ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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

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

(Required for testing of presymptomatic individuals)

Patient Name: _____ **Date of Birth:** _____ **Sex**: ☐ Female ☐ Male

- Participation in genetic testing is completely voluntary. Genetic counseling is highly recommended prior to and following testing for early-onset AD. See www.nsgc.org to find a genetic counselor in your area. The ordering health care provider or genetic counselor should explain results in person and be available for follow-up counseling. Individuals undergoing presymptomatic testing should be accompanied by a support person, who is not at risk for early-onset AD, for pre-test and post-test counseling.
- 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. Of those, <2% have the early-onset form defined by an AD diagnosis before age 65.
- This test will only detect the early-onset familial form of AD. The chance of identifying a disease-causing variant increases when disease onset in the family is earlier. Testing involves extracting DNA from a blood sample and sequencing the *APP*, *PSEN1*, and *PSEN2* genes. 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 accuracy of the DNA test result is 99%. Possible sources of error include sample mislabeling or contamination, transfusion, or bone marrow transplantation.
- Test results may reveal non-paternity or predict that other family members may be affected with, or at risk for developing early-onset AD.

There are three possible test results:

1. Negative: No pathogenic 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 causative or benign.
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 test result, indicating the patient may or may not develop symptoms, can be frustrating. A positive result could lead to serious psychological consequences including severe stress, depression, or feelings of futility.
- If a disease-causing variant is identified for early-onset AD, insurance rates, the ability to obtain disability and life insurance, and employability could be affected. The Genetic Information Nondiscrimination Act of 2008 extends some protections against genetic discrimination (http://www.genome.gov/10002328). All test results are released to the ordering health care provider and those parties entitled to them by federal, state, and local laws.
- Because ARUP is not a storage facility, most samples are discarded after testing is completed. Some samples may be stored indefinitely for test validation or education purposes after personal identifiers are removed. All New York samples are discarded 60 days following test completion. You may request disposal of your sample by calling ARUP Laboratories at 800-242-2787 ext. 3301.

Patient, Legal Guardian, Power of Attorney (POA): I have the legal authority to request ARUP Laboratories to test this sample for early-onset AD. I am the patient, his/her legal guardian, or POA. I have been counseled regarding the risks, benefits and limitations of this test and carefully considered the psychological impact the results may have on the patient and his/her family.

_____	_____	_____
Patient/Guardian Printed Name	Signature	Date

Ordering Healthcare Provider, Genetic Counselor: I have explained this genetic test and its risks, benefits and alternatives to the patient or legal guardian and addressed all their questions.

_____	_____	_____
Provider/Genetic Counselor Printed Name	Signature	Date