Head Shaking and Vestibulo-ocular Reflex in Congenital Nystagmus

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The authors investigated the mechanisms underlying the head shaking shown by some patients with congenital nystagmus (CN). In order to improve visual function by head shaking, a patient with CN must have (1) some visual acuity loss due to retinal image motion created by the nystagmus; (2) an abnormal vestibulo-ocular reflex (VOR); and (3) the head shaking must be correlated with the nystagmus. The authors measured the VOR gain (eye velocity/head velocity) and examined eye–head coordination in five patients with CN with various combinations of these three factors. One patient met all three criteria and was able to increase his acuity by shaking his head. Other patients who shook their heads either had no loss of visual acuity due to the nystagmus or had a normal VOR. In either case, head shaking was of no apparent visual benefit and may represent, instead, an associated pathologic tremor in the cephalomotor control system.

Some patients with congenital nystagmus (CN) also have head shaking that is usually intermittent and may have a frequency similar to that of their nystagmus. The etiology of the head shaking is unclear, but a few of these patients report improved vision with the head shaking. Gresty and Halmagyi1 have suggested that in some patients the head shaking reflects an adaptive strategy to nullify nystagmus, while in other patients it may only be an associated abnormal oscillation in the cephalomotor control mechanism. Vestibular function, however, has not been quantified in such patients to prove or disprove these hypotheses.

During head shaking in a normal individual, the vestibulo-ocular reflex (VOR) will create an eye movement in the orbit (the slow-phase) in the direction opposite to the head turning. The angular position of the eye in space is called gaze, and is determined by adding the movement of the eye in the orbit to the movement of the head. If the velocities of eye and head movement are the same but are oppositely directed, corresponding to a VOR gain (eye velocity/head velocity) of 1.0, the angular position of the eye in space (gaze) and, consequently, the direction of the visual axis in space does not change. This permits good visual acuity while one is moving about and can easily be demonstrated by reading an eye chart while shaking one’s head. While the cervico-ocular reflex is also stimulated during head shaking, it normally makes only a small contribution to compensatory slow-phases at the frequencies of rotation that comprise natural head motion.2–4

The decrease in visual acuity of many patients with CN may be due to retinal slip (movement of images on the retina) caused by their nystagmus,5 while other patients have associated abnormalities in their visual pathways that account for most of their visual loss. In some patients, the eye is still during a part of the nystagmus cycle and it is these periods of gaze stability that permit “foveation” and allow normal visual acuity.6 Patients with CN may also have an eye position (null point) in which the nystagmus amplitude is decreased and visual acuity is improved.

Several authors7–9 have reported an increase in visual acuity during head shaking in patients with CN, but vestibular function was not quantified. In order for a patient to be able to increase his visual acuity by shaking his head, several criteria must be met. At least some of the loss of acuity must be due to retinal slip caused by the nystagmus itself. The patient must also have a lowered or elevated VOR gain, at the frequency of head shaking, since with a normal VOR (gain = 1.0), head shaking per se does not allow a change in gaze which would be necessary.
to counteract the nystagmus. Finally, the head shaking must be coordinated with the nystagmus. If the VOR gain is low, the head movement must be opposite to the direction of the nystagmus drift; if it is high, the head movement must be in the same direction as the nystagmus drift. The most useful pattern of head motion is one in which the frequencies of head and eye oscillation are the same. If these conditions are met, a patient may be able to use head shaking to improve gaze stability during a part of each cycle of his nystagmus waveform, and thereby increase his visual acuity. An alternative possibility, reported in patients with nystagmus due to spasmus nutans, is that the head shaking transiently eliminates the nystagmus by an indirect, central, inhibitory effect. In this case, the VOR must have a normal gain, or the head shaking alone would create retinal slip and decrease acuity.

Several investigators have recorded vestibular responses from patients with CN. Forssman used caloric stimuli and found 44% of 88 patients had no response, and Yamazaki reported a unilateral decrease to caloric stimuli in three of five patients. When whole-body sinusoidal rotations are used to quantify the VOR, the frequency of stimulation is very important, as a gain of near 1.0 is normal at high frequencies with the gain progressively decreasing for values below about 1 Hz. Demer and Zee used a range of sinusoidal frequencies to test albino patients with CN and they found a loss of the low frequency response. Yee and colleagues tested 41 CN patients with 0.05 Hz rotations, where the normal gain was about 0.5, and found greater variation in the patient responses, although the mean was not decreased. None of these studies reported visual function or the presence of head shaking.

We have recorded both eye and head movements in five patients with congenital nystagmus in order to correlate their vestibular function, head shaking, and visual acuity. We selected these five patients because each one has a different combination of the three features. The VOR gain in both horizontal and vertical directions was measured, and the interaction of head shaking and nystagmus was examined during a reading task. Eye–head coordination during attempted gaze changes was also evaluated to look for VOR gain changes or any strategies compensatory for decreased vestibular function.

**Materials and Methods**

Five adult men with congenital nystagmus were studied. Their ages ranged from 30 to 60 yr. Patient 5 had ocular albinism; the others had no additional health problems. Ophthalmologic and neurologic examinations did not reveal any abnormalities except for the nystagmus and albinism. The nystagmus was conjugate in each patient. None of the patients had latent or manifest latent nystagmus. The direction, amplitude, frequency, and waveform of nystagmus was independent of whether either eye alone or both eyes together were viewing. Convergence may alter the waveform or amplitude of nystagmus in some patients with congenital nystagmus, but none of our patients had a change in acuity between near and distant testing. We eliminated any effects of changing convergence by making all recordings and reported acuity measurements at a viewing distance of 57 cm, so the state of convergence was always the same. The patients viewed a Snellen chart with the eyes in the orbital position that allowed best visual acuity. This orbital position corresponded to a null point in patients 1 and 3. For each patient, visual acuity was the same for each eye. Two of the patients (2 and 4) had themselves noted that they shook their heads, particularly when reading. The patient with albinism (5) had intermittent head shaking, but it did not seem to be associated with visual tasks. The patients were all volunteers in a congenital nystagmus study, and informed consent was obtained. One normal subject was tested in the same manner.

Eye movements were recorded using the magnetic field–search coil technique. This method utilizes a thin, soft plastic annulus in which a coil of wire is wound. After instillation of a topical anesthetic, the annulus is placed on the conjunctiva at the corneal–scleral junction so that it encircles but does not touch the cornea. The recording sessions were limited to 1 hr, and several patients were recorded on multiple occasions. Recordings were made monocularly with the search coil on the viewing eye. The patients wore a lightweight helmet that had a search coil attached to the top. This helmet allowed for unrestricted head movements in all directions. A 6-ft field coil system was used (CNC Engineering; Seattle, WA), allowing a patient to sit inside the coil and turn his head freely with minimal degradation of the system’s accuracy. The head and eye coils were calibrated before each use. The head and eyes were kept within 20 deg of straight ahead, where the system’s linearity is 2% or better. The analog eye and head position signals were low-pass filtered at 160 Hz (−3 db), and then were digitized at 100 Hz. The entire system had a noise level of 6 min of arc. Velocity traces were generated off-line using a finite impulse response digital filter with linear phase and a 0–10 Hz pass band. The patient was seated in a chair that allowed complete freedom of head movement. When immobilization of the head was desired, a chin rest was used, and the head movements were continually...
monitored to ensure that the head was still. The subjects were kept alert with verbal encouragement. Visual stimuli were under online computer control and were rear-projected onto a tangent screen 57 cm in front of the subject. The visual target was a 0.5-deg diameter spot of light. Each patient's nystagmus waveform was recorded with the head immobilized during visual fixation and in complete darkness.

Recordings were made while the patient voluntarily shook his head in the horizontal and then the vertical direction. Patients were verbally guided to achieve sinusoidal-appearing head oscillations with a range of frequencies from approximately 0.5 to 2.5 Hz. The peak to peak amplitudes varied from 5 to 40 deg, with the smaller amplitudes occurring at the higher frequencies. The peak velocities were 100 to 150 deg/sec. The patients were in complete darkness and were instructed to fixate an imaginary spot stationary in front of them. The relatively high frequency of rotation and the instructions to imagine a stationary spot comparable to that during normal head shaking. Passive head movements of similar frequency and amplitude were produced by firmly grasping the patient's head and turning it in the horizontal and vertical directions. Recordings were also made while the patient viewed newspaper-sized text that was held against the tangent screen, as this most effectively brought out the natural head shaking.

Eye–head coordination was tested using a saccadic refixation task in which a spot of light on the screen was unpredictably displaced either horizontally or vertically. Each patient was first recorded with his head fixed in the chin rest, and then again with his head free and instructed to follow the target with both the head and eyes.

The search coil method records angular position of the coil (eye or head) with respect to the (stationary) field coils, so the head coil trace was subtracted from the eye coil trace to give angular eye position in the orbit. The angular position of the eye in the orbit will be referred to as “eye;” and the angular position of the eye in space, as “gaze.” The numerical values for analysis were selected from a computer graphics terminal display of the position and velocity traces of the head and eye. The gain of the VOR was determined by comparing the velocities of the head and eye at the point of peak head velocity in each half cycle of head oscillation. The position of the eye in the orbit was near primary when these measurements were made. The eye and head movements are normally in opposite directions, so the ratio of eye to head velocity was by convention multiplied by -1.0 to create a positive value for the VOR gain. If a saccade appeared in the eye trace at the point of peak head velocity, that half cycle was not used for gain calculation. The waveforms of head movement were approximately sinusoidal, and no discernible phase shifts were expected or seen at the frequencies analyzed.

Measuring the VOR gain in patients with ongoing nystagmus is difficult because the measured eye velocity is the sum of the vestibularly-generated eye velocity and the eye velocity from the underlying CN. We measured the VOR gain during successive leftward and rightward half cycles of head movement. In patients 1, 3, and 4, the slow-phase was always in the same direction during our recording; so for half of the measurements, the slow-phase velocity from the CN was added to the vestibular eye velocity, and for the other half of the measurements, the CN was subtracted from it. In the other two patients, the slow-phases of nystagmus were not always in the same direction, but the direction of the slow-phase was not correlated with the direction of the head movement. Measurements made during passive head movements insured that no correlation was present. Therefore, for these patients too, the slow-phase velocities due to the CN were added to the vestibular response about half the time and subtracted from it about half of the time. Using this method, the underlying CN velocities are averaged out and do not affect the value of the mean VOR gain. The individual gain measurements, however, do reflect this interaction, and affect the variability of the gain, as seen in the standard deviations listed in Table 1. The larger standard deviations mostly occur when the slow-phase velocity of the underlying CN is high. While we measured the gain of the VOR when head velocity was maximum to increase the signal to noise ratio, the gain nevertheless remained constant throughout the full cycle of head movement, demonstrated in Figures 2 and 3. For example, measurements of the gain of the VOR taken from 10 equally spaced points covering a single cycle of head movement of patient 4 were all within 0.13 of the mean value, confirming that the gain of the VOR is independent of head position and velocity. This method of measuring the
Fig. 1. Representative waveform of each of five patients with congenital nystagmus. Each trace was recorded during fixation in the primary position. Patients 1 and 3 had a jerk left waveform and both also had a null point when looking to the right. Patient 4 had a jerk right waveform but there was little orbital position dependency. Patients 2 and 5 had more complicated waveforms, being sometimes pseudocycloid and sometimes pendular.

The gain of the VOR is valid for patients with nystagmus of any origin, and of any waveform.

Results

The gain of the VOR in the horizontal and vertical direction is given for each subject in Table 1. There was no systematic effect of frequency or amplitude on the VOR gain in the ranges tested (0.5–2.5 Hz, 5–40 deg). The gains determined for the passive and active conditions were also similar, so all conditions and frequencies were included in the computation of mean VOR gain. The statistical significance of the results was determined by using a two-tailed pooled t-test on the specified pairs of means. The horizontal gain for the normal subject was 0.97, which agrees well with the results of a study by Collewijn and colleagues in which similar active head oscillation in the dark at 0.3–1.3 Hz yielded a gain of 0.96 ± 0.04 for five normal subjects. The normal subject had a vertical VOR gain (0.98) that was essentially the same as his horizontal gain. Patients 1 and 2 had VOR gains in both the horizontal and vertical directions that were not significantly different from the normal subject, nor was the horizontal gain significantly different from the vertical.

Our three other patients (3, 4, 5) had horizontal VOR gains that were significantly below the normal ($P < 0.001$). In these three patients, the vertical VOR gain, though lower than normal, was significantly greater than the horizontal gain in each case ($P < 0.001$). The low values were recorded in spite of the fact that the verbal instructions and any possible augmentation by the cervico-ocular reflex should have led to an increased gain. Even with a visible fixation target, the horizontal VOR gains remained low, so that saccades were necessary to maintain fixation of the target.

A representative waveform of nystagmus that was recorded during fixation in primary position is shown for each patient in Figure 1. The frequency and

A. Horizontal pt #2

B. Vertical pt #2

Fig. 2. Traces recorded from patient 2 (normal VOR gain) during (A) horizontal and (B) vertical head rotations in the dark. Eye: eye position in the orbit; head: head position in space; gaze: eye position in space. The VOR compensates for the head movement, so the gaze traces are relatively stable during head rotation. The underlying congenital nystagmus is seen in the gaze trace, but is less intense than that observed during fixation in the light (compare to Fig. 1, noting different scales).
amplitude, but not the waveform, of each patient's nystagmus varied spontaneously and with the specific visual task. The gaze trace is equal to the sum of the eye trace and the head trace, so the gaze trace and eye trace are identical if there is no head motion. These waveforms were recorded with the head still, so only the gaze trace is shown. Patients 1 and 3 had jerk left waveforms in primary position, and each had a null point in right gaze where visual acuity was 20/20. Patient 4 also had a jerk waveform but no null position; with his head still, his best visual acuity in any orbital position was 20/60. The waveform of patient 2 was a pendular type, with refixating saccades occurring during fixation attempts. This patient's best acuity was 20/30. Patient 5, with albinism, had a visual acuity that was never better than 20/60. His waveform was variable, with periods of both pseudocycloid and pendular waveforms.

Figure 2 illustrates the effect of head rotations on the eye movements of patient 2, who had a nearly normal VOR in both horizontal and vertical directions. For horizontal rotation, the eye trace demonstrates the central addition of his underlying nystagmus and the compensatory vestibular eye movements. Since the VOR gain is 0.9, the vestibularly induced eye movement cancels 90% of the effect of the head movement, so that the gaze trace consists of the underlying nystagmus and about 10% of the head motion. In contrast, Figure 3 shows records from patient 4, who had a low horizontal gain but a nearly normal vertical gain. For vertical rotation, the vertical eye trace shows compensatory eye movements; while for horizontal rotation, the horizontal eye trace shows little compensatory movement. The eyes, therefore, move in space with the head so that the gaze trace is similar to the head trace. This demonstrates the instability of gaze during horizontal head rotations.

During attempted changes in gaze with coordinated head and eye movements, the horizontal gain of the VOR in these two patients had the same value as during head shaking with attempted fixation. A normal pattern of a shift of gaze using both eye and head movements is illustrated in the traces of patient 2 shown in Figure 4. A saccadic eye movement occurs first, acquiring the target. The head movement that follows is compensated for by the VOR so that the eye moves back toward a central position in the orbit, while the new position of gaze is maintained. On the other hand, patient 4, with a low horizontal VOR gain, cannot maintain gaze stability during gaze changes associated with head movements. Instead, he appears to have adapted for his low VOR gain by altering the normal pattern of eye–head coordination in a way similar to patients who have lost all labyrinthine function. His initial saccade is much smaller than the desired gaze change, and it is immediately followed by a backward drift of the eye that is not a direct vestibular response, since it begins before the head starts to move. During the head movement, the eye rotates little in the orbit due to the low gain of the VOR, so the gaze is moved with the head to the target.

The effect of head shaking while reading is shown for these same two patients in Figure 5. When reading, patient 2 does shake his head at a high frequency, but his normal VOR creates an additional slow-phase eye movement that effectively cancels the head movement. Therefore, he is not able to counteract the spontaneous nystagmus by head movements, and the resulting gaze trace reflects the (unchanged) underlying nystagmus waveform. Accordingly, the visual acuity of this patient is not improved by head shaking. In contrast, patient 4, who also shakes his head in the horizontal direction while reading, can, because of his low horizontal VOR gain, use head movement to change gaze and thereby...
nullify some of the unwanted gaze change created by his spontaneous nystagmus. The head shaking is at the same frequency, and oppositely directed to, the nystagmus slow-phase; the resulting gaze waveform has periods of complete stability that are not present when his head is still (compare Fig. 1 and Fig. 5). In this segment, the patient also utilized the strategy of a slight drift of his head to the right, which helped to compensate for his leftward nystagmus slow-phases. This patient could improve his visual acuity from 20/60 to 20/40 by shaking his head in this fashion.

Two other patients also had low VOR gains in the horizontal direction. Patient 3 had a little spontaneous head shaking and his visual acuity was 20/20 when his head was turned to bring his eyes to a null position. The patient with albinism (patient 5) could not increase his visual acuity by voluntary head shaking. He had spontaneous head shaking infrequently and did not have any while we were recording his eye and head movements.

**Discussion**

We chose a group of patients with CN who illustrate a variety of head shaking and nystagmus interactions. Three of the five patients had a low horizontal VOR gain, but only one of these three patients improved his acuity by appropriately shaking his head. One of the two patients with a normal VOR gain had head shaking in spite of the fact that it did not improve gaze stability or acuity. None of our patients appeared to turn off their nystagmus indirectly during head shaking, as has been reported in patients with spasmus nutans. If nystagmus were turned off, visual acuity could increase only if the patient had a normal VOR; therefore, improved vision during head shaking does not occur if the patient has a low VOR gain.

**Fig. 4.** Records of horizontal movement from patient 2 and from patient 4 during attempted gaze change. A, Patient 2 with a normal VOR has an initial saccade which rapidly shifts gaze, followed by a head movement and appropriate compensatory eye movement so that gaze remains stable. When the head is still, the nystagmus is present in both the eye and gaze trace. B, Patient 4 has a low horizontal VOR and used a different strategy to shift gaze. The initial saccade is much smaller than the desired gaze change, and the rest of the gaze shift is accomplished by the head turn while the eye hardly rotates backward in the orbit. Note the lack of gaze stability during the head movement.

**Fig. 5.** Horizontal records from the same two patients of Figure 4, taken while they were viewing a newspaper and spontaneously shaking their heads. A, Patient 2 with a normal VOR did not improve his fixational stability or change his nystagmus, which can still be seen in the gaze trace (compare to his fixation waveform in Figure 1). B, Patient 4, with a low horizontal VOR gain, could steady his fixation and improve his acuity by shaking his head at the right frequency and in the correct direction. Note the flat portions of the gaze trace (compare to his fixation waveform in Figure 1).
not necessarily indicate that the VOR gain is inappropriate. The clinical appearance of the head shaking of the two patients (Fig. 5) was similar, and it was not possible to be sure that the head movement was actually being used to compensate for the spontaneous nystagmus without analyzing the recordings. Because head shaking was found to be independent of an improvement in gaze stability, it is likely that it is due to pathologic mechanisms in some patients, and that it develops as a compensatory mechanism in others.

Patients with a normal visual acuity due to a null point (patients 1 and 3) or a waveform with foveation periods (patient 2) would not be expected to develop any compensatory head shaking, whether their VOR gain was low or normal. The patient with albinism (patient 5) has a reduced visual acuity that is probably primarily due to visual system abnormalities associated with the albinism. His pendular waveform presumably has a period of gaze stability associated with the direction reversal so that his nystagmus does not contribute much of his visual acuity loss of 20/60. Therefore, he would not be able to increase his visual acuity by shaking his head, even though his VOR gain is low.

One of the three patients with a low horizontal VOR gain was able to benefit from it by shaking his head to increase his visual acuity. During changes of gaze involving both head and eye movements, however, the low VOR gain works to his disadvantage, since during the head movement images would slip across the retina with consequent diminished visual acuity. None of the patients with a low horizontal VOR gain during head shaking showed any increase in gain in the combined eye–head refixation task. They were not able to voluntarily increase their gain even though, in this circumstance, it would have been useful to do so. This result did not confirm the suggestion of Gresty and Halmagyi that patients might learn to selectively suppress the VOR only during attempted fixation and not during refixations. In fact, our patients used alternative adaptive strategies to help stabilize gaze during eye–head movements, which is evidence of a persistently low VOR gain.

A possible mechanism for the development of a low horizontal VOR gain in congenital nystagmus is the loss of appropriate retinal slip (motion of image on the retina) that could be used to calibrate the reflex. The direction of the nystagmus in these patients was almost entirely horizontal, as is usual in congenital nystagmus. This creates a loss of gaze stability in the horizontal direction that is probably present at birth or shortly after. The presence of appropriate retinal slip during head movements seems to be necessary for development and calibration of the VOR. In CN, the retinal slip created by the nystagmus may interfere with this normal process of calibration. In humans blind from birth, the VOR is impaired and kittens reared in the dark have an abnormal VOR with a low gain. Experiments that manipulate the VOR gain by altering visual input have shown that the gain changes produced are specific to the direction of the visual manipulation. Eliminating retinal slip by strobe illumination has attenuated the plastic alteration of VOR gain and kittens reared in a strobe environment have a poor VOR. Patients with congenital nystagmus may have a nearly continuous horizontal retinal slip that is not a signal of an incorrect VOR gain, and cannot be used to improve the VOR accuracy. This loss of appropriate retinal slip in the horizontal direction may be the cause of the decreased VOR gain in the horizontal direction. The better vertical gaze stability would allow for more normal development of the vertical VOR, resulting in the difference in the vertical and horizontal VOR gains seen in our patients. This mechanism suggests that patients with normal visual acuity (due to periods when the eye is still) would be expected to have a normal VOR gain, as is the case for patients 1 and 2. The low horizontal VOR gain of patient 3, however, who has normal acuity at his null point is not explained by this mechanism. This patient also has a low vertical VOR gain, suggesting that there may be some other explanation for his abnormal VOR.

Some patients with CN may be able to use a low VOR gain and head shaking to improve gaze stability and visual acuity, while in other patients a low VOR gain or head shaking may be present without obvious benefit. A careful analysis of the vestibulo-ocular reflex and of the relationship between the head shaking and the nystagmus waveform is necessary to determine if the head shaking creates periods of retinal image stability.

Key words: vestibulo-ocular reflex, congenital nystagmus, head shaking, eye–head coordination, visual acuity

References