

bilateral ptosis, weakness of eye closure, facial muscles, shoulder, hand and hips. In Pedigree 2, the index patient developed ptosis at 17 years, and weakness progressed to other muscle groups over the next 7 years. Pedigree 3 index patient noted transient weakness of finger extensors in her twenties, which later progressed to mild involvement of wrist, neck and other muscles. Antibodies to AChR were absent in all three patients and there was no response to anticholinesterase treatment. EMG showed decrement of the CMAP and a repetitive response to a single nerve stimulus. Only Pedigree 3 showed a typical dominant inheritance pattern. (Croxon R, Hatton C, Shelley C et al. Recessive inheritance and variable penetrance of slow-channel congenital myasthenic syndromes. Neurology July (2 of 2) 2002;59:162-168). (Reprints: Dr David Beeson, Neurosciences Group, Weatherall Institute of Molecular Medicine, The John Radcliffe, Headington, Oxford OX3 9DS, UK).

COMMENT. Slow-channel congenital myasthenic syndromes are typically of dominant inheritance and caused by missense mutations in the muscle nicotinic acetylcholine receptor (AChR). Symptoms are present at birth or may be delayed until adulthood. Fatigable muscle weakness, selectively involving neck, shoulder and finger extensors, is mild or severe and tends to be slowly progressive. Response to anticholinesterase treatment is absent, and EMG shows a double response to a single nerve stimulus. Adding to the 11 previously described mutations underlying the SCCMS, the Oxford team describes two new mutations in the  $\epsilon$  subunit, with symptoms present only in the index patient, and the first reported examples of recessively inherited SCCMS.

## ANOXIC DISORDERS

### **OUTCOME FACTORS IN HYPOXIC ISCHEMIC ENCEPHALOPATHY**

The predictive value of history, examination, Glasgow Coma Scale (GCS) scores, EEG and sensory evoked potentials (SEP) in the prognosis of children with acute hypoxic-ischemic encephalopathy (HIE) was evaluated at the University Hospital of Lille, France. Of 53 consecutive children who were mechanically ventilated for HIE, 12 had died at 24 hours after admission, 3 were awake, and 42 showed impaired consciousness or were in coma (GCS <8).

In the 42 with uncertain prognosis, outcome was good in 12 and mild or moderate disability in 4 patients (a favorable outcome in 38%), and severe disability in 7 patients; 19 ultimately died. Predictors of an unfavorable outcome included: 1) an initial cardiopulmonary resuscitation duration longer than 10 minutes; 2) a GCS <5 at 24 hrs after admission; 3) EEG showing a discontinuous pattern and spikes or epileptiform pattern; and 4) bilateral absence of the N20 wave on SEPs. (Mandel R, Martinot A, Delepoulle F et al. Prediction of outcome after hypoxic-ischemic encephalopathy: a prospective clinical and electrophysiologic study. J Pediatr July 2002;141:45-50). (Reprints: Pr F Leclerc MD, Pediatric Intensive Care Unit, Hôpital Jeanne de Flandre, CHRU, 2 place O, Lambret, 59037 Lill9 Cedex, France).

COMMENT. At 24 hours after birth, clinical signs, the GCS, EEG and SEPs permit early prediction of prognosis of children with HIE.

Microcephaly after HIE may be predicted by serial head circumference measurements between birth and 4 months of age. A decrease in HC ratios of >3.1% by 4 months correlates with development of microcephaly and neurologic sequelae before 18 months. (see Progress in Pediatric Neurology III, 1997;p396).