What is multiple sulphatase deficiency?

Multiple sulphatase deficiency is a rare metabolic disease characterized by the defective activity of all known sulphatases. Individuals with multiple sulphatase deficiency have a reduction in the formylglycine-generating enzyme, which modifies sulphatases that help the body break down various hormones, fats, and sugars. Clinical features are caused by a buildup of sulfate-containing molecules in cells and may include ichthyosis, skeletal anomalies, distinctive facial features, neurologic deterioration, developmental delay, deafness, and hepatosplenomegaly. Age of onset ranges from neonatal to late childhood and survival is typically a few years after the onset of symptoms.

What are the symptoms of multiple sulphatase deficiency and what treatment is available?

Symptoms vary in severity and may include:

- Leukodystrophy (degeneration of white matter of the brain), which may lead to seizures and developmental delay
- Distinctive facial features
- Hypertrichosis (excessive body hair)
- Ichthyosis (dry, scaly skin)
- Skeletal abnormalities
- Deafness
- Heart malformations
- Hepatosplenomegaly (enlarged liver and spleen)

Life expectancy is shortened. Treatment is supportive and focuses on managing the symptoms.

How is multiple sulphatase deficiency inherited?

Multiple sulphatase deficiency is an autosomal recessive disease caused by mutations in the SUMF1 gene. An individual who inherits one copy of a SUMF1 gene mutation is a carrier. Carriers are not affected with multiple sulphatase deficiency. An individual who inherits two SUMF1 mutations, one from each parent, is expected to be affected with multiple sulphatase deficiency.

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 multiple sulphatase deficiency?

Multiple sulphatase deficiency is a rare condition that can occur in individuals of all races and ethnicities.

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?


National Organization for Rare Disorders (NORD): http://rarediseases.org/rare-diseases/multiple-sulfatase-deficiency/

Children Living with Inherited Metabolic Diseases (CLIMB): http://www.climb.org.uk/

References