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Klippel Trenaunay and Proteus Syndrome overlap--a diagnostic dilemma

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Abstract:

Klippel Trenaunay syndrome is a rare mixed vascular malformation characterized by a triad of port wine stain, varicose veins, and bony, soft tissue hypertrophy involving an extremity [1]. A rare case involving face and all the limbs is being reported. This is a case of interest also because of having clinical overlap with Proteus syndrome.

Introduction

The exact cause of Klippel-Trenaunay syndrome remains to be elucidated, although several theories exist like intrauterine damage to the sympathetic ganglia or intermediolateral tract, or deep vein abnormalities, or a mesodermal defect during fetal development. Most cases are sporadic, although a few cases in the literature report an autosomal dominant pattern of inheritance [2]. This syndrome is characterized by a triad of extensive capillary malformation, underlying venous varicosities, and underlying soft-tissue or bony hypertrophy. The port-wine stain is typically of the nevus flammeus type. Other vascular malformations can be angiokeratomas, angiodermatitis or even lymphangioma circumscriptum [3].

The lower limb is the site of malformation in the majority of patients. The hypertrophy involves the length as well as the circumference of the involved extremity and is caused by local hyperemia and venous stasis secondary to the vascular anomaly [4].

Proteus syndrome is a hamartomatous disorder which is characterized by asymmetrical overgrowth of any part of the body, verrucous epidermal naevi, vascular malformations and lipoma like subcutaneous hamartomas. Macrodactyly has been regarded characteristic. Often, cerebriform thickening of the soles and palms is also present [5].

Case report

A 20 years old female born out of non- consanguineous parents presented with a history of hemihypertrophy of left side of face, tongue and also disproportionate hypertrophy of all the limbs, more pronounced on left side since birth. She also had history of presence of Macrodactyly of left middle finger; port wine stain over the trunk, and all the limbs which did not show any change in colour till now. Patient also had venous varicosities over both the lower limbs as well as left upper limb since early childhood. Since last two years the patient had developed grouped vesiculo-papular lesions on left lower leg with history of frequent bleeding from them. There was no family history of such lesions and no history of any other significant illness in patient.

On cutaneous examination hemihypertrophy of the left side of face including the palate and tongue was present with a linear central depression over the left side of forehead.

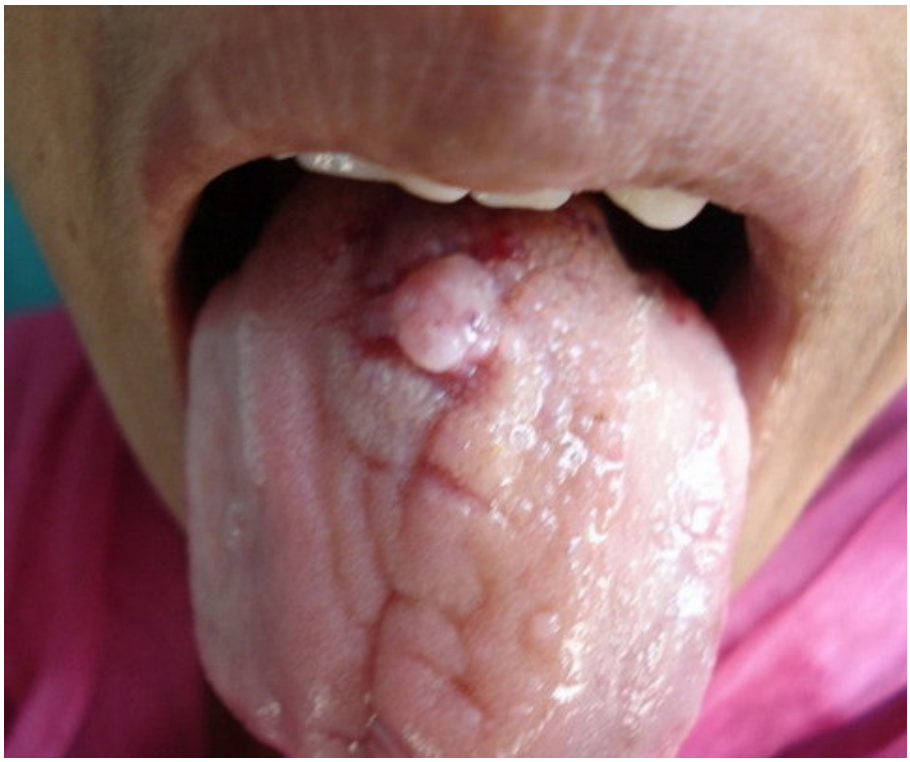

Fig 1: Showing hemihypertrophy of the tongue.

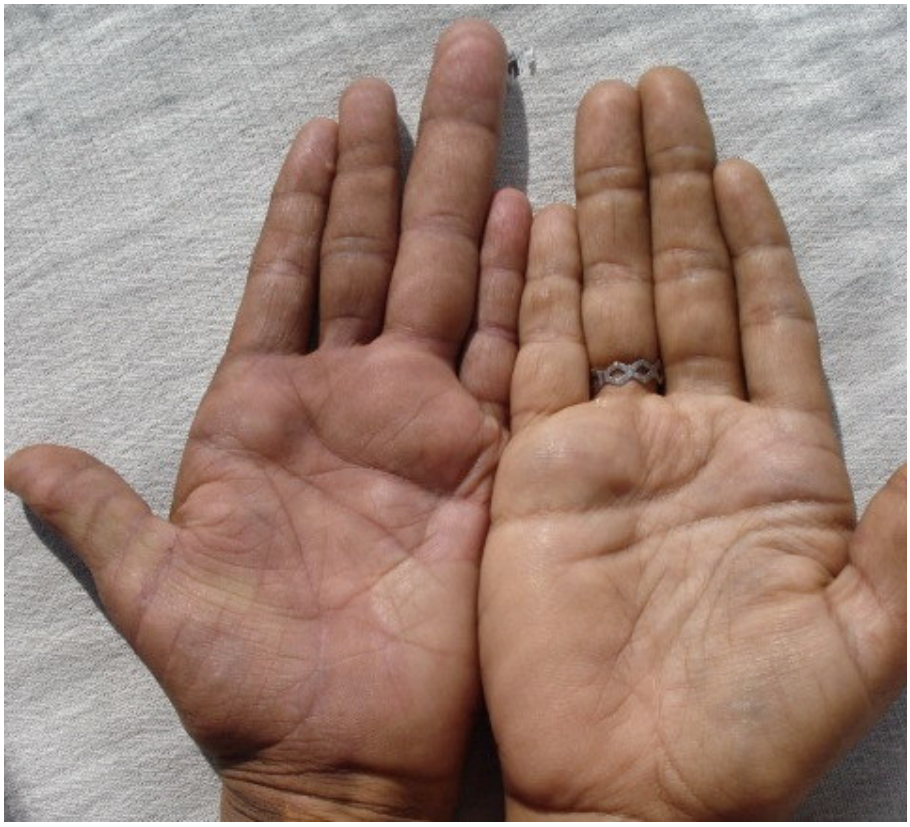

Fig 2: Showing macrodactyly of left middle finger.

Tongue showed deep furrowing on left side and a central 2cm x 2cm nodular swelling **{figure-1}**. Hypertrophy of both upper and lower limbs was present more pronounced on the left side. Left little finger showed macrodactyly **{figure-2}**.

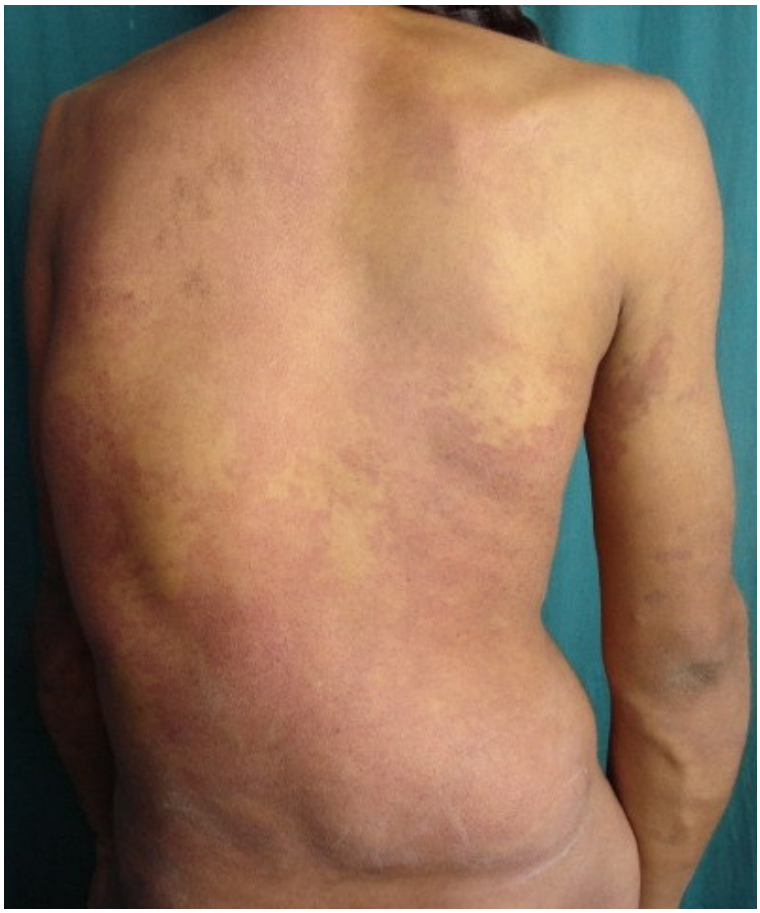

Fig 3: Port wine stain on back and scoliosis.

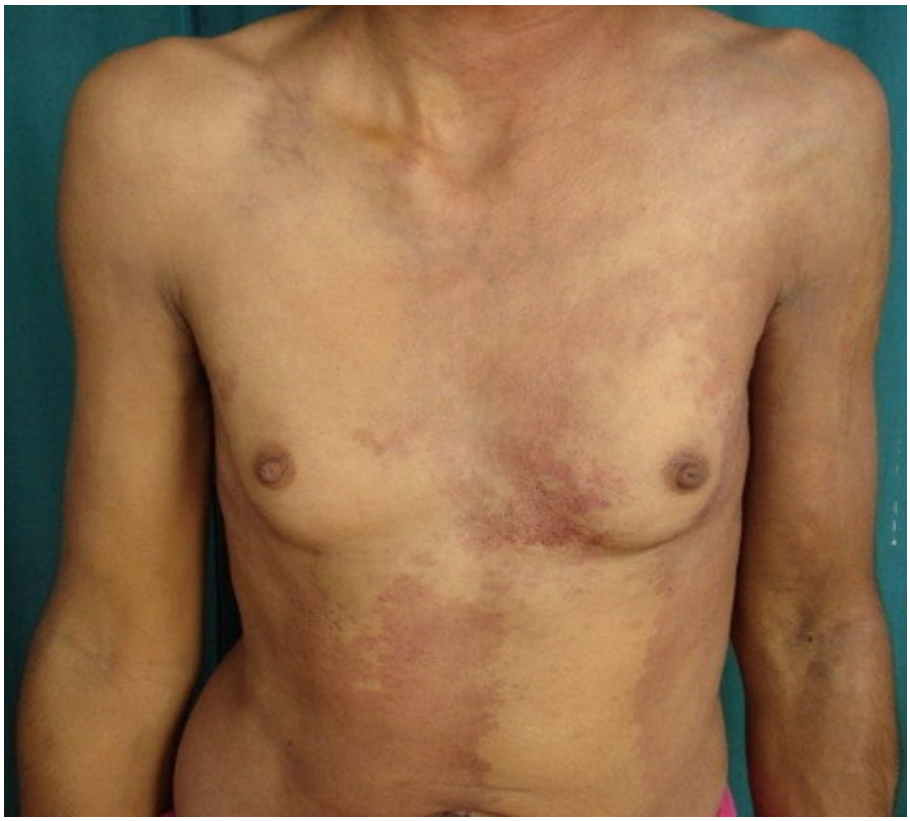

Fig 4: Showing extensive port wine stain.

Widespread port wine stain was present over the neck, trunk, and all the limbs and also extending over the left palm **{figure 3, 4}**. Dilated tortuous veins were present on all the four limbs. Multiple hyperpigmented grouped papulovesicular growths were present on left lower legs **{figure-5}**. Similar growths were diffusely present over the left popletial fossa, dorsum of left foot and also the ankle. Both the dorsum of feet showed hyperpigmented diffuse velvety plaques extending over the toes **{figure-6}**.

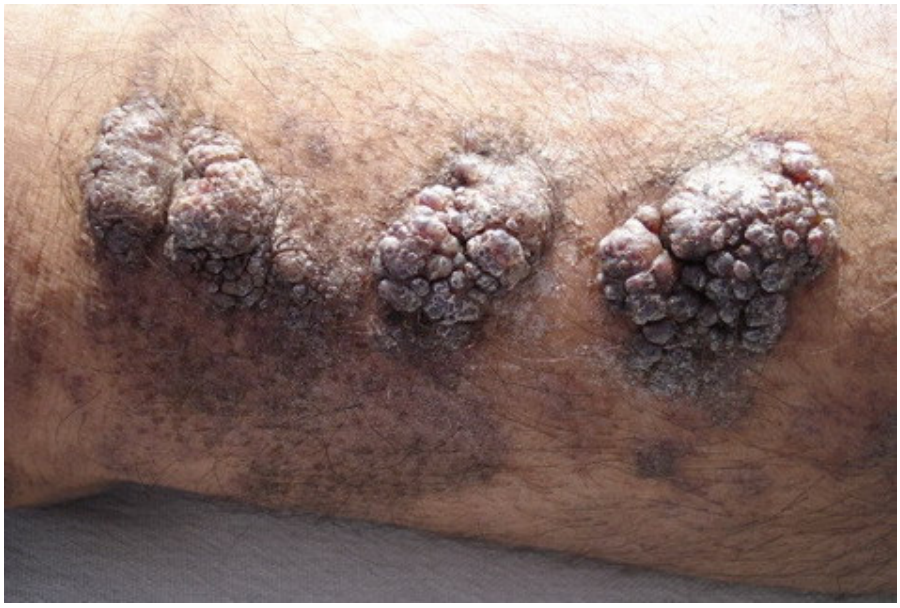

Fig 5: Showing lymphangioma circumscriptum.

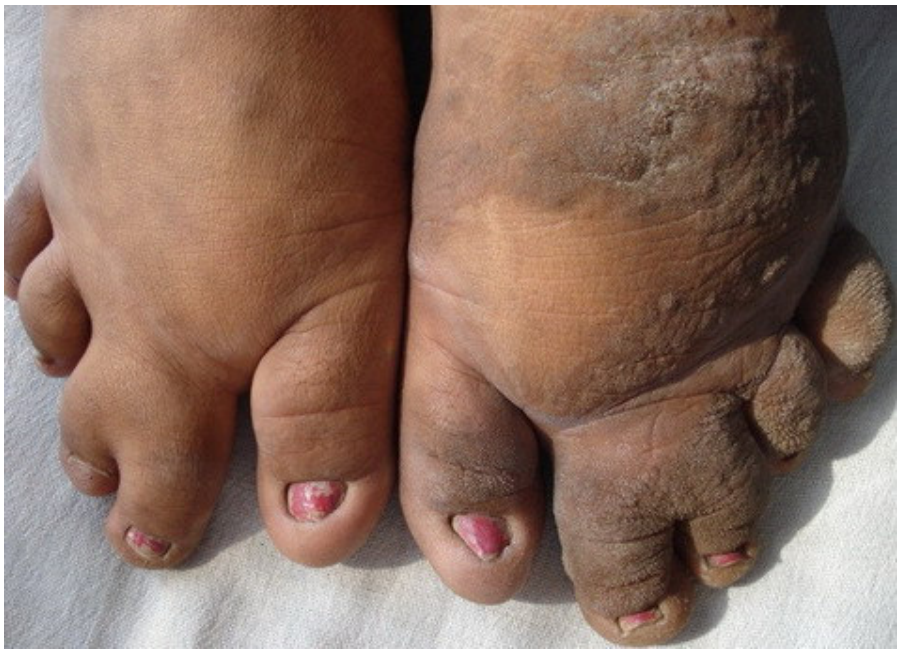

Fig 6: Showing syndactyly of 2nd and 3rd toes and velvety plaques.

Both the lower legs showed non pitting type of oedema. Other associated features were scoliosis to left, syndactyly of 2nd and 3rd toes of both the feet **{figure-3,6}**. All routine investigations were in normal limits. X ray both feet confirmed the syndactyly and showed overgrowth of 2nd left metatarsal. X ray both hands showed overgrowth of left ring finger phalanges. Colour Doppler both lower limbs showed varicosities and sapheno-femoral incompetence.

Biopsy of the papulovesicular lesions suggested the presence of lymphangioma circumscriptum **{figure-7}**.

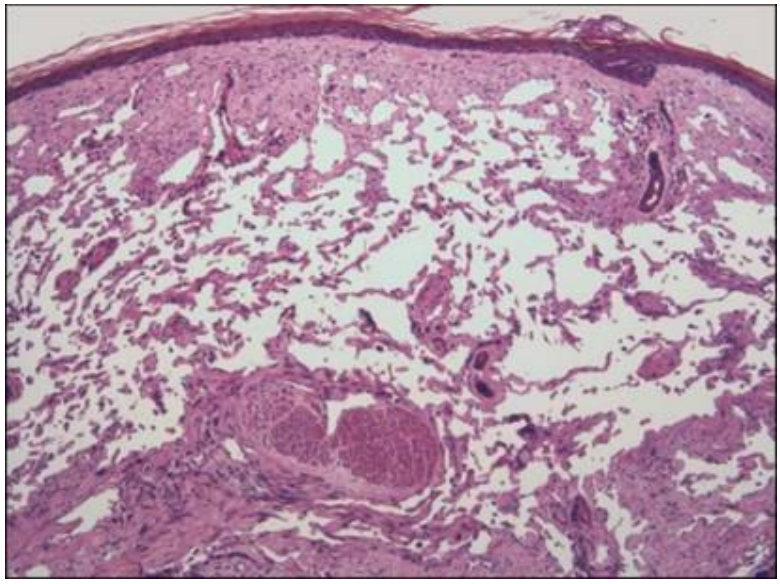

Fig 7: Showing histopathology suggestive of lymphangioma circumscriptum.

Discussion

In Klippel Trenaunay syndrome characteristic cutaneous lesions comprise of port wine stain which are usually present at birth. They occur on the affected limb but may sometimes extend beyond it involving several limbs. They have a tendency to stop in the midline. Various other rare vascular malformations include- small angio-keratomas, angiodermatitis, lymphangioma circumscriptum, lymphoedema. The hypertrophy present is of more commonly legs than arms and involves the growth of soft tissue and the bone. Rarely involvement of the face may be seen. Compensatory scoliosis is present when there is a leg length difference. Venous varicosity of the affected limb is a common presentation which often presents in the early childhood. There is a high risk of deep vein thrombosis to occur. Other associated features include- verrucous epidermal naevi, polydactyly, syndactyly [5].

Proteus syndrome comprises characteristically of asymmetrical hypertrophy of face, part or whole of one or both limbs, trunk. Macrodactyly is another characteristic feature. Soft tissue growth over the palms and soles in a cerebriform pattern is found [6,7]. The three main types of skin findings include- epidermal naevi which is

generally of linear verrucous variety, vascular malformations which are similar to Klippel Trenaunay syndrome, and soft subcutaneous masses which are highly characteristic and may represent complex hamartomatous malformations. Other associated features include scoliosis, kyphosis, cataract, strabismus, hypodontia with normal intelligence [5].

Our patient whose clinical features are summarized in **Table -1** exhibits the characteristic feature of hemihypertrophy of limbs a feature shared by both the syndromes but involvement of face and all the limbs favours Proteus syndrome. Presence of port- wine stain is again a feature common to both but its extensive involvement is a rarer presentation in Klippel Trenaunay syndrome than in proteus syndrome. Varicosities is present in both syndromes but more so in Klippel Trenaunay syndrome. Macrodactyly is a characteristic feature suggesting Proteus whereas presence of syndactyly and lymphangioma circumscriptum suggested more likely a possibility of Klippel Trenaunay Syndrome. Absence of verrucous naevi and sub cutaneous lipoma like nodules negated Proteus syndrome.

Clinical feature	Proteus Syndrome	Klippel Trenaunay syndrome
Facial hypertrophy	++	+
Hypertrophy of limbs	++ But involvement of many limbs is commoner in Proteus	++
Port wine staining	+ But extensive involvement is commoner here.	++
Varicosities	+	++
Lymphangioma circumscriptum	+	++
Macrodactyly	++	+
Syndactyly	+	++

Table 1. Summary of features of the patient comparing Proteus and Klippel Trenaunay syndrome
+ = may occur,
++ = characteristic of the condition

Conclusions

The present case is, hence, being reported because of its rarity & overlapping of features of both the syndromes i.e. Klippel Trenaunay and Proteus Syndrome in the same patient thereby creating a diagnostic dilemma.

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