Heritable Equine Regional Dermal Asthenia

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KEYWORDS

• Quarter Horse • Hereditary • Skin tearing • Collagen • HERDA • DNA testing

KEY POINTS

- Hereditary equine regional dermal asthenia (HERDA) is a form of Ehlers-Danlos syndrome that is most commonly seen in Quarter Horses, Appaloosas and American Paints.
- HERDA has an autosomal recessive mode of inheritance; DNA testing can establish normal, carrier and affected status.
- Affected horses are typically born normal and develop lesions within the first 2 years of life.
- More severely affected horses experience spontaneous skin sloughing and extensive lacerations, hematomas, and seromas from minor trauma, frequently resulting in disfiguring scars.
- Affected horses have a higher than expected incidence of corneal ulcers.
- Palliative therapy is available, but no curative treatment exists.

INTRODUCTION

Hereditary equine regional dermal asthenia (HERDA) is an autosomal recessive disorder of collagen observed primarily in Quarter Horses. It is a form of Ehlers-Danlos syndrome (EDS), which occurs in man and a variety of other species, such as cattle, sheep, dogs, cats, and mink. Ehlers-Danlos syndrome is a group of genetically heterogeneous connective tissue disorders that result from mutations in genes encoding various collagen types, enzymes that modify collagens, and other extracellular matrix proteins. When EDS occurs in animals, it is known by a variety of names, including *dermatosparaxis*, *cutaneous asthenia*, and *hyperelastosis cutis*.

HERDA has been extensively reported in the veterinary literature, beginning in 1978.[1–20] Because of the common practice of line breeding carrier horses, HERDA has recently become one of the most commonly reported inherited diseases in the

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equine industry, and has been reported in Quarter Horses, Appaloosas, and American Paints in several countries, including the United States, Canada, Mexico, Brazil, England, France, and the Netherlands.

AUTOSOMAL MODE OF INHERITANCE

Heterozygous (N/Hrd) horses are carriers of the recessive Hrd allele, yet appear normal, without cutaneous signs of the disease. Horses that are homozygous for the recessive allele (Hrd/Hrd) are physically affected, and exhibit hyperextensible skin, cutaneous lesions, skin sloughing, and characteristic scarring. **Fig. 1** shows the typical pattern of inheritance for HERDA. When a carrier mare (N/Hrd) is bred to a carrier stallion (N/Hrd) there is a 25% chance that the foal will have HERDA (Hrd/Hrd), a 50% chance that the foal will be a carrier (N/Hrd) and a 25% chance that the foal will be totally normal (N/N). When a normal mare or stallion is bred to a carrier stallion or mare, a 50% chance exists that the foal will be a carrier, but a 0% chance that the foal will be affected. When an affected mare or stallion (Hrd/Hrd) is bred to a normal mare or stallion (N/N), all the foals will be carriers (N/Hrd). An affected foal can never be produced from a normal parent. Both parents must carry at least one copy of the HERDA gene.

SIGNIFICANCE WITHIN THE QUARTER HORSE INDUSTRY

Although cutting horses are most commonly affected, pleasure, reining, working cow, and foundation bred horses are also affected. In cutting horses alone, 3 of the 10 leading lifetime sires (leading lifetime cutting horse sires, all ages, all divisions[6]) are confirmed carriers. As of 2012, these 3 sires alone have produced 5792 total registered offspring, including performing and nonperforming offspring. By virtue of HERDA's autosomal recessive mode of inheritance, one-half of these offspring (2896) are HERDA carriers or affected animals. The total lifetime earnings of these 3 carriers is $109,008,304; an average earning of $31,583 per performance offspring.

Carrier to carrier mating:

	N	Hrd
N	N/N	N/Hrd
Hrd	N/Hrd	Hrd/Hrd

N = Normal allele Hrd = Abnormal allele

Normal horse mated to carrier:

	N	N
N	N/N	N/N
Hrd	N/Hrd	N/Hrd

N = Normal allele Hrd = Abnormal allele

Fig. 1. Typical pattern of inheritance for HERDA showing matings of carrier to carrier and carrier to normal horse.