

## **RARA gene**

retinoic acid receptor alpha

### **Normal Function**

The *RARA* gene provides instructions for making a transcription factor called the retinoic acid receptor, alpha (RAR $\alpha$ ). A transcription factor is a protein that attaches (binds) to specific regions of DNA and helps control the activity of particular genes. The RAR $\alpha$  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 RAR $\alpha$  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 RAR $\alpha$  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-RAR $\alpha$  protein, functions differently than the protein products of the normal *PML* and *RARA* genes.

The PML-RAR $\alpha$  protein binds to DNA and represses gene transcription, like the normal RAR $\alpha$  protein. However, the PML-RAR $\alpha$  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-RAR $\alpha$  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.

## Other Names for This Gene

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

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of RARA ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5914\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5914[geneid]))

## Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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; RARA (<https://omim.org/entry/180240>)

## Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5914>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RARA\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RARA[gene]))

## References

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## Genomic Location

The *RARA* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

**Last updated April 1, 2011**