Unusual Presentation of Bibrachial Amyotrophic Lateral Sclerosis with a Remarkably Stable Clinical Course: A Case of TARDBP-Associated ALS10 in a Young Female

Dr Pragnya Panda (M.D,D.M)
Assistant Professor
Department of Neurology
AIIMS,Raebareli

Aim

- Amyotrophic lateral sclerosis (ALS) is a relentlessly progressive neurodegenerative disorder, yet certain genotypic variants may present with atypical trajectories. Bibrachial-onset ALS is rare and often underrecognized, particularly in younger patients. Mutations in the TARDBP gene, implicated in ALS10, typically confer variable phenotypic expression.

Methodology

- We describe the clinical course, neurophysiological findings, imaging, genetic testing, and treatment response of a 31-year-old female diagnosed with TARDBP-associated ALS.

Case report

- 31-year-old woman with progressive symmetrical upper limb weakness and wasting.
- Stable for 3 years.
- No bulbar or respiratory compromise,lower limb, cognitive and behavioral involvement.
- Exam: distal amyotrophy, fasciculations, UMN signs.
- EMG: active and chronic denervation.
- Genetic test: pathogenic TARDBP variant confirmed ALS10.
- The patient has remained clinically stable over 18 months after edaravone therapy.

Variant Summary

Gene / Variant	Genotype	Assessment	Mode of Inheritance	Phenotype
TARDBP c.1043C>T p.(Ala348Val) g.11062509C>T	Heterozygous	Likely Pathogenic	dominant	Amyotrophic lateral sclerosis 10

Discussion

- Unusual bibrachial phenotype distinguished by:
- Early onset
- Selective upper limb involvement
- Indolent progression
- Stable trajectory contrasts with aggressive ALS.
- Highlights heterogeneity from genetic determinants.
- Expands clinical spectrum of TARDBP-associated ALS.

Conclusion

- Bibrachial-onset ALS with prolonged stability is rare.
- Recognition prevents diagnostic delay.
- Genotype–phenotype correlation aids prognosis.
- TARDBP-ALS10 may guide research in disease-modifying strategies.