

Review checklist for Neurofibromatosis Type 1

Adult (over 16/18 years) – Annual Review Recommended

- All patients should be referred to a clinical genetics service at the time of (possible) diagnosis and the age of transition from paediatric to adult services.
- Adults should be evaluated annually by their primary care physician and ideally seen by a health profession with expertise in caring for people with NF1 once every three years.
- Individuals with complex NF should be seen at or cared for in consultation with a centre with expertise in managing NF1.
- **Record height, weight, and blood pressure at every visit.**

	WHAT TO LOOK FOR	WHEN TO REFER
IMAGING	At transition to adulthood (16-18 years) whole body-MRI (WB-MRI) to detect internal tumour load from plexiform neurofibromas including CNS-MRI as baseline for spinal tumour screening.	URGENT REFERRAL if symptomatic CNS glioma. REFERRAL to NF centre if high internal tumour load (increased risk for MPNST); or if incidental (asymptomatic) glioma.
PSYCHOLOGICAL BURDEN	Psychological burden is underestimated. Disfigurement may lead to feelings of social isolation and depression. Patients may be reluctant to talk about these issues and need encouragement.	Consider REFERRAL to relevant service for psychological assessment, intervention and/or support.
SKIN	Check for symptomatic lesions, plexiform neurofibroma and lumps requiring excision due to disfigurement.	URGENT REFERRAL to oncology or sarcoma surgeon for rapidly growing, painful or changing plexiform neurofibroma lesions. REFERRAL to dermatology or plastic surgeon for lesions needing removal for other reasons.
EYES	It is uncommon for optic pathway tumours to develop in adulthood. Visual signs/symptoms should be investigated urgently . Baseline ophthalmologist assessment if not done.	URGENT REFERRAL to ophthalmologist if there are concerns about the eyes or visual symptoms.
NEUROLOGICAL	Review for neurological symptoms, particularly new onset seizures, headaches, nerve pain and visual and gait disturbances . Consider nerve conduction studies to detect neuropathy and MRI for CNS or peripheral nerve sheath tumours.	REFERRAL to neurologist if increase in frequency and/or severity of headaches or onset of new symptoms.
BLOOD PRESSURE	Check blood pressure at least annually. If hypertensive , consider renovascular lesions (usually <20 years) or phaeochromocytoma (any age).	REFERRAL to endocrinologist if phaeochromocytoma is a possibility; or nephrologist for renovascular assessment.
WOMEN	Women have an increased chance of breast cancer between age 30 to age 50 years.	REFERRAL to breast specialist from age 30 years. Annual surveillance (MRI preferred) from age 30 to age 50 years. After age 50, surveillance as per population screening.
PREGNANCY	Prenatal & preimplantation testing is available but preconception genetic work-up needed. During pregnancy, neurofibromas may increase in size and/or itchiness. Consider phaeochromocytoma/renal artery stenosis in pregnant women with particularly high BP, especially if it persists post-delivery.	REFERRAL for men and women to clinical genetics prior to conception to discuss the chance of NF1 for offspring and to discuss pregnancy planning options.
ANY OTHER NEW SYMPTOMS	Consider other possible complications and increase risk for malignancy in NF1, including increased risk of gastro-intestinal stromal tumours (GIST) and other tumours.	REFERRAL to relevant specialist, preferably with NF1 expertise, and oncology expertise.

UNSURE? Contact your patient's paediatrician, other relevant specialist or your local clinical genetics service.

This care pathway is adapted for use by:

Contact details are: