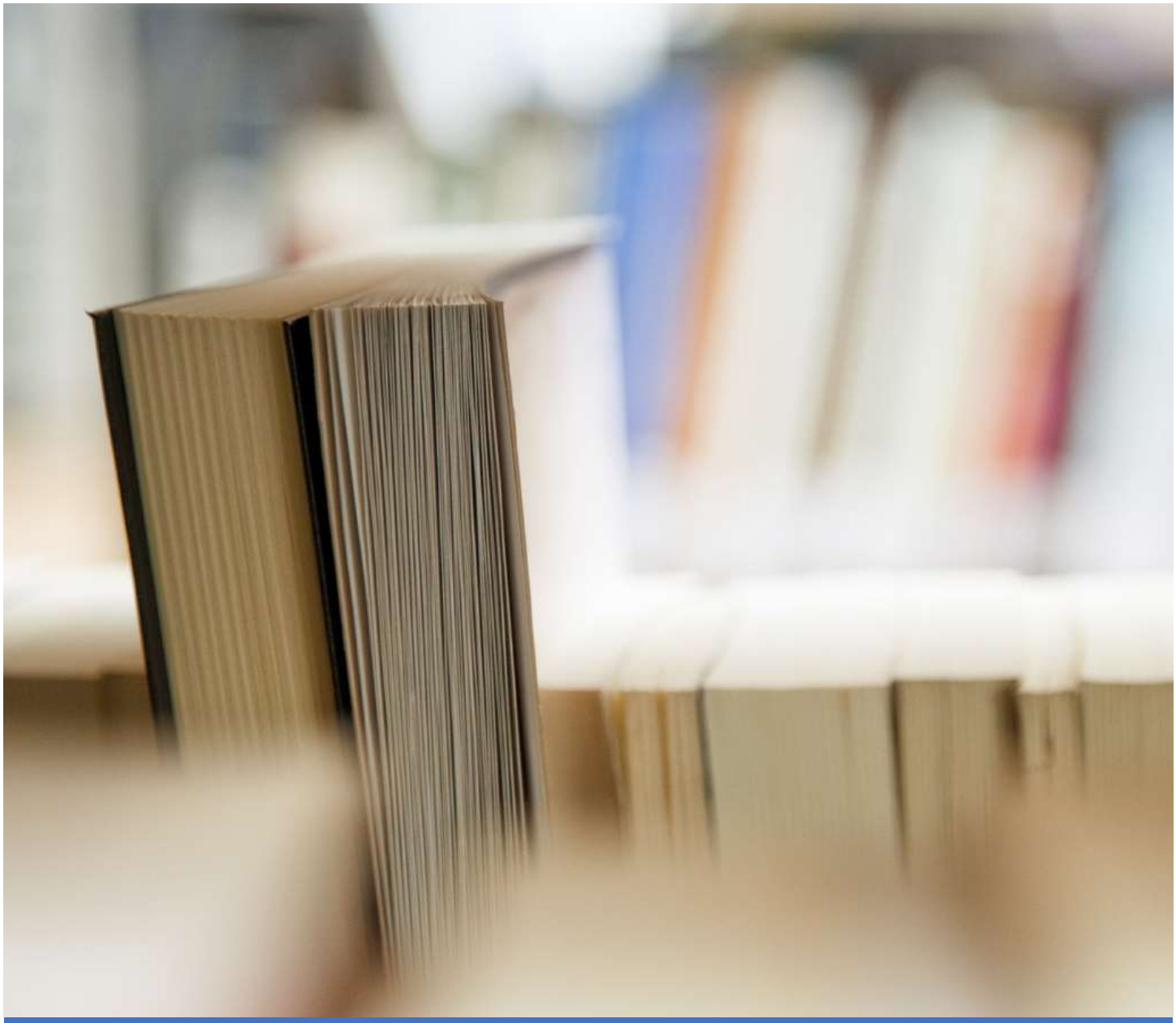

Relationships Between Genes, Genotypes and Phenotypes

USER

BIO 101

3.1: 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.

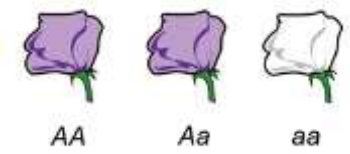

Figure 3.3: *Relationship between genotype and phenotype for an allele that is completely dominant to another allele. Original-Deholos (Fireworks)-CC:AN)*

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.

Examples	Interpretation
A and *a*	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.
a^+ and a^1	Superscripts or subscripts are used to indicate alleles. For wild type alleles the symbol is a superscript +.
AA or *A/A*	Sometimes a forward slash is used to indicate that the two symbols are alleles of the same gene, but on homologous chromosomes.

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.

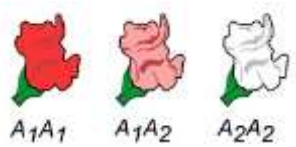

Figure 3.4: *Relationship between genotype and phenotype for incompletely dominant alleles affecting petal colour in carnations.(Original-Deholos-CC:AN)*

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^AI^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.

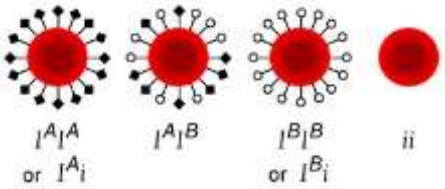

Figure 3.5: *Relationship between genotype and phenotype for three alleles of the human ABO gene. The I^A and I^B alleles show co-dominance. The I^A allele is completely dominant to the i allele. The I^B allele is completely dominant to the i allele. (Original-Deholos - CC:AN)*

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.

Terms Used

alternation of generations

life-cycle type in which the diploid and haploid stages alternate

diploid-dominant

life-cycle type in which the multicellular diploid stage is prevalent

haploid-dominant

life-cycle type in which the multicellular haploid stage is prevalent

gametophyte

a multicellular haploid life-cycle stage that produces gametes

germ cells

specialized cell line that produces gametes, such as eggs or sperm

life cycle

the sequence of events in the development of an organism and the production of cells that produce offspring

sporophyte

a multicellular diploid life-cycle stage that produces haploid spores by meiosis

blending theory of inheritance

hypothetical inheritance pattern in which parental traits are blended together in the offspring to produce an intermediate physical appearance

continuous variation

inheritance pattern in which a character shows a range of trait values with small gradations rather than large gaps between them

discontinuous variation

inheritance pattern in which traits are distinct and are transmitted independently of one another

dominant

trait which confers the same physical appearance whether an individual has two copies of the trait or one copy of the dominant trait and one copy of the recessive trait

F_1

first filial generation in a cross; the offspring of the parental generation

F_2

second filial generation produced when F individuals are self-crossed or fertilized with each other

hybridization

process of mating two individuals that differ with the goal of achieving a certain characteristic in their offspring

model system

species or biological system used to study a specific biological phenomenon to be applied to other different species

P_0

parental generation in a cross

product rule

probability of two independent events occurring simultaneously can be calculated by multiplying the individual probabilities of each event occurring alone

recessive

trait that appears “latent” or non-expressed when the individual also carries a dominant trait for that same characteristic; when present as two identical copies, the recessive trait is expressed

reciprocal cross

paired cross in which the respective traits of the male and female in one cross become the respective traits of the female and male in the other cross

sum rule

probability of the occurrence of at least one of two mutually exclusive events is the sum of their individual probabilities

trait

variation in the physical appearance of a heritable characteristic

chiasmata

(singular, *chiasma*) the structure that forms at the crossover points after genetic material is exchanged

cohesin

proteins that form a complex that seals sister chromatids together at their centromeres until anaphase II of meiosis

crossover

exchange of genetic material between non-sister chromatids resulting in chromosomes that incorporate genes from both parents of the organism

fertilization

union of two haploid cells from two individual organisms

interkinesis

(also, *interphase II*) brief period of rest between meiosis I and meiosis II

meiosis

a nuclear division process that results in four haploid cells

meiosis I

first round of meiotic cell division; referred to as reduction division because the ploidy level is reduced from diploid to haploid

meiosis II

second round of meiotic cell division following meiosis I; sister chromatids are separated into individual chromosomes, and the result is four unique haploid cells

recombination nodules

protein assemblies formed on the synaptonemal complex that mark the points of crossover events and mediate the multistep process of genetic recombination between non-sister chromatids

reduction division

nuclear division that produces daughter nuclei each having one-half as many chromosomes sets as the parental

nucleus; meiosis I is a reduction division

somatic cell

all the cells of a multicellular organism except the gametes or reproductive cells

spore

haploid cell that can produce a haploid multicellular organism or can fuse with another spore to form a diploid cell

synapsis

formation of a close association between homologous chromosomes during prophase I

synaptonemal complex

protein lattice that forms between homologous chromosomes during prophase I, supporting crossover

tetrad

two duplicated homologous chromosomes (four chromatids) bound together by chiasmata during prophase