Bionano Announces Publication of Interim Readout from Clinical Trial Run by Consortium Analyzing Optical Genome Mapping as Potential Standard of Care for Prenatal Testing

January 18, 2023

IRB-approved, multi-site, ongoing study evaluated 200 samples representing 123 unique prenatal cases across 9 sites for interim measures of key endpoints:

- *Results of OGM analysis were comparable, in a single assay, to the results of two separate standard of care (SOC) tests needed to reach a diagnosis in 56% of cases (69/123) and to three separate SOC tests needed to reach a diagnosis in 19% of cases (23/123)*
- *Concordance with SOC for all calls – 100% [200 out of 200 samples]*
- *Concordance with SOC for variant calls – 100% [78 out of 78 variants]*
- *Reproducibility of analytical QC from site-to-site – 100% [83 out of 83 replicates]*
- *Reproducibility of variant calls from site-to-site – 100% [83 out of 83 replicates]*

SAN DIEGO, Jan. 18, 2023 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (BNGO) today announced the publication of an interim report from an ongoing clinical trial designed to support establishing OGM as part of standard of care (SOC) in diagnosis of genetic disease for prenatal subjects. This publication reports on the prenatal genetic disease clinical trial program to evaluate OGM as an alternative to SOC workflows. This prenatal study focuses on comparing OGM to SOC, including concordance, reproducibility, technical success rates, turnaround time (TAT), diagnostic yield, and health economics. This first interim readout is designed to evaluate endpoints connected to analytical performance in key areas of technical performance and reproducibility of OGM.

“The process of establishing a consortium like this one to conduct a multi-site trial program is made possible by capable principal investigators and leading sites,” commented Alka Chaubey, PhD, FACMG, chief medical officer of Bionano. “We believe the trial is off to a terrific start, with a total of 414 subjects enrolled to date and with an interim readout of 123 subjects and 200 sample runs that show OGM performing very well. We look forward to the investigators proceeding with the remaining samples and evaluating other critical endpoints like comparative diagnostic yields, turnaround times and health economic impacts.”

Study Design
The study is an Institutional Review Board-approved, multicenter, double-blinded trial with samples from 123 clinical research subjects analyzed in a total of 200 sample runs to date. All samples had been previously tested with traditional methods like karyotyping, fluorescence in situ hybridization (FISH) and chromosomal microarray (CMA). The samples were from cases with known pathogenic or likely pathogenic variants (78), cases with known variants of uncertain significance (27), cases with no known reportable variant (18) and genomic controls (17).

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

- Equanimitas (Dr. Roger Stevenson)
- Cincinnati Children’s Hospital Medical Center (Dr. Jie Liu)
- University of Rochester Medical Center (Dr. Anwar Iqbal)
- Greenwood Genetic Center (Dr. Barbara DuPont)
- University of California San Francisco (Dr. Aleksander Rajkovic)
- Brigham and Women’s Hospital, and Harvard Medical School (Dr. Adrian M. Dubuc)
- Columbia University Irving Medical Center (Dr. Brynn Levy)
- Quest Diagnostics Nichols Institute (Dr. Peter Bui)
- Augusta University (Dr. Ravindra Kolhe)

Key Findings
This publication describes OGM performance metrics like robustness and reproducibility from site-to-site, operator-to-operator and run-to-run for the first time ever and for the largest number of prenatal samples investigated with OGM to date.

Key findings for the technical endpoints were reported as follows:

- *Results of OGM analysis were comparable, in a single assay, to the results of two separate SOC tests needed to reach a diagnosis in 56% of cases (69/123) and to three separate SOC tests needed to reach a diagnosis in 19% of cases (23/123)*
- *Concordance with SOC – 100% [200 out of 200 samples]*
- *Concordance with SOC for variant calls – 100% [78 out of 78 variants]*
- *Reproducibility of analytical QC from site-to-site – 100% [83 out of 83 replicates]*
- *Reproducibility of variant calls from site-to-site – 100% [83 out of 83 replicates]*

Key Takeaways
The publication concluded that these results demonstrate high technical performance of the OGM workflow from DNA isolation through data analysis. The authors reported that replicate run performance demonstrates reproducibility of OGM, suggesting it can be adapted and validated. The authors further pointed out that OGM is not limited to structural variant and copy number variation analysis alone but can also resolve repeat expansions greater than 500bp. The authors also cited OGM's ability to run an additional analysis pipeline for the screening of individuals with an expanded allele in the *FMR1* gene that could be causative of Fragile X syndrome. Screening for this repeat expansion is currently performed as a separate SOC test. The authors concluded that a single approach, like OGM, can allow genetic laboratories to provide rapid results with a cost-effective solution.

"Development and validation of OGM assays for prenatal 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 this publication demonstrating OGM's performance across multiple sites and its potential ability to perform in a single assay what requires two to three technologies in the SOC tests in practice today to reach a conclusive answer," commented Erik Holmlin, PhD, president and chief executive officer of Bionano. "I am eager to see the outcome for all trial subjects across the remaining endpoints. We are thrilled with the authors' recommendations for inclusion of OGM in the SOC testing in prenatal genetic analysis and these studies can provide important supporting data."

The publication can be found online at https://www.medrxiv.org/content/10.1101/2022.12.19.22283552v1.full-text

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.bionanogenomics.com, www.bionanolaboratories.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," "could," "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 include statements regarding our intentions, beliefs, projections, outlook, analyses or current expectations concerning, among other things, the potential of OGM to become a SOC prenatal test; the utility of OGM when used for the analysis of prenatal samples; and the ability of OGM to accurately detect genetic disorders, including structural variants, copy number variants, repeat expansions greater than 500bp and Fragile X syndrome, and, as a single technology, to result in a workflow that is cost-effective, highly sensitive and has a faster time to results than traditional cytogenetic methods. 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: global and macroeconomic events, such as the impact of the COVID-19 pandemic and the ongoing Ukraine-Russian conflict and related sanctions, 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 of OGM to accurately detect genetic disorders, including structural variants, copy number variants, repeat expansions greater than 500bp and Fragile X syndrome, and, as a single technology, to result in a workflow that is cost-effective, highly sensitive and has a faster time to results than traditional cytogenetic methods; future study results contradicting the results reported in the publication referenced above; 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; 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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Source: Bionano Genomics