Combined Hamartoma of the Retina and RPE in Neurofibromatosis type 2: A Multimodal Imaging Interpretation

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PURPOSE: To report a patient with NF-2 diagnosis and bilateral combined hamartoma of the retina and RPE at multimodal assessment.

METHODS: Case report of 15-year-old girl complaining of divergent deviation progression in OS. BCVA of 20/70 (OD) and 20/150 (OS). Clinical evaluation revealed lagophthalmos in OD due peripheral VII-nerve palsy associated to hypoacusis and several skin café au lait spots. Ocular fundus depicted superficial whitish macular reflex changes associated to deep yellowish dots at parafoveal area (fig 1, 2AE) which corroborate to hyperautofluorescent (fig 2BF), hyperfluorescent (fig 2CG) and hyporeflectance (fig 2DH) by autofluorescent, fluorescein angiography and near-IR, respectively in the multimodal ocular imaging (fig 2). SS-OCT revealing intraretinal disorganization with optical hyperreflectivity mainly at OPL [(omega sign (Ω), fig 3] and SD-OCT showing intraretinal RPE inward changes (fig 4). The OCTA (fig 5) shows capillary plexus dragging up through folded retinal layers (Ω). MRI (fig 6) of the head revealed bilateral vestibular schwannoma, which confirm neurofibromatosis type 2 diagnosis.

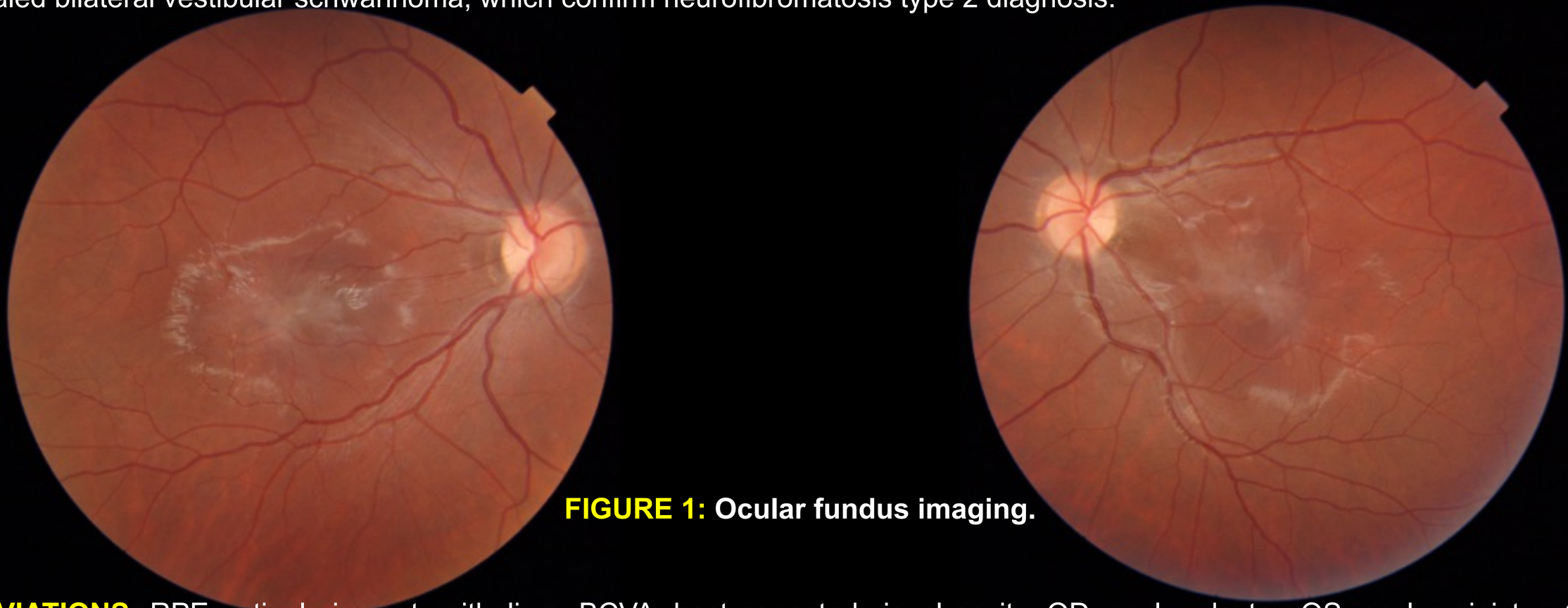


FIGURE 1: Ocular fundus imaging.

ABREVIATIONS: RPE, retinal pigment epithelium; BCVA, best corrected visual acuity; OD, oculus dexter; OS, oculus sinister; OPL, outer plexiform layer; SD-OCT, spectral-domain optical coherence tomography; SS-OCT, sweep source OCT; IR, near-infrared reflectance; AF, autofluorescence; OCTA, OCT angiography; CHRRPE, combined hamartoma of the retina and RPE; MRI, Magnetic ressonance imaging.

FIGURE 2

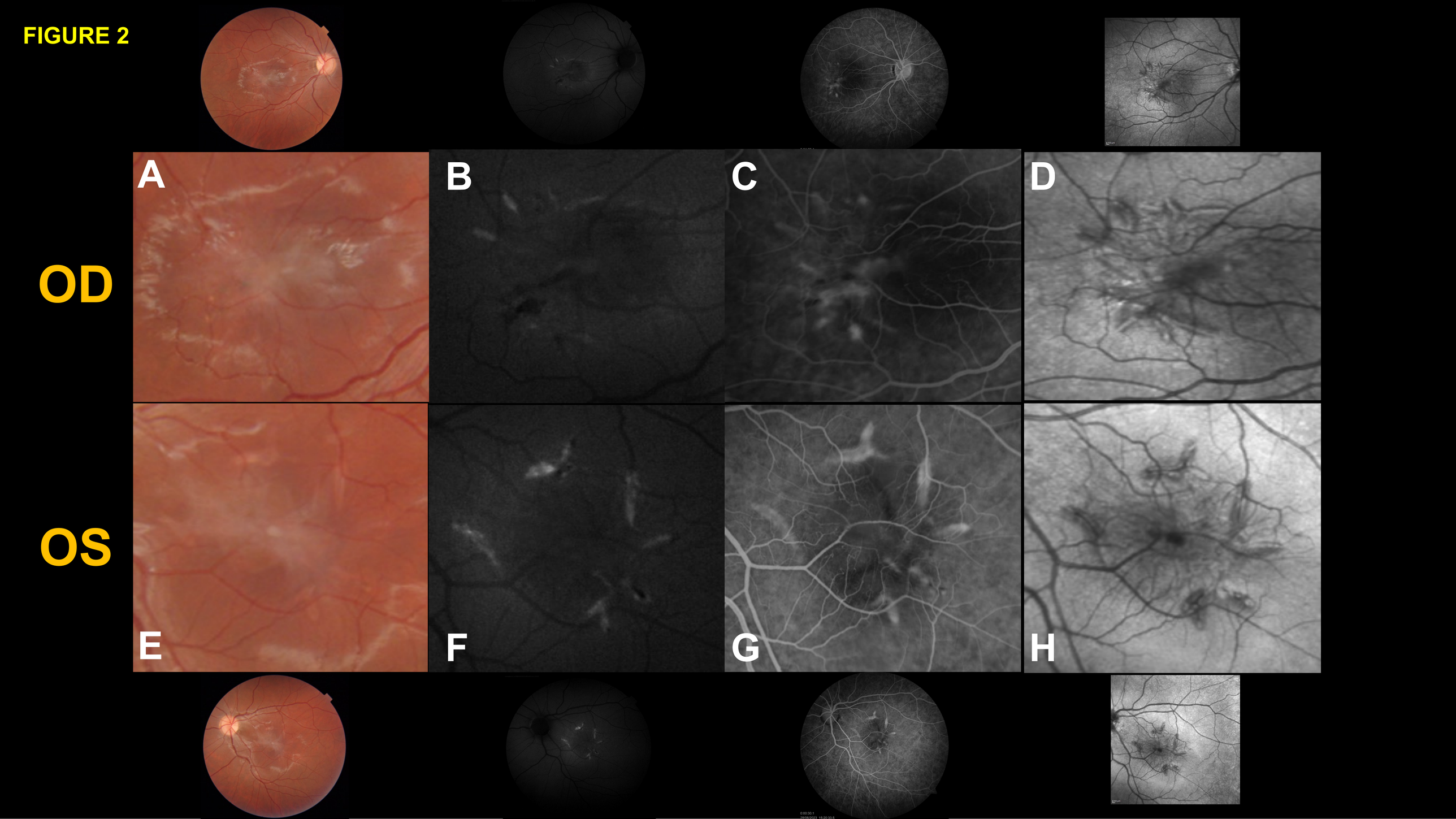


FIGURE 3. SS-OCT

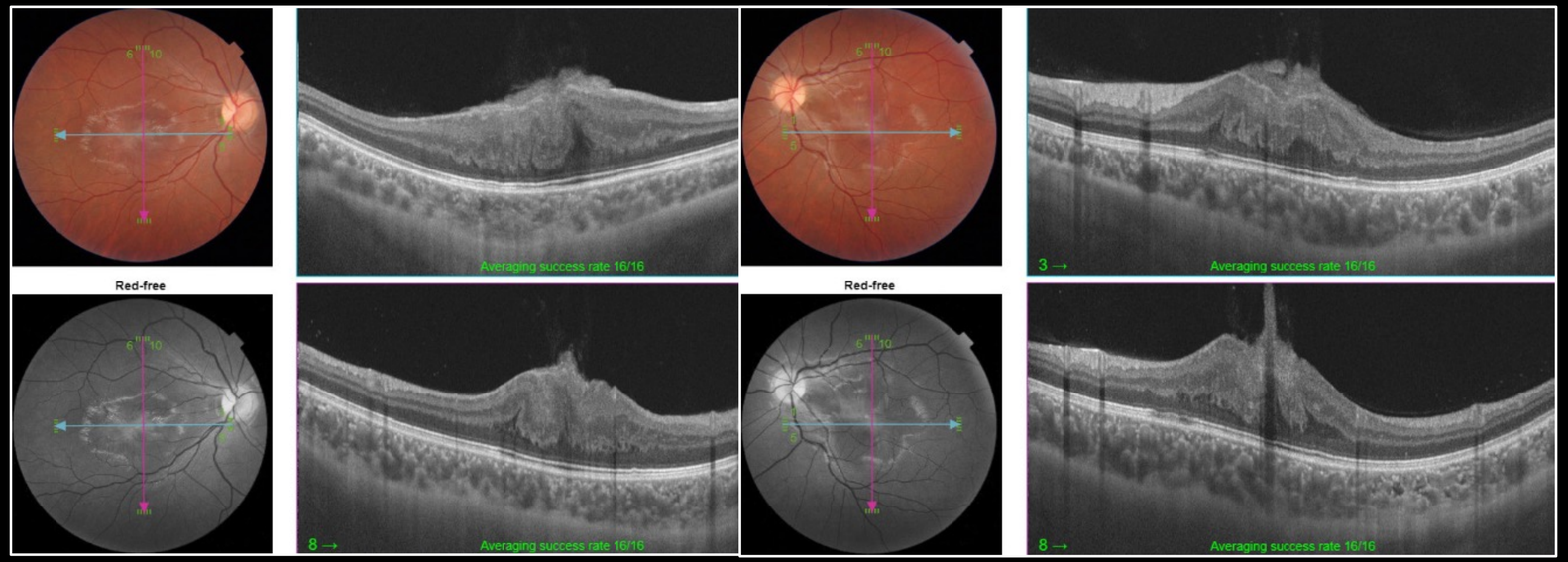


FIGURE 4. SD-OCT

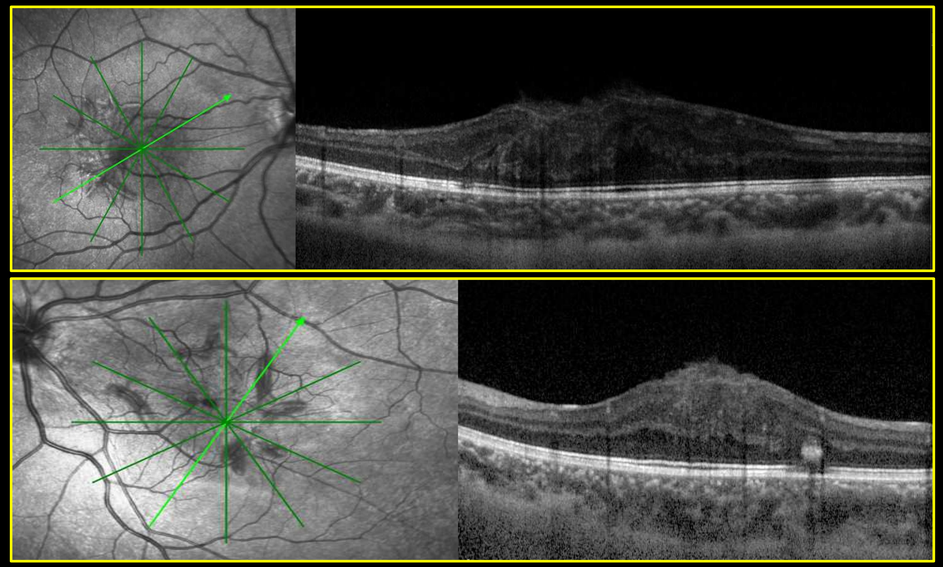
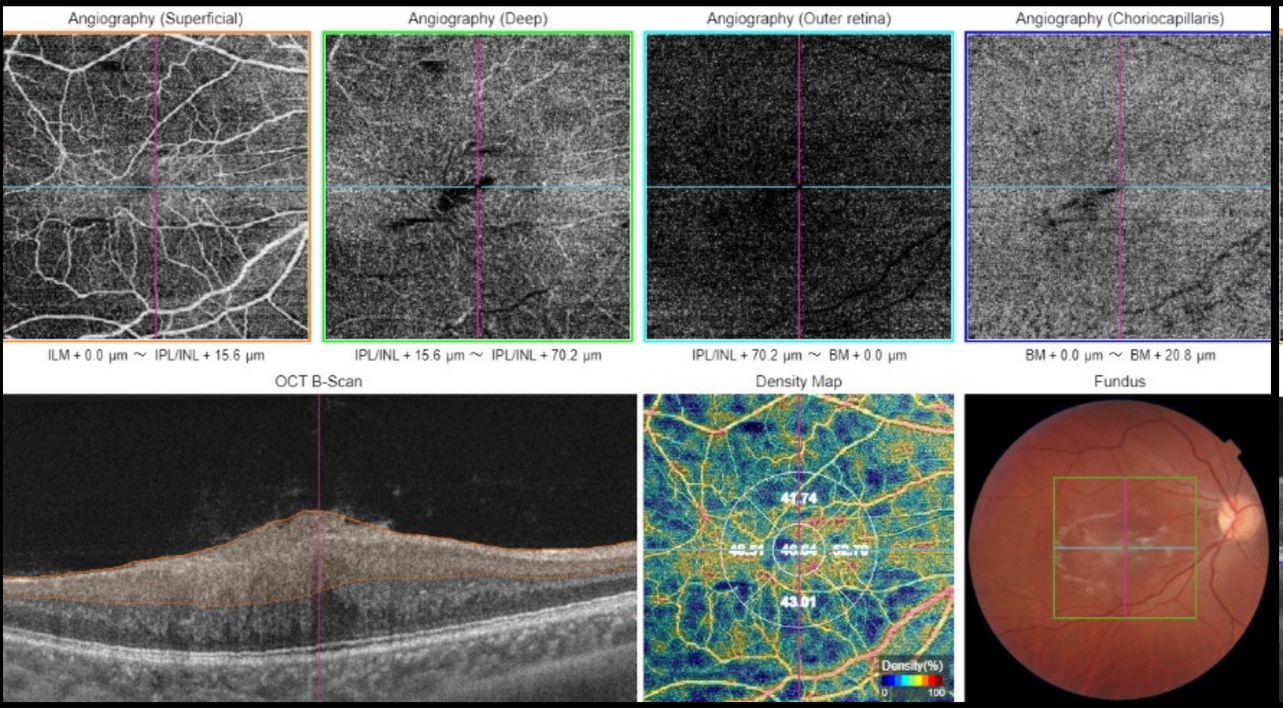
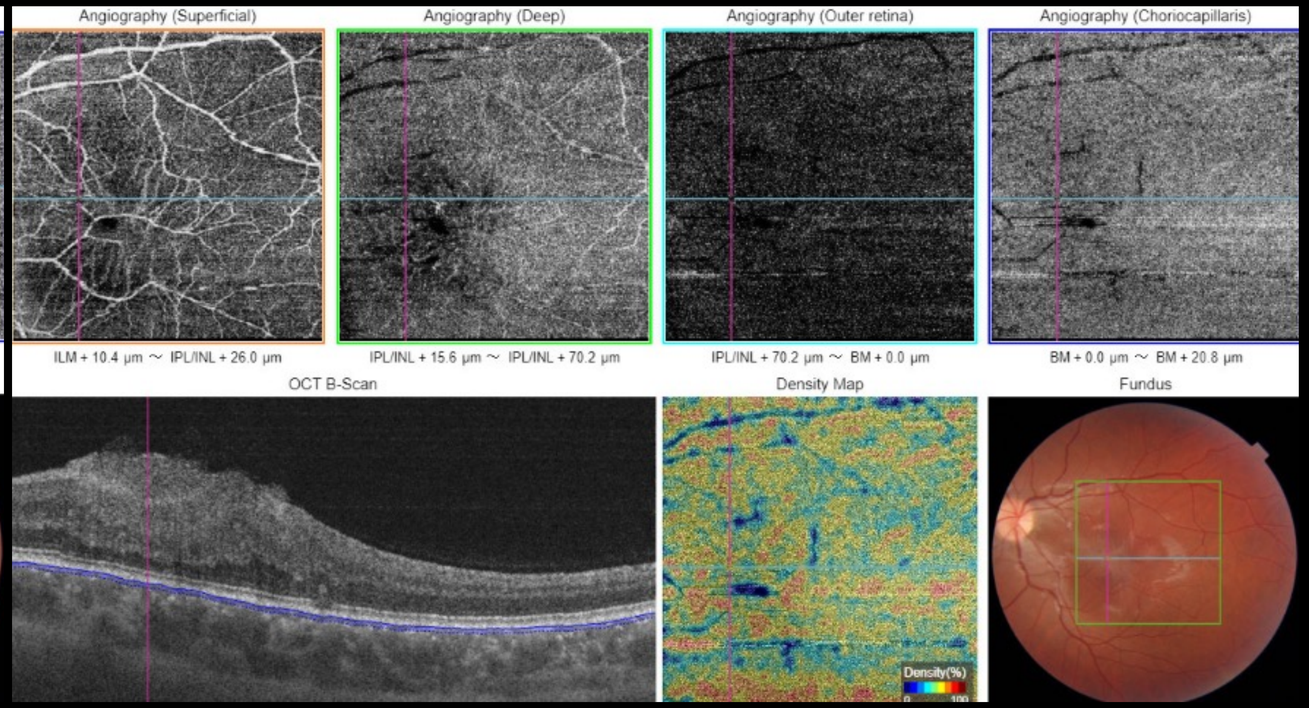


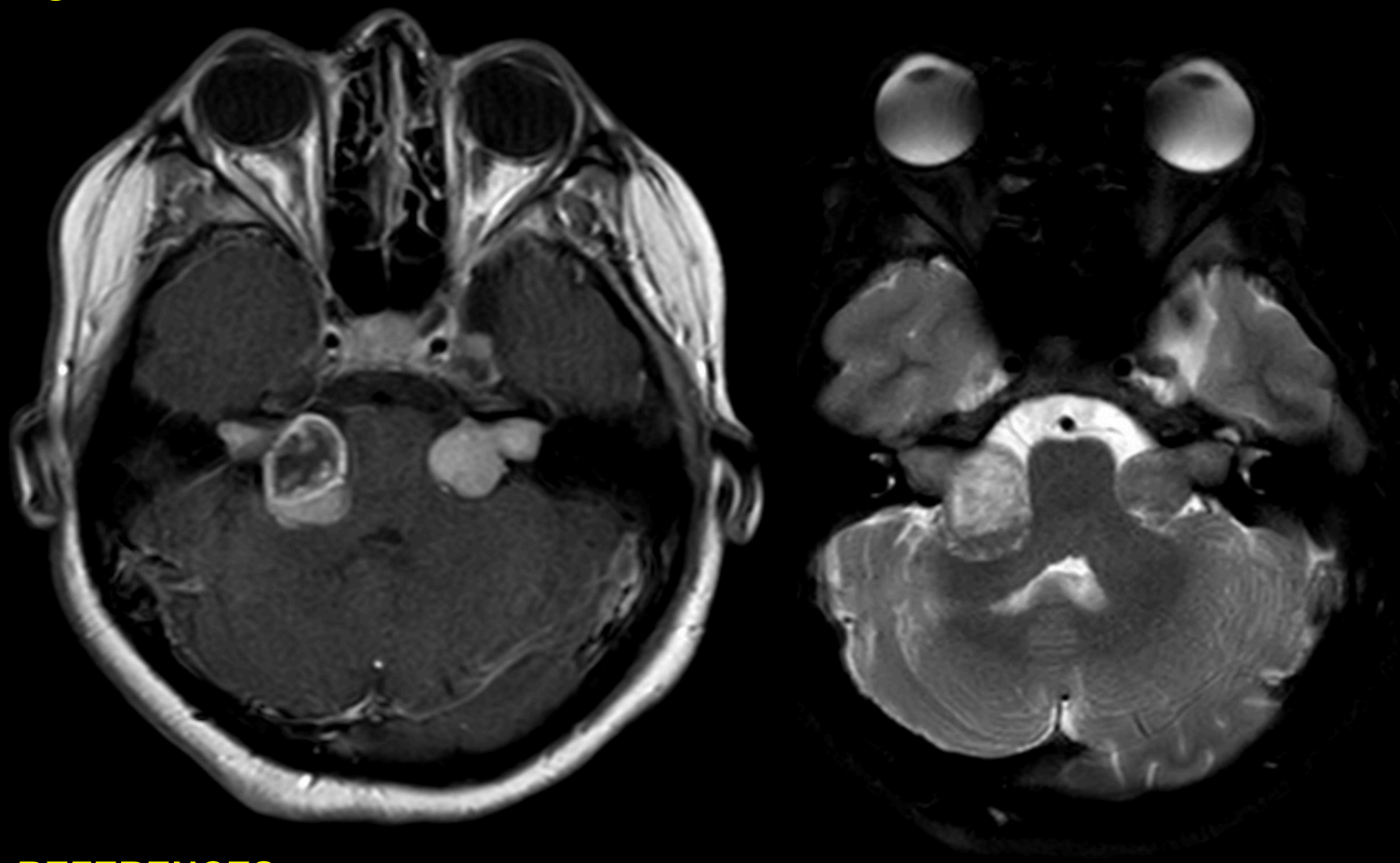
FIGURE 5. OCTA.





RESULTS: The patient is undergoing surgical programming with a neurosurgery team and strabismus follow-up.

Figure 6. MRI



DISCUSSION: Neurofibromatosis type 2 is an autosomal dominant disease, characterized by vestibular schwannomas, cataracts, retinal hamartomas, and tumors of the peripheral and central nervous system, demonstrating a variety of expression. NF2 results from loss-of-function alterations in the NF2 gene on chromosome 22, with consequent dysfunction of its protein product merlin. Despite the high morbidity associated with NF2 in severe cases, management of NF2-associated lesions consists primarily of surgical resection. Some clinical manifestations of NF2, such as ocular abnormalities, can be detected in infancy; therefore, clinical screening for at-risk members of NF2 families can start at birth, with the first magnetic resonance (MRI) scan. The CHRRPE is characterized these lesions based on the predominant tissue subtype, including melanocytic, vascular, or glial.

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