

WHY CHOOSE ExomeNext

Whole exome sequencing (WES) looks at the DNA where 85% of known genetic variations reside and may indicate a disease-causing mutation. ExomeNext provides a clear picture of a patient's health in a timely and cost-effective manner that can lead to better patient outcomes quickly and cost-effectively.

BENEFIT	WHY THIS MATTERS
<p>High Diagnostic Rate</p> <p>WES is 2-3 times more likely to identify the underlying genetic defect than traditional tests</p>	<ul style="list-style-type: none"> A diagnosis brings an end to the expensive, time-consuming, and potentially invasive diagnostic odyssey that burdens patients, families, and the healthcare system. The diagnostic rate for WES ranges from 25-35% based on published literature.¹⁻⁸ The rate is 2-3 times as likely as traditional methods (single gene, panels, etc.) to identify the underlying cause of a patient's genetic disease.⁹⁻¹³ Exome sequencing reanalysis following an originally negative report is more cost-effective and results in a much better diagnostic rate than further genetic testing.¹⁴ The rate is continually increasing due to the rapid rate of new gene-disease discoveries.^{2,15} Today, a peer-reviewed study reporting a newly-discovered disease gene is published every other day on average.¹⁵
<p>Impacts Medical Management</p> <p>Positive results alter medical management for virtually all patients</p>	<ul style="list-style-type: none"> An early and accurate diagnosis can lead to optimal care and dramatic prognostic improvements for patients and their families.^{6,7,16-20} One clinic recently reported that medical management was impacted for 100% of their patients with positive WES results.⁶ Clinical utility benefits include: <ul style="list-style-type: none"> Tailored medical management^{6,18,20,21,24} Prophylactic therapy²³ Minimization of invasive diagnostic procedures (i.e. muscle biopsies, MRIs w/ anesthesia)^{7,20} Reproductive planning^{6,18,20,21,24} Eligibility for clinical trials^{7,8}
<p>Cost Effective</p> <p>WES is about half to one-quarter the cost compared to traditional genetic tests</p>	<ul style="list-style-type: none"> WES is 25-50% the cost of traditional genetic testing trajectory.^{13,21,24} Several studies show WES as a first line approach vs. standard clinical practice saves \$1,000-\$7,640 per case.^{11,13,22} Identification of the underlying cause of genetic disease leads to informed family planning; this reduces costs as family members can then undergo less expensive targeted single site analysis.²² Targeted treatment options improve patients' conditions and also reduce costly hospital fees, along with wasteful non-effective treatment modalities.^{7,20}

ABOUT THE TEST

Ambry's ExomeNext™ is performed using whole exome sequencing, a robust technology that sequences all of the functionally relevant regions of essentially all 20,000 genes of the human genome. The beauty of this test is that it has the capacity to pinpoint rare mutations in an unbiased and efficient way. WES in a diagnostic setting is the most cost-effective and comprehensive method to rapidly detect the underlying etiology in patients with genetic disease.^{10-13,22,25}

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