Intracranial Variant of Encephalotrigeminal Angiomatosis – A Case Report

Abstract

Encephalotrigeminal Angiomatosis is a rare developmental phakomatoses characterized by the occurrence of nevus flammeus (port-wine stain) along the distribution of branches of trigeminal nerve, vascular angiomas in the eye, and leptomeningeal angiomas affecting 1 in 1,00,000 South Asian population. Herewith, such a rare case of such encephalotrigeminal angiomatosis in a 24-year-old male is described.

Keywords: Hemianopsia, phakomatoses, port-wine stain, status epilepticus, trigeminal nerve

Encephalotrigeminal angiomatosis is a rare, nonhereditary developmental condition that is characterized by hamartomatous vascular proliferation involving the tissues of the brain and face. It is believed to be caused by the persistence of a vascular plexus around the cephalic portion of the neural tube. This plexus develops during the sixth week of intrauterine development but normally undergoes regression during the ninth week.

A 24-year-old male patient reported to our Department of Oral Medicine and Radiology with a chief complaint of bleeding from his gums in the left upper back tooth region only during brushing his teeth. History reveals that he has episodes of head ache and seizures for the past 2 years. Extraoral examination reveals an unilateral purplish irregular nodular swelling seen unilaterally only on his left side of his vermilion border of his upper lip [Figure 1]. His left lateral profile view reveals an extraoral swelling on his left side of the face extending superiorly 3.5 cm away from the left lower eyelid, inferiorly extending 1.5 cm below the ala-tragal plane, anteriorly extending 1 cm away from the left side of the ala of the nose, posteriorly it extends 1.5 cm in front of the tragus of left ear and discrete areas of scars seen over the surface of the swelling on the skin of the left side of his face and his left eye is reddish shows dilated blood vessels in the sclera of the eye [Figure 2]. Intraoral examination reveals an irregular gingival overgrowth of marginal and attached gingiva covering almost the buccal aspect of the crowns in relation to 24, 25, 26, 27 region due to the hyperplastic growth in relation to 23, 24 region [Figure 3] and an area of unilateral bright reddish area on labial aspect of marginal and attached gingiva in 31, 32 tooth region [Figure 4] and along the left side of the posterior most region of the hard palate [Figure 5].

Considering the presence of bright reddish hyperplastic growth on gingiva and bleeding from the gingiva, it is provisionally diagnosed as of pyogenic granuloma or leukemia.

Differential diagnosis of encephalotrigeminal angiomatosis are tuberous sclerosis, Klippel–Trenaunay syndrome, Von Hippel Lindau syndrome, Wyburn–Mason syndrome, neurofibromatosis, PHACE syndrome, Cobb syndrome, Maffucci syndrome, Gorham–Stout syndrome, and Parkes Weber syndrome.

Conventional topographic maxillary occlusal radiograph reveals areas of tiny discrete flecks of calcification in the region of the left side of the Hard Palate [Figure 6]. Digital orthopantamographic image does not reveal any erosion of the floor of the maxillary sinus or left pterygoid plates, but only revealed a horizontally impacted tooth 48 [Figure 7]. Digital lateral cephalometric radiograph revealed tramline calcification due to leptomeningeal calcifications [Figure 8].

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Discussion

Encephalotrigeminal Angiomatosis also called as Sturge–Weber Angiomatosis or Sturge–Weber syndrome or Sturge–Weber Phakomatoses is a rare phakomatoses characterised by the association of a tiny portwine capillary vascular malformation along the distribution of the branches of the trigeminal nerve on the skin of the face called portwine stain or naevus flammeus with ipsilateral vascular glaucoma and vascular malformation of the eye, and leptomeningeal angioma.[1] The gene that causes Sturge–Weber syndrome was discovered at the Kennedy
William Allen Sturge and Frederick Parkes Weber described this disorder in a 6-year-old male child. Sturge–Weber syndrome is caused by a somatic mutation in the GNAQ Guanine Nucleotide Adenine nucleotide-binding protein G (q) subunit alpha (Gαq) gene on chromosome 9q 21.2.[2]

Dysregulation of vascular MAPK and/or PI3K signaling during human embryonic development plays a part in the pathogenesis and progression of Sturge–Weber syndrome.[3]

In our case, irregular nodular swelling seen unilaterally only on the left side of vermilion border of upper lip and discrete areas of scars seen on the skin of the left side of the face; these are due to the involution of port-wine stain that occurred unilaterally on the left side of the cheek on the skin of the face resulting in facial asymmetry as described by Ogul.[4] Port-wine stains are seen unilaterally on the posterior most hard palate region, suggesting the involvement of maxillary division of trigeminal nerve (V2) and the labial aspect of attached gingiva in relation to 31, 32 region, suggestive of involvement of mandibular division of trigeminal nerve (V3) and leptomeningeal calcifications and absence of unilateral port-wine stain on the skin of the face suggests it as a case of intracranial variant of Sturge–Weber syndrome.

Portwine stains are a type of fine capillary hemangiomomas, if when affects the gingiva makes it to appear more bright red in colour, hyperplastic resulting in gingival enlargement and easily friable on slight provocation resulting in bleeding. This is the cause of bleeding from the Gums and such lesions can mimic pyogenic granuloma. Sturge–Weber syndrome affects 1 in 50,000 live births. The main symptom is intracranial leptomeningeal angiomatosis, which mostly affects the occipital and posterior parietal lobes and can occur unilaterally and also bilaterally. Facial cutaneous vascular alterations occur ipsilaterally in the form of port-wine stains (nevus flammeus), which are normally found in the area of distribution of the trigeminal branch VI. Other clinical symptoms associated with Sturge–Weber syndrome are ocular manifestations such as glaucoma (30-70%), strabismus, hemianopsia (40-45%), visual field defects, and even blindness due to retinal detachment and episcleral hemangiomomas.[3] Neurological manifestations such as headaches (40–60%), seizures (75–90%), transient neurological stroke-like episodes, and cognitive impairment. Neurological cortical symptoms often include hemiparesis (25–60%) and hemiatrophy. The various imaging modalities includes intraoral periapical radiograph, in which occasionally cupping or resorption of alveolar bone may be seen, occlusal radiograph, which may reveal discrete flecks of calcifications due to the reason that vascular malformation can produce stasis of blood increasing the chance of thrombi from easily rupturable single endothelial lining of capillaries favoring dystrophic calcifications.

Krieger Institute in 2013 by Dr. Anne Comi and her collaborators.[3]

Figure 7: Digital orthopantamographic image did not revealed any erosion of floor of maxillary sinus or pterygoid plates and horizontally impacted tooth 48

Figure 8: Lateral cephalometric radiographic image showing tramline calcifications in the skull

Figure 9: Gadolinium contrast enhanced MRI scan-axial section reveals hypointense areas near the left buccinator muscle
The brush sign is an abnormally accentuated signal drop of the subependymal and deep medullary veins in T2 weighted MRI images in patients with encephalotrigeminal angiomatosis.[6]

Fluorescein angiography of eye helps to evaluate retinal vein to vein anastomosis in patients with Sturge–Weber syndrome.[7]

Recently, optical coherence tomography helps in the visualization of port-wine stains in patients with Sturge–Weber syndrome.[8] Gadolinium contrast enhanced magnetic resonance imaging (MRI) reveals irregular areas of variable low intensity (Hypointense) signal near the Left Buccinator muscle affected by Capillary hemangioma [Figure 9].

FDG–PET (Fluorodeoxyglucose–Positron Emission Tomography) helps to evaluate the metabolic activity and decreased blood flow due to vascular malformation in patients with encephalotrigeminal angiomatosis and proton magnetic resonance spectroscopic imaging helps to evaluate glutamate turnover.[9]

Ruthenium 106 plaque radiotherapy is effective for management of choroidal angiomas in Sturge–Weber syndrome.[10]

Verteporfin photo dynamic therapy is an effective and safe treatment for patients with choroidal hemangioma associated with Sturge–Weber syndrome.[11]

The treatment and prognosis of Sturge–Weber syndrome depends on the nature and severity of the possible clinical features. The facial port-wine stains and hyperplastic gingiva can be managed by the use of newer flash lamp pulsed dye lasers.[12]

Topical rapamycin combined with pulsed dye laser helps to effectively treat port-wine stains on the skin.[13] Port-wine stain affecting the gingiva can make the flossing and dental prophylaxis difficult. Recently, cryotherapy is one of the recommended treatment modality for management of gingival enlargement in patients with Sturge–Weber syndrome.[14]

Dentists must deal with such lesions with great care, when performing surgical procedures in the affected areas of the mouth because severe haemorrhage may be encountered. Epilepsy in this patient can be managed by i.v. Lorazepam 0.04 mg/kg, 4 mg/dose slowly intravenously, dose not to exceed 10 mg/day.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient has given his consent for his images and other clinical information reported in the journal. The patient understand that his name and initial will not be published and due efforts is made to conceal his identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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