REVIEW CLINICAL PRACTICE

Genetic Counseling for Obstetricians

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ABSTRACT

Today many chromosomal defects can be suspected and diagnosed by the currently available biochemical testing and invasive tests analysis of chromosomes.

It is necessary for the clinician to have a through knowledge of chromosomal defects so a proper genetic counseling can be done. This review article discusses the genetic counseling for the various defects which can occur in the unborn fetus.

Keywords: Genetic counseling, Chromosomal disorders, Prenatal diagnosis, Gene defects, Biochemical tests.

Clinical genetics is one of the most rapidly advancing fields in medicine. Spectacular progress has been achieved in this century with unraveling of the entire draft sequence of the human genome and identification of greater than 14,000 single gene disorders. A major contribution of these advances has been in diagnosis, management and prenatal diagnosis of genetic disorders, as the treatment in most cases is difficult or impossible, and where available beyond the means of most families. With increase in the knowledge about genetic disorders, genetic counseling has become an integral part of management of any disorder with genetic or probable genetic etiology. Counseling should be undertaken by a physician with proper understanding of genetic mechanisms.

GENETIC COUNSELING[1-3]

An accurate diagnosis of the disorder is very essential for any genetic counseling. It is defined as "the process by which patients or relatives at risk of a disorder are advised of the consequences of the disorder, the probability of developing and transmitting it, and ways in which this can be ameliorated." The objective of genetic counseling is to make the individual or the family understand the scientific information about the disease, thus helping the individual or family to choose a course of action which seems appropriate to them in view of their risks, their family goals, and their ethical and religious standards. Further, it should act in accordance with the decision, and also to make the best possible adjustments to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

Hence, the main elements of genetic counseling can be summarized as:

- Diagnostic and clinical aspects
- Documentation of family and pedigree information
- Recognition of inheritance patterns and risk estimation
- Communication and empathy
- Information on available options
- Support in decision making.

It also aims at providing social and psychological support to the affected family by the removal or lessening of guilt or anxiety and helping the individuals/couples achieve their parenting goals.

Nondirectiveness in Genetic Counseling

The main principle of genetic counseling is nondirectiveness, which is the art of presenting facts without influencing decision. It promotes the autonomy or self-determination and personal control of the client. To maintain the sense of psychological well-being amongst the clients, genetic counseling has also been defined as a dynamic psychoeducational process centered on genetic information. The goal is to facilitate client's ability to use genetic information in a personally meaningful way that minimizes psychological distress and increases personal control. It promotes understanding, decision making, personal control, adaptation to stress inducing events, and reduces psychological distress.

Psychological Issues

The diagnosis of any significant medical condition in a child or adult may have psychological, financial and social implications, but if the condition has a genetic basis, a number of additional issues arise. These include guilt and blame, the impact on future reproductive decisions and the genetic implications to the extended family.

Guilt and Blame

Feelings of guilt arise in relation to a genetic diagnosis in the family in many different situations. Parents very often express guilt at having transmitted a genetic disorder to their children, even when they had no previous knowledge of the risk. On the

other hand, parents may also feel guilty for having taken the decision to terminate an affected pregnancy. Healthy members of a family may feel guilty that they have been more fortunate than their affected relatives, and at-risk individuals may feel guilty about imposing a burden onto their partner and partner's family. Although in most situations where the person expressing guilt has played no objective causal role, it is important to allow him or her to express these concerns and for the counselor to reinforce that this is a normal human reaction to the predicament. Blame can sometimes occur in families where only one member of a couple carries the genetic risk (it wasn't *our* side), but again this is less likely to occur when the genetic situation has been explained and understood.

Reproductive Decision Making

Couples aware of an increased genetic risk to their offspring must decide whether or not this knowledge will affect their plans for a family. Some couples may be faced with a perplexing range of options including different methods of prenatal diagnosis and the use of assisted reproductive technologies. For others the only available option will be to choose between taking the risk of having an affected child and remaining childless. Couples may need to reconsider these choices on repeated occasions during their reproductive years. Decision making may be more difficult in particular circumstances, including marital disagreement, religious or cultural conflict, and situations where the prognosis for an affected child is uncertain. For many genetic disorders with variable severity, although prenatal diagnosis can be offered, the clinical prognosis for the fetus cannot be predicted. When considering reproductive decisions, it can also be difficult for a couple to reconcile their love for an affected child or family member, with a desire to prevent the birth of a further affected child.

Impact on the Extended Family

The implications of a genetic diagnosis usually reverberate well beyond the affected individual and his or her nuclear family. For example, the parents of a boy just diagnosed with Duchenne muscular dystrophy will not only be coming to terms with his anticipated physical deterioration, but may have concerns that a younger son could be affected and that daughters could be carriers. They also face the need to discuss the possible family implications with the mother's sisters and female cousins who may already be having their own children.

Bereavement

Bereavement issues arise frequently in genetic counseling sessions. These may pertain to losses that have occurred recently or in the past. A genetic disorder may lead to reproductive loss or death of a close family member. The grief experienced after termination of pregnancy following diagnosis of abnormality is like that of other bereavement reactions and may be made more intense by parents' feelings of guilt. After the birth of a baby with congenital malformations, parents mourn the loss of the imagined healthy child in addition to their sadness about their child's disabilities, and this chronic sorrow may be ongoing throughout the affected child's life.

Long-term Support and Follow-up

Many families will require ongoing information and support following the initial genetic counseling session, whether coping with an actual diagnosis or the continued risk of a genetic disorder. Follow-up sessions may be needed to reinforce the informations (usually forgotten or wrongly remembered by the consultants), answer new queries, provide latest information and provide psychological support to the family during the process of coping up, till the acceptance and adjustments take place.

Indications for Genetic Counseling

There are certain situations which can be identified before or after conception, in which genetic counseling and prenatal diagnosis may be required. These indications are:

- Advanced maternal age (> 35 years)
- Recurrent miscarriages (3 or more)/infertility/primary amenorrhia
- Previous child with
  - Dysmorphism/single or multiple malformations like cardiac, renal, brain defects/short stature/neuromuscular disorder/neurogenetic disorder/metabolic disorder/unexplained MR/cerebral palsy/autism/chromosomal abnormality/deafness/thalassemia/hemophilia.
- Previous unexplained still birth/s, neonatal or infantile deaths with or without congenital malformations
- Family history of a genetic disorder or any chromosomal abnormality like Down's syndrome, thalassemia, spinal muscular atrophy, hemophilia, congenital deafness or Gaucher's disease
- Consanguinity, especially with a history of suspected genetic disorder
- Maternal diseases like diabetes, hypothyroidism to identify high-risk fetuses through level II ultrasound
- Positive maternal serum screen either first or second trimester/abnormal fetal ultrasound
- Exposure to known or suspected teratogen during pregnancy
- Amniotic fluid abnormalities in second/third trimester especially in association with growth retardation
- Maternal infection (TORCH infection).

Steps in an Antenatal Case Management[1,2]

The skills required to make a genetic diagnosis are similar to those used for more common health problems, including history taking, physical examination, and proper laboratory testing. The practice of referring high-risk obstetric patients for genetic counseling improves the detection of identifiable genetic risk factors.[4]

History

The pregnancy history of the patient's mother might disclose maternal disease potentially causative of or related to the fetal condition, as seen in certain metabolic disorders, such as untreated maternal PKU or fatty acid oxidation disorders. Sometimes, maternal disorders (diabetes) and environmental or drug exposures (valproate, warfarin, etc.) during pregnancy can cause multiple malformations such as in fetal valproate syndrome or warfarin embryopathy. Medical history of maternal disorders like SLE, hypothyroidism is also important for better fetal outcome.

Family History

A thorough family history includes detailed information on relatives' ages, current and past medical health (including developmental or learning problems), birth defects, obvious dysmorphism, and surgeries. Specifically, questions about miscarriages, stillbirths, infant deaths and infertility should be asked. For deceased family members, age and cause of death should be documented. The racial and ethnic background is of importance in identifying higher risk groups. In addition, the possibility of consanguinity in the family history should be explored when clinically relevant. Drawing a family tree (pedigree) that symbolically (Fig. 1) represents the family and demonstrates a relationship between affected family members is an efficient and highly informative exercise. A three-generation pedigree should be constructed using the below mentioned symbols. The proband is the individual through which the family is ascertained. Large families will commonly have several proband. The person who seeks genetic counseling is called the consultand or counselee and the one who gives it is the counselor. In addition to medical specialists, trained persons with various backgrounds like nursing, social work education and psychology can function as genetic counselors.

History of any Genetic Disorders in the Family

Family photographs or medical records may be of help, particularly if other family members are suspected to have same genetic disorder.

Examination of the couple is required especially when there is a family history of a particular genetic disorder like neurofibromatosis, tuberous sclerosis, or incontinentia pigmenti.

Specialized Investigation

The importance of precise diagnosis for genetic counseling cannot be overemphasized. However, specialized tests like chromosomal analysis, enzyme analysis, and DNA analysis are required to arrive at a final diagnosis. Before these tests are ordered, information should be obtained on the type and volume of the specimen required (blood, urine, fibroblasts, amniocytes), type of tube in which the specimen should be kept, and conditions under which the specimen should be sent.

DNA Based Tests (Molecular Tests)

DNA testing investigates alterations in a gene that result in disease. Confirmed molecular diagnosis in index case would also help in carrying out prenatal diagnosis (by amniocentesis or chorionic villi sampling) for the respective disorder. Unless the type of mutation(s) in the proband or carrier parents is identified, prenatal diagnosis is not feasible. It should preferably be identified before next pregnancy. Examples of widely available molecular genetic tests include thalassemias, muscular dystrophies, spinal muscle atrophy, fragile X syndrome, hemophilia A and B, cystic fibrosis, albinism, achondroplasia, etc.

Chromosomal Analysis (Cytogenetics)

Chromosomal abnormalities can be diagnosed after birth using a blood test, or before birth using prenatal tests (amniocentesis

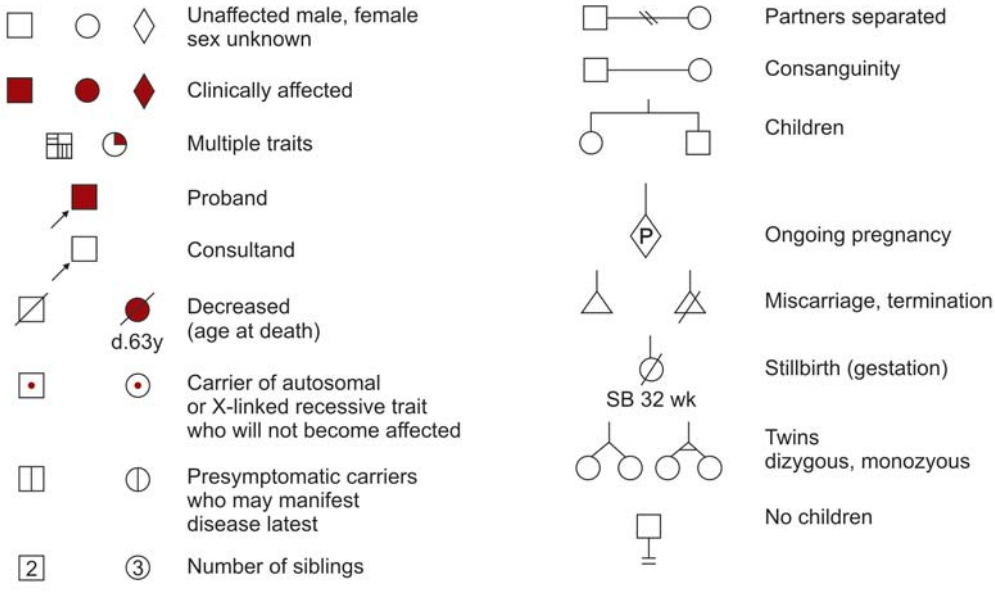


Fig. 1: The main symbols used in constructing a pedigree

or chorionic villi sampling). Tissues most commonly used are lymphocytes and amniocytes. Any abnormal finding has its own implication and management. Cytogenetic analysis on bone marrow also helps in diagnosis and prognosticating, especially in cancers. It takes on average 1 to 3 weeks to obtain a definitive result, the time depending on the method.

Newer diagnostic techniques include:

- Rapid-FISH (Rapid Fluorescence *in situ* Hybridization)
- MLPA (Multiple Ligation PCR Amplification)
- QF-PCR (Quantitative Fluorescent Polymerase Chain Reaction).

These methods of analysis do not require culturing, the amount of the sample material may be very small and the result is obtained in just few days. In comparison, classical cytogenetic analysis (karyotyping) after amniocentesis requires 15 to 20 ml of amniotic fluid, culturing of fetal cells (amniocytes) and takes around 10 to 21 days to produce the result.

Biochemical Testing

Biochemical testing refers to analyses of metabolites that are either the substrates or the products of a deficient enzyme. Thus, increase or decrease of metabolite concentrations are indirect indicators of metabolic disorders caused by an enzyme deficiency. If abnormal metabolites are identified, the disease may then be confirmed by enzyme analysis when available like in mucopolysaccharidoses, Gaucher's disease, Tay-Sachs disease, etc. Enzyme analysis often requires a fibroblast culture or a fresh liver biopsy. Some enzyme tests can be done on serum, red blood cells, or leukocytes.

GENETIC COUNSELING FOR VARIOUS GENETIC DISORDERS

Chromosomal Disorders

Chromosomal disorders are due to changes in the number or structure of chromosome from the normal state that results in a gain or loss of genetic material, such as Down's syndrome or Turner syndrome. These abnormalities account for approximately 6% of birth defects in industrialized countries. Majority of chromosomal disorders (Down's syndrome, Edward's syndrome [Trisomy18], Patau's syndrome [Trisomy 13]) have associated mental retardation. Of these, Down's syndrome is the commonest cause of mental retardation with a prevalence of about 1 in every 800 to 1,000 births, majority being due to an extra chromosome 21. In all *de novo* numerical *(Trisomic Down's syndrome and other autosomal trisomies)* and structural chromosomal abnormalities have a low risk of recurrence (< 1%) with the exception of very rare families possibly because of predisposition to nondisjunction or gonadal mosaicism. In practice, most couples with previous affected child do elect for prenatal diagnosis in subsequent pregnancy with full knowledge of procedural risks and low probability of recurrence. For other relatives of a child with isolated trisomic Down's syndrome, there is no evidence of an increased risk in their offspring.

Translocation Down's syndrome comprises only 5% of all cases of Down's syndrome and the recurrence risk is higher if either of the parents is carrying a balanced translocation. The risks for the offspring of balanced carriers vary from 2.5 to 10% for father and mother as a carrier, respectively for 14/21 and 21/22 translocation and 100% for 21/21 translocation. So precise type can be established from chromosomal analysis of index case, and the relatives at risk should be investigated using blood before pregnancy occurs. Where the parents of a child with translocation Down's syndrome are both chromosomally normal, the risk of further affected child is also low, probably similar to that trisomic Down's and other autosomal trisomies, i.e. < 1 percent.

Advanced maternal age carries an increased risk for Down's syndrome with a lesser risk of other trisomies. As a woman's age (maternal age) increases, the risk of having a Down's syndrome baby increases significantly and varies from 1:1925 at age 20, 1:885 at 30; 1:365 at 35; 1:110 at 40 to more than 1:50 at > 45. There is general agreement that all women aged 35 years or above should be informed of the risks of Down's syndrome and offered amniocentesis, however uptake of amniocentesis varies greatly between countries and region, and has been considerably affected by the development of second trimester maternal serum screening.

Noninvasive prenatal screening (1st and/or 2nd trimester) for common chromosomal disorders has good sensitivity using maternal serum biochemical markers and ultrasonography (discussed in other section). Definitive diagnosis can be provided by chromosomal studies on amniotic fluid, chorionic villus biopsy or cord blood sample. Knowledge of the loss rate of affected fetus is important for counseling prospective parents and for assessing the available screening methods at varying times of gestation. Before offering any kind of genetic testing, pre- and post-test counseling remains the biggest challenge and should always be done religiously and cautiously. The most important points to emphasize are—a positive test does not mean that the fetus has Down's syndrome and a negative test does not rule out Down's syndrome. The screening tests are done to relieve the anxiety, but in most situations, if done without counseling can make the couple very anxious. In India, as the tests have been introduced recently and pretest and post-test counseling is inadequate, it creates more confusion than solving the problems in many situations in authors' experience. It is imperative that the clinicians ordering the test should clearly understand the interpretation.

Presence of multiple abnormalities also raises the risk of any chromosomal abnormality to 35% (Table 1) and counseling should be individualized.

There is little point performing chromosomal analysis on patients with single congenital malformation, single gene disorders or with recognizable nonchromosomal syndrome. However, a chromosomal analysis of a clinically obvious Down's syndrome child is extremely important because depending on the type of chromosomal anomaly, the risk of recurrence may vary from 1 to 100% as mentioned above.

Table 1: Aneuploidy risk with major structural fetal malformations

Malformation	*Aneuploidy risk*
Cystic hygroma	60-75%
Hydrops	30-80%
Hydrocephalus	3-8%
Holoprosencephaly	40-60%
Cardiac defects	5-30%
Diaphragmatic hernia	20-25%
Omphalocele	30-40%
Gastroschisis	Non-minimal
Duodenal atresia	20-30%
Facial cleft	1%
Bladder outlet obstruction	20-25%
Limb reduction	8%
Clubfoot	20-30%
Single umbilical artery	Minimal

Multifactorially Inherited Disorders

Multifactorially inherited disorders account for 20 to 30% of all birth defects, a number of which are lethal. Examples of multifactorial birth defects are numerous, are usually malformations of a single organ system or limb, and include congenital heart disease, neural tube defects, cleft lip and cleft palate, clubfoot and developmental dysplasia of the hip.

i. *Antenatal screening for neural tube defect*: The prevalence of NTDs in different parts of India varies from 0.5 to 11 per 1000 live births. Most cases are straightforward and involve an isolated NTD, either anencephaly or spina bifida, or both. Care is needed to ensure that one is dealing with a primary NTD or NTD as a part of syndrome, teratogenic exposure or chromosomal anomaly. Vertebral anomalies and hydronephrosis are commonly seen in isolated NTDs. It can be uniformly fatal as in anencephaly, iniencephaly, acrania, total craniospinal rachisis or prognosis may be variable depending upon type of defect.

   a. *Maternal serum screening:* Maternal serum AFP levels are measured at 16 to 18 weeks of gestation. A cut-off > 2.5 MoM detects > 90% cases of anencephaly and 80% cases of spina bifida cystica. Though the specificity of the test is not very high, being increased in abortion, twin pregnancy, exomphalos, etc., yet it has been implemented widely and led to a striking decline in the incidence of open NTD.

   b. *Ultrasound screening:* Anencephaly is detectable at 10 to 12 weeks of gestation; spina bifida is detectable at 16 weeks onwards; large defects may be visible earlier. Prevention—Folic acid supplementation (4 mg per day) started two months before to three months after conception (periconceptional) prevents recurrence in about 72% of cases. Primary prevention (about 50%) by the use of 0.4 mg of folic acid periconceptionally has also been recommended.

Table 2: Empiric risk of recurrence of isolated malformation

Malformation	*Frequency per 1000 births*	*Recurrence for normal parents of one affected child*
Anencephaly/spina bifida	4-5	5 %
Cardiac malformation	6-8	3-4 %
Cleft lip and cleft palate	2	4-5 %
Cleft palate alone	0.5	2-6 %
Pyloric stenosis	2-3	3 %
Talipes equinovarus	3-4	2-8 %
Dislocation of hip	3-4	3-4 %
Hirschsprung's disease	0.1	6 %

ii. *Malformations with uncertain prognosis*: It is a challenge to the counselor as well as a dilemma for the family. Some of the malformations with uncertain prognosis are ventriculomegaly, meningocele, omphalocele, multicystic kidneys, hydronephrosis, urinary bladder obstruction, cystic adenomatoid lung malformation, diaphragmatic hernia, cardiac defects, cyst/calcification inside abdomen, mild limb shortening. Counseling and risk of recurrence in these disorders is based on empiric risk figures, and many anatomic anomalies indicate a heritable tendency. Prenatal diagnosis is possible in some defects using fetal ultrasonography. After diagnosis of any malformation on fetal anomaly scan, one should apply the knowledge of various genetic disorders and syndromes to provide appropriate nondirective genetic counseling and management. Table 2 shows risk of recurrence of some common malformations. Counseling in multifactorial disorders is to be done carefully, as there is a close overlap between hereditary and nonhereditary disorders. Prenatal diagnosis of major malformation is possible and the risk prediction can be calculated more accurately. There are guidelines which can help the at-risk families to plan the family or can be of great help in premarriage counseling.

Single Gene Defects

Single gene defects are caused by alterations in gene structure called mutations, which result in abnormal gene functioning. All single gene defects combined account for an estimated 7.5% of birth defects in industrialized countries. The usual patterns of transmission are autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, and Y-linked. Over half are inherited in an autosomal dominant fashion, about one-third as recessive and one-tenth as X-linked. Recurrence risk for the next pregnancy varies depending upon the inheritance pattern.

For most of the common genetic single gene disorders, either enzyme based or molecular diagnosis is available though facilities for diagnosis of all disorders are not available in our country. Prenatal diagnosis is possible by using the same techniques on fetal tissues—chorionic villus biopsy or amniotic fluid.

X-Linked Disorders

In pregnancies at high risk for X-linked disorder, fetal sexing offers the possibility of determining whether or not the fetus is indeed at risk.[2] However, in most disorders direct molecular prenatal diagnosis of an affected male is now possible, and even where it is not, linked DNA markers can often be used. First trimester fetal sexing using both DNA and cytogenetic methods is now feasible with CVS. Where a woman is only a possible carrier, it is vital to estimate the risk before fetal sexing is undertaken and to use methods of carrier detection, where applicable. The most successful approach to prenatal diagnosis is where no prenatal procedure is required at all because the carrier state is excluded. As far as possible, this should be approached as a planned procedure.

Autosomal Recessive Disorders

The disease usually occurs in one generation only. The risk of occurrence and recurrence is 25% in such cases (Fig. 2). Identification of mutation in such couples or proband is the prerequisite for prenatal diagnosis. So, to have an affected child both parents need to be carriers and prenatal diagnosis is also indicated in this situation. There is no risk of having an affected child if only one partner is a carrier or is affected (Figs 3 and 4). If only one partner is a carrier, there is a 50% chance of the baby being a carrier and all babies will be carriers if one partner is affected with an autosomal recessive hemoglobinopathy. Identification of carrier status is easy and possible by measurement of HbA2, which is almost always high in carriers except in a rare situation of silent carrier status when HbA2 may be normal and carrier status can only be confirmed by molecular studies. The risk of recurrence should be explained, if necessary with the help of diagrams. The risk, for example, 1 in 4 should be explained in both ways, i.e. 1 in 4 and 25%. It should be made clear that there may be 2 or 3 or more consecutively affected children in a family as chance does not have memory. The probability can be explained by an example of tossing a coin. It is often useful to compare this recurrence risk against the general population risk for the condition and for common birth defects (See Table 2). It should be stressed that any family can have children affected with genetic diseases or congenital malformations and parenting such a child or carrier status for genetic disease is not a stigma.

Autosomal Dominant Disorders

Here the risk of recurrence for the *sib is* 50% if one of the parents is affected. If it is a sporadic case (new mutation), risk of recurrence in *sib* is very low but gonadal mosaicism cannot be ruled out. In X-linked recessive disorders, risk of recurrence for boys is 50% whereas females usually do not manifest.

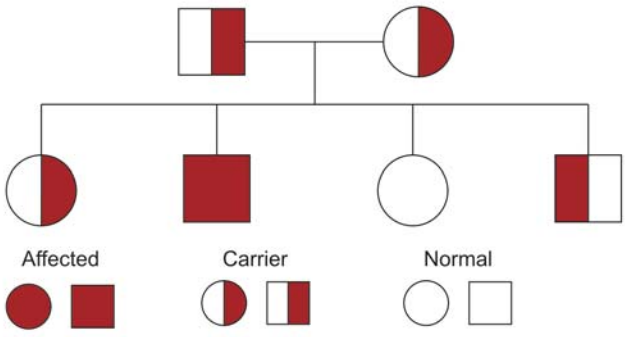


Fig. 2: 25% risk of affected child if both parents are carrier

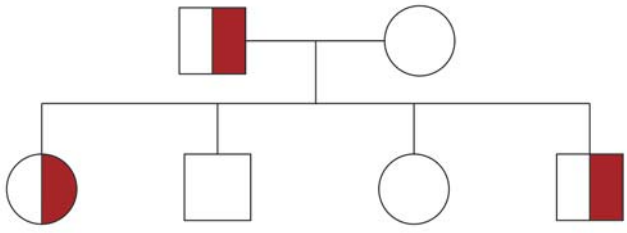


Fig. 3: No risk of affected child if only one parent is carrier

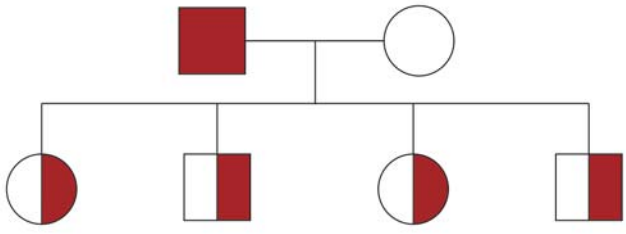
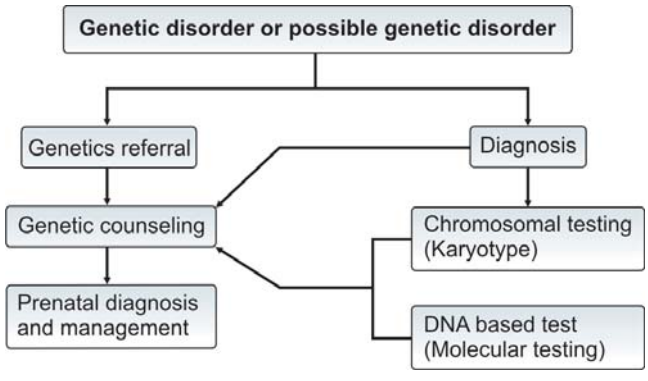


Fig. 4: No risk of affected child if one parent is affected

Role of Obstetricians in Management of Patients with Genetic Disorder and Antenatal Detection of Malformation



Special Problems in Genetic Counseling

In some of the situations, especially known autosomal recessive and dominant disorders, counseling and presentation of risks is relatively straightforward based on the principles of Mendelian

inheritance. However, in the presence of reduced penetrance, delayed onset, and genetic heterogeneity (a disorder caused by more than one genetic mechanism) extending into different modes of inheritance, the counseling becomes more difficult. There are several situations that can pose problems while counseling.

- *Consanguinity*: Relationship between blood relatives who have at least one common ancestor no more remote than a great-great grand parent. In Indian scenario, uncle niece relationship (second degree) is the commonest. The absolute risk of abnormal offspring (stillbirth, neonatal death and congenital malformation) for marriages between first cousins is 3 to 5%, about double the overall background risk of 2 to 3% for offspring of unrelated individuals. Similarly, the probability that first cousins will have a child with an autosomal recessive disorder is approximately 3%, although the risk can be greater if there is a family history of a specific genetic disorder.
- *Disputed Paternity*: Difficulty in genetic counseling may arise if the socially accepted father is not the real biologic father.
- Problems with chromosomal prenatal diagnosis.

Mosaicism: Its likely consequences and severity is often difficult to interpret especially in CVS samples, where mosaicism is often confined to the placenta.

Culture Failure

Unexpected or unrelated adverse findings: Occasionally, prenatal chromosomal analysis performed to rule out aneuploidy may reveal a common variant, a rare rearrangement or a marker chromosome. As its significance in fetus is not known, parental chromosomal analysis should be done to rule out a *de novo* or inherited abnormality. Unbalanced or *de novo* structural rearrangements may cause serious fetal abnormalities.

KEY POINTS TO REMEMBER

1. Hereditary diseases may manifest at the time of birth or several years later in life.
2. All congenital defects observed at the time of birth are not necessarily inherited. Some of these may be due to teratogenic effect of drugs, infections or irradiation during the first trimester of pregnancy.
3. A degree of clinical variability exists in the presentation of certain genetic disorders. This variable expression of the mutant gene is attributed to the degree of penetration of the gene. Thus, one member of the family may show all the features of a genetic disease, while his or her siblings may show only mild forms of the disorders with one or the other sign.
4. Genetic counselor should interpret the anticipated risk of recurrence of the inherited disorder in the future siblings in a meaningful manner, so that the family can arrive at a rational decision. The counselor has a particularly important responsibility in reassuring the parents that the risk of recurrence is low in case of disorders with multifactorial inheritance. In sporadic mutations and most of chromosomal disorders, there is only a small or no risk of recurrence.
5. While conveying information to the parents, the physician should be extremely cautions and should take special care not to infuse a sense of guilt in the parents. In case of X-linked disorders, it will be desirable to temper the blame on the mother, lest she is castigated by her husband or in-laws (Indian scenario).

CONCLUSION

Genetic counseling in association with modern prenatal diagnostic procedures constitutes a basic element of prevention of congenital anomalies and genetic disorders. Pediatricians and gynecologists are the primary physicians for the diagnosis, and management of children and high-risk couples with genetic disorders. They should make the parents or couple aware of the genetic disorder, risk of recurrence, prognosis, and prenatal diagnosis.

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