LAFORA DISEASE:A CASE SERIES

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INTRODUCTION

- Lafora disease is a rare, autosomal recessive progressive myoclonic epilepsy.
- Characterized by intractable seizures, cognitive decline, and the accumulation of Lafora bodies.
- Caused by mutations in either the EPM2A (laforin) or NHLRC1 (malin) genes.
- Hallmark pathological feature is the intracellular accumulation of polyglucosan bodies (Lafora bodies).

METHODS

- **Study Design: Retrospective observational case series.**
- **Setting: Institute of Human Behaviour and Allied Sciences (IHBAS), Delhi, between 2023 and 2025.**
- **Inclusion Criteria: Patients with clinical features of progressive myoclonus epilepsy (PME) and genetic or histopathological confirmation of Lafora disease.**

CASES

S. No.	Age/Gender	Onset Age	Symptoms	EEG Findings	MRI Brain	Gene mutation	Biopsy Findings
1.	16 Years/ Male	14 Years	Atonic seizures, Ataxia	Generalized background slowing with spike discharges	Normal	Not done	PAS-positive granules (Lafora bodies)
2.	15 Years/ Male	8 Years	Atonic seizures, cognitive decline	Posterior slowing	Normal	NHLRC1	Not done
3.	24 Years/ Male	22 Years	GTCS, Cognitive decline	Occipital spikes	Normal	EPM2A	Not done

CONCLUSION

- Lafora disease phenotypes and genotypes in India are similar to the phenotypes and genotypes as found in other parts of the world.
- Early seizures, rapid cognitive decline, typical EEG suggest diagnosis.
- Genetic testing or skin biopsy recommended.
- Early detection aids genetic counseling and supportive care.
- No curative treatment available.