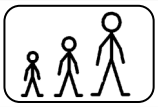
Version 18. 01/19

Neurofibromatosis Type 1

Review Guidelines



Annual Review Recommended

At time of diagnosis, or possible diagnosis, ALL patients should be seen in a genetics department. Those with significant complications will be followed up as appropriate through the nationally-funded Complex NF1 Service. Annual review should be undertaken by a Community/District Paediatrician and GP throughout childhood, and by a GP in adulthood. Patients, paediatricians and GPs have telephone access to the NF Service in Genetic Medicine for NF-related concerns.

AGE	GENETICS APPOINTMENT	NF1 REVIEWS CARRIED OUT BY	VISION CHECKS
<6 & 50% risk	In first year and then at 2 and 5*[1].	Care coordinated by Genetics.	Symptom check at NF1 review
<8 affected	Confirmation of diagnosis & assessment. Genetic counselling for family.	GP and Community/ District Paediatrician. Liaison with NF service for complex cases.	At least annual with paediatric ophthalmologist.
8 – 15 affected	On request		Annual with optician/orthoptist
16—18 affected	Appointment for counselling re: adult complications and genetics	Care coordinated by GP.	Symptom check at NF1 review
>16 affected*[2]	On request		

*[1] **If no café au lait spots by 5 years, NF1 can be excluded in the majority of NF1 families.**
Mutation testing can be considered to confirm or exclude the diagnosis and clarify the need follow up

*[2] **Women aged 40—50 should be referred for annual mammography as per 'moderate risk' NICE guidelines.**

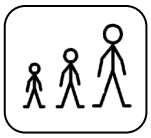
Review Checklist—Children (0—16)

Record **height, weight** and **head circumference.** Take **blood pressure** as soon as feasible.
If raised, see the Adult Review Checklist (p.2) for info.

	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 to Complex NF1 Service or Specialist sarcoma team**
SKELETON	**Scoliosis–** look for signs during entire growth period, and especially at puberty and during adolescent growth spurts. **Pseudarthrosis** - tibia most commonly affected but radius and ulna may be involved.	**Any curvature or bowing- REFER to NF1 specialist orthopaedic surgeon**
EYES	Have regular ophthalmic reviews taken place for those aged 0-7 years? Is there any evidence of a **squint, proptosis,** or **reduced visual acuity?**	**URGENT REFERRAL to ophthalmologist if there are concerns about the eye or visual symptoms**
NEUROLOGICAL	Neurological symptom review, particularly **ataxia, seizures, headaches** and **visual disturbance.**	**REFER to Complex 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 the NF1 team if you have any queries—contact details are at the bottom of p.2

Neurofibromatosis Type 1
Review Checklist—Adults (16+)



	WHAT TO LOOK FOR	WHEN TO REFER
GENERAL	Check for **symptomatic (painful, hard, rapidly enlarging or affecting function) subcutaneous or plexiform neurofibromas.** Cutaneous neurofibromas causing distress or irritation should also be identified.	**Rapidly growing, painful or changing lesions: URGENT REFERRAL to Complex 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, seizures, nerve pain,** and **visual and gait disturbances.**	**REFER to Complex NF1 Service or neurologist if increase in frequency and/or severity of headaches or onset of other symptoms.**
BLOOD PRESSURE	Check blood pressure. If **hypertensive** consider **renovascular lesions** (usually <20 years) or **phaeochromocytoma** (any age).	**REFER to endocrinologist if phaeochromocytoma is a possibility**
EYES	Newly symptomatic optic pathway tumours UNCOMMON in adults but any unusual visual signs/ symptoms need investigation.	**URGENT REFERRAL to ophthalmologist if there are any concerns about the eyes or visual symptoms**
WOMEN	Women with NF1 have an increased risk of developing **breast cancer** between the ages of 40 and 50, classified as ''moderate'' (between 3 and 8% according to NICE guidelines).	**REFERRAL to local breast screening centre for annual mammography from 40 years.**
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.	**Couples considering reproductive options should be REFERRED to clinical genetics**
ANY OTHER NEW SYMPTOMS	Relative risk of many tumours and other medical issues may be increased in NF1.	**Consider appropriate REFERRAL**

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