Choroidal Abnormalities in a Patient with Neurofibromatosis Type 1

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A 35-year-old man with neurofibromatosis type 1 (NF1) was referred to our clinic for an ophthalmological examination. He had no visual symptoms. There were 10 or more Lisch nodules bilaterally. Funduscopy did not show any abnormalities (Picture 1). However, infrared fundus autofluorescence (IR-FAF) demonstrated multiple, bright patchy lesions in the choroid (Picture 2).

Yasunari et al. (1) suggested that choroidal abnormalities

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occur more frequently than Lisch nodules. Viola et al. (2) reported that the highest accuracy is obtained at a cutoff value of 1.5 choroidal nodules detected on IR-FAF. IR-FAF originates from the retinal pigment epithelium (RPE) and choroid, and the melanin content of the RPE and choroid gives rise to the IR-FAF. In NF1 patients, the choroid exhibits proliferation of neural crest-derived melanocytes and thickening of the posterior fundus.

In conclusion, conducting an IR-FAF examination to detect choroidal involvement is useful for confirming the diagnosis of NF1.

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References