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DERMATOLOGY IMAGES

Muir-Torre syndrome type II: beyond mismatch repair deficiency

Síndrome de Muir–Torre tipo II: uma via distinta de reparação do ADN

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Muir-Torre syndrome (MTS) is a hereditary cancer syndrome characterized by sebaceous neoplasms and internal malignancies, most commonly colorectal cancer. Classical MTS is inherited in an autosomal dominant pattern and associated with pathogenic variants in DNA mismatch repair genes (*MLH1*, *MSH2*, *MSH6*, and *PMS2*), typically resulting in microsatellite instability.[1] In contrast, a rare autosomal recessive form, historically designated MTS type II, results from biallelic pathogenic variants in *MUTYH*, a base excision repair gene, and is generally microsatellite stable.[2,3]

We report a 72-year-old man with *MUTYH*-associated polyposis. Genetic testing identified two pathogenic MUTYH variants in compound heterozygosity. His medical history included subtotal colectomy for multiple colorectal adenomas and segmental enterectomy for stage II mucinous adenocarcinoma of the jejunum. His family history included colorectal cancer in his father and sister and hemicolectomy for multiple colorectal adenomas in a brother.

Cutaneous examination revealed numerous yellowish papules over the frontal and bilateral malar regions (Fig. 1). A prominent, intermittently bleeding lesion on the left frontal region (arrow) was excised, and histopathological examination revealed a sebaceous adenoma.

The coexistence of a sebaceous neoplasm and visceral malignancy in a patient with biallelic pathogenic *MUTYH* variants supports the diagnosis of autosomal recessive MTS type II. The patient remains under surveillance and is undergoing CO_2 laser treatment of the most bothersome facial lesions.

This case highlights the cutaneous presentation and genetic heterogeneity of MTS, drawing attention to the less recognized *MUTYH*-associated form, which arises through a DNA repair pathway distinct from mismatch repair deficiency.

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Figure 1. Multiple yellowish facial papules involving the frontal and bilateral malar regions. The arrow indicates the histologically confirmed sebaceous adenoma.

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Conflicts of interest

The authors declare no conflicts of interest.

Ethical considerations

Protection of human subjects and animals. The authors declare to have followed the ethical standards of the relevant experimentation committee, according to the World Medical Association and the Declaration of Helsinki. The procedures were approved by the institutional Ethics Committee.

Confidentiality, informed consent, and ethical approval. The authors have followed their institution's confidentiality protocols, obtained informed consent from all patients, and secured approval from the Ethics Committee. SAGER guidelines have been followed as applicable to the nature of the study.

Declaration on the use of artificial intelligence. The authors declare that no generative artificial intelligence was used in the writing or creation of the content of this manuscript.

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