Lavender Foal Syndrome (LFS)/Coat Color Dilution Lethal (CCDL)

Lavender Foal Syndrome (LFS), also known as Coat Color Dilution Lethal (CCDL), is a lethal neurologic disorder caused by a mutation of the MYO5A gene on chromosome 1. In addition to being born with the unique diluted coat color that can appear to be pale lavender, pale pink or silver, affected foals are unable to stand and will have episodes of tetany where the foal will lay on its side rigidly extending its limbs, neck and back. During these tetanic episodes, the foal will also frequently make paddling motions. In addition, affected foals are often large and have a difficult delivery (dystocia). It is important to note that not all foals will have the noticeable dilute coloration (although the coat color may appear more washed out than normal), so an affected foal may be initially misdiagnosed with neonatal maladjustment syndrome ("dummy foal") associated with the difficult delivery, a spinal cord injury, or encephalitis. The availability of a genetic test for LFS will now allow for easier differentiation among these conditions. Although an affected foal is unable to sit upright or stand on its own to nurse, it may have a strong suckle reflex and may be bottle fed. However, there is no treatment for LFS, and affected foals will either die or need to be euthanized, generally within days after birth.

LFS is rare and is considered to be an autosomal recessive trait. "Autosomal" means that there is no sex linkage, so both males and females can be equally affected. "Recessive" means that in order for a foal to be affected, it must have received two copies of the mutated gene, inheriting one copy from each parent. Horses that have one copy of the mutated gene, in combination with one copy of the normal gene, are physically normal but are considered carriers and have a 50% probability, each time they are bred, of passing the mutation along to their offspring. Of additional note, there has also been a proposed link between LFS and Juvenile Idiopathic Epilepsy; further work is being pursued to investigate this theory. While LFS is generally associated with horses of Egyptian breeding, the disorder has been reported in other bloodline groups. Since owners and breeders now have the ability to test their breeding stock to determine LFS clear, carrier or affected status, informed choices can be made for breeding selections, and there never needs to be another LFS affected foal born.

As noted by veterinary neurologist, Dr. Alexander de Lahunta, "necropsy studies have found no gross or microscopic abnormality in the central nervous system of these foals. It is therefore suspected that a neurochemical or submicroscopic structural abnormality is the cause of the clinical signs." The protein myosin Va, coded for by the MYO5A gene, is involved with the movement and depositing of pigment, which affects hair color; in particular, the MYO5A mutation causes a clumping of pigment granules, which results in the diluted appearance of the hair coat. MYO5A is also involved in neuron function. As a result, the alteration in the normal coding for this gene could explain the neurologic aspects and dilute coat color associated with LFS.

In 2008, Cornell University in conjunction with the Arabian Horse Foundation initiated a new study on LFS to locate the mutation responsible and develop a direct DNA test. Using single nucleotide polymorphism (SNP) technology made available from the completion of the equine genome sequence in 2007, a small bank of 6 samples previously obtained from LFS affected foals during the past decade were

analyzed and a specific location (locus) of interest on chromosome 1 was quickly identified. Through further sequencing of the candidate genes found at this locus, a single base deletion in the MYO5A gene was detected. Not only has the mapping and discovery of this mutation been beneficial for the development of a LFS test, this project also represents the first time whole genome SNP scanning in the horse has been successfully used for any trait, which is an important accomplishment for the research arena.

LFS testing can be done through the following labs:

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.

*****Combination testing for SCID/CA/LFS is now available from VetGen/special pricing with FOAL*****

- 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.
- Test orders can be placed 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
- Bulk order pricing directly from VetGen: http://www.vetgen.com/documents/order-form-equine.pdf

For additional information on LFS please contact:

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Reference Materials

1) Brooks S, et al. Whole-Genome SNP Association in the Horse: Identification of a Deletion in Myosin Va Responsible for Lavender Foal Syndrome. PloS Gen 2010;6(4)e1000909. http://www.plosgenetics.org/article/info:doi/10.1371/journal.pgen.1000909

2) Gabreski N, et al. Mapping of Lavender Foal Syndrome using the EquineSNP50 Chip. J Eq Vet Sci 2009;29(5):321-22.

3) Page P, et al. Clinical, Clinicopathologic, Postmortem Examination Findings and Familial History of 3 Arabians with Lavender Foal Syndrome. J Vet Intern Med 2006;20:1491–94.

4) Fanelli HH. Coat colour dilution lethal ('lavender foal syndrome'): a tetany syndrome of Arabian foals. Eq Vet Ed 2005;17(5):260-63. http://www.foal.org/Fanelli_260_263.pdf

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