COMBINED HAMARTOMA OF THE RETINA AND RETINA PIGMENT EPITHELIUM WITH OPTIC NERVE INVOLVEMENT: CASE REPORT

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Purpose

To present the follow up of a case of combined hamartoma of the retina and retina pigment epithelium (CHR-RPE) in a 10-year-old girl which was admitted to ophthalmology service after right eye exotropia.

Case report

A healthy 11-year-old girl referenced for an ophthalmologic assessment for the first time in 2020 with right eye exotropia noticed by her mother 4 months prior.

During the ophthalmologic evaluation, her visual acuity (VA) was hand motion (HM) on her right eye (OD) and 1.0 on her left eye (OS).

Ophthalmoscopy of OD revealed a gray elevated lesion with vascular tortuosity and covered by fibroglial tissue. The lesion extended to macula and affected optic nerve.

The OCT showed a hyperreflective lesion in the inner retina, leading to signal attenuation in the underlying tissues. Furthermore, it shows compression of the other retinal layers with invasion of the external retina and presence of intraretinal fluids.

The fluorescein angiography showed areas hyperfluorescence with signs of leakage or neovascularization.

The lesion is still stable after 2 years of follow-up and the patient is followed up with regular retinal imaging.

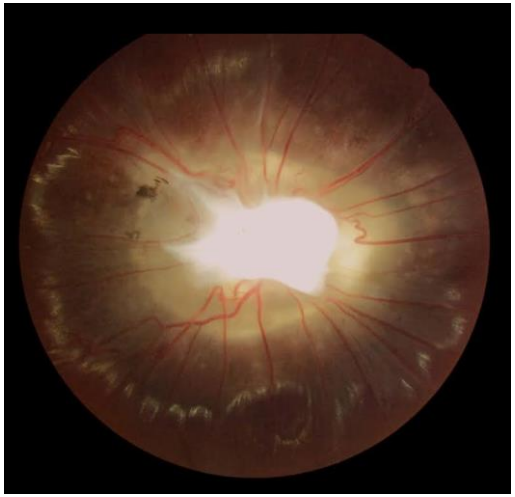

Figure 1: Color fundus of right eye showing optic disc involvement of the CHR-RPE

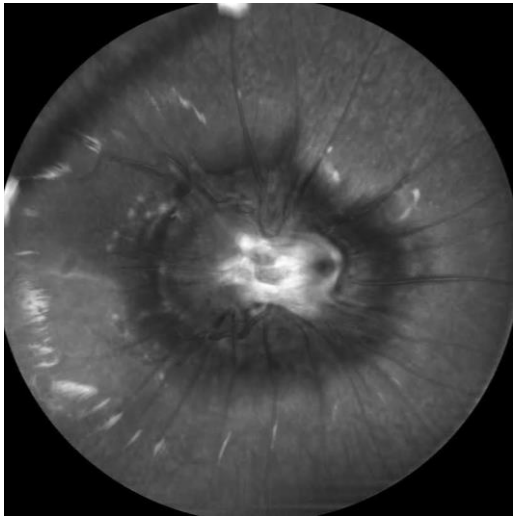

Figure 2: Infrared fundus of right eye showing optic disc involvement of CHR-RPE

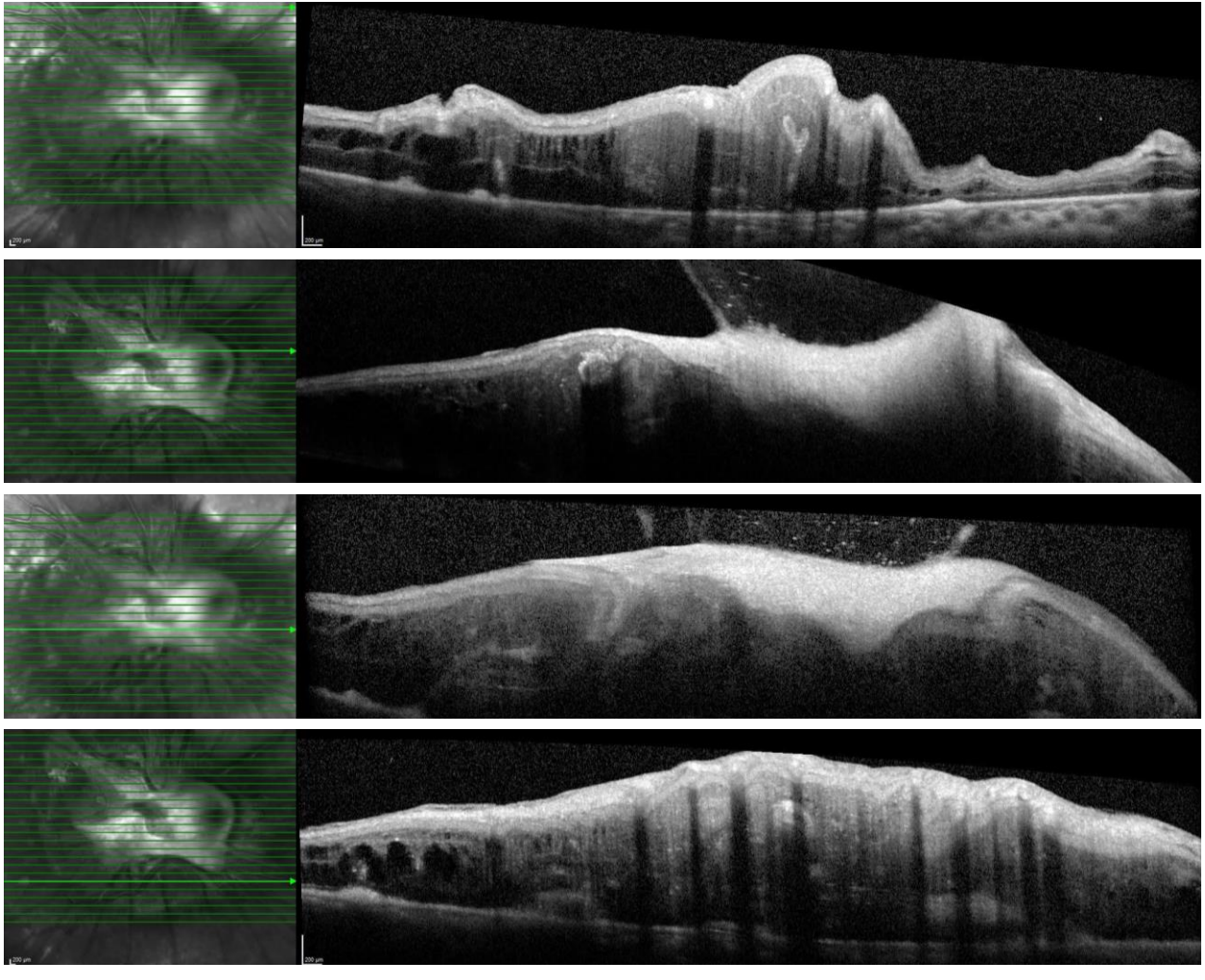

Figure 3,4,5,6: OCT showing typical disorganization of all retinal layers and RPE and also absence of involvement of the underlying choroid.

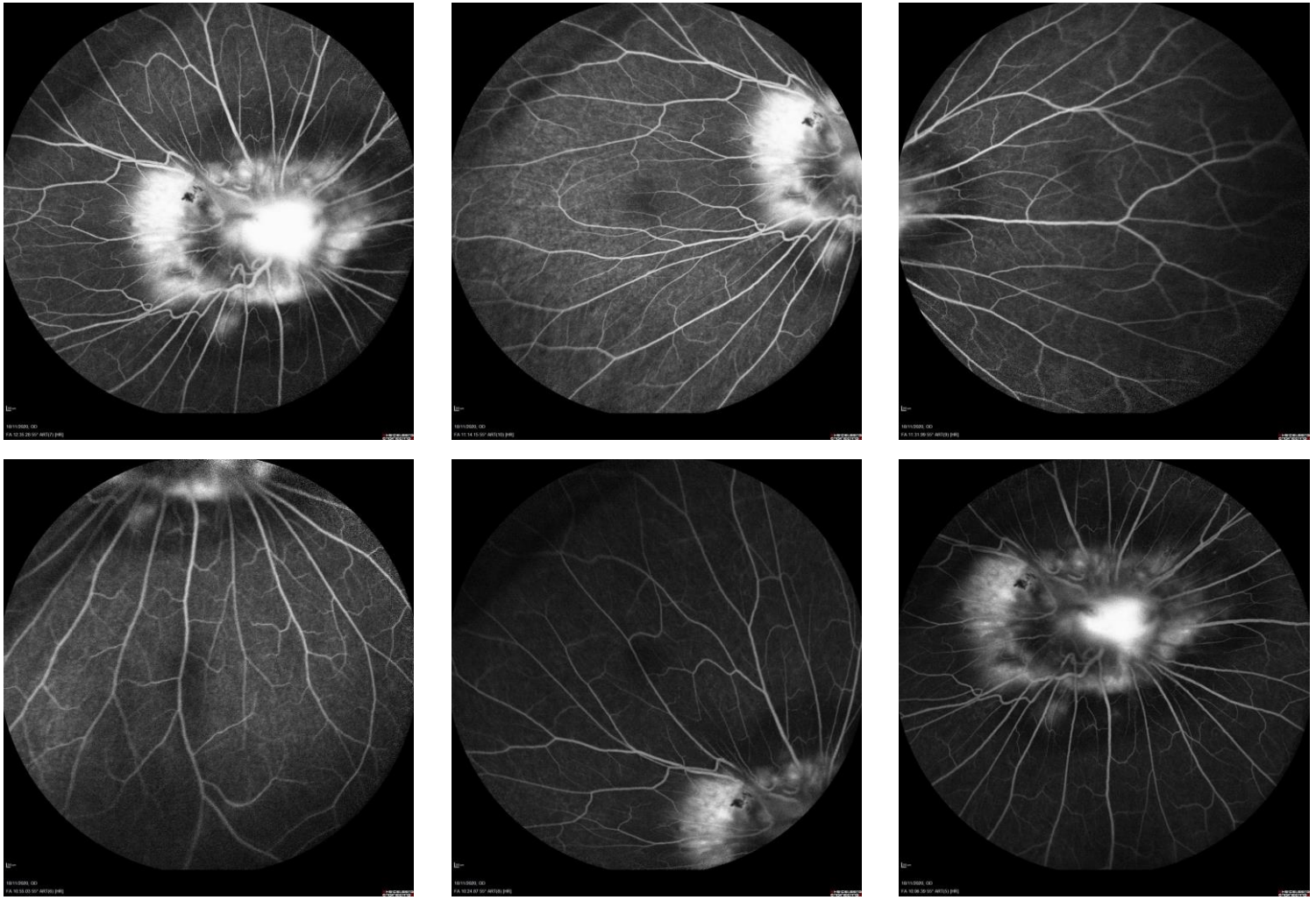

Figure 7-12: Fluorescein Angiography showing areas of hyperfluorescence and leakage

Results

A continuous follow-up of a rare case of a CHR-RPE that affected the optic nerve diagnosed in a 10-year-old girl. The lesion remains stable as well as her visual acuity. The patient continues her follow-up every 6 months in the ocular oncology service with regular retinal imaging.

Discussion

CHR-RPE is congenital, rare and benign tumor with a mixture of retina, RPE, glial tissue and retinal vessels. The tumor is usually asymptomatic and unilateral and is commonly diagnosed in young children with strabismus and reduced VA symptoms. The diagnostic is clinical and OCT and fluorescein angiography helps with it. Differential diagnoses include choroid nevus and melanoma, RPE adenoma and adenocarcinoma

Discussion

The lesion is usually observed twice a year and the treatment is required only if complications arise, such as tractional retinal detachment, epiretinal membranes, choroidal neovascularization and vitreous hemorrhage. The prognosis depends on the size, localization of the tumor and associated complications.

References

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