CONGENITALNYSTAGMUS
& other types of ‘ophthalmic’ nystagmus
…for neurologists 2009

LIONEL KOWAL

Ophthalmologist, practice restricted to Strabismus

Ocular Motility Clinic, Royal Victorian Eye and Ear Hospital, Melbourne

LARRY ABEL PhD

Optometry School, University of Melbourne

ELAINE WONG

Royal Victorian Eye and Ear Hospital,

CERA, University of Melbourne

SELECTION BIAS

Blind men and an elephant

From Wikipedia, the free encyclopedia

The story of the **blind men and an elephant** originated from India.

In various versions of the tale, a group of blind men (or men in the dark) touch an elephant to learn what it is like. Each one touches a different part, but only one part, such as the side or the tusk. They then compare notes on what they felt, and learn they are in complete disagreement. The story is used to indicate that reality may be viewed differently depending upon one's perspective, suggesting that what seems an absolute truth may be relative due to the deceptive nature of half-truths.

"Blind monks examining an elephant", an 1888 ukiyo-e print by Hanabusa Itchō.

Selection Bias :
N seen by ENT, ophthalmologist, neurologist, neurosurgeon –
all very different

Is it important to detect ‘ophthalmic’ nystagmus?

- If it’s confidently ‘congenital’ or ‘ophthalmic’ you don’t have to worry about any neurological problem

Terrible Terminology

2 main types of ‘congenital’ N

- lower case ‘c’: cN:

any type of early onset N

2 specific types of cN:

- Upper case ‘C’: CN: Congenital N
- LMLN: Latent Manifest Latent N

Congenital N = CN

aka Congenital Motor N CMN,
Congenital Sensory N CSN,
Idiopathic Infantile N IIN
Infantile N Syndrome ***INS*** [‘latest’ label]

Subtype: Periodic Alternating N PAN
≈ 10% in Melbourne

LMLN

Latent Manifest Latent Nystagmus

aka FMNS = Fusional Maldevelopment N Syndrome.

Can be obvious = manifest = MLN [Manifest Latent Nystagmus] or

'Pure' LN [Latent Nystagmus], only apparent when one eye is covered

*neurologically innocent

CN

1. **Convergence null ***
2. **Eccentric null**
3. L beat to L of null
4. R beat to R of null
5. ± Latent component
6. Horizontal, can be vertical**
7. ± Oscillopsia**
8. ± Strabismus

LMLN

1. **Fast beat to fixing eye***
2. Adduction null
3. N on lateral gaze
4. Latent component
5. Horizontal, can be torsional **
6. ± Oscillopsia**
7. Strabismus in nearly all

** NOT well known

VIDEOS CN

- 3 videos:
- 1. N on LG, PP
- 2. null on RG
- 3. conv null

VIDEOS CN

- 3 videos:
- 1. N on LG, PP
- 2. Null on RG - Will adopt face turn to L when he wants to see clearly
- 3. conv null

VIDEOS CN

- 3 videos:
- 1. N on LG, PP
- 2. null on RG
- 3. Conv null

VIDEOS LMLN LN

Why does CN occur? 1

- Poor ***symmetric*** acuity @ a critical time of visual development

Sometimes there is continuing evidence of the initiating cause – eg**bilateral optic n hypoplasia**, symmetric terrible refraction, cone dystrophy, cataract OU, **albinism** [macular hypoplasia or disc dysplasia]

. . .and sometimes there is no evidence of the presumed / possible initiating cause – macular hemorrhages, terrible refraction that got better

Why does CN occur? 2 *other*

- Genetic reasons – the N waveform itself can be inherited as an isolated issue

In CSNB: pathognomic waveform

- Abnormal CNS : Peri Ventricular Leukomalacia PVL

Many hypotheses / ?Multiple causes

- ? abnormal circuit between fixation and ocular stabilisation systems
- Abnormal proprioceptors in enthesis* [? cause, ?effect]

* where tendon inserts into sclera

Why does LMLN occur?

- ***Asymmetry*** in motor or sensory development @ a critical time

Strabismus, amblyopia, monocular cataract, PVL, unilateral optic n hypoplasia,…

Effects of LMLN

Unusual apparently incomitant strabismus

- DHD – H strabismus [usueXodeviation] that can vary depending on wch eye is fixing
- DVD – V strabismus …..

Torsional & Horizontal N

- ….no vestibular symptoms
- ..drives variable face turns and head tilts
- ...head shaking

Whatever caused it, cN will then degrade acuity further.

The amount of CN – associated degradation of acuity can be:

1. Predicted by mathematically dissecting the waveform [NAFX factor - Cleveland]
2. Estimated by assessing foveation time [the duration when N speed <5° / sec as it changes direction]

Acuity is *not* related to *amplitude / frequency* of N

NICE 5/2009: too many mistakes

2. Abnormal head posture

30° into R gaze	15° into R gaze	Primary position	15° into L gaze	30° into L gaze
6/24	6/12	6/18	6/30	6/48

… will have Face Turn to L ~ 15°

- 3. Reduced visual field

Any large face turn: effectively restricted field of best acuity

- 4. Strabismus in ?30%

Background ?3%

Principles of treatment of CN

- 1. Improve the foveation time / NAFX
- 2. Improve 2° effects such as face turn
- 3. Broaden the null zone

Principles of treatment of CN :

1. Improve foveation time & broaden null zone

- Drugs – gabapentin, memantine

Fairly new. Often effective.

? help a CN pt to pass a driving licence test!

- Contact lenses

SCL & HCL have an effect > optical effect

?interfere with local proprioception

- Prism glasses [BOΔ]: induce convergence
- Surgery

Are the effects of these sometimes additive?

2. Improve 2° effects such as face turn

- Prism glasses

Induce a conv null for distance

- Surgery

Effects of treatment of CN

- Improved waveform may result in improved acuity potential limited by any associated pathology
- Expanded null zone = improved field of same or better acuity &cosmetically improved N
- Improved face turn – improved appearance and improved field

Effects / Treatment of LMLN

- Reduced acuity : make *perfectly* straight and convert MLN to LN
- Face turn from adduction null: MR surgery
- Head tilt from intorsion null: torsional surgery

Defining the type of N

- Do we need Eye movement recordings EMR ?

Clinical Features : It should be possible to separate CN from LMLN in the office

CN

1. Convergence null (better reading VA)
2. L beat in L gaze / R beat in R gaze (either side of null zone)
3. Eccentric null
4. Latent component (can be pseudo-latent)
5. Usu. horizontal, can be torsionalor vertical or all

LMLN

1. Fast beat to fixing eye
2. Adduction null – can cause face turn
3. Nystagmus on lateral gaze (moving away from adduction null)
4. Latent component
5. Can be horizontal or torsional or both

Discriminating CN from LMLN in the office

- CONVERGENCE NULL :
- Both can have this – different mechanisms
- Adduction null of LMLN can look like conv null of CN

- ECCENTRIC NULL :
- Both can have this – different mechanisms:
- LMLN: null in aDuction or Intorsion. If RE dominant, will have FT to R ± head tilt to R with both eyes. Different with LE fixing.
- CN: eccentric null with RE fixing = LE fixing

Discriminating CN from LMLN in the office

- STRABISMUS :

LMLN nearly 100%

Non-expert may not always pick minimal strabismus

CN ?30%

- VERTICAL / TORSIONAL COMPONENT:

- LMLN: T more than V
- CN: V more than T

CN c.f. LMLN : L beat on L gaze, R beat on R gaze [Alexander's Law]

- CN: N → LE on L of null zone, N → RE on R of null zone
- LMLN :
- LE is fixing & is in LG: BE have N → L

- RE is fixing & is in RG: BE have N → R

…resembles CN

- LATENT COMPONENT:

Both can have this – ? different mechanisms

‘I AM WORSE THAN I USED TO BE’

- LMLN:
- Has lost binocularity and LN → MLN

- CN:
- new neurological lesion has caused deterioration,

or

- Stress – common as a temporary mechanism, rarely permanent

CN c.f. LMLN : L beat on L gaze, R beat on R gaze [Alexander's Law]

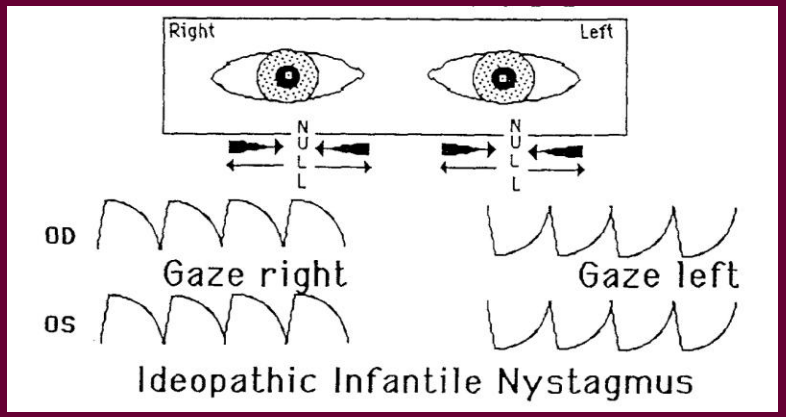
- CN: N → LE on L of null zone, N → RE on R of null zone
- Resembles 'N to fixing eye' of LMLN esp if has ET or XT as well.

- *SO – IT'S NOT ALWAYS EASY TELLING CN & LMLN APART*

EMR features – usually diagnostic

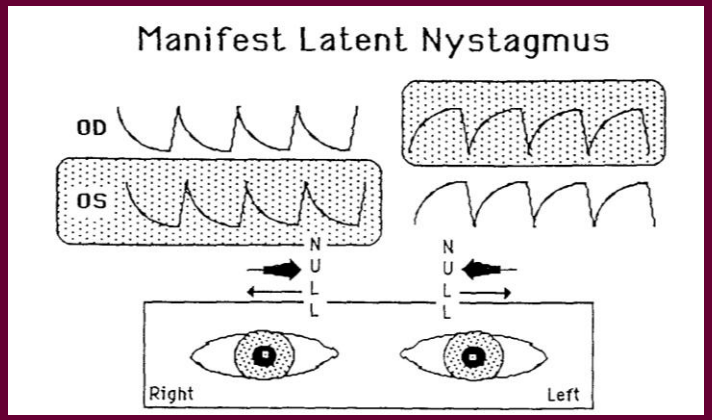
CN

- Exponential increase in velocity in slow phase (unique)
- Fast phase brings eye back to foveation



LMLN

- Decreasing velocity in slow phase (GPN)
- Can be asymmetric in phase, amplitude or frequency



PERIODIC ALTERNATING N PAN

- CN waveform + alternating face turns
  = PAN

PAN:

Acquired form : ‘tight’ periodicity - Every 2 minutes

Congenital form: usually **aperiodic**

e.g. 8 minutes one way, 2 minutes other direction

Not always recordable – takes too long for pt to maintain concentration on target

AUDIT OF LK's CLINICAL EXPERTISE – DIAGNOSING cN TYPE IN THE OFFICE

- 1. By L Abel, the recording engineer by reviewing his findings and my referral notes
- 2. By my [then] Fellow E. Wong who reviewed my files

LARRY ABEL

- 35 patients from the practice of author LK, a highly experienced clinician, were referred to the eye movement lab at my department for recording.
- Clinical diagnoses of nystagmus type(s) present was correlated with what was found upon recording

Results of LA's audit

- Clinical diagnosis: CN (10 patients)
  - Recording: 9 CN, 1 CN and LMLN
- Clinical diagnosis: LMLN (12 patients)
  - Recording:6 CN, 1 CN and LMLN, 5 LMLN
- Clinical diagnosis: CN and LMLN (11 patients)
  - Recording: 3 CN, 4 CN and LMLN, 4 LMLN
- Clinical diagnosis: unknown (2 patients)
  - Recording: 1 CN, 1 LMLN

- Clinical diagnosis CN : usually correct, many missed
- Clinical diagnosis LMLN: ~50% reliable. Overdiagnosed in the office, and many missed
- Clinical diagnosis CN with LMLN: Overdiagnosed in the office, some missed

Clinical correlations : Dr Elaine Wong [then Fellow]

- Chart Review of cN pts n=84
- 60 with EMR
- 42 EMR & LK pre-EMR office diagnosis
  - 5 cases of PAN were excluded from analysis

Overview: office c.f. lab diagnosis

Clinical and EMR Correlation

Office diagnosis of CN is correct 80+% of the time
Office diagnoses of LMLN & CN-LMLN not reliable

Predictive Values of Clinical Features – CN

Conv null present: 70+% are CN

Conv null absent: ≥1/2 are CN **- absence of conv null doesn't exclude CN**

Predictive Values of Clinical Features – CN

These office findings are not diagnostic

Predictive Values of Clinical Features – LMLN

If you don't have N
to fixing eye, 90% are not LMLN

If don't have SPA,

Latent N suggests LMLN ~60%

If you demonstrate SPA, <40% of time it LMLN.

Predictive Values of Clinical Features – CN & LMLN

Office findings are some guide

Conclusions : Clinical and EMR Correlation

- Presence of convergence null – suggestive of CN
- Eccentric null and nystagmus to direction of gaze – less predictive of CN
- No reliable clinical signs for LMLN

Do EMR!!!

Gold standard for diagnosing types of cN

No cardiologist would evaluate an arrhythmia without ECG

No neurologist would evaluate epilepsy without EEG

- Limitations exist! Can miss PAN!

Does everyone with N need to be recorded?

- Not if you're absolutely certain about the diagnosis and have all the information you need for management

OR

- If you don't need to know e.g. N with insignificant face turn, good acuity, no neurological symptoms/ signs….

SURGERY IN cN : LMLN

- Straighten eyes perfectly
- ± 10 Δ not enough: 0 Δ is needed
- Will convert MLN to LN
- Improve acuity, fix oscillopsia
- Other surgeries for face turns and head tilts

SURGERY IN cN : CN

- Eccentric null
- Convergence null
- Null in primary position
- No definite null

SURGERY IN CN : Hertle's operation

- Meta-analysis: ANY horizontal surgery in horizontal CN may improve acuity [probably by improving the waveform] even if the main aim of surgery has not been accomplished e.g. still has residual face turn
- So: Why not try the most basic component of muscle surgery, 'tenotomy – resuture', and see if that improves the waveform?
  - IT DOES!

Eccentric null ≤ 20° L face turn

Recess LMR 7mm & RLR 10 mm + tenotomy – resuture of the other two horizontal recti ~1 mm resection.

If a small duction/version paresis is not created then the head posture will usually return.

Eccentric null ≥ 25° L face turn

Recess LMR 7mm , RLR 10 mm recess

Resect other 2 recti for total 17mm per eye.

If a small duction/version paresis is not created then the head posture will usually return.

SURGERY IN CN : Convergence null

- Prism adapt [no Fresnel] for max BOΔ
- BMR *x* mm based on max BO Δ * + tenotomy – resuture the lateral recti

* *Some: max BMR 3mm*

SURGERY IN CN : No null or null in primary

- Tenotomy – resuture the horizontal recti

Hertle series 400 CN surgeries: 8%

SURGERY IN cN : PAN

- Look for convergence null

Prism adapt view BMR

or

- Tenotomy – resuture horizontal recti

SURGERY IN cN : warnings

- Albinism patients: have positive angle kappa & look divergent when they are straight to cover test

OTHER TYPES OF CONGENITAL / OPHTHALMIC NYSTAGMUS

1. Spasmusnutans
2. Heiman Bielschowsky phenomenon

Spasmusnutans

- Early < 12mo
- Asymmetric N
- Head bobbing
- DD: chiasmal tumour

- Late
- Persisting asymmetric N

HBP

- Slow vertical N in an eye that has developed or did have profound visual loss
- Very under recognised
- ~1Hz
- Vision improving surgery [eg cataract surgery] can cause diplopia

THE RECENT PAST AND THE IMMINENT FUTURE

- Increasing use of EMR to study the effects of different treatments and their combinations on our patients with cN will help us understand their condition better and plan more effective treatments

THANK YOU TO THOSE WHO TAUGHT & STILL TEACH ME ABOUT NYSTAGMUS: DRS. REINECKE, SPIELMAN, ABEL, DELLOSSO & HERTLE

& THANK YOU TO THESE 2 ACHIASMATIC BEAGLES WITH CN WHO CONTRIBUTED TO THE CLINICAL RECOMMENDATIONS IN THS TALK