Investigation of infantile onset nystagmus

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Financial Disclosures

NONE

Infantile Nystagmus

- bilateral visual impairment
- neurological disease
- congenital idiopathic motor nystagmus

Nystagmus is uncommon with higher visual pathway damage

Infantile nystagmus

early onset
- abnormal eye examination
- normal eye examination

acquired
- spasmus nutans
- chiasmal disease
Infantile nystagmus: Key questions
- can the child see?
- is there a family history of nystagmus?
- light sensitivity?
- is the child neurologically normal?
- is the eye examination normal?
- early onset or acquired?
- typical or atypical form?
- neurologically localising form?
- is this CIMN?

Assessment of visual responses
 observes
- Visual attention
- Fixation shifts
- Is vision consistent with a diagnosis of CIMN or worse than this?

Character of nystagmus
- Pendular or jerk?
- Symmetrical or asymmetrical?
- Horizontal vertical or rotary?
- Continuous or discontinuous?
- Varies in different gaze positions?

Refraction
- High refractive errors in:
  - Albinism
  - Aniridia
  - Retinal dystrophies
Nystagmus associated with anatomical abnormality of eyes

Always exclude iris translucency and examine mothers of male infants

Nystagmus associated with systemic disorder

X-linked ocular albinism
- reduced acuity
- nystagmus
- ocular signs of albinism
- chiasmal misrouting
- normal skin pigmentation

Examine mothers of male infants with albinism
X-linked ocular albinism: female heterozygotes

Albino misrouting

VEPS useful in atypical ocular hypopigmentation

F, 7 yrs. EDD 35661. Oculo-cutaneous albinism. VAR, VAL 6/24

Pattern appearance

Flash
Subtle ocular abnormalities: optic nerve hypoplasia

- High index of suspicion in infants with nystagmus and poor pupil responses
- Direct ophthalmoscopy
- Need for repeat ophthalmoscopy in infants with nystagmus
- Optic atrophy or hypoplasia?

Optic nerve hypoplasia

Infantile retinal dystrophies

- Lebers amaurosis
- Rod monochromatism
- S-cone monochromatism
- Congenital stationary night blindness

Needs ERG to confirm diagnosis

Lebers amaurosis

- Infantile onset rod-cone dystrophy
- AR inheritance
- Poor vision from infancy
- Nystagmus
- High hyperopia
- Variable fundus appearance
- Non-recordable ERG
F, 1.5 yrs. Leber congenital amaurosis

ERG: Peri-orbital electrodes

(Courtesy of G. Holder)

Achromatopsia
- infantile onset
- photophobia
- nystagmus
- reduced acuity
- normal retinal appearance
- absent photopic ERG
- normal rod ERG

X-linked CSNB
VAR 6/18; VAL 6/36

(Courtesy Graham Holder)

Cortical visual impairment and Nystagmus

Most children with CVI do not have nystagmus

Exceptions are:-

- Extensive brain injury
- Cerebellar abnormality
- Ex preterm infants with PVL
**Congenital idiopathic motor nystagmus**
- usually horizontal-all positions gaze
- may be head nodding or AHP
- relatively good acuity
- damps on convergence
- normal pupil responses
- normal fundus examination
- normal ERG
- Usually sporadic but X-L and Ad forms exist

**Manifest latent nystagmus**
- Associated with strabismus
  - Esotropia and DVD
- Horizontal
- Worse on covering one eye
- Fast phase towards fixing eye

**Nystagmus diagnosis : key messages**
- Detailed history
- Look for iris translucency
- ERG in all cases with normal fundus
- VEP if suspicion of mild albinism
- MRI scan in:-
  - Acquired nystagmus
  - Neurologically localising forms (rare in infants)
  - Unocular or atypical nystagmus
- Keep diagnosis under review
- Molecular genetic testing on horizon