



## Lec 5 \ Principles of Cytogenetic

قسم علوم التقنيات الاحيائية الطبية  
المرحلة الاولى

اعداد

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الايمل :

رمز الصف :

## جو هين تجيو + ألبرت ليفان

في عام 1956، داخل مختبر متواضع في السويد، كان عالمان—جو هين تجيو (Joe Hin Tjio) وألبرت ليفان (Albert Levan) يحاولان حل لغز حيّر العلماء لسنوات طويلة. حتى ذلك الوقت، كان العالم يعتقد أن عدد الكروموسومات في الإنسان هو 48 كروموسومًا .

لكن شيئًا لم يكن مقنعًا...وقف تجيو أمام المجهر لساعات طويلة، يُحضّر الخلايا بدقة غير مسبقة، مستخدمًا تقنيات جديدة لإيقاف الخلايا في مرحلة الانقسام، ثم قام بفرد الكروموسومات بعناية.

نظر مرة... ثم أعاد العد. ثم مرة أخرى...46 فقط. تردد للحظة... هل أخطأ؟  
أعاد التجربة... نفس النتيجة. أعادها مرة ثالثة... 46 كروموسومًا. في تلك اللحظة، لم يكن مجرد رقم قد تغير، بل تصحيح لواحد من أهم المفاهيم في علم الإنسان. نُشر الاكتشاف، وبدأت ثورة علمية: أصبح بإمكان العلماء ربط تغير عدد أو شكل الكروموسومات بالأمراض، ومن هنا وُلد علم الوراثة الخلوية الحديثة.

# Cytogenetics

Cytogenetics is the branch of genetics that deals with the study of chromosomes, their structure, function, and role in inheritance and genetic disorders. In general, genetics is the study of inherited traits and their variation. Some people confuse genetics with genealogy, which consider relationships but not traits .

Genetics is a life science associated with other sciences by one or more than elements and describes the results reveals from their genetics. Genetics is unlike other life sciences in how directly and intimately it affects our lives. It obviously impacts our health, because we inherit certain diseases and disease susceptibilities.

# Cytogenetics types:

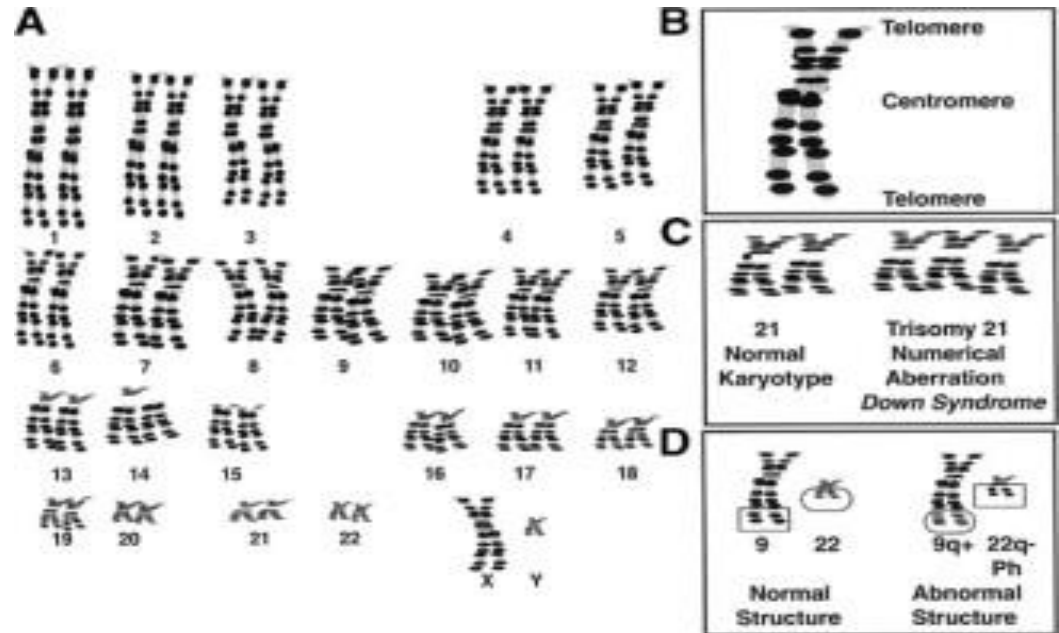
- 1- Physiological Cytogenetics
- 2- Molecular Cytogenetics
- 3- Immune Cytogenetics
- 4- Biochemical Cytogenetics



# Levels of Genetics:

There are many levels accomplished to study the genetics represented by:

- 1- Human Cytogenetics
- 2- Animals Cytogenetics
- 3- Plants Cytogenetics



# There are many levels accomplished to study the genetics represented by:

- DNA & RNA
- Genes , chromosomes and Genomes
- Cells ,Tissues and Organs
- Individual
- Family
- Population
- Evolution

# Gene, chromosome & Genome

**Genes** are the units of heredity, the sets of biochemical instructions that tell cells, the basic units of life, how to manufacture certain proteins. These proteins ultimately underlie specific traits; a **missing protein blood-clotting factor**, for example, **causes the inherited disease hemophilia**.

Individual genes come in variants **that differ from each other by small changes in the DNA** base sequence. The variants of a gene are **called alleles**, and these changes in DNA sequence arise by a **process called mutation**.

# chromosome

Genes are **part of larger** structures called chromosomes, which also include proteins that the DNA wraps around. A human cell has **23 pairs of chromosomes**. **22 pairs are autosomes**, or chromosomes that do not differ between the sexes. The autosomes are numbered from 1 to 22, with 1 being the largest. The other 2 chromosomes, the X and the Y, are sex chromosomes.

**Genome**, is The complete set of genetic **information characteristic** of an organism, including protein encoding genes and other DNA sequences, constitutes a genome

# Cells, Tissues, and Organs

A human body consists of trillions of cells. Most cells contain all of the genetic instructions, but cells differ in appearance and function by using only some of their genes, in a process called differentiation.

**For example:** a muscle cells manufactures its abundant protein fiber, skin cells manufacture keratins, collagen and elastin proteins characteristic of connective tissue cells.

Specialized cells with related functions aggregate and interact to form tissues, which in turn form the organs and organ systems of the individual.

# Individual

Two terms distinguish between the alleles that are present in an individual and the alleles that are expressed. The **genotype** refers to the underlying instructions (**alleles present**), and the **phenotype** is the visible trait, biochemical change, or effect on health (**alleles expressed**). Alleles are further distinguished by how many copies it takes to affect the phenotype.

A **dominant allele** produces an effect when present in just one copy (**on one chromosome**), whereas a **recessive allele** must be present **on both chromosomes to be expressed**. (Alleles on the Y chromosome are an exception; recessive alleles on the X chromosome in males are expressed because there is no second X chromosome to block expression.)

# Family

Individuals are genetically connected into families. Traditionally, the study of traits in families has been called transmission genetics or Mendelian genetics.

Molecular genetics, which considers DNA, RNA, and proteins, often begins with transmission genetics, when an interesting trait or illness in a family comes to a researcher's attention. Charts called pedigrees are used to represent the members of a family and to indicate which individuals have particular inherited traits. , but an unusual one—a family with identical triplets.

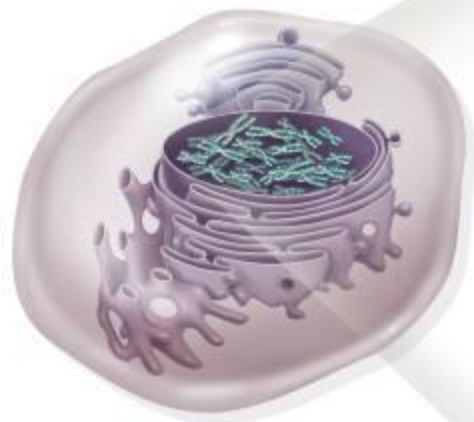
## Population

Above the family level of genetic organization is the population. In a strict biological sense, a population is a group of interbreeding individuals. In a genetic sense, a population is a large collection of alleles, distinguished by the frequency of particular alleles.

## Evolution

التطور

Geneticists have known for decades that comparing DNA sequences for individual genes, or the amino acid sequences of the proteins that the genes encode, can reveal how closely related different types of organisms are. The underlying assumption is that the more similar the sequences are, the more recently two species diverged from a shared ancestor. .



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