# Bionano Announces Two Publications from its Clinical Study Designed to Support OGM as Part of Standard of Care (SOC) in Genetic Disease Testing including the First Peer-Reviewed Publication on the Multi-Site Evaluation of OGM Against SOC

March 2, 2023

- Peer-reviewed publication from Iqbal, *et al.* describes the multi-site evaluation of optical genome mapping (OGM) for postnatal genetic disorders with 404 samples and shows:
    - Concordance of OGM against various standard of care (SOC) methods –99.5% [399 out of 401 samples]
    - First-pass success rate for OGM – 90.2% [369 out of 409 samples]
    - Overall success rate – 98.8% [404 out of 409 samples]
    - Inter- and intrasite repeatability –100%
- Pre-print publication from Broeckel, *et al.* describes the multi-site evaluation of OGM in an additional 560 unique samples, with a total of 749 unique samples to-date from 1,037 datapoints and shows:
    - Concordance for all combined samples against SOC methods – 99.6% [746 out of 749 samples]
    - For a subset of 79 prospectively collected samples from patients suspected of a genetic disorder, SOC had reportable findings in 19 cases [24%]; OGM had reportable findings in 27 cases [34%], corresponding to a 42% increase in the number of cases with reportable findings when OGM was used
    - For another subset of cases consisting of 135 retrospectively collected samples from patients suspected of autism spectrum disorders (ASD), SOC found reportable variants in 63 samples [46%]; OGM identified reportable variants in 83 samples [61%] corresponding to a 32% increase in the number of cases with reportable findings when OGM was used

SAN DIEGO, March 02, 2023 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (BNGO), today announced two publications detailing results from the clinical trial designed to support establishing optical genome mapping (OGM) as part of the standard of care (SOC) in diagnosis of genetic disease for postnatal patients. The clinical trial is designed to compare OGM to SOC, including concordance, reproducibility, technical success rate and the rate of detecting reportable findings in cases. A peer-reviewed publication covered an interim readout of the study, which showed OGM's high technical performance and reproducibility across sites versus SOC analysis. The preprint publication extended the study to additional patients and measured the rate of detecting reportable findings by OGM compared to that of SOC methods in analysis of samples from individuals with neurodevelopmental disorders, including developmental delay, intellectual disability and autism spectrum disorder (ASD).

The sites conducting the study and their principal investigators are as follows:

- University of Rochester Medical Center (Dr. M. Anwar Iqbal)
- Medical College of Wisconsin (Dr. Ulrich Broeckel)
- Columbia University Medical Center (Dr. Brynn Levy)
- Greenwood Genetic Center (Dr. Roger Stevenson)
- Medical College of Georgia, Augusta University (Dr. Ravindra Kolhe)
- Praxis Genomics (Dr. Peter L. Nagy)
- University of Iowa Health Clinics (Dr. Aaron Stence)
- H. Lee Moffitt Cancer Center (Dr. Aaron Bossler)

**Key Findings**

The peer-reviewed publication describes OGM performance metrics like first pass success rate and reproducibility from site-to-site, operator-to-operator and run-to-run for the first time ever and for the largest number of samples investigated with OGM to date.

Key findings were reported as follows:

- Unblinded concordance with standard of care (SOC) – 99.5% [399 out of 401 samples]
- Partially concordant with SOC – 0.5% [2 out of 401 samples]
- Blinded concordance with SOC – 97.6% [364 out of 373 samples]
- First-pass success rate for OGM – 90.2% [369 out of 409 samples]
- Overall success rate – 98.8% [404 out of 409 samples]
- 100% agreement on Fragile X syndrome (FXS) calls for "full expansion or not full expansion" in 401 samples

The pre-print publication describes OGM performance metrics compared to SOC methods for challenging samples from diagnosed and undiagnosed

rare diseases.

Key findings were reported as follows:

- Concordance for all combined samples against SOC methods – 99.6% [746 out of 749 samples]
- For a subset of 79 prospectively collected samples from patients suspected of a genetic disorder, SOC had reportable findings in 19 cases [24%]; OGM had reportable findings in 27 cases [34%], corresponding to a 42% increase in the number of cases with reportable findings when OGM was used
- For another subset of cases consisting of 135 retrospectively collected samples from patients suspected of autism spectrum disorders (ASD), SOC found reportable variants in 63 samples [46%]; OGM identified reportable variants in 83 samples [61%] corresponding to a 32% increase in the number of cases with reportable findings when OGM was used

**Key Takeaways**

Authors of the peer-reviewed publication concluded that study results demonstrate the high technical performance of the OGM workflow for postnatal samples. The authors reported that intersite, interrun, and intrarun performance demonstrates the reproducibility of the OGM workflow, suggesting the potential for easy adoption and validation. The authors further underscored that OGM is not limited to copy number variation (CNV) analysis alone, but can also resolve balanced structural rearrangements, size repeat expansions like *FMR1* and repeat contractions like D4Z4 and noted that OGM identified additional variants that were undetected by SOC. In summary, the authors concluded that a single approach, like OGM, can allow genetic laboratories to provide rapid results with a cost-effective solution.

The pre-print publication results demonstrate the potential of an OGM workflow to detect all classes of SVs with higher resolution, including aneuploidies, triploidy, translocations, inversions, insertions, microdeletions, microduplications, nucleotide repeat expansions or contractions, and absence of heterozygosity (AOH). In contrast to variants of uncertain significance (VUSs) that are detected by microarray, which are limited to gains and losses, the VUSs reported by OGM include multiple types of SVs, several of which reside in candidate genes associated with the phenotype. The authors concluded that OGM can offer a simple and streamlined workflow that can detect relevant genomic aberrations and mitigate the need for numerous testing platforms and time-consuming wet lab work, potentially improving lab performance by reducing the associated time and costs.

“Development and validation of OGM assays for postnatal analysis is an area where we believe our technology can have tremendous global impact. The performance we have seen matches our expectations. We are extremely happy with these publications demonstrating OGM’s performance across multiple sites and its potential ability to perform in a single assay what today requires multiple technologies,” commented Erik Holmlin, PhD, president and chief executive officer of Bionano.

“The process of establishing a trial program with a consortium like this one is made possible by capable principal investigators and leading sites,” commented Alka Chaubey, PhD, FACMG, chief medical officer of Bionano. “We are particularly pleased with the study findings related to the potential utility of OGM to increase reportable yield in autism spectrum disorder samples.”

The peer-reviewed publication is available at https://www.sciencedirect.com/science/article/pii/S1525157823000028; the pre-print publication is available at https://www.medrxiv.org/content/10.1101/2022.12.26.22283900v1.

**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, Inc. d/b/a Bionano Laboratories 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.bionano.com, www.bionanolaboratories.com or www.biodiscovery.com

This press release contains forward-looking statements contains forward-looking statements within the meaning of the Private Securities Litigation Reform Act of 1995. Words such as “can,” “believe,” “potential,” and similar expressions (as well as other words or expressions referencing future events, conditions or circumstances and the negatives thereof) 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 performance of OGM compared to existing technologies including karyotyping, FISH and CMA for the identification of structural variants; the ability and utility of OGM to detect structural variants in postnatal and rare disease samples; the ability and utility of OGM to be adopted for the identification of structural variants in postnatal and rare disease samples; the ability and utility of OGM to improve laboratory performance in terms of time and costs; and other statements that are not historical facts.

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 geopolitical and macroeconomic developments, such as the ongoing Ukraine-Russia conflict, related sanctions and the COVID-19 pandemic, on our business and the global economy; challenges inherent in developing, manufacturing and commercializing products; our ability to further deploy new products and applications and expand the markets for our technology platforms; failure of our OGM solutions to be adopted of the analysis postnatal and rare disease samples; the failure of OGM to detect structural variants consistent with the study results described in this press release; future study results that contradict the study results described in this press release; future study results that do not support the study results described in this press release; 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 obtain sufficient financing to fund our strategic plans and commercialization efforts; and 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 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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