RARA gene
retinoic acid receptor alpha

Normal Function

The RARA gene provides instructions for making a transcription factor called the retinoic acid receptor, alpha (RARα). A transcription factor is a protein that attaches (binds) to specific regions of DNA and helps control the activity of particular genes. The RARα protein controls the activity (transcription) of genes that are important for the maturation (differentiation) of immature white blood cells beyond a particular stage called the promyelocyte.

The RARα protein binds to specific regions of DNA and attracts other proteins that help block (repress) gene transcription, the first step in protein production. In response to a specific signal, the repressive proteins are removed and other proteins that induce gene transcription bind to the RARα protein, allowing gene transcription and cell differentiation.

Health Conditions Related to Genetic Changes

Acute promyelocytic leukemia

Gene mutations can be acquired during a person's lifetime and are present only in certain cells. These mutations are called somatic mutations, and they are not inherited. A somatic mutation involving the RARA gene causes acute promyelocytic leukemia, a cancer of the blood forming tissue (bone marrow). Acute promyelocytic leukemia is characterized by an accumulation of promyelocytes in the bone marrow. A rearrangement (translocation) of genetic material between chromosomes 15 and 17, written as t(15;17), fuses part of the RARA gene on chromosome 17 with part of another gene on chromosome 15 called PML. The protein produced from this fused gene, the PML-RARα protein, functions differently than the protein products of the normal PML and RARA genes.

The PML-RARα protein binds to DNA and represses gene transcription, like the normal RARα protein. However, the PML-RARα protein does not respond to the signal to induce transcription of genes, so the genes remain repressed.

Additionally, the function of the PML protein, the product of the PML gene, is disrupted. The PML protein blocks cell growth and division (proliferation) and induces self-destruction (apoptosis) in combination with other proteins. However, the PML-RARα protein does not block proliferation or induce apoptosis.
The PML-RAR\(\alpha\) protein blocks the differentiation of blood cells at the promyelocyte stage and allows abnormal cell proliferation. As a result, excess promyelocytes accumulate in the bone marrow and normal white blood cells cannot form, leading to acute promyelocytic leukemia.

**Chromosomal Location**

Cytogenetic Location: 17q21.2, which is the long (q) arm of chromosome 17 at position 21.2

Molecular Location: base pairs 40,309,180 to 40,357,643 on chromosome 17 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- NR1B1
- nuclear receptor subfamily 1 group B member 1
- RAR
- RAR-alpha
- retinoic acid receptor, alpha

**Additional Information & Resources**

**Educational Resources**

  https://www.ncbi.nlm.nih.gov/books/NBK12465/#A44441

**Scientific Articles on PubMed**

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RARA%5BTIAB%5D%29+OR+%28retinoic+acid+receptor+alpha%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

- RETINOIC ACID RECEPTOR, ALPHA
  http://omim.org/entry/180240

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/RARAID46.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RARA%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5914
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P10276

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12357341
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11257111
- OMIM: RETINOIC ACID RECEPTOR, ALPHA
  http://omim.org/entry/180240
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