3.2: Relationships Between Genes, Genotypes and Phenotypes

3.2.1 Terminology

A specific position along a chromosome is called a **locus**. Each gene occupies a specific locus (so the terms locus and gene are often used interchangeably). Each locus will have an allelic form (allele). The complete set of alleles (at all loci of interest) in an individual is its **genotype**. Typically, when writing out a genotype, only the alleles at the locus (loci) of interest are considered – all the others are present and assumed to be wild type. The visible or detectable effect of these alleles on the structure or function of that individual is called its **phenotype** – what it looks like. The phenotype studied in any particular genetic experiment may range from simple, visible traits such as hair color, to more complex phenotypes including disease susceptibility or behavior. If two alleles are present in an individual, then various interactions between them may influence their expression in the phenotype.

3.2.2 Complete Dominance

Let us return to an example of a simple phenotype: flower color in Mendel’s peas. We have already said that one allele as a homozygote produces purple flowers, while the other allele as a homozygote produces white flowers (see Figures 1.8 and 3.3). But what about an individual that has one purple allele and one white allele; what is the phenotype of an individual whose genotype is heterozygous? This can only be determined by experimental observation. We know from observation that individuals heterozygous for the purple and white alleles of the flower color gene have purple flowers. Thus, the allele associated with purple color is therefore said to be **dominant** to the allele that produces the white color. The white allele, whose phenotype is masked by the purple allele in a heterozygote, is **recessive** to the purple allele.
Figure 3.3: Relationship between genotype and phenotype for an allele that is completely dominant to another allele. Original-Deholos (Fireworks)-CC:AN)

To represent this relationship, often, a dominant allele will be represented by a capital letter (e.g. $A$) while a recessive allele will be represented in lower case (e.g. $a$). However, many different systems of genetic symbols are in use. The most common are shown in Table 3.1. Also note that genes and alleles are usually written in *italics* and chromosomes and proteins are not. For example, the *white* gene in *Drosophila melanogaster* on the X chromosome encodes a protein called WHITE.

**Table 3.1: Examples of symbols used to represent genes and alleles.**

<table>
<thead>
<tr>
<th>Examples</th>
<th>Interpretation</th>
</tr>
</thead>
<tbody>
<tr>
<td>$A$ and $a$</td>
<td>Uppercase letters represent dominant alleles and lowercase letters indicate recessive alleles. Mendel invented this system but it is not commonly used because not all alleles show complete dominance and many genes have more than two alleles.</td>
</tr>
<tr>
<td>$a^+$ and $a^1$</td>
<td>Superscripts or subscripts are used to indicate alleles. For wild type alleles the symbol is a superscript +.</td>
</tr>
<tr>
<td>$AA$ or $A/A$</td>
<td>Sometimes a forward slash is used to indicate that the two symbols are alleles of the same gene, but on homologous chromosomes.</td>
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</table>

3.2.3 Incomplete Dominance

Besides dominance and recessivity, other relationships can exist between alleles. In *incomplete dominance* (also called *semi-dominance*, Figure 3.4), both alleles affect the trait additively, and the phenotype of the heterozygote is...
intermediate between either of the homozygotes. For example, alleles for color in carnation flowers (and many other species) exhibit incomplete dominance. Plants with an allele for red petals ($A_1$) and an allele for white petals ($A_2$) have pink petals. We say that the $A_1$ and the $A_2$ alleles show incomplete dominance because neither allele is completely dominant over the other.

![Figure 3.4: Relationship between genotype and phenotype for incompletely dominant alleles affecting petal colour in carnations.](Original-Deholos-CC:AN)

### 3.2.4 Co-Dominance

Co-dominance is another type of allelic relationship, in which a heterozygous individual expresses the phenotype of both alleles simultaneously. An example of co-dominance is found within the ABO blood group of humans. The ABO gene has three common alleles which were named (for historical reasons) $I^A$, $I^B$, and $i$. People homozygous for $I^A$ or $I^B$ display only A or B type antigens, respectively, on the surface of their blood cells, and therefore have either type A or type B blood (Figure 3.5). Heterozygous $I^A I^B$ individuals have both A and B antigens on their cells, and so have type AB blood. Notice that the heterozygote expresses both alleles simultaneously, and is not some kind of novel intermediate between A and B. Co-dominance is therefore distinct from incomplete dominance, although they are sometimes confused.

![Figure 3.5: Relationship between genotype and phenotype for three alleles of the human ABO gene. The $I^A$ and $I^B$ alleles show co-dominance. The $I^A$ allele is completely dominant to the $i$ allele. The $I^B$ allele is completely dominant to the $i$ allele.](Original-Deholos -CC:AN)

It is also important to note that the third allele, $i$, does not make either antigen and is recessive to the other alleles. $I^A i$ or $I^B i$ individuals display A or B antigens respectively. People homozygous for the $i$ allele have type O blood. This is a useful reminder that different types of dominance relationships can exist, even for alleles of the same gene. Many types of molecular markers, which we will discuss in a later chapter, display a co-dominant relationship among alleles.
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