



Familial adenomatous polyposis

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What is familial adenomatous polyposis?

Familial adenomatous polyposis (FAP) is an inherited disorder characterized by cancer of the large intestine (colon) and rectum. People with the classic type of familial adenomatous polyposis may begin to develop multiple noncancerous (benign) growths (polyps) in the colon as early as their teenage years. Unless the colon is removed, these polyps will become malignant (cancerous). The average age at which an individual develops colon cancer in classic familial adenomatous polyposis is 39 years. Some people have a variant of the disorder, called attenuated familial adenomatous polyposis, in which polyp growth is delayed. The average age of colorectal cancer onset for attenuated familial adenomatous polyposis is 55 years.

In people with classic familial adenomatous polyposis, the number of polyps increases with age, and hundreds to thousands of polyps can develop in the colon. Also of particular significance are noncancerous growths called desmoid tumors. These fibrous tumors usually occur in the tissue covering the intestines and may be provoked by surgery to remove the colon. Desmoid tumors tend to recur after they are surgically removed. In both classic familial adenomatous polyposis and its attenuated variant, benign and malignant tumors are sometimes found in other places in the body, including the duodenum (a section of the small intestine), stomach, bones, skin, and other tissues. People who have colon polyps as well as growths outside the colon are sometimes described as having Gardner syndrome.

A milder type of familial adenomatous polyposis, called autosomal recessive familial adenomatous polyposis, has also been identified. People with the autosomal recessive type of this disorder have fewer polyps than those with the classic type. Fewer than 100 polyps typically develop, rather than hundreds or thousands. The autosomal recessive type of this disorder is caused by mutations in a different gene than the classic and attenuated types of familial adenomatous polyposis.

How common is familial adenomatous polyposis?

The reported incidence of familial adenomatous polyposis varies from 1 in 7,000 to 1 in 22,000 individuals.

What genes are related to familial adenomatous polyposis?

Mutations in the *APC* gene cause both classic and attenuated familial adenomatous polyposis. These mutations affect the ability of the cell to maintain normal growth and

function. Cell overgrowth resulting from mutations in the *APC* gene leads to the colon polyps seen in familial adenomatous polyposis. Although most people with mutations in the *APC* gene will develop colorectal cancer, the number of polyps and the time frame in which they become malignant depend on the location of the mutation in the gene.

Mutations in the *MUTYH* gene cause autosomal recessive familial adenomatous polyposis (also called MYH-associated polyposis). Mutations in this gene prevent cells from correcting mistakes that are made when DNA is copied (DNA replication) in preparation for cell division. As these mistakes build up in a person's DNA, the likelihood of cell overgrowth increases, leading to colon polyps and the possibility of colon cancer.

Related Gene(s)

Changes in these genes are associated with familial adenomatous polyposis.

- APC
- MUTYH

How do people inherit familial adenomatous polyposis?

Familial adenomatous polyposis can have different inheritance patterns.

When familial adenomatous polyposis results from mutations in the *APC* gene, it is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one parent with the condition.

When familial adenomatous polyposis results from mutations in the *MUTYH* gene, it is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. Most often, the parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but do not show signs and symptoms of the condition.

Where can I find information about diagnosis or management of familial adenomatous polyposis?

These resources address the diagnosis or management of familial adenomatous polyposis and may include treatment providers.

- American Medical Association and National Coalition for Health Professional Education in Genetics: Understand the Basics of Genetic Testing for Hereditary Colorectal Cancer (<http://www.nchpeg.org/documents/crc/Basics%20of%20genetic%20testing.pdf>)
- Gene Review: APC-Associated Polyposis Conditions (<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=fap>)
- Gene Tests: APC-Associated Polyposis Conditions (<http://www.ncbi.nlm.nih.gov/sites/>)

GeneTests/lab/clinical_disease_id/2144)

- Gene Tests: MUTYH-Associated Polyposis (http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/213916)
- Genetic Alliance (<http://www.geneticalliance.org/site-search/results/polyposis+fap+adenomatous+familial+taxonomy%3A13>)
- MedlinePlus Encyclopedia: Colon Cancer (<http://www.nlm.nih.gov/medlineplus/ency/article/000262.htm>)
- MedlinePlus Encyclopedia: Colorectal polyps (<http://www.nlm.nih.gov/medlineplus/ency/article/000266.htm>)

You might also find information on the diagnosis or management of familial adenomatous polyposis in Educational resources (<http://ghr.nlm.nih.gov/condition/familial-adenomatous-polyposis/show/Educational+resources>) and Patient support (<http://ghr.nlm.nih.gov/condition/familial-adenomatous-polyposis/show/Patient+support>).

To locate a healthcare provider, see How can I find a genetics professional in my area? (<http://ghr.nlm.nih.gov/handbook/consult/findingprofessional>) in the Handbook.

Where can I find additional information about familial adenomatous polyposis?

You may find the following resources about familial adenomatous polyposis helpful. These materials are written for the general public.

- **MedlinePlus - Health information**
 - Encyclopedia: Colon Cancer (<http://www.nlm.nih.gov/medlineplus/ency/article/000262.htm>)
 - Encyclopedia: Colorectal polyps (<http://www.nlm.nih.gov/medlineplus/ency/article/000266.htm>)
 - Health Topic: Cancer--Living with Cancer (<http://www.nlm.nih.gov/medlineplus/cancerlivingwithcancer.html>)
 - Health Topic: Colonic Polyps (<http://www.nlm.nih.gov/medlineplus/colonicpolyps.html>)
 - Health Topic: Colorectal Cancer (<http://www.nlm.nih.gov/medlineplus/colorectalcancer.html>)
- **Genetic and Rare Diseases Information Center - Information about genetic conditions and rare diseases**
 - Genetic and Rare Diseases Information Center: Attenuated familial adenomatous polyposis (<http://rarediseases.info.nih.gov/GARD/Disease.aspx?PageID=4&>

DiseaseID=8532)

- Genetic and Rare Diseases Information Center: Familial adenomatous polyposis (<http://rarediseases.info.nih.gov/GARD/Disease.aspx?PageID=4&DiseaseID=6408>)
- Genetic and Rare Diseases Information Center: Gardner syndrome (<http://rarediseases.info.nih.gov/GARD/Disease.aspx?PageID=4&DiseaseID=6482>)

- **Additional NIH Resources - National Institutes of Health**

- National Cancer Institute: Colon and Rectal Cancer (<http://www.cancer.gov/cancertopics/types/colon-and-rectal>)
- National Cancer Institute: Genetics of Colorectal Cancer (<http://www.cancer.gov/cancertopics/pdq/genetics/colorectal/HealthProfessional/page1>)
- National Human Genome Research Institute: Learning About Colon Cancer (<http://www.genome.gov/10000466>)
- National Institute of Diabetes and Digestive and Kidney Diseases: Colon Polyps (http://digestive.niddk.nih.gov/ddiseases/pubs/colonpolyps_ez/index.aspx)
- NIH Senior Health: Colorectal Cancer (<http://nihseniorhealth.gov/colorectalcancer/colorectalcancerdefined/01.html>)

- **Educational resources - Information pages**

- American Cancer Society: Colon and Rectum Cancer (<http://www.cancer.org/Cancer/TesticularCancer/OverviewGuide/index?rnav=criov&dt=10>)
- CDC: Colorectal (Colon) Cancer (<http://www.cdc.gov/cancer/colorectal/>)
- Genetic Science Learning Center, University of Utah (<http://learn.genetics.utah.edu/content/disorders/whataregd/colon/index.html>)
- Johns Hopkins Cancer Risk Assessment Program: Polyposis and the MYH Gene (<http://www.macgn.org/factsheets/Polyposis%20and%20MYH%201.pdf>)
- Madisons Foundation (http://www.madisonsfoundation.org/index.php?option=com_mpower&task=disease&diseaseID=381)
- Mayo Clinic (<http://www.mayoclinic.org/familial-adenomatous-polyposis/index.html>)
- Mount Sinai Hospital Kid's Corner: FAP & You (<http://www.zanecohencentre.com/gi-cancers/fgicr/kids-korner/fap-a-you>)
- National Organization for Rare Disorders (<http://www.rarediseases.org/rare-disease-information/rare-diseases/byID/142/viewAbstract>)
- New York Online Access to Health: Colon, Rectal and Anal Cancer

(<http://www.noah-health.org/en/cancer/types/colon/>)

- Orphanet (http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=733)

- **Patient support - For patients and families**

- American Society of Colon and Rectal Surgeons : Inherited Colorectal Cancer Registries (http://www.fascrs.org/patients/family_history_registries/)
- Colon Cancer Alliance (<http://www.ccalliance.org/>)
- Colorectal Cancer Coalition (<http://fightcolorectalcancer.org/>)

You may also be interested in these resources, which are designed for healthcare professionals and researchers.

- Gene Reviews - Clinical summary (<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=fap>)

- **Gene Tests - DNA tests ordered by healthcare professionals**

- Gene Tests: APC-Associated Polyposis Conditions (http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2144)
- Gene Tests: MUTYH-Associated Polyposis (http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/213916)

- ClinicalTrials.gov - Linking patients to medical research (<http://clinicaltrials.gov/ct2/results?cond=%22familial%20adenomatous%20polyposis%22>)
- PubMed - Recent literature ([http://www.ncbi.nlm.nih.gov/pubmed?term=\(Adenomatous%20Polyposis%20Coli%5BMAJR%5D\)%20AND%20\(\(familial%20adenomatous%20polyposis%5BTI%5D\)%20AND%20\(FAP%5BTIAB%5D\)%20AND%20\(colorectal\)\)%20AND%20english%5Bla%5D%20AND%20human%5Bmh%5D%20AND%20%22last%20720%20days%22%5Bdp%5D](http://www.ncbi.nlm.nih.gov/pubmed?term=(Adenomatous%20Polyposis%20Coli%5BMAJR%5D)%20AND%20((familial%20adenomatous%20polyposis%5BTI%5D)%20AND%20(FAP%5BTIAB%5D)%20AND%20(colorectal))%20AND%20english%5Bla%5D%20AND%20human%5Bmh%5D%20AND%20%22last%20720%20days%22%5Bdp%5D))

- **OMIM - Genetic disorder catalog**

- FAMILIAL ADENOMATOUS POLYPOSIS 1 (<http://omim.org/entry/175100>)
- FAMILIAL ADENOMATOUS POLYPOSIS, 2 (<http://omim.org/entry/608456>)
- DESMOID DISEASE, HEREDITARY (<http://omim.org/entry/135290>)

What other names do people use for familial adenomatous polyposis?

- Adenomatous Polyposis Coli
- Adenomatous Polyposis of the Colon
- Colon cancer, familial

- Familial intestinal polyposis
- Familial Multiple Polyposis
- Familial multiple polyposis syndrome
- Familial Polyposis Coli
- Familial Polyposis Syndrome
- FAP
- Hereditary Polyposis Coli
- MYH-associated polyposis
- Polyposis coli

For more information about naming genetic conditions, see the Genetics Home Reference Condition Naming Guidelines (<http://ghr.nlm.nih.gov/ConditionNameGuide>) and How are genetic conditions and genes named? (<http://ghr.nlm.nih.gov/handbook/mutationsanddisorders/naming>) in the Handbook.

What if I still have specific questions about familial adenomatous polyposis?

Ask the Genetic and Rare Diseases Information Center (<http://rarediseases.info.nih.gov/GARD/>).

What glossary definitions help with understanding familial adenomatous polyposis?

autosomal ; autosomal dominant ; autosomal recessive ; benign ; cancer ; cell ; cell division ; colon ; colon polyp ; colorectal ; desmoid ; DNA ; DNA replication ; duodenum ; familial ; gene ; incidence ; inheritance ; inheritance pattern ; intestine ; mutation ; polyp ; polyposis ; recessive ; rectum ; sign ; stomach ; surgery ; symptom ; syndrome ; teenage ; tissue ; tumor

You may find definitions for these and many other terms in the Genetics Home Reference Glossary (<http://ghr.nlm.nih.gov/glossary>).

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The resources on this site should not be used as a substitute for professional medical care or advice. Users seeking information about a personal genetic disease, syndrome, or condition should consult with a qualified healthcare professional. See [How can I find a genetics professional in my area?](http://ghr.nlm.nih.gov/handbook/consult/findingprofessional) (<http://ghr.nlm.nih.gov/handbook/consult/findingprofessional>) in the Handbook.

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