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Hemimegalencephaly with prominent ipsilateral facial hypertrophy

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Abstract

Hemimegalencephaly is an uncommon congenital malformation with unilateral enlargement of the hemicerebrum. Here we report a 1 month old male child who presented at our neurosurgery outpatient clinic with recurrent seizures and facial asymmetry. CT and MRI of the brain revealed diagnostic characteristic features of hemimegalencephaly associated with ipsilateral facial congenital infiltrating lipomatosis.

Keywords

Hemimegalencephaly, Dyke-Davidoff-Masson syndrome, Nerve growth factor, Ventriculo-perotoneal shunt, intracranial pressure. Congenital-infiltrating lipomatosis

Introduction

Hemimegalencephaly [HME] is a congenital disorder characterized by abnormal cortical formation with hamartomatous overgrowth in all or a part of a cerebral hemisphere. HME results from either increased proliferation or decreased apoptosis of developing neurons.[1] This disorder is a cryptogenic congenital disorder, does not have a recognized racial or gender predilection and accounts for only 0.2% of cases of childhood epilepsy.[2] Here we describe a 1 month old boy with features of HME and unilateral soft and bony facial hypertrophy due to congenital infiltrative lipomatosis.

Case report

A 1 month old male child was born at 35 weeks of gestation by caesarian section. Birth weight was 3.300 kg and head circumference was 37 cm. He stayed in the neonatal intensive care unit (NICU) for 5 days and received anti-epileptic drugs due to neonatal convulsions. The anterior fontanel was soft upon physical examination and he was able to move all his limbs equally. Left side facial asymmetry was noticed (Figure 1 A, B, C, D, E). A computerized tomography (CT scan) and MRI of the brain were obtained which revealed left cerebral hemimegalencephaly, lateral ventricular dilatation (LVD) and contralateral displacement of the falx cerebri attachment (Figure 2 A, B, C).

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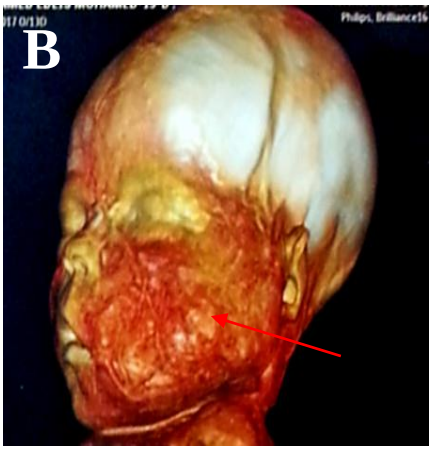

Figure 1 A, B
asymmetrical enlargement of the left cheek of the patient causing gross facial deformity with significant disfigurement of the face

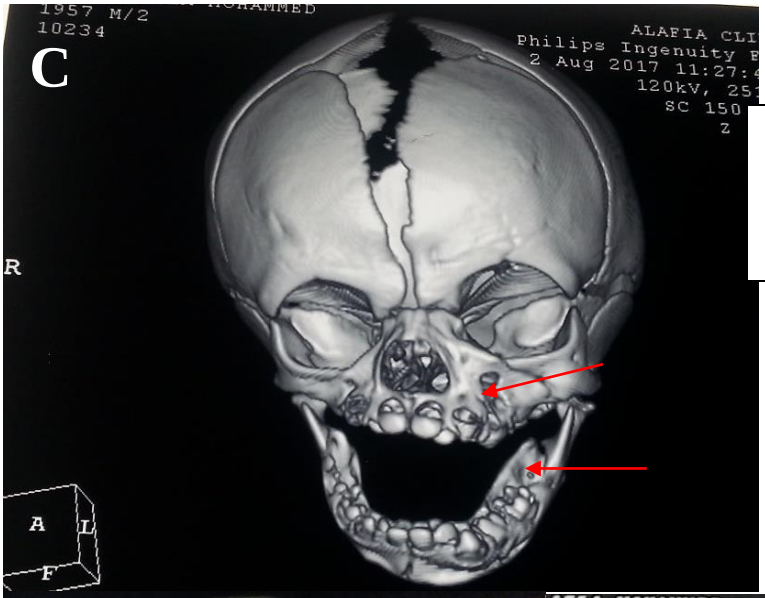

Figure 1 C 3D CT skull and soft tissue shows enlargement of the left mandible and maxilla [Red arrows]

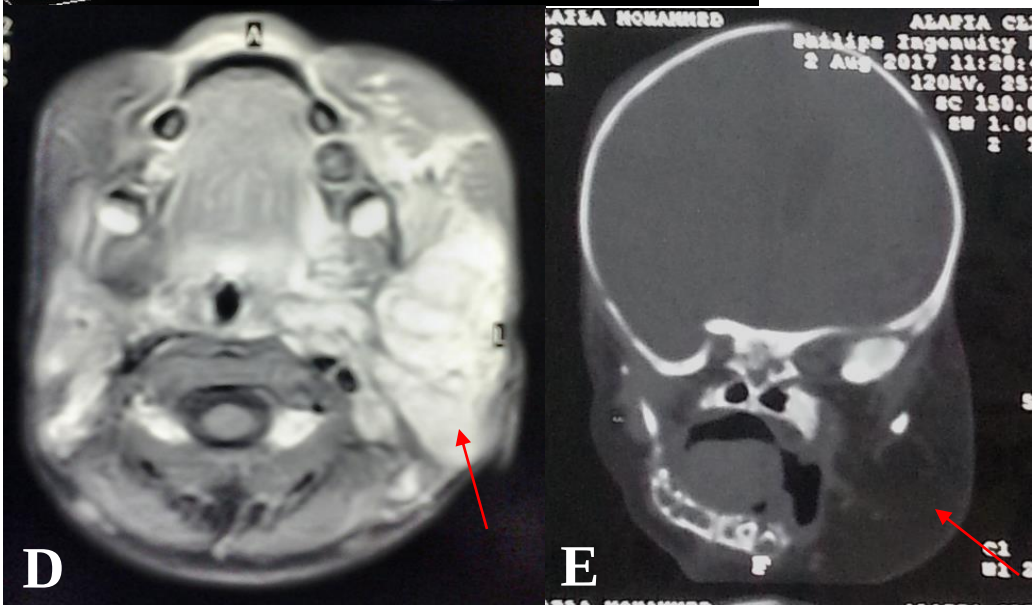

Figure 1 D, E (D) axial contrast MRI demonstrates an extensive, non-encapsulated left facial mass with fat signal located in the cheek **(E)** facial CT showing lipomatous infiltration of left side [Red arrows]

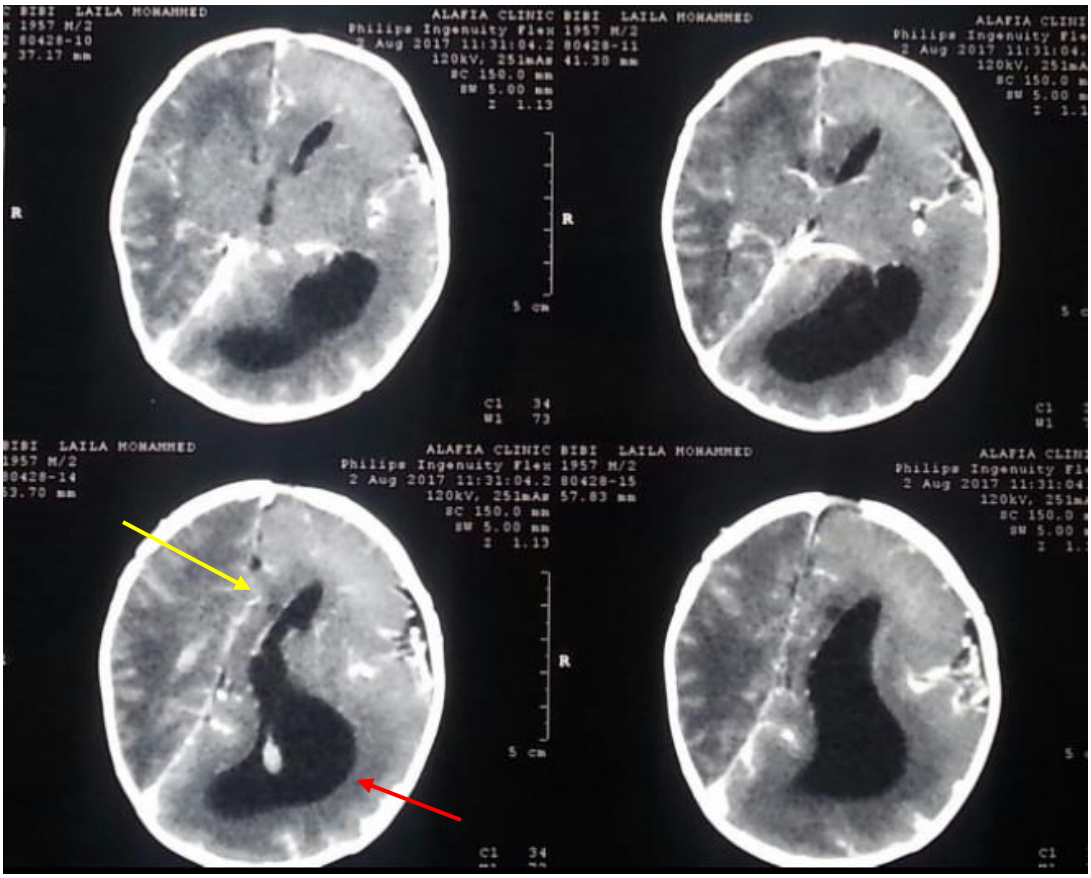

Figure 2 [A]….

Contrast CT scan of the brain showing left cerebral megalencephaly with lateral ventricular dilatation (LVD) [red arrow] and contralateral falx displacement (FD) [yellow arrow]

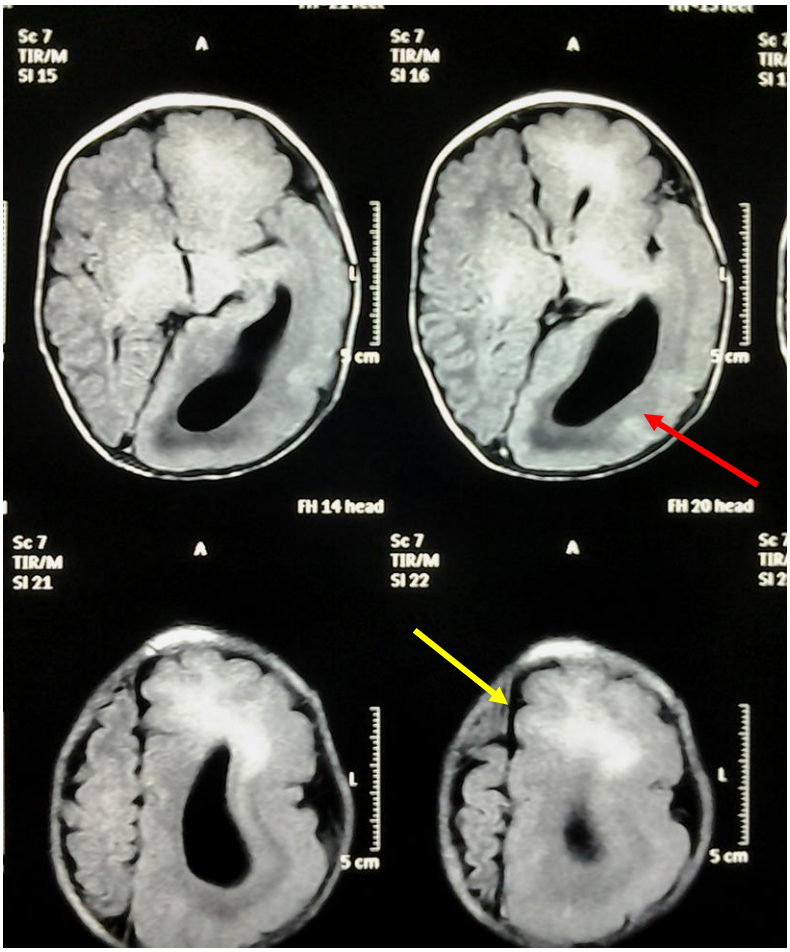

Figure 2 B

MRI T1-weighted axial and coronal view of the brain showing left cerebral megalencephaly with lateral ventricular dilatation [Red arrow] and contralateral falx displacement [yellow arrow]

Discussion

Hemimegalencephaly is a rare condition characterized by enlargement of all or parts of a cerebral hemisphere. The affected side of the hemisphere may have localized or diffuse neuronal migration defects, with areas of polymicrogyria, pachygyria, and heterotopia.[5-10]The etiology of this condition is unknown, but it could be due to abnormalities of neuronal differentiation and cell migration in a single cerebrum hemisphere. Nerve growth factor (NGF), which is produced and released by brain cells, is involved in the regulation of choline acetyltransferase activity, which is highly expressed in regions of the central nervous system innervated by the magnocellular cholinergic neurons of the basal forebrain including the hippocampus, olfactory bulb, and neocortex.[12-13] Antonelli et al demonstrated increased tissue levels of NGF and numerous high-affinity NGF-receptor-positive cells in hemimegalencephaly tissues compared with control brain tissues and these changes appear to be also associated with abnormal NGF-receptor expression in subcortical blood vessels. In addition, the low level of cortical choline acetyltransferase immunoreactivity is strongly suggestive of a dysregulation in the NGF differentiative activity in this site that could lead to the pathogenesis of HME.[14]

Facial asymmetry associated with HME has been described in few cases in literature. Aydingoz et al reported a case of hemimegalencephaly and ipsilateral congenital-infiltrating lipomatosis of the face (CILF) in a three month old infant who had ipsilateral scalp lipoma.[15] CILF includes soft-tissue and skeletal hypertrophy, premature dental eruption, and regional macrodontia.[16] The exact mechanism of the association between ipsilateral congenital-infiltrating lipomatosis of the face and HME is still unclear. Despite the fact that CILF is a benign condition, radical excision is very difficult due to high recurrence rate of regrowth.[17] The combination of HME and ipsilateral facial bony enlargement of the maxilla and the mandible due to CILF as in our case has not been illustrated before and this observation will expand the spectrum of this rare condition.

The imaging study of choice for diagnosis of this condition is magnetic resonance imaging (MRI). The radiographic features of the affected cerebrum include increased lateral ventricle size (may sometimes be small), shallow sulci enlarged gyri, enlarged thickened calvaria, contralateral displacement of the posterior falx or white matter calcification.[1] Other abnormalities found on MRI which can accompany this condition include abnormal gyral pattern with a thick cortex, gliosis in the white matter on the affected side, and abnormal myelination.[3,4] Dyke-Davidoff-Masson syndrome (DDMS) is an uncommon condition, which was reported first by CG Dyke**,** LM Davidoff and CB Masson in 1933.[11] This rare disorder must be in the differential diagnosis because the majority of patients with DDMS usually present with refractory seizures and facial asymmetry.

The treatment of HME is targeted to the control of epilepsy, which can be challenging to manage medically. In refractory cases, hemispherectomy is the treatment of choice and results in seizure control in at least 60% of cases, when carefully selected. Patients with contralateral malformations have a poorer surgical outcome.[2] In our case the plan of management is to follow up the patient closely in our neurosurgery clinic as the patient may need ventriculo-peritoneal shunt (V-P SHUNT) in the future if signs of high intracranial pressure are noticed. Regular follow up in neurology department and checking the anti-epileptic drugs level for optimal seizure control are essential.

Conclusion

We have described a rare case of hemimegancephaly, presented with infantile convulsions and unusual bony facial asymmetry due to congenital infiltrative lipomatosis. The main treatment in these cases is seizure control, either by anticonvulsant medications or hemispherectomy as last resort.

Acknowledgments

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