after birth from respiratory failure secondary to the narrow chest cavity and hypoplastic lungs.[4]

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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) demyelinization, and multiple Rosenthal fibers are characteristic features of the disease. Glial 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.

Conclucions 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 neuromethabolic 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.