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Unilateral vestibular schwannoma and meningiomas in a patient with PIK3CA-related segmental overgrowth: Co-occurrence of mosaicism for 2 rare disorders.

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A 28-year-old female with PIK3CA-related segmental overgrowth presented with headaches. She also had a unilateral vestibular schwannoma (VS), as well as 3 small (<2 cm) meningiomas, which according to the Manchester consensus diagnostic criteria for neurofibromatosis 2 (NF2) is sufficient for a clinical diagnosis. Analysis of blood revealed a mosaic PIK3CA c.2740G>A (p.Gly914Arg) mutation, confirming the diagnosis of PIK3CA-related overgrowth, but no mutations in NF2 were detected. Although VS has not previously been reported in PIK3CA-related segmental overgrowth, meningiomas have, raising the question of whether this patient's VS and meningiomas represent coincidental NF2 or phenotypic extension of her overgrowth syndrome. Genetic analysis of the VS revealed a heterozygous NF2 mutation c.784C>T (p.Arg262Ter) and loss of a portion of 22q, including NF2, SMARCB1, and LZTR1 genes. These results suggest that the patient has 2 different mosaic disorders, NF2 and PIK3CA-related overgrowth. The PIK3CA mutation was also present in the VS. Confirmation of the clinical diagnosis of mosaic NF2 in this patient has implications for monitoring and highlights the possibility of co-occurrence of mosaicism for multiple rare disorders in a single patient.

KEYWORDS: NF2; PIK3CA; mosaic; neurofibromatosis; overgrowth

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