Familial GIST Syndrome

Primary familial GIST syndrome is a rare inherited condition that increases a person's risk of developing **gastrointestinal stromal tumors (GISTs)**.

depending on which gene change is causing the syndrome (see “What are the symptoms of primary familial GIST syndrome?” below).

How common is primary familial GIST syndrome?

Primary familial GIST is rare. There are an estimated 50 to 100 known families across the world with this diagnosis.

What causes primary familial GIST syndrome?

Primary familial GIST syndrome is passed down in families through an inherited gene change (mutation). Most often, this syndrome is caused by changes in 1 of 2 genes:

Most often, this syndrome is caused by an abnormal ***KIT gene*** that is passed from parent to child. This is the same gene that is mutated (changed) in most sporadic (non-inherited) GISTs. (See What Causes Gastrointestinal Stromal Tumors?[2]) People who inherited this abnormal gene from a parent have it in all their cells, while people with sporadic GISTs only have it in the cancer cells.

Less often, an inherited change in the ***PDGFRA gene*** causes this syndrome. (Defects in the *PDGFRA* gene are also found in a small percentage of sporadic GISTs.)

Researchers continue to look for specific genes that may be related to the development of familial GIST and which may be used to help decide the patient’s treatment plan.

Options exist for people who carry a *KIT* or *PDGFRA* gene mutation and might want to have children. For more information, talk with an assisted reproduction specialist at a fertility clinic.

What are signs and symptoms of primary familial GIST syndrome?

People with primary familial GIST syndrome tend to develop GISTs before age 50. They are also more likely to have more than one GIST.

GISTs can cause symptoms such as:

- Pain or discomfort in the abdomen (belly)
- A mass or swelling in the abdomen
- GI bleeding, which might show up as dark stools or blood in the stool
- Fatigue due to anemia (having too few red blood cells)

Some GISTs might spread to other parts of the body, such as the liver, omentum (fat tissue in the abdominal cavity), or peritoneum (membrane lining the abdominal cavity), where they might cause other symptoms.

People with primary familial GIST syndrome might also have other health issues, depending on which gene change is causing the syndrome.

When this syndrome is caused by a change (mutation) in the *KIT* gene, other issues

- Two or more close relatives have been diagnosed with a GIST
- A person with a GIST also has a close relative with another rare tumor type
- A person with a GIST also has neurofibromatosis type 1 (NF1), multiple moles, or uncommon skin issues

To diagnose primary familial GIST syndrome, a person must have an inherited (germline) mutation in either the *KIT* or *PDGFRA* gene. This can be determined with genetic testing[6] of a sample of a person's saliva or blood. A doctor or genetic counselor can review your personal and family health history to help determine if you should consider this type of testing.

It is important to note that genetic testing for primary familial GIST syndrome is still evolving. There may be other genes that cause the syndrome, and current tests may not identify all families who have mutations in those specific genes.

What types of cancer are linked to primary familial GIST syndrome?

People with familial GIST syndrome have an increased risk of developing one or more GISTs. Having a mutation in the *KIT* gene is linked with a particularly high risk of developing one or more GISTs during a person's lifetime. However, not everyone with a known mutation will develop a GIST.

It is not known if people with primary familial GIST syndrome have an increased risk of other types of cancer.

How often should people with primary familial GIST syndrome be screened for cancer before a tumor develops?

Currently, there is no standard screening schedule for people who have a known genetic mutation but who have not developed a GIST. Your doctor will work with you to determine a personal screening plan that is best for you.

In general, different types of imaging tests such as CT or MRI scans, or endoscopy procedures might be done to monitor people for the development of GISTs.

Screening options may change over time as new technologies are developed and more is learned about primary familial GIST syndrome.

How is primary familial GIST syndrome managed?

Treatment options for GISTs depend on several factors, including where the tumor(s) is, if it has spread, and if tumors are affecting other parts of the body.

There is some evidence that familial GISTs tend to spread slower than sporadic GISTs, which may affect treatment decisions. Given the rare nature of this disease, people are encouraged to talk with doctors who have specific expertise in this area of medicine.

To learn more about how GISTs are treated, see Treating Gastrointestinal Stromal Tumors[7].

Questions to ask the health care team

If you are concerned about your risk of developing a GIST, talk with your health care team. It might be helpful to bring someone along to your appointments to take notes. Consider asking your health care team the following questions:

- What is my risk of developing a GIST?
- What is my risk of developing other types of cancer?
- What can I do to lower my risk of developing cancer?
- What cancer screening tests should I have?

If you are concerned about your family history and think your family may have primary familial GIST syndrome, consider asking the following questions:

- Does my family history increase my risk of developing a GIST?
- Should I meet with a genetic counselor or another genetics specialist? Can you refer me?
- Should I consider genetic testing?
- Should my family members consider testing?

Hyperlinks

1. www.cancer.org/cancer/types/gastrointestinal-stromal-tumor.html
2. www.cancer.org/cancer/types/gastrointestinal-stromal-tumor/causes-risks-prevention/what-causes.html
3. www.cancer.org/cancer/types/gastrointestinal-stromal-tumor/detection-diagnosis-

staging/how-diagnosed.html
4. www.cancer.org/cancer/diagnosis-staging/tests/endoscopy/upper-endoscopy.html
5. www.cancer.org/cancer/diagnosis-staging/tests/endoscopy/colonoscopy.html
6. www.cancer.org/cancer/risk-prevention/genetics/genetic-testing-for-cancer-risk.html
7. www.cancer.org/cancer/types/gastrointestinal-stromal-tumor/treating.html

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