63rd ANNUAL DIAGNOSTIC SLIDE SESSION 2022

CASE 2022-4

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Clinical History:
This 40-year-old woman had exercise intolerance and difficulty keeping up with her peers since childhood. As teenager, she was noted to have limited eye movements and developed progressive limb weakness. The patient and her mother (asymptomatic at time of testing) were found to have a heterozygous *POLG* mutation (c.2740A>C, p.Thr914Pro).

In her 30s, she developed episodes of right hemibody sensory symptoms and ipsilateral hemiparesis with associated epileptogenic activity. She was initiated on anti-epileptics for partial seizures with good control of the symptoms.

In her 40s, she experienced an episode of visual changes. An MRI demonstrated T2 signal change and enhancement centered at the right temporoparietal occipital junction and a suprasellar mass, measuring approximately 1.9 cm. Four months later, she developed sudden onset dysarthria preceded by intermittent right lower limb numbness. An MRI demonstrated diffusion restriction in the right parietal lobe and T2 cortical signal changes within the right temporoparietooccipital junction and a stable appearance of the suprasellar mass. She subsequently developed status epilepticus and acute hypoxic respiratory failure requiring intubation. Due to her declining status, she was transitioned to comfort care measures and died.

Autopsy findings:
At the time of autopsy, she was found to have acute and organizing bronchopneumonia. The brain appeared normally developed with mild generalized swelling; brain weight 1188 grams (fixed). There was patchy mottling of the bilateral cerebral cortices. A lobular and cystic white-tan mass was adherent to the base of the brain overlying the optic chiasm, 1.9 cm in greatest dimension.

Material Submitted:
1 H&E right lateral occipital lobe.

Points for Discussion
1. Differential Diagnosis
2. Clinical, pathologic, and molecular correlation