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CASE 2007-02

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Clinical History. A previously healthy 3-year-old boy initially presented with left leg pain. His family history disclosed that he is the only son of a non-consanguineous couple. There is no family or personal history of hematologic, metabolic or neurologic diseases. Surgical history is pertinent for a left inguinal hernia repair four days prior to his initial presentation. He returned to the emergency room several times thereafter with bouts of abdominal pain and emesis. Later on he developed a fever 101.5 (rising to a maximum of 103°F), headaches and dizziness, followed by ataxia, and paleness. On admission, he had splenomegaly, fluctuating pancytopenia (Hb 7.0 g/L, Hct 20.1%, WBC $4.1x10^9$, platelets $119x10^9$/L) with low reticulocytes, absolute neutropenia ($0.5x10^9$/L), and slight predominance of lymphocytes. MRI of the brain showed extensive, infra and supratentorial, white matter disease which progressed during the hospital course. CSF analysis revealed increased protein (114 mg/dl) and WBC (64 $x10^6$) all monocytes. Bacterial, viral, fungal and mycoplasmal infection, metabolic defects, drug intoxication and rheumatic disease were ruled out. He then developed tonic clonic seizures which progressed to status epilepticus, and subsequently rapidly progressive neurologic deterioration with brain death, two and a half months after initial presentation.

Material Submitted: 1 H&E stained section of the cerebrum, brainstem or cerebellum.

Necropsy Findings. The autopsy examination showed hepatosplenomegaly. The brain weighed 1440 g. The brain was edematous and diffusely softened as well as focally liquefied, particularly in the posterior fossa (brainstem and partial cerebellum).

Points for Discussion.

1. Diagnosis
2. Pathogenesis