Genetics Uncoded:

Clarity Genetics

Facts about Muscle-Eye-Brain Disease

What Your Test Results Mean

Carriers typically show no symptoms of Muscle-Eye-Brain Disease (MEB); however, carriers are at an increased risk of having a child with MEB. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Muscle-Eye-Brain Disease Explained

Muscle-eye-brain disease is a type of congenital muscular dystrophy characterized by brain malformations, eye abnormalities, muscle weakness, and developmental delay. Individuals with MEB are not able to make the proper chemical modifications to an enzyme called alpha-dystroglycan, which is essential for the development of muscle fibers and neural cells. Without the chemical modifications, alpha-dystroglycan cannot function properly, leading to the symptoms of the disease. There have been mild forms reported in recent years with onset in late childhood and minimal brain involvement. Treatment of individuals with MEB typically includes supportive care.

How the Genetics Work

Muscle-Eye-Brain Disease is an autosomal recessive muscular dystrophy caused by variants in the *POMGNT1* gene. In general, individuals have two copies of the *POMGNT1* gene. Carriers of MEB have a single variant in one copy of the *POMGNT1* gene while individuals with *POMGNT1*-related MEB 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 **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

Clarity Genetics, LLC
5231 NW 33rd Ave
Ft. Lauderdale, FL 33309
Revised 12/15/16