Poster P300
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SEQUENCE™ MD

Clinical Utility and Diagnostic Yield of Comprehensive Genetic Testing in Adults with Intellectual and Developmental Disabilities (IDD): *A Single-Center Experience*

Our Research

A retrospective analysis on **69 adult IDD patients** (ages 18-83, median = 30) with genetic testing results.

- **Inclusion:** Age ≥ 18 years, referral included ICD-10 codes F70-F84, R41, R48 and R62.
- **Exclusion:** Without a neurodevelopmental (NDD) assessment, with a previously obtained IDD diagnosis, who did not undergo genetic testing for IDD/NDD indications, and/or if genetic test results are pending.

What We Found

Comprehensive genetic testing yielded diagnostic rate on par with pediatrics* in adult IDD patients and

- Broad spectrum of genetic diagnoses
- Tangible benefits of diagnosis

Findings advocate extending ACMG pediatric standards to adults with IDD to:

- Bridge an existing healthcare gap
- Ensure equitable access to genetic evaluations
- Optimize care across the lifespan
- Reduce diagnostic odyssey and associated costs

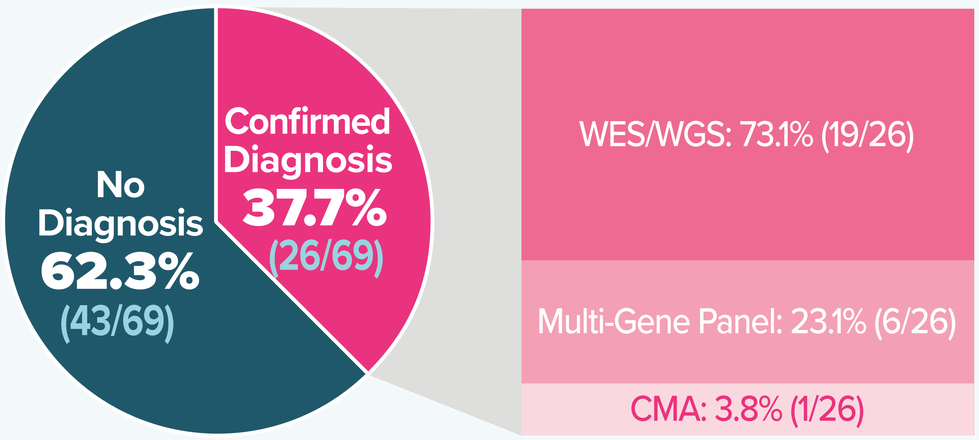
WES/WGS showed:

- Highest diagnostic efficiency
- Lower follow-up requirements
- Broader clinical impact

DUAL DIAGNOSES in 3 Patients

Actionable Medication Implications 15%

Diagnostic Yield



Disease Categories

- Neurological Disorder: 34.5% (10/29)
- Deletion/Duplication Syndrome: 27.6% (8/29)
- Monogenic Syndrome: 24.1% (7/29)
- Cerebrovascular Malformation: 3.4% (1/29)
- Endocrinopathy: 3.4% (1/29)
- Inborn Error of Metabolism: 3.4% (1/29)
- Mitochondrial Disorder: 3.4% (1/29)

Clinical Utility of Genetic Testing

IMMEDIATE MANAGEMENT 27.5%
Direct therapeutic intervention
Medication modification
Acute surveillance initiation

PREVENTIVE CARE 34.8%
Disease-specific monitoring protocols
Risk-reduction strategies
Specialist referral networks

LONG-TERM PLANNING 37.7%
Family cascade testing
Reproductive counseling
Resource coordination

DISCUSSION: Natural history discovery, justification of SSI for patients (disability income), uncovering carrier, pharmaco and secondary findings through opportunistic screening

Conclusion

Our results demonstrate that the diagnostic yield of comprehensive genetic testing is consistent with that of the pediatric population with IDD. There are additional benefits in the form of opportunistic screening in adults with IDD, including preventive cancer screening, medication modification, long-term planning, establishing social services, and support.

Future Direction

More research is needed to determine a standardized approach for measuring clinical and life outcomes in the adult population with IDD.

FOR MORE RESEARCH FINDINGS AND DATA FROM SEQUENCEMD, THE CLINICAL GENETICS PRACTICE, SCAN THE CODE.

* PMID: 34211152, 32203227 and 30002876