



The Importance of Equine Genetic Testing

The Educated Horseman: Reproduction Series



Horses can be affected by a variety of genetically linked disorders. In 2009, the whole horse genome sequence was categorized. This advancement in genetics has produced affordable genetic testing, advanced management and medical treatment of affected animals and helped to create breeding protocols that focus on reducing the impact genetic diseases have on the horse industry.

Of particular interest to owners of quarter horses is a five-panel test that examines polysaccharide storage myopathy (PSSM), malignant hyperthermia (MH), hyperkalemic periodic paralysis (HYPP), hereditary equine regional dermal Asthenia (HERDA) and glycogen branching enzyme deficiency (GBED). Additionally, beginning in 2015, the American Quarter Horse Association began requiring that ALL stallions have a five-panel genetic test completed before their 2016 foals can be registered. A detailed look at the diseases tested follows.

Polysaccharide Storage Myopathy

PSSM is a dominant autosomal hereditary condition caused by the mutation of the glycogen synthase 1 (GYS1) gene. This disease creates a muscle condition similar to tying up that affects 11 percent of quarter horses. Since it is a dominant condition, only one copy of the GYS1 gene needs to be present for its offspring to be affected. The test has three potential results:

1. P1/P1. This means your horse is positive for the dominant PSSM gene mutation and carries two copies of the GYS1 gene. Horses that are homozygous (having two copies) will pass the disease to 100 percent of their offspring.
2. n/P1. This means your horse has one copy of the GYS1 gene, and it is affected by PSSM. Horses that are heterozygous (having one copy) will pass the disease to 50 percent of their offspring.
3. n/n. This horse is negative for PSSM and cannot pass on the gene mutation to any offspring and should not display any symptoms of the disease.

Horses that have inherited the genetic mutation causing PSSM overproduce glycogen, which leads to excess sugar stored in the muscle that is not easily accessed during exercise. The exercise causes an energy deficit within the

muscle, causing muscle pain, stiffness, skin twitching, sweating, weakness, reluctance to move, gait abnormalities, mild colic and mild muscle wasting. These horses should be maintained on a low-starch and low-sugar diet with regular and consistent exercise.

Malignant Hyperthermia

MH is an autosomal dominant disease caused by mutation in the ryanodine receptor 1 (RyR1). This disease creates a rare muscle disorder that affects any horse related to a quarter horse. Since it is a dominant disease only one copy of RyR1 is required for the condition to exist. The test has three potential results:

1. MH/MH. This means your horse is positive for the MH mutation and indicates the horse carries two copies of the mutated gene. Homozygous horses will pass the disease onto 100 percent of its offspring.
2. n/MH. This means your horse has one copy of the MH mutation and indicates the horse is positive for MH. Heterozygous horses have a 50 percent chance of spreading this disease to its offspring.
3. n/n-. This horse is negative for MH and does not carry the gene mutation. It will not pass the condition onto its offspring and will not be affected by the disease.

Horses that have inherited the MH gene have a malfunctioning calcium-release channel within skeletal muscle cells. This causes excess calcium to be released into the portion of the cell that causes contractions and increases muscle metabolism. Horses affected by MH may not show any physical signs of the disorder until it is triggered by extreme exercise, stress or specific anesthetic (halothane). Horses experiencing symptoms of MH will have a fever often exceeding 109 F, excessive sweating, increased heart rate, abnormal heart rate, high blood pressure, acidosis and muscle damage. If not treated immediately, this condition can be fatal. Horses affected by MH can be managed during surgery if known prior to the procedure.

Hyperkalemic Periodic Paralysis

HYPP is an autosomal dominant disease caused by point mutation in the SCN4A gene that affects 1.5 percent

of quarter horses and up to 56 percent of halter horses. HYPP is a muscular disease that has been traced back to a quarter horse stallion named Impressive. Since it is a dominant disease, only one copy of the mutation is needed for the condition to exist. The test has three potential results:

1. H/H. This means your horse carries two copies of the mutated gene. It will pass this disease to 100 percent of its offspring and is positive for HYPP.
2. n/H. This means your horse carries one copy of the mutated gene. It is affected by HYPP and has a 50 percent it will pass the disease onto its offspring.
3. n/n. This horse is negative for HYPP, shows no symptoms and cannot pass it on to offspring.

Horses affected by HYPP have a mutation in the sodium-potassium pump system that is involved in controlling muscle contraction. This disease causes involuntary muscle contraction that may result in tremors, temporary paralysis and respiratory dysfunction and even death. Positive horses need to be maintained on a strict diet and exercise program to reduce excess potassium in their diet and help them maintain appropriate muscle function. Owners of positive horses should be cautious when riding or handling these horses because HYPP attacks are unpredictable.

Hereditary Equine Regional Dermal Asthenia

HERDA is an autosomal recessive disease caused by a mutation in peptidyl-prolyl isomerase B (PPIB) gene that affects 3.5 percent of quarter horses. HERDA causes a collagen deficiency that restricts adhesion of the skin layers linked to quarter horse stallion Poco Bueno. Since this is a recessive disease, a horse must have two copies of the mutated gene to be affected. The test has three potential results:

1. Hrd/Hrd. This means your horse is positive for HERDA and carries two copies of the gene.
2. Hrd/n. This means your horse is a carrier and only has one copy of the gene. It will not show any symptoms of the disease but will have a 50 percent chance of passing the mutated gene to its offspring.
3. n/n. This means your horse is negative, not affected by HERDA and will not pass the mutation onto any offspring.

Horses positive with HERDA have multiple slow-healing skin abrasions not typically noticed until training begins. The friction and pressure caused by training aids and saddles cause large sheets of skin to separate. There is no cure for this disorder or a way to effectively manage it. Positive horses are typically euthanized. Breeding carrier animals also is not advised.

Glycogen Branching Enzyme Deficiency

GBED is an autosomal recessive disease caused by a mutation in the GBE1 gene that affects 8 to 10 percent of

quarter horses. Paints, Appaloosas and other descendent breeds of quarter horses can be affected. GBED is a fatal condition that causes the inability to properly store sugar. Since it is a recessive disease, a horse must have two copies of the mutated gene to express the condition. The test has three potential results:

1. Gb/Gb. This means your horse has two copies of the GBED mutation. It is affected with GBED and will not live to pass on disease to offspring.
2. n/Gb. This means your horse is a carrier of the disease and has one copy of the GBED mutation. If bred, this horse has a 50 percent chance of passing the disease on to its offspring.
3. n/n. This means your horse is not affected by GBED and will not pass it on to its offspring.

Horses that inherit GBED are incapable of producing the enzyme needed to connect glycogen structures that prevent the horse from storing sugar. This prevents the horse from storing energy needed to fuel its organs, muscles and brain. Foals born positive for GBED display a wide range of symptoms including weakness, difficulty standing, low body temperature, seizures, contracted muscles and death. GBED commonly results in second and third trimester abortions and stillborn foals. Foals that survive foaling and not euthanized often die within eight weeks. Homozygous GBED is always fatal, and carrier animals should not be bred.

Understanding the genetic status of your horses is an important tool to use for management and breeding decisions. Fortunately, many labs exist and offer affordable genetic panels that can help you make an informed decision. If you have any question regarding your horse's genetic status, contact your local veterinarian.

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