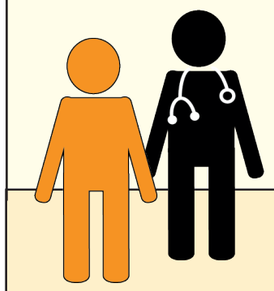
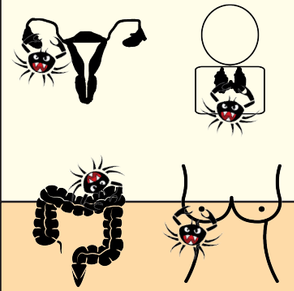
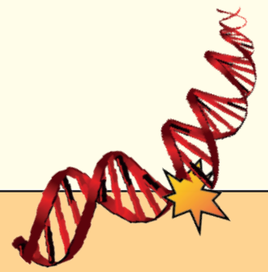
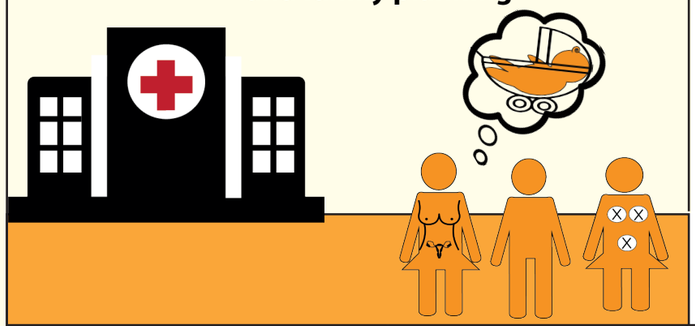
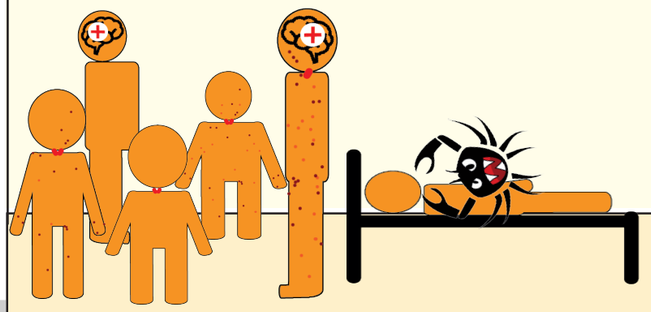
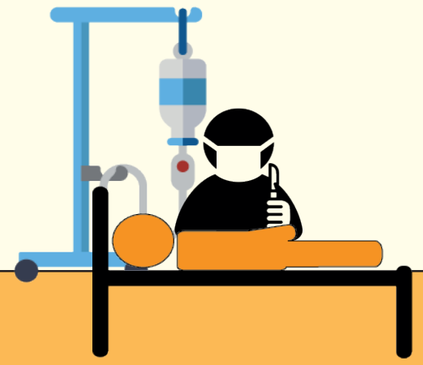
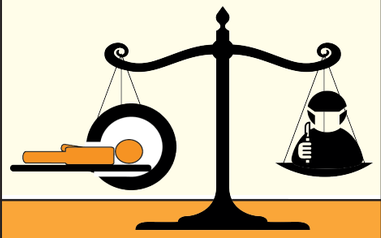
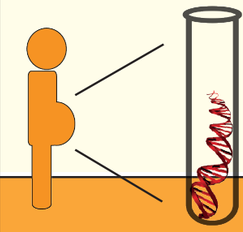
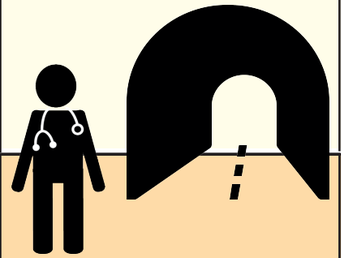
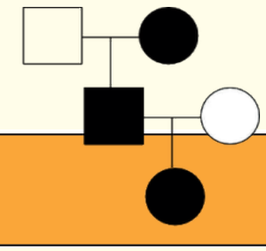
Patient Journey PTEN hamartoma tumour syndrome (PHTS)

	1st symptoms	Cancer diagnosis	Genetic diagnosis	Cancer treatment and (preventive) surgery	Surveillance and family planning	
Disease						
Clinic	Range of symptoms 	Involvement of broad multidisciplinary team 		Options: surveillance or preventive surgery 	Prenatal diagnosis 	
Challenge	Early detection 	Clinicians considering aspects outside their expertise 	Quick genetic diagnosis 	Personalised treatment 	Psychological support 	50% probability of a child with mutation 
Goal	Awareness and education for GPs and clinicians 	Guidelines to identify red flags 	Genetic tests for relatives 		Personalised follow up 	

European Reference Network

Genetic Tumour Risk Syndromes (ERN GENTURIS)

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Patient Journey PTEN hamartoma tumour syndrome (PHTS)

Disease

1st symptoms

Symptoms are variable:
- First symptoms present in a wide range of ages
- Severity of symptoms varies

Cancer diagnosis

Cancer risks:
Breast (67-85 %)
Thyroid (6-38 %)
Endometrial (19-28 %)
Kidney (2-34 %)
Colorectal (9-20 %)
Melanoma (0-6 %)

Genetic diagnosis

Based on clinical signs a presumptive diagnosis can be made.

The actual diagnosis of PHTS is based on a blood test that identifies a PTEN mutation.

Cancer treatment and (preventive) surgery

Cancer treatment:
- Breast
- Endometrial
- Thyroid
- Kidney
- Colorectal
- Melanoma

Often at a more early age than in the general population.

Involvement of multidisciplinary team of experts including a geneticist to counsel about options for surveillance and preventive surgery of the breasts.

Surveillance and family planning

Surveillance:
- Breast: yearly MRI from 30 years of age
  every 2 years mammography from 40 years of age
- Thyroid: yearly ultrasound from 18 years of age
- Kidney: every 2 years ultrasound from 40 years of age
- Endometrial: not recommended
- Colorectal: baseline colonoscopy at 35-40 years of age
- Melanoma: baseline skin examination at 30 years of age

Counselling on family planning

Clinic

Range of symptoms

1 or more of these symptoms can be seen in PHTS patients

Symptoms:
Macrocephaly
Thyroid lesions
Vascular abnormalities
Cutaneous spots
Intestinal polyps
Autism spectrum disorders
Etc.

Cancers:
Breast
Thyroid
Endometrial
Kidney
Colorectal
Melanoma

Involvement of broad multidisciplinary team

Multiple specialist have to work together, e.g:
- Geneticist
- Oncologist
- Surgeon
- Paediatrician
- Neurologist
- Endocrinologist
- Gastroenterologist
- Gynecologist

Options: surveillance or preventive surgery

Counselling on options:
- Enhanced surveillance
- Risk reducing surgery of the breasts

Prenatal diagnosis

Counselling on options for family planning:
- Prenatal diagnostics
- Preimplantation genetic diagnosis

Challenge

Early detection

Clinicians should be aware of genetic tumour risk syndromes and consider early onset cancer.

Early detection and treatment are vital for outcome!

Clinicians considering aspects outside their expertise

After diagnosis experts should be aware of all aspects of PHTS.

Patients should be referred to different expert centres and experts according to their needs and symptoms.

Quick genetic diagnosis

PHTS patients have to be detected as early as possible to be able to enrol in surveillance programs and take preventive measures.

Personalised treatment

There is no cure for PHTS.
Treatment is symptomatic.

Need for:
- Expert centres
- Center of excellence for the surgery
- PHTS specific treatment
- Research efforts
- Clinical trials

Psychological support

Psychological support to deal with:
- Anxiety after cancer treatment
- Choice for risk reducing surgery (mastectomy)
- Consequences for patient and relatives.

Exchange of views with other PHTS families or support groups.

50% probability of a child with mutation

PHTS patients with a child (wish) have 50% of probability to having a child with the disease causing variant

Counselling on dealing with the consequences.

Goal

Awareness and education for GPs and clinicians

GPs and clinicians are aware of:
- existence of PHTS
- the combination of clinical signs

Guidelines to identify red flags

GPs and clinicians should be able to recognise PHTS.

GPs and clinicians should know experts to refer to.

Genetic tests for relatives

Support for PHTS patients and their relatives.

Counselling on consequences of genetic testing for relatives.

Personalised follow-up

Expert centres for surveillance that offers:
- Knowledge of PHTS clinical guidelines
- Multidisciplinary team
- Experts according to patients needs

This patient journeys is according to the following ERN GENTURIS guideline: Cancer surveillance guideline for individuals with PTEN Hamartoma Tumour Syndrome (PHTS). Authors: Dr. Marc Tischkowitz, U.K., Dr. Chrystelle Colas, France, Dr Sjaak Pouwels, The Netherlands, Prof Nicoline Hoogerbrugge, The Netherlands. This guideline and further information on PHTS, ERN GENTURIS and its centes can be found on **www.genturis.eu .**

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