TWIST AND SHOUT: DELAYED PRESENTATION OF SKELETAL DYSPLASIA - CASE REPORT OF AN INFANT

CT Angiogram showed upper periaortic soft tissue surrounding the origin of the coeliac axis and superior mesenteric artery (SMA) causing marked stenosis of the SMA.

A working diagnosis of an inflammatory or infective Aortitis involving the proximal intra-abdominal Aorta, extending from the diaphragm to the renal vessels was made.

An extensive infectious work up was performed which proved negative including exclusion of tuberculosis.

C3 and IgA were marginally raised. Serum Anti Nuclear Antibodies, Anti-Double Stranded DNA Antibodies, Rheumatoid factor and urinary catecholamines were all within normal limits.

PET scan confirmed uptake in the proximal abdominal aorta with associated periaortic soft tissue suggestive of Aortitis.

A diagnosis of Takayasu arteritis - large vessel granulomatous vasculitis, was made.

Treatment was instigated with high dose intravenous methylprednisolone for three days followed by high dose oral prednisolone and subcutaneous methotrexate at a dose of 15 mg/m² weekly.

Inflammatory markers slowly began to normalise with immunosuppressive treatment. Follow up ultrasound at one month showed interval improvement in the aortic mass with increase in the aortic lumen size. On corticosteroid wean a further ultrasound 6 weeks later showed no improvement in the mass and was associated with a rise in inflammatory markers. Biologic therapy with adalimumab subcutaneously has since been added with plan for serial imaging to assess response to therapy.

Conclusion This is a rare presentation of a large vessel vasculitis - Takayasu arteritis in a male child of Caucasian origin.

SKELETAL DYSPLASIA - CASE REPORT OF AN INFANT WITH THANATOPHORIC DYSPLASIA

Background An infant with respiratory failure and features of skeletal dysplasia

Case report A male infant dichorionic twin weighing 2.3 Kg with antenatally suspected skeletal dysplasia had initial examination findings of frontal bossing, a flat nasal bridge, a wide anterior fontanelle and shortened upper and lower limbs along with a markedly narrow chest and short ribs. His systemic exam revealed no other abdominal or cardiovascular abnormalities. His twin had normal growth parameters and phenotypic appearance.

He was intubated within a few minutes of life and commenced on high frequency oscillatory ventilation. A skeletal survey confirmed his thorax to be narrow with shortened ribs and handlebar clavicles. He also had vertebral abnormalities with platyspondyly and U-shaped vertebra. His limbs showed generalised micromelia with short bowed femurs and humeri. He had no pneumothoraces.

The infant displayed progressive respiratory failure despite maximum oxygen concentration at 50 hours of life and interventions were discontinued due to futility. His clinical and radiological findings were most in keeping with Thanatophoric dysplasia I and this was confirmed genetically on microarray showing heterozygous FGFR3 mutation.

Discussion Thanatophoric dysplasia is the most frequent form of lethal osteochondrodysplasia with an estimated incidence of 1 in 60,000 births. There are two subtypes: Type 1 is characterised by a short, curved femur, and a straighter femur with cloverleaf skull characterises type II. Affected infants show marked underdevelopment of the skeleton and short limbs due to sporadic mutations of fibroblast growth factor receptor 3 gene (FGFR3). Infants are usually stillborn or die shortly after birth.
after birth from respiratory failure secondary to the narrow chest cavity and hypoplastic lungs.4

REFERENCES

P72 HEMATEMESIS IN A NEONATE; A FACTOR VII DEFICIENCY CASE REPORT

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Introduction or background Hemorrhage in neonates is an alarming sign that requires prompt recognition and management. Causes can be as simple and contained as cephalohematoma to life-threatening bleeding. One of the rare causes is having an underlying factor VII deficiency. It is a rare autosomal recessive disorder that involves disruption of the cascade of the extrinsic coagulation pathway leading to early onset bleeding.

Objectives To describe a case of factor VII deficiency and provide a literature review.

Clinical Case A 3 days old term female neonate, who had an uneventful perinatal course, discharged home and then presented to our facility with significant hematemesis as well as deranged coagulation profile. PT was 78 (ref. 13.5 – 16.4) and INR of 7.44 (ref. 1.05 – 1.35). PT normalized to 13.2 after the ‘mixing study’. Factor VII level was 1.4 (ref. 35–143) very low. She was diagnosed as having factor VII deficiency and given FFP and recombinant factor VII. She was discharged home with subsequent follow ups. Her diagnosis was confirmed by genetic testing.

Conclusion(s) Review of the Literature reveals that there are few reports on factor VII deficiency. This condition is rare and physicians need to have more awareness of it as it is crucial to establish prompt diagnosis and treatment to prevent major complications.

P73 A RARE CASE: THROMBOTIC THROMBOCYTOPENIC PURPURA PRESENTING WITH ACUTE KIDNEY INJURY

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Introduction Thrombotic thrombocytopenic purpura (TTP) is characterized by microangiopathic hemolytic anemia, thrombocytopenia, neurologic abnormalities and renal dysfunction. Acute kidney injury (AKI) is seen very rarely in TTP. In this case report, a case of TTP presenting with severe AKI was reported.

Case A 14-year-old girl was admitted to the emergency department with the complaint of the absence of urine output for two days and side pain. On physical examination, there was no other finding except costovertebral angle sensitivity.

It was learned that she had applied to the hospital with the complaint of absence of urine output nine years ago. She had been diagnosed with atypical hemolytic uremic syndrome. ADAMTS activity could not be studied at that time. Fresh frozen plasma (FFP) infusion was applied and hemodialysis was performed three times. Then the patient was improved. However, the patient did not come for follow-up.

The laboratory findings of the patient in the latest emergency presentation were urea: 211 mg/dl, cre: 8.56 mg/dl, LDH: 5388 U/L, PLT: 103×10^3/μl, and coagulation values were normal. Direct coombs test was negative, and peripheral blood smear revealed schistocytes and fragmented erythrocytes.

The patient was diagnosed with TTP due to low ADAMTS 13 activity (<0.2%). Plasmapheresis and dialysis treatment was performed and also started pulse prednisolone treatment. The platelet count increased to over 150,000 on the 5th day and urine output improved on the 10th day. The patient was discharged with an oral steroid therapy.

Conclusion In this case report, we want to emphasize that severe AKI may rarely occur in TTP patients in childhood. We should evaluate the ADAMTS level of each patient with AKI and hemolytic anemia and regulate the treatment.

P74 PATIENT WITH INTERMITTENT POSTURE ABNORMALITY: AN ALEXANDER DISEASE CASE REPORT

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Background Alexander disease (AD) is a rare neurodegenerative condition defined as fatal infantile leukodystrophy. Among its three forms being described (infantile, juvenile and adult AD), infantile form is the most common form of the disease. Megalencephaly, (which mostly detected in infantile form) demyelination, and multiple Rosenthal fibers are characteristic features of the disease. Gial fibrillar acidic protein (GFAP) mutations have been identified as genetic defects.

Goal We aimed to present diagnostic process of juvenile AD in a male patient with intermittent postural abnormality and a GFAP mutation.

Patient A 12-year-old male patient was admitted to our outpatient clinic with complaints of speech abnormality when he was nervous and a mild hunchback from time to time. His history was unremarkable and his unrelated parents have not any family history of neurological disorders. All laboratory tests, including metabolic scans, were normal. AD was considered due to the localization of the lesions (frontal predominance) detected through cranial magnetic resonance imaging (MRI). Genetic examination revealed a heterozygous GFAP mutation.

Conclusions Rigidity and postural abnormality may be indicative of some neurodegenerative diseases in late childhood and adolescence. Clinical and radiological follow-up is important in the diagnosis of neurometabolic disease. By reporting the current case, we also aimed to draw attention to the fact that postural abnormality may be the first sign of neurodegenerative diseases even when it is not permanent.