Prowess of Bionano Genomics' Saphyr System in Uncovering Novel Genetic Variations That Cause Cancer and Genetic Disease in Full Display at ASHG 2020

October 27, 2020

Top researchers present discoveries in cancer predisposition, neurodegenerative diseases, microdeletion syndromes and other genetic disorders made using Bionano's optical genome mapping technology

SAN DIEGO, Oct. 27, 2020 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO) announced that human genetics researchers using the Saphyr system will present their results at the American Society of Human Genetics (ASHG) Annual Meeting, being held virtually at www.ashg.org between October 27-30. The impact of structural variation analysis using the Saphyr system will be demonstrated at ASHG with 18 oral and poster presentations which cover an expanding array of diseases like cancer predisposition, microdeletion syndromes, repeat expansion disorders, neurodegenerative diseases, disorders of sex development and a variety of other genetic diseases. Additionally, these presentations show Saphyr's abilities to elucidate the exact structure of complex genomic rearrangements such as large inversions, chromothripsis and low copy repeats.

"The scientific importance and quality of the studies utilizing Saphyr and presented at ASHG have increased year over year," said Erik Holmlin, Ph.D., CEO of Bionano. "As more scientists present and publish their important discoveries made with Saphyr, an increasing number of potential future Saphyr users become aware of its prowess in uncovering novel genetic variants that contribute to cancer and genetic disease, which could drive more adoption and utilization for basic genetic research and clinical studies alike."

Below is a summary of key presentations to be given at ASHG 2020 featuring the use of Bionano's optical genome mapping technology:

Live Presentation – October 29, 2020, 11:45AM-12:00PM
Deciphering Genomic Inversions
Christopher M. Grochowski, Baylor College of Medicine
Genomic inversions are a class of structural variation (SV) relevant in evolution, speciation, and human disease but challenging to detect and resolve using current genomic assays. While short-read WGS can detect a fraction of copy number neutral inversions, those mediated by repeats or accompanied by CNVs remain challenging. The utilization of multiple technologies and visualization of unbroken DNA through long molecule approaches facilitate detection of *in cis* events and resolution of SVs containing two or more breakpoint junctions.

The following Co-Labs, Poster Sessions and Abstracts are available for on-demand viewing during and after ASHG 2020:

Bionano Laboratory Co-Lab Session: *Resolving Complex Haplotypes Implicated in Alzheimer's and Other Neurodegenerative Diseases.*
Mark T. W. Ebbert, Neuroscience Department, Mayo Clinic
Alzheimer's disease is genetically complex with no meaningful therapies or pre-symptomatic disease diagnostics. Most of the genes implicated in Alzheimer's disease do not have a known functional mutation, meaning there are no known molecular mechanisms to help understand disease etiology.

In this co-lab session, Mark T. W. Ebbert of the Mayo Clinic will discuss his team's work toward identifying functional structural mutations that drive disease in order to facilitate a meaningful therapy and pre-symptomatic disease diagnostic. Some of the genes and regions implicated in Alzheimer's disease are genomically complex and cannot be resolved with short-read sequencing technologies. These regions include MAPT, CR1, and the histocompatibility complex (including the HLA genes).

3342 – Bionano Poster Session: *High Throughput Analysis of Disease Repeat Expansions and Contractions by Optical Mapping*
Ernest Lam, Sr Manager Bioinformatics, Bionano Genomics
Repeat expansions and contractions are associated with degenerative disorders such as facioscapulohumeral muscular dystrophy (FSHD). Southern Blotting is the gold standard for long repeat analysis but has many limitations. Optical genome mapping allows for efficient analysis of diseases associated with repeat expansion and contraction.

2190 – Bionano Poster Session: *Rapid Automated large Structural Variation Detection in Mouse Genome by Whole Genome Sequencing*
Jill Lai, Sr Applications Scientist, Bionano Genomics
Identifying SVs for key model organisms such as mouse and rat is essential for genome interpretation and disease studies but has been historically difficult due to limitations inherent to available genome technologies. We updated the Saphyr analysis pipeline such that copy number variant (CNV) and SV analyses could now be applied to mouse and other non-human species, and constructed a control SV database for annotating variants, and identified strain-specific SVs/CNVs as well as variation shared among strains.

Additional presentations/abstracts featuring optical genome mapping:

3208 - *Long-read sequencing and optical mapping decipher structural composition of ATXN10 repeat in kindred with spinocerebellar ataxia and Parkinson's disease*
Presented by Birgitt Schuele, Associate Professor, Department of Pathology, Stanford University School of Medicine

3270 - *Uniparental isodisomy, structural and noncoding variants involved in inherited retinal degeneration (IRD) in three pedigrees*
Presented by Pooja Biswas, Ophthalmology Department, University of California, San Diego

Data CoLab: Whole Genome Map Assembly and Structural Variation Analysis with Hitachi Human Chromosome Explorer™
Presented by Hitachi-High-Tech America, Inc.

2123 - *High-throughput sequencing and mapping technologies applied to 10 human genomes with chromothripsis-like rearrangements*
Presented by Uirá Souto Melo, Mundlos Lab, Max Planck Institute for Molecular Genetics, Berlin, Germany

2165 - nanotatoR: A tool for enhanced annotation of genomic structural variants
Presented by Emmanuele Delot, Center for Genetic Medicine Research, Children's National Hospital, Washington, DC

2998 - *Highly variable structure and organization of the human 3q29 subtelomeric segmental duplications*
Presented by Umamaheswaran Gurusamy, Cardiovascular Research Institute, University of California San Francisco

2304 - Enlightening the dark matter of the genome: Whole genome imaging identifies a germline retrotransposon insertion in SMARCB1in two siblings with atypical teratoid rhabdoid tumor
Presented by Mariangela Sabatella, Princess Máxima Center for Pediatric Oncology, Utrecht, Netherlands

2318 - *FaNDOM: Fast Nested Distance-based seeding of Optical Maps*
Presented by Siavash Raeisi Dehkordi, Computer Science & Engineering, University of California San Diego, La Jolla

3023 - *Structural hypervariability of low copy repeats on chromosome 22 is human specific*
Presented by Lisanne Vervoort, Department of Human Genetics, KU Leuven, Leuven, Belgium

3024 - *Telomere-to-telomere assembly and complete comparative sequence analysis of the human chromosome 8 centromere*
Reviewer's Choice Award Recipient
Presented by Glennis Logsdon, Genome Sciences, University of Washington, Seattle, WA

3311 - *Comprehensive structural variant identification with optical genome mapping and short-read sequencing for diagnosis of disorders/differences of sex development (DSD)*
Reviewer's Choice Award Recipient
Presented by Hayk Barseghyan, Center for Genetic Medicine Research, Children's National Hospital, Washington, DC

3318 - *De novo mutation and skewed X-inactivation in girl with BCAP31-related syndrome*
Presented by H.J. Kao, Institute of Biomedical Sciences, Academia Sinica, Taipei, Taiwan

3560 - *Resolving genomic structures in MECP2 Duplication Syndrome provides insight into genotype-phenotype correlations*
Reviewer's Choice Award Recipient
Presented by Davut Pehlivan, Molecular and Human Genetics, Baylor College of Medicine, Houston, TX

2157 - *methometR: quantification of long-range haplotype specific methylation levels from Optical Genome Maps*
Presented by Surajit Bhattacharya, Center for Genetic Medicine Research, Children's Research Institute, Children's National Hospital, Washington, DC

About Bionano Genomics
Bionano is a genome analysis company providing tools and services based on its Saphyr system to scientists and clinicians conducting genetic research and patient testing, and providing diagnostic testing for those with autism spectrum disorder (ASD) and other neurodevelopmental disabilities through its Lineagen business. Bionano's Saphyr system is a platform for ultra-sensitive and ultra-specific structural variation detection that enables researchers and clinicians to accelerate the search for new diagnostics and therapeutic targets and to streamline the study of changes in chromosomes, which is known as cytogenetics. The Saphyr system is comprised of an instrument, chip consumables, reagents and a suite of data analysis tools, and genome analysis services to provide access to data generated by the Saphyr system for researchers who prefer not to adopt the Saphyr system in their labs. Lineagen has been providing genetic testing services to families and their healthcare providers for over nine years and has performed over 65,000 tests for those with neurodevelopmental concerns. For more information, visit www.bionanogenomics.com or www.lineagen.com.

Forward-Looking Statements
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 timing and content of the presentations identified in this press release; the effectiveness and utility of Bionano's technology in basic genetic research and clinical settings; the contribution of Saphyr to uncovering novel genetic variants that contribute to cancer and genetic disease; the benefits of Bionano's optical mapping technology and its ability to facilitate genomic analysis in future studies; and Bionano's strategic plans. 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 and the introduction of competitive products; 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; the loss of key members of management and our commercial team; 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, 2019 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.

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

Investor Relations Contact:

Ashley R. Robinson
LifeSci Advisors, LLC
+1 (617) 430-7577
arr@lifesciadvisors.com

Media Contact:
Darren Opland, PhD
LifeSci Communications
+1 (617) 733-7668
darren@lifescicomms.com

Source: Bionano Genomics