

Possible treatments

The following list of treatments are often advised for KTS:

- No treatment; just keep an eye on it.
- Laser treatment of the port-wine stain.
- Pain killers.
- Compression stockings.
- Blood Thinners.
- Surgery of body parts with overgrowth/ inhibiting growth of the longer leg.
- Varicose Vein treatment.
- Embolization of (localized) vascular malformations by the interventional radiologist.
- Treatment targeting the PIK3CA mutation with Sirolimus (or other medicine).

Do's and don'ts

Do:

- Maintain a healthy lifestyle.
- Compression stockings (in case of complaints).
- Consult experts.

Don't:

- Smoke.
- Use estrogen-based contraception.
- Work that requires standing.

MORE INFORMATION:
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Klippel-Trenaunay Syndrome (KTS)

Klippel-Trenaunay Syndrome (KTS) has three main features: a large port-wine stain birthmark on one or more limbs (often one leg), blood vessel abnormalities and overgrowth of the limb with the Port-Wine stain birthmark.

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Detailed description

Klippel Trenaunay Syndrome (KTS) is a rare, non-hereditary, medical condition that was first described in 1900 by the French physicians Maurice Klippel and Paul Trénaunay as the triad of; a large port-wine stain on one or more limbs (often one leg) in combination with a vascular malformation of the veins and overgrowth of the limb. Sometimes there are additional symptoms such as syndactyly (fusion of toes or fingers) and lymphedema (the build up of fluids in the soft tissue).

Cause

In 2016, it was discovered that KTS is a condition based on a change in DNA (a mutation) that is only present in the affected tissue (such as the port-wine stain or tissue with overgrowth) and not in the rest of the body. This is called a DNA mosaic. This means that KTS is not hereditary.

The mutation is in the PIK3CA gene and develops early (weeks 5-8) in pregnancy. The PIK3CA mutation ensures that the affected cells become more active, resulting in increased vessel growth.

Signs and Symptoms of KTS

- Limb overgrowth. In childhood, the overgrowth and especially the leg length difference should be monitored. This often remains in proportion.
- In the port-wine stain birthmark of KTS, purple blisters are sometimes seen that weep or bleed.
- At a later age, KTS can cause more issues with varicose veins and complaints based on chronic venous insufficiency such as tired and heavy legs, swelling and eventually a chance of a venous ulcer.
- Sometimes the swelling develops due to a poorly functioning lymphatic drainage system, resulting in lymphedema. Lymphedema also makes people more susceptible to erysipelas (also referred to as cellulitis).
- The risk of thrombosis is increased in people with KTS. This can manifest itself in painful vein inflammations, but also as a Deep Vein Thrombosis of the leg or arm. The risk of pulmonary embolism is also increased. Smoking and the use of estrogen-based contraception increases the risk of thrombosis which is not recommended. Contraception with 'progesterone-only' such as hormone coil or mini-pill can be safely used.
- Pain is a common complaint with KTS. In addition to the above causes, there may also be growing pains, arthritis and neuropathic pain.

- In some people with KTS there is also an association of abdominal and pelvic organ issues; this can lead to gynecological problems (severe periods or problems during pregnancy), urological and intestinal problems (bleeding). For women who want to become pregnant, a consultation with a gynecologist before pregnancy is advised.

Possible complications

- Leg length difference: The leg length difference should be monitored at a young age so that the patient does not have a leg length difference when older.
- Weeping and bleeding from the port-wine stain birthmark.
- Lymphedema and erysipelas.
- Varicose veins: in the long term, someone with KTS may develop more varicose veins issues such as tiredness, heaviness, swelling and venous ulcer.
- Thrombosis and pulmonary embolism: we see more often in KTS patients and it can occur at a young age. In the case of unexplained leg or lung complaints, this complication should be considered.

