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Pediatrics

INFANTILE KRABBE'S DISEASE – A CASE REPORT

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ABSTRACT Krabbe's disease is a rapidly progressing genetic disorder that affects the central and peripheral nervous systems. Early Infantile form of Krabbe's Disease is the most severe form, initial symptoms typically start between 3 to 6 months of age when the demyelination process begins. These children usually die before 2 years of age. We report a rare case of infantile krabbe's in an one year old child.

KEYWORDS : Krabbe's disease, Recurrent seizures, Global developmental delay, Demyelination disorder.

INTRODUCTION

Krabbe's disease or globoid cell leukodystrophy is a rare autosomal recessive disorder characterised by defective functioning of beta galactocerebrosidase, leading to severe mylein loss due to increased destruction with myelin formation being normal. Initial symptoms include loss of previously attained milestones and feeding difficulties. This disease is often misdiagnosed as colic, reflux, food/milk allergy, or even cerebral palsy.

CASE REPORT

One year old male child born out of third degree consanguinous marraige having complaints of recurrent seizures since day two of life, child subsequently had global developmental delay, excessive irritability and persisting seizures despite being on multiple antiepileptics. Child presented to our hospital with episodes of active seizures, h/o nasal regurgitation, drooling of saliva, failure to thrive, progressive spasticity of muscles all over the body.

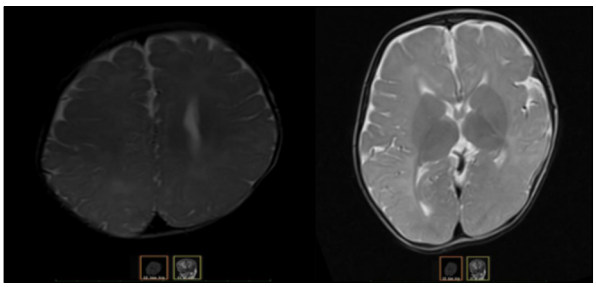
On Examination

Child was awake, supine with varying degree of flexion in all four limbs, drooling of saliva present, pallor present, no visual fixation, no neurocutaneous markers. VITALS : PR-124/min, RR-30/min, BP-80/50 mmHg

CNS : Increased tone in both upper and lower limbs DTR Exaggerated Plantar Extensor on both sidesFundus examination : Pale retinal background present, no optic atrophy.Hearing evaluation : Normal.Other systems : Normal.

Investigationsmri

showed lack of normal myelination signals in centrum semi ovale close to bilateral frontal and subcortical white matter at bilateral parietal region for age. Low signal in bilateral basal ganglia and ventro lateral thalami on T2 weighted image suggestive of hypomyelination disorder.



CSF analysis showed normal study. EEG showed generalized spikes with burst supression pattern

Genetic Study: Sent, reports awaited.

Treatment Given

Seizures treated with levetiracetam, phenobarbitone, clobazam. Physiotherapy given. Nutritional advice given and proper feeding methods taught to the parents.

DISCUSSION

Infantile Krabbe's is characterised by onset before 3 months, Neuropsychomotor deterioration along with seizures and irritability, Hypertonia, Microcephaly, Multiple spontaneous spasms charecterised by hyperextension of limbs and head. Treatment modalities include symptomatic and supporive treatment for seizures and hypertonia. Cord blood stem cell transplantation can prolong the life expectancy of the child as it slows down the neurological degeneration.

CONCLUSION

Prognosis of krabbe's disease is poor owing to progressive demyelination caused by the toxicity of psychosine. However with biochemical and molecular genetics evaluation, the diagnosis may be established early. This emphazise the importance of pediatrician in confirmation of diagnosis and subsequent genetic advice for family since there is risk of recurrence for future generations.

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