Title: Prism Management of Anomalous Head Posture (AHP) Secondary to Infantile Nystagmus

Abstract: Anomalous head postures (AHP) are often adopted by patients with nystagmus in order to maximize visual acuity. The following describes an infant with an AHP secondary to infantile nystagmus who is managed with base-out prism.

I. Case History

- Patient demographics: 1 year, 4 months old Caucasian male
- Chief complaint: Patient presents for a doctor-ordered nystagmus follow-up exam. At the previous exam, the patient was prescribed glasses with base-out prism (9 base-out in each eye) in order to reduce the AHP.
- Ocular history:
  - (+) Infantile nystagmus (first noticed at 6 weeks of age, diagnosed at 4 months of age)
  - (-) Ocular trauma
  - (-) Ocular albinism (confirmed by laboratory tests)
  - (+) Past exams at the University Eye Institute: originally presented on 11/2015; follow-up exams on 03/2016, 06/2016
- Medical history
  - No developmental concerns
  - Unremarkable pregnancy and birth
  - Born full-term at 38 weeks
  - No significant illnesses, injuries, hospitalizations or disabilities
  - Immunizations are current
- Medications
  - No reported systemic or ocular medications
  - No reported allergies to systemic or ocular medications
- Family medical and ocular history
  - (-) Nystagmus
  - (+) Diabetes Mellitus. Paternal grandmother
- Social history
  - Stays home with mom
  - Qualifies for Early Childhood Intervention Services (ECI)

II. Pertinent findings (the following includes findings from current & previous exams)

- Clinical
  - Unaided Acuity (with Teller acuity cards @ 55 cm):
    - OD: 20/190 (3.2 cyc/cm) OS: 20/94 (6.5 cyc/cm)
    - **Monocular visual acuities were measured at the initial exam, but were unable to be obtained at subsequent exams as patient gets significantly distracted and upset when either eye is covered. At follow-up exams, binocular visual acuity was 20/94 (6.4 cyc/cm), measured with Teller acuity cards
  - Cycloplegic retinoscopy: OD: +1.75-0.50X090
    - OS: +2.00-1.00X090
Confrontation fields: Monocular confrontation fields full and symmetric in all meridians, OD and OS

Alignment (measured with Krimsky):
- 15 CRXT with Krimsky (same with or without prism glasses)
- Current glasses prescription: OD: +0.75-0.50X090 9pd BO
  OS: +1.00-0.50X090 9pd BO

Pupils: equal, round and reactive to direct and consensual stimulation with no afferent pupillary defect detected OD, OS

EOMS: no underactions or overactions OD, OS
- Gross convergence was observed

Stereopsis measured with Lang I card: inconclusive as patient would not attend to card

Color Discrimination (measured with Pease-Allen color test):
- OD: 4/4 Red/green plate (pass)
- OS: 4/4 Blue/yellow plate (pass)

Contrast Sensitivity (measured with Hiding Heidi):
- Normal contrast sensitivity (measured with both eyes, 1.25%)

Slit lamp biomicroscopy:
- All structures normal OU
- (-) iris transillumination defects OU

Fundus exam:
- (+) Blonde fundus OU
- C/D ratio: 0.20/0.20 OD, OS

- Physical
  - Nystagmus: horizontal, left-beat nystagmus with moderate amplitude and low frequency
  - Head posture:
    - Without prism glasses: extreme right head turn with downward tilt (looking in superior left gaze)
    - With prism glasses: significant improvement in head posture; slight right head tilt
  - (-) Head nodding

III. Differential diagnosis
- Leading/Primary diagnosis
  - Idiopathic infantile nystagmus with AHP
- Others
  - Fusion maldevelopment syndrome nystagmus (latent/manifest latent nystagmus)
  - Spasmus nutans
    - Triad: nystagmus, head nodding, and anomalous head posture
  - Acquired nystagmus
  - Albinism-associated nystagmus

IV. Diagnosis and discussion
- Infantile Nystagmus
  - Idiopathic infantile nystagmus is the most common type of infantile nystagmus, followed by nystagmus associated with an ocular disease.
  - Characteristics
    - Involuntary, bilateral, conjugate, eye movements.
    - The nystagmus develops within the first 6 months of life (usually between birth to 12 weeks).
The waveform of the nystagmus is usually pendular initially, but with age develops into jerk-like nystagmus. The jerk component is developed in order to extend foveation period, allowing for target to be on the fovea for a longer period of time.

Foveation improves up to around 1.5-2 years but remains stable thereafter. The nystagmus can dampen with convergence, resulting in better visual acuity at near than at distance. Up to 64% of patients with nystagmus can also present with strabismus. Patients usually do not report oscillopsia.

Recent PET-MRI study shows down regulation of cortical activity in the area of MT/V5 bilaterally in a patient with infantile nystagmus. This area of the brain plays an important role in motion processing.

The nystagmus can have a strong genetic component or be singly affected. Most common inheritance pattern is x-linked. The gene FRMD7 (FERM-domain-containing-7 gene on chromosome Xq26.2) has been found to be a major cause of hereditary X-linked nystagmus.

The expected visual acuity is 6/12 (20/40) or better.

Compensatory strategies

Foveation strategy
- It is generally developed during visual development in order to maximize vision.
- Foveating saccades allows the individual to maximize amount of time eyes are moving slowly (foveation periods) to allow more time for fovea to line up with target.

Anomalous head posture
- This is generally adopted in order reduce nystagmus (occurs when null point does not occur in primary gaze position).
- Helps improve visual acuity.
- This can take the form of one or a combination of the following: head turn, head tilt, and/or chin up/down.

It is important to rule out other ocular or systemic pathology that may be associated with the nystagmus such as albinism, congenital cataract, retinal dystrophy, optic atrophy, or neurological defects. Ancillary tests: electrophysiology, lab tests, neurological, and imaging work-up.

Abadi and Bjerre’s study on infantile nystagmus showed that 62% were classified as idiopathic, 28% albinism, 10% had other ocular pathologies.

Idiopathic infantile nystagmus affects between 1 in 1000 to 1500 children. Two to three-fold male predominance.

Pathogenesis
- Two theories
  - Efferent: result from a primary defect in ocular motor control system.
• Afferent: result from a primary defect in the visual sensory system.

V. Treatment, Management

• Specific to this patient
  o Continue to wear glasses full-time
    ▪ OD: +0.75-0.50X090 9pd BO
    ▪ OS: +1.00-0.50X090 9pd BO
  o Despite this patient not being binocular, he has some relative gross convergence ability as demonstrated by his strabismus magnitude being equal with and without the addition of 18pd BO. This relative convergence with the BO prism is helping to dampen the nystagmus and reduce the AHP.
  o Patient is scheduled for a comprehensive eye exam and follow-up of the infantile nystagmus and AHP in three months.

• General treatment/management options
  o Refractive management
    ▪ Optimal refractive error correction is addressed first. 
    ▪ Yoked prism for patients with anomalous head posture.
      • Apex is pointed in the direction of the patient’s gaze. 
    ▪ Base-out prisms are prescribed binocularly to stimulate convergence for patients with fusion whose nystagmus dampens in convergence. 
      • This is usually only effective in those with binocular vision.
  o Contact lens
    ▪ This has been found to partially suppress the nystagmus. 
    ▪ The lenses allow for the optical center of correction on patient’s visual axis to be maintained.
    ▪ The lenses also broaden the high foveation-quality field.
  o Surgical options
    ▪ The aim is to correct anomalous head posture or to improve visual acuity by decreasing nystagmus amplitude or frequency.
    ▪ Anderson-Kestenbaum procedure: includes paired resections and resection of rectus muscles pairs. 
      • Shifts the position of the null point to primary gaze. 
    ▪ Cuppers’ divergence procedure involves bilateral recession of the medial rectus muscles and essentially diverge the eyes in patients whose nystagmus dampens with convergence. 
    ▪ Tenotomy and reattachment procedure: “broaden the null zone and reduce the intensity” of the nystagmus. 
      • This is not a first-line treatment. 
      • Tendons of EOMs are cut at the point of insertion and reattached.
    ▪ Surgery usually deferred until 6-8 years of age to ensure that AHP has stabilized. 
    ▪ There is a possibility of recurrence of AHP following surgery.
  o Pharmacological options
    ▪ Gabapentin and Memantine
      • GABA analogs. 
      • Positive effect in visual acuities, reduced nystagmus intensity and improved foveation when compared to placebo. 
      • The drugs showed positive effects in patients with idiopathic infantile nystagmus as well as patients with infantile nystagmus associated with other visual/ocular disorders.
• Botulinum toxin injections
  • This is rarely used due to its limited period of action and tendency to cause adverse reactions.1,3,12
  • Adverse effects include ptosis and diplopia.5

VI. Conclusions

• Infantile nystagmus can present by itself or can be associated with other ocular or neurological diseases. Although the majority of infantile nystagmus is idiopathic in nature, it is still important to take a thorough history and decide whether further ancillary tests are necessary.

• Clinical Pearls
  o Don’t be afraid to think outside of the box. Typically in strabismic patients with nystagmus, base-out prism would not be expected to make a significant difference, as these patients do not have good convergence ability. However, in this particular case, despite not being binocular, the patient demonstrated some degree of relative convergence with the base-out prism. This significantly improved his anomalous head posture.
  o Be aware of treatment options that can help relieve the patient’s symptoms, even if it is temporary. In this particular case, having the significant head turn and head tilt at such a young age can be detrimental to the patient’s musculoskeletal development. Although surgery may be an option in the future, it is important to come up with a treatment plan that can be implemented until the time that the patient is old enough and the AHP is stable enough to warrant surgical intervention.