Horse Genetics

Night Owl Education and Equestrian
About the Course

On this course you’ll learn about numerous aspects of genetics as it relates to horses.

Whether you are a recreational rider curious about genetics, a breeder wanting to make some informed choices or an equine science or genetics student there is something here for you. There are also some horse genetics research findings that have been assimilated and summarised especially for this course, providing information in a form you won’t (yet!) find elsewhere.

I remember a trick horse trainer once saying that with horses you should “make haste slowly”! Anyone who’s trained a horse to do anything will understand what he meant. The same is generally true for people learning something new, or about which they have only a patchy starting knowledge. Start off slowly, get the basics right, don’t let anyone else push you or rush you - go at your own pace. Gradually, as you become familiar with the basics, the speed and ease with which you pick up new things will increase. This course is built to be like that - one which you can do at your own pace, in your own time, and progressing only when you are ready.

The first four modules of this course introduce you to a wide range of genetic concepts and techniques. All of them are relevant to horses, and essential for a good basic understanding of horse genetics. They cover much more than just basic genetics and Mendelian inheritance, which many enthusiasts learn to some degree. There’s also plenty to help you understand more complex inheritance, such as that affecting performance, conformation and character traits.

The introduction to molecular genetics in module three covers some medical genetics, as the two heavily overlap. In fact various aspects of medical genetics are covered throughout the course, where appropriate. I have chosen to specifically reflect what’s happening now, and to discuss a few techniques most likely to be used in the future. The recent advances in molecular genetics have provided breeders with many previously unavailable tests. Such
tests will be to the eventual good health of horses, and will also aid many breeders in achieving their aims. Future advances in molecular genetics will directly lead to further improvements in veterinary medicine, to the benefit of horses and horse owners in general.

The subject of evolution is treated from the point of view that horse breeding is the application of artificially driven evolution, while equine conservationists can only make progress if they take evolutionary forces into account. Inbreeding, founder horses and selection, for example, are integral parts of horse breeding and conservation. With molecular genetic technology progressing apace I foresee that there could be some interesting and exciting developments in the worlds of horse breeding and conservation. This could well include an improved ability to select for traits controlled by several genes at once. In addition the fascinating subjects of domestication, ancient breeds and breed development are all benefiting, as advances in genetics help to unravel their stories, while preserving the odd mystery for us to contemplate.

The last module on horse colour and pattern genetics is different. There are a few new concepts introduced here and there. Generally though concepts already learned are illustrated, sometimes in depth, with reference to what is known about this ever popular aspect of horse genetics. It is this area of genetics that often triggers a person's interest in horse genetics, as it did my own. It is also this area that has benefited exceptionally well from recent research, with an explosion of information answering previous questions, and sometimes posing us with new ones.

The budding horse geneticist should realise that horse genetics is a rapidly expanding science (which hopefully, with proper funding, may remain so). There is still plenty to discover. There will still be exciting challenges in the coming years, followed by new revelations to delight and help us all. Advances in genetics will take away some of the frustrating, time-wasting, costly, and occasionally heart-breaking, aspects of horse breeding. The horse breeder will nevertheless still find his or her hobby or profession a mixture of challenges and achievements - enough to fire a life-time of enthusiasm!
The lesson plan for this course is given below:

**Module one: Genetics and basic inheritance**
What is Horse Genetics?
Lesson one: Genes, alleles and chromosomes
Lesson two: Inheritance and reproduction in the horse
Lesson three: Mendelian inheritance
Module one assignment

**Module Two: Modified and complex inheritance**
Lesson four: Modified Ratios
Lesson five: Genetic Linkage
Lesson six: Complex Traits and Polygenic Inheritance
Module two assignment

**Module three: Equine molecular genetics**
Lesson seven: An introduction to molecular genetics
Lesson eight: Finding and characterising genes for a particular phenotype
Lesson nine: Molecular genetics testing
Module three assignment

**Module four: Evolution and natural selection**
Lesson 10: Evolution
Lesson 11: Breeding Systems
Lesson 12: Domestication and ancient breeds
Module four assignment

**Module five: The genetics of coat colours and patterns**
Module Introduction
Lesson 13: Basic colour genetics
Lesson 14: Dilute colours
Lesson 15: White and white patterns
Lesson 16: Appaloosa patterns
Module five assignment
Final assignment
The module assessments are a mixture of multi-choice, short answer and analytical questions. They help you to review and reinforce your knowledge, with some emphasis on learning to apply it to realistic horse genetics problems. The final assessment is in the form of either an essay or report on some aspect of horse genetics of your own choosing. This allows you to develop your understanding of a particular aspect of horse genetics that is of interest or significance to you. It is your chance to express yourself and demonstrate your own individual enthusiasms - it is your chance to shine! Many possible examples are provided to help you choose, if you are uncertain about the opportunities available. Each of the six assignments carries equal weighting.

**Horse Genetics Website**

I have made a website, which some of you will probably have seen, at

Remember the website probably won’t be as up-to-date as the course notes you’re studying, nor is it as comprehensive. Nevertheless you may find it fun (and useful) to look at the photos of the beautiful horses and ponies presented on there, especially when studying the final module on horse colour and patterns.
Hello to you all, and thank you for choosing to study with me. I hope you enjoy the experience and learn many interesting things along the way.

My name is Glynis Scott and I have been interested in genetics, horses and animal breeding since I was a child. At age 11 I got my first bay cob called Ches, a quiet horse who taught me to ride. Later came a chestnut Anglo-Arab called Little John. He was anything but quiet, but taught me a great deal about horse behaviour and training. Without any real help I somehow turned him from a rearing tearaway into a quiet gentleman, who I will forever miss. Although Ches and John were both geldings I always dreamed of being able to breed horses. My then blacksmith, Lou Friar, owned a cremello mare that produced a string of palomino (and by luck filly) foals. I heard about this and was fascinated (but probably too shy to ask him much about it at the time). I had to find out how and why, and I knew that one day I would too own such a mare - somehow! For a long time though I stuck to breeding ducks, rabbits and the school mice!

After taking a degree in science and a doctorate in genetics I went on to become a professional theoretical and research geneticist, and a lecturer of genetics at the local university. This included co-authoring a genetics text-book and teaching some horse molecular genetics lectures on a Masters in Equine Science. I bred hamsters for a few years, using them to demonstrate genetics principles to my students (live exhibits only, and all of them well loved pets). It’s surprising how much a few cuddly cute animals can inspire students to take an interest in a lesson!

For many years my life was one of work, work, work - and little else. Then, a few years ago, I decided it was time to change that, and re-indulge some of my former hobbies. I bought a beautiful Foundation bred Quarter Horse cremello filly foal called Kitty, thus fulfilling that ambition I had as a child. She is surely a fabulous and intelligent horse. I plan to breed from Kitty, to produce palomino pure-bred Quarter Horse foals (and at last I have my eye on a
stallion, just recently arrived in Wales, and by co-incidence related to my other mare).

Recently I imported another mare, Smart Little Emily, otherwise known as Tyra. She’s a sweet natured chestnut mare from the US, and related to (and the spitting image of!) Smart Little Lena, the US cutting stallion. She’s small at about 13.3hh (possibly the shortest AQHA mare in the UK), and I plan to breed foals suitable for children and adults. I hope for her to become my sons horse in time, if he shows an interest. For a while I also had the sweetest half Arab grey mare, who I taught to do the odd trick. I swapped her, a few months ago, for a black Dartmoor gelding for my son James - and I’m busy trying to bomb-proof him right now!

Apart from the real horses I started to develop a greater interest in their genetics, and developed a website: horse-genetics.com. I got a lot of interest in the site, and a lot of enthusiastic help with the provision of pictures and information - for which I am truly grateful.

Over time I came to realise that it would be nice to thoroughly update and extend the information on horse genetics and produce a course on it, especially as the horse genome project had lead to a sudden explosion of knowledge in the area of horse genetics. The course would give enthusiasts a good grounding in genetics, as it pertains to horses, without being inaccessible to students who don’t already have a degree to their name. At about that time I myself took a diploma in animal behaviour (getting a distinction). That, I thought, could be the answer. There are no other such courses, but clearly there’s an interest in horse genetics. And so this course was born.
Horse Genetics
Module One
Genetics and Basic Inheritance
Module one: Genetics and basic inheritance

What is Horse Genetics?
Horse genetics is the science of heredity as applied to horses

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Genes occur in different forms called alleles
Each gene is represented twice in any particular individual
Genes are organised on chromosomes
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Summary

Module one assignment
What is Horse Genetics?

**Horse genetics is the science of heredity as applied to horses**

The word ‘genetics’ was first used to describe the *science of heredity* by William Bateson in 1907. Horse genetics is therefore the science of heredity as applied to horses. Heredity is the transmission of characteristics from parents to offspring.

Heredity is the reason for the similarity between parents and their offspring: it’s why plants, animals and microbes reproduce progeny of their own species, and not of some other kind. Your parents, and your children if you have them, are human, and not cats or chimpanzees. Your horse’s parents are horses and not zebras or donkeys!

The basic principles of heredity were first clearly described in 1866 by the Austrian monk Gregor Mendel, but his work remained largely unknown until it was “rediscovered” in 1900. Although Mendel established the principles of heredity through his work on pea plants they apply equally to horses as they do to plants and other animals.

Even though the science of genetics, and therefore of horse genetics, has only formally existed since the 1900s people have recognized the value of selectively breeding their crops and animals for at least 4000 years, probably for much longer. Recent evidence (2008) indicates that dogs were probably originally domesticated from wolves up to 100,000 years ago. Humans may’ve been shaping their evolution ever since, sometimes unwittingly, but often intentionally.

Horse domestication is thought to have first occurred on the Eurasian Steppes in the Near East, possibly around 4500 BC. The nomads living there had already domesticated dogs, cows, sheep, and goats, and so were probably aware of the benefits of selective animal breeding. Horses were used to carry their belongings, but they were also used to provide milk, meat and skins. The early horse breeders doubtless discovered that some desirable characters could be passed from parents to offspring: they were practising horse genetics even though they knew nothing about the principles and theory of horse genetics.
These days when we study horse genetics we want to know how heredity can account for the differences (variation) as well as the similarities between individual horses.

Modern day genetics - including modern horse genetics - encompasses not only the principles of heredity but also the study of the molecular nature of genes, and the biochemical reactions with which they are associated. Not only do geneticists study how characters are transmitted through generations they also explore the structure and working of genes and the other genetic material that control those characters. With ever increasing detail, geneticists are learning about how genes control and influence the characters they do. This is true for horse genetics too.

The structure and function of various genes has been elucidated, including those for some important horse genetic disorders, such as severe equine combined immunodeficiency that particularly occurs in Arabians and hyperkalemic periodic paralysis disorder in horses descended from the Quarter Horse stallion Impressive. Horse genetics has long been associated with the genetics of coat colour and this too is now being understood in ever increasing detail, although more still remains to be learned.

Students of horse genetics should always remember that variation is caused by both heredity and the environment (and the interaction between the two). Different breeds of horses have distinctive physical features, including, for example, their average height and build, and the shape of their heads. We know that these differences are largely due to heredity because horses retain their breed characteristics regardless of the environments in which they live. American Quarter horses look like Quarter horses whether they live on a large ranch in America, a small stud in Europe or as a youth’s horse in Australia. On the other hand a Shire and a Shetland will look different from one another even if they are raised at the same stables and trained by the same person.

The environment will nevertheless influence such characters as weight and muscle size, which will vary according to nutrition and exercise. Athletic ability and temperament also depend on various environmental factors, including nutrition, exercise, and the skill and temperament of trainers. A dressage champion may only have champion offspring if those
offspring are trained and ridden by expert trainers and riders. Accident is another form of environmental influence that can affect a horse’s appearance. Mares and stallions with a blemish might still make good breeding stock if the blemish is non-hereditary.

**Remember**

- genetics is the science of heredity
- heredity is the reason for the similarities and differences between parents and their offspring
- variation is caused by both heredity and the environment
Lesson one: Genes, alleles and chromosomes

Introduction

If you are fairly new to genetics there will be a lot of new terminology in this section. DON’T PANIC!

This section takes you through some of the basic principles relevant to horse genetics, gradually introducing new terms and concepts which will soon become familiar to you. The more carefully you read and think about this section the easier you will find the rest of the course.

You may well find that before too long friends will start asking you questions about their own horse genetics interests. If you prepare yourself well now you may be able to help them sort out problems and curiosities which to them had seemed quite baffling!

Remember: you may learn a lot of new terms, but you’ll soon become used to them.

Genes are the units of heredity

Genes are the units of heredity. They are too small to be seen directly, but the results of their action can be seen in all of life, including horses. Anyone who studies horse genetics should have some understanding of what genes are and what they do, even if it's only at an elementary level.

Genes are like pieces of code that say how the molecular building blocks of living organisms will be built, what will be put or done where, and when. This in turn determines what those organisms look like and how they work (or don't). The genes are encoded by a molecule called DNA (deoxyribonucleic acid). Since Watson and Crick discovered the structure of DNA in 1953 there has been an explosion of knowledge that has revolutionised genetics, including horse genetics. As a consequence we now have quite a
According to the human genome project humans have about 30,000 genes, which is many fewer than was thought. This is only a few thousand more than mice are thought to have. Horses have about the same number of genes as us. With the exception of some very special cells, which are involved in making our immune systems work, each of our cells contains all of the 30,000 or so genes - the same is true of horses. Since (almost) all body cells inherit the same genetic information we might ask why they aren’t all identical to one another. In general the reason is to do with the way in which the different parts of the genetic information are used (or not used) in different cells. Different cells may have the same genes but only a small proportion of them are working at any one time. The genes that are working in one sort of cell may be switched off in another, and vice-versa.

When we study horse genetics we might often only be concerned with how genes are passed on from parents to offspring. This kind of information can answer questions about a foal’s possible colour, or the likelihood of it inheriting some genetic disorder. Knowledge of this aspect of horse genetics can be used to plan breeding programs to optimise the likelihood of foals with a certain sets of characteristics, and/or to minimise the spread of undesirable characters and genetic disorders.

Another branch of horse genetics is more concerned with the molecular nature (construction) of genes and the way they work, or don’t work. Research in this area leads to an increased understanding of genetic disorders and can lead to genetic tests for identifying carriers, as well as to new or improved methods of managing or treating disorders in known sufferers.

**Remember**

- genes are the units of heredity
- genes are encoded by a molecule called DNA (deoxyribonucleic acid)
- most cells of a body have the same genes, but different genes are on and off in different types of cells, and at different times of development
Genes occur in different forms called alleles

Genes for any particular character may occur in slightly different forms, called alleles. **Alleles are therefore alternative forms of a particular gene.** To understand horse genetics you need to understand about alleles and how they work.

Alleles are gene variants caused by mutations ("genetic mistakes"). Each allele has a slightly different code and may make a slightly different product, or control a process in a slightly different way. It is rather like models of a particular make of car, in that each model is essentially similar with just some minor differences. (Hopefully!) each model of car will transport you around wherever you want to go, but some will do it faster, some will be more comfortable, or look sleeker, while others will have greater fuel efficiency, and yet others will be better for particular types of terrain.

**Remember**
- alleles are alternative forms of a gene
- alleles are gene variants caused by mutations

Each gene is represented twice in any particular individual

Each gene is represented twice in each cell. This means that any individual can have its 2 copies of the gene as the same allele, when it is called **homozygous** for that gene, or as two different alleles, when it is termed **heterozygous** for that gene. These two terms crop up again and again it’s important that you know what they mean and understand why each gene is represented twice. I’ve provided definitions of the terms below. After that follows a discussion of chromosomes, and then heredity and reproduction in lesson 2. These will help you to understand how genes are inherited between individual horses.

**Remember**
- **Homozygous:** Both copies of the gene of interest are the same in the individual in question: they are a copy of the same allele.
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- **Heterozygous**: An individual's two copies of the gene in question are different: there are two different alleles for the gene.

**Genes are organised on chromosomes**

Genes don't just float around in cells. They are spaced out along a (relatively) few long continuous strands of DNA. Often the analogy of beads on a string is used. This is quite a good analogy, though the beads aren't spaced out evenly: sometimes there are clusters, sometimes there are long spaces between the genes. Furthermore we now know that there's much more string than beads, with much DNA not coding for anything at all!

*The DNA strands are very long and are folded, coiled and held together in a way that makes up structures called chromosomes.* These can be fitted into cells without becoming tangled and broken, when genes might otherwise be damaged or lost. They help to ensure that heredity takes place without making too many mistakes that might otherwise result in abnormal and in-viable cells and offspring.

*Horse cells have 64 chromosomes each. The chromosomes of each cell are located in a sub-cellular compartment called the nucleus.* Particular places on a chromosome are called loci (singular locus). Each particular gene has a particular place, or locus, on a particular chromosome. *A particular gene is always located at the same locus.*

**Remember**

- genes are arranged on chromosomes
- chromosomes help package long DNA strands, which might otherwise be damaged
- the site of a particular gene is called a locus
- horses have 64 chromosomes in each body cell nucleus
Chromosomes occur in pairs

Each cell of a horse’s body contains **two copies of each chromosome** - one from the dam (maternal) and one from the sire (paternal). **In horses there are 32 pairs of chromosomes.** The paternal and maternal chromosome of each pair are said to be **homologous.** This means that they have same genes, and other structural features, at corresponding places along their length (and they are therefore structurally the same). The same applies to most other animals, including us. It is referred to as **diploidy.**

As each cell derives from one original fertilised egg cell all the cells have the same genetic material (the immune system cells have only part of this, rearranged somewhat, but they are a special case).

Because of diploidy each gene is represented twice - once on the maternal chromosome of a pair, once on the paternal chromosome. (Remember that each individual can be homozygous or heterozygous for any particular gene.)

The sex chromosomes are non-homologous

The exception to chromosomes occurring in homologous pairs is the chromosomes that determine sex. Female mammals, including mares, have two copies of a chromosome called the X chromosome - **these are homologous.**

Males though have only one X chromosome (from their mum) and one smaller chromosome, called a Y chromosome (from their dad). **The X and Y chromosomes are not homologous.** This is why genes on the X chromosome may be inherited differently in males and females (males can only have one copy of each gene that occurs on the X chromosome). **Because of this X chromosome genes are said to be sex-linked.** The Y chromosome has very few genes, which are, of course, only present in males.

An example of a sex-linked gene is that causing haemophilia A (factor VIII deficiency).
Although the example is not unique to horses, haemophilia A has been reported rarely in Thoroughbred, Quarter Horse and Standardbred colts (Archer, 1961, Henninger, 1988, Hutchins et al, 1967). Sex linkage is discussed in more detail later.

**Remember**

- **chromosomes occur in homologous pairs, which is called diploidy**
- **one chromosome of each pair is from the dam, the other is from the sire**
- **each body cell is derived from a single fertilised egg cell and is therefore genetically the same as all the others, immune system cells are the exception and the genetic material in these cells is altered after they are made**
- **the sex chromosomes are not homologous**
- **only colts inherit Y chromosomes, fillies do not**

**A few genes are not in the cell nucleus**

A few genes reside in cellular organelles called mitochondria. Mitochondria are cell organelles involved with energy metabolism. They **have their own genome** and are found outside of the cell's nucleus. Their genomes contain genes involved with energy release in muscle cells.

Mitochondria are thought to have originated from separate organisms that lived symbiotically within the cell, gradually evolving so as to become a vital part of it, and unable to live without it. There are thousands of mitochondria in a typical cell, muscles have an estimated average of about 10,000 copies per cell. Mitochondrial DNA (mtDNA) is passed from mother to child, that is to say it is **maternally inherited**. It can be passed to a colt and function within him, but stallions do not pass on mitochondrial DNA to their offspring.

Harrison and Turrion-Gomez (2006) reported that mitochondrial gene variation may
contribute to performance variability between thoroughbred racehorses. They observed extensive mitochondrial gene variation in the thoroughbred racehorse population, and found associations between mitochondrial genotypes and aspects of racing performance. This isn’t surprising given the function of mitochondrial genes in energy production and release in muscles. The genes are already known to affect the athletic performance of humans, with mutations being associated with muscular disorders, heart muscle problems and exercise intolerance.

Remember

- Mitochondria are maternally inherited cellular organelles that have their own genome.
- The genes in mitochondria are involved with energy metabolism and contribute towards performance characteristics in horses, especially important in racehorses.

Summary

Genes are the units of heredity, encoded by DNA and packaged into special structures called chromosomes. The site of a particular gene on a chromosome is called a locus. Horses have 64 chromosomes in each body cell nucleus, with all but the sex chromosomes in males occurring in homologous pairs. Body cells are said to be diploid, with each gene (except sex linked ones in males) represented twice, once on each chromosome of a pair. Alternative forms of a gene can occur through mutation, and are called alleles. If both copies of a gene are of the same allele the individual is said to be homozygous for that gene, otherwise it is heterozygous. Only colts inherit Y chromosomes, fillies do not.

Of each chromosome pair one is inherited from the dam, the other is from the sire. Each body cell is derived from a single fertilised egg cell and is therefore genetically the same
as all the others. The difference between cells is due to different genes being on and off in different types of cells, and at different times of development. Immune system cells are the exception as the genetic material in these cells is altered after they are made.

A few genes are in the genomes of sub cellular organelles called mitochondria, which are thought to have a bacterial origin. These genes are almost always only inherited from the dam, being present in egg cells. They're therefore said to be maternally inherited. Mitochondria are important for energy metabolism and may affect performance, especially in race horses.

References

Lesson two: Inheritance and reproduction in the horse

Introduction

This “heredity and reproduction” discussion is not about the practicalities of breeding horses. Rather it is a basic account of what happens to genes and chromosomes during reproduction. The example at the end of the lesson illustrates some of the principles and concepts from this and the first lesson.

An introduction to inheritance and reproduction in the horse

During horse reproduction the dam donates an egg and the sire donates sperm to each of its progeny. A single sperm fuses with the egg to produce a fertilised egg – called a zygote (in the case of non-identical twins the dam produces two eggs, each of which is fertilised by a single sperm). From the zygote a new individual may develop. The division from the single-celled zygote leads to the production of all the cells that make up the body. These body cells are called somatic cells and are the units of structure and function that comprise all living organisms. Each somatic cell inherits a copy of the genetic material present in the zygote.

The eggs and sperm can be thought of as the vehicles of heredity: they carry the genes that determine the similarity of the offspring to their parents, and the heritable differences between offspring. It is during their formation in the male and female reproductive organs that the processes that give rise to heredity and variation occur.

If all body cells inherit the same genetic information we might ask why they aren’t all identical to one another. In general the reason is to do with the way in which the different parts of the genetic information are used (or not used) in different cells. Eventually the sex organs will develop from somatic cells, and within these the next generation of sperms or eggs will form.
The genetic part of the process that gives rise to eggs and sperm is called **meiosis**. It causes the division of a body cell into two gametic cells (the general name for eggs and sperm cells and their precursors). Each gametic cell has only one copy of each chromosome pair present in the body cells. For each chromosome pair any particular gametic cell inherits one at random – in some cells it might be the maternal chromosome and others the paternal chromosome. In this way the chromosomes are mixed up and each gametic cell is genetically different. Such cells are said to be **haploid**, compared with somatic cells which are diploid. The haploid chromosome number is denoted $n$, the diploid number is $2n$. In horses $n=32$ and $2n=64$.

When two haploid gametes are fused at fertilisation, the diploid number of chromosomes is restored in the zygote. **The alternation of meiosis and fertilisation in the life cycle maintains the constancy of the chromosome complement between generations** (see figure 1).

During the process of meiosis, the paternal and maternal chromosomes of each pair come together and may exchange parts with one another (a process called **crossing over**, for obvious reasons). This occurs in such a precise way that the recombined chromosomes neither lose or gain information, but simply carry new combinations of the different alleles that were present in the maternal and paternal partners (i.e. alleles are swapped). So the maternal chromosomes will carry some alleles originally present in the paternal chromosomes and vice-versa, **creating new combinations not present in the body cells of the parents**. This causes extra variation in foals compared to the parents, so the foals carry some characters of each parent, but there is still variation between siblings (brothers and sisters).

**Remember**

- A horse's body cells are diploid and have 32 pairs of chromosomes.
- The eggs and sperm, and their precursor cells, are called gametic cells.
- Gametic cells are haploid and have 32 chromosomes, one of each pair.
- Meiosis creates gametic cells out of body cells.
gametic cells contain mixes of maternal and paternal chromosomes creating variation

fertilisation fuses an egg and sperm to form a diploid zygote, which hopefully is the beginning of a new and unique foal!

Figure 1: The alternation of meiosis and fertilisation in the life cycle maintains the constancy of the chromosome complement from one generation to the next.
Genes, alleles, chromosomes and inheritance

Meiosis distributes a complete haploid set of chromosomes into each of the gametes. The gametes in turn transmit their chromosome sets to the zygotes formed at fertilisation. In this way genes are handed on from parents to their offspring.

Meiosis allows for variation since different gametes have different combinations of parental chromosomes. The diploid cells which undergo meiosis have two sets of chromosomes - one set from each parent, which we call the maternal and paternal sets. Although each set carries a full quota of genetic information, the genes which specify individual character differences may be present as different alleles.

*The genetic make-up of an individual (or a cell) is called its genotype.* Individuals with different genotypes may look different to one another, when they will have different characteristics. It is these characteristics which are the external expression of the genotype, which is called the **phenotype**. Sometimes two genotypes can give the same phenotype.

**Example:**

The gene which determines whether a horse will turn grey occurs in two forms, alleles which are symbolised as $G^+$ and $G^g$. Each horse has two copies of the grey gene - one from its dam, one from its sire - and may therefore have genotype $G^+G^+$, $G^+G^g$ or $G^gG^g$ for this particular gene. Each egg or sperm has only one copy of the grey gene, which may be of allele $G^+$ or $G^g$.

Individuals with genotypes $G^+G^+$ and $G^gG^g$ are homozygous. They will produce gametes of only one genotype at the grey locus.

Individuals with genotype $G^+G^g$ are heterozygous. They will produce gametes of both genotypes at the grey locus, with approximately equal proportions of both types, $G^+$ and $G^g$. 
G^G^G and G^*G^G horses have the grey allele and are therefore grey: their phenotype for this gene is grey. Horses of genotype G^*G^* have coats of some other color (non-grey).

In this example allele G^G is said to be dominant over G^* because the presence of one copy of G^G causes greying: horses of genotype G^G^G and G^*G^G look the same.

Allele G^* is said to be recessive to allele G^G and its effect is over-ridden in the presence of allele G^G (i.e. in heterozygotes of genotype G^*G^G). Some genes have more than two alleles, and so have more than three genotypes.

Remember:

- the genetic make-up of an individual is called its genotype
- the external expression of the genotype is called the phenotype
- if an allele has to be homozygous to affect the phenotype it is said to be recessive
- if an allele over-rides another it affects the phenotype even when it is heterozygous - the over-riding allele is said to be dominant

The curly coat genes: an example of some important concepts

Most horses have a smooth coat that lies somewhat flatly against the skin. Some horses though have a wavy or curly coat. In the extreme the mane and tail may hang like ringlets, with the hair said to be “cork-screwed”.

There are at least two separate genes that cause a curly coat. One which occasionally occurs, for example in Mustangs and Rocky Mountain Horses, is dominant – that’s to say the curly coat allele is dominant over the smooth coat allele. Another is recessive and has rarely occurred in Quarter Horses and other breeds.

One breed, the Bashkir Curly, has been developed around curly coated horses. A very few
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breeders specialise in curlies in other breeds. In these breeds there has been positive artificial selection for horses with “curly alleles” at the curly coat genes, causing an increase in the frequency of such horses.

Selecting for the gene with a dominant curly allele is easier than selecting for a gene with a recessive curly allele. This is because when an allele is rare it usually occurs in heterozygotes: a rare allele is more likely to partnered with the common one than another rare one. Heterozygous individuals with a dominant curly allele have the curly phenotype. If they are crossed with smooth coated horses about 50% of their foals would be curly coated also. Therefore there can be out-crossing to non curlies to increase the genetic base of curlies and avoid some of the perils of in-breeding (which we will consider in more depth later in the course). This is quite important since initially (i.e. when the allele is rare) individuals carrying it will be quite closely related, all descending from the horse in which the mutation first occurred.

Some breed authorities forbid the registration of horses that have a curly coat, for example, the American Quarter Horse Association. This effectively acts as negative artificial selection since curly horses are “lost” from the breed. Such horses can pass their genes on to their offspring, but those offspring would no longer be considered part of the breed! Dominant genes would quickly be lost from the breed. However genes with recessive curly alleles would remain in rare heterozygotes since their phenotype would be smooth coated. These would be indistinguishable from other smooth coated horses unless they had a curly foal. Occasionally two heterozygotes might be mated by chance and a curly coated foal could then occur, but not be allowed to be registered, even if both parents were pure-bred Quarter Horses.

Starting a breeding program from curlies with a recessive curly alleles could be very difficult and require a lot of time (and money!). This is because the gene is rare and mostly in heterozygous smooth coated horses who are related to one another to some degree. You would need to be able to find horses separated from each other by several generations, preferably at least two stallions and two mares (so you could later breed from their offspring). At some point new blood would need to be introduced to avoid inbreeding, most likely from smooth coated individuals who don’t carry curly alleles. Then there would
be some generations of foals that were carriers of the curly alleles but had smooth coated phenotypes.

If there was a popular stallion who happened to carry a recessive curly allele then about 50% of his offspring would also carry it (a carrier is an individual heterozygous for some recessive trait). Some stallions can sire very many offspring in a lifetime. Furthermore some people “line breed” to a particularly successful stallion. If this were to happen then the frequency of the curly allele would increase in the population. It might take several generations but curly might eventually start cropping up in the breed of the stallion. It would then become easier for an enthusiast to start breeding recessive curlies. This argument is true for any rare recessive trait.

This discussion might seem academic – most of you probably prefer smooth coats anyway, or don’t really care one way or the other. However there is another feature of curly coats that could make them an increasingly popular choice in the future: curly coated horses are hypoallergenic. People who suffer from allergy to horse’s hair tend not to be bothered by curly coated horses. There are more such people than you might think, and some of them have to give up riding and horses altogether. How much better if these people could have a hypoallergenic horse! My own husband is allergic (and by coincidence also my next-door-but-one neighbour, who also has a horsey wife). If my son (who is only 18 months old right now) turns out to be allergic to horses but wants to ride I know what I’ll be doing!

The curly coat seems to be associated with other characters, possibly because genes for them are linked to (i.e. on the same chromosome as) the curly mutation. They are said to have a particularly quiet temperament, with them being less “flighty” than many horses. This could make them a popular choice with parents! In addition their curly coats, which are most curly in the winter, have better insulating properties than smooth coats, providing greater protection from cold and wet, and therefore making curlies particularly hardy.
Remember:

- **it is easier to breed for traits that are caused by rare dominant alleles than rare recessive ones**
- **within a breed, horses with rare recessive alleles tend to be heterozygous and related**
- **line breeding increases the frequency of rare recessive alleles, when they may start occurring as homozygotes**

Because line breeding increases the frequency of rare recessive alleles and individuals homozygous for them it can also increase the frequency of rare genetic disorders. This important aspect of in-breeding is discussed in more detail later in the course.

**Summary**

As we saw in the first lesson a horses body cells are diploid and have 32 pairs of chromosomes. The eggs and sperm are (gametic cells) and are haploid with only 32 chromosomes, one of each pair. Meiosis creates gametic cells out of body cells, mixing maternal and paternal chromosomes to create variation. The fusion of an egg and sperm during fertilisation leads to a diploid zygote, which hopefully is the beginning of a new and unique foal.

The genetic make-up of an individual is called its genotype, while the external expression of the genotype is called the phenotype. Recessive alleles have to be homozygous to affect the phenotype, while dominant alleles over-ride recessive ones and affect the phenotype even when they're heterozygous. It is easier to breed for traits that are caused by rare dominant alleles than rare recessive ones. Within a breed, horses with rare recessive alleles tend to be heterozygous and related. Line breeding increases the frequency of rare recessive alleles, when they may start occurring as homozygotes. Both desirable and undesirable recessive traits can start occurring during line breeding.
Lesson three: Mendelian inheritance

Introduction

Gregor Mendel unravelled the laws of heredity - before either chromosomes or genes were known about - by observing the inheritance patterns of various characters of garden pea plants. Although Mendel established the principles of heredity through his work on pea plants they apply equally to horses. The science of horse genetics is based on an understanding of Mendelian inheritance.

Mendel's successors expressed his findings in the terms of two laws of inheritance, usually using modern terminology that Mendel himself would've been unaware of. The observations now discussed make up the basis of what is often called Mendel’s First Law, or ‘The Law of Segregation’.

Remember: to understand horse genetics you will need to understand Mendelian inheritance

Mendel’s First Law of Segregation

Mendel realised that inheritance could only be explained if the elements that determine characters existed in pairs. These are now known to be the allele pairs of a gene, existing on homologous chromosomes (but Mendel didn’t know that at the time).

Mendel worked out that allele pairs are separated in the gametes, so that eggs and sperms carry just one member of each pair (actually for him it was the eggs and pollen, since he mainly worked on peas, but the same applies to animals). Whether any particular gamete carries one allele or the alternative one is determined by chance so that either is equally likely. So, for example, if we consider the dun gene for a heterozygous dun stallion approximately half its sperm will have the dun allele
and the half will have the non-dun allele.

When an egg and sperm fuse during fertilisation new allele pairs are formed. One generation thus carries the new pairs brought together from the parents of previous generation.

Mendel’s deductions can be summarised into his “first law of segregation” thus: *Contrasting forms of a character are controlled by pairs of unlike alleles that separate in equal numbers in the gametes during meiosis.*

Applying Mendel’s First Law to horse genetics we can work out the possible phenotypes of a foal from a particular mating, as long as one main gene controls the feature of interest and we know the genotype of the parents at that locus.

For example we’ll consider the probability of a grey foal from a grey mare and a chestnut stallion. The chestnut stallion is of genotype $G^+G^+$ (i.e. is homozygous for “non grey” alleles). The grey mare had only one grey parent herself and so must be heterozygous for the grey allele, i.e. of genotype $G^+G^G$. We can construct something called a Punnett Square to visualise this cross (always reminds me of strawberries!)

<table>
<thead>
<tr>
<th>Genetic contribution from mare</th>
<th>Genetic contribution from stallion is $G^+$</th>
</tr>
</thead>
<tbody>
<tr>
<td>50% chance of either allele in the egg</td>
<td>Only $G^+$ alleles in the sperm</td>
</tr>
<tr>
<td>$G^+$</td>
<td>50% chance: $G^+G^+$ not grey</td>
</tr>
<tr>
<td>$G^G$</td>
<td>50% chance: $G^G^+G^+$ grey</td>
</tr>
</tbody>
</table>
If we had a grey heterozygous mare and a grey heterozygous stallion, i.e. both of genotype G\(^+\)G\(^G\) then the cross would be as follows:

<table>
<thead>
<tr>
<th>Genetic contributions from dam (egg)</th>
<th>from sire (sperm)</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>50% chance of either</td>
<td></td>
<td></td>
</tr>
<tr>
<td>G(^+)</td>
<td>G(^+)</td>
<td>G(^G)</td>
</tr>
<tr>
<td>25% chance:</td>
<td>25% chance:</td>
<td></td>
</tr>
<tr>
<td>G(^+)G(^+)</td>
<td>G(^+)G(^G)</td>
<td></td>
</tr>
<tr>
<td>Not grey</td>
<td>Grey</td>
<td></td>
</tr>
<tr>
<td>G(^G)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>25% chance:</td>
<td></td>
<td></td>
</tr>
<tr>
<td>G(^G)G(^+)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Grey</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Grey</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Altogether there’s a 75% chance of a grey foal (25%+25%+25%).

Another way of saying this is that there’s a 3:1 ratio of grey to not grey. If the same two had several foals roughly ¼ of them would be non-grey, the more foals there were the closer would be the real observed ratio to this theoretical ratio. If we consider all matings that have been made between two heterozygous grey horses then the observed ratio would be even closer to the theoretical ratio.

The 3:1 ratio is what you normally expect when mating together heterozygous horses (for any one character). Approximately ¾ of foals are expected to be of the
dominant phenotype while \( \frac{1}{4} \) of them are expected to be of the recessive phenotype. **Geneticists call this kind of mating a monohybrid cross.** The “mono” part of monohybrid refers to the fact that only one gene is being considered. The “hybrid” part refers to the fact that both parents are heterozygous.

**Remember**
- **Mendel’s first law of segregation:** Contrasting forms of a character are controlled by pairs of unlike alleles that separate in equal numbers in the gametes during meiosis.
- **Mating together horses that are heterozygous for any particular character of interest is to make a monohybrid cross.**
- **Monohybrid crosses result in expected 3:1 ratios:** approximately \( \frac{3}{4} \) of foals are expected to be of the dominant phenotype while \( \frac{1}{4} \) of them are expected to be of the recessive phenotype.
- **If only a single monohybrid cross is made then the 3:1 ratio can be thought of in terms of chance,** there being a 75% chance of the dominant phenotype and a 25% chance of the recessive phenotype.

**Considering two characters at once**

Mendel wanted to know what happens when two pairs of contrasting characters are combined together in the same hybrid. We can illustrate what he found by considering examples. Let’s first consider an example of the inheritance of a colour and a pattern character in horses.

Mary has two bay mares that are homozygous for the tobiano spotting allele \((To^T)\) also for the bay allele at the agouti locus \((A^a)\).

A neighbour has a couple of homozygous black stallions and Mary took both her mares to the neighbour’s stallions. The foals would have to inherit one copy each of the bay and black alleles at the agouti locus (i.e. they will be \(A^aA^a\)). They will also be heterozygous for the tobiano spotting allele \((To^T)\). The foals, one colt and one filly,
were both bay and spotted. (Note that all horses were of genotype $E^*E^+$ at the extension locus so no red foals are possible.)

After a few years Mary intends to breed the filly and colt together (they’re unrelated). By constructing a Punnett Square she is able to know the chances of producing foals of the 4 possible different phenotypes:

<table>
<thead>
<tr>
<th>Genetic contributions from sire</th>
<th>from dam</th>
</tr>
</thead>
<tbody>
<tr>
<td>$T^*A^A$</td>
<td>$T^*A^A$</td>
</tr>
<tr>
<td>$T^*T^*A^A^A$</td>
<td>$T^*T^*A^A^A$ bay tobiano</td>
</tr>
<tr>
<td>$T^*T^*A^A^A$</td>
<td>$T^*T^*A^A^A$ bay tobiano</td>
</tr>
<tr>
<td>$T^*T^*T^*A^A^A$</td>
<td>$T^*T^*T^*A^A^A$ bay tobiano</td>
</tr>
<tr>
<td>$T^*T^*T^*T^*A^A^A$</td>
<td>$T^*T^*T^*T^*A^A^A$ bay tobiano</td>
</tr>
<tr>
<td>$T^+A^A$</td>
<td>$T^+A^A$</td>
</tr>
<tr>
<td>$T^+T^+A^A^A$</td>
<td>$T^+T^+A^A^A$ bay tobiano</td>
</tr>
<tr>
<td>$T^+T^+T^+A^A^A$</td>
<td>$T^+T^+T^+A^A^A$ bay tobiano</td>
</tr>
<tr>
<td>$T^+T^+T^+T^+A^A^A$</td>
<td>$T^+T^+T^+T^+A^A^A$ bay</td>
</tr>
<tr>
<td>$T^+A^A$</td>
<td>$T^+A^A$</td>
</tr>
<tr>
<td>$T^+T^+A^A^A$</td>
<td>$T^+T^+A^A^A$ bay tobiano</td>
</tr>
<tr>
<td>$T^+T^+T^+A^A^A$</td>
<td>$T^+T^+T^+A^A^A$ bay tobiano</td>
</tr>
<tr>
<td>$T^+T^+T^+T^+A^A^A$</td>
<td>$T^+T^+T^+T^+A^A^A$ bay</td>
</tr>
<tr>
<td>$T^+A^A$</td>
<td>$T^+A^A$</td>
</tr>
<tr>
<td>$T^+T^+A^A^A$</td>
<td>$T^+T^+A^A^A$ bay tobiano</td>
</tr>
<tr>
<td>$T^+T^+T^+A^A^A$</td>
<td>$T^+T^+T^+A^A^A$ bay tobiano</td>
</tr>
<tr>
<td>$T^+T^+T^+T^+A^A^A$</td>
<td>$T^+T^+T^+T^+A^A^A$ bay</td>
</tr>
</tbody>
</table>

There is now an expected 9:3:3:1 ratio of bay tobiano to black tobiano to solid bay to solid black (solid means with no white patches). This is the usual ratio expected when mating together two horses heterozygous for two particular characters. Geneticists call this kind of mating a dihybrid cross.
Since in this case there was just one foal we can think about the ratios in terms of chance. Thus the chance of a foal exhibiting both dominant characteristics (i.e. bay and tobiano) is 9/16 (56.25%).

The chance of a foal exhibiting one particular dominant characteristic and the other recessive characteristic is 3/16 (18.75%), with a 3/16 chance of exhibiting the alternative dominant and recessive characteristics.

The chance of a foal exhibiting both recessive characteristics (here solid and black) is only 1/16 (6.25%).

Another way of thinking about this is to consider what could happen when lots of similar di-hybrid crosses were made. Then 56.25% of foals would exhibit the dominant phenotype at both loci, 18.75% would show each of the mixes (i.e. one dominant and one recessive phenotype) while just 6.25% would show both recessive phenotypes. Of course these are the theoretical expectations and in real life the observed percentages would be approximate. The more such crosses that were considered the closer the real percentages would be to the theoretical ones.

If we consider each trait separately there’s a 3:1 ratio of bay to black and a 3:1 ratio of tobiano to solid colored (or non tobiano). (Note that 12:4 = 3:1). As noted previously this is what you expect from mating together two horses heterozygous for any particular one trait, which is called a **monohybrid cross**.

For those of you with some mathematical ability you will notice that the dihybrid ratio can easily be derived from multiplying out the two monohybrid ratios, e.g.

$$(3 \text{ bay} + 1 \text{ black}) (3 \text{ tobiano} + 1 \text{ solid}) =$$
$$3 \times 3 \text{ bay tobiano} + 3 \times 1 \text{ bay solid} + 1 \times 3 \text{ black tobiano} + 1 \times 1 \text{ black solid} =$$
$$9 \text{ bay tobiano} + 3 \text{ bay} + 3 \text{ black tobiano} + 1 \text{ black}$$

This way of working it out can be extended to 3 phenotypes – to work out a tri-hybrid ratio. For example, adding dun to the above (note that a black dun is called a grullo):
(9 bay tobiano + 3 bay + 3 black tobiano + 1 black) (3 dun + 1 non-dun) =

27 bay tobiano dun + 9 bay dun + 9 tobiano grullo + 3 grullo + 9 bay tobiano + 3 bay + 3 black tobiano + 1 black

Remember

- Mating together horses that are heterozygous for any 2 particular character of interest is to make a dihybrid cross.
- Dihybrid crosses result in expected 9:3:3:1 ratios: approximately 9/16 of foals are expected to be of both dominant phenotypes, 3/16 have one of the dominant phenotypes only with another 3/16 having the alternative one. 1/16 are expected show both recessive phenotypes.
- If only a single dihybrid cross is made then the 9:3:3:1 ratio can be thought of in terms of chance, for example there being a 56.25% chance of the double dominant phenotype occurring in the foal and a 6.25% chance of the double recessive phenotype.

How many foals would I need to get a certain phenotype?

The hybrid ratios tell you how likely any particular phenotype is from a particular cross. It doesn’t tell you how many foals to breed to increase the chances of getting one with a particularly desirable phenotype. There is a branch of statistics (binomial statistics) that allows one to calculate such things – but it is beyond the scope of this course (though many statistics programs can do it for you!).

However for your reference the following look-up table shows the number of foals to breed for certain levels of chance, given some of the more common
ratios. If the chance of getting the phenotype is quite high then you expect to breed fewer foals before getting the desired phenotype. When the chance is small then several or even many foals might be bred before the rare phenotype shows up, though there is always the possibility that one will be lucky! If you want to produce foals of a phenotype that is rare in the cross you’re thinking of making then it may be worth considering if a different stallion, and possibly even a different mare, might be used instead.

<table>
<thead>
<tr>
<th>chance of success</th>
<th>phenotype ratio</th>
<th>1/16</th>
<th>3/16</th>
<th>9/16</th>
<th>¼ (4/16)</th>
<th>¾ (12/16)</th>
<th>½ (8/16)</th>
</tr>
</thead>
<tbody>
<tr>
<td>50%</td>
<td></td>
<td>11</td>
<td>4</td>
<td>2</td>
<td>3</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>60%</td>
<td></td>
<td>15</td>
<td>5</td>
<td>2</td>
<td>4</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>70%</td>
<td></td>
<td>19</td>
<td>6</td>
<td>2</td>
<td>5</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>80%</td>
<td></td>
<td>25</td>
<td>8</td>
<td>2</td>
<td>6</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td>90%</td>
<td></td>
<td>36</td>
<td>12</td>
<td>3</td>
<td>8</td>
<td>2</td>
<td>4</td>
</tr>
<tr>
<td>95%</td>
<td></td>
<td>47</td>
<td>15</td>
<td>4</td>
<td>11</td>
<td>3</td>
<td>5</td>
</tr>
<tr>
<td>99%</td>
<td></td>
<td>71</td>
<td>23</td>
<td>6</td>
<td>16</td>
<td>4</td>
<td>7</td>
</tr>
</tbody>
</table>

**Summary**

To understand horse genetics you will need to understand Mendelian inheritance, which applies to all diploid organisms, including horses. The modernised version of Mendel’s first law of segregation states that contrasting forms of a character are controlled by pairs of different alleles that separate in equal numbers in the gametes during meiosis.

In genetics language the mating together horses that are both heterozygous for any
particular character of interest is to make a monohybrid cross. Monohybrid crosses result in expected 3:1 ratios: approximately ¾ of foals are expected to be of the dominant phenotype while ¼ of them are expected to be of the recessive phenotype. The 3:1 ratio can be thought of in terms of chance, there being a 75% chance of the dominant phenotype and a 25% chance of the recessive phenotype, for any particular cross.

Mating together horses that are heterozygous for any 2 particular character of interest is to make a dihybrid cross. Dihybrid crosses result in expected 9:3:3:1 ratios: approximately 9/16 of foals are expected to be of both dominant phenotypes, 3/16 have one of the dominant phenotypes only with another 3/16 having the alternative one. 1/16 are expected show both recessive phenotypes. When a particular dihybrid cross is made then the 9:3:3:1 ratio can be thought of in terms of chance, for example there being a 56.25% chance of the double dominant phenotype occurring in the foal and a 6.25% chance of the double recessive phenotype.

The hybrid ratios tell you how likely any particular phenotype is from a particular cross. A table is provided to tell you how many foals you are likely to need to breed to get one with a particular desirable phenotype. The smaller the chance of a foal from a particular cross the greater the number of foals that might have to be bred from that cross to achieve the desired phenotype, although of course (and since the numbers are based on averages), some breeders will be luckier than others!
Module one assignment

Q1. What is heredity?
A. Heredity is the transmission of characters from parents to offspring.
B. Heredity describes why the environment affects us.
C. Heredity is the transmission of characteristics from offspring to parents.
D. Heredity is the modern term for genetics.
E. Heredity is something that only occurs in pea plants.

Q2. Which best describes modern day genetics?
A. Genetics is just about the study of heredity.
B. Genetics is about studying the molecular control of genes only.
C. Genetics encompasses the study of heredity, the molecular nature of genes, and the biochemical reactions with which genes are associated. Geneticists learn about how genes control and influence characters.
D. Genetics is concerned only with the passing of characters from parents to offspring.
E. Genetics concerns the making of jeans.

Q3. What are the causes of variation between horses?
A. The environment only.
B. Genetics only.
C. Accidents.
D. Both the environment and genetic differences.
E. Being bought up on the same ranch.
Q4. Which of the following statements about genes is **false**?

A. genes are the units of heredity  
B. genes are encoded by a molecule called DNA (deoxyribonucleic acid)  
C. most cells of a body have different genes from most others  
D. different genes are on and off in different types of cells, and at different times of development  
E. genes are too small to be seen directly, but the results of their action can be seen in all of life  

Q5. Which of the following best describes what alleles are?

A. alleles are proteins  
B. alleles are structures discovered by Watson and Crick in 1953  
C. alleles are a sort of chromosome  
D. alleles are genes that only occur in immune system cells  
E. alleles are alternative forms of a gene caused by mutation  

Q6. Which of following statements is **false**?

A. genes are arranged on chromosomes and the site of a particular gene is called a locus  
B. chromosomes help package long DNA strands, which might otherwise be damaged  
C. horses are diploid, having 32 pairs of chromosomes in each body cell nucleus  
D. only fillies inherit Y chromosomes, colts do not  
E. each of a horses body cells derives from one fertilised egg cell
Q7. Which of following statements is \textbf{false}?

A. a mares eggs are haploid and have 32 chromosomes
B. a process called meiosis creates gametic cells (eggs and sperm) out of body cells
C. a stallions sperm contains a mix of chromosomes originating from his own dam and sire
D. fertilisation fuses an egg and sperm to form a diploid zygote, which hopefully is the beginning of a new and unique foal
E. a stallions sperm is diploid and has 64 chromosomes

The gene which determines whether a horse will be dun occurs in two forms, alleles which are symbolised as $D^D$ and $D^+$. Horses of genotype $D^D D^+$ are dun, those of genotype $D^+ D^+$ are non-dun.

For the \textbf{next 3 questions} choose an answer from the following possibilities:

A. $D^D D^+$
B. $D^+ D^+$
C. $D^D D^D$
D. $D^+$
E. $D^D$

Q8. Which is the homozygous genotype for dun?
Q9. Which is the heterozygous genotype?
Q10. Which allele is the dominant one?
Q11. What is the \textbf{phenotype} of horses with genotype $D^D D^D$ (one word is sufficient)?

Sally has a mare with a red dun phenotype. She has had four foals by the same chestnut (non-dun) stallion. Two foals were red dun and two were chestnut.
Q12. What is the genotype of the red dun mare at the dun locus?
Q13. Is she homozygous or heterozygous at the dun locus?
Q14. What are the possible genotypes of her eggs?
Q15. If Sally wants to be sure of having a dun foal from her mare what would you suggest she does when choosing a stallion?
Q16. Can you suggest any way that Sally can get a good idea of the genotype of potential dun stallions?

Emma has a grey Arab stallion who’s had three foals. One is chestnut, the other two were born chestnut but are now starting to turn grey. All were born to chestnut mares.

Q17. What is the genotype (at the grey locus) of the mares?
Q18. What is the genotype (at the grey locus) of the stallion?
Q19. If the stallion is bred to a chestnut mare what is the chance that the foal will be grey?

Q20. Champagne is a dominantly inherited trait that dilutes the colour of the coat (i.e. makes it paler). It’s affects on coat colour can be really attractive, for example champagne combined with chestnut can mimic golden palomino. Briefly explain how you think human selection has affected the frequency of the champagne allele in breeds such as the Tennessee Walking Horse, the American Cream Draft and the American Quarter Horse.

Q21. What does line breeding do to the frequency of rare recessive alleles? Briefly state one advantage and one possible disadvantage of using line breeding.

Q22. A tobiano mare and tobiano stallion are mated together. Both themselves had
one tobiano parent and one non tobiano parent. Are they heterozygous or homozygous for the tobiano spotting allele (To¹)?

The non-spotting allele is symbolised To⁺. Illustrate the cross below, using a Punnett square. What kind of cross is it?

What is the chance of the foal being Tobiano?

Q23. What’s the probability (or % chance) of a curly coated foal born of two heterozygous carriers of the recessive curly coat allele? What can you say about the genotype of a curly coated foal from such a cross?

What’s the probability (or % chance) of a curly coated foal born of two heterozygous curly coated horses? What can you say about the genotype of a curly coated foal from such a cross?

Q24. Coat colour in horses is governed by several genes. One of these (which you’ll learn more about later) determines whether the main melanin pigment in the coat is only red, as in chestnuts, or black. Other genes control the modification of these two pigments, as occurs in bays, browns and palominos, for example. Here we are just concerned with the red/black gene which is at the extension locus, with allele symbols e for the red allele and E⁺ for the black allele.

Black horses are of genotype E⁺E⁺ or E⁺e, chestnut horses are of genotype ee. Please ignore all other colour genes when answering these questions:

a. Which allele is the dominant one?
b. Could a black foal have two chestnut parents? Very briefly explain your answer.
c. Could a chestnut foal be born to two black parents? Briefly explain your answer.
d. What colour foals would you expect if several chestnut mares were bred to a black stallion? What could you infer about the stallions genotype by observing these foals?
e. Jerry is thinking of buying a black stallion called Black Magic. He has never sired any foals. He wants to know if the stallion will produce any red foals, or if he is
true-breeding (i.e. homozygous) black. Suggest ways in which Jerry might find out more about Magic's red/black genotype before buying him (and without Magic siring any foals).

Q25. What is the possible outcome of breeding together a black dun mare and stallion, if both are heterozygous for black \( (E^+e) \) and dun \( (D^0D^+) \). (Black dun is a colour pattern also known as grullo). Use a Punnett square if you prefer to (it’s up to you – I won’t mark you down for doing it another way - as long as you clearly show how). Give the phenotype names as well as the numerical ratio.

Answer the following, either as a fraction, e.g. 5/16, or using a percentage.

a. What proportion (or percentage) are expected to be true-breeding for dun?
b. What proportion (or percentage) are expected to be true-breeding for black (with and without dun)?
c. What proportion (or percentage) are expected to be true-breeding for both dun and black at once?

Q26. Which of the following is true about the inheritance of mitochondrial genes in a stallion (assume no inbreeding in its pedigree)?
A. The stallion will pass on these genes to his daughters.
B. The genes were inherited through the maternal grand-dam.
C. Stallions do not inherit mitochondrial genes.
D. The genes were inherited through the paternal grand-dam.
E. The stallion will share the same genes with a half-sister from the same sire.