# Bionano Announces Publication Showing OGM can Provide an Accurate and Cost-Effective Solution for Sizing Large Repeat Expansions in Constitutional Genetic Disease

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SAN DIEGO, April 23, 2025 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO) today announced a peer-reviewed publication in *Genome Research* demonstrating that optical genome Mapping (OGM) can be an accurate, cost-effective method for detecting and sizing large repeat expansions, which are a class of structural variation (SV) linked to as many as 40 genetic disorders. Led by Alexander Hoischen and researchers at Radboud University Medical Center in the Netherlands, the study provides one of the most comprehensive evaluations to date of OGM as a single workflow for characterizing large repeat expansions. Researchers evaluated 85 subjects containing known pathogenic expansions in three genes, *DMPK* (associated with myotonic dystrophy (DM) type 1), *CNBP* (associated with DM type 2), and *RFC1* (associated with cerebellar ataxia, neuropathy, and vestibular areflexia syndrome (CANVAS)). DM is a genetic condition causing progressive muscle weakness and wasting, affecting both skeletal and smooth muscles and CANVAS is a genetic condition characterized by progressive loss of coordination, sensory nerve damage, and impaired balance reflexes.

**Key Findings:**

- **High detection accuracy:** OGM identified 84 out of 85 (98.8%) pathogenic repeat expansions.
- **No apparent upper size limit:** OGM measured accurately the repeat sizes, even for extremely long expansions exceeding 7,000 repeat units.
- **Somatic instability revealed:** Evidence of somatic instability in 36 out of 85 samples, with variability observed in 30% of *DMPK* samples, 92% of *CNBP* samples, and 16% of *RFC1* samples. These findings provide new insights into the unpredictable nature of these disorders.
- **Single technique workflow:** Unlike traditional methods that require multiple gene-specific assays, OGM delivers a single, comprehensive assay that detects large repeat expansions across multiple loci, potentially reducing turnaround times and lowering costs.

This publication appears alongside two other OGM studies in Part II of a series released by *Genome Research* focusing on long-read methods such as OGM and long-read sequencing (LRS) for genome analysis. In an accompanying editorial, the editors of the series describe how OGM has the potential to address long-standing challenges in genome analysis, from structural variation to paralog resolution and repeat expansion detection, sizing and evaluation of their somatic instability.

“Repeat expansions are a clinically relevant class of structural variations (SVs). Their size, stability and variability make them challenging to analyze using conventional methods such as PCR, Southern blotting and next-generation sequencing (NGS). LRS may be useful, but LRS can be too costly for routine use,” commented Erik Holmlin, president and CEO of Bionano. “This study demonstrates how can OGM address a critical limitation in genome analysis and lays the foundation for researchers around the world to broaden their scope of OGM use in research to include this class of SVs, which we believe may pave the way for future development of better diagnostics and therapeutics for genetic diseases like repeat expansion disorders.”

The full research publication is available at: https://genome.cshlp.org/content/early/2025/03/19/gr.279491.124.full.pdf+html

The editorial accompanying the special issue of *Genome Research* is available at: https://genome.cshlp.org/content/35/4/xi.full.pdf+html

**About Bionano**

Bionano 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 optical genome mapping (OGM) solutions, diagnostic services and software. The Company offers OGM solutions for applications across basic, translational and clinical research. The Company also offers an industry-leading, platform-agnostic genome analysis software solution, and nucleic acid extraction and purification solutions using proprietary isotachophoresis (ITP) technology. Through its Lineagen, Inc. d/b/a Bionano Laboratories business, the Company also offers OGM-based diagnostic testing services.

For more information, visit www.bionano.com or www.bionanolaboratories.com.

Bionano’s products are for research use only and not for use in diagnostic procedures.

**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,” “may,” “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 describe future expectations, plans, results, or strategies, among other things, and in this release include, but are not limited to, statements OGM’s ability to be an accurate, cost-effective method for detecting and sizing large repeat expansions; OGM’s ability to identify pathogenic repeat expansions; OGM’s ability to measure accurately expansion sizes, even for extremely long expansions; OGM’s ability to show evidence of somatic

instability; OGM's ability to deliver a single, comprehensive assay that detects large repeat expansions across multiple loci, reducing turnaround times and lowering costs; the ability of OGM to address critical limitations in genomics analysis; the ability of OGM to pave the way for future development of better diagnostics and therapeutics for genetic diseases like repeat expansion disorders; and other statements not of historical fact. Such statements are subject to a multitude of risks and uncertainties that could cause future circumstances, events, or results to differ materially from those projected in the forward-looking statements. 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 impact of adverse geopolitical and macroeconomic events, such as bank failures, the ongoing conflicts between Ukraine and Russia and in the Middle East and related sanctions, regional or global pandemics, uncertain market conditions, including tariffs and inflation, and supply chain disruptions on our business and the global economy; the failure of OGM to be an accurate, cost-effective method for detecting and sizing large repeat expansions; the failure of OGM to identify pathogenic repeat expansions; the failure of OGM to measure accurately expansion sizes, even for extremely long expansions; the failure of OGM to show evidence of somatic instability; the failure of OGM to deliver a single, comprehensive assay that detects large repeat expansions across multiple loci, reducing turnaround times and lowering costs; the failure of OGM to address critical limitations in genomics analysis; the failure of OGM to pave the way for future development of better diagnostics and therapeutics for genetic diseases like repeat expansion disorders; the inability to achieve results similar to those referenced in this press release using the methods described therein; the failure of the publication referenced in this press release to prove to be an important advancement in repeat expansion research; the failure of the method described in this press release to further open researchers' ability to expand their OGM menu; future publications that contradict the findings of the publication referenced in this press release; the failure of our ability to drive adoption and utilization of optical genome mapping as a replacement to traditional cytogenetic techniques; challenges inherent in developing, manufacturing and commercializing products; our ability to further deploy new products and applications for our technology platforms; our expectations and beliefs regarding future growth of the business and the markets in which we operate; changes in our strategic and commercial plans; our ability to continue as a "going concern," which requires us to manage costs and obtain significant additional financing to fund our strategic plans and commercialization efforts; our ability to consummate any strategic alternatives; the risk that if we fail to obtain additional financing we may seek relief under applicable insolvency laws; and other risks and uncertainties including those described in our filings with the Securities and Exchange Commission ("SEC"), including, without limitation, our Annual Report on Form 10-K for the year ended December 31, 2024 and in other filings subsequently made by us with the SEC. All forward-looking statements contained in this report 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 are under no duty to update any of these forward-looking statements after the date they are made to conform these statements to actual results or revised expectations, except as required by law. You should, therefore, not rely on these forward-looking statements as representing our views as of any date subsequent to the date the statements are made. Moreover, except as required by law, neither we nor any other person assumes responsibility for the accuracy and completeness of the forward-looking statements contained in this press release.

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