

When veterinary ophthalmologist Dr. Lynne Sandmeyer began her investigation of *congenital stationary night blindness* (CSNB) in Appaloosa horses nearly three years ago, there was plenty of speculation, theories and anecdotes that associated the disease to particular coat patterns in the Appaloosa breed — but not much else in terms of solid veterinary research.

Now, findings from the Western College of Veterinary Medicine (WCVM) study show a definite association between the eye disease and the *Lp* (leopard complex) gene that's responsible for "turning on" the breed's spotting coat patterns. The study results were published in the November 2007 issue of *Veterinary Ophthalmology*.*

"All 10 of the homozygous *Lp* (few-spot or snowcap-patterned) horses in our study have been diagnosed with CSNB, but none of the other horses has the disease," reports Sandmeyer, an associate professor in WCVM's Department of Small Animal Clinical Sciences. "It's a relatively small sample, but the evidence seems conclusive. Whether the problem is with the *Lp* gene itself or with a nearby gene that's linked to it — we don't know that yet."

Sandmeyer and her research collaborators selected 30 purebred Appaloosa horses that displayed three different coat patterns found within the breed to participate in the two-year study. Ten horses had the "few spot" or "snowcap" pattern (homozygous for the *Lp* gene, or carrying two copies of the gene), 10 animals were "leopard" or "blanket spotted" (heterozygous *Lp*, or carrying one copy), and 10 were solid-coloured (not carrying the *Lp* mutation).

Sandmeyer thoroughly examined the eyes of all of the horses and conducted *electroretinographic* (ERG) testing to confirm the presence or absence of CSNB. Tissue and blood samples were also taken from each of the horses.

Confirming the association between coat patterns and genetics is just one aim of the study; determining where the problem lies and how to fix it are also important goals of the investigation that's backed by the College's Equine Health Research Fund.

"The eyes of the affected horses appear completely normal, but the ERG testing shows that there's a disruption somewhere in the signal transmission. Information is not getting through from the horses' eyes to their brains," explains Sandmeyer. Although it's still not clear where the broken link in the signal transmission occurs, she suspects the problem might be with a receptor, and that transmembrane proteins may be involved.

As for other aspects of the study, it will take about a year to complete the immunohistochemical analysis of the horses' tissue samples — testing that will help to pin down any physiological differences between the eyes of normal horses and the eyes of horses diagnosed with CSNB.

Sandmeyer's CSNB study is part of a much larger, North America-wide initiative called the Appaloosa Project that's exploring the genetic makeup of the equine breed. Dr. Rebecca Bellone, a genetic researcher from the University of Tampa and one of the Appaloosa Project's key collaborators, is responsible for analyzing the horses' blood samples.

"We'll start our investigation into the puzzle at this end, they'll start at the other, and we hope to meet somewhere in the middle with a few of the answers," says Sandmeyer.

Once researchers have pinpointed what causes the transmission problems and the gene responsible for the disease has been identified and located, then scientists can focus on developing potential gene therapy for CSNB.



By Roberta Pattison

Research Sheds New Light on NIGHT BLINDNESS

Several types of night blindness have been identified in people, and it's likely that one or more of these types share similarities with CSNB in horses, says Sandmeyer. "We can learn from the work that's been going on in human medicine, and they can learn from studies such as ours. Information goes both ways."

Meanwhile, Sandmeyer is interested in examining and identifying other horses afflicted with CSNB. It only takes about 40 minutes to conduct the physical examination and the testing required for diagnosing the disease — and she has already found a few more CSNB-affected horses. She suspects that part of the reason why the disease has gone unnoticed for so long is because it's seldom diagnosed: many horse owners simply have no idea that their animals can't see in the dark.

"Horses are adaptable creatures, and when they live from birth with a condition like this, they learn to cope. As well, people aren't around their horses much at night so often aren't aware that there's a problem," explains Sandmeyer, who welcomes inquiries from veterinarians or horse owners about the disease.

"This is an important problem that affects probably a quarter of the Appaloosa population. It's also a manageable disorder. Affected horses get by very well as it is, but once you know your horse has CSNB, you can make life a little easier for him." **H**

*Sandmeyer LS, Breaux CB, Archer S, Grahn BH. "Clinical and electroretinographic characteristics of congenital stationary night blindness in the Appaloosa and the association with the leopard complex." *Veterinary Ophthalmology*. 10(6): 368-375.

Visit www.ebrf.usask.ca to view the Fall 2005 issue of *Horse Health Lines* that includes more background information about WCVM's investigation of CSNB. Contact Dr. Lynne Sandmeyer for more details (306-966-1336; lynne.sandmeyer@usask.ca).

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