Case 11835

Krabbe disease
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Section: Neuroradiology
Area of Interest: Neuroradiology brain
Procedure: Diagnostic procedure
Imaging Technique: MR-Diffusion/Perfusion
Imaging Technique: MR
Imaging Technique: CT
Special Focus: Metabolic disorders Case Type: Clinical
Cases
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Patient: 5 months, female

Clinical History:

The patient had a delayed development, hypertonia and hyper-reflexia. She was not capable of holding her head or showing a social smile. She also suffered of low grade fever.

Imaging Findings:

On non-contrast MRI brain, abnormal altered signal intensity lesions was noted in bilateral peri-ventricular white matter, posterior limb of internal capsule, bilateral cerebral peduncles of midbrain, pons, ventral medulla, bilateral cerebellar white matter and bilateral cerebellar nuclei.
The lesions appeared hyperintense on T2 and FLAIR images and hypointense on T1 images and showed restricted diffusion in bilateral perirolandic areas. The lesions involved the corticospinal tract and sparing of sub cortical U fibres.

On non-contrast CT, bilateral symmetrical hyperdensity was noted in bilateral thalami. Faint calcification was noted in bilateral peri-ventricular white matter and bilateral corona-radiata.

In laboratory evaluation, there was very little activity (2SD below the reference value) of galactocerebroside β-galactosidase in blood leukocytes.

Discussion:

Krabbe disease is an autosomal recessive disorder. It is due to deficiency of galactocerebroside β-galactosidase enzyme. This enzyme degrades cerebroside in myelin. When myelination starts, myelin turnover is not properly done and cerebrosides accumulate in the lysosomes of macrophages within the white matter and cause demyelination. These macrophages show a globoid cell appearance. This disease is also called globoid cell leukodystrophy. This enzyme deficiency is due to a gene defect on chromosome 14q. [1]

There are four forms of Krabbe disease: Infantile, late infantile, juvenile and adult. The clinical manifestation varies according to age of presentation. The infantile form is the most common. In infantile form, patients present with hypertonia, irritability, delayed milestones, loss of developed milestones, fever, myoclonus, opisthotonus and nystagmus. Krabbe disease is fatal and rapidly progresses in infantile form. [1]

On CT brain imaging, bilateral thalami show hyperdensity. Sometimes both caudate nuclei, peri-ventricular white matter, posterior limb of internal capsule and brain stem also show hyperdensity with faint calcification. [2]

On MRI brain imaging, abnormal hyperintensity is noted in bilateral periventricular white matter, corona radiata, centrum semiovale, posterior limb of internal capsule, brain stem, cerebellar nuclei and white matter on T2 images.
with predominant involvement of cortico-spinal tract. [1]
Active demyelination shows restricted diffusion. On post-contrast study, there is enhancement in active
demyelination areas and spinal nerve roots. [3]
On MR spectroscopy, there is elevation of choline and myoinositol and reduction of NAA. [4]
Galactosylceramide beta-galactosidase activity measurement in peripheral blood leukocytes, cultured fibroblasts,
cultured amniocytes and chorionic villi can help confirm a diagnosis of Krabbe disease. [5]
Overall outcome of this disease is poor but now haematopoietic stem cell transplantation can be done and
substantially improve outcome of patients by delivering α-galactocerebrosidase. [6]
Take home message: Imaging should be carried out in early onset infantile seizures and in older children with
spasticity and ataxia to rule out this disease. [5] Early diagnosis of this disease improve outcome of patients by stem
cell transplantation. [6]
Teaching points: Corticospinal Tract involvement in MRI and hyperdense thalami in CT are the important findings
that help to diagnose this disease. [5]

**Differential Diagnosis List:** Krabbe disease, GM2 gangliosidosis, Amyotrophic lateral sclerosis

**Final Diagnosis:** Krabbe disease

**References:**

Radiographics 22 (3): 461-76 (PMID: 12006681)
(PMID: 6331756)
Given CA 2nd, Santos CC, Durden DD. (2001) Intracranial and Spinal MR Imaging Findings Associated with
Krabbe’s Disease: Case Report. AJNR 22: 1782-1785 (PMID: 11673180)
AJNR 21: 1478-1482 (PMID: 11003282)
SB Grover, P Gupta, M Jain, A Kumar, P Gulati (2005) Characteristic CT and MR features of Krabbe’s disease : A
case report. Ind J Radiology Imaging 15:4:503-506
Description: Abnormal hyperintensity in bilateral periventricular white matter with sparing of subcortical U fibres. Origin: Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
Description: There is loss of normal hypointensity within posterior limb of bilateral internal capsule, predominantly on the left. Both thalami appear hypointense. Origin: Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
Description: Both cerebral peduncles show hyperintensity instead of normal hypointensity at this age.
Origin: Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
Description: abnormal hyperintensity is noted in corticospinal tract in pons. Origin: Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
Description: Abnormal hyperintensity is noted in pons and bilateral middle cerebellar peduncle. Origin: Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
Description: Abnormal hyperintensity is noted in corticospinal tract in medulla and bilateral cerebellar nuclei & bilateral cerebellar white matter. Origin: Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
Description: Abnormal hypointensity is noted in bilateral periventricular white matter predominantly on the right side. Origin: Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
Description: Both cerebral peduncles appear mildly hypointense instead of normal hyperintensity on T1 image. Origin: Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
**Description:** Abnormal hypointensity is noted in corticospinal track in pons. **Origin:** Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
**Description:** Abnormal hypointensity is noted in ventral medulla and bilateral cerebellar nuclei. **Origin:** Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
Description: On axial image of CT Brain, hyperdensity in bilateral thalami. Origin: Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
**Description:** On axial image of CT Brain, faint calcification in bilateral periventricular white matter.

**Origin:** Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
**Description:** On axial image of CT Brain, faint calcification in bilateral periventricular white matter.

**Origin:** Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
**Description:** On coronal reformatted image of CT Brain, hyperdensity in bilateral thalami and calcification in bilateral periventricular white matter. **Origin:** Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.
Description: Restricted diffusion is noted in bilateral perirolandic areas. Origin: Sanya Diagnostics Center, Rajkot Civil Hospital, Rajkot, Gujarat, India.