

Haemophilia B presenting with bilateral cephalhaematomas

*Niraj Kumar Dipak¹, Ruchi Nimish Nanavati¹, Nand Kishore Kabra¹

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Introduction

Haemophilia is the commonest inherited bleeding disorder to present in the neonatal age group¹. Recent studies suggest that 15–33% of newborns with inherited bleeding disorders initially present with bleeding manifestations in the neonatal period². Factor IX deficiency (Haemophilia B, Christmas disease) is an X-linked recessive coagulation disorder. Its prevalence is estimated to be around 1 in 30,000 males. In most neonates with haemophilia B, initial presentation of spontaneous bleeding may be in form of skin bleeding, extracranial haemorrhage (ECH) or intracranial haemorrhage (ICH)^{3,4,5}. Other spontaneous bleeding events reported sporadically were gastrointestinal bleeding⁶, bleeding from spleen⁴ as well as occult bleeding like adrenal and liver haematomas⁷. As far as we are aware there are no reports in the literature of haemophilia A or B presenting with bilateral cephalhaematomas.

Case report

A male child, born to a 24 year old fourth gravida mother, was admitted to our institution on the 9th day of life with gradually increasing swellings over both parietal regions, noticed from the 3rd day of life (Figure 1). The mother has 3 other healthy children, a 5 year old boy, a 4 year old girl and an 8 month old girl respectively. The mother had a full term normal vaginal delivery with an uneventful antenatal period. Birth weight was 2.5 kg. The baby cried immediately after birth and the Apgar score was 9 at one minute. Vitamin K was given to the baby at birth. Examination revealed bilaterally symmetrical soft, fluctuant swellings measuring 3.5–4 cm in diameter over the parietal regions

¹Department of Neonatology, Seth GS Medical College and KEM Hospital, Acharya Donde Marg, Parel, Mumbai 400012, Maharashtra, India

*Correspondence: neonatafundas@gmail.com

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Figure 1: Bilateral cephalhaematomas

*Permission given by parents to publish photograph

suggestive of cephalhaematomas. The child was pale but haemodynamically stable. There was no evidence of bleeding from any other site. X-ray skull did not show an underlying linear skull fracture and there was no evidence of intraventricular / intra-parenchymal haemorrhage on ultrasonography of the cranium. There was no family history of bleeding tendencies.

The haemoglobin level was 9.8g%, packed cell volume 31%, total leucocyte count 9,500/cu mm and platelet count 280,000/cu mm. The prothrombin time was 12.6 seconds, activated partial thromboplastin time (aPTT) 130 seconds and the thrombin clotting time 20.4 seconds. In view of the prolonged aPTT, factor assay was performed. Factor VIII was 58.7% and factor IX was less than 1% indicating a severe deficiency of factor IX. Factor IX concentrate transfusion was given (40 IU/kg) every third day till 21 days of admission. He was discharged at 1 month of age and was advised to come for follow-up.

Discussion

Early diagnosis of haemophilia in neonates is very important for appropriate management, early prophylaxis, parental education and for minimizing the risk of mortality. Timely diagnosis of haemophilia in neonates poses a challenge in cases where there is no family history. The difficulties in diagnosis are mainly due to failure to identify the

presence of abnormal bleeding in neonates, which is often different from that typically observed in older children with haemophilia. In addition, diagnostic investigations are complicated by physiological differences in the neonatal haemostatic system². Both intracranial haemorrhage (ICH) and extracranial haemorrhage (ECH) are observed in neonates with haemophilia and the incidence of ICH in neonates with severe haemophilia is estimated to be 1–4%^{8,9}. In a literature review, ICH was more common than ECH and the most frequent site of haemorrhage was subdural⁹. Both ECH and ICH are associated with a significant risk of mortality as a consequence of massive blood loss even in apparently normal infants¹⁰. Anaemia and jaundice are the most frequent clinical manifestations in cases of ICH and ECH, followed by shock and hypotension⁹. However, in our case the newborn presented with bilateral cephalhaematomas without severe pallor, icterus or shock. The significance of ECH should not be underestimated as it can be life-threatening due to massive blood loss resulting in hypovolaemic shock and carries a significant risk of mortality even in apparently normal neonates¹⁰. In neonates presenting with massive cephalhaematomas without shock, anaemia, trauma or instrumentation at the time of delivery, the possibility of bleeding disorders should be borne in mind.

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