CASE 13 – “Peter’s anomaly”

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CASE REPORT

- 49 day old female
- Referred for corneal “thickening” in both eyes
- Family history: no similar findings
- Otherwise well
EYE EXAMINATION UNDER ANESTHESIA

• Corneal opacities in both eyes
• Axial length: 17 mm both eyes
• IOP: 31 – 35 mmHg both eyes
• Fundus: difficult to visualise due to opacity
B-SCAN ULTRASOUND
ULTRASOUND BIOMICROSCOPY

- Corneal thickening in both eyes
- Membrane behind posterior corneal surface, possibly Descemet membrane?, making a cystic space
- Anterior subcapsular cataract
WHAT DID WE DO?

We chose to perform a Trabeculectomy in both eyes.
FOLLOW-UP AFTER SURGERY

- IOP: 17/17
- BMC: central corneal scarring, 10 mm both eyes.
- Axial length: 21 OD, 22 OS
FOLLOW-UP 6 MONTHS AFTER SURGERY

- IOP controlled
- Pharmacological midriasis to allow light to enter the eye
- Early visual stimulation
- Nystagmus
1- Which of these genes do you expect to be mutated in this case?
   a – PAX6, PITX2, CYP1B1, FOXC1
   b – KRT3, KRT12, TGFBI, SLC4A11
   c – COL8A2, ZEB1, TACSTD2, TGFBI

2- Which of these would you consider as differential diagnosis in this case?
   a - Sclerocornea
   b – Endothelial dystrophy
   c – Dermoid
   d - Rubella
   e – All of them

3- Would you recommend surgery for visual rehabilitation (cornea/lens) now, age 7 months?
   a – Yes
   b – No
THANK YOU!