ORIGINAL RESEARCH PAPER

Radiodiagnosis

PERIVENTRICULAR NODULAR HETEROTOPIA ASSOCIATED WITH POLYMICROGYRI AND PARTIAL AGENESIS OF CORPUS CALLOSUM

KEY WORDS: Periventricular nodular heterotopias, Grey matter, Lateral ventricles

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ABSTRACT

Periventricular nodular heterotopia (PNH)is the most common form of grey matter heterotopia which is characterized by nodules of grey matter located along the lateral ventricles. It is mostly bilateral and occurs due to improper immigration of neurons during early development of fetal brain.

The patient most commonly presents with epilepsy and sometimes with developmental delay. Two different types of periventricular nodular heterotopia have been described in literature, namely bilateral and unilateral PNH. Bilateral PNH follows an X-linked dominant inheritance pattern. Most affected individuals are females. There are only a few reports of affected males, since X-linked inheritance means that development of most of the male embryos fails to progress.

We report a case of 33 year old male presented to us with history of headache , seizure and delayed milestones since 20 years.

INTRODUCTION

Periventricular heterotopia is a condition in which the neurons fail to migrate properly during the initial development of the fetal brain, from 6th week to 24th week of gestation. Few of the neurons fail to migrate to their proper position and form clumps around the ventricles in brain .

It becomes evident when patient presents with seizure, which is usually noticed during 2nd decade of life. Affected individuals may have normal intelligence, although some have developmental delay like difficulty with reading , spelling and movement disorders.

CASE HISTORY

We report a case of 33-year-old male who presented with a history of headache , seizure for the past 20 years .The onset of convulsions was at the age of 10 years occurring with a frequency of 2-3 episodes per month. He was initially treated with sodium valproate with a starting dose which was, gradually increased . In view of partial response to valproate, phenobarbitone was also added. Despite the use of two antiepileptic drugs, his convulsions persisted . His prenatal history was unremarkable. He had history of delayed developmental milestones. On examination, his intellectual ability was also normal. There was no family history of seizures. There were no signs of cognitive decline , progressive ataxia or cerebellar signs.

Patient was then advised MRI brain for further evaluation, which revealed asymmetric moderate dilatation of left lateral ventricle as compared to right with nodular appearance of its lateral margin which followed gray- matter signal intensity suggestive of periventricular nodular heterotopia.

Subtle similar changes were also noted in atrium and occipital horn of right ventricle.

There was increase in number of mildly thickened gyri in left fronto-temporo-parietal region and right temporo-parietal region suggestive of polymicrogyri.

Posterior portion of body and splenium of corpus callosum appeared hypoplastic.

Persistent cavum septum pellucidum was noted A 3.3 x 2.5 cm sized extra axial altered CSF intensity lesion was noted in the posterior fossa without any significant scalloping of occipital bone which was more likely suggestive of mega cisterna magna than arachnoid cyst

DISCUSSION:

Gray matter heterotopias are common malformations of cortical development and result due to improper immigration of neurons during early development of fetal brain.

Since the last two decades, with the advent of MRI, the presence of heterotopia is detected much more earlier. Bilateral and symmetrical subependymal gray matter heterotopia (SEH) is the most common form of SEH and is characterized by familial occurrence, a clear female predominance and a positive family history of epilepsy. Therefore, genetic factors are believed to play a major role in its etiology.

Patients with bilateral symmetrical SEH are usually normal individuals affected by focal epilepsy, frequently drug-resistant but not characterized by high frequency seizures. Bilateral symmetrical SEH is associated with posterior fossa malformations such as cerebellar hypoplasia and megacisterna magna.

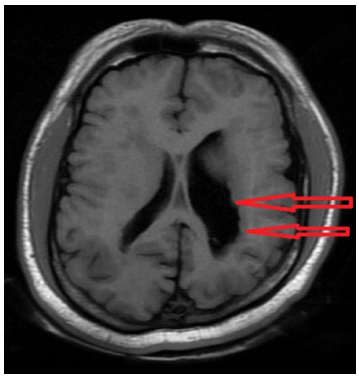
On CT scan irregularities along the lateral margins of the lateral ventricles may be the only clue for the diagnosis.

MRI is better and investigation of choice and can accurately delineate the morphology, distribution and extent. On MRI, subependymal heterotopias appear as ovoid lesions within the subependymal location. No evidence of perilesional edema or contrast enhancement is seen.The signal intensities of the nodules are identical to those of gray matter on all sequences, including contrast-enhanced series. EEG may be normal or with focal abnormalities

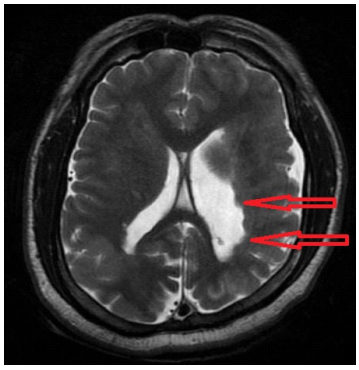
CONCLUSION-

The heterotopias are a rare subgroup of cortical malformations characterized by abnormal neuronal migration during the foetal life and it usually presents as refractory epilepsy.

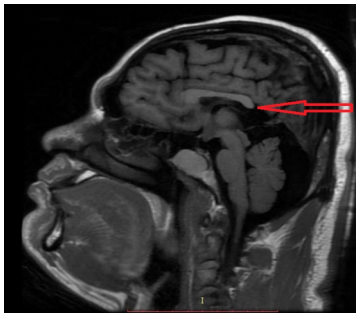
Being X-linked dominant, very few cases of bilateral periventricular nodular heterotopia associated with polymicrogyri and partial agenesis of corpus callosum have been reported in males.



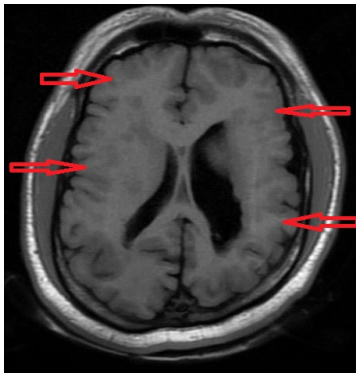
AXIAL T1W image of the brain showing asymmetric moderate dilatation of left lateral ventricle as compared to right with nodular appearance of its lateral margin which follows gray matter signal intensity suggestive of periventricular nodular heterotopias.



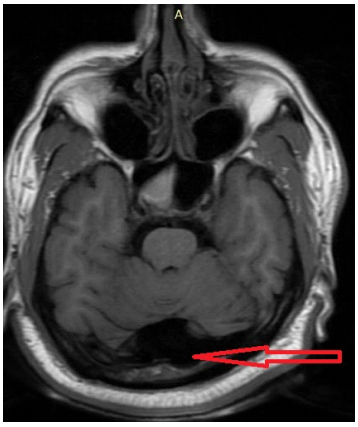
AXIAL T2W image of the brain showing similar features.



SAGITAL T1W image of the brain showing hypoplastic posterior portion of body and splenium of corpus callosum



AXIAL T1W image of the brain showing increase number of mildly thickened gyri in left fronto-temporo-parietal region and right temporo-parietal region suggestive of polymicrogyri



AXIAL T1W image of brain showing extra axial altered CSF intensity lesion in the posterior fossa without any significant scalloping of occipital bone suggestive of mega cisterna magna more likely than arachnoid cyst.

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