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Abstracts

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ABSTRACTS OF THE CANCELLED ANNUAL MEETING 2020

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Neonatal thrombocytopenia: a retrospective study of 120 cases

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Introduction: Neonatal thrombocytopenia (NT) is the most common neonatal hemostasis abnormality. It is defined by a blood platelet count of less than 150,000 / mm^3. This condition is sometimes a source of neurosensory sequelae in cases of intracranial hemorrhage. The objective of this study is to analyze the epidemiological, clinical, etiological, therapeutic and evolutionary aspects of NT.

Materials and methods: Retrospective descriptive and analytical study of all NT files, collected within our neonatal department, during a two-year period from 01 January 2017 to 31 December 2018. Included was any newborn with confirmed thrombocytopenia. All the files were analyzed using an exploitation sheet.

Results: During the study period, 3000 newborns were hospitalized, 120 of them presented a NT, which represents 4% of hospitalization. 70 cases were male with a sex ratio = 1.4. 60% came from a premature delivery. The age of onset of thrombocytopenia was predominant in the first week of life (n = 108, 90%) . 57 cases had low birth weight. 52.5% were eutrophic. Respiratory distress, prematurity and jaundice were the main reasons for hospitalization. 91 cases did not present hemorrhagic symptomatology. 48 cases had moderate thrombocytopenia and 29 cases had severe thrombocytopenia. The etiologies were dominated by neonatal infection in 61 cases, the autoimmune origin in 20 cases, 17 cases of feto-maternal erythrocyte incompatibility and two cases of genetic origin. 74 cases had received antibiotic therapy, 20 cases received a platelet transfusion, 18 cases received immunoglobulins and no treatment in 8 cases. The mortality rate was 15% with 18 deaths among the 120 cases included.

Conclusion: NT is often asymptomatic and discovered during an assessment in the context of another disease. Several factors can be at its origin antenatal, perinatal or postnatal. A multidisciplinary collaboration between the obstetrical team, the pediatrician and the hematologist is essential for an etiological and preventive approach as well as for early management

2

Euphrasia eye drops as a treatment option in preterm neonates with first signs of congenital nasolacrimal duct obstruction: a randomized double-blind placebo-controlled trial

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Aim: To investigate whether the early administration of Euphrasia eye drops® in preterm neonates presenting with ocular discharge with or without tearing and reddened eye fosters the resolution of the ocular discharge and reduces the need for topical antibiotic therapy, as compared to placebo.

Methods: We conducted a randomized double-blind placebo-controlled trial at the University Children's Hospital Bern, Switzerland, between May 2011 and December 2016. Preterm neonates with first signs of congestion of the nasolacrimal duct, defined as white, yellow, or green ocular discharge with or without tearing and reddened eye were included. Infants were randomly assigned (1:1) to the Euphrasia arm (Euphrasia eye drops®, Weleda AG, Arlesheim) or the placebo arm (NaCl 0.9%). Euphrasia or placebo was administrated at a dose of one drop in each eye four times a day over a period of 96 hours. The primary outcome was the treatment success, defined as no ocular discharge at 96 hours and no use of topical antibiotic therapy during the 96-hour intervention.

Results: A total of 114 neonates were screened and 84 were randomized. Among neonates in the Euphrasia arm, 22 (55.0%) achieved our primary outcome compared to 21 (51.2%) in the placebo arm (p = 0.85). In the Euphrasia arm, time to resolution of reddening tended to fall within the shorter bracket of 24 to 48 hours (24 (92.3%) versus 12 (80.0%) in the placebo arm, p = 0.34) and relapse or first signs of reddening during the 96-hour intervention tended to be lower (3 (7.9%) eyes versus 8 (18.2%) eyes in the placebo arm, p = 0.17). Tearing at 96 hours tended to be lower in the Euphrasia arm (5 (12.8%) eyes in the Euphrasia arm versus 12 (27.3%) eyes in the placebo arm, p = 0.10).

Discussion: Euphrasia did not significantly improve treatment success, defined as no ocular discharge at 96 hours and no use of topical antibiotic therapy during the 96-hour intervention. However, results suggest that Euphrasia may be of benefit for symptoms such as reddening and tearing, and thus improve the comfort of patients.

3

Vaccine hesitancy and under-immunization in Switzerland: A National Research Program comparing perspectives of providers and consumers of biomedicine and complementary and alternative medicine (CAM)

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Background and aims: Complementary and alternative medicine (CAM) is associated with vaccine critical attitudes in research and public opinion. CAM is integrated into the Swiss healthcare system, with certain CAM services reimbursed by basic mandatory health insurance when offered by biomedical doctors with additional postgraduate training in CAM. 25-50% of the Swiss population report CAM use and favorable attitudes. We therefore explored vaccine hesitancy (VH) by describing interactions between patients and providers of CAM/biomedicine.

Methods: We conducted qualitative interviews with 17 CAM providers, 20 biomedical doctors, and 30 parents. We observed 34 vaccination consultations (N = 18 CAM consultations; N = 16 biomedical consultations) and wrote narrative notes.

Results: As we have shown (Deml et al. 2019, Social Science & Medicine), the majority of CAM providers were not categorically opposed to vaccination and expressed ambivalent/favorable vaccination attitudes. Most framed vaccination decisions as choices at individual/family levels rather than focusing on public health. Biomedical doctors described difficult consultations with VH patients, describing them as "problem patients." Some parents described biomedicine and health authorities as being influenced by financial interests and criticized one-size-fits-all approaches. VH parents proactively sought out the "truth" and perceived CAM providers as offering "neutral" information. Parents emphasized the importance of trust, affect, and choice, with many explaining how trust was a prerequisite for productive vaccination consultations.

Conclusions: Our research suggests that healthcare professionals need support in addressing VH and might benefit from increased understanding of VH parents' rationales, particularly the importance parents attach to trust, affect, and choice. Parents generally preferred consultation approaches incorporating parents' vaccination knowledge/views and social/health contexts. These preferences aligned with CAM provider approaches, suggesting that biomedical providers could learn from CAM provider communication approaches in clinical encounters with VH parents.

4

Prevalence of complementary medicine use by pediatric oncology patients at different time intervals

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Background: the prevalence and modalities of complementary medicine (CM) use by pediatric oncology patients vary widely across studies, and changes in CM use over the course of treatment is understudied. Thus, this study aims to explore 1) CM use by pediatric oncology patients in relation to specific time intervals and 2) communication on CM use between parents of patient and pediatric oncologists.

Procedure: a retrospective cross-sectional study conducted among parents of children diagnosed with cancer at a Swiss pediatric hematology-oncology centre, by means of an online questionnaire. Questions were related to their child's CM use over 5 different time intervals; to sources of information about CM use; and to communication with the oncologists.

Results: the study included 140 participants. CM were used before the diagnosis by 54.3% of patients and 69.3% since the diagnosis. During each defined time intervals, between 50-60% of the patients used at least one CM. Homeopathy was the most popular CM modality used during the oncology treatment (27.1%), the first year after (33.9%) and between 1 to 5 years after the end of the oncology treatment (36.3%), and osteopathy was the most popular 5 years after the end of the oncology treatment or after (28.0%). If patients used CM before the diagnosis, they tended to continue to use them during the oncology treatment as well as 1 year after the end of the oncology treatment. Forty percent of participants did not discuss CM with their pediatric oncologist and 60% of CM users disclosed CM use to them. Among participants discussing CM use with their oncologist, more than 80% stated that they initiated discussion about CM use. Would the oncologist propose CM, 74.3% of participants would try it for their child.

Conclusion: The high prevalence of CM use and the different trend of use during the oncology care pathway and after underlines the need to increase communication on CM in the pediatric oncology settings, notably on benefits and interaction risks with oncology treatment. Furthermore, physicians' information and training on CM is required to improve communication.

5

The challenge to measure disease-specific QoL in rare disease; Results: from a prospective cohort study of patients with Hirschsprung disease

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Introduction: Measuring quality of life (QoL) adequately in rare diseases remains a challenge. The aim of this study is to investigate the functional outcome and the disease-specific QoL in patients with Hirschsprung disease (HSCR) with determination of its predictive factors.

Methods: Prospective transversal and case-control study in a French-Canadian cohort of HSCR patients. The questionnaires 'HAQL' (disease-specific QoL), 'PedsQL' (global health related QoL), 'Stressful life events', a questionnaire about the socio-economic state and a stool diary were requested to fill in. Hair cortisol concentration (HCC) (measure of chronic stress) was measured using an enzyme-linked immunosorbent assay kit (ELISA).

Results: 72 patients (52 males, 72%) and 117 controls (76 males, 65%) were included in the final analysis. Median [IQR1, IQR3] age at study inclusion was 12.1 years [8,17.5] in patients and 12.6 years [10.2,15.1] in controls. The mean scores of global health related QoL were comparable between patients and controls (toddlers 5 to 7 years old: 77.1/100 vs 85.7/100, p = 0.09; children 8 to 12 years old: 86/100 vs 81.6/100, p = 0.3; adolescents 13 to 18 years old: 79.9/100 vs 82/100, p = 0.5). Disease-specific score of QoL was low in children (mean score 539.3± 66.5/700), with the dimension 'Fecal continence during daytime' as the most affected one (mean score 52.6± 25.3/100). 'Physical functioning' was the most affected dimension in adolescents (mean score 74.2± 16.1/100). Prevalence of fecal incontinence/soiling was 85% in children, 40% in adolescents and 22% in adults. Nocturnal fecal incontinence was associated with poorer disease-specific QoL (coefficient = -0.11, p = 0.03). No association was seen between presence of fecal soiling/incontinence and HCC (p = 0.88).

Conclusion: The 'PedsQL'Global health related QoL questionnaire seems not to be specific enough to conclude adequately on QoL in patients with HSCR. Disease-specific QoL investigation is mandatory, to recognize the real burden of disease on QoL and might be an important step to improve patient's care.

6

Prevalence of tuberculosis in migrant children in Switzerland and relevance of current screening guidelines

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Aims: Since 2016, Swiss guidelines recommend to screen all migrant children <5 years old for tuberculosis (TB) and to screen older children only if they have risk factors for TB. Our goals were to describe the epidemiology of latent tuberculosis (LTBI) in migrant children at the Lausanne University Hospital, to identify determinants of LTBI and tuberculosis disease (TBD), and to evaluate the risk of a false-positive tuberculin skin test (TST) when using a positivity limit of 5 mm.

Methods: Newly arrived migrant children 0–18 years old were prospectively enrolled from 31.08.2015–31.08.2017. Every migrant child was assessed for the risk of TB exposure and TBD and was administered a TST. A TB-spot test was performed in children ≥ 5 years old when the TST was positive. Children with clinical and/or radiological signs of TBD were further investigated. Children ≥ 5 years of age with a positive TB-spot test and children <5 years of age with a positive TST, without clinico-radiological signs of TBD received a diagnosis of LTBI. A false-positive TST result was diagnosed in children ≥ 5 years old when the TB-spot test was negative. Potential determinants of LTBI, TBD and false-positive TSTs were identified.

Results: 253 patients were eligible. Median age of the patients was 8.1 years (IQR 4.5–12.8) and 104 (41%) were female. 24% (62/253) came from a country with a moderate–high incidence of TBD (≥ 80 cases per 100,000 individuals). 28 patients (11%) had positive TSTs, and TB was confirmed in 17 (6.7%) of these patients (16 with LTBI, 1 with TBD). A moderate–high incidence of TBD in the country of origin (aOR 18.8, 95%CI 5.1–68.6, p <0.001), older age (aOR 1.1, 95%CI 1.0–1.3, p = 0.025), and contact with a TBD patient (aOR = 8, 95%CI 1.8–36.2, p = 0.007) were associated with a diagnosis of TB, on multivariate analysis. Among the 23 children over 5 years of age who had a positive TST with measurement available, a measure between 5-9 mm was more frequent in case of a false-positive TST (5/9, 56% vs 0/14, 0%, p = 0.002). BCG vaccination was the only predictor of a false-positive TST (p = 0.03).

Conclusion: Screening migrant children ≥ 5 years old for TB could confer a public health benefit even in the absence of other risk factors. The limit of TST positivity could be raised from ≥ 5 mm to ≥ 10 mm to decrease the rate of false-positive results. A national assessment of migrant children between the ages of 5 and 15 should be carried out to confirm our findings.

7

Non-pharmacological home remedies in pediatrics: a cross-sectional study

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Introduction: Non-pharmacological home remedies (defined here as traditional remedies that cannot be obtained in a commercially available drug formulation and that do not require help from a therapist) are frequently used alongside conventional medicine. They are often composed of simple ingredients present in the household. For benign symptoms, they have the potential to reduce urgent consultations. The aim of this study was to identify the range of home remedies proposed by primary care pediatricians in their practice, as well as those used by parents of pediatric patients in Geneva.

Methods: This was a cross sectional study in two parts: 1.) Postal survey addressed to all primary care pediatricians in the canton of Geneva; 2.) Direct survey of parents of patients present in the waiting room of the pediatric emergency department of Geneva University Hospitals. The survey comprised a list of home remedies compiled through a review of the literature and discussions with experienced primary care pediatricians. Participants were asked to indicate whether they proposed / used these remedies. They could also add remedies that were not in the list. Data were transferred to a Qualtrics interface and analyzed in a descriptive way.

Results: 68 pediatricians (response rate: 46%) et 100 parents completed the surveys. The most common remedies were nearly identical in both populations. For example: salt water instillations to treat common colds were proposed by 77% pediatricians and used by 46% parents; black tea pads for viral conjunctivitis proposed by 66% pediatricians, and used by 17% parents; honey to relieve cough was proposed by 48% pediatricians and 49% parents. Potentially dangerous remedies (for example boiled garlic cataplasm for earache) were not commonly used.

Conclusion: Home remedies are commonly recommended by pediatricians and often used in the population. Documenting this in the consultation is important so that pediatricians can identify and advise about both safe and unsafe use of these remedies, and discuss interesting alternatives to pharmaceutical treatments in everyday pediatric practice.

8

Presentation, causes and disease duration of Lipschütz's acute vulvar ulcer: a systematic review

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Objectives: In previously healthy subjects, vulvar ulcers are mostly caused by sexually transmitted microorganisms. Lipschütz's acute vulvar ulceration, first reported in 1912, is a non-sexually acquired condition characterized by sudden onset of a few genital ulcers. The aim of this analysis was to investigate presentation, underlying causes and disease duration.

Methods: A systematic review of reports including cases of apparently previously healthy females affected by Lipschütz's ulceration was performed in Excerpta Medica, National Library of Medicine and Web of Science.

Results: The literature search disclosed 152 cases published after 1965. Almost 90 percent of the cases were ≤20 years of age (N = 136) and sexually inactive (N = 129). Lesions were usually one to about three, painful, ≥10 mm large, well-delimited, with a fibrinous, necrotic or purulent center and sometimes a symmetric vulvar distribution. Voiding disorders and enlarged inguinal lymph nodes were observed in a large subset of cases. Canker sores were noted in 10% of patients. Lipschütz's vulvar ulceration occurred concomitantly with an infectious disease in 135 cases: a flu-like syndrome in 79 and a well-defined infection in 56 cases. Infectious mononucleosis syndrome (N = 39) was the most frequently detected well-defined infection, followed by mycoplasma species infections (N = 11). The disease resolved after ≤3 weeks.

Conclusions: Lipschütz's ulceration mainly affects both sexually inactive and, less frequently, sexually active subjects ≤20 years of age, presents with ≤3 vulvar ulcers, resolves without recurrences within 3 weeks and is temporarily associated with an infection, most frequently a flu-like illness or an infectious mononucleosis syndrome.

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Prevalence and risk factors of lactic acidosis in children with acute moderate and severe asthma, a prospective observational study.

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Introduction: Lactic acidosis is a known complication of acute asthma but data are scarce in paediatric population. The aim of this study was to describe the prevalence and risk factors contributing to lactic acidosis in children hospitalised for moderate and severe acute asthma.

Patients and methods: A total of 154 children 2-17 years of age hospitalised for moderate or severe asthma was enrolled in the observational prevalence monocentric study. All patients had capillary blood gas assessment 4 hours after the first dose of salbutamol at hospital. The primary endpoint was the prevalence of lactic acidosis. Potential contributing factors as: age, sex, body mass index, initial degree of asthma severity, type of salbutamol administration (nebuliser or inhaler), steroids, ipratropium bromide and glucose containing maintenance fluid, represented secondary endpoints.

Results / observations: Prevalence of lactic acidosis was 27% in our population. In univariate analysis age older than 6, female sex, severe crisis, obesity, hyperglycaemia and salbutamol received by nebuliser were significantly related to lactic acidosis. In multivariate analysis single salbutamol administered by nebuliser (OR = 10, IC95% 2.2-45), age older than 6 (OR = 2.8, IC95% 1.8-6.4) and hyperglycaemia (OR = 2.6, IC95% 1.3-5.9) were identified as risk factors of lactic acidosis.

Conclusion: Lactic acidosis is frequent in children with acute moderate or severe asthma receiving salbutamol. Salbutamol administered by nebuliser, hyperglycaemia and age older than 6 are risk factors of lactic acidosis during acute asthma.

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Non-urgent paediatric emergency department visits in two Swiss university hospitals: Preliminary Results: of a mixed prospective and retrospective observational study

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Background: A growing proportion of visits to paediatric emergency departments (PEDs) in Switzerland are non-urgent. PED visits for non-urgent complaints put a growing burden on the health care system and impact the quality of patient care. Little is known about factors leading to non-urgent PED consultations in Switzerland.

We aim to investigate low-acuity consultations in two PEDs representing the French and German speaking regions of Switzerland, in order to assess the proportion of non-urgent PED visits, characterize them and understand possible causes.

Methods: We present preliminary results from the site of CHUV of a mixed prospective and retrospective observational study conducted in the PEDs of Hôpital de l'Enfance, CHUV, Lausanne, and Notfallzentrum für Kinder und Jugendliche, Inselspital, Bern.

Retrospective part: extraction of administrative and medical data from the clinic information systems for all visits that occurred in the PED between January and December 2018.

Prospective part: prospective, systematic sample of 1000 patients and their families visiting the PEDs. Questionnaire on demographic features and medical resources and chart extraction of detailed medical information from the same visit.

We classify PED visits in two categories: high-acuity and low-acuity. We define low-acuity visits as a combination of: triage level 4 and 5 on ATS scale, no radiologic study or laboratory test performed, no treatment prescribed other than over-the-counter drugs, and patient discharged home.

Results: Preliminary analysis of the extraction of 32'606 visits that occurred in 2018 at the PED of CHUV shows that about 60% of those visits met our criteria for low-acuity.

Recruitment for the prospective part started on the 01.09.2019 and we plan to include 1000 patients. As of end of January 2020, 407 patients have been recruited from both sites (consent rate 84%).

Analysis of the first 109 visits included at the PEDs of CHUV shows that 58% meet our criteria for low-acuity. In this sample group, the most frequent reasons for consultation were musculoskeletal injury, fever and abdominal pain.

Discussion: Most PED visits at CHUV are low-acuity visits. Our ongoing study is important because it will help understand the reasons behind the inappropriate use of PEDs in Switzerland. This will be helpful for future decisions regarding the allocation of resources and improvement of care.

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Cyanosis and food: a not too uncommon association

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Background: Methemoglobin (M) is an abnormal condition of hemoglobin that happens when the iron ion is oxidized in its ferric state, reducing the ability to bind O2 molecules impairing the normal loading of oxygen in the lungs and its delivery to the peripheral tissues. Congenital causes of M are due to autosomal dominant mutations of the hemoglobin chains (HbM) or, most commonly, to an autosomal recessive deficiency of Cytochrome b5 or Cytochrome b5 reductase (b5R). Acquired causes of M result of an increased methemoglobin formation by various hexogen agents, such as toxic products, medications or food.

Cases description: We describe two cases of previously healthy infants of 6 (A) and 8 (C) months. A arrived to our E.R. with cyanosis, apathy, cough, fever and 84% oxygen saturation without any response to oxygen administration. C arrived to our E.R. for sudden presentation of peripheral and central cyanosis and 88% of oxygen saturation without other symptoms. In the case of A, subsequently to the initial suspicion of sepsis, an antibiotic therapy with ceftriaxone was started. In both cases, a peripheral venous blood gas analysis was performed, that showed a hypermethemoglobinemia (A: 20.8%, C: 12.1%). Both presented a spontaneous normalization of methemoglobin levels. We found out, that a few hours before the onset of symptoms, A had eaten beans and C freshly milled spinach. Nitrate quantitative analysis revealed toxic values that confirmed the suspicion of a food-related nitrate intoxication.

Discussion: In infants, since the erythrocyte b5R activity ranges normally from 50 to 60% of adult activity, the development of M is more common even with low doses of externally induced products. M therapy consists in avoiding the exposure to offending agents and preventing the consequent formation of high levels of the oxidized form of HbA. Other possible treatments depend on M blood concentration: from no therapy in lesser degrees (<20%) to administration of agents with a reducing potential such as Methylene Blue or Ascorbic acid at high concentration and even to life support or blood transfusion in case of shock.

Conclusion: Nitrate intoxication should be considered as a potential diagnosis in any case of sudden cyanosis. It's fundamental to take the patient's clinical history into consideration. Parents should be advised regarding cooking and storage of foods that naturally contain high doses of nitrate.

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An almost fatal attraction

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Introduction: Foreign body (FB) ingestion is a common problem in children. The majority of FBs pass spontaneously through the gastrointestinal tract, however magnetic FBs pose a particular risk for obstruction due to attraction force through the intestinal wall. Whereas ingestion of a single magnetic FB may, in most cases, be managed as a simple FB ingestion, the ingestion of multiple magnetic FBs is associated with a high risk of complications and requires aggressive management.

Case report: A 20-month-old healthy girl was addressed to our emergency department for apathy, refusal to eat and drink and incoercible non-bilious vomiting in the prior three days. No history of fever nor diarrhoea. In the past three days, she passed gas but not stools. She was apathetic but responsive, she showed signs of dehydration. Vital parameters were within normal range. Abdomen was tender. After IV rehydration general conditions did not improve. Blood exams were within normal range, with the exception of a compensated metabolic acidosis. Because of worsening of neurological status and in absence of signs of dehydration, a neurologic aetiology was ruled out with a brain CT and a lumbar puncture, which were normal.. In the following days, she developed abdominal pain and distension. Abdominal x-ray revealed 15 spherical foreign bodies and bowel dilatation. The patient underwent emergency laparotomy. Multiple bowel perforations and a volvulus were found, caused by the magnetic attraction with several entero-enteric fistula. Removal of all magnets along with double intestinal resection and anastomosis were performed. After few days the child recovered well without sequelae.

Conclusions: Vomiting in children is most commonly due to acute infectious gastroenteritis; however, it is a nonspecific symptom and may be the initial presentation of serious medical conditions, including infections (meningitis, septicaemia); metabolic disease and anatomical abnormalities (obstruction, volvulus). FB ingestions are frequent in children. Ingestion of multiple magnets is very rare and poses a unique hazard of intestinal obstruction and perforation. The ingested magnets may reside in different intestinal loops and magnetic attraction force causes pressure bowel necrosis with perforation, fistula, or volvulus requiring prompt surgical management.

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Needle in a haystack or finding the underlying cause of a protein-losing enteropathy.

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Case report: A 3½y.o girl was referred for weight loss,diarrhea, abdominal pain and oedema over 2 months. Past medical history is marked by recurrent upper respiratory tract infections, gastro-enteritis, aphteous oral lesions and one episode of self-resolving periorbital oedema with increase fecal calprotectin 7 months prior to admission.

Work-up revealed severe hypoalbuminemia, hypochromic microcytic anemia, normal urine workup, decreased total immunoglobulin. Protein losing enteropathy(PLE) was diagnosed.

Gastrointestinal endoscopies showed edematous, altered mucosa with sign of colitis.Histological analysis revealed duodenal lymphangiectasia, severe gastric mucosal inflammation with erosive lesion, focal active colitis and epithelia dystrophic mucosa. Pancolitis was final diagnosis due to either very early onset inflammatory bowel disease (VEOIBD) or cow's milk protein allergy (CMPA). Primary immunodeficiency was discussed according to history. VEOIBD genetic panel was negative.

Treatment was IV Immunoglobulin, albumin, iron replacement therapy and blood transfusion.

Eviction of cow's milk protein (CMP) was made for high suspicion of CMPA.

Due to clinical deterioration, steroids were introduced with rapid clinical and biological improvement. Weaning was made over 4 months with good clinical outcome.

Control endoscopy revealed absence of sign of chronic colitis but presence of lymphagectasia in duodenum. The likely diagnosis of PLE is CMPA according to the findings, as with lymphangiectasis there is more mucosa contact with CMP.

As the diagnosis remains unclear, strict follow-up is mandatory.

Discussion: PLE is a rare complication characterized by excessive loss of proteins into the gastrointestinal tract due to impaired integrity of the mucosa.

Clinical presentation is variable, but mainly consists of edema due to hypoproteinemia. Diagnosis of PLE is commonly based on determination of fecal -1 antitrypsin clearance. Treatment of PLE targets the underlying disease but also includes dietary modification, supportive care, and maintenance of nutritional status.

Conclusion: PLE should be considered in patients with hyoproteinemia after exclusion of malnutrition, defective synthesis or proteinuria.

Finding underlying disease is challenging.

Gastrointestinal causes of PLE are various: CPMA, VEOIBD,eosinophilic gastroenteritis, infectious and primary immune deficiency with intestinal inflammation.

Prognosis of PLE is unknown and depends upon underlying disease.

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A case report of a pediatric patient with PTEN-Hamartroma Tumor Syndrome (PHTS): a diagnostic and follow-up challenge.

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Background: PTEN hamartroma tumour syndromes (PHTS) are a group of rare syndromes associated with germline mutation of PTEN (phosphatase and tensin), a tumor suppressor gene. PHTS include Cowden syndrome and Bannayan-Riley-Ruvacalba syndrome, characterized by a spectrum of features as macrocephaly with/without intellectual disability, benign hamartromas and an increased lifelong risk of certains cancers.

Case description: Our patient is a previously healthy child, born from healthy unrelated parents from Kenya. At the age of 11 years old, he was referred to our gastroenterology department for a severe iron deficient anemia, that was recurrent despite an oral and intra-venous iron replacement during more than 6 months. Hemoglobin electrophoresis was normal. Subsequent esophagogastroduodenoscopy revealed gastritis, histologically compatible with H. Pylori. Six months later, anemia persisted, despite successful eradication treatment. Phisical examination was notable for severe macrocephaly (head circumference >P97; +5.4SD), multiple facial thrichilemmomes and penile hyperpigmented maculae. A neuropsychological evaluation reported difficulties with concentration and with cognitive flexibility; he was assigned to a special school. A colonscopy showed several juvenile polyps and multiple large base polyps in the left colon and rectum. The conjoint occurrence of polyposis with macrocephaly, learning difficulties and dermatological typical lesions was suggestive of PHTS. Mutation analysis identified a novel c.80A>G mutation in the PTEN gene, confirming the diagnosis. Family testing for this mutation was negative. Thyroid ultrasound at diagnosis was normal. According to some authors, we will repeat thyroid ultrasound annually due to the risk of thyroid cancer since childhood. Colonscopies will be performed by 35 years of age due to the risk of gastro-intestinal cancer at younger age, then every 1-2 years if multiple/adenomatous polyps present, or every 3-5 years if they are sparse or not present.

Conclusion: Diagnosis is challenging in pediatric patients as some manifestation usually develops later in life. A presentation with recurrent anemia due to chronic gastro-intestinal bleeding is not common: Hansen-Kiss et al. described a proportion of only 6%. Early pediatric diagnosis allows an adequate multi-disciplinary follow-up, as well as the testing other at-risk family members. At this time, consensual guidelines for follow-up in pediatric patients are lacking.

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Use of Skim Breast Milk in a Preterm Infant with Congenital Chylothorax

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Introduction: Congenital chylothorax is a rare condition in which fluid accumulates in the pleural space during fetal development. The affected fetus and neonate may present with nonimmune hydrops fetalis. It can cause significant respiratory morbidity, as well as lead to malnutrition, immunodeficiency and increased risk of thrombosis. First-line therapy is fat-free enteral nutrition, usually a fat-free formula, combined with either intravenous lipids or enteral medium chain triglycerides.

Case: Our patient was a preterm female infant born at 32+1 weeks of gestation. A caesarean section was performed because of increasing hydrops fetalis of unknown aetiology. The infant required ventilatory support und surfactant therapy. Pleural drains were inserted bilaterally, and voluminous effusions were tapped. The effusions persisted and were consistent with chylothorax, being rich in triglycerides after three days of feeding with mothers own milk (MOM). In view of the prematurity and the positive effects of colostrum, we decided to feed the patient with fat-reduced MOM, combined with parenteral nutrition. The fat content of the MOM was reduced with a centrifugation and separation process. Macronutrients were measured pre- and post-separation-process.

A steady reduction in chylus could be observed during this intervention. The pleural drains could be removed. Yet, the infant required a period of entirely fat-free enteral feeds. At the moment several tests on aetiology of the chylothorax, including genetics, is pending

Conclusion: On the basis of the clear protective and positive effects of human milk, especially in preterm infants, a fat-reduced MOM might be tried as an alternative to fat-free enteral formula, in the initial treatment of congenital chylothorax.

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Paracetamol intoxication causing acute kidney injury in absence of the hepatotoxicity: Pediatric case report.

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Purpose: The risk of hepatic toxicity due to paracetamol intoxication is known to all practitioners. Nevertheless, paracetamol nephrotoxicity exists and occurs in approximately 0.4% to 8.9% of patients with paracetamol overdose, sometimes occurring without hepatotoxicity. Its onset and the peak of serum creatinine concentration are important to know to guide detection of this important complication. We report one pediatric case of acute kidney injury related to paracetamol intoxication without hepatotoxicity.

Clinical features: A 15-year girl, in good health with no routine treatment nor significant medical history, consulted the emergency department 48 hours after a voluntary drug intoxication with paracetamol (a single dose of 24g, 540 mg/kg), without any co-intoxication. Serum paracetamol level was not tested due to the late consultation of the young patient. Hepatic blood tests were normal, therefore N-acetylcysteine treatment was not indicated. Clinically, she initially presented diffuse abdominal pain and vomiting. Progressive localization to bilateral kidney pain was observed by 72 hours post-ingestion, lasting for another 4 days. In parallel, serum creatinine concentration, which was noted to be elevated at 100 µmol/l 48 hours post-ingestion, increased further to reach a maximum serum creatinine concentration at 171 µmol/l on the 8th day (eGFR, estimated glomerular filtration rate, was 28 ml/min). Urine testing revealed non-nephrotic proteinuria (proteinuria/creatininuria ratio 44g/mol) and hematuria. There were no hyaline casts or acanthocytes in the urine sediment, nor electrolyte or acid-base disorders. She did not present oliguria nor hypertension. Renal ultrasound results were normal. Renal function improved gradually with no particular intervention, allow-

ing discharge from hospital on the 9th day post-ingestion. Serum creatinine concentration returned to previous baseline level one week later as confirmed during a follow-up consultation with her pediatrician.

Conclusion: Renal paracetamol toxicity consists of acute tubular necrosis. Its pathophysiology remains unclear and there is no known effective treatment to interrupt renal tubular damage. In the majority of cases, however, acute kidney injury is spontaneously reversible. The aim in discussing this case is to highlight the importance of following serum creatinine level during the first week in cases of paracetamol overdose, regardless of the quantity of paracetamol ingested.

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Posterior urethral valves: a pathology with neonatal revelation

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Introduction: Posterior urethral valves (PUVs) are the most common congenital obstructive malformative uropathy of the boy's most frequent bladder seat and of unknown etiology. The incidence is relatively rare estimated at 1 in 4000 to 8000 births. Treatment remains a challenge requiring antenatal and neonatal multidisciplinary management and then in childhood and adolescence to prevent chronic renal failure.

Observation: A newborn male born from a pregnancy followed and completed. Cesarean born for a bi-cicatricial uterus with oligo-amnios. The telltale sign was urinary type of late urination. The renal balance and the blood ionogram were normal. Renal-bladder ultrasonography showed bilateral uretero-hydronephrosis associated with a bladder control with the presence of a right urinoma. The urinary cystography revealed a grade 5 bilateral vesico-ureteral reflux. The rest of the malformative assessment was normal.

Comment: PUVs are the main cause of Congenital Bladder Obstruction. The diagnosis may be suspected antenatal. Neonatal telltale signs may be urinary or extra-renal. Their long-term prognosis depends on the severity of the obstruction and its impact on bladder and kidney function. Management should be aggressive, with close monitoring of renal function.

Conclusion: PUVs represent a complex pathology, with significant repercussions throughout life. They constitute a pediatric urological emergency directly threatening the renal function and thus the child's vital prognosis. Interest of an early diagnosis. Antenatal diagnosis is possible. The care is multidisciplinary.

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Naguib Richeiri Costa syndrome

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Naguib-Richeiri-costa syndrome is a type 2 acro facial dysostosis associated with genitourinary anomalies; The affected patients have no intellectual deficit.

Also known as:

Hypertelorism-Hypospadias-Polysyndactyly Syndrome

Naguib Syndrome

Rare, described only in three families, it has a neonatal onset, its transmission in autosomal recessive

- Prevalence 1/100000.
- Girls and boys can be reached.
- All races and ethnicities can be affected by this syndrome.
- So far, no risk factor has been clearly identified.

In this clinical case, a condition of an 18 months old infant is exposed

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Acute interstitial nephritis in infant presenting with anuria – two rare case reports

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Introduction: Tubulointerstitial nephritis (TIN) is a heterogeneous group of conditions characterized by an inflammation and edema of the renal interstitium with secondary involvement of the tubules. It is rarely reported in the paediatric age range. The aetiologies are multiples, the presentation is variable and the treatment remains controversial.

Methods (case reports): We report 2 cases of Acute Interstitial Nephritis (AIN) in two infants aged 9 (case A) and 13 months (case B). Both presented with a KDIGO stage 3 anuric Acute Kidney Injury (AKI) in the context of febrile Ear Nose and Throat (ENT) Infection. A urine sediment in both cases showed sterile leukocyturia and proteinuria. In case B it resulted to be nephrotic proteinuria. A renal biopsy showed an infectious vs toxin origin in case A and an infectious with a possible pyelonephritic component in case B. Nonsteroidal anti-inflammatory drugs (NSAIDs) and amoxicillin (AMX) – were discontinued in case A. Both cases were treated with intravenous antibiotics, peritoneal dialysis, diuretics, corticosteroids and antihypertensive drugs. The doses and duration of steroid therapy were not equal in both cases.

Conclusion: AIN is a rare and often self-limiting pathology. The presentation with acute anuric AKI and nephrotic proteinuria is even less frequent. Despite the severe presentation, the different aetiology and treatment, the clinical and biological outcome of both cases was rapidly favourable. Diuresis was restored within 2 days and the renal function was normalized during the following weeks. Secondary complications (hypertension and anaemia) were also resolved with supportive therapy. Steroid treatment is controversial and further research is needed to determine dosage, way of administration and duration of treatment in this kind of patients. Stopping the suspected toxic substance is imperative.

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Rare case of primary hyperoxaluria in asymptomatic 15 month old child with bilateral nephrolithiasis.

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Introduction: A risk factor is identified in 80 percents of pediatric nephrolithiasis, usually urinary infection, urinary tract malformation, or urinary metabolic anomaly. Early or multiple lithiasis should raise awareness for metabolic disorders.

Case: Multiples bilateral nephrolithiasis were found in a healthy and asymptomatic 15 month old Egyptian infant during a follow-up ultrasound performed for minimal pelvic dilatation. Family history was negative for consanguinity and renal disease. Growth and renal function were normal. There were no acidosis or hypercalciuria, but urine showed hyperoxaluria (oxalate/creatinine ratio of 600-850 mmol/mol (normal value <170 mmol/mol at 15 months)). Genetic assessment is ongoing. Patient is under supportive treatment by vitamin B6, potassium citrate, and hyper-hydration.

Oxalate is an end product of several liver metabolic pathways and is eliminated in urine. In primary hyperoxaluria, rare autosomal recessive disorder, three liver enzymatic defects lead to an overproduction of oxalate. Type 1 is the most severe form and causes progressive loss of renal function. Whewellite crystals accumulate in kidney tissue. Once the glomerular filtration rate is below 30-45ml/min/1.73m2, oxalate production exceeds kidney capacity to eliminate it and precipitate in multiple organs.

Initial care focuses on decreasing crystallization with potassium citrate and hyper-hydration. Pyridoxine (vitamin B6), the main co-factor of the defective enzyme, is often given, but only decreases oxalate in a third of the patients with type 1. The only curative treatment is a combined liver and kidney transplants, correcting the enzymatic defect and restoring renal function. A promising therapy by RNA interference blocking the metabolism of glyoxylate is on trial.

Conclusion: The association of lithiasis, nephrocalcinosis and altered renal function is highly suggestive of hyperoxaluria. There is an important delay of 3-5 years between the first stones and the diagnosis of hyperoxaluria, with potential loss of kidney function in the meantime and important morbidity.

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Shone complex: a rare congenital pathology

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Introduction: The Shone complex (ShC) is an extremely rare congenital heart disease, characterized by the presence of several obstructive abnormalities of the left heart. Currently, eight lesions are considered part of this syndrome. A person must have at least three of these lesions to be diagnosed. The first four lesions were described: supra-mitral valve, mitral valve parachute, subaortic stenosis and coarctation of the aorta.

Observation: A newborn female born from a pregnancy followed and led to 38 Weeks Amenorrhé, the first born of a non-consanguineous couple. Born vaginally with good adaptation to extra-uterine life. Hospitalized from birth. Clinical examination revealed dysharmonious intrauterine growth retardation, cyanosis which regressed under simple oxygen therapy and a weak left femoral pulse. Moreover, there were no signs of acute heart failure. Cardiac Doppler ultrasound revealed severe hypoplasia of the left ventricle, coarctation with a hypoplastic aortic arch, aortic valve atresia and mitral atresia with interauricular communication. The rest of the malformative balance sheet returned without any anomaly.

Comment: Due to the large number of defects involved, individuals present themselves in different ways, with a wide range of combinations of defects, symptoms. The prognosis will depend on the severity and the number of defects. Repair of the mitral valve is usually done early to prevent the onset of pulmonary hypertension.

Conclusion: ShC also called Shone syndrome or abnormality is an extremely rare cardiac pathology. There are incomplete forms of the disease in which only two or three lesions are present. The treatment consists of a surgical correction of the constituent defects.

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A tracheal compression syndrome revealing a double aortic arch

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Introduction: Aortic artery abnormalities represent a group of rare congenital malformations. Spiral angiography has become the key examination with a dual diagnostic and therapeutic interest.

Observation: a 15-day-old female newborn who had persistent respiratory distress since birth. The clinical examination at admission revealed a general mean condition, moderate respiratory distress with presence of laryngeal stridor and diffuse bronchial rales on auscultation. The frontal chest X-ray and the cardiac Doppler ultrasound were without abnormalities. The oeso-gastroduodenal transit revealed a double regular impression of the two oesophageal margins above the hull. The thoracic angioscanner showed a double aortic arch with hypoplastic appearance of the left arch encircling the oeso-tracheal axis. The operative indication is posed, a thoracotomy was performed and the operative follow-ups were simple.

Conclusion: The double aortic arch remains a rather rare pathology that must be considered in the face of neonatal respiratory distress without obvious etiology. Surgical treatment is indicated if the symptomatology is frank and / or life-threatening. Generally the postoperative evolution is satisfactory.

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Cardiac rhabdomyoma revealing tuberous sclerosis of bourneville: (2) case and a review of the literature

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Introduction: Tuberous sclerosis of bourneville (TSB) is a rare genetic disorder characterized by the multi-systemic development of hamartomas. Cardiac rhabdomyomas are benign hamartomas that may represent the first manifestation of this phacomatosis with which they are associated in more than half of the cases. The association of cerebral and cardiac lesions during the neonatal period is rare.

Observation N° 1: This is a new male born from a pregnancy followed and completed. Hospitalized for management of persistent respiratory distress. The chest x-ray showed large vascular hils. Cardiac echo-Doppler showed pulmonary arterial hypertension with multiple myocardial tumors, one of which impeded the left ejection route. The operative indication is posed, a thoracotomy was performed and the operative follow-ups were simple. The brain MRI returned without abnormalities.

Observation N° 2: This is a newborn female born from a pregnancy followed and carried to a term. His mother carries a TSB. hospitalized from birth. An echocardiography performed showed the presence of multiple echogenic myocardial nodules at the two ventricles, non obstructive. Electrocardiogram showed no disturbance of rhythm or conduction. In front of these cardiac rhabdomyomas, a cerebral MRI was requested in search of characteristic lesions. In addition, the newborn did not show any clinical signs.

Comment: The diagnosis of TSB is radiological-clinical. Intracardiac rhabdomyomas are often the first and the earliest clinical manifestations of TSB. These rhabdomyomas regress spontaneously in most cases but can sometimes be complicated by arrhythmia or obstruction requiring medical-surgical treatment.

Conclusion: TSB is a multi-systemic, autosomal dominant genetic disorder characterized by the development of benign tumors, hamartomas. It is due to mutations of two genes TSC1 and TSC2. The antenatal diagnosis is feasible. Interest of genetic counseling.

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Total anomalous pulmonary venous return: a very serious congenital pathology

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Introduction: The abnormal pulmonary venous return includes the malformations concerning the connection of the pulmonary veins. When there is no pulmonary vein in the left atrium, it is a total anomalous pulmonary venous return (TAPVR). These total forms represent 1 to 2% of all congenital heart defects.

Observation: Newborn baby, 20 days old, female, from a completed pregnancy and an inbred marriage. The clinical examination identified the signs of cardiac decompensation (Tachycardia, polypnea and hepatomegaly). The chest X-ray showed cardiomegaly with large vascular hilum. Echocardiography demonstrated PHT with an inter-ear communication. The CT angiography revealed an infra-cardiac TAPVR. The report of other malformations returned without anomaly.

Comment: The clinical course will depend on the presence of an obstruction to the venous pulmonary return. Our patient had a narrowing of the connector at the portocave shunt area. curative surgery in the neonatal period is necessary.

Conclusion: TAPVR is a rare but serious congenital anomaly. The CT angiography is the essential examination to better appreciate its anatomy. The treatment is surgical.

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Absent femoral pulses and normal aortic arch anatomy: Search beyond the prevalent disease

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Introduction: Middle aortic coarctation is a rare clinical entity in childhood (<1/1'000'000) characterized by narrowing of the abdominal and/or distal descending thoracic aorta, and associated with significant morbidity and mortality. Most cases of midaortic syndrome (MAS) are idiopathic, but some have been described associated with genetic and acquired diseases. It presents with varying involvement of the visceral and renal arteries. The symptoms vary depending on the degree and location of stenosis: most patients present severe renovascular hypertension, absent femoral pulses, abdominal bruit, and claudication. Most cases undergo endovascular or surgical management with residual hypertension in some cases, requiring medication or reintervention.

Case report: We report the case of a female neonate, born at term by vaginal delivery after an uneventful pregnancy. Maternal serologic tests were normal and family history was silent. Good adaptation to extrauterine life and anthropometric parameters were within normal range. Examination of the newborn revealed absent femoral pulses, other than that good general conditions. Blood pressure values were normal in the 4 limbs. On her second day of life, an echocardiography was conducted which showed normal anatomy and function of the heart, as well as a normal aortic anatomy. The left ventricle was slightly hypertrophic. On her 26th day of life, a blood pressure gradient of 60 mmHg was found between the upper and lower extremities (right arm 127 mmHg, legs 65 mmHg), an echocardiography was repeated with results comparable to the previous one, however it also showed a severe stenosis of abdominal aorta immediately below the diaphragm (diameter 1 mm) and several abdominal collateral vessels. An MRI confirmed the diagnosis with stenosis of the right renal artery. A 4 mm stent was implanted in the abdominal aorta requiring dilation 2 months later. The procedure was successful. Upon check, at 6 months of age, she presented a regular growth and development, normal blood pressure in the four limbs and the echocardiography showed a 4 mm wide stent with anterograde flow, with a maximum gradient of 8 mmHg.

Conclusions: Patients with MAS often have additional visceral stenosis. Persistent hypertension after intervention is common and requires long-term monitoring. Further studies are needed to better understand etiology, long-term effectiveness of treatment, and to determine the optimal management of this condition.

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Bernard-Soulier syndrome with neonatal revelation: a rare pathology

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Introduction: Bernard-Soulier syndrome (BSS) is a hereditary hemorrhagic disorder affecting the megakaryocyte / platelet lineage. It is characterized by a haemorrhagic tendency, giant blood platelets and thrombocytopenia. This syndrome is extremely rare, with about 100 cases reported in the literature. It is due to a quantitative or functional deficit of the glycoprotein GPIb-V-IX platelet complex.

Observation: We report the case of a new male born from a pregnancy followed and completed, the 4th of a consanguineous couple. Hospitalized for exploration and management of severe thrombocytopenia considered to be related to maternal autoimmune thrombocytopenia. In view of the persistence of this thrombocytopenia despite the immunoglobulin treatments instituted, the exploration was started. The blood smear revealed a macro-thrombopenia with giant platelets. The study of platelet function found an absence of agglutination induced by ristocetin. The study of platelets by flow cytometry revealed a collapsed GPIb level.

Comment: Correct and early diagnosis of BSS is essential. The unpredictable severity of hemorrhagic symptoms is independent of low levels of GPIb. Our patient required platelet transfusions. The prognosis is generally good with appropriate treatment.

Conclusion: BSS also called hemorrhagic thrombocytic dystrophy is a rare inherited pathology, of autosomal recessive inheritance. With good prophylaxis, a fairly normal quality of life can be maintained. In addition to other therapies, the BS could be a candidate for future gene therapy.

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Feto-maternal erythrocyte incompatibility no ABO no anti-RH1: a pathology not to be ignored

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Introduction: Fetomaternal maternal erythrocyte incompatibilities (FMEI) are an area of perinatal immunohematology that is always manifested after birth, but sometimes during pregnancy. They represent 0.5 / 1000 births. The incompatibilities of anti-JK1, -MNS1, -RH4, -RH2 specificity are rare, especially since they are not accessible to immunoprophylaxis. The care requires a multidisciplinary approach by specialists of this period of development.

Observation N° 1: A newborn male born from a pregnancy followed and completed, hospitalized for management of early neonatal jaundice. Blood grouping of the newborn: A rhesus positive CC ee K negative. Mother: A rhesus positive CC ee K negative. father: O Rhesus positive cc Ee K negative. Total Bilirubin = 175 mg / l, Hemoglobin = 10.6 g / dl. Coombs test ++++ with presence of anti-RH4 alloantibodies.

Observation N° 2: A newborn female born from a pregnancy followed and completed, who underwent 3 exchange transfusions in utero. Hospitalized for the management of early neonatal jaundice. Blood grouping of the newborn: Cc ee K negative Rh positive. Mother: B Rhesus positive. Father: B Rhesus positive. Total Bilirubin = 66 mg / l, Hemoglobin = 16.4 g / dl. Coombs test +++ with presence of anti-MSN1 allo-antibodies.

Observation N° 3: A new male born from a pregnancy followed and completed, who underwent 3 exchange transfusion in utero. Hospitalized for the management of early neonatal jaundice. Blood grouping of the newborn: O rhesus negative cc ee K negative. Mother: A negative rhesus cc ee K negative. Father: O rhesus positive CC Ee K negative. Total Bilirubin = 90 mg / l, Hemoglobin = 12 g / dl. Coombs test +++ with the presence of anti-JK1, anti-RH2 and anti-RH1 allo-antibodies.

Discussion: In addition to rhesus FMEIs, today the pediatrician is essentially confronted with incompatibilities in the RH4, RH2 and MNS1 groups, which can sometimes be very severe. Interest of the noninvasive antenatal diagnosis of these compatibilities (genotyping of blood groups of the fetus on maternal blood) and that of their fetal anemic complications (Doppler velocimetry of the cerebral artery).

Conclusion: FMEI remains valid because of their persistence despite prophylactic measures and because of considerable progress in management methods. Interest in coordinating obstetric-pediatric efforts for immediate management of FMEIs in the birth room and optimize detection of jaundice and phototherapy.

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Role for CD48 in controlling immune hyperinflammatory syndrome

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CD48 is expressed on nearly all cells of haematopoietic origin. CD48 interacts with the 2B4 receptor on NK cells, gamma delta T lymphocytes and activated cytotoxic T lymphocytes. Phenotypes and mechanistic studies of an inherited defect in CD48 have never been described in humans. A deleterious heterozygous mutation in CD48 was found in a patient suffering from recurrent episodes of fever, rash, bi-cytopenia, hy-

perferritinaemia and hypercytokinaemia. The patient's heterozygous mutation led to reduced cell surface expression of CD48. This was associated with a defect in NK cell maturation and activation In vitro assays with CD48 haploinsufficient cells showed a dampening in cytotoxic effectiveness. In addition, a decreased susceptibility to killing of CD48 haploinsufficient target cells was observed. Patient's PBMC transcriptome also indicates a defect in 2B4-mediated NK cell activation, susceptibility to viral infections and LAT-dependent cytokine overexpression. Viral infections in CD48-deficient mice induced interleukin 6 hypercytokinaemia, similar to the cytokine pattern observed in our patient during flares. This study provides evidence that sufficient CD48 expression is needed to control inflammation by ensuring a correct NK cell maturation, functionality and target cell immune regulation by cytotoxicity.

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Congenital mediastinal neuroblastoma as cause of a neonatal respiratory distress

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Background: Respiratory distress is a common cause for hospitalisation in the newborn. Possible uncommon causes such as extrinsic lung compression should not be underestimated. Neuroblastoma represents only about 10 percent of childhood cancers but is the most common solid tumour needing chemotherapy in neonates besides Retinoblastoma. Located in the mediastinum, respiratory distress can result.

We present the case of a congenital neuroblastoma of the mediastinum with liver metastases, unrecognized in utero.

Case: The pregnancy and birth of the fullterm girl were normal. In the first minutes of life, she developed severe respiratory distress. Despite stimulation and ventilation she did not improve, a pre- and postductal saturation difference of more than 20% was evident and the chest X-ray showed an enlarged mediastinum.

She was transferred to Berne with the suspicion of a severe congenital heart defect. The echocardiography showed no structural anomalies but a mass that compressed the heart and the lungs and moved the aorta anteriorly. During the prenatal sonographic check-ups the presence of a mediastinal mass was not reported.

An MRI was performed on the first day of life showing a large mediastinal tumour with compression of the lungs and the left atrium with an intravertebral component with 50% infiltration of the spinal canal as well as multiple metastases in the liver. Because of these life threatening symptoms, it was not possible to perform a biopsy before starting treatment. Localisation of the tumour and the metastases allowed the diagnosis of a neuroblastoma. This was confirmed later by highly elevated homovanillic and vanillylmandelic acid in the urine taken before starting treatment. Chemotherapy according to the SIOPEN Lines protocol was started immediately. Due to severe tumorlysis syndrome and low renal function on the first day of life hemodialysis was needed. 14 days after starting chemotherapy, it was possible to stabilise the patient and to obtain the histological and molecular confirmation of a histologically and clinically favourable neuroblastoma.

Conclusion: Neonatal neuroblastomas can have an uncommon presentation. The diagnosis must be prompt so a multidisciplinary treatment can be started immediately in particular if life threatening symptoms are present.

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Epistaxis in infant: frequent parental concern or red flag?

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Background: Nose bleeding is a frequent occurrence in childhood affecting up to 60% of paediatric patients. The most common causes after two years of age are traumatic or idiopathic aetiologies without underlying serious illness. In children younger than two years of age, epistaxis is rare (1:10,000) and is associated with trauma, accidental or non accidental, or serious illness e.g., acute leukaemia, being the most common cancer of childhood, with 4000 new cases annually in the USA and 450 in the UK.

Case report, results and follow-up: We report on a 3-year old girl who presented with a first epistaxis episode. On clinical examination active nasal bleeding, pallor, tachycardia and a modest hepatomegaly were noted. Despite nasal ala pressing haemorrhage persisted. Laboratory work-up showed severe anaemia and thrombocytopenia with high degree lymphocytosis. Because of signs of haemorrhagic shock and the alarming laboratory results (Haemoglobin 41 g/dL, Thrombocytes 7x9/L, Leucocytes 7100 cell/mm3 of which 95% Lymphocytes), the girl was hospitalized in intensive care during 4 days. During the hospitalization, the diagnosis of Common B cell ALL was confirmed. The chemotherapy treatment according to the AEIOP-BFM 2009 protocol was successful. After a 14-day hospitalization in the hematologic department the girl was discharged with a schedule for a chemotherapy treatment in a day hospital setting.

Discussion: Recent meta-analysis revealed over 50% of children with leukaemia have palpable livers, palpable spleens, pallor, fever or bruising on diagnosis. About 25% present mucosal bleeding and only 10% present epistaxis on diagnosis. Paediatric leukaemia is a low-prevalence disease in primary care, emergency departments and general paediatric settings. A general practitioner is likely to encounter a child with this cancer only once every 20 years. To improve the early diagnosis of leukaemia, clinicians and general practitioners should keep leukaemia in mind when faced with everyday symptoms like nasal bleeding. Occurrence of multiple symptoms and signs should be considered as red flags.

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Serum sickness like reaction following amoxicillin administration

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Introduction: Drug allergy in children is a major public problem mainly due to diagnosis difficulties, leading to overdiagnosis. Nonimmediate reactions are the most common type of drug allergy with a large spectrum of clinical manifestations, ranging from the benign maculopapular exanthema to most severe reactions such as Steven Johnson syndrome.

Case report: A 6-year-old girl presented to the Pediatric Emergency Department with a 3 day-history of diffuse erythrodermia with superimposed petechial rash involving the face, upper limbs and upper truncus, bilateral knee pain, and persistent fever. Symptoms occurred 24 hours after amoxicillin administration for streptococcal pharyngitis. Her past medical history revealed no previous allergic reaction, no atopy and no previous antibiotic treatment administered.

Physical examination revealed fever (38.7°C) and tachycardia without any sign of shock, a diffuse erythematous macular exanthema with a negative Nikolsky's sign and without any mucosal involvement, associated with petechial lesions over the face, upper limbs and upper truncus. Limb examination showed no limitation in joint mobility although knee movements were painful, and no swelling.

Complete blood count was normal (leukocyte 11.4 G/L, platelet count 187 G/L) with a mild elevation of C-reactive protein (55 mg/L) and an erythrocyte sedimentation rate at 20 mm/h. Renal function testing was normal. Rapid antigen detection test for influenza was negative.

Antibiotic treatment was switched for clindamycin and a 0.6 mg/kg single dose dexamethasone was administered. Twelve hours later, fever and erythroderma had resolved and joint pain had reduced.

Conclusion: This case describes suspected serum sickness like reaction (SS-L-R) following the administration of amoxicillin. This clinical syndrome is more common in children than adults, and can be induced by a drug (typicall cefaclor or other betalactams) and/or a viral infection. This condition should be suspected in any patient presenting with skin rash associated with fever and/or arthralgia/arthritis, and is classified as a type III hypersensitivity reaction. Unlike SS-R, detectable immune complexes and decreased complement C3 and C4 are typically absent. An allergic cause should be excluded or confirmed by a complete allergological

workup including a drug provocation test performed 6 to 12 months after the initial episode.

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Japanese pediatric acupuncture accelerates language development in a late talker

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Introduction: About 15% of children with otherwise normal development have a delayed language development at the age of 24 months (i.e., 'late talker'), showing an active vocabulary less than 50 words and an absence of two-word combinations, often having long-term reduced language abilities. Few experimental studies did show a positive effect of acupuncture in children with autism, mental retardation or cerebral palsy and language delay. We report a case of a late talker with normal cognitive and receptive language development treated with Japanese pediatric acupuncture (i.e., Shonishin).

Case report: A 2-year-old boy was referred to our clinic for treatment with Shonishin. Cognitive, motor, and receptive language development was normal, but he spoke only 5 words. He showed frequent temper tantrums including hitting and biting when he was frustrated because unable to express himself. After two Shonishin treatments 2 weeks apart, the boy started to speak more and to imitate words. After the third treatment, there was further improvement of active vocabulary, and temper tantrums had ceased. After the fourth treatment (6 weeks after start of treatment), the boy started to use two-word combinations and correct pronouns. The treatment was continued monthly up to age 31 months. No more language problems were reported at a follow-up consultation at age 37 months.

Conclusions: Japanese acupuncture did show a positive effect on language development in a late talker. This finding, in combination with data from literature and personal clinical experience, shows that acupuncture can help language development in children with autism, cerebral palsy, or mental retardation and language delay. The effect of acupuncture on language development in children with isolated language delay remains understudied, highlighting the need for further investigation.

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Preventive care for adolescent migrants: a systematic review

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Introduction: Migrant adolescents present health compromising behaviours which are associated with increased risk of developing non-communicable diseases in later life. Primary care physicians are ideally positioned to provide preventive services to young migrants so as to reduce this risk.

Aim: To review preventive health guidance for migrant adolescents in Europe.

Method: Systematic review of the available preventive care guidance on migrant adolescents including: position statements, expert opinions, guidelines, clinical recommendations. Sources: PubMed and "Grey" literature (Google and Google scholar) using the following keywords and their synonyms: "adolescent/teenager/children"; "migrant", "prevention", "preventive services", "Europe". Articles published between 01.01.2010 and 31.12.2019 in English and French were included. Publications were summarised by the following main themes: infectious diseases, immunizations, mental health disorders and non-communicable diseases.

Results: We identified 21 publications. Most of them focused on mental health, infectious diseases and immunisation schemes. No publications focused solely on non-communicable diseases. 16 sources focused on mental health disorders, 8 on immunisation and infectious diseases. A majority of sources proposed general guidance towards migrant health, 7 had a paediatric focus and only 3 proposed adolescent and age-appropriate guidance. Among the 6 Swiss sources, all of them covered mental health, 2 covered all 4 themes and 3 had a paediatric focus.

Conclusions: There is a lack of age-appropriate and culture-appropriate guidance on preventive care for adolescent migrants. Solid data is needed in order to further develop recommendations on the early identification of their health compromising behaviours.

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Severe hyponatremia and acute symptomatic seizure in an infant with respiratory syncytial virus infection

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Background: Respiratory syncytial virus (RSV) is a common cause of bronchiolitis in infants. Hyponatremia secondary to Syndrome of inappropriate antidiuretic hormone secretion (SIADH) can occur in bronchiolitis. However, severe acute hyponatremia associated with seizures in RSV bronchiolitis is extremely rare.

Case report: We report the case of a 7-week-old infant with an acute transient seizure associated to severe hyponatremia in the context of a mild RSV bronchiolitis. The infant was born at term and had no past medical history. During his 3rd day of hospitalisation, he developed a focal tonic-clonic seizure associated to a sodium level of 114 mmol/l. The infant was initially treated with a bolus of intravenous saline 3% (3 ml/kg) with a favourable neurological response and the seizure was stopped after a couple of minutes. A perfusion with an isotonic saline was then started and followed by oral sodium substitution. The sodium treatment was decreased gradually and discontinued after one week with a favourable outcome and no further seizure. The Vasopressin level was increased.

Literature review: We conducted a systematic review of the literature in PUBMED in order to better define the clinical presentation, treatment, outcome and risk factors of RSV bronchiolitis associated with symptomatic severe hyponatremia.

Results: Eleven cases were reported (ours included). All had severe hyponatremia (<= 125mmol/l). Four of 11 were born prematurely (30- 32 weeks gestation). Six of 11 were treated with hypotonic infusions. There was one case reported with neurological sequelae. The severity of the bronchiolitis infection was mild in all cases.

Conclusions: Hyponatremia due to SIADH is common among infants with RSV bronchiolitis. Neurological complications are rare and usually related to severe hyponatremia. The outcome is generally good. Risks factors identified were history of prematurity, hypotonic infusion and severe hyponatremia. Prompt diagnosis and correct treatment of acute severe hyponatremia is imperative and can prevent further complications.

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Can't see the wood for the trees - Point-of-care Ultrasound for Evaluation of cervical lymphadenitis - Watch out for complications

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Introduction: Cervical lymphadenitis is a common presentation in the pediatric Emergency Departement (ED). With a handful of very common differential diagnoses in mind, the management in ED is relatively straightforward. Point-of-care ultrasound can be used for evaluation and timely management. Rare complications can be visualized as well, which can help prioritizing consults and advanced imaging.

Case presentation: A 4 year old girl was referred to our pediatric emergency department (ED) for evaluation with bilateral earache and torticollis. On presentation to ED she was mildly unwell appearing and normal vital signs. She had torticollis to the left without trismus and meningeal signs were absent. The left TM was mildly injected, the right was red and bulging. Left mastoid was red and tender, the pinna displaced laterally. The left neck appeared swollen with tender lymphnodes. Labs revealed leukocytes 20.5G/l, thrombocytes 510 G/l and CRP of 83 mg/l. Our differential diagnosis was either a cervical lymphadenitis with abscess formation or mastoiditis. Point-of-care ultrasound of the neck was

performed to look for cervical abscess, which showed multiple enlarged cervical lymphnodes with normal architecture. An urgent ENT consult for suspected mastoiditis was obtained. Computer tomography (CT) of the head and neck with contrast was ordered and demonstrated bilateral otomastoiditis with an abcess within the left sternocleidomastoid muscle (SCM), a small left sided retroauricular subperiosteal abscess, a thin abscess layer on the left temporal bone and thrombophlebitis in the left sigmoid sinus. Our patient was taken to the operating room for left mastoidectomy and epidural abscess drainage. Fusobacterium necrophorum was identified via PCR.

POCUS findings: The examination was difficult due to limited cooperation of the patient. Some dermal thickening was present and multiple lymphnodes with a normal architecture on Doppler were visualized. Perinodal tissue appeared hyperechogenic. No collections surrounding the lymphnodes were noted. However, on post-hoc review among the many hypoechogenic lymphnodes, an anechogenic structure inside the SCM was identified, which corresponded to a Bezold abscess seen on CT.

Conclusion: Bezold abscess is a rare, but serious complication of otitis media. Distinction between lymphadenitis and abscesses is key. Also check for abscesses in unusual locations like the SCM or the mastoid process.

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Idiopathic systemic capillary leak syndrome: case report

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Background: Idiopathic systemic capillary leak syndrome is an unexplained condition, usually receded by an acute intercurrent illness, characterized by episodes of acute increase in vascular permeability resulting in loss of protein-rich fluid into the interstitial compartment. It mainly affects middle-aged adults with a monoclonal gammopathy but can also affect healthy children. In most severe cases it can be life threatening leading to hypovolemic shock.

Case report: A 7 years old girl presented with fever, flu-like symptoms, periorbital edema and chest pain. Personal history was marked by an episode of para-infectious capillary leak two years earlier. At admission, blood test showed bicitopenia (Lc 4.4 G/L, Tc 128 G/L), slight hypoproteinemia (59 g/L) with normal albumin, CRP and creatinine. Proteinuria was absent. Patient resulted positive for Influenza-virus typ B. Echocardiography showed a pericardial effusion of 6 mm. During the hospitalization we assist to a rapid worsening of general conditions associated to dyspnoea, abdominal pain and headache. Laboratory workup showed hypoalbuminemia and overt hypoproteinemia (30 g/L and 46 g/L respectively). Pleural and abdominal sonography confirmed the presence of bilateral pleural and abdominal effusion. We started an anti-viral therapy (Oseltamivir) for five days and an endovenous steroid therapy for three days. Albumin replacement and diuretic therapy were also prescribed. General conditions progressively improved with reduction of peripheral oedema and dyspnoea. Blood test showed normalisation of albumin and protein values. In order to exclude an immunological dysfunction or a monoclonal gammopathy we performed an Ig dosage which showed normal IgA, IgM and IgG as well as normal plasma and urinary electrophorese.

Conclusion: Idiopathic capillary leak syndrome should be suspect in case of edema, interstitial effusion and hypoalbuminemia associated to a viral infection in otherwise healthy children. First line treatment is supportive with fluid resuscitation. More severe cases could need steroid therapy, albumin replacement, diuretics and IVIG. In recurrent cases a prophylactic treatment with IVIG may be considered.

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Collodion baby: a rare phenotype of congenital ichthyosis

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Introduction: Congenital ichthyosis type collodion baby (CB) is a rare genetic dermatosis. This phenotype is estimated to be between 1 in 50.000 to 1 per 100.000 births. About 30 cases have been reported in the literature. It is due to the abnormal retention of a horny layer in utero. The clinical diagnosis can be confirmed by molecular biology. Antenatal diagnosis is possible.

Observation: A newborn female born from a pregnancy followed and completed, the first born of a non-consanguineous couple. Born vaginally with good adaptation to extra-uterine life. Hospitalized from birth. The clinical examination found the entire integument covered with a glossy diaphragm resembling collodion with an ectropion, an eversion of the lips, wrinkled ear auricles and tapered finger tips with cracks. The newborn has no eyebrows and has little hair.

Comment: The prognosis of CB depends on the neonatal complications hydro-electrolytic, infectious and respiratory, while in the long term, the underlying pathology. The management must be early and adequate. Our patient had a favorable evolution.

Conclusion: CB is a rare but easy to recognize phenotype. It can evolve into different clinical forms of variable severity. Management must be early and rigorous in the neonatal period. Molecular diagnosis is possible. Interest of genetic counseling.

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Erythema nodosum revealing a streptococcus pharyngitis

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Introduction: Erythema nodosum is a relatively rare symptom seen in pediatrics emergency. It is characterized by septal inflammation of subcutaneous fat tissue, usually in the area of the shins. Its cause can be multiple and range from infectious disease to sarcoidosis, enteropathy, medication use or paraneoplasia. Its exact cause can often not be defined and there is almost 1/3 erythema nodosum idiopathic.

Case report: We report a 1 year and 6 months old girl, in good health, who developed a rash on the anterior shins bilaterally the previous day in the morning, and significant pain on palpation in the morning, without fever. There is no history of insect bite, nor trauma, nor wound. There was no new food introduced, no new washing powder, no shower product, no new clothes. She has had a normal intestinal transit without blood and no urinary symptoms. No one is ill in the family and she hasn't travelled lately. Vaccines are up to date. No known allergy.

Good general condition, feverish at 38.9

Dermatological exam: multiple lesions, macular, erythematous, 2x2cm, approximately 5 in each leg in the anterior tibial, significant induration in relation to the lesions, 2 lesions 1 cm diameter at the level of the left buttock, some erythematous lesions 2mm in diameter at the level of the right thigh.

ENT exam: calm eardrums, slightly erythematous back of throat, small cervical lymphadenopathy bilaterally 2mm in diameter.

Urinary stix: pH 5, density 1025, 2x leucocytes, 3x blood, rest was normal

Streptotest: positive

Erythrocytes: 4.1 T/l, Hemoglobin: 112 g/l, MCV: 78 fl, Thrombocytes: 429 G/l, Leucocytes: 13.2 G/l, Neutrophiles total: 61.3%, Monocytes: 7.5%

ASAT: 27 U/l, ALAT: 11 U/l

C reactiv protein: 56.1 mg/l

Bacteriology of the throat: streptococcus pyogenes

ASLO: negative

Discussion: In younger children, Erythema nodosum is often related to infectious diseases. Streptococcus is the most frequent agent (sometimes other agents according to the location and population). It is important to keep in mind other ethiologies mentionned before.

Even in a young aged girl we performed a rapid streptococcus test. The test was positive and the rest of the exams were consistent with this result. We decided to treat with Phenoxymethylpenicillin-Benzathin (0.75 Mio UI) (Penicillin V), per os, 3.5ml 2x/day, during 10 days, and suportive tratment.

Children with erythema nodosum should always be offered further analysis such as testing for streptococcus even in young children under age 3.

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Vulvar itching: an underdiagnosed clinical picture

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Background: vulvar lichen sclerosus (VLS) is a chronic inflammatory dermatosis of unknown aetiology characterized by atrophic plaques with translucent surface, affecting the genital area. Unfortunately, a delayed diagnosis is not infrequent; thus, it may have potentially invalidating sequelae. There is no definitive cure.

Case: A four-year-old girl was referred to our emergency with a 2 months history of vulvar and vaginal itching. Initially treated as common vulvovaginal nonspecific inflammation, the symptoms gradually worsened along with the appearance of an area of slight depigmentation of the vulvar labia and of the perineum. No vaginal secretion was reported, nor any voiding disorder. A paediatric gynaecologist was consulted and, in the light of the clinical and anatomical features, a diagnosis of VLS was made. A topical therapy, based on high concentration corticosteroid (clobetasol propionate 0,5 mg/g) was undertaken and the patient rapidly improved. A clinical follow-up after 4 weeks revealed a complete remission of the symptoms. The skin discolouration persisted, however supposed to improve in a much longer period.

Discussion: VLS has not a clear aetiology, but genetic, autoimmune, infectious, environmental, and hormonal factors are supposed to be involved. Local irritation or traumas seem to play a role in some cases. It is however important to acknowledge that VLS often has spontaneous resolution with the onset of puberty but it is burdened by a high grade of recurrences during menopause, when oestrogens decrease. Symptoms of VLS are slowly progressive, making the diagnosis difficult and frequently delayed; they consist in intense itching, skin depigmentation sometimes involving perianal area, in a "figure of eight" configuration, irritation, dysuria and, in most advanced cases, urinary and faecal incontinence. Complications normally follow the anatomical changes with atrophy and chronic inflammation with sexual, urinary and defecation dysfunction. Fissures might develop with rectal pain and bleeding in case of ulceration. The mainstay of treatment is topical therapy with steroids and, in a few selected cases, systemic retinoid.

Conclusion: the delay in diagnosis can lead to potentially invalidating sequelae that can be reduced by an aggressive therapy. A tight follow-up is necessary to assess the therapy effect and to evaluate the progression as this illness can both have a relapse and usually a spontaneous resolution during the puberty.

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Do we respect the achievable benchmarks of care for bronchiolitis?

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Introduction: Although clinical guidelines for management of bronchiolitis have been published in the last decades, there is still a lack of consensus regarding the application of good practice recommendations. Our aim was to evaluate if we respected the achievable benchmarks of care proposed in the literature (information from the Pediatric Health Information System database and Systematic Reviews) regarding the percentage of children who received bronchodilators (less than 19%), corticosteroids (less than 6%), antibiotics (less than 19%) and X-Ray (less than 32%).

Methods: Retrospective study in children aged <1 year admitted to the Emergency Department (ED) in our institution with bronchiolitis during two RSV seasons.

Results: We analyzed 787 episodes of bronchiolitis. The mean age was 4.7 ± 3.1 months; 469/778 (60.28%) had an increased respiratory rate, 477/787 (60.61%) had moderate/severe retractions, and the SpO2 at admission was 96.7 ± 3.6%. At the ED, 179/787 (22.7%) received bronchodilators, 2/787 (0.2%) received inhaled corticosteroids, 28/787 (3.5%) received oral corticosteroids, 60/787 (7.6%) received antibiotics, and 67/787 (8.5%) had an X-Ray. Also, of the 457 patients under the age of 6 months, 17 (3.7%) received bronchodilators.

Conclusions: We respected the achievable benchmarks of care proposed in the literature for the management of bronchiolitis. However, as these medical interventions are not routinely recommended by the American Academy of Pediatrics more can still be done to further decrease their use and comply with better practice recommendations.

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Benign transient hyperphosphatasemia; nothing to worry about?

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Background: Benign transient hyperphosphatasemia (BTH) is a condition in which serum alkaline phosphatase (ALP) is transiently elevated in the absence of other detectable systemic pathology. The increase may be marked and reach values around 5000 U/l with a return to normal values within a few months. BTH is rarely seen in adults and occurs mainly in infants and children. Its prevalence reaches up to 6% in children below 2 years of age. Important differential diagnoses include liver, bone and kidney disease, as well as hematological disease and acute bone injuries. The origin of BTH is obscure. In the literature an immature ALP clearance mechanisms is postulated as a possible source.

Case description: We describe two cases with similar presentation and course. Patient A was hospitalized at the age of 18 months for the investigation of mental retardation. ALP was markedly increased at 4437 U/l (normal range: 142-335 U/l) without clinical or laboratory signs (complete blood count, aspartate aminotransferase, alanine aminotransferase, total and direct bilirubin, gamma-glutamyl transferase, calcium, phophorus, 25-hydroxyvitamin D, parathyroid hormone, creatinine) of an underlying condition. Within a few weeks the ALP declined to a normal value of 188 U/l. The mental retardation is thought to be of genetic etiology. Patient B was referred to our endocrinological outpatient clinic. The elevated ALP was incidentally found during a routine investigation at the pediatrician's office. In patient B there were neither clinical nor laboratory signs of an underlying condition. Within a few weeks the ALP declined to 330 U/l which is in the normal range.

Conclusion: We describe two cases with the classical presentation of BTH. In accordance to literature the two patients were detected between September to November, the elevation of ALP reached a 10-fold higher value than the pediatric upper reference and the patients were under 2 years of age at presentation. From the sole value of the elevated ALP no conclusions to its origin can be drawn. In case of elevated ALP the above mentioned differential diagnoses should be considered and excluded. Once no causative condition can be found and a rapid decline of the ALP is observed diagnosis of BTH can be made. Hence unnecessary procedures and concerns can be avoided.

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Late decompressive hemicraniectomy in a child with malignant stroke following severe myocarditis and ECMO: never too late!

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Pediatric stroke is a rare condition associated with significant morbidity. Diagnostic delay is a major point preventing form optimized manage-

ment of stroke in children. Decompressive hemicraniectomy is rarely reported but could be a lifesaving option in children with stroke and malignant edema.

We report the case of a previously healthy 2-years-old girl, who present with enterovirus myocarditis, complicated by cardiac dysfonction, multiple episodes of arrhythmia and cardiac dysfonction who required extracorporeal membrane oxygenation (ECMO). Under ECMO, a left hemisyndrome was progressively observed and subsequently a right eye anisocoria was noticed. A possible cerebrovascular event was considered, but imaging was postponed to allow for decannulation and removal of pace wires. Brain MRI performed 48 h after symptoms onset showed a right middle cerebral artery stroke with malignant edema, on-going uncal engagement and secondary bilateral posterior ischemic stroke. Despite the delay in diagnosis, it was decided in agreement with the family to perform an urgent decompressive craniectomy. After identification of a large thrombus in the left cardiac ventricle and aortic root a surgical thrombectomy was also performed and the child was treated 3 months with anticoagulation.

Early in the course, the child exhibited cerebral visual impairment with absent fixation, bilateral opthalmoplegia and flaccid left-sided weakness. An intensive rehabilitation program was initiated. On follow-up ten months later, the child is able to make independent steps, has fully recovered vision and eye movements, and is producing two-word sentences. She has a persisting left upper limp palsy with limited distal movements. She had developed focal epilepsy, initially poorly responsive, and now under remission under two antiepileptic drugs.

This case serve to illustrate the rare occurrence of malignant stroke in children, who are at risk being less prone to accommodate brain swelling having less physiological brain atrophy than their adults counterpart. It also emphasize the difficulty to detect and to manage perioperative stroke in the intensive care unit because of sedations and technical obstacles. As shown in adults, decompressive craniectomy might improve outcome in children with malignant stroke and delayed craniectomy is probably still therapeutic.

Source: Management of stroke in neonates and children, Stroke 2019; Ferriero et al.

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A rare presentation of Rett syndrome

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Background: Rett syndrome is a neurodevelopmental disorder affecting primarily females and results in profound cognitive and physical disabilities. Around 80% of the patients develop epileptic seizures in the first 5 years after diagnosis. Continuous spike-waves during slow-wave sleep (CSWS) syndrome is an electroencephalographic term describing near-continuous epileptic activity in non-rapid eye movement (non-REM) sleep associated with neuropsychological regression. Possible causes include structural and non-structural pathologies. CSWS syndrome is described in only few cases as a presenting symptom of Rett syndrome.

Case description: We describe the case of a patient referred to our neuropediatric outpatient clinic at the age of 3 years. The girl is the first child of non-consanguineous parents and presented with language regression and autistic behavior. The rest of the neurological exam was normal. Due to the language dominant regression an electroencephalogram (EEG) during sleep was performed and detected a CSWS syndrome. The CSWS syndrome was refractory to antiepileptic therapy consisting of Levetiracetam, Clobazam, pulsatile Dexamethasone and ketogenic diet. At the age of 3.5 years secondary microcephaly was noted on clinical examination and hand stereotypies were observed shortly after. Genetic testing came back positive for a pathogen MECP2 mutation and diagnosis of Rett syndrome was made. Up to date no clinical detectable seizures were noted.

Conclusion: This is a very rare presentation of Rett syndrome. Epilepsy is a common feature of this congenital disease, whereas CSWS syndrome as initial clinical presentation is only described in few cases. Furthermore, normal global motor skills and in particular normal hand function up to the age of almost 4 years is uncommon for patients with Rett syndrome. According to the current diagnostic criteria of Rett syndrome, our patient can be classified as an atypical variant, whereat further classification is not possible (e.g. Zappella variant, Hanefeld variant). This case reflects the importance of thorough clinical examination as well as early genetic investigation. Although no causative treatment for patients with Rett syndrome is available, timely diagnosis helps to avoid unnecessary examinations and prevents extended uncertainty.

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A strange case of stridor

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Background: stridor is the main sign of vocal fold paralysis in childhood and its causes may be neurological, anatomical (tracheoesophageal fistula, aneurism, etc), idiopathic and birth trauma. The most common neurological cause is the Chiari malformation and it usually manifests itself in the 3rd month of life.

Case: A 13 months old boy with a 10 months history of dysphagia (mainly for liquids), poor weight gain and recurrent episodes of airways infections was addressed to our department where Celiac disease, IBD, cystic fibrosis, cardiac or endocrinological problems had been excluded. No anatomical alterations were found in the contrast esophagogram; EGDS and pH-impedancemetry were unremarcable. A fibroscopy, performed because of worsening stridor and hoarseness, revealed a bilateral vocal cord paralysis. Radiological differential diagnosis of recurrent laryngeal nerve palsy included MR of brain, spine and mediastinum and CT of the cervical spine. Chiari I malformation and platybasia, with downward displacement of cerebellar tonsils through the foramen magnum, causing bilateral compression of the emerging roots of the IX and X cranial nerves, were detected. Hydrocephalus and syringomyelia were excluded. Polysomnography revealed signs of central respiratory dysregulation. Neurosurgical extradural decompression of the posterior fossa and C1 laminectomy were performed, resulting in a progressive improvement of neurological symptoms in the early follow-up.

Discussion: Chiari malformations are an heterogeneous group of congenital disorders, defined by downward displacement of the cerebellum alone or with other anatomical anomalies and can be clinically associated with lower cranial nerve palsy. Detection of symptoms in children is insidious and related to oropharyngeal dysfunction.

In our patient, all the symptoms appeared justified by structural cranial nerve and lower brainstem compression with hoarseness, stridor, dysphagia and recurrent aspirations presenting as main clinical signs. The management of symptomatic CM-I with lower cranial nerve palsy is surgical decompression often burdened by intra and post-operative complications and by a high rate of recurrences.

Conclusions: Although its rare clinical onset during early childhood, Chiari I Malformation should be considered for differential diagnosis of stridor and dysphagia. Clinical presentation may be slowly progressive, hindering the diagnostic process; surgical intervention is the mainstay of therapy.

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Neuropsychological profile in a patient with Coffin-Siris-Syndrom: a case report

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Coffin-Siris syndrome (CSS) is a very rare genetic disorder that occurs worldwide with no ethnic predisposition. Until now approximately 200 cases have been documented.

The disorder is characterized by coarse facial features, hypertrichosis, hypoplastic to absent fifth fingernails or toenails and agenesis of the corpus callosum might occur. In these children mild to severe intellectual disability have been reported. Affected children may also have speech delays, where the expressive language seems to be more affected than the receptive language, as well as attention problems/hyperactivity and delays in motor development.

Here we describe a 5 years and 7 month old patient with a newly diagnosed CSS with a mutation in the ARID1A-gene that underwent neuropsychological testing. Intelligence was measured using the K-ABC II and the expressive language abilities performing the P-ITPA. To measure the graphomotor skills the subtest "patterns" from the SON-R 2 ½ - 7 was used. To assess behavioural difficulties and attention problems the VBV 3-6 and DISYPS III questionnaires were handed out to the parents as well as the kindergarten teacher.

Results: showed an intellectual profile in the range of a learning disability with a firm weakness in the expressive language, the verbal short-term memory as well as graphomotor skills. On a behavioural level, the patient was seen to be easily distracted which was in line with the information of the questionnaires that pointed out difficulties in the area of attention problems and motor restlessness.

Altogether these neuropsychological findings are consistent with the previous literature pointing out strong speech difficulties, behavioural and motor problems, even though in our case, the overall cognitive impairment seems to be milder. These data underline the importance of cognitive testing then, besides somatic stigmata, a neuropsychological profile can be helpful in detecting rare genetic disorders.

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Experience with cannabidiol (CBD) in the multimodal management of children and young adults with Cerebral palsy (CP) and complex comorbidities.

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Introduction: The «Center of Developmental and Pediatric Neurorehabilitation» (C.D.N) in Switzerland is a neuropediatric competence centre for children and adolescents diagnosed with acquired and innate brain pathologies. The aim of our experience-based study is to describe the experience in our centre with a regimen of medical cannabidiol oil. CBD is a non-psychotomimetic substance with a promising therapeutic potential in different medical conditions.

Patients and methods: The included patients were diagnosed severe spastic CP, refractory epilepsy and different comorbid conditions such as chronic pain, sleep disorders, irritability and nausea.

The selected formula contained CBD in sunflower oil for a sublingual application.

Clinical effects were assessed with semi-quantitative questionnaires.

Results: Promising results were reported in management of the refractory epilepsy, with two patients who became seizure-free. Unsatisfactory results were observed concerning decreasing of severe spasticity.

Very good results were shown regarding reduction of irritability, pain and nausea and amelioration of sleep quality.

Conclusion: Literature shows strong evidence for clinical benefits of CBD in different medical conditions. Our experience with CBD on a population of children and young adults with CP and complex comorbidities is varied and depends on the symptom/disease treated. In the long term therapy ineffective results were shown in decreasing of severe spasticity as opposed to very good results in controlling the number of seizure in patients with refractory epilepsy. CBD appears as well as a good ally in clinical management of additional symptoms (irritability, pain, sickness and sleep disorders) in multiple comorbidity patients, improving quality of life.

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Children born with Hypoxic-ischaemic Encephalopathy have specific deficit in attention

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Background: It is known that children, born with Hypoxic-ischaemic Encephalopathy (HIE), have a risk for neurodevelopmental disorders, particularly for delay in development of executive abilities. However, it is important to investigate the specific impact of HIE on executive abilities in different stage of development. The goal of this research was to examine the hypothesis that children at the age of 5–6, born with HIE, have a specific deficit in sustained attention.

Participants and methods: The experimental group included 20 children aged 5–6 years (mean age = 5.3). They were born full-term with perinatal Hypoxic-ischaemic Encephalopathy. The control group included 20 typically developing children. The children from experimental and control group were matched for gender and age. Executive abilities of children from both groups were assessed by 5 subtests from NEPSY (Auditory Attention and Response Set, Visual Attention, Design Fluency, Statue, Tower).

Results: One-way ANOVA has revealed significant ($p<.05$) group differences in 3 subtests from NEPSY (Auditory Attention and Response Set, Visual Attention, Statue). These subtests are designed to assess the attention, specifically sustained attention. However, we have not revealed significant ($p<.05$) differences between children from experimental and control group in Design Fluency and Tower subtest.

Conclusion: In view of the obtained results it can be assumed that the Hypoxic-ischaemic Encephalopathy has a specific (not global) negative effect on development of executive abilities in children at the age of 5-6 years. Children, born with HIE, have deficit in sustained visual and auditory attention.

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Specific deficit of memory in preschool children with ADHD

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Background: It was shown that children with ADHD have deficit in prefrontal cortex functions, including deficit in working memory (Martinussen et al., 2012). In our previous research we have revealed that ADHD children at the age of 8-9 years have deficit in memory in delayed recall condition (Kiselev et al., 2017). The goal of this research was to examine the hypothesis that preschool children with ADHD have the same deficit in memory in delayed recall condition.

Methods: The experimental group included 16 children with ADHD at the age of 5-6 years. The control group included 16 typically developing children. The children from experimental and control group were matched for IQ, gender and age. Children from both groups were assessed with visual memory subtest from Luria's neuropsychological assessment battery. This subtest is designed to assess the ability to perform visual memory for objects in immediate and delayed recall conditions. Two-way ANOVA was used to reveal group differences in reproducing the objects in two conditions.

Results: We have not revealed significant differences between children from experimental and control group in reproducing the objects in immediate condition. However, the interaction of condition type and group was significant ($p\leq0.05$). ADHD children were less successful in reproducing the objects in delayed recall condition.

Conclusion: In view of the obtained results, it can be assumed that preschool children with ADHD have specific deficit in memory, specifically they have impairment in memory in delayed recall condition. However, we need to do further research for understanding cognitive impairments in children with ADHD.

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Preschool children can benefit from maternal mindfulness training during pregnancy

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Background: It is known that maternal anxiety during pregnancy can affect child outcomes. It was shown that mindfulness training during pregnancy can reduce the anxiety of mother. It can be assumed that low level of maternal anxiety during pregnancy can have positive effect on the child development. The goal of this study is to evaluate the effect of the maternal mindfulness training during pregnancy on executive abilities in 5 years old children.

Methods: In current study we included 22 women who participated in the maternal mindfulness training during pregnancy. The control group

included 22 women who did not participate in this training during pregnancy. When the offspring of the target pregnancies were 5 years of age, their executive abilities was assessed by 4 subtests from NEPSY (Tower, Auditory Attention and Response Set, Visual Attention, Statue).

Results: One-way ANOVA was used to reveal group differences in performing executive tasks. We have revealed the significant differences ($p<.05$) between groups in 3 subtests from NEPSY (Tower, Auditory Attention and Response Set, Visual Attention). The children from the experimental group had better results in these subtests.

Conclusion: The received results suggest that maternal mindfulness training during pregnancy can have positive effect on neurocognitive development of children, particularly executive abilities can benefit from this training. We are going to investigate the long-term effect of maternal mindfulness training on the further neurocognitive development of children.

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Body-oriented training has positive effect on executive abilities in children with ADD

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Background: it is known that children with ADD have deficit in executive abilities. The goal of this study was to reveal effect of body-oriented training on executive abilities in ADD children. Particularly we compared the efficacy of two methods of training (body-oriented training for children vs. conventional motor exercises) in a randomized controlled pilot study.

Methods: 20 children with ADD between 6 to 7 years of age were included and randomly assigned to treatment conditions according to a 2×2 cross-over design. The body-oriented training included the exercises from yoga and breathing techniques.

To assess the executive functions and attention in children we used 5 subtests from NEPSY (Tower, Auditory Attention and Response Set, Visual Attention, Statue, Design Fluency). Effects of treatment were analyzed by means of an ANOVA for repeated measurements.

Results: The ANOVA has revealed ($p<.05$) that for all 5 subtests on executive functions and attention the body-oriented training was superior to the conventional motor training, with effect sizes in the medium-to-high range (0.39-0.87).

Conclusions: The findings from this pilot study suggest that body-oriented training can effectively influence the executive abilities in children with attention deficit disorder. However, it is necessary to do further research for revealing the impact of body-oriented therapies on the prevention and treatment of ADD in children.

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Acceptance of universal varicella vaccination among Swiss pediatricians and GPs who treat pediatric patients: Results: of an internet survey

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Background: Over the last two decades, several countries have initiated universal varicella vaccination (UVV) programs in infants. For countries starting such programs, there is a theoretical concern that suboptimal vaccination uptake could lead to a shift in the varicella incidence resulting in a higher burden of disease in older age groups, and potentially impacting complication rates. To achieve a high vaccination coverage, it is important that the relevant physicians show a high acceptance of a recommendation for UVV.

We studied the perception of varicella disease and the current vaccination behavior among Swiss pediatricians and general practitioners (GPs) who treat children. We also assessed their intention to advise parents to vaccinate their children against varicella in the event the Ministry of Health (MoH) recommends UVV.

Methods: Primary data was collected through a structured, 20-minute online survey with Swiss pediatricians and GPs who treat children. Eligible participants were identified through predefined screening questions and recruited by QualiPro, an agency specialized in Swiss healthcare professional recruitment.

Results: The questionnaire was sent to 1'208 GPs and 1'054 pediatricians and completed by 150 (6.6%) physicians: 40 GPs in the German-speaking part, 20 GPs in the French-speaking part, 67 Pediatricians in the German-speaking part, and 23 Pediatricians in the French-speaking part.

The majority (64%) of all participants reported that they currently recommend varicella vaccination for risk groups according to the national immunization plan. About one third of physicians (35%) – predominantly pediatricians – currently recommend it for all infants. In these situations, a MMRV combination vaccine is currently used by 58% for the first dose and by 59% for the second dose.

86% of participants stated that they would advise parents to have their children vaccinated against varicella in case of a recommendation for UVV by the MoH. 68% responded that they expect many questions from parents and 65% agreed that they have good arguments to convince parents of the importance of varicella vaccination.

Conclusions: We showed that the majority of Swiss physicians in this study have a high intention to vaccinate universally against varicella in case of an official UVV recommendation by the MoH. However, arguments in favor of UVV would need to be made available broadly to support physicians and achieve high coverage of varicella vaccination.

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Multifocal symmetrical osteomyelitis in a healthy 10-year-old boy

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Osteomyelitis more frequently affects young children (less than 5 years) and is commonly localised in the lower extremities. Multifocal osteomyelitis is only seen in 5-10% of cases, especially in newborns and infants. We present a case of multifocal osteomyelitis with favorable outcome in an older child without any known risk factor.

Case presentation: A 10-year-old boy consulted with a complaint of 6 days of pain, swelling and redness in both ankles, initially noted in the right and then bilaterally. He was also febrile at 40°C during this period. Physical examination showed swelling, erythema and warmth over the left lateral malleolus. Weight bearing was not possible on the left lower limb. Laboratory tests revealed a leucocytosis (14,5/mm3) and raised inflammatory markers (CRP 98 mg/dl, ESR 70mm/h). Lower limb magnetic resonance imaging revealed bone marrow oedema throughout the distal third of both tibias and right fibula, as well as soft tissue infiltration over the left lateral malleolus. There was no sign of arthritis nor a significant collection. Three blood cultures taken over 48 hours resulted positive for MSSA. Parenteral antibiotic therapy was promptly initiated. During the hospitalisation, the laboratory test results and the clinical condition showed signs of improvement. Complications such as deep vein thrombosis and endocarditis were excluded by ultrasound. Consequently, antibiotic therapy was switched to oral administration after 3 weeks of intravenous therapy and was stopped 4 weeks after the first negative hemoculture. The follow-up visits showed complete recovery, without significant effect on either growth or mobility.

Discussion Osteomyelitis is rarely seen in more than one bone simultaneously. Given the age of our patient and the peculiar symmetrical pattern of the lesions, our differential diagnosis was wide including rheumatological (CRMO) and tumoral causes. Concerning the risk of a bacterial infection in our patient, he did not have a relevant past medical history nor a history of acute trauma; he did not present signs of upper respiratory tract infection, nor any wound. We were able to reasonably exclude diabetes, sickle cell anemia, leukemia and an immunological deficit by laboratory tests. Considered patient's usual physical activity (rugby), repetitive microtrauma was the only identifiable risk factor.

Conclusion: Our aim is to highlight the incidence of severe infections in older children and question our knowledge of risk factors.

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Polymicrobial endocarditis due to Coxiella Burnetii and Triadelphia pulvinata

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Case report: A 13-year-old Moroccan boy, known for Tetralogy of Fallot and right ventricular outflow tract (RVOT) surgery at 19 months of age, was referred to our institution because of severe stenosis of his pulmonary bovine jugular valved conduit (Contegra). He presented with dyspnea, fever, weight loss, fatigue, hepatosplenomegaly and pancytopenia. After an extensive workup, serologies confirmed chronic Q fever and we initiated Doxycycline and Hydroxychloroquine. His Contegra conduit was replaced with a Biopulmonic conduit. Specific PCR and culture of his tube were positive for Coxiella burnettii, confirming our diagnosis of Q fever endocarditis. Following surgery, he presented with recurrent fever and dyspnea, prompting a chest MRI and PET-scan. Imaging revealed mediastinitis with a collection extending around the RVOT and the patient underwent replacement of his tube by an aortic homograft. The culture of the collection, the tube and blood cultures were sustainly positive for Triadelphia pulvinata, which was treated with Amphotericine and Voriconazole. Finally, metagenomic sequencing confirmed that Triadelphia pulvinata was already in the initial surgical specimen alongside Coxiella burnettii, approximately one month prior to the diagnosis of Triadelphia pulvinata infection, confirming a polymicrobial endocarditis. The patient improved and was transferred to his country with Doxycycline, Hydroxychloroquine, Amphotericine and Voriconazole.

Discussion Q fever is a zoonosis caused by intracellular Gram-negative bacteria, Coxiella burnetii. Animals are the main reservoir and transmission occurs mostly by inhalation of contaminated aerosols. Acute Q fever is usually presented by flu-like illness, granulomatous hepatitis or atypical pneumonia. Chronic Q fever develops in an estimated 1-5% of all infected humans and has high mortality and morbidity rates if left untreated. Both endocarditis and Q fever are uncommon diagnoses in children. The majority of paediatric cases of Coxiella burnetii endocarditis have been associated with congenital heart disease. Treatment consists of combination of doxycycline and hydroxychloroquine for 18-24 moths. Triadelphia pulvinata is a rare dematiaceous fungus found in soil. There are only 3 cases reported in the literature.

Conclusion: Chronic Q fever in children can present with endocarditis, requiring prolonged therapy with antibiotics.

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Chronic recurrent multifocal osteomyelitis involving sacrum and maxilla.

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Background: Chronic recurrent multifocal osteomyelitis (CRMO) is a condition described as polyostotic with a relapsing and remitting course, preferentially affecting the metaphysis of tubular bones in the pediatric population. Inflammatory foci may also involve clavicle, spine and jaw. We report a case of CRMO with unusual localization.

Case presentation: A 12 years old girl was admitted for nocturnal and disabling sacral pain since 4 weeks without history of trauma. Clinical exam was unremarkable except pain at the sacral region. Blood test showed normal white blood cell count and LDH, increase in inflammatory parameters (CRP 37mg/L, ESR 40mm/h) with negative blood cultures. MRI with injection revealed signs of discitis at the level of S3-S4 associated with reactive osteomyelitis of S3 and S4 and a presacral sovrafluid collection. CT-guided biopsy was performed: it showed lymphoytic and histiocytic infiltrate and allowed to exclude infection and malignancy. Pain resolved with NSAID and laboratory values normalized. We suspected CRMO onset with unique localization.

After 2 months the patient developed disabling pain at the right maxillary bone. At the clinical exam we found trismus and a single palpable right submandibular lymph node. Blood test showed normal white blood cell count and increase in inflammatory parameters (CRP 13mg/L, ESR 35mm/h). MRI of facial massive was performed, showing inflammation around the non-erupted third molar, at the right pterygopalatine fossa and maxillary sinus. Because of the atypical localization for a CRMO, we initially suspected an infectious process. Antibiotic therapy with Co-Amoxicilline was started in parallel to NSAID. After an initial improvement, pain worsened and became irresponsive to NSAID and paracetamol. A control by MRI and maxillary CT scan was performed two weeks after, confirming previous MRI findings of edema and better revealing the lytic effect on the bone surrounding the molar pocket. A novel biopsy with curettage allowed to exclude an infectious or malignant process. Total body MRI showed reduction of the sacral lesion without other inflammatory foci. Maxillary pain progressively improved under NSAID and laboratory values normalized.

Conclusions: CRMO is an exclusion diagnosis and can begin as an unifocal disease. In the course of disease, differential diagnosis should be reconsidered in case of unusual localization and atypical radiologic aspect. Is the second case reported with maxillary localization.

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Current management of acute bronchiolitis in Switzerland

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Background: Acute bronchiolitis (AB) is the most common viral lower respiratory tract infection in the first year of life. Despite international guidelines and numerous Cochrane reviews which emphasize that drugs do not alter the natural course of AB, still many drugs are prescribed. A questionnaire on treatment of AB was sent to all Swiss paediatricians before (2001) and after (2006) introducing national guidelines in 2004. The aim of this study was to evaluate the current management of AB and the long-term impact of national guidelines.

Methods: An online questionnaire AB was sent to all Swiss paediatricians to assess their current prescribing practice.

Results: From a total of 2040 questionnaires (1644 registered paediatricians, 396 trainees), 1079 were returned (response rate overall 52.9%). Of the respondents, 908 (44.5%) treated children with AB. Up to 89% of the registered paediatricians are still prescribing bronchodilators in outpatients and 58% in inpatients, but mostly not on a regular basis (see table). Steroids were less prescribed (37% in outpatients, 23% in inpatients), also not on a regular basis. The use of salbutamol and steroids was much lower compared to 2006. Most paediatricians (55%) are using SpO2 of ≤90% as a cut-off to apply oxygen, 36% use ≤92%, and 10% use ≤88%.

Conclusion: Bronchodilators and steroids are still widely prescribed for the treatment of AB in Switzerland, but much less compared to the survey in 2006. National guidelines and regular information can have a long-term impact on prescribing patterns.

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A challenging case identified by the recently implemented Swiss newborn screening for severe combined immunodeficiency

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Early detection of severe combined immunodeficiency (SCID) allows taking timely measures to prevent infections and increases chances for a successful definitive treatment with stem cell transplantation. In Switzerland, newborn screening for SCID by measuring T cell receptor excision circles (TREC) from dried blood stains was started on January 1st, 2019. Low levels of TRECs are not specific for SCID and can sometimes even be only a sign of a transient lymphocytopenia making the management of some of the patients identified by the screening very difficult. We present the challenging case of a newborn girl with low TRECs identified by the screening program. The otherwise healthy newborn showed a lymphocytopenia (1.21x109/L) with abnormally low levels of T

cells (0.18x109/L) including low recent thymic emigrants, naive T cells, helper T cells and cytotoxic T cells but normal levels of B and NK cells. These, together with other laboratory results, were consistent with either SCID or DiGeorge syndrome and infection prevention measures were started. A comparative genomic hybridization array revealed a microduplication of approximately 729 kb in chromosome bands 22q11.22 to q11.23, a chromosomal change of unclear significance in this context also found in the newborn's father with frequent respiratory viral infections and profuse skin warts. Whole exome sequence analysis of the patient and the parents did not reveal any known pathological mutations in SCID genes. In order to investigate whether the duplication impacts the expression level of nearby genes (such as TBX1, which is known to be associated with DiGeorge syndrome when mutated), a transcriptomic analysis is being performed. To distinguish between a hematopoietic versus a thymic defect, a T cell differentiation assay on the patient's CD34+ bone marrow cells using a specifically developed cell culture system is being conducted. During the first nine weeks of life, the newborn was hospitalised once for intravenous antibacterial und antifungal therapy due to neonatal cephalic pustulosis with a bacterial superinfection but remained otherwise free from infections. The patient showed neither a clear change of total lymphocyte counts nor of any of the lymphocyte subsets. As the usual genetic studies have remained inconclusive to date and the patient has not suffered recurring severe infections, the definite diagnosis is still unclear and watchful waiting has been adopted while further analyses are ongoing.

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Pregnancy: getting ready for the baby!

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Background: Beckwith-Wiedemann syndrome (BWS) is a congenital overgrowth disorder characterized by rare features such as macroglossia, abdominal wall defects and increased risk for embryonal tumors.

Case: A 37 years old G1P0 was diagnosed at 16 weeks pregnancy with fetal omphalocele. Given the US's findings, amniocentesis and genetic analysis were performed, which revealed a normal 46 XX karyotype and methylation defect on IC2 locus, diagnosing BWS. As planned, for the presence of the omphalocele, the mother was referred to a tertiary hospital.

At 34+2 weeks' gestation, the patient was born by spontaneous delivery showing normal neonatal adaption with Apgar scores of 7, 9, 10. Physical examination at birth showed macroglossia and 6 cm's omphalocele associated with Wharton's jelly cyst (15 cm diameter). On the first 24 hours, she presented multiple hypoglycemias, minimum at 2.1 mmol/L, promptly managed via parenteral route. On the first day she underwent surgery for omphalocele's correction, appendectomy and resection of perforated Meckel's diverticulum with termino-terminal anastomosis. Following the surgery, she benefited of total parenteral nutrition for 18 days.

Being she born premature, she developed respiratory distress syndrome in a contest of immature lung remaining intubated for 3 days.

Subsequently CPAP was applied for 6 days. Moreover, as premature, she developed anemia with haemoglobin minimum at 88 g/L and therapy with Aktiferrin 4 mg/kg was started.

Complementary were performed a cardiac US, normal, and brain US which evidenced grade I bleeding. The following brain US showed 2 millimetric cystic formations compatible with post-haemorrhagic outcomes. On macroglossia, thanks to appropriate physiotherapy sessions, on discharge she is fed just by baby bottle.

She could go home without further complication and she is having, since then, strict follow-ups. Abdominal US and alpha-fetoprotein levels will be performed every three months for 3 years, then every 6 months up to 6 years, then yearly in order to detect potential malignancy in time.

Conclusion: BWS is a rare disorder commonly diagnosed in the postnatal period. Our case depicts how, using prenatal imaging, BWS may be detected before delivery and transfer, in utero, to a tertiary center. Moreover, children with BWS are more risk for tumor devolpment with a range of risk between 4% and 21% varying on the genetic mutation; so prenatal diagnosis of BWS is key to start malignancy screening on time.

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Hypogonadotropic hypogonadism or constitutional delay? An oligogenic pattern of inheritance with complex malformations in one family

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Background: Congenital hypogonadotropic hypogonadism (CHH, termed Kallmann Syndrome in the presence of anosmia/ hyposmia) is a rare disease with a heterogeneous clinical picture and genetic background. Many investigations concerning the affected genes, which are acting alone (monogenetic) or in combination (oligogenetic), have been done, so that mutations can be found nowadays in up to 50%.

Case presentation: A term born boy showed a cleft lip and palate. At routine audiologic examination congenital deafness was found left. Speech development was normal until the age of 4 years, when speech expression went to be unclear and deafness of the right ear was also diagnosed and he received a cochlear implant. Although at the age of 6 years, school physician found retractile testis left and small phallus, no further referral was done. Only at the age of 16 years the boy presented with a familial constitutional delay (adrenarche only with 15 years, no growth spurt, bone age 14 years) at our clinics with pubertal stage P2, G1 (left testis inguinal, right descended, small phallus <-2 SD), ax.1. By LHRH and HCG test we diagnosed a hypogonadotropic hypogonadism and testosterone injections were started. The patient responded well, also with an increasing testis volume up to 6 ml over the next 3 years. Family history was conspicuous for a complex CHH (our patient was son 3): delayed puberty in the parents and the 4 youngest sons, cleft lip/palate in the father and the 4 youngest sons, oligotonia in the 3 last sons, cryptorchidism in the 3 middle sons, renal duplication in the 4th son, hearing impairment in the 3rd son. Therefore, further genetic tests were done.

Results: A monoallelic NOS1 mutation was found in the father and the three middle sons, consistent with an autosomal dominant mode of inheritance, but so far only our patient had CHH. This can be explained by having inherited different delayed puberty genetic backgrounds of the father (NOS1 mutation) and the mother (other unknown mutation) showing the oligogenetic mode of inheritance.

Conclusion: Pedigree drawing can be essential in finding the correct diagnosis. Searching the mutation(s) of CHH is promising and important. NOS1 (8% incidence in CHH) is next to FGFR1 and CHD7 the most frequently mutated gene in CHH patients. Investigations for causal treatment of gonadal insufficiency with nitric oxide synthase inhibitor in a cryptorchid murine model are running. (DeFoor J Urol. 2004).

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Double trouble – New-onset type 1 diabetes and encephalitis following hand-foot-mouth disease in a toddler

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Introduction: Enteroviruses are common pathogens in the paediatric population and cause a variety of diseases such as infectious meningoencephalitis. Virus-induced diabetes has also been described. In particular coxsackie viruses have been linked to autoimmune destruction of pancreatic cells.

Case report: An 18-month old boy was admitted with a history of fever and vomiting for 2 days resulting in moderate dehydration. A maculopapular rash on hands and back was interpreted as hand-foot-mouth disease. Despite oral rehydration he remained in reduced condition and progressively started hyperventilating. Hyperglycaemia of 19 mmol/l confirmed the suspicion of new-onset diabetes with mild ketoacidosis (pH 7.30). Diabetes management was performed according to the clinics protocol. Despite initial clinical improvement with declining glucose levels, fever and notable somnolence persisted for which he was transferred to the intensive care unit. Neuroimaging was normal without signs of cerebral edema. Lumbar puncture with raised cell count (25/µl

leucocytes, 19% plasma cells) and EEG were indicative for meningoencephalitis, which was empirically stated as viral since neither liquor PCR nor analysis of genome showed any pathogen. Antiviral treatment was given until PCR on HSV were negative and initial anticonvulsive treatment was stopped in absence of convulsions. Insulin-dependency remained, leading to the diagnosis of diabetes type 1 despite negative antibodies. The boy had gradually completely recovered after two months without any neurological sequelae.

Conclusion: Here, we observed a viral infection with concurrent new-onset type 1 diabetes and meningoencephalitis. In the acute phase distinction between diabetes and a central process leading to reduced vigilance was difficult. However, the initial blood gas analysis showed a hint of central affection with very low pCO2 (1.68 kPa) compared to acidosis (pH 7.30) indicating hyperventilation not only as a compensatory mechanism but due to central affection. Patients with new-onset type 1 diabetes typically show lower pH values. In the acute treatment of diabetes, brain edema should be suspected as a reason for reduced vigilance, but it is crucial to consider non-diabetes related diagnoses.

This case emphasises the need of further investigations when facing neurological non-improvement or worsening despite adequate treatment.

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Age appropriate reference intervals for eight serum kidney biomarkers in healthy infants, children and adolescents

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Background: The use of kidney biomarkers for early detection of drug-related glomerular or tubular kidney injury in infants, children and adolescents requires age-specific data on reference intervals in a pediatric healthy population. Therefore, we performed a study to characterize serum values for eight kidney biomarkers in healthy infants, children and adolescents.

Methods: A single center prospective observational study was conducted between December 2018 and June 2019. Single serum sample from 142 healthy infants, children and adolescents aged between 0 and 16 years were collected. Statistical analyses for eight kidney biomarkers (albumin, beta 2-microglobulin, beta-trace protein, creatinine, cystatin C, kidney injury molecule-1, neutrophil gelatinase-associated lipocalin, uromodulin) were performed to obtain reference intervals and associations with age, sex and weight were investigated (Pearson correlation, linear and piecewise regression).

Results: Albumin and creatinine increased with age ($p < 0.01$), whereas beta 2-microglobulin, beta-trace protein and kidney injury molecule-1 values decreased with advancing age ($p < 0.05$) in this healthy pediatric study population. Cystatin C showed dependency on sex (lower concentration in females) and decreased with age until reaching approximately 1.8 years; thereafter an increase with age was seen. Neutrophil gelatinase-associated lipocalin and uromodulin did not show any age-dependency.

Conclusion: This study provides age appropriate reference intervals for key serum kidney biomarkers determined in healthy infants, children and adolescents. Such reference intervals facilitate the interpretation of kidney biomarker changes in daily practice, and allow early detection of glomerular and tubular injury in infancy, childhood and adolescence.

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Developmental programming of arterial hypertension in a rat model of intrauterine growth restriction: impaired functionality of endothelial progenitor cells associated to oxidative stress and cellular senescence

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Introduction: Infants born after intrauterine growth restriction (IUGR) are at risk to develop arterial hypertension later in life. The endothelium is a major contributor to arterial hypertension. The endothelial colony forming cells (ECFCs) are critical circulating compounds of the endothelium and have the capacity to regenerate an injured endothelial monolayer. Therefore, an altered status of ECFCs is a marker of endothelial dysfunction and so of vascular disease. We have previously shown a reduced number and impaired angiogenic capacities of ECFCs in the cord blood of infants born with a low birth weight. However, it is not yet identified whether the alterations of ECFCs persist at adulthood and could contribute to the development of arterial hypertension.

Materials and methods: During gestation, pregnant rats received a control diet (casein 23%, CTRL) or a low protein diet (casein 9%) to induce IUGR. Male offspring were studied at 6 months of age: blood pressure was measured by tail-cuff plethysmography; ECFCs were isolated from bone marrow, quantified by flow cytometry, and their capacities to proliferate (BrdU incorporation) and to form a vascular network (matrigel test) were evaluated. Oxidative stress (superoxide anion detection by dihydroethidine), NO production (DAF-2DA) and cellular senescence (beta-galactosidase activity) were investigated. For each parameter, the results are presented as a mean +/- standard deviation and compared with Mann-Whitney U test. The significance level was set at $p < 0.05$.

Results: Compared to the CTRL group, IUGR males displayed an increased systolic blood pressure (CTRL vs. IUGR (n = 10)(mmHg): 126.7 +/- 3.6 vs. 143.1 +/-8.7; $p<0.001$) and a decreased number of ECFCs (CTRL vs. IUGR (n = 4)(%): 17.2 +/- 1.6 vs. 12.2 +/-1.5; $p<0.05$). IUGR ECFCs had a reduced ability to proliferate (-40%) and to form a vascular network (length of vascular sprouts (mm), CTRL vs. IUGR (n = 4): 394.9+/-164.7 vs. 260.9+/-204.5; $p<0.05$). These cells had also an increased superoxide anion level (CTRL vs. IUGR (n = 4) A.U: 2.1 +/- 0.4 vs. 5.7 +/-1.16; $p<0.05$), a higher beta-galactosidase activity (CTRL vs. IUGR (n = 4) A.U: 0.9 +/- 0.6 vs. 3.4 +/-1.6; $p<0.001$), and a decreased NO production (CTRL vs. IUGR (n = 4) A.U: 0.6 +/- 0.1 vs. 0.3 +/- 0.1; $p<0.001$).

Conclusion: IUGR induced in rats by a maternal low protein diet leads in adult males to hypertension and to impaired number and functionality of ECFCs associated to oxidative stress and cellular senescence.

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Continuous monitoring of health data with a wearable device in pediatric patients undergoing chemotherapy for cancer – a feasibility pilot study

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Background: Pediatric patients under chemotherapy are at high risk to develop severe infections. Delayed diagnosis and treatment can increase mortality. It has been shown in the ICU setting, that infections can trigger changes of vital signs, like heart rate variability, long before further clinical symptoms. Continuous monitoring with a non-invasive wearable device (WD), may detect such changes earlier than discrete measurements. We aimed to assess the feasibility of continuous monitoring of health data in pediatric patients under chemotherapy for cancer using a WD.

Methods: This single-center pilot study included 20 pediatric patients under chemotherapy for cancer. The WD (Everion®, by Biovotion) was worn on the upper arm or leg, for 14 days. Data was transferred to a

cloud database via a mobile phone app. The primary outcome was acceptable quality of monitored heart rate, during a cumulative duration of ≥18/24h per day on ≥7 consecutive days. Secondary outcomes included other vital signs, side effects, effort for the investigators and acceptability by parents and patients.

Results: Twenty patients, with median age of 6 years (range 3 to 16y; 9 patients <6y), were included. In preliminary data analysis, 8 of 20 patients, from 3 to 16 years, fulfilled the primary outcome. The main reason for non-fulfillment was that the device was not worn, while insufficient data quality was rare. One patient developed a superficial skin lesion after scratching. Side effects reported were irritated skin (n = 3), itching (n = 2) and sweating (n = 7). The effort for the investigators was high due to a time-consuming inclusion process, daily data availability checks, and 52 contacts, mostly for data problems. The most important problem identified was low motivation to wear the WD, mostly due to the lack of a direct benefit for patients and missing feedback by the WD. Eighteen of 20 patients or parents indicated that the Everion® is a suitable device to measure health data in children. Definite analysis, including data comparison with discrete measurements, is ongoing.

Conclusion: This study is the first to show that continuous monitoring of health data by a WD is feasible across the entire age range of pediatric oncology patients, although the predefined feasibility criterion (15 of 20 patients fulfilling the primary outcome), was not reached. The results of this study will influence the design of future WD studies, which aim to identify patterns predicting imminent fever or infection.

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DRESS syndrome - Drug Reaction with Eosinophilia and Systemic Symptoms syndrome

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Introduction: The DRESS syndrome is a rare but potentially life-threatening adverse reaction with cutaneous manifestations and internal organ involvement.

Case report: A 15-year-old teenager is hospitalized for debridement and intravenous antibiotic therapy after infection of a left femur osteosynthesis material. Management is complicated, requiring several antibiotics adaptations. After four weeks, he develops a pruritic febrile maculopapular exanthema with edema of the face and is transferred to the intensive care unit for a hypotensive shock. An anaphylactic versus septic shock is suspected and treatment is started accordingly, with good hemodynamical response. However, the clinical course is unfavorable. The adolescent stays in a reduced general condition with high fever and worsening of his rash which becomes more pronounced, without affecting the mucous membranes, and evolving towards an erythroderma and purpura of the extremities. The biological assessment shows a moderate eosinophilia (max 0.9 G/l) with a slight impairment of liver function. Skin biopsy is performed which shows necrotic keratinocytes and presence of eosinophils in the dermis. A DRESS syndrome is suspected with a RegiSCAR (probability score for DRESS) scored at 4. Upon reception of the results, all current treatments are stopped (vancomycin, meropenem, ibuprofen and acetaminophen). Treatment with high dose solumedrol followed by prednisone 1mg/kg/day is started. The clinical course is favorable, with resolution of rash and fever after five days. We haved planned a follow-up in allergology to help to identify the responsible agent.

Discussion: Dress syndrome is a serious form of cutaneous drug adverse reaction. The overall mortality rate is 10-20%. The time of onset of symptoms is usually delayed by 2 or 6 weeks after the culprit drug is initiated. The most common features are high fever, erythematous morbiliform rash and lymphadenopathy. Skin biopsy typically reveals a perivascular lymphocytic infiltrate in the papillary dermis with eosinophils. The diagnosis is difficult and primarily established on clinical and laboratory abnormal findings.

Conclusion: Prompt diagnosis using clinical criteria, laboratory values and histopathology is imperative. The lack of knowledge of this condition can lead to delay of diagnosis and high risk of mortality.

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The Infantile Hemangioma Referral Score (IHReS): A Validated Tool for Physicians

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Objective: Infantile hemangiomas (IH) are very common; some cases require timely referral and treatment to prevent complications. We developed and validated a reliable instrument for timely and adequate referral of patients with IH to experts by non-expert primary physicians.

Methods: This multicenter, cross-sectional, observational study used a three-stage process: 1) development of the Infantile Hemangioma Referral Score (IHReS) tool by IH experts who selected a representative set of 42 IH cases comprising images and short clinical history; 2) definition of the Gold Standard for the 42 cases by a second independent committee of IH experts; 3) IHReS validation using the 42 Gold Standard cases by non-expert primary physicians.

Results: A total of 60 primary physicians from seven different countries evaluated the 42 Gold Standard cases (without reference to the IHReS tool); 45 primary physicians evaluated these cases using the IHReS questionnaire, and 44 completed re-testing using the instrument. IHReS had a sensitivity of 96.9% (95% confidence interval [CI] 96.1 to 97.8) and a specificity of 55.0% (95% CI 51.0 to 59.0). The PPV and NPV were 40.5% and 98.3%, respectively. Validation by experts and primary physicians showed substantial agreement for inter-rater reliability and intra-rater repeatability.

Conclusions: IHReS, a two-part algorithm with a total of 12 questions, is an easy-to-use tool for primary physicians with the purpose of facilitating correct and timely referral of patients with IH. IHReS may help practitioners in their decision to refer patients to expert centers.

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External therapies of anthroposophical medicine in a children's oncological ward – from a feasibility study to a randomized controlled design

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Introduction: Complementary and alternative medicine (CAM) is of growing interest in parents of pediatric cancer patients. Among CAM, external anthroposophical therapies are particularly attractive, because they enable parents to actively take part in the healing process by learning to perform them. External CAM techniques may serve as an additional supportive therapy. So far there is no study of CAM external therapies in pediatric oncology.

Material and methods: In the present retrospective observational study, data of patients 0 - 18 years old with a diagnosis of tumour and one or more external nursing therapies of CAM was collected in our oncology ward between 2015 and 2018. The following therapies were offered: Aurum Lavandula comp. chest compress, Oxalis Folium 20% abdominal compress, Plantago and Cera flawa compress and Homunculus foot massage. Additionally, an evaluation questionnaire was completed by the nursing staff before and after the study.

Results: Out of an average of 50 patients per year with oncological inpatient care, 12 patients in 2015, 21 patients in 2016, 21 patients in 2017 and 14 patients in 2018 underwent one or more CAM external nursing applications. The following effects were documented as observed by the nurses: relaxation, falling asleep and reduction of pain. No side effects were observed. Data collected from the nursing staff on feasibility of CAM on a pediatric oncological ward and its influence on the process of care will be presented.

Conclusion: While the present observations show the feasibility of CAM nursing external application on a pediatric oncological ward, they also highlight the ethical and scientific challenges of studies in seriously ill children. As next step, we will perform a randomized study in oncological hospitalized patients with 3 treatment groups: 1. CAM nursing external application, 2. standard massages with almond oil and 3. regular standardised talks between patients and nurses. The duration of supportive treatment will be the same in each group. Before and after treatment, respiratory and pulse rate, blood pressure, skin conduction and cortisol level in saliva and blood will be measured, pain will be scored using a visual analogue scale and a questionnaire completed by patients and/or parents. Statistical analysis will evaluate, whether CAM nursing external applications have a specific impact on pain and stress levels of children under oncological treatments.

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