

# Review Checklist

## Neurofibromatosis Type 1



### Recommended Review Intervals

At time of diagnosis, or possible diagnosis, ALL patients should be reviewed by clinical genetics.

AGE	RECOMMENDED REVIEW PERIOD	GENERAL NF1 REVIEWS CARRIED OUT BY	VISION CHECKS
<8 Affected	Annual (after confirmation of diagnosis & assessment by clinical genetics)	Community or District Paediatrician coordinates care. Liaison with NF clinic for complex cases	At least annual with paediatric ophthalmologist.
<6 & 50% risk	In first year and then at 2 and 5 <sup>+</sup>		
8 – 15	Annual	Care coordinated by NF clinic – All patients and their GPs have phone/clinic access for review of NF1 related concerns.	Symptom check and basic eye examination when patient has their NF1 review.
16 – 25	Annual		
25 – 40	2 Years #		
> 40*	3 Years #		
*If no café au lait spots by 5 years, NF1 can be excluded in the majority of NF1 families			
# >25 and high disease burden – Annual Review			
* Women should be referred for annual mammography between the ages of 40 and 50 as per 'moderate risk' NICE guidelines.			

### CHILDREN (0 - 16 years)

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

#### 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 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 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 eyes or visual symptoms.

NEUROLOGICAL

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

**REFER** to 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 and ADHD**), 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 in clinical genetics if you have any queries**

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