Question 6 – September 9

A 4 year old presents to your local neurologist with cataplexy and developmental regression. He calls because you previously evaluated the child in infancy for cholestasis, which subsequently resolved. You review the liver biopsy which shows accumulation of fat, foamy macrophages, and bridging fibrosis.

You tell the neurologist you think the diagnosis is:

A. Alpha1antitrypsin deficiency
B. Ornithine transcarbamoylase deficiency
C. Niemann-Pick C
D. Citrin deficiency
E. Lead poisoning

Answer: C

Niemann Pick C is a lysosomal storage disease. It can present at various ages, but in infancy presents with neonatal cholestasis that later resolves, although cirrhosis often persists. Accumulation of lipids in the brain leads to neurologic symptoms and regressions years later. (Alpha 1 does not cause CNS disease. OTC is a urea cycle defect, not a storage disease. Citrin deficiency causes neonatal cholestasis, especially in Asian children. Lead poisoning would not present with neonatal liver disease)

http://www.ncbi.nlm.nih.gov/books/NBK1296/ (Gene Reviews)