case study

By

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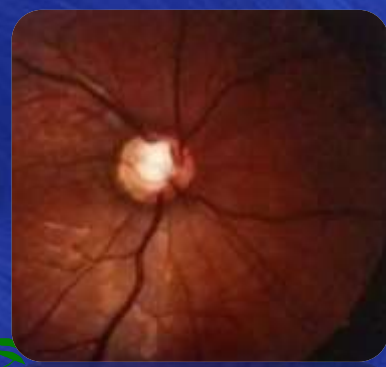
HISTORY

- Male child 16 years old.
- Chief Complaint: Blurring of vision and presented to my clinic for glasses prescription.
- Past History: No previous ocular trauma or surgery.
- Past Medical History: Unremarkable.
- Family History: positive glaucoma

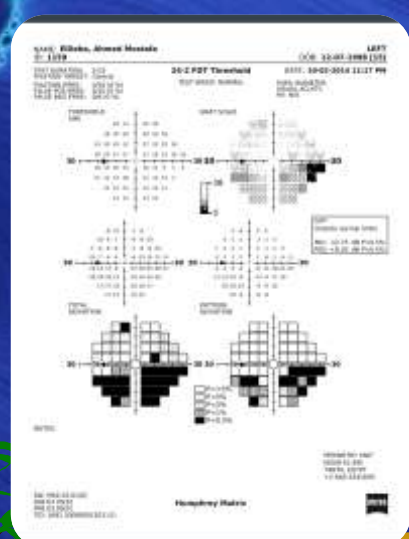
ON EXAMINATION

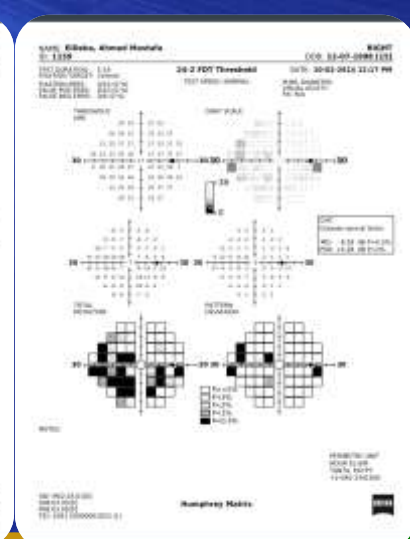
- Best corrected visual acuities: 20/20 OD, 20/20 OS.
- Rx: -0.75 sph OD, -1.25 - 0.5 x 135 OS.
- EOM: full.
- Pupils: RRR.
- Gonioscopy: Wide-open angles OU.
- IOP: 50 mmHg OD, 44 mmHg OS(applanation).
- SLE: Normal OU.
- CCT: 534 OU.

Optic Disc Examination

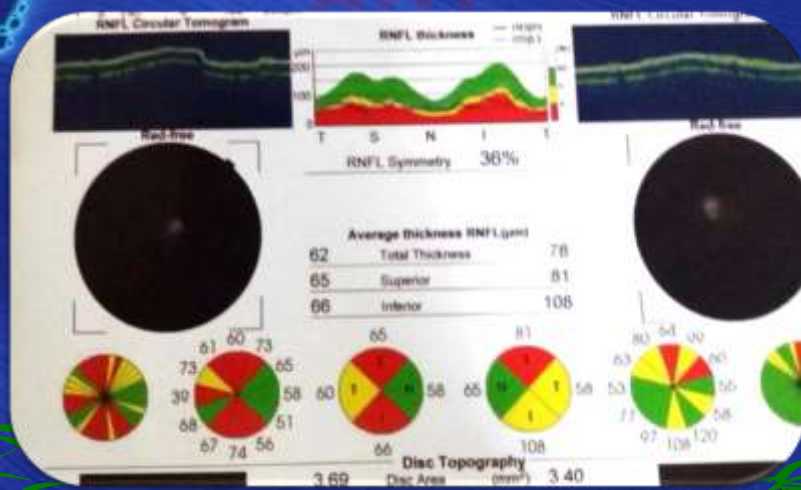


24-2 VF test





OCT



DIAGNOSIS

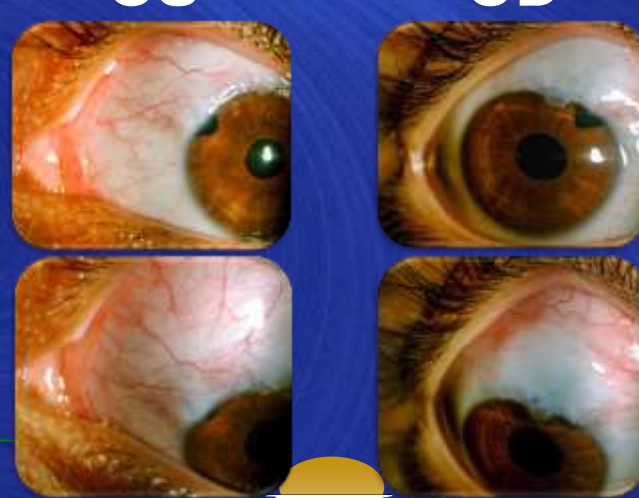
- Juvenile Onset Primary Open Angle Glaucoma (JOPOAG).

MANAGEMENT

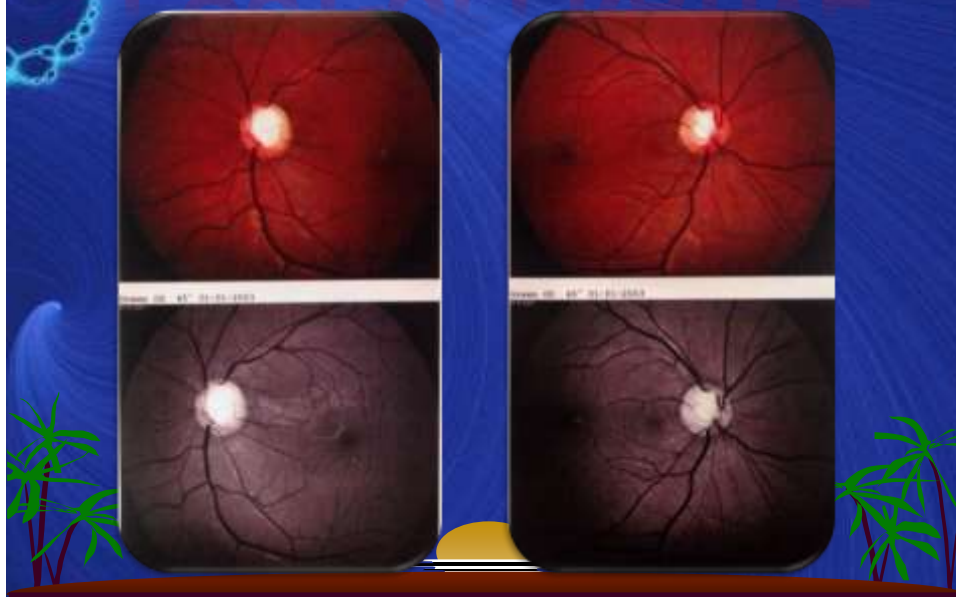
- Maximal anti-glaucoma medication, but without adequate IOP control.
- Trabeculectomies with MMC, were eventually performed in both eyes with good pressure control (OD 13mmhg, OS 14mmgh).

POST- TRAB (MMC)

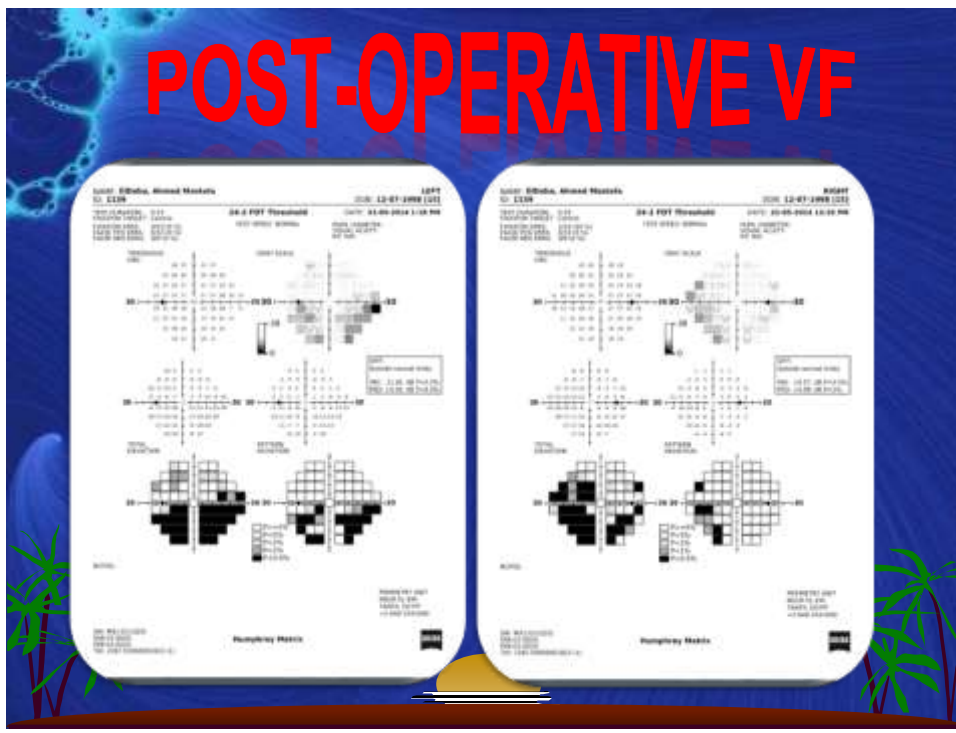
OS OD



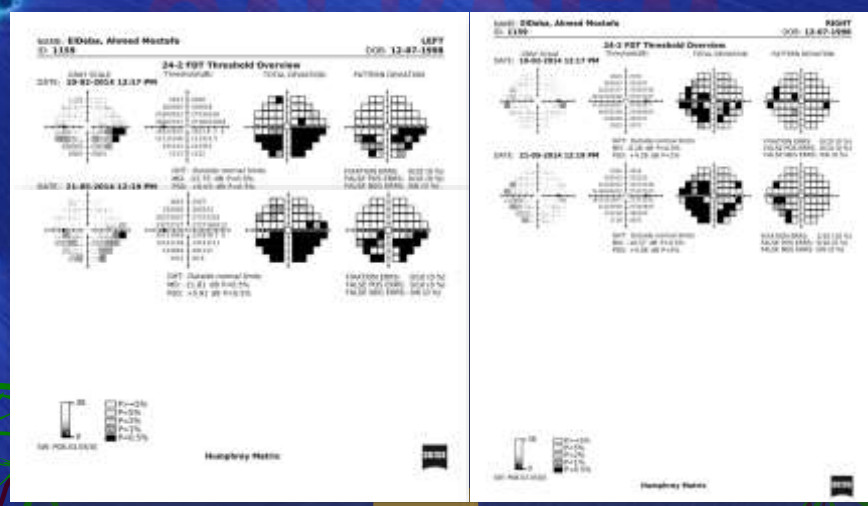
POST-OPERATIVE



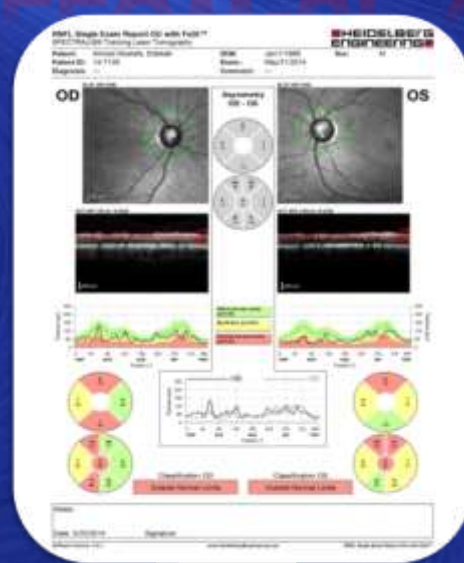
POST-OPERATIVE VF



OVERVIEW



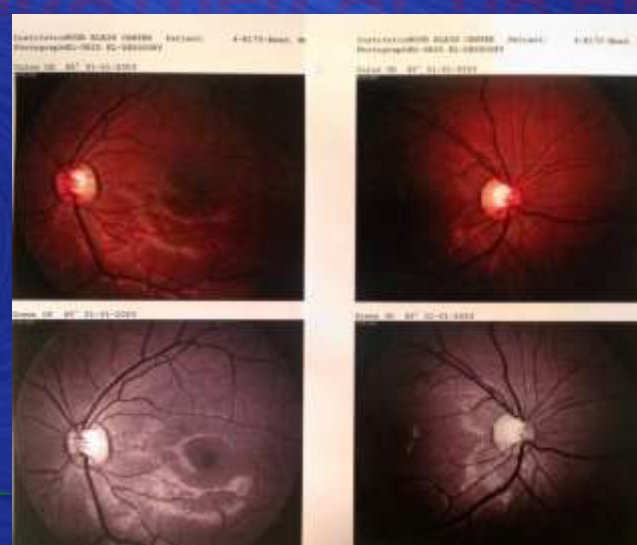
FOLLOW-UP OCT



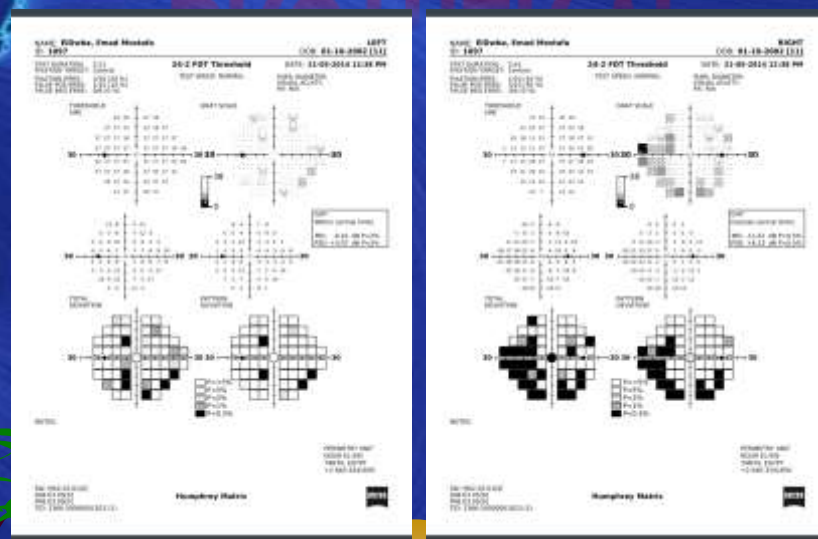
FAMILY SCREENING

- Other family members were screened for glaucoma.
- Revealed that his brother 11 y. old and his sister 8 y. old have suspicious glaucomatous optic disc with IOP 42 OD, 30 OS for the boy and 23 OD, 19 OS for the girl

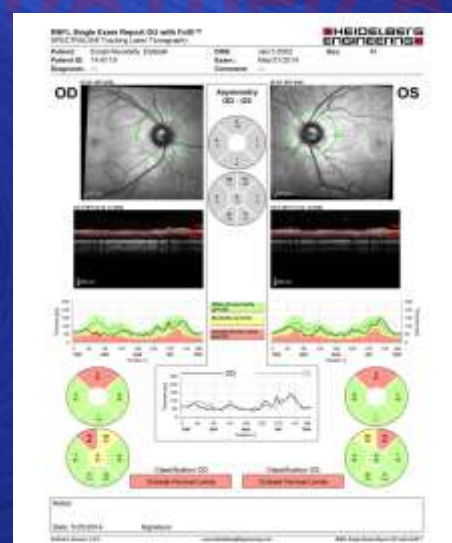
BROTHER OPTIC DISC



BROTHER VF

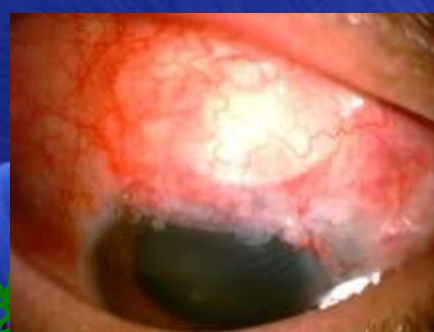


BROTHER OCT

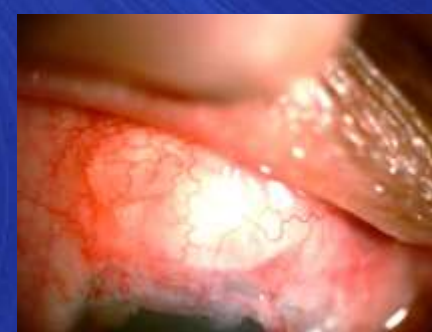


BROTHER POST TRAB (MMC AND OLOGEN)

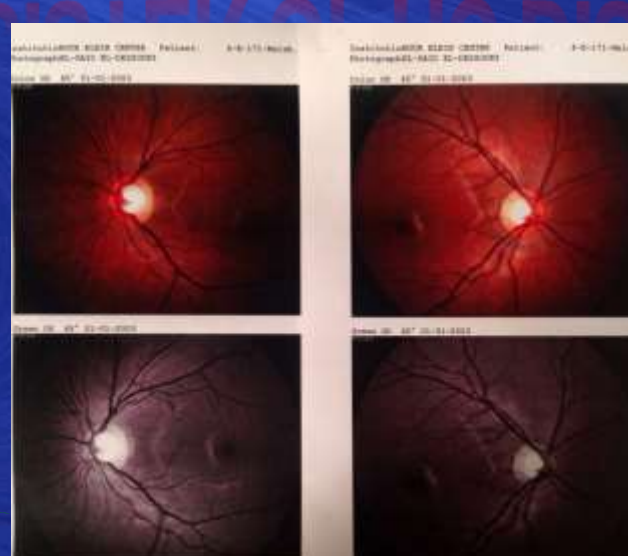
OS



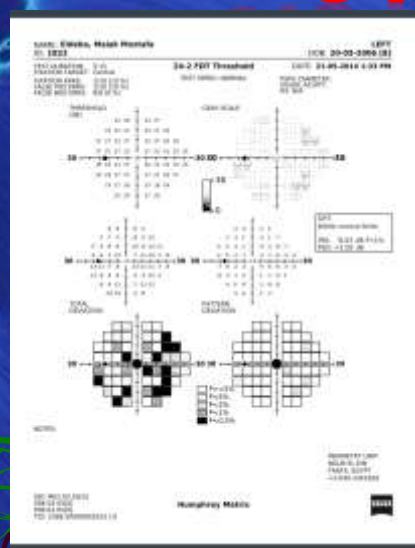
OD

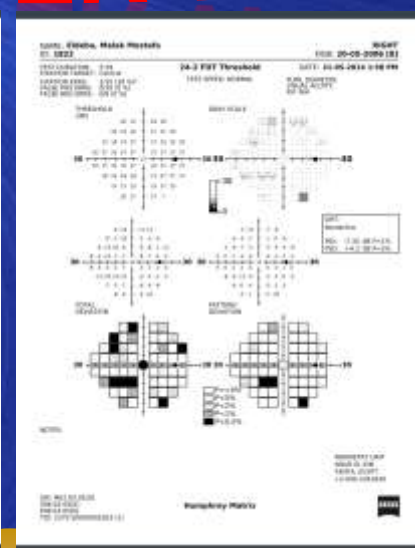


SISTER OPTIC DISC

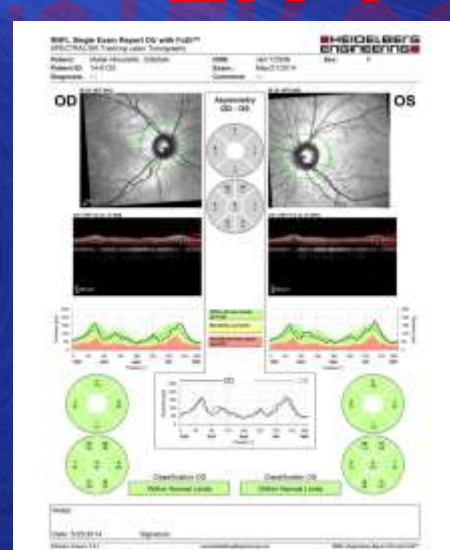


SISTER VF





SISTER OCT



DISCUSSION

- JOPOAG differs from adult onset POAG in the age of onset and often in magnitude of IOP elevation (often have extremely high IOP, some times more than 50 mm.Hg).

DISCUSSION

- JOPOAG is differentiated from late congenital glaucoma and other childhood glaucoma by the absence of large globe, Descemet's breaks (Haab's striae), or anterior segment dysgenesis.

DISCUSSION

- JOAG is inherited as an autosomal dominant pattern of inheritance.
- Myocilin mutations are responsible for a significant fraction of JOAG cases.
- Some of the case series in the literature shows association of JOAG with keratoconus and myopia pointing out the importance of proper ophthalmological work up of a patient with JOAG.

ROLE OF GENITIC TESTING

- Genetic family testing may be used to identify those family members at greatest risk for developing glaucoma so that appropriate medical and surgical options could be offered without delay.
- Also, those family members with no myocilin mutations are not at increased risk for developing JOAG, however, they still had the population risk of developing glaucoma that is unrelated to mutations in the myocilin gene.

POINTS TO REMEMBER

- Full ophthalmological examination for every case, even in apparently straight forward cases.
- Put glaucoma in your mind for early detection.
- Early screening of other members of the family (irrespective of age) with a known glaucoma patient can help to diagnose JOAG at an earlier stage and could probably prevent or postpone the final visual deterioration.

POINTS TO REMEMBER

- Medical therapies often have limited success in managing JOAG.
- Surgical management should be undertaken without any further delay because of the severity of the disease and it's early age of onset.


Thank you