# Argininosuccinic Aciduria

NxGen MDx

## What Your Results Mean

Test results indicate that you are a carrier of argininosuccinic aciduria (ASA). Carriers are not expected to show symptoms. You and your partner would both have to be carriers of ASA 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 ASA, each of your children has a 1 in 4 (25%) chance to have the condition.

# Argininosuccinic Aciduria Explained

## What is Argininosuccinic Aciduria?

Argininosuccinic aciduria is an inherited condition caused by a urea cycle defect that causes ammonia to accumulate in the blood. Symptoms typically present in infancy and may include lethargy or unwillingness to eat and a poorly controlled breathing rate or body temperature. Some babies may experience seizures, unusual body movements, and may go into a coma. Intellectual and developmental delays may also occur.

## Prognosis

Prognosis is typically poor. Early diagnosis and treatment are associated with best prognosis; however, severity of the disease is variable, mortality is high, and neurological damage is frequent.

## Treatment

Long-term treatment includes dietary restriction of protein and supplementation of arginine. For individuals who are non-responsive to dietary changes, oral nitrogen scavenging therapy can be considered. Orthotopic liver transplantation is considered only in patients with severe cases of this disease.

### Resources

**Genetics Home Reference**
https://ghr.nlm.nih.gov/condition/argininosuccinic-aciduria
**Urea Cycle Disorders Consortium**
https://www.rarediseasesnetwork.org/cms/ucdc/Learn-More/Disorder-Definitions
**National Organization for Rare Disorders**
https://rarediseases.org/rare-diseases/argininosuccinic-aciduria/
**National Society of Genetic Counselors**
https://www.nsgc.org/

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