

The Genetics Journal – Part Three

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In our last entry in the Genetics Journal we discussed Gregor Mendel's experiments with pea plants looking at the effects of a simple dominance effect on traits determined by one gene with two possible alleles. We also introduced the concept of a useful tool called a Punnett square. In this entry we look at four different effects on simply inherited traits: co-dominance, partial dominance, epistasis and sex-linked traits. But first we will expand on the concept of the Punnett square to show how two simply inherited traits exhibiting simple dominance can be evaluated at the same time to look at all of the possible genotypic and phenotypic combinations for both traits together.

Our example will use Angus and Hereford cattle and will look at two traits. One trait is coat color, which can be either Black or Red. Black exerts complete dominance so we will use the letters B and b for Black and Red respectively. The other trait is polled (no horns) or horned with polled being dominant so we will use P and p.

Purebred Angus cattle are primarily homozygous polled and black, BBPP, whereas purebred Herefords are red and horned, bbpp. Let's look at a cross between these two. The offspring of course have only one possible genotype, BbPp, since both parents are homozygous. These "Black Baldies", black with no horns, are of course all heterozygous for both traits. What happens then when we cross two Black Baldies?

Black Baldie Dam
BbPb

	BP	Bp	bP	bp	
Black Baldie Sire BbPb	BP	BBPP	BBPp	BbPP	BbPp
	Bp	BBPb	BBpp	BbPb	Bbpp
	bP	BbPP	BbPp	bbPP	bbPp
	bp	BbPp	Bbpp	bbPp	bbpp

If we analyze these results we find that there are nine different possible genotypes, and yet due to the complete dominance there are only four possible phenotypes. We can have BBPP, BBPp, BbPP and BbPp, all of which will be black and polled; BBpp, Bbpp which will all be black with horns; bbPP and bbPp which will be red and polled; and bbpp which will be red and horned.

Any number of simply inherited traits can be looked at in this way with a Punnett square. The more genes we study at once, the more complicated the diagram, but the technique is the same in any case. If we were to look at three genes, we would have eight columns and eight rows, for a total of 64 cells. The author and his wife breed French Angora rabbits for their fine fleece. The color genetics of rabbits is very well known and documented and is controlled by a pattern of simple inheritance. There are five gene locations involved however, which means that if you had two parent rabbits, both heterozygous for each gene, you could make a Punnett square that would accurately predict

the possible color outcomes of the offspring, as well as the percentage probability of each color, but the Punnett square would have 32 rows and 32 columns with a total of 1024 squares within the diagram to tabulate. We do this, but it is very time consuming and complex. Never-the-less it does tell us exactly what possible colors and what the probability of each color offspring is. We look forward to the day when we can be that accurate in predicting color possibilities in alpacas.

Another relationship that can exist between two genes at a single locus is simple inheritance without dominance. **Co-dominance** allows heterozygous offspring to have a resultant phenotype that is intermediate between the two homozygous possibilities. A good example is Shorthorn cattle coat colors. Shorthorns can have coats that are red, white or roan. The roan is a distinct appearance that comes from having both red and white hairs. It is called Co-dominance because each hair is either all red or all white. We will call red R and white r. RR cattle are red, rr cattle are white and Rr cattle are Roan. Let's use Punnett squares again to visualize what is happening. If we breed red to white it looks like this:

	Red Dam RR		
	R	R	
White Sire rr	r	Rr	Rr
	r	Rr	Rr

Again, since both parents are homozygous, the offspring are all heterozygous, in this case Roan (Rr). If we breed two of the Roan Shorthorns we can see the result:

	Roan Dam Rr		
	R	r	
Roan Sire Rr	R	RR	Rr
	r	Rr	rr

Again we have three different genotypes, just as in our examples with tall and short peas and Suri and Huacaya alpacas, but this time there are also three distinct phenotypes: Red, White and Roan. As you can see, this situation makes it much easier for the breeder to select animals to breed because each genotype can be determined just by evaluating the phenotype. The recessive genes are not hidden as they are in cases of complete dominance.

A similar but slightly different situation called **partial dominance** can also occur. A good example is red and white carnations. A heterozygous cross between the homozygous red and homozygous white produces a pink carnation. The single red gene cannot exert as much influence on the pigment production as two red genes can, hence a pink color. The Punnett squares would look exactly like those for Shorthorn cattle above, with the Rr heterozygote having the pink phenotype. A cross between two Rr pink carnations would yield 25% red, 50% pink and 25% white flowers.

There is another situation for us to consider in the case of simply inherited traits. When a gene at a second locus can effect the expression of a gene at the first locus we have an effect known as **epistasis**.

Epistasis means that the genes at one locus can have an effect on genes at other loci.

“A simply-inherited example of epistasis that is relatively easy to understand is coat color in Labrador retrievers. Labs come in three basic colors: black, chocolate and yellow. Genes at two loci determine these colors: the B (black) locus and E (extension of pigmentation) locus, as follows:

B_E_ => black
bbE_ => chocolate
__ee => yellow

The dashes in these genotypes indicate that either allele could be substituted without changing the phenotype. Black Labradors, for example, can be BBEE, BBEe, BbEE, or BbEe. Yellow labs can be BBee, Bbee, or bbee. Note that the expression of genes at the black locus depends on the alleles present at the extension locus. So long as there is at least one E allele at the extension locus, there appears to be complete dominance at the black locus, with black being dominant to chocolate. However, if the genotype at the extension locus is ee, then genes at the black locus become irrelevant—all animals will be yellow”¹

In this example then, genes at the E locus are epistatic over genes at the B locus. Epistasis is just one type of **gene combination effect**. Gene combination effects can be very tricky when it comes to making genetic predictions about animal breeding and we will refer to them again in the future when we begin to look at determining breeding values, which are a way of describing an animal’s genetic value as a producer of characteristics in an offspring.

What would Labradors that are heterozygous at both locations look like? They would be BbEe and would have the black phenotype, just like two other possible genotypes, BBEe and BBEE. Once more then we have the effect of complete dominance hiding recessive genes and phenotypic selection is dangerous.

Another topic to consider in modifications to Mendelian inheritance is **Sex-Linked inheritance**. Mammals have one pair of chromosomes that are called the sex chromosomes. They are usually referred to as X and Y. If an animal has two X chromosomes it is female. If it has one X and one Y chromosome then it is a male. Even though these chromosomes do form a pair, the X and Y are totally different from each other. A female, with two X chromosomes, can have two copies of an allele at each locus, and therefore can be homozygous or heterozygous at that locus. If the trait is one that exhibits no dominance, such as tortoiseshell color in cats, then the female could have three possible phenotypic expressions of this gene. The gene in question here is the gene for orange color,

which is dominant, O. The recessive gene, o, represents “non-orange”. A heterozygote, with a copy of each, Oo, will have the tortoise shell color. The male on the other hand, has only one X chromosome, and so he can only have an O or an o. Males then will be either orange or non-orange. Only females can exhibit the tortoiseshell color which is therefore said to be a sex-linked characteristic. Numerous other characteristics are known to be sex-linked in mammals, and behave according to this same type of pattern, where the female can be heterozygous or homozygous for any of the alleles and the male can only express the trait caused by a single copy of any of the alleles.

Another type of situation can arise in certain characteristics that are governed by a simple dominance relationship of expression of the alleles. For some traits there can be more than two alleles at a single locus. One example is the presence or absence of horns in Merino sheep. The characteristic of having horns, or being polled (without horns), is controlled by three different alleles at one locus. The most dominant allele causes the animal to be polled. The intermediate gene causes males to have horns, but females to be polled. The most recessive gene causes horns in both sexes. Thus we have a hierarchy of genes that range across a scale from most dominant to most recessive. We could call them P for polled, P’ for horned males/polled females, and p for horns in both sexes. The typical genetic notation used to show this range of dominance would be to write $P > P' > p$. Of course any one sheep can only have at most two of these three alleles, one on each chromosome. Although there are only two possible phenotypes, horned or polled, there are six possible genotypes: PP, PP’, Pp, P’P’, P’p, pp. Only one of these six will result in a female with horns, which is why most flocks of merino sheep do not have horned females. Yet many of the females may carry one p gene which is hidden by P or P’ at that locus on the other chromosomes. Therefore even in a flock of Merinos that has all polled sheep, an occasional horned female will “pop up” when she inherits two of these most recessive p alleles. Multiple alleles at one locus can make it very difficult to do genetic predictions of matings in these so called “simple” inheritance situations. To go back to my example of our French Angora rabbits, I mentioned that there are five loci that affect coat color. If that is not complex enough, the C locus has five different possible alleles that a given rabbit can have any two of. This means that at just that one locus there are 15 possible genotypes.

As alpaca breeders we cannot just gloss over this concept, because it appears that, at least in the case of color genetics, this same situation may exist in alpacas. Dr. D. Phillip Sponenberg, a leading authority on color genetics, thinks that there may be six or more different alleles at the Agouti locus in a hierarchy of dominance that affect color in alpacas at this one of several color loci.

So far in the first three entries to the Genetics Journal we have reviewed the Basic Mendelian Principles for the inheritance of simply inherited traits including the concepts of genes, alleles, dominant and recessive traits, the Mendelian laws of Gene Segregation and Independent Assortment, genotypes, phenotypes, Punnett squares and

simple dominance. We also discussed modifications of Mendelian ratios such as partial dominance, co-dominance, epistasis and sex linked characteristics. These are the basic building blocks of genetics, but the genetics of animal breeding are much more complex in most cases. In the future we will build on these foundational principles to begin to understand advanced principles such as polygenic traits, quantitative genetics including predicted breeding values and estimated progeny differences, population genetics, and other advanced principles.