# Gardner Syndrome

**This section has been reviewed and approved by the Cancer.Net Editorial Board [1], May / 2012**

Overview

**What is Gardner syndrome?**

Gardner syndrome is a subtype of familial adenomatous polyposis (FAP or classic FAP) [2]. In people with Gardner syndrome, masses of noncancerous tissue tend to form in many different organs, such as:

- Multiple adenomatous colon polyps. An adenomatous polyp is an area where the normal cells that line the inside of the colon and begin to make mucous and form a mass on the inside of the intestinal tract.
- Benign (noncancerous) tumors, including:
  - sebaceous cysts (closed sac filled with liquid found under the skin)
  - epidermoid cysts (lumps in or under the skin often filled with liquid)
  - fibromas (fibrous tumors)
  - desmoid tumors (fibrous tumors that can develop anywhere in the body)
  - osteomas (bony growths, usually found on the jaw)

People with Gardner syndrome also have a higher risk of developing colorectal cancer [3] and other FAP-related cancers [2]. Other features of Gardner syndrome that are similar to classic FAP include extra or unerupted teeth and congenital (present at birth) hypertrophy of the retinal pigment epithelium (CHRPE), an eye condition that does not affect vision but which a doctor can find by doing an examination with a special instrument called an ophthalmoscope.

**What causes Gardner syndrome?**

Gardner syndrome is a genetic condition. This means that the risk of Gardner syndrome can be passed from generation to generation in a family. The *APC* gene is linked to Gardner syndrome; *APC* stands for adenomatous polyposis coli. A mutation (alteration) in the *APC* gene gives a person an increased lifetime risk of developing polyps, benign tumors, and cancer.

**How is Gardner syndrome inherited?**

Normally, every cell has two copies of each gene: one inherited from the mother and one inherited from the father. Gardner syndrome follows an autosomal dominant inheritance pattern, in which 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 gene mutation also has a 50% chance of having the same mutation.

**How common is Gardner syndrome?**

Gardner syndrome is considered to be rare.

**How is Gardner syndrome diagnosed?**

A person with multiple adenomatous colon polyps and/or colorectal cancer [3] along with some of the benign tumors listed above is suspected of having Gardner syndrome. People suspected to have Gardner syndrome can 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 Gardner syndrome if they are tested and have the same gene mutation.

**What are the estimated cancer risks associated with Gardner syndrome?**

The cancer risks for Gardner syndrome are similar to those for classic FAP [2]. Cancer risks for classic FAP include:

- Colorectal cancer [3] almost 100% if not treated
- Small bowel [4] (intestines) 4% to 12%
- Pancreatic cancer [5] 2%
- Papillary thyroid cancer [6] 2%
- Hepatoblastoma [7] (a type of liver cancer) 1.5%
- Brain [8] or central nervous system tumor [9] less than 1%
- Stomach cancer [10] 0.5%
- Bile duct cancer [11] small, but increased
- Adrenal gland cancer [12] small, but increased

**What are the screening options for Gardner syndrome?**

The screening options for Gardner syndrome are considered to be similar to those for classic FAP, with the addition of regular dermatologic (skin) examinations.

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

Screening options for classic FAP [2] include:

- Yearly screening for hepatoblastoma [7], from birth to age 5 in children at risk, including a physical examination, abdominal ultrasound [13], and a blood test [14] to measure alpha-fetoprotein (AFP) levels
- Yearly flexible sigmoidoscopy [15], beginning between the ages of 10 to 12 for children at risk for FAP
- Colonoscopy [16] 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 [3]
- Upper endoscopy [17] (EGD) every one to three years, beginning at age 25 or after polyps are detected
- X-ray or computed tomography [18] (CT or CAT) scan of the small bowel if adenomas are found on EGD or before a colectomy; repeat every one to three years depending on symptoms
- Yearly physical examination, including thyroid evaluation
- Yearly dermatologic examination of skin (or more frequently if necessary)

Screening options may change over time as new technologies are developed and more is learned about Gardner syndrome. 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 [19].

**Questions to ask the doctor**

If you are concerned about your risk of colorectal cancer [3] 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 cumulative (total) colon polyps have I had?
- What type of colon polyps have I had? (The two most common kinds are hyperplastic and adenomatous.)
- What is my risk of developing other types 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 Gardner syndrome, consider asking the following questions:

- Does my family history increase my risk of colorectal cancer [3] or other types of cancer?
- Should I meet with a dermatologist (a doctor who specializes in diseases and conditions of the skin)?
- Should I meet with a genetic counselor?
- Should I consider genetic testing [20]?

**Additional resources**

Guide to Colorectal Cancer [3]

The Genetics of Colorectal Cancer [21]

What to Expect When You Meet With a Genetic Counselor [22]

**Colon Cancer Alliance**
www.ccalliance.org [23]

**Colorectal Cancer Coalition (C3)**
http://fightcolorectalcancer.org [24]

**National Cancer Institute**
www.cancer.gov [25]

**American Cancer Society**
www.cancer.org [26]

**Cancer*Care***
www.cancercare.org [27]

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

National Society of Genetic Counselors
www.nsgc.org [28]

National Cancer Institute: Cancer Genetics Services Directory
http://www.cancer.gov/cancertopics/genetics/directory [29]

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**Links:**
[1] http://www.cancer.net/about-us
[2] http://www.cancer.net/node/18852
[3] http://www.cancer.net/node/18701
[4] http://www.cancer.net/node/19632
[5] http://www.cancer.net/node/19495
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[27] http://www.cancercare.org/
[28] http://www.nsgc.org/
[29] http://www.cancer.gov/cancertopics/genetics/directory