Methods We describe the clinical presentation, results of laboratory and radiological investigations, treatment and outcome to date.

A review of current available literature on this topic was also undertaken.

Results An 8 year old boy presented to the PED with severe nausea and vomiting with ayclical pressure type right upper quadrant pain for 7 hours. On presentation the pain had reduced significantly. No symptoms of infection, no concerning vomit contents, diarrhoea or constipation.

This was the 10th similar episode in the previous 2 months. Previous investigations including blood panel, urine were normal and symptoms had resolved on attendance.

No abnormality was found on clinical exam.

Abdominal ultrasound demonstrated a large right sided hydrenephrosis secondary to PUJO confirmed by CT KUB. A renogram demonstrated a partial obstruction and surgical management was planned electively.

Conclusion Our patient had experienced multiple episodes of Dietl’s Crisis which had resolved independently. PUJO is not a common first time presentation in children of this age. We suggest that Paediatricians consider this diagnosis when the other more common differentials have been outruled while being mindful that clinical examinations, radiological and laboratory investigations may be normal in between episodes of Dietl’s crises.

Imaging investigations X-Ray Right knee reported florid callus formation surrounding the distal femoral metaphysis, representing an ossifying subperiosteal haematomat along with a bony fragment in relation to the anterolateral aspect of the distal metaphysis in keeping with an avulsion fracture.

Conclusion Based on antenatal, perinatal and postnatal history, revision of maternal case notes, and photographic evidence while being inpatient in the maternity ward, along with the presence of callus formation on X-Ray implied an injury older than 10 days. This suggests that the femoral fracture is most likely due to External Cephalic Version performed 6 days prior to delivery. A decision was made by the paediatric consultant to withhold any further safeguarding investigations as an aetiology for the child’s fracture was detected.

Abstracts

P571 FOETAL FEMUR FRACTURE AND EXTERNAL CEPHALIC VERSION: A CASE REPORT OF METAPHYSEAL INJURY AND LITERATURE REVIEW

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Introduction External Cephalic Version (ECV) is a procedure used to turn a foetus from a breech or transverse position into a cephalic position prior to the onset of labour. It is recommended by the UK national guidelines to enable vaginal delivery for breech presentation of a singleton pregnancy. ECV is considered a safe manoeuvre when dealing with breech presentation and fractured femur is a rare but recognised complication of this procedure, with only 3 cases reported in the specialty literature.

Case We herein report a 4 months old boy, born at 38+4/40 weeks gestational age by Kiwi extraction and episiotomy, with a birth weight of 4180 g, Apgar score 8 at 1 minute and 9 at 5 minutes of life, no active resuscitation was required. The pregnancy was carefully monitored due to the maternal nutritional controlled gestational diabetes and foeto-pelvic unstable lie. Was breech at 37+5 weeks and ECV manoeuvre performed, discharged home in stable condition afterwards. Postnatally paediatric team or parents did not raise any concern regarding pain or swelling of the knee. He was admitted to our Children’s Ward at 10 days of age by a community midwife with suspected non-accidental injury. Physical examination revealed right lower limb in a flexed, antalgic position, non-ecchymotic swelling of the right knee, with tenderness to touch. Full examination revealed no additional injuries. Retrospective review of pictures taken by parents in the first day of life revealed a swollen knee being held in a flexed position.

P572 PARTIAL DOUBLE TRISOMY 9 AND 13-FIRST REPORTED CASE IN MEDICAL LITERATURE

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Background Both trisomy 9p and partial trisomy 13q have been recognised in past with characteristics clinical anomaly, our case is the first reported case of combined partial double trisomy involving chromosome 9p and 13q. Phenotypic Characteristics vary based on the regions of the chromosomes involved and the gene dosages effect. Characteristics of our index case would not only help clinician in genotype-phenotype correlation of any such future cases but would also add up to the already described consequences in offspring of balanced reciprocal translocations in either parents.

Case report A female infant was born at 41 weeks gestation by normal delivery with birth weight 3.3 kgs. The pregnancy was uneventful. Baby had an episode of hypoglycaemia during very first day of life.

Physical examination of the baby revealed profound central hypotonia, head lag, low set ears, depressed nasal bridge and increased nuchal pad of fat. Cardiac examination revealed soft systolic murmur of grade 2/6 which subsequently on echocardiography was noted to arise from a small atrial septal defect. Remainder systemic examination was within normal limits. Further course in the special care baby unit was complicated by recurrent apneas, desaturations and poor feeding. She also developed symptoms of cow milk protein’s intolerance and gastro oesophageal reflux later on in life. Cranial ultrasound, Electroencephalography, MRI of the brain, renal ultrasound, sleep study, Laryngo bronchoscopy, chest and thoracic inlet X-rays were all normal. Array comparative genomic hybridisation, using a 60K Agilent chip showed a gain of chromosome 9 material of approximately 30.9Mb at bands 9p24.3–9p21.1 between base pair coordinates 204193 and 31104204, and an another gain of chromosome 13 material of approximately 11.2Mb at bands 13q12.11–13q12.3 between base pair coordinates 20407295 and 31578124, with the former representing the most proximal probe on this platform. Subsequent analysis of GBand metaphase chromosomes demonstrated an abnormal female karyotype with an additional chromosome consistent with a der(13)(9;13)(p21.1;q12).

Karyotypic analysis of the parents showed that the mother carried a balanced t(9;13) translocations. Therefore the transferred genetic defect in the index case was a product of 3:1...
segregation error of maternal reciprocal translocation t(9;13) (p21;q12).

Conclusion Balanced reciprocal translocations in either parents can amplify and produce unbalanced gamets leading to defective conceptus. Prenatal diagnosis is strongly recommended where balanced translocation is found in parent. Clinical features of the affected conceptus depends largely on the regions of chromosome involved.

A CASE OF INSIDIOUS RECURRENT ABDOMINAL PAIN

Porphyrias are a group of inherited metabolic disorders of heme biosynthesis leading to excessive accumulation and excretion of porphyrins. The variable clinical manifestations may determine a delay in the diagnosis, followed by a possible negative clinical outcome.

We describe a case of a 14-year-old girl with Shwachman-Diamond syndrome (SDS) and epilepsy admitted with severe cyclic abdominal pain not responsive to antalgic therapy, localized in the epigastric area, irradiating to the whole abdomen and the back, associated to aspecific symptoms (diarrhea, fever, vomiting). During hospitalization, the girl was asymptomatic. We excluded infectious, autoimmune, endocrinological causes, a relapse of SDS and performed a screening for porphyria that resulted negative. After each attack, she referred presenting urinary retention followed by hyperchromic urines, anxiety and paresthesias. Past clinical data showed hypotenretremia, tachycardia and hypertension. Eventually, increased values of ALA (8.21 mg/l) and PBG (3.96 mg/l) were found; exposure of fresh urines to sunlight caused a change of their color. Finally, genetic analysis was negative. These findings allowed the diagnosis of porphyria, therefore we prescribed a normocolorihyperglucidic diet and preventive glucose solutions in stressing situations, with regression of symptoms. Acute porphyrias present with life-threatening crisis secondary to the injury of central, peripheral and autonomic nervous system. They can be triggered by drugs, alcohol, infections, reduced caloric intake, endogenous hormone cycles and stressing situations. The characteristic manifestations are severe cyclic abdominal pain, neurological or psychiatric symptoms and/or hyponatremia. Diagnostic is the assessment of plasmatic PBG/ALA, always increased during an attack, normal during remission. Treatment is human haemin although, in mild attacks, a diet with high carbohydrates and/or preventive glucose infusions is effective; a negative genetic evaluation should never exclude the diagnosis.

It is important considering acute porphyrin in the differential diagnosis of severe cyclic abdominal pain, particularly in subjects affected by different and/or rare clinical disorders.