When would someone become suspicious that his/her family has familial GIST?

Any of the following features may suggest that familial GIST is present, however it is important to remember that Familial GIST is very rare. You should not overly concern yourself until you consult with your physician:

- One person in the family who has developed more than one separate primary (new GIST, not a metastasis) GIST tumor
- Two or more relatives with GIST
- One person in the family with GIST who also has a close relative with another rare type of tumor
- One person in the family with GIST who also has a personal or family history of unusual skin findings, multiple moles or Neurofibromatosis Type 1

Hyperpigmentation

Research

The Life Raft Group is cooperating with some of the top GIST specialty centers in the Project FLAG study. Project FLAG is a research study to learn more about Familial GIST, also known as hereditary GIST. Since only a small number of families with hereditary GIST have been described in the medical literature, not much is known about familial GIST.

Key goals of Project FLAG are to:

- Look at which genes cause GIST to develop in families
- Learn what other cancers may develop in families with GIST
- Learn about other symptoms, such as non-cancerous skin growths, that may be associated with hereditary GIST.

By looking at the genes of people with GIST, Project FLAG can help to clarify the risk for close family members to develop GIST. By looking at a large number of families with GIST, they hope to collect information that will help to develop cancer screening recommendations for people who have a risk of developing GIST. This would mean that GIST could possibly be found earlier and may be easier to treat.

People who take part in Project FLAG may also agree to be part of a registry of GIST families. These families can decide to take part in studies of hereditary GIST in the future.

If you have a diagnosis of GIST and are 18 years or older, you are eligible for FLAG.

There are no additional requirements: even if you do not have a family history of GIST your participation is needed! Enroll at www.ProjectFLAG.org

Familial GIST: Does it Affect Me?

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The Life Raft Group is an international patient support organization for people with GIST and their family members, whose mission is to ensure the survival of GIST patients while maintaining the quality of their lives. To accomplish this mission the Life Raft Group devotes its efforts to information and support, treatment surveillance, research, patient consultation and assistance, and advocacy.

Contact Us

The Life Raft Group
155 US Highway 46, Suite 202
Wayne, NJ 07470
Phone: 973-837-9092
Fax: 973-837-9095
www.liferaftgroup.org
E-mail: liferaft@liferaftgroup.org

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The Basics

Cancer arises from gene mutations. For most GISTS, the mutations are somatic, which means they are confined to one area of the body and not passed down. Germ-line mutations are heritable, the mutation is present in the egg or sperm of a parent and passed down to his/her children. The mutation is then present in all of the children's cells. This means that they can test family members for familial GIST relatively simply: by taking blood. If the inherited mutation is present in all cells of the body, it will be present in blood. While the exact genetic alteration might not be the same from one family to another, it will be the same within a family.

Features of Familial GIST

Some of the features seen in familial GIST cases are:
- Multiple family members with GIST
- More than one generation with GIST
- Specific skin findings: areas with more or less pigmentation, certain types of moles, a specific rash called urticaria and swallowing disorders

At this time, there is no evidence that inherited GIST is any more or less sensitive to modern treatments than non-inherited GIST. If you suspect that your family may have an inherited form of GIST, your first step should be to discuss your concerns with a physician or genetic health professional. Based on this discussion and your family history, the next step may be to undergo genetic testing.

The Numbers

We have a lot to learn about familial GIST. In a small number of families where more than one relative has GIST, mutations have been found in the known GIST susceptibility genes. Family members who carry the familial gene mutation seem to be more likely to develop GIST.
- Close relatives (siblings, parents) have up to a 50% chance of also carrying the mutation that was found in the patient.
- Parents have a 50% chance of passing the mutation on to each of their children.

Genetic Testing

Some reasons why you may want to get testing are:
- Find a reason for your cancer
- Find out your risk of developing a GIST (if you have not been diagnosed with GIST)
- Find out the cancer risks for siblings and children
- Find out what steps should be taken to reduce risk

If a heritable mutation is found in the GIST patient, close relatives can then be tested to see whether they also carry the familial alteration. There is limited information on the cancer risks associated with germline alterations in the GIST-susceptibility genes. We also need to learn more about what type of screening is most effective for detecting GISTS or other cancers early and whether those cancers can be prevented.

Research is underway to learn more about cancer risks and screening in families with GIST. It is unknown whether most insurance companies will cover genetic testing for familial GIST. However, some patients may be eligible for testing through research.

Due to the sensitivity of this issue, genetic testing should involve a genetic health professional. To try and locate a genetic counselor in your area, you can go to the website for the National Society of Genetic Counselors (www.nsgc.org).

Actions & Reactions

When faced with the possibility of a hereditary risk in the family, people can experience a range of emotions. Family members may ask themselves questions like, Did I give my disease to my children? Did my father pass GIST on to me? and What if my sister has the mutation and I don’t? All of these questions are valid, but can be quite stressful.

A genetic counselor or other genetics professional can help you with these questions and can also help you and your family navigate through the testing process. Genetic counseling can also help to identify which family members would be at risk to carry a gene alteration.