The annual incidence of leukemias in children is 4.5 cases per 100,000 children. Acute lymphoblastic leukemia accounts for approximately 77% of cases. The common presentations in this age group are pallor, irritability and bone pain. Leukaemia cutis can be a rare presenting sign of ALL (3% of cases).

Gitelman syndrome is a rare autosomal recessive renal tubular disorder characterized by hypoglycaemia, metabolic alkalosis, hypomagnesemia and hypocalciuria, most often it is caused by a mutation in the solute carrier family 21 member 3 (SLC12A3) gene. Prevalence is approximately 1/40000 and the prevalence of hetero-zygotes is approximately 1% in Caucasian populations, making it one of the most inherited renal tubular disorders.

We report a case of 7 year old boy who presented to emergency department with 4 days history of abdominal pain. Past history revealed that he always likes salty and pickled food, having intermittent weakness and low energy but no paraesthesia or muscle cramps.

He was born at term with uneventful neonatal course. His development is normal and is fully vaccinated. There is no family history of renal disease, Dad has IDDM, and he has one healthy brother.

On examination he was a slim boy, weight was 21.2 kg, height 119 cm (both between 9–25th centile) his vitals including blood pressure were normal. Systemic examination was normal apart from mild peri-umbilical tenderness. Abdominal X-ray showed faecal loading of colon and us abdomen was normal. He was referred for paediatric assessment by surgical team because of incidental finding of low serum potassium. His venous blood gas showed metabolic alkalosis and serum magnesium was low normal. ECG showed sinus rhythm.

He was admitted and started on intravenous fluids with added potassium chloride as well as oral K supplements. Other investigations including early morning urine, albumin/creatinine ratio, urine for electrolytes, urine amino acids and urine for retinol binding proteins (RBP) were all normal. Blood sample for genetics was sent for Gitelman syndrome.

His oral potassium dose was increased gradually while weaning on potassium supplement in intravenous fluids. He was discharged home on oral potassium after his serum potassium level improved and booked for repeat bloods in day ward. His repeat blood showed low serum magnesium, so he started on oral magnesium supplement. He also needed further increase in his potassium dose to maintain his serum level ≥2.8 mmol/L. His genetics tests confirmed two mutations in the SLC12A3 gene consistent with autosomal recessive Gitelman Syndrome.

Currently he is doing well and under regular follow up with general paediatric and paediatric nephrology.

Although it is a rare disorder, Gitelman Syndrome should be considered in children with unexplained hypokalaemia, with adequate treatment patients usually have excellent prognosis.

**P569**

**DEJERINE-SOTTAS SYNDROME AND CRANIO-FACIAL DYSMORPHISMS: A CASE REPORT**

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**Introduction** Dejerine-Sottas syndrome (DSS) is a rare hereditary motor-sensor neuropathy transmitted as either autosomal dominant or recessive and classified as a severe degenerative neuropathy of the Charcot-Marie-Tooth type.

DSS is characterized by demyelination and remyelination features with an extensive nerve and root hypertrophy that results in a decreased nerve conduction velocity (<10–12 m/s).

The hallmark clinical manifestations develop in early infancy with hypotonia, developmental motor delay and areflexia. Although arthrogryposis and spine deformities are frequent features, there are no direct associations with other dysmorphic features.

**Case** This report describes a rare association between DSS and craniofacial syndrome.

A 5 months old boy first presented in our Clinic with an early onset of motor symptoms manifested by congenital hypotonia, joint laxity particularly involving his lower limbs, failure to thrive, short stature and a significant psychomotor developmental delay.

On clinical examination he showed clear dysmorphic features with epicantic folds, hypertelorism, long philtrum, low set ears, downwarding of the eyes particularly the left eye and a convergent left eye squint.

**Comment** The presence of ‘soft’ clinical signs can distract from typical features of an underlying neurological syndrome leading to subsequent delayed or misdiagnosis in children with DSS.

In our experience, it is therefore important that Paediatricians can be aware of this possible association with this diagnosis and seek expert specialist geneticist advice if suspicious while simultaneously developing a management plan that supports and encourages attainment of maximal developmental process for the child.

**P570**

**AN UNUSUAL CAUSE OF RECURRENT VOMITING IN A SCHOOLBOY: DIETL’S CRISIS**

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**Background** Congenital Pelvic Ureteric Junction Obstruction (PUJO) is caused by an intrinsic stenosis of the PUJ or external compression by an accessory renal artery.

Dietl’s crisis is defined as episodic crampy upper abdominal pain, nausea and vomiting secondary to PUJO. Symptoms can be severe, increasing in nature with diuresis. The patient may also be asymptomatic between events.

Childhood vomiting is a regular presentation to Paediatric Emergency Departments (PED) worldwide.

**Aims** Our aim is to report a case of an eight year old boy who presented to our PED with recurrent episodes of vomiting and abdominal pain, the ultimate diagnosis for which was Dietl’s crisis.
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 a cyclical 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 hydronephrosis 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 haematoma 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.

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

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 chromosome 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 30.9 Mb at bands 9p24.3 and 13q12.11 with the former representing 11.2 Mb at bands 13q12.11–13q12.3 between base pair coordinates 20407295 and 31578124, with the latter representing an additional chromosome consistent with a der(13)(t(9;13)(p21.1;q12)).