CONDITION

# Gastrointestinal stromal tumors

Gastrointestinal stromal tumors are uncommon growths that develop in the wall of the digestive tract.

**WHAT IT IS**
Tumor arising in the digestive tract wall

**BODY SYSTEM**
Digestive system

**MAIN SYMPTOMS**
Abdominal pain, bleeding, early fullness, tiredness

**WHAT CAUSES IT**
Acquired gene changes, usually without a known trigger

**HOW IT IS DIAGNOSED**
Biopsy or surgical tissue examined by pathology

**MAIN TREATMENT**
Surgery and mutation-matched targeted medicines

**CAN IT BE CURED?**
Sometimes - surgery cures some localized tumors

**IS IT AN EMERGENCY?**
Sometimes - heavy bleeding or sudden severe abdominal pain

**LONG-TERM OUTLOOK**
Driven by spread, mutation, and treatment response

## Overview

A gastrointestinal stromal tumor, or GIST, is a growth that starts in the wall of the digestive tract. Most develop in the stomach or small intestine, although they can occur elsewhere in or near the digestive system.

GISTs differ from common cancers that begin in the lining of the stomach or bowel. They arise from cells related to the interstitial cells of Cajal, which help coordinate the muscle movements that move food through the digestive tract.

Some GISTs are small and slow-growing. Others invade nearby structures or spread, most often to the liver or the lining of the abdomen. Doctors estimate risk using the tumor's size, location, rate of cell division, rupture status, and genetic features.

A GIST may be found during a scan or procedure performed for another reason. Even one that causes no symptoms needs specialist assessment because symptoms alone cannot predict its behavior.

## Symptoms

Small GISTs may cause no symptoms. When symptoms occur, they depend on the tumor's location, size, and whether it bleeds.

Possible symptoms include:

Anemia can develop when a GIST bleeds slowly into the digestive tract. Bleeding may cause black, sticky stools or, particularly lower in the bowel, red blood.

Faster bleeding may cause vomiting of blood, dizziness, fainting, or a rapid heartbeat. A tumor can also narrow the stomach or bowel and prevent food or stool from passing, causing an obstruction.

These symptoms have many possible causes and do not necessarily mean you have a GIST.

- Abdominal pain or discomfort
- Feeling full after eating a small amount
- Nausea, vomiting, or loss of appetite
- A lump or swelling in the abdomen
- Unplanned weight loss
- Tiredness, weakness, or shortness of breath from anemia

## Causes and risk factors

Most GISTs develop after a digestive-tract cell acquires a mutation in a gene that controls growth. The mutation is usually limited to the tumor cells, so it was not inherited and cannot be passed to children.

Many GISTs have a mutation in KIT . Others have changes in PDGFRA . These mutations can keep growth signals switched on. Less commonly, GISTs involve succinate dehydrogenase, known as SDH, or genes in the RAS signaling pathway.

The cause of these acquired mutations is usually unknown. GIST is not known to result from a particular food, injury, or everyday behavior.

Rare inherited forms include those associated with neurofibromatosis type 1, inherited KIT or

PDGFRA mutations, and some SDH-related conditions. They may occur at a younger age or cause multiple tumors. If your history suggests an inherited form, genetic counseling can explain testing and what a result could mean for relatives.

## Diagnosis

A contrast-enhanced CT scan of the abdomen and pelvis can show a mass and whether it has spread. MRI may provide more detail in selected areas or be used when CT contrast is unsuitable. PET scans are sometimes used to assess an early treatment response but are not needed for everyone.

During an endoscopy, a thin camera is used to examine the stomach or bowel. Endoscopic ultrasound can show which layer of the digestive wall contains the mass and may guide a biopsy.

A biopsy removes tissue for examination by a pathologist. Special tests look for proteins such as KIT, also called CD117, and DOG1. Tissue removed during surgery is assessed in the same way.

Biopsy must be planned carefully because puncturing or rupturing the tumor can release cells into the abdomen. Some clearly removable tumors are taken out without a separate biopsy. A biopsy is usually needed before drug treatment or when the diagnosis is uncertain.

Molecular testing identifies mutations such as KIT and PDGFRA and helps determine which targeted medicines may work. After surgery, the pathology report records the tumor's size, location, mitotic rate, surgical margins, and whether it ruptured. These details help estimate the chance of recurrence.

## Treatment and management

Treatment is usually planned by a team experienced in GIST and other sarcomas.

Surgery is the main treatment for a localized GIST that can be removed safely. The aim is to remove it intact with a margin of healthy tissue. Extensive lymph-node removal is not usually needed because GIST rarely spreads first to lymph nodes.

Selected very small stomach tumors may be monitored with endoscopic ultrasound instead of being removed immediately. This requires specialist review and scheduled follow-up.

Targeted medicines block growth signals in tumor cells. Imatinib works against many GISTs with sensitive KIT or PDGFRA mutations. It may be given before surgery to shrink a difficult

tumor or afterward when recurrence risk is significant.

Mutation testing is essential because responses differ. For example, GIST with a PDGFRA D842V mutation generally does not respond to imatinib, while avapritinib can target this mutation. If an advanced GIST stops responding or treatment cannot be tolerated, options may include sunitinib, regorafenib, or ripretinib.

For GIST that has spread, targeted treatment may control growth for long periods. Treatment commonly continues while it is effective and side effects remain manageable. Surgery or another local procedure may help in selected cases.

Side effects vary and can include swelling, nausea, diarrhea, tiredness, skin changes, high blood pressure, and changes in blood counts or liver tests. Regular blood tests and scans monitor safety and response. Tell your team about all prescriptions, over-the-counter medicines, and supplements because interactions can alter drug levels. Do not stop targeted treatment without speaking to your team.

Standard chemotherapy and radiotherapy usually have limited roles, although radiotherapy may occasionally relieve symptoms in a specific area.

## Complications

A GIST can bleed into the stomach or bowel. Slow bleeding may cause anemia, while heavy bleeding can cause fainting and circulatory problems.

A growing tumor may obstruct the digestive tract. Rarely, it creates a hole in the stomach or bowel, called a perforation. Tumor rupture can release cells into the abdomen and raises the risk of recurrence.

GIST can return after surgery or spread, particularly to the liver or abdominal lining. Targeted medicines may also stop working when tumor cells acquire additional mutations, but other treatments may remain available.

## Prevention

Most GISTs cannot currently be prevented. No diet, supplement, or lifestyle change has been proven to stop the acquired mutations responsible for most cases, and there is no routine screening program for people without symptoms or inherited risk.

People with a rare inherited condition linked to GIST may be offered specialist monitoring. Genetic counseling can help relatives understand whether testing or follow-up is relevant. This

supports earlier detection but does not guarantee prevention.

## Living with it and what to expect

The outlook depends on where the GIST began, whether it was completely removed, its mutation and other risk features, and whether it has spread.

After surgery for localized disease, some people need only scheduled scans. Others take targeted medicine to reduce recurrence risk. Follow-up often continues for years because GIST can return long after treatment. The scan schedule is tailored to the tumor's risk and treatment stage.

A GIST that has spread may be managed as a long-term condition with ongoing targeted treatment. Scans show whether tumors are shrinking, stable, or growing. Growth in one area does not always mean every tumor has stopped responding.

Many people continue work, exercise, family activities, and travel, although side effects and appointments may require adjustments. Take targeted medicine consistently. If side effects interfere with eating, drinking, sleep, movement, or daily life, contact your team rather than changing or stopping treatment yourself.

Living between scans can be stressful. A written treatment record, planning for scan days, and support from a sarcoma service or support group may help. Emotional support is appropriate during diagnosis, remission, and long-term treatment.

## When to get medical help

Arrange a routine appointment for ongoing abdominal discomfort, unexplained early fullness, persistent nausea, unplanned weight loss, unusual tiredness, or a lasting change in bowel habits. These symptoms are often caused by other conditions, but persistent changes need assessment.

Contact your treatment team promptly for new black stools, mild bleeding, worsening pain, repeated vomiting, increasing abdominal swelling, or side effects that make it hard to eat, drink, or take medicine. Report new symptoms during follow-up rather than waiting for the next scan.

Seek emergency care now for:

These symptoms may indicate major bleeding, obstruction, perforation, or another urgent problem.

- Vomiting blood or passing a large amount of blood
- Fainting, severe dizziness, confusion, or clammy skin
- Sudden or severe abdominal pain
- A rigid or very swollen abdomen
- Repeated vomiting with inability to pass stool or gas
- Severe shortness of breath or chest pain

## Common questions

Is a gastrointestinal stromal tumor cancer?

A GIST can behave as a cancer by invading nearby tissue, returning, or spreading. Some small GISTs remain slow-growing. Pathology and imaging are needed to estimate likely behavior.

Can a GIST be completely cured?

Some localized GISTs are cured by surgery. The likelihood depends on size, location, mitotic rate, rupture status, and whether the tumor was completely removed. A GIST that has spread is less likely to be cured, but targeted medicines may control it for a long time.

Why does the mutation result matter?

The mutation can predict whether a targeted medicine is likely to block the tumor's growth signal. Testing helps avoid medicines unlikely to work and identifies better-matched options.

Will I need treatment after surgery?

Not everyone does. People with a very low recurrence risk may have scans alone. Targeted treatment may be recommended for higher-risk disease, provided the mutation is sensitive to that medicine.

Can a GIST come back years later?

Yes. Recurrence can happen months or years after surgery, although risk varies greatly. This is why follow-up may continue after treatment ends. Report new symptoms to your team.

Is GIST inherited by my children?

Usually not. Most mutations occur only in tumor cells. A small minority of GISTs are associated with inherited conditions. Multiple tumors, young age at diagnosis, or particular tumor features may prompt genetic counseling.

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