a case of von williebrand's disease

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Von Williebrand's disease is a hereditary haemorrhagic disorder inherited as a dominant characteristic and affecting both sexes. The disease is characterised by three important laboratory findings viz.,

1. An increased bleeding time — this in the acute phase, at other times it may be perfectly normal.
2. A positive Hess's test.
3. A secondary hypochromic anaemia.

The disease is due to an inherited abnormality of the skin capillaries and mucous membranes to which is added in many cases a deficiency in clotting factor VIII. The general features of the disease are not unlike haemophilia, in fact in the old medical literature we see it referred to as haemophilia of the female. The main symptom as is to be expected is a bleeding tendency, which may appear at birth or be latent for years. Commonly affected are the nose, gums, uterus and less commonly the stomach, intestines, urinary tract and joints. There is no associated splenomegaly.

Variants of the above mentioned disease reported in the literature are:

1. Disease + deficiency of factor VIII so called Angiohaemophilia (Klespner and Achenbach 1957).
2. Disease + deficiency of factor XI (Fick et al 1959).
3. Disease + deficiency of factors XI and VIII (Perry et al 1964).
4. Disease + deficiency of factor IX, so called Angiohaemophilia B (Klespner and Achenbach 1957).
5. Disease + deficiency of factor VIII + platelet abnormality (Raccuglia and Neel 1960)

CASE REPORT

This case patient was clerked by me during my stay at the Cardiff Royal Infirmary in August 1969.

K. S. a boy 13 years old was referred to the William Diamond ward with a history of spontaneous epistaxis from the left nostril two days prior to admission. A piece of cotton wool soaked in adrenaline pushed up the nostril controlled the bleeding temporarily, but this occurred again the day before admission, and the patient was sent to hospital.

On examination there was nothing abnormal in any of the boy's systems but a look at his past notes revealed that he was referred to the Infirmary in 1962 because of recurrent spontaneous epistaxis with no other physical signs: a number of investigations were done on him at that time: *they illustrate very well how a haemorrhagic disorder should be investigated in the laboratory.*

Bleeding time: 15 minutes + (control 2 — 7 minutes)
Platelets: 401,000
Clotting time: 7 minutes (control 5 — 10 minutes)
One stage prothrombin time: 15 seconds
Thromboplastin generation test: N.A.D.
Prothrombin Consumption index: 17%
Factor VIII assay: 65% of normal
Clot retraction test: 55% (control 45 — 64%)
Platelet thrombotic function reported normal in comparison with normal platelets in a dilution of 1 : 4
Result: a prolonged bleeding time and low factor VIII.

The blood of the boy's father and paternal uncle who also suffered from recurrent spontaneous epistaxis was tested and prolonged bleeding times were found in both

Father : 10 minutes
Paternal uncle : 15 minutes +

Thus due to:

1. An abnormal bleeding time in association with perfectly normal platet count clotting time and other relevant values as seen above.
2. Presence of a low factor VIII level
3. Familial trend.

A diagnosis of Von Williebrand's disease was made.

No specific treatment was recommended, but the boy's family was asked to send him to hospital should any haemorrhagic manifestations occur. In 1968 the patient suffered a dislocated radial epiphyses on which surgical intervention was considered necessary. A laboratory

check on the patient's blood showed that the bleeding time was 15 minutes and the factor VIII level was 33%: naturally this was not an ideal state of affairs in which to operate, so the boy was given one litre of frozen plasma — this to act as a source of factor VIII, as a result of which the bleeding time fell to 7 minutes and the factor VIII level rose to 50%. The operation was successful and the patient leads a very active life: he is a member of the school rugby team and plays cricket and soccer.

TREATMENT

Thus no specific treatment is possible: one lets the patient lead an active and healthy life and in cases of bleeding episodes such as epistaxis or prolonged bleeding after a tooth extraction, cotton wool swabs soaked in adrenaline are advisable and if these fail to stop the bleeding, early hospitalisation is indicated. In hospital symptomatic treatment is given and in severe cases factor VIII may be supplied (when necessary) by frozen plasma infusions. The intrinsic defect in the capillary wall, the whole basis of Von Williebrand's disease, cannot, for the time being at least, be affected by any method of treatment.

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