



Genetic Testing

COMMON QUESTIONS

- Is there a genetic test that accurately predicts Alzheimer's disease?
 - If a test were available, under what conditions should it be administered?
 - What are the potential risks associated with genetic testing for Alzheimer's disease?
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BACKGROUND INFORMATION

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?

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 five 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-ε4 on chromosome 19, is linked to a greater risk or 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-ε4 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-ε4 have a greater risk of getting Alzheimer's than people with other forms of the gene, and people with two copies of APOE-ε4 have an even greater risk. However, having one or two copies of the APOE-ε4 does not mean a person will necessarily develop Alzheimer's.

Consequently, the gene has no reliable value for predicting Alzheimer's, and it is medically useless information with regard to treatment. Tests for the gene, at present, would be of no use to an individual and only invite problems. Having the gene could cause unnecessary anxiety about getting the disease, and the information could result in discrimination in obtaining disability or long-term care insurance. Presently, genetic tests to determine susceptibility to Alzheimer's would only be of value in a research setting that is investigating the role of genes in the onset and progression of the disease.

ASSOCIATION POSITIONS

The Alzheimer's Association position on genetic testing includes six 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 a gene associated with Alzheimer's disease does not mean a person has the disease.
2. The presence of a gene is not a basis for underwriting insurance premiums for health care, long-term care, or life insurance.
3. The presence of a gene associated with Alzheimer's disease should not be used to deny access to housing, employment, health care, or any other goods and services.
4. The presence of a gene associated with Alz-

heimer's disease does not qualify an individual for disability-related benefits. Disability support should be based on functional criteria rather than on a genetic test.

5. 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.
6. 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.

To receive additional Association materials on this topic, log onto the Association's Web site (<http://www.alz.org>) or call (800) 272-3900.