التقنيات الاحيائية

علم الوراثة

الفصل الدراسي الثاني

اساتذة المادة:

أ.م.د.اسماء محمد سعود

أ.م.د.رنا كاظم محمد م.د.رامينا ميخائيل

The Cell Cycle

I. Prokaryotic cell division

A prokaryotic cell divides into two nearly equal halves by binary fission. A circular DNA replication starts at the replication origin and goes both ways. By the end, there are two complete, identical circles, each attached to the plasma membrane.

II. Chromosomal DNA molecules in eukaryotic.

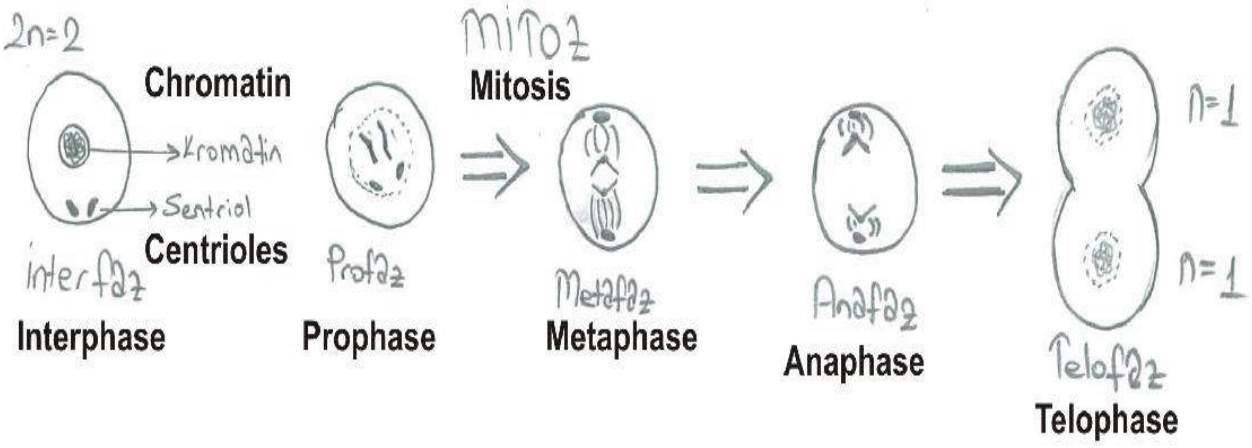
Each chromosome is made up of chromatin, which is a long DNA molecule with proteins attached to it. During cell division, chromatin is packaged into dense chromosomes. When cells are not dividing, the chromosomes are unpacked (decondensed), which helps to ensure proper DNA distribution during cell division.

III. Eukaryotic cell cycle.

The cell cycle defines as the cell's growth and division status. Cells either stop growing or must divide when they reach a certain size. Most, but not all, eukaryotic cells are capable of dividing. In general, Eukaryotic cell generation to take from 8 to 20 hours.

Mitosis

Prophase, metaphase, anaphase, and telophase are the four stages of mitosis, respectively (PMAT)



Meiosis

Meiosis I & II

Prophase, prometaphase, metaphase, anaphase, and telophase are all stages of cell division. In these stages, chromosomes behave similarly but not identically.

Meiosis I:

Chromosomes in a diploid cell resegregate during meiosis I , resulting in four haploid daughter cells. Genetic variation is generated during this stage of meiosis.

Meiosis vs. Mitosis

- **Chromosome behavior**

1- Mitosis: Independent of homologous chromosomes

2- Meiosis: Until anaphase I, homologous chromosomes pair forming bivalents.

- **In meiosis, the number of chromosomes is reduced.**

1- Mitosis: similar daughter cells

2- Meiosis: haploid daughter cells

- **Progeny's genetic identity:**

1-Mitosis: similar daughter cells.

2-Meiosis: The parental chromosomes in daughter cells are rearranged.

3- Meiosis: Crossing over of chromatids, not similar.

Laws of Genetics

1- Law of Segregation
2- Law of Independent Assortment
3- Law of Dominance

Mendel's Laws of Inheritance

- Mendel discovered that crossing plants with white flowers and purple flowers did not produce hybrid offspring. The offspring was purple-flowered, rather than a mixture of the two.

- Mendel's results were condensed into three inheritance laws: the Law of Dominance, the Law of Segregation, and the Law of Independent Assortment.

- He then came up with the concept of "factors," which are two types of heredity units, one recessive and the other dominant. Factors, later called genes, are usually found in pairs in ordinary body cells, but segregate during the development of sex cells, according to Mendel.

The law of Segregationist (Purity of Gametes)

- "Each gene separates from the others during gamete formation, resulting in only one allele for each gene in each gamete."
- According to the Law of Segregation, when an individual creates gametes, the copies of a gene separate so that each gamete only receives one copy.
- One allele or the other will be received by a gamete. The direct proof of this was later found in the process of meiosis.
- The paternal and maternal chromosomes are separated during meiosis, and the alleles with the characters are divided into two gametes.
- For example, pure tall plants are homozygous for genes (factors) TT, while dwarf plants have genes tt. Tallness and dwarfism are two distinct but related variables or determinants. Pure tall plants produce T-type gametes, while dwarf plants produce t type gametes.
- Gametes with T and t combine during cross-fertilization to create F1 hybrids. Tt is the genotype of these hybrids. It means that F1 plants have one gene for tallness and one gene for dwarfness, even though they are tall phenotypically. It seems that the tall and dwarf characters have been contaminated, with just the tall character remaining.
- The genes T (for tallness) and t (for dwarfness) separate and are passed on to separate gametes during gamete formation. As a result, the heterozygote produces two forms of gametes in equal numbers. Gene T is present in 50% of the gametes, while gene t is present in 50% of the gametes.
- As a result, these gametes are either pure for height or pure for dwarfism. (This is why the low of segregation is also known as the law of gamete purity.)

Law of Independent Assortment

- "The law of independent assortment states that the alleles of different genes are inherited independently within the organisms that reproduce sexually."
- The inheritance of several pairs of characters (two or more) is studied at the same time, with the factors or genes for each pair sorting out independently of the others. From the results of a dihybrid cross, Mendel formulated this law.
- In eukaryotic species, independent assortment occurs during meiosis I, specifically metaphase I of meiosis, to produce a gamete with a mixture of maternal and paternal chromosomes. This process, like a chromosomal crossover, adds to genetic diversity by forming genetic combinations.
- The chromosomes that end up in a newly formed gamete are selected randomly from all possible combinations of maternal and paternal chromosomes in the independent assortment.
- Gametes are assorted individually since they end up with a random mix rather than a pre-defined "set" from either parent. As a result, the gamete may have any mix of paternal and maternal chromosomes.
- He selected round-yellow seed and wrinkled green seed for the dihybrid cross and crossed them. In the F1 generation, he only got round yellow seeds. Self-pollination of F1 progeny in four separate seed combinations in the F2 generation. In the phenotypic ratio 9: 3: 3: 1, he obtained round-yellow, wrinkled-yellow, round green, and wrinkled green seeds.
- The phenotypic ratios of 3: 1, yellow: green color, and ratios of 3: 1, round: wrinkled seed form from the monohybrid cross was also retained in the dihybrid cross. As a result, he came to the conclusion that characters are distributed and inherited in separate ways. He came up with his second law, the Law of Independent Assortment, based on this discovery.

Pedigree Analysis

- For genetics researchers and genetic counselors, families are tools, and the bigger the family the better—the more children in a generation, the easier it is to see a mode of inheritance.
- Charts are called pedigrees to display family relationships and depict which relatives have specific phenotypes and, sometimes, genotypes. A human pedigree serves the same purpose as one for purebred dogs or cats or thoroughbred horses—it represents relationships.
- A pedigree in genetics differs from a family tree in genealogy, and a genogram in social work, in that it indicates disorders or traits as well as relationships and ancestry. Pedigrees may also include molecular data, test results, and information on variants of multiple genes.

- A pedigree consists of lines that connect shapes.
- Vertical lines represent generations; horizontal lines that connect two shapes at their centers depict partners; shapes connected by vertical lines that are joined horizontally represent siblings.
- Squares indicate males; circles, females; and diamonds, individuals of unspecified sex. Roman numerals designate generations. Arabic numerals or names indicate individuals.

Modes of Inheritance

1- Autosomal Dominant Inheritance

Criteria for an Autosomal Dominant Trait

1. Males and females can be affected. Male-to-male transmission can occur.
2. Males and females transmit the trait with equal frequency.
3. Successive generations are affected (no skip generations).
4. Transmission stops after a generation in which no one inherits the mutation.

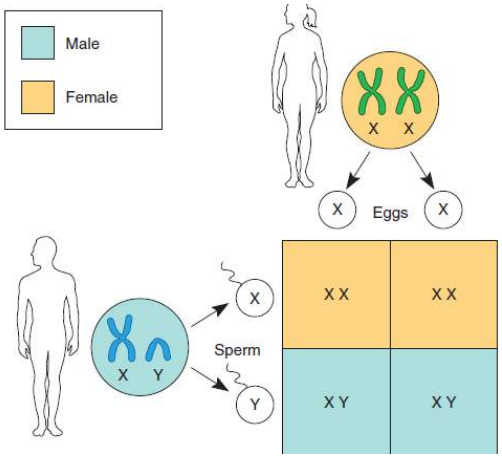
2- Autosomal Recessive Inheritance

Criteria for an Autosomal Recessive Trait

1. Males and females can be affected.
2. Affected males and females can transmit the gene unless it causes death before reproductive age.
3. The trait can skip generations.
4. Parents of an affected individual are heterozygous or have the trait.

➢ Distinguish between Y and X chromosome

- Y-linked traits are rare because the chromosome has few genes, and many have counterparts on the X chromosome.
- Y-linked traits are passed from male to male because a female does not have a Y chromosome.
- Some traits at first attributed to the Y chromosome are genes that are inserted into the chromosome from other chromosomes, such as a deafness gene.
- Genes on the X chromosome have different patterns of expression in females and males because a female has two X chromosomes and a male just one.
- In females, X-linked traits are passed just like autosomal traits—that is, two copies are required for expression of a recessive allele, and one copy for a dominant allele.
- In males, however, a single copy of an X-linked allele causes expression of the trait or illness because there is no copy of the gene on a second X chromosome to mask the effect. A man inherits an X-linked trait only from his mother.
- The human male is considered **hemizygous** for X-linked traits because he has only one set of X-linked genes.



X-Linked Recessive Inheritance

: **Criteria for an X-Linked Recessive Trait**

1. Always expressed in the male.
2. Expressed in a female **homozygote** and very rarely in a heterozygote.
3. Affected male inherits a trait from **heterozygote or homozygote mother.**
4. Affected female inherits a trait from affected father and affected or heterozygote mother.

- A daughter can inherit an X-linked recessive disorder or trait if her father is affected and her mother is a carrier because the daughter inherits one affected X chromosome from each parent.
- Without a biochemical test, though, an unaffected woman would not know she is a carrier for an X-linked recessive trait unless she has an affected son.
- A woman whose brother has hemophilia B has a 1 in 2 risk of being a carrier. Both her parents are healthy, but her mother must be a carrier because her brother is affected. **Her risk** is the chance that she has inherited the X chromosome bearing the hemophilia allele from her mother.
- The chance of the woman conceiving a son is 1 in 2, and of that son inheriting hemophilia is 1 in 2. Using the product rule, the risk that she is a carrier and will have a son with hemophilia, out of all the possible children she can conceive, is 1/2 × 1/2 × 1/2, or 1/8.

2- X-Linked Dominant Inheritance

Criteria for an X-Linked Dominant Trait

1. Expressed in females in one copy
2. Much more severe effects in males
3. High rates of miscarriage due to early lethality in males
4. Passed from male to all daughters but no sons

- A female who inherits a dominant X-linked allele or in whom the mutation originates has the associated trait or illness, but a male who inherits the allele is usually more severely affected because he has no other allele to mask its effect.
- The children of a normal man and a woman with a dominant, disease-causing allele on the X chromosome face the risks.

- For example in a severe condition, such as **Rett syndrome**, females may be too disabled to have children. Rett syndrome and similar X-linked dominant conditions, therefore, affect only girls, because sons would have to inherit the X chromosome bearing the mutation from their affected mothers.
- Another X-linked dominant condition, congenital generalized **hypertrichosis,** produces many extra hair follicles, and hence denser and more abundant upper body hair .
- Hair growth is milder and patchier in females because of hormonal differences and the presence of a second X chromosome.
- The affected man in the pedigree passed the trait to all four daughters, but none of his nine sons. Because sons inherit the X chromosome from their mother, and only the Y from their father, they could not have inherited the hairiness from their father.