Genetics

- Is the science of heredity. It includes the study of genes: how they carry information, how they replicate and pass to subsequent generation of cells or between organisms, and how the expression of their information within an organism determines its characteristics.

Vocabulary

- **Genome** – genetic information in a cell, includes chromosomes and plasmids
- **Gene** – a segment of DNA (usually) that codes for a functional product
- **Short tandem repeats (STRs)** – noncoding regions in genome
- **Genomics** – sequencing of genome

Central dogma of biology

- Francis Crick - 1956
- Genes are expressed when the functional product (i.e. protein) has been produced
More vocabulary

• **Genotype** – genetic makeup of an organism
  – Most bacteria are haploid
  – So, any mutation will be expressed

• **Phenotype** – expressed properties
  – Environmental factors play a role in determining what genes are turned on or off, for example . . .
  – milk in media can induce capsule formation
  – low nutrient content can cause sporogenesis

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**Prokaryotic chromosome**

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**Flow of genetic information**

**Horizontal vs. vertical gene transfer**

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**DNA Replication**

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Fig. 2.4 (Silverthorn)

DNA replication is semiconservative

DNA strands are antiparallel

Fig. 8.3

DNA replication

Fig. 8.5
**DNA replication**

- **Helicase** unwinds the two DNA strands
- **Topoisomerase (gyrase)** relaxes the supercoiling of the DNA
- **DNA polymerase** builds the complementary DNA strand
- **Primase** builds short RNA primers
- **Ligase** joins nucleotides together

**Bidirectional replication**

**Prokaryotic protein synthesis**

- **Transcription**
- **Translation**

**Transcription**
The genetic code

Fig. 8.8

Transfer RNA

Anticodon: Binds to codon on mRNA, following complementary base-pairing rules.

Translation

Fig. 8.9

Translation

Fig. 8.9

1 Components needed to begin translation come together.

2 On the assembled ribosome, a tRNA carrying the first amino acid is paired with the start codon on the mRNA. A tRNA carrying the second amino acid approaches.

3 The second codon of the mRNA pairs with a tRNA carrying the second amino acid at the A site. The first amino acid joins to the second by a peptide bond. This tRNA then moves to the P site.

4 The ribosomes move along the mRNA until the second tRNA is in the P site. The next codon to be translated is brought into the A site. The first tRNA now occupies the E site.
Control of gene expression

• Constitutive

• Pre – transcriptional control

• Post – transcriptional control
Pre–transcriptional control

- **Induction**
  - lac operon (E. coli)
  - off when no lactose
- **Repression**
  - trp operon (E. coli)
  - turned off in response to the overabundance of an end product of some metabolic pathway

**Inducible operon**

Fig. 8.12

**Inducible (lac) operon**

Fig. 8.12
Repressible operon

Operon

Control region Structural genes

DNA

Regulatory gene Promoter Operator

Structure of the operon. The operon consists of the promoter (P) and operator (O) sites and structural genes that code for the protein. The operon is regulated by the product of the regulatory gene (I).

Fig. 8.13

Repressible (trp) operon

Positive control – lac operon

Expression of the lac operon is also dependent on glucose levels in the medium.

Fig. 8.13

Expression of the lac operon is also dependent on glucose levels in the medium.
Epigenetic control

- Eukaryotic and bacterial cells can turn genes off via methylation of specific nucleotides

- The methylated (off) genes are passed to offspring, but the genes can be turned on again

Post – transcriptional control

- In eukaryotic cells, micro RNAs (miRNAs) inhibit protein production
  - Allows human cells to make different proteins

- In bacterial cells, miRNAs enable the cell to cope with environmental stress

(a) Lactose present, glucose scarce (cAMP level high). If glucose is scarce, the high level of cAMP activates CAP, and the lac operon produces large amounts of mRNA for lactose digestion.

(b) Lactose present, glucose present (cAMP level low). When glucose is present, cAMP is scarce, and CAP is unable to stimulate transcription.
Changes in genetic material

- Mutations
- Horizontal gene transfer

Mutations

- A mutation is a permanent change in the base sequence of DNA
- This may cause a change in the product encoded by the gene
- The mutation may convey an advantage or disadvantage to the organism

Types of mutations

- Silent
- Base substitution (point mutation)

![Fig. 8.17](image1)

Changes in genetic material

![Fig. 8.18](image2)
Mutagens

- Are agents that can bring about mutations
- Nitrous acid

![Fig. 8.19](Image)

Mutagens

- Nucleoside analogs

![Fig. 8.20](Image)

Mutagens

- Aflatoxin
  - *Aspergillus flavus*, which grows on grains
  - Causes a frameshift mutation

Mutagens

- Radiation
  - X and gamma rays change electron distributions and the resulting ions and free radicals are very reactive
  - Some ions oxidize bases in DNA, resulting in errors in DNA replication and repair
  - Breakage of bonds in the sugar-phosphate backbone of DNA, which can cause chromosomal abnormalities
UV radiation

- Formation of thymine dimers when exposed to UV light (260 nm)
- Enzymes like photolyases, endonucleases, exonucleases, ligases, etc. aid in the repair

Mutations

- Spontaneous mutation rate is low, but mutagens will increase that rate by a factor of 10 to 1000 times
- Essential for adaptation of species to surrounding environment

Identifying mutants

- Positive (direct) selection
  - Detection of mutant cells by selecting against unmutated (parent) cells
  - i.e. Penicillin resistance
- Negative (indirect) selection
  - Selects for cells that can not perform a specific function
  - Use of replica plating

Replica plating
Ames test
• A means by which bacteria can be used to detect carcinogens
• Exposure of mutant bacteria to a mutagen may cause new mutations that change the phenotype
• Used to evaluate pollutants

Genetic recombination
• Refers to the exchange of genes between two DNA molecules to form new combinations of genes on a chromosome

Recombination and bacteria
• Vertical gene transfer — genes are passed from parent to offspring
• Horizontal gene transfer — bacteria can also pass their genes laterally, to other microbes of the same generation
  — Involves a donor cell that gives a portion of its total DNA to a recipient cell

Bacterial genetic transfer occurs via . . .
• Transformation
• Conjugation
• Transduction
Transformation in bacteria

- Genes are transferred from one bacterium to another as “naked” DNA in solution.

Conjugation

- Mediated by plasmids
  - A plasmid is a circular piece of DNA that replicates independently from the cell’s chromosome
  - Plasmids carry genes usually not essential for growth of the cell under normal conditions
- Requires direct cell-to-cell contact
- Conjugating cells must be of opposite mating types
  - Donor cells contain plasmid, recipient cells usually do not
In gram-negative bacteria, the plasmid carries genes that promote the formation of sex pili on the donor cell.

Genetic map of *E. coli* chromosome

Bacterial DNA is transferred from a donor cell to a recipient inside a bacteriophage (a virus that infects bacteria).
Plasmids

- Plasmids are self-replicating, gene-containing, circular pieces of DNA about 1 – 5% the size of the bacterial chromosome
- Found mainly in bacteria
- Under certain conditions genes carried by plasmids can be crucial to the survival and growth of the cell

Types of plasmids

- Conjugative – i.e. F plasmid
- Dissimilation – code for enzymes that trigger the catabolism of unusual sugars and hydrocarbons
- Some code for proteins that enhance the pathogenicity of a bacterium
- Resistance factors (R factors)

R factor

- R factors carry genes that convey resistance to antibiotics, heavy metals, or cellular toxins
- Resistance transfer factors (RTF) – plasmid replication & conjugation
- r-determinant – resistance genes

Transposons

- Small segments of DNA that can move from one region of a DNA molecule to another