



NEWS RELEASE

Athena Diagnostics Improves Ataxia Movement Disorder Testing with Novel Long-Read Gene Sequencing

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New test utilizes technology based on PacBio's PureTarget sequencing method to uncover genetic causes of disease

BOSTON, July 29, 2025 /PRNewswire/ -- Athena Diagnostics, the specialty neurology business of Quest Diagnostics (NYSE: DGX), a leading provider of diagnostic information services, today announced a new laboratory test service intended to provide insights into genetic causes of ataxia, a group of progressive, degenerative neurological disorders that affect movement or coordination.

The new service is a laboratory developed test that identifies sequences of DNA that repeat many times across a long stretch of the genome. Conventional sequencing technologies are adept at elucidating short DNA sequences but may fail to discern complex or large repeating sequences. Genetic causes of ataxia include expansions of repeating sequences in various genes, which go undetected in many conventional genetic sequencing panels. The new service will be used to confirm initial screening results delivered using polymerase chain reaction (PCR) testing. Healthcare providers may now order the test from Athena Diagnostics.

Under a collaboration agreement with PacBio (NASDAQ: PACB), Quest developed and validated the new lab developed test using specialized technology based on PacBio's PureTarget method. Athena used the technology to develop and validate its ataxia test in Athena's advanced laboratory in Marlborough, Mass. Athena is also exploring options to develop and deploy a test based on PacBio technology to improve carrier screening.

In a study available as a preprint, a PureTarget gene panel was found to identify a specific repeating gene pattern associated with Spinocerebellar ataxia type 10 that may be associated with age of disease onset, information that

may help guide care decisions.

"Extensive iterative testing is sometimes needed to identify the genes implicated in neurological disease like ataxia," said Mark Gardner, Senior Vice President of Oncology, Genomics and R&D at Quest Diagnostics. "Interrogating long DNA strands may identify patterns that other methods may miss, improving confirmatory testing quality."

"Athena Diagnostics is a well-regarded leader in neurological and rare disease testing, and we are excited about the potential of this new Athena Diagnostics test, empowered with our expertise in long-read sequencing, so more families get the answers they need," said Christian Henry, President and Chief Executive Officer of PacBio.

Ataxia is a degenerative disease of the nervous system, wherein affected individuals may experience lack of coordination using muscles in their fingers and hands, arms, legs, walking, speaking or moving their eyes, resulting from dysfunction of brain areas. Recent studies have found the prevalence of ataxia in children to be 26 per 100,000, with 10 per 100,000 being caused by genetics.

About Athena Diagnostics

Athena Diagnostics, a business of Quest Diagnostics, is a leader in diagnostic testing for neurological diseases and offers innovative tests for Alzheimer's disease, muscular dystrophy and other neuromuscular and developmental disorders. Athena is dedicated to providing neurologists and other physicians and specialists with insights that can improve patient health. For more information, visit [AthenaDiagnostics.com](https://www.AthenaDiagnostics.com).

About PacBio

PacBio (NASDAQ: PACB) is a premier life science technology company that is designing, developing and manufacturing advanced sequencing solutions to help scientists and clinical researchers resolve genetically complex problems. Our products and technologies stem from two highly differentiated core technologies focused on accuracy, quality and completeness which include our HiFi long-read sequencing and our SBB® short-read sequencing technologies. Our products address solutions across a broad set of research applications including human germline sequencing, plant and animal sciences, infectious disease and microbiology, and oncology. For more information, please visit www.pacb.com and follow @PacBio.

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