Definitive Antemortem And Postmortem Diagnostics For NAD

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The goal of this study is to develop diagnostic tests for eNAD/EDM, Equine Neuroxoanal Dystrophy/Equine Degenerative Myeloencephalopathy.

Equine neuroaxonal dystrophy/degenerative myeloencephalopathy (eNAD/EDM) is an inherited neurologic disease in horses. Both a genetic susceptibility and a vitamin E deficiency during the first year of life are required for a foal to develop eNAD/EDM. However, despite many years of extensive research by our laboratory and others, no causative genetic mutations have been discovered.

One of the likely reasons for the lack of progress is the fact that we have never been able to investigate the genetics of eNAD/EDM within one family of horses that were raised in the same environment. In the spring of this year, we were contacted by a breeding farm in Texas that had half of their yearlings and two-year old horses demonstrating the incoordination observed with eNAD/EDM, while the other half were normal. All young horses were vitamin E deficient and only two sires were used on this farm, with both producing affected and normal foals.

Through a new collaboration with the Texas Veterinary Diagnostic Laboratory, we were able to confirm eNAD/EDM after death in 9 young horses. The remaining 8 young horses were neurologically normal or only very mildly affected. Blood samples for DNA were collected from all young horses and their dams and sires. With this unique cohort of related horses, all raised in the same vitamin E deficient environment, we propose to develop tests for eNAD/EDM that can be used while a horse is still alive and other tests that could be used after a horse has been euthanized.

The reason for needing two types of tests is that a genetic test could be specific to only one family or breed of horse (i.e. Quarter Horse) while a “biomarker” test after euthanasia would likely encompass all cases of eNAD/EDM.

Therefore, we will leverage this unique sample of horses to perform DNA sequencing to try and identify a genetic marker of eNAD/EDM while, at the same time, using tissue samples from these horses to identify biomarkers of eNAD/EDM that we can use to diagnose the disease after death in tissue samples.

The overall goal is to develop highly accurate diagnostic tests for this disease.

Importance to Industry: Since the 1970’s, the top three causes of spinal incoordination in the horse remain unchanged. These include cervical vertebral compressive myelopathy (CVCM, “Wobblers disease”), equine neuroaxonal dystrophy / equine degenerative myeloencephalopathy (eNAD/EDM) and equine protozoal myeloencephalitis (EPM). Tests for CVCM now include CT myelography, and blood and spinal fluid tests are available for EPM. In contrast, eNAD/EDM remains a diagnosis of exclusion, with definitive confirmation requiring detailed evaluation after the horse has died.

Our laboratory is committed to identifying a genetic cause for eNAD/EDM. Despite extensive investment of time and money, the genetic basis for eNAD/EDM is still unsolved. The unique family of Quarter horses that we have identified in Texas will allow, for the first time, a genetic study within a small family of horses with eNAD/EDM. Diagnostic tests that can be used while the horse is still alive and after death for eNAD/EDM can be developed from this cohort of horses and provide a definitive diagnosis for this highly prevalent cause of incoordination in horses.

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