

Limb Size Discrepancy in a 29 yo Male

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HISTORY: 29 yo M who first noticed his right leg was “more developed” than the left when he was in high school and this has persisted since that time. In addition to the limb being larger, he complains of swelling of the extremity with prolonged standing/sitting. He wears compression stockings when at work which helps with the swelling. He also gets discomfort and tightness with prolonged physical activity. He reports prominent veins on his right foot for years. He has been able to play soccer and is an active young male. He denies numbness, tingling, calf rubor, hair changes, back pain, weakness, pain at night, bowel or bladder dysfunction, fevers, chills, abdominal pain, nausea, vomiting, joint swelling, joint pain, and lymphadenopathy. He has been seen by various other physicians and has had ultrasounds and an MRI which have not revealed a diagnosis. Negative for prior abdominal/inguinal surgeries. Family history negative for bone/joint disorders or limb size discrepancy.

PHYSICAL EXAM: Vital signs within normal limits. Well appearing male with right leg larger than left. Arms symmetric. Skin exam with varicose veins on right foot, multiple erythematous, macular blanching lesions on right leg, and an irregular purple/reddish macular lesion that partially blanches present over lumbosacral area. Gait normal. Standing, right iliac crest is 2 inches superior compared to the left. Supine, his right medial malleolus was 1.5 inches inferior compared to left. Neurovascularly intact. No edema or cyanosis. Strength 5/5 throughout. The rest of his physical exam was within normal limits.

DIFFERENTIAL DIAGNOSIS:

Klippel-Trenaunay syndrome

Isolated hemihyperplasia

Lymphatic malformation

WORKUP:

Obtain records of ultrasounds/MRIs

WORKING DIAGNOSIS:

Klippel-Trenaunay Syndrome

TREATMENT/OUTCOMES:

There is no specific treatment for Klippel-Trenaunay syndrome or Isolated hemihyperplasia. As some conditions associated with hemihyperplasia are inherited, plan to refer to genetics. Isolated hemihyperplasia is associated with malignancy and screening is recommended for children with the disorder and may have relevance for future generations. Patients with Klippel-Trenaunay syndrome are at increased risk of DVTs, cellulitis, lymphedema and internal bleeding from abnormal blood vessels.