

RARE HERMANSKY-PUDLAK SYNDROME TYPE -2 IN A PREGNANT PATIENT: A CASE REPORT

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INTRODUCTION

Hermansky-Pudlak syndrome (HPS) is a rare autosomal recessive disease characterized by oculocutaneous albinism, a bleeding diathesis, and other organ involvement specific to certain types. The HPS-2 subtype is distinguished by the presence of neutropenia. In this case we discuss the management of pregnant patient known to have HPS-2 made complex by a consanguineous marriage as the status of baby affected by the condition was unknown.

CASE REPORT

A 25 years old Pakistani woman, with a known diagnosis of HPS-2, was managed by the Obstetric Haematology team during her pregnancy.

She and her sister were diagnosed at a young age. She suffered from recurrent respiratory infections as a child and was found to be immunodeficient. She was on long term Granulocyte Colony Stimulating Factor (G-CSF) and prophylactic Azithromycin for neutropenia but had poor compliance. She bruised easily on trauma and had a history of epistaxis requiring tranexamic acid. She had paler hair and skin than the rest of her siblings. She was registered as sight impaired with a right sided esotropia and nystagmus. The specific diagnosis of HPS-2 was based on genetic testing done at the age of 15.

Her parents had a consanguineous marriage and she also married her cousin. She had a spontaneous miscarriage when she first conceived but did not report any heavy bleeding. When she became pregnant again, she booked late at 20+4 weeks. She had stopped taking G-CSF of her own accord but did not suffer from any recurrent infections nor bleeding during her pregnancy. Her bloods showed thrombocytopenia and neutropenia. She was counselled that being related to her husband, there was a 1 in 4 risk of the baby being affected.

A detailed intrapartum plan was made by the haematology team. This involved avoiding any neuraxial anaesthesia, ventouse,

rotational forceps, fetal blood sampling or fetal scalp electrodes. A plan was made to give 1g Tranexamic Acid (TXA) intravenously (IV) when in established labour and to continue oral Tranexamic Acid 1g four times a day for 7 days post-delivery. 1 unit of platelets was to be arranged when the patient went in to labour. Plan was to transfuse the platelets if she had a Caesarean section. In case of a vaginal birth with excessive bleeding despite TXA, she was to receive the platelets. Cord blood was to be taken at the time to delivery to check baby's bloods for any abnormality.

She was induced at 39+1 weeks for estimated fetal weight above the 90th centile on a customised growth chart. She had a normal vaginal delivery with an episiotomy. The third stage was actively managed with an oxytocin infusion. Episiotomy was stitched in the room to minimize blood loss. The placenta did not deliver more than half an hour following delivery despite controlled cord traction. She was transferred to theatre for adherent placenta with a lower threshold for blood loss in view of her haematological condition. The placenta was manually removed under general anaesthetic. Vaginal wall tears noted in theatre were sutured. A vaginal pack was placed because of continuous oozing from the friable vaginal mucosa. She had a total blood loss of 1800mls following delivery. She received 3 units of red cells and 2 units of platelets. She was also covered with antibiotics. Baby's full blood count came back normal so no further testing was organized. The patient recovered well and went back to her haematology follow ups.

DISCUSSION

- ❑ HPS was first discovered in 1959. It has a prevalence of 1 to 9 in 1,000,000 individuals worldwide. But more common in Puerto Rico where the incidence is 1 in 1800, and ~ 1 in 22 individuals are carriers.
- ❑ Eleven types of HPS associated with mutations in 11 different genes have been recognised till now. These mutations cause altered biogenesis of lysosomes and lysosome-related organelles and defective intracellular protein trafficking. HPS-2 is a rare subtype caused by mutations in AP3B1 gene.
- ❑ Recurrent infections because of neutropenia and immunodeficiency have been described in HPS 2 and 10. Pulmonary fibrosis similar to idiopathic fibrosis occurs in patients with HPS types 1,2 and 4. These patients require long term follow up.
- ❑ HPS can cause life threatening haemorrhage in obstetrics and gynaecology patients.

CONCLUSION

Obstetric patients with HPS need multidisciplinary input for safe management. It is crucial to optimise primary haemostasis at the time of delivery in the case of a known diagnosis of HPS with a detailed intrapartum plan beforehand. Preconception counselling is important with a 25% risk of baby being affected if both parents are carriers.