Canavan disease: MRI with pathognomic MR spectroscopy findings
Published on 08.10.2019

DOI: 10.35100/eurorad/case.16489
ISSN: 1563-4086
Section: Paediatric radiology
Area of Interest: Head and neck Paediatric
Procedure: Diagnostic procedure
Imaging Technique: MR
Imaging Technique: MR-Diffusion/Perfusion
Imaging Technique: MR-Spectroscopy
Special Focus: Congenital Genetic defects Case Type: Clinical Cases
Authors: Dr Jay Satapara1, Dr Hiral Parekh2, Dr Nandini Bahri3
Patient: 18 months, female

Clinical History:
An 18-months old female child presented with macrocephaly and developmental delay. Patient was only child and had no siblings. Patient had normal birth history and had no history of previous hospital admission. On clinical examination macrocephaly was evident with delayed developmental milestones. Patient was sent for MRI.

Imaging Findings:
Magnetic Resonance Imaging (MRI) showed altered signal intensity areas which appeared hypointense on T1-weighted image, hyperintense on T2-weighted image and showed restricted diffusion on diffusion weighted image (DWI). Involved areas were subcortical and periventricular white matter, bilateral thalamus, bilateral globus pallidus, splenium of corpus callosum and dorsal part of midbrain and pons with sparing of bilateral caudate and putamen nucleus, bilateral internal capsule, ventral part of crus cerebri of midbrain and ventral part of pons. MR spectroscopy showed markedly elevated NAA and NAA-creatine ratio, which is pathognomic for canavan disease.

Discussion:
Canavan disease is a rare autosomal recessive leukodystrophy, caused by genetic mutation involving short arm of 17th chromosome. Which leads to deficiency of N-acetyl aspartoacylase, an enzyme required for myelin synthesis [1, 2]. In general population prevalence is 1:100000 [3]. Patients present with macrocephaly and neurological deficit usually during infancy and may have normal birth history. Neurological deficit ranges from developmental delay, lethargy and hypotonia to spasticity, blindness, seizures, decerebrate posturing and eventual leading to death. Sometimes in early stage macrocephaly could be the only finding evident [4]. Diagnosis is based on detection of increased N-acetyl aspartic acid (NAA) in urine, CSF and brain. CT and MRI are the initial investigation of choice [3]. In early stage CT shows megalencephaly with decreased attenuation of subcortical white matter without abnormal
contrast enhancement. In late stage CT shows signs of cortical atrophy in the form of dilatation of ventricles and prominent sulci and basal cisterns [2]. MRI provide more details of white matter involvement due to excellent gray white matter resolution. T1 weighted images show low signal intensity whereas T2 weighted and FLAIR images show high signal intensity within subcortical white matter, globus pallidus and thalamus usually sparing corpus callosum, putamen nucleus and internal capsule. As the disease progresses, periventricular white matter is also involved with resultant dilatation of ventricles. Diffusion-weighted images show restricted diffusion within involved white matter [2-8]. MR spectroscopy shows increased NAA and NAA-creatine ratio, which is pathognomonic for canavan disease [3]. Canavan disease is fatal condition; death occurs around 5 years of age. No definite treatment is available for the disease [2]. Generic therapy is being tried [3]. Take home message: Imaging with MRI and MR spectroscopy individually or in conjunction with each other leads to diagnosis of canavan disease in almost all instances. Written informed patient consent for publication has been obtained.

Differential Diagnosis List: Canavan disease, Alexander disease, Pelizaeus-Merzbacher disease, Adrenoleukodystrophy, Metachromatic leukodystrophy

Final Diagnosis: Canavan disease

References:

Adachi M, Schneck L, Cara J, Volk BW. Spongy degeneration of the central nervous system (van Bogaert and Bertrand type; Canavan's disease). A review. Hum Pathol. 1973 Sep;4(3):331-47. (PMID: 4593851)


Description: T2-weighted axial image showed hyperintense areas involving subcortical and periventricular white matter, bilateral thalamus, bilateral globus pallidus with sparing of bilateral caudate and putamen nucleus and bilateral internal capsule. 

Origin: © Department of Radio diagnosis, G. G. Hospital, M. P. Shah Medical College, Jamnagar, India.
Description: T2-weighted sagittal image showed hyperintense areas involving subcortical and periventricular white matter, splenium of corpus callosum and dorsal part of midbrain and pons.

Origin: © Department of Radio diagnosis, G. G. Hospital, M. P. Shah Medical College, Jamnagar, India.
Description: T2-weighted axial image showed hyperintense areas involving subcortical and periventricular white matter and dorsal part of midbrain with sparing of ventral part of crus cerebri of midbrain. Origin: © Department of Radio diagnosis, G. G. Hospital, M. P. Shah Medical College, Jamnagar, India.
Description: T1-weighted axial image showed hypointense areas involving subcortical and periventricular white matter, bilateral thalamus, bilateral globus pallidus with sparing of bilateral caudate and putamen nucleus and bilateral internal capsule Origin: © Department of Radio diagnosis, G. G. Hospital, M. P. Shah Medical College, Jamnagar, India.
Description: Diffusion-weighted image (DWI) showed restricted diffusion involving subcortical and periventricular white matter, bilateral thalamus, bilateral globus pallidus with sparing of bilateral caudate and putamen nucleus and bilateral internal capsule. Origin: © Department of Radio diagnosis, G. G. Hospital, M. P. Shah Medical College, Jamnagar, India.
Figure 4

Description: MR spectroscopy showed markedly elevated NAA and NAA-creatine ratio, which is pathognomic for canavan disease. Origin: © Department of Radio diagnosis, G. G. Hospital, M. P. Shah Medical College, Jamnagar, India.