FAMILIES' VIEWS ON DISCONTINUING ANTICONVULSANTS

The opinions of families of 76 children with epilepsy (>3 months seizure-free) and their 4 physician epilepsy specialists regarding acceptable risks of seizure recurrence (RSR) after AED withdrawal were investigated by questionnaire at the IWK Children's Hospital, Halifax, Nova Scotia, Canada. Families' responses were very variable: a RSR of 25% was unacceptable to 42% of families, whereas a >75% risk was considered acceptable by 20%. Families responses were dependent on previous seizure frequency, multiple seizure types, school grades repeated, and the *habits of playing the lottery*. The degree of risk acceptable to a particular family was not predicted by their physicians. Physician's opinions of acceptable RSR were more consistent, a median of 40%, but they varied from 0 to 90% for individual children. Families gave the primary responsibility for discontinuing AEDs to the physician in 80% of cases, but 46% reserved a secondary role in the decision for the parents. The majority of families (89%) denied the physician an exclusive role in the decision making. (Gordon K et al. Families are content to discontinue antiepileptic drugs at different risks than their physicians. Epilepsia June 1996;37:557-562). (Reprints: Dr K Gordon, IWK Children's Hospital, 5850 University Ave, Box 3070, Halifax, Nova Scotia, Canada, B3J 3G9).

COMMENT. This center's practice of discontinuing AEDs after a seizure-free 2 year period, with an expected seizure recurrence of 30-40%, results in an unacceptable degree of risk for more than half the families in this study. Withdrawal of AEDs after a 1 year seizure-free period provided a similar high risk of recurrence of 30-40%, in a recent study from the same center (Dooley J, Gordon K et al. Neurology April 1996;46:969-974).

Physicians should be aware of the parents' opinions and attitudes in regard to risk of seizure recurrence, when advising on the time to discontinue treatment of a child with epilepsy, but they cannot allow a "parent's strategy of playing lotteries" to intervene in this important decision. Each child with epilepsy is an individual, and the time for anticonvulsant withdrawal should be determined on an individual basis, having regard to varying predictive factors, not based on a generalized fixed period of 1 or 2 years. An expected seizure recurrence of 30-40% should be unacceptable in current practice, and the consequences of a single seizure relapse, especially in adolescents and young adults, should demand a more conservative and individual approach to this important decision. See Progress in Pediatric Neurology I, PNB Publishers, 1991, pp 100-104, for further references and commentaries on anticonvulsant withdrawal practices.

ATAXIA DISORDERS

NON-PROGRESSIVE ATAXIAS

A population-based study of 78 Swedish children with non-progressive ataxia is reported from the Department of Paediatrics, Malarsjukhuset, Eskilstuna; Department of Neuroradiology, Karolinska Institute, Stockholm; and Department of Paediatrics, University of Goteborg, Sweden. Criteria for inclusion were an ataxic gait, dyssynergia, dysmetria and intention tremor, resulting from prenatal (45%) or perinatal events (4%), and excluding patients with spasticity (51% were unclassifiable). The prevalence was 0.13 per thousand of 6- to 22-year-old children and adolescents. CT or MRI, available in

70 patients (90%), showed infratentorial pathology in 27%, including focal maldevelopments of the cerebellum: Dandy-Walker (2), Joubert syndrome (1), encephalocele (1), and hypoplasia (1). A genetic anomaly was found in 18 patients, including Angelman syndrome in 3. Forty-seven patients (60%) had mental retardation, severe in 25 and mild in 22; sixty nine (88%) had delayed speech development; 58% had visual dysfunction; and only 17 (22%) had normal cognitive function. (Esscher E et al. Non-progressive ataxia: origins, brain pathology and impairments in 78 Swedish children. Dev Med Child Neurol April 1996;38:285-296). (Respond: Dr Eva Esscher, Department of Paediatrics, Malarsjukhuset, Eskilstuna, S-633 88, Sweden).

COMMENT. Simple non-progressive ataxias comprise less than 10% of cases of cerebral palsy. Of 50% with known pathologies, the majority were prenatal in origin, many with a genetic background. No abnormality was revealed by imaging studies in 61%.

MACHADO-JOSEPH DISEASE

The frequency, and clinical, molecular, and neuropathological features of spinocerebellar ataxia 3 (SCA3) and Machado-Joseph disease (MJD) in 125 autosomal dominant cerebellar ataxia (ADCA) families were analyzed at the Service de Neuropathologie, Hopital de la Salpetriere, Paris, and Service de Neurologie, Hopital de Haut Leveque, Pessac, France; and Service de Neurologie, Hopital des Specialites, Rabat, Morocco. Thirty four families (126 patients) carried the expanded CAG repeat in the MJD1 gene. The length of the CAG repeat influenced the age at onset and the frequency of clinical signs associated with cerebellar ataxia (abnormal DTRs, decreased vibration sense). The frequency of supranuclear ophthalmoplegia, swallowing difficulties, and amyotrophy was significantly correlated with the disease duration. The age at onset varied from 14 to 70 years, mean 36 yrs, for the SCA3/MJD. One patient with SCA2 had an onset of ataxia at 8 years, whereas the youngest with SCA1 was 21. Neuropathological lesions distinguished the varieties of SCA, eg. basal ganglia lesions were more severe in SCA3/MJD than in SCA1. (Durr A et al. Spinocerebellar ataxia 3 and Machado-Joseph disease: Clinical, molecular, and neuropathological features. Ann Neurol April 1996;39:490-499). (Respond: Dr Durr, INSERM U289, Hopital de la Salpetriere, 4 boulevard de l'Hopital, 75651 Paris Cedex 13, France).

COMMENT. Despite the infrequent occurrence of Machado-Joseph disease in children, three recent reports from different parts of the world, France, Japan, and Australia, prompted commentary. MJD is an autosomal dominant spinocerebellar degeneration, occurring mainly in people of Portuguese descent. An unstable trinucleotide CAG repeat in MJD maps to the same region of chromosome 14 as the SCA3 locus. Two of 3 patients from Japan noted gait unsteadiness at age 18 years, followed 2 years later by involuntary movements, dysarthria, dysphagia, and hand incoordination. (Sakai T et al. A family with Machado-Joseph disease, previously diagnosed as dentatorubral-pallidoluysian atrophy. Neurology April 1996;46:1154-1156). Four families of Australian aboriginal people with MJD exhibited anticipation and an earlier age of onset. (Burt T et al. Machado-Joseph disease in east Arnhem Land, Australia: Chromosome 14q32.1 expanded repeat confirmed in four families. Neurology April 1996;46:1118-1122). MJD should be included in the differential diagnosis of a progressive ataxia with onset in later childhood, adolescence, or adulthood.