Free research paper on genetic testing

Science, Genetics

Genetic testing also known as DNA based test, involves direct examination of DNA molecules to test for genetic disorders and diseases (Gene testing, 2010). According to the Human Genome Project Information (2010), Genetic testing can be used for various reasons to include:

- Identifying people who carry one copy of genes causing diseases such as Huntington’s disease.
- Pre-symptomatic testing of adult onset diseases/disorders such as Huntington’s disease
- Forensic testing and determining paternity.
- Newborn screening and pre- natal testing.

DNA stands for Deoxyribonucleic acid, and these are chemical Data bases that carry information about your body function. It can reveal information of your genes that can cause diseases or illness to yourself or your offspring. Each time a cell divides in the body the DNA also splits into two single strands and this process is called DNA Replication. As replication takes place there is also a process called Transcription, which involves copying of DNA into messenger RNA (mRNA), which carries information needed for protein synthesis in the cell.

Transcription is then followed by the process of transferring the mRNA out of the nucleus of the cell into the cytoplasm and to the ribosome which is the cell’s protein synthesis factory. The process by which messenger mRNA directs the synthesis of proteins through help of transfer RNA (tRNA) is called translation.

Some genetic disorders are as a result of genomic DNA sequence variations or diseases causing mutations in human cells. Studies of genetic disorders

and genetic testing are usually done through family based studies.

Some common disorders or diseases that affect human beings and are associated with genetic predisposition include; Huntington’s disease, Down syndrome, Alzheimer’s disease, Turner syndrome, Muscular dystrophy, Lynch disease, Cystic fibrosis, Alleles etc

Huntington’s disease is a disorder that affects the central nervous system of humans causing irreversible damage and degeneration of the whole system. It has a very poor prognosis and eventually leads to death of the victims. Studies indicate that children born to parents with Huntington’s disease, regardless of gender have a 50% chance of inheriting the gene that causes this disease.

Huntington’s disease can be tested using a direct gene test and this can apply to three different situations or purposes, namely;

- For confirmation of suspected Huntington’s disease.
- Testing asymptomatic individuals with high risk of carrying the genes that cause HD
- For prenatal testing

For someone to be tested and diagnosed with a genetic disorder like Huntington’s disease or any other progressively incurable disease that is genetically related, he or she requires psychological and emotional support to help him deal with the problem. The news and disclosure of the test results is traumatic and devastating and there is need for professional counseling and support to victims before and after the test.

Huntington’s disease is an autosomal disorder that affects people mainly in the age bracket of 30-35years but some isolated cases of the disease have

manifested itself in early childhood or in old age.

There has been advancement of research that evolved from clinical trials and observational tests for possible treatment. The first breakthrough of genetic testing was when individuals were tested genetically to determine the risk of developing HD before symptoms occurred. The HD gene was cloned and mutation was identified as an unusual type of a CAG repeat expansion gene. The HD test that turns out positive shows the person already acquired half of chromosomes with HD genes from one parent. HD is not necessarily environmentally related.

Diabetes type II is considered to be a genetically related disease but environmental factors also play a great influence in its development. There is a connection between family history and lineage to diabetes type 2 but lifestyle factors also influences its development and progression. For people with type II diabetes it may be difficult to figure out whether their diabetes is due to lifestyle factors or genetic susceptibility (Kang, Ripsin and Urban, 2009).

Majority of people with diabetes type II are either overweight or obese; environmental factors such as food and lack of exercise play a major role in its development. Rare forms of diabetes type II are caused by gene mutation and it is a very difficult process to test or link diabetes type II to genetic causes. This process may involve carrying out the test on all families and their siblings for generations and generations.

Some advantages of modern genetic testing and allowing commercial companies to market and sell genetic testing kits are many. Accessibility of tests kits directly to consumers (DTC testing,) allows consumers to directly

purchase the kits from dealers and administer the tests themselves without necessarily having to go through a doctor. This has made it possible to test for breast cancer alleles and mutation linked to cystic fibrosis. This helps to promote proactive healthcare and maintain privacy of genetic information. Commercial genetic testing kits has also increased the number of pre-symptomatic testing in individuals at risk. This has helped reduce the degree of uncertainty and possibility to plan for early treatment and prevention of complications.

Some of the limitations involved with commercializing genetic testing are as follows:

- Lack o f government regulation and potential misinterpretation of results.
- Medical legal issues e. g. consent on testing especially minors, privacy of data, confidentiality, insurance, etc.

Risks involved in unregulated advertising and marketing of these test kits adds up to commercializing healthcare. Exaggerated and inaccurate information on the connection between test results and the risk of developing the disease may destabilize the patient and cause serious damage to their health and wellbeing.

Genetic testing has become a common phenomenon in modern clinical practice. It helps to diagnose individual’s vulnerability to inherited diseases and also for the possible presence of a genetic disease itself. Some individuals possess mutant forms of genes that increases their vulnerability and risk of developing certain forms of genetic disorders. Results of a genetic test can either confirm or rule out presence of genes causing a genetic disease, the risk of some individuals developing the disease or the

probability of someone passing on a genetic disorder. But critical things to consider before undertaking any test include consent, privacy and confidentiality. I believe with proper preparation and counseling on how to cope with results more people would consider undergoing this test. Genetic testing is a complex process that requires reliable laboratory procedure and accuracy in interpretation. Results of genetic testing are usually challenging to interpret and divulge. The sensitivity and specificity of these tests vary depending on the method used, the expertise, and the family history of individuals.

However, in my case if I tested positive for Huntington's disease, I would choose to have children because this does not mean that I will automatically pass the genes to my offspring. Chances of my offspring acquiring Huntington's gene are 50-50 (Walker, 2007). A negative result does not guarantee that you won't have a certain disorder and a positive result does not also guarantee that you must have a disorder or you must pass it to your offspring.

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