Your next steps

Find a specialist
Consult a gastroenterologist and genetic specialist, who might refer you to the other specialsts.

Follow medical guidelines
Regular specialist checkup: endoscopy.

Monitor symptoms
Keep track of new or changing health issues.

Consider treatment options
Discuss available therapies and potential surgical interventions.

Stay updated on Polyposis syndromes research
Clinical trials may offer new treatment possibilities.

Find a patient organisation
They can provide guidance and support.

Visit **www.genturis.eu** for medical guidelines and treatment centres.

About ERN GENTURIS

ERN GENTURIS is a European expert network for hereditary tumour syndromes. While we do not offer direct treatment, we provide patients and healthcare professionals with trusted information, guidance, and connections to specialists.

More info and contact

www.genturis.eu
E-mail: genturis@radbodumc.nl

European Reference Network

Funded by the European Union

Patient information

Polyposis syndromes: what this diagnosis means for your health and next steps

If you or a family member has been diagnosed with a Polyposis syndrome this guide provides clear medical information and practical next steps.

European Reference Network
for rare or low prevalence complex diseases
Network Genetic Tumour Risk Syndromes (ERN GENTURIS)

ERN GENTURIS
With every diagnosis we can help an entire family

What are Polyposis syndromes?

Polyposis syndromes are a group of inherited conditions characterised by the development of multiple polyps (abnormal growths) in the gastrointestinal tract. Depending on the syndrome, individuals may develop tens to thousands of polyps, which can vary in type, including adenomatous, hamartomatous, or serrated. Polyps can cause complaints like bleeding, pain and sometimes even bowel obstruction. If left undetected or untreated, some polyps can progress to colorectal cancer. Many polyposis syndromes are also associated with an increased risk of cancers in other organs, including the stomach, small intestine, pancreas, breast, and cervical cancer, depending on the specific syndrome. Major categories of polyposis syndromes include:

- **Adenomatous polyposis syndromes**, e.g. Familial adenomatous polyposis (FAP), MUTYH-associated polyposis (MAP).
- **Hamartomatous polyposis syndromes**: Juvenile polyposis syndrome (JPS) and Peutz-Jeghers syndrome (PJS).
- **Other polyposis syndromes:** Serrated polyposis syndrome (SPS), Hereditary mixed polyposis syndrome (HMPS), and Gastric adenocarcinoma and proximal polyposis of the stomach (GAPPS).

How are Polyposis syndromes inherited?

Most polyposis syndromes are inherited, although inheritance patterns vary depending on the underlying genetic cause. It might be

1. ***autosomal dominant inheritance:*** only one altered copy of the gene is needed to cause the condition, e.g. Familial adenomatous polyposis (FAP).
2. ***autosomal recessive inheritance:*** two altered copies of the gene (one inherited from each parent) are required for the condition to develop, e.g. MUTYH-associated polyposis (MAP).

What causes Polyposis syndromes?

Most polyposis syndromes are caused by inherited pathogenic (disease-causing) variants in genes involved in:

- DNA repair, which corrects DNA damage,
- Tumour suppression, which regulates cell growth and prevents uncontrolled cell division.

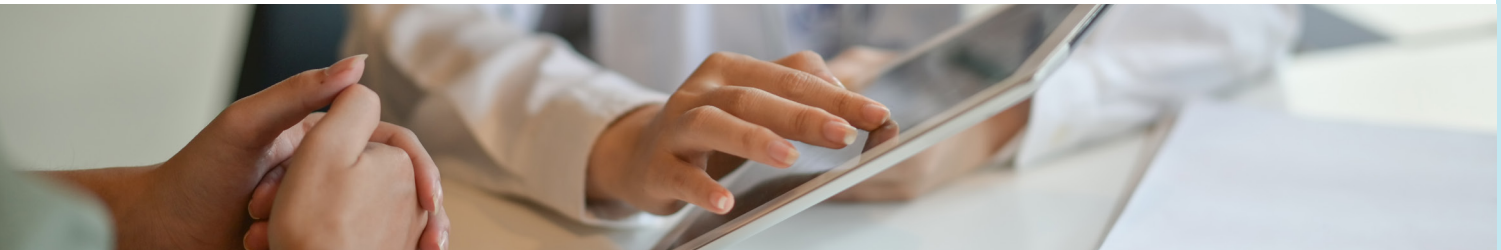
Some pathogenic variants are inherited from a parent, while others arise for the first time (de novo) or occur in only a proportion of the body's cells (mosaicism).

What are the diagnostic/testing options?

Diagnosis of a polyposis syndrome is based on a combination of:

- Clinical assessment, including personal and family history of polyps and cancer.
- Endoscopic examination (colonoscopy and upper gastrointestinal endoscopy) to identify and characterise polyps.
- Histopathology, to determine the type of polyps.
- Genetic testing, using multigene panels to identify pathogenic variants associated with inherited polyposis syndromes.

The diagnosis might be relevant for family planning.



What are the surveillance recommendations?

Surveillance in most polyposis syndromes aims to detect and remove polyps before they become cancerous and to identify associated cancers at an early stage.
Recommendations vary according to the specific syndrome, gene involved, and clinical presentation.

General surveillance recommendations include:

- Regular endoscopy with removal of polyps.
- Preventive surgery, such as colectomy, may be considered when the number or size of polyps cannot be safely managed endoscopically.
- Imaging and surveillance for extraintestinal cancers may also be recommended depending on the syndrome.

Typical starting ages include:

- FAP: surveillance begins at approximately 12 years of age.
- Attenuated or later-onset adenomatous polyposis syndromes: typically 18–20 years.
- Juvenile polyposis syndrome: surveillance usually begins between 12 and 15 years of age.
- Peutz-Jeghers syndrome: surveillance begins at 8 years, with additional screening for associated cancers in adulthood.

ERN GENTURIS provides information and resources to help you to understand this condition and guide your medical team toward the best possible treatment.

More information on **Polyposis syndromes**: