

What is mucopolysaccharidosis type II?

Mucopolysaccharidosis type II is an inherited metabolic disease that affects many organs and parts of the body. Individuals with mucopolysaccharidosis type II have defects in the enzyme iduronate 2-sulfatase, which breaks down large sugar molecules known as glycosaminoglycans or mucopolysaccharides.² The symptoms of mucopolysaccharidosis type II are due to the build-up of the large sugar molecules heparan sulfate and dermatan sulfate within lysosomes in cells.² Mucopolysaccharidosis type II belongs to a group of diseases called lysosomal storage disorders and is also known as Hunter syndrome.¹

What are the symptoms of mucopolysaccharidosis type II and what treatment is available?

Symptoms of mucopolysaccharidosis type II vary in age at onset and severity. Affected individuals are almost always male and typically do not show symptoms at birth.³ Signs and symptoms are progressive and may include:³

- Distinctive facial features
- Umbilical or groin hernias
- Recurrent infections
- Hepatosplenomegaly (enlarged liver and spleen)
- Macrocephaly (large head) and hydrocephalus (fluid build-up in the brain)
- Enlarged vocal cords and tongue and hoarse voice
- Narrowing of the airway, causing respiratory disease and sleep apnea
- Hearing loss and reduced vision
- Intellectual decline and developmental regression
- Skeletal abnormalities and short stature
- Heart valve disease and ventricular hypertrophy (enlarged heart)

Symptoms of the severe form of the disease typically appear at 2 to 4 years of age and life expectancy may be up to 10 to 20 years.³ Symptoms of milder forms may progress more slowly and may not include neurological involvement.³ Life expectancy may be into adulthood.³

There is no cure for mucopolysaccharidosis type II. Treatment may include enzyme replacement therapy to improve some of the physical symptoms, supportive care, and surgical intervention as needed.³

Mucopolysaccharidosis type II is included in newborn screening panels in a few states in the United States.⁴

How is mucopolysaccharidosis type II inherited?

Mucopolysaccharidosis type II is an X-linked recessive disease caused by mutations in the *IDS* gene.¹ A male who inherits one copy of an *IDS* gene mutation is affected with mucopolysaccharidosis type II. A female who inherits one copy of an *IDS* gene mutation is a carrier and is not expected to have related health problems, but in some cases may have symptoms. A female who inherits two *IDS* mutations, one from each parent, is affected with mucopolysaccharidosis type II, although this is an uncommon occurrence.

If a female is a carrier, the risk for each son to be affected is 50% and the risk for each daughter to be a carrier is 50%. If a male is affected, each son is unaffected and each daughter is an obligate carrier.

Who is at risk for mucopolysaccharidosis type II?

Mucopolysaccharidosis type II can occur in individuals of all races and ethnicities with an estimated incidence of 1 in 100,000 male live births.^{5,6}

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-ii>

National NPS Society: <http://mpssociety.org/mps/mps-ii/>

National Organization for Rare Disorders (NORD): <http://rarediseases.org/rare-diseases/hunter-syndrome/>

References

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