Choroidal abnormalities in neurofibromatosis: unveil and reveal

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ABSTRACT

To report a case of choroidal abnormalities in a patient with underlying Neurofibromatosis Type 1 (NF1). A 19-year-old boy with underlying bronchial asthma was referred for bilateral eye floaters for 1 year duration. He is not myopic. Visual acuity for both eyes was 6/9. Bilateral eyes have no relative afferent pupillary defect and optic nerve function test was normal. Anterior segment revealed multiple scattered Lisch nodules. Bilateral eye cup disc ratio was 0.6, otherwise unremarkable fundus examination and with no specific retinochoroidal abnormalities. There was no abnormality detected from optical coherence tomography (OCT) of the macula. However near infrared images showed multiple patches of bright choroidal lesion in the posterior pole of both eyes. Subsequently, indocyanine green angiography (ICGA) was done and revealed patches of hypercyanescence at early phase consistent with whitish lesion seen in infrared. Fluorescein angiography revealed no dye leakage or capillary fall-out and no vasculitis. Systemic examination showed multiple small café au lait spots and neurofibromas at the back, neck and upper limb. On further investigation, child's mother had neurofibromas at her face and similar findings in infrared images. It is therefore important to detect choroidal abnormalities in patient with NF1 as it has been added as an ocular diagnostic criteria recently since 2021.