SMW
Established in 1871

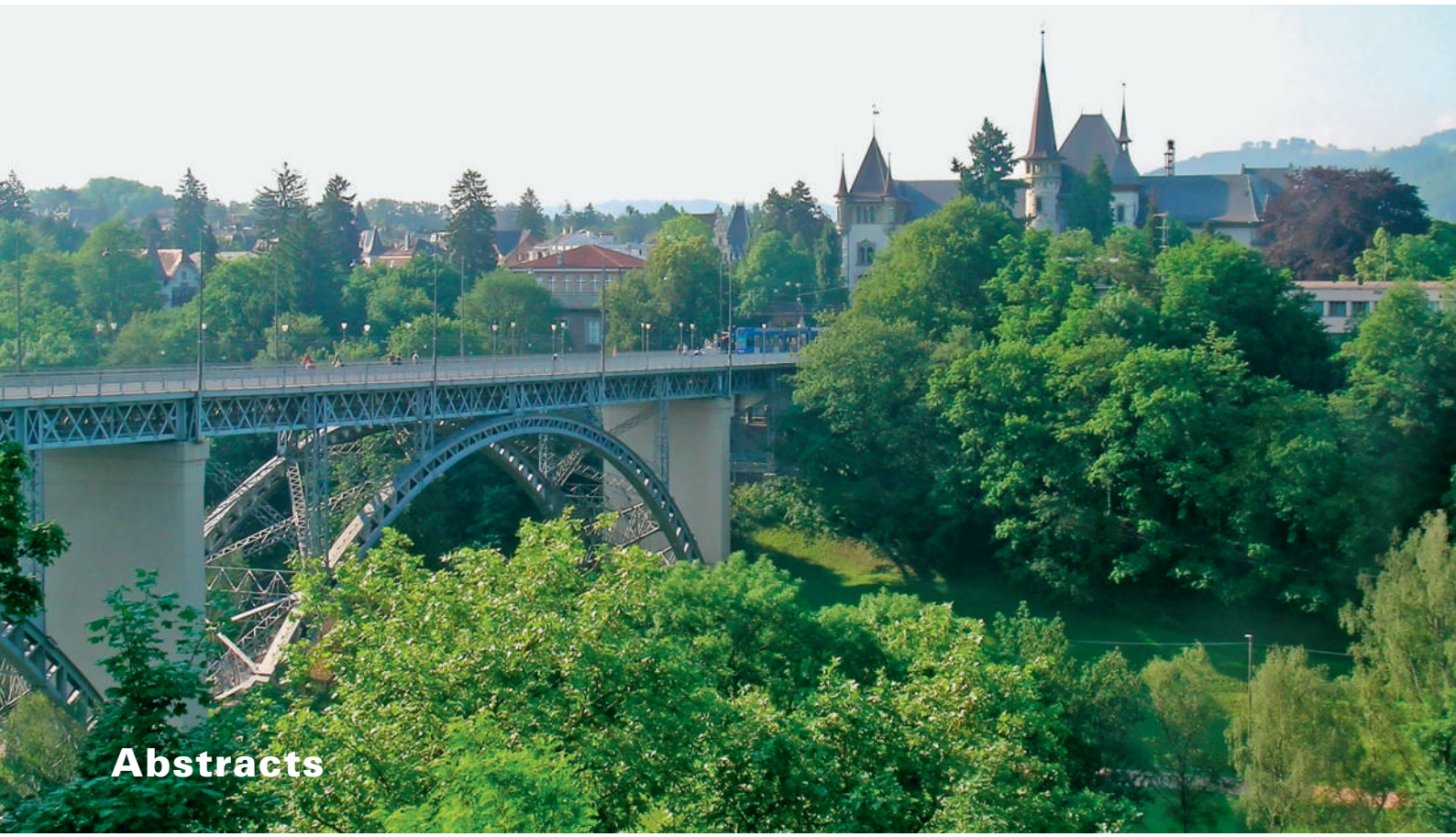
Swiss Medical Weekly

Formerly: Schweizerische Medizinische Wochenschrift
An open access, online journal • www.smw.ch

Supplementum 215
ad Swiss Med Wkly
2016;146
May 25, 2016

Annual Meeting of the Swiss Society of Paediatrics (SSP)

Bern (Switzerland), June 9–10, 2016



Abstracts

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CH-4132 Muttenz, Switzerland
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Submission to this journal proceeds totally on-line: www.smw.ch

ISSN printed version: 1424-7860
ISSN online version: 1424–3997

printed in switzerland

FM 1

Thrombolysis and thrombectomy in children with acute ischaemic stroke

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Background: Thrombolysis is an established treatment in adults with arterial ischaemic stroke (AIS). Case reports suggest, that thrombolysis might be effective in childhood stroke, however, larger studies are lacking. This study describes the effect of thrombolysis/thrombectomy (TT) on outcome in children with AIS.
Methods: Prospective case series of consecutive pediatric AIS patients receiving TT between January 2000 and December 2015. Outcome was assessed using the Paediatric Stroke Outcome Measure (PSOM) at discharge and after 6 months. TT-patients were pedNIHSS- and age-matched with AIS patients receiving standard treatment (ST). The sign test assessed the difference of PSOM in the paired samples.
Results: 15 TT-patients (9 boys, 60%) were compared to a total of 19 ST-patients (9 boys, 47%). Median (range) age and pedNIHSS at stroke onset were 11.9 years (1.5–15.3) and 13 points (7–28) respectively. Median (range) time to diagnosis/treatment-decision in the TT and ST group was four (1–48) and 24 (1–160) hours respectively. I.v. thrombolysis was performed in five, i.a. thrombolysis in four and thrombectomy in six patients. A total of 2 patients per group died during the acute episode (TT: 13%; ST: 10.5%). Median (range) PSOM at discharge and 6 months was 2.5 (0–8) and 1.75 (0–7) in the TT-group and 3.75 (0–10) and 2.0 (0–7) in the ST-group respectively ($p = 0.27$; $p = 1.0$).
Conclusions: Although there is a clinical difference in outcome between the two groups, the results did not reach statistical significance. Larger, prospective studies are needed to identify the subgroup of AIS patients that might benefit best from TT treatment.

FM 2

Additional corticosteroid treatment improves outcome in pediatric stroke due to focal arteriopathy

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Background: Focal cerebral arteriopathy (FCA) accounts for up to 35% of arterial ischemic stroke (AIS) in children, and is the most important predictor of stroke recurrence. The study objective was to compare outcomes for children with FCA treated with combined corticosteroid antithrombotic treatment (CAT) to those receiving antithrombotic treatment (AT) alone.
Methods: This multicenter retrospective Swiss/Australian cohort study analyzed consecutive children, aged 1 month-18 years, presenting with first AIS due to a FCA, from 1999–2014. Children with CAT were compared to those treated with AT. Primary outcome was the neurological deficit at 6 months post AIS, as measured by the Pediatric Stroke Outcome Measure (PSOM). Secondary outcomes included resolution of stenosis and stroke recurrence.
Results: 73 patients (51% males) were identified, 21 (29%) of whom received CAT. Mean (SD) age at stroke for the entire group was 7.9 years (4.7). Median (IQR) pedNIHSS was 3 (2.0–8.0) in the CAT-group and 5 (3.0–9.0) in the AT-group ($p = 0.098$). Median (IQR) PSOM 6 months post AIS was 0.5 (0–1.5) in the CAT-group, compared to 1.0 (0.5–2.0) in the AT-group ($p = 0.035$). Complete resolution of stenosis at last MRI was noted in 17 (81%) in the CAT-group compared to 24 (59%) in the AT- group ($p = 0.197$). Stroke recurrence occurred in one patient in each group.
Conclusion: Corticosteroid treatment appears to provide additional benefit over antithrombotic treatment for improved neurological outcome in childhood AIS due to FCA. Larger prospective studies are warranted to further investigate these differences and understand mechanisms by which steroids modify outcome.

FM 3

Mini-puberty as a window of opportunity in the investigation of congenital cryptorchidism

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Cryptorchidism is defined as the presence of the testicles in an extra-scrotal position.Testicular descent requires normal hormonal activity during fetal masculinization, hypothalamo-hypophyseal axis integrity, and is also influenced by environmental factors. Etiological workup remains a challenge. An 11-month old infant is addressed to our Endocrinology Unit for a pre-operatory evaluation of a persistent bilateral congenital cryptorchidism. Ultrasound confirmed the presence of the two testicles in an extra-scrotal position with the left testicle lying on the inner inguinal ring and the right testicle on the superficial inguinal ring. There is no hypospadias or micro-penis nor other dysmorphic signs. The maternal medical history reveals an uncomplicated pregnancy and no drug consumption. She had menarche at 16 years. Her father developed testicular cancer at 20 years after operation of cryptorchidism at 13 years. His twin brother developed testicular cancer at 40 years. The patient was born full term and eutrophic; adaptation to extra-uterine life was excellent. The first clinical examination revealed a bilateral cryptorchidism without other associated malformations. During the neonatal period the patient did not develop hypoglycemia or prolonged jaundice. Developmental milestones and growth were normal. Paraclinical investigation before orchiopexy showed normal inhibin, anti- Müllerian hormone values, and high testosterone levels after HCG stimulation which confirms normal Sertoli-cell and Leydig-cell function. Androgen receptor analysis, urinary steroid profiling and karyotype are on the way. Testicular US examination revealed micro-calcification, which in this family context could be the first sign of malignancy. The eventual diagnosis of hypogonadotropic hypogonadism (HH) in the context of delayed puberty in the mother will be delayed until puberty. Mini-puberty is the hormonal surge of gonadotropins and testosterone which occurs between 6 weeks and 6 month of life. It constitutes an indicator of the Leydig-cell function and the integrity of the hypothalamo-hypophyseal axis. During this period, defects in testosterone synthesis as well as HH can be diagnosed without external stimulation of the axis. If not evaluated during minipuberty, HCG test for investigation is necessary and HH diagnosis is delayed until puberty. We recommend referral of cases of congenital cryptorchidism for endocrine evaluation at the time of mini-puberty.

FM 4

Next generation sequencing for diagnosis of monogenic diabetes in the Swiss population

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Background: Monogenic diabetes is a heterogeneous group of diabetes due to a single gene mutation and includes neonatal diabetes (NDM), maturity diabetes of the young (MODY) and rare forms of syndromic diabetes. These forms of diabetes remain undiagnosed in probably more than 90% of patients. Diagnosis is however important to adjust diabetes treatment. The aim of the study was to identify mutations causing monogenic diabetes and to validate this method for diagnostic testing.
Methods: Swiss endocrinologists were proposed to participate in the study and to send blood samples of their patients with suspected monogenic diabetes. Inclusion criteria were NDM, autoantibody negative type 1 diabetes mellitus, type 2 diabetes mellitus diagnosed before the age of forty-five years without metabolic features and syndromic diabetes regardless of treatment. The analyses were performed by a targeted next-generation sequencing (NGS) assay sequencing 323 potential diabetes genes using the Haloplex technology. All the variants were confirmed by Saenger sequencing.
Results: So far we have analyzed 147 diabetic probands by NGS, including 4 neonates, 44 children and 99 adults with a mean age at diagnosis of 1.5 month, 10.8 years and 35 years, respectively. Among the neonates we diagnosed one patient with a Wolcott Rallison syndrome *(EIF2AK3* gene), one with a mutation in the *KCNJ11* gene and one with digenic variants in the *KCNJ11* and *HNF4A* genes. Only one patient had a type 1 diabetes harboring a variant in a type 1

susceptibility gene *(IL18RAP* gene). Among the pediatric cohort 41% had a mutation in one of the known 13 putative MODY genes *(HNF4A, GCK, HNF1A, PDX1, HNF1B, NEUROD1, KLF11, CEL, PAX4, INS, BLK, ABCC8, KCNJ11)*. Among the adult population we identified MODY mutations in 35% of the probands. In the latter two groups, mutations in the GCK gene were the most frequently diagnosed (20% and 26%, respectively) and in addition 8 novel *GCK* mutations were detected. The second most frequent mutations were found in the *HNF1A* gene (11% and 3%).
Conclusion: Overall, we achieved a diagnosis rate of 39% with the described selection criteria, which is high in comparison to other studies. GCK diabetes typically doesn't need any pharmacological treatment; however before the molecular diagnosis 10% of these patients were treated with insulin which could be stopped afterwards. This report therefore validates our approach as a diagnostic tool.

FM 5

Focal convulsions in a boy with hyperinsulinism

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Introduction: Hypoglycaemia may be caused by different pathologies. Here we report the case of an infant suffering from congenital hyperinsulinism, which was initially clinically diagnosed and treated as Watanabe syndrome (a benign infantile epilepsy characterized by focal seizures in otherwise healthy children with a regular interictal EEG).
The case: A five months old boy, third child of healthy parents without history of epilepsy, born at term after an uneventful pregnancy and so far healthy, presented for the first time with a cry out of sleep, followed by opening of his eyes with cloni of the right arm and full consciousness for a duration of a few minutes. After the second episode of again focal seizure out of sleep with isolated twitching of the right arm, fulfilling all the clinical diagnostic criteria of a Watanabe syndrome, therapy with Carbamazepin was initiated. After a six weeks seizure-free interval the boy had three seizures within three days, all characterized with twitching of the right arm. Consequently, the patient was hospitalized for further diagnostic evaluation.
Results: Thorough neurological examination, EEG and cerebral MRI were unremarkable. Long term video EEG monitoring revealed continuous slowing, which pointed to a metabolic disorder involving the glucose system. Laboratory investigations after a fasting period revealed a highly pathological plasma glucose of 1.3 mmol/l and low urinary ketone bodies. Concomitantly, insulin was not suppressed during this hypoketotic hypoglycemic episode and glucagon test was positive, i.e. significant increase of glucose after intravenous glucagon injection. Primary hyperinsulinism was diagnosed. Initially, glucose was stabilized by continuous infusion of intravenous glucose solution. Therapy with diazoxide and hydrochlorthiazide was initiated and intravenous glucose supply could be reduced and eventually stopped. A molecular genetic evaluation is ongoing.
Conclusion: Commonly hypoglycemic convulsions are characterized by generalized bilateral seizures, but – as in our case – they can be associated with focal motor seizures.

FM 6

Suspect myopathy? Don't forget vitamin D deficiency!

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Introduction: Vitamin D is essential in bone and muscle health. Vitamin D deficiency is a global health problem and diagnosis should not be missed, especially with the increasing migration of children from countries where vitamin D prophylaxis is not established. Severe vitamin D deficiency typically causes rickets in children, but may also present with myalgia and muscle weakness. Vitamin D deficiency should be considered in the differential diagnosis of myopathic disease.
Case report: We report five female patients (age range, 4–16), all with a migration background. The patients were diagnosed to suffer from severe vitamin D deficiency induced myopathy. First symptoms were muscle pain, especially of lower extremities, increasing on exertion. Clinical findings included general and proximal muscle weakness and gait abnormalities resulting in impairment of daily acitivies. Two patients underwent distinct diagnostic investigations in search of myopathic disease. In three patients laboratory investigation showed elevated Creatinkinase (CK) and 25-hydroxyvitamin D levels were markedly decreased in all patients. One patient suffered from rickets-associated myopathy. Another patient showed secondary hyperparathyroidism as a consequence of the vitamin D deficiency. All patients were substituted with therapeutic doses of vitamin D. Symptoms improved rapidly after a few weeks of treatment and clinical findings were completlyreversible. 25-hydroxyvitamin D levels turned to normal values or increased markedly during treatment. All patients are still supplemented with vitamin D.
Conclusion: Vitamin D deficiency can present with significant muscle impairment, mimicing myopathic disease. In case of unexplained myopathy, don't forget vitamin D defiency. Diagnosis is made easy and treatment is simple and effective.

FM 7

Low incidence of severe hyperbilirubinaemia in Switzerland: good, but it could be better!

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Introduction: Severe hyperbilirubinaemia is the most common cause of neonatal hospital readmissions. It can lead to acute encephalopathy and potentially to persistent neurodevelopmental sequeals. Early discharge from nurseries and a lack of follow-up of the jaundiced infants seem to be the major causes for a surge in recent years. This nationwide, population based study aimed to determine incidence and etiology of severe hyperbilirubinemia in term and late preterm infants in Switzerland.
Patients and methods: The study was conducted prospectively over 5 years (2007–2011) in collaboration with all 33 pediatric hospitals in Switzerland, the Swiss Pediatric Surveillance Unit and the Swiss Federal Offices of Statistics and of Public Health. An anonymous, two step reporting system was used with a check-off form and secondarily, with a detailed questionnaire. All newborn infants with gestational age (GA) ≥35 weeks with at least one value of total serum bilirubin (TSB) exceeding the age specific exchange transfusion limit (ETL) were included. ETL was defined as TSB ≥430 µmol/L in healthy term infants, ≥370 µmol/L in sick term infants or with hemolysis, and ≥320 µmol/L in term infants with birth weight <2500 g or in premature infants.
Results: During the study period, 379'280 live births (LB) with GA ≥35 weeks were recorded in Switzerland of which 129 developed severe hyperbilirubinemia (incidence: 34.0/100'000 LB). Preterm infants (>200/100'000 preterm LB) and boys (65.9%) were overrepresented. Incidence of very high TSB peaks (>514 µmol/L; >30 mg/dL) was 1.8/100'000 LB. The etiology was identified in 63 cases (58.8%) of which 58 were related to blood group incompatibility (ABO: 63.6%; Rhesus and subgroups: 24.3%) and 3 cases to severe hematoma (4.5%). The first TSB measurement was performed within the first 12h only in 9.6%, although the risk factors (mother's blood group and hematoma) were known at birth. In 54 (43.2%) of the 125 patients with available data, the first bilirubin measurement was already above the ETL. In 16 (29.6%) of these cases, the first TSB was measured only >12h after first notice of jaundice.
Conclusions: The incidence of severe hyperbilirubinemia in Switzerland is lower than in other countries. However, many of the cases could have been avoided by a better and earlier recognition of the risk factors (mother's blood group, hematoma and prematurity). In many cases, there is a delay between recognized clinical jaundice and first TSB measurement.

FM 8

Neonatal resuscitation practical training in 8 rural health centres in Mali

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Introduction: Every year 2.7 million newborn infants are recorded dead around the world, of which 98% die in low income countries. Perinatal asphyxia remains one of the leading causes of neonatal and under 5 mortality. Practical training in neonatal resuscitation (NR) is a key challenge in very remote health facilities, though with a high potential impact to reduce neonatal mortality by 47% (Pediatrics [2013];131:e353–e360). Terre des hommes Foundation has recently implemented, in partnership with the Clinic of Neonatology of the University Hospital of Lausanne (CHUV), a NR training program in its rural intervention area in Mali.
Method: In the health districts of Macina and Markala of the Ségou region, 8 health care structures (2 regional hospitals and 6 community clinics) were selected to be the pilot facilities (PF). During 6 months, the project was carried out in 3 phases. *Phase 1* (nov. 2014): training of PF health workers and 2 midwife instructors to the *Helping Babies Breathe* curriculum. Each PF has been equipped with one NR low-cost simulation kit (ventilaton bag and masks, mannequin, aspirator, stethoscope, teaching material) for autonomous training. *Phase 2* (5 months): regular simulation training sessions in each PF by midwife instructors. *Phase 3* (mai 2015): evaluation in each PF of simulation and resuscitation material conditions as well as NR theoretical knowledge and practical skills.
Results: Fifty health workers have been trained and drilled to manage NR. Six months after the initial training, 52% of trained health workers have been evaluated in 7/8 PFs (87.5%). 86% of the NR equipment was present and functional. The median level of theoretical NR knowledge gained (12 items) was 75% (ranging from 46% to 88%, depending on the PF). The median level of NR practical skills gained (14 items) was 82% (ranging from 50% to 93%).
Conclusion: Neonatal resuscitation skills are an area of competence that can be introduced and taught efficiently to the most remote health care workers, at low cost and without sophisticated technology. Such a model of decentralized low-cost simulation training could be expanded to other essential newborn care programs, like kangaroo mother care and neonatal sepsis management.

FM 9

Parent empowerment program for health professionnals to prevent Abusive head trauma

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Background: Becoming parents is a challenge and caring for an infant with persistent crying is one of the most stressful events for parents. A newborns' risk of sustaining an abusive head injury resulting from shaking during the first year of life is described by scientific literature. In France and Switzerland, health professionals often feel helpless on how to interact with family and to prevent Abusive head trauma. Low-cost prevention programs delivered by health professionals can substantially reduce stress, burn out and shaking baby. Health care family centered programs during the postnatal period should be developed in order to answer family needs, enhancing parenting skills and resources.
Objective: To plan, implement and evaluate a family-centered care neglect prevention program with direct health-care providers. Methods: Mixed methods were used in order to evaluate the collaborative program health-care providers. This study used a pre / post-intervention design with measurement of the helpgiving practices. This design allowed the description and analysis of predictive factors of change in knowledge and practice. Data were collected using questionnaires designed to measure empowerment helpgiving practices and using interviews.
Results: The program was recognized as useful and innovative by the majority of participants. It improves communication and collaboration between parents and health providers using systemic tools. The analysis of results on a sample of professionals showed a significant increase in knowledge about family centered care used to prevent risk of negligence.
Conclusions: This family centered care program was perceived as helpful in improving the family empowerment and the interactions between parents and babies. To assist in the transfer of empowerment skills in practical intervention with parents, new tools such as cards and video were developped. Health care professionnals responded positively to the new approach and used the tools developped to reach best pratices' targets.

FM 10

Clustering of cardiovascular disease risk factors among male youths in Southern Switzerland: preliminary study

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Background: The distribution of cardiovascular risk factors among youths in Southern Switzerland is poorly understood. We aimed at describing the prevalence of cardiovascular disease risk factors in 18- to 20-year-old males undergoing medical examination to assess fitness for recruitment into the army.
Methods: Between 2009 and 2013, 1541 (21%) out of 7310 conscripts volunteered for answering a structured questionnaire addressing smoking behavior, sedentarity and familial cardiovascular risk factors, as well as measuring blood pressure, lipidemia and waist circumference.
Results: 1. Height, weight, body fatness and blood pressure were not statistically different between conscripts who had respectively had not volunteered the study. 2. Following cardiovascular disease risk factors were detected: smoking (N = 656; 43% of the study participants), sedentarity (N = 594; 39%), positive cardiovascular family history (N = 235; 15%), blood pressure ≥140/90 mm Hg (N = 88; 5.7%), total cholesterol ≥5.2 mmol/L (N = 83; 5.4%), waist circumference ≥1.02 m (N = 55; 3.6%). No cardiovascular risk factor was detected in 434 (28%), one factor in 612 (40%) and two or more in 495 (32%) participants.
Limitations: Recognized cardiovascular risk factors such as diabetes, hyperuricemia and hypovitaminosis D were not addressed. Noninvasive surrogate biomarkers for atherosclerotic disease such as carotid intima-medial thickness or arterial stiffness were not measured.
Conclusions: This preliminary cross-sectional survey generates the first analysis of cardiovascular disease risk factors among ostensibly healthy male youths living in Southern Switzerland. The main finding is that two or more cardiovascular disease risk factors are present in approximately one third of them. Since smoking and sedentarity, the most commonly detected cardiovascular risk factors, are preventable, youths represent an opportunity for the promotion of lifestyles that will affect the development and progression of atherosclerotic disease.

FM 11

Cow's milk proteins in phytosanitary products: a new public health threat? "A propos" of a case of brittle asthma death

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Introduction: Phytosanitary products containing cow's milk proteins (CMP) are supposed to be more environment friendly and are becoming widely used. A case of death due to a severe asthma attack in a patient allergic to CMP rises concerns about their impact on public health.
Case report: Following non strenuous outdoor exercise, a 7 years old girl, known for asthma, showed signs of severe dyspnoe. In spite of salbutamol administration she suffered a cardio-respiratory arrest 30 min thereafter. Despite basic life support followed by intensive advanced life support with intubation, ventilation and epinephrin administration, she remained asystolic and resuscitation was discontinued 75 min after the first symptom. The post mortem examination showed gross pulmonary overdistension, bronchial inflammation and mucus plugs, but no laryngeal or glottic swelling. Tryptase level was 67.6 mcg/l. The patient was highly allergic to CMP and had already experienced two episodes of anaphylactic reactions requiering intramuscular epinephrin, once after contact with only traces of CMP. She was highly aware of dietary implications of her allergy and had verbalised these implications just one hour before the attack started. Spirometry, ergospirometry and FeNO measurment had very recently showed her asthma was well controlled with budesonide/formoterol inhalation BID, and salbutamol before exercise. The history (time lapse between exercise or meal and first symptoms), the symptoms (silent chest, no angioedema, no urticarial rash) and the post-mortem examination allows us to think that death was due to brittle asthma and not to an anaphylactic shock. In search for a trigger, we discovered that a vineyard near to the sport field had just been treated with phytosanitary products containing CMP.

Discussion: We postulate that the deadly brittle asthma attack in our patient can have been triggered by inhalation of CMP contained in phytosanitary products. As this kind of products is more and more widely used, and as cow's milk protein allergy is not uncommon in the population, even in adults, we imagine this use could represent a new public health threat.
Conclusion: We suggest a cautious evaluation of the risks of CMP use in phytosanitary products for patients allergic to CMP. A high index of suspicion should be raised in presence of severe asthma attack in patients allergic to CMP in areas where and seasons when these products are used, especially when other triggers cannot be found.

FM 12

Unplanned return visits to a paediatric emergency department

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Objectives: Unplanned return visits (URVs) to emergency departments (EDs) account internationally for 2.5 to 5.2%. A first ED visit usually ends with the recommendation to have a follow up visit at the pediatrician's office. However, there are patients who return unplanned to the ED. This study aimed to assess factors for unplanned return visits at the ED of the University Children's Hospital in Zurich.
Methods: All patients with an URV to the ED between January until December 2013 were included in the study. Data were taken retrospectively from the electronic patient files und different variables were defined and analyzed.
Results: 1682 (4.6%) out of a total of 36'618 treated patients had an URV and were included in this study. Out of all URVs, 1206 (71.7%) were infants and toddlers. 437 patients (26%) returned within 24 hours and more than 75% of patients within 72 hours after their first ED visit. The URVs were independent of the weekdays and mostly occurred between 10 o'clock and 22 o'clock. In 85% of the cases, the URVs were judged as unnecessary, and in 15% a hospitalization was indicated, mainly for children with a worsening respiratory illness.
Conclusions: The prevalence of URVs in the ED of our institution was within the prevalence reported in the literature. While URVs lead to hospitalization in some patients, the majority of URVs were unnecessary. These results indicate that a correct evaluation of the child's health state by parents is often challenging and requires repeated medical attendance following a first ED visit. Intensive counselling and scheduled short-term follow up consultation at the pediatrician's office could prevent URVs to the ED.

FM 13

Cyst of the pineal gland in retinoblastoma: what link with disease and treatments

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Objectives: Retinoblastoma (Rb) represents the most frequent intraocular pediatric tumor, with an average incidence of 1:14'000 to 1:34'000 births. In 2006 we were first to report on pineal cysts that appeared to be more common in children with hereditary bilateral Rb. The aim of our study was to review in a larger patient cohort the occurrence of pineal cysts and to study the link with disease caracteristics and treatments received.
Methods and Patients: Observational retrospective study of clinical and radiological data of 103 patients treated for Rb who had undergone a cerebral magnetic resonance imaging (MRI) between 2006 and 2013 and had a follow-up. Clinical records were reviewed for sex, age at diagnosis, hereditary pattern of disease, tumor laterality, stage according to the International Classification of Retinoblastoma, age at first MRI, treatments received, date of first and last treatment and last follow-up, response to treatment, long-term outcome, time interval from diagnosis of Rb to the diagnosis of a pineal cyst, and genetic data if known. Radiological reports and brain images were reviewed for each patient with pineal cyst to record its size and change over time.
Results: Of 103 patients with Rb 56 had unilateral and 47 bilateral disease. Ninety- five were sporadic Rb while 8 were familial. Forty-nine MRIs out of 103 (47.6%) presented a pineal cyst and were reviewed by a neuroradiologist to verify aspect and the size of the pineal gland. Occurrence of cysts was more frequent in bilateral disease, sporadic disease, in presence of a documented genetic mutation and in group D or E, but without statistically significance. No impact of treatment on the occurrence of cysts could be demonstrated. At 1 year of follow-up, cysts had higher growth in bilateral retinoblastoma and those with documented genetic mutation, but without statistical significance. None of the other parameters showed significant impact on growth.
Conclusions: We found a high incidence of pineal cysts in Rb patients, but could not demonstrate a significant relationship to the hereditary subgroup or genetic mutation. The evolution was benign without malignant transformation in absence of atypical radiological signs. The higher incidence however compared to a healthy pediatric population clearly indicates that pineal cysts are part of the disease-related midline brain abnormalities.

FM 14

Changes in management of febrile neutropenia and impact on lenght of hospital stay in a pediatric oncology unit

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Background: Chemotherapy-induced febrile neutropenia (FN) is a frequent complication in cancer treatments. Various empiric antibiotic (AB) regimens are recommended worldwide to treat quickly and effectively children with fever. Since 2000s, patients were stratified into low and high risk group for severe infection according to their underlying immunosuppression and were respectively treated with a 3rd or 4th generation cephalosporin or piperacillin/tazobactam with an aminoglycosid or a broad spectrum monotherapy of beta-lactamin. This study addresses the impact of empiric antibiotic strategies (bitherapy with ceftriaxone/amikacine for all children with FN vs meropenem monotherapy for high risk patients and bitherapy with ceftriaxone/amikacine in low-risk children) on the length of hospital stay (LOS).
Procedure: 10 years retrospective analysis of the first episode of FN in children followed at the University Hospital of Lausanne. We selected important demographic, clinical and treatment-related predictors potentially influencing length of hospital stay.
Results: One hundred and fifty-six episodes of FN were reported, which identified various predictors potentially associated with prolonged LOS. Moreover, antibiotic stratification resulted on average in a shorter LOS by 25% (irr = 0.754, P = 0.002).
Conclusions: Our study supports antibiotic stratification according to the underlying risk of developing severe infections among children with FN.

FM 15

Delayed decrease in SpO_2 values in infants with bronchiolitis: ED observation time for safe discharge

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Guidelines for bronchiolitis management are mainly established for hospitalization and discharge criteria after hospitalization. However the emergency department (ED) observation time necessary to allow safe discharge in patients with SpO_2 above 90–92% is still to be defined. Our aims were to identify the risk factors associated with a delayed decrease in SpO_2 in patients with SpO_2 ≥92% at ED arrival and to identify the ED observation time necessary to allow safe discharge. We retrospectively studied children <1 year admitted to the ED with bronchiolitis. Among 581 episodes of bronchiolitis, 47/581 (8%) patients had SpO_2 <92% at ED arrival, however 106 (18%) had a delayed decrease in SpO_2 (<92%) after a few hours of ED observation. Gender female, age <3 months, readmission and a more severe initial clinical presentation were associated with an increased risk for a delayed decrease in SpO_2 values with odds ratios varying from 1.7 to 7.5. In patients <3 months the decrease in SpO_2 values was slower [6.0 hours (IQR 3.0–14.0) vs. 3.0 hours (IQR 2.0–6.0), P = 0.018]. In 90% this decrease occurred in 20 hours for patients <3 months and 10 hours for patients ≥3 months. Patients <3 months with higher age-specific respiratory rates were more likely to have a faster decrease in SpO_2 values when compared to patients <3 months with

normal respiratory rates [4.4 hours (IQR) 3.0–11.7) vs. 14.6 hours (IQR 7.6–22.2), P = 0.037]. In conclusion, patients at risk for a delayed decrease in SpO_2 values could be safely discharged after an ED observation time of 10h for patients ≥3 months and 20h for patients <3 months if no decrease in SpO_2 values happens within this period and child is feeding well.

FM 16

Excellent results after reduced intensity conditioning and T-cell replete allogeneic blood stem cell transplantation in haemophagocytic lymphohistiosis

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Introduction: Reduced intensity conditioning (RIC) regimens containing submyeloablative iv.busulfan (cum. AUC 45–65 mg/Lxh), high-dose fludarabine 180 mg/sqm (d–8 to –3; in infants <9 kg 6× 1.2 mg/kg), serotherapy (rabbit Anti-T-cell globulin or low-dose alemtuzumab) and Graft-versus-Host-Disease (GVHD) prophylaxis comprising Cyclosporine A (until d-3 to d+160) and Mofetil-Mycophenolate (until d0 to +80–100) have shown excellent results in high-risk chronic granulomatous disease patients. Its efficacy in other non-malignant diseases, e.g. Haemophagocytic Lymphohistiocytosis (HLH), is still unknown.
Material (or patients) and methods: In this present study (6/2009 to 6/2015), we analyzed the outcome after administration of this protocol in children and adolescents/young adults with haemophagocytic lymphohistiocytosis (age: med 0.83; range 0.27–21 yrs): n = 3 with MUNC 18-2, n = 5 with MUNC-13-4, n = 2 with XLP (Purtillo's Disease), n = 1 Chédiak-Higashi-Syndrom and n = 2 with HLH with unknown genetic cause but abnormal degranulation assays.
Results: Transplants comprised n = 9 unrelated (n = 7 HLA-10/10 and n = 2 HLA-9/10) and n = 4 related HLA-identical donors. The donor sources were n = 7 bone marrow (BM,) n = 4 peripheral blood stem cells (PBSC) and n = 2 cord blood (CB). Intravenous total busulfan doses ranged between 4.4 and 17.2 mg/kg (median 12 mg/kg) corresponding to cumulative AUC of 48–77 mg/Lxh (med. 63.5). Fludarabine was administered at a total dose 180 mg/sqm (n =10) or 6 × 1.2 mg/kg (n = 3) depending on body weight. Serotherapy comprised Alemtuzumab (n = 9; range 0.8–2.5, median 0.6 mg/kg) and Anti-T-cell globulin (n = 1, ATG-Fresenius, 40 mg/kg; n = 3, Thymoglobuline 7.5 mg/kg). All patients engrafted. No graft failures, 2 cases of reversible hepatic venoocclusive disease and one case of reversible pulmonary hypertension were observed. After a median follow-up of 34 months (range 4–65 mo), the overall and event free survival rates are 100/100%, respectively. The rates of acute GVHD >III–IV and of chronic GHVD were 0/0%, respectively, at last follow-up. All surviving patients exhibit a stable myeloid donor chimerism (n = 3 80–90%; n = 9>95%).
Conclusion: Submyeloablative targeted busulfan, fludarabine and serotherapy containing RIC-regimens are excellent treatment options in HLH-patients with T-cells replete transplants including unrelated cords.

FM 17

Fever in neutropenia in children and adolescents: Evolution of main characteristics over two decades, 1993–2012, in a single center

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Objectives: This study aimed to describe the long-time evolution of fever in neutropenia (FN) characteristics at presentation, its management, and its outcome, in children and adolescents treated with chemotherapy for cancer.
Methods: This retrospective cohort study in children and adolescents <17 years presenting with FN covered two decades (1993 to 2012), in a single institution. FN was defined as fever in severe neutropenia (absolute neutrophil count <0.5 G/L) induced by chemotherapy for cancer. Mixed logistic regression, accounting for multiple FN episodes per patient, was used for analysis, results are given per decade.
Results: In total, 703 FN episodes were reported in 291 (50%) of 583 patients with chemotherapy (maximum per patient, 9). Characteristics at presentation: the proportion of FN in patients with acute lymphoblastic leukemia (ALL) versus other diagnoses increased over time (overall proportion 46%, odds ratio [OR] per decade, 1.66, 95% CI, 1.25 to 2.19). Central venous catheters (CVC; 66%; OR 21.1; 10.9 to 40.9), clinical signs of bacterial (23%; OR 2.39; 1.65 to 3.46) and of viral infections (37%; OR 1.54; 1.11 to 2.13), and fever itself (overall median, 39.1 °C; 0.10 °C per decade; 0.04 to 0.17) all significantly increased over time.
Management: the empirical use of ceftriaxone/amikacin decreased over time (86%; OR 0.56; 0.35 to 0.91), the duration of intravenous antibiotics remained stable (median 6 days; –0.4 per decade; –1.5 to 0.7), but it was more frequently escalated (23%; OR 8.47; 4.93 to 14.6). Hospitalization duration remained stable (median 5 days; 0.2; –0.9 to 1.2).
Outcomes: microbiologically defined infections increased over time (34%; OR 1.71; 1.25 to 2.33), because viral (10%; OR 6.02; 3.26 to 11.1) and fungal (7%; OR 6.77; 1.46 to 31.4) infections were more frequently diagnosed, while bacteremia (21%; OR, 0.93; 0.64 to 1.35), remained stable.
Conclusion: Significant and clinically relevant changes were detected in characteristics at FN presentation, management, and outcome over two decades. Clinically, they reflect changes over time towards routine use of CVC, towards higher chemotherapy intensity in ALL; towards a more liberal definition of fever, and thus FN itself; and towards increased diagnostics for viral and fungal infections. Scientifically, these changes need to be accounted for in longitudinal research projects.

FM 18

N_2-MBW in comparison to spirometry in children after lung transplantation

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Chronic lung allograft dysfunction is the main cause of deaths after the first year post transplantation (LTX) and bronchiolitis obliterans syndrome (BOS) is the leading cause of lung allograft dysfunction. Up to now BOS is a clinical diagnosis defined as an irreversible decrease of FEV_1 <80% (compared to best FEV_1 post LTX). New lung function tests like the nitrogen-multiple breath washout test (N_2 -MBW) detect small airway disease early in patients with chronic lung disease and seem thus promising for diagnosis of BOS. The aim of this study was to compare N_2-MBW and conventional spirometry outcomes during a short and long term follow-up of children who underwent lung transplantation at a single center. 38 children, age 7–18 years, performed spirometry and N_2-MBW in a clinical stable phase at least three month after LTX. N_2-MBW gives results on global (lung clearance index; LCI), conductive (Scond) and acinar (Sacin) VI. Patients performed lung function tests after LTX. Depending on the timepoint we categorized follow-up into short term (<18 months; n = 21) and long-term (>18 months; n = 22). Five patients were included in both follow-up groups. Z-scores were calculated using data from a healthy aged-matched control group. Mean (SD) LCI was 5.8 (6.1) z-scores, while FEV_1/FVC was –1.0 (1.4) z-scores at short- term follow-up. At long-term follow-up LCI was 3.8 (3.3) and FEV_1/FVC –0.9 (1.4) z- scores. The number of patients with elevated LCI values was 16/21 at short-term and 18/22 at long-term follow-up in comparison to 8/21 and 6/22, respectively for FEV_1/FVC. On both time points poor agreement between both parameters (kappa <0.4) was found. In this cross-sectional study LCI seems more sensitive to detect lung disease compared to FEV_1/FVC. Longitudinal follow-up will show if this is an inherent phenomena after LTX or indicative of early BOS.

FM 19

Correct environmental settings and simple changes in software algorithms can improve the quality of infant multiple breath washout outcomes

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Introduction: Multiple breath washout (MBW) is an established lung function test in infancy and serves as a primary outcome in clinical studies. However, the robustness of the analysis software has never been evaluated so far. In this study, using MBW measurements of healthy infants we tried to identify software inaccuracies that potentially influence the results. We also attempted to improve the quality of the results by modifying the algorithms used for the analysis.
Methods: MBW tests were performed in healthy infants from two centers in Switzerland (N = 12 per center) by ultrasonic flowmeter using sulfur hexafluoride (SF_6) as the tracer gas. Best quality tracings (n = 36 per center) were analysed with the current analysis software as well as with alternative analysis settings, where we reset correct environmental parameters and applied simple changes in the software algorithms.
Results: Using the current analysis software, the coefficient of variation (CV) for functional residual capacity (FRC) differed significantly between centres (mean ± SD (%): 9.8 ± 5.6 and 5.8 ± 2.9, respectively, p = 0.039). In addition, FRC values calculated during the washout differed between –25 and +30% from those of the washin of the same tracing. Temperature recordings had a great influence on the results. Algorithm changes normalized the CV of FRC between the two centers (mean ± SD (%): 4.6 ± 2.6 and 5.2 ± 3.1, respectively, p = 0.6) and lowered the FRC differences between washin and washout (between –15 and +9%).
Discussion: We identified high intra-subject and inter-center differences in FRC values that could not be physiologically explained when using the current software algorithms. We show that the use of correct environmental settings and alternative algorithms can improve the robustness of infant MBW outcomes. Thus, we recommend their use for more realistic between-centre comparisons of these outcomes.

FM 20

Clinical Research Management: PedNet Bern as a third space worker

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PedNet Bern is a service provider on the field of clinical paediatric research. Service provided includes theoretical and practical support. On the theoretic side PedNet is a signpost for newcomer on the field, helps to develop protocols, advises on financial and regulatory aspects. Practical support can be study coordination, study nurse or study investigator assistance, conduction of clinical studies (either with a specialist or the PedNet doctor as principal investigator, PI). Aim of the platform must be to provide optimal support for optimal outcome. During our work we detected several pitfalls for the clinical researcher in conducting clinical paediatric trials: 1. The GCP compliant implementation of a study cannot be ensured only through participation in a GCP course. The legal requirements especially for paediatric studies are high and hardly feasible additionally to daily clinical practice. 2. Expenses incurred by planning and setting up a clinical paediatric study are extremely underestimated and cannot be compensated by the proceeds of study conduction. 3. Financial flows and calculations are not objectively comprehensible independent from the involved party (pharma industries, hospital, PI). 4. The effective study conduction after a good planning and set up of a study is NOT a problem itself. As PedNet Bern we address the obstacles above with our experiences in clinical studies. First of all we help PIs to completely adhere to the procedures as described in the study protocol. Furthermore, a GCP compliant study performance is strongly connected to an optimal document handling. Investigators are lacking time resources to take care of the documents in a GCP compliant way besides their daily clinical workload. PedNet provides regulatory knowledge and time resources in this field. Other regulatory necessary solutions like monitored and locked IMP fridges were established and made accessible to in house PIs. Point 2 and 3 is addressed by a constant discussion with all involved parties to improve financial understanding within a paediatric clinical project. This includes but is not limited to the search for funding, advice in contract negotiations with the sponsors and advocacy in terms of paediatric clinical studies in our hospital.

FM 21

A pilot study for early intervention in late talking children in private paediatric outpatient setting

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Background: Problems of language development are frequent. About 65% of late talkers at age 2 years have a language delay at 3 years. Early intervention might improve outcome. We present a pilot study to evaluate feasibility of a paediatric private clinic based study to compare 3 different short term interventions for late talkers.
Methods: In 3 different private paediatric outpatient clinics children were screened at age 24 months with a standardised, for Switzerland adapted screening test (SBE-2-KT, Sprachbeurteilung durch Eltern mit 2 Jahren). Late talkers were offered randomly: 1) Heidelberg Parent-based Language Intervention (HPLI), a highly structured and interactive instruction programm (n = 5) ii) a shorter parent instruction based on the DVD "Sprache kitzeln", Steiner, Braun, HPF ZH, (n = 3) iii) isolated distribution of the DVD for instruction without direct training of parents (n = 8). By a semi-structured interview the three interventions were evaluated in the applicability and user friendliness. Children included into the study were tested at the start by the SETK-2 (Sprachentwicklungstest für Kinder 2 Jahre) and at age 3 by the SETK-3-5 (für drei bis fünfjährige Kinder) and the AWST-R (Wortschatztest).
Results: The SBE-2-KT showed to be a valuable screening instrument with 15.7% late talkers of all children screened. All screened late talkers, in whom parents agreed for an intervention did show corresponding problems in the SETK-2. However, only 20% of parents with late talkers agreed to participate in the study. An interview showed both direct parent instruction courses to be accepted and valued by parents and speech therapists. More extensive speech testing at 2 years and especially at three years were difficult due to missing Swiss standards and non compliance in 79% of the 3 year old children. Small and unequal group numbers made statistical analyses impossible. Comparison of percentile ranking at 2 and 3 years did show for all three groups an improvement in speech production, but also a percentile decrease in speech comprehension.
Discussion: The pilot revealed, that parents are not yet ready for time consuming interventions for a 2 year old late talker and that cooperation for testing at age 3 years is of major concern. Future steps will be to evaluate by a survey on concerns by late talking and readiness for interventions in parents and professionals. Also the problems of adequate tests at 3 years needs some further investigations.

FM 22

Use of simulation-based medical training in Swiss pediatric health care institutions: A national survey

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Background: Simulation-based medical training (SBMT) is a powerful tool for continuing medical education. Despite of medical SBMT rising popularity over the past decade, best practices guidelines pertaining to design and implementation have yet to be established. The aim of this national survey is to assess, describe and analyze the current state of SBMT in Swiss pediatric health care institutions.
Methods: Between June and August 2015 a national survey designed by the authors was carried out with respective medical education representatives of every FMH recognized Swiss pediatric health care institution. The survey reference day was May 31st 2015.
Results: 35/36 Swiss pediatric health care institutions/units answered our survey (response rate 97.2%). Sixty-nine percent were offering simulation-based training. More than 90% were offering their training in-situ and 62.5% were using high-fidelity mannequins. Technical skills, communication and leadership ranked among the top training priorities. All institutions were providing their training to inter-professional participants. The vast majority conducted training that was neither embedded within a larger educational curriculum (79.2%) nor evaluated (66.6%) by its participants. Only 5 institutions (20.8%) extended their training to at least two thirds of their hospital staff (physicians, nursing staff).
Conclusions: More than two third of the FMH recognized Swiss pediatric health care institutions are offering SBMT to their staff. The absence of a programmatic approach along with extending to a small number of healthcare employees are were identified as shortcomings that need to be addressed to further improve continuing medical education.

FM 23

Analysis of Medication Prescribing Errors in critically ill Children

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Objectives: Medication prescribing errors (MPE) can result in serious consequences for patients. In order to reduce errors, we need to know more about the frequency, the type and the severity of such errors.
Methodes: We performed therefore a prospective observational study to determine the number and type of medication prescribing errors in critically ill children in a paediatric intensive care unit (PICU). Prescribing errors were prospectively identified by a clinical pharmacist.
Results: A total of 1'129 medication orders were analysed. There were 151 prescribing errors, giving an overall error rate of 14% (95% CI 11 to 16). The medication groups with the highest proportion of MPEs were antihypertensives, antimycotics and drugs for nasal preparation with error rates of each 50%, followed by antiasthmatic drugs (25%), antibiotics (15%) and analgesics (14%). 104 errors (70%) were classified as MPEs which required interventions and/or resulted in patient harm equivalent to 9% of all medication orders (95% CI 6.5 to 14.4). 45 MPEs (30%) did not result in patient harm.
Conclusion: With a view to reduce MPEs and to improve patient-safety, our data may help to prevent errors before they occur.

FM 24

Evaluation of CIRS-reports concerning medication errors in a paediatric hospital

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Introduction: Medication errors contribute to the emergence of preventable adverse drug events and therefore should be minimized. The paediatric population is especially vulnerable for medication errors because of several risk factors like weight- or age- based dosing.
Objectives: The objective of this project was the analysis of medication errors reported by the critical incident reporting system (CIRS).
Method: Retrospective evaluation of all CIRS-reports filed during a 24-month period in the paediatric department of a Swiss hospital providing tertiary care. All reports included concerned drugs. A single report can refer to more than one error. The errors were categorised by the step of the medication process concerned and by the cause of the error, such as insufficient knowledge, inadvertency or neglect of internal policies and guidelines. This categorisation was done in function of explications and reporter- mentioned causes within the reports.
Results: During the evaluation period 287 reports were filed, including 297 errors of which 99 (33%) were identified as medication errors. 27% of these concerned the prescription, 41% the preparation, 31% the administration and 1% the documentation of drugs. 77% of all medication errors were caused by inadvertency, 18% by insufficient knowledge and 5% of medication errors were due to neglect of internal policies. Almost half of all reported errors during prescribing or preparation concerned the dosing. Administration errors concerned mostly the timing. Most reports (89%) were submitted by nurses.
Conclusion: The high amount of errors caused by inadvertency stresses the importance of identifying risk factors and building barriers in the medication process to prevent this type of errors. Almost every 5th error was caused by insufficient knowledge, mostly due to lacking access to drug information supporting the importance of easily accessible drug information. Nearly ¾ of errors reported were caused by nurses. This could be biased by the low reporting frequency of physicians. Nevertheless it stresses the importance of training and identifying measures to reduce errors during preparation and administration.

FM 26

Chronic pancreatitis in childhood – would you think about genetics?

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Background: Paediatric pancreatitis has received increasing attention over the last decade. Pancreatitis is defined as the histological occurrence of inflammation in the pancreatic parenchyma. In children, pancreatitis may be caused by congenital variants of the biliary and pancreatic ducts, drug toxicity, trauma, idiopathic and systemic disease. In addition, rare genetic predisposition may cause pancreatitis, mutations of PRSS1, SPINK1, CTRC and CFTR being the most common.
Case Report: A 15 years old female adolescent presented to our emergency room with acute abdominal pain, emesis, and coprostasis. Her prior history included recurrent episodes of acute abdominal pain for 2–3 months. On admission, clinical assessment was normal except for a resistance in the descending colon corresponding to coprostasis. The laboratory work-up revealed significantly elevated pancreatic enzymes: amylase 509 U/L (normal range <110 U/L), lipase 860 U/L (normal range 13–60 U/L). Laboratory testing for the most common etiologies of pancreatitis was unremarkable. Ultrasonography of the abdomen revealed no pathologies apart from distinct coprostasis. Emergency computed tomography scanning of the abdomen showed a cystoid enlargement of the pancreatic duct. Magnetic resonance cholangiopancreaticography demonstrated reduced pancreatic parenchyma, and three filling defects consistent with non-calcified concrements were detected. Symptomatic therapy led to a marked clinical improvement, amylase and lipase levels normalized. Further investigation included endoscopic retrograde cholangiopancreatography, which identified stones in the pancreatic duct, and chronic pancreatitis Cambridge IV was diagnosed. A hereditary form of pancreatitis was considered as etiology even though the family history was unremarkable. The genetic testing revealed homozygosity for the chronic pancreatitis predisposing SPINK1 mutation p.Asn34Ser.
Conclusion: Abdominal pain in children and adolescents warrants careful evaluation. Laboratory findings consistent with pancreatitis require etiological investigation with adequate radiological work-up. In the absence of common etiologies, genetic testing should be considered. In our case, the protective mechanism of SPINK1 against early activation of trypsinogen in the pancreas was lowered by homozygosity for the most frequent disease causing SPINK1 mutation (p.Asn34Ser). This explains the patient's development of recurrent acute and subsequent chronic pancreatitis at a young age.

FM 27

Apps for paediatric dosing – an evaluation

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Background: The website www.kinderdosierungen.ch provides health professionals with paediatric dosages. To increase usability, we aim to develop a mobile app. Many apps containing paediatric dosages are already available on the market.
Purpose: Since we are interested to see whether the available apps are safe to use in daily practice and to identify areas for possible improvement, we evaluated their quality and content.
Material and Methods: The Internet, apple app store and google play were screened for apps focusing on paediatric dosages. The apps found were analysed according to criteria including age, costs or number of active ingredients. For a more in-depth evaluation, apps with a dosage calculator and either more than 70 active ingredients, or a calculator specific for preterm infants were selected and assessed in the main categories quality/content, quantity, calculator, features, usability, and additional professional information.

Results: Of the 43 apps evaluated, more than a third (N = 15) are available for free. Nearly half of the apps (N = 19) contain between 20 and 100 active ingredients, while approximately a quarter contain more than 100 active ingredients. Eighteen apps (40%) fulfilled our criteria for further evaluation. With a maximal possible score of 30, the highest score reached was 20 (Safe Dose, Epocrates and Lexicomp), followed by 18 (AGN Emergency Booklet) and 17 (Peds Meds). The app Safe Dose ranked first in the category features and second in quality/content and additional professional information. Epocrates ranked third in all categories with the exception of the calculator feature, which received a low rank. Lexicomp was top in the categories quality/content, quantity and additional professional information however scored poorly with regards to usability and calculator function. Importantly, regarding the lowest ranked apps, none were identified that would be dangerous to use.
Conclusion: There is room for improvement for paediatric dosing apps, especially regarding integration of preterm infant calculations into apps that are not specifically designed for neonatology. Prior to using an app, a short evaluation is recommended since the appropriate app depends on the contents and features that are important for the user.

FM 28

Evaluation and comparison of the predictive value of the "Braden Q" scale and of the "Neonatal Skin Risk Assessment Scale (NSRAS) for the assessment of pressure ulcer risk in neonates

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Background: Pressure ulcers are an iatrogenic complication of intensive care. Their prevalence is a quality indicator. Hospitalized neonates are particularly at risk of developing pressure ulcer. In this vulnerable population, ulcers are believed to increase the morbi-mortality. Validated tools to predict, report and compare the risk of pressure ulcers in neonates are then needed. So far, such a tool does not exist in French.
Objectives: Our study aimed at translating, testing and comparing the predictive positive value of two scales evaluating pressure ulcer risk in neonates.
Methods: Translation of the Braden Q and NSRAS (Neonatal Skin Risk Assessment Scale) scales in French according to Wild's method, followed by an observational prospective study. The study included 80 neonates that were hospitalized in the Neonatology Intensive Care Unit of a tertiary referral hospital in Switzerland, between October 2015 and January 2016. It has been approved by the local ethics committee. Following parental consent, the included patients were observed from the first 48 hours of life, and followed up to 10 days. Two clinicians independently assessed daily the pressure ulcer risk, each using either the Braden Q or the NSRAS. Simultaneously, the nurse in charge of the patient assessed the presence of pressure ulcer with the Skin Assessment Tool (SAT; Gold standard). Clinicians and nurse were blinded with respect to each other's assessments. Pressure ulcers were classified according to NPUAP graduation and pictures were done. The patients dropped out when a pressure ulcer appeared or when the patient leaved the department or after 10 days of life, whatever came first. Sensitivity, specificity and positive predictive value for pressure ulcer appearance within the 24 hours following each observation are being calculated and compared between the two scales using Mc Nemar's tests. Their concordance will be estimated using Bland and Altman's method after standardising the scores of both scales.
Results: Among the 80 patients included, 10 of whom (12.5%) developed a pressure ulcer. Further statistical analyses are underway.
Conclusion: The development and the use of appropriate pressure ulcer risk scales should contribute to the improvement of strategies and recommendations for the prevention and management of pressure ulcers in neonates. It thus plays an important part in the improvement of the quality of care and patients safety.

FM 29

Specific positioning of preterm infants and new-born babies for optimal physiological development

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Background: Preterm infants and sick new-born babies need comprehensive and adequate medical care. Within the framework of various implemented measures optimal positioning is an efficient and cost effective intervention to support children in their development. Nurses and physiotherapists are equally involved in this domain but they have different functions, perspectives and education. Knowledge and experience of both professional groups should be enhanced by targeted interdisciplinary exchange and thus directly benefiting the children.
Methods: Nurses und physiotherapists of the University Children's Hospital Bern who work as a kinaesthetic trainer, coach for basal stimulation, clinical nurse specialist, physiotherapist and physiotherapy expert have joined to define objectives and principles of positioning premature and new-born babies based on current medical knowledge, literature search, established concepts and already existing guidelines. Extensive professional experience and considerations on the feasibility in the clinical setting were also included and discussed. A further aspect of this matter was the practicability after discharge from hospital. Parents should also have the opportunity to continue individual and safe positioning of their baby at home.
Results: The present guidelines form the theoretical basis for the implementation of optimal positioning in clinical practice. The multidisciplinary approach helps to integrate knowledge and skills of nurses, physiotherapists and parents leading to reciprocal impulses. A common approach considerably facilitates uniform practices particularly with regard to individual adaptations for children with special needs. Joint principles and instructions are also helpful to integrate new members of staff.
Conclusions: Active interprofessional exchange concerning optimal positioning of premature and new-born infants is beneficial for all participants, especially for the small patients. The aim of these basic principles and goals is not to limit the users thinking and acting but to encourage careful individual observation of each child. The understanding of the infant's condition and needs is essential to offer him individual body positions that support the best possible development.

FM 30

Reasons for spontaneous visits to the school nurse in a population of young migrants in Switzerland: Stressing the importance of low-threshold access to care

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Objectives: Young migrants are a vulnerable population. Cultural and migration factors increase the risk of ill-health and complicate the access to health services. Schools can facilitate the access to adequate care. Our study was conducted in one public school in Geneva (Switzerland), providing education to recently arrived young migrants over the age of 15 years, who have free access to the school nurse's services. Our objective was to describe the primary complaints for spontaneous visits to the school nurse, placing them in the context of the school nurses' role in addressing the health needs of young migrants.
Methods: The population consisted of 450 students over 15 years old, enrolled at one public school in Geneva (Service de l'accueil du postobligatoire). Students' spontaneous visits to the nurse's clinic and their primary complaints were recorded between September 2013 and June 2014. The complaints were grouped into categories and described using frequencies.
Results: A total of 231 visits were recorded, and the primary complaint was available for 212 of them (92%). Somatic complaints were the most frequent (76%), whereas mental health issues were the initial complaint in only 4% of consultations. Headaches were the most frequent reason for consultation (19%), followed by osteoarticular complaints (12%) and skin problems (including small wounds, 11%). Other reasons included social problems, help for taking medical appointments, and follow-up visits.

Conclusions: The young migrants in our population mainly consult for somatic complaints, as previously described in the literature. They usually present with minor medical problems, not needing urgent care. Nevertheless, these visits are opportunities to address other issues, such as somatic and mental health and psychosocial problems, frequent in this population. This stresses the role of the school nurse as a low-threshold point of access to the health system and as an aid to orientation towards appropriate care.

FM 31

The research culture among nurses working in the paediatric department of a university hospital in western Switzerland

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Background: Nursing research is essential to build up evidence-based practice, which improves quality of care, patient outcome, and nurse satisfaction in the workplace. Furthermore, it responds to societal and legal requirements. Despite an increasing amount of evidence-based recommendations for the practice, they remain underused in practice.
Aim: This study aimed at defining the research culture among nurses working in the paediatric department of a university hospital in western Switzerland.
Method: The design was a descriptive and correlational study. An online self-administered questionnaire: the Research culture questionnaire was chosen. It contains 120 items, which explore knowledge, attitudes and use of research by nurses as well as barriers and facilitators to read and conduct research. The questionnaire was first translated to French and adapted to the Swiss context. Then it was sent by e-mail to all nurses of the department.
Results: Descriptive and correlational analyzes showed that nurses were interested in research. Of the 201 nurses who answered the questionnaire (41% response ratio), half participated in research promotion activities but only a third considered their level of knowledge in research as high or medium. A third of them regularly read scientific articles, however only 11.3% often used research results to develop clinical guidelines or to make clinical decisions. Attitudes towards research were positive. Most considered it essential for developing care and for the quality of care. The main barriers identified were lack of time, conviction, financial and emotional support and knowledge. The facilitators identified were training in research and personal motivation.
Conclusions: Despite positive attitudes towards research, only one nurse out of ten will use research results to develop clinical guidelines or to make clinical decisions. The barriers and facilitators identified demonstrate a significant need of time and training in research. Recommendations for the practice would be to reinforce the links with researchers, already present through the existing partnership within the department. A way to achieve this goal could be to invite nurses to a more regular presence at conferences and setting up workshops or journal clubs.

FM 32

Haemophilus influenzae type b epiglottitis and meningitis despite adequate vaccination

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Background: Monovalent Haemophilus influenzae type b (Hib) conjugate vaccine leads to protection against invasive Hib disease after completion of three doses in 98–100% of all patients [1]. Nevertheless, invasive Hib disease can occur despite routine infant immunization against Hib.
Case presentation: Case 1: A 3 2/12 year old boy vaccinated four times with conjugate Hib vaccine at the age of 2, 4, 6 and 18 months, presented with epiglottitis and severe respiratory distress. Hib was cultured both in blood and tracheal secretion. He was ventilated for 5 days, given amoxicillin/clavulanic acid for 10 days, and discharged in good condition after 12 days. Immunologic evaluation five months later showed normal concentration of IgM, IgA and IgG (and subclasses). Hib antibodies were low (<0.15 ug/ml), but showed good response to a booster Hib vaccine (3.2 ug/ml). Case 2: A 1 3/12 year old girl vaccinated three times at the age of 2, 4 and 6 months was admitted with bacterial meningitis. Hib was cultured both in blood and cerebral spinal fluid. Immunologic evaluation revealed low Hib antibody concentration (<0.15 ug/ml), low total IgM (0.41 g/l) and normal total IgG. She was given ceftriaxone for 7 days, and was discharged in good condition after 7 days. The immunologic response to a booster Hib vaccine isunknown.
Discussion and Conclusion: Estimated monovalent Hib conjugate vaccine failure is rare, occurring in 2.2/100,000 vaccinees [2].Clinical presentation encompasses meningitis (65%), epiglottitis (20%), pneumonia (6%) and septic arthritis/osteomyelitis (6%) [3]. We present two previously healthy children with invasive Hib disease and insufficient Hib antibody concentration despite adequate Hib vaccination. Reasons for vaccine failure include an underlying immunodeficiency or fading immune protection secondary to physiologically time- dependent declining Hib antibodies. The majority of cases respond to a booster Hib vaccine with an adequate increase of Hib antibodies [3].

References: 1. Fritzell B, et al., J Pediatr. 1992;121:355.
2. Ladhani SN. Clin Ther. 2012;34:385.
3. Heath PT, et al., Clin Infect Dis.2000;31:973.

FM 33

Violence, love and bitter pills

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Various reasons, especially the size of pills and the bitter taste of medications, often lead to the refusal of oral medications in children. In order to fulfil their responsibility to administer the drugs correctly, parents use different strategies to overcome this dilemma of which one is violence. Parents usually don't speak about these powerful strategies. This taboo is generated by the expectation to satisfy their mission – to administer the pills, correctly – and the love of their child. At first they experience pity, which then turns into feelings of guilt. These two behavioural roles are not compatible and generate a taboo. What do we know about the contexts in the paediatric oral medication management from research? Primarily there are some publications with statements on the acceptability of various pharmaceutical forms in children. Furthermore, there are studies that have examined the effectiveness of a structured pill swallowing training in children. In some of these studies, experiences of parents in such challenging situations are described, but not scientifically analysed. The Children's Hospital of Eastern Switzerland, support children with an evidence-based pill swallowing training. This includes a professional approach to educate the children and the use of a vast assortment of education material, adapted to the developmental stage of the children. Examples of such educational units will be shown in form of a movie. A comprehensive education in paediatrics should not only focus on the child, but also include their parents. This is a necessity, for parents and their children are to be understood as one unit in which the members influence each other. An integration of the parents into the existing education would improve the quality of treatment to a great extent. Therefore physicians should emphasize this problem very early in the stage of their treatment and when problems occur, send those children to a nurse specialized in pill swallowing training. The next step should and will be generating knowledge about the experience of parents exposed to ethical dilemmas in oral medication management of their children, using grounded theory as a qualitative, inductive design.

FM 34

The use of 5-aminosalicylic acid in children and adolescents with inflammatory bowel disease in Switzerland

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Background: Guidelines for the treatment of paediatric inflammatory bowel disease (IBD) include local and systemic therapeutic options. In ulcerative colitis (UC) 5-aminosalicylic acid (5-ASA) is recommended as primary therapy for mild to moderate disease; furthermore there is strong evidence demonstrating the benefits of topical 5-ASA in distal

disease and in combination therapy for extensive disease. In Crohn's disease (CD) the evidence for a beneficial role of 5-ASA is weak.
Methods: Data of patients <18 years, registered between April 2008 and February 2015 in the Swiss Inflammatory Bowel Disease Cohort, were analyzed.
Results: 280 pediatric IBD patients were included: 149 with CD and 131 with UC. Most UC patients presented with extensive colitis or pancolitis (85 [65%]) and only one quarter with left-sided colitis or proctitis (33 [25%]). One third of UC patients (44 [34%]) received topical 5-ASA therapy and one quarter (33 [25%]) combination therapy. UC patients with left-sided colitis or proctitis were more likely to receive topical or combination therapy as compared to patients with pancolitis ($p < 0.001$ and $p = 0.004$, respectively). Analyzing the usage per year, an increase in the use of topical 5-ASA therapy in UC patients was noted over time (from 3% in 2009 to 20% in 2014). The use of topical 5-ASA was found to be independent of disease duration and age of diagnosis. Interestingly, topical 5-ASA users were more likely to be female than male ($p = 0.044$). Most CD patients had colonic or ileo-colonic disease (117 [79%]), 13% ileal disease. Fifty percent of CD patients (74 [50%]) were treated with oral 5-ASA at some point during their disease course. The use was independent of disease location – ileal, colonic or ileo-colonic ($p = 0.32$). The usage of oral 5-ASA was stable over time, with each year 15–20% of CD patients receiving oral 5-ASA.
Conclusion: In recent years a very positive trend showing an increase in topical 5-ASA therapy in children and adolescents with UC has been demonstrated. However topical therapy is still underused, especially in patients with a more extensive disease. Conversely, despite weak evidence supporting 5-ASA use in CD patients it was frequently prescribed. Physicians should continue to encourage their UC patients to use topical therapy.

FM 35

Prolonged postictal asystole in a child with focal epilepsy and cerebellar atrophy

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Background: Cardiac arrhythmias can be observed in the context of seizures. Seizure-induced asystole has been associated with focal epilepsies originating from the temporal and frontal lobe. We present a case with video recording during sleep EEG with postictal asystole and discuss the risk factors for SUDEP (sudden unexplained death in epilepsy).
Case presentation: A 12.5 years old girl was diagnosed with a focal epilepsy at the age of 6 years. Initial neurocognitive testing as well as MRI of the brain were normal. Under antiepileptic treatment a seizure frequency of 1–2/year was reported and regular follow-up EEGs were normal. Marked neurocognitive decline initiated follow-up investigations. Brain MRI revealed progressive cerebellar atrophy. Sleep-EEG recorded a secondary generalized seizure with left fronto-temporal origin lasting for 85 seconds followed by postictal asystole for 7 seconds. After an isolated QRS complex, asystole continued for another 27 seconds with a low voltage EEG trace. Another 3 isolated QRS complexes were noted over 14 seconds before regular heart beat reoccurred. Flat EEG trace lasted for 130 seconds. An interdisciplinary decision was taken and the patient admitted for transvenous single chamber pacemaker implantation.
Conclusion: The coexistence of cerebellar atrophy and fronto-temporal epilepsy increases the risk for SUDEP. Repetitive postictal asystole with prolonged EEG recovery time, together with progressive cerebellar atrophy might be responsible for the neurocognitive decline in this patient. Careful monitoring is mandatory to identify patients at risk for SUDEP. Decision for preventive pacemaker implantation should be taken individually for each patient.

FM 36

Which electronic database do Swiss healthcare professionals consult for paediatric dosages?

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Background and aim: Determining the correct dosage of a drug for children may be delicate. Drugs are often used "off-label" or "unlicensed" and often no recommendations are available in the summary of product characteristics. The aim of our survey was to analyze the behavior of Swiss paediatricians and pharmacists regarding their use of electronic databases with children's dosages.
Methods and Results: An online-survey was sent to Swiss paediatricians (N = 1806) and pharmacists (N = 2073) by e-mail. The rate of return was 23% (N = 882). Over 70% of all the participants use electronic drug databases at least once a week, 36% of them daily. Six percent answered, that they never use electronic drug databases. By trend, with increasing experience healthcare professionals use electronic databases less frequently. Focusing on children's dosages, the utilization of different information systems was analyzed: websites on the PC are most often used (53% use them at least once a week), followed by guidelines of the respective institution (24%). For paediatricians on the children's ward those guidelines are crucial, 57% use them at least once a week. In contrast, paediatricians in a paediatric practice use websites most often (50% at least once a week). However, only 13% of the Swiss healthcare professionals use Apps at least once a week to look up pediatric dosages, 71% never use Apps for this purpose. Most utilized database to look for a pediatric dosages is compendium.ch, 56% use it at least once a week. Second leading database is the website of the University Children's Hospital Zurich (kinderdosierungen.ch or posologies-pediatriques.ch). This website is consulted by 19% of the Swiss healthcare professionals at least once weekly. Analyzing the subgroup paediatricians showed that they mostly use the compendium.ch (60% at least once a week) followed by kinderdosierungen.ch/posologies- pediatriques.ch (25%) and the "Frank Shann" (19%).
Conclusion: Our results show that electronic information about paediatric dosages is crucial. The most important source for paediatric dosages is the summary of product characteristics. The paediatric dosage database of the University Children's Hospital Zurich is consulted by paediatricians and pharmacists alike. We guess that especially in case of "off-label" or "unlicensed use", this website is a helpful link for Swiss healthcare professionals.

FM 37

Prevalence of medication errors in paediatrics and correlation with risk factors

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Introduction: Medication errors contribute to the emergence of preventable adverse drug events and therefore should be minimised. The paediatric population is especially vulnerable for medication errors.
Objectives: The focus of this project was on medication errors during drug preparation on the ward. The prevalence of medication errors was determined and potential risk factors for medication errors identified by analysing frequency and number of medication errors. The analysed potential risk factors were: route of administration, number of interruptions during drug preparation, the level of nurses' work experience.
Setting: Swiss central hospital with paediatric department providing tertiary care.
Method: Analysis of processes during prescribing, drug preparation, administration and documentation between December 2013 and March 2014. Prescription and documentation errors were identified by reviewing medication order sheets and medication administration records. Dispensing and preparation errors were identified by direct observation. Medication administration errors were identified by interviewing staff. Medication errors were divided into mistakes or slips and violations.
Main outcome measures: Frequency of medication errors, number of medication errors per medication events.
Results: 402 medications events were recorded. 125 medication events were correct (31%), the same amount had mistakes or slips. 152 medication events had only violations (38%). A total of 427 medication errors were identified of which 170 were mistakes or slips and 257 were violations. A higher risk for occurrence of medication errors was detected for drugs applied via enteral feeding tube. Interruptions were positively correlated with mistakes and slips during drug preparation. Nurses' work experience showed no impact on the quality of medication events.
Conclusion: Prevalence of mistakes and slips in the medication process were similar to previous observations. Patients receiving drugs via enteral feeding tube seem to be a population at risk for medication errors. There are clear opportunities for making system changes in order to reduce medication error rates.

SPN 1

Motor and mental outcome of symptomatic neonatal arterial ischaemic stroke: Prognostic value of early neuroimaging findings

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Background: Neonatal ischaemic stroke is known to be associated with cerebral palsy (CP), epilepsy and cognitive impairment. Involvement of the corticospinal tract is known to be predictive for poor motor outcome.
Aim: The aim of the study is to test the prognostic value of neuroimaging data at stroke onset regarding CP symptoms and mental development after 2 years.
Method: We included 58 children with a mean age of 2.2 ± 2.1 days at stroke manifestation. MRI was performed within the first 15 days of life. Infarct localizations were compared with CP and mental development at 2years.
Result: Of all children, 37.9% had CP and 24.1% showed delayed mental development. The infarct was bilateral in 24.1%. All infarctions involved the middle cerebral artery (MCA) territory, 37.9% had also an involvement of the posterior cerebral artery(PCA). Children with MCA/PCA involvement had a higher risk for CP and for impaired mental development. The highest odds ratio regarding CP at 2 year was found for the involvement of the thalamus and the basal ganglia (OR = 17.0, 95% CI 3.26–88.77). Children with thalamus involvement are 8.1 times as likely to have impaired mental development as children without thalamus involvement (95% CI 1.99–33.05).
Conclusion: Early MRI in children with neonatal ischaemic stroke has a prognostic value for motor and mental development. Interestingly, involvement of the thalamus and the basal ganglia was more predictive for CP than the internal capsule. Moreover, involvement of thalamus seems to play a crucial role in predicting mental development 2 years post stroke.

SPN 2

Longer is not better in eradication of *Pseudomonas aeruginosa* primo-infection in paediatric Cystic Fibrosis patients

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Introduction: Patients with Cystic fibrosis (CF) are more susceptible to pathogens like *P. aeruginosa* (PA). PA primo-infection requires particular attention as failure in eradication is associated with accelerated lung deterioration. However, there is non consensus in the literature towards an optimal eradication protocol. The main aim of this study is to assess the rate of PA eradication following our particular protocol with 3 months of inhaled tobramycin and oral ciprofloxacin, putting it in relation to published data of shorter treatment regimen.
Methods: Retrospective single centre study with data analysis from June 1st 2007 to December 31st 2015 of patients with 1 or more PA primo-infection(s) exclusively treated by 3 x 28 days of inhaled tobramycin and oral ciprofloxacin for the first and last 21 days. Success in eradication is defined by ≥3 negative cultures for 6 months after the beginning of the protocol. Eradication is failed if ≥1 culture is positive in the same period.
Provisional results: Out of 46 patients with PA primo-infection(s), 31 followed the eradication protocol and were included in our analysis (11 girls and 20 boys). Girls had 17 primo-infections and boys had 27. Among these 44 primo-infections, 32 (72.7%) had an overall success in eradication and 12 (27.3%) a failure. Rate of success was 82.4% for girls and 66.7% for boys.
Provisional conclusion: Our protocol succeeded in an overall eradication rate of 72.7%, which is similar to published data of shorter eradication protocols. We found also a difference between boys and girls, but not significant (p-value 0.25428). Detailed analysis of individual patients will be performed regarding compliance, but as our protocol is also associated with increased treatment burden and costs, we might change to a shorter eradication protocol.

SPN 3

Neonatal programming of functional gastrointestinal disorders in infants

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Objectives and study: Functional gastrointestinal disorders (FGIDs) are common in infants, represent a frequent condition of parental distress and pediatric referral. To date, predisposing or protective factors for FGIDs still need to be clarified but could be crucial to identify preventive strategies. The aim of this study was to assess the influence of different neonatal factors on the incidence of FGIDs in the first months of life.
Methods: This is a prospective multicenter study including preterm and at term newborns consecutively enrolled at birth and followed up till one year of age. Exclusion criteria were represented by: malformations, (any kind of) surgery, neurological, immune, metabolic, cardiac or renal diseases or incomplete follow-up. FGIDs were classified according to Rome III criteria and assessed through a standardized interview by a dedicated physician at each hospital. Data were collected using a specific form at 1,3,6, and 12 months. Gestational age, mode of delivery, feeding pattern, antibiotic administration in neonatal period, and duration of hospitalization at birth were considered. Statistical analysis was performed by JMP program (version 11) using chi square test and multivariateanalysis.
Results: 1152 newborns (gestational age 163–297 days, 337 preterm, 29%, and 815, 71%, at term newborns) were recruited and completed the study. Preliminary analysis showed an overall significantly ($p < 0.0001$) higher incidence of FGIDs during the first year of life in preterm compared to at term newborns, and particularly of regurgitation (47% vs. 39%, $p = 0.019$) and colic (60% vs. 45%, $p < 0.001$). Overall FGIDs were significantly ($p < 0.001$) more reported in infants born with caesarean section (OR 1.7) or given antibiotics in the first week of life (OR 2.1), or with a long hospitalization at birth (OR 1.9). Regurgitation was also more frequent ($p < 0.001$) in infants who had a longer (>7 days) hospitalization at birth. Colic was significantly associated with preterm delivery, low birth weight, neonatal antibiotics, duration of hospitalization and formula feeding.
Conclusion: Preterm delivery and neonatal use of antibiotic are associated with an increased incidence of FGIDs in the first months of life. Caesarean section, formula feeding and longer hospital staying at birth may represent additional risk factors determining the higher prevalence of FGIDs in preterm compared to at term newborns and need to be further analysed.

SPN 4

The influence of different fever definitions on diagnostics and treatment after diagnosis of fever in chemotherapy-induced neutropenia in children with cancer

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Background: There is no evidence-based definition of the temperature limit defining fever (TLDF). Lowering the TLDF is known to increase the number of episodes of fever in neutropenia (FN). This study aimed to investigate the influence of a lower versus standard TLDF on diagnostics and therapy after FN diagnosis.
Methods: In a single pediatric cancer center using a high standard TLDF (39 °C tympanic temperature) patients undergoing chemotherapy were observed prospectively (NCT01683370). Results of all temperature measurements and key procedures of diagnostics and therapy during FN were recorded. The effect of applying lower TLDFs (range 37.5 °C to 38.9 °C; reported here, 38.0 °C) versus 39.0 °C on these measures was simulated in silicon.
Results: In reality, 45 FN episodes were diagnosed in 20 of 39 study patients (maximum, 6 episodes per patient). Of 3391 temperatures measured, 193 were ≥39.0 °C, and 937 ≥38.0 °C. For persisting fever ≥24 hours, additional blood cultures were taken after start of antibiotics

in 31 (69%) episodes in reality. This number decreased to 22 (49%) by virtually applying 39.0 °C, and increased to 33 for 38.0°C (73%; plus 11 episodes; plus 24%; 95% CI, 13 to 40). For persisting fever ≥48 hours, intravenous antibiotics were escalated to broader coverage in 25 (56%) episodes. This number decreased to 15 (33%) by virtually applying 39.0°C, and increased to 26 for 38.0°C (58%; plus 11 episodes; plus 24%; 95% CI, 13 to 40). For persisting fever ≥120 hours, intravenous antifungals were added in 4 (9%) episodes. This number increased to 6 (13%) by virtually applying 39.0°C, and nearly doubled to 11 for 38.0°C (24%; plus 5 episodes; plus 11%; 95% CI, 4 to 24). In reality, the median length of stay was 5.7 days (range, 0.8 to 43.4). In 43 episodes with hospital discharge beyond 24 hours, virtually applying 38.0°C instead of 39.0°C led to discharge delay by ≥12 hours in 24 episodes (56%; 95% CI, 40 to 71), with a median delay of 13 hours, and a cumulative delay of 68 days.
Conclusion: Applying a low versus standard TLDF led to relevant increases of diagnostics, antimicrobial therapy, and length of stay in children with FN. This, in turn, may increase treatment-related side effects and costs, and may decrease quality of life. The differences between management in reality versus simply applying 39.0° as TLDF reflect the important impact of clinical assessment, which is not modeled here.

SPN 5

Pertussis immunisation in pregnant women: Adherence to OFSP's recommendations

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Introduction: In 2013, the Office fédéral de la santé publique (OFSP) issued a recommendations advising booster immunisation of pregnant women against pertussis if the last infection or immunisation was more than five years old. We had the impression that the implementation of these recommendations was incomplete and decided to design a study to confirm this impression and help search for improvement.
Method: We prepared a questionnaire in order to gather information concerning the vaccinal status of women just after delivery. Given the results of the first period of study, we decided to issue a mailing to the obstetricians reminding them of the recommendations of the OFSP. We repeated this study 1 year thereafter to evaluate the impact of our action.
Results: During the period between 15 March 2014 to 30 April 2014, 21.6% pregnant women were proposed the immunisation according to the recommendations. Among the 82.4% remaining, only 4.1% had a sufficient immunisation status. We concluded that the opportunity to immunise pregnant women according to the recommendations was missed in 74.2%. The adherence of the OFSP guidelines was just 24.7%. The second survey conducted between 14 September 2015 to 25 October 2015 did not show any improvement despite a mailing reminding of the OFSP guidelines and proposing help and counseling. Adherence to guidelines was 24.4% and opportunity was missed in 66%. Our action clearly missed its target. The acceptance was excellent amongst women. In 2014, 76.2% accepted the immunisation if it was proposed during pregnancy and 67% accepted the backup immunisation proposed after delivery. In 2015, the acceptance was 100% and 50% respectively.
Conclusion: The implementation of the OFSP guidelines is clearly insufficient in our region. Further work is needed to evaluate the adherence to the recommendations in Switzerland and to search for an effective way to improve their application.

SPN 6

EEG sleep slow wave activity as a marker of load-dependent executive function deficits in very preterm children

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Background: Many children born very preterm experience difficulties in executive functions, e.g., planning, with the deficits often only becoming evident when the cognitive load is high. Sleep slow wave activity (SWA, 1–4.5 Hz EEG power), the key characteristic of deep sleep, reflects the degree of synaptic strength and network synchrony. SWA has been shown to be locally increased after the intense use of a certain cortical region, e.g., after an intensive cognitive training. This study investigated whether sleep SWA is an electrophysiological marker of local alterations in executive function networks in very preterm children.
Methods: A group of 38 very preterm children without any severe neonatal brain injuries (age at assessment [M ± SD] 12.9 ± 1.7 years) and 43 health term-born peers [13.1 ± 2.0 years] were assessed with a comprehensive battery of executive function measures. A composite score was calculated to reflect the ability to cope with increasing load. All-night high-density sleep EEG (128 electrodes) was recorded in all participants. SWA averaged across the first hour of NREM sleep was obtained and correlated with the composite score.
Results: Sleep efficiency was high in both groups (approximately 90%). The architecture and quality of sleep were not significantly different between the groups ($p > .41$). Very preterm participants had significantly lower abilities to cope with increasing load ($p < .001$). Looking at all participants, the composite score was positively correlated with SWA in a cluster of 14 electrodes over frontal brain regions ($r = .32 \pm .06$, $p < .05$). Within this cluster, SWA was higher in those participants with better coping abilities than in those with worse coping abilities ($p < .05$). Additionally, comparing the two groups, very preterm participants showed higher SWA compared to term-born participants when task demands were taken into account ($p < .05$).
Conclusion: The local increase of SWA in very preterm children compared to similarly performing term-born peers may reflect the more intense use of neuronal networks underlying executive functions to achieve normal performance. However, if demands are highest, this compensatory mechanism may fail and lead to the load-dependent executive function deficits often reported in very preterm children. This study shows that sleep SWA may represent a marker for load-dependent alterations in brain networks known to be involved in executive functioning.

SPN 7

Neurodevelopmental Outcome in children with congenital heart disease at school-age

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Background: Children with severe congenital heart disease (CHD) are at risk for adverse neurodevelopmental outcome. There is limited information on neurodevelopmental outcome at school-age and most studies excluded children with genetic syndromes. This study reports neurodevelopmental outcome and its risk factors for a current cohort of children with CHD including those with a genetic disorder (GD).
Methods: Outcome at six years was assessed in 233 (64 with GD) prospectively recruited children with severe CHD after infant full-flow cardiopulmonary bypass surgery at the University Children's Hospital Zurich. 198 had a biventricular (d-TGA n = 43) and 35 a univentricular heart defect (HLHS n = 11). Neuromotor and cognitive testing was performed using age-appropriate, standardised tests.
Results: Median IQ was 95 (range 47–135) in children without a GD and 55 (3–115) in children with a GD (median trisomy 21: 49, microdeletion 22q11: 74.5, other GD: 59). IQ scores were below the norm for both groups (non-GD $p < 0.001$, GD $p < 0.001$). The rate of IQ scores below 85 (–1 SD below 100) in children without GD was higher than the norm (22.5% versus 15.9, $p = 0.02$). Children with and without GD showed significantly lower z-scores in all neuromotor components. Independent predictors for IQ at 6-years included: Presence of GD, lower socioeconomic status, 1' Apgar, abnormal preoperative neurological examination, preoperative feeding difficulties, postoperative seizures, poorer cognitive outcome at 1-year and cardiac medication at the time of assessment (R^2 0.70, $p < 0.01$ for the model).
Conclusions: Children with CHD show mild neurodevelopmental impairments at school-age. Risk factors for poorer outcome include GD, perioperative neurological complications, and cardiac insufficiency at follow-up while surgical factors are less important. Close neurodevelopmental surveillance of these children is necessary to provide early therapeutic support.

SPN 8

Childhood arterial ischaemic stroke: the influence of lesion location on outcome

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Background: Childhood arterial ischaemic stroke (AIS) causes considerable sequelae affecting daily life activities. Data regarding the effect of lesion location on outcome are inconsistent. This study aimed to analyse the impact of lesion location on long-term outcome in children with AIS.
Methods: Prospective multicentre cohort study including consecutive children aged between 1 month and 16 years experiencing AIS from 2000–2014. Localisation was divided into: cortex/white matter, thalamus, basal ganglia and infratentorial. Longitudinal outcome assessment was performed using the Paediatric Stroke Outcome Measure (PSOM) at discharge, 6 and 24 months after AIS. PSOM was dichotomized into good (0, 0.5) and poor outcome. Logistic regression models were used to adjust for confounders.
Results: A total of 206 children (129 males, 63%) with a median (IQR) age of 5.9 years (2.1–12.0) were included in the study. Median (IQR) pedNIHSS at stroke onset was 6 (3.0–10.0). Poor outcome was noted in 122/202 (60%), 73/166 patients (44%) and 55/130 (42%) at discharge, 6 and 24 months after stroke respectively. Higher pedNIHSS at stroke onset ($p < 0.001$) as well as lesions affecting cortex/white matter (discharge: $p = 0.007$; 6 months: $p = 0.021$; 24 months: $p = 0.006$) or basal ganglia (discharge: $p = 0.027$; 6 months: $p = 0.004$; 24 months: $p = 0.005$) correlated with poor outcome at each assessment.
Conclusions: Almost half of the patients with AIS show a poor outcome 2 years after acute stroke. Ischaemic lesions located in the basal ganglia or cortex/white matter and pedNIHSS at stroke onset are independent predictors for poor outcome throughout the recovery period.

SPN 9

School-based TBC screening in recently arrived young migrants in Geneva: Constraints and benefits

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Introduction: Systematic health checks by the school nurse can help identify young people's health needs and orient them within the health system. In Geneva, tuberculosis (TB) screening by tuberculin skin testing (TST) is offered to all recently arrived young migrants coming from countries with high TB incidence (according to WHO criteria) during their first year at school. Recommendations concerning TB screening in these populations vary: WHO and experts in Geneva are in favor, whereas the Swiss Lung Association estimates the risk to be too low to recommend systematic screening. The aim of our study is to review data from Geneva and put them in the context of current procedures and the international literature.
Method: We analyzed TST's in young migrant pupils aged 16 to 19 years during one academic year (2013–14) in one school in Geneva. Pupils with reactions of 5 mm or more were oriented towards a primary care physician for IGRA (Interferon Gamma Release Assays) testing. We also reviewed the recent international literature on school-based TB screening.
Results: Over one year, 57 TST's were performed, 7 adolescents were sent for medical consultation because of a positive TST, 5 of which had a positive IGRA test (4 were diagnosed with latent TB, one had active TB). The literature review showed that TST's are more often used for screening than IGRA. Recently arrived young people from countries with high TB prevalence are considered at risk for active TB and school- based screening is offered in several countries. We found no publications describing other benefits of the contact with the school nurse.
Conclusion: School-based screening identified 4 cases of latent TB and one case of active TB in one year, although our findings are limited to one year and one school. In the future more complete data should be obtained, and compliance to treatment and the health system use of these young immigrants should be further studied. The coordination between school health services and primary care physicians is important for psychosocial aspects as well as the prevention of infectious diseases.

SPN 10

Respiratory viruses in healthy infants and infants with Cystic Fibrosis. A prospective cohort study

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Background: Acute respiratory tract infections in children with Cystic Fibrosis (CF) are known to cause exacerbation of disease, lower lung function and hospitalization. Viral infections are reported as the most frequent underlying cause. The implementation of the newborn screening for CF allows early diagnosis and opens new opportunities in preventative and therapeutic approaches early in life. Before this can be done, a better understanding of respiratory health and viral colonization during infancy is needed.
Methods: 31 infants with CF from the Swiss Cystic Fibrosis Infant Lung Development Cohort (SCILD) and 32 unselected, healthy infants (HC) from the Basel Bern Infant Lung Development Cohort (BILD) cohort were included in this prospective longitudinal study within the first year of life. Pre- and perinatal information was collected from the families. Respiratory symptoms were assessed with a weekly standardized telephone interview. Biweekly nasal swabs were taken and analyzed for 12 different viruses with Multiplex PCR (CF = 576, HC = 718).
Results: While viral colonization in general did not differ between the two groups (mean 42% vs. 43%), respiratory symptoms during infections were significantly less frequent in infants with CF (38% vs. 49%; OR 0.62; 95% CI 0.41–0.93; $p = 0.022$). This finding was pronounced for symptomatic infection with Human Rhinovirus (HRV) (6% vs. 10%; OR 0.49; 95% CI 0.26–0.95; $p = 0.035$), whereas colonization with Bocavirus occurred significantly more frequent in infants with CF (10% vs. 3%; OR = 2.55; 95% CI 1.22–5.34; $p = 0.013$).
Conclusion: Surprisingly, in this study, viral colonization was not more frequent in infants with CF compared to healthy controls and respiratory symptoms during virus infection occurred even less often in infants with CF. While we can only speculate about underlying reasons, our findings indicate a less important role of viruses in early CF lung disease compared to older patients with CF.

SPN 11

Brazil Micronutrient project

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The field of nutrition science today is evolving from the analysis of a single or a few nutrients that may affect a specific phenotype (e.g., growth or disease) to a more holistic analysis of the system. In the past, researchers and policy makers focused primarily on carbohydrate, lipid and protein as the cause of children under nutrition or obesity. Focusing on a single nutritional cause obscured the broader malnutrition that may exist in a child with obesity. The role of multiple micronutrients and their interactions has not been studied in health nor in chronic diseases. For example, vitamin D and B group vitamins have been reported to be deficient particularly in morbidly obese individuals and in those with metabolic syndrome. Obese individuals with metabolic syndrome have also been shown to have a lower level of the antioxidant vitamins C and E. However, whether micronutrients insufficiencies are affected by obesity or contribute to its cause remains difficult to be established, especially in humans. Reductionist approaches to complex phenotypes resulted in: (1) exaggerated claims for nutrition as a cure or prevention of disease; (2) the wide use of empirically based dietary regimens, as if one fits all; and (3) frequent disappointment of consumers, patients, and healthcare providers about the real impact nutrition can make on medicine and health. We described here a multi-micronutrient (12 vitamins and 5 minerals) intervention study in children aged 9 to 13. It was a community based participatory n-of-1 study of Brazilian children and adolescents in Ribeirão Preto, Brazil, based on the dietary, anthropometric, and physiological measures at baseline, after 6 weeks of 5 day per week supplementation, and after 6 weeks without supplementation. This

systems approach study was performed in two consecutive years (2013 and 2014) with identical methodology and with repeated or new participants in 2014. Fasting levels of glucose, LDL-cholesterol, and total cholesterol decreased after the intervention at the population level (the average response from baseline to post intervention). The results were replicated and validated in 2014. In depth phenotypic responses based on proteomic and metabolomics analysis along with whole genome genotyping (5M features) and exome analysis were also done. We will present preliminary population data and show interindividual variability in response to multi-micronutrient supplementation.

POSTERFLASH POSTERS

PF 1

Perinatal infratentorial haemorrhage: a rare but possibly life-threatening condition

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Background: Perinatal infratentorial haemorrhage (PIH) is a rare and clinically difficult to diagnose condition and has been described as a complication of abnormal labor and vacuum extraction.
Case presentation: After an uneventful pregnancy, a baby boy was born at 41 weeks gestational age. Due to fetal bradycardias birth was facilitated by vacuum extraction (Kiwi). Initial pH was 7.20 and Apgar Score 3/6/9. After initial resuscitation insufficient breathing, areactive pupils and absence of spontaneous movements were noted. The newborn required intubation and was transferred to the ICU. Diagnosis of neonatal asphyxia with hypoxic- ischemic encephalopathy (HIE) Sarnat stage III was made and therapeutic hypothermia (TH) for 72 hours was initiated. A cerebral ultrasound was conducted, which showed no particularities apart from a mildly hyperechogenic periventricular substance. There were no signs of cardiac, renal or hepatic affection. At physiological body temperature, routine cerebral MRI showed a subdural haemorrhage in the posterior fossa with compression of the 4th ventricle. Signs for HIE had resolved, but periodic breathing and transient bradycardia were still present. Close ultrasonographical and clinical controls were installed to observe for hydrocephalus development, a surgical intervention was not necessary.
Conclusion: Perinatal intracranial haemorrhage occurs in up to 25% of full term neonates, mostly localized in the ventricles. PIH is an atypical location which can be easily missed with ultrasound. It can be life-threatening with compression of the brainstem and development of hydrocephalus requiring surgical intervention. Therefore, in children with birth trauma, PIH may be an important differential diagnosis of HIE.

PF 2

Sports for Children with Myelomeningocele (MMC) – Ready, set, go!

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Introduction: There is a necessity of physical activity and sports as prerequisite for a good functional outcome for children with postnatal and in particular children with fetal MMC repair. However, there is still a gap between the actual knowledge regarding the positive effects of sports and its implementation in real life. This work aims to examine the impacts of sports and to convey practical recommendations in order to increase sports activities of children with MMC.
Methods: This survey is based on current literature in medical databases and literature of sports education for disabled children as well as interviews with health professionals and parents.
Results: Due to many reasons children with MMC develop an inactive lifestyle during adolescence. Obesity and a lack of physical activity lead to a vicious circle of loss of functional independence and mobility and eventually social isolation. This leads to a critical decrease of the health related quality of life of children with MMC. Children with fetal repair benefit from a better ambulation function in early childhood. The postnatal repair group shows that good ambulation function in early childhood does not prevent obesity. To preserve the benefit of the fetal repair, these children have to be encouraged even more to do sports. Frequent sports activities enables children to achieve a higher level of functional independence and mobility and leads to many peer relations. Children with MMC can perform many sports like handbiking, swimming or skiing. Sport clubs offer every kind of wheelchair sports to these children. Sports for children with MMC is only contraindicated after scoliosis surgery and new onset of symptoms of tethered cord. Moreover jumping on a trampoline regularly is not recommended. It is very important to inform parents about the benefits of sports for their child and about the offers of sport clubs and camps nearby. Parents are the ones who enable their child to participate in sports and the child needs the family's support.
Conclusion: Exercising regularly improves the health related quality of life in children with MMC. Peer relations prevent these children from social isolation. Based on current data sports and a high level of physical activity in daily life are recommended for children with MMC – it is important to start in early childhood and stay active during adolescence.

PF 3

Quasi-spontaneous epidural haematoma in a 7-year-old girl

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Traumatic and spontaneous spinal epidural hematomas (SSEH) are extremely rare in children, with an incidence of 0.1/100'000 infants per year. We report a 7-year old girl who presented with a 10 hours history of neck pain following a trivial flexion/extension neck movement during her music class. Within 40 hours, with gradually increasing cervical pain, she developed progressive right-sided hemiparesis leading to a cervical cord compression suspicion. An emergency spine MRI demonstrated an acute/early subacute right posterior epidural haematoma extending from C2/3 to T1, compressing the spinal cord. She underwent an emergency right C4-6 hemilaminectomy with evacuation of the underlying haematoma. No evidence of vascular malformation was found in the neuroimaging studies. Coagulation studies remained without abnormalities. Clinical follow-up 6 months later revealed a complete neurologic recovery. On top of being little-known, SSEH clinical presentation is often nonspecific, leading to diagnosis delay and, thus, post operative residual neurological deficits. A comprehensive review of literature of SSEH revealed 104 patients younger than 18 years old with reported clinical outcomes. Ours is the 105th case. It highlights that any discrepancy between major pain and minor, trivial (or even lack of) trauma should alert the clinicians.

PF 4

Epilepsy surgery in children and adolescents: first experiences of a collaborative project in Zurich

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Background: The incidence of childhood-onset pharmacoresistant focal epilepsy is estimated at 11.3 per 100 000 per year. Epilepsy surgery is an established treatment option for selected pediatric candidates, primarily aiming at seizure alleviation and reduction of antiepileptic drug burden. In Zurich, the Department of Neuropediatrics, University Children's Hospital, the Swiss Epilepsy Centre, Clinic Lengg, and the Department of Neurosurgery, University Hospital, have recently joined forces in a collaborative project, the Centre for Epileptology and Epilepsy Surgery (ZEE), thus also promoting paediatric epilepsy surgery.
Methods: We are presenting the presurgical findings, surgical procedures and seizure outcomes of 13 children and adolescents that underwent presurgical workup and subsequent resective surgery for pharmacoresistant focal epilepsy in the ZEE Zurich from January 14th, 2015 to January 13th, 2016.
Results: Seizure onset was in the first year of life in 5 cases (range 0.4–12.3 years). Four children suffered daily seizures prior to surgery. Mean age at surgery was 10.6 years (range 1.1–17.3). Invasive EEG recordings were required in 3 cases and intraoperative electrocorticography (ECoG) in 7 cases. Resections were temporal in 6 cases, frontal in 3 and parietal or occipital in 3, whereas one resection involved the temporo-occipital region. Etiology included focal cortical dysplasia in 9 cases and benign tumors, peri- or postnatal ischemic lesions, and hippocampal sclerosis following meningoencephalitis in infancy in one case each. At last postsurgical follow-up to date (mean 5 months), 11 of 13 children achieved seizure freedom, one demonstrated worthwhile improvement and only one did not benefit from surgery.
Conclusions: Our findings underline that epilepsy surgery in children and adolescents is highly effective in terms of seizure control. Presurgical assessment in an epilepsy center with pediatric expertise should be integrated in therapeutic regimes at an early stage, parallel to conservative management, in order to improve access and prevent treatment delay. This is crucial, since shortening the duration of epilepsy in children might result in improved long-term seizure outcome, optimal cognitive development and improved quality of life.

PF 5

Chiari Malformation Type I and sleep disorders: a case report

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We report a 5-year-old boy with a history of excessive snoring at night with respiratory pauses which had been noted by his mother. He presented also daytime sleepness with no other neurological symptom, and normal development, no previous illness. Physical examination was normal; initially obstructive apnea from adenoid hypertrophy was thought to be the most probable cause. Polysomnography was performed to investigate the sleep disorder and demonstrate 50 central apneas per hour, a respiratory rate of 5/min, with a baseline saturation of 95.4%. Cerebral MRI was performed and showed a Chiari malformation type I resulting in crowding at the craniocervical junction. He underwent nocturnal ventilator therapy, and posterior fossa decompression. Snoring diminished after decompression and physiologic improvement was confirmed by futher polysomnography studies. Chiari type I malformation typically presents in adulthood, and is assumed to rarely present clinically before adolescence. In comparison, in a young child with a history of snoring, enlarged tonsils and adenoids are the most common abnormality seen on polysomnography. The importance of the identifying sleep-disordered breathing in children is becoming more recognized. Still, practicioners do not routinely ask about sleep behavior beyond infancy, thereby missing an opportunity for early intervention and treatment. We report this case to raise awereness of this condition and to highlight the importance of obtaining a sleep history during routine health visits to detect a potentiel lifethreatening condition such as Chiari Malformation type I.

FM 6

A case of Crouzon-like craniosynostosis: correlation of intracranial hypertension with optic nerve sheath diameter measured by sonography

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Introduction: Around 30% of children with craniofacial dysostosis syndrome present with intracranial hypertension [1]. Craniosynostosis occurs most commonly as a nonsyndromic form, with an isolated synostosis. Syndromic forms are associated with a large spectrum of phenotypic features, and multiple genes have been identified [2]. Fundoscopic examination and radiological imaging provide indirect signs of intracranial hypertension, but clinical findings are often limited in young children. The gold-standard measurement of intracranial pressure is placement of an intracranial pressure (ICP) probe, but this requires a surgical procedure. Ultrasonographic measurement of the optic nerve sheath diameter (ONSD) has been investigated previously in traumatic brain injuries and intracerebral hemorrhage and shows good diagnostic accuracy with a pooled sensitivity of 90% and a pooled specificity of 85% [3]. There are fewer studies, that have compared it with invasive monitoring devices like ICP monitors. Normative values are not as yet well defined, but previous studies have defined ONSD above 5 mm as pathological [4].
Methods: We describe here, a case of a 6-month-old infant, who presented with typical phenotypic features of a Crouzon syndrome, but genetic tests (FGFR 1-2-3 and TWIST) were negative. ICP monitoring was performed before surgery, which revealed abnormally high values. Sonographic measurement of the ONSD was performed during ICP monitoring, before and after 360° decompressive craniectomy with fronto-orbital advancement.
Results: Pre-operative ultrasonographic measurements of the ONSD were 7.4 mm for the right eye, and 5.5 mm for the left eye. These pathological values correlated with ICP monitoring showing values around 30 mm Hg over a 24-hour period. Immediately after 360° decompressive craniectomy and fronto-orbital advancement, values for the ONSD were 5.9 for the right eye, and 4.9 mm for the left eye.
Conclusion: Ultrasonographic measurement of the ONSD is feasible and easy to perform at the patient's bedside, before and after surgery. It seems to correlate well with intracranial pressure measurement in cases of craniosynostosis. Further prospective studies are warranted, and therefore we have started to collect data for our patients with craniosynostosis who undergo surgical correction of this cranial malformation.

PF 7

Confusion in an adolescent: psychiatric problem, encephalitis, or ...what else?

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Case report: A pubescent Swiss-african 11-y old girl was hospitalised for headache and apyretic acute confusion: speaking difficulties, prostration, aggressiveness, disorientation. No hallucination. Workup showed uncomplicated arachnoid cyst, no other MRI cerebral anomaly; and encephalitis symptoms: CSF discrete pleiocytosis (10 leucocytes) and proteinorachy (510 mg/l) with oligoclonal bands, EEG slow waves. Despite presumptive immunotherapy, dystonic movements appeared. No immune or infectious/parasitic aetiology. Normal thyroxin. Metabolic assessment revealed hyperhomocystinaemia (HC) and methylmalonic aciduria (MMA) with normal cobalamin (B12) and folate, suggesting a cobalamin C (cblC) deficiency, a congenital disorder of B12 intracellular metabolism. Whether EEG and CSF anomalies were encephalitic trigger or cblC defect manifestation was not clear. B12 and cofactors improved situation. Ophthalmologic examination was normal. Her brother's metabolic workup was normal.
Discussion: In front of behavioural/psychotic manifestations, inborn errors of metabolism (IEM) should be searched, after exclusion of neurologic disease or intoxication, before concluding to psychiatric diagnosis. Successive steps analyses include blood count and gas, lactate, glucose, ammoniaemia, uric acid, ketone bodies; homocystinaemia, ceruleoplasmin, porphyrin; amino acid and acylcarnitin, organic acid; other specific anomalies. B12 metabolism is

implicated in central nervous and digestive functions, haematopoiesis, and vessels. It interferes with myelin synthesis, DNA and branched aminoacid metabolism. Beside nutritional deficiency; absorption, transport and intracellular metabolism anomalies are involved. CblC permits conversion of B12 in active forms. Gene responsible is MMACHC. Infant manifestations of Cblc defect are: hypotonia, seizures, lethargy/coma, failure to thrive, atypical haemolytic uremic syndrome (aHUS), cardiomyopathy, retinopathy. Late-onset: psychiatric symptoms, vascular thrombosis, myelopathy, macrocytic anaemia, digestive trouble, aHUS. Hallmarks are HC, MMA and hypomethioninemia without B 12 insufficiency. Even with vitamin and cofactors supplementation, long-term outcome is altered by neurologic and ocular impairment.
Conclusion: Treatable IEM are medical emergencies we should keep in mind in psychiatric situation, at any age. Late-onset CblC deficiency, the most common intracellular B12 metabolism error, is one of them.

PF 8

A novel synonymous mutation in *FGFR1* causes Hartsfield syndrome

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Hartsfield syndrome is a rare clinical entity characterized by the triad of holoprosencephaly, ectrodactyly and cleft lip/palate. In addition to these symptoms patients with Hartsfield syndrome can show developmental delay of variable severity, isolated hypogonadotropic hypogonadism (IHH), central diabetes insipidus, vertebral anomalies, eye anomalies and cardiac malformations. Mutations in *FGFR1* have been described to cause a wide phenotypic spectrum such as Hartsfield syndrome, hypogonadotropic hypogonadism with or without anosmia, Jackson-Weiss syndrome, osteoglophonic dysplasia, Pfeiffer syndrome and trigonocephaly type 1. Here we describe a novel synonymous mutation in *FGFR1* identified by exome sequencing in two siblings born to non-consanguineous healthy Swiss parents. The male patient presented with lobular holoprosencephaly with a single maxillary incisor, ectro-/syndactyly on both feet, syndactyly on both hands, craniosynostosis of the sagittal suture, delayed puberty and developmental delay. His younger sister was diagnosed with diabetes insipidus, in addition to syndactyly of the right foot and an aortic isthmus stenosis. A cranial MRI showed agenesis of corpus callosum and colpocephaly. The novel synonymous missense mutation c.1029G>A (p.Ala343Ala) *FGFR1* detected in both affected siblings, was excluded in DNA extracted from leukocytes of the parents and their healthy sister. Therefore, we assume a gonadal mosaicism or somatic mosaicism including germ cells of this mutation. In one of the *FGFR1* isoforms this guanine to adenine substitution is located in a specific splice acceptor region, which we currently are functionally validating *in vitro.*

PF 9

Exome analysis establishes the diagnosis of microvillus inclusion disease (MVID) in a family with cholestatic liver disorder as the predominant clinical feature

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Microvillus inclusion disease (MVID) is a very rare severe congenital enteropathy characterized by intracytoplasmatic microvillus inclusions and brush border atrophy in intestinal epithelial cells. It is an autosomal recessive disorder caused by mutations in the *MYO5B* gene (MIM#606540) encoding the myosin Vb protein. The disorder is characterized by intractable secretory diarrhoea in infancy and manifests either in the first days (early-onset form) or in the first two months (late-onset form) of life. Intestinal failure secondary to diarrhea is frequent and children with MVID are dependent on parenteral nutrition. Long-term outcome is generally poor, due to metabolic decompensation, repeated states of dehydration, infectious and liver complications related to parenteral nutrition. In contrast to children with early-onset MVID, the diarrhea is often less severe in children with the late-onset form of the disease. We report a patient who is now 30 years of age and who presented with unclassified cholestatic liver disorder and recurrent diarrhoea four weeks after birth, requiring parenteral nutrition for several months. The two older brothers of the patient had died of an unclassified liver disorder and intermittent massive diarrhoea at the age of 10 months and 5 years, respectively. The clinical course in the patient reported here stabilized in the second year of life after which he suffered from occasional episodes of diarrhoea only, but presented with severe cholestatic liver disorder as the predominant clinical feature. The putative diagnosis of progressive familial intrahepatic cholestasis (PFIC1/2) was established and the patient underwent cholezysto-jejunostomy at the age of 8 years. However, molecular analysis of the *ATP8B1* and *ABCB11* genes did not reveal any pathogenic mutations, excluding PIFC1/2 as the likely diagnosis in this family. In order to establish the correct diagnosis and deliver appropriate counselling, we performed exome sequencing. This analysis revealed two heterozygous disease- causing mutations in the *MYO5B* gene (c.242A>G, p.His81Arg and c.4798C>T, p.Gln1600*). Analysis of the healthy parents confirmed that they carry one of the mutations, thereby establishing microvillus inclusion disease (MVID) as the diagnosis in the patient. In summary, we present further evidence for the broad clinical variability in MVID and confirm that this disorder can manifest predominantly as cholestatic liver disorder.

PF 10

Identification of novel genes associated with mitochondrial disorders

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Background: Mitochondrial disorders may originate from defects in pyruvate oxidation, β-oxidation or oxidative phosphorylation (OXPHOS) and are due to mutations in the mitochondrial genome (mtDNA) or in one of many nuclear genes (nDNA) controlling the organelle function. The large numbers of potential disease-causing genes, together with the little guidance offered by most clinical phenotypes as to which gene may be causative, are a great challenge for the molecular diagnosis of these disorders.
Methods: We developed a gene panel for mitochondrial disorders relying on in solution capturing coupled to next-generation sequencing. The panel consists of 1476 nuclear-encoded genes implicated in mitochondrial biology, mitochondrial disease, or monogenic disorders with phenotypic overlap.
Results: Here, we present our results of targeted resequencing in a diverse cohort of 13 atients with suspected mitochondrial disorders based on clinical, biochemical, histological and/or molecular findings. With our panel we found mutations in 6 patients in genes, which are known to be disease-causing as well as in 3 candidate genes which are not yet described as disease-associated.
Conclusions: With our panel we identified mutations in ECHS1, which was not yet described as a disease-associated gene. ECHS1 encodes a short-chain enoyl-CoA hydratase involved in mitochondrial β-oxidation and it is active in the valine catabolic pathway. Mutations in this gene lead to an impaired energy supply resulting in multiple organ dysfunction. We also identified a mutation in MRPS14, which encodes a mitochondrial ribosomal protein of the 28S small mitoribosomal subunit, involved in the intramitochondrial protein synthesis, with yet unknown function. Furthermore, in a patient we identified a candidate gene called PLIN5, which is involved in lipid droplet homeostasis with a yet unknown function. Our study indicates that the use of next generation sequencing technology holds great promise as a tool for screening mitochondrial disorders. In addition, the proposed approach has the potential to identify new mutations in candidate genes, to expand the spectrum of causative genes responsible for mitochondrial disorders. The availability of a comprehensive molecular diagnostic tool will increase the capacity for early and rapid identification of mitochondrial disorders.

PF 11

Elevated 17-hydroxyprogesterone in newborn screening and no signs for congenital adrenal hyperplasia: Are HIV drugs to blame?

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Case Report: We report the case of a preterm, normal weight female infant, born at 26 weeks gestation with elevated 17-hydroxyprogesterone (17OHP) at newborn screening. There were no signs of congenital adrenal hyperplasia such as genital virilization; and sodium, potassium and cortisol were normal. The 17OHP was in the normal range for gestational age on the first day of life, but was elevated at 292 nmol/l 14 days later. The urinary steroid profile showed elevated progesterone and androgen metabolites with low-normal cortisol metabolites indicating diminished 21-hydroxylase enzyme activity. During pregnancy the mother was treated with Atripla® (Efavirenz, Tenofovir and Emtricitabin) and viral load was suppressed. Furthermore prophylactic antiretroviral therapy with Retrovir® (Zidovudin) was started on the first day of life to prevent vertical transmission of HIV. After delivery a single dose of hydrocortisone was administered for arterial hypotension.
Background and Hypothesis: Transient neonatal adrenal dysfunction has been described in association with antiretroviral therapy with Lopinavir-Ritonavir, but not with Zidovudin. Other drugs have not been tested. Therefore, we hypothesized that the elevated 17OHP might be due to the HIV treatment even though there have been no reports of adrenal dysfunction associated with the drug Atripla®.
Follow-up: In our patient 17 OHP levels normalized within 4 weeks and after termination of the antiretroviral prophylaxis indicating that the abnormal 17OHP was caused by the drugs.
Conclusion and Perspective: HIV drugs may affect steroid hormone biosynthesis in newborns and lead to abnormal neonatal screening tests. Further tests of HIV drugs are needed to investigate which compounds may cause a relevant (transient) adrenal dysfunction that may even require emergency supplementation of glucocorticoids.

PF 12

Characterization of the backdoor pathway for dihydrotestosterone synthesis in a virilizing childhood adrenocortical carcinoma

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Background and aim: Tumors of the adrenal cortex (ACT) may produce massive amounts of androgens and thereby lead to virilization. In children, this causes precocious pseudopuberty. Recently, we and others have described a novel steroid pathway for the synthesis of dihydrotestosterone (DHT), the most potent, natural androgen. In this alternative, backdoor pathway DHT is produced from 17-hydroxyprogesterone without going through testosterone like in the classic synthesis path. We therefore wondered, whether androgen synthesis in ACTs may involve the backdoor pathway.
Case report: A 2-years-old girl was referred to us for premature pubarche, recurrent facial exanthema and sweating. Clinical examination revealed pubic hair Tanner stage P3, clitoral hypertrophy and facial acne. Breast was Tanner stage B1. Her abdomen was protruding. Blood pressure was abnormal (110/79 mm Hg). Laboratory work-up showed grossly elevated androgens and mild hypercortisolism. By ultrasound a solid mass was found arising from the left adrenal gland; the MRI-scan confirmed an ACT with signs of necrosis. Tumor resection required nephrectomy, but allowed removing an adrenal tumor of 10 x 12 cm within the capsule. Fortunately, no signs of malignancy were found in the surrounding lymph nodes. However, genetic work-up revealed that the patient carried a TP53 tumor suppressor gene mutation compatible with the diagnosis of Li-Fraumeni syndrome, putting her at high risk for developing further tumors.
Materials and methods: The tissue of the ACT was investigated immunohistochemically using newly developed antibodies against proteins, which are characteristic for the steroid backdoor pathway of DHT synthesis. Results were compared to normal human adrenal cortex tissues.
Results: Histopathologic exam of the ACT revealed a carcinoma belonging to the high risk group showing extensive necrosis and invasion of blood vessels into the capsule. Specific immunohistochemical studies showed a strong upregulation of HSD17B6 and AKR1C3; these are enzymes, which convert androsterone to DHT in the last steps of the backdoor pathway. By contrast, enzymes upstream of androsterone (e.g. HSD3B2, SRD5A1 and AKR1C2/C4) were found downregulated in the ACT when compared to normal adrenal tissue.
Conclusion: Androgen overproduction in ACT may involve the steroid backdoor pathway for DHT synthesis, which shows clear alterations in specific protein expression levels when compared to normal human adrenal tissues.

PF 13

Neonatal onset propionic acidemia: a case report

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Proprionic acidemia (PA) is a rare autosomal recessive disorder of organic acid metabolism, caused by a deficiency of proprionyl-coenzyme A carboxylase (PCC). It has varied clinical and metabolic presentations: the usual presentation is life-threatening ketoacidosis and hyperammonemia. While metabolic acidosis is a key feature of early onset PA, there are several reports of patients where metabolic acidosis was not a persisting finding. We reported a case of PA in a 8-days-old female infant, third born of a non-consanguineous parentage. Pregnancy and delivery were uncomplicated (APGAR 9-9-10); she was born term with birth weight of 4350 g (250 gr >97%ile). Postnatal initial course was remarkable for progressive weight loss (725 gr weight loss). At 8th days of age she was referred to our istitution for feeding difficulties, poor sucking, vomiting, hypotonia and hypoactivity. Vital parameter were stable. Neurological exam reveald sonnolence, minimal spontaneus activity. All reflexes were present. She had a minumum decreased of flexor and extensor head tonus as well as of axial tonus. She presented a valid crying under stimulation. The rest of the phisycal examination was normal. Laboratory investigation revealed increasing level of serum ammonia (max 227 umol/L).Urinalysis revealed moderate ketonuria. Venous blood gas analysis and serum lactate concentration were normal at the onset. Urine organic acid study and the analysis of the Guthrie samples established the diagnosis of PA. Metabolic acidosis was not observed during the onset period of the disease. According to the pediatric metabolic specialist, we stopped the milk intake and performed rehydratation and prevention of the catabolic state by provision of adequate calories (by giving glucose 10% iv), while organizing the trasfert of the baby to a tertiary care center. We conclued that PA should be considered in the differential diagnosis of patients with neurologic syntoms and hyperammonemia with or without acidosis. Mild-to-moderate mental retardation is common in patients with PA even with good compliance to therapy. Neurodevelopmental deficits are usually due to reccurrent episodes of hyper ammoniemia and acidosis as well as long-term exposure of brain to abnormal metabolities (ammonia and its toxic compounds). Early diagnosis and treatment, even with peritoneal dyalisis, are important for better long-term prognosis.

PF 14

The erroneous perception of a clinical sign – jaundice in severe bacterial infection mimicking hepatitis in an adolescent patient

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Background: Cholestasis is a frequent complication of sepsis, especially in newborns and infants, but is rarely seen as the first manifestation of bacteremia before other systemic signs are present.
Case report: A previously healthy 14-year-old adolescent presented to our pediatric emergency department (PED) with a 5-day history of left ankle pain and jaundice without fever. Previously performed outpatient X-rays and computed tomography (CT) images of his left ankle joint were reported to be normal. He had raised cholestasis (ALAT 178 U/L, yGT 533 U/L, total bilirubin 103 µmol/L) and inflammatory markers (C-reactive protein (CRP) 195 mg/L), whereas a full blood count and

coagulation tests were within normal limits. With a recent history of camping in Italy, the patient was discharged home with a preliminary diagnosis of viral hepatitis. Five days later, he represented to our PED in distinctly reduced general condition but still apyrexial. Additionally to the jaundice, swelling and erythema of his left ankle, both wrists and extensor sides of his forearms, generalized allodynia and a diffuse palpable punctiform non-blanching purpuric rash were noted. Laboratory testing showed progressive inflammation (CRP 223 g/L, leucocytes 29.7 G/L), thrombocytopenia (thrombocytes 53 G/L) and raised kidney function tests (creatinine 166 umol/L). The patient was admitted to the pediatric intensive care unit with a working diagnosis of *sepsis* versus *systemic vasculitis*. He was started on empiric broad-spectrum intravenous antibiotic therapy. Subsequently, blood cultures became positive for *Staphylococcus aureus*. With a considerable delay of 7 days since first admission to PED, osteomyelitis of the left calcaneus and right femur, myositis and fasciitis of the right upper arm, right thigh and left lower leg, and multiple lung abscesses were documented by total-body magnetic resonance imaging.
Conclusion: This case of prolonged jaundice without fever prior to hospital admission with severe sepsis due to multifocal S. aureus osteomyelitis highlights the importance of considering jaundice as an early sign of bacteraemia in children of *all* ages, even if this association remains rare in adolescents. We assume that correct initial interpretation of cholestatic jaundice as the first sign of bacterial infection could have prevented severe sepsis in our patient. We suggest physicians aim to avoid fixation errors when interpreting frequent and more rare clinical signs and symptoms.

PF 15

Kawasaki – still a challenge!

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Introduction: Kawasaki disease (KD) is an acute self-limited vasculitis of unknown etiology that occurs predominantly in infants and young children. The diagnosis and management of KD is especially challenging in very young infants or if clinical criteria are not fulfilled.
Case I: A 4-years-old boy was admitted from our ID department because of fever for 14 days, lymphadenopathy – for which he was treated inpatient with antibiotics 7 days earlier – and in echocardiography slightly altered coronary arteries with signs of inflammation. We started with IVIG and aspirin, after which he developed a rash, but had no more fever 48 hours later. Other signs of KD were never apparent, and cardiac findings normalized over time.
Case II: A 3-years-old girl presented with a history of fever for 6 days, non- suppurative bilateral conjunctivitis, highly elevated inflammatory markers and back pain. MRI ruled out spondylodiscitis or osteomyelitis, but showed enlarged cervical lymph nodes. Liver enzymes were mildly elevated and in the 2nd week of illness platelets rose. After 8 days fever stopped and the patient slowly recovered. IVIG and aspirin were not given. Exfoliation of the fingertips occurred 2 weeks after the onset of symptoms. Repeated echocardiography was always normal.
Case III: A 3-years-old girl was admitted to our hospital with suspicion of KD. She had a history of 4 days fever, intermittent rash, edema of the lower legs, erythema of the lips and oral mucosa, and bilateral conjunctivitis. Inflammatory markers were elevated and there was a mild hypoalbuminemia. She spontaneously defervesced on day 5 and remained afebrile during inpatient surveillance over several days. Follow-up echocardiography remained normal.
Conclusion: The diagnosis of KD is a challenge and needs expert opinion. Some patients lack sufficient clinical signs to fulfill the classical criteria, so the diagnosis of an incomplete KD should be considered in the differential diagnosis of every child with fever for several days. Therefore laboratory tests (CRP, ESR, PLC, BNP, albumin) are useful but nonspecific. Echocardiography can help confirm the diagnosis, but new data implicate that more sensitive parameters are needed, including functional assessment, because we may underestimate cardiac involvement by classical characteristics.

PF 16

Adverse events and risk factors during emergency intubation in a tertiary paediatric emergency department: a cross-sectional study

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Background: Rapid sequence induction (RSI) and intubation in the emergency department (ED) can be life-saving procedures but require the appropriate skills, experience and preparation to avoid complications ranging from simple trauma to life-threatening desaturation. To date, only scarce data exists in the published literature on complications following RSI and intubation in children, and most guidelines are extrapolated from the adult population.
Methods: We reviewed all emergency intubations of patients in our tertiary PED within a two- year period to estimate the incidence of complications and to analyse risk factors associated with this procedure.
Results: Seventy-two children were intubated within the two-year period; complications occurred in 1 in 4 children (25%). Median age was 2 years (range: 0 days to 16 years). The most common reason for intubation was altered level of consciousness (LOC) and the most common diagnosis at the time of intubation was seizure/status epilepticus. Complications were related to intravenous (IV) access (n = 2) or equipment failure (n = 3), erroneous or insufficient drug preparation (n = 1), desaturation (n = 6) and other reasons, not further specified (n = 6). There was no significant association of complications with the child's age or weight, time of arrival to ED, pre-intubation hypotension or combination of drugs used.
Conclusion: Complications of induction and intubation, both relatively low-frequency procedures in the PED, occurred in 1 of 4 children. We suggest the use of an intubation checklist including the preparation of equipment and recommendations for drug use for frequent diagnoses would minimize the occurrence of adverse events of intubation in children.

PF 17

Neonatal cardiomyopathy – a chameleon challenging to diagnose

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Introduction: Neonatal cardiomyopathy (CM) is a rare but severe disease. Aetiologies and clinical presentation are broad. We present three cases of newborns with different forms of CM, which presented diagnostic and clinical challenges.
Cases: 1. Ventricular disproportion with right ventricular (RV) dilatation was noted during pregnancy and confirmed postnatally. The initially asymptomatic girl developed sinustachycardia and impaired left ventricular (LV) function at the age of 5 weeks when dilated CM was diagnosed. Metabolic disease was ruled out and treatment with ACE inhibitor started. Though LV function normalized, polyvalvular dysplasia with regurgitation of both atrioventricular (av)-valves and mild aortic regurgitation was observed at the age of 4yrs. Currently, at the age of 10yrs, clinical suspicion of connective tissue disorder exists, but the family refuses further genetic testing. 2. A preterm dysmorphic boy presented at day 3 with tachypnea, desaturation and supraventricular extrasystoles. Echo showed regurgitation of both av-valves, enlarged RV and dilated atria. Subsequently, LV function deteriorated and mitral regurgitation (MR) increased. Noonan syndrome with mitral valve dysplasia and LV dysfunction was diagnosed. Under medical support, LV contractility and MR normalized, but restrictive dysfunction persisted. In the following weeks a severe obstructive hypertrophic CM developed. The boy died due to therapy refractory arrhythmias and progressive heart failure. 3. A preterm asymptomatic girl presented after birth with dilated LV, moderate MR and slightly reduced LV function (EF 50%). Dilated cardiomyopathy was diagnosed. Three weeks later RV dilatation with markedly reduced function, severe pulmonary hypertension and tricuspid regurgitation were found. Clinical conditions deteriorated dramatically, and extracorporeal membrane oxygenation (ECMO) was installed. Myocardial biopsie revealed a severe form of biventricular Morbus Uhl. Due to very limited

therapeutic options and disease's severity, compassionate care was decided with the parents and mechanical support was withdrawn.
Conclusion: Newborns with CM may initially present asymptomatic, but can rapidly deteriorate. Initial echo may be unspecific before the full picture is shown with a delay of days to weeks. Thus, close echocardiographic follow-up is essential. The haemodynamic changes during the perinatal period and following weeks can make neonatal CM a diagnostic and clinical chameleon.

PF 18

Supraventricular tachycardia in neonates and infants: initial treatment and follow-up

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Background: Supraventricular tachycardias (SVT) occur with a peak incidence in infancy and resolve often spontaneously after the first year of life. In neonates and infants, recurrent SVT is prevented by administration of antiarrhythmic drugs for 12 months. The current evidence concerning type and exact duration of this prophylaxis is low and often depends on local institutional experience. We reviewed our patients regarding type and safety of this preventive treatment and arrhythmia outcome.
Methods: Retrospective single centre observational analysis: All neonates and infants with SVT, including prenatal diagnosed tachycardia, with anatomically normal heart presenting between 1/2005–7/2015 were included in the study.
Results: From 72 patients included 23 (32%) were prenatally diagnosed and 14 (19%) were born premature. Median age of first SVT episode was 12 (0–254) days. Arrhythmia diagnosed by surface ECG: Atrioventricular reentry tachycardia in 48 (67%), atrial ectopic tachycardia in 17 (24%), atrioventricular nodal reentry tachycardia in 1 (1%) and unclassified (history of insistent intrauterine tachycardia) in 6 (8%) patients. Tachycardia related LV dysfunction occurred in 6 (8%) infants with complete recovery in all. First line medications were beta-receptor blocking agents in 41 (57%), propafenone in 15 (21%) and amiodarone in 14 (19%) patients. Propafenone was discontinued in 7 patients due to proarrhythmic effects (QRS widening in 6, ventricular tachycardia in 1 patient). Median hospitalization following the first episode was 20 (0–166) days. Arrhythmia control was achieved with single antiarrhythmic drug therapy in 53 (74%), 14 (19%) had a twofold and 3 (4%) had a triple or more combination of antiarrhythmic drugs. There was no SVT related mortality. SVT relapsed in 12/72 (17%) patients after withdrawal of the prophylactic treatment; 6/23 (26%) of whom had fetal tachycardias.
Conclusions: Prophylactic antiarrhythmic medication for SVT in infancy is generally safe and well tolerated. In this cohort, proarrhythmic effects were only seen after 1C antiarrhythmic medication. Arrhythmia control is achieved with one medication in majority of patients. After cessation of the prophylactic treatment most patients are free of recurrences. Infants with episodes of fetal tachycardia tend to be more prone to suffer from SVT relapses after cessation of antiarrhythmic medication.

PF 19

The continued issue of pulmonary haemorrhage in ELBW infants

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Background: Pulmonary hemorrhage (PH) is a rare but life-threatening event associated with a high mortality. Numerous findings such as sepsis, coagulopathy, thrombocytopenia, surfactant application, intraventricular haemorrhage (IVH), and patent ductus arteriosus (PDA) are associated with PH. In the prevention of PH, the rationale of transfusing platelets and plasma is controversial. We reassessed risk factor for PH with specific focus on routine coagulation screens, including platelet counts.
Methods: Retrospective case-control study consisting of 20 ELBW infants with PH and 40 age- and weight-matched controls. PH was defined by way of tracheal blood that was found to have decreasing hematocrit levels requiring red blood cell transfusions. Neonatal infection and septicemia was assessed. Echocardiographic was performed for PDA diagnosis. For statistical analysis of platelet counts, the first blood examination at birth, and the most recent examination prior to PH, was considered. These data were compared with platelet counts from the second routine blood examination performed within 24–96 h postnatally in case-controls. We assessed thrombocytopenia (platelet count below 150/nL), anti-thrombin-III, fibrinogen, and activated partial thromboplastin time.
Results: All cases of PH occurred within 96 h after birth. Mortality until day 7 was higher after PH compared to controls ($p = 0.03$). Rates of antenatal steroids were lower in infants with PH ($p = 0.007$) compared to case-controls, while rates of surfactant administration, indomethacin, and ibuprofen did not differ. Platelet counts prior to PH were significantly lower than the lowest platelet counts during the first 96 h in controls ($p = 0.002$). Despite significant differences in the incidence of severe thrombocytopenia (below 50/nl) or abnormal coagulation screens, significantly more patients with PH received platelet transfusions and/or fresh-frozen plasma prior to later hemorrhage.
Conclusion: Antenatal steroids appear helpful in preventing PH, and ELBW infants born without prior antenatal steroids should be considered candidates for prophylactic indomethacin. Given that our study identifies transfusion of adult platelets and plasma to be associated with later PH, clinical trials on laboratory based values as transfusion criteria would be justified. In vivo experiments are needed to understand the suggested adverse effects on primary haemostasis when transfusing adult platelet and plasma to ELBW infants.

PF 20

Neonatal Purpura fulminans

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We present a case of neonatal purpura fulminans in a 3rd child of a healthy family. Intrauterine MRI at 28 gestational weeks revealed a hydrocephalus due to bilateral grade IV hemorrhage dated 2–7 days before. No underlying family history for bleeding disorders, medication, infection or trauma could be identified. The baby boy was born by caesarian section at term to avoid perinatal complications and showed normal primary adaption. In the first hours of life, gluteal and lower extremity hematomas were noted and evolved rapidly into purpura fulminans. In absence of any evidence for infectious trigger the clinical presentation led to severe congenital Protein C (PC) deficiency. Genetic testing of the family revealed a compound heterozygous genetic disorder. Immediate treatment with fresh frozen plasma (15 ml/kg/6h) iv was initiated and ceased progress of cutaneous lesions. As soon as available, plasma-derived PC-concentrate was initiated at a dose of 100 U/kg stat followed by 60 U/kg/ 6h. PC substitution was titrated under plasmatic level control to doses of 70 Units PC/kg/6h iv, which resulted in good clinical response. Timely application of treatment seemed crucial, as treatment delay was associated with clinical deterioration, respiratory insufficiency, intestinal problems and clinical hypercoagulability. Most probably these symptoms were due to microthrombus formation with low trough levels (<30% PC in aPTT method). Therapeutic enoxaparin was started for central venous line associated thrombosis in both jugular veins. Further investigation showed bilateral vitreous and retinal hemorrhage leading to blindness. The hydrocephalus remained stable without evidence of further intracerebral thrombosis or bleeding. Secondary wound healing of purpura fulminans lesions was excellent after subcutaneous surgical debridement. The child was discharged at the age of 2 month and is currently under subcutaneous PC infusion of 200 Units/kg over 12 hours every day and enoxaparin 2 mg/kg/24 h with no further evidence of thrombosis.
Conclusion: The triad of prenatal intraventricular hemorrhage in the 3rd trimenon og gestation, bilateral retinal hemorrhage and purpura fulminans in the first hours of life is a typical presentation of severe (homocygous or compound heterocygous) PC deficiency. Immediate diagnose and timely initiation of treatment is crucial to stop life-threatening disseminated intravascular coagulation (DIC).

PF 21

A dangerous but rare airway obstruction: Epiglottic cyst

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Introduction: Laryngeal cysts are a rare entity and constitute only 4.3% to 6% of all benign laryngeal tumors. However, epiglottic cysts are one of the top five laryngeal diseases in Korea. It was first described in 1864. Epiglottic cysts seem to be more prevalent in the 6th decade of life but they can affect and have been reported in all ages. Due to a smaller diameter of the respiratory tract in infants and children, an epiglottic cyst may easily obstruct the airway in this age group. Large cysts may present as stridor, cyanosis while feeding, respiratory difficulties, and may potentially be life-threatening. A positive association between an epiglottic cyst and smoking as well as allergic rhinitis has been reported.
Case report: A 30 day-old boy, was admitted to our hospital due to an insufficient weight gain during the preceding two weeks. The previous couple of days he often fell asleep during breast-feeding and was found to have jaundice. Additionally, the mother reported two very unsettled episodes with crying where he obviously had difficulties breathing and showed a cyanosis around the mouth for a short time. In emergency the physical examination revealed no obvious abnormality except a weight below the 3rd percentile and an obvious scleral and skin icterus. Laboratory findings showed only an elevated indirect bilirubin. During the admission when the boy became upset the saturation suddenly dropped to a minimum of 48% with obvious cyanosis and he showed remarkable subcostal and sternal retractions while he had a silent chest on auscultation. The most impressive finding, however, was a panicky facial expression. Under oxygen supply the boy recovered over several minutes. The thoracic radiograph showed unspecific bilateral reticular interstitial infiltrate without consolidation. As an intermittent upper respiratory tract obstruction was obvious, with indication for further investigations with endoscopy, the patient was transferred to the University Children's Hospital Zuerich. The endoscopy by the ENT specialists two days later revealed an epiglottic cyst. It was easily removed with a CO_2 laser. The boy recovered quickly and was discharged on day 2 after the intervention.
Conclusion: Laryngeal cysts are relatively rare benign lesions of the larynx. Congenital cysts can cause neonatal respiratory distress and death. Therefore removal is advised. In ENT surgery the most broadly used laser is the carbon dioxide laser.

PF 22

The importance of the assessment of the red reflex (Bruckner testing) in the newborn

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Introduction: Cataract is one of the main causes of blindness in children. The prevalence of childhood cataract is 1 to 15 per 10'000 children. The etiology can be classified in three types: congenital, developmental and traumatic. Hereditary cataracts are most frequent in an autosomal-dominant way, rarely there exist autosomal recessive and X-linked forms. In addition the cataract could be disease-associated, such as in metabolic disease, chromosomal anomalies and craniofacial disorders. In Africa the deficiency of vitamin A and the measles are the two main reasons of cataract in children. Surgery is the only known treatment for an optimal visual development and reduces the risk of amblyopia. After surgery the child needs a close follow up for early detection of refraction problems and strabismus, which is a serious and rather frequent problem.
Case report: In our primary care pediatricians office a 4-week old boy was brought in by his parents for the one month healthy child visit. He's a full term newborn, born after a uneventful pregnancy and a spontaneous uncomplicated delivery at a Swiss hospital. It is the third child of an immigrated family from Afghanistan. When he was two weeks old he was hospitalized for the reason of a fever of unknown origin, probably a viral infection. At the 1month check the development was age-based, but the red reflex of the eyes was bilateral inexistent. The immediate examination by the ophthalmologist revealed a bilateral congenital cataract. The indicated surgery entails removal of the opaque lens and was organized as an emergency intervention. The family had to be convinced that the surgery is very important for the child's eyesight. As we could understand there are several uncles and aunts also having problems with the eyes back in Afghanistan. Later also his 4 year old sister was operated for her incomplete congenital cataract.
Conclusion: The screening procedure in newborn examination is important to prevent a later amblyopia. Congenital cataract is an important disease, which can when detected and treated as soon as possible prevent later blindness or severe visual handicap. After operation it needs a intensive visual rehabilitation. In the case report, the cataract is most likely inherited in an autosomal dominant fashion, based on a bilateral appearance.

PF 23

Superior mesenteric artery syndrom after corrective spinal surgery for scoliosis

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Introduction: The superior mesenteric artery syndrom is a rare cause of proximal intestinal obstruction. Due to a narrow space between the superior mesenteric artery and aorta the distal duodenum suffers a compression. Mostly it is caused by a reduction of the mesenteric fat pad due to massive weight loss. In young persons it is also described as a complication after a spinal surgery for scoliosis. Lenghtening the spine decreases the aorto-mesenteric space, especially in patients with an asthenic habitus and therefore a preoperative smaller mesenteric fat pad.
Case report: A 14-year-old girl was admitted to our hospital complaining about vomiting and abdominal pain for two days without a fever. Her personal history revealed a dorsal spondylodesis because of a thoracic scoliosis 11 days ago. The physical examination showed an abdominal distention with a diffuse tenderness of the abdomen. Plain abdominal radiography revealed air-fluid-levels in the stomach and the duodenum. Abdominal ultrasound showed a distended stomach and part I and II of the duodenum without an obvious reason. The girl was admitted and after a day of improving symptoms she developed again increasing pain and restarted vomiting. A computer tomography was obtained which revealed multiple signs for a superior mesenteric artery syndrome with a compressed third part of the duodenum, an aorto-mesenteric angle of 18° and an aorto-mesenteric distance of 3 mm. The patient was transferred to the department of paediatric surgery at the University children's hospital of Zurich. A conservative therapy was initiated. For jejunal nutrition and decompression of the duodenum a duodenal tube was inserted in an endoscopic exploration. Few days later regular oral feed could be reintroduced.
Conclusion: – In patients with a history of scoliosis surgery in the last few weeks presenting with abdominal pain and vomiting a superior mesenteric artery syndrome should be suspected. – Plain abdominal films, a radiograph with oral contrasts or a computer tomography may help to make the diagnosis. A duodenal obstruction in the third part, an aorto-mesenteric angle ≤25° and an aorto-mesenteric distance ≤8 mm are the radiological criteria. – Usually conservative therapy is successful. With a jejunal feeding tube the stomach and the duodenum can be decompressed. In cases with failure of the conservative measures surgery may be needed.

PF 24

A neonatal presentation of breath holding spell

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We report the case of a newborn presenting with breath holding spells (BHS). A ten days old boy, born at term, after an uneventful pregnancy, was hospitalized for investigation of a stridor with inconstant desaturations and cyanosis during cries. These episodes already appeared the second day of life and were associated with a hoarse voice and were not concomitant with breastfeeding. Different investigations were performed, such as chest X-ray and laboratory test (haemoglobin, electrolytes and ferric balance) were all normal. Blood gases showed a hypercapnia maximal at 6.7 kPa. Cardiac ultrasound concluded to patent foramen ovale without any other anomaly.
A cerebral origin was excluded by normal magnetic resonance imaging and no relevant argument for epileptic disorder was retained. A nasofibroscopy showed an absence of laryngomalacia and normal vocal cords' movements. Indirect sign of reflux with arytenoid inflammation was noticed. However, under endoscopic visualization, breath- holding respiration occurred during cry with appearance of desaturation and cyanosis. Familial history revealed that the father presented the same pattern of BHS when he was 3 month-old. After

exclusion of all severe aetiologies and with the familial history, the diagnosis of BHS was retained. This child was discharged from hospital after 10 days with anti-reflux treatment. The evolution was good with complete disappearance of stridor and cyanotic episode at 2 months of life. Capnography realized during the control after 2 month was alsonormal. A review of the literature revealed a very small number of BHS case in the neonatal period. Indeed, BHS, the most common form of non-epileptic paroxysmal event, is described in children between 6 months to 6 years of life. Nowadays, the pathophysiology of BHS is not fully understood. Iron-deficiency anemia (IDA) has been outlined as possible contributing factor and was not found in our case. In conclusion, BHS is a current paediatric diagnosis, but very rare in neonatal period. No treatment has been shown to improve significantly this pathology. BHS diagnosis could be retained only after exclusion of otheretiology.

PF 25

Esophageal web as a rare cause of dysphagia in a 7-year-old girl

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Introduction: Feeding and swallowing disorders (also known as *dysphagia*) include difficulties in any step of the feeding process. Dysphagia to solid foods is a rare condition in children and a reason for further evaluation. Common causes are developmental disability, neurological disorders, behavioral factors and structural abnormalities. Signs and symptoms of swallowing and feeding disorders vary, based on the age of the child. Congenital disorders usually become apparent with the introduction of solid foods during infancy or early childhood.
Case report: A 7-year-old girl was referred to our hospital with a history of dysphagia and refusal of solid foods since early childhood. The patient was nourished with liquid and soft food only. She did not complain about odynophagia. There was no history of prematurity, gastro-esophageal reflux or neurological abnormalities. Physical examination was normal, weight 28 kg (P 75–90) and height 128 cm (P 50–75) except a remarkable overbite and jaw deformation due to the lack of chewing over years. Lab work-up revealed an iron deficiency anemia. A barium swallow revealed a narrowing in the proximal esophagus. Upper endoscopy showed a thin, web-like diaphragm at 5–6 cm with a small central opening of a few millimeters. The endoscope with an external diameter of 5.6 mm could not be passed beyond this web. Endoscopic laser division of the web was performed, using a ND-YAG laser (bare fiber 0.6 mm). Afterwards the web was dilated up to 9 mm. A follow-up endoscopy 2 weeks later showed improvement of the narrowing; no scar tissue at the laser division site was seen. Since laser incision and dilatation of the stenosis, the girl was able to eat solid food and enjoys the new acquired diversity of the nutrition.
Conclusion: Esophageal web is a rare cause of dysphagia. Clinical symptoms include disturbed eating habits (refusal of solid food), dysphagia and food impaction. Differential diagnosis of dysphagia includes eosinophilic esophagitis, gastro-esophageal reflux and functional disorders. A barium esophagram and an upper endoscopy are mandatory in the work-up of persisting dysphagia in children.

PF 26

Acute renal failure in severe hypercalcemia

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Introduction: The most common causes of hypercalcaemia in the pediatric population are vitamin D intoxication, excessive calcium intake, hyperparathyroidism of different origin and paraneoplastic. Main features of severe hypercalcaemia are gastrointestinal, neurological and psychiatric complaints, but also renal symptoms with polyuria, nephrolithiasis and renal failure can occur. We report the case of a 8 years old boy with acute renal failure due to severe hypercalcaemia in the setting of vitamin D and calcium supplementation for primary hypoparathyroidism.
Case report: A 8 years old boy with congenital hypoparathyroidism was referred to our outpatient clinic because of acute renal failure. He was known for hypoplastic right kidney with compensatory hypertrophic left kidney and normal renal function (creatinine 47 µmol/l 5 month before). The congenital hypoparathyroidism was treated with vitamin D and calcium supplements. Because of persistent hypocalcaemia and low vitamin D levels both supplements needed to be increased consecutively over the last three month. He presented in reduced general condition with a history of nausea, vomiting and weight-loss over several weeks. The parents also noticed polyuria with secondary nocturnal enuresis. Laboratory evaluation revealed a creatinine of 140 µmol/l, severe hypercalcaemia (total calcium of 3.45 mmol/l and ionized calcium of 1.69 mmol/l) as well as hypercalciuria (1.8 mol/mol creatinine). Blood pressure values were measured >P95. Vitamin D was reduced and calcium supplementation discontinued. Calcium values slowly decreased and with a delay of approximately 4 weeks renal function started to recover. Within some weeks general conditions improved, nausea and vomiting disappeared and he started to gain weight again. Calcium supplementation was resumed cautiously because of values below the normal limit and blood levels remained stable within the low normal range. Renal function continued to recover to a creatinine level of 56 µmol/l, blood pressure normalized as well and hypercalciuria disappeared.
Conclusion: In the paediatric population severe hypercalcemia is a rare, but important reason for acute renal failure. The pathophysiologic mechanisms are not fully understood, but renal failure is potentially reversible if hypercalcaemia is promptly treated. This case highlights the need of careful monitoring in case of calcium or vitamin D supplementation.

PF 27

Once seen, never forgotten: severe spinal cord compression due to atlantoaxial instability in a girl with Down syndrome

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Background/Purpose: Down syndrome is the commonest recognized chromosomal abnormality in man, the incidence today being 1:700 live births. Importantly, atlantoaxial instability (AAI) due to syndrome-typical ligamentous laxity can lead to spinal cord compression with devastating consequences if nottreated.
Case description: A 13 year old girl with Down syndrome was referred to our neuropaediatric department from a paediatric rehabilitation centre, where she had been admitted for locomotion training by her therapist. History revealed a deterioration of gait with kneeing- and toeing-in and more falls since 1–2 years. No bladder or gastrointestinal dysfunctions were reported. Reviewing her chart showed that the girl had been radiologically diagnosed with atlantoaxial instability already at the age of 7 month. The clinical examination showed spastic signs of both legs with cloni, hyperreflexia and shortening of achilles tendons as well as elevated muscle tone with hyperreflexia of both arms. MRI of the head and spine showed no signs for Moyamoya syndrome, nor other vascular or parenchymal cerebral pathology. At C1-C2-level, there was a significant spinal cord compression up to minimally 2–3 millimeters due to atlantodental instability with posterior subluxation of the dens axis. The girl was referred to the orthopaedic surgeon and a surgical stabilization within the next months is planned.
Conclusions/discussion: AAI occurs in about 10–30% of patients with Down syndrome, depending on the criteria used to define it. Symptoms of spinal cord compression can develop in up to 15% of those affected with AAI, which equates to 2–4% of all children with Down syndrome. Symptoms may present at any age and need urgent medical evaluation. They may include progressive spasticity as in our patient, mechanical neck or occipital pain in extended neck position, torticollis, and recent onset incontinence. In case of symptoms, there should be a low threshold to undertake MRI. Radiological screening for asymptomatic AAI is not recommended as the predictive value is poor, the measurement of the subluxation can be difficult and the long term natural history is very variable. The definitive management in symptomatic patients is surgical.

PF 28

Abdominal pain and dyspnoea – not necessarily lower lobe pneumonia

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Introduction: Abdominal pain, respiratory symptoms and infiltrate on chest x-ray are suggestive for lower lobe pneumonia. We present a child with a very unusual cause of pleuralpneumonia.

Case report: A 4-year-old boy presented at our emergency department with a two-week-history of abdominal pain and dyspnoea without fever. Chest x-ray (CXR) showed a postero-basal infiltrate and pleural effusion on the left side. Blood tests revealed mildly elevated inflammatory markers (CRP 13 mg/l, ESR 28 mm/h) and thrombocytosis (669 G/l). Suspecting pneumonia, the child was treated with amoxicillin, but showed no change of clinical symptoms, inflammatory markers or radiological findings. Suspecting atypical pneumonia, clarithromycine was added without any improvement. CT-scan revealed huge pleural effusion on the left side and multiple ubiquitous pulmonary nodules and bronchoscopy showed no mucosa inflammation. Puncture of the pleural effusion revealed atypical cells. Video-assisted thoracoscopic surgery (VATS) demonstrated multiple lesions of the lung and a biopsy of the lung was taken. Pathological work-up of the biopsy revealed a very high suspicion of epitheloid haemangioendothelioma with immunohistochemical detection of CAMTA1, highly specific for this tumor. The final confirmation of the reference pathology center in Kiel is still pending. PET-CT showed no further metastases.
Discussion: In patients with persistent pneumonia-like symptoms not responding to adequate antibiotic treatment, rare lung diseases should be evaluated. Epitheloid haemangioendothelioma is a very rare tumor in humans, especially in children; so far, the youngest described case in the WHO data base is 7 years old. As experience with this tumor in children is very limited, an experimental therapy with the tyrosine-kinase inhibitor pazopanib is considered as surgical intervention in both lungs is not possible. Prognosis of this tumor is variable.

PF 29

Neonatal stress during ambulance transportation: a feasibility study

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Background: Discomfort and stress during air or ground transportation have been described in adults. Each year, the Clinic of Neonatology of the Lausanne University Hospital (CHUV) carries out around 500 transports, and half of them are ambulance transfers of clinically stable neonates. During ambulance transports, environmental stimuli can cause stress in neonates and impact on their health, but it has not been described in the literature.
Aim: The aim of this study was to determine the feasibility of stress measurement in neonates and describe stress of neonates during transport by ambulance from the Clinic of Neonatology of the CHUV (level III) to a secondary referral hospital (level II).
Method: This feasibility study, including a convenience sample of neonates admitted to the Clinic of Neonatology of the CHUV, was conducted between September 2015 and January 2016. Following ethics approval and parental consent, neonates were included in the study, if they were transported by ambulance from the level III to a level II hospital. Exclusion criteria were neonates >44 weeks of corrected gestational age and patients with massive brain damage. Discomfort and stress were measured at baseline, every 15 minutes during transport, and at arrival in the cot. Saliva was collected using the SalivaBio Infant's Swab to extract cortisol at baseline and at arrival. Heart rate, respiratory rate, and oxygen saturation were continuously measured. Behaviour was observed with the Comfort Behavior and the Premature Infant Pain Profile-Revised scales at baseline, every 15 minutes, and at arrival. Descriptive and inferential data analyses will be performed using STATA, in which each participant is its own control.
Preliminary Results: Out of 96 screened, 20 neonates were included in the study. Patients characteristics were boys (40%) and girls (60%). The gestation groups were over 40 weeks (15%), 36–39 +6 weeks (35%), 32–35 + 6 weeks (45%), 28–31 + 6 weeks (5%). Sixteen neonates (80%) had no respiratory support, two (10%) had nasal continuous positive airway pressure, and two (10%) had oxygen therapy (High Flow Nasal) during transport.
Conclusion: The aim of this feasibility study was to test whether stress can be measured in neonates during transports. If the results for stable neonates were to indicate stress during transportation, effect of comfort interventions to reduce neonatal stress during ambulance transportation could be tested in a subsequent study.

PF 30

Story of a very small young man or old boy – unexpected diagnosis in times of migration

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Case report: We report of an 18.5 years old patient, refugee from Eritrea, who was admitted to our outpatient clinic because of very short stature and lacking signs of puberty. He's been living in Switzerland since 5 months, whereto he came only with his mother. He had always been smaller than his peers, but has never been seriously ill so far. Birth and birth weight has been reported as normal. Mother's height 150 cm, father's height approx. 168 cm, MPH 165.5 cm.
In clinical examination, we saw a well orientated young boy of prepubertal impression, height 123 cm (<< P3, –7.3 SDS), weight 21.7 kg (<P3, –5.5SDS) compared to 18y on WHO charts, HC 52 cm (<P3, -3.2 SDS), no dysmorphic signs, slight puffyness of the skin , Tanner stage P1, G1, testicles 2 ml descended, clear voice. Laboratory findings showed low values for TSH and fT4, low IGF-BP3 and IGF-1, low gonadotropins and testosterone values, no signs of celiac disease. In growth hormone stimulation testing there was no increase of HGH levels over the testing time, but persistent low cortisol seen. The radiological bone age was retarded at 9–10 years and in following MRI the suspicion of a malformation of the pituitary gland with aplasia of the anterior part, ectopy of the posterior part and aplasia of the pituitary stalk was confirmed. The "young man" or "old boy" was suffering from a severe panhypopituitarism due to congenital malformation of the pituitary gland, involving the growth, the thyroid, the corticoid and the pubertal axis, but no signs of diabetes insipidus were found. The questions of hormonal replacement therapies (cortisol, hGH, L-thyroxine, testosterone) concerning timing, combination and start of puberty induction are discussed on the poster, all in order to achieve the best growth and development result for his patient. On the other hand we tried to evaluate the cognitive and neuropsychological level of this adult young man living in the body of a 10 year old boy. Questions of schooling possibilities and daily or legal problems of this formally adult but very small and young patient are discussed.
Conclusion: In times of migration we should be aware to see more rare diseases and experiments of nature, like in this young man or old boy of 18 years, who as refugee was surviving without evident crisis a potentially lethal combined pituitary hormone deficiency. The combined and stepwise hormonal substitution of all axes in an adult but immature patient has to be considered carefully.

P 1

Prolonged complete av-block: a rare complication after balloon aortic valvuloplasty

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Background: Balloon valvuloplasty (BAV) is a standard procedure for aortic stenosis in all paediatric age groups. Serious complications are rare and mostly occur in neonates and young infants, whereas in older children the procedure is safe. We present two cases of prolonged complete atrioventricular (av) block after standard BAV.
Case descriptions: Patient 1 had VSD patch closure and resection of a subaortic membrane at age 6y, meanwhile progressive aortic valvular stenosis developed and at the age of 16y he underwent an attempt of BAV, peak gradient at valvular level was 124 mm Hg. A 16 mm Tyshak balloon was chosen for a 17 mm anulus. After the first dilatation done under rapid LV pacing (220 bpm) he had 2:1 and complete av block with bradycardia of 40–50 bpm, which persisted until 6 hours after the procedure, when normofrequent sinus rhythm recovered. No further episodes of block occurred (follow-up 10 months). Patient 2 was a boy with slowly progressive aortic valvular stenosis but no other cardiological antecedants. He underwent an attempt of BAV at the age of 11y, peak valvular gradient was 46 mm Hg and a 16 mm Tyshak ballon was chosen for a 16.7 mm anulus. Immediately after a successful dilatation (LV rapid pacing at 220 bpm) he was in complete av block with a wide QRS escape rhythm of 70 bpm which was poorly tolerated due to new moderate aortic regurgitation, he had to be treated with transjugular implantation of a temporary pacing catheter. Av block resolved after 72 hours and the pacing catheter was removed after stable sinus rhythm was seen for 24 hours. The patient was in sinus rhythm ever since (follow up 8 months).
Conclusion: To our knowledge prolonged av block after paediatric BAV has not been reported. Whereas in the patient with earlier resection of a subaortic membrane it might be speculated that conduction system was damaged during operation, it remains completely unclear why in the other patient with an untouched heart prolonged block occurred. This report might alert the interventional cardiologists about the rare but possible occurrence of such a complication.

P 2

Which approach should be taken in treating glyocosuria in children?

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Introduction: There are some affordable analyses to perform when treating glycosuria in a child. The first pathology to rule out is diabetes with blood sugar and osmolarity. The next step is to focus on kidney diseases.
Case report: A 21-month girl, was hospitalized for an acute gastroenteritis with moderate signs of dehydratation without any other clinical abnormalities on physical examination. She had repeated glycosuria. We found a metabolic acidosis with a normal anion gap, without any other electrolyte disturbances, hyperosmolarity or kidney failure. The urinary analyses showed proteinuria and glycinuria. Parathormone was normal with a slight elevation of vitamin D. An abdominal US ruled out any anatomical malformation. Wilson disease was excluded by normal ceruloplasmin and cystinosis excluded by normal leucocyte cystine content. She never had aminoglycoside or antiviral treatment, excluding a drug-related cause. A normal ophtalmologic control ruled out Lowe syndrome and cystinosis. In the absence of liver failure, hepatomegaly and repeated vomiting, we were able to rule out galactosaemia, tyrosinaemia and congenital fructose intolerance. She didn't have all the typical laboratory features for a Fanconi syndrome but this is still the most probable diagnosis. No exact cause for these abnormal findings has been found to date. Further analyses are still pending for mitochochondrial cytopathies, suspected by elevated lactates on blood gases. She was treated with oral bicarbonate, starting with 4 meq/kg, which normalized the blood acidosis.
Discussion: Further laboratory tests are only required when repeated normoglycemic glycosuria is obtained. Fanconi syndrome is a spectrum of pathologies which affect the proximal tubule transporters, where the sugar is reabsorbed. The laboratory findings are those of the nutrients which are no longer reabsorbed: glycosuria, aminoaciduria, phosphaturia, hyperuricosuria, metabolic acidosis and low-weight proteinuria (vitamin D, β-2 microglobulin) without hematuria. The main causes of Fanconi syndrome are: exposition to heavy metal, medication (tenofovir, aminoglycoside), hereditary diseases (cystinose, fructose intolerance, galactosaemia, mitochondrial cytopathies, Lowe syndrome). Symptoms include polyuria, polydypsia, dehydratation, rickets and growth retardation. Treatment is the replacement of all lost electrolytes or proteins and the prevention of osteomalacia.

P 3

Severe lower-extremity bowing: a clinical sign suggestive of X-linked hypophosphatemia?

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Background: X-linked hypophosphosphatemia is a prototypic disorder of renal phosphate wasting and the most common form of heritable rickets. Nevertheless, rachitic skeletal changes of nutritional and hereditary forms of rickets are indistinguishable.
Case report: Two 21-month-old toddlers were addressed to the orthopaedic clinic because of lower-extremity bowing, evident since the onset of weight bearing. Both toddlers showed radiologic signs of rickets, hypophosphatemia, a reduced tubular reabsorption of phosphate and normal 25-hydroxyvitamin D (25OHvitD) serum concentration. The family history showed, in one case (male), a mother who suffered from spontaneous dental abscesses and bone cysts. His family was known for short stature. In the other case (female) the father was adopted and his family history was unknown. Hypophosphataemic rickets was suspected and both patients were treated with oral phosphorus supplementation and active vitamin D analogs.
Both patients had genetic testing which revealed X-linked hypophosphataemic rickets (XLH) due to loss-of- function mutations in PHEX, an endopeptidase encoded by a gene localized on the X chromosome.
Discussion: Hypophosphataemic rickets is a group of hereditary and acquired conditions characterized by renal phosphate wasting, leading to hypophosphataemia and associated with inappropriately normal to low 25OHvitD serum concentration, causing osteomalacia and/or rickets. Different types of rickets can be distinguished by biochemical testing: in hypophosphataemic rickets, serum concentrations of 25OHvitD and calcium are normal, whereas in vitamin D-deficient rickets 25OHvitD serum concentration is low and the calcium concentration may be low or normal. Patients present with lower limbs deformities (bowlegs or knock knees), becoming evident at the beginning of walking. Dental alterations (poor dental development and teeth abscesses), short stature, bone pain and enthesopathy are other possible clinical manifestations of these disorders. Mode of inheritance and molecular genetic testing help distinguish different forms of hereditary hypophosphataemic rickets. Most children with XLH are treated until growth is complete.
Conclusion: Treating rickets will promote progressive correction of leg deformities and facilitate teeth mineralization. Earlier treatment appears to lead to better results. Variable spectrum of the disease and the suboptimal therapeutic approach makes the treatment of XLH patients challenging.

P 4

Management of ovarian lesions with vaginal bleeding in childhood

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Introduction: Large ovarian cysts >2 cm are rare in prepubertal girls after the first year of life and need further evaluation to exclude torsions and neoplasms. Estrogens are produced in 5% and 90% of the cysts sized >5 cm show spontaneous regression. Depending on the hospital, either pediatric surgeons, gynaecologists, endocrinologists or oncologists are in charge of diagnostics and treatment, but official guidelines are lacking.
Case report: A 2.5 year old girl presented with fever, abdominal pain, self-limited vaginal bleeding and premature thelarche. Ultrasound showed a cystic formation of about 10 ml in the left ovary with displacement of the ovarian stroma backwards, but no signs of ovarian torsion. Uterus was enlarged with endometrial proliferation. Besides

estradiol elevated to 114 pmol/L, tumour markers were negative, namely inhibin B. Because an ovarian tumour could not be excluded by MRI, two international specialists were involved: The Comprehensive Ovarian Cancer Center took the view of an ovarian tumour and recommended ovarectomy, while the radiologist specialized in paediatric gonadal tumours favoured to watch and wait and to perform cyst- or ovarectomy only after an increase in the cyst's size. In a closely monitored outpatient setting, normalization of estradiol and complete regression of pubertal signs and an infantile uterine and ovarian size cyst in ultrasound were found within 3 weeks. After 10 weeks, there was no more evidence of the cyst in ultrasound.
Conclusions: In this case, paediatric surgeons were concerned to miss an ovarian torsion or tumour, while paediatric endocrionologists and gynecologists were afraid of ovarectomy. As a multidisciplinary management of ovarian lesions in childhood is advocated, we designed such recommendations after review of the literature. We suggest further evaluation in cysts >2 cm together with paediatric endocrinologists, surgeons, radiologists, oncologists and gynecologists. Tumour markers and pelvic MRI should be done. Surgery is necessary if ovarian tumour or torsion is suspected, but abdominal puncture of cysts is obsolete. The least invasive techniques with preservation of the ovary are favoured because even small residues of the ovary can be functional and fertile later in life.

P 5

Fragility fractures in children: vitamin-D deficiency, malabsorption or more...?

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Introduction: Fragility fractures result from minimal trauma. A significant fracture history points to osteoporosis if including 1 or more vertebral fractures or at least 2 long bone fractures <age 10 or at least 3 long bone fractures <age 19, plus decreased bone mineral density, e.g. in dual-energy X-ray absorptiometry (DEXA). Primary bone disorders are rare; the most common cause is osteogenesis imperfecta (OI), due to defects in the quality or quantity of collagen I. Any child with osteoporosis plus blue sclerae, dental involvement or hearing problems should be screened for OI. Complications include visual disorders up to retinal detachment, hearing loss and cardiovascular problems like aortic root dilatation. Secondary causes of fragility fractures include Vitamin D deficiency, immobilisation, glucocorticoid excess, celiac disease, bone marrow diseases, rheumatologic diseases and endocrine disorders. Physical abuse should always be considered in children with repeated unexplained fractures.
Case report: An 11 years old boy was presented for back pain since 1 month without a history of recent trauma and after prescription of analgesics, physiotherapy and benzodiazepins without any effect. In his personal history he had 3 minimal trauma fractures. Family history war non-contributory. In the physical exam, normal height in the target height range, obesity and joint laxity were found. The spine X-rays showed vertebral fractures of T1&L1, fish vertebra T9-11 and osteopenic bones. A low spine z-score of –3.0 in DEXA confirmed the diagnosis of osteoporosis.
Results: Extended investigations revealed no evidence of Vitamin-D deficiency, metabolic bone or malabsorptive diseases, endocrinopathies or hematologic disorders. Because of pain, vertebral fractures and osteoporosis the patient was wearing a vertebral orthesis and started on physical therapy, weight-bearing exercise, vitamin D prophylaxis and bisphosphonate (neridronate) infusions every 3 months. Over the course of 12 months, he experienced a decrease in back pain, no more fractures and his height was stabilised. Despite lacking typical signs, biochemical analysis on skin fibroblasts revealed alterations in the production of type I collagen confirming the diagnosis of OI. No cardiovascular, auditory or visual complications were found.
Conclusion: Even when typical symptoms are missing, diagnosis of OI is important, because lifetime management of bone fragility and of additional disorders is needed.

P 6

Immunomodulation in Pompe disease: an effective strategy to resume enzyme replacement therapy after life-threatening adverse reactions

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Pompe Disease (PD) is an inborn error of metabolism characterized by lysosomal storage of glycogen, due to deficient acid a-glucosidase. Classical PD presents in infancy with severe cardiomyopathy. Late onset PD is characterized by progressive skeletal myopathy in childhood or at adult age. Non classical infantile PD presents in infancy with rapidly progressive skeletal involvement and moderate heart enlargment. Enzyme replacement therapy (ERT) with alglucosidase alfa has become standard therapy for all forms of PD. Clinical efficacy is variable: whereas lysosomal storage material is effectively removed from cardiomyocytes and correlates with reversal of cardiomyopathy, damage to skeletal muscle is permanent. Most patients develop antibodies against the infused enzyme and infusion related adverse reactions (IAR) are common. Very high antibody titers limit the clinical efficacy of ERT. Desensitization protocols have succeeded in controlling IAR. Immunomodulation is recommended in ERT-naive classical PD and has been used in rare patients with high sustained antibody titers. Immunomodulation to control IAR has not been described. A 6 yo tracheotomized and fully ventilated girl with non classical infantile PD had been treated by ERT with alglucosidase alfa since diagnosis at age 2 11/12. Three months after ERT initiation, she developed IAR (anxiety, facial flushing, and decreased O_2 saturation). IAR was controlled during 18 mo by slowing infusion rate and premedication with antihistamines. Anti alglucosidase IgG were 1/25'600, while specific IgE, tryptase and complement were normal. IAR then worsened over 3 mo until she presented a severe bronchospasm requiring manual ventilation with a rebound effect several hours after the acute episode. The subsequent infusion had to be stopped at 0.1 mg/kg/h. While ERT was on hold, the patient continued to decline. Following expert advice, we applied an immunomodulation protocol with Bortezomib, Rituximab, Methotrexate and immunoglobulins (Banugaria et al. 2013). We resumed ERT without any IAR and infusion rate was again increased. Anti-alglucosidase IgG remained stable at 1/6'400. The overall clinical benefit proved however deceiving with continued clinical deterioration. Immunomodulation is becoming a key strategy to conduct successful ERT in lysosomal storage disorders. When ERT is compromised by severe IARs, immunomodulation may be considered. Risks of immune suppression must be weighed against clinical benefits of ERT.

P 7

A case of bowel obstruction in a newborn

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Introduction: Bowel obstruction is one of the most common surgical emergencies in newborns. The incidence is 1 in 2000 live births. The 4 cardinal signs of intestinal obstruction in newborns are maternal polyhydramnios, bilious emesis, failure to pass meconium in the first day of life, and abdominal distention. The main causes of bowel obstruction may be: duodenal atresia, malrotation with volvulus, jejunoileal atresia, distal obstruction, meconium ileus, plug syndrome, Hirschsprung disease, imperforate anus and sepsis or necrotizing enterocolitis.
Case report: We present a term newborn with one episode of bilious (light green) vomiting and failure to pass meconium within the first 36 hours after delivery. First baby, term born at 40 6/7 weeks of gestational age, birth weight 3590 g, vaginal delivery to a 31 year old woman. No complications at birth. At 6 hours after delivery, normal physical examination. At 36 hours old examination he presents abdominal distention, abdominal pain, abdominal skin erythema, hypoactive bowel sounds, normal respiratory and hemodynamic examination and no signs of sepsis. An abdominal x-ray shows a dilated small bowel and multiple gas-fluid levels. Abdominal US is non conclusive. An oeso-gastro-duodenal transit remains normal. A barium enema shows a transition zone between sigmoid colon and rectum. A rectal suction biopsy at day 4 of life shows the absence of ganglion cells that confirms the diagnosis of Hirschprung's disease. After a period of

48 hours fast, intra venous antibiotics and serial rectal irrigation, the abdominal distension disappears. Antibiotics are stopped (negative blood cultures). Day 9 of life, the patient is discharged with enteral feeding and enemas twice per day.
Discussion: Hirschsprung's disease occurs in 1 out of 5000 births. The disease is caused by the failure of ganglion cells to migrate cephalocaudally through the neural crest during weeks 4 to 12 of gestation, causing an absence of ganglion cells in all or part of the colon. Varying lengths of the distal colon are unable to relax, causing functional colonic obstruction. The most common history associated with Hirschsprung disease of a term newborn is either failure to pass meconium in the first 24 hours of life or chronic constipation after discharge from the nursery. Failure to recognize Hirschsprung disease may eventuate in toxic megacolon and death. With proper treatment, most patients live normal adult lives.
(Photos are available)

P 8

Acute complicated appendicitis mimicking intussusception (case report)

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Introduction: Acute appendicitis is a rare and potentially fatal disease in infants. Diagnosis is often made at a complicated stage with high risk of morbidity and mortality thus early diagnosis remains critical. Ultrasonography (US) is helpful but is operator-dependent.
Case report: 5-month old female infant, referred with suspicion of intussusception (palpable mass in sub hepatic area). Feverish (39.5 °C) and decreased food-intake for 3 days, vomited once the day before first examination. Morning of admission, anamnestic was repeated crying spells and normal stools. At admission, no clues for localised or systemic infection, white blood cell count was 18.3 G/l and CRP was 132 mg/l. US was inconclusive for intussusception (adult radiologist); later transferred to paediatric-surgical department with weak abdominal guarding and apyretic. Laparoscopy showed sub-hepatic ulcero-phlegmonous appendicitis with localised pus around the appendix confirmed by pathologist to be an incipient perforated appendix. Patient was discharged after 5-day course of cefuroxim and metronidazole.
Discussion: Acute appendicitis is the most common surgical emergency in children. Incidence varies from 110–140 cases/100,000, of which less than 0.4% occur before age of 1. In infants, initial symptoms are more elusive. Laboratory exams are also of little value. Perforation is a major complication which determines morbidity and mortality and is closely associated with delay in treatment. Nance reported a 100% perforation rate in 10 children under 12 months. Abdominal US remains a valuable tool for evaluation of the acute abdomen.
Conclusions: Despite the rarity, paediatricians and radiologists must think about appendicitis in infants as early diagnosis is crucial.

Bibliography
Nance ML, Adamson WT, Hedrick HL. Appendicitis in the young child: a continuing diagnostic challenge. Pediatr Emerg Care 2000;16:160–162.

P 9

Severe failure to thrive and chronic diarrhea in a 4 months old infant – don't miss Cystic fibrosis despite neonatal screening test

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Introduction: Newborn screening (NBS) for Cystic Fibrosis (CF) was introduced in Switzerland in 2011 using an IRT-DNA-IRT algorithm followed by a sweat test. However, even the best NBS program has some false negative results which should be <5%. Physicians often do not consider CF in the differential diagnosis of an infant with failure to thrive in the presence of a negative NBS result.
Case presentation: A 4 month old girl was admitted to our hospital for further investigations because of failure to thrive, chronic diarrhea and severe malnutrition. She was born preterm in the 37th week of gestation with a birth weight of 2200 g. The mother reported poor feeding and high frequency of greenish stools (6–8 per day). At admission clinical examination was normal except for low weight (P –3.3 SD), length (P –4.2 SD) and head circumference (P –0.78 SD). Work up revealed a non detectable fecal elastase (<15 ug/g stool) and sweat test was elevated (chloride 81 mmol/l). CF was confirmed on genetic testing (homozygous for F508del). Pancreatic enzymes replacement was started, upon which stool frequency decreased and catch up weight gain and growth were achieved. Serum IRT concentration from NBS was 49 ng/mL (cut off 50 ng/mL). The girl did not have delayed meconium passage.
Discussion: Although CF NBS is as a valuable tool for early diagnosis, this case illustrates the fact that NBS does not detect all cases of CF. As every NBS has some false negative results, it is important to consider CF in infants who present with suspicious symptoms regardless of NBS results. Typical CF symptoms in early infancy include failure to thrive, excessive appetite and abnormal stool. Respiratory symptoms in early infancy appear less common than gastrointestinal symptoms.

P 10

Nonbilious emesis and failure to thrive – a neonatal case of primary recurrent gastric volvulus

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Case report: A 10-day-old girl was admitted for repeated postprandial emesis and poor weight gain. She was a full term baby with weight, height and cranial perimeter in normal percentiles. Pregnancy was uneventful. Initially feeding was well tolerated but after the 1st week of life, she progressively had postprandial and projectile vomiting. These episodes were always nonbilious and associated with abdominal pain and vagal-type malaise. Besides an oral thrush, rest of history and physical examination were unremarkable. An anatomical obstruction was suspected. No malrotation, annular pancreas, pyloric stenosis nor gastric volvulus was seen by ultrasound. Eventually, diagnosis of organoaxial gastric volvulus was made with an upper gastrointestinal (UGI) study pointing a horizontalized stomach with inversion of the curvatures. There was also a mild gastroesophageal reflux disease. With treatment of domperidone and milk thickener, patient was discharged in good condition and with less symptoms. Follow up at 2 and 11-week old showed complete resolution of the symptomatology and regular weight gain.
Discussion: Gastric volvulus is an abnormal rotation of the stomach >180°, leading to a gastric torsion and foregut obstruction. It is primary when associated with an anomalous attachment of the stomach to the abdominal wall. When secondary, it is caused by an intrinsic gastric disorder or of the adjacent organs. It can occur along the longitudinal axis, producing an organoaxial volvulus. In radiological studies, the stomach will be horizontalized and "upside down". It can also rotate along the transverse axis, giving a mesenteroaxial volvulus. It will then appear vertical on UGI study. A third type is a combined one. There are many common features between acute and chronic forms, especially nonbilious emesis, epigastric distention and abdominal pain. Yet because of the risk of strangulation and perforation, acute cases imply emergent surgery whereas chronic volvulus is linked to feeding difficulties and failure to thrive. The latter tend also to be associated with primary etiology. Diagnosis is made by imaging- abdominal x-ray, ultrasound and UGI study but is helpful only if symptomatic.
Conclusion: Because of its intermittent nature and vague symptoms, chronic volvulus is more difficult to diagnose but should suspected in case of nonbilious emesis and failure to thrive. Surgery, while necessary for acute form, is controversial in recurrent cases.

P 11

Acalculous cholecystitis and pancreatitis in Epstein-Barr virus infectious mononucleosis – systematic review of the literature

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Background: Acute acalculous cholecystitis and pancreatitis have been occasionally reported in primary Epstein-Barr virus infectious mononucleosis.
Methods: We completed a review of the literature in the US National Library of Medicine and Excerpta Medica databases.
Results: Forty-seven peer-reviewed scientific reports published between 1966 and 2015 were retained for the final analysis. Acute acalculous cholecystitis was recognized in 36 and acute pancreatitis in

14 patients with primary Epstein-Barr virus infectious mononucleosis ranging in age from 3 to 53, median 17 years. None of the 50 patients concurrently suffered from both acalculous cholecystitis and pancreatitis. In all patients, the symptoms of acute acalculous cholecystitis and pancreatitis concurrently developed with those of infectious mononucleosis. Epstein-Barr virus infectious mononucleosis, acute acalculous cholecystitis and pancreatitis resolved spontaneously following a hospital stay of 25 days or less.
Limitations: This work results from the small number of reported cases, often poorly documented and without or with a very brief follow-up. In addition, we were able to document the length of hospital stay but not that of recovery.
Conclusions: This analysis indicates that acute acalculous cholecystitis and pancreatitis are unusual but plausible complications of Epstein-Barr virus infectious mononucleosis. Cholecystitis and pancreatitis deserve consideration in cases with severe abdominal pain. Usually, these complications are rather mild and resolve spontaneously without sequelae.

P 12

Blisters and milk allergy in infancy

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Case report: A 3-month old boy diagnosed with atopic dermatitis 20 days earlier, presented a mildly pruritic eruption on trunk and extremities one week after the introduction of gluten-free cereals and milk formula. Urticaria or Erythema Mutiforme (EM) were suspected. Two days later liquid-filled vesicles appeared on cheeks and tense bullae on extremities. No mucous lesion. Nikolsky's sign was negative. No fever or other systemic symptom. Laboratory work-up showed normal blood cell count, high level of IgE against cow milk proteins (34.9 kU/L, N: <0.35) and elevated tryptase (14.7 µg/L, N: 0–10). Feeding with hydrolysed formula slightly improved erythematous plaques and pruritus. Topical steroids were later administered because of the apparition of new bullae (20/day). Punch biopsy detected subepidermal bulla, dermal inflammatory neutrophilic and eosinophilic infiltrate, IgG/C3 deposits. ELISA-IgG anti BP180 was strongly positive. This is the classic hallmark in bullous pemphigoid (BP). Subsequent clinical remission was fast.
Discussion: BP occurs predominantly in elderly adults. About one hundred of cases are reported in children, mostly in infants. After exclusion of infections and drug reactions, acquired auto-immune blistering disorder should be suspected in case of cutaneous blisters. Skin biopsy is necessary. Differential diagnosis in our case included EM and acquired epidermolysis bullosa. In BP, autoantibodies against proteins BP230 and BP180 of hemidesmosome and lamina lucida are found and have a clear pathogenic role. BP abnormal immune response can be triggered by many factors. Most frequently vaccination is reported, while just one case was related to milk allergy with a complete healing after dietary changes alone. Systemic steroids are otherwise often required, sometimes immunomodulators. In other reports, allergies were not investigated.
Conclusion: A bullous disease in infants can be immune-mediated and triggered by cow milk protein allergy. Dietary changes alone can be helpful without a heavier treatment.

P 13

A case of laparoschisis with biotinidase deficiency

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Case: L, a girl, was born after a pregnancy marked by the findings on ultrasound of a laparoschisis. There were no other anomalies and the pregnancy was uneventful. A c- section took place at 35 weeks because of a suspicion of intestinal distress. APGAR was 9/9/9 and surgical team took care of the laparoschis with the placement into silo of the small bowel and colon. Bladder was also outside the abdominal cavity and was immediately repositioned. Silo was maintained for 9 days. Then a synthetic patch was placed over the abdominal wall defect for progressive abdominal closure. The definitive closure was done at 20th day of life. Post-operative course was uneventful. The neonatal screening showed a biotinidase deficiency (BD). L. was asymptomatic of her deficiency. We performed a second screening at day 13 and added a daily supplement of biotin. It came back also pathological, so enzymatic analysis was performed which showed severe BD.
About the case: to the best of our knowledge, it's the first case of laparoschisis with a biotinidase deficiency (BD). Biotinidase is part of the biotin cycle. Biotin is a cofactor for 4 carboxylases whith roles in fatty acids and amino acids metabolism and gluconeogenesis. Biotinidase cleaves the bond between biotin and the amino acid lysine. It allows the liberation of free biotin from peptide generated through carboxylase degradation or from dietary protein. If not diagnosed with neonatal screening, BD could lead to various lesions, especially mental retardation, ophthalmologic anomalies or hearing loss. Acute presentation occurs usually after several months of life with a child appearing lethargic, hypotonic with seizures, hearing and visual problems, cutaneous rash and alopecia. Unresponsive seizures to medication in young children could be a symptom of BD and a trial of biotin will stop convulsions from minutes to hours. BD incidence is 1/100'000 for severe cases. In Switzerland, the incidence is twice higher at 1/55'000. It's an autosomal recessive disease and exists in 2 forms: severe and partial deficiency, depending on BD activity. Treatment is lifetime biotin supplement. For patients detected with neonatal screening, the outcome is excellent.
Conclusion: BD is the perfect example of a disease with neonatal screening with a potentially serious illness, nearly benign if found early with a simple and safe treatment. Moreover, in neonatal period, BD is asymptomatic and only screening could detect it before symptoms appear.

P 14

New discovered patient with microdeletion 17p13.2p13.1: case report and review of the literature

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Background: The array comparative genome hybridisation is the gold standard for the diagnostic clarification of patients with mental disability when no typical signs or karyotype for a known syndrome are present. The deletion 17p13.1 has been reported in only a few patients with dysmorphic signs and mental disability. Until now, no clear description of symptoms belonging to the disease has been reported. The object of the study is to describe the phenotypic characteristics of this patient and to compare these findings with the few cases that are present in the literature.
Case report: We used array-CGH to identify the cause a mental retardation in a 10-year old girl and we found a de novo microdeletion of chromosome 17p13.2p13.1. Our study concerns the description of this new patient, a 10-year old girl who was born premature after an uneventful pregnancy. Already during the first months of life some signs of psychomotor developmental delay associated to an important muscular hypotonia appeared and more examinations by her pediatrician have shown that other organs together with some facial dysmorphic signs were involved.
Discussion: Mental retardation, facial dysmorphisms, muscular hypotonia and visual impairment with or without strabismus are the prevalent characteristics. Not clear remains the correlation between the imaging findings and the 4 cases of seizures reported. 3/21 children show a cardiovascular involvement (two ventricular septal defect and one patent ductus arteriosus) surgical- or self-resolved, but the prognosis of the patients is still unclear and as unique information are the 33 years old for the oldest patients of the studied group.
Conclusion: Only a very few cases have been reported. For this reason, array-CGH remains a fundamental exam for these children. This minimal part of chromosome contains several already descripted genes, which are plausible to be the cause for the phenotype characteristics and especially for the intellectual disability of the patients. In conclusion, due the little group of reported patients with 17p13.1 microdeletion, the identification of key signs or symptoms together with the prognosis remains difficult and unclear, but the array CGH can be use as a diagnostic gold standard for all these children with intellectual disability when no typical signs for an already known syndrome are present.

P 15

Failure to thrive, developmental delay and lactic acidosis: Maternally Inherited Leigh Syndrome

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Introduction: Mitochondrial disorders (MD) are the most common congenital metabolic disorders (1:5000 births) characterized by respiratory chain defects with phenotypic and genetic heterogeneity. They are coded by nuclear or mitochondrial (mtDNA) genes, explaining Mendelian or maternal inheritance. Clinical features are progressive, multisystemic, mostly associated with lactic acidosis. Leigh syndrome (LS) is a type of MD.
Case report: A 4 month-old girl from non-consanguineous parents was referred to hospital for feeding difficulties, failure to thrive and dystonic movements. The maternal aunt had NARP (Neuropathy, Ataxia, Retinitis Pigmentosa) due to MT-ATP6, m.8993T>G, her 2 daughters died at 2 and 18 years from LS. The patient exhibited low weight, delayed development, axial hypotonia, peripheral hypertonia. Investigations showed lactic acidosis with elevated pyruvate, lactate peak on MRS. Genetic testing revealed homoplasmy (>99% altered mtDNA) for the family mutation, confirming a Maternally Inherited Leigh Syndrome (MILS). Supportive therapy was started. She died of respiratory failure at 8 months.
Discussion: MILS is characterized by developmental delay, abnormal muscle tone, dystonic movements, ataxia, visual loss, seizures, failure to thrive. Lactic acidosis with elevated pyruvate, symmetrical necrotizing lesions in basal ganglia and lactate peak in MRS are typical. Maternally inherited mutations in mtDNA encoded MT-ATP6 are associated with variable phenotypes from adult NARP to infantile MILS, depending on the heteroplasmy of the mutation (NARP<90%<MILS). Treatment is supportive. Death occurs early, often from respiratory or cardiac failure. If undiagnosed, death may appear to be sudden and unexpected.
Conclusion: Hypotonia, developmental delay, failure to thrive, and lactic acidosis are suggestive of MD. Family history plays a key role for early diagnosis. Only genetic tests confirm the diagnosis. Infantile presentations have poor prognosis.

P 16

Congenital toxoplasmosis in Switzerland: screening or no screening?

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Congenital toxoplasmosis (CT) is still a controversial issue regarding the most effective prevention strategy. In the past twenty years, different strategies have been proposed in Switzerland. However, little epidemiological data about their impact have been reported. The present study provides epidemiological data obtained in Lausanne, Switzerland, regarding CT prevalence after the proposed discontinuation of the sero- surveillance of seronegative expecting mothers. Blood samples from mothers and newborns (cord samples) were screened for toxoplasmosis (IgA, IgG and IgM; Western blot) prospectively over a period of 3 years. Newborns with any positive results were further investigated for CT. During the study period, a total of 8090 women were admitted for delivery. 5477 women accepted to enrol. 5585 newborns samples were collected for analysis. Seven newborns appeared to have CT. The prevalence of CT in this study was then of 0.13% (7/5585). 3/7 newborns were lost of follow-up. On the 4 others, none did have symptomatic CT.
Conclusions: we observed a significant higher prevalence in CT compared to earlier observations in the same geographical area. Further studies about incidence and prevalence of CT should be performed in order to confirm or exclude our results and inform about the appropriate prevention strategy to apply in Switzerland.

P 17

Gallbladder and pancreas in Henoch-Schönlein purpura – systematic review of the literature

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Background: Involvement of the bilio-pancreatic system has been occasionally observed in Henoch-Schönleinpurpura.
Methods: We completed a systematic review of the literature in the US National Library of Medicine and Excerpta Medica databases.
Results: Fifty-one reports published between 1977 and 2015 were retained for the analysis. A bilio-pancreatic involvement was recognized in 34 individually well- described cases (male : female = 19 : 15; age between 5 and 55 years) with severe abdominal pain: pancreatits (N = 20), acalculous cholecystitis (N = 11), both pancreatitis and cholecystitis (N = 3). In all pancreatitis cases, full recovery occurred (within ≤3 weeks in three-quarter of the cases). Cholecystectomy was performed in 8 cholecystitis cases. Histopathology of gallbladder, performed in 7 cases, revealed a leukocytoclastic vasculitis in 4 and the distinctive signs of cholecystitis in the remaining 3 patients.
Limitations: This work results from the small number of reported cases, often without or with a brief follow-up.
Conclusions: This analysis indicates that bilio-pancreatic involvement is unusual in Henoch-Schönlein purpura. This complication deserves consideration in patients with especially severe abdominal pain.

P 18

Invasive group A streptococcal disease – three instructive cases

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Introduction: Group A streptococcus (GAS) is a frequent cause of infections in children but invasive disease is uncommon. We present three different manifestations of invasive GAS infection in children.
Case reports: Case 1: A 7-months-old boy was referred to our pediatric intensive care unit with catecholamine dependent septic shock, multi-organ failure and DIC. He also developed an erythematous, desquamative macular rash. GAS was isolated in blood and urine culture confirming the diagnosis of streptococcal toxic shock syndrome. Despite broad- spectrum antibiotic therapy and full intensive care support, he died 15 days after admission from hypoxic ischemic brain damage. Case 2: An 11-months-old boy presented to our emergency department with prolonged febrile seizures. Bacterial meningitis was diagnosed by lumbar puncture and antibiotic therapy started. GAS grew in blood and cerebrospinal fluid (CSF) culture. Clinical course was complicated by persistent seizures, hydrocephalus internus, and subdural hygroma. An external ventricular drain was inserted on day two of hospitalisation. Despite slow clinical improvement he had persistent fever spikes over more than 5 days. Follow-up CSF cultures, first repeated on day two of hospitalisation, were always sterile. Case 3: A 5-year-old girl was admitted to our hospital with cervical lymphadenitis. Three weeks prior GAS tonsillitis with scarlet fever had been treated with oral antibiotics according to current guidelines. Despite intravenous antibiotic therapy the cervical lymphadenitis progressed to an abscess. Surgical incision and drainage were performed and GAS was cultivated from the abscess fluid. Concurrently she complained of hip pain and ultrasound documented bilateral coxitis, interpreted as poststreptococcal arthritis. Echocardiography was performed to exclude cardial involvement.
Conclusion: The course of invasive GAS infections often is complicated by severe and sometimes uncontrollable host inflammation. The management of these infections remains a challenge despite distinct sensitivity of GAS to antimicrobial treatment.

P 19

Complicated influenza infection in three family members

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Introduction: Influenza infection can cause severe and even fatal complications in immunocompromised and elderly patients. However, healthy and in particular younger children can be seriously affected. We

report on a family with a complicated course in two grandsons and the grandmother.
Clinical cases: Case 1. A two year old previously healthy boy with high fever and cough for five days presented in poor general condition to the emergency room. At admission, he was dyspnoeic, but had normal transcutaneous pO_2 saturation (100%) at ambient air and normal blood gas analysis (pCO_2 4.5 kPa). Influenza A antigen was detected on nasopharyngeal secretions; X-ray was consistent with pneumonia. He was admitted to intensive care unit because of suspected sepsis, and treatment with i.v. amoxicillin/clavulanic acid was initiated with short-term clinical improvement. His clinical course, however, deteriorated. Right pleural effusion was diagnosed and subsequently drained. As the boy's condition further deteriorated, lung CT scan was performed. It revealed interlobar encapsulated fluid which had to be drained with a second chest tube, followed by gradual clinical improvement. Blood and pleural cultures were negative, but PCR in pleural fluid was positive for Streptococcus pyogenes. The boy was discharged after 15 days on antibiotics and 20 days of hospitalization in good general and pulmonary condition. Case 2. In the meantime, the boy's four year old previously healthy brother was admitted to the paediatric ward with pneumonia. He was also treated with i.v. amoxicillin/clavulanic acid. Nasopharyngeal secretions were negative for influenza antigen. He developed left pleural effusion requiring drainage and had to be hospitalized for 15 days. No bacteria could be identified in blood and pleural fluid. Case 3. The boys' grandmother, aged 60 years, was hospitalized in the same week in the intensive care unit with influenza A infection complicated by pneumonia with effusion and sepsis. She developed severe ARDS with multiorgan failure and is still hospitalized at the time of abstract-writing (>4 weeks). Streptococcus pyogenes grew both in blood and tracheal secretions.
Conclusion: Viral or host factors may be responsible for this unusual cluster of complicated influenza A in a single previously healthy and non-influenza vaccinated family. Although the brothers' and family history did not reveal any warning signs of immunodeficiency, immunological workup of the family is underway.

P 20

Acute liver failure associated with Human Parvovirus B19 infection: a case report

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Background: Parvovirus B19 has been associated with large spectrum of clinical manifestations including benign erythema infectiosum in children, transient aplastic crisis in patients with hemolytic anaemia, and congenital hydrops fetalis. However, hepatic involvement and especially hepatic failure has rarely been reported in children with parvovirus B19 infection.
Case Presentation: In this work, we report a case of acute hepatitis associated with acute parvovirus B19 infection which evolved towards an acute transitory liver failure in an immunocompetent 13-year-old female adolescent. The patient was hospitalized for acute right lower quadrant abdominal pain and suspicion of appendicitis. Already prior to the laparoscopic appendectomy her liver function tests were elevated, but the patient evolved towards acute liver failure postoperatively only. Parvovirus IgM and IgG for parvovirus B19 were found to be elevated. After five days of supportive therapy, the first signs of hepatic recovery were visible and the patient was discharged a few days later. Immuno-histochemistry could not document the presence of parvovirus in the appendiceal tissue. No abnormality of bone marrow activity was noted during the following post-operative months.
Conclusion: The patient's transient acute liver failure would probably have passed unnoticed without the acute appendicitis. The epidemiology and clinical presentation of parvovirus B19 infections, and especially those with hepatic involvement, are not very well known.

P 21

Septic granulomatosis: perinatal management of a male fetus of a X-linked CGD carrier mother

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Septic granulomatosis or chronic granulomatosis disease (CGD) is a rare inherited primary immunodeficiency due to the defect of the nicotinamide adenine dinucleotide phosphate (NADPH) oxidase that leads to a failure of phagocytic leukocytes to produce microbicidal reactive oxygen species (ROS). Patients are susceptible to severe life- threatening bacterial and fungal infections and excessive inflammation with the development of granulomas in visceral organs. Mutations in any of 5 genes encoding structural or regulatory subunits of the NADPH oxidase complex are responsible for the disease. The X-linked disorder, caused by mutations in the CYBB gene, is the most common form, which is also associated with the most severe clinical phenotype, that could manifest already in the neonatal period with severe infections, especially cutaneous. The other 4 genes are NCFI, CYBA, NCF2 and NCF4 which are inherited in an autosomal recessive manner. A 31 years old pregnant woman was known to be a carrier for X-linked CGD, otherwise healthy. Her male infant was delivered at term. After birth, the ROS production of the NADPH oxidase activity of the newborn was measured and the genetic analysis conducted. The postnatal course was uneventful, apart from an intense erythema with skin lesions consisted of small pustules and papulovesicles on the face, the neck, the upper arms and thighs. A CRP of 42 mg/l was found. He was referred to the neonatal intermediate care unit for observation and further controls. No antibiotic therapy was administered. No changes of the exanthema were observed by the days and an erythema toxicum neonatorum could have been finally diagnosed. Meanwhile, the ROS production resulted clearly pathological, with a strong suspicion of NADPH oxidase inactivity suggesting a CGD. The genetic analysis is still ongoing. The infant was discharged home on the fourth day of life by antibiotic and antifungal prophylaxis. The surviving rate to adulthood is nowadays around the 90%, but the prognosis of the disease is strictly correlated to the promptness of the diagnosis and early treatment. In our case, the familiar anamnesis was essential to perform the biochemical and genetic analysis at birth and to start immediately a prophylaxis. In case of a known maternal carrier for X-linked CGD early measurement of ROS production is fundamental for a timely recognition of the disease and to prevent infectious complications by a prophylactic therapy.

P 22

Pneumococcus horribilis

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Introduction: We present the case of a 4-year old boy with abscess-forming pneumococcal pneumonia necessitating lobectomy and complicated by a nephritic- nephrotic syndrome.
Case report: The patient was admitted with a history of 5 days of cough and fever to a peripheral hospital. Chest X-ray and sonography showed pneumonia of the left lower lobe and antibiotic treatment with amoxicillin and clavulanic acid was started. On day 6 he was referred to our hospital due to recurrence of fever and edema formation. At arrival the boy was pale and in slightly reduced general condition. He showed acroedema and marked elevated blood pressure, proteinuria and hypalbuminaemia. Chest X-ray, sonography and finally CT revealed a pleural empyema with septation and an intrapulmonary abscess of 5 cm diameter in the left lower lobe. Due to the intrapulmonary lesions the patient had to undergo partial lobectomy. In the histologic specimens of the necrotic material gram-positive diplococci could be found. Antibiotic treatment with amoxicillin/clavulanic acid was continued for 21 days. The concomitant nephritic-nephrotic syndrome with repeated hypertensive crises had to be treated with a course of prednisone and a combination of antihypertensive drugs. Within 2 weeks the patient recovered completely and was discharged home.
Discussion: This case demonstrates a rare complication of a common disease. We hypothesize that abscess formation and nephritic-nephrotic syndrome were both caused by pneumococci. We found no underlying disease until now, sweat test and immunoglobulins were normal. In children with pneumonia literature shows that resection of lung tissue is required only in very rare situations.
Conclusion: Pneumococcal pneumonia in children may be complicated not only by pleural effusion but also by abscess formation. Children with pneumonia should be observed closely. Todays favored evaluation by ultrasound alone may miss important intrapulmonary complications and should therefore be accomplished by at least a chest X-ray when unexpected deterioration happens.

P 23

Identifying reservoirs of infections caused by K. kingae: a case-control study about oropharyngeal carriage of K. kingae among healthy adults

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Background: K. kingae is currently recognized as a significant pathogen in the pediatric population. Nevertheless, the possibility for adults to serve as a reservoir of healthy carriers has not been studied.
Method: We conducted a monocentric transversal study on 228 healthy adults to define the carriage rate. Participants were recruited among the staff of a children's hospital, a population exposed to aerosolized droplets from children. A secondary analysis using a case-control method was conducted to assess risk factors for carriage.
Results: We demonstrated an oropharyngeal carriage rate of 2.2% in this population. However, we noticed a striking similarity in the carriage rate among children younger than 4 years of age and adults living with children of that age group (8.8%). Use of day-care facilities for their own children was also demonstrated a risk factor for adultcarriage.
Conclusions: We were able to demonstrate the existence of adult carriage of K. kingae but our results point to a transmission from children to adults. Our results do not allow us to conclude that professional exposure in a hospital setting is a risk factor for oropharyngeal carriage.

P 24

Wolff-Parkinson-White supraventricular tachycardia in Children: a case report

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We report about a thirteen year old boy presenting in our emergency room with chest pain, dyspnea and palpitations. He described two similar episodes in the previous months. At the cardiovascular clinical examination tachycardia with HR: 200/Min, no hearth murmurs, no jugular venous distention, BP 119/63 mm Hg, peripheral pulses present and symmetrical. By the lung examination normal chest excursion, symmetrical vesicular murmur, no wheezing no respiratory distress sign. The electrocardiogram during the tachycardia showed the characteristic delta wave, which is a slurred upstroke in the QRS complex that is associated with a short PR interval. The short PR interval and slurring of the QRS complex is actually the impulse making it through to the ventricles prematurely (across the accessory pathway) without the usual delay experienced in the AV node. So we diagnosed a Wolff Parkinson White Syndrome. The echocardiogram was normal. The Wolff Parkinson White is a supraventricular tachycardia, in which the electrical conduction does not follow the normal way through the atrioventricular node, the bundles of His and the Purkinje fibers but through an accessory pathway antegrade and / or retrograde, located mostly on the left side, which directly connects the atria and ventricles. The prevalence is less than 1 % of the general population. It manifests clinically with palpitations, dizziness, syncope and / or syncope, chest pain and sudden cardiac death. Diagnosis is based on the ECG that showed: reduction of the PR interval (<0:12 sec), prolongation of the QRS (>0.110 seconds), the appearance of delta waves. The differential diagnosis is with ventricular premature beats, bundle branch block, congenital heart disease. Treatment of Wolff-Parkinson-White (WPW)-associated arrhythmias is directed at the underlying cause through the use of radiofrequency ablation of the accessory pathway.

P 25

Drug – Drug interaction between valproic acid and carbapenems: an interaction to know

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Valproate (VPA) is one of the most commonly used antiepileptic drugs in children. VPA is a complicated medication to use because of its complex metabolism and its narrow therapeutic window. Its metabolism is primarily hepatic with 3 routes: glucuronidation, beta oxidation and oxidation by the cytochrome P450 (CYP) 2C9 and 2C19. Due to its metabolism and its capacity to induce CYP, over 600 drugs have reported interactions with VPA.
Case presentation: A 14-year-old boy with LAMM Syndrome and complex focal epilepsy in the context of a folate receptor 1 deficiency was treated for many years with VPA with good seizure control and trough concentrations in the normal range. He was admitted at the UKBB for management of pneumonia with intravenous antibiotics. Meropenem was introduced from 30.6 to 6.7.2015. On 1.7. and 6.7.2015, serum VPA concentrations showed subtherapeutic levels, respectively 7.3 mg/l and 2.8 mg/l (normal range 50–100 mg/l). There were no inducers of CYP2C9 or CYP2C19 in the concomitant treatment and adherence was good. The child didn't present increase of severity or frequency of seizures. After meropenem discontinuation, VPA serum concentration increased and was back in therapeutic range fourteen days later: 50.9 mg/l on 20.7.2015.
Discussion: Regardless of patient's age, several case reports and case series have described significant, rapid and severe decreases in serum VPA concentrations (usually to subtherapeutic values) following initiation of therapy with a carbapenem antibiotic, including meropenem. Although often clinically unnoticed, this interaction may result in loss of previous seizure control. As in our case report, VPA concentrations generally recovered completely within 3 to 14 days. The mechanism responsible for this interaction is unclear. Decreased enteric hydrolysis of the valproate glucuronide (i.e. decreased entherohepatic recirculation) is thought to be principally responsible. Other possible mechanisms include decreased intestinal absorption, decreased hepatic hydrolysis of the valproate glucuronide, increased valproate glucuronidation ans increased renal clearance of valproate glucuronide.
Conclusion: The interaction between VPA and carbapenems is well described in the literature. However, it is often inknown by clinicians. Clinicians should be aware of this interaction and its risk of clinical seizures. If possible, alternative antibiotics should be considered. If not possible, VPA levels should be monitored.

P 26

A case of codeine accidental poisoning

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Opioids have analgesic and central nervous system depressant effects. Prescription of opioid medications are effective in the treatment of acute and chronic pain and as anesthetic agents. They have the potential to be abused for these effects and the associated feeling of euphoria, but on the other side, they may be the source of significant toxicity after exploratory ingestion in young children. Codeine is a natural alkaloid of opium, related to morphine, causing effects on central and peripheral nervous system similar to those of morphine (e.g.analgesia, sedation, cough, constipation, respiratory depression). Accidental ingestion of codeine is ferquently described in children. We report a case of a 36 months old child living in a refugee camp. Report of the first assessment: the patient was pale, apathic, drowsy, hypotonic, disoriented, and with walking deficiency. On clinical examination, he presented a generalized maculopapular rash, respiratory depression and heart rate variability; there was no fever, but an infection of the upper respiratory tract was detected. All the first level blood test were normal with exeption of gasometry, which schowed a mixed acidotyc status. All these findings led us to exclude hypoglycaemia, sepsis or a central nervous system infection. The possible reason for the patient's status could had been an acute intoxication or drug abuse, suggesting us to procede with a drug urine test, which was, infact, risulting positive for codeine and its metabolites. Further parent's description revealed the use of low doses codeine cough suppressant, but considered not enough to justify the test results. This discrepancy between the supposed quantity of assumed codeine and neurological symptoms reported by patient, drove us to suppose about a isoenzyme 2D6 mutation with phenotype "Codein ultra-metabolizer". The patient was treated with naloxone, having complete regression of symptoms after 2 doses administration. This case focuse our attention on possibility of a morphine overdose, using codeine, not related to the administered dose, but to genetic alterations of the metabolism.

P 27

An unusual cause of neonatal liver failure

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Introduction: Galactosemia is an autosomal-recessive inborn error of lactose metabolism with an estimated incidence of 1 in 30,000 to 40,000 livebirths in Europe. In Switzerland, about 2 cases are detected annually by the newborn screening. Classical galactosemia is caused by complete inactivity of the galactose-1-phosphate uridyl transferase (GALT), the enzyme converting galactose-1-P to UDPgalactose. Typical clinical signs are liver dysfunction, failure to thrive, sepsis and cataract occurring within the first days of life when a newborn is fed with breast milk or cow milk based formula. Excluding galactose and lactose from diet is the only long term therapy.
Case report: A preterm breast-fed boy born at gestational age of 35 2/7 weeks (no consanguinity, birth weight 2430 g, APGAR 5/8/10, initial need of CPAP, than good primary adaptation) showed progressive sucking weakness and muscular hypotonia from day 3. Additionally, jaundice persisted despite phototherapy. These symptoms were initially attributed to the prematurity. On day six of life, lethargy and weakness increased and he was transferred to the neonatology unit. Metabolic work-up revealed acute liver failure with severe coagulopathy (INR >7), elevated liver enzymes (ASAT 1210 U/L, ALAT 413 U/l, GGT 253 U/l) and cholestasis (total bilirubin 244 umol/l, direct bilirubin 49 umol/l, ALP 1074 U/l). Simultaneously, the result of the newborn screening was communicated via phone call from the screening centre in Zürich: Increased Galactose (>40'000 umol/l) and inactive GALT, compatible with classical galactosemia. Feeding was immediately switched to lactose/galactose-free formula milk; liver failure was treated with serial transfusions of fresh frozen plasma, substitution with intravenous vitamin K and oral tranexamic acid. Liver function normalized after 12 days. The boy recovered without clinical or sonographic signs of hemorrhage or cataract, and was discharged in good general condition after 21 days.
Conclusion: Galactosemia is rare but has to be considered if a newborn presents with unspecific symptoms like lethargy, jaundice and muscular hypotonia. Because consequences of untreated galactosemia may be deleterious, breast milk or bovine formula milk should immediately be replaced by lactose/galactose-free formula and liver function tests be performed in unclear cases, especially if the result of the newborn screening is pending. Urine testing for reducing substances (clinitest) could help early detection.

P 28

Extremly rare cause of brain death in a newborn

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Case report: The mother presented at 41 1/7 weeks of gestation due to diminished child movements since the previous day. The pregnancy had been uneventful with normal clinical and ultrasound examinations. Because of a pathologic CTG, the baby was delivered via emergency C-Section with Apgar scores of 3/4//4. The girl remained floppy and apnoic and was intubated and transferred to the NICU. Blood gas analysis and lactate were normal. Her head circumference was 39 cm (P>>97) with a wide and bulging fontanel and wide sutures. Her muscular tone remained floppy and there were no spontaneous movements or reflexes. Pupils were fixed and dilated. Reflective retraction of lower extremities probably due to spinal reflexes were present. Cranial ultrasound showed an inhomogenous diencephalic mass lesion of 6.5 × 6.5 × 5 cm and a bedside EEG showed bilateral continuous background activity. MRI examination confirmed the presence of the supraciasmatic lesion and showed severe symmetric impairment of perfusion of all intracranial arteries. Additionally, thrombosis of all cerebral venous sinus was described. The findings were suggestive for an initial cerebral sinus venous thrombosis due to a possible clotting disturbance 7–10 days prior delivery with consecutive bleeding, increase of intracranial pressure and consecutive thrombosis of all other cerebral sinus. Within 10 hours, head circumference increased by 2 cm and cortical background activity in the bedside EEG vanished to inactive tracing. Considering the findings, the parents agreed to redirection of care and the girl died after withdrawal of life support at the first day of her life. To our surprise histological work-up of the lesion at autopsy revealed a primitive neuroectodermal tumor (PNET) with secondary hemorrhage. PNET is a rare embryogenic WHO IV° tumor (highly malignant, rapidly growing) with poor prognosis comprising 0.5–2% of all CNS neoplasias. In neuroimaging, it typically shows a heterogeneous picture with cystic, necrotic and a-/hypervascularized regions. In very rare cases, it occurs neonatally.
Conclusion: Brain death in a newborn due to a rapid growing CNS-neoplasia is extremly rare. Neuroradiologically, neonatal PNET may be difficult to distinguish from a much more common brain hemorrhage. An autopsy is essential in establishing a definitive diagnosis, which allows for genetic counselling.

P 29

From suspected plexus palsy to the diagnosis of a high-grade vesicoureteral reflux (VUR)

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Case: A 4 weeks old boy was sent to our hospital with a palsy of the right arm. The pregnancy was complicated by gestational diabetes mellitus, birth at term by uncomplicated, rapid vaginal delivery. Birth weight was 3540 g, good primary adaptation. At the age of 2 weeks, parents measured a rectal temperature of 39.5 °C on a single occasion without any clinical signs of infection. The temperature returned to normal within 3 hours. At admission, he was well, afebrile. He had a weakness of the right forearm, normal deep tendon reflexes and a discrete swelling of the elbow. Typical radiological findings and markedly elevated inflammation markers led to the diagnosis of osteomyelitis of the distal humerus and intravenous antibiotic therapy with Cefuroxim was started. In the aspirate of a subperiostal abscess grew Enterobacter cloacae, resistant to Cefuroxime, why the antibiotic treatment was changed to Ertapenem. Further workup revealed sonographic signs of pyelonephritis, duplex kidney on the right side and bilateral hydronephrosis. The patient was discharged after 21 days of intravenous antibiotics with antibiotic prophylaxis with sulfamethoxazol/trimethoprim. Voiding cysto-ureterography (VCUG) at 3 months of age showed a VUR grade IV–V on the right and grade II on the left side. The child developed neutropenia (min. ANC 0.46 G/l) which was interpreted as drug induced by sulfamethoxazol/trimethoprim. After switching to nitrofurantoin prophylaxis, neutrophil count returned to normal.
Discussion: In this child presenting with forearm palsy, discrete swelling of the elbow led to further investigations and diagnosis of osteomyelitis of the distal humeral metaphysis. Enterobacter cloacae is an unusual organism causing osteomyelitis. We presume a clinically nearly silent pyelonephritis with hematogenous dissemination to be the primary source of the osteomyelitis.
Conclusions: A newborn with forearm palsy not always has a neurologic problem. If an unusual organism is detected in osteomyelitis, an underlying source of infection should be suspected. In newborns and infants, pyelonephritis can lead to hematogenous complications without causing severe symptoms. Think of neutropenia as a rare complication of antibiotic prophylaxis with sulfamethoxazol/trimethoprim.

P 30

Na, K, Cl or acid-base imbalance in children with urinary tract infections: narrative review

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Background: Recent guidelines on the diagnosis and management of urinary tract infections in childhood do not address the issue of abnormalities in Na, K, Cl and acid-base balance.
Methods: We have conducted a narrative review of the literature with the aim to describe the underlying mechanisms of these abnormalities and to suggest therapeutic maneuvers.
Results: Febrile urinary tract infections are often (approximately 50%) associated with abnormalities in Na and, less frequently, K, Cl or acid-base balance in inpatients 3 years of age or less. Abnormalities may result from a) factitious laboratory results, b) signs and symptoms (such as excessive sweating, poor fluid intake, vomiting and passage of loose stools) of the infection itself, c) improper parenteral fluid management or d) the prescribed antimicrobials. Furthermore, two transient renal tubular dysfunctions may occur in infants with infectious renal parenchymal involvement: 1. a reduced capacity to concentrate urine and 2. pseudohypoaldosteronism secondary to renal tubular unresponsiveness to aldosterone that presents with hyponatremia,

hyperkalemia and acidosis. In addition to antimicrobials, volume resuscitation with an isotonic solution is required in these children. In secondary pseudohypoaldosteronism, isotonic solutions (such as 0.9% saline or lactated Ringer) correct not only the volume depletion but also hyperkalemia and acidosis.
Conclusions: This narrative review suggests that in inpatients 3 years of age or less with renal parenchymal infections, non-renal and renal factors concur to cause fluid volume depletion and abnormalities in electrolyte or acid–base balance, most frequently as hyponatremia.

P 31

Sphenoidal mucocele: rare in children but potentially life-threatening.

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Introduction: Mucocele is a benign cystic lesion of the face sinuses. Only 1% of them involve sphenoid sinus, making this pathology very rare, especially in children younger than 12 years because sinus pneumatization is not yet achieved.
Case report: Healthy 14-year old boy was admitted for a week-long headache, initially treated with paracetamol and NSAIDs. Clinical examination showed low-grade fever (37.9 °C), tumefaction of left eyelid, mild neck stiffness and photophobia. Initial biological inflammatory parameters were elevated (CRP 222 mg/l, PCT 0.62 mcg/l). WBCs count was 14.4 G/l with band neutrophils 9.5%. Cerebrospinal fluid (CSF) revealed 73 leukocytes/ml, proteinorrachia 410 mg/l and glycorrachia 4.0 mmo/l (glycemia 6.1 mmol/l). Enterovirus-PCR was negative. CSF was unfortunately lost for culture. Patient condition deteriorated in the next 24 hours with evident meningeal signs. Laboratory values worsened (CRP 323 mg/l, PCT 1.78 mcg/l, WBCs 10.4 G/L with band neutrophils 30.5%). A second CSF sample showed augmented leukocytes to 2.800/ml, with 75% of neutrophils, hyperproteinorrachia 790 mg/l and hypoglycorrachia 3.6 mmol/l (glycemia 9.2 mmol/l). Ceftriaxone and dexamethasone were started. Hemocultures showed Streptococcus mitis on three occasions but CSF culture remained negative. So, a parameningeal infection diagnosis was retained. MRI showed sphenoidal mucocele complicated by osteitis of the clivus and dorsum sellae, with multiple cerebral thrombi. Work-up for thrombophilia showed heterozygote mutation for Leiden's factor V.
Discussion: Pathophysiology of mucocele is unclear. Sinus obstruction could be the primum movens. Symptoms are primarily caused by compression of the adjacent structures. Progressive and bilateral headache is common, as well as cranial nerve palsy. Eye- threatening conditions with potential permanent visual loss may reveal the lesion. Complications are due to bony erosion and exteriorized sinusitis into the orbit, cranial cavity and nasopharynx. Neuroimaging (CT and/or MRI) has to be done without delay when mucocele is suspected. Early antibiotherapy is necessary but definitive treatment consists in endoscopic sinus drainage and marsupialisation.
Conclusion: In case of febrile persistent headache without evident etiology, sinus mucocele should be considered before neurological complications occur.

P 32

Doctor, I'm tired all the time! A frequent complaint in adolescents care – clinical cases and investigation plan

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Fatigue is a subjective complaint. There is no precise, reliable definition that is well admitted and clinically useful. There is no score for quantification. Fatigue can occur as difficulty or inability initiating activity (perception of generalized weakness); as reduced capacity to maintain activity (easy fatigability); and/or as difficulty in concentration, memory and emotional stability (mental fatigue). Duration of fatigue can be recent (less than one month), prolonged (more than one month), or chronic (over six month). The presence of chronic fatigue does not necessarily imply the presence of the chronic fatigue syndrome. During adolescence, fatigue is a common symptom around the world. Up to 30% of adolescents in different countries suffer from significant morning tiredness more than one day each week, girls more than boys. Chronic fatigue is less likely (0.4% in UK) to occur, and the diagnosis of chronic fatigue syndrome is made on a low level (0.1%). Chronic fatigue and chronic fatigue syndrome seem to be more prominent in adolescence but on a long term prognosis seem to be better than in adulthood. A cause for fatigue in adults is found in about 50% of cases, regarding adolescents, no data exist. We present three clinical cases out of our cohort to illustrate typical findings. The first case is a 13 11/12 years old male adolescent having presented an episode of fever 6 weeks ago with sore throat and cervical/submandibular adenopathy during several days. Since then he developped fatigue and an increased need for sleeping. For 5 days he has to face with an axillary adenopathy. The second one, a 13 8/12 years old male adolescent with fatigue since about 4 months ago with concentration problems and a reduction in school skills. The physical examination doesn't show any pathology. The last one, a male adolescent, 15 6/12 years old, with a constant fatigue from morning to evening and lack of concentration, for more than 6 months. The medical history and physical examination are normal. We will compare their history and findings to the literature and try to propose an investigation plan for adolescents suffering from fatigue in regard to medical history, physical examination and complementary exams. We highlight some physiological changes in adolescent that may influence the level of fatigue. Furthermore, the chronic fatigue syndrome or sleeping disorder will also quickly approach.

P 33

Frontal Fibrous dysplasia: a case report

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Introduction: Fibrous dysplasia (FD) is a rare congenital disorder. It is a primary developmental abnormality of the bone forming mesenchyme in which fibrous tissue gradually expands and replaces the bone. It has been reported to account for 2.5% to 7% of all benign bone tumors.
Case report: A 14-year old Kurdish male in good medical condition, presented to the Emergency Department with a complaint of pain above the arch of the left eyebrow. The pain symptoms had already been present for 7 months. There was no history of trauma, headaches or visual disturbances. On physical examination, there was a left- sided proptosis and cranial nerves were grossly intact. There was no frontal prominence, no café au lait skin macules, and no tender mass was palpated. He had Tanner stage 5. Both cerebral CT and MRI imaging indicated a FD of the frontal bone. The patient was referred to the Neurosurgery Department at the CHUV where further exams are pending.
Discussion: The diagnosis of FD is based on clinical and radiological findings. FD shows no gender bias, is usually appears during periods of growth (early teen and adolescent years). Most commonly, it presents as a monostotic or polyostotic type. Sometimes it is associated with Mc Cune Albright syndrome.The bones most commonly affected are craniofacial skeleton, long bones, and costa. Depending on the type and location of cranial FD, the common symptom is a gradual, painless enlargement of the involved bone, clinically seen as a facial asymmetry, up to severe deformity with devastating functional and aesthetic consequences. The presentation is often unilateral and limited to the craniofacial bones. However this was not the case with our patient. His tumorous mass crossed over the midline towards the roof of the contralateral orbit. Monostotic and polyostotic forms of FD usually produce no new symptoms from the age of puberty and the condition then remains stable throughout the lifespan. Monitoring of these patients involves screening for complications such as fractures and secondary aneurysmal cysts. The prognosis is relative to the risk of sarcomatous degeneration. Biphosphonate therapy has not proven to be effective. Surgery addresses FD if symptomatic and if it is voluminous and weakens the bone depending on the location of the lesions.
Conclusion: Management of patients with FD is a real challenge and requires a multidisciplinary involvement for optimal care.

P 34

Tiptoe walking in a child: have you thought of ...?

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Case report: 6 y.o. boy, born at 34 6/7 GW without perinatal complication, referred for tiptoe walking. Normal walking was acquired by 16 months, and a progressive tiptoe walking was noted since 4 y.o. Family history was negative for neurological diseases. Clinical examination revealed bilateral equinus deformity with pyramidal signs mainly of lower limbs: spasticity, deep tendon hyperreflexia with diffusion, bilateral Babinski and Hoffman signs. Superficial sensitivity

was normal but inferior limbs pallesthesia was decreased to 6/8. No sphincter disorders. Brain and spinal cord MRI with spectroscopy were normal. Metabolic work-up excluded methylene tetrahydrofolate reductase (MTHFR) deficiency, cobalamine C disease and urea cycle disorder. A diagnosis of Hereditary Spastic Paraparesis (HSP), also known as Strümpell-Lorrain disease, is strongly suspected. Genetic testing is pending.
Discussion: Spastic diplegia is frequent: most patients have a history of prematurity, with periventricular leucomolacia on brain MRI. If diplegia is acquired or progressive, one has to first exclude by neuroimaging brain white matter diseases or spinal cord compression, such as tethered cord syndrome, as well as some rare neurometabolic disorders. In the absence of such conditions, HSP is the next diagnosis to consider. It is characterized by progressive weakness and spasticity of lower limbs, and can be pure, as in our case, or complicated (associated with other neurologic abnormality). The major pathologic change occurs in the spinal cord, consisting in axonal degeneration at distal ends of corticospinal tract, and sometimes including dorsal column fibers or anterior horn cells. With a prevalence of 5/100'000, HSP is genetically very heterogeneous, all modes of inheritance being possible. Mutations in at least 59 genes can cause the disease. Thanks to the recent technological advances such as high throughput sequencing, molecular analysis at once of all known genes is now possible. Treatment of HSP is directed towards reducing symptoms through physical therapy, and possibly botulinum toxin injection or intrathecal baclofen pump.
Conclusion: Think of HSP in any child walking on tiptoe and showing pyramidal signs, especially in the absence of a clear etiology such as prematurity or leukodystrophy.

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Paediatric Lyme Neuroborreliosis: a case report

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Lyme disease, caused by the spirochete Borrelia burgdorferi sensu lato, is the most common tick-borne illness. Lyme disease usually begins with the characteristic skin lesion, erythema migrans, at the site of the tick bite. Following haematogenous dissemination, neurologic, cardiac, and/or rheumatologic involvement may occur. Neurologic involvement occurs in up to 15% of untreated B. burgdorferi infection. We introduce a 12-year boy who presents in our emergency room with peripheral facial nerve palsy. No fever is referred and the remaining neurological examination is normal. Parents report a tick bite in the upper zone of the left leg who occurs about two months ago. After about ten days sign of erythema migrans appeared at near by and was treated with local antibiotic therapy. Supposing a neuroborreliose we performed serologic testing to search a Borrelia Infection.By positive IgG and IgM and positive Western Blot we performed a lombar punction. The serology in cerebrospinal fluid was negative. Our patient was treated with intravenous antibiotic therapy with Ceftriaxone (2 g once daily fot 14 days with a good responce and complete regression the facial palsy). From literature it is known that the diagnosis and management of Lyme disease in children is similar to that in adults with a few clinically relevant exceptions. Children may present with insidious onset; prompt diagnosis and treatment of this condition is important. Children who acquire Lyme disease have an excellent prognosis even when they present with the late disseminated manifestation of Lyme arthritis. Guidance on the judicious use of serologic tests is provided. Pediatricians should be familiar with the prevention and management of tick bites, which are common in children.

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Rapid lethal early infantile encephalopathy due to a mutated SLC19A3 gene

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Introduction: Early onset infantile encephalopathy is a heterogeneous entity with variations in the clinical course, variable genetic background and diverse treatment options. We present the dramatic clinical course of a 2 month old girl affected by lethal early onset encephalopathy due to a genetic defect in the SLC19A3 gene.
Case Presentation: At the age of 6 weeks, a full-term girl with an uneventful history during pregnancy, birth and first weeks of life developed within a few days a dramatic regression of neurological functions with somnolence, feeding difficulties and muscular hypotension. The main laboratory abnormality was an elevated blood lactate level (3.3 mmol/L, normal 1.0–1.8). Brain MRI 4 days after onset of symptoms revealed a pronounced vacuolating encephalopathy with cystic necrosis of white and grey matter including basal ganglia, brainstem and cerebellum and a restricted diffusion in cerebellum, mesencephalon, pons and corpus callosum. Since already the older brother had presented at the age of 5 weeks with similar symptoms and died at the age of 5 months of respiratory failure, a common inherited aetiology was suspected. When RT-PCR was done in both siblings, there was no SCL19A3 transcript in cDNA from fibroblasts. A low level of thiamine in cerebrospinal fluid (9.2 nmol/L, normal 28–106) supported this.
Discussion: MRI pattern recognition has been proposed to classify a new group of early onset encephalopathies with mutations of the SLC19A3 gene (1). SLC19A3 encodes the second thiamine transporter and is ubiquitously expressed. So far three different phenotypes caused by mutations in SLC19A3 are known with milder clinical courses and variable brain MRI patterns. The clinical course, brain MRI, positive family history and the genetic results in our patient corresponds to the findings in the new group with SLC19A3 mutations.
Conclusion: In early onset encephalopathy with a dramatic and rapid regression of neurologic functions and MRI findings of a subtotal brain degeneration with cystic necrosis of white and grey matter, basal ganglia, brainstem and cerebellum a mutation of SLC19A3 should be considered. Even though in this disease brain damage is already extensive at the time of diagnosis and despite a lack of treatment options, a confirmation of the diagnosis is needed for genetic counselling of the family.

P 37

Congenital hypomagnesemia – a rare cause for neonatal seizures

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Case presentation: Neonatal seizures are not a rare condition and mostly caused by hypoglycemia. We report on a 9-day old female, full-term newborn, who presented with recurrent, self-limiting movements of the right leg and finally was brought to our emergency room for a tonic-clonic seizure. Physical examination, cerebral ultrasound and the EEG were normal, but in the laboratory work-up a severe hypocalcemia and hypomagnesemia were found. These electrolyte disturbances were corrected by intravenous calcium and magnesium supplementations at first and followed by oral calcium replacement. Due to the fact that the hypocalcemia and the hypomagnesemia appeared in concert with an inadequate normal PTH value, we thought of a primary hypomagnesemia with secondary hypocalcemia. Also, because the urinary loss of magnesium was low, an intestinal magnesium resorption defect was hypothesized rather than a renal tubular defect. TRPM 6 mutations are known to cause this clinical phenotype. Therefore, we adjusted our therapy to this working hypothesis, stopped the calcium and started a magnesium supplementation only. The clinical follow-up was favorable and the laboratory follow up showed normal values for both, magnesium and calcium. She had no further seizures and develops normally. To date the genetic analysis for TRPM 6 mutations is pending.
Background: Hypomagnesemia with secondary hypocalcemia was first described by Paunier from Geneva in 1968. Today we know that TRPM 6 mutations are responsible for this condition. TRPM 6 is an intestinal magnesium transporter, which is responsible for the transcellular magnesium transport in the intestine specifically. Children with TRPM 6 mutations typically present with generalized seizures. They have extremely low levels of magnesium with moderate hypocalcemia, which is due to the impairment of the PTH function. Affected children are successfully treated with high doses of oral magnesium supplementation, which is needed lifelong. Neurological outcome is very good, if diagnosed early, avoiding secondary damage by seizures.

P 38

An atypical case of neuroborreliosis

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Introduction: Neuroborreliosis is a tick-transmitted infectious disorder of the nervous system caused by the spir ochete Borrelia sp. Clinical manifestations of Lyme disease vary most probably because of different subspecies but in generally may include systemic symptoms and dermatologic, neurological, cardiac and musculo-skeletical involvement. We are reporting this case as an unusual pediatric presentation of the disease.
Case report: A previously healthy 5 year-old girl presented to our Pediatric ER with intermittent headache over the last 5 months without fever or other neurological symptoms. For the past 10 days she had been complaining of intermittent bilateral lower limb muscle pain. There was no recollection of tick bites or other skin findings.On presentation the vital signs and the physical exam were within normal limits. A head MRI was normal. Initial serologic laboratory tests in serum revealed the presence of Borrelia Burgdorferi specific antibodies IgG et IgM. Western blot assay confirmed the diagnosis. Cerebrospinal fluid (CSF) revealed an elevated cell count of predominantly lymphocytes, an elevated CSF protein and the presence of Bb-specific antibodies (IgG). Based on these findings a 2-week treatment of intravenous Ceftriaxone was initiated with subsequent symptom resolution.
Discussion: Neurological symptoms in neuroborreliosis usually occur 4 to 6 weeks after a tick bite; it may affect the central and/or peripheral nervous systems. Facial paralysis is the most common manifestation in pediatric patients with neuroborreliosis (55%). However only 40–50% of the patients can recall a tick bite and only 20% report a local reaction. The reported case was atypical because of the absence of typical symptoms and the overall benign, presenting general condition of the patient.
Conclusion: Neuroborreliosis should be considered in a child presenting even with minimal neurological symptoms especially if no other typical etiology can be identified. Specialized investigations on serum and CSF are required for the confirmation of the diagnosis.

P 39

An atypical case of Arnold Chiari type 1

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Introduction: Arnold Chiari malformation type 1 is a disorder usually diagnosed in adults, defined by anatomic anomalies of the cerebellum and the cranio-cervical junction in association with syringomyelia. Symptoms and signs involve various areas of the central nervous system including the visual system, the cerebellar system and its pathways as well as motor and sensory pathways.
Case report: A previously healthy 20 months-old boy presented with a 4 day history of headache and generalized weakness; the appearance of an unsteady gait for a few hours and one episode of emesis prompted the parents to consult the Pediatric ER. The mother mentioned a head trauma one week ago. Clinically the patient was afebrile, fully conscious with an age-appropriate behavior. The neurological examination revealed an ataxia and a horizontal nystagmus. The sensory and motor examinations were within normal limits as was the rest of the clinical examination. The initial laboratory findings were unremarkable. A brain MRI showed an abnormal cerebellar tonsillar downward displacement associated with a syringomyelia. The patient was subsequently transferred to the Vaud Cantonal University Hospital for further neurosurgical care.
Discussion: This case is atypical because of the age at presentation, the acute onset and the atypical clinical signs. The clinical presentations of Arnold Chiari type 1 in a toddler are rare; the mean age of diagnosis is reported around 18 years. However a few cases of Arnold Chiari type 1 have been reported in children a few days following a head trauma. The symptoms may occur with variable frequency and in variable combinations, which explains the particularly challenging diagnosis.
Conclusion: Although the presentation of Arnold Chiari type 1 is atypical in children, it should be suspected in previously healthy children with acute onset cerebelar signs, in particular after a head trauma.

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Long-term outcome after arterial ischemic stroke in children and young adults

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Objective: To compare long-term outcome of children and young adults with arterial ischemic stroke (AIS) from 2 large registries.
Methods: Prospective cohort study comparing functional and psychosocial long-term outcome (≥2 years after AIS) in patients who had AIS during childhood (1 month–16 years) or young adulthood (16.1–45 years) between January 2000 and December 2008, who consented to follow- up. Data of children were collected prospectively in the Swiss Neuropediatric Stroke Registry, young adults in the Bernese stroke database.
Results: Follow-up information was available in 95/116 children and 154/187 young adults. Median follow-up of survivors was 6.9 years (interquartile range 4.7–9.4) and did not differ between the groups ($p = 0.122$). Long-term functional outcome was similar ($p = 0.896$): 53 (56%) children and 84 (55%) young adults had a favorable outcome (modified Rankin Scale 0–1). Mortality in children was 14% (13/95) and in young adults 7% (11/154) ($p = 0.121$) and recurrence rate did not differ ($p = 0.759$). Overall psychosocial impairment and quality of life did not differ, except for more behavioral problems among children (13% vs 5%, $p = 0.040$) and more frequent reports of an impact of AIS on everyday life among adults (27% vs 64%, $p < 0.001$). In a multivariate regression analysis, low Pediatric NIH Stroke Scale/NIH Stroke Scale score was the most important predictor of favorable outcome ($p < 0.001$).
Conclusion: There were no major differences in long-termoutcome after AIS in children and young adults for mortality, disability, quality of life, psychological, or social variables.

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Two cases of an unusual cause of headache and papilledema not to be missed

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Background/Purpose: Chronic headache is a frequent reason for referral to paediatric neurologists. Rarely, but important, history and clinical findings can lead to the diagnosis of a genetically determined, treatable form of headache caused by chronic meningeal inflammation.
Case description: 2 children aged 5 resp. 6 years presented with chronic headache, spontaneously resolving fever periods and papilledema. One child, furthermore had a chronic monarthritis of the left knee (bacterial arthritis/osteomyelitis excluded), the other child showed a non-pruritic rash waxing and waning since birth. Both children had elevated cerebrospinal fluid opening pressures (CSFOP) with 30 cm H_2O and 32 cm H_2O, respectively. In one child, audiological investigations showed a mild to moderate sensorineuronal hearing impairment. Molecular genetic testing for periodic fever syndromes revealed mutations in the NLRP3-Gene in both. Both patients responded to treatment with acetazolamide and canakinumab with disappearance of headaches, normalization of CSFOP and hearing impairment.
Conclusions/discussion: In both patients with slightly different phenotypes, the diagnosis of chronic infantile neurologic, cutaneous and arthritis syndrome (CINCA), the most severe form of the cryopyrin-associated periodic syndromes (CAPS), was confirmed. Affected patients may present with chronic central nervous system and inner ear inflammation, episodic fever, a neutrophilic rash and arthritis. CINCA typically occurs sporadically with de novo mutations of the NLRP3-gene, which encodes for a protein involved in IL-1β-activation. In attempt to prevent end organ damage with IL-1β-blocking therapy, early diagnosis of CINCA in patients with headache and/or papilledema is crucial.

P 42

Critical illness polyneuropathy and myopathy (CIPNM): a challenging diagnosis in children

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Severe muscle weakness and failure to wean from the ventilator are considered characteristic features of critical illness polyneuropathy and myopathy (CIPNM). There are only a few published data regarding definition, clinical criteria, incidence, aetiology, management, and long-term outcomes of CIPNM in children. We retrospectively reviewed over a 1-year period, data of 6 critically ill children with severe muscular weakness and long-term ventilator support with regards to clinical symptoms, risk factors, diagnostic investigations, and outcome. Sepsis or systemic inflammatory response syndrome as a risk factor for CIPNM was present in all children. One patient, the only male, experienced a severe systemic drug reaction with full recovery. In two girls diagnoses for encephalopathy and encephalitis, respectively, were made during the course of hospitalisation. Three of our 6 patients met the clinical criteria of CIPNM including proximal muscle weakness, sensory deficits, and absent reflexes. In terms of CIPNM outcome, two girls died while one experienced full recovery without sequelae. Only 1 girl received a complete CIPNM positive diagnostic investigation with electrophysiological examination and muscle biopsy. Though severe muscle weakness and atrophy, pathologic reflexes, sensory deficits, and failure to wean from the ventilator are common findings in paediatric critical care settings, a systematic approach and examination to identify CIPNM is lacking. Given the impact on morbidity and outcome we conclude that CIPNM should be ruled out in all children presenting anamnestic risk factors and typical clinical signs.

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Melkersson-Rosenthal syndrome in paediatrics: case report and literature review

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Background: Melkersson-Rosenthal syndrome is a rare neuromucocutaneous disorder of uncertain etiology, characterized by a triad of orofacial edema, recurrent facial nerve paralysis and a fissured tongue. The classic triad is rarely seen at initial presentation, presenting more commonly in its oligosymptomatic form. It is a syndrome usually seen in adolescence with only a few reported pediatric cases.
Case report: We present the case of a 15-year-old girl with two episodes of unilateral facial nerve paralysis, painless orofacial edema and swelling of the tongue. The first episode was at the age of nine and persisted for three months. The second episode was at the age of 13 and persisted for five months. In both episodes, the laboratory values were within normal ranges. Instrumental assessment like electroneuronography and head MRI supported the diagnosis of Melkersson-Rosenthal syndrome. Oral steroid therapy was administered with full symptomatic resolution. At two years follow up, there were no signs of recurrence.
Discussion: Melkersson-Rosenthal syndrome is a rare disease in the paediatric population partly because of misdiagnosis of the oligosymptomatic form. This case report and literature review focuses on paediatric patients, supplying additional information in the diagnosis and treatment of children with recurrent facial nerve paralysis and orofacial swelling.

P 44

Nursing screening of mental suffering in male adolescents in Emergency

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Background: The adolescent mental health is a major concern. "Half of all mental health disorders in adulthood appears at age 14, but most cases are undetected and untreated (WHO, 2014). The first cause of the mental disease in adolescence is the depression. In Switzerland, the prevalence of the mental diseases in hospitalized adolescents aged between 10 and 14 years is 8.4% and in those aged between15 and 19 years is 13%. Globally, around 1.3 millions adolescents die each year. After road injury and HIV, suicide is the third cause of death. In Switzerland, suicide remains the leading cause of death among young people aged 15–24 years, especialy in male adolescents. Rates of adolescent suicide attempts are underestimated and only 10–20% of them receive an appropriate care. Male adolescent show their unhappiness differently than girls: behavior disorder, risk conducted, and somatic complaints. Nowadays, when male adolescents consult Pediatrics Emergency Departments (PED) for somatic reasons, assessment and treatment of physical pain stay the priority and mental suffering lacking often. Screening for psychological distress with somatic manifestations in adolescence remains the best prevention of suicide.
Aims: To develop a nursing intervention in PED for the early detection of the mental suffering among male adolescent with somatic complaints. Study objectives: 1. Describe the components and characteristics of mental suffering of male adolescents. 2. Develop a systematic screening intervention of mental suffering in male adolescents and a nurse consultation in male adolescent health. 3. Evaluate the feasibility of the intervention.
Methods: This feasibility study will be organized with a sequential exploratory mixed design.
Conclusions: This research will contribute to highlight the particularity of the nursing care in PED and to determine mental suffering in male adolescents presenting with somatic complaints at the emergencies' departement and provide appropriate care.

P 45

Seizing the opportunity: The role of the school nurse in secondary schools in Geneva, Switzerland

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Objectives: Adolescents consult health professionals mainly for somatic complaints, but have difficulties in seeking help for psychological problems. Schools play an important role in detecting health issues in adolescents. In Geneva (Switzerland), school nurses are present part-time in public schools, offering consultations without appointment. When the nurse is absent, pupils are received by administrative personnel delivering basic care. Our objectives were to record the reasons for spontaneous visits to the school nurse in 12 to 15-year old adolescents, to describe the management by the school nurse, and to compare it to the care delivered by administrative personnel.
Methods: Data from four secondary schools in Geneva were analyzed. The reasons for spontaneous consultations and their management were recorded by school nurses and administrative personnel, during varying time periods between 2005 and 2010. Four school nurses additionally provided detailed descriptions of the management of spontaneous visits during one month.
Results: Spontaneous visits to the school nurse varied from 280 to over 900 per academic year. Headaches and abdominal pain accounted for two thirds of all visits. Pupils received by administrative personnel were mostly sent home (87%), whereas two thirds of pupils stayed at school when seen by the nurse. The nurses' detailed descriptions showed that in addition to the acute care, they offered follow-up visits and evaluated the psychosocial situation.
Discussion: Adolescents at secondary schools in Geneva mainly consult for somatic complaints, confirming previous findings. Visits to the school nurse decrease the amount of days missed. Nurses also evaluate health issues in further detail and may thus contribute to improve management of adolescents' health concerns and increase access to preventive services. We suggest points for reflection on how these services can be further developed in line with the principles of Youth Friendly Health Services.

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Unilateral tonsillar enlargement: Think about cancer!

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Unilateral tonsil hypertrophy with swelling is common in the course of acute pharyngitis (viral or bacterial) and usually point at a local bacterial complication. A malignancy should be identified as a possible although rare aetiology of unilateral tonsil enlargement. Non-Hodgkin lymphomas (NHL) of the palatine tonsil are the most common cause of extra-nodal NHL. We report the case of a teenager with a Burkitt's

lymphoma of the left tonsil. An otherwise healthy 14 years old girl presented with a five days history of dysphagia and left sided otalgia with snoring. She had no other loco-regional nor systemic complaints, like fever, night sweats or weight loss (B-symptoms). She was in good general condition, showing only a distinct enlargement of the left palatine tonsil, with mucous discharge and without cervical lymphadenopathy. Complete blood count was normal, rapid test for Group A streptococcus negative. An acute upper airway viral infection was diagnosed and ibuprofen started without relieving the symptoms. By persisting unilateral tonsillar hypertrophy of unclear aetiology, a biopsy was performed by an ENT specialist one week later and secured the diagnosis of Burkitt's lymphoma. Tumor staging showed no other localisation of the disease. The girl was treated with intensive systemic chemotherapy over 3 months and achieved a prompt and complete remission. Upper respiratory infections are the most frequent causes of illness in childhood. Pharyngitis with a rapid unilateral tonsil hypertrophy without clinical improvement under non-steroid anti-inflammatory drugs is a red flag and should lead to look for a malignancy. Lymphomas are the second most common haematological malignancies in children. Approximately 15% of head and neck lymphomas in children affect the Waldeyer's ring. Extra-nodal involvement is more common in non-Hodgkin Lymphoma (NHL) and the palatine tonsil is the most frequent site of involvement for extra-nodal NHL. The most common clinical manifestations of lymphoma in palatine tonsils are: unilateral and usually painless tonsil enlargement, alteration in the appearance of the mucous and cervical lymphadenopathy. B-symptoms are present in 16% of patients only. A biopsy must be done early in these circumstances. The prevalence of malignancy in unilateral tonsil enlargement without other clinical symptoms is very low; a rapid enlargement of the tonsil, local mucous changes or signs of loco-regional extension are a redflag.

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Score for individual surgical strategy definition in bone tumors – what for?

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The choice of surgical strategy after neoadjuvant chemotherapy remains difficult because of the diversity of locations, vascular implications, age of the child. We developed a strategic score objectifying and defining management for local tumor control in children. 9 criteria were included in scoring system: Age, tumorlocation, diaphyseal /epiphyseal, tumorsize, metastases, therapy resistent pain, joint invasion, neurological/vascular invasion, pathological fracture. Dependent on importance of influence in decision addition (0–5 points) vs multiplication (×0.1–1) was chosen based on clinical experience. Still retaining a large potential for growth in children reconstruction pose problems due to residual growth. Tumor location may implicate severe consequences if amputation especially in the upper limb seems inevitable. Simple resection of diaphyseal lesions fascilate strategic opportunities. Joint invasion complicates the resection, reconstruction remains challenging in growing children. Invasion of the neurovascular bundle is important for local tumor control and possibilities of functional reconstruction vs amputation. Millimetric values may be tolerated depending on the localization/compartment. Pathological fracture theoretically required amputation in order to achieve extra-compartmental resection. The decision remains controversial in the literature. Metastasis except skip lesions will not influence resection strategy, but delay in resumption of chemotherapy should be avoided. This score allow to determine the absolute indication for amputation/ conservative surgery. Intermediate scores allow conservative surgery but with difficulties to be highlighted.Used as part of the initial tumor board decision leading to a comprehensive and interdisciplinary definition of an individual strategy and so to a clear information of the patient and the family.

P 48

The risk of children and adolescents with cancer to develop fever in neutropenia: Evolution over two decades, 1993–2012, in a single center

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Background: Fever in neutropenia (FN) is the most frequent potentially life- threatening complication of chemotherapy in children with cancer. This study aimed to investigate the long-time evolution of the risk to develop FN and associated risk factors.
Methods: This retrospective cohort study in children and adolescents diagnosed with cancer before 17 years covered two decades (1993 to 2012) in a single institution. FN was defined as fever in severe neutropenia (absolute neutrophil count <0.5 G/L) induced by chemotherapy. Poisson and mixed Poisson regression was used for analysis.
Results: Cancer, including Langerhans cell histiocytosis and all tumors of the central nervous system, was diagnosed in 800 patients. Of them, 583 (73%) received chemotherapy and could be studied here. During 692 years of cumulative chemotherapy exposure time, 712 FN episodes were diagnosed (rate 1.03 per year; exact 95% confidence interval, 0.95 to 1.11), 154 of them with bacteremia (0.22 per year; 0.19 to 0.26). The risk to develop FN (rate ratio per decade, 0.97; 0.72 to 1.30; p = 0.83) and FN with bacteremia (0.88; 0.38 to 2.04; p = 0.76) did not change significantly over time. Significant changes over time were detected in 4 of 11 characteristics potentially associated with FN. In univariate analysis, 8 of these 11 characteristics were significantly associated with FN. A significant interaction of these associations with time was detected in 5 of them. In multivariate analysis, the risk to develop FN was significantly and independently associated with 6 of 11 variables studied: higher chemotherapy intensity, bone marrow involvement, central venous access device, prior FN, shorter time since diagnosis, and relapse. Only higher chemotherapy intensity and bone marrow involvement were significantly and independently associated with the risk to develop FN with bacteremia.
Conclusion: Significant and clinically relevant changes over time were detected in characteristics associated with the risk to develop FN. Additionally, significant interactions with time were detected in the majority of the respective associations. Clinically, they reflect changes in therapy of paediatric cancer over time. Scientifically, these changes need to be accounted for in longitudinal research projects.

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Incidence, distribution of age gender, serotypes and presentation of clinical manfestation in Switzerland 1999–2010

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Meningococcemia especially in children and young adults is still life-threatening. Fulminant sepsis is the most severe complication with high morbidity and mortality.In 1999–2002 in Switzerland, as in other European countries, incidence of meningococcemia increased substantially. We present a retrospective non-interventional cohort study of 710 patients aged 0–19 years, suffering from meningococcemia in 1999–2010 regarding incidence, distribution of age, gender, serotypes and clinical manifestation (meningitis, sepsis, septic shock, waterhouse friederichssen) and mortality. In conclusion the bimodal distribution of age has been confirmed and remained unaltered in the time of epidemic outbreak 1999–2002. Boys are at higher risk. Serotype distribution was the same in either meningitis or sepsis, as their incidence rose proportionally to the incidence of meningococcemia. However the incidence of fatal waterhouse friederichssen's manifestation did not rise. The fatal outcome was more likely associated with serotype C, whereas serotype B was linked to less severe clinical outcome. In 50% of cases no subtype could be detected.
Discussion: High incidence of meningitis and lower frequency of fatal waterhouse friederichssen during meningococci epidemy was probably the most important reason for proportionally lower lethality 1999–2002. However in this time period also a high alerteness for the illness and an aggressive management of septic children could explain reduced mortality.

P 50

Symptomatic essential thrombocythemia in a female adolescent. A case report

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Introduction: In paediatric patients reactive thrombocytosis is the most common cause of thrombocytosis and usually clinically silent and self-limited. Further investigations are required for persistent symptomatic thrombocytosis.
Case: A 14 years old female patient consulted her general practitioner with a two weeks history of recurrent stabbing pain and erythema at the soles of her feet and fingertips. Her blood count revealed a massive thrombocytosis of 2373 × 10^9/L. Vital signs and clinical findings, including a detailed neurological examination, were normal. Retrospectively, elevated platelet counts were documented for more than 12 months before the initial presentation at our service. After exclusion of familial thrombocythemia, we suspected essential thrombocythemia (ET). Cerebral ischemia was excluded by MRI scan. Ultrasound showed splenomegaly of 14.5 cm and bone marrow morphology demonstrated elevated numbers of mature megakaryocytes typically observed in ET and no evidence of myelofibrosis. Molecular analyses were negative for JAKV617F as well as mutations in CALR and MPL which are frequently associated with ET. Further laboratory analyses showed acquired von Willebrand syndrome and clinically suspected secondary thrombocytopathy with two episodes of bleeding tendency under the influence of acetylsalicylic acid. Von Willebrand factor antigen (48%) and activity (27%) were reduced. After initial thrombapheresis, therapy with high dose hydroxycarbamide (20–40 mg/kg/day) and acetylsalicylic acid helped to significantly reduce platelet counts to normal and showed a clinically stable follow-up during 11 months. Dose-reduction or short-term discontinuation of hydroxycarbamid due to neutropenia led to immediate relapse of thrombocytosis.
Background: ET is a chronic myeloproliferative neoplasm with clonal megakaryocyte proliferation. It is an extremely rare cause of thrombocytosis in childhood and adolescence with an estimated annual incidence of 0.09 per million in children under 15 years of age. If treated adequately, complications of thrombocytosis due to thromboembolism or bleeding are rare and the presence of somatic markers are uncommon in childhood ET.
Conclusion: Essential thrombocythemia should be considered as differential diagnosis in children and adolescents with persistent thrombocytosis after exclusion of other specific reasons like infection or thrombocytosis after splenectomy.

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4-Year Follow-up of Ultrasound-Based Diagnosis and Non-surgical Treatment of Developmental Dysplasia of the Hip in Mongolia: A Prospective Cohort Study

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Purpose: Avascular necrosis of the femoral head and residual dysplasia can occur after non-surgical treatment of developmental dysplasia of the hip (DDH). In former studies, 0 to 14 percent of treated hips developed avascular necrosis of the femoral head; 2 to 29 percent developed a residual hip dysplasia. Both are indications for surgical procedures and cause pain and early osteoarthritis despite interventions. We therefore aimed to determine their prevalence in a prospective cohort study of Mongolian newborns.
Materials and Methods: Hips of all children born within one year in the largest pediatric hospital of Mongolia (n = 8356) were examined by ultrasound at a median age of one day and treated with Tubinger splint if DDH was present (n = 107). All treated children could be discharged with heathy type 1 hips after monthly checks by ultrasound. A representative sample of 51/107 treated children was followed up at 3–4 years of age with conventional radiography. We determined 1) the formation of the femoral head (condensed) and joint space (narrowed) as signs for avascular necrosis; and 2) the acetabular angle (≥28 degrees in ≤3 year old participants or ≥25 degrees in those >3 years) as sign for residual dysplasia. Furthermore, we asked the parents about swaddling.
Results: No child showed signs for avascular necrosis. One child had a sign for residual dysplasia (25.8 degrees on the left at age 3.5 years). Angles in all other children were below the thresholds and highly variable, ranging from 11.1 to 26.2 degrees. They were slightly higher in girls than boys, and on the left compared to the right. Swaddling behavior did not affect the results.
Conclusion: Ultrasonographic diagnosis of DDH and treatment with Tubinger splint within the first few weeks of life is safe and efficient in preventing surgical interventions. The prevalence of avascular necrosis of the femoral head and residual dysplasia in Mongolia is among the lowest in literature.

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"My eye is tearing and red": common symptoms but…

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Introduction: Clear discharge and red eye are common symptoms seen in emergency pediatrics. The causes range from mild conditions to sight-threatening ocular emergencies. Juvenile xanthogranuloma (JXG) is a rare benign, proliferative histiocytic disorder of dermal dendrocyte origin. It occurs predominantly in young children with the eye being the most frequent site of extracutaneous involvement.
Case report: A 3-month old girl presents with a history of watery discharge and conjunctival irritation of the left eye for 3 weeks. A course of 10 days topical antibiotics had shown no improvement. Her mother additionally reported a progressive discoloration of the inferior part of the iris. There was no history of obvious ocular traumatism and no general symptoms were reported. The left eye showed a reactive mydriasis with hyphema and loss of red reflex. The girl was immediately referred to the University Eye Clinic. Ophthalmic examination revealed corneal opacity and an elevated intraocular pressure (IOP) of 40 mm Hg was estimated. The infant received an intensive topical corticosteroid and IOP-lowering treatment (Pred Forte 1% 3×/d and Cosopt 2×/d) with normalisation of the IOP within 10 days. The slit lamp examination showed a crescent-shaped beige-brown material in the inferior quadrant of the anterior chamber and on the anterior iris surface. Based on the clinical findings, JXG of the iris was suspected. The examination of the entire skin showed one yellow-reddish abdominal lesion of 5-milimeter diameter. The histology of the abdominal lesion confirmed the diagnosis of a JXG.
Discussion: JXG is a rare and benign, proliferative disorder of histiocytic cells. Lesions usually appear in the first two years of life. The skin is mostly affected, but every organ or system can be involved, typically the uveal tract of eye. Ocular JXG affects 0.3 to 0.5% of all cases. The treatment depends on the site of involvement and the presence of complications.
Conclusion: Children with persistent discharge should always be referred to an ophthalmologist for further investigations. A rigorous clinical examination should be performed to detect any abnormality. Complications as glaucoma or hyphema could lead to amblyopia or blindness and need proper management.

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Triple-therapy in ABCA3 deficiency

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Introduction: Recessive mutations in the ATP-binding cassette transporter A3 (ABCA3) are rare causes of childhood interstitial lung disease (chILD). ABCA3 is a protein expressed in alveolar type II cells which plays an important role in the transport of phospholipids needed for pulmonary surfactant production. ABCA3 deficiency causes severe neonatal respiratory distress. Most affected infants die in the first year of life. There is no established therapy for infants affected by the disease.
Case report: We report on a 4 week old term-born female referred because of failure to thrive and tachypnea. She presented with mild signs of respiratory distress and normal oxygen saturation. Chest X-ray showed diffuse shadowing, but no pulmonary infection could be diagnosed. Over the next weeks, respiratory distress worsened and oxygen therapy had to be implemented. Because of poor feeding, nutrition via nasogastric tube was started. CT scan of the chest demonstrated bilateral interstitial shadowing compatible with chILD. Lung biopsy and genetic blood testing enabled to diagnose ABCA3 deficiency. Meanwhile the child had severe respiratory distress and needed CPAP support with FiO2 up to 100%. Triple-therapy with

hydroxychloroquine, azithromycine and prednisolone was started, which resulted in gradual improvement of the pulmonary situation. The child could be discharded home at the age of 3 months without supplemental oxygen.
Conclusion/Discussion: Triple-therapy with hydroxychloroquine, azithromycine and prednisolone may offer a treatment option for infants with ABCA3 deficiency.

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Pulmonary artery sling – an unusual cause of respiratory failure

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Background: Pulmonary artery sling is a rare congenital condition in which the left pulmonary artery (LPA) originates from the right pulmonary artery (RPA), and passes between the trachea and the esophagus to reach the left lung, thereby forming a sling around the airway. It is often associated with intrinsic tracheal stenosis due to complete cartilaginous rings, and usually presents during the first months of life with progressive respiratory symptoms which can be fatal.
Case presentation: A 4-weeks-old boy presented with respiratory distress and difficulty feeding due to RSV bronchiolitis. The clinical course was marked by acute respiratory failure on the 2nd day in hospital requiring intubation. Blood gas analysis showed severe respiratory acidosis with a pH of 6.9, pCO_2 of 115 mm Hg and lactic acid of 7.7 mmol/l. Chest X-ray was compatible with postobstructive pulmonary edema, raising the suspicion of severe upper airway obstruction. Because of intermittent hampered mechanical ventilation, flexible bronchoscopy was performed. This examination led to the diagnosis of a long-segment distal tracheal stenosis. A chest CT scan confirmed this finding and demonstrated an aberrant origin of the LPA as seen with pulmonary artery sling. After establishing the diagnosis, the patient underwent surgical intervention, which consisted of reimplantation of the LPA, thereby eliminating the extrinsic compression of the airway, and concomitant slide tracheoplasty to enlarge the tracheal lumen. After a long hospitalization the patient is doing fine 2 years after the procedure with no respiratory symptoms. However, he presents a stenosis of the LPA and the RPA at the anastomosis and suture sites, which may require further intervention in the future.
Conclusion: The case illustrates a vascular malformation being the cause of respiratory failure in infancy. RSV bronchiolitis, the initial diagnosis of the patient, may cause severe affection of the respiratory system requiring mechanical ventilation. However, if the clinical course is unusual and not improving, this should raise suspicion of an underlying cause. Pulmonary artery sling, often associated with anomalies of the trachea, should be included in the differential diagnosis, as surgical treatment is curative of the affection.

P 55

Mucositis, skin lesions and dysuria – painful symptoms of Mycoplasma pneumoniae-associated mucositis (MPAM)

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Background: Mycoplasma pneumoniae infection is often associated with extrapulmonary disease. It may induce mucosal inflammation, referred to as Mycoplasma pneumoniae-associated mucositis (MPAM). The disease typically affects male adolescents and is characterized by oral, ocular and uro-genital lesions, and less often by skin lesions.
Case report: A 12-year-old boy was referred because of increasing oral pain with feeding difficulties, haematuria, dysuria and poor general condition. During the previous week, he had a febrile respiratory infection which was treated with co-amoxicillin by his pediatrician. On clinical examination, he showed impressive bilateral heamorrhagic conjunctivitis, swollen lips with enanthema, mouth ulcers, bullous skin lesions and genital ulcerations. Chest x-Ray was consistent with atypical pneumonia, and oxygen had to be administrated for 7 days. Urinanalysis was normal. PCR for mycoplasma pneumoniae was positive in nasopharyngeal secretions, and specific IgM to mycoplasma pneumoniae were detected in serum. Because of severe mucosal pain, intravenous nalbuphine therapy and urine catheterization were necessary. The boy was treated with azithromycin for 10 days. Neither systemic steroids nor immunoglobulins were administrated. The boy could be discharged home after 13 days of hospitalization.
Conclusion: Mycoplasma pneumoniae-associated mucositis, which belongs to the spectrum of erythema multiforme and Stevens-Johnson syndrome, can lead to severe and painful disease requiring prolonged hospitalisation.

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Abnormal findings in the lower respiratory system

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We describe the case of an 11 8/12 year old girl with abnormal findings in her lower respiratory tract. Based on predominant exercise intolerance with in- and expiratory stridor several tests were performed: Lung function detected a both restrictive and obstructive pattern with flattening of the expiratory part of the flow-volume loop. Allergy was excluded due to normal exhaled nitric oxide as a marker of eosinophilic inflammation of the airways and normal in vitro test for detecting common inhalative allergen-specific IgE. As a result of the biphasic stridor flexible endoscopy was organized subsequently. Endoscopy showed an abnormal anatomy of the bronchial tree with narrowing of the distal trachea and a hypoplastic left main bronchus. CT-thorax revealed an abnormal branching: What was thought to be the main carina represents the branching of the whole right upper lobe bronchus that originates from the trachea as a so called "pig lobe". The assumed hypoplastic left main bronchus disclosed as hypoplastic distal trachea. Additionally the right middle lobe was missing and a pulmonary artery sling was present. Treatment options are still in discussion including resection of the hypoplastic distal trachea and end-to-end-anastomosis.

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New reference values for N_2 multiple breath washout outcomes in pre-school and school-aged children

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Background: N_2 multiple breath washout measurements (N_2MBW) are widely used to assess ventilation inhomogeneity in pre-school and school-aged children. However, normal values for this age group do not exist so far.
Aim: The aim of this study was to generate reference values for N_2MBW outcomes in a big cohort of healthy Caucasian children from two centres.
Methods: 2–3 N_2MBW measurements were performed using the ultrasonic flowmeter (Exhalyzer D, Ecomedics, Duernten, Switzerland) in 96 healthy children (42 males, age range 5.9–18.1) from the University Children's Hospital of Bern, Switzerland and 42 healthy children (18 males, age range 2.8–17.2) from the Hospital for Sick Children in Toronto, Canada. Data were analysed with the newest version of the software provided by the manufacturer (Spiroware, 3.2.0, Ecomedics AG). Functional residual capacity (FRC) adjusted per weight and lung clearance index (LCI) are reported.
Results: 138 healthy Caucasian children aged from 2.8 to 18.1 years were included in this study. As expected, FRC increased with age, but LCI remained stable. Mean FRC per weight (SD) was 43.9 (9.8) ml/kg and mean LCI (SD) was 7.3 (0.6). The 95% limits of agreement for LCI were between 6.3 and 8.3. The between-centre comparison showed no significant difference both for FRC per weight (t-test, $p = 0.7$) and LCI (t- test, $p = 0.9$). Reference equations for FRC are given.
Conclusion: We report reference values for N_2 MBW outcomes in pre-school and school-aged children that can be used in clinical practice to define lung volumes and detect the size of ventilation inhomogeneity in pediatric respiratory diseases.

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Pulmonary embolism as a complication of deep leg vein thrombosis in a child without known risk factors

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Case report: C.A. 2004. A 10 year old boy was admitted to our emergency department with a 3-week history of lower leg pain and consecutive limp. No fever, B symptoms or trauma reported. The pain was reproducible on palpation; there was localized swelling just below the knee. An x-ray ruled out a fracture or neoplasm, slightly elevated blood inflammatory markers were found. The boy was discharged with NSAIDs and leg immobilization based on suspected muscular pain. In the follow-up the pain had almost subsided, but inflammatory markers remained high so that an MRI was performed, revealing slight edema around the neurovascular bundles of the thigh muscles. Two weeks later the boy returned with right-sided upper abdominal pain and dyspnea. The chest x-ray was unremarkable, the inflammatory markers stationary, an abdominal ultrasound revealed a thrombosis of the right common and external iliac vein. A chest CT confirmed the diagnosis: paracentral pulmonary embolism of the right lower and middle lobes with infarction of the lower right lobe. The boy was transferred to the University Children's Hospital Zurich, where he was treated with unfractionated heparin and discharged fully anticoagulated with a vitamin K antagonist 9 days after admission. In retrospect, a deep leg vein thrombosis was visible in the MRI.
Discussion: Despite its rareness the incidence of venous thromboembolism in the pediatric population is increasing, mainly in the in-patient hospital setting. The majority is associated with the presence of risk factors favoring the build-up of blood clots, especially central venous catheters, cancer, congenital heart disease, surgery, preterm birth and anti-phospholipid syndrome. Only five percent are spontaneous, as in our case report; in 60% of these cases a congenital thrombophilic disorder can be found. Today screening for congenital thrombophilia is not recommended in patients with secondary thrombosis or asymptomatic patients with a positive family history. Treatment includes the use of anticoagulant drugs such as heparins, coumarin derivates and rarely thrombolytic agents. Knowledge of the efficacy and safety of these treatments is limited by the lack of evidence-based trials in the pediatric population, warranting the need for further studies.
Conclusion: Consider venous thrombosis in persistent leg pain in an otherwise well-appearing child. Negative family history and absence of thrombophilic risk factors does not rule out the possibility of thrombosis.

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Magnesium in cystic fibrosis – systematic review of the literature

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Background: The metabolism of sodium, potassium and chloride and the acid-base balance are sometimes altered in cystic fibrosis. Textbooks and reviews only marginally address the homeostasis of magnesium in cystic fibrosis.
Methods: We performed a search of the Medical Subject Headings terms (cystic fibrosis OR mucoviscidosis) AND (magnesium OR hypomagnes[a]emia) in the US National Library of Medicine and Excerpta Medicadatabases.
Results: We identified 25 reports dealing with magnesium and cystic fibrosis. First, hypomagnesemia affects more than half of the cystic fibrosis patients with advanced disease; second, magnesemia, which is normally age-independent, relevantly decreases with age in cystic fibrosis; third, aminoglycoside antimicrobials frequently induce both acute and chronic renal magnesium-wasting; fourth, sweat magnesium concentration was normal in cystic fibrosis patients; fifth, limited data suggest the existence of an impaired intestinal magnesium balance. Finally, stimulating observations suggest that magnesium supplements might achieve an improvement in respiratory muscle strength and mucolytic activity of both recombinant and endogenous deoxyribonuclease.
Limitations: The main limitation of the present systematic review is that it was based on the scanty literature available. Furthermore, we were not able to collect individual patient data to calculate the influence of age on circulating magnesium level.
Conclusions: The first comprehensive review of the literature confirms that, despite being one of the most prevalent minerals in the body, the importance of magnesium in cystic fibrosis is largely overlooked. In these patients, hypomagnesemia should be sought once a year. Furthermore, the potential of supplementation with this cation deserves more attention.

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Foreign body aspiration and ingestion in children in a Swiss tertiary paediatric emergency department. A retrospective, observational study

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Background: Ingestion and aspiration of foreign bodies (FB) is a common problem in the pediatric emergency department (PED) and can be fatal. We aimed to analyse the occurrence of FB ingestion and aspiration in a Swiss tertiary PED.
Methods: We conducted a chart review of all children ≤16 years with FB aspiration or ingestion seen in our PED from August 2013 to January 2016.
Results: Ingestion occurred in 147 (23 non-accidental) and aspiration in 42 patients, respectively; 97 males (56%) and 77 females (44%) aged 15 days to 15 years (mean = 4.5 years). The majority of incidents occurred in children 0–3 years of age (n = 108, 62%), whereas intentional intake was seen in children ≥12 years only. Causative agents for ingestion included undrinkable liquids (n = 23), magnets (n = 11), coins (n = 11), batteries (n = 8); and jewellery (n = 8), apples (n = 10), nuts (n = 5), carrots (n = 4) and other (n = 23) for aspiration, respectively. Symptoms from ingestion included vomiting (n = 27, 18%), retching (n = 21, 14%), dysphagia (n = 26, 18%), hypersalivation (n = 11, 7%) or none (n = 58, 44%); those from aspiration included cough (n = 31, 74%), retching (n = 10, 24%), respiratory distress (n = 23, 55%), cyanosis (n = 5, 12%) and only 2 patients were asymptomatic. Chest or abdominal X-ray was required in 46 subjects (31%) with ingestion and in 16 (38%) with aspiration, in each case; upper gastrointestinal tract fluoroscopy in 6 (14%). Fiberoptic laryngoscopy was performed in 6 patients, however, without location of a FB in any child. 10 subjects underwent surgical removal of oesophageal FB in 10 (7%), 16 (25%) gastroscopy to pick magnets (n = 4), glass shards (n = 1) and yew needles (n = 13); secondary laparotomy was required in 2 cases and bronchoscopy was indicated in 24 (57%); but, in only 9 subjects a FB was removed (right lung (n = 6), left lung (n = 2), trachea (n = 1)). Prolonged clinical observation >6 h was needed in 66 of all FB cases (35%); complications occurred in only 3 ingestions (oesophageal corrosion n = 2, small intestine necrosis, n = 1) and one aspiration (acute respiratory distress syndrome (ARDS) with pneumothorax).
Conclusion: Aspiration and ingestion of FB remains a frequent presenting problem, especially in children aged 0–3 years. We suggest, physicians should be aware of the most dangerous objects and their sequelae, such as ingestion of several magnets or battery in the oesophagus. Then management requires urgent specialty expertise to ensure quick removal and thus, prevent serious complication.

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Increasing respiratory distress in a child with cystic fibrosis

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We present the case of a 8-year old female with cystic fibrosis (CF), admitted in our hospital with increasing cough and dyspnea. She was diagnosed with a homozygote mutation F508del at the age of 4 months after failure to thrive. Her airway cultures were positive for staphylococcus aureus and haemophilus influenzae, last detection of pseudomonas aeruginosa was at the age of 3 years. Her first second forced expiratory volume (FEV_1) two weeks before was 91% of predicted. On day of admission she presented with tachypnea of 60/min, deep retractions and strong wheezing on auscultation, oxygen saturation was 86%. Laboratory results showed CRP 16 mg/l and balanced blood gas analysis. Chest X-ray showed only bronchial wall thickening. Suspecting a virus induced pulmonary exacerbation she was prescribed oral steroids, intensified inhalation with salbutamol and ipatropiumbromid, oxygen supplementation and antibiotics. The following day she developed a silent chest with increasing dyspnea and oxygen need. Despite CPAP on intensive care unit she developed respiratory insufficiency, bilateral pneumothoraces, pneumomediastinum and soft tissue emphysema. She was then mechanically ventilated and pneumothoraces were treated with chest tubes. On the first day total serum IgE was 368 kU/l. We repeated the

measurement and found IgE to be highly elevated at 3844 kU/L. Airway cultures showed staphylococcus aureus and aspergillus fumigatus. Along with the clinical deterioration and the radiologic changes these results indicate an allergic bronchopulmonary aspergillosis (ABPA). We increased intravenous steroids and continued antibiotherapy. She was dismissed with 2.5 mg/kg steroids. The evolution was good, after 6 weeks, FEV1 was 75% and 3 months later 98% of predicted. In CF, 1–15% of patients may develop ABPA. Therefore the possibility of ABPA should always be considered in pulmonary exacerbations particularly with poor response to conventional treatment.

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Introducing liaison psychology within a paediatric unit dealing with diabetic patients: "a pilot experiment"

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The paediatric unit dealing with diabetic patients of the eastern part of canton Vaud was set up in March 2010 within the healthcare network of the Haut Léman region, the Réseau Santé Haut-Léman. In 2014, this unit approached the child psychiatry unit of the Fondation de Nant, the mental health service provider of the eastern part of canton Vaud. Together both units shared observations and considerations relating to the care given to young diabetic patients and their families. Our exchanges were specifically focussed on addressing the emotional needs of patients and families faced with a chronic illness. A pilot experiment was set up further to these exchanges involving liaison psychology within the paediatric team. As of February 2015, one of the clinical psychologists working within the child psychiatry unit joined the paediatric team on a part-time basis. After 10 monthes of intense collaboration, the results surpass the expectations of both the paediatric and the child psychiatry units. Access to psychological counselling has so far had a positive impact on treatment compliance and on family environment. Liaison psychology has also proved effective within the team in the global understanding of the diabetic patient. Depressive signs & symptoms related to decrease of compliance to treatment have been observed in a number of cases and allow us to formulate new hypotheses on the impact of adolescence on the management of diabetes.

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Reaching the Limits of Immunomodulatory Therapy in a Patient with Systemic JIA; a Case Report

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New immunomodulatory therapies have emerged over the past years that have greatly improved success in autoimmune disease treatment. We present a case of refractory systemic JIA (sJIA) that finally is in a relatively steady state after having exhausted all new immunomodulatory drugs, thus illustrating benefits but also limitations of therapeutical options for sJIA available today. An 11-year-old girl presented with typical signs of sJIA (recurring fever over 2 weeks, typical rash, splenomegaly) and was initially treated with oral steroids and NSAR. One month after stopping the steroids we saw reactivation of the sJIA now with gonarthritis and started therapy with methotrexate (MTX) and steroids. The steroids were again tapered and then stopped. One month after therapy solely with MTX we saw a third reactivation of sJIA, so we initiated therapy with Tocilizumab and steroids. This therapy did not show the desired effect, symptoms such as fever, rash and arthritis persisted, so the medication was changed to Canakinumab. This was also insufficient, 10 days after the first injection the patient showed high fever and arthritis, which continued after the second injection that was performed early (after 24 days), so the medication again was changed to Anakinra. The initial dose of 40 mg/d was eventually increased to 200 mg/d after two steroid pulse therapies had been performed. The first because of persistent signs of systemic inflammation and the second pulse one week later because of macrophage activation syndrome. At this point we also extended the therapy with Cyclosporine. Today, 4 months later, the patient is in a comparatively stable state under Anakinra, high dose Prednisolone and Cyclosporine, but still showing regular signs of reactivation after slight colds or immunisations, so that tapering of prednisolone is proving very difficult. This exemplary case illustrates a two-year search for effective therapy in a patient with sJIA, the whole spectrum of current therapeutic possibilities as well as their limitations.

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Simulate where it hurts – development of a multidisciplinary, interprofessonal in-situ simulation-based team training for the management of paediatric trauma in a tertiary swiss adult emergency department

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Background: At our institution, severely injured children require admission to the adult emergency department (ED), where sectional imaging and low dose X-ray (LODOX) are readily available. However, multiple challenges remain for optimally ensuring patient safety with a consistently alternating trauma team consisting of adult and pediatric physicians not routinely workingtogether.
Methods: To address this issue, we implemented regular in-situ simulation-based multidisciplinary, interprofessional team training into routine clinical practice. From January to December 2015, nursing and medical staff conjointly participated in our quarterly half-day scenario-based program. We aimed to evaluate the impact of simulation on the preparedness for paediatric trauma by a post-course survey.
Results: Twenty-eight participants (5 nurses, 21 physicians, 2 medical students) from adult and pediatric emergency medicine and pediatric surgery completed the survey; 23/28 had previous simulation experience; 13/28 advanced trauma life support (ATLS) provider certification. Realism was felt to be very strong by 10/28 participants. Half of the attendees reported to have gained significant new medical (n = 15/28) and crisis resource management (CRM) knowledge (n = 14/28), respectively. Four in 5 attendees very strongly thought that simulation was an effective teaching tool. General feedback included the request for more frequent team trainings tailored to the real situation, increased team thinking and self-confidence.
Discussion: Patient outcome strongly depends on medical and non-technical skills of a multidisciplinary body. Implementation of continuous team training for all personnel involved in the acute management of critically injured children in our institution increases team preparedness and self-confidence. The value of in-situ simulation-based team training in realistic circumstances is highlighted.

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Impact of interventions to increase the proportion of medical students choosing a primary care career: A systematic review

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Background: Increasing the attractiveness of primary care careers is a key step in addressing the growing shortage of primary care physicians, including general pediatricians. The purpose of this review was to (1) identify interventions aimed at increasing the proportion of undergraduate medical students choosing a primary care specialty, (2) describe the characteristics of these interventions, (3) assess the quality of the studies, and (4) compare the findings to those of a previous literature review within a global context.
Methods: We searched MEDLINE, EMBASE, ERIC, CINAHL, PsycINFO, The Cochrane Library, and Dissertations & Theses A&I for articles published between 1993 and February 20, 2015. We included quantitative and qualitative studies reporting on primary care specialty choice outcomes of interventions in the undergraduate medical curriculum, without geographic restrictions. Data extracted included study characteristics, intervention details, and relevant outcomes. Studies were assessed for quality and strength of findings using a five-point scale.
Results: The review included 72 articles reporting on 66 different interventions. Longitudinal programs were the only intervention consistently associated with an increased proportion of students choosing primary care. Successful interventions were characterized by diverse teaching formats, student selection, and good-quality teaching. Study quality had not improved since recommendations were published in 1995. Many studies used cross-sectional designs and non-validated surveys, did not include control groups, and were not based on a theory or conceptual framework.
Discussion: Our review supports the value of longitudinal, multifaceted, primary care programs to increase the proportion of

students choosing primary care specialties. Isolated modules or clerkships did not appear to be effective. Our results are in line with the conclusions from previous reviews and add an international perspective, but the evidence is limited by the overall low methodological quality of the included studies. Future research should use more rigorous evaluation methods and include long-term outcomes.

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IgG4-related abdominal inflammatory pseudotumor: case report and overview of the diagnosis

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Case report: We report the case of a previously healthy 15 year old boy who presented with acute flank pain. Clinical investigation revealed an abdominal mass. Ultrasound and magnetic resonance imaging confirmed a large polylobulated tumor in the mesenterium. Laboratory investigations showed elevated inflammatory markers (CRP 160 mg/l, ESR 75 mm per hour). An ultrasound-guided needle biopsy of the lesion was performed. The histological result was consistent with the diagnosis of a fibroinflammatory tumor. We therefore additionally measured the patient's IgG4 level, which turned out to be substantially increased. Altogether, these findings were highly suggestive of an IgG4-related calcifying fibrous pseudotumor. Due to the tumor's size and its clinical symptoms, the indication for a surgical tumor removal was given. Postoperative histologic workup confirmed the preoperative diagnosis.
Conclusion: IgG4-related disease is an emerging and possibly underdiagnosed group of disorders. Our poster provides an overview of the characteristic clinical, laboratory and histological findings, as well as the diagnostic and therapeutic features of this rare and still largely unexplored entity.

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