

Optic Nerve Hypoplasia: Why Acuity and Nystagmus Are Not Enough—A Clinical Perspective on Functional Vision

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Abstract

Visual disability is commonly assessed through visual acuity and visual-field measurements. Although important, these measures may not fully capture how a person processes and uses visual information in everyday life, particularly in complex environments or over sustained periods.

This paper begins from the lived experience of an individual whose lifelong diagnosis of congenital nystagmus did not adequately capture the complexity of congenital optic nerve abnormality, functional visual impairment and real-world disability. It uses this case-informed analysis to examine whether congenital or early-life optic pathway disruption, particularly optic nerve hypoplasia, may contribute to later neurodevelopmental visual processing difficulties.

Current definitions of cerebral visual impairment recognise that anterior visual pathway disorders may coexist with CVI where they do not fully explain the level or pattern of impairment (Chang et al., 2024). Building on this distinction, the paper does not seek to collapse ONH and CVI into a single diagnosis but suggests that they may need to be considered within a connected developmental visual pathway. Clinically, functional difficulty warrants investigation and support without first proving which mechanism caused it.

The paper does not propose a formal research protocol but identifies a two-strand clinical research direction. The first strand would examine young adults aged 18–24 with congenital or early-life optic nerve abnormalities, using structural imaging, electrophysiological, oculomotor and functional vision assessments to investigate the status of the optic nerve and visual pathway.

This would allow consideration of whether structural and electrophysiological findings correlate not only with acuity and nystagmus, but also with functional markers such as impaired depth perception, light-induced visual disturbance, crowding, figure-ground difficulty, motion perception, spatial orientation, visual fatigue and overload in visually complex environments.

This approach would also allow investigation of whether different components of visual pathway function are affected unevenly, so that some capacities may be relatively preserved while acuity, binocular depth perception, spatial integration, motion processing or visually guided function are significantly impaired. The second strand would apply the same structural, electrophysiological and functional approach earlier in life, in children aged 5–7 with suspected or confirmed optic nerve hypoplasia. Its purpose would be to improve early recognition of likely functional visual difficulty and to develop a more reliable basis for anticipating support needs before educational and developmental disadvantage becomes entrenched.

Together, these two strands would help clarify whether acuity, visual fields and nystagmus adequately describe lived visual disability in individuals with congenital optic nerve or visual pathway abnormality. They would also support a developmentally informed model of assessment that considers potentially uneven abilities across visual input, visual processing and functional visual use.

Keywords: Optic nerve hypoplasia; nystagmus; cerebral visual impairment; integrated clinical and functional assessment; visual pathway development; educational disadvantage.

PART 1

I. Why This Paper is Relevant

This paper proposes reframing optic nerve hypoplasia, visual pathway function, and functional vision as interconnected elements within a developing visual system, rather than as separate ocular, neurological, and functional domains. It asks whether congenital or early-life optic nerve abnormality may contribute not only to reduced visual input or nystagmus, but also to later difficulties in visual processing and functional use.

The immediate clinical proposition is that a person with ONH should have functional visual difficulty investigated and supported even when its mechanism is uncertain. The separate research question is how altered optic-nerve input, associated cerebral findings, gaze control and visual experience contribute to the later functional profile. The need for support does not depend on resolving that question first.

This question also points toward a staged clinical and research approach: first, to examine how optic nerve and visual pathway abnormalities relate to later functional vision in young adults aged 18–24; and second, to examine age-appropriate findings in children aged 5–7 with congenital optic nerve abnormality.

The visual process begins when the eye captures light and converts it into neural signals. The optic nerve transmits those signals to the brain, where subcortical structures and distributed cerebral networks construct the experienced world of space, motion, depth, and meaning. Vision is therefore not simply a sensory input, but a neurological process.

Current clinical and service frameworks often fragment this process into separate domains. Ophthalmology addresses the eye and optic nerve, neurology the brain, while education and social care assess functional performance.

Between these domains are individuals particularly children, whose visual processing is unstable, effortful, and cognitively demanding, yet whose difficulties may not be adequately recognised by systems organised around separate ocular, neurological, educational, and social-care categories.

This fragmentation is particularly evident in children with optic nerve hypoplasia and those diagnosed with cerebral visual impairment. Optic nerve hypoplasia is generally understood as a congenital abnormality of the anterior afferent visual pathway, commonly assessed within ophthalmology or neuro-ophthalmology, while cerebral visual impairment is understood as a brain-based visual disorder. In practice, however, both may involve disruption within an integrated visual system, resulting in difficulty constructing a stable and meaningful perceptual world.

A staged assessment model may help bridge this gap by linking structural findings, electrophysiological evidence and functional visual assessment, rather than treating these as separate forms of evidence belonging to separate services. Such an approach would also allow investigation of variance: why some individuals with congenital optic nerve abnormality appear to show relatively preserved function in certain visual tasks, while others experience marked difficulty with visual complexity, spatial organisation, movement, fatigue or real-world navigation.

The consequences are significant. Children whose visual systems develop on degraded or inconsistent input may be assessed primarily through acuity-based measures, mischaracterised by cognitive tests that rely heavily on visual processing, or denied appropriate support on the basis that they ‘see too well’ or are unlikely to benefit. These are not failures of intent, but of classification.

This paper proposes a unifying clinical framework for investigating whether, in some individuals, early optic pathway disruption is associated with functional impairment extending into cerebral visual processing. It does not assume that impairment at one anatomical level necessarily establishes impairment at another. While the distinction between eye disease and brain disease may be clinically and administratively useful, it does not fully capture the functional reality of visual impairment in these cases, and may constrain appropriate assessment, support, and intervention.

II. Optic Nerve Hypoplasia Within the Developing Visual System

Optic nerve hypoplasia (ONH) is not solely an anatomical abnormality of the eye but may also be understood as a developmental disruption to the brain’s primary source of visual input (Garcia-Filion and Borchert, 2013). Clinically, ONH is an underdevelopment of the optic nerve and may be associated with reduced visual acuity, nystagmus, strabismus, visual-field defects and wider developmental concerns (Kaur et al., 2013; Taylor, 2007).

ONH arises prenatally and, from birth, reduces the number of retinal ganglion-cell axons available to convey visual information. Depending on its severity and associated conditions, the signal reaching the visual cortex may therefore be reduced or atypical, while associated nystagmus or impaired gaze control may introduce additional retinal-image instability. Clinical electrophysiology may help identify reduced, delayed, asymmetric or poorly formed visual responses in ONH.

Visual evoked potentials record cortical responses to visual stimulation and can provide evidence about post-retinal visual-system function, while retinal electrophysiology can help distinguish retinal from post-retinal contributions (McCulloch et al., 2010). These tests should be interpreted alongside structural, ophthalmological and functional findings rather than as providing a complete neurological account of vision.

Where visual evoked potentials are used, assessment should follow current clinical standards for VEP recording and interpretation (Šuštar Habjan et al., 2025). During early development, the visual brain is shaped by patterns of incoming sensory input. Where that input is compromised from the outset, visual development may be affected in ways that extend beyond acuity alone.

Disruption during this period may have lasting effects on visual processing, including spatial localisation, motion tracking, depth perception, object recognition and visual attention. Rather than developing under conditions of stable and reliable input, the brain may have to organise visual processing under conditions of reduced signal and increased noise. Neuroplastic adaptation in this context may support function but may also leave the individual with a visual system that is less stable, less efficient and more cognitively demanding to use.

Where higher-order visual processing is also affected the proposed model predicts that this may present functionally as difficulty stabilising visual scenes, distinguishing figure from background, constructing consistent spatial representations, and extracting meaning from complex visual environments. Individuals may be able to detect visual stimuli while experiencing disorientation, fatigue, or overload in real-world settings. By school age, such patterns may already be established, reflecting a visual system that has developed under conditions of degraded input.

In this sense, ONH may provide a developmental context in which to investigate whether early optic pathway disruption contributes to higher-order visual processing difficulties consistent with those described in cerebral visual impairment (CVI). This perspective suggests a possible developmental relationship between optic nerve pathology and cerebral visual processing impairment, while preserving the clinical distinction between them.

III. Cerebral Visual Impairment and the Functional Use of Vision

Cerebral visual impairment is widely misunderstood, not only by the public but also across medicine, education, and disability systems. It is often described as “brain-based vision loss,” yet this characterisation does not fully capture its nature. CVI is not primarily a deficit of visual acuity, nor simply the result of a focal lesion of the visual cortex. Rather, it may be better understood as a disturbance in the processing, integration and functional use of visual information (Royal College of Ophthalmologists, 2023).

In a typically developing visual system, signals from the eyes and optic nerves are transformed into spatial representations, object identities, motion trajectories, depth relationships and a stable perceptual field. These functions depend on large-scale networks linking the primary visual cortex with dorsal “where/how” and ventral “what” processing streams. CVI may arise when these networks are disrupted, atypically developed or functionally impaired.

When this transformation process is compromised through neurological injury or atypical development, the visual system may operate inconsistently and inefficiently. Individuals may have difficulty navigating complex or cluttered environments, judging depth, recognising faces or objects, managing motion, and sustaining visual attention. They may also experience rapid visual fatigue, even where standard measures indicate relatively preserved acuity. The present hypothesis asks whether, in some individuals, chronically degraded or atypical early visual input may contribute to such developmental disruption.

The defining feature of CVI is therefore not simply what the eyes detect, but how effectively the brain can organise, interpret, and use visual information. CVI is best understood as a network-level condition rather than a single-lesion disorder. While the dorsal and ventral streams may be differentially affected, the unifying feature is disruption in the integration, prioritisation, and stabilisation of visual input into a functional perceptual world.

This framework also helps explain the possible overlap between CVI and optic nerve hypoplasia. Traditional definitions often distinguish CVI from visual impairment explained by ocular or optic nerve pathology. This distinction remains clinically important, but it may also obscure functional relationships across the developing visual system. From a developmental perspective, the critical issue is not solely the anatomical site of disruption, but the quality, timing, and consistency of visual input available during early neurodevelopment.

The National Institutes of Health workshop working definition describes CVI as brain-related visual dysfunction greater than expected from coexisting ocular conditions alone (Chang et al., 2024). An optic nerve finding should therefore neither establish CVI nor preclude investigation of functional difficulties it may not fully explain. Their cause requires clinical assessment rather than assignment to one anatomical site by assumption.

IV. Possible Developmental Relationships Requiring Investigation

Neuroimaging evidence:

Neuroimaging studies of congenital disorders affecting the eye, optic nerve, and visual pathways demonstrate that early disruption of visual input may be associated with large-scale reorganisation of the visual cortex and its networks. When patterned visual signals are absent or degraded from birth, the brain does not simply receive a weaker image; it develops under altered input conditions. In some congenital visual disorders, retinotopic organisation, inter-hemispheric connectivity or higher-order visual pathways may develop atypically, with lasting implications for visual processing. Importantly, such cortical changes may occur even when the primary site of disruption lies in the eye or optic nerve rather than in the brain itself (Millington et al., 2014).

These findings establish the biological plausibility of pathway-wide developmental effects, but they do not establish that isolated ONH necessarily causes cerebral visual impairment or the higher-order difficulties considered in this paper. Building on this, ONH may be understood not simply as a reduction in visual input, but as a developmental alteration in the reliability, structure, and consistency of the signal available to the visual brain.

For the developing visual system, these conditions are significant. Neural circuits are shaped through experience-dependent exposure to patterned sensory input. When that input is degraded or variable, the resulting organisation of visual networks may reflect those constraints. Neuroplastic adaptation may support functional use of vision but can also result in systems that are less stable, less efficient, and more sensitive to noise.

Neuroplastic adaptation within this model should not be understood as wholly maladaptive. Where one visual function is impaired, an individual can learn to rely, subject to their being available, on preserved sources of visual information. For example, a person with reduced or absent stereopsis may use colour and luminance contrast, shading, texture, perspective and motion to identify changes in depth or surface level. Such strategies can support effective functioning in favourable conditions, but they do not restore stereopsis, and their usefulness depends on the quality and availability of the alternative cues. The resulting visual profile can therefore be uneven, combining substantial impairment in one domain with preserved abilities and effective compensation in another.

Several developmental pathways could contribute to the observed functional profile. One possibility is that early optic pathway disruption alters input during development and contributes to later differences in spatial mapping, motion processing, object recognition or visual attention. Neuroplastic reorganisation is a biologically plausible contributor, but its particular role in ONH has not been demonstrated here. A later functional profile resembling aspects of CVI cannot, by itself, establish that sequence or exclude associated cerebral pathology.

From a systems perspective, congenital ONH may require higher-order visual networks to operate with reduced or atypical afferent input. The proposed model is that, in some individuals, this may contribute to increased cognitive demand, perceptual instability and variation in performance across environments. ONH identifies an abnormality of the anterior afferent visual pathway, whereas CVI identifies brain-based visual dysfunction that is greater than would be expected from coexisting ocular or anterior-pathway abnormalities alone. They should therefore remain diagnostically distinct, while their possible developmental relationship is investigated.

V. Why Acuity, Cognitive Testing, and Educational Systems May Misclassify Neurovisual Impairment

Visual disability involving higher-order processing difficulties may not be adequately captured by assessment frameworks dominated by visual acuity. Because acuity may be relatively preserved, and because affected children may demonstrate intact or high cognitive ability, their functional difficulties may be underestimated or judged as insufficient to justify specialist support. This risks a category error: the assumption that the presence and severity of visual disability can be adequately inferred from acuity alone.

Visual acuity measures the ability of the visual system to resolve fine spatial detail under standardised testing conditions. It does not by itself assess how the brain separates figure from background, tracks motion, constructs spatial representations, stabilises visual scenes or manages visual attention under conditions of complexity. Higher visual-function difficulties may therefore be present independently of the severity of acuity loss (Chandna et al., 2024). A child may perform adequately on a standard acuity test while experiencing significant functional difficulty in real-world environments.

Assessment should also ask whether a task can be sustained and repeated, how performance changes with fatigue, and how much recovery it requires. Performance during a brief controlled test does not establish sustainable functional capacity throughout a school, university or working day.

A similar limitation applies to cognitive and educational assessment. Some commonly used tests, particularly the non-verbal, performance and processing-speed components of standardised intelligence tests, rely heavily on visually mediated tasks, including drawing, block design, pattern recognition, visual memory and symbol manipulation. Research involving children with CVI indicates that visual acuity, contrast sensitivity and oculomotor function can influence cognitive, cognitive-visual and academic performance (Morelli et al., 2022). Where the visual demands of an assessment are not identified or appropriately adapted, performance may therefore underestimate underlying cognitive ability or produce an uneven and potentially misleading cognitive profile. If such results are interpreted as measures of general intellectual potential, they may also generate unduly pessimistic expectations of a child's capacity for educational attainment.

Consequently, children with neurovisual processing difficulties may be misinterpreted or misclassified. A child who cannot reliably stabilise a visual scene may appear confused; one overwhelmed by visual complexity may seem inattentive; and one experiencing severe visual fatigue may be regarded as disengaged. In each case, behaviour arising from visual difficulty may be attributed primarily to cognition, attention, motivation or conduct.

A related pattern may be observed in motor and physical development. Difficulties in coordination may be attributed solely to primary motor impairment but may also reflect, or be compounded by, impaired visual-spatial processing, including reduced ability to judge distance, track moving objects or integrate visual information with body position. Dorsal-stream networks support visual-spatial attention and visually guided movement, and disruption within these functions can affect physical performance even where basic motor capacity is relatively preserved (Dutton et al., 2004).

Children with neurovisual processing impairment may not be identified through acuity-centred visual-impairment pathways, may demonstrate intact cognitive ability while performing inconsistently on visually mediated tasks, and may not fit readily within established categories of need. Although the formal SEND framework is based on educational and functional need rather than visual acuity alone, access to appropriate support depends on those needs being recognised and evidenced.

Failure to identify the visual basis of difficulty may therefore delay or limit access to specialist visual-impairment services, SEND provision and reasonable adjustments. This pattern may be understood within a developmental visual-pathway framework in which visual disability can arise from impaired processing as well as reduced acuity, and in which assessment must consider functional visual use across clinical, cognitive, educational and real-world settings.

Spectacles can correct refractive error, and magnification or larger materials can improve access to a particular task. Their provision does not necessarily address absent stereopsis, visual overload, navigation or the effort of interpreting complex information. The relevant question is whether an aid meets the person's needs in the setting and over the duration in which it must be used.

VI. Potential Scale and Public Health Implications

Optic nerve hypoplasia (ONH) is an important cause of congenital visual impairment, while cerebral visual impairment (CVI) is increasingly recognised as a major cause of visual disability in children (Rahi and Cable, 2003; Garcia-Filion and Borchert, 2013; Chang et al., 2024). Although these conditions are classified separately, both concern the functioning of the visual pathway: ONH alters afferent visual input from early development, whereas CVI concerns difficulties in the cerebral processing and functional use of visual information.

All individuals with ONH are, by definition, affected by reduced or atypical afferent visual input, although the degree and functional expression of that impairment vary. The hypothesis advanced in this paper is that, among some individuals, the developing brain's adaptation to this compromised input contributes to higher-order visual-processing difficulties beyond those adequately explained by acuity, visual-field loss or nystagmus alone.

Existing prevalence estimates do not establish how many individuals with ONH experience such additional functional visual difficulties or how frequently these difficulties overlap with current definitions of CVI. The absence of prevalence data may partly reflect clinical pathways that document acuity, visual fields and nystagmus but do not routinely assess higher-order functional visual processing. Difficulties that are not systematically investigated or consistently recorded cannot readily be counted.

Nevertheless, the developmental framework proposed in this paper and the lived experience informing it provide grounds to investigate whether the population affected by higher-order visual-processing difficulties is broader than is currently recognised. It may include individuals described as having mild visual impairment, unexplained learning difficulties, coordination problems, attentional difficulties or inconsistent educational performance despite apparent cognitive ability. These features are not specific to neurovisual impairment and should not be treated as diagnostic evidence in isolation. Their possible visual contribution requires structured clinical, electrophysiological and functional investigation.

The potential public health implications are significant. Individuals whose neurovisual processing difficulties remain unrecognised may encounter barriers to education, mobility, social participation and employment, as well as an increased risk of fatigue, reduced confidence and adverse mental-health consequences. Where the visual basis of these difficulties is not identified, support may be delayed, misdirected or withheld.

Recognising this possibility is therefore not solely a matter of diagnostic clarification. It has implications for population-level identification, clinical, educational and workplace assessment, service design and resource allocation. Research is required to establish the prevalence and range of higher-order visual-processing difficulties among people with ONH, to distinguish these difficulties from those arising through other developmental pathways, and to determine which clinical and functional assessments most reliably identify individuals requiring additional support or reasonable adjustments.

VII. The Institutional Gap

Current service structures reveal a gap in how visual impairment is conceptualised and managed across clinical and support systems. This gap does not arise from any single discipline, but from the way responsibilities are distributed across them.

The visual system is often addressed as if it comprised separate domains: an ocular or afferent visual-pathway problem for ophthalmology, and a brain-based or functional problem for neurology, education and disability services. In practice, however, vision depends on an integrated visual system of multiple interconnected pathways. When disruption occurs during development, its effects may extend beyond the site of the original abnormality to influence how visual information is processed and used.

Ophthalmological assessment appropriately focuses on structural and measurable aspects of visual function, including acuity, visual fields, retinal integrity, optic nerve structure and eye movements. ONH can be identified through examination of the optic disc, supported where appropriate by quantitative disc measurement, fundus imaging or optical coherence tomography. Detection is more straightforward, however, in pronounced or markedly asymmetrical cases than in mild, segmental or less typical presentations. Even where the optic nerve abnormality is recognised, its broader and continuing functional consequences can fall outside the scope of routine assessment, particularly where no further medical or surgical intervention is indicated.

In England, certification as sight impaired or severely sight impaired remains ophthalmologist led. Although neurological and functional evidence can inform the consultant ophthalmologist's professional judgement, certification does not itself require a structured assessment of higher-order visual function. The extent to which such evidence is sought, recorded or incorporated can therefore vary between clinicians and services.

At the same time, definitions of CVI commonly emphasise visual dysfunction that cannot be fully explained by ocular findings or abnormalities of the anterior visual pathways. Although ocular and optic nerve abnormalities can coexist with CVI, their presence may create classification uncertainty if they are assumed to explain the individual's complete functional visual profile. Consequently, higher-order visual-processing difficulties risk remaining insufficiently investigated or documented.

The result is a gap in classification and service responsibility. Individuals experiencing significant functional visual impairment in educational and everyday contexts may not be fully captured within existing diagnostic or support pathways. This gap reflects differences in how visual impairment is defined and assessed across systems, rather than an absence of need. It highlights the importance of recognising the possible continuity between early visual-pathway disruption and later functional outcomes. A more integrated framework is therefore needed: one that considers the eye, optic nerve, visual pathways and brain as parts of a single developing visual system and assesses functional impairment accordingly

VIII. Conclusion

This paper has argued that the functional consequences of ONH cannot always be adequately understood through optic nerve structure, visual acuity, visual-field loss and nystagmus alone. When visual input is degraded, atypical or unstable during early development, the brain may develop its organisation and use of vision under constraint. Among some individuals, this could contribute to lasting difficulties in visual processing, including stabilising visual scenes, managing visual complexity, judging spatial relationships and using vision efficiently in real-world environments. The resulting functional profile can share important characteristics with CVI.

Visual disability is commonly assessed through visual acuity and visual-field measurements. Although important, these measures may not fully capture how a person processes and uses visual information, particularly in complex settings or over sustained periods. A person may perform relatively well on conventional tests yet experience substantial difficulty in daily life. This gap between measured vision and functional experience can leave support needs unrecognised across the life course. It points to limitations in how visual impairment is defined, assessed and classified, rather than a failure of any single service.

Within this context, a possible developmental contribution of altered early optic-nerve input offers a testable hypothesis alongside associated cerebral pathology and other explanations. This framework recognises that disability can arise both from the quality of visual input and from how that input is processed, organised, stabilised and functionally used by the developing brain. In cases involving early visual-pathway disruption, visual disability should therefore be approached as a distributed neurosensory process rather than as a condition confined exclusively to either the eye or the brain.

This framework does not propose that ONH and CVI are interchangeable diagnoses, or that ONH necessarily results in CVI. Rather, it identifies a possible developmental relationship requiring systematic investigation and seeks to prevent the presence of an optic nerve abnormality from prematurely foreclosing examination of additional higher-order visual-

processing difficulties. Approaching the visual system as a unified neurosensory pathway has implications across clinical, educational, occupational and social domains. It could support earlier recognition of need, more appropriately directed intervention and improved long-term support.

The argument also has implications for ophthalmology-led certification. Although consultant ophthalmologists can exercise professional judgement alongside acuity and visual-field guidance, certification does not itself require structured assessment of higher-order visual function. Where appropriate functional and, when indicated, neurological assessments are absent, pathway-related and brain-based visual difficulties risk remaining under-recognised or insufficiently documented, with consequences for access to support and reasonable adjustments.

Further research is required to determine whether altered early optic-nerve input contributes to later visual-processing difficulties in some people with ONH. It should also examine whether combined structural, electrophysiological, oculomotor and functional assessment adds useful information about support needs beyond acuity, visual fields and nystagmus, and how the findings might inform assessment, certification and intervention.

PART 2

The situation described in Part 1 already demands change as a matter of clinical fairness, educational justice and social justice. The two-stage research proposal is one possible disciplined way of producing the evidence needed to support that change.

Introduction

Optic nerve hypoplasia is a congenital underdevelopment of the optic nerve within the afferent visual pathway. This paper argues that it should also be considered within a wider developmental visual-pathway framework. Where early visual input is reduced, atypical or unstable, the consequences can extend beyond visual acuity alone, affecting binocular vision, stereopsis, visual fields, gaze stability and the functional use of vision in everyday environments.

Bilateral ONH can be associated with severe visual impairment or certification as severely sight impaired, although visual outcomes vary substantially. Where the presenting or recorded diagnosis is congenital nystagmus, there is a risk that the visible eye movement becomes the dominant clinical label, while an underlying optic nerve abnormality, binocular impairment, absent or reduced stereopsis, visual-field difficulties and real-world functional limitations receive insufficient attention. Some children may consequently proceed through childhood and education without the level of investigation, certification, educational support or functional recognition that their condition requires.

Heterogeneity and causal uncertainty

ONH may occur in relative isolation or alongside septo-optic dysplasia, endocrine dysfunction, cerebral malformations or other developmental conditions (Garcia-Filion and Borchert, 2013). The same functional complaint may reflect reduced or unstable afferent input, an associated cerebral abnormality, shared developmental processes, impaired gaze control, accumulated experience, or an interaction among these. These possibilities should be documented and investigated rather than presumed to form one sequence. Where later research examines the developmental contribution of altered optic-nerve input, existing MRI findings, supplemented by further imaging where clinically appropriate, could help identify associated cerebral abnormalities, including those linked to septo-optic dysplasia. These findings would need to be considered separately when interpreting the results.

Causal uncertainty is a reason for more comprehensive investigation, not a reason to disregard demonstrated functional difficulty. Where a neurological disorder is suspected, appropriate investigation may clarify diagnosis and care; where the cause remains uncertain, a support-oriented account of what the person can do, under what conditions, and at what cost remains valuable. The young person's own report should be considered alongside clinical observations and evidence from home and education.

The Two-Strand Approach

The paper does not propose a formal research protocol but identifies a two-strand clinical research direction. The first strand would examine young adults aged 18–24 with congenital or early-life optic nerve abnormalities, using structural, electrophysiological, oculomotor and functional assessments to obtain complementary evidence about optic nerve structure, visual-pathway function, gaze control and functional visual use.

The central question is not simply whether ONH or a related early optic nerve abnormality reduces visual acuity, but whether reduced, unstable or degraded visual input reaching the brain during development can contribute to uneven patterns of higher-order visual processing and functional visual use that are inadequately explained by acuity and nystagmus alone.

Functional visual impairment can remain concealed while educational tasks are simple, visually uncluttered and individually supported. A cognitively able child may compensate sufficiently to read short passages and meet early educational expectations despite underlying visual-processing difficulties. As educational demands increase, crowded text, diagrams, visual search, copying, spatial navigation, sustained visual attention and rapid processing can expose difficulties that were previously masked. Their emergence does not necessarily represent deterioration in vision; it can reveal a longstanding impairment that earlier assessments and less demanding environments did not identify. Without reassessment at important developmental transitions, a child can enter a substantially more complex educational environment unprepared and unsupported.

Assessment Domains

Structural imaging could identify reduced optic nerve size, abnormal optic-disc morphology, thinning of the retinal nerve fibre layer or ganglion-cell complex, optic chiasm abnormalities or wider differences affecting the visual pathways.

Electrophysiological assessment could provide complementary information about retinal and post-retinal visual function. Electroretinography can help distinguish retinal dysfunction from abnormalities arising farther along the visual pathway, while visual evoked potentials characterise cortical responses to visual stimulation and can reveal reduced amplitude, delayed, asymmetric or morphologically atypical responses. VEP testing has previously been used alongside optic-disc assessment in the clinical evaluation of ONH (Borchert et al., 1995). No individual electrophysiological test, however, establishes the complete neurological or functional interpretation of vision.

Functional vision testing would be required to investigate how structural and electrophysiological findings relate to everyday visual performance. Relevant domains include stereopsis and depth judgement, contrast sensitivity, visual fields, crowding, figure-ground perception, motion perception, light-induced disturbance, spatial orientation, visual fatigue and overload in visually complex environments.

Assessment of oculomotor control and gaze behaviour would also be important because functional vision depends not only on visual information reaching the brain, but on how gaze is directed to gather and update that information. A person can have difficulty maintaining fixation, shifting gaze, scanning complex scenes, following movement or coordinating visual attention under conditions of fatigue or visual overload. Assessing these functions would help clarify whether everyday visual difficulty reflects impaired visual input, higher-order visual-processing difficulty, inefficient gaze control or an interaction between these factors.

From Assessment to a Two-Stage Proof-of-Concept Pilot

The preceding section has argued that structural imaging, electrophysiological assessment, oculomotor assessment and functional vision testing should not be treated as separate or competing forms of evidence. In individuals with congenital or early-life optic nerve abnormalities, these forms of evidence need to be considered together to investigate relationships among optic nerve structure, visual-pathway function, gaze stability and everyday visual use.

The central proposition is that some individuals with congenital or early-life optic nerve abnormalities experience functional visual difficulties that are not adequately explained by acuity, visual-field loss or the visible presence of nystagmus alone. The wider objective is not simply improved diagnostic classification, but better recognition of functional need.

This paper does not propose a complete research protocol. It suggests that the next step could be a limited two-stage proof-of-concept pilot examining whether combined clinical and functional assessment adds useful information about functional needs beyond conventional diagnostic labels and acuity-centred assessment.

The proposed work would proceed in two stages: first, identifying potentially relevant markers in young adults aged 18–24; and second, examining age-appropriate counterparts in children aged 5–7.

Older participants can demonstrate the later functional consequences of congenital or early-life optic nerve abnormality, including cases in which the childhood condition was described principally as “nystagmus.” Younger children represent the stage at which earlier recognition and support could reduce the risk of educational and social disadvantage becoming embedded.

The pilot would neither treat ONH, nystagmus and related congenital optic nerve abnormalities as wholly separate from brain-based vision nor assume that ONH and CVI are interchangeable. It would investigate whether disruption of afferent visual input during development is associated, among some individuals, with a recognisable pattern of functional visual difficulty extending beyond that explained by acuity, visual-field loss and nystagmus.

Relevant difficulties could include impaired stereopsis and depth judgement, crowding, visual fatigue, difficulty within visually complex environments, spatial uncertainty, light-induced disturbance and inefficient functional use of vision. The pilot would examine whether these difficulties correlate with structural, electrophysiological and oculomotor markers and whether the resulting evidence could help identify children requiring additional assessment or support before avoidable disadvantage occurs.

Stage 1: Young Adults Aged 18–24

The first stage would examine young adults aged 18–24 with congenital or early-life optic nerve abnormalities, including confirmed or suspected ONH and related optic nerve abnormalities or injury. This group is important because participants are old enough for longer-term functional consequences to have become apparent across education, mobility, independence, transition to higher education, employment preparation and social participation. At the same time, their childhood clinical and educational histories may be more accessible than those of older cohorts, allowing examination of relationships among early clinical description, support received or denied and later functional outcomes.

Stage 1 would seek candidate markers across several domains:

Structural findings, including optic-disc appearance, optic nerve size, retinal nerve fibre layer thickness and ganglion-cell complex findings.

Electrophysiological findings, including reduced amplitude, delayed, asymmetric or morphologically atypical visual responses.

Oculomotor and gaze findings, including nystagmus, fixation instability, gaze control, scanning behaviour and visual search.

Binocular and functional findings, including reduced or absent stereopsis, crowding, visual fatigue, difficulty with sustained reading, navigation and visually complex environments.

Life-course outcome evidence, including education, access to support, educational transitions, independence, mobility, assistive technology and any disparity between clinical description and lived visual disability. In this context, “nystagmus” should be interpreted carefully. The issue is not that nystagmus was necessarily identified incorrectly, but whether it sometimes functioned as the dominant visible label for ocular-motor instability associated with an underlying optic nerve abnormality without adequately explaining the wider functional disability. Nystagmus can describe what is observed without necessarily explaining the person’s complete visual profile.

Stage 1 would therefore examine whether young adults with congenital or early-life optic nerve abnormalities demonstrate recognisable relationships among optic nerve structure, electrophysiological pathway function, oculomotor instability and real-world functional visual difficulty.

Stage 2: Children Aged 5–7

The second stage would apply the candidate markers identified or refined in Stage 1 to children aged 5–7 during the early primary-school years. Children within this age range are generally better able than infants to participate in a wider range of structural, electrophysiological, oculomotor and functional assessments. Early educational demands also provide opportunities to observe reading, handwriting, classroom navigation, visual attention, confidence, peer participation and adaptation to the school environment.

The selection of this age group does not assume that every relevant difficulty will already be visible. Cognitively able children can compensate successfully while tasks remain simple or visually uncluttered. The purpose would be to determine whether the combined assessment identifies functional vulnerability before increasing educational complexity exposes it through underachievement, fatigue, distress or loss of confidence.

Stage 2 would therefore examine whether candidate structural, electrophysiological, oculomotor and functional markers can contribute to an early functional-risk profile. Such a profile would not constitute a deterministic prediction or a diagnosis of CVI. It would identify areas requiring observation, further assessment or anticipatory support, including reading load, crowding, visual fatigue, classroom navigation, board or screen access, visual search, mobility, environmental complexity and the use of vision in unfamiliar settings.

The intended advantage would be prospective rather than reactive support planning. A child should not have to fail, fall behind or become exhausted before their functional visual needs are investigated and recognised.

Intervention and Transition Objective

A further objective would be to determine whether the functional-risk profile can be translated into practical assessment, intervention and support planning.

The immediate proof-of-concept question would concern support during early primary education. If the approach proved informative, it could provide a basis for planned reassessment at later educational and life transitions, including:

preparation for transition from primary to secondary school;

preparation for college or sixth form;

transition to university or other higher education;

transition into employment, supported employment or adult independence.

At each transition, assessment and support planning should consider not only acuity, but also visual fatigue, crowding, reading load, navigation, visual search, screen use, mobility, environmental complexity, assistive technology, reasonable adjustments and the child or young person's own account of functional difficulty. For younger children, parental observations and evidence from educational professionals would also be relevant.

Reassessment at these transition points would not require indiscriminate repetition of the complete testing battery. It would update the clinical and functional domains relevant to changing demands, emerging difficulties and current support needs. The aim would be to prevent support from being delayed until difficulty is already expressed through underachievement, distress or loss of confidence. Instead, the assessment profile would help anticipate need and enable support to be adjusted before disadvantage becomes entrenched.

Participant Profile

The pilot would focus on participants with congenital or early-life optic nerve abnormalities, including confirmed or suspected ONH and other clearly defined developmental abnormalities or early injuries affecting the optic nerve. Individuals whose childhood condition was described as congenital or infantile nystagmus could be included where this was used in its historical or descriptive sense: as a visible ocular-motor abnormality potentially associated with an underlying optic nerve abnormality, rather than as a complete explanation of the visual disability. A historical diagnosis of nystagmus alone would not establish eligibility; current clinical characterisation would be required.

For an initial proof-of-concept pilot, it could be appropriate to begin with participants without major unrelated neurological, developmental or acquired conditions capable of independently accounting for the functional visual profile. This would make it easier to investigate whether a combined marker pattern is associated with optic nerve or anterior visual-pathway abnormality. Such selection would, however, limit the generalisability of the findings, and coexisting conditions should be documented rather than assumed to be irrelevant.

Recruitment should seek reasonable diversity in sex, severity of visual impairment, unilateral or bilateral involvement and previous access to support. Exact balance within a small pilot should not take precedence over clearly defined eligibility and adequate clinical characterisation. Without comparison groups, the initial pilot could examine feasibility and relationships within the participant cohort but could not establish that any resulting marker pattern is specific to ONH. A later controlled study could include participants with infantile nystagmus without identified optic nerve abnormality and appropriately matched participants without known visual-pathway impairment.

Expected Output

The primary output would be a functional visual profile, not a new diagnostic category. It would describe strengths and difficulties, conditions that alter performance, endurance, repeatability and recovery, useful adaptations and any questions requiring further clinical investigation. For young adults, the profile would describe relationships among early visual-pathway abnormality, structural and electrophysiological findings, gaze control, functional visual performance and later educational, mobility and independence outcomes.

For children aged 5–7, the same assessment framework could produce a provisional, support-oriented functional-risk profile. This might indicate that acuity alone does not adequately represent possible difficulty with crowding, visual fatigue, reading load, classroom navigation or visually complex environments. Rather than predicting an individual child's outcome with certainty, the profile would identify areas requiring observation, further assessment or anticipatory educational adjustment.

Later Follow-Up and Expansion

If the proof-of-concept pilot identifies clinically meaningful candidate markers of functional visual risk, prospective follow-up at important developmental transitions would be justified.

Assessment at approximately 11, 16 and 24 years could examine whether early structural, electrophysiological, oculomotor and functional markers remain associated with—or help anticipate—visual needs as educational, social, occupational and environmental demands change.

The purpose would not be to repeat every test routinely. Reassessment would update the domains relevant to the person's developmental stage, changing circumstances and emerging functional difficulties. It would enable examination of functional vision during transition to secondary education, public examinations, further or higher education, employment, mobility and independent adult life.

The project could subsequently be expanded through additional age-banded cohorts. These could include children aged 8–11, to examine later primary-school demands; young people aged 12–16, to examine secondary-school visual load, examinations, mobility and social complexity; post-16 students, to examine transition to college, training or employment; and older adults, to investigate longer-term life-course outcomes. The first pilot should nevertheless remain deliberately limited.

The immediate purpose is to test the signal: whether combined structural, electrophysiological, oculomotor and functional assessment adds information about functional needs that conventional diagnostic labels and acuity-centred assessment may miss.

Conclusion

This paper has argued that congenital or early-life optic nerve abnormalities should not be understood solely by reference to optic nerve structure, visual acuity, visual-field loss or the visible presence of nystagmus. These measures remain important, but they may not fully explain the functional visual difficulties experienced by some individuals whose visual systems developed under conditions of reduced, unstable or degraded input.

The central issue is developmental as well as anatomical. Vision is constructed in the brain. Where the developing visual brain receives compromised input from the beginning of life, its organisation and functional use of vision may develop under constraint, alongside any associated cerebral or developmental conditions. The observed profile can be uneven: some visual capacities remain relatively preserved, while stereopsis and depth judgement, crowding, spatial orientation, visual endurance, visually guided movement, light tolerance, motion processing or performance in complex environments are substantially impaired.

The proposed two-stage proof-of-concept pilot is not intended to create a new diagnostic category, to assume that ONH and CVI are interchangeable, or to claim that ONH causes CVI in every case. Its purpose is more limited and practical: to examine whether combined structural, electrophysiological, oculomotor and functional assessment adds useful information about functional needs beyond acuity, visual fields and nystagmus.

Stage 1 would examine young adults aged 18–24 to identify candidate markers and investigate their relationships with later educational, functional and life-course outcomes.

Stage 2 would evaluate age-appropriate counterparts of those candidate markers among children aged 5–7. Recognition at this stage could enable further assessment and support before avoidable educational and social disadvantage becomes entrenched. If clinically meaningful markers emerge, prospective follow-up at key transition points—including approximately ages 11, 16 and 24—could examine whether early findings remain associated with, or help anticipate, changing support needs as educational, social, occupational and environmental demands increase.

The wider aim is not simply improved diagnosis, but improved recognition of functional need. A child should not have to fail before support is considered. A young person should not have to be misclassified before their visual difficulty is believed. An adult should not have to carry the consequences of childhood under-recognition before the limitations of acuity-centred assessment are questioned.

The proposed pilot is therefore both a clinical project and a social-justice project. It asks whether existing systems can move beyond considering only what the eye can resolve and examine how the person uses vision in the real world. If the approach proves informative and is supported by subsequent validation, it could contribute to a more integrated model in which optic nerve structure, visual-pathway function, eye-movement control and lived visual performance are considered together. Within such a model, functional visual difficulty should not be dismissed or minimised merely because it has not yet been adequately investigated or readily classified.

Declarations

Limitations

This paper proposes a hypothesis rather than a settled conclusion. It does not claim that optic nerve hypoplasia causes cerebral visual impairment in every case, or that all individuals with congenital optic nerve abnormality experience the same functional difficulties. Its purpose is to consider whether combined structural, electrophysiological, oculomotor and functional assessment may identify needs that conventional diagnostic labels and acuity-based assessments overlook.

Ethical statement

This proposal rests on a principle of clinical and educational fairness: functional visual need should not be dismissed merely because it is difficult to classify. Where congenital visual-pathway abnormality is present, systems should investigate reported functional difficulties before minimising or dismissing them, and support should not depend on a child first being allowed to fail.

Ethics approval and consent to participate

Not applicable. This manuscript presents a de-identified, case-informed clinical, policy and systems analysis. It does not report a research study, involve the collection of new research data, describe a clinical intervention or include research procedures involving human participants.

Consent for publication

The manuscript contains no identifiable personal information. Any case-informed material has been sufficiently de-identified to protect the identity of the individual concerned.

Data availability

Not applicable. No datasets were generated or analysed for this manuscript.

Competing interests

The author declares that there are no competing interests.

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