Optic atrophy

What is optic atrophy?

Optic atrophy (also termed optic neuropathy) is the loss of some or all of the nerve fibres in the optic nerve. It is an important sign of advanced optic nerve disease and is frequently seen in visual loss. It is said to be primary if it occurs without any preceding optic nerve head oedema and secondary if it is preceded by oedema. It may also be described according to the underlying aetiology (ie whether this relates to primary disease of the retina or whether the problem originates at the level of the optic nerve).

Optic atrophy is the end stage of a variety of causes of damage to the optic nerve anywhere along its length. There is most often no known cause; however, possible causes include direct trauma, pressure on or toxic damage to the nerve, and nutritional deficiencies.

Pathophysiology

There is a loss of axons and shrinkage of myelin, leading to widening of the optic cup.

Optic atrophy symptoms [1]

This depends on the causative condition. Optic disc atrophy in isolation results in the following symptoms and signs.

Reduction or loss of vision which may be central or peripheral depending on the underlying condition. Subtle damage may lead to loss of contrast or colour vision without measurable loss of acuity.

In unilateral optic atrophy there may be decreased perception of brightness in one eye relative to the other.

Signs

The disc is pale - comparison with the fellow eye may help elicit this sign. There is usually a reduction of the small blood vessels crossing the disc surface. In cases of secondary atrophy, the disc margin may be poorly delineated (due to gliosis rather than oedema). The appearance may offer some clues to the pathology.

- Where the atrophy is glaucomatous in origin, disc cupping will also be present.
- Sector disc pallor in older patients can suggest non-arteritic anterior ischaemic neuropathy.
- Severe optic atrophy in an elderly patient may be due to giant cell arteritis.
- Papilloedema may leave retinal folds and glistening bodies in the optic nerve head.

History

Signs and symptoms - speed of onset

- A rapid onset suggests demyelination, inflammation, ischaemia or trauma.
- More gradual onset suggests compressive, toxic/nutritional and hereditary causes.
- Very long history (years) suggests compressive or hereditary conditions.

Associated optic atrophy symptoms[1]

It is essential to ask about associated symptoms which might point towards an underlying cause.

- In a young patient, previous history of eye pain, paraesthesiae, ataxia or weakness suggest demyelination

- In an older patient, previous history of transient visual loss, diplopia, temporal pain, jaw claudication, fatigue, weight loss and myalgia suggests arteritic ischaemic optic neuropathy due to giant cell arteritis.
- In children, history of flu-like illness or vaccination could suggest para-infectious or post-vaccinial optic neuritis.
- Diplopia and facial pain suggest multiple cranial neuropathies due to inflammatory or neoplastic lesions behind the eye.
- Medication history should be noted - in particular, drugs which can be toxic to the optic nerve (eg, ethambutol, amiodarone, alcohol, methotrexate, ciclosporin).
- History of diabetes, hypercholesterolaemia and hypertension is common in patients with non-arteritic anterior optic neuropathy.
- Patients with known malignancy may have infiltrative or para-neoplastic optic neuropathy.
- Detailed family history may suggest hereditary autosomal and mitochondrial optic neuropathies. [2]

Examination [1]

- This should include acuities, colour vision, visual field check and pupillary reflexes.
- Visual acuity is not always impaired, as central vision may be initially spared.
- The assessment of colour vision should also observe speed of recognition, as this may detect relative differences between eyes. Red colour desaturation is a common early sign.
- A swinging light may be used to assess for relative afferent pupillary defect.
- Fundus examination may reveal a swollen or pale disc, although it may look normal. The disc is usually swollen in non-arteritic anterior ischaemic neuropathy and inflammatory (non-demyelinating) optic neuritis.

- A pale optic disc suggests a long-standing condition such as compressive, hereditary or toxic neuropathies.
- Cupping is seen in glaucoma, end-stage giant cell arteritis, congenital optic disc anomalies, compressive optic neuropathy, hereditary optic atrophy, radiation optic neuropathy and methanol poisoning.
- Some rare congenital conditions result in an anomalous appearance to the optic nerve head.

Investigations

- Optic atrophy is diagnosed on fundoscopy and may be confirmed with optical coherence tomography (a quick and painless imaging technique that can be performed in the outpatient clinic).
- Further investigation may then be required to assess function, such as formal visual field and colour testing.
- For a detailed account of assessing optic disc function, see the separate Examination of the Eye article.
- Neuroimaging of the brain is indicated where demyelination or compression are suspected. Multiple visually evoked potentials may also be helpful.
- Depending on the associated findings and suspected underlying cause, further investigations may be carried out - for example, to assess for the presence of a tumour (full neurological examination, imaging), to identify genetic abnormalities, or to diagnose suspected toxic neuropathies.

Causes of optic atrophy[1]

A study into the causes of optic atrophy in Saudi Arabia found:[3]

- The most common causes were tumours (62.2%), followed by hereditary disorders (17.1%). Autoimmune diseases (multiples sclerosis, antiphospholipid syndrome, neuromyelitis optica, systemic lupus erythematosus, and neuro-Behcet's), accounted 12.7% of cases).

- Less frequent causes included vascular/ischemic disorders (non-arteritic optic neuropathy, cerebral vascular accidents, sagittal sinus thrombosis and arteriovenous malformation), congenital malformations (hydrocephalus, arachnoid cysts, myelomeningocele and isolated craniosynostosis), infections, trauma and toxicity.

A review of children presenting in India has found causes in this age group to include congenital, inflammatory, infective, traumatic and vascular, including perinatal insults, space-occupying lesions and hypoxic ischaemia encephalopathy.[4]

Common causes of optic atrophy

Primary optic nerve disease

- Chronic glaucoma.
- Retrobulbar optic neuritis - eg, due to multiple sclerosis.
- Traumatic optic neuropathy.
- Lesions compressing the optic nerve (eg, tumour, aneurysms, Paget's disease of bone).

Primary retinal disease

- Central retinal artery occlusion or central retinal vein occlusion.

Secondary optic nerve disease

- Ischaemic optic neuropathy, which may be:
  - Arteritic ischaemic optic neuropathy - usually giant cell arteritis.
  - Non-arteritic anterior ischaemic optic neuropathy.
- Chronic papilloedema.
- Chronic optic neuritis.

Less common causes

- Hereditary optic neuropathies (eg, Leber's optic neuropathy).

- Toxic optic neuropathies:[5]
  - Methanol toxicity remains a significant problem in some parts of the world.
  - Possible drug causes of toxic optic neuropathy are disulfiram, halogenated hydroquinolones (amoebicides), ethambutol, isoniazid, chloramphenicol, vincristine and ciclosporin. Cimetidine has (rarely) been associated with optic neuropathy which reversed on stopping the drug.
  - People who abuse alcohol and tobacco and who are also malnourished are at greater risk, probably through deficiency of B-complex vitamins.
  - Metabolic disorders such as severe renal impairment may cause toxic optic neuropathy through build-up of toxins.
- Retinal degeneration (eg, retinitis pigmentosa).
- Retinal storage diseases (eg, Tay-Sachs disease).
- Radiation neuropathy.
- Syphilis.
- A rare autosomal dominant condition, optic atrophy 1 (also known as juvenile optic atrophy, or Kjer-type optic atrophy) is characterised by insidious onset of visual impairment in early childhood.[2]

Treatment

This depends on the associated disease.

Complications

Visual loss, the degree and nature of which will depend on the severity and type of underlying disease.

Prognosis

The optic nerve is a part of the central rather than the peripheral nervous system as it is derived from the diencephalon during embryonic development. Central nervous system nerve tracts are not capable of regeneration in the way that the peripheral nerves are. Optic atrophy is therefore irreversible.

Treatment, where available, will be aimed at limiting the progression of optic atrophy. The optic atrophy related to optic neuritis may, in some cases, be limited by the use of steroids.

How to prevent optic atrophy

Some causative conditions such as glaucoma and toxic, alcohol, tobacco and nutritional retinopathies can be limited by early diagnosis and optimal management of the underlying problem.

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