**Familial Adenomatous Polyposis**

**What is familial adenomatous polyposis?**

Classic familial adenomatous polyposis, called FAP or classic FAP, is a genetic condition. It is diagnosed when a person develops more than 100 adenomatous colon polyps.

An adenomatous polyp is an area where normal cells that line the inside of a person’s colon form a mass on the inside of the intestinal tract.

The average age for polyps to develop in people with FAP is in the **mid-teens**.

More than 95% of people with FAP will have **multiple colon polyps by age 35.**

If FAP is not recognized and treated, there is almost a 100% chance that a person will develop [**colorectal cancer**](http://www.cancer.net/node/31317)**.**

There is also an increased chance of developing cancer in the [stomach](http://www.cancer.net/node/31376) and/or [small intestine](http://www.cancer.net/node/31377).

Other types of cancer found in families with FAP include [**hepatoblastoma**](http://www.cancer.net/node/31274), a type of liver cancer seen in young children; **desmoid tumors/desmoid fibromatosis**, a locally aggressive tumor that does not metastasize; [**papillary thyroid cancer**](http://www.cancer.net/node/31262); [**pancreatic**](http://www.cancer.net/node/31388)**,** [**adrenal**](http://www.cancer.net/node/31341)**, and bile duct tumors**; and a type of brain tumor called [**medulloblastoma**](http://www.cancer.net/node/31266).

Not all symptoms of FAP are cancer-related.

Some additional features of FAP may include:

* ***Osteomas***, which are noncancerous bony growths, usually found on the jaw
* ***Extra, missing, or unerupted teeth***
* ***Congenital, meaning present at birth***,
* ***Hypertrophy of the retinal pigment epithelium (CHRPE).***
* ***Benign (noncancerous) skin changes, such as epidermoid cysts and fibromas***
* ***Adrenal masses***

There are three subtypes of classic FAP called [**attenuated FAP (AFAP)**](http://www.cancer.net/node/18503)**,** [**Gardner syndrome**](http://www.cancer.net/node/18869)**, and** [**Turcot syndrome**](http://www.cancer.net/node/19307). This section addresses classic FAP.

**What causes classic FAP?**

FAP is passed from generation to generation in a family. The APC gene is linked to FAP; APC stands for adenomatous polyposis coli. A mutation (alteration) in the APC gene gives a person an increased lifetime risk of developing colorectal cancer or other cancers of the digestive tract.

**How is classic FAP inherited?**

Normally, every cell has two copies of each gene: one inherited from the mother and one inherited from the father. FAP follows **an autosomal dominant inheritance pattern**. In autosomal dominant inheritance, a mutation happens in only one copy of the gene. This means that a parent with a gene mutation may pass along a copy of their normal gene or a copy of the gene with the mutation. Therefore, a child who has a parent with a mutation has a 50% chance of inheriting that mutation. A brother, sister, or parent of a person who has a mutation also has a 50% chance of having the same mutation.

Options exist for couples interested in having a child when they know that one of them carries a gene mutation that increases the risk for this hereditary cancer syndrome.

Preimplantation genetic diagnosis (PGD) is a medical procedure done in conjunction with in-vitro fertilization (IVF). It allows people who carry a specific known genetic mutation to have children who do not carry the mutation.

**How common is classic FAP?**

FAP is uncommon; specific estimates on how many people have FAP vary from one in 22,000 up to one in 7,000. About 30% of people with FAP do not have any family history of the condition; they have a de novo (new) mutation in the APC gene.

Most colorectal cancer is sporadic, meaning it occurs by chance, and is not related to FAP or other known inherited genetic changes. Less than 1% of all colorectal cancer is thought to be due to FAP.

**How is classic FAP diagnosed?**

Classic FAP is a clinical diagnosis. This means that it is typically diagnosed when the doctor finds many polyps, rather than by the results of a laboratory test. A person with more than 100 adenomatous colon polyps is considered to have FAP. People with FAP can also have a blood test to look for a mutation in the APC gene. If an APC gene mutation is found, other family members may be diagnosed with FAP if they are tested and have the same gene mutation.

**What are the estimated cancer risks associated with classic FAP?**

Colorectal cancer                                            almost 100% if not treated

Desmoid tumor                                               10% to 20%

Small bowel (intestines)                                  4% to 12%

Pancreatic cancer                                            2%

Papillary thyroid cancer                                  2%

Hepatoblastoma (a type of liver cancer)         1.5%

Brain or central nervous system tumor           less than 1%

Stomach cancer                                               0.5%

Bile duct cancer                                              small, but increased

Adrenal gland cancer                                      small, but increased

**What are the screening options for classic FAP?**

ASCO recommends the following screening for people with FAP. It is important to discuss these options with your doctor, as each individual is different:

***Sigmoidoscopy or colonoscopy every one to two years, starting at age 10 to 11***

***Yearly*** [***colonoscopy***](http://www.cancer.net/node/24481) ***once polyps are found until a colectomy is planned***.

People with classic FAP may need a colectomy, the surgical removal of the entire colon, at some point due to a high number of polyps and the high risk of colorectal cancer.

This is a major surgery and possible side effects may include the need for [colostomy](http://www.cancer.net/node/24724). Talk with your doctor about what to expect during and after this surgery.

After colon surgery, surveillance of the lower tract with sigmoidoscopy should continue with regular frequency, depending on type of surgery.

**Every 6 to12 months if some rectal tissue remains**

**Every 1 to 4 years if all rectal tissue has been removed (small intestinal pouch)**

[**Upper endoscopy**](http://www.cancer.net/node/24731) **(EGD) at age 25 to 30 or once colorectal polyps are detected, whichever occurs first**

**Yearly ultrasound of the thyroid may be considered starting at age 25 to 30**

Computed tomography (CT) scan or magnetic resonance imaging (MRI) if a person has a family history of desmoid tumors or a mutation on the APC gene that is linked with these tumors