Case Report
Olgu Sunumu
DOI: 10.4274/turkderm.39297
Turkderm - Arch Turk Dermatol Venerology 2016;50

A case of overlapping of Sturge Weber syndrome-Klippel Trenaunay syndrome and ophthalmological findings

Sturge Weber sendromu-Klippel Trenaunay sendromu birlikteliği olgusu ve göz bulguları

Ersin Aydın, Yakup Aksoy*, Ercan Karabacak, Bilal Doğan, Murat Velioğlu, Kürşat Göker**

Sultan Abdülhamid Han Training and Research Hospital, *Clinic of Ophtalmology, **Clinic of Radiology, İstanbul, Turkey

Abstract

Sturge-Weber syndrome (SWS) is a neurocutaneous syndrome characterized by facial port wine stains, vascular lesions in the ipsilateral brain and meninges, and glaucoma. Klippel-Trenaunay syndrome (KTS) is a rare congenital malformation associated with cutaneous vascular malformation, bony or soft tissue hypertrophy and venous varicosities in the affected limb. Although some cases have been reported in the literature, an overlap between these two phakomatoses is extremely rare and they have systemic and ocular affects. Here, we present a case showing the properties of both SWS and KTS and having interesting ophthalmological findings. This case is presented to emphasize that eye related complications might also be seen in these syndromes.
Keywords: Sturge-Weber syndrome, Klippel-Trenaunay syndrome, corneal thickness

Öz

Sturge-Weber sendromu (SWS) yüzde Porto şarabı lekesi, aynı taraf beyinde ve meninkste vasküler lezyonlar ve gözde glokom ile karakterize bir nörokutanöz sendromdur. Klippel-Trenaunay sendromu (KTS) ise kutanöz vasküler malformasyon, kemik ya da yumuşak doku hipertrofisi ve etkilenen ekstremitede venöz genişlemelerle karekterize nadir görülen bir konjenital malformasyondur. Literatürde az sayıda olgu bildirilmiş olmakla birlikte bu iki fakomatozun birlikte görülmesi nadir bir durumdur ve sistemik, oftalmolojik patolojilere neden olabilmektir. Burada SWS ve KTS özelliklerini bir arada gösteren, aynı zamanda ilginç oftalmolojik bulguları da olan bir olgu sunulmuştur. Bu olgu sunumu, bu sendromlarda gözle ilgili komplikasyonların da görülebileceğini vurgulamak amacı ile sunulmuştur.
Anahtar Kelimeler: Sturge-Weber sendromu, Klippel-Trenaunay sendromu, kornea kalınlığı

Introduction

Sturge-Weber syndrome (SWS) is characterized by port-wine nevus on face, leptomeningeal angiomatosis and cerebral calcification that involves one or both hemispheres and choroidal vascular lesions associated with glaucoma. Additionally early onset seizure, neurologic deterioration and neurodevelopmental delay can be seen[1,2]. Klippel-Trenaunay syndrome (KTS) is defined by triad of cutaneous and visceral hemangiomas which can appear at any site of the body, venous varicosity and hypertrophy of soft tissue and bone of the affected limb[3]. However extremely rare, many cases with overlapping of SWS and KTS have been reported[4,5]. We report here an unusual case with features of SWS and KTS and corneal thickness difference between two eyes.

Case Report

A 20 year old male presented with red macules and plaques on the right side of the body and face that had beennpresent since birth. There were no characteristic family story and medical history. On dermatological examination well-defined, non-blanchable, erythematous macules and plaques on right

Address for Correspondence/Yazışma Adresi: Yakup Aksoy Sultan Abdülhamid Han Training and Research Hospital, Clinic of Ophtalmology, İstanbul, Turkey
Phone.: +90 553 594 35 36 E-mail: dryakupaksoy@gmail.com **Received/Geliş Tarihi:** 15.10.2015 **Accepted/Kabul Tarihi:** 19.02.2016

half of the face, and hard palate have been detected. On face it was matching distribution of the ophthalmic and maxillary branches of the trigeminal nerve. The nevus flammeus was also continued to right shoulder, chest, back, both anterior and posterior faces of upper and lower limbs (Figures 1a, 1b). The right upper and lower limbs were taller and hypertrophic than left limbs. The upper limbs were measured from humerus head to radius distal in whole-extremity anteroposterior (AP) radiographs (orthoroentgenogram). Right upper limb was 2.4 cm (right:left 55.8:53.4) taller than the left. The lower limbs were measured from femur head to tibia distal in whole-extremity AP radiographs (orthoroentgenogram). Right lower limb was 0,4 cm (right:left 79.5:79.9) taller than the left. There were also 2 cm difference in circumference of thighs (right:left 53.5:51.5) and 1cm difference in circumference of arms (right:left 32:31). Osteohypertrophy was not present in orthoroentgenogram of our case. Cranial computed tomography (CT) and magnetic resonance imaging (MRI) was done. CT imaging showed coarse calcifications in the inferior temporal gyrus and hypertrophy in the frontal sinus ipsilateral to port-wine nevus (Figure 2a). In the cranial MRI there were hypertrophy in the lateral ventricle choroid plexus, mild dural thickening and choroidal thickening in the eye which were also ipsilateral to port-wine nevus (Figure 2b, 2c, 2d). The biochemical, hematologic and urinary laboratory tests were normal. Whole abdominal ultrasonography for visceral involvement and chest X-ray showed no remarkable finding. Doppler ultrasound examination of the right limb showed no arteriovenous fistula. The neurological examination was normal and no mental or motor anomaly was detected.

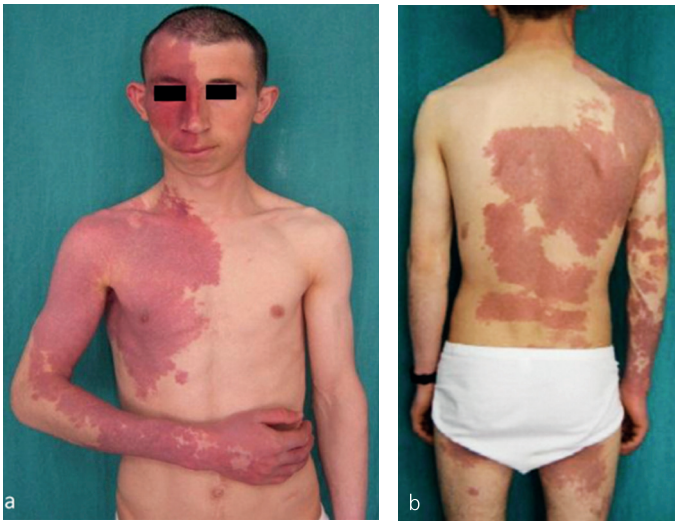

Figure 1. a) The nevus flammeus on right shoulder, chest and anterior face of upper and lower limbs b) The nevus flammeus on right shoulder, back and posterior face of upper and lower limbs

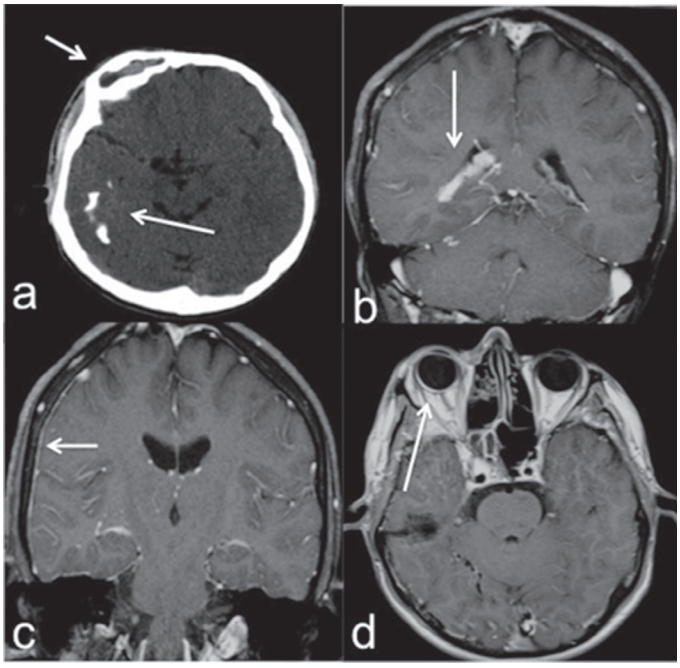

Figure 2. a) Axial cranial computed tomography image shows right temporal calcifications (long arrow) and hypertrophic right frontal sinus (short arrow). b) Contrast enhanced coronal T1 weighted image shows hypertrophy of right choroid plexus (white arrow). c) Contrast enhanced coronal T1 weighted image shows right mild dural thickening (white arrow). d) Contrast enhanced axial T1 weighted image shows right choroidal thickening in the right eye (white arrow)

On ophthalmological examination eyelids were purple colored due to subcutaneous port wine stain. Visual acuity was 20/20 in both eyes. Conjunctival and scleral vessels were large, tortuous and especially near the limbus these vessels had formed a ring appearance on limbus in right eye (Figure 3a, 3b). Fundus reflex was red in right eye while it's orange in left. Choroidal vessels were larger and more tortuous than left eye in right. The right eye had a glaucomatous cupping of the optic disc with the 5/10 cup/disc ratio while 2/10 in the left. Ocular tension was 30 mmHg on right and 15 mmHg on the left. The refraction was -0.25 dioptre in right and -3.00 dioptre on left. Ultrasonographic axial length (from fovea to corneal surface) was 22.23 mm on right and 23.07 mm on left. Pachymetric central corneal thickness (from anterior surface to posterior surface of cornea) was 605 µm in the right eye and 585 µm in the left.

Discussion

SWS was first defined by Sturge in 1879, and then was expanded by Weber more clearly. The incidence of SWS is 1/50000. SWS also called encephalofacial or encephalotrigeminal angiomatosis, is characterized by purple-colored flat cutaneous cranial port wine stain, most commonly along the trigeminal nerve, glaucoma and vascular lesions in the ipsilateral brain and meninges[2]. KTS is present at birth and usually involves a lower limb but may involve more than one and portions of trunk or face. It includes irregular and asymmetrical capillary and cavernous hemangiomas on the trunk or limbs, arteriovenous

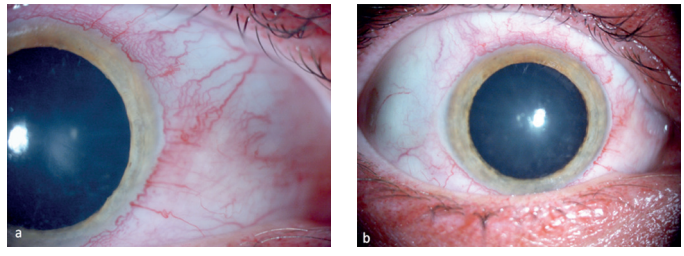

Figure 3. a) The large and tortuous conjunctival and scleral vessels b) The large conjunctival and scleral vessels form a ring appearance on limbus

fistulae, lymphedema, varicosities, asymmetrical hypertrophy and visceromegaly. Enlargement of the soft tissues may be gradual and may involve the entire limb, a portion of it, or selected digits. Most complications of this syndrome are related to the underlying vascular pathology. These include stasis dermatitis, thrombophlebitis, limb disparity. More serious sequelaes are thrombosis, pulmonary embolism, cerebral arteriovenous malformations, spinal arteriovenous malformations, macrocephaly, microcephaly, orbito-frontal varices and congestive heart failure[6]. Also they rare and the pathogeneses are still not clearly elucidated; the majority of cases are thought to result from somatic mutations of genes that play significant roles in embryonic vasculogenesis and angiogenesis[7]. Both diseases occur almost always sporadically, but a dominant autosomal inheritance has already been described in some families[8]. Additionally it's estimated that both of these two syndromes are the same which are presented differently due to difference in involvement.

Our case had port-wine staining on right half of the face, glaucoma and cerebral calcification on CT which are compatible with SWS and upper and lower limb hypertrophy and port wine stain which are compatible with KTS. Our case was diagnosed to SWS+KTS overlapping with these findings. These syndromes may have many systemic affects[6]. But our case had no other systemic involvements more than classical findings and ophthalmological findings. Other than glaucoma some ocular manifestations such as, buphthalmus, choroidal angioma, iris hypochromia, nevoid marks or vascular dilatation of the episclera and retinal detachment also have been reported[9]. In our case cup/disc ratio of right eye was glaucomatous due to 30 mmHg intraocular pressure which is higher than normal level. Axial length of right eye was about 1 mm shorter than left possibly due to thicker choroid layer of right eye. This difference in axial length caused about 3 dioptre difference between two eyes. Normal thickness of central cornea is about 551.3±26.2 μm[10]. Central cornea was thicker in the right eye than the left. Also we couldn't find any knowledge in the literature, we thought that this difference in corneal thickness could be due to hypertrophic affect in soft tissue that is seen in KTS.

We informed the patient about his disorder, begin anti glaucomateus treatment and prescribed glasses. There is no specific curative treatment for this combined disorder. Supportive care includes compression bandages for varicosities and surgical treatment may help selected patients.

In this report, we accentuate the importance of thorough ocular and radiological evaluation of cases that present with SWS+KTS. Recognition of possible underlying systemic and local anomalies and complications in overlapping of SWS and klippel trenaunay weber syndrome helps management of these syndromes.

Ethics

Informed Consent: Consent form was filled out by all participants.
Peer-review: Externally and internally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: Yakup Aksoy, Ersin Aydın, Murat Velioğlu, Concept: Ersin Aydın, Yakup Aksoy, Ercan Karabacak, Bilal Doğan, Kürşat Göker, Design: Ersin Aydın, Yakup Aksoy, Ercan Karabacak, Murat Velioğlu, Data Collection or Processing: Ersin Aydın, Yakup Aksoy, Ercan Karabacak, Bilal Doğan, Kürşat Göker, Analysis or Interpretation: Ersin Aydın, Yakup Aksoy, Ercan Karabacak, Bilal Doğan, Murat Velioğlu, Kürşat Göker, Literature Search: Ersin Aydın, Yakup Aksoy, Bilal Doğan, Murat Velioğlu, Kürşat Göker, Writing: Ersin Aydın, Yakup Aksoy.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study received no financial support.

References

1. Purkait R, Samanta T, Thakur S, Dhar S: Neurocutaneous syndrome: a prospective study. Indian J Dermatol 2011;56:375-9.
2. Bilir Göksügür S, Şereflican B, Göksügür N, et al: Fakomatozis Pigmentovaskülaris Tip IIB ile Sturge Weber Sendromu ve Klippel Trenaunay Sendromu Birlikteliği. Dermatoz 2014;4:1-4.
3. Timur AA, Driscoll DJ, Wang Q: Biomedicine and diseases: the Klippel-Trenaunay syndrome, vascular anomalies and vascular morphogenesis. Cell Mol Life Sci 2005;62:1434-47.
4. Sfaihi L, Aissa K, Fourati H, et al: Klippel Trenaunay syndrome in association with Sturge Weber syndrome about one case. Tunis Med 2014;92:173-4.
5. Purkait R, Samanta T, Sinhamahapatra T, Chatterjee M: Overlap of sturge-weber syndrome and klippel-trenaunay syndrome. Indian J Dermatol 2011;56:755-7.
6. Thomas-Sohl KA, Vaslow DF, Maria BL: Sturge-Weber syndrome: a review. Pediatr Neurol 2004;30:303-10.
7. Kihiczak GG, Meine JG, Schwartz RA, Janniger CK: Klippel-Trenaunay syndrome: a multisystem disorder possibly resulting from a pathogenic gene for vascular and tissue overgrowth. Int J Dermatol 2006;45:883-90.
8. Garzon MC, Huang JT, Enjolras O, Frieden IJ: Vascular malformations. Part II: associated syndromes. J Am Acad Dermatol 2007;56:541-64.
9. Cohen MM Jr: Klippel-Trenaunay syndrome. Am J Med Genet 2000;93:171-5.
10. Sen S, Bala S, Halder C, Ahar R, Gangopadhyay A: Phakomatosis pigmentovascularis presenting with sturge-weber syndrome and klippel-trenaunay syndrome. Indian J Dermatol 2015;60:77-9.
11. Chhajed M, Pandit S, Dhawan N, Jain A: Klippel-Trenaunay and Sturge-Weber overlap syndrome with phakomatosis pigmentovascularis. J Pediatr Neurosci 2010;5:138-40.
12. Abdolrahimzadeh S, Scavella V, Felli L, et al: Ophthalmic Alterations in the Sturge-Weber Syndrome, Klippel-Trenaunay Syndrome, and the Phakomatosis Pigmentovascularis: An Independent Group of Conditions? Biomed Res Int 2015;2015:786519.
13. Sharma P, Arya AV, Azad RV: Unusual retinal manifestation in a combination of Sturge-Weber and Klipplel-Trenaunay syndrome a case report. Indian J Ophthalmol 1990;38:195-7.
14. Doors M, Cruysberg LP, Berendschot TT, et al: Comparison of central corneal thickness and anterior chamber depth measurements using three imaging technologies in normal eyes and after phakic intraocular lens implantation. Graefes Arch Clin Exp Ophthalmol 2009;247:1139-46.