

Klippel-Trenaunay-Weber syndrome: literature review and case report

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Abstract

Klippel-Trenaunay-Weber syndrome is characterized by limb hypertrophy, varicose veins, and vascular nevus. The orofacial manifestations include early eruption of permanent teeth and hemifacial hypertrophy. This 5-year-old male patient had facial asymmetry, limb abnormalities, and a thumb-sucking habit. Cephalometric analysis revealed a Class II open bite occlusion. (Pediatr Dent 16:231–35, 1994)

Introduction

Klippel-Trenaunay-Weber syndrome (KTW) (angio-osteohypertrophy) was first reported by Klippel and Trenaunay in 1900.¹ Approximately 900 cases have been reported thus far, most of them in the European literature. Orofacial findings and dental management have been described infrequently.

The original description of KTW syndrome included limb hypertrophy, varicose veins, and vascular (port-wine) nevus, which were characterized as a clinical triad. Hemangiomas is the most frequent finding in these patients and is usually present at birth.² The vascular lesions may be bilateral or unilateral and can be present in the oral cavity or on the trunk, buttocks, limbs, head, and/or neck. Nevus flammeus, congenital arteriovenous aneurysm, cutaneous and subcutaneous capillary hemangioma, cavernous hemangioma, varicosities, and phlebectasia have been included in the vascular lesions of this syndrome.³ These findings may also appear in patients with Sturge-Weber syndrome, causing diagnostic confusion.⁴ The occurrence of both syndromes in the same patient has also been reported.⁴

Hypertrophy is another clinical finding that usually is present at birth, but may occur at any age and usually increases with age.² Hypertrophy can be of the soft and/or hard tissues and usually results in facial or extremity asymmetry. The hypertrophy can be contralateral or bilateral (e.g., legs) and occurs ipsilateral to hemangiomatotic skin lesions.^{2,3} Osteohypertrophy may lead to increased girth and/or length of the extremities. Rarely, an increase in the size of proximal or distal ends of limbs has been observed.² Infrequently the involved limb may atrophy rather than hypertrophy. Atrophy of soft tissue and/or underlying bones has been reported by Mullins.³ Macrodactyly, polydactyly, syndactyly, and oligodactyly can occur.²

Lindenauer⁵ reported 18 cases of KTW syndrome, and of these, nine exhibited an increase in both length and girth of the involved limbs, five increased length or girth of the limb extremities, two smaller limb length or girth, and one smaller limb length and girth on the

affected side. The remaining patient demonstrated normal limb size with hemangioma and varicosities.⁵

Contralateral convulsion and mental retardation have been reported in KTW syndrome.^{2,4} Other associated features reported include dry, scaly skin,¹ congenital dislocation of hip or shoulders, spina bifida, and scoliosis, among others.⁶

Vascular involvement of the internal organs can contribute to additional anomalies and dysfunctions.⁷

The etiology of KTW syndrome is still unknown, but Koch suggested an autosomal dominant form of inheritance.⁸ Others have excluded familial inheritance and propose sporadic occurrence.^{2,9,10} In their report You, et al. review the opinion of most authors that an intrauterine insult between the third and sixth week of gestation at the time of vascular differentiation and invasion of the limb bud is the cause.⁶ This may be substantiated by some of the mesodermal patterns observed.⁶ Others suggest injury to the sympathetic ganglia or intermediate lateral tract may account for the pattern of anomalies.^{6, 11, 12} Venous stasis and inherited disorders of phakomatosis have also been suggested.⁶ There is no apparent gender predilection.

Little has been written about the oral and maxillofacial manifestations of this syndrome.¹⁰ Petschelt reported that 90% of cases involve the upper and lower extremities and 10% the head and/or trunk.¹³ Gloviczki, et al.¹⁴ suggested 5% of cases involve the head and neck. Meischer reported that three-fourths of cases involving the facial region presented with oral changes—mostly nevus flammeus.¹⁵ Early tooth eruption, gingival hyperplasia, tongue and soft tissue overgrowth, and jaw bone osteohypertrophy causing malocclusion have been recognized.^{2, 4, 16} Some patients may have hemangioma unilaterally on the gingiva, palate, pharynx, and/or tongue.¹⁰ Intraoral cyst formation and intraosseous masses, mucosal ulcers, and sebaceous and salivary gland hypertrophy have also been reported.^{1, 17–19}

There has been confusion between KTW syndrome and Parkes-Weber syndrome.²⁰ Lindenauer proposed that KTW syndrome presents with varicosity, hyper-



Fig 1. A 5-year-old boy with KTW syndrome. Note slight facial asymmetry.

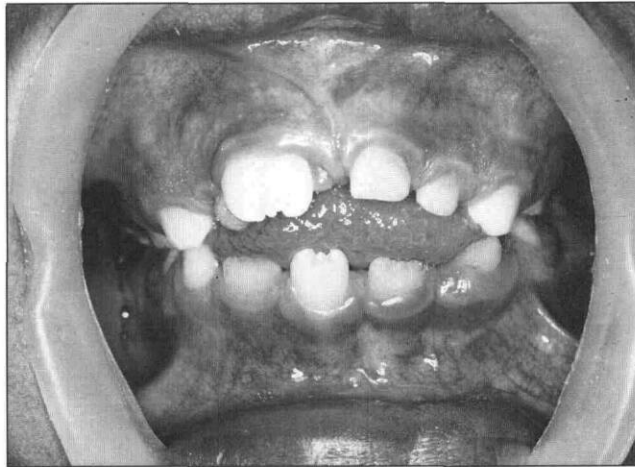


Fig 2. Early eruption of permanent teeth. Anterior open bite with tongue thrust. A granuloma-like soft mass was located at the distal-lingual surface of tooth #9.

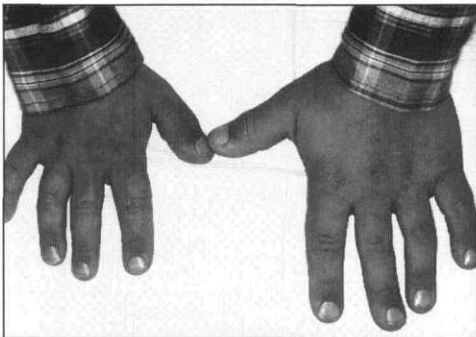


Fig 3. Asymmetric osteohypertrophy on the right hand. Note hemangiomatic lesions on the left hand.

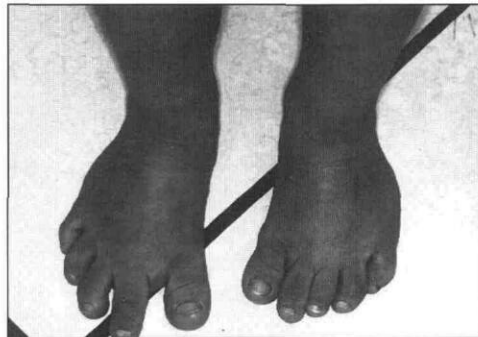


Fig 4. The left leg was larger than the right. Note the elongated toes on the left foot.

trophy, and hemangioma without arteriovenous fistula, while Parkes-Weber syndrome presents with a congenital arteriovenous fistula in association with hemangiectatic hypertrophy or phlebarteriectasis.⁵ In contrast, Sturge-Weber syndrome usually consists of craniofacial angiomatosis, port-wine nevus, and cerebral calcification. These features distinguish it from KTW syndrome. The syndrome originally described by Klippel and Trenaunay in 1900 should be considered a specific entity.

Case report

Chief complaint and history of present illness

A 5-year-old Caucasian male sought examination at the Children's Clinic of the University of Alabama School of Dentistry with a chief complaint of an anterior open bite. This was the patient's first dental visit.

KTW syndrome had been diagnosed at birth. At that time, the right hand was larger than his left and the left leg was larger than the right. Both legs were diagnosed as genua varum (bowed and twisted inwardly). A cutaneous and subcutaneous hemangioma was present throughout the left half of the body. The patient had worn an orthopedic appliance for three years

to correct his leg alignment. The patient's past medical history was otherwise unremarkable. Both siblings were healthy and without any characteristic findings of KTW syndrome. Parents reported an active thumb-sucking habit.

Physical examination

The initial evaluation revealed a 5-year-old male of average height with a large head, square muscular face, convex profile, large maxilla with spaces between upper teeth and an anterior open bite with tongue thrust (Figs 1 and 2).

The patient was friendly and communicative, though somewhat shy. His father stated that he was slow in learning. The patient appeared to have normal social and mental skills for his age.

General examination revealed an extensive nevus flammeus on the left half of the neck and

trunk and on the left arm, hand, leg, and foot. The left hand was of average size for the patient's age, but the right hand was larger (Fig 3). All five fingers of the right hand were uniformly large. His right foot was normal in size and no nevus flammeus was observed on it. His left foot was abnormally large with macrodactyly and elongated toes (Fig 4). The left portion of the face was slightly longer than the right side but without severe facial asymmetry.

Oral examination revealed no intraoral hemangiomas, nor soft tissue enlargement except for a pedunculated pink soft mass on the distal-lingual surface of tooth #9, which later proved to be an irritation fibroma. The maxillary arch appeared enlarged with spaces noted between all maxillary teeth. The anterior open bite measured 7.5 mm. Nine permanent teeth were clinically noted in the child's mixed dentition. The maxillary left central incisor had erupted, but the right central incisor had not and tooth F was retained. Oral hygiene was good and gingival inflammation was minimal. No carious lesions were noted.

Radiographic evaluation

A radiographic examination was performed and

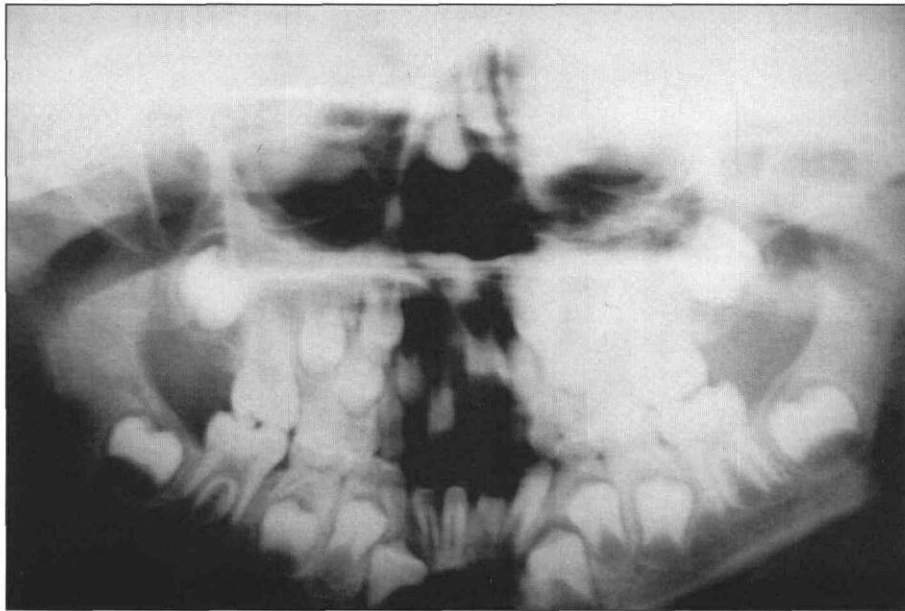


Fig 5. Panorex at age 5 years. Note early eruption of permanent teeth.

based on calcification and root formation, his dental age was estimated to be about 8 years (Fig 5).

Cephalometric analysis using the Modified Steiner, Ricketts, McNamara, and Moyer's analyses²¹⁻²⁴ (Figs 6 and 7; Table) indicated that the patient had a skeletal Class II pattern, skeletal open bite, longer anterior lower facial height, steep mandibular plane, and bimaxillary protrusion. The maxilla exhibited excessive horizontal growth while the mandible was of normal length. Facial asymmetry was noted in the A-P analysis with the left side 8 mm wider than the right (Fig 7).

Discussion

This case represents a variant example of KTW syndrome. The strict unilateral involvement of extensive nevus flammeus, cutaneous/subcutaneous hemangioma, and osteohypertrophy are consistent with this disease. However, the occurrence of osteohypertrophy with macrodactyly of the hand on the contralateral side is unusual. The English literature describing the orofacial findings of KTW syndrome is very sparse^{10, 15} (only three cases reported in the last 50 years). Cephalo-

metric analysis and longitudinal study of the craniofacial structure of these patients are lacking. This report attempts to address this information gap.

KTW syndrome is a rare disorder with very unusual findings. The three most common oral findings all have implications for the dentist. Early dental development and eruption necessitates orienting any dental treatment to the patient's dental rather than chronological age. Hypertrophy of the bone and/or gingiva could create severe malocclusions and present difficulties in designing and utilizing prostheses. Hemangiomatic areas that need any surgical procedures require extra attention regarding hemorrhage control.

Early eruption of the permanent teeth is a frequently reported orofacial manifestation of the syndrome.^{2, 4} Stellmach also reported premature eruption of a permanent central incisor on the side of maxillary enlargement.²⁵

Premature permanent tooth eruption may also occur in the absence of any underlying systemic disorder or can be associated with a variety of craniofacial syn-

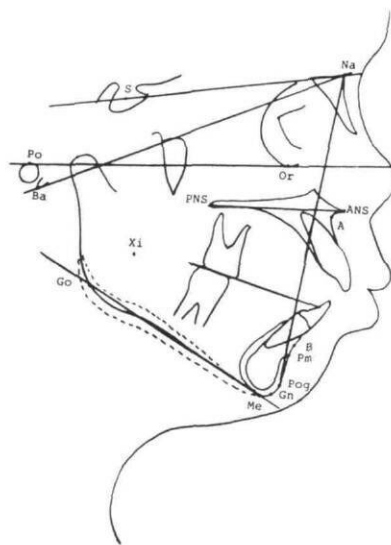


Fig 6. Lateral cephalometric tracing. Na, nasion; S, sella; Or, orbitale; Po, porion; Ba, basion; ANS, anterior nasal spine; PNS, posterior nasal spine; Xi, Xi point; A, point A; B, point B; Pm, supra pogonion; Pog, pogonion; Gn, gnathion; Me, menton; Go, gonion.

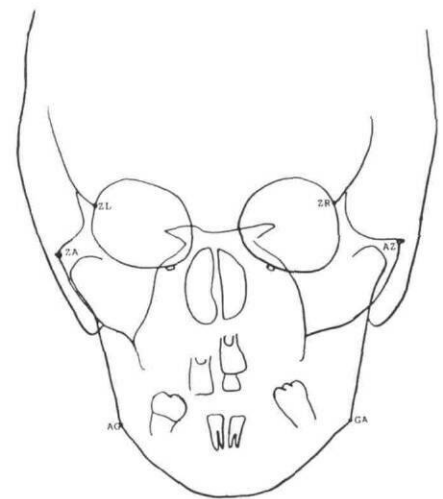


Fig 7. Posteroanterior cephalometric tracing. ZL(ZR), zygomatic bilateral points on the medial margin of the zygomaticofrontal suture at the intersections of the orbits; ZL-left, ZR-right. ZA(AZ), zygomatic center of the root of the zygomatic arch, midpoints. ZA-left, AZ-right. AG(GA), points at lateral inferior margin of the Antigonial protuberances. AG-left, GA-right.

Table. Cephalometric analysis

<i>Modified Steiner Analysis</i>		
	<i>Ref. Norm</i>	<i>Patient</i>
SNA	82°	80°
SNB	80°	73°
ANB	2°	7°
WITS appraisal (M/F)	-1/0	-1
Upper facial height	50%	43%
Lower facial height	50%	57%
GoGn to SN	32°	37°
SL	55 mm	41 mm
Interincisal angle	131°	101°
<i>Ricketts Analysis</i>		
	<i>Ref. Norm</i>	<i>Patient</i>
Convexity	2 ± 2 mm	6 mm
Max. depth	90°	86°
Facial depth	87 ± 3°	78°
Cranial length	55 ± 2.5 mm	64 mm
Porion length	-39 ± 2.2 mm	-43.5 mm
Mand. arc.	26 ± 4°	20°
Cranial deflection	27°	21.5°
Maxillary height	53 ± 3 mm	45 mm
<i>McNamara Analysis</i>		
<i>6 Y/O</i>	<i>Ref. Norm</i>	<i>Patient</i>
Max. length	81.7 ± 3.4 mm	88 mm
Mand. length	99.3 ± 3.6 mm	100 mm
LFH (ANS to Menton)	58.4 ± 3.1 mm	66 mm
Pog to N-FH perpendicular	-0.3 ± 13.8 mm	-20 mm
<i>Moyer's Facial Morphologic Analysis</i>		
	<i>Ref. Norm</i>	<i>Patient</i>
Ant. upper facial height mm	40.91 ± 2.41	44
Post. upper facial height mm	42.50 ± 2.91	37
Post. lower facial height mm	36.67 ± 3.31	44.5
Ant. max. height mm	24.94 ± 3.14	30
Post. max. height mm	9.06 ± 5.51	16
Mand. skeletal effective length	46.22 ± 1.69	40.5
Ant. cranial base (SE-FMN) mm	46.22 ± 1.69	41

dromes, including clefts, oculomandibulodyscephaly, chondroectodermal dysplasia, and hemifacial hypertrophy.¹⁹ Pyogenic granuloma and early eruption of teeth has been associated with increased vascularity in this syndrome.⁴ It has been proposed that changes of vascular pressure in the pulp will induce tooth erup-

tion.^{26, 27} Servelle studied venograms in 768 cases of KTW syndrome and concluded that if the main vein of a limb is ligated during childhood, the resulted venous stasis produces an elongation of the limb.²⁸ Based on this theory, venous vascular malformation leading to venous stasis might also cause premature dental eruption. However, this hypothesis cannot completely account for the phenomenon.^{26, 29} Premature dental eruption did not occur ipsilaterally in our case, nor the cases reported by Steiner.³⁰ The reason for early eruption in this syndrome needs further investigation.

Hemifacial hypertrophy and malocclusion have been reported previously.^{10, 16} However, the overall incidence rate has not been reported. Sciubba and Brown reported an increased bone density of one side of the mandible in one patient and moderate enlargement of one side of the maxilla in another.¹⁰ Cheruy and Heller described an adult female with hemihypertrophy of the ear, eye, nostril, lips, tonsils, and tongue (which was also papillomatous and scrotal).³¹ She also presented with hemimegalencephaly and dysphagia.

An initial cephalometric analysis was performed for this patient in order to establish baseline data of the dentoalveolar relationships. The maxilla enlargement and anterior open bite are similar to the reports of Steelmach, Miescher, and Steiner.^{15, 25, 30} Our study is the first to report these findings cephalometrically. The increased cranial length, maxillary length, and posterior lower facial height might account for these two findings. Macroglossia is another possible cause of anterior open bite in this case and those of Steiner. Frohlich reported that an excessively large tongue exerts expansive forces on the dental arches.³² He studied 27 cases and found that surgically reduced tongue size resulted in reduction of pressure from tongue on the teeth and a decrease in the freeway space of the mandible. The patient's Class II skeletal open-bite will need continuous monitoring to implement timely treatment that best augments the patient's potentially unusual craniofacial growth.

At present, the patient does not have severe facial imbalance (asymmetry). Should more pronounced asymmetry develop, treatment will be necessary. Asymmetry could be corrected by functional appliances. Proffit proposed the use of a custom-designed "hybrid" functional appliance to correct facial asymmetry.³³ However, in his original idea the appliance was designed for a patient with hemifacial microstomia. Another concern in considering the use of a functional appliance is whether the patient with KTW will or will not have the same growth spurt and growth timing as in the normal population. Howswell, et al., who reported two cases of primary hemihypertrophy of the face, noted that the process stops when growth ceases.³⁴ Their treatment involved surgical reconstructive procedures when growth ceased, e.g., soft tissue debulking, face lifts, osteotomies, and/or orthognathic surgery.

In the present case, the patient's physician reported an unusually powerful muscular contraction of the affected hand as that normally found in 16-year-old males. Therefore, possible facial imbalance resulting from unilateral hypertrophy of masticatory muscles needs special consideration in designing any type of therapy including functional appliances.

The major problem associated with dental management of the patient with this syndrome is excessive hemorrhage from any oral hemangiomatic lesions and delayed healing of surgical wounds. Sciubba reported one case of oozing and delayed healing of an extraction site on the affected side and normal healing response on the normal side.¹⁰ Surgical procedures require careful preoperative consideration in these patients. The use of hemostatic agents, electric cauterization, and ligation of the involved vessels should be taken into consideration. In planning a surgical procedure on a patient with KTW, radiographs can help detect intrabony hemangiomas lesions and venograms may be necessary to demonstrate deep venous channels.³⁵ Moreover oral-pharyngeal intubation should be considered to eliminate any post-operative bleeding complications via a nasal route.

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