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Original Article

Quantitative fibrinogen disorders: A retrospective study from a tertiary care centre in India

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Background and objectives: Congenital fibrinogen disorders encompass both quantitative and qualitative abnormalities of fibrinogen, characterised by variable bleeding or thrombotic tendencies. Due to their rarity and varied presentations, delays in diagnosis are common, which can significantly impact patient outcomes. This study was conducted to describe the clinical and demographic profiles of patients with congenital quantitative fibrinogen disorders over a 20-year period (2003-2024) at a tertiary care centre.

Methods: A retrospective analysis was conducted using laboratory records to identify 17 patients with quantitative fibrinogen disorders, characterised by prolonged prothrombin time (PT), activated partial thromboplastin time (APTT), and undetectable or low fibrinogen levels. Only 10 out of 17 patients could be contacted for follow up. The clinical presentation, including scoring of bleeding symptoms using the International Society on Thrombosis and Haemostasis - Bleeding Assessment Tool (ISTH-BAT), as well as demographic variables such as age of onset, first visit to our hospital, and diagnostic delays, were recorded. Treatment and follow up data were recorded.

Results: Majority of patients showed symptoms in the neonatal period, with umbilical bleeding as the most common initial manifestation, followed by post-traumatic and cutaneous bleeding. Despite early symptom onset, a median diagnostic delay of four years was observed. Seven cases were under regular follow up with on-demand therapy. Three patients died during follow up. Most of the cases received fresh frozen plasma.

Interpretation and conclusions: This study describes the clinical profile of quantitative fibrinogen disorders and emphasises the importance of increased awareness, early diagnosis, and standardised management in these patients. Given the significant risk of bleeding, proactive measures such as patient education and close monitoring are crucial.

Keywords Afibrinogenemia; Coagulation disorder; Fibrinogen deficiency; Neonatal period; Umbilical stump bleeding