Inheritest® CARRIER SCREEN

Arthrogryposis, mental retardation, and seizures (AMRS)

What is AMRS?

AMRS is an inherited disease characterized by arthrogryposis, intellectual and behavioral disabilities, and epilepsy.[1,2] In individuals with AMRS, there appears to be in a change in the surface of cells because a reduced amount of a nucleotide sugar (uridine diphosphate N-acetylglucosamine) is transported to the cells. The signs and symptoms of AMRS may be the result of changes in the way that cells grow and differentiate.[3]

What are the symptoms of AMRS and what treatment is available?

AMRS is a disease with variable severity and age of onset. Symptoms may include:[1,2]

- Arthrogryposis (joint contractures)
- Intellectual disability
- Autism spectrum disorder
- Epilepsy (seizures)
- Limb malformations
- Distal joint problems
- Microcephaly (small head)
- Retro/micrognathia (small chin)
- Hypotonia (low muscle tone)

There is no cure for AMRS. Treatment is supportive.

How is AMRS inherited?

AMRS is an autosomal recessive disease caused by mutations in the *SLC35A3* gene.[1,2,3] An individual who inherits one copy of a *SLC35A3* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two *SLC35A3* mutations, one from each parent, is expected to be affected with AMRS.

If both members of a couple are carriers of mutations in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for AMRS?

AMRS is rare. It has been reported in the Ashkenazi Jewish population.[3]

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Genetics Home Reference: http://ghr.nlm.nih.gov/gene/SLC35A3

References

1. SLC35A3. *Genetics Home Reference*. Available at http://ghr.nlm.nih.gov/gene/SLC35A3. Accessed February 4, 2016
2. Online Mendelian Inheritance in Man, OMIM®. Johns Hopkins University, Baltimore, MD. MIM Number: [61553]. Available at http://www.omim.org/entry/615553. Accessed February 4, 2016
3. Edvardson, S. *et al*.. Mutations in SLC35A3 cause autism spectrum disorder, epilepsy and arthrogryposis. J. *Med. Genet.* 2013;50:733-739.