Unexpected intra-operative bleeding due to Hermansky-Pudlak Syndrome

Sir,

Hermansky–Pudlak syndrome (HPS), first described in 1959 by Hermansky and Pudlak, is an autosomal recessive disorder that results in oculocutaneous albinism and bleeding problems due to an abnormality in platelet aggregation.[1] It affects 1 in 500,000–1,000,000 worldwide, but in Puerto Ricans, its prevalence is significantly higher, that is, 1 in 1800.[2] We report an undiagnosed HPS with intra-operative bleeding. Early diagnosis and the subsequent need to adopt necessary pre-cautions before major surgery render this syndrome very important for anaesthetists because 15% of mortalities are caused by haemorrhagic manifestations.[3]

A 35-year-old female patient with oculocutaneous albinism presented to our clinic with complaints of extremely ptotic and hypertrophic breasts. The patient was hospitalised for reduction mammoplasty. Investigations revealed haemoglobin of 11.9 g/dL; platelet count of 357 × 10^3/μL, prothrombin time (PT) 13.3 s and partial thromboplastin time (PTT) of 30.2 s. Her bleeding and clotting times were 1.5 min and 10.5 min, respectively. The patient underwent bilateral mammoplasty under general anaesthesia, lasting 5.5 h. Owing to intra-operative bleeding, patient received 6 units of packed red blood cells (PRBCs) and 2 units of fresh frozen plasma (FFP) during surgery. The post-operative blood parameters were; haemoglobin 11.3 g/dL, platelet count, 268 × 10^3/μL, PT 15.3 s and PTT 32.8 s. Due to continued bleeding in the post-operative period, the patient underwent a consultation at a haematology clinic and was clinically diagnosed with HPS. The patient developed hypotension and tachycardia and was administered 2 units of PRBCs and 2 units of FFP in the next 2 days. Bleeding persisted and on post-operative day 2, the patient underwent surgery again to control the bleeding under general anaesthesia, with laryngeal mask airway. Pre-operatively, on the advice of the haematologist, the patient was administered intravenous infusion of vasopressin (0.5 μg/kg). Following surgery, the patient recovered without any problems and was discharged from the hospital on post-operative day 6.

In general, HPS patients present to hospital during childhood with mild bleeding. Such bleeding usually includes mucosal bleeding and bleeding following tooth extraction or surgery, as seen in platelet function disorders.[4] The history of HPS patients generally includes ecchymoses in the body during childhood. The absence of childhood ecchymoses in our patient led us to miss the diagnosis of HPS. The blood counts, PT, PTT, international normalised ratio, and platelet count were also normal, but prolonged clotting time (10.5 min) was observed. In addition, the presence of oculocutaneous albinism suggested the clinical diagnosis of HPS. The diagnosis was confirmed by the absence of delta granules in platelets under an electron microscope.

No definitive treatment except symptomatic relief is available for HPS. General recommendations include the use of pre-operative intravenous desmopressin and having platelets 'on standby' for procedural surgeries.[5] Bleeding diathesis can be treated with the infusion of desmopressin (0.3–0.5 μg/kg) and platelet transfusion during surgery.[6]

In conclusion, HPS should be considered in the diagnosis of patients with oculocutaneous albinism; even in the absence of bleeding problems (bleeding may be mild or missed). If not diagnosed and treated early, such cases may present with extremely severe bleeding in major surgeries.

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