

## Understanding Your Negative Neurodevelopmental Disorder Genetic Test Result

INFORMATION FOR PATIENTS

| Result                | NEGATIVE                 | The testing done for you/your family member did not find any pathogenic mutations (disease-causing changes in the gene, like a spelling mistake) that cause a neurodevelopmental disorder (NDD), including epilepsy, intellectual disability (ID), or autism spectrum disorders (ASDs). You/your family member may have a mutation in a gene that was not included in this test or you/your family member's NDD may not be caused by a gene change. |
|-----------------------|--------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Gene                  | DEFINITION               | Genes are instructions for how our bodies work and develop. Everyone has two copies of each gene, one from each parent. Mutations in certain genes can cause NDDs. These mutations may be passed down in families or be brand new for the person with NDD. Even if there is no one else with an ASD in your family, it can still be caused by a change in a gene.                                                                                   |
| Diagnosis             | NO CHANGE                | This testing does not change your/your family member's diagnosis of a NDD. If you or your family member has been diagnosed with a NDD, that remains the same.                                                                                                                                                                                                                                                                                       |
| Further<br>Testing    | DISCUSS                  | More genetic testing may be needed for you or your family member. Please talk about this with your healthcare provider.                                                                                                                                                                                                                                                                                                                             |
| Management<br>Options | FOR PEOPLE<br>WITH A NDD | Management options vary by condition and other factors. Talk to your healthcare provider about which may be right for you/your family member.                                                                                                                                                                                                                                                                                                       |
| Next Steps            | DISCUSS                  | Even though no gene mutations were found from testing, there may still be a gene change causing the NDD in you/your family member. Please share this information with family members so they can discuss with their healthcare provider and learn more.                                                                                                                                                                                             |
| Reach Out             | RESOURCES                | Ambry's Neurology Site for Families ambrygen.com/patients/neurology American Epilepsy Society aesnet.org Autism Speaks autismspeaks.org The Arc thearc.org Child Neurology Foundation childneurologyfoundation.org National Society of Genetic Counselors nsgc.org Canadian Association of Genetic Counsellors cagc-accg.ca                                                                                                                         |

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 interpreted as medical advice.