CASE STUDY

A case of Factor XIII deficiency in New Zealand

C Sydney Shepherd

ABSTRACT

Factor XIII is a rare clotting deficiency whereby the formed fibrin clot is unstable and leads to a bleeding tendency. Routine clotting factor assays are usually normal leading to this disorder being missed in the first instance. In this case report a historic case of Factor XIII deficiency in New Zealand is provided and highlights the classical initial problems with identifying this inherited disorder.
Key words: Factor XIII deficiency, missed diagnosis, fibrin stability.

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INTRODUCTION

Formerly known as fibrin stabilising factor or Laki-Lorand factor, Factor XIII is a pro-transglutaminase for a plasma transglutaminase and is converted to the active form (XIIIa) by the action of thrombin in the presence of calcium ions. In the terminal clotting stage, Factor XIIIa acts by cross-linking the formed fibrin clots to stabilise and protect them from fibrinolysis (1).

First formally described by Duckert *et al.* (2,3) in a young boy with a cut to his head associated with a prolonged bleeding time, which was subsequently identified as Factor XIII deficiency. In Factor XIII deficiency, clots form as normal after bleeding or tissue damage but tend to break down if not stabilised by Factor XIIIa leading to a delay in the healing process. Factor XIII deficiency (with less than 1 per cent of the normal Factor XIII) is inherited as an autosomal recessive trait that affects about one in three million people (1). However, there are a number of missense and nonsense point mutations in the Factor XIII gene which will produce a range of bleeding tendencies, the majority of which may be familial and are considered to be autosomal recessive in origin (4).

CASE STUDY

MP presented to our laboratory for investigation of bruising episodes especially around his legs when about two years old, routine blood screening showed his haematology results to be normal including platelet count and coagulation screening tests (PCT, APPT, TCT, fibrinogen) were all normal.

The patient again presented for investigation some two and a half years later, this time with the added information that the bruising on his arms and legs appeared to occur in fairly regular cycles of about six-week intervals. Again, all the coagulation screening tests were within normal range. However, careful observation of the clots formed during these tests noted that the clots gradually broke down and showed dissolution.

A study of the literature indicated the possibility of Factor XIII deficiency and samples were submitted to Professor Duckert's reference laboratory in Geneva where confirmation showed that MP had a Factor XIII level of between 10 to 15% of normal range. Family studies showed that MP's two siblings both had normal levels of Factor XIII, but interestingly both parents showed subnormal levels (40 to 50% of normal). Unfortunately, we were unable to obtain samples from any other extended family members due to privacy reasons.

Treatment was begun with transfusion of cryoprecipitate with dosage appropriate for age and body size at six weeks intervals and the bruising episodes subsided. Sadly, the patient was killed in an accident before long-term survey of his condition could be monitored.

In conclusion, Factor XIII deficiency should always be considered and either confirmed or excluded in investigation of bleeding, bruising disorders when all clotting screening test show normal results.

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