Breakthrough Genomics helps expand access to precision pediatric care through its Rapid Whole Genome Sequencing Test

IRVINE, CALIFORNIA, USA, September 11, 2023 /EINPresswire.com/ -- Breakthrough Genomics, a leader in the clinical interpretation of genetic data, announces the launch of its new Rapid Whole Genome Sequencing Tests for diagnosing infants and children suffering from rare genetic disorders.

Traditional tests which include MRIs, spinal taps, and gene panel tests often miss the correct diagnosis leaving children and their families in distress while healthcare professionals grapple with the uncertainty of test results.

The Rapid Whole Genome Sequencing Test by Breakthrough Genomics provides doctors with a comprehensive view of a patient's genetic makeup and can pinpoint the genetic underpinnings of a child's condition within just a few days.

Leveraging AI-powered data interpretation technology, this test can rapidly analyze an individual's entire genome, enabling medical geneticists to successfully identify rare genetic conditions that may otherwise go undetected for an extended period. The test covers thousands of potential conditions within a wide range of medical specialties from neurological disorders like epilepsy to metabolic diseases, hormone imbalances, and congenital cardiovascular anomalies.

The company's whole genome test is now available in a number of pediatric care settings including Neonatal and Pediatric Intensive Care Units (NICUs and PICU's) and outpatient clinics.

"It's really incredible to hear the difference that these whole genome tests can make in a young person's life," says Scott Braman, the company's marketing director. He adds, "With the correct diagnosis in hand, doctors can determine the most appropriate treatment and families can feel confident that their child is on the right path."

This cutting-edge test with fast turnaround times is powered by Breakthrough Genomics' AI-powered interpretation platform Virtual GeneticistTM which is capable of analyzing a number of whole genome tests simultaneously. Virtual GeneticistTM employs advanced algorithms and machine learning models to quickly analyze vast amounts of genetic data to successfully zero in

on potential disease-causing variants. The platform not only expedites the diagnostic process but also enhances the accuracy of identifying rare diseases.

The company's founder and CEO, Dr. Laura Li, has deep expertise in rare disease diagnosis and sees the expanded access to Rapid Whole Genome Tests as one of the biggest breakthroughs in the field of pediatrics. "We are excited to offer our Rapid Whole Genome Test throughout the pediatric healthcare community as we know that it is truly a groundbreaking technology that has the ability to transform the lives of countless children and their families." - Dr. Laura Li Founder and CEO of Breakthrough Genomics.

For more information about Breakthrough Genomics' Rapid Whole Genome Sequencing Test, please visit BTGenomics.com

Media Contact:
Scott Braman
Director of Marketing
scott@BTGenomics.com

Laura Li
Breakthrough Genomics
+1 9492290094
email us here
