



Genetic Disorders and the Arabian Horse – An Overview

(Information partially excerpted from “Caution and Knowledge: An introduction to the genetic diseases and disorders that affect the Arabian horse” by the AHA Equine Stress, Research and Education Committee, with special thanks to Lisa Goodwin-Campiglio, Beth Minnich and Brenda Wahler, published in the Aug/Sept 2007 issue of Modern Arabian Horse magazine - updated information provided by Beth Minnich, April 2010, January 2011, January and December 2012)

- **Severe Combined Immunodeficiency Disorder (SCID):** An affected foal is born with a severely weakened immune system. Because their natural defense system against infection is not functioning properly, by the time they are five months or so of age, they generally die of an opportunistic infection (such as pneumonia) or they are euthanized. *[note: a test is available for SCID – see “testing” section for details]*
- **Cerebellar Abiotrophy (CA):** The Purkinje cells in the brain’s cerebellum begin to die, resulting in a severe lack of coordination. The degree of severity can vary, but most affected individuals are euthanized before adulthood, due to the hazard they present to themselves and others.

An affected foal is usually born without clinical signs. However, as they begin to grow, the degeneration of the Purkinje cells begins. Clinical signs include a head tremor and severe incoordination, combined with an inability to accurately gauge distance. Additional signs include an exaggerated gait and when at rest, a widelegged stance. Young horses with CA are also hyper-reactive and somewhat more prone to rearing than ordinary horses, with the frequent result that they lose their balance and fall. Clinical signs may not appear immediately, but are often first noticed at times when the foal is under close scrutiny, such as weaning.

CA is often mistakenly diagnosed as Wobbler’s Syndrome or as head trauma from an injury. Wobbler’s Syndrome is caused by compression of the spinal cord, due to malformation of the cervical vertebrae during growth, and can be diagnosed with the assistance of radiographs. Clinical signs can also be misdiagnosed as head trauma from an accident, because foals often injure themselves by falling over backwards or colliding with a fence. However, both conditions are quite different from CA and care should be taken to differentiate them.

Although clinical signs and case history can lead to a diagnosis of CA, at this point in time, the only way to confirm such a diagnosis is to examine the brain tissue after euthanasia. *[note: a test is available for CA – see “testing” section for details]*

- **Lavender Foal Syndrome (LFS) [also known as Coat Color Dilution Lethal (CCDL) or Dilute Lethal]:** This is a neurological disorder thought to be caused by a brain lesion. An affected foal cannot stand at birth and usually has seizures. LFS foals are frequently born with a telltale diluted coat color that can make the hairs appear to be a dull lavender, a pinkish-brown or somewhat silvery. In many cases, the foal was also a product of a difficult delivery. If the coat color is overlooked or not present, foals may be misdiagnosed as having neonatal maladjustment syndrome (known as “dummy” foals), due to a lack of oxygen from the dystocia.

It is critical for breeders to support the ability of modern science to locate the genes that cause these conditions. Developing genetic tests is a must!



Foals are usually euthanized within a few days after birth. *[note: a test is available for LFS – see “testing” section for details]*

- **Juvenile Epilepsy:** Although not generally fatal, it can be disabling and there has been a suggested genetic link to Lavender Foal Syndrome. Affected foals are born normal, but will have periodic epileptic seizures, beginning anywhere from 2 days to 6 months after birth. Between seizures, they appear normal. Treatment can include the use of traditional anti-seizure medications, which may reduce the severity of the clinical signs. Affected individuals usually outgrow the condition between 12 and 18 months.
- **Guttural Pouch Tympany (GPT):** This is a condition caused by the guttural pouches becoming distended with air, due to a malformation of the Eustachian tubes that allows for air to enter the guttural pouches, but not escape. GPT is seen in young horses, ranging from birth to a year or so in age and fillies are two to four times more likely to be affected than colts. Diagnosis of GPT can generally be done based on clinical signs, which include the classic non-painful swelling in the area just behind the cheek in the parotid region, along with radiographs of the skull. Because horses suffering from GPT can acquire respiratory infections, including pneumonia, use of anti-inflammatory and antimicrobial medications are sometimes needed. In order to correct the physical defect, surgery is required.
- **Occipito-Atlanto-Axial Malformation (OAAM):** This is a condition where the cervical vertebrae fuse together and to the base of the skull, causing compression and injury to the spinal cord. Affected foals are often unable to stand and nurse, but in other cases, the clinical signs may not become noticeable for several weeks. Clinical signs range from mild incoordination to the paralysis of both front and rear legs. This is the only cervical spinal cord disorder seen in horses less than one month of age, and a radiograph can assist in diagnosing the condition.

Frequently Asked Questions

How are they inherited?

SCID is known to be an autosomal recessive trait and breeding experiments conducted at the UC Davis Veterinary Genetics Laboratory indicate that CA is also an autosomal recessive trait. In addition, research on LFS indicates that it too is an autosomal recessive disorder. While the genetic nature of OAAM is not yet fully understood, some researchers have suggested that this is also an autosomal recessive condition. "Autosomal" means the trait is not sex linked, and "recessive" means that in order for a foal to be affected, it must have two copies of the mutated allele, receiving one copy from each parent.

The mode of inheritance for Juvenile Epilepsy has not yet been determined, however research from UC Davis indicates it is an autosomal dominant trait. A “dominant” trait means that an affected foal only needs to inherit one copy of the mutated allele to show clinical signs. An example of a dominant trait is Hyperkalemic Periodic Paralysis (HYPP) in American Quarter Horses. Recent research updates for GPT indicate there is some sex influence (fillies are more likely to be affected than colts) and it is a polygenic trait (multiple genes involved).

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Does linebreeding or inbreeding "cause" these diseases?

No. However, the more the gene pool is reduced by closely breeding related horses and if carriers are present, the frequency of the mutated allele(s) has the potential to increase in the population.

Is there any bloodline of Arabian horse more or less likely to have genetic conditions?

No, all lines in the Arabian breed have the potential to carry genetic conditions. For example, SCID and CA have appeared in virtually all modern major bloodline groups and appear to be distributed throughout the breeding population. In addition, while LFS is often associated with horses of Straight Egyptian or heavy Egyptian breeding, it has been identified in other breeding groups.

Did one horse or bloodline "cause" these diseases? Aren't these diseases a sign of "impure" breeding?

No and No. Cellular mutations are caused every time a cell divides; usually they have little to no effect. However, sometimes these mutations do end up altering a cell's function. In addition, while some historical animals are very likely to have been carriers (based on pedigree analysis of get or grandget who produced affected foals), there is anecdotal evidence that at least some of these conditions appeared even in desert bred Arabians. To quote the late Dr. Ann Bowling: *"Deleterious mutations that occur in purebred breeds are usually chance "hitch-hikers" in highly successful breeding lines, otherwise, the homozygous genotypes that produce the problem conditions would be so rarely encountered as to be overlooked or written-off as problems with unknown causes."*

Can't we eliminate genetic diseases by not breeding known carriers?

Without a test, it is impossible to identify all carriers. Just because a horse has "so far" never produced an affected foal does not mean it is "clear." Because stallions can sire many offspring, they are more likely to eventually be revealed as a carrier. Mares, however, may be unrecognized carriers. Even if bred to a carrier stallion, statistical probability may mean that they never produce an affected foal. Thus, a recessive gene can be passed on for generations without an affected foal being born.

Further, by removing all known carriers from the breeding population, the already limited Arabian gene pool would become even more limited and the breed could lose some highly valued bloodlines. In addition, a less-diverse breeding population may actually allow other genetic conditions to become more widely distributed. The success of SCID testing indicates that if known carriers are bred, with careful, selective breeding and testing of offspring, even carrier lines can be "cleared" of the gene.

The SCID test allows people to avoid ever breeding an affected animal, and allows breeders to make an informed choice to breed or not breed a carrier, as they may choose. However, for the remaining conditions, until there is a DNA test, there is no way to know which horses carry the gene, short of producing an affected foal. Likewise, even when two carries are bred, there is an equal statistical probability that their offspring will be clear or affected, thus guilt by association may unnecessarily taint the reputation of horses that actually do not carry the gene at all.

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Additional information on research projects:

Cerebellar Abiotrophy – UC Davis

Dr. Cecilia Penedo - mctorrespenedo@ucdavis.edu,

phone: 530-752-7460

<http://www.vgl.ucdavis.edu/genomic/cerebellar/>

Lavender Foal Syndrome – Cornell University

Dr. Samantha A. Brooks - equinegenetics@cornell.edu

phone: 530-752-1170 or 530-752-7267

Guttural Pouch Tympany - University of Veterinary Medicine Hannover, Germany

Dr. Ottmar Distl - ottmar.distl@tiho-hannover.de

http://www.tiho-hannover.de/einricht/zucht/mol_gen/luftsack_e.htm

Are tests available to determine carriers? [Updated December 2012]*****

Testing Information

- ***Cerebellar Abiotrophy (CA)***

UC Davis Veterinary Genetics Laboratory (VGL) - Cost is \$40 per test. Test can be ordered online from the VGL: <http://www.vgl.ucdavis.edu/services/horse.php> or call (530) 752-2211.

VetGen - Cost is \$35 - \$50 per test, depending on the number ordered. Test can be ordered through VetGen: <http://www.vetgen.com/documents/order-form-equine.pdf> or call (800) 483-8436.

- ***Lavender Foal Syndrome (LFS)***

Cornell Animal Health Diagnostic Center (AHDC) - Cost is \$47 per test (\$45 for the test, plus \$2 accession fee). Test can be ordered through the AHDC: <http://ahdc.vet.cornell.edu/news/lfs.cfm> or call (607) 253-3900.

VetGen - Cost is \$35 - \$50 per test, depending on the number ordered. Test can be ordered through VetGen: <http://www.vetgen.com/documents/order-form-equine.pdf> or call (800) 483-8436.

- ***Severe Combined Immunodeficiency (SCID) testing available from the VetGen*** - Cost is \$99 per test when ordered through FOAL. Test can be ordered through FOAL: http://www.foal.org/SCID_CA_LFS_KitOrder.pdf or contact: Arabian F.O.A.L. Association, Marguerite Illing, Treasurer, PO Box 198, Parksville, NY 12768-5336

- ***Combination testing for CA/LFS is now available from UC Davis*** - Cost is \$40 for one test per animal OR \$60 for both tests per animal. Tests can be ordered online from: <http://www.vgl.ucdavis.edu/services/horse.php> If you are unable to place an online order, please contact the VGL - phone (530) 752-2211.

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- **Combination testing for SCID/CA/LFS is now available from VetGen - Special pricing is available through FOAL - http://www.foal.org/SCID_CA_LFS_KitOrder.pdf**

- Cost for CA test is \$42.50 when added to \$99 SCID test order for the same horse.
- Cost for LFS test is \$42.50 when added to \$99 SCID test order for the same horse.
- Cost for combined SCID/CA/LFS test for the same horse is \$184.

Note: If a horse was previously tested for SCID or CA, a test for LFS can be done on that horse for \$42.50.

- Bulk order pricing directly from VetGen: <http://www.vetgen.com/documents/order-form-equine.pdf>

Voluntary reporting of test results: FOAL.org accepts test results for SCID, CA and LFS for inclusion on their publically available test results list. Please visit <http://foal.org/Test-Kits---Forms.html> for more information.

For additional information on genetic disorders affecting Arabians, please visit <http://www.arabianhorses.org/education/genetic/default.asp>

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