

Genetic Disorders and the Importance of Testing (for purebred AND PARTBRED Arabian horses)

Genetic testing for SCID, LFS and CA is now available as a three panel test via Massey University for \$95, or \$35 per individual test. Genetic testing can be done at the same time as DNA for registration, or samples from older horses may be tested. Massey is able to use hair samples from registered horses that are already on file provided there are enough viable hairs. Please note there may be a time delay if there are not enough hairs on file, therefore, for live horses, it may be best to send a new hair sample.

The NZAHBS would like to record voluntary results in our online pedigree database (under construction); please send copies of results you wish to be recorded. For legal reasons, results will be only accepted from the current recorded owner or leasee. Once recorded they will not be removed from the database. Note: under WAHO rules, genetic results cannot be recorded on registration certificates or in physical studbooks.

Instructions: Pull approximately 40 hairs with roots, from tail or mane and enclose in envelope as for DNA. Fill out the Massey University submission form and post.

Horses, like other species such as dogs, cats and even humans, can be affected by a variety of genetic disorders. Currently, DNA testing is available for ten equine genetic disorders, with three of those disorders of particular interest to Arabian horse owners and breeders: Severe Combined Immunodeficiency (SCID), Cerebellar Abiotrophy (CA) and Lavender Foal Syndrome (LFS).

All three of these disorders are recessive traits; this means that in order for a foal to be affected with the disease, it must have received a copy of the gene mutation from each parent. Horses that have only one copy of the gene mutation are physically normal, but are still carriers and can pass the mutation along to their offspring. While the vast majority of Arabian horses are clear, and do not possess the

mutations responsible for these disorders in their genetic code, it is important to be aware that these genetic disorders do exist within the breed.

Owners and breeders now have the ability to test to determine if their horse is clear, carrier, or affected. Testing allows breeders to make informed choices, so the production of affected foals can be avoided.

Inheritance of Recessive Genetic Disorders

Two copies of the mutated allele (gene) are needed for expression

% Probability at each mating	Carrier stallion	Clear stallion
Carrier mare	50% Carrier 25% Clear 25% Affected	50% Carrier 50% Clear
Clear mare	50% Carrier 50% Clear	100% Clear

Severe Combined Immunodeficiency (SCID)

Severe Combined Immunodeficiency (SCID) is a lethal genetic disorder that results in an affected foal being born with a severely weakened immune system. Because the foal's natural defence system against infection is not functioning properly, they die at a very early age of opportunistic infections (such as pneumonia) or require euthanasia.

Cerebellar Abiotrophy (CA)

Cerebellar Abiotrophy (CA) is a neurologic genetic disorder which causes the degeneration of specialized nerve cells in the brain that are responsible for balance and coordination. Foals affected with CA may appear normal at birth. However, as the nerve cells begin to die (often by a few months of age, but can frequently occur later in life), the foal will begin to show signs which may include a head tremor, lack of balance, exaggerated movement of the limbs, a wide-based stance, sensitivity to sound and sudden movements, and falling. The range and severity of the physical signs of CA can vary widely. Affected foals are generally euthanized or restricted to life as pasture pets, as their lack of coordination presents a danger to themselves and others, and also prevents them from being ridden safely.

Lavender Foal Syndrome (LFS)

Lavender Foal Syndrome (LFS), also known as Coat Colour Dilution Lethal (CCDL), is a lethal neurologic genetic disorder. Affected foals are unable to stand and often have episodes of tetany where the foal will lay on its side rigidly extending its limbs, neck and back. Many of these foals are also born with a unique diluted coat color that may appear as pale lavender, pale pink or silver. Because these foals are often large and may have had a difficult birth, they can initially be mistaken as 'dummy foals' (Neonatal Maladjustment Syndrome). There is no treatment for LFS and affected foals will die quickly or require euthanasia.

Other Disorders

Although not yet testable, disorders such as Guttural Pouch Tympany (GPT), Juvenile Idiopathic Epilepsy (JIE) and Occipitoatlantoaxial Malformation (OAAM) are also thought to have a genetic basis.

Half-Arabian owners and breeders also need to be aware of genetic conditions found in other breeds, such as Hyperkalemic Periodic Paralysis (HYPP) in Quarter Horses. Because HYPP is a dominant trait, it is possible for a half-Arabian to be HYPP affected. Owners should check with the registry of any breed they are utilizing for additional information on breed associated genetic disorders.

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These Registration Commission materials were prepared in conjunction with the Equine Stress, Research and Education Subcommittee on Genetic Disorders with the assistance of Beth Minnich, Jennifer Ramsey, DVM and Scott Benjamin

Breeding Considerations

The decision to use carriers of recessive genetic disorders as breeding stock is an individual choice; there is no right or wrong answer. The primary goal is to not produce any affected foals. A secondary goal is to reduce the frequency of the mutation in the gene pool while maintaining the genetic diversity that is vital for the long term welfare of the Arabian breed. Breeding decisions must be made in an informed and careful manner to prevent needless suffering of foals and to preserve the health of the Arabian horse.

When the decision is made to use a carrier, they should never be bred to another carrier, as this may lead to affected foals.

Affected animals should never be used for breeding, as they will always pass on a copy of the mutated gene.

The information provided here is meant as a basic overview, to introduce owners and breeders to common genetic disorders that can affect Arabian and half-Arabian horses. Additional information is available at <http://www.arabianhorses.org/education/genetic/default.asp> and also consult with your veterinarian.