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Mystery of a giant leg

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DESCRIPTION

A 20-year-old man presented with gradual enlargement and disability of the left lower limb since birth. At birth his parents noticed that his left lower limb was a bit bigger than his right, and since then it gradually enlarged in length and breadth, with a distorted shape. His left leg became so big that he could not use it for the last 10 years and became bed bound. Clinical examination revealed a port wine stain on his left lower abdominal wall and part of the left lower limb. His left lower limb was hugely enlarged from his left buttock to the toes. The skin over that limb was hyperpigmented, dry and scaly without any skin ulceration or engorged vessels. His left leg resembled an elephant's leg, with a length of 5 feet 1 inches (approximately 155 cm) and maximum diameter of 47 inches (approximately 119 cm; figure 1). His right leg was normal. An x-ray of the left limb showed all the bones and soft tissue shadow were grossly enlarged. The head of the femur was dislocated from the hip joint and all the tarsal and metatarsal bones were widely displaced from each other. With all the above features, he was diagnosed as having Klippel–Trenaunay–Weber syndrome. To the best of our knowledge, there is no previous report on Klippel–Trenaunay–Weber syndrome with such an enlarged limb.

Klippel–Trenaunay–Weber syndrome is a combination of venous and capillary malformations associated with soft

tissue and/or bony hypertrophy, with or without lymphatic malformations.¹ Typically, only one limb is involved, with the lower limbs representing 70% of cases.² The aetiology of this syndrome is unknown, but some authors have suggested that it results from a mesodermal abnormality that occurs during fetal development.³ Bone hypertrophy usually results in limb length discrepancy. Treatment depends on the extent of the lesion, the severity of the symptoms and disability. The common surgical operations attempted are the excision, repair of venous malformation and debulking operations for tissue deformity with partial or complete amputations of the limbs.¹

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Competing interests None.

Patient consent Obtained.

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Figure 1 Giant left leg (5 feet 1 inches (approximately 155 cm) in length and maximum diameter of 47 inches (approximately 119 cm)).

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