PEDIATRIC / ADULT

Introducing Whole Genome Sequencing (WGS), a comprehensive genetic test bringing your patient closer to reaching a diagnosis in one test.

BAYLOR GENETICS

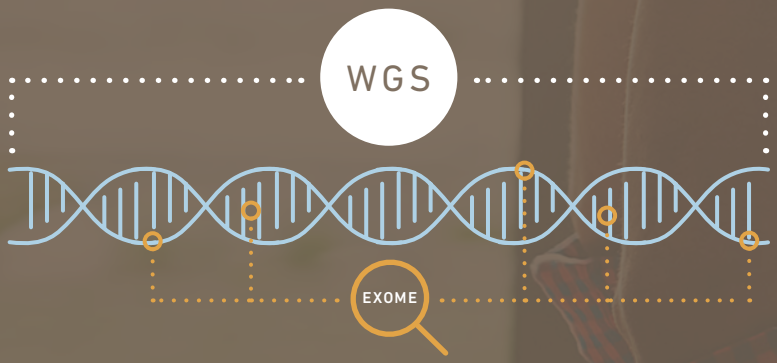
WGS
Whole Genome Sequencing

Diagnosis made possible with Whole Genome Sequencing

Whole Genome Sequencing (WGS) is a comprehensive test that covers up to 98% of the whole human genome.

Advancements in next-generation sequencing technology are poised to revolutionize diagnostic testing by the introduction of Whole Genome Sequencing (WGS). Your patient and family may have spent years undergoing multiple genetic testing without avail, which is why WGS may be the perfect test to potentially end their diagnostic odyssey. Alternatively, you may be managing a new patient or newborn with complex medical issues for whom a comprehensive approach would be the appropriate first-line test.

WGS is a powerful tool for detecting known and potential disease-causing variations. While other traditional genetic tests, such as whole exome sequencing and chromosomal microarray analysis, impart only particular changes in a patient's DNA, WGS is advantageous as a single test to detect variants that may not be amenable to current genetic testing.



The exome accounts for only 1 – 2% of the whole genome making WGS the most comprehensive test for your patient.

WGS can capture virtually all disease-causing genetic variations including single-nucleoatide variants, small insertion/deletions, and copy number variants. WGS has multiple applications ranging from being an effective diagnostic strategy for the clinical diagnosis of genetic and inherited disorders to treatment planning and pharmacogenomic uses.

WHEN TO ORDER WGS?

If your patient has a broad spectrum of phenotypic features without an obvious clinical diagnosis, instead of ordering multiple genetic tests, take advantage of WGS as the single genetic test to provide your patient with a genetic diagnosis.

If your patient and family have spent years undergoing multiple genetic testing without avail, WGS is the perfect test to potentially end their diagnostic odyssey.

	WHOLE GENOME SEQUENCING
Test Code	1800
Specimen Types	Blood, Cultured Cell Lines, DNA
Consent	REQUIRED
Parents Needed	REQUIRED
Parental Report Included	✓
TAT (weeks)	8 – 10
Can Elect to Receive Carrier Finding(s)	✓
Can Elect to Receive Incidental Finding(s)	✓
Raw Data Available	✓

Testing Details

Methodology

- Next-generation sequencing (NGS) on NovaSeq
- Custom-developed bioinformatics pipeline

Gene Coverage

- All genes
- Single nucleotide variants/indels in coding and non-coding regions
- Copy number variants (CNV)
- Depth/Coverage: Minimum 40x genome-wide
- PCR-free: Better CNV/SV detection
- 2x150bp, 550 insert size: Better CNV/SV/TNR detection and better mapping to distinguish pseudogenes
- Bioinformatic analysis performed on the newest genome build, GRCh38

Blood Cultured Cell Lines DNA

Understanding the Results

Positive Results

Positive or "abnormal" results mean there is a genetic change related to the patient's medical issues. Positive results can come in the form of the following genetic changes:

Related to the Patient's Indications

- Single nucleotide variants/indels
- Copy number variants (CNV)
  - Structural variants (SV)
  - Absence of Heterozygosity (AOH)

Unrelated to the Patient's Indications (Opt-in only)

- Carrier findings
  - Disorders recommended by the ACMG & ACOG
- Incidental findings
  - Incidental findings recommended by the ACMG

Negative Results

A genetic change related to the patient's issues was not detected using this test. This does not guarantee that the patient is free from genetic disorders.

Results of Unclear Significance

WGS 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 these changes cause medical issues. Studies of family members may help resolve the uncertainty.

Incidental Findings

WGS 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.

Specimen Requirements

TYPE	REQUIREMENTS	SHIPPING CONDITIONS
BLOOD	3-5 cc (2 yrs-adult), 2-3 cc (newborn-2 yrs) in EDTA (purple-top) tube (molecular testing). Attach clinical notes and concurrent laboratory reports.	Ship at room or refrigerated temperature in an insulated container by overnight courier. Do not heat or freeze. Specimen should arrive in the laboratory within 24-48 hours of collection.
CULTURED CELL LINES	Send two T25 flasks at 80-100% confluence.	Ship at ambient temperature (18-25°C/64-77°F) in an insulated container by overnight courier. Cell line specimens should arrive in the laboratory within 48 hrs of collection. Do not heat or freeze.
DNA (Extracted)	At least 20 ug with a minimum average concentration of 50 ng/µL. Attach clinical notes and/or concurrent laboratory reports as applicable. Extracted DNA will only be accepted if the isolation of nucleic acids for clinical testing occurs in a CLIA-certified laboratory or a laboratory meeting equivalent requirements as determined by the CAP and/or the CMS.	Ship at room temperature in an insulated container by overnight courier. May also be shipped frozen on a minimum of 10 lbs of dry ice in an insulated container by overnight courier.

Indications for Testing

- MULTIPLE CONGENITAL ANOMALIES
- AUTISM SPECTRUM DISORDERS
- NEURODEVELOPMENTAL DISORDERS
- DEVELOPMENTAL DELAY
- INTELLECTUAL DISABILITY
- FAILURE TO THRIVE
- DYSMORPHIC FEATURES
- EPILEPSY SYNDROMES
- PATIENTS WITH AN EXTENSIVE DIFFERENTIAL DIAGNOSIS

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 Miraca Holdings, Inc. and Baylor College of Medicine, including the #1NIH funded Department of Molecular and Human Genetics. A pioneer of precision medicine for nearly 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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