

Young Leaders' Program Colloquium

Genetic Testing of Children for the Sake of Other Family Members: Ethical and Legal Issues

(Nagoya University School of Medicine)

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Purpose of This Colloquium

In this colloquium, I would like to discuss the ethical and legal problems of genetic testing of minors for the sake of other family members using a case report by professor Mariko Tamai of Shinshu University.

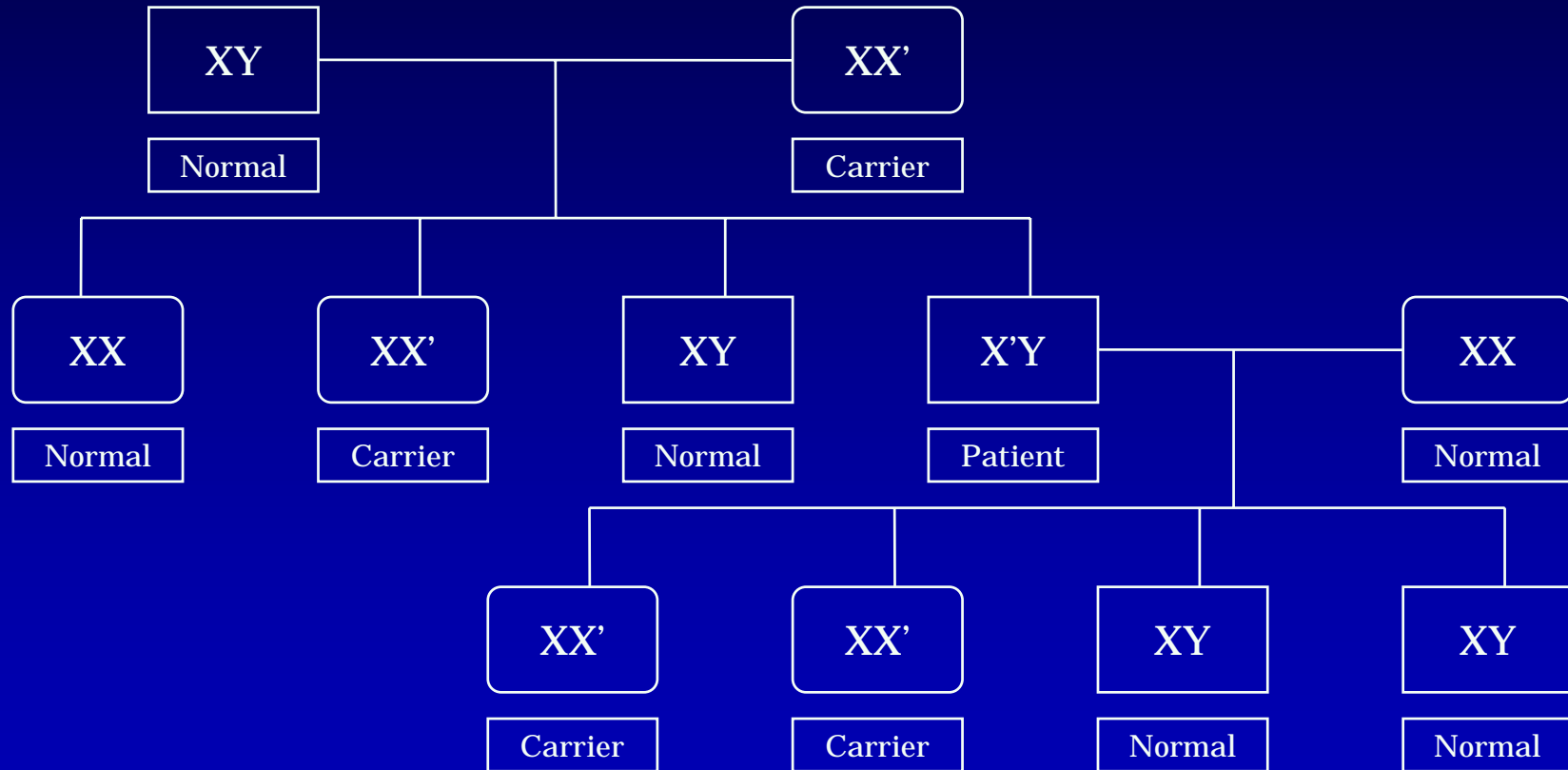
Professor Tamai is a clinical psychologist at the Department of Clinical and Molecular Genetics of Shinshu University Hospital. Professor Tamai kindly gave me permission to use her materials in this colloquium.

Notes for Professor Tamai's Case

Professor Tamai's Case is concerned with the genetic testing of two boys who were suffering from Duchenne type muscular dystrophy (DMD).

DMD is a hereditary (X-linked recessive inheritance) muscle disorder with onset usually before age 6. Characterized by symmetric weakness and wasting of first the pelvic and crural muscles and then the pectoral and proximal upper extremity muscles. Progression is steady. Most patients are confined to a wheelchair by age 10 or 12 and used to die of respiratory complications by age 20 yr. However, recently with the help of a respirator some patients survive into their thirties.

X-linked Recessive Inheritance



Professor Tamai's Case

(The following case was posted on her home page*, and with her permission, is quoted here with some changes.)

Professor Tamai's client, in her 40s at that time, had two male children. Both had been diagnosed as Duchenne muscular dystrophy (DMD) based on their neurologist's clinical observations. The older boy was 14 year old. The younger one was an 11 year old. The client became pregnant unexpectedly. When she visited the clinical genetic department of Shinshu University hoping for a healthy baby, she was in the 10th week of pregnancy.

[*http://square.umin.ac.jp/~mtamai/gene_info.htm]

Professor Tamai's Case

The client wanted to know if the fetus had DMD. She said that it would be too much for her to raise another child with DMD since she already had two with the disease. After careful considerations, she decided to visit Professor Tamai's department. She had not told either of the boys about her pregnancy.

In order to find out if the fetus had DMD, it was necessary to investigate the genes of both boys to identify the specific type of mutation. In this case, neither of the boys had been genetically tested.

Professor Tamai's Case

As a cause of the DMD, there are three types of mutation of dystrophin gene; deletion, duplication and point mutation. More than half of all the cases are due to deletion. If their mutation type was deletion, the fetus could be genetically tested using their genetic information. In order to obtain their genetic information, blood samples of both boys had to be taken.

Professor Tamai's Case

The client did not want to tell her boys about her pregnancy and the necessity of their genetic information.

One week after the first visit, she came to the outpatient department with her two boys. She had told the boys only that they would go to the hospital for some tests.

After some general checkups, blood samples were taken from them without telling them the nature and purpose of the test.

Question 1

For the purpose of prenatal diagnosis, is it acceptable to take a blood sample from the parents' other children (who are already born and suffering from the disease), use that blood sample to carry out an examination for genetic mutation related to the disease, then use the resulting genetic information to avoid giving birth to another child having the same disease?

Suppose the child is 14, 11, 7 or 2 year old. Does the degree of youngness of the child affect your answer?

Professor Tamai's Case

After taking the blood from the boys, the information of their gene mutation was obtained. It identified the mutation as deletion. Then my client in the 15th week of pregnancy had an amniocentesis. The chromosomal test determined the fetus was female. As the X-linked recessive disorder usually appears only in a male person (a female fetus has 50 percent chance of being a carrier of the disorder), and the risk of the fetus having the DMD decreased to a negligible level, she continued her pregnancy and gave birth to an apparently healthy girl. The boys genetic information was not used at this stage.

Professor Tamai's Case

However, when the girl grows up and if she wants to know before her marriage whether she carries the same mutation as her mother and brothers, the information kept in each brother's file will be needed.

Question 2

If, 20 years later, the sister decides to find out whether she is a carrier of the dystrophin gene mutation, should her brother's genetic information be made accessible to her (her doctors) without her brothers' informed consent?

In the meanwhile, how should the parents and doctors behave toward her regarding this matter, anticipating she, someday, will become to like to know her own carrier status of the brothers' disease.

Problems

1. Should it be allowed to perform a genetic test of a minor patient suffering from X-linked recessive disorder such as Duchenne muscular dystrophy only with the consent of his parents, when the sole purpose of the test is making it possible to conduct prenatal diagnosis of the fetus who, if born, will be his sibling, or carrier testing of his female (already born) siblings or aunts? If it is allowed, what justification can be offered for it?
2. If the genetic test of the patient has already been done, and using his genetic information, it is possible to carry out such prenatal diagnosis or carrier testing, may it be used only with the consent of his parents? If so, on what basis is it permissible?

Principles

1. Informed consent

In order to carry out a genetic test, informed consent must be obtained from the individual to be tested. If the patient is a young child and incompetent to give his own consent, proxy consent must be obtained from his parents. Parents are authorized to give proxy consent, as they are expected to act so as to further the best interest of their child. Therefore, their authority to give proxy consent for their child is limited to the cases where the child will benefit from the procedure.

Principles

2. Confidentiality of Medical Information

The genetic information obtained from the genetic test is confidential information of the tested individual.

It should be disclosed to him as long as he wants to be told.

It may be disclosed to his relatives or family members if he consents to it being disclosed. It may not be disclosed to them against his will.

UNESCO, International Declaration on Human Genetic Data (October 2003)

Article 8: Consent

- (a) Prior, free, informed and express consent, without inducement by financial or other personal gain, should be obtained for the collection of human genetic data, human proteomic data or biological samples***, and for their subsequent processing, use and storage***. Limitations on this principle of consent should only be prescribed for compelling reasons by domestic law, consistent with the international law of human rights.
- (b) When, in accordance with domestic law, a person is incapable of giving informed consent, authorization should be obtained from the legal representative, in accordance with domestic law. The legal representative should have regard to the best interest of the person concerned.

UNESCO, International Declaration on Human Genetic Data (October 2003)

Article 8: Consent

- (c) An adult not able to consent should as far as possible take part in the authorization procedure. The opinion of a minor should be taken into consideration as an increasingly determining factor in proportion to age and degree of maturity.
- (d) In diagnosis and health care, genetic screening and testing of minors and adults not able to consent will normally only be ethically acceptable when it has important implications for the health of the person and has regard to his or her best interest.

UNESCO, International Declaration on Human Genetic Data (October 2003)

Article 14: Privacy and Confidentiality

(b) Human genetic data, human proteomic data and biological samples linked to an identifiable person should not be disclosed or made accessible to third parties, in particular, employers, insurance companies, educational institutions and the family, except for an important public interest reason in cases restrictively provided for by domestic law that is consistent with the international law of human rights or where the prior, free, informed and express consent of the person concerned has been obtained provided that such consent is in accordance with domestic law and the international law of human rights. ***

My Tentative Answer

Testing incompetent minors (and using their genetic information) for the sole purpose of prenatal diagnosis or carrier testing is permissible only if

1. The minor is a symptomatic patient.
2. Testing carries only minimal risk to the minor.
3. The minor himself does not indicate his opposition to his being tested (or his information being used) after given adequate information in accordance with his power of understanding.
4. The minor has an intimate relation with the person who will utilize test result.
5. There is practically no alternative to the testing of the minor.
6. Ethics committee approval has been secured.