Discussion
Bernard-Soulier syndrome is a hereditary disorder of platelet function first described in 1948 by a special form of hereditary thrombocytopenia in which the platelets were of giant size and yet could not produce a normal prothrombin consumption. The platelet counts and clot retraction were normal. The inheritance of the syndrome was described to be autosomal recessive.

The first step of the process of coagulation is adhesion of platelets to the subendothelium of the blood vessel. This leads to aggregation (primary and secondary) and release of platelet factor 3 from platelet membrane, which participates in the clotting process. In BSS the aggregation is defective and hence the bleeding tendency.

BSS belongs to the group of congenital qualitative disorders of platelets, which also include Glanzmann's thrombasthenia. In von Willebrand disease, however, the basic defect is with plasma and, although the picture may mimic BSS, the platelets are normal in size, PTT(k) is abnormal and factor VIII is reduced. All these are correctable by plasma infusion, as against the defect in BSS for which platelet transfusion is the only remedy.

Another condition where giant platelets are found is May-Hegglin anomaly. This is a rare hereditary disorder transmitted as an autosomal dominant trait. Basophilic inclusion bodies (upto 3) called "Doll's bodies" are found in the granulocytes of these patients. The patients with this disorder present with a variable bleeding tendency, especially epistaxis and purpura. The platelet count is low and the clot retraction depends on the platelet count. Other investigations including bone marrow examination are usually normal.

When suspected, BSS is diagnosed by a prolonged bleeding time, giant platelets and defective platelet aggregation with ristocetin. It commonly manifests as gastrointestinal haemorrhage, uterine bleeding, epistaxis and purpura. Over 30% of the patients suffering from BSS die due to gastrointestinal haemorrhage. However, this is a rare condition and only about 60 cases have been reported in world literature.

The treatment of this condition is difficult. Splenectomy and corticosteroids are ineffective. Platelet transfusion during the episode of bleeding is the only effective therapy. However, antplatelet antibodies develop slowly and therefore after some time this therapy becomes useless. It is thus advisable to use platelets from HLA-matched donors, preferably the patient's siblings. The patient should avoid all drugs which impair platelet function and cause gastrointestinal haemorrhage.

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