Lisch Nodules: A Key Diagnostic Sign of Neurofibromatosis Type 1

Nódulos de Lisch: una clave diagnóstica en neurofibromatosis tipo 1

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Figure 1

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Lisch nodules are included in the diagnostic criteria of type 1 neurofibromatosis when ≥2 occur. They are considered to be melanocytic hamartomas of the iris that are frequently observed before the age of 6 years and increase in number and prevalence with age. They are usually light or dark brown in color and are more apparent in lighter colored eyes.

A 9-year-old girl in follow-up for suspected neurofibromatosis presented with 6 café-au-lait spots measuring >5 mm on the trunk and limbs and axillary freckling. Physical examination revealed 3 light brown spots on the right iris (Fig. 1A). The finding was confirmed by the ophthalmologist and corroborated the diagnosis.

A 72-year-old woman with type 1 neurofibromatosis verified by genetic testing presented with multiple light brown spots on both blue irises (Fig. 1B). In contrast with the previous case, the clinical presentation was very obvious.

Identification of Lisch nodules during the first years of life can confirm a diagnosis in patients who do not yet fulfill a sufficient number of classic criteria. We feel that it is important for the dermatologist to search for this clinical manifestation, which can prove useful in the differential diagnosis of other neurocutaneous syndromes in younger patients.