Klippel-Trenaunay Syndrome of the Upper Limb - A Rare Congenital Anomaly

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A 48 year old Indian male presented with a painless swelling of the right upper limb since birth associated with an overlying discolouration involving the right chest wall. At presentation, he complained of increased sweating and a tingling sensation over the involved limb. Clinical examination revealed a port wine stain over the right upper limb and hemithorax extending from the base of the neck to the T3 dermatome limited by the midline (Figure 1), with a hypertrophy of the ipsilateral limb (Figure 2). Examination was negative for digital anomalies, arteriovenous malformations and spina bifida. Based on these features, a diagnosis of Klippel-Trenaunay syndrome of the right upper limb was made.

The Klippel-Trenaunay syndrome, first described in 1900, is characterized by a triad of port-wine stain, varicose veins, and musculoskeletal hypertrophy, with only 63% of patients presenting with all three signs and any two of three features being sufficient for a diagnosis. Additional findings include spina bifida, hypospadias, polydactyly, syndactyly, oligodactyly, hyperhidrosis, hypertrichosis, and paresthesias. It generally involves a single extremity with the leg being the most common site affected. An exclusive upper limb involvement is rare and has been identified in 11% of patients. Diagnosis is essentially clinical. Major differentials include the Parkes-Weber syndrome, differentiated by the additional presence of arteriovenous fistulae and the Proteus syndrome. Complications include venous thrombosis, cellulitis, lymphedema, bleeding and Kassabach-Merritt syndrome. Treatment is mainly conservative and symptomatic.

References


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