Genetic Disorders A BRIEF GUIDE FOR BREEDERS

The Arabian horse, like other equine breeds and other animals, carries mutated genes. Some cause problems; some do not. Six disorders in Arabian horses have been identified as being caused by, or are likely to be caused by, mutations that are inherited:

- Severe Combined Immunodeficiency (SCID)
- Cerebellar Abiotrophy (CA)
- Lavender Foal Syndrome (LFS)
- Juvenile Epilepsy (JE)
- Gutteral Pouch Tympany (GPT)
- Occipito-Atlanto-Axial Malformation (OAAM). (See below and on the next page for a short description of each disorder.)

For three of these, SCID, CA, and LFS, there are tests. The SCID and LFS tests directly identify the mutated gene ("direct gene" test). The current CA test identifies two overlapping genes associated with the disorder.
All three of these disorders are autosomal recessive. Autosomal means that the gender of the parents or foal is not relevant. Recessive means that animals must inherit a copy of the mutated allele from BOTH parents to be affected (a foal inherits one allele from each parent; the combination of two alleles constitutes a gene).

SCID and LFS are both fatal in foals that inherit a copy of the mutated allele from both parents. CA is different in that it causes a variable amount of brain damage in horses that have two copies of the mutated allele. The mechanism that determines how badly a horse will be affected by CA is not yet well understood. Some horses can live into adulthood and may even be parents; some are not able to survive.

A horse with both copies of the allele being the mutated version will be AFFECTED. A horse that has only one copy of the mutated allele and one normal copy is often called a CARRIER. This is sometimes misunderstood to mean that the horse is somehow contagious; it is not. The only way to produce a foal that suffers from a disorder is if both sire and dam have at least one copy of the mutated allele, and a mutated allele is inherited from both parents. An AFFECTED SCID or LFS foal WILL DIE. An AFFECTED CA foal may not.

For all three disorders, CARRIERS will seem normal. The probabilities are much like flipping a pair of coins where one coin is the sire and one is the dam. But it is important to understand that EACH breeding is like a NEW flip of the coins. The numbers below are probabilities, not predictions or guarantees. Just as each foal has a 50% chance of being a colt and 50% chance of being a filly, there is no guarantee that if a mare produces three fillies in a row, the next one has to be a colt. Each breeding is independent of all previous breedings. The probabilities shown are for each breeding.

In the chart below, an AFFECTED is a horse where both copies of the allele are the mutated version. A CARRIER has one copy of the mutated allele and one copy of the normal allele. A CLEAR has two copies of the normal allele. It does not matter which parent is the sire and which is the dam; gender is irrelevant to how these disorders are inherited. By testing and then breeding only clear-to-clear or clear-to-carrier, breeders can be certain NOT to produce an affected foal. It is not necessary, nor is it desirable from a breed-diversity standpoint, to eliminate all carriers from the breeding pool. There is not yet enough data to say, with certainty, which lines are involved in each disorder. Testing and disclosure will expand our knowledge and, ultimately, help us all be better custodians of the Arabian breed.

Parent 1	Parent 2	Probability of an AFFECTED foal	Probability of a CARRIER foal	Probability of a CLEAR foal
Affected	Affected	100%	0%	0%
Affected	Carrier	50%	50%	0%
Carrier	Carrier	25%	50%	25%
Carrier	Clear	0%	50%	50%
Clear	Clear	0%	0%	100%

SCID — Severe Combined Immunodeficiency: Foals are born with a weak immune system, much like the "Bubble Boy" syndrome in humans. Foals usually die within weeks or months from an infection that would usually be fought off by a normally functioning immune system.

LFS — Lavender Foal Syndrome: Foals usually cannot stand at birth and may have seizures. Some are unusually large, with associated difficulty during delivery. Some are born with a pale coat color (sometimes described as lavender or silvery). LFS may be misdiagnosed as "dummy" foal (lack of oxygen at birth). Most are euthanized soon after delivery.

CA — Cerebellar Abiotrophy: May or may not be apparent at birth. Symptoms may not be noticeable until months or, in a few cases, years later, although the disorder is usually identified by 6-18 months of age. The disorder involves the death of Purkinje cells in the cerebellum, which affects coordination. Symptoms include a head tremor that may be noticeable when the foal/weanling is trying to nurse, drink, or eat ("intentional tremor"). Gait may be exaggerated, the forelegs that seem to be trying to climb a hill even on flat ground, or the horse may exhibit a chronically wide stance. Some just seem a little "off" or "clumsy" or "accident prone." Diagnosis can be confirmed by necropsy (laboratory evaluation of the cerebellum). The amount that an animal may be affected is unpredictable.

JE — Juvenile Epilepsy: Foals appear normal at birth but begin to have seizures within a few days to 6 months. The seizures are sometimes treated with medication; the foal appears normal between seizures. Most outgrow the seizures by 18 months. Researchers suspect it is inherited as a dominant, possibly with a link to LFS.

GPT — Gutteral Pouch Tympany: Usually apparent within the first year. A malformation in the Eustachian tube allows air to enter but not leave the guttural pouches. The result is swelling just behind the cheek. The area may become infected. Researchers suspect a genetic cause that may be related to gender, as fillies are more likely to be affected than colts.

OAAM — Occipito-Atlanto-Axial Malformation: Caused by fusion of cervical vertebrae and incorrect formation of the area where the vertebrae connect to the skull. Foals may show signs at birth or within a few weeks. They may not be able to stand and nurse. Compression of the spinal cord may lead to more severe symptoms, including paralysis of the legs. No research is currently being conducted on this disorder.

More Information

Information about Genetic Disorders can be found at the Institute website www.desertarabian.org and the AHA website www.arabianhorses.org/education/genetic/default.asp

A site devoted to Cerebellar Abiotrophy is at www.cerebellar-abiotrophy.org. It includes detailed scientific information, videos of affected horses, a list of horses with test results that have been made public, and much more.

The organization F.O.A.L. offers discounts on testing and maintains a public list of horses with the reported results. See www.foal.org.

The following labs in the US offer tests or one or more of these disorders:

VetGen http://vetgen.com/

Cornell Animal Health Diagnostic Center (LFS only) http://ahdc.vet.cornell.edu/news/lfs.cfm

UC Davis Veterinary Genetics Laboratory (VGL) http://www.vgl.ucdavis.edu/services/horse.php

Animal Genetics http://www.animalgenetics.com