

Family Education Sheet



Familial Adenomatous Polyposis (FAP)

What is Familial Adenomatous Polyposis (FAP)?

- FAP is a colorectal (colo- = colon; -rectal = rectum) syndrome characterized by dozens to hundreds of polyps (abnormal growths or tumors) in the body. In FAP, the polyps are called adenomas. FAP is an inherited syndrome, which means it is passed down from your parents or grandparents.
- Almost everyone who is diagnosed with FAP develops colorectal cancer (CRC) by the age of 40-50 years if the entire colon is not removed.
- Attenuated FAP (AFAP) is a milder kind of FAP. AFAP is characterized by fewer polyps and the development of colorectal cancer at a later age. Less than 10% of families with FAP have an attenuated form.
- Polyps are generally found in the large intestine. However, many patients also develop polyps in other areas of the gastrointestinal (GI) tract, such as the stomach and small intestine. The risk of developing cancer in these areas is much less than in the large intestine.

What is a polyp?

- A gastrointestinal polyp is an abnormal growth of tissue that is either attached to the intestinal wall by a stalk (pedunculated) or growing directly from the wall (sessile).

How common is FAP?

- FAP occurs in approximately 1 in 10,000 people and affects both males and females equally.
- FAP is responsible for less than 1% of cases of colorectal cancer.

What causes FAP?

- FAP is usually caused by a mutation of the adenomatous polyposis coli (*APC*) gene. Rarely, other gene mutations may also give rise to this condition.
- The gene mutation can happen in two ways:
 1. It can be passed down from a parent (80-85% of cases).
 2. It can be a new mutation in the *APC* gene (15-20% of cases).

What are the symptoms and possible complications of FAP?

- Individuals may have no symptoms in the early development of FAP. Abnormal signs and symptoms that may develop in the course of the disease include:
 - Abdominal pain
 - Rectal pain or bleeding
 - Diarrhea
 - Anemia

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- Individuals with FAP may have other abnormal growths or lesions in other areas of the body, such as the thyroid, pancreas, stomach, brain, liver, eyes, teeth, and bone.
- These can be either benign (non-cancerous) or malignant (cancerous) lesions. Types of these lesions are listed below in order from most common to least common.
 - **Benign Lesions**
 - Small black dots in the back of the eye (Congenital hypertrophy of the retinal pigmented epithelium or CHRPE)
 - Fluid sac on the surface of the skin (Epidermoid cysts)
 - Abnormal bone growth (Osteoma)
 - Extra teeth (Supernumerary teeth)
 - Tumor in the abdominal wall (Desmoid tumor)
 - Tumor in the adrenal gland (Adrenal gland adenomas)
 - **Malignant Lesions**
 - Thyroid cancer
 - Brain tumor
 - Liver tumor (Hepatoblastoma)

Who should be screened for FAP?

- You should be screened for FAP if you have:
 - A family history of FAP or colon cancer at a young age, or
 - Any of the unusual growths or lesions listed above.
- An initial screening should be done:
 - By 8-10 years of age for a high-risk patient,
 - By 10-12 years of age for an average-risk patient, or
 - At the time of first symptoms.
- Average- and high-risk patients are determined based on family history and/or known high-risk gene mutations.

How are patients screened for FAP?

- Patients are screened for FAP by performing a colonoscopy. A doctor uses a long, flexible tube with a light and camera on the end to look inside the rectum and up into the large intestine. An upper GI endoscopy is often done at the same time. An upper GI endoscopy also uses a long, flexible tube but looks down into the esophagus ("food tube"), stomach, and small intestine.
- Another way to screen for FAP is by genetic testing. Detection of the gene mutation is very accurate, detecting approximately 95% of cases and with 98% accuracy.
- Other screening studies may include radiology imaging.

How is FAP treated?

- The most important treatment for FAP is the early removal of the entire colon (prophylactic colectomy) to prevent the development of cancer. The timing of surgery is based on the patient's age or other high-risk factors, including family history, polyp number, and pre-malignant changes in the polyps.

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- There is no cure for FAP. Treatment is directed to reduce complications, such as abdominal pain, bowel obstruction, GI bleeding, and cancer.

A [Spanish](#) version of this is available from your provider

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