CHAT gene

choline O-acetyltransferase

Normal Function

The *CHAT* gene provides instructions for making a protein called choline acetyltransferase. This protein is located at the ends of nerve cells in specialized areas called presynaptic terminals. Choline acetyltransferase facilitates the production of a molecule called acetylcholine. Acetylcholine is essential for normal muscle movement. When acetylcholine is released from the presynaptic terminal, it attaches (binds) to a receptor protein located in the membrane of muscle cells. When acetylcholine binds to its receptor protein, specialized channels in the receptor then open, allowing certain charged atoms (ions) to flow into and out of muscle cells. The flow of these ions allows for muscle contraction and relaxation, resulting in muscle movement.

Health Conditions Related to Genetic Changes

Congenital myasthenic syndrome

More than 30 mutations in the *CHAT* gene have been found to cause congenital myasthenic syndrome. Most of these mutations replace single DNA building blocks (nucleotides) in the *CHAT* gene. The mutations lead to decreased production of choline acetyltransferase or the production of a protein with decreased ability to aid in the production of acetylcholine. The resulting lack of acetylcholine decreases the availability of open receptors, impairing ion flow through muscle cells. A reduction in muscle cell ion flow decreases muscle movement leading to muscle weakness characteristic of congenital myasthenic syndrome. In addition, people with congenital myasthenic syndrome who have mutations in the *CHAT* gene are more likely than affected individuals with mutations in other genes to have short pauses in breathing (apnea), but the cause for this association is unclear.

Other Names for This Gene

- acetyl CoA:choline O-acetyltransferase
- CHOACTASE
- choline acetylase
- CLAT_HUMAN
- CMS1A

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CHAT (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1103[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CHAT%5BTIAB%5D%29+OR+%28choline+acetyltransferase%5BTIAB%5D%29+NOT+%28Cri+du+chat%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CHOLINE ACETYLTRANSFERASE; CHAT (https://omim.org/entry/118490)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1103)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CHAT[gene])

References

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- Engel AG, Shen XM, Selcen D, Sine SM. What have we learned from the congenitalmyasthenic syndromes. J Mol Neurosci. 2010 Jan;40(1-2):143-53. doi:10.1007/s12031-009-9229-0. Epub 2009 Aug 18. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19688192) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3050586/)
- Schara U, Christen HJ, Durmus H, Hietala M, Krabetz K, Rodolico C, SchreiberG, Topaloglu H, Talim B, Voss W, Pihko H, Abicht A, Muller JS, Lochmuller H.Long-term follow-up in patients with congenital myasthenic syndrome due to CHATmutations. Eur J Paediatr Neurol. 2010 Jul;14(4):326-33. doi:10.1016/j.ejpn.2009.09.009. Epub 2009 Nov 8. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19900826)

Genomic Location

The *CHAT* gene is found on chromosome 10 (https://medlineplus.gov/genetics/chromosome/10/).

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