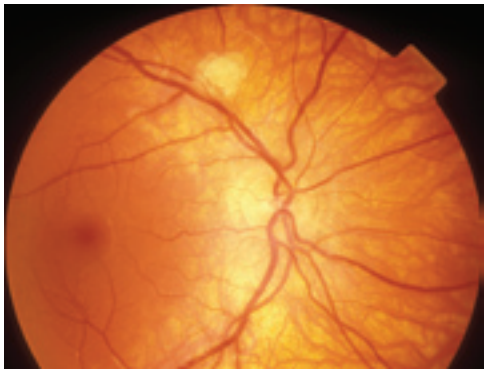
Neurofibromatosis type 2

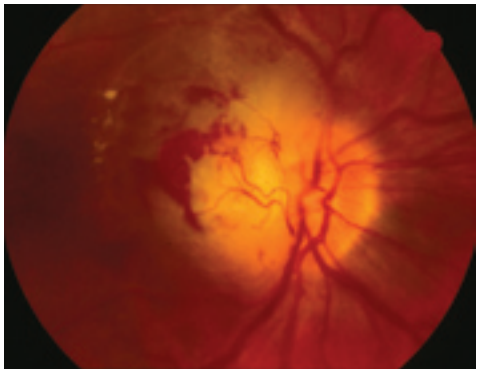
DESCRIPTION

Neurofibromatosis is one of the phakomatoses ('birthmarks'), the congenital or heritable conditions characterised by hamartomas and neoplasms throughout the body. The phakomatoses include neurofibromatosis, tuberous sclerosis, von Hippel-Lindau disease (retinal capillary haemangioma), Sturge-Weber syndrome, ataxia-telangiectasia and Wyburn-Mason syndrome.

Neurofibromatosis types 1 and 2 share some clinical features; and both are inherited in an autosomal dominant pattern. Type 2 (NF2) is characterised by bilateral acoustic neuromas and a high risk of cataract. The affected gene is on chromosome 22, and encodes a protein called Merlin, which has been implicated in cell surface remodelling and tumour suppression. Patients develop acoustic neuromas (vestibular schwannomas), which are often bilateral. Other central nervous system tumours are less common, and include neurofibromas, meningiomas and gliomas. The principal ocular complication is juvenile posterior sub capsular cataract, which affects two-thirds of patients and usually develops before age 30. Hamartomas of the retinal pigment epithelium and retina are common.



Retinal astrocytic hamartoma in the right superior fundus of a 10-year-old boy, appearing as a 'fish egg' like lesion. Tests for tuberous sclerosis and neurofibromatosis proved negative and this was presumed to be an isolated finding. Image courtesy of *Clinical and Experimental Optometry* journal



Optic nerve glioma with haemorrhage, associated with neurofibromatosis

SYMPTOMS

Acoustic neuromas typically present in the second or third decade with hearing loss, tinnitus and/or loss of balance. Symptoms of fifth, sixth or seventh cranial nerve dysfunction may ensue. Juvenile posterior sub-capsular cataracts may cause blurred vision or glare. As with NF1, *café au lait* patches are asymptomatic; and neurofibromas may cause cosmetic disfigurement or discomfort.

SIGNS

Café au lait spots occur in approximately two-thirds of patients. They are less numerous than in NF1, and tend to be restricted to the trunk. Cutaneous neurofibromas occur in approximately 30 per cent, while Lisch nodules are rare. (See Neurofibromatosis Type 1, *Optician*, September 21, 2007). Thickened corneal nerves, with a coarse silvery appearance, are more common in NF2 than NF1

Posterior sub-capsular cataracts have a characteristic granular appearance immediately anterior to the posterior lens capsule. Neurological examination, with particular attention to the cranial nerves, may reveal signs such as hearing or balance impairment, as in acoustic neuroma. Extraocular motility defects occur in 10 per cent of patients.

INCIDENCE

Very rare (1 per million). Optic nerve gliomas are rare (1/100,000).

SIGNIFICANCE

Cataract may cause significant loss of vision. Central nervous system neoplasms have the potential to substantially impair neurological function, and may require major surgery with its attendant risks.

SEE ALSO

Neurofibromatosis Type 1, Cataract, Tumours of the retinal pigment epithelium.

MANAGEMENT

Genetics

After the collection of a detailed family history and examination of all family members, the patient should be advised that, on average, half of their children will be affected by this autosomal dominant condition.

Additional investigations

Suspected central nervous system tumours are investigated with computed tomography (CT) or magnetic resonance imaging (MRI). Electroencephalogram (EEG) and audiography are employed in the preoperative assessment of acoustic neuroma.

Surgery

Several methods, involving surgery and radiotherapy, are employed in the removal of acoustic neuromas. They are best removed at an early stage for optimal preservation of hearing and facial nerve function.

REVIEW

Thorough neurological examination at regular intervals is appropriate.

The full series of these articles will be available in the book *Posterior Eye Disease and Glaucoma A-Z* by Bruce AS, O'Day J, McKay D and Swann P. £39.99. For further information click on the Bookstore at **opticianonline.net**

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