CHD2 gene

chromodomain helicase DNA binding protein 2

Normal Function

The *CHD2* gene provides instructions for making a protein called chromodomain DNA helicase protein 2. This protein is found in cells throughout the body and regulates gene activity (expression) through a process known as chromatin remodeling. Chromatin is the complex of DNA and proteins that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. When DNA is tightly packed, gene expression is lower than when DNA is loosely packed. Chromodomain DNA helicase protein 2 appears to play an important role in the brain, although its function is not well understood. Research suggests that it may help control development or functioning of nerve cells (neurons).

Health Conditions Related to Genetic Changes

CHD2 myoclonic encephalopathy

At least 30 mutations in the *CHD2* gene have been found to cause *CHD2* myoclonic encephalopathy, a condition characterized by recurrent seizures (epilepsy), abnormal brain function (encephalopathy), and intellectual disability beginning in childhood. About half of these mutations delete pieces of DNA from the *CHD2* gene. These and other *CHD2* gene mutations either prevent the production of any chromodomain DNA helicase protein 2 or lead to the production of a nonfunctional version of the protein. As a result, chromatin remodeling and gene expression normally regulated by the chromodomain DNA helicase protein 2 are disrupted. It is unclear why *CHD2* gene mutations seem to only affect nerve cells in the brain or how they lead to the signs and symptoms of *CHD2* myoclonic encephalopathy.

Autism spectrum disorder

At least nine *CHD2* gene mutations have been identified in people with autism spectrum disorder (ASD), a varied condition characterized by impaired social skills, communication problems, and repetitive behaviors. Mutations in the *CHD2* gene impair the function of the CHD2 protein, resulting in small changes in the expression of many genes, the effects of which combine to affect brain development and increase the risk of ASD. Normal variations in other genes, as well as environmental risk factors, such as parental age, birth complications, and others that have not been identified, also affect

an individual's risk of developing this complex condition.

Lennox-Gastaut syndrome

MedlinePlus Genetics provides information about Lennox-Gastaut syndrome

Other Names for This Gene

- ATP-dependent helicase CHD2
- CHD-2
- EEOC
- FLJ38614

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CHD2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1106[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CHD2%5BTIAB%5D%29+OR+%28chromodomain+helicase+DNA+binding+protein+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- CHROMODOMAIN HELICASE DNA-BINDING PROTEIN 2; CHD2 (https://omim.org/entry/602119)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1106)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CHD2[gene])

References

- O'Roak BJ, Stessman HA, Boyle EA, Witherspoon KT, Martin B, Lee C, Vives L,Baker C, Hiatt JB, Nickerson DA, Bernier R, Shendure J, Eichler EE. Recurrent denovo mutations implicate novel genes underlying simplex autism risk. Nat Commun.2014 Nov 24;5:5595. doi: 10.1038/ncomms6595. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25418537) or Free article on PubMed Central (https

://www.ncbi.nlm.nih.gov/pmc/articles/PMC4249945/)

- Pinto D, Delaby E, Merico D, Barbosa M, Merikangas A, Klei L,Thiruvahindrapuram B, Xu X, Ziman R, Wang Z, Vorstman JA, Thompson A, Regan R,Pilorge M, Pellecchia G, Pagnamenta AT, Oliveira B, Marshall CR, Magalhaes TR,Lowe JK, Howe JL, Griswold AJ, Gilbert J, Duketis E, Dombroski BA, De Jonge MV,Cuccaro M, Crawford EL, Correia CT, Conroy J, Conceicao IC, Chiocchetti AG, CaseyJP, Cai G, Cabrol C, Bolshakova N, Bacchelli E, Anney R, Gallinger S, CotterchioM, Casey G, Zwaigenbaum L, Wittemeyer K, Wing K, Wallace S, van Engeland H,Tryfon A, Thomson S, Soorya L, Roge B, Roberts W, Poustka F, Mouga S, Minshew N, McInnes LA, McGrew SG, Lord C, Leboyer M, Le Couteur AS, Kolevzon A, JimenezGonzalez P, Jacob S, Holt R, Guter S, Green J, Green A, Gillberg C, Fernandez BA,Duque F, Delorme R, Dawson G, Chaste P, Cafe C, Brennan S, Bourgeron T, BoltonPF, Bolte S, Bernier R, Baird G, Bailey AJ, Anagnostou E, Almeida J, Wijsman EM,Vieland VJ, Vicente AM, Schellenberg GD, Pericak-Vance M, Paterson AD, Parr JR,Oliveira G, Nurnberger JI, Monaco AP, Maestrini E, Klauck SM, Hakonarson H,Haines JL, Geschwind DH, Freitag CM, Folstein SE, Ennis S, Coon H, Battaglia A,Szatmari P, Sutcliffe JS, Hallmayer J, Gill M, Cook EH, Buxbaum JD, Devlin B,Gallagher L, Betancur C, Scherer SW. Convergence of genes and cellular pathwaysdysregulated in autism spectrum disorders. Am J Hum Genet. 2014 May1;94(5):677-94. doi: 10.1016/j.ajhg.2014.03.018. Epub 2014 Apr 24. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24768552) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4067558/)
- Suls A, Jaehn JA, Kecskes A, Weber Y, Weckhuysen S, Craiu DC, Siekierska A, Djemie T, Afrikanova T, Gormley P, von Spiczak S, Kluger G, Iliescu CM, Talvik T, Talvik I, Meral C, Caglayan HS, Giraldez BG, Serratosa J, Lemke JR,Hoffman-Zacharska D, Szczepanik E, Barisic N, Komarek V, Hjalgrim H, Moller RS,Linnankivi T, Dimova P, Striano P, Zara F, Marini C, Guerrini R, Depienne C,Baulac S, Kuhlenbaumer G, Crawford AD, Lehesjoki AE, de Witte PA, Palotie A,Lerche H, Esguerra CV, De Jonghe P, Helbig I; EuroEPINOMICS RES Consortium. Denovo loss-of-function mutations in CHD2 cause a fever-sensitive myoclonicepileptic encephalopathy sharing features with Dravet syndrome. Am J Hum Genet.2013 Nov 7;93(5):967-75. doi: 10.1016/j.ajhg.2013.09.017. Epub 2013 Oct 24. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24207121) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3824114/)
- Thomas RH, Zhang LM, Carvill GL, Archer JS, Heavin SB, Mandelstam SA, Craiu D, Berkovic SF, Gill DS, Mefford HC, Scheffer IE; EuroEPINOMICS RES Consortium. CHD2myoclonic encephalopathy is frequently associated with self-induced seizures. Neurology. 2015 Mar 3;84(9):951-8. doi: 10.1212/WNL.0000000000001305. Epub 2015Feb 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25672921) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4351660/)
- Trivisano M, Striano P, Sartorelli J, Giordano L, Traverso M, Accorsi P,Cappelletti S, Claps DJ, Vigevano F, Zara F, Specchio N. CHD2 mutations are arare cause of generalized epilepsy with myoclonic-atonic seizures. EpilepsyBehav. 2015 Oct;51:53-6. doi: 10.1016/j.yebeh.2015.06.029. Epub 2015 Aug 7. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/26262932)

Genomic Location

The *CHD2* gene is found on chromosome 15 (https://medlineplus.gov/genetics/chromosome/15/).

Last updated June 1, 2017