CONDITION

Klippel-trenaunay-weber syndrome

A condition present from birth that causes abnormal blood vessels and uneven tissue or limb overgrowth.

WHAT IT IS
Congenital blood vessel malformations with uneven tissue overgrowth

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

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

WHAT CAUSES IT
Usually a non-inherited gene change before birth

HOW IT IS DIAGNOSED
Examination, Doppler ultrasound, and MRI

MAIN TREATMENT
Compression, specialist medicines, procedures, and surgery

CAN IT BE CURED?
No - existing malformations cannot be completely removed

IS IT AN EMERGENCY?
Sometimes - sudden breathlessness, major bleeding, or painful limb swelling

LONG-TERM OUTLOOK
Lifelong condition often manageable with specialist follow-up

Overview

Klippel-trenaunay-weber syndrome, or KTW syndrome, is an older name for a condition involving abnormal blood vessels and overgrowth of part of the body. It is present from birth, although some features become clearer as a child grows.

The name can be confusing. Klippel-Trenaunay syndrome usually involves slow-flow malformations of veins, tiny skin vessels, or lymph vessels. "Slow-flow" means blood does not pass through them under high pressure. Parkes Weber syndrome is a different condition involving fast-flow connections between arteries and veins.

Historically, both were sometimes called Klippel-trenaunay-weber syndrome. Specialists now try to separate them because their causes, complications, and treatments are not identical.

Typical features include a red or purple birthmark called a capillary malformation, enlarged or twisted veins, and overgrowth of bone or soft tissue. One leg is affected most often, but an arm, the trunk, or several areas may be involved.

The condition matters because abnormal vessels may cause pain, swelling, skin problems, bleeding, or blood clots. Overgrowth can affect walking, footwear, joint movement, and body image. People with fast-flow artery-to-vein connections may also place extra strain on the heart.

Symptoms

Symptoms differ according to which tissues and blood vessels are involved. Some features are visible at birth, while enlarged veins and swelling may become more noticeable during growth, puberty, pregnancy, or periods of reduced mobility.

Common features include:

Overgrowth may involve bone, muscle, fat, or skin. A difference in leg length can affect posture and walking. The larger limb is not always the longer one, and some people have tissue wasting rather than overgrowth in certain areas.

Small fluid-filled blisters may form when lymph vessels are involved. These can leak clear or blood-stained fluid. Bleeding may also occur from fragile skin, veins, or malformations inside the bowel, bladder, or reproductive organs.

A blood clot in a deep vein can cause new pain, warmth, tenderness, or swelling, usually in one limb. A clot that travels to the lungs may cause sudden breathlessness, chest pain, faintness, or coughing up blood.

Fast-flow artery-to-vein connections suggest Parkes Weber syndrome rather than classic Klippel-Trenaunay syndrome. They may make an area feel unusually warm or cause a pulse, vibration, or rushing sound. Extensive fast-flow malformations can make the heart work harder.

- A flat pink, red, or purple port-wine birthmark
- An arm or leg that is longer, wider, or both
- Enlarged surface veins or varicose veins
- Swelling caused by abnormal veins or lymph vessels
- Aching, heaviness, cramps, or reduced movement
- Repeated skin irritation, infection, or slow-healing wounds

Causes and risk factors

Classic Klippel-Trenaunay syndrome is usually linked to a change in the PIK3CA gene. This gene helps control cell growth. The change happens by chance in some cells during early development before birth. This is called a somatic or mosaic gene change because only part of the body carries it.

The condition is therefore usually not inherited from a parent. Parents did not cause it through anything they did before or during pregnancy.

Parkes Weber syndrome can be associated with changes in genes such as RASA1 or EPHB4 . Some of these changes can be inherited. Genetic counselling may be helpful when fast-flow malformations, several affected relatives, or multiple small capillary birthmarks suggest an inherited condition.

There are no established lifestyle risk factors for classic Klippel-Trenaunay syndrome. Its severity is influenced by where the changed cells developed and which tissues are involved.

Diagnosis

Diagnosis starts with a physical examination and a detailed history. The pattern of the birthmark, abnormal veins, swelling, and tissue or bone growth may strongly suggest the condition.

Imaging shows how deep the malformations extend and whether blood flow is slow or fast. Tests may include:

Blood tests may be used before procedures or when there is concern about bleeding or clotting. Some extensive venous malformations use up clotting proteins within the affected area, even without a large visible clot.

Genetic testing may help clarify the diagnosis. Because a PIK3CA change may exist only in affected tissue, a blood test can be negative even when the condition is present. Testing a tissue sample is sometimes more informative, but it is not needed or appropriate for everyone.

Assessment by a vascular anomalies team can help distinguish Klippel-Trenaunay syndrome from Parkes Weber syndrome and other overgrowth conditions.

- Doppler ultrasound to assess blood flow and look for clots
- MRI to map veins, lymph vessels, muscles, and other soft tissues
- MR angiography or other vascular imaging when fast flow is suspected

- X-rays or standing limb measurements to compare bone length

Treatment and management

There is no single treatment that removes every feature. Care is based on the symptoms, blood-flow pattern, tissues involved, and effect on daily life. A specialist team may include experts in vascular medicine, dermatology, radiology, haematology, orthopaedics, rehabilitation, genetics, and surgery.

Compression garments can reduce swelling, aching, and blood pooling. They need to be measured and fitted carefully. Lymphoedema therapy may include compression, movement, skin care, and techniques that help fluid drain from the limb.

Regular activity supports circulation and joint movement. Some people benefit from physiotherapy, supportive footwear, shoe lifts, or other mobility aids. Skin care is important when swelling, leaking lymph vessels, or fragile areas are present.

Procedures may be considered for troublesome malformations:

Removing a large malformation completely may not be possible and can damage nearby nerves, muscles, or healthy vessels. Procedures are therefore planned around a specific goal, such as reducing pain, bleeding, swelling, or heart strain.

Medicines that target abnormal cell-growth pathways, including sirolimus or alpelisib, may be considered for selected people with severe or complicated disease. These treatments require specialist supervision and monitoring because they can cause significant side effects.

Blood-thinning medicine may be used for a confirmed clot or around certain high-risk procedures. It is not routinely suitable for everyone, especially when abnormal vessels also create a bleeding risk.

- Laser treatment can lighten some capillary birthmarks or treat surface bleeding
- Sclerotherapy closes selected abnormal veins or lymphatic spaces
- Endovascular procedures treat vessels from inside using small tubes
- Embolization blocks fast-flow artery-to-vein connections
- Surgery may remove selected tissue, treat veins, or address limb-length differences

Complications

Possible complications include chronic pain, reduced mobility, arthritis, skin breakdown, and

repeated skin infections. Persistent swelling can make clothing and footwear difficult to fit.

Slow blood flow in enlarged veins raises the risk of superficial clots and deep vein thrombosis. A deep vein thrombosis can sometimes lead to a pulmonary embolism, meaning a clot has reached the lungs.

Bleeding ranges from minor oozing to more substantial blood loss. Internal vascular malformations may cause blood in the urine or bowel movements. Heavy menstrual bleeding can also occur.

Large differences in limb length may affect the hips, knees, spine, or walking pattern. Overgrowth around a joint can restrict movement.

Fast-flow malformations can divert a large amount of blood directly from arteries into veins. If extensive, this may eventually strain the heart and cause breathlessness, tiredness, fluid retention, or a racing heartbeat.

Living with a visible birthmark or a differently sized limb can affect confidence and emotional wellbeing. Psychological support or contact with people who have similar vascular conditions may be useful.

Prevention

Klippel-Trenaunay syndrome cannot currently be prevented. The usual PIK3CA change happens by chance during development before birth and is not caused by diet, activity, medicines, or parenting.

Complications may sometimes be reduced through planned follow-up, properly fitted compression, skin care, and early assessment of new pain, swelling, bleeding, or wounds. Before surgery, pregnancy, or a long period of immobility, the care team may reassess clotting and bleeding risks.

When an inherited fast-flow condition is suspected, genetic counselling can explain family testing and the chance of passing the gene change to a child. This does not apply to most people with classic Klippel-Trenaunay syndrome.

Living with it and what to expect

KTW syndrome is lifelong. Existing vascular malformations do not disappear on their own, and the affected limb usually remains different in size or appearance.

Symptoms can change over time. Growth spurts, hormonal changes, pregnancy, weight

changes, injuries, operations, and reduced movement may increase swelling or make abnormal veins more noticeable. Treatment may improve a specific problem, but further procedures can be needed if symptoms return or different vessels become troublesome.

Children may need repeated measurements of limb length and growth. Treatment plans can change as the skeleton matures. Adults are more likely to need monitoring for pain, skin problems, clots, bleeding, or reduced mobility.

Many people attend school, work, exercise, travel, and have families. Practical adjustments may include custom clothing, compression garments, suitable footwear, planned movement during long journeys, or changes to activities that repeatedly injure fragile skin.

Pregnancy is possible, but it can increase venous swelling and clotting or bleeding concerns. Planning with an obstetric team and vascular specialists allows these risks to be assessed before and during pregnancy.

Routine follow-up is generally intended to monitor symptoms, growth, mobility, skin health, and treatment results. New symptoms should not automatically be assumed to come from KTW syndrome, so ordinary health concerns still deserve appropriate assessment.

When to get medical help

Arrange routine medical review for increasing swelling, persistent pain, new visible veins, leaking lymph blisters, changes in walking, or a wound that is slow to heal. Blood in urine or bowel movements and unusually heavy menstrual bleeding also need timely assessment.

Seek urgent same-day help for:

Call emergency services for sudden breathlessness, chest pain, fainting, coughing up blood, severe uncontrolled bleeding, or new confusion. These symptoms may indicate a clot in the lungs, major blood loss, or another serious problem.

New breathlessness with ankle swelling, marked tiredness, or a racing heartbeat also needs prompt assessment, particularly when fast-flow malformations are present.

- Sudden new pain, warmth, redness, or swelling in a limb
- Bleeding that is difficult to stop with firm pressure
- A spreading red, hot, painful skin area, especially with fever
- Rapidly worsening pain or swelling after an injury or procedure

Common questions

Is Klippel-trenaunay-weber syndrome the same as Klippel-Trenaunay syndrome?

The names have often been used interchangeably. However, modern classification separates classic slow-flow Klippel-Trenaunay syndrome from Parkes Weber syndrome, which has fast-flow connections between arteries and veins. Imaging helps establish which pattern is present.

Is KTW syndrome inherited?

Classic Klippel-Trenaunay syndrome is usually caused by a PIK3CA change that occurs by chance in some cells before birth. It is not usually inherited. Some Parkes Weber cases involve inherited RASA1 or EPHB4 gene changes.

Will the affected limb keep growing?

The limb generally grows as the person grows, but the difference between limbs may become more or less noticeable. Growth is monitored during childhood because bone length and soft-tissue size do not always change at the same rate.

Can compression garments make the condition go away?

No. Compression does not remove abnormal vessels or overgrown tissue. It can help control swelling, aching, and blood pooling while it is being worn.

Can I exercise with KTW syndrome?

Many people can exercise, and movement often helps circulation and joint function. Suitable activities depend on pain, skin condition, mobility, clot history, and the type of vascular malformation. Activities that repeatedly injure fragile tissue may need modification.

Can the port-wine stain become cancerous?

A capillary birthmark is not usually cancerous. Changes such as repeated bleeding, a new lump, an open sore, or a rapid change in appearance should still be assessed rather than assumed to be part of the birthmark.

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