

# Genetic Testing as a Tool to Identify Horses with or at Risk for Ocular Disorders



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## KEYWORDS

- Genetics • Ocular disorders • Night blindness • Recurrent uveitis
- Squamous cell carcinoma • Multiple congenital ocular anomalies • Horses

## KEY POINTS

- Genetic testing can help identify horses with ocular disorders and tests are available for congenital stationary night blindness and multiple congenital ocular anomalies.
- Genetic testing can also help identify horses at risk for developing ocular disorders, including equine recurrent uveitis and squamous cell carcinoma.
- Screening horses for genetic mutations can help inform management decisions for an earlier diagnosis and better prognosis.

## INTRODUCTION

Sequencing the genome (the totality of an organisms DNA) of a gray thoroughbred mare named Twilight has allowed the rapid development of tools and resources to aid in the understanding of the genetics of economically and medically important traits in the horse.<sup>1</sup> Twilight's genome has served as a reference with which to compare sequence information from other horses and has enabled finding the causative mutations for several disorders, including those that can affect ocular function. These findings allow the creation of genetic tests offered by several commercial laboratories around the globe. Use of these tests can influence clinical management and breeding decisions.

Most inherited ocular disorders or those disorders with ocular manifestations are breed specific or affect breeds that are closely related. The genetics of 8 different ocular disorders have been investigated (**Table 1**). Approximately half of the disorders studied have an associated or causative variant identified. These disorders include congenital stationary night blindness (CSNB) and equine recurrent uveitis (ERU) in

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Disclosure: R.R. Bellone is affiliated with the Veterinary Genetics Laboratory, a genetic testing laboratory offering diagnostic tests in horses and other species.

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Table 1 Inherited ocular disorders				
Ocular Disorder	Alias Names	Breeds	DNA Test Available and Name	Important Clinical Considerations
Congenital stationary night blindness	CSNB	Appaloosa, American miniature horse, Knabstrupper, Noriker, Pony of the Americas	Yes: LP	All <i>LP/LP</i> horses are night blind
Equine recurrent uveitis	Moon blindness/ ERU	Appaloosa Warmblood breeds	Yes: LP No	In the Appaloosa breed risk for ERU is defined as <i>LP/LP &gt; LP &gt; N &gt; N/N</i>
MCOA	Anterior segment dysgenesis/ MCOA	Rocky Mountain horse, Kentucky mountain saddle horse, Kentucky mountain pleasure horse, Icelandic horse, Shetland pony, American miniature horse, Comtois, and other breeds with silver, including American quarter horse and Morgan	Yes: silver	<i>Z/Z</i> horses typically have a more severe phenotype than <i>Z/N</i>
Limbal SCC	Ocular SCC	Haflinger	Yes: ocular SCC	<i>R/R</i> 5.5 times more likely to develop ocular SCC than <i>R/N</i> or <i>N/N</i>
Cataracts		Exmoor pony, American quarter horse, Arabian, thoroughbred, and Morgan	No	Females seem to be more frequently affected in the Morgan and Exmoor breeds
Corneal dystrophy		Friesian	No	May be related to other collagen disorders in the breed
Distichiasis		Friesian	No	Unknown mode of inheritance
Aniridia		Belgian and American quarter horse	No	Thought to be inherited as a dominant mutation so offspring of affected horses should be carefully monitored

Abbreviations: CSNB, congenital stationary night blindness; MCOA, multiple congenital ocular anomalies.

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