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.

**WHAT TO LOOK FOR**

**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.

**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.
- 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?

**EYES**
- Neurological symptom review, particularly **ataxia**, headaches, loss of consciousness and visual disturbance.

**NEUROLOGICAL**
- Review development – noting in particular **coordination** and **speech difficulties**. There may be short stature & macrocephaly. **Precocious** or **late puberty** should be investigated.

**DEVELOPMENT**
- 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.

**EDUCATION & BEHAVIOUR**
- **Rapidly growing, painful** or changing lesions: **URGENT REFERRAL** by specialised NF1 service/specialist sarcoma team.
- **Any curvature or bowing** – **REFER** to orthopaedic surgeon.
- **URGENT REFERRAL** to ophthalmologist if there are concerns about the eyes or visual symptoms.
- **REFER** to specialised NF1 service or neurologist if increase in frequency and/or severity of headaches or onset of other symptoms.
- **Consider REFERRAL** to paediatric specialist.
- **Consider REFERRAL** for professional assessment of educational needs.

**REVIEW CHECKLIST Neurofibromatosis Type 1**

**CHILDREN (0-16 years)**

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

**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

SKIN
Check for symptomatic lesions, plexiform neurofibromas, any lumps requiring excision for non-medical reasons.

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.

NEUROLOGICAL
Neurological symptom review, particularly headaches, nerve pain, and visual and gait disturbances.

BLOOD PRESSURE
Check blood pressure annually. If hypertensive consider renovascular lesions (usually <20 years) or pheochromocytoma (any age).

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.

WOMEN
Women have an increased risk of developing breast cancer, particularly between the ages of 35-50. Classified as moderate risk.

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 pheochromocytoma/renal artery stenosis in women with particularly high BP, especially if it persists post-delivery.

ANY OTHER NEW SYMPTOMS
Consider other possible complications.

WHEN TO REFER

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.

Consider REFERRAL to an appropriate counselling service and/or a plastic surgeon & dermatologist.

REFER to specialised NF1 service or neurologist if increase in frequency and/or severity of headaches or onset of other symptoms.

REFER to endocrinologist if pheochromocytoma is a possibility.

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

REFER to breast screening centre. 35-40 years: MRI +/- US 40-50: MRI +/- MMG, US >50: MMG +/-US

Women who are planning pregnancy should be REFERRED to clinical genetics.

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.