



CTDP1 gene

CTD phosphatase subunit 1

Normal Function

The *CTDP1* gene provides instructions for making a protein called carboxy-terminal domain phosphatase 1. This protein helps regulate the activity of an enzyme called RNA polymerase II. The RNA polymerase II enzyme initiates transcription, which is a key step in using the information carried by genes to direct the production (synthesis) of proteins.

Health Conditions Related to Genetic Changes

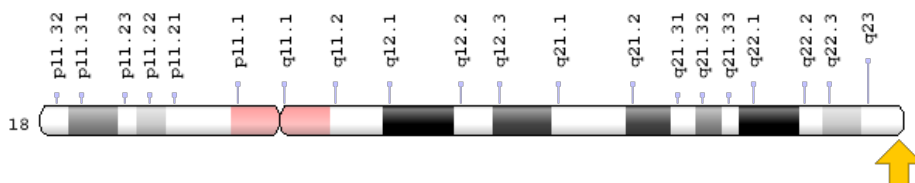
Congenital cataracts, facial dysmorphism, and neuropathy

Thus far, everyone with CCFDN has had the same mutation in both copies of the *CTDP1* gene in each cell. This mutation, written as IVS6+389C>T, alters the way the gene's instructions are pieced together to produce the carboxy-terminal domain phosphatase 1 protein. The altered instructions introduce a premature stop signal, resulting in an abnormally short, nonfunctional protein that cannot regulate transcription. Defective regulation of the transcription process affects the development and function of many parts of the body. It is not known how nonfunctional carboxy-terminal domain phosphatase 1 protein results in the specific signs and symptoms of CCFDN.

Chromosomal Location

Cytogenetic Location: 18q23, which is the long (q) arm of chromosome 18 at position 23

Molecular Location: base pairs 79,679,801 to 79,756,625 on chromosome 18 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CCFDN
- CTD (carboxy-terminal domain, RNA polymerase II, polypeptide A) phosphatase, subunit 1
- CTD of POLR2A, phosphatase of, subunit 1
- CTDP1_HUMAN
- FCP1
- RNA polymerase II subunit A C-terminal domain phosphatase
- RNA polymerase II subunit A C-terminal domain phosphatase isoform FCP1a
- RNA polymerase II subunit A C-terminal domain phosphatase isoform FCP1b
- serine phosphatase FCP1a
- TFIIIF-associating CTD phosphatase 1
- transcription factor IIF-associating CTD phosphatase 1

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): RNA Polymerase II Requires General Transcription Factors
<https://www.ncbi.nlm.nih.gov/books/NBK26887/#A999>

Clinical Information from GeneReviews

- Congenital Cataracts, Facial Dysmorphism, and Neuropathy
<https://www.ncbi.nlm.nih.gov/books/NBK25565>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28CTDP1%5BTIAB%5D%29+OR+%28%28FCP1%5BTIAB%5D%29+OR+%28CCFDN%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+2520+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- C-TERMINAL DOMAIN OF RNA POLYMERASE II SUBUNIT A, PHOSPHATASE OF, SUBUNIT 1
<http://omim.org/entry/604927>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CTDP1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CTDP1%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:2498
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:9150>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/9150>
- UniProt
<https://www.uniprot.org/uniprot/Q9Y5B0>

Sources for This Summary

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