

What is

# ADA-SCID?

Adenosine deaminase severe  
combined immunodeficiency

A rare and  
life-threatening  
inherited immune  
disorder



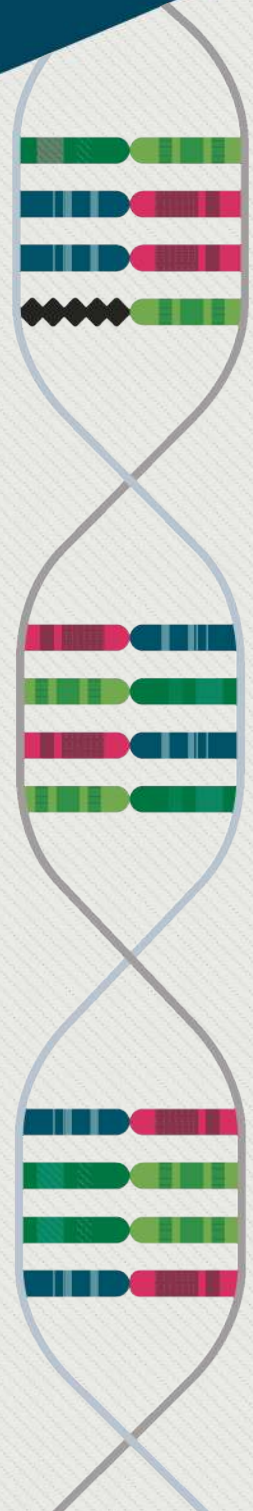
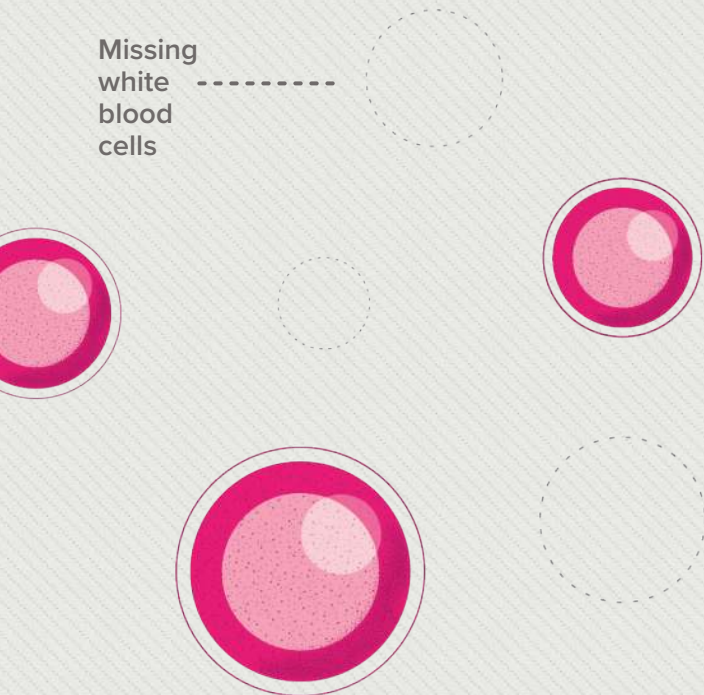
# What causes ADA-SCID?

Faulty gene  
in the DNA

ADA-SCID is caused by a change, or mutation in the *ADA* gene. This leads to absent or very low levels of the ADA enzyme such that the immune system is severely compromised or completely lacking.

When there is not enough of the ADA enzyme, the white blood cells responsible for fighting infections are missing, or do not work properly.

Missing  
white  
blood  
cells



# How is ADA-SCID inherited?

Approximately  
**1 in 500,000**  
babies born will  
have ADA-SCID

## Autosomal recessive inheritance

ADA-SCID is an autosomal recessive condition. This means that a child must inherit the mutated gene from both parents to have the condition.

ADA-SCID affects both boys and girls.



Affected



Unaffected



Carrier

Carrier father



Carrier mother



Affected child



Carrier child



Carrier child



Unaffected child

# How does ADA-SCID affect the body?

Children with ADA-SCID are at risk of serious, repeated infections. The ADA enzyme is found throughout the body and a shortage may cause other problems.

## Symptoms may include:

Developmental delay

Serious infections

Skeletal problems

Slower to grow and thrive

Skin inflammation

Frequent diarrhoea

Recurrent lung infections

Risk of deafness

For more information about ADA-SCID and its management, please consult your healthcare provider.



# Useful terms

## Adenosine deaminase (ADA)

An enzyme essential for the development and functioning of the immune system.

## 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 ADA-SCID.

## Genes

Small sections of DNA that contain the instructions for individual characteristics, like eye and hair colour, 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.

## Enzyme

A type of protein produced by the body's cells that increases the rate of chemical reactions, enabling the body to build up, or break down substances that are necessary for life and normal functioning.

## Immune disorder

A dysfunction of the immune system.

## Immune system

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

## 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

- [www.orpha.net](http://www.orpha.net): Serve combined immunodeficiency due to adenosine deaminase deficiency
- Whitmore KV, Gaspar HB. Front Immunol. 2016;7:314. Genetics Home Reference [Internet]. Bethesda (MD): The Library Published: April 19, 2021. Available from <https://ghr.nlm.nih.gov/primer/inheritance/inheritancepatterns>

