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 provi[illegible]nswers for families.

- >97% of the exome covered with a minimum depth of coverage of 20X
- Detects gross deletions and duplications ≥5 exons
- W[illegible] mitochondrial DNA, >5% [illegible]roplasmy is detecte[illegible]

Tests

Ambry offers a comprehensive set of test options for timely, cost[illegible]ective results using advanced technology and state-of-the-art bioinfor[illegible]s.

Ask about the buccal swab sample option

	ExomeNext®-*Proband*	[illegible]Next®-*Duo*	ExomeNext®-*Trio*
Turnaround time	6-[illegible]	6-[illegible]ks	6–8 weeks
Uncharacterized genes analyzed	[illegible]	No	Yes
Mitochondrial genome	Optio[illegible]	[illegible]ptional	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 [illegible] of REALI[illegible] Inc., excels at translating scientific research into clinically actionable test results based on a dee[illegible]erstan[illegible]f the hu[illegible]enome and the biology behind genetic disease. Ambry has an unparalleled track record of dis[illegible]eries over 20 y[illegible]and a da[illegible]e that continually expands through collaboration with academic, corporate and pharm[illegible]utical partners. Be[illegible]rst to ma[illegible]with innovative products and comprehensive analysis, Ambry enables clinician[illegible]confidently inform pati[illegible]health decisions. For more information, please visit ambrygen.com.

**Reports includ[illegible] for full ACMG second[illegible]findings results,[5] for all exome-sequenced individuals*

References

1. Smith ED, et al. Classifica[illegible]enes: St[illegible]dized clinical validity assessment of gene-disease associations aids diagnostic exome analysis and reclassifications. *Hum Mutat* 2017.
2. Farwell KD, et al. Enhanced u[illegible]fa[illegible]entered diagnostic exome sequencing with inheritance model-based analysis: results from 500 unselected families with undiagnosed genetic conditions. *Genet Med.*[illegible]
3. Farwell Hagman KD, et al. Candidat[illegible]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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