An Analysis and Study on Genetic Diseases

Dr. Anitha S
Department of Biomedical engineering
ACSCE
Banglore, India

Hemanth Kumar G
Department of Biomedical engineering
ACSCE
Banglore, India

Sushma P
Department of Biomedical engineering
ACSCE
Banglore, India

Meghana R
Department of Biomedical engineering
ACSCE
Banglore, India

***Abstract*-- Every living being is comprise of units known as cells, which are also called as basic building blocks of living organisms. The main component of cell is nucleus which consists of a networking line or thread like structure known as chromatin networks. This network is basically made up of DNA(deoxyribonucleic acid), this is the one which carries genes that are segments in it. Genes are the ones which carries the characters of a person like eye color, hair color, height, blood type etc. A genetic disorder is a disease caused in whole or in part of a body by a change in the DNA sequence. Genetic disorders can be caused by a mutation in one gene, mutations in multiple genes, combination of gene mutations and environmental factors, or by occurrence of any damage to chromosomes. There are many methods for detection of abnormal DNA.**

Keywords— Deoxyribonucleic acid, genes, dna, chromatin networks, genetic disorder.

I. INTRODUCTION

As we unlock the secrets of the complete set of human genes, many diseases have a genetic component. The reasons for these are by mutations that are inherited from the parents and are also present in that individual at birth, by acquired mutations in a gene or a group of genes that occur during a person's life due to some environmental exposure this type is not inherited from their parents.

Geneticists group genetic disorders into three categories:

Monogenetic disorders: are caused by a mutation in a single gene. The mutation may be present on one or both chromosomes as each one chromosome inherited from each parent. Monogenic disorders are relatively rare in comparison with more commonly-occurring diseases, such as diabetes and heart disease. A major distinction among monogenic disorders is between "dominant" and "recessive" diseases. Dominant diseases are caused by the presence of the disease gene on just one of the two inherited parental chromosomes. In dominant diseases, the chance of a child inheriting the disease is 50 percent. In a family situation, for example, if the parents have four children, it may be possible that two of those children inherit the disease gene. Recessive diseases require the presence of the disease gene on both of the inherited parental chromosomes. In this case, the chance of a child inheriting a recessive disease is 25 percent. In the family example, if the parents have four children, it may be more likely that only one child will develop the disease. Examples of recessive diseases include cystic fibrosis and Tay-Sachs disease.

Multifactorial inheritance disorders: are caused by a combination of small inherited variations in genes, often acting together with environmental factors. Heart disease, diabetes, and most cancers are examples of such disorders. Behaviors are also multifactorial, involving multiple genes that are affected by a variety of other factors. Researchers are learning more about the genetic contribution to behavioral disorders such as alcoholism, obesity, mental illness and Alzheimer's disease.

Chromosome disorders: are caused by an excess or deficiency of the genes that are located on chromosomes, or by structural changes within chromosomes. although no individual gene on the chromosome is abnormal. The absence or non-expression of a group of genes on chromosome 15 causes Prader willi syndrome. A specific form of blood cancer may be caused by a chromosomal translocation, in which portions of two chromosomes (chromosomes 9 and 22) are exchanged. No chromosomal material is gained or lost, but a new, abnormal gene is formed that leads to formation of cancer.

Genetic disorders typically involve the inheritance of a particular mutated disease-causing gene, such as sickle cell disease, cystic fibrosis, and Tay-Sachs disease. The mutated gene is passed down through a family, and each generation of children can inherit the gene that causes the disease. Rarely, one of these monogenic diseases can occur spontaneously in a child when his/her parents do not have the disease gene, or there is no history of the disease in the family. This can result from a new mutation occurring in the egg or sperm that gave rise to that child.

Most genetic disorders, however, are "multifactorial inheritance disorders," meaning they are caused by a combination of inherited mutations in multiple genes, often acting together with environmental factors. Examples of such diseases include many commonly-occurring diseases, such as heart disease and diabetes, which are present in many people in different populations around the world.

Research on the human genome has shown that although many commonly occurring diseases are usually caused by

inheritance of mutations in multiple genes at once, such common diseases can also be caused by rare hereditary mutations in a single gene. In these cases, gene mutations that cause or strongly predispose a person to these diseases run in a family, and can significantly increase each family member's risk of developing the disease. One example is breast cancer, where inheritance of a mutated BRCA1 or BRCA2 gene confers significant risk of developing the disease.

II. METHODOLOGY

Genetic testing uses laboratory methods to look at our genes.

The procedure as follows:

Identify gene changes that are responsible for an already diagnosed disease

Determine the severity of a disease

Guide doctors in deciding on the best medicine or treatment to use for certain individuals

Identify gene changes that may increase the risk to develop a disease

Identify gene changes that could be passed on to children

Screen newborn babies for certain treatable conditions

Diagnostic testing is used to precisely identify the disease that is making a person ill. The results of a diagnostic test may help you make choices about how to treat or manage your health.

Predictive and pre-symptomatic genetic tests are used to find gene changes that increase a person's likelihood of developing diseases. The results of these tests provide you with information about your risk of developing a specific disease. Such information may be useful in decisions about your lifestyle and healthcare.

Carrier testing is used to find people who "carry" a change in a gene that is linked to disease. Carriers may show no signs of the disease; however, they have the ability to pass on the gene change to their children, who may develop the disease or become carriers themselves. Some diseases require a gene change to be inherited from both parents for the disease to occur. This type of testing usually is offered to people who have a family history of a specific inherited disease or who belong to certain ethnic groups that have a higher risk of specific inherited diseases.

Prenatal testing is offered during pregnancy to help identify fetuses that have certain diseases.

Newborn screening is used to test babies one or two days after birth to find out if they have certain diseases known to cause problems with health and development.

Pharmacogenomic testing gives information about how certain medicines are processed by an individual's body. This type of testing can help your healthcare provider choose the medicines that work best with your genetic makeup.

Research genetic testing is used to learn more about the contributions of genes to health and to disease. Sometimes the results may not be directly helpful to participants, but they may benefit others by helping researchers expand their understanding of the human body, health, and disease.

Some of the tests done in fetus is given bellow:

A. Chorionic villus sampling:

Chorionic villus sampling (CVS) is a prenatal test in which a sample of chorionic villi is removed from the placenta for testing. The sample can be taken through the cervix (transcervical) or the abdominal wall (transabdominal).During pregnancy, the placenta provides oxygen and nutrients to the growing baby and removes waste products from the baby's blood. The chorionic villi are wispy projections of placental tissue that share the baby's genetic makeup.Chorionic villus sampling is usually done between weeks 10 and 13 of pregnancy earlier than other prenatal diagnostic tests, such as amniocentesis. Chorionic villus sampling can reveal whether a baby has a chromosomal condition, such as Down syndrome. Chorionic villus sampling can also be used to test for other genetic conditions, such as cystic fibrosis.Chorionic villus sampling can provide information about your baby's genetic makeup. Generally, chorionic villus sampling is offered when the test results might have a significant impact on the management of the pregnancy or your desire to continue the pregnancy.It is usually done between weeks 10 and 13 of pregnancy earlier than other prenatal diagnostic tests, such as amniocentesis.

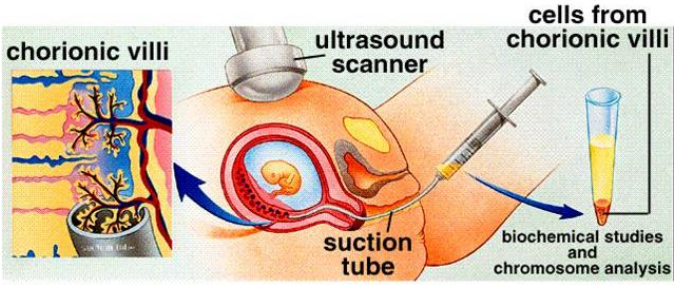

Fig. 1. Chorionic villi sampling

B. Percutaneous Umbilical Cord Blood Sampling:

Percutaneous umbilical cord blood sampling (PUBS) is a diagnostic procedure in which a doctor extracts a sample of fetal blood from the vein in the umbilical cord. This blood can be analyzed to detect chromosomal defects or other abnormalities. PUBS is also known as umbilical vein sampling, fetal blood sampling, and cordocentesis. It can detect chromosomal abnormalities, anemia, anemia, toxoplasmosis and rubella. A specially trained doctor extracts fetal blood. She does this by inserting a fine needle through the woman's abdomen into the fetal vein in the umbilical cord.

C. Amniocentesis:

Amniocentesis (also referred to as amnio, amniotic fluid test or AFT) is a prenatal testing procedure with significant accuracy. It involves collecting an amniotic fluid sample from the uterus and testing it in the laboratory for certain fetal abnormalities and genetic conditions. It is done for detecting hereditary conditions like spina bifida , Down syndrome and

fragile X syndrome, assessing the lung maturity of the baby if an early delivery is required due to some pregnancy complication, detecting intrauterine infections, finding out whether the baby has any life threatening conditions like Edward's syndrome (Trisomy 18), Patau syndrome (Trisomy 13) and Tay-Sachs disease, assessing the health of the baby in case the mother has a blood sensitization, like Rh sensitization (a serious condition that occurs when the mother has a different blood type than the baby). But at present, obstetricians often use a Doppler ultrasound instead of AFT for detecting such conditions, determining the gender of the baby, decreasing the amniotic fluid volume within the uterus.

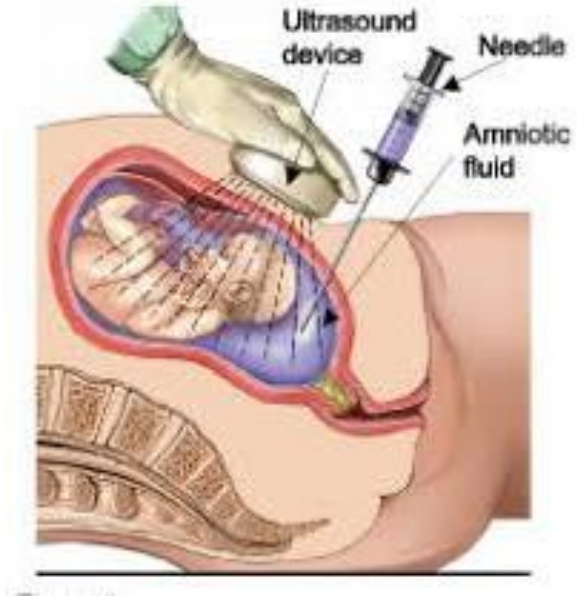

Fig. 2. Amniocentesis

III. CONCLUSION

Genetic test results can be hard to understand, however specialists like geneticists and genetic counselors can help explain what results might mean to you and your family. Because genetic testing tells you information about your DNA, which is shared with other family members, sometimes a genetic test result may have implications for blood relatives of the person who had testing.

REFERENCE

[1] "Gene, mutation, human inherited diseases at dawn of age of personalized genomice" David N Cooper, Jain min chen, Andrew D Phillips.

[2] "Strategies for finding disease genes" Dennis Drayna.

[3] "Genetic diseases symptoms, causes, Treatment, and Prognosis" Melissa conrad stoppler, William C. Shiel Jr.

[4] Wolpaw JR, Birbaumer N, McFarland DJ, Pfurtscheller G,Vaughan TM. Brain-computer interfaces for communication and control. Clin Neurophysiol.

[5] Cook AM, Hussey SM. Assistive technologies: Principles and practice. 2nd ed. St. Louis (MO):