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 and 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 (overleaf) 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 and/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 overleaf...

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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.**

Dr. Grace Vassallo, January 2019

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