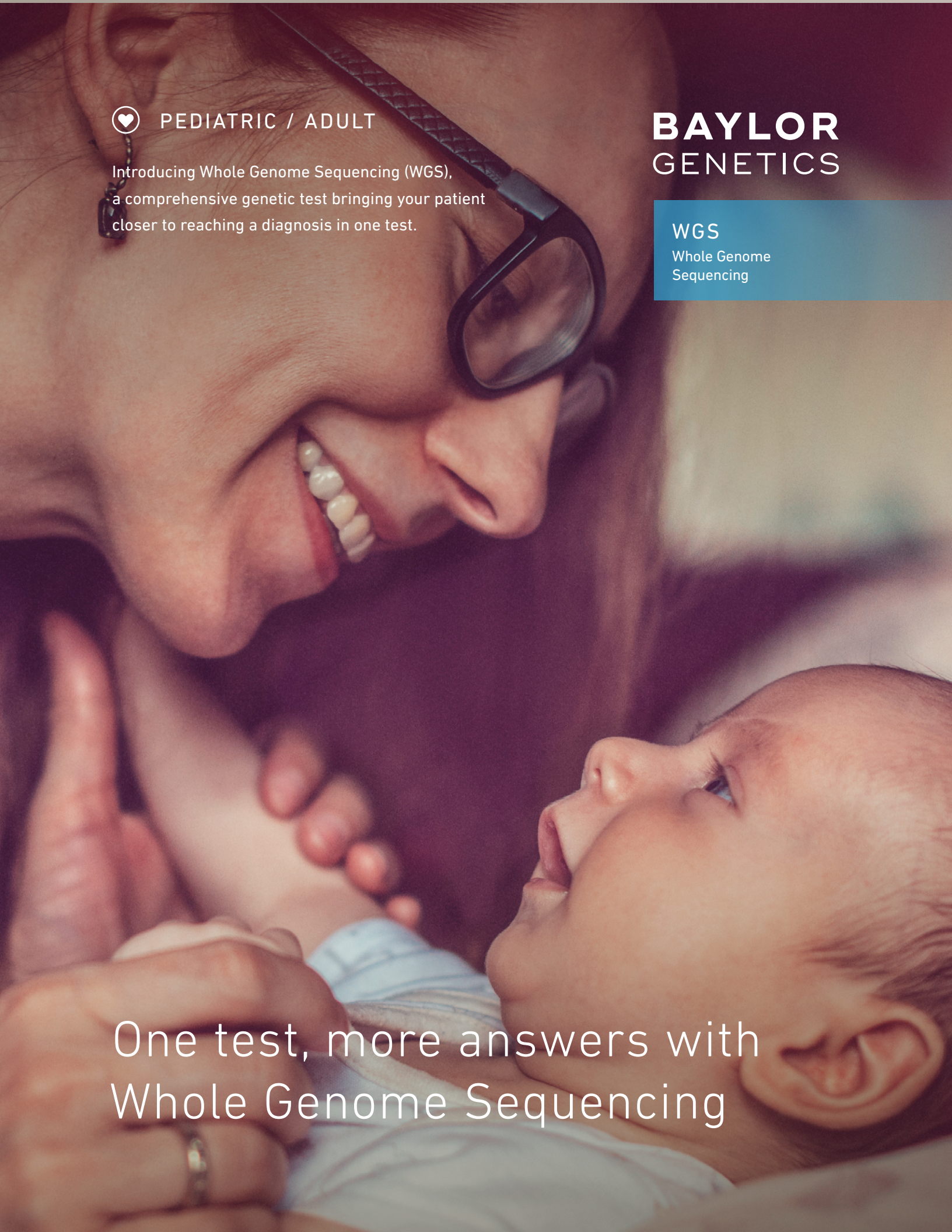


PEDIATRIC / ADULT

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

One test, more answers with Whole Genome Sequencing

Indications for Testing

PREVIOUS GENETIC TESTING UNINFORMATIVE

MULTIPLE CONGENITAL ANOMALIES

AUTISM SPECTRUM DISORDERS

NEURODEVELOPMENTAL DISORDERS

DEVELOPMENTAL DELAY

INTELLECTUAL DISABILITY

FAILURE TO THRIVE

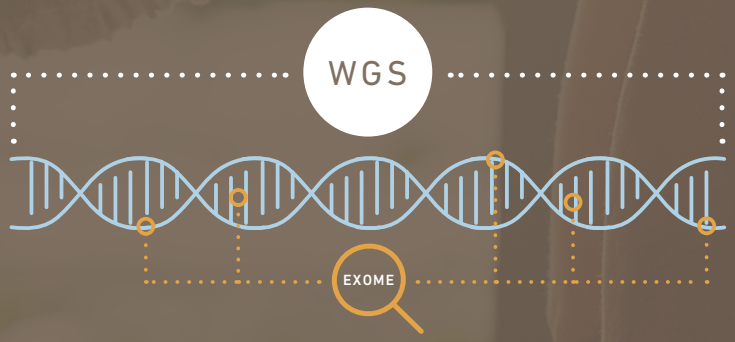
DYSMORPHIC FEATURES

EPILEPSY SYNDROMES

PATIENTS WITH AN EXTENSIVE DIFFERENTIAL DIAGNOSIS

Whole Genome Sequencing through Baylor Genetics is a comprehensive test that covers up to 98% of the whole human genome.

Whole Genome Sequencing (WGS) assesses the vast majority of the human genome and is a powerful tool for detecting known and potential disease-causing variations. Other genetic tests, such as whole exome sequencing and chromosomal microarray analysis, only detect specific types of disease-causing variants. WGS, however, is a comprehensive single test that can detect a broad range of variants that may not be identified on more targeted genetic testing.



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

WGS captures virtually all disease-causing genetic variations including single-nucleotide variants, small insertion/deletions, and copy number variants. Furthermore, WGS can also identify variants within the mitochondrial DNA as well as trinucleotide repeat expansions, adding additional utility compared to other tests.

WHEN TO ORDER WGS?

WGS should be considered for symptomatic patients lacking a sufficient personal or family history to suggest a specific genetic (or non-genetic) cause for intellectual disability, developmental delay, autism spectrum disorder, or multiple congenital anomalies, or if previous targeted testing or exome sequencing (WES) was uninformative.

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.

Comparison Chart

	RAPID TRIO WGS	TRIO WGS	RAPID DUO WGS	DUO WGS	RAPID PROBAND WGS	PROBAND WGS
Test Code	1822	1800	1823	1803	1829	1810
Consent	REQUIRED	REQUIRED	REQUIRED	REQUIRED	REQUIRED	REQUIRED
Parents Needed*	REQUIRED	REQUIRED	REQUIRED	REQUIRED	OPTIONAL	OPTIONAL
Parental Report Included*	✓	✓	✓	✓	✗	✗
TAT (weeks)	1 (starting at 5 days)	10	1 (starting at 5 days)	10	1 (starting at 5 days)	10
Can Elect to Receive Secondary Finding(s)	✓	✓	✓	✓	✓	✓
Raw Data Available	✓	✓	✓	✓	✓	✓

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

Testing Details

Gene Coverage

- All genes
- Single nucleotide variants/indels in coding and non-coding regions
- Copy number variants (CNV)
- Includes pathogenic mitochondrial variants
- Includes trinucleotide repeats
- Depth/Coverage: Average 40x genome-wide
- PCR-free: Better CNV
- 2x150bp Sequencing Length: Better CNV/TNR detection and mapping for complex genomic regions
- Bioinformatic analysis performed on the newest genome build, GRCh38

Methodology

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

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 include the following types of variants:

- Single nucleotide variants/indels (SNV)
- Copy number variants (CNV)
- Structural variants (SV)
- Absence of heterozygosity (AOH)
- Uniparental disomy (UPD)
- Short tandem repeats (STR) for 27 genes
- Mitochondrial genome variants

Negative Results

Negative results mean no relevant genetic change could be detected using WGS. 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

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 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 (WGS 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 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.

Specimen Requirements

TYPE	REQUIREMENTS	SHIPPING CONDITIONS
BLOOD	Draw blood in an EDTA (purple-top) tube(s) and send 3-5 cc for adults/children or 3 cc for infants < 2 years.	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.
BUCCAL SWAB	Collect with ORAcollect•Dx (OCD-100) self-collection kit (provided by Baylor Genetics with instructions). We highly recommend the sample be collected by a healthcare professional.	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
CORD BLOOD	Draw blood in an EDTA (purple-top) tube(s) and send 3-5 cc for adults/children or 3 cc for infants < 2 years. 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.
SALIVA	Saliva should be collected with an Orange DNA self-collection Kit.	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.

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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