Case 2

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A 1-day-old baby is born through emergent C-section at 36 weeks of gestational age due to sudden signs of fetal distress. At birth, he presented bleeding from the umbilical stump and epistaxis. Apgar score was 3/6, with the need for mechanical ventilation. There is familiar history of one brother deceased after birth due to prenatal brain bleeding of uncertain etiology and another healthy brother. Initial platelet count and coagulation screening tests were normal.

Questions

1. Describe the imaging findings in A and B.
2. What does the lesion in C (white arrow) suggest?
3. Taking into account the clinical information and ultrasound, what would be your main differential diagnosis?

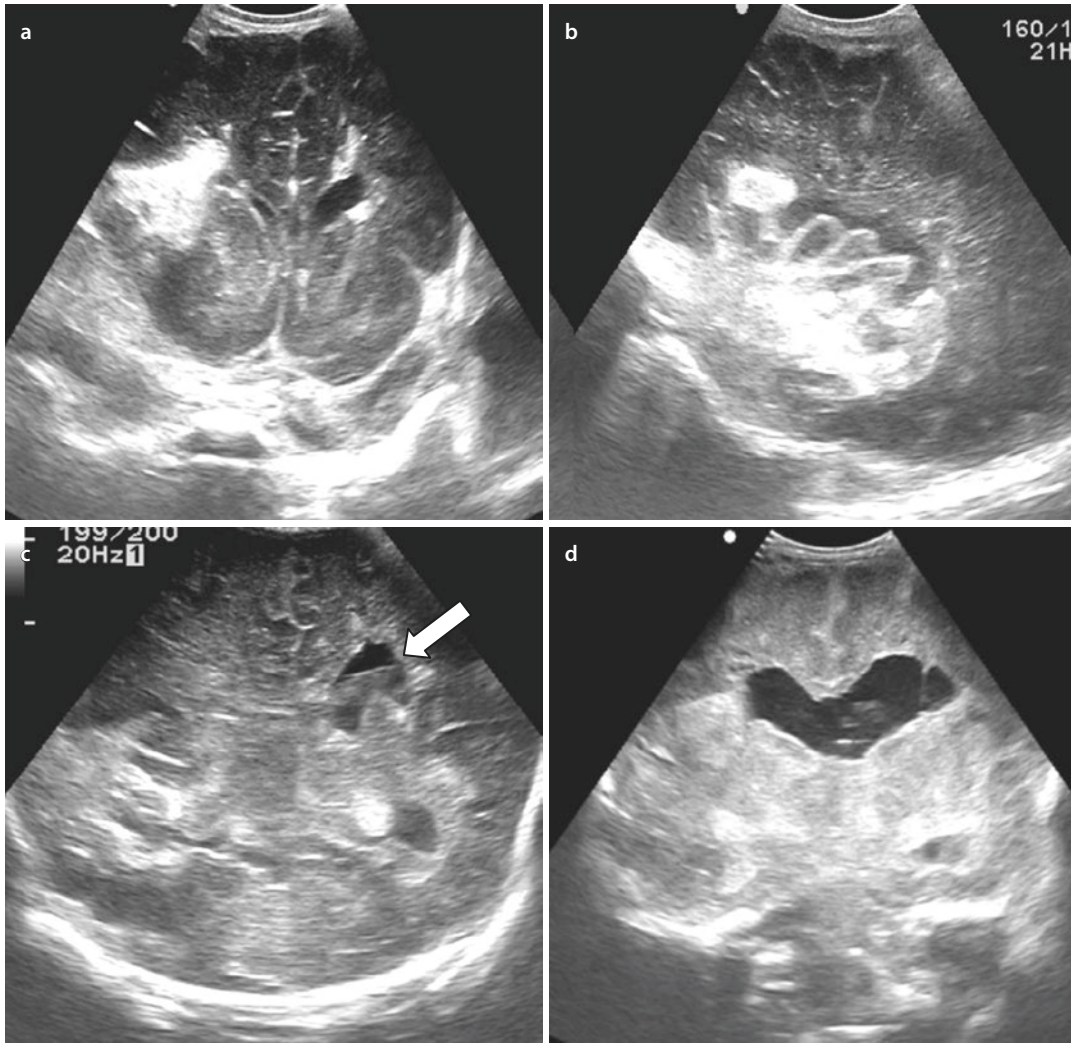

Fig. 2.1 Head ultrasound **a** and **c** coronal and **b** sagittal images at day 1. Coronal image of head ultrasound performed at day 7 **d**

Diagnosis Congenital factor XIII deficiency

Answers

1. Bilateral acute/recent parenchymal hemorrhagic lesions, without evidence of intraventricular blood. The one on the right has ruptured to the subarachnoid space.
2. Left hemisphere old periventricular lesion, with cavitation and marked irregularity of the ventricular contour.
3. Congenital bleeding disorders, including vasculopathies, coagulopathies, and metabolic disorders.

2.1 Comments

Congenital FXIII deficiency is a rare genetic bleeding disorder, with an estimated prevalence of 1 case per 2–3 million individuals. It affects males and females equally and is inherited as an autosomal recessive trait.

The symptoms of factor XIII deficiency have variable severity and may become apparent at any age, although most cases are identified during infancy. Common symptoms include bleeding from the umbilical stump, epistaxis, muscle hematoma, subcutaneous bleeding, and high risk of miscarriage. About one third of patients present intracerebral bleeding, a risk greater than in other related bleeding disorders and the leading cause of mortality [1].

The diagnosis of this disorder is a challenge, not only due to its rarity but also because typical coagulation screening and platelet function tests are normal.

Intracranial hemorrhage in neonates is not a rare finding and can occur as a result of prematurity, birth trauma, platelet disorders (congenital, alloimmune), coagulation disorders, infections, and hypoxia.

In this case, the relevant familiar history and the presence of brain lesions of different ages (◘ Fig. 2.1a and c) suggested a genetic inherited etiology. Differential diagnoses include congenital coagulopathies and vasculopathies but also some metabolic diseases, such as congenital disorders of glycosylation and mitochondrial respiratory chain defects.

Reference

1. Bertamino M, et al. Diagnosis and management of severe congenital factor XIII deficiency in the emergent department. Blood Transfus. 2015;13:324–7.

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