CLIL

Genetics

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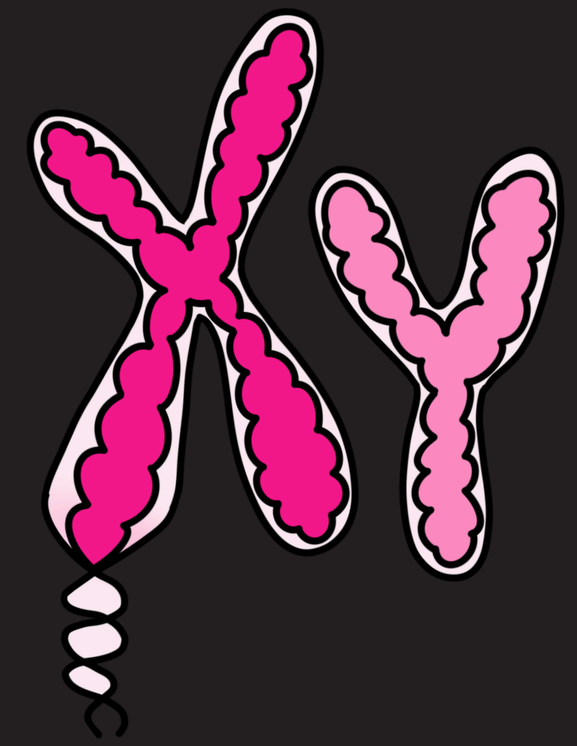
The chromosome theory of inheritance

- ***Mendel's "factors"*** were initially seen as abstract (no knowledge of chromosomes or nucleus)
- ***Walther Flemming*** (1882): observed mitosis and identified chromosomes
- ***Meiosis*** showed chromosome behavior matching Mendelian segregation
- ***Walter Sutton*** (1903): proposed genes are located on chromosomes (Chromosome Theory)
- Later discoveries confirmed chromosomes as carriers of ***heredity***

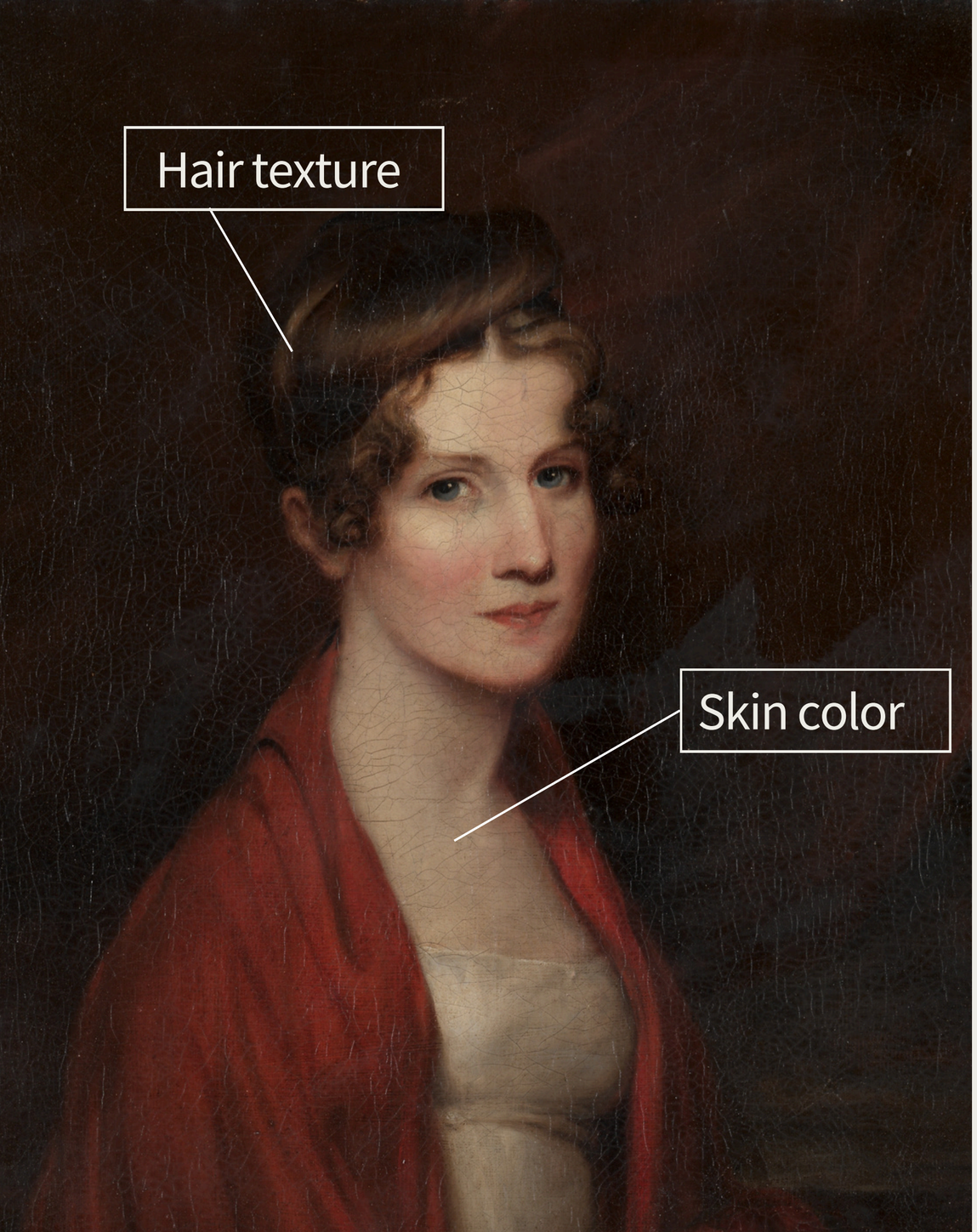
The determination of sex

* ***Females*** (XX), ***Males*** (XY) → Y determines male development
* ***X chromosome***: large, many genes
  Y chromosome: small, few genes
* ***Hemizygosity***: males have only one allele for X-linked genes
* Explains higher frequency of ***X-linked disorders*** in males



Sex-linked charcteristics: to Drosophila to humans

- ***Thomas Hunt Morgan*** used ***Drosophila melanogaster*** as a model organism (fast life cycle, many offspring, XX/XY system)
- Discovered a ***white-eye mutation*** in male flies (recessive trait)
- Demonstrated that some traits are linked to the ***X chromosome***
- Provided first experimental proof of the ***Chromosome Theory of inheritance***



GENETICS

Family trees are essential for tracing and **illustrating** the hereditary **transmission** of genetic **diseases** through generations.
In the 19th century, **hemophilia** spread through the **descendants** of **Queen Victoria**.
The spread of the disease was caused by **dynastic marriages,** used at the time to seal political alliances between European states.

THOMAS MORGAN AND ALFRED STURTEVANT

T. **Morgan**, studying *Drosophila melanogaster*, discovered that Mendel's rule does not apply to genes located on the same chromosome. During meiosis, crossing over allows the **physical exchange** of segments between **homologous chromosomes**.
Alfred **Sturtevant** realized that the frequency of crossing over depends on the physical distance between genes on a chromosome.

- **Distant genes** = **more frequent crossing over.**
- **Close genes** = **rare crossing over**

The unit of measurement is the **centiMorgan** (cM), where 1 cM corresponds to a recombination frequency of 1%.

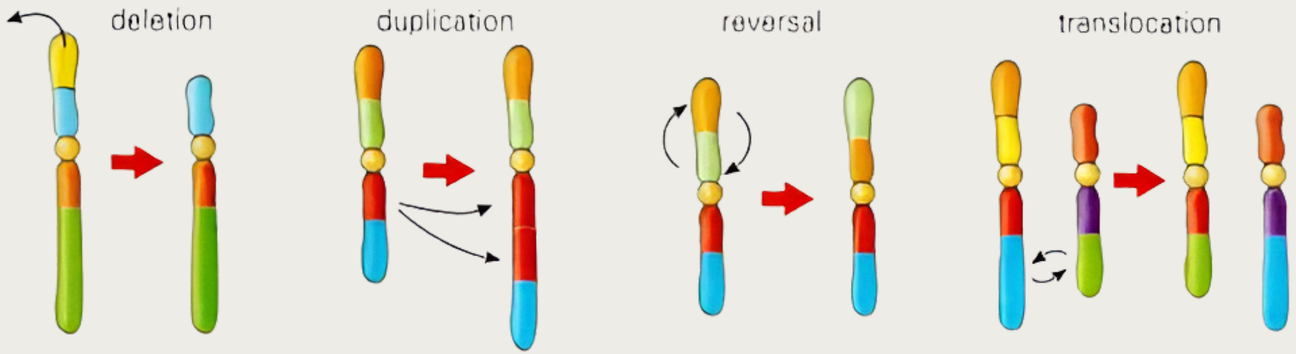
INHERITANCE AND GENERATIONS

Polygenic inheritances are pathologies caused by the combined action of **multiple genes** and **environmental factors**, whose role is crucial for the actual onset of the disease.The genes underlying these diseases are inherited in a **cumulative manner**: the **more defective genes** an individual receives from their parents, the **higher the probability** of developing the disease. Some of these conditions are visible at birth, such as **congenital hip dislocation, cryptorchidism, and cleft lip and palate.**

Anomalies in chromosome structure

These errors during chromosome crossing over can cause structural changes like deletions, duplications, translocations, and inversions. Often the embryos with these genetic defects end in miscarriage, although sometimes they complete development, but lead to the birth of individuals with serious illnesses.

- **deletion**: occurs when a fragment of a chromosome is lost;
- **duplication**: occurs when a piece of chromosome is present twice in the same chromosome;
- **translocation**: occurs when two non-homologous chromosomes exchange fragments;
- **inversion**: occurs when a piece of chromosome first detaches and then reattaches to the chromosome, but is reversed.

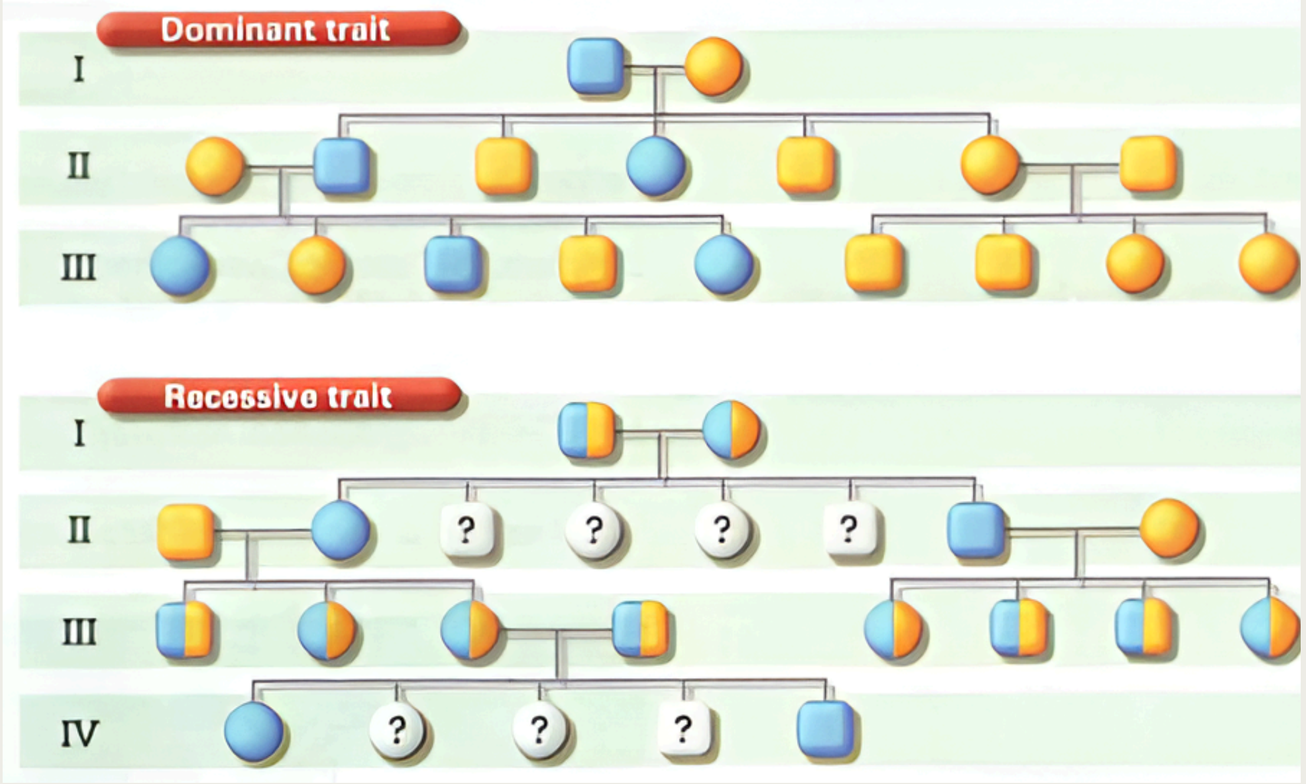


HUMAN GENETICS

Mendel’s laws apply to humans, and while some traits are determined by single genes, most human characteristics are **polygenic**. This means they depend on multiple genes, resulting in continuous variation or varying symptom intensity in genetic diseases.

Studying human genetics is challenging due to ethical limits on experimental methods like selective breeding or induced mutations. Instead, researchers rely on **family trees** (**pedigrees**). These studies help identify patterns of inheritance and genetic disorders caused by mutations. They can be:

- **genetic mutations**, affecting a single gene;
- **chromosomal mutations**, altering the structure of a chromosome:
- **genomic mutations**, that alter the number of chromosomes increasing it by one or more units (**aneuploidy**), or even doubling or tripling (**polyploidy**).



Gene mutations disrupt a single gene and lead to **hereditary diseases**, as carriers pass the defective gene to their descendants following Mendelian inheritance rules. These diseases are categorized based on the faulty gene, such as **recessive autosomal**, **recessive heterosomal** and **dominant**.

In contrast, chromosomal mutations and aneuploid genomic ones, result in **chromosomal anomalies** that do not follow Mendelian inheritance patterns.

While genomic mutations that multiply the entire genome, are typically incompatible with life in animals, they may occur in plants.

DISEASES

Hereditary diseases can be autosomal recessive, sex-linked recessive, or dominant. **Autosomal recessive diseases** are caused by a defective recessive allele on a non-sex chromosome. The risk is higher in children of closely related parents. **Sex-linked recessive diseases** are caused by a recessive allele on the X chromosome; **Dominant diseases** are caused by a dominant defective allele, so both AA and Aa individuals are affected; if an affected parent is heterozygous, each child has a 50% chance of inheriting the disease.
The genetic contribution to a trait is called heritability.

ANORMALIES

Chromosomal anomalies are abnormalities caused by errors during meiosis that affect whole chromosomes or large chromosome segments.These anomalies are divided into numerical abnormalities and structural abnormalities. **Numerical abnormalities** occur when homologous chromosomes fail to separate correctly during meiosis, a process called non-disjunction. An important exception is **Down syndrome**, which is caused by an extra copy of chromosome 21.
Abnormalities can also affect the sex chromosomes. **Turner syndrome** occurs in females who have only one X chromosome. **Klinefelter syndrome** occurs in males with an extra X chromosome.

Thank you for your attention!