

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.

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

Tests Listed in the Genetic Testing Registry

- Tests of CTD1P1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9150\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9150[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28CTDP1%5BTIAB%5D%29+OR+%28CCFDN%5BTIAB%5D%29%29+AND+%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%29>)

Catalog of Genes and Diseases from OMIM

- C-TERMINAL DOMAIN OF RNA POLYMERASE II SUBUNIT A, PHOSPHATASE OF, SUBUNIT 1; CTD1P1 (<https://omim.org/entry/604927>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/9150>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CTDP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CTDP1[gene]))

References

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Genomic Location

The *CTDP1* gene is found on chromosome 18 (<https://medlineplus.gov/genetics/chromosome/18/>).

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