Genetics Uncoded:

Facts about

Hurler Syndrome (Mucopolysaccharidosis Type 1)

What Your Test Results Mean

Carriers typically show no symptoms of Hurler syndrome; however, carriers are at an increased risk of having a child with Hurler syndrome. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Hurler Syndrome Explained

Hurler syndrome is an inherited lysosomal storage disorder in which glycosaminoglycans (sugar molecules) accumulate in the body and can damage organs. Individuals with Hurler syndrome do not make enough of the lysosomal enzyme alpha-L-iduronidase. Without enough of this enzyme, the body cannot properly break down glycosaminoglycans. The build-up of glycosaminoglycans leads to multisystem organ damage.

Individuals with Hurler syndrome may appear normal at birth but typically develop the symptoms within the first two years of life. Cloudy corneas can develop, leading to vision issues and even blindness, heart valve issues, and narrowed arteries may occur. Lung disease with frequent infections can develop, and decreased brain development can also occur. Life expectancy is typically ten years of age but with treatment may be increased.

Available treatment includes enzyme replacement therapy — the incorporation of the missing enzyme into the body through the veins. Incorporation of this enzyme may reduce the excess glycosaminoglycans and lead to functional improvements; however, the enzyme does not cross the blood-brain barrier and the neurological complications of the disorder does not typically improve with enzyme replacement therapy. Bone marrow transplant is possible and can help expand the lifespan of the affected individual.

How the Genetics Work

Hurler syndrome is an autosomal recessive disorder caused by pathogenic variants in the *IDUA* gene. In general, individuals have two copies of the *IDUA* gene. Carriers of Hurler syndrome have a single variant in one copy of the *IDUA* gene while individuals with Hurler syndrome have variants in both copies their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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