Sturge Weber Syndrome, Klippel Trenaunay or Port Wine Stain? The Challenge of Diagnosis

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Abstract

Background: Sturge-Weber and Klippel-Trenaunay are congenital disorders characterized by the presence of vascular malformation that may indicate an incomplete mild clinical presentation and may even be overlapped. Thus, the challenge of diagnosis and the appropriate follow-up of these patients become necessary due to the possible late onset of alterations that are characteristic in these syndromes in order to provide a better quality of life for these patients. The aim of this case report is to describe a clinical case of a patient presenting signs suggestive of Sturge-Weber and Klippel-Trenaunay syndromes.

Case Report: A 5-year-old girl that was examined at a Stomatolgy Department in a Public University Hospital in the city of Rio de Janeiro – Brazil, who presented signs of one of the Sturge-Weber and Klippel-Trénaunay syndromes. She reported spontaneous nasal bleeding occasionally and oral bleeding in the area of the right mandible. Inspection showed gum hyperplasia, soft teeth and mild hypertrophy of lower left limb. The patient has been followed-up at the Clinical Genetics unit as well as the Stomatolgy Department.

Conclusion: This case report presents the challenges in making a diagnosis, treatment and clinical follow up on children presenting combined signs of rare congenital disorders such as Sturge-Weber and Klippel-Trenaunay syndromes.

Introduction

Vascular malformations are defined as developmental anomalies caused by deregulation in signaling related to proper formation of vascular tree, consisting of abnormal vessel channels lined with quiescent endothelium. Among many hereditary vascular malformation syndromes, there are congenital sporadic variants that can be accompanied by growth disturbances of soft or skeletal tissue [1-2]. Sturge-Weber syndrome and Klippel-Trenaunay syndrome are two examples of well known conditions that have received many names and classifications such as Klippel-Trenanay-Weber syndrome, Sturge-Weber disease, Sturge-Weber-Krabbe syndrome, Sturge-Kalischer-Weber syndrome, encephalotrigeminal angiomatosis and meningofacial angiomatosis [3].

Klippel-Trenaunay syndrome (KTS) consists of a cutaneous vascular nesus over the trunk or limbs in an asymmetrical distribution, associated with varicosities and hypertrophy of all or part of a limb, according to the triad: capillary malformation of one leg, ipsilateral hypertrophy and varicose veins. There are cutaneous port-wine stains, bone and soft tissue hypertrophy involving one or more limbs [4]. The central nervous system is rarely involved, but related abnormalities include cerebral and spinal arteriovenous malformations, orbital-frontal varicose veins, microcephaly or macrocephaly [5]. KTS patients with extensive lymphatic-venous malformations may develop a chronic destructive coagulopathy in which blood platelet count is minimally lowered [6]. Treatment is symptomatic.

Sturge-Weber syndrome (SWS) is characterized by leptomeningeleal and choroidal venous angiomatosis and ipsilateral capillary malformations in the face, usually following trigeminal nerve distribution and showing as a port-wine stain, present at birth and nonprogressive. Ipsilateral gyriform calcification of the cerebral cortex, sight defects (if ocular area is affected), mental retardation, epileptic fits, contra-lateral hemiplegia and affliction of oral mucosa and gums can also occur [2,7]. Vascular malformations can occur in the eye and may affect the conjunctiva, episclera, choroid, or retina. Hemangioma can be found on other regions of the body, mouth or internal viscera. The Roach scale classifies cases of SWS into three types, according to clinical features. Epileptic seizures are part of the disease spectrum [8-10].

Overlapping of KTS and SWS may occur. Since several case reports of patients presenting with characteristics of both diseases are being published in the last two decades, showing that there is no clear distinction between them, the two conditions can even represent different patterns of a same clinical entity. In this regard, we aim to present a case of a 5-year-old girl who shares clinical features between KTS and SWS, as well as to describe the oral aspects concerning treatment and follow-up with regards to its dental aspects.

Case Report

A 5-year-old Caucasian girl was referred to the Oral Medicine...
Clinic of a public teaching hospital, whose chief complaint was haemorrhagia during and after eruption of the first lower molar. During medical and dental history taking, her mother told that her father had learning difficulties as a child and his cousin had dysrythmia. Her paternal grandfather had dysrhythmia, hemangioma on the forehead, eye and nose on the left side and her paternal uncles and her sister had several areas of redness of the skin. The physical examination revealed an hemangioma on the right side of the face, extending to scalp, auricle, cheek, chin and neck, giving rise to facial asymmetry (Figure 1), a 10cm hemangioma on the left ankle and another of 0.5 cm on abdominal region. In all affected areas, there was increased temperature and the lower left member showed mild temperature difference and a higher volume of soft tissue in the area of calf and ankle when compared to right limb (Figure 2).

Intraoral examination showed an increase in the lower right alveolar region in the area of teeth 46, 85 and 84, conferring an appearance of open bite. Affected oral mucosa had an erythematous color extending over three quarters of the tongue, leading to macroglossia (Figure 3). This child presented neither apparent carious lesions nor any periodontal illness upon visual and touch inspection. Panoramic radiographic examination showed dental anomalies such as an increase in the right-hand mandible body of the menton foramen, and distal displacement of the bud of tooth 47 in relation to tooth 46 (Figure 4) CT scan was normal and there were no ophthalmological alterations.

The patient was referred to the Vascular Surgery unit of the same institution for evaluation and subsequent surgery. After that, the patient underwent two sessions of arterial embolisation with PVA particles on right facial and lingual arteries, the first with 300 ml and the second with 500 ml. Post-operative of both procedures occurred with no complications.

Follow Up

The patient is now receiving outpatient monitoring. By the time she was 10 and after three years of follow-up, she was referred to the Clinical Genetics unit. She reported spontaneous nasal bleeding occasionally and oral bleeding in the area of the right mandible. Inspection showed gum hyperplasia, soft teeth and mild hypertrophy of lower left limb. Upon drawing a pedigree, a pattern of autosomal dominant inheritance could be identified due to paternal relatives affected by lesions of vascular aetiology. The patient underwent abdominal ultrasound, echocardiography, and ophthalmological evaluation, all normal. Her follow up continues to date.

Discussion

This is a patient who has a complex congenital syndrome of vascular malformations with facial haemangioma, oral alterations compromising gum and teeth, combined with bone and soft tissue hypertrophy of one leg. According to the eponymous classification, this patient would meet the criteria for both Sturge-Weber syndrome as well as Klippel-Trenaunay syndrome. Based on several different published cases over the last 20 years, there is not much clarity in the distinction between KTS and SWS2, although it is well known that not every person who has a port-wine stain has SWS. In fact, 8 -33% of people with port-wine stain actually have the syndrome and also concluded that SWS only occurs when port-wine stains involve V1 distribution of the trigeminal nerve. Lesions are usually 85% unilateral and 15% bilateral. However, at the Sturge-Weber Foundation, unilateral lesions represent 49% of the reported vascular hemangiomata.

KTS diagnosis is different to SWS, as the two conditions share certain clinical and radiological features and over 40 combined cases have been previously related, including one with a combination of
encephalofacial angiomatosis and KTS, leading to an increase of the homolateral mandibular ramus [3,9]. In the present report, there is also a combination of clinical findings. The presence of macrogllossia and hypertrophy of the affected maxillary bone, causing malocclusion and facial asymmetry is a commonly related finding in SWS patients, and reconciles with the patient's clinical signs in this report.

Children with mild cases should be very well monitored for risk of possible development of medical problems that may occur later, such as vision problems, epilepsy and slow intellectual development. We must remember that the age for appearance of glaucoma varies from birth up to 41 years and convulsions, from birth to 23 years [8,11].

Patients with buccal alterations should be submitted to periodic examinations and detailed control of bacterial plaque as it is known that this can aggravate the vascular situation. Any treatment of surgical nature in areas associated to haemangiomata is considered of extreme risk of fatal haemorrhage and should be avoided. If truly necessary, such procedures should be carried out at a hospital facility with resources capable of being rapidly assessed in case of a bleeding emergency.

The case reported here leads us to consider it a rough or incomplete form, very similar to both SWS and KTS, neither of which can be ruled out. The hereditary pattern was discovered upon analysis of the family's pedigree. Autosomal dominant inheritance has been described previously in KTS, but not in SWS. Another clinical feature is that there are neither ocular problems nor any evidence of involvement of trigeminal V1, leading us to favor KTS over SWS, or still SWS type II, according to the abovementioned Roach classification. Thus we recommend periodic monitoring of this patient as we can see that SWS glaucoma and other KTS alterations may manifest later on. In addition, advances in embryology may clarify the origin of SWS variants. Normally diagnosis is made based on patient's history and his/her clinical examination, using complementary imaging exams for confirmation and documentation of the case. However, in unclear cases, a genetic evaluation made by a clinical geneticist may help state the diagnosis and from this moment on, the approach can be extendable to the whole family; creating bases provide accurate genetic counseling.

Oral alterations, being so frequent in these syndromes, makes the participation of a dental surgeon on the multidisciplinary team leading to affected patients something of utmost importance, in order to set the diagnosis and conducts. We also observe that in the light of so many alterations, both oral and systemic, a multidisciplinary and specialized team should monitor these patients so as to allow early diagnosis, preventive control and choice of adequate and suitable treatment.

**References**