Neuronal ceroid lipofuscinosis: Magnetic Resonance Imaging findings of brain
Published on 17.08.2016

DOI: 10.1594/EURORAD/CASE.13887
ISSN: 1563-4086
Section: Paediatric radiology
Area of Interest: Neuroradiology brain
Procedure: Education
Imaging Technique: MR
Special Focus: Congenital Case Type: Clinical Cases

Authors: Anjana Trivedi1, Krushnadas Radadiya1, Jagruti Kalola, Chetna Dodia, Anirudh Chawla, Mayur Pankhania

Patient: 8 years, male

Clinical History:
An 8-year-old boy presented with a history of convulsions, visual difficulties for the last year, and delayed milestones. He was a known case of neuronal ceroid lipofuscinosis and was referred for follow-up brain magnetic resonance imaging.

Imaging Findings:
Brain MRI demonstrated marked prominence of the cerebral sulci and cerebellar folia with diffuse knife-blade cortical atrophy. There was thinning of the corpus callosum and brain stem atrophy. Atrophy of both basal ganglia was noted, with “boxcar ventricles” on coronal sequences due to caudate nucleus atrophy. There was moderate to severe ex vacuo dilatation of the ventricular system with markedly prominent sylvian fissures, cerebral sulci, basal cisterns, cisterna magna and cerebellar folia. Bilateral periventricular hyperintensities were seen on T2 weighted and FLAIR sequences. Normal flow voids were noted on T2 weighted images. No restricted diffusion was evident.

Discussion:
Neuronal Ceroid Lipofuscinosis (NCL) is a group of inherited neurodegenerative disorders which are broadly subdivided into infantile, late infantile, juvenile and adult types. NCL is also classified into several subtypes based on the affected stage of sphingolipid metabolism. It is suspected that there are common pathways for many of the variants. Its prevalence is 1 in 12, 500 in some populations. The prevalence of NCL is highest in Scandinavian countries, especially Finland. It is classified into CLN1 to CLN8 on the basis of transfecting CLN1 (ceroid lipofuscinosis, neuronal 1) to CLN8 deficient cells [1, 2]

NCL has a heterogeneous clinical presentation depending upon the age of presentation. In the infantile form, its presentation is developmental delay and growth retardation. In late infantile and juvenile forms, it presents with cognitive dysfunction, visual failure, myoclonus and learning disability. The adult phenotype usually presents in the third decade with loss of balance, psychiatric illness, Parkinsonian features, progressive loss of vision, and progressive impairment of memory and intellectual function sufficient to interfere with social skills. There is impairment of orientation, the ability to learn, and higher executive functions such as planning, organizing and sequencing. The diagnosis is confirmed by measuring the specific defective enzyme in leukocytes, cultured fibroblasts, dried blood spots and saliva. [1, 2, 3]

Findings on MRI include diffuse cerebral and cerebellar atrophy of both grey and white matter. There is ex vacuo
dilatation of the ventricular system. T2 weighted images show hyper-intense periventricular white matter and low signal intensity in the thalami and striatum. There is thinning of corpus callosum. There is no restricted diffusion on diffusion weighted imaging. Proton MR spectroscopy reveals progressive changes, with a reduction of NAA and an increase of myoinositol and glutamate/glutamine. In long-standing late infantile NCL, myoinositol increases substantially with a marked reduction of lactate. [2, 3, 4, 5]

Treatment is symptomatic such as for convulsions. Neuronal Ceroid Lipofuscinosis is a progressive disorder with reduced life expectancy. There is no specific treatment or cure for any type of NCL, and a delay in diagnosis has no impact on its clinical course or on the prognosis.

**Differential Diagnosis List:** Late infantile neuronal ceroid lipofuscinosis, Epileptic encephalopathies, Infections, Inherited metabolic disorders

**Final Diagnosis:** Late infantile neuronal ceroid lipofuscinosis

**References:**


Description: Axial T2W FLAIR image shows marked prominence of cerebral sulci. Origin: Department of radiodiagnosis, P.D.U. Medical college Rajkot
Description: Sagittal T1 Inversion Recovery Image reveals diffuse knife blade atrophy of cerebral cortex with marked prominence of cerebral sulci and cerebellar folia. Origin: Department of radiodiagnosis, P.D.U. Medical college Rajkot
Figure 3

Description: Axial T2W image shows prominent cerebellar folia and knife blade atrophy in the temporal lobes. There is dilatation of the forth ventricle, and the preponine and ambient cisterns. Origin: Department of radiodiagnosis, P.D.U. Medical college Rajkot.
Description: Sagittal T2W image shows thinning of corpus callosum, and atrophy of the cerebellum and brain stem. There is dilatation of the third and fourth ventricles, the cisterna megha and pre-pontine cistern. Origin: Department of radiodiagnosis, P.D.U. Medical college Rajkot
Description: Axial T2W image shows bilateral periventricular white matter hyperintensity with moderate to severe dilatation of the ventricular system and diffuse knife blade atrophy of cerebral cortex. Origin: Department of radiodiagnosis, P.D.U. Medical college Rajkot.
Description: Coronal T2W image shows "boxcar ventricles" with ventriculomegaly associated with atrophy of both caudate nuclei. Diffuse knife blade cortical atrophy is noted with prominent sylvian fissures and cerebral sulci. Origin: Department of radiodiagnosis, P.D.U. Medical college Rajkot.