



# Interpreting Equine Genetic Defect Testing Results

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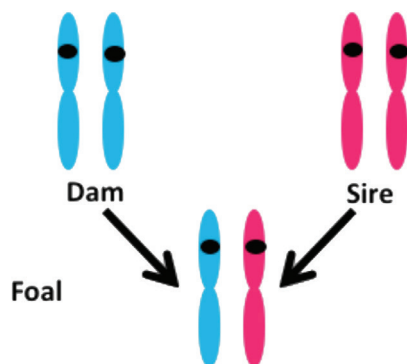
The American Quarter Horse Association (AQHA) has genetic tests available to test horses for five genetic defects. In this fact sheet, we will discuss how these defects are inherited, how to understand the genetic testing results received from AQHA and Animal Genetics, Inc., and the probability of producing an affected horse.

## Inheritance

The equine genome is made up of 31 pairs of autosomal chromosomes plus the sex chromosomes. Genetic defects are a result of alleles that cause a lethal condition or that severely handicap the performance of an individual. Simply speaking, at any location in the genome where there is a mutation (a change in the DNA sequence), there are two alleles, which can be thought of as alternate forms of a gene. Each individual has two alleles (found on a chromosome): one inherited from the sire and one inherited from the dam (Figure 1). The mode of inheritance describes if an allele is dominant, which means they mask the effects of other allele, or recessive, whose effect can be hidden by the dominant allele. The five defects tested for by the AQHA have either an autosomal dominant or autosomal recessive inheritance. In their testing results, they display a normal allele as n or N. They display a disease allele as d or D.

## Autosomal Dominant

An autosomal dominant defect means that the mutation is found on one of the 31 autosomal chromosomes (not the sex



**Figure 1.** One Chromosome (pink or blue), which contains alleles (black dots), is inherited from each parent.

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chromosomes) and will mask normal alleles. An autosomal dominant mutation only requires one copy of the dominant allele (D) to produce an affected horse, so affected horses can either be heterozygous (normal/Defect) or homozygous for the defect (Defect/Defect). Unaffected animals will always be homozygous normal (normal/normal). Hyperkalemic Periodic Paralysis (HYPP), Malignant Hyperthermia (MH), and Polysaccharide Storage Myopathy (PSM) are examples of autosomal dominant diseases tested for by AQHA.

## Autosomal recessive

The term autosomal recessive means the mutation is on one of the 31 autosomal chromosomes (not a sex chromosome), and it is not expressed unless the animal receives two copies of the defective allele (one on each of the chromosomes inherited from its parents). Because these traits are recessive, possessing only one copy of each of these disease alleles is not enough to change the phenotype, because it is masked by a dominant normal allele. Affected animals will always be homozygous for the disease allele (defect/defect). Glycogen Branching Enzyme Deficiency (GBED) and Hereditary Equine Regional Dermal Asthenia (HERDA) are autosomal recessive diseases tested for by AQHA.

## Reading genetic test results

AQHA and Animal Genetics, Inc. report the same information for each allele inherited by an animal, which is called a genotype. Both tests report the number of normal alleles (N or n, depending on the test) inherited by an animal. If an animal has 2 normal alleles for a defect (NN or nn, depending on the test), they are homozygous normal, are unaffected by the disease, and cannot pass on that disease allele to their progeny. If they are homozygous for the disease allele (Defect/Defect) they will always be affected and will always pass that disease allele on to their progeny. Where the two tests differ slightly is in the interpretation of the results for animals that are heterozygous (have one normal allele and one allele for a defect).

The AQHA reports the results of their genetic tests using all capital letters. For autosomal dominant diseases (HYPP, MH, and PSM), if your animal is heterozygous (N/Defect) or homozygous for the disease allele (Defect/Defect), they will be affected. Additionally, they can pass this disease allele on to their offspring. For autosomal recessive diseases

(HERDA, GBED), heterozygous animals are denoted as N/Defect, and can pass on the disease allele to its offspring, but the animal will not be affected itself. These animals are often times referred to as carriers.

The Animal Genetics, Inc. test makes it easy to identify the mode of inheritance because they use a combination of upper and lowercase letters to denote the type of inheritance of each defect. With their results, a capital letter means a dominant allele (and you only need one to see that phenotype, either normal or disease) and a lowercase letter means a recessive allele (you need two copies to see that phenotype, either normal or disease). It is important to remember that the mode of inheritance describes the inheritance for the defective allele, so if a disease is recessive, the normal allele is dominant and vice versa. Therefore, normal alleles will be lowercase (n) for autosomal dominant defects and uppercase (N) for autosomal recessive defects. A heterozygous animal will be affected if the defective allele is paired with a lowercase n (n/D=affected because the defect is dominant) and will be a carrier if it is paired with a capital N (N/d=unaffected carrier because the defect is recessive).

## Determination of Risk

Knowing the genotype of an animal for a genetic defect allows us to determine the risk of producing an affected foal from mating two animals. An animal passes on one of the two alleles it possesses. As a result, a homozygous normal animal (denoted as either nn or NN, depending on the test and mode of inheritance) will pass on normal alleles to its

progeny 100 percent of the time. Conversely, a homozygous affected animal (d/d or D/D) will pass on disease alleles to its progeny 100 percent of the time. Heterozygous animals pass on a defective allele 50 percent of the time and a normal allele 50 percent of the time. The following scenarios illustrate your risk of producing affected progeny (D & d stand for either dominant or recessive disease alleles, respectively). It is important to note that these numbers are what we expect to see, on average, with a large number of matings (i.e. more than 100 matings). It is possible to see different percentages with a small number of matings.

### Autosomal dominant:

$n/D \times n/n = 50\%$  affected

$n/n \times n/D = 50\%$  affected

$n/D \times n/D = 75\%$  affected

any D/D mating =  
100% affected

### Autosomal recessive:

$N/d \times N/N = 0\%$  affected, 50% carriers

$N/d \times N/d = 50\%$  carriers, 25% affected

$d/d \times N/N = 100\%$  carriers

$d/d \times N/d = 50\%$  affected, 50% carriers

Knowing the genotypes of the animals you are mating and how genetic defects are inherited can assist in making good management decisions both to preserve genetic progress and reduce or eliminate economic losses associated with genetic defects. In addition, awareness of genetic defect testing and the risk of producing affected progeny will help in analyzing risk and managing the incidence of genetic defects, which will improve the health and welfare of the animals in your herd.

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