Thrombocytopenic Purpura without Anemia and Leukopenia in Gaucher’s Disease

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THROMBOCYTOPENIA associated with anemia and leukopenia is a common finding in Gaucher’s disease and occasionally is accompanied by bleeding. On the other hand thrombocytopenic purpura without anemia or leukopenia is rare in Gaucher’s disease. Only two such cases have been reported. In the present communication a case of Gaucher’s disease is presented in which the main clinical feature was thrombocytopenic purpura and in which there was only slight enlargement of the spleen and no anemia or leukopenia.

CASE REPORT

C. S., a 27 year old Jewish female clerk was admitted on July 19, 1953, because of subcutaneous hemorrhage, epistaxis, gingival bleeding and meno-metrorrhagia.

Family history. One brother died three days after birth, deeply jaundiced. A sister apparently had jaundice from birth, remained physically and mentally underdeveloped and died at the age of six. The mother died at the age of 53 of cancer of the breast. The father, 60 years of age, apparently is in good health. There is no history of bleeding tendency in the family.

Previous history. At the age of 14 the patient had mumps followed by temporary obesity. Two years previous to admission she developed slight jaundice accompanied by nausea, vomiting, slight upper abdominal discomfort and low grade fever lasting for a few weeks. During the course of a year and a half similar episodes occurred a number of times. The menarche was at the age of 15.

Present illness. Since early childhood the patient had bruised easily, and had shown petechiae and spontaneous subcutaneous hemorrhages as well as frequent nose bleeds and gingival bleeding. Three years previous to admission she had melena. Menstrual periods lasted up to seven days and were accompanied by considerable blood loss.

Physical examination. The patient was in good nutritional state with neither pallor nor jaundice. The temperature, pulse and blood pressure were normal. Numerous ecchymoses and petechiae were seen scattered over the body, especially on the legs. The lymph nodes were not enlarged. The head, neck and chest did not reveal abnormal findings. The abdomen was soft, the liver was palpated 5 cm. below the costal margin; it was firm and not tender. The spleen was palpated just below the costal margin and was firm and not tender. Except for the purpuric manifestations no abnormalities were seen on the legs.

Laboratory findings. The urine was normal. Blood sedimentation rate (Westergren) 9 mm. in the first hour and 26 mm. in the second. Hemoglobin 12 Gm. per cent; red blood count 4.37 million/cu.mm.; reticulocyte count 0.9 per cent; white blood count 9,600/cu.mm. Differential count: 67 per cent polymorph neutrophils, 4 per cent band forms, 2 per cent eosinophils, 22 per cent lymphocytes and 5 per cent monocytes. Thrombocytes 80,000/cu.mm., morphologically normal. The sternal bone marrow was normal, with megakaryocytes normal in number and morphology but with diminished platelet formation. The bleeding time was 6 minutes, clotting time 4½ minutes, clot retraction complete after 75 minutes, tourniquet test positive, plasma prothrombin 60 per cent, serum prothrombin 7 per cent (normal), serum prothrombin conversion accelerator 60 per cent (normal) and fibrinogen 380 mg. per cent. The Coombs, Kahn and Wassermann tests were negative. Blood urea, glucose and
bilirubin were normal. The routine liver function tests, thymol turbidity, thymol flocculation and Takata Ara were normal. The serum total protein was 8.4 Gm. per cent, albumin 4.6 Gm. per cent, and globulin 3.8 Gm. per cent.

X-ray of the chest and the electrocardiogram were normal.

The diagnosis of idiopathic thrombocytopenic purpura was made. The patient was given ACTH by intravenous drip, 5 mg. daily for a period of ten days. No new hemorrhages appeared. The thrombocyte count on the tenth day of treatment had risen to 130,000/cu.mm., the tourniquet test was negative, the bleeding time 5 minutes, the clot retraction normal and the serum prothrombin 7 per cent.

Splenectomy was performed on the 10th of August, 1953. The thrombocyte count immediately preceding operation was 90,000/cu.mm., the tourniquet test slightly positive amid the bleeding time 3 minutes. The spleen was found to be of normal shape but slightly enlarged, weighing 250 Gm. The cut surface appeared homogenous and rich in pulp. Microscopic examination of the spleen revealed small groups of Gaucher cells scattered in the intersinusoidal tissue. No megakaryocytes were seen in the sections.

Six hours following splenectomy the thrombocyte count had risen to 160,000/cu.mm., the white blood count to 15,000/cu.mm., and the red blood count was 4 million/cu.mm. The bleeding time was 1½ minutes and the tourniquet test was negative.

After the diagnosis of Gaucher's disease had been established on histologic examination of the spleen, the preoperative bone marrow smear was re-examined and a few Gaucher cells were found. X-ray examination showed the characteristic Erlenmeyer flask-shaped lesion of the lower third of the femur. The patient was discharged one week after operation when her condition was satisfactory and there were no signs of bleeding. During the next two months repeated blood examinations revealed thrombocyte counts of 140,000–200,000 per cu.mm. and normal tourniquet test, bleeding time and clot retraction.

On the 14th of February, 1954, the patient was examined in the third month of pregnancy. She had no bleeding phenomena and the thrombocyte count was 220,000/cu.mm. During pregnancy the patient felt well and thrombocyte counts ranged from 200,000 to 300,000 per cu.mm., until the 9th month of pregnancy when a minimum of 120,000/cu.mm. was found and the routine hemostatic tests were normal. On the 17th of August, 1954, the patient delivered a healthy female baby. The cord blood showed 185,000 thrombocytes/cu.mm. Two days after birth the thrombocyte count of the infant was 80,000 and on the 5th day 185,000/cu.mm. During the week after delivery the mother's daily platelet counts ranged from 130,000 to 160,000/cu.mm.

**Comment**

This case of Gaucher's disease is unusual in that its main feature was thrombocytopenic purpura in the absence of anemia, leukopenia and with only slight enlargement of the spleen. However the possibility must be considered that both Gaucher's disease and thrombocytopenic purpura in this patient were present coincidentally and not related to each other.

The absence of marked splenomegaly in Gaucher's disease, although rare, is known to occur especially in cases in which symptoms appear late in the course of the disease and the rate of progression is slow. In most of such cases anemia was an outstanding feature and is ascribed to crowding out of the bone marrow by Gaucher cells. In the present case there was no anemia despite the bone destruction demonstrated in the X-ray of the femur.

Thrombocytopenic purpura as the only clinical manifestation in Gaucher's disease is rare. Davis et al. described an adult patient with this syndrome who was successfully treated by splenectomy. In their case the liver and spleen were markedly enlarged and both these organs as well as the bone marrow contained large numbers of Gaucher cells. Wintrobe refers to a similar case in a child of 8 years.
Although thrombocytopenic purpura as the sole manifestation of Gaucher's disease is very rare, this disease should be considered in the differential diagnosis of idiopathic thrombocytopenic purpura.

SUMMARY

A patient with Gaucher's disease is described who had thrombocytopenic purpura, but no anemia or leukopenia and only slight enlargement of the spleen. The hemorrhagic diathesis and platelet count responded well to splenectomy.

SUMMARIO IN INTERLINGUA

Es describite un patiente comi morbo de Gaucher qui habeva purpura thrombocytopenic sed ni anemia ni leucopenia e solmente un leve allargamento del splen. Le diathese hemorrhagic e le conto del placchettas respondeva ben a splenectomia.

REFERENCES

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