Assessment of nystagmus

The assessment of a child with nystagmus involves first of all ensuring that the observed eye movement disturbance is genuine nystagmus rather than one of the stimulating disorders, particularly opsinclusus. Rather than a rhythmic to and fro oscillation as seen in nystagmus, children with opsinclusus demonstrate bursts of rapid (saccadic) eye movements that are multidirectional (horizontal, oblique, and vertical) and often distressing to the child. While such a disturbance of eye movements may be entirely normal in the first two to three months of life, and should settle spontaneously, its persistence or appearance in an older child may denote the presence of an otherwise occult neuroblastoma and requires appropriate investigation.

Childhood nystagmus has been the subject of intensive investigation and the underlying pathophysiology has recently been reviewed. However, using a simple clinical paradigm it is possible to arrive at an accurate assessment in most cases without recourse to detailed eye movement recording. It is traditionally divided into ‘congenital’ and ‘acquired’ forms, with the term congenital being somewhat loosely applied to nystagmus appearing in the first two to three months of life. Because of this imprecise usage, a recent review has recommended abandonment of the term congenital in favour of ‘early onset nystagmus’. As yet this is not established in our literature, and will not, therefore, be used in this annotation.

The congenital form of nystagmus (incidence 1/6550 live births) may in turn be subdivided into sensory deprivation nystagmus in which a disturbance of vision is the primary defect, and congenital idiopathic nystagmus in which there is no underlying disturbance of the afferent visual pathways. Of those children presenting to a department of paediatric ophthalmology with nystagmus, the great majority will have sensory deprivation nystagmus.

Congenital nystagmus

Though the history will usually give a clear indication of early onset of nystagmus, this is not always the case and other features of congenital nystagmus may need to be identified. The nystagmus is typically uniplanar, remaining horizontal in all directions of gaze. It dampens with convergence and may increase in intensity with attempted fixation on a target. A less constant but diagnostic characteristic when present is that of inversion of the optokinetic response. Non-typical patterns of congenital nystagmus are, however, common and include vertical, oblique, and torsional movements in addition to the horizontal oscillations. These atypical patterns are particularly prominent in sensory deprivation nystagmus.

SENSORY DEPRIVATION NYSTAGMUS

A range of ocular disorders will so degrade the visual input that they prevent stable ocular fixation and lead to sensory deprivation nystagmus (table). Many are clinically obvious such as cataracts or corneal opacities, while others may be strongly suspected on the basis of the child’s history, for example retinopathy of prematurity. In some children, however, the eyes may appear grossly normal, and may remain so for several years, and it is in these circumstances that ancillary investigations are essential to identify the underlying pathology accurately. Of particular importance in this context are children with retinal disorders. Leber’s amauro-

Aetiology of sensory deprivation nystagmus

<table>
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<th>Usually clinically obvious</th>
<th>Anterior segment</th>
<th>Posterior segment</th>
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<tr>
<td></td>
<td>Corneal opacities</td>
<td>Retinopathy of prematurity</td>
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<td>Cataract</td>
<td>Destructive retinitis (for example cytomegalovirus)</td>
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<td>Anisocoria</td>
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<td>Glaucoma</td>
<td>Macula scarring</td>
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<td>Albinism</td>
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Require neurophysiological confirmation

- Leber’s amaurosis
- Achromatopsia
- Cone dystrophy
- Congenital stationary night blindness

Require investigation using imaging techniques

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<thead>
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<th>Optic nerve</th>
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<tr>
<td>Optic nerve hypoplasia</td>
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<td>Optic atrophy</td>
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CONGENITAL IDIOPATHIC NYSTAGMUS

Congenital idiopathic nystagmus can be diagnosed in a child with the typical pattern of nystagmus when clinical examination and electrophysiological testing fail to demonstrate any underlying sensory cause. It can also be strongly suspected, without resorting to extensive investigation, when the pattern of eye movements conforms to that already described, if the visual acuity is good (6/18 or better).

Congenital idiopathic nystagmus is commonly familial, and though autosomal dominant and recessive forms have been described, the commonest pattern of inheritance is X linked with some of the female carriers showing evidence of nystagmus. If the pattern of nystagmus is more atypical, a diagnosis of congenital idiopathic nystagmus must be one of exclusion, and full investigation is necessary before attaching such a diagnostic label. If electrophysiological testing is normal, the children require central nervous system imaging, preferably using magnetic resonance imaging, before the paediatrician can be certain there is no underlying pathology.

Children with known neurodevelopmental pathology, such as cerebral palsy, Down’s syndrome, etc, present a particular problem when they also have nystagmus. The great majority of children thus affected do not have nystagmus, and its presence should stimulate a full evaluation as already described. Nystagmus does not occur in children who are ‘cortically blind’ in isolation and the discovery of cataracts or optic atrophy will significantly alter a child’s visual prognosis, and may require specific treatment or specialised support to deal with the visual difficulty.

Acquired nystagmus

Nystagmus starting for the first time after the establishment of normal, stable ocular fixation has sinister connotations. Bilateral severe visual loss occurring before the age of 4 years will commonly give rise to an acquired form of sensory
deprivation nystagmus, but then the visual loss is the usual presenting complaint. However, nystagmus may be the presenting sign of neurological or metabolic disease, and the nystagmus may have important localising features. It is helpful to be familiar with those patterns of nystagmus that have localising value or that tend to indicate a specific pathology.

VERTICAL NYSTAGMUS
When the fast phase of nystagmus is directed downward (usually best identified with the eyes looking down and to the side) it is referred to as down beat and usually denotes pathology arising at the craniocervical junction. The commonest cause in childhood is the Arnold-Chiari malformation but cerebellar abnormalities and in older children syringomyelia may be responsible. Up beat nystagmus has less localising value and occurs as a toxic reaction, particularly to anticonvulsants, but in the absence of any toxic effect suggests posterior fossa pathology and has been seen in a variety of disorders ranging from demyelination to medullablastoma. See-saw nystagmus is so called because the pattern of movement resembles a child’s see-saw and involves one eye elevating and intorting, while the fellow eye depresses and extorts in a rhythmic manner. It is associated with suprasellar lesions, and in particular craniopharyngiomas.

If during attempted upgaze, particularly rapid upward movements, the eyes instead perform a beating pattern of convergence associated with globe retraction, the pattern of movements is known as convergence retraction nystagmus. We have seen this as a congenital movement disorder in a dysmorphic child with no neurological pathology, but it most commonly denotes pressure in the region of the Sylvian aqueduct and is typically seen in hydrocephalus or more rarely pineal mass lesions.

HORIZONTAL NYSTAGMUS
Monocular or grossly asymmetrical, horizontal nystagmus should alert the paediatrician to the possibility of an underlying neurological problem. The combination of an asymmetric nystagmus, abnormal head posture, and horizontal head shaking may well prove to be spasmus nutans, which behaves in a benign manner and carries a good prognosis. However, an exactly similar presentation has been seen with diencephalic tumours, and, therefore, detailed imaging is mandatory, and a benign diagnosis can only be made by observing full recovery after negative investigation.

Other forms of monocular nystagmus include that associated with severe unilateral visual loss, the nystagmus associated with internuclear ophthalmoplegia (most often demyelinating in origin, though occasionally vascular) and it may also be seen in association with ataxia and disc swelling in cerebellar and cerebellopontine angle lesions.

Periodic alternating nystagmus is a fascinating and rare form of horizontal nystagmus in which the fast phase changes direction in a cyclical manner with each cycle lasting 90 to 180 seconds. There is commonly a compensatory head posture which also changes direction. While it is often congenital in origin, once again it may be associated with craniocervical junction pathology or anticonvulsant ingestion.

Children with metabolic or degenerative neurological disorders may well develop nystagmus during the evolution of their disorder. Hypothyroidism, leukodystrophies, disordered lipid storage, etc, will often show horizontal or complex nystagmus patterns at some stage in the disease once again demonstrating the need for careful evaluation of any atypical pattern of nystagmus.

Management
Having established the diagnosis, instituted any appropriate treatment and provided genetic counselling, the final stage in assessment is to determine whether any intervention will specifically reduce the nystagmus and permit a higher level of visual acuity. The paediatrician should be aware that a number of treatments have been advocated, and while there is doubt about their efficacy, they should none the less be considered. Some (such as biofeedback) have had no extensive clinical evaluation. Others such as the surgical option have been shown to be successful in both eliminating head postures and improving vision, at least in the medium term. Pharmacological intervention has some value in adults, but the role of agents such as baclofen and benzhexol is unknown in children.

Conclusion
Children with nystagmus present infrequently but are a source of great anxiety. Proper assessment requires a combination of clinical skills, neurophysiological techniques, and neurological imaging. For this reason the initial assessment is often best carried out using a combined neuro-ophthalmic approach. Armed with an accurate diagnosis the paediatrician can offer counselling, support, and occasionally treatment to minimise the visual impact of the nystagmus.

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