Understanding Nystagmus: Diagnosis, Related Disorders, Treatment, and Research

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Nystagmus

“All that wiggles is not nystagmus.”

**Definition**: Involuntary oscillation of the eyes, characterized by a slow phase, followed by a fast phase.
Physiologic Nystagmus

- End-point (eccentric-gaze) nystagmus
Physiologic Nystagmus

Vestibular Nystagmus
Physiologic Nystagmus

Optokinetic Nystagmus
Infantile-Onset Nystagmus

- **Congenital nystagmus**
  (Infantile nystagmus syndrome*)
- **Latent / manifest latent nystagmus**
  (Fusional maldevelopment nystagmus syndrome*)
- **Spasmus nutans**
  (Spasmus nutans syndrome*)
- **Others**: congenital see-saw, downbeat, upbeat, or familial vertical pendular

(*: Proposed by Classification of Eye Movement Abnormalities and Strabismus Working Group, NIH, 2001)
Acquired Nystagmus

- See-saw nystagmus
- Periodic alternating nystagmus
- Downbeat nystagmus
- Upbeat nystagmus
- Gaze-evoked nystagmus
- Vestibular (central or peripheral) nystagmus
- Others, including acquired pendular forms
Congenital Nystagmus: Related Disorders

- **Amblyopia**: poor vision caused by abnormal visual development, due to abnormal visual stimulation
- **Susceptible ages**: birth to 7 or 8 years
Congenital Nystagmus: Related Disorders

Strabismus
Congenital Nystagmus: Related Disorders

Albinism
Congenital Nystagmus: Related Disorders

Optic Nerve Hypoplasia
Congenital Nystagmus: Related Disorders

Aniridia
Congenital Nystagmus: Related Disorders

Retinal Dystrophies

Rod Cone

Rods & Cones

capillary network of inner blood-retinal barrier

outer blood-retinal barrier

choroid
Peter’s Anomaly—
Congenital Corneal Opacity

Congenital Nystagmus: Related Disorders
Congenital Cataracts
Latent / Manifest Latent
Nystagmus:
Related Disorders

- Any eye condition that affects normal development of “fusion,” usually by affecting vision in at least one eye

- **Amblyopia**: poor vision caused by abnormal visual development, due to abnormal visual stimulation
  - Susceptible ages: birth to 7 or 8 years
Latent / Manifest Latent Nystagmus: Related Disorders Strabismus
Latent/Manifest Latent Nystagmus: Related Disorders

Congenital Cataracts

Déjà vu? Yes, there is considerable overlap!
Peter’s Anomaly—Congenital Corneal Opacity

Latent/Manifest Latent
Nystagmus:
Related Disorders
Spasmus Nutans: Related Disorders

- Strabismus
- Amblyopia
- In spasmus nutans-like disease: brain abnormalities
Diagnosis

- **History**
  - Onset
  - Symptoms
    - Light-sensitivity
    - Difficulty in darkness
    - Oscillopsia (sensation of the world shaking or moving)
    - Dizziness, etc.
Diagnosis (cont.)

- Eye exam
- Eye movement recordings
  - To verify type of nystagmus
  - To characterize details, such as convergence-damping, null position, etc.
  - To determine best treatment options
Eye Movement Recording

Infrared Oculography

Magnetic Search Coil
Congenital Nystagmus: Waveforms
Latent / Manifest Latent Nystagmus: Waveforms

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<td>Square-wave jerks</td>
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Spasmus Nutans: Waveform
Diagnosis (cont.)

- Electoretinography (ERG): for diagnosis of retinal dystrophies in particular (cone and/or rod dystrophies)
Diagnosis (cont.)

- **Neuroimaging:** MRI or CT scan—including:
  - For acquired nystagmus
  - For any nystagmus with other associated neurologic abnormalities

- Other tests
Treatment: Optical Correction/Aids

- Glasses, including with tints, prisms
- Contact lenses
- Low vision aids
An optical method of shifting gaze to reduce AHP.
A: The eyes are shifted to a "null" position in right gaze.
B: By introduction of the prism before each eye with the base toward the preferred gaze position (apex toward the AHP), the gaze is shifted straight ahead.
Optical method of stimulating convergence.
Effect of Prisms on CN

Red: without prisms
Green: with prisms
Optical method of stabilizing images on the retina
Treatment:
Somatosensory Stimuli

- Contact lenses, soft or rigid gas permeable
- Neck / face electrical stimulation or vibration*
- Acupuncture*

*Problem: practicality

(*DISCLAIMER: The presenter does not necessarily promote these treatments.*)
Treatment: Medications

- Congenital periodic alternating nystagmus:
  - Baclofen (not as useful as for acquired periodic alternating nystagmus)

- Pendular nystagmus, in multiple sclerosis:
  - Gabapentin
  - Memantine
Retrobulbar Space

Botox Injection

Treatment: Medications (cont.)
**Treatment: Medications (cont.)**

- **Botox** injection into orbit
- **Disadvantages:**
  - Possible **droopy eyelid** ("ptosis")
  - Possible **double vision**
  - **Wears off in about 8 weeks, requiring repeated injections**
Treatment: Surgery

- Strabismus correction
Treatment: Surgery

- Head posture correction (e.g. Anderson-Kestenbaum procedure)
Treatment: Surgery

- Improvement of vision
  - Artificial divergence surgery
  - Anderson-Kestenbaum
  - Combined artificial divergence and Anderson-Kestenbaum
Eye Muscle Surgery

Site of disinsertion during surgery
Eye Muscle Surgery

Eye Muscle (hooked and sutured)

Suture

Nose

Suture

Forehead
STABLE OCULAR MOTOR SYSTEM
New Research: Congenital Nystagmus

- Underlying cause—proposed:
  - Lack of normal visual motion during a critical period of development in infancy
  - Possible abnormal proprioception (sensation of position)
New Research: Congenital Nystagmus

- Diagnosis of waveforms is possible in infancy: 6 months of age or less
- Correlation with vision
New Research: Congenital Nystagmus

- **Oscillopsia** (=sensation / perception of world moving / shaking)
  - A debilitating problem in very few congenital nystagmus patients
  - Associated with decompensated motor or sensory status (e.g. strabismus, retinal dystrophy)
  - May be helped with treatment for the underlying cause
New Research: Congenital Nystagmus

- **Refractive errors** (i.e. astigmatism, far-sightedness, near-sightedness)
  - Wider range in CN / albinism patients
  - CN / albinism patients with refractive errors have less tendency to progress toward “emmetropia” (no refractive error)
New Research: Congenital Nystagmus

- Torsional component of CN
  - Does not significantly affect vision in the null position
  - Generated centrally (in brain), as opposed to peripherally (in eye muscles)
New Research: Congenital Nystagmus

- Psychophysical tests
  - Orientation thresholds:
    - Lines of different orientations and differing lengths presented for differing periods of time
    - CN patients require more time to recognize the orientations
    - CN patients have a harder time with vertical lines than horizontal lines
New Research: Congenital Nystagmus

Psycho-physical tests

• Latency of optotype recognition

Plasma Screen
At or above visual acuity, CN patients required more time to recognize the “E” than controls.
At or above visual acuity, CN patients required more time to recognize the “E” than controls
New Research: Congenital Nystagmus

- **Neuroanatomic study** of the muscle/eye junction ("enthesis")
  - Differences compared to patients without nystagmus
Enthesial Area
CONTROL HUMAN ENTHESIS

Myelin
Axon
Nerve Ending
Capillary

Labels:
- Myelin
- Axon
- Nerve Ending
- Capillary
INS IDIOPATHIC 18 Mos M

A. Unmyelinated Nerve Ending

B. Nerve Endings c Dilated RER

C. Nerve Endings c Dilated RER

D. Fragmented Neurolemma
New Research: Congenital Nystagmus

- **Neuroanatomic study** of the muscle/eye junction ("enthesis")
  - Suggests a possible role for abnormal proprioception (sensation of position) as cause of disease
New Research: Congenital Nystagmus

Medication effect:

- One adult CN patient improved with diet pill*
- One child with CN improved with ADHD pill*
- Medications improved vision, strabismus, and depth perception, despite *increased* nystagmus intensity
- Likely reason: *more foveations per second*

*These drugs may carry serious side effects and have NOT undergone clinical trial for treatment of nystagmus.
New Research: Congenital Nystagmus

- **Surgical treatments**
  1. Horizontal rectus tenotomy study in humans (NIH study)
     - Improved visual acuity in the children
     - Improved vision subjectively in adults

(To be published soon—stay tuned...)
New Research: Congenital Nystagmus

- Surgical treatments
  2. LASIK (laser-assisted in situ keratomileusis, refractive surgery)
    - In 47 y.o. woman with CN
      - Moderate amplitude and frequency
      - Convergence-damping
      - No eccentric-gaze null
    - After 2 treatments with pupil-tracking laser system:
      - 20/40 each eye pre-op with glasses
      - 20/40 each eye post-op without glasses
New Research: Congenital Nystagmus

Surgical Treatment: LASIK
New Research: Congenital Nystagmus

Surgical Treatment: LASIK

Corneal Flap

Laser applied to the stroma
New Research: Congenital Nystagmus

- LASIK for CN (INS) patient??? Only if:
  - Convergence-damping nystagmus
  - No eccentric null-point
  - Smaller nystagmus frequencies and amplitudes
  - Experienced surgeon
  - Advanced pupil-tracking laser systems
  - With full knowledge of the patient about problems with eccentric ablation zones, including monocular diplopia, glare, halos, and other debilitating symptoms
New Research: Acquired Nystagmus

- Localizing defects in the brain
- **Optical treatment**: an electro-optic device, for pendular nystagmus, e.g. in multiple sclerosis
- **Medications**, e.g. in multiple sclerosis, periodic alternating nystagmus
Thank you for your attention