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A RARE CASE REPORT OF KLIPPEL-TRENAUNAY SYNDROME

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ABSTRACT: *Klippel-Trenaunay syndrome has sometimes been used interchangeably with Klippel-Trenaunay-Weber syndrome. Klippel-Trenaunay syndrome is the term used for the triad of congenital anomalies. Klippel-Trenaunay-Weber syndrome or Parkes-Weber syndrome consists of the triad of Klippel-Trenaunay syndrome accompanied by a clinically apparent arteriovenous fistula. Hemodynamically insignificant arteriovenous malformations do not preclude a diagnosis of Klippel-Trenaunay syndrome. We report a case of term male neonate with clinical findings of Port-wine stain and hemi- hypertrophy of left side of body, consistent with Klippel-Trenaunay syndrome.*

Keywords: Klippel-Trenaunay Syndrome, Parkes-Weber syndrome, port-wine stain, vascular malformations, hypertrophies.

INTRODUCTION: ***K***lippel-Trenaunay syndrome (KTS) is a rare congenital condition usually presenting with port wine stains, excessive growth of bones and soft tissue and varicose veins which most commonly occurs in the legs, but it also may affect the arms, face, head, or internal organs. Not all cases have the full triad of features. There is wide variation in the clinical manifestations of the condition. Mostly, it is idiopathic in origin; however, sometimes it may occur as an autosomal dominant trait. Klippel Trenaunay Syndrome is estimated to affect at least 1 in 100000 people worldwide irrespective of sex and races.[1]The characteristic capillary haemangioma is visible from birth in the vast majority of cases (98% in one series).[2]

The vascular malformation is usually limited to a single extremity, though multiple extremities can be involved. Limb abnormalities may present initially as gait disturbances. The digits may be affected with macrodactyly, syndactyly, polydactyly or oligodactyly.

CASE REPORT: A 2 days male baby born of 22 years old Gravida 1, Para 1 presented to our SNCU, M.K.C.G Medical college and Hospital, with complaints of multiple red patches on left half of body with ipsilateral hemi hypertrophy .The baby was born at term by spontaneous vaginal delivery at a local hospital. On examination, the neonate had 4000 grams weight on admission with normal Vital signs. Multiple Port-wine stains (Fig 1) were found on left half of the body with same side hemi hypertrophy. (Fig 2) There was no cardiac murmur; no distended vessels, on investigation, thrombocyte count was normal and routine septic screen were within normal limit. ECHO was normal and cardiothoracic team concluded that the baby didn't need any vascular intervention. The neonate was clinically diagnosed as a case of Klippel Trenaunay Syndrome.

Fig.1. Showing port wine stains.

Fig 2 showing left sided hemi hypertrophy

DISCUSSION: Klippel-Trenaunay Syndrome was first described by two French doctors, Klippel and Trenaunay in 1900. It is a triad of capillary malformations, usually port-wine stains, soft tissue and bone hypertrophy and varicose veins or venous malformations (Klippel & Trenaunay, 1900), [3] Port-wine stains may be present at birth. These vascular malformations consist of mature dilated dermal capillaries. These lesions are macular, sharply circumscribed, and pink to purple in color and tremendously variable in size. Port-wine stains can occur as a component of Klippel-Trenaunay-Weber Syndrome associated with soft tissue enlargement (i.e. hypertrophy of an extremity or a part of it) [4].

In this case, upper limbs are hypertrophied and port-wine stains are present. There may be varicose veins, in this case no such findings were found on physical examination. There may be some rare complications, e.g. thrombophlebitis, dislocation of joints, gangrene of the affected extremities, heart failure, hematuria secondary to angiomatous involvement of urinary tract, rectal bleeding from lesions of gastrointestinal tract, pulmonary lesions and malformation of lymphatic vessels.[5] No such complication was found in this case.

Although the cause of KTS is still unknown, it is hypothesized that it is caused by a mesodermal abnormality during fetal development leading to vascular and soft tissue malformations in the affected limb (Baskerville et al, 1985). Mc Grory&Amadio(1993) believed that an underlying mixed mesodermal and ectodermal dysplasia was responsible for development of KTWS. Klippel Trenaunay Syndrome might develop due to a single gene defect. Rarely it can be inherited as an autosomal dominant trait. KTW syndrome associated with a reciprocal t(5;11)(q13.3;p15.1) or the de novo translocation t(8;14) (q22.3;q13). The association between the angiogenic factor gene AGGF1 and KTS appears to be significant.[6]

CONCLUSION:

In many patients, a thorough medical history and physical examination are sufficient to make the diagnosis. However, a number of imaging studies are useful when there are complications. There is no curative therapy Management requires a multidisciplinary and individualized approach, aiming to ameliorate the patient's symptoms and correct the consequences of limb-length discrepancy.

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