University of Basel, Basel, Switzerland; ⁶Department of Women, Child and Adolescent, Division of Pediatric Oncology and Hematology, Geneva University Hospital, Switzerland; ⁷Division of Pediatric Hematology/Oncology, Department of Pediatrics, Inselspital, Bern University Hospital, University of Bern, Bern, Switzerland

Background

Hearing loss is a known late effect in childhood cancer survivors (CCS) after ototoxic treatments – in particular platinum chemotherapy, cranial radiation with doses of ≥30 Gray, and surgery involving the auditory system. To date, we have not studied the prevalence and clinical risk factors for severe hearing loss based on audiogram data from CCS at a national level in Switzerland.

Aim

To assess the prevalence of severe hearing loss in CCS after platinum chemotherapy using audiogram data and to determine clinical risk factors for severe hearing loss.

Method

As part of the international collaborative research project PanCareLIFE, we performed a chart review of all CCS who were diagnosed at age ≤18 years in Switzerland and treated with platinum chemotherapy in the nine pediatric oncology clinics between 1990 and 2014. Audiograms, treatment-related information and socio-demographic data were extracted from medical records and from the Childhood Cancer Registry (ChCR). Two raters independently graded all audiograms based on the Muenster Ototoxicity Scale. We defined severe hearing loss as ≥grade 3 of the last audiogram. We used multivariable logistic regression to assess associations between risk factors and hearing loss.

Results

We analyzed data from 302 CCS (56% male; mean age at diagnosis 7.2 years (SD: 5.2)), mean time from cancer diagnosis to last audiogram 5.4 years (SD: 4.3). Prevalence of severe hearing loss (Muenster \geq grade 3) was 23% (95% CI: 19-28). Severe hearing loss was associated with younger age at diagnosis (odds ratio [OR] for <5 years 2.6, 95% CI: 1.4-5.0), treatment in 1990-1999 vs. 2000-2014 (OR 2.2, 95% CI: 1.2-4.0), higher cumulative doses of cisplatin (OR 2.5, 95% CI: 1.1-5.5 for 300-500 mg/m² and OR 3.8, 95% CI: 1.5-9.7 for >500 mg/m²;) and concomitant cranial radiation with cumulative doses over 30 Gray (OR: 2.2, 95% CI: 1.1-4.3), but not with gender and cumulative doses of carboplatin.

Conclusion

One in four CCS treated with platinum chemotherapy in Switzerland suffers from severe hearing loss. Patients aged less than 5 years when treated with high cumulative doses of cisplatin and cranial radiation could profit from close monitoring of their hearing function.

Funding

We would like to express our gratitude to the CANSEARCH Foundation and to the Swiss Cancer League (Grant No: HSR-4951-11-2019) for the financial support of the study.

SPN 10

Children with congenital heart disease after open-heart surgery – A survey on developmental follow-up and early interventions, along with an assessment of parental needs.

Ursprung P^{S1}, Knirsch W², Kretschmar O², Von Rhein M¹, Jenni O¹

¹Developmental Paediatrics, University Children's Hospital Zurich, Switzerland; ²Children's Heart Center Zurich, University Children's Hospital Zurich, Switzerland

Background

Congenital heart disease (CHD) is the most common birth defect affecting approximately 6-8:1'000 life born children. In Switzerland, 600-800 children are born with a CHD every year. Swiss recommendations suggest systematic evaluation of development at 1, 2 and 5 years of age to accompany and consult families, detect developmental delays, and to initiate early interventions, if necessary. The implementation of this recommended follow-up (FU) has not yet been examined in Switzerland. First analyses of the Swiss neurodevelopmental Outcome Registry of Children with severe congenital heart Disease (ORCHID), which has been established in 2019, showed that the follow-up rate of this at-risk population is far behind expectations. Furthermore, parents of children with CHD are facing increased stress, which can have a negative impact on their mental and physical health. So far, it is unclear which support services actually help them in coping with everyday life and in dealing with their children's illness, or which ones are still lacking.

Methods

Our study includes children with CHD (born in 2015/2016) in whom open-heart surgery on cardiopulmonary bypass was performed at the University Children's Hospital Zurich in the first year of life (N = 221). Exclusion criteria are death (n = 21), residence abroad (n = 5), and poor German language skills. We just started a telephone survey for quality assurance with the parents of these children and combine results with data from medical records. Following this, we conduct a more detailed survey of developmental delays, their treatment, and the parental needs. The study is expected to be completed in the spring of 2021.

The talk will present the follow-up rate of children with CHD after open heart surgery, where FU is being performed, and if children with developmental impairments are successfully allocated to early interventions. Furthermore, we will present factors which interfere with a high follow-up rate, and whether the existing early therapeutic and support services match the needs of families with children with operated CHD.

SPN 11

Consultation and Communication with Vaccine Hesitant Patients: An Expert Focus Group Discussion Study

Buhl A^{1,2}, Schmid Thurneysen L^{1,3}, Deml M^{1,2}, Tarr P^{1,3}

¹University of Basel, Basel, Switzerland; ²Swiss Tropical and Public Health Institute (Swiss TPH), Basel, Switzerland; ³University Department of Medicine and Infectious Diseases Service, Kantonsspital Baselland, Bruderholz, Switzerland; *contributed equally

Background

Trusting relationships between patients and healthcare providers (HCPs) during vaccine consultations play determinative roles in vaccination decisions. Our National Research Program (NRP74) has a particular focus on the role of complementary/alternative medicine (CAM) providers in vaccination decision-making since they and their patients have often been depicted as vaccine hesitant (VH). However, our research has shown that CAM provider consultations typically focus on establishing trust and involving patients in vaccination decisions.

Methods

We conducted a longitudinal qualitative focus group discussion (FGD) study between April 2019 and November 2020 with a total of 6 FGDs with 5-10 experienced HCPs with different backgrounds ("core group" of 5 CAM providers in each FGD, complemented by paediatricians, general practitioners, vaccine experts from federal office of public health, and federal vaccination commission). Our aim was to gain insights into HCPs' approaches to achieving effective vaccination consultations with VH patients. FGDs followed pre-determined topics with stimulus material used to elicit debates.

Results

Participants agreed that rather than providing standardized explanations, providers should tailor vaccination counseling to patients' individual needs to be effective, i.e. sincere communication to build trust and address concerns. CAM participants did not describe VH patients as problematic. Rather, they interpreted VH as an expression of patients' active, critical appraisal of vaccination information. Participants discussed the need to identify trustworthy, scientifically sound vaccination information and to translate this into advice that was useful for patients. CAM participants often perceived vaccine messaging by health authorities as "pro-vaccination propaganda". They recommended individualizing the Swiss vaccination schedule and they rejected vaccine mandates.

Conclusion

Interestingly, including experts with diverse training backgrounds led to productive discussions on VH rather than controversy. All participants considered the FGDs to be enriching, fruitful, and a valuable opportunity to exchange ideas. The FGD results informed the design of an online survey for HCPs relating to their vaccine communication needs, and the ongoing development of a vaccination counseling intervention for HCPs.

SPN 12

5-years outcome in pediatric celiac disease in the era of the non-biopsy diagnostic approach

Klöti S¹, Schaad J^{1,2,3}, Sokollik C², Spalinger J^{2,3}, Righini F³

¹Department of pediatrics, Children's hospital Lucerne, Switzerland; ²Department of pediatric gastroenterology, hepatology and nutrition, University children's hospital Inselspital Bern, Switzerland; ³Department of pediatric gastroenterology, hepatology and nutrition, Children's hospital Lucerne, Switzerland

Introduction

Until 2011, histological proof of villous atrophy in the small bowel was the gold standard to diagnose celiac disease (CD) in pediatric patients. After 2011, diagnosis of CD was possible without biopsy when following criteria were fulfilled: symptoms suggestive of CD, positivity of HLA-DQ2 or -DQ8, tissue transglutaminase antibodies (anti-tTG-IgA) \geq 10 times the upper limit of normal and positivity of endomysium antibodies (EMA). Latest ESPGHAN-guidelines in 2019 allowed diagnosis of CD with the non-biopsy approach in a subgroup with very high antibodies even without consideration of the clinical presentation and genetic testing. Up to date, there hardly exists data comparing the biopsy versus the non-biopsy diagnostic approach. The aim of this study was to compare outcomes in patients with non-biopsy diagnosed CD to those diagnosed with additional biopsy.

Method

Retrospective study of CD patients during a five-year follow-up. All patients with diagnosis of CD and follow-up available in the pediatric gastroenterology at Children's hospital in Lucerne between January 2017 and December 2019 were included.

Patients diagnosed by biopsy were compared to patients with exclusively serological confirmed diagnosis. Values are presented as medians (interquartiles) for continuous variables and as frequency (%) for qualitative variables. Chi2 and Wilcoxon rank sum test are used for univariate analysis. All analyses were performed using SAS software, version 9.3 (SAS Institute, Cary, NC).

Results

In a CD pediatric cohort with more than ten-fold elevated anti-tTG-IgA at diagnosis, 115 patients with CD diagnosis confirmed by biopsy were compared to 25 patients with exclusively serological confirmed diagnosis. Median age at diagnosis was 8.58 years (3.7, 11.2) vs 4.92 years (3, 9.6) at time of diagnosis ($p = 0.12$). Median time to negativity of anti-tTG-IgA was comparable in both groups (13 months (12, 25) vs 13 months (12, 24.5) respectively, $p = 0.66$). Weight and height were comparable in both groups at diagnosis and during follow-up ($p > 0.05$).

Conclusion

This is one of the first studies, comparing outcomes of patients with CD diagnosis confirmed by biopsy to those with exclusively serological confirmed diagnosis. In absence of significant difference of laboratory values and anthropometric data between the two groups during follow-up, this study is supporting the validity of the non-biopsy diagnostic approach.

SPN 13**Early diagnosis and treatment in children with autism spectrum disorder: Experience from Switzerland's Italian region.**

Riederer C¹, Pifferini R¹, Rizzi E¹, Glaser B¹, Zanda N¹, Soldini E², Ramelli GP¹

¹*Servizio di Neuropediatria, Istituto Pediatrico della Svizzera Italiana, Bellinzona, Switzerland;* ²*Dipartimento economia aziendale, Sanità e socialità, Scuola Università Professionale;* ³*della Svizzera Italiana, Manno, Switzerland*

Introduction

There is not yet a definitive cure for autism spectrum disorder (ASD), but early therapeutic intervention can allow a child to acquire social skills that significantly improve his or her quality of life and allow for integration into the mainstream school system. In the canton of Ticino, an outreach program targeted paediatrician awareness of early signs of ASD and established a database for recording data on the patients. The objective of this pilot study was to better understand therapeutic approaches that lead to integration into regular school.

Method

Data were collected from 126 children diagnosed with an autism spectrum disorder at the Cantonal Neuropediatric Department between January 2009 and December 2019. Three children were excluded because they moved out of Ticino during the study. Only children who reached school-age (i.e. born between 2005 and 2014, 83 in total) were included in the current study. Univariate descriptive statistics were used to describe the sample. The relationships between type of schooling and other therapeutic variables were explored using non-parametric statistical tests (chi-squared tests, Mann-Whitney tests, Kruskal-Wallis tests).

Results

The sample consisted of 83 children (70 boys and 13 girls). 50 (60%) children were mainstreamed and 33 (40%) of them were in special education. Among participants, three categories of treatment were reported: intensive behavioural therapy (> 15 hours/month) ($n = 23$ children); limited therapy (< 15 hours/month) ($n = 30$ children) and no behavioural therapy ($n = 30$ children). The bivariate analyses demonstrated a significant difference between the two groups of children (those who were included in mainstream schooling and those in a special education program) for the following variables: age at diagnosis, M-CHAT screening test scores, ADOS-2 test scores, IQ and behavioural therapy program. The results showed that intensive behavioural therapy is significantly associated with regular schooling. Without this intensive behavioural therapy, integration into regular schooling is possible only for high functioning children.

Conclusion

This is the first Swiss study based on a regional population of children with autism that highlights the importance of early diagnosis and behavioural therapy for integration into regular schooling. Our results also show that the type of treatment makes the biggest difference to the outcomes of children with medium-to-severe autism.

SPN 14**Does age influence self-perception of the soft-tissue profile in children?**

Varatharaju Vysnave¹, Caffish Marianne², Soroken Cindy², Kiliaridis Stavros¹, Antonarakis Gregory S.¹

¹*Division of Orthodontics, University Clinics of Dental Medicine, University of Geneva, Switzerland;* ²*Department of Pediatrics, Geneva University Hospitals, Switzerland*

Introduction

Appreciation of the soft-tissue profile is important in orthodontic diagnosis and treatment. However, are the patients themselves aware of their profile appearance? We aimed to evaluate if age influences self-perception of the soft-tissue profile in children.

Methods

The study population for this prospective cross-sectional investigation consisted of 3 groups of 60 patients, classified according to age (<12 years; 12-15 years; >15 years). Each subject's right-sided facial profile was photographed to obtain a silhouette. Facial profile silhouette templates were created to represent the local population. Each subject's photograph was inserted into the corresponding template, and the subjects were asked to identify themselves. Facial profile self-recognition was recorded as a binary variable (yes or no). Other recorded variables included age, sex, and sexual maturity rating (using Tanner staging). Chi-square tests were used to analyze facial profile self-recognition between different subgroups, and stepwise multiple regression was used to predict the probabilities of facial profile self-recognition, with age, sexual maturity rating, and other recorded variables as independent variables.

Results

Eighty percent of subjects aged >15 years recognized their own profile, compared with only 55% and 50% of subjects aged 12-15 years and <12 years, respectively. Subjects aged >15 years were significantly more likely to recognize their profile than younger subjects ($P = 0.001$). Similarly, subjects with the most advanced sexual maturity rating (stage V) were significantly more likely to recognize their profile (85% self-recognition) than those in groups I-IV ($P < 0.001$). Girls were more likely to recognize their profiles than boys ($P = 0.028$). When using multiple regression analysis, sexual maturity rating appears to be the only significant predictor for facial profile self-recognition ($R^2 = 0.25$; $P < 0.001$).

Conclusions

Facial profile self-recognition seems to improve with age and sexual maturity (sexual maturity rating stage V). Because orthodontic treatment planning takes possible soft-tissue changes into account, it is important to evaluate the degree of self-perception of the patients to adapt our goals and treatment discussions.

SPN 15**The importance of a comprehensive Primary Ciliary Dyskinesia diagnostic center – Experience of the first 100 patients at the PCD-UniBe Diagnostic Center**

Müller Loretta¹, Savas Sibel T¹, Tschanz Stefan², Kieninger Elisabeth¹, Bullo Marina¹, Casaulta Carmen¹, Latzin Philipp¹, PCD-UniBe study group

¹*Pediatric Pneumologie, Kinderklinik, Inselspital Bern, Department for BioMedical Research (DBMR), Universität Bern, Schweiz;* ²*Institut für Anatomie, Universität Bern, Schweiz*

Primary ciliary dyskinesia (PCD) is a rare genetic disorder characterized by immotile/dyskinetic respiratory cilia. Patients suffer from respiratory symptoms since birth. Since there is no gold standard test, diagnosis is challenging and requires different methods and a lot of expertise. Therefore, we established the comprehensive PCD-UniBe diagnostic center.

Our diagnostic approach includes nasal nitric oxide measurement, nasal brushing and air-liquid-interface (ALI) cell culture for all patients with subsequent analysis of ciliary motility by high-speed-video-microscopy (HSVM) and immunofluorescence staining (IF) of structural proteins. In selected patients, we perform electron microscopy (TEM) of the ciliary ultrastructure and genetic analysis. An interdisciplinary PCD-board brings the results to a diagnosis.

Between 01/2018 and 04/2020, we were assigned the first 100 cases from all over Switzerland (mean age 12.9a, median 5.6a, range: 16 days to 68.9a, 17 adults). For all cases, we performed fresh HSVM and cell culture (90% success rate). Cell cultures provide samples without secondary effects, more cell groups and cilia to analyze. We performed HSVM ALI on successful cultures

and IF on all cases (51x fresh, 78x ALI, some both). For 29 patients we performed TEM (27x ALI, 5x fresh; 3x both) and genetic panel analysis. The use of cell cultures dramatically reduced inconclusive findings for HSVM (37% fresh versus 12% ALI) and IF (53% versus 15%). We did not find any evidence of PCD in 78 cases, diagnosed PCD in 18 patients and further analyses are ongoing in four patients. HSVM was diagnostically significant in twelve, IF in 14, TEM in five, and genetics in ten cases.

This indicates that none of the methods alone was sufficient to diagnose all 18 PCD cases and that a comprehensive approach is essential for accurate PCD-diagnosis. Cell culture was crucial and prevented re-brushings in 90% of cases. This is particularly important in pediatric patients.

SPN 16

Paediatric SARS-COV2 infections in Switzerland

Uka A^{1,2}, Zimmermann P^{1,2,3}, Ritz N^{4,5}

¹Faculty of Science and Medicine, University of Fribourg, Fribourg, Switzerland; ²Department of Paediatrics, Fribourg Hospital HFR, Fribourg, Switzerland; ³Infectious Diseases Research Group, Murdoch Children's Research Institute, Parkville, Australia; ⁴Paediatric Infectious Diseases and Vaccinology, University of Basel Children's Hospital Basel, Switzerland; ⁵Department of Paediatrics, The Royal Children's Hospital Melbourne, The University of Melbourne, Australia

Background

Coronavirus disease 2019 (COVID-19) is usually less severe in children compared to adults, with mostly mild or asymptomatic courses and a low case-fatality rate. This study describes detailed clinical characteristics, treatment and outcomes of children with laboratory confirmed COVID-19 in a non-hospitalised and hospitalised setting and quantifies factors associated with admission to hospital and intensive care unit in children and adolescents with SARS-CoV-2 infection on a nationwide level.

Methods

Data were collected through the Swiss Paediatric Surveillance Unit (SPSU) from children < 18 years with laboratory-confirmed SARS-CoV-2 infection. All 33 paediatric hospitals in Switzerland reported non-hospitalised and hospitalised cases from March 1 to October 31, 2020 during both pandemic peaks.

Findings

In total, 678 children and adolescents were included. The median age was 12.2 (IQR 5.0 – 14.6) years, 316 (47%) were female and 106 (16%) had comorbidities. Comorbidities were the only factor associated with hospital admission in a multivariable regression analysis (odds ratio 3.23, 95%CI 1.89 to 5.50; p-value <0.01). Hospitalised children more often presented with fever [96 [76%] vs 209 [38%], p-value<0.01] and rash [16 [1%] vs 6 [1%], p-value<0.01]. Anosmia/dysgeusia was more prevalent in non-hospitalised children (73 [13.3%] vs 3 [2.4%], p-value<0.01). In the hospitalised children, oxygen treatment was required in 34 (27.0%), intensive care admission in 16 (12.7%) inotropes in 9 (7.3%) and mechanical ventilation in 8 (6.3%). Complications were reported in 28 (22.2%) children with cardiovascular complications being most frequent (11 [1.6%]). Three deaths were recorded.

Interpretation

This study confirms that COVID-19 is mostly a mild disease in children and adolescents with low mortality. Fever, rash, and comorbidities are associated with higher admission rates. Continuous observation is necessary to further understand paediatric COVID-19, guide therapy and evaluate the necessity for vaccination in children.

SPN 17

Cardiovascular magnetic resonance predictive of outcomes in paediatric pulmonary hypertension

Capello Mainardi C^{1,2}, Muthurangu V², Ferrucci E¹

¹Department of Paediatrics, Paediatric Institute of Italian Switzerland (IPSI), Ospedale Civico di Lugano, EOC, Lugano, Switzerland; ²Department of Paediatric Cardiology, Unit of cardiac MRI, Great Ormond Street Hospital, London, United Kingdom

In the era of goal-driven therapies, the ability of predicting prognosis is essential in the management of patients with pulmonary hypertension (PH). Since paediatric PH differs from the adult form, what applies to adults does not necessarily translate to children as well and unfortunately, there are no sufficient studies conducted on paediatric populations. In PH, cardiac magnetic resonance (CMR) is the most accurate way of assessing right ventricle (RV) function and pulmonary haemodynamics non-invasively. Given that prognosis is heavily influenced by RV function, CMR may also prove useful in providing prognostic

information. While the prognostic role of CMR is well-established in adults, to date there is only one study demonstrating this correlation in children. The aim of this study was to investigate if other CMR-derived variables correlate with survival in paediatric PH.

The study was conducted at Great Ormond Street Hospital in London. The study population included 223 patients with PH, with a median age of 8.9 years. All images were acquired by real-time radial k-t SENSE CMR, a dynamic imaging technique that allows to scan in shorter times and without breath holding, thus avoiding the need for general anaesthesia. For each patient, measures of RV and left ventricle (LV) function were assessed, and among all CMR parameters, those that correlated the most with survival on univariate analysis were right ventricular ejection fraction (RVEF) and septal curvature ratio (SCR), a measure of RV bulging into the LV. This was also reflected in quartile-based Kaplan-Meier survival curves: being in the lowest quartile for SCR and RVEF was correlated to a poorer prognosis.

While previous literature already proved the prognostic significance of RVEF in children, this study demonstrates for the first time that SCR correlates with survival. These new results could be the basis for finding new treatment goals specific for children and new clinical endpoints for paediatric clinical trials, with the final aim of optimizing treatment strategies and offering a better chance of survival. Moreover, the use of real-time techniques makes CMR performable in children as well, since it does not need sedation and controlled ventilation. Hence, the implementation of this technique could make CMR a widespread and convenient auxiliary diagnostic tool in paediatric PH, thus allowing to rely less on invasive procedures to obtain prognostic information.

SPN 18

Epidemiology of COVID-19 in Swiss PEDIatric Populations and Their Families (ECOPED): Quality of Life in the Diabetes Cohort

Vural S¹, Kahlert CR², Roduit C³, Lauener R⁴, Allemann D¹

¹Department of Endocrinology / Diabetology, Children's Hospital of Eastern Switzerland, St. Gallen, Switzerland; ²Infectious Diseases and Hospital Epidemiology, Children's Hospital of Eastern Switzerland, St. Gallen, Switzerland; ³Department of Allergology, Children's Hospital of Eastern Switzerland, St. Gallen, Switzerland; ⁴Department of Pediatrics, Children's Hospital of Eastern Switzerland, St. Gallen, Switzerland

Introduction

During the current SARS-CoV-2 pandemic, diabetes is associated with increased morbidity and mortality with COVID-19 in adults. Less is known about children with diabetes including the impact of social restrictions and anxiety about COVID-19 on the diabetes management and their mental health. The present study aims to examine, whether children with diabetes mellitus (DM) have a poorer quality of life as compared to a Swiss pediatric reference population because they suffer from social isolation or classify themselves as a risk group for COVID-19 and might be concerned about their health.

Methods

This monocentric study started in April 2020, with 2 longitudinal cohorts including children from the Childhood Allergy, Nutrition and Environment (CARE) birth cohort study and the outpatient diabetes clinic, and of these, the results are presented here. Informed consent was obtained from children and/or parents. In addition to the assessment of SARS-CoV2 antibodies, metabolic parameters, physical activity and mental health of parents, we focus here on the health-related quality of life (HrQoL) which was examined longitudinally using the Kidscreen-10 Questionnaire (Self-reported by children > 8 years or by parents for children < 8 years) submitted at baseline, monthly during 16 weeks follow up and at 10 months' final visit. The mean of the normal Swiss Kidscreen reference population before COVID-19 is a T of 50, values > 50 represent a better and values < 50 a poorer quality of life.

Results

In the descriptive interim analyses until February 2021, HrQoL was assessed in 56 children with Type1 DM and two with MODY 2; 88% of them had completed the 4-month examination (Baseline characteristics, medians and ranges: 53.4% male, 46.6% female; 12 years, 1 to 19; HbA1c 7.3%, 5.6 to 12.5%). The patients showed decreased HrQoL scores in months with high restrictions, as in May 2020 (median 46.7, min 32.2 - max 65.2), and from Nov 2020 on (46.7, 36.4 - 77.1), or after back to school in September (46.7, 31.3 - 83.5), but otherwise normal T-values even above 50 during holiday seasons June to Aug 20 (50.7, 32.4 - 83.6)

Conclusions

In the diabetes cohort we found a fairly decent metabolic management and a normal HrQoL, but there was a tendency towards lower scores than in the KID-screen reference population during the months with higher restrictions through COVID-19.

POSTERS

P 1

Polyhydramnios: Several causes – broad phenotypeStelling L¹, Hodel M², Kretschmar O³, Neuhaus T¹, Luhmann-Lunt C⁴¹Department of Pediatrics, Children's Hospital Lucerne, Switzerland; ²Obstetric Clinic, Lucerne Cantonal Hospital, Switzerland; ³Department of Pediatric Cardiology, University Children's Hospital Zurich, Switzerland; ⁴Pediatric and Neonatal Care Unit, Children's Hospital Lucerne, Switzerland**Background**

The prevalence of polyhydramnios (PH) is 1-2%; severe cases requiring amnioreduction are rare. PH is associated with fetal and perinatal morbidity. There are several conditions with PH: multiple gestations, fetal anemia, gestational diabetes, fetal infections (TORCH), anatomical or chromosomal/genetic anomalies (e.g. RASopathies, VACTERL, Bartter).

Methods

Review of our local severe cases between 2015 and 2019.

Results

Case 1: Preterm girl (35 6/7 weeks gestation, birth weight 2685g) with PH requiring repeated amnioreduction. Genetic analysis confirmed Bartter type 2. Postnatal course included polyuria with metabolic alkalosis, hyponatraemia and transient hyperkalemia followed by hypokalemia. Renal ultrasound showed medullary nephrocalcinosis. The girl's development and growth is normal on indomethacin and sodium supplement. Cases 2 and 3: Girl (38 5/7 weeks, 2950g) and her brother (38 2/7 weeks, 3500g) had PH. The postnatal course was uneventful with normal clinical examination, laboratory findings and renal ultrasound leading to final diagnosis of familial idiopathic PH. Case 4: Girl (37 5/7 weeks, 2380g) has prenatally been diagnosed with atrioventricular septal defect and PH. Noonan syndrome was confirmed postnatally based on family history, typical facial features and genetic testing. The patient died at 5 months secondary to heart failure. Case 5: Preterm girl (33 5/7 weeks, 2140g) presented with severe PH without other anomalies suggestive of Bartter's syndrome requiring amnioreduction. The postnatal course was complicated by hyponatraemia and polyuria, but also severe hypertension (130 mm Hg). Ultrasound on day 2 showed apparently 2 normal kidneys; repeated ultrasound on day 5 revealed impaired arterial flow of the left kidney and dilated infrarenal aorta. MRI and angiography confirmed unilateral left renal artery stenosis and mid-aortic syndrome. Subsequent balloon dilatation of the renal artery and aorta resulted in normalization of urine volume and electrolytes.

Conclusion

Polyhydramnios can be categorised into inherited, idiopathic or acquired cases. The prenatal course does often not predict a specific diagnosis, making counseling of parents difficult. Final diagnosis is based on postnatal course, clinical and laboratory findings and genetic analysis.

P 2

EBV-related dacryocystitis: diagnostic challenges and therapeutic pitfallsSternberg J¹, Lambiel S², Cao Van H², Massa H³, Landis BN²¹Department of Pediatrics, University Hospital Geneva, Switzerland; ²Department of Clinical Neurosciences, Oto-rhino-laryngology and cervico-facial surgery Unit, University Hospital Geneva, Switzerland; ³Department of Clinical Neurosciences, Ophthalmology Unit, University Hospital Geneva, Switzerland**Objective**

Acute dacryocystitis is an atypical and rare manifestation of pediatric mononucleosis. Still unknown by many clinicians, we want to present a case to highlight the challenges and adequate treatments.

Case report

A six year old girl with no ocular history was admitted to our hospital for intravenous antibiotic therapy of a right pre-septal cellulitis. During hospitalization, she developed a fluctuating bump in the nasolacrimal region which clinically and radiologically looked like an abscess. Serology was positive for mononu-

cleosis and we concluded to an acute EBV-related dacryocystitis. After multi-disciplinary discussion, she was treated conservatively with digital lacrimal sac massages and intra-venous antibiotic therapy with an excellent result.

Discussion: Precise diagnose was difficult because initial symptoms were unspecific (rhinitis, fever, edematous and sensible red eye lids, no purulent discharge and moderate bilateral cervical lymphadenopathy) and this presentation of EBV very uncommon and thus barely known. Nevertheless, making the difference between dacryocystitis and abscess is crucial to choose the appropriate treatment and avoid unnecessary and even potentially deleterious surgery. Conservative management of dacryocystitis is the gold-standard according to the scarce literature.

Conclusion

Acute dacryocystitis in children free of ocular history should arise the suspicion of EBV primo-infection. With a conservative treatment, prognosis seems excellent and therefore surgery should be avoided as much as possible.

P 3

A not so insipid start in life ...Dubail M¹, Sternberg J¹, De Mul A², Parvex P², Stollar F¹¹Department of Pediatrics, Geneva University Hospital, Switzerland; ²Department of Pediatric Nephrology, Geneva University Hospital, Switzerland**Introduction**

Hereditary nephrogenic diabetes insipidus is a rare recessively transmitted disease characterized by the inability to concentrate urine. Polyuria and polydipsia are cardinal symptoms but their recognition in an unmonitored setting represents a challenge for parents and physicians. Furthermore, other symptoms are non-specific. Diagnosis is often delayed and made at around 9 months. A prompt recognition and treatment are primordial because of the risk of cerebral damage due to recurrent and severe hypernatremic dehydration episodes.

Case presentation

We report the case of a four months old female infant, born appropriate for gestational age, from a consanguineous union. She was admitted at Geneva Children's Hospital due to failure to thrive since two months of age. She also presented recurrent vomiting, significant thirst, constipation and daily episodes of fever of unknown origin for the last two weeks. Psychomotor development was within normal limits. She was breastfed since birth and formula was added two weeks before admission. All growth parameters were under third percentile and physical examination showed dehydration signs.

After several blood and imaging tests excluding infectious, gastro-intestinal and neurological causes, an inability to concentrate urine was suspected because of hypernatremia, hyperosmolality, along with low urine density and osmolality. High plasmatic Copeptin levels allowed differentiation between a nephrogenic and central cause. Diagnosis was confirmed with genetic testing showing homozygous mutations pAla147Thr of aquaporine-2 channel. Free water intake (to compensate polyuria) and low osmolar diet (low sodium and normal to low protein) were introduced, enabling rapid improvement in vomiting and laboratory findings and an adequate growth. Diuretics and antiemetics were also introduced with a careful follow-up by nephrologists.

Conclusion

Nephrogenic diabetes insipidus should be considered when an infant presents with hypernatremic dehydration. Clinically, failure to thrive, vomiting, recurrent episodes of fever and apparent thirst should arise suspicion. Prompt recognition, diagnosis and treatment are essential for a proper growth and development, and to prevent life threatening consequences.

P 4

A forgotten disease presenting with bilateral leg weakness, petechiae and spontaneous gingival bleeding in a 3-year-old toddlerFelsler A¹, Agyeman P¹, Horn M¹, Stranzinger E², Kopp MV¹, Moser B¹¹Department of Paediatrics, Inselspital, University Hospital Berne, Switzerland; ²Department of Diagnostic, Interventional and Paediatric Radiology, Inselspital, University Hospital Berne, Switzerland**Introduction**

A forgotten disease presenting with pseudoparalysis, petechiae and gum disease.

Case presentation

A 3-year-old previously healthy Caucasian boy presented with a brief history of non-weight bearing to our Emergency Department. Clinical examination revealed a pseudoparalysis and multiple small petechiae on the legs, swollen gums with spontaneous bleeding and no fever. Hips and knees were semi-flexed with painful full range of movement and swollen knees. His work up showed: CRP 13 mg/L, WBC 10.7 G/L, Ery 4.78 T/L, Hb 113 g/L, Plt 299 G/L, ESR 23 mm/h, INR 1.06, aPTT 29.7 sec. Magnetic resonance imaging (MRI) of the spine and pelvis revealed multiple contrast-enhancing osseous lesions in the spine, and T2 hyperintense lesions of the distal and proximal femoral metaphysis, the right acetabulum and both os ischii. Dietary history revealed a fussy eater with a restricted diet based entirely on cow's milk with dissolved cookies. The clinical findings combined with the restricted diet and MRI patterns led to the suspicion of vitamin C deficiency. He showed minimal response to analgesia but good response to empirical treatment with 200 mg oral vitamin C daily, with all clinical symptoms resolved within one week. For his feeding difficulties he was enrolled in a feeding program. His initially taken vitamin C level returned undetectable (<5 µmol/l, normal range 26-97 µmol/l) confirming the clinically suspected diagnosis of scurvy.

Conclusion

Scurvy is best known as a historical disease of ancient sailors in long distance sea travel, caused by nutritional deficiency of vitamin C. Vitamin C is an essential nutrient acting as an antioxidant and important cofactor enabling tissue growth and repair, as well as maintaining function of certain neurotransmitters and proper immune function. The clinical spectrum varies with nonspecific initial symptoms like irritability, loss of appetite or low-grade fever. Later signs are symmetrical myalgia, arthralgia, joint and limb swelling, skin changes with easy bruising, perifollicular petechiae and poor wound healing but also gum disease with bleeding, loosening of teeth and finally death. Although a rare disease and more commonly reported from developing countries with prevalent malnutrition, this case report highlights the importance of a thorough clinical and dietary history sparing the patient unnecessary investigations and treatments. Supplementation with vitamin C usually leads to a swift recovery.

P 5

Low hematocrit leading to tacrolimus toxicityPiletta-Zanin A^{1,2}, De Mul A², Rock N², Lescuyer P³, Samer CF^{1,4}, Rodieux F¹¹Division of Clinical Pharmacology and Toxicology, Department of Anesthesiology, Pharmacology, Intensive care and Emergency medicine, Geneva University Hospitals, Switzerland; ²Division of Pediatric Specialties, Department of Women, Children and Adolescents, Geneva University Hospitals, Switzerland; ³Division of Laboratory Medicine, Department of Diagnostic, Geneva University Hospitals, Switzerland; ⁴Institute of Pharmaceutical Sciences of Western Switzerland (ISPSO), University of Geneva, Geneva, Switzerland

Tacrolimus is a calcineurin inhibitor and one of the most frequently prescribed immunosuppressant after solid organ transplant. Tacrolimus is characterized by a narrow therapeutic index and high intra- and inter-individual pharmacokinetic variability. In order to maximize the effectiveness and minimize the toxicity, Therapeutic drug monitoring in whole-blood is the standard procedure. However tacrolimus extensively binds to erythrocytes, and changes in hematocrit affect tacrolimus whole-blood distribution and thus influence unbound (therapeutically active) tacrolimus concentration. Hematocrit is therefore a key factor in the interpretation of tacrolimus whole blood concentrations.

We report a case of a 16-year-old girl with liver and kidney transplantation in whom of low hematocrit has led to a misinterpretation of infratherapeutic tacrolimus concentration leading to a significant increase in tacrolimus dosing and toxicity.

P 6

Recognizing the Presentation and Typical Complications of Congenital Portosystemic Shunts in Children in General Practice: a case studyBahadori Atesa¹, Kuhlmann Beatrice², Debray Dominique³, Franchi-Abella Stephanie⁴, Wacker Bou Puigdefabregas Julie¹, Beghetti Maurice¹, Wildhaber Barbara⁵, McLin Valérie¹¹Pediatric Specialties Division, Geneva University Hospitals (HUG), Geneva, Switzerland; ²Pediatric Endocrinology, Cantonal Hospital Aarau, Aarau, Switzerland; ³Pediatric Gastroenterology, Hepatology & Nutrition, Necker Hospital, Paris, France; ⁴Pediatric Radiology, Bicêtre Hospital, Paris, France; ⁵Child and Adolescent Surgery Division, Geneva University Hospitals (HUG), Geneva, Switzerland**Background**

Congenital porto-systemic shunts (CPSS) are rare anatomical vascular anomalies resulting in communications between the portal venous system and the systemic circulation, affecting an estimated 30'000 to 50'000 children. CPSS can present at any age as multi-systemic disease of variable severity mimicking common pediatric conditions, including tall stature.

Case presentation

A nine year old girl was addressed to an endocrinologist for tall stature. Initial work up shows insulin resistance with hyperinsulinism, acanthosis nigricans and hyperandrogenemia without menarche. The following were also identified over the course of the work-up: intermittent hyperammonemia, cognitive impairment with hyperintense globus pallidus on T1 weighted cerebral Magnetic Resonance Imaging, intermittent macrohematuria and a vascular malformation was suspected on abdominal ultrasound. Cardiac echocardiogram was suggestive of elevated right sided pressures. She was referred to a specialized liver center at eleven years of age for suspicion of pulmonary hypertension.

An angio-CT scan confirmed the presence of a CPSS between the left portal vein and the left hepatic vein, with two nodules in segments VII and VIII of the liver. Angiography with balloon occlusion confirmed the presence of a single shunt with a normal right hepatic portal venogram. Right heart catheterization confirmed moderate pulmonary hypertension (mean pulmonary arterial pressure: 40 mmHg; pulmonary vascular resistance index: 7 WUm²). Liver nodule histology revealed hepatic adenomas with nuclear translocation of beta-catenin, a mutation at risk of malignant transformation.

Management and outcome

Pulmonary hypertension was treated using combined therapy by an endothelin receptor antagonist and a phosphodiesterase type 5. Next, the CPSS was successfully closed using an endovascular Amplatzer plug. Finally, liver nodules were surgically resected after several months of normal portal flow.

Conclusion

This case illustrates most of the common chief complaints and complications of CPSS. Abdominal Doppler ultrasound is the key to diagnosis. Considering porto-systemic shunts in the diagnostic work-up of a patient with unexplained endocrine, liver, gastro-intestinal, cardiovascular, hematological, renal or neurocognitive disorder is important as prompt referral to a specialized center may significantly impact patient outcome.

P 7

Benign COVID19 in a highly vulnerable adolescent with type 1 diabetes and leukemiaSarkisian J¹, Klee P^{1,2}, Dirlwanger M^{1,2}, Bernard F^{3,4}, Baleyrier F^{3,4}, Ansari M^{3,4}, Schwitzgebel V^{1,2}¹Pediatric Endocrine and Diabetes Unit, Division of growth and development, Department of pediatrics, gynecology and obstetrics, University Hospitals of Geneva, 1211 Geneva, Switzerland; ²Diabetes Center, Faculty of Medicine, University of Geneva, 1211 Geneva, Switzerland; ³Oncology and Hematology Unit, Department of pediatrics, gynecology and obstetrics, University Hospitals of Geneva, 1211 Geneva, Switzerland; ⁴CANSEARCH research platform in pediatric oncology and haematology, Department of pediatrics, gynecology and obstetrics, University of Geneva, Switzerland**Background**

SARS-COV-2 infection is currently a major health concern. Several studies have sought to determine the risk factors related to the mortality and severity of this infection. Diabetes and hematologic cancers are among these risk factors in the adult population, but it is not clear if these risk factors apply to the pediatric population.

Methods

SARS-COV-2 detection was done with COBAS® 6800 system (Roche).

Clinical presentation

This is a 17-year-old adolescent with type 1 diabetes diagnosed 7 years ago. In January 2020 a Philadelphia chromosome-positive acute lymphoblastic leukemia was detected. In July 2020 the patient needed an haplo-identical allogeneic stem cells transplantation and is currently under immunosuppressive therapy (tacrolimus). On 11/11/2020, the patient consulted for asthenia associated with inappetence, episodes of vomiting and diarrhea, without fever. He tested positive for SARS-CoV-2 by PCR (CT 26.7 (day of infection (DI) 1). The patient was hospitalized during 6 days for observation and symptomatic treatment. After discharge, fluctuating asthenia persisted associated with rhinorrhea and a cough. Two weeks later, he consulted again with fever at 39.5°C. The SARS-CoV-2 PCR test was repeated and still positive (CT 19.4, DI 22). Additional investigations revealed a Legionella infection with positive PCR in a bronchoalveolar lavage sample and positive urinary antigen. The clinical and biological course was favorable under antibiotic therapy, without the need for respiratory or hemodynamic support. Several PCR tests for SARS-CoV-2 were subsequently carried out to monitor the kinetics of the infection (CT 26,5 on DI 29 – CT 25,9 on DI 39 – CT 24,6 on DI 54). It came back negative on DI 84.

Conclusions

This case illustrates a surprisingly benign course of COVID-19 for an adolescent with a high risk profile, having type 1 diabetes, leukemia and a co-infection with Legionella. Clearance of SARS-COV-2 took almost three months. The age-dependent severity of COVID-19 has already been demonstrated, however not in the presence of a hematologic malignancy combined with diabetes. Further studies are necessary to dissect the mechanisms predisposing to severe infection in the pediatric population and the role of the immunosuppression on the severity of COVID-19.

P 8**Early identification of children with developmental delay. Primary health care study in the canton of Zurich.**

Moser Martin¹, Mu?llner Corina¹, Ferro Patricia¹, Jenni Oskar^{1,2}, von Rhein Michael^{1,2}

¹University Children's Hospital Zurich, Child Development Center; ²Children's Research Center, Zurich, Switzerland

Background

Early identification of children with developmental delay (DD) is one of the main goals in well-child visits. In the canton of Zurich, most children between 0-4 years are referred for early intervention (EI) by pediatricians and GPs. However, rates of EI-utilization are below expectations. We therefore sought to investigate the perspective of primary care providers on early identification, ways to access, and their overall satisfaction with the system in the Canton of Zurich.

Methods

Our online survey included 59.2% of all pediatricians and 11.3% of all GPs in the Canton of Zurich.

Results

The vast majority of children (95.7%) with DD is being referred to EI by pediatricians. GPs are underrepresented given that they account for 11.7% of all preschool consultations. Self-confidence levels with regard to identifying DD differs between the two groups of primary care providers with better self-ratings of pediatricians. In the subgroup of pediatricians, the self-confidence level is associated with the number of well-child visits per week and the years of experience in private praxis. The system of care is well appreciated, but a shortage of resources was criticized by study participants.

Conclusions

Primary care providers have a central role in identifying children with DD and referring them to early intervention. However, ongoing education is essential to improve the early identification of DD, especially for new and foreign primary care providers. Pediatric health care research - ideally in cooperation with primary care providers - is crucial to analyze and optimize the pediatric health care in Switzerland.

P 9**Headache and meningism without a cough – Tuberculosis?**

Peter AM¹, Simma L², Buettcher M³

¹Department of Pediatrics, Lucerne Children's Hospital; ²Emergency Department, Lucerne Children's Hospital; ³Department of Paediatric Infectious Diseases, Lucerne Children's Hospital

Introduction

Central nervous system tuberculosis is still a common problem in endemic countries but rarely seen in Switzerland. The clinical manifestation may be nonspecific and the diagnosis challenging.

Case presentation

A 13-year-old adolescent presented with fever, headache and lumbar pain over four days. She had severe meningism but normal mentation. Pleocytosis (cell count (CC) 821/ul, 50% polynuclear (PN), 50% mononuclear (MN)), hypoglycorrhachia (CSF / serum glucose ratio: 0.15), elevated levels of albumin (525mg/l) and lactate (7.1mmol/l) were found. She had been evaluated for possible mycobacterium tuberculosis (MTB) infection post exposure to an index patient (IP) (negative interferon gamma release assay (IGRA)). The last contact with the IP was two months before. Our patient did not complain of any cough or b-symptoms. She was started on ceftriaxone to cover a bacterial meningitis, but the clinical picture was atypical. After three days without clinical improvement and negative blood and CSF cultures (incl. PCR) a second CSF sample was obtained (CC 482/ul: 28% PN, 72% MN, Glucose 1mmol/l, albumin 657mg/l, lactate 9.8mmol/l) and MTB investigations were initiated. Gene Xpert real time PCR Ultra (and later culture) were positive for MTB (Rifampicin rpoB-Gene wildtype) on the first and second CSF sample. Microscopy was negative. MRI of brain and spine revealed no signs of inflammation. Surprisingly the MRI revealed bilateral apical consolidations suggestive for concomitant pulmonary tuberculosis. IGRA and tuberculin skin tests were negative. PCR and culture of induced sputum sampling were positive, microscopy negative. Treatment was started with rifampicin, isoniazid, pyrazinamide, moxifloxacin and dexamethasone. Neurological symptoms improved rapidly after 24-48 hours.

Conclusion

Hypoglycorrhachia may be a sign for acute bacterial meningitis, which is far more common in MTB non-endemic countries. However the combination of hypoglycorrhachia and a predominant MN pleocytosis is an important sign suggesting MTB. Concomitant clinical pulmonary signs and symptoms are not obligatory. For diagnosis one should never rely on immunological MTB investigations alone as they can be negative in tuberculosis disease. Gene Xpert MTB Ultra PCR is a very sensitive and fast test particularly for the examination of CSF in children. Early diagnosis and rapid initiation of treatment is the most important factor for a positive outcome.

P 10**Abdominal tuberculosis.About three pediatric cases.**

Ait idir k¹, Benaïed MA², Tibouk A³

¹Service de pédiatrie. Centre de Consultations Spécialisées de l'Armée. Hussein Dey. Algiers. Algeria.; ²Service de chirurgie infantile. Hôpital Central de l'Armée. Algiers. Algeria.; ³Service d'anatomie pathologique. Hôpital Central de l'Armée. Algiers. Algeria.

Introduction

abdominal tuberculosis (TB) is uncommon in children. It can affect the abdominal cavity and the digestive tract from the oesophagus until the anus. It is characterized by unspecific symptoms. Microbiology, endoscopy and histopathology are the foundations of the exploration, although sometimes they may not contribute to an accurate diagnosis.

Material and methods

the study was retrospective and descriptive of a mini-series of three pediatric cases enrolled from 2015 to 2020. The diagnosis was

established according to the World Health Organization diagnostic criteria of tuberculosis.

Results

three girls aged 3,4 and 7 years. All of them with a history of BCG vaccination. Only one patient had contact with a tuberculosis case. Clinical features were: abdominal pain (3/3), fever (2/3), weight loss (2/3), acute appendicitis (1/3). Regarding location, abdominal TB involved peritoneum (3times), abdominal lymph nodes (2times), intestine (2times) with fistula (1 time), liver and spleen (1time).The histopathological study was conclusive in 2/3 of the cases. The evolution was favourable under TB chemotherapy introduced following the Algerian TB program.

Conclusion

abdominal TB is a rare and insidious infection .Children appear to have a higher risk of having extrapulmonary TB affecting any organ. In TB endemic country, as Algeria, abdominal TB must be considered in front of patients with nonspecific features. In the absence of histological evidence, a set of arguments (clinical, biologicals, radiologicals., endoscopicals) is required to confirm the diagnosis

P 11

Colorectal polyps in children. About a serie of 36 cases

Ait idir K¹, Tibouk A², Benaired MA³

¹Service de pédiatrie. Centre de Consultations Spécialisées de l'Armée. Hussein Dey. Algiers. Algeria.; ²Service de chirurgie infantile. Hôpital Central de l'Armée. Algiers. Algeria.; ³Service d'anatomie pathologique. Hôpital Central de l'Armée. Algiers. Algeria.

Introduction

Colorectal polyps are not rare in children. The objective of this study was to describe the demographic, clinical, endoscopic and histological features of children with colorectal polyps.

Material and Methods

The study was retrospective and descriptive of children with colorectal polyps enrolled from 2013 to 2020 and managed in centre de consultations spécialisées de l'armée Hussein Dey in Algiers

Results

36 children were registered. 24 boys and 12 girls. The mean age at presentation was 9 ± 5.8 years [2-15]. Concerning familial history, we founded colorectal cancer (1 case), familial adenomatous polyposis (1 case). About personal history, our objective diabete milletus (1 case), Down syndrome (1case), focal nodular hyperplasia (1 case). Hematochezia was the principal symptom (98%), followed by abdominal pain (86%), chronic diarrhea (36%),lost weight (22%),prolapse of the polyp from the anus (18%) and intestinal intussusception (5,4%). Severe anemia was noted in two patients who need blood transfusion, iron deficiency anemia (66%), inflammatory syndrome (12%). Endoscopy coupled to biopsy were performed. The most frequent polyp location where the rectosigmoid colon (76%), descending colon (34%), transverse colon (20%), ascending colon (15%), duodenum (8,3%) and solitary polyps (2,7%). The histopathological examination founded: 25 (70%) typical juvenile polyps, 3 (8,3%) with adenomatous polyps, 2 (5,5%) with inflammatory polyps, 2 (5,5%) with inflammatory polyps, 1 (2,7%) with PeutzJeghers syndrome, 1 (2.7%) patient familial adenomatous polyposis, 1 (2.7%) colon cancer, 1 (2.7%) rectal neuroendocrine tumor.

Conclusion

Colorectal polyps are the most common tumors of the colon and rectum in children. The symptomatology is nonspecific. Both of endoscopy and histology are obligatory for the diagnosis. The origin of polyps is generally benign but can be malignant, fortunately it's exceptional.

P 12

HPV vaccine awareness, information sources, and hesitancy among young men and women in Switzerland

Schwendener CL^{*1,2}, Kiener LM^{*1,2}, Jafflin K^{2,3}, Schärli S⁴, Muggli F⁵, Gültekin N⁶, Huber BM⁷, Merten S^{2,3}, Deml MJ^{2,3}, Tarr PE^{1,2}

¹University Department of Medicine, Kantonsspital Baselland, University of Basel, 4101 Bruderholz, Switzerland; ²University of Basel, Petersplatz 1, 4051 Basel, Switzerland; ³Swiss Tropical and Public Health Institute, Socinstr. 57, 4051 Basel, Switzerland; ⁴Rekrutierungszentrum Aarau, Schweizer Armee, Aarau, Switzerland; ⁵Rekrutierungszentrum Monte Ceneri, Schweizer Armee, Monte Ceneri, Switzerland; ⁶Kompetenzzentrum für Militär- und Katastrophenmedizin, Eidgenössisches Departement für Verteidigung, Bevölkerungsschutz und Sport VBS Schweizer Armee, Ittigen, Switzerland; ⁷Department of Pediatrics, HFR Fribourg Cantonal Hospital, Fribourg, Switzerland; ⁸*equal contribution

Background

HPV vaccine has been recommended in Switzerland for young women and young men since 2007 and 2015, respectively. Identifying factors associated with HPV vaccine uptake, including vaccine hesitancy (VH), is essential for designing successful vaccination programs. We examined the association between awareness, knowledge and VH with HPV vaccine uptake in Swiss males and females, 15-26 years of age.

Methods

We recruited participants in the offices of physicians providing biomedicine and complementary/alternative medicine (CAM), during military enlistment, and in a sexual health clinic. We conducted interviews (25-35 minutes) and gathered information on socio-demographics, HPV vaccine awareness, knowledge, information sources and HPV vaccination status. We measured VH with a modified Parent Attitudes about Childhood Vaccinations (PACV) survey that has been validated for use in German, French, and Italian. We used univariate and multi-level logistic regression analysis to assess determinants of HPV vaccine uptake.

Results

Of 994 participants, 674 participants provided a vaccination certificate (415 males, 259 females). 151 (58%) female and 64 (15%) male participants had received ≥1 dose of HPV vaccine. 694 (42%) participants were aware/knowledgeable about the HPV vaccine. Females were more likely to be aware/knowledgeable about HPV vaccination than males (339/409 [83%] vs. 356/585 [61%]; p<0.01). Younger (born after 1.7.02) compared to older (born before 1.7.02) participants were more likely to be aware about HPV vaccine (103/149 [69%] vs. 357/845 [42%]; p<0.01). The 3 most frequently mentioned information sources were school health programs, health care providers, and participants' social networks. 81 (31%) female and 92 (22%) male participants were VH (PACV score ≥50). The odds of being unvaccinated were higher among VH females than non-VH females, odds ratio (OR) = 4.90 (95% confidence interval [CI], 2.53-9.50), but similar among VH and non-VH males, OR = 1.90 (95% CI, 0.84-4.31).

Conclusion

HPV vaccine awareness and knowledge were higher among females vs. males, and higher among younger vs. older participants. To our knowledge, this is the first assessment of adolescents' hesitancy towards HPV vaccine using the PACV survey. VH is associated with lower HPV vaccine uptake in young Swiss women but not young Swiss men whose low immunization rates may be related to awareness and access issues.

P 13

Acute onset of a drug induced extrapyramidal syndrome in a 14 years old healthy girl.

Sebti Moustapha^{1,2}, Ritter-schenk Céline¹

¹1. Department of pediatrics, hôpital fribourgeois, Switzerland; ²2. Department of endocrinology, metabolic science and cardiovascular system, University of fribourg, Switzerland

A 14 year old girl, with no past medical history, was brought to the emergency department for a psychological state alteration that began abruptly 6 hours earlier with inadequate gaze and expressive dysphasia. Subsequently, she developed abnormal positioning of the head, dyskinesia, resting tremor and hypersalivation. The patient was well oriented and had a GCS of 15. In the anamnesis, it appears that the patient was put on a weight-appropriate dose of amoxicillin-clavulanate 3 days earlier by her attending physician for a bacterial tonsillitis (2 days history of fever and dysphagia, positive pharyngeal streptest).

Upon admission in the ED, assessment of airway, breathing and circulation were unremarkable. After exclusion of life-threatening etiologies, especially intracerebral complications of the tonsillitis, a re-assessment of patient's recent history was conducted, and the possibility of an intoxication was evoked. The mother then reported that the child was put on supratherapeutic doses of metoclopramide (10 mg, 3 x/day, body weight: 42 kg) 72 hours earlier for vomiting and nausea, which she took systematically. This information led us to a drug induced extrapyramidal syndrome and an IV anticholinergic therapy was started (biperiden, 2.5 mg). A few minutes after the initiation of treatment, a complete resolution of neurological symptoms was achieved. Metoclopramide treatment was interrupted. The patient reported blurred vision during a few minutes following anticholinergic therapy. This symptom spontaneously disappeared within a few hours. She remained stable during a 12 hours neurological surveillance and was discharged the next day.

P 14

Assessing cough in epidemiological studies: does it matter which question is used?

Mallet MC¹, Mozun R¹, Ardura-Garcia C¹, Latzin P², Moeller A³, Kuehni CE¹

¹Institute of Social and Preventive Medicine, University of Bern, Bern (Switzerland); ²Paediatric Respiratory Medicine, University Children's Hospital Bern, University of Bern, Bern (Switzerland); ³Division of Respiratory Medicine, University Children's Hospital Zurich and Children's Research Centre, University of Zurich, Zurich (Switzerland)

Introduction

Epidemiological studies use different questions to assess cough in children. We compared parents' answers to different questions on cough in their children and how this affected prevalence estimates.

Methods

We analysed data from Luftibus in the School, a study on respiratory health in schoolchildren, conducted in Switzerland, in 2013-16. Parents completed a questionnaire with three different questions on their child's cough used in large epidemiological studies: does your child "cough apart from colds" (1), "has dry cough at night apart from a cough associated with a cold or a chest infection" (2) and "cough more than other children" (3). We assessed how many parents answered "yes" to each question, how the answers overlapped and how this differed by age, sex and presence of wheeze.

Results

We included 3427 children (age 6-17 years, 50% girls). Among them, 8% (280) reported to have wheezed in the past 12 months. The prevalence of cough differed substantially depending on the question asked. 872 (25%, 95%CI 24-27%) parents reported cough apart from colds, 390 (11%, 95%CI 10-12%) dry night cough, and 158 (5%, 95%CI 4-5%) said that their child coughs more than other children. We found only partial overlap between answers to the different cough questions, with only 88 (3%) parents saying yes to all three questions. Overlap was broader for children who also reported wheeze, but did not vary by age or sex.

Conclusion

The questions used to assess cough in children strongly affect prevalence estimates of cough in children. We therefore need to be very careful when interpreting and comparing studies.

Funding: SNSF:320030_182628; Lunge Zürich

1. Ferris B. Epidemiology Standardization Project (American Thoracic Society). *Am Rev Respir Dis* 1978; 118: 1-120
2. Asher et al. International Study of Asthma and Allergies in Childhood (ISAAC): rationale and methods. *Eur Respir J* 1995; 8: 483-491
3. Clifford et al. *Arch Dis Child* 1989; 64: 1118-1125

P 15

Neonatal hypoglycemia: open the diapers !

Tossetti L¹, Fournier C¹, Kayemba Kay's S¹, Dalaqua M², Llor J¹, Paccaud Y¹, Hernandez E¹, Phan-Hug F³

¹Pediatric Department, Hôpital du Valais, Sion, Switzerland; ²Neuroradiology, Hôpital du Valais, Sion, Switzerland; ³Pediatric Endocrinology, Hôpital du Valais, Sion, Switzerland.

A term newborn presented with hypoglycemia (0.9mmol/l) at H8 of life. Clinical examination revealed mild dysmorphic facial features (micrognathism and low-set ears with helix malformation), micropenis and small testis. He was admitted for hypoglycemia. He received glucose 10% infusion and phototherapy for jaundice. Early enteral feeding was difficult due to fatigue. He presented recurrent hypoglycemia until day 6 with total glucose supply of 7mg/kg/min.

Blood tests during hypoglycemia showed low cortisol (2.5nmol/l) and ACTH (7ng/l). Insulin levels were normally suppressed (0.4mU/l). Abdominal ultrasound showed normal adrenal glands. Oral hydrocortison (20mg/m2) was started at day 5. Recurrent hypoglycemia, hypogonadism and hypocortisolemia strongly suggested a central hormonal deficit. GH (1.58µg/l) and free T4 levels were low without TSH elevation (10.7 pmol/l, 5.41mU/l). Cerebral MRI revealed an ectopic posterior hypophysis and no pituitary stalk, confirming the diagnosis of pituitary stalk interruption syndrome (PSIS). L-tyroxin (8mcg/kg) substitution was started the day after hydrocortison substitution and subcutaneous recombinant GH (35µg/kg/day) was added at day 8, resolving persistent hypoglycemia. Prolactin (243mcg/l) was normal and there was no sign of diabetes insipidus. Symptoms resolved after hormonal replacement therapy. Blood test at 2 months of age (minipuberty) confirmed gonadotrophin deficiency.

Discussion

PSIS is a rare cause of hypopituitarism. Diagnosis is challenging in the neonatal period, as assessment of hypothalamus-hypophysis axis differs from the other stages of life and hormonal values are difficult to interpret. Reported median age at diagnosis is 4 years. Pituitary hormone deficiency can develop over time. GH deficiency is present in all patients and contributes to neonatal hypoglycemia and micropenis. ACTH deficiency is life-threatening and must be rapidly diagnosed and treated. Posterior pituitary hormones are often unaffected and prolactin level is typically normal.

Conclusion

PSIS is a rare cause of persistent neonatal hypoglycemia. It must be suspected whenever hypoglycemia is associated with hypogonadism. The combination of clinical, hormonal and radiological findings permitted to confirm diagnosis in the first week of life. Prompt hormonal replacement therapy allowed quick resolution of symptoms and prevention of life-threatening complications.

P 16

Osteopathia Striata with Cranial Sclerosis (OSCS) can occur in males

Petter M¹, Wolfer S¹, Hirsch-Delpy J², Bottani A³, Llor J¹, Paccaud Y¹, Russo M¹

¹Service de Pédiatrie, Hôpital du Valais, CHVR, Sion, Switzerland; ²Service de radiologie, Hôpital du Valais, CHVR, Sion, Switzerland; ³Service de génétique médicale, Hôpital du Valais, ICH, Sion, Switzerland

Case report

15-year-old male who, while showering and moving his head, suddenly heard a cracking noise in his cervical bones, lost consciousness and felt. Development was normal without history of seizures.

There was no history of structural heart disease, previous syncope or family history of sudden death.

Clinical examination showed a broad and high forehead with macrocrania, posteriorly-rotated ears, mild prognathism, bilateral hallux valgus with long toes. Genitalia were normal. Head circumference was 59.5 cm (>P97), weight 52.3kg (P25-50) and height 175.5 cm (P50-75).

Head CT-scan revealed a generalized thickening and increased density of all bones. X-ray of pelvis and long bones showed radial, resp. longitudinal striations of metaphyses and diaphyses. MRI of the cervical spine in neutral, hyperflexed and hyperextended positions confirmed the thickening of skull base and of the foramina magna without functional implication on the vessels. Echodoppler examination of carotid and vertebral arteries was normal. Audiogram and ophthalmic examination were normal.

Based on radiological findings, the diagnosis of OSCS was made. Blood, skin, buccal swabs, and urine DNA analysis is pending.

Discussion

OSCS is a rare X-linked dominant sclerosing bone dysplasia caused by mutations in the AMER1 (also called WTX) gene and thus affects predominantly females. Most males with a mutation in this gene die in utero or during neonatal period and their Xrays usually do not show metaphyseal striations. Few surviving males with a milder phenotype have been reported and somatic mosaicism for a mutation in AMER1 has been proven in 4 of them. Progressive cranial sclerosis may lead to sensorineural hearing loss, ophthalmoplegia, facial palsy or medullary compression, thus prompting a multidisciplinary surveillance. Main differential diagnosis is osteopetrosis.

Conclusion

Do not discard the diagnosis of OSCS in a male with suggestive radiological findings.

References

- Holman SK et al. The male phenotype in Osteopathia Striata Congenita with Cranial Sclerosis. *Am J Med Genet* 2011 Part A 155: 2397-2408.
- Hague J et al. Male child with somatic mosaic Osteopathia Striata with Cranial Sclerosis caused by a novel pathogenic AMER1 frameshift mutation. *Am J Med Genet* 2017 173A: 1931-1935

P 17**Biotinidase deficiency in a newborn**

Papasavva D¹, Peralta M², Ballhausen D², Paccaud Y¹, Russo M¹, Hernandez E¹, Llor J¹

¹Service de Pédiatrie, Hôpital du Valais, Centre Hospitalier du Valais Romand, Sion, Switzerland; ²Unité pédiatrique des maladies métaboliques, CHUV, Lausanne

Case report

A late preterm boy presented hypotonia and tachypnea at birth, systolic cardiac murmur (2/6) and hepatomegaly. X-rays showed cardiomegaly. Blood test: lactic acidosis (10mmol/l) and hypoglycemia (0.8mmol/L). Due to respiratory distress, he was intubated.

The day after, he presented a rash and hypotension treated with fluid and amines.

Transaminases were abnormal (AST 198U/l, ALT 172U/l), lactate elevated (10mmol/L) and ammonium was normal.

Based on symptoms (hypotonia, respiratory distress, rash) and findings (lactic acidosis, hypoglycemia, cardiomegaly, hepatomegaly) a metabolic disorder was suspected.

Biotinidase deficiency (BTD) was diagnosed (enzyme activity 18%, ↑ Lactate/Pyruvate, ↑ CK, ↑ lactate. Urines: ↑ 3-hydroxyisovaleric acid, ↑ lactate). Because of the severe clinical presentation, association to a holocarboxylase or pyruvate carboxylase deficiency was suspected and skin biopsy for fibroblasts analysis performed (still pending).

With the appropriate diet (proteins, lipids) and oral supplements of biotin, riboflavin and thiamine he globally improved and was intubated after one week. One month later he was discharged with biotin, thiamine and B2 supplements. Development was favourable at three month.

Discussion

BTD is an autosomal recessive disorder (incidence of 1:60000), on chromosome 3q25. Biotin is important for fatty-acid synthesis, amino-acid catabolism and gluconeogenesis. Newborn screening has resulted in the detection of partial (10-30% of mean normal activity) and profound BTD (<10%).

Clinical manifestations of profound BTD include neurological (hypotonia, seizures, developmental delay), dermatological (rash, alopecia) and immunological abnormalities. Biological features are metabolic acidosis, high lactate and ammonium level, organic aciduria (↑3-hydroxyisovaleric acid, ↑ 3-methylcrotonylglycine) and hypoglycaemia. Enzyme assay for biotinidase reveals low activity. Newborns are rarely symptomatic and in that case, other metabolic disorders (holocarboxylase/ pyruvate carboxylase deficiency) might be associated. Treatment by oral biotin should start as soon as possible to avoid neurological damage.

Conclusion

BTD is rarely symptomatic in a newborn. It should be considered in case of neurological and respiratory symptoms with lactic acidosis. In case of early presentation, like our case, association to other metabolic disorders should be excluded. Immediate treatment (biotin max 40mg/d) assure a good prognosis.

P 18**Umbilical abnormalities : when silver is not the answer**

Hertli M¹, Ramseyer P², Llor J¹, Alexe E²

¹Service de Pédiatrie, Hôpital du Valais, Centre Hospitalier du Valais Romand, Sion, Switzerland; ²Service de Chirurgie Pédiatrique, Hôpital du Valais, Centre Hospitalier du Valais Romand, Sion, Switzerland

Case report

A 4-d.o. full term baby boy presented with umbilical erythema and yellow discharge. Good general condition, except a subfebrile state at 37.9°C the same day. He was exclusive breastfeeding with normal stools and urine.

Induced delivery for premature and prolonged rupture of membranes with excellent neonatal adaptation. He left hospital at day 3 of life with a normal clinical examination.

Upon arrival: erythematous umbilicus with visible fistula, yellow discharge increased by abdominal palpation. Normal bowel sounds, soft and painless abdomen. No mass or organomegaly. Normal blood tests.

Abdominal sonography showed a patent omphalomesenteric duct (POMD) without associated malformations (i.e. malrotation).

Surgical exploration: resection of the POMD with termino-terminal ileo-ileal anastomosis and umbilicoplasty. Parenteral nutrition for 48h in the NICU followed by complete enteral feeding. Discharged 4 days after surgery.

Discussion

OMD's malformations are rare (2%) and occur by abnormal involution between 8th-9th week of gestation. The most common is Meckel's diverticulum with its complications (bowel obstruction, GI bleeding, infection). Umbilical discharge in the neonatal period should raise the suspicion of POMD. Sometimes the presentation is atypical like an umbilical granuloma not responding to silver nitrate treatment. Abdominal echography and fistulography can help differentiate POMD from patent urachus. Treatment is surgical with POMD resection and ileo-ileal anastomosis. It prevents from complications such as intestinal prolapses, bowel obstruction, peptic ulcer with hemorrhage or omphalitis and peritonitis, with mortality rate of 10%.

Conclusion

Think about a POMD when examining an infant with umbilical discharge or persistent umbilical granuloma.

Early surgical treatment prevents potential dangerous complications.

P 19**Cannabis intoxication in a toddler**

Fracheboud N¹, Russo M¹, Donzé N², Paccaud Y¹, Hernandez E¹, Llor J¹

¹Service de Pédiatrie, Hôpital du Valais, Centre Hospitalier du Valais Romand, Sion, Switzerland; ²Service de Chimie clinique et Toxicologie, Hôpital du Valais, Institut Central des Hôpitaux, Sion, Switzerland

Case report

A previously well 17 m.o. girl was referred by ambulance for an altered level of consciousness. She was unusually sleepy since the night before and remained difficult to wake the following morning. She had no history of fever, nor infectious symptoms or head trauma.

At arrival, she was lethargic with Glasgow 11, had reactive mydriasis and global hypotonia but vital signs were normal. Clinical examination was otherwise normal.

She had normal glycemia and no inflammatory syndrome. Gas study showed a compensated hyperchloremic metabolic acidosis without lactate.

Later on, parents admitted that she accidentally ingested cannabis resin that was on the table. Rapid urinary test was positive for cannabis and blood level of THC-COOH (inactive metabolite of THC) was extremely elevated, >200 µg/l.

She was hospitalized 24h in intermediate care for monitoring and hydration. The next day, she had a normal level of consciousness and was discharged home. The case was reported to the judge and Child Protective Services.

Discussion

Cannabis is the most frequently used illegal drug in Switzerland. 33.8% of swiss population aged 15 and over has used cannabis at least once in their lifetime.

Pediatric admissions in France for children aged < 6 years increased from 1.7 to 16.1 per 100'000 admissions per year between 2004 and 2014. 71% were 18-month-old or younger. Resin stick is the most common source of intoxication and can be mistaken with chocolate bar.

The main presenting symptom is lethargy. Other frequent signs include ataxia, mydriasis, hypotonia, tachycardia and hypoventilation. The major risk is coma and respiratory insufficiency. Lethargy is a non-specific symptom that can lead to invasive investigations such as lumbar puncture and brain computed tomography. Considering cannabis intoxication can avoid further examinations.

When cannabis intoxication is suspected, start with rapid qualitative urine toxicology screen (immunochemical) and then confirm with quantitative test (GCMS) in urine or blood. The elimination half-life of THC is greater than 24 hours.

Management consists of hemodynamic and respiratory support. There is no specific antidote. Sometimes, naloxone can be used.

Conclusion

Cannabis intoxication should be considered in presence of altered mental status in afebrile children. Rapid drug screening can reduce the number of invasive tests.

The Child Protective Services should be informed.

P 20**At first glance it seems like conjunctivitis - Look beyond the red eye !**

Dosso L¹, Wolfer S¹, Gaillard MC², Stathopoulos C², Munier F², Choucair ML³, Beck-Popovic M³, Llor J¹, Boldea R⁴

¹Service de Pédiatrie, Hôpital du Valais, Centre Hospitalier du Valais Romand, Sion, Switzerland; ²Jules-Gonin Eye Hospital, University of Lausanne, Lausanne, Switzerland; ³Unité d'hémato-oncologie pédiatrique, Service de Pédiatrie, CHUV, Lausanne; ⁴Service d'ophtalmologie, Hôpital du Valais, Centre Hospitalier du Valais Romand, Sion, Switzerland

A 5-y.o. girl was referred by her pediatrician for a painful right red eye associated with photophobia, high fever and vomiting for 3 days, unresponsive to classical analgesia. Due to atypical presentation, emergency physicians requested an ophthalmic exam by a specialist. On examination, visual acuity of the right eye was light perception only. Anterior segment showed conjunctival hyperemia with clear discharge, corneal edema, mild anterior chamber inflammation, rubeosis iridis and elevated intraocular pressure (50mmHg). Right eye funduscopy showed dense vitreous cellularity suggestive of an infectious versus neoplastic disorder. Left eye was normal. Laboratory tests revealed elevated inflammatory parameters (ESR: 55 mm/h, CRP 64 mg/l) with normal leukocyte count. Toxoplasmosis, toxocarosis and herpes serologies were negative. The child was referred to the Eye Hospital Jules-Gonin, the Swiss national reference center for retinoblastoma, for further investigations. Dilated indirect ophthalmoscopy and B-scan ultrasonography revealed a retinal mass with intravitreal seeding obscuring view of the optic nerve head and highly suggestive of retinoblastoma. Brain and orbit MRI were performed and showed a possible retrolaminar invasion of the right optic nerve. Pineal gland was normal. Lumbar puncture was negative for tumor cells. Diagnosis of a unilateral retinoblastoma was made. Systemic chemotherapy (etoposide and carboplatin) and intravitreal chemotherapy was initiated.

Discussion

Retinoblastoma is the most common intraocular cancer in childhood (1:17 000 live births). It is caused by a somatic or germline mutation in the RB1 gene. Familial cases and bilateral retinoblastoma are usually detected earlier than the unilateral forms (4-12 months versus 24 months). Typical clinical signs are leukocoria and strabismus (80% of cases). Atypical presentation (20%) includes uveitis, glaucoma, hypopyon, buphthalmia, cataract or subluxated lens and intraocular hemorrhage. Extraocular extension can manifest with proptosis, orbital cellulitis or proptosis. Pineal and parasellar regions can also be affected (trilateral retinoblastoma).

Conclusion

Retinoblastoma is usually diagnosed before the age of 3 years (90% of patients), but can occur in older children. General practitioners should be aware that retinoblastoma can mimic ocular inflammation and any red and painful eye, regardless of age, should have a prompt dilated ophthalmologic evaluation.

P 21**Be aware of floppy but alert baby**

Andrade Borges S¹, Papasavva D¹, Russo M¹, Llor J¹, Paccaud Y¹, Hernandez E¹, Faignart N², Poloni C²

¹Service de Pédiatrie, Hôpital du Valais, Centre Hospitalier du Valais Romand, Sion, Switzerland; ²Unité de neuropédiatrie, Service de Pédiatrie, Hôpital du Valais, Centre Hospitalier du Valais Romand, Sion, Switzerland

Case report

3-m.o. boy admitted for persistent hypotonia, feeding difficulties and paradoxical breathing, without fever or prior infection.

History of term birth (induced for decreased fetal movements), hospitalized for primary adaptation disorder (APGAR 5/7/9), transient tachypnea, mild axial hypotonia (head lag) and hypoglycemia. Family history was not contributive.

Neurologic exam showed an alert baby, good visual contact and expressive mimicry, severe axial (head lag) and peripheral hypotonia (hips in external rotation, knees in flexion or frog-leg posture, scarf sign, poor spontaneous motility), areflexia, tongue fasciculations, good suction, rapid, irregular belly

breathing and bell-shaped chest. Laboratory: normal blood count, gazometry and thyroid tests. CK at 263 U/L (N< 190). Genetic analysis: deletion in SMN1.

Diagnosis of spinal muscular atrophy type 1 (SMA type 1) was retained. Palliative care or nusinersen treatment were discussed with parents and first option was chosen due to already severe respiratory insufficiency. Clinical course was unfavorable, with death at 4 months.

Discussion

SMA type 1 is a severe autosomal recessive neuromuscular disorder caused by inactivating mutations in SMN1 (incidence of 1:10000), leading to insufficient SMN protein production. Clinical picture in SMA is variable due to SMN2 paralog, providing partial function. SMA type 1 is most severe, neonatal form. Absence or low levels of SMN protein lead to apoptosis of lower motor neurons in anterior horn and lower brainstem, resulting in weakness and muscular atrophy. Onset of symptoms is before 6 months of age, rarely achieving motor developmental milestones, and mortality by the age of 2.

Diagnosis is clinical and confirmed by genetic testing. Floppy but alert baby with tongue fasciculations and areflexia should be investigated for SMA.

CK can be elevated, EMG can be helpful. Muscle biopsy is not indicated.

Therapy was mainly supportive but nusinersen, antisense oligonucleotide in intrathecal injection, which corrects defective splicing and converts thus SMN2 functionally to SMN1, can improve motor function and delay disease onset. Another new treatment has been recently developed, onasemnogene ABEPR-VOVEC (AVXS-101), using virus capsid to deliver SMN functional gene copy to motor neurons.

Conclusion

SMA type 1 is a rare but well-known cause of hypotonia. Clinical features are typical. Early diagnosis is essential, because new treatments are available.

P 22**Persistent ductus arteriosus: persistent to treat or not to treat challenge in current NICU**

Colombo R¹, Salomon C¹, Barcos-Munoz F¹

¹Neonatal and Pediatric Intensive Care, Geneva University Hospitals, Switzerland

Background

Persistent ductus arteriosus (PDA) is a major challenge associated with prematurity and its incidence is inversely proportional with gestational age (GA) at birth. In preterm ≤ 28 weeks' GA incidence is over 50%. PDA can lead to multi-organ morbidities, remarkably on the cardiorespiratory system, brain, gastrointestinal tract and kidney. From 1980s on, the introduction of NSAID, along with other neonatal advancements, improved the survival rate of the preterms. Better survival rate brought about a younger preterm population with a reduced response to treatments.

Case

We present a 690g dizygotic twin delivered at 27+4 weeks gestation, who developed respiratory instability during the first 2 weeks of life consistent with the presence of PDA. On the 4th day, due to persistent respiratory symptoms despite surfactant administration at day 1, the first cardiac echo was performed and showed a dilated ductus arteriosus (DA) with significant left cavities dilatation. A cerebral ultrasound revealed altered doppler values. In this context of hemodynamic significant PDA (hsPDA), a first ibuprofen cure was administered. At 7 days of life (DOL) he was treated by vancomycin-gentamicin due to a late-onset sepsis caused by *Staphylococcus Warneri*. At 8 DOL he received a second ibuprofen cure (high oral dose) without success with increase in respiratory distress and an impact on cerebral and intestinal blood flow. The follow up by echocardiography confirmed once again a hsPDA and suspicion of aortic coarctation. Finally, at 12 DOL the patient underwent surgical ligation with careful monitoring of 4 limb tension and aortic morphology on echocardiography. Following the intervention, we witnessed a rapid clinical and hemodynamic stabilization without sign of coarctation.

Discussion

PDA management is an increasingly evolving challenge in NICUs. Physician has to take into account not only the effects related to hsPDA, such as excessive pulmonary blood flow (risk of pulmonary oedema, hemorrhage and bronchopulmonary dysplasia) and reduced systemic blood flow due to PDA diastolic steal phenomenon (risk of NEC, IVH), but also the consequences related to treatments (notably low urine output due to NSAID). The balance achieved is a tailored medicine only partially supported by evidence-based choices.

P 23**Resistant skin lesions, think about zinc !**Vasey L¹, Papadimitriou V¹, Alipour Therany Y², Stollar F¹¹Department of Pediatrics, Geneva University Hospital, Geneva, Switzerland; ²Department of Dermatology and Venereology, Geneva University Hospital, Geneva, Switzerland**Background and importance**

Zinc is an essential element of our body. The skin is the third largest consumer of zinc in the organism, guaranteeing the effectiveness of the epithelial barrier at the epidermal level via the proliferation of keratinocytes and via the self-regulation of the hydric load of water. Zinc is involved in cellular immunity through physiological immune suppression.

The diagnosis of zinc deficiency is often delayed. Its prompt recognition is important in cases treatment resistant skin lesion and candida infections in order to provide the correct treatment allowing a rapid improvement of the condition.

Case presentation

We report the case of a 5 months old male infant, who was admitted at Geneva University children Hospital due to diffuse erythematous-squamous, desquamating lesions, well delimited on the face and periorificial (with sparing of the scalp and the upper lip), the inguinal folds, the penis and the seat. The first skin lesions appeared a month ago, at the same time as an oral thrush. Initially, a treatment of topical and oral antifungals was tried, but after a lack of response and in view of a negative mycotic smear, a seborrheic dermatitis has been suspected. A local and oral treatment of corticosteroids was therefore started without any success. Moreover, the oral thrush did not disappear despite the local antifungal treatment. Psychomotor development was within normal limits with normal cerebral ultrasound and MRI. He was breastfed since birth with good weight growth but poor statural growth despite breast milk enrichment. Immune function was measured in normal range. He was born at 24 3/7th gestational weeks and presented a neonatal candidemia. Family history shows that both parents have a history of eczema. A zinc dosage was performed, diagnosis of zinc deficiency was confirmed, and the infant had rapid improvement with zinc supplementation. The zinc level has been dosed out in normal range in the maternal blood and milk.

Conclusion

In pediatrics, zinc deficiency should be suspected in the presence of recurrent infections, particularly candida infections, and skin disease refractory to treatment, especially in premature babies. Skin lesions take may be erythematous, confluent and eczematous with significant desquamation, which may resemble seborrheic or atopic dermatitis and have a typical facial and sacral localization

P 24**Hyperinsulinism as a cause of recurrent hypoglycemia in a low birth weight newborn**Crédeville M¹, Salomon C¹, Barcos-Munoz F²¹Service de pédiatrie générale, Département de la Femme, de l'Enfant et de l'Adolescent, Hôpitaux Universitaires de Genève; ²Service de Néonatalogie et des Soins Intensifs Pédiatriques, Département de la Femme, de l'Enfant et de l'Adolescent, Hôpitaux Universitaires de Genève**Introduction**

Neonatal hypoglycemia frequently occurs in newborn at risk, is often asymptomatic, and could lead to neurological damage if left untreated. Hyperinsulinism is a known cause of recurrent hypoglycemia with an incidence of 1:40 000 births, usually in children of diabetic mothers. Due to alteration of fetal metabolism, preterm and low birth weight newborns are at risk of hyperinsulinemic hypoglycemia. We'd like to share our experience, likely to be encountered in any maternity ward.

Case report

We report the case of a male preterm, born at 35 4/7 weeks of amenorrhea in a secondary medical center in Geneva. Intrauterine growth restriction was early discovered during the pregnancy follow-up, without chromosomal anomalies detected nor maternal diabetes. Childbirth occurred after labor induction because of oligohydramnios and growth restriction. The adaptation to extrauterine life was unremarkable. He underwent a first hypoglycemia at 0.3 mmol/l at two hours of life, discovered through a hypothermia at 35 degree Celsius. He was treated with IV bolus of dextrose 10% in water, followed by IV infusion and continuous enteral feeding. Severe hypoglycemia recurred at 24 hours of

life, after switching to discontinuous enteral feeding, without any symptom. To optimize patient care, he was transferred to a tertiary hospital at 36 hours of life. There he underwent hypoglycemia work-up, showing insulin level at 20mUI/l, consistent with hyperinsulinism. No other metabolic disturbance was found. The case was discussed through multidisciplinary meetings with the endocrinologists and the diagnosis of transient hyperinsulinism due to fetal growth restriction was retained. Significant hypoglycemia occurred repeatedly until 3 days of life. In order to maintain blood sugar levels in normal range, glucose infusion rate was increased up to 15mg/kg/min. No other treatment was given. Glucose intake was slowly decreased with good tolerance. A subcutaneous glucose monitoring was set up, helping us to know when the glycemia had a downward trend.

Conclusion

Even though transient hyperinsulinism is the most common cause of neonatal hypoglycemia, it remains pretty difficult to manage when hypoglycemia is recurrent. This occurrence may last days, even weeks before going back to normal and sometimes requires other treatments. Although screening of patients at risk is recommended, it might be worth it to regularly reassess during a few days after a first episode of hypoglycemia.

P 25**Not all bone lesions in children are osteomyelitis****Case report**Rouge Elton Paola Andrea¹, Rossé Mélanie¹, Zimmermann Petra^{1,2}¹Department of Paediatrics, Fribourg Hospital HFR, Fribourg, Switzerland; ²Faculty of Science and Medicine, University of Fribourg, Fribourg, Switzerland**Introduction**

Early diagnosis and treatment of osteomyelitis in children is critical to prevent long-term sequelae. However, not all bone lesions in children correspond to an osteomyelitis. We present an infant with decreased leg movements who was found to have multiple bone lesions in atypical locations for osteomyelitis.

Case

A 5-month-old, previously healthy boy presented with intermittent fever since 2 weeks and decreased leg movements. The physical exam revealed painful passive movement of the left hip without local signs of inflammation. The osteotendinous reflexes, as well as the rest of the neurologic exam were normal. Laboratory tests did not show signs of inflammation but a hypochromic normocytic anaemia with a haemoglobin (Hb) level of 92 g/l. Urine analysis, a chest X-ray and an abdominal ultrasound were normal. An ultrasound of the hips did not show an effusion. The child was started on intravenous antibiotics covering typical bacteria which cause osteomyelitis. A magnetic resonance imaging (MRI) showed multiple bone lesions with oedema-like signal alterations in the spinous process of the second lumbar vertebra as well as in the left ischium, left fibula, right cuboid bone, and in several ribs.

Because the bone lesions were in atypical locations for an osteomyelitis at that age, further tests were done to exclude haematological and oncological causes of the bone lesions, as well as immunodeficiency. A bone biopsy did not show signs of infection and cultures remained sterile. With the help of a haemoglobin electrophoresis the diagnosis of homozygous sickle-cell disease (SCD) was made. The HbS was 63%.

Conclusion

Differentiation of vaso-occlusive ischemia/bone infarction from osteomyelitis in children with SCD remains challenging. There is a limited evidence guiding management, in case of doubt, a bone biopsy or aspiration should be done

P 26**Presumptive foodborne botulism in a 5-month-old infant caused by home-made food**Schaub B¹, Ott A², Luhmann-Lunt C³, Mandanis X⁴, Dorner M⁵, Bauder F¹, Buettcher M⁶¹ Department of Paediatric Neurology, Lucerne Children's Hospital; ² Department of General Paediatrics, Lucerne Children's Hospital; ³ Department of Paediatric Intensive Care Medicine, Lucerne Children's Hospital; ⁴ Department of Paediatric Emergency, Lucerne Children's Hospital; ⁵ German National Consultant Laboratory for Neurotoxin-producing Clostridia, Robert Koch Institute, Berlin; ⁶ Department of Paediatric Infectious Diseases, Lucerne Children's Hospital

Introduction

Botulism is a rare and potential life-threatening disease caused by the toxins of *Clostridium botulinum*, an anaerobic bacillus found ubiquitously in the soil. Foodborne botulism caused by ingestion of pre-formed toxins in food needs to be differentiated from infant botulism, where ingested spores germinate and release toxins in the infantile gut. Anyway, early diagnosis and treatment is crucial to prevent mortality and reduce the duration of symptoms and hospital stay. Clinical presentation in infants can be less obvious and very few foodborne botulism cases have been described in this age group. To the best of our knowledge, this case presents the youngest infant with laboratory confirmed foodborne botulism, so far.

Case

A 5-month-old, previously healthy Caucasian boy presented with acute onset of refusal to drink and slightly increased sleepiness. Over the next hours he developed non reactive mydriasis, reduced spontaneous eye-opening and facial expression, aphagia without increased drooling as well as a descending flaccid paralysis with intact muscle reflexes. Admission to the ICU, with intubation for airway protection, was necessary about 9 hours after admission. The ICU stay was complicated by a systemic inflammatory response and paralytic ileus. Botulism was suspected early and heptavalent antitoxin could be administered about 24 hours after presentation. Toxicology screening, cranial MRI, CSF analysis and aEEG were normal. A more detailed history revealed ingestion of insufficiently heated home-made baby food around 30h prior to his symptom onset. Botulism could be confirmed by the presence of neurotoxin type B (BoNT/B) producing *Clostridium botulinum* in the patient's stool and home-made food. He recovered slowly and could be extubated after 5 days. 3 weeks after admission he was transferred to a rehabilitation clinic with a nasogastric feeding tube, only.

Discussion & conclusion

Although infant botulism is more common at the patient's age-group, especially the rapid progression of symptoms makes foodborne botulism more likely. If suspected at any time, treatment with heptavalent antitoxin should never be delayed. The aim of this presentation is to report the onset and recovery of each symptom as detailed as possible to (re)raise awareness of this rare disease and to provide a reference course for future cases of foodborne botulism in infants.

P 27

Acquired methaemoglobinaemia – a rare cause of cyanosis in infants

Rimle C¹, Herenger Y^{1,2}, Neuhaus TJ¹

¹Children's Hospital Lucerne, Department of Paediatrics, Lucerne, Switzerland; ²Laboratories Genetica, Zurich, Switzerland

Introduction

Cyanosis in children mostly results from cardiac or pulmonary disorders. However, it is important not to miss other reversible causes. We present a case of acquired methaemoglobinaemia in an infant.

Case report

A 6-week-old boy presented with blue-grey skin colour and decreased oxygen saturation with no signs of respiratory distress. Peripheral saturation was 86% and did not improve with supplementary oxygen. There were no clinical signs of heart failure or pulmonary disease. The patient was on antibiotic prophylaxis with Cotrimoxazole after two episodes of pyelonephritis (*Enterococcus faecalis* and *Klebsiella oxytoca*). Despite the blue-grey skin and decreased peripheral oxygen saturation, he was in good general condition. Capillary blood gas analysis showed a methaemoglobin fraction of 23.8%. Methylene blue was given intravenously as emergency treatment in a total dose of 2mg/kg. Methaemoglobin levels and skin colour normalized quickly after treatment. Diagnostic investigations included haemoglobin electrophoresis, Glucose-6-phosphate dehydrogenase (G6PD) activity and genetic analysis (Cytochrome-b5 reductase gene; CYB5R3), which all were normal. Cotrimoxazole was stopped and the parents were instructed to avoid drugs that potentially act as oxidizing agents. So far, methaemoglobin level has remained normal.

Discussion

Methaemoglobinemia leads to decreased transport and delivery of oxygen. Clinical signs are blue-grey appearing skin without respiratory distress, decreased peripheral and normal arterial oxygen saturation. Methaemoglobinemia results from exposure to oxidants or rarely from congenital deficiency of G6PD or CYB5R3. Infants are more susceptible because levels of methaemo-

globin reductase are lower than in adults. Since the infant in our case had already presented with slightly elevated levels of methaemoglobin prior to Cotrimoxazole intake, a genetic cause was suspected. However, no pathogenic variation in the CYB5R3 gene was detectable.

Conclusion

Methaemoglobinemia as a cause of cyanosis is rare, but should be considered if peripheral oxygen saturation is low and does not improve with supplementary oxygen. Diagnosis is based on blood gas analysis or multiwave pulse oximetry. Emergency treatment consists of intravenous Methylene blue. Differential diagnosis includes genetic diseases or acquired causes, in particular medications as suggested in our case.

P 28

A case of meningitis and Herpes Zoster from varicella reactivation in an immunocompetent adolescent

Marangoni Alice Serena¹, Bombeli Audrey¹, Zemmouri Abdelaziz¹, Bopst Léa¹, Chariatte Vincent¹

¹Department of Pediatrics, EHC - Ensemble Hospitalier de la Côte, Switzerland

Background

Varicella zoster virus (VZV) is a member of the herpesvirus family and causes varicella (chickenpox). After the primary infection, VZV establishes latency in the cranial nerve and dorsal root ganglia. The development of neurological complications due VZV reactivation is relatively uncommon, particularly in immunocompetent paediatric patients. The virus reactivation frequently results in the painful dermatomal rash of herpes zoster.

Case presentation

A previously healthy 14-years-old girl presented a 3-days history of increased pulsatile occipitotemporal headache, backpain, neck stiffness, and petechiae on feet and ankles, a 1-day of intermittent nocturnal food vomiting, dizziness during changes of position and skin rash compatible with Herpes Zoster. The patient had chickenpox when she was 3 years old and presented no known immunodeficiency.

Upon admission, her physical examination revealed normal hemodynamic parameters, no fever, a maculopapular rash evolving into vesicles with erythematous regions on the left side of her chest, and normal neurological examination results. Routine blood examination results shown microcytic anaemia associated with leukopenia with no increase in inflammatory biomarkers. The encephalic RM findings were normal; CSF revealed the presence of VZV, associated with leucocytosis and hyperproteinaemia. Bacterial culture of CSF yielded no growth, and the blood culture was negative. A swab from a skin lesion was positive for VZV.

The diagnosis made was of VZV reactivation meningitis and Herpes Zoster.

The patient was treated with intravenous Acyclovir (10 mg/kg every 8 hours). Her symptoms improved, and she was discharged after the 3rd day of hospitalization with PVC to complete a 10-days course of acyclovir.

Conclusions

Although rare, the reactivation of VZV should be recognized as a potential cause of meningitis in immunocompetent paediatric patient but it is not one of the most severe neurological complications and usually does not lead to the major brain damage characterising VZV myelopathy, vasculopathy or eye disease. Therefore, high level of suspicion is required even for those patients in which suggestive clinical features are not present to guide the diagnosis, especially in rare cases where Herpes Zoster may be absent, or it may appear later than other symptoms.

Intravenous Acyclovir represents the treatment of choice to obtain a fast clinical response, preventing the onset of late-term complications.

P 29

Back Pain: A Royal Illness?

Beck Samuel¹, Büttcher Michael²

¹General Paediatrics, Children's Hospital Lucerne, Cantonal Hospital Lucerne, Switzerland; ²Paediatric Infectious Diseases, Children's Hospital Lucerne, Cantonal Hospital Lucerne, Switzerland; ³Pediatric Surgery, Children's Hospital Lucerne, Cantonal Hospital Lucerne, Switzerland

Background

Spondylodiscitis is a rare condition among children, accounting for around 3% of the osteoarticular infections in children. Clinical symptoms are unspecific,

including low grade fever, back pain and only in few cases neurological manifestations. Because the early symptoms are unspecific, the diagnosis is often delayed. Cultivation of a pathogen from blood or vertebral fluid is difficult. Among the positive cultures, *Staphylococcus aureus* is detected in the majority of cases. For spondylodiscitis among children from 6-48 months, *Kingella kingae* is considered the leading cause.

Case presentation

Summary A 14-months-old, previously healthy and very active girl was evaluated for refusal of walking, decreased activity, low grade fever, weight loss and a swelling of the lower back. She had normal vital parameters and showed an unsteady, limping gait with a stiff back and reduced arm movement. The manual muscle testing of the lower extremities as well as the reflex status were normal. The laboratory findings showed slightly increased inflammation parameters with a white blood cell count of 11.7 G/L, a C-reactive protein of 23 mg/l and an erythrocyte sedimentation reaction of 46mm/h. The MRI demonstrated a T2 hyperintensity of the lumbar vertebrae L1 and L2 including the disc in between, as well as in the left psoas muscle. Cultures of a puncture of the muscle were positive for *Kingella kingae*. Amoxicillin/clavulanic acid therapy was administered intravenously for seven days and afterwards continued orally for a total duration of four weeks. An orthopedic corset was purpose-built. The child recovered without residues.

Discussion / learning points

Spondylodiscitis is a rare reason for back pain and subfebrile temperatures in children. Laboratory findings only show a slight inflammation. The increasing availability of MRIs led to a more frequent and earlier diagnosis. *Kingella kingae* (KK) is considered the leading cause of osteoarthritis (and therefore spondylodiscitis) in children between 6-48 months. Cultivation of KK is difficult, PCR should be performed in addition. If a PCR on an oropharyngeal swab is positive for KK, an antibiotic therapy with betalactams can be started. For spondylodiscitis in general, it is recommended to start intravenous treatment with broad-spectrum antibiotics, which are active against *S. aureus*. Recommendations regarding the treatment duration vary from 3-14 days of i.v. antibiotic therapy and a total duration of 3-4 weeks.

P 30

A 3-month-old infant with failure to thrive and hyponatremia: remember the role of mineralocorticoids.

Fillistorf Laura¹, Papathanasiou Matthildi¹, Pittet Anne¹, Armengaud Jean-Baptiste¹, Gehri Mario¹, Hauschild Michael²

¹Service of Pediatrics, Department Women-Mother-Child, Lausanne University Hospital, Lausanne, Switzerland; ²Endocrinology Diabetology Unit, Service of Pediatrics, Department Women-Mother-Child, Lausanne University Hospital, Lausanne, Switzerland

Failure to thrive is a common cause for consultation in primary care setting, and its differential diagnosis is large. Thoughtful consideration is needed in order to quickly recognize rare but life-threatening conditions.

We report the case of a 3-month-old female, born at term to non-consanguineous healthy parents, presenting with severe failure to thrive (weight 3.075kg (-5.6 SDS according to WHO), length 56cm (-2.97 SDS) and BMI 9.8kg/m² (-5.69 SDS)). She was breastfed and temporarily supplemented with formula milk. Except for mild dehydration, physical exam was unremarkable, including normal development for age. The newborn screening was normal. Laboratory workup revealed severe hyponatremia (124 mmol/l, N 134-142), hyperkalemia (max. 6.6 mmol/l, N 3.5-5.6) and hyperreninemia (> 100 ng/ml/h, N < 3). LC-MS (liquid chromatography-tandem mass spectrometry) steroid profile showed a marked elevation of the aldosterone-precursors 11-deoxycorticosterone and corticosterone. Serum aldosterone level was normal to high. The rapid response to mineralocorticoids (fludrocortisone) and oral sodium supplementation, with normalization of natremia and sustained weight gain, argued against a type 1 pseudohypoaldosteronism (PHA1) due to mineralocorticoid-receptor loss of function, therefore suggesting a type II aldosterone synthase deficiency (ASD). ASD is coded by the CYP11B2 gene, and is a rare form of congenital hyperreninemic hypoaldosteronism inherited as either an autosomal recessive or autosomal mixed penetrant dominant trait.

The presentation of an infant with insufficient mineralocorticoid action may vary, but typically infants with ASD or PHA1 present early in the first weeks of life with vomiting, failure to thrive and a salt-wasting syndrome which can be life-threatening. Specific hormonal and genetic analysis are required for final diagnosis.

With this case report, we aim to sensitize pediatricians about the clinical presentation of this entity as well as immediate action to take, including emergency lifesaving treatment.

Acknowledgement: Dr Cachat

P 31

Case report: Rapunzel, Rapunzel, you look so pale. Pica syndrome as clinical manifestation of celiac disease

Dusoczky N¹, Wehrli L³, Spalinger J^{1,2}, Neuhaus TJ¹, Büttcher M¹, Righini-Grunder F^{1,2}

¹Department of Pediatrics, Children's Hospital, Lucerne, Switzerland; ²Department of Pediatric Gastroenterology, Hepatology and Nutrition, Children's Hospital, Lucerne, Switzerland; ³Department of Pediatric Surgery, Children's Hospital, Lucerne, Switzerland

Introduction

Pica syndrome is usually a psychological eating disorder characterized by an appetite for largely non-nutritive substances, such as hair (trichophagia). However, Pica syndrome may also occur secondary due to deficiency of micronutrients, such as iron.

We present the case of a six-year-old girl with trichophagia, resulting in a bezoar and extending from the stomach to the duodenum (Rapunzel syndrome).

Results

A 6-year-old girl was referred to our pediatric gastroenterology outpatient clinic with chronic epigastric pain syndrome, resulting in loss of appetite and failure to thrive.

Clinically, patchy alopecia and a hard mass (8x10cm), in the upper abdomen were detected. Ultrasound and abdominal X-ray revealed a distended stomach and proximal duodenum due to an intragastric mass. On further questioning, the parents reported that she had been plucking hairs and ingesting these over the past 6 months.

Laboratory investigations demonstrated iron-deficiency anemia (Hb 10mg/l, MCV 69fl, MCH 23pg, Ferritin 8µg/l). Work-up for an underlying cause revealed a more than ten-fold elevated tissue transglutaminase IgA, positive Endomysium IgA and positive genetic predisposition (HLA-DQ2 positive) suggesting Celiac Disease. The trichobezoar, extending from the stomach to the duodenal bulb, was removed by laparoscopic gastrotomy. Additional mucosal biopsies confirmed an early stage of celiac disease (Marsh-Oberhuber-Classification Type 1).

Conclusion

Iron-deficiency anaemia is a common finding in ambulatory care. In infancy often due to malnutrition, however, in older children other causes have to be ruled out. Deficiency of micronutrients such as iron or vitamins may lead to peculiar eating behaviors such as trichophagia, presenting as Pica syndrome. In patients with Pica syndrome, an underlying somatic disease leading to intestinal malabsorption, such as celiac disease, should, therefore, be ruled out.

P 32

Rumination syndrome, a frequently overlooked entity

Rosenblatt L^{1,2}, Sokollik C³, Spalinger J^{1,2}, Righini-Grunder F^{1,2}

¹Department of Pediatrics, Children's Hospital Lucerne, Switzerland; ²Department of Pediatric Gastroenterology, Children's Hospital Lucerne, Switzerland; ³Department of Pediatric Gastroenterology, Children's university hospital Inselspital, Berne, Switzerland

Introduction

Rumination syndrome, a functional gastrointestinal disorder, is infrequently recognized or misdiagnosed leading to delayed diagnosis, unnecessary investigations and treatments, and might have a negative impact on patients' quality of life. It is characterized by recurrent effortless regurgitation of recently ingested food followed by re-swallowing or spitting. Rumination syndrome is primarily a clinical diagnosis using Rome IV criteria, whereas high resolution esophageal manometry and impedance-pH probe can help to support the clinical diagnosis.

Aim

We aim to raise awareness of Rumination syndrome in Swiss patients and the role of impedance-pH probe and high resolution esophageal manometry.

Methods

Case series

Results

We report on four cases (Three males, one female, median age 13 years) who were referred to us for a second opinion. Persistent symptoms in form of effortless regurgitation, belching and abdominal discomfort have been reported over a median time of 3.5 years prior to diagnosis of Rumination syndrome. Previous investigations were gastroscopies in three patients, therefore two with multiple gastroscopies. All but one had one or multiple swallow studies. All patients were treated with PPI without clinical response. Impedance-pH probe was performed in three patients, showing high numbers of retrograde liquid flow in the esophagus during daytime, but no pathological reflux episodes during night. High resolution manometry was performed in 3 patients. Normal esophageal peristalsis was confirmed in all patients and in two patients' rumination episodes were observed. After patient education and behavioral therapy, two out of four patients stated subjective improvement. One needed baclofen as a peroral treatment to diminish frequency of transient lower esophageal sphincter relaxation.

Conclusion

Physicians should consider rumination syndrome as a differential diagnosis in patients presenting with regurgitation, especially when occurring immediately after food ingestion and without retching. Diagnosis is crucial to prevent unnecessary costly tests and to implement appropriate treatment to reduce patient suffering. If rumination syndrome is suspected, impedance-pH probe and high resolution manometry might be helpful tools to confirm diagnosis and rule out gastro-esophageal reflux disease.

P 33**Three cases – one rare manifestation: hypercalcaemia in infants**

van Ransbeeck A¹, Lurà M¹, Herenger Y^{1,2}, Neuhaus T J

¹Department of Paediatrics, Children's Hospital Lucerne, Switzerland; ²Genetica, Zurich, Switzerland

Introduction

Symptomatic hypercalcaemia in infants is rare, but has serious consequences. Clinical symptoms include failure to thrive, dehydration, hypertension and muscular hypotension.

Case 1

A 2 month-old girl presented with peripheral pulmonary stenosis, supra-aortic stenosis and several dysmorphic features. Williams-Beuren syndrome was genetically confirmed with de-novo heterozygote deletion 7q11.23. Further development showed decreased oral intake and failure to thrive. Mother had stopped vitamin D. At 9 months length and weight were <P3, leading to gastrostomy (PEG). Blood tests revealed hypercalcaemia (ionized 2.2 mmol/l) and suppressed PTH (7 pg/ml). Ultrasound showed medullary nephrocalcinosis. Hypercalcaemia persisted with repeated episodes of dehydration and prerenal failure (creatinine 142 umol/l). A single-dose of intravenous bisphosphonate (Pamidronate, 1mg/kg) was given, normalising calcium within 6 days. High fluid intake and low calcium diet were introduced. Two years later, calcium has remained normal; growth and development have improved.

Case 2

A 6 month-old girl presented with failure to thrive (weight <P3). She was breast-fed with extra BEBA FM85. Blood tests revealed hypercalcaemia (ionized 1.54 mmol/l), low phosphate (1.17 mmol/l) and suppressed PTH (5 pg/ml). Creatinine was normal, but renin and aldosterone were high, suggesting dehydration. Ultrasound showed medullary nephrocalcinosis. Vitamin D and FM85 were stopped; oral phosphate substitution was started with rapid improvement of general condition and weight. Genetic analysis is pending for infantile hypercalcaemia type II (SC34A1).

Case 3

A 4 month-old girl presented with urinary tract infection. Ultrasound revealed medullary nephrocalcinosis. Blood tests showed mild hypercalcaemia (total 2.69 mmol/l), normal phosphate (1.9 mmol/l) and slightly low PTH (14.9 pg/ml). Vitamin D was stopped, resulting in normalized calcium and PTH level. Genetic analysis is pending for infantile hypercalcaemia type I (CYP24A1). The girl has a dysmelia with no known assimilation with hypercalcaemia and will be treated as independent diagnosis.

Conclusions

Hypercalcaemia requires an interdisciplinary diagnostic and therapeutic approach to differentiate genetic, syndromic and acquired causes. Treatment includes vitamin D avoidance and high fluid intake, in selected cases low calcium diet and bisphosphonates.

P 34**Neonatal seizures – triggered by the child or by the mother?**

Danioth LP¹, Bauder F^{1,2}, Lurà M^{1,3}, Büttcher M^{1,4}, Neuhaus TJ^{1,5}

¹Department of Paediatrics, Lucerne, Switzerland; ²Department of Paediatric Neurology, Lucerne, Switzerland; ³Department of Paediatric Pulmonology, Lucerne, Switzerland; ⁴Department of Paediatric Infectious Diseases, Lucerne, Switzerland; ⁵Department of Paediatric Nephrology, Lucerne, Switzerland

Background

Maternal vitamin D deficiency during pregnancy can cause neonatal seizures.

Case 1

7-day-old term Pakistani male neonate presented in status epilepticus with multifocal clonic and myoclonic seizures. Pregnancy was complicated by gestational diabetes. A presumed infectious cause was treated (amoxicillin, amikacin, acyclovir). Full sepsis work-up was negative and treatment stopped. Cranial sonography/MRI/EEG were normal. Severe hypocalcaemia (ion.0.77, tot.1.37 mmol/l), hypomagnesaemia (0.49mmol/l), hyperphosphataemia (2.97mmol/l) and inadequately normal PTH (49 pg/ml, range 15-65) were detected. Despite intravenous calcium gluconate (0.5-1.5 mmol/kg/d) and magnesium (1mmol/d), seizures persisted. Phenobarbital was started and the seizures stopped. Review of his mother revealed severe vitamin D deficiency (9.2nmol/L) and elevated PTH (191pg/ml, range 15-65). She was not wearing a Hijab and had no Vitamin D supplementation. The calcium substitution was continued and additionally vitamin D (1000 U/d) was given. His calcium, magnesium and phosphate levels normalized within one week. Phenobarbital was stopped after 7 days. At 2 week follow-up no further seizures/myoclonia were noted and all laboratory values had normalised and remained stable at 3 months.

Case 2

7-day-old term Caucasian female neonate presented with multifocal clonic and myoclonic seizures. Paternal history was positive for seizures. Cranial sonography/MRI were normal. Significant hypocalcaemia (ion.0.94, tot.1.73 mmol/l), hypomagnesaemia (0.57mmol/l), hyperphosphataemia (3.14 mmol/l) and inadequately normal PTH (45 pg/ml) were detected. Therapy included calcium gluconate (0.5mmol/kg/d), vitamin D (1000 U/d) and magnesium (1mmol/d). For the first 24 hours seizures increased. After starting phenobarbital seizures stopped. Review of her mother blood revealed Vitamin D deficiency (49.7nmol/L) and normal PTH (40.6pg/ml). Calcium and vitamin D were continued, phenobarbital was stopped after 2 and magnesium after 4 days. Within one week all laboratory values had normalised and remained stable at 2 months of age.

Discussion

Both neonates suffered from hypocalcaemia secondary to transient hypoparathyroidism, leading to seizures. Severe maternal vitamin D deficiency with secondary hyperparathyroidism during pregnancy causes transient suppression of the foetal/neonatal parathyroid gland. Maternal vitamin D deficiency in pregnancy is common, particularly in women of colour or with limited sun exposure.

P 35**Discovering Resources Beyond the Standard of Care – Impact of an integrative care program for children with cancer in Brazil**

Tröndle Marc¹, Stritter Wiebke¹, Odone Vicente², Peron Karina², Ghelman Ricardo², Seifert Georg^{1,2}

¹Department of Pediatric Oncology/Hematology, Otto-Heubner Centre for Pediatric and Adolescent Medicine (OHC), Charité – Universitätsmedizin Berlin, corporate member of Freie Universität Berlin, Humboldt-Universität zu Berlin and Berlin Institute of Health, Berlin, Germany; ²Universidade de São Paulo, Faculdade de Medicina, Departamento de Pediatria, Instituto de Tratamento do Câncer Infantil (ITACI), São Paulo, São Paulo, Brazil

Introduction

This article examines the feedback of health care providers within the implementation of an Integrative Care project in a clinic for pediatric oncology in São Paulo, Brazil. Since 2018 the project has implemented external anthroposophic therapies in the activities of daily nursing. The objective is to evaluate how the project evolved and what impact it had on the daily operation of the hospital. A special focus emphasizes the perspective of study nurses.

Materials and Methods

Qualitative semi-structured interviews were conducted. Audio files were transcribed, translated to German and underwent a MAXQDA software assisted analysis. Using a thematic approach, coherent cross-case topics were defined.

Results

Three main topics emerged from analysis of the data. 1.) The implementation and its effects on daily patient care demonstrated positive outcomes in patients and were well-accepted with minimal changes in daily activities. 2.) The perspective of study nurses showed a large motivation due to beneficial and stress-relieving effects of the application and a growing patient-caregiver relationship. 3.) Problems and aspirations for improvement were the lack of time and the urge to make the project grow in the future.

Conclusion

Not only patients but also health care providers seem to benefit from integrative methods. They have the potential to improve the working atmosphere and to strengthen relations between patients, caregivers and family members. General feedback was positive and acceptance in the team arose over time when beneficial effects became visible.

P 36

A neonatal case of Wolff-Parkinson-White syndrome

Mazzara C¹, Leoni-Foglia C¹, Balmer C², Stefani-Gluecksberg A¹

¹Istituto Pediatrico della Svizzera Italiana, Ospedale San Giovanni, Bellinzona, Switzerland; ²Universitäts-Kinderspital, Zürich, Switzerland

Background

The pattern of Wolff-Parkinson-White (WPW) is an abnormality of the electrical conduction with an accessory pathway which transmits electrical impulses from the atria to ventricles outside of normal conduction system. This produces a typical Delta wave or pre-excitation in the electrocardiogram (ECG). WPW syndrome is associated with supraventricular tachycardia (SVT) as orthodromic or antidromic atrioventricular reentrant tachycardia (AVRT). Usually is a benign pathology although the incidence of sudden cardiac death is estimated at 0.15-0.39% and the risk of heart failure (HF) is higher in newborns and infants.

Case description

A 24 days old female newborn was conducted to our emergency room for 24 hours history of decreased appetite, persistent crying and apathy. No history of fever, vomiting or diarrhoea. At clinical examination she presented marbled skin and slowed refill. Blood pressure was 86/57 mmHg and heart rate (HR) 270 bpm. Respiratory rate was increased with normal O₂ saturation. Inferior hepatic margin was palpable at 3 cm from costal arch. Haemogram and inflammatory test were negative; moderate acidosis was registered. The ECG evidenced a SVT not responsive to vagal manoeuvres. An echocardiogram showed low left ventricular ejection fraction (20-30%), mitral insufficiency and dilatation of inferior vena cava. Because of progressive clinical worsening in a reactive and awake patient, a pharmacological cardioversion with IV adenosine 0.2 mg/kg under ECG monitoring was performed, in operatory room, without response. Adenosine at 0.4 mg/kg was repeated with restoration of sinus rhythm, normal HR and Delta waves. The diagnosis of WPW syndrome with AVRT and secondary HF was confirmed. Because of worsening of acidosis and of respiratory activity the patient was admitted to Paediatric ICU. After 3 months she had a normal heart function under Propanolol and Flecainide without new episodes of TPS.

Discussion

The suspicion or clinical diagnosis of WPW syndrome in paediatric patients is easy because the children present palpitations, chest discomfort, shortness of breath, dizziness, fatigue, irritability, anxiety, ashen colour and syncope. The challenge is to recognize the AVRT in newborns and infants. HF is developed in short time in foetus and neonates and has a relation with the HR of SVT. Recognizing and promptly treating patients with WPW syndrome in this special age group is fundamental to avoid complications and the risk of death.

P 37

Experience of monitoring children in need of multidisciplinary intervention in a primary care unit

Mendes Pontes Porto ME¹, Ximenes Ribeiro Lima R^{1,2,3}, Ximenes Lima MC¹, Farias Sobreira Bezerra FM²

¹Universidade de Fortaleza; ²Unidade Básica de Saúde - Roberto da Silva Bruno; ³Pre-feitura de Fortaleza

Introduction

Due to intense neuroplasticity, childhood is defined as a critical period for development and growth, therefore, it is the most appropriate time for stimulation and rehabilitation of children with development disorders. However, due to the service's great demand, distance from the service locations and errors in the referral process, treatment in specialized care centers is often delayed. A small number of basic health units in Fortaleza are set up with a multidisciplinary team promoting "intermediate" care for children until they reach advanced intervention in secondary or tertiary care.

Objectives

To report the experience of monitoring children in need of multidisciplinary intervention in a primary care unit.

Methods

This is an observational study conducted in a basic health unit in the city of Fortaleza, state of Ceará from January 2020 until nowadays. The multidisciplinary team is composed by 1 pediatrician, 1 physiotherapist, 1 occupational therapist and 2 psychologists. Children assisted in the child development department at the basic health unit from January 2020 until now, were included. The ones who abandoned the treatment during the same period, were excluded due to the impossibility of evaluating the results. The study contains the following aspects: age, gender, and response to the proposed treatment in the motor and communication domains of development.

Results

This resulted in a sample of 20 children, with ages varying from 6 months to 8 years. Among them, 14 boys and 6 girls with a median age of 45m and 34.5 months, respectively. Achievement of speech function was observed in 1 male participant after completing the age of 2. Improved speech function, reduced echolalia and increased dexterity in spontaneous search for communication with family members was reported in 8 of 14 boys and 3 of the 6 girls. All children achieved motor progress, especially the ones under one year old who showed improvement in cephalic support and axial tone. Those older than one year also experienced improvement in gross and fine motor coordination.

Conclusion

Despite the absence of a speech therapist on the team, the context of absences during the pandemic, and the visits being made only once a week, it was possible to observe significant improvements on the children assisted in this unfavorable social context. Therefore, the presence of "intermediate assistance" in basic health units benefits the children with difficult access to services.

P 38

Severe thromboembolic event in a healthy adolescent with Covid-19

Mazzara C¹, Corigliano T¹, Kottanattu L¹, Vanoni F¹, Özkartal T², Cristallo Lacalamita M³, Simonetti GD^{3,4}, Brazzola PL¹

¹Pediatric Institute of Southern Switzerland, Bellinzona, Switzerland; ²Department of Cardiology, Ospedale Regionale Bellinzona e Valli, Bellinzona, Switzerland; ³Institute of Imaging of Southern Switzerland, Bellinzona, Switzerland; ⁴University of Southern Switzerland, Lugano, Switzerland

Case description

A previous healthy 15-year-old boy presented in March 2020 to the pediatric emergency department because of pain in the right gluteal region during the last 3 days and acute swelling of the homolateral leg. Personal and family history were silent, especially for thromboembolic risk factors or events. Because of the lockdown strategy, the patient drastically changed his habits becoming suddenly sedentary. His father presented fever and cough in the last 6 days. On clinical examination he was tachycardic and the right leg was swollen, livid and painful. The leg's pulses were less palpable. Tanner stadium was 5. Magnetic resonance showed instead a deep venous thrombosis, extending from the renal veins to the iliac and leg's vessels. A pulmonary angio-CT was performed demonstrating the presence of bilateral pulmonary embolisms. A nasopharyngeal swab for SARS-CoV-2 was performed, confirming the positivity.

Subcutaneous anticoagulation treatment with Enoxaparin (80 mg q12h) was started and switched to oral anticoagulation with a vitamin K antagonist, after reaching the therapeutic window. The screening for acquired and hereditary thrombophilia resulted positive for Lupus anticoagulant (LA, 1.41 ratio), heterozygous mutation for the prothrombin gene (G20210A) and heterozygous mutation for the ATIII gene (SERPINC1).

Discussion

There is evidence that pediatric patients have an increased risk of developing antiphospholipid antibodies following viral infections. Several studies had reported hypercoagulability in patients with COVID-19 infection, with the need of prophylactic or therapeutic anticoagulation. At that time, there were no indication about prophylactic anticoagulation in pediatric patients. This case reports a pediatric patient with SARS-CoV-2 infection without respiratory symptoms associated with a severe thromboembolic event. Its etiology is most likely multifactorial with genetic factors playing a major role. The recognized hypercoagulability status related to SARS-CoV-2 infection and the sedentaryness caused by the pandemic “#stayhome” strategy could represent potential triggers for overt clinical manifestations of a genetic prothrombotic predisposition. In conclusion, in case of SARS-CoV-2 infection possible thromboembolic complications should be actively searched also in pediatric teen age patients.

P 39

Successful treatment with metreleptin in congenital generalized lipodystrophy

Heldt Katrin¹, Dintheer-ter Velde Anneco¹, l'Allemand Dagmar¹

¹Pediatric Endocrinology and Diabetology, Eastern Switzerland Children's Hospital St. Gallen, Switzerland

Introduction

Congenital generalized lipodystrophy (CGL) is a rare autosomal recessive disease, characterized by almost complete absence of adipose tissue, e.g. due to a defect in phospholipid biosynthesis, resulting in hypertriglyceridemia, severe insulin resistance and ectopic fat deposition. The main clinical features are near total absence of adipose tissue and muscular hypertrophy, accompanied by hyperphagia, acanthosis nigricans, hepatomegaly /hepatic steatosis and polycystic ovarian syndrome; other manifestations include splenomegaly, intellectual disability, hypertrophic cardiomyopathy and lytic bone-lesions. Severe metabolic complications like diabetes, pancreatitis, non-alcoholic steatohepatitis, cirrhosis of the liver and infertility often occur early. Deficiency of adipose tissue leads to low levels of circulating leptin, an adipokine mediating energy sufficiency.

Intervention and results

CGL type 1 in our patient is linked to a homozygous mutation in the AGPAT2 gene and was diagnosed at the age of 4 weeks thanks to the typical subtotal loss of subcutaneous adipose tissue, muscular appearance, eruptive xanthomas of the skin and hepatomegaly. Biochemically, a massive hypertriglyceridemia, insulin resistance and decreased leptin level were found. Ultrasound revealed hepatic steatosis, slight splenomegaly, ovarian hypertrophy and a cardiac septal hypertrophy. We started a modified low-fat diet with 25% total energy content from fat, which led to a reduction in triglyceride level, insulin resistance, hepatic steatosis and ovarian hypertrophy. With good adherence and tolerability of the diet, the fat content could be increased to 35% of the total energy over the course under restriction of fructose and simple sugars, with slight but tolerable hypertriglyceridemia. Due to the persistence of pronounced leptin deficiency, we started an additional therapy with subcutaneous metreleptin at the age of 2.6 years as suggested. As a result, triglyceride and insulin levels as well as the sizes of liver, cardiac septum and ovaries normalized.

Conclusions

Hormone replacement therapy with metreleptin in our patient with CGL was well tolerated and resulted in sustained improvements in metabolic parameters like hypertriglyceridemia and insulin resistance but even normalization of hepatic steatosis, liver size and cardiac septum volume. This reduces the risk of acute exacerbations such as pancreatitis and long-term metabolic complications.

P 40

Case report: acute upper airway obstruction in children

Della Franca M¹, Pescia L¹, Calciolari J¹, Pasquale MF¹, Ferrucci E¹

¹Service of Paediatrics, Paediatrics Institute of Southern Switzerland, Ospedale Regionale di Lugano, EOC, Lugano, Switzerland

A 4 y.o. boy presented to E.R. for suspected foreign body (FB) inhalation. He was playing with some Legos when he suddenly started to cough and to complain about a persistent discomfort at the supra-jugular notch level. Upon arrival at the E.R. his conditions were stable: the respiratory status showed no particular signs except for a mild stridor. Plain chest x-rays revealed no FB but showed a relevant narrowing of the upper airways. Suddenly, a few minutes later, the child started having vigorous cough followed by an episode of vomit and the appearance of respiratory distress, with no evidence of haemodynamic decompensation. An emergency bronchoscopy was thus performed which identified a significant laryngeal oedema. Airway patency was ensured by orotracheal intubation, which was maintained for the next 48 hours. In the following two days, a fleeting rash appeared on the whole body and further laboratory investigations isolated two different viruses in the tracheal secretions (Coronavirus NL63 and Rhino-Enterovirus). A second bronchoscopy confirmed the actual absence of any FB. Upper airway obstruction is a frequent occurrence in paediatrics. For their anatomical structure, children are vulnerable to it and life-threatening cases are not rare. Therefore, regardless of the initial degree of obstruction, rapid assessment and careful monitoring of clinical evolution is necessary. The present case offers the opportunity to discuss about all the possible aetiologies underlying acute upper airway obstruction in paediatric population, whose presentation can be so variable that they may represent a real medical challenge. Aspiration of foreign bodies is one of the most frequent causes in children younger than 5 years of age. If infectious symptoms are present, the clinical suspicion mainly relies on viral or bacterial infections affecting the upper airways (croup, epiglottitis, laryngotracheitis) which can be real emergencies. Anaphylactic reactions may be severe when oedema involves the retro-pharynx or the larynx. Symptoms onset in these cases is usually sudden, and other clinical signs such as urticaria may be associated. Hereditary angioedema should be suspected in patients with progressive upper airway oedema not responsive to intramuscular epinephrine. Contusive, penetrating or burning injuries can directly or indirectly involve the upper airways causing rapid obstruction for which prompt evaluation and airway protection is necessary.

P 41

Multiple cutaneous abscesses due to *Pseudomonas aeruginosa* in a six-month-old girl

Dupriez Ch¹, Creuven M², Bernier V¹

¹Sector of Pediatric Dermatology, Department of Pediatrics, CHC-Liège, Belgium; ²Sector of Infectious Pediatrics, Department of Pediatrics, CHC-Liège, Belgium

Pseudomonas aeruginosa (PA) is rarely encountered in skin lesions in children, except in ecthyma gangrenosum or bathtub folliculitis. To our knowledge, multiple cutaneous abscesses in a healthy infant have never been reported in the literature so far.

We report the case of a 6-month-old girl with a 3-day course of fever and bronchitis. She presented with three erythematous cutaneous nodules on the right arm and the left leg. A blood analysis showed a C-Reactive Protein at 177.1 mg/L with a hyperleukocytosis. Rapidly the nodules reached the number of eight and a systemic antibiotherapy was started with intramuscular ceftriaxone first due to lack of easy venous access and then intravenous amoxicilline plus clavulanic acid. A neutropenia appeared while the CRP level decreased. In the following days, the nodules evolved into abscesses. A biopsy of a nodule showed panniculitis with pus, leukocytes infiltrate and the culture grew full of PA. Surgical punctures of the other abscesses also drained off pus with cultured PA. The child went rapidly well. The plastic bathtub, the tap, the aerosol mask and connector, and the « Babycook » device were tested negative for

PA. Her twin brother and the family had no cutaneous lesion. Systemic antibiotics were discontinued and oral ciprofloxacin was started during two weeks. Repeated blood cultures remained negative. An extensive work-up did not reveal a deep infectious source (heart, abdomen, bone). The humoral and cellular immunity was normal. The child experienced a progressive decrease of the abscesses with and after antibiotics.

Nodular panniculitis due to PA is a rare condition. Differential diagnosis with erythema nodosum or other causes of panniculitis may be challenging before evolution to abscess. A positive bacteremia, an infected skin wound or contaminated liquid containing devices are often considered the origin of the cutaneous lesions.

This report aims to answer questions such as the source of the contamination, the usefulness of the antibiotics in the treatment, the need for surgical drainage, and the link between leukopenia and infection or immunodeficiency.

P 42

Plant-based nutrition in adolescents: first results from a focus group study

Stritter W¹, Ngoumou G¹, Seifert G¹

¹Department of Pediatric Oncology and Hematology, Charité Universitätsmedizin Berlin, Germany

Background

Plant-based diets are becoming increasingly popular amongst adolescents. At the same time environmental protection and animal welfare are central issues for young people, not just because of the Friday for Future movement.

Question: What is the main motivation for young people to change their diet towards a plant-based nutrition?

Methods

We conducted three focus groups with adolescents and young adults (aged 14–21), with female, male and mixed gender participants respectively. Main themes of the focus group discussions were: reasons or cause for diet change, challenges in and motivations for maintaining a plant-based diet and attitudes toward healthy nutrition. The recordings were transcribed verbatim and analyzed according to thematic analysis.

Results

Analysis is still ongoing. First results indicate that young people mainly use environmental and animal protection reasons to justify the change in diet. They wish for background knowledge about nutrition to be integrated into school lessons, particularly with a view to animal welfare, the environment and health. The need for reliable information for a better orientation in the large network of circulating information was emphasized.

Conclusion

Since more and more adolescents are changing their diet toward a plant-based nutrition there is a great need for age-appropriate information about balanced and healthy plant-based nutrition as well as concepts to bring this information to the target groups.

P 43

From Influenza A to cardiac arrest in two days

Nagel C¹, Brotschi B², Binz N¹

¹Department of paediatric intensive care, Kantonsspital Graubünden, Chur, Switzerland;

²Department of paediatric intensive care, University Children's Hospital Zürich, Switzerland

Introduction

Whilst Influenza A virus infections (IAVI) are common, renal complications of IAVI are uncommon and its pathogenesis underlying renal injuries has not been delineated. Here, we describe a previously healthy patient without previous risk factors for severe complications who developed rhabdomyolysis induced renal injury and subsequent cardiac arrest.

Case

A six year old female patient was hospitalized with myositis due to an IAVI with a creatinine kinase (CK) level of 19'195 U/L. After initial improvement of her clinical condition, she developed dark brown oliguria, increasing muscle pain and cold mottled limbs on the second day. The next day she presented in our emergency unit in a septic shock like clinical condition.

Upon clinical hypothesis of septic shock, fluids and ceftriaxone were administered and rapid sequence intubation under catecholamines and further fluid

resuscitation was performed. Laboratory showed a hyperkalemia, hyponatremia and CK level of 408'800 U/l.

Arrhythmia due to hyperkalemia resulted in pulseless ventricular arrhythmia four times; return of spontaneous circulation was each time achieved after defibrillation under mechanical and pharmacological resuscitation.

As hyperkalemia persisted despite maximally escalated medication, the patient was transferred to a tertiary care hospital with possibility of hemodiafiltration.

Continuous renal replacement therapy, fasciotomy due to compartment syndrome and multimodal long time rehabilitation (PTSD, muscle atrophy, renal recovery, ect.) led to minor neurocognitive deficits with near complete recovery of activities of daily living by orthotic support of the lower limbs.

Discussion

Data on complication rates of IAVI are scarce, but most cases of IAVI have benign outcome.

Renal complications of IAVI are uncommon, but have been described and include acute kidney injury, rhabdomyolysis, haemolytic uremic syndrome, acute glomerulonephritis, disseminated intravascular coagulation amongst others.

The case of our patient is remarkable because the patient developed rhabdomyolysis and subsequently renal failure without any prior risk factors.

Summary

Rhabdomyolysis and renal injury combined with septic shock led to cardiac arrest in a previously healthy child with IAVI requiring cardiopulmonary resuscitation. The outcome in our patient was considerably good given the initial deterioration.

Conclusion

Children with IAVI and significant myositis should be continuously monitored for severe complications.

P 44

A rare case of vascular thoracic outlet syndrome in a young athlete

Capello Mainardi C¹, Paolucci L¹, Corigliano T¹, Queirolo S¹, Brazzola P², Prouse G³

¹Department of Paediatrics, Paediatric Institute of Italian Switzerland (IPSI), Ospedale Civico of Lugano, EOC, Lugano, Switzerland; ²Department of Paediatrics and paediatric Oncology and Haematology, Paediatric Institute of Italian Switzerland (IPSI), Ospedale San Giovanni, EOC, Bellinzona, Switzerland; ³Department of Vascular Surgery, Ospedale Civico of Lugano, EOC, Lugano, Switzerland

A 15 y.o. patient presented to the E.R. with left shoulder pain started 48 hours before, after vigorous upper limb exercises, without any previous traumatic injury. Clinically, his left arm appeared bluish in color and slightly edematous compared to the contralateral one. The limb range of movement was not impeded, but he complained about tingling sensation and pain at the pectoralis maximum insertion. In suspicion of a thrombotic event, a Duplex US was performed identifying a deep vein thrombosis of both left axillary and subclavian veins. Laboratory tests showed no remarkable abnormalities except for an elevation of D-dimers (1.14 mg/L, URL 0.5 mg/L). A nasopharyngeal swab for SARS-CoV-2 was also performed, considering the most recent case reports about SARS-CoV2-induced venous thrombosis, but it was negative. Intravenous heparin was promptly initiated and then a percutaneous thrombus aspiration was successfully performed. However, during a control Duplex US the following day, a relapse was found with reformation of an equally extended thrombus. Following another complete thrombus aspiration, the patient underwent a second relapse. The localization of the venous thrombosis and its two consecutive relapses in a young, healthy patient without past or family history of cardiovascular risks and a normal thrombophilic screening appeared very unusual. Therefore, an MRI angiography with provocative test was performed, showing the presence of thoracic outlet stenosis compressing the left subclavian vein at the crossing point between clavicle and 1 rib. A diagnosis of vascular thoracic outlet syndrome (also called Paget Schroetter syndrome or effort thrombosis) was thus made, and the patient was discharged with an anticoagulation therapy with rivaroxaban. During follow-up no complications were described. In addition, to prevent relapses after anticoagulation suspension, surgical decompression was performed with 1 left rib resection and intra-procedural angiography.

In conclusion, different etiological scenarios may cause shoulder pain in a young athlete, especially if upper extremity exercises are performed vigor-

ously and repetitively. A thoracic outlet syndrome, especially its vascular subtype, should be considered among the differential diagnosis despite its incidence of <1-2/100000 per year. In fact, its early diagnosis is fundamental to promptly initiate treatment thus offering a chance of recanalization and avoiding post-thrombotic syndrome.

P 45

A chronic nephropathy preceded by an episode of IgA vasculitis and acute kidney injury after ASLO-positive pharyngitis. Is there a link?

Cufi i Prat M¹, Zoubir S A¹, Hopfer H², Goischke A³

¹Service de pédiatrie, Hôpital du Jura, site de Delémont; ²Universitätsspital Basel, Pathologie, Basel; ³Universitäts-Kinderspital beider Basel, Pädiatrische Nephrologie, Basel

Background

Clinical patterns in pediatric nephrology often correspond to specific kidney or systemic diseases. A diagnosis becomes challenging if two common patterns overlap in time and the renal biopsy does not match with neither expected disease.

Case report

A previously healthy vaccinated 9-year-old girl was admitted twice to our pediatric service. During the first admission, a clinical diagnosis of non-complicated IgA vasculitis (Purpura Schönlein Henoch) was given and the girl was discharged with renal function follow-up. One month later, she was re-admitted in the context of acute renal failure suspected to be due to a post-streptococcal glomerulonephritis (PSGN). The laboratory investigations confirmed low C3 and high ASLO. In the course, a nephrotic syndrome developed with subsequent worsening of renal function, which led us to perform a kidney biopsy.

The biopsy revealed a diffuse proliferative immune complex glomerulonephritis (GN) with co-dominant polyclonal IgA, IgG3 and C3c deposits by immunofluorescence and structured deposits by electron microscopy.

On RAAS (renin-angiotensin-aldosterone system) blockade, the girl showed only partial remission of proteinuria on RAAS blocker, but renal function improved quickly. In additional laboratory tests performed after the renal biopsy, cryoglobulinemia type III and a questionable C3Nef were detected.

Discussion

Surprisingly, the kidney biopsy performed because of the unfavourable clinical course neither showed the typical morphological pattern of IgA vasculitis nor PSGN. The leading differential diagnosis was an IgA-dominant infectious GN, a diagnosis well established in the adult population (Satoskar et al, CJASN 6:1179-1186). Type III cryoglobulins have been described in some of these patients. If a genetic or autoimmune dysregulation of the complement system contributes to the protracted course in this case, is still under investigation.

Conclusion

Diagnoses of common kidney diseases during childhood are frequently given clinically, especially if presentation and additional laboratory investigations are congruent with the diagnostic hypothesis. Nevertheless, a broader differential diagnosis is important in patients with an unexpected course. A diagnostic kidney biopsy will help to reach a diagnosis.

P 46

Successful conservative management of dural sinus malformation in a newborn

Piffer A¹, Cristallo Lacalamita M², Goeggel Simonetti B¹, Delcò Volontè C¹

¹Istituto Pediatrico della Svizzera Italiana, San Giovanni Hospital, Bellinzona, Switzerland; ²Istituto di Imaging Svizzera italiana, San Giovanni Hospital, Bellinzona, Switzerland

Background

Dural sinus malformation (DSM) is a rare congenital condition of unknown etiology characterized by a substantial enlargement of one of the dural venous sinuses.

Case

We present the case of a male infant born at 34 weeks of gestation after an uneventful pregnancy who was admitted to our neonatal unit for CPAP treatment due to a neonatal respiratory distress syndrome (wet lung). Birth weight was 2075 g (25-75thP), length 46.5cm (50thP) and head circumference 29 cm (3-10thP). Apgar scores were 6/6/6 and arterial and venous umbilical cord pH values of 7.22 and 7.34. Apart from an occipital bossing of the skull, the neurological examination was normal. Family history was unremarkable.

On the 4th DOL (Day Of Life), in the context of hyperbilirubinemia, a blood count showed thrombocytopenia (86 G/L thrombocytes). Cranial ultrasound revealed a hypoechogenic lesion in the posterior fossa with local mass effect, but no signs of hydrocephalus. MRI showed a posterior extra-axial mass with a large thrombosed venous lake involving the torcular Herophili. This lesion had a mass effect with anterior dislocation of the cerebellum and the brainstem. Imaging was compatible with the diagnosis of a thrombosed congenital dural sinus malformation (DSM). Blood coagulation test were normal. The patient was transferred to a tertiary care center where a contrast enhanced MR-angiography showed no signs of an AV fistula.

On DOL 5 full therapeutic anticoagulation was started with i.v. heparine and four substitutions of antithrombin III. On DOL 18 the treatment was switched to low molecular weight heparin. Ten days after the beginning of full heparinisation (DOL 15), a control cMRI demonstrated incipient thrombus organisation.

Further laboratory analysis did not reveal an underlying coagulation disorders (antithrombin III, factor X, proteine C and S), negative maternal anticardiolipin antibodies, lupus anticoagulants, ac-beta2-microglobulin).

At present, the baby is 3 months old and shows a normal neurological development.

Conclusion

DSM is a rare disease with high mortality. It may be diagnosed antenatally via ultrasound and/or MRI, or during first months of life presenting with macrocephaly or neurological symptoms. We diagnosed DSM in our patient due to thrombocytopenia. In this case, conservative management with regular neuropaediatric follow-up visits and imaging has so far been successful.

P 47

Perimenstrual asthma: case report of a pattern not to be underestimated

Ridolfi A¹, Pesca L G A¹, Zanolari M¹, Ferrucci E¹

¹Istituto Pediatrico della Svizzera Italiana, Ente Ospedaliero Cantonale, Switzerland

Background

“Global Initiative for Asthma” affirms that under the age of 14, asthma is more common in boys than in girls but after this age, the epidemiology is reversed and asthma becomes more frequent in women. The prevalence of asthma is about 9.7% in women and 5.5% in men. It is estimated that between 19% and 40% of asthmatic women in childbearing age may experience a cyclical clinical deterioration during the luteal phase and/or during the first days of menstruation. The impact of female steroid sex hormones on the function of the respiratory system and inflammation in the bronchi may play a central role in this phenomenon, known as perimenstrual asthma (PMA). Although it is a quite frequent event, there are not many studies in the literature. The number of studies decreases if the paediatric age is considered. Case study: We report about a 16-year-old girl, with a history of allergic asthma. After puberty, she developed asthma exacerbations related with menstruation. Dysmenorrhea and difficult control of the asthmatic symptoms were recorded since the menarche (age of 10). Since the age of 10 to 16 years old, she came to the emergency four times for asthma attacks that were not manageable at home. She was treated with a controller therapy combined with Formoterol and Fluticasone (500ug/day), enhanced with Montelukast, although this regularly had to resort to reliver therapy with Salbutamol and systemic steroids. A complete, detailed diary of the crisis kept by the patient, shows that every month she has a similar clinic seven days before the menstruation. A PMA was then supposed, and a oestrogenic-progestinal therapy was started, with complete disappearance of dysmenorrhoea and an almost complete decline of the crises of Asthma.

Conclusion

PMA has often a more severe course than other asthma phenotypes, and the achieving and maintaining control over the symptoms is more difficult than in non-PMA asthma. In young women of childbearing age, with frequent asthma attacks without clear trigger factors, it's worth keeping a diary of crises. This in order to study a possible correlation with hormone fluctuations of the late luteal phase and thus to diagnose a PMA. In these cases, especially when a high dose therapy is ineffective, it is suitable to implement a oestrogenic-progestinal therapy. In most cases, it leads to a much better outcome.

INDEX OF FIRST AUTHORS

The numbers refer to the numbers of the abstracts.

Andrade Borges Stéphanie P 21
 Andrea Felser P 4
 Ardura-Garcia Cristina SPN 6, SPN 7

 Bahadori Atessa P 6
 Beck Samuel P 29
 Beiner Matthias FM 12
 Buettcher Michael FM 14

 Capello Mainardi Carlotta SPN 17, P 44
 Colombo Ron P 22
 Crédeville Mathilde P 24
 Cufi i Prat Meritxell P 45

 Danioth Laura Patricia P 34
 Della Franca Michela P 40
 Deml Michael FM 3
 Dosso Leila P 20
 Dubail Mathilde P 3
 Dupriez Charlotte P 41
 Dusoczky Nicole P 31

 Ehrler Melanie FM 17

 Fillistorf Laura P 30
 Fracheboud Noémie P 19
 Fux Michaela SPN 5

 Gonza Ngoumou FM 7
 Gummy-Pause Fabienne SPN 4

 Heldt Katrin P 39
 Hertli Muriel P 18
 Hofer Seline FM 6

 Karim Ait P 10, P 11
 Keller Michèle FM 5
 Klöti Simon SPN 12
 König Naoko FM 9

 Lucas Ramanathan Pia FM 4

 Mallet Maria Christina P 14
 Marangoni Alice Serena P 28
 Mazzara Calogero P 36, P 38
 Moussaoui Dehlia SPN 2
 Müller Loretta SPN 15

 Naef Nadja FM 10
 Nagel Christina P 43

 Ogal Mercedes FM 2

 Papasava Dimitra P 17
 Papathanasiou Terzi Matthildi Athina SPN 8
 Perrin Anne FM 8
 Peter Anna Maria P 9
 Petter Marie P 16
 Piffer Arianna P 46
 Piletta-Zanin Alexandre P 5
 Porto Maria Eduarda P 37

 Ramelli Gian Paolo SPN 13
 Ridolfi Andrea P 47
 Rimle Carole P 27
 Rosenblatt Lara P 32
 Rouge Elton Paola Andrea P 25

 Sahyoun Cyril SPN 3
 Sarkisian Jessica P 7
 Schaffner Thierry Oliver FM 15
 Schaub Barbara P 26
 Schmid Thurneysen Lisa SPN 11
 Schneider Nadja FM 1
 Schuler Mirjam FM 18
 Schwendener Corina Ladina P 12
 Sebti Moustapha P 13
 Stebler Anita FM 13
 Stelling Lisa P 1
 Sternberg Julie P 2
 Strebel Sven SPN 9
 Stritter Wiebke P 42

 Tosetti Lorenzo P 15

 Tröndle Marc P 35
 Uka Anita SPN 16
 Ursprung Pascale Seline SPN 10

 van Ransbeeck Andrea P 33
 Varatharaju Vysnave SPN 14
 Vasey Léa P 23
 von Rhein Michael FM 11, FM 16, P 8
 Vural Serpil SPN 18

SWISS MEDICAL WEEKLY

Editorial board:

Prof. Adriano Aguzzi (editor in chief)
Prof. Manuel Battegay (deputy editor in chief)
Academic editors: see www.smw.ch

Managing editors: Natalie Marty, MD,
and Jan Roth, MD

Guidelines for authors and online submission:
www.smw.ch

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
Phone +41 61 467 85 55
office@smw.ch

ISSN online supplement: 2504-1622

© Swiss Medical Weekly Supporting Association,
2021.

Swiss Medical Weekly is an open access publication. Accordingly, SMW grants to all users on the basis of the Creative Commons license "Attribution – Non commercial – No Derivative Works" for an unlimited period the right to copy, distribute, display, and perform the work as well as to make it publicly available on condition that (1) the work is clearly attributed to the author or licensor (2) the work is not used for commercial purposes and (3) the work is not altered, transformed, or built upon. Any use of the work for commercial purposes needs the explicit prior authorisation on the basis of a written agreement.

Cover image: © Christoph Frei, Bern