The Genetics of Horse Coat Color

From top left: Chincoteague Pony, Friesian, Quarter Horse, Palomino
Photos by Jim Shambhu. Come see these breeds and more at the Kentucky Horse Park!

Coat color has long been an identifying feature for horses. Throughout time, some breeds have even been bred for specific colors. Breeders and owners alike have been curious about what coat color a foal will inherit. Using basic genetic principles, breeders can predict the coat colors of offspring, and then will mate horses to produce foals with desired coat colors and viability, such as in the case of Dominant White and Frame Overo horses. This activity will demonstrate some of these genetic principles which can then be applied to other characteristics and species.

*This educational packet is intended for high school biology students.

Name:_____Date: _____

Class: _____

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Why does coat color matter?

Some horse breeders have selectively bred for certain coat colors for centuries. Though coat color has shown no correlation to improved work or performance by a horse, breeders still have a preference for certain coat colors. These preferences exist for many reasons. Some breeds, their registries, and their traditions value some colors over others. For example, breeders of the Marwari from India tend to favor and breed for grey, piebald, or skewbald, and try to avoid all black as that is thought to be unlucky.

Some pairs of horses pull wagons and carriages in parades and shows throughout the country. Often, these pairs (or more) have matching coats to accentuate their role as a team. The trolley here at the Kentucky Horse Park is a daily example of this.

There are also many people that simply have a favorite color of horse that is aesthetically pleasing to them and want to own horses of that color.

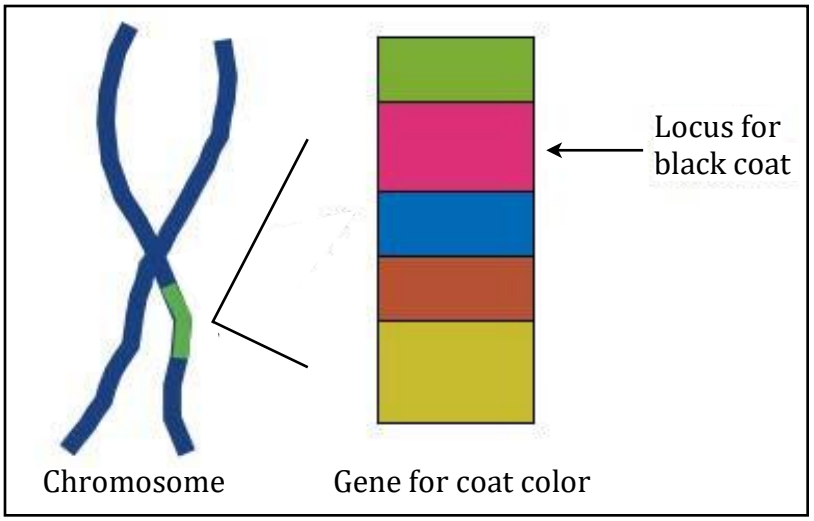
An Arabian horse at a show. Photo by Jim Shambhu.

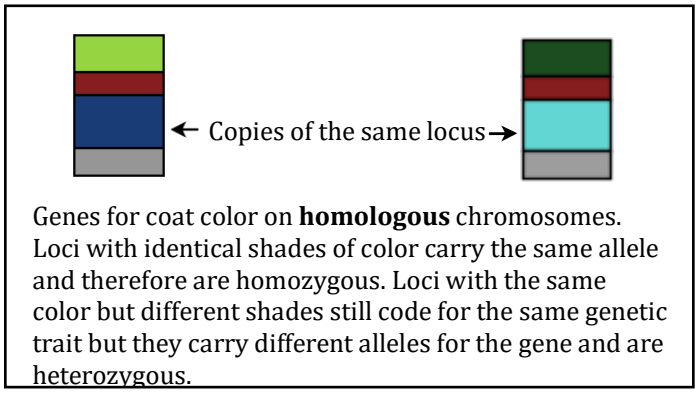
Clydesdales Colin and TJ pulling a trolley. Photo by pixbysteve.com. Come see Colin and TJ at the Kentucky Horse Park!

The Genetic Blueprint: Genes

Coat color is **inherited**, meaning that the condition of the trait is passed directly from parent to offspring. An understanding of this mechanism allows breeders to predict the coat colors of foals based upon their parents.

The genetic "information" about all of an organism's traits, such as a horse's coat color, is contained on **chromosomes** inside their cells. A **gene**, a segment of the chromosome, contains the information for a specific trait or suite of traits. Genes are further divided into individual units called **loci**. Each locus, either alone or in conjunction with other loci, influences part of the trait controlled by its corresponding gene. For example, coat color is determined by a single gene, and several loci within that gene determine color.



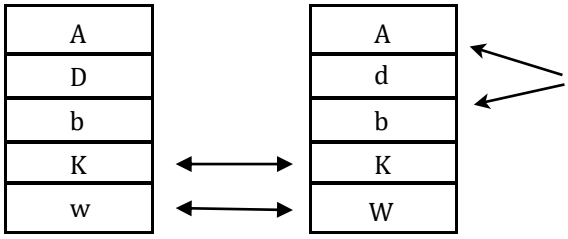


Genes for coat color on **homologous** chromosomes. Loci with identical shades of color carry the same allele and therefore are homozygous. Loci with the same color but different shades still code for the same genetic trait but they carry different alleles for the gene and are heterozygous.

Because each chromosome is paired with its **homologous** chromosome, another chromosome that contains genes for the same traits, there are two copies of each gene and locus in the genome that are not necessarily identical due to the variation in **alleles** (their similarity is represented here by color). The offspring will receive a combination of these inherited alleles from their parents. We will later see how geneticists analyze and predict the genetic outcome for an offspring using the parents' already known genotypes.

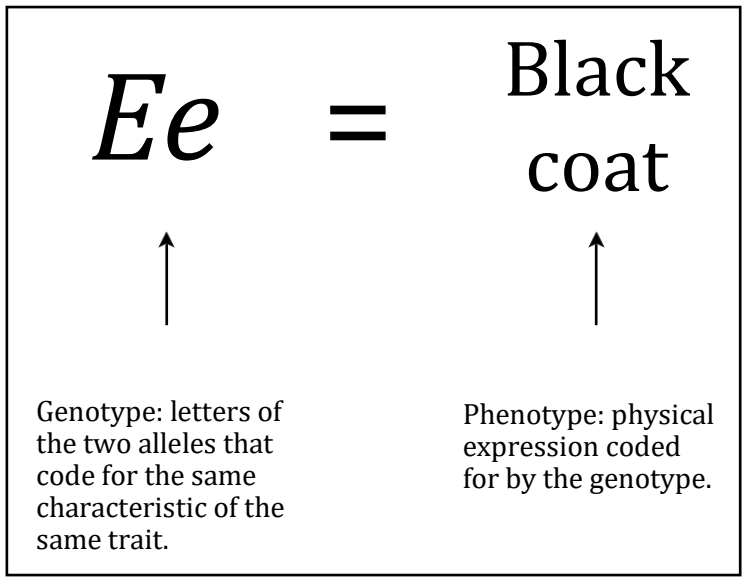
How do we study genes?

The molecular structure of a locus determines the genotype of the gene at that locus for the organism which will then refer into its phenotype. The two copies of a locus may or may not be identical due to the variation in alleles inherited from the parents, otherwise the loci are stable in that they are fixed positions on the chromosomes, essentially standing as genetic markers. Two single letters, either capital or lowercase, commonly represent the genetic alleles from the parents on the locus. These letters are assigned by the scientist studying the locus and denotes a specific variation of a genetic trait. Two different loci will have different letters (A and B, for example), but the two copies of the alleles at a single locus will have the same letter and be written together (AA, aa, or Aa) to create a specific genotype.



Loci on homologous chromosomes that code for the same genetic trait have the same letter, but sometimes have different cases denoting the variation in alleles. This concept was illustrated by the different shades of the same color in the diagram on the previous page.

Genes for coat color on homologous chromosomes.



The letters that represent alleles are known together as a **genotype**. Every genotype has a corresponding **phenotype**, which is the physical expression of the trait that results from the genotype's molecular configuration. Next, let's use horse coat color as an example of genotype and phenotype.

The Genetics of Horse Coat Color

One locus determines black and chestnut coat color. At the black and chestnut color locus, an *E* allele denotes black and an *e* allele denotes chestnut. The *E* allele is **dominant**, meaning that any genotype with this allele will produce the *E* phenotype. Both the **homozygous** *EE* and **heterozygous** *Ee* genotypes produce a black coat. The e allele is **recessive**, meaning that it is masked by a dominant allele in an *Ee* genotype but is expressed in an *ee* genotype. The *ee* genotype, then, produces a chestnut coat. If you know the genotypes of two parents, you can predict the genotype and phenotype of their offspring.

EE = Black coat

Ee = Black coat

The E allele is dominant, so its phenotype is expressed, even when the e allele (recessive) is present in the genotype.

ee = Chestnut coat

Genetic Tool: Punnett Square

A **Punnett square** is a simple genetic tool that can be used to demonstrate the possible offspring between two parents using their genotype. Let's set up a square for a chestnut horse and an *Ee* black horse. One cross is done for you as an example.

Write the first allele for the *Ee* horse here.

Write the second allele for the *Ee* horse here.

Write the first allele for the *ee* horse here.

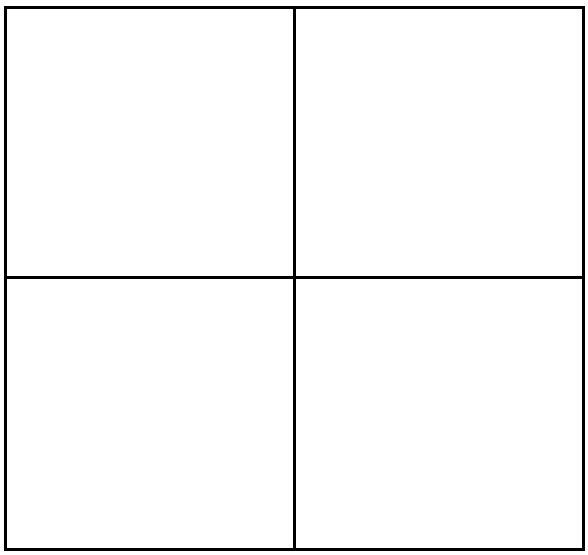
Write the second allele for the *ee* horse here.

	E	___
e	*Ee*	
___		

The crosses in the boxes represent the possible genotypes of the offspring from this mating. In this case, we see that statistically, half of the offspring of these parents over their breeding time will be black and half will be chestnut.

Genetic Tool: Punnett Square (cont.)

Let's see what happens when two *Ee* black horses breed. Fill in the following Punnett square:



Fill in the blanks:

_____(fraction) of the offspring will be black, but two different genotypes are represented among the black offspring. Of the black offspring, 1/3 will be_____(genotype) and 2/3 will be_____(genotype). The remaining_____(fraction) will be chestnut.

Whites, Greys, & Lethal Whites: What's the Genetics?

White coats have long been a desirable color for their beauty and rarity. But what is really the difference between them and their genetics? Most people may see a horse that appears to have an all-white coat, but are really seeing the common grey horse that has 'greyed out' and mistaking it for a white horse.

Neapolitano IV La Sada (Beamer), the Lipizzan representative from our Breeds Barn shows. He is a perfect example of a horse that has 'greyed out.' Note the darker muzzle and eyes.

Grey horses have a dominant ***G* gene**, which is responsible for the exclusion of pigment from hair. This means that they were born a solid color, such as black or chestnut, and will maintain that skin color while the pigment in their hair fades out over their lifetime. Eventually, they reach a full 'greyed out' or white appearance similar to humans. One way to tell they are grey, besides pushing around the hair and getting a glimpse of their skin, is that their muzzles and the skin around their eyes will be darker and not pink.

A grey horse will have a genotype of either *GG* or *Gg*, while all non-grey horses will be recessive (*gg*) at the grey locus.

White Prince, a rare white Thoroughbred . Photo by Jim Shambhu.

In the case of a true white coat, which is much rarer and a result of an autosomal dominant genetic mutation of the **KIT gene.** This is responsible for the white spotting of horses, dogs, cats, swine, humans, cattle, and rats or mice. The resulting white gene is represented by the allele *W.* There are multiple mutations that have the possibility of occurring on the KIT gene for any white with only one actually occurring per offspring. These mutations are numbered as such: W^5, W^{10}, and W^{20} being the three most common. There may also be an *N* present, which means that no mutation gene was passed from one of the parents. This is where **autosomal dominant** will come into play.

When an individual has inherited one copy of the mutant gene (*W*) and one normal gene (*N*), there will be at least some degree of white spotting or full white. For example if Individual A has a mutation genotype of N/W^5 then they will express some white spotting because the W^5 mutation is present, and if Individual B has a genotype of *N/N* then there are no mutations and the horse will possess no white coloring and be solid. Along the same lines, when looking past the mutation to broader color genes, in the presence of a dominant *W* gene mutation an **epistatic** effect takes hold over another color gene, such as black (*E*). This means that the dominant *W* gene will suppress the effect of the *E* gene.

Table of possible white mutations of KIT gene and their results:

N/N	No mutations detected
N/W^5	One copy of W^5 mutation; some white markings
W^5/W^{10}	One copy of W^5 and W^{10} mutations; white markings present, could be all white
W^5/W^{20}	One copy of W^5 and W^{20} mutations; white markings present, could be all white
W^5/W^5	Two copies of W^5 mutation; white markings or all white *not viable*
N/W^{10}	One copy of W^{10} mutation; some white markings
W^{10}/W^{20}	One copy of W^{10} and W^{20} mutations; white markings present, could be all white
W^{10}/W^{10}	Two copies of W^{10} mutations; white markings or all white *not viable*
N/W^{20}	One copy of W^{20} mutation; extended white markings might be present
W^{20}/W^{20}	Two copies of W^{20} mutation; more chance for extended white markings

As mentioned above, the white gene is seen through mutations of the KIT gene. A breeder can come across some major trouble if they are not careful and pay close attention to which mutation the parents will be supplying. What this means is that there are two mutation genotypes that can cause the resulting embryo to not be viable and be reabsorbed or aborted early on in the pregnancy. This occurs with the dominant homozygous pairing of the W^5 and W^{10} mutations: W^5/W^5 or W^{10}/W^{10}. The only dominant homozygous mutation pairing that won't ultimately result in a non-viable pregnancy is the W^{20} mutation: W^{20}/W^{20}. This will just produce a larger chance of extended white markings, such as stockings that go above the knee or hock, sabino coloring and more. However, the two non-viable dominant homozygous white mutation pairings should not be confused with, as they often are, **Lethal White Syndrome (LWS/LWO).** This (unfortunately always) lethal condition occurs mostly within the **overo** coat pattern of the Paint horse and some Thoroughbreds, more specifically the majority within the **frame overo** pattern. It needs to be noted that this is only the majority. There have been cases noted in the tobiano pattern and solid overo patterns, but will always have overo genes contributing as horses can carry the gene. When two horses mate that both carry the lethal *O* gene, there will be a chance of producing a lethal white foal which is only possible in the homozygous form. The subsequent effect is that the foal will born all white and will die or be humanely euthanized within hours or days after birth from intestinal tract abnormalities, ***ileocolonic aganglionosis***. Thankfully, this condition is now becoming easier to avoid by doing a genetic test on possible parents and not breeding two carriers.

An LWO foal with frame overo mother. Photo Credit: Stephanie Valberg, University of Minnesota

_____ Devoid

_____ Carrier

_____ LWO

Activity: If N denotes the sequence for non-overo and O represents the lethal white gene, show in the Punnett square to the left the outcomes of an NO X NO pairing. What are the chances for an offspring completely devoid of the lethal white gene, of producing a carrier, and of producing a lethal white foal (LWO)? Express in percentage.

A breeder has a mostly solid mare with some degree of white spotting and a full white stallion that he wishes to breed with each other. However, he does have some concern about what white mutations they could possibly pass on, so he has some genetic testing done on the mare while already knowing the stallion's mutation genotype. The stallion has a mutation genotype of W^5/W^{20} and he found that the mare has a mutation genotype of N/W^5. Knowing this information, list the possible mutations that could be passed to offspring from the sire and from the dam {sire=dad, dam=mom}

Possible genetic mutations from sire:

Possible genetic mutations from dam:

Now set up the Punnett square for the cross between these two parents:

Referencing the white mutation chart on pg. 10, what are the chances of this pair producing a foal that could be fully white, a foal that displays some markings, a foal that could show extended markings, and having a non-viable pregnancy? List the ratios of each genotype below.

Critical Thinking

Imagine the foal these two produced ended up being a filly with a true white coat. The breeder believes this to be sheer luck because of the small chance. However, he would like to have another true white foal down the line and he believes this filly would have a greater chance of producing a white foal than her mother. Years later, when this mare is ready to be bred after a successful racing career, the breeder finds the seemingly perfect true white stallion. The owner of the stallion informs the breeder that his genetic mutation is W^5/W^{10}. Would this be a good match? Explain why or why not by creating a Punnett square and using the information from the mutation chart from pg.10 and the results from your Punnett square on the previous page.

Vocabulary

Autosomal Dominant: Specifically for mutated genes; Inheritance pattern where there is one copy of the mutant gene and one normal gene present in an individual and the mutant gene trait overshadows the normal and is present

Allele: Variant form of a gene located at a specific position (locus) of a chromosome; combination of at least two of these comprise a genotype

Chromosome: Threadlike strand of DNA that determines heredity through the genetic material they hold and are located in the nucleus of eukaryotic cells

Dominant: Masks recessive alleles in a heterozygous genotype; therefore is expressed in the phenotype; represented by an uppercase letter

Epistasis (Epistatic): One gene masks the phenotypic expression of another gene when there is interaction between genes at two or more loci; epistasis is the process, epistatic describes the gene that performs this

Frame Overo: Specific overo Paint pattern that are the typical carriers of the Lethal White Syndrome gene; although they are the most common they are not the only ones and do not always carry it

Gene: Segment of a chromosome; contains information for a specific trait or suite of traits; the fundamental, physical, and functional unit of heredity

Genotype: set of genes/alleles in the DNA which is responsible for a particular trait

Heterozygous: Have one dominant and one recessive allele at the same locus

Homologous: Chromosome pairs (one from each parent) that have similar length and gene location but genes may contain different alleles

Homozygous: Have either two dominant or two recessive alleles at a single locus

***Ileocolonic aganglionosis*:** Intestinal tract abnormalities resulting in colic and death in foals affected by Lethal White Syndrome

Inheritance: Process through which a trait is passed from parents to offspring

KIT Gene: Mutations of this gene causes white spotting/markings in horses, cattle, humans, cat, dogs, rats/mice, and swine; responsible for more white patterns than any other gene in horses

Lethal White Syndrome (LWS/LWO): Autosomal genetic disorder where a foal is born almost pure white and dies due to intestinal complications within either hours or days after birth; usually humanely euthanized due to related pain

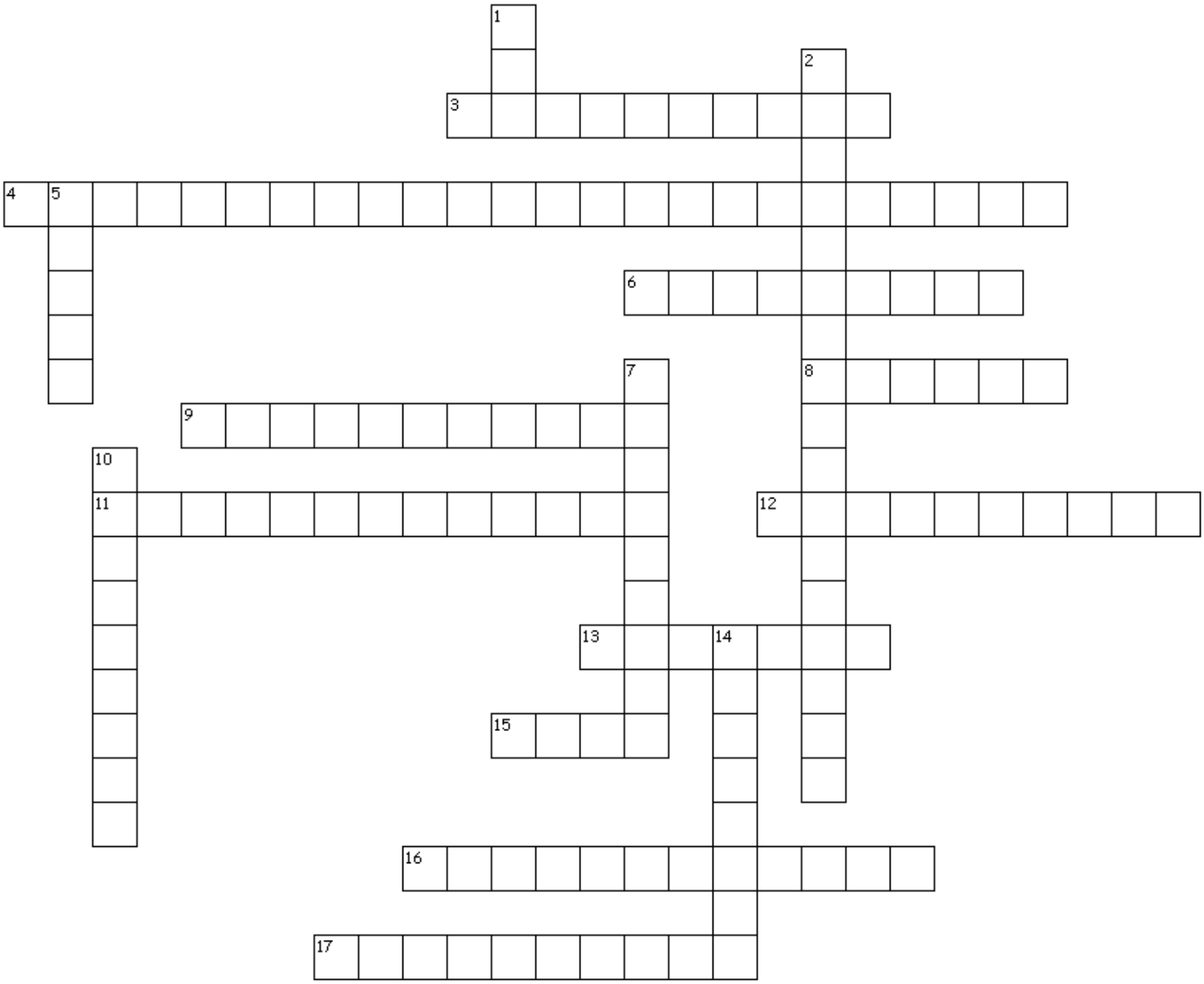
Locus (plural Loci): fixed position on a chromosome for a gene acting as a genetic marker

Phenotype: Physical expression of an organism's genotype

Punnett square: Diagram used in the study of genetics used to predict the genotype outcome of a breeding

Recessive: Masked by a dominant allele in a heterozygous genotype and only shows in the homozygous form; represented by a lowercase letter; trait that shows in the homozygous recessive form is a recessive trait

Genetics Crossword



All answers from vocab

Across:

3: Two of the same alleles, either dominant or recessive, that make up the genotype

4: Intestinal abnormality that is found in lethal white foals that causes colic and death

6: Physical expression of a genotypic trait

8: Variant form of a gene at a locus

9: Process of traits passing from parents to offspring

11: Diagram used to predict genotypes of a breeding

12: Pair of chromosomes, one from each parent, with similar gene locations, but with different genetic variations

13: Mutation of this is responsible for white spotting/markings and all white coat in horses

15: Unit of heredity holding information for a specific trait or suite of traits

16: *Ee*

17: Determines heredity through genetic material; located in nucleus

Down:

1: Abbreviation for deadly disease in affected homozygous frame overo foals

2: When a genetic mutation overshadows normal genes and becomes present

5: Specific location on a chromosome for genes; genetic marker

7: *e* in ***Ee***

10: Describes a gene that masks another gene

14: set of genes from each parent that determine a particular trait