

Whole-Exome Sequencing Enhances Patient Care

RESULTS FROM WHOLE-EXOME SEQUENCING (WES) CAN PROVIDE INSIGHTS TO INFORM MEDICAL MANAGEMENT DECISIONS



A recent collaboration between Ambry Genetics and The Kennedy Krieger Institute published in *Molecular Genetics and Genomic Medicine* reveals health care providers' practices and medical management changes based on WES results.

WHY THIS MATTERS TO YOU

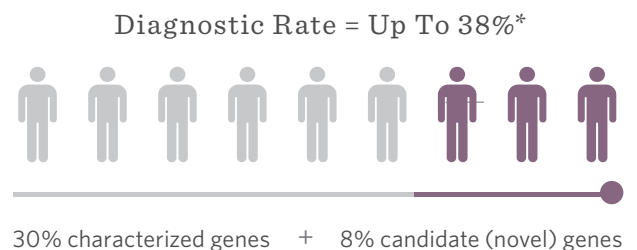
WES is one of the most accurate and efficient diagnostic tools for patients with undiagnosed genetic disorders. Data from Ambry's new published study suggest that results from WES, regardless of whether the result is positive, negative, or uncertain, can inform new treatment decisions to improve the care of your patients.¹

BACKGROUND

- A growing body of literature supports the use of WES as a first-tier test as it can substantially reduce cost and time to diagnosis.²⁻¹⁰
- WES provides a diagnostic rate ranging from 25-40%, which is two to three times higher than traditional genetic testing methods.¹¹⁻¹⁸
- An early and accurate molecular diagnosis can lead to optimal care and dramatic prognostic improvements for patients and their families.^{12,15,18-22}
- We evaluated the clinical utility of WES by surveying healthcare providers (HCPs) about changes in clinical management subsequent to receiving their patient's WES test results. The survey gathered information from the ordering HCP about changes in medication and treatment plans, prognosis and risk assessment, reproductive planning, and educational services subsequent to WES testing (findings below).

POINTS FOR YOUR PRACTICE

- Ambry's ExomeNext has a 38% diagnostic yield.¹¹
- ExomeNext-*Trio* evaluates characterized and uncharacterized genes in the exome, possibly leading to gene discovery and new diagnoses that could not otherwise have been achieved with traditional diagnostic testing.
- Ambry's 'Patient for Life' initiative involves continuously analyzing negative exome data in hopes of eventually find an answer for each patient. The ordering HCP will always be alerted when a test result changes.

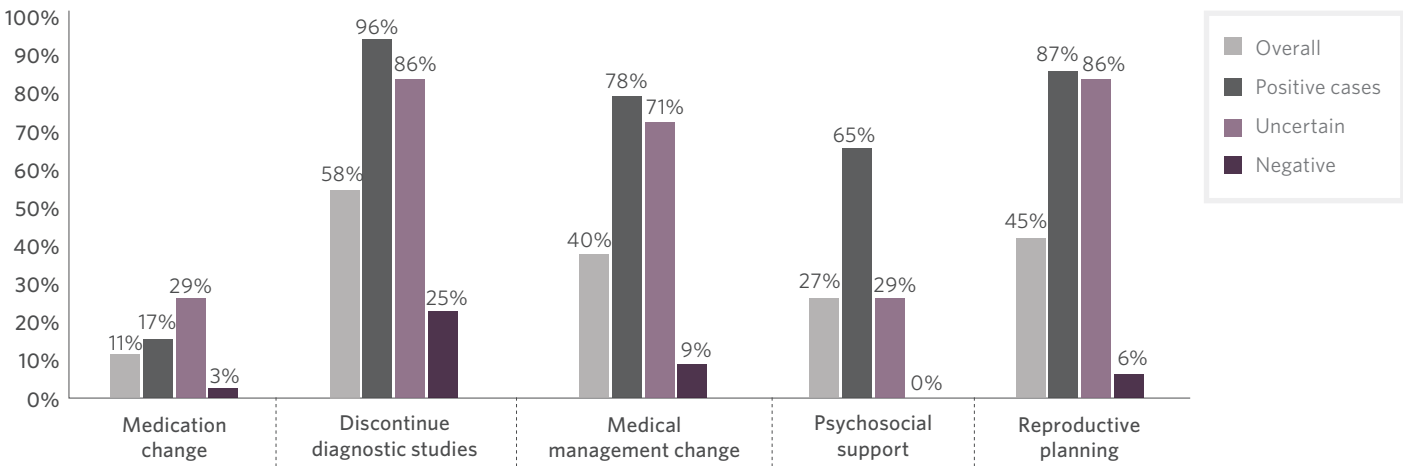


*Diagnostic rates vary based on test ordered. Trio test options provide ~2x higher detection rates than proband only.

SIGNIFICANT FINDINGS

- 95.7% of patients with positive WES results and 83.3% of patients with uncertain WES results were able to discontinue further diagnostic studies, including invasive procedures.¹
- Uncertain and negative WES results also impacted medical management (e.g. medication changes, referrals to specialists, reproductive planning-Figure 1), in some cases as much as a positive result.

Figure 1. Percentage of cases where each category of medical management or medical impact was affected.



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