

Gastrointestinal stromal tumors (GISTs) belong to a group of cancers known as soft tissue sarcomas. The number of new cases in the United States annually has been estimated to be 5,000-6,000. Tumors usually arise from the intestinal tract with the most common site being the stomach, followed by the small intestine, and then the colon/rectum with rare cases arising in the esophagus. There are also tumors that appear to arise in the membranous tissue lining the wall of the stomach (peritoneum) or in a fold of such membranous tissue (the omentum). There are also case reports of tumors arising in the appendix and/or pancreas. These tumors most commonly present with abdominal pain, bleeding or signs of intestinal obstruction. They spread most commonly to sites within the abdominal cavity and to the liver, although there are rare cases of spread to the lungs and bone. Some GISTs are noncancerous (benign) and do not spread (indolent); others are aggressive with extensive local invasion as well as distant metastases. Most cases result from a change (mutation) in one of two genes, KIT or PDGFR, which leads to continued growth and division of tumor cells. There are a few reported cases of families in which a gene mutation is inherited; however, the majority of tumors occur randomly for no apparent reason (sporadically) and not inherited (acquired mutation). Most cases arise in older adults. Leiomyosarcomas of the intestinal tract arise in a similar location and can be confused with GIST. Leiomyosarcoma is a malignant (cancerous) tumor that arises from smooth muscle cells. There are essentially two types of muscles in the body – voluntary and involuntary. Smooth muscles are involuntary muscles – the brain has no conscious control over them. Smooth muscles react involuntarily in response to various stimuli. For example, smooth muscle that lines the walls of the digestive tract causes wave-like contractions (peristalsis) that aid in the digestion and transport of food. Smooth muscles in the salivary glands cause the glands to squirt saliva into the mouth in response to taking a bite of food. Smooth muscle in the skin causes goose bumps to form in response to cold. Leiomyosarcoma usually spreads via the bloodstream. It is very rare to see it in lymphatics. Since smooth muscle is found all over the body, a leiomyosarcoma can form almost anywhere where there are blood vessels, heart, liver, pancreas, genitourinary and gastrointestinal tract, the space behind the abdominal cavity (retroperitoneum), uterus, skin. The uterus is the most common location for a leiomyosarcoma. Most leiomyosarcomas of the gastrointestinal tract are now reclassified as GISTs. The exact cause of leiomyosarcoma, including uterine leiomyosarcoma, is unknown. (For more information on these tumors, choose “leiomyosarcoma” as your search term in the Rare Disease Database.)