Clarity Genetics

Genetics Uncoded:

Facts about

Niemann-Pick Disease Type C

What Your Test Results Mean

Carriers typically show no symptoms of Niemann-Pick disease type C (NPC); however, carriers are at an increased risk of having a child with NPC. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Niemann-Pick Disease Type C Explained

NPC is an inherited lysosomal storage disorder that impairs fat metabolism causing harmful amounts of lipids to accumulate in the spleen, liver, lungs, bone marrow, and brain. NPC usually appears in childhood, although infant and adult onsets are possible. Signs of NPC include severe liver disease, breathing difficulties, developmental delay, seizures, poor muscle tone, lack of coordination, problems with feeding, and an inability to move the eyes vertically. People with this disorder can survive about 10-20 years after diagnosis. There is no cure for NPC; however, seizure medications and physical and speech therapy may be recommended.

How the Genetics Work

NPC is an autosomal recessive disorder caused by pathogenic variants in the *NPC1* gene. In general, individuals have two copies of the *NPC1* gene. Carriers of NPC have a single variant in one copy of the *NPC1* gene while individuals with NPC 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