NEUROMUSCULAR DISORDERS

GUILLAIN-BARRE SYNDROME OUTCOME

The prognostic indicators and outcome in 297 patients with Guillain-Barre syndrome were analysed at the Istituto di Ricerche Farmacologiche 'Mario Negri', Milan, Italy. Patients, recruited from Italian centers, were followed for 24 months or until recovered. Only 5% were children <15 years; approximately 50% were older than 50 years. The commonest antecedent illnesses were influenza (26%), URI (15%), and gastroenteritis (9%). Immunizations were causative in 5 patients: influenza vaccine in 4 and DPT in 1. The mean times to nadir, improvement and recovery were 12, 28 and 200 days; 71% recovered, 16% had residua, and 11% died. Cardiac arrest with dysautonomic syndrome was the main cause of death. The percentage with respiratory failure increased with age (7% <35 years, 31% >35 years). Predictors of a more favorable outcome included young age at onset, influenza antecedent illness, mild symptoms and signs, short latency to nadir, short duration of active disease, and normal or demyelinating electrodiagnostic features. Prognosis was less favorable in older patients, with gastroenteritis antecedent illness, respiratory insufficiency, long latency to nadir, long duration of active disease, and axonal damage. Treatments were effective only during the active phase and did not influence chance of recovery. (Beghi E, Bono A, Bogliun G, et al. The prognosis and main prognostic indicators of Guillain-Barre syndrome. A multicentre prospective study of 297 patients. Brain Dec 1996;119:2053-2061). (Respond: Dr Ettore Beghi, Instituto di Ricerche Farmacologiche 'Mario Negri', Via Eritrea, 62-20157-Milano, Italy).

COMMENT. Guillain-Barre syndrome can no longer be regarded as an acute or subacute demyelinating inflammatory polyradiculopathy with a favorable outcome and complete recovery in a majority of patients. Recent reports of cases with a less favorable prognosis have been asociated with antecedent *Campylobacter* enteritis, axonal damage and a positive response to anti-GM1 antibodies. This multicenter, prospective study supports the concept of a heterogeneous disorder with variable prognosis, dependent on age, antecedent illness, axonal damage, and duration of active disease. Perhaps the antecedent illness and the causative role of vaccines, especially influenza, require more attention.

Immune globulins are effective and safe in severe childhood-onset Guillain-Barre syndrome, according to a multicentre prospective study of 26 children reported from the Rambam Medical Center, Haifa, Israel. (Shahar E, Shorer Z, Roifman CM, et al. Pediatr Neurol Jan 1997;16:32-36).

HEREDITARY MOTOR AND SENSORY NEUROPATHY IIB

The clinical and electrodiagnostic features of 10 adults in a family with axonal, autosomal dominant, hereditary motor and sensory neuropathy (HMSN IIB), with linkage to chromosome 3q, are reported from Washington University School of Medicine, St Louis, MO. Symptomatic age at onset is in the second or early third decade; 7 of 10 subjects noted foot drop or weakness while in high school. Some complained of diminished sensation in the feet and toe/foot ulcerations. Intrinsic hand muscles were mildly affected in younger, and wrist and finger extensors in older patients. All had pes cavus or hammer toes. Ankle reflexes were reduced. Electrodiagnostic studies showed a distal sensorimotor axonopathy with normal motor conduction velocities. The

disorder was thought to be a distinct subtype of HMSN II. (Elliott JL, Kwon JM, Goodfellow PJ, Yee W-C. Hereditary motor and sensory neuropathy IIB: clinical and electrodiagnostic characteristics. Neurology Jan 1997;48:23-28). (Reprints: Dr Jeffrey L Elliott, Department of Neurology, Washington University School of Medicine, 660 S Euclid Ave, St Louis, MO 63110).

COMMENT. Autosomal dominant hereditary motor and sensory neuropathies (HMSNs) are either predominantly demyelinating in type (HMSN I), with slowed conduction velocities and hypertrophic nerves, or axonal (HMSN II), with normal conduction velocities, small compound muscle action potentials, and axonal degeneration. The above report of HMSN IIB appears to be a homogeneous entity, exhibiting linkage to chromosome 3q, younger age at onset, and characteristic clinical and electrodiagnostic features.

A novel point mutation in the peripheral myelin protein 22 (PMP22) gene is reported in association with HMSN I (Charcot-Marie-Tooth type 1A) from the University of Cagliari, Italy. (Marrosu MG, Vaccargiu S, Marrosu G, et al. Neurology Feb 1997;48:489-493). PMP22 protein maintains normal function of peripheral nerve myelin, and the location of the mutation determines the phenotype of HMSN I disease.

DEVELOPMENTAL AND DEGENERATIVE DISORDERS

NEUROANATOMY IN RETT SYNDROME

Volumetric MRI analyses of the cerebral cortex and posterior fossa of 20 girls with Rett syndrome (RS) were compared with individually-matched normal controls at the Kennedy Krieger Institute, The Johns Hopkins University School of Medicine, Baltimore, MD. Gray and white-matter tissue volumes, with the exception of the pons, showed reductions in RS compared to controls. The caudate nucleus showed a disproportionate volume reduction. A reduction in cerebellar measurements followed the general reduction in brain size in RS. Age-related changes were not different from controls, and a progressive neurodegeneration was not evident. Brains of monozygotic twins discordant for RS revealed reduced gray-matter volumes in the RS twin but not in her sister. (Subramaniam B, Naidu S, Reiss AL. Neuroanatomy in Rett syndrome: cerebral cortex and posterior fossa. Neurology Feb 1997;48:399-407). (Reprints: Dr Allan L Reiss, Kennedy Krieger Institute, 707 N Broadway, Rm 509, Baltimore, MD 21205).

COMMENT. These volumetric neuroanatomical studies of Rett syndrome are important in our understanding of developmental and clinical-anatomical correlations. The etiology of RS remains an enigma, and even the genetics of the disorder are undetermined. My colleague, Dr John Wilson, in his introduction to a chapter on RS in Vol III, Progress in Pediatric Neurology, 1997, suggests the process of *apoptosis* as a possible explanation for RS.

Apoptosis in development and disease of the nervous system: 1. Naturally occurring cell death in the developing nervous system, is discussed by Narayanan V, University of Pittsburgh, PA. (Pediatr Neurol Jan 1997;16:9-13). The occurrence of cell degeneration during normal neural development has been studied experimentally in chick embryos, and apoptosis is proposed as the mechanism of infantile spinal muscular atrophy. Substances produced by the target tissue influence the survival of developing neurons. Limb bud removal causes cell degeneration in brachial or lumbosacral