Chapter 5
Raw material: heritable variation among individuals

Proteins are chains of amino acids

DNA codes for protein

Eukaryotic DNA is organized into chromosomes

Chromosomes come in homologous pairs

Ploidy can vary

Ploidy: Number of copies of unique chromosomes in a cell
Production of protein from DNA requires transcription and translation

Gene expression: process by which information from a gene is transformed into product

Ribosomes translate mRNA into protein

Gene expression can be regulated in a number of ways

RNA splicing can create multiple proteins from a single gene

Regulation of gene expression is flexible

Non-coding regions make up most of the genome

- Non-coding regions include:
  - RNA genes
  - Pseudogenes
  - Mobile genic elements
microRNA can affect phenotypes

Petunia

Snapdragon

Wild Type

Mutant

Variation in genome size and complexity

<table>
<thead>
<tr>
<th>Organism</th>
<th>Classification</th>
<th>Number of Chromosomes</th>
<th>MR in Genome</th>
<th>Approximate Number of Proteins Coding Genes</th>
</tr>
</thead>
<tbody>
<tr>
<td>A. fava</td>
<td>Liliaceae</td>
<td>1</td>
<td>18</td>
<td>1500</td>
</tr>
<tr>
<td>A. lutea</td>
<td>Liliaceae</td>
<td>1</td>
<td>18</td>
<td>1500</td>
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<tr>
<td>A. officinalis</td>
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<td>1</td>
<td>18</td>
<td>1500</td>
</tr>
<tr>
<td>A. thaliana</td>
<td>Brassicaceae</td>
<td>5</td>
<td>18</td>
<td>26,000</td>
</tr>
<tr>
<td>N. tabacum</td>
<td>Solanaceae</td>
<td>12</td>
<td>18</td>
<td>32,000</td>
</tr>
<tr>
<td>N. vectorii</td>
<td>Solanaceae</td>
<td>12</td>
<td>18</td>
<td>32,000</td>
</tr>
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<td>A. sativus</td>
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<td>18</td>
<td>32,000</td>
</tr>
</tbody>
</table>

Most variation in size due to differences in mobile genetic elements

Key Concepts

• Most proteins function in four ways:
  – Enzymes
  – Cell-cell signaling
  – Receptors
  – Structural elements

• Mutations are the raw material for evolution
  – In diploid organisms, deleterious mutations may be masked by a functional gene copy

Key Concepts

• All cells use mRNA to carry genetic information
  – Some viruses use RNA instead of DNA for the genome
• Non-coding RNA plays critical roles in gene regulation

Types of mutation

Different types of mutation can alter the phenotype

<table>
<thead>
<tr>
<th>Location of Mutation</th>
<th>Type of Mutation</th>
<th>Consequence for Gene Action</th>
</tr>
</thead>
<tbody>
<tr>
<td>Coding Region</td>
<td>Substitution</td>
<td>Alters the product of the gene, and thus its function or activity.</td>
</tr>
<tr>
<td></td>
<td>Insertion</td>
<td>Alters the expression level of the gene.</td>
</tr>
<tr>
<td></td>
<td>Deletion</td>
<td>Alters the expression level of the gene.</td>
</tr>
<tr>
<td></td>
<td>Duplication</td>
<td>Alters the expression level of the gene.</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Intragenic Regulatory Regions</th>
<th>Mutation to coding region of transcription factor</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Alters the binding affinity and the activity of a transcription factor.</td>
</tr>
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<th>Intragenic Regulatory Regions</th>
<th>Mutation to site in transcription regulatory regions</th>
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<tbody>
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<td>Alters the expression level of the gene.</td>
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<th>Mutation to site in trans-regulatory regions</th>
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<th>Physiological</th>
<th>Phenotypic</th>
<th>Consequence for Gene Action</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mutations</td>
<td>Alleles</td>
<td>Alters the expression level of the gene.</td>
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</tbody>
</table>
Examples of point mutations

Germ line mutations are heritable

- **Somatic mutations**: affect cells in the body of an organism; not heritable
- **Germ-line mutations**: affect gametes; heritable and relevant to evolution

Key Concepts

- Changes in coding sequences and gene expression are heritable
- Gene expression changes can affect when, where, and how much a gene is expressed

Recombination generates variation

Independent assortment ensures novel combinations of alleles

Key Concepts

- Meiosis generates considerable genetic variation
  - Recombination
  - Independent assortment
Linking genotype and phenotype

- **Genotype**: the genetic make-up of an individual
- **Phenotype**: an observable measurable characteristic of an organism

Simple polymorphisms can produce differences in phenotype

Sometimes a single genotype can produce multiple phenotypes

Polyphenic trait: single genotype produces multiple phenotypes depending on environment

Quantitative traits influenced by genes and the environment

Francis Galton (1822-1911)

Quantitative traits influenced by multiple genes; generate a normal distribution

Human height has genetic component

QTL analysis can help discover genes influencing quantitative traits
Key Concepts

- Polyphenisms often result from a developmental threshold mechanism
- Continuously varying traits are called quantitative traits
- Evolutionary biologists study variation in the expression of phenotypic traits
  - Caused by genetic and environmental factors

Environmental influences on gene expression

- **Phenotypic plasticity**: changes in phenotype produced by a single genotype in different environments
  - Tailors organism to environment

Key Concept

- Gene expression often influenced by signals from the environment
  - Allows match to environmental circumstances