

EPOR gene

erythropoietin receptor

Normal Function

The *EPOR* gene provides instructions for making a protein called the erythropoietin receptor. Erythropoietin is a hormone that directs the production of new red blood cells (erythrocytes) in the bone marrow. Red blood cells make up about half of total blood volume, and their primary function is to carry oxygen from the lungs to tissues and organs throughout the body. New red blood cells are constantly being produced by the body as worn-out red blood cells are broken down. To trigger the production of red blood cells, erythropoietin attaches (binds) to the erythropoietin receptor. This binding turns on (activates) the receptor, which stimulates several signaling pathways (particularly a cascade of signals known as the JAK/STAT pathway) that lead to the formation and maturation of red blood cells.

Health Conditions Related to Genetic Changes

Familial erythrocytosis

At least 16 mutations in the *EPOR* gene have been found to cause familial erythrocytosis, an inherited condition characterized by an increased number of red blood cells and an elevated risk of abnormal blood clots. When familial erythrocytosis results from *EPOR* gene mutations, it is often designated ECYT1.

Most of the identified mutations in the *EPOR* gene lead to the production of an abnormally short version of the erythropoietin receptor. A few mutations change single protein building blocks (amino acids) in the receptor. All of these mutations alter the structure of the receptor, causing it to remain activated for an abnormally long time after binding to erythropoietin. The overactive receptor signals the production of red blood cells even when no more are needed, which leads to an excess of these cells in the bloodstream.

Other Names for This Gene

- EPO-R
- EPOR_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of EPOR ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2057\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2057[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28EPOR%5BTI%5D%29+OR+%28erythropoietin+receptor%5BTI%5D%29+OR+%28EPO-R%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- ERYTHROPOIETIN RECEPTOR; EPOR (<https://omim.org/entry/133171>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2057>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=EPOR\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=EPOR[gene]))

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Genomic Location

The *EPOR* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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