

Charlesworth et al. combined linkage analysis with [whole-exome sequencing](#) of two individuals to identify candidate causal variants in a moderately-sized UK kindred exhibiting [autosomal](#)-dominant inheritance of [craniocervical dystonia](#). Subsequent [screening](#) of these candidate causal variants in a large number of familial and sporadic cases of [cervical dystonia](#) led to the identification of a total of six putatively pathogenic mutations in [ANO3](#), a gene encoding a predicted Ca(2+)-gated chloride [channel](#) that they show to be highly expressed in the [striatum](#). Functional studies using Ca(2+) imaging in case and control fibroblasts demonstrated clear abnormalities in endoplasmic-reticulum-dependent Ca(2+) signaling. They conclude that mutations in ANO3 are a cause of autosomal-dominant craniocervical dystonia. The locus [DYL23](#) has been reserved as a synonym for this gene. The implication of an ion channel in the pathogenesis of dystonia provides insights into an alternative mechanism that opens fresh avenues for further research ((Charlesworth G, Plagnol V, Holmström KM, Bras J, Sheerin UM, Preza E, Rubio-Agusti I, Ryten M, Schneider SA, Stamelou M, Trabzuni D, Abramov AY, Bhatia KP, Wood NW. Mutations in ANO3 cause dominant craniocervical dystonia: ion channel implicated in pathogenesis. *Am J Hum Genet*. 2012 Dec 7;91(6):1041-50. doi: 10.1016/j.ajhg.2012.10.024. Epub 2012 Nov 29. PMID: 23200863; PMCID: PMC3516598.)

From:

<https://neurosurgerywiki.com/wiki/> - **Neurosurgery Wiki**

Permanent link:

<https://neurosurgerywiki.com/wiki/doku.php?id=dyl23>

Last update: **2024/06/07 02:52**

