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Letter to the Editor

Familial isolated congenital nystagmus in a Turkish infant

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Nystagmus is an involuntary, rhythmic oscillation of the eyes in which at least one phase is slow. Its congenital form may be associated with blindness, familial and idiopathic. In familial form, the pattern of inheritance may be autosomal recessive, autosomal dominant, or X linked [1,2]. Herein, we report an infant with familial isolated congenital nystagmus due to unusual presentation. To the best of our knowledge this is the first Turkish family in the English literature.

A 3-month-old boy was admitted with rhythmic oscillations on both eyes, which were firstly noted just after birth. There was no any decrease or increase in his complaint since birth. He was the product of a 40 weeks term uncomplicated gestation and labor. There was no consanguinity between the parents. He was the 2nd baby of the family, and the elder child was healthy. Similarly, the patient's mother and the mother's father and the mother's grandmother had isolated congenital nystagmus. On physical examination, head control was excellent. Bilateral horizontal nystagmus (pendular type) was diagnosed. Fundoscopic examination was normal. Other systemic and neurological findings were normal. On laboratory studies urine and blood amino acids, serum lactic acid and pyruvic acid concentrations were normal. Flash visual evoked potential was also unremarkable. Cranial magnetic resonance imaging was normal. His mother's physical and neurological examination was also normal with the exception of horizontal nystagmus (pendular type). The mother's visual acuity was intact, but she had bilateral myopia, 2.5 Diopters on the right and 0.5 Diopters on the left. Her optic nerve was bilateral normal.

Familial isolated congenital nystagmus is infrequently reported in the literature [1–3]. Lorenz and Gampe [3] reviewed 180 patients with congenital nystagmus and found that a sensory defect nystagmus was present in 142 patients (79%). The diagnoses were as follows: albinism (any form) in 56 patients (30%), progressive photoreceptor dystrophy in 20 patients (11%), stationary cone dysfunction in 18 patients (10%), bilateral optic nerve hypoplasia in 15 patients (8%), chorioretinal or optic nerve colobomata in 10 patients (6%), aniridia and its variants in 10 patients (6%), familial isolated nystagmus in 8 patients (5%), and congenital stationary night blindness in 5 patients (3%). Thirty-eight patients (21%) could not (yet) be classified [3]. Ohba [2] reported a four-generation, family which presented with autosomal dominantly inherited congenital nystagmus, peripheral corneal opacity, and foveal hypoplasia without any iris tissue malformation. Recently, Kerrison et al. [1] have reported that NYS1 ap-

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pears to be a common gene for familial congenital idiopathic nystagmus. Our patient and his family members had isolated congenital nystagmus without any other abnormality. Based on these data the inheritance of congenital isolated nystagmus was accepted as autosomal dominant.

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