

Article ▶ Low Vision Management For Infantile Nystagmus: A Case Study

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ABSTRACT

Background: Infantile nystagmus is a common cause of visual impairment. It can occur in isolation, but it is often found in association with other conditions, such as albinism. The condition results in varying levels of vision impairments ranging from mild to severe. Characteristics include early onset, bilateral involvement, pendular and jerk waveforms, the presence of a null point, and the lack of optokinetic nystagmus and oscillopsia (environment moving). This article provides a review of pathogenesis, epidemiology, and treatment of infantile nystagmus. A case study highlights low vision strategies and management for the condition.

Case Report: A 12-year-old male with infantile nystagmus presented to the satellite low vision pediatric clinic for initial evaluation. He reported difficulty with different tasks at school secondary to his visual impairment. Low vision intervention utilized various devices and strategies to meet his distance and near goals.

Discussion: With no known cure, low vision rehabilitation is important for individuals with nystagmus to allow them to reach their highest potential. Rehabilitation techniques can easily be applied by a primary care optometrist.

Keywords: low vision, nystagmus, reading acuity, rehabilitation, visual impairment



A video review of congenital nystagmus.

Introduction

Nystagmus is an involuntary, periodic eye movement caused by a slow drift fixation that may be followed by a fast (jerk form) or slow (pendular form) re-fixation saccade.¹ Congenital nystagmus is usually a benign condition that is associated with normal or reduced vision.

Although the underlying cause for nystagmus is present at birth, nystagmus tends not to arise until age two to four months when visual fixation typically develops.¹ When not present at birth, the more appropriate nomenclature for nystagmus that occurs before six months of age is infantile nystagmus. However, published articles frequently use many terms interchangeably for this condition including congenital nystagmus (CN), infantile nystagmus (IN), and infantile nystagmus syndrome (INS). Characteristics include early onset, bilateral involvement, pendular and jerk waveforms, the presence of a null point, and the lack of optokinetic nystagmus and oscillopsia.¹⁻⁵

IN can occur with and without additional ocular and/or central nervous system deficits. A recent extensive survey performed in the

United Kingdom found the total prevalence of IN to be 1.4 per 1,000.⁶ It is important to determine whether the nystagmus occurs in isolation or is associated with other conditions in order to give the proper prognosis. IN can be associated with an afferent visual pathway problem, a cerebral anomaly, or metabolic disease such as an optic nerve glioma, cerebellar lesion, or Tay-Sachs disease. However, IN can also occur in isolation. Nystagmus associated with ocular pathology such as aniridia is often termed sensory or secondary nystagmus, while nystagmus without association is termed motor or primary nystagmus.^{1,2} Idiopathic infantile nystagmus (IIN) is also another term commonly encountered for infantile nystagmus found in isolation.

Even though nystagmus can be associated with different conditions, Hertle and Dell'Osso strongly insist that these two scenarios should not be separated into different categories of nystagmus.⁷ They assert that this division, often used in the literature, is very misleading to the true etiology of the nystagmus. This is because IN occurring with or without additional anomalies demonstrates the same waveform patterns and is due to the same mechanism. Although the nuances of this mechanism have yet to be discovered, it is generally hypothesized that the nystagmus occurs because of a deficit in the ocular motor pathway. More specifically, the nystagmus results from instability of the slow eye movement and gaze-holding circuitries. This includes connections between the visual cortex, superior colliculus, pontine nuclei, cerebellum, and oculomotor nuclei. They contend that for a person with both a sensory anomaly such as albinism and congenital nystagmus, neither condition directly results from the other, i.e. the nystagmus is not technically the direct result of albinism, but rather a result of motor circuitry dysfunction. Thus, Hertle and Dell'Osso argue that each disorder, nystagmus and albinism,

should be defined as a distinct entity in order to avoid confusion.^{4,7}

However, they do note that the motor and sensory systems do not develop in isolation from one another, but instead there is much "cross-talk" between the two pathways during maturation. Collaboration between these two systems should result in overall smooth and coordinated function. This does not occur in IIN; instead the motor system develops erroneously. This ocular motor pathway malfunction can therefore result from a primary defect of this pathway (what others term motor/primary nystagmus) or may result from the effects of the abnormal cross talk between two pathways caused by the anomalous sensory system (what others term sensory/secondary nystagmus). Proponents of IN's dual classification system argue that although inherently inaccurate, this categorization is useful for clinical diagnosis, management, and explanation of prognosis.^{2,8}

Nystagmus associated with a sensory deficit demonstrates a significant reduction in foveation time and greater visual impairment as compared to IIN.⁹ Infantile nystagmus cases are often associated with conditions that disrupt the afferent pathway including albinism, optic nerve hypoplasia, and retinal dystrophies such as achromatopsia. The recent large scale UK survey found the greatest association of IN with albinism. Differentiation between the two etiologies may be made through direct observation such as in the case of albinism or optic nerve hypoplasia. However, if direct observation is inconclusive, ancillary testing including color vision, visual fields, electroretinography, magnetic resonance imaging, and/or visual evoked potential should be performed.²

IIN can occur sporadically but is most often hereditary. Inheritance is most commonly X-linked but can also be autosomal dominant or recessive; recessive pattern is least likely. A mutation of the FRMD7 gene has been discovered as the most frequent reason for

the X-linked form. FRMD7 is expressed in the cerebellum, developing neural retina, and lateral ventricles. However, it is unknown whether the nystagmus happens because of a loss or a gain of gene function. A small cohort study showed that people with the FRMD7 mutation were less likely to demonstrate an increase in amplitude of nystagmus on primary gaze and anomalous head posture. However, there is currently no definitive way to distinguish inheritance pattern from clinical observation.¹⁰

To date, no treatment has been found for IN, largely in part because of the complexity of etiology. Pharmacologic therapy has been successful in adult acquired nystagmus where pathophysiology is better understood. Gabapentin and Memantine have shown promise in minimizing nystagmus when occurring in isolation, but only small studies have been performed.^{3,11} Surgical intervention has consistently showed improvement of anomalous head posture but varied results in improvement of visual acuity.^{3,12} This is likely due to the many factors that influence visual acuity such as patient age and overall development of motor and sensory pathways. With no known cure, low vision rehabilitation is important for individuals with IN to allow them to reach their highest potential.

Case Report

History

A 12-year-old male presented to the satellite low vision pediatric clinic for an initial evaluation. He and his mother reported a reduction in distance and near acuity secondary to bilateral congenital nystagmus. Specifically, he reported difficulty seeing the board at school, noting that it was especially challenging when words were written in white chalk on the green chalkboard instead of a SMART Board. He and his mother also stated that he became tired after reading for roughly 20 minutes. His mother expressed concern

about his hunched over posture when reading as a result of his significantly reduced reading distance. Goals for this visit were to identify any devices or strategies that may help alleviate or eliminate these problems.

Aside from the nystagmus, his ocular and systemic histories were unremarkable. He was being followed regularly by an ophthalmologist for his condition and wore a pair of distance-only spectacles. He reported that his vision was stable. This was confirmed at his last visit (<6 months prior) with his ophthalmologist, during which his distance correction was only slightly adjusted.

Examination Findings

Entering distance acuity with habitual correction (OD: +0.50-1.50x002, OS: -0.25-1.75x005) was 20/60 monocularly and binocularly using a modified Feinbloom Chart (SVOSH Salus University, Philadelphia, PA). Entering near acuity with habitual correction was .25/1M binocularly (20/80 Snellen equivalent at 40 cm). A continuous text Lighthouse reading card (Lighthouse International, New York, NY) and an MNRead card (Precision Vision, LaSalle, IL) were alternated to prevent memorization of the text. It was noted that the patient's reading was slow with much effort during initial acuities. He was asked to re-read the card, this time largest print to smallest. The smallest print size at which the patient's reading was effortless and fluid was recorded as .45/2.5M (20/140 Snellen equivalent). It was also noted that the patient displayed a horizontal pendulum-like head movement while reading.

Updated refraction confirmed that the patient's habitual distance prescription was appropriate. A +2.00 DS add for both eyes was determined best to meet the patient's near needs; see discussion below. Acuities through the near prescription were .3/1.6M (20/110 Snellen equivalent) and .19/.8M (20/80 Snellen equivalent), all read with ease and fluidity.

Subjectively, the patient also stated a decrease in effort to see near material.

Entrance testing was unremarkable with the exception of nystagmus. In primary gaze, the nystagmus demonstrated a low frequency, moderate amplitude, horizontal pendular waveform; while in right and left gaze, the waveform was right jerk and left jerk, respectively. The amplitude and frequency of the nystagmus increased on left and right gaze and decreased in up and down gaze as well as upon convergence. There was not a strong latent component. Binocular color testing was normal with all Ishihara pseudoisochromatic plates identified correctly. Binocular contrast sensitivity was measured at 5% binocularly using the Lea Symbol contrast test, indicative of a moderate contrast impairment.

External and internal ocular health was unremarkable bilaterally; no pathology was noted. Dilation was deferred secondary to the patient's recent evaluation by his ophthalmologist and his desire to return to school after the low vision exam.

Discussion

Classification

The patient's IN can be classified as idiopathic since it was not associated with another deficiency. Direct observation showed no additional disorders such as albinism, aniridia, or optic nerve hypoplasia. Color vision was normal, and visual fields were full. The patient's nystagmus demonstrated the classic mixed waveform of IIN, horizontal pendular nystagmus in primary gaze that becomes a jerk wave in horizontal end gaze. Additionally, his mother reported that as an infant he had a very extensive nystagmus work up, and all tests showed no further involvement.

Low Vision Rehabilitation

First and foremost, an accurate refraction must be performed to ensure that the patient receives maximum optical clarity.¹³ It should

be noted that infantile nystagmus is found to be associated with a significant amount of with-the-rule astigmatism.^{13,14} This was true of the patient's prescription.

As mentioned above, people with nystagmus may also develop anomalous head posture.⁴ The compensatory head turn or tilt allows their eyes to be positioned at their null point.² Although our patient did not demonstrate a definitive head turn or tilt, he did display a pendular head movement while reading. This head movement was likely a counterbalance for the nystagmus.² When questioned about his head movement, the patient was unaware of the motion, indicating that he did so subconsciously. This is a positive adaptation to his condition, and therefore, no effort was made for correction. In order to help the patient meet his visual goals, his complaints were grouped into distance and near tasks.

For distance, a telescopic system would enable him to read material written on the board. A hand held telescope as opposed to a mounted system was evaluated since the inability to view the board was not a constant concern. The hand held device gave him the freedom to use the telescope when necessary both during school as well as during mobility-related activities. Additionally, it was less noticeable than a mounted system, allowing him to feel more comfortable when using the device in school. A Keplarian model as opposed to a Galilean was chosen due to its larger field of view and better optical quality. A variable focus system was used to allow the patient to focus the telescope for different distances: for instance, if he sat further or closer to the board or wanted to use the telescope outside the classroom.

Goal VA for viewing print on a classroom board is generally agreed upon as 20/40 to 20/50.¹³ Given the patient's best-corrected acuity, 20/60, he would require 1.5X magnification to achieve this. However, the lowest-powered hand held telescope available

during the evaluation was 2.75X. Although mathematically a 2.75X telescope would increase his acuity enough to view the board, it has been our experience that the lower magnification telescope's smaller size makes it more difficult to manipulate. This is especially true for the novice user. Since this was the patient's first experience with a telescope, it was important to reduce any potential frustration that would reduce the patient's motivation to use the device. Therefore, he was evaluated with a 4X telescope. In the future after he is comfortable with the telescope, a lower magnification device could be evaluated. The lower magnification may be preferred because of the enlarged field of view.

During the exam, the patient was very successful with a 4X Specwell Keplarian hand held telescope. Since his monocular spectacle-corrected acuity was the same, he looked through the telescope with his preferred eye. Had the acuities differed, he would have used the telescope with his better-seeing eye. He was able to achieve 20/20 acuity with his right eye. He also used the telescope to spot far-off objects through the window and was excited by his new ability to see distant targets.

In addition to difficulty with board work secondary to acuity constraints, the patient was educated that the lower contrast of the chalk on the green chalkboard was also contributing to his reading difficulties as compared to the higher contrast SMART board. It is well established that there is a range of contrast levels for which one can maintain maximum reading speed in both normally sighted and visually impaired individuals. However, visually impaired individuals are often much more sensitive to contrast changes and thus show a truncated range.^{15,16} This applies to the patient since he demonstrated a mild contrast impairment; he is more susceptible to changes in contrast such as using a SMART Board versus a chalkboard compared to his classmates. The role of contrast in his school work was noted in his

exam summary for his vision itinerant teacher. Additionally, he would also benefit from high contrast reading and writing materials. The use of bold line paper and high contrast pencils and markers was recommended.

For near, the additional plus prescribed should alleviate some strain on his accommodative system and thus prevent or delay visual fatigue. Alternation between the Lighthouse Near Acuity and MNRead continuous text cards was performed in order to avoid memorization. A concerted effort was made to evaluate both his near acuity (NA) and reading acuity (RA). Memorization can result in falsely elevated RA and NA. Reading acuity is defined in the literature as the smallest print size that can be read without significant errors.¹⁷ A formula incorporating sentences and words read along with errors on the MNRead card has been developed to determine RA accurately.¹⁷ However, this information was not collected during our exam secondary to time constraints, so the RA referred to in this paper was an estimate. The RA was determined by carefully listening to the patient while he read the near card aloud, largest print to smallest. The RA was the size at which he read with ease and efficiency, i.e. smoothly and quickly with little effort. Near acuity was the smallest print size he was able to read regardless of the errors.

Critical print size (CPS) is the font size below which reading is suboptimal. M notation continuous text cards were used to determine near acuity as it is well established that continuous text acuity is a better indicator of overall reading rate than single letter acuity.¹⁷⁻²⁰ However, some research shows that although there is a correlation between continuous text acuity and reading rates, continuous text acuity is still often not an accurate predictor of reading rates.¹⁸ This is most likely secondary to the multifactorial aspect of reading. It is expected that IN patients will have decreased reading rates compared to normally sighted

individuals because the involuntary nystagmus disrupts the sequence of saccades and fixations required for reading.¹⁷

There was a noticeable difference between RA and NA. Although the patient could decipher 1M print without any additional magnification, he did so taxingly; reading was fluid at 2.5M print. This difference supports the patient's initial complaint of visual fatigue since it emphasizes the large effort needed to complete near work. It is understandable that the patient would become tired after performing in this manner for long periods of time. The disparity is also supported by research studies that have shown an acuity reserve of 3:1 to be necessary for maximum reading rates.²¹ Acuity reserve is the ratio between goal acuity and NA. Applying this to the patient, starting with his NA of 1M, theoretically one could expect him to read fluidly at 3M given the 3:1 ratio. This assumption correlates well with his measured RA occurring at 2.5M. RA was used for further near calculations because we felt this rate more accurately portrayed the patient's near acuity and habitual effort.

Again using reading reserve to achieve maximum reading rates, starting with RA and CPS with habitual correction, .45/2.5M, the goal acuity would be about 0.8M with a 3:1 reading reserve. Based on relative size magnification, the patient would need to hold his material three times closer to achieve this print size. He would have to shorten the working distance from the initial 45cm to 15cm. At this working distance, he would need roughly an additional +6.75 DS to achieve best optical clarity.

However, although the patient's accommodative ability was reduced, he was still pre-presbyopic and therefore unlikely to accept this entire near prescription. Furthermore, another goal of the near magnification was to extend the patient's working distance; the full add would force him to hold material much closer to maintain best clarity. Instead, a +2.50 DS add was chosen as a starting point based on

previous experience with low vision children. With this correction, his CPS improved to 0.8M while maintaining a comfortable working distance of 20 cm. He was able to achieve 1.6M when asked to extend his working distance even further to 30 cm (about 12 inches). In the end, a +2.00 DS add was prescribed because his RA remained the same as with +2.50 DS, but his range of clarity increased with the power reduction.

A flat top bifocal prescription for classroom and long-term writing or reading was recommended. The bifocal is user friendly since the patient does not need to switch between glasses. This is very helpful in an educational setting when students are frequently switching between distance (board) and near (book) tasks. The bifocal location has an added benefit for this patient because the design causes him to look downward into a position where his nystagmus is dampened. Although the additional plus will also reduce convergence, another null position for the patient, the ratio between accommodative convergence and stimulus to accommodate (AC/A) was not measured, so the bifocal's full effect on eye posture is unknown. Additionally, it was indicated that the bifocal be measured slightly higher than normal, from the lower pupil margin, to guarantee utilization of the additional magnification. The patient was cautioned about wearing the bifocal full time as it may cause mobility issues.

The CPS with which the patient should be presented was determined using the equivalency 1M=8 point and M size for RA at maximum working distance of 30 cm. Given 1.6M as RA at 30 cm, the smallest font size at this distance would be 13 point. A range of 13- to 16-point font size was recommended because overall letter size and stroke width are widely variable given font type. The optimal font would be one with a uniform stroke width such as Tahoma or Arial. This will enable the patient to view his reading material at a more

comfortable 12- to 16-inch viewing distance. Additionally, good quality task lighting and the use of a slant board to improve posture were recommended.

Conclusion

Infantile nystagmus can be associated with or appear without other systemic and ocular complications. When first noted, much care should be taken to determine its etiology. Simple low vision concepts and strategies can be applied to meet visual needs. Patient goals should be broken down into distance and near tasks. Both optical and non-optical compensations should be considered to maximize habilitation. A primary care optometrist can easily use these low vision principles to provide great patient satisfaction. The information can also be used to modify occupational and educational settings to allow an individual with nystagmus to attain greatest success.

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