PEDIATRIC NEUROLOGY BRIEFS

A MONTHLY JOURNAL REVIEW

J. GORDON MILLICHAP, M.D., F.R.C.P., EDITOR

Vol. 4, No. 8 August 1990

SEIZURE DISORDERS

MYOCLONUS EPILEPSIES

Progressive myoclonus epilepsies and related disorders were classified at an international workshop in Marseille, France. Myoclonus refers to sudden, brief, shock-like involuntary movements. The progressive myoclonus epilepsies (PME) are a group of rare genetic disorders characterized by myoclonus, epileptic seizures and progressive neurological deterioration, particularly dementia and ataxia. Many specific diseases can cause the PME syndrome. Electrophysiological findings share many similarities including generalized spike wave discharges, photosensitivity, focal epileptiform discharges, vertex spikes in rapid eye movement sleep, and giant somatosensory evoked potentials. Slow background activity occurs particularly in PME caused by diffuse neuronal damage or storage diseases. Five disease entities account for most of the cases: 1) Unverricht-Lundborg ("Baltic myoclonus"), 2) LaFora disease, 3) neuronal ceroid-lipofuscinoses, 4) mitochondrial disorders, and 5) the sialidoses. The term Ramsay Hunt syndrome was discarded in favor of two main syndromic categories: 1) Progressive myoclonus epilepsies and 2) progressive myoclonic ataxias (PMA). PMA comprises myoclonus, and progressive cerebellar ataxia with infrequent or absent epileptic seizures and little or no dementia. The causes of PME syndrome are better defined than those of PMA. PMA may be caused by spinal cerebellar degenerations, mitochondrial disease, early cases of Unverricht-Lundborg disease, sialidosis and in association with celiac disease. Progressive myoclonus is distinguished from static myoclonus encephalopathies such as post anoxic myoclonus (Lance-Adams syndrome).

PEDIATRIC NEUROLOGY BRIEFS (ISSN 1043-3155) ©1990 covers selected articles from the world literature and is published monthly. Subscription requests ($28 US or £15 UK annually; add $5 (£3) for airmail outside North America) may be sent to: Pediatric Neurology Briefs - J. Gordon Millichap, M.D., F.R.C.P. - Editor, P.O. Box 11931, Chicago, IL 60611, USA, or Nat Wst Bnk, 94 Kensington High Street, London W8, UK. The Editor is Professor of Neurology and Pediatrics at Northwestern University Medical School, Chicago, and is presently at Southern Illinois University School of Medicine, Springfield, Illinois, USA.

Phenytoin may exacerbate myoclonus whereas treatment with benzodiazepines and valproate is most effective. (Andermann E et al. Classification of progressive myoclonus epilepsies and related disorders. Marseille Consensus Group, Ann Neurol July 1990; 28:113-116).

COMMENT Myoclonus and its relation to monoamine, GABA and other receptors is reviewed from the Departments of Neurology and Pediatrics,University of Southern California School of Medicine, Los Angeles, CA. (Snodgrass SR. FASEBJ July 1990; 4:2775-2788). The term myoclonus was first used in 1881 by Freidreich with reference to a progressive movement disorder of a gradual onset in a middle-aged man. By 1891 Unverricht had written a book on myoclonus, describing a family with myoclonus and progressive deterioration. Snodgrass, in the present article, classifies myoclonus in four categories: a) stimulus-sensitive myoclonus, b) stimulus-insensitive myoclonus, c) sleep myoclonus, and d) asterexis and negative myoclonus. Distinction between epileptic and nonepileptic myoclonus is often difficult. Posthypoxic action myoclonus (Lance-Adams syndrome) is associated with reduced CSF levels of 5-HIAA, the serotonin metabolite, and 50% respond to treatment with 5-HTP. Benzodiazepines and valproic acid are the most useful drugs for patients unresponsive to HTP. Various animal models of myoclonus are described. (Gundlach AL. FASEB J July 1990; 4:2761-2766).

"Severe myoclonic epilepsy of infancy" is discussed by Hurst DL (Epilepsia July/August 1990; 31:397-400) from the Department of Medical and Surgical Neurology, Texas Tech University, Lubbock, Texas. The central features of this syndrome include: 1) normal development before the onset of seizure activity, 2) repeated prolonged febrile seizures, 3) later onset of mixed/myoclonus epilepsy, 4) developmental slowing with onset of seizure activity, and 5) evolving EEG abnormalities. The incidence of the syndrome is estimated at one in 40,000 children. Dravet, who first described the syndrome in 1978, found a 30% prevalence in a group of 142 children with myoclonic epilepsy. A more vigorous, rather than conservative, approach to the management of complex febrile seizures might be suggested by this report.

RASMUSSEN ENCEPHALITIS

Immunological abnormalities associated with chronic encephalitis, epilepsy, and progressive hemiplegia in a three-year, nine-month old girl are reported from the University of Utah School of Medicine and the Primary Children's Medical Center, Salt Lake City, Utah. The child developed simple partial seizures refractory to medications and associated with a contralateral hemicerebral atrophy. Immunologic abnormalities were indicated by elevated antinuclear antibody, CSF oligoclonal bands and elevated immunoglobulin G (IgG). A right subtotal hemispherectomy resulted in control of the seizures. Pathological study showed widespread cerebral vasculitis and severe