ABSTRACT BOOK
ONSP DAYS 2015—12th ITALIAN MEETING
OF NATIONAL OBSERVATORY FOR TRAINEES AND YOUNG
PAEDIATRICIANS

Padua (Italy) 29 September—3 October 2015

ISSN 2279-7165 - Euromediterranean Biomedical Journal [online]
A STRANGE FISSURED TONGUE

C. Campanale¹, E. Sacco¹, M. Germano², M. Crisetti³, M.C. Sacco³

¹ School of Specialization in Paediatrics, University of Foggia, Italy
² Child Neuropsychiatry, IRCCS “Casa Sollievo della Sofferenza”, San Giovanni Rotondo (FG), Italy
³ Division of Pediatrics, IRCCS “Casa Sollievo della Sofferenza”, San Giovanni Rotondo (FG), Italy

Matteo, 6 yrs, presented with lagophthalmos and forehead not corrugated on the right, and buccal rhyme deviation on the left. He had a brother affected by type 1 diabetes mellitus. Matteo was discharged with diagnosis of Bell’s palsy and treatment with steroid therapy, B vitamins, artificial tears and ocular bandage overnight. After 1 month, he was hospitalized again for signs of contralateral peripheral facial paralysis and edema of the lips. Routine, infectious and liquor exams were normal. Audioimpedensiometric exam demonstrated a missing stapedial reflex on the left; ENG revealed the reduced amplitude of the motor response of the left facial nerve with increased distal motor latency compared to the contralateral. The brain MRI demonstrated the presence of a focal and nuanced enhancement of contrast solution in the first horizontal intrapetrosal trait of the 7th cranial nerve, and in the geniculate ganglion, both on the left side. During a careful neurological examination, Matteo’s tongue was fissured and a new diagnosis emerged: Melkersson-Rosenthal syndrome. Clinical and neuro-radiological control were improved after 20 days of therapy. Therefore, in a case of recurring and alternating peripheral facial paralysis associated with orofacial edema and fissured tongue, the possibility of Melkersson-Rosenthal syndrome should not be overlooked. It is a neuro-mucocutaneous granulomatosis and is described as a rare pediatric autoimmune disease, which requires close follow up due to the increased risk of associated autoimmune diseases.

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A SUSPECTED DIAGNOSIS OF NIE-MANN PICK DISEASE, TYPE B

A.Cioffi¹, A. Romano¹, P. Strisciuglio¹

¹ Department of Translational Medical Sciences, Section of Pediatrics - University of Naples Federico II

R. is 23 months old, his parents are Moroccans and consanguineous. He was well up to 9 months old, when he had a bacterial bronchopneumonia. During hospitalization an important hypertransaminasemia was detected. The parents reported a second hospitalization for febrile illness, postprandial vomiting, diarrhea hypertransaminasemia (not correlated to viral infections) and anemia at the age of 22 months. When R. arrived at our hospital we found: an important hepatosplenomegaly confirmed at ultrasound (liver size increased, right lobe 125 mm, left lobe106 mm, caudate 66 mm; Spleen size increased DL 135 mm), an accentuation of the vascular markings and defined radiopacity areas with scissural thickening on the right at chest X-ray, hypertransaminasemia (positive IgG and IgM antibodies to CMV without urine o blood CMV DNA), microcytic hypochromic anemia, hypercholesterolaemia and hypertriglyceridaemia, low positive values of specific IgE for milk associated with an increased total IgE. Global neurodevelopment was slightly delayed. The levels of serum chitotriosidase and plasma oxysterols were compatible with a diagnosis of Niemann Pick type B (NPDB). We are awaiting levels of sphingomyelinase activity on fibroblasts.

NPDB is caused by an inherited deficiency in acid sphingomyelinase activity. Patients show involvement of the spleen, liver, and lungs, but remain free of neurological manifestations despite the massive visceral involvement. They often survive into adulthood.

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POSTERIOR REVERSIBLE ENCEPHALOPATHY SYNDROME, AN URGENCY WITH A HAPPY ENDING

Ferretti M¹, Micalizzi C¹, Palmisani E¹,
Posterior reversible encephalopathy syndrome (PRES) is an acute clinico-neuroradiological entity seen in children with various associated conditions: malignancies, drug toxicity, autoimmune diseases and infections. Clinical manifestations may consist of headache, vomiting, visual abnormalities, seizures and altered mental status. Brain magnetic resonance imaging (MRI) would be the gold standard for diagnosis, and it should be performed promptly in order to remove the potential trigger factors. Typical finding is white-grey matter oedema involving the cerebral posterior regions. PRES is generally reversible, although severe life-threatening complications can also occur.

We describe the case of an 8 year-old boy with acute lymphoblastic leukaemia, who developed PRES during the induction phase of chemotherapy. His first symptoms were nervousness and agitation, followed by confusion, visual disturbances and seizures. Because of the rapid neurological worsening, an urgent head computed tomography was performed and revealed a bilateral cortico-subcortical hypodensity of the parieto-occipital lobes, suggesting a PRES. The electroencephalography showed diffuse slow activity with repeated elements in the occipital region. The first measures involved controlling hypertension, steroid tapering and initiation of an anti-epileptic therapy. The neurological outcome of the patient was favourable. The follow-up brain MRI showed complete resolution, although signs of previous hypoperfusion were detected.

Case 1. A previously healthy 15 year-old boy presented with sub-acute onset of vertigo, diplopia and nausea followed by an episode of loss of consciousness. On physical examination he showed hypotension, loss of motor coordination and somnolence.

Case 2. A 14 year-old boy presented a first episode of migraine with aura. A few days later, he developed vertigo, nausea, limb weakness and fatigue with a gradual worsening during the next three weeks. On physical examination, he showed signs of cerebellar dysfunction with ataxic gait and loss of motor coordination.

Both patients attended the same school. Further investigations, including neuroimaging (CT and MRI), EEG, lumbar puncture, serum and urine drug screen, showed no significant alterations. Serum titers for neurotropic viruses and Mycoplasma were obtained. EBV antibodies pattern documented a recent infection.

Discussion: ACA is a relatively rare condition occurring after viral infections or less frequently after immunization. It can be due either to direct infection or to autoimmune demyelination mechanism. This clinical syndrome is usually characterized by acute onset, rapid evolution and good prognosis. While case 1 showed complete resolution of symptoms within few days, case 2 had a slower resolution which occurred several weeks later. Since cases of ACA complicated by cerebellar atrophy and raised intracranial pressure have been reported, a strict clinical and radiological follow-up is recommended.
A COMPLEX CASE OF ESOPHAGEAL ATRESIA

D. Martinelli, M. Capozza, M. Baldassarre, N. Laforgia

Neonatal Intensive Care Unit-Hospital of Bari, University of Bari

We examine the case of a preterm born at 35 weeks and 5 days of gestation by C-Section due to polidramnios and with a prenatal diagnosis of esophageal atresia type III. After three days of life, he underwent surgical intervention with segmental resection of the esophagus with termino-terminal anastomosis. Back on the ward, he needed respiratory assistance with mechanical ventilation and subsequently with High Flow Therapy (HFT). He had exclusive parenteral nutrition for 9 days, beginning enteral feeding on the tenth day of life (six days post intervention). On 8th of January, during an endoscopic evaluation, a cartilaginous ring was found. Consequently, a further operation was planned. He was near to experiencing serious respiratory difficulty and there was ultrasonographic evidence of atelectasis. During hospitalization he has undergone tests for contrastographic control (negative), esophageal Impedance pH (negative) and broncoscopic evaluation (tracheomalacia). To complete the diagnostic process he was sent to Meyer Hospital in Florence. The endoscopic investigation underlined the necessity for surgical aortopexy for a vascular ring, performed in the hospital of Massa Carrara. On the 04/06/15 he was transferred to our U.O. for difficulty in oral feeding, and is now recovering a proper bottle suction capacity with increasing volumes of milk.

ACUTE THROMBOTIC AND HEMORRHAGIC COMPLICATIONS IN ACUTE PROMYELOCYTIC LEUKEMIA.

Mazzoni M¹, Micalizzi C¹, Palmisani E¹, Rossi A², Morana G², Dufour C¹.

¹Clinical and Experimental Haematology Unit, Giannina Gaslini Children's Hospital, Genova, Italy.
²Pediatric Neuroradiology Unit, Giannina Gaslini Children's Hospital, Genova, Italy.

Acute promyelocytic leukemia (APL) deserves special mention both for its clinical presentation (risk of developing thrombotic and hemorrhagic complications caused by the release of procoagulant factors and activators of fibrinolysis), and for its pathogenic molecular translocation mechanism which determines altered function of the receptor for retinoic acid, essential to myeloid differentiation. On 12/3/2013 Francesco, 8 years old, was sent, by his Pediatrician, to Gaslini Hospital’s emergency department, suspecting appendicitis. His blood test showed marked leukocytosis (250000/mm³) and thrombocytopenia (48000/mm³); the morphological evaluation of the peripheral blood smear was compatible with M3V APL. The next morning treatment with all-trans retinoic acid (ATRA) and chemotherapy were undertaken. 48 hours later the child developed an acute event characterized by left hemiparesis: head CT showed haemorrhagic lesions in the right parietal lobe, and brain MRI showed acute cerebral ischemia due to arterial ischemic multidistrict stroke. During the night, the child had seizures, so he was intubated and transferred to the ICU, where antiepileptic treatment and unfractionated heparin were undertaken. Chemotherapy and ATRA by nasogastric tube were successfully continued. Currently the child has completed the consolidation and he has achieved morphologic remission. Physiotherapy and speech therapy have been started, with progressive improvement: now he can walk autonomously.

THE BOY WITH “LITTLE BIT STRANGE FACE”: A MENKES DISEASE CASE-REPORT

V. Chierici ¹, A. Marolda ¹, C. Peruzzi ², A. Papa ², M. Viri ², G. Bona ¹

¹Clinica Pediatrica, A.O.U. Maggiore della Carità, UPO, Novara
²U.O. Neuropsichiatria Infantile, A.O.U.
Maggiore della Carità, Novara

Male, born at term, AGA, biparietal cephalohematoma, left clavicle fracture. 1° hospitalization at 7 days: palleness, excessive crying, poor weight gain. At 45 days new admission for ALTE: cyanosis, hypotonia, staring and loss of consciousness. Anemia (Hb 6.4 g/dl), UTI, GER were found. Normal EEG. At 4 months new hospitalization: hyporeactivity, lack of appetite, vomiting and face abnormalities (prominent frontal skull bones, flat top, head dermatitis, “moon” face). Psychomotor retardation. He had focal seizures and dyskinesias, EEG with right temporal-occipital focalities. Started Phenobarbital and Levetiracetam. Brain MRI with after seizures alterations of left hippocampus, extreme tortuosity of brain vessels and platybasia. Normal CGH Array setting. He underwent genetic analysis of ATP7A gene mutation for suspected Menkes syndrome (positive in the child and in his mother). At 11 months, he needed midazolam and continuous morphine infusion due to a dystonic state. At 14 months, he underwent percutaneous endoscopic gastrostomy. At present, he is staying in a long-term care center.

Menkes syndrome (Menkes sdr) is a rare (1/300000) inborn error of metabolism in which cells can absorb copper but are unable to release it. It is caused by a defect in the ATP7A gene that is inherited. The gene is on the X-chromosome so mothers that carry the defective gene can transmit it to sons. Symptoms are: bone spurs, kinky hair, feeding difficulties, irritability, hypotonia, low body temperature, mental deterioration, rosy cheeks, seizures, skeletal changes, bladder diverticulosis.

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LETHAL SEPSIS BY AEROMONAS SOBRIA IN A PATIENT WITH ACUTE LYMPHOBLASTIC LEUKEMIA

G. Del Baldo, E. Franceschini, N. Caporelli, M. Rossi, P. Coccia, P. Pierani

Pediatric Hematology-Oncology, Department of Pediatrics, Università Politecnica delle Marche, Ancona

Aeromonas Sobria (AS) is a rare cause of systemic infection occurring both in immunocompromised and competent subjects, with a wide range of severe manifestations. A 14 year old boy affected by acute lymphoblastic leukemia, was admitted to our department because of chest pain and limb edema occurring during re-induction phase after anthracycline administration. Electrocardiogram and chest X-ray were negative. Blood tests showed neutropenia, thrombocytopenia and hypokalemia. He was started on anti-viral, bacterial and fungal prophylaxis. The day after, Gabapentin and Tramadol were started for persistent chest pain, pleural and abdominal effusion were found on CT and Aspergillus antigen was detected in serum. On 3rd day he experienced deterioration of clinical condition with fever and severe cutaneous hyperesthesia; within a few hours, he showed erythematous lesions similar to those for necrotizing fasciitis, severe hypotension and obtunilation. He was started on Meropenem and Vancomycin, and admitted to intensive care for septic shock. He died after 3 hours due to multi organ failure. Blood culture was positive for AS after death. AS is generally isolated in water, seafood and vegetables. In our patients, we suspected infection by contaminated fish. Average time of death is 3 days, as in our case. In immune-compromised patients with signs of necrotizing fasciitis, it is important to consider the possible involvement of AS. Early recognition and aggressive therapy are the primary determinants of outcome.

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RECURRENT HYPOGLYCAEMIA IN TYPE-1 DIABETES MELLITUS MAY UNRAVEL THE ASSOCIATION WITH ADDISON’S DISEASE: A CASE REPORT

T. Timpanaro, S. Passanisi, D. Lo Presti, M. Caruso-Nicoletti

Pediatric Department, AOU Policlinico-
Vittorio Emanuele, Catania, Italy

A 16-year-old boy had been affected by T1DM from the age of 16 months. At the age of 4 years, autoimmune thyroiditis was diagnosed and treatment with levo-tiroxine was started. During a follow-up visit, he reported episodes of hypoglycaemia in the preceding three months; one of these episodes was severe with loss of consciousness, and required admission to the hospital. The patient was reevaluated three months later: other episodes of hypoglycaemia had occurred with one episode of severe hypoglycaemia characterized by seizure, and treated with glucagon. Glycated haemoglobin was significantly reduced, routine blood chemistry resulted negative, excluding the presence of liver and kidney diseases. Screening for celiac disease was also negative. Hence, we decided to further reduce the total dose of insulin. At the following visit, the patient reported frequent hypoglycaemic episodes and marked asthenia. At clinical examination, the patient presented scrotal hyperpigmentation and blood pressure = 80/50 mmHg. Adrenal insufficiency was immediately suspected, and the patient was admitted to the hospital for further investigation. The diagnosis of AD was confirmed and cortisone acetate was prescribed. T1DM, thyroiditis, and AD are associated in a clinical disorder known as Schmidt’s syndrome or type 2 autoimmune polyglandular syndrome. However, an early onset of T1DM and thyroiditis in the first years of life could precede the disease.

SEVERE SINUSOIDAL OBSTRUCTION SYNDROME AS AN ACUTE COMPLICATION OF LEUKEMIA TREATMENT: A CASE REPORT.

M. Leoni (1), M. Ferretti (1), C. Micalizzi (2), M. Faraci(3), E. Lanino(3).

(1) Scuola di Specializzazione di Pediatria – IRCCS G Gaslini, Genova – Università degli Studi di Genova.
(2) U.O. Emato-oncologia Pediatrica – IRCCS G Gaslini, Genova.

Gabriele, 6 years old, suffering from acute lymphoblastic leukemia B-cell common high risk, is in treatment according to the protocol AIEOP-ALL-2009. After the first reinduction cycle, containing thioguanine, he presented transfusion-refractory thrombocytopenia, with rapid consumption of transfused platelets. He was admitted to our hospital because of fever and diarrhoea with neutropenia. Piperacillin + tazobactam and amikacina were started. At laboratory tests: C-reactive-protein (CRP) 5 mg/dl, AST/ALT 106/114 U/l, and bilirubin 3,81 mg/dl; blood cultures negative; clostridium difficile was found, so metronidazole was started. Later, he worsened: he presented respiratory insufficiency, weight gain, and hepatomegaly. Blood tests revealed CRP 8,86 mg/dl, AST/ALT 1721/675 U/l, GGT 106 U/l, bilirubin 13,22 mg/dl (direct 8,87 mg/dl), and alteration of coagulative parameters (PT 22%, aPTT 61,9 sec, D-Dimer > 30 mg/l). Chest’s Computed Tomography revealed bilateral pleural effusion and right lung consolidation; abdominal ultrasound revealed ascitis, hepatomegaly, and reverse portal flow with normal sovra-hepatic veins the consequent diagnosis was of Sinusoidal Obstruction Syndrome (SOS). We started therapy with defibrotide, associated with supportive care and abdominal drainage, which resulted in progressive resolution of the disease. SOS is a potentially life-threatening syndrome, which can complicate antineoplastic treatment; the best therapy, also in severe cases, is defibrotide.

ADEM: WE SHOULD THINK ABOUT PLASMAPHERESIS

G. Brindisi

Aldo Moro University, Bari

FC, sixteen year old girl, with fever for three days, asthenia, frontal headache
treated with antipyretic, antibiotic and cortisone. Later apyrexia, vomit, lack of appetite, pains in lower limbs, difficulty walking, constipation and urinary disorders appeared. At this point, she was admitted to the Giovanni XXIII Hospital Neurology department in Bari. On admission: lower limbs flaccid: paraplegia, TRO absence, pain and tactile hypoesthesia, Lasegue positivity, upper extremity weakness, nuchal rigidity, c.n. integrity, good space-time orientation.

Laboratory examinations: increased CRP and fibrinogen, Ab Anti Borrelia and Mycoplasma positivity. CSF analysis: intrathecal synthesis of oligoclonal IgG and G paraproteins. Negativity of Ab anti GM1,GM2,NMO.

Brain-marrow RMN: focal hyperintense unmodified after contrast, medium injury in left frontal parietal area and from D1 to D10. Clinical signs compatible with acute disseminated encephalomyelitis. Oral cortisone therapy was started with acyclovir, ceftriaxone and clarithromycin. Then administration of Ig iv and initiation of plasmapheresis (9 sessions), with a gradual reduction in leg weakness and regaining of sphincter control. Discharged after one month with a referral for rehabilitation treatment.

Clinical improvement confirmed in follow-up examinations. Brain-marrow RMN three months after: absence of altered signal areas in brain or marrow, in support of the efficacy of the apheresis treatment performed.

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BILATERAL FACIAL NERVE PARALYSIS: A CASE REPORT

Franceschini E1, Del Baldo G1, Antonini L1, Cionna C1, Cesaroni E2, Zamponi N2.

1Department of Paediatrics, Università Politecnica delle Marche, Ancona, Italy
2Pediatric Neurology Department, Ospedali Riuniti, Ancona, Italy

An 11-year-old male presented with a 2-day history of asthenia, headache, vomiting, neck pain mainly on the right side, difficulty in closing both eyes and eating and impairment of taste. Symptoms of upper tract respiratory infection were reported three weeks before, no history of travel, insect bites, facial or head injury. At admission, he showed bilateral facial nerve paralysis, more evident on the right side of the face, without any other neurologic symptoms. Brain MRI and electroencephalogram were normal. Serological tests revealed IgM anti-CMV and anti-GM2 ganglioside antibodies. Spinal fluid analysis showed significantly elevated albumino-cytologic dissociation. Nerve conduction studies confirmed widespread demyelination involving the spinal roots, peripheral nerves and bilateral facial nerves, leading to a diagnosis of Guillain-Barré Syndrome. The patient received two courses of intravenous immune globulin and oral steroid therapy. During the hospital stay the patient showed a total recovery of the left facial nerve in four days, with residual deficit of the right facial nerve. He never manifested limb paresthesias, hyporeflexia or paralysis; no other cranial nerves were involved. At follow-up, after 1 month, the electroneurography showed axonotmesis of the right facial nerve and normal conduction of the left one. Bilateral facial palsy is very uncommon. The diagnosis of Guillain-Barré syndrome should be taken into account, even without limb hyporeflexia or paresthesias.

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THE DARK SIDE OF PERINATAL ASPHYXIA

M.S. Lieggi, L. De Cosmo, N. Laforgia

UO di Terapia intensiva neonatale e Neonatologia – Azienda ospedaliera Policlinico di Bari
Università degli studi “Aldo Moro” di Bari

S.S. is a baby born post-term at 42 + 4w of EG. Family history silent, non-consanguineous parents and sister in good health. In obstetric history only polyhydramnios and poor fetal movements were detected. After birth, the baby was transferred to our NICU due to severe perinatal asphyxia (Apgar 2/3): she showed a very serious generalized hypotonia, associated with an absolute absence of spontaneous motility, ROT
A RESTLESS CHILD.

E. Sciorio¹, A. Romano¹

M., a 7 year old boy, presented at our hospital suffering from multiple worsening motor and vocal tics for 6 months. During hospitalization, high CPK levels and localized, non-traumatic, bruising on both upper and lower limbs were noted. Toxic and infective causes were excluded, as inflammatory markers, throat swab and EBV were negative. MRI, EEG and SPECT showed no pathological findings. Epilepsy was also excluded. The YGTSS (Yale Tic Severity Scale) showed a score of 80/100. According to the Diagnostic and Statistical Manual of Mental Disorders DSM V criteria and in view of the recent onset of symptoms (less than 12 months), the condition of Transient Tics was diagnosed. A treatment with Pimozide (1 mg/day) was therefore started. After 48h of treatment, a prompt clinical and laboratoristic response was observed, with reduction of tics, resolution of limb bruises and CPK normalization. Currently, after 17 months, M. still suffers from tics. Because of symptoms persisting for more than 12 months, according to DSM V criteria, the diagnosis was changed to Tourette Syndrome. Tourette S. is characterized by multiple motor and vocal tics starting before the age of 18. These tics tend to vanish at maturity and may be associated with Attention Deficit Hyperactivity Disorder (ADHD) and Obsessive Compulsive Disorders (OCD).

NOT ALWAYS ANXIETY

C. Ciullo, G. Raimondi

Scuola di Specializzazione in Pediatría, Università di Bari

M., a 12-year-old girl, arrives at the Emergency Room for tremors spreading to the whole body. She has a history of allergic asthma and is on therapy with antihistamine and fluticasone propionate-salmeterol inhalation. Her mother reported that after the administration of

¹Department of Translational Medical Science, Section of Pediatrics, University of Naples "Federico II".

and sucking, while the ocular motility was preserved. We began to assume that it could not be a simple case of asphyxia, so we began to perform an extended panel of investigations (karyotype and array-CGH, gene for Prader-Willy, surveys specific metabolic); however, everything was normal. Even the instrumental investigations performed (echocardiogram, renal ultrasound, ultrasound and MRI brain) were negative, as well as the muscle biopsy, which revealed signs of nonspecific myopathy. The only data which turned out to be useful were the EMG (which revealed a lack of spontaneous and electrically induced motility bilaterally at the level of all the muscular stretches analyzed) and the X-ray of the skeleton (which revealed the presence of multiple fractures). We decided to take a molecular analysis of DNA, which demonstrated the presence of a point mutation of the gene SMA1 (that was inactivated) and only 1 copy of the SMA2, whose number of copies is inversely proportional to the severity of the clinical manifestation.

At this point we diagnosed SMA 0, a very serious and extremely rare variant of spinal muscular atrophy, which has fetal onset and is lethal within a few months of birth. SMA is an autosomal recessive genetic disorder (second only to cystic fibrosis), characterized by the degeneration of the anterior horn of the spinal cord; usually it occurs in three forms (1-2-3) with variable onset and clinical picture, characterized by hypotonia and generalized weakness of muscles, with reduction of ROT and conservation of eye motility. The most severe forms have early onset and lead to death of the patient within a few years of life. They cause the progressive involvement of the muscles involved in swallowing and breathing, which intensify a clinical picture which is already compromised.

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the asthma therapy, M had a fight with her father followed by symptoms of tremors and palpitations. An anxiety crisis was suspected, and M was taken to the Emergency Room. On the arrival M had good functional parameters: heart rate 126, 100%O₂ saturation, blood pressure 75/96mmHg. The objective examination was negative except for coarse tremor, which was not resolved by distracting her nor by administration of placebo. Investigating the history in more detail, we found that the same morning M. had started a new therapy for asthma control: 25mg of prednisone and 4 puffs of salbutamol and that the tremors had begun a few seconds before the fight with her father. This new therapy had been prescribed the previous afternoon by the specialist, because the spirometry performed by her had shown a worsening in asthma symptoms. On this basis, the specialist had decided to change the therapy, although the previous therapy had been able to completely control the asthma. Consequently the tremors that M. was presenting were actually due to an overdose of medication in an asymptomatic child. The tremors resolved spontaneously after about an hour and the therapy was suspended.

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**A CRAZY HEART**

M. Arigliani¹

¹Department of Clinical and Experimental Medical Sciences, Unit of Paediatrics, University-Hospital of Udine, Udine, Italy

Monkole Hospital, Kinshasa, D.R. Congo. We received a full term infant who was born a few hours before elsewhere. The pregnancy and vaginal delivery occurred without complications. At birth, the baby presented with the heart outside the chest. He didn’t need any assistance. The child had a complete ectopia cordis: an open sternum and extrathoracic heart covered only by visceral pericardium. At presentation he was pink, awake, eutrophic, mildly polyphonic with SpO₂ 94%. He had low-set ears and mild micrognathia, no other dysmorphisms. He had a regular heart rate of 130 bpm while sleeping, with a 2/6-heart murmur in mesocardium. Neurological and thoracic-abdominal examination was regular. A cardiac US showed a small atrial septal defect; heart chambers, the arrangement of the great vessels and diaphragm were normal; cardiac output was good. An abdominal US was regular. On day 7th he showed signs of sepsis with hypothermia, pallor and increased CRP. He died on 8th day of life.

**DISCUSSION**

Complete ectopia cordis is extremely rare. Many cases have an associated intra-cardiac defect. If untreated, it is universally lethal. There are only few reported survivors following surgical correction which is usually a staged repair aimed to: (1) provide soft tissue cover of the heart; (2) reduce the heart into the thorax; (3) palliation or repair of any intra-cardiac defect; and (4) reconstruction of the chest wall. Unfortunately in D.R. Congo there is no Pediatric Cardiac Surgery.

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**WHEN PULSE OXIMETRY SUGGESTS A DIAGNOSIS**

M. Arigliani¹, E. Valentini¹

¹Department of Clinical and Experimental Medical Sciences, Unit of Paediatrics, University-Hospital of Udine, Udine, Italy

Luanda, Angola. Malnutrition Center, afternoon of December 31st 2014. A 13 month-old child is admitted with moderate malnutrition, three-day fever and cough. She appears well, febrile and mildly tachypneic, SpO₂ 90%. Cardio-respiratory and abdominal examinations are normal. Mother’s HIV status is unknown. Radiology is already closed. Nasal oxygen and I.V. penicillin G are started empirically, suspecting pneumonia for low SpO₂, which seems disproportionate to clinical signs. Laboratory results will arrive on January 2nd, when we revaluate the child: now she is very sick, febrile,
pale and drowsy. Respiratory rate is 75/min with paradoxical breathing and SpO2 85%. Her heart rate is 180 bpm, refill time <2 sec. Cardio-thoracic auscultation is negative. She has neutrophilic leucocytosis, CRP +++, positive HIV test. A chest x-ray shows bilateral perihilar infiltrates.

We increase oxygen and start I.V. cotrimoxazole and oral prednisolone suspecting severe Pneumocystis pneumonia. This was my last day in Angola. The child died two days later.

**DISCUSSION**

Pneumocystis carinii pneumonia is the main cause of death in HIV+ infants. Low SpO2 with a negative thoracic auscultation is common. Chest x-ray shows bilateral perihilar infiltrates, becoming increasingly homogeneous and diffuse as the disease progresses. This type of pneumonia should be suspected in African infants with respiratory infection and severe desaturation when thoracic auscultation is negative and maternal HIV status is unknown.

### MARKET SURVEY ABOUT THE USE OF IRON CHELATION THERAPY IN ITALIAN THALASSEMIC PATIENTS

F. Urbano¹, R. Conte², F. Bonifazi ², G. Lassandro ¹, M. Valente ³, G.C. Del Vecchio, P. Giordano¹

¹Università degli Studi di Bari – Dipartimento di Scienze Biomediche e Oncologia Umana Azienda Ospedaliero Universitaria Consorziale Policlinico-Ospedale “Giovanni XXIII” – U.O. Pediatría “Bruno Trambusti”-Bari
²Fondazione per la Ricerca Farmacologica Gianni Benzi Onlus – Valenzano (BA)
³Università Commerciale “Luigi Bocconi” – Milano

β-thalassemia is an inherited disorder of hemoglobin synthesis characterized by severe anemia. Sufferers are dependent on regular transfusions which are responsible for toxic iron overload. The main complications related to iron overload affect liver, cardiac, musculoskeletal and endocrine system and have an effect on clinical history as well as cost. The introduction of iron chelation therapy, has on the one hand reduced the primary manifestations of the disease leading to an improvement of expectation and quality of life, but on the other hand led to the onset of complications and comorbidities secondary to the adverse events of therapy itself. The main aim of the study is to characterize the economic impact of iron chelation therapy and of its complications in 272 thalassemic patients belonging to 60 Centres connected to the Interregional Register of Thalassemia and then to compare the different chelation regimens. Iron chelation therapy erodes the largest share of costs with significant differences according to the chelation regimen. The use of oral chelators, Deferasirox (DFX) and Deferiprone (DFP), is increasing among pediatric patients as a result of their better compliance and tolerability and affects the overall costs more than Deferoxamine (DFO). On the other hand, the combination DFO/DFP therapy is increasing because of its positive effect on morbidity and survival. However, there is no standardized protocol for the combination therapy according to a real risk-benefit ratio for patients as yet.

### IS THERE AN ASSOCIATION BETWEEN FUNCTIONAL GASTROINTESTINAL DISORDERS IN THE FIRST THREE MONTHS OF LIFE AND MATERNAL PSYCHOLOGICAL PROBLEMS?

P. Drimaco¹, N. Laforgia¹, A. Resta¹, S. Varano¹, G. Castoro¹, R. Maurogiovanni¹, M. Fanelli¹, M. Capozza¹, V. Santoiemma¹, M.E. Baldassarre¹

¹Neonatology and Neonatal Intensive Care Unit, Department of Biomedical Science and Human Oncology, University of Bari “Aldo Moro”, Bari, Italy

Aim of the study: to investigate whether functional gastrointestinal disorders are
associated with postpartum mood disorders. 113 mother/child pairs were enrolled in this perspective, longitudinal study. Maternal depressive symptoms were evaluated at birth, one and three months after delivery using Maternity Blues, Edinburgh Postpartum Depression Score and Symptom Check List for Anxiety and Depression. The Adult Attachment Interview was used to determine the attachment style of the mother. Any sign/symptom was recorded weekly for the first three months of life, together with type of feeding. Analysis: #2 test, student t-test, linear regression. Results: 37(32.7%) newborns were exclusively BF. 16(14.2%) newborns had regurgitation, 10(9.7%) colic, 4(3.5%) dischezia and 10 (9.7%) constipation. 60(53.1%) mothers had postpartum depression and/or anxiety. 53.6% of infants with regurgitation had a depressed mother vs 23% of infants without regurgitation(#2 =10.63, p= 0.003); 45.2% of infants with colic had a depressed mother vs 15.9% of infants without colic (#2 =10.63, p= 0.001). A mother’s insecure attachment style was found in 36% of infants with persistence of regurgitation until third months of life vs 1.8% of infants with mother’s secure attachment style (p<0.001). Postpartum maternal depressive symptoms and anxiety are associated with infantile colic and regurgitation. Early intervention in cases of postpartum depression could be useful to avoid inappropriate treatments.

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**PEDIATRICS IN ANCIENT GREECE: THE MYTH OF MELAMPUS**

F. Saettini¹, M.F. Olivieri², A.J. Kesel³, L. Bonalume⁴, F. Marzari⁵.

¹San Gerardo Hospital, University of Milano-Bicocca, Department of Pediatrics, Italy.  
²University of Milano, Department of Historical Studies, Italy.  
³Chammünsterstr. 47, D-81827 München, Germany.  
⁴University of Milano-Bicocca, Department of Psychology, Italy.  
⁵University of Siena, Centro Antropologia e Mondo Antico, Italy.

**Aim:** ancient Greek medicine has been the subject of thorough investigation, yet the history of Greek paediatrics is still in need of firm documentation. With this aim, we detail one of the earliest documented paediatric cases which can be traced to Melampus.

**The myth of melampus:** Melampus, a renowned soothsayer and healer of ancient Greek mythology, acquired the art of prophecy after his ears had been purged by some snakes. The most famous episode of his life was the healing of the daughters of Proetus, “Proetides”. Having offended a god, they were punished with madness, lust, vitiligo, and hair loss. Ancient sources from Hippocrates to Galen suggest that Melampus used helleborus to cure them.

**Discussion:** connected with the god Asclepius (Melampus was related to Apollo, the father of Asclepius), the presence of the snakes reinforces this character not only as a seer but also as a healer. Current research on helleborus proves this plant to be an effective drug with antidepressive, sedative, antipsychotic and anti-inflammatory effects. As we estimate the age of the Proetides to be between 14-18, this myth demonstrates the concern that existed for pediatric conditions; the Greeks searched for the causes of pediatric conditions and actively tried to cure them. The novelty represented by the story of Melampus lies in the use of herbs, and this was recognized by later authors in antiquity who named the hellebore ‘melampodion’.

**Conclusions:** this investigation provides insight into ancient Greek medicine and highlights Melampus among the founding figures of ancient pediatrics.

###

**LEISHMANIA...NOT ALWAYS ALONE...OFTEN IN BAD COMPANY!**

F. De Angelis¹, M. Grilli¹, L. Antonini¹, N. Caporelli¹, N. Oggiano¹, C. Catassi¹

¹Clinica Pediatrica, Ospedale Salesi Ancona, Università Politecnica delle Marche.
Leishmaniasis is an infectious disease caused by the protozoan Leishmania spp, occasionally associated with secondary infections. A case of a 5-year-old girl with a right basal pneumonia, persistent paracetamol-resistant fever and respiratory failure is discussed. She had been living in Senegal for the 2 months before admission; contact with a domestic dog which died during the following 6 months was reported. Initial examination and investigations revealed acute respiratory failure, drowsiness, hyperpyrexia, anemia, thrombocytopenia, hepatosplenomegaly, jaundice, inflammatory marker increase. Wide basal bilateral lung infiltrates, ground glassed areas, mediastinal lymphadenopathy was detected by Chest TC. She was assigned to the Intensive Pediatric Care Unit due to rapid clinical and radiological worsening and she was managed with high flows O2 therapy and IV cotrimoxazole and meropenem. Serologic tests were done in order to discriminate between an infectious and oncohaematological aetiology. The identification of parasites in the bone marrow smear and serum positive for Aspergillus antigen (galactomannan index (GMI) 7:0) led to a diagnosis of visceral Leishmaniosis with Aspergillus super-infection. She was started on IV Amphotericin B and Voriconazolo, showing good response. In Italy, visceral Leishmaniosis is a rare disease, and it can be associated with uncommon infections. However, in this case, early diagnosis and treatment led to gradual clinical and radiological improvement, until complete healing.

###

**TETANY AS A RARE PRESENTATION OF INTESTINAL OBSTRUCTION IN A CHILD**

Palpacelli A.\(^1\), Gatti S.\(^1\), Romagnoli V.\(^1\), Cobellis G.\(^2\), Catassi C.\(^1\)

\(^1\) Clinica Pediatrica, Ospedale Salesi Ancona, Università Politecnica delle Marche.

HEMOPTYSIS IN PATIENT WITH FEBRILE NEUTROPENIA IN ACUTE EARLY T PRECURSOR LEUKEMIA

Bertelli E.\(^o\), Castagnola E.\(\ast\), Svahn J.\(^o\), Micalizzi C.\(^o\)

\(^o\)Clinical and Experimental Haematology Unit, Istituto Giannina Gaslini, Genova

\(\ast\)Infectious Diseases Unit, Istituto Giannina Gaslini, Genova

We report the case of a duodenal obstruction and malrotation in a 15 month old girl presenting with signs of tetany. She was admitted with carpo-pedal spasms, dehydration and irritability and had vomited several times in the last 24 hours. At barium meal examination signs of malrotation were found, and consequently, Ladd’s procedure was performed. Seven days after surgery the girl newly presented with several episodes of bilious vomiting and progressive deterioration. A repeated upper endoscopy finally showed the presence of impacted food bolus in the first part of the duodenum and after removal, an incomplete duodenal membrane was found between the first and the second portion of the duodenum. The web was surgically removed and a duodenoplasty was performed. Congenital intestinal abnormalities are frequently detected in the first days/months of life. Congenital duodenal obstruction is an uncommon condition with an incidence of 1 in 4000-15000 live births. The association between duodenal obstruction and small bowel malrotation has rarely been described. The presence of another intestinal abnormality is a possible confounding factor in the identification of duodenal membranes. The most common presenting sign of both the conditions is vomiting with bile or without, a hypochloraemic, hypokaemic metabolic alkalosis with hypocalcemia leading to tetany is rarely seen in modern medicine especially in children.
Case report: 16-year-old girl, diagnosis Early T Precursor Leukemia (ETP) one month before, chemotherapy with LAL 2009 HR (prednisone poor responder-PPR). At h 22.00 of 26th day of IA induction, including dexamethasone, she develops febrile neutropenia (WBC 100/uL). After blood cultures she receives empirical therapy with ceftriaxone and amikacin according to internal protocol. Clinical condition improves and she remains afebrile for 24 h. The following day at h 04.00 she suddenly develops cough with hemoptysis. At this time fibrinogen is 167 mg/dL, and platelets 35000/uL. Chest X-ray shows thickening of basal medium right field, and chest CT scan shows a nodule with central hypodense area (necrotic-colliquiative evolution of the infiltration). In spite of supplementation with platelets, red blood cells, plasma, fibrinogen, antithrombin, activated factor VII, clinical condition worsens rapidly and the pulmonary hemorrhage becomes uncontrollable. She died at h 12.00. Blood cultures yielded a strain of Pseudomonas aeruginosa, with reduced sensitivity to antibiotics. Aspergillosis in the most frequent infectious cause of massive pulmonary hemorrhage in pediatric leukemias, while bacterial pneumonia or TBC are by far less frequent. Aspergillus requires the presence of neutrophils to cause hemoptysis or cavitation, while bacteria (Gram-positives or negatives) can cause pulmonary necrosis also in the presence of neutropenia.

###

NEONATAL HYPOXIC - ISCHEMIC HEART FAILURE

M. Capozza, D. Martinelli, N. Laforgia

Neonatologia e Terapia Intensiva Neonatale, Dimo, Università degli Studi di Bari

Heart failure is a clinical syndrome in which the heart is unable to guarantee the needs of the organism. The newborn is very susceptible to this because in new born babies the factors regulating heart function are less efficient. We report the case of Anna, born at term by urgent C-section because of severe decelerations in CTG and IUGR and subsequently subjected to CPR and intubation. She presented with severe anemia (Hb 2.3 g/dl) because of chronic fetal-maternal transfusion and received urgent transfusion of RBC. A dose of Surfactant was administered. She underwent HFO for 10 days and conventional ventilation for 7 days, then high flows for 3 days. She received Dopamine, Dobutamine and Milrinone for clinical and echocardiographic signs of myocardial dysfunction (arterial hypotension, iposfigmia of arterial pulses, pallor, reduced ventricular contractility, pulmonary hypertension) for 6 days which by inhibiting phosphodiesterase causes increase of cAMP improving cardiac contractility and vasodilation. Peritoneal dialysis was performed for 4 days to treat acute renal failure. Brain ultrasound showed hyperechogenicity and cerebral edema. Phenobarbital was started for generalized tonic seizures and a discontinuous and hypovolted pattern of EEG. 20 days later, the brain MRI showed HIE. Anna was discharged at one month of life. Renal and respiratoy function are normal. She continues therapy with oral Luminale. Last cardiac ultrasound revealed hypertrophy of septum and of posterior wall of left ventricle. Clinical follow-up and a new brain MRI would be useful to study the neurological outcome.

###

PARTIAL STATUS EPILEPTICUS AS ONSET OF A BENIGN CHILDHOOD EPILEPSY WITH CENTRO-TEMPORAL SPIKES (BCECTS)

Laccetta G. 1, Orsini A. 1, Del Pistoia M. 1, De Tata R. 1, Bertelloni S. 1, Cesaretti G. 1, Bonuccelli Al. 1

1Scuola di specializzazione in Pediatria, Università di Pisa – Azienda Ospedaliera Universitaria Pisana

T. was born at 29 weeks of gestation because of placental venous thrombosis; at birth he was small for gestational age. The child presented a normal psychomotor development; at 4 years of age he started growth hormone therapy for impaired linear growth, according to Italian medical laws. Family history was nega-
tive for neurologic diseases and related conditions.
At 5 years of age T. presented an episode of sialorrhea, dysarthria and left deviation of his mouth with partial loss of consciousness. About 15 minutes after the onset of symptoms, the child was at the Emergency Department showing left deviation of the mouth and orbicular oris muscle flaccid paralysis. General physical examination was normal.
On admission to our Pediatric Department T. was totally asymptomatic; therefore, a partial status epilepticus was suspected. Laboratory results, electrocardiogram and echocardiogram were normal. Electroencephalograms performed while in awake and sleep showed a focal epileptiform activity compatible with Benign Childhood Epilepsy with Centro- Temporal Spikes (BCECTS). Brain magnetic resonance showed no abnormalities.
Six days after discharge from hospital, T. had an episode of partial seizure with secondary generalization during sleep; this episode was interrupted by the administration of rectal diazepam. After the occurrence of the second epileptic episode valproic acid therapy was started achieving control of the seizures; growth hormone therapy was not discontinued.

####

**HYPERTROPHIC CARDIOMYOPATHY IN CHILDHOOD: A CASE REPORT**

C. Lo Verso¹, V. Duca², B. Giuffrè², R. Conti².

¹Dipartimento di Scienze per la Promozione della Salute e Materno Infantile "Giuseppe D’Alessandro", Università degli Studi di Palermo.

Hypertrophic cardiomyopathy is a heart muscle disease with increased heart wall thickness, in the absence of systemic/cardiac causes. Prevalence of 1:500-1000 in the general population. It was the first cardiomyopathy to be identified with a specific genetic etiology: it is autosomal dominant. The frequently involved genes are: MYH7; MYBPC3; TNNT2. Epigenetics also affects gene expression with important changes in clinical condition. We describe the case of a male patient aged 8 months who came to our observation due to lack of growth and difficulty eating. His father and uncle suffer from Hypertrophic Cardiomyopathy, with ICD from the age of 30 years. On examination the patient had a systolic expulsion murmur. ECG showed abnormal ST-T. Echocardiography showed interventricular septum hypertrophy, reduced cardiac relaxation and through Doppler we detected ventricular outflow obstruction with pressure gradient (70 mmHg). In children, the diagnosis of HCM requires an LV wall thickness more than two standard deviations greater than the predicted mean. However, DNA analysis gives diagnostic certainty (60% cases) and makes "family genetic screening" possible. Our patient began treatment with propranolol, and the ultrasound follow-up revealed an improvement in cardiac function. The risk of mortality is greater for children than for the adult population. Genetic diagnosis of this disease allows therapeutic intervention before symptoms manifest seriously, preventing disease progression.

####

**A CASE OF HYPOGLOSSAL NERVE PALSY CAUSED BY TEETH DEVICE**

T. Timpanaro; MC. Balistreri; S.D. Marino; L. Schiavone; P. Smilari; F. Greco; G. Sorge

Pediatric Department, AOU Policlinico-Vittorio Emanuele, Catania, Italy

We describe the case of a 12 year-old female, admitted to our hospital for a tongue deviation toward the right side and fasciculation observed during protrusion. The patient complained of difficulty swallowing (dysphagia) and demonstrated slurred speech (dysarthria). She had used a dental device for the previous two years, which had been removed for pain. Moreover, she had received logopedic treatment for 3 months, obtaining
partial benefit. The patient underwent electromyographic and imaging studies. EMG-VCN of the genioglossus muscle and of the Hypoglossal nerve (CN XII) showed axonal motor neuropathy of the left CN XII. Brain magnetic resonance imaging (MRI) revealed asymmetry of the vertebral arteries, the course of left hypoglossal nerve was regular. Angio MRI showed a small reduction both in the volume and the enhancement after contrast at the half left of the tongue, without neurovascular conflict. We diagnosed axonal motor neuropathy of left CN XII caused by trauma (dental device). Hypoglossal nerve injuries are rare and the most common causes include: malignancy, trauma, occipital-cervical junction fracture, iatrogenic causes, syringomyelia, cranial nerve injury and infection. The prognosis is good, and most isolated unilateral CN XII palsies resolve within 6 months.

###

**GAIT INSTABILITY, TENDON HYPERREFLEXIA AND DYSARTHRIA IN A CHILD. A CASE OF EARLY ONSET OF SPINOCEREBELLAR ATAXIA TYPE 8 (SCA8)**

M. Grisolia 1, A. Nicoletti 1, F. Falvo 1, E. Pascale 1, I. Mascaro 1, F. Ceravolo 1, D. Concolino 1

1 Department of Pediatrics, University Magna Graecia, Catanzaro, Italy

Introduction: SCA8 is a slowly progressing ataxia with disease onset typically occurring in adulthood. Common initial symptoms are scanning dysarthria, gait instability and tendon reflex hyperreflexity. SCA8 phenotype is caused by an expansion mutation involving two overlapping genes, ATXN8OS and ATXN8. We report the case of an 11 year old girl with ATXN8OS mutation, evaluated for gait instability and dysarthria. Case report: the proband was the first child of healthy unrelated parents. Pregnancy, delivery and perinatal event was uneventful. At the age of 3 she started to show developmental delay and gait impairment. Clinical examination at 11 showed ataxia, tendon hyperreflexia, positive finger to nose and Romberg’s tests, dysarthria, dysphagia for solid foods and mild cognitive impairment. Brain imaging showed cerebellar vermis hypoplasia. Laboratory tests excluded inherited metabolic disorders. Molecular analysis of spinocerebellar ataxias (SCA) has found an expansion of triplets on ATXN8OS gene, responsible for SCA8. Conclusions: gait impairment in children often remains a medical challenge causing delays in diagnosis of hereditary neurological disorders. Early diagnosis can prevent some complications. SCA8 is transmitted in AD manner with low penetrance, so accurate genetic counseling is required. All affected individuals have one parent with an ATXN8OS expanded allele. Therefore, it is important to test the family as the disorder can also appear clinically in adulthood.

###

**TRAUMATIC DIAPHRAGMATIC ROPURE: A RARE CASE OF RESPIRATORY DISTRESS**


1 Università di Udine, Clinica Pediatrica, Azienda Ospedaliero-Universitaria di Udine
2 Dipartimento di Scienze della Salute, Università degli Studi di Firenze, Azienda Ospedaliero Universitaria Meyer Firenze
3 I Clinica Pediatrica, Seconda Università degli Studi di Napoli
4 Dipartimento di Emergenza Accettazione, Trauma Center, Azienda Ospedaliero Universitaria Meyer Firenze.
5 Dipartimento di Emergenza Accettazione, Azienda Ospedaliero Universitaria Meyer Firenze.

A 6-year-old boy was brought by the Emergency Medical Service to the Emergency Department of our Children’s Hospital with abdominal pain after being involved in a high speed motor vehicle crash in which he was a restrained passenger. Primary survey revealed normal breath sounds throughout the thorax. A
A 22-month-old boy was brought to our Emergency Department for colicky abdominal pain, icy cold sweating and fever. The symptoms had started 24 hours earlier. Good general conditions, pallor, normal breath sounds and cough. The abdominal ultrasound (US) showed the presence of adenomesenteritis. The child was discharged after the relief of pain, but returned after 36 hours for abdominal pain, sweating, vomiting, poor feeding and reduced diuresis. The clinical examination revealed a CRT of 3”, shallow breathing with prolonged expiratory phase. Blood tests were normal. The abdominal US was unchanged. He was admitted for observation. US was repeated 3 days later for the persistence of the symptoms and reduction of breath sounds. US bilateral pleural fluid and the chest X-ray showed cardiomegaly and pulmonary vascular congestion. He was transfered to the intensive care unit due to heart failure caused by myocarditis. Myocarditis is an inflammatory disease of the heart, usually caused by an infection, rare and potentially deadly. Diagnosis is difficult because of the variety of non-specific symptoms at the onset. It is the most common extra abdominal cause of abdominal pain in children <2years. There are no non-invasive tests for diagnostic certainty, it is therefore necessary to integrate clinical, laboratory and instrumental examinations (ECG, echocardiogram, chest X-ray, MRI heart).

###

AN UNCOMMON CASE OF ABDOMINAL PAIN IN THE EMERGENCY DEPARTMENT

M. Giacalone1, S. Montano1, S. Salvadei1, N. Parri2, S. Masi3.

1 Dipartimento di Scienze della Salute, Università degli Studi di Firenze, Azienda Ospedaliero Universitaria Meyer Firenze
2 Dipartimento di Emergenza Accettazione, Trauma Center, Azienda Ospedaliero Universitaria Meyer Firenze.
3 Dipartimento di Emergenza Accettazione, Azienda Ospedaliero Universitaria Meyer Firenze.

A 22-month-old boy was noticed. Initial blood pressure was 100/55, heart rate 125 min. The Glasgow Coma Scale was 15. A total body CT scan demonstrated a liver laceration (grade IV AAST classification) and a raised right hemi-diaphragm with ipsilateral reduction of the lung volume and pleural effusion. No other injuries were seen on the CT scan. Patient was admitted to the pediatric intensive care unit where he remained stable with spontaneous breathing. 3 days later, a CXR was obtained because of respiratory distress and worsening clinical conditions. CXR revealed a complete opacification of the right hemithorax and an elevated right emidiaphragm. A CT scan of chest and abdomen showed a right diaphragmatic herniation and a right pleural effusion with a mediastinal shift. On diagnostic thoracocentesis, haemorrhagic fluid was drained confirming the diagnosis of right-sided hemithorax. Exploratory thoracotomy performed with a right subcostal incision (VI-VII rib) revealed a traumatic rupture of the right diaphragm with liver herniation into the chest. The diaphragmatic injury was repaired. Post-operatively, the patient did well.

###

A CASE OF HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS INVOLVING THE CENTRAL NERVOUS SYSTEM

C.Coppari1, M.Rossi1, S.Bacelli1, G.Del Baldo1, C.Cionna1, S.Gobbi1, P.Pierani1, S.Pizzi2.

1Department of Paediatrics Oncohematology, Università Politecnica delle Marche, Ancona, Italy
2 Pediatric Intensive Care Department, Ospedali Riuniti, Ancona, Italy

A 3 year-old girl, hospitalized for systemic EBV infection, presented a seizure unresponsive to pharmacological treatment and severe respiratory distress, requiring admission to the intensive care unit. History revealed she had suffered from necrotizing pneumonia, treated with antibiotic therapy and VATS one month
before. On admission she was critically ill, non-convulsive and with important hepatosplenomegaly. Analysis of CSF showed pleocytosis and elevated CSF proteins, EEG revealed widespread suffering and brain MRI was normal. Blood examination revealed normocytic anemia, thrombocytopenia, mild neutropenia, hypertriglyceridemia, increased transaminases, hyperbilirubinemia, hypofibrinogenemia, elevated serum ferritin level and soluble IL-2 receptor. Analysis of the bone marrow demonstrated hemophagocytosis. The genetic investigation did not show pathogenetic mutations of familiar erythrophagocytic lymphohistiocytosis (HLH)-related genes. EBV infection was treated with rituximab because of the persistence of high viremia. On the basis of Euro-Hit diagnostic criteria for HLH the diagnosis was established, and she started a specific therapy according to the Euro-Hit-HLH protocol. Induction therapy and continuation therapy were performed until she underwent hematopoietic stem cell transplantation because of the evidence of disease reactivation.

####
A CASE OF AN ASYMPTOMATIC INTRACARDIAC MASS ASSOCIATED WITH A RARE MALIGNANT TUMOR.

Cionna C\textsuperscript{1}, Bacelli S\textsuperscript{1}, Caterina C\textsuperscript{1}, Ferrito L\textsuperscript{1}, Gobbi S\textsuperscript{1}, Pierani P\textsuperscript{1}, Pozzi M\textsuperscript{2}, Merlino E\textsuperscript{2}

\textsuperscript{1}Department of Paediatrics Oncohematology, Università Politecnica delle Marche, Ancona, Italy
\textsuperscript{2}Heart Pediatric Surgery Department, Ospedali Riuniti, Ancona, Italy

A 7-year-old girl presented at our hospital with an incidental finding at echocardiography control of a cylindrical floating mass in the right atrium and in the right ventricle near the pulmonary infundibulum (4mmx3cm), starting from the inferior vena cava. She did not present any pathological findings on physical examination, except for a II/VI diastolic rumbling murmur. Serological tests revealed the following: LDH 507 UI/l, NSE 86 UI/l. A contrast-enhanced CT scan showed: large right suprarenal mass (5x4.5cm) involving the right renal artery, the superior mesenteric artery, the right renal vein, the left renal vein and inferior vena cava, determining a filling defect and a thrombotic lesion extending up to the right atrium and right ventricle. After clinical and instrumental evaluation, cardiovascular surgical treatment was suggested. The surgery was performed by the cardiovascular surgical team, and revealed the mass in the right atrium, right ventricle, and inferior vena cava, during deep hypothermic circulatory arrest. On histological examination, the lesion presented clear characteristics of adrenal cortical carcinoma. Biochemically, plasma catecholamines and adrenal cortex hormones were normal, showing that it was a non-functioning tumor. An angio-CT confirmed that the right suprarenal mass was currently unresectable and a bone scintigraphy showed that there were not any bone metastases. In order to reduce the size of the mass and to make the tumor completely resectable with surgery, the patient started a specific therapy with Mitotane plus EDP (Etoposide, doxorubicin, cisplatin). This therapeutic approach is currently ongoing.

####
STATUS EPILEPTICUS IN A CHILD WITH SUSPECTED CROUZON SYNDROME

G.Romano1, M.Scavone1, E.Stefanelli1, E.Carboni1, V.Talarico1, I.Mascaro1, M.Barreca2, MC Galati3, G.Raiola2

1. U.O. di Pediatria Universitaria, Università “Magna Graecia” di Catanzaro
2. U.O. di Pediatria, Azienda ospedaliera “Pugliese-Ciaccio” di Catanzaro
3. U.O. di Oncologia pediatrica, Azienda ospedaliera “Pugliese-Ciaccio” di Catanzaro

An 8 year old child was referred to our unit for repeated, unremitting epileptic seizures with dyspnoea. Medical history: obstructive sleep apnea. Physical examination: mental retardation, exophthalmos, strabismus, hypertelorism, deviated
nasal septum, brachycephaly, protruded lower jaw, narrow arched palate, crowding of teeth. He was immediately treated with iv Midazolam and ventilatory support. Rx of the skull: hammered-silver appearance with an enlarged sella turcica and lack of fusion of the sutures. Brain MR: ectasia of the ventricular system, bilateral exophthalmos, reduction of anteroposterior diameters. Ocular examination: divergent strabismus and marked proptosis with bilateral optic subatrophy. EEG: epileptiform electrical cerebral activity; therefore, he was treated with Carbamazepine. Combining clinical and instrumental data, we suspected Crouzon’s syndrome (CS). The FGFR-2 gene analysis was requested.

He was discharged home on anticonvulsant therapy and the craniotomy was scheduled. Discussion: CS, an autosomal dominant disorder, is the most frequently occurring member of craniofaciosynostosis, with variable expressivity. The premature fusion of sutures exerts pressure on the skull as well as on the developing brain. In these children, the clinical manifestations of abnormally increased intracranial pressure (ICP) are difficult to detect. Early detection of ICP and subsequent treatment is important in order to reduce the risks for brain development and visual function.

BRODIE’S ABSCCESS: HOW, WHEN AND WHY

V. Dolcemascolo1, C. Santelia2, S. Celestino2, D. Driul3, E. Passone2, G. Cricchiutti3

1 Scuola di Specializzazione in Pediatría, Trieste
2 Scuola di Specializzazione in Pediatría, Udine
3 Clinica Pediatrica, Azienda Ospedaliero-Universitaria SSMM, DPMSC, Udine

An 18 month old child presented with a 1 month history of mild limping, swelling of the right ankle and progressive load rejection unresponsive to ibuprofen. Blood samples showed SER 41 mm/h and negative blood culture. X-ray disclosed a tibio-distal rarefaction and MR an area with a sclerotic border from the tibio-distal epiphyseal ossification nucleus to metaphyseal area, compatible with Brodie’s abscess. Curettage was not possible so intravenous therapy was started with cephoxaxim and clindamycin and continued for 4 weeks followed by a month of oral amoxicillin and rifamicyne with subsequent recovery. Brodie’s abscess is a subacute osteomyelitis, more frequent in children, that usually affects tibia, femur and clavicle. In infants younger than 18 months it can involve epiphysis and metaphysis due to a vascular communication present until this age. The causative organism is usually CoA-positive Staphylococcus but in 50% of cases no organism can be cultured. If pain, minimal loss of function and limping are the most consistent complaints, symptoms can often be aspecific. White blood cell count, SER, CRP are usually normal or slightly elevated. MR is the gold standard for the diagnosis showing a well demarcated hyperintense lesion with or without sequestrum area surrounding sclerosis. Differential diagnosis includes malignant diseases such as Langerhans cell histiocytosis, Ewing’s sarcoma or lymphoma. Brodie’s abscess treatment is prolonged antibiotic therapy based on antibiograms, but lesion curettage is also described.

TRIORCHIDISM: A RARE GENITOURINARY ABNORMALITY.

Papia B.1, Castiglione M.C.1, Insinga V.1, Zambaiti E.2, Cimador M.2, Corsello G.1

1 Dip. di Scienze per la Promozione della Salute Materno Infantile "G. D’Alessandro”, Univ. degli Studi di Palermo-Direttore prof. G. Corsello
2U.O.C. Chirurgia Pediatrica

Introduction: Polyorchidism refers to the presence of more than two testicles. Polyorchidism is usually discovered incidentally. The most common anomalies associated are inguinal hernia (30%), maldescended testis (15% to 30%), tes-
ticular torsion (13%) and hydrocele (9%).

Case report: L.F.P., 2 years, with neonatal diagnosis of monolateral undescended testes (right) with normal milestones. Local examination revealed normal male external genitalia and scrotum, penis shaft with synechiae. Left testis was palpable in the scrotum, normal for texture and volume. Right testis was absent in the scrotum, palpable in the inguinal canal. The surgery consists of transverse incision of the right inguinal canal, with insulation, ligature and section of peritoneum-vaginalis duct. During the downward traction a second deferens vas was revealed, that ended in a second atrophic testis.

Conclusion: Laung described the first anatomical classification. In this case, we see a type IV, with complete duplication of testes, epididymis and vas. Management of polyorchidism has been subject of much debate, in particular when it is found incidentally. The orchidectomy of the supernumerary testes is advocated only when it is atrophic, without connection to the vas deferens. On the basis of the available literature, it is appropriate to preserve the supernumerary testis by performing scrotal orchidopexy.

###

HAEMOLYTIC URAEMIC SYNDROME AS A COMPLICATION OF INDUCTION CHEMOTHERAPY FOR ACUTE LYMPHOBLASTIC LEUKEMIA

Olivieri I.°, Micalizzi C.°

*Clinical and Experimental Haematology Unit, Istituto Giannina Gaslini, Genova*

Case report: A four year old boy with ALL, treated with ALL R 2006 protocol. At day 33 of IA induction (including dexamethasone, 8-L-asparaginase and 4 vincristina+daunoblastina) he develops haemolytic anaemia, thrombocytopenia, gross haematuria and finally acute renal failure. The urine culture is positive for E.Coli, so the diagnosis of haemolytic uraemic syndrome (HUS) is made. He immediately stops the chemotherapy and starts with haemodialysis, plasma exchanges and transfusions. After 20 days, he is completely dialysis and transfusion independent with a normal serum creatinine and a normal urine output. 25 days after discharge, his original chemotherapy regimen is re-instituted with no ill effects.

**Discussion:** The existence of an association between ALL and HUS is reported, and mortality in the acute phase of the illness is 3-5%, so it is very important to consider HUS in the differential diagnosis in all patients with ALL who develop haemolytic anaemia, thrombocytopenia and/or acute renal failure.

####

A FEVER THAT SMELLS

C. Stocco¹, V. Dolcemascolo¹, E. Miorin², G. Crichiutti²

¹Scuola di Specializzazione in Pediatria di Trieste; ²Clinica Pediatrica – Ospedale S. Maria della Misericordia di Udine

A 7 year old boy presented with a two week history of chest pain, cough, weight loss and fever unresponsive to azithromycin and amoxicillin-clavulanate. He had returned from a trip to India six months before. Blood tests showed WBC 14590/mmc (N 8910/mmc, L 4610/mmc), CRP 85 mg/L, SER 73 mm/h and negative culture. Chest X-ray disclosed two upper lobe cavitary bilateral lesions, one of which with direct connection to a segmentary bronchus later defined by TC. Tuberculin skin test (TST) was negative but quantiferon resulted positive. Smear microscopy and PCR resulted negative for M. tuberculosis (MBT) on blood, gastric aspirates, urine and bronchoalveolar lavage. Cultures for MBT are still in progress. Antitubercular therapy with isoniazid, rifampin and pyrazinamide was started with rapid clinical improvement. India has the highest number of tuberculosis (TB) cases in the world, with a rise in multidrug-resistant disease in the last decade. TB in children is often
diagnosed clinically, as up to 40% of immunocompetent children with culture-confirmed TB may have negative TST, and most of them are sputum-negative for acid-fast bacilli. Interferon-γ release assays have greater specificity than TST and radiological findings, including lymphadenopathy, cavitation and communication with bronchial structures can suggest the diagnosis. When laboratory confirmation is not established a presumptive diagnosis may be made based on clinical and radiographic response to empiric treatment.

###

**PSYCHOMOTOR AGITATION, EPILEPTIC SEIZURES AND MEMORY IMPAIRMENT: A CASE OF LIMBIC ENCEPHALITIS**

F. Vendemini, A. Aldrovandi, A. Rizzello, DM. Cordelli, E. Franzoni, A. Pession

1 Scuola di Specializzazione in Pediatria, Università di Bologna
2 UO Neuropsichiatria Infantile – Policlinico Sant’Orsola Malpighi
3 Direttore Scuola di Specializzazione in Pediatria, Clinica Pediatrica Università di Bologna – Policlinico Sant’Orsola Malpighi

A 16 year-old boy was admitted to the adult Emergency Department for a first episode of generalized seizure. During observation, secondary generalized seizures relapsed and were treated with diazepam. EEG and brain CT were normal and levetiracetam therapy was started.

The boy was admitted to our Unit two days after symptom onset. Due to persistence of seizures, intravenous (IV) midazolam was added with subsequent reduction of seizure frequency. However, the boy began to show behavioral changes, notably agitation, anxiety, aggressivity toward parents and staff along with short-term memory impairment and episodes of confusion. MRI brain imaging showed bilateral low hyperintense FLAIR hippocampal and amygdala signals. Cerebrospinal fluid evaluation was normal. Suspecting limbic encephalitis (LE), IV methylprednisolone (subsequently replaced with oral prednisone) and immunoglobulin were started, while olanzapine and lorazepam were administrated to rapidly control psychiatric symptoms. Infections, metabolic and connective tissue disorders were excluded by laboratory analyses. Antibodies to neuronal and synaptic antigens were negative and concomitant neoplasia was ruled out. Seizures receded while psychomotor agitation lasted for a further three days and then gradually disappeared; amnesia with regard to the hospitalization period persisted.

This case suggests considering LE as a differential diagnosis in patients presenting with psychomotor agitation along with seizures and memory defects.

###

**HERPES SIMPLEX VIRUS TYPE 1 (HSV1) ENCEPHALITIS IN AN IMMUNOCOMPETENT CHILD WHO PRESENTED SUDDEN DYSPHASIA**

Tonelli L, Marabini C, Palmas G

Clinica Pediatria Ospedale Salesi Ancona, Università Politecnica delle Marche, Italia

An immunocompetent 5-year-old girl was referred for dysphasia, fluctuation of consciousness and a seizure resolved by rectal diazepam. Laboratory tests resulted negative for inflammatory markers. Electroencephalography and brain tomography were unremarkable, while lumbar puncture (LP) revealed cell count of 292 in the cerebral spinal fluid (CSF). CSF polymerase chain reaction (PCR) was positive for HSV1 therefore highdose acyclovir intravenous treatment (10 mg/kg every 8hours for 21days) was started. Magnetic resonance (MR) detected whole thalamic, frontal and temporal lobe inflammatory feature. In the following 24hours, a deterioration in neurological status appeared, marked by stupor, pain, weak response to verbal stimulation and blurred vision. A new MR showed diffuse cerebral edema, consequently, a mannitol and furosemide treatment was
started. After 4 days, MR reported gradual brain edema resolution and improved consciousness was detected. Subsequently, she no longer presented seizures or other encephalitis signs. Dysphasia, disorientation and speech disturbances (difficulty in remembering common words) were still present, combined with behavioural changes such as irritability and opposition. After 21 days antiviral therapy, CSF-PCR for HSV1 resulted negative and RM re-evaluation showed a left hemispheric fronto-temporo-parietal wide signal impairment and ipsilateral capsule-thalamic hemorrhagic findings. She was discharged with antiepileptic therapy and a clinical-instrumental follow-up was started.

###

PRENATAL DIAGNOSIS OF CONGENITAL PORTO-SYSTEMIC SHUNT: A CASE REPORT

Giulia Paolella, Marcello Farallo, Irene Degrassi, Silvia Bettocchi, Gabriella Nebbia

Servizio di Epatologia e Nutrizione Pediatrica, Fondazione IRCCS Ca’ Granda Ospedale Maggiore Policlinico - Milano

Background: Congenital porto-systemic shunts are rare vascular malformations classified as either intra or extra-hepatic shunts; extra-hepatic shunts are also divided into extra-hepatic shunt type 1 (Abernethy malformation) characterized by absence of portal vein and extra-hepatic shunt type 2 with hypoplastic intrahepatic portal vein.

Case report: F. was born at the 39W of gestation with APGAR score 9-10. Fetal ultrasound showed agenesis of ductus venosus, intrahepatic calcifications, hyperechoic bowel, and dilated vena cava. Fetal MRI confirmed the absence of fetal ductus venosus with dilated umbilical vein draining in inferior vena cava. Amniocentesis was normal. Newborn metabolic screening was also normal. On the second day of life, echocardiogram showed a small patent ductus arteriosus and patent foramen ovale. Abdomen radiography confirmed liver calcifications. Abdominal ultrasound showed two hyperechoic lesions in the right hepatic lobe near the hepatic veins; furthermore, a semilunar shaped calcified lesion, with posterior shadow cone situated in proximity of Aranzio duct was also detected. Abdominal CT angiography showed hepatic calcifications, and the portal vein draining to the inferior vena cava. An ectatic extension at the level of the vena cava fueled by mesenteric venous vessel was also detected. The patient was affected by Abernethy malformations. He will be eligible for fistula closure after portal system permeability evaluation.

###

.. AND ONE DAY JAUNDICE APPEARED

Monica Malamisura¹, Paola De Angelis², Filippo Torroni², Erminia Romeo², Francesca Rea², Simona Faraci², Giulia Angelino¹, Jolanda Pianese¹, Giusy Ferro¹, Stefania Bernardi¹ e Luigi Dall’Oglio²

1 University Department of Pediatrics, Unit of Immune and Infectious Diseases, Children’s Hospital Bambino Gesù, Rome, Italy
2 Digestive Surgery and Endoscopy Unit, Children’s Hospital Bambino Gesù, Rome, Italy

A 12-year-old boy was brought to the emergency department of another hospital for jaundice, abdominal pain and vomiting. His clinical history was silent, his mother died of meningitis infection as a result of AIDS, when he was three years old. On arrival, he rated the pain at 8 on a scale of 0 to 10 (with 10 indicating the most severe pain). At clinical examination, the abdomen was soft with moderate tenderness in the epigastrium and right upper quadrant with positive Murphy’s sign, the skin was jaundiced, the remainder of the examination was normal. The blood tests resulted in high bilirubin and pancreatic enzymes. He was diagnosed with acute pancreatitis and started on specific therapy (intravenous hydration, fasting, antibiotics, UDCA and octreotide). At cholangiopancreatography MRI, pancreatic head appeared increased...
in volume and alteration in inhomogeneous parenchymal signal evident. Ectasia of Wirsung all along its course (up to 4 mm caliber), ectasia of the biliary intrahepatic extrahepatic and ectasia biliary main (16-17 mm). The patient was tested for HIV infection, considering his clinical history, unfortunately he tested positive and started antiretroviral therapy (at the start the CD4 were 90 UI/μL, 8% of lymphocytes with 19000 copies of virus). The patient was then transferred to our hospital for surgical evaluation. On arrival his clinical conditions were stable, total bilirubin was 13,6 mg/dl, (direct 10,84 mg/dl) with GGT 451 UI/L and rise in pancreatic enzymes, lipase 241 UI/L and amylase 137 UI/L. One day after admission an ERCP was performed, coleducic duct was hardly sondable and not dilatable. Computered Tomography (CT) showed newly oval formation, uneven between the liver and the pancreas (DL 4 cm transverse 3 cm sagittal 2,6 cm) and biliar ducts dilated. Abdominal ultrasonography, complicated cystic formation suggested the presence of a duodenal duplication cyst. Tumoral markers were negative. Five days after admission, outer transhepatic biliary drainage was put in place via percutaneous transhepatic cholangiography, of biliary fluid sample was collected to test for cryptosporidium, cryptococcal and mycobacteria, which were absent. Ten days after the admission a joint procedure was performed with surgeons and interventional radiologists with the placement of an internal biliary drainage via sphincterotomy and rendezvous between the internal and external biliary drainage. On CT examination, performed nine days after the procedure, the bile ducts were not dilated and the newly formed oval was no longer viewable. Twenty days after admission the external drain was removed, and the patient was discharged in good clinical conditions, bilirubin was 1,64 mg/dl, GGT 43 UI/l. At follow-up, one month later, no signs of cholestasis and cytolysis, the internal stent was replaced, the ERCP showed a formation jutting into the lumen of the second portion of the duodenum at the injection of the contrast, evidenced by dye gathering round the wall of the duodenum suggesting duplicating or spreading submucosa. At biopsy biliary tract was normal. New follow-up is ongoing. The patient responded dramatically to antiretroviral therapy, CD4 12,5%, viral load was absent. Gastrointestinal tract duplications are rare congenital anomalies, only a small percentage have been described in the duodenum, they are usually diagnosed in the adult population.

####

HYPERAMMONEMIA OR NOT HYPERAMMONEMIA: THAT IS THE QUESTION?

A CASE OF CONGENITAL NEPHROTIC SYNDROME MISDIAGNOSED BY A NEONATAL HYPERAMMONEMIA

C. Spagnoli, R. Annibali, C. Pannunzi Proietti

Clinica Pediatrica Ospedale Salesi Ancona

A term male infant was referred to our NICU for hyperammonemia, 3 days after birth. The family history was silent and the course of pregnancy regular. The physical examination revealed marked dysmorphic features, pale skin, poor sucking and mild feet edema. First level exams showed marked lipemic serum; liver and kidney function were normal. Hyperammoniemia was not confirmed. The subsequent laboratory tests revealed hyper-triglyceridemia (TG), hypercholesterolemia (COL), hypoalbuminemia and severe proteinuria. Five days after birth, he started fasting for 24h, with significant reduction of TG and COL levels and than he started Monogen formula. He needed repeated albumin infusions, but his general conditions remained stable. His clinical features and his laboratory tests appeared suggestive for congenital nephrotic syndrome. Seven days after birth, we started a diuretic treatment with furosemide, ramipril/losartane and idroclorotiazide/spironolattone. His pressure levels remained under normal values and his proteinuria decreased. He continued to need daily albumin infusions, but his general conditions remained stable. His clinical features and his laboratory tests appeared suggestive for congenital nephrotic syndrome. Seven days after birth, we started a diuretic treatment with furosemide, ramipril/losartane and idroclorotiazide/spironolattone. His pressure levels remained under normal values and his proteinuria decreased. He continued to need daily albumin infusions. Twelve days after birth, he developed leukocytopenia, while hemoglobin level and platelet count remained normal.
When he reached 5 kg of weight, a renal biopsy was performed and it showed a focal segmental glomerulosclerosis. The analysis of the gene SMARCAL1 confirmed the diagnosis of immune bone dysplasia Schimke type.

###

**SIROLIMUS FOR DIFFUSE NEONATAL LYMPHANGIOMATOSIS: A CASE REPORT**

Mongelli G., Lieggi S., Varano S., Piccarreta P., Santoiemma V., Laforgia N.

Neonatology and NICU, University of Bari, Italy.

Vascular anomalies represent a heterogeneous group of benign disorders, but some of these lesions can impair vital structures, deforming, or even becoming life-threatening.

We report a case of a diffuse lymphangiomatosis in a term newborn, for which a prenatal diagnosis of laterocervical lymphatic malformation was made. During the first days of life, a significant increase in volume of the laterocervical lymphatic malformation was seen together with respiratory distress that required intubation and mechanical ventilation. Total body MRI showed diffuse lymphangiomatosis with other lesions involving submandibular space, glottis, mediastinal space and abdomen.

The mTOR inhibitor Sirolimus treatment we had already used in another case resulted in significant reduction of all lesions without side effects. Mammalian target of rapamycin (mTOR) is a serine/threonine kinase regulated by phosphoinositide-3-kinase. mTOR acts as a master switch for numerous cellular processes, including cellular catabolism and anabolism, cell motility, angiogenesis, and cell growth.

At the moment, our patient is four months old and she is well. Neither surgical nor sclerosing therapy has been performed.

###

**NON-CONVULSIVE STATUS EPILEPTICUS IN A CHILD WITH PANHYPOPITUITARISM AND RATHKE CLEFT CYST**

I. Parente1, E. Sciorio1, N. Improda2, G. Montefusco1, C. Gagliardo1, M. Salerno2, A. Romano1

1 Settore di Neuropsichiatria Infantile, A.O.U. Federico II di Napoli
2 Settore di Endocrinologia Pediatrica, A.O.U. Federico II di Napoli

Introduction: Rathke cleft cysts (RCC) are rare benign embryonic remnants of the Rathke's pouch. In children, most RCCs are asymptomatic. When symptomatic, they can cause headache, visual disturbance, or pituitary hormone deficits. Hypothalamic-pituitary lesions may be associated with forms of epilepsy. Epilepsy has not been reported in association with cysts. B is a boy followed at our Department from the age of 5 years for RCC. Since the age of 7 years, he has shown hypopituitarism requiring replacement therapy with hydrocortisone, L-thyroxine and GH. Despite treatment, the mother reported persistence of sporadic morning abatement episodes. Therefore, the dose of hydrocortisone was gradually increased up to 18 mg / kg / day, with partial symptom improvement. At the age of 8 he came to our attention for persisting numbness and confusion. At physical examination, he showed delayed response to stimuli and snappy leg tendon reflexes. The EEG showed generalized spike-polispikes, configuring status epilepticus. Midazolam ev. was administered with the resolution of the status epilepticus. The evaluation of the hormonal profile and electrolytes as well as neurometabolic screening was normal. He started treatment with valproic acid with good symptom control.

Conclusions: convulsive disorders may represent acute complications of endocrinological conditions. However, epilepsy should always be kept in mind when evaluating a child with adrenal insufficiency presenting atypical symptoms or who seems unresponsive to hormone replacement therapy. Although there are no data at present, it is possible that the presence of...
RCC can be pathogenetically involved in the genesis of some forms of epilepsy.

### NEUROLOGIC RED FLAGS IN A PATIENT AT RISK: THE UNEXPECTED RELAPSE

Prezioso G.¹, Marsili. M.¹, Guidone P.¹, Chiarelli F.¹, Onofrillo D.², Sau A.², Pillon M.³, Cecinati V.²

¹: Department of Pediatrics, University of Chieti, Via dei Vestini 5, 66100, Chieti, Italy
²: Pediatric Hematology and Oncology Unit, Department of Hematology, Spirito Santo Hospital, Via Fonte Romana 8, 65124 Pescara, Italy
³: Department of Women's and Children’s Health, Clinic of Pediatric Hemato-Oncology, University of Padua, Padua, Italy

A 16-year-old boy complained of worsening headache with morning vomiting for about 2 weeks and diplopia for 2 days. A bilateral papilledema was detected and the brain CT and MRI scans showed a heterogeneous mass in the left semioval center, with perifocal edema extending to the corpus callosum and compressing the left ventricle. Contrast enhancement also involved the cerebral falx. He had been diagnosed 14 months before with primary mediastinal large B cell lymphoma (PMLBCL), stage IIIb with bulky presentation, extranodal involvement not affecting the CNS and superior vena cava syndrome. He was treated with DA-EPOCH-R and, after 6 cycles, the boy was disease-free. The patient was admitted for intracranial hypertension and antiedemigen therapy with dexamethasone and mannitol was undertaken. Additional lesions were excluded by a PET-CT. The stereotaxic biopsy confirmed the CNS relapse of the lymphoma. Thus the LNH 97 protocol was started and the boy is now at the III course with good clinical tolerance and partial mass reduction. CNS relapse in PMLBCL is infrequent and usually occurs in the first year. Once neurological complications are controlled by supportive treatment, chemotherapy must be restarted. Several risk factors and prophylaxes have been suggested but data are conflicting. Promising results have been reported with the DA-EPOCH-R protocol. However, as showed by our case, CNS relapses cannot be accurately predicted and prevented nowadays.

### AN UNUSUAL CASE OF REFUSAL TO WALK IN A CHILD

Grilli M.¹, Coppari C.¹, Rossi M.¹, Lattanzi B.², Pieroni G.³, Cesaroni E.⁴, Zamponi N.⁴

¹ Department of Pediatrics, Università Politecnica delle Marche, Ancona, Italy
² Department of Pediatrics, Ospedali Riuniti, Ancona, Italy
³ Department of Pediatric Radiology, Ospedali Riuniti, Ancona, Italy
⁴ Department of Pediatric Neuropsychiatry, Ospedali Riuniti, Ancona, Italy

We report a case of a 3-year-old girl admitted to the neuropsychiatric department of our hospital for an acute refusal to walk. About 15 days before, she suffered from a lower limb trauma, followed by weakness and limping. On history, she had a mild neurodevelopmental delay with a very restrictive eating pattern. Upon physical examination, she was very irritable, suffering, and unable to stand, with the hips and knees semiflexed. Gingival swelling and intermittent bleeding were present, despite a normal complete blood count and clotting. Blood examinations revealed only a decrease in the parathyroid hormone, with calcium, phosphorus and D vitamin within the normal range, and a slight increase in inflammatory markers. Brain and medulla MRI excluded a cerebral mass or a medullary compression. Analysis of CSF and ENG were normal. X-rays of the lower limb excluded fractures, but demonstrated irregularities in the right distal femoral metaphysis of uncertain significance. Hip US was normal, however, hip and vertebral radiographs underlined a marked reduction of calcic tone. The whole body MRI showed signaling altera-
tion of all the large joints, because of periarticular effusion. These clinical and radiological findings led to the suspicion of a metabolic bone disease and investigations were carried out for scurvy. Vitamin C level was 0.9 mg/L (normal: 5-15) confirming the diagnosis. Therefore, she started a specific supplementation with a clear improvement in her clinical conditions.

###

**GESTATIONAL WEIGHT GAIN AND FETAL GROWTH IN UNDERWEIGHT WOMEN: A POPULATION-BASED STUDY IN NORTH-EASTERN ITALY**

Luca Bonadies, Francesca Volpe, Lara Giliberti, Alessandro Mazza, Antuan Divisic, Anita Cappellari, Ivana Grbin, Gianluca Straface, Vincenzo Zanardo

Division of Perinatal Medicine, Policlinico Abano Terme, Abano Terme, Italy.

Objective. Maternal underweight is a common occurrence with potential adverse perinatal outcomes. We aimed to investigate the relationship between gestational weight gain (GWG) and fetal growth in underweight women.

Methods. Maternal and neonatal data were collected on the maternity ward of Policlinico Abano Terme (Italy), from January 2014 to June 2015. 792 women were categorized according to pre-pregnancy body mass index (BMI) and guidelines for optimal GWG. Neonatal weight, length and head circumference were also collected.

Results. Pre-pregnancy BMI was by far the strongest predictor of the neonatal fetal growth. Offspring of underweight women were comparable in size at birth to neonates of normal weight women, but they resulted significantly lighter than offspring of both overweight and obese women. Conversely, GWG was higher when pre-pregnancy BMI was lower and within the range recommended by IOM guidelines. In addition, while LBW occurrence was unaffected, underweight women presented almost a 50% reduction in LGA as well as in operative delivery: caesarean section and vacuum extractor use in comparison to normal weight women while this reduction is more enhanced in the comparison to overweight and obese women.

Conclusions. Pre-pregnancy underweight does not impact on birth weight of term neonates if GWG is normal. It can be supposed that efforts to reach optimal GWG could be a leading choice for many women living in industrialized and in low-income countries.

###

**AGENESSION OF THE CORPUS CALLOSUM, LISSENCEPHALY AND CEREBELLAR HYPOPLASIA AT PRENATAL ULTRASOUND: POSSIBLE DIAGNOSIS OF THE SYNDROME OF SMITH-LEMLI-OPITZ.**

M.A. Caiazzo1, C. Gagliardo1, L. Capasso1, R. Pisanti1, M. Rosa1, F. Raimondi2

Dipartimento di Neonatologia e terapia intensiva neonatale, Università di Napoli AOU Federico II

We present the case of a female newborn (39 weeks), small for gestational age, the first born after 4 miscarriages. Fetal ultrasounds up to the 5th month were normal; from the sixth month ultrasounds showed growth retardation, agenesis of the corpus callosum, microcephaly, lissencephaly, hydrocephalus, cerebellar hypoplasia, bone dysplasia. Fetal MRI at 31 weeks confirmed the ultrasound abnormalities including bilateral ventriculomegaly. Screening for chromosomopaty (18 and 21), perinatal infections and molecular analysis for cystic fibrosis were negative. After birth we found some dysmorphic features: hypotelorism, dysmorphic low-set ears, micrognathia, cleft of the soft palate, axial polydactyly of the feet with syndactyly bilaterally and hypotonia. We suspect Smith-Lemli-Opitz syndrome(SLOS) because of clinical features and the low level of cholesterol. This diagnosis was confirmed by measuring plasma sterol. SLOS(1.20000-1.40000) is characterized by multiple congenital defects, mental retardation and behavioral disorders. It is
caused by a defect of cholesterol synthesis resulting from a deficiency of beta-3-hydroxysteroid-delta7-reductase. SLOS may be suspected during prenatal life in the presence of heart disease, growth retardation, ambiguous genitalia, hydrodrops and genitourinary abnormalities. In our case ultrasounds and MRI showed mainly brain abnormalities (corpus callosum agenesis, lissencephaly and cerebellar hypoplasia) that could represent a new association for the suspicion of SLOS in a newborn.

EVALUATION OF BONE DENSITY IN PATIENTS WITH NOONAN SYNDROME

MR Cozzolino, C. Gagliardo, G Minopoli, A Del Puente, A Esposito, I. Parente, D Melis

1 Dipartimento di Scienze Mediche Translazionali, Sezione di Pediatria, Università Federico II Napoli
2 Dipartimento Reumatologia Federico II

Noonan syndrome (NS) is an autosomal dominant disorder characterized by facial dysmorphism, short stature, heart defects and skeletal dysplasia, involving MAPK pathway which regulates cell differentiation and homeostasis of bone tissue. We enrolled 12 patients with NS, 8 males and 4 females (mean age 13.3 years), 6 with PTPN11 mutation, 1 with SHOC2 mutation, 1 with BRAF mutation, two with alteration of SOS1 and 2 with genetic analysis still in progress. We performed axial x-ray densitometry (DEXA) and/or bone ultrasound (BUA) to evaluate mineral bone density. In 5 patients we performed DEXA and BUA, in 4 patients only BUA and for 3 only DEXA. Results: 4/8 patients (50%) with DEXA presented a mineral profile compatible with osteopenia (Zscore <-1), 2 patients (25%) had osteoporosis (Zscore <-2.5), 2 patients (25%) had normal bone mineral density; BUA showed osteopenia in 5 of 9 patients (55%) while the examination was normal in the remaining patients (4/9 equal to 44%). The subjects in which BUA and/or DEXA were normal had mutation in PTPN11, SOS1 and BRAF. Conclusion: to date there are limited studies on the mineralization of bone tissue in patients with NS. The data from our study indicate the presence of osteopenia in 50% of patients. This result suggests the importance of careful monitoring of bone mineral density in patients with NS in order to start a specific therapy if necessary.

OSTEODYSPLASIA, BONE MARROW AND EXOCRINE PANCREATIC FAILURE... A CASE OF SHWACHMAN-DIAMOND SYNDROME

C. Gagliardo, L. Capasso, R. Pisanti, J. Cerullo, MA Caiazzo, MR Pirozzi, F. Raimondi

1 Dipartimento Neonatologia e terapia intensiva neonatale AOU Federico II, Napoli

We report the case of triplets born at 33+5/7. The first two (monochorionic diamniotic) showed intrauterine growth retardation, thoracic hypoplasia, short limbs, osteodysplasia, dysmorphic features. The first born showed also Bronchopulmonary dysplasia, recurrent infections, bone marrow failure (neutropenia, thrombocytopenia, anemia) while the second born showed poor growth, seborrheic dermatitis and signs of exocrine pancreatic insufficiency. The Juene Syndrome, Ellis-van Creveld disease (associated with abnormalities of the chest), cystic fibrosis, Pearson and Blizzard syndrome (associated with exocrine pancreatic insufficiency) were suspected. We also suspected Shwachman-Diamond syndrome (SDS) because of the association of skeletal anomalies, failure of the exocrine pancreas and bone marrow. SDS was confirmed by molecular diagnosis (double heterozygous for 2 mutations causative of disease c.258+2T>C;c.107delT). The diagnosis of SDS (1:76000) is difficult in newborns because some clinical manifestations (bone marrow depression, immunodeficiency, dermatitis) can occur later in child or adulthood. Thoracic hypoplasia at birth is
a rare presentation of SDS and is associated with mortality in the first year of life. Conclusion: the clinical presentation of the SDS is variable; SDS should also be suspected for patients with skeletal abnormalities of the chest; for these you should also plan a follow-up to detect pancreatic insufficiency and bone marrow that can also occur with time.

###

CARDIAC INVOLVEMENT IN PATIENTS WITH MUTATION IN PTPN11 AND NOONAN/NOONAN LIKE SYNDROME

G Minopoli¹, C. Gagliardo¹, MR Cozzolino¹, G. Andria¹, P. Strisciuglio¹, D Melis¹

¹ Dipartimento di Scienze Mediche Traslazionali, Sezione di Pediatria, Università Federico II Napoli

Noonan and Noonan-like syndrome (AD) are diseases characterized by neuro-cardio-facial-cutaneous involvement and psychomotor development. The most frequent heart diseases are pulmonary valvular stenosis and hypertrophic cardiomyopathy. PTPN11 gene is mutated in 50% of patients. We examined 17 patients, 12 males and 5 females (8 months -22 years), with mutation in PTPN11 and clinical phenotype of Noonan and Noonan-like syndrome. All patients underwent cardiology consultation, echocardiogram, electrocardiogram: 9/17 showed pulmonary valve stenosis, 2 patients pulmonary dysplasia, 3/17 alterations of the mitral valve; 4 patients atrial septal defect ostium II, 2 VSD, 5 patent foramen ovale, 18% non-obstructive hypertrophic cardiomyopathy. At electrocardiogram 3/17 (all with abnormalities of pulmonary valve) showed a pathological trace: right bundle branch block, sinus arrhythmia. 23% of patients showed no heart disease. Conclusions: in Noonan and Noonan-like syndrome, frequency of heart disease is 50%-80%. Data from our study indicate that 77% of patients with mutation of PTPN11 have heart disease (morphological and functional). The majority (over 60%) has a valve involvement (stenosis, dysplasia), and 18% have hypertrophic cardiomyopathy. In the literature, mutations of PTPN11 are associated with pulmonary valve stenosis, short stature and mild dysmorphism. The association between the coexistence of conduction defects and pulmonary valve stenosis remains to be clarified.

###

A SEVERE CASE OF NEONATAL HYPOTONIA

R. Panza, A. Resta, A.N. Abbaticchio, D. Capodiferro, N. Resta, L. De Cosmo, N. Laforgia

Unità Operativa Neonatologia e Terapia Intensiva Neonatale, Università Degli Studi Di Bari; D.A.I. Scienze e Chirurgia Pediatrica

“Floppy infant” is a hypotonic newborn who may present respiratory distress at birth, low Apgar score, weak suckling and swallowing. The diagnosis is not easy in the newborn period as it may be related to numerous conditions. An accurate neurological examination, the assessment of Prechtl’s General Movements, the study of spontaneous motility of passive and active muscle tone and of primitive reflexes, may help understand if hypotonia is due to central or peripheral causes. MRI, US, laboratory tests, specific tests such as muscle biopsy, EMG and genetic tests are required to make a diagnosis. We present the case of a newborn, born at 42+4 weeks of GA by vaginal delivery. Admitted to our UTIN because of perinatal asphyxia, she required therapeutic hypothermia and ventilatory support for one month; she died 30 days later. She presented severe generalized hypotonia, no spontaneous movements of the limbs, no appropriate suck-swallow pattern, no deep tendon reflex. The family history was unremarkable, but the mother reported reduced fetal movements and polyhydramnios. Brain MRI was normal, Total Body Rx revealed multiple spontaneous fractures of tibia and femur, EMG showed absence of spontaneous and induced motility and muscle
biopsy revealed non-specific signs of myopathy. Karyotype and genetic test for Prader-Willi were normal. Molecular test for SMA revealed a point mutation of the SMN1 gene that made it non-functional. SMA type I with neonatal onset is also named SMA 0, and always represents a fatal prognosis.

###

**UNUSUAL CAUSE OF HEMOPERITONEUM IN A CHILD - A CASE REPORT**

C.R. Stanca¹, A. Lacatusu¹, C. Olaru², M. Burlea², P. Plamadeala³, D. Mihaila³, S.G. Aprodul, N. Gimiga¹, S. Diaconescu²

¹ „St. Mary” Children’s Emergency Hospital, Gastroenterology Unit, Jassy, Romania,
² „Gr. T. Popa” University of Medicine and Pharmacy, Department of Pediatrics, Jassy, Romania
³ “St. Mary” Children’s Emergency Hospital, Pathology Department, Jassy, Romania
⁴ “St. Mary” Children’s Emergency Hospital, Surgery Department, Jassy, Romania

Children with intestinal vascular malformations may suffer from abdominal pain, emesis, gastrointestinal bleeding, obstruction or intussusception; some cases are asymptomatic.

A boy aged 5 presented at the ER unit with impaired general condition, fever, vomiting, abdominal pain and constipation for three days. Abdominal guarding, contracture and a firm tumoral mass in the right abdominal flank and fossa were found. Laboratory data showed pancitopenia, acidosis, azotemia, hypoproteinemia, hepatic cytolysis. US and plain X-Ray showed massive overflow fluid in the peritoneum. Emergency surgery found hemoperitoneum, intestinal oclusion caused by a „tumor” including the cecum and ascending colon and perforation of the transverse colon; a right extended hemicolecotomy was performed. Postoperatively condistions worsened progressively, with DIC, hemorrhagic shock, sepsis and exitus within 24 hours. Histopathology identified a giant cavernous vascular malformation of the cecum and the ascending colon.

Benign vascular lesions of the gastrointestinal tract, extremely rare in children, include hemangiomas, lymphangiomas and vascular malformations. Early imaging is essential for diagnosis: abdominal ultrasound, barium enema, colonoscopy, scintigraphy, CT, MRI and also mesenteric arteriography are useful. In our case, the malformation had a silent evolution with a huge increase in volume that led to intraperitoneal rupture with severe postoperative complications and exitus.

###

**WIDENING THE SCOPE OF THE 15q13.3 MICRODUPLICATION SYNDROME. PATIENT REPORT AND GENOTYPE-PHENOTYPE CORRELATION.**


Department of Sciences for Health Promotion and Mother and Child Care. University of Palermo. Palermo, Italy.

We report the case of a 6 year-old male infant, first son of healthy non-consanguineous parents. He was born at 35.5 WG due a cearean section performed as an emergency because of eclamptic gestosis. At birth he needed to be hospitalized in a Neonatal Intensive Care Unit due to respiratory difficulties and hypotonia. By the age of 9 months, his clinical history was characterized by developmental milestone delay, especially in receptive and expressive language, fine motor and visual motor integration areas. At the age of 6 years, as a result of the diagnosis of a slight intellectual disability (ID) associated with mild dysmorphic features, the patient was referred to the Genetics Department. Our patient’s evaluation included chromo-
some and molecular analysis for fragile X syndrome which were normal, while the CGH Array analysis revealed a paternal derived microduplication of 0.5 Mb in size at the 15q13.3 chromosome. Copy number variants (CNVs) at the 15q13 chromosome have been implicated in multiple neuropsychiatric conditions, including autism spectrum disorder (ASD), schizophrenia and ID. Chromosome 15q13 is a hotspot for CNVs due to the presence of low copy repeat (LCRs) elements that predispose it to homologous recombination events and mediate nonallelic homologous recombinations (NAHR), resulting in genomic rearrangements of the 15q13.3 chromosome. Interestingly, this microduplication encloses the CHRNA7 gene locus which codifies for the α7 nicotinic acetylcholine receptor in the human brain. A dosage sensitivity effect of this protein has been proposed to play a major role in impaired cognitive and behavioral phenotypes. Furthermore, clinical data and family histories of patients with small 15q13.3 microduplications actually suggest that these rearrangements might be associated with developmental delay/intellectual disability, muscular hypotonia, and a variety of neuropsychiatric disorders with a wide range of severity and partially penetrant phenotypes.

###

A CONTINUOUS SEARCH... A CASE OF ACUTE DISSEMINATED ENCEPHALOMYELITIS, ADEM

Caporelli N. 1, Antonini L. 1, De Angelis F. 1, Siliquini S. 2, Osimani P. 3

1 Department of Pediatrics, Università Politecnica delle Marche, Children Hospital Salesi, Ancona, Italy.
2 Pediatric Neurology Department, Children Hospital Salesi, Ancona, Italy.
3 Division of Pediatric Infectious Diseases, Children Hospital Salesi, Ancona, Italy.

ADEM is a rare disease of CNS. It is a diagnosis of exclusion and neuroimaging might be normal at onset. A.A., 3-year-old girl, was brought to our Hospital with irritability and numbness. Fever and cold 7 days before admission. Neurological examination revealed irritability, nucal rigidity, back pain, Kerning and Brudzinski signs. Raised inflammatory markers (WBC 19000/mm c N 83% e CRP 5.9 mg/dl). EEG revealed generalized cerebral dysfunction with no definitive irritable foci. She was admitted to the Pediatric Department and managed with Cefotaxime. MRI of brain was normal. Lumbar puncture: CSF cell count was 54/mm c, negative CSF cultures. The antibiotic therapy led to an initial improvement with defervescence on day 3. On day 8 clinical condition worsened, she experienced fever without shiver, mild hand-tremors and feet-clonus. A second MRI revealed disseminated multifocal lesions consistent with edema, inflammation, and demyelination. Diagnosis of ADEM was considered and she was started on IV methylprednisolone pulse therapy for 5 days, followed by prednisone with good response and gradual neurological improvement. On follow up after 2 months: neurological examination was normal and MRI showed a decrease in the brain lesions. Sometimes during initial course of disease BRAIN MRI can be normal: initial MRI scan in our case showed no evidence of ADEM but later it revealed its characteristic features. Early diagnosis and treatment holds the key to a favorable outcome.

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SHINGLES AND MOVEMENT DISORDERS ... NOT ONLY ATAXIA!

Antonini L. 1, Caporelli N. 1, Grilli M. 1, De Angelis F. 1, Cesaroni E. 2, Osimani P. 3

1 Department of Pediatrics, Università Politecnica delle Marche, Children Hospital Salesi, Ancona, Italy.
2 Pediatric Neurology Department, Children Hospital Salesi, Ancona, Italy.
3 Division of Pediatric Infectious Diseases, Children Hospital Salesi, Ancona, Italy.

The case of a 9-year-old girl who presented with a 1-week history of movement disorders during Shingles is dis-
cussed. Chickenpox at 8 years of age; febrile pharyngitis treated by a 7-day course of oral amoxicillin 3 months before admission. Recent medical history: agitation and motor difficulties the day after a booster injection. For these symptoms and onset of vesicles in the right hemithorax, she was initially admitted to the ER and with the suspicion of cerebellitis associated to Zoster, transferred to the Infective Department. At medical examination: evidence of crops of vesicles, consistent with Shingles; 3/6 ejection systolic murmur, involuntary generalized movements, psychomotor agitation, emotional lability and speech impairment. ASLO 1900 UI/ml, CRP 2.1 mg/dl and negative throat culture for GABHS. Echocardiogram revealed left atrial enlargement, severe mitral insufficiency (MI), thickened valvular leaflets. A diagnosis of Sidenham’s Chorea was made. Treatment with prednisone, furosemide, potassium canrenoate, captopril and carvedilol was begun. Beta-lactam, acyclovir and BDZ were also administered. She subsequently improved and at 2 months follow-up regression of neurological symptoms was observed and moderate MI with good hemodynamic compensation was still detectable. Although the diagnosis of Chorea is made infrequently, it should be included in the differential diagnosis of movement disorders. An accurate cardiological evaluation should be performed to rule out rheumatic fever.

TUBEROUS SCLEROSIS COMPLEX IN A PATIENT CARRYING AN ATYPICAL GENOMIC REARRANGEMENT


Department of Sciences for Health Promotion and Mother and Child Care. University of Palermo. Palermo, Italy.

Tuberous sclerosis, also called tuberous sclerosis complex (TSC), is a rare, multi-system genetic disease that causes benign tumors to grow in the brain and on other vital organs such as the kidneys, heart, eyes, lungs, and skin. It usually affects the central nervous system and results in a combination of symptoms including seizures, developmental delay, behavioral problems, skin abnormalities, and kidney disease. The disorder affects as many as 25,000 to 40,000 individuals, with an estimated prevalence of one in 6,000 newborns. The wide range of organs affected by the disease implies an important role for TSC1 and TSC2 genes located at 9q34 and 16p13 chromosomes. These genes encode respectively for hamartin and tuberin proteins which are involved in cell proliferation and differentiation. We report on clinical findings detected in a patient with TSC and glomerulonephritis carrying a 1 Mb deletion at the 12q12.3q13 chromosome detected by an CGH-Array analysis but without mutations at the TSC1 and TSC2 loci. Furthermore, the 1 Mb deletion we report on, harbors only 16 known genes, and since there is no gross functional imbalance, we can speculate that the phenotype observed in our patient probably results from a haploinsufficiency effect of one or more genes located within the deleted region. Finally this evidence, in combination, provides a unique opportunity to make a genotype-phenotype correlation never before reported.
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