

What is MPS?

Mucopolysaccharidoses

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



What are the types & cause of MPS?

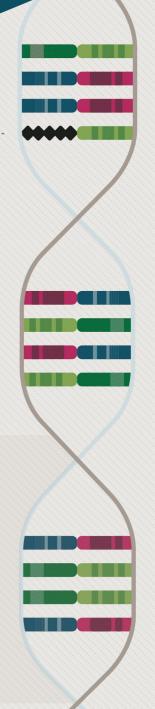
Faulty gene in the DNA

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

MPSI	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?*

1 in every 25,000

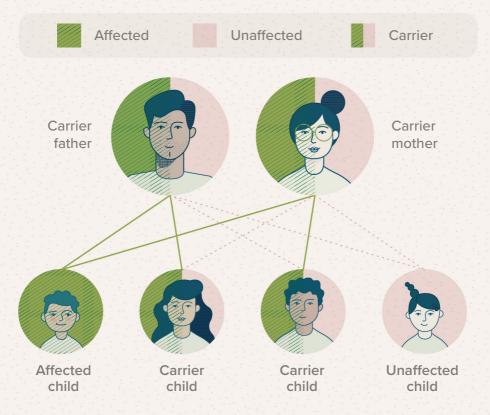
babies born will have some form of MPS

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*.



*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?

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

provider.

its management, please consult your healthcare Symptoms may include:

Bone and joint problems

Distinct facial features

Short stature

Recurrent respiratory infections

Intellectual disability

Behavior 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 color, and how to make roteins, 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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orchard-tx.com MPS-GLB-001 September 2020