

SEIZURE DISORDERS

CORTICAL EXCITABILITY MEASURES IN PATIENTS AND UNAFFECTED SIBLINGS

Researchers at St Vincent's Hospital, Victoria, Australia, measured cortical excitability using transcranial magnetic stimulation in 157 patients with epilepsy (95 generalized and 62 focal) and their asymptomatic siblings and results were compared to those of 12 controls and 20 of their siblings. No differences were observed in cortical excitability between healthy controls and their siblings. Compared to controls, cortical excitability was higher in siblings of patients with generalized or focal seizures. Motor threshold was lower in patients with juvenile myoclonic epilepsy compared with their siblings. The disturbance in cortical excitability appears to involve intracortical inhibitory circuits even in siblings of patients with a structural abnormality and acquired epilepsy. (Badawy RAB, Vogrin SJ, Lai A, Cook MJ. Capturing the epileptic trait: cortical excitability measures in patients and their unaffected siblings. *Brain* 2013 Apr;136(Pt 4):1177-91). (Response: Dr Radwa Badawy. E-mail: badawyr@unimelb.edu.au).

COMMENT. The authors conclude that certain genetic factors that predispose to epilepsy and a complex genetic/environmental interaction determine the clinical phenotype.

Gene mutations in progressive myoclonus epilepsies.

A mutation in the potassium channel associated gene CNTN2 was the cause of a cortical myoclonic tremor and epilepsy in a consanguineous Egyptian family (Stogmann E, et al. *Brain* 2013 Apr;136(Pt 4):1155-60)

A mutation in the GOSR2 gene was identified in 12 patients with "North Sea" progressive myoclonus epilepsy. Early onset ataxia at 2 years of age was followed by myoclonic seizures at average age 6.5 years, followed by multiple seizure types. All patients developed scoliosis by adolescence, an important diagnostic clue, some had pes cavus or syndactyly, and all had elevated serum creatine kinase (mean 734 IU) and normal muscle biopsies. EEG showed generalized S/W with posterior predominance and photosensitivity. With progressive decline, patients became wheelchair bound by mean age 13 years. The cases all came from countries bounding the North Sea. The relentless course distinguished "North Sea" progressive myoclonus epilepsy (PME) from other PMEs (Lomax LB, et al. *Brain* 2013 Apr;136(Pt 4):1146-54). Other PMEs include Unverricht-Lundborg disease (gene CSTB mutation), Lafora's disease (EPM2A), Northern with mental retardation (CLN8), and teenage-onset PME (CLNB) (Andrade DM, et al. *Pediatr Neurol* 2012 Sep;47(3):205-8).

TREATMENT OF SYMPTOMATIC INFANTILE SPASMS

Investigators at Tokyo Women's Medical University studied the clinical, radiological, and EEG characteristics of 69 patients with infantile spasms (IS) followed for 3-74 months (mean 18 months) after initial cessation of epileptic spasms (ES).