Bionano Genomics Announces Publication of Interim Readout from the Consortium Conducting a Postnatal Clinical Trial Intended to Support Establishing Optical Genome Mapping as Part of Standard of Care in Genetic Disease Diagnosis

January 10, 2022

IRB-approved, multi-site, ongoing study evaluated 331 individual sample runs from 202 unique samples across 5 sites for interim measures of key endpoints:

- *Concordance with standard of care (SOC) – 97.7% [214 out of 219 samples]*
- *Partially concordant with SOC – 2.3% [5 out of 219 samples]*
- *Concordance with SOC for pathogenic variant calls – 100% [219 out of 219 samples]*
- *Concordance with chromosomal microarray (CMA) – 100% [103 out of 103 samples]*
- *First-pass success rate for OGM – 94% [311 out of 331 samples]*
- *Reproducibility of analytical QC from site-to-site – 98.8% [171 out of 173 replicates]*
- *Reproducibility of pathogenic variant calls from site-to-site – 100% [173 out of 173 replicates]*

SAN DIEGO, Jan. 10, 2022 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (BNGO), pioneer of optical genome mapping (OGM) solutions on the Saphyr® system and provider of N_XClinical™, the leading software solutions for visualization, interpretation and reporting of genomic data, today announced the publication of the first readout from the ongoing clinical trial designed to support establishing OGM as part of standard of care (SOC) in diagnosis of genetic disease for postnatal patients. This publication reports on the postnatal genetic disease diagnostic arm of Bionano's study to evaluate OGM as an alternative to SOC workflows in four key clinical areas: prenatal and postnatal genetic diseases, hematologic malignancies and solid tumors. The studies will compare OGM to SOC, including concordance, reproducibility, technical success rates, turnaround time (TAT), diagnostic yield, health economics and patient outcomes. 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 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 believe the trial is off to a terrific start, with a total of 813 subjects enrolled to date and as the interim readout of 202 subjects and 331 sample runs shows, OGM has performed 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 202 clinical research subjects analyzed in a total of 331 sample runs. 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 a genetic diagnosis (152), cases without a genetic diagnosis (6) and controls (44).

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 Bossler)

Key Findings
This 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 for the technical endpoints were reported as follows:

- Concordance with SOC – 97.7% [214 out of 219 samples]
- Partially concordant with SOC – 2.3% [5 out of 219 samples]
- Concordance with SOC for pathogenic variant calls – 100% [219 out of 219 samples]
- Concordance with CMA – 100% [103 out of 103 samples]
- First-pass success rate for OGM – 94% [311 out of 331 samples]
- Reproducibility of analytical QC from site-to-site – 98.8% [171 out of 173 replicates]
- Reproducibility of pathogenic variant calls from site-to-site – 100% [173 out of 173 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 copy number variation analysis alone, but can also resolve balanced structural rearrangements, size repeat expansions like *FMR1* and repeat contractions like D4Z4. In summary, the authors concluded that a single approach, like OGM, can allow genetic laboratories to provide rapid results with a cost-effective solution, which can benefit both the lab and the affected individuals.

"The OGM community is evaluating the whole workflow. The performance we have seen matches our expectations and we are happy with this publication announcing that OGM is performing well across multiple sites," commented Erik Holmlin, PhD, president and chief executive officer of Bionano. "Congratulations to this team for getting this paper published in 2021 and congratulations to Dr. Chaubey on the progress of her program. I am eager to see the outcome for all trial subjects across the remaining endpoints. We believe we can change the standard of care in genetic testing with OGM and these studies can provide important supporting data."

The full publication can be found online at https://www.medrxiv.org/content/10.1101/2021.12.27.21268432v1

For more information related to OGM and its application in genetic diseases and cancer, attend 2022 Symposium, Bionano's event for the OGM community. Symposium starts today, January 10, and runs until Thursday, January 13. For more information, visit www.bionanogenomics.com and a link to register for 2022 Symposium is available at https://www.labroots.com/ms/virtual-event/bngo2022.

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 "may," "will," "expect," "plan," "anticipate," "estimate," "intend" 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 reproducibility of the OGM technique and its ability to be easily adapted and validated; Bionano's clinical trial's ability to support establishing OGM as part of the SOC and to successfully measure critical endpoints comparing OGM to SOC; and the potential for OGM generally to become part of the SOC. 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, including the introduction of competitive technologies or improvements in existing technologies; failure of future study results to support those demonstrated in the publication referenced in this press release; 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 OGM or 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, 2020 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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