

Managing Nystagmus in Childhood

Date of Publication: April 2023

Date of Review: April 2025

Introduction

The onset of a spontaneous oscillation of the eyes can occur at any time in life but is most commonly encountered during childhood. In the UK, nystagmus in the general population has been reported to have a prevalence of 2.4 in 1000¹. It can occur as an isolated disorder, in association with a number of different eye conditions, or as a result of a range of neurological disorders. The onset of nystagmus in childhood is not rare and can be the cause of significant clinical and parental concern, and sometimes requires urgent investigation. There is currently no standard clinical approach to investigating nystagmus in childhood^{2,3}.

The goal of this Clinical Practice Points (CPP) Document is to provide a single point of reference for busy clinicians when managing these complex patients from differential diagnosis, through long-term management, to discharge. Equally important, nystagmus is usually associated with visual impairment, and has been shown to adversely impact on quality of life⁴. Therefore, attention is also paid here to an element of care which is often overlooked⁵: the provision of support for patients and carers throughout and beyond clinical care pathways.

Nystagmus and other eye oscillations can also arise in later life, often due to acquired neurological conditions. This document is specific to nystagmus in children.

How to investigate children with Nystagmus?

Diagnosing children with nystagmus can be complex and involve a number of investigations. However, there is a general consensus that a basic clinical investigation, including history taking, orthoptic assessment, anterior and posterior segment examinations, refraction, and examination of the nystagmus itself, comprise the minimum of an initial work-up^{3,15} and are important prerequisites to more individualised investigations.

Subsequent investigations should be dictated by findings from the basic history and examination and by clinical priority (see figure 1). Local access to resources or expertise may alter the order of investigations but should not preclude thorough and complete diagnostic investigation.

Electrodiagnostic testing continues to have a key role in diagnosing children with nystagmus in addition to providing information about visual prognosis^{3,14}. Eye movement recordings (EMRs) can also aid diagnosis but are not used routinely for diagnosis in most centres.

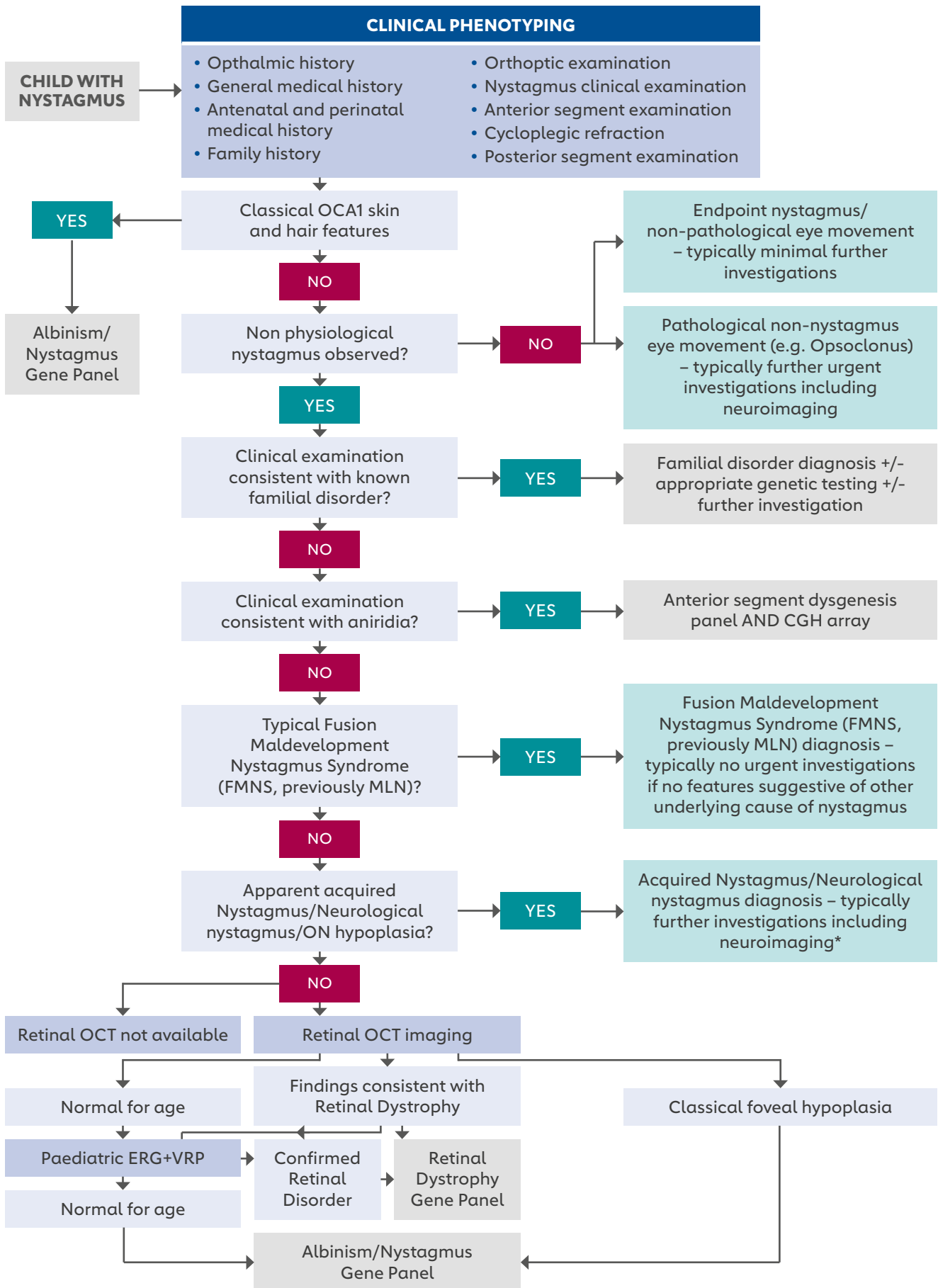
The advent of genomic testing by next generation sequencing is now facilitating faster and more accurate diagnosis of the underlying aetiology for more children with nystagmus than ever before. Specific gene panels for nystagmus and associated disorders have been developed^{3,6-9}, and the National Genomic Test Directory (UK) includes an 'Albinism or congenital nystagmus' gene panel (currently the R39 panel): (www.england.nhs.uk/publication/national-genomic-test-directories).

Similarly, advances in Optical Coherence Tomography (OCT) technology, including handheld devices, have increased the role for this modality in diagnosing nystagmus in children.

The Leicester Grading System for foveal hypoplasia is able to help classify grades of foveal hypoplasia, which not only correlate to visual acuity^{10,11} but also with specific diagnoses,^{12,13} and can be used to help estimate future vision in young infants with nystagmus¹¹.

A typical diagnostic workflow is presented in figure 1 and forms the basis of the clinical practice in the UK.

Figure 1: Diagnostic workflow for children with nystagmus (adapted from Self et al.³)



* Where there is a clear history of prenatal drug exposure, neuroimaging may not be necessary in cases with atypical nystagmus. However, phenotyping should still be completed in all cases to seek clues of other underlying causes. All cases should be reviewed by a general paediatrician. If the history is less clear then the risk of the nystagmus being due to exposure should be considered but this should not automatically preclude further investigation according to the standard diagnostic workflow.

What are the **RED** flags when investigating a child with nystagmus?

When investigating a child with nystagmus, the range of potential underlying diagnoses can be very broad, particularly at the start of the diagnostic journey and when the child is very young. As such, diagnostic pathways prioritise identifying those underlying conditions which necessitate the most urgent investigation, treatment, referral, or management. **RED** flags can be considered those parts of the history, examination or investigations that point towards the more urgent underlying conditions. Conversely, a number of **GREEN** flags exist that point towards the less urgent underlying causes.

Table 1: List of RED and GREEN flags when investigating a child with nystagmus^{3, 14, 15}

RED flag	GREEN flag	Explanation
Acute onset after 6 months of age		Infantile nystagmus usually presents before 6/12 age. Acute onset after 6/12 age is more likely to be acquired
New onset oscillopsia and no good long-standing nystagmus history	Infrequent or no oscillopsia	Absence of persistent oscillopsia in nystagmus suggests early onset and therefore less likely to be acquired
Vertical nystagmus	Horizontal nystagmus only	Vertical nystagmus is more likely due to neurological causes or retinal dystrophy than uniplanar horizontal only nystagmus
Difference in nystagmus intensity between the two eyes		An interocular difference in intensity (incl. apparent uniocular nystagmus) makes a neurological cause more likely. Uncommon in more benign congenital forms
	Beats in the direction of gaze horizontally	Typical feature of Infantile Nystagmus Syndrome (INS) and so neurological cause less likely
	Only seen in extreme gaze positions	Typical feature of physiological end-point nystagmus
	Other ocular findings including congenital cataract, strabismus, etc.	Other ocular features suggest an 'ocular' cause rather than a neurological cause
No slow component to eye movements and therefore not nystagmus		Manifest uncontrolled eye movements besides nystagmus are more likely neurological in origin, e.g. opsoclonus
	Intermittent 'nystagmus' seen with forced convergence and no other clinic features in a well child	Voluntary nystagmus (not actually nystagmus as no slow phases) is more easily generated with convergence
Coincident neurological symptoms/signs, e.g. ataxia or headache		Neurological cause more likely
	Presence of null zone +/- anomalous head posture	A common feature in INS and rarely seen in acute onset neurological nystagmus
	Significant change in nystagmus intensity and direction with monocular occlusion	Typical feature of Fusion Maldevelopment Nystagmus Syndrome (FMNS, aka MLN, Manifest Latent Nystagmus)

No one of these **RED** or **GREEN** flags is pathognomonic for a single underlying cause. However, they can be used to direct and prioritise the urgency of the subsequent diagnostic workup.

Management

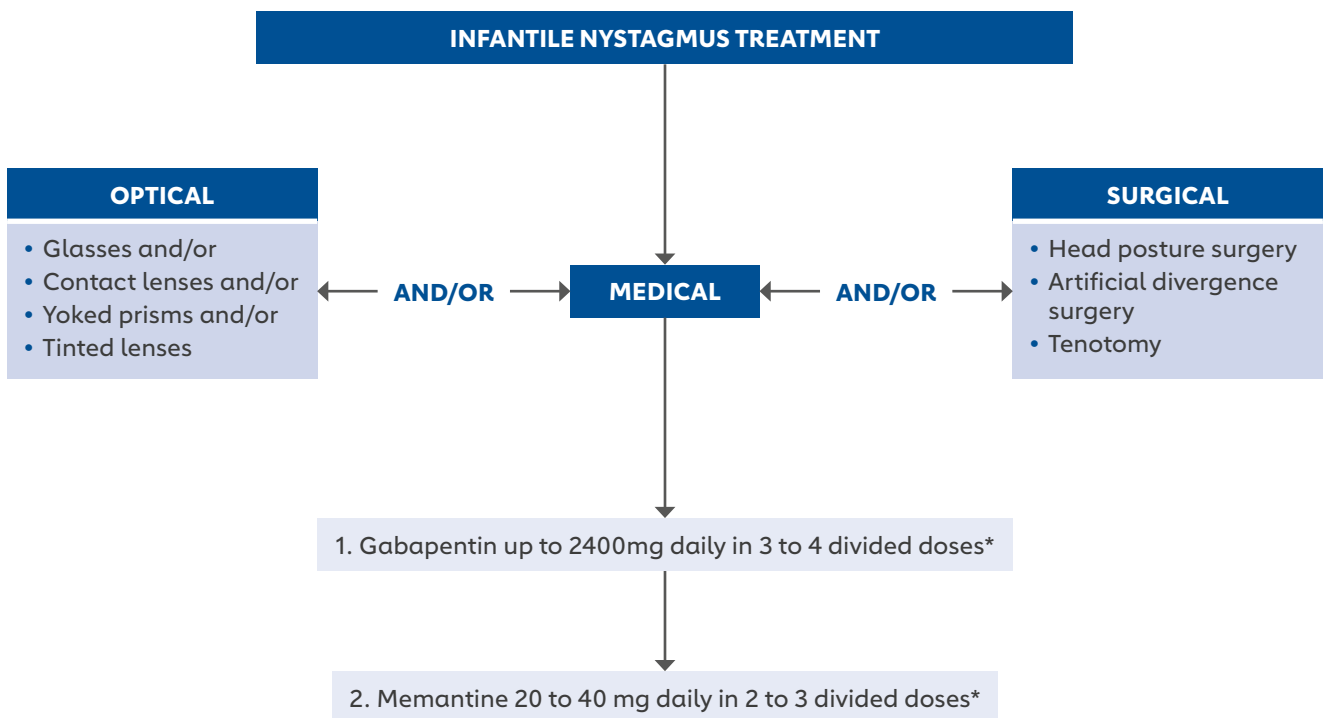
As detailed above, a broad range of underlying conditions can be associated with nystagmus, and treatments may be specific to these underlying aetiologies (e.g. retinal dystrophy, optic nerve disease, etc.).

Independent of aetiology, problems that may need to be addressed in children with nystagmus include:

- Optimisation of visual function
- Management of neck strain associated with significant anomalous head postures (AHPs)
- Improvement of ocular alignment/appearance.

Management of nystagmus includes an array of optical, medical, and surgical options as outlined in figure 2.

Figure 2: Options for management of nystagmus



* These doses are applicable to adults only.

Optical

Refractive correction using either spectacles or contact lenses is important for maximising visual function^{3,16,17}. Contact lenses, which may also dampen nystagmus through their proprioceptive effects in some patients, are especially useful in cases where there is a significant AHP and/or significant astigmatism. Tinted lenses can be beneficial in cases of nystagmus associated with photophobia. Prisms can also be incorporated into spectacles to take advantage of a null zone (the direction of gaze with minimal nystagmus) or induce artificial divergence in cases where there is a convergence null (dampening of the nystagmus and improved visual function on near viewing).

Medical

Gabapentin (up to 2400 mg in divided doses) and Memantine (up to 40 mg in divided doses) treatment have been shown to be effective in reducing nystagmus intensity and improving visual function in some adult patients¹⁸. Baclofen can be effective in congenital periodic alternating nystagmus¹⁹. These are rarely prescribed for children.

Surgical

Extraocular muscle surgery can be beneficial, both in terms of visual function and management of associated torticollis, to correct significant AHPs ($> 20^\circ$). Carrying out this surgery as soon as possible can potentially maximise visual development in children. This is achieved by shifting the null zone to a more central gaze position (i.e. Kestenbaum-Anderson procedure)³. The presence of Periodic Alternating Nystagmus (PAN), where the nystagmus in primary gaze spontaneously changes direction along with the resulting null zone, should be determined before considering surgery. AHP surgery can be combined with the correction of any associated strabismus or with artificial divergence surgery (to take advantage of a convergence null). In cases of FMNS, AHP surgery should be carried out on the fixing eye, with surgery for any residual heterotropia performed on the non-fixing eye.

Referral for long term support

Families should be referred by the clinician to the following services as soon as they are ready to receive such support:

- The local authority sensory team, in particular the QTVI (Qualified Teacher of Visually Impaired children), should assess special educational needs
- The ECLO (Eye Clinic Liaison Officer) can advise about local support groups, habilitation services, mobility training and registration as sight impaired.

The vast majority of children with nystagmus, even those with good visual acuity, experience visual impairment for real-world tasks. For this reason, most children in the UK with nystagmus are eligible for sight impairment registration through the Certificate of Visual Impairment (CVI) process. CVI registration should be considered in all children with nystagmus.

Clinicians should also consider signposting families to the following:

- The [Nystagmus Network](#), a registered charity in England and Wales, provides reliable, up-to-date information on most of the long-term needs of children and adults living with nystagmus and welcomes referrals from clinical professionals. info@nystagmusnet.org
- The [Albinism Fellowship](#) supports families in the UK and Ireland
- [Aniridia Network](#) supports families and adults in the UK and Ireland
- National vision impairment charities, such as [RNIB](#) and [Guide Dogs](#), can also provide information and support.

Referral/Discharge

Referral criteria into paediatric HES

All children with nystagmus should be referred to Hospital Eye Services. Urgent (typically on the day) referrals should be considered for children with any of the **RED** flag features (table 1):

- Acute onset after 6 months of age
- New oscillopsia and no good longstanding nystagmus history
- Vertical nystagmus
- Difference in nystagmus intensity between the two eyes
- No slow component to eye movements and therefore not nystagmus
- Coincident neurological symptoms/signs, e.g. ataxia.

Referral criteria from paediatric HES services into specialist nystagmus services

Typically, most children will be referred to a specialist nystagmus service:

- Where a suitable investigation pathway is not available locally to assist with diagnosis, e.g. OCT, ERG and VEP, genetics (referral may be for these only rather than for a specialist clinic review, according to locally agreed networks).
- Where specialist investigations (e.g. EMR) are required to assist with diagnosis/management or if a second opinion is sought.
- Where treatment options are not accessible/available locally (optical/medical/surgical)

Recommendations for discharge

Discharge from specialist centre to local HES if investigation and/or treatment pathway has been completed in the infant/younger child, following locally agreed network arrangements.

Discharge into the community from the non-specialist/specialist centre if investigation and/or treatment pathway has been completed in the older cooperative child (>8yo) or adult and a long-term management/support plan has been agreed.

Access to detailed information relating to nystagmus management and treatment should be made available to all patients/carers, ensuring efficient transition and continued management in the community setting.

List of Abbreviations

AHP	Anomalous Head Posture
CGH	Comparative Genomic Hybridization
EMR	Eye Movement Recording
ERG	Electro-RetinoGram
FMNS	Fusion Maldevelopment Nystagmus Syndrome
HES	Hospital Eye Services
INS	Infantile Nystagmus Syndrome
MLN	Manifest Latent Nystagmus
OCA	Oculo-Cutaneous Albinism
OCT	Optical Coherence Tomography
PAN	Periodic Alternating Nystagmus
VEP	Visually Evoked Potential

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