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**Leiomyosarcoma**

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# Disease Overview

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 wavelike 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 is a form of cancer. The term “cancer” refers to a group of diseases characterized by abnormal, uncontrolled cellular growth that invades surrounding tissues and may spread (metastasize) to distant body sites via the bloodstream, the lymphatic system, or other means. Different forms of cancer, including leiomyosarcomas, may be classified based upon the cell type involved, the specific nature of the malignancy, and the disease’s clinical course. Leiomyosa 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 gastrointestinal stromal tumors (GIST – see below)

Leiomyosarcoma is classified as a soft tissue sarcoma. Sarcomas are malignant tumors that arise from the connective tissue, which connects, supports and surrounds various structures and organs in the body. Soft tissue includes fat, muscle, nerves, tendons, and blood and lymph vessels. The exact cause of leiomyosarcoma, including uterine leiomyosarcoma, is unknown.

# Signs & Symptoms

The symptoms of a leiomyosarcoma vary depending upon the exact location, size, and spread of the tumor. A leiomyosarcoma, especially in the early stages, may not be associated with any obvious symptoms (asymptomatic). General symptoms associated with cancer may occur including fatigue, fever, weight loss, a general feeling of ill health (malaise), and nausea and vomiting.

Pain may occur in the affected area but is uncommon. Swelling usually occurs and a mass is commonly detected. Additional symptoms are specific to the exact location of the tumor. The tumors may cause bleeding in the gastrointestinal tract and cause black, tarry, foulsmelling stools (melena), or vomiting of blood (hematemesis) or abdominal discomforts). A leiomyosarcoma of the uterus may cause abnormal bleeding from the uterus into and the vagina, with abnormal vaginal discharge, and a change in bladder or bowel habits.

Most forms of leiomyosarcoma are aggressive tumors that may spread (metastasize) to other areas of the body such as the lungs or liver, potentially causing life-threatening complications. Leiomyosarcoma has a high risk of recurring after treatment, if not diagnosed early.

# Causes

The exact cause of leiomyosarcoma is unknown. Researchers speculate that genetic factors may play a contributing role in causing LMS.

In individuals with cancer, including leiomyosarcoma, malignancies may develop due to abnormal changes in the structure and orientation of certain cells known as oncogenes or tumor suppressor genes. Oncogenes control cell growth; tumor suppressor genes control cell division and ensure that cells die at the proper time. The specific cause of changes to these genes is unknown. However, current research suggests that abnormalities of DNA (deoxyribonucleic acid), which is the carrier of the body’s genetic code, are the underlying basis of cellular malignant transformation. These abnormal genetic changes may occur spontaneously for unknown reasons or, more rarely, may be inherited.

# Affected populations

Leiomyosarcomas affect both males and females. Leiomyosarcoma is a form of soft tissue sarcoma. According to the American Cancer Society, at least 15,000 new cases of soft tissue sarcoma occur each year in the U.S. Soft tissue sarcomas affect men and women equally and occur more often in adults than children or adolescents. Soft tissue sarcomas account for 1 percent of all adult cancers in the U.S. According to one estimate, leiomyosarcomas account for 7-11 percent of all cases of soft tissue sarcomas.

# Disorders with Similar Symptoms

Symptoms of the following disorders can be similar to those of a leiomyosarcoma. Comparisons may be useful for a differential diagnosis.

Leiomyomas are benign tumors arising from the smooth muscle. The uterus and the gastrointestinal tract are the most common locations for this tumor. In some cases, multiple leiomyomas form. Leiomyomas often do not cause symptoms (asymptomatic). In some cases they may be associated with pain or swelling in the affected area. Leiomyomas may grow large enough to compress nearby structures causing a variety of symptoms and necessitating surgical removal. In extremely rare cases, leiomyomas may progress to become cancerous (malignant transformation). The exact cause of leiomyomas is unknown. (For more information on this condition, choose “leiomyoma” as your search term in the Rare Disease Database.)

Gastrointestinal stromal tumors (GIST) belong to a group of cancers known as soft tissue sarcomas. Tumors usually arise from the intestinal tract with the most common site being the stomach, followed by the small intestine, and 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. GIST results from a change 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 are sporadic and not inherited. Treatment is with surgery. In individuals in whom the disease has spread, treatment may include surgery when possible and the administration of imatinib mesylate (Gleevec, Glivec), a tyrosine kinase inhibitor that inhibits the KIT or PDGFR responsible for tumor growth. Imatinib mesylate can delay or prevent recurrence of GIST when given after the tumor has been removed (resection). The role of imatinib mesylate in pediatric GIST is being studied at this time. (For more information on this disorder, choose “GIST” as your search term in the Rare Disease Database.)

# Diagnosis

A diagnosis of a leiomyosarcoma may be made based upon a detailed patient history, a thorough clinical evaluation and a variety of tests including blood tests, surgical removal and microscopic examination of tissue (biopsies) and various imaging techniques. In some cases, individuals may notice a painful lump or mass in the affected area. A key diagnostic aspect is distinguishing malignant leiomyosarcoma from its benign counterpart, the leiomyoma.

To confirm a diagnosis of leiomyosarcoma a fine-needle aspiration may be performed. Fine-need aspiration (FNA) is a diagnostic technique in which a thin, hollow needle is passed though the skin and inserted into the nodule or mass to withdraw small samples of tissue. The collected tissue is then studied under a microscope. In some cases, FNA may prove inconclusive and physicians may perform a core (Trucut) biopsy or an incisional biopsy. During Trucut or incisional biopsy, a small sample tissue is surgically removed and sent to a pathology laboratory where it is processed and studied to determine its microscopic structure and makeup (histopathology).

Specialized imaging techniques may be used to help evaluate the size, placement, and extension of the tumor and to serve as an aid for future surgical procedures, among individuals with leiomyosarcomas. Such imaging techniques may include computerized tomography (CT) scanning, magnetic resonance imaging (MRI) and ultrasound. During CT scanning, a computer and x-rays are used to create a film showing cross-sectional images of certain tissue structures. An MRI uses a magnetic field and radio waves to produce cross-sectional images of particular organs and bodily tissues. During an ultrasound reflected sound waves create an image of internal organs and other structures within the body.

Laboratory tests and specialized imaging tests may also be conducted to determine possible infiltration of regional lymph nodes and the presence of distant metastases.

# Standard Therapies

## Treatment

The therapeutic management of individuals with a leiomyosarcoma may require the coordinated efforts of a team of medical professionals, such as physicians who specialize in the diagnosis and treatment of cancer (medical oncologists), specialists in the use of radiation to treat cancer (radiation oncologists), surgeons (surgical oncologist), oncology nurses, and other specialists.

Specific therapeutic procedures and interventions may vary, depending upon numerous factors, such as primary tumor location, extent of the primary tumor (stage), and degree of malignancy (grade); whether the tumor has spread to distant sites; an individual’s age and general health; and/or other elements. Decisions concerning the use of particular interventions should be made by physicians and other members of the health care team in careful consultation with the patient, based upon the specifics of the case; a thorough discussion of the potential benefits and risks; patient preference; and other appropriate factors.

The main form of treatment for leiomyosarcomas is surgical excision and removal of the entire tumor and surrounding tissue (resection). Depending upon the location of the primary tumor, surgical procedures may also include the use of certain reconstructive techniques. Surgical options are dictated by the size, location and spread of a tumor.

In addition, based upon primary tumor site, size, and other factors, recommended standard therapy may often include postoperative radiation to help treat known or possible residual disease. If initial surgery is not an option due to the specific location and/or progression of the malignancy, therapy may include radiation alone. Radiation therapy preferentially destroys or injures rapidly dividing cells, primarily cancerous cells. However, some healthy cells (e.g., hair follicles, bone marrow, etc.) may also be damaged, leading to certain side effects. Thus, during such therapy, the radiation is passed through diseased tissue in carefully calculated dosages to destroy cancer cells while minimizing exposure and damage to normal cells. Radiation therapy works to destroy cancer cells by depositing energy that damages their genetic material, preventing or slowing their growth and replication.

For some affected individuals, particularly those who have locally advanced, metastatic, or recurrent disease, therapy with certain anticancer drugs (chemotherapy) may also be recommended, possibly in combination with surgical procedures and/or radiation; physicians may recommend combination therapy with multiple chemotherapeutic drugs that have different modes of action in destroying tumor cells and/or preventing them from multiplying.

In most cases, however, chemotherapy and radiation therapy have had only limited success in slowing or stopping progression of leiomyosarcomas. Because of the rarity of leiomyosarcomas no standard or overall effective type of chemotherapy or radiation therapy has been identified. The use of chemotherapy and radiation therapy for the treatment of leiomyosarcomas remains under investigation. (Please see the “Investigational Therapies” section below.)

In 2015, Yondelis was approved as a chemotherapy for liposarcoma and leiomyosarcoma that cannot be removed by surgery or patients that previously underwent treatment that contained anthracycline. Yondelis is marketed by Janssen Products.

# Clinical Trials and Studies

Medical centers and hospitals that specialize in sarcomas (sarcoma centers) are conducting research into new treatments for individuals with soft tissue sarcomas including new chemotherapeutic drugs, new combinations of chemotherapeutic drugs, angiogenesis inhibitors that prevent the formation of new blood vessels needed to supply blood tumors, and various biologic therapies that involve the immune system in fighting cancer. (For information on sarcoma centers, contact the sarcoma specific nonprofit organizations listed in the Resources section of this report.)

Information on current clinical trials is posted on the Internet at www.clinicaltrials.gov. All studies receiving U.S. government funding, and some supported by private industry, are posted on this government web site.

For information about clinical trials being conducted at the NIH Clinical Center in Bethesda, MD, contact the NIH Patient Recruitment Office:

Tollfree: (800) 411-1222

TTY: (866) 411-1010

Email: prpl@cc.nih.gov

For information about clinical trials sponsored by private sources, contact: www.centerwatch.com

# References

TEXTBOOKS

De Vita Jr Vt, Hellman S, Rosenburg SA. Eds. Cancer: Principles and

Practice on Oncology. 5th ed. Philadelphia, PA: J.B. Lippincott Company; 1997:1748-9.

INTERNET

Martinez GE, Brasinikas G. Intestinal Leiomyosarcoma. Emedicine. https://emedicine.medscape.com/article/179839-overview. Updated January 7, 2010. Accessed August 29, 2012.

Sarcoma – Adult Soft Tissue Cancer. American Cancer Society. https://www.cancer.org/Cancer/Sarcoma-

AdultSoftTissueCancer/index. Accessed August 29, 2012.

Boston Children’s Hospital. Leiomyosarcoma.

https://www.childrenshospital.org/az/Site1098/mainpageS1098P0.html.

Accessed August 29, 2012.

# Programs & Resources RareCare® Assistance Programs

NORD strives to open new assistance programs as funding allows. If we don’t have a program for you now, please continue to check back with us.

# Additional Assistance Programs

|  |
| --- |
| **Rare Disease Educational Support Program**  Ensuring that patients and caregivers are armed with the tools they need to live their best lives while managing their rare condition is a vital part of NORD’s mission.  [https://rarediseases.org/patient-assistance-programs/raredisease-educational-support/](https://rarediseases.org/patient-assistance-programs/rare-disease-educational-support/) |
| **Rare Caregiver Respite Program**  This first-of-its-kind assistance program is designed for caregivers of a child or adult diagnosed with a rare disorder.  [https://rarediseases.org/patient-assistance-programs/caregiverrespite/](https://rarediseases.org/patient-assistance-programs/caregiver-respite/) |

# Patient Organizations

[**National Leiomyosarcoma Foundation**](https://rarediseases.org/organizations/national-leiomyosarcoma-foundation/)

|  |
| --- |
| *NORD Member*  Phone: 303-783-0924 Email: annieachee@aol.com  [https://rarediseases.org/organizations/national-leiomyosarcomafoundation/](https://rarediseases.org/organizations/national-leiomyosarcoma-foundation/) |
| [**American Cancer Society, Inc.**](https://rarediseases.org/organizations/american-cancer-society-inc/) Phone: 404-320-3333  [https://rarediseases.org/organizations/american-cancer-societyinc/](https://rarediseases.org/organizations/american-cancer-society-inc/) |
| [**National Cancer Institute**](https://rarediseases.org/organizations/national-cancer-institute/)  Phone: 301-435-3848 Email: cancergovstaff@mail.nih.gov  <https://rarediseases.org/organizations/national-cancer-institute/> |
| [**OncoLink: The University of Pennsylvania Cancer Center Resource**](https://rarediseases.org/organizations/oncolink-the-university-of-pennsylvania-cancer-center-resource/)  Email: hampshire@uphs.upenn.edu  [https://rarediseases.org/organizations/oncolink-the-university-ofpennsylvania-cancer-center-resource/](https://rarediseases.org/organizations/oncolink-the-university-of-pennsylvania-cancer-center-resource/) |
| [**Sarcoma Alliance**](https://rarediseases.org/organizations/sarcoma-alliance/)  Phone: 415-381-7236 Email: info@sarcomaalliance.org  Fax: 415-381-7235  <https://rarediseases.org/organizations/sarcoma-alliance/> |
| [**Rare Cancer Alliance** https://rarediseases.org/organizations/rare-cancer-alliance/](https://rarediseases.org/organizations/rare-cancer-alliance/) |
| [**BeatSarcoma**](https://rarediseases.org/organizations/beatsarcoma/)  Phone: 415-826-0474 Email: info@beatsarcoma.org  <https://rarediseases.org/organizations/beatsarcoma/> |
| [**Northwest Sarcoma Foundation**](https://rarediseases.org/organizations/northwest-sarcoma-foundation/)  Phone: 503-954-5740 Email: melissa@nwsarcoma.org  [https://rarediseases.org/organizations/northwest-sarcomafoundation/](https://rarediseases.org/organizations/northwest-sarcoma-foundation/) |
| [**Sarcoma Foundation of America**](https://rarediseases.org/organizations/sarcoma-foundation-of-america/) Email: info@curesarcoma.org [https://rarediseases.org/organizations/sarcoma-foundation-ofamerica/](https://rarediseases.org/organizations/sarcoma-foundation-of-america/) |
| [**Leiomyosarcoma Support & Direct Research Foundation**](https://rarediseases.org/organizations/leiomyosarcoma-support-direct-research-foundation/) Email: info@LMSDR.org  [https://rarediseases.org/organizations/leiomyosarcoma-supportdirect-research-foundation/](https://rarediseases.org/organizations/leiomyosarcoma-support-direct-research-foundation/) |

# More Information

*The information provided on this page is for informational purposes only. The National Organization for Rare Disorders (NORD) does not endorse the information presented. The content has been gathered in partnership with the MONDO Disease Ontology. Please consult with a healthcare professional for medical advice and treatment.*

## GARD Disease Summary

The Genetic and Rare Diseases Information Center (GARD) has information and resources for patients, caregivers, and families that may be helpful before and after diagnosis of this condition. GARD is a program of the National Center for Advancing Translational Sciences (NCATS), part of the National Institutes of Health (NIH).

[**View report**](https://rarediseases.info.nih.gov/diseases/6880/x)

## Orphanet

Orphanet has a summary about this condition that may include information on the diagnosis, care, and treatment as well as other resources. Some of the information and resources are available in languages other than English. The summary may include medical terms, so we encourage you to share and discuss this information with your doctor. Orphanet is the French National Institute for Health and

Medical Research and the Health Programme of the European Union.

[**View report**](https://www.orpha.net/en/disease/detail/64720)