

INTERNATIONAL ETHICS

Emerging issues in genetic testing in Sri Lanka

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The ethical problems associated with genetic testing have been widely discussed. These issues include informed consent prior to testing, confidentiality of results, prenatal diagnosis and abortion of affected fetuses. The following cases illustrate some of the ethical issues that have arisen in Sri Lanka and are also likely to occur in other countries where genetics services are being introduced.

The first case is of a one-year-old boy with Down's syndrome (trisomy 21) whose parents were asked by the paediatrician to get a chromosome analysis conducted on him. The parents accepted this advice and paid Rs 5,000 for the test. The test confirmed the diagnosis. It did not change the care given by the parents or the paediatrician. The parents had already decided that they would not have more children. A review of the chromosome result suggested errors in the identification of the chromosomes although the diagnosis of Down's syndrome was confirmed.

The second case was of a newborn baby with multiple congenital anomalies who was suspected of having a severe chromosome disorder, trisomy 13. The disorder is associated with reduced life expectancy and severe problems if the child survives. A confirmed chromosome result would have enabled the parents and doctors to make an informed decision regarding the future care of the child. The parents were unable to pay for the test and therefore the test was not performed.

Testing does not always help

In an ideal situation testing would be recommended for all cases of Down's syndrome. When the parents have to pay for such tests, it is more important but also difficult to balance the advantages and the disadvantages of testing. In the first case, the diagnosis, made on the basis of the child's clinical features, was not in doubt. Confirming it with chromosome tests made no difference to his care. In some cases a chromosome rearrangement can occur and increase the risk of another affected child in the family. Even if this had been the case, the parents were not planning to have another child. If a chromosome rearrangement was identified, what could have been done about it? Prenatal diagnosis using amniocentesis is being introduced in Sri Lanka, but it is expensive and only available in the private sector. Even if an anomaly is confirmed by amniocentesis and the parents request an abortion, Sri Lankan law does not permit such a choice. The only benefit is that the parents are made aware of the problem before birth of

the child but would the cost be justified?

In the second case, decisions regarding aggressive versus supportive therapy could have been made more confidently if accurate chromosome tests were available. If the test had been performed, the parents should have been made aware of the implications of the diagnosis and of future care if trisomy 13 was confirmed. Both cases illustrate the need for the involvement of parents and for making informed decisions prior to genetic testing.

Is informed decision making possible?

In countries where genetic services are offered as part of an organised system, genetic counselling is usually offered prior to testing. Counselling enables clients to make an informed choice about genetic testing. Many factors can make it difficult to make informed decisions in Sri Lanka. People's knowledge of genetics is poor and access to information in local languages from sources such as the Internet is limited. Parent support groups are also scarce. Referring physicians are likely to get information from the laboratories offering genetic testing, and they may not be adequately aware of the limitations of testing. The influence of the doctor may make it difficult for parents to refuse genetic testing even if they feel it is unnecessary or unaffordable.

The limited availability of independent, trained genetic counsellors adds to the problem in Sri Lanka. Hospitals offering testing also offer genetic counselling, but the counsellors' financial links with the hospitals and laboratories can lead to conflicts of interest. Maintaining an adequate number of specimens is necessary for the laboratory to function in a cost effective manner and the lab may feel it necessary, or may be forced, to increase the number of specimens being analysed.

The average monthly salary in Sri Lanka is Rs 3,057. The average family income is Rs 12,803. A chromosome test that costs Rs 5,000 is a significant expense for most families. This is illustrated by the second case, where genetic testing was arguably justified but the parents could not afford the expense.

The incorrect pairing of the chromosomes in the first case suggests that there was inadequate quality control at the laboratory. A laboratory offering genetic testing should be accredited as having met quality control standards. In deciding to recommend a test, referring doctors should be aware of the quality standards of a laboratory as an inaccurate result may

mislead them in their clinical decisions. The clients should also be aware of this possibility because they have to decide whether to pay for a test of poor quality, not have a test at all or opt for investigations performed by an accredited but more expensive laboratory.

The introduction of testing for human genetic disease in Sri Lanka has raised many issues about the benefits of the tests. Further assessment is required to determine the true costs and benefits of such tests in developing countries where patients have to fund their tests.

Did I kill a man?

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When I refused to give a false certificate to a 15-year-old girl which stated that she was sick and therefore could not attend school for 15 days, little did I know that I was signing the death warrant of a man I had never seen in my life. Manjula, the girl, pleaded with me, saying that her teacher was very strict, but I did not relent. I firmly showed her the door.

Two months later, I had forgotten about Manjula, assuming that she must have got the certificate from some of my less rigid colleagues. Her mother suddenly brought me back into the picture. Manjula, she told me, was so scared of the teacher that she had not attended school for many days. Meanwhile, Krishna, a young man, eloped with the lonely girl. A police hunt subsequently forced him to bring her back. The abduction of a minor girl is a serious offence. A desperate Krishna committed suicide to avoid police action and infamy.

Did my refusal to issue a certificate somehow trigger this sequence of events? At the time, I was very clear that I did not wish to issue a false certificate. However, later events forced me to revisit my opinion. Was I too rigid? Should I have judged the need for a certificate keeping in mind the circumstances?

If someone had asked me to issue a certificate of sickness to avoid being present in court, I could have been justified in refusing it outright. If an employee wanted to use sick leave that was

lapsing, I would never have issued a certificate. But here was an innocent girl who only wanted to escape the wrath of a schoolteacher.

I could not have foreseen the events that unfolded. However, I could have at least perceived that she did not ask for the certificate for monetary or any other gain. By issuing the certificate, I was not going to harm or cause any loss to anyone. Maybe I could have asked Manjula to bring along her mother before issuing the false certificate. Perhaps I could then have felt satisfied that I had done my duty by informing the parent.

Are we sometimes too rigid in implementing the law? Do we follow it more in letter than in spirit? Making exceptions to any law can, of course, be dangerous. An ultrasonologist may justify doing a sex determination test by saying that the mother did not intend to perform an MTP. Or that she already had five female children and it was humane to perform the sex determination and even an MTP. To extend the analogy, should euthanasia not be banned and instead be performed after considering the specifics of each case?

I still debate with myself. Did I do the right thing? Would Krishna still be alive if I had given the certificate? I do not have an answer.

(All names have been changed to protect identities.)