an incorrect diagnosis had been made. The presenters made pleas for better education of professionals and a higher index of suspicion about FXS.

Psychologists proposed testing for patients with MR, using the Stanford Binet for IQ and the Vineland for Adaptive Behavior as a first stage, and areas of strength and weaknesses and more specific functions in second stage tests. Comparing cognitive and behavioral abilities in children with FXS and with non-FXS autism, IQ scores in FXS show a significant decline with time, whereas individuals with autism manifest relatively no change in cognitive levels. In contrast, both groups show significant longitudinal decreases in adaptive behavior. In another presentation, therapy of ADHD symptoms in 11 children with FXS, some showed an increase in performance on executive function tasks using Adderall. Other sessions on FXS were on population screening and the molecular basis of FXS. (Fryns J-P, Borghgraef M, Brown TW et al. Conference report. Am J Med Genet October 2000;94:345-360). (Respond: Jean-Pierre Fryns, Center for Human Genetics, Herestraat 49, B-3000 Leuven, Belgium).

COMMENT. The frequency of missed diagnosis in cases of FXS is of interest. Guidelines have previously been suggested to aid the practicing physician in determining which children should have a chromosomal analysis. (see Progress in Pediatric Neurology I, PNB Publishers, 1991;pp196-7).

An estimate of the prevalence of X-Linked MR syndromes is about 1:1000 mentally retarded males, 50% having FXS. The clinical manifestations of FXS include: mental retardation, developmental delay, speech delay, ADHD, temper tantrums, mouthing of objects, autistic behaviors, and impaired gross motor coordination. ADHD is diagnosed in 50% or more of FXS patients. Epilepsy occurs in 25%. A positive family history for mental retardation is reported in 65%, and 90% of cases have a family history of either retardation, learning disability, or hyperactivity.

Physical findings include: large ears (75%), prominent jaw and long face (70%), macrocephaly (40%), hypertelorism (40%), simian palmar creases (35%), and large testicles (17%). A chromosomal analysis for FX is most likely to be positive for FXS in young males with developmental delay (especially speech delay), a maternal family history for MR, and facial dysmorphism (especially large ears, long face, and prominent jaw). For further articles on FXS, see Progress in Pediatric Neurology II, PNB, 1994;295-299. Despite the guidelines and a high index of suspicion, particularly among boys with ADHD, I still find the selection of likely candidates for chromosome testing to be difficult, with few positives in my practice of pediatric neurology.

CONGENITAL MALFORMATION SYNDROMES

GOMEZ-LOPEZ-HERNANDEZ SYNDROME: EXPANDED PHENOTYPE

Since the first patient with cerebello-trigeminal-dermal dysplasia, later called Gomez-Lopez-Hernandez syndrome, was reported in 1979 by Gomez et al, six more cases have been added to the literature. Abnormalities involve the cerebellum (rhombencephalosynapsis), cranial nerves (trigeminal anesthesia), and scalp (alopecia). A further case is reported in a 19 year-old male seen at Tufts University School of Medicine, Boston, MA. The typical manifestations of the syndrome were present. In addition, some unusual findings include seizures, growth hormone deficiency and short stature, and prominent behavioral and psychiatric problems (hyperactivity, depression, self-injurious behavior, and bipolar disorder). Hyperactivity first noted at 4 years of age was later associated with attention deficit disorder. Chromosomal analysis was normal. (Brocks D,

Irons M, Sadeghi-Najad A, McCauley R, Wheeler P. Gomez-Lopez-Hernandez syndrome: Expansion of the phenotype. Am J Med Genet October 2000;94:405-408). (Respond: Patricia G Wheeler MD, Division of Genetics, NEMC Box 394, New England Medical Center, 750 Washington St, Boston, MA 02111).

COMMENT. The GLH syndrome is rare, only 8 cases reported. Physical findings that should alert suspicion are a high forehead with turricephaly, hypertelorism, narrowed palpebral fissures, temporal balding, posteriorly rotated ears, long fingers with Dupuytren contracture, and short stature. Corneal opacities may develop over time because of trigeminal anesthesia and corneal insensitivity. Head banging and head rocking, and other self-injurious behavior may occur as a result of pain insensitivity. Psychiatric problems may lead to increased risk of suicide. MRI is preferable to CT in demonstration of the cerebellar anomaly. Rhombencephalosynapsis consists of fusion of the cerebellar hemispheres and absence of the cerebellar vermis. In Greenfield's Neuropathology text (London, Arnold, 1958;p335), aplasia of the cerebellum, due to failure of the two alar plates of the rhombencephalon to unite in the formation of the corpus cerebelli, is called *palaeocerebellar aplasia (cerebellar rachischisis).* Various nuclear atrophies are also associated, including the inferior olives and dentate nuclei.

MRI IN MOBIUS SYNDROME

The MRI findings in three patients with Mobius syndrome are reported from the Hospital Vall d'Hebron, Barcelona, Spain. An 8 year-old girl had facial diplegia and convergent strabismus secondary to bilateral palsy of the facial and abducens nerves. Associated anomalies included blindness with coloboma, deafness, ear malformation, micrognatia, and mental retardation. MRI showed alterations in the shape of the brainstem and floor of the fourth ventricle, at the level of the inferior pons, with absence of the medial eminence.

A 28 year-old woman presented with congenital bilateral facial palsy, and convergent strabismus with paralysis of horizontal gaze. Associated features included small mandible, deafness, tongue hemiatrophy, and swallowing difficulties. MRI showed brainstem anomalies, including straightening of the floor of the fourth ventricle, absence of the medial colliculus of the pons (VIth and VIIth nuclei), and absence of the hypoglossal eminence in the medulla oblongata (XIIth hypoglossal nuclei). Brainstem auditory evoked potentials were abnormal. Another 28 year-old woman was referred because of headache. She had facial diplegia and convergent strabismus, with limited bilateral ocular abduction. MRI showed a V-shaped depression in the floor of the fourth ventricle due to absence of the facial colliculus. (Pedraza S, Gamez J, Rovira A, Zamora A et al. MRI findings in Mobius syndrome: correlation with clinical features. Neurology October (1 of 2) 2000;55:1058-1060). (Reprints: Dr Salvador Pedraza, Unidad de RM. IDI, Servicio de Radiologia, Hospitals Vall d'Hebron, P. Vall d'Hebron s/n, Barcelona, Spain).

COMMENT. Cranial nerves most often involved in Mobius syndrome are VII, VI, III, and VIII. The IVth, IXth, Xth, and XIIth can also be affected. Anomalies of the cerebellum, hypothalamus, pituitary gland, and limbs are sometimes noted. Genetic, ischemic, infectious, and toxic (thalidomide, alcohol) factors have been suggested in etiology. Congenital agenesis of the brainstem nuclei and malformation of the floor of the fourth ventricle are the most common pathological findings, now demonstrated by MRI. Primary nerve or myopathic involvement has also been suggested in some cases. Electrophysiologic studies support the involvement of brainstem nuclei and internuclear connections.