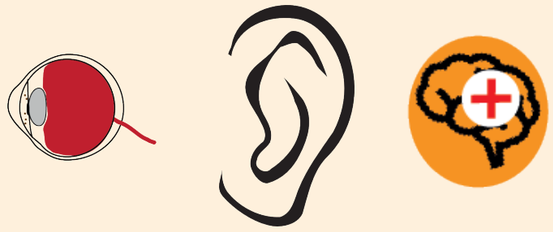
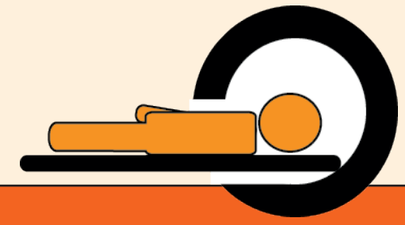
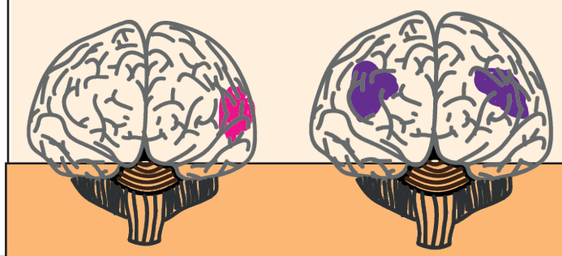
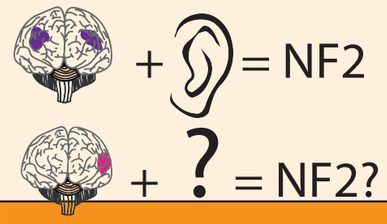
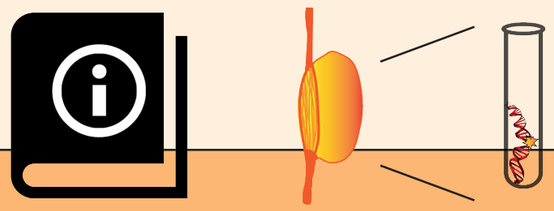
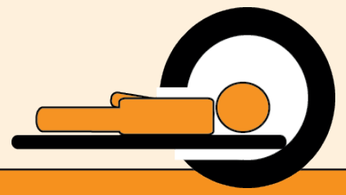
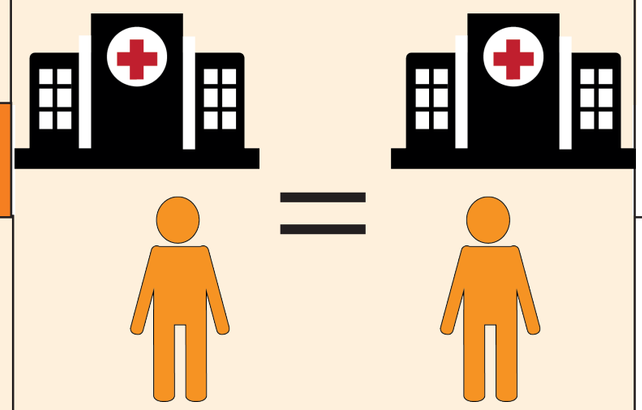
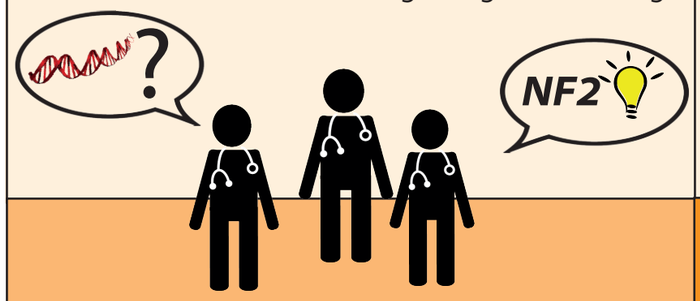
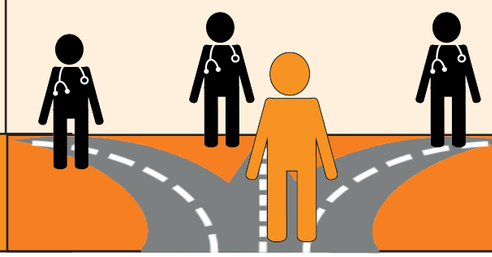
Patient Journey *NF2*-related schwannomatosis

	1st symptoms	Diagnosis	Treatment	Follow-up and surveillance
Disease				
Clinic	Benigne growth variable symptoms 	Hard to diagnose conform latest criteria 		Follow-up 
Challenge	Awareness among clinicians genetic testing on neoplastic tissue 	Diagnosis is based on MRI 	*NF2*-related schwannomatosis centers with multidisciplinary teams 	Equal surveillance and quality of life for all *NF2*-related schwannomatosis patients 
Goal	All clinicians are aware of *NF2*-related schwannomatosis and challenges in genetic testing 	Diagnostic criteria for clinicians are up-to-date 	Clear clinical pathway 	

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Patient Journey *NF2*-related schwannomatosis

Disease

1st symptoms

Symptoms appear at any age, from birth.
Mostly in children, teenagers and young adults.
100% penetrance.
- Hearing loss and/or tinnitus
- Balance problems
- Vision problems
- Neurological signs or symptoms
- Specific skin nodules (plaques)

Diagnosis

- Bilateral vestibular (hearing and balance nerve) schwannomas: clear *NF2*-related schwannomatosis hallmark
- Parent or child with *NF2*-related schwannomatosis OR unilateral vestibular schwannoma OR 2 or more meningiomas AND two other *NF2*-related schwannomatosis features (including 2 other *NF2*-related schwannomatosis related tumours).

REMEMBER no relatives with the disease in 50-60% of the cases.

Treatment

Neurosurgery is the treatment of choice, but only if necessary, based on:
- Position
- Volume
- Speed of growth

Often tumours are already too large or in a risky part of the nervous system, to be removed without iatrogenic consequences.

Bevacizumab is very expensive and not available in many countries.

Radiosurgery is contraindicated in young age for the risk of provoking a secondary tumour.

There may be >10 tumours throughout life.

Follow-up and surveillance

- Annual MRI startingat age 10-12
- Hearing evaluation
- Annual complete eye examination.

Clinic

Benigne growth variable symptoms

Severity of symptoms is dependent on type, location and volume:

- Vestibular schwannoma: hearing loss
- Brain meningioma close to optic pathways: vision problems
- Nerve tumours

Hard to diagnose conform latest criteria

Latest diagnostic criteria:
Diagnosis is based on MRI (brain, spine and peripheral nerves):
Two vestibular schwannomas OR one vestibular schwannoma PLUS two or more nerve tumours (schwannomas, ependymomas, meningiomas)

Follow-up

Wait and scan:
There may be > 10 tumours throughout life.

Assessment of risk of tumour versus risk of surgery of newly identified tumours, considering:
- Position
- Volume
- Speed of growth

Challenge

Awareness among clinicians genetic testing on neoplastic tissue

The genetic diagnosis, on the *NF2* gene, is prognostic and vital for pre-symptomatic diagnosis.
Genetic testing on tumour tissue is recommended (blood samples do not necessarily give a result).

NF2-related schwannomatosis is one of the 500 most frequent rare diseases but it is under diagnosed and wrongly considered a mild disease. Tumours do not show metastasis but the effect on the brain and spine is highly disabling and can even be lethal.

Diagnosis is based on MRI

Diagnosis is based on MRI, because of difficulties in genetic testing.

If there MRI image is not straight forward, *NF2*-related schwannomatosis diagnosis is hard.

The delay in diagnosis can take several years.

NF2-related schwannomatosis centers with multidisciplinary teams

Only very skilled neurosurgeons, can remove multi-lobulated tumours. 40% of *NF2*-related schwannomatosis tumours are multi-lobulated.
NF2-related schwannomatosis centres in every country with multidisciplinary teams of:
Neurosurgeons, neurologists, audiologists, radiologists, geneticists, eye specialists, psychologists, language, balance therapists, physiotherapists, occupational therapists etc.

Equal surveillance and quality of life for all *NF2*-related schwannomatosis patients

Good quality *NF2*-related schwannomatosis centres are present in:
-United Kingdom
- France
- Spain
- Germany
- Belgium.

In other countries *NF2*-related schwannomatosis care is not organised /specialised.

If *NF2*-related schwannomatosis patients decide to go to France or UK for a second opinion and neurosurgery, they have to arrange their own funding.

Quality of life and survival for *NF2*-related schwannomatosis patients are low outside *NF2*-related schwannomatosis specialised centres.

All *NF2*-related schwannomatosis patients should be entitled to good surveillance, care, and the best possible quality of life.

Reliable genetic testing should be available for prenatal screening and pre-implantation genetic diagnosis.

Goal

All clinicians are aware of *NF2*-related schwannomatosis and challenges in genetic testing

When the result of a DNA blood test is negative, tumour tissue is used for genetic testing.

Clinicians are aware that *NF2*-related schwannomatosis is not Neurofibromatosis type 1.

Diagnostic criteria for clinicians are up-to-date

Clinicians are aware of latest diagnostic criteria:
Two vestibular schwannomas OR one vestibular schwannoma PLUS two or more nerve tumours OR two meningiomas and two more nerve tumours.

Borderline cases are referred to specialists.

Clear clinical pathway

Clinical care is available for every *NF2*-related schwannomatosispatient, without economical restrictions.

Treatment options are explained and the patients are involved in the decision making process.

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