BAYLOR GENETICS

Inpatient | Outpatient

Neonatal · Pediatric · Adult

Whole Genome Sequencing

The Most Comprehensive First-Tier Test for Diagnosing Rare Disease, and Unexplained Symptoms

Answers to Guide Patient Care and Outcomes

More Informed Results with Multimodal Data, AI-Enhanced Expert Interpretation, and End-to-End Support

GOLD 2024 STEVIE WINNER FOR TECHNOLOGY EXCELLENCE

Table of Contents

For over 45 years, Baylor Genetics has advanced genetic testing—delivering answers that inform care for patients and providers alike.

Our Whole Genome Sequencing (WGS) workflow analyzes multimodal data across diverse clinical modalities and incorporates AI-driven insights alongside clinically curated databases to generate informed results. This enables one of the fastest and most comprehensive genomic tests available to support differential diagnosis in complex or urgent clinical scenarios, particularly those involving rare diseases or unexplained symptoms.

Considerations and Limitations. It is important to understand that genetic tests, even if negative, cannot rule out the presence of every variant. Genetic testing, while highly accurate, might not detect a variant present in the gene(s) tested. This can be due to limitations of the information available about the gene(s) being tested, or limitations of the testing technology. It is not possible to exclude risks for all genetic diseases for you/your child and your family members. It is possible that even if the test identifies the underlying genetic cause for the disease in your family, this information may not help in predicting the progression of disease or change management or treatment of disease.

Testing Options

Rapid WGS (rWGS) for Inpatient Care

Results in 5 Days*

Comprehensive genetic testing ideal for critically-ill patients in the NICU/PICU/CVICU with a suspected genetic disease.

Rapid Proband WGS (Test Code: 1829)
Single-Patient (Proband) Testing

Rapid Duo WGS (Test Code: 1823)
Includes Patient (Proband) + One Biological Parent

Rapid Trio WGS (Test Code: 1822)
Includes Patient (Proband) + Both Biological Parents

Rapid Quad WGS (Test Code: 1824)
Includes Patient (Proband) + Both Biological Parents + Closely Related Family Member

Indications for Testing

- Cardiac Arrest
- Congenital Anomalies (e.g., cardiac, skeletal, & genitourinary anomalies)
- Developmental Delays
- Epilepsy
- Extensive Differential Diagnosis
- Failure to Thrive
- Hypotonia
- Intellectual Disability
- Immunodeficiencies
- Metabolic Disturbances
- Neurodevelopmental Disorders
- Neuromuscular Disorders
- Previous Genetic Testing Uninformative
- Prolonged and / or Recurrent Hospital Stays
- Respiratory Insufficiency at Term

WGS for Outpatient Care

Results in 3 Weeks*

Comprehensive genetic testing for patients suspected to have a genetic condition with a complex phenotype.

Proband WGS (Test Code: 1810)
Single-Patient (Proband) Testing

Duo WGS (Test Code: 1803)
Includes Patient (Proband) + One Biological Parent

Trio WGS (Test Code: 1800)
Includes Patient (Proband) + Both Biological Parents

Quad WGS (Test Code: 1804)
Includes Patient (Proband) + Both Biological Parents + Closely Related Family Member

Indications for Testing

- Autism Spectrum Disorder
- Cerebral Palsy
- Congenital Anomalies (e.g., cardiac, skeletal, & genitourinary anomalies)
- Developmental Delays
- Epilepsy
- Extensive Differential Diagnosis
- Failure to Thrive
- Hypotonia
- Intellectual Disability
- Immunodeficiencies
- Metabolic Disturbances
- Neurodevelopmental Disorders
- Neuromuscular Disorders
- Previous Genetic Testing Uninformative
- Prolonged and / or Recurrent Hospital Stays
- Vision & Hearing Loss

* Turnaround time can vary based on factors such as collection date, sample quality quantity and completeness of patient information provided.

Multimodal Data

Baylor Genetics' Whole Genome Sequencing (WGS) includes a range of modalities and analysis methods that extend beyond standard sequencing. This strategy offers a broader analysis and deeper insight into disease mechanisms, helping to inform accurate diagnosis and support clinical care decisions.

Explore Additional Modalities Beyond Traditional Sequencing

Genome Analysis

This is the starting point of our multimodal approach. It enables the detection of variants across the genome, which may explain a patient's symptoms. This comprehensive view may help uncover insightful information by identifying multiple types of variants (see page 11 for details) and sets the foundation for deeper insights through additional layers of multimodal analysis.

RNA Sequencing Analysis (RNA-Seq)

Supplemental targeted testing for WGS adds functional evidence to strengthen the interpretation of patient-specific variants, helping reduce variants of uncertain significance (VUS) and increase diagnostic yield.[1]

Complex Structural Variant (SV) Analysis

Complex Structural (SV) Analysis: Complex SVs like rearrangements can disrupt the normal function of a gene and contribute to a broad range of genetic diseases. SV analysis can provide better clarity for patients than traditional sequencing methods by using Optical Genome Mapping (OGM) for unmatched high-resolution detail.[2]

Short Tandem Repeat (STR) Analysis

STRs are repetitive DNA sequences that can lead to many neurological, neuromuscular, and other genetic disorders. While challenging to detect with WGS alone, advanced testing strategies, including long-read sequencing (LR), are used to assess and confirm 58 clinically significant STRs (see pages 13-14 for more details).

Methylation Analysis of *FMR1*

Methylation analysis of the *FMR1* gene can help confirm Fragile X-associated conditions in rare situations where traditional indicators (STR size) do not reflect the patient's clinical presentation. This analysis evaluates gene expression for functional genomic information to guide patient diagnosis.

Mitochondrial DNA (mtDNA) Analysis

High-depth mtDNA sequencing enables sensitive detection of mitochondrial variants, including a mixture of healthy and affected mtDNA (heteroplasmy) down to 5%.[3] This improves identification of mitochondrial disorders, which often present with neurological and multi-system features.

Uniparental Disomy (UPD) Analysis

UPD occurs when both copies of a chromosome or chromosomal region are inherited from the same parent. When parental samples are available, WGS can identify these patterns to help explain imprinting-related conditions.[4]

Metabolomic Analysis

Evaluate hundreds of metabolites with Global MAPS® to assess functional disruptions in biochemical pathways for inborn errors of metabolism. When paired with WGS, it can show how genetic changes may affect patients, helping to clarify uncertain or unexplained findings.[5]

NOTE: This is ordered as a separate test (Global MAPS®) and is reported separately from WGS.

1 RNA-Seq is only performed on qualified variants that meet the prediction algorithm criteria that suggests additional functional evidence can be provided. If a qualified variant is found by WGS, RNA-Seq can only be performed on blood samples. Additional specimens may be requested as needed.

2 For select cases when appropriate as determined by Baylor Genetics, WGS may be supplemented with Optical Genome Mapping (OGM) to aid with result clarification for complex structural variant analysis. Complex structural variant analysis can only be performed on blood samples. Additional specimens may be requested as needed.

3 Based on internal data.

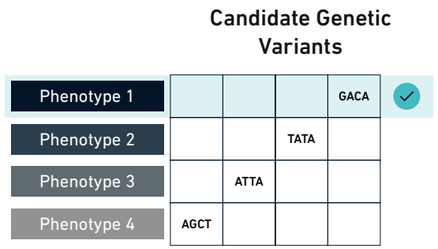
4 Requires comparator samples from both biological parents.

5 Global MAPS is a semi-quantitative metabolomic profiling screen that analyzes disruptions in both individual analytes and pathways related to biochemical abnormalities. Must be ordered separately from WGS testing.

Complex SV analysis with OGM as well as STR and methylation analyses using long-read sequencing (LR) are not approved in New York State. A final report will be issued prior to initiating RNA-Seq or complex SV analysis, with any resulting updates provided through an addended report. Comparator samples are excluded from RNA-Seq, complex SV (with OGM), and STR analyses. RNA-Seq, methylation, complex SV, and some STR analyses are not available as stand-alone tests.

AI-Enhanced Expert Interpretation

Baylor Genetics is committed to quality and delivering prompt, accurate results through a clinically validated workflow—integrating AI technology, unique rare disease data sets and a specialized medical/scientific staff.



Artificial Intelligence (AI) Workflow

Our proprietary interpretation pipeline is integrated with an industry-leading AI solution to accelerate candidate gene variant prioritization—leveraging globally-esteemed databases and curated clinical literature to reveal underlying causes of disease.

Expert Interpretation with BCM Faculty

Unique to Baylor Genetics, many of our ABMGG-certified directors are also Baylor College of Medicine (BCM) faculty. Bringing their deep clinical and research expertise in rare disease they lead reporting of Whole Genome Sequencing performed at Baylor Genetics. This close collaboration reinforces our commitment to expert interpretation and quality assurance to deliver the highest standard of genetic testing.

Exclusive UDN Database Access

Baylor Genetics has been the sequencing core site and an active partner of the Undiagnosed Diseases Network (UDN) since 2015. As an integral contributor to the UDN, Baylor Genetics collaborates closely on rare disease cases and leverages data from previously uncharacterized conditions. This partnership enables deeper understandings and more definitive diagnostic support, particularly in complex or atypical clinical presentations.

Moving Clinical Genetics Forward with Research

For over 45 years, Baylor Genetics has been at the forefront of genetic testing, empowering patients and leading genomic research, with publications in the *New England Journal of Medicine, Genome Medicine*, and other highly respected journals. We routinely present at scientific conferences including the Child Neurology Society, American College of Medical Genetics and Genomics, American Society of Human Genetics, and National Society of Genetic Counselors.

End-to-End Support

Our full-service team of clinical geneticists, genetic counselors and client service professionals work alongside you—from order to report—ensuring fast, prompt and reliable support throughout the testing journey.

Available Services for WGS Patients

Post-Test Genetic Counseling

Patients who receive a positive or uncertain result with WGS may qualify for genetic counseling with a certified genetic counselor who can help explain test results. Restrictions apply.

Phlebotomy Services

If a patient is not able to get access to a blood draw service, we may have options to help find a convenient phlebotomy service and prevent further delays in testing.

Patient Financial Assistance Team

Dedicated support to guide patients and their family with understanding insurance coverage, explore qualifying financial support options, and explain any out of pockets costs.

Multiple Testing Types

Comparator testing is available (Duo, Trio, and Quad), which involves testing the patient along with biological family members—such as parents or siblings—as comparators to more accurately identify genetic causes of disease. Rapid testing is also available with clinical reports provided in 5 days.*

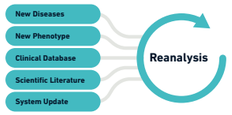
EMR Integration with Epic Aura

Organizations using Epic Aura will have the ability to order WGS from Baylor Genetics' and view results directly in the EHR—supporting fast, informed treatment decisions and improving the provider experience.

Flexible Sample Types

Our WGS workflow is validated for multiple sample types to making testing more accessible and reliable for patients with rare diseases. This ensures high-quality results, even when traditional sample collection isn't possible.

- Whole Blood
- Buccal Swab
- Saliva
- Cord Blood
- Extracted DNA
- Cultured Skin Fibroblast



Physician-Initiated Reanalysis

As our understanding of genomics continues to evolve, physician-initiated reanalysis enables reassessment of the patient's genetic data and presentation in light of new clinical findings or new research and insights. This process offers patients and providers a renewed opportunity to uncover meaningful answers.

* Turnaround time can vary based on factors such as collection date, sample quality/quantity, and completeness of patient information provided.

WGS Test Features and Details

WGS is the most comprehensive test available through Baylor Genetics. It analyzes up to 98% of the human genome, detecting known and potential disease-causing variants that may not be identified by more targeted genetic testing methods. Additionally, WGS covers both the protein-coding exons and clinically significant non-coding regions of the genome.

PCR-Free WGS

Compared to PCR-based WGS methods, our WGS provides uniform genome sequencing coverage to reduce implication bias, which helps better variant calling accuracy.

Genomic Findings

- Single Nucleotide Variants (SNVs)
- Structural Variants (SVs)
- Short Tandem Repeat (STRs)
- Copy Number Variants (CNVs)
- Methylation of *FMR1*
- Mitochondrial DNA (mtDNA) Analysis
- Uniparental Disomy (UPD)
- Regions of Homozygosity (ROH)

40x Sequencing Coverage

At 40x coverage, the genome is read an average of 40 times, significantly increasing the accuracy and reliability of variant detection.

Secondary & Incidental Findings

Medically actionable variants that might not be associated with a patient's current disease still might provide important information for their medical management in the future. Baylor Genetics provides the option for the reporting of pathogenic and likely pathogenic variants in genes on the ACMG Secondary Findings list.

In addition, other incidental findings unrelated to the patient's phenotype but that might impact their medical management can be separately opted into.

Short Tandem Repeat (STR) Analysis

We analyze a curated set of genes known to contain STRs, which are associated with a variety of genetic disorders. The following is a list of genes with STRs that we evaluate as part of our comprehensive WGS-based testing approach.

ASSOCIATED DISEASE	GENE
Blepharophimosis, epicanthus inversus, and ptosis, types 1 and 2 [MIM:110100]	*FOXL2*
Central hypoventilation syndrome, congenital, with or without Hirschsprung disease [MIM:209880]	*PHOX2B*
Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome [MIM:614575]	*RFC1*
Cleidocranial dysplasia [MIM:119600]	*RUNX2*
Corneal dystrophy, Fuchs endothelial, 3 [MIM:613267]	*TCF4*
Creutzfeldt–Jakob disease [MIM:123400]; Gerstmann-Sträussler-Scheinker syndrome [MIM:137440]; Huntington disease-like 1 [MIM:603218]	*PRNP*
Dentatorubral-pallidoluysian atrophy [MIM:125370]	*ATN1*
Desbuquois dysplasia 2 [MIM:615777]	*XYLT1*
Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg) [MIM:254800]	*CSTB*
Epiphyseal dysplasia, multiple, 1 [MIM:132400] Pseudoachondroplasia [MIM:177170]	*COMP*
Familial adult myoclonic epilepsy 2 [MIM:607876]	*STARD7*
Familial adult myoclonic epilepsy 4 [MIM:615127]	*YEATS2*
Familial adult myoclonic epilepsy type 1 [MIM:601068]	*SAMD12*
Familial adult myoclonic epilepsy type 3 [MIM:613608]	*MARCHF6*
Fragile X syndrome [MIM:300624]; Fragile X tremor/ataxia syndrome [MIM:300623]; Premature ovarian failure 1 [MIM:311360]	*FMR1*
Friedreich ataxia [MIM:229300]	*FXN*
Frontotemporal dementia and/or amyotrophic lateral sclerosis 1 [MIM:105550]	*C9orf72*
Global developmental delay, progressive ataxia, and elevated glutamine [MIM:618412]	*GLS*
Hand-foot-genital syndrome [MIM:140000]	*HOXA13*
Holoprosencephaly-5 [MIM:609637]	*ZIC2*
Huntington disease [MIM:143100]	*HTT*
Huntington disease-like 2 [MIM:606438]	*JPH3*
Intellectual developmental disorder, X-linked 109 [MIM:309548]	*AFF2*
Intellectual Disability, FRA12A type [MIM:136630]	*DIP2B*
KINSSHIP Syndrome [MIM:619297]	*AFF3*
Machado-Joseph disease [MIM:109150]	*ATXN3*
Myotonic dystrophy 1 [MIM:160900]	*DMPK*
Myotonic dystrophy 2 [MIM:602668]	*CNBP*
Neuronal intranuclear inclusion disease [MIM:603472]; Tremor, hereditary essential, 6 [MIM:618866]	*NOTCH2NLC*

Short Tandem Repeat (STR) Analysis (continued)

ASSOCIATED DISEASE	GENE
Neuronopathy, distal hereditary motor, autosomal recessive 7 [MIM:619216]	*VWA1*
Neuropathy, hereditary sensory and autonomic, type VIII [MIM:616488]	*PRDM12*
Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia [MIM:613563]	*CBL*
Oculopharyngeal muscular dystrophy [MIM:164300]	*PABPN1*
Oculopharyngeal myopathy with leukoencephalopathy 1 [MIM:618637]	*NUTM2B-AS1*
Oculopharyngodistal myopathy 1 [MIM:164310]	*LRP12*
Oculopharyngodistal myopathy 2 [MIM:618940]	*GIPC1*
Oculopharyngodistal myopathy type 4 [MIM:619790]	*RILPL1*
Oculopharyngodistal myopathy-5 [MIM:621446]	*ABCD3*
Partington syndrome [MIM:309510]; Intellectual developmental disorder, X-linked 29 [MIM:300419]; Developmental and epileptic encephalopathy-1 [MIM:308350]	*ARX*
Robin sequence with cleft mandible and limb anomalies [MIM:268305]	*EIF4A3*
Spinal and bulbar muscular atrophy of Kennedy [MIM:313200]	*AR*
Spinocerebellar ataxia 1 [MIM:164400]	*ATXN1*
Spinocerebellar ataxia 10 [MIM:603516]	*ATXN10*
Spinocerebellar ataxia 12 [MIM:604326]	*PPP2R2B*
Spinocerebellar ataxia 17 [MIM:607136]	*TBP*
Spinocerebellar ataxia 2 [MIM:183090]	*ATXN2*
Spinocerebellar ataxia 27B [MIM:620174]	*FGF14*
Spinocerebellar ataxia 31 [MIM:117210]	*BEAN1*
Spinocerebellar ataxia 36 [MIM:614153]	*NOP56*
Spinocerebellar ataxia 37 [MIM:615945]	*DAB1*
Spinocerebellar ataxia 4 [MIM:600223]	*ZFHX3*
Spinocerebellar ataxia 51 [MIM:620947]	*THAP11*
Spinocerebellar ataxia 6 [MIM:183086]	*CACNA1A*
Spinocerebellar ataxia 7 [MIM:164500]	*ATXN7*
Spinocerebellar ataxia 8 [MIM:608768]	*ATXN8OS/ATXN8*
Synpolydactyly [MIM:186000]	*HOXD13*
X-linked panhypopituitarism [MIM:312000]; X-linked intellectual disability with isolated growth hormone deficiency [MIM:300123]	*SOX3*
X-linked VACTERL syndrome [MIM:314390]	*ZIC3*

45+ YEARS OF INNOVATION

4 MILLION+ CLINICAL TESTS PERFORMED

1 MILLION+ FAMILIES HELPED

1 MISSION EMPOWERING YOU WITH ANSWERS THAT MATTER

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

A pioneer of precision medicine for over 45 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. 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.

1.800.411.4363 | BAYLORGENETICS.COM

The tests described have been developed and their performance characteristics determined by the CLIA-certified and CAP-accredited laboratory performing the test. These tests are laboratory-developed tests (LDTs) and have not been cleared or approved by the U.S. Food and Drug Administration (FDA). Clinical testing is performed in compliance with the Clinical Laboratory Improvement Amendments (CLIA) and the standards of the College of American Pathologists (CAP), ensuring high quality and reliability in laboratory practices. © 2025 Baylor Genetics, Inc. All Rights Reserved.

03.23.26