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GLAUCOMA IN STURGE WEBER SYNDROME IN AN INFANT-AN INTERESTING CASE REPORT

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INTRODUCTION

Sturge weber syndrome (SWS) [encephalotrigeminal angiomatosis] is a congenital sporadic phacomatosis, characterised by the classical triad of Nevus Flammeus [portwine stain]of face, Leptomeningeal angiomatosis with cerebral gyriform calcifications and Choroidal haemangioma with or without glaucoma. Not all patients have all the three components of the syndrome, presence of two is required for diagnosis [bisystem disease-face + eyes/leptomeninges]. Other symptoms that may be present are seizures,hemiparesis, stroke like episodes, intracranial calcifications and in many cases mental retardation. Presence of facial haemangioma prompts investigation for ocular or brain pathology but over 85% of pts. with facial haemangioma do not have ocular/neural problems.On the contrary,there are no pts. with neurologic and ocular disease without a cutaneous haemangioma. S.W.S occurs sporadically with a frequency of approx. 1/ 50000 live births.

ETIOPATHOGENESIS

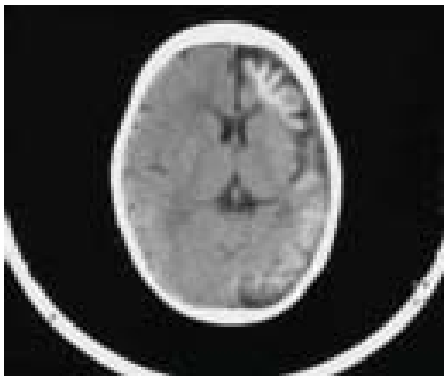
The condition is thought to result from anomalous development of primordial vascular bed in the early stages of cerebral vascularisation.At this stage,the blood supply to the brain,meninges and face is undergoing reorganization,while the primitive ectoderm in the region differentiates into skin of upper face and occipital lobe of cerebrum.In pts. with SWS overlying leptomeninges are richly vascularised and the brain beneath becomes atrophic and calcified,particularly in the molecular layer of cortex.The cutaneous haemangioma are capillary haemangioma/ telangiectasias,composed of dilated capillaries,hence appear bright red/portwine in colour.Choroidal and leptomeningeal haemangioma are cavernous haemangioma consisting of dilated and anastomotic vascular spaces

CASE REPORT

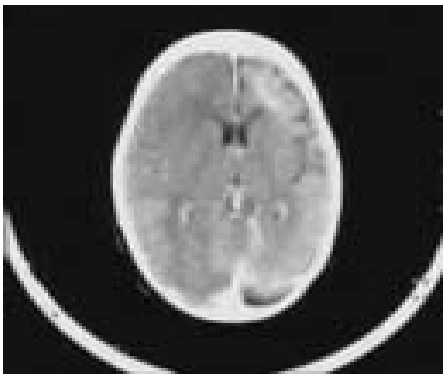
A 6 month old male infant presented to paediatric dept.from a village near Berhampur. His mother complained of enlarged and hazy black part of the left eye with photophobia in same eye since birth,weakness of right side limbs,delayed milestones,with portwine stain on left side of face mostly on upper half involving lids.He was referred to ophthalmology dept.for detailed examination of the eye.On clinical examination of left eye,we found corneal oedema,megalocornea measuring about 14 mm.Pt. was examined under anaesthesia, IOP was raised to 29 mm of Hg as measured with Schiotz tonometer. AC was deep.Fundus picture couldn't be assessed due to corneal oedema.Gonioscopy couldn't be done as the pt. was a infant.CT scan of pt. showed left temporo-parietal calcification with associated cerebral atrophy.Contrast -enhanced CT showed enhancement of angiomatous malformation and atrophy of the ipsilateral hemisphere.Axial non-enhanced Ct showed left hemiatrophy of the cerebral cortex and typical gyral calcification.Pt.was put on topical medication with beta blocker plus Carbonic Anhydrase Inhibitor combination for 1 month for control of IOP. It was refractory. So trabeculectomy surgery was planned for left eye. IOP of pt after trabeculectomy reduced to 17.3 mm of Hg and gradual clearing of corneal edema was noticed in 6 wks time post. op. Fundus picture was normal ophthalmoscopically. As photophobia and corneal edema had reduced significantly postoperatively,we assessed the vision must have improved in left eye.Clinical exam of right eye was normal in all aspects.Pt.was put on systemic drugs for seizure by paed.dept.

DISCUSSION

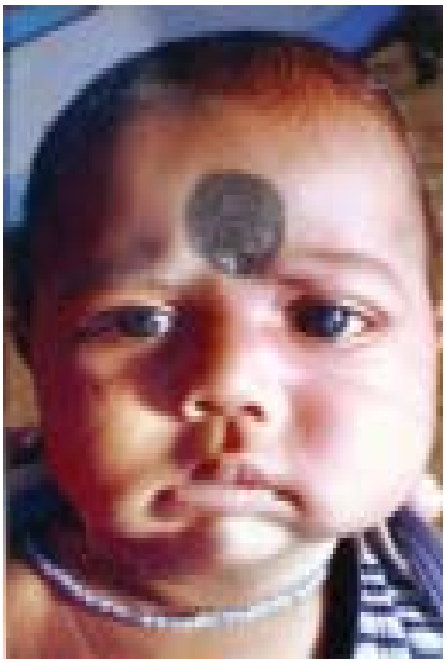
Facial angioma is present at birth,tends to be unilateral and always involves the upper face and eyelid.It



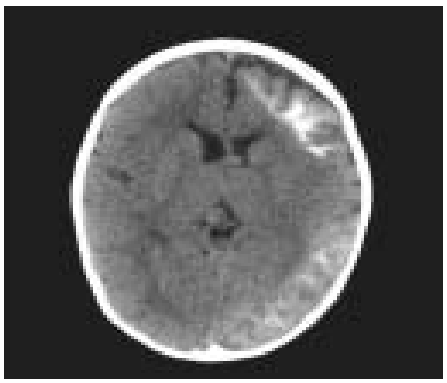
Axial nonenhanced CT scan shows left hemiatrophy of the cerebral cortex and typical gyral calcification.



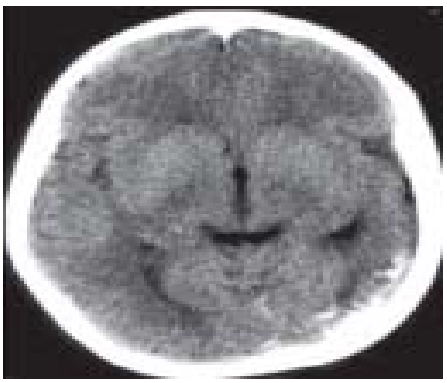
Contrast-enhanced CT showing enhancement of angiomatus malformation and atrophy of the ipsilateral hemisphere



Sturge Weber Syndrome showing Portwine stain on the left side of face and Bupthalmus of left eye.



CT Scan showing extensive pial malformation over the left cerebral hemisphere with volume loss



CT Scan of patient showing Left-temporal parietal calcificaiton with associated atrophy

occurs along the distribution of the ophthalmic,maxillary and rarely mandibular division of the trigeminal nerve.Lesion usually stops along the facial midline but may cross it.Nevus may also be evident over the lower face,trunk and in the mucosa of mouth and pharynx.This doesn't respond to steroids,rather treated with CO2 laser.CNS vascular malformations are most often confined to the pial vessels of the occipitoparietal area.Slow flow of blood through these vessels leads to hypoxia, encephalomalacia, cortical atrophy,subsequent calcification. Majority of pts.have intracranial calcification in a "serpentine/rail road track/tramline"fashion. About 80% of pts. develop focal or generalized tonic clonic seizures contralateral to the side of facial nevus.Seizures develop mostly within 1st year of life and is a poor prognostic factor.Seizure becomes refractory to anticonvulsants and may be associated with developmental delay and slowly progressive hemiparesis, hemiplegia or hemianopia. Transient stroke like episodes or visual defects persisting for several days and unrelated to seizure activity are common and probably result from thrombosis of cortical veins in the affected region. Although neurodevelopment appears to be normal in the 1st yr of life,mental retardation or severe learning disabilities are present in atleast 50% in later childhood, probably the result of prolonged generalized seizures and increasing cerebral atrophy secondary to local hypoxia and use of numerous anticonvulsants.

Ophthalmic findings : 1) Congenital glaucoma in SWS is almost always associated with involvement of both lids and areas of distribution v1 and v2 branches of trigeminal nerve by portwine stain.In one study 60-71% of pts. with SWS developed glaucoma,among whom 2/3rd case developed glaucoma <2 yrs age resulting in buphthalmos.In the rest of the cases glaucoma may

develop anytime from infancy to adulthood.Pathogenesis involves a)outflow obstruction because of abnormal angle development and vascularisation of iris

b) Isolated trabeculodysgenesis in infants

c) Increased episcleral pressure with decreased outflow[ass. with arteriovenous communication in an episcleral haemangioma] may be responsible in older pts.

d) Secondary angle closure in older pts. with choroidal haemangioma,retinal detachment and neovascular glaucoma

e) Hypersecretion of aqueous

f) Hyperpermeability of blood vessel walls of choroidal haemangioma

g) Adult open angle glaucoma mechanisms

2) Episcleral haemangiomas-69% pts.

3) Choroidal haemangiomas-55% of pts.These cavernous haemangiomas are usually diffuse in SWS.Most often located temporal to optic discand are most elevated in the macular area.Haemangiomas do not cause any visual disturbances early in life but may lead to late development of overlying retinal cystoid changes or exudative retinal detachment

4) Other ocular findings-heterochromia iridis, conjunctival angioma,retinal aneurysms ass. with an arteriovenous angioma of the thalamus and the midbrain.Visual field defects may be present in pts.with occipital lobe meningeal involvement and cortical atrophy.

In general more extensive the haemangioma, more severe the glaucoma and earlier its presentation. Corneal diameter is enlarged but no Haab's striae are found. Gonioscopy and histopathological studies reveal goniodysgenesis in infantile cases, uveal meshwork is thick, sclera spur is poorly developed, iris insertion may be high, there may be a Barkan's membrane or vascularisation of angle and trabecular meshwork.

Glaucoma is resistant to medical therapy,but tried first.Prostaglandin analogues,beta blockers,CAI,topically can be given.If IOP controlled, surgery is averted / delayed.If IOP remains high, goniotomy/ trabeculectomy is the initial procedure of choice.

* 1-2 yrs age-goniodysgenesis is present which responds to goniotomy [response is less than in primary congenital glaucoma]
* older age-primary trabeculectomy
* combined trabeculectomy with trabeculotomy gives good results. Trabeculotomy-addresses barrier to aqueous outflow posed by a congenital angle anomaly

  Trabeculectomy-bypasses the episcleral veins
* filtering procedures can be combined with antimetabolites

Complications of surgery: Due to high episcleral venous pressures, choroidal effusions develop after any form of pressure lowering surgery.Too sudden decompression of the globe may give rise to expulsive choroidal haemorrhage.Filtration surgeries must be coupled with measures to minimize any period of hypotony,such as intraoperative infusion and tight releasable sutures.

CONCLUSION

Seizures-treated by anticonvulsants but are usually refractory.If lesion is limited to one hemisphere and child is less than 1-2 yrs,hemispherectomy may be done

Portwine stain-responds well to flash lamp pulsed laser therapy especially if located on forehead.

Special educational facilities have to be given to mentally disabled children

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