CONDITION

Klippel-trenaunay syndrome

Klippel-Trenaunay syndrome is a lifelong condition involving abnormal blood vessels and overgrowth, usually in one limb.

WHAT IT IS
Congenital blood vessel malformations with tissue and bone overgrowth

BODY SYSTEM
Blood vessels, lymph vessels, skin, bones, and soft tissues

MAIN SYMPTOMS
Port-wine stain, enlarged veins, limb overgrowth, swelling

WHAT CAUSES IT
Usually a non-inherited PIK3CA gene change

HOW IT IS DIAGNOSED
Clinical examination, Doppler ultrasound, and MRI

MAIN TREATMENT
Compression, symptom control, medicines, and specialist procedures

CAN IT BE CURED?
No, but symptoms can be managed

IS IT AN EMERGENCY?
Sometimes, with major bleeding or possible blood clot

LONG-TERM OUTLOOK
Lifelong condition with symptoms ranging from mild to disabling

Overview

Klippel-Trenaunay syndrome, often shortened to KTS, is a condition present from birth. It affects the development of blood vessels, lymph vessels, bones, and soft tissues. It most often involves one leg, although an arm or several areas can be affected.

KTS is traditionally recognized by three features:

A person does not always have every feature. Some changes are visible at birth, while enlarged veins, swelling, or differences in limb size may become clearer as a child grows.

The abnormal vessels in KTS are usually slow-flow malformations . This means blood or lymph moves through them slowly. KTS does not usually include abnormal high-flow connections between arteries and veins. Those connections may point to a different condition, such as

Parkes Weber syndrome.

KTS matters because it can cause pain, swelling, skin problems, bleeding, difficulty moving, and blood clots. The effects range widely. Some people need little treatment, while others benefit from long-term care involving several specialties.

- A red or purple capillary birthmark, often called a port-wine stain
- Abnormal veins or lymph vessels
- Overgrowth of bone or soft tissue in the affected area

Symptoms

A port-wine stain is often the first visible sign. It is caused by widened tiny blood vessels near the skin. The stain is usually pink, red, or purple and may darken or become thicker over time. It is not a bruise and does not fade when the cause of a normal bruise would have resolved.

Vein abnormalities may include varicose veins or deeper malformed veins. Varicose veins are enlarged, twisted veins near the skin. These changes can lead to aching, heaviness, warmth, or swelling, especially after standing.

Lymphatic malformations affect vessels that normally drain fluid from tissues. They may cause ongoing swelling, small fluid-filled blisters, leakage from the skin, or repeated skin infections.

Overgrowth can affect the length or width of a limb. One leg may become longer, larger, or both. Toes or fingers can also be enlarged. A difference in leg length may change walking and place extra strain on the hips, knees, or back.

Other possible symptoms include:

Symptoms differ between people and between affected body areas. Having KTS does not mean that every possible problem will develop.

- Pain, cramping, or a feeling of pressure
- Easy bleeding from fragile skin or abnormal vessels
- Skin thickening, sores, or slow-healing wounds
- Reduced movement or difficulty finding suitable clothes and shoes
- Blood in the urine or stool if internal vessels are involved

Causes and risk factors

KTS is usually linked to a change in the PIK3CA gene. This gene helps control how cells grow. The change happens by chance in one cell during very early development before birth. Cells that develop from that cell carry the change, while many other cells do not. This pattern is called mosaicism .

Because only some cells are affected, a blood test may not find the gene change. Testing a sample from affected tissue is sometimes more informative, although genetic testing is not needed in every case.

KTS is part of a broader group called PIK3CA-related overgrowth spectrum, or PROS. These conditions share patterns of tissue overgrowth and blood vessel abnormalities.

The gene change is generally not inherited from a parent. People with KTS usually do not have a family history of the condition, and parents did not cause it through anything they did during pregnancy.

There are no established lifestyle or environmental risk factors. KTS affects development before birth and is not contagious.

Diagnosis

Diagnosis begins with a medical history and examination. A clinician looks at the birthmark, veins, swelling, limb size, skin, movement, and differences between the two sides of the body.

A Doppler ultrasound uses sound waves to show blood flow. It can identify abnormal veins and help check for a clot. It can also show whether a malformation has slow or fast blood flow.

Magnetic resonance imaging, or MRI, provides detailed pictures of soft tissue, blood vessels, lymphatic malformations, and overgrowth. Magnetic resonance venography may be used to map veins, especially when a procedure is being considered.

Other tests may be used for a specific concern. These can include X-rays to measure bone length, blood tests when clotting or bleeding is suspected, or imaging of the abdomen and pelvis.

Genetic testing can support the diagnosis by finding a PIK3CA change. A negative result does not necessarily rule out KTS because the change may not be present in the sample tested.

The care team may also check for another vascular condition. In particular, high-flow connections between arteries and veins suggest a diagnosis other than classic KTS.

Treatment and management

There is no single treatment that removes all features of KTS. Care focuses on reducing symptoms, protecting movement and skin, and treating complications. A vascular anomalies service may bring together specialists in blood vessels, skin, blood disorders, imaging, orthopaedics, physical therapy, and surgery.

Compression garments can help control swelling, aching, and pooling of blood in an affected limb. They need to fit correctly. The right type and pressure are chosen after the limb and its circulation have been assessed.

Skin care is important when swelling or lymphatic problems are present. Keeping the skin clean and moisturized, treating small injuries promptly, and using recommended wound care can reduce irritation and infection.

Physical therapy may support strength, joint movement, balance, and walking. Shoe lifts or other orthotic devices can help with some differences in leg length. Larger differences may require assessment by an orthopaedic specialist while a child is growing.

Procedures are selected according to the vessels involved:

Operations can be more complex because abnormal vessels may increase bleeding and clotting risks. Detailed imaging and planning are therefore important.

Medicines that affect cell-growth pathways, including sirolimus or alpelisib, may be considered for some people with severe or complicated PIK3CA-related overgrowth. These medicines require specialist supervision and monitoring. They are not suitable or necessary for everyone.

Blood-thinning medicine may be used to treat a clot or in selected high-risk situations. It is not routinely taken by every person with KTS because abnormal vessels can also increase bleeding risk.

- Laser treatment may lighten a port-wine stain or treat small bleeding vessels
- Sclerotherapy injects a substance into selected abnormal vessels to close them
- Vein procedures or surgery may help carefully selected problems
- Orthopaedic surgery may address a substantial limb-length difference
- Surgery may remove limited overgrown tissue when expected benefits outweigh risks

Complications

Slow blood flow in malformed or enlarged veins can increase the risk of deep vein thrombosis, or DVT. This is a blood clot in a deep vein, usually in a leg. A clot can sometimes travel to the lungs and cause a pulmonary embolism.

Some people develop repeated small clots within a venous malformation. This can cause tender, firm areas and may affect blood-clotting test results. Specialists call this localized intravascular coagulopathy.

Other possible complications include:

Pregnancy can place extra pressure on veins and may worsen swelling or clotting risk. Anyone with KTS who is pregnant or planning pregnancy may benefit from early specialist assessment.

- Chronic swelling and discomfort
- Cellulitis, a bacterial infection of the deeper skin
- Skin breakdown, ulcers, or fluid leakage
- Bleeding from skin, bowel, bladder, or reproductive organs
- Reduced mobility from overgrowth or joint strain
- Anaemia from repeated or significant bleeding
- Emotional distress related to pain, appearance, or daily limitations

Prevention

KTS cannot currently be prevented. The gene change usually occurs randomly during early development and is not caused by behaviour, diet, or pregnancy choices.

Some complications may be reduced through planned care. This can include using prescribed compression, staying active within comfortable limits, caring for the skin, and attending follow-up for limb growth or vein problems.

Before surgery, pregnancy, or long periods of reduced movement, the care team may assess clotting and bleeding risks. Preventive blood-thinning treatment is individualized because both risks can be present in KTS.

Living with it and what to expect

KTS is lifelong, but it does not affect everyone in the same way. The birthmark and malformed vessels usually remain. Tissue or bone overgrowth may become more noticeable during childhood and adolescence as the body grows.

Symptoms may also change with hormones, pregnancy, weight changes, injuries, surgery, or long periods of limited movement. Swelling and aching may be worse later in the day or after prolonged standing.

Some people have mild changes and take part in most activities without major limits. Others need repeated procedures, compression, mobility support, pain management, or help with wounds and infections. Treatment can reduce many symptoms, but new concerns may appear over time.

Routine follow-up is based on the areas affected. Children may need regular measurement of limb length and growth. Adults may need review for changing pain, swelling, skin problems, bleeding, or vein symptoms.

Daily life can involve practical adjustments, such as fitted compression clothing, supportive footwear, rest breaks, or planning around treatments. Support for body image, anxiety, or low mood can also be valuable. These concerns are part of living with a visible and sometimes painful condition, not a personal failing.

When to get medical help

Seek emergency help for symptoms that could indicate a pulmonary embolism or severe bleeding:

Get urgent medical assessment for new one-sided swelling, warmth, redness, or pain, especially if it begins suddenly. These symptoms can occur with a DVT. Fever with rapidly spreading redness, increasing pain, or feeling very unwell may indicate cellulitis and also needs prompt care.

Contact your usual care team soon for repeated bleeding, blood in urine or stool, a new wound, persistent fluid leakage, increasing pain, or a noticeable change in limb size or function.

Routine appointments are appropriate for reviewing compression garments, monitoring growth, discussing a stable birthmark, or managing ongoing swelling that has not suddenly changed.

- Sudden shortness of breath
- Chest pain, especially if worse when breathing in
- Coughing up blood
- Fainting or severe weakness
- Heavy bleeding that does not stop with firm pressure

Common questions

Is Klippel-Trenaunay syndrome cancer?

No. KTS is a developmental condition involving overgrowth and malformed vessels. It is not cancer, and the affected tissue does not spread through the body like cancer.

Is Klippel-Trenaunay syndrome inherited?

Usually not. The PIK3CA change generally happens by chance after conception and affects only some cells. Most people with KTS did not inherit it from a parent and do not pass on the mosaic change through families.

Does a port-wine stain mean someone has KTS?

No. Port-wine stains can occur on their own and are more common than KTS. KTS is considered when the stain appears with abnormal veins or lymph vessels and overgrowth of the affected area.

Can exercise make KTS worse?

Appropriate movement often supports circulation, strength, and joint health. The best activity depends on pain, swelling, skin condition, and the vessels involved. High-impact or contact activities may need adjustment when there is significant overgrowth, fragile skin, or bleeding risk.

Can the affected limb be made the same size as the other one?

Some differences can be improved with compression, shoe lifts, procedures, or orthopaedic treatment. Complete symmetry may not be possible. Treatment usually aims to improve comfort, movement, skin health, and everyday function rather than appearance alone.

What is the difference between KTS and Parkes Weber syndrome?

Both can cause a birthmark and limb overgrowth. Classic KTS involves slow-flow venous, capillary, and lymphatic malformations. Parkes Weber syndrome includes high-flow connections between arteries and veins. Doppler ultrasound and MRI help distinguish them because their monitoring and treatment differ.

Sources

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