Equine neuroaxonal dystrophy (eNAD) is an inherited neurodegenerative disease characterized by early onset ataxia and histologic lesions within the cerebral brainstem and spinal cord. The clinical and histologic features of eNAD resemble an inherited neurodegenerative condition in humans known as ataxia with vitamin E deficiency (AVED). AVED was once referred to as Friedrich’s Ataxia with isolated Vitamin E deficiency because the neurologic deficits associated with Friedrich’s Ataxia are indistinguishable from those of AVED. Cardiomyopathy is reported in the majority of Friedrich’s Ataxia patients, contributing to death in 83.3% of cases in one postmortem study. The goal of this project is to compare the cardiac structure and function evaluated by echocardiography and electrocardiography in Quarter horses with eNAD and unaffected breed-matched controls. Characterizing the cardiac phenotype of horses affected by eNAD will improve understanding of the systemic effects of this complex heritable condition.

Hypothesis
Quarter horses with eNAD exhibit subclinical cardiomyopathy that manifests as myocardial thinning and a higher prevalence of arrhythmias.

Methods
Horses for this study were selected from the Center for Equine Health (CEH) herd at UC Davis. Cases included 7 eNAD suspect Quarter horses and 7 age and breed-matched controls.

Electrocardiography: Cardiac Electrophysiology

Blood work: Cardiac Troponin I
Blood drawn at the time of each horse’s ECG was evaluated at the University of Pennsylvania for cardiac troponin I quantification.

Echocardiography: Cardiac Structure and Function
Echocardiography was performed with the Vivid IQ ultrasound system, using 6 standardized equine echocardiographic views in short and long axis and a long axis 2 chamber view from the left.

Statistics
Data was assessed for normally using a Shapiro-Wilk test, and most variables were compared between cases and controls with a student’s T-test or Mann Whitney U test, where appropriate. Arrhythmia prevalence data was compared between cases and controls using a chi-squared test.

Conclusions and Future Directions
Cardiac structure: Horses in the eNAD group had a significantly decreased left ventricular free wall thickness in diastole (p=0.024) and significantly decreased relative wall thickness (p=0.047).

No significant differences were detected between the two groups in:
- Cardiac electrophysiology
- Cardiac troponin I levels
- Cardiac function

Future directions: Identify evidence of changes in left ventricular mass, and accumulation of oxidative damage to DNA and apoptosis in the myocardium of horses with eNAD submitted for post-mortem evaluation compared to breed-matched controls.

References