

There are now at least eight commercially available genetic tests for inherited diseases in horses. This presentation will cover these diseases and use of genetic tests in breeding programs.

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Conflict of interest statement; Drs Valberg, Mickelson and McCue hold the patent for the PSSM test and receive royalties.

The selective breeding of animal populations may give rise to a common founder that can disseminate a genetic trait to many thousands of related offspring within a few years. A DNA mutation has been identified in only 8 equine genetic diseases to date (Oct 2007), however , there will likely be many more identified in the near future with the rapid development of genetic tools specific for horses. Five of the 8 genetic diseases are found in Quarter Horses. In our opinion this breed distribution reflects the larger number of Quarter Horses in the USA relative to other breeds, a tendency to line breed, and the openness and dedication of the Quarter Horse Association to finance and support investigation into equine genetics.

The 5 known genetic mutations in Quarter Horses include Hyperkalemic Periodic Paralysis (HYPP), Glycogen Branching Enzyme Deficiency (GBED), Hereditary Equine Regional Dermal asthenia (HERDA), Polysaccharide Storage Myopathy (PSSM) and Malignant Hyperthermia (MH). Three other genetic diseases with known mutations not affecting Quarter Horses are Severe Combined Immunodeficiency (SCID) in Arabians (Vetgen does testing) and Overo Lethal White Syndrome (OLWS) in Paints (UC Davis does testing), and Junctional Epidermolysis

Bulosa (JEB) in Belgians (UCDavis does testing).

The pattern of inheritance of these traits is either autosomal dominant or autosomal recessive.

Autosomal dominant traits: Require only one copy of the mutant gene to cause disease. Breeding of an affected heterozygous horse (one copy of the defective gene) to a normal horse results in a 50% chance of producing an affected horse. Breeding two heterozygous affected horses has a 50% chance of producing a heterozygous affected, 25% chance of a homozygous affected (2 copies of the defective gene) and a 25% chance of a homozygous normal being born.

Autosomal recessive traits: Requires two copies of the mutant gene to cause disease. Breeding two affected horses results in a 100% chance of producing an affected horse. Breeding two carriers results in a 25% chance of producing an affected horse, a 25% chance of a normal horse and a 50% chance of a carrier. Breeding an affected and a carrier results in a 50% chance of producing an affected horse and a 50% chance of producing a carrier.

We are continuing to work on which bloodlines are affected. This summary contains some preliminary results on the types of Quarter horses affected by these diseases.