

## 61<sup>st</sup> ANNUAL DIAGNOSTIC SLIDE SESSSION 2020.

### CASE 2020-4

#### **Submitted By:**

Lucy P. Evans BS, Edwin Stone MD, PhD, Robert Mullins PhD, Budd Tucker PhD, Katherine Gibson-Corley DVM, PhD, Karra A. Jones MD, PhD  
The University of Iowa, Department of Pathology, Iowa City, IA 52242

#### **Clinical History:**

The patient initially presented in childhood with visual problems and was diagnosed with retinitis pigmentosa that progressed to clinical blindness. She started experiencing CNS symptoms in her early 20s including word finding difficulties and memory loss followed by generalized tonic-clonic seizures. Brain MRI showed generalized cortical atrophy of unknown etiology. Her CNS symptoms continued to progress until her death at 27 years of age. She had a cousin diagnosed with retinitis pigmentosa in her 20s, but no other pertinent family history.

**Autopsy Findings:** An autopsy limited to brain and eyes was performed. Gross evaluation of the brain showed generalized cortical atrophy with a weight of 950 grams. Sectioning revealed a mildly thin cortical ribbon and highly atrophic bilateral lateral geniculate nuclei.

#### **Material Submitted:**

Gross images of the brain  
Gross image of coronal section including basal ganglia  
Gross image of coronal section including thalami/hippocampi  
H&E-stained section of the hippocampus

#### **Points for Discussion:**

1. Differential diagnosis
2. Ancillary studies and molecular analysis
3. Pathogenesis