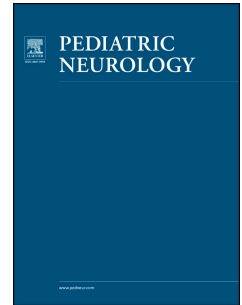


Accepted Manuscript



Is this actually CIN? Comment on: "Congenital Idiopathic Nystagmus: An Observational Diagnosis"

Jay E. Self, BM FRCOphth PhD, Ahmed Salman, MSc, BSc (Hons), Marion F. Hedley-Lewis, DBO(T)

PII: S0887-8994(15)00230-1

DOI: [10.1016/j.pediatrneurol.2015.05.005](https://doi.org/10.1016/j.pediatrneurol.2015.05.005)

Reference: PNU 8664

To appear in: *Pediatric Neurology*

Received Date: 8 May 2015

Accepted Date: 10 May 2015

Please cite this article as: Self JE, Salman A, Hedley-Lewis MF, Is this actually CIN? Comment on: "Congenital Idiopathic Nystagmus: An Observational Diagnosis", *Pediatric Neurology* (2015), doi: 10.1016/j.pediatrneurol.2015.05.005.

This is a PDF file of an unedited manuscript that has been accepted for publication. As a service to our customers we are providing this early version of the manuscript. The manuscript will undergo copyediting, typesetting, and review of the resulting proof before it is published in its final form. Please note that during the production process errors may be discovered which could affect the content, and all legal disclaimers that apply to the journal pertain.

Corresponding Author Details:

Name: Mr Jay Self BM FRCOphth PhD

Role: Associate Professor and Consultant Paediatric Ophthalmologist

Address: Eye Unit and Faculty of Medicine
Room OC16 C Level MP104
Southampton Eye Unit
Tremona Road
Southampton General Hospital
Southampton
SO16 6YD
Tel: 02381 203617
Fax: 02381 204120
Email: jes3@soton.ac.uk

Is this actually CIN?

Comment on: "Congenital Idiopathic Nystagmus: An Observational Diagnosis"

We read with interest the article by Ghosh and colleagues; 'Congenital Idiopathic Nystagmus: An Observational Diagnosis'[1]. Several details require comment.

The 'Patient Description' is missing key information required to diagnose Congenital Idiopathic Nystagmus (CIN) (now more commonly called idiopathic Infantile Nystagmus Syndrome (INS)[2]). For example; there is no description of the direction of the nystagmus in any position of gaze (it typically beats in the direction of horizontal gaze and stays horizontal in up/down-gaze in INS), the effect of monocular viewing (in Fusion Maldevelopment Nystagmus Syndrome, FMNS it can change direction depending on which eye is occluded) or the symptom of oscillopsia.

The nystagmus is described to 'improve significantly when fixing on an object'. If this object is a near target, then the patient will converge and dampening of the nystagmus in this setting (convergence dampening) is a recognised feature of INS[3, 4]. However, INS typically increases with fixation effort, particularly in the distance. Therefore, the details of the 'object' are key here.

Vision is presented in a single measurement, which is presumably, binocular acuity. However, it is important to measure monocular visual acuities and to perform at least a cover test when considering that FMNS is one of the most common causes of nystagmus and is associated with asymmetrical visual acuities/strabismus.

Furthermore, it is unclear whether basic bedside supranuclear eye movement tests have been performed i.e. the Vestibulo-Ocular Reflex, Smooth Pursuit, Optokinetic Reflex and Vergences all of which inform regarding the cause of nystagmus. This is particularly key given the patient history.

Some of the clinical details are misleading and use incorrect terminology. The patient is described as preferring 'her head tilted, with her chin to the left' because her nystagmus was reduced in right gaze. The term head 'tilt' should be used to describe the head posture created by touching the ear to the shoulder and would not be expected to aid nystagmus, which reduces on right gaze. A patient with reduced nystagmus on right gaze might be expected to have a left face turn (the head posture created by looking over ones left shoulder), not a left head tilt.

We agree with the authors comment that INS is characterised by a few key features which can help differentiate it from other causes of nystagmus in children[5]. However, those stated are not the key features, which aid the clinician and many features are missing. We would also caution that nystagmus will commonly worsen with gaze in the direction of the fast phases in many forms of nystagmus (Alexander's law) which is not the same as a null zone seen in INS. Furthermore, a key aspect to diagnosis of INS, when the cardinal features are incomplete, is eye-movement recordings showing accelerating nystagmus slow-phases (only seen in INS).

Therefore, we would caution that a diagnosis of INS requires more information than is provided here and that many of the key features, required to exclude other diagnoses are not presented or discussed.

1. Ghosh, D. and A.D. Rothner, *Congenital Idiopathic Nystagmus: An Observational Diagnosis*. *Pediatr Neurol*, 2015.
2. Workshop, C., *Classification of Eye Movement Abnormalities and Strabismus (CEMAS) Workshop report*. 2001: National Eye Institute Sponsored Workshop.
3. Self, J. and A. Lotery, *A review of the molecular genetics of congenital idiopathic nystagmus (CIN)*. *Ophthalmic Genetics*, 2007. **28**(4): p. 187-191.
4. Casteels, I., et al., *Nystagmus in infancy*. *Br.J.Ophthalmol.*, 1992. **76**(7): p. 434-437.
5. Self, J., et al., *Infantile nystagmus and late onset ataxia associated with a CACNA1A mutation in the intracellular loop between s4 and s5 of domain 3*. *Eye*, 2009.