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 parent.

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 hypotonia, head lag, low set ears, depressed nasal bridge and low set ears.

Physical examination revealed 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.
A CASE OF INSIDIOUS RECURRENT ABDOMINAL PAIN

1Anita Spirito, 1,2,3Enrica Manca*, 4Claudio Carmine Guida, 1Angela Maggio, 1,3Maria Savino, 3Filippo Ascella, 1Massimo Petticello-Mantovani, 1Savetio Ladogana. 1Department of Pediatrics, Oncology-Hematology Unit, 1Casa Sollievo della Sofferenza' Scientific Institute, San Giovanni Rotondo, Foggia, Italy; 2Residency Program in Pediatrics, University of Foggia, Foggia, Italy; 3Department of Pediatrics, Pediatric Unit, 4Department of Transfusion Medicine and Analysis Laboratory, Casa Sollievo della Sofferenza' Scientific Institute, San Giovanni Rotondo, Foggia, Italy; 3Immunogenetic Laboratory- Department of Transfusion Medicine and Analysis Laboratory, Casa Sollievo della Sofferenza' Scientific Institute, San Giovanni Rotondo, Foggia, Italy; 5Institute of Pharmacology-“Casa Sollievo della Sofferenza’ Scientific Institute, San Giovanni Rotondo, Foggia, Italy.

10.1136/archdischild-2019-epa.907

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, radiating 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 hyperchomeric urines, anxiety and paresthesias. Past clinical data showed hypesymatrinemia, 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 normocalorichyperglucidic 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. Diagnosis 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 porphyria in the differential diagnosis of severe cyclic abdominal pain, particularly in subjects affected by different and/or rare clinical disorders.