CONDITION

Rhabdomyosarcoma

Rhabdomyosarcoma is a rare cancer that develops from immature muscle-forming cells in soft tissue.

WHAT IT IS
Cancer of immature muscle-forming cells

BODY SYSTEM
Muscles and other soft tissues throughout the body

MAIN SYMPTOMS
Growing lump, swelling, pain, unexplained bleeding

WHAT CAUSES IT
Usually unknown; acquired cell changes drive growth

HOW IT IS DIAGNOSED
Biopsy with tissue testing; imaging maps spread

MAIN TREATMENT
Combination chemotherapy with surgery and radiation

CAN IT BE CURED?
Sometimes - especially when localized and treatment-responsive

IS IT AN EMERGENCY?
Sometimes - breathing trouble, severe bleeding, or new weakness

LONG-TERM OUTLOOK
Often curable when localized; spread or recurrence worsens outlook

Overview

Rhabdomyosarcoma is a cancer of soft tissue. It begins in immature cells that would normally develop into skeletal muscle, the type of muscle used to move the body. However, it can develop in places with little or no skeletal muscle, such as the bladder, prostate, vagina, or around the eye.

It most often affects children and teenagers, but adults can also develop it. Common locations include the head and neck, urinary or reproductive organs, arms, legs, trunk, abdomen, and pelvis.

Rhabdomyosarcoma can grow into nearby tissue and may spread through the blood or lymphatic system. The lymphatic system is a network of vessels and glands involved in immune function. When this cancer spreads, it may reach the lungs, bones, bone marrow, or

lymph nodes.

There are several subtypes. Embryonal and alveolar rhabdomyosarcoma are the best known. Laboratory testing can identify cell and gene features that help predict how the cancer may behave. These findings, along with its location, size, spread, and response to treatment, guide care.

Symptoms

Symptoms depend mainly on where the tumour is growing. A lump may be painless at first, so pain is not required for rhabdomyosarcoma to be present.

Possible symptoms include:

These symptoms have many more common causes, including infections, injuries, cysts, and non-cancerous growths. Symptoms alone cannot confirm rhabdomyosarcoma.

A tumour pressing on the brain, spinal cord, airway, or urinary tract can cause more urgent problems. Examples include breathing difficulty, new weakness, loss of bladder control, or inability to pass urine.

- A new or enlarging lump, swelling, or area of firmness
- Pain or reduced movement near a tumour in an arm, leg, or joint
- A swollen or bulging eye, drooping eyelid, or changes in vision
- A blocked nose, repeated nosebleeds, or discharge from one side
- Blood in the urine or difficulty passing urine
- Vaginal bleeding or discharge that is not expected
- Swelling in the scrotum or groin
- Abdominal swelling, constipation, vomiting, or ongoing abdominal pain

Causes and risk factors

The exact cause is usually unknown. Rhabdomyosarcoma develops when a cell acquires changes in its DNA, the instructions that control cell growth. These changes allow the cell to keep multiplying instead of developing and stopping normally.

Most cases are not inherited and are not caused by anything a person or parent did. Diet, exercise, minor injuries, and everyday activities are not known causes.

A small proportion of people have an inherited condition that increases their risk. Examples include Li-Fraumeni syndrome, neurofibromatosis type 1, Beckwith-Wiedemann syndrome, and Costello syndrome. These conditions affect genes involved in growth or protection from cancer.

Having one of these syndromes does not mean rhabdomyosarcoma will develop. Likewise, most people with rhabdomyosarcoma do not have a known inherited syndrome.

A genetics assessment may be offered when the person is very young, has a suggestive personal or family history, or has certain tumour features.

Diagnosis

A physical examination and imaging can show where a mass is located, but a biopsy is needed to confirm rhabdomyosarcoma. A biopsy removes a small sample of tissue so a specialist can examine it under a microscope.

Biopsy planning is important because the position of the biopsy cut may affect later surgery. When possible, assessment by a sarcoma team before a suspicious mass is removed can help preserve treatment options.

The tissue sample is tested to determine the tumour type. Testing may include immunohistochemistry, which uses stains to identify proteins in the cells, and molecular testing for gene changes. For example, some alveolar tumours contain a change involving a gene called `FOXO1`. This result can affect risk assessment and treatment planning.

Imaging is used to measure the tumour and look for spread. Tests may include:

Bone marrow tests, bone imaging, or examination of the fluid around the brain and spinal cord may be needed in particular cases. Not everyone requires every test.

The care team combines the biopsy findings and scans to classify the cancer. Important features include its location, size, subtype, gene findings, lymph-node involvement, whether it has spread, and how much could be removed safely.

- MRI to show the tumour and nearby muscles, nerves, and blood vessels
- CT scans, often including the chest to examine the lungs
- PET-CT or another whole-body scan in selected situations
- Ultrasound for some abdominal, pelvic, or scrotal masses

Treatment and management

Rhabdomyosarcoma treatment usually combines chemotherapy with surgery, radiation therapy, or both. Care is generally coordinated by a specialist sarcoma team because treatment needs to control the cancer while protecting growth, movement, appearance, and organ function where possible.

Chemotherapy uses medicines that travel through the bloodstream. It treats the known tumour and cancer cells that may be elsewhere but are too small to appear on scans. Treatment usually uses several medicines over a period of months. The exact combination and length are based on the cancer's risk group and the treatment protocol being followed.

Surgery aims to remove the tumour with a rim of healthy tissue when this can be done without unacceptable harm. Immediate complete removal is not always possible or appropriate. For example, a tumour near the eye, bladder, major nerves, or blood vessels may first be treated with chemotherapy.

Radiation therapy directs high-energy beams at a defined area. It may treat cancer that could not be removed, cells left behind after surgery, or tumours in locations where surgery would cause major loss of function. Proton therapy may be considered in some centres because it can reduce radiation exposure to nearby tissue, although it is not suitable or available for everyone.

Scans and examinations during treatment show how the tumour is responding. Treatment may be adjusted if serious side effects occur or the cancer does not respond as expected.

If rhabdomyosarcoma returns, options may include further chemotherapy, surgery, radiation, targeted treatment for particular tumour changes, or a clinical trial. Treatment choices depend on where it returned, previous treatment, and the person's overall health.

Supportive care is also important. It may include medicines for nausea or pain, infection treatment, nutrition support, physiotherapy, psychological support, and help with school, work, fertility, or family life.

Complications

The tumour itself can damage nearby structures. Possible complications include blocked urine flow, vision problems, reduced movement, nerve pressure, bleeding, or breathing difficulty. Cancer that has spread can cause bone pain, breathing symptoms, or reduced production of blood cells in the bone marrow.

Treatment can cause temporary effects such as tiredness, nausea, hair loss, mouth soreness, low blood cell counts, and greater infection risk. The likely effects differ between treatments.

Some effects may appear months or years later. Depending on the person's age and the area treated, these may involve growth, fertility, hormone function, hearing, heart health, kidney function, movement, or development of another cancer. These risks do not occur in everyone. Long-term follow-up is used to look for them early.

Prevention

Most rhabdomyosarcomas cannot be prevented. There is no known lifestyle change that reliably prevents this cancer, and there is no routine screening test for people at average risk.

Inherited cancer syndromes cannot be prevented either. If one is suspected or confirmed, genetic counselling can explain testing, family implications, and any specialist monitoring that may be appropriate. Monitoring may find a tumour earlier, but it cannot guarantee prevention.

Living with it and what to expect

Treatment often continues for many months and can affect school, work, sport, eating, sleep, and family routines. Some periods are more intensive than others. Plans may include time away from usual activities, but many people continue parts of school or work with adjustments.

After treatment, follow-up usually includes examinations and scans. Visits are more frequent at first and become less frequent over time if there are no signs of recurrence. Follow-up also checks growth, movement, organ function, emotional health, and delayed treatment effects.

The outlook is shaped by several factors. Localized tumours that respond well to treatment are often curable. Tumours that have spread to distant sites, return after treatment, or have certain high-risk biological features are harder to cure. Statistics for broad groups cannot predict exactly what will happen to one person.

Recovery may include physiotherapy, occupational therapy, speech therapy, or support with body-image changes. Children and teenagers may need educational help during treatment and support as they return to school. Adults may need workplace adjustments or financial and fertility guidance.

Fear of recurrence is common, especially near scans or appointments. Clear follow-up plans, psychological support, and contact with age-appropriate cancer support services can make this uncertainty easier to manage.

When to get medical help

Arrange a routine medical assessment for a lump that is growing, unexplained swelling, repeated bleeding, ongoing pain, or symptoms that do not settle. A painless lump still deserves assessment if it persists or enlarges.

Seek urgent medical advice for rapidly increasing swelling, difficulty passing urine, blood in the urine, repeated vomiting, a bulging eye, or new changes in vision.

Get emergency help for:

A fever during chemotherapy can be a sign of a serious infection because chemotherapy may lower infection-fighting white blood cells. Follow the oncology team's fever instructions immediately rather than waiting for a routine appointment.

- Trouble breathing or signs that the airway is blocked
- Severe or uncontrolled bleeding
- New weakness, numbness, difficulty walking, or loss of bladder control
- Sudden severe vision loss
- Severe illness or unusual sleepiness during cancer treatment

Common questions

Is rhabdomyosarcoma a type of cancer?

Yes. It is a soft-tissue sarcoma, meaning a cancer that begins in tissues such as muscle, fat, nerves, or blood vessels. Rhabdomyosarcoma starts from immature muscle-forming cells.

Does every growing lump mean rhabdomyosarcoma?

No. Most lumps have another cause, such as a cyst, infection, injury, or non-cancerous growth. A lump that persists, becomes larger, or causes other symptoms should be medically assessed.

Can adults get rhabdomyosarcoma?

Yes, although it is more common in children and teenagers. Diagnosis and treatment in adults still require specialist sarcoma care. The tumour's location and biological features may differ between age groups.

Does rhabdomyosarcoma spread quickly?

It can be aggressive and may spread, but its behaviour is not the same in every person. Subtype, gene findings, location, size, and response to treatment all matter.

Can rhabdomyosarcoma come back after treatment?

Yes. It may return in the original area or elsewhere in the body. Follow-up examinations and scans are intended to check for recurrence and monitor treatment effects. The risk is different for each risk group.

Is rhabdomyosarcoma inherited?

Usually not. Most cases result from DNA changes that occur in tumour cells during life. A small number are linked to inherited cancer syndromes, so genetic counselling may be suggested when personal or family features raise concern.

Sources

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