Understanding Birth Defects

Dilemmas of a Genetic Counselor

❍ **Puloma Shah**

As a genetic counselor with more than twenty years of experience, it is my delicate task to impart knowledge of human genetics to those seeking such counsel, usually families with a challenged child, or families running the risk of producing such a child due to a family history of genetically determined diseases such as the blood disorders Hemophilia and Thalassemia, as well as Down Syndrome. These diseases often recur in families with a history of that particular disease. The chances of recurrence depend upon the pattern of inheritance of the disease. Genetic counseling helps parents and families learn about these diseases, and guides potential parents on ways to decrease the risk of bearing children with a genetic disease, as well as on available treatments for those already born.

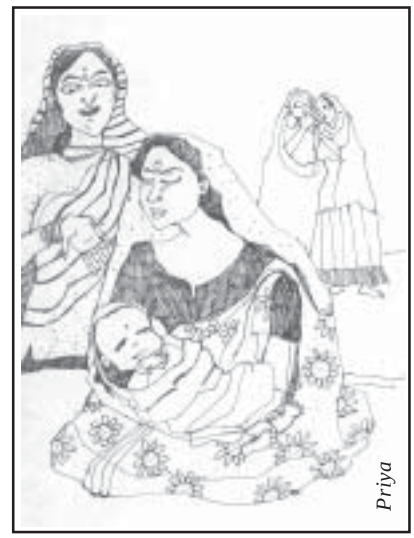
In my counseling work I interact with people from various professions, social-strata and diverse backgrounds. For many of them, their dreams and hopes have turned into despair and frustration, guilt and sadness due to the deleterious effects of genetically determined diseases. The severe physical, emotional and financial toll of genetic diseases has changed their lives forever.

Whether it is the birth of a daughter, a challenged child or infertility, it is most often the woman who is held responsible, leading to harassment, domestic violence, abandonment, or divorce.

In a country where families are obsessed with having sons, the birth of a daughter is usually unwelcome. Most families blame women for not having sons, though it is the sperm of the male that contains the genetic sex determinant.

There is a similar bias when a married couple has to face the trauma of infertility. The woman very often is considered at fault. About 10 per cent of married couples face infertility problems. Infertility is as likely to be caused by the man as by the woman. Approximately 40 per cent of the cases of infertility are due to the man, and 40 per cent due to the woman, while in the remaining 20 per cent the cause is unknown. When a couple visits a fertility clinic, multiple tests are done to find the reason for the infertility. In many cases, when the test results reveal that the problem is with the husband, he is shocked and finds it hard to accept. Wives are far more likely to accept the results of the tests, and marriages rarely break up if the man is the one who is infertile. However, the same is often not the case in the reverse situation.

Pregnancy and childbirth are a very special and vulnerable time in a woman's life. When a child is born with a birth defect, the new mother often gets into a terrible depression and suffers from an acute sense of worthlessness. This is the time when she needs support from everyone around. Instead, many women face emotional and mental harassment from their in-laws and husband because the defect is frequently blamed on her. Two to three percent of all newborn babies have a birth defect of some kind. Often, the family feels the birth defect is due to some flaw in the behaviour or the physiology of the woman. Unable to defend herself since she is dependent and often unaware of the true reasons for the defect, the woman often feels helpless and rejected.

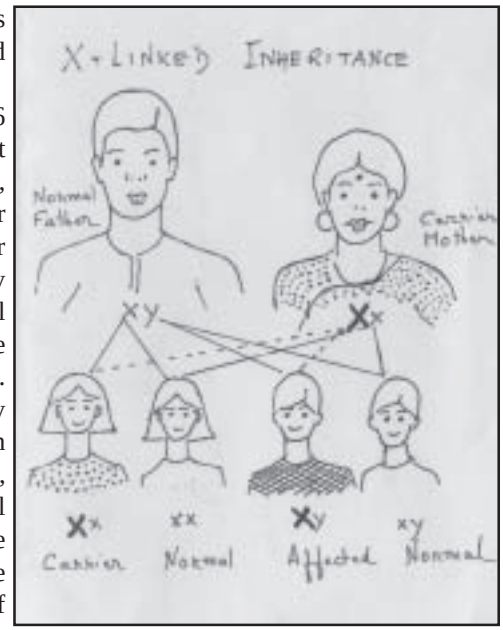


X-linked diseases occur when the mother carries a faulty gene on one of her X-chromosomes (even though she does not show any signs of the disease). In such a case, a son has a 50 per cent risk of inheriting the faulty gene and manifesting the disease. A daughter has an equal chance of being a "carrier" like her mother, remaining unaffected by the disease herself but capable of transmitting it to her sons. Fortunately prenatal diagnosis is now available, where one can detect whether the male foetus is affected or not. Women with a family history of X-linked diseases should take advantage of this technology.

There are rare cases when families with no history of such X–linked diseases unexpectedly have a son with this disease. This can happen when a mutation (change) from a normal gene to a faulty one occurs in a woman's genetic material. In such a case, if the husband is told that his wife is the "carrier" of the faulty gene, he often blames her or abandons her and his child, and

remarries. Sadly, the woman ends up forced to raise the challenged child single-handedly.

Every person is a carrier of 5-6 recessive disease-causing genes. It is a matter of sheer chance that, sometimes, a person who is a carrier of a faulty gene marries another "carrier" with the same kind of faulty gene. Unaware of this mutual condition, they sometimes produce a child that has a genetic disease. For such a couple there is commonly a recurrent risk of 25 per cent with each pregnancy. In such situations, carrier detection tests and prenatal tests are recommended to the couple so that the health of the child can be ascertained in the early months of future pregnancies.



In such cases also, the information is sometimes used as a tool against the woman - as if her faulty gene alone has created this problem. The difficulty in dealing with such situations is because of societal ignorance about the role of genetic inheritance. There are many instances when husbands feel that the option of a second marriage is more convenient than undertaking prenatal diagnostic tests for future pregnancies. Thalassemia (a blood disorder) is an example of the relatively infrequent diseases that show this kind of recessive inheritance pattern.

Genetic diseases differ from other medical disorders in their impact on the family because of the risk of recurrence. The risks vary from 5 per cent to 50 per cent depending on the inheritance pattern.

As genetic counselors we explain the intricacies of heredity to families in simple ways. We tell them how a man and a woman both have a role to play in transmitting heredity. We try to help them understand how these diseases are inherited. We stress that women's genetic composition are not more determinative of a child's health than men's. We see to it that the woman does not feel guilty for something not her fault, and family members do not accuse her unfairly. As the family's beliefs are often still deeply rooted in their religious, cultural and social heritage, we clarify misunderstandings and try to dispel ignorance and superstition.

Counseling families is a complex task for genetic counselors because of the varying socio-economic and cultural backgrounds of their clients. For example, the experiences of urban, literate, independent women are different from rural, illiterate women living in joint families. All these factors are taken into consideration before counseling on any aspects of genetic diseases. At the same time, one of the most important points of genetic counseling is the value placed on honesty. This raises a lot of ethical quandaries. For instance, genetic counseling becomes very tricky when a woman is found to be the 'carrier' of an X-linked disease. Imagine a couple from a rural background, illiterate and with one affected child. You look at the wife and see her vulnerability. Then you wonder whether it would be wise to tell the couple that the woman is the 'carrier' of the disease causing gene, or if it would be wiser to leave that information out, and just restrict your counseling to urging them to go for a prenatal diagnosis for future pregnancies, which would prevent any more children being born with the disease.

Every person is a carrier of 5-6 recessive disease-causing genes. It is a matter of sheer chance that, sometimes, a person who is a carrier of a faulty gene marries another "carrier" with the same kind of faulty gene.

Sometimes, there are instances where detection of a chromosomal (genetic material) rearrangement in one of the parents is a cause of infertility, recurrent abortions or a malformed child, though in most cases prenatal diagnosis can help.

Such situations also lead to the same kind of counseling dilemmas. If the responsible parent is the husband, the wife is often likely to accept it, but if it is the wife, the implications can be serious. Marital discord may follow. She can be blamed for the problem, harassed and sent back to her natal home. Ultimately, it is not only she who suffers, but her children too.

If her husband does decide to divorce her it is only the beginning of the ordeal. An even more difficult battle for the woman is getting maintenance for herself and her children and being allowed to lead a life of dignity after divorce.

Taking into consideration the social reality, it is very important that we protect these women from any discrimination or injustice they may face solely because of their genetic make up. ❐