BREAKTHROUGH
GENOMICS

Rare Disease
Diagnosis

Genetic Testing

Whole Exome & Whole Genome Tests

HIPAA COMPLIANT

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

CLIA Certified

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COMPREHENSIVE GENETIC ANALYSIS IN JUST ONE TEST

Through our best-in-class WGS and WES Genetic Testing, BREAKTHOUGH GENOMICS provides advanced AI-driven analysis to help diagnose more than 5600 rare genetic diseases. With fast turnaround times, high diagnostic yields, and decades of experience in the field of rare disease diagnosis, BREAKTHROUGH GENOMICS offers patients of all ages a precise diagnosis and a way end their diagnostic odyssey.

Whole Exome (WES) and Whole Genome (WGS) Tests Feature:

- Higher diagnostic yields than commercial labs
- Analysis of newly-discovered pathogenic variants
- Previously unrecognized clinical conditions
- Variants not covered in standard gene panels
- Personalized interpretation that leverages the most up-to-date scientific research and over 30 million publications
- High-throughput data sequencing technology with deep gene coverage

Scientists estimate that there are over 150 million people around the world that suffer from rare genetic diseases, with a vast majority being undiagnosed

Benefits to a successful diagnosis include:

- Improved disease management and outcomes
- Options for new therapeutic treatments
- Opportunity to participate in clinical trials
- Understanding of potential risks to family members
- Reduced expenses by avoiding a long diagnostic odyssey

BREAKTHROUGH GENOMICS

Rare Disease Diagnosis

70% of rare genetic diseases and conditions begin in childhood

Children whose rare genetic disease or condition goes undiagnosed can suffer from a number of serious consequences, including: misdiagnosis, incorrect interventions or the prescription of unnecesssary medications, and social and emotional isolation.
Whole Exome and Whole Genome Tests can help parents uncover the genetic underpinnings to many conditions that can be difficult to diagnose and can have a significant on their children.

Common conditions that can be diagnosed through genetic testing:

- Mental Disabilities
- Metabolic Disorders
- Development Delay
- Malformations
- Neurology
- Gastroenterology
- Cardiology

Rapid WES and WGS

BREAKTHROUGH GENOMICS's WGS & WES Tests can also be deployed in neonatal and pediatric intensive care units (NICU and PICU) with different levels of expedited service available upon request

What's included:

- ✓ Comprehensive Analysis requiring only a saliva sample
- ✓ All recommended ACMG Genes and incidental findings
- ✓ Secure Data Protection and Reporting
- ✓ High Quality Sequencing Data with coverage over 30X
- ✓ Leverage of proprietary AI-powered bioinformatic platforms to resolve Variants of Unknown Significance
- ✓ Up-to-date Literature Review
- ✓ Individualized Analysis of each case by U.S. Board-certified Medical Geneticist

A new study conducted by a children's clinician at BC Children's Hospital found that commercial labs missed over 10% of the correct diagnoses in WES cases.

BREAKTHROUGH GENOMICS' Comprehensive Whole Genome Test (WGS) includes additional coverage of rare conditions including:

- Copy Number Variations (CNV Analysis)
- Trinucleotide Repeat Expansion
- Spinal Dystrophy

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