CHD3 gene

chromodomain helicase DNA binding protein 3

Normal Function

The *CHD3* gene provides instructions for making a protein that regulates gene activity (expression) by a process known as chromatin remodeling. Chromatin is the complex of DNA and protein 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. Chromatin remodeling is one way gene expression is regulated during development. The CHD3 protein helps with chromatin remodeling by moving components called nucleosomes, that help bundle DNA in a tight package. Moving nucleosomes helps make DNA more accessible for gene expression. The CHD3 protein provides energy for this remodeling by breaking down a molecule called ATP.

Through its ability to regulate gene activity, the CHD3 protein is involved in many processes during development, including maintenance of the structure and integrity of DNA, the maturation process that determines the type of cell an immature cell will ultimately become (cell fate determination), and the growth of cells as they progress through the step-by-step process they take to replicate themselves (the cell cycle).

Health Conditions Related to Genetic Changes

Snijders Blok-Campeau syndrome

More than 25 mutations in the *CHD3* gene have been found to cause Snijders Blok-Campeau syndrome. This condition is characterized by intellectual disability, developmental delay, speech delay, and distinctive facial features.

Most *CHD3* gene mutations change single protein building blocks (amino acids) in the CHD3 protein. The majority of mutations alter an area of the protein that is involved in breaking down ATP to provide the energy for chromatin remodeling. *CHD3* gene mutations can either increase or decrease the protein's chromatin remodeling activity. These changes seem to affect the activity of genes that direct the development of many different organs and tissues before birth. It is unclear how increased and decreased protein function both lead to the signs and symptoms of Snijders Blok-Campeau syndrome.

Other Names for This Gene

- Mi-2a
- Mi2-ALPHA

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CHD3 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1107[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CHD3%5BTIAB%5D%29+OR+%28chromodomain+helicase+DNA+binding+protein+3%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+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CHROMODOMAIN HELICASE DNA-BINDING PROTEIN 3; CHD3 (https://omim.org/entry/602120)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1107)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CHD3[gene])

References

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- Torchy MP, Hamiche A, Klaholz BP. Structure and function insights into theNuRD chromatin remodeling complex. Cell Mol Life Sci. 2015 Jul;72(13):2491-507.doi: 10.1007/s00018-015-1880-8. Epub 2015 Mar 22. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25796366)

Genomic Location

The *CHD3* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

Last updated July 1, 2020