Genetics Uncoded:

Facts about

Arthrogryposis, Mental Retardation, and Seizures

What Your Test Results Mean

Carriers typically show no symptoms of arthrogryposis, mental retardation, and seizures (AMRS). Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Arthrogryposis, Mental Retardation, and Seizures Explained

AMRS is an inherited condition that is caused by abnormalities in a glycan transporter. Carbohydrate chains (glycans) are normally attached to the outside of the cell membrane so that cells can communicate with each other and tissues can be properly developed and maintained. These carbohydrate structures cannot be moved to the cell membrane without the transporter, leading to serious health problems for individuals with AMRS. The major symptoms of AMRS are described in its name—affected individuals suffer from arthrogryposis (immobility of the joints due to muscle fibrosis), mental retardation, and seizures. In addition, individuals with AMRS usually have small head size, poor muscle tone, and autism. There is no cure for AMRS.

How the Genetics Work

AMRS is an autosomal recessive disorder that can be explained by variants in the *SLC35A3* gene. In general, individuals have two copies of the *SLC35A3* gene. Carriers of AMRS have a single variant in one copy of the *SLC35A3* gene while individuals with AMRS 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.

NxGenMDx, LLC
801 Broadway Ave NW, Suite 203
Grand Rapids, MI, 49504

Revised 12/15/16