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CASE REPORT

Case Report: Keloid Scar in A Rare Congenital (Inherited) Coagulation Factor Deficiency

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Abstract:
Factor XIII deficiency is one of the rare bleeding diathesis, the presentation of which varies from multiple ecchymosis, hematomas, poor wound healing, spontaneous abortion to even a sever life-threatening intracranial hemorrhage. A rare manifestation of this disorder is excessive scar formation and keloids. Here we report a case of a 35 years old lady who we diagnosed to have Factor XIII deficiency as she presented with an excessive scar formation, keloid, following a burn trauma

Key Words: *Factor 13, Deficiency, Bleeding, Disorder, Keloid*

Introduction

Factor XIII deficiency is one of the rare yet sever causes of bleeding disorders. It could either be inherited as an autosomal recessive disease or acquired in some cases. The clinical manifestations of such deficiency may vary from a delayed umbilical stump bleeding early in life, to a subcutaneous, intramuscular to even intracranial bleeding, as well as impaired wound healing, menorrhagia and spontaneous abortion in pregnancy [1]. However, a very rare manifestation of this disease is excessive scar formation, Keloid, and it's been seen in some patients.

Case Report

This is a 35 years old, Saudi lady, who's a known case of combined iron and B12 deficiency anemia, as well as PCO (polycystic ovarian disease), which she was diagnosed with following a prolonged history of menorrhagia; and is on hormonal therapy and iron supplements since her diagnosis. This patient presented to us complaining of a brown, hypertrophic lesions on both of her shoulders and left arm following a history of a burn trauma. The lesions were extending beyond the original boundaries of the initial burnt areas; they were firm, painless, non-itchy, and had irregular margins [Figure1, 2]. Apart from having a family history of keloid scar affecting her father, the patient didn't report any previous history of excessive bleeding with traumas, dental procedures, bruises or hematoma, and not even a familial history of a known bleeding or coagulation disorders.

Her laboratory workup showed the followings, a hematocrit of 27.7%, Hgb of 8.7, WBC of 11.5 K/Ul with normal differentials, platelet count was 445K/Ul. Her PT was 13.7 sec, PTT was 29.7 sec, PFA collagen/EPI was 100 sec (82-150 sec). Her iron profile showed serum iron < 10 ug/dl, Ferritin of 2.46 ng/ml, and TIBC of 495 ug/dl. The

patient was also screened for factor XIII deficiency after narrowing down the differential diagnosis, and the measurement of that factor was as low as 56.6% (70-140%).

The patient's keloid was managed with intralesional steroid injection and cryotherapy in private hospital, and the scar has partially improved in terms of size reduction since then.

Discussion

Factor XIII is one of the essential factors in the clotting cascade to secure a normal homeostasis. This factor has also a role in the process of wound healing and tissue repair. Factor XIII is mainly found in the plasma, platelets and monocytes [2]. Composition wise, this factor consists of two, non-identical polypeptide subunits, known as "A-chain" and "B-chain". Together, these two subunits form a tetrameric molecule of two units of the A-chain and two other units of the B-chain. Activation of factor XIII requires thrombin and calcium to aid the exposure of an active cysteine residue on the A-chains and the subsequent dissociation of the B-chain dimer to ultimately having this factor catalyse the formation of covalent bonds between the gamma chains and the alpha chains of fibrin. The end result of this process is the formation of a clot that is both mechanically stable and resistant to the degradation by fibrinolysis [3].

As mentioned earlier, deficiency of factor XIII can either be congenital or acquired sometimes during the patient's life. The congenital deficiency is inherited in an autosomally recessive manner; and it's a rare disorder, affecting only one in 3-5 millions people [4], and accounting for about 6% of all of the rare bleeding disorders [5]. The deficiency of this factor can manifest at different stages of life, but in 80% of the affected people, it often presents early during the neonatal period with umbilical stump bleeding just a few days after birth. To a lesser extent, it can present with a severe life-threatening intracranial hemorrhage, reported in 25-30% of the cases. However, the latter presentation comprises a more frequent manifestation in Factor XIII deficiency than what's seen with haemophilia A and B [4]. Other common manifestations of this disorder include delayed wound healing and recurrent spontaneous miscarriages. On the other hand, excessive scar formation, or keloid, is quite a rare manifestation.

A retrospective analysis of 17 patients with Factor XIII deficiency over a period of 20 years, done in the year 2002 at a tertiary hospital in Riyadh, Saudi Arabia, showed that 71% of the cases were first manifested with ecchymosis and recurrent hematomas. Another 55% of the patients, who were males, presented with bleeding after circumcision, and about 41% presented with umbilical stump bleeding. Poor wound healing and keloids were seen in only 3 patients accounting for a total of 18% of the cases, and an equal percentage was also seen with intracranial hemorrhage, where 3 patients presented with that. Other manifestations, seen in one patient each, were cephalhematoma, abortion, abruptio placenta, and intraperitonial bleeding [6].

Diagnosis of factor XIII deficiency is made based on the measurement of its quantitative activity. Clotting solubility test may also be used, the finding of which is having a stable clot of more than 24 hours in case of factor XIII deficiency. Other tests such as measurement of aPTT and PT can't be incorporated in the diagnosis as the results in these patients are often normal. Molecular tests are also available, however, not quite needed to establish the diagnosis [8].

Conclusion

Factor 13 deficiency with keloid scar is a rare manifestation, thus, the clinician is likely to miss it. One should keep in mind the possibility of this rare disease when the patient presents with a poor wound healing or a keloid scar after an injury. The occurrence of keloid scar in our patient may reflect the poor quality of the clotting cascade, associated with the loss of the tensile strength of the fibrin polymers, caused by factor XIII deficiency and leading to an abnormally large scar formation.

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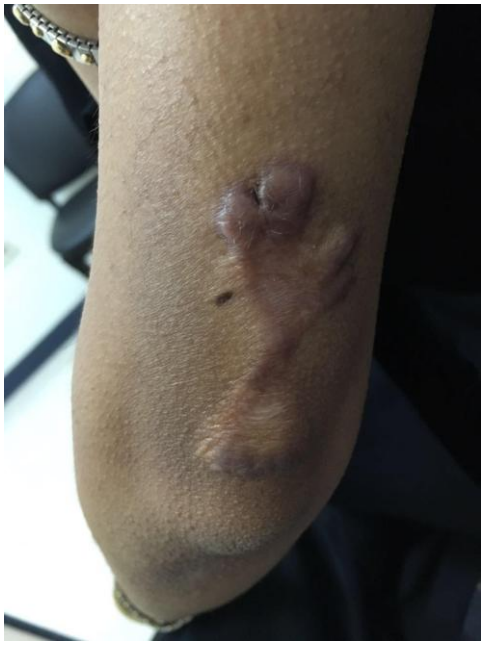

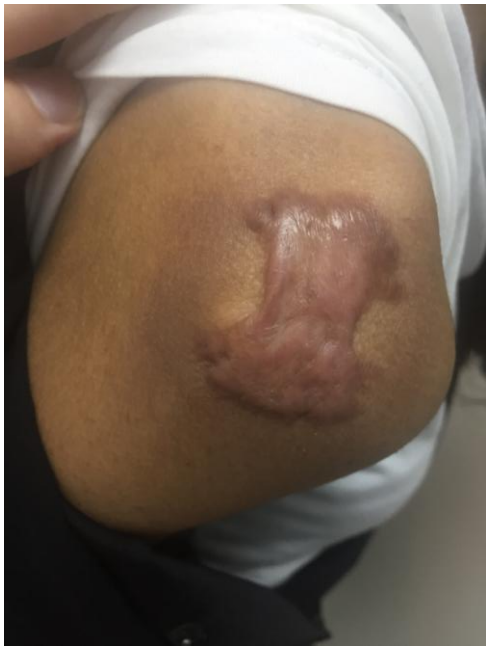

Figure 1 and 2: Keloid over the left arm and rights shoulders

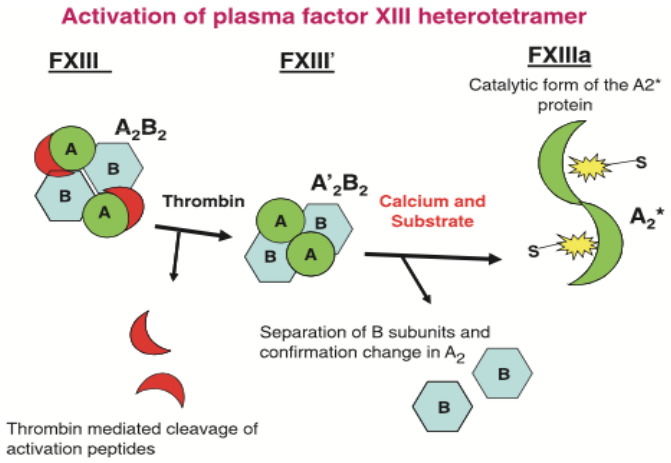

Figure 3: Activation of plasma factor XIII heterotetramer. *From (HSIEH, L. and NUGENT, D. (2008). Factor XIII deficiency. Haemophilia, 14(6), pp.1190-1200)*

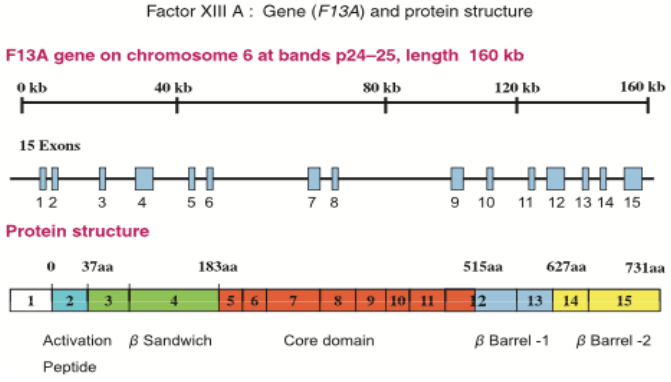

Figure 4: Factor XIII A: gene (F13A) and protein structure. *From (HSIEH, L. and NUGENT, D. (2008). Factor XIII deficiency. Haemophilia, 14(6), pp.1190-1200)*

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