L # 4: Genetics: Basic Principles

What is Genetics?

Genetics is the study of heredity and the variation of inherited characteristics. In dentistry, understanding genetic principles is essential for diagnosing, managing, and preventing oral diseases with a genetic basis.

Important Definitions

1- Chromatin:

The uncoiled, thread-like structure of DNA and proteins. It is found during interphase (G1, S, G2 phases). Not visible under a light microscope.

2- Chromosome

A highly condensed and organized structure of DNA carrying genetic information. Visible during metaphase and anaphase; returns to chromatin in telophase. Visible under a microscope during mitosis or meiosis.

In humans, there are 23 pairs of chromosomes (46 total), with one set inherited from each parent. These include 22 pairs of autosomes and one pair of sex chromosomes (XX for females and XY for males). Chromosomes are visible under a light microscope during cell division when they condense and become tightly coiled.

3- Chromatid:

A single chromosome strand has been copied and is part of a sister chromatid pair. It is found during mitosis and meiosis.

4- Locus

A **locus** (plural: loci) refers to the specific, fixed position on a chromosome where a particular gene or genetic marker is located. Think of a chromosome as a long map, and the locus is like an exact address on that map indicating where a gene resides. The locus is identified by the chromosome number, the arm (p for short arm, q for long arm), and the specific region, band, and sub-band numbers.

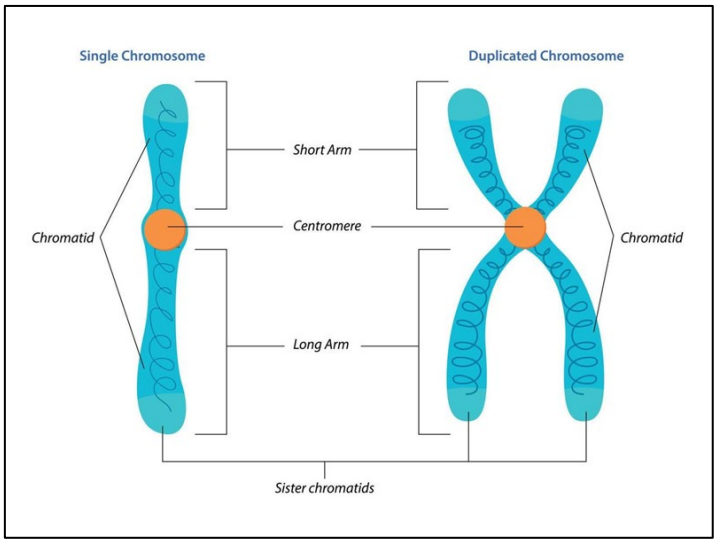
Key Features of a Locus:

- **Specificity**: Each gene occupies a unique locus on a chromosome.
- **Notation**: Described using a standardized nomenclature (e.g., 7q31.2 denotes chromosome 7, long arm, region 3, band 1, sub-band 2).
- **Importance**: Identifying loci is crucial for genetic mapping, studying inheritance patterns, and diagnosing genetic disorders.

Example to Illustrate the Concepts:

- **Gene Location**: The gene responsible for cystic fibrosis, CFTR, is located on the **long arm** of chromosome **7** at position **q31.2**. So, its locus is **7q31.2**.
- **Chromosome**: Chromosome 7 is one of the 22 pairs of autosomes in humans and carries many genes essential for normal development.

Figure 1: locus on chromosome



Genotype vs. Phenotype

Definitions

- **Gene**: A segment of DNA that contains coding information for a protein or RNA molecule.
- **Allele**: Different versions of a gene found at the same locus on homologous chromosomes.

Relationship Between Genotype and Phenotype

- **Genotype**: The genetic makeup of an organism; the combination of alleles for a given gene.
- **Phenotype**: The observable characteristics or traits resulting from the interaction of the genotype with the environment.

In dentistry, variations in genotypes can influence tooth development, enamel formation, and susceptibility to oral diseases.

Mendelian Inheritance

– **Laws of Inheritance**

Gregor Mendel's experiments established foundational principles:

1. **Law of Segregation**: Each individual carries two alleles for each gene, which segregate during gamete formation.
2. **Law of Independent Assortment**: Genes for different traits are inherited independently of each other.

– **Punnett Squares**

A Punnett square is a diagram that predicts the genotype and phenotype combinations in genetic crosses.

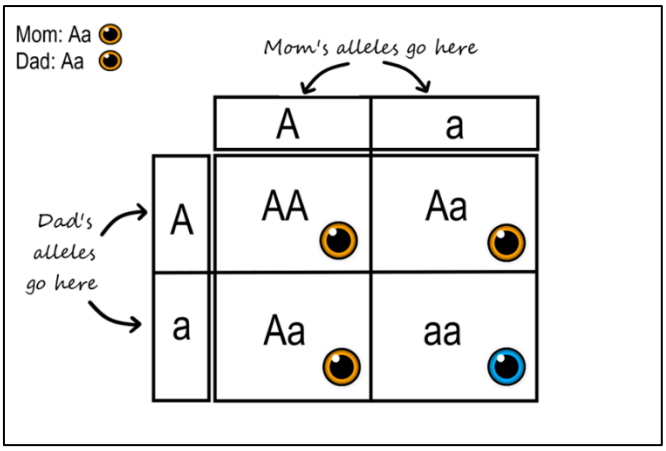


Figure 2: Punnett Square for a Monohybrid Cross—illustrating the potential genotypes resulting from a cross between heterozygous parents for a single trait.

Non-Mendelian Inheritance Patterns

a) Incomplete Dominance

The heterozygote displays a phenotype intermediate between both homozygotes.

b) Codominance

Both alleles in the heterozygote are fully expressed.

c) Multiple Alleles

More than two allele forms exist for a gene within a population (e.g., ABO blood groups).

d) Polygenic Traits

Traits influenced by multiple genes, such as tooth size and shape.

e) Mitochondrial Inheritance

Genes transmitted through mitochondrial DNA, inherited maternally. Mitochondrial mutations can affect cellular energy and impact oral tissues.

f) Epigenetics

Heritable changes in gene expression without altering DNA sequence, often through DNA methylation or histone modification.

Review Questions Of Lecture Notes #3

1. Define genetics and explain its relevance to dentistry.
2. What is a chromosome, chromatid and chromatin?
3. Explain the difference between genotype and phenotype.
4. How are chromosomes organized in humans, and what is the significance of the 23 pairs?
5. Describe the concept of a locus and its role in genetic mapping. Give an example with a specific gene and its locus.
6. What are Mendel's Laws of Inheritance? How are they applied to predict genetic conditions?
7. What distinguishes Mendelian from non-Mendelian inheritance patterns?
8. Explain the genetic and environmental factors involved in cleft lip and palate. How does this condition impact oral health?
9. What role does mitochondrial inheritance play in oral health, and how does it differ from nuclear inheritance?
10. Define epigenetics and discuss how it might influence gene expression.