



Genetic Testing

A position paper – 05/01

A. Executive Summary

1. The present paper constitutes the input of Alzheimer Europe and its member organisations to the ongoing discussions within Europe about genetic testing (in the context of Alzheimer's disease and other forms of dementia).
2. Alzheimer Europe would like to recall some general principles which guide this present response:
 - a) Having a gene associated with Alzheimer's disease or another form of dementia does not mean that a person has the disease.
 - b) People who have a gene linked to Alzheimer's disease or another form of dementia have the same rights as anyone else.
 - c) Genetic testing does not only affect the person taking the test. It may also reveal information about other relatives who might not want to know.
 - d) No genetic test is 100% accurate.
 - e) The extent to which health cover is provided to citizens by the State social security system and/or privately contracted by individuals differs from one country to the next.
3. On the basis of these principles, Alzheimer Europe has developed the following position with regard to genetic testing:
 - a) Alzheimer Europe firmly believes that the use and/or possession of genetic information by insurance companies should be prohibited.
 - b) Alzheimer Europe strongly supports research into the genetic factors linked to dementia which might further our understanding of the cause and development of the disease and possibly contribute to future treatment.
 - c) Based on its current information, Alzheimer Europe does not encourage the use of any genetic test for dementia UNLESS such test has a high and proven success rate either in assessing the risk of developing the disease (or not as the case may be) or in detecting the existence of it in a particular individual.
 - d) Alzheimer Europe requests further information on the accuracy, reliability and predictive value of any genetic tests for dementia.
 - e) Genetic testing should always be accompanied by adequate pre- and post-test counselling.
 - f) Anonymous testing should be possible so that individuals can ensure that such information does not remain in their medical files against their will.

B. Introduction

4. It is extremely important for people with dementia to be diagnosed as soon as possible. In the case of Alzheimer's disease, an early diagnosis may enable the person concerned to benefit from medication, which treats the global symptoms of the disease and is most

effective in the early to mid stages of the disease. Most forms of dementia involve the gradual deterioration of mental faculties (e.g. memory, language and thinking etc.) but in the early stages, it is still possible for the person affected to make decisions concerning his/her finances and care etc. – hence the importance of an early diagnosis.

5. If it were possible to detect dementia before the first symptoms became obvious, this would give people a greater opportunity to make informed decisions about their future lives. This is one of the potential benefits of genetic testing.
6. On the other hand, such information could clearly be used in ways which would be contrary to their personal interests, perhaps resulting in employment discrimination, loss of opportunities, stigmatisation, increased health insurance costs or even loss of health insurance to name but a few examples.
7. The present discussion paper outlines some of the recommendations of Alzheimer Europe and its member organisations and raises a few points which deserve further clarification and discussion.

C. The necessity for a response by Alzheimer Europe

8. In the last few years, the issue of genetic testing has been increasingly debated. In certain European countries there are already companies offering such tests. Unfortunately, the general public do not always fully understand what the results of such tests imply and there are no regulations governing how they are carried out i.e. what kind of information people receive, how the results are presented, whether there is any kind of counselling afterwards and the issue of confidentiality etc.
9. In order to provide information to people with dementia and other people interesting in knowing about their own state of health and in order to protect them from the unscrupulous use of the results of genetic tests, Alzheimer Europe has developed the present Position Paper.

D. General principles

10. At its Annual General Meeting in Munich on 15 October 2000, Alzheimer Europe adopted recommendations on how to improve the legal rights and protection of adults with incapacity due to dementia. This included a section on bioethical issues. These recommendations obviously need to guide any response of the organisation regarding genetic testing for people who suspect or fear they may have dementia and also those who have taken the test and did develop dementia.
 - a) The adult with incapacity has the right to be informed about his/her state of health.
 - b) Information should, where appropriate, cover the following: the diagnosis, the person's general state of health, treatment possibilities, potential risks and consequences of having or not having a particular treatment, side-effects, prognosis and alternative treatments.
 - c) Such information should not be withheld solely on the grounds that the adult is suffering from dementia and/or has communication difficulties. Attempts should be made to provide information in such a way as to maximise his/her ability to understand, making use of technology and other available techniques to enhance communication. Attention should be paid to any possible difficulty understanding, retaining information and communicating, as well as his/her level of education, reasoning capacity and cultural background. Care should be taken to avoid causing unnecessary anxiety and suffering.
 - d) Written as well as verbal information should always be provided as a back-up. The adult should be granted access to his/her medical file(s). S/he should also have the opportunity to discuss the contents of the medical file(s) with a person of his/her

choice (e.g. a doctor) and/or to appoint someone to receive information on his/her behalf.

- e) Information should not be given against the will of the adult with incapacity.
- f) The confidentiality of information should extend beyond the lifetime of the adult with incapacity. If any information is used for research or statistical purposes, the identity of the adult with incapacity should remain anonymous and the information should not be traceable back to him/her (in accordance with the provisions of national laws on respect for the confidentiality of personal information). Consideration should be given to access to information where abuse is suspected.
- g) A clear refusal by the adult with incapacity to grant access to information to any third party should be respected regardless of the extent of his/her incapacity, unless this would be clearly against his/her best interests e.g. carers should have provided to them information on a need to know basis to enable them to care effectively for the adult with incapacity.
- h) People who receive information about an adult with incapacity in connection with their work (either voluntary or paid) should be obliged to treat such information with confidentiality.

E. Genetic testing

11. These general principles as well as the Convention of Human Rights and Biomedicine (see Annex 1) and the Universal Declaration on the Human Genome and Human Rights (see Annex 2) dictate Alzheimer Europe's position with regard to genetic testing.
12. Alzheimer Europe would like to draw a distinction between tests which detect existing Alzheimer's disease and tests which assess the risk of developing dementia Alzheimer's disease at some time in the future:
 - a) **Diagnostic testing:** Familial early onset Alzheimer's disease (FAD) is associated with 3 genes. These are the amyloid precursor protein (APP), presenilin-1 and presenilin-2. These genetic mutations can be detected by genetic testing. However, it is important to note that the test only relates to those people with FAD (i.e. about 1% of all people with Alzheimer's disease). In the extremely limited number of families with this dominant genetic disorder, family members inherit from one of their parents the part of the DNA (the genetic make-up), which causes the disease. On average, half the children of an affected parent will develop the disease. For those who do, the age of onset tends to be relatively low, usually between 35 and 60.
 - b) **Assessment for risk testing:** Whether or not members of one's family have Alzheimer's disease, everyone risks developing the disease at some time. However, it is now known that there is a gene, which can affect this risk. This gene is found on chromosome 19 and it is responsible for the production of a protein called apolipoprotein E (ApoE). There are three main types of this protein, one of which (ApoE4), although uncommon, makes it more likely that Alzheimer's disease will occur. However, it does not cause the disease, but merely increases the likelihood. For example, a person of 50, would have a 2 in 1,000 chance of developing Alzheimer's disease instead of the usual 1 in 1,000, but might never actually develop it. Only 50% of people with Alzheimer's disease have ApoE4 and not everyone with ApoE4 suffers from it.

There is no way to accurately predict whether a particular person will develop the disease. It is possible to test for the ApoE4 gene mentioned above, but strictly speaking such a test does not predict whether a particular person will develop Alzheimer's disease or not. It merely indicates that he or she is at greater risk. There are in fact people who have had the ApoE4 gene, lived well into old age and never

developed Alzheimer's disease, just as there are people who did not have ApoE4, who did develop the disease. Therefore taking such a test carries the risk of unduly alarming or comforting somebody.

13. Alzheimer Europe agrees with diagnostic genetic testing provided that pre- and post-test counselling is provided, including a full discussion of the implications of the test and that the results remain confidential.
14. We do not actually encourage the use of genetic testing for assessing the risk of developing Alzheimer's disease. We feel that it is somewhat unethical as it does not entail any health benefit and the results cannot actually predict whether a person will develop dementia (irrespective of the particular form of ApoE s/he may have).
15. We are totally opposed to insurance companies having access to results from genetic tests for the following reasons:
 - a) This would be in clear opposition to the fundamental principle of insurance which is the mutualisation of risk through large numbers (a kind of solidarity whereby the vast majority who have relatively good health share the cost with those who are less fortunate).
 - b) Failure to respect this principle would create an uninsurable underclass and lead to a genetically inferior group.
 - c) This in turn could entail the further stigmatisation of people with dementia and their carers.
 - d) In some countries, insurance companies manage to reach decisions on risk and coverage without access to genetic data.
16. We therefore urge governments and the relevant European bodies to take the necessary action to prohibit the use or possession of genetic data by insurance companies.
17. Alzheimer Europe recognises the importance of research into the genetic determinants of Alzheimer's disease and other forms of dementia. Consequently,
 - a) we support the use of genetic testing for the purposes of research provided that the person concerned has given informed consent and that the data is treated with utmost confidentiality; and
 - b) we would also welcome further discussion about the problem of data management.
18. In our opinion, any individual who wishes to take a genetic test should be able to choose to do so anonymously in order to ensure that such information does not remain in his/her medical file.

F. Other considerations

19. People who take genetic tests and do not receive adequate pre and post test counselling may suffer adverse effects.
20. Fear of discrimination based on genetic information may deter people from taking genetic tests which could be useful for research into the role of genes in the development of dementia.
21. Certain tests may be relevant for more than one medical condition. For example, the ApoE test is used in certain countries as part of the diagnosis and treatment of heart disease. There is therefore a risk that a person might consent for one type of medical test and have the results used for a different reason.

**G. Annex 1: The Convention on Human Rights and Biomedicine
(Council of Europe - Oviedo, 1997)**

22. The following extracts from the above mentioned convention should be borne in mind:

- Any form of discrimination against a person on grounds of his or her genetic heritage is prohibited.
- Everyone has the right to respect for private life in relation to information about his or her health.
- Everyone is entitled to know any information collected about his or her health. However, the wishes of individuals not to be so informed shall be observed.
- Tests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counselling.

H. Annex 2: Universal Declaration on the Human Genome and Human Rights (UNESCO, 1997)

23. The following extracts from the above mentioned declaration should be borne in mind:

- Everyone has a right to respect for their dignity and for their human rights regardless of their genetic characteristics.
- That dignity makes it imperative not to reduce individuals to their genetic characteristics and to respect their uniqueness and diversity.
- The right of each individual to decide whether to be informed or not of the results of genetic examination and the resulting consequences should be respected.
- No one shall be subjected to discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights, fundamental freedoms and human dignity.
- Genetic data associated with an identifiable person and stored or processed for the purposes of research or any other purpose must be held confidential in the conditions foreseen by law.