Hemimegalencephaly with Seizure: A Rare Congenital Malformation in a 22 months Old Boy

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Case Report

Keywords: Hemimegaloencephaly, Pediatric Age Group, Seizure

Posted Date: April 5th, 2021

DOI: https://doi.org/10.21203/rs.3.rs-379150/v1

Abstract

Isolated hemimegalencephaly (HME) is a rare congenital malformation of brain development, remarkable for its extreme asymmetry, and is characterized by the overgrowth of part or whole hemisphere. The enlarged hemisphere is manifested by hamartomatous characteristics with a dysplastic cell array of atypical morphology. Traditionally the genetic theories regarding the pathogenesis of HME are considered due to disturbance in cell signaling during neuroblast migration, cell differentiation, and proliferation, pattering, and symmetry. HME can present as isolated or associated with several neurocutaneous syndromes. The clinical picture varies depending on the severity of the malformation; however, HME patients typically exhibit refractory epilepsy, macrocephaly, colpocephaly, global developmental delay, intellectual disability, hemibody hypertrophy, and hemiparesis. Early diagnosis is crucial because despite neuroimaging and pathologic evidence, hemimegalencephaly sometimes still is unrecognized. Also, misdiagnosed as obstructive hydrocephalus or cerebral neoplasm can lead to unnecessary surgical procedures. Although hemispherectomy has high morbidity, it is recommended early for patients with severe, intractable epilepsy. We report a diagnosed case of a 22-month-old boy hemimegalencephaly who presented with seizures attack and was successfully treated with antiepileptic medications.

Background

Hemimegalencephaly is a rare cortical malformation marked by the enlargement of one cerebral hemisphere. The pathogenesis is not fully understood although there is a link of gene and a link with mTOR has been found. This disease can be presented with other diseases but most commonly represents with seizure. Some times seizures are precipitated with drugs, but sometimes not, then hemispherectomy is commonly done.

In our case it is a 22 months old boy with hemimegalencephaly and seizure. Seizures are precipitated after giving bolus dose of levetiracetam (20mg/kg) at ED. He was started on topiramate 30mg twice a day, clonazepam 0.25mg twice a day, injectable lorazepam in case of seizure activity, and acetaminophen as needed. As it is a rare case, so we are presenting this case through this case report.

Case Report

A 22-month-old boy with a past medical history of hemimegalencephaly, infantile spasms, developmental delay, and complex partial seizures presented to the emergency department with increased frequency of seizure, subjective fever, diarrhea, rhinorrhea, and rash from the last 2 days. Previously his seizure episode lasts for few seconds and was consistent with staring and going limp. Now, it lasts for one minute and consists of falling to the floor associated with abnormal movements. On examination, his weight was 12.3 kg, blood pressure was 110/63 mmHg, pulse rate was 116/minutes, the temperature was 98.9 F, SpO2 was 100% and respiratory rate was 23/minute. The rest of the examination findings were unremarkable. Baseline investigations along with metabolic profile was sent, the results of which were unremarkable (table 1).

Table 1: Laboratory Findings

Test	Result
Hemoglobin	12.4 g/Dl
Total leukocyte count	8.8(x10^9/l)
RBC	4.3 (x10^{12}/l)
Platelets	290x10^9/l
Prothrombin time	12 seconds (12 seconds control)
Activated partial thromboplastin time	28 seconds (28 seconds control)
Alanine aminotransaminase (ALT)	32 U/L
Aspartate aminotransaminase (AST)	28 U/L
Alkaline phosphatase	43 U/L
Total bilirubin	0.2 mg/dl
Blood urea	21 mg/dL
Creatinine	0.4
Sodium	137.2 mEq/L
Potassium	3.92 mEq/L
Chloride	103 mEq/L
S.calcium	9.8 mg/dL
Total cholesterol	115 mg/dL
Triglycerides	109 mg/dL
High-density lipids	41 mg/dL
Low-density lipids	170 mg/dL

He was given a bolus dose of levetiracetam (20mg/kg) at ED and no seizure activity was noted after that. He was started on topiramate 30mg twice a day, clonazepam 0.25mg twice a day, injectable lorazepam in case of seizure activity, and acetaminophen as needed. Electroencephalogram was advised which showed poor regional organization and lack of a posterior dominant rhythm as well as continuous epileptiform activity in the right occipital-temporal-parietal region. MRI brain with intravenous contrast was done which showed asymmetric enlargement of the right cerebral hemisphere with hyper myelination, mild ventriculomegaly, and displaced posterior falx most in keeping with hemimegalencephaly along with asymmetric right-hemispheric hyperperfusion which might be related to

recent seizure activity (Figure 1). He was admitted to the ward and was kept under observation for the next 48 hours. No seizure activity was noted and he was discharged home on antiepileptic medications.

Discussion

Hemimagalencephaly is a rare genetic condition which is associated with Seizures. It is a rare cortical malformation marked by the enlargement of one cerebral hemisphere. (1) The pathogenesis of this disease is thought to be characterized by abnormal activation of the mTOR signaling pathway. (1) So mTOR inhibitor like everolimus plays a key role in the treatment sometimes. In an Youtube Video lecture from UCLA Health (2) it has been also mentioned that drugs can't control this seizure in many cases, then Hemispherectomy is done to save the patient from severe seizures.

Currently, hereditary pathogenesis hypotheses and modern histopathology offer a foundation for this complex malformation as a key disruption in neuronal lineage, division, and proliferation, interfering with a disturbance in body symmetry gene expression, and initiation earlier than radial neuroblast migration. (3)

Neurofibromatosis, epidermal nevus syndrome, Ito's hypomelanosis, and Klippel–Trenonay–Weber syndrome are all neurocutaneous conditions that may induce Hemimegalencephlaly. A serious and drug-resistant epilepsy typically dominates the clinical image. Macrocrania, moderate/severe mental retardation, unilateral motor deficiency, and hemianopia are other typical findings. (4) The EEG demonstrates a number of irregular patterns, the most prominent of which are inhibition bursts and/or hemihypsarrhythmia. While standard observations on neuroimaging and histologic investigations (enlarged hemisphere, malformed ventricular system, modification of regular gyration) are common, creating a differential diagnosis with other disorders of neuronal and glial proliferation may be challenging. (4) In later age group it can show dementia also. (5)

Last, we had presented a case of WHO GRADE III Anaplastic astrocytoma in an 72 years old male (6), in that patient the treatment was not so much complicated like this one.

In Our case, a 22 months old boy came with a seizure. The MRI image shows a right sided enlargement of the cerebral hemisphere with hypermyelination and ventriculomegaly. He was given a bolus dose of levetiracetam (20mg/kg) at ED and no seizure activity was noted after that. He was started on topiramate 30mg twice a day, clonazepam 0.25mg twice a day, injectable lorazepam in case of seizure activity, and acetaminophen as needed. So, fortunately the seizure stopped after induction of the drug. But in many cases seizure does not precipitate with drugsm then Hemispherectomy is the treatment of choice. This surgery has some adverse effects like, Post operative bleeding, Surgical infections, Meningitis which can be prevented by prescribing steroids and hydrocephalous (may be early or delayed)

Conclusion

This is a case of Hemimegalencephaly with Seizure. The Seizures are precipitated after administration of drug. The patient was discharged with antiseizure drugs in prescription.

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Declarations

Conflict of Interest: Authors doesn't have conflict of Interest

Funding: No Funding is attached

Patient's Consent: Patient's consent has been taken for publication

Acknowledgements: We are thankful to the professors of the Universities and our parents.

Figures

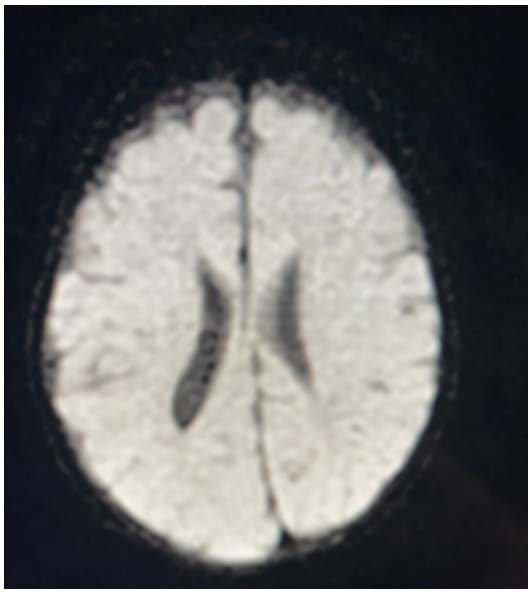


Figure 1

Figures of MRI and CT shows the right Cerebral enlargement i.e Hemimegalencephaly with causes herniation towards the left cerebral hemisphere and enlargement of right lateral ventricle.

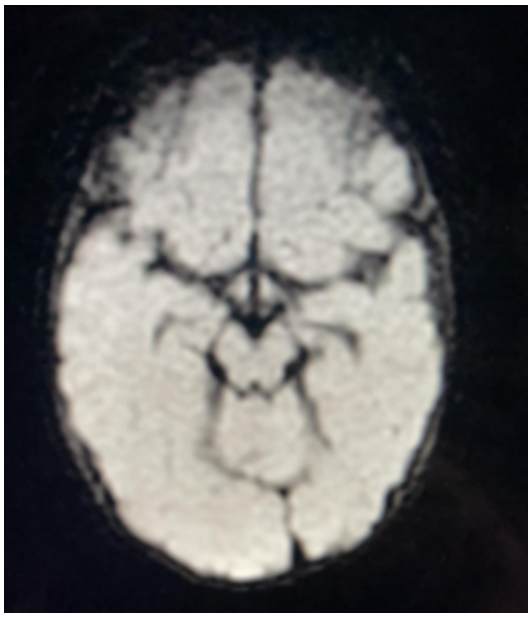
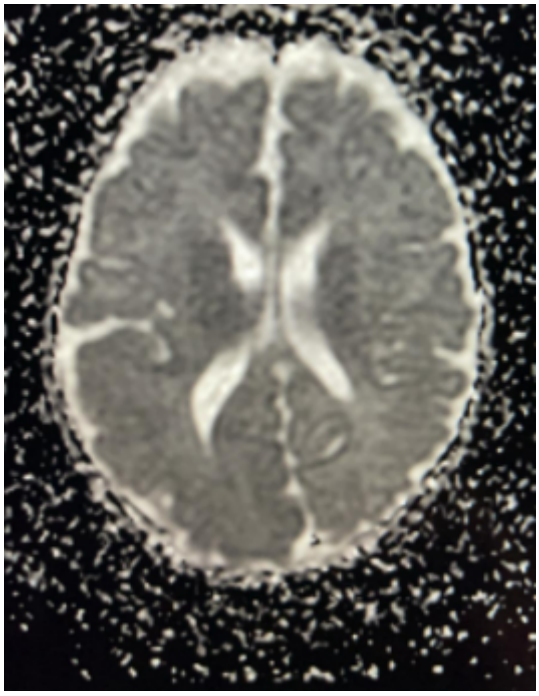


Figure 2

Figures of MRI and CT shows the right Cerebral enlargement i.e Hemimegalencephaly with causes herniation towards the left cerebral hemisphere and enlargement of right lateral ventricle.



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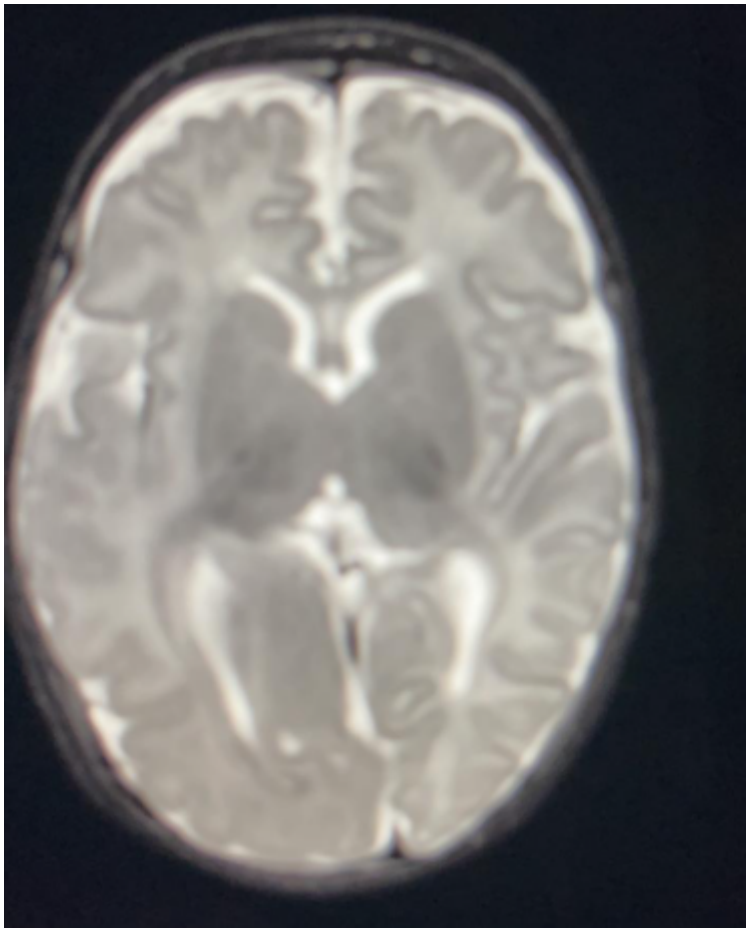

Figure 4

Figures of MRI and CT shows the right Cerebral enlargement i.e Hemimegalencephaly with causes herniation towards the left cerebral hemisphere and enlargement of right lateral ventricle.

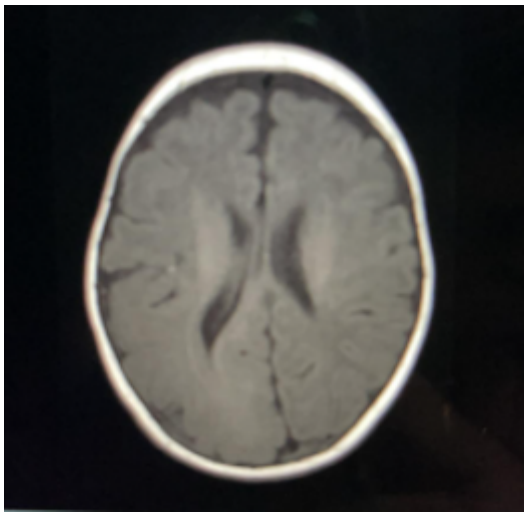

Figure 5

Figures of MRI and CT shows the right Cerebral enlargement i.e Hemimegalencephaly with causes herniation towards the left cerebral hemisphere and enlargement of right lateral ventricle.

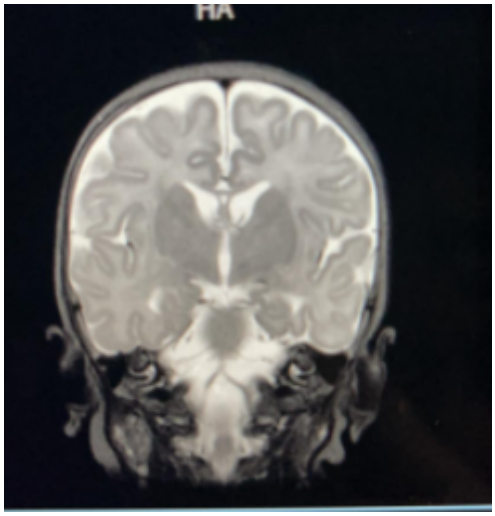

Figure 6

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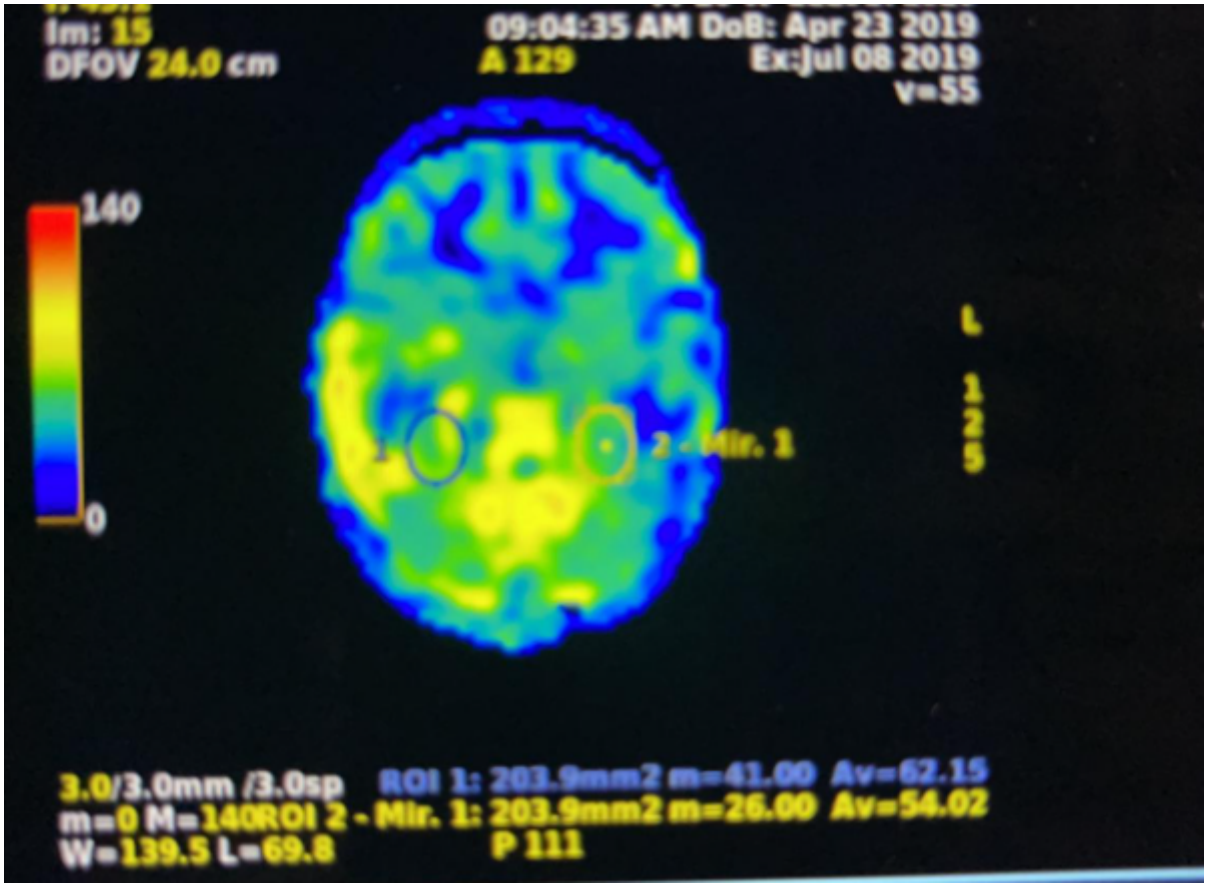

Figure 7

Figures of MRI and CT shows the right Cerebral enlargement i.e Hemimegalencephaly with causes herniation towards the left cerebral hemisphere and enlargement of right lateral ventricle.