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RARE CASE OF GLOBOID CELL LEUKODYSTROPHY (KRABBE'S DISEASE)

Radio-diagnosis

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KEYWORDS

Krabbe's Disease, Globoid Cell Leukodystrophy, Autosomal Recessive Lysosomal storage disease, Demyelination, Enzyme beta galactosidase

INTRODUCTION:

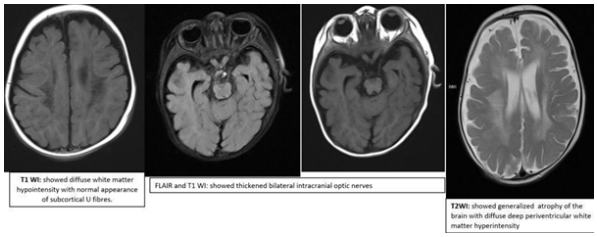
There are some classes of childhood intracranial disease processes who have typical pattern of findings in imaging with a great range of variations. Some of these are quite rare in occurrence and diagnosis is not often histologically confirmed or proper follow up could not be done. This is why their pattern is not quite familiar to new radiologist and often difficult to be identified at once. One such disease is Krabbe's Disease, also known as Globoid Cell Leukodystrophy which is an Autosomal Recessive Lysosomal Storage Disease with faulty Galactose cleavage due to deficiency of enzyme galactocerebroside Beta-galactosidase resulting in progressive accumulation of psychosine in large globoid epithelioid cells. There is progressive and is a rarity but have peculiar imaging features which aid to the diagnosis.

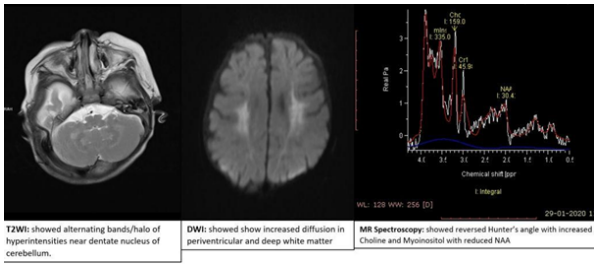
BACKGROUND:

A 7-month-old female child with non-significant birth history (full term gestation, delivery by CS with immediate cry and no NICU admission) came with recent unprovoked episodes tonic convulsions lasting a few minutes followed by loss of consciousness with drowsiness after gaining consciousness back, manifested two to three times per day with diurnal and nocturnal attacks.

A series of magnetic resonance imaging (MRI) scans of the patient's brain revealed diffuse white matter demyelination with sparing of subcortical U fibres. There is thickening of intracranial part of optic nerves. Areas surrounding the dentate nuclei show T1 hypointensity with T2 hyperintense alternating bands in cerebellum. There is mild generalised atrophy.

There was sparing of thalamus in this case which is usually involved in Krabbe's disease. There are confluent poorly defined hyperdensities are found in CT images involving Thalamus and corona radiata which appear hypodense in T1W MR images, typical for this disease.





Based on clinical and imaging findings, a provisional diagnosis of Krabbe's Disease was made which was subsequently confirmed by histopathological examination. Parents of the affected baby were counselled and were informed about the disease and possible outcomes with and without available treatments.

DISCUSSION:

This is a rarely encountered condition with most common age of presentation being 3-6 months. There is accumulation of Psychosine due to deficiency of galactose cleaving enzyme Galactocerebroside beta galactosidase which is caused by a gene mutation which shows autosomal recessive inheritance. Psychosine is toxic to oligodendrocytes and is accumulated to more than 100 times of its usual normal levels. Psychosine is not found in most normal brains. which ultimately leads to destruction of oligodendrocytes and demyelination.

More common in females and infantile form being the most common one. Infants are usually presented with complaints of extreme irritability and poor feeding but any other symptom, like seizure, according to the brain involvement can be expected.

Imaging findings includes volume loss with white matter thinning, dilated ventricles and enlarged sulci. Subcortical U fibres are usually spared.

This disease is progressive in natures. Disease being progressive, is usually fatal over time. Bone marrow transplant is an option which helps less symptomatic patients but does not completely improve the condition of the patient.

Differential diagnosis in children should include:
1. Metachromatic leukodystrophy
2. Vanishing white matter disease
3. Neuronal ceroid lipofuscinosis
4. GM2 Gangliosidosis

CONCLUSION:

Pattern recognition is the key to diagnose Krabbe's disease. Although Krabbe's disease is fatal and progressive, early diagnosis might help to improve the prognosis and reduce the severity of its manifestation in children.

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