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WHEN AN ECG MAKES DIFFERENCE.... QUANDO UN ECG FA LA DIFFERENZA ...

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This is the case of a 6-year-old girl who presented with sudden loss of consciousness (LOC) after a crying episode, associated with cyanosis, ocular retroversion and generalized hypotonia, which lasted about two minutes. A similar episode occurred 24 hours later, preceded by dizziness and auditory and visual disturbances. For this reason, she was admitted to a Pediatric Department where routine laboratory tests and a brain CT and EEG were performed and resulted negative. After 24 hours, she had a recurrence, so biochemical examination of CSF was performed to rule out encephalitis and empirical antibiotic and antiviral therapy was started. Approximately 30 hours after hospital admittance, the patient displayed a new LOC associated with perioral cyanosis and bradycardia with a spontaneous resolution and rapid recovery (<1 ') of consciousness. At this point an ECG was performed which identified a prolonged QT. The girl was transferred to the Pediatric Cardiology Department. Upon admittance, the patient exhibited arrhythmic instability with evidence of T wave alternation, frequent premature ventricular polymorphic beats, and prolonged QTc up to 600 msec. Pulseless Torsades de Pointes occurred and was treated with CPR and Defibrillation, I.V. infusion of MgSO4, Isoproterenol (to maintain baseline HR>100bpm), KCl (up to 60 mEq/day during beta-agonist I.V. therapy). As mid-long term treatment, PMK-ICD was implanted and Nadolol was introduced. A genetic study documented the mutation 1873 G>A missense of the KCNH2 gene, compatible with LQT2. About 3-5% of cases of pediatric syncope have a cardiac origin. Nevertheless, the ECG must be an initial and inevitable screening tool.

####

HOW MANY SPECIALISTS FOR A SIMPLE DIAGNOSIS?
QUANTI SPECIALISTI PER UNA DIAGNOSI SEMPLICE?

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This is a case of a 15.5 year-old male football player, who, 10 months before, began suffering from vomiting and diarrhea, reduced heat tolerance and fatigue, sweating while at rest, hyperphagia without weight gain, and palpitations. He was diagnosed with pneumonia, but, despite a course of antibiotics, all the clinical features persisted. The echocardiogram was normal. His gastroenterological function was evaluated, and binge eating disorder and autoimmune intestinal diseases were excluded. Other examinations identified abnormal thyroid function: TSH <0.005 mUI/l, fT4 35.5 ng/l (9.3-16). At the first visit to our Pediatric Endocrinology department, he was 60 kg per 179.5 cm and showed neither thyroid goitre nor exophthalmos. BP was 140/60 mmHg with hyperkinetic cardiac signs and relative tachycardia (90 bpm, previous reported HR at rest 50 bpm). The thyroid ultrasound showed enlargement of the gland with evidence of chronic thyroiditis, without nodes and low vascularization at ECD. Recurrence of autoimmune disorders (celiac disease, Hashimoto thyroiditis and rheumatoid arthritis) in the maternal family was reported. We confirmed the diagnosis of Graves' thyroiditis, with borderline positive TSHR-Ab. Tiamazole (0,3 mg/kg/die) and Bisoprolol were prescribed and symptoms improved and thyroid hormone levels were reduced. Graves' thyroiditis is the most common

cause of hyperthyroidism in children and adolescents. Delay in its diagnosis occurs when the symptoms are evaluated singularly instead of collectively.

####

SOMETIMES THEY COME BACK
A VOLTE RITORNANO

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F. was an 8-year-old boy admitted to hospital for intermittent fever that started 10 days before and persisted despite 5 days of therapy with amoxicillin -clavulanate for pharyngitis. The boy also had a headache, low back pain, and an alternating stool pattern since a few weeks before. The physical examination showed pallor, dehydrated lips, cervical lymphadenopathy, red and swollen tonsils and a 2/6 systolic heart murmur. Laboratory tests revealed elevated CRP, ESR, ferritin and Anti-streptolysin O. A chest X-Ray was normal, while an abdomen ultrasound showed moderate splenomegaly and a thickened sigmoid colon wall with surrounding lymphadenopathy. An echocardiogram was also performed revealing thickened aortic and mitral valves - the first one with moderate insufficiency, and the second one with mild insufficiency - confirming our clinical suspiction of rheumatic fever (R.F.). The patient started treatment with benzathine penicillin G, prednisone and salicylates. After two days he was apyretic.
Due to its high prevalence in low- and middle-income countries, R.F. is the most common pediatric heart disease in the world and, in many countries, the most common cause of death in children and young adults. Appropriate treatment of streptococcal pharyngitis in high risk populations eradicate most of S. pyogenes infections thus preventing R.F. This case is interesting because rheumatic fever is a rare disease in Europe and this child developed it after an episode of pharyngitis properly treated with antibiotics.

####

GINGIVAL ENLARGEMENT IN A TEENAGER
UNA TEENAGER CON IPERPLASIA GENGIVALE

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Gingival hyperplasia can be the primary sign of a systemic disease. Drugs, tumors, vitamin deficiency, granulomatosis diseases and genetic disorders should be considered for differential diagnosis.
S., a 14-year-old female, was admitted to our department for severe anemia. She was asymptomatic, except for asthenia after physical exercise. At clinical evaluation, we only found gum overgrowth, which had begun to appear four years earlier. She had already undergone a gingival biopsy, which identified a granulomatous lesion, and an autoantibodies profile, including ANCA, which resulted negative. Because of severe microcytic hypochromic anemia, S. received an iron transfusion. Under clinical suspicion of scurvy (she referred to having a very selective diet), vitamin C was prescribed and she began oral supplementation. The lower limbs X-ray resulted negative for scurvy bone changes, while the vitamin C serum level was just below the lowest normal limit. During the exams, we found ANCA positivity, ESR elevation and mild proteinuria. The pulmonary function test, including DLCO, and chest CT scan showed pulmonary involvement with restrictive patterns.
In conclusion, even if S. didn't present typical "strawberry gingivitis", her gum hyperplasia was the first manifestation of

granulomatosis with polyangiitis (Wegener's granulomatosis - W.G.), probably enhanced by a mild scurvy.
W.G. is a multisystemic autoimmune vasculitis that can present with intraoral lesions long before multiorgan involvement occurs.

####

NEONATAL CHOLESTASIS AND DOWN SYNDROME: CASE REPORT AND LITERATURE REVIEW
COLESTASI NEONATALE E SINDROME DI DOWN: CASO CLINICO E REVISIONE DELLA LETTERATURA

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Introduction. Neonatal cholestasis (NC) is an uncommon condition (1:2500 neonates) that always indicates a hepatobiliary dysfunction. Causes include biliary obstruction, hepatic or systemic infections, metabolic diseases, toxic and alloimmune insults. Biliary atresia is the main cause in otherwise healthy term neonates. The prevalence of Down syndrome in Italy is 1 in 1000 newborns; in this population, NC occurs more frequently with a prevalence of 3.9%.
Case report. A 27-day-old male, born at term and affected by Trisomy 21, presented with jaundice and acholic stools. The baby was in good general conditions and displayed regular growth. Lab tests showed conjugated hyperbilirubinemia, moderate hypertransaminasemia and GGT elevation. Ultrasound identified a hyperechoic liver and a normal gallbladder. No other congenital malformations or haematological abnormalities were present. Liver histology showed a giant cell cholestatic hepatitis with paucity of intrahepatic bile ducts. Subsequently, liver tests gradually improved.
Conclusion. Infants affected by Down syndrome carry a 100-times increased risk of developing NC in comparison to healthy term neonates. The main cause is paucity of intrahepatic bile ducts that usually shows spontaneous resolution overtime. However, when associated to concomitant transient abnormal myelopoiesis (TAM), NC may be more severe and lead to liver failure. In infants with Down syndrome, prolonged jaundice and acholic stools should always prompt the investigation of NC.

####

D-LACTIC ACIDOSIS COMPLICATING ACUTE PANCREATITIS IN A PATIENT WITH SHORT BOWEL SYNDROME
ACIDOSI D-LATTICA COME COMPLICANZA DI PANCREATITE ACUTA IN UN PAZIENTE CON SINDROME DELL'INTESTINO CORTO

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A 6-year-old boy was admitted to the hospital because of 2-day history of emesis. His medical history was notable for a prolonged NICU hospitalization due to prematurity, respiratory distress and necrotizing enterocolitis resulting in short bowel syndrome (SBS). Blood tests showed a rise in amylase and lipase and hypocalcemia, suggesting acute pancreatitis. We implemented I.V. rehydration, fasting and parenteral nutrition for 5 days, followed by standard pancreatitis diet regimen. On day 19, the child developed neurological symptoms such as altered mental status, drowsiness and impaired motor coordination, with normal EEG. Physical examination disclosed marked abdominal distension and pain. Arterial blood gas analysis revealed a severe metabolic acidosis: pH 7.25, HCO3- 9.6 mM, PaCO2 22 mmHg, normal lactate (L-isoform) and high serum anion

gap. A diagnosis of D-lactic acidosis was made. Treatment consisted of I.V. bicarbonate and rehydration, I.V. metronidazole, IM thiamine, and exclusion of the primary source of D-lactate production as simple sugar or yogurt. Blood tests and neurological status were normalized within 24 hours. D-lactic acidosis should be considered in patients with SBS who develop unexplained metabolic acidosis with encephalopathy. The main mechanism explaining D-lactic acidosis in SBS is carbohydrate malabsorption. Undigested carbohydrates are fermented by colonic microbiota capable of producing D-lactic acid. Serum D-lactate>3 mM supports diagnosis.

####

A RESISTANT KAWASAKI DISEASE
UNA MALATTIA DI KAWASAKI AL LIMITE DELLE POSSIBILITÀ TERAPEUTICHE

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Introduction: Kawasaki disease (KD) is an autoimmune disease occurring mostly in children under the age of 5 years, characterized by an acute inflammation of the blood vessels. The etiopathogenesis is still unknown and, even if it is usually self-limiting, some patients may develop cardiac abnormalities. Treatment consists of intravenous immune globulin (IVIG) and aspirin, but a small percentage of patients require additional therapy. Case report: An 8-year-old male presented with 3 days of remittent fever, pharyngitis, lymphadenitis and scarlet rash. Routine examination showed neutrofilic leucocytosis and increased inflammation indicators. We initially suspected scarlet fever, however the fever lasted over 5 days and was associated with other typical clinical signs, which led to a diagnosis of KD. Treatment: The first step of therapy was a single-infusion of IVIG and high-dose aspirin. Since the fever persisted, we performed a second cycle of IVIG without clinical improvement, followed by infusion of IV corticosteroids (30mg/kg once-daily for 3 days) that resolved the fever. After 5 healthy days, our patient's high fever returned, so he underwent a second cycle of steroids (1mg/kg twice-a-day for 2 days). Conclusions: KD may be considered in children over 5 years. Fever resistant to IVIG, male gender and increased C-reactive protein are predictors of complications of the disease. A second cycle of steroids may be considered before immunomodulatory or cytotoxic therapy.

####

COMMON DRUGS MAY HAVE UNUSUAL CONSEQUENCES
FARMACI ABITUALI POSSONO AVERE CONSEGUENZE INUSUALI

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A 2-year-old Moroccan girl was transferred to our unit from a local pediatric unit because her symptoms of cutaneous lesions, adenitis and a persistent fever, treated with clarithromycin therapy without clinical improvement, were worsening. Clinical examination revealed widespread vesciobullous lesions on the chest, purpuric macules on the back, palpebral edema, and ulceration with hemorrhagic crusting of the lips. We suspected a Stevens-Johnson syndrome

(SJS). Blood exams showed: 20240/uL WBC (73,4% N), Hb 10,6 g/dL, PLT 478000/uL, albumin 3,3 g/dL, CRP 85,3 mg/dL, PCT 6,92 ng/mL. TORCH, Coxackie, Mycoplasma, Parvovirus B19, and hepatitis A, B, C serologies were negative. Clarithromycin was withdrawn and moisture-retentive ointments, corticosteroid, and ceftriaxone therapy was started. On the third day, epidermal sheet-like desquamation started from the extremities and perineum. IVIG were started and she showed gradual improvement of the cutaneous lesions. SJS is a severe dermatologic reaction characterized by keratinocyte apoptosis and epidermal detachment. Although it is often caused by drugs, including sulfonamides, anticonvulsants and NSAIDs, or infections, the precise pathogenesis of SJS is unclear. We reported this case because SJS has rarely been associated with Clarithromycin oral suspension, and therapy with corticosteroids and IVIG is still debated. Considering the high mortality rates of SJS, it is important to recognize and promptly treat this condition.

####

WHOLE EXOME SEQUENCING IN AN UNUSUAL CASE OF PERSISTENT EARLY ONSET HYPERTRANSAMINASEMIA
WHOLE EXOME SEQUENCING IN UN INSOLITO CASO DI IPERTRANSAMINASEMIA A ESORDIO PRECOCE

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This is a case report of persistent hypertransaminasemia and cholestasis in a male infant (conceived by ICSI, 32 gestation weeks). He was admitted to NICU with respiratory distress and transient diffuse muscular hypertone. At 4 months he presented with food refusal, severe poor growth, mild muscular hypertone and increased transaminases, gamma-GT and alkaline phosphatase. All autoimmune, metabolic and infective blood screening, and brain and abdominal ultrasound imaging were normal. Sleep-EEG showed synchronous sharp elements on occipital regions. Hepatic scintigraphy showed hepatocellular damage, with no bile pathways dilation on cholangio-MRI. At 8 months, transaminasemia and cholestasis indices worsened, abdominal ultrasound showed hyperechogenous hepatomegaly, brain MRI showed a periventricular hyperintensity on T2-weighted sequences and mild atrophy. Hepatic biopsy showed hepatocellular massive steatosis. We excluded Niemann -Pick disease type C, lysosomal acid lipase deficiencies, glycogenoses, galactosemia, fructosemia, and fatty acid beta-oxidation defects. Whole Exome Sequencing (WES) was finally performed, showing a p.Gly290Arg homozygous variant in the TUFM gene, determining a combined deficit of type-4 oxidative phosphorylation. Based on this outcome, metabolic diseases should be considered in differential diagnosis of infants with progressive hypertransaminasemia, cholestasis and hepatomegaly. Genetic tests such as WES, which are able to detect novel mutations in genetically heterogenous diseases, should also be utilized.

####

ACUTE ABDOMEN IN A FEMALE TEENAGER
ADDOME ACUTO IN UNA ADOLESCENTE

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A 12-year-old girl came to pediatric emergency department for vomiting, diarrhea, hypogastric abdominal pain and fever. Blood tests showed neutrophilic leukocytosis, elevated CRP and IgM positivity for Mycoplasma pn., the abdominal

ultrasound showed a right ovarian cyst, and the chest X-ray detected lung parenchymal thickening. Antibiotic therapy with clarithromycin was started. Several days later, the girl complained again about abdominal pain on the right and hypogastric area. Blood tests demonstrated signs of inflammation but subsequent abdomen ultrasound were always regular, but did not show the appendix. For this reason an abdomen-CT scan was performed which showed inflammation and multiple lymphnodes in mesenteric adipose tissue, greater in the pelvis, perivescical, in Douglas'pouch. Suspecting a flogistic pathology of the appendix, an exploratory laparoscopy was performed and revealed a gangrenous perivescical appendicitis, which led to a laparotomic appendicectomy.
Diagnosis of acute abdomen in adolescents is typically insidious. The possible causes in young patients range from gastro-intestinal, to genital and urinary disorders. The appendix atypical sites increase the possibility of clinical manifestations involving other organs (e.g urinary irritation in pelvic and perivescical localization). On the other hand, right basal pneumonia often simulates an acute abdomen.

####

SYNCOPE OR NOT SYNCOPE, THIS IS THE PROBLEM
SINCOPE O NON SINCOPE, QUESTO E' IL PROBLEMA

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A 16-year-old girl was admitted to our hospital for recurrent episodes of transient loss of consciousness. Her mother suffers vagal vascular syncopes. In our patient all the episodes were characterized by prodromic symptoms (dizziness, malaise, nausea, visual blurring) and triggers (pain, prolonged orthostatism). Over the past 3 years, she also reported short term episodes of palpitations. During a previous hospitalization, blood tests, EEG, brain-CT, echocardiogram and ECG were all negative, but an HolterECG showed supraventricular tachycardia (180 bpm). During her first hospitalization in our structure, the tilt test was positive for vagal vascular syncopes and the transesophageal electrophysiological study showed a inducible supraventricular tachycardia. She began tilt-training. One year later, the patient presented with new episodes of loss of consciousness, without trigger and prodromic symptoms, but with some significant traumas. A loop recorder was implanted, which excluded the association between the episodes and supraventricular tachycardia. She began therapy with Ivabradina, without benefit, so she was back at our hospital. During the second hospitalization, a positive EEG (isolated sharp waves, with a right occipital parietal temporal focal face) was discovered, but another sleep-deprived EEG resulted normal, so no therapy was prescribed. One year later, due to the presence of clonus and muscle hypertonus during an episode, therapy with levetiracetam was started. No more episodes over the following 18 months were detected.

####

A 2-YEAR-OLD GIRL WITH COMPLICATED GASTROENTERITIS
UNA GASTROENTERITE COMPLICATA

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A 2-year-old girl with no significant past medical history was taken to the emergency ward with a 7-day history of diarrhea and vomiting, complicated by drowsiness and oliguria. At admittance, she was unconscious, pale, febrile, hypotonic with intermittent hypertonus of the right arm; HR 170 bpm, BP 69/35 mmHg, SpO2 100%. Blood exams showed metabolic acidosis (pH 7.06, HCO3- 5 mmol/L, BE -23 mmol/L, AG 32 mmol/L), hypernatremia (Na 162 mmol/L), increase in creatinine (592 umol/l) and urea (121 mmol/l) rates, normal blood count, except for mild leukocytosis, no signs of hemolysis. Head CT scan was negative. In the Pediatric Intensive Care Unit she received supportive care with intravenous fluid resuscitation and anticonvulsant therapy and showed improvement. Renal function resolved within 48 hours. Thrombosis of the superior sagittal sinus was detected with a cerebral MRI, with associated ischemic lesions. Intravenous heparin was started. Underlying metabolic disorders and thrombophilia were excluded and microbiological investigations were negative. Complete neurological recovery was achieved after two weeks. Follow-up MRI showed complete recanalization.

Conclusions: Differential diagnosis in a dehydrated child with neurological impairment includes electrolyte and metabolic imbalance, viral encephalitis, hemolitic-uremic syndrome but also rare conditions such as cerebral sinovenous thrombosis. A prompt diagnosis and treatment can avoid neurological sequelae.

####

BASM: NOT JUST BILIARY ATRESIA
BASM: NON SOLO ATRESIA DELLE VIE BILIARI

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Introduction. The rare association of biliary atresia (BA) and splenic abnormalities (polysplenia, asplenia etc) is defined by the acronym BASM. Other major defects (cardiac ones, situs inversus) can also be present.

Case Report. M., female, born at 37 wks from CD, BW 3.2 kg, presented jaundice, pale stools, dark urine, failure to thrive since the first days of life. Blood exams performed on 30th day of life (date of hospital admission) highlighted cholestatic hepatitis (AST 245U/L, ALT 219U/L, GGT 1682U/L, Tot Bil 7.8mg/dl, Dir Bil 4.3mg/dl). Other main causes of neonatal cholestasis were excluded. Abdomen ultrasound showed hepatomegaly, triangular cord and a cavernous transformation with slow flux of the portal vein left branch. Based on these findings, M. underwent a diagnostic laparotomy, which confirmed the diagnosis of BA type III, cavernous venous malformation and splenic agenesis. Surgeons were not able to use Kasai procedure (KP) in this complex malformation, so M. was discharged with total enteral feeding (special formula with MCT), vitamin subministration and UDCA. The aim was to allow M. to grow in order to get on the waiting list for liver transplant.

Conclusion. Although BASM is very rare, it appears to be a distinct subgroup among infants with BA. Long term studies show a severe failure to thrive and a worse prognosis of liver disease for these patients. KP is not always possible in BASM syndrome and often OLT is the only therapeutic choice.

####

FIBRODYSPLASIA OSSIFICANS PROGRESSIVA IN A 14-MONTH-OLD FEMALE INFANT: FEW SIGNS TO ALLOW FOR EARLY DETECTION OF A VERY RARE DISEASE

FIBRODISPLASIA OSSIFICANTE PROGRESSIVE IN UNA BAMBINA DI 14 MESI: POCHI SEGNI PER SOSPETTARE PRECOCEMENTE UNA MALATTIA MOLTO RARA

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Fibrodysplasia ossificans progressiva (FOP) is a severe inheritable disorder of connective tissue (prevalence 1:2000000), characterized by big toe malformation (hallux valgus/abnormal first metatarsal/ monophalangism) and progressive heterotopic endochondral ossification. Affected patients appear normal at birth, except for toe malformations. Episodes of painful soft tissue swelling (flare-ups) are reported in the first decade of life, often precipitated by injury or viral infection. Any invasive intervention on these lesions could lead to their transformation into heterotopic bone. A 14-month-old female infant was referred to our Clinical Genetic Unit under suspicion of a congenital malformation syndrome. She had non-consanguineous healthy parents and older sister, unremarkable information on pregnancy and neonatal period, normal anthropometric size. At 2 months of age, she was first evaluated for bilateral hallux valgus and, in the following months, for recurrent upper airways flogistic episodes. Finally, at 14 months of age, she presented subcutaneous soft swollen areas on the neck and was hospitalized suspecting a neoplastic process. Routine blood exams and infectious tests, urinary VMA, Mantoux test, abdomen echography, cardiological and neurological consultations were all normal. Neck echography showed thickening of the muscular walls and some big lymph nodes with tissue edema, and the neck-chest-abdomen CT scan confirmed these findings. Tissue biopsy was proposed but FOP was also considered. Molecular analysis of ACVR1 gene was performed and demonstrated a pathogenetic heterozygous activating mutation (617G>A; R206H) of the gene. Clinical findings confirmed by genetic analysis, avoiding the use of invasive tests such as biopsy, allowed for the precocious diagnosis of our patient.

####

TOXIC SHOCK SYNDROME OR KAWASAKI DISEASE, A CASE OF DIFFICULT DIFFERENTIAL DIAGNOSIS

SINDROME DELLO SHOCK TOSSICO O MALATTIA DI KAWASAKI, UN CASO DI DIFFICILE DIAGNOSI DIFFERENZIALE

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A 6-year-old girl presented with a history of profuse diarrhea and fever the preceding 2 days and impetigo one week before. She was ill-appearing, tachycardic, hypotensive with poor peripheral perfusion. Her physical exam showed non-purulent conjunctival injection, generalized oedema and oliguria, mild lymphadenopathy and a maculo-papular rash in the malar area, trunk and limbs. Blood

exams showed neutrophilic leucocytosis, increase of C-Reactive Protein and Procalcitonin, acute renal injury (AKI), hyponatremia, hypokalaemia, hypocalcaemia, hypophosphatemia, hypoalbuminemia, hyper-CPKemia and coagulopathy. Echocardiogram, abdominal ultrasound and thoracic X-rays were normal. Immediate fluid resuscitation was administered. Progressive deterioration of the patient's hemodynamic status required Intensive Care monitoring and I.V. infusion of dopamine and diuretics. The child was initially treated as Kawasaki Disease (KD)/ Toxic Shock syndrome (TSS) with IVIG and IV clindamycin which improved her conditions. After the resolution of her skin rash and fever, she presented with desquamation of the soles of her feet, of her hands, perianal and oral areas. Bacterial cultures were all negative. As recently reported in the literature, KD can, on rare occasions, present as Kawasaki Shock Syndrome (KSS). Although the acute management of both TSS and KSS is based on supportive treatment and IVIG, recognizing KSS is of utmost importance for optimal long-term patient management.

####

A DOMINANT…TREMOR
UN TREMORE…DOMINANTE

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Early-onset encephalopathy STXBP1-gene related is inherited in an autosomal dominant manner. 95% of patients have epilepsy, often difficult to control, with a non-specific EEG pattern. Most patients have intellectual disability, behavioral and movement disorder. Maddalena was a newborn admitted because of generalized tonic-clonic seizures occuring since day 3 of life, treated with phenobarbital. Metabolic screening laboratory studies and septic work-up were negative. Brain imaging was unremarkable. EEG was characterized by generalized spike-and-slow waves. After discharge Maddalena developed refractory epilepsy and infantile spasms, despite multiple antiepileptic therapies, and her EEG showed hypsarrhythmia. Therapy with levetiracetam, nitrazepam and ACTH effectively managed the seizure episodes. Presently, she is 4 months old and shows a good psychomotor development. Investigation of genes associated with infantile forms of epilepsy identified as de *novo STXBP1* deletion variant (C.1057_1060del). This previously unreported STXBP1 deletion, in a subject with neonatal-onset focal seizures, broadens the spectrum of clinically relevant human disorders STXBP1-gene correleted. Treatment of STXBP1 encephalopathy warrants a multidisciplinary approach, and currently consists of symptomatic treatment of seizures and of behavioral and locomotor problems with physical therapy and occupational therapy to maximize the developmental potential.

####

PYOMYOSITIS: THE DIAGNOSIS LIES IN SUSPECTING IT
PIOMIOSITE: LA DIAGNOSI STA NEL SOSPETTARLA

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Introduction. Pyomyositis is a muscular infection mainly by Staphylococcus aureus and trauma could be the main predisposing factor. Clinical suspicion, supported by Magnetic Resonance Imaging (MRI), is pivotal in order to start endovenous antibiotic therapy precociously. Here we describe three cases.
Case report. All three patients came to

our attention for fever and localized pain without history of trauma. Blood examinations were compatible with acute inflammation.
Blood culture was positive for meticillin sensitive S. aureus, so antibiotic therapy was started precociously and suspended when MRI resulted negative.
M. was 12 years old and he had right lower limb pain. Computed tomography (CT) was negative, while MRI showed an abscess in iliac-obturator region, extended up to gluteal, sacral and pelvic region.
E. was a 5-year-old girl presenting with pain and swelling of right hip. CT and MRI showed abscess at the right hip abductor. Antibiotic therapy and surgical drainage were necessary.
P. was 13 years old and she had pain in the right shoulder. CT and MRI revealed abscess of the right intercostal muscles associated with the homolateral scalene ones, involving the surrounding tissue.
Discussion. Fever, localized pain and a history of trauma are suggestive of pyomyositis: precocious suspicion allows antibiotic therapy to begin as soon as possible. There are no guidelines regarding treatment of pyomyositis, but our clinical experience suggests to continue antibiotic use for at least 3 weeks.

####

HYPERTRANSAMINASEMIA WITH HEPATOMEGALY: LOOK FOR A DIAGNOSIS!
IPERTRANSAMINASEMIA ED EPATOMEGALIA: CERCA UNA DIAGNOSI!

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A 7-year-old male with Gilbert syndrome and autistic behaviors with developmental delay, had occasional isolated mild increase of ALT (85 U/L) confirmed with subsequent controls and associated with hepatomegaly. CRP, ESR, CPK and liver function (gGT, ALP, coagulation test, serum protein electrophoresis) were normal. A persistent and asymptomatic increase in transaminases with normal gGT was part of a pure chronic cytologic syndrome and when associated with hepatomegaly, always requires an in-depth diagnostic approach. Viral hepatitis, endocrinopathies and celiac disease were excluded. There was no hypergammaglobulinemia and ANA, ASMA, LKM were also negative. Ceruloplasmin and alpha1-antitrypsin levels were regular, as well as those of emogasanalysis, ammonium, lactate, and uric acid. Exams performed did not help in the diagnosis, so the clinical features of the child were reviewed: the symptomology including development delay, hepatomegaly, hypertransaminasemia in a child with stunted growth and mild facial coarseness had raised the suspicion of Mucopolysaccharidose. Analysis of the urine analysis revealed a marked increase in heparan sulfate and the diagnosis of MPS III was confirmed by genetic analysis. Hypertransaminasemia is one of the most common laboratory test alterations that pediatricians must to deal with. It is imperative that any child with chronic hypertransaminasemia and hepatomegaly receives an appropriate investigation to help elucidate an etiology, looking for possible rare underlying causes.

####

ABDOMINAL PAIN AND… ALOPECIA
DOLORE ADDOMINALE E… ALOPECIA

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An 11-year-old girl was admitted to a regional hospital referring abdominal pain and lack of appetite. Past medical history revealed weight loss and occasional diarrhea over the past year. On physical examination, she did not appear well and had an area of non-scarring occipital alopecia. Urgent blood tests and abdominal ultrasound were not significant. During the hospitalization, clinical suspicion of trichotillomania was formulated. A CT-scan revealed a gastric trichobezoar that was surgically removed. Additionally, she began psychotherapy for trichotillomania. Two months later she came to our attention for the persistence of intermittent abdominal pain and diarrhea. She denied recent trichophagia. Blood tests revealed IgA anti-transglutaminase >200 U/ml in a subject with HLA-DQ2. She was diagnosed with celiac disease (without duodenal biopsies). She showed a rapid improvement in general conditions from the beginning of the gluten free diet. At one year follow-up, she was asymptomatic with improved weight gain and negative IgA anti-transglutaminase. A few cases of trichobezoar associated with celiac disease have been reported in the literature. Trichotillomania most frequently affects female subjects in the first two decades of life; trichophagia is a form of pica (compulsion to eat non-nutritional substances). The result is the trichobezoar, which causes abdominal pain, malabsorption and intestinal obstruction; very often the bezoar requires laparotomy surgical removal.

####

IPLV EVEN WITH NO DIARRHEA
IPLV ANCHE SENZA DIARREA

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This case resport is of a four-month-old infant, hospitalized for a serious dystrophy. Birth weight 3600 g, exclusive breast milk for one month, then mixed feeding and since the third month infant formula. Good growth during the first month, then a slow-down and a stop since the third. At the anamnesis, gradual reduction of the quantities of milk intake, with desultory vomiting (never bilious) and bowel generally constipated. When the child was admitted, a marked dystrophy came into sight (weight 4380g) with hypodermic essentially absent, globose abdomen, excellent psychomotor development. An abdomen x-ray and scan excluded "obstructive" causes, acid–base balance excluded acidosis, blood count and biochemical profile were normal. Considering the serious dystrophy, a formula feeding for NGT was started, also as ex-iuvantibus. Stool samples for elastase, blood, antitrypsin-1 and eosinophil were collected.
On elementary formula, the child gained weight, vomit disappeared, and three days later oral feeding was resumed with release on the seventh day. At the 20-day follow-up, he had grown (720 grams) and a hydrolysate formula was started. In stool samples, finding showed normal elastase, absent faecal eosinophil, faecal fats and occult blood present and alpha1-antitripsian faecal slightly increased.
Enteropathy by IPLV (today also classified as chronic PFIES) is one of the cause of failure to thrive and dystrophy even without diarrhea.

####

THE IMPACT OF PERTUSSIS IN ITALY: A REAL DECREASING TREND OR ONLY AN UNDERESTIMATED DISEASE?

L'IMPATTO DELLA PERTOSSE IN ITALIA: UN REALE TREND IN DECREMENTO O SEMPLICEMENTE UNA MALATTIA SOTTOSTIMATA?

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A 1-month-old infant was admitted to our hospital suffering from whooping cough, rhinorrhea and difficulty breathing since the previous week.

Physical examination showed nasal flaring, retractions, grunting. Respiratory rate was 30 breaths/min, SpO2 98%. Blood tests revealed leukocytosis (46.970 cells/mm3) with lymphocytosis (>60%); CRP was normal. Chest x-ray detected apical right field thickening, thus Ampicillin was introduced. Later, feeds were started via gavage because of inability of oral feeding. The PCR of nasopharyngeal swab confirmed suspicion of pertussis. Clarithromycin was administered.

Bordetella Pertussis (BP) is a highly contagious disease, that causes significant morbidity and mortality. Recently, there have been large outbreaks of BP in Europe, the United States and Australia. In contrast, Italy has not experienced a resurgence of BP since 2002, when 96% of immunization coverage was achieved. However, data from the statutory notification system indicates that BP is greatly underestimated, likely due to the lack of awareness and recognition by clinicians. A recent Italian seroepidemiological study suggests that BP actually circulates widely among adolescents and adults, because of waning immunity and diminished herd immunity. They may not present typical symptoms of BP, representing a potential source of infection for infants, who are not yet completely immunized. This suggests the need to recommend a primary vaccination course, including a booster in adults.

####

A RARE CAUSE OF PERIUMBILICAL PAIN

UNA RARA CAUSA DI DOLORE PERIOMBELICALE

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G., 12, was admitted in our ED for fever having begun 24 hours prior and abdominal pain, localized in the periumbilical region. Blood tests and abdominal US were negative, but, due to intense colic meteorism, the appendix wasn't found. Due to the persistence of abdominal pain, she was hospitalized. Upon physical examination, the abdomen was treatable, but at the palpation there was a small painful swelling below the umbilicus, on the median line. A second abdominal US showed a cystic mass of about 1x2cm of mixed echogenicity and signs of inflammation of surrounding fat tissues. Surgical consult confirmed the diagnosis of inflammation of urachal remnants. Antibiotic therapy was started, with complete pain relief. A cystouretrography was planned to complete the diagnosis workup. The urachus is a tubular structure that connects the allantois to the dome of the bladder during fetal develop-

ment. Normally, it obliterates completely after birth giving rise to a fibrous cord running from the umbilicus to the dome of the bladder: the median umbilical ligament. Urachal remnants represent a failure in the obliteration process. It can result in several anomalies: urachal cyst (30%), sinus (15%), diverticulum (5%) and a patent urachus (50%).The majority of patients are asymptomatic. However, they may become symptomatic if infection takes place. Complete excision is indicated both in case of persistent symptomatic remnants and asymptomatic ones, because of the associated risk of malignant degeneration.

####

LOBAR EMPHYSEMA IN 7-YEAR-OLD-GIRL: ACQUIRED FORM OR LATE ONSET CONGENITAL FORM?
UN CASO DI ENFISEMA LOBARE IN UNA BAMBINA DI SETTE ANNI: FORMA ACQUISITA O CONGENITA AD ESORDIO TARDIVO?

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A 7-year-old female child was admitted to our centre due to severe respiratory distress, fever and hypoxia. The medical history showed bronchoscopy for foreign body extraction, followed by pneumonia at the age of 2, and thoracothomic surgery for atrial septal defect (ASD) at the age of 4. Treatment with antibiotics, bronchodilators, oxygen and steroids was partially successful. Chest angio-CT showed a large air cyst (62 mm x 48 mm) with thin and regular wall, compatible with Lobar Emphysema (LE). Surgical resection was performed with progressive clinical improvement and without any complications. Histological examination confirmed the diagnosis of LE. Congenital Lobar Emphysema is a rare disease, characterized by lobar over-distension, leading to compression and displacement of adjacent normal lung tissue. Congenital heart disease may be found in 12–20% of cases. Respiratory symptoms usually appear during the first 6 months of life. Acquired Lobar Emphysema is less frequent and may be a complication of foreign body inhalation or airways inflammation. First diagnostic hypothesis is based on chest X-ray. Chest CT scan or MRI confirm diagnosis and are essential to choose the correct surgery strategy. The management of LE in symptomatic children is surgical resection. In our case the etiopathogenesis is probably acquired due to the late onset of the symptoms, ascribing foreign body extraction followed by pneumonia and thoracotomy for ASD which are causal factors.

####

THE DARK SIDE OF LEUCINE
IL LATO OSCURO DELLA LEUCINA

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Introduction. 3-Methylcrotonyl-CoA carboxylase deficiency (MCCD) is an autosomal recessive inborn error of leucine catabolism. In MCCD, an elevated urinary excretion of 3-hydroxyisovaleriate (3-

HIV) and 3-methylcrotonylglycine (3-MCG) is often present, and there is sometimes a secondary carnitine deficit. The clinical presentation can be asymptomatic or associated with neurologic abnormalities and death.
Case report. S., an Asian female born at term, was admitted at 6 days of age because of elevated C50H detected by newborn screening programs. Family history: parents are first cousins in good health, while her sister has a delay in psychomotor development. Clinically, S. showed poor sucking and hyporeactivity. Laboratory tests: hyperammonemia and increased urinary excretion of 3-HIV and 3-MCG. Discharged without therapy, after a week she was hospitalized again because of seizures. S. began therapy with phenobarbital, low-protein diet, biotin and carnitin, with success. Family compliance was poor - after 3 days she presented again with repeated seizures controlled by midazolam. Resolution was achieved after several days of therapy (drugs and diet). Currently she is 15 months old and seizure-free (therapy: phenobarbital, biotin and carnitine), but has delays in psychomotor development.
Conclusion. Molecular analysis revealed omozygosity for MCC1-H476P, not previously described. Open questions: How does consanguinity influence phenotype? New mutation: does a genotype-phenotype correlation exist? Use of biotin/carnitin: is it rational?

####

GREY MACULES ON THE SKIN: WHAT ARE THEY?
CHIAZZE GRIGIASTRE SULLA CUTE: DI COSA SI TRATTA?

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Erythema Discromicum Perstans or Ashy Dermatitis is an acquired idiopathic disorder caracterized by ovalar grey macules. They commonly affect the body in the order of trunk, face, arms and neck, with symmetrical distribution. Frequently, it occurs in patients with skin type III and IV and without any sistemic signs and symptoms. Diagnosis is clinical and histopathological. Most cases resolve themselves over many years.
A 9-year-old girl presented with blue-grey lesions to the trunk, neither itchy nor sore, that appeared 7 months prior. No clinical history of recent infection, bleeding or trauma. Upon examination, many hypermelanotic grey macules were observed on the trunk and arms. No signs of scratching. Cardiac-thoracic-abdominal examination was normal. Because of the clinical suspicion of grey macules due to phthiriasis, permetrin therapy was started. Since the lesions had grown, blood tests with coagulation testing were performed and resulted normal. Assuming Ashy disease, skin biopsy was executed and it confirmed the suspected diagnosis.
Our clinical experience suggests that Ashy disease can often be confused with: phthiriasis (cerulean macules in gluteal-lumbar-sacral region, nits and lice to the eyelashes); bruising from maltreatment (generally accompanied with other violence signs); coagulation and aggregation disease (petechiae and other bleeding signs); mongolian spot (slate grey macules confluent to buttocks and shoulders present at birth).

####

IDIOPATIC EOSINOPHILIC PNEUMONIA, A PAEDIATRIC CASE
PNEUMOPATIA EOSINOFILICA EREDITARIA, UN CASO PEDIATRICO

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Eosinophilic lung disease is a group of heterogeneous disorders divided into secondary forms and rare idiopatic forms (acute eosinophilic pneumonia, chronic EP, idiopathic hypereosinophilic syndrome). A 12-year-old asthmatic girl was admitted to Tirane Hospital for severe respiratory distress. Chest x-ray showed bilateral multiple thickening areas with mediastinic pleural involvement confirmed with CT scan. Based upon clinical and radiological reasoning, the worsening of symptoms and nonresponsiveness to antibiotics and steroids, she was transferred to the Pediatric Unit. Upon admission, breathing sounds were slightly decreased. The first blood samples, arterial EGA and CBC, were all normal. Chest x-ray showed multifocal and circumscribed bilateral pulmonary areas of consolidation with lamellar aspect, circumscribed ground glass opacities on CT scan. Infectivological investigations and the sweat test were negative. We repeated CBC with the unexpected finding of hypereosinophilia, confirmed by microscopic peripheral blood examination (37%). We extended the investigation panel to exclude all possible causes of secondary pulmonary hypereosinophilia. Bronchoscopy with BAL examination was performed showing hypereosinophilia (38%). These findings, with the characteristic radiological ground glass pattern and the persistence of symptoms for about 5 weeks were suggestive of ICEP. We started therapy with a high dose of prednisone for 2 months and slowly tapering off over 6 months, until radiological and clinical normalization resulted.

####

A CASE REPORT OF MYCOPLASMA PNEUMONIAE ASSOCIATED MUCOSITIS (MPAM): AN ATYPICAL STEVEN JOHNSON SYNDROME?

UN CASO DI MYCOPLASMA PNEUMONIAE ASSOCIATE A MUCOSITE: UNA STRANA SINDROME DI STEVEN JOHNSON?

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A 6-year-old boy was admitted with feverish bronchitis not responding to amoxicillin, and wide oral ulcers preventing feeding. On examination: painful ulcerations covered with yellowish serofibrinous exudates on oral mucosa, lips and tongue. Blood examinations: leukocytosis (WBC 16.000/ml), neutrophils 78%, CRP 7.7 mg/dl. He was treated with intravenous hydration and clarythromicin infusion, aerosol therapy with salbutamol and ipratropium. On day 2, bronchitis clinically improved, but increasing oral ulcers were present as well as the occurrence of deep bilateral serous conjunctivitis, genitalia skin ulcers, urinary pain and hematuria. All of these features clinically indicated Steven Johnson syndrome without skin affection, as described in MPAM. Serum enzyme immunoassay confirmed the diagnosis revealing M. Pneumoniae IgG and IgM. In the literature, no data was found about treating children for MPAM, and there was no strong evidence regarding the effectiveness of IVIG or steroid therapy. We decided to treat him with methylprednisolone (2 mg/kg/die) for 5 days. We noticed rapid improvement of mucosal lesions, so we started tapering on day 12. Conjunctivitis was treated with azythromicin eye-drops. According to data

showing clarythromicin as a possible role in SJS, it was safer to replace it with ciprofloxacin (20 mg/kg/die). We noticed a remarkably fast improvement of clinical conditions and the boy was discharged on day 7. Complete healing of the lesions and recovery were achieved on day 15.

####

BULLSEYE: ERYTHEMA MULTIFORME
ERITEMA MULTIFORME: BERSAGLIO COLPITO

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Erythema multiforme (EM) is an uncommon immune-mediated vesiculobullous skin disorder, characterized by typical target-like lesions, with or without mucosal involvement. Herpes simplex virus (HSV) is the most common trigger etiology.
An 11-year-old girl presented with fever and vesiculobullous lesions with an erythematous border on her trunk, arms and legs. She also presented conjunctivitis and ulcerated bullae on her lips and oral mucosa, affecting her oral intake. These lesions developed twelve days after a herpetic cutaneous infection on her left shoulder, which was treated with a full course of oral Aciclovir. As EM major was suspected, the child received oral prednisone, as well as supportive treatment (pain relief and intravenous hydration). HSV serology was positive for recent infection.
Few studies support the use of corticosteroids in the treatment of EM. In this case, the patient promptly responded to steroid therapy and progressively improved. Although differentiation from Stevens-Johnson syndrome can be difficult, particularly in early stages, EM is more often caused by infections and has a better prognosis. Since the diagnosis is clinical, it is important to identify early clues of EM, through a detailed history and a thorough physical examination, in order to start a supportive and etiology-related treatment as soon as possible.

####

MY SKIN IS BLUE: A STRANGE CASE OF DYSPNEA
LA MIA PELLE È BLU: UNO STRANO CASO DI DISPNEA

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R, an 8-years-old child, came to our centre with a history of dyspnea. Her anamnesis evidenced only recurrent upper airway infections since she was 2 years old and an IgA deficiency.
Clinical examination revealed perioral cyanosis, clubbing, and a small teleangectasia on the face. Her vital signs were: respiratory rate of 24 breaths/minute, oxygen saturation of 90% on room air and blood pressure in a normal range for age and sex. Her blood sample showed polycythemia with elevated haemoglobin, white blood cells and platelets in range.
Chest radiography showed a suspected vascular ectasia in the right middle lung lobe. Then, a pulmonary arteriovenous malformation (PAVM) was suspected. In order to confirm the diagnosis, she underwent a complete cardiologic evaluation including a transthoracic contrast echocardiography, which resulted positive for an intrapulmonary shunt. Then, we performed a contrast chest CT scan, which revealed a high-flow PAVM on the right lower lobe.
A subsequent genetic investigation diagnosed hereditary hemorrhagic telangectasian (HHT) in the child.
PAVMs are arteriovenous malformations that result in an intrapulmonary right-to-left shunt, bypassing normal circulation and connecting a pulmonary artery to a pulmonary vein. Depending on the degree of the shunt, the major symptoms

are dyspnea, cyanosis and hypoxemia. PAVMs could be idiopathic post-infective, but between 80-90% occur in patients with HHT.

####

A RARE NEONATAL WATERY STOOL
UNA RARA FORMA DI DIARREA ACQUOSA NEONATALE

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This case report is of a probable lower intestinal obstruction. At 32^weeks' gestation prenatal ultrasonography showed fetal bowel dilatation, from rectum to small intestine, polyhydramnios and honeycomb sign, common for anorectal malformation or Hirchsprung disease. Parents were consanguineous (cousins). At birth, plain abdominal x-rays of the newborn revealed normal bowel gas, so intestinal obstruction was ruled out. There was no meconium emission, only watery stool (like urine). Considering family consanguineity, prenatal history and clinical findings, Congenital secretory Chloride Diarrhea (CCD) was suspected, and stool chlorine dosage was performed. It revealed high chlorine levels, confirming CCD diagnosis. Genetic exams were underway. A repleacement therapy with satisfactory electrolites equilibrium was initiated. For the watery stool, proton pomp inhibitors were started, but with no improvement. According to recent literature, we administered Short Chain Fatty Acid and Sodium butirrate (SB), and diarrhea became less frequent and stools more compacted. CCD is a chronic diarrhea characterized by excretion of watery stool high in chlorine, dehydration, hypokalemia and metabolic alkalosis. It needs an adeguate replacement therapy, and SB can be useful. In conclusion, when ultrasound reveals signs of fetal intestinal obstruction, not confirmed at birth, with watery stools, one should consider CCD and start therapy as soon as possible. If the condition is diagnosed early, the prognosis will improve.

####

HEPATOSPLENOMEGALY IN A 3-MONTH-OLD CHILD WITH "MILKY" BLOOD
EPATOSPLENOMEGALIA IN UNA BAMBINA DI 3 MESI CON SIERO LATTESCENTE

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A 3-month-old child of consanguineous parents was referred for evaluation of hepatosplenomegaly (HSM). There was a family history of dyslipidemia. At admission: height, weight, head circumference were within the 50th pc. Physical examination was negative with the exception of a distended abdomen with palpable liver and spleen. The abdominal US confirmed the HSM and revealed a hyperechogenic liver. A blood sample showed a "milky" serum. Laboratory tests revealed: cholesterol 105 mg/dl, HDL-cholesterol 16 mg/dl, triglycerides (T) 792 mg/dl, GOT 128 U/l, GPT 95 U/l. Thyroid, kidney and pancreatic functions, alkaline phosphatase, serum electrolytes and glucose were all within normal range. Gaucher disease, lysosomal acid lipase deficiency, mucopolysaccharidosis were ruled out. Finally, a presumptive diagnosis of familial chylomicronemia (FC) was made and the analysis of the candidate genes was requested. She was started on a low-fat formula and T rapidly decreased (159 mg/dl). She did not have any disease-related complications. FC is a rare recessive disorder, due to lipoprotein lipase (LPL) deficiency or its cofactors. Clinical features include failure to thrive, eruptive xanthomas, lipemia retinalis, hepatosplenomegaly, recurrent abdominal pain and episodes of acute pancreatitis. In infants, hypertriglyceridemia is often an incidental finding during routine laboratory tests. Low-fat diet is usually sufficient, while pharmacotherapy plays a less important role.

####

SHOULD LAB TESTS FOR MYCOPLASMA PNEUMONIAE (MP) ALWAYS HAVE A CLINICAL IMPACT IN CHILDREN WITH COMMUNITY ACQUIRED PNEUMONIA?
I TEST DI LABORATORIO PER MYCOPLASMA PNEUMONIAE (MP) DOVREBBERO SEMPRE AVERE UN IMPATTO CLINICO NEI BAMBINI CON POLMONITE ACQUISITA IN COMUNITÀ?

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A 7-year-old boy with a 2-day history of fever and left-sided pleuritic chest pain presented at our hospital with mild cough, slightly reduced SaO2 and tachypnea. Other vital signs were normal. PE revealed superficial breathing with reduced TVF, increased dullness and decreased breath sounds in the left lower lung fields. PA and lateral chest x-rays noted a blunt left costophrenic angle. Lab tests showed polymorphonulear leukocytosis and CRP elevation. Ultrasound scan identified pleural locular fluid. MP IgM antibody titre was slightly increased (12) as compared to reference lab results (<10). IV ceftriaxone was initiated and his clinical conditions improved on day 1. He was discharged on day 6, switching to oral therapy with cefixime. Two weeks after admission, specific MP IgM title was confirmed (12).
The detection of serum antibodies against MP still has some limitations because of the existence of long-term carriers. The patient had a significant clinical response to empirical treatment. Considering MP antibodies borderline results and the patient's clinical improvement, specific treatment for MP was not initated. The patient's clinical course and serological reassessment confirmed it was an adequate approach. This case underlines once more how clinical evaluation continues to hold a privileged role in deciding treatment, but is it always secure not to use a safe antibiotic effective on MP infection with positive, or even without, lab results?

####

KARTAGENER SYNDROME : A CASE REPORT
SINDROME DI KARTAGENER : UN CASO CLINICO

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A 5-year-old child was hospitalized for dyspnea and cough, suggestive of pneumonia. An accurate history showed a dextrocardia diagnosed at birth and a chronic catarrhal cough with mucopurulent expectoration that appeared in the first year of life. An abdominal ultrasound showed a situs inversus viscerum and a chest CT scan detected bronchiectasis. Studying the mucociliary clearance and ultrastructural examination of cilia revealed pathological findings, confirming the suspicion of a primary ciliary dyskinesia. Kartagener Syndrome is a phenotype of the ciliary dyskinesia, characterized by the triad situs inversus viscerum, bronchiectasis and nasal polyps. Currently, our patient is six years old and he is well, being treated with antibiotics, inhaled steroids and bronchodilators, and respiratory physiokinesitherapy.
Primary ciliary dyskinesia is a rare genetic disease characterized by structural and functional abnormalities of cilia. An altered mucociliary clearance is responsible for chronic respiratory symptoms and other symptoms with considerable phenotypic variability.
This case highlights the importance of careful patient history recording and physical examination when there is a presence of chronic cough, to make a differential diagnosis.

####

A STRANGE CASE OF CYTOPENIA… ALPS!
UNO STRANO CASO DI CITOPENIA… ALPS!

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A 9-year-old boy, previously diagnosed with celiac disease, was admitted at our pediatric hospital for asthenia and fever. At admission, he appeared extremely pale. A complete blood count (CBC) revealed a severe cytopenia: PLT4000/mmc, Hb 6,2g/dl, WBC 1510/mmc(N 290/mmc). Infectivological tests were negative but Coombs tests and anti-neutrophil antibodies were positive. Bone marrow aspiration excluded leukemia and FISH for MDS abnormalities was negative. Evans syndrome was suggested for the patient so corticosteroids plus Intravenous Immunoglobulin was started. Clinical and laboratoristic remission were observed and a follow-up program was required. However, during the tapering of prednisone, CBC decreased, so Cyclosporin was started, but the patient did not improve. After the failure of common therapies (PDN, HDIg, CyA), laboratory tests were repeated. The lymphocyte subsets reported increased numbers of double-negative T cells (CD3+TCRαβ+CD4-CD8-) so the diagnosis of Autoimmune lymphoproliferative syndrome (ALPS) was confirmed for the patient. Mycophenolate therapy was successfully started resulting in CBC normalization. ALPS is a disorder of the lymphoid system regulation (defect in lymphocyte apoptosis) characterized by splenomegaly, lymphadenopathy, cytopenias and other autoimmune diseases. The diagnosis was based on clinical findings and laboratory abnormalities: abnormal biomarker testing IL-10, FasL, IL-18, and vitamin B12; somatic o germinal mutation in FAS, FASL or CASP10; elevated levels of DNTs(≥2.5 %).

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FREQUENT REGURGITATION AND HEPATOMEGALY IN A BREASTFEEDING INFANT: AN UNEXPECTED DIAGNOSIS
RIGURGITI FREQUENTI E EPATOMEGALIA NEL LATTANTE: UNA DIAGNOSI INATTESA

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M. was sent for a consultation from her Pediatrician at the age of 4 months, after discovering hepatosplenomegaly and gastroesophageal reflux symptoms. Physical examination findings were significant for the presence of visceromegaly, mild hypotonia and frontal bossing. Laboratory tests revealed elevated cytolysis markers in the absence of liver function alteration, negative infectivological investigations, and normal values of ammonia levels and pre- and post-prandial blood glucose levels. Abdomen Ultrasound confirmed the hepatomegaly with hyperechoic structure and diffuse moderate periportal spaces hyperechogenicity and splenomegaly with uniform hyperechoic structure. A full cardiologic exam was normal, as were the audiometric, ocular and orthopedic evaluations. Metabolic tests detected the absence of sphingomyelinase activity on leucocytes and a significantly high level of chitotriosidase activity. The diagnosis of lysosomal storage disease, namely Niemann-Pick disease, was made and confirmed by genetic investigation which resulted positive for two pathogenetic mutations of SMPD1 gene, defining the disease as type A/B. The clinical course was characterized by a rapid deterioration of liver function with ascites, coagulopathy and worsening jaundice. The exitus occurred at the age of 8 months for an Acinectobacter Baumanii Complex sepsis, which led to a multiorgan failure and ended with a fatal metabolic imbalance.

PERSISTENT DYSPNOEA IN A ONE-YEAR-OLD CHILD
DISPNEA PERSISTENTE IN UN BAMBINO DI UN ANNO

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A one-year-old child was admitted to hospital due to the sudden onset of cough, dyspnoea and fever. On physical examination, he had tachycardia and tachypnoea with severe respiratory distress. Blood tests showed neutrophilic leukocytosis, increase in CRP and positive Adenovirus serology with a left lower lobe consolidation in the chest x-ray. He was treated as a pneumonia patient with antibiotics. At discharge, after two weeks of treatment, signs of respiratory distress persisted. Recurrent and worsening dyspnoea was observed over a two-month period with repeated admissions. For this reason, he was referred to our bronchopneumology unit. When he arrived, his general conditions were poor with hyperinflation of the chest, widespread crepitations and rhonchi. A chest-X-ray showed signs of interstitial infiltrate, peribronchial thickening and areas of subsegmental atelectasis. Cystic fibrosis, tuberculosis, congestive heart disease, immunodeficiency, α-1 antitrypsin deficiency were excluded. A high resolution chest-CT identified mosaic perfusion, air trapping, vascular attenuation, bronchiectasis and peribronchial thickening. The clinical presentation along with the previous positivity for Adenovirus serology and the typical radiological pattern led to the diagnosis of bronchiolitis obliterans. The condition was treated with steroids, azithromycine and oxygen and the patient showed gradual improvement.

####

FRIDAY NIGHT FEVER
LA FEBBRE DEL VENERDÌ SERA

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A 15-year-old boy had periodic fever associated with thoracic and abdominal pain during the last six months. Fever episodes presented each seven days and lasted about 48 hours. At the first visit, the patient was febrile (TC 38°C) and reported pain in the sternum-collarbone joints. The blood tests showed increase of white blood cells (WBC 10820/mm3, N 80%, L 10%, M 9%), and inflammatory indices (RCP 13.4 mg/dl, ESR 32 mm/h, SAA 513 mg/l). Throat swab and urine analysis were negative. A bowel wall thickening was excluded through abdominal scan. Two-dimensional echocardiography showed a pericardial and left pleural thickening. This febrile episode was treated with paracetamol and disappeared after 48 hours. When he was asymptomatic, blood tests were again performed, both WBC and inflammatory indices were normal. The patient experienced other febrile episodes with: abdominal and thoracic pain, lateral cervical lymphadenopathy and arthritis of the ankle with spontaneous remission. For these clinical and laboratory findings, Familial Mediterranean Fever was suspected, so his DNA was analyzed to detect the MEFV gene. Analysis showed the presence of homozygous mutation of c.2080A>G p.Met694Val that encodes mutated protein pyrin, an important player in the innate immune system and the component of inflammasome. The patient started therapy with colchicine (1 mg/die) and immediately recovered with a significant improvement in the quality of life.

####

MALARIA CAUSED BY P. FALCIUPARUM COMPLICATED BY ACUTE ACALCULOUS CHOLECYSTITIS (AAC)
MALARIA DA P. FALCIPARUM COMPLICATA DA COLECISTITE ACUTA ALITIASICA

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Introduction. AAC is a rare complication during acute malaria. It has been more commonly reported in adults, in association both with P. Falciparum and P. vivax infection. Case report. A 13-year-old girl was admitted to Chiulo Hospital (Angola), for headache and high fever over the last 4 days. On physical examination, she was pale, restless, the liver was enlarged and diffuse abdominal tenderness was noticed. A rapid diagnostic test for P. Falciparum was positive. IV quinine was started. Her general conditions progressively improved. However, after 4 days, she was still febrile, abdomen was still painful, mainly at the right upper quadrant, Murphy sign was positive, Blumberg was negative. Abdominal US revealed a distended gallbladder with biliary sludge inside and pericholecystic fluid. ACC was diagnosed. IV Ciprofloxacin combined with Metronidazole were added. After 2 days, fever and vomiting subsided and abdominal pain disappeared. On the 5th day, she was discharged with oral Ciprofloxacin and Metronidazole, to complete a 10-day course of antibiotic. Sickle cell test was negative. Comments. ACC is a rare condition among children but should be considered in the differential diagnosis of abdominal pain. Acute malaria infection is an uncommon risk factor for AAC, but under reported in low-income countries. The ultimate diagnosis of AAC mainly relies on ultrasound.

####

ACUTE RHEUMATIC FEVER AND CHOLECYSTITIS: WHAT IS THEIR RELATIONSHIP?
MALATTIA REUMATICA E COLECISTITE: QUALE RELAZIONE?

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Acute cholecystitis is uncommon in children and is usually caused by infection. Pathogens include Streptococci (groups A and B) and Gram-negative organisms.
T., an 8-year-old male, was admitted to our department with severe fatigue, diffuse osteoarticular pain and subcutaneous nodules. Three weeks before, he had been recovered for acute cholecystitis of unknown aetiology, treated with antibiotic therapy.
Because of the important osteoarticular pain, which was unresponsive to common analgesic drugs, onco-haematological diseases were excluded after performing bone marrow aspirate and biopsy, total body scintigraphy and abdominal ultrasound, all of which were negative.
At clinical examination, a systolic apical murmur was detected, which had previously gone undetected, and echocardiography revealed a mild mitral valve regurgitation. Subcutaneous nodules were firm, symmetric, painless, and located over extensor aspect of elbows, knees, ankles and fingers. The overlying skin was not inflamed. Haematological exams showed significant ESR elevation and rising antistreptolysin O (ASLO) and antideoxyribonuclease B (aDNAsi-B) titers.
Based upon the Jones Criteria 2015, the diagnosis of Acute Rheumatic Fever (ARF) was determined: T. presented two major criteria (carditis, subcutaneous nodules) and two minor criteria (arthralgias, elevated acute phase reactants). It can be hypothesized that ARF was a result of (streptococcal) cholecystitis.

####

IF NEONATAL JAUNDICE PERSISTS
SE L'ITTERO NEONATALE PERSISTE

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R, a male, preterm born to non-consanguineous parents, was referred at 2 months old to our department for investigation of persistent jaundice with con-

jugated hyperbilirubinemia, which appeared at the 4th day of life, supposedly caused by parenteral nutrition and treated with UDCA. Blood exams showed hyperammonemia, hypertransaminasemia with normal γ-GT, high levels of PA, hyperlactatemia and reduced levels of fat -soluble vitamins. Coagulation tests and albumin levels were normal. Coombs test was negative. Urinary bile acid profile was consistent with cholestasis. Lysosomal acid lipase levels were normal. Abdominal US showed no hepatobiliary lesions. Liver tissue histology revealed canalicular cholestasis, portal fibrosis and giant cell hepatitis, while immunohistochemistry showed negative immunoreaction of BSEP (Bile Salt Export Pump). Considering these data, on suspicion of PFIC (Progressive Familial Intrahepatic Cholestasis), genetic analysis was performed confirming 2 heterozygotic mutations in ABCB11 gene, causing PFIC2. This disease was characterized by: its early onset, no extrahepatic signs, rapid progression to liver failure, increased risk of hepatobiliary carcinoma, and by the clinical-lab findings we described above. The patient is now receiving UDCA therapy and supplementation of fat-soluble vitamins and MCT. Some patients may also benefit from surgical biliary diversion. If liver failure or cirrhosis occurs, transplantation represents the only effective therapy.

####

HYPEREOSINOPHILIA AND ATOPIC DERMATITIS IN AN INFANT: FOOD ALLERGY OR PRIMARY IMMUNODEFICIENCY SYNDROMES?

IPEREOSINOFILIA E DERMATITE ATOPICA IN UN BAMBINO: ALLERGIA ALIMENTARE O SINDROME DA IMMUNODEFICIENZA PRIMARIA?

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A four-month-old infant was admitted to our department due to a recent history of severe atopic dermatitis and edema. He was born at 36 gw, birthweight 4.285 kg. Fed with breastmilk and formula. Regular growth at 50th percentile. When admitted, he presented with extended impetiginized eczema, edema, and reduced diuresis. Blood exams were performed evidencing leucocitosis with hypereosinophilia, hypoalbuminemia, IgG deficiency and hyper-IgE. He had normal heart, liver and kidney function and resulted negative to infectivological screening. Suspecting cow's milk protein allergy, serum specific IgE were performed resulting positive (class I).

Considering the previous findings, primary immunodeficiency syndromes with hypereosinophilia were also suspected (quantitative serum Ig levels, isohemagglutinins, lymphocytes proliferation assays, genetic tests, skin biopsy) which, after being investigated, resulted negative.

The patient was treated with albumin and furosemide, systemic steroid, antibiotics and antihistamines and a free amino acids formula was started. This resulted in an improvement of his general condition as well as the atopic dermatititis.

When weaning was started at the age of 6 months, the patient presented a worsening of the eczema when wheat was introduced. RAST for the most common allergens were performed resulting positive for wheat, fish, eggs and cow milk, so a hypoallergenic diet was prescribed.

In conclusion, food-allergy is a relatively common condition in children, but primary immunodeficiency syndromes should be ruled out in patients with severe eczema and hypereosinophilia combined with hyper-IgE and IgG deficit.

####

LATE PRESENTATION OF DOUBLE AORTIC ARCH IN SCHOOL-AGE CHILDREN

IL RAGAZZO DEGLI ANELLI

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A 14-year-old male presented with thoracic pain and respiratory difficulty during physical activity at school. At hospital admission, he was awake, oriented and with normal vital signs. He also had hand paresthesia and dry mouth. He referred five similar episodes over the past year. A week before, during a sport medical visit, they talked about abnormalities detected on the flow-volume curve, but medical records were missing. The boy never suffered from asthma or wheezing. He was treated for gastroesophageal reflux, without benefits for recurrent epigastralgia. At physical examination, the boy was worried about his status, but appeared in good general condition. Ha had antalgic breath, without evidence of respiratory distress or abnormal sounds. ECG was normal, chest radiograph disclosed a mediastinic enlargement toward the right part, spirometry revealed an obstructive ventilatory defect, the flow-volume curve showed plateaus on both inspiratory and expiratory. This combination of findings prompted further chest imaging investigations with computer tomography scan, which revealed a double aortic arch and lusory right succlavian artery. The subsequent surgical treatment resulted in resolution of his respiratory symptoms. Vascular rings can cause respiratory symptoms that usually result in prompt diagnosis during early infancy. However, the diagnosis can be delayed for many years and it could be addressed by the analysis of the flow-volume curve.

####

SEIZURES IN AN ANEMIC NEWBORN BORN AT TERM: A CASE OF CEREBRAL VENOUS SINUS THROMBOSIS

CONVULSIONI IN NEONATA A TERMINE CON ANEMIA: UN CASO DI TROMBOSI DEI SENI VENOSI CEREBRALI

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Alberta is a term baby born from an urgent cesarean section due to a non-reassuring CTG (Apgar 8-9). After 2 days of wellness, anemia (Hb: 9.3 g/dL) without other clinical signs was discovered and oral supplementation was started. At day 7, jerks of the left arm with ipsilateral head deviation appeared. An ictal EEG revealed rhythmic S/PS-W complexes over the right hemisphere. Phenobarbital (PHB) load and maintenance was administered. Diffuse periventricular hyperintensity was found by ultrasound, so a brain MRI and MRV was performed, showing a cerebral venous sinus thrombosis (CVST) of the deep cerebral veins with right thalamus edema. The patient received enoxaparin therapy and laboratory work-up was completed with thrombophilia screening, resulting negative.
After 1 month, the MRV showed a near-complete regression of the thrombus. Seizures were well controlled by PHB and neuropsychological follow-up is ongoing.
CVST is a rare but important cause of morbidity and mortality in neonates. Complicated delivery and anemia may be risk factors. Seizures and/or encephalopathy are the most common clinical presentations.
MRI and MRV are pivotal for the diagnosis and prognosis. Internal cerebral veins, as in this case, are less frequently involved. In newborns, anticoagulant therapy is suggested in association with supportive and symptomatic measures, also to prevent recurrences, but a standardized approach is still missing. Long-term outcomes include developmental delay and epilepsy.

####

A CASE OF NEURORETINITIS CAUSED BY VARICELLA-ZOSTER VIRUS REACTIVATION

UN CASO DI NEURORETINITE DA RIATTIVAZIONE DI HERPES VARICELLA-ZOSTER VIRUS

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An 11-year-old boy was visited for impaired vision in the left eye, conjunctival injection and painful eye movement lasting for 2 weeks. Topic atropine and steroid/cloramfenicol treatment previously started was ineffective. No skin lesions were noticed. Clinical history reported chickenpox at 15 days of age and thoracic herpes zoster at 5 years. Basal exams were normal. Toxoplasma, herpes simplex and cytomegalovirus antibodies (Ig) resulted negative as well as chest x-rays and tuberculin skin test. Varicella-zoster virus (VZV) IgG were positive while IgM were negative. Ophthalmologic evaluation showed neuroretinitis and chorioretinitis of left eye. Diagnosis of VZV reactivation involving ophthalmic branch of the trigeminal nerve was confirmed by detecting of VZV-DNA in aqueous humor. The patient completed a 14-day regimen of i.v. Aciclovir (10mg/kg/die) and Prednisone followed by 6-weeks of oral treatment with Valaciclovir (1g q8h). A total of 2 months of topical treatment (atropine and prednisone eye drops) was performed. Ophthalmologic controls demonstrated a good recovery.

Neuroretinitis associated with herpes zoster (HZ) sine herpete is a rare condition that should be considered in children presenting with worsening vision. VZV infection during gestation or the first year of life is the major risk factor for childhood onset HZ because the infant is protected by transplacentally transferred maternal VZV-Ig but is unable to develop adequate cellular and humoral immunity to VZV.

####

SEVERE JAUNDICE IN A NEWBORN AFFECTED BY HEREDITARY SPHEROCYTOSIS

ITTERO SEVERO IN NEONATa AFFETTO DA SFEROCITOSI EREDITARIA

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A 3 kg female was born at 37 GA. She developed severe and persistent jaundice within the first day of life despite phototherapy. Investigations revealed high indirect bilirubin, anemia, negative DAT, normal thyroid function and G6PD activity. A careful review revealed maternal history of hereditary spherocytosis (HS). Blood smear and osmotic fragility test were performed revealing the presence of spherocytes and low erythrocyte resistance and a HS diagnosis was made. On the 14th day rEPO was started. Phototherapy was stopped on the 10th day. HS is a type of hemolitic anemia in which abnormalities of erythrocyte structural proteins lead to poorly deformable red blood cells with a shortened life span. Jaundice is the most common presenting feature of HS in neonates. Diagnosis is often delayed: more than one-half of neonates with HS are not anemic in the 1st wk of life, spherocytes are observed only on 1/3 of blood smears and osmotic fragility test often shows false negative response. Treatment is supportive with phototherapy and blood transfusion. Early administration of rEPO seems to be effective in reducing blood transfusion requirement in the first year of life. In this case, we made early diagnosis based on family history of HS, confirmed by blood smear and osmotic fragility test, which allowed us to begin rEPO early. Today, at 4 months old, the baby has required only 2 transfusions and she has stable hemoglobin levels. In cases of severe neonatal jaundice, HS is an important cause to be considered.

####

CHEST PAIN IN A CHILD AFFECTED BY HENOCH-SCHÖNLEIN PURPURA

DOLORE TORACICO IN UN BAMBINO CON PORPORA DI SCHÖNLEIN-HENOCH

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G.M. is a 14-year-old boy affected by Henoch-Schönlein purpura and melena treated with oral steroids. Subsequently, he had right chest pain and a right upper lung density so he was treated with parenteral antibiotic therapy and the thickening resolved. Two days later, the right chest pain reappeared associated with a "metallic" cough. The new radiography showed right tension pneumothorax (PNX) associated with controlateral displacement of the mediastinal structures. A pleural drain was inserted and the PNX improved. After 48 hours, the drain was clamped but the PNX impaired. The chest CT showed a pseudonodular density on the right upper lobe with air content, communication with the pleural cavity, and an apical right PNX. A chest drain was inserterd for aspiration for 72 hours with clamping, and it was removed after 48 hours based upon the normalization of the radiography.
DISCUSSION: Necrotizing pneumonia (NP) is rare in children but it may be associated with bronchial pleural fistula (BPF) with PNX, empyema and septic shock. The chest CT is the gold standard examination for diagnosis. Hoffer et. al. hypothesized that BPF is due to the rupture of the visceral pleura adjacent to the necrotic lung parenchyma.
Several studies have shown association between the use of anti-inflammatory agents, steroids and the development of complications in community-acquired pneumonia.
We hypothesize that the use of steroids has facilitated the emergence of NP resulting in BPF and PNX.

####

ARTHRITIS AND BARBECUE: WHAT IS THE CONNECTION?
ARTRITE E BARBECUE: QUALE LEGAME?

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A 16 year-old boy was admitted to our unit for fever, bloody diarrhea and swelling of the left knee. Fever and diarrhea started seven days before admission; left knee joint arthritis and cervical pain developed 5 days later. On admission, the knee joint was warm and swollen with left elbow and ipsilateral midfoot arthritis. Laboratory tests showed normal white blood cells count with increase of C-reactive protein (13, 75 mg/dl). Empiric Ceftriaxone therapy and Ibuprofen were started. Ultrasounds showed left knee effusion. Fecal calprotectin was weakly increased. Autoantibodies, blood and urine cultures were negative. On the fourth day, stool cultures became positive for Campylobacter and the patient reported he had barbecue some days before. Azithromycin oral therapy was started with improvement in clinic and blood exams. After 3 weeks, on the follow up visit, joint pain was absent and mobility recovered. In this case, first suspicions were: septic arthritis or bowel inflammatory disease (IBD). Considering the occurrence of oligoarthritis and diarrhea, the hypothesis of IBD and Reactive Arthritis (RA) increased. In differential diagnosis, first line tests are: complete blood count, sierologies, blood and stool cultures, fecal calprotectin and autoimmunity asset. In conclusion, intestinal infections related to food poisoning such as Campylobacter should be considered as causes of RA.

####

PERSISTENT CRYING IN AN INFANT: A SIGN OF TROUBLE?
PIANTO PERSISTENTE IN UN BAMBINO: UN SEGNO DI PREOCCUPAZIONE?

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A 4-month old infant presented to a first-level Emergency Department with vomit and inconsolable crying. An abdominal ultrasound showed a probable intestinal intussusception, hence the patient was transferred to a pediatric hospital. At admission he was afebrile, with normal vital signs, a soft abdomen and active bowel sounds, but still plaintive and persistently crying. Abdominal ultrasound showed hyperechogenic fat in the right quadrant and no signs of intestinal intussusception. Abdominal X-ray resulted normal. Two days after the initial evaluation, the patient became febrile, with leukocytosis, elevated CRP and PCT, and negative blood and stool culture. A wide spectrum antibiotic therapy was initiated. Repeated abdominal ultrasounds only showed reactive mesenteric lymph nodes, until the 8th day of hospitalization, when an abscess was found in the right iliac fossa, with a small hyperechogenic imagine consistent with appendicolith, suggesting the hypothesis of acute appendicitis. An exploratory laparoscopy confirmed the diagnosis of perforated appendicitis. Acute appendicitis in infants is uncommon, and has a high rate of complication and should be taken into consideration in patients presenting with unexplained, excessive crying. Ultrasound is recommended as a first-line modality of imaging. It should be repeated if initially negative, considering secondary inflammatory sonographic signs as potential indicators of acute appendicitis.

####

A CASE OF CHRONIC DIARRHEA: IS IT UNTREATABLE DIARRHEA? HOW DO WE MANAGE IT?
UN CASO DI DIARREA CRONICA: SI TRATTA DI DIARREA INTRATTABILE? COME LA TRATTIAMO?

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Introduction: autoimmune enteropathy (AE) is a rare diagnosis that should be considered in the presence of a protracted unexplained diarrhea, histologic changes in the bowel (villous atrophy and presence of apoptotic bodies), and after exclusion of a primary immunodeficiency. Treatment requires nutritional support and immunosuppressive therapy (IT).
Case report: a 7-month-old boy presented with protracted secretory diarrhea (8-12 ev/day of liquid stools), weight loss and poor general conditions. Initial investigations showed: hypogammaglobulinemia, hyponatremia, hypoalbuminemia, and enteroRNA virus positive in stools. The UGI and colon biopsies showed severe villous atrophy, lymphoplasmatic cell infiltrate and presence of apoptotic bodies in the duodenum, and mucosal inflammation with apoptotic bodies in the glandular epithelium in the colon. A primary immunodeficiency and other causes of early onset diarrhea were excluded. In keeping with clinical and histological findings, a diagnosis of AE was made. Total parenteral nutrition was required and IV 6-methylprednisolone therapy was started with tacrolimus introduced 1 month later. The second endoscopy revealed persistence of intestinal damage, requiring the introduction of azathioprine. At 18 months, the patient developed candida Parapsilosis infection, requiring the withdrawal of IT. A third endoscopy, performed with the patient off-treatment, showed normal duodenal morphology and mild colon inflammation. AZT was restarted and elemental feeds were slowly reintroduced with good tolerance.
Conclusion: AE is a challenging diagnosis in children requiring prolonged nutritional and immunosuppressive therapies.

####

A NEWBORN SHAKING: A MATTER…OF WHITE MATTER
UN NEONATO CHE TREMA: IN SOSTANZA…SOSTANZA BIANCA

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Human Parechovirus (HPeV) is a rare but significant cause of neonatal viral encephalitis, presenting with clinical deterioration, fever, seizures and characteristic MRI findings of white matter injuries. Diagnosis is confirmed by virus isolation on CSF. Severe clinical manifestations (sepsis, HLH-like) with hyperferritinemia and cytopenia have been reported.
A 15-day-old newborn was admitted to Pediatric Emergency for fever, poor feeding, drowsiness and weak crying. No signs of focal infection at physical examination or in the laboratory studies, a normal cranial ultrasound, and apneas onset. Antibiotic and antiviral prophylaxis was started. Onset of asymmetric right tonic-clonic seizures with unconsciousness 24 hours later, spontaneously resolving after 40 seconds. After HLH development, steroid therapy was started. Sleep EEG identified a bilateral anomaly with asymmetric spikes on left hemispheric derivations, and DWI-MRI revealed multifocal white matter lesions with capsule and callosum involvement, suggesting viral encephalitis vs MAS brain involvement. HPeV RNA on CSF was confirmed. Neurological follow-up during rehabilitation assessed mild development delay, progressive EEG and right side deficit normalization. MRI at 24 months showed atrophy-gliosis resulting from multifocal white matter injury.
As extensive involvement may lead to neurodevelopmental sequelae, HPeV should be considered as part of a suspected neonatal sepsis or atypical seizures workup. Characteristic neuroimaging findings may aid in early diagnosis.

####

DOES SCURVY STILL EXIST IN WESTERN COUNTRIES IN 2017?
ESISTE ANCORA LO SCORBUTO NEI PAESI OCCIDENTALI NEL 2017?

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Scurvy is a rare disease, caused by vitamin C deficiency and characterized by hemorrhages, hyperkeratosis, hypochondriasis and hematological abnormalities.
A 10-year-old child presented with global developmental delay, vomit and dehydration. During the anamnesis, long-standing history of food selectivity without fruits and vegetables and poor growth (weight and height <3°%ile) with normal somatomedine C, anti-tTG, and thyroid function. Vitamin D deficiency was corrected with oral supplementation.
Physical examination: haemorrhagic gingivitis and petechiae, dyschromia, follicular hyperkeratosis, and hypotrophic muscles. Laboratory examinations showed mild thrombocytopenia and leukopenia. Liver, kidney, electrolytes, coagulation, malabsorption tests, inflammatory and infectious indices were normal. Skeletal x-rays showed irregularity and thickening of the provisional zones of calcification of lower limbs. To rule out nutritional deficiency we dosed plasma vitamin C levels and found severe deficit (<2 umol/L) with very low PTH and ALP. After fifteen days of treatment with oral vitamin C, the patient showed normalization of vitamin C plasma level, white blood cells and platelets counts and bone metabolism tests with resolution of hemorrhages and skin manifestations. After 3 months, the patient started to grow again.
Conclusions: although the incidence of scurvy has become low in modern societies, it can still occur and early recognition is important because of the excellent outcome.

####

THE RIGHT WAY FOR A PERSISTENT NEONATAL JAUNDICE

LA GIUSTA VIA PER UN ITTERO NEONATALE PERSISTENTE

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An AGA breastfed term infant was referred to our Neonatal Unit for jaundice and phototherapy in the 5th day of life (Direct Coombs Test: negative). Physical examination revealed a cleft lip and right cryptorchidism. Echocardiography confirmed the prenatal finding of a tricuspid valve incompetence. Due to the persistence of jaundice and the appearance of dark urine, on day 7, complete blood examination was performed and liver function tests showed: aspartate and alanine transaminase: 93 U/L and 130 U/L respectively (normal range below 40U/L), total serum bilirubin: 22,4 mg/dl (direct bilirubin: 3,13 mg/dl), and serum alkaline phosphatase: 927 IU/L (max 550U/L). The coagulation profile revealed a prothrombin of 14% (normal range 70-130), activated partial thromboplastin time of 128s (normal range 27-40s), and an international normalized ratio of 4,56. Plasma and exchange transfusions were performed and supplementation of fat-soluble vitamins was started. Abdomen ultrasound was normal. Infections, congenital hypopituitarism, hemophagocytic lymphohistiocytosis and neonatal hemochromatosis were ruled out. Metabolic screening for inborn errors of metabolism showed an increase in galactose urinary excretion and the diagnosis of galactosemia was highly suspected. A lactose-free diet was started and liver function completely recovered. The diagnosis of galactosemia was confirmed by genetic analysis that revealed the presence of a omozygous p.Lys285Asn mutation (exon 9: c.855G>T) in the GALT gene.

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VOMITING AND LETHARGY IN NEWBORN: AN UNEXPECTED CASE.

VOMITO E LETARGIA IN UN NEONATO: UN CASO PARTICOLARE.

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Pseudohypoaldosteronism (PHA) type 1 is a rare salt wasting syndrome caused by aldosterone resistance that may occur in the first weeks of life with vomiting, lethargy, dehydration and failure to thrive.
There are two forms, with distinct inheritance:
- Renal PHA type 1 (autosomal dominant) caused by a mutation of the mineral corticoid receptor confined to the kidney, with mild hyperkalemia that tend to remit with growth.
- Systemic PHA type 1 (autosomal recessive) caused by a mutation of the epithelial Na+ channel with multisystemic salt wasting, acidosis and severe hyperkalemia that can lead to cardiac arrest.
Case report: A.P. was admitted to our ward on the 10th day of life for vomiting and inappetence. She was born at 39 weeks of gestational age by non-consanguineous parents, without complications. Family history was negative. Physical examination showed a hyporeactive, hypotonic, irritable child with poor skin perfusion, capillary refill was less than 3 seconds. Regular respiration. Blood pressure was 109/91 mmHg. Weight 2.950 kg (-14% from birth weight). Blood exams showed K+ 10.6 mEq/L, Na+ 126 mEq/L, with metabolic acidosis. In differential diagnosis the following were excluded:
- Hypoaldosteronism (the aldosterone levels were higher than the normal: 1500 pg/mL, v.n. 300)
- Congenital adrenal hyperplasia (the child had normal genitalia without clitoromegaly, serum cortisol was normal: 3.6 ug/mL, v.n. 0.1-10)

- Bartter Syndrome (plasma renin was high: 490 pg/ml, v.n. 105-161)
- Secondary transient Pseudohypoaldosteronism (renal ultrasound was normal and urinary tract infection was excluded).

After six months, the genetic analysis confirmed our clinical hypothesis of systemic Pseudohypoaldosteronism type 1.

PHA type 1 is a rare syndrome that occurs in newborns with common symptoms, such as vomiting and inappetence, and it is life-threatening if not recognized and treated in a timely manner.

Genetic analysis is essential for the correct diagnosis but targeted research of the mutations needs time. Clinical suspicion and exclusion of other diseases sharing similar features are essential for an early clinical diagnosis and a prompt management.

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