

46th ANNUAL DIAGNOSTIC SLIDE SESSION 2005.

CASE 2005-1

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Clinical History: This 2½ year-old girl presented to the Emergency Department in partial complex status epilepticus with shaking of the right arm and leg. Initial CT and MRI scans of the brain were unremarkable. Laboratory studies, including assays for lactate, very long chain fatty acids, amino acids, ammonia, ceruloplasmin, viral serologies, and spinal fluid studies were unremarkable. Pyruvate was elevated (0.14 mM) with a normal lactate:pyruvate ratio. On day 6 of hospitalization, due to no improvement in controlling the seizures with conventional anticonvulsant therapy, she was placed in a pentobarbital coma, with the addition of Trileptal for complete control. Pentobarbital was discontinued on the 14th day of hospitalization, at which time she was observed to have occasional choreoathetoid movements and left leg weakness. MRI performed 10 days after the initial MRI study showed abnormal T2 and FLAIR hyperintensity involving bilateral caudate nuclei, globus pallidus, and putamen. MRI obtained 28 days after the initial study showed cystic changes in the globus pallidus bilaterally. The seizures were controlled and despite initial difficulties with eating and drinking, she received rehabilitation, improved, and was discharged to her home approximately 56 days after admission.

Three days prior to this presentation, the child was seen at another hospital for clenching of her body, urinary incontinence, and 5-10 minutes of unconsciousness. Lab studies and a head CT were reportedly unremarkable. She had a medical history of asthma requiring occasional albuterol and one past hospitalization for pneumonia. She lived with her mother and two healthy siblings. A brother died at 5 months of age with situs inversus and complex congenital heart defects. At age 7 months, a sister had died of sudden infant death syndrome as determined by autopsy examination.

Four days following discharge, the child was seen in the Emergency Department in cardiopulmonary arrest. Resuscitation efforts ceased after 25 minutes of asystole. A complete autopsy was performed.

Necropsy findings: No external abnormalities were identified. The peripheral organs appeared normal with the exception of petechiae involving the thymus and epicardial surface of the heart. The brain, weighing 1160 grams, and its coverings were normal externally. After formalin fixation, coronal sections revealed softening, cystic change, and dark tan discoloration involving both segments of globus pallidus bilaterally with apparent sparing of the medial medullary lamina. Softening and red-brown discoloration of bilateral putamen was also noted. Microscopic lesions in the left primary motor cortex and both substantia reticulata consisted of gliosis and macrophages with some neuronal loss. Mild to moderate neuropil vacuolation and scattered pyknotic neurons were present. Hippocampi were unremarkable. No abnormalities were revealed in the spinal cord. Sections from peripheral muscle and nerve, pituitary gland, and eye were histologically unremarkable. No mitochondrial ultrastructural abnormalities were detected in a sample from cerebellar cortex. Purkinje cells were well preserved and contained cytoplasmic crystalloid-like inclusions.

Toxicology studies were negative for alcohols, Trileptal, and other drugs tested. Enterovirus was cultured from stool.

Materials submitted: 2 x 2-inch projection slide, and an H&E section of the right or left pallidum.

Points for discussion: 1. Diagnosis; 2. Differential diagnosis