DEATH DUE TO PULMONARY EMBOLISM AND PULMONARY HYPERTENSION IN KLIPPEL TRENAUNAY SYNDROME.

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ABSTRACT Death due to Pulmonary Embolism and Pulmonary Hypertension in Klippel Trenaunay Syndrome. Klippel Trenaunay Syndrome (KTS) is a congenital disorder characterized by bony and soft tissue hypertrophy, capillary malformation, and varicosities. This syndrome has several complications such as sepsis, hypercoagulability, rectal and bladder bleeding, venous thrombosis, and pulmonary embolism. Massive haemorrhage due to rupture of malformed leg vessels has been reported as a cause of death. We present a case of KTS where an adult Caucasian male died suddenly at home. Clinically, the decedent had chronic pain and warmth in both legs, port wine stain, a longer and hypertrophied left leg, vascular malformation, big toes, and swollen soft tissues. Autopsy did not reveal cardiomegaly or intrusive coronary artery disease. A ropy, granular, organized, and formed anti-mortem blood clot is observed in the pulmonary trunk, branches, and lungs. He had the abnormal structure of the feet, as well as varicose veins and swelling of the legs. Grossly, the left calf circumference is greater than the right calf circumference, varicosity of the left lower extremity, bony and soft tissue hypertrophy, especially the lower left extremity, swollen and large toes, and cerebral swelling. Microscopic finding of lungs includes thickened pulmonary interstitium with congestion and oedema. Pulmonary arterioles showed hypertrophied muscular layers with numerous thrombi of different ages, ranging from recent to remote, with the complete organization of some vessels; trichrome stains confirmed the presence of recanalization and some plexiform lesions. There is a thick muscular wall, organized thrombotic material, intimal fibrosis, and artery recanalisation.

KEYWORDS Klippel Trenaunay Syndrome, KTS, Autopsy, Pulmonary embolism, bone and soft tissue hypertrophy

Introduction

Klippel Trenaunay Syndrome (KTS) is a congenital disorder characterized by bony and soft tissue hypertrophy, capillary malformation, and varicosities (1). This syndrome has several complications such as sepsis, hypercoagulability, rectal and bladder bleeding, venous thrombosis, and pulmonary embolism. Many clinicians, pathologists, and forensic pathologists are unfamiliar with this rare entity. Forensic pathologists can narrow down the complications and carefully look for the potential causes of death in KTS. For example, massive haemorrhage due to rupture of malformed leg vessels has been reported to cause death.

Those with large arteriovenous malformations are at risk of the formation of blood clots in the blood vessels. If a large-volume blood flow occurs, high-output cardiac failure may develop. We present a case of KTS where an adult Caucasian male lived alone and died suddenly at home. Clinically, the decedent had chronic pain and warmth in both legs. The left was worse than the right, with a longer and hypertrophied left leg, vascular malformation, big toes, and swollen soft tissues (Fig 1,2,3,5). Additionally, he had port wine stain (Fig. 4). However, the
left leg is most commonly hypertrophied, and multiple affected limbs have been reported, such as arms, trunk, and rarely the head and neck. Confirmatory toxicological analyses indicate no evidence of prescription or non-prescription drug intoxication.

An autopsy revealed no traumatic injury. The usual treatment of KTS includes compression garments, support stockings, pumps, laser treatment, surgical intervention, and preventive anticoagulation. The decedent refused to routinely wear support stockings. Reportedly, he received anticoagulation therapy infrequently. Although the decedent had large venous malformations, organomegaly, and chronic lymphedema, there was no evidence of haemorrhage from vascular malformations. There is also no evidence of cardiomegaly or intrusive coronary artery disease. A ropy, granular, organized, and formed anti-mortem blood clot is observed in the pulmonary trunk, branches, and lungs (Fig 6,7).

A 39-year-old male was found dead sitting on a recliner in the living room. The decedent was sitting upright but slumped
slightly to his right in the recliner, appearing to have just fallen asleep. He was wearing sweat pants and a t-shirt. Reportedly, he came to a developmental center in the morning and evening and was given his medications. The developmental center reportedly stated that the decedent suffered from a congenital syndrome with the clinical diagnosis of Klippel-Trenaunay Syndrome. He had the abnormal structure of the feet, as well as varicose veins and swelling of the legs. Grossly, the left calf circumference is greater than the right calf circumference, varicosity of the left lower extremity, bony and soft tissue hypertrophy, especially the lower left extremity, swollen and large toes (Fig. 1-5), cerebral swelling, but no evidence of arteriovenous malformation in the brain, pulmonary congestion and oedema with focal thickening of the pulmonary arterioles. Microscopic finding of lungs includes thickened pulmonary interstitium with congestion and oedema; pulmonary arterioles showed hypertrophied muscular layers with numerous thrombi of different ages, ranging from recent to remote, with the complete organization of some vessels. Trichrome stains (Fig. 9) confirmed the presence of recanalization and some plexiform lesions. There is a thick muscular wall, organized thrombotic material, intimal fibrosis, and artery recanalisation. Most vessels showed evidence of thromboembolic vasculopathy. There is no evidence of gross arteriovenous malformation of the brain, no significant traumatic injuries or tumours in the liver, the throat, or the brain. KTS is a congenital syndrome involving enlarged veins and arteries, limb hypertrophy, and capillary malformations. In KTS,

lymphedema may be present, true hypertrophy of the affected soft tissues is always present, and hemangiomas (enlarged or abnormal vessels/collection of vessels, i.e. port wine stains) may be present vary in depth. There may be limited to the skin or extend deeper into the subcutaneous tissue, including muscle and bone. Visceral organs, such as the pleura, the spleen, the liver, the urinary bladder, and the colon, may also be affected.

KTS generally affects a single extremity, although cases of multiple affected limbs have been reported. The leg is the most common site, followed by the arms, the trunk, and rarely the head or neck. Limb hypertrophy causes increased length (bony involvement) and/or girth (soft tissue involvement). Secondary to PE, the decedent had Cor pulmonale, pulmonary hypertension with recanalization and a few plexogenic changes in the pulmonary artery. Internal examination revealed right ventricular hypertrophy of the heart and congested lungs. The cause of death in this decedent was pulmonary Embolism and Pulmonary Hypertension in a case of Klippel Trenaunay Syndrome.

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