IDIOPATHIC INFANTILE NYSTAGMUS, WITH AND WITHOUT *FRMD7* GENE MUTATIONS

Clinical features and eye movement recordings of 90 subjects with mutations in the gene (*FRMD7* group) were compared to 48 without mutations (non-*FRMD7* group) but with clinical idiopathic infantile nystagmus (IIN), in a study at University of Leicester, Leicester Royal Infirmary, Leeds General Infirmary, Royal Preston Hospital, Addenbrooks Hospital, Cambridge, UK; Wills Eye Hospital, Philadelphia, USA; and Medical University Graz, Austria. Visual acuity and binocular vision were generally normal in both groups. Prevalence of strabismus was similar and occurred in 7.8% of mutation and 10% of non-FRMD7 patients. Anomalous head posture was significantly more frequent in the non-FRMD7 group ($P<0.0001$); moderate (5-15 degrees) in 24% and severe (>15) in 27% vs 17% only moderately affected in the FRMD7 group. Amplitude of nystagmus was lower at primary position in the FRMD7 group ($P<0.0001$) compared to non-FRMD7 group ($P=0.83$). Pendular nystagmus was more frequent in the FRMD7 group ($P=0.003$). Obligate female carriers of an FRMD7 mutation were clinically affected in 53%. Visual acuity of affected females was better than affected males ($P=0.014$). FRMD7 is a major cause of X-linked IIN. The findings are helpful in genetic counselling of patients with idiopathic infantile nystagmus. (Thomas S, Proudlock FA, Sarvananthan N, et al. Phenotypical characteristics of idiopathic infantile nystagmus with and without mutations in FRMD7. **Brain** May 2008;131:1259-1267). (Respond: Prof Irene Gottlob, Ophthalmology Group, University of Leicester, Faculty of Medicine & Biological Sciences, Robert Kilpatrick Clinical Sciences Building, Leicester Royal Infirmary, PO Box 65, Leicester, LE2 7LX, UK. E-mail: ig15@le.ac.uk).

COMMENT. Idiopathic infantile nystagmus is noted in the first months of life, and diagnosis is dependent on the absence of albinism and congenital night blindness or achromatopsia. Prevalence of nystagmus is estimated at 2.4/1000 (Sarvananthan N et al. **Invest Ophthalmol Vis Sci** 2006;47:E-Abstract 2656). Inheritance is X-linked most commonly, and the present authors report multiple mutations in FRMD7 gene localized to chromosome X (NYS1). In addition, autosomal dominant inheritance is described, localized to chromosome 6, as well as autosomal recessive inheritance. In patients with idiopathic infantile nystagmus, clinical characteristics of those with FRMD7 mutation are similar to a non-FRMD7 mutation group, except abnormal head posture is significantly less frequent and amplitude of nystagmus in the primary position is lower in those with mutations. Most patients with IIN have good visual acuity and stereopsis and strabismus is infrequent, in both those with and without mutations. Mutations in FRMD7 are the major cause of inherited IIN. Cerebellar dysfunction is the most likely cause of nystagmus related to the FRMD7 gene in IIN. (Glasauer S. **Ann NY Acad Sci** 2003;1004:206-219).

MANAGEMENT OF NEONATAL SEIZURES

The generally accepted clinical approaches to neonatal seizures were determined by questionnaires completed by pediatric neurologists and neonatologists representing all the pediatric neurology units and all departments of neonatology in Isrrael, and were evaluated at