The Neurofibromatoses
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The neurofibromatoses (NF) are the most common member of a family of disorders (neurocutaneous disorders) which simultaneously affect the skin and nervous system. The characteristic lesion in this disorder is the neurofibroma which is generally benign and derived from neural tissue. Diagnoses are generally made in children and symptoms become progressive with age and growth of neurofibromas. Other neurocutaneous disorders include tuberous sclerosis, Sturge-Weber syndrome and von Hippel-Lindau disease. There are two main types of neurofibromatosis (Type I and II) which constitute separate entities and are best discussed separately.

Neurofibromatosis type I
Neurofibromatosis type I (NF-1) is also known as Von Recklinghausen's disease or peripheral NF. It accounts for up to 90% of NF cases and affects around 1 in 3000 live births. It is a genetic disorder which is inherited in an autosomal dominant pattern, but despite this, its manifestation can vary greatly and up to half of new cases are sporadic and do not have a family history. Most, if not all, cases will be diagnosed by the age of five.

The diagnosis is made based on its clinical manifestations. As its name suggests (peripheral NF) the majority of its lesions (neurofibromas) affect peripheral nerves in the skin or nerve plexuses or melanocytes. Therefore, it generally manifests with Café au Lait spots (dark brown skin spots), freckling in the groin or armpit, pigmented iris hamartomas, cutaneous neurofibromas (usually palpable or visible as skin lumps), plexiform neurofibromas (neurofibromas which are associated with nerve plexuses), tumours of the optic nerves and skull bone abnormalities. Although less common, it can affect the brain and spinal cord, and so may be associated with brain tumours (gliomas), epilepsy and cognitive impairment (though this is usually not severe).

The symptoms and therefore morbidity of NF-1 are often due to the plexiform neurofibromas (weakness, paralysis, numbness, pain), optic gliomas (visual disturbance), or peripheral neurofibromas (pain, numbness, weakness) which can cause spinal cord compression (paralysis or weakness). Plexiform neurofibromas can rarely (<3%) transform to become malignant.

Neurofibromatosis type II
Neurofibromatosis type II (NF-2) is also known as "Central Neurofibromatosis". Therefore, as its name suggests, neurofibromas in this type of NF always cause brain or spinal tumours. The neurofibromas characteristically manifest as bilateral acoustic neuromas (tumours of the hearing or balance nerves) and so progressive hearing loss is a common presentation. Patients may also develop multiple meningiomas and so other symptoms may include headaches, balance problems (ataxia), tinnitus, vertigo, facial weakness or numbness, and limb weakness or paralysis. Lesions can also affect the spinal cord (gliomas or ependymomas). NF-2 is also a genetic disorder and is inherited in an autosomal dominant fashion. It is far less common than NF-1, affecting only 1 in 50,000 live births and so accounts for around 10% of NF cases.

Treatment
The treatment revolves around symptom management. In cases where there are compressive lesions, surgery is the mainstay. Multiple surgeries may be required during the life of the patient. In those rare cases that become malignant, chemotherapy may be used. Genetic screening and counseling is available.

Prognosis

Although the genes have been identified in both NF-1 and NF-2, there is currently no cure. The lesions are generally benign but in rare cases (<3%) the neurofibromas can undergo malignant transformation.

Support and Information

There are several national organizations that provide support for those challenged by neurofibromatosis. Some include:

- Neurofibromatosis Association of Australia (NFAA)
  http://www.nfaa.org.au/support_local_support.htm
- Better Health Channel (VIC)
  http://www.betterhealth.vic.gov.au/bhcv2/bhcarticles.nsf/LFourPagesMoreInfo/Neurofibromatosis
- The Children's Tumor Foundation (CTF) (a non-profit organization working towards a cure for Neurofibromatosis in the United States)
- Neurofibromatosis Inc. (a non-profit organization working towards Neurofibromatosis patient support in the United States)
- Neurofibromatosis Cafe (a non-profit organization working providing patient education, awareness and support of Neurofibromatosis in the United States)