Relationships Between Visual Acuity and Anomalous Head Posture in Patients With Congenital Nystagmus

Deanna J. Stevens, MD; and Richard W. Herle, MD

ABSTRACT

Purpose: To determine whether patients with congenital nystagmus and an anomalous head posture have better binocular visual acuity than such patients without an anomalous head posture.

Patients and Methods: This was an observational case series of prospectively collected data for 125 patients with clinical and oculographically confirmed congenital nystagmus. Clinical data were tabulated using computer software. Statistical analyses compared binocular visual acuity with and without the presence of a clinically evident anomalous head posture.

The authors are from the Laboratory of Visual and Ocular Motor Physiology and the Departments of Ophthalmology, Columbus Children's Hospital and The Ohio State University, Columbus, Ohio.


Address reprint requests to Richard W. Herle, MD, Pediatric Ophthalmology Associates, Inc., Children's Hospital of Columbus, 555 S. 18th Street, Suite 402, Columbus, OH 43205.

Presented at the Annual Meeting of the American Association for Pediatric Ophthalmology and Strabismus; March 23-27, 2003; Kauai, Hawaii.

The authors have no industry relationships to disclose.

In accordance with ACCME policies, the audience is advised that this continuing medical education activity may contain references to unlabeled uses of FDA-approved products or to products not approved by the FDA for use in the United States. The faculty members have been made aware of their obligation to disclose such usage.

The material presented at or in any SLACK Incorporated continuing medical education activities does not necessarily reflect the views and opinions of SLACK Incorporated. Neither SLACK Incorporated nor the faculty endorse or recommend any techniques, commercial products, or manufacturers. The faculty/teachers may discuss the use of materials and/or products that have not yet been approved by the U.S. Food and Drug Administration. All readers and continuing education participants should verify all information before treating patients or utilizing any product.

EDUCATIONAL OBJECTIVES

1. To review the general characteristics of congenital nystagmus, how visual acuity is related to it, and why patients with congenital nystagmus assume anomalous head postures.

2. To determine the relationship between visual acuity and anomalous head posture in patients with congenital nystagmus with and without associated sensory system deficits.

3. To describe how the results of this study can assist with visual acuity prognosis in preverbal or nonverbal children diagnosed as having congenital nystagmus.

See quiz on page 297; no payment required.

Results: The mean visual acuity was 20/42 (log of the minimal angle of resolution [MAR], 0.32) in patients with an anomalous head posture and 20/83 (logMAR, 0.62) in patients with no anomalous head posture (P < .001). Among patients with disease of the sensory system, those with an anomalous head posture had a mean visual acuity of 20/55 (logMAR, 0.44) and those without an anomalous head posture had a mean visual acuity of 20/108 (logMAR, 0.73; P < .001).
Conclusions: Visual acuity was found to be significantly better in patients with congenital nystagmus who had an anomalous head posture versus those without such a head posture. Our findings indicate that the presence of an anomalous head posture in a patient with congenital nystagmus correlates with good vision and thus may be considered a positive prognostic sign in a preverbal child.


INTRODUCTION

Congenital nystagmus is an ocular motor disorder presenting at birth or during early infancy characterized by involuntary oscillations of the eyes. The involuntary eye movements have both a slow phase and a fast phase and are usually in the horizontal plane. Ocular motor recordings can be used to distinguish congenital nystagmus from other forms of nystagmus. Oculographically, congenital nystagmus is identified by slow phases of the waveforms with predominantly increasing velocity characteristics, foveation periods, and breaking saccades.1,2

The ocular oscillation interferes with vision in many ways, but primarily by preventing a visual target from remaining on the fovea. Congenital nystagmus is aggravated by anxiety, stress, anger, and increasing fixation demands. Congenital nystagmus is decreased with sedation, fatigue, sleep, extraocular muscle surgery, and some medications.3,7 Associated conditions may include strabismus, significant refractive error, and diseases of the sensory system such as retinal dystrophies, albinism, and congenital optic nerve abnormalities.3,7

Different positions of gaze can render different intensities of oscillation in a child with congenital nystagmus. The null zone is defined as that position of gaze where nystagmus intensity (amplitude × frequency) is least. Children with congenital nystagmus may adopt an anomalous head posture (eg, chin up or down, head turned or tilted) to improve their vision by maintaining gaze in the direction of the null zone.8,9 Vision may also be enhanced by increasing the foveation time (the period of time during which the eyes are within 30 minutes of arc of the target and moving at less than 4° per second) or the beat-to-beat stability of foveation (ie, “jitter” is reduced).10-12

By adopting an anomalous head posture and moving the null zone toward the position of primary gaze, a patient with congenital nystagmus is using a compensatory mechanism whereby nystagmus is stabilized and visual acuity is improved. Patients with congenital nystagmus may not exhibit an anomalous head posture because their null zone is located in the primary position, they may not have a null zone, or their sensory system deficit precludes any visual benefit from using a null zone. An anomalous head posture may be present in patients with congenital nystagmus for reasons other than nystagmus. Ocular conditions such as strabismus and uncorrected astigmatism, in addition to nonocular entities such as vertebral and muscular disorders, may also lead to a head turn or tilt.8,9,13

Ophthalmologists have the responsibility of being among the first to evaluate infants and children with involuntary eye movements. When faced with the diagnosis of congenital nystagmus, parents naturally experience some degree of anxiety concerning their child’s condition, with most of their questions revolving around the child’s visual prognosis. It is our hypothesis that the presence of an anomalous head posture might be indicative of better visual acuity among patients with congenital nystagmus. To date, no known study has evaluated the validity of this hypothesis. Our study aimed to demonstrate whether an anomalous head posture can be considered a predictor of visual acuity in patients with congenital nystagmus.

PATIENTS AND METHODS

This was an observational case series of prospectively collected data for 125 patients with clinical and oculographically confirmed congenital nystagmus. Of the 125 patients with congenital nystagmus evaluated in the Ocular Motor Neurophysiology Laboratory of the National Eye Institute during 1998 through 2001 (NEI Intramural Protocol 99-E1-0035), 64 children with congenital nystagmus and an anomalous head posture were included in this study. Their clinical and oculographic data were collected prospectively for this analysis.

To be eligible for this study, patients had to have been oculographically diagnosed as having congenital nystagmus and an anomalous head posture and had to have signed informed consent.
Inclusion criteria further included having best optical correction in place and the ability to cooperate for standardized monocular and binocular visual acuity testing as well as a complete ophthalmic evaluation. Those patients with congenital nystagmus evaluated at the National Eye Institute who were unable to cooperate with visual acuity testing were excluded from the study.

The etiology of the anomalous head posture was determined by clinical examination plus information obtained by ocuography. Each examination included assessment of monocular and binocular visual acuity using the Amblyopia Treatment Study Visual Acuity Testing HOTV optotype protocol or the Early Treatment Diabetic Retinopathy Study chart. The following parameters were also evaluated: spontaneous anomalous head posture, anomalous head posture during visual acuity testing, or both; intensity of nystagmus in relation to gaze, changing head position, and monocular cover; refraction; ocular motility; binocular function; results of dilated fundus examination; and ocular motor recordings. Each examination was performed in a standardized fashion by a qualified pediatric ophthalmologist (RWH). Patients were considered to have an anomalous head posture due to a null zone if they moved their chin up or down or turned or tilted their head during spontaneous activity or acuity testing and this was associated with a corresponding null zone on ocular motility recordings.

All data were tabulated using computer software. Patients who were unable to provide reliable data regarding visual acuity due to their young age or other deficiency and patients with mixed congenital and latent nystagmus were excluded from the study.

A statistical analysis was performed on clinical data from the 64 patients meeting the criteria for entry into the study. Patients were divided into two groups based on the presence or absence of a clinically significant anomalous head posture. The two groups were compared regarding age, gender, and the presence of strabismus. A primary analysis examined binocular visual acuity in relation to the presence or absence of an anomalous head posture. A secondary analysis compared visual acuity in those patients with and without associated disease of the sensory system. The statistical analysis involved use of a one-tailed t test.

**RESULTS**

Sixty-four patients were included in this study. Their ages ranged from 3 to 61 years, with an average age of 16 years. Seventy-six percent of the patients were male. Fifty-seven percent of the patients had coexisting strabismus.

Thirty-eight (59.4%) of the patients displayed an anomalous head posture, whereas 26 (40.6%) of the patients did not (*P* = .031). The average age of patients with an anomalous head posture was 17.6 years (standard deviation [SD], ±16.5 years) and the average age of patients without an anomalous head posture was 15.6 years (SD, ±13.2 years; *P* = .603). The male-to-female ratio of the 38 patients with an anomalous head posture was 30 to 8, whereas the ratio among the 26 patients without an anomalous head posture was 19 to 7 (*P* = .591). Twenty-three (60.5%) of the 38 patients with an anomalous head posture and 20 (76.9%) of the 26 patients without an anomalous head posture had coexisting strabismus (*P* = .152). Seven (18.4%) of the 38 patients with an anomalous head posture were noted during clinical and ocuographic evaluation to exhibit an aperiodic, changing head position. These patients were considered a subgroup with aperiodic alternating nystagmus and their visual acuity was analyzed separately. The mean visual acuity was calculated at 20/58 (log of the minimal angle of resolution [MAR], 0.46) for the 7 patients with aperiodic alternating nystagmus and 20/39 (logMAR, 0.29) for the remaining 31 patients with unchanging anomalous head posture (*P* = .145).

The patients with aperiodic alternating nystagmus with changing anomalous head posture were thereafter included in the larger group of all patients with anomalous head posture because of the statistically insignificant difference in visual acuity between the two groups.

The mean visual acuity was 20/42 (logMAR, 0.32) among all 38 patients with an anomalous head posture and 20/83 (logMAR, 0.62) among the 26 patients with no anomalous head posture (*P* < .001) (Fig. 1).

Thirty-one (48.4%) of the 64 patients in this study had one or multiple forms of associated visual sensory system disease identified. These conditions included albinism (*n* = 11), optic nerve dysplasia or hypoplasia (*n* = 17), foveal hypoplasia (*n* = 2), cone dystrophy (*n* = 3), congenital cataracts (*n* =
1), congenital stationary night blindness (n = 1), dyschromatopsia (n = 1), and aniridia (n = 1). Patients without a defined abnormality but with a visual acuity of less than 20/50 were also considered to have a disturbance of the sensory system, as the diagnosis of congenital nystagmus alone would not be enough to account for such decreased acuity.

Seventeen (44.7%) of 38 patients with an anomalous head posture and 20 (76.9%) of 26 patients without an anomalous head posture showed some evidence of sensory disease (P = .005). Among patients with sensory disease, those with an anomalous head posture had a mean visual acuity of 20/55 (logMAR, 0.44) and those without an anomalous head posture had a mean visual acuity of 20/108 (logMAR, 0.73; P < .001) (Fig. 1).

Twenty-seven patients showed no evidence of disease of the sensory system. Among these patients, mean visual acuity was 20/33 (logMAR, 0.22) in 21 patients with an anomalous head posture and 20/34 (logMAR, 0.23) in 6 patients without an anomalous head posture (P = .92) (Fig. 1).

Twelve (46%) of 26 patients without an anomalous head posture were found to have a clinical or oculographic null zone in primary position. These patients were analyzed as a separate subgroup because they had a primary position null zone and therefore theoretically needed no anomalous head posture. Their mean visual acuity was 20/69 (logMAR, 0.54), significantly less than the mean visual acuity of all patients with an anomalous head posture (20/42; logMAR, 0.32; P = .03) (Fig. 2).

**DISCUSSION**

Head posturing associated with abnormalities of the ocular motor system is usually compensatory. The purpose of this head posturing is to enhance bifoveal fixation, singular binocular vision, or visual acuity. The ocular etiologies of an anomalous head posture include strabismus, nystagmus, refractive errors, dissociated ocular deviations, and lid anomalies. Children with nystagmus will have an anomalous head posture due to a "gaze null" associated with congenital nystagmus or an "adduction null" associated with latent or manifest latent nystagmus, a head tilt associated with spasmus nutans, or improved fusion and ocular position associated with strabismus. Individuals with congenital nystagmus plus asymmetric aperiodic alternating nystagmus may also exhibit an anomalous head posture, sometimes in both directions. In a review of the ocular causes of abnormal head postures in 188 patients, Kushner listed 8 separate conditions. Ocular oscillations were responsible in 27.4% of the cases if the patients he described as having "congenital esotropia with ocular posture" are included. Dell'Osso found that 43% of his first 100 patients with congenital nystagmus or latent or manifest latent nystagmus had strabismus. Of the patients with strabismus, 53% had congenital nystagmus and 35% had latent or manifest latent nystagmus as their oscillation type. It has been reported in other series of patients that from 19% to 57% of those with strabismus will have associated nystagmus. Although the clinical differen-
tiation of these disorders is accurately accomplished after a thorough history and ocular examination, the etiology of an anomalous head posture is often elusive in patients with a combination of strabismus and nystagmus. A complete ophthalmic, ocular motor, and oculographic evaluation is the only sure way to differentiate the etiology of an anomalous head posture in many of these patients.

Previous studies have attempted to correlate visual acuity with measurable parameters in patients with congenital nystagmus. Guo et al. found a positive association between visual acuity and the duration of foveation periods in patients with congenital nystagmus. Abadi and Worfolk identified a significant correlation between visual acuity and wave form shape. Bedell and Loshin, however, failed to show a significant correlation between visual acuity and the amplitude, frequency, and intensity of nystagmus and the duration of foveation periods, although the intensity and amplitude of nystagmus were favorably associated with acuity in some subgroups. The current study is the first known to examine the relationships between visual acuity and the presence or absence of an anomalous head posture.

The goal of this study was to determine whether patients with congenital nystagmus and an anomalous head posture have better binocular visual acuity than such patients without an anomalous head posture. The presence of an anomalous head posture was independent of age, gender, and the presence or absence of strabismus. In this study, patients with an anomalous head posture were actually less likely to have coexistent strabismus compared with patients without an anomalous head posture.

Visual acuity was significantly better in those patients who displayed an anomalous head posture versus those who did not. This was true of all patients with an anomalous head posture as a group, including those patients with a periodic alternating nystagmus.

Patients without an anomalous head posture were significantly more likely to show evidence of an abnormality of the sensory system. Similarly, as a group, patients with disease of the sensory system were less likely to demonstrate an anomalous head posture. These findings indicate that the presence of an abnormality of the sensory system, with its inherent visual deficit, might reduce the stimulus for using the null zone because less visual benefit is derived. Importantly, among patients with an associated sensory disease, vision was shown to be significantly better in those who had an anomalous head posture.

Among patients without sensory disease, an analysis failed to show a statistically significant difference in visual acuity between those with and without an anomalous head posture. No difference was expected, as the group was selected by visual acuities better than 20/50. Nevertheless, the evidence holds that an anomalous head posture, regardless of the presence or absence of sensory disease, is predictive of a better visual acuity in a patient with congenital nystagmus.

Interestingly, patients with a primary position null zone, thought to use their null zone without an anomalous head posture, had decreased visual acuity when compared with patients with an anomalous head posture. By convention, a null zone at primary position is located within 10° vertically or horizontally. A patient with an apparent primary position null zone could have a null zone that is just outside of primary position, but no anomalous head posture is manifest because the patient does not use the null zone and therefore gains no visual benefit from it. Further oculographic study may better determine which patients with null zones fail to use them and why.

Our findings indicate that the presence of an anomalous head posture in a patient with congenital nystagmus correlates with good vision. In a preverbal child, this may be considered a positive prognostic sign, thus providing new information to the patient's family and practitioner.

REFERENCES

9. Herltse RW, Zhu X. Oculographic and clinical characterization of thirty-seven children with anomalous head postures, nystagmus,