ARUP LABORATORIES

A nonprofit enterprise of the University of Utah and its Department of Pathology

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EARLY-ONSET ALZHEIMER'S DISEASE (AD) TESTING INFORMED CONSENT

Patient Name: _____ **Date of Birth:** _____

Sex Assigned at Birth: ☐Female ☐Male ☐Intersex **Gender Identity (optional):** ☐Female ☐Male ☐_____

Symptoms: ☐ No ☐ Yes **If yes, describe:** _____ **Age of onset:** _____

Is there a family history? ☐ No ☐ Yes

If yes, describe relative's relationship to patient: _____ **Age of onset:** _____

Was DNA testing performed on relative? ☐ No ☐ Yes **If yes, describe variant identified:** _____

- Participation in genetic testing is completely voluntary.
- The ordering healthcare provider or genetic counselor should provide pretest and posttest genetic counseling (including a discussion of results) in person. At risk/ presymptomatic individuals should be accompanied by a support person, who is not at risk for early-onset AD.
- See nsgc.org to find a genetic counselor near you.
- AD is characterized by progressive memory loss leading to dementia; there is currently no cure or effective treatment. Up to 25% of individuals with AD have a hereditary form. Less than 2% have the early-onset form defined by an AD diagnosis at younger than age 65.
- This test only detects the early-onset familial form of AD. The chance of identifying a disease-causing variant increases with younger age of onset. Testing involves sequencing the DNA of the *APP*, *PSEN1*, and *PSEN2* genes from a blood sample. Disease-causing DNA variants are only identified in 60–80% of individuals with early-onset AD. In 20–40% of affected individuals, a causative variant cannot be identified. The analytical accuracy of the DNA test result is 99%. Possible sources of error include sample mislabeling, contamination, transfusion, or bone marrow transplantation.
- If a disease-causing variant is identified, insurance rates, ability to obtain disability and life insurance, and employability could be affected. The Genetic Information Nondiscrimination Act extends some protections against genetic discrimination (genome.gov/10002328). Test results are released to the ordering provider and other parties legally entitled to them.
- Test results may reveal nonpaternity or predict that other family members are affected or at risk for developing early-onset AD.

There are three possible test results:

1. Negative: No disease-causing variants were identified. This result reduces, but does not exclude, the risk for early-onset AD.
2. Uncertain: A variant of uncertain significance was identified. There may or may not be an increased risk for early-onset AD. More information is needed to determine if the variant is harmful or not.
3. Positive: A disease-causing variant was identified and there is an increased risk for early-onset AD. Offspring have a 50% risk for inheriting a genetic predisposition for early onset AD.

- There are psychological risks associated with early-onset AD testing. A negative result can produce feelings of joy and guilt. An uncertain result can be frustrating. A positive result could cause severe stress, depression, or feelings of futility.
- ARUP is not a storage facility. Most samples are discarded after testing. Some samples are stored indefinitely for test validation or education purposes after personal identifiers are removed. New York samples are discarded after testing. You may request sample disposal by calling ARUP Laboratories at 800-242-2787 ext. 3301.
- In cooperation with the National Institutes of Health's effort to improve understanding of specific genetic variants, ARUP submits HIPAA-compliant, deidentified (cannot be traced back to the patient) genetic test results and health information to public databases. The confidentiality of each sample is maintained. If you prefer that your test result not be shared, call ARUP at 800-242-2787 ext. 3301. Your deidentified information will not be disclosed to public databases after your request is received, but a separate request is required for each genetic test. Additionally, patients have the opportunity to participate in patient registries and research. To learn more, visit ARUP's Genetics Resources website at aruplab.com/genetics/resources.

Patient, Legal Guardian, Power of Attorney (POA): I am the patient, their legal guardian, or POA. I have been counseled regarding the risks, benefits, and limitations of testing for early onset AD and carefully considered the psychological impact the results may have on the patient and their family.

_____	_____	_____
Patient/Guardian Printed Name	Signature	Date

Ordering Provider/Genetic Counselor: I have explained the risks, benefits, and alternatives of this genetic test to the patient/guardian.

_____	_____	_____
Provider/Genetic Counselor Printed Name	Signature	Date