Using Optical Coherence Tomography and Optical Coherence Tomography Angiography to Understand Pathoanatomical Changes of the Vitreoretinal Interface and Inner Retina in Patients with Neurofibromatosis 2

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Purpose

To assess and better understand the ocular presentations in patients with neurofibromatosis 2 (NF2) using multimodal imaging of optical coherence tomography (OCT) and optical coherence tomography angiography (OCT-A)

Materials and Methods

- OCT and OCT-A data from four patients (ages 9, 11, 13, and 34) with NF2-associated ocular presentations were collected for this retrospective study
- OCT (Spectralis; Heidelberg Engineering, Heidelberg, Germany) and OCT-A (Solix, Optovue, Fremont, USA) were used to capture images during a standard comprehensive clinical examination
- All patients were screened for genetic testing upon suspicion for NF2. Three out of the four cases were confirmed to be NF2 positive (one of which was confirmed to be mosaic NF2)
- Pathoanatomical changes were documented and compared among the four patients to look for associations

Imaging Results

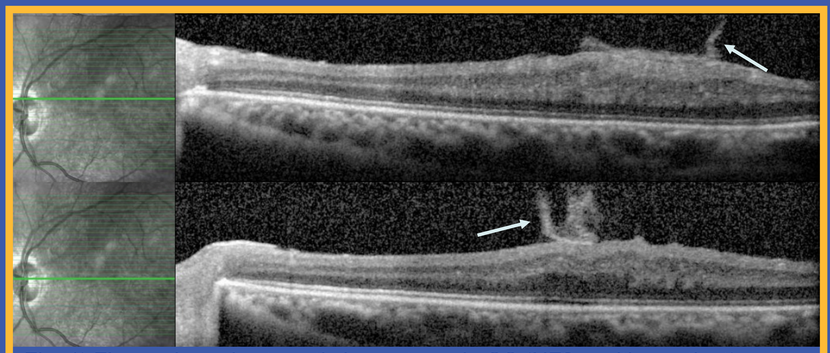

Fig. 1) Flame-shaped astrocytic hamartoma in OS (NF2 positive patient)

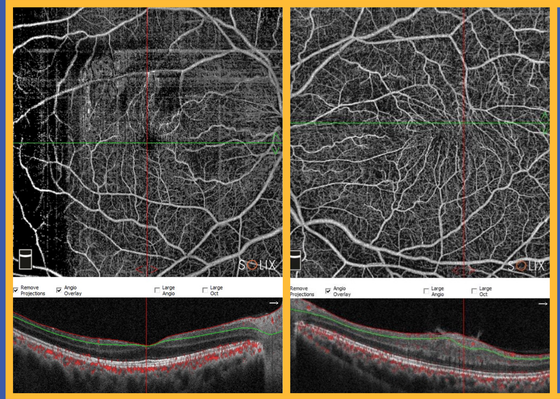

Fig. 3a) Left column: OCT-A of OD superficial layer("control")

Fig. 3b) Right column: OCT-A of OS superficial layer (abnormal)

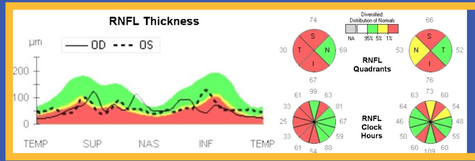

Fig. 4) OCT shows retinal nerve fiber layer thinning OU

Results

- Eight eyes from four patients presented with ocular abnormalities typically associated with NF2
- Retinal astrocytic hamartomas were documented in all four patients in at least one eye
- Optic nerve atrophy was present in two out of the three patients with a NF2 positive genetic screen
- OCT-A shows absence of the foveal avascular zone and an anatomical shift of blood vessels

Patient	Age	NF2 genetic screening	Meningioma	ERM	Astrocytic hamartoma	Optic nerve atrophy
1	9	Negative		OD	OS	
2	11	Positive	Present	OS>OD	OS	glioma OD
3	13	Positive (mosaic)			OU	OD
4	34	Positive	Present		OS	OS > OD

Conclusion

- Astrocytic hamartoma with a positive NF2 genetic screen is consistent with previously seen clinical presentation of NF2 in the eye
- Optic atrophy seems to be associated with NF2-related pathoanatomical changes but further evaluation with MRI imaging, medical, and ocular history is needed