PROGRESSIVE ENCEPHALOPATHY IN BOYS WITH SYMPTOMS OF RETT SYNDROME AND *MECP2* MUTATIONS

Four young boys with neonatal onset of encephalopathy, a progressive course, and *MECP2* mutations are reported from the University of Alabama, Birmingham, AL Symptoms suggestive of Rett syndrome included failure to thrive, respiratory insufficiency, microcephaly, hypotonia, movement disorder, with myoclonic, dyskinetic, and choreiform patterns, and repetitive face scratching or nose rubbing stereotypies. *MECP2* mutations, characteristic of girls with Rett syndrome, are previously reported in 11 boys with progressive encephalopathy, 7 having affected sisters. The authors report 4 de novo male cases, 3 of whom died at 14 to 27 months of age. Details of the 11 previously published male patients are tabulated, 10 of whom died in infancy. (Kankirawatana P, Leonard H, Ellaway C et al. Early progressive encephalopathy in boys and *MECP2* mutations. **Neurology** July (1 of 2) 2006;67:164-166). (Reprints: Dr Alan K Percy, CIRC 320E, 1530 3rd Avenue South, Birmingham, AL 35294).

COMMENT. Young boys presenting with early progressive encephalopathy, respiratory insufficiency, movement disorder, seizures, or changes in muscle tone should be tested for *MECP2* pathology and a possible diagnosis of Rett syndrome. Molecular confirmation is important in defining the cause and prognosis of this progressive encephalopathy and in providing genetic counseling.

NEUROMUSCULAR DISEASES

INFANTILE ONSET MYASTHENIA GRAVIS WITH MuSK ANTIBODIES

A late infantile case of myasthenia gravis (MG) with anti-muscle-specific receptor tyrosine kinase (MuSK) high antibody (Ab) titer is reported from Kyushu University, Fukuoka; and Nagasaki University, Japan. The onset with bilateral ptosis and abnormal phonation occurred after febrile seizures at 2 years of age. The diagnosis by a positive edrophonium test at age 3 years was followed by treatment with oral prednisolone. Ocular symptoms responded but bulbar symptoms persisted and worsened by age 12 years. The addition of pyridostigmine and increase in dose of steroid had limited benefit. Thymectomy was performed at age 26 years, with no response. On admission at age 29, he had marked facial weakness, nasal voice and dysphagia, and choked on drinking water. The tongue was atrophic with a triple-furrowed appearance, and the neck and proximal muscles of extremities were weak, without fasciculations. Tendon reflexes and sensation were normal. Edrophonium infusion improved speech and swallowing. Anti-MuSK Ab measured by radioimmunoassay was 4.80 nmol/L (normal, <0.05). His voice, swallowing and muscle strength improved after plasma exchange using Spectra. (Murai H, Noda T, Himeno E et al. Infantile onset myasthenia gravis with MuSK antibodies. **Neurology** July (1 of 2) 2006;67:174-4).