CONDITION

Familial chordoma

Familial chordoma is an inherited tendency to develop rare tumors along the skull base or spine.

WHAT IT IS
Inherited tendency to develop chordoma tumors

BODY SYSTEM
Bones, spine, skull base, and nervous system

MAIN SYMPTOMS
Persistent pain, headaches, double vision, bowel or bladder changes

WHAT CAUSES IT
Inherited genetic changes, sometimes unknown

HOW IT IS DIAGNOSED
MRI or CT, biopsy, and genetic testing

MAIN TREATMENT
Specialist surgery and high-dose radiation

CAN IT BE CURED?
Sometimes - localized tumors may be eradicated

IS IT AN EMERGENCY?
Sometimes - new spinal cord compression symptoms

LONG-TERM OUTLOOK
Recurrence risk requires lifelong specialist monitoring

Overview

Familial chordoma is an inherited tendency to develop chordoma, a rare cancerous bone tumor. Chordomas grow from remnants of the notochord, a structure that helps form the spine before birth and normally disappears during development.

Notochord remnants may remain near the skull base, within the mobile spine, or in the sacrum at the bottom of the spine. A chordoma can arise from them years later.

"Familial" means chordoma has occurred in more than one blood relative and may be linked to an inherited genetic change. In some families, the cause is an extra copy, or duplication, of the TBXT gene, which helps direct notochord development. Other families have no identifiable TBXT change.

Chordomas usually grow slowly, but they can invade bone and soft tissue or press on nerves, the brainstem, spinal cord, bowel, or bladder. Their location may make complete removal difficult, and they can return after treatment.

A family history does not mean every relative will develop a chordoma. Genetic counseling can help clarify risk and whether testing or monitoring may be useful.

Symptoms

Symptoms depend on the tumor's location and often develop gradually. They can resemble common conditions, and having one does not necessarily mean you have chordoma.

A skull-base chordoma may cause:

A chordoma in the neck or middle spine may cause pain, tingling, numbness, or weakness that can spread into an arm or leg. Spinal cord pressure may affect balance, walking, or bowel and bladder control.

A sacral chordoma may cause persistent lower-back, buttock, or tailbone pain, as well as leg symptoms, constipation, difficulty passing urine, or changes in sexual function. Diagnosis can be delayed because these symptoms are common and the tumor may be deep within the pelvis.

Some tumors are found before symptoms develop, either during imaging for another reason or through monitoring arranged because of a family history.

- Persistent headaches or pain behind the eyes
- Double vision or other eyesight changes
- Facial numbness or weakness
- Difficulty swallowing, speaking, or hearing

Causes and risk factors

Familial chordoma results from an inherited susceptibility. It is not caused by injury, diet, activity, infection, or anything a person did.

Some affected families have a germline duplication of TBXT , meaning the change is present from birth throughout the body. TBXT provides instructions for making brachyury, a protein involved in notochord development and commonly found at high levels in chordoma cells.

A familial TBXT duplication can be inherited in an autosomal dominant pattern. Each child of

someone carrying the duplication has a 1 in 2 chance of inheriting it. However, inheriting the change does not guarantee that chordoma will develop, and the likely age of onset cannot be predicted precisely.

Some families have several affected relatives but no currently recognized genetic cause. Research into other inherited factors continues.

A chordoma in someone without an affected relative is usually considered sporadic. Certain common TBXT variants may influence risk, but they differ from the rare duplications found in some families.

Diagnosis

MRI is used to show soft tissue, the brain, and the spinal canal, while CT can show bone damage and assist surgical planning. Imaging also helps define the tumor's relationship to nerves, blood vessels, and other structures.

A biopsy is usually required to confirm chordoma. A pathologist examines a small tissue sample, sometimes testing it for brachyury and other markers. Because chordoma can resemble other tumors, review by a pathologist experienced in bone and soft-tissue tumors is valuable.

Biopsy must be planned by the specialist team that would provide definitive treatment. The route used matters because tumor cells can sometimes remain along the biopsy path and affect later surgery.

Genetic testing serves a different purpose. A blood or saliva test may identify an inherited TBXT duplication when family history suggests familial chordoma, but it cannot show whether a tumor is present.

Before testing, a genetics professional should explain what positive, negative, or uncertain results could mean for you and your relatives.

Treatment and management

Treatment is planned by a specialist team experienced in chordoma. This may include surgeons, radiation and medical oncologists, radiologists, pathologists, rehabilitation specialists, and genetics professionals.

Surgery is often the main treatment for localized chordoma. When possible, the goal is to remove the tumor in one piece with a rim of healthy tissue, known as wide or negative-margin

removal. This may reduce local recurrence.

Complete removal is not always safe. Skull-base tumors may be close to the brainstem, cranial nerves, or major blood vessels. Spinal or sacral surgery may affect strength, sensation, spinal stability, or bowel and bladder control. The team should explain the expected benefits and possible long-term effects.

Chordoma is less sensitive to standard radiation doses than many cancers. High-dose, precisely targeted radiation may be given after surgery, before surgery in selected cases, or when surgery is not possible. Proton-beam therapy is often considered because it can limit radiation exposure to nearby tissue. Other advanced techniques may also be suitable.

Standard chemotherapy generally has limited activity against chordoma. Targeted medicines or clinical trials may be considered for tumors that cannot be removed, have returned, or have spread. Choices depend on the tumor, previous treatment, overall health, and available evidence.

Supportive care may include pain control, rehabilitation, mobility support, and treatment for bowel, bladder, swallowing, vision, or nerve problems. Regular MRI or CT scans are essential because chordoma can return years later.

Complications

The main complication is local recurrence, in which the tumor returns at or near its original site. This can occur even after apparently complete treatment. Further treatment may be more difficult because of scar tissue, previous radiation, and nearby vital structures.

Pressure or direct tumor growth can cause chronic pain, weakness, loss of sensation, difficulty walking, double vision, swallowing problems, or changes in bowel, bladder, and sexual function.

Chordoma can also spread to distant sites, including the lungs, bones, liver, and lymph nodes. Local growth is often the more immediate concern, but follow-up may assess both recurrence and spread.

Treatment can cause lasting effects such as reduced mobility, nerve damage, hearing or vision problems, changes in appearance, hormone problems after skull-base treatment, or bowel and bladder difficulties. Rehabilitation and symptom-focused care can help maintain independence and quality of life.

Prevention

An inherited susceptibility to chordoma cannot currently be prevented. Lifestyle changes cannot remove a TBXT duplication or guarantee that a tumor will not develop.

When a familial genetic change is known, relatives may choose genetic counseling and testing. Identifying a carrier may allow monitoring before symptoms appear. There is no universally accepted screening schedule, so a genetics and chordoma team should tailor the imaging type and timing.

Reproductive genetic counseling can explain options for people concerned about passing a known change to a child. These are personal decisions, and counseling should be non-directive.

Living with it and what to expect

Familial chordoma can affect families over many years. Some people who inherit a susceptibility never develop a known tumor, while affected relatives may be diagnosed at different ages or tumor locations.

Recovery after treatment may take months. Skull-base or spinal surgery can require rehabilitation for strength, balance, speech, swallowing, or daily activities. Some effects improve with healing and therapy, while others may persist.

Follow-up usually continues for life because recurrence can occur after a long interval. It may include symptom reviews, examinations, and imaging of the treated area. Other scans may be used if there is concern about spread. The schedule depends on tumor location, treatment, and previous results.

Many people return to work, education, family responsibilities, and valued activities, sometimes with adjustments. Pain management, rehabilitation, mobility aids, workplace changes, and psychological support can help.

Uncertainty before scans and concern about relatives are common. Genetic counselors can help with family communication and testing decisions, while specialist nurses, counselors, and peer groups can provide emotional support.

When to get medical help

Arrange a routine appointment if you have a family history of chordoma and want genetic counseling. Seek assessment for persistent or worsening pain, headaches, double vision, numbness, weakness, swallowing problems, or unexplained bowel or bladder changes.

If you have been treated for chordoma, contact your specialist team about new symptoms,

returning symptoms, or changes between follow-up visits. Do not wait for your next scan if symptoms are progressing.

Seek urgent medical care for:

These symptoms may indicate significant pressure on the spinal cord, lower spinal nerves, brainstem, or airway and require emergency assessment.

- New or rapidly worsening weakness in an arm or leg
- New numbness around the genitals, buttocks, or inner thighs
- Sudden loss of bladder or bowel control
- Inability to pass urine with back pain or leg symptoms
- New severe difficulty swallowing or breathing

Common questions

Is familial chordoma cancer?

Chordoma is a malignant, or cancerous, bone tumor. It may grow slowly, but it can invade nearby tissue, return after treatment, and sometimes spread.

Does everyone with a TBXT duplication get chordoma?

No. A duplication increases susceptibility but does not make chordoma certain. Precise risk estimates are not available for every family.

Can familial chordoma skip a generation?

It can appear to if someone inherits the change but never develops a recognized tumor. Each child of a parent with a familial TBXT duplication has a 1 in 2 chance of inheriting it.

Should my relatives have genetic testing?

Testing may help when a disease-causing familial change is known or several blood relatives have chordoma. Genetic counseling should come first so each person understands the possible results, limitations, and implications for relatives.

Can a chordoma come back after surgery?

Yes. Chordoma can return near its original site, including years after treatment. Complete removal and specialist radiation may reduce but cannot eliminate this risk, so long-term imaging is important.

Is chordoma the same as a slipped disc or chordoma-like benign tumor?

No. A slipped disc affects the cushioning disc between spinal bones and is not a tumor. Benign

notochordal cell tumors arise from similar embryonic tissue but usually behave differently. Specialist imaging and, when needed, pathology distinguish them.

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