

MODULE 11 Part 2

COURSE CODE: C-7984

Optometric management of posterior segment eye disease – Optic nerve anomalies



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As the saying goes, the eyes are a window to the soul. They are also a window to the brain, and one can discover a number of conditions, sometimes life-threatening, affecting the brain by their effect on optic nerve appearance and function.

The main concerns when assessing any abnormalities are (1) 'Do I need to refer?' and (2) 'If so, how urgently?' In general, abnormalities that are of acute onset, associated with symptoms or signs of compromised optic nerve function, even when the nerve head looks normal, or with pain, tend to be more urgent than those that are long-standing, or congenital, and asymptomatic.

Important considerations when suspecting optic nerve disease

A general history and ophthalmic examination, including checking of intraocular pressures and full fundal inspection is useful in assessing any ocular disease, including optic nerve problems.

The following are important specific questions to bear in mind when assessing any potential optic nerve abnormalities.

- Does the disc look abnormal?
- Is it swollen?
- Are symptoms of recent onset?
- Are there specific patterns of pain?
- Is there evidence of abnormal nerve function?

Generally if the answer is yes to some/all of the above, the more worried one is and the more likely that referral is warranted.

Disc appearance

Abnormal disc features are often not in themselves specific for one particular pathology, except in the case of particular congenital malformations which will be mentioned later. Of course, the presence of a congenital malformation does not exclude acquired disease, and so one should be wary in attributing any new symptoms to a congenital anomaly.

Key pathological features to look out for include the following:

- Swelling
- Hyperaemia
- Pallor
- New vessels
- Absent venous pulsation
- Abnormal cupping
- Disc haemorrhages

Swelling

Swollen discs (elevated, with indistinct margins) may mean papilloedema,

which is defined as bilateral swollen discs due to raised intracranial pressure (e.g. due to an intracranial tumour) and can hence indicate potentially life-threatening disease. Discs may also be swollen in a number of other states, but bilateral swollen, hyperaemic discs, perhaps with peripapillary haemorrhages and associated with a headache makes papilloedema more likely. Other causes of disc swelling include accelerated hypertension and inflammation (papillitis) as well as acute anterior ischaemic optic neuropathy, though in this case the disc is pale, rather than hyperaemic. The optic disc may also look swollen in a number of normal states – e.g. optic disc drusen, tilted discs, peripapillary myelination and in the crowded, small discs of hypermetropes.

The optic disc may also be swollen in central retinal vein occlusions, posterior uveitis and in ocular hypotony, hence a full fundal examination and measurement of intraocular pressure is also important.

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Hyperaemia

Although a pink disc is healthy, hyperaemic, swollen discs occur in papillitis and papilloedema as above.

Pallor

Disc pallor occurs in anterior ischaemic optic neuropathy (AION). If the disc is swollen as well, this may indicate that the ischaemia is acute and hence referral is more urgent, particularly as the cause may be giant cell arteritis (GCA) – a neuro-ophthalmic emergency. Specific patterns of pain are associated with GCA. Non-arteritic AION is less urgent, but usually occurs only in the context of small, crowded discs. A pale, flat atrophic disc indicates longstanding ischaemia and so referral is less urgent. Pale, grey discs can also occur after chronic papilloedema (e.g. idiopathic intracranial hypertension) and papillitis. These conditions where the disc is atrophic and pale, but had been swollen previously, are known as *secondary optic atrophy*.

A pale disc with crisp margins, i.e. where there has been no prior swelling, indicates *primary optic atrophy*, and this can occur in hereditary or toxic optic neuropathies and also following any lesions affecting the optic nerve behind the eye, i.e. anywhere from the retrolaminar portion up to the lateral geniculate body where the optic nerve fibres end. Hence, this can include past retrobulbar optic neuritis and compression due to tumours or aneurysms. Lesions at the optic chiasm or further back may cause areas of optic atrophy in both eyes. In addition, atrophy can be diffuse or sectoral depending on which nerve fibres are affected. A pale, cupped disc occurs in advanced glaucoma.

New vessels

New vessels at the disc are usually a sign of proliferative diabetic retinopathy and will often be seen in the context of other diabetic retinopathy. This is an indication for urgent referral for treatment (laser photocoagulation of the retina to stop the hypoxic drive).

Absent venous pulsation

Absent venous pulsation can indicate raised intracranial pressure, but pulsation is also absent in 20% of normal individuals, so on its own may not be

significant (unless it is clear that the absence of pulsation is new).

Abnormal cupping

Abnormal cupping, where the neuroretinal rim containing the nerve fibres leaving the eye is thinned, may be indicative of glaucoma. A cup-to-disc ratio of 0.3 or less is normal. A higher cup-disc ratio may also be normal – termed physiological cupping – when there is a normal number of nerve fibres leaving the eye, but the scleral canal through which they exit is large (e.g. in myopic eyes), and so the cup-disc ratio is larger than normal. A difference in cup-disc ratios between the two eyes of more than 0.2 is suspicious.

Haemorrhages

Disc haemorrhages may be seen in the context of papilloedema, diabetic retinopathy, central retinal vein occlusion, glaucoma and other conditions.

Duration of symptoms

Optic nerve disease can often be asymptomatic, and picked up only at routine optometric examination. This is especially true of congenital anomalies, but also can be true of acquired conditions such as glaucoma and even papilloedema.

When symptoms are present, the more recent the onset, the more urgent the problem might be. When symptoms of visual loss are in one eye only, establishing their time-course may be difficult. Long-standing symptoms, especially the negative symptoms characteristic of optic nerve disease, might only be noticed when the patient happens to rub or close the other eye. It can be helpful to ask when vision was last known to be normal in that eye, and thus establish an upper limit for the duration of visual loss.

Symptoms that are worsening or changing, e.g. transient visual obscurations are also more worrying than those that are static.

As vascular events tend to be rapid, visual loss in anterior ischemic optic neuropathy is normally sudden, although there are cases where it develops over a couple of weeks. In optic neuritis, visual loss usually develops over a number of days, often associated with pain on eye

movement, and will start to improve over the next few weeks, although brightness perception may remain impaired for several months. Gradually, slowly progressive visual loss might suggest a compressive cause. Chronic, symmetrical, bilateral progressive or stable visual loss is seen in toxic or nutritional optic neuropathies. In congenital anomalies, symptoms, if any, tend to be stable, unless a complication arises, such as serous macular detachment in individuals with optic disc pits.

Leber's hereditary optic neuropathy usually manifests as visual loss over four to six weeks with about eight weeks' delay between the first and second eye. It usually affects young men in their teens having a family history of vision loss in males.

Patterns of pain

Patients often present to optometrists with a headache, and it is important to distinguish relatively benign tension headaches from more serious causes. The headaches of raised intracranial pressure tend to be worse in the morning on waking or at night, as CSF pressure rises when lying flat. They may also be worse on bending over or straining as these also cause rises in pressure, and may be associated with nausea and vomiting. Tension headaches, however, are intermittent, usually come on later in the day, and are frequently described as a frontal or retro-orbital band of tightness.

Pain on eye movements is a frequent feature of optic neuritis. The intra-orbital portion of the optic nerve sheath gives rise to part of the origin of the superior and inferior rectus muscles, and so is stretched by eye movements.

Tenderness over the temporal artery, scalp pain, e.g. on combing one's hair, and jaw claudication (pain in the jaws on chewing, relieved by rest) are features of GCA (also known as temporal arteritis) and hence could indicate an emergency situation where urgent steroid treatment is required to prevent vascular occlusion and blindness.

Evidence of abnormal disc function

Visual acuity and contrast sensitivity

Visual acuity should be measured, e.g. with a Snellen chart, with full refractive

correction, or with a pinhole. It should be tested for near, e.g. with close reading charts, as well as for distance. In the early stages of disease, the visual acuity may be only slightly decreased. Contrast sensitivity tests spatial resolution more thoroughly and can pick up more subtle visual loss.

Afferent pupillary defect

This is an *objective* test. A relative afferent pupillary defect (RAPD) is useful in detecting current or previous optic nerve disease, and in ruling out functional or non-organic visual loss. It is important to remember that it signifies an asymmetry of conduction between the two optic nerves, so is only present in unilateral or asymmetric optic nerve disease – if, for example, there is bilateral optic nerve disease, e.g. a pituitary tumour causing compression at the chiasm, then it may be absent.

Reduced colour vision

Colour vision is reduced in optic nerve disease, although it can also be impaired in macular disease. However, in the presence of reasonably good visual acuity, an acquired colour vision defect is quite a good indicator of optic neuropathy. One can test this by simply asking the patient to look at a red object with each eye in turn and report whether the 'redness' was the same. One can also test colour vision more formally. Kollner's rule holds that red-green dyschromatopsia occurs in optic nerve disease whereas blue-yellow defects

occur in retinal disease. However, there are some optic nerve conditions in which blue dyschromatopsia may occur including glaucoma, autosomal dominant optic atrophy, chronic papilloedema and even demyelinating optic neuropathy.

Reduced brightness perception

This can also be tested by asking the patient to compare brightness perceived in each eye in turn, and may remain impaired even after visual acuity has returned to normal, e.g. after an episode of optic neuritis.

Visual field defects

Optic nerve diseases cause negative visual symptoms, i.e. loss of an area of vision, whilst macular diseases often give positive symptoms (distortion of images, etc.). One can, however, experience photopsia with optic nerve disease. Intermittent transient obscurations of vision may occur with papilloedema: vision disappears for a second or two, often when the intracranial pressure is raised such as bending down or coughing. Loss of vision associated with gaze is characteristic of lesions that compress the orbital optic nerve, classically optic nerve sheath meningioma.

The pattern of field loss may hint at the cause. A *central scotoma* may be seen in neuropathies associated with demyelination (diffuse loss might be seen on a Humphrey visual field), toxic or nutritional states (when the scotoma is

small) and Leber's hereditary optic neuropathy (when the scotoma is large). *Enlargement of the blind spot* may occur in papilloedema and congenital optic nerve anomalies.

Field defects that respect the *horizontal meridian* (i.e. they do not cross it) occur in anterior ischaemic optic neuropathy (inferior altitudinal), optic nerve head drusen, papilloedema and glaucoma. In advanced papilloedema or glaucoma, the whole field is constricted.

Field defects due to optic nerve compression at or behind the chiasm tend to respect the *vertical meridian* and may progress. Tilted optic discs may give field defects that look as if they respect the vertical meridian; usually, however, they do cross the midline somewhere and, as they are congenital, they do not progress.

Disorders affecting the chiasm or optic tracts will give field defects in both eyes (bitemporal hemianopia in chiasmal compression, homonymous defects in more posterior lesions), whilst those affecting the nerve before the chiasm will give monocular defects only.

Main types of optic nerve abnormality

Optic neuritis

Optic neuritis refers to an inflammatory or demyelinating process affecting the optic nerve.

Anatomically, it may be classified as retrobulbar neuritis (affecting the nerve behind the eye), papillitis (affecting the disc) or neuroretinitis (affecting the disc and the retinal nerve fibre layer also) – see Table 1.

Causes of optic neuritis include the following:

- Demyelination (commonest in adults)
- Post-viral (commonest in children)
- Adjacent infection
- Granulomatous inflammation or autoimmune vasculitides

Demyelination

Optic nerve fibres are myelinated from the lamina cribrosa onwards, and demyelinating optic neuritis involves loss of this myelin sheath and deposition of a plaque (which can be visualised on T2-weighted MRI scans).

Retrobulbar neuritis Optic nerve head is not involved, so the disc looks normal. This is the most common type in adults; often represents demyelination.

Papillitis Optic disc is inflamed, swollen and hyperaemic. There may be peripapillary flame-shaped haemorrhages. This is the most common type in children, often bilateral and post-viral.

Neuroretinitis Inflammation at the disc and also the retinal nerve fibre layer. A "macular star" develops as disc swelling resolves. Less common than the other types; rarely represents demyelination. 25% of cases are idiopathic (Leber idiopathic stellate neuroretinitis). Other cases associated with cat-scratch fever (60%) or other conditions.

➔Table 1

Anatomical classification of optic neuritis



Women are more commonly affected than men, and presentation is usually between the ages of 20 and 40 years, often with pain on eye movements or a retro-orbital headache which may resolve over a week or so, associated with progressive visual loss in the affected eye. Acuity usually falls to between 6/18 and 6/60. Some patients experience tiny white flashes (phosphenes). A relative afferent pupillary defect, loss of colour vision, and a centro-caecal scotoma, or diffuse depression of the central visual field, may be demonstrated. The disc usually looks normal as the neuritis is usually retrobulbar. Much later there may be optic atrophy. If the patient has had previous optic neuritis in the other eye, there may be some atrophy evident at the disc which will give a clue to the diagnosis.

Optic neuritis may be an isolated episode in many cases. However, there is a 38% risk of developing multiple sclerosis (MS) (a disease characterised by episodes of demyelination of different parts of the central nervous system separated in time) over the next 10 years. The risk is 56% if T2-signal lesions are seen on MRI (see Fig. 1), and 22% if no such lesions are seen. Other factors that increase the risk of developing MS are a winter onset of optic neuritis, patients who are genetically HLA-DR2 positive, and those whose symptoms are worse with elevation of body temperature (the Uhthoff phenomenon).

If the diagnosis of optic neuritis is in

doubt, electrodiagnostic tests can be helpful to check for delayed visual evoked potentials. In classic optic neuritis, investigations are not always needed. Although MRI can identify individuals at higher risk of MS, there is no real preventative treatment and no way of predicting when the next attack might occur or how mild or severe future disease might be. Thus discussing possibilities of MS with patients is probably not helpful unless patients themselves ask or have had any other neurological symptoms making MS very likely.

In terms of prognosis, 75% of patients recover to a visual acuity of 6/9 or better, although colour vision and brightness perception may remain abnormal. A mild afferent pupillary defect may also persist. In total 10% of patients may develop chronic optic neuritis.

Treatment does not influence eventual visual outcome – this was shown by the Optic Neuritis Treatment Trial. Intravenous and oral steroids may speed up recovery, but have no effect on final outcome, and can give rise to side-effects. Oral prednisolone alone has been associated with a higher recurrence rate. Intramuscular interferon beta-1a has been shown to produce a small benefit.

Other causes of optic neuritis

Post-viral optic neuritis occurs, most commonly in children, one to three weeks after infections such as measles, mumps, chickenpox, whooping cough,

rubella and glandular fever, or even after immunisation. Bilateral papillitis is usually seen, and there may be other neurological features, from headaches to seizures. Treatment in most patients is not required, as prognosis is good, but intravenous steroids are considered in severe visual loss.

Infectious optic neuritis can be sinus-related, associated with severe headache and sphenothymoidal sinusitis. Treatment is with systemic antibiotics and sometimes surgical drainage. Other infectious causes include cat-scratch fever, syphilis, Lyme disease and Varicella zoster virus. In terms of non-infectious causes, optic neuritis may be seen in some autoimmune vasculitides and in sarcoidosis – up to 5% of patients with neurosarcoid may be affected, with the optic disc sometimes having a lumpy appearance, suggestive of the granulomatous inflammation characteristic of sarcoid.

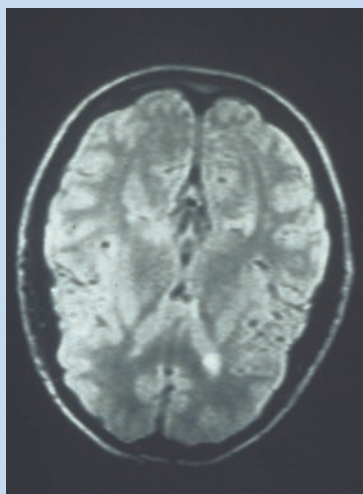
Anterior ischaemic optic neuropathy

Anterior ischaemic optic neuropathy (AION) is due to occlusion of the short posterior ciliary arteries which supply the optic nerve head. The cause can be non-arteritic, which is more common, or arteritic, which is more serious.

Non-arteritic AION

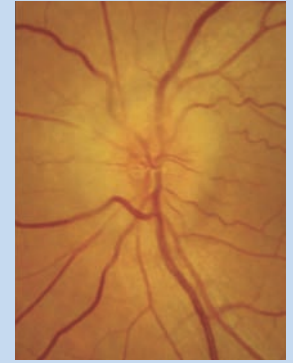
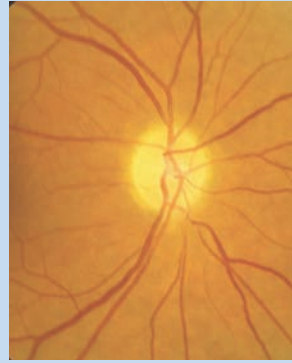
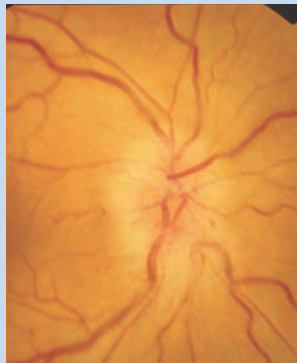
This is the most common optic neuropathy in the elderly. Men are more commonly affected, and presentation is usually between 50 and 70 years, with sudden, painless visual loss; occasionally it may be progressive in a step-wise manner. The extent of acuity and colour vision impairment may vary. An altitudinal field defect is commonly seen, i.e. either the upper or lower field. Acutely, the optic disc is swollen and pale (Fig. 2a), especially the sector that is infarcted. The disc later becomes atrophic (Fig. 2b).

An important risk factor is having a small, crowded, hypermetropic looking disc, with a very small cup, the so-called “disc at risk” (see Fig. 2a). Other risk factors include those for general cardiovascular disease, i.e. hypertension, diabetes, high cholesterol, antiphospholipid syndrome, elevated serum homocysteine levels, etc. and also sudden hypotensive events, cataract



➔ **Figure 1. MRI scan**

Two slices of the MRI scan of a 23 year old patient with optic neuritis. The white lesions seen are typical of those seen in the periventricular area in multiple sclerosis, and indicate a higher risk of developing further neurological problems.



⇒ **Figure 2a. Anterior ischaemic optic neuropathy**

Fig 2a shows the discs of a 48 year old man with AION affecting his right eye, with a classic pale swollen disc, associated with a nerve fibre layer haemorrhage, and the small “disc at risk” can also be noted in left eye.

⇒ **Figure 2b. Anterior ischaemic optic neuropathy**

Fig 2b shows the same patient's discs photographed 9 months later: he has optic atrophy in the right disc, but has unfortunately presented with loss of vision in his left eye, with AION now affecting this eye.

surgery and even use of sildenafil (Viagra). Thus, a general medical referral is sometimes indicated.

A total of 6% of patients have recurrences in the same eye. In all 10% will have involvement of the other eye in the next two years and 15% after five years. There is no definitive treatment to improve vision or prevent deterioration, although aspirin may have a role, and general cardiovascular risk factors such as diabetes and hypertension should be addressed.

Arteritic AION

Arteritic AION is associated with giant cell arteritis (GCA) and gives the same disc appearance as non-arteritic AION. There may be associated nerve fibre layer haemorrhages or even choroidal infarcts, and the visual loss tends to be more severe. Women are more

commonly affected, and patients are usually over 60. Systemic features of GCA may be noted, including headache, scalp tenderness, malaise, fever, weight loss and jaw claudication, although in 20% these features are absent at presentation (occult GCA). The condition is serious as, if untreated, visual loss may rapidly become bilateral, in up to 65% within three weeks. Hence, if suspected, urgent hospital referral is indicated. We would strongly recommend optometrists refer any patient with AION to hospital urgently, allowing ophthalmologists the responsibility to decide whether the AION is arteritic or non-arteritic, because of the implications of untreated GCA.

Blood tests may aid the diagnosis: an elevated ESR – erythrocyte sedimentation rate – or CRP – C-reactive protein – show evidence of

inflammation. A temporal artery biopsy shows “giant cells” under microscopy. Treatment is usually initiated beforehand if the diagnosis is strongly suspected, and consists of high dose oral steroids, e.g. 80mg of prednisolone, subsequently reduced over time.

Papilloedema

This refers to swollen optic discs due to raised intracranial pressure (ICP). It necessitates emergency referral if detected. Swollen discs due to other causes are not papilloedema, and discs that appear falsely swollen are termed pseudopapilloedema. It is important to note that visual function (acuity, colour vision, visual field testing) is usually normal in early papilloedema. However, most patients have symptoms.

Presenting symptoms include the characteristic headache as described above. Note that children can also get papilloedema, so it is worth attempting to examine their discs also. Patients may complain of transient visual obscurations – loss of vision lasting seconds, often with changes in posture or straining, e.g. the Valsalva manoeuvre (forced breathing out against a closed glottis). There might also be other signs of raised ICP such as a sixth nerve palsy, giving diplopia on lateral gaze: this is the “pseudo-localising sign” (the nerve is stretched by the high ICP). Visual acuity is initially normal, but later falls. Visual field tests show an enlarged blind spot initially and progressive field constriction in late stages.

Chronologically papilloedema has

Mechanical Elevation of the optic nerve head
Blurring of disc margins
Filling in of the physiological cup
Oedema of the peripapillary nerve fibre layer
Retinal and/or choroidal folds
Vintage papilloedema: disc pallor with white refractile bodies

Vascular Hyperaemia of the disc
Loss of spontaneous venous pulsation
Venous dilation
Obscuration of small surface vessels on disc
Peripapillary haemorrhages (flame-shaped)
Exudates in or around the disc
Nerve fibre layer infarcts (cotton wool spots)

⇒ **Table 2**

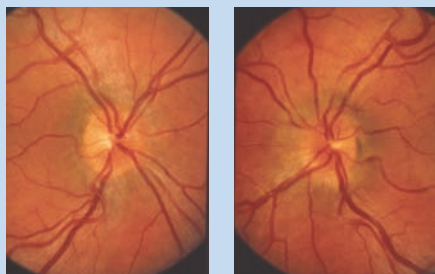
Signs of papilloedema

been divided into early, acute, chronic and vintage stages (see Fig. 3), with gradually increasing severity of symptoms and signs. Early signs include blurring of the disc margins (initially nasal, then superior, inferior and temporal), disc hyperaemia and loss of spontaneous venous pulsation. Venous pulsations can be detected using the high magnification of a direct ophthalmoscope and observing a vein just as it leaves the disc margin. If pulsations are present, raised ICP is very unlikely. If absent, this does not necessarily mean papilloedema as pulsations are absent in a fifth of normal people. Table 2 lists fundus signs of papilloedema.

Causes of pseudopapilloedema include hypermetropic discs, which are small, crowded, with a small cup, and hence may appear swollen, or myopic tilted discs, which may appear swollen superonasally (see Fig. 4). Other causes include peripapillary nerve fibre myelination (which may also give the appearance of exudates) and buried optic disc drusen. Table 3 summarises the important questions to ask when papilloedema.

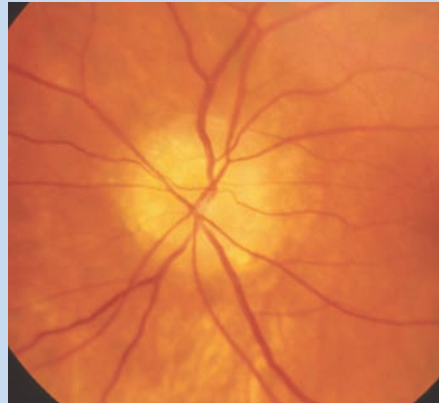
Visual field testing may be useful in showing an enlarged blind spot, or, in later stages, constriction of the field. Fluorescein angiography can be helpful in confirming the diagnosis – the early angiogram shows a dilated capillary network over the disc and the late angiogram shows leakage from the disc (Fig. 5).

Causes of papilloedema include intracranial masses (brain tumours),



➔ **Figure 3. Papilloedema**

These three sets of optic disc photographs show early, established and chronic papilloedema. Fig 3a shows blurring of the nasal optic disc margin and a hyperaemic disc, and it would be important to look for lack of spontaneous venous pulsation to confirm the suspicious appearance.



➔ **Figure 4. Pseudopapilloedema**

Figure 4a shows buried disc drusen. The disc looks swollen, but note the irregular "lumpy" edge to the disc, the anomalous retinal vessels and the yellowish appearance.

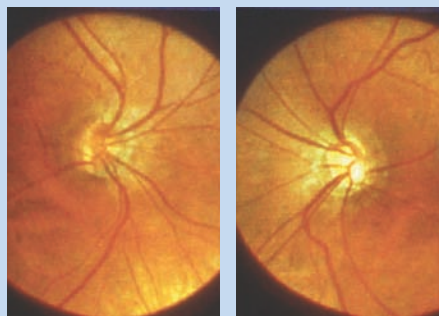


Fig 4b illustrates tilted small discs, which are sometimes confused for swollen discs, especially the temporal edge.

hydrocephalus (raised ICP due to blockage of flow of cerebrospinal fluid), venous sinus thrombosis, meningitis and idiopathic intracranial hypertension. CT (computerised tomography) or MRI (magnetic resonance imaging) brain scans can help

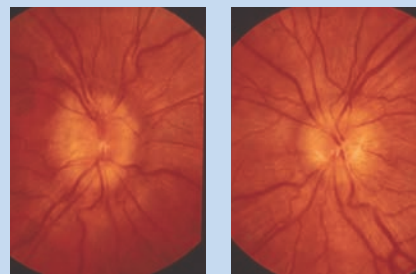


Fig 3b shows established papilloedema with elevated nerve head and loss of the optic disc cup.

Are there symptoms of raised ICP?

Typical headache
Transient visual obscurations
Diplopia on lateral gaze (VI nerve palsy)

What is the refraction of the eye?

Disc may look swollen in:
– hypermetropes
– myopes with tilted discs

Is there spontaneous venous pulsation?

Presence makes papilloedema unlikely

Is there anomalous retinal vasculature?

Can be associated with disc drusen

Are there vascular signs of papilloedema?

Peripapillary haemorrhages
Exudates or retinal cotton wool spots
(See Table 2)

➔ **Table 3.**

Questions when faced with swollen discs suspicious of papilloedema

identify the cause, and sometimes a lumbar puncture is required to measure the pressure and to examine the cerebrospinal fluid (CSF).

Idiopathic intracranial hypertension refers to raised ICP of unknown cause, i.e. not due to a mass lesion, and if untreated can lead to blindness. Typically it affects obese, young women with no other neurological symptoms, often with recent weight gain. The most serious complication is optic atrophy

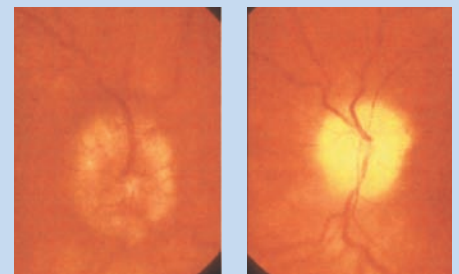
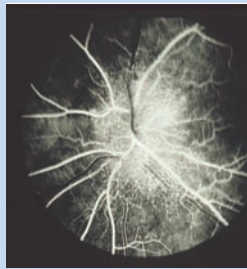
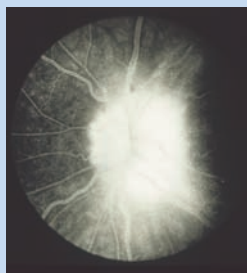


Fig 3c shows vintage papilloedema in the right disc with some refractile white crystals on the surface, and the left disc is already atrophic due to chronic papilloedema – this eye has already lost significant vision in a 24 year old patient with idiopathic intracranial hypertension.



➔ **Figure 5. Fluorescein angiogram of an optic disc in a patient with papilloedema**

Fig 5a is the early arterial phase of the dye transit and shows dilated capillaries over the surface of the optic disc. The arteries are filled with dye (white) and the veins still empty (black). Fig 5b shows the late photograph of this disc with fluorescein leakage from the optic disc.

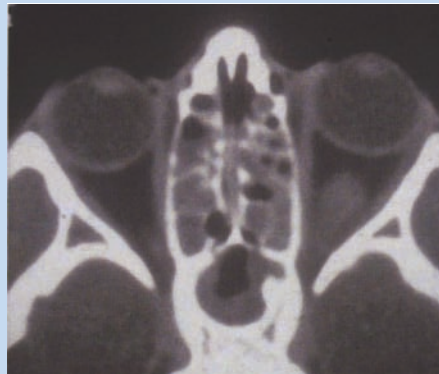


with constriction of peripheral fields. It can be associated with steroid use/withdrawal and with tetracyclines and vitamin A. Treatment includes encouragement of weight loss, which may be curative. Acetazolamide reduces CSF production and can be helpful. However, if there is evidence of progressive field constriction, surgical procedures may be required, which include shunting, where the CSF is diverted to the abdominal peritoneal cavity, or optic nerve sheath fenestration, where the optic nerve sheath is incised behind the eye to allow CSF to leak.

Compressive optic neuropathy

The optic nerve glioma (Fig. 6a) and optic nerve sheath meningioma (Fig. 6b) are the two main tumours that cause visual loss by compressing the optic nerve (as opposed to those which compress the optic chiasm, such as pituitary tumours, resulting in the characteristic bitemporal hemianopia). Pathologies that compress the nerve in the orbit, such as thyroid eye disease, may also cause proptosis.

In total 75% of optic nerve gliomas present with visual loss in the first decade of life, and 90% by the age of 20. The course of the disease is usually benign, and surgery is not performed as it involves severing the optic nerve. Gliomas can be associated with



➔ **Figure 6. Compressive optic neuropathies**

Figure 6a shows the CT scan of a child with a swelling of the optic nerve which is an optic nerve glioma.

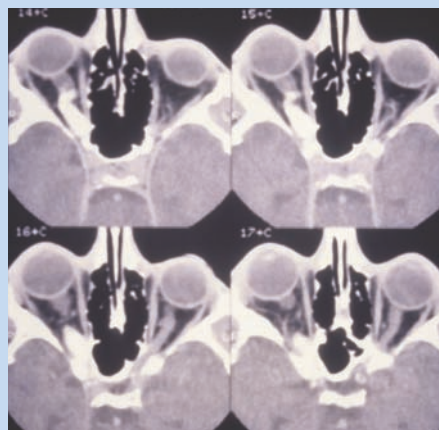
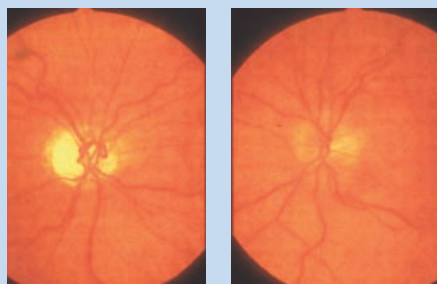


Figure 6b is the CT scan of a patient with an optic nerve sheath meningioma which can be seen on the different cuts as the tortuous thickened and calcified nerve sheath of the right optic nerve (seen above).



The optic discs of this patient are seen in figure 6c; the left disc is normal but the right disc displays the classic triad of signs of optic atrophy, disc swelling and opticociliary shunt vessels.

neurofibromatosis Type 1, an autosomal dominant inherited disease that involves benign nerve tumours in different nerves throughout the body.

Patients with an optic nerve sheath meningioma are usually adults, typically middle-aged women, presenting with the classic triad of visual loss, optic atrophy and opticociliary shunt vessels (on the disc between the posterior ciliary and retinal circulations) – see Fig. 6c. They often have a proptosis if the meningioma is intraorbital. Surgery is difficult as the tumour can rarely be removed, but the tumours may respond to radiotherapy, and some are hormone-dependent, responding to anti-hormonal treatment.

Diabetic papillopathy

This is a rare condition affecting diabetics, usually Type 1 diabetics around their twenties. It involves mild disc swelling, which is bilateral in around 75% of cases, and which may be associated with mild visual loss – acuity is rarely worse than 6/12. It usually resolves spontaneously, but may take several months. The aetiology is unclear but it may represent a mild ischaemic optic neuropathy. Clearly other causes of disc swelling should be excluded first before attributing it to the diabetes.

Toxic or nutritional optic neuropathy

This condition is sometimes called “tobacco-alcohol” amblyopia as it frequently presents in individuals, who are smokers and heavy drinkers. In nutritional optic neuropathy, there is usually a deficiency of B vitamins – B1 (thiamine) or B12 (seen in bowel absorption problems). Treatment is with vitamin replacement.

Presentation is with fairly sudden, progressive loss of vision, and a centro-caecal scotoma, fairly symmetrical in both eyes, with impaired colour vision. The visual field defect may be difficult to define using a white target, but tends to be larger and easier to plot with a red target. Optic discs may be normal initially, or show subtle temporal pallor, with splinter-shaped haemorrhage and sometimes minimal disc oedema. In the early stages, the visual loss is reversible, but later advanced stages are often unresponsive to treatment.

Leber's hereditary optic neuropathy

This is a rare cause of bilateral optic neuropathy, usually presenting in males, who are affected nine times more commonly than females, between the ages of 10 and 35, with a peak incidence in the teens. It results from mitochondrial mutations, thus inheritance is always through the mother. Atypical cases may affect females and present at other ages, so it should be considered in any patient with bilateral optic neuropathy. There is typically painless loss of vision in one eye over a few weeks, followed by similar loss in the other eye (usually after an eight-week delay). The disc looks hyperaemic, with indistinct margins – this is pseudo-oedema, as there is no leak on fluorescein angiography. Dilated capillaries also may be seen on the disc surface and adjacent retina, and this telangiectatic microangiopathy may be seen in the fundi of asymptomatic female relatives. Visual loss is variable, with some mutations having a worse prognosis. Treatment is generally ineffective.

Other hereditary optic atrophies

There are other hereditary optic atrophies that are rarer still. These include dominant optic atrophy (Kjer syndrome: autosomal dominant inheritance, presenting typically before the age of 20), Behr syndrome (autosomal recessive inheritance, presenting in the first decade, associated with abnormal gait and mental problems) and Wolfram syndrome (autosomal recessive inheritance, presenting by the age of 21, associated with diabetes mellitus, diabetes insipidus, deafness, mental problems and other abnormalities).

Congenital anomalies

Congenital anomalies of the optic disc are common. They are frequently insignificant, but can give rise to visual field defects, be associated with other neurological defects or give rise to macular problems. Some may be confused with papilloedema, particularly optic disc drusen, the small discs of hypermetropes and tilted discs.

Anomaly	Features, significant associations or complications
Tilted disc	<ul style="list-style-type: none"> • Nerve enters eye obliquely. Disc usually directed inferonasally. Elevated side may have indistinct margin. Temporal vessels may deviate nasally initially (situs inversus). • May have superotemporal field defects that do not respect the midline • Associations: myopic astigmatic refractive error, chorioretinal thinning
Optic disc pit	<ul style="list-style-type: none"> • Large disc with pit, usually temporal. Field defects common. • 45% of non-central pits will develop serous macular detachment (25% resolve; some might need laser or vitrectomy).
Optic disc coloboma	<ul style="list-style-type: none"> • Due to incomplete closure of optic fissure during development. Excavation inferiorly (reduced fluorescence on fluorescein angiography) • Acuity often reduced. Superior field defect. Complications include serous macular detachment. • Associations: iris/fundus colobomas, microphthalmos, chromosomal abnormalities and systemic syndromes (e.g. CHARGE syndrome).
Morning glory anomaly	<ul style="list-style-type: none"> • Very rare, usually unilateral and sporadic. Ring of chorioretinal disturbance around a large disc with a funnel-shaped excavation. White tuft of glial tissue over centre. • Increased number of vessels, emerging like spokes from rim of disc. • Visual acuity may be normal. Serous retinal detachment in 30%. • Choroidal neovascularisation may develop. • Associations: frontonasal dysplasia, neurofibromatosis type 2 and PHACE syndrome.
Optic nerve hypoplasia	<ul style="list-style-type: none"> • Small grey disc with surrounding halo of hypopigmentation (double ring sign). Reduced number of nerve fibres. Distance from fovea to temporal disc border is 3x disc diameter or more. • Vision variable from blindness to normal acuity. If unilateral, may present with squint. • Associated with a range of midline brain defects. Predispositions: maternal use of alcohol, LSD, quinine, steroids or other agents during gestation.
Prepapillary loop	<ul style="list-style-type: none"> • Unilateral vessel loop from disc into vitreous. • 10% develop obstruction of the retinal artery supplying the loop
Bergmeister papilla	<ul style="list-style-type: none"> • Glial tissue on the disc surface (unilateral)
Megalopapilla	<ul style="list-style-type: none"> • Horizontal and vertical diameters are 2.1 mm or more
Peripapillary staphyloma	<ul style="list-style-type: none"> • Normal disc at the bottom of a deep excavation. Acuity reduced. • Complications: local retinal detachment
Optic nerve aplasia	<ul style="list-style-type: none"> • Very rare: absent or rudimentary disc.
Papillorenal syndrome	<ul style="list-style-type: none"> • Autosomal dominant condition, central excavation in disc, central retinal vessels replaced by cilioretinal vessels. Kidney abnormalities.
Myelinated retinal nerve fibres	<ul style="list-style-type: none"> • Ganglion cell axons are normally not myelinated until they pass out of the eye through the lamina cribrosa. Abnormal myelination in the retina or near the disc can be mistaken for exudates or disc swelling (if at the disc margin).
Optic disc dysplasia	<ul style="list-style-type: none"> • A deformed disc that doesn't conform to any recognised category.

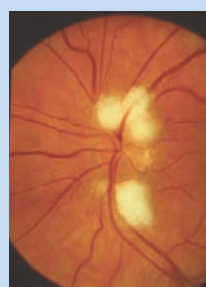
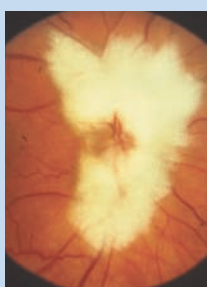
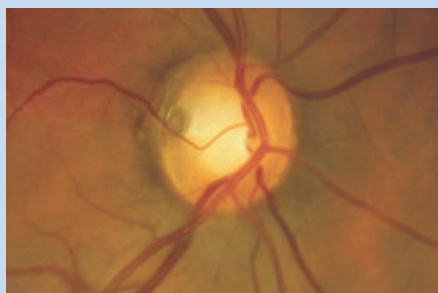
Disc drusen

This refers to hyaline-like calcific material within the optic nerve head, affecting 0.3-1% of the population. The condition is bilateral in approximately 75% of cases, and may have autosomal dominant inheritance. Buried drusen can be confused with disc swelling, as they can give an irregular disc margin with an absent cup. Patients are usually asymptomatic. Once they have appeared on the surface as whitish lumps, which may happen during the teens, they are less easily confused with papilloedema. In addition, they are more likely to have anomalous vasculature, such as trifurcation of first order retinal vessels, and the disc is more pink/yellow, rather than truly hyperaemic.

Disc drusen can be associated with retinitis pigmentosa, angioid streaks and Alagille syndrome (a genetic condition with pale fundi, hypertelorism, biliary, vertebral and heart problems). Complications are rare, but include juxtapapillary choroidal neovascularisation, disc new vessels, central retinal arterial/venous occlusion and limited field loss in 75%.

Other anomalies

Tilted discs and small, hypermetropic discs have been mentioned. Other anomalies include optic disc pits (Fig. 7a), which can lead to serous macular



➔ Figure 7. Congenital disc anomalies

Figure 7a is an example of an optic disc pit (a small pit on the temporal edge of the disc) which may result in visual loss due to serous fluid leaking underneath the macula. Figure 7b displays myelinated nerve fibres, commonly misdiagnosed as exudates.

detachment, colobomas due to incomplete closure of the optic fissure during development and myelinated nerve fibres (Fig. 7b), which can be confused with a swollen disc (if at the margin) or exudates. Table 4 summarises some of these anomalies.

Conclusions

Optic nerve abnormalities range from incidental congenital anomalies to manifestations of sight-threatening conditions such as temporal arteritis or even life-threatening disease, such as a brain tumour. A careful, systematic approach, examining optic disc appearance, assessing patterns and chronology of symptoms, and looking for evidence of optic nerve dysfunction, will help one arrive at a differential diagnosis and hence decide on appropriate management.

Further reading

Riordan-Eva P. Clinical assessment of optic nerve disorders. *Eye* 2004; **18**, 1161-1168

Taylor D. Developmental abnormalities of the optic nerve and chiasm. *Eye* 2007; **21**, 1271-1284

Kanski JJ *Clinical Ophthalmology: A systematic approach*. 6th ed. London: Elsevier; 2007. pp. 82-89, 786-802

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Module questions

Course code: c-7984

Please note, there is only one correct answer. Enter online or by form provided

An answer return form is included in this issue. It should be completed and returned to CET initiatives (c-7984) OT, Ten Alps plc, 9 Savoy Street, London WC2E 7HR by March 5 2008.

1. Which one of the following is correct? Causes of optic atrophy include:

- A vitamin C deficiency
- B an optic disc pit
- C idiopathic intracranial hypertension
- D postural hypotension

2. Symptoms of raised intracranial pressure are most likely to include all the following except:

- A obscurations
- B nausea and vomiting
- C diplopia
- D madarosis

3. Which one of the following is correct? Adult patients with retrobulbar optic neuritis usually have:

- A peripheral field constriction
- B pain on eye movements/headache
- C bilateral swollen optic discs
- D bilateral severe visual loss

4. Which one of the following is correct? Non-arteritic anterior ischaemic optic neuropathy:

- A produces an homonymous visual field defect
- B occurs in large myopic "discs at risk"
- C may affect the second eye after the first eye
- D is associated with an elevated erythrocyte sedimentation

5. Patients with early papilloedema are most likely to present with which one of the following?

- A a central scotoma
- B headaches
- C a relative afferent pupillary defect
- D severe loss of vision

6. Which one of the following is correct? Clinical signs of early papilloedema include:

- A optociliary shunt vessels
- B retinal and choroidal folds
- C loss of spontaneous pulsation
- D venous sheathing

7. Which one of the following does NOT cause papilloedema in adults?

- A idiopathic intracranial hypertension
- B optic neuritis
- C a brain tumour
- D venous sinus thrombosis

8. Which one of the following is correct? Idiopathic intracranial hypertension:

- A always results in optic atrophy
- B is caused by venous sinus thrombosis
- C may cause peripheral visual field constriction
- D usually affects young men

9. Which one of the following is correct? Optic nerve sheath meningioma:

- A peaks in incidence in the second decade of life
- B may cause proptosis
- C rarely causes visual loss
- D is associated with neurofibromatosis Type 1

10. Which one of the following is correct regarding diabetic papillopathy?

- A it is common
- B it generally affects people with Type II diabetes
- C bilateral swelling of the optic disc occurs in around 75% of cases
- D patients will generally complain of extensive visual loss

11. Which one of the following regarding Leber's hereditary optic neuropathy is incorrect?

- A it affects males more than females
- B the peak incidence of the condition occurs in the teens
- C at the initial stages, patients suffer a bilateral loss of vision
- D inheritance is through the maternal side

12. Which one of the following is correct? Optic disc drusen are usually:

- A unilateral
- B symptomatic
- C associated with anomalous vasculature
- D caused by myelinated nerve fibres

Please complete on-line by midnight on March 5 2008 - You will be unable to submit exams after this date - answers to the module will be published in our March 7 issue



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