KREP

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KARNATAKA RADIOLOGY EDUCATION PROGRAM

CASE PRESENTATION

Case of neuronal ceroid Lipofuscinoses VS spinocerebellar ataxia

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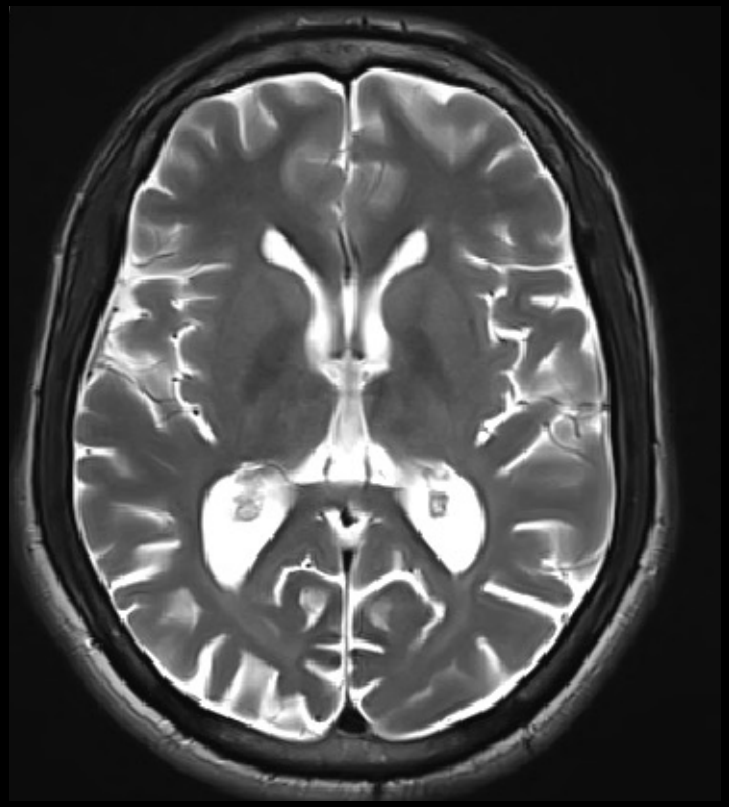
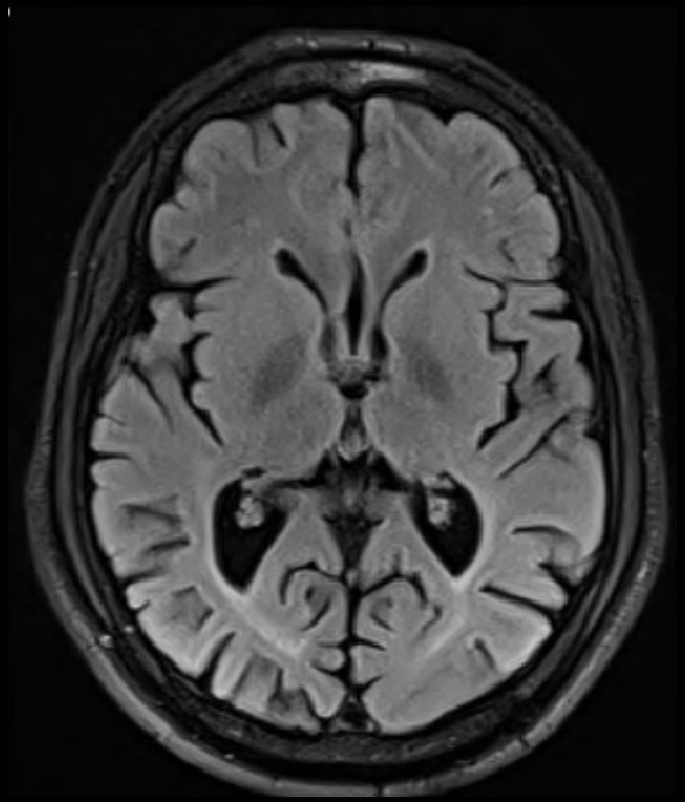
Case -1

- 2O year old male patient with complaints of inability to walk for 8 years.
- Birth history : normal
- On clinical examination – ataxia was present
- No history of visual disturbance

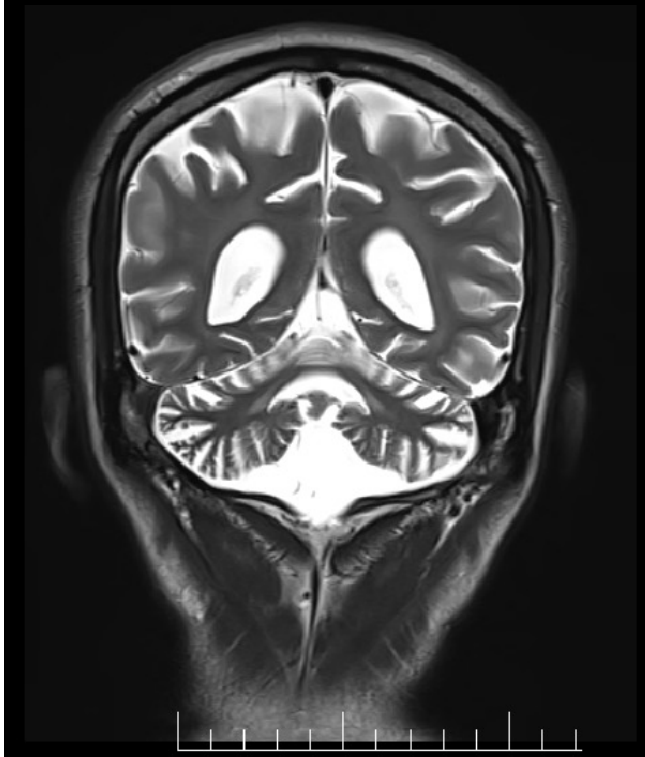
T2 axial



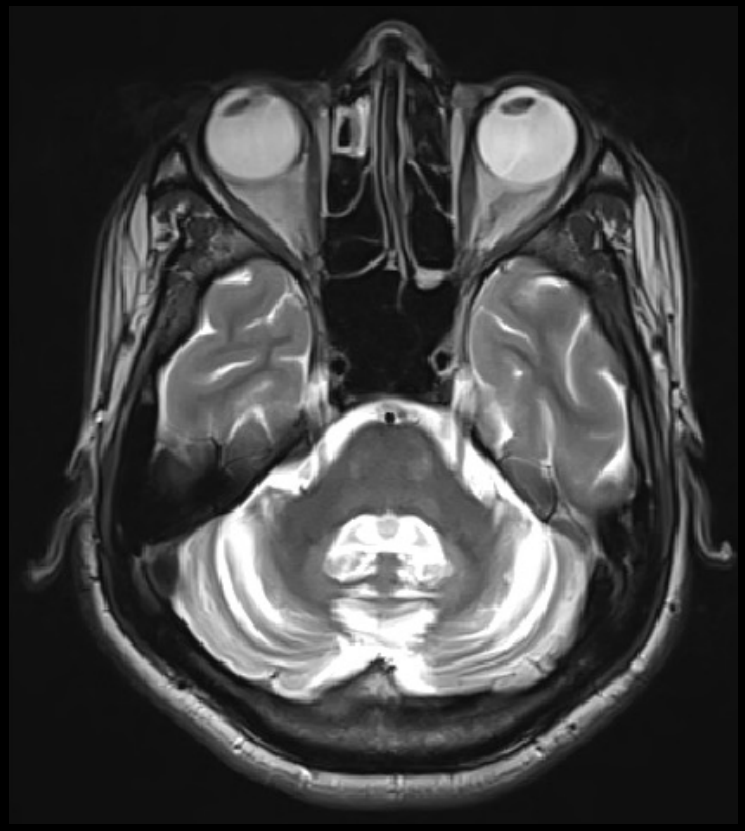
FLAIR axial



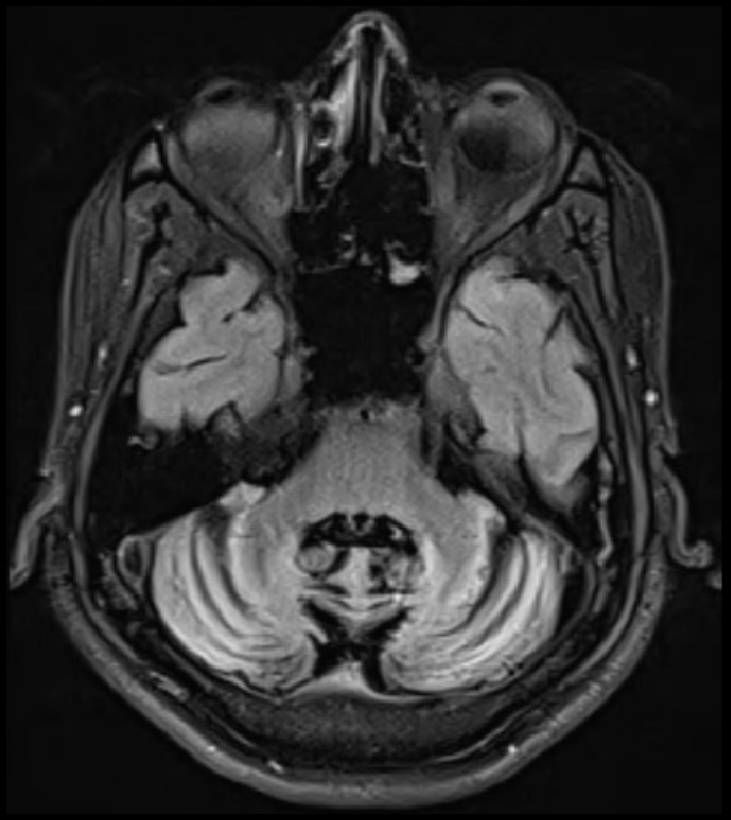
T2 coronal



T2 axial



FLAIR axial



Findings:

- Symmetrical T2 & FLAIR hyperintensities noted in periventricular and peritrigonal regions
- T2 & FLAIR hypointensities in bilateral thalami
- Prominent cerebellar foliae (cerebellar atrophy)
- Atrophy of middle cerebellar peduncle
- T2 & FLAIR hyperintensity involving dentate nuclei

DIFFERENTIALS

- Neuronal ceroid lipofuscinosis
- Spinocerebellar ataxia

Neuronal ceroid lipofuscinosis

POINTS IN FAVOUR	POINTS AGAINST THE DIAGNOSIS
Symmetrical periventricular hyperintensities	Absence of visual symptoms
T2/FLAIR hypointensity of thalami	
Cerebellar atrophy	
Hyperintensity of dentate nuclei	

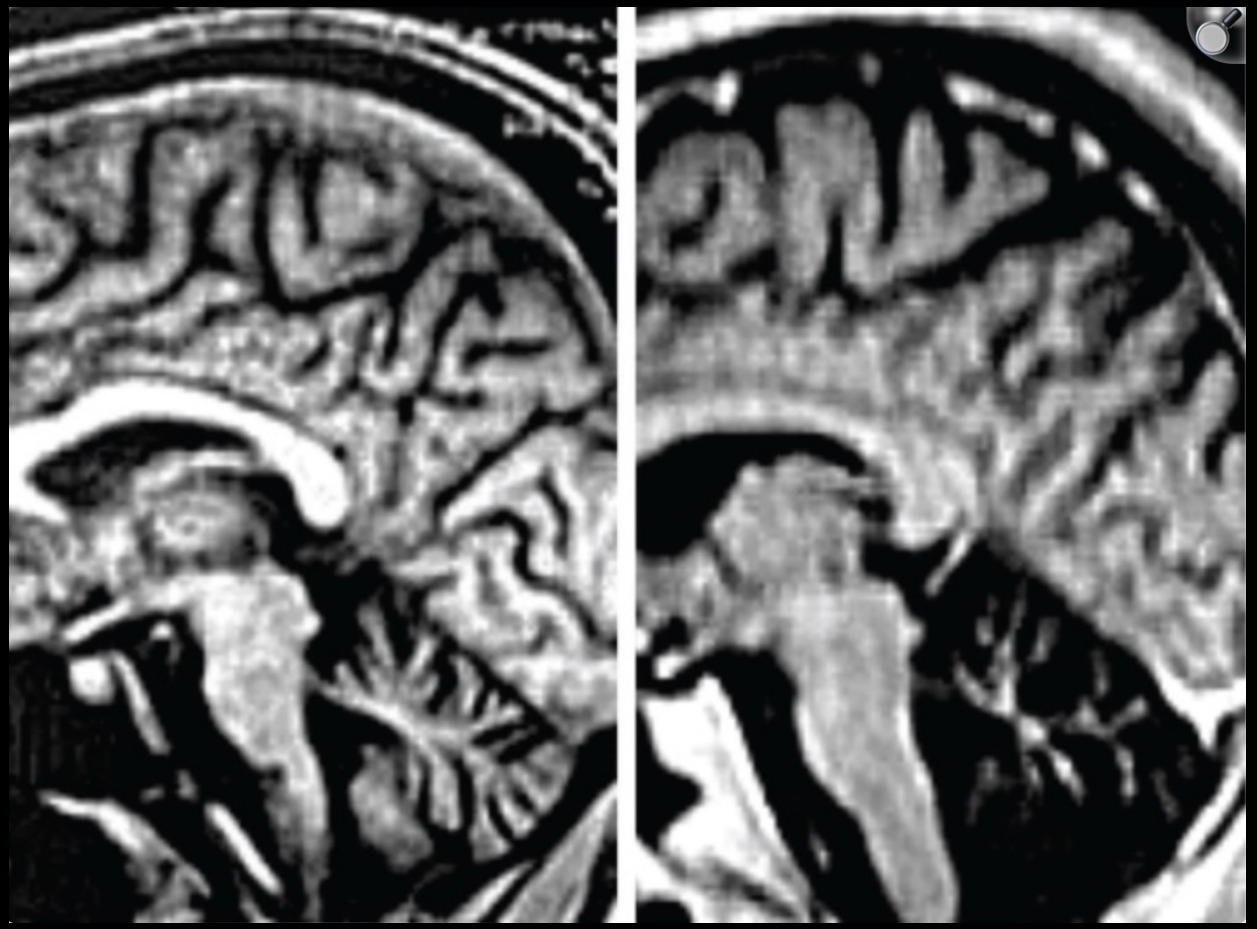
NCL Type	Typical Onset	MRI Findings	Differential Features
CLN1 (Infantile NCL)	6–24 months	- Diffuse **cerebral atrophy**(early) - **Thinning of corpus callosum** - **Delayed/diminished myelination** - Cerebellar atrophy (later)	Early, rapid cerebral atrophy in infancy is a key differentiator.
CLN2 (Late Infantile NCL)	2–4 years	- **Periventricular white matter hyperintensities** - Progressive **cerebral and cerebellar atrophy** - **Thalami hypointensity** on T2 - May show **subcortical U-fiber sparing**	Thalamic T2 hypointensity is more specific; cerebellar atrophy appears early.
CLN3 (Juvenile NCL / Batten Disease)	4–7 years	- Progressive **cerebral and cerebellar atrophy** - **Occipital lobe atrophy** is common - **Thalami and basal ganglia volume loss**	Occipital atrophy and visual loss precede generalized atrophy.

NCL Type	Typical Onset	MRI Findings	Differential Features
CLN5 (Variant Late Infantile NCL)	4–10 years	- **Cerebral and cerebellar atrophy** - Thalamic and brainstem involvement (variable)	Imaging overlaps with CLN2 but onset is later and less rapid.
CLN6 (Variant Late Infantile / Juvenile NCL)	Variable (childhood)	- Similar to CLN2/CLN5 - Cerebral and cerebellar atrophy - Periventricular white matter changes	Diagnosis is often clinical-genetic; imaging overlaps with CLN2 and CLN5.
CLN7 (MFSD8-related)	Childhood	- Diffuse **cortical and cerebellar atrophy** - **Corpus callosum thinning** - **Hypomyelination**	Resembles CLN2/CLN6; gene testing needed to differentiate.

NCL Type	Typical Onset	MRI Findings	Differential Features
CLN8 (Northern epilepsy variant)	Late infantile / Juvenile	- Mild or slow-progressing **brain atrophy** - May show **cortical thinning**, especially in parietal and occipital regions	Slower progression than CLN2; neurocognitive symptoms may precede imaging changes.
Adult NCL (e.g., CLN4)	Late adolescence to adulthood	- Mild **cerebral atrophy** - **Cortical signal abnormalities** (variable) - Basal ganglia changes may be present	Less specific findings; diagnosis often relies on clinical and genetic data.

Spinocerebellar ataxia

POINTS IN FAVOUR	POINTS AGAINST THE DIAGNOSIS
Cerebellar atrophy	Lack of extensive white matter changes
Middle cerebellar peduncle atrophy	Thalamic hypointensity
	Normal brainstem
	No history of visual disturbance



CASE OF SCA: Brain stem and cerebellum atrophy are clearly visible from the onset of clinical manifestations. With the evolution of the disease, there is an increase mainly in cortical and cerebellar atrophy.

Follow up

- Patient was given multivitamin tablets and managed out patient basis