Understanding and Treatment of Infantile Nystagmus Syndrome

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Examination Techniques: Highlights

- **Acuity**
  - Binocular and Monocular
  - Gaze-Dependent

- **Color, Contrast**

- **Ocular Motor**
  - Strabismus
  - Nystagmus – “nulls”
  - Head Posture

- **Accommodation**

- **Refraction**
  - Objective
Visual Acuity Testing

20/400  20/200  20/100  20/50  20/25
Evaluation Techniques: Afferent System

- Vision testing procedures
  - Behavioral Vision Testing (acuity, color, stereo)
  - Visual Evoked Responses (flash, pattern, sweep)
  - Electoretinography (flash, pattern)
  - Contrast, Color and Visual Field Testing
Evaluation: Efferent System
Eye Movement Recordings

• Methods
  ➢ High speed photographic methods.
  ➢ “Contact” electrooculography.
  ➢ Infrared reflectance oculography.
  ➢ Scleral contact lens/magnetic search coils.
Eye Movement Recordings

- Diagnosis/Differentiation of Eye Movement Disorders.
- Utility as an “Outcome Measure” in Clinical Research.
Eye Movement Recordings

- Value of data
  - Diagnosis.
  - Classification.
  - Etiology.
  - Therapy.
  - Research.

685 Patients 1998-2005
Afferent System

Conception

Development

Birth

Infancy

Efferent System

Vision

Vergence, Versions

STABLE OCULAR MOTOR SYSTEM
**CEMAS**

| Disease Name | INFANTILE NYSTAGMUS SYNDROME (INS)  
[Old Congenital Nystagmus and “Motor and Sensory” Nystagmus] |
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<td>Criteria</td>
<td>Infantile onset, ocular motor recordings show diagnostic (accelerating) slow phases</td>
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| Common Associated Findings | Conjugate, horizontal-torsional, increases with fixation attempt, progression from pendular to jerk, family history often positive, constant, conjugate, with or without associated sensory system deficits (e.g., albinism, achromatopsia), associated strabismus or refractive error, decreases with convergence, null and neutral zones present, associated head posture or head shaking, may exhibit a ”latent” component, “reversal” with OKN stimulus or (a)periodicity to the oscillation. Candidates on Chromosome X and 6  
May decrease with induced convergence, increased fusion, extraocular muscle surgery, contact lenses and sedation. |
| General Comments | Waveforms may change in early infancy, head posture usually evident by 4 years of age. Vision prognosis dependent on integrity of sensory system. |
Nystagmus and Vision

• “Sensory” System
  - Refractive Error
  - Amblyopia
  - Abnormal Binocular Vision
  - Ocular Media Damage
  - Retinal Disease
  - Nycloptia/Photophobia
  - Optic Nerve Disease
  - Visual Cortex Disease

• “Motor” System
  - Oscillation
  - Strabismus
  - Abnormal Pursuit (tracking)
  - Abnormal Saccades (fast eye movements)
“MOTOR” SYSTEM TREATMENT

- Medications
- Visual Training (strabismus, binocular dysfunction)
- Acupuncture
- Biofeedback
- Vibratory Stimulation
- Prisms, Telescopes, Contact Lenses
- Botox
- Eye Muscle Surgery
Medical Treatments

- *Spectacles*
- *Contact Lenses*
- *Low Vision Aids*
- *Penalization (patching, drops)*
Medical Treatments

- Photophobia
- Nystagmus
  - Sedatives, Hypnotics, Neuroleptics, Anti-seizure drugs
  - Acupuncture, Biofeedback, Vibratory Stimulation
- Strabismus and binocular dysfunction
  - Orthoptics
  - Spectacles
  - Penalization
“Nystagmus” Surgery

• Effect a Positive Change on the Oscillation
  ➢ Improve Waveform
  ➢ Increase Foveation
  ➢ Broaden Null Position
  ➢ Improve Periodicity

• Treat Anomalous Head Positions
ANIMAL MODEL OF INS

- Achiasmatic Belgian Sheepdogs
- Ocular Motor Behavior
- Ocular Motor Analysis
- Infrared Oculography Recording
- Preoperative and Postoperative
  - Visual Behavior
  - Eye Movement Recordings
HUMAN CLINICAL TRIALS
EYE MUSCLE SURGERY AND INS

- Simple tenotomy of all 4 horizontal recti
- Reattachment at the original insertion
- Final Effect related to underlying visual system disease

Hertle RW, Dell’Osso LF, FitzGibbon, EJ, Yang D, Mellow SD.
Horizontal Rectus Muscle Tenotomy In Children with Infantile Nystagmus Syndrome: A Pilot Study.
Journal of AAPOS 2004;8;539-548

Hertle RW, Dell’Osso LF, FitzGibbon, EJ, Thompson DJS, Yang D, Mellow S.
Horizontal Rectus Tenotomy In Patients with Congenital Nystagmus: Results In Ten Adults
Ophthalmology 2003;11;2097-2115
**HUMAN CLINICAL TRIALS**

**EYE MUSCLE SURGERY AND INS**

- **Increased Foveation** (amount of time during a beat of INS during which the eye is moving at <4 deg/sec and within a few degrees of the target – when the eye/brain “sees”)

[Graph showing eye movement before and after surgery]

**Preferred OD Fixing Under Binocular Conditions**
Improved Waveforms (Pure Jerk and Pendular to Jerk/Pendular with foveation)

HUMAN CLINICAL TRIALS
EYE MUSCLE SURGERY AND INS

Pre-operation Nystagmus

Post-operation Nystagmus

Preferred OD Fixing Under Binocular Conditions
HUMAN CLINICAL TRIALS
EYE MUSCLE SURGERY AND INS

- Increased Breadth of The Null Zone
HUMAN CLINICAL TRIALS
EYE MUSCLE SURGERY AND INS

• 1-3 Lines of Recognition Acuity Increase
HUMAN CLINICAL TRIALS
EYE MUSCLE SURGERY AND INS

• Improved Visual Recognition Time (Speed of Recognition)
GAZE DEPENDENT VISUAL ACUITY

Fig. 1. Gaze angle
• Improved Gaze Dependent Visual Acuity (GDVA)

**PRE-POST GDVA PT. 19**

**PRE-POST GDVA PT. 25**
“Myotendon”

Annulus Of Zinn

Enthesial Area
CONTROL HUMAN ENTHESIS

A. Myelin

B. Nerve Ending

Capillary
TREATMENT: ANIMAL MODEL

Etiologic

- INS with Gene Defect (RPE65 – Leber’s in Humans)
- Genetic Therapy*
Conclusions
Ask For:

- **Accurate Evaluation**
  - Afferent System
  - Efferent System

- **Accurate Diagnosis**
  - Sensory System Deficits
  - Nystagmus Type
  - Strabismus
  - Head Posturing

- **Medical Treatment Options**
- **Surgical Treatment Options**
- **Treatment versus “CURE”**