

REVIEW CHECKLIST for Neurofibromatosis Type 1

- At time of (possible) diagnosis patients should be referred to a clinical genetics service.
- **Children** with NF1 should be managed by a paediatrician and/or multidisciplinary team.
- Most **adults** with NF1 can be managed by their general practitioner with referral to specialised services as needed. Refer more complex cases/problems to a specialist/multidisciplinary team familiar with NF1.
- Patients with **mosaic or segmental NF1** may require less intensive review depending on their individual clinical features.

Children (0-16 years)

Record height, weight, head circumference and blood pressure at every visit.

WHAT TO LOOK FOR

WHEN TO REFER

SKIN

Neurofibromas – can be itchy, and sometimes tender. May be cutaneous or subcutaneous.
Plexiform neurofibromas – note location, appearance, size and hardness.
Monitor large areas of café au lait pigmentation &/or excessive hair growth for development of a plexiform neurofibroma.

URGENT REFERRAL to oncology or sarcoma surgeon for rapidly growing, painful or changing lesions.

SKELETON

Scoliosis – look for signs during entire growth period, but especially at puberty and during adolescent growth spurts.
Pseudoarthrosis – tibia most commonly affected but radius and ulna may be involved.

REFERRAL to orthopaedic surgeon if any curvature or bowing.

EYES

Regular ophthalmic review from 0-16 years (6-monthly to age 4, then 12-monthly to age 16). Specifically assess for **squint, proptosis** or **reduced visual acuity**.

URGENT REFERRAL to ophthalmologist if there are concerns about the eyes or visual symptoms.

NEUROLOGICAL

Review for neurological symptoms, particularly **ataxia, headaches, disturbed level of consciousness** and **visual disturbance**.

REFERRAL to neurologist if increase in frequency and/or severity of headaches or onset of new symptoms.

DEVELOPMENT

Review development.
 Assess for **coordination** and **speech difficulties**.
 There may be short stature & macrocephaly.
 Investigate **precocious** or **late puberty**.

REFERRAL to relevant specialist if concerns.

EDUCATION & BEHAVIOUR

There is an increased incidence of **learning** and **behaviour difficulties** in NF1 (particularly attention difficulties, ADHD and autism spectrum disorder).
 Identify possible special needs and appropriate resources to assess and address them.

Consider **REFERRAL** to relevant service for psychological assessment, intervention and/or support.

UNSURE? Contact your patient's paediatrician, other relevant specialist or your local clinical genetics service.
 In [enter State] clinical genetics services are available [enter details of service and how to contact]

TO BE ADJUSTED BY SERVICE USING THE CHECKLIST

REVIEW CHECKLIST for Neurofibromatosis Type 1

ADULT (over 16 years)

Record height, weight and blood pressure at every visit.

WHAT TO LOOK FOR

WHEN TO REFER

PSYCHOLOGICAL BURDEN

Psychological burden is underestimated. Disfigurement may lead to feelings of social isolation, and depression. Psychological problems are common but 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 for non-medical reasons.

URGENT REFERRAL to oncology or sarcoma surgeon for rapidly growing, painful or changing lesions.
REFERRAL to dermatology or plastic surgeon for removal of lesions needing removal for other reasons.

EYES

It is uncommon for optic pathway tumours (OPT) to develop in adulthood. Visual signs/symptoms should be investigated. 3- to 5-yearly examination by ophthalmologist if no tumour arisen during childhood.

URGENT REFERRAL to ophthalmologist if there are concerns about the eyes or visual symptoms.

NEUROLOGICAL

Review for neurological symptoms, particularly **headaches, nerve pain and visual and gait disturbances.**

REFER 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).

REFER to endocrinologist if phaeochromocytoma is a possibility.

WOMEN

Women have an increased chance of **breast cancer**, particularly between ages 30-50. Classified as moderate risk.

REFER to breast specialist by age 30 years
annual surveillance 30-50 yrs
30-40 yrs: MRI +/- US
40-50 yrs: MRI +/- MMG, US
2nd yearly surveillance >50 yrs
MMG +/- US

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.

REFER men and women to clinical genetics prior to conception to discuss risk of NF1 for offspring and reproductive options.

ANY OTHER NEW SYMPTOMS

Consider other possible complications

REFER to relevant specialist.

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