International Symposium on Bioethics

Genetic Testing of Children and Confidentiality of Their Genetic Data

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Problems

1. Is it allowed, in your country, to perform a genetic test of a minor patient suffering from X-linked recessive genetic disorder such as Duchenne muscular dystrophy only with the consent of his parents, when the only 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 it being disclosed. It may not disclosed to them against his will.

Professor Tamai's Case

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

The client, in her 40s at that time, had two male children with muscular dystrophy. Both were diagnosed as Duchenne muscular dystrophy (DMD) based on their neurologist's clinical observations. The older boy was a 14-year-old 8th grader. The younger one was an 11-year-old 5th grader. The client became pregnant unexpectedly. When she visited the clinical genetic department hoping for a healthy baby, she was in the 10th week of pregnancy.

Professor Tamai's Case

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

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.

Professor Tamai's Case

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

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 following case was posted on her home page and, with her permission, is quoted here with some slight revisions.)

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 in your country to take a blood sample from the parents' other children (who are already 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 in this stage.

However, when the girl grows up and if she wants to know before her marriage whether she carries the same mutation, 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.

My Tentative Answer

Testing incompetent minors 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 subjected to the test 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.**