



♡ 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 a better diagnosis so they can focus on the future.

BAYLOR GENETICS

WES

Whole Exome
Sequencing

One test, more answers.
Whole Exome Sequencing.



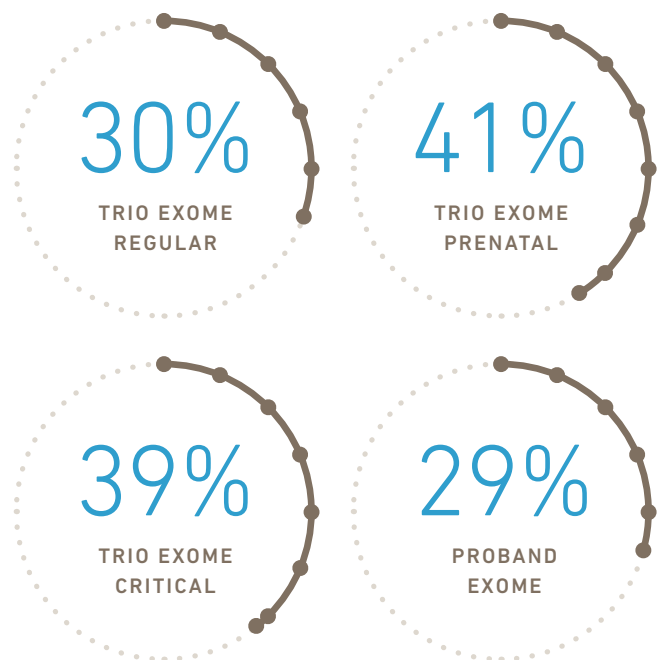
Indications for Testing

EXHAUSTED GENETIC TESTING OPTIONS

PATIENTS WITH A LONG LIST OF
DIFFERENTIAL DIAGNOSES

ATYPICAL PRESENTATION OF DISEASE

Baylor Genetics Exome Diagnosis Rates



THE EXOME DIAGNOSIS RATE AT BAYLOR GENETICS IS APPROXIMATELY 30% FOR PROBAND OR TRIO EXOME, AND APPROXIMATELY 40% FOR PRENATAL TRIO AND CRITICAL TRIO EXOME. ORDERING THE EXOME TEST WITH A SHORTER TURN-AROUND TIME CAN IMPACT HEALTH OUTCOMES IN A SIGNIFICANT WAY, ESPECIALLY FOR YOUNGER PATIENTS.

When a patient's medical history and physical exam strongly suggests an underlying genetic cause, we recommend whole exome sequencing.

Whole exome sequencing (WES) is available to patients who are searching for a unifying diagnosis for multiple medical issues. Unlike older technology where only one gene could be tested at a time, Baylor Genetics uses state-of-the-art technology to study a person's exome. The exome refers to all our exons, which are the important protein-coding sections of DNA that are contained in our genes. The majority of DNA changes that may cause a genetic disorder are found in exons.



Think of genes like sentences in the instruction manual for our bodies.




























Scientists know what the sentences should say. Sequencing is a technology that looks at each letter in every sentence of DNA in the exome allowing scientists to compare the DNA of a person who has medical issues to the DNA from a person without medical issues. WES simultaneously looks at thousands of portions of genetic material at the same time. However, this technology cannot see all genetic changes that may lead to genetic conditions, which is why additional testing may be recommended.

Finding an answer to your patient's medical condition through whole exome sequencing can be life changing. Results can open options to treatment and additional research opportunities through Baylor College of Medicine.

ALL IT TAKES IS ONE TEST TO GET MORE ANSWERS



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)	10	1 (starting at 5 days)	10	1 (starting at 5 days)	6
Can Elect to Receive Incidental Finding					
Raw Data Available					
Consent Available in Other Language					



Blood



Saliva

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

Considerations for Test Selection

WES Trio (1600)

WES Trio is beneficial when both biological parents are available and want to know all disorder-causing genetic changes.

Critical WES Trio (1722)

When your patient's baby is critically ill, rapid results are necessary. Both biological parents must be available for this test.

WES Duo (1603, 1623)

WES Duo is beneficial when only one biological parent is available and wants to know all disorder-causing genetic changes. A rapid option is available for patients that are critically ill.

Proband WES (1500, 1729)

Proband WES is an option when one or both biological parents are not available. A rapid option is available for patients that are critically ill.

Blueprint Custom Panel (1300)

The Blueprint panel is a genetic test that enables physicians to order a customized panel of genes based on your patient's clinical symptoms.

Total Blueprint Panel (1390)























The Total Blueprint Panel will analyze all the 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 concerns.

Adult Screening Exome Sequencing (1605)

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

The following specimen types are accepted for all exome testing options: blood, buccal swab, 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.

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

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

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 or “normal” results mean no relevant genetic change could be detected using WES. This does not mean that there is no genetic change, but it may mean WES could not detect it.






Results of Unclear Significance

WES can detect change(s) in DNA that do not have a clear meaning. Every person has changes in their DNA; not all of these changes cause medical issues.

Incidental Findings

WES can sometimes detect a change in a person’s DNA unrelated to the reason for the sample being sent for testing. If this change is medically significant, it is called an incidental finding. Possible incidental findings include mutations that increase a person’s risk for cancer or heart disease. It is optional to receive results on incidental findings. Information regarding adult-onset dementia syndromes, such as Alzheimer’s disease, or other adult-onset neurological conditions will not be reported.

PRENATAL WES

Test Code	1622
Consent	REQUIRED
Parents Needed	 REQUIRED
Method of Parent Testing	EXOME
TAT (weeks)	3 (not including tissue culture)
Can Elect to Receive Incidental Finding	 FETAL  PARENTAL
Raw Data Available	
Consent Available in Other Language	

Prenatal WES Trio is used when prenatal imaging detects an anomaly that strongly suggests there is an underlying genetic etiology. Prenatal WES Trio is often considered after fetal chromosomal microarray analysis or other prenatal testing has been non-diagnostic.

FETAL REPORT INCLUDES:

- Pathogenic or likely pathogenic variants in disease genes related to the prenatal indications
- Variants in disease genes unrelated to the prenatal indications, but likely to cause significant disorders during childhood

PARENTAL REPORT INCLUDES

- Pathogenic variants will be reported as medically actionable if discovered in genes included in the ACMG policy statement regarding recommendations for reporting of incidental findings
- If requested, only included for WES Trio (1600) and Critical WES Trio (1722)



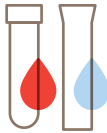
40+ YEARS OF INNOVATION



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 #1 NIH 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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