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).

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.

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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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”.

Paediatric NF1 Patient Journey Visual

Paediatric NF1 Clinical Support Tool