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

REFERENCES

HEMATEMESIS IN A NEONATE; A FACTOR VII PATIENT WITH INTERMITTENT POSTURE ABNORMALITY: A RARE CASE

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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.

PATIENT WITH INTERMITTENT POSTURE ABNORMALITY: AN ALEXANDER DISEASE CASE REPORT

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Introduction 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. Gilral 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.