IMAGES IN DERMATOLOGY

Neurofibromatosis type 1: Multiple cutaneous manifestations

Carolina Narváez,* Javier Rosero,* Ernesto Rosero**



Figure 1: Diffuse nodular swelling and dark brown macules coalescing into patches, with a few skin-colored papules over the malar areas.



Figure 2: Soft papules and nodules, pedunculated and sessile, similar in color to the surrounding skin or slightly pigmented. Café-au-lait macules.

A 45-year-old female with a maternal family history of neurofibromatosis type 1 presented to the dermatology outpatient clinic with multiple long-standing cutaneous lesions that had progressively evolved since childhood.

Physical examination revealed dermatosis localized to the face, characterized by diffuse nodular swelling predominantly affecting the infra-facial region, resulting in asymmetry and dysmorphia. In addition, multiple dark brown hyperpigmented macules coalescing into patches were observed, predominantly on the forehead and bilateral cheeks, along with a few skin-colored papules over the malar areas (Figure 1).

On the posterior thoracic and lumbosacral regions, multiple papules and nodules of varying sizes were distributed asymmetrically. These lesions were soft, pedunculated or sessile, skin-colored or slightly pigmented, and exhibited a positive "buttonhole sign" (invagination of the lesion upon pressure). Numerous café-au-lait macules of varying dimensions were also present, some measuring more than 15 mm in diameter (Figure 2).

Following detailed history and physical examination, the patient fulfilled more than two of the classic NIH (National Institutes of Health) diagnostic criteria for neurofibromatosis type 1. She also had a genetic study performed years earlier confirming a pathogenic NF1 gene mutation. The patient is currently under multidisciplinary management involving dermatology, ophthalmology, neurology, and cardiology.

* Universidad Tecnológica Equinoccial. Quito, Ecuador.
** Independent Researcher. Quito, Ecuador

Correspondence:
Carolina Narváez
caro93narvaez@gmail.com
Quito-Ecuador.
Teléfono: 0987866885

Neurofibromatosis type 1 (NF1) is an autosomal dominant genodermatosis with an incidence of approximately 1 in 3,000 live births. It is caused by mutations in the NF1 gene located on chromosome 17q11.2, which encodes neurofibromin, a tumor suppressor protein that negatively regulates the RAS-MAPK pathway.[1] Cutaneous manifestations represent cardinal findings and include café-au-lait macules (present in >95% of patients), cutaneous and plexiform neurofibromas, axillary and inguinal freckling, and Lisch nodules of the iris.[2] Plexiform neurofibromas, present in 30–50% of cases, are congenital lesions with progressive growth potential and risk of malignant transformation.[3]

Diagnosis is established according to the revised clinical criteria published in 2021 by Legius et al., requiring two or more of the following: ≥6 café-au-lait macules (>5 mm in prepubertal individuals or >15 mm in postpubertal individuals), ≥2 neurofibromas or one plexiform neurofibroma, axillary or inguinal freckling, optic glioma, ≥2 Lisch nodules, characteristic osseous lesion (sphenoid dysplasia or pseudarthrosis), or a first-degree relative with NF1.[4] Molecular confirmation identifies pathogenic NF1 mutations in approximately 95% of cases and is particularly useful in atypical presentations.[5] The main dermatologic complication is malignant transformation of plexiform neurofibromas into malignant peripheral nerve sheath tumors (MPNST), occurring in 8–13% of patients, with warning signs including rapid growth, pain, and changes in consistency.[6]

Management requires multidisciplinary follow-up, including annual dermatologic evaluations to monitor neurofibromas and enable early detection of MPNST, ophthalmologic assessment for optic gliomas, and neurologic evaluation for central nervous system complications. Surgical resection is reserved for symptomatic, disfiguring, or suspicious lesions. Genetic counseling is essential given the autosomal dominant inheritance pattern with complete penetrance.[2–3]

INFORMED CONSENT

The patient included in this study signed an informed consent form authorizing the use of her images and clinical data exclusively for research and scientific publication purposes. It is guaranteed that no personal data were disclosed and that no photographs allowing identification were used.

REFERENCES

1. Gutmann DH, Ferner RE, Listernick RH, et al. Neurofibromatosis type 1. Nat Rev Dis Primers. 2017;3:17004.
2. Bergqvist C, Servy A, Valeyrie-Allanore L, et al. Neurofibromatosis 1 French national guidelines based on an extensive literature review since 2010. Orphanet J Rare Dis. 2020;15(1):37.
3. Haldeman-Englert CR, Scheuerle AE, Schultz M, et al. Diagnosis and management of neurofibromatosis type 1. Transl Pediatr. 2021;10(4):1082-1094.
4. Legius E, Messiaen L, Wolkenstein P, et al. Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genet Med. 2021;23(8):1506-1513.
5. Miller DT, Freedenberg D, Schorry E, et al. Health supervision for children with neurofibromatosis type 1. Pediatrics. 2019;143(5):e20190660.
6. Miettinen MM, Antonescu CR, Fletcher CDM, et al. Histopathologic evaluation of atypical neurofibromatous tumors and their transformation into malignant peripheral nerve sheath tumor in patients with neurofibromatosis 1–a consensus overview. Hum Pathol. 2017;67:1-10.

ORCID

Carolina Narváez https://orcid.org/0009-0003-5405-1882
Javier Rosero https://orcid.org/0000-0002-3635-6653
Ernesto Rosero https://orcid.org/0009-0008-8228-7258

How to cite this article: Narváez C., Rosero J., Rosero E. Neurofibromatosis tipo 1: manifestaciones cutáneas múltiples. - Dermatología en Imágenes. Rev Dermatol Cent Úraga. 2026;8(1).

Date of receipt: 14/9/2025
Date of acceptance: 2/2/2026