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.
Chromosomal Location

Cytogenetic Location: 17q22, which is the long (q) arm of chromosome 17 at position 22

Molecular Location: base pairs 58,192,726 to 58,205,174 on chromosome 17 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

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

Additional Information & Resources

Educational Resources

• Immunobiology: The Immune System in Health and Disease (fifth edition, 2001): Eosinophils are Normally Under Tight Control to Prevent Inappropriate Toxic Responses
  https://www.ncbi.nlm.nih.gov/books/NBK27112/#A1739

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EPX%5BTI%5D%29+OR+%28eosinophil+peroxidase%5BTI%5D%29%29+AND+%28%28Genes%5BMH%29+OR+%28Genetic+Phenomena%5BMH%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

• EOSINOPHIL PEROXIDASE
  http://omim.org/entry/131399
Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_EPX.html
- ClinVar
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:8288
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P11678

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/24802755
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4067173/
- OMIM: EOSINOPHIL PEROXIDASE
  http://omim.org/entry/131399
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10848988
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8034708

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