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Case Report

Klippel Trenaunay Syndrome in a 5 Year Old Boy: A Rare Embryological Vascular Anomaly

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ABSTRACT

We present a case history of 5 years old boy, born of a non-consanguineous marriage with uneventful vaginal delivery brought to OPD with complaints of limb length discrepancy where the length and girth of right leg was greater than the left leg, dilated veins over right inguinal area and port wine stain over right half of the body for 4 years, the condition progresses with progressing age of child. There was no history of ulceration, Raynauds phenomenon, paresthesia, difficulty in walking or pedal edema. Baseline investigations, Color Doppler, and X-ray lower limb were normal. Klippel trenaunay syndrome is a vascular malformation disorder comprising a triad of port wine stain, limb hypertrophy and varicosities with a global incidence of 5 cases/100000.

Keywords: Klippeltrenaunay syndrome; PIK3CA-related overgrowth spectrum; Phlebotomy; Port Wine Stain

Abbreviations: PIK3CA: Phosphatidylinositol 3-Kinase Enzyme; PROS: PIK3CA-Related Overgrowth Spectrum

INTRODUCTION

Klippel trenaunay syndrome is a congenital vascular malformation disorder comprising a triad of port wine stain, limb hypertrophy and varicosities with a global incidence of 5 cases/100000 populations, with no sexual or racial predilection. Etiological factors include-translocation at t (8;14) (q 22.3;q13) chromosomes, [1] and PIK3CA gene (phosphatidylinositol 3-kinase enzyme) mutation, where PIK3CA is responsible for sending chemical signals for cell proliferation, migration, and survival. Mutation in PIK3CA gene allows cells to grow and divide unchecked, which leads to abnormal growth of the bones, soft tissues, and blood vessels [2].

Klippel Trenaunay syndrome refers to a rare congenital anomaly which is characterized by capillary malformation, venous malformation and sometimes lymphatic malformation associated with overgrowth of a limb, with soft tissue hypertrophy and/or bony hypertrophy. The anomal generally presents since birth and usually involves the lower limbs as well as portion of trunk, face, upper limb or head [3]. We report a case of a 5 year old boy having port wine stain, varicose veins and excessive growth of soft tissue of right lower limb clinically consistent with Klippel Trenaunay Syndrome.

CASE REPORT

A 5 year old boy, born of a non-consanguineous marriage and delivered at full term by uneventful vaginal delivery, presented to skin outpatient department with complaints of limb length discrepancy where the length and girth of right leg was greater than the left leg, dilated veins over right inguinal area and right side of trunk, and cutaneous pigmentation over right half of the body since 4 years, the condition progresses with age and anthropometric development of child. There was no history of ulceration over limbs, Raynauds phenomenon, paresthesia, difficulty or pain while walking and pedal edema.

On examination-multiple superficial varicose veins were present over right inguinal area and right side of trunk,(Figure 1) hyperpigmented to erythematous raised plaque (Port Wine Stain) was present in right half of the body (Figure 2) excluding face and neck, limb length discrepancy was found as on measurement the length of left lower limb came out to be 41 cm as compared to 44 cm of right limb (3 cm difference)(Figure 3) Baseline investigations, Color Doppler, and X-ray of lower limbs were normal.

Based on clinical and radiological imaging findings, a diagnosis of Klippel Trenaunay syndrome was made.

The attendants were counselled about the progressing course and the possible complications of the disease. An orthopedician reference was taken; compression stockings were prescribed to check overgrowth of the right lower limb. Parents were explained about the possible treatment plans in future (ND YAG, Pulsed dye lasers) for the pigmentary component of the disease after he

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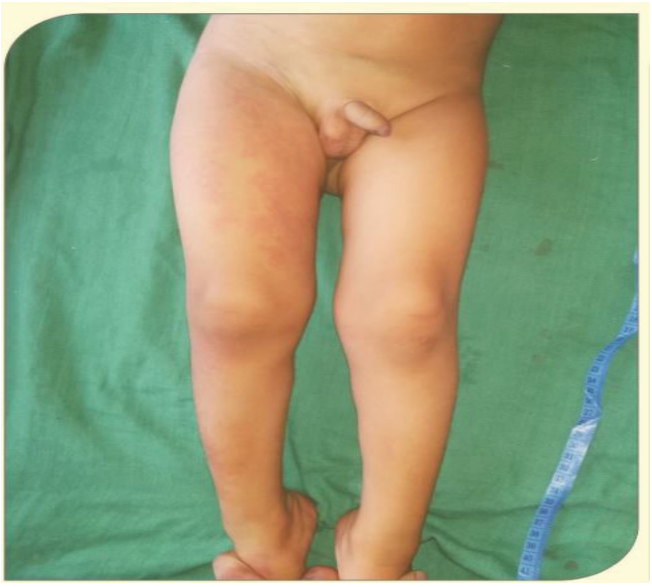

Figure 1: Showing multiple dilated veins over right inguinal area.

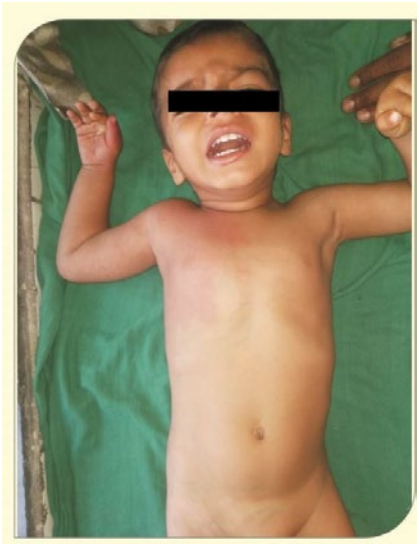

Figure 2: Showing erythematous cutaneous pigmentation 'Port Wine Stain' over right half of the body.

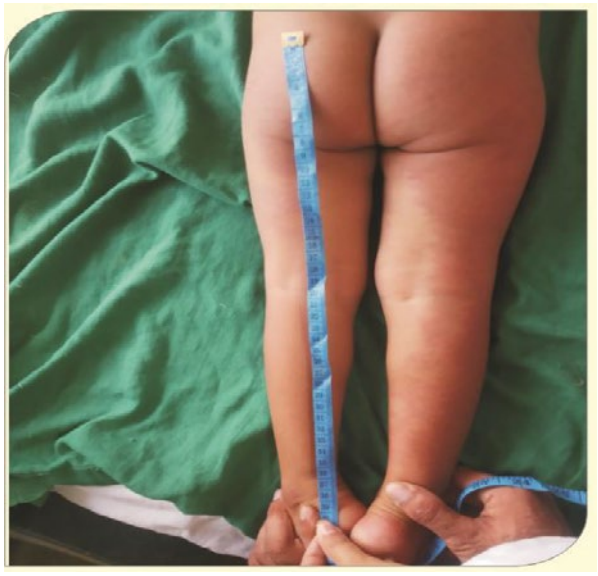

Figure 3: Showing limb length discrepancy where right lower limb is hypertrophied (increased length and girth) in comparison to left lower limb.

had attained full growth of the limbs and once the port wine stain component also gets stabilised.

RESULTS AND DISCUSSION

Klippel-Trenaunay-Weber syndrome is a rare congenital medical condition in which blood vessels and/or lymph vessels fail to form properly. The three main features are naevusflammeus (port-wine stain), venous, lymphatic malformations and soft-tissue hypertrophy of the affected limb. Classified on the basis of clinical features present into-Typical-All features are present and Atypical-All features are present except port-wine stain [4].

Klippel-Trenaunay syndrome is one of several overgrowth syndromes, including megalencephaly-capillary malformation syndrome that are caused by mutations in the PIK3CA gene. Together, these conditions are known as the PIK3CA-Related Overgrowth Spectrum (PROS) [5].

Port Wine Stain-is a birthmark caused by swelling of small blood vessels near the surface of the skin. They are typically flat and can vary from pale pink to deep maroon in color, usually covering part of one limb. The affected area may become lighter or darker with age [6].

Overgrowth of bones and soft tissues beginning in infancy, limiting to one limb, most often one leg but overgrowth can also affect the arms or torso. The limb length discrepancy may cause difficulty in conducting daily activities later in life like pain, a feeling of heaviness, reduced movement in the affected area and problems with walking [7].

The third major feature-vein malformations varicose veins-swollen twisted veins that appear superficially, present on the sides of the upper legs and calves. Sometimes deep veins may also get involved which in inturn increase the risk of Deep Vein Thrombosis (DVT) [8-10].

CONCLUSION

Other complications of Klippel-Trenaunay syndrome include limb cellulites, lymphedema and blister formation or internal bleeding from port wine stain. Less commonly, syndactyly or polydactyly may be present. Aneurysms in medium and large vessels are associated most common being abdominal aortic aneurysm.

Diagnostic tests include Multidetector row computed tomography arteriography; color Doppler, MR angiography, contrast venography and arteriography, and nuclear MRI.

Treatment-Compressive therapy-compressive bandages can be offered to prevent high output flow, edema and hypertrophy of soft tissues which is a beneficial preventative measure in children. For varicose vein-sclerotherapy can be offered. Once the naevusflammeus component gets stabilised-ND:Yag laser, pulsed dye laser could be offered. Surgical Debulking of the discrepent limb can be done to improve the quality of life.

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