Nonsyndromic mental retardation

There are many well-defined syndromes that cause mental retardation: Down syndrome, severe perinatal birth injury, and metabolic disorders, among many other causes.

Nonsyndromic mental retardation is the most common developmental disorder but its causes have not been defined. Higgins et al. (p. 335) studied a kindred with mental retardation within a large genealogic database. Patients with mental retardation appeared otherwise normal. Mental retardation was autosomal recessive and discovered to be linked to chromosome 3p25pter. This region has also been implicated in mental retardation with the chromosome 3p-deletion syndrome. ♦ The accompanying editorial by Opitz (p. 328) considers the inherent limitations of defining genes that cause mental retardation and points out that terming the mental retardation “nonsyndromal” is likely to be incorrect as new techniques for the study of facial morphology are applied.

Bioinformatics in Lafora's disease: From mutation to disease mechanism

Lafora's disease—a progressive myoclonic epilepsy of children—is caused by mutation in the gene for laforin. The function of the protein and the range of mutations are not known.

Minassian et al. (p. 341) detected seven new mutations in exon 1 of the laforin gene and used bioinformatic analyses to unravel laforin's function, suggesting it binds and cleaves carbohydrates. A disturbance of its function results in the polyglucosan bodies that

characterize the disease. ♦ The accompanying editorial by Graf (p. 331) defines terms and concepts and offers a vision of how bioinformatics will take us from defining the sequence of the human genome to an understanding of how genes control cell expression of proteins crucial for CNS function.

Monitoring retinal function during drug treatment

Vigabatrin is an anticonvulsant in wide use in Canada and abroad, where it is considered first-line treatment for infantile spasms. It is often acquired for use by the parents of patients in the United States. Vigabatrin use is associated with visual field defects—a subject of previous *Neurology* articles. Harding et al. (p. 347) studied the ability of electroretinography to detect and quantitate abnormalities—an observation of particular importance in children or incapacitated adults who cannot perform reliable visual field tests. The electroretinography proved of great value, a topic amplified by Miller in the accompanying editorial (p. 333). Miller discusses the practical uses of electroretinography and why it is so valuable, particularly because other GABAergic drugs are coming into use and electroretinography appears to detect toxicity before it becomes clinically evident.

Stroke: Pseudo abducens palsy and convergence retraction nystagmus

Pullicino et al. (p. 352) present the clinical and imaging findings of seven patients with a pseudo VIth palsy and top-of-the basilar infarction. A pseudo VIth palsy may suggest a pontine lesion to

the clinician but localizes to the diencephalon. The combination of pseudo VIth palsy and convergence retraction nystagmus points to a lesion rostral to the oculomotor nucleus in the midbrain.

Anticipation in proximal myotonic myopathy (PROMM)

For some PROMM kindreds, the responsible gene is on chromosome 3q. Schneider et al. (p. 383) report 3q-linked PROMM families (also termed myotonic dystrophy type 2) in which there was evidence for the disease having an earlier age at onset in successive generations. Such anticipation suggests that PROMM, like myotonic dystrophy type 1, may be a trinucleotide repeat disorder.

Globus pallidus stimulation: Effects on cognition and mood in PD

Pillon et al. (p. 411) studied deep brain stimulation for neuro-behavioral effects during deep brain stimulation “on” and “off” in 56 patients 3 and 12 months after implantation. With deep brain stimulation “on,” patients improved in psychomotor speed and working memory. The only evident worsening was in verbal fluency.

AIDS myelopathy and CSF/HIV viral load

Viral load has become a standard measure for following HIV-infected patients being treated with antiretroviral agents. CSF viral load correlates with neurocognitive impairment. Geraci et al. (p. 440) compared 16 patients with AIDS myelopathy to 16 HIV-infected patients without myelopathy. CSF HIV-1 RNA did not correlate with the presence or severity of myelopathy.

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