Multiple Gestation in a Patient with Hermansky-Pudlak Syndrome

Abstract Type: Case Report/Case Series
Lisa A. Rhodes, M.D.; John J. Kowalczyk, B.Sc.; Bridget M. Marroquin, M.D.
Strong Memorial Hospital, University of Rochester

Hermansky-Pudlak syndrome (HPS) is an autosomal recessive disorder defined by oculocutaneous albinism, platelet storage dysfunction, and abnormal lipofuscin deposits. Pulmonary fibrosis or granulomatous colitis may be present in severe cases. This syndrome creates unique circumstances for the parturient. Patients are at high risk for morbidity due to their bleeding diathesis.

A 33 yo G4P2 Puerto Rican female with HPS and triplet gestation was admitted at 32 weeks EGA for preterm labor. The patient had a history of severe postpartum hemorrhage with her first vaginal delivery. Postpartum hemorrhage was a concern because of the patient’s bleeding diathesis and increased potential for uterine atony with the triplet gestation. The patient was taken for urgent Cesarean section due to fetal distress. Desmopressin (0.3 mcg/kg) was administered prior to surgery. In the operating room, an arterial line and large bore peripheral intravenous access were obtained. General endotracheal anesthesia was induced and triplets were delivered uneventfully. After delivery, the patient was treated with oxytocin and methylegonovine to improve uterine tone. The patient’s mild thrombocytopenia (100K) was treated with five units of platelets. Estimated blood loss was 800ml. Transfusion of red blood cells was unnecessary. The patient was extubated and recovered without incident. The triplets were taken to the NICU for further monitoring.

HPS occurs in one in 500,000 worldwide but more commonly in Puerto Rico, with one in 1,800 persons affected. The bleeding diathesis results from the absence of dense bodies, contained within platelets. Dense bodies release factors which assist in platelet aggregation.

Our patient was of fair complexion, with nystagmus, decreased visual acuity, and mild pulmonary fibrosis. Her PT and PTT were normal, but bleeding time was prolonged, which is typical for HPS.

To avoid excessive bleeding in the pregnant HPS patient, intravenous desmopressin (0.3 mcg/kg) should be given prior to delivery. This causes an increase in release of factor VIII and Von Willebrand’s factor from storage sites. The mechanism of action of desmopressin in HPS is unclear, but case reports have demonstrated its efficacy. Platelets and packed red cells should be available for possible transfusion. Neuraxial anesthesia is contraindicated in HPS patients.


