Neurofibromatosis Type 1
Review Guidelines

Annual Review Recommended
At time of diagnosis, or possible diagnosis, ALL patients should be seen in a genetics department. 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 | *If no café au lait spots by 5 years, NF1 can be excluded in the majority of NF1 families.

*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.

WHEN TO REFER
Rapidly growing, painful or changing lesions: URGENT REFERRAL to Complex NF1 Service or Specialist sarcoma team.
Any curvature or bowing: REFER to orthopaedic surgeon
URGENT REFERRAL to ophthalmologist if there are concerns about the eye or visual symptoms
REFER to Complex 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.

WHAT TO LOOK FOR
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.
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.
Have regular ophthalmic reviews taken place for those aged 0-7 years? Is there any evidence of a squint, proptosis, or reduced visual acuity?
Neurological symptom review, particularly ataxia, headaches, loss of consciousness and visual disturbance.
Review development - noting in particular coordination and speech difficulties. There may be short stature & macrocephaly. Precocious or late puberty should be investigated.
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.

UNSURE? Do not hesitate to contact the NF1 team if you have any queries—contact details are at the bottom of p.2.
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 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, 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
Optic pathway tumours UNCOMMON in adults but any unusual visual signs/symptoms warrant 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 - in practice very few requests for testing are received. 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.