Rural Economy
& Development
Programme

Genetics and Breeding of Irish Horses

Teagasc
Agriculture and Food Development Authority

Foreword

The equine industry is a growing, vibrant and diverse one which contributes substantially to the overall economy. The annual economic value of the combined sport horse/equestrian, horseracing and breeding industries in Europe is estimated at €52.1 billion per annum. In an Irish context the Irish thoroughbred breeding and racing industries are valued at €1.84 billion while the Irish sport horse sector is estimated to be worth in the region of €816 million to the economy. A significant part of both industries is breeding.

Knowledge about equine genetics lags far behind that for other domestic animals such as bovines. However, each year the demand for such knowledge is increasing significantly which is a positive development. Teagasc has recognised this demand, which has resulted in the production of this book.

This book highlights the importance of equine genetics and breeding for the industry as a whole. It stresses the importance of selectivity and using the best males and females, to genetically improve a population, in a certain direction, based on a predefined goal. It provides an overview of various genetic principles. Animal breeding has the advantage of being cumulative and permanent. This suggests that genetic gain can be progressively improved generation by generation.

Teagasc is one of the main providers of full-time education, lifelong learning and advice to the growing cohorts of equine breeders, producers and riders primarily based in rural Ireland. Recent years have seen an increase in demand for both full-time educational and advisory supports for riders and young breeders in particular. To fill this gap Teagasc have provided and are providing short blended learning courses which are of enormous benefit to breeders.

Some equine breeders have untapped potential. If an optimal breeding approach is implemented, significant genetic advances can be achieved in the coming years. The key to this is knowledge.

I wish to acknowledge the hard work, dedication and commitment which has been exhibited by the author in the production of this book. It is a useful resource which provides key information for breeders, owners and all associated with the equine industry. I also wish to commend the work that has been undertaken by the Teagasc Equine Knowledge Transfer programme for their key role in the sport horse strategy ‘Reaching New Heights’ and in the suite of Knowledge Transfer, Education and Training delivered across the equine sector. I thank them for their dedication and innovative spirit, particularly in the recent development of webinars, distance learning courses and new online information. To quote Kofi Annan: “Knowledge is power. Information is liberating. Education is the premise of progress, in every society”.

G. E. Boyle

Professor Gerry Boyle
Director, Teagasc

Executive Summary

This book has been designed to educate those interested in equine studies. It outlines several aspects of equine genetics and breeding and illustrates how breeders can improve the genetic merit of Irish horses backed by science. The book aims to be a blueprint for equine genetic improvement. The terms commonly used by animal breeders to describe the characteristics of a population are described in an easy to understand format. The book is designed to educate breeders to use available resources to make more informed breeding decisions. This is an essential book for horse breeders and owners, students of equine studies, animal breeding and veterinary science who want to learn more about equine breeding and genetics. References are included throughout to provide greater details about a particular topic. A glossary at the end of the book provides the reader with a summary of definitions used.

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Introduction

Over the last century, the productivity of domesticated animals has increased substantially in line with scientific advances in several areas. Primarily, the introduction of quantitative genetics beginning in the 1930s has greatly enhanced the rate of change (Gianola and Rosa, 2015). Furthermore, improvements in animal nutrition enabled animals to reach their genetic potential, while improvements in reproductive technologies allowed producers to accelerate genetic gain by only selecting genetically elite animals to reproduce. In several species, technological developments and computerised record-keeping facilitated accurate identification of genetically elite animals. Genetic evaluations exploited the resulting data to ensure superior genotypes were selected to suit a particular breeding goal, thereby enhancing genetic gain. In horses, the contribution of breeding to improvements in performance has been much slower in the past decades. When planning coverings, some horse breeders still use theories that rely on hunches, or rule-of-thumb assumptions that usually have no scientific basis even though the science of genetics is over 100 years old.

The equine industry in Ireland is dominated by the Thoroughbred sector and the Sport Horse sector. The Thoroughbred breeding industry produces close to half of all Thoroughbreds in Europe. There are 6,777 breeders, but 92% of these own less than 5 broodmares (Horse Racing Ireland, 2017). Ireland is the third largest producer of Thoroughbred foals worldwide. In 2020, 9,182 Thoroughbred foals were born (Weatherbys Fact Book, 2020). The Irish Thoroughbred breeding and racing industries are valued at €1.84 billion to the economy; breeding accounted for total expenditure of €583 million (Horse Racing Ireland, 2017).

The sport horse sector in Ireland is broad and diverse consisting of many different activities and businesses. At the centre of this industry is the Irish bred sport horse. The definition of a sport horse is ‘a riding horse of a single breed or a combination of breeds used for (or intended for) recreational and competitive activities other than racing’ (Corbally and Fahey, 2017). In 2017, the sport horse population was estimated at 135,715 animals with 20,388 active broodmares; there were 14,830 active breeders. In 2019, the total number of live foals registered was 4,912 (Horse Sport Ireland, 2019). The Irish Sport Horse sector was estimated to be worth €816 million to the economy (Corbally and Fahey, 2017); breeding is the largest sector accounting for total expenditure of €271 million. While some Irish Sport Horses are performing very well on the world stage it is clear that the overall Irish sport horse industry is not realising its full potential. In 2020, the World Breeding Federation for Sport Horses (WBFSH) ranked the Irish Sport Horse studbook in 4st and 14th position for eventing and show-jumping, respectively. The Irish Sport Horse studbook does not, as yet, have a WBFSH ranking for dressage horses. It is apparent that competitor studbooks are overtaking the Irish Sport Horse studbook in the WBFSH rankings. Nonetheless, significant genetic advances can be achieved in the coming years if an optimal breeding approach is implemented. This will require significant acceptance from breeders and industry both in terms of recording data and implementation of available resources.

Essential genetics for breeders

The horse, evolved as a prey animal, who depended on flight as its primary means of survival. As a result, horses were naturally selected for speed and the ability to roam long distances. Horses were domesticated on the Eurasian steppe some 6,000 years ago (Levine, 1999). Since domestication horses have been selected for several different traits but predominately strength, speed and endurance traits. This selection has led to the creation of diverse breeds. The development of specific breeds has resulted in selection for athletic phenotypes that enable the use of the horse for riding, recreation, sport, and racing (Chowdhary, 2013).

At its most fundamental level, genetic selection involves the passing of allele variants (both favourable and unfavourable) from parents to offspring, and unlike management (i.e., nutrition, exercise, and health) genetic selection is cumulative and permanent. This implies that improvements achieved are compounded each successive generation. For example, in poultry, Zuidhof et al. (2014) demonstrated that the kilograms of feed necessary to yield a kilogram of meat (i.e., feed conversation ratio) was predicted to have reduced by 50% and growth rate increased by over 400%, between the years 1960 and 2005. The power of genetic selection was shown by Havenstein et al. (2003) who documented that in chickens approximately 85% to 90% of the improvements in feed efficiency were due to genetics.

It's difficult for management to compensate for bad genetics (and vice versa). Therefore, genetic selection and optimum management when used together, provide the best opportunity for improvement, yielding enormous benefits over time. For example, if a breeder introduces good genetics for traits such as soundness, performance and athleticism they can be improved each successive generation. In direct contrast, however, any indirect consequences of breeding are also compounded every generation. It is imperative that breeding schemes are optimised, where possible to achieve gains in performance where all traits effecting performance are accounted.

Chapter 1. Basic Equine Genetics

One limiting factor for people's understanding of genetics is the language used by experts in the field. An understanding of basic genetic principles can go a long way towards understanding modern approaches to genetic improvement. The basic principles of genetics were established by the Austrian monk Gregor Mendel from his work using garden peas in 1866. In this chapter many basic genetic concepts are explained which lay the foundations for more complex concepts defined later.

DNA

Deoxyribonucleic acid (DNA) is located within the nucleus of a cell. DNA is made of long chains of nucleotides. A nucleotide is the basic building block of nucleic acids consisting of a sugar molecule attached to a phosphate group and a nitrogen-containing base (Figure 1.1).

Molecules of DNA are made up of two strands with cross linkages made up of nitrogen-containing bases. Four different bases make up the cross linkages: adenine (A), thymine (T), guanine (G) and cytosine (C). The linkages form in a particular way where thymine pairs with adenine while guanine pairs with cytosine. The combination is referred to as base pairs. The structure is known as a double helix. As a result, if the sequence of one strand is known then the other strand can be worked out (Figure 1.2).

In multicellular organisms (i.e., plants and animals), the DNA molecule contains all the information necessary to transform a single cell, specifically a fertilised egg, into a complex, multicellular individual. Scientists were initially surprised that a molecule with only four basic units could deliver sufficient information. However, the information was found to be contained in the precise order of bases along the molecule.

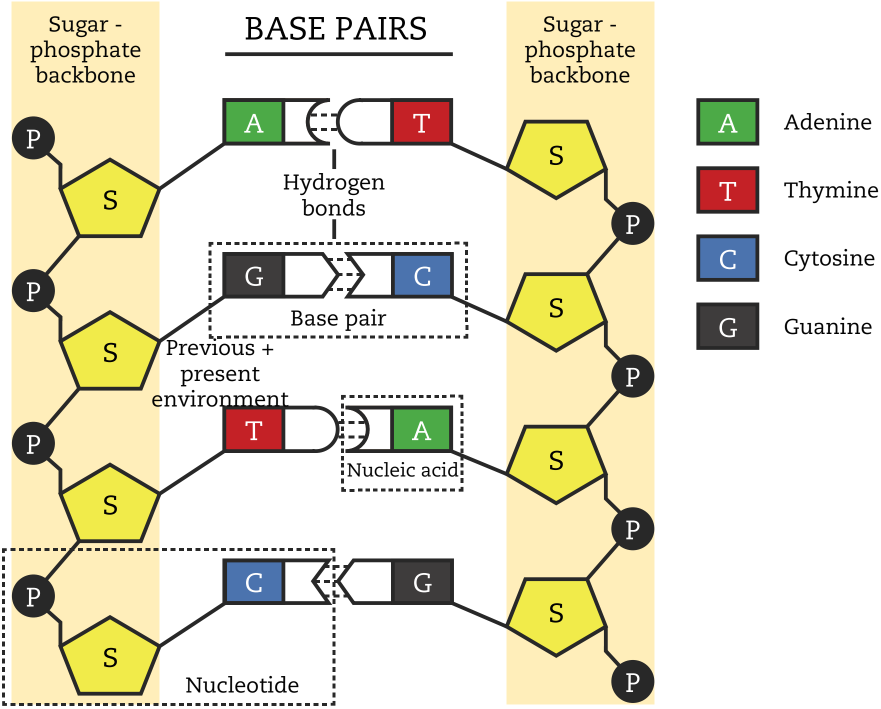


Figure 1.1. DNA structure. *National Human Genome Research Institute*

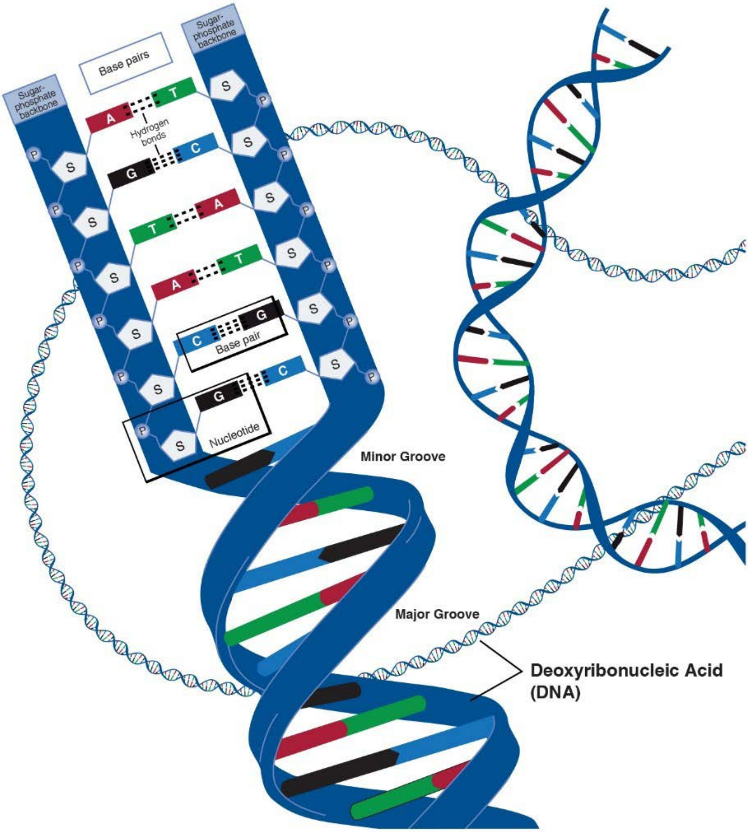


Figure 1.2. DNA structure. *National Human Genome Research Institute*

Genes

A gene is a sequence of DNA which contains the instructions to make a protein or other gene products. Genes provide instructions as to, for example, what the animal will look like and how it will perform. Genes are strung together in long strands of DNA called chromosomes. All animals have multiple pairs of chromosomes (one from each parent) located in the nucleus of their cells. A healthy horse has 32 pairs of chromosomes (this is known as the diploid number of chromosomes) including two sex chromosomes, XX in females and XY in males. Humans have 23 pairs of chromosomes.

The non-sex chromosomes (31 pairs) of a healthy horse are known as autosomes. Of the 64 chromosomes found in each cell of a healthy horse, 32 come from the sire and 32 come from the dam (Bailey and Brooks, 2020). The complete set of chromosomes is known as a karyotype. One chromosome can contain hundreds and even thousands of genes. It is the genes, in combination with the environment (e.g., nutrition and management) that determines the performance of the horse such as jumping ability or disease susceptibility. All genes are composed of DNA, and it is the variation within the DNA that makes horses different.

Sex chromosome abnormalities

In horses, reproductive failure can be caused by several factors. One factor commonly overlooked is abnormality of chromosome or karyotype. Mares that exhibit infertility, reduced cyclicality or small ovaries may have an abnormal chromosome complement known as equine Turner syndrome or XO monosomy. This occurs where a mare is missing one X chromosome (XO instead of the normal XX); these mares have a karyotype of 63, XO instead of the normal complement of 64, XX (Figure 1.3). In certain cases mares with equine Turner syndrome can exhibit symptoms such as reduced size, angular limb deformity and poor conformation (Samper et al., 2006).

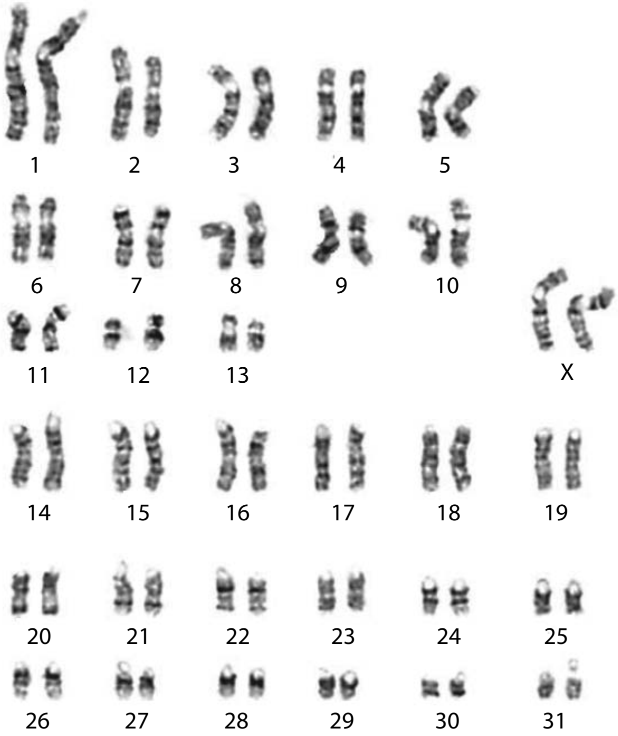


Figure 1.3. A karyotype of healthy mare containing 32 pairs of chromosomes including two sex chromosomes (64, XX). Samper et al. (2006).

Some infertile mares can have an abnormality called XY sex-reversal syndrome. In this case, the mare has a female phenotype and male karyotype 64, XY. In this particular abnormality the mare can have normal genitalia but no estrous cycles and may also exhibit stallion like behaviour.

In some mares, a few cases of X chromosome duplication, or trisomy, have also occurred. In these cases the mares has a karyotype of 65, XXX and appear phenotypically normal but are infertile (Samper et al., 2006).

Locus and alleles

Genes or nucleotides are located at a physical position on the chromosomes called the locus (the plural is loci). The locus is in the same position on the chromosome for all animals of a species; in some instances however, this position can be altered due to deletions (removal of DNA), duplications (repeated sequence of DNA), inversions (sections of the chromosome rotated within a chromosome) and translocations (sections swapped from one chromosome to another).

An allele is a variant of a locus (or region) from changes caused by mutations. There is one allele on each chromosome, but they may have changes in sequence. Each allele present in the foal should be present in at least one of the parents; however, in some cases this is not the case due to mutations and/or deletions. Alleles found on the locus can be either homozygous “AA, aa” (same) or heterozygous “Aa” (different). Some horses with the best performance (phenotypes) do not always make the best breeding stock.

Alleles found on a locus can be either dominant, usually donated by capital letters or recessive, usually donated by small letters when written in a Punnett square. Dominant alleles mean that only one copy is needed for it to be expressed. For example, it only takes one dominant allele to express grey horse colour, it will always be expressed with only a few exceptions. Recessive means that an allele will always be covered up by a dominant allele if present. As a result, two copies of the allele are needed for the trait to be expressed in most cases. An example is the red allele of the Extension gene. Chestnut horses are homozygous for the recessive allele – see Chapter 4 for further information.

Genotype

Genotype refers to all the information contained within the animal’s DNA. This DNA contains a huge volume of information, 2.7 billion base pairs or building blocks separated into approximately 15,000 separate genes. In general, each gene is inherited 50% from the sire and 50% from the dam at the point of fertilisation and is fixed from that point with the same DNA code in every cell. However, these percentages can change where there are changes in autosomes and sex chromosomes. In addition, the dam contributes mitochondrial DNA which is passed from the dam directly to the offspring (Chapter 2).

Each horse is born with a fixed genotype which remains the same throughout their life. For example, an observed phenotype (physical expression of a gene) for coat colour can be black or white, while its genotype for coat colour may be either AA, Aa or aa. Although genotypes AA and Aa both correspond to a black coat colour and only aa results in a white coat colour, the animal's phenotype that produces a black coat colour can be derived from one of two gene variants.

Phenotype

The phenotype is the observed physical traits of an animal "in the field" (e.g., coat colour, height etc.); it's the physical expression of the animal's genotype. The variation in a population's phenotypic performance is dependent on two components, genetics and environment (and their interaction). The phenotype is influenced by many factors in the animal's environment. Differences due to genetics in its simplest form constitute differences between breeds while environmental variance constitute differences in nutritional intake, herd management and biological factors such as age and gender. For example, two horses with identical genotypes could develop dissimilar phenotypes if raised in contrasting environments and exposed to different nutrition and training. Figure 1.4 gives an overview of an animal's lifespan highlighting important events that indicate changes in environmental influence. Despite the best efforts of geneticists, genetically perfect animals can still have poor performance as several environmental factors can impede performance such as training, diet and the experience of the jockey.

$$\text{Phenotype} = \text{genotype} + \text{environment} + \text{genotype}^{*}\text{environment}$$

$$+ \text{covar (genotype;environment)}$$

where **genotype** refers to the information contained within the DNA of the horse; **environment** refers to the variation in the physical environment in which the horse is exposed (e.g., training, nutrition etc.); **genotype*environment** is the interaction between the genotype and the environment. This interaction occurs where the difference in performance between two genotypes depends on the environment in which performance is measured. A simple example is where a stallion has progeny in Ireland and other European countries. The environments in these countries differ (e.g., nutrition and training). Therefore, the progeny from one stallion could perform better in one environment than the other. As a result, the environment in which the animal is raised can effect performance and must be evaluated. **Covar (genotype; environment)** is the covariance between genotype and the environment. For example, horses from an exceptional family might be treated better than their contemporaries; this is known as preferential treatment. Good trainers might not train horses with mediocre expected genetic merit. Another example is where parents with a high IQ provide an IQ stimulating environment for their children (Visscher et al., 2008).

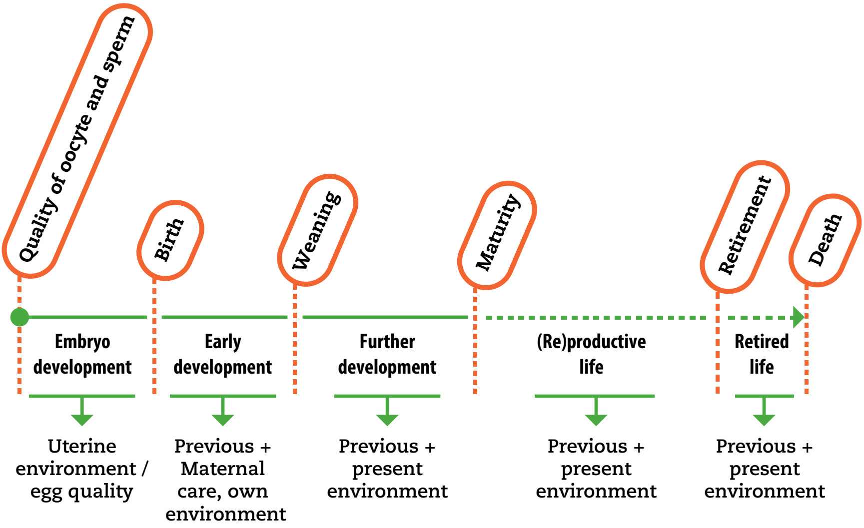


Figure 1.4. An overview of animal's lifespan highlighting important events that indicate changes in environmental influence. A horse is influenced by its current environment but also its past environments as emphasised in the boxes underneath. The mare determines part of the environment until weaning. Oldenbroek and van der Waaij (2015).

Genetic variation

Genetic variation is a measure of the variation or differences within a population that are due to the differences in genetic merit of the animals. Genetic variation occurs both within and among populations. Without genetic variation, genetic improvement is not possible. Recently, McGivney et al. (2019) found that in the last five decades there has been a highly significant decline in global genetic diversity in the Thoroughbred. The results of this study indicate that market-driven emphasis on highly valuable pedigrees and the common practice of inbreeding to successful ancestors in attempts to reinforce favourable genes in offspring has resulted in a global reduction in genetic diversity.

Breed formation

The process of breed formation is a process of reducing genetic diversity in a population. Breeders want to increase alleles that contribute to the desired phenotype and eliminate alleles that adversely affect the horse. Genetic variability within a population is a function of 5 evolutionary forces which are genetic drift, migration (also called gene flow), mutation, recombination, and selection. Two processes predominate: genetic drift and selection.

Genetic drift

The amount of variation within a breed is determined by the number and variety of horses that make up that breed. As long as the studbook maintains an open book, new gene variants can be introduced into the population with new horses. Once the studbook is closed (e.g., Thoroughbred studbook), the sum total of genes is determined by the founders of the breed. Mutations can still create new genetic variants in a closed population.

Events unrelated to selection can have an impact on the presence of an allele. One of these is called genetic drift. Alleles that are rare in the population are at great risk of being lost. If a genetic variant occurs in only one individual, or in only a few individuals, and by chance that allele is not passed on to the next generation, then the allele will no longer exist in that breed. Therefore, genetic diversity will decrease. Conservation biologists are concerned about loss of genetic diversity because some genes may provide the capacity to resist disease or increase health and fitness. The value of each and every gene cannot be known, therefore breeders are encouraged to value genetic diversity in breeding programmes.

Selection

Selection causes changes in allele frequencies as a result of choices made by breeders to use particular animals as breeding stock. These choices should lead to the gain of alleles that make the horse more desirable and the loss of alleles that do not improve the phenotype. Until recently, scientists have not been able to identify any performance genes at the molecular level, so direct selection for genes has not occurred. Consequently, scientists can only speculate that differences in gene frequency for certain genes are related to performance. In the coming years, there should be direct evidence about the role of genes and performance for certain traits. For example, Hill et al. (2020) found that the myostatin gene has a direct association with sprinting ability and stamina in Thoroughbred horses. The myostatin gene can control the rate of development of muscle and the type of muscle that is formed (fast-twitch or slow-twitch). Studies have shown that there are three types of speed gene known as C:C. C:T and T:T – see Chapter 6 for further information. It is likely that selection for speed has increased the sprinting allele for Flat Horses, while selection for stamina has led to the endurance allele being most common among National Hunt horses.

Chapter 2. Statistical and Genetic Terminology

Statistical Terminology

Mean

Also known as the average, the sample mean is calculated as the sum of all values in a dataset divided by the total number of records in that dataset.

Standard deviation

Knowing the standard deviation allows inferences to be made on datasets that are normally distributed. Assuming a normally distributed dataset the empirical rule states that a) approximately 68% of data lies ± 1 standard deviation from the mean, b) approximately 95% of data lies ± 2 standard deviations from the mean, and c) approximately 99.7% of data lies ± 3 standard deviations from the mean.

Coefficient of Variation

The coefficient of variation expresses the standard deviation relative to the mean and is expressed as a percentage. Unlike the mean and standard deviation (which are expressed in the units of the trait) the coefficient of variation is independent of the unit of measurement; therefore, comparison between traits with different units can be made.

Correlation

A correlation measures the strength of a linear relationship between two variables x and y. Correlations range from -1 to +1, where -1 indicates that when variable x is high, variable y is always low (or vice versa), 0 indicates no linear relationship between variable x and y, and +1 indicates that when variable x is high variable y is also high (and vice versa). Figure 2.1 illustrates different types of correlations.

Genetic correlations measure the strength of the linear relationship between two traits that is due to genetics (Berry et al., 2011). A negative genetic correlation indicates that when variable x is high, variable y is low (and vice versa) while a positive genetic correlation indicates that when variable x is high variable y is also high (and vice versa). Genetic correlations occur due to pleiotropy or linkage among genes. Genetic correlations are unit-less ranging from -1 (strong negative relationship) to 0 (no relationship) to +1 (strong positive relationship between two variables) (Falconer, 1996).

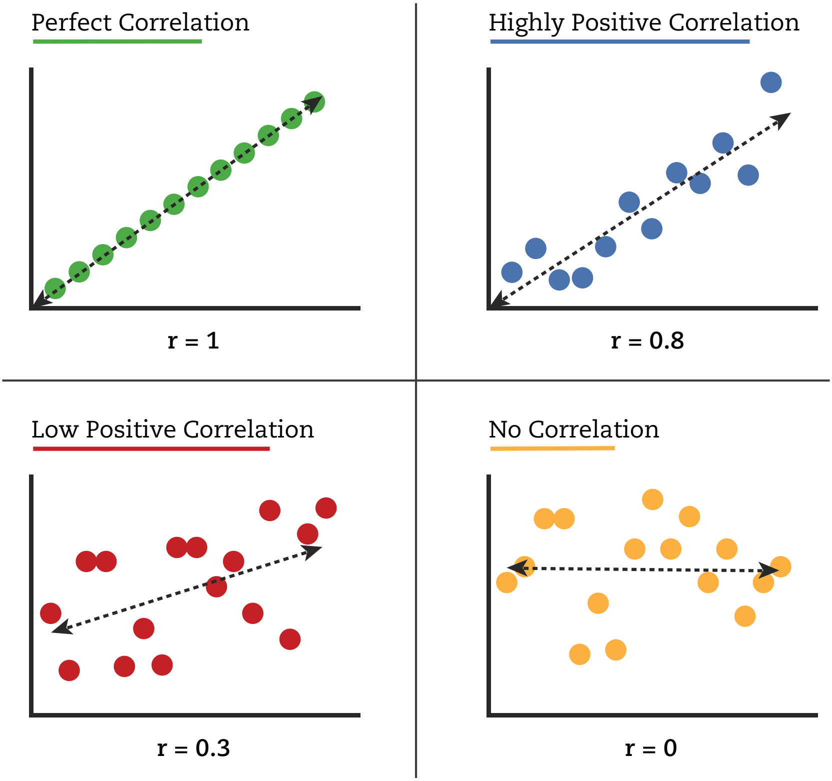


Figure 2.1. Examples of correlations.

Genetic Terminology

Heritability

Heritability (h^2) summarises the proportion of phenotypic variation, or differences among a cohort of animals, attributable to genetic variation between individuals (Berry et al., 2011). Put simply, how much of the observed performance (phenotype) independent of management is due to genetics. Heritability is estimated for a specific population in a specific environment, thus, it reflects the genetic variation for a trait in that particular population (Oldenbroek and van der Waaij, 2015). The specific alleles that contribute to a phenotype may vary greatly in different environments. The particular alleles that a population possess results from their origin and prior selection. Most traits cannot be explained by few genes but influenced most likely by thousands of genes.

Heritability ranges from 0 (not heritable) to 1 (fully heritable), where 0 infers that none of the observed performance in a population is due to additive genetic variation while 1 indicates that all of the observed performance in a population is due to additive genetic variation. If heritability is high, it is expected that a large proportion of the phenotypic differences of the parents will be passed on to the progeny. Human height for example is a highly heritable trait (approximately 80% heritable). Many characteristics such as athletic ability, coat colour and behavioural tendencies are all under genetic control. Heritability values are influenced by factors such as the accuracy of controlling environmental effects, gender and age.

$$h^2 = \frac{\text{additive genetic variation}}{\text{phenotypic variation}}$$

Reliability

Reliability is a measure of accuracy or degree of confidence in the published genetic merit of an animal being a true reflection of the genetic merit of that animal. The genetic evaluation process, using sophisticated statistical techniques, provides an estimate of an animal's genetic merit based on available data – see Chapter 5 for further information.

Reliability ranges from 0 to 99% and is important when evaluating an animal's genetic index. For example, a sire who has a lot of data backing up his genetic evaluation is more 'reliable' (i.e., >90%); therefore, the extent to which his genetic evaluation changes in the future is reduced. Conversely, a sire that has less data backing up his genetic evaluation is less 'reliable' (i.e., 30-50%); therefore, the extent to which his genetic evaluation changes in the future is increased. Genetic evaluation changes can be either upwards or downwards. Consequently, the more information available on an animal's pedigree, progeny and performance results in a higher reliability.

Generation Interval

The generation interval is the average age of the parents when their offspring are born or the average length of time between the birth of an animal and the birth of its replacement offspring (Thiruvenkadan et al., 2009). This can be different for males and females. The shorter the generation interval, everything else being equal, the faster the rate of genetic progress per year resulting from selection. Horses have a long generation interval varying from 9 to 12 years. This contrasts with other domestic animals whose values are approximately half that or less (Table 1.2).

Thoroughbreds can have a long generation interval as many Thoroughbreds begin their breeding only after completing their career in racing; excellent racehorses might race for a long time. In contract, reproductive technologies (e.g., artificial insemination (AI), embryo transfer (ET) or intracytoplasmic sperm injection (ICSI)) are accepted for breeding sport horses. Therefore, using the correct scientific breeding approach faster genetic progress could be achieved due to a reduction in the generation interval. Assuming genetic gain is occurring the youngest generation are genetically the best horses.

Table 1.2. Generation interval of domestic animals.

Species	Generation Interval
Horses	9 to 12 years
Dairy cattle	4 to 6 years
Beef cattle	4 to 6 years
Pigs	1.5 to 2 years
Chickens	1 to 1.5 years
Sheep	3 to 5 years

Genomics

When evaluating a pedigree breeders are trying to identify key traits that the horse has potentially inherited from its ancestors. Today, it is possible to assess the exact DNA make-up of a horse, as opposed to the inferred genetic make-up that is assumed from pedigree. Although pedigree offers a wealth of information there are occasions where pedigree can be misleading. This is where an evaluation of DNA can unlock some of the puzzle.

Genomics is the study of an animal's DNA which are the building blocks of genes. The horse genome was sequenced in 2007 with the final report published in 2009 (Wade et al., 2009). As a result, the 3 billion units of genetic code that spell out the instructions to make an individual horse are now known. This has allowed researchers to understand how differences in a horse's DNA make that individual horse unique. It is now possible to take a blood, hair or tissue sample from a new-born foal to determine its complete DNA profile and this can be used to predict the subsequent performance of that individual. For example, mature height and muscle mass can be difficult to estimate in a young foal but genetic testing of the same foal can give an insight into the physical makeup as a mature horse. In addition, because DNA is transmitted from one generation to the next, the DNA of a foal (e.g., potential stallion) could also be used to predict the expected performance of its progeny.

Breeders need to remember that genomic predictions for performance are still only predictions. If a genetically elite mare is not managed appropriately then her genetic potential will not be realised. This is why genomic predictions are not (and never will be) 100% accurate in predicting performance. Nonetheless, over the last decade rapid technical advances in molecular genetics and bioinformatics have enabled the integration of genomic information in increasing numbers of routine applications in animal breeding.

Genomic technology is currently used by some studbooks to, for example, screen for Osteochondrosis, lethal genes as well as confirm parentage and identify and correct errors. Most of the traits (e.g., athletic performance, health etc.) selected in performance horses are complex involving numerous genes. As a result, using genomics to select horses for these desirable traits is more difficult but still achievable.

Figure 2.2. Two different horses each with a different strand of DNA. Both horses have different combinations of nucleotides (i.e., A, T, C and G) in the DNA sequence. The single difference between both animals is called a single nucleotide polymorphism (highlighted by the red box).

Single Nucleotide Polymorphisms

Genotyping is the process of determining the genetic constitution (i.e., the genotype) of an individual by examining its DNA sequence; it enables the underlying genetic variants in DNA to be explored. Different genetic variants exist including single nucleotide polymorphisms (SNPs) which refer to a sequence change caused by a single base-pair mutation at a specific locus in the DNA sequence (Figure 2.2). Genetic background analysis is commonly done using either microsatellite or SNP analysis. Single nucleotide polymorphism genotyping is currently a popular genomic technology as SNPs are abundant and widely distributed across the genome. However, depending on the application both methods of genome scanning have their advantages and disadvantages.

Y chromosome and mitochondrial DNA

According to Gregor Mendel, genes are inherited from the sire and dam and may behave with a dominant, codominant or recessive modes of inheritance. In the cases of the sex chromosomes and mitochondria each has its own DNA and follow a unique pattern of inheritance.

Only found in males the Y chromosomes is the male determining piece of DNA. In the absence of the Y chromosome, embryos develop female characteristics. The Y chromosome follows the ‘tail-male line of a pedigree (Chapter 3). As a result, the Y chromosome is used as a measure of a stallion’s influence, over many generations, on a population. As an illustration, over 95% of modern-day Thoroughbreds trace their ancestry to the Darley Arabian through the male side of the pedigree. Cunningham et al. (2001) determined that 158 founders contributed 81% of the DNA to the modern-day Thoroughbred. The Y chromosome has limited genetic variation.

Breeders might often wonder why some mares have a special place of influence on a breed. Quite often these mares have excellent performance records. This suggests they are already an elite group. Nonetheless, some of these mares are only recorded as broodmares with no performance records. Their daughters however were covered with good sires, and the line traces down the generations to produce good horses. A genetic explanation for the continuing influence of a specific ‘foundation’ mare within a pedigree could be due to the role of the mitochondria and mitochondrial DNA. Mitochondria are found in nearly all plants and animals. They are small structures that carry out important functions of energy creation for cells and commonly referred to as the “powerhouse of the cell” for this reason. Mitochondria have their own DNA encoding for 37 genes (Bailey and Brooks, 2020).

Mitochondrial DNA is passed from the dam directly to the offspring (Lande and Kirkpatrick, 1990). Unlike the Y chromosome, the mitochondrial DNA has been found to contain lots of genetic variation in horses (Achilli et al., 2012). When an egg is fertilised, the sperm contributes its chromosomal DNA only, and minimal mitochondrial DNA. Mitochondrial DNA is maternal regardless of the sex of the embryo. Therefore mitochondrial DNA is a measure of the influence of female bloodlines. The mitochondrial DNA is passed down generation after generation through the tail-female line (Chapter 3). This could be the link between the 'foundation' mare with the foals in her line. The excellent performance of some horses sharing tail-female connections could be based on maternal inheritance through mitochondria. However, any such relationship between performance traits and specific mitochondrial genes currently awaits validation.

Epigenetics

Epigenetic changes occur where there are modifications to gene expression without changes to the DNA sequence; therefore, external environmental effects can influence the expression of the phenotype. The genetic sequence (DNA) of an individual is fixed from birth. However, expression of the genetic information is not fixed from birth as epigenetic control is dynamic and responsive to external inputs. Research suggests that the success of the offspring may actually be determined well before the mare is scanned in-foal as the gametes can be influenced by external environmental influences on the sire and dam before fertilisation. Such environmental influences include nutrition and training, environmental chemicals, drugs/pharmaceuticals and aging. The genetics that the offspring possess do not always determine success. For example, bee larvae start with the exact same genetics and depending on the selected diet (i.e., royal jelly or worker jelly) as larvae they either become a queen or worker bee. This is due to the way due to the way genes are switched on or off in response to the specific diet (Wojciechowski et al., 2018). The queen bee lives a completely different life to the worker bee where she can reproduce and live for >2 years. In contrast, the worker bee is unable to reproduce with no active ovaries and only lives for 3-6 weeks.

In horses, the periods where this can occur is the periconception period (i.e., before the male and female gametes meet) and the point of fertilisation and during pregnancy. Most of the research to date has focused on the gestation period. Rossdale and Ousey (2002) proposed that an adverse foetal environment could potentially limit future health and athletic performance of the horse.

Epigenetics and the recipient mare

If breeders are considering using artificial methods of reproduction such as ET or ICSI they should carefully consider their choice of recipient mare. Breeders should ensure the recipient mare is the closest match to the donor mare in terms of type, breed and height as possible; this is based on research evidence that shows uterine size, withers height and body condition score can affect foal growth and metabolism (Allen, 2005, Peugnet et al., 2014). This is to ensure the foal gets the most appropriate environment during pregnancy that it was supposed to get from the donor mare had she carried the pregnancy.

Genomic imprinting

The contribution of the stallion and mare to the offspring can be hotly debated among breeders. Genomic imprinting occurs where the expression of a gene is dependent on the parent of origin. During egg and sperm development, certain genes are activated while others are deactivated. The epigenetic mechanisms of DNA methylation and modifications of chromatin regulates the expression of the parent of origin (Surani et al., 1986). Only a small percentage of genes, less than 2% of the genome, is thought to exhibit this genomic imprinting.

It has been suggested that genomic imprinting is responsible for the maternal granddam effect. For example, certain Thoroughbred stallions are poor producers of racehorses, however through their mare offspring they become excellent grandsires. The explanation for this is unclear but it has been suggested that these stallions have genes that are superior for racing performance, but are only expressed when they are transmitted by a female (Bailey and Brooks, 2020).

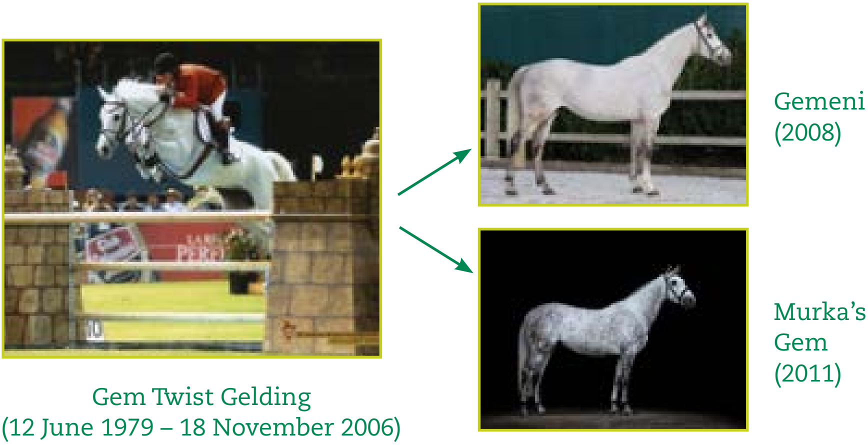
Cloning

Cloning, also known as somatic cell nuclear transfer, results in an organism that is genetically identical to the individual from which it was derived. Somatic cell nuclear transfer is a technique where scientists remove the nucleus from a healthy unfertilised egg and replace it with the nucleus from a somatic cell (any cell other than a reproductive cell as these do not have the required number of chromosomes) from the animal being cloned. The resulting embryo is implanted into a surrogate. The surrogate then gives birth to a clone of the animal that donated the nucleus. The first cloned horse foal was born in 2003 using adult somatic cells (Galli et al., 2003).

Cloned offspring will never be identical to the original individual. This is due to the effect of the environment on the expression of genetic merit. Some traits are more under genetic control than others. Lowly heritable traits will be influenced more by variation in their environment (e.g., how the clone is raised and managed compared to the original) than their genetic merit.

Several problems can sometimes affect cloned foals. For example, foals can have contracted tendons in the front legs, enlarged umbilical cord and weakness or maladjustment. Nonetheless, defects in the cloned foal tend to be erased when the clone reproduces.

Cloning in sport horses is not a tool of genetic improvement as offspring are a genetic copy of the donor animal. Figure 2.3 shows an example of a successfully cloned horse Gem Twist and his clones Gemeni and Murka's Gem.



Gem Twist Gelding
(12 June 1979 – 18 November 2006)

Gemeni
(2008)

Murka's
Gem
(2011)

Figure 2.3. An example of a successfully cloned horse Gem Twist and his clones Gemeni and Murka's Gem.

Heterosis

Heterosis, or hybrid vigour, is defined as the extra performance above mid-parent mean. This means that the performance of a crossbred animal can exceed that of both parents (Figure 2.4). However, for this to happen the heterosis effect must be greater than the difference between the mean and the better of the two parent breeds. Heterosis is an artefact of increased genomic heterozygosity, exploiting dominance variance at a locus but also reducing the probability that a locus is homozygous for recessive deleterious alleles affecting performance.

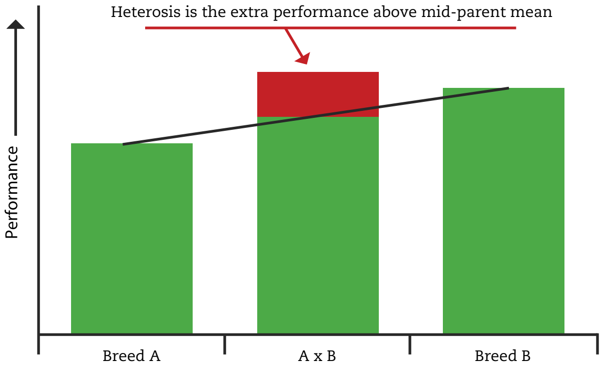


Figure 2.4. The performance of a crossbred animal will exceed that of both parents.

Chapter 3. Pedigree and Breeding Schemes

Pedigree

The conventional pedigree format is illustrated in Figure 3.1. The offspring is named on the left. The first column to the right lists the parents arranged with the sire on the top and the dam on the bottom. The next column gives the parents of the parents (i.e., grandparents of the offspring). Although the offspring resemble their parents to a varying degree, the genetic contribution of each parent remains similar: 50% of the offspring's DNA come from the father and 50% from the mother. However, these percentages can differ where there are changes in autosomes and sex chromosomes as well as the mitochondrial DNA contributed by the dam (Chapter 1 and 2); for simplicity we can assume 50%. Parent-offspring relationships have a relatedness coefficient of 50% or 0.5; grandparents-offspring relationships have a relatedness coefficient of 25% or 0.25 (Table 3.1). These relatedness coefficients are not exact proportions as the gene proportion could be larger or smaller than the calculated relatedness coefficient. Nonetheless, these coefficients are the most probable percentages.

For every locus, the offspring have four possible allelic combinations (assuming the parents are heterozygous for different alleles): 25% of the time the offspring will receive the same alleles from their dam; 25% of the time the offspring will receive the same alleles from their sire; 25% of the time the offspring will receive no alleles in common from sire and dam; and 25% of the time the offspring will receive the same alleles from both sire and dam. Geneticists are unable to predict the exact proportion of genes that any two full siblings have in common due to the random assortment of chromosome pairs during gamete formation. As a result, geneticists provide an average value for all genes of full siblings as a group (Bailey and Brooks, 2013). Therefore, one cannot assume that a full brother to a proven stallion will be an equivalently successful sire. For example, a full brother to a proven Group 1 winning Thoroughbred stallion will have a higher likelihood of being successful given his genetic relationship with the proven stallion compared to an unrelated stallion. However, there is no assurance that the full brother has obtained the group of genes that sets his brother apart from the rest of the breed.

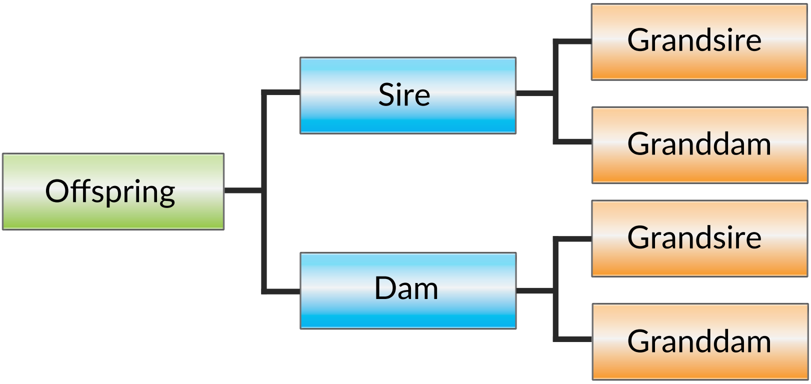


Figure 3.1. The conventional pedigree format.

Table 3.1. Percentage influence of individuals within a pedigree, by generation position.

Generation	Relationship	DNA	Total ancestors
	Colt/Filly	100%	1
1	Parents	50%	2
2	Grandparents	25%	4
3	Great-grandparents	12.5%	8
4	GG-grandparents	6.25%	16
5	GGG-grandparents	3.125%	32
6	GGGG-grandparents	1.56%	64
7	GGGGG-grandparents	0.78%	128
8	GGGGGG-grandparents	0.39%	256

Pedigree generation influence

Inexperienced breeders maybe intimidated by a four- or five generational pedigrees. Therefore some breeders reduce confusion by forgetting about horses after a certain generation (usually the fourth generation) (Bailey and Brooks, 2013). The genome of any horse is always a composite of contributions from the entire pedigree. On average, gene percentages of ancestors in distant generations will be small, however if a sire was exceptional then he can have a greater influence on the modern stock. This results in a higher level of inbreeding and is a big issue in the Thoroughbred.

Figure 3.2 illustrates an example where coat colour (G; grey) can be traced through a pedigree. The grey horse in the fourth generation on the dam's side (indicated 'G') cannot be the source of BOUNCER'S grey colour. The grey ancestor in the fourth generation on the sire's side can be traced from one generation to the next and is clearly the source of BOUNCER'S grey colour. The G on the sire's side had a 50% chance of being lost from the pedigree with each generation but was passed forward in each case.

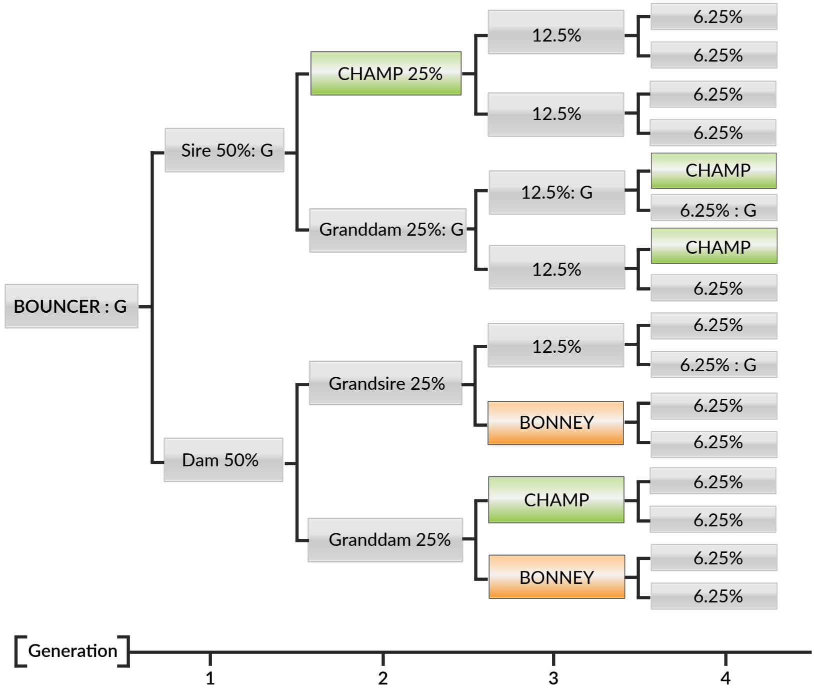


Figure 3.2. For a hypothetical case, consider the calculation of a pedigree for BOUNCER. BOUNCER'S relatedness coefficient to BONNEY is 25%. Gray coloured horses are classified with G.

Tail-male and tail-female lineages

Tail-male lineage

Tail-male pedigree lineage is referring to the 'top line' of a conventional pedigree format (i.e., sire, grandsire, great-grandsire etc.). Tail-male pedigree lineage are classified by breeders as those that trace to a notable founder stallion. As illustrated in Figure 3.3 BOUNCER, JACK, HERO and JAKE share tail-male lineage and a Y chromosome from the 'superior' stallion CHAMP. Clustering male horses based on their shared tail-male relationship makes biological sense in the light of transmission of the Y chromosome from male to male (Chapter 2). These male horses share the genes present on the Y chromosome (except for the effects of recombination and mutation).

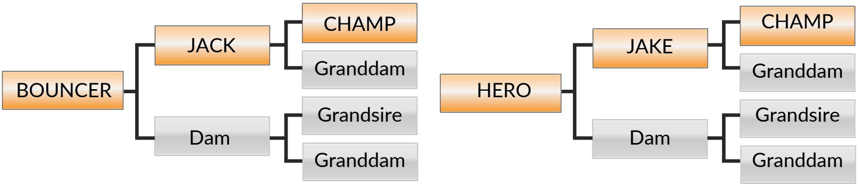


Figure 3.3. BOUNCER, JACK, HERO and JAKE share tail-male lineage and a Y chromosome from the stallion CHAMP.

Tail-female lineage

Tail-female pedigree lineage is referring to the ‘bottom line’ of a conventional pedigree format (i.e., dam, granddam, great-granddam etc.). Breeders use the word ‘family’ to mean several things but are generally referring to horses that are selectively bred for several generations from a subset of animals within a breed. Frequently, this reflects the long-standing successful breeding programme of a particular stud farm (Bailey and Brooks, 2020). However, some breeders associate many traits to tail-female pedigree connections. See Figure 3.3, the ‘foundation mare’ is BONNEY. Many breeders strive for multiple generations of breeding within the same line (sires and dams of the same line). However, any underlying biological basis for attaining excellence in this way currently awaits validation (Bailey and Brooks, 2020).

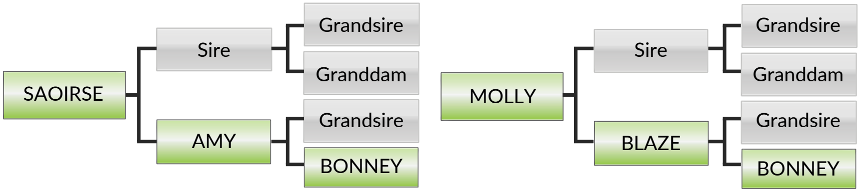


Figure 3.4. SAOIRSE, AMY, MOLLY and BLAZE share tail-female lineage to the super mare BONNEY.

Inbreeding

Many horse breeds have small closed populations with high levels of inbreeding and homozygosity which require appropriate breeding management. Inbreeding occurs from the mating of two genetically related individuals (e.g., father and daughter, mother and son, brother and sister or cousins). The inbreeding coefficient, *F*, defines the probability that two alleles at any locus of an individual are identical (homozygoes) by descent (Malécot, 1948).
Some examples of expected *F* values are:

- Parent and Offspring = 0.25
- Uncle and Niece = 0.125
- First cousins = 0.0625
- Second cousins 0.016

See Figure 3.2; BOUNCER and his sire are inbred to CHAMP. As an illustration, BOUNCER F value is 0.0825. As a result, approximately 8% of BOUNCERS genes would be expected to be identical to those from CHAMP. BOUNCER'S dam is expected to be inbred to the mare BONNEY, but CHAMP is not expected to be inbred to her, because she does not occur on both sides of his pedigree. Inbreeding increases the proportion of genes that trace to a given ancestor. In addition, the proportion of genes that are homozygous is increased. However, in certain cases the desirable genes in the ancestor may not have been homozygous in the ancestor. Therefore, accurate genetic tests are required to identify carriers of undesirable traits when working with inbred pedigrees (Bailey and Brooks, 2020).

Inbreeding has intensified in line with the progress in reproductive technologies, such as ET and ICSI (i.e., with sport horses), both of which can result in using fewer parents to provide the next generation of breeding animals. In addition, there has been an increase in the number of foals sired by popular stallions. These trends decrease the effective population size (i.e., the number of genetically distinct individuals) and, over time, will result in increased inbreeding. For example, the Thoroughbred has been shown to be the most inbred of all the horse breeds (Bowling, 1996). It was estimated that 78% of the genetic variation present in the modern Thoroughbred population was contributed by as few as 30 of the founder animals; the average inbreeding co-efficient based on pedigrees was 12.5% (Cunningham et al., 2001). A slower rate of inbreeding is not (normally) as bad as faster rates. Increased inbreeding has been connected with negative effects on fitness traits in many populations. Reproductive depression is one common sign that a population is compromised by high levels of inbreeding (Binns et al., 2012).

Within purebred breeding programmes, extreme genetic uniformity is usually undesirable as reproductive soundness, hardiness and vigour will reduce with increasing homozygosity (Bailey and Brooks, 2013). Within a large (breed) group, genetic diversity can be maintained by establishing several inbred lines. Breeders might be surprised to find how few founders their purebred animals trace from if distant generations could be viewed as easily as more recent ones.

Some closed studbooks, such as the Thoroughbred have created a closed gene pool. This has resulted as their requirements prevent the use of individuals from outside the herdbook registry. The objective of a closed studbooks is to produce a consistent type of stock with desirable traits specific to that breed. The gene pool of these populations could be genetically diverse if a large founder population exists with no historic periods of restriction (i.e., bottlenecks) (Bailey and Brooks, 2020). In the global Thoroughbred population, McGivney et al. (2020) reported that the significant increase in the levels of inbreeding during the last five decades are unlikely to be stopped due to current Thoroughbred breeding practices. Increasing genetic diversity within closed studbooks is challenging. The introduction of genetically diverse individuals would help increase genetic diversity in these populations. New gene combinations can be produced through mutations or undetected crossbreeding.

Line-breeding

Breeders use the term line-breeding to describe their breeding programmes based on several pedigree crosses to one or more selected ancestors. The same ancestor usually occurs in the pedigree of both the sire and dam. Hence, it is believed that breeding with the same ancestor on the top and bottom of the pedigree will condense bloodlines to bring out desirable traits of the ancestor in the resulting offspring. All line-breeding is a form of inbreeding and reduced genetic variance. See Figure 3.2; BOUNCER is line-bred to the “exceptional sire” CHAMP.

Crossbreeding

In direct contrast to inbreeding and line-breeding, crossbreeding (often termed outcrossing) is predominately implemented for two reasons. First, the introduction of favourable genes and second, to exploit heterosis or hybrid vigour (Chapter 2). Outcrossing is defined as the breeding together of unrelated individuals. In closed studbooks, such as the Thoroughbred this would mean avoiding or minimising the duplication of names in pedigrees However, it is still likely that animals are related in distant generations. In sport horses, the KWPN studbook for example, utilise outcrossing in combination with performance testing to identify show-jumpers and dressage horses. Their breeding stock are selected for a subset of traits associated with excellence in these disciplines.

Assortative mating

This is a form of selection involves mating like with like individuals. Without regard for pedigree, the perceived phenotypic similarities of both individuals is called positive assortative mating. A racehorse breeder would usually select positive assortative mating for speed (e.g., mating a sprinter to a sprinter). However, a breeder could also incorporate negative assortative matings to improve traits such as conformation (e.g., light-boned animals mated to heavier boned animals) (Bailey and Brooks, 2013).

Mating horses opposite in phenotype is known as negative assortative mating or corrective mating. For example, breeding a mare who is toed in to a stallion who is toed out. However, different genetic mechanisms could be causing both conditions. As a consequence, it will be difficult to predict the offspring's leg structure. Therefore, the best strategy would be to breed the mare to a stallion with the most correct structure that fits into the breeder's economic restraints and other selection criteria.

Chapter 4. Genetics of Coat Colour

Breeders can easily recognise most coat colours, however, coat colour genetics can cause confusion in the equine community. Breed societies and owners often record colour as one of the main physical compositions of identifying animals. Since sequencing the equine genome in 2007, geneticists were able to identify mutations for basic coat colours. Many coat colour genetic tests are now commercially available to horse breeders. Only a few different genes are responsible for the variation in coat colour that we see in horses.

Black, bay and chestnut

The base coat colours of horses are black, bay and red (or chestnut). Other horse coat colour can result from modifications of the base colours. The two major genes that determine the base coat colours are Agouti (A or gene *ASIP*) and Extension (E or gene *MC1R*). An understanding of these genes gives a good basis towards understanding coat colour genetics. Several other genes exist to control the modification or absence of colour and/or patterning.

The Extension gene is credited with producing the black pigment observed in horses that are black, brown, bay, dun as well as the red pigment seen in chestnuts. Two alleles of Extension are assigned to account for the black/chestnut colour variation in horses. The alleles either extend (E) or diminish (e) the amount of black in the coat. In simple terms, the Extension gene determines whether black pigment is found in the hair and skin (EE or Ee), or only in the skin (ee) (Table 4.1).

Table 4.1. Phenotype and genotype for the Extension (E) allele.

Phenotype	Genotype
Black, brown or bay	EE or ee
Chestnut (red)	ee

An important aspect of the black hair pigment in horses is its distribution. Black hair may be distributed uniformly (black horse) or in a pattern called bay where it is restricted to the mane, tail and legs but reduced or absent on the body. Black patterns characteristics are due to the Agouti gene.

The dominant allele (A) of the Agouti gene causes the distribution of black or brown hair to be restricted to certain areas (e.g., mane, tail, lower legs, ear rims). The recessive allele (a) does not restrict the distribution of black hair and when homozygous in the presence of E, produces a uniformly black horse (Table 4.2).

Table 4.2. Phenotypes for combinations of the E and A alleles.

Phenotype	Genotype
Bay or Brown	EEAA, EeAA, EEAa or EeAa
Black	EEaa or Eeaa
Chestnut (red)	eeAA, eeAa or eeaa

Greying

A young horse that has a progressive greying allele can be born any colour but soon after birth the horse starts to turn grey. The timeframe towards a complete grey horse depends primarily on the base coat colours. Chestnut (ee) base coloured horses will acquire white hairs at a much faster rate than darker base colour horses (Pielberg et al., 2008).

The grey coat colour (*STX17* gene) is represented by a dominant (G) or recessive genotype (g) (Table 4.3). Grey interacts epistatically with all other coat colour genes except white, obstructing their actions. Therefore it is difficult or impossible to determine what coat colour genes it possesses.

Epistasis occurs when the presence of an allele at one locus masks the effect of an allele at another locus (i.e., there is an interaction of alleles at different loci).

As grey colour is produced by the actions of a dominant gene, at least one parent of a grey horse must be grey. A foal with grey coloured parents has at least a 25% chance to be homozygous for grey. Homozygous greys should only have grey offspring. Figure 4.2 illustrates an example of where grey coat colour (G; grey) can be traced through a pedigree. For further reading see: The genetics of the horse, 2000. A.T. Bowling and A. Ruvinsky

Table 4.3. Grey gene phenotype and genotype.

Phenotype	Genotype
Grey	Gg, GG (dominant genes)
Non-grey	Gg (recessive genes)

Chapter 5. Scientific Breeding Programme

What is a scientific breeding programme? It is a programme with a defined breeding goal for the production of the next generation of animals. It is trying to identify individuals in the population with good genetics outside of environment effects (e.g., training, nutrition, health programme, rider etc.). For breeding purposes, only the horse's genetic merit is important as this component can be passed to their offspring. The construction of a breeding programme involves recording selected traits, estimating breeding values, selection of potential parents and a mating programme for the selected parents including appropriate (artificial) reproduction methods.

Breeding programmes are focused on improving the performance of a population. A structured breeding programme should provide a system for collating performance information for individual animals and provide the ability to rank animals based on their genetic merit for performance. Moreover, breeding programmes should provide appropriate dissemination means to allow producers make informed decision that enables mating of the best males with the best females.

Breeding goal

Fundamental to any breeding programme is a clearly defined breeding goal. The breeding goal is defined to improve the genetic ability of an animal's progeny to generate greater profitability. Every breeder should have a specific breeding goal. For example, the breeding goal of the Irish

Sport Horse Studbook is ‘to produce a performance horse that is sound, athletic with good paces and suitable temperament to be competitive at the highest international levels in the FEI disciplines. Similarly, the breeding goal of the KWPN studbook is to breed horses that can perform at Grand Prix level in show-jumping or dressage. A show-jumping breeder might have the goal of producing a horse of international fame that will potentially jump 1.60m, as another illustration. Equally a breeder may have the goal to produce a show hunter. The chances of success for a horse that is “built” for the job, is greater than that of a horse whose build works against him. Moreover, because horses have a long generation interval, the likely circumstances that will prevail in several years need to be taken into account; not only which animal characteristics will be important, but also their degree of importance relative to other traits (i.e., genetic correlations). Figure 5.1 illustrates the key steps of a successful long-term breeding strategy, whatever the species.

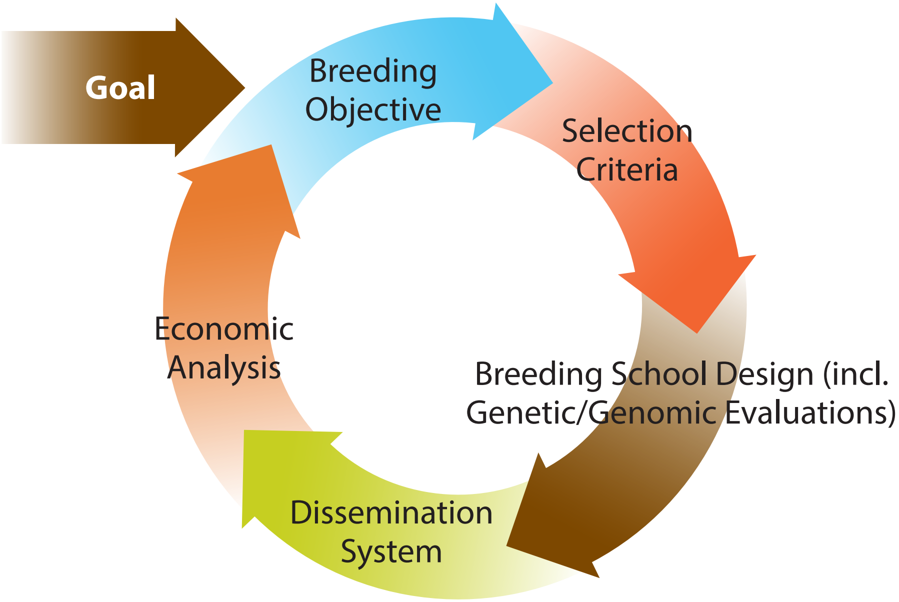


Figure 5.1. Outline of an optimal long-term breeding strategy.

Breeding objective

A breeding objective includes all traits that contribute to the overall goal of the breeding programme. These are traits that breeders or industry would like to improve, specified according to their relative importance. The breeding objective generally includes several traits each appropriately weighted (e.g., how much more/less important is jumping ability relative to health). There should be a long-term emphasis on trait improvement. In general, the more traits included in the breeding objective the slower the rate of genetic progress for each trait per generation. Historically, horses have been selected for a number of traits over time such as speed, jumping ability, strength and for more conventional traits like temperament. Globally, many horse breeds have been selected for preserving and improving traits of interest to their particular studbook. The breeding objective should strike a balance between performance, soundness and health traits. Novel traits such as fertility, longevity, and health should be included in breeding objectives. The healthier the horse, the greater the chance that it can reach its genetic potential and that it can be maintained for a long period.

Any breeding programme should focus on quality over quantity using only proven mares and stallions with good genetics for performance, soundness and health traits. The resulting foals should meet an industry demand. With a view to the intensive and prolonged training path, a horse that is uncomplicated to handle, easy to ride as well as intelligent and diligent, are very important criteria in the pursuit of reaching the highest sport level attainable. A horse could have the best genetics for jumping ability, but if it isn’t sound and therefore cannot compete in competitions this is impractical.

Breeders should be mindful and note that environmental effects can mask genetic effects where possible “bad” genetics can look better than “good” genetics due to environmental effects. For example, if a horse jumps a clear round, that achievement may be partly attributed to its genetic ability and partly due to its environment such as the skill of its rider and quality of its training. For breeding purposes, a horse’s genetic ability is the only factor that is important, as a rider’s talent cannot be passed on to a foal. It’s difficult for management to compensate for bad genetics (and vice versa). Therefore, it is important that all traits are analysed appropriately and breeding decisions are made on genetic information and not phenotypic information. To reduce environmental bias, data are needed from multiple sources including phenotypic data, management/environmental data, pedigree data, and where possible DNA/genomic data. Once this information is known, it allows calculating what proportion of the observed variance in a trait due to genetic influences and what is due to environment/management factors.

Breeding scheme design

The success of a breeding programme revolves around an accurate national database which includes pedigree and performance data. Accurate knowledge of the full ancestry of all animals within a database is a vital component of a successful breeding programme. Genomic testing methodologies as alluded to in Chapter 2 can be used to verify a pedigree and resolve possible mistakes. Every animal should only have one (and only one) unique national identification and this identification method should be common across all countries.

Accurate identification of animals combined with accurate performance recording is essential to identify the genetically elite individuals within a population. All data must be available, good or bad. There is a variance in the performance among a sire's offspring as while a foal receive half of their DNA from their sire, the make-up of that half can differ. Therefore, the distribution of genetic effects inherited can vary around a mean of the sire itself. If only data from animals that received the "good" complement of genes is included in the national genetic evaluations then unless properly accounted for, the sire's genetic merit would be inflated. Similarly, if for some reason data were not available on the elite progeny of an animal then the sire's estimate of genetic merit may be poorer than its true genetic merit. Furthermore, knowledge on where the horse resided as well as other factors such as age should be available facilitating the removal of such environmental effects from the performance record.

Breeding programme

Once accurate genetic evaluations are available, consideration must be given to which horses are mated taking cognisance of expected genetic gain but also any accumulation of inbreeding as this can affect long-term genetic gain. Inbreeding as defined in Chapter 3 is the mating of related animals and results in inbreeding depression, which it is known to have, on average, an unfavourable effect on most performance traits especially those associated with fitness. Although, in some instances, inbreeding can result in favourable characteristics in the offspring. In the absence of DNA information on individual animals, control of inbreeding cannot be achieved without knowledge of the pedigree of an animal. Therefore, all pedigree information must be accurately recorded.

BLUP

A methodology called best linear unbiased prediction (BLUP) has been successful for making genetic improvement in many other livestock species and is the system responsible for generating breeding values. Henderson (1949) originally developed the system for use in cows in the 1950's. In horses, Árnason (1980) was the first to report the actual use of BLUP to evaluate gait for Icelandic horses. The BLUP methodology aims to maximise the correlation between true and predicted breeding values while minimising error variance (Mrode, 2014). BLUP is commonly used in genetic evaluations as it can incorporate all information from all relatives, handle unbalanced designs and account for most selection bias. Today, BULP has evolved into a very sophisticated system and is used by several studbooks to evaluate complex traits such as performance, conformation, movement and athleticism. BLUP relies on good genetic parameters (i.e., parameters that reflect the true populations). As a result, parameters should be estimated in a population frequently.

Genetic evaluation

A genetic evaluation is a statistical assessment of pedigree and performance data from a population taking account of environmental effects. The results of genetic evaluations are published as breeding values. These breeding values estimate the genetic merit of an individual as a parent for a particular trait or series of traits based on an evaluation of all available data on the phenotypic performance of an animal, and close relatives, for a trait (Berry et al., 2011, Oldenbroek and van der Waaij, 2015). Breeding values allow animals to be ranked according to their expected genetic value as a parent. Like heritability, breeding values are population and trait specific.

Routine access to phenotypic performance data and animal ancestry is required to perform accurate genetic evaluations. Phenotypic performance data needs to be collected for traits that are directly related to the breeding goal (e.g., if the breeding goal is to improve jumping ability then all show-jumping results must be collected). Alternatively, this can be done through known correlated traits.

In order to genetically evaluate equine traits, it is necessary to make allowances for the known sources of environmental variation. Methods of genetic evaluation and improvement rest heavily upon comparisons of the performance of animals which have been treated similarly. As an illustration, animals born within a close timeframe, on the same farm, in the same year and managed similarly (i.e., nutrition, training etc.). These animals are commonly referred to as contemporaries and belong to a grouping called contemporary groups. The accuracy of selection is improved by ensuring animals within a contemporary group receive similar treatment as far as possible. With large contemporary groups, the interaction between genetics and the environment can be disentangled more effectively. Various statistical methods exists to decide on the optimal contemporary group size.

Young horses are routinely evaluated in many European studbooks in both show-jumping and dressage. These tests are very popular to breeders and owners who find it important that their horses score well. Often talented riders are hired to train and show the horses at their best. These evaluations provide data for genetic evaluations of young horses as well as their parents, principally by applying the BLUP animal model. The outcomes of these tests also give the breeder an early predictor of the genetic ability of the horse.

Rate of genetic gain

Many discussions on breeding programmes and genetic gain focus on heritability. However, heritability is only one of the factors that influences genetic gain. The rate of genetic progress per generation for a given trait is described as (Rendel and Robertson, 1950):

$$\textit{Annual rate of genetic gain}, \Delta G = \frac{\text{selection intensity} * \text{accuracy} * \text{genetic standard deviation}}{\text{generation interval}}$$

The annual rate of genetic gain is influenced by selection intensity (i.e., the proportion of the population selected to be parents), accuracy of selection (i.e., the information available for the trait), genetic variation for the given trait (i.e., the trait must be transmissible between generations and have variance) and the generation interval (i.e., the average age of parents when their offspring are born). One of the biggest obstacles of genetic improvement with horses is the slow rate of genetic progress due to the long generation interval. For the advancement of breeding it is essential to get younger stallions breeding. If a breeding programme is successful then young stallions should be on average genetically better than older stallions.

When a small proportion of genetically elite animals are selected to be parents, selection intensity is maximised. Selection intensity can depend on reproductive and breeding tools available to producers (Oldenbroek and van der Waaij, 2015); for example, artificial insemination, ET, ICIS and sexed semen all increase the availability of genetically elite breeding animals. This results in a smaller proportion of parents being selected for breeding. Ample genetic variation as well as a shorter generation interval results in a faster rate of genetic gain (e.g., chickens have a much shorter generation interval than horses). Accuracy of selection is influenced by heritability and the quantity of information available on a particular trait. When a trait is lowly heritable, the accuracy of selection will typically also be low unless many records are available. However, if sufficient records are available for lowly heritability traits (e.g., reproductive traits) it is still possible to achieve the same accuracy of selection as traits with a high heritability (Berry et al., 2011).

Genomic selection

Fundamental to a successful breeding programme is the accurate identification of the best (and worst) animals. At birth, a prediction of an animal's genetic merit is obtained by averaging the genetic merit of the respective sire and dam. However, because progeny inherit different pieces of DNA from the parent, in a relatively random process, an accurate prediction of the actual genetic merit is not known until the animal has performance records itself and/or has many progeny with performance records.

Performance is determined by the genes of the horse and how those genes are affected by the environment the horse is exposed to. Genes, which are made up of DNA, remain with an animal throughout life and are identical in every cell of the body. Therefore, the genes in the follicles of a new born foal's hair are the same as the genes that horse contains when jumping a class several years later. Therefore, knowing the genes of a foal and how each gene affects performance allows geneticists to more accurately determine how that animal (and their progeny) would perform in the average environment many years later. This is the science underpinning genomic selection.

Meuwissen et al. (2001) defined genomic selection as the ability to predict individual animal breeding values using whole genome information. These breeding values can be predicted for any genotyped animal, thus providing an indication of the future performance capabilities of that animal and its offspring. In horses, significantly less genomic data has been available but genomic selection has enormous future potential to improve the genetics of horses given their long generation interval (Stock and Reents, 2013).

The process behind genomic selection includes generating SNP (Chapter 2) effects for a genotyped reference population with recorded phenotypes for a specific trait (or number of traits). In this way, the effects of all loci that contribute to the genetic variation are captured, even if the effects of the individual loci are small and not significantly different from zero. Selection candidates (in subsequent generations) are then genotyped and their genotypes are combined with the estimated SNP effects to estimate a genomic breeding values for each animal. Principles of genomic selection are illustrated in Figure 5.2.

The main differences between the traditional method of estimating breeding values (i.e., BLUP) and genomic selection are that pedigree recording is essential for traditional BLUP- breeding values but not required for genomic selection. In addition, unlike BLUP, elite breeding animals (i.e., the selection candidates or their progeny) do not have to be trait recorded (phenotyped) for genomic selection (Meuwissen et al., 2016). There are significant advantages of implementing genomic selection into an equine breeding programme. Animal DNA mapping allows genomic breeding values to be predicted for a foal at only a few days old. Colt foals with excellent genomic breeding values could be used on mares when the animal starts to produce semen. As a result, a large decrease in the generation interval could be obtained as animals can be bred at an earlier age. In addition, genomic selection significantly increased the rate of genetic gain by improving the reliability of breeding values. With traditional breeding many stallions are old before breeders know they are excellent sires.

A major limitation of genomic selection is the large number of markers required and the cost of genotyping to obtain genetic markers. However, both these limitations have been largely overcome following the sequencing of the livestock genomes, allowing for the subsequent availability of hundreds of thousands of single nucleotide polymorphisms (SNP), (Hayes and Goddard, 2001).

Currently 65,000 pieces of DNA (i.e., Equine SNP70 Beadchip) are measured in the horse although the technology is available to measure a much higher amount of DNA (High Density genotyping platform). Nonetheless, of greatest importance is accurate knowledge of the association between each piece of DNA and the range of performance traits where data is available.

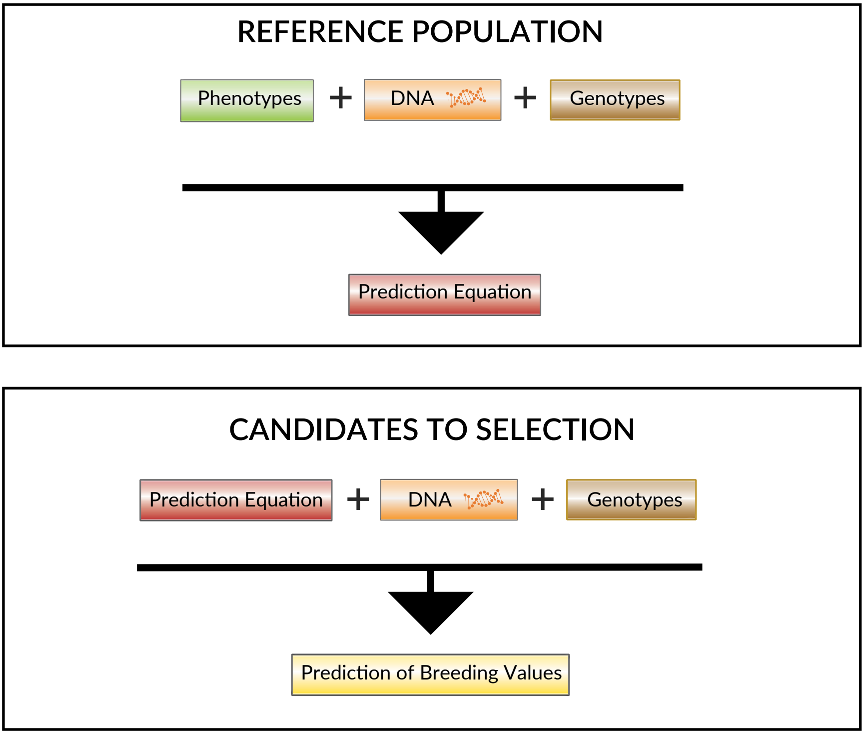


Figure 5.2. Top: a prediction equation is obtained from a reference population with phenotypes and genotypes; bottom: this prediction equation is used on candidates with genotypic information only. *Boichard et al., 2016.*

Dissemination of genetics

The wide impact of genetic improvement depends on the dissemination of genetic material. Artificial insemination is one of the most effective methods to disseminate superior genetics. In 2016, Corbally and Fahey (2017) reported that 42% of Irish Sport Horse breeders covered their mares through AI. This represented an increase of 10% since 2011 when research was previously conducted. However, the authors concluded that although a significant increase has been seen in Ireland, the extent of AI usage still lags well behind its use in the main European studbooks (i.e., KWPN, Belgian Warmblood, and Swedish Warmblood. Selle Francais, Holstein and Hannoverian) where the use of AI varies from 70% to 100%.

Embryo transfer is another effective method to disseminate superior genetics. Increasing the reproductive rate of females by ET is mainly useful in species with low reproductive rates such as horses. In the horse, ET was first performed successfully in 1972.

Intracytoplasmic Sperm Injection is a more complex reproductive technology and its use in mares is increasing. This reproductive technology involves injecting a single sperm into an oocyte through a microscopically fine needle in vitro (Allen, 2005). The use of ICSI requires highly developed technical skills and is expensive.

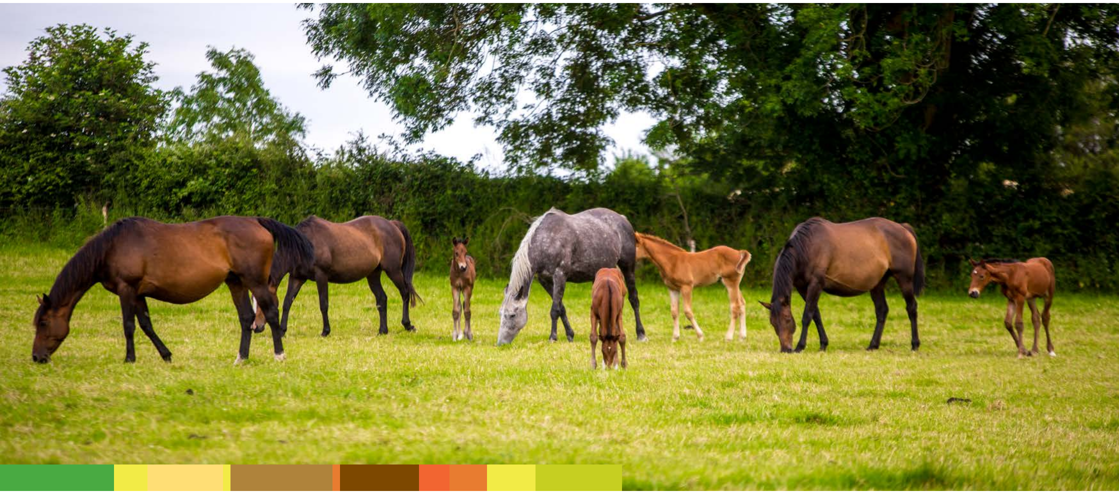
The use of ET and ICSI have several advantages, chief amongst which is producing multiple offspring from 'superior' mares in a single year. Embryos can be harvested from the mare without removing her from competition. Both reproductive technologies result in greater selection intensity on the mare line, as well as more accurate estimation of breeding values (i.e., mares will get a greater number of related phenotypic records).

Economic analysis

Breeding horses is a slow process but genetics is permanent and cumulative. Therefore, breeders must aim to improve genetic merit each generation. Genetic gain must be constantly reviewed and the impact of genetic differences in (economic) performance quantified.

Any economic analysis should consider both income and expenses. Given horse breeding is a slow and long-term process, returns from breeding decisions may only be realised several years later. Breeders need to have a long-term approach as considerations that may not be important in the short-term could have significant impacts in the long-term.

The bases for an economic analysis is net profit and return on investment. When breeders develop their breeding goals, emphasis should be placed on maximising profitability. This is advisable unless there are clear reasons to deviate from this strategy.



Horse Sport Ireland

In Ireland, Horse Sport Ireland (HSI) as the national governing body for equestrian sport carry out genetic evaluations for all show-jumping horses. Breeding values for show-jumping ability at international level are produced which measures the difference between each horse's genetic ability for international level show-jumping and the average genetic ability for show-jumping in the Irish population. If genetic evaluations are accurate then genetically elite horses should, on average, outperform genetic inferior horses (HSI, 2020).

Measurement of show-jumping ability

The breeding objective of the Irish Sport Horse Studbook is "to produce a performance horse that is sound, athletic with good paces and suitable temperament and capable of winning at the highest international level in FEI disciplines". It is vital that the genetic evaluation reflects a horses' ability to produce progeny that can compete successfully at the highest levels and this requires direct measurement of international performance.

In 2009, HSI published a new breeding policy for the Irish Sport Horse studbook which introduced the award of star ratings to horses that are successful as performance horses. These ratings enable breeders to quickly identify older stallions with a proven track record and younger stallions that are out-performing their contemporaries and are potentially future stars. Star ratings and merits for stallions, mares and geldings are periodically reviewed and are based on all verifiable performances. Star ratings are based on the highest level in sport that a horse has achieved successfully (i.e., with at least two double clear rounds or a top four placing in single-round speed competitions).

Lifetime performance ratings are based on the star rating system and have been chosen as the measurement of performance for the genetic evaluation. This is based on comprehensive national level information from the Show-Jumping Ireland (SJI) database and international results collated by HSI from the FEI and other sources.

Breeding values generated for stallions and mares available to Irish breeders consider successful international level performances and reflect how well the stallions cross with Irish mares. Breeding values can also be generated for foreign based stallions based on their own performance record. One difference between the genetic evaluation and the star ratings awarded to stallions is that the lifetime performance rating of all competing progeny of a stallion are included in the genetic evaluation, whereas star ratings are based on a stallion's best two progeny (HSI, 2020). Star ratings are a phenotypic measurement of performance reflecting the best of a stallion's progeny. Breeding values are a genetic measurement of the average improvement in performance of a stallion's progeny when compared to those of other stallions (HSI, 2020).

Lifetime performance rating in the Irish Sport Horse Studbook are in Table 5.1. Lifetime performance ratings allow for the incorporation of international level performances in a routine and timely fashion. The methodology can be easily extended for use in other disciplines such as eventing when sufficient data are available and can be integrated with linear profiling information in the future.

Lifetime performance ratings are converted into a numeric score for each horse. This score reflects the percentage of Irish Sport Horses that compete successfully at this level and measure the difference between the horse and the average Irish Sport Horse show-jumper in standard deviation units.

Table 5.1. Lifetime performance rating in the Irish Sport Horse Studbook.

Lifetime Performance rating	Criteria	Equivalent stallion star rating	Equivalent mare/ gelding star rating	Comparison with Irish Sport Horse show-jumpers
FEI CSI Championship	Competed at major international championships	5*	5*	Top 0.2%
FEI CSI 5*	Two double clear rounds at FEI CSI 5* Grand Prix or Nations Cup level	5*	5*	Top 0.4%
FEI CSI 4*	Two double clear rounds at FEI CSI 4* Grand Prix or Nations Cup level	4*	5*	Top 0.6%
FEI CSI 3*	Two double clear rounds at FEI CSI 3* level	3*	4*	Top 1.5%
FEI CSI/National 1.50m	Two double clear rounds at FEI CSI or National 1.50m level	2*	3*/4*	Top 2.5%
National 1.40m	Two double clear rounds at National 1.40m level	1*	3*	Top 3.5%
National 1.30m	Two double clear rounds at National 1.30m level		3*	Top 12%
National 1.20m	Two double clear rounds at National 1.20m level		2*	Top 25%
National 1.10m	Two double clear rounds at National 1.10m level		1*	Above average
National 1.00m	Two double clear rounds at National 1.00m level			Average
Below national 1.00m	Has not achieved two double clear rounds at National 1.00m level			Below average

Source: HSI (2020)

Breeding values

Estimating breeding values are expressed as an index with an average value of 100. This baseline value of 100 reflects the average genetic merit of active breeding Irish Sport Horse mares born between 1999 and 2013 and enables comparisons between horses. Every year, this baseline moves to include mares between four and eighteen years of age so that the baseline value of 100 reflects the progress being made within the breed. The spread of the breeding values is set to a standard deviation of 20. The spread of breeding values follows a normal distribution with approximately two-thirds of horses with breeding values between 80 and 120 and are considered average for the breed (HSI, 2020).

For example, horses with breeding values of over 120 are considered to be of high genetic merit and are breed improvers for that trait. Horses with breeding values over 130 considered to be superior breed improvers for that trait. The true genetic merit of a horse can never be known exactly. Although both a horse's own performance and its progeny provide information about its genetic merit (and parentage), performances are affected by factors other than genetics and progeny share just half of their parents' genes (Chapter 1).

Breeding values are estimates of genetic merit and should always be considered by breeders as well as the reliability. Breeding values can change as more data becomes available; for example, as the number of a stallion's progeny competing increases. More information on a horse results in a higher reliability. The reliability value reflects the amount and type of information used in the calculation of a breeding value and ranges from 0 to 99% (Chapter 2). All Irish Sport Horse stallions with show-jumping breeding values with a reliability greater than 75% have at least 20 progeny included in their genetic evaluation (HSI, 2020).

In general, less information is available on foreign and Thoroughbred stallions as few have parents that have been genetically evaluated in Ireland. This leads to lower reliabilities for these horses. Horses with low amounts of information available receive breeding values close to 100 as this is the average of the breed. Breeding values for foreign stallions may also be available from their studbook of origin (e.g. studbooks in the Netherlands, Germany, Belgium, Sweden and France). Although breeding values between countries are not directly comparable, they may be useful for stallions standing abroad without sufficient numbers of progeny recorded in Ireland. Performances at the national level in the UK and the USA have been included in the genetic evaluation for many horses, where possible (HSI, 2020).

Why breeders should refer to Breeding Values when choosing a stallion?

Show-jumping breeding values predict the show-jumping ability of a stallion's offspring. For Irish Sport Horse stallions with show-jumping breeding values greater than 130, over twenty times as many of their offspring will compete at international level compared to the offspring of stallions with show-jumping breeding values less than 100. Foals by stallions with show-jumping breeding values above 130 achieve, on average, €445 more at auction than foals by stallions with show-jumping breeding values below 100 (HSI, 2020). For further reading see: The Irish Sport Horse Studbook Genetic Evaluation Report 2020. horsesportireland.ie/wp-content/uploads/2020/12/final-genetic-evaluation-report-2020.pdf.

KWPN studbook: Most successful scientific breeding programme

In 2020, the WBFSH ranked the KWPN (Studbook of the Royal Dutch Sport Horse) as the number one studbook in all three disciplines of show-jumping, dressage and eventing, The KWPN distinguishes four breeding directions: Riding horses, (subdivided into show-jumping and dressage disciplines), harness and Gelder horses. Riding horses represent the largest group (85-90%). Even though each breeding direction has defined breeding objectives for that particular discipline, all horses fall under the general KWPN breeding goal which aims to:

- breed a competition horse that can perform at Grand Prix level
- have long usefulness
- have the character that supports the will to perform as well as being friendly towards people
- have functional conformation that enable good performance
- have attractive exterior, along with refinement, nobility and quality

Scientific research and market trends can lead to reconsideration of breeding objectives.

KWPN mare and stallion selection

Performance testing of both mares and stallions includes options for 'station testing' at centers where the conditions are standardised for all animals. In addition, professional riders are availed of for the training period.

Stallion selections

For stallions, there is an obligatory path to become a studbook approved stallion. The stallion path has four steps:

1. Inspections of the stallion: stallions are inspected on hard surfaces for correctness of legs and body and on loose jumping (jumping horses) or free movement (dressage stallions).
2. Health checks: in the various stages of the stallion selection process, stallions must meet minimum demands for clinical correctness of body and functioning, radiographs and semen quality. In addition, endoscopic examinations are performed for roaring (i.e., recurrent laryngeal neuropathy).
3. Station performance test: depending upon their age, stallions must prove their sport ability during a central performance test of a maximum of 70 days.
4. When a stallion has offspring, these offspring are inspected and/or followed in sport.

   Based on the collected information, various breeding values are estimated for each stallion. Based on their estimated breeding values, the stallions are evaluated when their eldest offspring is 1 year, 3 years, 7 years and 11 years of age. The evaluation of foals is undertaken to observe the quality of a young stallion's first crop of foals and the type of mare best suited for that stallion. After the final evaluation stage, the committee either approves a stallion for life or can dismiss him from breeding.

Stallions whose progeny have demonstrated excellence in performance are granted recognised status even if they do not fully fulfil the requirements for conformation or radiograph results. Once they have proven themselves superior in performance or breeding, they are granted the same status as approved stallions. Since 2009, the KWPN require standard radiographs of newly approved stallions' first yearlings in order to gather information to eliminate Osteochondrosis (OC) from the population. Once a stallion passes the test and are approved for breeding, the breeding value for OC is published in the KWPN Stallion Database (and updated each year).

Mare selections

Inspection of mares and foals is performed at district level and then the best go on to central inspection, and finally to national inspection offering an opportunity for foals and mares to be benchmarked against the best for their age category. Mares also have the option of undergoing one-day performance testing under saddle (IBOP) or either a five- or two-week station test (EPTM) is possible.

The selection and use of the best mares is stimulated by the studbook by issuing PROK (Project Radiographical research KWPN) certificates. To be issued with the PROK certificate radiographs are taken of the front and hind legs. The navicular bone, sesamoid bone, the fetlock joint and coffin joint are evaluated. In the hind legs, the hock, stifle and fetlock joint are evaluated. A PROK predicate is awarded to KWPN horses that comply with the x-ray requirements of the studbook. The mare can earn PROK certificates based on her own qualities (i.e., performance, conformation and heath) or based on qualities of the offspring (i.e., performance and conformation).

Data collected by KWPN

Linear scores of conformation and movement from 20 random foals of each new approved stallion are recorded. In addition, radiographs for OC from 20 random yearlings of each new approved stallion are recorded. Linear scores on conformation and movement of 3 year old mares offered for studbook inspection are recorded. Jumping and free movement of all young stallions offered for the stallion selection process are recorded. Marks obtained in one day performance tests for mares (IBOP) and station tests (EPT) are all recorded. Competition results of all registered horses are continually recorded. This data is then used to predict breeding values for all stallions and mares.

KWPN breeding values

Breeding values are estimated for all linear scored traits (conformation, movement, loose jumping), OC, show-jumping and dressage. These breeding values are communicated to breeders, and used during their selection process to evaluate the breeding programme and to identify optimal combinations of the stallion and mare.

Scores from the performance tests are used for breeding value estimation. This is due to little information being available for young horses as these horses don't have sport results at this stage. Research by the KWPN shows that the results from the performance tests have a strong correlation with later sport results. Therefore, these tests have a high predictive value. The performance tests are the foundation of breeding values.

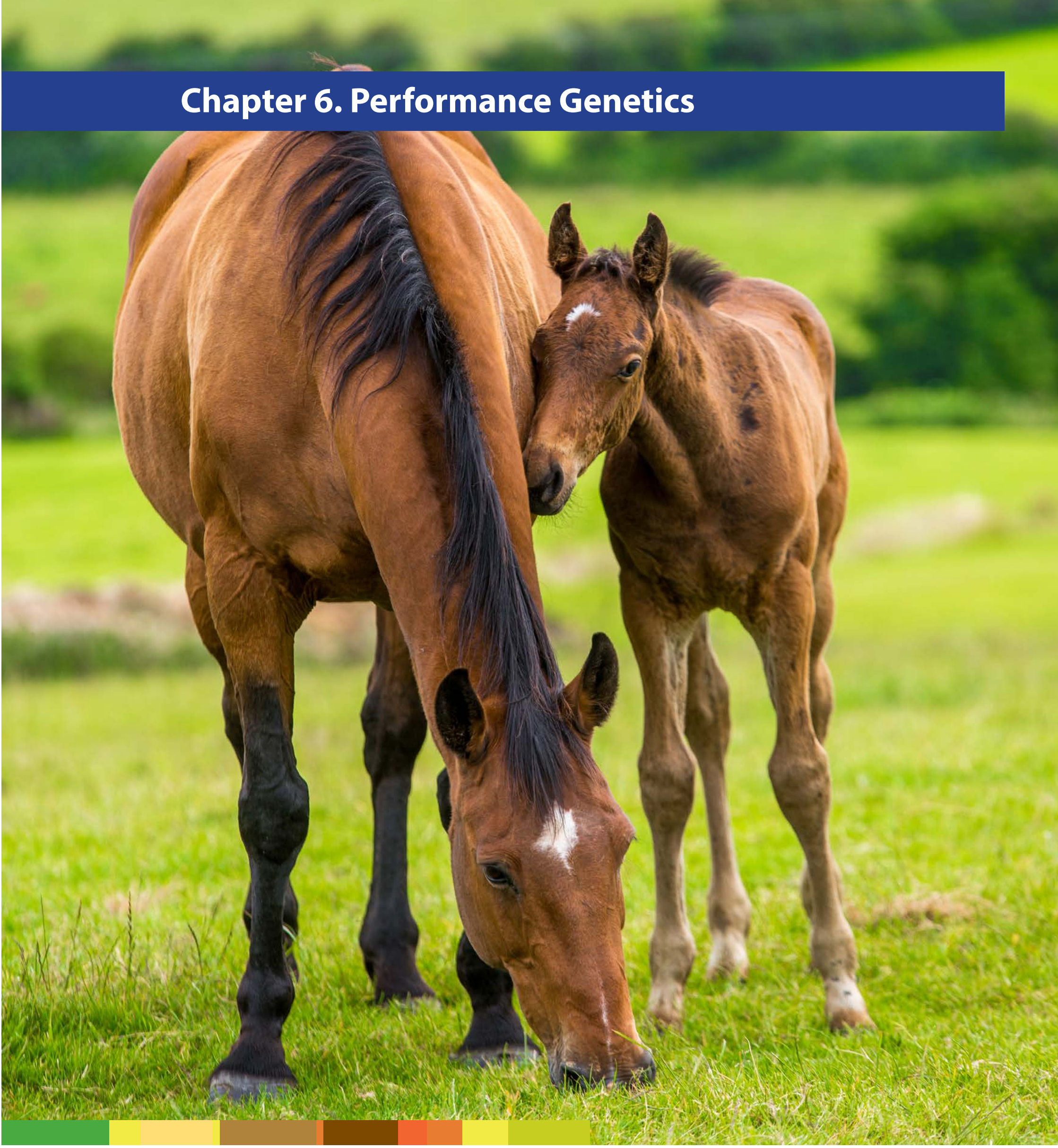
DNA Information

Since 2016, the KWPN has predicted genomic breeding values for OC. While OC is strongly influenced by environmental factors (e.g., management and nutrition) a large part of the trait is heritable (Chapter 7). This means, through selective breeding, the incidence of this trait can be greatly reduced.

Genomic predictions are so far only available for the OC trait in both breeding directions of dressage and show-jumping. The index expresses a stallion's tendency to pass on OC to his progeny. The KWPN studbook radiographs a random sample of a stallion's progeny at 9-18 months to indicate genetic transmission of OC. The prevalence of OC in the population is continuously updated through x-raying young animals. The DNA composition of a random sample is added as a source of information to the breeding values estimated for OC which can provide a good assessment of the heritability of OC without need for an offspring test.

The KWPN studbook normally refuses to licence stallions with a severe OC-remarks; however, in some cases where the stallion demonstrates exceptional talent, they can be approved. Having this information available to breeders is highly valuable. Breeders have the option to then avoid stallions that have a high tendency to pass on OC to their progeny which can result in an immense loss in value of the animal.

Chapter 6. Performance Genetics



Given the vast differences among horses and breeds it is obvious that performance has a genetic component. This is recognised by breeders who select superior preforming horses for breeding stock. The Thoroughbred, Standardbred, French Trotter, Swedish Trotter and Quarter Horse breeds are particularly suited to racing (Hill et al., 2013). Sport horses have been strongly selected for jumping and other athletic phenotypes (Table 6.1).

Table 6.1. World Breeding Federation for Sport Horses Stud Book Rankings 2020 for Eventing, Show-Jumping, and Dressage.

Eventing	Show-Jumping	Dressage
1. KWPN	1. KWPN	1. KWPN
2. Selle Francais	2. Selle Francais	2. Oldenburger
3. Hannoverian	3. Belgium warmblood	3. Westfälisches
4. Irish Sport Horse	4. Zangersheide	4. Hannoverian
5. Holstein	5. Oldenburger International	5. Deutsches Sportpferd
6. Oldenburger	6. Deutsches Sportpferd	6. Lusitano
7. Deutsches Sportpferd	7. Hannoverian	7. Danish Warmblood
8. Trakener	8. Holstein	8. Rheinisches
9. Swedish Warmblood	9. Oldenburger	9. Belgian Warmblood
10. Anglo European	10. SBS (Belgian Sport Horse)	10. Trakhener
11. Anglo Arabe	11. Anglo European	11. British Hanoverian
12. Belgium warmblood	12. Westfälisches	12. Swedish Warmblood
13. Great Britain Sport Horse	13. Mecklenburg	13. Holsteiner
14. Zangersheide	14. Irish Sport Horse	14. New Zealand Hanoverian
15. Oldenburger International	15. Swedish Warmblood	15. KWPN of North America

Heritability of Performance

When a breeder evaluates a pedigree they are trying to evaluate the genetic contributions from ancestors in the belief that racing performance is inherited. Heritability estimates how much of the variation in athletic phenotypes are due to genetics independent of management (Chapter 2). The basic components of performance genetics are to establish trait heritability, determine breeding values for individuals and discover genes that influence performance traits (Bailey and Brooks, 2013).

Sport Horses

In some European countries, performance testing of sport horses has been carried out in a comparable and systematic way over the last few decades. This allowed for genetic analyses to be carried out on reasonably large data sets with thousands of individual horses sired by at least 100 stallions (Bowling and Ruvinsky, 2000). The methods used for estimating genetic parameters are based on BLUP techniques (Chapter 5). The average heritability estimates of performance traits for young stallions and mares from several countries are in Table 6.2. These estimates of heritability are based on data from Warmblood riding horses in Germany, Netherlands, Sweden and Switzerland (Bowling and Ruvinsky, 2000).

Table 6.2. Average heritability estimates of performance traits for young stallions and mares (Bowling and Ruvinsky, 2000).

Tests	Trait	Heritability
Stallions' test	Temperament	0.41
	Walk	0.55
	Trot	0.58
	Gallop	0.56
	Riding ability	0.44
	Free Jumping	0.56
Mares' test	Walk	0.25
	Trot	0.30
	Gallop	0.27
	Riding ability	0.20
	Free Jumping	0.24

Thoroughbreds

Much of the research investigating heritability of performance traits has occurred in Thoroughbreds. Heritability estimates vary depending on the analytical model and the performance measure. Several studies use earnings, handicap ratings, and racing times (Chowdhary, 2013). Heritability estimates for earnings range from 0.23 to 0.56 among Thoroughbreds (Tolley, 1985). Between Japanese Thoroughbreds, heritability for lifetime earnings has been estimated as 0.12 (Tozaki et al., 2010). Heritability estimates of race time vary from 0.09 to 0.78 (Tolley, 1985). In addition, the heritability of race time has been observed to decrease as the race length increases (Oki et al., 1995). Handicap rating heritability estimates fluctuated from 0.24 to 0.76 (Tolley, 1985, Gaffney and Cunningham, 1988). In Australian Thoroughbreds, best race distance was a strongly inherited performance trait which has been estimated as 0.94 (Williamson and Beilharz, 1998). It is estimated that genetics is responsible for up to 35-55% of a horse's racetrack success (Gaffney and Cunningham, 1988, Mota et al., 2005); therefore, while genetics is very important in determining success it will never be the only factor.

Thoroughbred gene tests

The speed gene

The myostatin gene *(MSTN)* has been shown to be responsible for muscle development and muscle fibre type in a range of species (Grobet et al., 1997). Horses, but particularly Thoroughbreds, have a very high muscle mass to body weight ratio (55%) compared to other mammalian species (30-40%) (Gunn, 1987). In 2009, Prof Emmeline Hill, an equine genomics scientist at the Animal Genomics Laboratory at University College Dublin established that the myostatin gene had an association with sprinting ability and stamina in Thoroughbreds. The myostatin gene can control the development rate of muscle and the type of muscle that is formed (fast-twitch or slow-twitch). The Thoroughbred horse has a high level of muscling; therefore any difference in muscling caused by genetic variations is thought to have a significant effect on racing performance. Studies have shown that there are three types of speed gene known as C:C. C:T and T:T (Hill et al., 2010).

- **C:C horses** have more muscle and more fast-twitch muscle and best suited to sprint distances (5-8 furlongs). They tend to perform well at yearling sales.
- **C:T horses** have a mix of fast-twitch and slow twitch muscles meaning they are suited to middle distances (6-10 furlongs).
- **T:T horses** have a predominance of slow-twitch muscles making them best suited to longer distance races (8 furlongs plus).

Muscle mass at two years old is significantly greater (7%) in C:C horses compared with T:T horses. This suggests a more precocious development of the skeletal musculature with C:C horses. In addition, C:C horses earned up to 13 times more in prize money than T:T horses at two years old, when race distances are primarily limited to ≤8 f (Hill et al., 2013).

A genetic test is now available to horse breeders, trainers and owners. *The Equinome Speed Gene Test* can be used to establish if their horse is more genetically suited to be a sprinter, middle-distance or long-distance racehorse. This can improve decision making in selection, breeding and training.

Elite performance test

Equinome have developed a second test called the Equinome Elite Performance Test. Thoroughbred horse breeders, trainers and owners have the option to test for genetic markers that could indicate if foals and yearlings are likely to perform at the elite level. The genetic information provided by the Equinome genetic tests can be applied at all stages of an individual horse's lifecycle to inform and optimise critical breeding, selection, purchasing and training decisions.

Chapter 7. Health Genetics

Poor herd health has the potential to have devastating effects. This can be the risk to animals themselves but also from an economic perspective where performance capacity is significantly reduced. Genetic disease, for example, can be very costly to control. To avoid these circumstances the aim should be prevention or control. Many health disorders have genetic causes, but, in horses the emphasis has predominately focused on keeping high standards of hygiene, diet and management rather than focusing on genetic improvement of disease resistance (Bowling and Ruvinsky, 2000).

Genetics of skeletal disorders

In horses, two of the most common skeletal diseases are OC and navicular disease; both of which can lead to reduced performance, early retirement an large financial losses. Current research has focused primarily on the molecular mechanisms involved. The substantial progress of comparative genomics and the horse genome sequencing project provide a very effective approach to unravelling the genetic basis of OC and navicular disease. Radiological examinations to diagnose skeletal diseases have become an integral part in veterinary orthopaedic practices and pre-purchase examinations. The outcome of radiological examinations has considerable economic impact on the market value of a horse and as a selection criterion for breeding stallions.

Osteochondrosis

Osteochondrosis is caused by a disturbance of the endochondral (within cartilage) ossification of the epiphyseal growth cartilage (Jeffcott, 1996). At the epiphyseal growth cartilage, the disturbance can lead to the formation of partially or completely detached fragments (osteochondrosis dissecans (OCD), fissures, or subchondral bone cysts, leading to inflammation. Areas of the growth cartilage fail to become converted to bone (Chowdhary, 2013). Lesions occur at predisposed sites that are specific to the species and joint and lead to joint inflammation and osteoarthritis. Osteochondrosis is a frequent cause of pain, lameness and reduced performance in young athletic horses (McIlwraith, 1993).

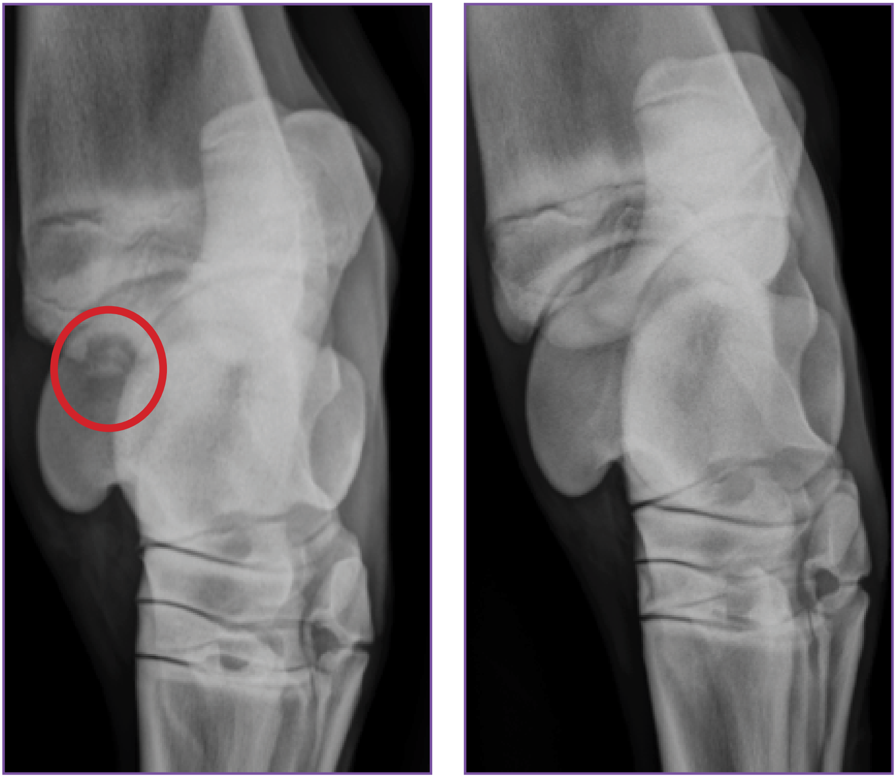
The fetlock, hock, and stifle joints are the sites mainly affected due to this developmental disease. The disease is detected using radiographs. It is acknowledged that OC is a major problem in the equine industry. Harst et al. (2005) demonstrated that OC contributed three times more to economic losses compared to any other equine disease. Large differences in prevalence have been reported. Wittwer et al. (2006) and Van Grevenhof et al. (2009) reported frequencies between 10% and 64% across a range of Warmblood and trotter horse populations. Environmental conditions, breed and type of sampled data in these studies could be attributed to the large variation in prevalence.

One presentation of OC is OCD which results in failure of the developing and maturing process of growing cartilage that may affect either the growth plate or articular cartilage. Cartilage at the top of the growth plates doesn't undergo the normal hardening process and may break down. Extra fluid can form in the joints due to the inflammation caused. It can also present as fragments or cracks in cartilage and cysts in underlying subchondral bone. Sometimes bone and cartilage fragments break off from the main bone and float free in the joint fluid (joint mice). Many OCD lesions heal within a few months. However, those that do not can cause permanent problems when not correctly treated. Figures 7.1 and 7.2 illustrate examples of OCD in the hock and stifle joints. It is estimated that 20-25% of newborn foals will develop some form of OCD while the number of affected horses worldwide is increasing (Bourebaba et al., 2019). Furthermore, OCD is a serious welfare issue in horses, due to extensive stress on leg joints during exercise.

Osteochondritis dissecans occurs primarily in young and growing horses (4 - 18 months of age). Osteochondritis dissecans occurs in the stifle from 6 - 24 months; in the hock from 6 - 18 months and up to 36 months; and in the shoulder from 12 - 18 months of age. Joint swelling, and mild lameness are often present and young foals may have difficulty in rising.

The causes of OCD are poorly understood. Nonetheless, a number of factors have been implicated including genetics, nutrition, skeletal growth rates, minerals, physical activity, and hormones (Al-Hizab et al., 2002). Heritability estimates for OCD in Warmblood and Standardbred horses ranged from 10% to 40% (Distl, 2013). Stock and Distl (2005) illustrated that it is now possible to select for health (e.g., reducing OCD incidence).

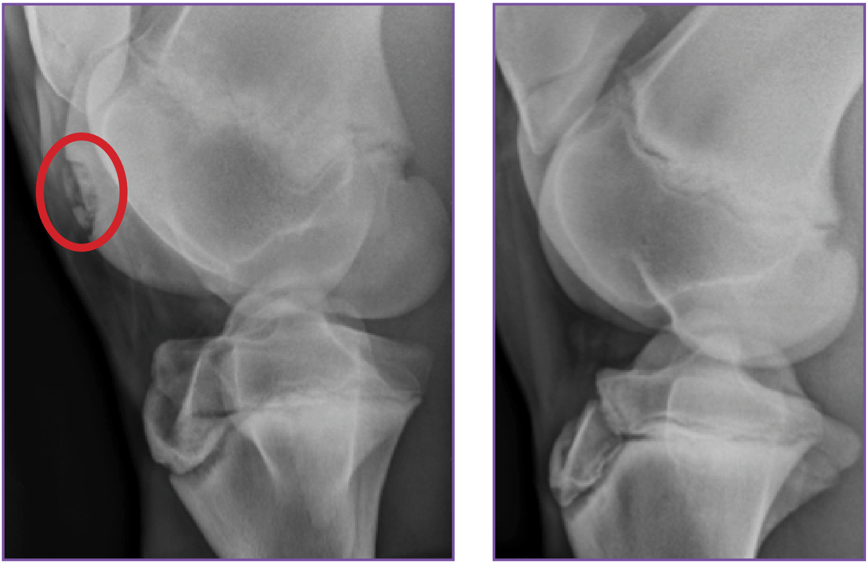
Osteochondrosis is heritable as illustrated in numerous studies (Table 7.1). Reported heritability estimates with adequate genetic variation suggest selection for skeletal soundness may be achievable (Chowdhary, 2013). In 2016, the KWPN studbook introduced genomic breeding values for OC. The breeding values for OC provides breeders with an estimate of the likelihood the horse will pass OC to the next generation; a horse has a higher or lower chance than average to pass OC on to his or her offspring.



OCD in hock (circled in red)

Normal hock

Figure 7.1. Example radiograph of OCD in the hock (image left) and normal hock (image right).



OCD in stifle (circled in red)

Normal stifle

Figure 7.2. Example radiograph of OCD in the stifle (image left) and normal stifle (image right).

Table 7.1. Heritability estimates for OC in different limb joints in different horse breeds.

Population and number of horses investigated	Radiographic findings	Heritability estimate	Reference
Norwegian trotters (n = 644)	OC[2] (hock)	0.21	Grøndahl and Dolvik (1993)
Dutch warmblood (mares; n = 590)	OF[1] (hock)	0.14 ± 0.17	KWPN (1994)
Dutch warmblood (stallions; n = 1,965)	OC[2] (hock) OC[2] (stifle)	0.11 0.09	Der Kinderen (2005)
Dutch warmblood (foals; n = 811)	OC[2] (fetlock) OC[2] (hock) OC[2] (stifle) OCD[3] (fetlock) OCD[3] (hock) OCD[3] (stifle)	0.08 ± 0.10 0.15 ± 0.08 0.07 ± 0.06 0.06 ± 0.07 0.26 ± 0.09 0.02 ± 0.04	Van Grevenhof et al. (2009)
Hanoverian warmblood (n = 3,725)	OF[1] (fetlock) OF[1] (hock)	0.19 ± 0.03 0.37 ± 0.06	Stock et al. (2005)

[1] Osteochondral fragments.
[2] Osteochondrosis.
[3] Osteochondrosis dissecans.

Navicular disease

Navicular disease is one of the main causes of chronic forelimb lameness in middle-aged horses. Because the condition is both chronic and degenerative, it can be managed in some horses but not cured. Riding horses show very often (up to 70–80%) slight or moderate changes in the navicular bone, whereas moderate to severe changes are at low frequencies of 2-5% (Chowdhary, 2013). Heritability estimates for radiological changes of different severity in the navicular bone ranged from 0.10 to 0.34 (KWPN, 1994, Stock and Distl, 2006).

Genetic disorders

Warmblood fragile foal syndrome

Warmblood fragile foal syndrome (WFFS) is a relatively new condition for breeders in Europe although it has, for example, affected the Warmblood population in the United States for some time. This is a heritable genetic condition found in Warmblood horses. Foals can be born prematurely and/or born with abnormalities usually skin abnormalities. The skin is elastic and easily tears causing wounds to arise. Hyperextension of the joints is another common symptom; foals are not viable and are usually euthanised soon after birth. Warmblood Fragile Foal Syndrome is caused by a single mutation in the lysyl hydroxylase 1 *(LH1)* gene. An affected WFFS foal is born with two copies of the mutated *LH1* gene, one coming from each parent. Warmblood Fragile Foal Syndrome is an autosomal recessive trait; this means that a foal can only be affected if the foal inherits the disease from both parents. Additionally, parents that are carriers do not have any symptoms associated with WFFS. However, they will pass on a copy of the defective gene to their offspring 50% of the time. If breeding two carriers, the foal has a 25% chance of being affected and a 50% chance of being a carrier. Table 7.2 illustrates the inheritance patterns of WFFS. This disease can be eradicated if all animals are tested for the disease to avoid matings that could result in affected foals being born.

Table 7.2. Warmblood fragile foal syndrome inheritance.

Symbol	Status	Description
WFFS/WFFS	Affected	The horse carries two copies of the WFFS mutation and is homozygous for WFFS. The horse is affected with the WFFS genetic disorder.
N/WFFS	Carrier	Both the normal and mutant alleles were detected. Horse tested heterozygous for WFFS and is a carrier of the WFFS mutation associated with your breed.
N/N	Clear	Horse tested negative for WFFS and does not carry the WFFS gene mutation. The horse will not pass on the defective gene to its offspring.

Hoof wall separation disease

Hoof wall separation disease (HWSD) is a debilitating genetic disease found in a small percentage (<15%) of Connemara ponies. This disease affects all four hooves and is incurable. The clinical sings of HWSD are not always visible but include breaking away of hoof wall (Figure 7.3), weight bearing issues, difficult to shoe and lameness. This disease is caused by a single mutation and can be inherited depending on the chosen matings. It is important that two carriers are not crossed as this could result in a 25% chance of the progeny having the condition.

This disease can be eradicated if all animals are tested for the disease and where breeders avoid matings that could result in affected foals being born. It is also important to understand that carriers should not be excluded from breeding as this would further reduce the gene pool.

Figure 7.3. Clinical sings of HWSD include breaking away of the hoof wall. *Connemara Pony Breeders' Society.*

Polysaccharide storage myopathy

Polysaccharide Storage Myopathy *(PSSM1)* is a glycogen storage disorder found in several breeds (e.g., Thoroughbred, Draft, Warmblood and many others). The condition is a form of tying-up disease is horses. Clinical sings include stiffness, sweating, muscle damage and an inability to move. McCue et al. (2008) used a genome wide association scan to identify *ECA10* and *GYS1 (glycogen synthase 1)* as candidate genes. A genetic mutation in the *GYS1* gene causes PSSM Type 1 *(PSSM1)* (McCue et al., 2008, Valberg, 2018). The severity of the disease is strongly influenced by a high grain diet (e.g., higher amounts of glycogen) as well as limited exercise. A genetic test is now available to identify a horse with the disease (Valberg, 2018). As a result, the diet of affected horses can be modified by reducing grain or using commercially available feed formulations designed for these horses.

Genetics of respiratory diseases

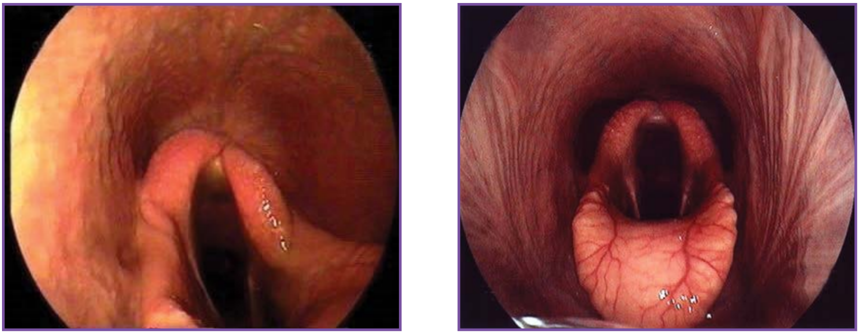
Recurrent Airway Obstruction

Recurrent airway obstruction (ROR) also known as heaves is a chronic respiratory disease associated with allergy. Results from gene mapping studies have identified genes in regions in *ECA13* and *ECA15* for two large sire families in Warmblood sport horses (Swinburne et al., 2009). Nonetheless, the influence of genetics is small compared to the role of environmental factors. The presence of allergens from poor quality hay are the main environmental factors causing the disease.

Recurrent laryngeal neuropathy

Recurrent Laryngeal Neuropathy (RLN) is an obstructive upper-airway disorder caused by degeneration of the recurrent laryngeal nerves and leading to paralysis of the muscles served by those nerves. Affected horses will make a roaring or whistling noise during exercise and the condition impairs performance during intense exercise. Figure 7.4 shows an example of RLN. This is a relatively common condition of racehorses causing the muscles of the larynx to function inadequately. Potential purchasers at sales increasingly ask for endoscopic examinations ('scoping') to be performed in an attempt to assess 'soundness of wind'. The disease has several names including laryngeal hemiplegia and laryngeal paralysis.

Previously, heritability studies indicated that genetics accounts for 20% of the RLN trait in Thoroughbreds (Ibi et al., 2003). A genome-wide association study (GWAS) in Thoroughbred horses highlighted a single locus for RLN susceptibility on ECA3 previously associated with height in horses (Boyko et al., 2014). Height is a risk factor for RLN, but not all loci that contribute to height also confer risk to RLN, as demonstrated by another large GWAS in the Belgian breed (Brooks et al., 2018).



Paralysed arytenoid cartilage

Normal larynx

Figure 7.4. The larynx of two horses seen through an endoscope. The horse experiencing RLN (image left) and normal larynx (image right).

Conformation

For centuries, horses have been evaluated based on their conformation. Conformation is defined as the degree of physical correctness regarding the skeletal and muscular framework of the horse (Van Weeren and Crevier-Denoix, 2006). Serious consideration is given to conformation when horses are evaluated for purchase. Poor conformation can significantly reduce the value of an animal. Conformation is commonly believed to influence susceptibility to injury and can either enhance or limit movement. Weak or crooked legs can be a sign that the horse is not going to stay healthy, particularly when such horses begin hard work. In biomechanical terms, better conformation will result in a more efficient animal. Simply put, horses will find it easier to do their work which puts less stress and strain on muscles, bones, tendons, ligaments and joints so horses will likely stay sounder for longer (Pilliner et al., 2009). Limb injuries are the top cause of early retirement of equine performance horses (Ducro et al., 2009).

Depending on the discipline, breeders favour certain characteristics. In all performance horses there are certain features that are preferred, which are generally considered “correct” conformation. Some examples include horses having balanced body proportions, straight limbs, and proper angles between important structural bones. The proportions and angles of the bones have the capacity to limit the horse’s range of motion. This will negatively impact performance. Figure 7.5 and Figure 7.6 illustrate examples of poor conformation.

Can conformation be used as a predictor of performance?

The characteristics that breeders advocate in their horses varies depending on the discipline for which the horse will be competing. As an illustration, distance racehorses generally have longer legs, powerful muscles, and light bodies, while dressage horses can be of a slightly heavier build while still maintaining gracefulness (Love et al. 2006). Much of the research on conformation to date has focused primarily on Thoroughbred and Warmblood horses.

Over a 7-year period, Love et al. (2006) evaluated conformation records of 3,916 Thoroughbred yearlings. In the study, conformation of all horses was evaluated using a fixed routine by a single veterinary observer. Most of the horses raced in the UK in flat races (72%) while 7% of the yearlings failed to race. Conformation defects in the sample of 3,916 Thoroughbred yearlings examined as well as heritability estimates are in Table 8.3. The most common recorded defect was toed-out feet (30%). The study found that there was a tendency towards a greater proportion of horses with defects in the group of unraced horses compared with horses that raced; however, this result was not statistically significant.

Significant associations between racing performance and conformational defects were found to be almost completely explained by an effect of sire. All of the conformational traits showed considerable evidence of genetic influence. The highest heritable trait was back-at-the-knee (i.e., 66%). In conclusion, the study found only weak associations between performance and conformation which could not be accounted for by the very strong relationship between pedigree and conformation.

Table 7.3. Conformation defects in the sample of 3,916 Thoroughbred yearlings examined as well as heritability estimates (Love et al., 2006).

Conformation trait	Prevalence	Heritability
Toed-out	30%	0.16
Toed-in	19%	0.17
Upright pasterns	18%	0.18
Base Narrow	13%	0.16
Offset knees	12%	0.42
Weak pasterns	6%	0.31
Weak hocks	5%	0.38
Back-at-the-knee	4%	0.66

Weller et al. (2006) investigated the relationship between conformation, injury and performance in 108 National Hunt racehorses used for racing over fences during a two year period. Medical and performance data of all horses in the study were recorded. Conformation was measured in 3D using a computerised motion analysis system. Weller et al. (2006) found that the only conformational parameters associated with increased earnings or placings in races were angle of hip, shoulder and width of the mandible (jaw).

Santschi et al. (2006) evaluated 119 Thoroughbreds monthly from 1 month old to 120 days and 70 of these every second month until yearlings. Results found that 94% had carpal valgus (knock-kneed) at 1week old, only 8% carpal valgus by 362 days, 56% of fetlocks defined as straight at 1 week but 79% were straight at 525 days. The study concluded that slight carpal valgus in foals is normal and naturally corrects over first year of life.

Ducro et al. (2009) investigated the heritability of uneven feet and its relationship to sports performance in the Dutch warmblood. Heritability estimates of foot conformation traits were moderate and ranged from 0.16 for heel height to 0.27 for hoof shape. The authors concluded that predisposition to uneven feet can be reduced by selection.

Breeders should learn to discern between severe and minor conformational faults. The best way to achieve this is to evaluate lots of different horses. Get a picture of “correct horses” and evaluate conformation in a routine, repeatable manor. The functionality of the cardiovascular system as well as other physiological traits will have an obvious link to performance. Nevertheless, independent of the discipline or training programme, the overall conformation of the horse will affect the performance ability of horses to a certain degree. The angles of bones and proportions have the capacity to limit range of motion, negatively impacting the performance of the horse.

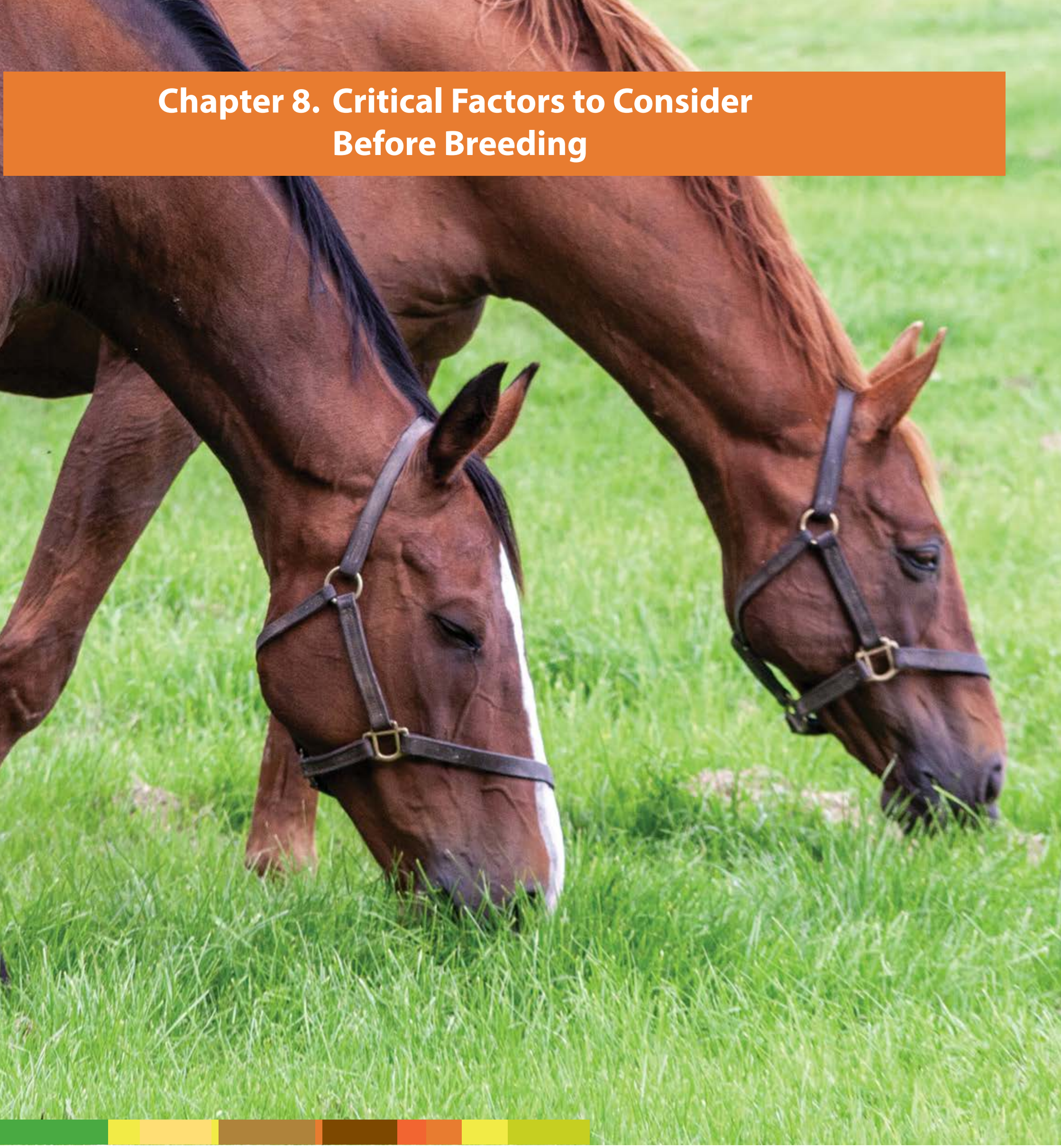
With all conformation studies, it is important to realise that athletic potential is difficult to standardise and performance depends on many factors apart from conformation (Weller et al., 2006). Several research studies on various breeds have found no single conformational trait having good predictive value for performance. It is possible for breeders to select against offset knees and back-at-the-knee conformation; and to a lesser extend against other faults (Table 8.3).

Conformational defect (forelimb) **Post corrective surgery**

Figure 7.5. The forelimb of a foal with a conformational defect pre- (image left) and post- corrective surgery (image right).

Figure 7.6. Foal with carpal valgus (toed-out) deformity corrected using an extension. Foal after 6 months.

Chapter 8. Critical Factors to Consider Before Breeding



The choice of the mare and stallion for breeding can be an intricate and time-consuming process. Frequently, not enough emphasis is put into this selection. As a result, the market becomes flooded with oversupply of mediocre or poor quality stock. This is clearly evident when evaluating sales results where many horses fail to cover their production costs.

Performance criteria varies considerably with the individual breeder and the type of horse required to match their specific breeding goal. The traits required to breed show horses, performance horses and leisure horses differ greatly. Nonetheless, regardless of breeding direction, reproductive competence and the ability to produce healthy offspring with minimal danger to the life and well-being of the dam should also be of prime importance.

Evaluating mares

Investment in a quality broodmare is essential for the success of a breeding enterprise. Mares are selected for temperament, conformation, movement, veterinary soundness, performance, pedigree and progeny (if available). Before attempting to select a stallion for the mare, the breeder needs to ascertain the type of mare they have or want to purchase. It is essential that breeders are absolutely clear on their breeding goal. Preferably, the mare should demonstrate an ability to perform in the sport the breeder has chosen to breed for or be well related to other animals that demonstrate the performance attributes the breeder is looking to reproduce. This gives the breeder the best opportunity to be successful. For example, if the breeding goal is to breed international show-jumpers the mare should to be well related to several Grand Prix horses.

Prior to purchase of a breeding mare a reproductive soundness examination should be conducted by a veterinary practitioner. The mare must be fertile, otherwise attempting to breed from her could prove a very costly.

Sport horse breeders should consider attending mare studbook inspections to get an independent evaluation of the strengths and weaknesses of their mare. The inspections give the breeder an independent viewpoint on how their mare compare against the rest of the population based on her linear score results. The information provided, particularly in relation to the athleticism traits will give a clear indication regarding suitability of the mare to produce for various markets. This information can greatly assist in making future breeding decisions.

The mare selection checklist in Table 8.1 should be used by breeders before considering breeding from a mare. In addition, Table 8.2 can be used as a template for breeders to evaluate progeny at different life stages during their development. It is suggested that this should be done as a foal and again at 6 months, 12 months and 36 months.

Table 8.1. Mare selection checklist.

How well do you know your mare	Notes
What is the breeding goal for the mare?	
What market is one aiming to breed for?	
What age will the progeny be sold?	
Veterinary soundness - any reproductive or wind issues? If there were barren years why did they occur?	
Conformational soundness – has the mare any physical problems or unsoundness?	
Is conformation, movement and athleticism of the mare appropriate to the chosen breeding goal?	
Was the mare inspected? what are her conformational strengths and weaknesses?	
Has a veterinary practitioner examined radiographs of the mare?	
Has the mare a performance record and to what level?	
Is the dam line capable of producing the type of horse outlined in your breeding goal?	
Has the dam line produced a horse at that level before?	
How closely related is the mare to that performance line?	
How many of the mare's siblings have already competed at the level your targeted breeding goal is aimed?	
Any temperament issues?	
*What is the breeding value of the mare (if available)?	
*What is the reliability of the breeding value (if available)?	

*See Irish Sport Horse Studbook Genetic Evaluation Report 2020:
horsesportireland.ie/wp-content/uploads/2020/12/final-genetic-evaluation-report-2020.pdf

Table 8.2. 5 minute horse evaluation scoresheet. Minimum assessment at different stages during development.

Horse Name:		**Score**	**Age (months)**		
			Foal	**6**	**36**
Height		S – XL			
Breed and sex character		1 – 10			
Health		1 – 10			
Balance		1 – 10			
Structure					
	Conformation	1 – 10			
Movement					
	Walk	1 – 10			
	Trot	1 – 10			
	Canter	1 – 10			
	Carefulness	1 – 10	_____	_____	
	Flexibility	1 – 10	_____	_____	
	Scope	1 – 10	_____	_____	
	Technique	1 – 10	_____	_____	
Muscling		1 – 10	_____	_____	
Temperament		1 – 10			
Overall		1 – 10			
Final remarks					

Breed and sex character: how well a horse represents its particular breed and sex (i.e., ‘type’).

Health: Is the horse physically healthy.

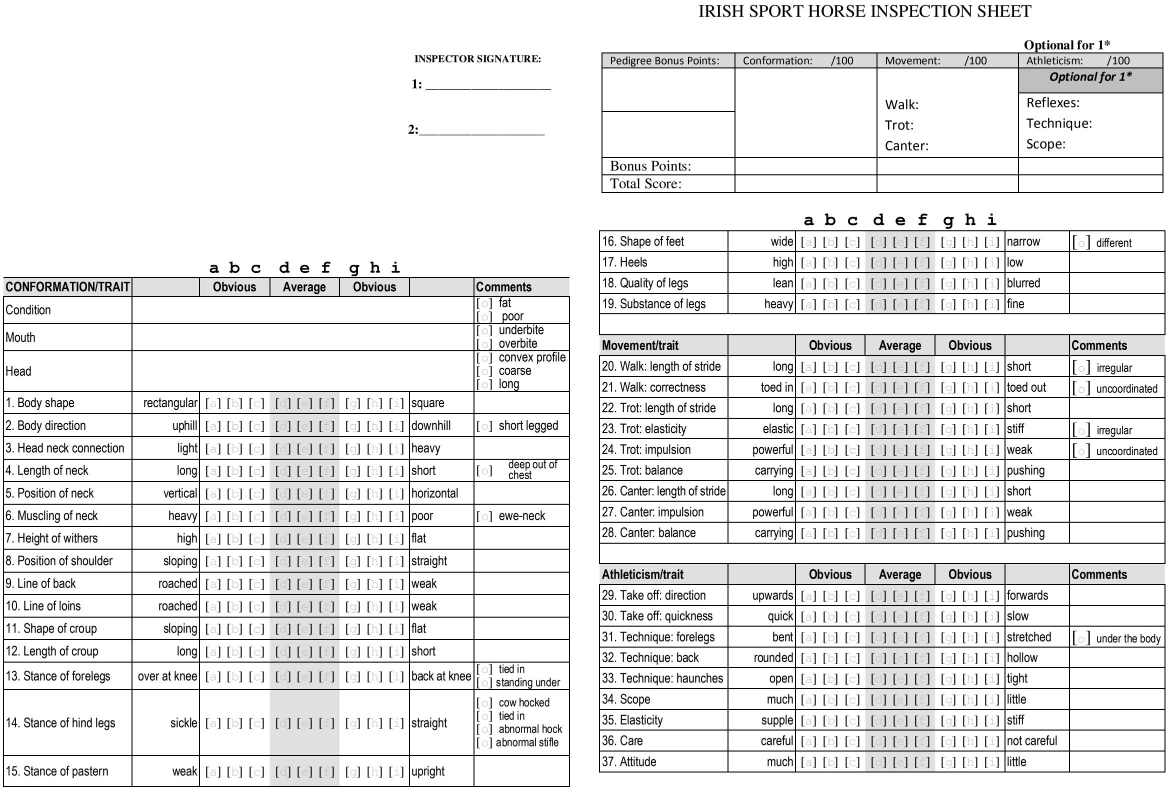
Balance: Is the horse in proportion.

Structure: Any conformational faults.

Movement: Enough athlete ability for chosen breeding goal. Dose the horse move economically.

Muscling: Quantity and quality of muscling.

Irish Sport Horse inspection sheet



IRISH SPORT HORSE INSPECTION SHEET

Optional for 1*

Pedigree Bonus Points:	Conformation: /100	Movement: /100	Athleticism: /100
			Optional for 1*
		Walk: Trot: Canter:	Reflexes: Technique: Scope:
Bonus Points:			
Total Score:			

INSPECTOR SIGNATURE:

1: ____________________

2: ____________________

		a b c	d e f	g h i		
CONFORMATION/TRAIT		**Obvious**	**Average**	**Obvious**		**Comments**
Condition						[o] fat [o] poor
Mouth						[o] underbite [o] overbite
Head						[o] convex profile [o] coarse [o] long
1. Body shape	rectangular	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	square	
2. Body direction	uphill	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	downhill	[o] short legged
3. Head neck connection	light	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	heavy	
4. Length of neck	long	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	short	[o] deep out of chest
5. Position of neck	vertical	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	horizontal	
6. Muscling of neck	heavy	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	poor	[o] ewe-neck
7. Height of withers	high	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	flat	
8. Position of shoulder	sloping	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	straight	
9. Line of back	roached	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	weak	
10. Line of loins	roached	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	weak	
11. Shape of croup	sloping	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	flat	
12. Length of croup	long	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	short	
13. Stance of forelegs	over at knee	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	back at knee	[o] tied in [o] standing under
14. Stance of hind legs	sickle	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	straight	[o] cow hocked [o] tied in [o] abnormal hock [o] abnormal stifle
15. Stance of pastern	weak	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	upright	

		a b c	d e f	g h i		
16. Shape of feet	wide	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	narrow	[o] different
17. Heels	high	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	low	
18. Quality of legs	lean	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	blurred	
19. Substance of legs	heavy	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	fine	
Movement/trait		**Obvious**	**Average**	**Obvious**		**Comments**
20. Walk: length of stride	long	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	short	[o] irregular
21. Walk: correctness	toed in	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	toed out	[o] uncoordinated
22. Trot: length of stride	long	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	short	
23. Trot: elasticity	elastic	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	stiff	[o] irregular
24. Trot: impulsion	powerful	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	weak	[o] uncoordinated
25. Trot: balance	carrying	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	pushing	
26. Canter: length of stride	long	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	short	
27. Canter: impulsion	powerful	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	weak	
28. Canter: balance	carrying	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	pushing	
Athleticism/trait		**Obvious**	**Average**	**Obvious**		**Comments**
29. Take off: direction	upwards	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	forwards	
30. Take off: quickness	quick	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	slow	
31. Technique: forelegs	bent	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	stretched	[o] under the body
32. Technique: back	rounded	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	hollow	
33. Technique: haunches	open	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	tight	
34. Scope	much	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	little	
35. Elasticity	supple	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	stiff	
36. Care	careful	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	not careful	
37. Attitude	much	[a] [b] [c]	[d] [e] [f]	[g] [h] [i]	little	

Details on the Irish Sport Horse studbook selections can be found by scanning the QR code.

SCAN ME

Evaluating stallions

The breeder needs to obtain as much information as possible before committing to a stallion. The mare's shortcomings should be compensated by using the correct stallion. Stallions are selected for temperament, conformation, movement, veterinary soundness, performance, pedigree and progeny. In addition, the stallion must have good fertility to enhance 6the mare's chances of conception.

The stallion selection checklist in Table 8.3 should be used by breeders before committing to using a particular stallion. To fully evaluate stallions, they and their progeny, should be viewed if possible. Alternatively, breeders are encouraged to view available videos (e.g., YouTube). In addition, the stallion's offspring should be viewed at competition or alternatively using video analysis. Breeders should also speak to the riders of the stallions where possible.

Table 8.4 illustrates the Irish Sport Horse studbook classification for stallions. The approved Irish studbook stallions are screened to eliminate unsoundness where the stallion must meet veterinary requirements. Where stallions from abroad are used, it is essential that the sport horse breeders become familiar with the approval system in the relevant studbook of origin and researches as much information as possible on the particular stallion. Of particular interest should be the veterinary requirements of the approval system the stallion has been appraised under.

Teagasc estimates the average cost of producing a foal for auction to be in the region of €1,500 to €2,250 while the average cost of producing a three-year-old for auction can be €4,000 to €4,500. These figures can be highly variable and the final cost can be considerably higher. These estimates include basic costs of production but do not take account of stud fees, mare depreciation, professional training fees and barren years. Breeders need to critically access their breeding strategies. They need to remain critical of their mares, breed for a particular market and consider upgrading poorer breeding mares. In addition, breeders are encouraged to attend foal sales and young horse events to evaluate progeny from various sires.

Table 8.3. Stallion selection checklist.

How well do you know your stallion	Notes
• What is the studbook classification for the stallion (e.g., stage 1, stage 2 or fully approved)? What are the veterinary requirements of the approval system?	
• If fully approved what genetic tests has the stallion been screened for?	
• Is his conformation and athleticism appropriate to the chosen breeding goal?	
• If required, has the stallion enough bone and blood?	
• How will this stallion complement your mare?	
• Has the stallion any physical problems or unsoundness? evaluation of radiographs?	
• Have you checked with other breeders/owners or with auctions to see if any of his progeny have had unsoundness issues? or undesirable traits?	
• Have you evaluated his pedigree, own performance and the performance of his offspring (what level are they preforming at)?	
• Any temperament issues?	
• Semen quality issues?	
• *What is the breeding value of the stallion (if available)?	
• *What is the reliability of the breeding value (if available)?	

*See Irish Sport Horse Studbook Genetic Evaluation Report 2020: horsesportireland.ie/wp-content/uploads/2020/12/final-genetic-evaluation-report-2020.pdf

Table 8.4. Irish Sport Horse studbook classification for stallions.

Approved (A)	*Stallions that have successfully met the criteria in all aspects of the veterinary inspection and inspection/performance requirements * Stallions are awarded star ratings based on their own performance or their progeny performance
Preliminary Approved (PA)	*Stallions that have met the veterinary & Stage 2 inspection requirements are classified as PA until they reach a max. 10 years old.
Not Approved (NA)	**NA1 -** *Stallions have met the veterinary inspection requirements but have not met the inspection/ performance requirements **NA2 -** *Stallions that have not met the veterinary requirements

Chapter 9. Frequently Asked Questions

Common questions submitted from Thoroughbred and sport horse breeders.

Q Can a chestnut mare have a bay foal and vice versa and how does grey comes about?

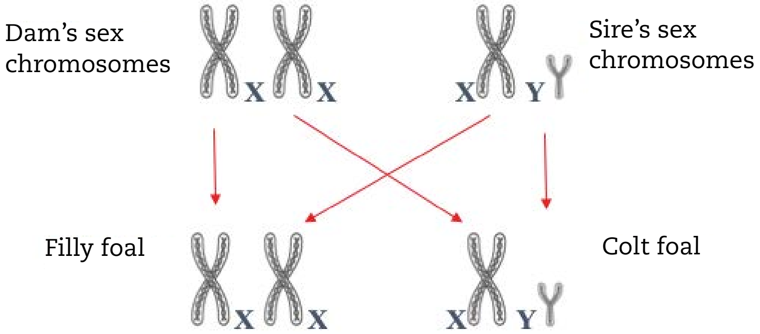
Coat colour in horses is influenced by a number of genes. The chestnut coat colour arises if a horse inherits two copies of the recessive version of the red (or chestnut) coat colour gene. Therefore, both parents must be carriers of the recessive version of the gene. That means they could both be bay (but carriers of the red gene version) or they may be chestnut themselves.

The grey gene on the other hand is dominant. This means that a grey horse must have at least one parent that is grey. If two grey horses have only one copy of the grey gene and are carriers of the red gene then there is a 25% chance that these two grey horses could produce a chestnut foal.

Nonetheless, phenotypic descriptions can only provide an approximation of the genes involved to predict the colour outcome of different mating's. Two horses with the same coat colour could have a different genotype. As a result, to accurately describe the alleles carried by an individual horses a DNA-based test for the colour genes should be used. These tests will describe the alleles carried by an individual horse, and therefore the potential to pass these alleles on to offspring.

Q How is sex of the foal inherited?

In mammals the method of inheritance of sex is purely down to a pair of sex chromosomes ‘X’ of ‘Y’. The Y chromosome are much smaller chromosomes and do not have as much genetic material as the X chromosome. Offspring that inherits an X and Y chromosome will be male and the offspring that inherits two X chromosomes will be female. In essence, every female only carries X sex chromosomes whereas every male will carry an X and Y chromosome; therefore it’s typically the father that determines the sex of the offspring. It’s purely random if 50% of the sperm will have X chromosomes and 50% will have Y chromosomes. It’s totally random whether the X or Y chromosome fertilises the ovum.



Q Why does one stallion ‘stamp’ their stock more than others?

Breeders often refer to a dominant sire as one who stamps his stock. This refers to a stallion that gives their physical resemblance to their progeny. The correct genetic reference is the termed dominance. Some horses can transmit certain characteristics to their offspring at a higher frequency than typically encountered with other breeding animals. Coat colour is a good example. A stallion whose offspring always or nearly always match the colour of the sire is described as a dominant sire. For some of the effects considered the correct terminology is not dominance but epistasis or homozygosity. An example can be a sire who is homozygous for grey coat colour. As a result, regardless of the colour of the mare every foal from that sire will be grey (white being the only possible exception) (Bailey and Brooks, 2013). Breeders need to remember that homozygous for colour does not mean that this is linked with the transmission of genes for other traits (e.g., jumping ability, conformation etc.). Breeders need to keep in mind that the mare is equally important in breeding and that no one stallion can be the perfect sire for every mares foal.

Q Would cloning the Tokyo Olympic gold medallist ‘Explosion W’ win another Olympic medal?

Cloning does not guarantee that the clone will be an exact replicate of the cloned animal. An Explosion W clone does not guarantee another Explosion W. This is because much of what makes up a horse isn’t down to the genetics but the environment. Some traits are influenced more by the environment than others (i.e., heritability). Traits that are lowly heritable will be affected more by variation in their environment (e.g., how the clone is raised and managed compared to the original) than their genetics. Examples of environmental variation include differences in nutrition, management and biological factors such as age and gender. As a result, it is unlikely that a clone will be identical from the animal which it was derived but would be genetically similar. The clone would have a greater chance of being a good performer but this can’t be guaranteed. Therefore, the chances of an Explosion W clone winning an Olympic medal are quite slim. Maybe this is beneficial as you can’t buy success just by cloning the best horses.

Cloning is beneficial to re-create a champion gelding like Explosion W with functional testes that would produce genetically identical spermatozoa and thereby enable the champion Explosion W to breed after all. In many sport horse disciplines a large number of high-class competing animals are males who were castrated at a young age. These geldings win several major events, therefore, it would be logical they be selected for breeding the next generation of champions and cloning could allow this (Allen, 2005).

Q What is a ‘blood horse’ and how are percentages calculated by HSI?

Breeders often quote “blood percentage” when evaluating horses. The percentage of blood is not a unit of inheritance. The percentage of blood is not an exact proportion, but an estimate. See example below, the offspring Thoroughbred blood percentage is predicted to be 62.5% (i.e., 50% of the offspring’s genes come from the father and 50% from the mother).

$$\frac{\text{(Sire 37.5\% Thoroughbred)}}{\text{(Dam 87.5\% Thoroughbred)}} = \text{Offspring 62.5\% Thoroughbred}$$

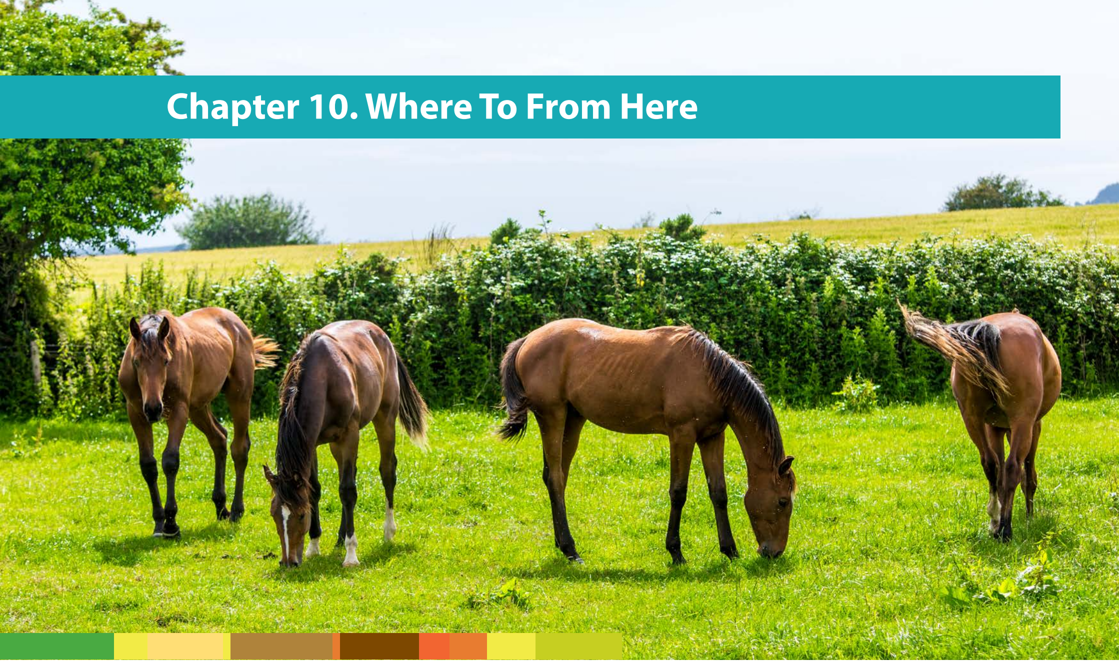
Breeders need to consider the phenotypic expression of ‘blood’. The progeny could be 62.5% Thoroughbred, however the offspring might not express their ‘percentage of blood’. Traditionally, in Ireland horses were bred by crossing an Irish Draught mare with a Thoroughbred sire.

The offspring in some cases however can reflect their Irish Draught ancestry more than their Thoroughbred ancestry. Therefore, the genotype is important but the expression of the phenotype is also very important. The athletic expression of a blood horse should be defined. Some breeders define a blood horse as a horse that should have energy, stamina, toughness, courage and quick reflexes in their athletic expression.

Q What is the probability that OCD chips in my Warmblood mare will be inherited by her offspring?

Heritability measures how much of the observed phenotype (OCD chips) independent of management are due to genetics. Some traits are highly heritable and some are not at all, but most fall somewhere in between. This means that the likelihood of a mare passing on defects to her progeny will depend on 1) the heritability of the trait and 2) whether the mare is a carrier of risk alleles. Research suggests that the heritability estimates for OCD in Warmblood horses ranged from 10% to 40%. This means that OCD is heritable and there is a moderate chance that the offspring will inherit this condition or be a carrier.

Chapter 10. Where To From Here



Breeding horses is competitive and can command a significant financial investment. The evaluation of performance has relied mostly on pedigree evaluation and observation of the physical characteristics of the horse. This has resulted in a lack of consensus among horse trainers, breeders, and owners about the most appropriate and informative scientific methods to select high genetic merit horses.

The sequencing of the horse genome has been the single greatest contribution to equine research. Before the genomics era, Mendelian genetics and statistics were used to predict genotypes and make general breeding recommendations. However, with the advent of molecular genetics it is possible to identify the DNA base changes responsible for the most valuable traits. With the discovery of genomics researchers can now identify specific genes of causation. Genomics has changed everything from human health to production of both crops and livestock that make up our food. It is going to greatly impact the equine industry in the future.

Genomic studies are rapidly increasing our knowledge of which genes are contributing to athletic performance. This growing field of research will lend to improved breeding strategies, training regimes and tailored nutrition (i.e., nutrigenomics) that may be optimised for individual horses; the unique genetic make-up of horses can affect their response to training and dietary nutrients. As a result, risks will be minimised while horses can be given the best opportunity to reach their genetic potential. Over the coming years, the list of genes contributing to performance is likely to increase as more sophisticated equine genomics resources become available. This will greatly increase our knowledge of the genes and molecular mechanisms underpinning athleticism and exercise adaptation.

Genomic selection approaches, in combination with reproductive technologies, have revolutionised the design and implementation of breeding programmes in other livestock species, particularly in cattle. Genomic selection has enormous future potential to improve the genetics of horses given their long generation interval. Once more genomic data is available breeders will likely have the opportunity to select horses based on their genomic breeding values.

Inbreeding and diversity

Breeders are understandably concerned about the existence of genetic diversity and inbreeding. As breeders select for a very precise phenotype, they increase the proportion of beneficial alleles at the expense of other potentially favourable alleles. If a genetic variant occurs in only one individual, or in only a few individuals, and by chance that allele is not passed on to the next generation, then the allele will no longer exist in that breed. With genomics, it is possible to monitor inbreeding levels and genetic diversity for several breeds.

Genetics of complex traits

There is currently no inexpensive way to discover genes for complex traits in the horse. Complex traits such as performance, conformation, and diseases are often caused by several genes interacting with the environment. Jumping and racing ability involves the respiratory system, muscle, heart, and mental aspects, but also requires very good nutrition and training. Major genes that contribute to racing performance have been discovered. Nonetheless, funding is required to study many other complex relationships.

Summary

Horses have been selected over time for a number of traits such as speed, jumping performances, strength or for more conventional traits like morphology or temperament. Worldwide, many horse breeds have been selected for preserving and improving traits of interest for sport performance or work. Novel traits such as fertility, longevity and health have been recently included or should be included in breeding decisions. One of the biggest obstacles of genetic improvement with horses is the slow rate of progress due to the long generation interval. Breeding younger stallions and mares (using AI, ET and ICIS for sport horses) would increase genetic gain by reducing the generation interval. Significant genetic advances can be achieved in the coming years. Identifying genes for performance at the molecular level is proving complex. Specifically, most traits are likely to be the product of a large number of genes plus environmental influences. It only makes sense that performance in horses is the product of genes affecting several areas. In conclusion, breeders should always remember that genetics create the potential but management can realise or destroy that potential.

Glossary

Allele	An allele is a variant of a gene. There can be multiple alleles for a gene and individuals will carry 2 alleles.
Ancestor	Any individual from which an animal is descended.
Autosomes	All chromosomes apart from sex chromosomes.
BLUP	Best Linear Unbiased Predictor (BLUP), is a statistical procedure for estimating breeding values. BLUP disentangles genetic effects from environmental effects (training, nutrition, age etc.) and produces predictions of breeding values.
Breeding value	A prediction of the average genetic merit of an individual as a parent.
Chromosome	Structures located in the nucleus of all cells, which are made up of DNA, and on which genes are located. In the nucleus chromosomes occur in pairs but occur singly in gametes (sperm and eggs). Of the 64 chromosomes found in each cell of a healthy horse, 32 come from the sire and 32 come from the dam. One chromosome can contain hundreds and even thousands of genes.
Covariance	Degree to which two measurements vary together.
Crossbreeding	Matings between horses of different breeds or lines.
Deletion	A mutation involving a loss of DNA; either one or a number of base pairs or a chromosomal fragment.
DNA	The chemical structure which chromosomes and genes are made of. Molecules of DNA are made up of two strands with cross linkages made up of chemical substances called bases (adenine, thymine, guanine and cytosine).
Diploid	The term for an organism's genome where chromosomes are organised in pairs, horse's diploid number is 64 from 32 pairs of chromosomes.
Dominance	Only one copy of the mutation is needed for it to be expressed outwardly. A dominant allele will always express even if a recessive allele is present.
ET	Embryo Transfer (ET) involves the removal of an embryo from the uterus of one mare, and the transfer of that embryo into the uterus of another mare.

Epistasis	Occurs when the presence of an allele at one locus masks the effect of an allele at another locus (i.e., there is an interaction of alleles at different loci). An example include interactions between alleles at different coat colour loci in horses.
Gamete	Sex cell: an unfertilised egg (ovum) in the female, and a sperm (spermatozoa) in the male.
Gene	A unit of heredity that provides instructions as to what the animal will look like, how it will perform etc. Genes are strung together in long strands of DNA called chromosomes.
Genetic correlation	Genetic correlations measure the strength of the linear relationship between two traits x and y that is due to genetics. Genetic correlations occur due to pleiotrophy or linkage among genes.
Gene expression	The functional relationship between deoxyribonucleic acid (DNA), ribonucleic acid (RNA), and protein.
Generation interval	The mean age of the parents when the progeny are born.
Genetic evaluation	The prediction of breeding values
Genome	The full complement of genetic information for an individual.
Genotype	Combination of genes which the horse inherits (e.g., Aa or AA).
Genetic variation	The variation or differences within a population that are due to differences in genetic merit of the animals.
Heritability	Proportion of phenotypic variation, or differences among a cohort of animals, attributable to genetic variation between individuals.
Heterosis	The degree to which the performance of a crossbred animal is better or worse than the average performance of the parents.
Heterozygous	Horses that carry two different alleles at a particular locus (e.g., Ab).
Homozygous	Horses that carry two copies of the same allele at a particular locus (e.g., AA). Can be homozygous dominant or homozygous recessive.

ICSI	Intracytoplasmic sperm injection (ICSI) involves micro-injection of a single sperm cell into the cytoplasm of a mature oocyte, which physically causes fertilization. The fertilised oocyte is returned to an incubator and allowed to develop into an embryo, which usually occurs within 6-8 days. After, the embryo can be implanted into a recipient mare.
Inbreeding	Involves mating closely related individuals. Can be done deliberately (e.g., line-breeding) or can result from long-term selection in closed populations.
Inbreeding depression	Reduced performance normally associated with increased levels of inbreeding. Many recessive genes result in undesired traits or reduced performance when expressed.
Karyotype	The complete set of chromosomes is known as a karyotype.
Line-breeding	Deliberately mating closely related individuals. Breeders use line-breeding to cross to an exceptional individual.
Linkage	Association of genes that are physically located on the same chromosome.
Locus	The site of the gene on a chromosome.
Marker	Specific sequences of the DNA molecule. The markers might or might not be functional genes.
Mutation	Random change in DNA structure.
Nucleus	The cellular organelle which contains all the genetic information of an organism.
Nutrigenomics	The interaction between genes and nutrition; how an individual's unique genetic make-up affects their response to dietary nutrients. The ultimate goal is to optimise equine health through the personalisation of diet.
Over-dominance	Where the heterozygous phenotype lays outside of the two homozygous phenotypes.
Pedigree	Diagram illustrating the relationships between members of an extended family. The diagram can also illustrate an inheritance pattern for a specific trait.
Punnett square	Square diagram which can be used to predict the genotypes of a particular cross.
Phenotype	The observed performance of an animal "in the field" (e.g., coat colour, height); it's the physical expression of the animal's genotype.

Pleiotropy	Pleiotropy occurs when an allele at a single locus influences more than one trait of the animal.
Recessive	An allele that will always be covered up by a dominant allele if present, is represented by lower case letters (e.g., aa).
Selection criteria	The character(s) upon which selection decisions are based.
Selection Index	The combining of measurements from several sources into an estimate of genetic value. More than one measurement on a trait, and/or measurements of the trait on relatives, and/or the measurements on more than one trait are combined into a single estimate of overall genetic value.
Selection objective	The character(s) which are intended to be modified by selection.
Zygote	The cell produced by the fusion of mature gametes (egg and sperm).

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Images

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Appendix

www.teagasc.ie

https://www.teagasc.ie/rural-economy/rural-development/equine/

www.horsesportireland.ie

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