Multiple Sulfatase Deficiency

What Your Results Mean

Test results indicate that you are a carrier of multiple sulfatase deficiency (MSD). Carriers typically show no symptoms. Risk for current or future pregnancies is dependent on your partner's carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner/donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner/donor are carriers for multiple sulfatase deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

Multiple Sulfatase Deficiency Explained

What is Multiple Sulfatase Deficiency?

Multiple sulfatase deficiency (MSD) is an inherited lysosomal storage disorder. The disorder prevents the cells in the body from properly processing a group of enzymes called sulfatases, which help to break down substances containing sulfates, including a variety of sugars, fats, and hormones. There are three major types of MSD, categorized by age of onset: neonatal, late-infantile, and juvenile. The neonatal form is most severe. Symptoms appear soon after birth and include seizures, developmental delay, slow growth, movement problems, ichthyosis (dry, scaly skin), hypertrichosis (excess hair growth), skeletal problems (including scoliosis and joint stiffness), and coarse facial features. Affected children can also have hearing loss, an enlarged liver and spleen, and heart abnormalities.

The late-infantile type is the most common form of the condition and shares some of the features seen in the neonatal type, including ichthyosis, skeletal problems, and coarse facial features. Children with the late-infantile type of MSD have normal development in early childhood but then progressively lose their cognitive and movement skills due to brain abnormalities. The juvenile type of MSD is rare, and onset of symptoms is in mid- to late-childhood. These children are typically normal at birth and in early childhood and then experience a loss of motor and cognitive skills; however, this loss of skills is typically slower. They also experience ichthyosis.

Prognosis

The prognosis for MSD is very poor. Children rarely live beyond age 10 and some die at younger ages.

Treatment

There is no curative treatment for MSD at this time. Physiotherapy and hydrotherapy can be useful to help individuals with MSD.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/condition/multiplesulfatasedeficiency

National Society of Genetic Counselors
https://www.nsgc.org/

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

Revised: 4/2022