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What is Salla disease?
Salla disease (SD) is an inherited disorder characterized by progressive damage to the nervous system, poor growth, and seizures. It involves defects in a protein called sialin, which is needed to move a substance called 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 in a group of diseases called sialic acid storage disorders. There is a moderate form called intermediate severe Salla disease and a severe form known as infantile free sialic acid-storage disease (ISSD).[1]

What are the symptoms of Salla disease 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 form of the sialic acid storage disorders. Treatment for Salla disease includes supportive care for symptoms and is associated with survival into adulthood.[2]

How is Salla disease inherited?
Sialic acid storage disorders 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 copies of the c.115C>T (p.R39C) mutation, one from each parent, is expected to be affected with Salla disease. Other mutation combinations in the *SLC17A5* gene may result in intermediate severe Salla disease or ISSD.[2]

If both members of a couple are carriers, 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 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 carrier frequency 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.

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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): http://rarediseases.info.nih.gov/
- Lysosomal Learning: An online resource on lysosomal storage disorders: www.lysosomallearning.com

References

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