

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  $\alpha$ -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 developmental delay. She was found to have Canvan disease. Although Canvan 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 marriage 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