diseases has been diagnosed as Type - Classic of Hodgkin lymphoma (mixed celularity).

Results The treatment is done according to protocol for Hodgkin lymphona, with three cycles of OEPA (Prednisone, Vincristine, Doxorubicine, Etoposide), and three cycles of COPP (Prednisone, Procarbazine, Vincristine, Cyclophosphamide).After therapy, the PET - scan has been made and its results show that nowhere in the body is present malignant infiltration.

Conclusion OEPA and COPP are very efficient therapy in the treatment of Hodgkin lymphoma.

527 CHEMOTERAPEUTIC TREATMENT OF RHABDOMYOSARCOMA BOTRYOIDE

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Introductions Rhabdomyosarcoma (RMS) is malignant tumor which arises from embryonic muscle cells. RMS is the most common type of soft-tissue cancer in children. It can develop on children at any age, but it's most common in those between 2 and 6 years old and 15 and 19 years old.

Purpose Presentation of the case with RMS botryoides, which was presented to the Hematology/Oncology Unit at Pediatric Clinic and has received chemotherapy treatment.

Materials and Methods A female child, 2 years old, who was initially admitted in Clinic University Centre of Tirana, with presence the mass of tumor in vulvovaginal region. The tumor mass appeared after the birth and it has been increasing in size since then. At University Clinic Center of Tirana the patient has been operated, and the tumor has been removed from her. Detailed Histopathological analyses has shown that the removal part of her was botryoide Rhabdomyosarcoma. The diagnose of the patient was made based on anamnesis, clinical examination, laboratory tests, radiological images, bone marrow aspiratione, histopathological analysis. After the diagnoses it was indicated the treatment protocol of chemotherapy for extracranial rhabdomyosarcoma VAC.

Conclusion The total Resection of the tumor mass, the application of the protocol for RMS botryoide, supportive care, monitoration of the chemotherapy toxicity has resulted in absence of minimal residual disease, which is confirmed by the follow up of her clinical status, laboratory tests, radiology tests, that has resulted the absence of secondary deposits.

528 IATROGENIC TRACHEAL RUPTURE IN A CHILD: CASE STUDY AND REVIEW OF LITERATURE

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Tracheal rupture is rare in childhood and optimal treatment is not clear. A 14-year old boy was admitted to a local hospital after sudden loss of consciousness. He underwent endotracheal intubation and was referred to our hospital. The patient's history revealed that he had voluntarily inhaled butane gas. The physical examination was consistent with coma and cardiogenic shock, and the chest x-ray showed pulmonary edema. The patient was admitted to the intensive care unit, and diuretic and inotropic therapy was started. In the third hour of monitoring of the patient under mechanical ventilation, subcutaneous emphysema and pneumothorax at the right hemithorax were observed without deterioration of the vital functions. Thoracic computed tomography (CT) scan findings were consistent with tracheal rupture. The patient was monitored conservatively without surgery. On the fifth day of hospitalization, his tube was removed, and he was discharged on the twelfth day with a positive prognosis. In this study, a tracheal rupture case after endotracheal intubation is presented in which the patient recovered completely with conservative therapy.

529 OSTEOGENISIS IMPERFECTA IN A DEFINED BIRTH COHORT: REGIONAL EXPERIENCE AND THE NEED FOR A NATIONAL PLAN

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Background and Aims We aim to analyse the clinical characteristics of OI within a 10 year regional birth cohort in Ireland and to highlight the management challenges and the need for a National level referral and treatment structure.

Methods Case notes of all patients with OI from 2002 to 2011 were reviewed for clinical characteristics, radiological findings, results of molecular genetics and the comprehensive management including treatment received abroad.

Results

Case1: Female type III OI born by caesarean section at term noticed to have several fractures in-utero and infancy; now 10 years with short stature & respiratory problems.
Case2: Male type IV OI, born by CS at term. Multiple antenatal fractures noted on postnatal skeletal survey. Presently 6 years old.
Case3: Male type III OI, presented at birth with multiple fractures, and significant ligamentous laxity. Presently 4 years old.
Case4: Female type V OI, born by CS at term. Diagnosed at 9 weeks when presented with fractures. Developed subsequent crush fractures of vertebrae despite being on pamidronate infusions; presently 3 years old.
Case5: Female type IV OI, born by CS at term with antenatal suspicion of skeletal dysplasia. Presently 2 years old with associated developmental delay.
Case6: Male type IV OI, followed up since 27 weeks gestation and intrauterine death following the development of non-immune hydrops at 34 wks.

Conclusion As more children are surviving nationally there is a need for a single specialised and comprehensive paediatric metabolic bone diseases unit providing a structured quality of care and avoiding travel abroad.

530 ACQUIRED MICROCEPHALY IN A CHILD WITH ABSENT SPEECH: HOW TO STUDY?

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Introduction Microcephaly is defined as reduce of the head circumference less than 2 SDS for age, sex and gestation.

Purpose Three different cases of acquired microcephaly and absent speech are presented with the purpose to find the better form to study patients with this clinical association.

Results

Case 1 - Eight-year old girl, has acquired microcephaly since 15 months of age, absent speech, delay motor skills, seizures and angelman-like features. Chromosome 15 study confirmed Angelman Syndrome.

Case 2 - Five-year old boy, has acquired microcephaly since 12 months of age, absent speech, mental retardation, behavioral problems. Creatine Transport Defect was confirmed with high levels of creatine in urine and hemizygous missense mutation in the SLC6A8 gene.
Case 3 - Eleven-months old boy, has acquired microcephaly since six months of age, absent speech, hypotonia. He also has α-1-antitrypsin deficiency, increased lactic acid and T4 low. Cerebral magnetic resonance showed global loss of volume of white matter. Muscle biopsy confirmed respiratory chain disorder with complex 2 deficiency - 25%.

Discussion If the patient showed acquired microcephaly and absent speech associated with convulsions and angelman-like features, the most probably diagnosed is Angelman syndrome. The screening for mutation in chromosome 15 diagnosed the syndrome. If the patient has also behavior disturbances with family history of learning disabilities, determination of urine creatine is obligatory to exclude creatine transport defect. If all these tests are negative and the patient has unrelated organs involved, we need to exclude respiratory chain disorder and muscle biopsy is mandatory.

531 CLINICAL AND THERAPEUTIC ASPECTS OF GAUCHER'S DISEASE IN CHILDREN

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Introduction Gaucher's disease is the most common of the inherited metabolic disorder known as lipid storage diseases. It is a lysosomal disease, autosomal recessive. It is caused by a deficiency of beta-glucocerebrosidase. The result is a substance called glucocerebroside to build up in cells of the body (Spleen, liver, lungs, bones and sometimes in the brain.

There are three clinicals types:

Type 1
95%
1/50000
Subacute
Infants/Children
Doesn't involve the brain

Type 2
1%
1/150000
Acute/Deadly
Newborn-06 months
Severe brain damage

Type 3;
5%
1/100000
Chronic
Juvenil/Adult

Brain-Liver-Spleen involvement appear gradually

Materials and Methods It's a baby 13 months old. He had hepatosplenomegaly with cytopenia. He had the neurological signs such pyramidal syndrome with contra version ocular without flutter.

The exploration concluded for the GD by the enzymatic dosage.

Results After six years of follow up, enzyme replacement therapy (Imiglucerase) has demonstrated its effectiveness as well as biological as clinical.

Our observation has been raised the possibility of signs of brain involvement in the type 1.
The finding joins a few cases in the literature.
This data calls into question the traditional classification cited from above.

Conclusion We emphasize the importance of studying more of cases (Clinic - Genetic) to put an update on the current classification.

The early therapeutic in the management of GD is still advantageous.

532 VACTERL ASSOCIATION: A NEW CASE WITH BIOTINIDASE DEFICIENCY AND ANNULAR PANCREAS

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VACTERL (V - Vertebral anomalies, A - Anal atresia, C - Cardiovascular anomalies, T - Tracheoesophageal fistula, E - Esophageal atresia, R - Renal (Kidney) and defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal dysplasia, and limb anomalies. Less frequent defects seen with VACTERL association are prenatal and postnatal growth deficiency, laryngeal stenosis, ear anomaly, large fontanels, defect of lower limb, rib anomaly, tethered cord, and defects of external genitalia. We report a case of VACTERL association who had concomitant biotinidase deficiency and annular pancreas, which has not been previously reported.

533 CANAVAN DISEASE: A CASE REPORT FROM KUWAIT

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A Kuwaiti female presented at age of three months with macrocephaly, hypotonia, and global developemental delay. She was found to have Cancvan disease. Although Cancvan disease is a rare autosomal recessive neurometabolic disorder which occurs mainly in Ashkanazi Jews, we are reporting this case to highlight that neurometabolic diseases as well as other rare autosomal-recessive disorders affect a relatively large number of patients in countries with high rate of consanguineous marrige like Kuwait and other Gulf areas. We believe that it is high time for molecular cytogenetic studies to be done on Canvan disease and other rare neurometabolic disorders affecting Kuwaiti patients.

534 A CHALLENGING CASE OF MAKING CRITICAL CARE DECISION ON THE WITHDRAWAL OF NEONATAL INTENSIVE CARE

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Background The RCPCH and the Nuffield Council of Bioethics provide guidance on making critical care decisions on withdrawal of intensive care. BLISS also provides guidance for families to work in partnership with professionals when making critical decisions.

Clinical Case Following an uneventful pregnancy, a baby boy was born at 30 weeks gestation to non-consanguineous parents. He was profoundly hypotonic with respiratory distress at birth. He remained ventilated for poor respiratory effort in the weeks following delivery. He failed attempts at extubation. He continued to have paucity of movement. He underwent extensive investigations for hypotonia and was reviewed by external specialists. His clinical picture suggested an extremely poor prognosis. Following extensive discussions and multidisciplinary meetings it was felt that it was not in the baby's best interest to continue with intensive care. Despite prolonged counselling of the parents over weeks regarding palliative care, they insisted on continuing intensive care. They sought an independent neonatal opinion through their solicitor. The opinion of the independent external professional was in