Gardner Syndrome [1]

What is Gardner syndrome?

Gardner syndrome is a subtype of familial adenomatous polyposis (FAP or classic FAP) [2], which usually causes benign, meaning noncancerous, tumors 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 begin to make mucus and form a mass on the inside of the intestinal tract.
- Benign tumors, including:
  - sebaceous cysts, which are closed sacs filled with liquid found under the skin
  - epidermoid cysts, which are lumps in or under the skin often filled with liquid
  - fibromas, which are fibrous tumors
  - desmoid tumors, which are fibrous tumors that can develop anywhere in the body
  - osteomas, which are 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 can be seen by looking at the retina using 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, meaning an 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.

Options exist for couples interested in having a child when they know that one of them carries a gene mutation that increases the risk for this hereditary cancer syndrome. Preimplantation Genetic Diagnosis (PGD) is a medical procedure done in conjunction with in-vitro fertilization (IVF). It allows people who carry a specific known genetic mutation to have children who do not carry the mutation. A woman's eggs are removed and fertilized in a laboratory. When the embryos reach a certain size, one cell is removed and is tested for the hereditary condition in question. The parents can then choose to transfer embryos which do not have the mutation. PGD has been in use for over a decade, and more recently has been used for several hereditary cancer predisposition syndromes. For more information, talk with an assisted reproduction specialist at a fertility clinic.

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 (intestines) cancer [4]	4% to 12%
• Pancreatic cancer [5]	2%
• Papillary thyroid cancer [6]	2%

• Hepatoblastoma [7], which is 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 skin examinations by a dermatologist, a doctor who specializes in diseases and conditions of the skin.

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, meaning 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 developing cancer, talk with your doctor. Consider asking the following questions of your doctor:

- What is my risk of developing colorectal cancer?
- How many colon polyps have I had in total?
- 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?
- Should I meet with a genetic counselor?
- Should I consider genetic testing [20]?

Additional resources

Guide to Colorectal Cancer [3]

What to Expect When You Meet With a Genetic Counselor [21]

Colon Cancer Alliance
www.ccalliance.org [22]

Colorectal Cancer Coalition (C3)
http://fightcolorectalcancer.org [23]

National Cancer Institute
www.cancer.gov [24]

American Cancer Society
www.cancer.org [25]

Cancer*Care*
www.cancercare.org [26]

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

National Society of Genetic Counselors
www.nsgc.org [27]

National Cancer Institute: Cancer Genetics Services Directory
www.cancer.gov/cancertopics/genetics/directory [28]

Links
[1] http://www.cancer.net/cancer-types/gardner-syndrome
[2] http://www.cancer.net/node/18852
[3] http://www.cancer.net/node/31317
[4] http://www.cancer.net/node/31377
[5] http://www.cancer.net/node/31388
[6] http://www.cancer.net/node/31262
[7] http://www.cancer.net/node/31274
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[23] http://fightcolorectalcancer.org/
[24] http://www.cancer.gov/
[25] http://www.cancer.org/
[26] http://www.cancercare.org/
[27] http://www.nsgc.org/
[28] http://www.cancer.gov/cancertopics/genetics/directory