

54th ANNUAL DIAGNOSTIC SLIDE SESSION 2013

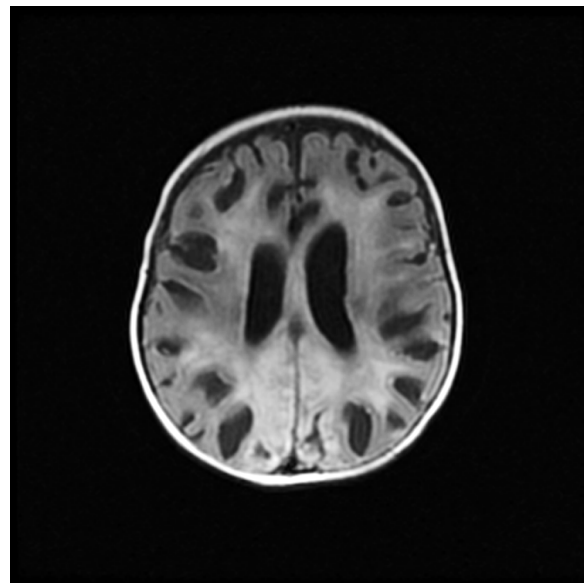
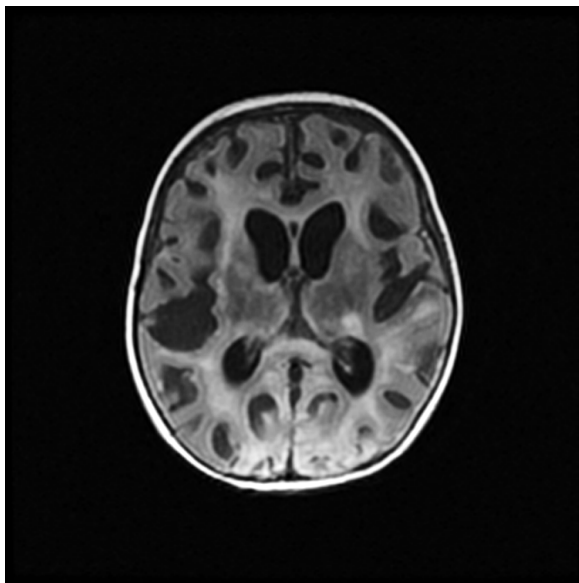
CASE 2013-9

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Clinical History: A 7-month-old infant, who had reached all developmental milestones, developed febrile seizures and watery diarrhea. After 3 days of illness, she was admitted to a community hospital for evaluation and treatment of seizures, diarrhea, and encephalopathy. EEG showed diffuse high-voltage slow-wave activity. Brain CT and MRI were unremarkable; work-up for infection was negative. The family history was significant for a sibling who died at the age of 5 after presenting with fever, hepatosplenomegaly, and cytopenia, but with no brain involvement. The infant was discharged home with presumed diagnosis of viral encephalitis after treatment with Keppra led to resolution of seizures. However, she continued to regress developmentally, leading to admission to a children's hospital, where she was evaluated by Neurology, Rheumatology, Infectious Disease, and Genetics. The patient's condition continued to worsen and she was transferred to a referral institution for specialized care. On admission, she showed decerebrate posturing, nystagmus, absent gag reflex, and tachypnea with subcostal retractions and stridor; repeat brain MRI is shown. Despite intensive support care, the infant continued to deteriorate and died 2 weeks later, at the age of 9 months.

Autopsy findings: At autopsy, there was hepatosplenomegaly and consolidation of the lungs; microscopic examination showed lymphocytic interstitial pneumonitis, lymphocytic infiltrates in the liver portal tracts and the kidney interstitium, and necrotizing lymphadenitis in a para-aortic lymph node. The brain (620 g; expected weight 810 ± 80 g) showed moderate diffuse edema, patchy subarachnoid hemorrhage, and severe parenchymal softening associated with white matter cavitation throughout the brain, but most prominently in the left frontal and occipital lobes.

Material Submitted: Brain MRI and one H&E-stained section from the left parietal lobe.



Points for discussion:

1. Diagnosis
2. Pathogenesis