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WHAT CAN BE BEHIND AN EPISODE OF BRUE/ALTE?
COSA PUÒ NASCONDERE UNA BRUE/ALTE?

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This is the case of a two-month-old infant, who was evaluated for an episode of generalized hypotonia and paleness, and noisy breathing for an unspecified amount of time with a parental resuscitation attempt. Upon admission, the infant was afebrile with regular vital parameters but persistent hypotonia. No significant personal, family and social history. The patient was treated like an ALTE (Apparent Life Threatening Event). Blood tests performed showed metabolic acidosis with hyperlactatemia and mild dehydration. Intravenous administration of 0.9% saline solution was started. During hospitalization, the infant's temperature rose to 38 °C, so blood and urine culture tests were ordered and ceftriaxone introduced. Neurological (fundus oculi, lumbar puncture, RMI brain scan) and cardiological (ECG, echocardiography) investigations conducted were regular. A fast defervescence and resolution of neurological symptoms with a simultaneous normalisation of blood chemistry tests was observed. Toxicological screening revealed cocaine in the hair, secondary to intrauterine exposure, and ethyl alcohol 0,3 g/L in urine. Additionally, urinocolture was positive for Klebsiella Pneumoniae, which can cause false identification of alcohol consumption. In conclusion, we observed an episode of ALTE due to a urinary tract infection and exposure to a toxin. Behind an episode of BRUE (Brief Resolved Unexplained Events)/ALTE can be an abuse experience. We suggest that a toxicological screening should be included in early management of a BRUE/ALTE.

####

RECURRENT HEMORRHAGIC TONSILLITIS IN AN INFANT WITH VON WILLEBRAND DISEASE: A CASE REPORT
TONSILLITE EMORRAGICA RICORRENTE IN UN LATTANTE CON MALATTIA DI VON WILLEBRAND: UN CASO CLINICO

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A 10-month-old boy was admitted to our hospital after an episode of hemoptysis during a febrile hypertrophic tonsillitis. In neonatal age, he underwent lingual frenotomy without major bleeding. Parents reported tendency to bruise after minor trauma. Additionally, his mother experienced post-partum hemorrhage. INR elongation and factor VII slight deficiency (48%) were detected. It was hypothesized that a temporary deficiency of vitamin K, secondary to infection, had caused the acute bleeding. Upon discharge, parents were advised to administer vitamin K in case of fever. Three months later, during chickenpox infection, he presented hemoptysis and bleeding gums despite vitamin K supplementation. Second level coagulation tests identified type 2 Von Willebrand Disease (VWD). During both admissions, erythrocytes, platelets and fresh-frozen plasma were transfused. After clinical stabilization, adeno-tonsillectomy was successfully performed. At high risk for surgical bleeding, the factor VIII and Von Willebrand factor complex were infused perioperatively. VWD is the most common inherited bleeding disorder. It is caused by a defect (qualitative or quantitative) of Von Willebrand Factor (VWF), a glycoprotein crucial for platelet adhesion to the subendothelium. Type 2 VWD patients show qualitative defects of VWF. It is crucial to consider Von Willebrand Disease in the presence of recurrent mucocutaneous bleeding which is unresponsive

to vitamin K administration, despite normal or lightly abnormal clotting tests.

####

SEPTIC HIP ARTHRITIS IN THE NEONATAL AGE: A CHALLENGING DIAGNOSIS
ARTRITE SETTICA DELL'ANCA IN EPOCA NEONATALE: UNA SFIDA DIAGNOSTICA

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Septic arthritis of the hip is rare in infants, following bacterial invasion of the bloodstream, adjacent infected sites or direct inoculation. A 6-day-old male infant, full-term, second-born from vaginal delivery, arrived in our Emergency Room due to sudden irritability and low motility of lower limbs. His birth weight and Apgar score were normal, whereas maternal vaginal swab was not performed. Ortolani-Barlow test revealed right hip instability. Upon physical examination, the baby was alert and afebrile with normal vital parameters. Right hip was flexed and extrarotated, with painful passive and active mobilization. Blood count and CRP were unrevealing. No bone fractures were found on X-rays. Coxo-femoral ultrasound scan showed intra-articular effusion, and intravenous antibiotics (amikacin plus ceftazidime) were started. Right hip arthritis with capsule relaxation was confirmed by MRI, requiring an echo-guided drainage of the affected joint. Synovial fluid colture revealed Staphylococcus epidermidis. Improvement of hip movements was noted in 7 days, verified by ultrasound which proved resolution of joint effusion. After discharge, antibiotic treatment was stopped on day 10 with no further disturbances. Early diagnosis (based on Morrey's criteria) and prompt treatment (with antimicrobials for <2 weeks and 1 joint aspiration) are crucial for optimizing outcome and avoiding long -term disabilities of septic hip arthritis in infants.

####

REBECCA: AN ARTHRITIS - MULTIPLE POSSIBLE DIAGNOSIS
REBECCA: UN'ARTRITE, MOLTEPLICI POSSIBILI DIAGNOSI

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Rebecca, 8 years old, has a history of recurrent oral ulcers. Family history of arthritis. Fever and migrant arthritis started with hip and cervical pain, then big articulations, followed by small articulations. No rash. History of non-treated tonsillitis, positive pharyngeal swab, elevated ASLOT. Normal blood count, increased CRP, ESR, C3. Negative serology for virus and borrelia. Echography of the hip: modest liquid layer. Therapy with NSAID and amoxicillin-clavulanate, no improvement. To cover infectious arthritis, ceftriaxone and clindamycin were prescrived with no improvement. She had a normal RF titer and elevated fecal calprotectin. Ultrasound of the abdomen: adenitis and small pelvic liquid layer (no celiac disease). ECG and echocardiography: normal. Ophthalmologic evaluation: normal. At our hospital, she presented anemia and leukocytosis, very increased ESR, CRP, C3, ASLOT, Ab anti Dnase, negative auto-antibodies, normal peripheral smear, and oscillating fecal calprotectin. ECG, CXR, echocardiography, and abdominal ultrasound: normal. Therapy with prednisone, with improvement, but persisting pain. After high ASLOT and anti DNase, prednisone was suspended and she started high doses of aspirin with no resolution of symptoms. Shifted to prednisone and naproxen, improvement, still pain at two fingers with enthesitis. Prophylaxis with diaminocillin. Rheumatologic evaluation: morning stiffness of finger and wrist. Oral ulcers. Increased calprotectin. EGDS, colonoscopy, and video capsule: small lesions of stomach and small intestine. When the presentation is weird, think bowel disease.

####

"THE BABY WITH SILVER HAIR"

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We will present a case of hemophagocytic lymphohistiocytosis (HLH) associated with Griscelli Syndrome type 2 (GS2) in a 3-month-old male who had been hospitalized for recurrent infections several times. During the last hospitalization he presented with high fever, scruff of the neck lymphadenopathy, hepatosplenomegaly and petechiae in both lower arms. Laboratory data revealed acute pancytopenia with hyperferritinemia, high values of LDH, hypertransaminasemia, hypofribinogemia, hypoproteinemia and coagulopathy. The bone marrow aspirate revealed some morphologically benign macrophages with haemophagocytic activity that confirmed the diagnosis of hemophagocytic lymphohistiocytosis. The baby was treated according to the EURO-HIT-HLH criteria with remission of the symptoms. The molecular analysis didn't show any mutations in the exons of the genes correlated with familial hemophagocytic lymphohistiocytosis. At 11 months of age, the baby had a relapse, so he was treated according to the HLH-2004 criteria followed by hematopoietic stem cell transplantation from an HLA-matched related donor. The diagnosis of GS2 was made at 2 years of age in response to a set of clinical features, such as: hypopigmentation of the skin and silver hair not associated to albinism, parents' consanguinity and a pathognomonic appearance by microscopic examination of a hair. The subsequent molecular analysis showed a homozygous mutation, inherited from both parents, of a splicing trait in the RAB27A-gene.

####

ERYTHEMA NODOSUM: WHAT DOES IT HIDE?
ERITEMA NODOSO: COSA NASCONDE?

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A 9-year-old girl from the Philippines was admitted to the ED because of a rash on both legs associated with low grade fever and mouth sores for one week. Medical history was normal. She hadn't taken any drugs. Her vaccination schedule was up-to-date. Upon physical examination, vital signs were normal, oral ulcers were noticed and a raised erythematous and painful rash was found on the extensory surface of the legs. Lab tests showed increased inflammatory values. Because of the oral ulcers, an eryhema nodosum associated to primary herpetic infection was suspected. Thus, treatment with acyclovir and bethametasone was started, improving the oral signs, while low-grade fever and skin lesions were still present. Lab tests and instrumental procedures were performed: throat swab culture, antistreptolysin O titer, fecal calprotectin, and abdomen ultrasound. All resulted negative. Further evaluation revealed a positive Mantoux reaction. Quantiferon assay and gastric aspirate samples for BK were negative. The chest X-ray was normal. Diagnosis of erythema nodosum associated with latent TB was made. Treatment with a three antitubercolar drug regimen was initiated with rapid fever response and progressive resolution of the nodules.

####

WEBER CHRISTIAN PANNICULITIS: COULD COLCHICINE BE EFFECTIVE?
PANNICULITE DI WEBER CHRISTIAN: LA TERAPIA CON COLCHICINA PUÒ ESSERE EFFICACE?

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A 13-year-old boy was admitted to our centre presenting reddish nodular swellings, located on the upper and lower limbs and on the buttocks. Skin lesions were painless, non-itchy and not associated to systemic symptoms such as fever, abdominal pain or weight loss. Laboratory evaluation showed normal red and white blood cell count, platelets, coagulation profile, CRP, ferritine, liver and renal functions, immunoglobulines, C3 and C4, IgE. Celiac disease and thyroid disfunction were also excluded. No evidence of ANA, ENA, nDNA and ANCA was found. Skin biopsy of the lesions revealed lobular panniculitis with chronic inflammatory infiltration of T and B lymphocytes (CD3+, CD4+, CD8+), foamy histiocytes and giant cells, making diagnosis of Weber-Christian panniculitis. Therapy with colchicine (1 mg/day) was soon started with good response, and after a 6-month follow-up, the boy presented only few nodular lesions over his finger tips. Weber Christian disease (WCD) is an infiltrative inflammation of fat tissue rarely reported in childhood and adolescence. WCD may only have cutaneous involvement or it can present as a severe systemic illness affecting heart, kidneys, liver and lungs. No specific treatment is available, even if steroids and immunosuppressive drugs are often used. As observed in our patient, a 1st step therapy with colchicine could be effective, based on the evidence of its wide antinflammatory use in several cutaneous diseases.

####

HEMORRHAGIC SKIN LESIONS: IS IT ALWAYS VASCULITIS?
LESIONI EMORRAGICHE CUTANEE: SI TRATTA SEMPRE DI VASCULITE?

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An eight-year-old child, who was sent to us from another hospital, presented at our clinic under suspicion of Henoch Schonlein Purpura, with a case history of fever, abdominal pain, vomiting and skin haemorrhagic lesions. Physical examination revealed several erythematous itchy elements and papules with central ulcers and necrotic crust distributed over the body's surface. The blood chemistry tests performed revealed no alterations, with the exception of slightly increased inflammatory indices. Case history, especially the combination of fever associated with abdominal pain and skin lesions, made it possible to exclude the diagnosis of Henoch Schonlein Purpura. Dermatological advice posed the suspicion of Pityriasis Lichenoides et varioliformis acuta (PLEVA) also known as Mucha Habermann disease, a papular dermatosis of uncertain etiology. Skin biopsy confirmed the diagnosis. In particular, the form presented in this patient was Febrile Ulceronecrotic Mucha Habermann disease, a rare variant of PLEVA, which is more destructive and associated with systemic involvement such as high fever and development of skin necroses, which may lead to death. Treatment with steroids was started with benefit, but with recrudescence of the same symptoms of onset when the drug was stopped. Due to steroid dependence, immunosuppressive therapy with methotrexate was initiated with significant improvement in quality of life and the possibility of steroid reduction.

####

THE CHILD WHO ALWAYS LOOKED UPWARD
IL BAMBINO CHE GUARDAVA SEMPRE VERSO L'ALTO

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This case is of achondroplasia, the most common non-lethal skeletal dysplasia, in a child who was born at term of gestation from a physiological pregnancy. He's now 1 year of age. Skeletal dysplasia was suspected at birth by the presence of clinical features typical of achondroplasia – macrocephaly with frontal bossing, flat nasal bridge, short limbs with rhizomelic shortening of upper limbs, trident hand.

Thus, follow-up diagnosis was carried out. A total body radiography was performed, with evidence of shorter than normal limbs. An orthopedic consultation confirmed the suspiscion. Transfontanellar cerebral ecography showed ectasia of liquoral spaces and interhemispheric fissure. This exam was performed every six months. The screening test for congenital hearing impairment was negative. Soon, he will undergo a polysomnography. During the first months of his life, length remained below the 3rd percentile, whereas head circumference was over the 95th percentile. Motor milestones were adequate for age. When he was 5 months of age, genetic testing looking for FGFR3 mutations was carried out. Waiting for the result, he was monitored every 3 months to assess his growth over time and to look for the appearance of complications. Finally, the genetic testing confirmed the suspicion – the analysis of the FGFR3 gene showed the substitution p.Gly380Arg in eterozigosis. This is the most common autosomical dominant mutation causing achondroplasia.

####

A LONG PROPHYLAXIS…
UNA LUNGA PROFILASSI…

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Rickets is a metabolic disease of the growing bone characterized by a mineralization defect. The distinction between nutritional rickets (lack of vitamin D/ calcium/phosphorus) or genetic rickets (mutations of genes involved in vitamin D metabolism or in calcium/phosphate homeostasis) is crucial for therapy. We present the case of a 3-year-old boy affected by short stature and bone deformities. In his family history, two maternal cousins had lower limb deformities. His perinatal anamnesis was normal along with his auxological parameters at birth and neurodevelopment. Starting at 4-months, his growth chart deflected: before he began crawling, his limbs had begun to bend with no improvement with cholecalciferol supplementation. Could it be vitamin D resistant rickets? Physical examination showed: frontal bossing, expanded costochondral junctions, lumbar hyperlordosis, valgism of the elbows, varism of the knees and enlarged ankles. His weight and height were 5th percentile and cranial circumference 75th percentile. Laboratory examinations showed normal levels of serum and urinary calcium, and normal levels of 25(OH)D but increased alkaline phosphatase and PTH levels, hypophosphatemia and high phosphaturia. A "metaphyseal flaring" was radiologically documented. The genetical analysis confirmed the diagnosis of X-linked hypophosphatemia, one of the most common forms of genetic rickets. This case underlines the importance of the anamnesis in order to avoid unnecessary therapies and make a quick diagnosis.

####

ASLO TITER: AN OFTEN CONFOUNDING FACTOR IN ARTHRITIS DEFINITION

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A previously healthy 6-year-old girl was admitted for a 5 week distrubance of her left knee and ankle pain and antalgic attitude in flexion without fever, causing severe functional impotence especially in the morning. Elevated in-crescendo ASLO titer was associated (2520 IU/ml) and two recent febrile tonsillitis requiring amoxicillin occurred respectively 7 weeks and 3 weeks before hospitalization. Lab tests revealed normal WBC but elevated RCP and ESR, while ANA were negative. Cardiac investigations were normal. Femoral X-rays showed condylar cortical irregularities and an areola of bone rarefaction with sclerotic labrum at the distal-third diaphysis. Joint effusion, synovial ankle thickening and hypervasculariza-

tion appeared in the ultrasound. Many hypothesis were considered. Jones criteria for ARF were not fulfilled and reactive forms were excluded after negative microbiological exams. Osteoid osteoma and subacute osteomyelitis with joint involvement were unlikely because of clinical course. PSRA was strongly suspected, but arthritis quickly evolved to involving the right knee and a toe of the left foot, suggesting a diagnosis of oligoarticular JIA. Ibuprofen was started and a specific therapeutic plan was defined. High ASLO titer is often a confounding factor, capable of delaying exact recognition of joint diseases. JIA requires a clinical-based diagnosis and should be suspected in patients under-16 with a history of arthritis for over 6 weeks when other diagnoses have been excluded.

####

FROM PRENATAL DIAGNOSIS OF PYELECTASIS TO MEKI EXPERIMENTAL TREATMENT (SELUMETINIB) FOR NF1

DALLA DIAGNOSI PRENATALE DI PIELECTASIA AL TRATTAMENTO SPERIMENTALE DELLA NF1 CON MEKI (SELUMETINIB)

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S. (male, 3y 6m) was prenatally diagnosed with bilateral hydroureteronephrosis. The abdominal ultrasound at 1 month of age confirmed severe bilateral hydroureteronephrosis and showed a solid formation between the rectum and bladder. The histologic exam identified a neurofibroma, in a child with two cafè au lait spots: the direct sequencing of the NF1 gene revealed a pathogenetic mutation (c.5902C>T (p.Arg1968*)). The mictional cystography was normal (no reflux, no bladder abnormalities). The MAG3 scintigram showed an obstructive pattern with dilated, tortuous ureters. The bladder was only slightly visible. After a first infection, antibiotic prophylaxis was indicated. S. underwent several interventions, the first, to resolve obstructive uropathy bilateral urethral reimplantation, failed. The second intervention was performed to avoid a compromised kidney (an initial loss of renal function was then evident). S. was provided first with a right ureterostomy, and then with a left nephrostomy. The check-up MRI showed important growth of neurofibroma with infiltration of the lower abdominal structures. Given the non-operability and the documented compression on adjacent structures, an experimental cytoreductive treatment for NF1 was proposed: selumetinib (MEKi), an oral selective inhibitor of MAPK kinase. Treatment started in June 2018. S. is now waiting for the check-up MRI to assess the response, and then define the following steps for his treatment.

####

BACK PAIN IN CHILDREN: WHAT IS THE DIAGNOSIS?

MAL DI SCHIENA NEI BAMBINI: QUALE DIAGNOSI?

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XX, 11y, was admitted to our hospital with subacute lower back pain after a minor trauma eighteen months prior. She

also had severe constipation over the last two weeks. Clinically, she complained of tenderness after palpation at the thoracic -lumbar spine. No other symptoms were reported (no fever, lymphadenopathy, skin rashes or abdominal mass). Spinal radiographs (XR) showed D11 vertebral body wedging. The blood exam only showed mild increase in inflammatory markers. Mantoux test was negative. Abdomen ultrasound was normal. The spinal MRI scan revealed multiple lumbar and dorsal bone thinning areas, osteosclerosis clusters and signs of bone marrow oedema (abnormal hyperintensity on STIR images). Bone marrow aspirate and biopsy were normal. Cultures from blood and bone biopsies were also negative. Total body scintigraphy showed increased uptake from known vertebral lesions and a new lesion on the right femur. The MRI scan of this site showed multi-focal areas of oedema-like bone marrow signs (as vertebral images), and an osteolytic lesion surrounded by sclerosis on XR. Bone biopsy of both lesions showed chronic, non specific, aseptic inflammation. After ruling out acute or chronic infections, malignancies and benign/malign bone tumors and metabolic disorders, the final diagnosis was of chronic recurrent multifocal osteomyelitis (CRMO). She began therapy with NSAIDs and bisphosphonates.

####

HYPOALBUMINEMIA AND ACUTE CYTOMEGALOVIRUS INFECTION: IS THERE A RELATIONSHIP?

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Menetriere's disease associated with Cytomegalovirus (CMV) infection is a rare entity in children, characterized by hypertrophic gastropathy and hypoalbuminemia, secondary to protein loss through the gastric mucosa. A previously healthy 1-year old girl presented with fever which had begun 4 days prior, anorexia, fatigue. Two episodes of vomiting and diarrhea with a soft stool was referred by parents over the past ten days. Clinical examination revealed peripheral edema. A low level of serum albumin (1,7 g/dl), without other alterations, was detected by laboratory exams. After admission to our ward, two intravenous (IV) infusions of albumin solution were needed, and I.V. fluid therapy was administered for seven days. Urine samples were negative for proteinuria. Ultrasonography showed mild hepatomegaly without splenomegaly or ascites. Serological exams revealed a recent CMV infection (high level of anti-CMV IgG and IgM), confirmed by quantitative blood CMV-DNA PCR. No macroscopic findings were recognized by a gastrointestinal upper endoscopy. Histological exam showed mild gastric foveolar hyperplasia and mixed inflammatory cells within the lamina propria. Immunohistochemical studies detected CMV Early Nuclear Antigen in gastric biopsy. The child fully recovered with supportive care without complications. We reported a case of a pediatric patient with hypoalbuminemia secondary to self-limiting CMV associated with protein-loss gastropathy, in absence of endoscopic findings of Menetriere's disease.

####

AN "ATYPICAL" CASE OF FEBRILE CHOLANGITIS

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A 7.5 year-old Angolan boy with familiarity for Sickle Cell Anemia was admitted with 1 day of high fever, abdominal pain, anorexia and decreased general conditions. Examination demonstrated a mild scleral icterus, a generalized maculopapular erythematous rash, an erythematous pharynx and a slightly tender hepatomegaly. Blood tests revealed cholestasis, mild transaminitis, elevated ESR and CRP. Ultrasonography showed an enlarged liver with minor distension of left hepatic duct. Serological tests for viruses, bacteria and protozoans, blood culture and throat swab for Group A

Streptococcus were negative; hemoglobin S wasn't found. Antibiotic therapy (Piperacillin/tazobactam) started on day 1 was ineffective. From day 8, the patient became afebrile, but elevated ESR and CRP persisted and thrombocytosis and anemia developed on day 13. Clinical features and laboratory findings suggested an incomplete Kawasaki disease (KD) (fever ≥5 days and 2 principal clinical criteria), so the patient was treated with a single dose of intravenous immunoglobulin (2 g/kg) and oral aspirin (3 mg/kg/day) with gradual decrease of inflammation markers and platelet count. Echocardiography on day 18 and a month later were normal. Now, a regular heart surveillance is continued. KD is often a major clinical challenge and its diagnosis shouldn't be underestimated. The case study shows that KD has to be considered in children with prolonged fever, signs of hepatobiliary disease and high inflammation markers.

####

CLINICALLY AMYOPATHIC DERMATOMYOSITIS (CADM) TESTED POSITIVE FOR MELANOMA DIFFERENTIATION ASSOCIATED PROTEIN 5 (MDA5) AUTOANTIBODIES IN AN ITALIAN LITTLE GIRL

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Patients with CADM lack clinical or laboratory evidence of muscle disease for at least six months after initial presentation, but show typical skin findings. We describe a case of a 6-year-old child, whose family and past medical history were negative, who was admitted to our unit in March 2018 after a 3 month history of lower limb pain. This pain did not limit her from performing daily activities and did not usually require medical treatment. Physical examination showed heliotrope rash, erythematous scaling lesions on the elbows and knees, and Gottron's-like papules on the proximal interphalangeal joints of the hands, which were confirmed by cutaneous biopsy. All other systems were normal. Laboratory investigations showed increased serum levels of AST and ALT, positive result for anti-MDA5 and Ro-52 antibodies, and at the same time the full blood count, CK, kidney function, ESR, CRP, C3, C4 and infectious diseases profile were all normal. EMG, MRI, ECG, chest X-ray, spirometry, capillaroscopy, abdominal ultrasound were normal. Based on the clinical, laboratory, and histopathological findings, the diagnosis of CADM was made. The patient was discharged on oral prednisone 1 mg/kg daily for 15 days, and then MTX. She showed clinical improvement and started follow-up. Since patients with CADM testing positive for anti-MDA5 antibodies often develop rapidly progressive interstitial lung disease, malignancy and cutaneous vasculopathy, it is important to detect and monitor blood level of these antibodies.

####

TB: IN HINDSIGHT EVERYTHING IS CLEAR
TB: COL SENNO DI POI TUTTO È CHIARO

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Despite significant improvements in the diagnosis achieved over the years, this infection remains difficult to identify. A 6-year-old African girl presented to the emergency department after 14 consecutive days of fever with no related clinical signs. No respiratory involvement. She was born in Italy. No history of recent travels. No known familiar health problems. On the first 2 presentations, examination was unremarkable. Third presentation showed a rising CPR (10.83 mg/dl) and ESR (120 mm/h). X-ray revealed a right upper lobe pneumonia. Ampicillin was started with no response. Despite changes of therapy

(ceftriaxone and clindamycin) the child continued to be febrile. The tuberculin skin test (TST) was unclear. HIV sierology was negative. CT-scan showed consolidation and linfoadenopathy as prevalent findings. The patient was suspected to have Tuberculosis (TB). Quantiferon TB-Gold Test would have had a positive result 2 weeks later. The PCR (direct polymerase chain reaction) on 2 different gastric smears confirmed the suspicion. TB-treatment (isoniazid, rifampicin, pyrazinamide) was started with good clinical response. This infectious disease has long been neglected. After the notification of infectious disease to the Public Health Service, it was discovered that an uncle had the infection which he failed to report. A neighbour child was also tested later and resulted positive for TST.

####

SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS (SJIA): BE AWARE ABOUT WHAT CAN APPEAR SIMULTANEOUSLY

ARTRITE IDIOPATICA GIOVANILE SISTEMICA: ATTENZIONE A CIÒ CHE PUÒ COMPARIRE SIMULTANEAMENTE

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A 14-year-old, ill-appearing girl presented with fatigue, myalgia, diffuse arthralgia, polyarticular arthritis and a typical salmon-colored rash during daily febrile episodes. Lab tests showed elevated WBC count with PMN leukocytes predominance, elevated ESR and CRP. Ultrasound showed minimal pericardial fluid and slight enlargement of liver and spleen. Considering these findings consistent with probable sJIA and excluding other etiologies, therapy with ibuprofen and oral prednisone was initiated. Despite a partial clinical response, she developed a diffuse fixed itchy rash unrelated to the onset of fever or the general status. These features were not consistent with a typical sJIA rash, but with an adverse cutaneous drug reaction. NSAID therapy was suspended and the rash disappeared. During hospitalization ferritin increased up to 3497 ng/ml, AST was 90 UI/L, platelet count, WBC and Hb levels were decreasing. MAS was suspected. Bone marrow examination showed rare elements of hemophagocytosis and excluded Leishmania infection. An impending, unrecognized MAS is a potentially lethal complication in sJIA. Currently, there are different criteria to help diagnosing MAS before further clinical worsening. Waiting for clear MAS can expose the patient to a higher risk. Treatment with a 3-day course of I.V. methylprednisolone was started with clinical improvement and partial lab test normalization. The patient was discharged on oral steroids and she is now followed as an outpatient.

####

A PAIN THAT CORNERS YOU

UN DOLORE CHE TI METTE ALL'ANGOLO

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The aorto-mesenteric syndrome is an uncommon finding but, when identified, it can represent a pediatric emergency. A 13-year-old girl presented to the pediatric emergency department with an history of increasing abdominal pain, fever and vomiting. The laboratory exams and abdominal echography were negative. The symptoms did not improve with antiemetic or with analgesic therapy. During the clinical observation two biliary vomits occurred, suggesting an obstruction of the upper gastrointestinal tract. Given the age, sex and symptoms, an angio-TC was performed which demonstrated a critical aorto-mesenteric angle (28°) and distance (5 mm), with a mild proximal

duodenal dilatation. The symptoms significantly improved following nasogastric tube decompression and fasting regimen. After a few days the symptoms disappeared and the barium swallow x-ray demonstrated a regular transit. The surgical treatment was no longer necessary and the girl was discharged from the pediatric unit.

####

I CAN RESIST EVERYTHING EXCEPT… TENTACLES

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An 8-year-old boy was referred to the Pediatric Emergency Department after being stung by a sea anemone (Anemonia viridis) in Croatia. A burning, prickling, stinging pain of both lips and local oedema with difficulties in feeding and speaking immediately followed. The patient was at first evaluated in Pola, where he was promptly administered high doses of I.V. corticosteroids and antihistamines. After one day, he moved to our Department, with stable vital parameters. The physical examination revealed persisting facial oedema, especially of the lips, associated with some irregular purplish erythematous chin linear plaques as marks of tentacles. The laboratory exams showed just a mild elevation of the neutrophil count. In the following days, the patient developed left cheek pain and homolateral mucosal erythema, so an appropriate antibiotic therapy was started. The salivary gland ultrasound displayed an irregular patchy thickening of the subcutaneous tissues of the left cheek without evidence of abscess. In accordance with the specialist of the Antivenom Medical Center of Pavia, during the following days we kept monitoring vital signs and symptoms, administering I.V. corticosteroids, antihistamines and analgesics. After a complete recovery of symptoms and signs, the child was discharged.

####

A NEUTROPENIA WITHOUT SOLUTION
UNA NEUTROPENIA IRRISOLTA

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Joseph, a 15-year-old boy from Cameroon, was admitted to our ED. He was in poor general condition, severely malnourished, coughing. He was recently treated for malaria and bronchitis in Cameroon, with incomplete recovery. Investigations showed bilateral pneumonia, oral candidiasis, anemia, extremely low level of CD4 (Hb 8,7 g/dl, CD4 3/mm^3) and HIV-RNA detectable. He was diagnosed with AIDS (stadium C3) in probable vertical HIV infection. Antibiotics, antitubercolar prophylaxis with Isoniazid for TB contact and antiretroviral therapy (lopinavir/ritonavir + emtricitabina/tenofovir plus raltegravir) were immediately started with clinical, biochemical and virological improvement (HIV-RNA < 37 cp/ml). Simultaneously, Joseph developed a progressive, severe persistent neutropenia (N 190/mm3), slightly responding to granulokines. All investigations resulted negative and excluded a superinfection or immunological origin of neutropenia. Myelosuppression and neutropenia in HIV-positive patients is frequent and multifactorial (HIV cytotoxicity, secondary infection, malignancy and drugs). Considering the myelosuppressive effect of Ritonavir and Isoniazid, after a careful evaluation of HIV-infection status and active Tuberculosis infection risk, we decided to interrupt these two agents. After one month Joseph's neutrophil count doubled (510/mm^3 – 9,5%), without effects on the viral load. Unfortunately, Joseph's last neutrophil count was 310/mm^3.

####

WHAT LIES BEHIND AN ARTHRITIS

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A previously healthy 10-year-old child presented with a two month history of bilateral knee pain and morning stiffness, and had been referred to the rheumatology department. He had no known trauma. The pain got worse in the morning and had slight improvement with Ketoprofen. Laboratory investigation showed elevated ESR (43 mm/h) and CRP levels (48 mg/L), microcytic anemia and elevated Anti-streptolysin O titer (393 units/mL). Echocardiogram was negative and, with the suspicion of post-streptococcical inflammatory syndrome, he was started on Diaminocillin therapy. Two months later, the patient presented recurrent oral aphthosis. Extensive investigation including serological tests for autoimmune diseases, pathergy test and eye examination with slit lamp were all-negative; inflammatory indices remained elevated and microcytic anemia was persistent despite iron supplementation. After 10 months from the onset of knee pain, the child was referred to a gastroenterologist for evaluation of intermittent abdominal pain and fatigue. There was no history of fever, weight loss, rectal bleeding or diarrhea. An abdomen ultrasound showed ileum thickening. Colonoscopy and EGD revealed multiple ulcers and biopsies confirmed CD diagnosis. Joint involvement is the most common extra intestinal manifestation (30%) in children with CD, and it can occur before suggestive symptoms of bowel disease. Therefore, CD should be considered in differential diagnosis of arthralgia/ arthritis to avoid diagnostic delay.

####

COSA SI NASCONDE DIETRO UNA FEBBRE CHE NON PASSA?

WHAT HIDES BEHIND A FEVER THAT DOESN'T PASS?

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F.A., 6 y.o, was admitted to our hospital with a week long history of fever, abdominal pain, diarrhea and vomiting. She had returned the day before the onset of fever from a stay in Morocco. Her abdomen was treatable but widely painful to palpation on all quadrants. WBC count was 23.84 migl./mmc with 74% neutrophils and CRP 22 mg/dl. Abdominal ultrasound only showed presence of some larger lymph nodes along the right iliac vessels (dimensions up to about 8 x 4 mm). Cecal appendix was not displayed; kidneys were of regular size and echostructure, without signs of hydro -ureteronephrosis.
During the hospitalization, treatment with Ceftriaxone was started, and then substituted with Gentamicin due to an adverse reaction. The serological tests for main viruses, bacteria and parasites were negative, as well as Quantiferon and Mantoux (performed under supsicion of abdominal tuberculosis) and blood cultures. Urine culture was found positive for E.Coli (1 million CFU multiresistent). In relation to the persistence of the symptomatology, the girl was subjected to an abdomen CT, which showed both kidneys discretely increased in volume, with loss of normal cortico-medullary differentiation and inhomogeneous contrast enhancement due to bilateral pyelonephritis with initial abscess. Meropenem was started for 2 weeks followed by oral Ciprofloxacin; the fever passed 4 days after starting Meropenem. Two weeks later, the MRI showed initial reduction of the renal abscesses. We could

not perform voiding cystouretrography (VCUG) because she moved abroad.

####

CITOPENIA E ALPS: NON SEMPRE LA PRIMA DIAGNOSI È QUELLA GIUSTA! CYTOPENIA AND ALPS: THE FIRST DIAGNOSIS IS NOT ALWAYS THE RIGHT ONE!

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A 12yo boy diagnosed with ALPS at 22 months was referred to us due to disease progression. Clinical presentation included splenomegaly, progressive pancytopenia (mainly thrombocytopenia, gradually worsening) with "dysplastic" aspects to the bone marrow findings, mild impairment of liver function tests, pathological αβ-DNT cells level and hypergammaglobulinemia. Apoptosis test was repetitively positive. Concomitant infectious causes were excluded. Never started specific therapy. Radiological investigations showed hepatomegaly and enlarged abdominal lymph nodes with worsening splenic framework. Our clinical evaluation confirmed visceromegaly and pancytopenia with a definitive ALPS diagnostic profile (according to NIH 2010 criteria). The analysis of bone marrow showed the presence of numerous foamy histiocytes suggesting Gaucher Disease (GD). GD was confirmed by chitotriosidase and β-glucosidase dosage. MRI of the femurs showed signs of the disease. Enzyme replacement therapy was started and a normalization of the haematological indices was observed together with the ALPS parameters after 6 months. In addition to clinical similarities, patients with GD can present hyper-infiammatory characteristics secondary to the accumulation and activation of macrophages and a pattern of immune dysregulation that can overlap the features of ALPS. Therefore, the differential diagnosis of GD must be taken into consideration during the diagnostic work-up of patients with an "ALPS phenotype".

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A CASE OF SEVERE AUTOIMMUNE THROMBOCYTOPENIA - (UN CASO DI SEVERA PIASTRINOPENIA AUTOIMMUNE)

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A 7-year-old boy was referred to the emergency department for diffuse petechiae and ecchymosis and he was hospitalized for suspected autoimmune thrombocytopenia (PLTs 4000/mmc) with positive direct Coombs test. His past medical history was significant for axillary lymphadenopathy (follicular reactive hyperplasia). On examination, several enlarged, nontender lymph nodes involving supraclavicular and axilla areas were detected and CT scan of the abdomen revealed generalized adenopathy with numerous prominent lymph nodes in the retroperitoneal area. Flow cytometry of peripheral blood revealed an expanded population of alpha/beta double-negative T cells and hypergammaglobulinemia. All of these findings suggested the hypothesis of Autoimmune Lymphoproliferative Syndrome (ALPS). All genetic tests were negative. ALPS is a chronic, non-malignant lymphoproliferation disorder due to an inability to regulate lymphocyte homeostasis through the process of lymphocyte apoptosis. The main features are splenomegaly, lymphadenopathy, hypergammaglobulinemia and autoimmunity. Flow cytometry approach is helpful for the diagnosis and an elevated level of vitamin B12 is a known biomarker for the presence of FAS mutation. Patients may not have all clinical features or genetic

mutation and, nevertheless, be affected by an ALPS-like disorder. Individuals with severe autoimmune hemolytic thrombocytopenia may benefit from IVIG in combination with corticosteroids, but often immunosuppressive drugs (Sirolimus) are required.

####

PROLONGED URTICARIAL RASH AND DAILY FEVER: AN UNUSUAL CASE OF SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS
UN RASH PERSISTENTE CON FEBBRE QUOTIDIANA: UN INSOLITO CASO DI ARTRITE IDIOPATICA GIOVANILE SISTEMICA

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An 8-year-old girl was hospitalized for an urticarial rash in a febrile pharyngitis. Positivity for S. pneumoniae in pharyngeal swabs was reported in her history. Blood sample showed neutrophilic leukocytosis, increased inflammatory markers and IgM for M. pneumoniae. Oral antibiotics and antihistaminic therapy were started. She was feverish with severe itching until betamethasone was administered.
Daily evening fever and macular rash reappeared when the steroid was discontinued.
She started complaining about migratory arthralgia in the lower limbs and a slight ankle edema was noticed. Chest X-ray, echocardiography and abdomen ultrasound excluded infectious foci. Mantoux test was negative. Bone marrow aspirate was normal.
Striking elevation of CRP, ESR, ferritin, and SAA, combined with clinical history, suggested an auto-inflammatory disorder. She was started on Naproxen with regression of fever, improvement of erythema and reduction of inflammatory markers.
One month later, despite therapy, fever and pruritic rash recurred accompanied by arthritis of the knee. The diagnostic criteria for systemic juvenile idiopathic arthritis (SJIA) were met. Oral corticosteroid therapy ameliorated both clinical and laboratory parameters. The SIJA is a rare, auto-inflammatory disease. Systemic symptoms can initially be preponderant. We present a challenging case starting with daily fever and itchy hives rash. Arthritis of the knee appeared two months later. SIJA is a diagnosis of exclusion.

####

ACUTE COMPLICATED UTI AS A MANIFESTATION OF CHRONIC KIDNEY DISEASE: A CASE REPORT
IVU COMPLICATA COME MANIFESTAZIONE DI MALATTIA RENALE CRONICA: CASE REPORT

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Most UTIs in children are uncomplicated and respond readily to antibiotic treatment without further sequelae. We present the case of a 5-year-old boy admitted with dysuria, high-degree fever and vomiting. The child presented distal hypospadia. At examination, he was sleepy, tachycardic, and hypotensive. Blood tests showed PCR 124 mg/L, PCT 12.222 ug/L, thrombocytopenia, coagulopathy (INR 2,24), creatinine 230 umol/L. Abdominal US showed a hyperechoic kidney with a reduction of cortico-medullary differentiation. The patient was hospitalized in PICU and broad-spectrum antibiotic therapy was started. Blood and urinary cultures resulted positive for E. Coli sensitive to prescribed antibiotics. However, due to persistent febrile episodes and increased inflammatory markers, an abdominal CT was performed documenting the presence of bilateral renal abscesses, mainly to the right kidney. This was associated

with severe kidney dysfunction, requiring dialysis. Due to persistent toxic appearance, a right nephrectomy was performed. After this, the fever resolved and inflammatory markers normalized. Renal function also improved, allowing the patient to stop dialysis, but continued to suffer from mild chronic renal failure. Further diagnostic studies revealed the presence of primary vescico-ureteral reflux nephropathy. Severe and complicated UTIs are uncommon and can be the first manifestation of a congenital anomaly of the kidney and urinary tract.

####

CLINICAL DIAGNOSIS OF MPS: THINK TREATABLE DISEASES FIRS
DIAGNOSI CLINICA DI MPS: CONSIDERA PRIMA LE PATOLOGIE TRATTABILI

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D.D. is a 3-year-old boy who was evaluated in February 2018 for pectus carinatum and short stature. His medical history and neurological development were both normal. His great-great grandparents were blood relatives. Physical examination revealed dental abnormalities, lower rib-cage flaring, kyphoscoliosis. His height and weight were under the average for the age, despite normal values at birth. Ophthalmic evaluation showed corneal clouding. We performed urinary GAGs, at the "Microcitemico" metabolism laboratory, which were elevated. Qualitative chromatography was negative. Despite this, for the clinical suspect of MPS IV, we performed skin biopsy to detect enzymatic GALNS activity, analyzed by the Gaslini Lab of Molecular Genetics, which was absent. Molecular analysis of GALNS gene is pending. He will start ERT in September. Elosulfase alfa has to be admistered IV at the dose of 2 mg/kg/w with antihistamine premedication. Follow-up data suggest improvements in daily activities, endurance, respiratory function, muscle strength, joint movements, pain and fatigue. The patient developed antibodies to elosulfase alfa, although this is not correlated with safety or efficacy. Further studies will provide comprehensive data of the effects on the growth patterns, especially in patients younger than 5 years. Optimal benefits from ERT would require the beginning of the treatment as soon as possible to avoid irreversible damage. It is advisable to first test treatable MPS, based on clinical signs.

####

ECZEMA HERPETICUM AND ATOPIC DERMATITIS
ECZEMA HERPETICO E DERMATITE ATOPICA

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Patients with atopic dermatitis are at risk for developing severe skin complications as a consequence of disseminated viral infections, such as eczema herpeticum. Even if it is a rare event, it is a potentially life-threatening disorder. We describe a case of a twelve-year-old girl who has been affected by atopic dermatitis since her first year of life with good control of the disease. She developed bilateral conjunctivitis with right preauricular adenopathy, otalgia and pain in the temporal mandibular joint. At the same time she developed a sudden skin eruption of painful clusters of monomorphic vescicles with erythema, approximately 5 -10 mm in diameter. The rash started around the eyes and it produced serous and yellow fluids. After three days, fever appeared and the rash progressed all over the face and the wrists. It evolved into crusted, punched-out skin erosions that coalesced into plaques. With the beginning of systemic antiviral therapy

with acyclovir, the fever disappeared and the rash stopped. Because of the risk for keratoconjunctivitis, topical therapy with vangancycolvir was started. In order to prevent bacterial infection of the skin, antibiotic therapy, both with systemic beta lactam and topic fusidic acid was given. No other organs were affected, and she never developed neurological symptoms. Serologic assay indicated a primary HSV-2 infection explaining the Kaposi varicelliform eruption, synonimous with eczema herpeticum.

####

AN UNUSUAL SWELLING OF THE EYE AFTER GYM
UNO STRANO GONFIORE OCULARE DOPO LA GINNASTICA

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A 6-year-old girl was admitted to our hospital for the appearance of swelling of the left eye after intense physical activity, which had started 8 months before. The symptomatology progressively worsened, presenting even upon awakening in the last few days. The visit showed a slight swelling of the lower eyelid of the left eye. Suspecting vascular disease, a doppler ultrasound was requested. The US showed a suspected venous malformation, confirmed by contrast-enhanced MRI of the periorbital region. Surgical reduction was preceded by solidification by an interventional radiologic procedure. Venous malformations have the peculiar clinical feature of increasing volume concurrently with physical exertion, fever, heat exposure and other conditions that result in increased blood flow. There is also the possibility of volumetric increase in conjunction with hormonal changes (puberty, pregnancy) or after trauma. Diagnosis is based on clinical evaluation and on the execution of radiological investigations (doppler US/MRI). Surgical excision and/or sclerotherapy is indicated when the malformation is of a significant dimension or affects delicate areas, such as the head/neck, where it can have a negative functional and aesthetic impact on the patient's life. In conclusion, when a child presents with a swelling that appears or worsens after physical activity, one should think about the possible vascular origin of the lesion and, in particular, of venous malformations.

####

A RARE CASE OF HEPATOSPLENOMEGALY: INFECTIOUS DISEASE OR HAEMOPATHY AT THE ONSET?
UN RARO CASO DI EPATOSPLENOMEGALIA: INFEZIONE O ESORDIO DI EMOPATIA?

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Alex, a 1-year-old, is admitted for evaluation of a massive hepatosplenomegaly and poor weight gain. He had high fever episodes occurring at 2-week intervals for the past 2 months. Physical examination revealed a febrile child with mucocutaneuous pallor with diffuse petechiae and ecchymosis, cervical and inguinal adenopathy, firm spleen and liver palpable on the transumbilical plane. Laboratory evaluation showed Hb 7,2 g/dl, WBC 19.490/mmc with N 60,6%, L 26,8% M 12%, PLT 40.000, PCR 56,5 mg/l, procalcitonin 10,6 ng/ml. Markers for TORCH and EBV infections were negative. Chest radiography and abdominal US were non-diagnostic. The bone marrow had a hypercellular and mielodysplastic aspect, with some atypical cells, few megakaryocytes, several myeloid and lymphoid cells. Hydration and empiric I.V. antibiotic therapy was started immediately. Diagnostic suspicion lead to visceral leishmaniasis, also because Alex comes from an endemic area of sandflies. However, total anti-Leismaniasis immunoglobulins and specific parasites were not found in his

blood. Moreover, a high level of HbF (20,7%), cytopenia and accurate examination of bone marrow samples supported the hypothesis of an onco-hematological disease. Furthermore, numerous fleeting and brownish skin nodules appeared as a cutaneous manifestation of leukemia. The presence of the genetic mutations PTPN11 finally confirmed that Alex was affected by juvenile myelomonocytic leukemia (JMML).

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EMOTTISI MASSIVE RICORRENTI: POSSIBILE INDICAZIONE AL TRAPIANTO POLMONARE IN FC
MASSIVE RECURRENT HAEMOPTYSIS: A POSSIBLE INDICATION FOR LUNG TRANSPLANTATION IN CF

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A 16-year-old female affected by Cystic Fibrosis was diagnosed at 2 months (CFTR genetics: 218AA>G/I148T-3199del). She presented chronic colonization by mucoid P. aeruginosa and MSSA, and exocrine pancreatic insufficiency. At the age of 6-years-old, US evidenced signs of CF-related hepatopathy, which had evolved into progressive biliary cirrhosis, portal hypertension and hypersplenism, esophageal varices (F1-F2) treated with endoscopic ligation. The portal hypertension worsened, and when she was 13, transjugular intrahepatic portosystemic shunt (TIPS) was performed and treatment with lactulose and branched amino acids for persistent hyperammonaemia was initiated. Coagulation patterns showed chronic PT alteration resistant to I.V. vitamin K and thrombocytopenia. At 14-years-old, she presented with recurrent episodes of moderate haemoptysis, so she underwent a selective embolization of bronchial arteries; blood extravasation in mediastinum required intubation in the intensive care unit (ICU). Afterwards, respiratory function worsened (actual FEV1 52% predicted) with frequent pulmonary exacerbations and recurrent moderate-massive haemoptysis. At 16-years-old, a new embolization was attempted without complete resolution. Since then, she has experienced three other episodes of massive haemoptysis (>200 ml) requiring I.V. tranexamic acid and sub-intensive monitoring. She is now on a waiting list for hepatic-pulmonary transplantation. In CF, the bronchial arterial system can become hypertrophic, with collateral vases and neo-angiogenesis, before haemoptysis manifests clinically. Is it preventable? If so, how? What are the advantages in terms of survival and pulmonary function preservation?

####

KAWASAKI DISEASE WITH ACUTE RESPIRATORY DISTRESS SYNDROME AFTER INTRAVENOUS IMMUNOGLOBULIN AND ALBUMIN INFUSION: A CASE REPORT
SINDROME DI KAWASAKI COMPLICATA DA ARDS DOPO INFUSIONE DI IMMUNOGLOBULINE E DI ALBUMINA: CASO CLINICO)

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We report the case of a patient with Kawasaki Disease, who unexpectedly developed two episodes of acute respiratory distress syndrome (ARDS); the first after IVIG infusion, and the second after albumin infusion. Were they both TRALI (Transfusion-related acute lung injury)? The patient was a 5-year-old girl, who was diagnosed with typical Kawasaki disease and treated with high-dose intravenous immunoglobulin. She developed ARDS during the IVIG infusion, and there was a relapse of ARDS after albumin infusion. The authors assume that ARSD after IVIG infusion in a patient with KD probably resulted from a generalized capillary leak after a systemic inflammatory immune response. TRALI's incidence in the pediatrics population is unknown. Only a few cases of TRALI after IVIG have been described in children. We found no cases of TRALI after albumin infusion in the literature. Although

the cause of ARDS after albumin infusion remains unanswered in our case, several hypotheses emerged: 1) Immune mediated mechanism: albumin, being a blood component, could boost generalized capillary leak and developed ARDS, in the same way as IVIG infusion. 2) Albumin infusion could cause ARDS by circulatory volume overload.

####

A CASE OF HEMORRHAGIC SYNDROME IN A DEVELOPING COUNTRY
UN CASO DI SINDROME EMORRAGICA IN PAESE IN VIA DI SVILUPPO.

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An 11-year-old boy was admitted to the emergency department of the Regional Hospital of Ziguinchor (Senegal) for epistaxis and hematemesis. Two days before, the child reported a mild head trauma, without loss of consciousness, from an accidental fall backwards.
At admittance, clinical examination of the back and head showed no bruising, tenderness, or deformity. The patient showed no haemodynamic failure (normal blood pressure and good tissue perfusion) but the state of consciousness was altered (GCS 11/15) and he was febrile (39°C). Tender hepatomegaly was observed; no peritoneal signs were found. No signs of mucocutaneous bleeding were identified. The boy also had abundant melena.
Blood tests showed moderate anemia (Hb 7,4 g/dl, HCT 22%) and thrombocytopenia (16.000/mm3). WBC count was 4.100/mm3 with neutrophilia (81%). After ORL examination, post-traumatic bleeding was initially suspected. The patient received transfusional support and underwent CT scan of the head, neck and abdomen, but no vascular or organ lesions were found. Finally, a rapid diagnostic test for malaria (TDR) was performed, showing a positive result. Diagnosis of complicated malaria with thrombocytopenia, haemorrhagic manifestations and cerebral involvement was therefore made. Quinidine I.V. therapy was administered on the first day, followed by oral therapy with dihydroartemisinin-piperaquine for 3 days. He was discharged after 5 days and had an uneventful recovery without further bleeding episodes.

####

A CASE REPORT OF MPS II: WHEN TO STOP ERT?
UN CASO DI MPS II: QUANDO SOSPENDERE LA TERAPIA ENZIMATICA SOSTITUTIVA?

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Mucopolysaccharidosis type II (MPS II, also known as Hunter syndrome) is an X-linked multisystem disorder characterized by the accumulation of glycosaminoglycans (GAGs) in lysosomes. Most of the affected patients are male. Clinical manifestations include CNS involvement (mainly characterized by progressive cognitive deterioration), progressive airway and cardiac disease, short stature, macrocephaly, macroglossia, hoarse voice, conductive and sensorineural hearing loss, hepato-splenomegaly, dysostosis multiplex, spinal stenosis, and carpal tunnel syndrome in early ages. Urine GAGs can suggest the presence of an MPS condition, but they are not specific. To confirm the diagnosis of MPS II, one must detect the deficiency of iduronate 2 -sulfatase (I2S) enzyme activity in white cells, fibroblasts, or plasma, and one must check molecular analysis. ERT (enzymatic replacement therapy) has been available since 2006 with idursulfase (Elaprase®), a recombinant form of human iduronate 2-sulfatase. A widely discussed topic in the scientific community is when to stop ERT if no evidence of clinical efficacy is seen over the years of

treatment. It is known that early treatment with ERT can slow or even improve somatic manifestations of MPS II, but since Elaprase® does not cross the blood -brain barrier, no effect on CNS disease is anticipated and the progressive neurological impairment is unavoidable. Although hematopoietic stem cell transplantation (HSCT) (using umbilical cord blood or bone marrow) could provide sufficient enzyme activity to slow or stop the progression of the disease, no controlled clinical studies have been conducted in MPS II. We present a case of a boy who was diagnosed with MPS II in 2006 at the age of 12-years-old. His clinical features included: weight 30 kg (>50° centile), height 115 cm (<3° centile), autonomous walking and feeding, toilet-trained, simple sentences, unclear language, IQ 48, and hearing devices. His performance at the 6 Minute Walking Test (6MWT): 244 metres. Hepatosplenomegaly (liver +6 cm, spleen +4 cm), umbilical hernia, claw hands, pes cavus, joint stiffness, moderate mitro-aortic regurgitation, and normal ejection fraction (EF) (taking captopril 8 mg for 3 times a day). The brain MRI showed ventriculomegaly and increased extracerebral spaces (i.e. brain atrophy). Urinary GAGs testing was 140 µg/g creatinine. We decided to start Elaprase®. After 1 year of ERT, we observed an improvement in the height and performance in 6MWT. Urinary GAGs testing was 19 µg/g creatinine. No cognitive improvement was observed. Over the next few years, although the growth and the urinary GAGs continued to improve, there was a progressive cognitive deterioration which included: no more walking, no sphincteric control, swallowing difficulties, no language, incapacity to understand simple orders, IQ non-testable, onset of seizures, and increased brain atrophy on the brain MRI. Furthermore, over the years, he underwent many orthopaedic interventions, was hospitalized six times for bronchopneumonia, and displayed deterioation of cardiac function. After 7 years of treatment, in agreement with the parents, ERT was stopped. No severe worsening was observed after stopping it. Parents refused gastrostomy. The patient died during the course of a bronchopneumonia at the age of 20.

####

ALWAYS LOOK AT YOUR PATIENTS SKIN
QUANDO IL DETTAGLIO NON DEVE SFUGGIRE

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A. was brought to our hospital because of the onset of a disseminated itchy rash, happening 3 days prior. During the previous week, she had frequently touched her mouth and showed polydipsia and lack of appetite. She had presented a single feverish peak two days before. Clinical evalutaiton showed disseminated skin lesions of different appearance. Since it was summer, we first hypothesized the traumatic lesions were due to her daily outdoor activities or insect bites. A closer look at her skin showed that she had different kinds of lesions, including hyperaemic and crusty papules (itching?), hard hyperaemic plates with petechiae and knee bruises. In her mouth, we observed little hemorrhagic ulcers near the molars. Blood examinations showed sever thrombocytopenia (5000/mmc) with regular count of the other blood cellular lines and GPT 170U/L without coagulation time abnormalities. We suspected a post-infectious autoimmune purpura (PTI) and we quickly started IVIG therapy (1 g/Kg), administered successfully without complications. After therapy she didn't show new symptoms and, considering the complete response (PLT 48.000/mmc), we dismissed her. Seven days later she appeared in good general condition, and didn't show new hemorrhagic lesions or other symptoms. Her platelet count was completely

back to normal (225.000/mmc). A simple case, but remember to always look at your patients skin.

####

A CASE OF MYCOPLASMA PNEUMONIA INDUCED RASH AND MUCOSITIS IN ASSOCIATION TO HHV6 CO-INFECTION
UN CASO DI RASH E MUCOSITE CAUSATI DA COINFEZIONE TRA MICOPLASMA PNEUMONIAE E HHV6

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A six-year-old girl presented to us with a history of fever and cough. She had conjunctivitis associated with mouth and genital mucositis. She had crackles at lung auscultation with interstitial pneumonia on the chest x-ray. Laboratory data showed luekocitosis and high CRP. Microbiological investigations were negative, except for HHV 6 and mycoplasma pneumonia (positive on swabs and in blood). Later, she developed a diffuse erythematous-edematous target like rash, and the mucosal sloughing worsened. Vesicles appeared in the centers of target lesions hesitating in burst with necrosis. Stevens Johnson Syndrome (SJS) was suspected and she was treated with clarytromicym, acyclovir, topic drugs and supportive care. The mild mucocutaneous involvement and the spontaneous recovery lead to a definitive diagnosis of Mycoplasma pneumoniae-induced rash and mucositis (MIRM) with HHV6 co-infection. MIRM is a rare, self-limiting condition affecting skin and mucosa caused by Mucoplasma. It is defined as a distinctive disease from SJS and Erythema multiforme major (EMM), and it has a various presentations, consisting of mucositis and a peculiar targetoid vescicolous rash. Muco-cutaneous criteria have been proposed to differentiate MIRM from SJS and EMM, but they are not yet validated. MIRM skin lesions seem to worsen with co-infection with other agent. Further characterizations of mycoplasma induced muco-cutaneuos diseases are needed in order to recognize children requiring intensive care at the onset.

####

A (NOT SO) STRANGE CASE OF A BABY ON FIRE
IL CASO STRANO (MA NON TROPPO) DELLA BAMBINA IN FIAMME

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A previously healthy 16-month-old girl presented a daily, but intermittent, fever (maximum 39,5°C), followed by a non-pruritic macular rash. She was admitted to the Pediatrics Ward of a nearby Hospital to start antibiotic therapy with I.V. Ceftriaxone. The laboratory findings showed elevated white blood cell and platelet counts, and an important increase in ESR (54 mm/h) and CRP (22,19 mg/dL). Blood and urine cultures were negative, as well as chest X-Ray, abdominal and cardiac US. On suspicion of atypical Kawasaki disease, because of lymphoadenopathy, two cycles of IV Immunoglobulins (2g/kg) were administered together with ASA at anti-inflammatory doses. After an initial improvement, the patient again had high fever with desquamation of the fingertips and thrombocytosis. She was transferred to our Pediatrics Unit for further investigation. The girl refused to walk and showed leucocytosis, hyperferritinaemia and hypertrigliceridaemia. A bone marrow aspiration was performed and neuron -specific enolase was dosed, both resulting normal. On clinical and laboratory basis, she was diagnosed with Systemic Juvenile Idiopathic Arthritis. Therapy with ASA was continued and steroid therapy with IM Methylprednisolone and then oral Prednisolone (10 mg/day) was started. The girl resumed a normal gait, experiencing stable remission while tapering steroid therapy at two-month follow-up.

AN ATYPICAL PRESENTATION OF FANCONI ANEMIA
UN CASO ATIPICO DI ANEMIA DI FANCONI

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Fanconi Anemia (FA) is a rare autosomal X-linked recessive disorder due to mutations in 16 genes, whose protein products collaborate in a DNA repair pathway. FA is characterized by physical anomalies (even though 1/3 of patients have no dysmorphic features), progressive bone marrow failure, hypersensitivity to DNA cross-linking agent and predisposition to malignancy. The usual diagnostic test for FA involves detection of chromosomal breakage in peripheral blood T lymphocytes cultured with a clastogenic agent such as diepoxybutyrate (DEB test). We report a case of a four-year-old caucasian boy affected by acute lymphoblastic leukemia (ALL) and enrolled in "AIEOP LLA 2009" protocol. Starting from day 38, during induction chemotherapy with 6 mercaptopurine and citarabine, he experienced prolonged neutropenia (absolute neutrophil count <500/mm3) for 28 days. Chemotherapy was interrupted and, on day 70, bone marrow biopsy was performed and medullary aplasia was reported. He underwent DEB test which confirmed the clinical suspect of FA. His therapy was therefore switched to sibling transplant, and bone marrow performed one year post-transplant achieved complete remission. Our case report is atypical because FA was diagnosed after leukemia due to patient hypersensitivity to chemotherapeutic drugs. Moreover, most FA are associated to myeloid (not lymphoblastic) leukemia.

####

EVALUATION OF THE STATE OF FILLING AND REHYDRATION IN PATIENTS AFFECTED BY NDI
VALUTAZIONE DELLO STATO DI RIEMPIMENTO E REIDRATAZIONE IN PAZIENTI AFFETTI DA DIABETE INSIPIDO NEFROGENICO

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A 21-month-old boy, affected by NDI and in treatment with hydrochlorothiazide and indomethacin, presented with 7 days of fever, episodes of vomiting, somnolence during the previous 24 hours, and a reported reduction in his water consumption. At first, weight was 9.730 Kg (9.870 kg one month before), apyretic, with poor clinical conditions, dry mucous membranes, crying without tears, capillary refill of 2 seconds, cool hands and feet, and normal skin elasticity. Blood pressure and cardiac frequency were 102/66 mmHg and 110 beats/min respectively and the pharynx was reddish. Creatinine 0.65 mg/dL (with eGFR of 70 mL/min/1.73m2), sodium 154 mEq/L. Indomethacin administration was stopped and intravenous 5% dextrose administration (30 ml/h) was started. After 12 hours, sodium 148 mEq/L and weight 9.780 kg. The child was sleepy with a fluid balance diary showing an input of 460 ml and an output of 430 ml (calculated on the basis of diaper weights). Physical examination showed a tense abdomen, painful during palpation, with dull sounds in hypogastric, umbilical and left iliac regions. Ultrasound showed a vesical globus extended 5 cm above the umbilical line with bilateral hydroureteronephrosis. The urinary catheterization drained 900 ml of urine with a subsequent new body weight of 8.830 kg. At this point, the velocity of infusion of 5% dextrose was modified to 50 ml/h, registering an improvement of the patient's general clinical conditions after 24 hours (the body weight was 9.2 kg). After three days, blood examinations showed sodium of 142 mEq/L and creatinine of 0.25 mg/dL. The catheter was removed and the patient was discharged. The management of dehydration in patients affected by NDI (similar to patients affected by nephritic syn-

drome) is different. It's important to consider the risk of hypernatremic dehydration, in which the skin turgor may be normal, and the thirst reduction should always be considered as an important red flag of severe dehydration.

####

DWI-MRI AS A GOLD STANDARD IN DIAGNOSIS AND FOLLOW-UP OF FEBRILE UTI?
DWI-MRI COME GOLD STANDARD PER LA DIAGNOSI E IL FOLLOW UP DELLE IVU FEBBRILI?

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A 4-year-old girl presented the first episode of urinary tract infection (UTI) by E. coli ESBL, with fever and dysuria, treated with amoxicillin/clavulanic acid therapy with apparent benefit. Ultrasonography (US) images of the urinary tract were normal and laboratory tests were not available. One month later, fever (39°C) and dysuria reappeared, with urine culture still positive for E. coli ESBL, non responsive to amoxicillin/clavulanic acid treatment. Laboratory tests showed neutrophilic leucocytosis and elevation of CRP (22.48 mg/dl). We performed US of the urinary tract with only evidence of a minor dilatation of the left renal pelvis. Then, a diffusion-weighted imaging (DWI) MRI was performed, showing, instead, multiple focal inflammatory areas of the right renal parenchyma. We started therapy (meropenem) with fast improvement of clinical conditions, decreased inflammatory markers and a sterile urine culture. Six months later, a DWI-MRI was repeated and showed a complete restitutio ad integrum of all the focal inflammatory areas, with no residual renal scars. UTI is the most common infectious disease in the pediatric population and this case report highlights the importance of using MRI DWI-sequences in making accurate diagnosis in patients with febrile UTI, because it can be performed quickly without exogenous contrast medium and irradiation for the patient. It is able to detect renal parenchyma involvement early on and to predict treatment response and future renal scarring.

####

ATAXIA WITHOUT NEUROLOGIC DEFECT OR ARTHRITIS WITHOUT ARTHRITIS?

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A 2-year-old girl was admitted to the hospital because of gait disturbance and refusal to walk, especially in the morning. No trauma, infectious episodes or autoimmune disease history. Two days before admission, she had the left lower limb in extra-rotation and instability in direction changes. RX scan of limb and hip and US were negative. During the admission neurological evaluation, she showed ataxic deambulation. Blood exams: PCR 54 mg/L. She had a fever (max T 37.8 ° C) for 4 days. Joint examination seemed normal, even if a careful physical examination was not possible due to poor compliance. US abdomen and brain MRI excluded masses. Rx thorax, EEG, urine catecholamines, Mycoplasma antibodies and PCR for neurotropic viruses were normal. The symptoms were self-limited. PCR decreased and she was discharged with a diagnosis of mild cerebellitis. Afterwards, she had no daily fever or stiffness of the lower limbs. After 3 weeks from the onset of symptoms, arthritis was noted on the elbow, knees and ankle bilaterally, and some joints of the hands. The exams showed an increase of PCR, ferritin and IgG; titer ANA, RF and PPD were negative. Considering wide joint involvement and lack of response to NSAIDs, polyarticular infiltration of steroids was performed and methotrexate was started. Periodic slit-lamp ophthalmic examination will be performed for detection of uveitis. Diagnosis: polyarticular

juvenile idiopathic arthritis with systemic symptoms.

####

AN UNUSUAL ABDOMINAL PAIN
UNA CAUSA INSOLITA DI ADDOMINALGIA

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C, a 6-year-old girl, was admitted to the hospital because of colicky left flank pain, nausea and vomiting, no fever. Physical examination showed no pathological features, no abdominal bulging, urine strip test was normal. Clinical history presented prenatal detection of minimal left hydronephrosis and an episode of urinary tract infection. The abdominal US scan showed a severe left hydronephrosis (pelvic APD 50 mm). A diagnosis of ureteropelvic junction obstruction (UPJO) was made, and she was admitted to the Nephrological department. There, a Color-Doppler US was performed with detection of an accessory left renal artery originating from aorta and crossing the UPJ. MR confirmed the severe left hydronephrosis due to a crossing vessel (CV) and showed an important decrease in left kidney function (separated GFR of 90% for the right and 10% for the left kidney). Antenatal US allows for early diagnosis of UPJO in most children. However, a lower polar CV causing extrinsic UPJO is usually undetected at routine scans. Color-Doppler US may be useful but is not routinely performed. Moreover, occasional colicky pain in children may be attributed to gastrointestinal rather than renal causes. These two facts may lead to a delay in referral. A recent study shows a high association of CV in children over the age of 2 and of female gender, abdominal pain, other anomalies and poor preoperative function of affected kidney on imaging studies. Calyceal dilatation is also more prominent in the UPJO due to CV than in the intrinsic one.

####

A STRANGE LESION OF RIGHT FOOT

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R.M., male, 11 years old, arrived in First Aid for pain at th right foot dorsum arising after trauma. Right foot Rx showed a lesion of the third metatarsus, interpreted as enchondroma. Total Body Scintigraphy demonstrated an increased signal uptake at right foot dorsum. After 1 year, a new Rx showed a volumetric increase of the lesion. For this reason, the patient underwent surgical intervention of curettage. Post-intervention Rx showed a thinning of cortical bone, poorly recognizable. The suspect of a malignant lesion was confirmed with bone biopsy which identified an osteosarcoma, grade IV, SATB2+, CD68+, S100-. No metastasis was found. The patient started chemotherapy according to protocol ISG-OS2. Enchondroma is one of the most common bone tumors, while osteosarcoma rarely occurs in the foot and typically produces dense ossified tumor matrix (internal areas of high density) within or around a bone and can produce an aggressive subperiosteal new bone formation. With tumor growth, destruction of bone typically results in a permeative pattern on radiographs with associated soft-tissue mass. In our patient, the radiological pattern of the lesion is not typical for a malignant lesion, while the pain and the increased uptake of signal of scintigraphy are more characteristic.

####

WHEN IS ENOUGH MORE THAN ENOUGH?
QUANDO È TROPPO, È TROPPO

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A 13-year-old boy presents to the E.R. with a three week history of distal leg

pain and migrant skin swellings, and ambulation possible only with crutches. He is depressed and has stopped attending school and doing sports. During physical exam, his mother starts listing his numerous diseases (allergies, celiac disease, GER, gastritis, AVM, epilepsy, diabetes) showing all the investigations performed: over 10 years, he underwent 9 US (for abdominal pain, low back pain, leg pain, DVT; some Doppler), 3 X-Rays, 9 MRIs (brain and legs, 3 with contrast), 1 EGD test, 4 EEGs, 2 lower gastrointestinal series (for stubborn constipation first, then encopresis), urodynamics, anorectal manometry, rectal biopsy (suspecting idiopathic megarectum) together with countless amounts of medical advice in gastroenterology, neurology, rheumatology, surgery, never receiving a certain diagnosis nor solving the reported symptoms. He takes carbamazepine without diagnosis of a specific type of epilepsy. Once offered EEG and a neuropsychiatric advice, his mother says they would be "useless". At physical examination, we only notice a strong pain after light pressure on both legs. We perform blood tests, legs MRI, EnoG and orthopaedic advice. The response is unanimous: radiological findings can't explain his symptoms and he must start walking again as soon as possible, in order to avoid algodystrophy. At discharge he's still using crutches, crying for pain.

####

SEIZURES AND…
CONVULSIONI E…

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A baby girl, birth weigh 2965 g (30th centile), was born at 39 weeks' physiological gestation. The Apgar score was 9-10 at 1 and 5 minutes. Referred clinical wellbeing up to 6 days of life, when she experiencend seizures and vomiting. Blood count, ionogram, blood glucose, CRP, coagulation, metabolic and thrombophilic profile were normal. Transfontanellar ultrasound scan revealed hyperechogenicity of thalamus/basal ganglia and superior sagittal sinus. Brain MRI and MRV showed a cerebral venous sinus thrombosis (CVST) of all cerebral veins with bilateral thalamic haemorrhage and periventricular hyperechogenicity. After 2 days, excluded acute bleeding, treatment with enoxaparin was started. After 37 days of anticoagulant therapy, the MRV showed a marked reperfusion of the cerebral venous sinuses. Currently, she is 4-years-old and presents a slight psychomotor delay accentuated in the last year, probably related to suspension of the rehabilitative and logopedic treatment. About 60% of neonatal CVST occurs within the first week of life. The most frequent manifestations are seizures, widespread or with focal neurological signs. The mortality rate is high, 33%-79% of patients report long-term sequelae: persistent epilepsy (40%), motor deficits (70%) and cognitive impairment (60%). Management of neonatal CVST remains controversial due to risk of bleeding. Despite the lack of randomized controlled trials, even in the presence of significant intracranial bleeding, treatment with UFH and LMWH should be considered in order to contrast propagation of the venous thrombosis.

####

AN INSIDIOUS ARTHRITIS
UN'ARTRITE INSIDIOSA

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A 14-year-old boy, affected by GH deficiency in treatment with rGH and history of dissecans osteochondritis of the knees, presented with right elbow pain and functional impairment appeared after a tennis match. Two weeks later, for the persistence of the pain, he was admitted with swelling, redness and antalgic flexion of the elbow. No history of infections, fever or morning stiffness. Laboratory findings and X-rays were normal, but ultrasound

examination showed joint effusion and synovial thickening. The first diagnostic hypothesis was reactive arthritis. Anti-inflammatory treatment with Ibuprofen and restriction of physical activities were started with improvement of pain but persistence of functional impotence. About 3 weeks later, due to fever associated with elevation of ESR and PCR, the hypothesis of septic arthritis was also considered. ASLO titer, PPD, pharyngeal swab and urine cultures were all negative and arthrocentesis showed hematic synovial fluid with negative microbiological examinations, so this hypothesis was excluded. Then computed tomography and magnetic resonance imaging were performed with evidence of humeral condyle dissecans osteochondritis and reactive synovitis. Treatment was based on analgesics and restrictions on impact activities. On this basis, the case was critically reconsidered: condition of short stature, recurrent dissecans osteochondritis and minor dysmorphic features with brachydactyly was suggestive of ACAN syndrome.

####

A SIMPLE DIAGNOSIS, A MULTIDISCIPLINARY MANAGEMENT. A CASE OF VITAMIN D-RESISTANT RICKETS
UNA DIAGNOSI SEMPLICE, UNA GESTIONE MULTIDISCIPLINARE. UN CASO DI RACHITISMO VITAMINA D-RESISTENTE

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Rickets can be divided in two families: Vitamin D-dependent and Vitamin D-resistant. The last one is less frequent, but an early diagnosis is important to start the right supplementation and reduce clinical complications. We describe the case of a boy who came to our attention at 1-year-old with signs of tibial varus deformity, ALP elevation (734 UI/L) and Hypophospatemia (2.5 mg/dl). Calcium (4.9 mEq/l), vitamin D (both 25OH and 1-25OH) and PTH were normal. His family history (mother with vitamin D-resistant rickets with no genetic determination) suggested an X-linked type, which led us to carry out a molecular analysis that looked for PHEX gene mutation. His history was characterized by a significant incidence of rickets complications: at 2-years-old, the boy had surgical correction for craniostenosis; since 3-years-old, he has undergone surgical correction of varus deformity of lower limbs; and he is on a periodic odontoiatric follow-up due to recurrent dental abscesses. Fortunately, there has not been any kidney involvement so far. An interesting aspect of his history is the immunological side: since 2016 he started suffering from conjunctivitis, which was identified in 2017 as vernal conjunctivitis, and it is now being treated with topical cyclosporine. Since the age of 2, he has had periodic fever with exudative pharyngotonsillitis compatible to PFAPA syndrome showed by the clinical presentation (almost no positive swabs) and the response to betamethasone.

####

STROKE AND CHICKENPOX: DID YOU THINK ABOUT IT?

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A 6-year-old girl presented sudden onset of hemichorea with no history of trauma, headache or other neurological symptoms. Nothing to highlight in her familial and clinical history, except for the heterozygotes mutation of coagulation factor II and a virulent VZV infection 7 months before. Blood tests, including first level trombophilic study and serology for CMV, HSV, PVB19, were negative, but EBV and VZV IgG were positive. Head CT showed hypodensity of the left pulvinar and MRI study confirmed a recent ischaemic insult in the left thalamo-capsular region. Contrast echocardiography was negative. She started ASA 5 mg/kg with gradual

complete resolution of clinical symptomatology. We saw her first 4 months after the event, documenting no residual clinical signs and reduction of the MRI ischaemic lesion. We suspected post-varicella angiopathy (PVA) and recommend ASA therapy for 2 years and imaging follow-up in one year. After one year still no clinical signs, but MRA showed a stenosis of some cortical tracts of the MCA with vessel wall CE. No other vascular district involved. A lumbar puncture was performed, showing a borderline specific antibodies index in the CSF. We started steroid and antiviral therapy on the hypothesis of a re-activation of PVA. MRA was performed after one, three and six months, showing a slowly improved neuroradiological imaging. PVA has some typical clinical and radiological features and can recur over a two-year-period from the infectious event.

####

A "TOO ACTIVE" BONE - UN OSSO TROPPO "ATTIVO"

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A 10-year-old girl presented with acute dorsal pain after probably a mild trauma. She was treated with anti-inflammatory drugs and osteopathic treatment with apparent clinical resolution. A few months later, she presented with acute right humerus swelling and pain. Furthermore, some erythematous-desquamative skin lesions appeared on the palm of the left hand and on the sole of the left foot. Blood exams showed only leukocytosis and inflammatory markers mildly increased. Right humerus radiography showed osteolysis and Computed Tomography (CT) revealed osteoaddensant bone lesions in the dorsal spine (D4-D5) and periosteal reaction in the right humerus. Positron Emission Tomography (PET) was positive only in humeral region. Bone biopsy of right humerus excluded neoplastic diseases. She was diagnosed with chronic osteomyielitis. Due to the persistence of pain she was admitted to our Hospital. Total body Magnetic Resonance Imaging (MRI) confirmed the presence of periosteal reaction in the right proximal humerus and showed a permeative cortical lesion in the same region. In agreement with oncologists and radiologists, we decided to perform another bone biopsy that showed chronic, non specific inflammation. Infectious diseases were also excluded. The combination of clinical, radiological, histopathological findings allowed us to diagnose SAPHO Syndrome. Treatment with bisphosphonates, oral steroids and sulfasalazine was started with clinical and radiologic improvement.

####

RECURRENT MICRO HAEMOPTYSIS FROM UPPER AIRWAYS: A CAUSE FOR CONCERN?

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A 13-year-old girl was admitted to Pneumology Unit with a 3 month history of recurrent emission of saliva tinted with blood, associated with upper airways congestion, no cough. Recurrent infections, bleeding, allergies denied. Menarche at 12 with regular cycles. No recent weight loss, fever or GI symptoms. No clinical benefit with tranexamic acid therapy, bleeding persisted with increasing frequency (weekly). First level blood tests were normal; coagulation screening showed mild reduction of PT, II, IV, VII clotting factors, no clinical significance according to haemostasis specialist. Negative PPD, normal chest X-ray. Infective and gastroenterological diseases excluded according to consultants. Nasal fibroscopy performed by ENT specialist showed mild adenoid hyperplasia (grade 2). Upon admission, normal physical examination, first level blood tests, coagulation, chest X-ray. Sensitization to house dust mites, normal spirometry. Fibro-bronchoscopy was performed,

showing hypertrophy of adenoids and lingual tonsil with submucosal telangiectasia; small plain submucosal angioma on the right side of trachea; no active bleeding. Hypertrophic lymphatic tissue and submucosal telangiectasia of lingual tonsil were likely responsible for symptoms. Recurrent infection and inflammation of oropharyngeal lymphatic tissues are common causes of blood emission from the upper airways, not abundant as in haemoptysis from lower airways and not associated with cough, but causing concern and medicalization.

####

A BURNING ESOPHAGUS
UN ESOFAGO IN FIAMME

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A 12-year-old male was admitted to the emergency department of G. Gaslini Children's Hospital because of stomach ache and some vomiting episodes (food and mucus), the last two episodes with bright red blood. He did not report any other symptom/signs. Ingestion of drugs or toxic substances were denied. He suffered from recurrent bronchospasm (dust mite allergy). His mother was celiac. Physical examination revealed epigastric tenderness, slight skin pallor with no sign of hypovolemia. Peripheral intravenous catheter was positioned and omeprazole administrated intravenously. Laboratory examinations showed normal complete blood count (Hb 14 g/dl, PLT 221000/mm^3) and coagulation. Albumin, transaminase, bilirubin, glycemia, amylase, C-reactive protein were normal. Chest radiograph and abdominal ultrasonography were normal. An hour after admission he presented another blood vomiting episode. Two hours after admission, Hb was 12.3 g/dl. Six hours after admission esophagogastroduodenoscopy was performed and showed distal esophagitis grade IV (Hassal) with linear furrows and white exudates in proximal esophagus. Histology examination showed: erosive esophagitis with maximum 50 eosinophils for HPF. He was treated with a cycle of PPI and topical steroid with histological remission at control after two months. Conclusion: children with Eosinophilic Esophagitis usually present with food impaction, RGE -like symptoms and dyspepsia, more rarely with hematemesis.

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ZEBRA VS HORSE: HYPOCALCEMIA IN COMPLEX PATHOLOGY
LA ZEBRA E IL CAVALLO: IPOCALCEMIA IN PATOLOGIA COMPLESSA

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Hypocalcemia is one of the most common disorders of mineral metabolism in children. It is caused by different conditions, mostly related to parathyroid hormone, calcium and vitamin D metabolism disease. Case Report: Male, 12,5 years old, surgically treated for tetralogy of Fallot at 4 months. On follow-up for vesicoureteral reflux, congenital hypoparathyroidism (CATCH 22 negative), iatrogenic nephrocalcinosis, transient hypothyroidism and poor growth. Replacement therapy with calcium carbonate and calcitriol. At 10-years-old, he started therapy with triptorelin for precocious puberty, suspended at 12. At 11 years old, he started to present repeated episodes of asthenia, irritability, paresthesia, with little alterations in calcium and phosphorus serum levels. Symptoms disappeared through adjustments in therapy. At 12-years-old, he presented acute hypocalcemia (muscle cramps, sign of Trousseau, laryngospasm, very low plasma calcium levels), and began treatment with intravenous calcium gluconate. During hospitalization, high levels of anti-TGA and HLA DQ2 positivity were detected. Histological results of mucosal biopsy specimen revealed duodenitis and celiac malabsorption syndrome stage 3B. Conclusion: Celiac disease is a rare cause of vitamin D deficiency rickets. The patient started a gluten free diet, with strong calcium and

vitamin D supplementation and resolution of hypocalcemia.

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NEUTROPENIA: SIGNAL OF AN UNDERLYING CONDITION.

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Occasional neutropenia was diagnosed in a 4-year-old boy, hospitalized for abdominal pain. After 10 days, he was re-evaluated for low-grade fever and asthenia; blood tests (BT) confirmed neutropenia associated with mild anemia. Diagnosis of post-infectious neutropenia was suspected, and hematological follow-up was recommended. Over the weeks, general conditions were good with occasional evening fever and hepatomegaly, no lymphoadeno/splenomegaly. Repeated BT confirmed leukopenia and severe neutropenia (median value 440/cmm) with normal platelets and mild normochromic anemia; no immature forms at peripheral smear, antibodies (Ab)/direct research of viruses (EBV, CMV, HBV) were negative. Urine and blood cultures excluded any infectious origin, while vaccine antigens response, immunoglobulin and IgG subclass dosage did not support immunodeficiency hypothesis. TCRab+CD4-CD8- (double negative–DN) T cells were increased (2.2%), so a lymphoproliferative disease secondary to one or repeated infections was suspected. The positivity of indirect antineutrophil Ab in serum reinforced clinical suspect of "auto/disimmune disorder". Two months later, due to the appearance of fever and persisting leuko/neutropenia, bone marrow aspirate was performed with evidence lymphoid blasts infiltrate, defining the diagnosis of acute lymphoblastic leukemia. In conclusion, the positivity of DNT cells plus IgM against neutrophils in an otherwise "healthy" child, were elements that mislead the true diagnosis.

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WHEN URTICARIA IS NOT FLEETING QUANDO L'ORTICARIA NON È FUGACE.

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Mastocytosis includes a group of disorders characterized by clonal expansion and accumulation of mast cells (MC) in various organs and tissues. Cutaneous mastocytosis (CM) is the most frequent manifestation with prevalence of 0.005-0.01% in Europe. Pediatric mastocytosis is uncommon, presenting mostly as isolated cutaneous lesions. We report a case of an infant with CM. Male infant aged 10 months, with no history of interest, hospitalized for multiple itchy brown macules from 0.2 to 2 cm of diameter. The lesions emerged at 4 months of age, initially on the trunk with centrifugal spread. He presented an isolated hyperchromic elevated cutaneous lesion on the right leg. Darier's sign was present. Serum tryptase (sT) was negative (4.48 µg/L): laboratory investigations and abdomen sonography excluded the systemic form. Skin biopsy was perfomed and showed MC infiltration (tryptase+, CD117+) occupying the dermis, confirming the diagnosis of polymorphic maculo-papular CM (MPCM). The patient was given antihistamine medication. In 2016, the WHO defined 3 forms of CM based on the extent and appearance of skin lesions: MPCM (monomorphic/polymorphic)(40-75%), mastocytoma(20-50%), diffuse CM(3-8%). Diagnosis requires typical skin lesions, skin biopsy and/or activating KIT mutation in lesional skin tissue. A sT> 20 µg/L demands bone marrow examination for systemic involvement. CM spontaneously regresses by adolescence and treatment is symptomatic.