PEDIATRIC / PRENATAL

Whole exome sequencing 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.

BAYLOR GENETICS

WES
Whole Exome Sequencing

Diagnosis made possible with Whole Exome Sequencing

Indications for Testing

MULTIPLE CONGENITAL ANOMALIES

AUTISM SPECTRUM DISORDERS

NEURODEVELOPMENTAL DISORDERS

DEVELOPMENTAL DELAY

INTELLECTUAL DISABILITY

PREVIOUS GENETIC TESTING WAS UNINFORMATIVE

FAILURE TO THRIVE

DYSMORPHIC FEATURES

EPILEPSY SYNDROMES

PATIENTS WITH AN EXTENSIVE DIFFERENTIAL DIAGNOSIS

Baylor Genetics Exome Diagnosis Rates

33%
TRIO EXOME REGULAR[1]

35%
TRIO EXOME PRENATAL[2]

51%
TRIO EXOME RAPID[1]

Baylor Genetics is committed to ending the diagnostic odyssey by providing best in class exome testing.

Identifying the underlying genetic cause is powerful in understanding a patient's diagnosis. In a study performed by Baylor Genetics experts, exome test results altered medical management in up to 72% of infants, allowing for redirection of care, initiation of new sub-specialist care, medication/dietary changes, and furthering life-saving procedures.[1]

1. Meng, Linyan et al. "Use of Exome Sequencing for Infants in Intensive Care Units: Ascertainment of Severe Single-Gene Disorders and Effect on Medical Management." JAMA pediatrics vol. 171,12 (2017): e173438. doi:10.1001/jamapediatrics.2017.3438

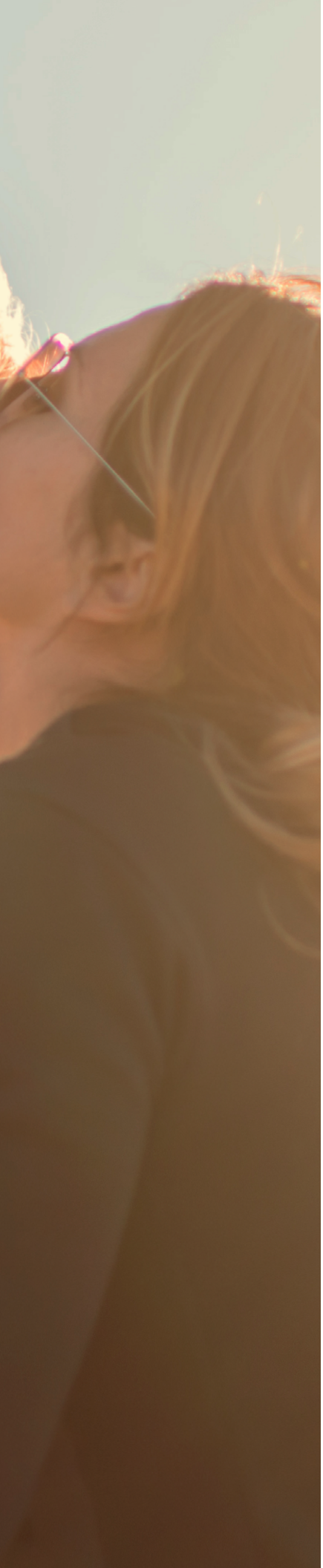
2. Normand, Elizabeth A et al. "Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder." Genome medicine vol. 10,1 74. 28 Sep. 2018, doi:10.1186/s13073-018-0582-x

When a patient's medical history and physical exam suggests an underlying genetic cause, we recommend Whole Exome Sequencing through Baylor Genetics.

Whole exome sequencing (WES) is comprehensive genetic testing which assesses the exome, the set of all exons (protein-coding sections within genes) within the human genome. Most genetic conditions are caused by variants found within these exons. Older testing technologies only look at one or a few genes at a time, which is time-consuming and could lead to several rounds of testing to reach a diagnosis. Using next-generation sequencing, WES accelerates the testing process by analyzing thousands of genes at the same time.

Finding the reason for your patient's medical condition 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.

TAKE THE FIRST STEP WITH WHOLE EXOME SEQUENCING



Comparison Chart

	PROBAND WES	RAPID PROBAND WES	DUO WES	RAPID DUO WES	TRIO WES
Test Code	1500	1729	1603	1723	1600
Consent	REQUIRED	REQUIRED	REQUIRED	REQUIRED	REQUIRED
Parents Required	or	OPTIONAL	or	REQUIRED	REQUIRED
Parental Report Included*	⊗	⊗	✓	✓	✓
TAT (weeks)	6	1 (starting at 5 days)	6	1 (starting at 5 days)	6
Can Elect to Receive Incidental Finding	✓	✓	✓	✓	✓
Raw Data Available	✓	✓	✓	✓	✓

Blood Saliva

* Parental Report is only included for certain test codes and if the parent(s) provide a sam

Considerations for Test Selection

Rapid WES Testing
When a baby is critically ill, rapid results are necessary.

BluePrint Custom Panel
The BluePrint Panel is a genetic test that enables providers to order a customized panel of up to 1,000 genes based on a patient's clinical symptoms.

Proband Only Testing
Proband WES is an option when biological parents are not available.

Total BluePrint Panel
The Total BluePrint Panel will analyze all exonic regions of the 4,800 known Mendelian disease-causing genes simultaneously to identify the rare changes in an individual's DNA that are contributing to your patient's medical condition.

The following specimen types are accepted for all exome testing options: blood, buccal swab, cord blood, cultured skin fibroblast, and purified DNA. For specimen requirements, please visit baylorgenetics.com/whole-exome-sequencing.

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

RAPID TRIO WES	BLUEPRINT CUSTOM PANEL	TOTAL BLUEPRINT PANEL	ADULT SCREENING EXOME
1722	1300	1390	1605
REQUIRED	REQUIRED	REQUIRED	REQUIRED
REQUIRED	or	or	or
✓	✗	✗	✗
1 (starting at 5 days)	6	8	10
✓	✗	✓	STANDARD REPORTING
✓	✗	✓	✓

ple. For Duo Whole Exome Sequencing, only one parent is required to submit a sample.

Duo Testing
WES Duo is an option when one biological parent is available.

Adult Screening Exome Sequencing
The Adult Screening Exome Sequencing test is used when an individual's medical history and physical exam findings are normal, but the person desires information about the potential future risk of developing a genetic disorder.

Trio Testing
WES Trio has the highest diagnostic rate of all WES testing options and is available when both biological parents are available.

Understanding the Results

Positive Results
Positive or "abnormal" results mean there is a change in the genetic material related to the patient's medical issues.

Negative Results
Negative results mean no relevant genetic change could be detected using WES. Genetic testing, while highly accurate, might not detect a change present in the genes tested. This can be due to limitations of the information available about the genes being tested, or limitations of the testing technology.

Results of Unclear Significance
WES can detect change(s) in DNA that do not have a clear meaning known as a variant of uncertain significance (VUS). Every person has changes in their DNA; not all of these changes cause medical issues. Studies of family members may help resolve the uncertainty. As our understanding of genetics increases, it may also be possible to determine the significance of these variants.

Additional Reporting Options

ACMG Secondary Findings
The American College of Medical Genetics (ACMG) has published guidelines for the reporting of medically actionable or secondary findings (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) findings in these genes. These findings are available on an opt-in basis.

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 understand if any disease.

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. These findings are available on an opt-in basis.

40+ YEARS OF INNOVATION

4 MILLION+ CLINICAL TESTS PERFORMED

1 MILLION+ FAMILIES HELPED

3 THOUSAND+ TESTS OFFERED

1 MISSION IMPROVE HEALTHCARE THROUGH GENETICS

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

Baylor Genetics is a joint venture of H.U. Group Holdings Inc. and Baylor College of Medicine, including the #1NIH funded Department of Molecular and Human Genetics. A pioneer of precision medicine for over 40 years, Baylor Genetics now offers a full spectrum of clinically relevant genetic testing, access to world-renowned experts, and the confidence to provide patients with the best care.

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