Neurofibromatosis Type 2 Care pathway

*The Patient Clinical Pathway is "the whole care pathway from identification, diagnostics, and multidisciplinary case discussions to surveillance", 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 and a specialized NF2 Centre.
Ideally all but very mildly affected patients will be followed up as appropriate through a nationally recognized reference NF2 centre.
Annual review should be undertaken by a recognised NF2 specialist multidisciplinary team.
Patients, paediatricians other local specialists and GPs have telephone access to the NF Reference Centre for NF-related concerns.

AGE and status	DIAGNOSTIC APPOINTMENT	NF2 REVIEWS CARRIED OUT BY	Audiology, MRI, and opthalmology
<10 years 50% risk or child of sporadic NF2 patient And at risk group*	In first year and then annually	Care coordinated by Genetics or NF2 Specialist paediatrician	Symptom check at NF2 review, ophthalmic and audiology
10-15 years 50% risk And at risk group*	Confirmation of diagnosis & assessment. Genetic testing & counselling for family Discharge if negative for family pathogenic variant or on linkage analysis in families without pathogenic variant identified.	Care coordinated by Genetics or NF2 Specialist paediatrician	Symptom check at NF2 review and audiology Baseline MRI brain MRI spine can be delayed to second visit 2-yearly if no tumours. Usually only brain MRI at first assessment unless concerns
<16 years affected with NF2 tumours and meeting diagnostic criteria	Confirmation of diagnosis & assessment. Genetic counselling for family.	Coordinated by NF2 centre paediatric service	At least annual with paediatric NF2 specialist. Ophthalmology at baseline and two yearly. 6-month MRI after diagnosis and then annual MRI head and 3 yearly spine unless symptomatic.
<16 years offspring of an NF2 patient and tested positive for pathogenic variant or affected on linkage	Confirmation of diagnosis & assessment. Genetic counselling for family.	Coordinated by NF2 centre paediatric service	At least annual with paediatric neurologist. Ophthalmology at baseline and two yearly. Two yearly MRI brain and spine until tumours identified from age 8-10.
>15 years affected	Confirmation of diagnosis & assessment. Genetic counselling for family.	Coordinated by NF2 multidisciplinary adult (or paediatric to 18 in some countries) team. Should include minimum of Neurosurgeon, neuroradiologist, ENT, Neurology, genetics, audiology and ophthalmology.	At least annual with team. Ophthalmology at baseline and as needed. 6-month MRI after diagnosis and then annual MRI head and 3 yearly spine unless symptomatic.
>15 years 50% risk And at risk group*	Confirmation of diagnosis & assessment. Genetic testing & counselling for family Discharge if negative for family PV or on linkage.	Care coordinated by Genetics or NF2 adult team	Symptom check at NF2 review, ophthalmology and audiology Baseline brain MRI and MRI 2-3 -yearly if no tumours

>15 years offspring of an NF2 patient and tested positive for pathogenic variant or affected on linkage	Confirmation of diagnosis & assessment. Genetic counselling for family.	Coordinated by NF2 adult team	At least annual with NF2 team. Ophthalmology at baseline. Two yearly MRI brain and 5 yearly spine until tumours identified.

*Unilateral vestibular schwannoma, other sporadic schwannoma or meningioma aged <30

Review Checklist—Children (0—16)

Assess vision check for strabismus and cataract, neurological examination, audiology, cutaneous examination

	WHAT TO LOOK FOR	WHEN TO REFER
SKIN	Plaque like thickened skin often hairy and slightly pigmented. Subcutaneous nodules that move beneath skin and can often feel on thickened nerve.	**REFERRAL** If symptomatic or needed for genetic diagnosis
EYES	Have regular ophthalmic reviews taken place for those aged 0-15 years. Is there any evidence of a squint, retinal hamartoma or cataract.	**REFERRAL** to ophthalmologist if there are concerns about the eye or visual symptoms in between ophthalmic assessments
PSYCHOLOGICAL BURDEN	Effects are underestimated. Psychological problems are common but children, may be reluctant to talk about these issues and need encouragement.	
NEUROLOGICAL	Neurological symptom review, particularly loss of neurological function and pain and visual and gait disturbances and loss of muscle function including mononeuropathy.	**REFERRAL** If loss of function or pain requiring treatment
AUDIOLOGY	Review development – Pure tone audiogram, speech discrimination	**REFERRAL** If hearing loss

Neurofibromatosis Type 2 - Review Checklist—Adults (16+)

	WHAT TO LOOK FOR	WHEN TO REFER
SKIN	Check for symptomatic lesions	**REFERRAL** If symptomatic or needed for genetic diagnosis
PSYCHOLOGICAL BURDEN	Effects are underestimated. Psychological problems are common but patients, both men and women, 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	**REFERRAL** If loss of function
EYES	Visual assessment and discs particularly if has multiple cranial tumours	**REFERRAL** to ophthalmologist if there are any concerns about the eyes or visual symptoms
AUDIOLOGY	Review development – Pure tone audiogram, speech discrimination	**REFERRAL** If hearing loss

Neurofibromatosis Type 2 (NF2) Clinical Pathway

European Reference Network for rare or low prevalence complex diseases
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 and a specialized NF2 Centre.
Ideally all but very mildly affected patients will be followed up as appropriate through a nationally recognized reference NF2 centre.
Annual review should be undertaken by a recognised NF2 specialist multidisciplinary team.
Patients, paediatricians other local specialists and GPs have telephone access to the NF Reference Centre for NF-related concerns.

AGE	DIAGNOSTIC APPOINTMENT	NF2 REVIEWS CARRIED OUT BY	AUDIOLOGY AND MRI
<10 years 50% risk or child of sporadic NF2 patient AND at risk group*	In first year and then annually	Care coordinated by Genetics or NF2 Specialist paediatrician	Symptom check at NF2 review, ophthalmic and audiology
10-15 years 50% risk AND at risk group*	Confirmation of diagnosis & assessment. Genetic testing & counselling for family Discharge if negative for family PV or on linkage analysis in families without PV identified	Care coordinated by Genetics or NF2 Specialist paediatrician	Symptom check at NF2 review and audiology Baseline MRI brain MRI spine can be delayed to second visit 2-yearly if no tumours. Usually only brain MRI at first assessment unless concerns
<16 years affected with NF2 tumours and meeting diagnostic criteria	Confirmation of diagnosis & assessment. Genetic counselling for family.	Coordinated by NF2 centre paediatric service	At least annual with paediatric NF2 specialist. Ophthalmology at baseline and two yearly. 6-month MRI after diagnosis and then annual MRI head and 3 yearly spine unless symptomatic
<16 years offspring of an NF2 patient and tested positive for pathogenic variant or affected on linkage	Confirmation of diagnosis & assessment. Genetic counselling for family.	Coordinated by NF2 centre paediatric service	At least annual with paediatric neurologist. Ophthalmology at baseline and two yearly. Two yearly MRI brain and spine until tumours identified from age 8-10
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>15 years 50% risk AND at risk group*	Confirmation of diagnosis & assessment. Genetic testing & counselling for family Discharge if negative for family PV or on linkage	Care coordinated by Genetics or NF2 adult team	Symptom check at NF2 review, ophthalmology and audiology Baseline brain MRI and MRI 2-3 -yearly if no tumours
>15 years offspring of an NF2 patient and tested positive for PV or affected on linkage	Confirmation of diagnosis & assessment. Genetic counselling for family.	Coordinated by NF2 adult team	At least annual with NF2 team. Ophthalmology at baseline. Two yearly MRI brain and 5 yearly spine until tumours identified

*Unilateral vestibular schwannoma, other sporadic schwannoma or meningioma aged <30. PV = pathogenic variant.

Review Checklist — Children and adults

General health check (record as soon as possible and then annually):

Height .....
Weight .....
Blood Pressure .....

Clinical Presentation:

..... ☐
..... ☐
Other symptoms:
.....
Genetic counselling completed ☐
Date Completed: .....
Clinical diagnosis
.....
Positive Genetic Test ☐
Diagnosis Date: .....

WHAT TO LOOK FOR (child)	WHAT TO LOOK FOR (adult)
SKIN: Plaque like thickened skin often hairy and slightly pigmented. Subcutaneous nodules that move beneath skin and can often feel on thickened nerve. **REFERRAL** If symptomatic or needed for genetic diagnosis. ☐ **Date Referred**:	**SKIN**: Check for symptomatic lesions. **REFERRAL** If symptomatic or needed for genetic diagnosis. ☐ **Date Referred**: .....
EYES: Have regular ophthalmic reviews taken place for those aged 0-15 years. Is there any evidence of a squint, retinal hamartoma or cataract? **REFERRAL** to ophthalmologist if there are concerns about the eye or visual symptoms in between ophthalmic assessments ☐ **Date Referred**:	**EYES:** Visual assessment and discs particularly if has multiple cranial tumours. **REFERRAL** to ophthalmologist if there are any concerns about the eyes or visual symptoms (adult). ☐ **Date Referred**: .....
NEUROLOGICAL: Neurological symptom review, particularly loss of neurological function and pain and visual and gait disturbances and loss of muscle function including mononeuropathy. **REFERRAL** If loss of function or pain requiring treatment ☐ **Date Referred**:	**NEUROLOGICAL:** Neurological symptom review, particularly headaches, nerve pain, and visual and gait disturbances and loss of muscle function including mononeuropathy. **REFERRAL** If loss of function. ☐ **Date Referred**: .....
AUDIOLOGY: Review development – Pure tone audiogram, speech discrimination. **REFERRAL** If hearing loss. ☐ **Date Referred**:	**AUDIOLOGY:** Review development – Pure tone audiogram, speech discrimination. **REFERRAL** If hearing loss. ☐ **Date Referred**: .....
PSYCHOLOGICAL BURDEN: Effects are underestimated. Psychological problems are common but children, may be reluctant to talk about these issues and need encouragement.	**PSYCHOLOGICAL BURDEN:** Effects are underestimated. Psychological problems are common but patients, both men and women, may be reluctant to talk about these issues and need treatment.

Doctor: .....Review date: ..... Faculty: .....

Notes:

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