CHRNE MUTATION AND CONGENITAL MYASTHENIA

The CHRNE e1293insG mutation was identified in 14 (60%) of 23 North African families with an early onset form of congenital myasthenic syndrome studied at centers in France, Tunisia, Algeria, and UK. The phenotypic expression was homogeneous with moderate hypotonia and oculobulbar involvement, mild and stable course, and good response to cholinesterase inhibitors. This mutation is thought to originate in an ancient single founder event in the North African population. (Richard P, Gaudon K, Haddad H, et al. The CHRNE 1293insG founder mutation is a frequent cause of congenital myasthenia in North Africa. **Neurology** Dec 1 2008;71:1967-1972). (Reprints: Dr Daniel Hantai, Inserm U582.47, Boulevard de l'Hopital, Paris, France 75651.
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COMMENT. Congenital myasthenic syndrome (CMS) is caused by various genetic defects that include recessive mutations in AChR subunit CHRNE gene. In North Africa, most patients are homozygous for the e1293insG mutation. In Europe, the mutation is associated with a heteroallelic mutation in CHRNE. The DOK7 mutation CMS has a broad clinical phenotype, distinguished by the absence of external ophthalmoplegia, a progressive deterioration of respiratory function, and lack of long-term response to esterase inhibitors (Muller JS et al. **Brain** 2007;30:1497-1506; **Ped Neur Briefs** June 2007;21:43-4). Molecular genetic testing is important in diagnosis and therapy of infants with CMS. In patients with COLQ mutations, esterase inhibitors may cause worsening of symptoms (**Ped Neur Briefs** March 2008;22:17-18). In an early report of 6 infants with a 'neonatal persistent (congenital) myasthenia gravis' treated at the Massachusetts General Hospital, Boston (Millichap & Dodge. **Neurology** 1960;10:1007-1014), mothers did not suffer from MG, but 2 cousins of 1 patient and a sister of another were affected. The principal presenting signs were ptosis, weak cry, and generalized weakness. In later childhood, ptosis and external ophthalmoplegia were most prominent, and involvement of bulbar musculature, weakness, and respiratory difficulty were uncommon and mild. Symptoms, especially ophthalmoplegia, were only partially relieved or refractory to pyridostigmine. In the MGH series of pediatric myasthenic patients, 10 infants had the neonatal transient type, 6 a congenital type, and 35 the juvenile type.

LANGUAGE DISORDERS

AUTOSOMAL DOMINANT PARTIAL EPILEPSY WITH AUDITORY FEATURES

Auditory and language processing in 17 subjects with autosomal dominant partial epilepsy with auditory features (ADPEAF) was investigated by MRI, fMRI, and MEG and compared to 26 controls, in a study at Columbia University, New York. Age of seizure onset was late childhood or adolescence. MRI revealed no structural abnormality, but fMRI and