Anticipation in HD and Creutzfeldt–Jakob disease

Two articles and an accompanying editorial consider important developments relating anticipation (the worsening of hereditary disease through successive generations) to the understanding of disease pathogenesis. MacDonald et al. (p. 1330) note that progressive trinucleotide expansion accounts for only 70% of the variation in the age at onset of Huntington's disease (HD). They found linkage of a chromosome 6 region to be associated with younger onset of HD. This region contains GluR6, the kainate receptor. If the kainate receptor is indeed the linked gene, it could relate to glutamatergic neurotoxicity proposed as pathogenetic in HD. The Brief Communication by Rosenmann et al. (p. 1328) reports anticipation in familial Creutzfeldt–Jakob disease in Libyan Jewish families. The cause of this observation is not clear. The editorial by Ashizawa and Conneally points out that ascertainment biases can result in anticipation that is apparent but not real. However, geneticists are now understandably cautious about dismissing anticipation because it was denied for decades until the trinucleotide repeat diseases were discovered. Ashizawa and Conneally note that there are over 25 diseases with reported anticipation, and understanding why may explain the causes of many of them.

Neuromuscular disease

Trojan et al. (p. 1225) report that pyridostigmine was of no benefit in postpolio syndrome (PPS). ♦ Dalakas' editorial (p. 1166) reviews the pathogenesis of PPS and discusses the inherent limitations that it places on any treatment strategy. Dalakas also outlines sensible ap-

proaches for counseling patients with PPS.

Restless legs syndrome

Entezari-Taher et al. (p. 1201) studied CNS motor circuitry with transcranial magnetic stimulation in patients with primary restless legs syndrome (RLS). (pRLS can be secondary to peripheral neuropathy.) Patients with pRLS had evidence for decreased supraspinal inhibition. This change in inhibition may account for the response of RLS to dopaminergic treatments because there is accumulating evidence for abnormal subcortical dopaminergic systems in RLS.

Multiple sclerosis

Vickrey et al. (p. 1190) questioned a large sample of MS patients to address a matter of great importance to neurologists: Does the general neurologist do equally well in caring for patients with MS as do MS specialists? In general, care was comparable. However, MS specialists were more likely to discuss, prescribe, and maintain interferon-beta 1b treatment. This type of study focuses attention on how neurologists—both generalists and subspecialists—can improve their patient care.

♦ Fernandez-Arquero et al. (p. 1361) studied factors determining genetic susceptibility in Spanish patients with MS. A polymorphism of tumor necrosis factor (TNF) correlated with MS susceptibility. The TNF and previously reported human leukocyte antigen influence were both important. This link with TNF, an inflammation-related protein, is noteworthy and has been seen in autoimmune diseases.

Chiari I malformations in children

Wu et al. (p. 1271) examined MRI findings and clinical symptoms

and signs in 49 children with Chiari I malformations ascertained by MRI testing. Although the study was retrospective, the data suggest that symptoms of Chiari I (headache, neck pain, and ataxia) often considered to occur only in adults are in fact frequent in children. The degree of tonsillar ectopia by MRI correlated with symptoms.

Epilepsy

Hennessy et al. (p. 1276) review the early and late mortality after temporal lobe epilepsy surgery in 299 of 305 patients operated on over a 20-year period (1975–1995) at The Maudsley Hospital, London. During an average follow-up period of 9 years, 20 of 299 patients died: 2 operative deaths and 18 later deaths. Thirteen of the later deaths were attributed to epilepsy, including 6 with sudden death. Surgery appeared to improve survival of epileptic patients with epilepsia overall, but not of those operated on for right-sided mesial temporal sclerosis.

Stroke

Gorter et al. (p. 1319) examined the predictors of hemorrhage in a population of 651 patients anticoagulated for arterial occlusive ischemic stroke as part of a clinical trial (Stroke Prevention in Reversible Ischemia Trial) for nonembolic strokes of arterial origin. The study had been terminated early because of the 7% incidence of major bleeding. Hemorrhage was more frequent with leukoaraiosis with those over 65 years of age, and with higher international normalized ratios. In comparison with anticoagulation for embolic strokes, there was a 19-fold increase in hemorrhage.

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