

## CHD8 gene

chromodomain helicase DNA binding protein 8

### Normal Function

The *CHD8* 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 CHD8 protein is thought to affect the expression of many other genes that are involved in brain development before birth. In particular, the CHD8 protein and the genes it regulates likely help control the development of neural progenitor cells, which give rise to nerve cells (neurons), and the growth and division (proliferation) and maturation (differentiation) of neurons. In this way, the CHD8 protein helps to control the number of neurons in the brain and prevent overgrowth.

### Health Conditions Related to Genetic Changes

#### Autism spectrum disorder

More than 30 *CHD8* 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 *CHD8* gene impair the function of the CHD8 protein, and may interfere with its ability to help control the number and growth of neurons in the brain. Excess neurons and overgrowth in parts of the brain are associated with ASD, but the relationship between these abnormalities and the behavioral features of the disorder is unknown.

### Other Names for This Gene

- ATP-dependent helicase CHD8
- AUTS18
- axis duplication inhibitor
- chromodomain-helicase-DNA-binding protein 8 isoform 1
- chromodomain-helicase-DNA-binding protein 8 isoform 2

- duplin
- helicase with SNF2 domain 1
- HELSNF1
- KIAA1564

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of CHD8 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57680\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57680[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CHD8%5BTIAB%5D%29+OR+%28chromodomain+helicase+DNA+binding+protein+8%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+1800+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- CHROMODOMAIN HELICASE DNA-BINDING PROTEIN 8; CHD8 (<https://omim.org/entry/610528>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/57680>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CHD8\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CHD8[gene]))

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## **Genomic Location**

The *CHD8* gene is found on chromosome 14 (<https://medlineplus.gov/genetics/chromosome/14/>).

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