Characterization of a New Inherited Ocular Disease in Rocky Mountain Horses

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Anterior segment dysgenesis syndrome is an inherited ocular defect in Rocky Mountain Horses. The disease is Mendelian with semidominant inheritance. Heterozygous animals have ciliary cysts, and homozygous animals have complex anterior segment dysgenesis. Authors’ address: Depts. of Large Animal Clinical Sciences (Ewart) and Small Animal Clinical Sciences (Ramsey), D202 Veterinary Medical Center, Michigan State University, East Lansing, MI 48824-1314. © 1998 AAEP.

1. Introduction
The Rocky Mountain Horse is a new breed that descends from the Mountain Pleasure Horse. This breed is rapidly gaining popularity, and the number of registered Rocky Mountain Horses has more than doubled in recent years. Many Rocky Mountain Horses are descendants of a single Mountain Pleasure Horse founder sire. The use of limited foundation stock and intensive line breeding has resulted in the widespread propagation of a heritable ophthalmic defect in this breed.1 The abnormalities are associated with developmental defects of the anterior segment of the eye.1 Abnormalities are variable and involve anterior segment structures along with the lens, ciliary body, and retinal epithelium. In similarly affected humans and mice, this syndrome is termed anterior segment dysgenesis.1 Anterior segment dysgenesis has not been previously described in Rocky Mountain Horses; therefore, many practitioners are unfamiliar with the disease and appropriate management recommendations. We have performed studies to classify the familial ocular lesions in Rocky Mountain Horses and to characterize the mode of inheritance of this syndrome.

2. Materials and Methods
We characterized the ophthalmic abnormalities in a large population of Rocky Mountain Horses that represented eight generations of an extended family. Physical and ophthalmic examinations were performed on all horses. Ophthalmic examinations included applanation tonometry; direct, focal, and diffuse slit-lamp biomicroscopy; and indirect ophthalmoscopy. To determine the hereditary basis of this disease, we collected and analyzed pedigrees from all the Rocky Mountain Horses that were examined.

3. Results
The most common abnormality of Rocky Mountain Horse eyes was large cysts arising from the temporal ciliary body or retina. An additional syndrome of multiple abnormalities of the anterior segment, lens, and retina was also noted. These lesions were consistent with anterior segment dysgenesis. Anterior segment abnormalities were only detected in horses that had ciliary cysts. The majority of the horses affected with cysts or anterior segment dysgenesis could be traced to one sire (generation IV: individual 7) who was a founda-
tion sire of the breed. The immediate progeny of IV:7 demonstrated cysts or a normal ocular phenotype. No immediate progeny of IV:7 had anterior segment dysgenesis. Individual IV:7 was not examined.

Genotypes were predicted based on the ocular phenotypes observed and on the potential gene dose (one or two) obtained from IV:7. The mode of inheritance appeared to be semidominant, with the cyst phenotype being expressed in heterozygous animals and the anterior segment dysgenesis phenotype being expressed in the homozygous state. Of animals with a genotype predicted to be heterozygous for the locus controlling cysts, 3% were phenotypically normal and thus were described as having incomplete penetrance.

4. Discussion

From these studies we concluded that cysts and anterior segment dysgenesis occur consistently and reproducibly in Rocky Mountain Horses. These phenotypes were documented throughout a large population of Rocky Mountain Horses, and the uniformity of observed lesions suggests a common cause for these abnormalities. Our results support the hypothesis that these lesions are inherited in a semidominant manner, with cysts being expressed in the heterozygous state and complex anterior segment dysgenesis lesions being expressed in the homozygous state. The disproportionate number of affected horses within an extended pedigree suggests the possibility that the disease is attributable to a common mutation inherited from a shared ancestor. To our knowledge, our assessment of the prevalence, epidemiology, and genetic model fitting of this disease in Rocky Mountain Horses provides the first documented evidence of the heritability of this condition in horses. Our long-term objective is to develop a DNA test to determine the genotype of affected animals, especially carrier animals with incomplete penetrance. A careful ophthalmic examination along with supportive DNA testing, when available, will be important tools for the selection of breeding stock in pedigrees in which these defects occur.

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References and Footnotes

