of rabbit β **globin gene** to mouse cells which were TK^+ and were, therefore, earlier selected for their transforming ability. In most cases the foreign DNA in these cases gets integrated into mammalian chromosomes. This transformation can rarely be brought about in the fertilized mouse eggs also, and it was shown that rabbit β globin was produced by the mouse developed from transformed egg and that the rabbit β globin gene thus introduced is inherited to the next generation.

Use of Human Genetics in Medical Science

If knowledge about genetics of human diseases is available, it can be used in a variety of ways to avoid or reduce the incidence of some of these diseases. This can be achieved in a variety of ways and we will describe five of them, namely (i) genetic counselling, (ii) antenatal diagnosis, (iii) gene therapy, (iv) making choice of baby's sex, and (v) DNA fingerprinting in forensic science.

Genetic counselling

Genetic counselling for couples who believe that there may be a risk of producing a defective child, has now become routine aspect of medical practice, particularly in the developed countries. These parents may either voluntarily abstain from having any child or may undergo selective abortions on suspicion or after ascertaining it through antenatal diagnosis.

A genetic counsellor should first be able to identify the disease and therefore should be first a clinician and then a geneticist. The simplest cases asking for genetic counselling will be those having a family history of disease and the parents may like to know the chances of having a child free of that disease. A couple may have one defective child and would like to know the chances of having a normal child on the next pregnancy. In such a case, if the defect is known to be single gene recessive and both parents are normal, the chance is three in four of having a normal child, although even a normal child will have a two third chance of being a carrier. The parents may like to give birth to such a child who may be a carrier, because the chance of his or her spouse also being a carrier will be remote. However, in such cases, even the possibility of having the defective grand child can be worked out, if the frequency of heterozygotes in the population available for marriage of the child is known. For instance, in case of **fibrocystic disease (cystic fibrosis)** of the pancreas, the frequency in general population (perhaps in UK) is 1/22. The chance of the normal child to be a carrier being 2/3 and the chance of spouse to be a carrier being 1/22, the chance of the grand child to be defective would be $2/3 \times 1/22 \times 1/4 = 1/132$ (since 1/4 is the chance that a child born to both heterozygote parents will be defective). A risk of one in 132 may or may not be worth taking depending upon temperament and circumstances. More detailed calculation can be done in these simple cases and also in cases of polygenic nature and variable penetrance. Readers are advised to consult a book on human genetics for further details.

In some cases, detection of heterozygote, may also be useful and possible. Following are the situations where it is possible. (i) When a heterozygote, though phenotypically normal, produces a particular enzyme activity intermediate to those found in two homozygotes, its presence can be detected using electrophoresis. In such cases, if a biochemistry laboratory is available, deficiency like **HGPRT (Lesch-Nyhan syndrome)** in heterozygous condition can be detected from a blood sample or some skin cells. (ii) When the mutant produced an altered form of gene product, the heterozygote may produce two different forms of protein that can be separated by electrophoresis to enable the identification of a heterozygote. (iii) If the defect is associated with a chromosome structure, then with the availability of a cytogenetics laboratory, such an abnormality in heterozygous condition can be identified.

Amniocentesis and antenatal diagnosis

When a pregnant woman is known to have a chance of bearing a child with a genetic defect, it may be desirable to diagnose the condition in the fetus. This can be done by taking some cells from the fetus by drawing a few millilitres of amniotic fluid with the help of a hypodermic needle; The technique is called **amniocentesis** (Fig. 24.8) and is usually performed at 15th week of pregnancy, to allow enough time for safe abortion if recommended. The amniotic fluid has free cells of fetal origin, which can be cultured and tested in various ways e.g. karyotype, enzyme production and restriction site pattern analysis of its DNA. At least 35 diseases which can be identified by this technique are known. If disease is detected through