

Hemimegalencephaly: Report of a Rare Case

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ABSTRACT

A 2-year-old boy is reported with hemimegalencephaly (HME) and associated congenital hyperplastic tissue in the pre maxillary region. Computed tomography not only evaluated the extent of the lesion, but also demonstrated the cerebral anomaly. Magnetic resonance imaging plays an important role in the assessment of conditions or lesions suspected of having hyperplastic fatty tissues. HME is a severe developmental malformation of the brain, remarkable for its extreme asymmetry. HME is of these forms: Isolated form, which is the most common form and found in 66% cases, syndromic form, which is associated with several neurocutaneous syndromes. We describe a case of congenital hypertrophy of the cheek associated with HME. The purpose of this case study is, when dental surgeon comes across this kind patient in clinical practice, the dental surgeon should have enough knowledge about this conditions and dental surgeon should know what kind of dental management give this kind of anomaly.

Keywords: Congenital, Epilepsy, Hemimegalencephaly, Polymicrogyria, Psychomotor, Seizures

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INTRODUCTION

As stated by many researchers, hemimegalencephaly (HME) can be a solitary abnormal embryonic or foetal development, or it can be attached to neurocutaneous syndromes, involving the brainstem and cerebellum, such as neurofibromatosis tuberoses sclerosis, Proteus syndrome, Klippel-Trenaunay-Weber syndrome, and linear sebaceous nevus syndrome.¹

Magnetic resonance imaging (MRI) shows the midsagittal structures in between two ventricles; these are similar to bands in HME. Some investigators studied it and they found that these bands like structures are not frequently found in between ventricles, and they used diffusion tensor MR and fiber tract (FT) reconstruction imaging for the study.²

Molecular causes of HME remain unclear, but some investigators done study on this and suggested that molecular cause of HME could be because of *de novo* somatic mutation in phosphatidylinositol 3-kinase-AKT3-mammalian target of rapamycin component pathway. Some authors done study on patients with HME and suggested that different diverse mechanism is the cause of anomaly and maybe it's a neoplastic deformation or a focal malformation.^{3,4}

In HME condition, the only one-sided cerebral hemisphere is distended. Exact cause of this condition is not known and has been hypothesised to happen due to injury during the second trimester of pregnancy or as early as the 3rd week of gestation, or as a hereditarily planned growing disorder linked to cellular lineage and formation of asymmetry. Investigators done a study on animal and concluded the role of AKT3 gene in controlling brain size. And investigator suggests that AKT3 activity forcefully modulates brain size in humans.⁵

HME is a complicated illness with multiform clinical presentations and epilepsy is the worst presentation. MRI is the most convenient way for showing the features of HME with congenital-infiltrating lipomatosis of the face. MRI can distinguish between congenital-infiltrating lipomatosis and HME associated with other neurocutaneous syndromes. HME also seen associated with other syndrome such as Proteus syndrome and Ohtahara syndrome.⁶⁻⁸

Authors theorize that the HME patients unable to control epileptic attacks which are responsible for growth delay of affected hemisphere. Some researchers have done a study on the patients with HME, and in that four patients had HME along with neurocutaneous syndromes and rest of the patients had solitary HME.

Two patients required surgical management for seizures in childhood. One patient had no intellectual affliction, and one patient had mild intellectual affliction, and three patients had severe intellectual disability. All patients had shown motor deficits ranging from mild hemiplegia. One patient, who had started getting seizures at the age of 7 years, had better seizure control and psychomotor growth in adulthood than patients in whom seizure onset occurred in the 1st year of life.^{9,10}

Computed tomography (CT) shows the number of the pathologies which may trigger epilepsy, especially those associated with the acute presentations such as haemorrhage, infarction and mass lesions as well as obvious malformations, and calcified lesion. And in some cases, where there is immediate management required CT is needed some condition such as Sturge-Weber or tuberous sclerosis and subtle cortical malformations, small tumors, mesial temporal sclerosis evaluated with the help of CT.¹¹

Some authors suggest that primary management of seizure is embolization and before doing any kind of invasive treatments, surgical or embolic, every effort must be made to establish seizure control medically. First advice is medication, then if seizures are not in control than advice for invasive treatment.¹²

CASE REPORT

A 2-year-old boy came to our Department of Oral medicine and Radiology with the complaint of right cheek swelling since birth. He was the second child of his parent. And he was a child of a full-term pregnancy of consanguineous parent. The prenatal history was normal, and the baby cried immediately after birth. There was no familial history of such complaints in the family. Physical examination showed right cheek enlargement caused by a mass, which is present since birth. The mass is huge in size and causes the obliteration of mouth, and the sagging of the right corner of the mouth is seen (Figure 1).

No other physical abnormalities were noted in the patient. This mass had ill-defined borders and normal skin color and texture extended anteriorly ala of nose to posteriorly preauricular region and superiorly periorbital region to inferiorly lower border of mandible and size is not possible to measure because of ill-defined border occupied the entire part of the right cheek and slightly large size of the right ear compared to left (Figure 2). The patient had epileptic seizures at the age of 6 months, and seizures were controlled by medication.

The dermatological examination was normal. Neurological examination was normal, and the patient

was holding his neck properly, this is showing the growth of the patient. The general examination revealed a normal, symmetrical development of the extremities, and the trunk. But, delayed milestones were noted in the patient.

For the complete study of the case, we did laboratory study of ionic calcium, electroencephalogram (EEG) mapping, CT of the face, and colour Doppler ultrasonography of the cheek. As per laboratory study, ionic calcium is in the normal range. The EEG mapping showed slowed background mainly over the right hemisphere and a generalised burst of a slow sharp wave. CT of the brain shows large sized right-sided cerebral hemisphere with mild dilatation of the right lateral ventricle (Figure 3). There is an atrophy of sinuses seen (Figure 4). Ultrasonography shows the hypertrophy in premaxillary region (Figure 5).

A provisional diagnosis of this malformation is hemihypertrophy, and we suspect Sturge-Weber



Figure 1: Asymmetrical enlargement of the right cheek of the patient causes gross facial deformity with significant disfigurement of the face



Figure 2: Enlarge right ear and left side ear which is of normal size

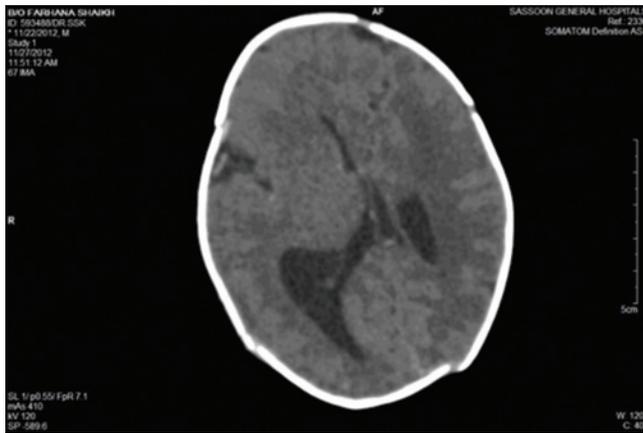


Figure 3: Computed tomography axial view revealed several right megancephaly with atrophy of left cerebral hemisphere causing midline shift to left the side and compressing lateral ventricle on the left side with ipsilateral dilatation of lateral ventricle. Widened non-fused sutures. Hypoplastic sinuses

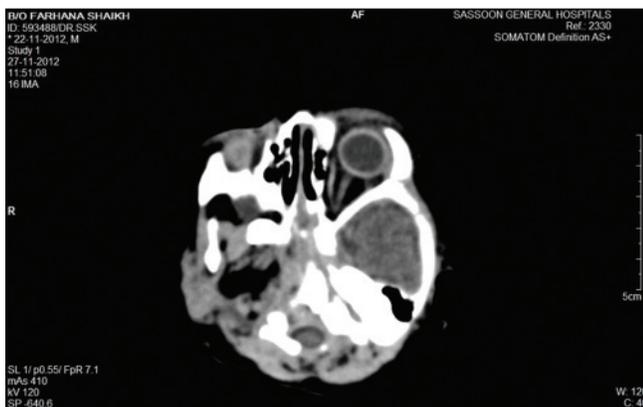


Figure 4: Hypoplastic sinuses in hemimegalencephaly there is an atrophy of sinuses seen



Figure 5: A diffused ill-defined swelling is noted on the right side of face involving the cheek region this appears to be a hypertrophied tissue

syndrome and ischemic hemiatrophy and unilateral polymicrogyria are a differential diagnosis of this anomaly.

The patient was prescribed carbamazepine, 20 mg/kg/day to be taken orally in 2-3 divided doses and physician advised not to stop the drug and recall after every 2 months. He has shown a very good response to seizure management for past 2 years against most of HME cases where patients have not responded to medical management.

Patient's parents have given their informed consent for publishing their case along with pictures and images.

DISCUSSION

HME (HME; also termed one-sided megalencephaly) is a moderately unusual but clinically impressive Defect of cortical development categorized by marked cerebral asymmetry (Friede, 1989). It is basically described by (Sims, 1835), and the author proposed that the cause of HME was the outcome of the augmented proliferation of progenitor cells in cell cycles. The cause of HME involves mechanisms that impede with late corticoneurogenesis with partial defeat of post-neurogenesis apoptosis (that is physiologic process) in the molecular layer.¹

Unusual midsagittal bands like structures in HME were exposed as aberrant fibres on FT reconstruction imaging. These kinds of structures were found in 50% of patients with HME studied. Diffusion tensor imaging is a beneficial way to show white matter structures which are not seen in routine imaging systems.²

In HME cases, exaggerated hemispheres demonstrated a large range of involvement. Variances of neuronal migration were present, and also there was an unevenly opposite connection among the severity of hemispheric participation and the degree of enlargement. This association is described via anticipated mechanism of a minor hemispheric injury during mid-to-end of second trimester.⁴

Some author studied cut-out section of HME soft tissue to examine whether the disorder might reveal somatic mutations affecting genes, which can hamper the growth of the brain. They concluded that HME tissue samples demonstrate trisomy of chromosome 1q, surrounding numerous genes, including AKT3, which is identified to control brain size.⁵

In focal lesions of HME, the post-surgery results are not that much excepting, but 40% improvement is noticed that is patient become seizure-free. But the purpose of surgery is preventing additional cognitive injury and developmental delay that is not achieved by doing surgeries like hemispheric surgery. And there are lots

of complications noticed during surgery, and those are avoided by minimal resections of hemisphere.⁶

Some author noticed several anatomic abnormalities outside the involved hemisphere such as extension of the homolateral brainstem, cerebellum and left lateral ventricle, in analysis of maximum numbers of HME cases.⁷

The investigator did a study on two Brazilian children who are suffered to Proteus syndrome. And common Proteus syndromic features diagnosed are uneven overdevelopment of tissue mass, reduced muscle strength, dermatologic defect and Intellectual disability. And in both of these cases, the refractory epilepsy is noted that is matched with Ohtahara syndrome. And HME along with disproportionate dispersal of facial fat are noted.⁸

The authors done a study on one case of HME with MRI imaging and suggested that the correct imaging techniques play an important role in the exact diagnosis of HME disorder and HME is hardly been found in adults patients. CT shows several pathologies, which may trigger epilepsy. Especially those associated with the acute presentations such as haemorrhage, infarction, and mass lesions as well as obvious malformations and calcified lesions. Often CT serves as first-line imaging in acute presentation where urgent treatment may be deserved.⁹⁻¹¹

Embolic hemispherectomy can be a supportive preoperative aide to surgical hemispherectomy in patients suffered from HME, and epilepsy. Early seizure control after embolic hemispherectomy is noted which is encouraging, but at this point investigators cannot propose embolic hemispherectomy as a primary therapeutic role, because right now this procedure is under trial, and additional research is required.¹²

In our case, the patient has HME with hypertrophy in the right cheek and the right ear is slightly large in size compared to the left ear and delayed milestones are seen. Epileptic seizures reported at the age of 6 months and seizures were controlled with medication. And the combination of these features is not seen in previously reported cases of HME.

CONCLUSION

HME is an unusual defect of the brain and complications are noted in during hemispheric surgery in a number of patients. So the medical management of seizures and proper oral prophylaxis is recommended for this anomaly.

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