

RNA Sequencing for WES and WGS – Delivering More Actionable Diagnostic Answers

RNA sequencing (RNAseq) enhances the diagnostic capability of WGS and WES by further assessing variants associated with splicing and gene expression. Insights from this testing provide functional evidence that can enable reclassification of these variants.

RNAseq To Enhance Rare Disease Insights

RNAseq analyzes gene expression and splicing events at the transcript level

- **Order:** When placing your WGS or WES order, check the box to reflex to RNAseq for qualified variants*
- **TAT†:** 28 days
- **Sample Requirements:** Blood in EDTA tube (if additional sample is required)
- **Report:** Provided as an updated (addendum) report
- **Pricing:** No additional charge

Variant Reclassification Can Impact Patient Outcomes

- Identifying treatment options
- Adjustments in medical management
- Providing patients with additional clinical trial eligibility and research opportunities
- Informing family planning decisions

* A qualified variant meets our prediction algorithm criteria that RNAseq will provide additional functional information.

† TAT for RNAseq is calculated from the release date of the WES/WGS report or from date of sample receipt if an additional sample is requested by the laboratory. Please call client services at 1-800-411-4636 for further information.

Improving Lives Through Personalized Care And Genetic Expertise

We provide diagnostic answers that matter – empowering patients and families to make informed decisions about their health and helping healthcare providers identify, treat, and prevent disease.

Whole Genome Sequencing And Whole Exome Sequencing Are The Most Comprehensive Genetic Testing Solutions

Whole Genome Sequencing (WGS)

WGS is the most comprehensive genetic testing solution and analyzes 98% of the genome, covering both the protein-coding exons and non-coding regions of the genome.

ANALYSIS:

Genome-wide detection of SNVs, indels, CNVs, regions of homozygosity (ROH), and short tandem repeats:

- Clinically significant exonic, intronic, and regulatory variants are reported
- Clinically significant CNVs
- Uniparental disomy (UPD) reported in trio cases only
- Short tandem repeat (STR) detection in 29 genes
- Detection of mitochondrial DNA (mtDNA) variants

Whole Exome Sequencing (WES)

WES is a genetic testing solution that analyzes all exons (protein-coding sections within genes) within the human genome. Most genetic conditions are caused by variants found within the exons.

ANALYSIS:

- Detection of sequence variants, e.g. single nucleotide variants (SNVs) and indels
- Detection of copy number variants (CNVs) 3 or more exons and homozygous copy number changes of any size

WGS and WES Tests Offer

- **RNAseq* for enhanced variant classification**
- Variant interpretation aided by concurrent, trio-based analysis
- Rapid testing options with written results starting at 5 days
- Flexible sample types accepted

Baylor Genetics: Making Genetic Testing More Accessible

- Simplifying the customer experience
- Online portal, streamlined ordering with genetic counselor support
- Billing support including prior authorization, verification of benefits, patient out-of-pocket cost estimation, and patient financial assistance program

Visit **baylorgenetics.com** to view our comprehensive test menu.