



## POLR1D gene

RNA polymerase I and III subunit D

### Normal Function

The *POLR1D* gene provides instructions for making one part (subunit) of two related enzymes called RNA polymerase I and RNA polymerase III. These enzymes are involved in the production (synthesis) of ribonucleic acid (RNA), a chemical cousin of DNA. Both enzymes help synthesize a form of RNA known as ribosomal RNA (rRNA). RNA polymerase III also plays a role in the synthesis of several other forms of RNA, including transfer RNA (tRNA). Ribosomal RNA and transfer RNA assemble protein building blocks (amino acids) into functioning proteins, which is essential for the normal functioning and survival of cells.

Based on its involvement in Treacher Collins syndrome, the *POLR1D* gene appears to play a critical role in the early development of structures that become bones and other tissues of the face.

### Health Conditions Related to Genetic Changes

#### Treacher Collins syndrome

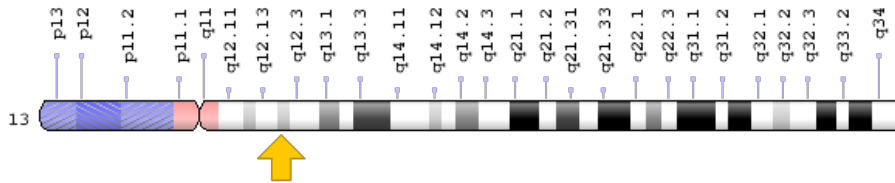
At least 20 mutations in the *POLR1D* gene have been identified in people with Treacher Collins syndrome, a condition that affects the development of bones and other tissues of the face. These mutations appear to alter the structure and function of the POLR1D protein, which reduces the amount of functional RNA polymerase I and RNA polymerase III in cells. Consequently, less rRNA is produced. Researchers speculate that a shortage of rRNA may trigger the self-destruction (apoptosis) of certain cells involved in the early development of facial bones and tissues. The abnormal cell death could underlie the specific problems with facial development found in Treacher Collins syndrome. However, it is unclear why the effects of a reduction in rRNA are limited to facial development.

#### Coloboma

## Chromosomal Location

Cytogenetic Location: 13q12.2, which is the long (q) arm of chromosome 13 at position 12.2

Molecular Location: base pairs 27,620,743 to 27,667,415 on chromosome 13 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- AC19
- DNA-directed RNA polymerase I subunit D
- DNA-directed RNA polymerases I and III subunit RPAC2
- FLJ20616
- MGC9850
- polymerase (RNA) I polypeptide D
- polymerase (RNA) I polypeptide D, 16kDa
- polymerase (RNA) I subunit D
- RNA polymerase I subunit D
- RNA polymerases I and III subunit AC2
- RPA9
- RPA16
- RPAC2
- RPAC2\_HUMAN
- RPC16
- RPO1-3

## **Additional Information & Resources**

### Educational Resources

- Biochemistry (fifth edition, 2002): RNA in Eukaryotic Cells Is Synthesized by Three Types of RNA Polymerase  
<https://www.ncbi.nlm.nih.gov/books/NBK22433/#A3981>
- Molecular Cell Biology (fourth edition, 2000): The Three Roles of RNA in Protein Synthesis  
<https://www.ncbi.nlm.nih.gov/books/NBK21603/>
- The Cell: A Molecular Approach (second edition, 2000): Eukaryotic RNA Polymerases  
<https://www.ncbi.nlm.nih.gov/books/NBK9935/#A981>

### Clinical Information from GeneReviews

- Treacher Collins Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1532>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28POLR1D%5BTIAB%5D%29+OR+%28%28AC19%5BTIAB%5D%29+OR+%28RPAC2%5BTIAB%5D%29+OR+%28TCS2%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>

### Catalog of Genes and Diseases from OMIM

- POLYMERASE I, RNA, SUBUNIT D  
<http://omim.org/entry/613715>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_POLR1D.html](http://atlasgeneticsoncology.org/Genes/GC_POLR1D.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=POLR1D%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:20422](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:20422)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:51082>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/51082>

- UniProt: RPAC2\_HUMAN  
<https://www.uniprot.org/uniprot/P0DPB6>
- UniProt: RPC22\_HUMAN  
<https://www.uniprot.org/uniprot/P0DPB5>

### Sources for This Summary

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- Fisher E. Exploring the genetic origins of Treacher Collins syndrome. *Clin Genet.* 2011 Apr;79(4):330-2. doi: 10.1111/j.1399-0004.2011.01632.x. Epub 2011 Feb 7.  
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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20301704>
- Yao Y, Yamamoto K, Nishi Y, Nogi Y, Muramatsu M. Mouse RNA polymerase I 16-kDa subunit able to associate with 40-kDa subunit is a homolog of yeast AC19 subunit of RNA polymerases I and III. *J Biol Chem.* 1996 Dec 20;271(51):32881-5.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/8955128>

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