A Rare Diagnosis Behind a Common Symptom: CLN8-Related NCL in a 7-Year-Old Boy

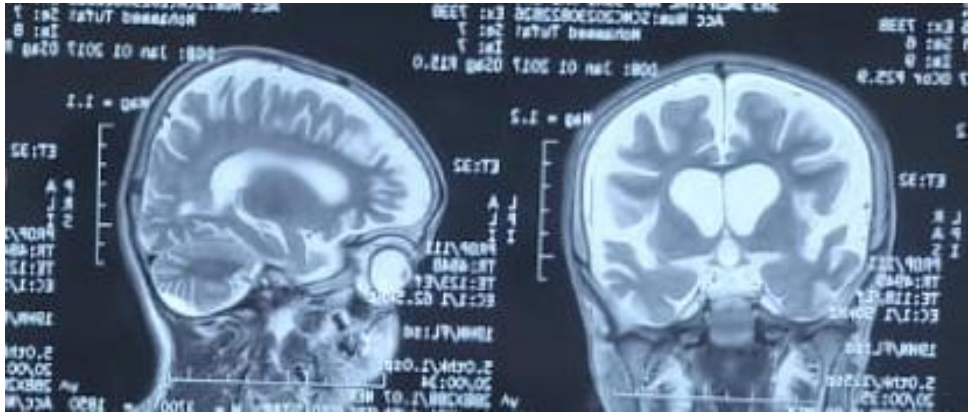
Case Report | IANCON 2025 E-Poster
id 517

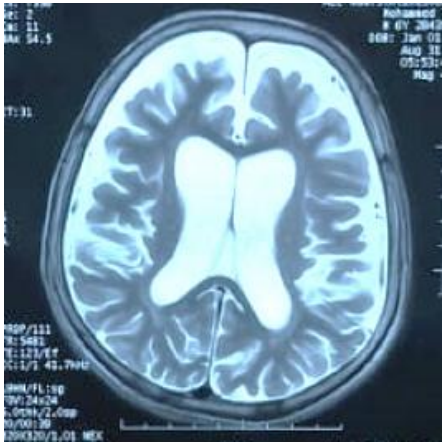
Introduction

- Neuronal Ceroid Lipofuscinosis (NCL) is a rare lysosomal storage disorder.
- Autosomal recessive; caused by mutations in CLN1–CLN14 genes.
- Characterized by accumulation of lipopigments in neurons caused neurodegeneration.
- Clinical: developmental regression, seizures, cognitive decline, vision loss.
- Classified: congenital, infantile, late-infantile, juvenile forms.
- MRI: cortical & cerebellar atrophy.
- Definitive diagnosis: genetic testing.

Case Presentation

- 7-year-old boy born out of consanguineous parents.
- Normal milestones until 4 years, then progressive regression.
- Symptoms -Begin at age of 4 years tremors, ataxia, frequent falls become bedridden in 2 years followed by progressive Vision loss, mutism, inability to follow commands for 6months . Seizure in form of GTCS (1 year), continuous myoclonic jerks (8 days).
- Examination:– GCS - E4V1M5 , Spasticity of all four limbs , brisk reflexes, neck weakness, extensor plantar
- MRI Brain: cerebral and cerebellar atrophy.
- Genetic testing: CLN8 mutation → Variant Late-Infantile NCL.





Discussion & Conclusion

Discussion:

• NCL classified by genotype (CLN1–CLN14) and phenotype. Triad of seizure , dementia and vision loss is found in most of cases

CLN8 is a rare form of Late-Infantile form presenting between age 2 and 5

MRI: cerebellar atrophy hallmark for NCL 8 type

• Early diagnosis vital for genetic counselling and supportive care.

Conclusion:

• NCL should be suspected in children with milestone regression, seizures, vision loss.

• MRI and genetic testing confirm diagnosis.

•Early recognition is crucial for genetic counseling, supportive management, and improving quality of life, as no definitive cure currently exists.