Eosinophil peroxidase deficiency

Description

Eosinophil peroxidase deficiency is a condition that affects certain white blood cells called eosinophils but causes no health problems in affected individuals. Eosinophils aid in the body's immune response. During a normal immune response, these cells are turned on (activated), 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. In eosinophil peroxidase deficiency, eosinophils have little or no eosinophil peroxidase. A lack of this protein does not seem to affect the eosinophils' ability to carry out an immune response.

Because eosinophil peroxidase deficiency does not cause any health problems, this condition is often diagnosed when blood tests are done for other reasons or when a family member has been diagnosed with the condition.

Frequency

Approximately 100 individuals with eosinophil peroxidase deficiency have been described in the scientific literature. Based on blood test data, varying estimates of the prevalence of the condition have been reported in specific populations. Eosinophil peroxidase deficiency is estimated to occur in 8.6 in 1,000 Yemenite Jews, in 3 in 1,000 North-African Jews, and in 1 in 1,000 Iraqi Jews. In northeastern Italy, the condition occurs in approximately 1 in 14,000 individuals; in Japan it occurs in 1 in 36,000 people; and in Luxembourg, eosinophil peroxidase deficiency is thought to occur in 1 in 100,000 people.

Causes

Mutations in the *EPX* gene cause eosinophil peroxidase deficiency. The *EPX* gene provides instructions for making the eosinophil peroxidase protein. During an immune response, activated eosinophils release eosinophil peroxidase at the site of injury. 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.

EPX gene 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. Other proteins within affected eosinophils are normal, and while the cells lacking eosinophil peroxidase are smaller and may have structural changes, the loss of eosinophil peroxidase does not appear to impair the function of eosinophils.

Learn more about the gene associated with Eosinophil peroxidase deficiency

- EPX

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- EPXD
- Peroxidase and phospholipid deficiency in eosinophils
- Presentey anomaly

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Eosinophil peroxidase deficiency (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1850000/)

Genetic and Rare Diseases Information Center

- Eosinophil peroxidase deficiency (https://rarediseases.info.nih.gov/diseases/12361/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- EOSINOPHIL PEROXIDASE DEFICIENCY; EPXD (https://omim.org/entry/261500)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28eosinophil+peroxidase+deficiency%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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