CONDITION

Gardner syndrome

Learn about bone tumor-epidermoid cyst-polyposis, an APC-related condition involving colorectal polyps, benign bone growths, skin cysts, and increased cancer risks.

WHAT IT IS
Inherited APC-associated polyposis condition

BODY SYSTEM
Digestive system

MAIN SYMPTOMS
Blood in stool, bowel habit changes, abdominal cramping, skin cysts

WHAT CAUSES IT
Inherited APC gene change

HOW IT IS DIAGNOSED
Colonoscopy with tissue examination, APC genetic testing

MAIN TREATMENT
Lifelong surveillance and preventive surgery

CAN IT BE CURED?
No, lifelong follow-up remains necessary

IS IT AN EMERGENCY?
Sometimes, severe or sudden abdominal pain

LONG-TERM OUTLOOK
Very high colorectal cancer risk without effective monitoring and treatment

Overview

Bone tumor-epidermoid cyst-polyposis is an older name for the combination of colorectal polyps, benign bone growths called osteomas, and epidermoid skin cysts. This pattern is most often associated with Gardner syndrome , which is now considered part of the broader group of APC-associated polyposis conditions .

The main APC-associated condition is familial adenomatous polyposis, often shortened to FAP. People with FAP develop many adenomatous polyps in the colon and rectum. Adenomas are growths that are not cancer, but some can become cancer over time. Without effective monitoring and treatment, the chance of colorectal cancer becomes very high.

The "bone tumors" in the older name are usually osteomas. These are noncancerous areas of extra bone, often affecting the jaw or skull. Epidermoid cysts are usually noncancerous lumps

beneath the skin that contain keratin, a protein found in skin, hair, and nails.

A person does not need to have all three features to have an APC-associated condition. Some people mainly develop colorectal polyps, while others have more noticeable findings outside the bowel. These may include dental abnormalities, desmoid tumors, eye findings, stomach or small-intestinal polyps, and certain cancers.

Recognizing the condition matters because colorectal polyps may not cause symptoms until they are advanced. Diagnosis allows a person to begin appropriate surveillance, discuss preventive treatment, and offer genetic testing to relatives who may also be at risk.

Symptoms

Symptoms vary, even among members of the same family. Some findings, such as jaw osteomas or skin cysts, may appear before intestinal symptoms.

Colorectal polyps often cause no symptoms. When symptoms occur, they may include:

Osteomas are usually hard, slow-growing, painless lumps. They most often occur in the jaw or skull but can develop in other bones. A jaw osteoma may change facial shape or be found during dental imaging.

Dental findings can include extra teeth, teeth that do not emerge normally, missing teeth, or unusual tooth roots. These changes may be noticed by a dentist before the intestinal condition is identified.

Epidermoid cysts are round lumps under the skin. They commonly occur on the face, scalp, arms, or legs. They may remain small and painless. If a cyst becomes inflamed or infected, it can become red, warm, swollen, or tender and may drain material.

Some people develop desmoid tumors . These are growths made from connective tissue. They do not spread to distant organs like cancer, but they can grow into nearby tissues. Desmoid tumors may form in the abdominal wall or inside the abdomen, sometimes after abdominal surgery. Symptoms depend on their size and location and can include a firm mass, pain, vomiting, or bowel blockage.

Other possible findings include polyps in the stomach or duodenum, which is the first part of the small intestine. A harmless eye finding called congenital hypertrophy of the retinal pigment epithelium may be seen during an eye examination. Some APC-associated conditions also increase the risk of thyroid and other less common cancers.

- Blood in or on the stool
- A change in usual bowel habits

- Diarrhea or constipation that does not settle
- Abdominal discomfort or cramping
- Unexplained iron-deficiency anemia, which can cause tiredness or shortness of breath
- Unintentional weight loss

Causes and risk factors

The condition is usually caused by a disease-causing change, also called a pathogenic variant, in the APC gene . This gene normally helps control cell growth. When it does not work properly, cells in the lining of the digestive tract are more likely to form adenomatous polyps.

APC-associated polyposis is inherited in an autosomal dominant pattern. This means a person with an APC variant has a 1 in 2 chance of passing it to each child. The chance is the same for every pregnancy, regardless of the child's sex.

Some people inherit the variant from an affected parent. In others, the variant begins as a new genetic change, so there may be no known family history. A person with a new variant can still pass it to their children.

The type and location of an APC variant can influence the pattern of disease, but they do not predict every outcome. Relatives carrying the same variant may have different numbers of polyps or different findings outside the colon.

A family history of numerous colorectal polyps, colorectal cancer at a young age, desmoid tumors, jaw osteomas, or several relatives with related findings may raise concern for an inherited polyposis condition. Other genes can also cause multiple colorectal polyps, so evaluation should not rely on symptoms or family history alone.

Diagnosis

Diagnosis usually combines medical history, family history, physical examination, endoscopy, and genetic testing.

A clinician may ask about colorectal polyps and cancer in parents, siblings, children, grandparents, aunts, uncles, and cousins. Information about ages at diagnosis is useful. A history of colectomy, jaw growths, desmoid tumors, or unexplained abdominal tumors can also provide important clues.

A colonoscopy allows a clinician to examine the colon and rectum using a flexible camera. Polyps can be counted, sampled, or removed. A pathologist then examines the tissue to

determine whether the polyps are adenomas and whether any show advanced changes or cancer.

Upper endoscopy may be used to examine the stomach and duodenum. The area where the bile and pancreatic ducts enter the duodenum, called the ampulla, requires careful assessment because polyps can develop there.

Genetic counseling is recommended before or alongside genetic testing. A blood or saliva test can look for a pathogenic APC variant. Testing may also include other genes associated with colorectal polyposis, especially when the person's features do not clearly identify one condition.

A positive APC result can confirm the diagnosis and guide testing for relatives. A negative result does not always rule out an inherited condition. Testing may not detect every genetic change, and some people with multiple polyps have changes in other genes or no currently identifiable cause.

Once a familial APC variant is known, relatives can have targeted testing for that specific variant. Those who did not inherit it generally do not need APC-related surveillance beyond care based on their personal and general population risks.

Treatment and management

Management is designed to prevent cancer, control symptoms, and monitor organs that may be affected. Care often involves a gastroenterologist, colorectal surgeon, medical geneticist or genetic counselor, pathologist, and primary care clinician. Other specialists may be needed for desmoid tumors, thyroid findings, dental problems, or skin cysts.

Regular colonoscopy can monitor polyp development and remove selected polyps. However, endoscopic removal may become impractical when polyps are too numerous, large, rapidly increasing, or showing concerning tissue changes.

Surgery to remove the colon is a central preventive treatment for classic FAP. The timing and type of operation depend on the number and features of polyps, involvement of the rectum, cancer findings, symptoms, overall health, and personal priorities.

One approach removes the colon while connecting the small intestine to the rectum. Another removes both the colon and rectum and creates an internal pouch from the small intestine. Each option has possible benefits and long-term effects on bowel function. Surgery reduces colorectal cancer risk but does not remove the need for follow-up. Polyps can still develop in remaining rectal tissue, an intestinal pouch, or other parts of the digestive tract.

Upper endoscopy is used to monitor duodenal and stomach polyps. The interval and treatment

plan depend on the number, size, location, and microscopic features of the polyps. Some can be removed endoscopically, while advanced disease may require specialized surgery.

Certain medicines can reduce colorectal or duodenal polyp burden in some people. They do not reliably replace surgery or endoscopic surveillance and can cause significant side effects. Medication decisions should be made with a specialist familiar with inherited polyposis.

Osteomas usually do not require treatment unless they cause pain, interfere with function, or create a major cosmetic concern. Epidermoid cysts may be observed or removed. An inflamed cyst needs medical assessment because inflammation does not always mean bacterial infection.

Desmoid tumors require individualized management by an experienced multidisciplinary team. Depending on the tumor's behavior and location, options may include observation, medication, or surgery. Immediate surgery is not appropriate for every desmoid tumor because some remain stable and surgery itself can be followed by additional growth.

Complications

The most important complication is colorectal cancer. The risk is driven by the continuing development of adenomatous polyps, which is why surveillance and preventive surgery are essential parts of care.

Polyps in the duodenum or around the ampulla can also become cancerous. Other cancer risks associated with APC variants may include thyroid, pancreatic, liver, brain, and stomach cancers, although the size and timing of these risks vary.

Desmoid tumors can press on the intestines, blood vessels, ureters, or other structures. This can cause pain, bowel obstruction, reduced blood flow, or kidney problems. Their behavior is unpredictable: some grow, some remain stable, and some shrink without treatment.

Complications can also result from repeated procedures or major bowel surgery. Possible concerns include bleeding, infection, bowel obstruction, changes in stool frequency, dehydration, and inflammation or polyps in remaining intestinal tissue. The surgical team should explain expected recovery and long-term bowel function before an operation.

The emotional effects of a hereditary condition can be substantial. People may worry about cancer, surgery, body image, bowel function, or passing the condition to children. Genetic counseling and mental health support can be useful parts of care.

Follow up

Follow-up continues throughout life, including after colon surgery. A personalized plan may include examinations of the remaining rectum or intestinal pouch, upper endoscopy, assessment for desmoid tumors, and evaluation of other APC-associated findings. The exact schedule depends on age, previous test results, the operation performed, and family history.

Routine appointments are appropriate for reviewing surveillance results, discussing new but stable skin or bone lumps, assessing bowel function, and arranging family genetic testing. New abdominal swelling, a persistent change in bowel habits, unexplained anemia, or blood in the stool should be reported promptly rather than waiting for the next scheduled visit.

Seek urgent medical assessment for worsening abdominal pain, repeated vomiting, increasing abdominal swelling, fever after a procedure, or a painful and rapidly enlarging mass.

Emergency care is appropriate for severe or sudden abdominal pain, vomiting with inability to pass stool or gas, heavy rectal bleeding, fainting, confusion, or signs of severe dehydration. These symptoms can have many causes, but they may indicate bowel obstruction, significant bleeding, or another complication needing immediate treatment.

Sources