spasms in 59, neck stiffness and spasms in 10, upper extremity stiffness and spasms in 4, abdominal wall stiffness and spasms in 26, respiratory symptoms with spasms in 6, and falls in 30. Symptoms were exacerbated by emotional stress, startle, cold, and movement. Seventy-nine were GAD65 antibody seropositive, and 53 (67%) had at least one coexisting autoimmune disease; 3 (4%) had cancer. GAD65 antibody values were significantly higher in patients with classic SMA than in those with partial or variant SMA. Treatment with diazepam (40 mg/day) provided sustained improvements. Immunotherapy gave additional improvements. Sixteen (64%) of 23 patients with extended follow-up remained ambulatory. (McKeon A, Robinson MT, McEvoy KM et al. Stiff-man syndrome and variants. Clinical course, treatments, and outcomes. **Arch Neurol** Feb 2012;69(2):230-238). (Respond: Andrew McKeon MD, Department of Neurology, Mayo Clinic. E-mail: mckeon.andrew@mayo.edu).

COMMENT. Stiff-man syndrome occurs mainly in adults but can occur in children. Diagnosis may be confirmed with EMG documentation of hyperexcitability of spinal motor neurons, GAD65 antibodies, and response to diazepam, first described by Howard FM Jr (**Proc Staff Meet Mayo Clin** 1963;38:203-212).

Several reports of stiff-child syndrome are uncovered by a PubMed search. The disorder must be distinguished from hyperexplexia or hereditary stiff-baby syndrome, an autosomal dominant disorder. The EMG shows persistent hyperexcitability at rest, abolished by diazepam. The hypertonia lessens during sleep and increases with the slightest startle or tactile stimulus. Nose tapping will elicit the hyperexplexic startle response in affected newborns. (Tohier C et al. **Arch Dis Child** 1991;66:460-461) (**Ped Neur Briefs** May 1991).

# MOVEMENT DISORDERS

## BENIGN HEREDITARY CHOREA: RESPONSE TO LEVODOPA

A case of sporadic non-progressive chorea is reported in a 6 year-old girl from Hospital Sant Joan de Deu, Barcelona University and other centers in Spain and The Netherlands. At age 21 months she was diagnosed with severe motor delay and gait disorder. Birth and perinatal history including screening test for hypothyroidism were normal. A diagnosis of subclinical hypothyroidism was made at 2 years of age and she was treated with oral L-thyroxine. Language and learning skills have been age appropriate. At 3 years of age she was hypotonic, reflexes were normal, but her gait was unstable, clumsy, and wide-based, with frequent, sudden falls. Choreiform movements were generalized, affecting the mouth, limbs, and trunk, and were not progressive. A *TITF-1* de novo gene mutation test was positive. Levodopa therapy started at age 3 years 6 months controlled the chorea. When therapy was temporarily interrupted after 1 year, symptoms recurred with frequent falls and clumsy gait. Discontinuation of therapy slowly after 3 years of treatment was successful without relapse. (Fons C, Rizzu P, Garcia-Cazorla A, et al. TITF-1 gene mutation in a case of sporadic non-progressive chorea. Response to levodopa treatment. **Brain Dev** 2012;34:255-257). (Respond. Dr Carmen Fons, Department of Pediatric Neurology, Hospital Sant Joan de Deu, Barcelona, Spain. E-mail: cfons@hsjdbcn.org).

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]).

# INFECTIOUS 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.