Equine Myopathy (PSSM2)

PSSM2 – What does it mean for my breeding program?

Information & Recommendations

Test	# H108
Sample	Hair or Blood
Price	200 € (net)
Time required	10 - 14 days

PSSM2 stands for "Polysaccharide Storage Myopathy Type 2" and is an umbrella term for multiple muscle diseases with similar symptoms.

The name was given because PSSM Type 1 has similar symptoms, and it can be difficult to distinguish between the types of disorders.

PSSM2 is actually not a defect of muscle polysaccharide storage. We now know that the symptoms of PSSM2 result from defects in muscle fibers themselves.

Until recently, PSSM2 was diagnosed by ruling out all other possibilities. If a genetic test for PSSM1 was negative, but a biopsy showed disrupted muscle formation, the horse was diagnosed with PSSM2. Four genetic variants associated with PSSM2 have now been identified. The test for these four variants is exclusively available in Europe from CAG.

Typical Symptoms of Equine Myopathy (PSSM2)

The first symptoms of PSSM2 are most often seen between 7-10 years of age.

- Changes in behaviour (likely related to pain)
- Shifting lameness (especially in the stifles)
- Severe muscle wasting (especially in the hindquarters and topline / shoulder girdle)
- Localized muscle wasting causing small divots, often resembling kick marks
- Coordination problems /.Ataxia
- Muscle tension / cramping / tying up
  - Stiff hindquarters
  - Muscle tremors
- Gait changes (reduced stride length, lack of drive from hindquarters, bunny hopping, rope walking, etc.).

Genetic Tests and Inheritance

Four mutations associated with PSSM2 have been identified and can be tested by CAG.

Myopathy Panel	Variants tested
#H108	P2, P3, P4, Px

All four variants have a semidominant mode of inheritance. This means that one copy of the variant is sufficient to increase the risk of a horse developing PSSM2. If a horse has two copies of a variant, it will develop symptoms at a younger age, and will have a more severe disease process. Because the mutations are in different genes, it is possible for a horse to have a combination of variants.

Caring for PSSM2 affected horses

Hereditary muscle diseases are not curable. Most PSSM2 affected horses benefit from diets with high fat and protein, and/or supplementation of the amino acids Lysine, Threonine, and Methionine, along with regular exercise.

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Breeding recommendations:

General Information

- Unhealthy horses (having symptoms of any disease) should not be used for breeding.
- Whether it is symptomatic or not, a horse with any copies of a dominant mutation has the probability of passing the mutation on to its offspring. With each breeding, a horse with one copy (affected heterozygote) has a 50% probability passing the mutation on to its foal. A horse with two copies (affected homozygote) has 100% probability of passing the mutation on to each foal.

PSSM2 – semidominant Inheritance, incomplete Penetrance

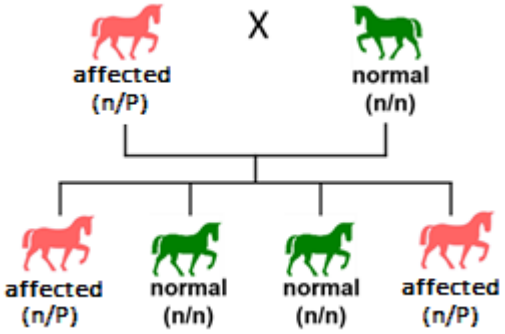
Semidominant hereditary diseases (having incomplete penetrance) have a great variability in how and when their symptoms are seen. Horses which have the same mutations can have very different clinical profiles. While one horse is mildly affected, another may be much more severely affected. This variation is seen because the symptoms can be influenced by other (known and unknown) genetic variants, and by environmental influences (nutrition, handling, stress). For these reasons, a full breeding ban on an asymptomatic horse with one copy of one of these known mutations is not recommended at this time. By removing all affected animals from a breeding population, the genetic diversity is artificially reduced. This often leads to other genetic diseases becoming more common, which can cause a breed even more problems. Decisions regarding the breeding of affected heterozygotes should be made on an individual basis.

Things to note:

- The first symptoms are usually seen in adult horses (7-10 years). At the age of breeding maturity, affected horses will still appear healthy. Only a genetic test allows an owner to determine if a horse has any of the PSSM2 variants, and to make an informed breeding decision. When a horse has no PSSM2 variants, it can be used for breeding without any concern. When a horse has one or more PSSM2 variants, the following points should be noted:

- Horses with more than one PSSM2 variant (for example, P2/P2, n/P2 + n/P4, etc.) should not be used for breeding. These horses will develop symptoms at a younger age than animals with only one variant, and the disease will be more severe.

- Horses with only one copy of a known mutation and which are otherwise healthy, excellent representatives of their breed and have a high genetic and breeding value, may be used for breeding after careful consideration of all factors with a veterinarian, breed club representative and an animal geneticist. In such a case, the horse should only be bred to a horse that has tested normal for all known variants.



EQUISEQ

CAG holds the exclusive license to offer the EquiSeq Equine Myopathy Panel in Europe. Other forms of equine myopathies are still being researched. Genetic tests for new variants will also be available at CAG.