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ANNOUNCEMENT

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Committee of Ethics
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Guidelines for genetic testing

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We have revised the two previous sets of guidelines proposed by the Japan Society of Human Genetics, "Guidelines for genetic counseling and prenatal diagnosis (1995)" and "Guidelines for genetic testing, using DNA analysis (1996)". We intend to draw the attention of medical research institutions, medical facilities, clinical testing companies, all persons involved with genetic testing, and the media, to the importance of genetic testing, and to explain the meaning of its importance so that people will respect and follow the guidelines.[1]

Advances in cell and molecular genetics have contributed to the development of human genetic research. However, some people point out that this new knowledge has created additional bioethical debates. As a background to this debate lies the fact that most private information relating to one's biological life is contained in the human genome. Also, genes are inherited by families and future generations. With currently available analytical technology, identification of an individual is possible, along with the detection of specific genotypes and chromosomal karyotypes. In particular, the number of genetic diseases that can be tested or diagnosed by DNA analysis is increasing year by year, and the usefulness of clinical genetic testing is widely accepted.

In this situation, important problems still remain, such as pre- and post-test genetic counseling, confirmation of informed consent prior to the test, ways of dealing with remaining samples, and ways of dealing with information obtained by the genetic tests. These problems need to be considered when genetic testing (including prenatal tests, carrier detection, pre-symptomatic tests, and susceptibility tests) is employed in clinical practice to elucidate and diagnose genetic information. Persons who are involved in genetic counseling or testing, in genetics clinics must protect the person taking the test (henceforth called the subject) and their family's human rights. Subjects must not be discriminated against because of their chromosomal karyotypes or their genotypes; also appropriate medical care should be given to persons who need care. The subject's decision must be respected in regard to the genetic testing. Accordingly, we propose the following guidelines be followed.

1. Genetic counseling should be conducted by a genetic counselor who has sufficient knowledge and experience of medical genetics.[2]
2. Genetic counselors must try to give the most recent and correct information to the subject (henceforth, called the client). This includes information on frequency of the disease, its natural history and rate of recurrence (genetic prognosis), and information about genetic tests such as carrier detection, prenatal tests, pre-symptomatic tests, and susceptibility tests. As there are various genotypes, phenotypes, prognoses, and responses to treatment, etc, within the same genetic diseases, persons involved in genetic counseling should keep this fact in mind.
3. Genetic counselors should try to use simple and easily comprehensible words to explain all matters. If the client wishes, and/or it is better to have a third person present, it may be useful to have one or more accompanying persons. Explanations should be written in a clinical record book, and kept for a set time period.
4. In counseling prior to genetic testing, the counselor must provide to the subject accurate information concerning the purpose, method, accuracy, and, in particular, the unfavorable limitations, of testing beyond the requirements of ordinary genetic counseling. In addition to oral explanations, written disease-specific information should be provided to ensure that there are no omissions.
5. The client and their family have both the right to know and the right not to know, which should be equally respected. Therefore, genetic counseling and genetic tests using personal identifying samples should be based on the autonomous decision made by the person taking the test. This decision should not be an instruction of, or directed, by the counselor. In this situation, the client has the alternative of not taking the test, and it should be explained to them that they will not suffer any disadvantage because of such a decision. In particular, for the pre-symptomatic diagnosis of adult-onset genetic disease, multiple counseling sessions should be conducted prior to any tests, and the decision of the subjects must

be shown to be the result of their own autonomous decision-making.

6. Genetic tests can be carried out only after informed consent is obtained.
7. Even though the client may desire genetic testing, the doctor can refuse to provide testing if the request is against social and ethical norms, or if it is against the doctor's personal principles. If refusing because of personal disagreement, the doctor should refer the subject to other medical facilities.
8. If a surrogate representative makes the decision because the subject is deemed to be unable to exercise autonomous decision-making, a decision for genetic testing must be made that protects the best interests of the subject. Therefore, the testing of children for adult-onset genetic disease that has no effective treatment or means of prevention should be avoided.
9. The fact that clinical features vary between subjects and rely on penetrance even if a susceptibility-related genotype is detected, should be explained to a subject who is to have susceptibility testing related to cancers or multifactorial inherited diseases. Also, the fact that even if the target gene does not have any susceptibility-related genotype, there is a possibility that disease will occur, should be explained. The kinds of medical approaches, if needed after the test, should be explained.
10. Genetic testing must be performed using only established and practiced techniques. The laboratories or organizations that are providing testing services should be monitored according to an established standard and they should conduct their own follow-up research and always strive to improve the accuracy of diagnosis.
11. The results of the genetic testing must be explained to the subject in easily understandable language. Even if the testing was unsuccessful or the result was inconsistent, the results must be explained to the subject.
12. If the genetic counselor deems it preferable to give the test results to the subject while they are accompanied by person(s) whom the subject trusts, rather than while they are on their own, the counselor should suggest this possibility. The subject can terminate analysis of the sample at any time, including in the middle of the analysis, and they can also decide not to know the results. Further, the subject should never suffer any disadvantage for taking that decision, and should be informal that this is the case.
13. Counseling after testing is essential, and should be continued as long as it is understood to be necessary. Also, medical/welfare support, including psychological and social support, should be prepared as needed.
14. All personal genetic information must be kept confidential and, basically, it cannot be given to another person unless the subject allows this. The utmost care is needed so that this information is not used as a source of discrimination.
15. It is suggested to the subject the result be disclosed to family members, and that they are tested, if the test result can be used to prevent or treat disease in the subject's family members, not only in regard to single-gene disorders but also for multifactorial disorders. If the subject does not confirm that they will reveal the information to their family after repeated efforts at persuasion, and if this information could certainly prevent the suffering of the family, it may be ethically acceptable to disclose the genetic information, only for diagnosis, prevention, and treatment, by the request of the subject's family. However, the decision as to whether to share the information or not should be made by the appropriate ethics committee, and not by the counselor.
16. The remainder of samples obtained for genetic testing can be stored for the future benefit of the subject and their family. The samples should not be used for purposes other than the original one. If the sample may provide useful information in the future on the disease in question or a related disease, written agreement to use the sample must be obtained from the subject after it is clearly explained that any identifying personal information will be deleted for such potential use.
17. In regard to prenatal testing, in consideration of currently available diagnostic technology and medical genetic knowledge, our views are expressed in the "Additional remarks" below. Treatment after the test should respect the subject's wishes, and the counselor may not participate in this decision-making. No matter what decision the subject makes, the subject and their family should be supported psychologically and socially. A system that allows for these factors is urgently needed.

Additional remark 1: opinions regarding prenatal diagnosis

1. Prenatal testing/diagnosis done in the first and second trimesters includes cytogenetic, biochemical, molecular, and pathological analysis, using amniotic fluid, chorionic villus, and fetal samples. In addition, instrumental diagnoses, including ultrasonic testing are also available.[3]
2. Invasive prenatal test/diagnosis procedures, such as chorionic villus sampling and amniocentesis, should be considered when the mother wishes to have a test/diagnosis in the following situations:
   a. If either one of the parents is a carrier of chromosomal abnormality
   b. If there is a history or experience of giving birth to a baby with chromosomal abnormality
   c. If the pregnant woman is of advanced maternal age
   d. If the pregnant woman is heterozygous for a serious X-linked disease
   e. If both of the parents are heterozygous for a serious autosomal recessive disease
   f. If either of the parents is heterozygous for a serious autosomal dominant disease
   g. If there is any other situation in which the fetus is predicted to contract a severe disease
3. Except when testing for an X-linked hereditary disease, the sex of the fetus should not be revealed.
4. Efforts should always be made to improve the accuracy of prenatal test/diagnosis technology.

5. As for maternal serum marker testing, we strongly recommend that it be conducted in accordance with the views previously stated by our committee and by the Ministry of Health and Welfare of Japan Health and Science Committee (Report of the Task Force on Maternal Serum Marker Testing).

Additional remark 2: pre-implantation testing/diagnosis[4]

Pre-implantation testing/diagnosis is one of the genetic tests. When (a) either parent is a carrier of chromosomal abnormality or (b) a carrier of severe autosomal dominant disease, or (c) both parents are carriers of severe autosomal recessive disease, or (d) the mother is a carrier of a severe X-linked disease, and, in addition, (e) does not wish for a genetic abortion, or (f) in an infertile couple who strongly wish to have the test under informed consent in accordance with these conditions, they may become subjects. However, this test needs highly developed techniques and it is recognized to be at the stage of clinical research. Therefore, institutions that are considering the conducting of pre-implantation testing/diagnosis should be aware of the ethical issues and proceed carefully.

Footnotes

1. The Japan Society of Human Genetics (JSHG), Council Committee of Ethics, has announced guidelines for genetic testing. These guidelines are valid for members of the Japan Society of Human Genetics. However, if actions that might be unethical, not in accordance with social norms, or inappropriate are carried out under these guidelines, and if person who is not a member of this Society takes the action, these guidelines do not have the power to prohibit such action. We intend to discuss this matter with the relevant scientific societies in the future, and we wish strongly for the government to set up a policy on genetic testing [see Holtzman and Watson (1998)].
2. At present, in Japan, there are medical doctors certified by the JSHG as "Clinical Geneticists" and those certified by the Japan Society for Genetic Counseling (the previous Japan Society of Clinical Genetics) as "Genetic Counselors"; these two certifications will be unified after discussion with the relevant societies. Also, there should be opportunities for the training of future medical genetic personnel, not only doctors but also co-medical staff, such as nurses and public health nurses, to better fulfill the needs of users.
3. Using ultrasonic tests, it is now possible to detect several fetal disorders, including some chromosomal abnormalities. Therefore, explanations should be given before the conducting of ultrasonic tests in accordance with the standard view given above in this guideline.
4. The Japan Society of Human Genetics, Board of Directors, respects the view stated by the Japan Society of Obstetrics and Gynecology with respect to pre-implantation test/diagnosis, and expresses the wish that this testing/diagnosis be conducted appropriately in an ethical and socially acceptable manner.

Reference

Holtzman NA, Watson MS (1998) Promoting safe and effective genetic testing in the United States. Final report of Task Force on Genetic Testing. The Johns Hopkins University Press, Baltimore