MRI pattern recognition of neuroceroid lipofuschinosis in children

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Introduction

- Neuronal ceroid lipofuscinosis (NCL) is a group of neurodegenerative disorders mainly affecting the gray matter that are characterized by seizures, cognitive decline, myoclonus, visual impairment and abnormal movements.
- These disorders are both clinically and genetically very heterogeneous thereby complicating its diagnoses.
- There are 14 types of NCL described so far
- MRI brain pattern recognition may help in recognizing the type of NCL in few cases
- This is very important in the current era when next generation sequencing is commonly used as one of the initial modality to diagnose neurodegenerative disorders.
- When we get variants of unknown significance results on genetic tests, a correct phenotyping both clinically and radiologically helps in the correct interpretation.
- While most studies in general mention that MRI shows diffuse cerebral and cerebellar atrophy with thalamic hypointensities, specific findings in different subytypes of NCL are lacking
- There have been very few studies on MRI in children with especially in the subtypes

Objectives

- To study the pattern of signal abnormalities seen in different types of confirmed cases (Enzymatically and/or genetically)

Material and Methods

- It was a retrospective study in a single tertiary care centre in south-India over the last decade.
- MRI brain of children with confirmed diagnosis of NCL (enzymatically and/or genetically) was carefully analysed.
- Different areas of the brain were looked meticulously for signal changes and marked using a standardized proforma
- The abnormalities found were compared with sub-type of NCL and MRI findings in a given subtype were compared at different time points when more than one scan was available.

Results

- MRI brain of 56 children with confirmed diagnosis of NCL (enzymatically and/or genetically) were analysed
- Most cases showed cerebral and cerebellar atrophy with signal changes in the basal ganglia including thalami.
- Many of them showed subtle white-matter hyperintensities in the periventricular white matter also.
- The salient MRI findings in different subtypes of NCL is shown in table-1.
- The changes are characteristic and suggestive of the subtype at presentation.
- Towards the end-stage of the disease the findings are nonspecific.

Table-1 showing salient neuroimaging findings in different subtypes of NCL in children

Subtype of NCL (No. of subjects)	MRI findings
CLN-1 (n-16)	Prominent cerebral atrophy with hypointense thalami. Cerebellum normal initially
CLN-2 (n-25)	Severe early cerebellar atrophy with periventricular white matter hyperintensities. Thalamus normal initially but later shows hypointensities
CLN-6 (n-4)	Thalamic hypointensities with periventricular white matter hyperintensities and mild diffuse cerebellar atrophy
CLN-7 (n-6)	Thalamic hypointensities with periventricular white matter hyperintensities and early severe diffuse cerebellar atrophy
CLN-8 (n-3)	Thalamic hypointensities with hypointensities in globus pallidi and posterior putamina, periventricular white matter changes and early severe cerebellar atrophy
CLN-11 (n-1)	Cerebellar hypoplasia with pure cerebellar atrophy. Supratentorial compartment normal at presentation
CLN-14 (n-2)	Initially MRI showed delayed myelination. Severe cerebellar atrophy with normal supratentorial compartment after years of presentation.

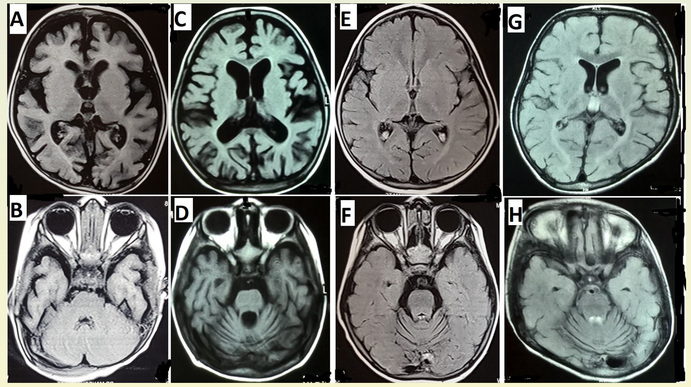

Fig 2 MRI brain of CLN1, CLN2, CLN6 and CLN7
1A, 1B: FLAIR axial section of the brain of CLN1 child at the level of basal ganglia and cerebellum respectively showing diffuse cerebral atrophy, thalamic hypointensities and sparing of cerebellum.
1C, 1D: FLAIR axial section of the brain of CLN2 child at the level of basal ganglia and cerebellum respectively showing diffuse cerebellar and cerebral atrophy (cerebellum more than cerebrum); normal thalamus and periventricular white matter hyperintensities.
1E, 1F: FLAIR axial section of the brain of CLN6 child at the level of basal ganglia and cerebellum respectively showing mild diffuse cerebellar atrophy, thalamic hypointensities and periventricular white matter hyperintensities.
1G, 1H: FLAIR axial section of the brain of CLN7 child at the level of basal ganglia and cerebellum respectively showing prominent diffuse cerebellar more than cerebral atrophy, thalamic hypointensities and periventricular white matter hyperintensities.

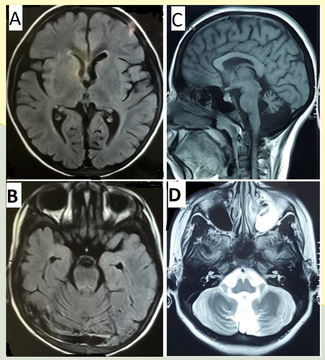

Fig:2 MRI of NCL8 and NCL 11
2A, 2B: FLAIR axial section of the brain of CLN8 child at the level of basal ganglia and cerebellum respectively showing mild diffuse cerebellar and cerebral atrophy, thalamic hypointensities and periventricular white matter hyperintensities.
2C, 2D: T1W sagittal section of the brain and T2W axial section of the brain at the level of cerebellum respectively showing isolated cerebellar atrophy (mainly vermian) with hypoplasia.

Conclusions

MRI at presentation in children with NCL can suggests the subtype of NCL in many cases .