REVIEW CHECKLIST
Neurofibromatosis Type 1

Recommended Management

- At time of diagnosis, or possible diagnosis, ALL patients should be seen in a genetics department.
- All **children** with NF1 should be managed by local specialists (e.g. paediatricians and/or a multidisciplinary team).
- **Adults** with NF1 could be managed by their general practitioner with referral to specialised services as necessary. More complex cases could be referred to a NF1 clinic or multidisciplinary team who have extensive experience with the condition.
- Individuals with **mosaic or segmental NF1** may require less intensive review depending on their clinical presentation.

CHILDREN (0-16 years)

Record **height**, **weight** and **head circumference**. Take **blood pressure** as soon as feasible.

	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.	Rapidly growing, painful or changing lesions: URGENT REFERRAL by specialised NF1 service/ specialist sarcoma team.
SKELETON	**Scoliosis** – look for signs during entire growth period, and especially at puberty and during adolescent growth spurts. **Pseudoarthosis** – tibia most commonly affected but radius and ulna may be involved.	Any curvature or bowing – REFER to orthopaedic surgeon.
EYES	Have regular ophthalmic reviews taken place for those aged 0-16 years? (6 monthly to age 4, 12 monthly to age 16) Is there any evidence of a **squint, proptosis,** or **reduced visual acuity?**	URGENT REFERRAL to ophthalmologist if there are concerns about the eyes or visual symptoms.
NEUROLOGICAL	Neurological symptom review, particularly **ataxia, headaches, loss of consciousness** and **visual disturbance.**	REFER to specialised NF1 service or neurologist if increase in frequency and/or severity of headaches or onset of other symptoms.
DEVELOPMENT	Review development – noting in particular **coordination** and **speech difficulties.** There may be short stature & macrocephaly. **Precocious** or **late puberty** should be investigated.	Consider REFERRAL to paediatric specialist.
EDUCATION & BEHAVIOUR	There is an increased incidence of **learning** and **behaviour** (particularly attention difficulties, **ADD**, **ADHD and ASD**) problems. Identify possible special needs and appropriate resources to assess them.	Consider REFERRAL for professional assessment of educational needs.

UNSURE? Do not hesitate to contact your NF1 managing doctor if you have any queries- contact details are at the bottom of page 2

REVIEW CHECKLIST
Neurofibromatosis Type 1

ADULTS (>16 years)

	WHAT TO LOOK FOR	WHEN TO REFER
SKIN	Check for **symptomatic lesions, plexiform neurofibromas,** any lumps requiring excision for non-medical reasons.	Rapidly growing, painful or changing lesions: URGENT REFERRAL to specialised NF1 service or specialist sarcoma team. Lesions being removed for other reasons need referral to plastic surgeon or dermatologist.
PSYCHOLOGICAL BURDEN	Effects are underestimated. Disfigurement may lead to feelings of social isolation, and depression. **Psychological problems** are common but patients, both men and women, may be reluctant to talk about these issues and need encouragement.	Consider REFERRAL to an appropriate counselling service and/or a plastic surgeon & dermatologist.
NEUROLOGICAL	Neurological symptom review, particularly **headaches, nerve pain,** and **visual and gait disturbances.**	REFER to specialised NF1 service or neurologist if increase in frequency and/or severity of headaches or onset of other symptoms.
BLOOD PRESSURE	Check blood pressure annually. If **hypertensive** consider **renovascular lesions** (usually <20 years) or **phaeochromocytoma** (any age).	REFER to endocrinologist if phaeochromocytoma is a possibility.
EYES	Optic Pathway tumours UNCOMMON in adults but any unusual visual signs/symptoms warrant investigation. 3-5 yearly examination by ophthalmologist recommended if no tumour arisen during childhood.	URGENT REFERRAL to ophthalmologist if there are concerns about the eyes or visual symptoms.
WOMEN	Women have an increased risk of developing **breast cancer,** particularly between the ages of 35-50. Classified as moderate risk.	REFER to breast screening centre. 35-40 years: MRI +/- US 40-50: MRI +/- MMG, US >50: MMG +/-US
PREGNANCY	Pre-natal and pre-implantation testing is available but relies on pre-pregnancy genetic work-up. During pregnancy, neurofibromas may increase in size and/or itchiness. Consider phaeochromocytoma/renal artery stenosis in women with particularly high BP, especially if it persists post-delivery.	Women who are planning pregnancy should be REFERRED to clinical genetics.
ANY OTHER NEW SYMPTOMS	Consider other possible complications.	REFER to appropriate specialist.

Do not hesitate to contact the Neurofibromatosis clinic, Department of Clinical Genetics, Royal North Shore Hospital. Email: NSLHD-ClinicalGenetics@health.nsw.gov.au. Phone: 02 9463 1727
Adapted by the RNSH team, with permission of the Manchester NF service. Version 1 March 2017.