ERN GENTURIS Plain Language Summary:

CLINICAL PRACTICE GUIDELINES FOR THE DIAGNOSIS, TREATMENT, MANAGEMENT AND SURVEILLANCE OF PEOPLE WITH SCHWANNOMATOSIS

INTRODUCTION

Schwannomatosis is characterised by the development of typically painful, benign nerve sheath tumours (schwannomas) on the spinal and peripheral nerves around the body. Clinical care for people with schwannomatosis varies substantially, as there is no specific guideline on schwannomatosis yet.

GUIDELINE AIMS

The schwannomatosis guideline has been created to assist healthcare professionals to give the most up-to-date diagnosis, clinical management and surveillance of people with schwannomatosis. This guideline has been drawn from the best available evidence and the consensus of experts in caring for people with schwannomatosis and it will be regularly updated to reflect changes in evidence. The expectation is that clinicians will follow this guideline unless there is a compelling clinical reason specific to an individual patient not to.

SCOPE & PURPOSE OF THE GUIDELINE

The guideline is intended to define the optimal diagnosis, clinical management and surveillance of people with schwannomatosis.

GUIDELINE SUMMARY

Exam or surveillance		**Interval**	**Age to start**	**Strength***
Schwannomatosis	Clinical examination and assessment for pain and neurological examination	Annual	12-14	Moderate
Schwannomas	Brain and spine MRI	According to specific gene / age recommendations	Diagnosis or 12-14 years	Strong

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	Whole-Body MRI	Baseline or soon after. Consider alternating with Craniospinal	Diagnosis or 12-14 years	Moderate
	Ultrasound	Consider for problem solving in limbs or intercostal	As appropriate	Moderate

*This grading is based on published articles and expert consensus.

KEY RECOMMENDATIONS

Clinical Overview	Life expectancy in schwannomatosis **is not** usually affected, unlike NF2. Pain **is** a prominent feature, especially for people with a *LZTR1* germline pathogenic variant.
Diagnosis	In people with schwannomatosis at reproductive age or at transition, a discussion of the likely risks of transmission to offspring and the options for testing in pregnancy and pre-implantation diagnosis **should** be undertaken.
Imaging	In patients with localised pain and/or associated neurologic focal deficit, without an obvious schwannoma localised MRI **should** be performed using thin slices (<3mm) in order to detect very small but functionally significant schwannomas.
	For targeted investigation of pain, ultrasound (in the hands of someone experienced at imaging schwannomas) **may be** a useful problem-solving modality.
Annual clinical assessment	At each review visit there **should** be: • Full assessment of pain history • Full neurological examination • Assessment of Quality of Life using a recognized tool e.g. EQ-5D • **Assessment of psychological needs of the patient**

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Non-surgical pain management	Multidisciplinary pain management focusing on symptom management and targeting pain related disability using a bio-psychosocial approach **should** be used.
	Painful schwannomas have a significant neuropathic component, drugs such as tricyclic antidepressants and gabapentinoids **should** be used first line, and SSRI or other ASD (Topiramate, Carbamazepine, Oxcarbazepine) second line.
Surgical intervention	Some lesions are not surgically removable, and operations are linked to increased morbidity. So, assessment of the likelihood of success and the risks of neurological deficit **should** include assessment by a surgeon with significant experience resecting nerve sheath tumours

PSYCHOLOGICAL NEEDS

While the physical manifestations of schwannomatosis are objective and describable, **it is important to consider the impact of schwannomatosis on patients' cognitive, psychological, emotional and social well-being**. Psychological distress can be caused **by pain, fatigue**, having to undergo multiple surgeries, uncertainties about disease progression, and fears related to family planning.

Patients' beliefs about their medical condition can be extremely strong determinants in their response to therapy, long term management and overall disability. Severity of physical disease does not always correlate with emotional distress, however pain is a significant factor in schwannomatosis. This is not surprising as **pain has a well-recognised and significant psychosocial correlation**.

Realistically, a formal psychological assessment cannot be performed in all patients diagnosed with schwannomatosis. However, certain risk factors should alert the clinician to consider early psychological involvement and referral.