Anesthetic Challenges For A Parturient With Hermansky-Pudlak Syndrome – A Case Report

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Introduction

- Hermansky-Pudlak Syndrome (HPS): a rare genetic metabolic disorder characterized by oculo-cutaneous albinism and platelet dysfunction.
  - ➢ Etiology of platelet dysfunction: absence of platelet dense bodies.
  - ➢ Qualitative bleeding disorder: bleeding time significantly prolonged despite normal PT, PTT, and platelet counts.[1]
- HPS presents challenges in the management of intra-and postpartum bleeding:
  - ➢ Especially in cases with obstetric comorbidities predisposing to blood loss.[2]
  - ➢ Methods of labor analgesia are also limited due to contraindications against using neuraxial blocks.[3]

Methods

- This case was reviewed retrospectively and the evaluation of current pertinent literature was completed.
- The patient consented to the creation and presentation of this case report.
- Given that the report does not include identifiable patient information, it is exempt from IRB review in accordance with Hartford Healthcare policy.

Case Presentation

- 24 year-old parturient (38 weeks gestation)
- Known HPS (easy bruising, heavy menses)
- Vaginal bleeding ongoing for 2 days (Hgb 9.8, platelet 209)
- Placental abruption concern

Case Management

Approach	• Multidisciplinary: Hematology, Maternal Fetal Medicine, and Obstetric Anesthesiology teams.
Pre OP:	• At admission: Transfusion 2 units cross-matched platelets • Prior to delivery: 2 units platelets, 1 g TXA (2 hours prior)
Intra OP:	• Platelets continuous transfusion • Induced vaginal delivery with Pitocin • No neuraxial anesthesia (risk of epidural hematoma), nitrous oxide used instead, pain score decreased from 8/10 to 2/10 • Backup plan: C-section with general anesthesia and blood products available
Post OP	• 2 units platelets (12 and 24 hours after initial and second transfusions) • Oral TXA for 7 days
Outcome	• Uncomplicated vaginal delivery with suboptimal but satisfactory pain control and no significant bleeding • No intra or postpartum hemorrhage; Hgb stable • Discharged on day 2 without complications

Discussion & Conclusion:

- Being mindful of the disease pathophysiology.
- Vigilance and preparation for adverse outcomes
  - ➢ Hemorrhagic event could lead to significant maternal morbidity and mortality.4
  - ➢ Blood products ready, with pre and post-delivery platelet transfusions
- Alternative pain management plans
  - ➢ Neuraxial anesthesia contraindicated
  - ➢ Nitrous oxide, systemic opioid administration, and non-opioid pharmacologic management.[3]
  - ➢ NSAIDs should be avoided in patients with HPS.[5]

References

1. International Journal of Gynecology Obstetrics, 154, 412–426, 2021.
2. Obstetric Medicine, 1(2), 95-96, 2008.
3. BMJ Case Reports, Volume 2016, 2016.
4. Heritable Disorders of Amino Acid Metabolism, 1974, 177-261, 1974.
5. American Journal of Obstetrics and Gynecology, 5, 564-565, 1985.