

Breakthrough Genomics Wins 2024 Innovation Award for AI-Driven Rare Disease Diagnosis and Early Cancer Detection

Breakthrough Genomics honored for AI advances in rare disease diagnosis & early cancer detection, transforming healthcare

IRVINE, CALIFORNIA, USA, May 20, 2024 /EINPresswire.com/ -- [Breakthrough Genomics](#) - a leader in the interpretation of genomic data - announced today that it has been awarded the 2024 Innovation Award at this year's California Investment Forum. The prize was given to recognize the company's development of novel AI-based techniques to improve the diagnosis of nearly 6000 rare diseases and expand the capabilities of its early cancer detection tests.

Since its genesis in 2017, Breakthrough Genomics has prioritized ongoing investments in AI and machine learning to continually improve its proprietary technology and best-in-class clinical tests. Along with its blood-based tests for the early detection of pancreatic and colorectal cancer, the company boasts two proprietary systems to analyze genomic data and quickly zero in on the genetic mutations that cause rare diseases, most often in children.

With millions of potential genetic variants to sort through in a short period of time, the need to leverage AI for the task was clear from the start.

"The field of human genetics poses unique challenges that go far beyond the capabilities of the typical language models deployed by ChatGPT and other AI-engines. For human genetics, it really takes a revolutionary approach to match the complexity of the field", said Breakthrough Genomics' CEO and Co-founder Dr. Laura Li (PhD, DABMGG).

In her talk at the Forum, Dr. Li highlighted the evolution of AI in precision medicine, starting with the use of the emerging technology in pathology labs tasked with detecting cancer cells in tissue samples. "From that initial image-based approach to the large language models we use today in our Virtual Geneticist TM platform, the effective adoption of AI has the clear potential to help improve and even save millions of lives," stressed Dr. Li.

But the challenge with AI is in the details, and the hard-won ability of companies like hers to constantly incorporate new AI capabilities into their existing models. That is where Breakthrough Genomics has excelled. In a 2023 study conducted at the University of British Columbia's Children's Hospital, the company's Virtual Geneticist TM platform was able to help diagnose an additional 10% of previously missed pediatric cases. This is critical, because without an accurate diagnosis, children and their parents often spend many agonizing years and tens of thousands of dollars trying to understand and treat their child's rare condition.

Another example of the company's commitment to push what is possible with AI is their [BT-Reveal](#) TM Early Pancreatic Cancer Test. The test was launched in the fall of 2023 as the first-of-its-kind test to help screen for pancreatic cancer - notorious for being extremely difficult to catch early enough to be treated. By leveraging the company's expertise in AI, Breakthrough Genomics was able to leverage a deep neural network model in their bioinformatic analysis of blood samples that improved the accuracy of the test and helped to reduce the number of false positive results.

And, just a few months ago, Breakthrough Genomics launched [Colon AiQ](#) TM - another ground-breaking test - this time a blood-based screening test for the early detection of colorectal cancer (CRC). The need for Colon AiQ TM is also clear as many eligible adults in the U.S. are not up-to-date on their CRC screening and are hesitant to follow through with colonoscopies and stool-based tests.

"It's amazing to see what Breakthrough Genomics has already been able to do to help fight two of the worst cancers affecting our communities." Says Fiona Ma - California State Treasurer and a strong supporter of precision medicine initiatives. She adds, "I can't wait to see what's next for Breakthrough Genomics as it helps to make California a leader in this important area."

Today, the company's BT-Reveal TM test can detect 83% of pancreatic cancers including cancers in Stages 1 and 2 and returns results with only 5% chance of being a false positive. "Many doctors still don't know that they can now screen high-risk patients for pancreatic cancer. AI is truly helping to make what was once considered impossible a reality," says Scott Braman, Breakthrough Genomics' Director of Marketing and Partnerships.

The recognition of Breakthrough Genomics with the 2024 CIF Innovation Award for "Breakthrough Medical Technology in Diagnostic"; marks a significant milestone in the integration of AI within the field of medical diagnostics. This accolade reflects the company's commitment to advancing healthcare through innovative solutions.

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