## DEVELOPMENTAL DISORDERS

### NEURAL TUBE DEFECTS AND CHROMOSOME DELETIONS

Patients with neural tube defects (NTDs) complicated by congenital heart defects, facial anomalies, thymic hypoplasia, cleft lip or palate, or hypocalcemia and a family history of NTDs and other anomalies were tested for 22q11 deletions at the Departments of Pediatrics and Molecular Genetics, Oregon Health Sciences University, Eugene, Oregon. Of 295 patients identified with NTDs, 22 had at least one more clinical anomaly and/or a positive family history. Fetal alcohol and valproate syndromes were excluded. Cytogenetic analysis and molecular testing on 16 revealed 22q11 deletions in 3 and normal results in 13. Deletion of 22q11 was an infrequent cause of NTDs. (Nickel RE, Magenis RE. Neural tube defects and deletions of 22q11. Am J Med Genet Dec 1996;66:25-27). (Reprints: Dr Robert E Nickel, 901 East 18th Avenue, Eugene, OR 97403).

COMMENT. Cytogenetic testing for the 22q11 deletion is recommended in infants with neural tube defects complicated by congenital heart defects, particularly conotruncal defect, and in those with a family history of the heart defect, velo-cardio-facial syndrome, or DiGeorge sequence.

### PRESYMPTOMATIC DIAGNOSIS OF NEUROFIBROMATOSIS 2

The clinical spectrum of neurofibromatosis 2 (NF2) at the time of presymptomatic DNA diagnosis in at-risk first-degree relatives in five families were studied at the Cedars-Sinai Medical Center, UCLA School of Medicine, Los Angeles, and the Neurofibromatosis Institute, La Crescenta, CA. With molecular genetic analysis, 11 first-degree relatives were predicted to be at high risk, and 20 at low risk of carrying an NF2 mutation. Five mutation carriers, including a 31-year-old, had no clinical manifestations, while 4, including a 7-year-old, had vestibular schwannomas (VS), early-onset cataracts, or both. The identification of presymptomatic NF2 mutation carriers by DNA diagnosis permits improved genetic counselling and clinical management in at-risk subjects. The early detection of VS by gadolinium-enhanced MRI can improve surgical outcome. (Baser ME, Mautner VF, Ragge NK et al. Presymptomatic diagnosis of neurofibromatosis 2 using linked genetic markers, neuroimaging, and ocular examinations. Neurology Nov 1996;47:1269-1277). (Reprints: Dr Michael E Blaser, 11746 Bellagio Rd, #308, Los Angeles, CA 90049 or Dr Stefan-M Pulst, Division of Neurology, Rm 8920 South Tower, Cedars-Sinai Medical Center, 8700 Beverly Blvd, Los Angeles, CA 90048).

COMMENT. In NF2 mutation carriers, DNA testing may lead to early diagnosis, and optimal treatment and counselling. However, ethical factors must be considered in testing children because of health insurance and other discriminating issues.

## HEADACHE DISORDERS

### JUVENILE IDIOPATHIC STABBING HEADACHE

A series of 83 juvenile patients with idiopathic stabbing headache, 3.3% of all juvenile patients referred because of recurrent headache, is reported from the Paediatric Neurology Services of the University of Ferrara and the University of Padua, Italy. Mean age at onset was 7 +/-3 years, and sexes were equally affected. The pain lasted a fraction of a second to a few minutes. The