

Supporting Patients with Paired DNA and RNA Testing



# Comprehensive RNA Analysis for Better Variant Classification

Paired DNA/RNA genetic testing with +RNAinsight analyzes functional RNA data to help classify DNA variants. It also identifies deep-intronic mutations that may go undetected with a DNA only or reflexive RNA testing approach. This novel functional evidence is especially important in non-White populations that have been underrepresented in research and clinical testing. As a result, diagnostic yield is higher and variant of uncertain significance rate is lower, providing clarity for patients and healthcare providers.<sup>1</sup>

- Identifies more positive results<sup>1,3</sup>
- > Resolves variants of uncertain significance<sup>1,4</sup>
- > Reduces evidence gaps in non-White populations<sup>1</sup>

### ₩**91**Genes

### For Maximum Coverage

Analyzes functional RNA data across more genes for better variant classification

>70%

#### Reclassified Inconclusive<sup>2</sup>

Decreases splicing variants of unknown significance

~1/50

### Positive Patients Impacted<sup>2</sup>

Results without +RNAinsight would have been negative or inconclusive



### +RNAinsight Gene Coverage, Compatibility, and Experience\*

**+RNAinsight analyzes up to 91 genes** associated with cancers of breast, ovarian, prostate, colon, pancreatic, uterine, and more. It can be paired with most\* Ambry Genetics hereditary cancer panels to provide functional RNA information to help identify and interpret DNA variants.

### RNA by the Numbers







1,500 Variants



34 RNA Scientists



29 Publications & Presentations

### **Experience That Matters**

In 2019, Ambry Genetics became the first clinical lab to introduce paired DNA/RNA testing. Since that time, we have performed concurrent DNA and RNA testing on over 800,000 patients and classified over 1,500 unique variants. We've grown our team to 34 scientists who analyze and interpret RNA results. To meet our continued commitment to data-sharing and transparency, we've contributed to over 30 scientific posters, presentations, and publications.

### **+RNA**insight

### **Commonly Ordered With**

### CancerNext®

> 34 genes

### CancerNext-Expanded®

> 71 Genes

#### CustomNext-Cancer®

> 91 Genes

\*Not available for STAT Testing or BRCAPlus.®



Requires EDTA (DNA) & PAXgene (RNA) Specimens



14-21 Day
Turn Around Time



CAP/CLIA Certified and New York State Approved

**Technical Details** +RNAinsight analyzes transcripts for up to 91 genes depending which Ambry Genetics DNA-based Hereditary Cancer Panel it is paired with and depending on the absence or presence of RNA transcripts expressed in the blood. The results from +RNAinsight are used to provide functional RNA information to further support classification of DNA variants. It is not intended to be used as a stand-alone diagnostic test.

## For More Information CONTACT YOUR SALES REP TODAY

#### References

- 1. Horton, C., Hoang, L., Zimmermann, H., Young, C., Grzybowski, J., Durda, K., Vuong, H., Burks, D., Cass, A., LaDuca, H., Richardson, M. E., Harrison, S., Chao, E. C., & Diagnostic outcomes of concurrent DNA and RNA sequencing in individuals undergoing hereditary cancer testing. JAMA Oncology. https://doi.org/10.1001/jamaoncol.2023.55862.
- 2. Horton, C., Cass, A., Conner, B. R., Hoang, L., Zimmermann, H., Abualkheir, N., ... & Karam, R. (2022). Mutational and splicing landscape in a cohort of 43,000 patients tested for hereditary cancer. NPJ Genomic Medicine, 7(1), 49.

  3. Landrith, T., Li, B., Cass, A. A., Conner, B. R., LaDuca, H., McKenna, D. B., ... & Karam, R. (2020). Splicing profile by capture RNA-seq identifies pathogenic germline variants in tumor suppressor genes. NPJ precision oncology, 4(1).
- 4. Karam, R., Conner, B., LaDuca, H., McGoldrick, K., Krempely, K., Richardson, M. E., ... & Chao, E. (2019). Assessment of diagnostic outcomes of RNA genetic testing for hereditary cancer.