Genetic Screening Tests available		
Symbol	**Name**	**Description of variant effects**
Equine Colour and Pattern Testing		
E	Red Factor/Extension (Formerly CCC)	Detects the two alleles of the MC1R gene which determines whether black pigment is expressed (horse is bay or black; E/-), or if the horse is chestnut (e/e).
A	Agouti (Formerly known as AG)	The Agouti gene controls the distribution of black pigment in a horse's coat. Note that agouti is only seen if the horse is not chestnut. Black pigment can either be restricted to the points (and the horse is bay; A/-) or is evenly distributed over the entire coat (horse is black; a/a).
Cr	Cream Dilution (Formerly known as CD)	Detects the mutation in the MATP gene responsible for palomino, buckskin, smoky black, cremello, perlino and smoky cream coat colours. If one copy of cream is detected (Cr/n), only red pigment is diluted and the horse is palomino, buckskin or smoky black, depending on its base colour. If two copies are detected (Cr/Cr), the horse is diluted to cremello, perlino or smoky cream.
W20	White spotting patterns 1 – 27 (formerly known as DW)	*** The EGRC will start with offering W20 first *** The W patterns are caused by numerous mutations in the KIT gene which causes white hairs over the coat. The distribution and pattern of white varies considerably, ranging from socks and face markings, white spotting covering the body, to a completely or nearly completely white coat. The skin is pink, but the eyes are brown. These patterns were previously known as Dominant White, but modern nomenclature uses W plus the number allocated to the known mutation. Many of the W mutations have been discovered only in specific families. W5, W10 and W20 are the most commonly tested W patters., although the EGRC can test for any of the known W mutations. Please contact us before ordering for further information to determine whether testing is relevant for your breed or line of horse.
G	Grey (formerly known as GR)	Grey causes accelerated loss of pigment in the hair. The foal is born a solid colour, then white hairs become mixed with coloured hairs around the body. Eventually the horse becomes all white. Rate of greying can vary significantly. Grey is also associated with the development of melanomas. Horses that are homozygous grey are more likely to develop melanoma earlier than heterozygotes. The melanomas are not cancerous; however, they can become large and obstructive. The grey phenotype is caused by a duplication in the STX17 gene.

Genetic Screening Tests available		
Symbol	**Name**	**Description of variant effects**
Lp	Leopard Complex/Appaloosa Spotting (Congenital Stationary Night Blindness, CSNB)	Leopard complex spotting (Lp), also known as Appaloosa spotting, describes a number of different spotting patterns. The pattern is often symmetrical and ranges from a few white patches on the rump to animals that are almost completely white. There may be progressive roaning of the coat with age. Mottled skin around the muzzle, eyes, coronets (causing striped hooves), genitalia and anus is characteristic. Some Appaloosa patterning is caused by the PATN gene working in conjunction with Lp. Lp/Lp horses have Congenital Stationary Night Blindness (CSNB) and are night blind. Leopard complex spotting is caused by a DNA insertion in the TRPM1 gene.
O	Overo Lethal White Foal Syndrome	A single O allele causes the "frame" or "frame overo" spotting pattern. Expression of white is highly variable, ranging from lots of white "framed" by the horse's base colour, to minimal or just a few white hairs on the belly. Overo Lethal White Foal Syndrome occurs when a horse is homozygous for the O mutation. These O/O foals are born almost or completely white, but do not have properly formed intestinal nerves and cannot pass faeces. They only survive a few days if not euthanised for compassionate reasons. Carrier horses (O/n) have no documented health issues. OLWS is associated with a two base pair change in the EDNRB gene. Because O can be minimally expressed, it is important to test any horse that might be a carrier even if it has little to no white on it, to prevent the birth of an affected foal.
D/nd1/nd2	Dun	Dun lightens the body, leaving the head, lower legs, mane and tail undiluted. It also causes a darker dorsal stripe, shoulder stripes, and sometimes leg barring and concentric marks on the forehead (known as 'primitive markings'). There are 3 important variations of the Dun gene. D/n causes dun dilution and primitive markings, nd1 does not cause dilution but there is some variable expression of primitive markings, and nd2 causes no-dilution and no primitive markings.
Sb1	Sabino1 (formerly known as SAB1)	Sabino is an older term used to describe a number of different spotting patterns. Sabino 1 is a specific mutation that causes (often high) white on the legs and belly, and a blaze. Roaning is generally evident, particularly at the edges of the white patterns. Homozygous horses (Sb1/Sb1) are almost completely, if not completely, white.

Genetic Screening Tests available		
Symbol	**Name**	**Description of variant effects**
SW1	Splashed White 1	SW1 is associated with an insertion in the MITF gene. The SW1 mutation has been identified in a number of breeds including Quarter Horse, Paint, Trakehner, Miniature Horse, Shetland Pony and Icelandic Horse; and it may be present in other breeds as well.
SW2	Splashed White 2	SW2 is associated with a SNP in the PAX3 gene. It has only been observed in certain lines of Quarter Horses and Paints.
SW3	Splashed White 3	SW3 is associated with a deletion in the MITF gene. It has only been observed in certain lines of Quarter Horses and Paints.
To	Tobiano (formerly known as TOB)	Tobiano is a white spotting pattern where the large white patches with clean edges extend across the spine. Legs often have high white and partial or entire blue eyes may be observed. Tobiano is associated with a large inversion on Chromosome 3.
Genetic Disease Testing		
CA	Cerebellar Abiotrophy	CA is a neurological disorder that affects the cells in the cerebellum, causing head tremors, ataxia and other effects. Affected horses are more likely to fall and are generally not safe to ride. Symptoms appear from 6 weeks to around 4 months old. CA has been linked to a mutation in the TOE1 gene and is a recessive disorder, meaning that a horse must be homozygous (CA/CA) to be affected. If a horse is a carrier (CA/n), it will not show any clinical signs of CA, but it will pass the variant on to approximately half its offspring. Mating to other carriers should be avoided to prevent the birth of an affected foal.
LFS	Lavender Foal Syndrome	LFS causes neurological dysfunction in foals. The symptoms include seizures, hyperextension of the limbs, neck and back, leg paddling, and inability to stand. As the name suggests, LFS also dilutes to the coat to a pale lavender pink or silver colour. The foal will not improve and will die, so it should be euthanised. LFS is a recessive disorder so two copies of the defective version of the MYO5A gene must be inherited (LFS/LFS) for a foal to be affected. If a horse is a carrier (LFS/n), it will not show any clinical signs of LFS. However, there is a 50% chance it will pass the variant to its offspring, so mating to other carriers should be avoided to prevent the birth of an affected foal. LFS is caused by a mutation in the MYO5A gene and is more frequently observed in (although is not limited to) Arabian horses with Egyptian heritage.

Genetic Screening Tests available		
Symbol	**Name**	**Description of variant effects**
SCID	Severe Combined Immune Deficiency	Foals affected by SCID lack a proper immune system which is critical for fighting infection. Once the maternal immune protection wears off, SCID foals develop signs of infection (e.g. fever, respiratory distress and/or diarrhea). SCID cannot be cured and affected foals will usually die from an infection before 6 months of age. SCID is a recessive disorder so two copies of the defective version of the DNA-PK gene must be inherited (SCID/SCID) for a foal to be affected. If a horse is a carrier (SCID/n), it will not show any clinical signs of SCID. However, there is a 50% chance it will pass the variant to its offspring, so mating to other carriers should be avoided to prevent the birth of an affected foal.
GBED	Glycogen branching enzyme deficiency	GBED is a lethal storage myopathy caused by a mutation in the GBE gene that prevents the foal from properly storing glucose. This means the horse will not have enough stored energy, which will eventually damage its organs. The symptoms observed are associated with the lack of energy preventing the organs from working correctly, and may include general weakness, low body temperature, seizures and difficulty rising. GBED is always fatal, with most affected foals dying before the age of 8 weeks. GBED can also cause foetuses to be aborted in utero. It occurs in Quarter horses, paint horses and related breeds and is inherited as a recessive trait, so only homozygous (GBED/GBED) horses are affected. If a horse is a carrier (GBED/n), it will not show any clinical signs of GBED. However, there is a 50% chance it will pass the variant to its offspring, so mating to other carriers should be avoided to prevent the birth of an affected foal.
HERDA	Hereditary equine regional dermal asthenia	HERDA is a skin disease found primarily in Quarter Horses. It is characterised by hyper-elastic skin which progresses to severe skin lesions, particularly along the back of the horse. The disorder affects the collagen that holds the skin in place, making it much easier to tear than normal. Any rubbing, such as that caused by saddling, will cause the skin to split so affected horses are unable to be ridden. The lesions are painful and prone to infection. HERDA is associated with horses from the Poco Bueno sire line. HERDA is caused by a mutation in the PPIB gene and is recessive, so the horse must be homozygous (HERDA / HERDA) to be affected. If a horse is a carrier (HERDA/n), it will not show any clinical signs of HERDA. However, there is a 50% chance it will pass the variant to its offspring, so mating to other carriers should be avoided to prevent the birth of an affected foal.

Genetic Screening Tests available		
Symbol	**Name**	**Description of variant effects**
HYPP	Hyperkalemic Periodic Paralysis	HYPP causes delayed muscle relaxation. Symptoms range from muscle twitches and weakness to severe muscle spasms, paralysis, respiratory noises, collapse and occasionally death, usually from cardio-respiratory effects. HYPP is a dominant trait, meaning a horse only needs 1 copy of the mutation (HYPP/n) to be affected. There is some evidence that homozygous horses (HYPP/HYPP) are more severely affected than heterozygotes. The severity and onset of symptoms can be managed with diet, particularly by avoiding high potassium feeds. HYPP is associated with Quarter Horses from the "Impressive" line and is caused by a mutation in the SCN4A gene.
PSSM1	Polysaccharide Storage Myopathy	PSSM1 causes a build-up of abnormal sugars in muscle. This is one of the causes of tying up, with clinical signs including muscle twitches, stiffness, sweating, reluctance to move and painful cramps. PSSM1 is found in many breeds including Quarter Horses and draft breeds. It can be controlled with changes to diet and other environmental factors. It is associated with a mutation in the GYS1 gene and is inherited in a dominant fashion, so a horse only needs to carry one copy (PSSM1/n) to be affected. There is some evidence that homozygous horses (PSSM1/PSSM1) are more severely affected than heterozygotes. Please note that this test only detects this one specific type of tying up, and horses may still exhibit signs of tying up even if they are not positive for PSSM1.
MH	Malignant Hyperthermia	MH is a muscle disorder that may only become apparent if the horse is subjected to an extreme stress or exposed to a halogenated anaesthetic. When exposed, the mutation triggers the release of excess calcium in skeletal muscle cells causing high temperature (hence the name), increased heart rate and blood pressure, sweating and muscle rigidity. MH sometimes occurs in horses which are also positive for PSSM1, causing them to have more severe tying up symptoms. MH is rare and only found in some Quarter Horse and paint families; however, because it is potentially fatal it is recommended all possible carriers be tested before undergoing anaesthesia. MH is associated with a mutation in the RyR1 gene and is a dominant trait, meaning a horse only needs 1 copy of the mutation (MH/n) to be affected.

Genetic Screening Tests available		
Symbol	**Name**	**Description of variant effects**
HWSD	Hoof Wall Separation Disease	Hoof Wall Separation Disease causes the hoof wall to easily crack and break. It is specific to Connemara ponies or horses carrying Connemara blood. In some ponies the disease is less severe, but in other cases they may need to be euthanised due to increasing pain and related infections. The symptoms can appear quite early in life and can affect all four feet. The disease is recessive, meaning only ponies that are homozygous (HWSD/HWSD) can be affected, although carriers are still capable of passing the mutation to their offspring. HWSD is caused by an insertion in the SERPINB11 gene.
Other Testing		
AME and SRY	Genetic Sex Determination (Note. This is not a karyotyping test).	Amelogenin detects the presence of the horse X and Y chromosomes, determining the genetic sex of an individual. SRY is a Y-chromosome specific marker used for ambiguous sex determination cases.
Genetic Screening Tests under development…. Coming soon!		
Symbol	**Name**	**Nature of Characteristic or Disease**
Equine Colour and Pattern Testing		
PATN1	Appaloosa Pattern-1	LP determines whether a horse will show leopard complex spotting, while other genes determine the amount of white shown. PATN1 is associated with increasing white in LP horses. If the horse is LP/N and PATN1/N, it will have a 'leopard' pattern. If the horse is LP/LP and PATN1/N, it likely has a 'few-spot' pattern.
Pan	Pangare	Pangare is the pattern observed on wild Przewalskis Horses. It is also known as 'mealy' and features pale hair around the muzzle, eyes and underside of the horse. The mutation thought to be responsible for Pangare has only recently been identified so this test will need some validation before we offer it commercially.
Genetic Disease Testing		
Symbol	**Name**	**Nature of Characteristic or Disease**
FFS1	(Warmblood) Fragile Foal Syndrome 1	FFS1 is a fatal skin disorder that causes the skin to be thin, hyper-extensible (fragile) and easily torn. Other clinical signs include swelling and haematoma, joint laxity and possibly abortions and premature births. Foals are severely affected and

Genetic Screening Tests available		
Symbol	**Name**	**Description of variant effects**
		are euthanised due to a poor prognosis for life. FFS1 is caused by a mutation in the PLOD1 gene and is autosomal recessive, so affected horses will have inherited the defective allele from each parent (FFS1/FFS1). If a horse is a carrier (FFS1/n), it will not show any clinical signs of FFS1. However, there is a 50% chance it will pass the variant to its offspring, so mating to other carriers should be avoided to prevent the birth of an affected foal. Whilst this disease is primarily recognised in Warmblood/Sport horses, the mutation has been seen at very low frequencies in other breeds.
OAAM	Occipitoatlantoaxial malformation	OAAM is a developmental defect where the first cervical vertebra is malformed and resembles the base of the skull. The second cervical vertebra resembles the first. This compresses the spinal cord near the base of the skull, causing neurologic effects. Symptoms vary from abnormal head carriage, reluctance to move, neck twisting, progressive incoordination and weakness, and inability to stand. There appears to be more than one mutation involved and there is a test available for only one of these.