

## Neurofibromatosis Type 1

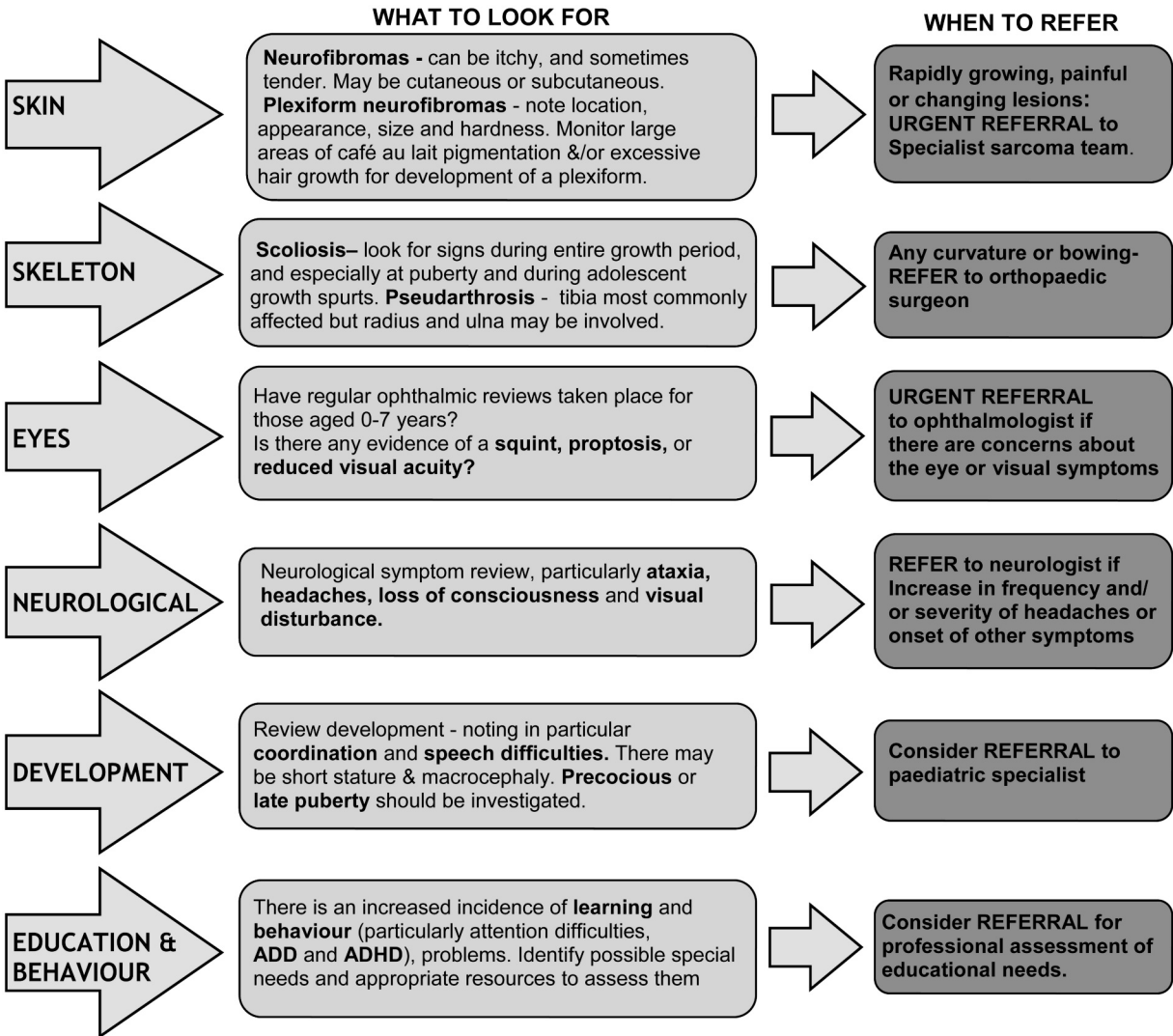
### Recommended Review Intervals

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

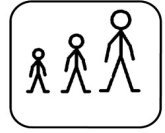
| AGE   | RECOMMENDED REVIEW PERIOD  | GENERAL NF1 REVIEWS CARRIED OUT BY   | VISION CHECKS  |
|---|--|--|--|
| <8 Affected   | Annual (after confirmation of diagnosis & assessment by clinical genetics) | Paediatrician coordinates care. Liaison with NF clinic for complex cases.  | At least annual with paediatric ophthalmologist.                           |
| <6 & 50% risk   | In first year and then at 2 and 5 <sup>+</sup>                             |  |  |
| 8 – 15  | Annual   | Care coordinated by GP – 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



**Unsure? Do not hesitate to contact our Helpline: +353 85 702 0024**



### ADULTS (> 16 years)

#### WHAT TO LOOK FOR

#### WHEN TO REFER

|                                      |  |   |
|--------------------------------------|--|---|
| <p><b>SKIN</b></p>                   | <p>Check for <b>symptomatic lesions, plexiform neurofibromas</b>, any lumps requiring excision for non-medical reasons</p>   | <p>Rapidly growing, painful or changing lesions: <b>URGENT REFERRAL to Specialist sarcoma team</b>. Lesions being removed for other reasons need referral to plastic surgeon or dermatologist</p> |
| <p><b>PSYCHOLOGICAL BURDEN</b></p>   | <p>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.</p>  | <p>Consider <b>REFERRAL to an appropriate counselling service and/or a plastic surgeon &amp; dermatologist</b>.</p>   |
| <p><b>NEUROLOGICAL</b></p>           | <p>Neurological symptom review, particularly <b>headaches, nerve pain, and visual and gait disturbances</b>.</p>   | <p><b>REFER to neurologist if increase in frequency and/or severity of headaches or onset of other symptoms.</b></p>  |
| <p><b>BLOOD PRESSURE</b></p>         | <p>Check Blood pressure. If <b>hypertensive</b> Consider <b>renovascular lesions</b> (usually &lt;20 years) or <b>phaeochromocytoma</b> (any age)</p>  | <p><b>REFER to endocrinologist if phaeochromocytoma is a possibility</b></p>  |
| <p><b>EYES</b></p>                   | <p>Optic pathway tumours <b>UNCOMMON</b> in adults but any usual visual signs/symptoms warrant investigation</p>   | <p><b>URGENT REFERRAL to ophthalmologist if there are any concerns about the eyes or visual symptoms</b></p>  |
| <p><b>WOMEN</b></p>                  | <p>Women with NF1 have an increased risk of developing <b>breast cancer</b> between the ages of 40 and 50, classified as "moderate" (between 3 and 8% according to NICE guidelines).</p>   | <p><b>REFERRAL to local breast screening centre for annual mammography from 40 years.</b></p>   |
| <p><b>PREGNANCY</b></p>              | <p><b>Pre-natal and pre-implantation testing</b> 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, particularly if it persists post-delivery.</p> | <p><b>Women who are planning pregnancy should be REFERRED to clinical genetics</b></p>  |
| <p><b>ANY OTHER NEW SYMPTOMS</b></p> | <p>Consider other possible complications</p>   | <p><b>REFER to appropriate specialist</b></p>   |

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