

CASE 2

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Clinical Summary: This 14 year old girl was born at 37 weeks' gestation to a 19 year old primigravida whose pregnancy was complicated by a severe upper respiratory infection, treated with antibiotics, in the fifth month of gestation. The patient's birth weight and head circumference were normal. At 2 months of age she was noticed to have staring episodes. At 4 months of age she had a generalized seizure. An EEG was abnormal, with polyspike bursts and frequent paroxysms of spike-wave activity. She was treated with phenobarbital and phenytoin. Although she showed some clinical improvement, her EEGs continued to be abnormal. She developed myoclonic seizures and was given a course of ACTH. Subsequently, she was treated with a variety of anticonvulsants, including nitrazepam, ethosuximide, methosuximide, clonazepam, primidone, diazepam, and chlorazepam.

At 20 months of age she had excision of a nasopharyngeal glioma.

At 5 years of age she stood and cruised holding onto furniture, finger fed, and crossed the midline to grasp objects. She did not follow commands, did not speak, and was not toilet-trained. She continued to have 2-3 clusters of myoclonic seizures and one grand mal seizure daily. At 6 years of age she could take 4 steps with help and understood 5 words. Over the following years she showed a decline in her ability to stand and became chair-ridden. At 11 years of age she was small and looked younger than she was. Her weight was 51 lbs and head circumference 48½ cm. She had mild hypoplasia of the right arm and leg, and increased leg muscle tone. Tendon reflexes were hyperactive, 3/4, and plantar responses were equivocal. She responded socially by looking at the examiner but did not speak nor follow commands. She required support to take steps. She had gaze-evoked nystagmus, followed visually but had disconjugate gaze. One year before death she was hospitalized on two occasions because of aspiration following grand mal seizures. She was found unresponsive in bed in the residential facility where she was placed five years before death.

Family History: Her maternal grandfather had posttraumatic seizures which were well controlled with phenytoin. A younger sister was completely normal and never had seizures.

An autopsy was performed.

The brain weighed 1050g. No encephalomalacia or cerebral atrophy was recognized. A small area of ectopic cortex was found adjacent to the posterior lateral ventricle. The cerebellum was sclerotic and atrophic. Microscopically, the material in the submitted slide was found abundantly throughout most of the cerebral cortex. occasional deposits were present in putamen and substantia innominata. None was seen in the caudate, amygdala, thalamus, cerebellum, brainstem or white matter.

Material Submitted: 1 stained and 1 unstained chrome gel mounted section.

Points for Discussion: What is the material?