

## Social and cultural issues in genetic counselling

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Medical genetics has rapidly emerged as a definitive branch of modern medicine with an increasing number of illnesses described as having an aetiological basis in alterations in one or more genes. Communication of the nature and implications of these disorders to the 'at risk' family is the basis of genetic counselling. The complexity of information to be given to a family necessitates a trained genetic counsellor. This article is based on the author's experience in counselling families with genetic disorders, public perceptions and misconceptions, and the influence of local customs and traditions on attitudes towards such disorders.

Formal training in genetic counselling is available in several countries ([www.agnc.org.uk](http://www.agnc.org.uk), [www.abgc.net](http://www.abgc.net), [www.hgsa.org.au](http://www.hgsa.org.au)) in which the spoken language is primarily English. In India, the counsellor working in an urban setting has to communicate with and counsel individuals from multiple language backgrounds in the course of a single day (Elackatt 2013). An easily understood word for 'gene' or 'genetic disorder' is not available in any vernacular language. Such issues, and the fact that a significant proportion of patients seeking genetic counselling are uneducated, pose a daily challenge to the counsellor. Also, unlike most other medical specializations, genetic medicine has implications not only for the affected individual but also for the wider family.

The questions most often asked by couples who have had an affected pregnancy or a child with a genetic disorder are:

- What is the diagnosis?
- Why did it happen?
- Was it because of something I did or did not do in pregnancy?
- Will it affect the life span, intelligence, prospects of marriage and child bearing?
- Is there a treatment?

- Will it happen again if I have more children?
- What can I do to prevent it from happening again?

Addressing these common questions in a manner that is understood by all individuals seeking counselling irrespective of their background is the basis of successful communication. Genetic counselling encompasses communication of information about a genetic disorder, its pattern of inheritance, prognosis for the affected individual, recurrence risks, future reproductive options, identification of other family members at risk and the availability of treatment options and support services (Harper 2010; Clarke and Thirlaway 2011).

Among the most common referrals in any genetic clinic are individuals with Down's Syndrome (DS). While the majority of such individuals have three copies of chromosome 21 (Trisomy 21), a few may have a translocation type of DS, which increases the risk of recurrence in subsequent pregnancies (Bawle 2012). The picture of a karyotype is often used in counselling to demonstrate the presence of the extra chromosome. The family being counselled sometimes derives their own interpretation of such images. Queries occasionally raised are whether the extra chromosome may have been contributed by a previous partner of one or the other spouse or whether it could be removed from the body by antibiotics as the chromosomes look like 'worms in the blood'. A family with an apparently balanced translocation segregating through various family members refused to consider prenatal diagnosis as an option believing that several sessions of '*pranic healing*' had rid the family of the genetic 'mistake'. So convinced were they that they had enthusiastically come forward to offer this therapy to other families visiting this counsellor's clinic!

For parents seeking counselling, whether it is a baby with a congenital anomaly or an individual with intellectual disability, one of the main questions asked or implied, is regarding future possibility of marriage and chances of bearing children. Males with DS are commonly thought to be infertile. Recent studies

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indicate that the level of intellectual disability in DS patients may affect the likelihood of natural procreation (Stefanidis *et al.* 2011). Nevertheless, in India increasing numbers of adult males with DS are able to find wives of normal intelligence (often from poorer or less educated families), probably as a result of parental perseverance and perhaps more as a life-long care giver rather than a partner for procreation.

Consider conditions with an abnormal number of sex chromosomes with primary infertility such as females with the Turner syndrome (45,XO karyotype) or males with the Klinefelter syndrome (47,XXY). In individuals in whom these conditions have been detected pre-maritally, seldom do families reveal these findings to a future spouse or his or her family in an arranged marriage. In a society where marriages are universal and bearing children is the natural consequence of marriage, there is genuine fear of the marriage proposal being rejected at the outset if the ability to bear children is in doubt.

Similar apprehensions are seen among individuals with inherited skin disorders. Genetic conditions such as neurofibromatosis type 1, oculocutaneous albinism and lamellar ichthyosis are usually seen in individuals with normal intellectual and physical capabilities. A commonly held perception with lamellar ichthyosis (autosomal recessive), a condition with patchy scaling of the skin, is '*naga-dosha*' or that the evil influence of a malevolent snake is on the individual. It is often very difficult for individuals with these conditions to find a spouse through an arranged relationship despite appropriate genetic counselling. So ingrained is the belief that individuals with congenital anomalies or genetic disorders are less than perfect, that they seldom find spouses who do not themselves have a disorder. Societal intolerance also forces individuals, especially wives, to conceal their 'defects'. A child was seen in clinic with deafness, cutaneous albinism and a white forelock of hair, indicating a diagnosis of the Waardenburg Syndrome. As this is an autosomal dominant (AD) condition, the parents were examined carefully and found to be normal. The mother later returned to the clinic to reveal what her husband was unaware of even after eight years of marriage, that she wore dark coloured contact lenses in one eye to conceal heterochromia irides (different coloured irides), the mildest manifestation of Waardenburg syndrome. The mother of another child diagnosed with AD oculocutaneous albinism (Goldenhar) syndrome had cleverly concealed from her husband and his parents her dysplastic ear with her hairstyle for over a decade!

Marriage patterns contribute to the incidence of genetic disorders in the population. In the southern states of India, consanguinity is common (Bittles *et al.* 1988, 2010). Among Hindus it is usually uncle-niece marriages, with a girl marrying her maternal uncle (where one exists), and among Muslims it is first cousin marriages. Such consanguineous unions are often multi-generational, posing a challenge to the counsellor attempting pedigree delineation in the clinic. Predictably, such inbreeding leads to an increased prevalence of

autosomal recessive (AR) disorders. Education and social class appear to make little difference to this practice, with increasing numbers of young urban professionals seeking counselling because they are 'married to a cousin'. Since beta thalassemia is the commonest recessive disorder in India, with a carrier frequency ranging from 1–4% (Verma *et al.* 2011; Mohanty *et al.* 2013), this counsellor suggests beta-thalassemia testing for all healthy consanguineous couples.

Another serious problem is genital ambiguity at birth. Genital ambiguity can be caused by recessive disorders, the commonest of these being congenital adrenal hyperplasia (CAH). CAH in a female infant can cause virilization of the external genitalia, occasionally requiring corrective surgery later in life. Investigations reveal a normal female karyotype and internal genitalia with elevated 17-hydroxyprogesterone levels. Steroid replacement for life ensures that these women can function as normal females with regard to menarche and pregnancy. Shockingly, however, some parents choose to discontinue therapy, leading to near certain death of the affected female, rather than deal with the long term issues posed by virilization. Such deliberate neglect is sometimes due to the unfounded fear that these women may end up as '*hijras*' (trans-genders adopting female appearance and attire).

In the case of XY individuals with a female phenotype, most often due to an androgen receptor mutation, sensitivity and care are required in counselling to protect confidentiality and the individual's privacy. The emotional impact of such a diagnosis on the affected individual is enormous and long-lasting and psychological support is therefore required for extended periods of time. In recent times, there have been sensational media reports about the gender and sexual orientation of some athletes and 'holy-men'. It is appalling to see medical personnel revealing details of the genetic results and genital abnormalities to the media with little concern about the impact of disclosing such information in public on the lives of these individuals (IndiaToday Online 2012).

Preference for a male child manifests itself in many ways. When the counsellor is preparing a pedigree chart and asks about the number of sibs, many individuals enumerate only male children. One family with several affected pregnancies chose to give birth to a severely affected offspring detected in the second trimester because antenatal scanning elsewhere had revealed it to be a male. This baby died in the newborn period from medical complications.

A group of recessive conditions, rare elsewhere but relatively common in southern India, are the lysosomal storage disorders (LSDs). Of the nearly 50 known LSDs treatment in the form of enzyme replacement therapy is available for seven (Beck 2010; Hollak and Wijburg 2014). Replacement enzymes are transfused weekly or fortnightly, costing between 30 and 50 lakh rupees a year for a child weighing ten kilograms. Unlike in Western societies the cost of this therapy is neither covered by the state nor by private health insurance schemes. In the few situations where parents are

able to pay for therapy and have more than one affected child, the affected male child is invariably treated in preference to an affected girl child. Such choices are often made in a spectrum of conditions where therapy is available but expensive.

Apart from the direct cost of therapy, practical issues such as schooling and securing jobs are also of constant concern to the parent. For children born with cranio-facial syndromes, the facial dysmorphology is sometimes worse than their intellectual disability. Many schools are reluctant to admit such children because of their appearance. The counsellor often has to write to educational authorities in support of the child's admission to the school. An adult with Crouzon syndrome with normal intellect had to seek legal redress as her application for a driving license was turned down due to the fact that 'she looked abnormal.' For children with progressive muscle weakness such as in Duchenne muscular dystrophy, the inability to climb upstairs usually manifests at the end of the first decade. The vast majority of 'normal' schools have middle and senior classes on the first floor and above, without any lifts or other climbing aids available on site. Children with glycogen storage disorders requiring feeding at frequent intervals to avoid hypoglycaemic seizures require multiple negotiations with school authorities to secure permission to do so.

Nowhere is genetic counselling as crucial as with prenatal diagnosis. In India, when a family is seen in the fetal medicine clinic, usually for an anomaly detected in a pregnancy scan or because of prior history of a genetic disorder, a number of religious, social and emotional factors are at play. The diagnostic options available to the couple, the decision to have a prenatal test, the implications of finding an adverse result and the long-term impact of a genetic disorder in the family, all need to be discussed at length and in a manner that the family is able to make their own choices. The couple seeking advice is often accompanied by a number of family members, usually parents or in-laws, who are not only a part of the counselling session but play a key role in the decision the couple will make for intervention in pregnancy. Mothers are often blamed or made to feel guilty about any disorder identified in pregnancy. This makes counselling in X-linked disorders such as Duchenne muscular dystrophy and haemophilia especially tricky (Shetty *et al.* 2001). In extreme situations the wife may be abandoned or left to her own resources to deal with the consequences of an affected pregnancy. Therefore, while collecting blood for carrier testing in X-linked disorders it is useful to collect a paternal sample simultaneously to avoid implicating only the mother.

In recessive disorders, when a couple understands the concept of both parents being carriers and a recurrence of 25% in a subsequent pregnancy, there have been instances when the husband has returned later with a 'second wife' for the sole purpose of having a normal child! One educated

couple, whose first child was diagnosed with Morquio syndrome, a rare lysosomal disorder, went a step further. The wife returned for a scan in her next pregnancy which was fathered by the husband's close friend in order to ensure a healthy child from conception. The counsellor can only take consolation in the realization that the principles of AR inheritance were well understood. As a consequence of such outcomes, many learned colleagues believe that non-directive genetic counselling, where the choices of testing and treatment are discussed with the family and decisions are made solely by the family members themselves, is not often feasible in our society.

Religion and social beliefs play a large role in determining a family's perception of the birth defect and its causation. A baby born with a huge protruding tongue as a result of Beckwith-Wiedemann syndrome had been reverentially named 'Durga' by her parents. Children with extra limbs are also believed to be incarnates of Hindu deities. Multiple recurrences of serious disorders are accepted with a resigned belief that it is a consequence of bad 'karma'. Birth defects are often believed to result from the expectant mother venturing out during an eclipse. Even now, branding the child with a hot object is not uncommon in rural southern India. Branding is usually done on the abdomen (for abdominal distension) and at the nape of the neck, and is usually performed by an elderly female family member (Rathi and Gupta 2015). Burning of eyelashes with a lit incense stick is also believed to ward off evil spirits. In northern India, potions containing cannabinoids are fed to infants to lull them to sleep, leading to pin-point pupils and chronic constipation. Eyes lined with kohl and dark spots on cheeks, palms and soles are commonly seen in infants to keep evil spirits at bay. Such deeply ingrained beliefs and customs contribute to the clinical profile and it is necessary to take them into account while counselling. With time, patience, and repeated meetings, explaining genetic concepts with everyday examples and with an appreciation of local sensibilities, it is possible to communicate with the families effectively and sensitively. For the counsellor too, understanding the family's beliefs and the reasoning that goes into decision-making are a continuous source of learning.

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