Chapter 5
Complex Patterns of Inheritance
Phenotypes Can Be Discontinuous or Continuous

- **Discontinuous variation** shows distinct phenotypes
  - Short and tall peas phenotypes

- **Continuous variation** shows a series of overlapping phenotypic classes
  - Height in humans
Continuous and Discontinuous Variation

Fig. 5.2
Continuous Variation in Humans

Fig. 5.1
Genotype + Environment

Produce the Phenotype

\[ P = G + E \]
Terms

• **Polygenic traits** are determined by two or more genes

• **Multifactorial traits** are controlled by two or more genes and show significant interaction with the environment

• **Complex traits** are ones where relative contribution of genes and environment are not yet established
Polygenic Inheritance

- Traits are usually quantified by measurement
- Two or more genes contribute to the phenotype
- Phenotypic variation varies across a wide range
- Better analyzed in populations than in individuals
- Example: human eye color
- As the number of loci increases, the number of classes increases.
- As classes increase, phenotypic difference between classes decreases.
- Averaging out of the phenotype is called **regression to the mean**.

Fig. 5.5
Multifactorial Traits

- Genotype does not change after fertilization (except by mutation)

- Phenotype is the sum of the observable characteristics and may change throughout life

- Environment includes all genetic and nongenetic factors
Characteristics of Multifactorial Traits

- Polygenic
- Genes controlling trait act additively
- Environmental factors interact with the genotype to produce the phenotype
- Assessing interactions can be difficult
Methods Used to Study Multifactorial Traits

• **Threshold model**
  Frequency of disorder among relatives is compared with the frequency of the disorder in the general population

• **Recurrence risk**
  Estimates the risk that the disease will recur
### Table 5.1 Familial Risks for Multifactorial Threshold Traits

<table>
<thead>
<tr>
<th>Multifactorial Trait</th>
<th>Risk Relative to General Population</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>MZ Twins</td>
</tr>
<tr>
<td>Clubfoot</td>
<td>300×</td>
</tr>
<tr>
<td>Cleft lip</td>
<td>400×</td>
</tr>
<tr>
<td>Congenital hip dislocation (females only)</td>
<td>200×</td>
</tr>
<tr>
<td>Congenital pyloric stenosis (males only)</td>
<td>80×</td>
</tr>
</tbody>
</table>
Phenotypic Variation

Sources of phenotypic variation
• Genotypes in the population
• Variation in the environment

**Heritability** – how much of the observed phenotypic variation is due to differences in genotype
Factors that Contribute to Phenotypic Variance

- **Genetic variance**
  Variance attributed to the genotypic differences

- **Environmental variance**
  Variance attributed to differences in the environment

- **Correlation coefficients**
  Measure the degree to which variables vary together
Heritability of Fingerprints

Table 5.2 Correlations between Relatives for Total Ridge Count (TRC)

<table>
<thead>
<tr>
<th>Relationship</th>
<th>Number of Pairs</th>
<th>Observed Correlation Coefficient</th>
<th>Expected Correlation Coefficient between Relatives</th>
<th>Heritability</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mother-child</td>
<td>405</td>
<td>0.48 ± 0.04</td>
<td>0.50</td>
<td>0.96</td>
</tr>
<tr>
<td>Father-child</td>
<td>405</td>
<td>0.49 ± 0.04</td>
<td>0.50</td>
<td>0.98</td>
</tr>
<tr>
<td>Husband-wife</td>
<td>200</td>
<td>0.05 ± 0.07</td>
<td>0.00</td>
<td>—</td>
</tr>
<tr>
<td>Sibling-sibling</td>
<td>642</td>
<td>0.50 ± 0.04</td>
<td>0.50</td>
<td>1.00</td>
</tr>
<tr>
<td>Monozygotic twins</td>
<td>80</td>
<td>0.95 ± 0.01</td>
<td>1.00</td>
<td>0.95</td>
</tr>
<tr>
<td>Dizygotic twins</td>
<td>92</td>
<td>0.49 ± 0.08</td>
<td>0.50</td>
<td>0.98</td>
</tr>
</tbody>
</table>

Twin Studies

- **Monozygotic twins**
  - Single fertilization
  - Genetically identical

- **Dizygotic twins**
  - Independent fertilizations
  - Share approximately half their genes

Fig. 5.10