

RP 115

Clinical Utility of Next-Generation Genomic Sequencing in Paediatric Clinical Genetics

A Cohort of 100 Cases

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Deploying Next-Generation Sequencing to Resolve Rare Paediatric Disorders

Transforming Diagnostics

Next-generation sequencing (NGS) has transformed the diagnosis of rare paediatric disorders with heterogeneous and overlapping phenotypes.

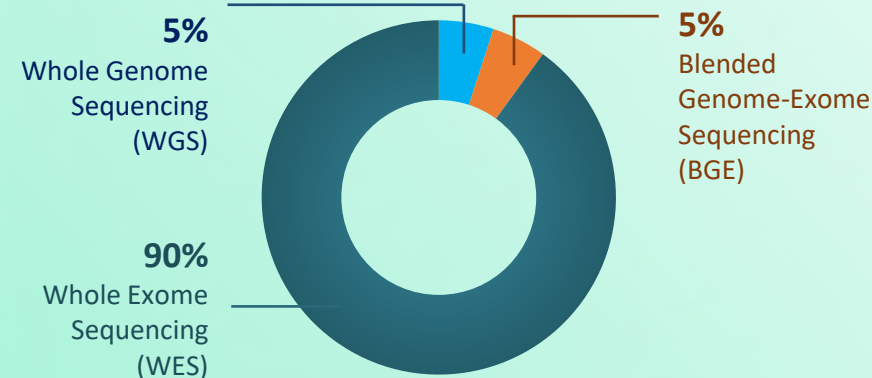
Study cohort (N=100)

Age Range: 1 day to 18 years

Demographic Split



Testing Modalities



Diagnostic Yield and Variant Spectrum

Molecular Efficacy

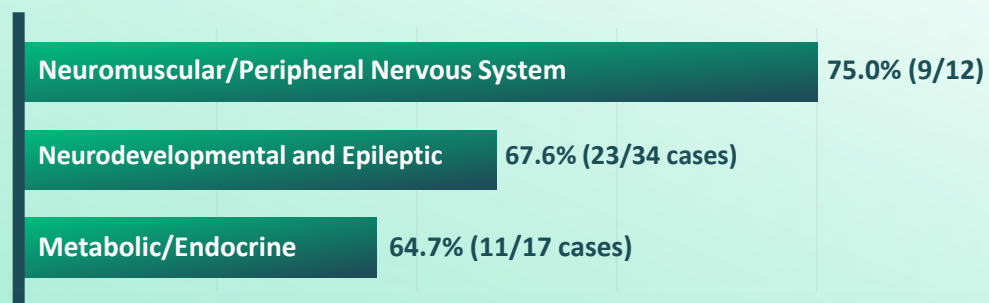
63%

Phenotype-related Molecular
Diagnosis Reached

84%

Yield of Actionable P/LP
Variants

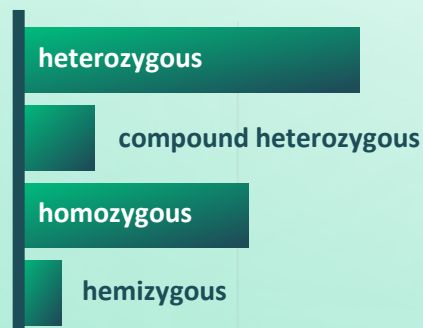
Highest Diagnostic Yields by Phenotype



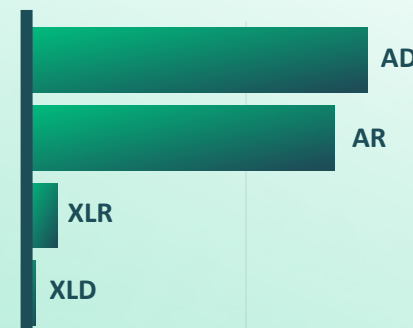
Variant Spectrum



Zygosity



Inheritance



Translating Genomic Findings into Definitive Clinical Management

Ending the Diagnostic Odyssey

- Establishes a definitive molecular diagnosis
- Reduces unnecessary investigations
- Provides diagnostic certainty for patients and families

Strategic Use of Genome Sequencing

- WES as the first-line test for most suspected monogenic disorders
- WGS for unresolved cases or when complex variants/CNVs are suspected

Precision Patient & Family Care

- Guides personalized treatment and clinical management
- Enables genetic counselling and recurrence risk assessment
- Supports family testing, reproductive planning, and targeted therapies