

Perry disease in an Argentine family due to the DCTN1 p.G67D variant

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Resumen

Sequence variants in exon 2 of the Dynactin-1 gene (DCTN1) have been reported as causative of Perry (disease) Syndrome (PeD, MIM 168605), a rare neurodegenerative disorder characterized by parkinsonism, psychiatric symptoms, weight loss and hypoventilation [1]. Here, we report the third family with the p.G67D variant in DCTN1. An Argentine family from the northeastern province of Misiones was evaluated at multiple centers. Three individuals were available for clinical, neuropsychological and genetic testing including the proband (Fig. 1, individual III.2), a symptomatic sister (III.3), and an asymptomatic brother (III.4). All family members and/or caregivers gave their informed consent to participate. This investigation was approved by the Institutional Ethics Committee.

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