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Practical Genetics

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Genetic diseases are variations in the DNA sequences of a particular gene which affect its function thus cause a particular disease. These changes may present from the conception moment, and will be found in the DNA of each cell in the body. These consist of cells in the gonads (sperms or ova) and such variations will be transmitted to children. Such variations are called germline variants. Other variations will be restricted to specific cells that are not gonads, and then will not be transmitted to children. In this case it is called somatic variants [1].

Genetic diseases are frequently divided to Mendelian or multi-factorial disorders. Factors affecting variable penetrance can be genetic and/or environmental. Multi-factorial genetic disorders are due to these combinations between gene mutations and environmental factors. Where, Mendelian (single gene or monogenic) disorders can affect only one individual in the family. This might be owed to autosomal recessive or X-linked inheritance (with unaffected carriers of the mutation in the family), non-penetrance (with some carriers of the mutation not being affected, often for unknown reasons). The affected person will have a new dominant mutation which take place in the egg or sperm from which they develop. Then, the key to understand Mendelian inheritance is to get an overview about the family tree and construct a pedigree data that include multi-generations [1,2].

Genetic testing is defined as "the investigation of human DNA, RNA, chromosomes, proteins and certain metabolites to notice heritable disorder-related genotypes, mutations, phenotypes or karyotypes for clinical purposes." Such purposes consist of predicting risk of diseases and develop curative strategies [3]. Prenatal, newborns, carriers and families in high risk, are included. Clinical data that include the finding of the physical examination, the medical and family history, is essential to select the significant diagnostic test and to observe the interpretation of the results [4].

Genetic testing of somatic cell observes the non-heritable mutation; examines tissue (usually cancer) for diagnostic or treatment purposes. Diagnostic testing for heritable mutation responsible for the disease; involves testing an affected person to recognize this mutation. Predictive testing for heritable mutation; involves check an unaffected relative for a germline mutation, so the risk of disease will vary depending on the mutated gene and the family history [1].

Disease name and clinical condition are targeted for the genetic testing. Epidemiological matters (prevalence, sex ratio, morbidity and mortality), pathophysiological (expected molecular genetic mechanism of disease), characterization of disease (symptoms, age of onset, complications and accurate natural course including prognosis), therapy (availability, effects, limitations or side effects, prevention and treatment method), genetic effects (mode of inheritance, penetrance, probability of germinal mosaicism, recurrence rate of the siblings and children, genotype-phenotype correlation and availability of genetic counseling); should be taken into consideration [5].

Genetic data do not demonstrate any variations during the individual's life; although they are slightly shared with biological relatives. Not only the genotype or phenotype of the individuals will be tested, but also their relatives can be participated, then it is possible to make a diagnosis for non-progressive carriers (who will rarely develop the disease in the future, but acquire a mutated gene, and this mutation may possibly be transmitted to the next generation). It is possible to predict the development of in-advance disease with almost 100% accuracy, then can be used for prenatal diagnosis, but if it is disclosed and/or performed incorrectly, it may cause several social troubles to the examinees and their relatives [5].

Prenatal tests which may be performed to the fetus may include ultrasound, chorionic villus sampling (CVS), aminocentesis and maternal serum α-fetoprotein. Screening for chromosomal abnormalities using the fluorescence in-situ hybridization (FISH) is a challenge. This technique is highly performed in interpretation for birth defects and mental retardation, microdeletion/microduplication syndromes and chromosome enumeration and ploidy states. These tests will provide information to decide whether to continue the pregnancy if fetal impairment is detected [6,7].

However, another hypothesis states that current methods do not detect all mutations that might occur in the gene. It may not suggest a clinically useful result, it provide probabilistic not deterministic information. There are many different genes that may cause the disease; some of these genes have not been identified yet, since a mutation in one gene can cause different diseases. Furthermore the genetic testing, like any medical test, is subjected to laboratory error; this gives a sight on the priority for genetic counseling [1].

Genetic counseling is a contact between a healthcare professional qualified in genetics field and an individual/family threatened by an inherited disease. This aimed to take account on promoting awareness of the medical situation of the affected person, understanding the role of heredity in the expression of the disease and the risk of recurrence, discussing the options offered for treating the disease and supporting families in finding the options that are most proper for them [8].

Finally, genetic testing is any test that yields a significant genetic data, definitely revealing underlying DNA information, either germline or somatic. Geneticists may need to be able to recognize patients with or at risk of genetic conditions, collect family history data, explain the results of a test, evaluate genetic risk by considering the implications of a genetic testing to other members of the patient's family, and then use multi-generational family history information to draw a pedigree [9].

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