Ambry Genetics | LifeStrands

ExomeNext®

Reference Guide

Providing a Clearer Path to Patient Health – *For Life*

ExomeNext leverages Ambry's expert analysis and interpretation to provide answers for otherwise undiagnosed patients, revealing a clearer path to better and more informed health.

With over 10 years of experience in exome, Ambry has pioneered gene-disease validity curation and continues its search for answers through proactive gene upgrade reporting, supporting the health of its patients indefinitely.

Industry-Leading Methodology for Clarity

Ambry's exome testing is based on published gene-disease validation criteria and family inheritance-based variant filtering, providing an industry-leading approach to variant assessment and helping to ensure relevant disease-causing variants are captured and reported.

Expert Support to Interpret Results

Ambry's knowledgeable Genomic Science Liaisons and field support are available to answer questions and assist providers with results interpretation to more effectively manage patient care.

Patient for Life Program

Our unique, premium service reviews patient data for pathogenic or likely pathogenic variants in newly-characterized genes and proactively issues reclassification reports to ordering clinicians. This is carried out indefinitely and free of charge.

A Clearer Path to Patient Health

ExomeNext adheres to Ambry's published methods for characterizing gene-disease relationships[1-3], allowing for the most clinically-relevant findings to be reported in each patient and supporting better-informed medical management decisions.

LEADERSHIP IN EXOME

First commercial lab to offer whole exome sequencing in 2011

First published criteria for assessing clinical validity of gene-disease relationships[1]

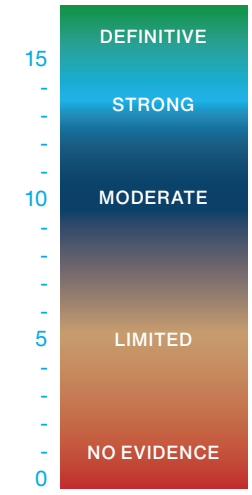
CLARITY

Ambry's gene-disease validity curation and expert analysis produces clear and accurate results.

The gene-disease validity assessment system classifies the strength of gene-disease relationships. This system is based on a weighted scoring system guided by the ClinGen gene curation criteria, with points assigned to evidence criteria supported by existing literature and data.[1]

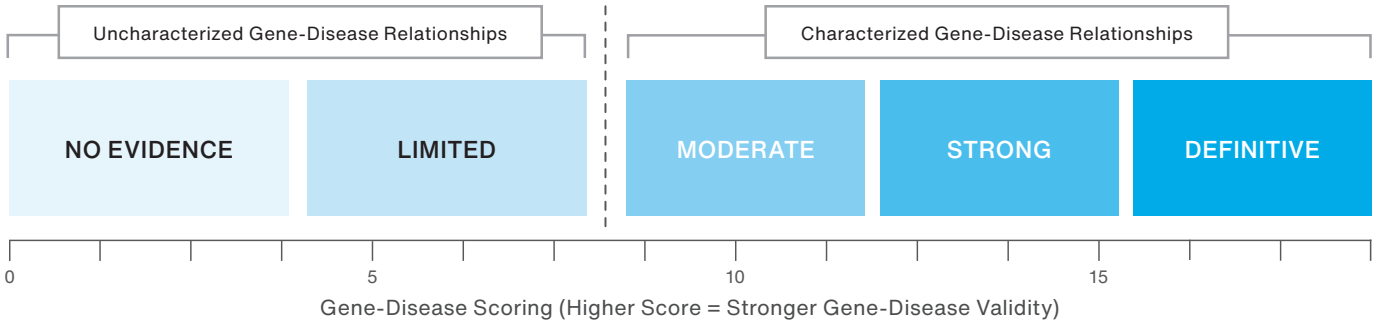
Gene-Disease Validity Scoring Criteria

CATEGORY OF EVIDENCE	POINTS
Number of unrelated patients	1 - 4
Other statistical evidence	0 - 1
Number of publications	0 - 3
Number of pathogenic variants	0 - 4
Gene function	0 - 2
Gene disruption	0 - 2
Model organism	0 - 2
SUM	



TRANSPARENCY

Ambry's objective gene-disease validity criteria defines characterized vs. uncharacterized genes. This tool supports consistent, evidence-based reporting and helps lead to new diagnoses in previously unsolved cases.[1]



ONE TEST, UNWAVERING SUPPORT

Patient for Life Program

As part of this free, inclusive service, Ambry's dedicated team of clinical scientists regularly reviews publications, databases, and other literature for gene-disease data. Ambry proactively issues reclassification reports based on published gene-disease validity standards, taking the initiative to support the health of patients indefinitely.

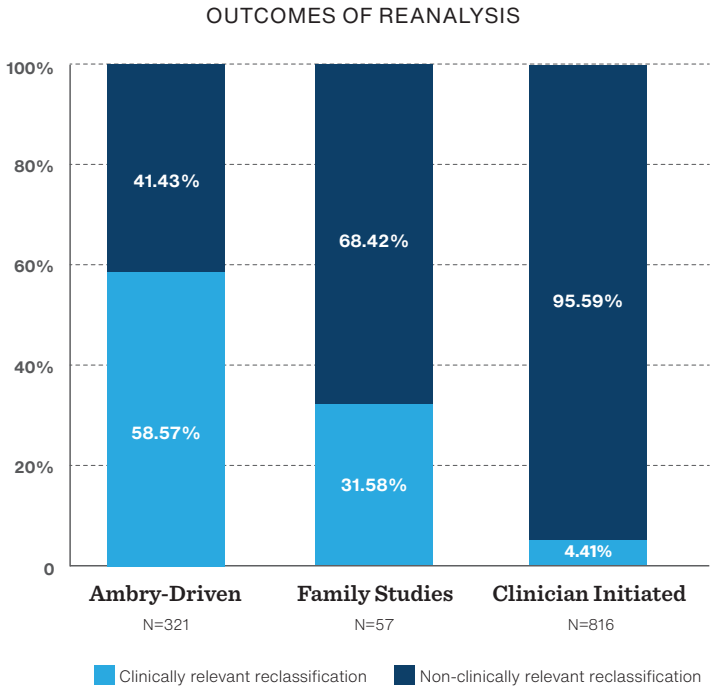
How the Program Works

Ambry's clinical scientists review newly described gene-disease relationships

Patients' exome data are reanalyzed and reclassification reports are issued for new clinically-relevant genes

Health care providers receive a reclassification report free of charge

Ambry-Driven Reanalysis Leads to Increased Clinically-Relevant Reclassifications



Backed by Ongoing Research

Our clinical scientists regularly review literature applying our published and highly-standardized gene-disease scoring system. Due to this proactive approach, recently characterized genes account for 20% of our positive findings, which may not be reported under standard processes.[1]

The Patient for Life Program's commitment to continuous database maintenance and gene-disease validity may allow for more timely identification of clinically-relevant updates and earlier patient management vs. family studies or clinician-initiated reanalysis driven by additional phenotypic data.[4]

FOR MORE DETAILS
visit ambrygen.com/patient-for-life.

Unparalleled Support and Commitment to Health

As part of our commitment to patient health, Ambry's clinically-knowledgeable Genomic Science Liaisons are available to help interpret results and answer questions that can aid in better informed medical management.

Comprehensive Coverage

ExomeNext tests offer excellent coverage across the entire genome, helping to provide answers for families.

- >97% of the exome covered with a minimum depth of coverage of 20X
- Detects gross deletions and duplications ≥5 exons
- Within mitochondrial DNA, >5% heteroplasmy is detected

Tests

Ambry offers a comprehensive set of test options for timely, cost-effective results using advanced technology and state-of-the-art bioinformatics.

Ask about the buccal swab sample option

	ExomeNext®-*Proband*	ExomeNext®-*Duo*	ExomeNext®-*Trio*
Turnaround time	6-8 weeks	6-8 weeks	6–8 weeks
Uncharacterized genes analyzed	No	No	Yes
Mitochondrial genome	Optional	Optional	Optional
Number of individuals sequenced	1	2	3
Co-segregation analysis	Included	Included	Included
Secondary findings results*	Optional	Optional	Optional

About Ambry Genetics

Ambry Genetics, a subsidiary of REALM IDx, Inc., excels at translating scientific research into clinically actionable test results based on a deep understanding of the human genome and the biology behind genetic disease. Ambry has an unparalleled track record of discoveries over 20 years and a database that continually expands through collaboration with academic, corporate and pharmaceutical partners. Being first to market with innovative products and comprehensive analysis, Ambry enables clinicians to confidently inform patient health decisions. For more information, please visit ambrygen.com.

**Reports include option for full ACMG secondary findings results,[5] for all exome-sequenced individuals*

References

1. Smith ED, et al. Classification of genes: Standardized clinical validity assessment of gene-disease associations aids diagnostic exome analysis and reclassifications. *Hum Mutat* 2017.
2. Farwell KD, et al. Enhanced utility of family-centered diagnostic exome sequencing with inheritance model-based analysis: results from 500 unselected families with undiagnosed genetic conditions. *Genet Med.* 2015.
3. Farwell Hagman KD, et al. Candidate-gene criteria for clinical reporting: diagnostic exome sequencing identifies altered candidate genes among 8% of patients with undiagnosed diseases. *Genet Med.* 2017.
4. Gage J, et al. (2021) Sustained, proactive clinical validity curation leads to higher quality panel development, reduction in VUSs, and Faster Variant Reclassification. In Patient-centered Laboratory Utilization Guidance Services Summit.

CONTACT INFORMATION

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