

# REVIEW CHECKLIST

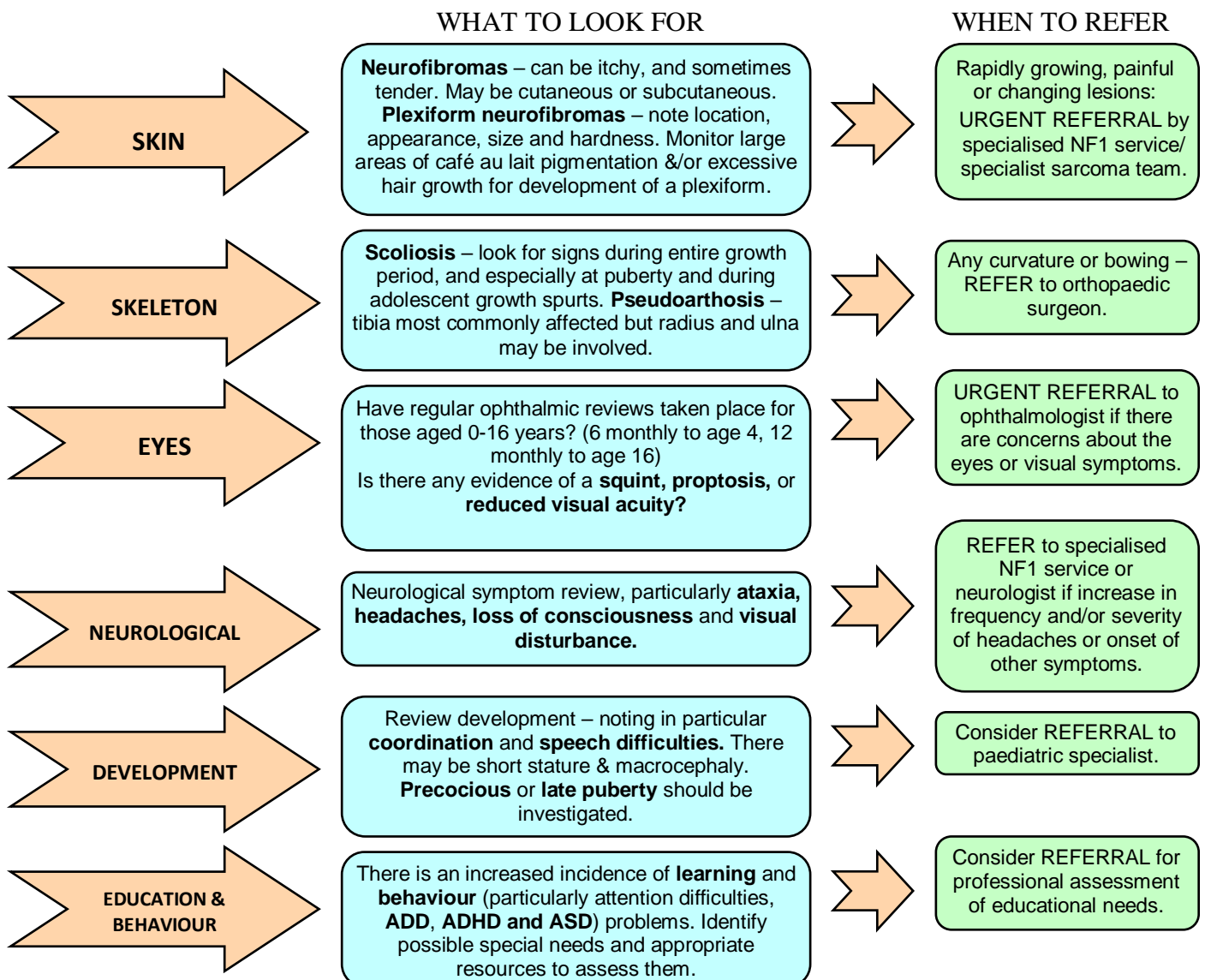
## Neurofibromatosis Type 1

### Recommended Management

- At time of diagnosis, or possible diagnosis, ALL patients should be seen in a genetics department.
- All **children** with NF1 should be managed by local specialists (e.g. paediatricians and/or a multidisciplinary team).
- **Adults** with NF1 could be managed by their general practitioner with referral to specialised services as necessary. More complex cases could be referred to a NF1 clinic or multidisciplinary team who have extensive experience with the condition.
- Individuals with **mosaic or segmental NF1** may require less intensive review depending on their clinical presentation.

### CHILDREN (0-16 years)

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



**UNSURE?** Do not hesitate to contact your NF1 managing doctor if you have any queries- contact details are at the bottom of page 2

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## Neurofibromatosis Type 1

### ADULTS (>16 years)

#### WHAT TO LOOK FOR

#### WHEN TO REFER

<b>SKIN</b>	Check for <b>symptomatic lesions, plexiform neurofibromas</b> , any lumps requiring excision for non-medical reasons.	Rapidly growing, painful or changing lesions: <b>URGENT REFERRAL</b> to specialised NF1 service or specialist sarcoma team. Lesions being removed for other reasons need referral to plastic surgeon or dermatologist.
<b>PSYCHOLOGICAL BURDEN</b>	Effects are underestimated. Disfigurement may lead to feelings of social isolation, and depression. <b>Psychological problems</b> 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.
<b>NEUROLOGICAL</b>	Neurological symptom review, particularly <b>headaches, nerve pain, and visual and gait disturbances</b> .	REFER to specialised NF1 service or neurologist if increase in frequency and/or severity of headaches or onset of other symptoms.
<b>BLOOD PRESSURE</b>	Check blood pressure annually. If <b>hypertensive</b> consider <b>renovascular lesions</b> (usually <20 years) or <b>phaeochromocytoma</b> (any age).	REFER to endocrinologist if phaeochromocytoma is a possibility.
<b>EYES</b>	Optic Pathway tumours UNCOMMON in adults but any unusual visual signs/symptoms warrant investigation. 3-5 yearly examination by ophthalmologist recommended if no tumour arisen during childhood.	<b>URGENT REFERRAL</b> to ophthalmologist if there are concerns about the eyes or visual symptoms.
<b>WOMEN</b>	Women have an increased risk of developing <b>breast cancer</b> , particularly between the ages of 35-50. Classified as moderate risk.	REFER to breast screening centre. 35-40 years: MRI +/- US 40-50: MRI +/- MMG, US >50: MMG +/-US
<b>PREGNANCY</b>	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.	Women who are planning pregnancy should be REFERRED to clinical genetics.
<b>ANY OTHER NEW SYMPTOMS</b>	Consider other possible complications.	REFER to appropriate specialist.

Do not hesitate to contact the Neurofibromatosis clinic, Department of Clinical Genetics, Royal North Shore Hospital. Email: [NSLHD-ClinicalGenetics@health.nsw.gov.au](mailto:NSLHD-ClinicalGenetics@health.nsw.gov.au). Phone: 02 9463 1727

Adapted by the RNSH team, with permission of the Manchester NF service. Version 1 March 2017.