NOTCH2NLC

NOTCH2NLC is 1 of 3 nearly identical, functional human NOTCH2 (600275)-like genes on chromosome 1q21.1 The NOTCH2L proteins appear to regulate Notch signaling pathway and promote cortical neurogenesis.

The objective of Yau et al. in a study was to determine the prevalence of the GGC-repeat expansion in NOTCH2NLC in whites presenting with movement disorders.

They searched for the GGC-repeat expansion in NOTCH2NLC using repeat-primed polymerase chain reaction in 203 patients with essential tremor, 825 patients with PD, 194 patients with spinocerebellar ataxia, 207 patients with “possible” or “probable” MSA, and 336 patients with pathologically confirmed MSA. They also screened 30,008 patients enrolled in the 100,000 Genomes Project for the same mutation using ExpansionHunter, followed by a repeat-primed polymerase chain reaction. All possible expansions were confirmed by Southern blotting and/or long-read sequencing.

They identified 1 patient who carried the NOTCH2NLC mutation in the essential tremor cohort, and 1 patient presenting with recurrent encephalopathy and postural tremor/parkinsonism in the 100,000 Genomes Project.

GGC-repeat expansion in NOTCH2NLC is rare in whites presenting with movement disorders. In addition, existing whole-genome sequencing data are useful in case ascertainment [1].

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Yau WY, Vandrovcova J, Sullivan R, Chen Z, Zecchinelli A, Cilia R, Stefano D, Murray M, Carmona S; Genomics England Research Consortium, Chelban V, Ishiura H, Tsuji S, Jaunmuktane Z, Turner C, Wood NW, Houlden H. Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients With Movement Disorders. Mov Disord. 2020 Oct 7. doi: 10.1002/mds.28302. Epub ahead of print. PMID: 33026126.