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Genotypic and Phenotypic Spectrum of Children with Genetic West Syndrome From India – A Multicentre study

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INTRODUCTION

Etiology of IESS (West syndrome) is diverse

Genetic | **Metabolic** | **Structural**

- Approximately in 30 % of West syndrome (WS), no etiology is identified
- Genetic cause accounts approximately for 40 % of unexplained IESS
- Paucity of literature globally on genetic IESS and even scarce from the Indian subcontinent
- Michaud JL, et al from Canada:

Need for large multicentric international cohorts to find possible crucial information which could have potential therapeutic and prognostic implication

OBJECTIVES

To describe the genotypic and phenotypic profile of children with West syndrome (WS) of genetic etiology.

MATERIALS AND METHODS

Study Design: Ambispective-observational, multicentric study

Place of Study: Pediatric Neurology Unit, PGIMER, Chandigarh in collaboration with five pediatric centres across India over 15 months

Study Period: January 2021 to March 2022.

MATERIALS AND METHODS

Inclusion Criteria

Children (under follow-up or newly diagnosed) with genetic IESS (confirmed after January 2018 by next generation sequencing or sanger sequencing or karyotype or chromosomal microarray and others)

Exclusion Criteria

Children with known structural genetic (like neurofibromatosis type 1, Tuberous Sclerosis Complex, structural malformations) and structural neurometabolic etiologies were excluded

Genetically confirmed WS - confirmed by CES, WES, Epilepsy panel, CMA, etc., (Newly diagnosed as well as in follow-up) (Only Pathogenic and likely pathogenic variants as per ACMG)

Detailed In-person assessment in the centre of follow up

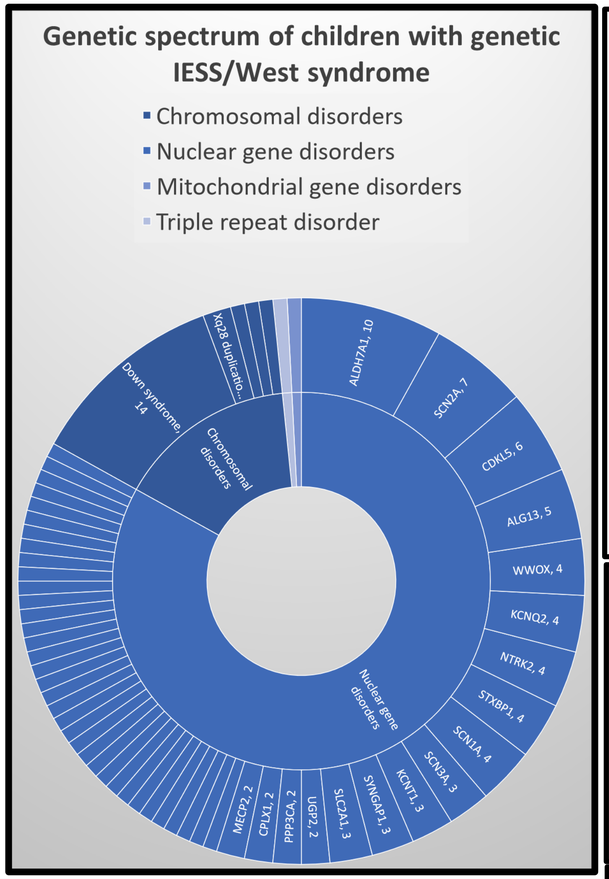
Telephonic assessment or/ and Retrospective review of case records

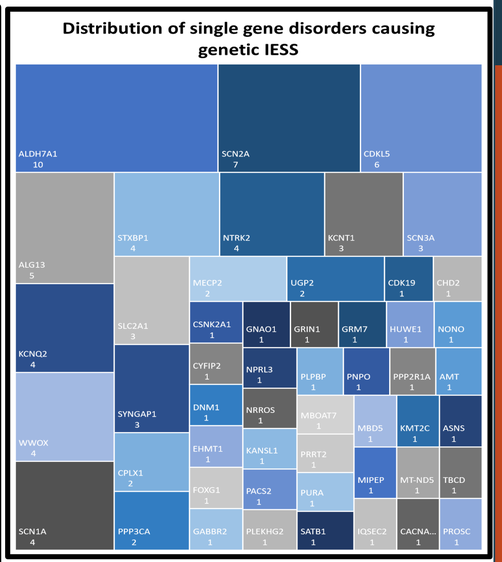
Collection and entry of data in pre-designed proforma/ Microsoft excel; Collation of data from all centres and statistical analysis

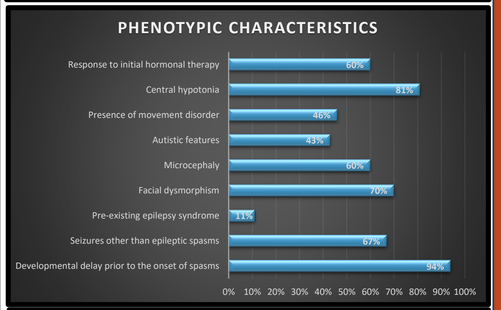
DISCUSSION

Largest cohort of exclusive genetic only West syndrome

RESULTS (Total 124 genetically confirmed cases)







Median age of onset of epileptic spasms: 5 months (22 children had late onset epileptic spasms)

CONCLUSION

- **Etiology of Genetic IESS/ West Syndrome is diverse.**
- **Down syndrome, ALDH7A1, SCN2A, CDKL5, ALG13 are the common causes of genetic WS**
- **Central hypotonia, developmental delay prior to the onset of spasms, autistic features, and facial dysmorphism were notable findings observed.**