

Case Report

KLIPPEL–TRENAUNAY SYNDROME (KTS): A CASE REPORT

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ABSTRACT

Klippel- Trénaunay syndrome (KTS) is an uncommon entity. This congenital malformation is characterized by the triad of soft tissue or bony hypertrophy, cutaneous vascular malformations, and atypical venous abnormalities. Only 5% of cases of KTS involve the head and neck region. We report here a case of KTS affecting the maxillofacial region and discuss the clinical features and differential diagnoses of these cases.

Keyword: Klippel–Trenaunay Syndrome

INTRODUCTION

Klippel–Trenaunay syndrome (KTS) is a congenital disorder characterized by triad of vascular nevi, venous varicosities and hyperplasia of soft and hard tissues in the affected area (Terezhalmay *et al.*, 2000). Alternative names given for Klippel- Trenaunay Syndrome are Klippel-Trenaunay-Weber syndrome; Angio osteohypertrophy; Nevus varicosus osteohypertrophicus syndrome; Hemangiectasia hypertrophicans and Nevus verucosus hypertrophicans.

Lesions in KTS are associated with capillary fragility and tend to bleed easily (Ita *et al.*, 2001). In addition to capillary fragility, platelet dysfunction, clotting and fibrinolytic abnormalities can also occur (Ita *et al.*, 2001). The purpose of reporting this case is to emphasize the importance of complete preoperative hematological tests in patients with KTS to avoid possible postoperative bleeding problems.

CASES

A 12 year old female patient reported with a chief complaint of red patch on the face since birth. Her parents had noticed the presence of a red discolored patch on his right cheek at the time of her birth. She gave the history of frequent bleeding from nose. The patient was apparently asymptomatic but since last 3 month she felt that left side of his face was growing faster than his right side. While taking history from patient, hoarse of voice was noted. She was from lower socio-economical strata.

On extra Oral examination, the port wine stain was noted on the left side of face. It was extended supero-inferiorly from left hair line on fore head to the imaginary horizontal line passing 2 cm above the inferior border of mandible and antero-posteriorly from midline of face to vertical line passing through posterior border of mandible. Left half sides of upper and lower lips were also affected with the lesion. A well demarcated line was seen at the center of vermilion border of the upper lip. On palpation, blanching of the patch on digital pressure had also noted. There was local rise in temperature with that patch and skin over the patch smooth with no scar marks. The margins of the patch were well demarcated and smooth (Figure 1).

There was additionally most obvious sign of hemi facial hypertrophy of left side of mandible resulting in facial asymmetry with deviation of chin towards right side. The lip line was also slanting towards the left side due to deviation of chin. Hypertrophy was also evident in the lips and cheeks of affected side (Figure 1).

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Figure 1: Port wine stain on left side of face with deviated chin to right side

Intraoral examination revealed port wine hemangiomas affecting mucosal surface of left side of upper lip which was well demarcated at midline from normal right side of lip (Figure 2A). Similar port wine stain was present with left half side of palate separated from right side at the midline (Figure 2B). Dilated varicose superficial veins were noted at the ventral surface of tongue (Figure 3A). Left side of alveolar processes appears to be hypertrophied with premature eruption of the 27, 37 & 33 were seen as compared to 17, 47, 43 (Figure 3B). The temporal difference in the dental development between two sides was 2-3 years.



Figure 2A: Hemangioma involving left side of upper lip with line of demarcation in center

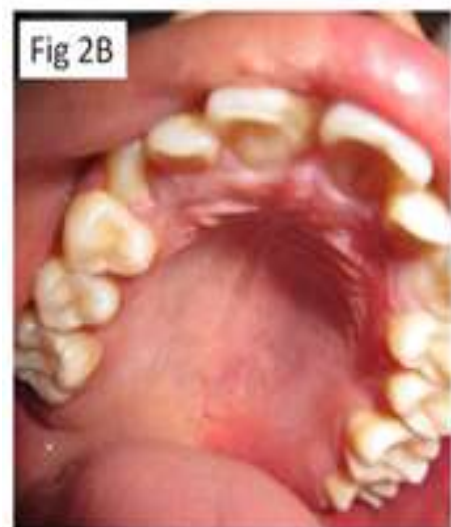


Figure 2B: Hemangioma involving left side of palate with line of demarcation at midpalatine raphe

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Figure 3A: Lingual Vericocities



Figure 3B: Difference in development of 33 & 43, as 33 has complexly erupted compared to 43

Radiographic examination with the OPG confirmed the temporal difference in development of 17 & 27 and 37 & 47. There was also temporal difference between the development of 38 & 48. The enamel formation of 48 has almost complete whereas the development of 38 had just to be started (Figure 4). Spacing between the teeth with labial tilting of the incisors was observed and also his upper incisors.



Figure 4: OPG revealing differences in development of 17 & 27, 37 & 47, 38 & 48

On the basis of classical features of hemi-facial hyperplasia of soft and hard tissues, a cutaneous vascular malformation and associated, a final diagnosis of KTS was made

DISCUSSION

Klippel-Trenaunay syndrome was first reported in 1900 by French doctors, Klippel and Trenaunay (Bathi RJ *et al.*, 2002). KTS is characterized by vascular malformations, venous/lymphatic varicosities and bony and soft tissue hypertrophy of the affected area. Capillary Hemangiomas are the most common type, which are called port wine stains due to its red and purple color and are often apparent at birth or by second week of age (Samuel & Spitz 1997). Diagnosis is based upon the presence of any two of these three features (Terezhalmay *et al.*, 2000).

It is hypothesized that it is caused by a mesodermal abnormality during fetal development leading to vascular and soft tissue malformations in the affected part of body (Baskerville *et al.*, 1985). McGrory & Amadio (1993) believed that an underlying mixed mesodermal and ectodermal dysplasia was responsible for development of KTWS. Happle (1993) had stated that Klippel-Trenaunay Syndrome might develop due to a single gene defect. Rarely, it can be inherited as an autosomal dominant trait (Ceballos-Quintal *et*

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al., 1996). Whelan *et al.*, (1995) reported a case of a girl with KTW syndrome associated with a reciprocal translocation: t (5;11) (q13.3;p15.1). The de novo translocation t (8;14) (q22.3;q13) has also been reported (Wang *et al.*, 2001). The association between the angiogenic factor gene *AGGF1* and KTS appears to be significant (Hu *et al.*, 2008).

However there can be some degree of variability regarding the localization and the extent of hyperplasia. In KTS there occurs a characteristic unilateral increase in dimensions of both hard and soft tissues, particularly involving the lips, cheeks, tongue and teeth. In our patient, the majority of these features were present but the tongue was normal in size but there was presence of venous varicosities on ventral surface of tongue. The possibility of hypertrophy of tongue in future age of life can't be denied.

Klippel-Trenaunay syndrome is characterized by an extreme degree of variability when affecting the craniofacial region, with the majority patients not exhibiting the classical triad of signs. The greatest variability pertains to vascular anomalies, which can range from cutaneous hemangioma to arterio-venous malformations (Ita *et al.*, 2001). The hyperplasia can be attributed to increased vascularity resulting from abnormal vascular development (Ita *et al.*, 2001). In our case also the vascular malformation and hyperplasia were observed on the same side. In addition to the classical features of KTS, associated additional findings like abdominal hemangioma, heart defects, syndactyly, polydactyly, oligodactyly and macrodactyly are also reported in the literature (Bathi *et al.*, 2002, Steiner *et al.*, 1987).

The development of features in KTS during the pubertal phase is rapid and the changes do not progress after puberty (Bathi *et al.*, 2002). Our patient was a teenager who had recently started with a pubertal growth spurt, which explains the rapid progression of the swelling.

Only 5% of cases of KTS involve the head and neck region (Bathi *et al.*, 2002). It is difficult to explain the reason for the segmental involvement of the face in KTS with unilateral enlargement of the maxilla and dentoalveolar region with normal growth of contralateral components (Ita *et al.*, 2001), as observed in our patient. The bleeding time and clotting time of the patients were with normal limits, so the recurrent epistaxis could be due to local factors.

Klippel-Trenaunay syndrome should be differentiated from other congenital vascular anomalies. When Klippel-Trenaunay Syndrome is associated with arteriovenous fistula, it is known as Klippel-Trenaunay-Weber Syndrome (Weber 1907). Parkes-Weber syndrome has similar features in addition to arterio-venous fistula (Bathi *et al.*, 2002). In Sturge Weber syndrome there are hemangiomas along the distribution of the trigeminal nerve with focal seizures, sensory and motor paralysis, calcifications of vessel walls and visual field defects (Bathi *et al.*, 2002). Our patient had none of these features.

The facial asymmetry and esthetics are the major concerns of the patient and that requires an interdisciplinary team approach between a dentist, an oral maxillofacial surgeon, plastic surgeon and an orthodontist who have to formulate a coordinated plan in the management of such patients. In all patients with KTS, even without craniofacial involvement, a dentist must advise detailed hematological investigations prior to any oro-surgical procedure to prevent a possible postoperative bleeding problem. Even these patients are not treated; they should be regularly followed up for every 6 month of interval.

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