

# ‘What would you do, doctor sahib?’

In urban India, there is an increasing public awareness regarding the role of genes in the incidence of disease and the possibility of making use of pre-natal testing (PNT). PNT is associated with modernity and good parenthood. In this paper, Jyotsna Agnihotri Gupta throws light on women’s use of PNT for pregnancy management and the decision making regarding genetic testing – and whether to continue the pregnancy following a ‘positive’ test result – in order to achieve the birth of ‘healthy’ children.

Jyotsna Agnihotri Gupta



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GENETICISTS IN INDIA could be the new pundits instead of the ones traditionally casting horoscopes after the birth of a child to predict the future course of its life. Since the second half of the 20th century genetics is slowly changing cultural understandings of health and illness, introducing a new dimension to health care. Seventy percent of India’s approximately 1.17 billion population lives in rural areas, where basic health care services are either unavailable or inadequate. India scores very low on important health indicators such as life expectancy, infant and child mortality and maternal mortality. Given the size of the country’s population, genetic diseases affect a small number of people, and when diagnosed, they are largely untreatable. However, for the families concerned they pose a considerable burden, since care and rehabilitation facilities for individuals with genetic disorders are very poor.

## Reproductive genetics

Over 25 million births occur annually in India. The infant and child mortality rate is very high, with infectious diseases being the primary cause of death. Most births still take place under the supervision of (skilled or unskilled) traditional birth attendants and there is a lack of simple ante-natal and post-natal services in rural areas. In the cities pre-natal check-ups of pregnancies occur in clinics and thus congenital malformations and genetic disorders may get diagnosed; they are increasingly reported as important causes of peri-natal mortality. If the incidence of birth defect is assumed to be two per cent, then 500,000 babies are born with some form of birth defect every year in India.<sup>1</sup> Paediatricians and geneticists argue that prevention is the best strategy, considering that cure after birth is difficult and costly. With the proliferation of genetic tests, testing is becoming a part of ‘good parenthood’ at the individual level. At the societal level, community genetics is emerging as a practice and as a future model in which the options offered by genetic testing technologies can be used optimally for the realisation of preventive health care. Certain genetic conditions are seen as a major drain on the quality of life of the individuals concerned and their families, as well as on health resources. While the prevalence of haemoglobinopathies (blood-related diseases), such as thalassaemia and sickle-cell disease and its variants has been noted in certain communities and regions of India, recent studies bear out that the primary reason for referral at genetic medicine departments is reproductive genetics. The main aim of reproductive genetics is early detection of a heightened risk of giving birth to children with a disorder which is untreatable and which could (in varying degrees) undermine the child’s quality of life and life span, depending on the severity of the condition.



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## Genetic testing

Genetic testing may be carried out at different times throughout life and through the use of various techniques. In general there are four moments of genetic testing: (1) carrier testing; (2) preconceptional testing of the embryo in assisted reproduction (IVF), referred to as pre-implantation genetic diagnosis (PGD); (3) pre-natal testing of the foetus during pregnancy (PNT); and (4) screening of neo-natals, or later in life for late onset genetic conditions.

The most common form of genetic testing is pre-natal testing (PNT). PNT refers to all methods used to ascertain the health of the developing foetus. These include biochemical screening (maternal serum alpha-fetoprotein, triple marker screening), ultrasound, chorionic villi sampling<sup>2</sup>, amniocentesis<sup>3</sup>, fluorescent in situ hybridisation technology (FISH), and DNA assays. Different techniques may be used for PNT, either individually or in combination. Some tests are not in themselves diagnostic and serve only as a first stage in screening for certain disorders; other tests, even using different methods of diagnosis, may be required to come to more or less reliable results. Genetic tests are quite expensive and not all families can afford them.

## Pre-natal genetic testing in India

Increasingly urban Indian women are undergoing PNT to produce ‘healthy’ children. During my empirical research in hospitals in Delhi and Mumbai in 2007 and 2008 I found that pregnant women registering with gynaecologists in cities were increasingly being advised to have a triple marker test in the first trimester of pregnancy. A very large number of them received a ‘positive’ diagnosis, i.e. a heightened risk of the foetus being affected, and were advised to undergo further tests such as chorionic villi sampling or amniocentesis in the second trimester. Often, women seeking further testing and genetic counselling were those who had suffered repeated miscarriages, or were ‘high-risk pregnancies’ due to advanced maternal age (cut-off point used is 35 years). Also, there were pregnant women and couples with an elevated risk – with a family history of a genetic disorder, knowledge of an abnormal gene in the family, a previous child with a disorder, or suspect findings from previous tests. However, on examination of the records of the cytogenetic laboratories of the two hospitals in Delhi where I conducted research, I discovered that in 95 percent of the cases sent for second trimester testing it was a false alarm and no abnormality was detected.

PNT plays upon two fundamental desires of all couples: to become parents and to produce healthy and ‘normal’ children. Pregnant women seemed to have ambivalent feelings regarding PNT. They were not sufficiently informed regarding genetic tests and procedures and what the diseases meant in terms of reduced functioning. The very existence of these technologies, coupled with the advice of their gynaecologists and/or genetic counsellors to undergo these tests, created a kind of pressure on women. As one woman put it ‘Is it necessary to go through this [amniocentesis]? No one in the family has a genetic condition.’ While they did not want to risk giving birth to a disabled child, many women mentioned that before the technology appeared women were giving birth to healthy babies, also at an advanced age. Yet, once advised testing, many saw it as a necessary evil and complained about the anxiety caused by the tests until the results were known, the inconclusiveness of certain tests entailing the need for further tests, and the concomitant risk of miscarriage.

It is remarkable that none of the women/couples I interviewed said they would continue the pregnancy following a ‘positive’ test result. The ‘pressure’ to choose abortion after PNT was rather subtle: it arose mainly from intending parents’ own understanding that it was ‘not fair toward a child to bring it to birth with a severe disability’. The main reasons for the

## Pre-natal testing in the socio-cultural context of India

proliferation of PNT are the dearth of institutional facilities for the disabled, and absence of medical insurance to bear the costs of often very expensive therapies for a child who suffers from a genetic condition. Caring for a disabled child results in an enormous financial and care burden for families, the latter falls mainly on the women. Also, fear of social stigmatisation not only of the affected child, but its parents and the whole family plays an important role. For this reason genetic testing and genetic counselling are increasingly sought.

## Genetic counselling

Clinical facilities for genetic investigation in India are rather poor and there is a shortage of trained clinical geneticists. There are only 25 genetic counselling centres in the whole country. According to geneticist I.C. Verma (personal communication):

*“Much of the work consists of providing information – helping patients to come to a decision. Many come with some diagnosis already; we make a more specific diagnosis. It often comes as a real shock to them. They are not ready to accept that there is no treatment for the condition. They require a period of adjustment. There is no rejection of affected children once they are born. There is more acceptance here than in the West... They do express their difficulty in looking after an affected child. Having a child with a genetic disease is quite tough... Patients wonder ‘why has it happened to us?’ Some put all their faith in the counsellor and listen to you; some check and recheck; they doubt everything you say. Some are angry at the doctor about what has happened. Financial reason is important. Family members’ views are very important. They discuss with their own parents; generally they don’t want to let other family members know, because of social stigma.”*

Although genetic counsellors uphold the principle of non-directive counselling, i.e. let women/couples themselves decide regarding (further) testing and whether to continue the pregnancy after an adverse test result, they say, that in general:

*“Parents want more directive advice. ‘What would you do, doctor sahib, in our situation?’ Non-directive advice is not acceptable, because they are not so literate about science. They say they will discuss with the family whether to go for further testing. Unlike the West, whenever there is an abnormality in a child the whole family comes for counselling here. It is very rare that only a couple comes; in fact the couple is the quietest... We try to take away the guilt of having given birth to an abnormal baby.”* (Dr I.C. Verma)

In principle, genetic tests may be empowering for women who are the child bearers and carry the primary responsibility for raising children, and caring for the sick and elderly. However, there may also be negative consequences of PNT for pregnant women. In the highly patriarchal Indian society men and the woman’s marital family traditionally make important decisions within the family. Women may face coercion in making reproductive choices regarding not only the number of children (quantity), but also the kind of children (quality) to be born, thus making it difficult to ensure their free and informed consent for use of genetic services. Also, the consequences of a ‘positive’ test may be disastrous for a daughter-in-law, as women often feel guilty or are blamed for producing a girl, or a disabled child. There were cases when husbands refused to be tested for carrier status for thalassaemia, although they were informed that the future offspring may actually manifest the disease only if both the parents were carriers.

## Enhancing informed decision making

Prevention of serious disease through genetic testing is a legitimate public health goal. It is imperative that the government invests in increasing health facilities for genetic testing in the public sector, so that for families at risk, if desired, genetic testing is affordable and accessible. Simple screening and diagnostic facilities at district health centres with the possibility of referral to specialised centres should be provided. Health literacy programmes for the public which include information on genetics and on societal implications of developments in the field of human genetics need serious attention.

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Developments in genetic technology concern all individuals, but since it is women who bear children and are the main carers, decisions affecting their pregnancy and outcome concern them more. Introducing pre-natal testing as part of standard pre-natal care should go hand-in-hand with an adequate system of informing and counselling women about pre-natal screening to ensure informed decision-making. Research has shown that decision aids are able to improve the quality and the level of informedness of pre-natal testing decisions and such decision aids should be developed and implemented in the pre-natal testing setting. Education materials need to be developed on genetic testing and screening that are culturally sensitive and in an accessible language. Also, the government needs to invest in institutions and care facilities for the disabled, and support for families with disabled children so that pregnant women do not feel compelled that terminating the pregnancy of an affected foetus is the only possible choice.

Jyotsna Agnihotri Gupta  
Research Fellow, SMAP  
International Institute for Asian Studies  
University for Humanistics, Utrecht  
jagupta@telfort.nl

## References

- Verma, I.C. and Bijarnia S. 2002. The burden of genetic disorders in India and a framework for community control. *Community Genetics*, 5, 192–196.
- Chorionic villus sampling (CVS) is a form of pre-natal diagnosis to determine chromosomal or genetic disorders in the foetus. It entails getting a sample of the placental tissue and testing it. CVS can be carried out 10-13 weeks after the last menstrual period, earlier than amniocentesis (which is carried out at 16-20 weeks).
- Amniocentesis is a medical procedure in which a small amount of amniotic fluid, which contains fetal tissues, is extracted from the amniotic sac surrounding a developing foetus, and the fetal DNA is examined for genetic abnormalities.



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