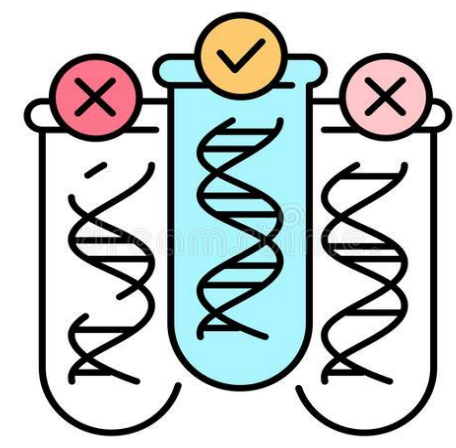


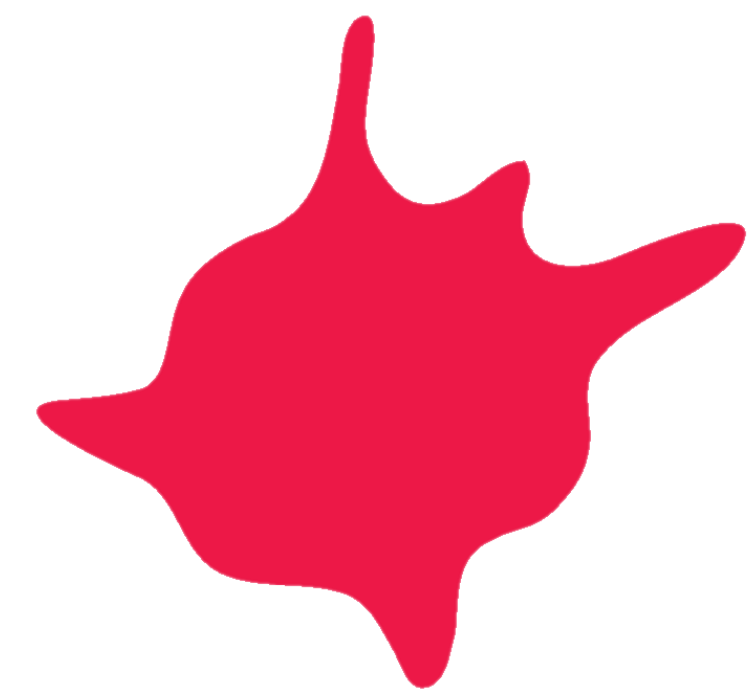
The Influence of Private Genetic Testing on the Peripartum Workup of Thrombocytopenia and Selection of Neuraxial Anesthesia for Cesarean Delivery

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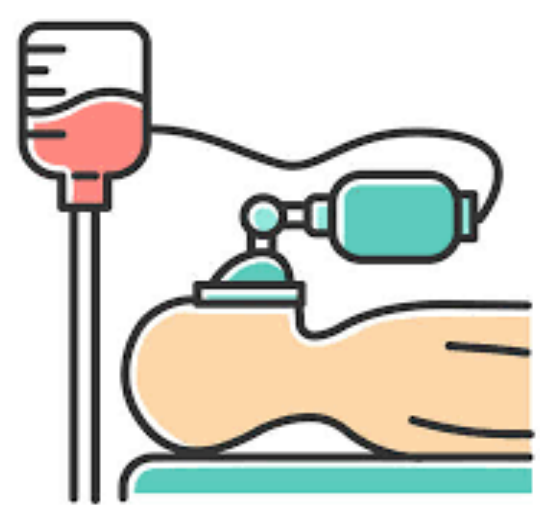
Genetic Testing



- In 2015, 200,000 women underwent elective carrier screening in the USA.¹



- ACOG recommends all testing options should be discussed with every pregnant woman.²



- The private company Invitae: “are able to screen for up to 280 conditions”.

Bernard-Soulier Syndrome (BSS)

- Very rare, autosomal recessive.
- Defect of the platelet membrane complex.
- Thrombocytopenia, prolonged bleeding time and giant platelets.
- Variable phenotype: easy bruising to major hemorrhage.³

Anesthetic Considerations for BSS

- Insufficient information to guide safety of neuraxial technique.
- Higher rates of primary and secondary postpartum hemorrhage reported.⁴

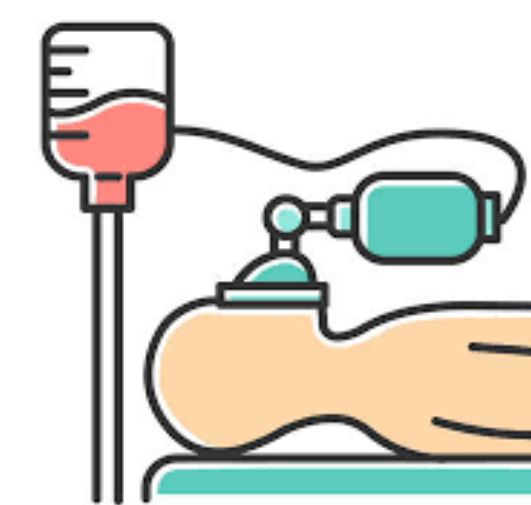
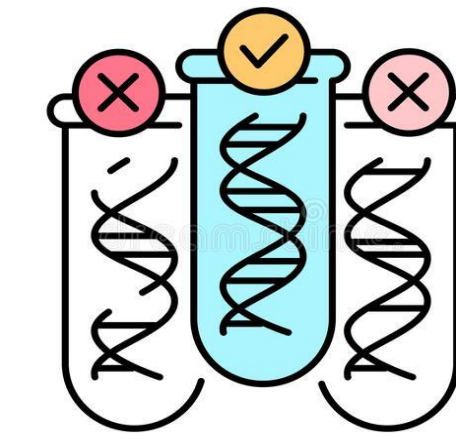


DEPARTMENT OF
ANESTHESIA

Case Presentation

Antepartum

- 32 yr G1P0 IVF twins.
- Pre-conception screening revealed BSS carrier state.
- Thrombocytopenia ($66-81 \times 10^9/L$).
- No bleeding episodes.
- Giant platelets but normal aggregometry
- Hematology -“interplay of gestational thrombocytopenia and BSS carrier status”.
- Trial of prednisone - no effect.
- Plan for elective cesarean delivery .



Cesarean Delivery

- Preoperative platelet count **68** $\times 10^9/L$.
- Risk/Benefit discussion with patient and partner.
- Decision: Single shot spinal with 25G, abandon if more than one attempt required.
- Spinal anesthetic - successful with one attempt.
- 500 mLs blood loss.

Postpartum

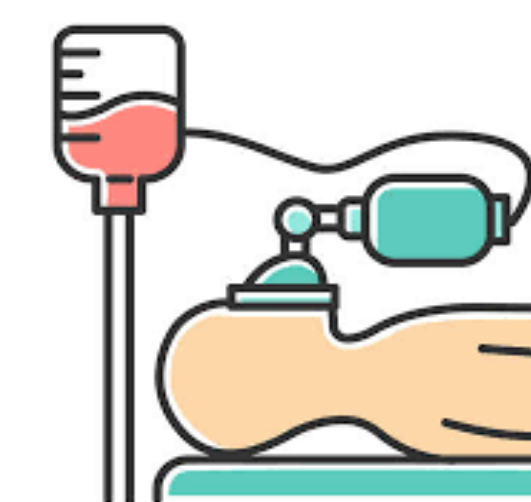
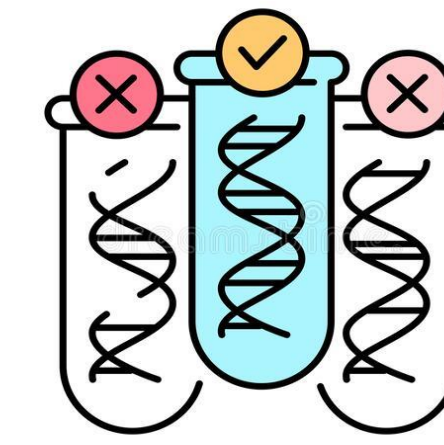
- Patient discharged home on day 2.
- Healthy twins born with no complications.

Discussion

Learning Points

1. Patients obtaining private whole-genome testing is becoming more common.

- May raise new clinical information that may significantly impact anesthetic choice and management.
- May require additional investigations and multidisciplinary consultations.
- Important to actively enquire about genetic testing during antenatal consultations.



2. The decision to perform neuraxial blockade in patients with BSS is challenging

- Many grey zones without clear-cut answers from guidelines.
- Paucity of cases in literature to guide management.
- Relies on clinical judgement and include patient/partner in decision making.

References

1. Committee Opinion No. 691: Obstet Gynecol. 2017
2. Lazarin et al. Semin perinatol 2016
3. Peitsitis et al. Haemop 2010
4. Bragadottir G et al. Am J Hematol 2015