Ambry Genetics®
A Konica Minolta Company

ExomeNext®

REFERENCE GUIDE

ExomeNext Can Shorten Your Patient's Diagnostic Odyssey

ExomeNext leverages Ambry's industry-best data analysis team and unique bioinformatics pipeline to help bring an end to the diagnostic odyssey experienced by many undiagnosed patients.

EXCELLENT COVERAGE ACROSS THE ENTIRE GENOME

- >97% of the exome covered with a minimum depth of coverage of 20X
- Detects gross deletions and duplications ≥ 5 exons

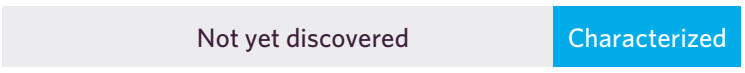
EXOME VS. PANELS

- >98% of point mutations found on panels also identified by ExomeNext.[1]

LEADERS IN EXOME ANALYSIS

First published criteria for assessing clinical validity of gene-disease relationships[2]

20,000 GENES TOTAL



Of the 20,000 genes, only ~5,000 have been characterized. Ambry is leading the efforts to define new gene-disease relationships for the community.

DIAGNOSE MORE PATIENTS

Provide an answer for **8% more patients** with our validated candidate gene analysis[3,4]

EXOMENEXT DIAGNOSTIC RATE = UP TO 38%*

30% characterized genes + 8% candidate (novel) genes

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

PATIENT FOR LIFE

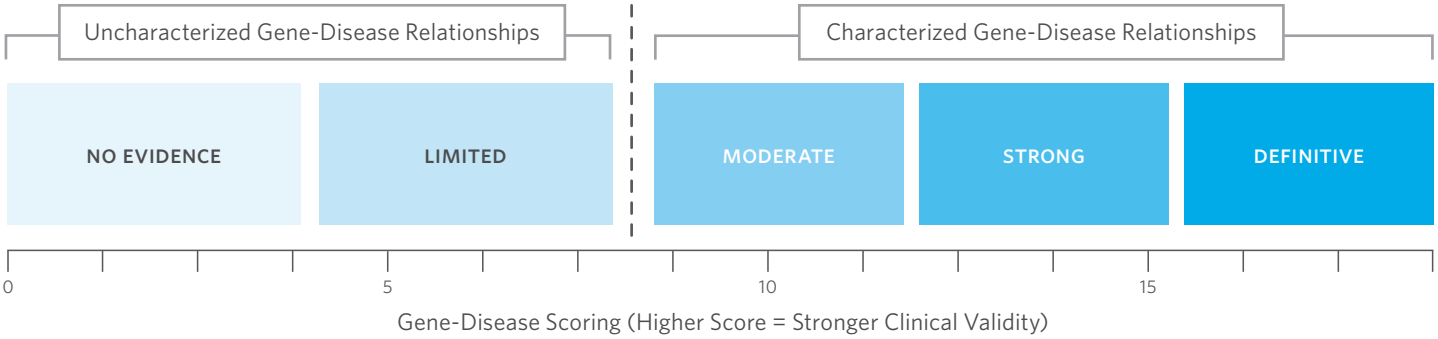
ExomeNext doesn't end with the initial report. Ambry's reanalysis program continues to look for new answers.

DATA SHARING + COLLABORATIONS + ONGOING INTERNAL RE-ANALYSIS + PHYSICIAN REQUESTED RE-ANALYSIS

6% overall reclassification

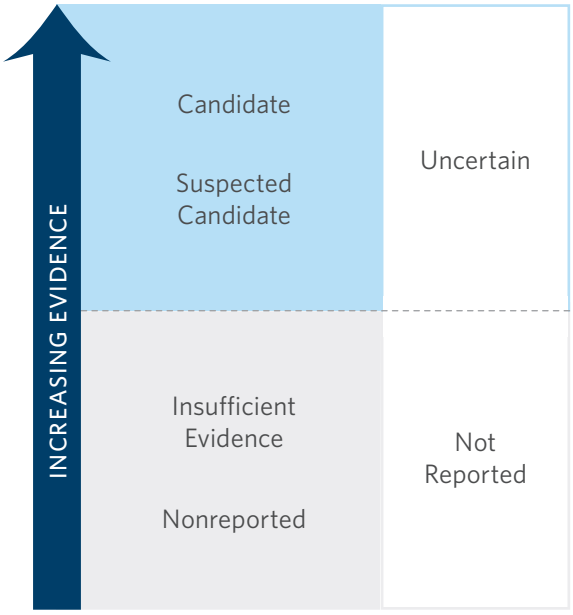
ExomeNext Adheres to Evidence-Based and Phenotype-Driven Analysis and Reporting Model[2-4]

AMBRY'S OBJECTIVE SCORING SYSTEM ENSURES CONSISTENT DATA INTERPRETATION



CLINICAL CORRELATION COMBINED WITH ACMG REPORTING CRITERIA PRODUCE ACCURATE RESULTS

CANDIDATE GENE REPORTING CRITERIA[4]



CATEGORIZATION OF CHARACTERIZED GENETIC ETIOLOGIES

Clinical Correlation	Alteration Classification	Overall Results Category
Positive	Pathogenic	POSITIVE
	Likely Pathogenic	LIKELY PATHOGENIC
	Uncertain	UNCERTAIN
Likely Positive	Pathogenic	LIKELY POSITIVE
	Likely Pathogenic	LIKELY POSITIVE
	Uncertain	UNCERTAIN
Uncertain	Likely Positive	UNCERTAIN
	Likely Pathogenic	UNCERTAIN
	Uncertain	UNCERTAIN

References

1. LaDuca H, *et al.* Exome Sequencing covers >98% of mutations identified on targeted next generation sequencing panels. PLoS One. 2017.
2. Smith ED, *et al.* Classification of genes: Standardized clinical validity assessment of gene-disease associations aids diagnostic exome analysis and reclassifications. Hum Mutat 2017.
3. 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.
4. 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.

Comprehensive Exome Testing Options for your Patient's Needs

	EXOMENEXT®- *PROBAND*	EXOMENEXT®- *DUO*	EXOMENEXT®- *TRIO*	EXOMENEXT- *RAPID®**	EXOMENEXT®- *SELECT*
Turnaround time	6-8 weeks	6-8 weeks	6-8 weeks	8 days**	2-4 weeks
Number of genes analyzed	~4,500	~4,500	Up to ~20,000	Up to ~20,000	Up to 500
Mitochondrial genome	Optional	Optional	Optional	Included	Not included
Number of individuals sequenced	1	2	3	3	1
Co-segregation analysis	Included	Included	Included	Included	Included
Secondary Findings Results	Included	Included	Included	Included	No

** Only institutional and cash billing are accepted*

*** Verbal result provided within 8 days when an informative trio is provided; full report including mtDNA analysis, co-segregation analysis, and Sanger confirmation provided within 14 days.*

Simple Ordering Process

ACCEPTED SPECIMENS: Proband and Family Members: Blood, Saliva, DNA

ORDER SUBMISSION

a. Proband specimen

b. Family Member specimens (Required for Duo/Trio orders)

c. Clinic Notes

d. Medical Necessity Form (insurance orders)

AMBRY INTERNAL ORDER REVIEW

a. Required documents & samples

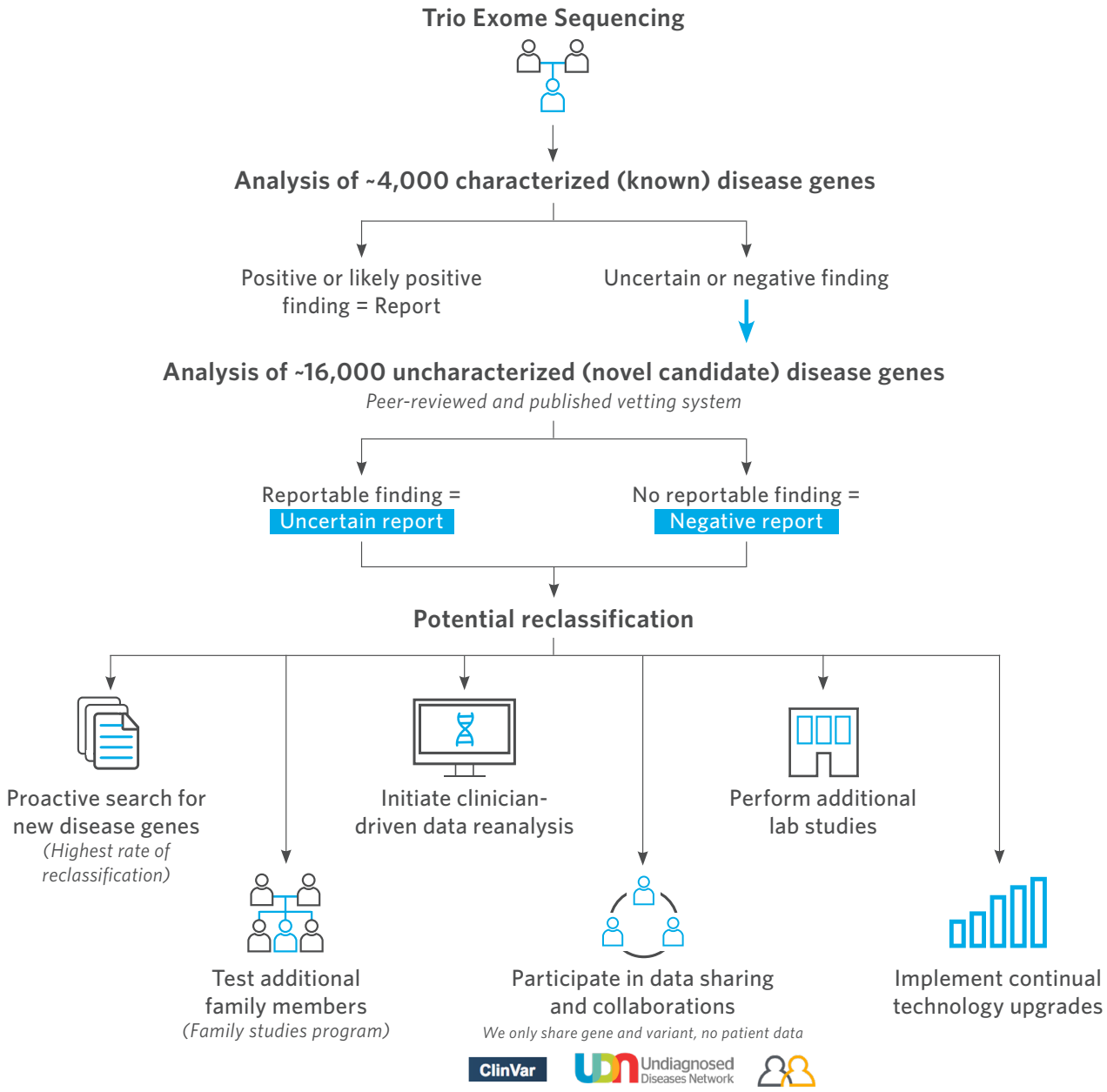
b. Test authorization (insurance orders)

RESULTS AVAILABLE IN 6-8 WEEKS OR LESS^

Results available through mail, fax, or online portal.

^Shorter TAT for ExomeNext-Rapid or ExomeNext-Select

Moving Science Forward: Finding Answers Program for Rare Disease Patients



About Ambry Genetics®

Ambry Genetics, as part of Konica Minolta Precision Medicine, excels at translating scientific research into clinically actionable test results based upon a deep understanding of the human genome and the biology behind genetic disease. Our unparalleled track record of discoveries over 20 years, and growing database that continues to expand in collaboration with academic, corporate and pharmaceutical partners, means we are first to market with innovative products and comprehensive analysis that enable clinicians to confidently inform patient health decisions. We care about what happens to real people, their families, and the people they love, and remain dedicated to providing them and their clinicians with deeper knowledge and fresh insights, so together they can make informed, potentially life-altering healthcare decisions. For more information, please visit ambrygen.com.

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