

Genetic Testing

Researchers have observed that having a parent or sibling with Alzheimer's disease does increase one's risk somewhat above the general population's risk of developing the disease, but such a family history should not cause undue anxiety. Nonetheless, some people with such family histories, and some without such histories, wish to have a genetic test that will answer the question: Will I be next?

Genetic risk factors for Alzheimer's

Alzheimer's disease is the object of intense genetic analysis. Researchers have identified four mutations, or variant forms, of genes associated with the disease. Three of those genes — located on chromosomes 21, 14, and 1 — are linked to the early-onset forms of Alzheimer's in which symptoms usually begin to appear between a person's early 40s and mid-50s. If someone has one of these gene mutations, he or she will at some point develop the disease. These incidents of Alzheimer's are very rare, possibly accounting for fewer than one percent of all cases.

Affected families are usually well aware of their unique history with the disease. For members of these families, a genetic test could indicate whether an individual carries the gene mutation and will eventually develop the disease. Many people in these families do not wish to know their genetic status, but some do get tested.

The fourth gene, APOE-e4 on chromosome 19, is linked to a greater risk of susceptibility for developing late-onset Alzheimer's, the more common form of the disease that is manifested after the age of 55 and generally associated with old age. APOE-e4 is a variant form of a gene that encodes the production of a protein called apolipoprotein E, which may play a role in repairing connections between brain cells. People with one copy of APOE-e4 have a greater risk of getting Alzheimer's than people with other forms of the gene, and people with two copies of APOE-e4 have an even greater risk.

Factors to weigh before considering genetic testing

Genetic testing for APOE-e4 is controversial and should only be undertaken after discussing the benefits and risks with a physician or genetic counselor. APOE-e4 increases the risk of developing Alzheimer's, but it is neither necessary (people without APOE-e4 develop the disease) nor sufficient (not all people with APOE-e4 develop Alzheimer's).

Although there is no way to change one's APOE-e4 status, lifestyle modifications may help reduce the potential effects of having APOE-e4. These lifestyle modifications include eating a heart-healthy diet, exercising and staying mentally active.

The risks of APOE-e4 genetic testing include anxiety if the test results are positive. However, recent large scale studies showed that anxiety is manageable with appropriate genetic counseling.

Before undergoing APOE-e4 testing, one should consider the possible effects of a positive test result on one's eligibility for disability, long-term care and life insurance. The Genetic Information Nondiscrimination Act of 2008 does not apply to these forms of insurance.

Association positions on genetic testing

The Alzheimer's Association position on genetic testing includes five principles intended to prevent genetic discrimination. These would apply to current tests for early-onset genes and to reliable tests that may eventually be developed to predict late-onset Alzheimer's.

1. Having the APOE-e4 gene does not mean a person has or will develop Alzheimer's disease.
2. The presence of a gene associated with Alzheimer's disease should not be used to deny access to housing, employment, health care, insurance or any other goods and services.
3. The presence of a gene associated with Alzheimer's disease does not qualify an individual for disability-related benefits. Disability support should be based on functional criteria rather than a genetic test.
4. Because of possible social consequences or discrimination, anonymous testing should be available, thereby making the fact of and results of genetic testing for Alzheimer's disease invisible on an individual's medical records.
5. If performed, genetic testing for Alzheimer's should be done with pre- and post-test counseling, which includes a full discussion of the implication of the test and provides the individual with all information necessary to make an informed decision. All genetic counseling and information should be provided in culturally and linguistically appropriate formats and should take into account an individual's literacy level.

The Alzheimer's Association is the leading voluntary health organization in Alzheimer care, support and research.

Updated November 2008