

# The "Patient Journey" for children with Neurofibromatosis Type 1: improving care by patients involvement

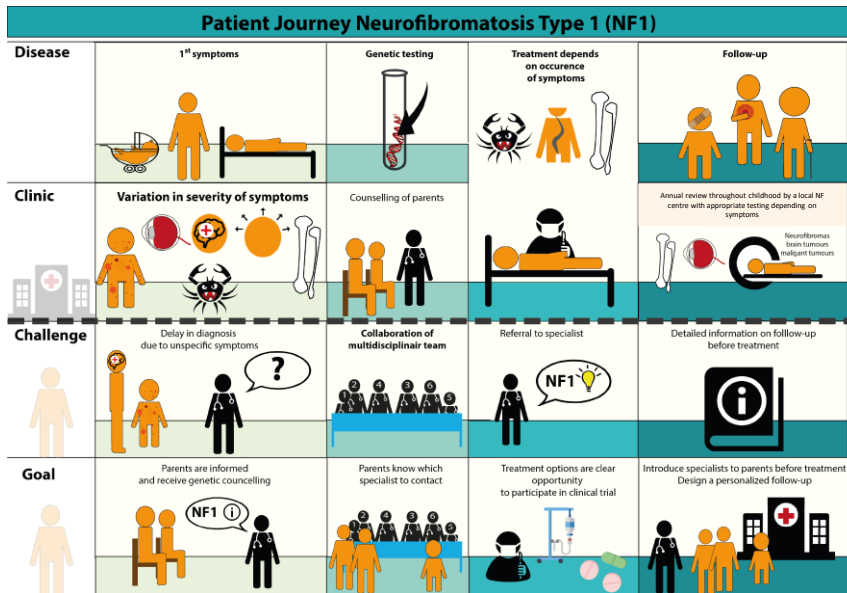
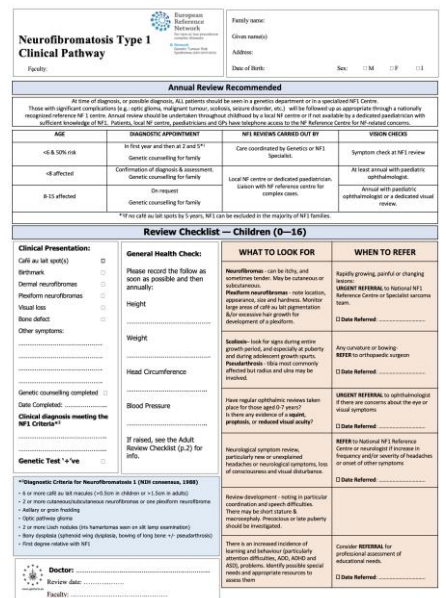
## ERN GENTURIS

- Patients with genetic tumour risk syndromes (genturis) are at very high risk of developing tumours in multiple organ systems at a young age.
- There are more than 40 different genturis, including Neurofibromatosis Type 1 (NF1).

*To overcome the hurdles every NF1-patient faces, ERN GENTURIS has developed an innovative visual approach for patients and parents input to share their expertise and experience: "Patient Journeys".*

## Method

- Patient representatives completed a mapping exercise of the needs of paediatric NF1-patients across the different stages of a "patient journey": first symptoms, (pre-)diagnosis, treatment, and follow-up. Each stage has three levels: clinical presentation, challenges and needs identified by patients, and their goal to improve care.
- The "patient journey" is visualized in a comprehensive manner, supported by a detailed information matrix.
- Final "patient journey" is reviewed by both patients and professional experts.
- The NF1 clinical pathway was thereafter reviewed:
  - Benchmarking the care and treatment on the needs identified in the "patient journey".
  - Turned into clinical support tool (piloted in clinical setting).

**Neurofibromatosis Type 1 Clinical Pathway**

Family name: \_\_\_\_\_ Given name: \_\_\_\_\_ Address: \_\_\_\_\_ Date of Birth: \_\_\_\_\_ Sex:  M  F  O

**Annual Review Recommended**

Age: \_\_\_\_\_ **REVIEW CATEGORY** \_\_\_\_\_

**REVIEW CATEGORY**

- < 6-8 months: In first year and then at 2 and 5\*
- < 6-affected: Confirmation of diagnosis & counselling. Genetic counselling for family
- < 6-15 affected: On request Genetic counselling for family

\* For request see page 10 of form. NF1 can be included in the majority of NF1 families.

**Review Checklist – Children (0–16)**

General Health Check:	WHAT TO LOOK FOR	WHEN TO REFER
<input type="checkbox"/> Birthmark	Neurofibromas – can be tiny, and sometimes tender. May be numerous or subcutaneous.	Any new or changing
<input type="checkbox"/> Dermal neurofibromas	Neurofibromas – note location, appearance, size and number. Monitor large areas for growth for development of disfigurement.	URGENT REFERRAL to National NF1 Reference Centre or specialist services team
<input type="checkbox"/> Height	Subtle: look for signs during entire growth period, and especially of puberty and during adolescent growth spurt.	Any constriction or bowing
<input type="checkbox"/> Blood Pressure	Phaeochromocytoma – like most commonly affected but rarely and also may be involved.	URGENT REFERRAL to National NF1 Reference Centre or specialist services team
<input type="checkbox"/> Blood Pressure	Neurological symptoms review.	URGENT REFERRAL to National NF1 Reference Centre or specialist services team
<input type="checkbox"/> Genetic Test *a-e	Neurological symptoms review.	URGENT REFERRAL to National NF1 Reference Centre or specialist services team

## Results

- The first symptoms and their severity vary greatly between NF1-patients. Patients indicated that this heterogeneity can cause a delay in diagnosis. The goal therefore is to inform parents and receive genetic counselling. Upon genetic testing and counselling the challenge is to form a multidisciplinary team, in which parents know which specialist they can contact.
- Treatment of NF1 depends on the occurrence of symptoms. It is therefore important to refer the patient to NF1-specialists, so that treatment options are clear, and patients will have the opportunity to participate in clinical trials. Moreover, detailed information on follow-up, which exist for MRI surveillance (if indicated, this is not routine without symptoms ) and annual physical and ophthalmologic examination, must be provided before treatment starts.

## Conclusion

Patients and their caregivers are able to discuss the individual needs of the NF1-patient using the "patient journey", while keeping in mind the expertise of both professional and patient leads.

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