



Bionano Genomics Announces First Publication Using OGM for Detection of Repeat Expansions in CANVAS and Adult-Onset Ataxia

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SAN DIEGO, Aug. 11, 2022 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO) today announced the publication of the first study to evaluate the utility of optical genome mapping (OGM) in the analysis of repeat expansion disorders. Repeat expansion disorders are a class of disorders that impact approximately 1 in 3,000 people and are caused by expansions of short tandem DNA repeats.

Researchers in this study describe how OGM can be adopted as an alternative to Southern blot analysis for the identification of repeat expansions in the *RFC1* gene that lead to cerebellar ataxia with neuropathy and bilateral vestibular areflexia syndrome (CANVAS) and adult-onset ataxia in multiple populations. Individuals with *RFC1* (AAGGG)_n repeat expansion disorder may present with a spectrum of features including cerebellar ataxia, neuropathy, and vestibular areflexia. Specifically, ataxias are a group of neurodegenerative repeat expansion disorders, characterized by atrophy of the cerebellum that leads to the inability to control balance and coordination. Even though there are many known cerebellar ataxia-causing genes and variations, some adult patients with adult-onset ataxia remain genetically undiagnosed, and studies to identify genetic causes are ongoing.

In the study, researchers screened 626 samples for the presence of (AAGGG)_n repeat expansions in the *RFC1* gene using a combination of PCR to detect repeat sequences, and OGM, to determine the size of the repeat expansions. OGM confirmed the presence of expanded *RFC1* alleles in all cases where it was used as part of the workflow. Repeats identified using OGM were in the range of 800–1299 repeat units. OGM is well-suited for analyzing large repeat expansion disorders that require the measurement of long, intact DNA molecules for accurate sizing, as was observed in this study. The study noted the successful use of OGM to replace the labor intensive and time-consuming Southern blot, which has been the “gold” standard for sizing repeat expansions up to now.

“We believe this study helps demonstrate the important role OGM can play in understanding the most complex regions of the genome and that OGM is well-suited for the analysis of repeat expansion disorders that require the measurement of long, intact DNA molecules for accurate sizing. We plan to continue developing better tools for the detection of a range of repeat expansion disorders, including those causing adult-onset ataxia and CANVAS. We are optimistic that adding OGM to repeat expansion disorder research can potentially lead to better diagnosis for patients and more accurate data for neuroscience researchers,” commented Erik Holmlin, PhD, president and chief executive officer of Bionano Genomics.

This publication is available at: <https://link.springer.com/article/10.1007/s00415-022-11275-9>

About Bionano Genomics

Bionano Genomics is a provider of genome analysis solutions that can enable researchers and clinicians to reveal answers to challenging questions in biology and medicine. The Company's mission is to transform the way the world sees the genome through OGM solutions, diagnostic services and software. The Company offers OGM solutions for applications across basic, translational and clinical research. Through its Lineagen business, the Company also provides diagnostic testing for patients with clinical presentations consistent with autism spectrum disorder and other neurodevelopmental disabilities. Through its BioDiscovery business, the Company also offers an industry-leading, platform-agnostic software solution, which integrates next-generation sequencing and microarray data designed to provide analysis, visualization, interpretation and reporting of copy number variants, single-nucleotide variants and absence of heterozygosity across the genome in one consolidated view. For more information, visit www.bionanogenomics.com, www.lineagen.com or www.biodiscovery.com

Forward-Looking Statements of Bionano Genomics

This press release contains forward-looking statements within the meaning of the Private Securities Litigation Reform Act of 1995. Words such as “believe,” “can,” “optimistic,” “plan,” “potential,” and similar expressions (as well as other words or expressions referencing future events, conditions or circumstances) convey uncertainty of future events or outcomes and are intended to identify these forward-looking statements. Forward-looking statements include statements regarding our intentions, beliefs, projections, outlook, analyses or current expectations concerning, among other things, the ability and utility of OGM to complement whole genome analysis in the detection of repeat expansions associated with CANVAS and adult-onset ataxia. Each of these forward-looking statements involves risks and uncertainties. Actual results or developments may differ materially from those projected or implied in these forward-looking statements. Factors that may cause such a difference include the risks and uncertainties associated with: the impact of the COVID-19 pandemic on our business and the global economy; general market conditions; changes in the competitive landscape and the introduction of competitive technologies or improvements to existing technologies; failure OGM to detect repeat expansions associated with CANVAS and adult-onset ataxia; changes in our strategic and commercial plans; our ability to obtain sufficient financing to fund our strategic plans and commercialization efforts; the ability of medical and research institutions to obtain funding to support adoption or continued use of our technologies; failure of future study results to support those demonstrated during the study referenced in this press release; and the risks and uncertainties associated with our business and financial condition in general, including the risks and uncertainties described in our filings with the Securities and Exchange Commission, including, without limitation, our Annual Report on Form 10-K for the year ended December 31, 2021 and in other filings subsequently made by us with the Securities and Exchange Commission. All forward-looking statements contained in this press release speak only as of the date on which they were made and are based on management's assumptions and estimates as of such date. We do not undertake any obligation to publicly update any forward-looking statements, whether as a result of the receipt of new information, the occurrence of future events or otherwise.

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