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Salla disease and other sialic acid storage diseases

What are sialic acid storage diseases?

Sialic acid storage diseases, including Salla disease, are inherited disorders characterized by progressive damage to the nervous system, poor growth, and seizures. Individuals with sialic storage diseases have a defect in a protein called sialin, which is needed to move free sialic acid within cells. Symptoms are due to the toxic build-up of sialic acid in the cells, particularly in the nervous system. Salla disease is the mildest form of the sialic acid storage diseases. There is a moderate form called intermediate severe Salla disease and a severe form known as infantile free sialic acid-storage disease.[1]

What are the symptoms of Salla disease and other sialic acid storage diseases and what treatment is available?

Individuals with Salla disease typically appear normal at birth. Symptoms usually begin by six months of age and include:[1,2]

- Hypotonia (low muscle tone)
- Ataxia (difficulty coordinating movements), typically by one year of age
- Poor growth in early childhood
- Slow, progressive intellectual and motor skills delays
- Seizures
- Spasticity (abnormally tight muscles)

The other forms of sialic acid storage disease have more severe symptoms, with some similarities to Salla disease. There is no cure for any of the sialic acid storage diseases. Treatment includes supportive care for symptoms. Salla disease is associated with survival into adulthood.[2]

How are sialic acid storage diseases inherited?

Sialic acid storage diseases are a group of autosomal recessive diseases caused by mutations in the *SLC17A5* gene.[1] An individual who inherits one *SLC17A5* gene mutation is a carrier of a sialic acid storage disorder and is not expected to have related health problems. An individual who inherits two *SLC17A5* gene mutations, one from each parent, is expected to be affected with a sialic acid storage disease, such as Salla disease.

If both members of a couple are carriers of a mutation in the same gene, the risk of having 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 Salla disease?

Salla disease is a rare disorder most frequently reported in individuals of Finnish ancestry, but occurring in other populations as well. The estimated frequency of carriers of Salla disease in Finland is 1 in 200.[3]

Having a relative who is a carrier or is affected can increase an individual's risk of being 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?

Genetic and Rare Diseases Information Center (GARD): https://rarediseases.info.nih.gov/gard/4754/salla-disease/resources/1

National Organization for Rare Disorders (NORD): http://rarediseases.org/rare-diseases/lysosomal-free-sialic-acid-storage-disorders/

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/sialic-acid-storage-disease

References

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2. Adams D, Gahl WA. Free Sialic Acid Storage Disorders. *GeneReviews*. Available at: http://www.ncbi.nlm.nih.gov/books/NBK1470/ Accessed: March 21, 2012.
3. Aula N *et al*. The Spectrum of *SLC17A5*-Gene Mutations Resulting in Free Sialic-Acid Storage Diseases Indicates Some Genotype-Phenotype Correlation. *Am J Hum Genet* 2000; 67:832-840.