Technical Seminar

On

Genetic Diseases; Comprehension and Perspectives

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Compendium

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College of Veterinary and Animal Sciences, Udgir
Dist- Latur 413517 (M.S.)

Common Genetic Disorders in Human

Narayan Jadhav

MD(Ayu.Med), MBA(HealthCare), PhD(Scho).
Professor & Head, Dept.of Rognidan & Vikruti Vigyan(Pathology).
Dhanwantari Ayurveda Medical College& Hospital, Udgir, Dist. Latur (MH)
Email- drnarayanj52@gmail.com

Introduction

A genetic disease is any disease caused by an abnormality in the genetic makeup of an individual. The genetic abnormality can range from minuscule to major -- from a discrete mutation in a single base in the DNA of a single gene to a gross chromosome abnormality involving the addition or subtraction of an entire chromosome or set of chromosomes.

Some people inherit genetic disorders from the parents, while acquired changes or mutations in a preexisting gene or group of genes cause other genetic diseases. Mutations can occur either randomly or due to some environmental exposure.

Types of genetic disorders:

There are a number of different types of genetic disorders (inherited), they are grouped in four as follow.

1).Single gene inheritance 2).Multifactorial inheritance 3).Chromosome abnormalities4).Mitochondrial inheritance 5).Developemental Defects : (Errors in Morphogenesis 6). Storage Diseases (Inborn errors of Metabolism)

1) Single gene genetic inheritance:

Single gene inheritance is also called Mendelian or monogenetic inheritance. Changes or mutations that occur in the DNA sequence of a single gene cause this type of inheritance. There are thousands of known single-gene disorders.

Single-gene disorders have different patterns of genetic inheritance, including

a) Autosomal dominant inheritance- in which only one copy of a defective gene (from either parent) is necessary to cause the condition;
b) Autosomal recessive inheritance- in which two copies of a defective gene (one from each parent) are necessary to cause the condition; and
c) X-linked inheritance- in which the defective gene is present on the female, or X-chromosome.

X-linked inheritance may be dominant or recessive.Some examples of single-gene disorders include, cystic fibrosis, alpha- and beta-thalassemias, sickle cell anemia,Marfan syndrome, fragile X syndrome, Huntington's disease, and hemochromatosis.

2) Multifactorial genetic inheritance:

Multifactorial inheritance is also called complex or polygenic inheritance. Multifactorial inheritance disorders are caused by a combination of environmental factors and mutations in multiple genes.

For example, different genes that influence breast cancer susceptibility have been found on chromosomes 6, 11, 13, 14, 15, 17, and 22. Some common chronic diseases are multifactorial disorders.

Examples of multifactorial inheritance include, heart disease, high blood pressure, Alzheimer's disease, arthritis,diabetes,cancer, and obesity.

Multifactorial inheritance also is associated with heritable traits such as fingerprint patterns, height, eye color, and skin color.

3) Chromosome abnormalities:

Chromosomes, distinct structures made up of DNA and protein, are located in the nucleus of each cell. Because chromosomes are the carriers of the genetic material, abnormalities in chromosome number or structure can result in disease. Abnormalities in chromosomes typically occur due to a problem with cell division.

For example- Down's syndrome or trisomy 21 is a common genetic disorder that occurs when a person has three copies of chromosome 21. There are many other chromosome abnormalities including: Turner syndrome (45,X0), Klinefelter syndrome (47, XXY), and Cri du chat syndrome, or the "cry of the cat" syndrome (46, XX or XY, 5p-).

Diseases may also occur because of chromosomal translocation in which portions of two chromosomes are exchanged.

4) Mitochondrial genetic inheritance:

This type of genetic disorder is caused by mutations in the non-nuclear DNA of mitochondria. Mitochondria are small round or rod-like organelles that are involved in cellular respiration and found in the cytoplasm of plant and animal cells. Each mitochondrion may contain 5 to 10 circular pieces of DNA. Since egg cells, but not sperm cells, keep their mitochondria during fertilization, mitochondrial DNA is always inherited from the female parent. Examples of mitochondrial disease include Leber's hereditary optic atrophy (LHON), an eye disease; myoclonic epilepsy with ragged red fibers (MERRF); and mitochondrial encephalopathy, lactic acidosis, and stroke -like episodes (MELAS), a rare form of dementia.

5) Developemental Defects : (Errors in Morphogenesis)

These defects are a group of abnormalities during feotal life due to errors in morphogenesis. The branch of science dealing with the study of developemental anomalies is

called Teratology. Certain chemicals, drugs, physical and biological agents are know to induce such birth defects are called teratogens.

The morphological abnormality or defects of an organ or anatomic regions of the body so prouduced is called Malformation. These are categorized as under .

1. Agenesis : The complete absence of an organ. e.g. Unilateral or bilateral agenesis of kidney.
2. Aplasia: The absence of development of an organ with presence of rudiment or anlage. e.g.Aplasia of lung with rudimentary bronchus.
3. Hypoplasia: It is incomplete development of an organ not reaching the normal adult size e.g. Microglossia.
4. Atresia: In complete formation of lumen in hollow Viscus.e.g. oesophageal atresia.
5. Developemental dysplasia: It is defective development of cells and tissues resulting in abnormal or primitive histogenetic structures .e.g. renal dysplasia.
6. Dystrophic anomalies: These are defects resulting from failure of fusion.e.g. Spina bifida.
7. Ectopia or heterotopia: It refers to abnormal location of tissue at ectopic site.e.g.Pancreatic hetrotopia in the wall of stomach.

6). Storage Diseases (Inborn errors of Metabolism):

These are biochemically distinct groups of disorders occurring due to genetic defect in the metabolism of Carbohydrates ,Lipids and Proteins resulting in intracellular accumulation of metabolites. All the storage diseases occur as a result of autosomal recessive or sex(x) linked recessive genetic transmission.Most but not all ,of the storage diseases are Lysosomal storage disease e.g. Type II-Pompe's disease.

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