The Reproductive Implications of a Rare Congenital Disorder

Gorlin-Chaudhry-Moss Syndrome

Growth retardation
Skeletal abnormalities
Conductive hearing loss
Genito-urinary malformations
Hair abnormalities: Hypertrichosis, low hair line
Lipodystrophy and increased risk on insulin resistance
Craniosynostocis: Premature closure of fibrous joints (sutures)
Cardiac abnormalities: Patent ductus arteriosus, cardiomegaly, valvular problems, propensity to endocarditis
Facial abnormalities: Mid-face hypoplasia, maxillary hypoplasia, high arched palate, microphthalmia, hypodontia, dental malocclusion

Case Review:

- 37yo with GCM presented to a fertility clinic with difficulty conceiving
- Preconception counselling led the couple to opt for pre-implantation genetic diagnosis (PGD)
- She conceived naturally prior to PGD

- PMHx:
  - *Mitral and tricuspid regurgitation*
  - *Dilatation of the aortic root, ascending aorta and aortic arch*
  - *Hirsutism (on Spironolactone)*
  - *Classic facies*
  - *Short stature*

Discussion:

In light of the rarity of the condition the potential obstetric complications were theoretical and as such only advisories exist from multidisciplinary input and interpretation of existing more generalised guidelines

Unfortunately the 12 week scan revealed anomalies of the lower spine and limbs, absence of an identifiable bladder and suspected heart defects. The couple opted for termination and post mortem confirmed the diagnosis of sirenomelia

The GCM gene was not identified

Antenatal and Intrapartum Care Plan:

- Spironolactone was discontinued due to feminising effects on a male fetus

- To reduce the risk of cardiac decompensation:
  - *Surveillance by antenatal echo each trimester*
  - *Beta blockers to reduce the risk of aortic dissection*
  - *Vigilance for hypertension*

- Screening for gestational diabetes

- Antenatal anaesthetic review:
  - *Facial abnormalities may pose difficulties in both face-mask ventilation and intubation*
  - *Skeletal malformations may complicate intra-operative positioning as well as venous access*
  - *Dermatoglyphs associated with GCM can preclude finger pulse oximetry*

- Elective caesarean section, indicated by virtue of her significant aortal disease, arguable may also have been warranted due to short stature and limited pelvic capacity.

- A contingency plan in the event she presented in advanced labour was made including strict fluid management and cardiac appropriate uterotonics

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