

Neurofibromatosis Type 1

Review Guidelines for Adults

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 for adults continued overleaf

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 pheochromocytoma (any age).



REFER to endocrinologist if pheochromocytoma 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 pheochromocytoma/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

Unsure? Do not hesitate to contact the NF1 team if you have any queries



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