Human genetic disorder

HUMAN GENETIC DISORDER

Introduction

Human Genetic Disorder is a kind of illness which is caused by chromosomes or genes abnormalities. Some disorders like cancer are in part of genetic disorders but they can also be caused by environmental factors. Many disorders are quite rare and they affect one person in a million. There are some types of recessive gene disorders which have an advantage in heterozygous states in some environments. Both genetic and environmental factors play a role in development of any kind of disorder. Human genetic disorder is caused by abnormalities of individuals of genetic materials. There are four types of genetic disorders; single-gene, chromosomal, multifactorial and mitochondrial. Single gene disorder is caused by a single mutated gene. This disorder can be passed on in successive generations in various ways. This disorder is caused mainly by mutation or changes of the DNA sequence in one gene. Multifactorial is caused by a combination of mutation of multiple genes and environmental factors. Chromosomal is caused by abnormalities caused by chromosomes. Mitochondrial is a genetic disorder caused by mutation in the mitochondria nonchromosomal DNA (Driel, et. al, 57).

Single-gene is caused by mutation in the DNA. Gene codes for proteins which are the molecules which carry out most of the work does most of functions in life and even make up various cellular structures. After gene mutation for its proteins product not to carry out its usual functions, this leads to disorder. There are many single-gene disorders which occur in one out of 200 births. Examples include sickle cell anemia, cystic fibrosis, Marfan syndrome, heredity hemochromatosis and Huntington’s disease. Single-gene disorders

are hereditary in identifiable structures. Combination of mutation of multiple genes and environmental factors leads to multifactorial (Bennet, 896). There are various types of genes which influence vulnerability to breast cancer which has been found on different chromosomes. Due to complicated nature of chromosomes, this makes it difficult to analyze chromosomal and single-gene disorders. Most of the common chronic disorders are multifactorial. Examples of this disorder include Alzheimer's, high blood pressure, heart disease, diabetes, arthritis, obesity and cancer. Inheritance of multifactorial is associated with genetic characteristics like height, fingerprint, skin color and eye color (Watson, 44).

Chromosomal disorder is associated with chromosomes. Chromosomes are distinct structures which make up protein and DNA. Chromosomes which are mainly the carriers of heritable materials like chromosome structures abnormalities as missing leads to disease. The major abnormalities of chromosomal can be detected using microscopic examination. Trisomy 21 or Down syndrome is a common disorder which occurs when one has copies of chromosome 21. Mitochondrial disorder caused by mutation in nonchromosomal mitochondria DNA. Many genes are named after the disorder they are associated with. The normal functioning of gene is encoded with protein and this is not associated with cause of illness. Diseases linked with genetics occur when genes are not in a position to work properly. The hemochromatosis gene really codes for membrane protein after mutation of the gene in a manner which prevents if from encoding a functional and normal protein products (Milunsky, 67).

Human genetic disorder is mainly caused by different kind of gene known as variation or modification of gene known as mutation. There are many diseases which are associated with the aspects of genes. Some like cancer are caused by gene mutation. Mutation can occur randomly or due to exposure to the environment like smoke from cigarette. Some of genetic disorders are inherited. Mutated genes are passed down from one generation to another through a family and each child can inherit those genes which causes the disorder. Other genetic disorders are because of problems with different number of packages of genes known as chromosomes like Down syndrome. Human genetic disorders are the main cause of death, disability and human tragedy. It is rare to find in a family which is entirely free from any kind of genetic disorder. Genetic defects are mainly known to cause pregnancy loss in developed countries and several spontaneous miscarriages involve fetus with abnormal chromosomes (McKusick, 15).

Human genetic disorder is caused by nondisjunction which is the failure of chromosomes to replicate during the Anaphase II. Genes lacking chromosomes are not able to produce an embryo which is viable. Nondisjunction is most often linked with 21st chromosome giving rise to Down syndrome which increases chances of developing Alzheimer’s disease. Human genetic disorder is caused by abnormal genes groups which are passed down from one generation to another. Spontaneous mutation of genes is caused by error in the replication of DNA resulting in substitution base or insertion or deletion of one or two pair base from the DNA. Other disorder like somatic genetic disease is caused by sudden appearance of abnormal type of gene in one part of the body like cancer. Others like

chromosomal abnormality are caused by abnormalities in the structure of chromosomes causing Down syndrome (Green and Waterston, 1968).

Human genetic disorder can be diagnosed. Genetic test can be performed to determine whether the person has or doesn’t have the disease even as early as during fetus. This testing is known as antenatal testing. Chromosome test can also be performed to diagnose human genetic disorder. Diagnostic of human genetic disorder is conducted for chromosome disorders, congenital malformations, mental retardation, infertility, learning disabilities, miscarriages, metabolic and molecular disorders and paternity. There are available genetic services for all prenatal, pediatric and adult genetic disorders such as Alzheimer’s, cancer, heart disease and other late disease onsets. Defects of birth lead to death of infants. There are infants who are diagnosed with the main genetic disorders. Genes and DNA performs are arranged on chromosomes. There are no genes which really cause disease. Mutations in the genes caused serious genetic disorder. Gene mutation in chromosomes caused disorder. Not all mutations of genes lead to genetic disorder because some are unexpressed without any noticeable effect to the organism (Baird, et. al, 678).

Conclusion

Human genetic disorder occurs due to abnormalities of individuals of genetic materials. Human genetic disorders are of different types depending on the causes. There are four types of genetic disorders; single-gene, chromosomal, multifactorial and mitochondrial. Genetic disorders are mainly caused by mutation of genes and DNA. Each type of disorder has different cause. For example chromosomal disorder is caused by mutation of chromosomes.

Swap of DNA by chromosomes to form variety of the gene pool can result to disorder leading to exchange of regions. Through abnormalities of genes and chromosomes human genetic disorder occurs. Some disease like cancer is as a result of genetic disorder but they can occur because of environmental factors. Most human genetic disorders are rare and they affect one person in a million. Some recessive gene disorders give advantage to the heterozygous states in some environments.

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