

CNBP gene

CCHC-type zinc finger nucleic acid binding protein

Normal Function

The *CNBP* gene provides instructions for making a protein called CCHC-type zinc finger nucleic acid binding protein. This protein has seven regions, called zinc finger domains, which are thought to attach (bind) to specific sites on DNA and its chemical cousin, RNA.

The CNBP protein is found in many of the body's tissues, but it is most abundant in the heart and in muscles used for movement (skeletal muscles). The CNBP protein regulates the activity of other genes and is necessary for normal development before birth, particularly of muscles.

One region of the *CNBP* gene contains a segment of four DNA building blocks (nucleotides) that is repeated multiple times. This sequence, which is written as CCTG, is called a tetranucleotide repeat. In most people, the CCTG sequence is repeated fewer than 26 times.

Health Conditions Related to Genetic Changes

Myotonic dystrophy

Mutations in the *CNBP* gene cause a form of myotonic dystrophy known as myotonic dystrophy type 2. Myotonic dystrophy is characterized by progressive muscle wasting and weakness. Muscle weakness in type 2 primarily involves muscles close to the center of the body (proximal muscles), such as the those of the neck, shoulders, elbows, and hips. People with this disorder often have prolonged muscle contractions (myotonia) and are not able to relax certain muscles after use.

The type of gene mutation that causes myotonic dystrophy type 2 is known as a tetranucleotide repeat expansion. This mutation increases the size of the repeated CCTG segment in the *CNBP* gene. People with myotonic dystrophy type 2 have from 75 to more than 11,000 CCTG repeats.

The mutated *CNBP* gene produces an altered version of messenger RNA, which is a molecular blueprint of the gene that guides the production of proteins. Researchers have found that the altered messenger RNA traps proteins to form clumps within the cell. The clumps interfere with the production of many other proteins. These changes

prevent muscle cells and cells in other tissues from functioning properly, leading to muscle weakness and the other features of myotonic dystrophy type 2.

Other Names for This Gene

- CCHC-type zinc finger, nucleic acid binding protein
- cellular nucleic acid binding protein
- cellular retroviral nucleic acid-binding protein 1
- CNBP1
- CNBP_HUMAN
- DM2
- ZCCHC22
- zinc finger 9 protein
- zinc finger protein 273
- zinc finger protein 9
- zinc finger protein 9 (a cellular retroviral nucleic acid binding protein)
- ZNF9

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CNBP ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7555\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7555[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ZNF9%5BTIAB%5D%29+OR+%28zinc+finger+protein+9%5BTIAB%5D%29%29+OR+%28%28Cellular+nucleic+acid+binding+protein%5BTIAB%5D%29+OR+%28CNBP%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+720+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- CCHC-TYPE ZINC FINGER NUCLEIC ACID-BINDING PROTEIN; CNBP (<https://omim.org/entry/116955>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/7555>)

- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CNBP\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CNBP[gene]))

References

- Armas P, Aguero TH, Borgognone M, Aybar MJ, Calcaterra NB. Dissecting CNBP, a zinc-finger protein required for neural crest development, in its structural and functional domains. *J Mol Biol.* 2008 Oct 17;382(4):1043-56. doi:10.1016/j.jmb.2008.07.079. Epub 2008 Aug 5. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18703071>)
- Armas P, Nasif S, Calcaterra NB. Cellular nucleic acid binding protein binds G-rich single-stranded nucleic acids and may function as a nucleic acid chaperone. *J Cell Biochem.* 2008 Feb 15;103(3):1013-36. doi: 10.1002/jcb.21474. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17661353>)
- Bachinski LL, Czernuszewicz T, Ramagli LS, Suominen T, Shriver MD, Udd B, Siciliano MJ, Krahe R. Premutation allele pool in myotonic dystrophy type 2. *Neurology.* 2009 Feb 10;72(6):490-7. doi: 10.1212/01.wnl.0000333665.01888.33. Epub 2008 Nov 19. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19020295>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2677510/>)
- Botta A, Caldarola S, Vallo L, Bonifazi E, Fruci D, Gullotta F, Massa R, Novelli G, Loreni F. Effect of the [CCTG]_n repeat expansion on ZNF9 expression in myotonic dystrophy type II (DM2). *Biochim Biophys Acta.* 2006 Mar;1762(3):329-34. doi: 10.1016/j.bbdis.2005.11.004. Epub 2005 Dec 6. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16376058>)
- David AP, Pipier A, Pascutti F, Binolfi A, Weiner AMJ, Challier E, Heckel S, Calsou P, Gomez D, Calcaterra NB, Armas P. CNBP controls transcription by unfolding DNA G-quadruplex structures. *Nucleic Acids Res.* 2019 Sep 5;47(15):7901-7913. doi: 10.1093/nar/gkz527. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/31219592>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6735679/>)
- Kleefeld F, Erdmann H, Schoser B. Myotonic Dystrophy Type 2. 2006 Sep 21 [updated 2025 Sep 25]. In: Adam MP, Bick S, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, editors. *GeneReviews*(R) [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2026. Available from <http://www.ncbi.nlm.nih.gov/books/NBK1466/> Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20301639>)
- Liquori CL, Ricker K, Moseley ML, Jacobsen JF, Kress W, Naylor SL, Day JW, Ranum LP. Myotonic dystrophy type 2 caused by a CCTG expansion in intron 1 of ZNF9. *Science.* 2001 Aug 3;293(5531):864-7. doi: 10.1126/science.1062125. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11486088>)
- Wei C, Stock L, Schneider-Gold C, Sommer C, Timchenko NA, Timchenko L. Reduction of Cellular Nucleic Acid Binding Protein Encoded by a Myotonic Dystrophy Type 2 Gene Causes Muscle Atrophy. *Mol Cell Biol.* 2018 Jun 28;38(14):e00649-17. doi: 10.1128/MCB.00649-17. Print 2018 Jul 15. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/29735719>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6064917/>)

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Genomic Location

The *CNBP* gene is found on chromosome 3 (<https://medlineplus.gov/genetics/chromosome/3/>).

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