Third stag **Medical Genetics**

Lab 4 medical genetic and inherited trace

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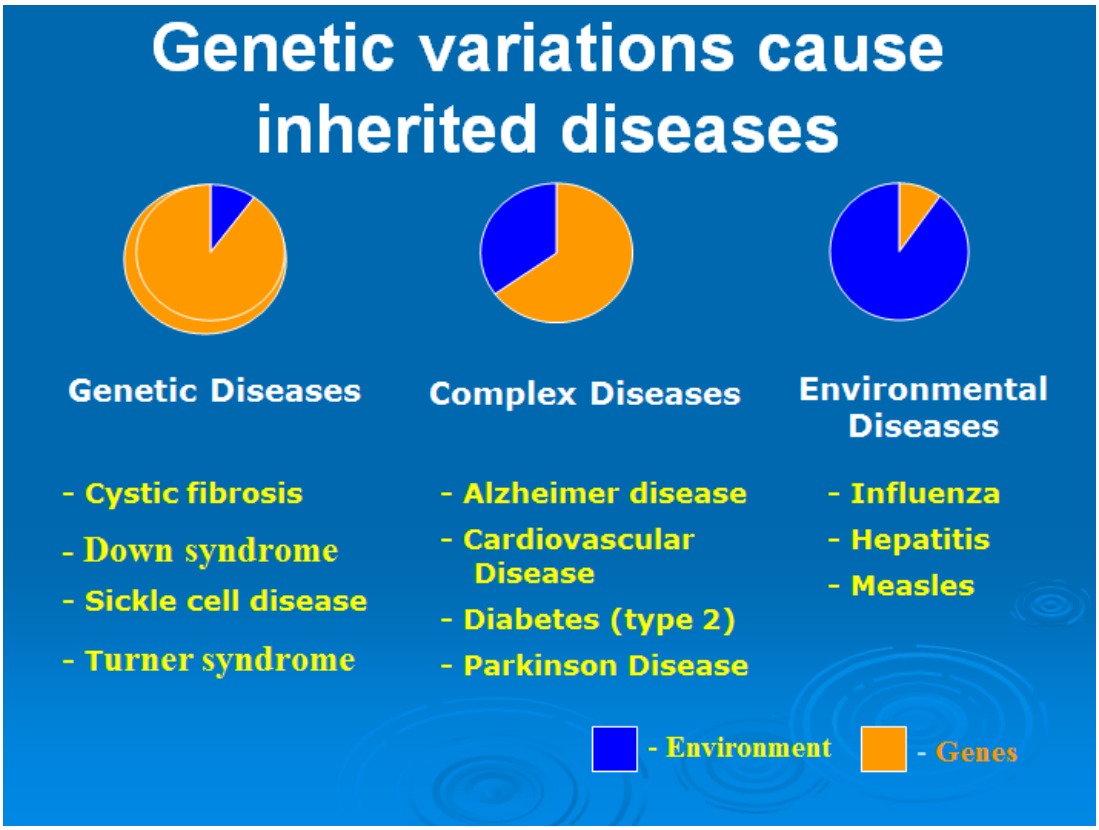
Lab 4

Medical genetics is the specialty of medicine that involves the diagnosis and management of hereditary disorders, Genetic medicine is a newer term for medical genetics and incorporates areas such as gene therapy, personalized medicine, and the rapidly emerging new medical specialty, predictive medicine.



Various specialties within medical genetics are interrelated:

1. Clinical Genetics.
2. Cytogenetics.

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3. Molecular Cytogenetics.

4. Molecular Genetics

5. Human genetics, molecular biology, genetic engineering, or biotechnology.

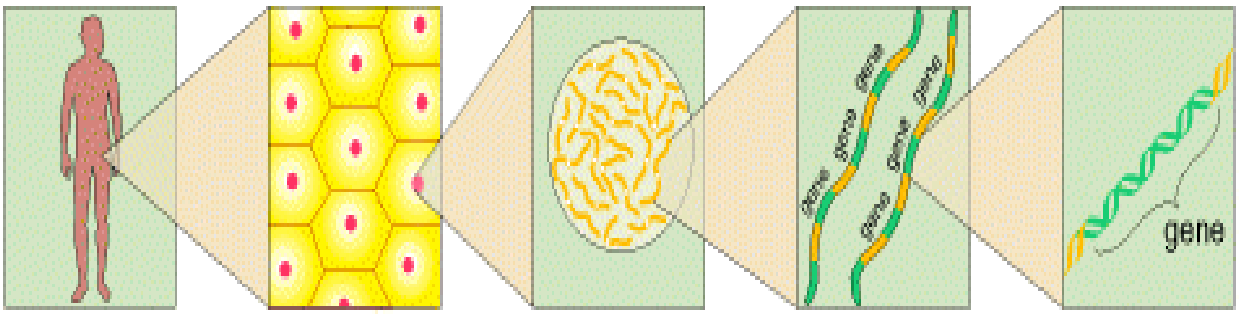
*** The diagnosis of a genetic disease requires a comprehensive clinical examination composed of three major elements:**

1.a physical examination

2.a detailed medical family history

3.clinical and laboratory testing if available.

While primary care providers may not always be able to make a definitive diagnosis of a genetic disease, their role is critical in collecting a detailed family history, considering the possibility of a genetic disease in the differential diagnosis, ordering testing as indicated and, when available, appropriately referring patients to genetic specialists**.**



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- All living organisms consist of cells.
- In each cell there is chromosomes.
- Chromosomes are strings of DNA
- Chromosome consists of genes, blocks of DNA.
- Each gene encodes a particular protein.

Forms of genetic variations

1- Single nucleotide substitution: replacement of one nucleotide with another

GATTTAGATC**G**CGATAGAG

GATTTAGATC**T**CGATAGAG

2- Microsatellites or minisatellites: these tandem repeats often present high levels of inter- and intra-specific polymorphism

3- Deletions or insertions: loss or addition of one or more nucleotides

4- Changes in chromosome number, segmental rearrangements and deletions.

Dominant genes

In dominant genetic disorders, if one affected parent has a disease-causing allele that dominates its normal counterpart, each child in the family has a 50-percent.

Examples include:

- **Achondroplasia.** This is a bone development disorder that causes dwarfism.
- **Marfan syndrome.** This is a connective tissue disorder that causes long limbs and heart defects.

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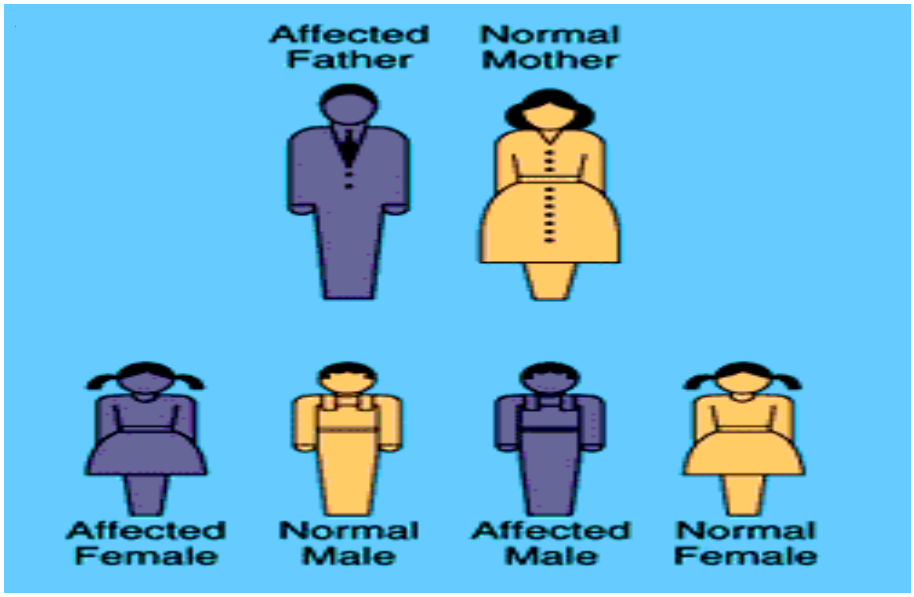
chance of inheriting the disease allele and the disorder.



Recessive genes

In diseases associated with altered recessive genes, both parents - though disease-free themselves- carry one normal allele and one altered allele.

Each child has one chance in four of inheriting two altered alleles and developing the disorder;

one chance in four of inheriting two normal alleles, two chances in four of inheriting one normal and one altered allele, and being a carrier like both parents.

Examples include:

 Cystic fibrosis. This is a disorder of glands that causes excess mucus in the lungs. It also causes problems with how the pancreas works and with how food is absorbed.

 Sickle cell disease. This condition causes abnormal red blood cells that don't carry oxygen normally.

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 Tay-Sachs disease. This is an inherited autosomal recessive condition that causes the central nervous system to decline. The condition is fatal, usually by age.