PSSM1 and Gypsy Vanner Horses

PSSM1 is present in some Gypsy Vanner Horses most likely due to the large draft breed heritage. This is also present in some light breeds as well as draft.

Type 1 Polysaccharide Storage Myopathy (PSSM1) is a muscle disease characterized by accumulation of abnormal complex sugars (glycogen) in skeletal muscles. The accumulation of these sugars can cause breakdown of muscle fibers which leads to muscle pain, weakness, skin twitching, sweating, and reluctance to move. The PSSM1 variant does not explain all cases of excessive abnormal glycogen accumulation in the muscle. It is likely that other genetic factors contribute to this condition, but to date no genetic variants for other types of PSSM have been identified or scientifically validated.

PSSM1 Testing Results explained:

- Horses with **n/n** genotype will not have PSSM1 and cannot transmit the PSSM1 variant to their offspring.
- Horses with **n/PSSM1** genotype will have the PSSM1 variant and may show signs of disease. These horses may transmit the PSSM1 variant to their offspring.
- Horses with **PSSM1/PSSM1** genotype are homozygous for the PSSM1 variant and may be more severely affected. These horses will transmit the PSSM1 variant to all of their offspring.

Managing Horses by Feed and Exercise

Certain management practices can help horses with polysaccharide storage myopathy (PSSM). Neither straight cereal grains such as plain oats nor textured or sweet feeds containing cereal grains should be fed. Specialized feeds for horses that are a low-starch, high-fat feed are most appropriate.

Exercise is also important to the management of symptoms.

PSSM1 Study funded by the GVHS with the University of Kentucky.

In order to obtain an estimate of the frequency of the PSSM1 mutant allele in horses registered with the Gypsy Vanner Horse Society, 100 hair samples were selected at random from the GVHS database, spanning the years 2003-2015. Samples were tested using the TaqMan assay system for the G>A SNP in exon 6 of the GYS1 gene.

Of the 100 samples, 76 had a normal genotype of N/N and 24 tested as heterozygous N/PSSM1. This translates into 176 normal alleles and 24 mutant alleles, giving an allele frequency of 88%

normal and 12% mutant. We did not find any samples that were homozygous for the mutant allele.

Using these observed values of allele frequency, our Hardy-Weinberg calculation of the EXPECTED genotype frequencies in the total population are as follows:

Expected genotype frequency for N/N: 77.44%
Expected genotype frequency for N/PSSM1: 21.12%
Expected genotype frequency for PSSM1/PSSM1: 1.44%

Therefore approximately 1 in 5 horses can be expected to be heterozygous for the mutation, and 1-2 horses per 100 horses can be expected to be homozygous for the mutation.

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Upon reviewing the results of this study, the GVHS has contacted the University of Minnesota Equine Center **Neuromuscular Diagnostic Laboratory,** for their direction on next steps to conduct the scientific research to fully understand what this means for the Gypsy Vanner Breed. More information will be forthcoming once the particulars of this further study have been finalized. GVHS management has finalized an agreement with the University of Kentucky to offer PSSM1 testing to GVHS owners as part of specially priced testing "bundles".
Until more study is completed this caution from Dr. Graves still applies:

"*Please advise your breeders and owners to wait until a scientific estimate of frequency is available and work together to decide how PSSM1 will be dealt with in your breed* ***before valuable breeding stock is lost****."*