ARABIAN HORSE FOUNDATION UPDATE – JANUARY 2011 (updated February 2011)

Equine Cerebellar Abiotrophy (CA) – Update on UC Davis Research Project: Potential causative mutation for CA identified

Background:

Equine Cerebellar Abiotrophy (CA) is a debilitating degenerative condition of the cerebellar portion of the brain which results in a severe lack of coordination. The degree of severity can vary among individual horses, but most affected horses are euthanized before adulthood, due to the hazard they present to themselves and others, and the current inability to treat or cure the condition.

Breeding experiments conducted at UC Davis indicate that CA is the result of an autosomal recessive gene mutation. *Autosomal* means the disorder is not sex linked (both sexes can be affected) and *recessive* means both parents must contribute the "CA gene" in order to have an affected foal (this is the same mode of inheritance as SCID and LFS).

Additional information on CA can be found here:
http://www.arabianhorses.org/education/genetic/default.asp

The New Year begins with a lot of positive news about the CA Project.

DNA Test Advancement

Research conducted at the UC Davis Veterinary Genetics Laboratory (VGL) by Drs. Leah Brault and Cecilia Penedo has identified a mutation that is associated with CA. This potential causative mutation involves 2 overlapping candidate genes that are undergoing further study. This is an important finding, as it moves the currently available CA test from a marker based screening test to a more fully developed DNA test. The availability of this test continues to help diagnose the defect in suspect foals and to help owners identify carriers in their breeding stock. The true value of testing is to help guide breeders in making safe mating selections, with the goal to never produce a CA affected foal.

The Arabian Horse Foundation has provided additional funding to the VGL to incorporate whole genome scanning into the project, to further validate the location of the CA gene in the horse genome.

Further information and details on the CA research and development of the CA test are published in the February 2011 issue of the journal, Genomics.

Owners interested in submitting samples for testing can order the test directly from the Veterinary Genetics Laboratory's website: http://www.vgl.ucdavis.edu/services/horse.php

******IMPORTANT NOTE FROM THE VETERINARY GENETICS LABORATORY (VGL), UC DAVIS FOR OWNERS WHO HAVE ALREADY TESTED FOR CA AT THE VGL******

******All cases from CBA492 onwards have already been tested for the mutation. The VGL will be retesting all cases reported as CA/CA and N/CA before CBA492 and the clients who submitted the samples in question will be updated. There will be no charge to owners for this retesting. N/N cases will not be retested.******

******UPDATE ON SAMPLE RETESTING (February 2011)******

******The UC Davis VGL recently retested samples that had been submitted for the Cerebellar Abiotrophy Screening test between April 2008 and September 2009 with the new mutation the VGL later discovered and found to be associated with CA.**

Although the indirect test that was available then had a high rate of accuracy, it was expected that, in a few exceptional cases, it might incorrectly indicate the presence of the CA gene. Among the 491 samples reanalyzed, 16 results changed based on the new test. This retesting indicates the indirect test had an accuracy rate of 97% relative to the new test. Cases which were previously reported as N/N based on the indirect test did not display the critical markers; their status remains N/N based on the new test.****

Owners who have foals suspected of having CA are encouraged to contact Dr. Penedo:
Cecilia Penedo, PhD
UC Davis Veterinary Genetics Laboratory
Tel: (530) 752-7460, Fax: (530) 752-3556
e-mail: mctorrespenedo@ucdavis.edu

Other CA Project Updates:

In addition to the publication in Genomics, the UC Davis group also has two additional papers slated for publication in the near future:

- *American Journal of Veterinary Research* – this paper reviews work done to determine an autosomal recessive mode of inheritance for CA.
- *Equine Veterinary Journal* – this paper discusses a survey of 31 breeds and finding the CA mutation that is being studied in Arabians also in the Trakehner, Bashkir Curly and Welsh Pony breeds.

The Arabian Horse Foundation would like to thank all of the donors who have helped support this important research project, along with the owners who have submitted samples and provided information to the UC Davis team.

Plans are currently being made for additional research this year to look at the functional aspects of the two candidate genes being studied. As such, **continued support** is important and needed.

Donations to the Arabian Horse Foundation can be made at:
http://www.arabianhorsefoundation.org/donate.html

For more information about the Arabian Horse Foundation, please contact:
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