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Atypical presentation (or a variant) of Klippel-Trenaunay-Weber syndrome: a case report and literature review

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Abstract

We report the case of a 65-year-old man who we thought was an atypical presentation or a variant of the Klippel-Trenaunay-Weber syndrome. Although it is primarily a disorder of infancy and childhood, it has been reported to present in adulthood also[1,2]. Our case report highlights the need to consider the differential diagnosis of Klippel-Trenaunay-Weber syndrome in patients presenting with suggestive symptoms and signs, irrespective of their age.

Keywords

Klippel-Trenaunay-Weber syndrome; capillary haemangioma; arteriovenous malformation; port wine stain.

Case history

A 65-year-old man presented with a 3-year history of gradually worsening swelling of the left side of his body, including his face, neck, arm and leg. He walked mainly on the right side of the body as the left side of his body felt heavy, numb, and he had a backache due to radiculopathy, restricting his body movements. He had difficulty for example combing his hair and hanging clothes using his left arm. His left hand had been gradually increasing in size and he had begun to drop items from the left hand. He suffered from intermittent headaches and reported an occasional right-sided chest swelling, which would disappear spontaneously before re-appearing. He had no history of anterior neck swellings, kyphosis, tongue enlargement, change in voice, increased sweating, prognathism, or visual problems. His symptoms were disabling and he needed his son's help with most of the activities of daily living. He had a past history of diabetes mellitus, hypertension, depression, rectal bleeding, bilateral carpal tunnel syndrome, and lumbosacral radiculopathy, for which he was considering an operation offered by the spinal surgeons.

On examination, he was an obese man with a blood pressure of 173/62 mm Hg, a regular heart rate of 62 bpm, oxygen saturation of 97% in room air, and a temperature of 36.3°C. He had bilateral gynaecomastia predominantly on the left side. His heart sounds were normal, without

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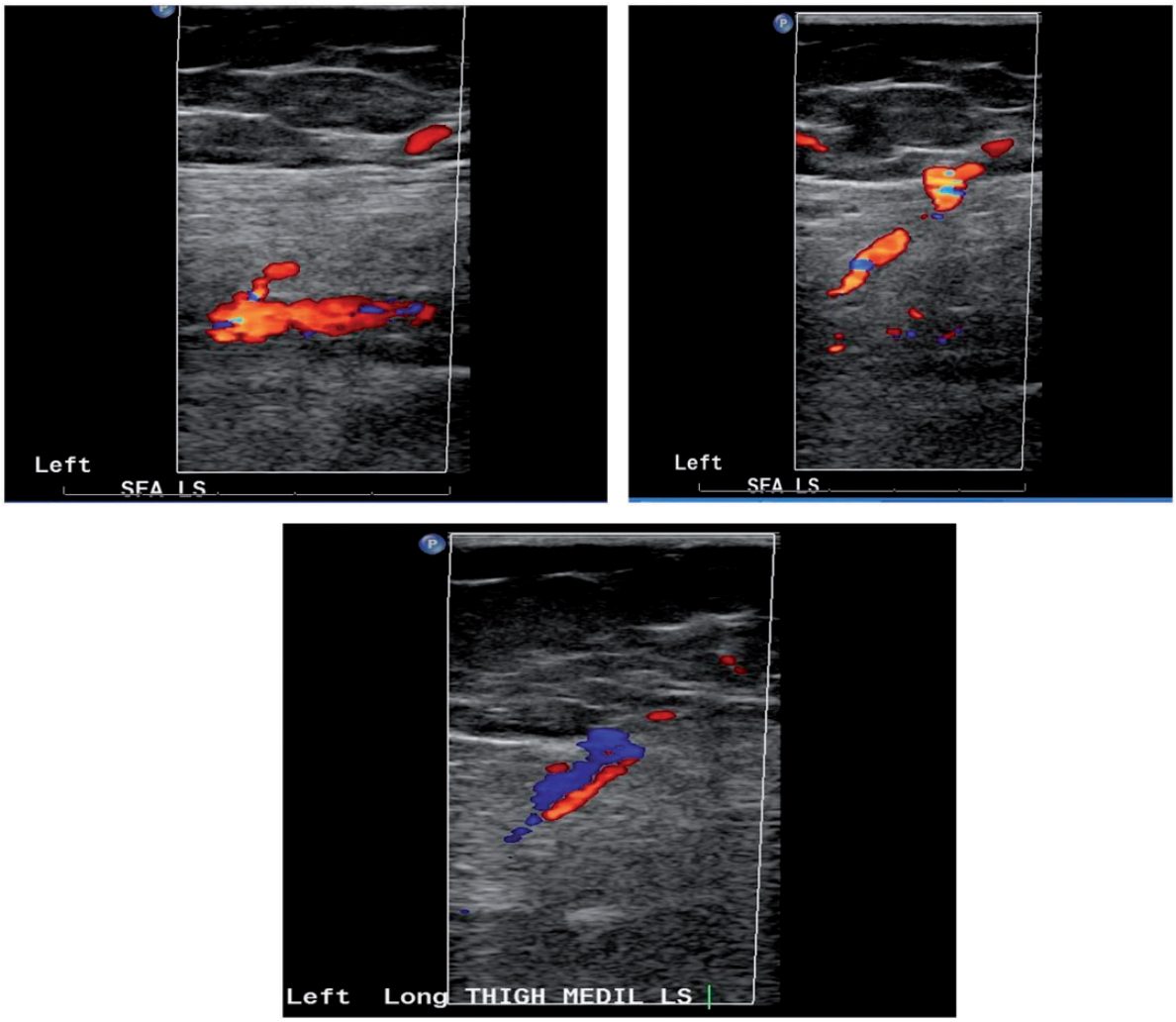

Fig. 1. Abnormal arterial and venous branches as mentioned in the text.

any murmurs, and his jugular venous pressure was not increased. He had bilateral leg swelling, predominantly the left leg, without pitting oedema. His left hand was slightly larger in size by 2 cm than the right hand. He had superficial varicose veins in both legs. The left upper and lower limbs were greater in circumference by a couple of centimetres from the right upper and lower limbs. His chest was clear on auscultation. His abdomen was soft and non-tender. The thyroid and lymph node examination was normal. His range of eye movements and visual fields were normal. A complete neurologic examination was difficult due to severe back pain, restricting his spinal and lower limb movements. However, he had a decreased sensation to light touch and impaired sense of joint position in the left upper and lower limbs.

The differential diagnoses formed included a vascular pathology, a hormonal disorder, and neuropathy-related hypertrophy/atrophy of the limbs. He was referred to the vascular surgeons for further investigations and management. A Doppler ultrasound scan (USS) of the left lower and left upper limbs was performed, which showed a small segment of an abnormal arterial tree in the lower limb of unknown significance. The findings were in the form of prominent arterial and venous branches ramifying through the muscles and subcutaneous fat particularly in the left thigh and the majority of the arterial branches appeared to be rising directly from the superficial femoral artery (Fig. 1). Blood tests including organ and hormonal profiles were normal. Based on the clinical history and the findings on the Doppler USS, the radiologist suggested the possible diagnosis of Klippel-Trenaunay-Weber syndrome. The vascular surgeon also raised the possibility of a whole-body Klippel-Trenaunay-Weber syndrome and advised excluding occlusion of the subclavian vein, iliac vein, and/or narrowing of the thoracic inlet/outlet. Magnetic resonance imaging (MRI) of the spine revealed degenerative changes in both cervical and lumbosacral regions, with bilateral stenosis at L4/L5 and L5/S1 regions, and

diffuse disc bulge at L4/L5 with changes in the facet joints. Magnetic resonance angiography (MRA) and venography (MRV) showed normal vessels supplying the upper and lower parts of the body.

Discussion

Klippel-Trenaunay-Weber syndrome is a rare congenital condition usually presenting early in infancy or childhood. It consists of a triad of port wine stain (capillary haemangioma), varicose veins, and hypertrophy of an affected limb. The French physicians, Klippel and Trenaunay, reported the first case in 1900[3]. In 1907, Weber reported these three findings with an additional arteriovenous malformation and angioma[4]. Therefore, for practical reasons the syndrome is often jointly called the Klippel-Trenaunay-Weber syndrome. Klippel-Trenaunay-Weber syndrome affects males and females equally, and the incidence rate is up to 1:40,000. The exact cause is not known. There are different theories of its manifestation especially on abnormal vasculogenesis during embryonic and foetal development. These theories involve intrauterine damage to the sympathetic ganglia[5], defect of the mesodermal duct during foetal development[6], and deep vein abnormalities leading to varicosities and hypertrophy of the limbs[7]. Usually it is sporadic; however, there are reports of autosomal dominant[8] and paradominant inheritance[9].

The port wine stain or the capillary haemangioma is usually the first presentation of the condition. The colour can vary from salmon pink to purple. The haemangioma can be superficial, or it can spread deeply into the tissues and internal organs, which in itself can present with internal or external bleeding[10]. Varicosities are usually congenital and superficial; however, they can be deeper and extensive involving some internal organs. In women, the varicosities can expand during pregnancy; therefore special care is needed, and serial ultrasonograms should be done to strictly monitor any expansion. Foetal growth retardation due to Klippel-Trenaunay-Weber syndrome has been reported[11]. Arteriovenous malformation associated with Klippel-Trenaunay-Weber syndrome can affect the spine, illustrating the importance of investigating their presence[12]. Limb hypertrophy usually develops during infancy and childhood. Other features that can be associated with Klippel-Trenaunay-Weber syndrome are polydactyly[13], syndactyly, seizures and hemimegalencephaly[14], spina bifida, lymphatic obstruction, chronic venous insufficiency, limb ulceration, and thromboembolism[15]. Association with tumours including angiosarcomas has been reported[16].

The diagnosis is always based on the history and the classic physical findings on examination. There is no laboratory test to confirm the condition. No uniform diagnostic criteria have been established[17] as the condition can present with a wide range of symptoms. A restrictive diagnostic criterion[18] has been proposed, which highlights that the growth can be increased or decreased, varicosities need not be present, and there can be arteriovenous fistulae. There are reports of unusual variants of the syndrome[19,20]. Doppler USS, computed tomography, MRI and MRA/MRV scans can be performed but they are not used to make a diagnosis. The treatment varies, and is mainly for symptom control as their in no permanent cure. Compression garments are used for venous and lymphatic stasis problems. Skin infections can be treated with antibiotics. Orthopaedic correction can be considered for significant limb length or girth discrepancies. Laser treatment has been in use as the main management for capillary haemangioma. The symptomatic superficial varicosities can be treated with stripping, ligation and sclerotherapy. Analgesics help pain control; however, early referral to a specialist pain clinic should be considered for long-term management. As there are multiple symptoms in patients with this condition, they are usually managed by multidisciplinary team input. Generally, patients with Klippel-Trenaunay-Weber syndrome live a good life with optimum symptom control, but the complications can be life threatening.

Our patient did not have a family history of Klippel-Trenaunay-Weber syndrome. He did not have capillary haemangioma either. In view of his unique and unusual clinical history, examination findings, abnormal arterial tree pattern on Doppler USS and the absence of some classic features of Klippel-Trenaunay-Weber syndrome, the diagnosis of an atypical presentation or a variant of the syndrome was considered. Genetic screening was not offered in view of an absent family history, and because the condition is usually sporadic in nature. He continued to be reviewed by the vascular surgeons and was having follow-up with the spinal surgeons with regard to lumbosacral radiculopathy.

Teaching points

- Klippel-Trenaunay-Weber syndrome mainly presents in infancy and childhood, however it has been reported to present in adults also. Hence, it is important to consider the differential diagnosis of this condition in adulthood as and when appropriate.
- The possibility of an atypical presentation (or a variant) of Klippel-Trenaunay-Weber syndrome should be borne in mind, and a specialist opinion be sought when there is a doubt about the diagnosis.
- While a diagnosis is being formed, the many disabling symptoms should be managed with multidisciplinary input.
- Further research is needed to learn more about this unique and rare condition to better understand the pathophysiology and its long-term management.

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