9. GENETIC TESTING

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Genetic testing is defined as the use of specific assays to determine the genetic status of individuals already suspected to be a high risk for a particular inherited condition. The terms genetic test, genetic assay and genetic analysis are used interchangeably to mean the actual laboratory examination of samples. The term genetic screening is used when genetic testing is done on a target population with latent, early or asymptomatic disease.

Areas of focus in genetic testing include prenatal diagnosis, newborn screening, carrier screening, forensic screening and susceptibility screening. Genetic profiling was established in Malaysia in 1994. The service is provided by the Government Chemistry Department, Kuala Lumpur. The laboratory is dedicated to excellence in service and provides the most accurate and powerful proven DNA technology. Chromosomal analysis to detect congenital abnormalities is performed at the Institute of Medical Research (IMR), National Family Planning Board and all the laboratories in government University Hospitals in Kuala Lumpur.

Prenatal diagnosis discerns whether a foetus is at risk for various identifiable genetic diseases or traits. Prenatal diagnosis is carried out using amniotic fluid, foetal cells, foetal or maternal blood cells obtained during amniocentesis testing. Since prenatal screening began as early as 1966, the number of metabolic defects and genetic disorders that can be diagnosed prenatally has expanded greatly. There are also discussions requiring tests for parents who are participating in in-vitro fertilization programmes and those who are at genetic risk. Preimplantation testing of embryos might ensure that only embryos free of genetic diseases or problems would be placed in the uterus.

Newborn screening involving the analysis of blood or tissue samples is used to detect genetic diseases like phenyketonuma, which results in retardation. This can be prevented by putting the afflicted infant on a special diet.

Carrier screening identifies individuals with an abnormal gene of chromosome that may cause problems either for the offspring of the person screened. The testing of blood or tissue samples can indicate the existence of a particular genetic trait, changes in the chromosomes or changes in DNA that are associated with inherited diseases in asymptomatic individuals. Such tests have been developed for cystic fibrosis, Duchenne muscular dystrophy, haemophilia and neurofibromatosa. It has also become possible to identify certain cancer-prone individuals through genetic testing.

Forensic testing using DNA technology has been used for paternity testing and forensic serological products. DNA profiling is a powerful tool in clearing the innocent and convicting the guilty where crimes are concerned.

Susceptibility screening is used to identify workers who may be susceptible to toxic substances that are found in their workplace and may cause future disabilities. It has been reported that in USA in 1986, about 400,000 workers become disabled by occupational illnesses and these illnesses are precipitated by genetic hyper-susceptibility as co-workers are unaffected.

Advantages of Genetic Testing

1. Genetic testing can be used for medical intervention and research, reproductive data, enumeration, monitoring and surveillance as well as for registries of genetic diseases and disabilities.

2. From a public health point of view, genetic testing will provide comprehensive information about both individuals and population groups. It may prevent costly treatment of a disease, protect third parties and give the person an option for treatment.

Disadvantages of Genetic Testing

1. After tests have been carried out, there are dangers of isolation of individuals, loss of insurance and lack of educational and job opportunities for persons diagnosed with incurable and costly disorders known from early childhood.

2. There are a few areas of sensitivity:

    a. The workplace, where employers may choose to test job applicants, or those already employed, for susceptibility to toxic substances or for genetic variations that may lead to future disabilities, thereby raising health or workmen's compensation costs.
    b. The insurers (either life or health insurance companies) might use genetic information or tests as criteria for denying coverage or might require reproductive testing to be done for cost containment purposes.
    c. Law enforcement officials may carry out genetic tests or use information obtained from the tests without consent.

Genetic Counseling

Non-directive counseling has two major elements. The first in the provision of accurate, full and unbiased information that individuals and families may use in making decisions. The second is an understanding, empathic relationship that offers guidance and helps people to work towards their own decisions. Full disclosure of test results includes ambiguous test results, new and controversial interpretations and differences among professional colleagues in regard to test interpretation. The ethical principles applied to genetic counseling are not listed out in this paper.

Recommendation by MMA

1. Genetic screening should always be voluntary and not mandatory.
2. Genetic testing and chromosomal analysis form an integral part of diagnosis and should be encouraged.
3. Amniotic fluid analysis for congenital abnormalities is an established and acceptable procedure.
4. Genetic experimentation and manipulation would be very difficult to supervise and control in developing countries and therefore should be discouraged.
5. Genetic testing just to determine the socially derived sex of an unborn baby must be resisted.

6. There are no ethical issues in forensic DNA profiling services since samples submitted are solely for the purposes of forensic identification whereby the genetic loci analysed are non-coding regions. This should be supported.
7. The Ministry of Health should form an Advisory Committee on genetic testing. Members of the Committee should include the stakeholders in genetic testing.

   - professionals involved in general medicine, genetics, pathology, genetic testing, the biotechnology industry, consumers, MMA, insurers and other interested parties like Society of Occupational Health.

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