CFS (60.9% of 23). None had neurological lesions requiring surgery. (Millichap JJ et al. Methods of investigation and management of infections causing febrile seizures. **Pediatr Neurol** 2008;39:381-386). CFS without neurologic signs of intracranial pathology is insufficient indication for emergent CT scan. Diagnostic criteria for CFS and indications for CT scan may require re-evaluation.

METABOLIC DISORDERS

COGNITIVE OUTCOME OF INFANTS WITH POMPE DISEASE RECEIVING ENZYME-REPLACEMENT THERAPY

Researchers at University Medical Center, Rotterdam, the Netherlands, and University of Leuven, Belgium prospectively assessed cognitive function in 10 children with classic infantile Pompe disease who had been treated with enzyme-replacement therapy (ERT) since 1999. Median age at diagnosis was 0.7 months (range 0.1-6.2 months). ERT was started at a median age of 2.3 months (range 0.1-8.3 months). Developmental scores in the first 4 years of life ranged from above average to severe delay. The type of IQ test used, severity of motor problems, speech/language delay, and age at start of ERT influenced the developmental scores. At young age poor motor functioning may interfere with reliable assessment of cognition. Scores in 5 children tested after 5 years of age ranged between normal and mild developmental delay. Nine children had hearing deficits and 7 had impaired vision. Brain imaging in 6 patients revealed periventricular white matter abnormalities in 4. (Ebbink BJ, Aarsen FK, van Gelder CM, et al. Cognitive outcome of patients with classic infantile Pompe disease receiving enzyme therapy. **Neurology** 2012 May 8;78:1512-1518). (Response and reprints: Prof van der Ploeg. E-mail: a.vanderploeg@erasmusmc.nl).

COMMENT. Pompe disease (glycogen storage disease type II; acid maltase deficiency) is an autosomal recessive, progressive metabolic myopathy due to lysosomal a-glucosidase deficiency. Enzyme activity is reduced to <1%, and glycogen stores accumulate in skeletal, cardiac, and smooth muscle, and in the brain. Pompe presents with neonatal hypotonia, macroglossia, cardiomegaly, and hepatomegaly. Patients usually die before 1 year of age with cardiorespiratory failure or aspiration pneumonia. ERT, using recombinant human a-glucosidase, improves motor development and lengthens life expectancy, but ERT does not cross the blood-brain-barrier. Glycogen is stored in the CNS and may cause cognitive deficits. This study shows that children treated with ERT who survive to school age may have normal to mildly delayed cognitive development. Testing of young children < 4 years is largely dependent on motor function. Since muscle involvement and weakness are prominent features of Pompe disease and are resistant to ERT, cognitive development will be underestimated in children younger than 5 years.

Early treatment and newborn screening for Pompe disease are recommended. (Chien YH et al. **Pediatrics** 2009 Dec;124(6):e1116-1125) (Burton BK. **Am J Med Genet C Semin Med Genet** 2012 Feb 15;160(1):8-12).