Aspartylglucosaminuria

What Your Results Mean

Test results indicate that you are a carrier of aspartylglucosaminuria. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of aspartylglucosaminuria for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a 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 or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for aspartylglucosaminuria, each of your children has a 1 in 4 (25%) chance to have the condition.

Aspartylglucosaminuria Explained

What is Aspartylglucosaminuria?

Aspartylglucosaminuria is an inherited disorder where individuals experience a decline in mental functioning over time. Infants appear healthy at birth, with signs and symptoms showing up within the first 2-3 years of life. Additional signs and symptoms include coarse facial features, behavioral problems, an intellectual disability, and skeletal abnormalities. Adults with aspartylglucosaminuria can also experience seizures and difficulty with movement.

Prognosis

Prognosis is considered unfavorable as the decline in mental functioning is progressive and individuals typically live until mid-adulthood.

Treatment

Treatment for aspartylglucosaminuria is symptomatic; there is currently no cure. Bone marrow transplant has been attempted with mixed results and enzyme replacement therapy is not yet clinically available.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/achromatopsia
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/aspartylglycosaminuria/
National Society of Genetic Counselors
https://www.nsgc.org/

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