Choroidal abnormalities in Neurofibromatosis type 1

Courtesy of Dr. Fabiana Mallone and Prof. Antonietta Moramarco.
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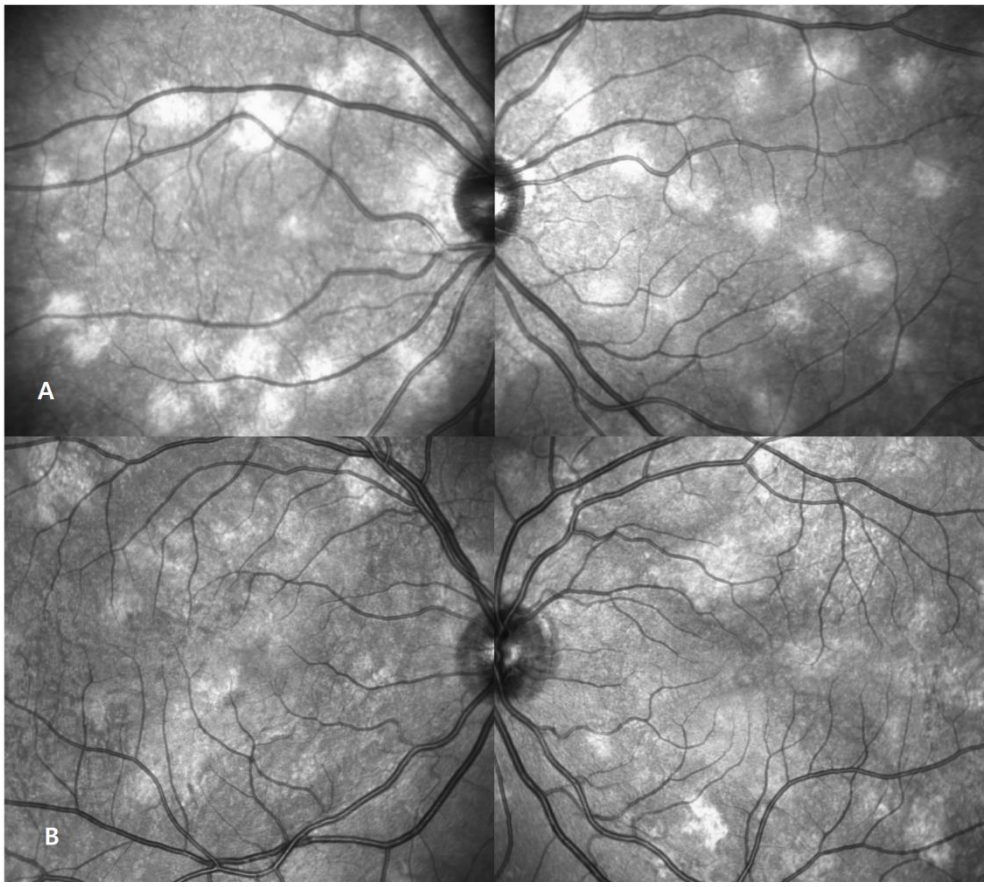

A 28-year-old man (*Patient 1*) and a 31-year-old woman (*Patient 2*), were referred to the eye clinic after dermatological finding of six and eight café-au-lait macules over 15 mm in maximum diameter, respectively. Medical history was unremarkable. Patients were fully asymptomatic and best corrected visual acuity (BCVA) was 20/20 in each eye. Biomicroscopic examination of the anterior segment revealed the presence of seven iris Lisch nodules in Patient 1 and no iris alterations in Patient 2. Mydriatic indirect fundus biomicroscopy was normal in all cases. While performing near-infrared reflectance (NIR) OCT evaluation, multiple bilateral hyperreflective, patchy choroidal abnormalities (CAs) were observed in both patients (Patient 1: top panels **A**, right and left eye; Patient 2: bottom panels **B**, right and left eye). However, as can be seen from comparing the images in top panels **A** with bottom panels **B,** CAs showed two different types of appearance: rounded, well-defined, easily detectable ‘dome-shaped’ CAs in one case (top panels A), and dull, poorly defined and

confluent ‘placcoid’ CAs in the other (bottom panels B). Due to recent introduction of CAs to the current diagnostic criteria for Neurofibromatosis type 1 (NF1), the presence of more than two CAs or more than two iris Lisch nodules, in association with six or more café-au-lait macules, allowed for diagnosis of NF1 in both patients. As observed in our patients, the appearance of CAs can vary considerably from patient to patient in terms of definition and morphology.