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.

Frequency

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

Causes

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 errors that are made when DNA is copied (DNA replication) in preparation for cell division. As these errors build up in a person’s DNA, the likelihood of cell overgrowth increases, leading to colon polyps and the possibility of colon cancer.

**Inheritance Pattern**
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.

**Other Names for This Condition**
- adenomatous familial polyposis
- adenomatous familial polyposis syndrome
- adenomatous polyposis coli
- familial multiple polyposis syndrome
- FAP
- MYH-associated polyposis

**Diagnosis & Management**

**Genetic Testing Information**
- What is genetic testing? /primer/testing/genetictesting
Research Studies from ClinicalTrials.gov
• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22familial+adenomatous+polyposis%22

Other Diagnosis and Management Resources
• GeneReview: APC-Associated Polyposis Conditions
  https://www.ncbi.nlm.nih.gov/books/NBK1345
• GeneReview: MUTYH-Associated Polyposis
  https://www.ncbi.nlm.nih.gov/books/NBK107219
• Genomics Education Programme (UK): Familial Adenomatous Polyposis
  https://www.genomicseducation.hee.nhs.uk/documents/familial-adenomatous-polyposis/
• Genomics Education Programme (UK): MYH-Associated Polyposis
• MedlinePlus Encyclopedia: Colon Cancer
  https://medlineplus.gov/ency/article/000262.htm
• MedlinePlus Encyclopedia: Colorectal polyps
  https://medlineplus.gov/ency/article/000266.htm
• National Cancer Institute: Genetic Testing for Hereditary Cancer Syndromes

Additional Information & Resources

Health Information from MedlinePlus
• Encyclopedia: Colon Cancer
  https://medlineplus.gov/ency/article/000262.htm
• Encyclopedia: Colorectal polyps
  https://medlineplus.gov/ency/article/000266.htm
• Health Topic: Cancer--Living with Cancer
  https://medlineplus.gov/cancerlivingwithcancer.html
• Health Topic: Colonic Polyps
  https://medlineplus.gov/colonicpolyps.html
• Health Topic: Colorectal Cancer
  https://medlineplus.gov/colorectalcancer.html
Genetic and Rare Diseases Information Center

- Attenuated familial adenomatous polyposis
  https://rarediseases.info.nih.gov/diseases/8532/attenuated-familial-adenomatous-polyposis
- Familial adenomatous polyposis
  https://rarediseases.info.nih.gov/diseases/6408/familial-adenomatous-polyposis
- Gardner syndrome
  https://rarediseases.info.nih.gov/diseases/6482/gardner-syndrome

Additional NIH Resources

- National Cancer Institute: Colorectal Cancer
  https://www.cancer.gov/types/colorectal
- National Cancer Institute: Genetics of Colorectal Cancer
  https://www.cancer.gov/types/colorectal/hp/colorectal-genetics-pdq
- National Human Genome Research Institute: Learning About Colon Cancer
  https://www.genome.gov/Genetic-Disorders/Colon-Cancer
- National Institute of Diabetes and Digestive and Kidney Diseases: Colon Polyps
  https://www.niddk.nih.gov/health-information/digestive-diseases/colon-polyps

Educational Resources

- American Cancer Society: Colon and Rectum Cancer
- CDC: Colorectal (Colon) Cancer
  https://www.cdc.gov/cancer/colorectal/
- Genetic Science Learning Center, University of Utah
  https://learn.genetics.utah.edu/content/disorders/multifactorial/
- Johns Hopkins Cancer Risk Assessment Program
- MalaCards: familial adenomatous polyposis
  https://www.malacards.org/card/familial_adenomatous_polyposis
- 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 (NORD)
  https://rarediseases.org/rare-diseases/familial-adenomatous-polyposis/
• Orphanet: Familial adenomatous polyposis  
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=733

• Your Genome from Wellcome Genome Campus  
  https://www.yourgenome.org/facts/what-is-familial-adenomatous-polyposis

Patient Support and Advocacy Resources

• American Society of Colon and Rectal Surgeons: Hereditary Colorectal Cancer Registries  
  https://www.fascrs.org/hereditary-colorectal-cancer-registries

• Colon Cancer Alliance  
  https://www.ccalliance.org

• Colorectal Cancer Coalition  
  https://fightcolorectalcancer.org

Clinical Information from GeneReviews

• APC-Associated Polyposis Conditions  
  https://www.ncbi.nlm.nih.gov/books/NBK1345

• MUTYH-Associated Polyposis  
  https://www.ncbi.nlm.nih.gov/books/NBK107219

Scientific Articles on PubMed

• PubMed  
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Adenomatous+Polyposis+Coli %5BMAJR%5D%29+AND+%28%28familial+adenomatous+polyposis%5BTI%5D %29+AND+%28FAP%5BTIAB%5D%29+AND+%28colorectal%29%29+AND+e nglish%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp %5D

Catalog of Genes and Diseases from OMIM

• DESMOID DISEASE, HEREDITARY  
  http://omim.org/entry/135290

• FAMILIAL ADENOMATOUS POLYPOSIS 1  
  http://omim.org/entry/175100

• FAMILIAL ADENOMATOUS POLYPOSIS 2  
  http://omim.org/entry/608456
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15089902

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15264271

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12646143

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/24124059

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15264268

- National Cancer Institute: Genetics of Colorectal Cancer 
  https://www.cancer.gov/types/colorectal/hp/colorectal-genetics-pdq

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15660526

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17938238
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12853198

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12606733

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15236166

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