14.2: Biochemistry of Gene Behavior

The Mechanics of Gene Expression

We often think of genes as “made of DNA”; they reside in the nucleus and endosymbiotic organelles of eukaryotes or the nucleoid region of prokaryotes. As described in Chapter 13, they are transcribed into an RNA message by RNA polymerase then interpreted by ribosomes that assemble particular amino acids into a polypeptide strand (also known as a protein) based on the sequence of nucleotides. In a cell, proteins can act as enzymes, structural features, pigments, and a host of other functions, including regulating the expression of other genes. This expression of genes leads to how an organism looks – its phenotype.

Dominant and Recessive; Homozygous, Heterozygous, and Hemizygous

Alleles themselves do not directly exert an effect on the phenotype of an organism. You will recall from Chapter 3 that genes are instructions, often for proteins. It is the effect of the protein that causes an organism to take on particular traits. In this chapter we will look at how to design symbols appropriate to communicate the characteristics of alleles you want to investigate, but keep in mind that the allele instructs what kind of protein to make.

Chapter 1 points out that organisms usually fall into the classes of being diploid or haploid. Humans and eukaryotic genetic systems usually assume the organism is diploid, which means that most chromosomes are represented as pairs. Each pair has homologous loci: the term homologous means “the same information” and refers to the gene at each locus. Note that although the loci, or genes, are the same, the alleles that comprise them may be different!

If an organism is diploid and homozygous for an allele (Figure A1.2, left and right), the gene at the same position of the homologous chromosomes is the same allele. Only one type of protein is made. If an organism is diploid and
heterozygous (Figure A1.2, middle), and the protein from one allele influences the phenotype more than the protein from the other allele, we use the terms **dominant** and **recessive**, respectively.

![AA, Aa, aa](Image)

**Figure A1.2: Relationship between genotype and phenotype for an allele that is completely dominant to another allele.**

(Original-M. Deyholos -CC:AN – from chapter 13)

If one copy of an allele makes enough protein to compensate for the absence of protein from the other allele in a heterozygote, it will influence the phenotype. If this phenotype looks identical to that of an organism homozygous for the “functional” allele, we consider the “functional” allele to be dominant to the “null”. We could also say the “null” is recessive to the “functional” allele. Keep in mind that the alleles themselves aren’t doing anything, but it’s common practice to label the alleles as dominant or recessive, although in reality we are talking about the expression of those alleles.

Why this is important is how the proteins from two alleles interact. If both proteins are identical (from a homozygous genotype) the phenotype that results will be that of the action of one “type” of protein, even if though there are two copies of the gene – they are both the same allele. There’s no real interaction because the proteins do the same thing. It’s in heterozygotes that we can see whether a particular allele is dominant over another or otherwise influences the phenotype in an interesting way.

Here’s another point about the “normal” allele: it was honed by natural selection over a long period of time. Genes are instructions for the protein tools of an organism’s cells. For this reason we often call the “normal” allele of a gene the “wild type” allele. This would be the allele most common in the wild, presumably because it provides a benefit to the organism. Thus, most mutations are likely to reduce the effectiveness of the wild type allele, although the process of evolution allows (and, in fact, requires) an occasional beneficial allele to permeate a population if it provides a selective advantage.

Note that we do not use the terms homozygous or heterozygous in haploid organisms. If they are haploid, their phenotype will reflect the genotype of the only allele present. The proper term for their genotype is hemizygous to reflect only one copy of each gene.

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**There are Many Kinds of Alleles for a Gene**

A mutation is a change in nucleotide sequence. Chapter 11 goes into more detail of what this means. What’s important now is that you understand that the amino acids of a protein can be different if we compare different alleles of a gene and they may behave differently – often one protein will “work better” than the other. If the promoter of a “functional” allele of a gene is damaged, the allele that is created might not even create an mRNA so no protein will be encoded by that allele. This is called an amorphic or “null” mutant (See Chapter 1).
Keep also in mind that a gene can be mutated in different parts of its sequence to create different alleles. A diploid organism can have a maximum of two alleles (aside from gene duplication or abnormal chromosome structure, but ignore that for now). In a population, though, there can be many, many different alleles. Perhaps allele one decreases protein function and allele 2 is even less effective. This describes an allelic series as follows: wild-type > allele 1 > allele 2 > any null allele. Wild-type alleles in this case encode the most effective protein. Null alleles represent catastrophic mutations that eliminate transcription or produces proteins that can’t function at all. See section 3.2 for another example of an allelic series.

Gender and Gene Interactions

Finally, here are a couple points about other interactions before we move on to gene symbols. Some organisms have a pair of sex chromosomes that dictate gender (see Chapter 4). We'll ignore incorporating ways to communicate if a chromosome is a sex chromosome in this chapter; Appendix 2 will show how we can indicate those using a symbol.