Indications for Testing

- MULTIPLE CONGENITAL ANOMALIES
- NEURODEVELOPMENTAL DISORDERS
- INTELLECTUAL DISABILITY AND/OR DEVELOPMENTAL DELAY
- FAILURE TO THRIVE
- DYSMORPHIC FEATURES
- EPILEPSY SYNDROMES
- EXTENSIVE DIFFERENTIAL DIAGNOSIS
- PREVIOUS GENETIC TESTING UNINFORMATIVE

In the NICU/PICU

With written results as early as five days, consider Rapid Whole Exome Sequencing (rWES) for your patients when a genetic etiology is suspected.

End Your Patient's Diagnostic Odyssey

Getting a diagnosis that explains your patient's symptoms can be life changing. Results provide treatment options, inform medical management, and open additional research opportunities so you can focus on the best care for your patient.

EARLY DIAGNOSIS FOR PATIENT CARE

- 32% of affected individuals had changes in medical care[1]
- Save an average of $12k - $15k per patient[1]
- On average, avoid ~525 days of hospitalization[1]
- 3 out of 4 families want answers and are in favor of diagnostic tests[2]

SOURCES:
1. Am J Hum Genet.2021 Jul 1; 108(7): 1231–1238.
2. Child Neurology Foundation 2020 Assessment Survey Summary

45+ YEARS OF INNOVATION

4 MILLION+ CLINICAL TESTS PERFORMED

1 MILLION+ FAMILIES HELPED

3 THOUSAND+ TESTS OFFERED

1 MISSION EMPOWERING YOU WITH ANSWERS THAT MATTER

Baylor Genetics pioneered the history of genetic testing. Now, we're leading the way in precision medicine.

A pioneer of precision medicine for over 40 years, Baylor Genetics is a leading diagnostic genomics partner offering a full spectrum of clinically relevant genetic testing, including Whole Genome Sequencing, Whole Exome Sequencing, and focused panels. A joint venture of H.U. Group Holdings, Inc. and Baylor College of Medicine, which has the #1 NIH-funded Department of Molecular and Human Genetics, Baylor Genetics couples the fastest and most comprehensive precision diagnostics options with the support of genetic counselors to help clinicians and patients avoid a lengthy diagnostic odyssey, guide medical management, and make sure no patient with a genetic disorder gets left behind. Our test menu spans from family planning, pregnancy, neonatal and pediatric testing, oncology, and beyond.

Baylor Genetics is located in Houston's Texas Medical Center and serves clients in 50 states and 16 countries.

1.800.411.4363
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PRENATAL / NICU / PICU / PEDIATRIC / ADULT

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WES
Whole Exome Sequencing

Whole Exome Sequencing (WES) searches thousands of genes to identify changes and discover the source of your patient's medical condition. Our team of world-renowned genetics experts focus on finding the genetic cause of each patient's medical or developmental problem, guiding them to an accurate diagnosis so they can focus on the future.

Diagnosis made possible with Whole Exome Sequencing (WES)

Whole Exome Sequencing (WES) — a comprehensive precision diagnostic test for actionable insights.

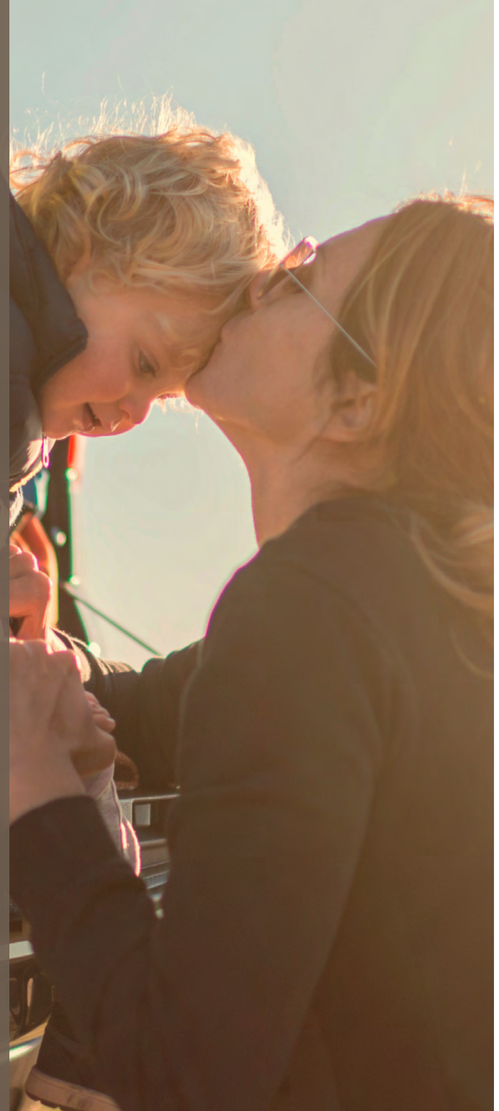
Finding the reason for your patient's medical condition can be life changing.

Whole Exome Sequencing (WES) assesses the exome, the set of all protein coding sections within the human genome. As most genetic conditions are caused by variants found within those exons, WES provides a higher diagnostic yield compared to Chromosomal Microarray Analysis (CMA) and targeted panel testing to allow for more clinically actionable insights.

BAYLOR GENETICS IS COMMITTED TO FINDING ANSWERS FOR YOU AND YOUR PATIENTS, WHICH IS WHY WE OFFER COMPANION TESTING FOR COMPLEMENTARY INSIGHTS

To assist with providing answers, our WES includes the following features:

- RNA sequencing (RNAseq), available as a reflex test for WES and Rapid WES (rWES), can help reclassify qualifying variants
- WES Reanalysis (Test Code 1900)
- CMA (Test Code 8665)
  - » Proband WES + CMA (Test Code 1530)
- Global MAPS® (Test Code 4900 & 4901)
- Comprehensive Mitochondrial DNA (mtDNA) Analysis (Test Code 2055)
  - » Trio WES + mtDNA (Test Code 1532)
  - » Trio rWES + mtDNA (Test Code 1533)
- Additional Affected Sibling (Test Code 1602)



Comparison Chart

	NICU / PICU					
	RAPID TRIO WES	RAPID DUO WES	RAPID PROBAND WES	TRIO WES	DUO WES	PROBAND WES
Test Code	1722	1723	1729	1600	1603	1500
Parental Report Included*	✓	✓	✗	✓	✓	✗
Turnaround Time (TAT) (weeks)	1 (starting at 5 days)†	1 (starting at 5 days)†	1 (starting at 5 days)†	3	3	3
Can Elect to Receive Secondary Finding	✓	✓	✓	✓	✓	✓
Raw Data Available	✓	✓	✓	✓	✓	✓

RNA Sequencing

RNA Sequencing (RNAseq) is a reflex option to our WES offerings to help reclassify qualifying variants.‡

Sample Requirements (if additional sample is required)	Blood in EDTA
TAT(days)	28¶
Report	Provided as an updated (addendum) report

For rapid testing orders, please email rapidtesting@baylorgenetics.com at the time samples are sent to the laboratory. This will alert the laboratory so that the patient's sample can be accessioned quickly. To secure emails that are sent, please add "[Secure]" in the subject line.

Test Details for Whole Exome Sequencing

GENE COVERAGE

- All genes
- Single nucleotide variants/indels in coding regions
- Copy number variants (CNV) >3 exons & homozygous copy number changes of any size
- Depth/Coverage: Average 100x genome-wide
- 2x150bp Sequencing Length: Better mapping for complex genomic regions
- Bioinformatic analysis performed on the newest genome build, GRCh38

METHODOLOGY

- Proprietary-developed bioinformatics pipeline

TURNAROUND TIME

- Written results starting at 5 days for rapid and 3 weeks for standard†

Sample Type Accepted

The following specimen types are accepted for all WES orders: blood, buccal swab, cord blood, cultured skin fibroblast, extracted DNA, and saliva. For specimen requirements, please visit www.baylorgenetics.com/whole-exome-sequencing.

Additional Reporting Options

AVAILABLE ON AN OPT-IN BASIS

ACMG Secondary Findings

The American College of Medical Genetics (ACMG) has published a series of guidelines for the reporting of these types of medically actionable or secondary findings (including PMID: 34012068). These guidelines include a list of genes, which are updated occasionally, that are considered medically actionable and indicate laboratories should report pathogenic (disease-causing) and likely pathogenic findings in these genes. In accordance with an update to this policy statement (PMID: 25356965), you may choose to opt in to receive this information.

Incidental Findings

Medically actionable variants are changes found in genes known to be associated with disease but not associated with the patient's current symptoms or clinical presentation. These variants are reported as they may cause severe, early-onset disease or may have implications for treatment and prognosis.

Potential clinically significant findings in genes with no known disease association (WES Trio only)

Rare variants including homozygous, hemizygous, compound heterozygous, and/or de novo variants in candidate genes for which there is limited available evidence of disease association are reported as variants of uncertain significance. Relevant literature is referenced if available. These are considered research findings, and further information would be required to determine the relationship to the patient's condition.

Additional Whole Exome Sequencing testing options are available. If interested, please contact your Baylor Genetics representative or email **help@baylorgenetics.com**.

* Parental Report is only included for certain test codes and if the parent(s) provide a sample. For Duo Whole Exome Sequencing, only one parent is required to submit a sample.

† The listed TAT is dependent on sample type. Please call our Client Services team at 1-800-411-4636 for further information.

‡ A qualified variant meets our prediction algorithm criteria (Splice AI) that RNAseq will provide additional functional information.

¶ The TAT for RNAseq is calculated from the release date of the WES report or from date of sample receipt if an additional sample is requested by the laboratory.