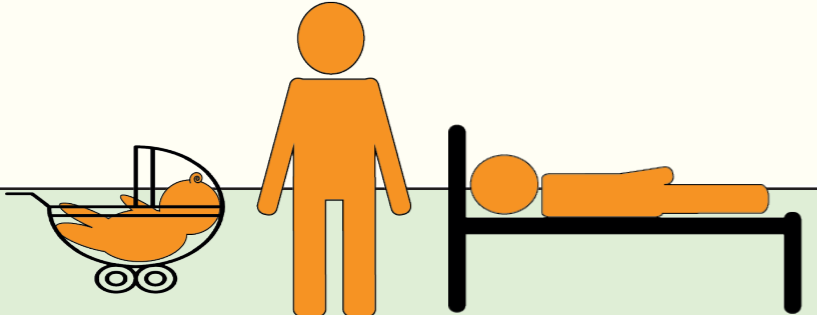


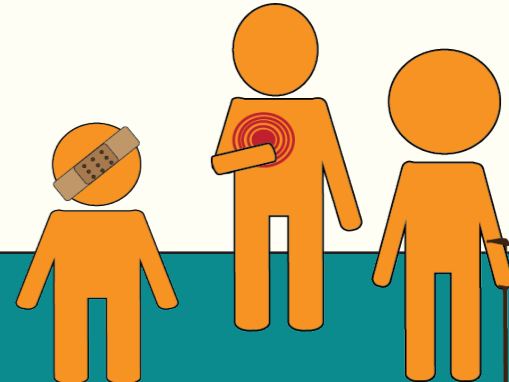
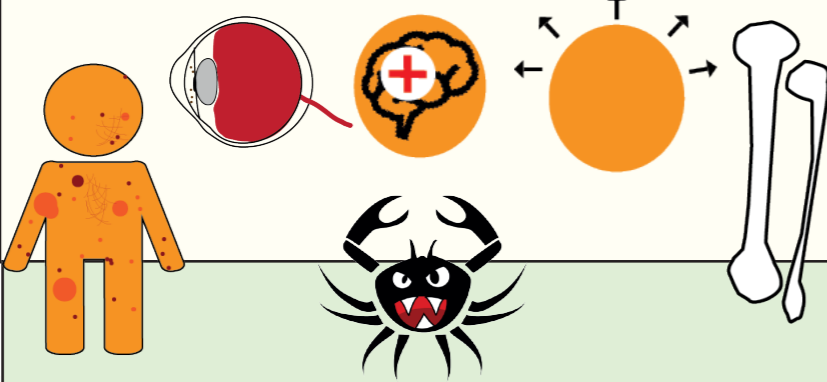
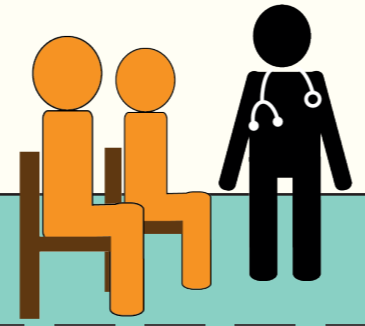
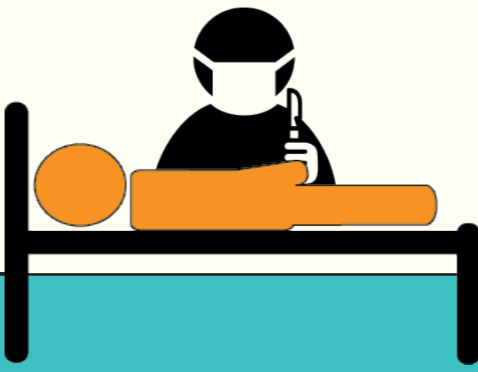
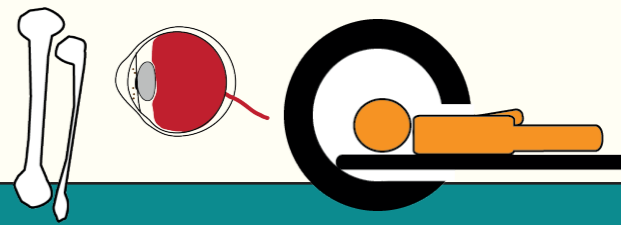
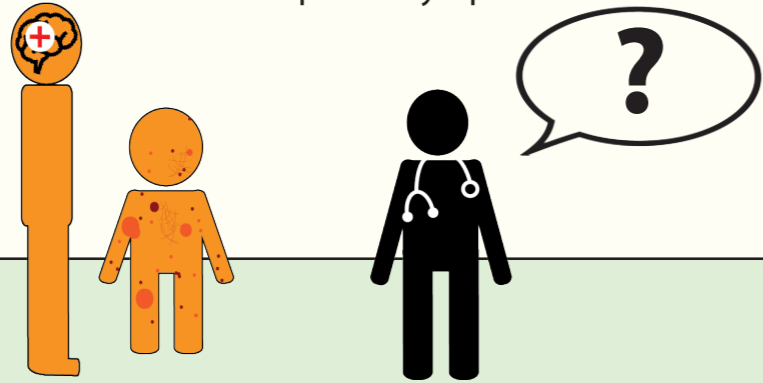



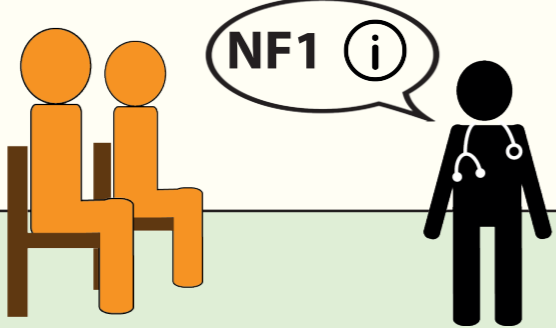
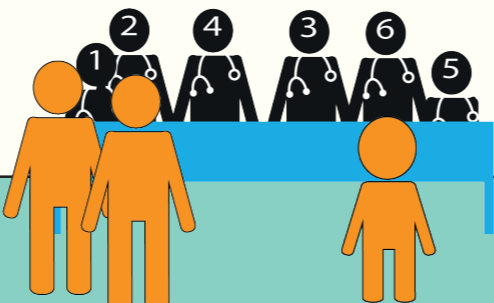
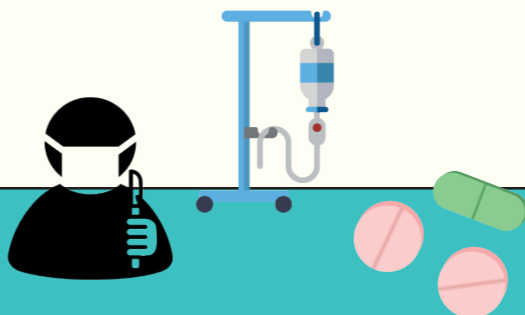


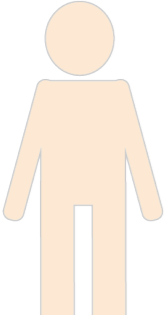
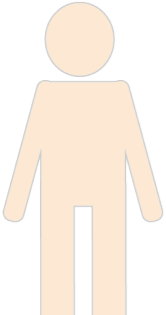


# Patient Journey Neurofibromatosis Type 1 (NF1)

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

# Patient Journey Neurofibromatosis Type 1 (NF1)

<b>Disease</b>	<p><b>1<sup>st</sup> symptoms</b></p> <p>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.</p> <p>Many clinical features are age dependent and increase in severity over time.</p>	<p><b>Genetic testing</b></p> <p>If symptoms raise suspicion for NF1, the patient – and parents- are referred to clinical genetics to confirm NF1 diagnosis.</p>	<p><b>Treatment depends on occurrence of symptoms</b></p> <p><b>Please refer to the ERN GENTURIS NF1 care pathway for specific referrals, look for:</b></p> <ul style="list-style-type: none"> <li>* café-au-lait macules</li> <li>* cutaneous neurofibroma</li> <li>* plexiform neurofibroma</li> <li>* neurological symptom review</li> <li>* ophthalmological review in children</li> <li>* Growth and (cognitive) development</li> <li>* blood pressure</li> <li>* scoliosis, pseudoarthrosis, bone-density, sphenoid dysplasia</li> <li>* Psychosocial wellbeing and neuropsychological functioning and educational or behavioural needs</li> <li>* pain</li> </ul>	<p><b>Age specific follow-up</b></p> <p><b>Physical care</b> (treatment depending on symptoms)</p> <p><b>Psychoeducation</b> and possible supportive therapy</p> <p><b>including physical examination, MRI surveillance at adolescent age and ophthalmology up to age of 8, thereafter visual screening until adulthood.</b></p> <p><b>Physical examination</b></p> <ul style="list-style-type: none"> <li>- skin/body: cutaneous and plexiform neurofibromas' neurologic deficit</li> <li>- Blood pressure: phaeochromocytoma/renovascular stenosis</li> <li>- Skeletal changes: scoliosis, vertebral changes, limb abnormalities, growth</li> </ul> <p><b>Check for precocious puberty</b></p> <p><b>Ophthalmologic examination:</b> OPG</p> <p><b>MRI:</b> in case of growth of benign tumours, suspicion of malignant tumours</p> <ul style="list-style-type: none"> <li>-screening MRI brain/WB-MRI at adolescent age to assess tumour burden</li> </ul>		
<b>Clinic</b> 	<p><b>Variation in severity of symptoms</b></p> <table border="0"> <tr> <td style="vertical-align: top;"> <p><b>Early (common) symptoms</b></p> <ul style="list-style-type: none"> <li>- Cafe au lait spots</li> <li>- Freckling</li> <li>- Naevus anaemicus</li> <li>- Larger head size</li> <li>- Developmental delay</li> <li>- Lisch nodules</li> <li>- Focal areas of signal intensity (FASI)</li> </ul> </td> <td style="vertical-align: top;"> <p><b>Age dependant (severe) symptoms</b></p> <ul style="list-style-type: none"> <li>- Bone deformities</li> <li>- Scoliosis</li> <li>- Deficit or epilepsy</li> <li>- Optic pathway glioma (OPG)</li> <li>- Plexiform neurofibromas</li> <li>- Cutaneous neurofibromas</li> <li>- Malignant peripheral nerve sheath tumours</li> <li>- Brain tumours</li> </ul> </td> </tr> </table>	<p><b>Early (common) symptoms</b></p> <ul style="list-style-type: none"> <li>- Cafe au lait spots</li> <li>- Freckling</li> <li>- Naevus anaemicus</li> <li>- Larger head size</li> <li>- Developmental delay</li> <li>- Lisch nodules</li> <li>- Focal areas of signal intensity (FASI)</li> </ul>	<p><b>Age dependant (severe) symptoms</b></p> <ul style="list-style-type: none"> <li>- Bone deformities</li> <li>- Scoliosis</li> <li>- Deficit or epilepsy</li> <li>- Optic pathway glioma (OPG)</li> <li>- Plexiform neurofibromas</li> <li>- Cutaneous neurofibromas</li> <li>- Malignant peripheral nerve sheath tumours</li> <li>- Brain tumours</li> </ul>	<p><b>Counselling of patients/parents</b></p> <p>Patients/parents needs psychoeducation to understand and to accept, and cope with the diagnosis and available treatment options.</p> <p>Patients/parents need to know who is going to take care of them and their child(ren).</p>	<p><b>Referral to specialist</b></p> <ul style="list-style-type: none"> <li>- Quick referral to specialist for treatment / surgery</li> <li>- Detailed information on treatment options</li> <li>- Detailed information about beneficial complementary therapies</li> <li>- "Case manager" is always up to date</li> <li>- Information on possibilities to participate in a clinical trial or how to get access to an experimental therapy.</li> </ul>	<p><b>Detailed information on follow-up before treatment</b> – balance wait and see versus treatment options by multidisciplinary team</p> <ul style="list-style-type: none"> <li>- Detailed information on follow up care options before treatment / surgery</li> <li>- Start of follow up care as soon as it makes sense</li> <li>- Continuous care by same (team of) specialist(s)</li> <li>- Information on possibilities to participate in a clinical trial and to get access to an experimental therapy.</li> <li>- Brain MRI and WB-MRI at transition age once</li> </ul>
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<b>Challenge</b> 	<p><b>Delay in diagnosis due to unspecific symptoms</b></p> <p>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.</p> <p>Education for GPs, paediatricians, and clinicians on NF1.</p> <p>GPs and clinicians need to be aware of the combination of (common) clinical symptoms that point towards NF1.</p> <p>Updated guidelines for NF1 detection.</p>	<p><b>Collaboration of multidisciplinary team</b></p> <p>After NF1 diagnosis a multidisciplinary team, led by a case manager, need to collaborate to realise personalised care:</p> <ul style="list-style-type: none"> <li>- geneticist: genetic counselling</li> <li>- paediatrician/neurologist: counselling on surveillance and possibly treatments</li> <li>- specialists of which surgeons: explanation of surveillance / treatment options</li> <li>- psychological support: counselling on impact and consequences</li> </ul>	<p><b>Referral to specialist</b></p> <ul style="list-style-type: none"> <li>- Quick referral to specialist for treatment / surgery</li> <li>- Detailed information on treatment options</li> <li>- Detailed information about beneficial complementary therapies</li> <li>- "Case manager" is always up to date</li> <li>- Information on possibilities to participate in a clinical trial or how to get access to an experimental therapy.</li> </ul>	<p><b>Detailed information on follow-up before treatment</b> – balance wait and see versus treatment options by multidisciplinary team</p> <ul style="list-style-type: none"> <li>- Detailed information on follow up care options before treatment / surgery</li> <li>- Start of follow up care as soon as it makes sense</li> <li>- Continuous care by same (team of) specialist(s)</li> <li>- Information on possibilities to participate in a clinical trial and to get access to an experimental therapy.</li> <li>- Brain MRI and WB-MRI at transition age once</li> </ul>		
<b>Goal</b> 	<p><b>Patient/parents are informed and receive genetic counselling at an appropriate time</b></p> <p>The patient/parents are well informed about genetic testing and the impact of the potential outcome:</p> <ul style="list-style-type: none"> <li>- genetic counselling before and after genetics testing.</li> <li>- Patient/parents are proactively offered psycho-education. A psychologist is part of the multidisciplinary team.</li> <li>- Patient/parents are referred to the patient organisation.</li> <li>- Patient/parents know which doctor is their "case manager" if NF1 is diagnosed.</li> </ul>	<p><b>Patient/parents know which specialist to contact</b></p> <ul style="list-style-type: none"> <li>- Patient/parents know who is their "case manager" who can answer their questions or can refer them to a specialist.</li> <li>- Patient/parents know the patient organisation and are informed about their program and services</li> <li>- Patients / relatives can give their feedback and express their concerns.</li> </ul>	<p><b>Treatment options are clear. Opportunity to participate in clinical trial.</b></p> <ul style="list-style-type: none"> <li>- Treatment options were explained and the patients / parents were involved in the decision making process</li> <li>- Patients / parents are able to participate in a clinical trial</li> <li>- Patients / parents can benefit from complementary therapies</li> <li>- Patients / relatives can give their feedback and express their comments.</li> </ul>	<p><b>Introduce specialist to patient/parents before treatment.</b></p> <p><b>Design a personalised follow-up.</b></p> <ul style="list-style-type: none"> <li>- Team of specialists for follow up care is introduced to the patient / relatives and wait and see versus intervention is discussed</li> <li>- A personalised follow-up care protocol is designed together with the patient/relatives</li> <li>- Patients/relatives can give their feedback and express their concerns.</li> <li>- There is a transition program for adolescent NF1 patients in place.</li> </ul>		