Northern Sydney Local Health District

Review checklist for Neurofibromatosis Type 1

Children (0–16/18 years) – Annual Review Recommended

- All patients should be referred to a clinical genetics service at the time of (possible) diagnosis and the age of transition from paediatric to adult services.
- Children should be evaluated by a health professional with expertise in caring for people with NF1 annually to age 10 years, then once every two years from age 10 years. Children over age 10 years should be evaluated annually by their paediatrician or primary care physician.
- Individuals with complex NF should be seen at or cared for in consultation with a centre with expertise in managing NF1.
- **Record height, weight, head circumference and blood pressure at every visit.**

	WHAT TO LOOK FOR	WHEN TO REFER
SKIN	**Neurofibromas** – can be itchy, or tender. May be cutaneous or subcutaneous. **Plexiform neurofibromas** – note location, appearance, size, hardness and any features of malignancy. Large areas of café au lait pigmentation &/or excessive hair may suggest a plexiform neurofibroma.	**URGENT REFERRAL** to oncology or sarcoma surgeon for rapidly growing, painful or changing lesions. **REFERRAL** to dermatologist or plastic surgeon for skin lesions needing removal.
SKELETON	**Scoliosis** – look for signs , especially at puberty. Dystrophic scoliosis may present earlier. **Pseudoarthosis** – tibia (mostly) or radius and ulna. **Decreased bone density** more frequent. Optimise Vit D. **Sphenoid dysplasia** may present with facial asymmetry.	**REFERRAL** to orthopaedic surgeon if any curvature or bowing. **REFERRAL** to craniofacial surgeon if facial asymmetry.
EYES	Specifically assess for **squint, proptosis** or **reduced visual acuity.** At least yearly ophthalmic review to age 8 years (6-monthly preferable); annually 9–18 years.	**URGENT REFERRAL** to ophthalmologist if there are concerns about the eyes or visual symptoms.
NEUROLOGICAL	Review for neurological symptoms, particularly **new onset or change in seizures, ataxia, headaches, disturbed level of consciousness** and **visual disturbance.**	**REFERRAL** to neurologist if increase in frequency and/or severity of headaches or onset of new symptoms.
DEVELOPMENT	Review development. Assess for **coordination** and **speech difficulties**. There may be short stature & macrocephaly. Investigate **precocious** or **late puberty.**	**REFERRAL** to relevant specialist if concerns.
EDUCATION & BEHAVIOUR	There is an increased incidence of **learning** and **behavioural difficulties** (particularly attention difficulties, ADHD and autism spectrum disorder). Identify needs and appropriate resources to assess and address them.	Consider **REFERRAL** to relevant service for psychological assessment, intervention and/or support.
IMAGING	At transition to adulthood (16-18 years) whole body-MRI (WB-MRI) to detect internal tumour load from plexiform neurofibromas including CNS-MRI as baseline for spinal tumour screening.	**URGENT REFERRAL** if symptomatic CNS glioma. **REFERRAL** to NF centre if high internal tumour load (increased risk for MPNST); or if incidental (asymptomatic) glioma.

UNSURE? Contact your patient's paediatrician, other relevant specialist or your local clinical genetics service.

This care pathway is adapted for use by: .....

Contact details are: .....

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Based on ERN GENTURIS guidelines and pathways (www.genturis.eu) and similar checklists by Royal North Shore Hospital and Manchester Neurofibromatosis services.
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