

EPX gene

eosinophil peroxidase

Normal Function

The *EPX* gene provides instructions for making a protein called eosinophil peroxidase. This protein is found within certain white blood cells called eosinophils. During a normal immune response, eosinophils are activated (turned on), and they travel to the area of injury or inflammation. The cells then release proteins and other compounds that have a toxic effect on severely damaged cells or invading organisms. One of these proteins is called eosinophil peroxidase. This protein helps form molecules that are highly toxic to bacteria and parasites. These toxic molecules also play a role in regulating inflammation by fighting microbial invaders.

The eosinophil peroxidase protein is produced as a long strand that is cut (cleaved) into two smaller pieces. The shorter piece is known as the light chain and the longer piece is known as the heavy chain. These two pieces are attached to each other to form functional eosinophil peroxidase.

Health Conditions Related to Genetic Changes

Eosinophil peroxidase deficiency

At least three mutations have been found to cause eosinophil peroxidase deficiency. This condition affects eosinophils but causes no health problems in affected individuals. These mutations reduce or prevent eosinophil peroxidase production or result in a protein that is unstable and nonfunctional. As a result, eosinophils have severely reduced amounts of eosinophil peroxidase or none at all. The cells lacking eosinophil peroxidase are smaller and may have structural changes, but the other proteins within affected eosinophils are normal and the loss of eosinophil peroxidase does not appear to impair the function of these cells.

Other Names for This Gene

- eosinophil peroxidase preproprotein
- EPO
- EPP
- EPX-PEN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of EPX ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8288\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8288[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28EPX%5BTI%5D%29+OR+%28eosinophil+peroxidase%5BTI%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

- EOSINOPHIL PEROXIDASE; EPX (<https://omim.org/entry/131399>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8288>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=EPX\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=EPX[gene]))

References

- Acharya KR, Ackerman SJ. Eosinophil granule proteins: form and function. *JBiol Chem*. 2014 Jun 20;289(25):17406-15. doi: 10.1074/jbc.R113.546218. Epub 2014 May 6. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24802755>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4067173/>)
- Romano M, Baralle FE, Patriarca P. Expression and characterization of recombinant human eosinophil peroxidase. Impact of the R286H substitution on the biosynthesis and activity of the enzyme. *Eur J Biochem*. 2000 Jun;267(12):3704-11. doi: 10.1046/j.1432-1327.2000.01403.x. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/10848988>)
- Yamaguchi Y, Zhang DE, Sun Z, Albee EA, Nagata S, Tenen DG, Ackerman SJ. Functional characterization of the promoter for the gene encoding human eosinophil peroxidase. *J Biol Chem*. 1994 Jul 29;269(30):19410-9. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/8034708>)

Genomic Location

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

Last updated December 1, 2014