Surveillance protocol for tumour screening / identification in individuals with neurofibromatosis type 1

This guideline for tumour management in Neurofibromatosis type 1 has been drawn from the best available evidence and the consensus of experts in this area and it is regularly updated to reflect changes in evidence.

The expectation is that clinicians will follow this guideline unless there is a compelling clinical reason to undertake different management, specific to an individual patient.

European Reference Network
for rare or low prevalence complex diseases
Network Genetic Tumour Risk Syndromes (ERN GENTURIS)

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	Surveillance	Interval	Age (years) / indication	Strength*	Refer^
Optic pathway glioma	Clinical assessment: 1. Visual assessment 2. Fundoscopy 3. Visual fields 4. Optic coherence tomography	1-3: At least yearly 4: When feasible	0 - 8	1. Strong 2. Strong 3. Moderate 4. Moderate	7.2 & 9.2 (rec. 1-4)
	Visual screening	Yearly	8 – transition adolescence to adult	Moderate	7.2 & 9.2 (rec. 5-6)
Brain or spine glioma	Patient history / Examination signs of brain tumours	Every visit	All ages	Moderate	7.3 & 9.3 (children) 7.4 & 9.4 (adults)
Plexiform neurofibroma	Clinical examination	Every visit	All ages	Moderate	7.5 & 9.5 (rec. 1-2)
	Whole body MRI	Once	Transition adolescence -adult	Weak	7.5 & 9.5 (rec. 3-4)
MPNST + ANNUBP	Clinical examination + history taking	Every visit	All ages	Strong	7.6 & 9.6 (rec. 1-2)
	Regional MRI combined with ^{18}FDG PET MRI or ^{18}FDG PET CT	On indication	Suspicion for malignancy	Moderate	7.6 & 9.6 (rec. 3)
Orbital & Periorbital Plexiform neurofibroma	Clinical assessment, refraction error, vision fields, ocular motility	Every visit	All ages	Strong	7.7 & 9.7 (rec. 1)
Cutaneous neurofibroma	Clinical examination	Every visit	All ages	Strong	7.8 & 9.8 (rec. 1)
Gastrointestinal stromal tumour	Clinical examination + history taking	Every visit	Adolescence and adults	Moderate	7.9 & 9.9 (rec. 1-2)
	Abdominal MRI or CT	On indication	Clinical suspicion of presence based on symptoms	Moderate	7.9 & 9.9 (rec. 4)
Phaeochromocytoma and paraganglioma	Biochemical screening	On indication	Raised blood pressure	Moderate	7.10 & 9.10 (rec. 2)
	Biochemical screening	On indication	Pregnant women Consider if elective surgery requiring general anaesthesia	Weak	7.10 & 9.10 (rec. 1 and 3)
Breast cancer	MRI	Yearly	30 - 50	Moderate	7.11 & 9.11 (rec. 2-3)
	Breast screening per national guideline for the general population		> 50	Moderate	7.11 & 9.11 (rec. 2-3)
Glomus tumours of the digits	Screening for symptoms and visual inspection	Every visit	All ages, clinical suspicion	Moderate (Age, weak)	7.12 & 9.12 (rec. 1-3)
Juvenile myelomonocytic leukaemia	As part of normal clinical routine: patient history and physical examination	Every visit	<12	Moderate	7.13 & 9.13 (rec. 1-2)
Psychosocial needs	Psychosocial wellbeing and neuropsychological functioning	Every visit	All ages	Weak	7.14 & 9.14 (rec.1-3)

* This grading is based on published articles and expert consensus: strong – expert consensus AND consistent evidence, moderate – expert consensus WITH inconsistent evidence AND/OR new evidence likely to support the recommendation, weak – expert majority decision WITHOUT consistent evidence. ^ If manifestation is found, please refer to the following chapters in the guideline for management and treatment of observed manifestation. MPNST = Malignant peripheral nerve sheath tumour, ANNUBP = Atypical neurofibromatous neoplasm with uncertain biologic potential. Note. MRI = magnetic resonance imaging; 18FDG PET MRI = 18F-fluorodeoxyglucose positron emission tomography magnetic resonance imaging; 18FDG PET CT = 18F-fluorodeoxyglucose positron emission tomography computed tomography; CT = computed tomography.