Patient Journey Neurofibromatosis Type 1 (NF1)

Disease	**1st symptoms**	**Genetic testing**	**Treatment depends on occurence of symptoms**	**Age specific follow-up**
Clinic	**Variation in severity of symptoms**	Counselling of parents		**including physical examination, MRI surveillance at adolescent age and ophthalmology up to age of 8, thereafter visual screening until adulthood.**
Challenge	Delay in diagnosis due to unspecific symptoms	**Collaboration of multidisciplinair team**	Referral to specialist	Detailed information on folllow-up before treatment
Goal	Patient/parents are informed and receive genetic counselling at an appropriate time	Patient/parents know which specialist to contact	Treatment options are clear opportunity to participate in clinical trial	Introduce specialist to patients/parents to balance wait and see versus treatment. Design a personalised follow-up.

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Disease

1st symptoms

Skin abnormalities are often present at birth or appear within six months. By the time a child is 6 years old, major symptoms usually are evident.

Many clinical features are age dependent and increase in severity over time.

Genetic testing

If symptoms raise suspicion for NF1, the patient – and parents- are referred to clinical genetics to confirm NF1 diagnosis.

Treatment depends on occurence of symptoms

Please refer to the ERN GENTURIS NF1 care pathway for specific referrals, look for:

* café-au-lait macules
* cutaneous neurofibroma
* plexiform neurofibroma
* neurological symptom review
* ophthalmological review in children
* Growth and (cognitive) development
* blood pressure
* scoliosis, pseudoarthrosis, bone-density, sphenoid dysplasia
* Psychosocial wellbeing and neuropsychological functioning and educational or behavioural needs
* pain

Age specific follow-up

Physical care (treatment depending on symptoms)

Psychoeducation and possible supportive therapy

including physical examination, MRI surveillance at adolescent age and ophthalmology up to age of 8, thereafter visual screening until adulthood.

Physical examination
- skin/body: cutaneous and plexiform neurofibromas' neurologic deficit
- Blood pressure: phaeochromocytoma/renovascular stenosis
- Skeletal changes: scoliosis, vertebral changes, limb abnormalities, growth

Check for precocious puberty

Ophthalmologic examination: OPG
MRI: in case of growth of benign tumours, suspicion of malignant tumours
-screening MRI brain/WB-MRI at adolescent age to assess tumour burden

Clinic

Variation in severity of symptoms

Early (common) symptoms
- Cafe au lait spots
- Freckling
- Naevus anaemicus
- Larger head size
- Developmental delay
- Lisch nodules
- Focal areas of signal intensity (FASI)

Age dependant (severe) symptoms
- Bone deformities
- Scoliosis
- Deficit or epilepsy
- Optic pathway glioma (OPG)
- Plexiform neurofibromas
- Cutaneous neurofibromas
- Malignant peripheral nerve sheath tumours
- Brain tumours

Counselling of patients/parents

Patients/parents needs psychoeducation to understand and to accept, and cope with the diagnosis and available treatment options.

Patients/parents need to know who is going to take care of them and their child(ren).

Challenge

Delay in diagnosis due to unspecific symptoms

Treatment of manifestations in NF may need a different approach than if they occur in non-NF patients.
Therefore it is important to diagnose NF1.

Education for GPs, paediatricians, and clinicians on NF1.

GPs and clinicians need to be aware of the combination of (common) clinical symptoms that point towards NF1.

Updated guidelines for NF1 detection.

Collaboration of multidisciplinair team

After NF1 diagnosis a multidisciplinary team, led by a case manager, need to collaborate to realise personalised care:
- geneticist: genetic counselling
- paediatrician/neurologist: counselling on surveillance and possibly treatments
- specialists of which surgeons: explanation of surveillance / treatment options
- psychological support: counselling on impact and consequences

Referral to specialist

- Quick referral to specialist for treatment / surgery
- Detailed information on treatment options
- Detailed information about beneficial complementary therapies
- "Case manager" is always up to date
- Information on possibilities to participate in a clinical trial or how to get access to an experimental therapy.

Detailed information on folllow-up before treatment – balance wait and see versus treatment options by multidisciplinary team

- Detailed information on follow up care options before treatment / surgery
- Start of follow up care as soon as it makes sense
- Continuous care by same (team of) specialist(s)
- Information on possibilities to participate in a clinical trial and to get access to an experimental therapy.
- Brain MRI and WB-MRI at transition age once

Goal

Patient/parents are informed and receive genetic counselling at an appropriate time

The patient/parents are well informed about genetic testing and the impact of the potential outcome:
- genetic counselling before and after genetics testing.
- Patient/parents are proactively offered psycho-education. A psychologist is part of the multidisciplinary team.
- Patient/parents are referred to the patient organisation.
- Patient/parents know which doctor is their "case manager" if NF1 is diagnosed.

Patient/parents know which specialist to contact

- Patient/parents know who is their "case manager" who can answer their questions or can refer them to a specialist.
- Patient/parents know the patient organisation and are informed about their program and services
- Patients / relatives can give their feedback and express their concerns.

Treatment options are clear. Opportunity to participate in clinical trial.

- Treatment options were explained and the patients / parents were involved in the decision making process
- Patients / parents are able to participate in a clinical trial
- Patients / parents can benefit from complementary therapies
- Patients / relatives can give their feedback and express their comments.

Introduce specialist to patient/parents before treatment.

Design a personalised follow-up.

- Team of specialists for follow up care is introduced to the patient / relatives and wait and see versus intervention is discussed
- A personalised follow-up care protocol is designed together with the patient/relatives
- Patients/relatives can give their feedback and express their concerns.
- There is a transition program for adolescent NF1 patients in place.

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