Mucopolysaccharidoses

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Mucopolysaccharidoses (MPS) refers to a group of inherited metabolic diseases which is caused by a specific missing or faulty (not properly working) enzyme.

Without the enzyme, the break down process to turn sugar chains into proteins and simpler molecules is incomplete. This enzyme and break down process is needed to help build bones, tendons, skin, cartilage, corneas and connective tissue.

Due to the incomplete break down process, people with this disease are permanently affected with progressive cellular damage, which affects their appearance, organ and system function, physical abilities and cognitive development (in most cases).

Frequency

Research has estimated that 1 in 22,465 Australian births result in a form of mucopolysaccharidoses.

Signs and Symptoms

- Varies by type (see below)

Treatment

- There is no cure for the forms of mucopolysaccharidoses at this point in time.

- Medical care for mucopolysaccharidoses is aimed at treating systemic conditions and improving the quality of life for people with the disorder.

Types specific to section 24 of the NDIS Act

MPS 1-H (Hurler Syndrome)

Mucopolysaccharidoses type 1-H, also known as Hurler Syndrome, generally becomes apparent within the first year of life.

Children with this type of MPS experience developmental delay, which is primarily seen through delayed speech and progressive cognitive and sensorial decline between the ages of 12 to 24 months.

Signs and symptoms include:

- Abnormal vertebral morphology
- Abnormal heart valve morphology
- Abnormality of the tonsils
- Cardiomyopathy (heart disease)
- Wide nasal bridge
- Full cheeks

MPS III (San Fillipo Syndrome)

Mucopolysaccharidoses type 3 (MPS III) primarily affects the spinal cord and the brain, causing progressive intellectual disability and developmental decline.

There are generally no signs or symptoms that someone has the disorder at birth, with them instead beginning to show during early childhood.

Children with MPS III may be restless and have sleeping issues, as well as showing features of autism spectrum disorder. As children get older and the disorder progresses, seizures and other movement disorders may develop.

Signs and symptoms include:

- Delayed development (slowing of learning ability)
- Deteriorating mental status
- Course facial features
- Behavioral problems
- Sleeping difficulties

Contact IDEAS

Free call: 1800 029 904

Text: 0458 296 602

Email: info@ideas.org.au

Web: www.ideas.org.au

Disclaimer

This fact sheet provides general information about the disability and is for informational purposes only. It is not a guarantee that you will meet the disability requirements in section 24 of the NDIS Act.

References

https://www.mpssociety.org.au/

https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Fact-Sheets/Mucopolysaccharidoses-Fact-Sheet#3195_1

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