NxGen MDx

Genetics Uncoded:

Facts about Aspartylglucosaminuria

What Your Test Results Mean

Carriers typically show no symptoms of aspartylglucosaminuria. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Aspartylglucosaminuria Explained

Aspartylglucosaminuria is an inherited lysosomal storage disorder caused by a deficiency of the enzyme aspartyl glucosidase. Aspartyl glucosidase is found in lysosomes, the recycling compartments in the cells of the body. Without this enzyme, carbohydrates accumulate and cause cell death and tissue damage. Aspartylglucosaminuria causes progressive decline in mental functioning, coarse facial features, and skeletal abnormalities. Bone marrow transplant has been attempted with mixed results and enzyme replacement therapy is not yet clinically available.

How the Genetics Work

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

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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