GLANZMANN'S P DISEASE

Pages with reference to book, From 84 To 86

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Glanzmann's disease is a rare disorder of platelet function. Specifically, the problem is of defective platelet aggregation and impairment of platelet factor 3. The disease is transmitted in an autosomal recessive pattern and affected children present with bleeding manifestations consistent with quantitative platelet disorder.

CASE REPORT

An 11 years male, weighing 22 Kgms, presented with complaints of easy bruisability since birth and bleeding from gums. His problems started at the time of circumcision (age 11 days) when he bled for 24 hou.rs. Numerous episodes of skin bruises were noticed after this episode. An episode of haematuria at age 6 months and bleeding &om the mouth after being slapped by his father brought him to the hospital at 6 years at which time he was given blood transfusion. He has since then been bleeding from gums off and on and has been given thirteen blood transfusions with a diagnosis of haemophilia.
Family Pedigree
1) 1st boy died soon after birth due to unknown cause. He had generalized bruises and ecchymoses.
2) 2nd boy — now 11 — the patient.
3) 3rd boy — died at 18 days due to pneumonia (had bruises at birth).
4) 4th a girl, 7 years old — absolutely healthy.
5) 5th a girl, 5 years old — developed bruises and ecchymoses at birth that disappeared in 2 weeks — now bleeding from nose and gums off and on for the last 2 years.
6) female — 2 years old — born with bruises and ecchymoses which disappeared at 2 weeks — baby is in good health.
7) male — 5 months — absolutely healthy. Father and mother are first cousins.
There was no family history of bleeding disorder in any family member. Examination revealed a young thin lean boy oriented in time and space. Pulse, respiration, temperature and blood pressure were within normal limits. Significant findings included pallor, small discrete nodes palpable in neck, groin and bruises on left arm and right leg. Gums were bleeding and caries of teeth was obvious. X-ray of the chest was normal. Test for aggregation of platelets = Negative (no aggregation). Platelet factor 3 availability = Normal. All members of the family were subjected to investigations.Results are shown in Table.

TABLE

Haematological Investigations of the Family.

Person	Age	Clotting Time	Bleeding Time	Aptt	Clot Retraction	Platelet Count	Platelet Morimology	Platelet Aggregation
Index Patient (Male)	Ilyrs	4':15"	14'	26/31sec	Abnormal	210,000/cmm	Normal	No Aggregates
Sister	7 yrs	6':45"	2':20"	36/40sec	Normal	220,000/cmm	Normal	Normal
Sister	5 yrs	7':30"	25':45"	38/40sec	Abnormal	190,000/cmm	Normal	No Aggregates
Sister	2yrs	4':50"	15':40"	28/30sec	Abnormal	240,000/cmm	Normal	No Aggregates
Brother	5mo	3':20"	3':15"	32/36sec	Normal	230,000/cmm	Normal	Normal
Father	40 yrs	4':15"	3':40"	37/30sec	Normal	220,000/cmm	Normal	Normal
Mother	30yrs	5':35"	4':50"	35/40sec	Normal	260,000/cmm	Normal	Normal
Control	30yrs	4':30"	3'	36/33sec	Normal	210,000/cmm	Normal	Normal

Pedigree is depicted in Figure.

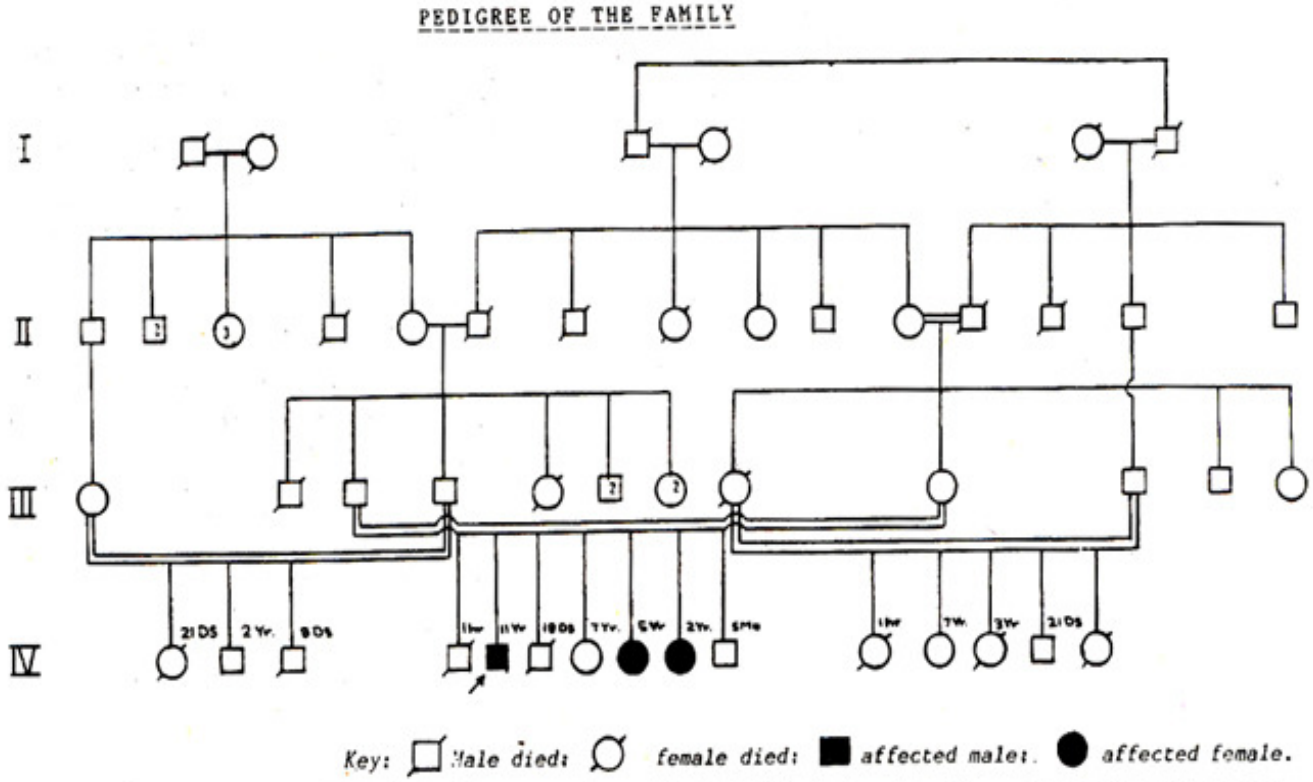

Figure - Pedigree of the affected family. Only important connections in the family have been shown. Ages of the children at the time of death or otherwise are shown above the block or circles in IV generation.

DISCUSSION

Bleeding from the gums, nose and petechiae point to platelet abnormality. Since the platelet num ber is adequate, this case represents a disorder of platelet function. Adhesion, aggregation and degranulation are three well-known functions of platelets and may result in the following well-recognized conditions:

I. Defective Adhesion:

1. Bernard-Soulier Syndrome

2. Von Willebrand's Disease
II. Defective Aggregation:
1. Glanzmann's Thrombasthenia
III. Defective Degranulation:
1. Storage Pool Disease
2. Aspirin-like Defects

Glanzmann described in 1918[1], the condition which is characterized by prolonged bleeding time and failure of platelet aggregation. The platelets fail to aggregate in response to ADP, epinephrine, collagen, thrombin, 5HT,arachidonic acid & PGE[2]. About 80% of cases also show varying levels of impairment of platelet factor[3-6].

Clinical Manifestations

The disease is transmitted as an autosomal recessive trait with consanguinity being a factor[7,8]. Carriers show no bleeding manifestations and demonstrate normal platelet function[9,10] although platelet membrane glycoprotein analysis has been demonstrated to reveal abnormalities, the bleeding manifestations are similar to those present in other platelet disorder, i.e., - mainly mucosal haemorrhage and may even occur in the neonatal period[4]. Occasionally bleeding manifestations decrease with increasing age[3]. Local measures are often sufficient but major bleeding requires multiple platelet transfusions. It remains a rare disorder with only about 150 cases having been reported[11].. Case reports have come from all over the world. Recently Kbanduri reported 42 cases from South India. Rare as it is, it remains one of the most common and dearly defined congenital qualitative platelet disorders[12].

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