



POLR1C gene

RNA polymerase I and III subunit C

Normal Function

The *POLR1C* 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 *POLR1C* 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

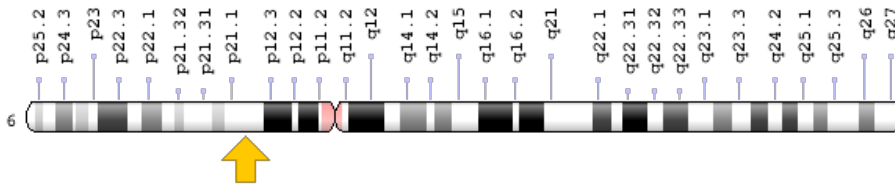
At least six mutations in the *POLR1C* gene have been identified in people with Treacher Collins syndrome, a condition that affects the development of bones and other tissues of the face. These mutations appear to alter the structure and function of the POLR1C protein, which reduces the amount of functional 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: 6p21.1, which is the short (p) arm of chromosome 6 at position 21.1

Molecular Location: base pairs 43,517,024 to 43,562,404 on chromosome 6 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AC40
- DNA-directed RNA polymerase I subunit C
- DNA-directed RNA polymerases I and III 40 kDa polypeptide
- DNA-directed RNA polymerases I and III subunit RPAC1
- polymerase (RNA) I polypeptide C
- polymerase (RNA) I polypeptide C, 30kDa
- polymerase (RNA) I subunit C
- RNA polymerase I subunit C
- RNA polymerases I and III subunit AC1
- RPA5
- RPA39
- RPA40
- RPAC1
- RPAC1_HUMAN
- RPC40

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=%28POLR1C%5BTIAB%5D%29+OR+%28%28RPA5%5BTIAB%5D%29+OR+%28TCS3%5BTIAB%5D%29+OR+%28RPA40%5BTIAB%5D%29+OR+%28RPAC1%5BTIAB%5D%29+OR+%28AC40%5BTIAB%5D%29+OR+%28RPC40%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 C
<http://omim.org/entry/610060>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=POLR1C%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:20194
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:9533>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/9533>
- UniProt
<https://www.uniprot.org/uniprot/O15160>

Sources for This Summary

- Dauwerse JG, Dixon J, Seland S, Ruivenkamp CA, van Haeringen A, Hoefsloot LH, Peters DJ, Boers AC, Daumer-Haas C, Maiwald R, Zweier C, Kerr B, Cobo AM, Toral JF, Hoogeboom AJ, Lohmann DR, Hehr U, Dixon MJ, Breuning MH, Wieczorek D. Mutations in genes encoding subunits of RNA polymerases I and III cause Treacher Collins syndrome. *Nat Genet.* 2011 Jan;43(1):20-2. doi: 10.1038/ng.724. Epub 2010 Dec 5.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21131976>
- 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.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21261602>
- Katsanis SH, Jabs EW. Treacher Collins Syndrome. 2004 Jul 20 [updated 2012 Aug 30]. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Ledbetter N, Mefford HC, Smith RJH, Stephens K, editors. *GeneReviews®* [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from <http://www.ncbi.nlm.nih.gov/books/NBK1532/>
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20301704>
- Mao M, Fu G, Wu JS, Zhang QH, Zhou J, Kan LX, Huang QH, He KL, Gu BW, Han ZG, Shen Y, Gu J, Yu YP, Xu SH, Wang YX, Chen SJ, Chen Z. Identification of genes expressed in human CD34(+) hematopoietic stem/progenitor cells by expressed sequence tags and efficient full-length cDNA cloning. *Proc Natl Acad Sci U S A.* 1998 Jul 7;95(14):8175-80.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/9653160>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC20949/>

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