
**DOPA-RESPONSIVE MOTOR DISORDER WITH SEPIAPTERIN REDUCTASE DEFICIENCY**

The clinical findings in 7 children from Malta, at first suspected to have cerebral palsy and later diagnosed with sepiapterin reductase deficiency, are reported from Great Ormond Street Hospital, London, UK, and St Luke’s Hospital, University of Malta. All had early motor delay with diurnal variation and cognitive impairment. Oculogyric crises occurred from an early age in 6, hypotonia followed by dystonia in 5, chorea in 4, bulbar involvement in 3, and Parkinsonian tremor in 2. Treatment with L-dopa was started at ages 1 to 10 years in doses of 1.5 to 4 mg/kg/day, and the response was dramatic, except for aggravation of chorea. Improvement was obtained predominantly in motor function and control of oculogyric crises, but cognitive function and learning remained moderately impaired. A worsening of symptoms in hot weather was relieved by an increase in dose of L-dopa. All had a novel mutation in the tetrahydrobiopterin pathway involving sepiapterin reductase, with autosomal recessive inheritance. None had an abnormality in the gene encoding guanosine triphosphate cyclohydrolase 1 (GTPCH1), consistent with the autosomal dominant Segawa disease. (Neville BGR, Parascandalo R, Farrugia R, Felice A. Sepiapterin reductase deficiency: a congenital dopa-responsive motor and cognitive disorder. Brain October 2005;128:2291-2296). (Professor BGR Neville, The Wolfson Centre, Mecklenburgh Square, London WC1N 2AP, UK).

COMMENT. This autosomal recessive, dopa-responsive congenital motor disorder, characterized by dystonia with diurnal variation and dramatic response to L-dopa, resembles in some respects the autosomal dominant GTPCH1, dopa responsive deficiency known as Segawa’s disease. Unusual features include a congenital onset, early hypotonia and frequent bulbar involvement, atypical manifestations of Segawa’s disease (Ann Neurol 2003;54(Supp 6);S32-45). Both conditions are often incorrectly diagnosed with unexplained cerebral palsy. The authors recommend that infants with unexplained cerebral palsy who have ITP antibodies be screened for sepiapterin reductase deficiency, although outside Malta, the disease is probably very rare. The report adds to a range of cases of dopa-responsive motor disorders and the necessity for a trial of L-dopa when the diagnosis is unclear.

**INFECTIOUS DISORDERS**

**AUTOANTIBODIES IN POST-EPSTEIN-BARR ACUTE CEREBELLAR ATAXIA**

Eight of 23 patients with acute cerebellar ataxia (ACA) following Epstein-Barr virus (EBV) infection (proven serologically) had increased IgM anti-triosephosphate isomerase (TPI) antibody titers, in a study at Kyorin University, Tokyo, and Kinki University, Osaka,