Course Name- Human Cytogenetics

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Unit 4: Genetic Disorders

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Genetic Disorders

- A genetic disorder is an illness caused by one or more abnormalities in the genome, especially a condition that is present from birth (congenital).
- Genes are the building blocks of heredity. They are passed from parent to child. They hold DNA, the instructions for making proteins. Proteins do most of the work in cells. They move molecules from one place to another, build structures, break down toxins, and do many other maintenance jobs.
- Sometime there is a mutation, a change in a gene or genes. The mutation changes the gene's instructions for making a protein, so the protein does not work properly or is missing entirely. This can cause a medical condition called a genetic disorder.

- Most genetic disorders are quite rare and affect one person in every several thousands or millions.
- In non-heritable genetic disorders, defects may be caused by new mutations or changes to the DNA.
- The defect will only be heritable if genetic disorder occurs in the line.
- Genetic disorder generally occur in two different varieties.

a) Chromosomal defect (chromosomal mutations)

b) Gene defect (gene mutations)

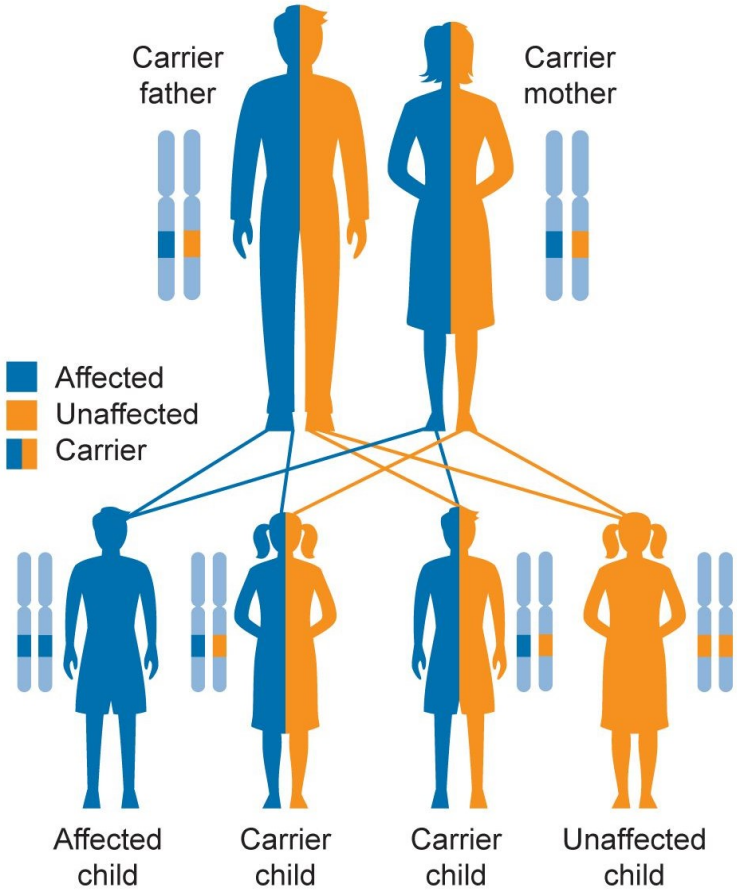
- There are a number of different types of genetic disorders (inherited) and include:

1. Single gene inheritance:

Single gene inheritance is also called Mendelian or monogenetic inheritance. Changes that occur in the DNA sequence of a single gene cause this type of inheritance. Single-gene disorder have different patterns of genetic inheritance, including

- Autosomal dominant inheritance, in which one copy of a defective gene (from either parent) is necessary to cause the condition

Autosomal Recessive Inheritance



- Autosomal ressive inheritance, in which two copies of a defective gene (one from each parent) are necessary to cause the condition and
- 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

1) cystic fibrosis,
2) alpha-and beta thalassemias,
3) sickle cell anemia,
4) Marfan syndrome,
5) Fragile X syndrome,
6) Huntington's disease and
7) hemochromatosis

2. Multifactorial inheritance:

- Multifactorial inheritance is also called complex or polygenic inheritance.
- Multifactorial inheritance disorder are caused by a combination of environmental factors and mutation in multiple genes. Examples of multifactorial inheritance include

1) heart disease,
2) high blood pressure,
3) Alzheimer's disease,
4) arthritis,
5) diabetes
6) cancer and
7) obesity

3. Chromosome abnormalities

Chromosome, 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.

There are many other chromosomal abnormalities including:

1) Turner syndrome (45,0),
2) Kinefelter syndrome (47,XXY), and
3) Cri-du-chat syndrome, or the “cry of the cat” syndrome (46,XX or XY, 5p-)

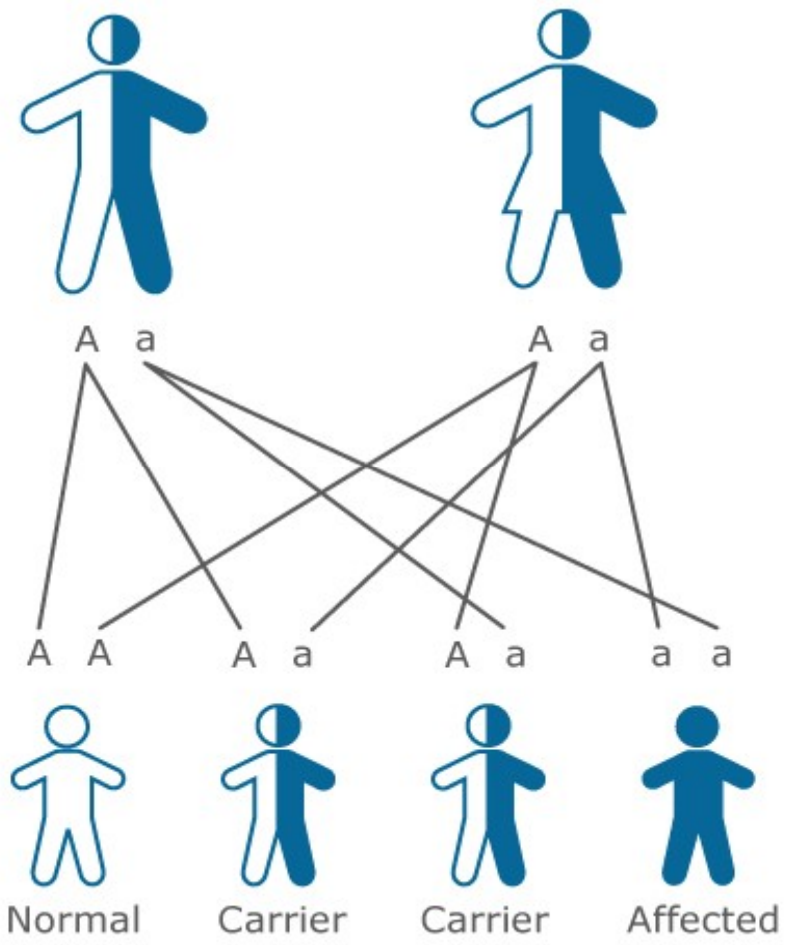
Disease may also occur because of chromosomal translocation in which portions of two chromosomes are exchanged.

4. Mitochondrial inheritance:

This types of genetic disorder is caused by mutations in the non-nuclear DNA of mitochondria.

Example of the mitochondrial disease include:

1) Leber's hereditary optic atrophy (LHON), an eye disease,
2) Myoclonic epilepsy with ragged red fibers (MERRF) and
3) Mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS), a rare form of dementia.



Genetic Disorders

History

- 1866- First there was Gregor Mendle, who studied inherited characteristics.
- This was followed by Francis crick and James Watson who unraveled the DNA molecule.
- This has led us to understanding the human genome sequence
- 1950's- Maurice Wilkins (1916), Rosalind Franklin (1920-1957), Francis H.C. Crick (1916-) of Britain and James D. Watson (1928-) of the U.S. Discover chemical structure of DNA, starting a new branch of science- molecular biology.
- Watson and Crick made a model of the DNA molecule and proved that genes determine heredity.

- 1957- Arthur Kornberg of the U.S. produced DNA in a test tube.
- 1966- The genetic code was discovered.
- 1983- “Genes are able to change position on chromosomes”- Barbara McClintock
- 1990- Gene therapy was used on patients for the first time.
- 1993- Dr. Kary Mullis discovered the PCR procedure, for which he was awarded the Novel prize.
- 1995- DNA testing in forensics cases gains fame in the O.J. Simpson trial.

Types of Genetic Disorders

There are three types of genetic disorders:

- Single-gene disorders, where a mutation affects one gene. Sickle cell anemia is an example.
- Chromosomal disorder, where chromosome (or parts of chromosomes) are missing or changed.

  Chromosomes are the structures that hold our genes.

  Down syndrome is a chromosomal disorder
- Complex disorder, where there are mutations in two or more genes.

  Often lifestyle and environment also play a role. Colon cancer is an example.

Diagnosing Genetic Disorder

Genetic tests are on blood and other tissue to find genetic disorders

Purpose of genetic tests

- Finding genetic diseases in unborn babies
- Finding out if people carry a gene for a disease and might pass it on to their children
- Screening embryos for disease
- Testing for genetic diseases in adults before they cause symptoms
- Making a diagnosis in a person who has disease symptoms

Physical Examination

- Certain physical characteristics, such as distinctive facial features, can suggest the diagnosis of a genetic disorder.
- A geneticist will do a through physical examination that may include measurements such as the distance around the head, the distance between the eyes, and the length of the arms and legs.
- Depending on the situation specialized examinations such as nervous system (neurological) or eye (ophthalmologic) exams may be performed.
- The doctor may also use imaging studies including x-rays, computerized tomography (CT) scans, or magnetic resonance imaging (MRI) to see structures inside the body.

Genetics, Disease Prevention and Treatment

- Most treatment options revolve around treating the symptoms of the disorders in an attempt to improve patient quality of life.
- Gene therapy refers to a form of treatment where a healthy gene is introduced to a patient.
- Check regularly for the disease.
- Follow a healthy diet.
- Get regular exercise.
- Avoid smoking tobacco and too much alcohol.
- Get specific genetic testing that can help with diagnosis and treatment.

THANK YOU