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Haemorrhagic infarct in a neonate with Christmas disease (Hemophilia –B) – A Case Report

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Abstract

Objective: To report the case of a newborn admitted to a tertiary care hospital Krishna Institute Of Medical Sciences, Karad and describing the clinical, laboratory and final diagnosis of hemophilia.

Case study: Male newborn Preterm infant, without perinatal events, showing ecchymoses at sites of venous puncture, cephalohematoma during hospital stay. After testing for coagulation factors, he was diagnosed with factor IX deficiency: mild hemophilia B or Christmas disease. He was discharged after clinical improvement.

Discussion: The presence of abnormal bleeding in newborns should always raise the suspicion of the diagnosis of hemophilia, since the early diagnosis of the disease is of very much importance in the prevention of hemorrhagic events.

Introduction

Hemophilia is one of the common hereditary bleeding disorder that may lead to severe life threatening bleeding. Most hemophilic patients experience no problem during the neonatal period, but bleeding including intracranial hemorrhage leading to severe mortality and morbidity may rarely be observed during this period. Early diagnosis is very important in terms of prognosis in these cases. However, the diagnosis is usually delayed due to confusion of clinical signs of intracranial bleeding with other conditions in the newborn period, absence of family history in 30% of cases and the lack of information about the mother being a carrier of hemophilia.

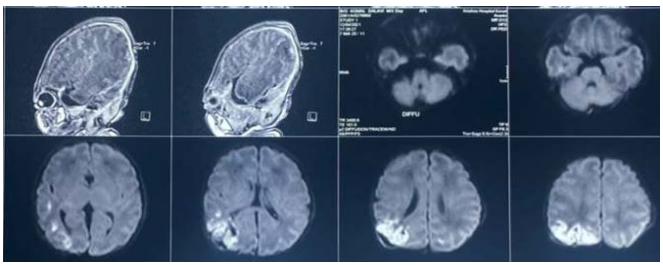
In this case report, we are reporting a newborn diagnosed with intracranial infarct with hemorrhagic conversion (ICH) on the 2nd postnatal day, who was shifted in intubated state to nicu due to respiratory arrest due to ICH. The newborn was diagnosed with mild hemophilia with no familial history. We aimed in showing out the fact that mild hemophilia could also rarely lead to intracranial hemorrhage in term newborns and early diagnosis and replacement treatment were of utmost importance.

Case

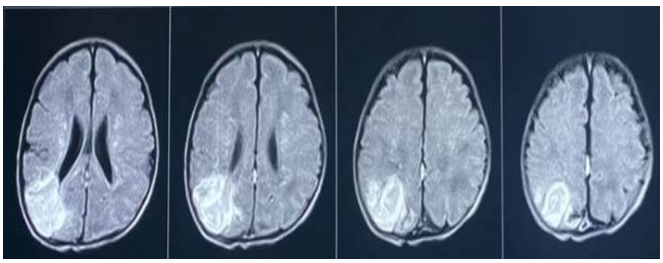
We report the case of a newborn male, born to a 22-year-old G1P1 mother at 34 weeks gestation through vaginal delivery, without any postpartum events , with Apgar scores at 1 and 5 min of 7 and 9, respectively, weighing 2358 g, which is appropriate for the gestational age. A tumor in the right parietal lobe suggestive of cephalohematoma was seen. He was transferred with his mother to obstetrics ward for

regular neonatal care. Vitamin K was administered at birth and breastfeeding was started.

At 20 hours of life of life, baby had sudden breathlessness following feed associated with cyanosis of peripheries, so baby was shifted to neonatal intensive care unit of Krishna hospital Karad in intubated state i/v/o ?aspiration. Baby had spontaneous activity and heart rate of 145 bpm, normal blood pressure (81/47 mmHg),oxygen saturation 95% on room air, hence was extubated and put on bubble cpap, Chest xray ruled out aspiration. Full blood count, coagulation study, blood chemistry and blood culture were performed. There was a 4 cm increase of head circumference (39 cm) since the day of delivery. Except for the bruises, no other findings were seen. Neonatal, pulse oximetry and red reflex test were performed, with normal results. Laboratory test results revealed: hemoglobin 10.1, hematocrit 31.2%, platelets 166,000, leukocytes 2500(23/75/00/02). Blood chemistry was unremarkable at first with a steady increase in indirect bilirubin. On day 3, Baby was noticed to have decreased activity and anterior fontanella was wide open boggy, NSG brain showed intraparenchymal haematoma in right parietal region, with periventricular flaring. MRI was done which showed acute infarct with hemorrhagic conversion in right parietal, temporal and bilateral occipital gyri, acute subdural haemorrhage along right parietal convexity was also noted. Patient's coagulation parameters were evaluated, PROTHROMBIN TIME 22, INR 1.9, PLASMA D-DIMER 0.70MG/L for which ffp was transfused.

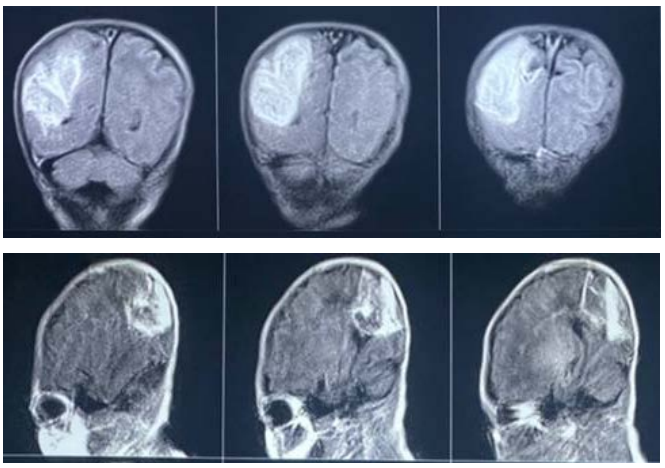


Multifocal areas of altered signal intensity noted involving right parietal and right temporal and bilateral occipital gyri, appearing hyperintense on T2WI, isointense on T1W1, hyperintense on DW! And hypointense on ADC equence (showing diffusion restriction) and showing areas of blooming on hemo- suggestive of acute infarct with hemorrhagic conversion.



Small acute subdural hemorrhage noted along right parietal convexity (maximum thickness – 4mm)

Acute infarct hemorrhagic conversion in right parietal , right temporal and bilateral occipital gyri.



On day 4, Baby had repeated episodes of desaturation and convulsion for which baby was intubated and was kept on IPPV mode of ventilation. Baby was started on anticonvulsants. Gradually baby improved and was

weaned off from ventilator, repeat mri brain after 10 days showed new acute infarct and decreased previous subacute infarct with haemorrhagic conversion. The diagnosis of factor IX deficiency was confirmed: mild hemophilia B after coagulation factor testing (factor IX 27.8% and factor VIII 57%) baby was gradually started on feeds and was discharged once stable. Baby was doing well without any episodes of convulsion since discharge.

Discussion

This patient had no family history of the disease, similar to nearly all patients with hemophilia, as 30% of hemophilia cases result from random mutation and a positive family history is not necessary. In cases of hereditary transmission of the disease, women tend to be only carriers, whereas men develop the pathology.

The disease is often diagnosed in infants when they begin to move about, due to the subsequent traumas, considering the low rate of suspicion during the neonatal period. Abnormal bleeding (spontaneous or iatrogenic) in newborns suggests the possibility of hemorrhagic disorders and should be promptly investigated to ensure that patients who have the disease begin treatment as soon as possible. In this report, the neonate presented with (cephalohematoma). Cephalohematomas can lead to death from hypovolemic shock if they continue to bleed. The patient also presented with other types of bleeding, which are frequently reported and helped assist in diagnosis, such as muscle hematoma hemarthrosis, ecchymosis, and bruising in venipuncture sites.

Laboratory assessment of the coagulation system is also important in the investigation of hemophilia. Prothrombin time (PT) evaluates the extrinsic pathway of the coagulation cascade and PTT evaluates the intrinsic pathway. In patients who present with bleeding with normal platelet levels, normal PT, and increased PTT, factor VIII, IX, or XI deficiency or the presence of the intrinsic pathway inhibitor should be suspected. The reference values are known to vary between laboratories. In this case, factor IX deficiency confirmed the diagnosis of hemophilia B.

After diagnosis, treatment on demand or prophylaxis should be initiated. Prophylaxis is meant to prevent new articular lesions; it consists of regular administration of coagulation factors and may be continuous (at least 45 weeks of treatment) or intermittent (<45 weeks of treatment). It is associated with better outcomes compared with treatment on demand when hemorrhage is occurring or to prevent an event, as in the case of pre-operative administration. Treatment is currently based on the replacement of the absent CFs from purified human plasma. Reposition of factor IX in hemophilia

B is 20–40 IU/kg/day, and its half-life is 18–24 h; it is therefore administered two to three times per week, also according to the severity of the disorder and the lifestyle of each patient. The development of products with a prolonged half-life has reduced the demand for treatment in many patients. In patients with hemophilia B, therapeutic levels can be obtained with replacement every 2 weeks. However, the high cost of prophylactic therapy and the risk of bleeding due to low coagulation factors have encouraged research in the area of gene therapy. Better-quality recombinant concentrates with increased kinetics are currently gaining popularity, and in the future, gene therapy is expected to allow patients to produce the deficient CF. In this way, techniques using viral vectors for the gene have been used with good therapeutic results, low toxicity, and initially without inhibitor emergence and are promising. Thus, early diagnosis allows anti-bleeding prophylaxis to be

initiated through regular replacement of coagulation factors, thereby reducing potential complications and providing a better quality of life for the patient. Neonatologists' role in recognizing signs suggesting abnormal bleeding in neonates is critical, especially in those with increased PTT only, and they should always bear in mind that a positive family history of hemophilia is not always necessary and that in more severe cases, massive intracranial hemorrhages may develop, which can lead to death in newborns.

Conclusions

After reviewing existent literature we conclude that our patient had a rare presentation of hemophilia. He presented with intraparenchymal haematoma in right parietal region after an uncomplicated vaginal delivery. Hemophilia diagnosis was made within 2 days after presentation and treatment towards hemophilia B a started, with no other bleeding episodes during inpatient stay. Imaging studies showed no other complications from the initial presentation and a normal neurodevelopment was achieved up to date. This case report reminds that hemophilia can present in several different ways. When facing a significant bleeding on a neonate with no risk factors, a blood coagulation disorder must be thought along with immediate aggressive support measures. An early diagnosis allows to an early directed treatment and better outcome.

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