AMHA Genetics Committee Report

Feb 20th 2009

- No committee members were present
- Discussions with members via phone were
- Concerns on the lack of encouragement of AMHA to recommend color testing for some horses being registered with ambiguous coat coloration.
- How AMHA should AMHA best handle Dwarfism testing and positive carriers - need to have something in place BEFORE the test is available

Silver Testing

- Silver gene test is recommended to be utilized more by the registrar in this breed
- Especially on some horses for registration applications that are called chestnuts, liver chestnuts, silver bays and champagne.

Latest Genetic News online - *TheHorse*

"With MAF funding, members of the ECGR are using SNP (single nucleotide polymorphism) chips--a genetic tool recently developed with funding from MAF, the USDA, and European partners--to study recurrent exertional rhabdomyolysis, commonly known as tying-up, in Thoroughbreds; lavender foal syndrome in Arabians; extreme lordosis, or swaying of the back, in Saddlebreds; dwarfism in Miniature Horses, as well as susceptibility to cribbing and immune/infectious diseases in a number of breeds.

In several cases, they are close to identifying the location of the genetic mutation(s) responsible for a particular disease."

"The SNP chips allow us to identify those genes that contribute to a genetic disease much more quickly," Mickelson explained. "We can then hope to more rapidly find the specific gene and mutation and develop a DNA test to determine a horse's genetic risk for susceptibility to that particular disease."

Research progress

- 2008 Acquired additional funding and support through MAF to become one of the first Equine genetic research projects to utilize the newly designed Mayo "Snip Chip" designed by Illumina in fall of 2008
- Analyzed data and found a candidate gene associated with Type I dwarfism
- Currently finalizing sequencing work on the associated gene
- Possible mutation found associated with Type I dwarfism in process of analyzing all current samples in hand
- Possible test available by summer 2009
- If there is interest in helping I request any and all samples of dwarfs- only hair and pic needed to determine type.

Presentation of work on Dwarfism

- Equine Science Society
- Conference in Keystone, Colorado
- Keystone Conference Center - May 29-31

Investigation of Dwarfism Among Miniature Horses using the Illumina Horse SNP50 Bead Chip.

J. Eberth*, T. Swerczak, E. Bailey; MH Gluck Equine Research Center/ University of Kentucky, Lexington, KY, USA

Introduction

There are dwarfism genes which greatly reduce statute and may negatively impact health and reproduction. This is not considered a desirable genetic trait for Miniature horses. Therefore, the following studies were conducted to discover the genetic basis for dwarfism.

Dwarfism Study

Materials and Methods

Pedigree records suggested that dwarfism occurs as a recessive genetic trait among miniature horses. Pathological examination, involving comparison of skeletal development and other phenotypic traits, suggested that there may be as many as four distinct types of dwarfism segregating among miniature horses.

Type I dwarves exhibit cranial abnormalities of a disproportionately large head, large bulging eyes and eye sockets, a forehead with relative frontal domed prominence and a relatively short stubbed muzzle.
An underbite of variable severity is commonly seen in this type however some have a normal bite.
The midface is often small with a flat nasal bridge and narrow nasal passages and the airway obstruction can be "central" in origin (due to foramen magnum compression) or "obstructive" in origin (due to narrowed nasal passages).Symptoms of airway obstruction include snoring.
Other characteristics are shortened limbs, enlarged joints, malformed or bowed legs with limited extension and flexion and overall disproportionate short stature. Progressive hoof deformities resulting from malformed limbs as well as progressive arthritis in the limbs become worse with age. Spinal abnormalities such as roachback may or may not develop later.

Dwarfism Study

To avoid pooling samples from different genetic forms of dwarfism, horses were selected based on a common phenotype and belonging to a common family line. This phenotype was identified as Type I dwarfism of miniature horses. DNA was isolated from blood or tissues of 20 horses exhibiting type I dwarfism and from 20 relatives that did not exhibit the dwarfism traits. The DNA was tested using the Illumina Equine SNP50 bead chip. The results were analyzed using PLINK v 1.04.

Results

A single chromosome region was strongly associated with the trait. EMP2 (empirical P value, corrected for all tests) value of 0.019 was obtained, strongly supporting this candidate region. Within the candidate region a candidate gene was found which causes dwarfism among humans.

Discussion

The candiate gene is currently being sequenced for horses with the hope of discovering a mutation responsible for the trait. Additional work will entail identifying a haplotype signature for Type I dwarfism that could be used to estimate its frequency in the population and determine the phenotypic heterogeneity associated with this haplotype.

ACKNOWLEDGEMENTS

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Champagne Test

We can accurately identify a champagne horse through two methods.

1. **Confirmation through the ICHR.** This may include a photo copy of the horse's ICHR registry papers. Photo copies can be checked against the ICHR on-line stud book for accuracy. ICHR is also willing to consult directly with the AMHA registrar on questionable horses.

(When the University of Kentucky was doing the final testing for the new champagne test, 100% of the ICHR registered horses whose owners submitted samples tested positive. The several rejected by ICHR tested negative.)

1. **2. Color DNA testing specific for champagne** done through one of several laboratories. Either one if these two measures should provide adequate proof that a stated horse is a true champagne, and will discourage those with dishonest intentions to continue to take advantage of the system.

Champagne Test

- For current members of the ICHR, use a copy of the ICHR paperwork as proof of champagne with AMHA.
- For those who do not wish to join ICHR, the DNA test for champagne at some labs is about $25.
- With more intentional or unintentional false claims of champagne being made, the Genetics committee would recommend that AMHA require DNA test or ICHR proof before registering a horse as champagne.
- As a side note, some of these horses incorrectly registered Champagne are most likely Silvers, however, this does NOT mean that a horse cannot be Silver AND Champagne, a horse very possibly can.
- This reason is why it is recommended to test for Silver as well.

AMHA Color List online for registration

- Quintin and I have been working on a revamping of the color listing for AMHA and how it will be utilized in the registration of horses in the registrars office.
- I currently do not know the reality if the change can occur, Quintin would need to comment
- I am very willing to assist Quintin and AMHA in this update that is so desperately needed