Case Report

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Journal of Rare Disorders: Diagnosis & Therapy
ISSN 2380-7245

2015
Vol. 1 No. 3:22

DOI: 10.21767/2380-7245.100022

MRI Features of a Rare Entity -Cystic Leukoencephalopathy without Megalencephaly

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Citation: Soni N, Biswas S, Kumar S. MRI Features of a Rare Entity -Cystic Leukoencephalopathy without Megalencephaly. J Rare Dis Diagn Ther. 2015, 1:3.

Abstract

Context: Cystic leukoencephalopathy without megalencephaly is a very rare entity in children, presenting clinically with non-progressive encephalopathy, normo or microcephaly, and early onset of severe psychomotor impairment. To date, fewer than 30 cases have been described. The association of this entity with bilateral sensorineural hearing loss has been reported previously in literature only in three case reports.

Case report: Here we report the case of a 4 year old normocephalic male child presenting with mental retardation and bilateral congenital deafness. An MRI (3 Tesla) study of brain revealed bilateral symmetrical anterior temporal lobe subcortical cysts, patchy multifocal subcortical and pericystic white matter hyperintensities and dilatation of temporal horns of both lateral ventricles. A BAER test confirmed the presence of bilateral sensorineural hearing loss in the child.

Conclusion: The clinical signs and cranial MRI findings were similar to a handful of reports published in literature and hence led to the diagnosis of this rare disease. Furthermore we report the association of this entity with bilateral sensorineural hearing loss.

Keywords : Cystic leukoencephalopathy; Megalencephaly; Magnetic resonance imaging; Anterior temporal lobe cysts; Sensorineural hearing loss

Abbreviations : MRI: Magnetic Resonance Imaging; BAER: Brainstem Auditory Evoked Response; LBATC: Leukoencephalopathy with Bilateral Anterior Temporal lobe Cysts; FIESTA: Fast Imaging Employing Steady state Acquisition; CMV: Cytomegalovirus

Received: September 28, 2015; **Accepted:** October 14, 2015; **Published:** October 22, 2015

Introduction

Cystic leukoencephalopathy without megalencephaly, otherwise known as Leukoencephalopathy with bilateral anterior temporal lobe cysts (LBATC) is a rarely diagnosed entity. Clinically the disease presents in early childhood with moderate to severe psychomotor retardation in an apparently non-progressive clinical course with normo- or microcephaly. Previously it was thought to be due to congential Cytomegalovirus infection of the developing brain. However recent genetic studies have linked it to a mutation of the RNAse T2 gene [1-4]. The classical MRI findings in brain include bilateral anterior temporal **lobe cysts in a subcortical location with multifocal** white matter hyperintensities, pericystic abnormal myelination and symmetric lesions in the periventricular regions [2,5]. Enlargement of the inferior horns of both lateral ventricles has also been reported [1]. The association of this rare disease with bilateral sensorineural hearing loss deafness has been previously reported only in three case reports [1-3].

In some aspects, this condition resembles other leuko-encephalopathies such as “megalencephalic leukoenceph-alopathy with subcortical cysts (van der Knaap disease)” and “leukoencephalopathy with vanishing white matter” due to the presence of subcortical cysts and white matter abnormalities. However the clinical presentation and other neuroimaging features are distinct [2]. Here we report a case of cystic leukoencephalopathy without megalencephaly in association with bilateral sensorineural hearing loss, the combination of which is indeed very rare.

Case Report

A 4 year old male child presenting with mental retardation and congenital deafness was referred to our department for an MRI of the brain. He was a single child born after an uneventful pregnancy to non-consanguineous parents. There was no history of perinatal asphyxia. On examination the head circumference was normal for age (measuring 50 cm, i.e., 50th percentile for age). An MRI of the brain showed bilateral symmetrical anterior temporal lobe subcortical cysts with pericystic white matter hyperintensities and dilatation of the temporal (inferior) horns of both lateral ventricles **(Figure 1)**. The frontal and occipital horns revealed no obvious abnormality. There was no diffusion restriction or post contrast enhancement in the cysts **(Figure 2)**. Multifocal areas of subcortical white matter hyperintensities were noted in the rest of the temporal lobes and bilateral parietal regions **(Figure 3)** without any periventricular or parenchymal calcifications. No obvious cortical abnormalities were noted. Axial FIESTA of the brain showed normal course and caliber of bilateral 7th and 8th cranial nerves in the cisternal and intracanalicular segments with normal appearing internal ear structures on both sides. Metabolic screening in the child was normal. Subsequently a BAER study was done which confirmed the presence of bilateral sensorineural hearing loss.

Discussion

Cystic leukoencephalopathy without megalencephaly was first described by Olivier et al. In 1998 and since then, fewer than 30 cases have been described [2]. It is characterized by an apparently non-progressive condition characterized by severe psychomotor delay with variable degrees of tone and reflex abnormalities, normo- or microcephaly. Patients with cystic leukoencephalopathy without megalencephaly are severely involved from the beginning with majority lacking speech development. Most of the patients achieve walking with aid, but some can only sit or crawl [5]. Other variable features include seizures, spasticity, athetoid hand movements, dystonia, nystagmus, and hearing loss.

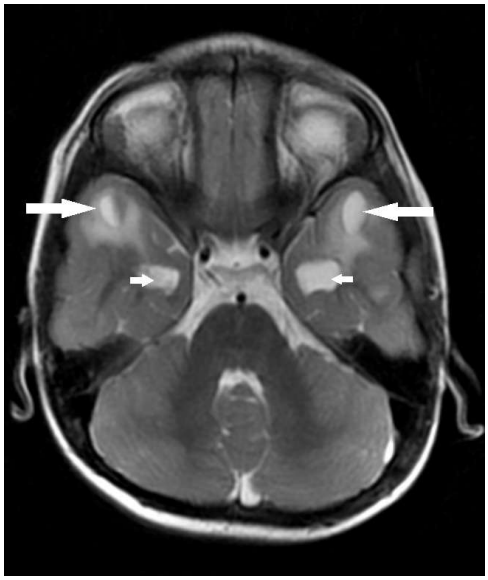

Figure 1 Axial T2 (TR/TE , 5600/100 ms) image shows the bilateral anterior temporal lobe subcortical cysts with pericystic white matter hyperintensities (long arrows) and dilated temporal horns of both lateral ventricles (short arrows).

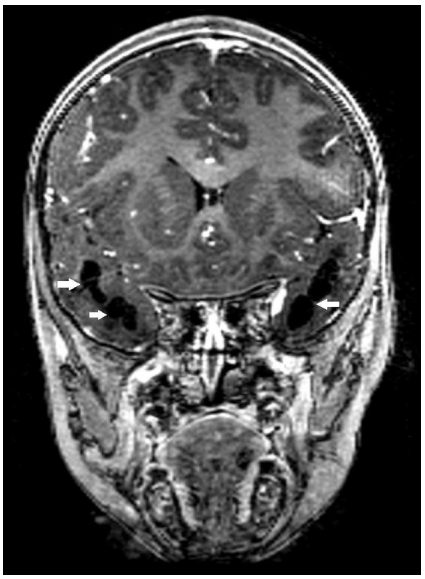

Figure 2 Coronal T1 image with contrast (TR/TE, 1300/12 ms) shows bilateral non enhancing anterior temporal lobe cysts (arrows).

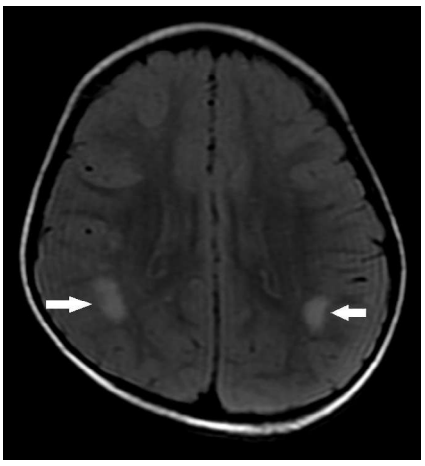

Figure 3 Axial T2 FLAIR (TR/TE/TI, 8800/100/2250 ms) image shows focal subcortical white matter hyperintensity (arrows) in bilateral parietal lobes.

The classical MRI finings in brain include extensive subcortical cysts within the anterior temporal lobes with white matter hyperintensities. The signal intensities of the cysts were identical to those of cerebrospinal fluid [6]. Reported findings also include dilatation of temporal horns of lateral ventricles, pericystic abnormal myelination and symmetric lesions in the periventricular regions [5].

Previously it was thought to be a sequelae of congenital CMV infection, however current genetic analysis have shown that loss-of-function mutations in the gene encoding the RNASET2 glycoprotein leads to cystic leukoencephalopathy [1]. Like CMV infection , RNAse T2 deficiency interferes with brain development

and myelination through angiogenesis or RNA metabolism.

The differential diagnosis in our case includes Megalencephalic leukoencephalopathy with subcortical cysts (also known as Van der Knaap disease), leukoencephalopathy with vanishing white matter (Vanishing white matter disease) and congenital CMV infection.

Megalencephalic leukoencephalopathy with subcortical cysts is characterized by macrocephaly developing within the 1st year of life with mild neurological deterioration initially but disproportionate and dramatic MRI changes. These include a typical pattern of diffuse white matter lesions and swelling with invariable anterior temporal and frontoparietal subcortical cysts. Genetic mutation of MLC 1 gene has been found to be responsible for this entity [7].

Patients with vanishing white matter disease have extensive cystic changes in the cerebral white matter; however the temporal lobes are not preferentially involved. Clinically it is characterized by a mild initial course, with later deterioration and bouts of sudden decline in function [2].

Children affected by congenital CMV infection usually present with microcephaly, seizures and delayed developmental milestones [7]. SNHL is usually not present at birth but develops later by 2 years of age [2]. The classical MRI findings include periventricular and anterior temporal lobe cysts, ventriculomegaly with periventricular calcifications and migration abnormalities (in the form of pachygyria, polyicrogyria, lissencephaly). Focal or confluent white matter hyperintensities may also be seen, often with a parietal predominance.

In summary this case report clearly demonstrates the classic MRI findings of cystic leukoencephalopathy without megalencephaly and its association with bilateral sensorineural hearing loss. Further testing for mutation defects of RNAseT2 gene can help to establish the genetic basis of this rare entity.

Conflict of Interest

The authors declare that they have no financial or personal relationship(s) that may have inappropriately influenced them in writing this article.

Authors' Contributions

Shamick Biswas was responsible for conception, design and drafting of the article and MRI image acquisition. Neetu Soni and Sunil Kumar were responsible for revising the article critically for intellectual content, analysis and interpretation of images and for final approval of the manuscript.

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