

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



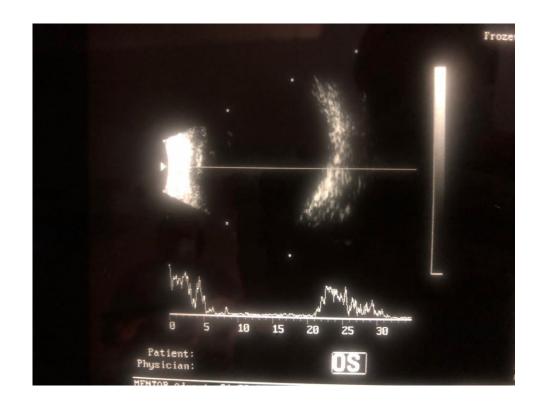






B-SCAN ULTRASOUND







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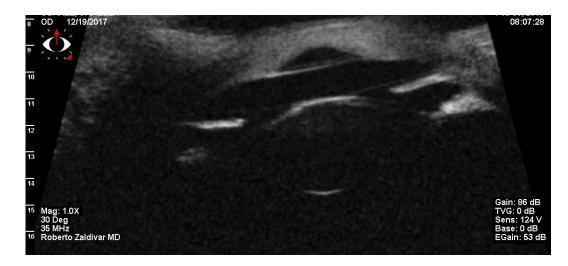


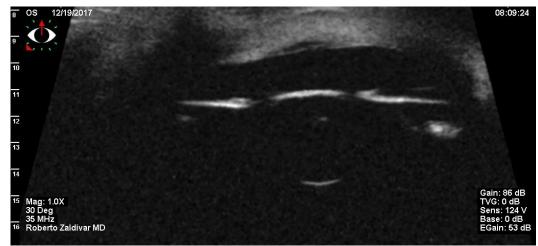




ULTRASOUND BIOMICROSCOPY

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















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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











Some questions about the case...

- 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 reccommend surgery for visual rehabilitation (cornea/lens) now, age 7 months?
- a Yes
- b No









We would like to ask the experts...

- 1- Would you proceed in a different way in this case?
- a Yes. (If so, what would you do?)
- b No
- 2- Would you consider Cyclo-G-6 to control IOP in this patient?
- a Yes
- b No
- 3- Please explain the UBM findings?
- 4- What would you do with the corneal opacity?
- a Corneal graft (PK) at this age
- b Rotational autokeratoplasty
- b Limbal stem cell transplantation
- c Wait (In this case, until when?)
- d Other: (In this case, what would you do?)









THANK YOU!







