Familial Adenomatous Polyposis

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Overview

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 begin to make mucous and 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 [2]. The risk of colon cancer is 87% by age 45.

There is also an increased chance of developing cancer in the stomach [3] and/or small intestines [4]. Other types of cancer found in families with FAP include hepatoblastoma [5] (a type of liver cancer seen in young children); papillary thyroid cancer [6]; pancreatic [7], adrenal [8], and bile duct cancers [9]; and a low risk of a type of brain cancer called medulloblastoma [10].

Not all symptoms of FAP are cancer-related. Some additional features of FAP may include:

- Osteomas (noncancerous bony growths, usually found on the jaw)
- Extra, missing, or unerupted teeth
- Congenital (present at birth) hypertrophy of the retinal pigment epithelium (CHRPE), an eye condition that does not affect vision but can be seen by looking at the retina using a special instrument called an ophthalmoscope.
- Benign (noncancerous) skin changes, such as epidermoid cysts and fibromas
- Desmoid tumors (noncancerous fibrous tumors that can develop anywhere in the body)
- Adrenal masses

There are three subtypes of classic FAP called attenuated FAP (AFAP) [11], Gardner syndrome [12], and Turcot syndrome [13]. This section addresses classic FAP.

What causes 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 [2] or other cancers of the digestive tract.

How is 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.

How common is 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 [2] is sporadic (occurs by chance) and is not related to FAP or other known inherited genetic changes [14]. Less than 1% of all colorectal cancer is thought to be due to FAP.

How is FAP diagnosed?

Classic FAP is a clinical diagnosis. This means that it is typically diagnosed by its symptoms rather than 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 FAP?

- Colorectal cancer [2] almost 100% if not treated
- Small bowel [4] (intestines) 4% to 12%
- Pancreatic cancer [7] 2%
- Papillary thyroid cancer [6] 2%
- Hepatoblastoma [5] (a type of liver cancer) 1.5%
- Brain [15] or central nervous system tumor [16] less than 1%
- Stomach cancer [3] 0.5%
- Bile duct cancer [9] small, but increased
- Adrenal gland cancer [8] small, but increased

What are the screening options for FAP?

It is important to discuss with your doctor the following screening options, as each individual is different:

- Yearly screening for hepatoblastoma [5] from birth to age 5 in children at risk, including a physical examination, abdominal ultrasound [17], and a blood test [18] to measure alpha-fetoprotein (AFP) levels
- Yearly flexible sigmoidoscopy [19], beginning between the ages of 10 to 12 for children at risk for FAP
- Colonoscopy [20] once polyps are detected; individuals with classic FAP will typically need a colectomy (the surgical removal of the entire colon) at some point due to the number of polyps and the high risk of colorectal cancer.
- In most cases a total colectomy is recommended to prevent colon cancer in young adults. If any of the rectum remains after surgery, flexible sigmoidoscopy should still be performed regularly.
- Upper endoscopy [21] (EGD) every one to three years, beginning at age 25 or after polyps are detected
- X-ray or computed tomography [22] (CT or CAT) scan of the small bowel if adenomas are found on the EGD or before a colectomy; repeat every one to three years depending on symptoms.
- Yearly physical examination, including thyroid evaluation

Screening options may change over time as new technologies are developed and more is learned about FAP. It is important to talk with your doctor about appropriate screening tests.

Learn more about what to expect when having common tests, procedures, and scans [23].

Questions to ask the doctor

If you are concerned about your risk of colorectal cancer [2] or other types of cancer, talk with your doctor. Consider asking the following questions of your doctor:

- What is my risk of developing colorectal cancer?
- How many colon polyps have I had in total?
- What kind of colon polyps have I had? (The two most common kinds are hyperplastic and adenomatous.)
- What is my risk of developing another type of cancer?
- What can I do to reduce my risk of cancer?
- What are my options for cancer screening?

If you are concerned about your family history and think your family may have FAP, consider asking the following questions:

- Does my family history increase my risk of colorectal cancer?
- Should I meet with a genetic counselor?
- Should I consider genetic testing [24]?

Additional resources

Guide to Colorectal Cancer [2]

The Genetics of Colorectal Cancer [14]

What to Expect When You Meet With a Genetic Counselor [25]

Colon Cancer Alliance

www.ccalliance.org [26]

Colorectal Cancer Coalition (C3)

http://fightcolorectalcancer.org/ [27]

National Cancer Institute

www.cancer.gov [28]

American Cancer Society

www.cancer.org [29]

Cancer*Care*

www.cancercare.org [30]

To find a genetic counselor in your area, ask your doctor or visit the following websites:

National Society of Genetic Counselors

www.nsgc.org [31]

National Cancer Institute: Cancer Genetics Services Directory

www.cancer.gov/cancertopics/genetics/directory [32]

Links:
[1] http://www.cancer.net/about-us
[2] http://www.cancer.net/node/18701
[3] http://www.cancer.net/node/19645
[4] http://www.cancer.net/node/19632
[5] http://www.cancer.net/node/19134
[6] http://www.cancer.net/node/19293
[7] http://www.cancer.net/node/19495
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[12] http://www.cancer.net/node/18869
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[25] http://www.cancer.net/node/24907
[26] http://www.ccalliance.org/
[27] http://fightcolorectalcancer.org/
[28] http://www.cancer.gov
[29] http://www.genetests.org/servlet/access?db=genestar&id=8888891&fcn=e&fcn2=refer&refer=http%3A%2F%2Fwww.cancer.org&type=resource&key=rqddkjwp16MpW
[30] http://www.genetests.org/servlet/access?db=genestar&id=8888891&fcn=e&fcn2=refer&refer=http%3A%2F%2Fwww.cancercare.org&type=resource&key=rqddkjwp16MpW
[31] http://www.nsgc.org/
[32] http://www.cancer.gov/cancertopics/genetics/directory