

Congenital idiopathic nystagmus in identical twins

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SUMMARY Using an infrared recording system we examined the nystagmus waveforms of a pair of monozygotic twin girls and found them to be dissimilar. It is proposed that in view of the common mode of inheritance the differences are a result of environmental influences.

The past decade has seen a transformation in the approach to the study of congenital nystagmus (CN). Though some research has been carried out on the sensory effects of the nystagmus¹⁻³ most has been directed towards understanding the motor characteristics of the disorder.⁴⁻¹³ Notably, Dell'Osso and his colleagues have carefully established that 12 distinct waveforms may be distinguished.^{8,13} These oscillations are not related to any one aetiology.^{9,13} Nor do they appear to have a common genetic heritage, since 3 members of the same family have been shown to have different nystagmus waveforms.⁷

In the present study the eye movements of a pair of identical twins with CN will be described. The twin girls, being genetically identical, should presumably

provide a pointer to the extent of environmental influences, since any difference arising between the monozygotic twins must be due to nongenetic factors.

Case report

The twins were full-term babies born on 12 August 1967, and the nystagmus was noticed soon after birth. There is no known family history of congenital nystagmus. The twins (Fig. 1) were 13 years old at the time of this study.

An analysis of finger print total ridge counts and serum and red cell polymorphic markers indicated that the girls are monozygotic twins. The twins were concordant for the following markers: ABO, MNS, rhesus, Lutheran, Kell, Lewis, Duffy, Kidd, secretor, haptoglobin, transferrin, GM, Inv, esterase 2,

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Fig. 1 Identical twin girls with a bilateral, conjugate, horizontal, idiopathic congenital nystagmus. Twin A is pictured on the left. Before this photograph was taken both girls had their left pupils dilated so that their fundi could be photographed.

acid phosphatase, PGM, 6PGD, and adenosine deaminase.

The ocular media and fundi appeared normal. Neither twin had a head turn nor any abnormal head oscillation. Both tanned well in the sun and could be shown to have macular pigment.¹⁴ Case histories and clinical tests suggested that the nystagmus is idiopathic.

For twin A refraction revealed OD +2.00/-4.75×40 6/9, OS +1.50/-6.00×160 6/12; and twin B, OD +1.50/-4.50×25, 6/18, OS +1.75/-5.00×155, 6/18-2. Visual acuities were taken when both the head and the eyes were in the primary position. Photoelectric keratoscope¹⁵ measurements confirmed that the astigmatic nature of the refractive errors reflected to a large extent the toroidal shape of the anterior surface of the corneas. The shape factor of each eye, which is a measure of the rate of peripheral flattening of the cornea, was computed and found not to differ significantly from that of the normal population.

Infrared eye movement recordings were made of

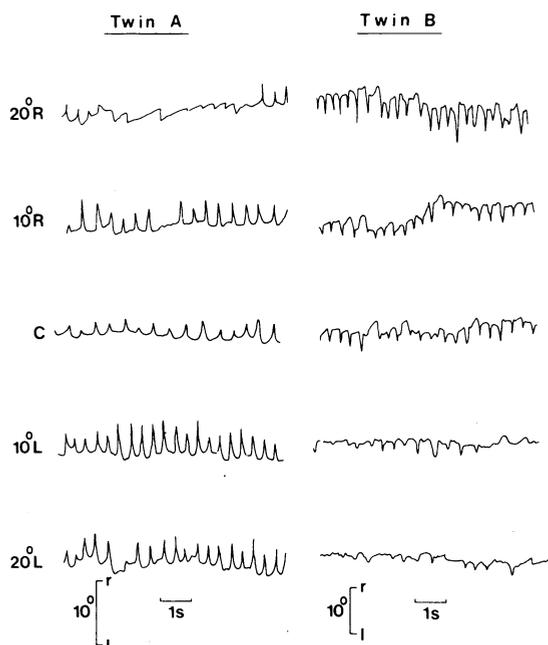


Fig. 2 Infrared eye movement recordings of the nystagmus of the twins taken at 5 different positions of gaze: Centre (C), 10° and 20° to the right (R) and 10° and 20° to the left (L). During central gaze both twins have a pseudocycloid waveform. The fast phase is directed towards the left for twin A and to the right for twin B. This accounts for the noticeable increase in the amplitude of the nystagmus when twin A looks to the left and twin B looks to the right. All recordings illustrate the movements of the right eye of each twin. Note different 10° calibration bars for the 2 subjects.

the nystagmus with both eyes being monitored simultaneously. For both twins the nystagmus was bilateral, and the movement was almost exclusively in the horizontal plane. Fig. 2 illustrates the nystagmus waveforms in 5 positions of gaze; the primary position (C), $\pm 10^\circ$ and $\pm 20^\circ$ from centre. During central fixation both twins have a pseudocycloid waveform, but the fast phases are in opposite directions, with the fast phase to the left for twin A to the right for twin B. As Dell'Osso and Daroff⁸ have pointed out, this waveform is often misidentified as being pendular if position only is monitored. With the aid of a velocity record the waveform can be seen to consist of an accelerating drift off target which is terminated by a braking saccade and followed by a decelerating slow eye movement which brings the eyes back on target. In accordance with Dell'Osso and Daroff⁸ we defined the direction of the pseudocycloid nystagmus as the direction of the braking saccade independent of its amplitude.

The flattened peaks seen in the waveform of twin A would suggest a foveation time greater in duration than that of her sister. This is probably the explanation for the differences found in their visual acuity, the higher visual acuity of twin A being associated with the retinal image spending longer periods of time within the foveal area. Null zones were found in opposite directions of gaze, being on gaze right for twin A and gaze left for twin B. This was not entirely unexpected, particularly in view of the direction of the fast beats monitored on central fixation. In neither twin did the act of convergence attenuate the nystagmus.

Discussion

Though Forssman and Ringner¹⁶ have estimated the frequency of idiopathic CN in the general population to be 1 in 1,000 males and 1 in 2,800 females, the chances of encountering monozygotic twin girls with idiopathic CN are, to say the least, small. It is therefore regrettable that the only other description of such a case to be found in the literature should omit any quantitative reference to the nystagmus waveform.¹⁷ In this study we have established that the characteristics of the nystagmus waveforms for our monozygotic twins are indeed dissimilar. This being the case, and in view of the common mode of inheritance, it seems reasonable to suppose that these differences result from environmental influences.

We are greatly indebted to Dr Michael Connor, of the West of Scotland Regional Genetics Centre, for confirming the monozygotic status of the twins; Ian Smith, of Glasgow Eye Infirmary, for photographing the twins; Ms W. Wiggie for translating the German paper¹⁷; and Ms Janice Patrick for typing the manuscript.

This study was supported by an MRC Project Grant G979/1025N. C.M.D. held an MRC studentship (77/7291).

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