COMMENT. A novel nonsense mutation in the TITF-1 gene is published simultaneously with the above case report in a Japanese family with benign hereditary chorea (Nakamura K et al. J Neurol Sci Feb 15, 2012;313(1-2):189-192). The proband showed severe generalized chorea, delayed motor development, subnormal intelligence, congenital hypothyroidism, bronchial asthma, and a history of pulmonary infection. These characteristics are features of Brain-Thyroid-Lung syndrome. Her brother and mother showed a mild benign hereditary chorea phenotype with congenital hypothyroidism. It is suggested that therapy with levodopa may compensate for underdeveloped dopaminergic pathways in this disorder.

Pathological findings in an autopsied Japanese adult with benign hereditary chorea 2 and hypotonia that presented at age 40 years showed mild degeneration of the striatum and cerebral white matter with astrocytosis. Non-progressive symptoms of chorea and hypotonia had persisted until the patient’s death at 83 years. (Yoshida Y et al. Neuropathology Jan 12, 2012 [Epub ahead of print]).

INFECTION DISORDERS

MANAGEMENT STRATEGY FOR CHILDHOOD ENCEPHALITIS

Researchers at the Children’s Hospital, University of Oxford, and Alder Hey Children’s NHS Foundation Trust, Liverpool, UK review the literature on encephalitis and suggest a management strategy. Encephalitis, defined as inflammation of brain parenchyma, is associated directly or indirectly with infectious agents (viruses or other microorganisms, fungi, parasites, rickettsiae) or caused by other inflammatory or immune-mediated pathologies (eg. ADEM, paraneoplastic, NMDAR encephalitis, voltage gated K channel limbic encephalitis). Herpes simplex virus (HSV) type 1 is the most common cause of sporadic encephalitis, either primary infection or via reactivation of virus in the trigeminal ganglion. Enteroviruses such as polio and arboviruses (Japanese encephalitis virus and West Nile virus) enter the brain across the blood-brain barrier. Etiology is undefined in 60% cases of encephalitis.

CSF should be sent: 1) to microbiology lab for microscopy, culture and sensitivity analysis; 2) to virology lab for PCR for HSV types 1 and 2, VZV, HHV-6 and -7, CMV, EBV, enteroviruses, respiratory viruses, HIV and C pneumonia; 3) to biochemistry for glucose (with paired plasma sample), lactate and oligoclonal bands; and 4) stored sample for future tests. Up to 10% of patients with viral encephalitis have a normal CSF. Some patients have a mononuclear pleocytosis and moderately elevated protein in the CSF, or raised red blood cell count (hemorrhagic encephalitis). Eosinophils suggest infection with helminthes, toxoplasma, Rickettsiae, or M pneumonia. Low CSF glucose suggests a bacterial, fungal or protozoal etiology. PCR may be negative early and after acyclovir. (Thompson C, Kneen R, Riordan A, Kelly D, Pollard AJ. Encephalitis in children. Arch Dis Child Feb 2012;97:150-161). (Respond: Dr Clara Thompson, C/o Professor AJ Pollard, Children’s Hospital, Oxford, UK. E-mail: clara.thompson@doctors.org.uk).

COMMENT. This excellent review also refers to the value and indications for EEG and MRI in diagnosis of encephalitis, treatment including acyclovir, and prognosis.