

What is

MPS?

Mucopolysaccharidoses

A group of rare,
life-limiting,
inherited genetic
diseases



What are the types & cause of MPS?

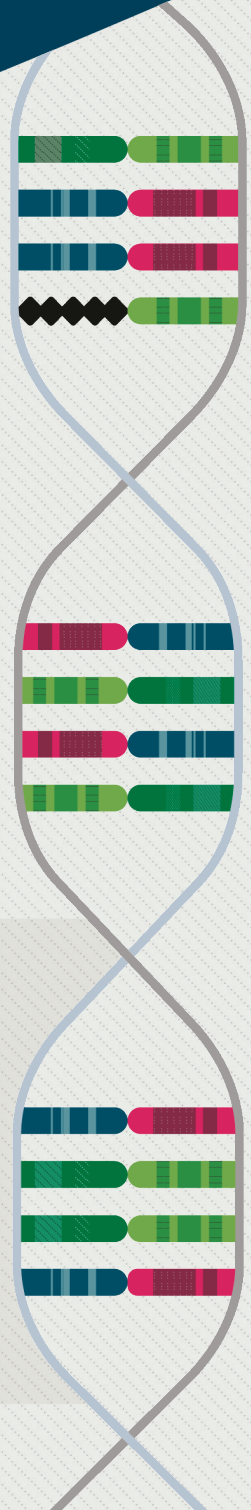
Faulty gene
in the DNA

There are different types and subtypes of mucopolysaccharidosis (MPS). Each type of MPS is caused by the absence or malfunction of a particular enzyme that the body needs to break down molecules called glycosaminoglycans (GAGs).

As a result, GAGs collect in the body's cells and tissues, leading to progressive cell damage and permanent effects on the body.

Types of MPS:

- MPS I** Hurler syndrome, Hurler-Scheie syndrome and Scheie syndrome
- MPS II** Hunter syndrome
- MPS III** Sanfilippo syndrome
- MPS IV** Morquio syndrome
- MPS VI** Maroteaux-Lamy syndrome
- MPS VII** Sly syndrome
- MPS IX** Hyaluronidase deficiency



How is MPS inherited?*

Approximately
**1 in every
25,000**

babies born will have
some form of MPS

Autosomal recessive inheritance

MPS is an autosomal recessive condition.

This means that a child must inherit the mutated gene from both parents to have the condition.

MPS affects both boys and girls*.



Affected

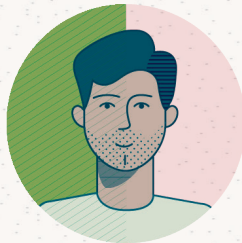


Unaffected



Carrier

Carrier
father



Carrier
mother



Affected
child



Carrier
child



Carrier
child



Unaffected
child

* All forms of MPS are autosomal recessive except MPS II, otherwise known as Hunter syndrome, which is an X-linked recessive disorder and affects boys only.

How does MPS affect the body?

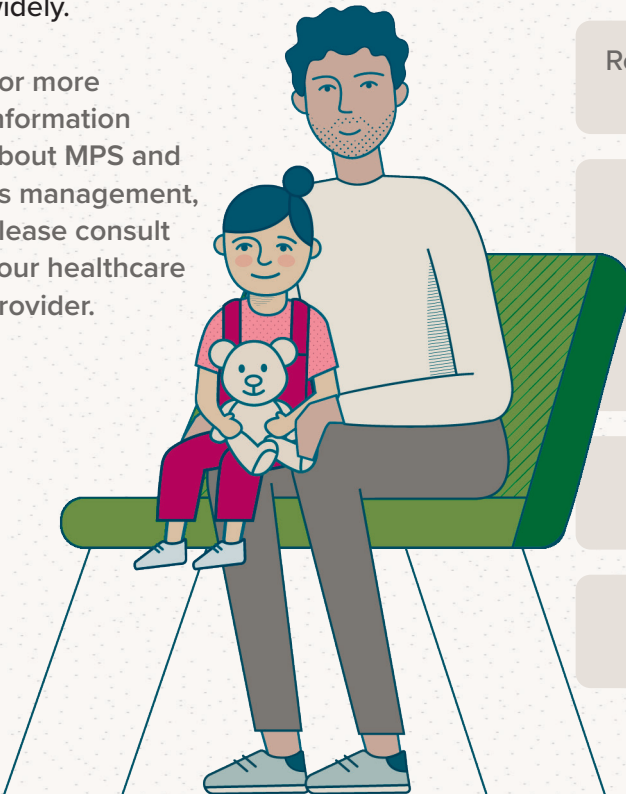
Symptoms may include:



Many symptoms are common among the different forms of MPS, but have different patterns and levels of severity depending on the form of the disease.

Usually, MPS symptoms are not present at birth. Most children experience a period of normal development followed by a decline in physical and/or cognitive function. The age at which symptoms begin can vary widely.

For more information about MPS and its management, please consult your healthcare provider.



Bone and joint problems

Distinct facial features

Short stature

Recurrent respiratory infections

Intellectual disability

Behaviour problems

Hearing and/or vision loss

Heart problems

Useful terms

Intellectual disability

A disability characterized by limitations in cognitive functioning and skills such as communication and ability to care for oneself.

X-linked recessive inheritance

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

Cognitive function

Each cognitive function works to help gather and process information, and they often work together in interrelated processes. These include attention, memory, language, perception, decision making and problem solving.

Glycosaminoglycans (GAGs)

Long chains of sugars (polysaccharides) in human cells that facilitate the development of bone, skin and connective tissue and are broken down by enzymes.

Gene

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.

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 MPS.

References

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