European Reference Network
for rare or low prevalence complex diseases

Network Genetic Tumour Risk Syndromes (ERN GENTURIS)

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European Reference Network on GENetic TUmour RIsk Syndromes

Care Pathway

Non-*NF2*-related schwannomatosis

version 1.3

Accepted: 26 September 2021

Non-*NF2*-related schwannomatosis (SCWN) CLINICAL PATHWAY

*The **Patient Clinical Pathway** is "the whole care pathway from identification, diagnostics, and multidisciplinary case discussions to surveillance and preventive surgery", so indeed a pathway in time, focusing on **HOW***

Annual Review Recommended

At time of diagnosis, or possible diagnosis, ALL patients should be seen in a genetics department.
Care can be co-ordinated through the schwannomatosis MDT team by an appropriate specialist
Annual review should be undertaken by a recognised specialist.
Patients, other local specialists and GPs have telephone access to the NF Reference Centre for NF-related concerns.

AGE	DIAGNOSTIC APPOINTMENT	REVIEWS CARRIED OUT BY	MRI head and spine
<12 50% risk or child of sporadic schwannomatosis patient or tested positive for family pathogenic variant (PV)	Based on symptoms	Care can be coordinated through the schwannomatosis MDT team by an appropriate specialist.	Not required unless symptomatic
12-15 50% risk	Confirmation of diagnosis^ & assessment. Genetic testing & counselling for family. Discharge if negative for family PV or on linkage	Care can be coordinated through the schwannomatosis MDT team by an appropriate specialist.	Symptom check at SCWN review and Baseline MRI and MRI 3-yearly if no tumours
<16 affected already with schwannomas	Confirmation of diagnosis^ & assessment. Genetic counselling for family.	Care can be coordinated through the schwannomatosis MDT team by an appropriate specialist. At least annual with paediatric neurologist.	6-month MRI after diagnosis and then 2-3 yearly MRI* unless symptomatic
12-15 offspring of a schwannomatosis patient and tested positive for PV	Confirmation of diagnosis^ & assessment. Genetic counselling for family.	Care can be coordinated through the schwannomatosis MDT team by an appropriate specialist. Annual with paediatric neurologist.	2-3 yearly MRI* until tumours identified
≥16 Affected with schwannomas	Confirmation of diagnosis^ & assessment. Genetic counselling for family.	Care can be coordinated through the schwannomatosis MDT team by an appropriate specialist. At least annual with team.	6-month MRI after diagnosis and then 2-3 yearly MRI* unless symptomatic
≥16 50% risk	Confirmation of diagnosis^ & assessment. Genetic testing & counselling for family Discharge if negative for family PV or on linkage	Care can be coordinated through the schwannomatosis MDT team by an appropriate specialist. Symptom check.	Baseline MRI and MRI 3-5 -yearly if no tumours. Stop screening aged 40-50.
≥16 offspring of a schwannomatosis patient and tested positive for PV	Confirmation of diagnosis^ & assessment. Genetic counselling for family.	Care can be coordinated through the schwannomatosis MDT team by an appropriate specialist. Symptom check.	Annual symptom check. Baseline MRI and MRI 2-3 -yearly* if no tumours. Stop screening age 70 if no tumours.

^ The name of all the schwannomatosis conditions reflect the underlying gene, thus *SMARCB1*-related schwannomatosis, *LZTR1*-related schwannomatosis, 22q-related schwannomatosis and schwannomatosis not otherwise specified (NOS)
*Whole Body MRI can be alternated

Non-*NF2*-related schwannomatosis Review Checklist—Children (0—16)		
	WHAT TO LOOK FOR	**WHEN TO REFER**
SKIN	Subcutaneous nodules that move beneath skin and can often feel on thickened nerve	If symptomatic or needed for genetic diagnosis
NEUROLOGICAL	Neurological symptom review, particularly loss of neurological function	If loss of function
Non-*NF2*-related schwannomatosis Review Checklist—Adults (16+)		
	WHAT TO LOOK FOR	**WHEN TO REFER**
SKIN	Check for symptomatic lesions	If symptomatic or needed for genetic diagnosis
PSYCHOLOGICAL BURDEN	Effects are underestimated. Psychological problems are common but patients, may be reluctant to talk about these issues and need encouragement.	
NEUROLOGICAL	Neurological symptom review, particularly headaches, nerve pain, and visual and gait disturbances and loss of muscle function including mononeuropathy	If loss of function
PAIN	It's important review quality, intensity and location of pain.	Uncontrolled pain

Non-NF2-related Schwannomatosis (SCWN) Clinical Pathway

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Faculty: .....

Family name:

Given name(s)

Address:

Date of Birth: Sex: ☐ M ☐ F ☐ I

Annual Review Recommended

At time of diagnosis, or possible diagnosis, ALL patients should be seen in a genetics department. **Care can be co-ordinated through the schwannomatosis MDT team by an appropriate specialist.** Annual review of symptoms should be undertaken by a recognised specialist. Patients, other local specialists and GPs have telephone access to the NF Reference Centre for NF-related concerns.

AGE	DIAGNOSTIC APPOINTMENT	ANNUAL SCWN REVIEWS OF SYMPTOMS CARRIED OUT BY	MRI head and spine
<12 50% risk or child of sporadic schwannomatosis patient or tested positive for family PV	Based on symptoms	Care can be co-ordinated through the schwannomatosis MDT team by an appropriate specialist.	Not required unless symptomatic
12-15 50% risk	Confirmation of diagnosis & assessment. Genetic testing & counselling for family. Discharge if negative for family PV or on linkage		Baseline MRI and MRI 3-yearly if no tumours
≥16 50% risk			Baseline MRI and MRI 3-5 -yearly if no tumours. Stop screening aged 40-50
12-15 offspring of a schwannomatosis patient and tested positive for PV	Confirmation of diagnosis & assessment. Genetic counselling for family.		2-3 yearly MRI* until tumours identified
≥16 offspring of a schwannomatosis patient and tested positive for PV			Baseline MRI and MRI 2-3 -yearly* if no tumours. Stop screening age 70 if no tumours
<16 affected already with schwannomas			6-month MRI after diagnosis and then 2-3 yearly MRI* unless symptomatic
≥16 Affected with schwannomas			6-month MRI after diagnosis and then 2-3 yearly MRI* unless symptomatic

* Whole Body MRI can be alternated

Review Checklist — Children (<16)

Clinical Presentation:

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Other symptoms:

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Genetic counselling completed ☐

Date Completed: .....

Clinical diagnosis

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Genetic Test '+'ve ☐

Diagnosis Date: .....

General Health Check:

Please record the follow as soon as possible and then annually:

Height

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Weight

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Blood Pressure

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WHAT TO LOOK FOR	WHEN TO REFER
SKIN: Subcutaneous nodules that move beneath skin and can often feel on thickened nerve	If symptomatic or needed for genetic diagnosis. ☐ **Date Referred**: .....
Neurological symptom review, particularly loss of neurological function	REFER to Complex National Reference Centre or neurologist if loss of function. ☐ **Date Referred**: .....

Notes:

The name of all the schwannomatosis conditions reflect the underlying gene, thus SMARCB1-related schwannomatosis, LZTR1-related schwannomatosis, 22q-related schwannomatosis and schwannomatosis not otherwise specified (NOS)

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Doctor: .....

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Review date: .....

Faculty: .....

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Network Genetic Tumour Risk Syndromes (ERN GENTURIS)

Faculty: .....

Family name:

Given name(s)

Address:

Date of Birth: Sex: ☐ M ☐ F ☐ I

Annual Review Recommended

At time of diagnosis, or possible diagnosis, ALL patients should be seen in a genetics department. **Care can be co-ordinated through the schwannomatosis MDT team by an appropriate specialist.** Annual review of symptoms should be undertaken by a recognised specialist. Patients, other local specialists and GPs have telephone access to the NF Reference Centre for NF-related concerns.

AGE	DIAGNOSTIC APPOINTMENT	ANNUAL SCWN REVIEWS OF SYMPTOMS CARRIED OUT BY	MRI head and spine
<12 50% risk or child of sporadic schwannomatosis patient or tested positive for family PV	Based on symptoms	Care can be co-ordinated through the schwannomatosis MDT team by an appropriate specialist.	Not required unless symptomatic
12-15 50% risk	Confirmation of diagnosis & assessment. Genetic testing & counselling for family. Discharge if negative for family PV or on linkage		Baseline MRI and MRI 3-yearly if no tumours
≥16 50% risk			Baseline MRI and MRI 3-5 -yearly if no tumours. Stop screening aged 40-50
12-15 offspring of a schwannomatosis patient and tested positive for PV	Confirmation of diagnosis & assessment. Genetic counselling for family.		2-3 yearly MRI* until tumours identified
≥16 offspring of a schwannomatosis patient and tested positive for PV			Baseline MRI and MRI 2-3 -yearly* if no tumours. Stop screening age 70 if no tumours
<16 affected already with schwannomas			6-month MRI after diagnosis and then 2-3 yearly MRI* unless symptomatic
≥16 Affected with schwannomas			6-month MRI after diagnosis and then 2-3 yearly MRI* unless symptomatic

* Whole Body MRI can be alternated

Review Checklist — Adults (16+)

Clinical Presentation:

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..... ☐

Other symptoms:

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Genetic counselling completed ☐

Date Completed: .....

Clinical diagnosis

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Genetic Test '+'ve ☐

Diagnosis Date: .....

General Health Check:

Please record the follow as soon as possible and then annually:

Height

.....

Weight

.....

Blood Pressure

.....

WHAT TO LOOK FOR	WHEN TO REFER
SKIN: check for symptomatic lesions.	If symptomatic or needed for genetic diagnosis. ☐ **Date Referred**: .....
Psychological burden: Effects are underestimated. Psychological problems are common but patients, may be reluctant to talk about these issues and need encouragement.	Consider **REFERRAL** to an appropriate counselling service. ☐ **Date Referred**: .....
Neurological symptom review, particularly headaches, nerve pain, visual and gait disturbances and loss of muscle function including mononeuropathy.	REFER to Complex National Reference Centre or neurologist if loss of function. ☐ **Date Referred**: .....
Pain: It is important to review quality, intensity and location of pain.	**REFER** to pain specialist if uncontrolled pain. ☐ **Date Referred**: .....

Notes:

The name of all the schwannomatosis conditions reflect the underlying gene, thus SMARCB1-related schwannomatosis, LZTR1-related schwannomatosis, 22q-related schwannomatosis and schwannomatosis not otherwise specified (NOS)

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Doctor: .....

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Review date: .....

Faculty: .....

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