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(CASE REPORT)

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Neonatal severe haemophilia: A case report

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Abstract

Hemophilia A is an inherited bleeding disorder caused by deficiency of coagulation factor VIII. It is transmitted in an X-linked recessive pattern from female carriers to male children.

We report the observation of a severe hemophilia A in a male newborn with a history of hemophilia in the siblings who present hemarthrosis of the left elbow. The diagnosis was suspected and confirmed through biologic investigations and imaging. Therefore, patient was put under factor VIII with good evolution.

It is a congenital hemorrhagic disease related to deficiency in anti-hemophilic factor. The severity of the bleeding syndrome is correlated to the factor level. There are three varieties of hemophilia: severe, moderate and minor. The main complications are hemarthrosis, hematomas of variable location and externalized hemorrhages. A favorable prognosis depends on rapid diagnosis, monitoring and immediate medical treatment.

Diagnosis of hemophilia A should be rule out in all male newborns with a family history of hemophilia, and suspected in the presence of cord bleeding at birth or during circumcision.

Keywords: Hemophilia; Neonate; Factor VIII; Hemarthrosis

1. Introduction

Hemophilia A is a bleeding disorder caused by a deficiency of coagulation factor VIII (FVIII). Hemophilia is a genetic disease of recessive transmission linked to the X chromosome. It therefore affects boys and is transmitted by women, who are therefore called carriers [1]. Hemophilia, although rare, remains the most common congenital coagulopathy. We report a case of severe hemophilia A in a newborn with a history of hemophilia in the sibling complicated by elbow hemarthrosis.

2. Case report

This was a male newborn at term through cesarian section on a scarred uterus. The mother was 39 years old and had a brother with hemophilia A. The newborn was hospitalized at 2 days of age for jaundice due to ABO incompatibility mother O+ newborn B+. The biological workup showed anemia at 9.4, total bilirubin at 92, indirect bilirubin at 87, prothrombin rate at 65%, activated partial thromboplastin time at 3 seconds, negative c-reactive protein, transfontanellar and adrenal ultrasound were without abnormalities. The coagulation factor assay showed an FVIII activity of 0.1% in favor of severe hemophilia A. At 10 days of age, the newborn presented a warm and painful elbow joint when mobilized (fig 1). Ultrasound of the elbow was in favor of an elbow hemarthrosis; the baby received intravenous factor VIII with good clinical evolution.

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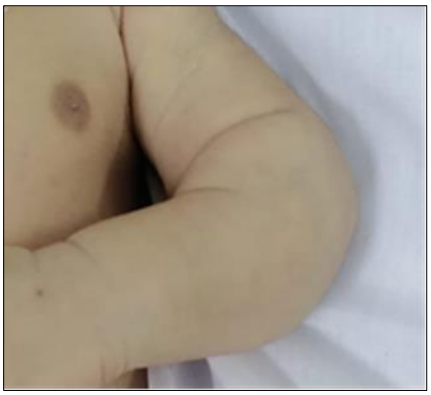

Figure 1 Hemarthrosis of the elbow in a newborn with hemophilia A

3. Discussion

Hemophilia A is a congenital bleeding disorder related to antihemophilic factor VIII deficiency. It affects more than 1.2 million individuals (mostly boys and men) worldwide. Hemophilia A occurs in all ethnic groups and throughout the world. It occurs in approximately 1 in every 4000 to 1 in every 5000 live male births [1].

Diagnosis of hemophilia A requires confirmation of a factor VIII activity level lower than 40% of normal (< 0.40 IU/mL). Therefore, according to the depth of the deficiency, three varieties of hemophilia can be distinguished: the first one is severe hemophilia: characterized by a profound deficiency of antihemophilic factor, the level of factor VIII or IX is less than 1% (compared with the normal level, which is 0.1 mg/L of plasma for factor VIII and 5 mg/L of plasma for factor IX), these patients bleed spontaneously and are particularly prone to intramuscular and joint hemorrhages; the second one is moderate hemophilia in which the level of factor VIII or IX is between 1 and 5%, in this type hemorrhagic bleeding is more moderate and usually secondary to trauma; the third category is minor hemophilia where the level of factor VIII or IX is between 6 and 30%, in this case, hemorrhagic accidents are rare and only provoked and rarely detected at birth [2]. Our patient had severe hemophilia with a factor VIII activity of 0.1%, discovered after a systematic investigation in the presence of a family history of hemophilia.

The differential diagnosis of hemophilia includes von Willebrand disease, factor XI deficiency or factor XIII deficiency and inherited platelet disorders. Nicolau syndrome, or embolia cutis medicamentosa, is another condition that would be considered in the differential diagnosis as a complication of intramuscular injection in neonates[3].

The main complications of neonatal hemophilia are central nervous system hemorrhage. Diagnosed by routine transfontanellar ultrasonography, some studies recommended this exam in all neonates before discharge.[4] Cranial magnetic resonance imaging and computed tomography (CT) scan should be performed just in symptomatic neonates with intracranial hemorrhage. other complications are cephalohematoma, and sites of medical interventions including circumcision, heel sticks, and venipunctures or hemarthroses affecting in particular the knee, the ankle and the elbow, knowing that all the joints can be affected, the hematomas can be serious either because of their location engaging the vital or functional prognosis, vascular or central nervous system compression or because of their size. More rarely, externalized hemorrhages: digestive hemorrhages or hematuria. [5,6].

Treatment is mainly based on the administration of antihemophilic factor VIII [7,8]. The rapidity of the diagnosis, the monitoring and the immediate implementation of the medical treatment condition a favorable prognosis [9,10]. In our observation, the newborn presented a hemarthrosis of the elbow and received factor VIII in emergency, with good clinical improvement.

4. Conclusion

Hemophilia A is a rare congenital disorder, but bleeding can be severe and life threatening.

Prevention involves ultrasound diagnosis of sex before birth in families at risk and administration of vitamin K to the mother prepartum to avoid hypovitaminosis.

Compliance with ethical standards

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Disclosure of conflict of interest

The authors declare that they have no conflict of interest.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study

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