GENETIC DISEASE RESULTS AND DESCRIPTIONS

Genes come in pairs; each parent contributes one copy to the offspring. With respect to the autosomal dominant genetic diseases listed below (HYPP, PSSM, and MH), only one defective gene is necessary to express the genetic disease in question. Such gene can be inherited from either of the parents or from both of the parents. With respect to autosomal recessive genetic diseases listed below (GBED and HERDA), typically, two copies of an abnormal gene (two copies = a pair) must be present in order for the horse to show signs of the genetic defect in question. Horses with only one copy of the defective gene are considered "carriers".

Effects of equine diseases

GBED – Glycogen Branching Enzyme Deficiency. Fatal disease of newborn foals caused by defect in glycogen storage. Affects heart and skeletal muscles and brain. Inherited as a recessive gene.

Possible results:

N/N	Normal - does not possess the disease-causing GBED gene.
N/G	Carrier - Heterozygous (one normal and one GBED gene).
G/G	Affected – horse has two copies of the GBED gene.

HERDA – Hereditary Equine Regional Dermal Asthenia. Skin disease characterized by hyperextensible skin, scarring, and severe lesions along the back of affected horses. Typical onset is around 2 years of age. Inherited as a recessive disease.

Possible results:

N/N	Normal - horse does not have the HERDA gene.
N/HRD	Carrier - horse carries one copy of the HERDA gene.
HRD/HRD	Affected - horse has two copies of the HERDA gene.

HYPP – Hyperkalemic Periodic Paralysis. Muscle disease caused by defect in sodium channel gene that causes involuntary muscle contraction and increased level of potassium in blood. Inherited as a dominant disease. Two copies of the defective gene produce more severe signs than one copy.

Possible results:

N/N	Normal - does not possess the disease-causing HYPP gene.
N/H	Hyperkalemic - Heterozygous (one normal and one HYPP gene).
H/H	Affected - horse has two copies of the HYPP gene.

MH – Malignant Hyperthermia. Rare but life-threatening skeletal muscle disease triggered by exposure to volatile anesthetics (halothane), depolarizing muscle relaxants (succinylcholine), and stress. Presumed inheritance as a dominant disease.

Possible results:

N/N	Normal - horse does not have the MH gene.
N/MH	Affected - horse has one copy of the MH gene.
MH/MH	Affected - horse has two copies of the MH gene.

PSSM1 – Polysaccharide Storage Myopathy Type 1. Muscle disease characterized by accumulation of abnormal complex sugars in skeletal muscles. Signs include muscle pain, stiffness, skin twitching, sweating, weakness and reluctance to move. Inherited as a dominant disease.

Possible results:

N/N	Normal - horse does not have the PSSM1 gene.
N/PSSM1	Affected - Horse has one copy of the PSSM1 gene.
PSSM1/PSSM1	Affected - horse has two copies of the PSSM1 gene.