



Genetics Uncoded:

Facts about

Inclusion Body Myopathy 2



What Your Test Results Mean

Carriers typically show no symptoms of inclusion body myopathy 2; however, carriers are at an increased risk of having a child with inclusion body myopathy 2. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

● Inclusion Body Myopathy 2 Explained

Inclusion body myopathy 2 is an inherited condition that slows the body's production of sialic acid, a sugar molecule that allows cells in a tissue to signal and adhere to one another. This disease causes progressive weakening of the skeletal muscles, usually beginning in an affected individual's late teens or early twenties. About 20 years after symptoms appear, the disease will have progressed to the point that the person can no longer walk and is confined to a wheelchair. Inclusion body myopathy 2 does not weaken the eye and heart muscles and does not cause neurological symptoms. There is no cure for this disease, but physical and occupational therapy can help individuals retain strength and mobility for as long as possible.

● How the Genetics Work

Inclusion body myopathy 2 is caused by pathogenic variants in the *GNE* gene. In general, individuals have two copies of the *GNE* gene. Carriers of inclusion body myopathy 2 have a single variant in one copy of the *GNE* gene while individuals with inclusion body myopathy 2 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.