

Genetic Screening Tests available		
Symbol	Name	Nature of Characteristic or Disease
Equine Colour and Pattern Testing		
AG	Agouti (black pigment distribution)	The Agouti gene controls the distribution of black pigment in a horse's coat. Black pigment can either be uniformly distributed (allele a) or it can be restricted to the points of the body eg. Bay (allele A). Two copies of the "a" allele (a/a) are required for uniform black pigment distribution.
CCC	Chestnut/Red Factor/Extension	Detects the two alleles of the MC1R gene that affect the amount of black or red pigment and so determine the base coat colour of chestnut or bay/brown/black.
CD	Cream Dilution	Detects the mutation in the MATP gene responsible for the palomino, buckskin, smoky black, cremello, perlino and smoky cream coat colours.
CHP	Champagne Dilution	Champagne is a coat colour dilution distinct from the Cream, Silver and Pearl Genes. Colours range from gold, amber, sable or classic champagne depending on the base colour of the horse. Champagne dilution is linked to a single base pair change in the SLC36A1 gene.
DW(W1-22)	Dominant White	Dominant white is one of several known depigmentation phenotypes in horses. Considerable phenotypic variation is seen ranging from completely white, to white spotting and white marking from birth. Skin is usually pink and eyes are dark. A series of 21 different mutations in the Kit gene (designated W1-W22) have been associated with white patterning/white coat colour in horses. Many of these mutations have been discovered in specific family/breeding groups and are not widespread throughout a particular breed. W10 for example is found in Quarter horses descended from the lineage of a stallion called GQ Santana. W20 was found to occur in a number of horse breeds but appears to have only a subtle effect on pigmentation – examples seen were a blaze and white socks. When W20 is combined with another W mutation, W5, more severe effects on pigmentation were observed. However the W5 mutation as only been observed in a specific family of Thoroughbreds and how widespread W5 and any of the other W mutations are in the Australian horse breeds/families is unknown at this stage. The AEGRC

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		can test for any of the known W mutations but please contact us for further information as to whether testing would be relevant/beneficial for your breed/horse.
GR	Grey	Grey is a coat colour mutation where horses are born with a darker coat colour but grey over a comparatively short time, eventually becoming completely white. The grey phenotype is caused by a duplication in the STX17 gene.
LP	Leopard Complex/Appaloosa Spotting (Congenital Stationary Night Blindness, CSNB)	Leopard complex spotting (LP), also known as Appaloosa spotting, describes a group of coat pattern phenotypes. The pattern is often symmetrical and ranges from a few white patches on the rump of the animal to animals that are almost completely white. Homozygosity for the LP mutation has also been associated with Congenital Stationary Night Blindness (CSNB), where the horse has impaired vision in dark conditions from birth. Leopard complex spotting has been associated with a DNA insertion in the TRPM1 gene.
OLWFS	Overo Lethal White Foal Syndrome	Overo lethal white syndrome (OLWS) is an inherited syndrome of foals born to parents of the overo coat-pattern lineage. Affected "O/O" foals are totally or almost totally white and die within days. OLWS is associated with a two base pair change in the EDNRB gene. Heterozygous "N/O" horses remain healthy and unaffected but are carriers and display patches of white coat colour known as "frame overo".
PRL	Pearl Dilution	Pearl is a rare dilution phenotype usually only seen in Quarter horses and Spanish horse breeds. This dilution phenotype is also known as "Barlink Factor". Horses carrying two copies of pearl will have a lightened coat, mane and tail, in addition to bright eye colours due to pigment changes caused by the gene. Pearl is also known to interact with Cream dilution to produce pseudo-double Cream dilute phenotypes. Pearl has been linked to a single base change in the SLC45A2 gene.
SAB1	Sabino1	Sabino describes a group of similar white spotting patterns, usually involving irregular spotting on the legs, belly and face with extensive roaning. Sabino1 patterning is associated with a particular single base

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		change in the KIT gene. Sabino1 is not the causative DNA change for all sabino-patterned horses.
SIL	Silver Dilution	The silver dilution gene dilutes black pigment in a horse's coat, but has no effect on red pigment. Horses with a black base colour will be diluted to chocolate coat colour with flaxen mane and tail while horses with a bay coat colour will have lightened black pigment on the lower legs with flaxen mane and tail. The silver dilution trait has been linked to two mutations in the PMEL17 gene.
SW1	Splashed White 1	<p>The Splashed White group of coat phenotypes in horses is variable, but is usually characterised by a large white blaze and blue eyes. The white markings may also extend to the rest of the head and neck, the legs and the underbelly. The Splashed White phenotype is also occasionally associated with congenital deafness, though the links are not well understood.</p> <p>Three different mutations have been identified as being associated with the Splashed White coat pattern. They are named SW1, SW2 and SW3</p> <p>SW1 is associated with an insertion in the MITF gene. Breed distribution of SW-1 includes Quarter Horse, Paint, Trakehner, Miniature Horse, Shetland Pony and Icelandic Horse and may be present in other breeds as well.</p>
SW2	Splashed White 2	SW2 is associated with a SNP in the PAX3 gene. Reported to only occur in certain lines of Quarter Horses and Paints.
SW3	Splashed White 3	SW3 is associated with a deletion in the MITF gene. Reported to only occur in certain lines of Quarter Horses and Paints.
TOB	Tobiano	Tobiano is a distinct white spotting pattern involving large defined white markings. Tobiano is associated with a large inversion on Chromosome 3.
Genetic Disease Testing		
CA	Cerebellar Abiotrophy	Neurological condition with head tremor, muscle weakness and lack of balance. CA has been linked to a

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Symbol	Name	Nature of Characteristic or Disease
		mutation in the TOE1 gene.
GBED	Glycogen branching enzyme deficiency	Metabolic genetic disease that is fatal in foetal and neonatal stages. GBED has been associated with a mutation in the GBE1 gene.
HERDA	Hereditary equine regional dermal asthenia	Severe skin blistering and lesions leading to secondary infections and early death. HERDA has been associated with a mutation in the PPIB gene.
HWSD	Hoof Wall Separation Disease	Inherited disorder characterised by separation and breaking of the dorsal hoof wall – appears to occur only in the Connemara Pony. HWSD is associated with a DNA insertion in the SERPINB11 gene.
HYPP	Hyperkalemic Periodic Paralysis	Potassium induced intermittent paralysis. HYPP is associated with a mutation in the SCN4A gene.
JEB1	Junctional Epidermolysis Bullosa (Draft Horses)	Skin and mouth lesions leading to secondary infections and death at 1-2 years. JEB in draft horses is associated with a DNA insertion in the LAMC2 gene.
JEB2	Junctional Epidermolysis Bullosa (Saddlebreds)	Skin and mouth lesions leading to secondary infections and death at 1-2 years. JEB in Saddlebreds is associated with a DNA deletion in the LAMA3 gene.
LFS	Lavender Foal Syndrome	Neurological abnormalities and neonatal death. LFS has been associated with a DNA deletion in the MYO5A gene.
MH	Malignant Hyperthermia	Disorder of skeletal muscle leading to tying up, elevated body temperature and adverse reactions to anaesthetic. MH has been associated with a mutation in the RyR1 gene. This mutation has also been found to increase the severity of PSSM when both the MH and PSSM1 mutations are present in the same horse.
PSSM1	Polysaccharide Storage Myopathy	Chronic exercise induced muscle breakdown (exertional rhabdomyolysis). Severity modified by other genes and environmental factors. PSSM1 is associated with a mutation in the GYS1 gene.
SCID	Severe Combined Immune	Disease where the immune system is severely weakened,

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	Deficiency	affected foals have an inability to fight off infections and die around 4-7 months of age. SCID is associated with a DNA deletion in DNA-PK.
Other Testing		
AME and SRY	Genetic Sex Determination (Note. This is <u>not</u> a karyotyping test).	Amelogenin detects the presence of the horse X and Y chromosomes, determining the genetic sex of an individual. SRY is used for ambiguous sex determination cases.

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