Congenital factor XIII deficiency in the heterozygous twins

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Introduction :

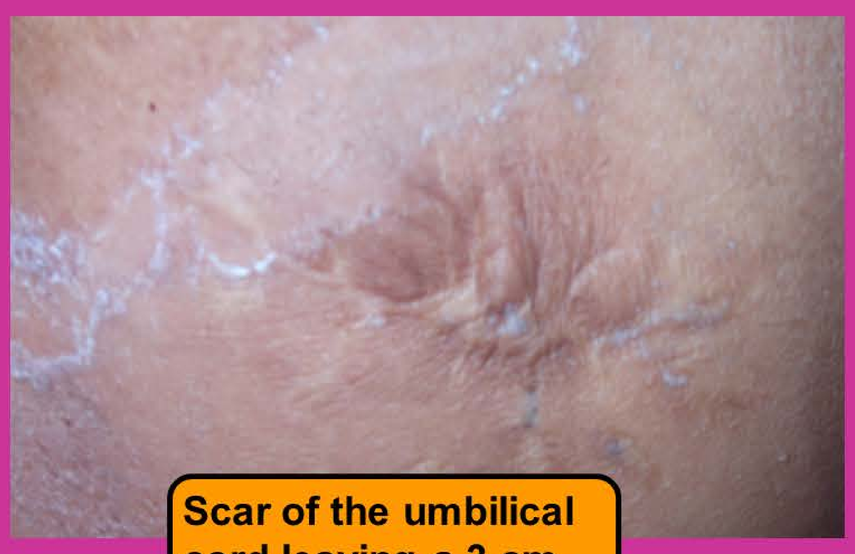
Factor XIII deficiency is suspected in a heterogeneous hemorrhagic table, associated with normal standard blood tests. Frquency is very rare as 1 case / 2-3 000 000 and autosomal recessive transmission. F XIII deficiency was first described in 1961 by Duckert .

Observation:

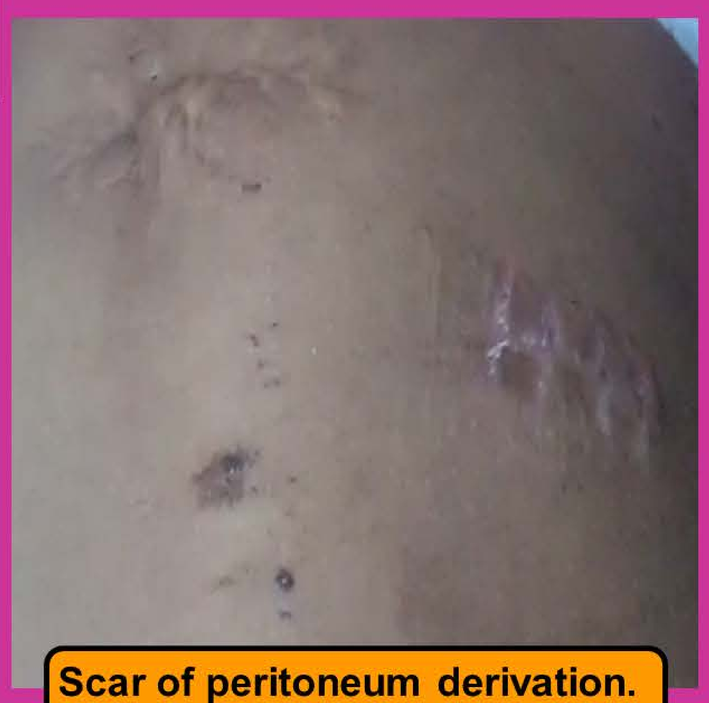
A girl of 6 years who has a brain hemorrhage after surgery. Questioning the mother is the notion of 2 nd degree of consanguinity among parents, caesarean section gives a birth of a heterozygous twin Apgar 9/10, which found bleeding history hemorrhage of the umbilical cord leaving a 3 cm scar, subcutaneous hematoma, spontaneous bruising and post-traumatic, nose bleeding minimal abundance. The twin has the same hemorrhagic symptoms in addition to bleeding post-circumcision for boy. The girl was operated at the age of 7 months for setting up a peritoneal valve for encephalomyelitis. She was hospitalized at the age of 6 years in neurosurgery for valve dysfunction , post- operative follow are marked by the appearance of an intra-dermal parenchymal hemorrhage in bilateral plantar flexion and galial hematoma, specialized neurological examination revealed signs intracranial hypertension. The scan shows a parenchymal hematoma 34x37x46 mm right parietal, bilateral occipital horns hematic flooding of both ventricles and subdural hematoma 17 mm thick.

Laboratory tests

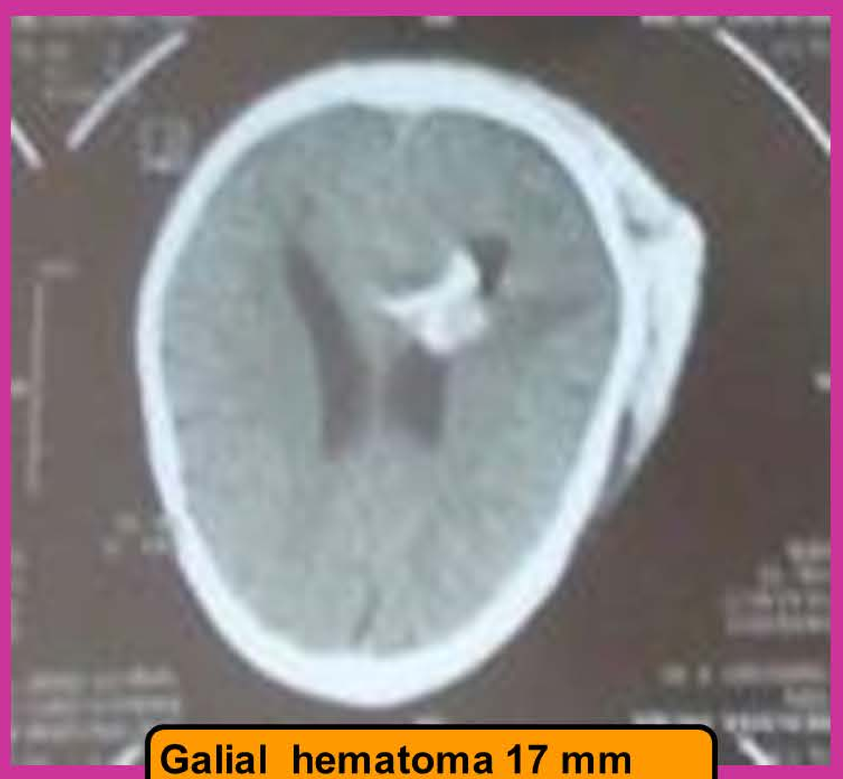
A normal rate of platelet 3750.000/mm3 and VPM 9.3 fl , research for any Thrombocytopathy by flow cytometry back normal. The classical coagulation tests found : APTT = P 26 (T : 30 sec) , TP : 90%, fibrinogen : 3 g / dl are normal The dosage of factor XIII found a decrease of plasma level at 25 %.



Scar of the umbilical cord leaving a 3 cm.



Scar of peritoneum derivation.



Galial hematoma 17 mm

Discussion:

1- The deficit of Factor XIII is manifested by a variety of symptomatic hemorrhagic :
- Bleeding of umbilical cord (81%).
- Hematoma (67%).
- Post- traumatic bruising (90%).
- Intracranial hemorrhage (23%).
- Hemorrhage a circumcision found in the boy.

2 - Delayed healing of the umbilical cord found was found fall in two children, very evocative. Many substrates in the blood circulating in the vessel wall, including fibronectin, von Willebrand factor, vitronectin , collagen, the lipoprotein activate Factor XIII. The Factor XIII facilitate repair of damaged tissues .

3 - lack of parallelism between the hemorrhagic symptoms is the degree of the deficit of factor XIII. Our patient had an obvious hemorrhagic syndrome despite a rate of 25% of Factor XIII.

4 Twins despite heterozygous (bi - chorionic) must have reached in this case .

5 - Since 1970, an F XIII concentrate obtained from human placenta (fibrogamine). In our patient is to use fresh frozen plasma to treating of hematoma girl, with a good clinical outcome.

6 - In terms of molecular biology, polymorphism Val34Leu could have significant clinical consequences. Indeed, the Leu allele may be protective for the occurrence of arterial and venous thrombosis , and increase the risk of primary brain hemorrhage. May be the case for our patients.

Conclusion:

The factor XIII deficiency is a rare constitutional coagulopathy à évoquer a cerebral hemorrhage in the newborn or infant and do not hesitate to make a simple examination of the umbilicus cord or any scar that will quickly guide our diagnostic and therapeutic management.

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