Eye (2004) 18, 1038–1048

www.nature.com/eye

CAMBRIDGE OPHTHALMOLOGICAL SYMPOSIUM

Congenital disorders of the optic nerve: excavations and hypoplasia

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Abstract

The principal congenital abnormalities of the optic disc that can significantly impair visual function are excavation of the optic disc and optic nerve hypoplasia. The excavated optic disc abnormalities comprise optic disc coloboma, morning glory syndrome, and peripapillary staphyloma. Optic nerve hypoplasia manifests as a small optic nerve, which may or may not be accompanied by a peripapillary ring (the double ring sign). In addition, the optic disc cupping, which occurs as a sequel to some cases of periventricular leucomalacia, can arguably be classified as a type of optic nerve hypoplasia. All of these conditions can be unilateral or bilateral and can impair visual function mildly or severely. It is essential that children with poor vision due to any of these conditions are managed by treating refractive errors, giving occlusion therapy in selected cases, and optimising the conditions at home and at school in an attempt to ensure that impaired vision does not impede development or education.
Eye (2004) **18,** 1038–1048.
doi:10.1038/sj.eye.6701545

Keywords: optic nerve; congenital; hypoplasia; coloboma

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Received: 4 September 2003
Accepted: 4 September 2003

Introduction

Congenital anomalies of the optic disc are fortunately rare, and in many cases either unilateral or asymmetrical involvement means that functional vision is not significantly impaired. However, children with poor vision due to these conditions are not uncommonly seen in paediatric practice and accurate diagnosis and good management are essential.

Excavations of the optic disc

Optic disc coloboma

Definition

Optic disc coloboma (Figure 1) comprises a clearly demarcated bowl-shaped excavation of the optic disc, which is typically decentred and deeper inferiorly.

Aetiology

Coloboma of the optic disc is thought to result from abnormal fusion of the two sides of the proximal end of the optic cup.[1] The condition can occur in association with multiple congenital abnormalities indicative of an insult to the developing foetus during the sixth week of gestation.[2]

Optic nerve coloboma may occur sporadically or be inherited with an autosomal dominant inheritance.[3] It has recently been shown to be associated with PAX2 gene mutations as part of the renal-coloboma syndrome.[4–7] Expression of PAX2 is restricted to cells of astrocytic lineage both during retinal development and in adulthood. Using immunohistochemistry, it has been found that adult retinal cells with the

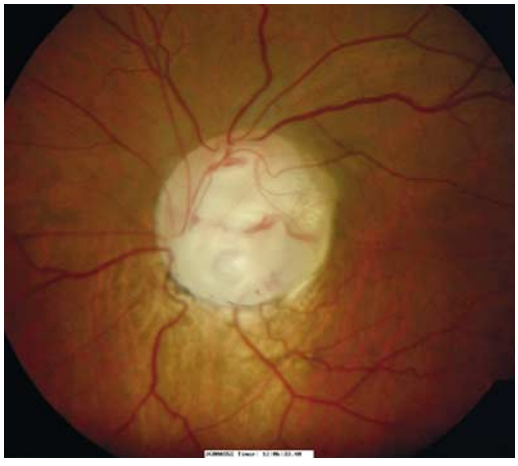

Figure 1 Coloboma of the optic nerve head.

antigenic phenotype present in mature perinatal astrocytes are found only in the region surrounding the optic nerve head, and that astrocyte precursor cells expressing PAX2 are found in a small region surrounding the optic nerve during early development. It has been argued that these findings suggest that coloboma formation may be associated with impaired astrocyte differentiation during development.[8]

The association of optic nerve coloboma with dysgenesis of the internal carotid artery and transphenoidal encephalocele with hypopituitarism has led to the suggestion that the link between these malformations is abnormal neural crest cell development.[9]

Clinical features

Unilateral and bilateral optic disc colobomata occur with similar frequencies.[10] The coloboma occupies the lower part of the optic nerve head. The neuro-retinal rim is absent inferiorly but is usually identifiable superiorly. In cases in which the adjacent inferior retina and choroid are deficient, microphthalmia may also be evident.[11]

Visual acuity is reduced to varying degrees. Careful analysis of the photographic appearances of colobomata involving the optic nerve has shown that the only feature that relates to visual outcome is the degree of foveal involvement by the coloboma.[12] The size of the coloboma, the colour of the optic nerve, and the presence of subfoveal pigment change are not related to visual outcome. Significant refractive error and anisometropia are common.[12]

Progressive optic nerve cupping and neural rim decrease have been documented in a patient with bilateral autosomal dominant optic nerve colobomas with no evidence of raised intraocular pressure and remarkably, no progressive visual field loss.[13]

Circumferential intrascleral smooth muscle has been observed histologically[14] and may account for the rare observation of spontaneous contractility of the colobomatous optic disc.[15]

Complications

A small proportion of cases are associated with cysts arising from the optic nerve sheath, which communicate with the subarachnoid space.[16] Rarely such cysts can enlarge and lead to compressive optic neuropathy.[17]

Peripapillary choroidal neovascularisation has been described in association with optic nerve coloboma.[18] Retinal detachment is also a recognised complication, and remarkably, spontaneous re-attachment may occur.[19–21] The source of the subretinal fluid is not known but could derive from fluid entering the retrobulbar space from surrounding orbital tissue, or from the choriocapillaris, or from CSF. In contrast to retinochoroidal colobomas, rhegmatogenous detachment is probably not a recognised association.[11]

Contrary to what is commonly taught, basal encephalocele is a rare association with optic nerve coloboma[11] (in contrast to the more common association with morning glory syndrome).

Associations

There is a wide range of associations. These have been reviewed by Brodsky[11] and include the CHARGE association (coloboma, choanal atresia, congenital heart disease, and multiple other abnormalities), Walker–Warburg syndrome, Goltz focal dermal hypoplasia, Aicardi syndrome, Goldenhar syndrome, and linear sebaceous naevus syndrome. More recently, associations with Dandy Walker malformation[22] and renal coloboma syndrome (with a mutation of PAX2 transcription)[4–7] have also been described.

Treatment

A trial of patching may result in improvement of vision in the child presenting early in life, and optimal refractive correction may be indicated.

Morning glory anomaly

Definition

The morning glory optic disc (Figure 2) anomaly is a congenital optic disc dysplasia in which a conical excavation of the posterior fundus includes the optic disc and is filled with glial tissue. The term reflects the morphological similarity to the flower of the morning glory plant.[23]

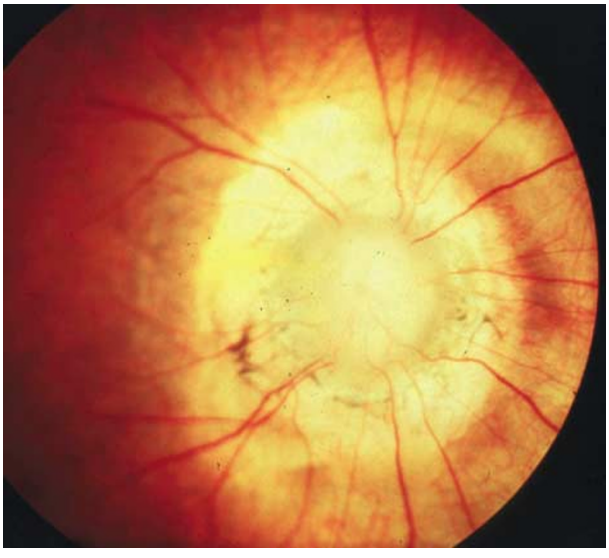

Figure 2 Morning glory syndrome (reproduced with permission from the British Journal of Ophthalmic Photography).

Aetiology

Morning glory syndrome is ostensibly a sporadic condition but it has been described occurring in isolation in a parent and child.[24]

The pathogenesis of the condition is unknown.[11] One hypothesis argues that the condition results from failure of closure of the foetal fissure and that it is a variant of optic nerve coloboma. Alternatively, a primary mesenchymal abnormality has been postulated on the basis of the glial tuft, the scleral and vascular abnormalities, and the finding of adipose and smooth muscle tissue around the terminal optic nerve. An alternative argument is that the symmetrical excavation of the optic nerve head, which characterises the disorder, is suggestive of dilation due to dysgenesis of the terminal optic stalk, which fails to close, leading to persistent excavation of the optic nerve head. The central gliosis and the vascular pattern suggest primary neuro-ectodermal dysgenesis.[10]

Morning glory syndrome is more common in female population and (in the USA) occurs less commonly in black people.[10]

Clinical features

Morning glory syndrome is usually unilateral and is characterised by a funnel-shaped, excavated optic disc (which may even include the macula) with central glial tissue surrounded by chorioretinal pigmentary disturbance. The central glial material may be either elevated or recessed.[10,11] The retinal vessels are increased in number and appear to arise from the disc margin from where they tend to run to the peripheral retina in a straighter course than usual. Fluorescein angiography has revealed arteriovenous communications near the optic disc.[25]

The visual acuity is usually reduced, being in the region of count fingers to 6/60, but it can be associated with no perception of light. Rarely the acuity can be as good as 6/6.

The remarkable phenomenon of contractile movements of the optic disc has been reported on a number of occasions. In one reported case, phasic contraction was observed by scanning laser ophthalmoscopy over 2-s periods of contraction and 20-s periods of recovery. The contractile movement was not related to exposure to strong light or the Valsalva manoeuvre but it seemed to be induced by massage of the eyeball.[26] In another reported case, both the contractions and the periods of relaxation lasted for 4-s, occurring between three and five times a minute.[27] Histologically, a cuff of smooth muscle tissue has been identified within the terminal optic nerve and this may account for the observed movement.[28,29]

Associations and complications

Morning glory syndrome tends to be an isolated condition and does not occur as part of a multisystem disorder. It can however be associated with transphenoidal encephalocele and hypopituitarism.[30,31] Patients with this condition tend to have a wide head and hypertelorism associated with a flattened nasal bridge. The encephalocele bulges into the nasopharynx where it can obstruct respiration. Incorrect identification and management of this pathology as a nasal polyp has been reported to lead to a fatal outcome.[32]

Rhegmatogenous retinal detachment (which can now be successfully treated) can be associated with a break at the optic disc margin[33] or within the optic disc cup,[34] and may occur in about one-third of cases.[11] Remarkably, optic nerve fenestration has been reported to lead to resolution of serous retinal detachment, suggesting that the source of subretinal fluid can be CSF.[35] Spontaneous resolution of retinal detachment has also been reported.[36]

Subretinal neovascularisation both under the peripapillary retina[37] and under the fovea[38] is a recognised complication.

Treatment

When the condition is first seen in young children, a trial of occlusion is warranted but should not be continued if improvement in vision does not take place.

Peripapillary staphyloma

Definition

Peripapillary staphyloma consists of a deep excavation of the area of the fundus surrounding the optic disc (Figure 3).

Epidemiology

The condition is sporadic, very rare, usually unilateral, and is not inherited.

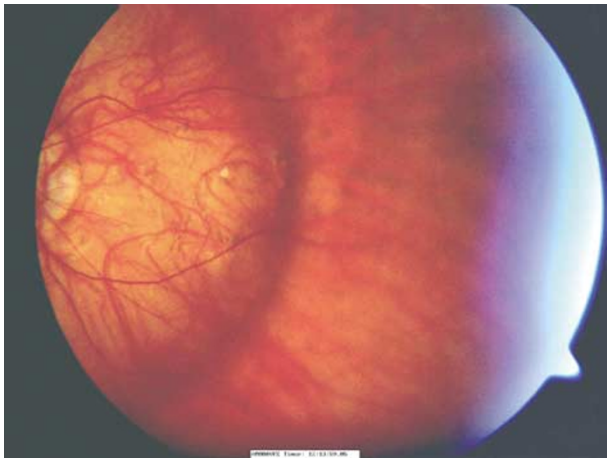

Figure 3 Peripapillary staphyloma.

Aetiology
The aetiology is unknown. It appears to arise as a sequel to disturbance of scleral development at about 20 weeks gestation, perhaps arising as a consequence of the new development of intraocular pressure causing scleral herniation at this stage of development.[10]

Clinical features
The optic nerve may look normal or manifest temporal pallor but it is situated within an excavated defect of the sclera lined by retina and choroid, which tend to be atrophic in appearance. Like optic nerve coloboma and morning glory syndrome, spontaneous contractility and relaxation of the staphyloma has been observed.[39,40]

Vision is usually markedly reduced in the affected eye but has occasionally been reported as near normal.[39] A centrocaecal scotoma may be present in eyes with reduced visual acuity. Myopia or emmetropia most commonly accompanies the condition.

Associations
In most reported cases, peripapillary staphyloma occurs in isolation and is unilateral.[41] Two out of six patients with frontonasal dysplasia in association with basal encephalocele were found to have either unilateral or bilateral peripapillary staphyloma.[42] Both facial capillary haemangioma[43] and linear nevus sebaceous syndrome are rare associations.[44]

Optic nerve hypoplasia

Definition
Optic nerve hypoplasia (ONH) is seen ophthalmoscopically as an abnormally small optic nerve head. Recently, the definition has been extended to include cases of optic nerve head cupping, which accompany periventricular leucomalacia (PVL). In both conditions, there are fewer fibres in the optic nerve from the time of birth.

Epidemiology
ONH is among the three leading causes of blindness in children in the United states. The other causes are cerebral damage and retinopathy of prematurity.[45]

Aetiology
In most cases, ONH is a sporadic occurrence for which there is no identifiable cause. There are, however, a number of associations, which are listed in Table 1. Supranormal regression of axons in the optic nerve

Table 1 Disorders associated with ONH (conditions listed in an alphabetical order)

Condition	*Comment*	*Reference*
Aicardi syndrome	Case report	106
Anticonvulsants in pregnancy	Seven cases	107
CHARGE association	Found in four out of 50 patients	108
Distal 5q deletion syndrome	Multiple abnormalities present	109
Dominant inheritance	Five individuals in four generations	67
Duane's retraction syndrome	Isolated association	110
Partial deletion of chromosome 6p	Multiple abnormalities present	111
Chromosome 7(q22→q34) and 7(q32–34) interstitial duplication	Multiple abnormalities present	112
		113
Chromosome 17 interstitial deletion	Multiple abnormalities present	71
Ethanol toxicity	Causes selective loss of small-diameter myelinated optic nerve axons (in animal models)	114
		51
Frontonasal dysplasia	Seen in seven of 23 patients with dysplasia	115
Goldenhar–Gorlin syndrome	Hypoplasia and optic nerve agenesis described	116
Idiopathic growth hormone deficiency	MRI study, pituitary gland defects common. ONH in 9%	117
Isotretinoin toxicity	Taken during pregnancy	118
Nevus sebaceous of Jadassohn	Cutaneous phacomatosis with malignant potential	119
Maternal diabetes mellitus	ONH is an isolated defect associated in general with good visual acuity	120
Muscle eye brain disease	Autosomal recessive condition Chromosome 1p anomaly	121
Orbital haemangioma	Association of orbital haemangioma lens coloboma, and ONH	122
Periventricular leucomalacia	Typical ONH or marked cupping of the optic discs can be seen. May be due to retrograde transynaptic degeneration	58, 59
Suprasellar teratoma	Case report	123
Valproic acid toxicity	SOD and other features	124

(rather than a primary failure of differentiation) is thought to be the principal mechanism of development of ONH.[46] Histopathologically, the lack of degenerated neurons in ONH is indicative of axonal loss due to apoptosis during development.[47]

Risk factors for ONH include young maternal age, first parity, maternal smoking, and preterm birth and its complications.[48] ONH is a known feature of foetal alcohol syndrome in both man[49–51] and experimental animal models.[52,53] Superior segmental ONH has been found in three of 34 children born to mothers with type I diabetes.[54]

ONH can occur in association with lesions at any site in the developing visual system[55,56] particularly in subcortical (rather than cortical) damage.[57] Loss of periventricular white matter due to PVL is commonly associated with optic disc anomalies.[58,59] As a rule, PVL of early onset is associated with small optic discs, whereas PVL of later onset is more likely to be associated with optic disc cupping. It has been hypothesised that retrograde trans-synaptic degeneration occurs when the periventricular visual pathways have been damaged, and that the pattern of optic nerve pathology that results depends on the timing of the damage.[58,59]

In a small but significant proportion of patients with ONH (both with and without associated brain abnormalities), either subnormal or negative like electroretinograms have been recorded. It has been argued that these findings may indicate that in a subset of cases with ONH and nystagmus, the retinal pathology may have been primary[60] or that trans-synaptic degeneration beyond the ganglion cell layer may take place.[61]

Septo-optic dysplasia (SOD) is now considered to comprise any combination of ONH, pituitary gland hypoplasia, and midline abnormalities of the brain. Recent studies have shown that in SOD, key mutations have been identified in Hesx-1, a protein that is involved in the mediation of normal development of the forebrain and the eyes during embryogenesis.[62]

In the embryonic mouse, optic nerve formation involves interaction between netrin-1, which is an axon guidance molecule on neuroepithelial cells at the optic disc, and the receptor for netrin-1 found on retinal ganglion cell axons. Deficiency of either of these proteins leads to deficits in retinal ganglion cell pathfinding, resulting in ONH and deficits in hypothalamic development.[63,64]

ONH has only rarely been reported in siblings,[65] identical twins,[66] or running in a family.[67]

Clinical features

A peripapillary ring around a small optic disc is the hallmark of ONH, but is not always present.[68] Biomicroscopy may reveal a pathological thinness of the nerve fibre layer.[69] ONH may be associated with tortuosity of the retinal vasculature.[70] This feature has in one study also been shown to be associated with the development of endocrinopathy.[70] A persistent grey appearance has also been described.[71]

The ratio of the distance between the centre of the optic disc to the centre of the macula (DM) and the mean optic disc diameter (DD) (ie the DM to DD ratio) provides an index of the size of the optic disc on clinical examination and on photography[72,73] (Figure 4). In one study, a ratio of 2.94 provided a one-tailed upper population limit of 95%.[74] For practical purposes, a DM : DD ratio of 3.00 should lead to serious consideration of the diagnosis[73] and a value of 4.00 probably accords a definitive diagnosis.[75] The visual acuity is associated with the DM : DD ratio. In one study of 19 children with ONH, all eyes with a DM : DD ratio of more than 3 had reduced visual acuities but all those with a ratio of less than 3 had normal acuities.[76] In another study, however, in 75% of bilateral cases, the eye with the relatively smaller optic disc was paradoxically found to have a better visual acuity than the fellow eye.[74] Both the degree of afferent pupillary dysfunction[76] and reduction in brightness sense[77] are also related to the level of acuity.

When the condition is asymmetrical, a relative afferent pupil defect adds weight to the diagnosis.[69] As one would expect, abnormality of the visual evoked potential is typically observed but the photopic and scotopic ERG tend to be normal.[78]

ONH is commonly asymptomatic and may first be detected by identification of visual field loss or observation of the optic nerve head. Segmental ONH,

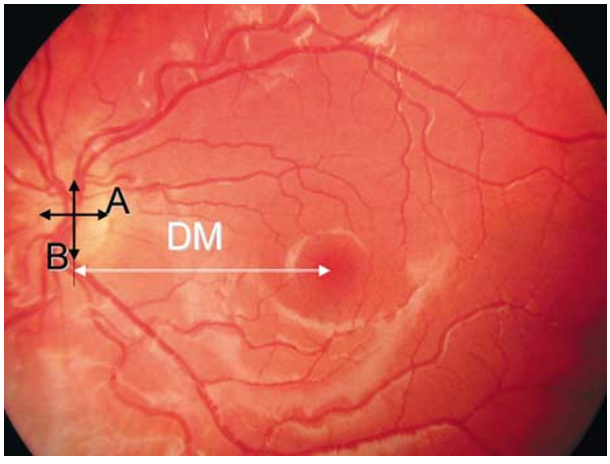

Figure 4 ONH. The mean disc diameter (DD) is (A + B)−2. The perpendicular distance between the centre of the disc and the macula is shown as DM (the distance between the centre of the disc and the centre of the macula). When the ratio of DM to DD is greater than 3, ONH is suspected, and when it is greater than 4, ONH is very likely.

which is commonly superior (causing a lower visual field defect),[79] can be identified by clinical observation, red free photography,[80] or by optical coherence tomography.[81]

Reduced optic nerve diameter (from the normal range of 4–4.5 mm) measured by ultrasound has been described, and the diameter of the optic canal may be reduced.[82] In addition to demonstrating associated intracranial disorders, magnetic resonance imaging of the intracranial visual pathways may demonstrate reduced size of the optic nerves[83] and diffuse chiasmal hypoplasia, which can lend considerable weight to the diagnosis of ONH.[84]

Associations

Magnetic resonance imaging has revealed coexistent CNS abnormalities in 30 out of 40 patients with ONH.[85] These authors describe a number of different features.

1. Isolated ONH.
2. Absent septum pellucidum. (This anomaly has not been found to be associated with significant intellectual, behavioural, or neurological dysfunction.[86])
3. Posterior pituitary ectopia (commonly associated with endocrine dysfunction).
4. Migrational anomalies in the cerebral hemispheres (for example, thinning of the corpus callosum, which is predictive of neurodevelopmental problems).

Other associated brain abnormalities include porencephaly, schizencephaly, intracranial arachnoid cyst, and intracranial epidermoid cyst.[56,87,88]

We have seen a number of infants presenting with neonatal hypoglycaemia who have later been shown to have SOD with pituitary insufficiency. ONH with an absent septum pellucidum has the highest incidence of multiple pituitary endocrinopathies[89] and of neonatal hypoglycaemia,[90] which may be associated with neonatal cholestatic jaundice.[91] Growth hormone deficiency, adrenocorticotrophic hormone deficiency, thyroid-stimulating hormone deficiency, and diabetes insipidus have all been described.[92] Identification of hypopituitarism to identify low cortisol is essential. Coma and convulsions due to hypoglycaemia have followed uncomplicated surgery under general anaesthetic.[93] Even adult death from associated adrenal hypoplasia has been described.[94] Preservation of secretion of gonadotrophins, however, is not uncommon.[95] Hypoplasia of the olfactory tract can be associated.[96]

Children with bilateral ONH affecting the whole of each optic nerve, and those with poor vision and nystagmus, are more likely to have non-ocular developmental abnormalities than those with unilateral or bilateral segmental hypoplasia.[97]

Treatment

In some cases patching can improve vision, but in others patching makes no difference.[98] If patching is not successful in improving visual function, it should be stopped to prevent distress. Careful refraction and prescription of spectacles is indicated in selected cases to prevent an additional amblyopic component from astigmatism, which has been found to be commoner in children with ONH.[99]

Prognosis

Some children with bilateral ONH, who present as blind in early infancy, may show a degree of delayed visual maturation and go on to develop mobility vision or better during the ensuing 2 years.[100]

Optic disc cupping in PVL — a variant of ONH?

PVL has recently emerged as an important and common cause of visual impairment in children.[101]Disturbance of development of posterior periventricular white matter at a time of intrauterine development between 29 and 34 weeks can give rise to reduced acuity with features of crowding, lower visual field impairment, and a range of cognitive disorders in visual function. In a significant proportion of patients, optic disc cupping with thinning of the neuro-retinal rim has been observed.[58]

Trans-synaptic degeneration probably accounts for the cupping. It is known for example that damage to the occipital lobes in young monkeys gives rise to loss of ganglion cells in the retina.[102,103] Such damage in the adult monkey leads to the same phenomenon, but to a much lesser degree.[104]

Jacobson *et al*[58] describe ONH in association with PVL of probable early onset, but optic disc cupping in cases in which the disorder probably occurred at a later stage. In cases in which the pattern of PVL is suggestive of pathology at an earlier stage of development (affecting frontal periventricular white matter) ONH was observed, whereas in cases of presumed later onset (subcortical posterior PVL) optic disc cupping was observed. These authors therefore propose that early-onset pathology results in contraction of the size of the optic nerve (resulting in a classical pattern of ONH) but that such pathology of later onset leads to a cupped optic disc, which they argue is a variant of ONH; the pattern of the latter is due to the lesser plasticity of the sclera to adapt to the neuronal loss at a later stage of development.

Management of the child with visual impairment due to disorders of the optic nerve

Bilateral disorders of the structure of the optic nerve commonly result in impaired functional vision presenting in early childhood. It is essential that the child and his/her family are well looked after from the outset.

Optimising visual function
Refractive error is common. Astigmatism needs to be corrected for eyes in which there is evidence of visual function. In our experience, dynamic retinoscopy commonly reveals impaired accommodation, and the correction of relatively small amounts of hypermetropia can be rewarded with significant visual improvement in some cases.

There may be super-added amblyopia. A short trial of patching of the eye with the worse visual function is warranted, but if there is no improvement this should not be continued.

Working with parents
Breaking the news that vision is impaired requires compassion and sensitivity. The first consultation rarely conveys much information, and a planned approach at a later stage to discussing the wide range of questions that arise is essential. Parents need accurate and clear information about the condition and its prognosis. Such information is currently available on the internet, but only dedicated sites warrant recommendation (see Appendix A). Parents also need advice concerning how to look after a visually impaired child. For infants, the developmental guide 'Show Me what My Friends Can See'[105] can prove very helpful. Genetic counselling is indicated when there is a clear family history.

Measuring visual function and working within visual limitations
Vision is required for communication, access to information, and mobility. The maximum distance at which faces can be seen, and facial expressions seen and learned from, needs to be determined and conveyed to parents and carers. The language framework required to compensate for reduced vision should be established. The size and contrast, and the proximity of toys and printed information required for maximum speed of access to the information need to be determined. The provision of low-vision aids at an early stage warrants consideration. The visual limitations to all aspects of mobility need to be understood and compensated for.

Ensuring that impaired vision does not lead to developmental delay
Early intervention can contribute significantly to a positive developmental outcome. This needs to be put in place at an early stage by notifying and informing the appropriate authorities.

Sources of information for parents and children
Reliable sources of information are very much appreciated by families. United Kingdom sources are listed in Appendix A.

In conclusion, in the majority of cases, disorders of optic nerve structure are sporadic and the aetiology is unknown. A positive approach aimed at habilitation and optimising visual function from an early stage during development reaps dividends. An approach that says 'there is nothing more I can do' should be a thing of the past!

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Appendix A

Organisations providing help for families with children with visual impairment.

Specific organisations

(1) Focus (Septo Optic Dysplasia/Optic Nerve Hypoplasia)
E-mail: uksupport@focusfamilies.org
Website: www.focusfamilies.org

(2) MACS (Micro & Anophthalmic Children's Society)
Coloboma is one of the eye conditions covered by MACS
MACS
1 Skyrmans Fee
Frinton-on-Sea
CO13 0RN
Tel/Fax: 0870 600 6227
E-mail: enquiries@macs.org.uk
Website: http://www.macs.org.uk

(3) Eyeless Trust
Eyeless Trust
PO Box 1248
Slough
SL2 3GH
Tel: 01494 672006
E-mail: ANDREW.paul@eyeless.org.uk
Website: http://www.eyeless.org.uk

General organisations helping families with visual impairment

(1) Visual Impairment Scotland
Visual Impairment Scotland is a new organisation that aims to provide information and support to children with visual impairments and their parents. Clear and understandable medical information documents are provided as well as a parent network support system.
Tel: 0131 651 6078
E-mail: viscotland@ed.ac.uk
Website: www.viscotland.org.uk

(2) Specific Eye Conditions (SPECS)
SPECS provides support to a wide range of organisations dedicated to helping people with eye conditions. The website provides a directory for patients, parents, and clinicians who are looking for information and support.
Specific Eye Conditions, PO Box 379, Margate CT9 1WD
Tel: 01843 292 435
E-mail: info@eyeconditions.org.uk
Website: www.eyeconditions.org.uk

(3) Contact a Family
Contact a Family is a national registered charity, founded in 1979, for families with disabled children. Contact a Family's aim is to enable families to organise their own systems of support and contact.
Contact a Family, 209-211 City Road, London EC1 V 1JN
Tel: 020 7608 8700; Fax 020 7608 8701
Helpline: 0808 808 3555 or Textphone: 0808 808 3556; Freephone for parents and families (1000–1600 hours, Mon–Fri)
E-mail: info@cafamily.org.uk
Website: http://www.cafamily.org.uk

(4) The National Blind Children's Society
This is a national organisation that offers a wide range of services to visually impaired people aged between 0 and 24.
NBCS, Bradbury House, Market Street, Highbridge, Somerset TA9 3BW
Tel: 01278 764 764
E-mail: enquires@nbcs.org.uk
Website: www.nbcs.org.uk

(5) Look
Look is a parent-run organisation that was established to give mutual support to families of visually impaired children. There are local UK branches.
LOOK, 49 Court Oak Road, Birmingham B17 9TG
Tel: 0121 428 5038
Website: www.look-uk.org

(6) Royal National Institute of the Blind (RNIB)
The RNIB offers a wide range of family support services.
105 Judd Street, London WC1 H 9NE
Telephone Helpline: 0845 766 99 99 (for the price of a local call, UK callers only)
Tel: 020 7388 1266 (switchboard/overseas callers)
Fax: 020 7388 2034
Website: www.rnib.org.uk