Eye disease in horses
Investigator: Dr. Mark Rutherford
University of Minnesota
Status: Nearly complete

Dr. Mark S. Rutherford received his PhD in immunology from the University of Illinois and is an associate professor in the department of veterinary and biomedical sciences at the University of Minnesota. His postdoctoral research fellowship was done through the department of biochemistry at St. Jude Children's Research Hospital in Memphis, Tenn.

What is equine recurrent uveitis? What are the symptoms?
Dr. Rutherford: ERU is an inflammatory disease of the eye, usually progressing slowly and in repeated episodes in one or both eyes. The inflammation leads to an autoimmunity against proteins in the eye. Symptoms include tearing (lacrimation), clouding of the eye, decreased ocular pressure, photophobia, and synechia, which is an adhesion between the iris and the lens or cornea.

How common is it?
Dr. Rutherford: ERU is thought to affect up to 10 percent of horses.

How is it diagnosed and traditionally treated? How effective is treatment?
Dr. Rutherford: An eye exam provides the best diagnosis, particularly early in the disease. Corneal edema, aqueous flare, and synechia are characteristic signs. Treatments usually begin with anti-inflammatory drugs to mute the autoimmune process, and often pain killers are included. As damage to the eye proceeds, often removal of the eye is necessary. There are several newer but highly invasive approaches, including implantation of drug pumps (to regulate immune cells), and vitrectomy where the contents of the eye are drained and replaced with saline. While these approaches show promise, both are very expensive options.

What do you hope to learn from your study?
Dr. Rutherford: First, we will catalog which alleles, or versions of specific genes, are present in appaloosas. Then, by comparing the frequency of specific gene changes between affected and normal horses, we hope to identify any alleles with a skewed frequency in ERU cases. This would suggest a strong link to ERU.

Results so far that you wish to share?
Dr. Rutherford: Our work to date shows that several of the genes we are sequencing have a high degree of polymorphism, meaning that many versions exist in the equine population. This is to be expected for the type of genes we are examining. Our

preliminary test also suggests a genetic contribution of one or more of these genes to ERU. We will complete sequencing for three of the genes in our equine population soon. Additional follow up studies using microsatellite genetic markers so far confirm our initial results.

Why should the average animal lover care about your study?

Dr. Rutherford: While devastating for any companion animal species, blindness in horses is perhaps even more difficult for the animal and the owner. Horses don't utilize the strong olfactory cues that a dog might use, and it becomes nearly impossible to ride a blind horse. Since the disease is so widespread and likely has a strong genetic component, it is our responsibility to identify the contributing genetic factors and remove those carrier animals from the breeding population. This is quite similar to approaches in other animal species and in other equine genetic diseases to lower the incidence of disease.