

What is **X-CGD?**

X-linked Chronic Granulomatous Disease

A rare and
life-threatening
inherited immune
disorder

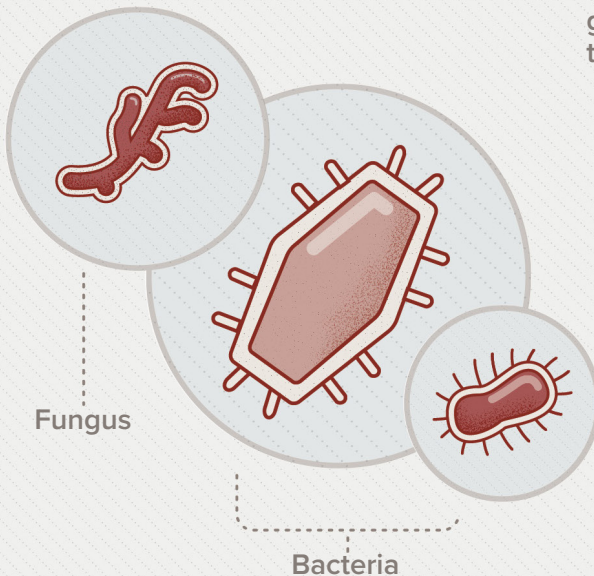


What are the causes of X-CGD?

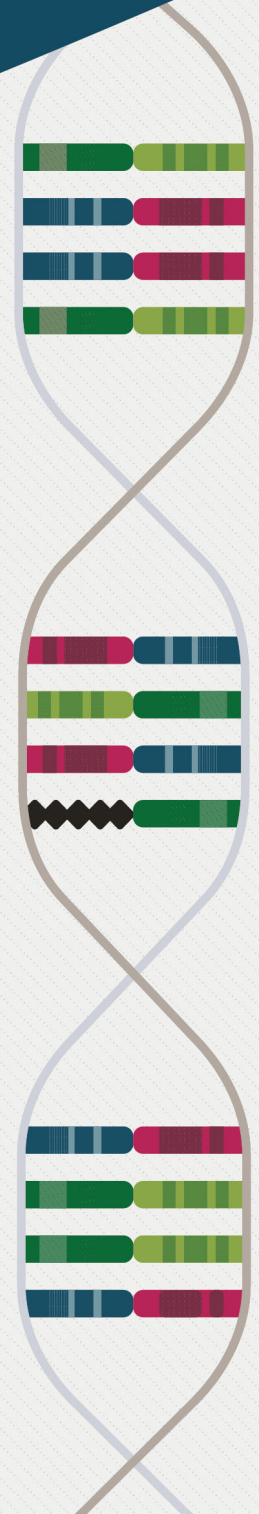
X-CGD is caused by a change or mutation in the CYBB gene, which provides instructions for making a protein that helps some immune system cells to function normally.

White blood cells called phagocytes target, eat and kill foreign invaders such as bacteria and fungi.

X-CGD prevents the production of a chemical needed to attack these invaders, leading to increased risk of bacterial and fungal infections.



Faulty gene in the DNA



How is X-CGD inherited?

Approximately
**between 1 in every
100,000-400,000**

**baby boys born
will have X-CGD**

X-linked recessive inheritance

X-CGD is inherited in an X-linked pattern, which means that X-CGD usually only affects boys.

Carriers of the mutation have sometimes been shown to have mild symptoms, similar to those of X-CGD.



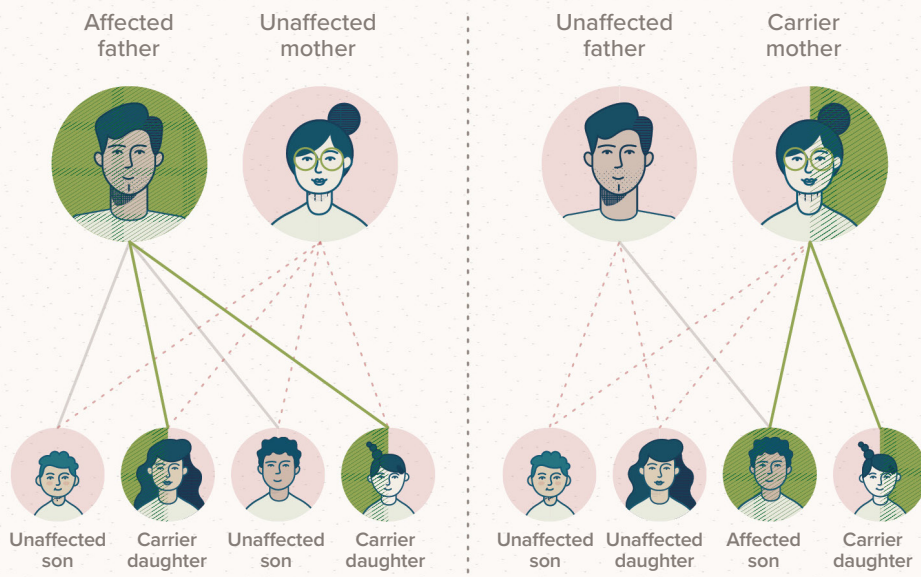
Affected



Unaffected



Carrier



How does X-CGD affect the body?

Boys with X-CGD are vulnerable to infection and excessive inflammation. Some may develop masses of immune cells (granulomas) that form at sites of infection or inflammation.

Symptoms may include:

Frequent & difficult to treat lung infections
(e.g. pneumonia)

Inflammatory bowel disease
(digestive problems, stomach pain, diarrhea)

Inflammation of genitourinary tract
(kidneys and bladder)

Skin infections
(e.g. skin abscesses)

Liver and/or brain abscesses

For more information about X-CGD and its management, please consult your healthcare provider.



Useful terms

Genes

Small sections of DNA that contain the instructions for individual characteristics, like eye and hair color, and how to make proteins, the functional building blocks of the cell. Proteins are responsible for making sure that the cells in the body function properly.

Mutation

A change in the structure of a gene or group of genes. Such changes can be passed on from parent to child. Many mutations cause no harm but others can cause genetic disorders, such as X-CGD.

CYBB gene

A small section of DNA that provides instructions to make a protein that helps some immune system cells to function normally.

Phagocytes

Cells that protect the body by ingesting harmful foreign invaders, like bacteria and fungi and dead or dying cells.

Immune disorder

A dysfunction of the immune system.

Immune system

Defends the body against foreign invaders, such as bacteria, viruses and fungi.

Granulomas

Masses of immune cells that form at sites of infection or inflammation.

X-linked recessive inheritance

A form of inheritance where the mutation, or disease, usually affects only boys.

Infection

The invasion and multiplication of micro-organisms, such as bacteria, viruses and fungi, which may cause symptoms or harmful effects within the body.

White blood cells (lymphocytes/leukocytes)

A type of blood cell that plays an important role in the immune system's response to infection. White blood cells are formed in the bone marrow.

References

- van den Berg et. al, PLoS One.2009;4(4):e5234
- Genetics Home Reference [Internet]. Bethesda (MD): The Library Published: June 23, 2020. Available from <https://ghr.nlm.nih.gov/primer/inheritance/inheritancepatterns>

