"Genetic Double Jeopardy"
A Patient With Stickler Syndrome and Haemophilia B Carrier

K Hoyland[1] and K Cheesman[2]

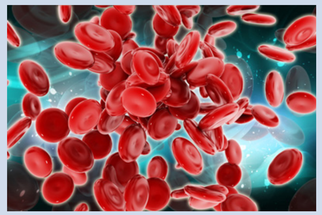
[1]Obstetric Anaesthetic Fellow, [2]Obstetric Anaesthetic Consultant, St Thomas' Hospital, London

Guy's and St Thomas' NHS Foundation Trust

INTRODUCTION

- Sticklers syndrome is a rare, multisystem connective tissue disorder [1]
  - Issues with associated airway features of Pierre Robin
  - Hypermobility – potential LA failure
  - Scoliosis
  - Mitral valve prolapse
- Haemophilia B is an X-linked bleeding disorder, due to lack of factor IX
  - 1 in 4 female carriers have bleeding symptoms
  - Risk of PPH [2]
- Limited literature in obstetric patients





CASE REPORT

- 21yr old primip
- Multi-disciplinary approach planned for safe delivery
- Sticklers syndrome
  - Manifested with retinal issues, sensorineural hearing loss, osteoarthritis, hypermobility
  - Main obstetric concern was retinal complications from prolonged 2nd stage
- Factor IX levels were normal throughout pregnancy
  - No concerns about regional anaesthesia
  - Bleeding concerns from patient due to maternal PPH in past
- Anaesthetic considerations:
  - Airway assessment showed flattened nasal bridge but no obvious difficult airway
  - USS lumbar spine unremarkable
  - Hypermobility raised concerns about LA resistance
- Patient opted for elective LSCS rather than induction with early epidural
- Underwent spinal anaesthetic with good block
- Tranexamic acid perioperatively
- Uncomplicated surgery – 350ml blood loss

DISCUSSION

- Sticklers syndrome and Haemophilia B are not related
- When presenting together can pose significant challenges to the anaesthetist
- In sticklers syndrome with airway concerns, cautious regional anaesthesia is advised with safe airway back up plans in case of failure
  - Potentially likely due to difficult spinal access from scoliosis or unpredictable LA effects
- History of bleeding is important in Haemophilia B carriers
  - Factor IX levels do not change significantly during pregnancy (unlike factor VIII)
  - Levels should be checked at presentation and during 3rd trimester [2]
  - Factor IX concentrate can be given if severe haemorrhage [2]
- The anaesthetic challenges that connective tissue disorders can cause are frequently underestimated by obstetric and midwifery colleagues
- These women are rarely reffered to anaesthesia clinics in our experience
- Early referral of these patients is important to aid planning and safe delivery

References

1. Gomez J, Rice SM, Makhamreh MM, Al-Kouatly HB (2020) Pregnancy management in a patient with stickler syndrome. Molecular Genetics and Genomic Medicine 8: e1311
2. Shah UJ, Narayanan M, Smith JG (2015) Anaesthetic considerations in patients with inherited disorders of coagulation. Continuing Education in Anaesthesia Critical Care and Pain 15(1): 26-31