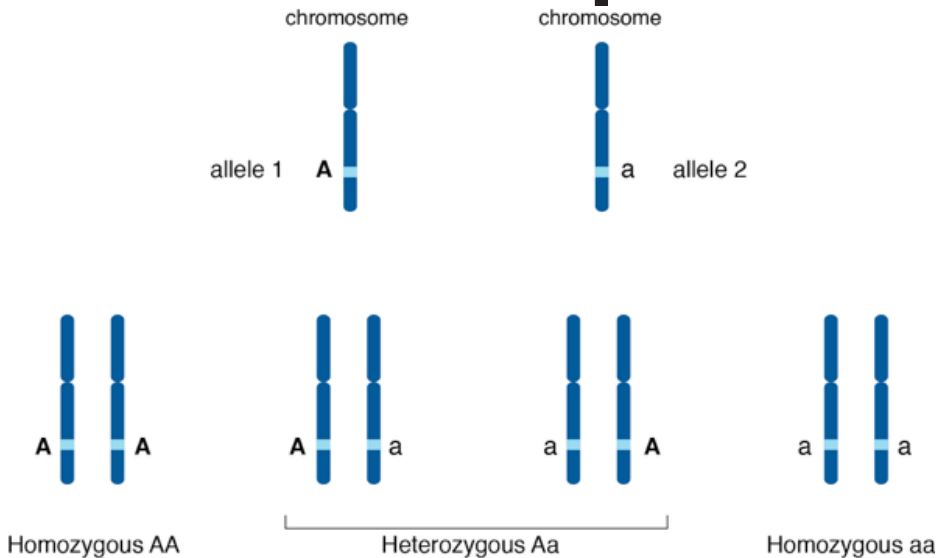


# Chapter 1

## Part 1: Simple Nomenclature



**Figure 1-1**

Two chromosomes with the same gene but different alleles, A and a.

(Image used with permission of [www.genome.gov](http://www.genome.gov) provided it includes the link <http://xoax.net/mediaGallery/Allele-dat-342>)

### INTRODUCTION

Classical genetics requires higher-level thinking and an organized strategy. You have probably encountered symbols in high school genetics and perhaps in previous chapters assigned to you in this course. Chapter 13 points out that a position on a chromosome is a gene locus, which simply means the location at which a gene is found. At that position, the gene could be one of several variants that we call alleles. The set of alleles comprise an organism's genotype, which might affect the organism's phenotype through the action of the genotype.

When you are solving genetics problems you have to keep track of different alleles of one or more genes. This can become confusing, so understanding how a gene works, how alleles vary, how they are expressed, and the influence they have on phenotype is important. A consistent nomenclature system – a set of rules dictating how you name alleles – becomes immensely helpful. Online Open Genetics has an activity that demonstrates why a good nomenclature system is helpful. The icon to the right indicates an online module.

### A WHY USE NOMENCLATURE?

Word and practical problems in biology can get confusing in a hurry, particularly if you're distracted by something like exam stress! When you are manipulating several ideas, it is good practice to be thoughtful and follow rules that keep you consistent in your interpretation. As an instructor, I have seen work in which the student clearly got flustered and forgot that the mutation he or she was working on was dominant. This often leads to an answer that is inconsistent with the data.

There are a few simple rules we can use for nomenclature. Essentially, we use capital letters to indicate an allele that, in a heterozygote, expresses the proteins from that particular allele. This first appendix uses a simplified system to communicate allele characteristics. The next appendix will show a more complicated system that carries even more information in the gene symbols.



<http://tinyurl.com/oog-name>

## B THE BIOCHEMISTRY OF GENE BEHAVIOUR

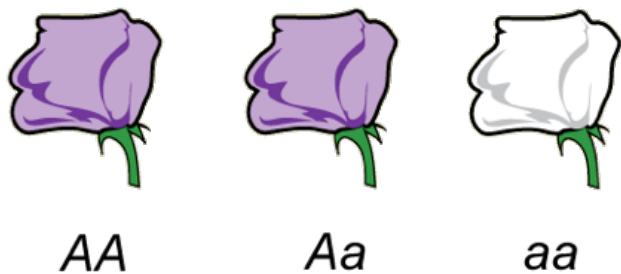
### B.1 THE MECHANICS OF GENE EXPRESSION

The biochemical action of gene expression is in Chapter 3. We often think of genes as “made of DNA” and they reside in the nucleus and endosymbiotic organelles of eukaryotes or the nucleoid region of prokaryotes. 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.

Chapter 2 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!

### B.2 DOMINANT AND RECESSIVE; HOMOZYGOUS, HETEROZYGOUS, AND HEMIZYGOUS

Alleles themselves do not directly exert an effect on the phenotype of an organism. You will recall 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



**Figure 1-2**

Relationship between genotype and phenotype for an allele that is completely dominant to another allele. (Original-M. Deyholos -CC:AN)

make. If an organism is diploid and homozygous for an allele (**Figure 1-2**, 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 1-2**, 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. Note that we don't 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.

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 2).

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.

<http://tinyurl.com/oog-biochem>

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 organ-

ism'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.

### B.3 THERE ARE MANY KINDS OF ALLELES FOR A GENE

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 on page 4 below for another example of an allelic series.

### B.4 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 how we indicate this as a symbol for now; the second part of this chapter will show a trick to indicate this.

Incomplete dominance, codominance, and epistasis are not accommodated in any of the nomenclature systems we present.

## C BASIC NOMENCLATURE

### C.1 SINGLE LETTER SYSTEM

Sometimes what you want to do is a little rough work for investigating your genetic model. A genetic model is a diagram of the logic that you propose for inheritance. For instance, if you cross a true-breeding purple plant with a true-breeding white plant (e.g. see Figure 1-2 on page 4; cross the outer two plants) you will get a heterozygote (the middle plant in Figure 1-2 on page 4, also shown at left). If we name the gene after the recessive trait (*a* is the first letter in *albino*), we know

that the heterozygote will have one capital letter "A" and a lower case "a". The heterozygote is the F<sub>1</sub> generation ("first filial", which means it's the first child from parents that are crossed). The F<sub>1</sub> is purple, which means the "a" allele is recessive; only one copy of the "A" allele is needed for enough purple pigment to make it identical to one true-breeding parent. This is complete dominance.

We can't know from the information given which allele is wild type or mutant. One hypothesis, though, is that purple pigments are required to attract pollinators and therefore would help the plant in the wild. Albino plants could be a mutant and might not generate as many seeds for lack of pollinators. If the context of your genetics problem doesn't indicate which allele is wild-type, it's good practice to name your allele based on the recessive trait. Often the recessive allele is the mutation. We might consider that the "a" allele is null and makes no pigment at all. One or two "A" alleles make enough protein to cause the plant to be purple.

Figure 1-2 on page 4 already assumes that the capital letter (*A*) stands for an allele that encodes a protein for purple pigment and the recessive allele (*a*) doesn't make pigment. Thus the *Aa* heterozygote is sufficient evidence to adopt upper- and lower-case letter "A"s to communicate the characteristics of purple and white alleles.

**A note of caution:** When you're writing down gene symbols for homework or on an exam, be sure to make the characters distinct. A typewritten "y" is easy to distinguish from the upper case "Y" but not as easy when writing it down. Instructors who ask you to show your work need to be able to follow your logic train. More important than that is YOU have to be able to follow your own reasoning. Students often switch symbols and come up with an answer that is inconsistent with the data given because of this. Consider underlining your capitals or putting a line through one of them to make it distinct (e.g.  $\text{\underline{Y}}$  for the dominant allele; *y* for the recessive).

### C.2 NAME THE GENE AFTER THE MUTANT PHENOTYPE

Some instructors would accept "P" for "purple" for the previous cross. However, the better answer is to follow an established system. During "exam fog", it's easy to get lost if you are inconsistent with how you develop your symbols. During your study period and when you're practicing genetics problems, be thoughtful

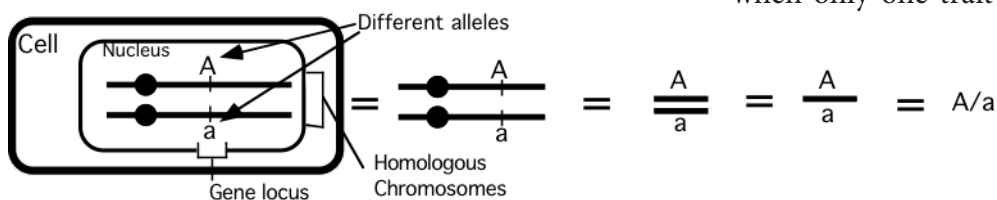
about the gene names you choose.

Let's always choose a letter based on the mutant phenotype for our gene symbol. If we are presented with a ladybug mutant that is small, we might choose "d" for "dwarf". Geneticists sometimes set up a research program based on unusual phenotypes of the organism they are studying. The fact that a mutant phenotype that is heritable exists tells us that there is a genetic control for the trait and that it might be isolated in the lab. When you look at your classmates, you don't necessarily note that none of them has an arm growing from the tops of their heads. If one student had this trait, however, you couldn't help but notice it. If you found out it showed up in that student's ancestry in a predictable fashion, you might reasonably suggest that there is a genetic basis for that. If it happened to be controlled by a single gene, you might call the gene "extra arm" or "arm head". If it happened to be a dominant trait, you might use the letter "X" (for "Xtra") or "A" (for "Arm") for the mutant allele. The wild type allele would be "x" or "a", respectively.

### C.3 IF YOU DON'T KNOW WHICH IS MUTANT, USE THE RECESSIVE TRAIT FOR THE GENE NAME

What if you don't know which allele is mutant? What if you're presented with two true-breeding frogs: one that is gold and one that is yellow. If you don't know what the predominant colour in nature is you can't know which one is mutant. If you crossed them and all the progeny are gold, then you know the dominant allele encodes a protein to make it gold. The recessive, therefore, is "yellow" and you should name the gene "y" after the recessive phenotype. This means the dominant allele would be "Y". Your offspring would therefore be Yy and the gold parent would be YY. The yellow parent would be yy.

Apply these ideas at the Online Open Genetics exercises.



**Figure 1-3**

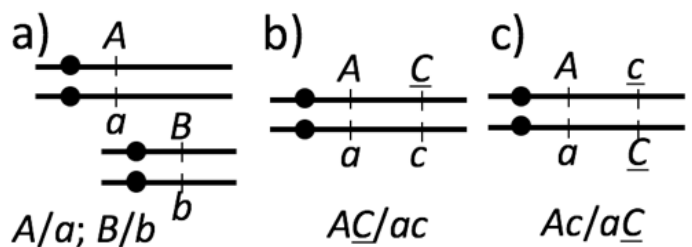
A diagram of how chromosomes, loci and alleles look in the cell, and how we depict them in written form. Note that only one gene is represented in this figure and chromosomes have not replicated. (Original-J.Locke- CC BY-NC 3.0)

## D LINKED GENES

Mendel was lucky. He studied a variety of traits in pea plants and his data were consistent with his idea of traits being encoded by pairs of discrete heritable units. He didn't call these "genes" and had no idea about their chromosomal origins or chemical makeup. It turns out that the genes he studied were either on different chromosomes and so assorted independently (See Chapter 9), or so far apart on the same chromosome that linkage could not be detected (See Chapter 10).



<http://tinyurl.com/oog-basic>



**Figure 1-4**

Three gene arrangements for cells of genotype  $AaBb$ . Chromosomes are replicated (shown with sister chromatids). a) demonstrates unlinked genes:  $A/a; B/b$ . Linked genes are shown in b) coupling  $AC/ac$  and c) repulsion  $Ac/aC$ . (Modified by T. Nickle from an original by J.Locke-CC BY-NC 3.0)

Symbols for a gene can be drawn on a page to communicate their position on a chromosome (Figure 1-3). To do this, we use a forward slash (/) to demonstrate what is on each chromosome. Figure 1-4 shows how we might conceptualize the position of a gene on two chromosomes by collapsing the chromosomes into a single line.

There's no question about where the gene is located when only one trait is under investigation: it will be at the same position on each homolog. Two genes, however, can be one of three possibilities (Figure 1-4). Each possibility has implications for gene mapping and predicting ratios from a dihybrid cross. Figure 1-4 shows the positions of genes for an unlinked situation as well as linked genes in coupling and

repulsion configurations. If genes are unlinked, put the allele symbols for one gene on either side of one slash followed by a semicolon (indicating that it's unlinked) and the other gene with the alleles separated by a second slash ( $A/a; B/b$ ). When genes are linked, only one slash is used: remember, the slash stands for a pair of homologous chromosomes. Genes in coupling would have the dominant genes together on one side of the slash and recessives on the other side ( $AB/ab$ ). Repulsion would represent the other arrangement ( $Ab/aB$ ).

Practice your skills with identifying linked and unlinked genes online. Some examples of different

forms of gene symbols are shown in Table 1-2 on page 10. Keep in mind that sometimes you have flexibility in which system of nomenclature you use, but sometimes it is dictated to you, for example in publications or other formal submissions. You are discouraged from inventing your own system or mixing up different systems because it will confuse your readers (or graders!).

A more advanced system of nomenclature is outlined in the next section. New rules are introduced to help you identify sex linked genes and predict phenotypes from gene symbols alone.



<http://tinyurl.com/oog-linked>

## Part 2: Advanced Nomenclature



**Figure 1-5**

Wildtype and mutant “flies” of *Suminospauci combibo*, an imaginary model system. These will be used in this chapter to show how to apply advanced nomenclature.

(Image created by D. Bird based on templates generously provided by The Imaginarium. Used with permission)

In the first part of this chapter, you saw how several nomenclature systems can be used to communicate the nature of the mutant allele such as the mutant phenotype and whether the mutant allele is dominant or recessive to the wild-type. This appendix extends the toolbox of symbols by introducing a powerful system that will help you practice thinking about genetics like a professional. It's a strictly-defined method of naming alleles and is designed for you to be able to look at a single gene symbol and determine its inheritance automatically. We will call it the Sumnospauci combibo convention (*Sóo-me-no spów-see*, a hypothetical organism used for genetics practice questions). We'll build up to this by working through extensions to the rules.

Many students find this formalized nomenclature scheme cumbersome at first. The rules are straightforward, but practice makes this system simple. I've had many students report that they hated it at first, but when using it they found it easier to track the alleles during complex problem solving.

## A SUPERSCRIPTS

Sometimes a letter is used as the name of a gene, and superscripts can modify it to indicate the different alleles (Figure 1-6). One common single letter code for an allelic series is “*I*”. Red blood cells can have their cell membranes modified by sugar tags that give rise to our blood type. One allele of *I* gives rise to blood type A and is therefore called *I<sup>A</sup>*. An enzyme encoded by *I<sup>B</sup>* modifies sugars to create blood type B. A heterozygote *I<sup>A</sup>I<sup>B</sup>* demonstrates both sugar tags because those alleles are expressed – they are codominant. People with blood type O only possess alleles for the *I* gene that don’t work and are therefore recessive – they don’t modify the extracellular sugar tags. Because it is recessive, individuals are homozygous for *i*: they are *ii*.

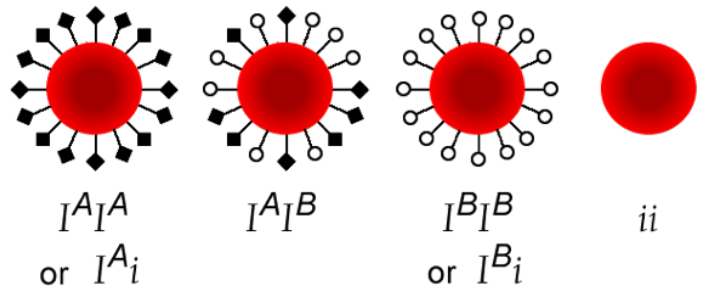
Sometimes a superscript “plus sign” is used to denote the wild type allele. One might use the symbol *W<sup>+</sup>* to indicate a wild-type allele that promotes wing growth. Note that the generic “wing” gene name isn’t a best practice – name the gene after the mutant phenotype! A wingless mutant would be *W<sup>-</sup>*. You should never use a “+” and shift the case of the letter unless you are dealing with a special case such as the codominance in the blood type example above. The capital “*I*” letter indicates it is dominant to “*i*”. The superscript A and B for the codominant alleles indicate the dominant alleles are different from each other.

Superscripts can be symbols, a single letter, or many letters (a few examples are in Table 1-1). They modify the gene name only in the superscripted symbols: the regular-sized letters are identical between them (see Table 1-1). This means that *Abc<sup>+</sup>* and *abc* would be different genes (*i.e.* not allelic); *Abc<sup>+</sup>* and *Abc* are alleles, as are *abc<sup>+</sup>* and *abc*. Note that a superscript is not mandatory for all alleles of that gene. It’s use depends on the convention being used.

Alleles of bacterial genes are typically indicated with a superscript + or -. For example, a bacterial allele that creates an enzyme that makes methionine would be *met<sup>+</sup>*, and a defective allele of that gene is *met<sup>-</sup>*.

**Table 1-1**  
Examples of genes using a superscript modifier.

<i>white<sup>apr</sup></i> or <i>white<sup>apricot</sup></i>	An allele of the <i>white</i> gene which has an “apricot” phenotype
<i>Abc<sup>+</sup></i> <i>Abc</i>	Two alleles for the <i>Abc</i> gene (wild-type and mutant, respectively). Note the mutant allele is dominant.
<i>w<sup>+</sup></i> ; <i>w<sup>a</sup></i> ; <i>w</i>	Three alleles in a series for the <i>w</i> gene. The first is wild type; the second two are different mutant alleles.
<i>bio<sup>+</sup></i> ; <i>bio<sup>-</sup></i>	A wild-type allele of a <i>biotin</i> gene and its recessive counterpart. This is likely a bacterial gene due to the convention.



**Figure 1-6**  
Relationship between genotype and phenotype for three alleles of the human ABO gene. The *I<sup>A</sup>* and *I<sup>B</sup>* alleles show co-dominance. The *I<sup>A</sup>* allele is completely dominant to the *i* allele. The *I<sup>B</sup>* allele is completely dominant to the *i* allele. (Original-Deholos -CC:AN)

## B THREE-LETTER SYMBOLS

It’s perfectly acceptable to use a single letter or even two letters. Sometimes, though, multiple traits spelled with the same first letter can get confusing. Using three letters for a gene symbol can make it easier to remember what the letters stand for. In fact, for some model systems those who study them adopt a defined nomenclature system. A plant often used for genetic studies, called *Arabidopsis thaliana*, has a three-letter code (<https://www.arabidopsis.org/portals/nomenclature/namerule.jsp>).

Just as we saw for the one-letter symbols, the dominant allele has the first letter capitalized and the last two letters are lower-case. Recessive alleles are all lower-case. With three letters, you can make gene names that are easier to keep track of. For example, you might see a fly with an extra set of wings. Instead of calling it “*w*” for “wings” (which is a poor choice because it doesn’t represent the mutant phenotype), you can instead call it “*exw*” for “extra wings”. Then, when you see it, you can sound out the abbreviation and remember that it stands for the mutation. If the wild-type allele for this is dominant, then you would write that one “*Exw*”. The first letter indicates that it is the dominant allele. What you can’t tell just from these examples is

that wild-type allele is dominant! *Hint:* remember in the previous section that the “+” superscript indicates the wild-type allele.



<http://tinyurl.com/oog-advanced>

## WHY USE THE *SUMINOSPAUCI COMBIBO* SYSTEM?

In the first part of the chapter you saw different systems of expressing gene inheritance. Some rules we established were:

- ◆ Alleles are instructions for a protein. It is the protein that determines the phenotype of a trait.
- ◆ Traditionally, we call an allele “dominant” or “recessive”, but these terms actually refer to the action of the protein the allele encodes. The alleles are merely “instructions”.
- ◆ Symbols should be chosen based on the phenotype of the mutant. If the mutant allele cannot be identified, the symbol should reflect the recessive phenotype.
- ◆ Symbols for “recessive” alleles are in lower case.
- ◆ Symbols for “dominant” alleles have a capitalized first letter (if the symbol has more than one letter, all remaining letters are lower case).
- ◆ Some mutant alleles act as recessive to wild-type, and some mutant alleles act as dominant to their wild-type counterpart.
- ◆ A “+” superscript indicates that an allele is wild-type. If this style of nomenclature is chosen, then all alleles are identical (including the type of letter: e.g.  $a/a^+$  reflect a recessive mutation and the corresponding wild type in a heterozygous individual;  $a/A^+$  is incorrect).

We noted that if a mutant allele is recessive to the wild-type, and the wild-type allele is dominant to the mutant allele. Conversely, if the mutant allele is “dominant”, the wild-type allele is “recessive”. When writing these things out fully, the descriptions get complex; and the more complex, the more likely you are to make a mistake. Likewise, if you’re writing out a description and start talking about the mutant alleles and switch to the wild-type allele later on, chances are good you’ll confuse the reader – and likely yourself!

There is a simple solution: always name gene after the mutant phenotype. If you can’t tell which phenotype is mutant, name the gene after the recessive. The mutant allele will have a capitalized first letter if it’s dominant. If it is recessive, it will be all in lower case. Be sure to italicize or underline the gene symbol. Once the mutant allele is set, use exactly the same letters (including the upper- or lower-case first letter) and put a super-

script “+” behind it.

The “+” means wild type. Always. But it also means “not”. Bear with me.

If you see a gene symbol and you know it follows the *S. combibo* convention, you can immediately know if it is dominant or recessive. If you see *wht* as a gene symbol, you know, without any extra information, that:

1. The mutant allele:
  - a) Is recessive (because the symbol is in all lower case)
  - b) Probably looks like something that *wht* would describe (perhaps “white”?)
2. the wild type allele:
  - a) Has the symbol  $wht^+$
  - b) Is “not” mutant (because of the “+”, but it should be obvious because we name the allele after the mutation)
  - c) Is “not” recessive (therefore it is dominant to *wht*)
  - d) Is “not” white (or whatever *wht* stands for)

That’s a lot of information in just three letters! Note that we can get the same information from the wild type symbol alone. Consider  $Pnk^+$ . It seems like it refers to something that’s pink when it displays the mutant phenotype (but this allele is NOT the mutant; it has the “+” so it is NOT pink). The capital “P” tells us the mutant allele is dominant, and so the wild-type allele is NOT dominant (it is recessive).

Finally, note that we’re using the term “gene name”. The gene is named after the mutation. Whether it is *wht* or  $wht^+$ , the “gene name” is *wht*. For *Pnk* or  $Pnk^+$ , the gene name is *Pnk*. We use the superscript when it is required. Try to get into the habit of asking exactly what the mutant symbol is trying to communicate.

## C MORE POWER: X-LINKAGE CHANGES THE GENE SYMBOL

So far we’ve dealt with symbols that involve autosomal genes. In earlier courses, you might have encountered sex-linkage (also known as X-linkage, because most of these genes are on the X chromosome). More information about sex-linked genes is in the next chapter (Chapter 4).

Sex chromosomes, by definition, are represented differently between males and females. In mammalian systems - the human system being most familiar to you - females have two X-chromosomes and males have one X- and one Y-chromosome. Be aware that this isn’t universal for all organisms!

Because males in the X/Y system do not have homologous chromosomes, it's obvious when a gene is X-linked: the male has only one allele compared to having two alleles for his autosomes. But how would you indicate that a female's genotype contains X-linked genes? The system so far doesn't contain enough information to illustrate this.

We'll conform to the three letter system but put these letters as a superscript after a large "X" for X-linked genes. Since there are very few Y-linked genes, the Y

chromosome is represented as just a "Y" with no superscript. Wild type alleles will still have a "+" after the three letters. For example, a male with an X-linked gene for a wild-type gene encoding black eyes would be represented as  $X^{bey+}/Y$ . A female heterozygous for this gene would be  $X^{bey+}/X^{bey}$ .

More examples are in **Table 1-2**.

Practice writing out genotypes and making gene symbols at

<http://opengenetics.net>.



<http://tinyurl.com/oog-sex>

**Table 1-2**

Examples of symbols used to represent genes and alleles.

Examples	Interpretation
<i>A</i> and <i>a</i>	Uppercase letters represent dominant alleles and lowercase letters indicate recessive alleles. Mendel invented this system but it is not commonly used in publications because not all alleles show complete dominance and many genes have more than two alleles. It's quick and easy for you to use when working out genetics problems when you are sure each gene involves only two alleles.
<i>a</i> <sup>+</sup> and <i>a</i>	Superscripts are used to indicate alleles. For wild type alleles the symbol is a superscript +. The mutant allele of gene <i>a</i> would be recessive.
<i>met</i> <sup>+</sup> and <i>met</i>	This is typical of a prokaryote gene symbol. It could be referring to wild-type (functional) and mutant (nonfunctional) alleles of a gene that makes a protein in the methionine synthesis pathway.
<i>AA</i> or <i>A/A</i>	Sometimes a forward slash is used to indicate that the two symbols are alleles of the same gene, but on homologous chromosomes. Both representations in this row are identical: it represents a homozygous dominant.
<i>Aa/Aa</i> or <i>Aa/aa</i>	Note that this example shows two alleles of the gene <i>Aa</i> . We know that the gene symbol is two letters because the slash separates the allele found on each of the homologous chromosomes. We cannot tell if the mutant phenotype is recessive because there's no indication which is wild type.
<i>Gm</i> <sup>+</sup> <i>shr</i> / <i>Gm</i> <i>shr</i>	The three-letter system is used here. " <i>Gm</i> " might mean that the phenotype is "green", but we can't be sure. What we do know is that the mutant allele codes for a protein leading to a dominant phenotype. The wild-type allele must be recessive to the mutant allele. Maybe " <i>shr</i> " means "shrunken" or "short", but we know that the mutant phenotype can only be seen in the homozygous recessive configuration. The phenotype for this organism is mutant for both <i>Gm</i> and <i>shr</i> traits. Final note: the genes are on the same chromosome based on the position of the slash.
<i>bob</i> <sup>+</sup> / <i>bob</i> ; <i>mia</i> / <i>mia</i>	This also uses the three-letter system. The organism is heterozygous for <i>bob</i> but shows the wild-type trait in its phenotype. It is homozygous recessive for <i>mia</i> and therefore shows that mutant phenotype. The genes are unlinked.

**SUMMARY:**

- ◆ Symbols are used to denote the alleles, or genotype, of a locus.
- ◆ Phenotype depends on the alleles that are present, their dominance relationships, and sometimes also interactions with the environment and other factors.
- ◆ In a diploid organism, the alleles can be homozygous, heterozygous or hemizygous.
- ◆ Allelic interactions at a locus can be described as dominant vs. recessive, incompletely dominant, or co-dominant.
- ◆ Two different genes can be on the same chromosome (linked) or on different ones (unlinked)
- ◆ Steps:
  - Can you identify the mutant? Name the gene after its phenotype
  - If you cannot tell which allele is mutant, name the gene after the recessive allele
  - Capitalize the first letter for the dominant allele; use lower-case for the recessive allele
  - If genes are linked, write the gene symbols together on each side of the slash. If genes are unlinked, they appear on different sides of a semicolon