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My46 Trait Profile

Familial adenomatous polyposis

Other Names: Gardner syndrome, Attenuated familial adenomatous polyposis

Familial adenomatous polyposis is a genetic condition characterized by the development of hundreds to thousands of colon polyps that if not removed, have the potential to become cancerous over time. It is caused by mutations in the APC gene which makes the adenomatous polyposis coli protein.

Characteristics of Familial adenomatous polyposis

Familial adenomatous polyposis (FAP) is a genetic condition characterized by the development of hundreds to thousands of colon polyps (abnormal growths that often look like mushrooms) called adenomas. Adenomas are growths that if not removed, have a high chance of becoming cancerous over time. Individuals with FAP usually start to develop colon polyps as a teenager or young adult. Without surgery, the risk of colon cancer in FAP is near 100%. A milder form of FAP called attenuated familial adenomatous polyposis (AFAP) is characterized by less than 100 colon polyps and a 70% lifetime risk of colon cancer.

Most individuals with FAP will also develop polyps in their stomach called fundic gland polyps. However, it is uncommon for someone with FAP to develop stomach cancer. Precancerous polyps and cancer in the duodenum (the first part of the small intestine) may also occur in FAP. Rarely, other cancers may be seen in FAP.

The lifetime cancer risks associated with FAP/AFAP include the following:

Cancer type	Lifetime risk
Colon	70-100%
Duodenal	4-12%
Thryoid	approximately 2%
Stomach	approximately 1%
Brain (typically medulloblastoma)	less than 1%
Liver (hepatoblastoma)	less than 1%

Individuals with FAP are also at increased risk for the following noncancerous (benign) findings:

Feature	Description
CHRPE	Freckle-like spots on the retina (i.e., the tissue that lines the back of the eye)
Desmoid tumor	Tumor most often found in the abdomen

Osteoma	Bony lump or bump on the skull or jaw
Epidermoid cyst	Cyst on the skin

The term Gardner syndrome was previously used for individuals with FAP who had these noncancerous findings. However, it is now known that FAP and Gardner syndrome are the same condition.

Diagnosis/Testing

Both FAP and AFAP are caused by a change or mutation in the APC gene. The APC gene is a tumor suppressor gene which means that it keeps cells from growing too fast. Mutations in the APC gene do not allow the APC protein to work normally and as a result, cells may grow uncontrollably. This uncontrolled growth is what can cause the tumors and cysts in affected individuals.

Prompt diagnosis of FAP is important for early detection and prevention of cancer. While mutations in the APC gene are responsible for causing FAP/AFAP, other conditions may look very similar to FAP/AFAP.

Management/Surveillance

Early detection and prevention of cancer are the goals of FAP management. This occurs through early and regular screenings, in addition to preventative surgeries (e.g., colectomy a surgical procedure to remove all or part of the colon). Although the timing of procedures may vary among individuals with FAP, colon screening typically begins in childhood, followed by surgery when polyps become too numerous to manage. Stomach and duodenal cancer screening typically starts in early adulthood. It is important that individuals seek management with healthcare providers who are experienced with FAP.

Mode of inheritance

FAP is inherited in an autosomal dominant pattern. This means inheriting one APC mutation is enough for an individual to be affected and show signs of FAP. The mutation is usually inherited from a parent, either the mother or the father. However, around 30% of the time, neither parent has FAP. In these cases, FAP occurs for the first time (de novo) in an affected child.

Risk to family members

The risk to family members depends on whether or not the individual with FAP has a parent affected with FAP. If a parent also has FAP, the risk of having a child with FAP is 50% with each pregnancy. If a parent does not have FAP, the risk of other siblings being affected is very low.

Special considerations

None

Resources

FAP Gene Support Group
http://www.fapgene.com
Genetics Home Reference: Familial adenomatous polyposis
http://ghr.nlm.nih.gov/condition/familial-adenomatous-polyposis
Cancer.Net: Familial Adenomatous Polyposis
http://www.cancer.net/cancer-types/familial-adenomatous-polyposis

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