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Be careful with exophthalmos

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Histiocytosis is a tumor-like condition, frequently seen in children between 1 and 15 years. However, the etiology is poorly defined. One hypothesis suggests that it may be caused by an alteration of the Langerhans cell by a somatic mutation or a viral infection at an early stage of development .The cell accumulates abnormally and produces proinflammatory cytokines responsible for the organ and systemic injury. Localization: 80% skeleton, 60% skin, 33% lymph nodes, 30% hepato-renal, 25% lung, 20% CNS. The final diagnosis is obteined by histological spacement. G.B., a 3 year old child, was hospitalized with bilateral exophthalmos prevalent on the left side. Blood tests normal, thyroid function normal. No ophthalmological or neurological symptoms were observed. MRI highlighted high cell density neoformation, involving skull base bones and both orbits. The CT guided biopsy confirmed the diagnosis of Langerhans cell histiocytosis. This case serves to remind us to take care with diagnosis when sympotoms are indicative of a particular illness. Indeed, exophthalmos should be treated with care, above all in children. Histiocytosis also should receive special consideration because it affects a large number of organs and systems and enters in the differential diagnosis of many clinical conditions.

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Subdural Hematoma and external hydrocephalus as clinical presentation of rare factor XIII deficiency in infant

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A 5 months baby was brought to our service due to vomiting, lethargy, poor reactivity and bulging fontanelle. On physical examination a blueish color of eyes was noticed, suggestive of osteogenesis imperfecta. Her medical history disclosed only a slight prematurity at birth (34 weeks) and left hypoacusis documented at hearing tests. Emergency brain ultrasounds and scans revealed external hydrocephalus. A brain NMR documented diffused subdural hematomas with varying evolutions. Retinal hemorrhages were noticed by the ophthalmologist. Neurosurgeons applied external drainage devices obtaining evacuation of clear LCR, resolution of hydrocephalus and rapid clinical improvement. A careful follow-up was performed until complete resolution of subdural hematomas. A defect in hemostasis was suspected, so a complete examination of the baby's coagulation was performed, disclosing a significant reduction of fXIII (29%). The same protein was found to be slightly reduced in her mother (63%). On suspecting osteogenesis imperfecta, a radiological assessment was performed, revealing no bone anomalies. Finally, an analysis of genes COL1A1 and COL1A2 was ordered which is still under evaluation. In conclusion, coagulation defects need to be excluded in infants presenting with intracranial hemorrhages, even if a deficit of fXIII and vWf is rare; the possible association with bone anomalies like Osteogenesis Imperfecta can cause the extension of hemorrhages and influence the clinical trend to healing.

####

A case of atypical persistent vomiting

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Vomiting in the neonatal period can be a sign of several pathologies not exclusively of gastrointestinal origin. D.C. was born by spontaneous delivery, 3200 g, Apgar 9-10, and presented several vomiting episodes in the first few hours of life. The first spontaneous emission of meconium occurred after 12 hours and in the following hours just after stimulation. Laboratory tests and eco abdomen were normal. Vomiting per-

sisted even with continuous nasogastric feeding. The abdomen X-ray even with contrast media, was negative. Enteral feeding with a hydrolyzed protein formula did not change the clinical situation. In the 5th day of life she had phototherapy, which was suspended after 4 days. From the 7th day there were signs of torpor and feeding rejection with apparent macroglossia not present in the early days. Waiting for outcome screening, the thyroid function was evaluated: 303 TSH mUI/L,fT4 0.7 ng/dl. Ultrasound and scintigraphy demonstrated thyroid agenesis. The femoral distal epiphyses X-ray showed bilateral Béclard nucleus. Anti TSH receptors antibodies (TRBAb), performed for previous maternal positivity, were negative. At the moment the baby is six months old and presents normal neuropsychological and auxological development. Conclusion: although hypothyroidism screening is carried out, suggestive clinical signs, such as vomiting sine causa, necessitate a thyroid function check in the first days of life. This strategy allows for the initiation of replacement therapy earlier, with long-term benefits.

####

Results of the application of WHO guidelines for malnutrition in children in a rural district hospital of Burkina Faso

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Malnutrition is one of the most important underlying causes of mortality in children under 5 years of age in the developing world. The aim of this study, carried out in a rural district hospital of Burkina Faso, was to evaluate the implementation of the World Health Organization (WHO) guidelines for the treatment of children with complicated acute malnutrition in an area with a high prevalence of co-morbidities. Between April 2008 and February 2012, 615 children were hospitalized and treated according to the WHO guidelines; 63,6% having complicated severe acute malnutrition (MAS) and 36,4% having complicated moderate acute malnutrition (MAM). Frequency of co-morbidity, mainly malaria, was very high (91,1%). Mean duration of hospitalization was 14,6 days. At discharge, 81,1% of children were improved or recovered; the mortality rate was 11,7%. After discharge, children were enrolled as outpatients; the percentage of children lost at follow up was very high especially during the rainy season and increased from 44% at first follow up to 78,9% at the eighth follow up. Our results demonstrate the effectiveness of WHO guidelines for inpatient treatment of children with complicated acute malnutrition also in areas with a high prevalence of co-morbidities and highlight the need for new outpatient treatment strategies. Therefore, in April 2012 we started production and free distribution of enriched flour made from local ingredients, prepared together with the mothers of malnourished children.

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Precocious puberty related to hamartoma and glioma

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M., 8 yrs 5 mths, presented at our clinic for larche and pubarche since age 8. Her previous history was not remarkable. Height had been >97°c since the age of 2, MPH was 75°c; pubertal stage PH2, I1, B3; clinical and neurological exams were normal; bone age: 11 yrs. GnRH test: LH (+30'): 3.4 mIU/mL, FSH (+90'): 9.8 mIU/mL. E2: 37 pg/mL. Pelvic ultrasonography: uterine length was 49 mm, right and left ovarian volume were 2.6 mL. Treatment for central precocious puberty (CPP) (triptorelin 2.5 mg every 28 days) was initiated and a brain MRI was performed which showed a optic-chiasmatic glioma and hamartomatous component possibly related to NF1. Ophthalmologic evaluation was regular. A neurosurgery specialist recommended repeating MRI two months later; it showed unchanged findings. No other criteria for diagnosis of NF1 were fulfilled. The prevalence of CNS lesions in presence of CPP is lower in females (5%), especially after the age of 6 and the male-female ratio is 2:1; they are represented mainly by hypotha-

lamic hamartomas and, in minor portion by optic gliomas. Treatment with GnRH-agonists and neuroradiological follow-up is the first choice for these patients while surgical removal should be reserved to selected cases. This case shows the importance of neuro-radiological investigations in CPP also when age, sex and symptoms onset do not suggest organic causes.

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Psychogenic polydipsia in a child of 16 months

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R., 16 mths old, presented at our clinic for polydipsia (up to 3 L/day) and polyuria. Her previous history was unremarkable except for upper respiratory tract infections and a HHV6 infection at the age of 1. Physical examination showed a good condition and hydration status and normal neurological findings. Weight was 10.2 kg (40°c), height was 80.5 cm (79°c); she showed no weight gain since 11 mths of age. R. was hospitalized in order to evaluate water balance and perform further investigations. She was allowed to drink water ad libitum. The daily fluid intake and urine output were 1500 ml/day (3 L/m²/day), confirming polydipsia and polyuria. Serum osmolality was 267 mOsm/kg, urinary osmolality was 166 mOsm/kg and urine specific gravity was 1.011. Other tests, including blood glucose, urine glucose and HbA1c, were normal and DMT1 were excluded. Because of her age, we chose not to perform a water-deprivation test and we ordered the gradutal reduction of fluid intake, transitioning from a baby-bottle to a cup. Two months later, serum osmolality was 268 mOsm/kg and urinary osmolality had increased to 338 mOsm/kg. Clinical examination was normal, she showed weight gain (11.3 kg, 56°c) and liquid intake reduced to 500 mL/day. On suspicion of psychogenic polydipsia we opted for strict follow-up and delayed a decision to perform further investigations.

####

Breastfeeding in the Mediterranean area: comparison with the WHO guidelines.

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Exclusive breastfeeding until month 6 and continued over the first year of life, in tandem with the weaning process, is important for a number of reasons. It prevents a number of conditions in children (i.e. obesity and allergies) maintains good nutrition, influences the immune system, and delays the occurrence of symptomatic disease (i.e. celiac disease). In the Mediterranean area, where breastfeeding had always been favored as a result of cultural and religious tradition (especially in the Islamic populations), a change has been seen in the few last decades. Firstly, in the economically developed countries (the European side) habits started to differ significantly from the WHO guidelines and later on the African side there has also been a progressive decline in the exclusive breastfeeding until month 6 . In comparison with the ideal (100% prevalence rate) and the achievable condition (close to 85%) in Mediterranean countries the prevalence rates are under 50% (see figure). Among them, Italy ranks in the last position; only partially mitigated by the observed rates of 35% in those regions where investment in an educational program on breastfeeding is quite strong. Investments in educational programs on breastfeeding are of high priority in the Mediterranean area both in Europe and in Africa.

####

Blueberry Muffin Baby Syndrome: a case report

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A female baby was born at term after an uncomplicated pregnancy. A few minutes after the delivery numerous brownish-red macular, papular and nodular lesions, ranging from 2 to 10 mm in diameter, appeared over the trunk, abdomen and lower limbs suggesting a diagnosis of "blue berry muffin baby syndrome ". The

rest of the physical examination and the laboratory tests (serological tests included) were negative. Chest and skeletal X-ray, abdominal and CNS ultrasound were normal. The skin biopsy revealed dermal histiocyte aggregates positive for S100 and CD1a. With these findings the diagnosis of congenital self-healing reticulohistiocytosis of Hashimoto-Pritzker was made. The baby was then discharged with a follow up plan. In the presence of "blue berry muffin baby syndrome" the differential diagnosis should consider: congenital infections (Cytomegalovirus, TORCH, Coxsackie, Epstein Barr, Syphilis), haematological disorders (spherocytosis) and neoplastic disorders (histiocytosis, congenital leukaemia, neuroblastoma, rhabdomyosarcoma). Self-healing reticulohistiocytosis is a benign form of Langerhans cell histiocytosis (LCH) characterized by the presence, usually in the neonatal period, of cutaneous lesions, which generally disappear within the first months. The absence of sistemic involvement at diagnosis and during the next follow up permits the distinction of this benign and self-healing form from the others types of LCH.

####

Antenatal combined therapy of an autoantibody-related congenital atrioventricular block: a case report.

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Autoantibody-related congenital atrioventricular block (CAVB) is an autoimmune condition in which transplacental passage of maternal autoantibodies causes damage to the developing heart conduction system of the foetus. We report a case of a 31-year-old woman, in a good clinical status, referred to our center in February 2013 during the 26th weeks of her first pregnancy after detection of bradycardia in a fetal ultrasonography. An echocardiography revealed a 3rd degree CAVB with an atrial rate of 154 bpm and a ventricular rate of 54 bpm, without any anatomical defects that structurally displace the heart conduction system. Anti-Ro/La-related CAVB was suspected, despite the mother being asymptomatic for autoimmune disease, detection of maternal anti-extractable nuclear antigens antibodies was performed. A high title of maternal anti-Ro/SSA was found (359,5 U/ml) and diagnosis of an autoantibody-related CAVB was made. A combination treatment protocol of oral betamethasone (4mg/day), plasmapheresis (weekly session) and IVIG (1gr/kg fortnightly) was begun and continued until the delivery date, to diminish the inflammatory response in the fetal conduction system and to lower the serum levels and placental transport of maternal autoantibodies. At birth, IVIG session (1gr/Kg fortnightly) for neonate was scheduled until maternal antibodies became undeletable. This therapy is known to be effective in reversing a 2nd degree CAVB but its benefit in treating 3rd degree CAVB remains to be proven.

####

A case of Macrophage Activation Syndrome in Cystic Fibrosis.

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We describe the case of a boy, affected by Cystic Fibrosis (CF), age 10.7 yrs, FEV1 in good health 60%, chronically colonized by Methicillin-susceptible Staphylococcus aureus (MSSA). In February 2011, the patient underwent intravenous antibiotic therapy for respiratory exacerbation. A week after onset he presented fever unresponsive to acetaminophen, alteration of consciousness, emathemesis and melena. Blood tests performed showed: PLTs 47000/mm3, ferritin 76150 ng/dl, LDH 7863 U/L, AST 642 U/L, aPTT 91.4 s, PT 21%, Fibrinogen 88 mg/dl. These first findings suggested an initial septic shock, so we changed intravenous antibiotic therapy: ciprofloxacin (30 mg/kg/die), amikacin (20 mg/kg/die), teicoplanin (10 mg/kg/die) and oseltamivir (120 mg/day for one week) after throat swab result (Real

time PCR positive for AH1N1 Virus). Were also performed: blood culture (negative), chest x-ray (thickening area in basal left lung), EGDS (evidence of fund hemorrhagic gastritis); abdominal ultrasound, x-ray brain and MRI were normal. These findings suggested a diagnosis of Macrophage Activation Syndrome (MAS) and we started intravenous therapy with prednisone for 15 days, followed by gradual tapering. In a week we observed resolution of clinical features with a complete normalization of macrophage activation marker. Currently the patient has stable respiratory conditions (last FEV1 64% pred).

####

Refetoff Syndrome associated to common variable immunodeficiency and psoriasis: a case report

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We describe the case of a 17-year-old Brasilian boy with common variable immunodeficiency and psoriasis who presented high levels of thyroid hormones with positivity for anti-TPO antibodies and normal values of TSH after TRH-stimuli test. The thyroid echography showed the presence of two nodules in the left lobe of the gland. The thyroid scintigraphy with 99-MTC-pertechnetate reported: "Gland in situs, slightly dysmorphic, with normal distribution of the tracer". So the genetic study of thyroid hormones receptors was started through PCR and sequencing of the coding regions and intron-exons junctions of the THRB gene. The results showed the presence, in the exon 9 of the THRB gene, of the M313T mutation due to a replacement cytosine-threonine (ATG-ACG) in etherozygosis. In addition, an unusual polymorphism rs13063628 was found in etherozygosis in position IVS9+9bp. The molecular genetic response is compatible with Refetoff syndrome, a very rare syndrome characterized by a mutation of the THRB gene responsible for a generalized resistance to the T3 nuclear receptor and sensorineural deafness. It can be associated to autoimmune diseases like vitiligo, autoimmune thyroiditis and Graves' disease. The association with common variable immunodeficiency and psoriasis has never been described in literature before, and for this reason it is necessary to research not only autoimmune diseases, but also umoral and cellular immunity disorders in all patients with Refetoff syndrome.

####

Agenesis of corpus callosus: a case report

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Corpus callosum is the main neocortical commissure of the cerebral hemispheres. Agenesis of corpus callosum (ACC) is a rare congenital disease. It can be complete or not, isolated or related to brain malformations, genetic syndromes or metabolic disorders. The karyotype is altered in 20% of cases. The diagnosis is made by ultrasound: colpocephaly of the occipital horns of lateral ventricles (LV), dislocation at the top of 3°ventricle, approachment of the frontal horns of LV ("bull-horn"), cerebral grooves radially around the roof of 3° ventricle. Case report: A.V. was born at the gestational age of 35 weeks by cesarean section in breech presentation. Pregnancy course was regular, no prenatal exposure to toxins reported and maternal serology for TORCH was negative. Apgar score: 7I-9V, weight: 2650g. At 30 weeks the obstetric ultrasonography showed ventriculomegaly. Physical examination at birth shows craniofacial asymmetry and mild axial hypotonia. Brain ultrasonography showed ACC aspect "bull-horn" of LV, absent cingulate gyrus and related structures, bilateral ventriculomegaly, colpocephaly of occipital horns. The MRI confirmed ultrasonographic data; the EEG was regular. The ultrasonographic screening excluded extracranial malformations. The karyotype and screening for metabolic disorders were normal. Neurological development is appropriate to baby's age at the age of 24 months.

####

Abdominal pain and migraine: abdominal migraine?

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A.F. is a child of 8 years and 7 months who came to our observation for recurrent abdominal pain. By the age of two years he had suffered from recurrent episodes of intussusception for which he had been subjected to two surgerical interventions. From the age of three years, this problem was no longer present; however, he began to complain of recurrent debilitating, long-lasting abdominal pain which never occurred at night. These pains appeared to be more pronounced during the summer and were non dependent on meals. The patient has also recently reported the occurrence of migraine, although there is no family history of this problem. The physical examination and blood tests did not show anything abnormal. Abdominal ultrasound showed a significant thickening of the loops of the ascending and transverse colon, with negative calprotectin. AF has also undergone a radiological examination with barium contrast medium, which confirmed the absence of structural and functional abnormalities and a normal EEG result. Because of the clinical characteristics of pain and its association with the headache, and negative investigations excluding organic causes of abdominal pain, abdominal migraine was suspected; therefore, the patient started therapy with flunarizine. The symptoms reported during therapy and after its suspension will allow us to confirm the diagnostic hypothesis.

####

An annoying itch of the face and the arts

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DMN, 4 years 6 months, was admitted to our facility for diagnostic investigations upon the appearance of pruritic dermatitis of the face and limbs, from the age of 4 months, initially defined as atopic dermatitis. Prick tests were performed for food: results negative, and environmental allergens: positive results for D.Pteronissus, D.Farinae, Grasses mix, the dosage of total IgE showed a value above normal (210 KU / L vn <70 KU / L), the RAST to foods and inhalants were normal. Therapy with Advantan, Zirtec, Bentelan, Gentalyn Beta was prescribed, with which there has been a clinical improvement. Given a recurrence of the stympotoms and the family history of celiac disease (maternal uncle), dermatitis herpetiformis was suspected. Serology for celiac disease was performed, that was negative and duodenal biopsy showed a normal duodenal mucosa, incisional biopsy of the skin lesion, also excluded dermatitis herpetiformis. DMN was diagnosed with papular urticaria comfirmed by the value of total IgE, the positive allergy tests together with a compatibility of histology and site of the lesions. Therapy with a 1st generation antihistamine, Fucicort Aloe and Zinc has been prescribed and as well as the treatment of the home environment with pyrethroids.

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Sickle cell dactylitis: a case report

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Nanoro, Burkina Faso. D. ,a 2 year-old child, came to our clinic due to symmetric swelling of the third and fourth fingers of both hands. At physical examination, fingers were hot, painful, with purulent exudate pouring out of the incisions made at home by her father with therapeutic purpose. The general examination showed conjunctival pallor and mild splenomegaly. The blood count showed anaemia (Hb 7g/dl) and neutrophilia. We also decided to do a hemoglobin electrophoresis om suspicion of drepanocytosis. While waiting for test result, due to the onset of fever, we started her on broad-spectrum antibiotics along with analgesic treatment and hydration. Nevertheless, after a slight improvement, on the fifth day the patient showed a rapid deterioration, with sudden, massive spleen growth. Suspecting an acute splenic

sequestration crisis, we asked for a blood unit for urgent transfusion; unfortunately, the evolution was so fast that the patient died before having the transfusion done. Some days after the death, the electrophoresis result showed SS homozygous pattern, confirming the clinical suspicion of sickle cell anaemia. Acute dactylitis is frequently the first sign of drepanocytosis in the first years of life, and it is associated with a negative prognosis. This case was worsened by the onset of an acute splenic sequestration crisis, one of the most dangerous complications of sickle cell anaemia.

####

Wheat related disorders: beyond celiac disease.

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Wheat is one of the most widely consumed food grains in the world. The introduction of gluten-containing grains may create the condition for different diseases: wheat allergy (WA), celiac disease (CD), and gluten sensitivity (GS). We report three different cases. A 21 month old boy referred to us with irritability, loose and frequent stools, abdominal pain and failure to thrive. Serological tests revealed an IgA deficit, normal level of IgAanti-tTG and IgAanti-AGA, positivity of IgGantiAGA and high IgGanti-tTG. HLA-DQ2/8 was detected in genetic evaluation. CD was confirmed by a small intestinal biopsy. A 16 year old girl referred to us with an episode of urticaria, after wheat ingestion, during physical activity. Wheat skin prick test and wheat radioallergosorbent test resulted positive. On the suspicion of wheat-dependent exercise induced anaphylaxis and to avoid the risk of an oral challenge, IgE to recombinant ω5-gliadin was performed with a positive result. WA diagnosis was confirmed. An 11 year old boy referred to us with weight loss, abdominal pain, bloating, vomiting, diarrhea and extra-gastrointestinal symptoms after wheat ingestion. During a previous visit both allergic and autoimmune mechanisms were ruled out. A gluten free diet (GFD), followed by a positive blind placebo/gluten challenge, confirmed GS. All patients responded well to the GFD. As illustrated, based on a combination of clinical, biological, genetic and histological data, it is possible to differentiate these diseases.

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A case of pelvic pyomyositis

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Pyomyositis (PM) is an acute pyogenic infection of skeletal muscle common in tropical countries. Children and young adults are most commonly affected (M/F: 2/1). The development of PM is assumed to be secondary to traumatic injury to a muscle during a period of bacteriemia. Staphylococcus aureus is the most common causative agent followed by Streptococcus pyogenes, Enterococcus faecalis and Neisseria gonorrhoeae. The diagnosis is based on clinical features and haematological and radiological investigations. Magnetic resonance imaging (MRI) is considered the gold standard in making a diagnosis. A 9-year-old girl was referred to our hospital with fever and sever left hip pain radiating to the thigh. The pain worsened to the point that walking became difficult. Laboratory findings revealed a neutrophilic leukocytosis and an increase in CRP (18.1 mg/dl, normal range <0.6) and pro-calcitonin (0.37 ng/ml, normal range <0,05). Ultrasound and radiographic studies of the hip were negative. MRI showed an enlarged left iliacus muscle with a high signal on the the T2-weighted images. Therapy with teicoplanin and cefotaxime was initiated with rapid normalization of inflammatory markers and clinical improvement. Early diagnosis of PM is important and it should be differentiated from other severe conditions such as the septic arthritis of the hip; late therapy can result in serious complications.

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Headache with vomiting in infancy

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C., 2.5 years old, came to our Emergency Department (ED) because of recurrent episodes of frontal headache and morning vomiting for 7 days. Family history was positive for migraine with aura and Meniere syndrome. On suspicion of a symptomatic headache, we perform blood tests which showed normal results and fundus oculi (FO) examination showed evidence of prominent optic disc. Brain CT scan and MRI were normal. Clinical conditions of C. improved rapidly with normalization of FO, so we decided not to treat. One year later, C. came back to our ED with a recurrence of symptoms; we performed blood tests again and FO exam showed optic disc swelling and hyperemia. Brain MRI showed expansion of the perioptic sheath. During hospitalization, C. still presented recurrent headache and vomiting, especially in the morning, poorly responsive to analgesics and antiemetics drugs. Because of her age, on suspicion of benign intracranial hypertension, we decided not to perform lumbar puncture and to start treatment with acetazolamide (250 mg to 375 mg/day). The little girl's conditions improved gradually and symptoms disappeared in about 5 days. Benign intracranial hypertension is a syndrome characterized by headache, papilledema, high pressure and normal composition of cerebrospinal fluid, in the absence of neuroimaging abnormalities and other identifiable causes. The incidence is about 0.9 /100 000 and increases in obese adolescents. This case is remarkable because of age of onset and absence of overweight.

####

The year of the snake is coming

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Poisonous snake bites are medical emergencies that require immediate intervention. Approximately 2.5 million cases of snakebite are observed in the world every year, more than one million in Sub-Saharian Africa, but their true incidence is underestimated. In particular in Burkina Faso, the registered number of admissions for snake bites in 2008 was 16259. Italian data are scarce and they show an incidence of about 250 bites/year. Three species of snakes are responsible for fatal poisonings worldwide: Elapidae, Vipers and Cobra. Their venom contains protease and toxins that cause local symptoms such as edema, pain and gangrene and/or systemic complications such as neurotoxic syndromes and serious coagulopathies. The only specific treatment for snakebite is the administration of the specific antivenum, in association with symptomatic supportive care. The aim of our study is to describe the epidemiological and clinical data and to compare the diagnostic and therapeutic approaches to snake bites in two different locations. We retrospectively studied the cases of snake bite observed from January to December 2012 in the paediatric population (<15aa) in a rural area of Burkina Faso - Saint Camille Hospital Nanoro – (16, 11M and 5F) and at the 'Meyer Hospital' in Florence (2, 2M). 10/16 (62.5%) of the cases observed in Burkina Faso presented a grade 2 severity, and 6/16 (37.5%) presented grade 3 (50% severe neurological symptoms). 2/2 Italian cases had severity 2. Both Italian children received monovalent serum antitivenum, while in African children the polyvalent serum was administered in 56% (9/16) of cases. No adverse reaction was observed after administration. In Burkina Faso 5/16 children (2M, 3F) died because of severe systemic complications.

####

About Breath Holding Spells…

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Giacomo is a 10 month old gypsy boy, who came to our attention because of two episodes characterized by prolonged expiratory apnea, unconsciousness, cyanosis and hypotonia lasting about 20 seconds and followed by spontaneous resolution. Giacomo's pregnancy was complicated by oligohydramnios and perinatal distress; the cesarean delivery was at 35 weeks of gestation. To clinical observation, Giacomo showed normal skin color for his gypsy ethnicity and 2/6 systolic heart murmure to centrum cordis due to small left>right jet at the level of oval fossa attributable to patent foramen ovale; at neurological examination the child presented slight asymmetry of active motility, less fluid movement in left upper extremity, capacity for grabbing but not of transferring objects, sitting position not fully captured. Blood examination was performed. A severe iron deficiency anemia is diagnosed (Hb 6,7 mg/dl; MCV 53,9fl; RDW 19%; ferritin 2ng/ml). EEG showed prevalence of right-sided slowest waves. Giacomo started iron therapy; he will have brain MRI and echocardiography checks. 8% of ALTE (Apparent Life-Threatening Events) episodes are due to BHS (Breath Holding Spells) the diagnosis of which is mainly of clinical type and is sometimes associated with that of iron deficiency anemia. Martial status study is therefore recommended in the evaluation of these patients, also in apparent absence of other signs of iron deficiency.

####

Idiopathic Systemic Capillary Leak Syndrome: Case Report

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Systemic capillary leak syndrome is an unusual entity which presents recurrent episodes of hypovolemic shock due to leakage of plasma to the extra-vascular compartment because of immunologic dysregulation. PG, a 9 year old female, came to our first aid service for abdominal pain, pallor and hypotension with no fever. Here an abdomen x-ray (negative) and a thorax x-ray (pulmonary opacity with pleural effusion) were performed. She was admitted, intubated and transferred to ICU because of respiratory distress. Total body TC showed bilateral pulmonary opacity and pleural, pericardial and abdominal effusion. PG took antibiotic, antimycotic and steroid. Laboratory exams showed severe hypoalbuminemia (2,1 g/dl) and anemia (Hb 7,5 g/dl). In the following days, we observed a worsening of the anasarcatic state with hemoconcentration (leukocytosis and elevation of hemoglobin-Hb 11.6 g/dl-). Therefore, PG began therapy with albumin, clonidine, milrinone and furosemide resulting in clinical improvement. SCSL is difficult to diagnose. The characteristic triad of hypotension, hemoconcentration (leukocytosis and an increase of hematocrit) and anasarcatic state, in the absence of other causes of shock, present in this patient, is typical of this syndrome.

####

A possible rare case of genetic SMA

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Acute infantile spinal muscular atrophy (SMA) is as an autosomal recessive congenital neuromuscular disorder with different clinical forms. Classical types (I, II and III) are defined according to time of onset and severity of symptoms, but, recently, other forms, with atypical clinical signs, have been defined (i.e. SMARD). We report a case of a single preterm male newborn (EG:34 weeks, BW:1690g) delivered by C-Section because of polyhydramnios. Parents were not consanguineous, family history was positive for two previous pregnancies complicated both by polyhydramnios with delivery by C-section at 34weeks. The first male died of respiratory failure on the 4th day of life and the second girl also died after 4 hours of life for the same reason. The male showed bilateral clubfoot. No diagnoses were made in either cases. Our newborn, immediately after birth,

required mechanical ventilation, because of very weak respiratory effort. He showed clubfoot, profound hypotonia, absent ROT and fractures of the left humerus and both femurs. Routine blood tests were all within normal limits, as well as CPK. His karyotype was 46 XY. EMG, done at DOL9, suggested myogenic etiology, but muscle biopsy (*m.gastrocnemius*) was not able to identify any classical myogenic etiology and suggested neurogenic etiology. Genetic tests for SMA type 1 were negative while genetic tests for SMARD are pending. Baby passed away after 26 days of life for cardio-respiratory arrest.

####

Evaluation of a handheld Near-infrared Device (InfrascannerTM) for Detection of Intracranial Haematoma in Children with Minor Head Injury.

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Background: CT scan is the gold standard in the identification of intracranial injury (ICI) in patients with minor head injury (MHI). However, the risks related to irradiation and to the sedation of uncooperative patients should not be overlooked. Infrascanner, a handheld near-infrared device, has shown good diagnostic accuracy for intracranial hematomas and hemorrhages in adults with head injury, but there are few data regarding its use in pediatric population. Objective: To determine the feasibility of use and potential accuracy of InfrascannerTM in children with MHI in Pediatric Emergency Department (PED). Design/Methods: Prospective observational study at PEDs of Padua and Treviso (Italy) on patients <15 years of age evaluated for MHI at intermediate-high risk of ICI according to a local adapted PECARN rule. Decision on CT scan and CT reporting were blinded and independent to Infrascanner result. Results: We enrolled 110 pts with a complete measurement ratio without sedation of 94%. Average time for measurement was 4,4±2,9 min. We had a positive Infrascanner result in 8 pts (8%): 1 with positive CT scan for extradural hematoma with the same localization, 4 with scalp hematoma (confounding factor), 3 performed at the beginning of the learning curve. No pts reported any problem at the phone-call follow-up. Conclusions: Infrascanner is an easy-to-use tool thanks to the high completion rate and the short time to completion. Further larger studies are needed to evaluate its diagnostic accuracy.

####

Type 1 Diabetes and celiac diasease: a retrospective study of prevalence and temporal correlation.

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Although the association between type 1 diabetes mellitus (T1DM) and celiac disease (CD) is well known, the exact estimate of the prevalence of this association and the temporal correlation between the two diseases have not been clearly defined to date. In our Center for Pediatric Diabetology in Catania includes 383 patients with T1DM, we identified 32 patients with a diagnosis of celiac disease, with a prevalence equal to 8.3%. We considered two groups: the first one included all those for which the diagnosis of celiac disease was made previously or simultaneously to that of diabetes; the second group included those patients for whom celiac disease had developed after diabetes. 59.3% patients belonged to the first group and had an average age at onset of CD of 4.2 ± 3.8 years (mean ± SD). The second group included 40.7% of the patients with an average age at onset of CD of 8.1 ± 4.2 years. The study, therefore, shows a high prevalence (8.3%) and, above all, a significant association between the age of the patient and the temporal correlation in the onset of both diseases ($p < 0.05$), emphasizing the importance of a more careful and aimed screening for T1DM in young patients with early onset of CD.

####

Cystic Fibrosis and Pregnancy…yes, we can!

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Cystic fibrosis (CF) is an autosomal recessive genetic disorder caused by mutation of the CFTR gene that affects mainly the lungs, pancreas, liver and intestines. We report a case of a 35-year-old woman affected by CF with pancreatic insufficiency, treated at our Center. The patient has a compromised pulmonary status (FEV1 usually between 32-52% and colonization by Pseudomonas aeruginosa (Pa) and Staphylococcus aureus (Sa), a deficient nutritional status (BMI 16-18) and CF-Related Diabetes (CFRD). In her medical history we report miscarriage during the 8th weeks (2011). In June 2012, she began a new pregnancy that proceeded in the normal range, despite a constant deficient state of nutrition (severe hypoalbuminemia) and hydration. During the 20th week, the patient was admitted for an acute respiratory exacerbation caused by Pa and Sa, with increased inflammatory markers and reduced spirometric indices; she had therapy (azithromycin, piperacillin/ tazobactam), resulting in clinical improvement. At the 24th and 28th weeks, the patient undergwent further exacerbations. In the following weeks the pregnancy progressed in the normal range with term birth and healthy newborn. To sum up, pregnancy in women with severe CF, does not aggravate the maternal prognosis, nor affect fetal health. It requires careful follow-up and planning of the pregnancy to coincide with a cycle of ev antibiotic treatment before the beginning of the pregnancy, to avoid the use of drugs in the first trimester.

####

A case of hepatocerebral form of mitochondrial DNA depletion syndrome

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DG was admitted to our hospital at the age of 3 months because of growth arrest and hypotonia. Case history: consanguineous parents, regular pregnancy and neonatal period, 2 cases of SIDS in mother's family. Suspected UTI at 1 month. Physical examination: jaundice, hepatomegaly, axial hypotonia, decreased spontaneous movements, diminished deep tendon reflexes, alert sensory. Laboratory-test results showed hypoproteinemia, transaminases and direct bilirubin elevation, high levels of plasma lactate and altered urinary excretion of organic acids in absence of succinylacetone. Viral hepatotropic markers were negative. Emogasanalysis, ammonium, plasma and urinary amino acids were normal. Ultrasound examination showed increased liver size and echogenicity and abdominal-pelvic spilling. Normal brain MRI, EMG and ENG. Mitochondrial disease was suspected and as a result we performed molecular testing in cultured skin fibroblasts that showed MPV17 gene deletion. Progressive liver disease led to coagulopathy and hypoglycemic crises. DG died as a result of respiratory distress at the age of 5 months. DNAm depletion syndrome is an autosomal recessive disease characterized by deletion of the last exon of the MPV171 gene leading to severe hepatocerebral involvement and exitus before the age of 1year. The therapy is only supportive. The extra-hepatic involvement makes TX liver ineffective. Stem cell transplantation could show promising results.

####

A possible etiology for intracranial hypertension in pediatric age.

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A.M., is a 14 year old girl, who presented with an episode of fever and sore throat associated to supraorbital pulsatile migraine with phonophobia, nausea and vomiting. The headache, exacerbated by eye movements, got worse in the following

days, requiring access to the Emergency Department: ophthalmological examination showed bilateral optic nerve edema with peripapillary hemorrhages (LE>RE). CTscan results were unremarkable so a lumbar puncture was performed, which was normal, but immediate improvement of headache was noted. VEP, ERG and MRI were all normal, confirming the hypothesis of benign intracranial hypertension. No evidence of endocrine dysfunction was identified. The blood virological test ruled out the main etiologic agents (1-2HSV, CMV, parvovirus B19, Adenovirus) and the search for other, uncommon, viruses are still in progress. An empirical therapy with i.v. corticosteroids and acetazolamide 15mg/kg/die was started and iv potassium for 4 days was given for drug-induced hypokalemia.The next ophtalmological check up after 6 days therapy showed complete recovery from the peripapillary hemorrhages and a partial reduction in edema of the papillae. After excluding a secondary etiology for intracranial hypertension a diagnosis of idiophatic pseudotumor cerebri was established. The girl was discharged in good condition with instructions to continue low dose oral steroids tapering down the dose before stopping it completely together with diuretics until the next clinical follow-up.

####

A girl affected by type1 and type2 oral-food allergies

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At 6 months S.R showed urticaria-angioedema after ingestion of gluten pasta, which occured again at 8 months. Therefore, a gluten-free diet was followed. At a year S.R showed the same type of reaction to egg. SPT showed a strong positivity for wheat, egg, fish and soy, so she started a diet free of these foods. At the age of 2 she had recurrent episodes of bronchospasm and oculo-rhinitis, and for this reason SPT was performed for inhalants, which showed a strong positivity for olive, cypress and mites. The execution ImmunoCAP test confirmed positivity for foods and inhalants and celiac disease was excluded. S.R. continues to have severe allergic reactions (anaphylactic), which require a strict diet and constant intake of antihistamines and bronchodilators. The RAST repeated in subsequent years should discover new positivities for foods (peanuts, hazelnuts, almonds, oranges, apple, peach, celery, tomatoes, strawberries, various fish, peas, lentils, barley, rye and oats) and inhalants (various grasses, ragweed pollen, ragweed, wormwood and cat dandruff), justifying the persistence of allergic reactions despite diet and therapy. We assume an allergy to profilin (SPT positive for grass-birch-composite-olive-parietaria associated with sensitization to many foods containing profilin), the syndrome of asthma and /or anaphylaxis by effort, defined by positivity for the molecule ώ-5-gliadin, and slow resolution of allergy to egg, positivity for Gal d1 at the age of 10.

####

A case of neonatal hypoglicemia: the importance of early diagnosis

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M.M. was born at term by eutocic birth, after a physiological pregnancy, weight at birth: 3380 g. A few hours after birth the child developed mild but persistent hypoglicemia so she was sent to UTIN. On the arrival at the department the child was torpid, with difficulties in suction and clonus. Haematic tests showed: hypoglycaemia, hypocorticism, hypothyroidism; levels of IGF1, aminoacidemia, aminoaciduria, Fehling test and sialotranferrine isoelettro focusing were normal. The child also underwent, as every child born in our Centre does, extensive metabolic screening that did not reveal any abnormalities. The child also underwent encephalic RMN which showed: “little and dysmorphic sella turcica with hypo intense signal”. Therefore, we made a diagnosis of panhypopituitarism. We prescribed replacement therapy with Hydrocortisone and Levothyroxine. Two months later, because of low levels of IGF1, we prescribed therapy with Somoatotropin. At the last ambulatory visit the child had normal growth and normal neurological development showing the impor-

tance of early diagnosis and early and careful replacement treatment for prevention of neurological retardation.

####

An acute headache due a nodulary swelling of the skull

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A.M., an 8-year-old male, was born at term by uneventful vaginal delivery to unrelated, healthy parents without a family history for genetic or neurological diseases. His clinical history was only characterized by the presence of two episodes of febrile seizure respectively at 18 and 23 months. At the age of 8 years, the patient was referred to our Department because of an acute temporo-parietal headache. Our evaluation included physical and neurologic examination, which were normal except for a 3 x 3 cm nodulary swelling in the right parietal region of the skull. No fever or neck stiffness was present. Laboratory evaluation demonstrated normal red blood cell, white blood cell, and platelet counts. The results of coagulation function tests and biochemistry profile analysis were normal. Neuroimaging studies were performed, skull X-rays revealed: "a right parietal thecal reduced-intensity bone area rounded of 20 mm in diameter" and a brain MRI showed: "a right parietal osteolytic lesion with oval shape of approximately 16 x 22 mm diameter. The mass marked the underlying cerebral convolutions and is not associated with edemigeni phenomena". The patient was submitted to neurosurgery for removal. Histological examination showed rich infiltration of eosinophils and dense proliferation of Langerhans cells (CD1a, S-100+); furthermore, the lesion was characterized by necrotic-hemorrhagic phenomena and by a growth fraction of about 2-3 mitoses XHPF. Histiocytosis X, an uncommon disorder characterized by an abnormal proliferation of Langerhans' cells, is a spectrum of diseases which should be classified among the wide spectrum of histiocytic disorders, consisting of proliferation of the mononuclear phagocyte system. The clinical manifestations reflect the site of histiocytic proliferation and may vary from a solitary bone lesion discovered by chance on X-ray film to a disease with a rapidly fatal course affecting almost any organ. Our case illustrates once again the clinical heterogeneity of headache as well as the complexity of Histiocytosis X. Therefore, a histiocytic disorder must be suspected in a child with a persistent cranial swelling and an acute secondary headache.

####

Dizzy with genetics

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A. is a 14-year-old Colombian girl adopted at the age of 8 after serious abuse and maltreatment. She came to our attention for dizziness and anomalies of eye movements, associated with severe migraine, photo-phonophobia and nausea, triggered by even small head movements. PMH: sporadic episodes of constrictive headache, bicornuate uterus, malformation of paranasal sinuses, upper calyceal distention of the right kidney with parapyelic cysts and Rathke's cleft cyst. Physical examination: rapid saccadic movements in all direction in ŌŌ accentuated during fixation, linear hypopigmentation on the medial side of the lower limbs along the lines of Blaschko. MRI of brain and cervical spine, ENT visit, eye examination, EEG all normal. The negativity of the investigations, clinical data (improvement in eye movements, persistent headache) and the gradual mood deflection, lead to a migrainous syndrome associated with anxiety somatization disorders. Considering the phenotypic aspects and malformations, genetic investigations were required. Array -CGH analysis revealed a 1,2 Mb duplication of the 17q12 region, containing approximately 15 genes. A few cases of this alteration have been described in the literature recently, associated with a wide phenotypic variability, particularly mental retardation and epilepsy, behavioural abnormalities, urogenital malformations, MODY, esophageal atresia and dyschromatosis.

####

New 2012 guidelines for diagnosis of celiac disease.
Farewell to biopsy?

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MM, a 2yr3m old girl, came to our attention with AGA-IgG (40.1 U/ml) and anti-TG2-IgA>128 U/ml, referred by her pediatrician because of prolonged diarrhea (34 days) after an episode of acute gastroenteritis. She showed normal growth and had not previously suffered from gastro enteric symptoms. Her mother is affected by autoimmune thyroiditis. At the visit, she no longer had diarrhea . We repeated AGA and anti-TG2-IgA in addition to EMA, HLA genotype and anti-actin IgA (AAA): AGA-IgG 24 U/ml, anti-TG2-IgA>70 U/ml, EMA highly positive, HLA-DQ2 positive and AAA negative. In this setting the ESPGHAN criteria 2012 could not be applied because the girl did not have celiac symptoms; therefore an EGD with duodenal biopsies was performed. Histological analysis showed a normal morphology. The girl continued therefore a gluten-containing diet. During the following months, anti-TG2-IgA and EMA gradually diminished to reach normal values after 12 months. In the literature, several cases of positivity for anti-TG2-IgA during acute infections are described, even if not of EMA. Our case report shows that, even if thanks to the new guidelines we can sometimes reach the diagnosis of celiac disease without the need for invasive biopsies, EGD + biopsy still remains the only choice when interpretation of results is difficult or dubious. Therefore, a very strict adhesion to the ESPGHAN criteria is needed to avoid false positives, regarding a diagnosis forcing patients to a gluten-free diet for the rest of their lives.

####

"As much as the love for salt"... A 20 year long story

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Michela is a 3-month-old child. She came to our attention for fever, cough and fidgetiness. Her clinical conditions were compromised. Her body temperature was 39.5° C, RR 60/min, CR 150 bpm, Sat O_2 96%. Fine crakles could be heared at the left apex. The blood count showed an increase of white blood cells. We diagnosed "an apical pneumonia and insufficient growth" and we chose to hospitalize the baby. Michela's mother told us that this was the fourth admission in three months of life. At those admissions Michela had shown signs of respiratory and enteric disease. It was necessary to make further investigations such as the sweat test. It turned out positive and we diagnosed Cystic Fibrosis. The global treatment of Cystic Fibrosis obtained a good growth and a good control on the respiratory pathology. When Michela was 5 years old, we observed an intermittent colonization from Pseudomonas Aeruginosa. When she was 10 years old, the infection became chronic, with progressive development of antibiotic resistance, decrease of respiratory functionality, need of O2 therapy and NIV. When Michela was 22 years old, we decided to list her for transplantation. On March 2013 we admitted her to our hospital for a new relapse of her chronic infection. Her clinical conditions were heavy compromised, so we moved her to an Italian transplantation centre. She started ECMO awaiting transplant, but she died during the operation.

####

Skin features and macrocephaly: hallmarks for specific syndrome

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C. a 5 year-old boy, came to our attention for developmental delay, autism, statural overgrowth and macrocephaly. No family history of intellectual disabilities, mother diagnosed with Klippel-Trenaunay, father

with macrocephaly (>+ 2 SD). On physical evaluation: stature at 90th centile, weight at 50th centile, OFC 55 cm (>+ 2 SD). He showed pigmented macules on the trunk and penis, lipoma in abdomen. We performed routine blood tests, thyroid examination, metabolic screening, molecular analysis for X-Fragile, S. Sotos, S. Simpson-Golabi-Behmel all normal. Karyotype 46, XY. EEG, PAE, brain MRI were all normal as well. On the strong suspicion of Bannayan Riley Ruvalcaba syndrome, we performed molecular analysis which revealed a PTEN gene mutation. We then performed a colonscopy which revealed several polypoid projections on the ileum, colon, and rectum. The microscopic examination of polyps showed a nodular lymphoid hyperplasia. BRRS is a polymalformative syndrome with A.D. transmission and high risk of tumors. Main clinical features are: macrocephaly, multiple lipomas, hamartomatous intestinal polyps, vascular malformations, pigmented macules on the penis. In 50% of cases there's hypotonia, developmental delay and variable intellectual disabilities. In 60% of cases there's a mutation of the PTEN gene while a deletion is seen in 11% of cases. BRRS shares features with Cowden syndrome, an A.D. condition with more females affected, where mutations in PTEN gene are present in 80% of cases.

####

I've got you on my skin…

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S.B., a one year old, was hospitalized in a pediatric department for hyperpyrexia with chills and diffuse maculopapular rash. Nothing to report on past medical history. CBC: Hb 8,7 g/dl, MCV 54,7 fl, WBC 123000 (N 22%, L 75%, M 0%), PLT 255000, the others blood tests and inflammatory markers were normal. Discharged with a diagnosis of interstitial pneumonia, atopic dermatitis and exantema subitum, she still had discontinuous fever, asthenia with progressive weight loss and deterioration of physical condition. Therefore she was hospitalized again: she had hepatosplenomegaly, seborrheic and pale skin, nothing to report upon physical examination; Hb 5,9 g/dl, reticolocites 4%, MCV 61 fl, WBC 11200 (N 26%, L 45%, M11%), PLT 105.000; ESR 33, CPR 1,1 mg/dl. Total body TC showed hepatosplenomegaly with the absence of focal lesions and multiple swollen lynph-nodes in the mesentery. Bone marrow aspirate showed no atypical cells, signs of dyserythropoiesis or block of maturation of metamyelocytes. During hospitalization the baby had episodes of absence (EEG and TC brain normal) and maculopapular disseminated lesions appeared. Skin biopsy showed an infiltratation of CD1 and S100 positive cells which provided for the diagnosis of Langerhans cell histiocytosis. She started therapy with LCH III protocol, with insufficient results; therefore, she was switched to a second-line therapy with 2 CDA (LCHS 2005 protocol), with which she achieved remission without presenting reactivation of the primitive disease. She is now undergoing a periodic follow up.

####

Erythrodermia, oral candidiasis and diarrhea

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CM, a 2-month-old girl, born at 40 wks with normal appearance, was admitted to our Clinic for a dermatitis that appeared during her 2nd wk of life and was diagnosed and treated as "atopic dermatitis" at another Pediatric Clinic. On admission she appeared to be ill, with fever, severe erythrodermia with diffuse scaly rash and cutaneous edema, oral candidiasis (resistant to Fluconazole gel) and diarrhea. Laboratory evaluation showed eosinophilia (WBC 13200/□L, N 63%, L 14%, M 8% And 15%), Hb 11.9 g/dL, PLT 348000/□L, IgG 341 mg/dL, IgM 19 mg/dL, IgA 16 mg/dL, IgE 14 IU/mL, lymphopenia with absence of B-cells and of naive CD4 T-cells (ALC 1850/□L,, CD3 1590/□L, CD4 1498/□L, CD4-CD45RA <1%, CD4-CD45RO 81%, CD8 110/□L, CD19 <1%, CD16-56 168/□L), absent *in vitro* lymphocyte proliferative

response to mitogens, positive oral *Candida albicans*, a paternal haplotype and one alone maternal haplotype in HLA typing. From the clinical features and laboratory data we made the diagnosis of Omenn Syndrome in SCID $T^-B^-NK^+$. CM was urgently hospitalized in the Pediatric HSCT Unit, treated with NPT, piperacillin-tazobactam, amikacin, liposomal amphotericin, acyclovir, Ig, MPN, topical steroids, ciclosporin, trimethoprim/sulfamethoxazole and received a haploidentical HSCT from her father. 10 years later, she shows normal hematological and immunological values. We found no mutation in her *RAG1/RAG2* genes.

####

A case of hipertensive hydrocephalus due to brain malformation

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Holoprosencephaly (HPE) is a brain malformation resulting from incomplete cleavage of the prosencephalon into two cerebral hemispheres. It can be alobar, semilobar or lobar. In 1993, another milder subtype called "Middle Interhemispheric Variant" (MIHF) was reported. The HPE can be due to environmental causes (diabetes, alcoholism, exposure to drugs and TORCH infections during the pregnancy) and genetic causes. CASE REPORT: Giovanni was born at gestational week 35 by cesarean delivery, with severe hydrocephalus. HPE was suspected on the fetal US at 20^ w. After birth, the child was admitted in NICU where the hydrocephalus was confirmed, without dysmorphic features. The brain US showed a picture of severe malformative hydrocephalus. The partial separation of frontal and occipital lobes and of the thalami didn't allow a differential diagnosis between HPE and MIHF with a dorsal cyst. The brain CT confirmed the US picture. Immediately after the diagnosis, Giovanni was transferred to a Pediatric Neurosurgery centre where a ventriculoperitoneal shunting was performed. CONCLUSIONS: HPE occurs in 1:16000 live births but its frequency may be as high as 1:250 pregnancies. The incidence of MIHF is unknown. The clinical manifestations depend on the severity of the HPE and range from severe cerebral paralysis (often Choreoathetosis) with endocrine and hypothalamic dysfunctions and dysmorphic features, to milder spasticity that doesn't prevent ambulation (MIHF).

####

A case of Anorexia

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G. 17 years, came to our observation for acute decompensation in chronic anorexia . A clinical history of gastroesophageal refux, recurrent tonsillitis and a sister with a history of anorexia (which strongly influenced the patient psychological condition) should be noted. G. presented in bad general condition, pale, with lower limb acrocyanosis, petechiae of the forearms volar surfaces, lanugo and palmar-plantar hypercarotenemia. Deambulation was difficult; she presented ankle edema and bradycardia. G. was uncooperative, she had dysmorphophobia, with altered body image, middle insomnia, weight phobia and anxiety symptoms. She presented also hypetransaminasemia, mild thrombocytopenia, hypoglycemia, hypocalcemia and a slight hypophosphoremia, even before therapeutic intervention. The echocardiography showed a mild to moderate pericardial effusion. To avoid refeeding syndrome we started the correction of electrolyte abnormalities. Only as a second step did we set a dietary plan and contemporary psychiatric and emotional support therapy, while waiting to start cognitive-behavioral therapy (not effective without a certain weight recovery). Anorexia nervosa is a serious medical and psychiatric emergency, and, for this reason, early intervention, the correction of electrolytes and monitoring of clinical parameters together with medical and psychiatric treatment are crucial for prognosis.

####

A rare case of a girl with precocious puberty

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L., a 3.4 year old, developed pubarche at 1 year. At first presentation height was 102.1 cm (96°%ile vs 3°%ile THT), height velocity (HV) 12.8 cm/year, pubertal stage AH1, B1, PH3. She had advanced bone age (4.4 years), elevated testosterone concentrations (T), normal pelvic and adrenal US. CAH was excluded, and in search for the source of T, an abdominal CT scan was then performed, which was normal. After 6 months she continued to show pubertal growth, increased pubic hair and increased T concentration. An abdominal MRI revealed the presence of a solid mass in her right adrenal gland (4*2.5*2.2 cm). The tumour was removed and it turned out to be a stage II carcinoma. She was then referred to the paediatric oncology unit, and started treatment with Mitotane (protocol Trep AIEOP) and Hydrocortisone. After 6 months (4.9 years) she developed bilateral thelarche (B3). Results of a LHRH test (FSH peak 10.6 mU/ml, LH peak 66.6 mU/ml) and elevated oestradiol (49.5 pg/ml) concentration indicated she had developed central precocious puberty. Treatment with GnRH analog was then initiated. Precocious puberty can occur after the removal of a virilising tumour. Excess androgens may lead to maturation of the hypothalamic-pituitary-gonads axis, and removal of the feedback effect exerted by sex steroids would trigger endogenous LHRH secretion, thus causing gonadotropin-dependent precocious puberty.

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Cerebellar hypoplasia and pneumonia

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An 8 month-old boy, born at 36 wks with IUGR and cerebellar hypoplasia, was admitted to our Clinic for a cough which had progressively worsened over a few days. On admission, he appeared to be ill, with high fever, tachypnea, grunting, retractions, and also slight microcephaly, oral candidiasis, pallor and petechiae. Chest radiography showed interstitial pneumonia, that suddenly worsened and intubation and mechanical ventilation was required. Laboratory evaluation showed WBC 8900/uL (N 36%, L 26%, M 35%, E 3%), anemia (Hb 7 gr/dL, reticulocytes 1.5%), thrombocytopenia (PLT 20000/uL), lymphopenia with absence of B-cells and NK-cells (ALC 2010/mcL, CD3 1980/mcL, CD4 1450/mcL, CD8 520/mcL, CD19 <1%, CD16-56 <1%), absent in vitro lymphocyte proliferative response to mitogens, agammaglobulinemia (IgG 80 mg/dL, not detected IgM and IgA), positive oral Candida albicans, positive BAL PCR-DNA Pneumocystis jirovecii. From the pathognomonic association of cerebellar hypoplasia and SCID T+B-NK- we made a diagnosis of Hoyeraal-Hreidarsson Syndrome (the severe infantile variant of dyskeratosis congenita; in this child the XL form, caused by a novel missense mutation in diskerin coding gene DKC1, exon 3 T113-->C, Ile38Thr). Pneumocystis pneumonia was cured, and the child received an allo-HSCT from her HLA-identical sister, with sustained correction of immunological and hematological defects. Unfortunately, her generalized telomerase defect caused a premature aging syndrome, with exitus (cirrhosis of the liver) at 10-years-old.

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Multicystic dysplastic kidney in children: conservative management and long term approach

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PURPOSE: Multicystic dysplastic kidney (MCDK) is a relatively common form of renal dysplasia. Associated urinary tract anomalies may accompany MCDK changing its natural history. The aim of this study is to evaluate the clinical course and the long-term (20-years) renal outcome of children with MCDK. MATERIAL AND METHODS: We retrospectively evaluated 79 children, 52 males (65,8%) and 27 females (34,2%) with unilateral MCDK. Mercaptoacetyl-triglycine (MAG3) renal scans were performed in all patients. The voiding cystourethrogram (VCUG) was performed only in children with a dilated urinary tract to exclude other uropathies. Was also monitored blood pressure and GFR. RESULTS: Diagno-

sis was made by prenatal US in 68 patients (86%). 16/79 patients (20,2%), had abnormalities in the contralateral kidney. Compensatory hypertrophy of the contralateral kidney occurred in 70 patients (88,6%). Two children had reduced GFR. Proteinuria was present in 2 of 79 patients. Hypertension occurred in 1 of 79 patients. There was complete involution of MCDK in 59 patients (74,6%) and no malignant transformation in any patients. Only four children had recurrent UTI. CONCLUSIONS: The majority of children with MCDK are apparently healthy, have favourable outcomes and do not need nephrectomy. VCUG is no longer recommended for all children and must be performed only in selected cases. Follow up must be more rigorous in patients with complex MCDK, who are at significant risk of developing UTI and renal failure.

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Mellitus Diabetes and eating disorders: a complex case

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M. a 13 year old came to our observation for a suspected eating disorder, lack of control over food assumption, binge eating and purging, in particular vomiting. The patient was suffering from diabetes mellitus type 1 without complications by the age of 8, and had a reasonable metabolic control (up to that point) but exhibited all the typical psychological implications of chronic diseases. At the time of the visit M showed depressive symptoms, she referred self-harm behavior and gradual caloric restriction, followed by episodes of loss of control. Although auxological and body composition parameters were not altered, bulimic symptoms were accompanied by a metabolic decompensation of diabetes, in particular blood glucose values, because of a poor compliance. Because of frequent hypoglycemic crises we decided to significantly reduce insulin units and to set a proper diet plan, aiming at vomiting interruption and metabolic correction. To improve depressive symptoms we prescribed therapy with sertraline. During hospitalization our patient also manifested nausea, headache, epigastric pain and left upper limb pain, and also anxiety symptoms, auditory hallucinations, which lead to convertion episode a few months later. We completed her therapeutic plan with alprazolam, lorazepam and risperidone. In time the mood and eating disorders improved, but the conversion disorder and her unstable psychological condition make the management of insulin therapy and metabolic control difficult .

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Outcome in children with vesicoureteral reflux: 19 years of follow-up

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PURPOSE: Vesico-ureteral reflux is a disease that affects 1-2% of the pediatric population, it can be associated with reflux nephropathy and the development of chronic renal failure in adults. The aim of this study was to evaluate the validity of the conservative approach to VUR and provide practical guidance regarding the management of patients with VUR and UTI. MATERIAL AND METHODS: This study included 181 patients with primitive RVU aged from 0 to 24 between 1993 and 2012. Patients were divided into group A (children younger than 2 years) and group B (patients aged between 2 and 24 years). All 181 patients underwent ultrasound of the urinary tract and voiding cystourethrogram. Kidney function was evaluated with GFR renal and scintigraphy. RESULTS: Of the 181 patients with VUR, 80 (44.1%) received a prenatal diagnosis. The children who did not receive a prenatal diagnosis and who were not prescribed prophylactic antibiotics after birth, developed febrile UTI in a percentage of 75% (group A) and 80.6% (group B). Only 29/181 patients underwent surgery. None of the 181 patients had chronic renal failure. CONCLUSIONS: Only selected cases require surgery. We cannot predict which patients will develop ESRD in adulthood. Prenatal diagnosis is important to prevent UTI in children with VUR under 2 years of age. For older patients, it is necessary to diagnose and treat voiding dysfunction and constipation to prevent UTI

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Not just an hypertransaminasemia

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A 3-year-old girl was referred to our hospital with hypertransaminasemia. Anamnesis: recent gastroenteritis and an episode of oral candidiasis six months before. Normal physical exam. The laboratory tests showed hypertransaminasemia, hypergammagt and hypergammaglobulinemia with normal restant parameters of liver function. Negative serology for hepatotropic viruses. Normal ceruloplasmin, serum and urine copper and alpha1antitrypsin. Positivity of ANA and anti-LKM with high title should be noted. Celiac disease and IBD were excluded. Liver biopsy revealed evidence of chronic injury of the intrahepatic bile ducts and lobules. After an evaluation of the girl's medical history, laboratory tests and histological exam AIH/SC overlap syndrome was diagnosed. Genetic investigation for APECED was carried out, which showed homozygosity for the mutation R139X. She was treated with corticosteroids and azathioprine and was followed up for the management of the disease. Colangioresonance and hepatic function had become normal. At 4 years she developed hypoparathyroidism and maintained a normal adrenal function, which is kept under control. Upon screening the whole family, the younger brother was found to be homozygous for the same mutation. The APECED is a rare genetic disorder of immunoregulation, autosomal recessive, caused by mutations in the AIRE gene. The typical symptomatic triad is: mucocutaneous candidiasis, hypoparathyroidism and adrenal insufficiency. Approximately, 15-20% of patients also develop a AIH.

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Influence and prevention of xerostomia in the oral manifestations of pediatric onchoematologic patient.

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Xerostomia plays a fundamental role in the genesis and clinical aggravation of oral cavity diseases. The purpose of this study was the evaluation of the role played by xerostomy in the oral cavity diseases of children affected by LAL and LMA. We enrolled 60 children: Group A (20 patients undergoing chemotherapy), Group B (20 patients in maintenance chemotherapy) and Group C (20 healthy children). We evaluated: salivary flow, basal salivary PH and PH after stimulation with paraffin, DMFT index, BEWE index and PCR. The results were: higher incidence of xerostomy in Group A (85%) compared with Group B (65%), and in Group A and B (75%) compared with Group C(10%).Increase of PCR index in Groups A(48,5%). Increase of BEWE index in Groups A and B (45%);Normal values in Group C (15%). Increase of DMFT index in Group A compared with Group B, Groups A and B compared with Group C. Increased PH and salivary flowmetry after paraffin mixtures. Xerostomia represents a statistically significant risk factor (p-value < 0,05) for the oral cavity.Paraffin may represent a valid aid in preventing xerostomy. Oncohaemathological young patients need: 1) more attention to chemo and radiotherapy-induced xerostomia 2) accurate information about the oral cavity 3) highly qualified staff that have to be trained and informed about specific therapies.