

What is sialidosis?

Sialidosis is an inherited metabolic disease with variable severity and age at onset. Sialidosis types I and II are recognized based on clinical features, severity, and age of onset. Individuals with sialidosis type I or type II have a deficiency in the neuraminidase 1 enzyme, which helps break down large sugar molecules.² Signs and symptoms of sialidosis are due to the buildup of molecules in the lysosomes.² Sialidosis is also known as mucopolipidosis type I and belongs to a group of diseases called lysosomal storage disorders.³

What are the symptoms of sialidosis and what treatment is available?

Individuals with sialidosis type I typically start showing signs and symptoms in their teens and twenties. Life expectancy and intelligence are not affected. Symptoms may include:^{1,3}

- Progressive problems with walking
- Vision problems
- Characteristic eye abnormality called a cherry-red spot
- Progressive myoclonus (muscle twitches)
- Ataxia (loss of muscle coordination)
- Seizures
- Hyperreflexia (over-responsive reflexes)

Individuals with sialidosis type II have more severe forms of the disease.¹ Signs and symptoms may be evident prior to birth, usually leading to death before or soon after birth, or evident by early childhood with life expectancy varying depending on the severity of symptoms.¹ Symptoms may include:^{1,3}

- Fetal hydrops (accumulation of fluid)
- Hepatosplenomegaly (enlarged liver and spleen)
- Bone abnormalities
- Distinctive facial features
- Short stature
- Intellectual disability and developmental delay
- Symptoms seen in type I sialidosis
- Hearing loss
- Tooth and gum abnormalities
- Angiokeratomas (benign skin lesions)
- Heart abnormalities

Treatment is supportive and focuses on prevention of complications and management of symptoms.³

How is sialidosis inherited?

Sialidosis is an autosomal recessive disease caused by mutations in the *NEU1* gene.¹ An individual who inherits one copy of a *NEU1* gene mutation is a carrier and are not expected to have related health problems. An individual who inherits two *NEU1* mutations, one from each parent, is expected to be affected with sialidosis.

If both members of a couple are carriers of mutations in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for sialidosis?

Sialidosis is a rare condition that can occur in individuals of all races and ethnicities. Its worldwide prevalence is unknown.¹

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/sialidosis>

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

The International Advocate for Glycoprotein Storage Diseases (ISM RD):
http://www.ismrd.org/glycoprotein_diseases/sialidosis

References

1. Sialidosis. *Genetics Home Reference*. Available at: <http://ghr.nlm.nih.gov/condition/sialidosis>. Accessed February 18, 2016.
2. *Neu1*. *Genetics Home Reference*. Available at: <http://ghr.nlm.nih.gov/gene/NEU1>. Accessed February 18, 2016.
3. Sialidosis. *National Organization for Rare Disorders*. Available at: <http://rarediseases.org/rare-diseases/sialidosis/>. Accessed February 18, 2016.