49th ANNUAL DIAGNOSTIC SLIDE SESSION 2008

CASE 2008-5

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Clinical History: We report 4 cases of young children, including two females and two males, with ages ranging from 4 to 9 years, who presented with early onset seizure disorders refractory to medical treatment. They all displayed mild to moderate developmental delay involving mainly language, fine motor skills and below the average intelligence. Imaging of the brains showed different findings, with two cases displaying focal cortical dysplasia with periventricular nodular heterotopias, one case with small bilateral periventricular nodular heterotopias and one with a focal non-specific "thickening" of the cerebral cortex. Non-neurological anomalies were also recorded with one small septal cardiac defect, bowel malrotation at birth and bilateral abnormalities of upper extremities respectively in three of the patients. The neurological familial medical history was variable from case to case, ranging from unknown to a case with a sibling with Prader-Willi syndrome, one history of seizure in a maternal nephew and maternal migraine. All children underwent neurosurgical intervention with hemispherectomies.

Material submitted: H&E section cerebral cortex from one of the cases

Points for discussion:

1. Main abnormal neuropathological finding
2. Diagnosis

Diagnosis: Seizure with filamin-positive cytoplasmic astrocytic inclusions

References:

1. Horoupian, DS et al., Astrocytic cytoplasmic inclusions within an epileptic focus in an otherwise neurologically intact patient. Hum.Pathol. (2003), 34 (7): 714-6
2. Minagawa, M. et al., Inclusion bodies in cerebral cortical astrocytes: a new change of astrocytes. Acta Neuropathol (1992), 84 (1): 113-6.
3. Kato, S. et al., Immunohistochemical studies on the new type astrocytic inclusions identified in a patient with brain malformation. Acta neuropathol (1992), 84 (4): 449-52.

Learning objectives:

Discuss the histopathology of seizure and possible etiologies.