Ophthalmologists are frequently consulted to aid in the timely diagnosis of congenital neurofibromatosis type 1 (NF-1). We are asked to perform a slit-lamp examination to determine the presence of iris nodules and to assess the visual acuity and appearance of the optic nerves.

The diagnosis of NF-1 is especially challenging in young children because many clinical findings are not present during childhood but develop with age. On optical coherence tomography (OCT), infrared reflectance is used for imaging deeper structures such as the choroid. Multiple patchy and bright choroidal abnormalities have been found using the infrared mode of OCT in patients with NF-1.

The aim of the study by Goktas et al. in this issue was to investigate the frequency of choroidal abnormalities using infrared reflectance imaging with OCT in pediatric patients with NF-1. Nineteen pediatric patients diagnosed as having NF-1 and 20 control patients underwent infrared reflectance imaging with OCT. Infrared reflectance revealed that 78.9% of the patients with NF-1 had choroidal abnormalities. This is higher than the frequency of Lisch nodules (47.4%) found in this same group. Of interest, the authors found multiple choroidal nodules in 2 patients who were 5 years of age, only one of whom had Lisch nodules. The study demonstrates that hyperreflective choroidal abnormalities in children can be easily observed by infrared light examination.

This noninvasive procedure has the potential to become the eighth criterion used in the clinical diagnosis of NF-1, which currently requires the presence of at least two of seven criteria. These include: café-au-lait spots, intertriginous freckling, Lisch nodules, neurofibromas, optic pathway gliomas, distinctive bony lesions, and a first-degree family relative with NF-1.

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