Understanding Your Positive Neuronal Ceroid Lipofuscinosis (NCL)/Batten Disease Genetic Test Result

INFORMATION FOR PATIENTS WITH TWO PATHOGENIC MUTATIONS OR TWO VARIANTS THAT ARE LIKELY PATHOGENIC

RESULT | MUTATION
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The testing done for you/your family member shows that you have a combination of two pathogenic or likely pathogenic (disease-causing) mutations in a gene (or genes) that cause NCL. Both of these should be treated as the same type of positive result.

GENE | DEFINITION
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Genes are instructions for how our bodies work and develop. Everyone has two copies of each gene, one from each parent. Mutations (changes in the gene, like spelling mistakes) in certain genes can cause NCL. Most types of NCL are caused by having two mutations in the same gene, but some types can be caused by having only one mutation. Even if there is no history of NCL in your family, it can still be caused by changes in a gene.

DIAGNOSIS | NCL
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This result means that you/your family member has a type of NCL. There are many types of NCL and most cause progressive problems with vision, movement, and thinking ability. NCL can begin during infancy, childhood, or adulthood depending on the type of NCL you/your family member has. Talk to your healthcare provider about you/your family member’s specific diagnosis.

MANAGEMENT OPTIONS | FOR PATIENTS WITH NCL
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Some NCL genetic test results can help identify specific treatments that are more or less likely to be helpful. Treatment options can include: medications and therapies to either help manage the symptoms of NCL or to treat the underlying cause. Knowing the genetic cause of you/your family member’s symptoms may help to avoid some tests or procedures. Talk to your doctor about which may be right for you/your family member.

SCREENING OPTIONS | FAMILY MEMBERS
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Family members of a person with two gene mutations in a NCL gene may have one or more mutation in the same gene. When a person has one mutation, they are a “carrier” and could have a child with NCL if their partner is also a carrier. Your adult family members may wish to be tested to see if they carry the mutation(s) found in your family.

NEXT STEPS | DISCUSS
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It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers. Talk to your doctor or genetic counselor about who in your family might benefit from considering this testing.

REACH OUT | RESOURCES
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• Ambry’s Neurology Site for Families patients.ambrygen.com/neurology
• Batten Disease Support and Research Association bdsra.org
• Beyond Batten Disease Foundation beyondbatten.org
• National Society of Genetic Counselors nsgc.org
• Canadian Association of Genetic Counsellors cagc-accg.ca

PARTICIPATE | RESEARCH OPPORTUNITIES
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Institute for Basic Research in Developmental Disabilities (IBR) Batten Disease Center opwdd.ny.gov/institute-for-basic-research/jervis-clinic/batten
The Batten Disease Registry housed at the IBR collects data on all NCL cases in the U.S. and some foreign countries. Participating in research is voluntary.

HOW NCL IS INHERITED
People who carry two mutations in an NCL gene have NCL. Most often, they inherited one of these mutations from their mother and one from their father. People who have a mutation in a NCL gene are “carriers” and do not usually have symptoms of NCL. Family members of a person who has two mutations in a NCL gene could also have a mutation in the same gene.

Please discuss this information with your healthcare provider. The field of genetics is continuously changing, so updates related to your genetic testing result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or taken as medical advice.