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Palace Hotel Bari (Italy)

TRAUMI SPORTIVI
ACUTE TRAUMA: GETTING THROUGH THE NIGHT

ABSTRACT BOOK

# ABSTRACT BOOK

SCIENTIFIC SESSION OF A.I.S.O.T. (ITALIAN ORTHOPEDIC AND TRAUMA RESIDENTS ASSOCIATION) DURING 47TH NATIONAL CONGRESS OF O.T.O.D.I. (ORTHOPEDIC AND TRAUMA SURGEONS OF ITALY)

**Bari (Italy) 12 May 2016**

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During the 47th National Congress of O.T.O.D.I. (Orthopedic and Trauma Surgeons of Italy) in Bari the Scientific Session of A.I.S.O.T. (Italian Orthopedic and Trauma Residents Association) was held.
A.I.S.O.T. promotes and supports the links and exchanges between Italian Ortho and Trauma Residents. The board includes 9 delegates who are elected annually and over 250 active members.
The scientific session, organized and overseen by the Residents of the University of Bari, was attended by residents from all Italian Ortho and Trauma Schools.

The TOPICS discussed were:

SPORT TRAUMA
- Quadriceps Tendon
- Achilles Tendon
- Elbow Luxation
- Muscular Lesions

ACUTE TRAUMA: GETTING THROUGH THE "RESIDENT" NIGHT
- Upper Limb
- Lower Limb
- Paediatrics
- Spine
- Damage Control
- Legal Aspects of the Italian Residents "on call"

## ABSTRACT BOOK

### NON OPERATIVE VERSUS OPERATIVE TREATMENT OF ACUTE ACHILLES TENDON RUPTURE: A META-ANALYSIS OF RANDOMIZED TRIALS

Vittorio Nappi, Biagio Moretti.

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**OBJECTIVES**: There are many treatment options for acute Achilles tendon rupture. The aim of our study was to compare, with a of meta-analysis review, conservative treatment with open repair and percutaneous repair for Acute Achilles Tendon Rupture(AATR)

**METHODS**: We systematically searched six electronic databases: Cochrane Bone, The Journal of Bone and Joint Surgery, Injury 2008, Foot and Ankle Surgery, Knee Surg Sports Traumatol Arthrosc (2008) and The American Journal of Sports Medicine, to identify randomised controlled trials (RCTs) and reference lists of articles in which non operative treatment was compared with operative treatment (open and percutaneous repair) for AATR.
**SELECTION CRITERIA:** All randomised and quasi-randomised trials comparing surgical versus non-surgical treatment or different surgical methods for acute Achilles tendon ruptures in adults.
**RESULTS**: The choice of surgery technique is not straightforward, and various forms of open surgery and percutaneous techniques exist. Open operative treatment of Achilles tendon rupture significantly reduces the risk of rerupture compared to non operative treatment, but this treatment is associated with a significantly higher risk of other complications including infection, adhesions, sural nerve injury or sensory disturbance. Percutaneous repair reduces postoperative complications, pain and operation times, and enables faster recovery, enhancing overall patient compliance, but has a higher risk of sural nerve injury. This complication may be associated mainly with lack of vision during surgery, which leads to damage of the nerve.
**DISCUSSION**: A meta-analysis of randomised controlled trials comparing conventional to minimally invasive approaches for repair of an Achilles tendon rupture have demonstrated that MIS ( Minimally Invasive Surgery) has a significantly reduced risk of superficial wound infections, reduces operation times, and enables faster recovery, enhancing overall patient compliance and satisfaction for good to excellent results compared with conventional open surgical approaches.
Postoperative splinting can be performed with a rigid cast (proximal or distal to the knee) or a more mobile functional brace that reduces the overall complication rate. Postoperative early weight bearing combined with early ankle motion exercises is associated with a lower minor

complication rate and achieves superior and more rapid functional recovery than conventional immobilization after surgical AT repair.
CONCLUSION: There is no agreement on the ideal type of surgical management for Achilles tendon rupture; the open and the percutaneous technique are both safe and effective in repairing the ruptured Achilles tendon and both afford the same degree of restoration of clinical, ultrasound and isokinetic patterns. Currently, open repairs may still be preferable in younger and more physically demanding individuals requiring greater push-off strength, although there are risks of wound complications. Modern percutaneous techniques followed by early functional rehabilitation are becoming increasingly popular with good early results.
The choice of rehabilitation management after the surgical repair of acute Achilles tendon (AT) ruptures remains controversial due to the insufficient clinical evidence.

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## TERRIBLE TRIAD OF THE ELBOW: ITS EFFECTS ON RETURN TO SPORTS

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*[2]Dario Camberlingo Hospital – Francavilla Fontana - Italy*

OBJECTIVE: To review the injury mechanism and the treatment progress of terrible triad of the elbow, and to analyze its effects on return to sports activities.
METHODS: Related literature concerning terrible triad of the elbow was extensively reviewed and comprehensively analysed but the clinical outcomes and its effects on return to sports are still debated.
RESULTS: The main treatment of terrible triad of the elbow is operation. The ultimate goal of treatment is to reconstruct sufficient stability of the elbow. The treatment includes fixation of the coronoid by suture, screw or plate; fixation of radial head by screw and plate, partial or complete replacement of the radial head; fixation of lateral collateral ligament and the medial collateral ligament by bone suture or anchors and the application of the external fixator. These surgical treatments have their own indications and advantages. The rate of return to ≥1sportswas 80% of patients at an average of 7.0 months postoperatively; 75.0% returned to their previous intensity and level of sport. Patients who played a sport involving throwing had problems, and felt that they could not throw a ball normally.
CONCLUSION: Generally, surgery is needed to maintain the stability of the elbow for patients with terrible triad elbow. However, whether medial ligament repair or not, the choice of approach, and mechanism of injury still need further study. 80% of patients return to full sports at an average of 7 months post injury. For throwing sports, patients complained of decreased range of motion during throwing. The possible outcome should be considered preoperatively in this type of candidate.

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## USE OF PLATELET RICH PLASMA IN MUSCLE INJURIES. OUR EXPERIENCE

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INTRODUCTION: Musculoskeletal injuries represent one of the most common causes of long term pain and physical disability, affecting hundreds of millions of people around the world and accounting for the majority of all sport-related injuries. The best management for muscle injuries has not been identified. The results of healing with conventional therapy including rest, ice, compression, and elevation (RICE) are often inadequate, and also the therapies based on Non steroidal Anti-inflammatory Drugs (NSAIDs) are quite controversial. For

these reasons, beside classic conservative treatments, new therapeutic approaches, such as the use of growth factors, were developed in order to enhance biologic response and healing processes. One of the most innovative systems is based on Platelet-Rich Plasma (PRP). PRP is a plasma layer, separated after centrifugation of an autologous blood sample, in which platelets are about 8 times more concentrated than in normal plasma. The rationale for this technique is based on scientific evidence of the importance of growth factors, contained in platelet granules, in the normal muscle tissue healing process.
METHODS: We report the effects on clinical, functional and instrumental outcomes of US-guided injections of autologous PRP in 20 patients with musculoskeletal injuries, aged between 18 and 43 years, treated in our clinic between 2010 and 2013.
RESULTS: At two month follow-up pain was totally absent in 13 patients and slightly reduced in 6 patients, while MRI showed a normal pattern in 16 cases and a significant reduction of tissue edema in 3 cases. In only 1 patient was there no clinical or instrumental improvement.

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## QUADRICEPS TENDON RUPTURE: A SYSTEMATIC REVIEW.

Giuseppe Lavecchia, Giovanni Vicenti, Massimiliano Carrozzo, Biagio Moretti.

*University of Study of Bari – Orthopaedic and Trauma Department – Bari - Italy*

INTRODUCTION: Rupture of the quadriceps tendon is an uncommon yet serious injury requiring prompt diagnosis and early surgical management. It is more common in older (>40 years) individuals and is sometimes associated with underlying medical conditions. In particular, bilateral spontaneous rupture may be associated with gout, diabetes, or use of steroids. Clinical findings typically include the triad of acute pain, impaired knee extension, and a suprapatellar gap. Imaging studies are useful in confirming the diagnosis. Although incomplete tears may be managed non surgically, complete ruptures are best treated with early surgical repair.
CONCLUSION: Worst results were noted in delayed repairs. Reported complications included heterotopic ossifications, deep venous thrombosis or pulmonary embolism, superficial and deep infection. It appears that the type of surgical repair does not influence the clinical results. The majority of the studies reported good or excellent ROM and return to pre-injury activities. The overall rate of re-rupture was 2%.

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## CHALLENGES OF THE SUB-AMPUTATED HUMERUS: TO AMPUTATE OR NOT? A CASE STUDY.

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OBJECTIVES: The goal of this case study is to demonstrate, based upon the literature, how, when and why it is necessary to save an upper limb.
METHODS: The goal of this case study is to demonstrate how, when and why to save the upper right limb of a 32-year-old male. At 3:30am, after a motorcycle accident against a guardrail, a patient was admitted to the G. Rummo Emergency Department with an exposed diaphyseal fracture of the humerus (Gutilo Anderson IIIC) with a torn off humeral artery lesion, massive loss of skin and muscle, MESS equal to 8, and ipsilateral fracture of the radial epiphysis and distal radial diaphysis. In the emergency room, an external fixator bridge was applied to perform osteosynthesis of the fractures. The saphenous vein was used to replace the damaged humeral

artery and an extensive debridement was performed with sliding flap closure for the contused, lacerated wound with loss of substance. The patient was monitored with a clinical follow up every 15 days and radiological follow-up for up to 12 months after surgery. Clinical parameters were: Constant Shoulder Score, the Mayo Elbow Score, the Mayo Wrist Score, and bone healing was evaluated radiologically.

**RESULTS:** The healing of the humerus-diaphyseal fracture occurred nine months after the trauma, while the diaphyseal and epiphyseal fractures of the distal radius occurred two months after surgery. All of the Constant Shoulder Scores, the Mayo Elbow Scores, and the Mayo Wrist Scores were comparable to pre-trauma. Additionally, the sliding flap took hold in 30 days.

**DSCUSSION/CONCLUSION:** Only 1.2% of all humeral fractures are exposed. Very often the exposed fractures of this region result from high energy trauma or crushing. The exposure of the fracture is most often associated with a significant neurovascular lesion of the upper limb. Unlike the lower limb, the decision to amputate is very difficult because the upper limb functional arthrodesis outcomes are more advantageous compared to the prosthesis. The decision to salvage or amputate a limb is generally made calculating the Mangled Extremity Severity Score (MESS). However, the reliability of this score in deciding to save or amputate the upper limb in ballistic or high speed traumas is challenging. The mortality and morbidity of sub-amputated limb trauma are generally associated with a delayed revascularization, the mechanism of injury, anatomy of the damaged site, associated lesions, age, and environmental circumstances. From the data available in the literature and from war experience, it can be concluded that the immediate irrigation, the debridement, the immediate osteosynthesis of open humeral fractures, and early revascularization lead to good-to-excellent outcomes.

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## OPEN BILATERAL SUPRAINTERCONDYLAR FEMORAL FRACTURE DURING A NIGHT SHIFT: TIMING AND MANAGEMENT

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**INTRODUCTION:** During a night shift, a 45 year old woman, victim of a car accident came to our attention, with clinical and radiological signs of supraintercondylar fracture of both femurs and right patella fracture.

Given the exposure of the fractures and the need to stabilize a bilateral femoral fracture in order to prevent the risk of immediate complications (fat embolism, anemia, vascular and nervous deficits), emergency procedure was activated.

**TREATMENT:** With patient in supine position on Maquet table, the exposed wounds were extensively washed with physiological water. Using the image intensifier, a series of images were taken including pelvis, proximal epiphisis of both femurs, both tibia, ankles and feet, these last revealing a dislocation of the left Chopart articulation with an associated fracture of the first MT proximal epiphisis, undiagnosed during the first radiographic examination.

The surgery started from the right knee. Given the fracture pattern and the short time since the trauma took place, the ostheosynthesis of the supraintercondylar fracture was performed with cannulated headless screws and a patella tension bending with a double wiring.

Subsequently, the procedure moved to the left knee: considering the type of fracture and the apparent loss of bone substance, the surgeons opted for a reduction and stabilization of the knee joint with a temporary external fixator. Finally, reduction and stabilization of the fracture -dislocation of the left Chopart articulation was accomplished by using K-wires. The postoperative course proceeded without major complications.

**CONCLUSIONS:** The complexity and the urgency of the patient's clinical situation guided the surgeon in choosing the best

surgical treatment for the two supraintercondylar fractures, in one case opting for a definitive treatment and in the other for a temporary ostheosynthes is that would be converted later.
Also, attention should be paid to the accurate pre-operative study of these patients, victims of high-energy trauma, of which accurate radiographs are often not performed in ER making it necessary, once in the operating room, to perform a series of shots with the image intensifier in order to exclude urgent interventions such as dislocations or over- or under segmental fractures over the prior fracture, which can influence the type of surgery that has to be performed.

####

## SUPRACONDYLAR FRACTURES OF THE HUMERUS, GARTLAND 3: PEDIATRIC URGENCY. CASE REPORT AND LITERATURE REVIEW.

Salvatore Di Giacinto[1], Luigi Meccariello[2]

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**OBJECTIVES:** Using a clinical case, we aim to show the proper management of humeral pallet fractures in children.
**METHODS:** A 6-year-old male child arrived at22:30 after falling from a trampoline. The injury had occurred 8hours earlier. Physical examination showed the presence of a vast dorsal hematoma of the entire humerus and the presence of Tucker Sign, but there were no neurovascular deficits present.
The patient was operated on 10 hours post-injury for closed reduction with lateral percutaneous pinning, and then packed with a humeral-wrist open brace for 45 days. We evaluated the patient with a clinical follow-up using the Mayo Elbow Performance Score (MepS) and x-rays with Carrying angle comparative to 15 days, 1-3-6-12 months post-injury. Our evaluation endpoint was 2 months after injury.
**RESULTS:** K-wire removal at 45 days. FU at 1 year: MEPS: 100; Δ Carrying angle (Carrying angle comparative): 0; Complete functional recovery and recovery of pronosupination.
**DISCUSSION / CONCLUSION:** Elbow fractures in children account for about 8% of upper limb fractures. The most common age for supracondylar fractures is between 5-10 years old, with a higher incidence in males (a ratio of 3 to 2) and more common in non-dominant limbs. These injuries occur when the limb is in extension. These fractures often generate anxiety and concern among experts due to the fact that they are rarely observed in non-specialized hospitals, and their treatment is mainly handled in super-specialized facilities. The most feared complications are definitely neurovascular. Volkmann's syndrome has been described in these injuries.
Neurovascular injury can stem from: damage derived directly from the traumatic mechanism, from kinking of the fracture site, and from imprisonment resulting from reduction maneuvers. The most feared complication is vascular compartment syndrome. In cases of neurovascular injuries (absence of peripheral pulses, ischemic hand) the emergency regimen is to treat these injuries urgently, within 6 hours.
The positive results obtained in this clinical case would suggest that urgent surgical treatment with closed reduction and percutaneous pinning is advisable for Gartland type 3 fractures.
This is a technique that requires an adequate learning curve: the choice of the surgical approach, which is necessary in cases of a non-satisfactory closed reduction, depends on the characteristics of the specific injury.

####

## MASSIVE LUMBAR DISC HERNIATION WITH COMPLETE DURAL SAC STENOSIS: A CASE REPORT

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**INTRODUCTION:** Lower back pain is a common orthopaedic problem. However, the incidence of symptomatic lumbar disc herniation is very rare. We report a case of massive lumbar disc herniation (LDH) with a complete dural sac stenosis.
**PRESENTATION OF CASE:** A 45-year-old man presented with lower back pain and leg pain below the knee. He had severe weakness of the left extension halluces longus, left ankle dorsiflexion. MRI showed a large disc herniation at L3/4 expanded further to the spinal canal. Complete dural sac stenosis was defined as no recognizable rootlet and cerebrospinal fluid signal on T2-weighed axial MR images.
The L3/4 discectomy with L3/5 stabilization was performed. In the immediate postoperative period, the patient experienced complete relief from the leg pain.
**CONCLUSION:** A massive LDH with complete dural sac stenosis is associated with severe back and leg pain at presentation, however surgical treatment cannot be deferred when significant neurological symptoms occur.

####

## FROM DAMAGE CONTROL ORTHOPAEDICS TO FINAL TREATMENT OF FLOATING KNEE FRASER IIB WITH BONE LOSS: A CASE REPORT.

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**OBJECTIVES:** The objective of this case report is to describe management of and optimize the possible therapeutic solutions for the treatment of femoral bone loss associated with an open fracture IIIB Gustilo Anderson (GA) in a polytrauma that includes Floating Knee (GF) Fraser IIB from Damage Control Orthopaedics to final treatment.
**METHODS:** After a high kinetic energy car accident, a 23-year-old patient arrived at our Emergency Department with an Oro-Tracheal Intubation and an Injury Severity Score of 24 points (moderate concussion, moderate splenic ulcerations, left distal femur open fracture IIIB GA and truncated diaphyseal open fracture of the leg IIIB GA). The patient was treated in the emergency room with an external fixator Femorotibial Bridge after extensive cleaning and debridement of open fractures. The patient was treated with a specific antibiotic prophylaxis and monitored with laboratory tests. The patient underwent an operation on the seventeenth day post-trauma with a LISS plate (Less Invasive Stabilization System) and screws with contralateral allograft bone strut. The follow-up was clinical using the Knee Injury and Osteoarthritis Outcome Score (KOOS), Short Form 12 Health Survey (SF-12) for quality of life, and radiological at 1-3-6-12-18-24-36 months.
**RESULTS:** Immediately post-surgery, the patient performed active and passive mobilization of the knee. Three months after the first surgery, the patient underwent surgery for the intramedullary nailing of the tibia. The patient walked with partial load up until the sixth month after injury, and then began a progression to a total load. At 24 months post-surgery, the patient had both the KOOS and SF-12 at 100 points.
**DISCUSSION / CONCLUSION:** In 1975, Blake & McBride defined Floating Knee as a lesion, consisting of ipsilateral fractures of the femur and tibia, which disengages the knee from the rest of the limb. The dramatic nature of this injury, which occurs in almost all multiple trauma patients, requires swift action and decision

by the orthopedist-traumatologist surgeon to improve patient outcomes. The surgical management of Floating Knee was heavily influenced by improvement in surgical techniques and synthetic methods. Not all Fraser IIB are equal, some have both the fracture points exposed and are associated with serious aesthetic conditions and severe impairment of the joint line. The timing of treatment should be discussed case by case. The surgical sequence should be respected: first, the fixation of the femur, and then the stabilization of the tibia, taking into account the condition of the skin, eventual exposure or the eventual level of sub-amputation of the limb. The clinical and radiographic results show how efficient damage can lay the foundations for an excellent definitive treatment.

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## ITALIAN JURISPRUDENCE AND THE LEGAL STATUS OF DOCTORS IN TRAINING DURING GUARD DUTY.

Davide Ferorelli[1], Francesco Emanuele Celentano[2].

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[2]*PhD Student in International and European Law at the Law Department of the University of Bari.*

The legal aspects that interest the doctor in training need to be based on clear standards and rules. Unfortunately, jurisprudence in this field is subject to many different interpretations and decisions related to the final application of law. It is for this reason that it has become more and more important, and necessary, to clarify which rules should be applied to the entire category of doctors in training, so that their own training can develop in an organized manner.
A fundamental starting point is the contract, signed by each doctor before starting their training, ruled by the provisions of art. 37, Legislative Decree n. 368/199 and the subsequent changes made to it subject to the contract prototype adopted by Prime Minister's Decree on July 7, 2007. The contract clearly states that training is aimed at acquiring the knowledge necessary for specialist competences in consideration of the European legislation in this filed. It also stresses the importance of the principle that the doctor in training could never replace the specialist doctor.
Unfortunately, as is well known in the medical field, that too often the doctor in training is considered a useful replacement for the fully trained personnel and this mistaken view of the roles causes the trainees training to suffer through loss of time. There are many cases brought against doctors in training for medical responsibility, despite their particular, and often not well defined, status of trainees with the guarantees that this should determine for them.
The analysis of some jurisprudential cases shows that frequently doctors in training are convicted for medical responsibility, unskillfulness and negligence in situations where they were accused of failure to reduce risk and, sometimes, also for refusing the task. In this field of analysis sentence 24/11/1999 n. 5311 of the Court of Cassation penal section seems fundamental. It states that considering the medical degree and the ability to practise, the trainee too has an important duty with regards to patient health. It seems clear, though, that the Italian Supreme Court has ignored the ontological limits for the category composed of doctors in specialist training. In another case, a doctor in training was convicted as part of a guilty medical *equipe* [18/05/2005 n.18568 of the Court of Cassation, penal section]. Frequently, in fact, the Supreme Court has denied the existence of a clear law that could limit the role of the doctor in training to that of a trainee. In various instances the Court explained that even if the trainee just acts under the guide of the tutor, they are obliged to control, in the limits of their competences, and eventually to contest and to refuse to apply, the medi-

cal choices made by the full doctor if they are considered wrong or dangerous for the patient. This ambiguous role has to be enacted, of course, with respect to the principle of "gradual responsibility" ruled by the law and jurisprudence [01/08/2008 n.32424 of the Court of Cassation, IV penal section].

In conclusion, it seems necessary for the legislator to create a uniform and clear legal framework that could override the enormous number of laws, regulations and contracts that now rule the work of doctors in training. A *no blame culture* should guide the legislator in the creation of new uniform regulation that could guarantee the transition to a new medical environment in which trainees could work, cooperate and act in a safe and, above all, legitimate way.

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# ABSTRACT BOOK
## 1° NATIONAL EVENT "IMAGES IN PNEUMOLOGY"

## Bari (Italy) 06-07 June 2016

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## A CASE OF ACUTE RESPIRATORY DISTRESS SYNDROME EXPOSING UNKNOWN ANTI JO-1 SYNDROME-RELATED INTERSTIAL LUNG DISEASE

Pierluigi Intiglietta, Pierluigi Carratù, Giorgio Castellana, Lorenzo Marra, Onofrio Resta.

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Marcantonio, a 65 year old caucasian male, complained to his GP of dry cough, fatigue and fever with shaking chills. A chest X-Ray examination was carried out; it showed multiple consolidations and interstitial thickening areas in medio -basal zones. Ciprofloxacin was empirically administered with no improvement. He was then hospitalized and HCV, HBV, HIV, Legionella and Pneumococus were ruled out; high levels of ESR and CRP were detected. A few days later, due to the occurrence of acute respiratory distress syndrome (ARDS), he was admitted to the Intensive Care Unit where Assisted Mechanical Ventilation was performed. A chest HRCT was then carried out revealing evidence of Non Specified Interstial Pneumonia (NSIP) with multiple consolidations with air bronchograms, diffuse ground-glass areas and pneumomediastinum (Figure 1); moreover, bronchial aspirate revealed Acinetobacter Baumani Complex Infection. Broad spectrum antibiotic therapy and high dose glucocorticoids were administered daily. Once stabilization had been achieved, the patient was transferred to the Respiratory Medicine Unit where autoimmunity tests were performed showing strong positivity to anti-ENA Jo-1 autoantibodies; after a rheumatic consultation, treatment with cyclophosphamide 100 mg die, in combination with methylprednisolone 32 mg die was established. The patient was discharged with a diagnosis of Acute Respiratory Failure secondary to bilateral Bronchopneumonia and Anti Jo-1 syndrome-related Interstitial Lung Disease (ILD). After two months of therapy a follow up HRCT was carried out showing neither consolidations nor pneumomediastinum, scarce interstial thickenings, and a persistence of the ground-glass areas (figure 2); spirometry showed restrictive lung function with moderate DLCO reduction. It is evident that previously unknown ILD related to rheumatic diseases may be discovered only when acute conditions, such as bronchopneumonia, occur. In these cases, antibiotic therapy followed by steroids and immunosuppressive drugs are fundamental to control such diseases.

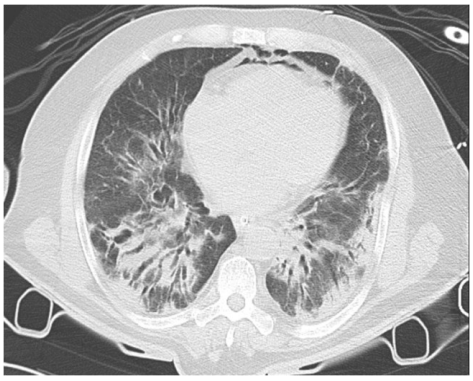

**Figure 1:** HRCT: radiological findings compatible with fibrotic NSIP

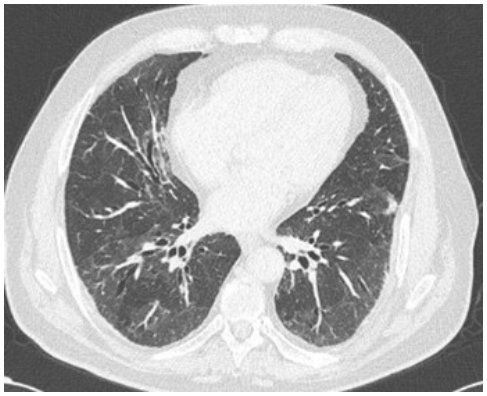

**Figure 2:** Follow up HRCT after two months of therapy.

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## A CASE OF SUBACUTE EXTRINSIC ALLERGIC ALVEOLITIS

Agnese Caringella, Luciana Labate, Silvano Dragonieri, Anna Rita Tassiello, Onofrio Resta.

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A 58-year-old woman, smoker (10 P/Y), suffering from high blood pressure, anxiety-depressive syndrome and reflux disease, has a history of cough and sputum from unspecified time. In early 2015, her condition exacerbates with the concurrent suspension of the tobacco habit and she complains about exertional dyspnea, weight loss and fatigue. She is on various antibiotics and in aerosol therapy with little benefit. In July 2015, she undergoes a HRTC that shows centrilobular micronodules, excepting lower lobes, and disseminated patchy areas of air trapping (Figure 1-2). She is therefore hospitalized. The physical examination reveals disseminated rhonchi and nail clubbing. Pulmonary function tests show moderate reduction of DLCO (54% of predicted). Laboratory tests are normal. Accordingly, she is subjected to fibrobronchoscopy and bronchoalveolar lavage (BAL) that highlight lymphocytosis (20%) and CD4/CD8 ratio < 0.2. The patient interview immediately emphasizes an intermittent but long history of exposure to birds (parrots, canaries, finches). After a multidisciplinary discussion among the pulmonologist, the radiologist and the pathologist, on the basis of the positive anamnesis for exposure to poultry and due to the compatible clinical, radiological and functional context, the patient is diagnosed as having subacute extrinsic allergic alveolitis. We therefore recommend: 1) the removal of poultry; 2) prednisone (37 mg per day; tapering every 4 weeks). After six months of therapy, the patient reports a sharp improvement in cough, sputum, dyspnea, radiological and pulmonary function tests (DLCO 62% of predicted).
Extrinsic allergic alveolitis, also known as hypersensitivity pneumonitis (HP), is a complex syndrome caused by continual inhalation of and sensitization to a large variety of aerosolized antigens which are widely classified in 5 categories: 1) Bacteria; 2) Fungi, yeasts; 3) Mycobacteria; 4) Animal proteins; 5) Chemicals. The most common causes of HP are avian antigens and microbial agents. Avoiding exposure to a suspected or certain causative agent and treatment with systemic corticosteroids are the basis of HP management.

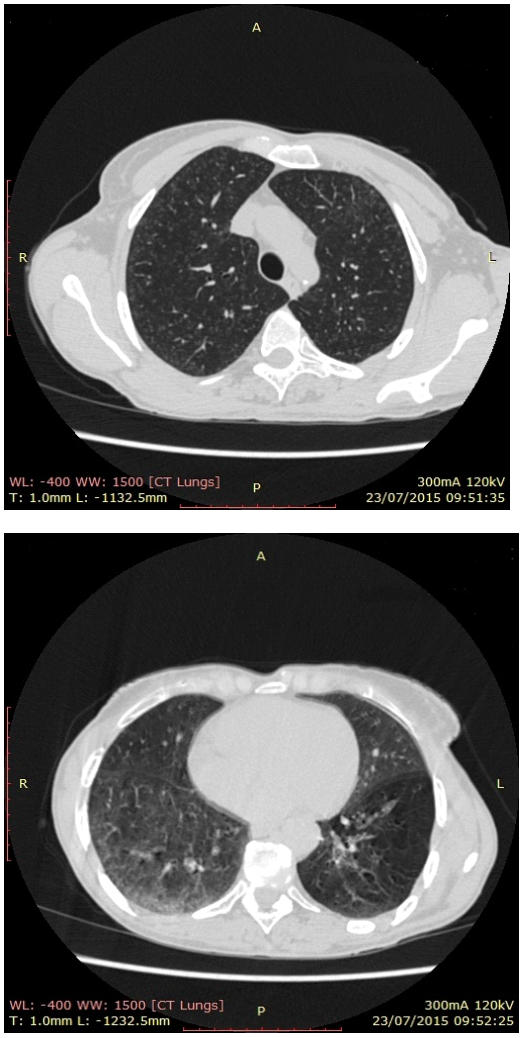


**Figure 1 -2:** HRTC showing centrilobular micronodules, excepting lower lobes, and disseminated patchy areas

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## SEVERE OSA: EFFICACY OF ORAL APPLIANCE THERAPY

Giorgio Castellana, Pierluigi Carratù, Agnese Caringella, Pierluigi Intiglietta, Onofrio Resta.

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D. is a 50 year old male, he has never smoked. His medical history includes arterial hypertension, dyslipidemia and gastroesophageal reflux. His wife reported breathing pauses during sleep.

When admitted to our outpatient department, he complained daytime sleepiness (Epworth sleepiness score 10 out of 24), memory deficit, dry mouth in the morning, loud snoring, nocturia (2-4 times for night) and frequent nocturnal awakenings. Physical examination revealed blood pressure of 140/90 mmHg, BMI 24.5, neck size 41 cm, Mallampati score 2, long uvula, overjet and class II malocclusion. STOP-BANG score: 7. Pulmonary function testing revealed normal values, flattening of expiratory curve, paCO2 45mmHg and HCO3- 29.9 mmol/L. The patient underwent nocturnal Cardio-Respiratory Monitoring (CRM) due to the strong suspicion of Obstructive Sleep Apnea (OSA): Apnea-Hypopnea Index (AHI) was 58.9; Oxygen Desaturation Index (ODI) was 56.2; the percentage of the total time thatoxygen saturation level was lower than 90% (T<90%) 19.2%; median SpO2 91.7% (Figure 1). The patient showed intolerance to Continued Positive Airway Pressure (CPAP) therapy and was therefore submitted to ENT (sleep endoscopy) and Dental evaluation: at ENT, hypertrophy of lingual tonsil with oropharyngeal collapse >75% was found, which reduced after the mandibular advancement maneuver; the Dental evaluation excluded contraindications to oral appliance (OA) therapy. Our proposed therapy consisted of Mandibular Advancement Device (MAD or AOm): it was gradually advanced up to 7 mm, with progressive improvement in symptoms and indices on CRM: AHI 14; ODI 15.5; T<90% 1%; median SpO2 95%.
OSA is the most frequent of breathing sleep disorders and its usual treatment is nasal CPAP therapy. More recently, OAm has been shown to be efficacious in OSA and current practice parameters of AASM indicate this as a first-line therapy in patients with mild-to-moderate OSA (AHI <30) and in severe OSA patients who fail treatment attempts with CPAP therapy.

####

## AN ODD CASE OF PULMONARY NODULE WITH POSITIVE PET-TC

Maria Rosaria Vulpi[1], Vincenzo Nicola Valerio[1], Paola Terlizzese[2], Silvano Dragonieri[1], Onofrio Resta[1].

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Paolo, a 65 year old man, smoker (30 P/Y), without any comorbidities, was admitted to our ward for fatigue and cough with purulent sputum. A chest X-ray showed a fibrous nodule in the upper right pulmonary lobe. A contrast-enhanced thoracic CT confirmed the fi-

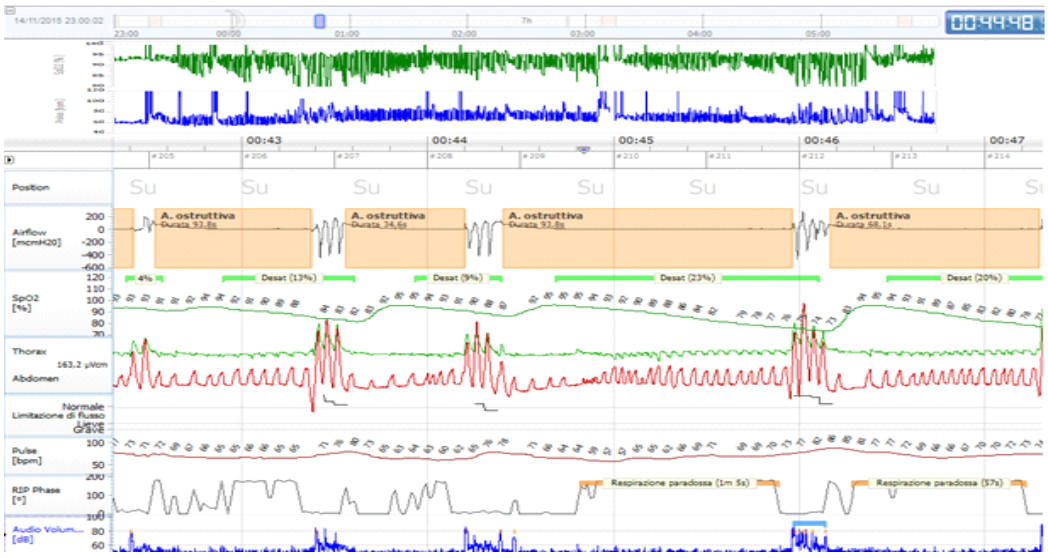


**Figure 1:** Nocturnal Cardio-Respiratory Monitoring

brotic nature of the lesion. The patient was discharged with the diagnosis of "COPD exacerbation". Two years later, the patient was readmitted to the hospital with similar symptoms. A chest X-ray revealed a new opacity with cavity in the upper left pulmonary lobe. A CT thorax revealed a speculated nodule of 2,5 cm diameter in that region (Figure 1). A bronchoscopy and broncho-alveolar lavage with cytological examination and microbiologic investigations were performed, and turned out negative. Furthermore, the tuberculin skin test and three sputum cultures were all negative for tuberculosis.

The analysis of two statistic models for the evaluation of malignancy risk of the nodule, the Logistic Regression and the Bayesian Analisis, revealed high risk.

In addition to that, the patient autonomously decided to undergo a PET-CT scan, that confirmed the suspicion of malignancy, showing a pathological uptake of FDG within the nodule (SUV max 4,5).

Eventually, the patient underwent surgery, which provided excision of the nodular lesion.

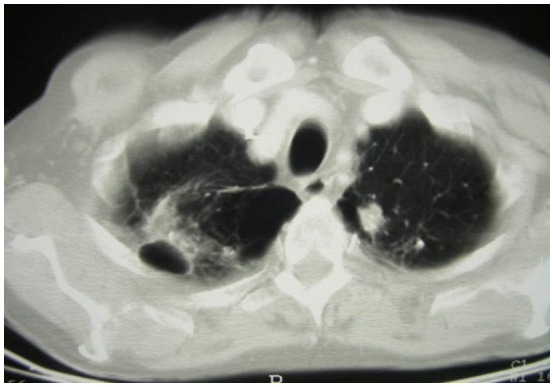

**Figure 1:** A CT thorax revealed a speculated nodule of 2,5 cm diameter in that region

The pathology report described "Necrotizing granulomas with multinucleated giant cells (foreign body appearance); presence of acid-fast bacilli noted; overall there is no evidence of malignancy". The cultures were positive for Mycobacterium Xenopi.

In conclusion, with regards to the differential diagnosis of a single pulmonary nodule, PET-CT scan should be performed in all patients with medium risk of malignancy, in whom this exam can help in orienting the diagnosis and guiding management. Furthermore, in the case of high malignancy risk, a cyto-histological exam of the nodule is of paramount importance.

####

## A CASE OF HIPOXIEMIA IN A PATIENT WITH ALS

Giuseppina D'Alba, Pierluigi Carratù, Vito Liotino, Giulia Gaudiuso, Onofrio Resta.

*Institute of Respiratory Diseases, Department of Surgical and Medical Sciences University of Bari*

A 54 year old man with arterial hypertension in his medical history was diagnosed5 months ago for spinal ALS . Spirometry F/V reported normal values with a Moderate reduction of the DLCO.

Blood –gas analysis in a supine was pH 7.41 pCO236 pO2 59 HCO3- 23 .Seated ABG showed pH 7.40 pCO2 37 pO2 63 HCO3- 23. The patient did not complain about nocturnal and or diurnal breathing symptoms and his laboratory tests were normal. The echocardiography, EKG, NT PRO-BNP plasma levels were normal. The chest X-ray did not show any parenchymal lesion. The Angiopulmonary CT excluded lung emboli and showed the presence of streaks of atelectasis in the inferior lung lobes (Figure 1-2). Polisomnographic findings showed TST 90: 45 % ODI:64 AHI:66SaO2 91%. The patient underwent mechanical ventilation (without oxygen FIO2: 21%) which was crucial for the resolution of respiratory distress and for the improvement of ODI:18,AHI:20 and SatO2: 94.3%

A new ABG on room air after nocturnal ventilation showed an increase in the pO2 of about 15 mmHg. The patient was discharged with a diagnosis of respiratory failure and severe OSAS with a prescription for NIV during night hours. During the outpatient follow-up the improvement in pO2 was confirmed. The respiratory disorders in patients with neuromuscular disease usually appear first during the night and then get worse turning into alveolar hypoventilation. It is much more

frequent to find patients with hypercapnia instead of only hypoxemia likely due to the pump failure, which is a common feature of this disease. In this patient, the reduction of diaphragm function, confirmed by a low SNIP value created a basal disventilation that disappeared only after a night of ventilation therapy, probably due to the recruitment of basal lung segments that are physiologically perfused and poorly ventilated. It is also important to underline that it is common to underestimate respiratory disease in neuromuscular patients

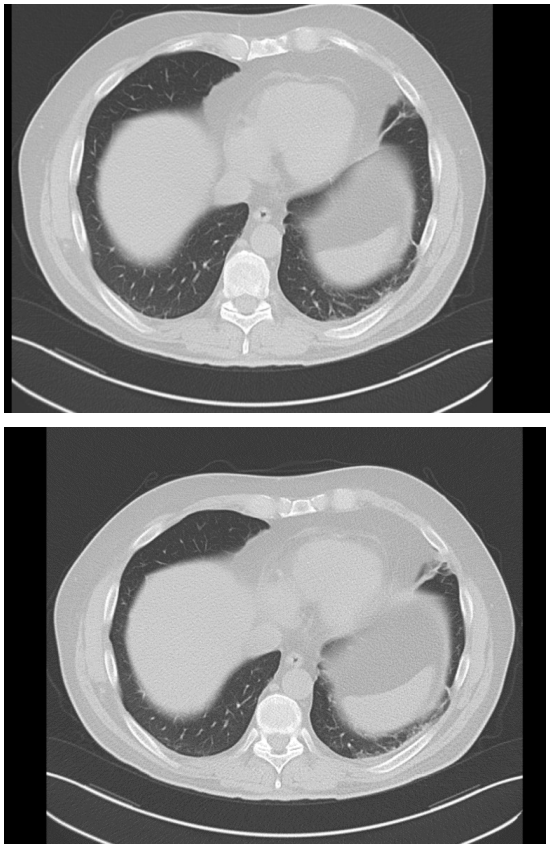

**Figure 1-2:** The AngioCT pulmonary excluded lung emboli and described the presence of streaks of atelectasis in the inferior lung lobes.

####

## ALLERGIC BRONCHOPULMONARY ASPERGILLOSIS: A DIFFERENTIAL DIAGNOSIS IN SEVERE ASTHMA

Lorenzo Marra, Silvano Dragonieri, Giuseppina D'Alba, Ina Ali, Onofrio Resta.

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A 58 year old woman with a diagnosis of asthma, in therapy with ICS + LABA. Due to the need for frequent cycles of oral corticosteroids, she underwent a new medical examination. She referred cough, breathlessness and periodic episodes of coughing with purulent brownish sputum, cyclically treated with antibiotic therapy. On thoracic examination, she had widespread groans and wheeze. Spirometry showed a mild obstructive ventilatory defect (Tiff 68%, FEV1 70.6%) and bronchoreversibility test was positive. Blood tests were required for a more careful diagnostic assessment, revealing peripheral blood eosinophilia and elevated total serum IgE. Prick Test was positive for parietaria, dermatophagoides and Aspergillus. A chest TC showed ground glass and tree in bud in upper left lobe and central bronchiectasis (figure 1). The bronchoscopy with Broncho-Alveolar Lavage (BAL) was negative on cytological examination, but macroscopic evaluation of the collected material showed the presence of flocculent brown mucous, which led us to identify aspergillus. Other blood test showed elevated serum IgE and IgG for Aspergillus. Autoimmunity testing and tests for cystic fibrosis were negative. Looking into Rosenberg-Patterson diagnostic criteria for allergic bronchopulmonary aspergillosis (ABPA) we achieved a diagnosis. The patient also reported the presence at her home of moisture collection areas on the walls, with environmental isolation of Aspergillus Niger. After environmental remediation and two months of therapy with oral prednisolone and itraconazole, plus inhalator therapy, she has full control of symptoms, a normal clinical examination and functional test, normal blood test and the new chest TC no longer shows ground glass or tree in bud (figure 2). Although the real prevalence of ABPA in severe asthma in unknown, it is estimated that is around 6%, and sensitization to various fungi predisposes to an increased severity of bronchial asthma. When presented with asthma,

we should take into account this differential diagnosis.

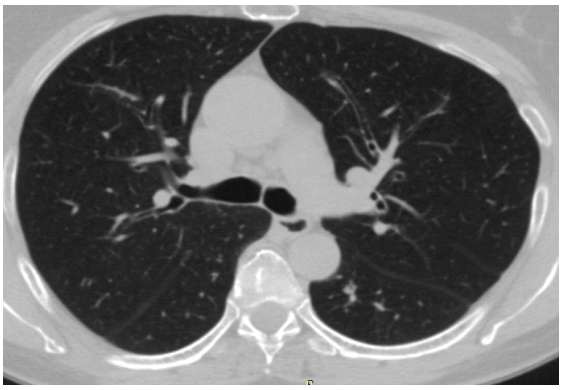

**Figure 1:** Chest TC showed ground glass and tree in bud in upper left lobe and then central bronchiectasis

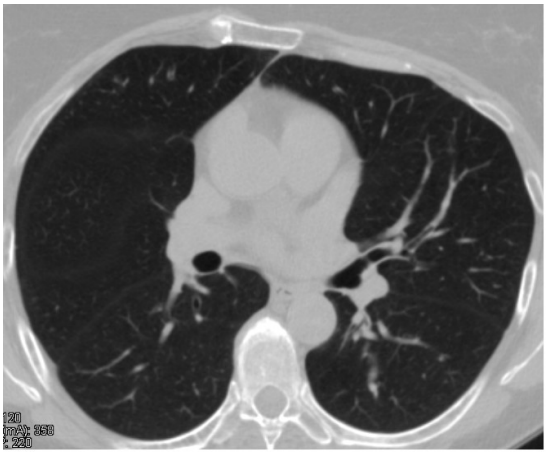

**Figure 2:** New chest TC no longer shows ground glass and tree in bud

####

## A CASE OF ACUTE PULMONARY EDEMA FOLLOWING LICORICE ABUSE

Vito Liotino, Vincenzo Nicola Valerio, Pierluigi Carratù, Maria Rosaria Vulpi, Onofrio Resta.

*Institute of Respiratory Diseases, Department of Surgical and Medical Sciences University of Bari*

Licorice has been known to cause hypertension, hypernatremia, hypokalemia through the mineralcorticoid effect of glycyrrhizic acid. 100 mg/day of glycyrrhizic acid is the lowest observed adverse effect level. P.T., a 36 year old previously healthy male, came to the ER after 48h of dyspnea, fatigue and leg edema. He was not taking any medication. Abuse of licorice during the previous 10 days (about 350 mg/day of glycyrrhizic acid) was the only variation of his usual diet. On physical examination he appeared in respiratory distress with inspiratory crackles on both lungs. His blood pressure was 150/90 mmHg. Laboratory investigations were remarkable for: potassium 3,6mmol/l, sodium 145mmol/L, NTproBNP 380pg/mL (normal < 125 pg/mL). Oxygen saturation in room air was

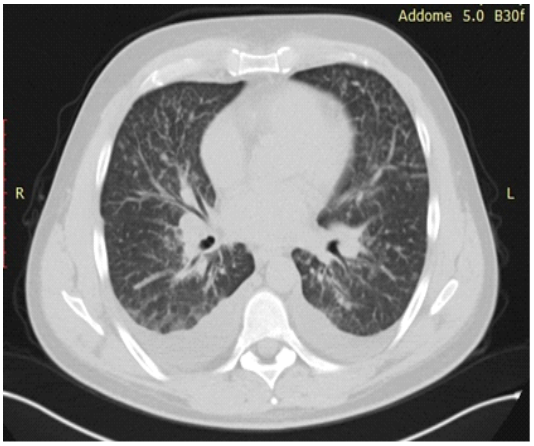


**Figure 1:** A CT scan with contrast was performed showing reticular diffuse interstitial thickening, bilateral pleural effusion, nodules and basal ground-glass areas.

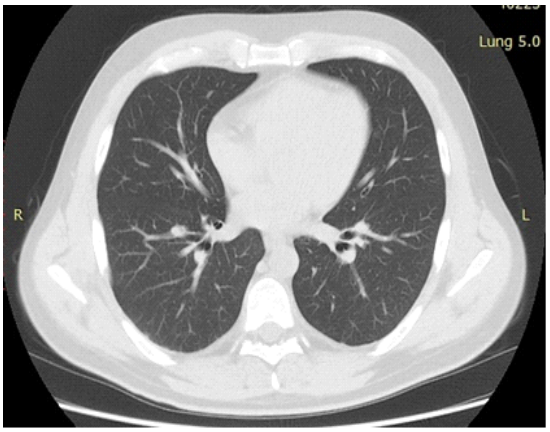


**Figure 2:** A lung CT scan was performed with complete resolution of radiographic findings.

86%. The radiograph of the chest showed diffuse interstitial thickening and right pleural effusion. Electrocardiogram and transthoracic echocardiogram were normal except for signs of systemic venous congestion. A CT scan with contrast was performed showing reticular diffuse interstitial thickening, bilateral pleural effusion, nodules and basal ground-glass areas; nothing to report in the abdomen (Figure 1). Occupational exposure to toxic substances was excluded. The urine drug test, oncomarkers, thyroid function, autoimmunity test and abdominal echography were all normal. A therapy with furosemide 40 mg/day, potassium canrenoate 50 mg/day and oxygen was established with rapid improvement of clinical condition. A new echocardiogram and a cardiac stress test were performed and did not reveal any pathology. A lung CT scan was performed with complete resolution of radiographic findings (Figure 2).On discharge he was normotensive, with normal potassium (4 mmol/L) and NTproBNP (50 pg/mL) levels. Abstaining from eating licorice, at 1 month follow-up, the patient was stably asymptomatic and normotensive. Despite the rarity of this event, pulmonary edema can result from abuse of licorice. This highlights the importance of investigating the eating habits of the patient in question carefully.

####

## IPAF CASE WITH FIRST PULMONARY MANIFESTATION, RESPONSIVE AT IMMUNOSUPPRESSIVE THERAPY

Ina Ali, Marco Patruno, Silvano Dragonieri, Giuseppe Cassano, Onofrio Resta.

*Institute of Respiratory Diseases, Department of Surgical and Medical Sciences University of Bari*

A case of a 65 year old man, caucasian, smoker, suffering from hypertension andhypothyroidism,evaluated in our clinic for appearance of hacking cough, worsening dyspnea, asthenia. Chest examination: crackles bilaterally. RCP and ESR increased, blood count normal. ChestX-ray showed diffuse interstitial thickening. Antibiotic therapy did not improve the symptoms. Chest HRCT showed crosslinking, bronchiectasis, and subpleural ground glass(Figure 1). Spirometry showed slight reduction of FEV1 and FVC. Moderate reduction in the DLCO. 6MWT: 425 m paths, no oxyhemoglobin desaturation. Transthoracic echocardiogram showed increased PAPs. Positive ANA (1/1280), antiENA Sm / RNP, Scl 70 antibodies antiENA. Multidisciplinary discussion: IPAF (UCTD with ILD) diagnosis and initiation of treatment with cyclophosphamide 100 mg / day and prednisone 25 mg / day. After 6 months of therapy chest HRCT showed resolution of the previous framework, modest thickening remains at thebases (Figure 2).

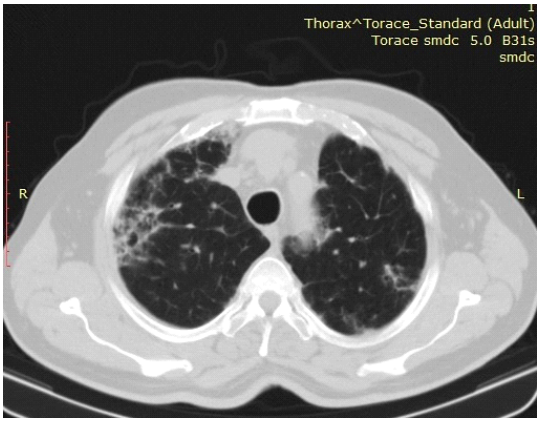


**Figure 1:** HRCT showed crosslinking, bronchiectasis, and subpleural ground glass

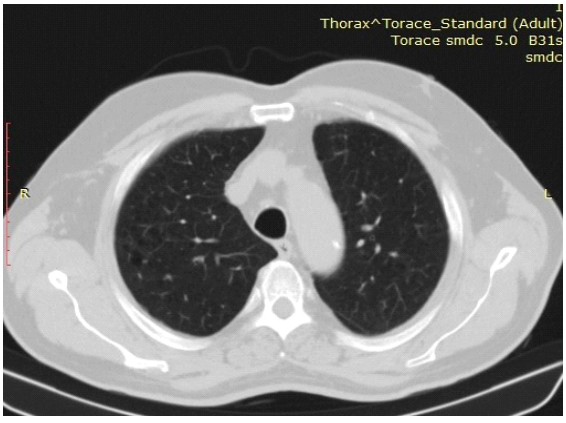


**Figure 2:** HRCT showed resolution of the previous framework, modest thickening remains at the bases.

6MWT: 600 m paths, no desaturation. Spirometry: FEV1, FVC and DLCO increased. Transthoracic echocardiogram showed PAPs reduction. We describe a case of IPAF thefirst manifestation of which is lung involvement with clinical, functional and radiological improvement after therapy.

####

## HIGH-RESOLUTION COMPUTED TOMOGRAPHY CRITERIA FOR UIP PATTERN.

Laura Gabrieli

*Section of Radiology - D.I.M. (Department of Interdisciplinay Medicine) - University of Bari.*

An 82 year old hypertensive diabetic woman reported to the emergency department, with syncope andheadinjury. She had a two week history of incessant cough, recently productive, withdyspnea and asthenia onminorexertion. At the emergency departments he was subjected to two skull Tc: negative for encephalic lesions. She performed lung scintigraphy, which excluded embolism, underwent cardiology consultation, echocardiogram (mild dilation of the limbs, mild tricuspid regurgitation) and chest x-ray (bilateral pulmonary reticulation). The patient was hospitalized and subjected to intravenous antibiotic therapy (Klacid and Rocefin) for ten days, on the basis of the following chest CT report: "Reticular abnormality, with subpleural and a basal predominance; traction bronchiectasis; increased volumetric lymphnodes (maximum 1.5 centimeters) in the lodges of Barety, sovracarenali and aorto-pulmonary window". After four days the patient was without fever. She was subjected to a new chest x-ray, which reported: "No observed changes compared

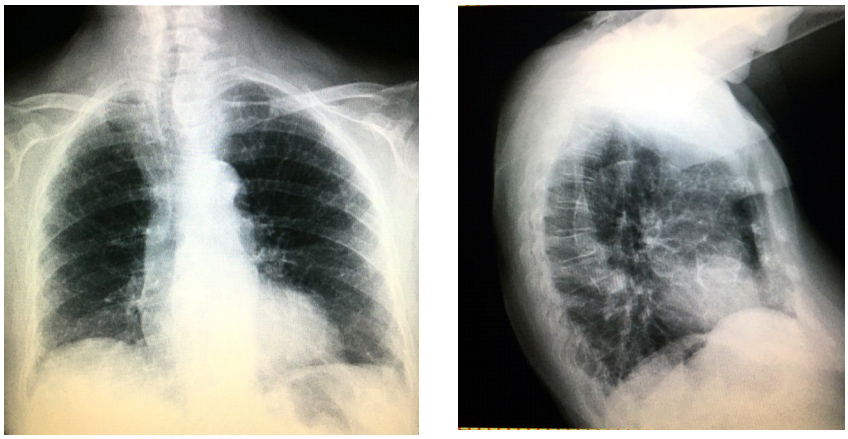

**Figure 1 – 2:** Chest X-Ray

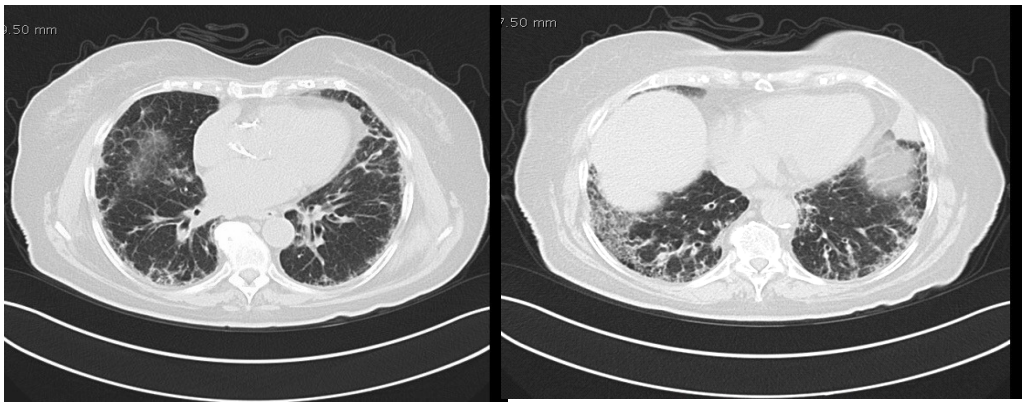

**Figure 3 – 4:** Chest CT

to the previous chest x-ray, characterized by bilateral pulmonary reticulation, highly suspicious of interstitial fibrosing pneumonia."Other tests were needed in pneumology. The patient's medical history was significant for the discovery of a CT pattern of DEFINITE or POSSIBLE UIP (Usual Interstitial Pneumonia). The different morphological features of UIP can be analyzed with high quality CT: thin section reconstruction (< 1.5 mm), reconstruction algorithm and suspended deep inspiration. Systematic approach to CT involves: evaluation of image quality; precise description of specific disease features; using standard terminology; consideration of disease distribution; is it a fibrotic DILD (Diffuse Interstitial Lung Disease) or non-fibrotic DILD?
Considering CT features of fibrosis (Honeycombing; Traction bronchiectasis; Volume loss) and evaluating these features, clinicians should ask whether the patient presents a fibrotic DIL Dora definite UIP? If not, is it possible UIP? What are the alternatives? An Official ATS/ERS/JRS/ALAT statement, reports the following evidence-based guidelines for diagnosis and management of Idiopatic Pulmonary Fibrosis:
FEATURES OF DEFINITE UIP: Subpleural, basal predominant; reticular abnormality; honeycombing with or without traction bronchiectasis; absence of features listed as inconsistent with UIP pattern.
FEATURES OF POSSIBLE UIP: subpleural, basal predominant; reticular abnormality; absence of features listed as inconsistent with UIP pattern.
Therefore, the presence of honey combing is critical for making a definite diagnosis. Honeycombing is manifested on HRCT as clustered cystic airspaces, typically of comparable diameters in the order of 3–10 mm but occasionally as large as 2.5 cm. It is usually subpleural and is characterized by well-defined walls. The alternatives are: DIP (Desquamative Interstitial Pneumonia) or NSIP (Non-Specific Interstitial Pneumonia). Any of the following seven features are inconsistent with the UIP pattern: upper or mid-lung predominant; peribronchovascular predominant; extensive ground glass abnormality (extent reticular abnormality); profuse micronodules (bilateral, predominantly upper lobes); discrete cysts (multiple, bilateral, away from areas of honeycombing); diffuse mosaic attenuation/air-trapping (bilateral, in three or more lobes); consolidation in bronchopulmonary segment (s)/lobe(s).
Final Diagnosis was a possible UIP, because the features of the chest CT were reticular abnormality, with subpleural and a basal predominance and traction bronchiectasis, without honeycombing.

####

## BIOMARKERS AND SEVERE ASTHMA

Giulia Scioscia, Marilena Lo Storto, Giovanna Elisiana Carpagano, Grazia Cotugno, Maria Pia Foschino Barbaro

*Institute of Respiratory Diseases, Department of Surgical and Medical Sciences University of Foggia*

Severe asthma is a heterogeneous disease, affecting among 5–10 % of asthmatic patients. Despite high-dose therapy, a large percentage of patients are not fully controlled and havea poor quality of life.
Currently, studies are beginning to identify different "phenotypes" defined by clinical manifestations, pathophysiological mechanisms and biomarkers. An ideal biomarker is easy to measure, not invasive nor expensive, and can be used to identify either clinical or treatment response phenotypes, evaluate changes in disease activity, or confirm a diagnosis. A 61 year-old woman with moderate persistent bronchial asthma, was evaluated at the clinic dedicated to the diagnosis and treatment of asthma of the Department of Respiratory Diseases of Foggia. She reported acute dyspnea, despite therapy with high dose of ICS/LABA ,antileukotriene and, when needed, oral steroids. Skin-prick test was positive for dermatophagoides and cat epithelium. Laboratory findings documented low Asthma Control Test (ACT 10); high total IgE level (465 IU / ml); moderate-severe obstruction on functional test, high blood and induced sputum count of eosinophils (8.1% - 13% respectively), abnormal bronchial fraction of exhaled nitric oxide

(FeNO 69 ppb) and exhaled breath temperature (EBT 34.8 °C).
On the basis of these results, we proceed to the step-up therapy with the addition of anti-IgE monoclonal antibody (Omalizumab). After four months, we observed an improvement of ACT (12 ppt) and functional parameters, reduction of blood and sputum eosinophils count (4,2% - 8% respectivtely), FeNO (50 ppt) and EBT (33,8°C). Based on these results, 4 months after beginning Omalizumab administration, it was possible to highlight the lack of efficacy of this therapy with the help of non-invasive biomarkers.

####

## UIP VS FIBROTIC NSIP: UTILIITY OF CRIOBIOPSY IN THE DIFFERENTIAL DIAGNOSIS

Giulia Patricelli, Donato Lacedonia, Giovanna Elisiana Carpagnano, Incoronata Caccavo, Maria Pia Foschino Barbaro.

*Institute of Respiratory Diseases, Department of Surgical and Medical Sciences University of Foggia*

A correct therapeutic approach to interstitial lung diseases requires, first of all, a specific diagnosis. Sometimes the complexity of clinical and functional framework and the non-specificity of the radiological pattern, do not allow for a definitive diagnosis. In these casesm a histologic analysis of the lung parenchyma is required.
A 68 year old man, who had never smoked, was evaluated at the clinic dedicated to the diagnosis and treatment of Rare Lung Disease at the Department of Respiratory Diseases in Foggia. He reported dry, persistent cough and dyspnea at the slightest effort. In anamnestic history no occupational exposure to fine particles, chemicals or other toxic inhalants was revealed. On clinical examination, he presented dry, inspiratory bibasilar "velcro-like" crackles on auscultation and showed digital clubbing. For a better clinical diagnosis, he performed chest X-ray (prominent reticular interstitial markings near the lung bases), pulmonary function tests (moderate restrictive defect), 6mwt (good exercise tolerance in the absence of oxygen desaturation) and autoimmune panel (negative). A consecutive HRCT (Figure 1) showed radiological findings compatible with "Possible UIP" or, alternatively, fibrotic NSIP. In consideration of his good general medical condition, the patient performed criobiopsy which confirmed UIP histological pattern with concomitant mucosal adenocarcinoma.
Histological evaluation of bioptic samples collected by endobronchial methods, is most helpful in the differential diagnosis of fibrotic diffuse parenchymal lung diseases when the clinical-radiological pattern is non-diagnostic. Transbronchial lung cryobiopsy (TBLC) has been shown to be useful for obtaining large and well-preserved biopsies of lung parenchyma.
The need to find biomarkers useful in monitoring treatment response is evident in clinical practice, however, their discovery is made difficult by the huge number of proteins involved in severe asthma pathogenesis.

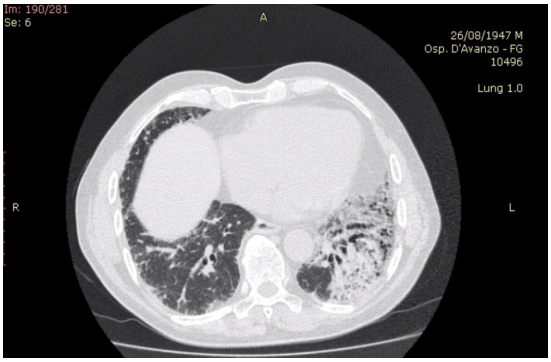


**Figure 1:** HRCT: radiological findings compatible with "Possible UIP" or, alternatively, fibrotic NSIP.

####

## ASYSTOLE RELATED TO OBSTRUCTIVE SLEEP APNEA (OSAS). EFFECT OF THE POSITIVE PRESSURE DEVICE (CPAP)

Valerio Saliani, Roberto Sabato, Giulia Patricelli, Maria Pia Foschino Barbaro.

*Department of Medical and Surgical Sci-*

*ences, Section of Respiratory Diseases - University of Foggia*

We report the case of a patient with severe OSAS, complicated by nocturnal respiratory failure, with cardiovascular and metabolic comorbidity and COPD that showed repeated OSA-related asystoles, who then started to CPAP therapy. A 54 year old (M), smoker, obese (BMI 32 kg / m2), was evaluated with nightly full cardio-respiratory monitoring (MC-R) 12-channel for high risk of OSAS. Clinical history of sub-airway obstruction, ischemic-hypertensive cardiomyopathy, stenosis of large coronary trunks, chronic obstructive pulmonary disease, type 2 diabetes and multinodular goiter in euthyroid. P.A. 140/80 mmHg. The ECG in vigil, showed sinus rhythm at 66 bpm, normal.
The 2D echocardiogram showed a left ventricle with dimensions on the higher limits, moderate hypertrophy, normal contractility (E.F. 60% ). Dilated left atrium; right sections within normal limits. Fibrotic valves. PAPs 40 mmHg. The diagnostic CR-monitoring showed severe OSAS with 85% of obstructive events (co -presence of COPD - Overlap syndrome) and arrhythmia related to obstructive events (several episodes of bradycardia associated with inconstant asystoles, lasting up to 10.9 ") (Figure 1). The

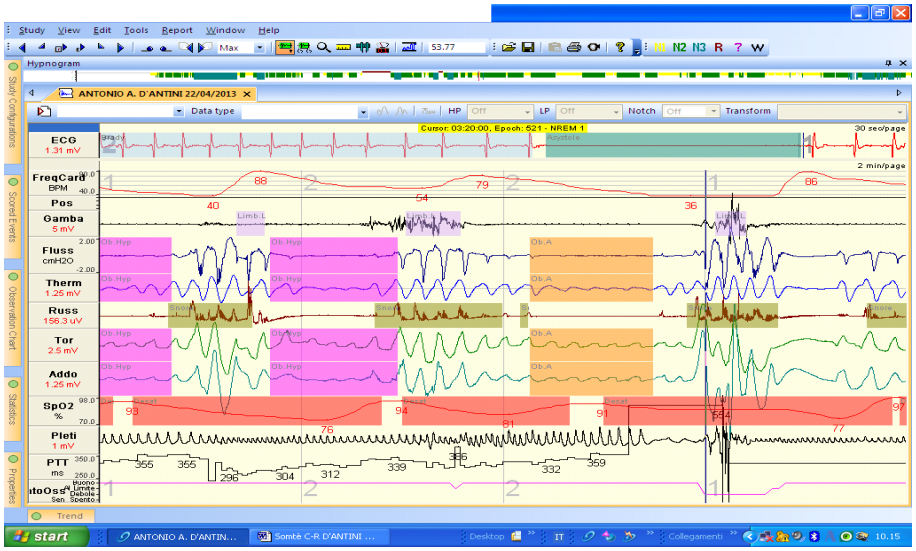


**Figure 1:** Spontaneous breathing (2 min)

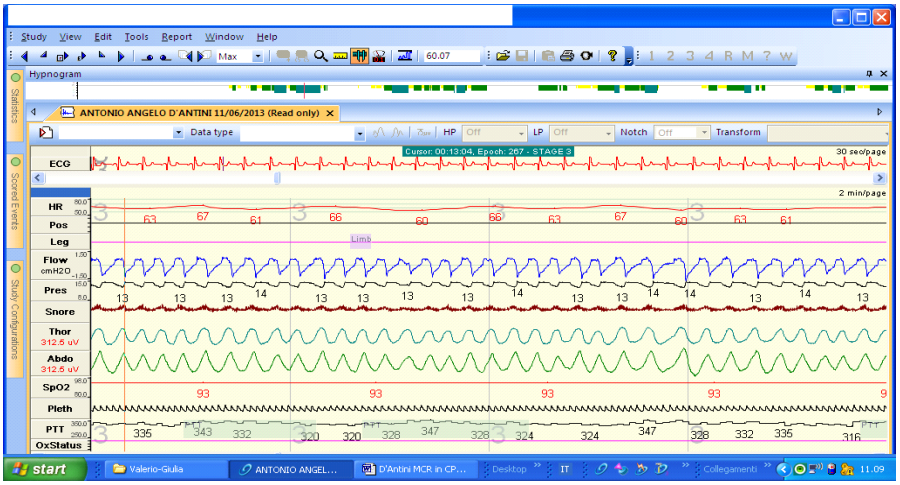


**Figure 2:** CPAP (2 min)

Holter-ECG showed sinus rhythm between 42 and 102 bpm, normal interventricular conduction; numerous night breaks (total 113, max 9220 msec); absence of ischemic ST-T changes.
The patient was fitted with the CPAP device and subjected to complete MC-R for therapeutic efficacy testing. With the correction of obstructive events, episodes of asystole disappeared (Figure 2).
The use, during sleep, of CPAP device corrects OSA-related disorders and may prevent cardiovascular complications. In the case reported, CPAP impacts positively on the serious nocturnal disturbances of cardiac rhythm in a patient with severe OSAS. It is possible to assume that the autonomic and hemodynamic changes, myocardial ischemia, and the structural and functional alterations of the atrial and ventricular chambers associated with the repetitiveness of nocturnal apneic episodes, are responsible for the triggering of cardiac arrhythmias in OSAS patients.

####

## PULMONARY CYSTS: A RARE CASE OF FAMILIAR DISEASE

Giuseppe Antonio Palmiotti[1], Donato Lacedonia[1], Giovanna Elisiana Carpagnano[1], Giovanna D'Andrea[2], Carlo Florio[3], Maurizio Margaglione[2], Maria Pia Foschino Barbaro[1].

*1 Institute of Respiratory Diseases, Department of Surgical and Medical Sciences University of Foggia*
*2 Medical Genetics, Department of Clinical and Experimental Medicine, University of Foggia, Italy*
*3 Oncologic Institute of Bari, Italy*

We describe the case of a 68 year old male patient, who had never smoked, suffering from hypertension in pharmacological treatment and nasal polyposis treated surgically, who came to our attention about two years ago for cough and shortness of breath. He also has a family history of spontaneous pneumothorax. He performed an HRTC scan that showed the presence of several cysts, spread ubiquitously, but most frequently at the lung bases (Figure1). Furthermore, examining one of his two sisters, we found the presence of fibrofolliculomas on her face that were absent in our patient (figure2). We also recuperated the HRCT images of the other sister that confirmed the presence of lung cysts. We then sent a blood sample from our patient and his sister for genetic testing to our genetics laboratory which confirmed a heterozygous nonsense mutation R477X of FLCN gene already described in literature. We diagnosed Birth-Hogg-Dube' syndrome. Birt–Hogg–Dubé syndrome (BHD) is an autosomal, dominantly inherited, monogenic condition characterised by multiple fibrofolliculomas, pulmonary cysts, pneumothorax, renal cysts and tumors due to a mutation of a gene, folliculin (FLCN), which is located on 17p11.2 . It is considered a rare disease with only roughly 600 reported families worldwide. Due to the lack of awareness, there is commonly a delay in

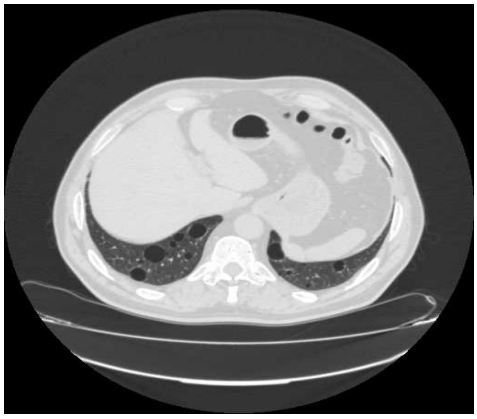

**Figure 1:** typical round cysts of Birth-Hogg-Dubè

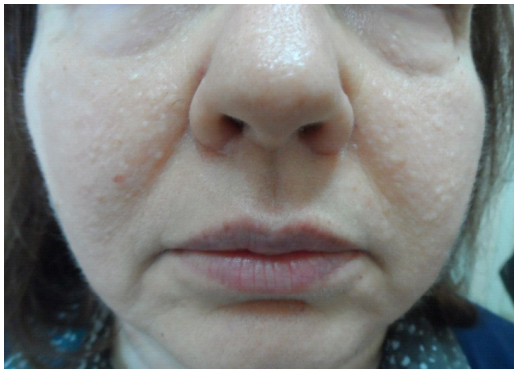

**Figure 2:** Fibrofolliculomas

the pulmonary diagnosis of BHD and patients are frequently mislabeled as having chronic obstructive lung disease, emphysema or common bulla and blebs. The purpose of this case report is to focus attention on a rare disease that clinicians should be aware of, especially pulmonologists, since the differential diagnosis concerns several respiratory diseases. We also emphasize the importance of a center specializing in rare diseases with a multidisciplinary team dedicated to them including genetists. This would help to avoid diagnostic delay and inappropriate therapy.

####

## A CASE OF MARIJUANA ABUSE PRESENTING AS "VANISHING LUNG SYNDROME"

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Gennaro, a 42-year old male, with a family history of respiratory diseases, complained of exertional dyspnea during daily activities. Over the last 30 years, he has smoked 20 cigarettes and two or three joints of marijuana per day. PFT showed an obstructive pattern and pulmonary hyperinflation, with an increase in Total Lung Capacity (TLC), Functional Residual Capacity (FRC) and Residual Volume (RV). Arterial blood gas analysis revealed mild hypoxemia with normal acid-base balance. A high resolution CT scan of the thorax showed multiple large emphysematous bullae distributed peripherally in both upper lobes, slightly prevalent in left upper lobe; the lower lobes showed centrolobular and paraseptal emphysema. Pulmonary perfusion scintigraphy documented a large perfusion defect in both upper lobes, involving many segments. A normal α1-antitrypsin level was detected. The conclusive diag-

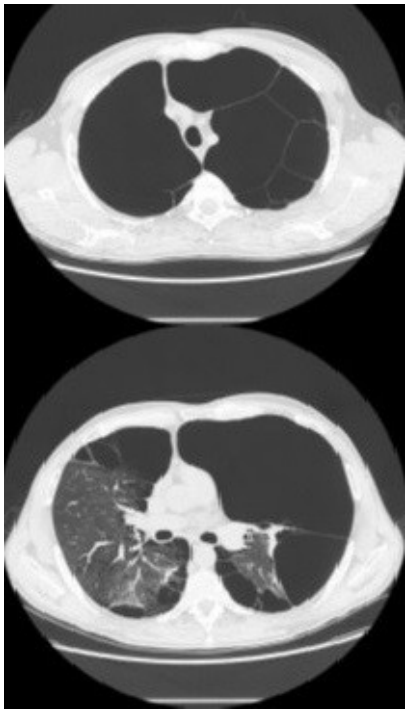

**Figure 1:** Chest CT scan cross-sections

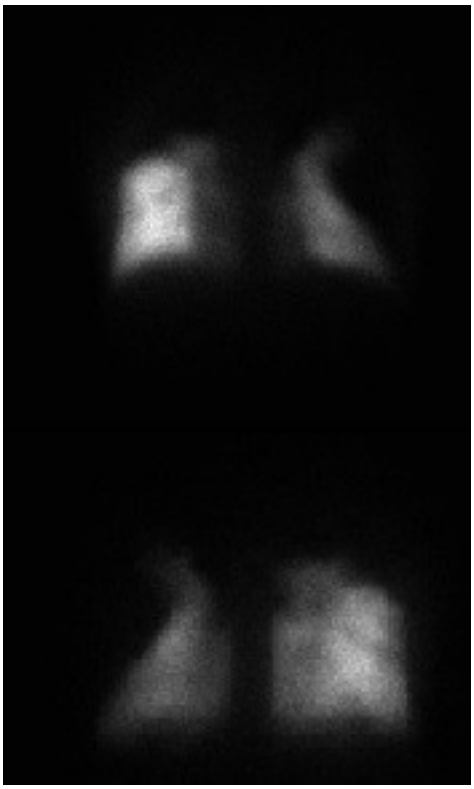

**Figure 2:** Perfusional scintigraphy bilateral bullae severe perfusion defect involving mainly the upper lobes

nosis was "Vanishing lung syndrome" (VLS) due to marijuana abuse. Cigarette and marijuana smoking were forbidden to the patient; LAMA and LABA were prescribed. Finally, the patient was added to the transplant list. VLS, also called giant bullous emphysema, is a progressive bullous disease characterized by large bullae that involve at least one-third of one or both hemithorax. Marijuana abuse has shown an increasing trend worldwide in the last decade. The principal physiological impairment with long-term cannabis smoking is on large airway function; in contrast, cannabis smoking was uncommonly associated with macroscopic emphysema.

####

## CHEST CT IN THE DIFFERENTIAL DIAGNOSIS OF PNEUMONIA: A CASE REPORT

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The diagnosis of pneumonia can be a "challenge" in elderly patients because symptoms and signs are often not specific. Chest X-ray is usually sufficient to confirm the diagnosis. Chest CT is reserved in patients with atypical radiographic pattern or those are not responding to treatments in order to discard other diagnostic alternatives. We report a case of a 77-year old man, non-smoker, who presented a tour outpatient clinic with a history of fatigue, mild fever, dyspnea, and dry cough lasting for 2 months despite a cycle of antibiotic therapy with Ceftriaxone. Physical examination was negative. Functional tests showed a mild restrictive ventilatory defect, a moderate reduction of lung CO transfer, and a mild hypoxemia. Laboratory assays revealed a mild neutrophilic leucocytosis, increased CRP levels, and negative urinary antigens tests for pneumococco and legionella. The tuberculin skin test was negative. At chest X-Ray, inhomogeneous bibasilar opacities were detected. On clinical suspicion of pneumonia, antibiotic therapy with levofloxacina and piperacillina-tazobactamwas started. After 2 weeks, both clinical and radiological abnormalities remained unchanged and a chest CT scan was performed. CT findings were suggestive of organized pneumonia (OP): patchy areas of consolidation with subpleural (Figure 1

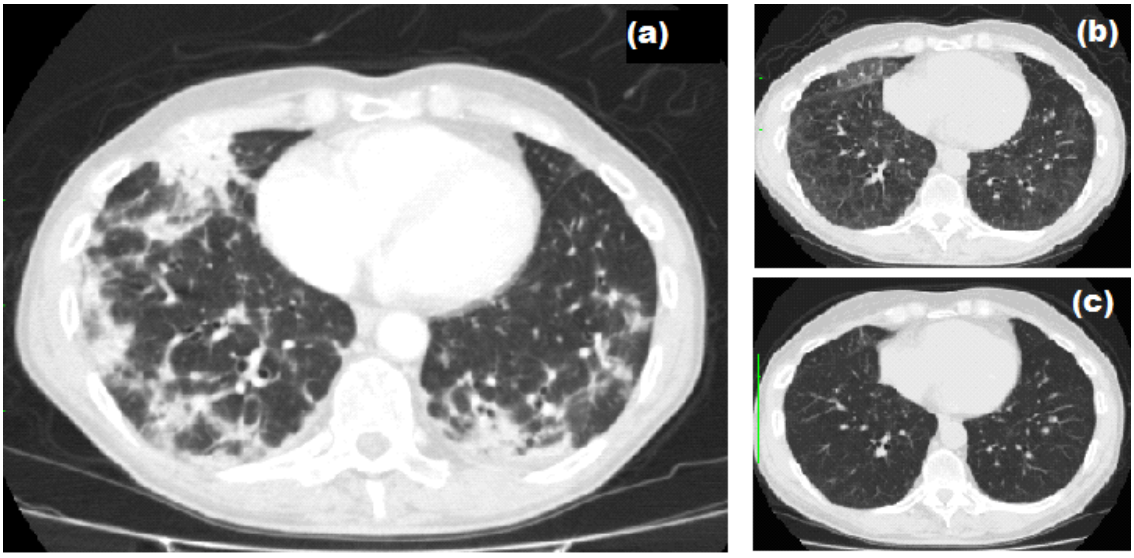


**Figure1:** (a) CT scan shows patchy areas of consolidation and ground glass opacities with subpleural and bilateral distribution in the lower lobes. High-resolution CT images performed after three (b) and six (c) months of corticosteroid treatmentshow the resolution of the consolidations.

a) and peribronchial distribution, ground glass opacities, peribronchial nodules, "band-like" pattern, and "reversed halo sign" (Figure2). Serum autoimmune markers were negative. Treatment with an initial dose of prednisone 50 mg/die gradually tapered over a six-month period determined an improvement of symptoms and a resolution of chest CT findings (Figure 1 b,c).

The differential diagnosis between community acquired pneumonia and Cryptogenic Organizing Pneumonia (COP) can be difficult. Furthermore, infectious pneumonia can be the cause of OP. Despite requiring a histological exam for definitive diagnosis, high resolution CT (HRTC) is highly accurate for OP diagnosis in the presence of the typical clinical features.

####

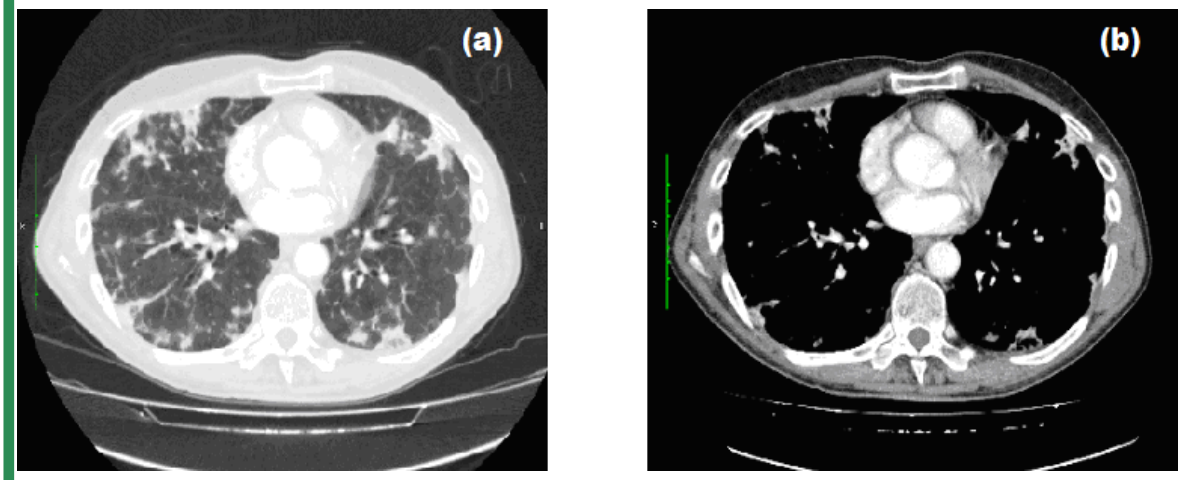


**Figure 2:** CT scan shows peripheral consolidation with reversed halo sign (RHS) in the left lower lobe (arrow) in lung (a) and mediastinal window settings (b).

Special Issue

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## ABSTRACT BOOK
## ONSP DAYS 2016—13th ITALIAN MEETING OF NATIONAL OBSERVATORY FOR TRAINEES AND YOUNG PAEDIATRICIANS

**Bologna (Italy) 5 - 8 October 2016**

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## A 7-YEAR-OLD GIRL WITH AN UNEXPECTED VAGINAL BLEEDING

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A 7-year-old girl was referred to our clinic with vaginal bleeding started 1 week before. Parents reported rapidly progressive bilateral breast budding, spread of pubic hair and growth acceleration. On examination we found a Tanner B4PH2. No axillar hair; height of 144 cm (>95°p) and weight of 42.8 kg (>95° p). No family history of precocious puberty. The left hand wrist X-ray showed a bone age of 10 years according to Greulich and Pyle. Neurological and general examination was normal. The hormonal assay revealed: estradiol 117.6 pmol/L, while basal and stimulated levels of LH and FSH were low. Tumor markers were negative. An abdominal ultrasound (US) documented a post-pubertal uterus with prominent endometrium and 3×2 cm mass in her left ovary, confirmed by pelvic MRI. A chest CT ruled out possible pathologic lymph nodes or masses. The patient underwent laparoscopic oophorectomy with a diagnosis of granulosa cell tumor (GCTs), juvenile type. The follow-up, 4 years later, showed no tumor recurrence/metastasis.
Precocious puberty in girls is defined by the development of sexual characteristics before the age of eight years. It is often secondary to a central idiopathic activation of the hypothalamic-pituitary-ovarian axis (central precocious puberty), otherwise a peripheral cause must be excluded. GCTs are the most common ovarian tumors responsible for this, causing estrogenic production. 50% are unilateral, with excellent prognosis after surgery.

####

## THE CONCURRENCE OF NEPHROTIC SYNDROME AND ACUTE GASTROENTERITIS IN A CHILD: AN UNCOMMON CAUSE OF OLIGO-ANURIA

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Nephrotic syndrome (NS) can be associated with acute kidney injury (AKI) and oliguria can be the presenting symptom of this.
An 8-year-old boy affected by steroid- and cyclosporine-dependent NS presented with both NS relapse for 2 days and acute gastroenteritis (AGE) with vomiting and diarrhea for 4 days. At AGE onset creatinine was 0.5mg/dL. He had been taking prednisone (60mg/m$^2$/day) since the NS relapse. He showed drowsiness, tachycardia (118beats/min), cold hands and feet, refill time of about 2 seconds and low-quantity micturition. He gained 0.8 kg in absence of peripheral oedema. Blood pressure was 90/50mmHg. Blood tests showed creatinine 4.2mg/dL, Na 140mEq/L, K 6.2mEq/L, urea 155mg/dL, albumin 1.9g/dL. As NS- and AGE-related hypovolaemia was evident, we started i.v. fluid administration (NaCl 0.9% 20mL/Kg in 1h) followed by 20% albumin i.v. solution (2.5mL/kg in 3h) and then i.v. furosemide (1mg/kg). Despite our treatment, 6h after admission he had only passed 66 mL (0.22mL/kg/h) of urine and levels of creatinine, potassium and urea were 4.6mg/dL, 6.5mEq/L and 260mg/dL, respectively. Despite hypovolaemia correction, he required dialysis. Conclusions: in children with both NS and AGE an edematous state can be misleading due to presenting paradoxical weight gain so clinicians must remain aware of the possible hypovolaemic state. Fluid administration without albumin infusion is not a correct treatment and i.v. NaCl 0.9% administration followed by i.v. 20% albumin and then furosemide should be the therapeutic option.

####

## A FIXED POLYPNEA

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XY, term baby, 3200g, 7-10 APGAR score. Family history: vegan mother; unexplained maternal uncle death in neonatal period. At thirty hours of life: irritability, pallor, fixed polypnea. Congenital heart defect (CHD), infective, respiratory and central etiology were excluded; unique clues: worsening respiratory alcalosis (RA) (pH 7.59, pCO2 15 mmHg), high NH4+ level= 270 µmol/L. Suspecting a metabolic disease, enteral nutrition was replaced by high dose glucose, L-arginine and Na-benzoate infusion. Despite hemodiafiltration, the baby died 7 days later. Ornithine transcarbamylase deficiency (OTCD) was diagnosed.
OTCD is the most common urea cycle defect (UCD), X-linked-inheritance. Males with OTCD are normal at birth, in a few hours they develop hyperammonemic encephalopathy, which explains hyperventilation and RA. In event of suspected OTCD, the main therapeutic goal is to lower blood NH4+ to ≤200 µmol/L. The diagnosis is primarily genetic. The prognosis mainly depends on the duration of high a NH4+ level, neither the level nor the seizures themselves [1,2,3].
Pediadvisor: be highly suspicious of UCD in a previously well newborn with a very fast deterioration; before lumbar puncture, look at NH4+ levels; be aware of a history of unexplained death in newborn males in the first week of life associated to a vegan mother; once CHD or sepsis is excluded, consider a "respiratory alcalosis in an encephalopathic individual who is hyperventilating, as pathognomonic of UCD"[4].

####

## HYPERLEUCOCITOSIS: A PREDICTOR OF SEVERE B. PERTUSSIS DISEASE

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We report a case of a 2-month-old infant, non vaccinated, with hyperleucocitosis and *B. pertussis* infection. The infant was admitted to the ED after 15 days of persistent cough, post-tussive vomiting and respiratory distress. During the observation she presented with several paroxysms of cough with cyanosis and apnea. Her peripheral white cell blood count was 56000 cells/microL and 66% were lymphocytes. A chest X-ray showed hilar infiltration and a diffused granular pattern with no signs of consolidation. The diagnosis of pertussis was confirmed by a positive culture and PCR assay of the nasopharyngeal aspirates. She was treated with Azithromycin oral therapy for 5 days and with hyperhydration (150cc/Kg) for 3 days until the blood count progressively reduced.
The predominant nonspecific laboratory indication of *B. pertussis* infection is a leukocytosis resulting from lymphocytosis. Marked leukocytosis (eg, >60,000 cells/microL) has been associated with increased pertussis severity, and has a major role in the cardiopulmonary compromise.
In infants, the WBC and lymphocyte count are directly correlated with disease severity, the mortality rate in infants is approximately 1%. Hyperhydration should be considered for all patients who present hyperleucocytosis. Exchange transfusion is indicated if the WBC is >100000 cells/microL due to pertussis related pneumonia, cardiogenic shock or neurological involvement.

####

## HYPOGLOSSAL NERVE PALSY AS A COMPLICATION OF METHICILLIN-RESISTANT STAPHYLOCOCCUS AUREUS (MRSA) PARAPHARYNGEAL ABSCESS IN A NEWBORN

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*[2] Neonatal Intensive Care Unit, Puericulture Institute and Neonatal Section, University of Cagliari, Italy*

Deep neck abscess (DNA) is a rare complication of upper airway bacterial infections and commonly occurs in infants; however, it is very rare in neonates. Airway obstruction and mediastinitis represent the principal complications in this period. Hypoglossal nerve palsy has been described in only 3 pediatric cases but never in neonates.
O. is a 27-day-old male neonate who arrived at the Emergency Room after 2 days of rhinitis and upper airway inflammation. Examination revealed left acute otitis media and rhinorrhea without fever. Investigation revealed leukocytosis, slightly increased CRP but negative PCT. We started intravenous (iv) antibiotic therapy, but 2 days later left laterocervical lymphadenopathy appeared. Clinical conditions of the newborn progressively deteriorated; he developed left hemytongue swelling with deviation of the tongue to the healthy side during crying. Computed tomography showed a DNA complicated by ipsilateral hypoglossal nerve palsy. Nasopharyngeal cultures were positive for MRSA and the baby started teicoplanin iv. Despite this, his conditions worsened: respiratory distress, desaturation and apnea crisis with retropulsion of head appeared. The baby was intubated and the DNA was drained with gradual improvement in mobility of the tongue. DNAs have an insidious clinical presentation in neonatal age, often without fever, that quickly progresses to airway obstruction. In this age, MRSA which does not respond to common antibiotic therapy should also be considered.

####

## RESPIRATORY DISTRESS UNRESPONSIVE TO SUPPLEMENTARY OXYGEN

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Total anomalous pulmonary venous return (TAPVR) is a rare congenital heart disease in which the four pulmonary veins, instead of connecting with the left atrium, drain through a common collector vein into systemic veins (superior vena cava) or directly into the right atrium. TAPVR is classified into different types, based on drainage site of pulmonary veins into the heart: supracardiac, cardiac, infracardiac and mixed.
We describe the case of a 2-day-old male neonate born after an uncomplicated pregnancy. The baby was admitted for pallor, poor perfusion, respiratory distress with tachypnea (100 breaths per minute) and cyanosis (SaO2 79%) unresponsive to supplementary oxygen. The echocardiogram revealed that the pulmonary veins drained into the right atrium through a collector behind the same. The chest X-ray showed a 'snowman' appearance with initial signs of pulmonary hyperflow. A diagnosis of supracardiac TAPVR with restrictive atrial septal defect (ASD) was made and the baby underwent heart surgery which consisted of an anastomosis between collector vein and left atrium and the closing of ASD and vertical vein.
TAPVR requires emergency heart surgery and early detection is very important. Therefore, it must always be considered in the differential diagnosis of neonates with respiratory distress and cyanosis unresponsive to oxygen therapy.

####

## HEART MURMUR IN NEWBORN: A CASE OF TETRALOGY OF FALLOT

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The Tetralogy of Fallot is one of the most common congenital heart disorders; it comprises right ventricular (RV) outflow tract obstruction (RVOTO), ventricular septal defect, aorta dextroposition, and RV hypertrophy. Physical findings at birth could be cyanosis of the lips and nail bed or difficulty with feeding, but a systolic murmur or thrill on left sternal border could be the only sign.
R. is a female newborn a few hours old, with uneventful pregnancy and delivery. She arrives at our observation because she has had one episode of desaturation (SaO2 88-92%) without cyanosis associated to a systolic murmur over the pulmonic area. The chest X-ray finds RV hypertrophy with diminished vascularity in the lungs and diminished prominence of the pulmonary arteries and the paediatric cardiologist confirms, with echography, the presence of the four signs of Tetralogy; the pulmonar artery is only slightly hypoplasic and the stenosis is both dynamic and fixed. The baby is well, so the surgery is planned once 6 Kg weight is achieved. The other examinations are normal, including the genetic studies.
Occasionally, it is possible to diagnose some cases of congenital heart disease in prenatal echography, before the onset of clinical symptoms or dramatic situations. It is important to listen carefully to every newborn's heart because often there are no other signs nor physical findings. In our case, the heart murmur was the only indication.

####

## A CHRONIC COUGH TO BE DISCOVERED

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Ioana is a 14-year-old girl that comes to our attention for chronic cough, which began at 7 years of age. She reports dry cough, shortness of breath after exertion and chest tightness.
The first tests she undergoes are blood exams (normal CBC, negative RCP), a first level screening for immunological study, a negative quantiferon, sweat test, nasal brushing and EKG. Chest X-rays are normal. Given her symptoms and age, the most plausible causes to exclude were asthma and GER. Her spirometry indicates no obstructive pattern, Tiffenau index is 0.97 and reversibility test 6%. A pH-metry is normal. Given the persistence of symptoms a chest CT scan is required that shows bronchiectasis and, in the left lower lobe, bronchial segmental branches that appear dysmorphic, alternating dilated and stenotic tracts. Upper airway endoscopy shows presence of granulation tissue obstructing the ostium at the bottom of the left bronchus. Necrotic vegetal tissue is found and removed.
7 years before, Ioana had inhaled a foreign body (FB) that was responsible for her respiratory symptoms and that made it difficult to diagnose her. Inhalation of FB is more frequent in early childhood years and in males than in females. They are mostly vegetables, nuts and seeds. The diagnosis and therapy can be made only with endoscopy. Complications include erosions, granulation, impact of FB, lung collapse and bronchiectasis.

####

## A CASE OF DIPLOPIA IN PATIENT WITH AUTOIMMUNE ENCEPHALOPATHY

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Acute disseminated encephalomyelitis (ADEM) is an inflammatory demyelinating disease of the central nervous system. It involves multifocal areas of the white matter, rarely the gray matter and spinal cord, and mainly affects children, 1-2 weeks after infections or rarely after vaccinations. We describe an 11-year-old girl admitted at the Unit of Clinic Pediatrics of the University of Catania because following a 3-week history of gastroenteritis, she had headache and fever, blurred vision, diplopia, scotoma and dysgraphia. She showed impairment of left upper limb strength and vivid patellar reflex.Laboratory tests were normal with type 2 IgG bands at cerebrospinal fluid. At the visual field test, hemianopsia was present in both eyes with reduced visual acuity of the left eye. Visual evoked potential test showed a reduction of the amplitude of P100 wave, normal latency. Brain magnetic exam imaging showed lesions in pons, middle left cerebral peduncle, basal ganglia, callosal corpum, optic nerve and spinal cord. Therapy with intravenous high-dose methylprednisolone (30mg/kg/day for 5 days), followed by prednisone (1mg/kg/day tapered for 6 weeks) resulted in a marked clinical improvement. Clinical, anamnestic and instrumental data were indicative of ADEM. ADEM is a cause of ophthalmological symptoms. Patients, such as this, treated with high-dose intravenous steroids followed by an oral steroid taper and/or intravenous immunoglobulin have good visual outcomes.

####

## SYSTOLIC HEART MURMUR AND SEVERE HYPOTONIA AS PRESENTING SIGNS OF A METABOLIC DISORDER

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We report on a 7-month-old male, admitted to the Unit of Pediatrics of the University of Catania because of generalized hypotonia and heart murmur. The physical and neurological examination confirmed a severe systolic murmur and generalized hypotonia. Laboratory tests showed increased levels of creatine phosphokinase and transaminases, while signs of biventricular overload with high voltage potential were present at ECG. At heart ultrasound, a hypertrophic cardiomyopathy was found, with severely increased left ventricular mass index (LVMI) of 215 gm/m$^2$. Suspecting Pompe disease we performed DBS test for acid alpha-glucosidase (GAA), which was totally absent. The molecular analysis showed a new heterozygous point mutation of the *GAA* c.693-2 A>C (IVS3-2 A>C) and the two exonic variants c.368G>A p.(Gly123Glu) and c.1288G>A p.(Glu430Lys).
Pompe disease is a rare, autosomal recessive lysosomal disorder caused by deficiency of GAA and characterized by lysosomal glycogen accumulation, mainly in muscular tissue. The response to enzyme replacement therapy with recombinant human GAA varies among patients. In the first months, our patient was treated with the recommended dose of 20 mg/kg biweekly, but his cardiac hypertrophy worsened (LVMI 230 gm/m$^2$) and he showed signs of respiratory insufficiency. Following the experience of Van Gelder at al, the patient was treated with with a higher dose (40 mg/kg/week), with noticeable improvement (LVMI 166 g/m$^2$).

####

## A RECURRENT DIPLOPIA

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A 14-year-old boy complained of diplopia followed by headache twice a week over the previous 6 months. His medical history was otherwise unremarkable. Neither vomiting nor other neurological signs were present. General physical examination was normal. The ophthalmologic evaluation confirmed diplopia with weakness of both medial rectus muscles (left>right), but no palsy. Considering this long history of headache, we requested brain MRIscan that showed an important cystic lesion in the sella turcica with compression of the hypophysis and upwards displacement of the optic chiasm. There was no intracystic alteration. A CT scan was also performed to exclude the presence of calcifications. Routine blood test and the hormone profile (pituitary gland fuction) were normal. The most probable diagnosis is Rathke's cyst. Our patient is waiting to undergo NCH surgery.
Diplopia is the perception of 2 images of a single object. It can be mono or binocular. Possible causes are: impairment of cranial nerves to extraocular muscles, mechanical interference with ocular motion (trauma, tumors, Graves disease) and neuromuscular transmission disorders.
Rathke's cyst is a benign lesion, well defined and without calcifications that arises from the residual of Rathke's pouch. It can be symptomatic (diplopia, headache, hypopituitarism) if it is large enough to compress intra-suprasellar structures. In these cases a surgical approach is needed because of the cyst's dimensions and the symptomatic effect on the patient.

####

## AN ATYPICAL ITCHY PETECHIAL RASH

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A 15-year-old boy presented with a two day history of an itchy petechial eruption and sore throat followed by fever. Eruption was extended to axillae, arms, hands, legs and abdomen while palms, soles and face were spared. Laboratory tests revealed: 4600/uL WBC, Hb 16,1 g/dl, 156000/uL platelets. Renal and hepatic functions, serum electrolytes and coagulation tests were normal, CRP was 19,5 mg/L. Urinalysis, nasopharyngeal swab, ADNasi and ASLOT were negative. TORCH and Coxsackie virus tests excluded active infections. Serum Parvovirus B19 (PVB19) IgM and IgG antibodies were positive, indicative of recent infection. Serological panel revealed an EBV reactivation, linked with immunosuppression due to PVB19 infection. On day 4 the rash began to subside without any specific treatment, except cetirizine for itch.
The most common clinical feature of PVB19 infection is erythema infectiosum (EI), but other unusual petechial/purpuric skin eruptions have been described. They are acute self-limiting manifestations often associated with fever and systemic symptoms. Patients are infectious during the rash because, in contrast to EI, purpuric/petechial eruptions are linked with the viremic phase of PVB19 infection. Histopathologically, an inflammatory perivascular lymphocytic infiltrate with erythrocytes extravasation in the dermis is present. In conclusion, PVB19 infection should be considered in every patients

presenting a petechial/purpuric eruption of unclear origin.

####

## VAN WYK-GRUMBACH SYNDROME: A CASE REPORT

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A 7-year-old girl was referred to us because of two episodes of vaginal bleeding. Patient's past medical history was unremarkable and family history was positive for thyroid disease. Auxological parameters were: height 112.3 cm (-2,18 SDS), weight 27 Kg (0.15 SDS), breast development at Tanner stage B2 with no pubic or axillary hair. No other physical abnormalities were observed. Laboratory data revealed TSH 75 µUI/ml, FT4 <0,3 ng/dl and high level of TPO antibodies. Basal LH and FSH were at prepubertal level while estrogens were 37,5 pg/mL, prolactin was 45,8 ng/ml and α-fetoprotein was 8,4 ng/ml. Bone age of a 5 year old. Pelvic US demonstrated a postpubertal uterus measuring 4,8 cm in length and a 5 mm thick endometrial stripe, ovaries were enlarged and cystic. Thyroid US showed an irregular pattern. Pituitary RMN was negative. GnRH test showed an FSH dominated prepubertal response. After L-T4 therapy, she had no further episodes of vaginal bleeding and ovarian cysts decreased in size, suggesting Van Wyk-Grumbach syndrome (VWGS).
VWGS is characterized by hypothyroidism, precocious puberty regressing after L-T4 therapy and delayed bone age, differing from other precocious puberty diseases. Although aetiology of VWGS is uncertain, ovarian stimulation and precocious puberty could be triggered by high level of TSH which can activate the FSH receptor by molecular mimicking. Therefore hypothyroidism should be always investigated in girls with vaginal bleeding and ovarian cysts.

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## WHEN PETECHIAE ARE A FAMILY ISSUE…

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J, male, 4 months, arrived at our hospital with the finding of petechiae on wrists and face and ecchymosis on the ankles which had appeared 24 hours before, during a low respiratory tract infection. No history of trauma, nor bleeding episodes. Past history of haematochezia in the first month of life, resolved after elimination of milk proteins and eggs from the mother's diet. Mother and sister were affected by non immune-mediated chronic thrombocytopenia. On examination: diffused atopic eczema. Blood cell count revealed PLT 28.000/mcl, with normal coagulation parameters. He was admitted to the Hematologic Division for assessment. Infectious tests, abdomen ultrasonography and bone marrow aspirate were normal. Prednisone treatment was set up for two months with temporary improvement of platelet values (105.000/mcl), which then decreased again (PLT 14.000/mcl); whereby intravenous immunoglobulins were administered. In consideration of the family history, associated with thrombocytopenia, low platelet volume (medium MPV 7.4 fl) and with the clinical finding of eczema, the analysis of expression of cytoplasmatic WASP protein was performed. It was compatible with the diagnosis of Wiskott-Aldrich syndrome, which was confirmed performing the molecular analysis of the gene WAS, where the Arg321 mutation was documented. Therefore, allogenic stem cell transplantation from a genotypically identical sibling donor was performed 3 months after diagnosis.

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## TUBERCULOSIS: AN EMERGING PROBLEM

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The term "scrofula" has been used for a long time to indicate a chronic lymphadenitis swelling of the cervical nodes. This is the most common site for childhood tuberculosis. We report an 7-month -old Albanian male, admitted to the Unit of Pediatrics of the University of Catania because of a right laterocervical swelling. Neck ultrasound showed swelling and liquefied nodes with inflammatory reaction that involved the parotid gland and were ulcerated at the skin level. Suspecting tuberculosis (TB), Mantoux test was performed and resulted positive, while TB test (T Spot) results were equivocal. Chest X-ray was negative and abdomen ultrasound showed hepatomegaly. The patient started treatment according to TB protocol with Rifampicin at 20 mg/Kg/day for 6 months, Isoniazid at 10 mg/Kg/day for 6 months, Pyrazinamide at 30 mg/Kg/day for 2 months and periodic drainages of scrofula, with appreciable clinical benefit. The increasing migration flows from countries with high levels of endemic TB, are contributing to an increase in the spreading of tubercular infections, especially in countries in which TB had been almost totally eradicated. In the first year of life, the risk of disseminated disease is about 40% and it reduces to 10% in school age. The extra pulmonary manifestations are significantly more frequent in younger children (infants and preschool) representing about 20% of cases of TB. In particular, scrofula is the most frequent form of extrapulmonary TB in children, at about 70%.

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## WHAT'S BEHIND A SYSTOLIC HEART MURMUR? A MARFAN SYNDROME CASE

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A 16-year-old boy, presented with systolic heart murmur of 3/6 Levine and overgrowth. History revealed he had suffered from aortic stenosis and aortic root dilatation (Z score >2) at age 3, treated with cardiac surgery. At clinical examination height was 188 cm (>95 pc), higher than the genetic target, with arm-span-to-height ratio of 1.05; furthermore, the patient presented a long face with deeply set eyes, arachnodactyly, thoracic deformity and wrist sign.
Marfan Syndrome (MFS) was suspected, and the patient underwent further investigation according to the systemic score of Ghent nosology. Ecocardiography assessed aortic valvolar disfunction, mitral valve prolapse and regurgitation. Eye examination was normal. Column radiography showed scoliosis and lumbosacral MRI ascertained dural ectasia. The systemic score of 7, the aortic dilatation and the negative family history led to diagnosis of MFS. Genetic analysis of mutation in FBN1 is still in progress.
MFS is a genetic disorder of the connective tissue with autosomal-dominant inheritance, usually caused by mutations in the Fibrillin-1 gene and has a prevalence of one in 5000-10000 individuals.
Conclusions: in children, diagnostic criteria for MFS may not be met on first investigation, and establishing the diagnosis may need a follow up of some years. However, early diagnosis is a key issue in the medical treatment of MFS, in consideration of the life expectancy of 32 years without therapy.

####

## A CASE WITH A CAPITAL C

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Scurvy is a rare nutritional disorder due to a prolonged deficiency in Vitamin C intake, which can be still identified in subjects with intellectual disabilities, psychiatric illness or unusual dietary habits.
The disease spectrum includes systemic symptoms and dermatological, haematological, rheumatic, dental and skeletal manifestations.
L. is a 6-year-old girl coming to our attention because of arthralgia with refusal to walk, petechiae on legs and anemia. She had receveid oral steroids, NSAIDs and antibiotics without improvement.
At our first examination she showed poor general conditions, depressed humor, paleness, tachycardia, ankles and knee swelling, petechiae, gingival swelling and dental enamel anomalies.
A more accurate investigation of dietary habits disclosed an extremely restricted diet (milk and biscuits) since she was 18 months old because of a strong refusal for solid food. The clinical history and the manifestations, together with the radiological investigations and the detection of low ascorbic acid blood level, led to the diagnosis of scurvy.
She was immediately put on Vitamin C and other dietary supplements and a strict logopedic, physiotherapeutic and psychological program of recovery was started. She showed a remarkable improvement in humour, dietary behaviour and motor skills within two months.

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## WHEN "TOO MUCH IS TOO MUCH"

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A 15-year-old boy presented with fever associated with left shoulder pain, vomiting and feeding difficulties; he was tired and suffering. A basal left thorax auscultation revealed a reduction of vescicular breathing and bronchial murmur. X-ray disclosed a large thickening of the lung. Blood samples showed CRP 340 mg/l and Creatinine 2.37 mg/dl. We started bolus of isotonic fluid; the urinalysis showed a low fractional sodium excretion < 1%, normal urinary sediments but increased level of B2microglobuline. The ultrasound revealed normal flow on renal artery. So why did the patient present with acute tubular damage in a state of dehydration? We discovered that he had taken a high dose of Ibuprofen for 4 days. He had developed a pre-renal failure due to hypovolemic status with nephrotoxicity caused by NSAID. In cases of volume depletion, the risk of renal toxicity is potentially increased by the use of NSAID drugs due to the loss of vascular compensatory mechanism. In similar cases, there is an upregulation of the renin-angiotensin system and an increase of renal prostaglandin production. The latter decreases the pre-glomerular resistance and preserves the renal perfusion and the glomerular filtration. This preservative effect is inhibited by the use of ibuprofen, resulting in renal vasoconstriction and reduction of the renal flow. Most cases of NSAID-induced renal failure are mild and self-limiting. Interrupting the ibruprofen treatment and hydration are fundamental for recovery.

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## FEVER AND ARTHRALGIA: NOT ONLY ARTHRITIS

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A four-year-old first born was sent to our division due to high fever (T max 40° C) lasting for two weeks. He was poorly responsive to antipyretic therapy, with arthromyalgia, conjunctival hyperemia, sore throat and maculopapular rash (on palm of hands, on plants of feet and on the proximal part of the thighs).
Blood exams showed neutrophil leukocytosis, thrombocytosis, elevated inflammatory markers and positivity to antinuclear antibodies (1/80).
Chest X-Ray, abdominal ultrasound and echocardiography were normal. Suspecting Kawasaki disease, we administered a bolus of intravenous immunoglobulin and, after a new event of fever, the treatment was repeated without clinical response.
For this reason, we considered a diagnosis of systemic juvenile idiopathic arthritis and we started anti-inflammatory therapy (ibuprofen at dosage of 40 mg/kg/day) with only a temporary clinical response. Therefore, we decided to review the medical history which allowed us to discover that a month before the child had presented a hyperemic and swollen tumefaction on his finger.
We performed blood serological tests revealing a positivity to anti-Borrelia IgG 1/80 title with confirmatory result positive of 5 bands. Ceftriaxone therapy was administrated and after about 24 hours the child had a high fever event similar to a Jarish-Herxheimer reaction. The therapy was continued for three weeks with progressive clinical improvement confirming the hypothesis of Lyme borreliosis disease.

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## METFORMIN VS PLACEBO IN NEWLY DIAGNOSED TYPE 1 DIABETES MELLITUS

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OBJECTIVES To evaluate whether metformin administration, as adjunctive treatment in adolescents recently diagnosed with T1DM, can preserve the residual ß-cell function preventing c-peptide secretion failure.
METHODS Study design: multicentric interventional clinical trial. Patients: 10-18 years enrolled between 2-12 weeks from the diagnosis, randomized to 2 groups (the metformin and the placebo group) and treated with a standard insulin regimen. As adjunct therapy, the patients in the metformin group were given 500 mg-metformin tablets, and the patients in the placebo group similar-looking placebo tablets. Follow-up: evaluation of HbA1c and basal c-peptide every 3 months .
RESULTS
•c-peptide is not reduced in V1 respect to V0, neither in V2 respect to V1 in patients treated with metformin, while it is significantly reduced in patients treated with placebo (V1 vs V0 p = 0.03; V2 vs V1 p = 0.012).
•HbA1c is not significantly different in both groups in V1 respects to V0 neither in V2 respect to V1
•HbA1c and c-peptide is not significantly different in both groups in V0, V1, V2

| | HbA1c (%) | | c-peptide (ng/ml) | |
|---|---|---|---|---|
| | *Metformina* | *Placebo* | *Metformina* | *Placebo* |
| start (V0) | 7.2 | 7.25 | 0.9 | 0.7 |
| After 6 months (V1) | 7.0 | 7.65 | 0.73 | 0.5 |
| After 12 months (V2) | 7.4 | 8.0 | 0.56 | 0.30 |

CONCLUSIONS These preliminary results show that the addition of Metformin (500 mg three times a day) within 12 weeks of diagnosis seems to slow down the progressive ß-cell function compared to patients treated with placebo.

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## CORRELATION BETWEEN SEPSIS AND MAJOR CLINICAL COMPLICATIONS IN VERY LOW BIRTH WEIGHT INFANTS (VLBW)

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Neonatal sepsis is inversely related to birth weight (BW) and gestational age (GA).The risk of sepsis for VLBW is high because of immature defenses. We evaluated the epidemiology of sepsis among the VLBW infants in NICU "Federico II" and the influence of sepsis on outcomes: mortality, retinopathy of preterm (ROP), O2-dependence, respiratory distress (RDS), cerebral hemorrhage (IVH), patent ductus arteriosus (PDA), necrotizing enterocolitis (NEC). 74 VLBW (year 2015/16) were divided into two groups (sepsis proven by blood culture positive or not) that were not statistically significant for GA, BW, twin birth, type of birth. For each newborn we considered intrapartum and antifungal prophylaxis, maternal risk factors, sepsis confirmed, localized infections, O2-dependence, ROP, IVH. The Statistical Package for Social Sciences was used for statistical analyzes. RESULTS: 37.8% of patients had at least one episode of sepsis confirmed. For these neonates the mortality (35,7% vs 13%), the incidence of O2-dependence, NEC, ROP and medical therapy for the PDA were higher than controls. There was no significant difference between the two groups for the RDS, IVH and surgery for the PDA. Coagulase Negative Staphylococci caused confirmed sepsis in 46% of cases; Candida, Gram negative and Gram positive were found in the 29%, 15% and 10% of cases, respectively. Our study shows that VLBW with confirmed sepsis may have increased risk of adverse outcomes; we need, however, more studies to confirm these hypotheses.

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## INFLUENCE OF MATERNAL CHORIOAMNIONITIS ON OUTCOMES IN VLBW INFANTS: PRELIMINARY RESULTS OF THE NICU AT UNIVERSITY FEDERICO II, NAPLES

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Chorioamnionitis is often a cause of preterm labor and can increase neonatal morbidity and mortality. We evaluated the influence of maternal chorioamnionitis in VLBW infants at NICU Federico II on outcomes such as sepsis, patent ductus arteriosus (PDA), respiratory distress (RDS), necrotizing enterocolitis (NEC), cerebral hemorrhage (IVH), periventricular leukomalacia (PLV) and mortality. 74 VLBW infants (year 2015/16) were divided into "cases" group (born from mothers with clinical and/or histological chorioamnionitis, as defined in the Infection Register of the Italian Neonatal Network) and into "control" group (without clinical and/or histological maternal chorioamnionitis). Statistical analysis were performed with the software Statistical Package for Social Sciences. E. Coli maternal chorioamnionitis was diagnosed in 8 newborns: 2 had clinical diagnosis, 4 had histological diagnosis; 2 had both clinical and histological diagnosis. The latter two infants died in the first day of life for E.Coli sepsis. One newborn with clinical chorioamnionitis died in the first day without confirmed sepsis. The cases showed mortality and incidence of severe IVH higher than controls. There were no significant differences between the two groups for the remaining outcomes. Many studies associate maternal chorioamnionitis with increased risk of early sepsis, NEC, PDA and neonatal mortality. Our preliminary data confirm the increased incidence of premature mortality and sepsis for VLBW born from mothers with

chorioamnionitis.

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## SPONTANEOUS SUB-CAPSULAR SPLENIC RUPTURE IN A GIRL WITH CHRONIC MYELOID LEUKEMIA

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A 10-year-old girl diagnosed with chronic myeloid leukemia (CML), received chemo/biological therapy without complete molecular remission. Two years later, she was admitted to the Pediatric Unit (Bari University Hospital) in lymphoid blast crisis. She had marked leukocytosis, severe anaemia and thrombocytopenia; she looked very uncomfortable with hepatosplenomegaly, respiratory distress and abdominal pain. A thoracic-abdominal CT scan showed left lung atelectasis with pleural effusion; hepatomegaly with micro-nodules; splenomegaly with inhomogeneous parenchyma and sub-capsular hematoma. She was hemodynamically stable with no evidence of retroperitoneal hemorrhage. *Aspergillus flavus* was cultured from sputum and its antigen detected in blood. She was supported with blood product transfusions, antibacterial and anti-fungal therapy and received intensive chemo/biological therapy. Splenic rupture was managed conservatively (bed rest, monitoring of vital signs). The patient responded with symptomatic and radiologic improvement. Splenic rupture is an uncommon complication of hematologic malignancies, only occasionally reported in children. Fungal infection, leukemic infiltration, thrombocytopenia may predispose children to parenchymal hemorrhages or sub-capsular hematoma. Management of splenic rupture ranges from conservative treatment in hemodynamically stable children, to emergency splenectomy. Splenic rupture should be considered when CML patients experience severe abdominal pain.

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## PSEUDOTHROMBOPHLEBITIS IN AN ADOLESCENT: WHAT'S BEHIND BAKER'S CYST?

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A 14-year -old girl was seen at our hospital with swollen left knee kept in flexion, calf swelling and a 5 week history of left posterior knee pain. There was no history of recent trauma or systemic illness before the presentation. Initial examination revealed swelling and warmth of left calf, positive Homans' sign and 1.5 cm difference in circumference between the 2 calves mimicking thrombophlebitis (TP) or deep venous thrombosis (DVT). D-dimer was normal and Doppler US was negative for TP and DVT but found a 47x11,9 mm Baker's cyst (BC). MRI confirmed the presence of a cyst localized in the popliteal fossa and excluded meniscal or ACL tears. There were no bony abnormalities on X-ray. Blood tests revealed normal CBC, normal thrombophilic profile and negative inflammatory markers. Negative dsDNA and ENA excluded underlying connective tissue disease or vasculitis, ANA were positive (1:320) with homogeneous pattern. Synovial fluid was collected via arthrocentesis, the culture was negative for common pathogens and cytologic evaluation showed an inflammatory aspect. These features allowed us to make a diagnosis of juvenile idiopathic arthritis (JIA).
Pseudothrombophlebitis syndrome (PS) is the occurrence of calf pain and swelling caused by extrinsic compression of the popliteal vessels by an enlarging BC, or as the result of its rupture. BC in children

is uncommon and few cases reported PS as a possible clinical manifestation. Often idiopathic, BC is less frequently related to JIA.

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## TYROSINEMIA TYPE I : A CASE REPORT

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We report a case of a full term newborn who, after an uneventful pregnancy and perinatal period, at the age of three months was hospitalized for worsening dyspnea and hyperpyrexia, suggestive of bronchiolitis. Blood tests showed acute hepatic failure, with increased transaminases and severe bleeding disorders indicating need for liver transplantation. Routine analysis excluded post infections, autoimmune and genetic disorders. Metabolic tests showed increased plasma levels of tyrosine, phenylalanine and methionine. The dosage of succinylacetone showed higher than normal values, confirming the diagnosis of tyrosinemia type I.
At the moment, our patient is nine months old and he is very well, under treatment with nitisinone and on a low tyrosine diet. Due to therapy, the hepatic adenomas and mild renal tubulopathy encountered at diagnosis improved until their disappearance.
Tyrosinemia type I is one of the disorders of tyrosine metabolism. Toxic substances that accumulate are responsable for hepato-renal damage and neoplastic degeneration of the liver. The treatment of tyrosinemia type I relies on the use of nitisinone, to be initiated as soon as possible to obtain a reduction in the risk of developing liver cancer.
This case highlights the importance of performing metabolic analysis in cases of unexplained acute liver failure, as early diagnosis can change the disease course. Liver transplantation was avoided in this case and slowed the hepatotoxic action of metabolite products.

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## PAIN MANAGEMENT IN CHILDREN WITH MUCOPOLYSACCHARIDOSES

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Mucopolysaccharidoses (MPS) comprise a group of rare, genetic lysosomal storage disorders caused by deficiencies of the lysosomal enzymes involved in the degradation of glycosaminoglycans (GAGs). Pain is a common feature in mucopolysaccharidoses; pathophysiology of pain in this group of diseases is still unclear and genesis of pain is multifactorial. Currently, there are poor data about pain management in these patients. Here, we present our clinical experience in complex pain management in three children with MPS.
A 12-years-old girl with MPS 3B was referred to our service for a history of chronic severe back pain with refusal to keep the sitting position; mixed pain was diagnosed and successfully treated with indomethacin and gabapentin. The second case concerns a 18-year-old girl with MPS IV who referred a chronic, severe, low back pain and a trigeminal cephalalgia, partially responding to strong opioids; cannabinoids were proposed with success. In the third case, a pure nociceptive back pain was diagnosed in a 12-year-old boy with MPS 3A and severe cognitive impairment, referred to our service for irritability. Acetaminophen at regular intervals was administered with success. In all cases a multidisciplinary approach was essential to assess and treat pain successfully.

####

## A CASE OF ESOPHAGEAL STENOSIS CAUSED BY UNDIAGNOSED HERPES SIMPLEX VIRUS PERINATAL INFECTION

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Herpetic esophagitis resulting from a disseminated infection by Herpes Simplex Virus is a rare condition in the general population. However, it is more prevalent in immunodeficient patients.
We report a case of esophageal stenosis, diagnosed with an upper endoscopy, in a 2-year-old child, who came to our observation for dysphagia. From the age of six months, the patient had struggled with the introduction of solid food to his diet. The patient was able to swallow only semi-liquid meals, retching and drooling when the parents tried to give him solid or semisolid foods. Furthermore, the parents reported a periodic localized vesicular exanthema, which spontaneously disappeared after about three weeks.
The patient's remote medical history reported a vesicular disseminated exanthema at 3 days of life, treated with infusion therapy with Aciclovir in a Neonatal Intensive Care Center, treated as a case of VZV infection. This infection was not confirmed by serological investigations and no information about the gestational age was available because the child had been adopted shortly after birth by the adoptive parents.
The serological exams that we performed, revealed a previous infection of HSV type 2 (high level of anti-HSV2 IgG; anti-HSV2 IgM negative), while antibodies to VZV (IgG and IgM) were negative. One month after the first endoscopy and after biopsies excluded the presence of esophageal inflammatory diseases that could cause stenosis, an endoscopic dilatation was performed. One month later, the patient was subjected to an upper digestive radiography with barium swallow that confirmed the resolution of stenosis. Currently, the child eats any food smoothly and does not require any drug therapy.
We have described a case of esophageal stenosis due to perinatal herpetic esophagitis, which was not initially diagnosed, and was identified due to the occurrence of a complication and subsequent serological investigations.

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## THROMBOCYTOPENIA AND SPLENOMEGALY: WHEN THE CLINICAL PICTURE WINS OVER THE LABORATORY FINDINGS

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Gaucher disease (GD) is an autosomal recessive disease due to lack of β glucosidase, the typical manifestations of which are splenomegaly and thrombocytopenia. Diagnosis is made by demonstrating reduced activity in glucocerebrosidase of leukocytes and fibroblasts[1].
A 2-year-old patient born to consanguineous parents, affected by thrombocytopenia, dysmorphic features and hepatosplenomegaly was referred to our division. Due to the presence of microcephaly, failure to thrive and dysmorphisms, the patient had been subjected to abdomen ultrasound, which showed hepatomegaly, brain MRI (normal), karyotype (46XX), CGH array (negative).
The patient showed<3rd pc weight and height, microcephaly, low hairline, low-set ears, diffuse petechiae, muscle hypotonia/hypotrophia, hepatosplenomegaly. The bone marrow analysis documented findings suggesting an Idiopathic Thrombocytopenic Purpura diagnosis, while laboratory tests showed an increased chitotriosidase value, with a normal dosage of leukocyte β glucocerebrosidase and urinary mucopolysaccharides. Suspecting GD, we performed skin biopsy for β glucocerebrosidase analysis of fibro-

blasts, the results of which were compatible with GD.
As highlighted by the recent literature, in order to avoid diagnostic delays, GD must be considered in the differential diagnosis of thrombocytopenia with splenomegaly. Our case demonstrates that a strong clinical suspicion should call laboratory results in to question, leading to the need to perform second-level investigations[1].

####

## INFECTIVE SUBACUTE ENDOCARDITIS: OUR EXPERIENCE

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A 15-year-old girl with subaortic restrictive interventricular septal defect was referred to us for recurrent fever, myalgias and arthralgias, fatigue and loss of weight over the previous six months. She had been treated with oral antibiotics and anti-inflammatory drugs without success. Medical history revealed a recent nasal piercing. Physical examination: cardiac 4/6 loud systolic murmur and hepatosplenomegaly. Laboratory tests showed leukocytosis and sideropenic anemia, CRP 91 mg/L, PCT 0.2 ng/ml. Reumatological and infectivological screenings were negative. Chest X-Ray and abdominal US were normal. Echocardiogram findings of mobile vegetations on the right side of the ventricular septal defect and on the aortic valve leaflets confirmed the diagnosis of Infective Endocarditis (IE). According to 2015 AHA Consensus empirical treatment was started. After the finding of three blood cultures positive for *Streptococcus mitis/ oralis,* therapy was changed to an etiological one. Fever decreased after two days of antimicrobial therapy and echocardiographic examination was negative 10 days later.
IE is a rare but life-threatening condition. Presentation is generally indolent with non-specific signs and symptoms. Differing from the past 50 years, the latest guidelines do not recommend antibiotic prophylaxis for dental and non-dental procedures. This case report highlights that in patients with predisposing cardiac condition and history of invasive procedures, IE should be always investigated.

####

## A PARTICULAR CASE OF KNEE SWELLING

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A 3-year-old girl began to limp and to complain of pain in the right lower limb in August 2009 apparently following a trauma. Right knee X-rays and ultrasound (US) were normal. The leg was immobilized by bandaging, a resolution of symptoms followed. A year later, the child started to complain again of pain in her right knee, which appeared swollen. Knee magnetic resonance imaging (MRI) showed thickening and lack of homogeneity of the medial collateral ligament and edema of the subcutaneous soft tissue. Plaster cast was positioned and NSAID therapy was administered for 20 days with pain relief, but the swelling persisted. The child came for the first time to our attention in March 2011. Joint examination showed considerable swelling in the medial region of the right knee,

painful on palpation. Inflammatory markers were negative. Knee MRI showed the presence of a lesion with hyperintense signal on T2 and with low contrast enhancement, between the medial collateral ligament and meniscusfemoral ligament, raising the diagnostic suspicion of bursitis of the medial collateral ligament or of low-flow vascular malformation. Given the diagnostic uncertainty and the persistence of symptoms, surgical excision of the lesion was carried out in May 2011. Histological features were diagnostic for intra-articular venous malformation (IAVM). Knee MRI, performed 1 and 3 years later, showed no recurrence of the vascular malformation. The child is symptom free with a full knee range of motion.

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## AN UNEXPLAINED SPLENOMEGALY: THE UNUSUAL ASSOCIATION OF NEUROBLASTOMA AND GAUCHER DISEASE

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Gaucher disease (GD) is the most frequent lysosomal disorder, that results from loss of function of acid β-glucosidase due to mutations in the glucocerebrosidase gene (GBA1). Clinical manifestations of GD include cytopenia, hepatosplenomegaly, bone disease, growth retardation and, depending on GD type, possible neurological involvement. GD patients have an increased risk of cancer, in particular of hematological origin, while the association between GD and neuroblastoma (NBL) has never been described.
Here we report the case of an adolescent, diagnosed with NBL, also presenting a severe splenomegaly and a history of persistent thrombocytopenia. The association with GD was suggested by the histological findings on bone marrow biopsy and later confirmed by enzymatic and genetic tests. Enzyme replacement therapy (ERT) was undertaken, in association to the treatment protocol for high risk metastatic NBL, and the boy is presently alive at stop therapy.
The evidence of this new association supports the necessity of further studies on GD comorbidities. Moreover, detection of glucocerebrosidase activity, in order to rule out GD, should be considered in the differential diagnosis of each unexplained splenomegaly, since delayed recognition of the disease leads to late treatment and a higher probability of irreversible consequences. An algorithm for early diagnosis of GD in pediatric patients, starting from splenomegaly as key presenting sign, has recently been published.

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## LATE CF DIAGNOSIS IN SCREENING ERA

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Safaa is an Arabian girl born in an Italian region where CF newborn screening test is normally performed. Her parents have a blood relationship.
Diarrhoea, constant rhinitis and chronic productive cough are reported during her first year of life. Growth is regular until the second year. Afterwards, there is an important deceleration along with a significant abdominal distension and constipation. Coealiac screening tests are negative and so are prick tests for food allergens and EGDS. Nevertheless, parents decide to start a gluten and lactose free diet without any improvement.
At the age of 3, E.S. arrives at our Institute for abdominal pain. An abdominal x-ray is performed, showing impacted stool. Between the exams for chronic constipation, sweat test is performed, resulting pathological twice ($Cl^-$ 122 – 113 mEq/l). Because of diagnostic suspicion of CF, a thorax x-ray is performed, demonstrating a pulmonary opacity on

the right lung, and so antibiotic therapy is started. Moreover a steatorrhoea condition is observed, so that substitutive therapy with pancreatic enzymes is started too, on the hypothesis of a pancreatic insufficiency, confirmed later with pathological fecal elastase. CFTR genetics were positive for an undescribed omozigotic mutation.
Safaa's IRT was border-line by Ligurian standards, but it was below cut-off in her locality. As a result, she was not recalled in the first instance and suffered a 3 year diagnostic delay.

####

## A MYSTERIOUS CASE OF DYSPNOEA IN AN INFANT

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G.G. is 6-month-old baby girl, delivered normally after 38+3 weeks of an uncomplicated pregnancy from unrelated and healthy parents. She is breastfed and grows regularly. Her doctor notes transient tachydyspnoea crisis, characterized by subcostal retractions and desaturation, with spontaneous resolution, not related to fever, cough or other symptoms and without any consciousness alteration. Auscultation is normal. She is admitted to another hospital where ECG, echocardiogram, thorax x-ray, fibroscopy, RSV PCR test, abdomen and transfontanellar ultrasound are performed, the results of which are completely normal. The only altered values during the hospitalization are the saturation levels: 91-92% when awake, 84-85% when asleep. She is discharged with PPI therapy, although no signs of GERD were identified. After one month of PPI therapy without improvement in her respiratory dynamic, she was admitted to our department.
A polysomnography is executed, showing median saturation value of 89,2%, nadir 79%. The thorax CT scan showed ground glass opacities of lungs evocative of neuroendocrine hyperplasia of infancy (NEHI). This is a recently described lung disease characterized by marked increases in neuroendocrine cells, affecting infants, mostly born at term, after an initial period of wellbeing, with a favourable outcome.
G.G. was discharged with just 0.25-0.5 L/min of supplemental O2 for the night time, and she is slowly but progressively improving.

####

## SUDDEN ONSET OF STRABISMUS AND DIPLOPIA: NOT ONLY MALIGNANCIES

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A 7-year-old girl was admitted to our ER because of convergent strabismus and worsening diplopia over the previous twnty four hours. No previous history of visual disturbances, headache, or recent head trauma. Stiff neck from one week only.
On neurological examination a right eyeball adduction and diplopia in all directions were detected.
An ophthalmological examination revealed bilateral papilledema with normal visus.
Differential diagnoses were malignancies, venous sinus thrombosis, haemorrhagic stroke and idiopathic intracranial hypertension.
Haematologic, biochemical and hormonal assays were within normal limits.
Brain CT, brain and spinal MRI were negative for any expansive lesions or stroke.
Second Day: The ophthalmological revaluation showed a worsening papilledema with peripapillary haemorrhages, vascular tortuosity, and visual acuity loss.
Considering idiopathic intracranial hypertension, acetazolamide was administered to decrease intracranial pressure.
Fifth Day: The cerebral hypertension increased with further visual acuity loss.
A lumbar puncture was successfully performed in deep sedation with a CSF pres-

sure measurement over 60 mbar. At day ten we changed to corticosteroid therapy because of lack of improvement with diuretics.
Twelfth Day: visual acuity was 4/10 right eye and 6/10 left eye, and a facial paralysis occurred. The patient underwent urgent ventricular peritoneal shunt surgery.
In the following days, rapid improvement of diplopia and of visual acuity were observed.

####

## A CASE OF ALEUKEMIC LEUKEMIA CUTIS WITH TESTICULAR INVOLVEMENT

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M. is an eleven –month-old boy who came to our attention with generalized cutaneous nodules and painless testicular tumefaction. His recent medical history was significantly marked by skin eruption with papules and small nodules which regressed spontaneously over a few weeks. A skin biopsy and a testicular biopsy showed the presence of a diffuse infiltration of atypical blast cells with monoblastic features. Immunophenotyping confirmed the common origin of the disorder in both tissues (CD56+, CD33+, HLADR+, CD34-, CD117-). Molecular study with RT-PCR depicted positivity for t(16;21) (AML1-MTG16) rearrangement and the presence of the nucleophosmin1 (NPM1) gene mutation. Bone marrow aspirate showed a low rate of blast cells with the same phenotype (0.02%). A diagnosis of AML with exclusive extramedullary involvement of skin and testis was made. The patient was categorised in the intermediate risk group and chemotherapy was subsequently started (AIEOP-AML 2013/01 protocol). After the first cycle of induction (ICE), the skin lesions disappeared leaving a darker ring in the site of the previous nodules; testis went back to normal. Bone marrow aspirate confirmed the absence of medullar disease. He continued with his consolidation therapies and he completed the last chemotherapy cycle a few days ago. Take home message: in an infant with suspicious skin lesions you must exclude the presence of aleukemic leukemia cutis.

####

## FAILURE TO THRIVE: WHEN VOMITING DOES NOT STOP

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A 7-month-old Italian boy was referred to Parma Children's Hospital for failure to thrive and feeding problems with several episodes of vomiting per day. The mother explained a history of polyuria and constipation in the last 2 months. He was the first child born to non-consanguineous parents at full term, after a pregnancy characterized by intrauterine growth restriction. Birth weight: 2.7 kg (3° pct).
On admission, the child appeared slightly dehydrated. Blood investigations revealed a metabolic acidosis; the urinalysis was strongly positive for glucose (with normal values of glycemia) and proteins, with a positive urine anion gap. An abdominal ultrasound was obtained, revealing normal intra-abdominal organs; however, global kidney dimensions were increased with preserved parenchymal structure, and swollen and hyperechoic cortex, with no pyelocaliceal ectasia. Suspecting an hereditary Fanconi syndrome, in which renal tubular acidosis is secondary to a metabolic disease, the patient was sent to the Pediatric Nephrology Department in Bologna, where a supportive treatment with sodium bicarbonate, phosphate solution, potassium chloride and Indomethacin was started. With the hypothesis of nephropathic cystinosis, the cystine content of peripheral blood leukocytes was determined

and was revealed to be elevated in two different samples, thus providing the diagnosis. Therefore, Cysteamine treatment at increasing doses was immediately undertaken. The genetic analysis of the CTNS gene is still underway.

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## ONE MORE REASON TO BLEED…

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A 10-month-old, full term infant with a birth weight of 3470g,was admitted for investigation of bloody diarrhoea. She was well on exclusive breast-feeding until 8 months old, but she developed diarrhoeal stools with blood 1 month after the introduction of cow's milk formula. On admission, she was afebrile, in good general condition. Her weight was between the 50$^{th}$ and 75$^{th}$C. Biochemical parameters were normal. RAST test for cow's milk protein was negative. Coagulation was normal. Microbiological and virological analysis of her stools was negative for enteropathogens. Faecal calprotectin was elevated. An elevated titer of IgG CMV in the infant's serum was noticed, additionally urine culture also revealed CMV, instead PCR for CMV in blood was negative. Meckel's scintigraphy with 99m Tc was normal, while abdominal ultrasonography revealed thickening of the wall of descending colon. A limited colonoscopy was performed and macroscopically colitis was seen with oedema, focal erythematous mucosa and aphtous-like ulcerations in rectum, sigmoid, and descending colon. The differential diagnosis comprised allergic colitis, CMV colitis and autoimmune or IBD colitis. Biopsies showed moderate destruction of crypts with oedema and moderate inflammation in lamina propria consisting of plasma cells, histiocytes and eosinophils in clusters. Immunohistochemical staining for CMV was negative. Probably both factors, CMV infection and cow's milk protein allergy could have contributed to the severe rectal bleeding.

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## A RARE CASE OF PRIMARY INTRAVENTRICULAR HEMORRHAGE IN AN ADOLESCENT BOY

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Case report. M, 14 years old, is admitted to the Emergency Department for sudden nuchal headache and vomit. He is suffering with a GCS of 14. Vital parameters, physical examination and blood tests are unremarkable and his past history is negative. Head CT scan shows a mild, primary tetraventricular hemorrhage. The patient's neurological conditions worsen (GCS 12) in a few hours and he becomes hypotensive and bradycardic. A second CT highlights an obstructive hydrocephalus due to the occlusion of the Monro Foramens with unchanged hematoma volume. A temporary external ventricular drain is urgently placed and the cerebral arteriography shows a cerebellar arteriovenous malformation (MAV) successfully embolized. His neurological recovery is complete in 2 weeks.
Discussion. Primary intraventricular hemorrhage (IVHp) is a rare condition in children. The onset is often sudden with headache, vomiting, impaired alertness or coma. MAVs cause up to 54% of IVHp but intraventricular tumors, coagulopathies or vasculitis are also described. Acute obstructive hydrocephalus, due to anatomic obstruction to cerebrospinal fluid flow, represents severe IVHp complication. Hydrocephalus must be suspected in any case of neurologic deterioration because exitus for brainstem dysfunction or cerebral transtentorial herni-

ation is possible. The treatment of IVHp focuses on cessation of bleeding and, if present, relieving hydrocephalus by external ventricular drain until intracranial pressure is normalized.

####

## THE ROLE OF ULTRASOUND TO PREDICT NON INVASIVE VENTILATION FAILURE IN NEWBORNS WITH RESPIRATORY DISTRESS SYNDROME

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The aim of the study was to evaluate the diagnostic ability of lung ultrasound to predict the failure of non-invasive ventilation in newborns treated for respiratory distress. Secondary outcomes were to verify inter-observer agreement and to evaluate any time difference in diagnosis between x-ray and lung ultrasound.
28 newborns with mild to moderate respiratory distress were enrolled.
Transthoracic ultrasound and chest x-ray were carried out within 2 hours from admission to the ward. A neonatologist, a resident neonatologist and a pediatric radiologist analyzed the findings.
15 newborns showed the more severe echographic images (type 1) and were diagnosed to have Respiratory Distress Syndrome (RDS); 11 of them received invasive ventilation. 13 patients showed images less severe and a diagnosis of Transient Tachypnea of the Newborn (TTN) was made (type 2-3).
The cumulative inter-observer agreement between neonatologist and resident neonatologist was found in 89,3% of cases. Concerning x-ray, the inter-observer agreement between pediatric radiologist and neonatologist was 71%, while between neonatologist and resident neonatologist it was 75%.
Diagnostic time for ultrasound and x-ray was, respectively, 5'30''± 19'' and 27'1''±1'57''.
Transthoracic ultrasonography is an effective method in the diagnosis and follow-up of RDS or TTN in newborns. It is safer and faster compared to x-ray, also showing better inter-observer agreement.

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## A CHALLENGING CASE OF INFANT CHOLESTATIC JAUNDICE: NEVER OVERRATE A CMV INFECTION

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In a child of unrelated parents, hailing from the same Moroccan village, ingravescent jaundice with failure to thrive and a rise of transaminases (AST 1530 U/l; ALT 1328 U/l), bilirubin (total 14,66 mg/dl, direct 7,36 mg/dl), ALP (642 U/l) with normal GGT levels were detected at one month of age. Ultrasounds did not reveal liver abnormalities but did show worsening splenomegaly. Liposoluble vitamin deficiency was associated with high levels of total serum bile acids. Alpha-fetoprotein levels were persistently high. Metabolic diseases (aminoacidopathies, fatty-acids disorders, organic acidemias, Gaucher and Niemann–Pick diseases) bile duct atresia, Rotor and Dubin-Jonson syndromes were excluded. CMV DNA was detected in urine and in blood. A magnetic resonance of the brain was performed and revealed germinolytic cysts, possibly, but not purely, related to CMV congenital infection. Since the hypothesis of CMV hepatitis was not convincing and cholestasis markers were persistently high, the child underwent hepatic biopsy showing hepatitis and cholestastis; CMV was not found at histologic evaluation. The persistently normal GGT values raised the suspicion of a Progressive Familial Intrahepatic Cholestasis (PFIC) type 1 or 2. Liver immunostaining showed no detectable BSEP (the main exporter protein of bile acids from hepatocytes to canaliculi). The final diagnosis of PFIC 2 was made. Genetic investigations for mutations of ABCB11 gene are in progress both in the child and in the parents.

####

## VAGINAL BLEEDING IN PREPUBERTAL AGE: A SYMPTOM THAT SHOULD NOT BE UNDERESTIMATED.

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A 3-year-old girl presented at our hospital for episodes of vaginal bleeding. She had no history suggestive of sexual abuse nor trauma to the genital area. The general physical examination was normal, height, weight and BMI were within normal limits. She did not present signs of early puberty. The genital examination did not show signs of infection or trauma, and no vaginal foreign body was visualized. She started therapy with macrolide and cicatrina oliment. She also did urine tests and vaginal swabs, that were negative. After one week of therapy a transabdominal pelvic sonogram showed hypoechoic area (8.4x6.54mm) on the uterine body (Polyp?Submucosal myoma?) and a normal prepuberal ovarian volume, without adnexal masses. Serum LH, FSH and estradiol were normal for prepuberal period, while the 17-hydroxyprogesterone was high. A pelvic RM showed: proliferative lesion (2,5x3x1,6cm) extended from the cervical posterior side to the upper third size of the vagina, with no homogeneous structure and many hemorrhagic areas in the context with high contrast enhancement. No lymph node metastases. No focal lesions in other organs. During the colposcopy a "bunch of grapes" lesion was found in the vagina cavity. On histological examination, the lesion showed clear characteristics of embryonal rhabdomyosarcoma variant botryoides. She started a therapy with IVA (ifosfamide-vincristine-actinomycin) according to protocol EpSSG RMS2005 ARM SR-C, in order to reduce the mass and make the tumor removable.

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## AN UNRESOLVED CASE OF NEONATAL DIABETES

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Neonatal diabetes (ND) is a monogenic diabetes with onset in the first six months of life [1]. Transient ND (TND) resolves by 18 months of age, predisposes to diabetes later in life [2], and is often due to imprinting defects of chromosome 6 [2]. Permanent ND (PND) is predominantly due to mutations of KCNJ11 and ABCC8 genes, encoding for subunits of the sulfonylurea receptor. PND can be caused also by INS and GCK gene mutations (2). We describe a full-term male born from healthy parents. Hyperglycaemia (about 200 mg/dl) was found at 24 hours of age. Blood glucose steadily increased until 10 days, when subcutaneous insulin was started. ABCC8, INS, KCNJ11, and GCK genes sequencing were normal. At 7 months, he started continuous subcutaneous insulin infusion, interrupted at 3 years due to complete remission. c-peptide was always low-normal. At 3.5 years, he presented absence attacks and performed an EEG showing abnormal sleep electroencephalographic patterns. Depakin was started. Brain MRI was normal. At 5.6 years: HbA1c 7.6% and Glargine (2 IU/day) was started. Abdominal MRI showed normal pancreas, specific auto-antibodies were negative, c-peptide was low, fundus oculi was normal. Whole exome sequencing was performed and was negative.
The clinical course of this case is atypical. Genetic testing of neonatal diabetes-related genes and WES were normal. This case suggests that a genetic cause may not be identified in all patients.

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## A CASE OF ACUTE PANCREATITIS: FAULT OF UNUSUAL BEHAVIOR?

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This is the case of R.Z., female 13 years old, hospitalized for vomiting and acute abdominal pain (epigastric and right upper quadrant). She had a history of recurrent abdominal pain and habit of deliberate ingestion of sewing thread. On admission: suffering patient, intense abdominal pain. Blood tests: neutrophilia, elevated transaminases, significant increase of amylase and lipase, negative acute-phase reactants. Abdominal ultrasound showed mild enlargement of the pancreas without alteration of Wirsung and biliary tract. Feeding was stopped, parenteral nutrition and drugs (analgesics, antibiotics, gastroprotective and Sandostatin and Gabesato mesylate) were started. On the second day, there was a significant reduction of amylase and lipase in absence of clinical improvement and then the patient underwent a cholangio-MRI which confirmed the diagnosis of pancreatitis; endoscopy was also performed and stringy greenish material was removed from the stomach. On the third day dyspnea appeared associated with hypophonesis at chest auscultation, there was a rise in inflammation indices and hypoalbuminemia; thoraco-abdominal CT showed peritoneal and pleural effusion which resolved quickly through treatment with albumin and furosemide. Over the following days, clinical, biochemical and ultrasound improvement was seen. In this case, it was not possible to define the etiology of the pancreatitis; however, the history of habitual ingestion of sewing thread confirmed endoscopically may have had a causative role.

####

## TOO MUCH ESR

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Luca is 9 years old when he complains of recurrent acute arthritis to his right knee, treated with NSAIDs and antibiotic therapy. His CRP and ESR are always elevated during the follow-up. 5 months later arthralgia and bone pain starts elsewhere. Full blood count, serologic screening for infections, auto-antibodies and echocardiography are negative. Uveitis is absent. Whole body MRI shows lesions at jaw, distal femur and proximal tibia bilaterally, some left ribs, and a large one at left calcaneus. The bone biopsy is consistent with Chronic Recurrent Multifocal Osteomyelitis (CRMO). Luca starts intravenous Pamidronate, followed by clinical improvement. He is currently well.
CRMO is a rare auto-inflammatory bone disease with a mean onset age of 10 years. Most children complain of pain with or without swelling at the site of bony lesion, typically sternum and clavicles; rarely oligoarthritis is the first sign. It is a systemic disease. Radiographs, technetium bone scans and MRI are useful in the work up and at follow up. Bone biopsy is diagnostic. First line therapy is NSAIDs, while second line therapy is mainly based on bisphosphonate[1].
Luca's case indicates some features which could help in the work up of oligoarthritis: he is too old for a typical reactive arthritis; elevated inflammatory markers are unusual in JIA; MRI is useful, as is bone biopsy, in persistent, generalised bone pain/arthritis mainly to rule out malignancy.

####

## A RARE CASE OF BACTERIAL MYOCARDITIS MIMICKING GASTROENTERITIS

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A 6-year-old girl presented at the ED for abdominal pain, dark stools, vomit and asthenia. She was apyretic, pale, sweaty with cold extremities, tachycardic and hypotensive. Rehydration was performed without any improvement. Blood tests yielded increased troponin I (111 ng/mL); echocardiogram showed dilatative cardiomyopathy with an ejection fraction of 20%. Transferred to our PICU, she required intubation, inotropes and IVIg. However, hemodynamics rapidly worsened with refractory hypotension, so V-A ECMO was started. Even though blood cultures and serologic tests were negative, we suspected a fulminant bacterial myocarditis because of the presence of purulent pericardial fluid as well as neutrophilic infiltrate in a myocardial biopsy specimen. S. Aureus and H. Influenzae were isolated in the bronchial aspirate. ECMO was successfully suspended after 8 days. However, clinical conditions and echocardiographic findings did not improve, so she was inserted on the heart transplant waiting list. After a further deterioration a second V-A ECMO course was necessary, but the patient unfortunately died as a result of cerebral air embolism.
Myocarditis may be difficult to diagnose, often being almost asymptomatic or mimicking other less severe illnesses. Bacterial myocarditis is rare (0.2-1.5%) but mortality is very high (75% in infants, 25% in children). This case is peculiar for the bacterial etiology and the clinical appearance of abdominal disease as a result of intestinal hypoperfusion due to cardiogenic shock.

####

## EVEN HEADACHES BLEED!

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Intracranial hypertension due to hemorrhages of CNS must always be considered when evaluating a child with acute onset of headache and vomiting, even if simulating a CNS infection. Timing of the outset and an accurate anamnestic evaluation are also useful in distinguishing between primary and secondary headache in child.
CT is the gold standard for the diagnosis and focal neurological signs are the main indication to the imaging, but not necessarily the only ones.
Sofia was 10 years old when she came to our Emergency Department complaining of fever (38.5°C) headache and vomiting which started at night. She denied being previously affected by headache. CRP and ANC were normal and she had neither seizure nor any other neurological signs. Chemical serological and microbiological tests on liquor were negative, but some bloody liquor came out during the lumbar puncture. CT, MRI and angiography of CNS confirmed intracranial hemorrhage, secondary to MAV rupture. Sofia needed neurosurgical removal of the hemorrhage with a complete restitutio ad integrum.

####

## A CASE OF GUTTATE PSORIASIS

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We describe the case of B., a 3 year old child, who arrived at the University Federico II in Naples with a pharyngitis treated with paracetamol and which resolved after about three days.
B. showed a new episode of pharyngitis after 10 days. The oropharyngeal swab (OS) resulted positive for group A beta-hemolytic streptococcus (SBEGA) and the child was treated with antibiotic therapy (amoxicillin and clavulanic acid) for 7

days with resolution of clinical signs and negativity of the OS. About 2 weeks later, B. presented a rash characterized by pink salmon colour patches. After a dermatological examination a guttate psoriasis (GP) related to streptococcal infection was diagnosed. All blood tests were normal and the OS resulted positive for SBEGA again. B. was treated with antibiotic therapy a second time. He ate a healthy diet and he was exposed to sunlight with resolution of symptoms. Two weeks later the child repeated the OS and it resulted still positive despite the absence of clinical signs.
Psoriasis is a relatively common disorder in children and can be triggered by an upper respiratory tract infection. It is known that streptococcal throat infections can trigger GP, but the ability to trigger guttate psoriasis is not serotype specific. Recent studies have suggested that T-cells play a critical role in the pathogenesis of psoriasis. T cells in acute GP skin lesions may be activated by streptococcal superantigens. Although anti-streptococcal management, such as antibiotics and tonsillectomy, have frequently been implicated in recurrent psoriasis, there is to date no evidence that either intervention is beneficial. The use of antibacterial therapy is not supported by the case described.

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## BRONCHIOLITIS AND CONVULSIONS: NOT ALWAYS DUE TO APNEAS

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INTRODUCTION: Bronchiolitis is an acute prognostically favorable respiratory condition. However, the presence of risk factors (prematurity, congenital heart disease, immunodeficiency, in the first 12 weeks of life) can lead to respiratory failure and complications.
CLINICAL CASE: M.D.,1 month, is hospitalized for acute respiratory failure. O.E.: severe general condition, pasty skin low perfused, marked hyporeactivity. At the chest there are poor bilateral air flows and gasps in small bubbles. He is intubated, placed in an incubator with cardiac and pulse ox monitoring and connected to ventilator in SIPPV mode. Start blood tests, chest X-ray and echocardiography (normal). During the first hours of hospitalization appearance of tonic-clonic seizures in the limbs treated with phenobarbital i.v. with regression of the symptoms. The finding of hyponatriemia (114 mEq/l) requires an adequate correction. Extubated on the third day, he is placed in "high flow" with gradual improvement of clinical-laboratory and fluid and electrolyte balance (at discharge Na 139 mEq/l).
CONCLUSIONS: SIADH during bronchiolitis is caused by lung over insufflation, secondary to the air trapping, and by hypovolemia, causing a pressure reduction in left atrium, activation of baroreceptors and increase of ADH. Another hypothesis is the hypercapnia that reduces renal blood flow with retention of water and sodium, edema and hyponatremia. The daily control of serum electrolytes and their correction is the gold-standard.

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## HEMOPHAGOCYTIC LIMPHOHISTIOCYTOSIS IN CHILDREN: FOUR CASE REPORTS

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Hemophagocytic lymphohistiocytosis (HLH) is a rare, life threatening disorder mediated by cytokine storm and characterized by fever, pancytopenia and hepatosplenomegaly, associated with laboratory features like hyperferritinemia, hypertriglyceridemia and hypofibrinogenemia. HLH is classified into primary or

familial HLH (FHL), usually diagnosed within the first two years of life and related to inherited gene defects; secondary HLH (sHLH) is triggered by severe infections, autoimmune or oncology disease. We describe four pediatric patients with sHLH who presented at our Institution in critical clinical condition with persistent fever, refractory to wide spectrum antimicrobial agents, hyperferritinemia, hypertriglyceridemia, coagulopathy and splenomegaly. Bone marrow aspiration was performed to detect morphological signs of hemophagocytosis and revealed acute lymphoblastic leukemia (ALL) in two patients, juvenile myelomonocytic leukemia (JMML) and leishmaniasis in the remaining two. The heterozygous A91V mutation in PRF1 was identified in the first patient affected by ALL. HLH was successfully managed by treating primary disease. HLH is a rare but potentially fatal disorder in children. High index of suspicion is crucial for early diagnosis of HLH to ensure a prompt diagnosis and opportune treatment of this potentially deadly condition.

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## JOINT SWELLING IN POST-INFECTIOUS URTICARIA: A CASE REPORT

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We report the case of a 2-year-old boy who came to our attention for the presence of swollen painful joints (proximal interphalangeal joints of the left hand, left elbow and both knees) associated to itchy pinpoint red papules on the back of the left hand, on the left elbow and around both ankles. About ten days before his presentation at the Hospital he had had a respiratory tract infection with airflow obstruction and a 5-day fever, treated with oral antibiotics and salbutamol aerosol therapy. Four days before the appearance of joint swelling he had experienced ocular discomfort in the absence of any sign of conjunctivitis.
Since our first impression was the one of a reactive arthritis, we performed the serology for Mycoplasma, Chlamydia, Yersinia, Campylobacter, EBV, CMV and HBV. In order to exclude a post-streptococcal arthritis, we verified the level of antistreptolysin-O titer and the level of C3 and C4 complement component.
After 24 hours of observation, the cutaneous lesions became purpuric and other joints became swollen, so we performed a urine analysis, an abdomen US and occult fecal blood test to exclude a Schonlein-Henoch Purpura.
In order to rule out our hypothesis, we performed a joint US, to understand whether we were facing an arthritis or a soft tissue involvement.
The joint US only showed oedema of surrounding tissues.
Joint swelling and skin rash responded to antihistamines, leading us to conclude the boy had a post-infection urticaria.

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## A CASE OF NEONATAL HYPERTROPHIC CARDIOMYOPATHY: KEEP NOONAN SYNDROME IN MIND

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The first male child of non consanguineous healthy parents was born at term with Apgar score of 9 at the1st and 10 at 5th minutes. The pregnancy was uneventful except for mild polyhydramnios. The baby was large for gestational age and his glucose profile was normal. A detailed clinical examination revealed only mild hypertelorism. A systolic murmur was detected on the third day of life and echocardiography showed a hemodynamically stable hypertrophic cardiomyopathy (HCM). After escluding maternal diabetes, we performed acylcarnitine profile which was normal. To exclude

other anomalies, abdominal ultrasound was performed and a horseshoe kidney was detected. Cerebral ultrasound was normal. Noonan syndrome (NS) was suspected and confirmed with the molecular analysis of PTPN11 gene. A neonatal HCM can be due to maternal diabetes, metabolic disorders (i.e. Pompe disease and fatty acid oxidation disorders), mitochondrial defect or syndromes. HCM is found with a frequency of 20-61% in RASopathy (i.e. NS, Costello syndrome), with a low frequency in Beckwith Wiedemann syndrome and Down syndrome. A differential diagnosis could be challenging for the neonatologist considering the phenotypic variability of these syndromes and because the clinical features could be not fully expressed early in life. Although the most common heart defect in NS is the pulmonary valve stenosis, this syndrome is one of the most common causes of genetic HCM in neonates and should always be considered.

####

## AN ACCIDENTAL DIAGNOSIS OF SPLENOMEGALY… SOMETIMES MOTHERS' CONCERNS ARE RIGHT

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B.M. is a 3 year old girl. From the age of one year, the mother reported a palpable mass in the left part of the abdomen, pallor and failure to thrive. At 3 years of age she was admitted to a local hospital for foreign body inhalation and she was found to have severe splenomegaly and pancytopenia (Hb 8.1 g/dl, Plt 67.000/mmc and WBC 2710/mmc). Microbiological investigations were negative. On admission to our hospital, the clinical examination showed pallor, petechiae, lymphadenopathy, severe splenomegaly, normal neurological evaluation and no bone pain. Suspecting Gaucher disease we performed serum/plasma chitotriosidase: 3173 nM/ml/h and leucocites beta-glucosidase: 3 nM/mg/h. The diagnosis was genetically confirmed by the presence of homozygous L444P mutation in GBA gene, that is a very high risk factor for developing neurological disease. Both parents were heterozygous for the same mutation. After a few days, she started treatment with Imiglucerase (60 UI/Kg) every 15 days. After 6 months of therapy, the laboratory investigations showed resolution of pancytopenia, reduction of splenomegaly and growth improvement.
Gaucher disease is a rare genetic disorder, caused by deficiency of the enzyme glucocerebrosidase. It is important to recognize early clinical signs to avoid a delayed diagnosis. Enzyme replacement therapy prevents progressive manifestations of Gaucher disease and improves Gaucher disease outcome.

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## I CAN SEE TWO MUMS! AN UNUSUAL MYCOPLASMA LOCALIZATION

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CASE REPORT: 7-year-old boy with an 11 day history of fever and catarrhal cough, currently treated with amoxiclav, arrives at our Emergency Unit. Chest-X-ray shows a parenchymal middle and upper right density with fissure thickening. On the following day, he complains of double vision and develops intermittent diplopia, psychomotor impairment and photophobia. Ophthalmologist confirms diplopia in binocular vision and, after neurology, an EEG showed an increase of the slow component of the temper-parietal-occipital bilateral derivation. Brain MRI shows hyperintensity T2/FLAIR of cerebellar cortex and peri-IV ventricle, and confirmed encephalitis; the PL shows oligoclonal band and the CRP was negative for mycoplasma but serology and pharyngeal swab revealed positive mycoplasma. An empirical treatment with ceftriaxone iv, oral clarithromycin and IV dexamethasone was started with good improvement.
DIFFERENTIAL DIAGNOSIS OF DIPLOPIA: isolated VI nerve palsies for traumatic,

iatrogenic (drugs), ictal (in vasculitis) or post-infectious causes.
CONCLUSIONS: M. pneumoniae is one of the main etiological agents of community -acquired pneumonia in paediatric age. Extra pulmonary localizations are rare (mainly, CNS, PNS, hepatitis, arthritis, and vasculitis). Our experience has shown that, even in common infections, we should never underestimate minor but unusual complaints when treating children ("I can see two mums"). Prompt diagnosis and treatment can lead to speedy recovery.

####

## A CASE OF PRECOCIOUS VAGINAL BLEEDING AND RECURRENT OVARIAN CYSTS

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We describe the case of a 6-year-old girl referred to us for vaginal bleeding. Her pubertal stage was PH1B2I$^-$ and she had one café-au-lait spot along the neck midline. Endocrine studies revealed: prepubertal basal and post stimulation gonadotropin level but elevated estradiol. Pelvic ultrasonography showed a voluminous left ovarian cyst (confirmed by MRI), that was endoscopically resected. Afterwards she presented recurrent vaginal bleedings with concomitant increase of estradiol level and intermittent ovarian cysts, unresponsive to medroxyprogesterone acetate. At the age of 9 years endocrine tests revealed a central precocious puberty, so she started decapeptyl therapy without clinical benefit. Finally, a genetic diagnosis of McCune Albright Syndrome (MAS) was made. Skeletal radiological examination showed multiple areas of fibrous dysplasia involving long bones, ribs and skull, even though she had neither pain nor skeletal deformities. No other endocrine disorders were found. MAS is a rare and complex disease involving skin, skeleton and endocrine system caused by a somatic activating mutation in the GNAS gene. It should be suspected in case of precocious puberty presenting with recurrent ovarian cysts, breast development and vaginal bleeding.

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## TWO UNUSUAL CASES OF NEONATAL DYSPNEA

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P.G., term infant, male, delivered spontaneously, Apgar 9-10, pregnancy uneventful. About 12 hours after birth he developed dyspnea (Silverman Index 4-5) and need for oxygen supplementation. Transferred to our NICU, empirical antibiotic treatment and nCPAP were started. Auscultation revealed diffuse bilateral crackles. At chest radiograph: mild interstitial thickening, destroposition of the heart and gastric air bubble, liver in left hypochondrium. Abdominal ultrasound and echocardiogram confirmed "Situs Viscerum Inversus Totalis". Routine blood and culture tests were normal. Dyspnea slowly resolved after 8 days; nCPAP was withdrawn after 2 weeks. Nasal brushing for ciliary motility evaluation is programmed. "Situs Viscerum Inversus totalis" was prenatally diagnosed in C.A., a female term infant, delivered spontaneously, Apgar 9-9. She developed dyspnea with oxygen-dependency and crackles on auscultation 5 days after birth. Routine blood and culture tests were normal. Chest radiograph showed mild interstitial thickening. Dyspnea gradually improved and spontaneously resolved within 1 week. Nasal brushing confirmed Primary Ciliary Dyskinesia (PCD). Genetic analysis (DNAH1) is in progress.
PCD is rarely diagnosed in the neonatal period; its symptoms are often mild and with quick and spontaneous resolution.

When you come across a late-onset dyspnea (12 hours of life-7 days) without an objective etiology, do not forget PCD!

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## A PECULIAR CASE OF PERSISTENT HEMOPHAGOCYTIC LYMPHOHYSTIOCYTOSIS

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We report the case of a 15-month-old Italian boy who presented with fever, hepatosplenomegaly, pancytopenia, and hyperferritinemia. Clinical and laboratory features were consistent with hemophagocytic lymphohystiocytosis (HLH) and bone marrow aspirate revealed macrophage hemophagocytosis. The child was treated (AIEOP 2004) with high doses of steroids and etoposide (8 infusions) followed by high doses of oral cyclosporine. In spite of this treatment, after 5 months no satisfactory improvement was obtained and the child presented persistent hyperferritinemia and severe cytopenia. On admission to our hospital, a new bone marrow aspirate was performed, which revealed amastigotes consistent with a diagnosis of visceral leishmaniasis. We started a daily administration of liposomal amphotericin B and gradually tapered systemic steroids and cyclosporine. This therapeutic approach led to progressive improvement of clinical and laboratory features with a significant reduction of hepatosplenomegaly and a marked reduction in ferritin levels.
In conclusion, visceral leishmaniasis is one of the main infectious triggers of hemophagocytosis and its clinical picture may be indistinguishable from that of primary HLH. Furthermore, leishmaniasis is not always detectable in the bone marrow aspirate, particularly in its early stages. Our experience suggests that in children with hemophagocytic syndrome who experience an incomplete response to treatment, a new bone marrow aspirate should be considered.

####

## ACUTE DISSEMINATED ENCEPHALOMYELITIS (ADEM): LET'S FOLLOW IT UP

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We report the case of a 2-year-old child with fever, refusal to walk, lethargy and irritability. Physical examination was normal. The anamnesis was positive for an accidental trauma and an episode of gastroenteritis. Blood tests, bacterial and viral serology, physical-chemical and microbiological analysis of cerebrospinal fluid (CSF) were all negative. Brain CT revealed a hypodense area in the left pericapsular region. On MRI, T2-weighted images showed multiple hyperintense focal lesions within the periventricular white matter, encephalic trunk, thalamus and right putamen. Sleep-deprived EEG showed spike-and-wave pattern. Diagnosis of ADEM was considered and he was treated with iv methylprednisone, subsequently, he was started on oral prednisone. Follow up at 1 and 5 months has shown clinical remission and an improvement of lesions detected on MRI. ADEM is a monophasic demyelinating disease of the central nervous system (CNS), common in young children, often preceded by an infection or a vaccination. It is characterised by various neurological manifestations, also present in other conditions such as multiple sclerosis (MS). A normal CSF analysis, typical MRI lesions in white matter and deep gray nuclei and the use

of MRI- Callen criteria, could orient the diagnosis, but there are no specific biomarkers of the disease. Up to 30% of ADEM patients receive a diagnosis of MS after relapses, consequently clinical and radiological follow-up is mandatory.

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## NEONATAL DYSPNEA... ON LATE!

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Riccardo was born at 26 weeks of gestational age with a birth weight of 750g by urgent cesarean section because of placenta previa hemorrhage. He was admitted to the neonatal intensive care unit (NICU). After a single administration of surfactant by InSurE technique, he was supported with nasal continuous positive airway pressure (nCPAP) for 16 days. The further clinical course was uneventful until the 36$^{th}$ day of life, when he showed sudden and rapidly worsening dyspnea. Antibiotics and nCPAP were promptly started, but he soon required endotracheal intubation because of severe respiratory failure. Chest X-ray following intubation showed diffuse opacity of the left lung, well positioned endotracheal tube, displaced tip of the PICC (previously positioned because of feeding intolerance) in left subclavian vein. At the second chest X-ray, the left lung appeared surrounded by massive pleural effusion, confirmed by thoracic ultrasonography. Milky fluid, similar to parenteral nutrition solution, was drained from the pleural space, with a progressive reduction of oxygen requirement. The PICC was removed. Complete clinical and radiological recovery was observed.
Pleural effusion in the neonatal period is rare, including congenital or acquired diseases (approximately 30% *vs* 70%). PICC is widely used in NICU for administering TPN to premature babies. Several complications have been reported, including pleural effusion, of which prevalence is described 0.05-1.1%.

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## A CASE OF "SURGICAL" DYSPNEA

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Case Report: Patient born at 39 weeks out-of-hospital with birth weight of 3435 g and APGAR score 9-10. On day 3 of life, he was transferred to our Neonatal Intensive Care Unit (NICU) because of worsening dyspnea. At admission, the patient appeared pink, reactive, in good general conditions. Vital signs (heart rate, SpO2 and blood pressure) were normal. Dyspnea was moderate (Silvermann Index 4), with clear intercostal and epigastric retractions. Thoracic auscultation revealed mild inspiratory stridor ("cornage"), which gradually increased on the subsequent days. Chest radiography was normal. Fiber optic laryngoscopy showed a large supraglottic arytenoid cyst with relevant inspiratory airway obstruction. Neck ultrasound and CT scan confirmed a laryngeal cyst (16X12X13 mm) causing airway deviation. On day 20 of life surgical excision of the saccular cyst was performed with marsupialization. The cyst wall was removed with laser CO2 therapy. Dyspnea rapidly resolved within the first few days following surgical treatment.
The patient was discharged on day 30 of life with minimal stridor (likely caused by post-surgical edema).
Conclusion: Congenital laryngeal cysts are an unusual cause of neonatal dyspnea and stridor. It is important that rare lesions such as laryngeal cysts are not overlooked. Early diagnosis and prompt surgical treatment are crucial in order to prevent progressive airway obstruction.

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## 12 YEARS OF RECTAL BLEEDING

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S. has a story of persistent haematochezia, starting in the second month of life. At first, it was thought to be milk protein allergy and treated with hydrolyzed formula feeding with initial benefit. From age 2, rectal bleeding reappeared, presenting at each evacuation and being associated with abdominal pain and tenesmus. Suspecting an inflammatory bowel disease, S. underwent several endoscopies which showed nodular follicular hyperplasia of the small intestine and vascular congestion of the rectosigmoid region; histological examinations revealed signs of aspecific inflammation. He was diagnosed with aspecific proctosigmoiditis and treated with oral Mesalazine and topical steroids from the age of 4 with only partial improvement of bleeding. Revision of the images from previous endoscopies suggested the presence of rectal hemangioma; last colonoscopy, performed at the age of 12, was consistent with this condition, showing significantly increased vascular markings in the ampulla and the rectosigmoid junction, with branched aspect and submucosal ectasias. Magnetic Resonance Angiography showed thickening of the recto-sigmoid wall, with spongy appearance after contrast, confirming a vascular dysplasia. Histological reexamination showed several dilated vascular structures within *lamina propria* and *muscularis mucosae*, consistent with angiomatous – angioectasic lesions. Conclusion: despite its rarity, rectal hemangioma should be considered as a cause of gastrointestinal bleeding.

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## SPLENOMEGALY? DON'T FORGET HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS!

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Hemophagocytic Lymphohistiocytosis (HLH) is a hyperinflammatory syndrome that occurs in various underlying genetic (familial HLH) and acquired disorders (infections, malignancies). Macrophage activation syndrome (MAS) is a very similar disorder characterized by a hyperinflammatory response to rheumatic diseases. A 16-month-old male presented at our E.D. with persistent high fever unresponsive to antibiotics, poor general condition, splenomegaly, pancytopenia, high level of PCR, AST, ALT, hyperferritinemia, hypertriglyceridemia, low ESR. Bone marrow examination showed increased hystiocites, bone marrow PCR and serology were positive for Leishmania, so we concluded HLH secondary to Leishmania. A 5-year-old male, 20 days after vaccination, developed fever, lymphadenopathy, splenomegaly, purpuric rash with increased level of PCR, ferritin, LDH, AST and low level of platelets and leukocytes, low ESR. Bone marrow aspirate was not conclusive. He started corticosteroid therapy, with transient benefit. Genetic analysis for familial HLH showed a mutation in XIAP gene. A 17-year-old female arrived at our E.D. due to persistent fever, arthritis, diffuse purpuric and malar rash, splenomegaly, leukopenia, thrombocytopenia and increased level of LDH, AST, ferritin and triglyceridis, ANA and anti dsDNA positive suggesting a MAS as initial presentation of SLE. HLH is a rare but life-threatening disease, therefore early diagnosis is mandatory to set up prompt and specific treatment.

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## A CASE OF "POLKA DOT" LIVER

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INTRODUCTION In literature there are only a few reports of "polka dot" liver in adults. Here we describe a case of polka

dot liver in a young boy in good clinical condition.
CASE REPORT A 14-year-old boy with normal perinatal anamnesis was admitted to our hospital for fever and abdominal pain. An abdominal trauma (a fall down stairs) was reported in anamnesis two months prior to admission. After this event he presented heartburn and nausea treated with proton pump inhibitors with no benefit. Blood tests revealed a rise in C-reactive protein level. Abdominal ultrasound showed liver at the upper limits with a diffusely inhomogeneous structure (cystic formations), a small inhomogeneous area was also detected in the spleen. The patient underwent esophagogastroduodenoscopy that showed hyperemic esophageal mucosa and disseminated small whitish nodules. Abdominal TC and MRI confirmed the presence of hepatic cystic formations. Bone marrow aspirate was performed to rule out lymphoproliferative causes and it was normal. Suspecting hepatic abscesses, a complete immunological evaluation for infectious diseases was carried out. Anti Candida antibodies were present and antifungal therapy was started. Rectal swab was also positive for Candida. For a possible anaerobic etiology intravenous antibiotic therapy was started with subsequent regression of cystic formations. The patient also presented regression of epigastric pain and normal blood tests.

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## INTRACRANIAL VENOUS SINUS THROMBOSIS AS A COMPLICATION OF OTITIS MEDIA IN A 6-YEAR-OLD FEMALE

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Acute mastoiditis (MA) is an intratemporal complication of acute otitis media (MOA) in children developing in 0.2% with adequate treatment. Despite adequate imaging and a proper use of antibiotics, complications of MA occur in 20%. Among them, otogenic transversus sinus thrombosis develops in 2% and carries a high mortality rate (5%) or can lead to long-term neurological morbidities if not promptly recognized. We report the case of a 6-year-old female, treated with amoxicillinclavulanate (80 mg/kg/day) for 7 days for clinical signs of MOA (otalgia, fever and tympanic hyperemia) with clinical resolution. Some days after stopping therapy, she was referred to an emergency ward for a sudden headache, diplopia, photophobia and right strabism. Right papilledema was seen without neurological findings. No infection parameters nor coagulative anomalies were found and thrombophilic screening was normal. MRI showed right transverse venous thrombosis with intense flogosis with no other thrombosis extension. Treatment included high dose broad spectrum antibiotic treatment (ceftazidime and gentamicin), intravenous steroids and low dose heparin anti coagulative dosage. During hospitalization her general conditions progressively improved even the right diplopia. She was discharged with an anti-prophylaxis low dose heparin and oral antibiotic. Two months later diplopia had almost disappeared and the MRI control showed a quite total right transverse senus vessel recanalization.

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## AN UNCOMMON AND SEVERE CAUSE OF ERYTHEMATOUS RASH AND PETECHIAE

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Female, 34 month old with a 2 month history of intermittent fever. Initially admitted to a local hospital, she presented with fever, hepatosplenomegaly,

lymphadenopathy, erythematous rash on face and trunk, petechiae, anemia, leukocytosis and thrombocytopenia. She was immediately referred to Palermo Children's Hospital for further investigations. On admission, laboratory findings showed: WBC 25.590/mm³, N 11.230/mm³, L 9.790/mm³, M 2.980/mm³, Eo 1.260/mm³, B 330/mm³, PLT 49.000/mm³, RBC 4.310.000/mm³, Hb 10.5 g/dl, MCV 80.5 fL, LDH 982 IU/L. After a few days the patient was transferred to the Onco-Hematology Pediatric Unit. On admission: Hb 9.5 g/dl, WBC 52.440/$mm^3$ (N 61%, L 28%, M 7%), PLTS 21.000/$mm^3$. Both LDH and Haemoglobin F were elevated: 1140 UI/L and 44,6%, respectively. Peripheral blood smear: anisopoikilocytosis, left shift with metamyelocytes, monocytosis and 5% blasts. Bone Marrow: hypocellularity and blasts less than 5%. She was diagnosed as suffering from juvenile myelomonocytic leukemia: cytogenetic analysis showed NRAS mutation (pQ6IH variant). The patient was referred to the Pediatric Hemato-Oncology Department at Bambino Gesù Hospital (Rome) where, after five cycles of Azacitidine, hematopoietic stem cell transplantation from unrelated donor was performed: the course was uneventful and the girl was discharged 35 days after Bone Marrow infusion.

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## PSEUDO-BARTTER'S SYNDROME AND DEHYDRATION: ATYPICAL PRESENTATION OF CYSTIC FIBROSIS

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Infants with Cystic fibrosis (CF) are prone to developing episodes of hyponatriemic hypocloremic dehydration with metabolic alkalosis, due to excessive salt loss with sweating, especially during wormer months.
An 8-month-old girl came to our observation because of somnolence and reduced response to stimuli. Weight loss of 5% over the previous 7 days and low-quantity micturition in the previous 24 hours were reported. She showed slightly dry mucous membranes, tachycardia (140beats/min), capillary refill time <2sec. As a first approach we administered i.v. 0.9% NaCl 20mL/kg in 2h with prompt improvement of the patient's general condition. Biochemical exams showed hyponatriemia (Na 135mEq/L), hypokaliemia (K 2,6mEq/L), hypocloremia (Cl 95mEq/L) and metabolic alkalosis (pH 7.57; $HCO^{3-}$30,3mmol/L), Fractional excretion of sodium was 0.98%. Therefore, i.v. 0.9% NaCl 100mL/kg in 24h was continued with the addition of KCL 40 mEq/$m^2$/die. After 24h we observed good general conditions, normalization of both blood parameters and diuresis. The recovery appeared far too prompt for that of a dehydrated patient with Bartter's syndrome. Therefore, the patient was submitted to the sweat test which resulted positive twice. Conclusions: Cystic fibrosis should be considered in differential diagnosis of any child presenting with unexplained dehydration, metabolic alkalosis, hyponatremia, hypocloremia, and hypokalemia. Low sodium and chloride urinary levels can help to exclude Bartter's syndrome.

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## MULTIPLE CONGENITAL ANOMALIES, HEPATOSPLENOMEGALY, PANCYTOPENIA, POLYSEROSITIS: THE COMPLEX CLINICAL SPECTRUM OF A RARE CYTOSKELETON DISEASE

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We report the case of a young boy, born preterm after uncomplicated pregnancy. He presented persistent ductus arteriosus, left ventricular hypertrophy and inguinal hernia. He also showed a failure to thrive with auxological parameters steadily under the third centile. In addi-

tion, he was affected by developemental delay, severe intellectual disability, bilateral sensoryneural deafness and facial dysmorphisms: mid-face hypoplasia and short philtrum. At 2 years of age, hepatosplenomegaly associated to persistent pancytopenia was noted, confirmed by consecutive complete blood counts. A minimal white blood cells value of 900 mcL, a platelet count of 41,000 mcL and an hemoglobin level of 5,3 g/dl were reported. The peripheral blood smear revealed the presence of a high number of megakaryocytes and pseudo-Pelger–Huët anomaly in neutrophils. Radiological investigations revealed pericardial, pleural and peritoneal effusion with concurrent hypoalbuminemia. Genetic analysis showed that this patient carried a mutation in the CDC42 gene which plays a critical role in the formation of actine cytoskeleton. His phenotypical, hematological and neurological anomalies are explained by the involvement of the protein in several biological processes such as the genesis of platelets from megakaryocytes and the proliferation of neuronal progenitor cells. This condition is known as Takenouchi-Kosaki syndrome, and it has been described in three other patients.

####

## OLIGURIA AND DIZZINESS: ADD SALT

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A 9-year-old girl was referred to our emergency department after a severe trauma. She had been intubated at the scene due to Glasgow Coma Scale (GCS) score of 6. She was hemodinamically stable with open wound on scalp, right homerus and knee with local hematoma. Patient underwent total body computed tomography (CT) scan that showed a displaced-exposed distal femur fracture and a parietal skull fracture without intracranial injuries. Patient was admitted to Intensive Care Unit and underwent open reduction fixation with K-wires and bridge external fixator. She was extubated after 36 hours and transferred to the ward. Blood test revealed anemia (8,3 g/dl). She showed post-traumatic stress disorder symptoms (anxiety and insomnia). Oral Lorazepam was administered. After 2 days of therapy she showed dizziness and aphasia, level of consciousness was impaired (GCS 13-14) and mild oliguria was demonstrated without peripheral edema. Lorazepam was stopped without improvement of clinical conditions. Laboratory data showed severe hyponatremia, increase of serum osmolarity and urine sodium concentration and decrease of urinary osmolarity. The subsequent evidence of polyuria allowed us to confirm a diagnosis of Cerebral salt wasting syndrome (CSW). Therapy with Fludrocortisone, normal saline and sodium chloride was given with normalization of sodium concentration.
This case is evidence of the importance of electrolyte balance evaluation in cases of delayed neurological symptoms after brain injury.

####

## UNEXPECTED SOUVENIRS

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An 8-year-old Peruvian girl arrived at the Emergency Department for occipital headache and vomit, followed by loss of contact with reality with left mouth and gaze deviation and left side twitching. Medical history was negative. She had lived in Peru for 10 months, 5 years before. Head CT scan showed a target lesion of 32x42 millimetres in right parieto-

occipital white matter. MRI showed a cyst with peripheral contrast enhancement, a central spot and thin contour, interpreted as cavernoma. Antiepileptic treatment was started, but wrongly interrupted by the family after a month. Two weeks later, she presented with headache, nausea, scintillating scotoma, difficulty recognising people and blurred vision. A second MRI led to the suspicion of a colloidal neurocysticercosis. However, serology for cysticercosis and stool examination were negative. Abdominal ultrasound and ocular evaluation were normal. Therapy with albendazole plus 7-days dexamethasone and carbamazepine was started. A month later, no comitial events had been reported and the control MRI showed a reduction of the cerebral lesion. Therefore, the antiparasitic therapy was stopped.
This case underlines the importance of considering neurocysticercosis in the differential diagnosis of comitial crisis, especially in children who have lived in or travelled to endemic areas. The diagnosis of this condition is difficult and relies mainly on a clinical and radiological picture, as serology can be falsely negative, particularly in the case of a single lesion.

####

## TB OR NOT TB?

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QuantiFERON-TB Gold In-Tube is a T-cell interferon-gamma release assay used for diagnosis of Mycobacterium tuberculosis (Mtb) infection as an alternative to tuberculin skin test (TST). It detects two antigens encoded by a specific genomic segment (Region of Difference 1), that is not present in BCG vaccine strains and in the majority of nontuberculous mycobacteria (NTM). We report two cases of cervical lymphadenopathy. CMV, EBV, Adenovirus,Listeria, Bartonella henselae, Toxoplasma gondii and Streptococcus pyogenes infections were ruled out. TST and QuantiFERON-TB test were positive in both cases. Suspecting a tubercular (TB) lymphadenopathy, a standard TB four-drug regimen was started. Culture of bioptic sample was positive for Mtb in one case, whereas for the other, M. kansasii was isolated. Therefore, the first patient continued with isoniazid and rifampicin for 6 months plus ethambutol and pyrazinamide for 2 months and the second patient was switched to clarithromycin and rifampicin for 3 months. In conclusion, QuantiFERON test is considered diagnostic for Mtb infection, negative in case of NTM infection or BCG vaccination. However, it cross-reacts with M. kansasii, M. szulgai, M. marinum, M. flavescens, and M. gastrii, so in case of cervical lymphadenitis with positive QuantiFERON test, culture must be performed to distinguish Mtb from NTM infection, thus, guiding clinical management.

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## A CASE OF GOLDBLOOM SYNDROME REVEALED BY STIR MRI

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A previously healthy 9-year-old girl experienced daily crisis of bone pain in the lower limbs, associated with fever spikes, limping and nocturnal awakenings. Only a partial response to ibuprofen was observed. Physical examination was unremarkable. Laboratory tests showed mild anemia, thrombocytosis, increased acute phase reactants and high antibody titer against streptolysin O (4280 UI/ml) and DNase-B (6310 UI/ml): an atypical dysproteinemia, characterized by hypoalbuminemia and increased a1, a2 and g globulinemia, was also noted. Throat swab was positive for group A β-

hemolytic streptococcus. X-ray of the lower limbs showed periosteal hyperostosis at femurs and tibias. On STIR MRI, these bones revealed hyperintense signal at the metaphysis. Histopathological examination showed signs of chronic inflammation. Two mg/kg prednisone was started, leading to complete recovery within a few days. Therapy was regularly reduced after two weeks and, finally, suspended after a month when the bone changes disappeared. No recurrence has been observed in the following years.
The clinical picture observed in our patient was consistent with the illness originally described by Goldbloom et al. So far, only a few cases have been reported in the literature. Herein, we report one further case, stressing the STIR MRI features, these may permit a timely diagnosis and a safe control of therapy outcome.

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## CCAMING SOON

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We present the case of male AGA, born at 38w, by spontaneous delivery. Perinatal clinical course was physiological.
At day 14 he was admitted to NICU for sepsis and he started antibiotic therapy. X-Ray showed a pleural effusion, so thoracentesis was performed (bacterial culture negative); after a few hours tension L-PTX appeared, so he was intubated and a chest drain was positioned.
A chest –TC was performed, due to clinical deterioration, which was indicative of lateCHD. The child was trasferred to our NICU on day 18.
The radiological examination was inconclusive between congenital lobar enphysema and pneumatocele. At day 27, after clinical and inflammatory marker improvement, the child underwent surgery with removal of lung IX-X segments + undefined cystic mass like a pneumatocele.
Histology revealed a cystic mass similar to a loculated pleural abscess, and a lung sample like a Congenital cystic adenomatoid malformation (CCAM type III).
CCAM of the lung is a rare anomaly of the lower respiratory tract characterized by cystic adenomatous overgrowth of the terminal bronchioles and airspaces.
80% of cases are usually identified during the first 2 years of life, and typical manifestations include progressive respiratory distress or recurrent respiratory tract infection.
Surgical resection of the affected part of the lung is the treatment of choice, because it may prevent recurrent respiratory tract infections and malignant transformation.

####

## A BABY-GIRL WITH FEEDING PROBLEMS: ALL THAT GLITTERS IS NOT…SILVER!

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I. is 7-month-old baby girl born at term, SGA. Neonatal hypoglycemia is known. She is admitted to our department due to failure to thrive and vomit, previously treated unsuccessfully with PPI. At physical examination we find weight <3rd, length <3rd, relative macrocephaly, triangular shaped face, hypotonia. I. is unable to tolerate oral feeding and needs enteral nutrition with NG tube for 6 months. During this period she grows very slowly with recurrent vomiting. Blood tests, esophageal pH monitoring, barium swallow X-rays, abdominal ultrasound, EGD and others exams (head CT, transfontanellar ultrasound) are normal. Cow milk protein intolerance is excluded. Pelvic ultrasound demonstrates a normal uterus and the absence of ovaries. Patient's clinical presentation suggests the diagnosis of Silver-Russell syndrome (SRS). SRS is caused by maternal uniparental disomy for chromosome 7 or

11p15 hypomethylation and its main features are growth retardation with antenatal onset, failure to thrive, typical facial phenotype (relative macrocephaly, triangular shaped face), frequent body asymmetry, feeding problems and severe vomiting under the age of 1 year.
Unexpectedly, we find a very high level of FSH and a kariotype exam shows a mosaicism 45X/46XY, a rare sex development disorder. As described in literature, infants affected may show SRS-like phenotype, including gastrointestinal problems as in this case.

####

## STRANGE RESPIRATORY DISTRESS

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Case report: DF, male, 14 years old, comes to our Emergency department at 1:30 p.m. for mild dyspnea and sore throat. Vital signs: SpO2 97%, body temperature 37.6 °C, cardiac frequency 147 bpm. Physical examination shows agitation, continuous belching, hyperemic pharynx, hypertrophic tonsils, so we start clinical monitoring.
At 3:30 p.m. he has a respiratory and cardiac arrest: doctors start RCP with AMBU (FiO2 100%), the ECG monitoring shows sinusal rhythm with 110 bpm. Anesthetists arrive and the patient is intubated: the laryngoscopy shows glottid oedema (however, he is intubated without problems). He is admitted to UTI where he starts antibiotic therapy and vital signs are monitored continuously. Blood exams and in particular the throat swab result positive for H. influenzae. Asking the family, it is revealed that the boy was not vaccinated for H. Influanzae when he was a child.
Differential diagnosis of inspiratory dyspnea: laryngitis, foreign body, epiglottitis.
Conclusions: Nowadays, an anti-vaccination attitude is spreading, so long forgotten are reemerging diseases: it is important to keep them in mind and never underestimate any symptom.

####

## A CASE OF LIGASE 4 (LIG-4) DEFICIENCY TREATED WITH ALLOGENEIC HEMATOPOIETIC STEM CELL TRANSPLANTATION (HSCT)

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LIG-4 enzyme plays a role in DNA repair and immunoglobulin and TCR gene rearrangements. Its deficiency leads to telomere shortening, deficit of B and T lymphocytes and can be associated with myelodysplasia and bone marrow failure.
We observed a 17-year-old boy with persistent leukopenia, low lymphocyte count and opportunistic infections since age 2, with normal phenotype, for the first time. In the previous year he had shown worsening anemia and thrombocytopenia. Bone marrow was myelodysplastic with megakaryoblastic hypoplasia. We found a shortening of telomeres in WBCs. NGS panel was employed to identify biallelic mutation of LIG-4.
Due to pronounced radio-chemosensitivity related to shortening of telomeres, he received a first bone marrow HSCT from his matched brother after a reduced immunosuppressive conditioning regimen (CR) according to DNA repair disease conditions (Fludarabine-FLU, Cyclophosphamide and Campath). The patient developed a primary rejection, and one month later he received a second HSCT from the same donor, but using peripheral blood stem cells and after a more intensive CR including FLU, Melphalan and low dose of Total Body Irradiation (200cGy). The patient did not develop acute toxic complications. Actually, one month after the second HSCT a complete engraftment of donor cells was seen. This is a second case of patient affected by LIG-4 deficiency who received allogeneic HSCT, in our experience the

use of more aggressive CR allowed for the engraftment.

####

## WHEN BLOODY DIARRHEA PERSISTS… REMEMBER FECAL CALPROTECTIN!

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We report the case of a 10-year-old boy who came to our attention with a history of bloody diarrhea, accompanied by urgency, tenesmus and abdominal pain. His medical history was unremarkable except for iron deficiency anemia treated unsuccessfully with oral iron and atopic dermatitis since the first months of age. He was on a two-year cow's milk free diet because of a previous diagnosis of cow's milk allergy due to positive RAST. His physical examination revealed bloating but soft and non tender abdomen without organomegaly and poor weight gain. Blood tests showed only increased CRP and ESR, with normal full blood count and coagulation. Hepatic, renal, pancreatic and thyroid function were also normal. Serum analyses for celiac disease and auto-antibodies (ANA, anti-DNA) tests were negative. Microbiological and virological stool analyses were negative. His fecal calprotectin was elevated (>500 mcg/g). A colonoscopy was performed which showed a rectum-sparing ulcerative colitis-like IBD pattern with an E4 proximal extension according to Paris classification. PUCAI was 50 (moderate disease activity). After ruling out viral and tubercular infections, he was treated with Methylprednisolone 1 mg/kg/die, tapered in 4 weeks, and Azathioprine, initially at 1 mg/kg dose, then increased to 2 mg/kg as it was well tolerated. After 6 months of therapy he experienced full clinical and endoscopic remission.

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## PERSISTENT RECTAL BLEEDING IN CHILDHOOD: NOT ONLY ANAL FISSURES

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An 8-year-old boy presents with a 6 month-history of alternating stool pattern and some episodes of rectorrhagia after defecation, during constipation periods. No other symptoms are reported. His birth, medical and family histories are unremarkable. At medical evaluation he presents with height and weight over the 95$^{th}$ percentile. Abdominal physical examination shows bloating but neither tenderness, nor palpable masses, while 2 deep anal fissures are noticed at 3 and 6 o'clock in lithotomy position. Blood count, faecal calprotectin and the other inflammatory biomarkers are negative. Suspecting a functional disorder, oral laxatives and a local therapy for fissures (steroid + lidocaine) are prescribed with full remission. After 6 months, the boy experiences a new episode of rectal bleeding with a normal stool pattern, successfully treated with local therapy for fissures. 4 months later, after a new episode of rectal bleeding with a normal stool pattern the boy undergoes colonoscopy that reveals one subocclusive bleeding polyp of size 4 cm in the ascending colon. Intraoperative polypectomy is performed and the histological examination rules out malignancies, showing an inflammatory pattern. In the follow-up visits the patient is healthy with no more rectal bleeding occurrence or defecation complaints. We report this case to emphasize the importance of not stopping at anal inspection when rectal bleeding is persistent.

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## IMMUNE THROMBOCYTOPENIA, HAEMOLYTIC ANAEMIA…IS THERE ANYTHING ELSE?

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Evans syndrome (ES) is the simultaneous or sequential combination of destruction of at least two blood cell lineages (frequently autoimmune haemolytic anaemia + immune thrombocytopenia). In 50% of the cases, this condition is secondary to other immune disorders. In this paper, we present the case of a 10-year-old boy with ES. Admitted to the hospital with one day history of cutaneous and mucosal petechiae, his past medical history included an immune thrombocytopenic purpura at 2 years treated with corticosteroids and intravenous immunoglobuline (IVIg). The patient was afebrile and did not present any alteration at physical examination except for presence of petechiae and splenomegaly. Blood cell count showed thrombocytopenia while there were no abnormalities on bone marrow aspiration. Direct IgG Coombs test and IgG vs Platelets test were positive, revealing ES (autoimmune haemolytic anaemia + immune thrombocytopenia). ES secondary causes were also tested but no sign of recent infections, Autoimmune Lymphoproliferative Syndrome (ALPS) or autoimmunity markers were found. Common Variable Immunodeficiency Disease (CVID) was diagnosed due to the low levels of IgG, low count of CD20+ and lack of response to vaccination (without other abnormalities to the immune system). After various attempts using IVIG, Prednisolone and Mycophenolate Mofetil the patient went into remission from the disease using a therapy that consisted of the use of eltrombopag + sirolimus.

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## SORE THROAT, PHARYNGITIS AND HEART MURMUR: THE GOOD, THE BAD AND THE UGLY.

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A 5-year-old child was brought to the emergency room for respiratory difficulty. Physical examination revealed BT 37.4 ° C; BP 94/53 mmHg; SpO2 100% in room air; HR 120 bpm; RR 40 bpm, poor general conditions, exudative pharyngitis, rales at pulmonary bases, a gallop rhythm with a 3/6 L systolic murmur with diastolic flow murmur and mild hepatomegaly. Blood tests showed rising inflammatory markers and at chest X-ray a moderate cardiomegaly without clear signs of congestion. Past medical history revealed a febrile pharyngitis followed by transient coxalgia and progressive noctnurnal dyspnea. Therefore a throat culture was performed while an urgent echocardiography showed massive organic and functional mitral regurgitation, and severe left atrial and ventricular dilatation. Because of the GAS positive swab and the ASO titer elevation a diagnosis of rheumatic carditis with severe mitral involvement was made. After one month of steroid, antibiotic and heart failure therapy the child underwent mitral valvuloplasty. Currently the child is doing well, continuing the cardiological follow-up with yearly echographic controls. Although acute rheumatic fever has declined in incidence in western countries the disease remains one of the most important causes of cardiovascular morbidity and mortality and it should always be kept in mind when facing a heart murmur after a pharyngitis.

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## DIPLOPIA, A MULTIDISCIPLINARY MONSTER

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INTRODUCTION: Diplopia is a symptom which can pose a diagnostic challenge for the neurologist and ophthalmologist. Its mechanism (monocular or binocular), type, temporal progression and associated symptoms are important for its etiological diagnosis.
CASE REPORT: A 13-year-old boy admitted to pediatric ER referring binocular diplopia and sporadic balance disorders for about 24 hours, reporting a gastrointestinal and upper respiratory tract infection some days before. He had a bilateral fixed mydriasis, binocular diplopia in the nine positions of gaze (involvement of III, IV and VI cranial nerve) and wide-based gait as minimal sensory ataxia. CT scan revealed neither ocular fundoscopy and vision field abnormalities nor pathological findings. He was hospitalized. Blood tests and EEG were not significant. Brain and brain stem MRI was performed, disclosing a rhinosinusitis with predominant ethmoid and sphenoid involvement. A clinical worsening with areflexia induced the suspicion of Miller-Fisher Syndrome (MFS). A lumbar puncture was performed looking for anti-GQ1b antibodies (Ab), which was in the normal range. Intravenous steroid led to rapid healing.
DISCUSSION: MFS is a variant of Guillain -Barre Syndrome characterized by ophthalmoplegia, ataxia, and areflexia. Diplopia is often the presenting symptom. It is associated with anti-GQ1b Ab; however, more than 10% of patients are seronegative.MFS is usually self-limiting and patients recover after few months.

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## A CASE OF VAGINAL BLEEDING DUE TO A BENIGN OVARIAN MASS. THE IMPORTANCE OF FOLLOW-UP FOR THE DIFFERENTIAL DIAGNOSIS BETWEEN OVARIAN CYST AND TUMOR

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A previously healthy 5-year-old girl came to our attention because of painless vaginal bleeding. She had neither abdominal pain nor abdominal masses and a B2–PH1 Tanner stage.
After GnRH stimulation, FSH and LH were at pre-pubertal levels, and estradiol was undetectable. Tumor markers were within the normal range. Pelvic ultrasonography revealed a right ovarian cystic mass of 37mm in diameter and uterine enlargement (43x24x12 mm). Bone age x-ray was about 1 year advanced.
She was carefully monitored and, after one month, symptoms spontaneously disappeared. A new pelvic ultrasonography showed pre-pubertal symmetric ovaries.
Suddenly, one month later, B2 Tanner stage reappeared, with normal serum estradiol levels. A right ovarian cystic mass 45 mm at largest diameter and uterine enlargement were detected by ultrasonographic investigation. Bone age x-ray was confirmed as stable.
Routine thyroid laboratory tests were in normal range and x-ray investigation did not show bone dysplasia.
Because of the peculiar recurrence of clinical signs, she underwent genetic analysys for McCune Albright syndrome (MCA): it was positive.
Conclusions: Clinical features of ovarian cyst may overlap with those associated to neoplastic masses. Furthermore, an ovarian cyst may be the only one feature in the McCune Albright syndrome. For these reasons a strictly clinical, ultrasonographic and laboratoristic follow-up is extremely important to pursue a precise diagnosis.

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## A PARTICULAR ASSOCIATION BETWEEN CYTOPENIA AND SPLENOMEGALY

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In this report we describe a case of a 9-year-old male affected by Evans Syndrome (mild chronic autoimmune multilineage cytopenias, 2 episodes of acute hemolytic anemia and 1 of immune thrombocytopenia) and associated splenomegaly, thoracic and axillary lymphadenopathy followed up at our Hematology and Oncology Outpatient Clinic. On initial evaluation, malignant, infectious and metabolic etiologies of splenomegaly and cytopenias were ruled out. Due to an elevated TCR+CD4-CD8- double-negative T cells (DNTs) count on lymphocyte subsets study an Autoimmune lymphoproliferative syndrome (ALPS) was suspected. We therefore performed a Fas receptor-induced T lymphocyte apoptosis assay on 2 different peripheral blood samples which gave conflicting results. In order to definitively confirm the diagnosis, we employed FAS (TNFRSF6) gene PCR analysis, according to the current diagnostic criteria.[1] The same heterozygous G856T mutation in exon 9 (death domain) of FASgene was also found in the patient's brother and mother who had increased DNTs,but no symptoms. In order to control persistent cytopenias resistant to corticosteroids and Mycophenolate Mofetil (MMF) treatment, in agreement with patient's family, 10 infusions of Rituximab were administered. The child developed hypogammaglobinemia which persisted after the suspension of Rituximab. He is currently in good clinical condition, no further hemolytic/thrombocytopenia episodes have occurred so far, subcutaneous immunoglobulin infusions are still ongoing.

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## A CHRISTMAS HUS

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Hemolytic Uremic Syndrome (HUS) is characterized by hemolytic anemia, thrombocytopenia and renal impairment. More than 90% of cases are secondary to Shiga-like toxins produced by different *Escherichia coli* serotypes. Atypical HUS is less common and involves abnormal activation of the complement alternative pathway and progression to end-stage renal disease.
We describe the case of a 2-month-old female twin with atypical HUS presenting in winter.
Born at 29 weeks 5 days gestation by caesarean section, S. was admitted to the NICU for prematurity, respiratory distress and jaundice. She was readmitted at 2 months for respiratory distress and diarrhoea and suddenly developed anemia, oedema and macrohematuria. Lab results: Hb 4.8 g/dl, PLT 51.000/ mmc, C3 53 mg/dl, C4 8 mg/dl. Creatinine rose to 3.88 mg/dl; urine output fell to 0.3 cc/kg/h despite continuous furosemide infusion (3 mg/kg/h), etacrynic acid and spironolactone boluses; several RBC transfusions were necessary. Shiga-like toxin and *S. pneumoniae* were not detected. Methylmalonic acidemia with homocystinuria (an inborn error of vitamin B12 metabolism) and ADAMTS-13 deficiency were excluded. Eculizumab, a monoclonal antibody against C5, was started and a great improvement in diuresis, renal function, Hb and PLT levels was noted. Atypical HUS should always be considered in young children (< 6 months) presenting with or without diarrhoea; hypocomplementemia and the absence of Shiga-like toxins further support this diagnosis.

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## AN ACUTE LATE-ONSET DYSPNEA

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Male, gestational age 23 weeks, birth weight 690g; surgical closure of patent ductus arteriosus on day 20. On day 65, he developed dyspnea with increased oxygen need, apneas and bradycardia. Chest X-ray and lung ultrasound (US) showed a huge left pleural effusion; 8 ml of milky fluid, whose composition was consistent with chyle, were removed by thoracentesis. Chest drainage was placed and enteral formula with medium-chain triglycerides (MCT) was started, with no improvement; octreotide was thus commenced, but proved ineffective. Due to the infant's weight and poor condition, surgical treatment was not viable; hence, fluid restriction and high-dose diuretics were undertaken, with no benefit. While replacing the chest drainage, a sudden bleeding from its insertion point occurred, with subsequent formation of fibrin clots. In the following days, chyle output progressively decreased and chylothorax resolution was finally achieved.
Chylothorax is the most common cause of neonatal pleural effusion and represents a possible complication of thoracic surgery. Chest X-ray and lung US are useful diagnostic tools, while thoracentesis is both a diagnostic and therapeutic procedure, as it allows respiratory symptoms relief and pleural fluid analysis. Medical management, including intravenous lipid emulsions, enteral MCT formulas and off-label octreotide or somatostatin, is effective in 80% of cases; if it fails, surgery should be considered in relation to the infant's characteristics.

####

## NEPHRADVISOR: PAY ATTENTION TO DRUG COCKTAIL

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Acyclovir and b-lactam are commonly used for empiric treatment of meningoencephalitis, according to different dose regimens. Their combination can strengthen renal damage through tubulotoxicity, thus caution should be exercised using these drugs in association.
A 7-year-old girl presented with fever of 39.9°C, headache, vomiting, lower leg pain and soft stool. Suspecting brain involvement, a CT scan was performed as well as sepsis screening (both normal). SCr at admission was 0,39 mg/dl. Ceftriaxone 80 mg/kg/day was started. The development of subjective vertigo and behavioural changes prompted an EEG (slow waves in the right temporal lobe) and acyclovir was begun (20 mg/kg, 3 times a day).
Blood exams showed progressive development of acute kidney injury (SCr 3.73 mg/dl, BUN 63 mg/dl). 48 hours from admission, urine analysis revealed proteinuria (PrU/CrU 7) with high beta-2 microglobulin (13 mg/dl; n.v. 0.5-1.6). US detected kidney cortical hyperechogenicity. The UO was 0.8-1.2 ml/kg/h, BP 120/70 mmHg. Drugs were discontinued and rehydration was started with restoration of renal function in 6 days (SCr 0.47 mg/dl) and BP normalization.
Nephrotoxicity is often underestimated. Acyclovir and ceftriaxone are both nephrotoxic drugs causing an interstitial nephritis with tubular necrosis and a combination therapy should be started only if strictly necessary. Furthermore, renal function should be monitored despite commonly used dosages of these drugs are administered.

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## THE CHILD WITH JOINT SWELLING … A

## TEAM GAME

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A 12-year-old boy started complaining of arthralgia and low-grade fever with a positive throat swab for Group A Streptococcus pyogenes. He was treated with a 6-day course of clarithromycine with clinical improvement. After ten days, because of the appearance of bilateral hip pain, the boy underwent hip ultrasound (negative) and blood tests (RCP 1.84 mg/dl). He was discharged on Amoxicillin Clavulanate therapy for 7 days. A few days later, a persistent swelling of the first toe and the first metatarsal of the right foot appeared. No evidence of erythema marginatum, chorea or subcutaneous nodules was present. More comprehensive blood tests revealed ESR 37 mm, RCP 1.7 mg/dl, ASO titer 1023 UI/ml. Cardiologic examination showed minimal aortic regurgitation with no thickened leaflets; suspecting ARF intramuscular Benzathine Penicillin and oral Aspirin were started.
The joint swelling improved slowly and resolved completely within 2 months. For the atypical clinical course of joint swelling compared to arthritis in ARF, cardiac examination was repeated at 6 months confirming the minimal aortic regurgitation described with unclear rheumatic involvement of the valve. Since the cardiologic follow-up at one year from diagnosis showed stable aortic insufficiency without resolution and no mitral involvement, the diagnosis of carditis was excluded by configuring a post-streptococcal reactive arthritis framework; prophylaxis was therefore stopped.

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## UNFORGETTABLE DISEASES: WHOOPING COUGH WITH NO COUGH

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A 24-day-old female infant, with uncomplicated reported perinatal history, arrived at our Emergency Unit for rhinitis and poor feeding, presenting tachypnea and diffuse chest crackles without fever. Chest X-ray (CXR) showed interstitial thickening and air-trapping. White blood cell (WBC) count and C reactive protein (CRP) were normal. She started nebulized hypertonic solution and Ampicillin. On the 1st day of hospitalization she had a respiratory worsening, requiring transfer to NICU for cPAP support. After a further deterioration of respiratory function, mechanical ventilation was started. CXR showed an upper right lobar consolidation. Suspecting chlamydial pneumonia, Clarithromycin was added. After 12 hrs, she presented generalized hypotonia and increasing oxygen requirement. CXR showed pleural effusion; leukocytosis (WBC 78710/mmc) and increased CRP (4 mg/dl) were found. Suspecting HSV disseminated disease, Acyclovir was added. Respiratory function fell until severe pulmonary hypertension (PH) occurred, unresponsive to full intensive care support. She died 96 hrs post-admission, with postmortem microbiological diagnosis of Bordetella pertussis infection.
Diagnosis of neonatal pertussis is difficult, since paroxysmal cough is not common. It may complicate with pneumonia, PH due to hyperleukocytosis, neurological involvement and high mortality. Clinicians should be careful about "forgotten" infectious diseases, since a high index of suspicion is needed to avoid delayed diagnosis.