CONDITION

Extragonadal germ cell tumors

Extragonadal germ cell tumors are growths from germ cells that develop outside the ovaries or testicles, most often in the chest, abdomen, or brain.

WHAT IT IS
Germ cell tumors arising outside the ovaries or testicles

BODY SYSTEM
Reproductive cells, often affecting the chest, abdomen, or brain

MAIN SYMPTOMS
Lump, chest pressure, headaches, vomiting

WHAT CAUSES IT
Misplaced early germ cells; exact trigger unknown

HOW IT IS DIAGNOSED
Imaging, tumor-marker tests, and usually biopsy

MAIN TREATMENT
Chemotherapy, surgery, and sometimes radiation therapy

CAN IT BE CURED?
Sometimes - type, site, and spread shape curability

IS IT AN EMERGENCY?
Sometimes - breathing trouble, seizures, or brain-pressure symptoms

LONG-TERM OUTLOOK
Often curable, but mediastinal nonseminoma is harder to treat

Overview

Extragonadal germ cell tumors begin outside the gonads-the testicles or ovaries. Germ cells normally develop into sperm or eggs. These tumors often form along the center of the body, where germ cells travel during early development.

Common locations include the mediastinum, the space between the lungs; the retroperitoneum, at the back of the abdomen; and the brain. In babies and young children, tumors can also develop near the tailbone.

Location and tumor type vary with age. Chest tumors are more common in adolescent and young adult males. Brain germ cell tumors often occur near the pineal or pituitary gland.

Some are seminomas, called germinomas in the brain, which often respond well to treatment.

Nonseminomatous tumors include yolk sac tumors, embryonal carcinoma, choriocarcinoma, and mixed germ cell tumors. Teratomas contain tissues resembling other body tissues and may be mature, immature, or mixed with a malignant tumor.

Identifying the exact type is important because treatment and outlook differ. Doctors also examine the ovaries or testicles because a tumor found elsewhere can sometimes be spread from a small or unnoticed gonadal tumor.

Symptoms

Symptoms depend on the tumor's location and size. Small tumors may be found during imaging for another reason.

A chest tumor may cause:

Facial or neck swelling may occur when a mass presses on the large vein returning blood from the upper body to the heart. This is called superior vena cava obstruction.

A tumor in the back of the abdomen may cause abdominal or lower-back pain, fullness, constipation, or a lump. Pressure on the kidneys or urinary tubes may affect urine flow.

A brain tumor can cause headaches, nausea, vomiting, balance problems, double vision, unusual sleepiness, or seizures. Tumors near the pituitary gland may cause extreme thirst and frequent urination, altered growth or puberty, menstrual changes, or reduced sexual function.

A tumor near the tailbone may appear as a lower-back or buttock mass and may affect bowel or bladder control.

General symptoms include tiredness, reduced appetite, fever, or unplanned weight loss. These have many possible causes and do not necessarily mean that a tumor is present.

- Persistent cough or shortness of breath
- Chest pain or pressure
- Trouble lying flat
- Swelling of the face, neck, or arms
- Prominent veins over the upper chest

Causes and risk factors

The exact cause is usually unknown. The leading explanation is that some early germ cells remain outside the ovaries or testicles during embryonic development and later form a tumor.

Extragonadal germ cell tumors are not usually caused by anything a person did, and they are not contagious.

Primary mediastinal nonseminomatous germ cell tumors are associated with Klinefelter syndrome, a chromosomal condition in which a male has an extra X chromosome. Most people with Klinefelter syndrome do not develop this tumor, and many affected people do not have the syndrome.

Age and sex are associated with certain patterns but are not direct causes. Most tumors are not passed from parent to child. Genetic assessment may be offered if a chromosomal or inherited condition is suspected.

Diagnosis

Diagnosis usually involves a medical history, physical examination, imaging, tumor-marker tests, and a biopsy. CT or MRI shows the tumor's location, size, and relationship to nearby structures. Additional scans may check whether it has spread.

Blood tests may measure:

AFP is not produced by a pure seminoma or germinoma, so a raised level suggests nonseminomatous cells. Beta-hCG may rise in several tumor types. LDH is less specific.

A biopsy removes tissue for examination under a microscope. It identifies the tumor type and helps rule out conditions such as lymphoma. In selected situations, characteristic imaging and tumor-marker results may allow treatment planning without an initial surgical biopsy.

For a suspected brain tumor, AFP and beta-hCG may be tested in blood and cerebrospinal fluid. MRI is central to assessment, although surgery or biopsy may be needed if findings are inconclusive.

Other tests may include:

Tumor type, location, marker levels, and spread guide treatment and help estimate outlook.

- Alpha-fetoprotein, or AFP
- Beta-human chorionic gonadotropin, or beta-hCG
- Lactate dehydrogenase, or LDH
- Ultrasound of the testicles or imaging of the ovaries
- Blood counts and kidney and liver tests
- Imaging of the chest, abdomen, pelvis, or brain

- Hearing and heart assessment before certain treatments
- Fertility and hormone assessment when relevant

Treatment and management

Treatment should be planned by a specialist team familiar with germ cell tumors. The approach depends on whether the tumor is a seminoma or germinoma, nonseminomatous tumor, teratoma, or mixed tumor.

Chemotherapy is a main treatment for many malignant tumors. Those outside the brain are often treated with cisplatin-based combinations. The medicines and number of cycles depend on tumor type, location, spread, general health, and organ function.

Surgery may remove a localized tumor or tissue remaining after chemotherapy. It is particularly important for mature teratomas, which do not reliably respond to chemotherapy. A residual mass may contain teratoma, active tumor, or scar tissue, so specialists assess whether surgery, imaging, or observation is appropriate.

Seminomas and brain germinomas are usually sensitive to chemotherapy and radiation therapy. Brain tumors require a central nervous system-specific plan, sometimes combining chemotherapy with targeted radiation. Surgery is selective because these tumors may be close to structures controlling vision, hormones, movement, and memory.

Scans and tumor-marker tests assess the response. A mass that remains visible is not always active cancer.

Treatment may affect fertility. Before chemotherapy, radiation, or major surgery, adolescents and adults should discuss sperm, egg, embryo, or ovarian-tissue preservation when time and circumstances allow. Hormone replacement may be needed if the tumor or treatment affects the pituitary gland.

Complications

A tumor may press on the lungs, major blood vessels, brain, spinal fluid pathways, kidneys, bowel, or bladder.

Brain tumors can block cerebrospinal fluid and cause hydrocephalus, raising pressure inside the skull. Signs include worsening headache, vomiting, drowsiness, and vision changes. Tumors near the pituitary may cause lasting hormone deficiencies.

Treatment effects can include infection, low blood counts, nausea, fatigue, hearing changes,

kidney problems, numbness, lung injury, and reduced fertility. Monitoring helps identify complications early.

Some mediastinal nonseminomatous tumors are associated with blood disorders or other cancerous cell types. A germ cell tumor may also return after treatment, making follow-up important.

Prevention

There is no established way to prevent an extragonadal germ cell tumor. It is thought to arise from events during early development rather than diet, exercise, sexual activity, or another avoidable behavior.

People with Klinefelter syndrome should receive usual care for that condition. Having the syndrome does not mean that a tumor will develop.

Living with it and what to expect

Treatment may last several months and involve chemotherapy visits, hospital stays, surgery, or radiation. Fatigue and appetite or activity changes are common.

Many seminomas, germinomas, and other germ cell tumors respond strongly to treatment, and some can be cured after spreading. Primary mediastinal nonseminomatous tumors generally have a less favorable outlook and often require care at an experienced center.

Follow-up commonly includes examinations, tumor-marker blood tests, and imaging. Visits are usually more frequent soon after treatment and become less frequent over time. The schedule depends on tumor type, treatment, and recurrence risk.

Long-term care may monitor hearing, kidney and lung function, cardiovascular health, fertility, hormone levels, and emotional well-being. People treated for brain tumors may need help with school, work, memory, vision, or hormone replacement.

Some people return to previous routines, while others need rehabilitation, medication, or adjustments at work or school. Follow-up remains important even when treatment has ended and you feel well.

When to get medical help

Arrange a medical appointment for a persistent lump, unexplained chest or abdominal

pressure, recurring headaches, ongoing vomiting, unusual thirst and urination, or symptoms that continue to worsen.

Seek urgent medical help for:

Call emergency services for severe breathing difficulty, fainting, a first seizure, a seizure lasting several minutes, or difficulty waking.

During chemotherapy, fever can indicate a serious infection when white blood cell levels are low. Follow the treatment team's emergency instructions and contact the cancer service immediately rather than waiting for a routine appointment.

- New facial, neck, or arm swelling with chest symptoms
- A severe or rapidly worsening headache with vomiting
- New double vision, weakness, confusion, or marked drowsiness
- Difficulty passing urine or sudden loss of bowel or bladder control

Common questions

Is an extragonadal germ cell tumor the same as testicular or ovarian cancer?

Not exactly. It arises from the same general cell family but is found outside the ovaries or testicles. Doctors examine the gonads to make sure the tumor did not begin there and spread.

Are all extragonadal germ cell tumors cancerous?

No. A mature teratoma may be noncancerous, although it can grow and press on nearby organs. Seminomas, germinomas, yolk sac tumors, embryonal carcinomas, choriocarcinomas, and many mixed tumors are malignant.

Why can a germ cell tumor grow in the chest or brain?

Germ cells move through the developing body before birth. Some may remain along this pathway and later form a tumor. The trigger is usually unknown.

Can an extragonadal germ cell tumor be cured?

Sometimes. Seminomas and germinomas are often highly responsive to treatment. Outlook also depends on location, tumor type, marker levels, spread, treatment response, and whether remaining tumor can be removed. Mediastinal nonseminomatous tumors are generally harder to cure.

Can the tumor come back after treatment?

Yes. Recurrence is possible, particularly during the earlier years after treatment. Regular examinations, scans, and tumor-marker tests help detect it.

Will treatment affect my ability to have children?

It can. Chemotherapy, radiation, surgery, hormone problems, and the tumor itself may reduce fertility. Discuss fertility preservation before treatment when possible, although options may also be available afterward.

Sources

- National Cancer Institute. Childhood Extracranial Germ Cell Tumors Treatment (PDQ®)-Health Professional Version .
- National Cancer Institute. Childhood Central Nervous System Germ Cell Tumors Treatment (PDQ®)-Health Professional Version .
- European Association of Urology. EAU Guidelines on Testicular Cancer: Disease Management .
- Honecker F, Aparicio J, Berney D, et al. "ESMO Consensus Conference on testicular germ cell cancer: diagnosis, treatment and follow-up." Annals of Oncology . 2018;29(8):1658-1686.
- Murray MJ, Bartels U, Nishikawa R, Fangusaro J, Matsutani M, Nicholson JC. "Consensus on the management of intracranial germ-cell tumours." Neuro-Oncology . 2015;17(7):888-894.
- Albany C, Einhorn LH. "Extragonadal germ cell tumors: clinical presentation and management." Current Opinion in Oncology . 2013;25(3):261-265.