The Torsional Component of "Horizontal" Congenital Nystagmus

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This article is dedicated to the memory of Lea Averbuch-Heller, MD (1958–2000). We began recording torsional data from occasional adult patients with congenital nystagmus earlier, but this study did not formally begin until 1997, while she was a fellow in the Ocular Motor Neurophysiology Laboratory and a member of the Department of Neurology, Case Western Reserve University. It was set aside while other studies were more actively pursued and resurrected the day after her tragic accidental death. Dr. Averbuch-Heller was uncommonly prolific during her foreshortened career (42 journal publications, 17 abstracts, four reviews, and six book chapters). A warm and compassionate woman, with a bright smile that could light up a room, Lea was a mensch. We who were fortunate enough to have known her have lost a true friend and are left with the sadness of an unfillable void.

Objectives: To study the relationship between the major horizontal and minor torsional components of congenital nystagmus to elucidate the diagnostic importance, effects on vision, and pathogenetic implications of the torsional components.

Methods: We recorded the eye movements of 13 subjects with congenital nystagmus using a three-dimensional magnetic search coil technique over a 15-year period. The subjects fixated on stationary targets straight ahead and along the horizontal and vertical meridians. Six of the 10 subjects with horizontal congenital nystagmus were asymptomatic; the remaining 4 (plus two with a vertical component to their congenital nystagmus) had adult-onset symptoms. An additional subject without symptoms had a vertical congenital nystagmus component plus seesaw nystagmus; one of the symptomatic subjects also had seesaw nystagmus.

Results: In all 13 subjects, the horizontal and torsional cycles were phase-locked, and positive horizontal (rightward), vertical (upward, if any), and torsional (clockwise) motion coincided in 10 subjects. That is, rightward horizontal eye rotation coincided with clockwise curvilinear motion (rightward and downward) of the upper pole of each eye. During the horizontal foveation periods, torsional motion was also of low velocity. In 2 of 13 subjects, the torsional waveforms differed from those in the horizontal plane; in others, the direction or the variation with gaze angle differed from that predicted by Listing. In each of the 13 subjects, the torsional components ranged from 8.16% to 94.42% (median, 32.94%) of the peak-to-peak magnitudes of the congenital nystagmus. In most cases, the measured torsion was far greater than that predicted by Listing's law for a worst-case analysis (range, 0.69–11.83%; median, 4.91%). The torsional components of the two subjects with seesaw nystagmus were 60.48% and 264.02%.

Conclusions: The manner in which the horizontal and torsional components of "horizontal" congenital nystagmus were phase-locked made clinical detection of the torsional component difficult. Most "horizontal" congenital nystagmus is actually horizontal-torsional congenital nystagmus. Visual acuity during horizontal foveation periods is not significantly diminished by torsional motion. In only one subject did the torsional component of the congenital nystagmus have an amplitude equivalent to Listing torsion; in the other 12 subjects, torsion exceeded our estimate of what Listing's law would predict. The torsional components of the seesaw nystagmus in two subjects also greatly exceeded the torsion predicted by Listing torsion. The most parsimonious explanation for our data is that the cyclic torsion in congenital nystagmus was generated centrally and not a result of Listing torsion, mechanical crosstalk, or normal or abnormal extraocular-muscle (plant)

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At the time of her death (September 26, 2000), Dr. Averbuch-Heller was a member of the Department of Neurology, Tel-Aviv University, Tel-Aviv, Israel.

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dynamics. Further measurements are needed to confirm this hypothesis.

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Congenital nystagmus usually manifests itself in the horizontal plane, with only rare cases having a vertical component. In patients with this configuration, the congenital nystagmus could have either a diagonal or an elliptical trajectory, depending on the relative amplitudes and phases of the oscillations in each plane. Because the magnitude and speed of the horizontal oscillations of congenital nystagmus are often large, it is difficult to determine, by inspection alone, whether there is an accompanying vertical or torsional component. The development of the means to make precise measurements of eye rotations in all three directions (1,2) has made it possible to address these issues. We present here our findings from 13 subjects whose waveforms typical of congenital nystagmus we have recorded over a 15-year period.

There have been only two other studies of the torsional component in subjects with clinically classified "horizontal" congenital nystagmus. One reported that the torsional component was related to the horizontal component in three of the four subjects recorded (3). The second found that the "basic configuration" of Listing's plane remained intact in congenital nystagmus but that there were short-term (cycle-to-cycle) violations of Listing's law (4). In this report, we examine further the torsional components of the "horizontal" congenital nystagmus in 13 individuals and discuss the cause of this clinically hidden oscillation. We investigate whether it is due to Listing torsion (5) or to an underlying instability in the torsion control subsystem and emphasize the relationship between both components of the congenital nystagmus during the foveation periods, when visual acuity is most affected by retinal image position and velocity.

METHODS

Patient Selection

During the past 15 years, we recorded the eye movements of 13 patients, using the three-dimensional magnetic search coil technique, who showed nystagmus waveforms typical of congenital nystagmus. Not all patients were thought to have congenital nystagmus when referred, and some presented with visual complaints or dizziness. Their clinical and oculographic features are summarized in Table 1. Over this same time period, we made a total of 465 recordings of subjects with congenital nystagmus, but mainly preferred to use an infrared technique, especially in children. Thus, the selection of the subjects reported here is influenced by age and also, in six patients, by visual complaints, sometimes appearing in adulthood. The eye movements of 13 subjects with congenital nystagmus (6 male and 7 female, ranging in age from 25 to 45 years) were recorded. In addition to six subjects (S1–S6) whose congenital nystagmus was predominantly horizontal and who exhibited no symptoms, we studied six subjects with congenital nystagmus plus adult-onset symptoms (including oscillopsia), suggesting possible additional oscillations. In the latter group, four cases were predominantly horizontal, one (S11) had diagonal congenital nystagmus, and one (S13) had seesaw nystagmus and diagonal congenital nystagmus. Also studied was the patient known as S12, who had no symptoms but had seesaw nystagmus and diagonal congenital nystagmus. This research, involving human subjects, followed the Declaration of Helsinki, and informed consent was obtained after the nature and possible consequences of the study were explained. The research was approved by an institutional review board.

Recording

Horizontal, vertical, and torsional rotations of both eyes were recorded in the Ocular Motor Neurophysiology Laboratory using the "double loop" scleral search coil method with 6-foot field coils (CNC Engineering, Seattle, WA). The coil system bandwidth was 0 to 150 Hz, with a linear range of greater than ±20 degrees and a sensitivity of 0.1 degree in all three planes. The subject's head remained within the 30 cm cube of the magnetic field, where the translation artifact was less than 0.03 degrees/cm. Horizontal and vertical rotations of the coils of up to 20 degrees produced less than 0.5 degrees of crosstalk in the torsional channel. Data were filtered (bandwidth 0–90 Hz) and digitized at 200 Hz (S6 at 400 Hz and S12 at 500 Hz) with 16-bit resolution using a PCI-MIO-16XE50 DAQ board (National Instruments, Austin, TX). Scleral-coil (Skalar, Delft, the Netherlands) gains were calibrated using a protractor device capable of rotations in each plane. Coil data were adjusted for bias during analysis. The mean foveation position of each eye was set to 0 degrees to align it to the target position during fixation in primary (central, see later discussion) position. This is routinely done for most other types of eye-movement recording methods, and, although it does not guarantee that the 0-degree eye position coincides with a target image on the center of the fovea, it does place 0 degrees at the subject's chosen point of fixation. Except for rare cases of extrafoveal fixation or certain types of foveal aplasia, it is reasonable to equate 0 degrees with the foveal center, especially when the subject has good vision.

Protocol

During search-coil recording, the subject was seated 1.2 m in front of a translucent screen upon which the targets were projected. The head was stabilized in the straight-ahead position and the subject was instructed to move only

TABLE 1. ***Torsional nystagmus components and waveforms***

S	Nystagmus	Waveforms			Quad	Average amplitudes			Symptoms
		Hor	Vert	Tor		Hor (°p-p)	Vert (°p-p)	Tor (°p-p)	
1	CN-h	$+PP_{fs}$	—	$+PP_{fs}$	T	1.80	—	0.87	
2	CN	$-J$	—	$-J$	T	9.82	—	2.47	
		$+J_{ef}$	—	$+J_{ef}$	N	4.46	—	0.80	
3	CN	$+J_{ef}$	—	$+J_{ef}$	N	0.69	—	0.65	
		$-PP_{fs}$	—	$-PP_{fs}$	N	2.56	—	1.29	
4*	CN-h	$-J_{ef}$	—	$+P_{fs}$	T	2.65	—	0.88	
		$+J_{ef}$	—	$-PC$	N	1.37	—	0.84	
5	CN	$-J_{ef}$	—	$-J_{ef}$	T	6.62	—	6.02	
6	CN/APAN	$-J_{ef}$	—	$-J_{ef}$	N	5.53	—	0.46	
7	CN†	$+J_{ef}$	—	$+J_{ef}$	N	1.17	—	0.26	OSOP, diplopia
8	CN, DYSM	$+J_{ef}$	—	$+J_{ef}$	N	2.62	—	0.36	Poor vision
9	CN, PAN	$+J$	—	$+J$	T	7.47	—	0.74	OSOP, dizziness
10	CN‡	$+J$	—	$+J$	N	10.74	—	3.56	OSOP, imbalance
11*	CN§	$-P_{fs}$	$-P_{fs}$	$+P_{fs}$	N	6.91	9.58	1.86	OSOP
		$-J$	$-PC$	$+PC$	N	3.51	4.05	1.58	
		$-J_{ef}$	$-PC$	$+PC$	N	2.83	4.92	2.94	
12	CN	$-PC$	$-PC$	$+PC$	T	5.72	2.12	2.45	
	SSN(RE)	$+P$	$+P$	$-P$		8.58	5.70	6.28	
13	CN	$-PC$	$+PC$	$-PC$	T	3.06	1.11	2.58	OSOP, blurring, diplopia
	SSN(RE)	$+P$	$+P$	$-P$		5.59	4.86	18.26	

*, torsional and horizontal waveforms differ; †, "aware his eyes were moving"; ‡, vestibular hypofunction; §, Lithium; +, rightward, upward, or clockwise; −, leftward, downward, or counterclockwise; APAN, asymmetric (a)periodic alternating nystagmus; CN, congenital nystagmus; CN-h, hereditary congenital nystagmus; DYSM, dysmetria (saccadic); FLUT, flutter; Hor, horizontal; J, jerk; J_{ef}, jerk with extended foveation; P, pendular; P_{fs}, pendular with foveating saccades; PC, pseudocycloid; PP_{fs}, pseudopendular with foveating saccades; OSOP, oscillopsia; Quad, quadrant (N—nasal, T—temporal); RE, right eye; S, subject; SSN, see-saw nystagmus; Tor, torsional; Vert, vertical; VN, vestibular nystagmus.

the eyes to view each target as it was turned on. Subjects attempted to fixate a laser spot located in a central position, or when it was displaced 5 to 15 degrees horizontally or vertically. The specific data used for this study were retrieved from those collected using our general protocol for congenital nystagmus patients, which includes horizontal and vertical saccades, smooth pursuit, and vestibulo-ocular reflex and fixation, saccades, and pursuit in depth. The main purpose of the recordings was to diagnose the nystagmus, identify null angles, and suggest therapies. Documenting the torsional components was a secondary aim, and there was no time to plot the torsion over multiple fixation points, as would be required to identify Listing primary position. All test procedures were carefully explained to the subject before the experiment began and were reinforced with verbal commands during the trials. The room light could be adjusted from dim to blackout to minimize extraneous visual stimuli. An experiment consisted of one to 10 trials, each lasting less than 1 minute, with extra time allowed between trials for the subject to rest. Trials were kept this short to guard against boredom, because congenital nystagmus intensity is known to decrease with inattention.

Ocular Motor Conventions

Standard, historical (Kestenbaum) ocular motor conventions were followed (6). For movements in all planes, directions are from the subject's point of view (positive horizontal, vertical, and torsional directions were right, up, and clockwise, respectively). These conventions preserve consistency between planes as well as between the directions of eye movement and those of perceived motion (oscillopsia), if present. Defined motions are independent of head position ("up" is toward the forehead and "clockwise" is curvilinear rotation of the top pole of the globe to the subject's right and down). In keeping with historical precedent and clinical usage, primary (or central) position corresponded to the position of the eyes during fixation of a distant target located straight ahead on the horizontal meridian. To avoid ambiguity, we used the term "Listing primary position" when referring to the position from which

all horizontal or vertical movements have no induced torsion. We note that primary/central position is the *same* fixed position in space for all normal individuals, whereas Listing primary position is idiosyncratic, can be time-varying, and does not represent a fixed position in space.

Analysis

Data analysis (and filtering, if required), statistical computation of means and standard deviations, and graphic presentation were performed using the MATLAB (The MathWorks, Natick, MA) software for scientific computing.

Using Fick coordinates, Listing torsion is given by the following equation:

$$\psi = \arcsin \{\sin \phi \sin \theta/(1 + \cos \phi \cos \theta)\},$$

where ψ is clockwise (positive) torsion, ϕ is rightward (positive) motion, and θ is upward (positive) motion (7). Lacking the data to accurately determine Listing primary position, we presumed it to be at primary position for our worst-case analysis. For the purposes of comparing the torsional component of congenital nystagmus with that predicted by Listing's law, congenital nystagmus magnitude was calculated as $(H^2 + V^2)^{1/2}$, where H and V are the respective horizontal and vertical (if any) peak-to-peak amplitudes of the congenital nystagmus cycle. The congenital nystagmus was treated as horizontal if the vertical component did not alter the above-calculated congenital nystagmus magnitude by 2% or more. In 6 of 10 such subjects, the change in congenital nystagmus magnitude was less than 0.5%.

To calculate the standard deviations of the congenital nystagmus waveform's foveation periods in a given interval of fixation (2–5 seconds), the NAFX function was used (8). The NAFX is an expansion of the NAF, a function that provides a measure of the foveation quality of a nystagmus waveform that is proportional to potential visual acuity (9). This measure of potential visual acuity for individuals with congenital nystagmus, who do not exhibit acuity-limiting symptoms, automatically calculates both the standard deviation of mean foveation position and velocity. The NAFX algorithm also calculates the foveation-period time interval. For those lacking well-developed foveation (within the ±0.5 degrees by ±4 degree/sec window), the NAFX allowed inclusion of foveation periods that fell within an expanded foveation window whose dimensions were chosen to fit each individual's foveation abilities. The effects of the torsional component of congenital nystagmus on visual acuity depend on the amplitude of the torsional oscillation during the foveation period and the standard deviation of the mean horizontal foveation positions. We are not referring to Listing torsion in this determination. If there were no horizontal motion, torsion about the center of the fovea would not be expected to diminish visual acuity. This is because a high-spatial-frequency (small) target would remain at the center of the fovea. However, if the eye is off target (as occurs when a subject with congenital nystagmus fails to exhibit well-developed foveation), torsional motion causes the image of the small target to move on the retina by an amount (Δd) equal to the product of the measured torsional angular motion (ψ_m) and the distance (r) of the target image from the center of the fovea ($\Delta d = r\psi_m$). Thus, the less well developed the horizontal foveation ability is (the greater the standard deviation of foveation positions, resulting in larger values of r), the greater the torsion-induced retinal image motion (Δd) and the greater the impact of torsional angular motion (ψ_m) on visual acuity. Conversely, the more well developed the horizontal foveation is, the less the effect of decrement torsional motion on visual acuity. Comparisons of NAFX values were made with and without inclusion of the torsional components of the congenital nystagmus.

We used nystagmus phase-plane analysis to study the simultaneous relationship between the position and velocity of the eye (and, hence, of retinal image). The trajectories seen on phase plane plots are always in a clockwise direction if the conventions of first quadrant direction and velocity being positive are adhered to. Saccadic movements appear as high-velocity clockwise loops; rightward saccades would show positive velocities and directions while leftward saccades would be negative. The trajectories of respective slow movements would also appear clockwise, with lower velocities. Phase planes provide an easy graphic identification of those periods when the target image is both stable and on the fovea. During smooth pursuit or vestibulo-ocular reflex (VOR) analysis, phase planes of retinal image motion or gaze identify those periods of stability indicative of good pursuit or VOR, respectively. Further details on the use of phase planes can be found elsewhere (10–12).

Nystagmus scanpath plots (vertical versus horizontal motion) of both position and velocity allow one to determine whether the minima for eye position and velocity are synchronous in the horizontal and vertical planes. Unlike the classic scanpaths of normal subjects during viewing of a complex scene, these are scanpaths of the eyes produced by ocular oscillations during fixation on a single target (nystagmus scanpaths). These and the phase planes can provide evidence of simultaneous satisfaction of the position and velocity criteria in both planes.

Nystagmus conjugacy plots (OS versus OD) for the horizontal, vertical, and torsional planes of motion demonstrate the relative amplitudes and phases of the motions of the two eyes.

Determining the exact Listing primary position is a time-consuming process requiring measurements at all points on a finely meshed grid. The complete evaluation of patients with nystagmus requires that a variety of clinically important measurements be made within the 30-minute time limit we impose for coil recordings. Because the data

gathered from our general recording protocol for nystagmus patients precluded evaluating Listing primary position, we used an indirect method of analysis of the torsional congenital nystagmus components. Listing torsion was plotted for the diagonal (tertiary) motion to yield the worst case (*i.e.*, greatest predicted Listing torsion) against which the torsional components of "horizontal" (or, in some cases, predominantly horizontal) congenital nystagmus were compared. The predicted, theoretical Listing torsion (LT) is set equal to that measured for tertiary motion and is given by the following equation:

$$LT = (0.1M)^2$$

where M is the magnitude of the tertiary motion [From Figure 6 (7)]. For tertiary motion measured in the nasal quadrant, the data in that same figure indicate that $LT = 1.5(0.1M)^2$.

The actual Listing torsion expected in the 10 cases with primarily horizontal congenital nystagmus motion is well below that of tertiary motion and should approach 0 for cases in which primary/central position corresponds to Listing primary position. In the three cases (S11, S12, and S13) in which the congenital nystagmus was truly diagonal, the torsional component should equal Listing torsion if that is its source.

RESULTS

The main clinical and oculographic findings of the patients studied are summarized in Table 1.

The signs of the waveforms allow interplanar comparisons of congenital nystagmus direction (positive is right, up, or clockwise, as defined in the Methods section); they were the same (positive or negative) in 10 subjects and different in three (S4, S11, and S12). In two subjects (*), the torsional and horizontal waveforms differed. The seesaw nystagmus was pendular in both S12 and S13; it was predominantly torsional in S13. To facilitate comparisons between the torsional components and Listing torsion, the quadrant of the presumed-to-be-tertiary congenital nystagmus motion was determined and included in Table 1.

Representative records from four subjects are summarized in Figures 1 through 6. Unless stated otherwise, the vertical components were either nondetectable or less than the limits described in the Methods section. In the time plots, both a foveating and a braking saccade (13,14) are identified to aid in deciphering the phase-plane plots, in which foveating and braking saccades are also identified. The phase planes also contain the foveation window, with foveal position and velocity limits for best acuity for each subject.

Nystagmus Waveforms

The plots in Figure 1 show the position, velocity, and phase planes of the pseudopendular waveform with

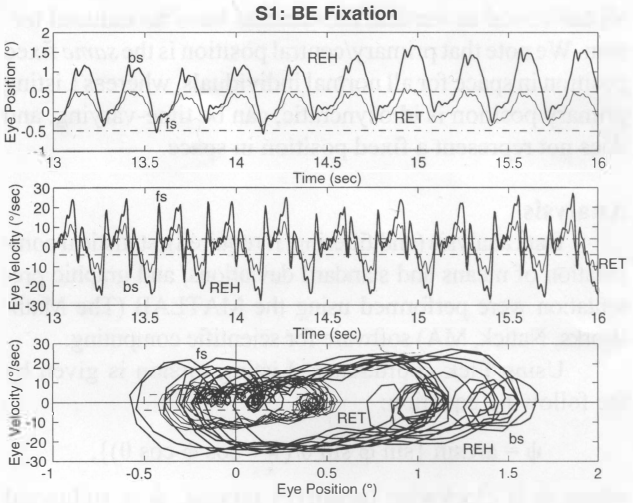

FIG. 1. Eye position (**top**) and velocity (**middle**) versus time and phase planes (**bottom**) of the horizontal and torsional components of S1's pseudopendular with foveating saccades congenital nystagmus waveform. *Dot-dashed lines* in the time plots indicate the extent of the fovea and ±4 degree/sec retinal slip velocity. *Dashed box* in the phase plane indicates the boundaries of the foveation window (±0.5 degrees by ±4 degree/sec) necessary for best visual acuity. BE, both eyes; R(L)EH, right (left) eye horizontal; R(L)ET, right (left) eye torsional; fs, foveating saccade; and bs, braking saccade. In Figures 1 through 6, horizontal motion is shown solid (heavy lines) and torsional–solid (light lines).

foveating saccades (PP_{fs}) of S1. This most complex of congenital nystagmus waveforms is mimicked exactly in the torsional plane (they are phase-locked, equivalent waveforms). In this illustration, the torsional component is roughly 50% as large as the 1.5-degree peak-to-peak horizontal component. This is the congenital nystagmus of a subject with well-developed foveation. The locations of both foveating and braking saccades are indicated in one of the congenital nystagmus cycles. The foveating and braking saccades in both planes were phase-locked, as can clearly be seen in the velocity plot. More importantly, the low-velocity foveation periods in both planes coincide.

Figure 2 shows the waveform and phase-plane plots of the jerk left waveform of S2. The torsional component mimics the horizontal component. The approximately 10- to 15-degree peak-to-peak congenital nystagmus amplitudes are larger than those of S1, and the torsional component, also larger, varies from 20% to 33% of the horizontal component. Also, the foveation is not as well developed, requiring an expanded ±2.5-degree position window and ±10-degree velocity window to define foveation. In Figure 3, we see another waveform of S2, jerk right with extended foveation (JR_{ef}). Note the dynamic overshoots in the horizontal foveating fast phases. Again, with the exception of the dynamic overshoots, the torsional and horizontal

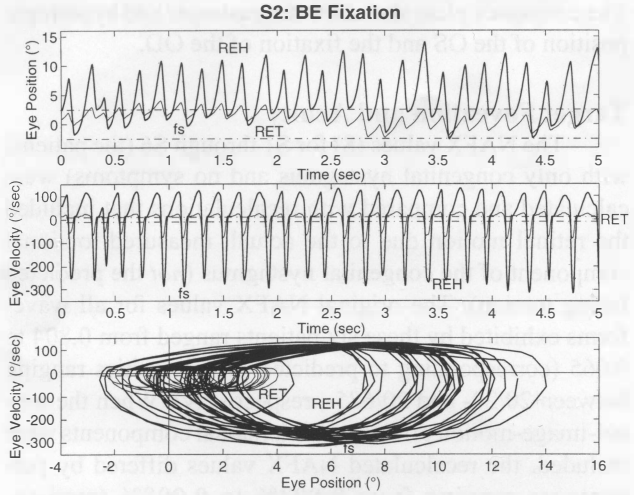

FIG. 2. Eye position (**top**) and velocity (**middle**) versus time and phase planes (**bottom**) of the horizontal and torsional components of S2's jerk left congenital nystagmus waveform. *Dashed lines* in the time plots indicate the extent of the expanded fovea (±2.5 degrees) and ±10 degree/sec retinal slip velocity. *Dashed box* in the phase plane indicates the boundaries of the expanded foveation window (±2.5 degrees by ±10 degrees/sec). BE, both eyes; R(L)EH, right (left) eye horizontal; R(L)ET, right (left) eye torsional; fs, foveating saccade; and bs, braking saccade.

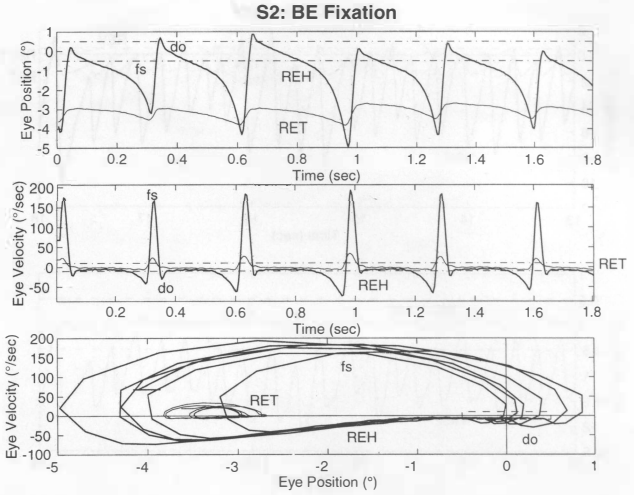

FIG. 3. Eye position (**top**) and velocity (**middle**) versus time and phase planes (**bottom**) of the horizontal and torsional components of S2's jerk right with extended foveation congenital nystagmus waveform. *Dot-dashed lines* in the time plots indicate the extent of the fovea and ±10 degrees/sec retinal slip velocity. *Dashed box* in the phase plane indicates the boundaries of the expanded foveation window (±0.5 degrees by ±10 degrees/sec). BE, both eyes; R(L)EH, right (left) eye horizontal; R(L)ET, right (left) eye torsional; fs, foveating saccade; and bs, braking saccade.

components (especially the foveation periods) are phase-locked and the waveforms equivalent. The 1-degree peak-to-peak torsional component is 20% to 25% of the 4- to 5-degree peak-to-peak horizontal component.

Figure 4 shows the differences in congenital nystagmus waveforms of S4's horizontal and torsional components. In this subject, spontaneous reversals of the J_{ef} waveform's horizontal direction produced two different waveforms in the torsional plane, pseudocycloid and pendular with foveating saccades (P_{fs}). The "foveating saccades" of the torsional waveforms are saccades that are phase-locked with the actual foveating saccades in the horizontal (and vertical, if present) direction. The counterclockwise fast phases of the pseudocycloid corresponded to the JR_{ef} waveform and the clockwise foveating saccades of the P_{fs} corresponded to the JL_{ef} waveform. Despite the waveform differences, they were phase-locked in both planes; this allowed the foveation periods in both planes to overlap. The phase plane shows that both jerk directions resulted in satisfying the foveation-window criteria.

In Figure 5, both the measured torsional component of S9's congenital nystagmus and that predicted by Listing torsion are plotted for comparison. In both panels, it is clear that the Listing torsion is of much lower amplitude than that actually measured. In the lower panel, where the Listing torsion is increased by a factor of 10 to enable comparison,

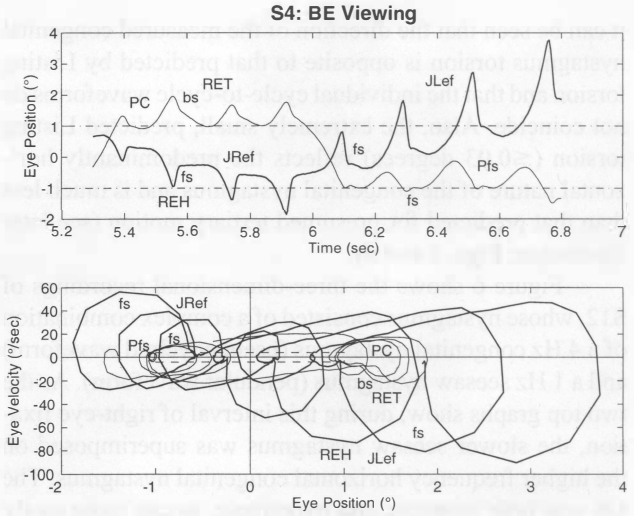

FIG. 4. Eye position (**top**) versus time and phase planes (**bottom**) of the horizontal and torsional components of S4's jerk right (JR_{ef}) and left (JL_{ef}) with extended foveation congenital nystagmus waveforms. Both torsional waveforms, pseudocycloid (PC) and pendular with foveating saccades (P_{fs}), differ from the horizontal waveform. *Dot-dashed lines* in the time plots indicate the extent of the ±1-degree extended foveation positions. *Dashed box* in the phase plane indicates the boundaries of the expanded foveation window (±1 degree by ±4 degrees/sec). BE, both eyes; R(L)EH, right (left) eye horizontal; R(L)ET, right (left) eye torsional; fs, foveating saccade; and bs, braking saccade.

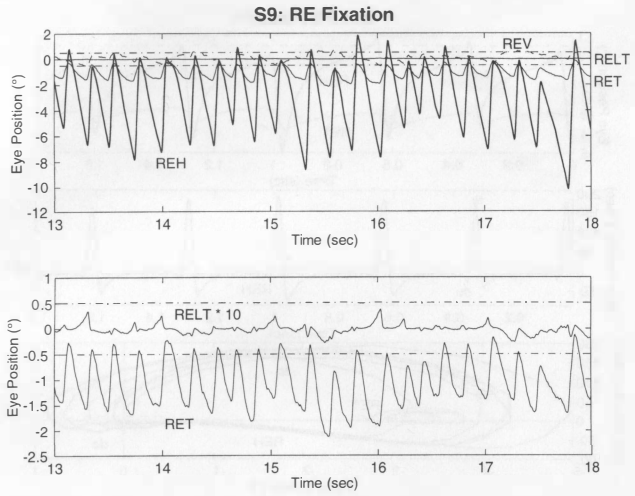

FIG. 5. Three-dimensional eye position versus time of S9's OD (**top**), including the Listing torsion component (RELT) calculated from the horizontal and vertical data using the equation given in the Methods section. A comparison of the Listing torsion (multiplied by a factor of 10) and the measured congenital nystagmus torsion (**bottom**). Horizontal, torsional, and Listing torsion shown in *solid*, *vertical*, and is shown *dashed lines*. *Dot-dashed lines* in the time plots indicate the extent of the fovea. BE, both eyes; R(L)EH, right (left) eye horizontal; R(L)ET, right (left) eye torsional; fs, foveating saccade; and bs, braking saccade.

it can be seen that the direction of the measured congenital nystagmus torsion is opposite to that predicted by Listing torsion and that the individual cycle-to-cycle waveforms do not coincide. Also, the extremely small, predicted Listing torsion (≤0.03 degrees) reflects the predominantly horizontal nature of the congenital nystagmus and is much less than that predicted for presumed tertiary motion (see later discussion; Figs. 8 and 9).

Figure 6 shows the three-dimensional recordings of S12, whose nystagmus consisted of a complex combination of a 4 Hz congenital nystagmus (pseudocycloid waveform) and a 1 Hz seesaw nystagmus (pendular waveform). As the two top graphs show, during this interval of right-eye fixation, the slower seesaw nystagmus was superimposed on the higher frequency horizontal congenital nystagmus. The OS was both esotropic and hypotropic, as can more easily be seen in the scanpaths at the lower left. The scanpaths (horizontal versus vertical motion) of both eyes also show that the slow vertical motion of the two eyes is superimposed on the rapid horizontal motion. In Figure 6 (lower right), conjugacy plots of the congenital nystagmus and the seesaw nystagmus in S12's two eyes (OD versus OS) demonstrate the horizontal and torsional conjugacy (positive-slope diagonal trajectories indicate 0-degree phase difference) and the vertical disconjugacy (negative-slope diagonal trajectories indicate 180-degree phase difference). The conjugacy plots also show the esotropic and hypotropic position of the OS and the fixation of the OD.

Target Foveation and Acuity

The NAFX values (8) for S1 through S6 (the patients with only congenital nystagmus and no symptoms) were calculated and compared with recalculations that included the retinal motion due to the actual, measured torsional component of the congenital nystagmus (*not* the predicted listing torsion). The original NAFX values for all waveforms exhibited by these six patients ranged from 0.804 to 0.065 (corresponding to predicted visual acuities ranging between 20/20- and 20/685-, respectively). When the retinal-image-motion effects of the torsional components were included, the recalculated NAFX values differed by percentages ranging from 0.002% to 0.098% (median, 0.015%), representing a median percentage change in predicted acuity of 0.02%.

Listing Torsion

We plotted the relative amounts of the torsional components of each subject's congenital nystagmus and compared them to that predicted by Listing torsion with the

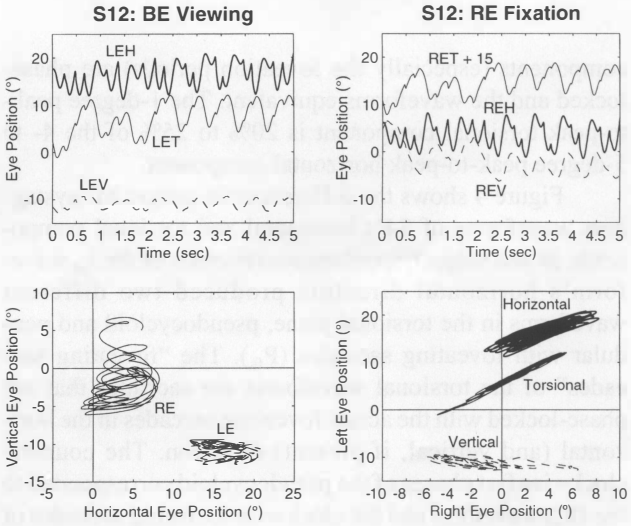

FIG. 6. Three-dimensional eye position versus time of S12's left (**top left**) and right (**top right**) eyes. Nystagmus scanpaths (**bottom left**) of S12's horizontal and vertical oscillations caused by the combination of congenital nystagmus and seesaw nystagmus showing the diagonal nature of the resulting motion. In the conjugacy plots (**bottom right**), the relative phases of the motion of both eyes in each plane are shown. In the time and conjugacy plots, vertical motion is shown as a *dashed line* (as is the LE motion in the scanpath), and in the top right panel, RET was shifted 15 degrees clockwise for clarity. BE, both eyes; R(L)EH, right (left) eye horizontal; R(L)ET, right (left) eye torsional; fs, foveating saccade; and bs, braking saccade.

worst-case presumption that the congenital nystagmus was diagonal. From Figure 7, it can be seen that there is a wide variation in the relative percentages of the torsional components of the congenital nystagmus across subjects and across waveforms; the relative percentages are idiosyncratic and vary from 8.16% to 94.42% (median, 32.94%). In S4, who exhibited two different torsional waveforms (both of which differed from the horizontal), the two percentages were 32.74% (P_{fs}) and 61.01% (pseudocycloid). In S11, the three percentages were 18.03% (P_{fs}), 29.52% (jerk-pseudocycloid), and 55.08% (pseudocycloid). Both subjects showed large waveform-dependent differences.

In Figure 8, the measured congenital nystagmus and torsional amplitudes are plotted for each subject and, for comparison, the predicted, theoretical Listing torsion for tertiary motion. In all cases, the measured torsion exceeded the predicted Listing torsion. The amounts by which actual measured torsion exceeded Listing torsion are shown more clearly in Figure 9, where the percentages of torsional to horizontal amplitude are plotted against congenital nystagmus amplitudes for each subject and the comparative Listing percentages are also shown.

In S5, we measured the torsional components at four secondary (±15 degrees horizontal and ±15 degrees vertical) and four tertiary (±15 degrees horizontal and vertical) positions. The torsional components for all positions are shown in Table 2. Both the measured values and the percentages they represented were lower in three of the four secondary positions and in three of the four tertiary positions than in the primary/central position.

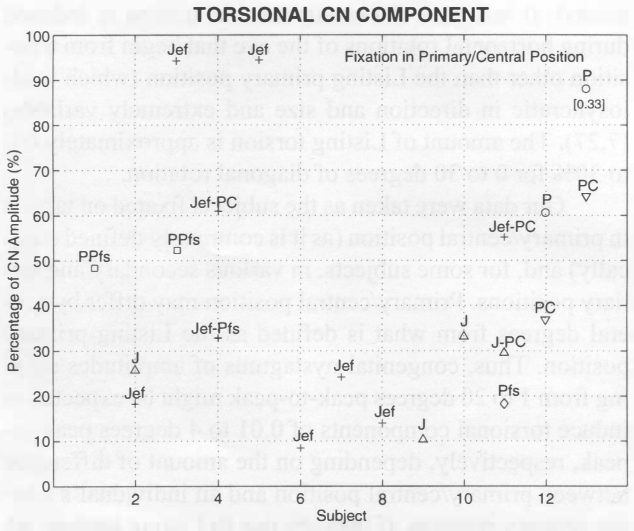

FIG. 7. A plot showing the average percentage of the total congenital nystagmus magnitude that the torsional component represents. Each subject's congenital nystagmus waveforms are shown and, for S4 and S11, the differing torsional waveforms are also indicated. In this and Figures 8 and 9, the symbols indicate the horizontal congenital nystagmus waveform: circle, pendular; diamond, pendular with foveating saccades; square, pseudopendular with foveating saccades; +, jerk with extended foveation; up-triangle, jerk; and down-triangle, pseudocycloid. As indicated in this Figure and Figure 9, the percentage of S13's seesaw nystagmus is plotted at 0.33 of its calculated value.

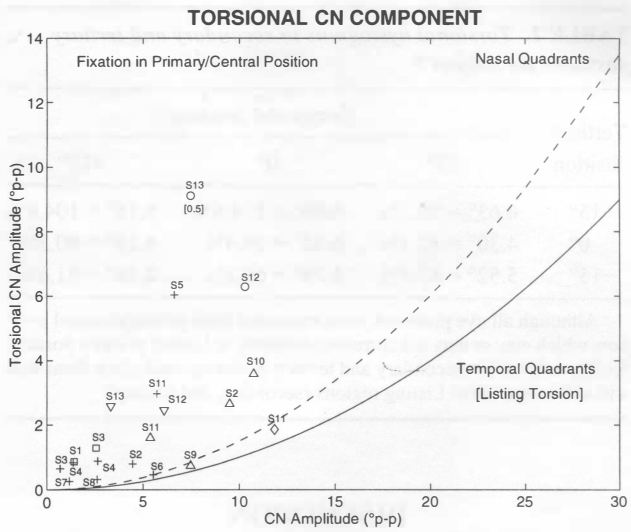

FIG. 8. The average absolute amplitudes of the torsional components are plotted versus the congenital nystagmus amplitude for each subject along with the predicted Listing torsion for tertiary motion. As indicated, the amplitude of S13's seesaw nystagmus is plotted at 0.5 of its measured value.

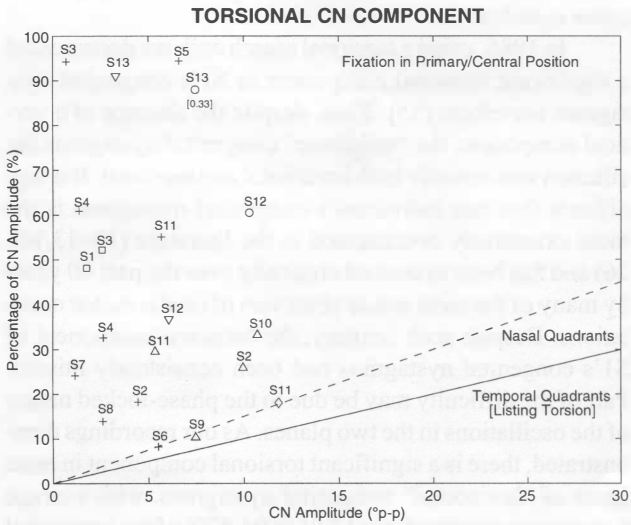

FIG. 9. The average percentages of congenital nystagmus amplitude of the torsional components plotted versus the congenital nystagmus amplitudes for each subject; the percentages of predicted Listing torsion for tertiary motion are also shown.

TABLE 2. ***Torsional nystagmus in secondary and tertiary positions for subject 5***

Vertical position	Horizontal position		
	−15°	0°	+15°
+15°	6.63° = 90.3%	6.06° = 114.6%	5.15° = 104.8%
0°	4.30° = 82.1%	6.02° = 94.4%	4.15° = 90.3%
−15°	5.52° = 87.4%	3.78° = 69.8%	2.76° = 91.6%

Although all eye positions were measured from primary/central position, which may or may not correspond exactly to Listing primary position for this subject, the secondary and tertiary positions used place them well within the equivalent Listing regions (secondary and tertiary).

DISCUSSION

Clinical Diagnosis

The existence of a torsional component to congenital nystagmus was originally suggested by an observation made by one of the authors of this study (LFD, who has congenital nystagmus). While he was sitting in a car and not attending to a visual task, the horizontal outline of the top of the dashboard (in his lower visual field) occasionally appeared to be rocking in a torsional manner. This torsional oscillopsia disappeared immediately when the subject (S1) fixated any object in front of him. Studies of S1's congenital nystagmus disclosed that there was no vertical component, and the congenital nystagmus was considered to be a uniplanar, "horizontal" oscillation. However, the occasional torsional oscillopsia suggested that, at least under some conditions, a torsional component might become manifest.

In 1986, using a torsional search coil, we documented a significant torsional component in S1's congenital nystagmus waveform (15). Thus, despite the absence of a vertical component, the "uniplanar" congenital nystagmus oscillation was actually both horizontal and torsional. It is significant that this individual's congenital nystagmus is the most extensively documented in the literature (10–13,16–26) and has been examined clinically over the past 40 years by many of the most astute observers of ocular motor oscillations. Despite such scrutiny, the torsional component of S1's congenital nystagmus had been consistently missed. Part of the difficulty may be due to the phase-locked nature of the oscillations in the two planes. As our recordings demonstrated, there is a significant torsional component in most cases of "horizontal" congenital nystagmus, with average percentages ranging from 13.91 to 94.42% of the congenital nystagmus amplitude. Thus, although pure horizontal congenital nystagmus may exist (as, perhaps, in S6), it appears to be much less common than horizontal-torsional congenital nystagmus.

Visual Acuity

We compared the best-corrected visual acuities of S1 through S6 predicted by NAFX values calculated using only the horizontal component of the congenital nystagmus with those calculated after inclusion of the torsional component. Our data confirmed the hypothesis that the predicted visual acuity of individuals with well-developed foveation is only minimally affected by torsional components of the congenital nystagmus (S2's percent NAFX change of 0.002% is equivalent to a 0.003% change in predicted visual acuity). The median change in the NAFX values of 0.015% corresponds to a change in predicted visual acuity that is well below the accuracy with which acuity can be measured. Thus, even for patients with significant torsional components of congenital nystagmus and lacking well-developed foveation, predicted visual acuity was virtually unaffected.

Torsional Mechanism

There are several possible mechanisms that might be responsible for the torsional component of "horizontal" congenital nystagmus. The two most probable are (1) a reflection of the Listing torsion seen in all tertiary positions of gaze, and (2) a centrally generated instability in the torsional control system (sites mediating torsional eye movements). It has been demonstrated that torsion is induced during horizontal rotations of the eye that begin from a position other than the Listing primary position (which is idiosyncratic in direction and size and extremely variable) (7,27). The amount of Listing torsion is approximately 0% to 30% for 0 to 30 degrees of diagonal rotation.

Our data were taken as the subjects fixated on targets in primary/central position (as it is commonly defined clinically) and, for some subjects, in various secondary and tertiary positions. Primary/central position may differ by several degrees from what is defined as the Listing primary position. Thus, congenital nystagmus of amplitudes ranging from 1 to 20 degrees peak-to-peak might be expected to induce torsional components of 0.01 to 4 degrees peak-to-peak, respectively, depending on the amount of difference between primary/central position and an individual's Listing primary position. If they are due to Listing torsion, we should expect that for large vertical gaze angles, the torsional components for the same horizontal congenital nystagmus should be larger than those in primary/central position, the latter being closer to the Listing primary position.

We found that the average torsional components of congenital nystagmus measured in primary/central position fell in the range of 8.16% to 94.42%. In fact, for S5, individual percentages greater than 100% (*e.g.*, 125.1%) were measured. These average percentages, which varied from 13.91% to 94.42% in 11 of 13 subjects, were far greater

than those predicted by Listing torsion for the respective congenital nystagmus motions (0.69–11.83%; median, 4.91%). Even in the close cases of S6, S9, and for one of S11's three waveforms, all values were greater than the measured Listing torsion for temporal-quadrant motion (equal to the predicted theoretical Listing torsion) and equal to that of measured nasal-quadrant motion. Because the congenital nystagmus motion for S9 was temporal (Table 1) and the measured congenital nystagmus torsion far exceeded the actual predicted Listing torsion (Fig. 5), only the torsion measured for S6 and one of three waveforms of S11 could be caused solely by Listing torsion in this worst-case calculation. However, the measured congenital nystagmus torsion for S6 also far exceeded (by a factor of 21.5) the predicted Listing torsion (based on the actual horizontal and vertical components). As in the data of Figure 7, waveform did not determine percentage differences between measured and predicted torsion across subjects. Owing to their magnitudes, the torsional components of S13's seesaw nystagmus were plotted at 0.33 (Figs. 7 and 9) or 0.5 (Fig. 8) of their actual values (264.02% and 18.26 degrees, respectively).

Furthermore, the data in Table 2 show that the torsional components of the congenital nystagmus (in patient S5) measured at secondary and tertiary gaze angles were not greater than those measured in primary/central position; in fact, in six of the eight positions, they were lower. A simple off set bias of the true Listing primary position would not yield such a map of torsional amplitudes. These data reflect the dependence of congenital nystagmus amplitude on eye position (nystagmus greater in upward or rightward gaze). They do not support the hypothesis that the torsional components of congenital nystagmus are due to Listing torsion. Finally, in S4 and S11, the torsional components of the congenital nystagmus had different waveforms than the horizontal components. This suggests independent instabilities and does not support either the Listing-torsion hypothesis or a simple mechanical crosstalk in the extraocular plant.

We also considered the possibility that our measure of primary/central position differed from Listing primary position by several degrees. The latter position is known to be variable, and its mean values have been found to be 6.9 degrees in the horizontal plane and 0.7 degrees in the vertical plane (7). Given the idiosyncratic nature of Listing primary position, one should expect that the individual variations of our subjects would be randomly distributed in both amplitudes and directions; they were not. Using the data of S9 in Figure 5 as an example, the torsional congenital nystagmus component exceeded by a factor of 37.2 that predicted by Listing torsion and it was in the wrong direction. Recalculating the Listing torsion for presumed ±6.9-degree errors in the position of either horizontal or vertical Listing primary position yielded amplitude difference factors ranging from 2.8 to 62.5, with the direction remaining incorrect in three of the four comparisons. Again, using our worst-case approach and shifting the points for all subjects rightward by 6.9 degrees in Figure 9 (*i.e.*, toward the Listing torsion curve), only the data from two additional subjects would come close to Listing torsion (for nasal quadrants only), S2 and S8. The data from S2 and S8 would still remain greater than Listing torsion, and the data from the remaining eight subjects would continue to far exceed Listing torsion. Even for this worst-case presumption of mislocalization of Listing primary position, 10 of the 13 subjects' torsion could not be attributed to Listing torsion (S4 and S11 had different waveforms, S9's torsion was in the wrong direction, and S5's torsion did not vary as Listing torsion in different gaze positions).

Taken together, the different horizontal and torsional waveforms that could simultaneously be exhibited in some patients, the higher torsional amplitudes in primary versus tertiary positions, the absence of a difference between predominantly horizontal and diagonal congenital nystagmus, the instances in which the amplitudes of the torsional components exceeded those of the horizontal components, and the higher than predicted percentages represented by the torsional components support the hypothesis that, in many cases, torsional congenital nystagmus is caused by instability of torsional control. Further study of this suggestion, including the identification of Listing primary position in congenital nystagmus subjects using search-coil systems that use three separate field coils, is warranted.

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