6.13.2. Childhood Nystagmus (II): Congenital nystagmus

periodic alternating nystagmus (PAN)

- types
  - congenital
    - associated with oculocutaneous albinism
    - Arnold-Chiari malformation
  - acquired
- clinical presentation
  - jerk nystagmus
  - strictly horizontal
  - nystagmus periodically changes direction
    - starts with a jerk nystagmus in 1 direction
    - slowly begins to damp, leading to a period of no nystagmus x 10 - 20 seconds
    - jerks in the opposite direction
    - cycle repeats = every 4 minutes
  - some children adopt an alternating head turn
- Periodic alternating nystagmus (PAN)
  - associated with oculocutaneous albinism
  - congenital or acquired
  - strictly horizontal
  - oscillation cycle of 2–4 minutes
    - any purely horizontal nystagmus that occurs in primary position should be observed for at least 2 minutes
    - change in amplitude and frequency
    - crescendo–decrescendo nystagmus (1)
    - 10-20 seconds
    - short period of downbeat or no nystagmus (2)
    - crescendo–decrescendo nystagmus in the opposite direction (3)
  - periodic alternating head turn
    - to minimize the nystagmus, in accordance with Alexander's law
  - dysfunction of the cerebellar nodulus and uvula
    - multiple sclerosis
    - cerebellar degeneration
    - Arnold-Chiari type I malformation
    - stroke
    - use of anticonvulsant medication
  - bilateral loss of vision
    - reversible
  - baclofen
    - can be effective for the acquired form

latent nystagmus

- occurs in children with decreased fusion
  - most commonly infantile esotropia
  - early-onset strabismus
  - decreased vision in 1 or both eyes
- clinical presentation
  - when 1 eye is occluded, a conjugate jerk nystagmus develops
    - fast phase directed toward the uncovered eye
      - left jerk nystagmus occurs when the right eye is covered
      - the only form that reverses direction with a change in fixation
    - the nystagmus damps when the fixating eye is in adduction
      - occlusion of the right eye causes left jerk nystagmus with a left head turn
      - head turn reverses with a change in fixation
      - Figure 13-2 (Courtesy of American Academy of Ophthalmology)
    - in CMN, nystagmus damps with convergence
  - asymmetries in amplitude, frequency, and velocity of the nystagmus can also be present
  - binocular visual acuity is better than monocular
  - manifest latent nystagmus
    - other eye is suppressed or amblyopic
    - nystgmus becomes manifest when both eyes are open but only 1 eye is being used for vision
  - nystagmus waveform
    - in contrast to exponentially increasing velocity of slow phase with CMN
    - exponentially decreasing velocity of slow phase
- Fusional Maldevelopment Nystagmus Syndrome (Latent Nystagmus)
  - clinical characteristics
    - high-plus lens or filter
    - partial optical blurring of 1 eye permits better visual acuity measurements
    - use of a standard occluder degrades visual acuity
    - begins (or accentuates) when binocular viewing is disrupted
    - early-onset
    - constant-velocity slow phase
      - in contrast to the increasing exponential slow phase of INS/CN
    - conjugate horizontal jerk nystagmus
    - direction changing with monocular occlusion
      - fast phase toward the viewing eye; slow phase toward the nose/occluded eye
      - both slow phase and eye are toward nose (occluded eye)
    - almost always associated with esotropia
    - subnormal stereopsis
    - may coexist with INS
    - frequently, with dissociated vertical deviation
  - manifest latent nystagmus (MLN)
    - nystagmus with characteristics of LN that is present when both eyes are open
    - nystagmus spontaneously and intermittently develops whenever the esotropic eye is physiologically suppressed
  - LN and MLN are benign entities

congenital motor nystagmus (CMN)

- not associated with central nervous system abnormalities
- binocular conjugate nystagmus
- nearly normal visual function
- plane of nystagmus is most often horizontal
  - remains so on upgaze and downgaze (uniplanar)
- exponential increase in velocity of the slow phase
  - Figure 13-1 (Courtesy of American Academy of Ophthalmology)
- ± null point
- ± abnormal head position
  - head position becomes more pronounced as child approaches school age
- head bobbing or movement
  - may be present initially but usually decreases with age
- oscillopsia is rare
- damped by convergence
  - can be associated with esotropia
  - nystagmus blockage syndrome
    - combination of nystagmus and esotropia
    - characteristically present with an esotropia that "eats up prism" on attempted measurement
    - exhibit an increased jerk nystagmus on attempted lateral gaze
- paradoxical inversion of OKN response
  - 2/3 of patients
  - patient with right jerk nystagmus views an OKN drum rotating to the patient's left
  - exhibits a damped right jerk nystagmus or possibly even a left jerk nystagmus
  - occurs only in CMN

Infantile Nystagmus Syndrome (INS)/ Congenital Nystagmus (CN)

- epidemiology
  - onset in first few months of life
  - may not become evident until several years of age
  - ± family history
- clinical presentation
  - no oscillopsia
  - conjugate
  - horizontal
    - remains horizontal in upgaze and downgaze
  - continuous or intermittent
  - jerk or pendular
    - depending on position of gaze
  - accentuation by visual attention & distant fixation
    - unlike peripheral vestibular nystagmus
  - diminishment by convergence
  - abolished in sleep
  - slow phase in the direction opposite that of a rotating optokinetic drum
    - reversal of normal pattern!
      - in contrast to constant velocity slow phase of latent nystagmus
  - exponential increasing velocity of slow phase with distance from fixation
    - requires eye movement recordings
  - there is frequently a null point
    - field of gaze in which nystagmus intensity is minimal
    - head turn or posture
      - if the null point is not in primary position
  - strabismus
    - 15%
  - not associated with afferent pathway disease
    - visual acuity is proportional to the foveation period
- sensory nystagmus
  - central visual acuity is too poor to develop a stable fixational mechanism
  - etiology
    - ocular albinism
    - achromatopsia
    - leber congenital amaurosis
    - aniridia
  - in any patient with early-onset nystagmus look for evidence of damage to the visual pathways
    - impairment of visual tracking
    - optic atrophy
    - presence of such abnormalities should prompt neuroimaging
    - electrophysiologic testing (ERG, VEP) is warranted

congenital sensory nystagmus

- pathogenesis
  - bilateral pregeniculate, afferent visual pathway abnormality
- if the visual pathway disorder is present at birth, the resulting nystagmus becomes apparent in the first 3 months of life
- clinical presentation
  - type of nystagmus
    - searching nystagmus
      - slow searching/wandering conjugate horizontal eye movements without fixation
      - usually seen in children whose visual acuity is < 20/200
    - pendular nystagmus
      - patients with visual acuity of >20/200 in at least 1 eye
    - jerk nystagmus
      - patients with visual acuity of 20/60 - 20/100
  - severity is somewhat correlated with the degree of vision loss
  - waveform cannot be distinguished from that of congenital motor nystagmus
  - congenital sensory nystagmus as a result of a retinal dystrophy
    - mild vascular attenuation
    - optic disc pallor
    - ± completely normal retinal appearance
    - electroretinogram may be required

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