UNVEILING MYSTERIES: OCULAR MANIFESTATIONS IN A CHILD WITH NEUROFIBROMATOSIS TYPE 1

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INTRODUCTION

- Neurofibromatosis 1(NF1) - one of the most common genetic diseases - is an autosomal dominant genetic disorder.
- The disease manifestations are extremely variable, even within a family. NF1 is characterized by:
  - Multiple cafe au lait spots
  - Axillary and inguinal freckling
  - Multiple discrete dermal neurofibromas
  - Iris Lisch nodules
- Plexiform neurofibromas (PNF) are neoplasms that are characteristic of NF1, often causing disfiguring effects (e.g., on the face), and are considered precancerous lesions.
- Glaucoma and associated globe enlargement are common in patients with NF1 and orbital-facial involvement.
- I present a case of a child with NF1 exhibiting atypical retinal findings in the other eye. Discussing these unusual manifestations emphasizes the need for tailored management strategies and can help refine diagnostic protocols and guidelines to ensure comprehensive care.

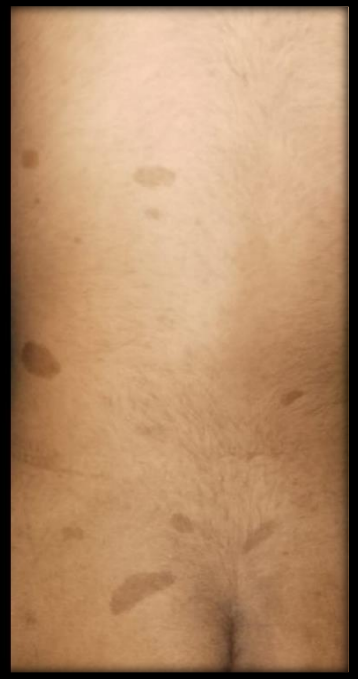
MATERIALS & METHODS

CASE REPORT

- A 3-year-old male with multiple café au lait macules on the trunk and the left side of the face, along with disfigurement of the left side of the face, presented to our hospital at the age of 2.5 months with the chief complaint of left eyeball enlargement.

- Birth history included delivery by cesarean section at full term, a birth weight of 2.8 kg, & no history of ICU admission.

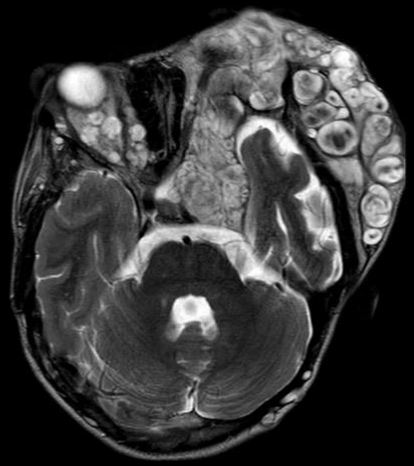
- During further evaluation of the medical history, it was discovered that both his mother and maternal grandfather also have Neurofibromatosis Type 1.



Parental consent was obtained before sharing the child's photos.

INVESTIGATIONS

Blood investigations were within normal limits.



MRI Brain was suggestive of plexiform neurofibroma with sphenoid wing dysplasia and homogenous smooth enhancement of choroidal layer of the left eye

Examination under General Anesthesia

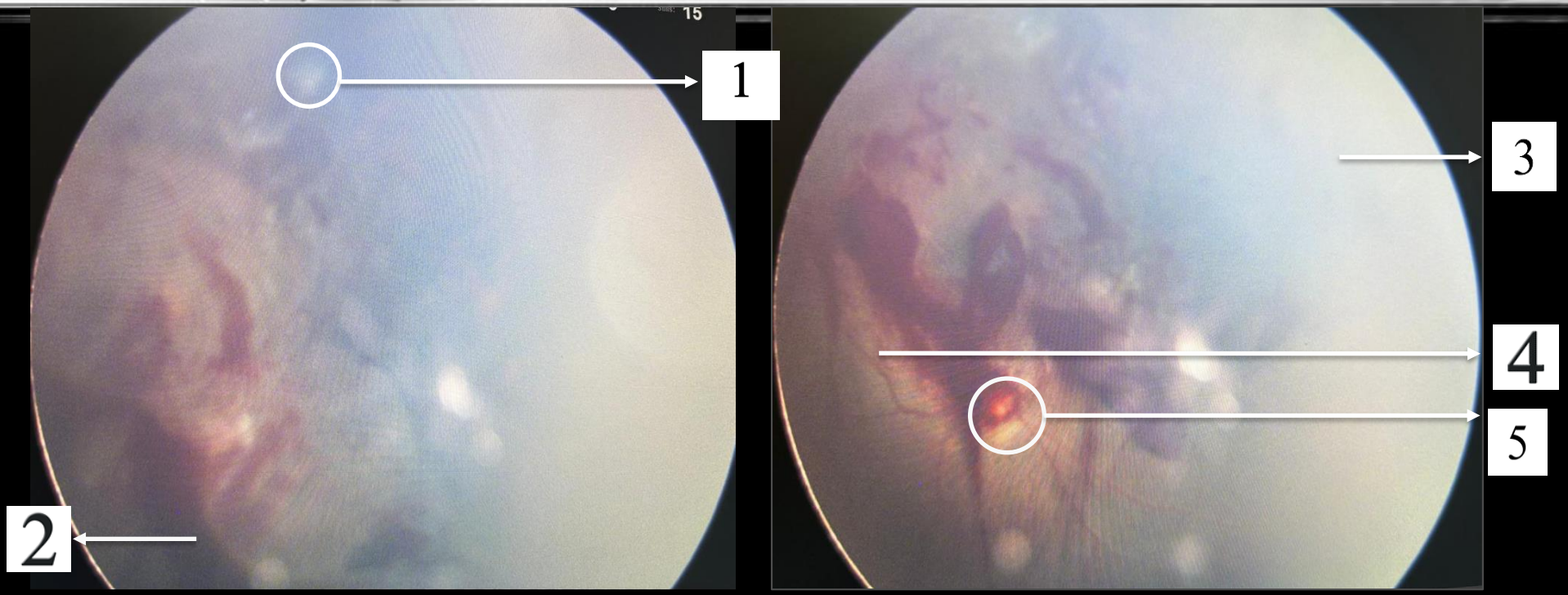
FINDINGS OF EXAMINATION UNDER GENERAL ANAESTHESIA

DATE	IOP (AT)		K(V)*K(H)		FUNDUS	
	RE	LE	RE	LE	RE	LE
13/09/21	14	26	11*10.5	14*13.5	0.2 CDR, healthy NRR	DNS
	LEFT EYE TRABECULECTOMY WITH MITOMYCIN C WAS DONE ON 13/09/21					
16/10/21	14	20	10*10.5	12*13	0.2 CDR, healthy NRR	Disc hazily seen, deep cup
15/11/21	16	24	11*11	13*13	0.2 CDR, healthy NRR	0.3 CDR, flame shaped hemorrhage inferior to the disc
31/01/22	10	16	10*10.5	13*13	0.2 CDR, healthy NRR	0.4-0.5 CDR
15/11/22	8	12	10.5*10	13*13	0.2 CDR, healthy NRR	0.7 CDR

- Subsequent follow-ups were sporadic as the patient had a history of epilepsy for which he was on anti-epileptics and physiotherapy.
- A fundus examination was performed on 22/07/24. To further assess the right eye in detail, a fundus image was captured using a RetCam device.

IOP (AT)		K(V)*K(H)		FUNDUS	
RE	LE	RE	LE	RE	LE
18	20	10*10.5	12.5*13	*Atypical findings	0.8-0.9 cup disc ratio with disc pallor and full retinal vascularization in the left eye

*FUNDUS FINDINGS IN THE RIGHT EYE



1. Fibrotic band in the infratemporal quadrant
2. Multiple retinal and pre-retinal hemorrhages
3. Abruption of vessels 360° with peripheral ischemia
4. Dilated and tortuous vessels
5. Hemorrhages over the disc

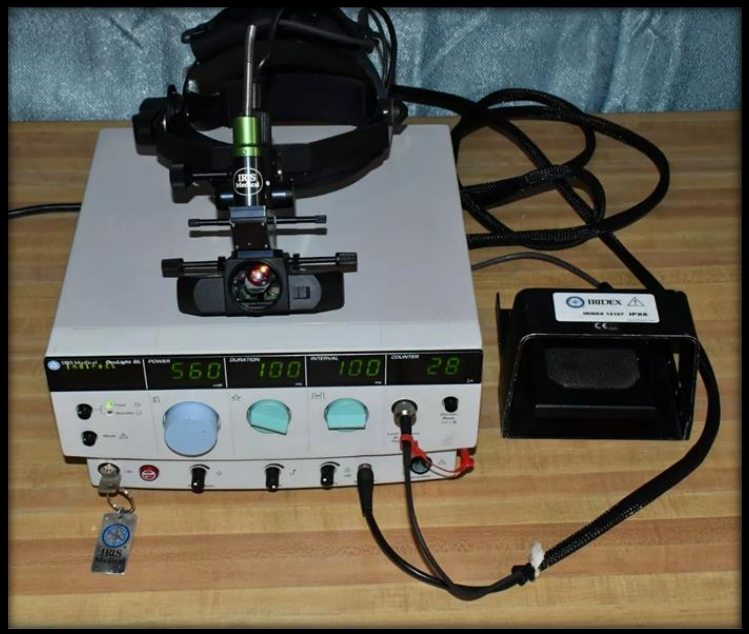
DISCUSSION: ?FAMILIAL EXUDATIVE VITREORETINOPATHY

- Full-term birth, normal birthweight, and no history of oxygen requirement may lead us to the diagnosis of FEVR.
- FEVR exhibits three different forms of inheritance: autosomal dominant, autosomal recessive, and x-linked recessive. Among these, autosomal dominant is the most common form.

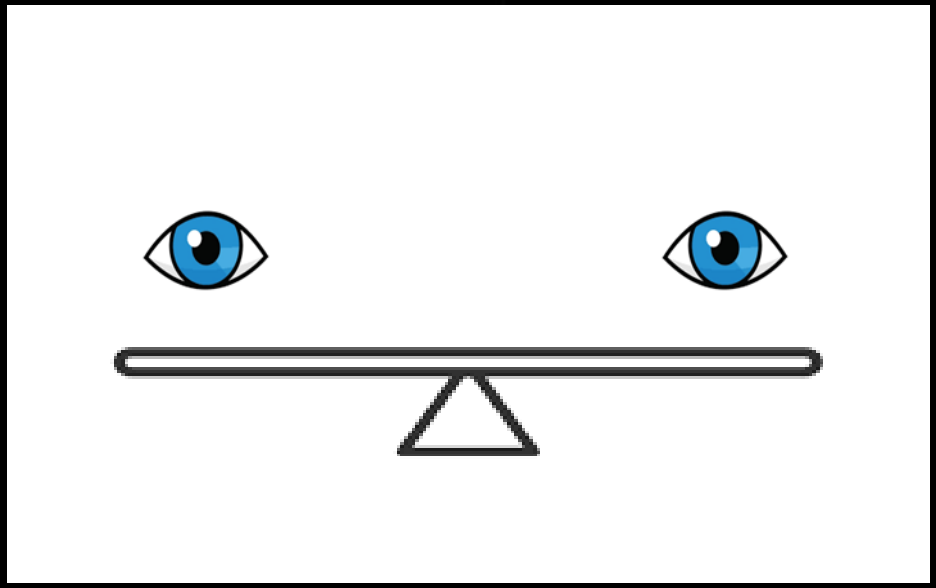
Revised FEVR Clinical Staging System 2014	
Stage	**Clinical Features**
1	Avascular periphery or anomalous intraretinal vascularization
1A	Without exudate or leakage
1B	With exudate or leakage
2	Avascular retinal periphery with extraretinal vascularization
2A	Without exudate or leakage
2B	With exudate or leakage
3	Extramacular retinal detachment
3A	Without exudate or leakage
4	Macula-involving retinal detachment, subtotal
4A	Without exudate or leakage
4B	With exudate or leakage
5	Total retinal detachment
5A	Open funnel
5B	Closed funnel

MANAGEMENT

- The patient has been called for a follow-up and will be managed with an anti-VEGF injection, followed by photocoagulation using a Frequency-Doubled Nd:Yag Laser (532 Nm).



CONCLUSION



Despite the right eye having appeared normal in all prior assessments over the years, recent findings underscore the importance of conducting thorough and regular examinations of **both eyes**.

REFERENCES

- Pinson S, Wolkenstein P. La neurofibromatose 1 (NF1) ou maladie de Von Recklinghausen [Neurofibromatosis type 1 or Von Recklinghausen's disease]. Rev Med Interne. 2005 Mar;26(3):196-215. French. doi: 10.1016/j.revmed.2004.06.011. PMID: 15777582.

- Friedrich RE, Christ G, Scheuer HA. Mandibular symmetry on posterior-anterior cephalograms of neurofibromatosis type 1 patients with facial plexiform neurofibroma. GMS Interdiscip Plast Reconstr Surg DGPW. 2023 Dec 11;12:Doc11. doi: 10.3205/iprs000181. PMID: 38111842; PMCID: PMC10726724.

- Abdolrahimzadeh B, Piraino DC, Albanese G, Cruciani F, Rahimi S. Neurofibromatosis: an update of ophthalmic characteristics and applications of optical coherence tomography. Clin Ophthalmol. 2016 May 13;10:851-60. doi: 10.2147/OPTH.S102830. PMID: 27257370; PMCID: PMC4874640.

- Sizmaz S, Yonekawa Y, Trese TM. Familial exudative vitreoretinopathy. Turk J Ophthalmol. 2015;45(3):164-8.
- Kashani AH, Learned D, Nudleman E, Drenser KA, Capone A, Trese MT. High prevalence of peripheral retinal vascular anomalies in family members of patients with familial exudative vitreoretinopathy. Ophthalmology. 2014;121(1):262-8.

"Vision is not just what we see, but how we strive to understand the world. Happy learning!"