Bionano Announces a 56% Increase in Publications Describing the Utility of OGM in Rare Diseases in Q1 2026 vs Q1 2025

May 5, 2026

Overall, studies show:

- *OGM Can Resolve Previously Unresolved Rare Disease Cases*
- *OGM Can Serve as a Gold Standard Technique for Characterization of SVs*
- *OGM Can Complement Sequencing for Better SV Sensitivity and Genomic Insights*
- *OGM Can Identify Actionable Disrupted Genes Across Rare Diseases*

SAN DIEGO, May 05, 2026 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO) reported that 28 publications describing the utility of optical genome mapping (OGM) for analysis of rare diseases were released in Q1 2026, representing an approximately 56% increase over last year. The total number of samples analyzed, 78, represents a 225% increase compared to the number analyzed in studies published in Q1 2025 (24). Publications describing OGM use in rare disease research come from institutions all around the world, including from Europe, Asia, South America, and the United States.

These studies describe the ability of OGM to enable clinical researchers to identify, characterize and interpret relevant structural variants (SVs) often missed by traditional cytogenetic methods such as karyotyping, fluorescence *in-situ* hybridization (FISH) and chromosomal microarrays (CMA) as well as next-generation sequencing (NGS) and long-read sequencing (LRS). They encompass a broad range of conditions, including neurodevelopmental, neuromuscular, neurodegenerative, immunological, and malformation syndromes.

“Rare disease is not just a scientific challenge – it is a deeply personal challenge for millions of patients and families searching for answers, often for years without clarity. With an estimated 1 in 15 people worldwide affected and ~70% of cases having a genetic origin, the unmet need for better research tools like OGM is profound. For too long, these studies have remained out of reach due to limitations of conventional technologies. OGM is changing that reality by breaking through these barriers, revealing hidden genomic complexity, and providing more clarity in cases once considered unsolvable. Building on its impact in hematological malignancies, OGM is now helping bring much-needed clarity to the rare disease research community as an alternative to traditional cytogenic analysis and as a strong complement to NGS and LRS,” said Erik Holmlin, Ph.D., president and chief executive officer of Bionano.

Key takeaways from 14 of the 28 publications are summarized in key categories below, all highlighting the unique value of OGM and its complementarity with sequencing.

OGM Resolves Previously Unresolved Rare Disease Cases

- OGM continues to demonstrate value in rare disease cases, where approximately 60% remain genetically unexplained after analysis by standard methods. Two studies in multisite European (n=57 trios), and USA–Taiwan (n=29) cohorts show that OGM increases yield by 5–17% after prior negative whole genome sequencing (WGS), and in some cases LRS. OGM identified previously missed SVs across a wide range of variant types, including balanced and unbalanced SVs, and complex rearrangements, some of which were more difficult to detect with only LRS data due to genomic complexity. Together, these studies underscore how OGM can be a powerful tool for helping to solve unresolved cases and uncover cryptic genomic variations that other methods miss.

OGM can Serve as a Gold Standard Technique for Characterization of SVs

- A study from Japan analyzed 30 cases with copy number variations (CNVs) and SVs that had been initially characterized by sequencing, and showed that OGM, alongside targeted LRS, improved the characterization of these SVs in 46% of cases. In 23% of the cases, including some in which LRS was constrained by read length, OGM findings unraveled significant novel gene-disrupting events including complex rearrangements and repetitive regions.

- Two studies from Brazil demonstrate the use of OGM to precisely identify the breakpoints of inversions affecting the *SOX3* and *SYT1* genes, both involved in malformation syndromes. By pinpointing these SVs, OGM was pivotal in linking the genetic alterations to dysregulation of these genes in gonadal tissue and in neurons, thereby providing a genetic link to the observed phenotypes.

- A paper outlining international guidelines with authors from the US, Latin America, Europe and Asia, recommends use of OGM to confirm structural variants initially identified by NGS.

OGM Complements Sequencing for Better SV Sensitivity and Genomic Insights

- A German study shows that OGM resolved a case by identifying a homozygous deletion in *KIF1C* that was missed after both whole-exome sequencing (WES) and WGS. A study from Turkey and Austria demonstrates that combining OGM with NGS enabled reclassification of an apparent heterozygous point mutation to a compound heterozygous event with an SV affecting the *TMC6/TMC8* tumor suppressor genes that was seen with OGM.

Other studies show that while SV detection by LRS has improved, OGM is useful for detecting larger SVs and helps to reduce false positives associate with LRS.

- In a study of Parkinson's disease in Germany and the US, OGM identified more variants than LRS with better accuracy, particularly in the 50–80 kbp range.

- A study from Spain and the Netherlands used OGM alongside LRS to study two families with angioedema, including a 20-year long genetic odyssey. OGM identified a previously missed insertion that was later confirmed to be an immobile element linked to the disorder.

- A study between Sweden and the US demonstrated that OGM enables complete reconstruction of a complex derivative X chromosome that could not be fully resolved by LRS.

OGM Enables Identification of Actionable Disrupted Genes Across Rare Diseases

Understanding precise breakpoints is critical across rare diseases, as it enables accurate molecular evaluation, informs genetic counseling, and has important implications for emerging gene-editing–based therapeutic strategies. Duchenne Muscular Dystrophy (DMD) is a prime example where accurate characterization of exon–intron disruptions is essential, as many therapies are designed for specific variant classes and therefore depend on precise genetic definition. OGM has been demonstrated to be a powerful tool in DMD, as illustrated by several studies.

- A collaborative study between Medical College of Wisconsin and Children's Wisconsin and University of Illinois-Chicago in the US, reported a case with persistent negative findings was resolved using OGM and LRS, which identified a novel inversion in the *DMD* gene.

- A study from China described how OGM enabled precise assessment of *DMD* exon duplications by resolving their genomic structure and insertion sites. OGM findings clarified a previously unexplained case with a dual phenotype involving both DMD and spinal muscular atrophy (SMA), where the SMA-associated mutation had been identified, but the DMD component had remained unresolved prior to OGM.

- A study by Cincinnati Children's Hospital Medical Center from USA on three samples with intragenic DMD duplications showed that OGM revised classification. In two cases, duplications were found outside the gene and reclassified as likely benign, while in one case a tandem duplication within the gene was classified as pathogenic, directly impacting management decisions.

These findings in DMD are consistent with observations across other rare diseases, where OGM similarly improves the detection and clinical research interpretation of complex SVs.

The following is the list of publications in the order presented in this release:

	Site and Country	Link
1	Multisite European study from **Finland, France, Germany and Netherlands**	https://doi.org/10.64898/2026.01.16.26344264
2	University of California from **USA** and Multisite from **Taiwan**	https://doi.org/10.1093/hmg/ddaf204
3	Multisite study from **Japan**	https://doi.org/10.1038/s41525-026-00561-4
4	Pacific Northwest Research Institute from **USA** and Multisite from **Brazil** – SOX3	https://doi.org/10.1186/s13293-025-00822-4
5	University of Sao Paulo from **Brazil** – SYT1	https://doi.org/10.1155/crig/6652420
6	Multisite International guideline study **USA, Latin America, Europe and Asia**	https://doi.org/10.1093/ejendo/lvaq013
7	University of Lübeck from **Germany** – KIF1C	https://doi.org/10.1007/s12311-026-01963-x
8	Hacettepe University Cancer Institute and Institute of Medical Genetics from **Turkey & Austria**	https://doi.org/10.1038/s41431-026-02043-8
9	Multisite study from **Germany and USA**	https://doi.org/10.1002/acn3.70332
10	Multisite from **Spain** and Radboud UMC from **Netherlands**	https://doi.org/10.1007/s10875-026-02015-z

11	Karolinska University Hospital, Baylor College of Medicine and Pacific Northwest Research Institute from **Sweden and USA**	https://doi.org/10.1101/gr.281175.125
12	Medical College of Wisconsin and Children's Wisconsin and University of Illinois-Chicago from **USA**	https://doi.org/10.1002/ajmg.a.70133
13	The Affiliated Women and Children's Hospital of Ningbo University from **China**	https://doi.org/10.1186/s13039-026-00751-w
14	Cincinnati Children's Hospital Medical Center from **USA**	https://doi.org/10.1016/j.nmd.2026.106335

About Bionano Genomics

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. All statements other than statements of historical facts contained in this press release, including statements regarding our future results of operations or financial condition, business strategy and plans, and objectives of management for future operations, are forward-looking statements. Words such as "anticipate," "believe," "can," "could," "estimate," "expect," "intend," "may," "plan," "potential," "predict," "project," "should," "target," "will," or "would" 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 substantially improve detection of relevant cytogenomic abnormalities in rare diseases; the ability and utility of OGM to be a useful tool to help solve unresolved cases and uncover cryptic genomic variations missed by other methods; the ability and utility of OGM to resolve previously unresolved rare disease cases; the ability and utility of OGM to serve as a gold standard technique for characterization of SVs; the ability and utility of OGM to completement sequencing for better SV sensitivity and genomic insights; the ability and utility of OGM to identify actionable disrupted genes across rare diseases; the ability and utility of OGM to provide comprehensive, genome-wide identification of structural variants, copy number alterations, cryptic rearrangements, and noncoding events; the ability and utility of OGM to provide critical genomic information that complements targeted NGS and may help advance future research in rare disease research; the ability of OGM to outperform legacy cytogenomic methods; continued research, presentations and publications involving OGM, its utility compared to traditional cytogenetics and our technologies; our ability to drive adoption of OGM and our technology solutions; and any other statements that are not of historical fact. Each of these forward-looking statements involves risks and uncertainties. Accordingly, investors and prospective investors are cautioned not to place undue reliance on these forward-looking statements as they involve inherent risk and uncertainty (both general and specific) and should note that they are provided as a general guide only and should not be relied on as an indication or guarantee of future performance. 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 failure of OGM to substantially improve detection of relevant cytogenomic abnormalities in rare diseases; the failure of OGM to be a useful tool to help solve unresolved cases and uncover cryptic genomic variations missed by other methods; the failure of OGM to resolve previously unresolved rare disease cases; the failure of OGM to serve as a gold standard technique for characterization of SVs; the failure of OGM to completement sequencing for better SV sensitivity and genomic insights; the failure of OGM to identify actionable disrupted genes across rare diseases; the failure of OGM to provide comprehensive, genome-wide identification of structural variants, copy number alterations, cryptic rearrangements, and noncoding events; the failure of OGM to provide critical genomic information that complements targeted NGS and may help advance future research in rare diseases; the failure of OGM to outperform legacy cytogenomic methods; our ability to obtain sufficient financing to fund our strategic plans and commercialization efforts and 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; the risk that if we fail to obtain additional financing we may seek relief under applicable insolvency laws; the impact of adverse geopolitical and macroeconomic events, such as the ongoing international conflicts and uncertain market conditions, including inflation, tariffs, and supply chain disruptions, 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; changes in our strategic and commercial plans; the ability of medical and research institutions to obtain funding to support adoption or continued use of our technologies; study results that differ or contradict the results mentioned in this press release; and the risks and uncertainties associated with our business and financial condition in general, including the 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, 2025, our Quarterly Reports on Form 10-Q and in other filings subsequently made by us with the SEC. 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, except as may be required by law.

CONTACTS

Company Contact:
Erik Holmlin, CEO
Bionano Genomics, Inc.
+1 (858) 888-7610
eholmlin@bionano.com

Investor Relations:
Webb Campbell

Gilmartin Group
+1 (858) 888-7625

Source: Bionano Genomics