Chapter 14: Human Heredity

Class	Biology

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14.1 — Human Chromosomes

Karyotypes

Human cells are not dissimilar from other animals.

- Genetic instructions (**genome**) is different

Genome: full set of genetic information that an organism carries in its DNA

- Cells are photographed during mitosis to easily see condensed chromosomes
- Chromosomes are cut out and arranged to form an image called a *karyotype*

Karyotype → shows the complete diploid set of chromosomes grouped together in pairs, arranged in order of decreasing size

- Contains 46 chromosomes in 23 pairs (one from the sperm and one from the egg in each pair)

Autosomal & Sex Chromosomes

Sex Chromosomes → 2 of the 46 chromosomes in the human genome

- Determine an individual's gender
  - Females → XX
  - Males → XY
    - Ensures approximately half of offspring are male and half are female
- Y chromosome is a lot smaller than the X chromosome
  - X Chromosome → 1200+ genes
  - Y Chromosome → 140

Autosomes / Autosomal Chromosomes → rest 44 chromosomes

Transmission of Human Traits

Most human traits follow Mendel's *law of dominance*.

- Certain alleles of genes are dominant and others are recessive

- Codominance & Multiple Alleles → multiple alleles are showcased in offspring

Sex-Linked Inheritance

Genes on the X and Y chromosomes are **sex-linked**.

- Located on sex chromosomes
- Genes found on the Y chromosome are only found in males (passed directly from father to son)

Some traits are dominant and other are recessive.

X-Chromosome Inactivation

In female cells, most of the genes in female cells are *randomly* switched off.

- **Barr Body:** dense region in the nucleus of randomly switched off genes

Human Pedigrees

Pedigree → shows the presence/absence of a trait according to the relationship between parents, siblings, and offspring

- Applying principles of Mendelian genetics to humans
- Determine the nature of genes

14.2 — Human Genetic Disorders

From Molecule to Phenotype

Genes influence phenotype → Errors are very costly

- Changes in genetic sequence directly influence the amino acid chains that lead to a phenotype

Disorders Caused by Individual Genes

Sickle Cell Disease → defective allele for beta-globin (makes it more soluble)

- Molecules clump into fibers to make a sickle-shaped blood cell

- Sickle cells get stuck in the capillaries

Cystic Fibrosis (CF) → deletion of three bases for the protein *cystic fibrosis transmembrane conductance regulator (CTFR)*

- CFTR normally allows Cl^- ions to pass across cell membranes
- Amino acid is removed → Improper folding
- Tissues throughout the body malfunction without access to Cl^- ions
- Recessive disorder that is very harmful in children

Huntington's Disease → dominant allele for a protein in brain cells

- Mental deterioration and uncontrollable movements
- More codon repeats → earlier disease appears, more severe symptoms

Sickle cell disease & CF were helpful to combat other disease and were genetic advantages **AT TIMES**.

Chromosomal Disorders

Nondisjunction: chromosomes failing to separate during meiosis → disorders in chromosome numbers

- Trisomy → when individuals have three copies of a chromosome as some copies dont separate (ex. Down syndrome)
- Nondisjunction of X chromosomes → Turner's syndrome (females have one X chromosome)
- Extra X chromosome in males → Klinefelter's syndrome (babies have never been born without an X chromosome)

14.3 — Studying the Human Genome

Manipulating DNA

Scientists can **READ** DNA base sequences by cutting, separating, and replicating DNA.

- Allows for the study of genomes in living organisms

Cutting DNA

> DNA molecules are **too large** to be read as a whole, so they must be separated.

- **Restriction Enzymes:** enzymes produced by bacteria that cut DNA into precise pieces (*restriction fragments*)

Separating DNA

Gel Electrophoresis → separates and analyzes differently-sized fragments of DNA

1. Mixture of DNA fragments is placed at the end of a porous gel
2. Electric voltage is applied
3. Negative DNA molecule move to the positive end
    - Smaller molecules move farther

Gel electrophoresis produces bands of DNA based on fragment size (made visible by staining).

Reading DNA

1. Single-stranded DNA fragments are placed in a test tube with DNA polymerase
2. Chemical dyes are attached
    - Produces color-coded DNA fragments of different lengths
        - Order of bands → nucleotide sequence of DNA

The process is automated and computer-controlled → DNA sequencing can read thousands of bases in a few seconds

The Human Genome Project

Human Genome Project: 13-year international project to sequence all DNA and identify human genes

- Developed technology to support genetic research
- Explored gene function
- Studied human variation

1. Researchers break up the genome into manageable pieces
2. Regions are used as markers

Sequencing & Identifying Genes

“Shotgun Sequencing” → cutting DNA into random fragments

- Determines base sequence of each fragment
- Puts fragments together by linking their overlapping areas

> Once **promoters** are located, *genes* can be discovered.

Comparing Sequences

Most DNA matches base-to-base.

- 1 base in 1200 between two people will **NOT** match
  - Single base differences → **Single Nucleotide Polymorphisms (SNPs)**

Certain sets of SNPs are closely linked together many times.

- **Haplotypes** (*haploid genotypes*)**:** collections of linked SNPs
  - International HapMap Project → works to identify haplotypes associated with diseases and conditions

Sharing Data

> The **Human Genome Project** ended in 2003.

- Human genome and other organisms' genomes are freely found on the Internet
- DNA sequences acan be studied online for free

Bioinformatics: the creation, development, and operation of databases and other computing tools to collect, organize, and interpret data about life science

- Computer programs were needed to identify the specific sequences
- Led to **genomics** (the study of whole genomes, including genes and their functions)

Future of the Human Genome Project

Only 2% of the human genome codes for proteins.

- Most chromosomes are large areas with few genes.

> The Human Genome Project raises many ethical questions.

- Who owns and controls genetic information?
- Who should have access to personal genetic information?

Treatments can be specialized to genomes to fight disease best.

Beyond DNA

DNA is not the only regulator of phenotype.

- Specific enzymes regulate gene expression (enzymes are affected by environmental factors)
- Some genes are imprinted to be “silenced”