Dystonia 24

Mutations or variations in the ANO3 gene have been associated with certain neurological conditions. For example, variations in ANO3 have been linked to a movement disorder known as autosomal dominant craniocervical dystonia (formerly known as Dystonia 24 or DYT24). craniocervical dystonia is characterized by involuntary muscle contractions in the neck and face, leading to abnormal movements and postures.

Dystonia 24 was first reported in 2000 as an autosomal dominant cause of dystonia caused by variants in the ANO3 gene [1)].

Although many adults have been described with dystonia 24, since 2014, an increasing number of children have also been reported. Dystonia 24 should also be considered in the differential of a child with unexplained dystonia.

DYT24 is associated with mutations in the ANO3 gene, which encodes the Anoctamin 3 protein. Anoctamins are involved in ion channel regulation and are found in various tissues, including the brain.

DYT24 is a type of autosomal dominant dystonia, meaning that a single copy of the mutated gene from either parent is sufficient to cause the disorder. This form of dystonia often involves focal or segmental dystonia affecting the neck, face, or larynx. The age of onset can vary, but symptoms typically begin in adulthood.

It's important to note that research in the field of genetics and dystonia is ongoing, and classifications or understanding of specific genetic forms of dystonia may evolve. If you or someone you know is affected by DYT24 or if there have been updates in research or terminology since my last update, consulting with a healthcare professional or a genetic counselor would provide the most accurate and up-to-date information.

It's important to note that genetic information and its associations with specific conditions may be subject to ongoing research, and the understanding of the ANO3 gene's role in health and disease may evolve.

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 DYT23 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 [2)]

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Treatment

see Dystonia treatment.

Case reports

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[2] [3]

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