MANAGEMENT GUIDELINES FOR ADULTS WITH NEUROFIBROMATOSIS TYPE 1 (NF1)

Tumour Foundation of BC

EMPOWERING INDIVIDUALS WITH NF

OVERALL RECOMMENDATION

All adults with NF1 should be assessed by a physician at least once a year with particular attention paid to blood pressure and age-related complications. While all symptoms should be investigated promptly, additional routine screening of asymptomatic patients is not recommended. The disease mainly affects the skin and nervous system. However, there are many potential complications that must be monitored.

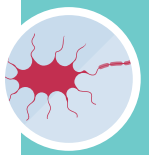
Referral recommendations are indicated where appropriate.

PSYCHOLOGICAL & LEARNING ISSUES

psychologist or psychiatrist

The pressure and stress of carrying a genetic disorder is considerable for both men and women, no matter how severely they are affected. If your patient needs counselling, be sure to refer to an appropriate mental health professional. Look for someone experienced in dealing with medical challenges.

Your patients with NF1 may be feeling anxious, socially isolated, or depressed. These symptoms may arise from cosmetic problems caused by neurofibromas and from the complex and unpredictable nature of the disease. As well, learning disabilities and ADHD are significantly more common in patients with NF1 than the general population (50% vs. 15%) and may have led to trauma in school. You should carefully evaluate symptoms and refer to a psychologist or psychiatrist as necessary. This need may be ongoing. Referral to the Tumour Foundation of BC may also help some patients.



NEUROLOGICAL ISSUES

neurologist or NF1 team

If your patient experience headaches that are not usual for them (seizures, visual disturbances, nerve pain or gait disturbances), be sure to request an immediate referral to a neurologist.

Patients with NF1 who experience headaches, seizures, visual disturbances, nerve pain, or gait disturbances need immediate referral to a neurologist or an NF1 team. They should be investigated for both cerebrovascular disease and brain tumours. Tumours (such as cerebral, spinal and optic pathway gliomas) require at least yearly MRIs for five years and then ongoing clinical monitoring.

BONE HEALTH

neurosurgeon or orthopedic surgeon

Men and women with NF1 have an increased risk of osteoporosis. Examine risk factors and suggest diet and exercise options, particularly if patient is female and close to menopause.

Some 10% of patients with NF1 have scoliosis and 5% will require surgery. Patients with scoliosis should be referred for spinal orthopedic assessment. Women with NF1 have an increased risk of osteoporosis at menopause in a large registry-based study, a fivefold increase in fracture risk was reported for adults older than 40 years.

CARDIOVASCULAR ISSUES

endocrinologist

People with NF1 often have high blood pressure. Be sure to check blood pressure at every visit, or at least once a year.
Note: Hypertension can begin in childhood in patients with NF1.

Blood pressure should be checked annually. If patient is hypertensive, consider renovascular lesions (especially in patients under 20) or phaeochromocytoma (at any age). In the case of the latter, refer to an endocrinologist. Essential hypertension should be treated as in the general population.

Other issues might arise, including: vascular abnormalities include arterial stenosis, moyamoya arteriopathy, aneurysms, arteriovenous malformations, cardiac valve abnormalities, and vessel compression and/or invasion by NF-related tumours.

TUMOURS

Examine lumps and bumps — anywhere on the body, whether painful or not — that are growing or changing.

Neurofibromas are a benign peripheral nerve sheath tumour. There are three main types:

1. **CUTANEOUS NEUROFIBROMAS**

   plastic or dermatological surgeon

   Patients will benefit from referral to a plastic or dermatological surgeon for removal.

2. **SUBCUTANEOUS NEUROFIBROMAS**

   NF1 specialist or soft tissue tumour/peripheral nerve surgeon

   Present in 15% of NF1 patients, be sure to seek advice from an NF1 specialist or a soft tissue tumour/peripheral nerve surgeon. This is because such removal may result in neurological deficits.

3. **PLEXIFORM NEUROFIBROMAS**

   NF1 specialist or soft tissue tumour/plastic surgeon

   Present in 30 to 50% of NF1 patients; problematic in about 6% of cases. Be sure to seek advice from an NF1 specialist or an experienced soft tissue tumour/plastic surgeon. Surgery can cause severe haemorrhage or encroach on vital organs. Chemotherapy is sometimes a therapeutic option for such tumours. Note that some patients can have many deep neurofibromas without symptoms.

   Note: People with plexiform neurofibromas are at increased risk of developing malignant peripheral nerve sheath tumours (MPNST). Signs of such a development include progressive/persistent pain and rapid growth.

VISION ISSUES

ophthamologist

Patients with NF1 may have experienced eye problems as children (especially optic nerve tumours). Whatever childhood vision issues have been reported, an urgent assessment is highly recommended if any visual symptoms present themselves.

GASTRO-INTESTINAL ISSUES

further investigation

Bloating, pain, constipation or bleeding can be caused by NF1.

Symptoms of bloating, pain, dyspepsia, constipation, bleeding or anemia may indicate a neurofibroma or a gastrointestinal stromal tumour in the digestive tract and should be investigated. Patients are also at an increased risk of carcinoid tumours (usually in the duodenum).

PREGNANCY

clinician with experience in NF1

Genetic testing prior to pregnancy will reveal the risk of a patient passing this disorder on to their children. Seek genetic testing if your patient is considering pregnancy.

If your patient wishes to become pregnant, it is important to seek advice from a clinician with experience in NF1. The severity of the condition cannot be predicted within families. All people with NF1 should receive genetic counselling prior to conception. In addition to the risks to the unborn child, risks to the patient include: renal artery stenosis or phaeochromocytoma as a cause for hypertension. Steps should be taken to ensure that pelvic neurofibromas do not impede delivery.

BREAST CANCER RISK IN WOMEN

Hereditary Breast Cancer program

Women age 30-50 years have an increased breast cancer risk of four to five-fold until 50 years (not substantially increased thereafter). Annual screening is recommended between the ages of 30 and 50 and becomes even more important if there is additional family history of breast cancer. Depending on availability in your community, the Hereditary Breast Cancer program may be able to provide screening via MRI so as to reduce the risk of additional exposure to radiation that a mammogram would entail. Please request this service for your patients.

FOR FURTHER INFORMATION:

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CITATIONS

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2 Friedman JM, Adam MP, Ardinger HH, Pagon RA, Wallace SE, Stephens K, & Amemiya A. (1998). *Neurofibromatosis 1*. PubMed. Retrieved from www.ncbi.nlm.nih.gov/pubmed/20301288

3 Sharif, S., Moran, A., Huson, S. M., Iddenden, R., Shenton, A., Howard, E., & Evans, D. G. R. (2007). *Women with neurofibromatosis 1 are at a moderately increased risk of developing breast cancer and should be considered for early screening*. Journal of Medical Genetics, 44(8), 481–484. doi: 10.1136/jmg.2007.049346

4 Stewart, D. R., Korf, B. R., Nathanson, K. L., Stevenson, D. A., & Yohay, K. (2018). *Care of adults with neurofibromatosis type 1: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG)*. Genetics in Medicine, 20(7), 671–682. doi: 10.1038/gim.2018.28

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