



Top Researchers to Present Discoveries Made Possible by Bionano's Saphyr System for Genome Imaging Technology at the ASHG 2019 Annual Meeting

Findings to be presented cover broad range of scientifically and clinically relevant areas including schizophrenia, sex development, cancer and muscular dystrophy

SAN DIEGO, CA, Tuesday Oct. 15, 2019 – Bionano Genomics, Inc. (Nasdaq: BNGO) today announced that disease researchers using Bionano's Saphyr system for whole genome imaging will present their results at the American Society of Human Genetics (ASHG) Annual Meeting, between October 15-19 in Houston, Texas.

The impact of analysis using the Saphyr system for ultra-sensitive and ultra-specific genome-wide detection of structural variation will be presented at ASHG with 22 oral and poster presentations and an Educational Event hosted by Bionano.

"ASHG 2019 represents a milestone for Bionano, with a record number of presentations demonstrating novel discoveries through our genome mapping technology," said Erik Holmlin, Ph.D., CEO of Bionano. "The growing use of the Saphyr system in disease research illustrates the value in identifying genomic variations for deep understanding of disease origin and diagnostic development."

Optical mapping through Saphyr enables the direct observation of large genomic variations through imaging of fluorescently labeled, megabase-size native DNA molecules. Next-generation sequencing (NGS), in contrast, relies on short-reads that piece together sequence fragments in an attempt to rebuild the actual structure of the genome. NGS often misses large DNA variations, such as deletions, insertions, duplications, and translocations and inversions. Genome mapping resolves these structural variations for more insight into the genetic variations that cause disease.

Below is a summary of key presentations to be given at ASHG 2019 featuring the use of optical genome mapping:

Genetic diagnosis of sex development disorders through optical mapping

Half of disorders of sex development (DSD) patients lack a firm diagnosis. Prof. Eric Vilain, from George Washington University and Children's National Medical Center, will present research validating the diagnostic and gene discovery use of Bionano genome mapping to identify structural variants in patients with DSD. The talk, entitled *Integration of optical genome mapping and sequencing technologies for identification of structural variants in DSD*, will be presented on Wed. Oct. 16 at 5:15 - 5:30 pm in the convention center Level 3, Room 361D.

Genomic mapping has the potential to replace a combination of current cytogenetic techniques

Currently, a comprehensive clinical analysis of genomic aberrations requires a combination of various assays such as CNV-microarrays, karyotyping and fluorescence *in situ* hybridization (FISH).
Dr. Tuomo

Mantere, from Radboud University Medical Center, will present data directly comparing traditional cytogenetic assays with Bionano mapping in leukemia patient samples to illustrate that genome mapping can identify all aberrations found by the three conventional technologies combined, and additional variants as well. The poster, entitled *Next-generation cytogenetics: High-resolution optical mapping to replace FISH, karyotyping and CNV-microarrays* will be presented on Thurs. Oct. 17, between 2 - 3pm, PgmNr 2533/T.

Genomic architecture reveals critical factors that may contribute to schizophrenia-associated 3q29 chromosomal deletion

Deletions at the 3q29 chromosomal locus are associated with a 40-fold increase in risk for schizophrenia. Knowing the features that contribute to genomic instability is critical for identifying risk factors of chromosomal deletions. Trenal Mosley, from Emory University, will present the discovery of novel genomic structural characteristics found in 12 patients with 3q29 deletion and their parents using Saphyr. The poster entitled, *Optical mapping of the schizophrenia-associated 3q29 deletion reveals new features of genomic architecture*, will be presented on Wed. Oct. 16, between 2 - 3pm, PgmNr 1389/W.

Bionano and NGS resolve complex rearrangements in extrachromosomal, circular DNA in glioblastoma

The rapid growth of aggressive tumors such as glioblastoma is partially caused by the rapid amplification of oncogenes in circular structures outside of native chromosomes. Because these structures do not occur in the reference genome, standard analysis methods fail to correctly assemble them. Jens Luebeck, from the University of California, San Diego, demonstrates that a combination of Bionano genome mapping and NGS resolves important breakpoints and gene amplifications in extrachromosomal DNA. The talk, entitled *Integrated Analysis of NGS and Optical Mapping Resolves the Complex Structure of Highly Rearranged Focal Amplifications in Cancer*, will be presented on Sat. Oct. 19, from 10:15 - 10:30am PgmNr: 323

Bionano Educational Event will feature research on muscular dystrophy, prenatal development & neurodegenerative disorders

At Bionano's educational event, Dr. Alka Chaubey from Perkin Elmer Genomics, Dr. Frances High from Mass General Hospital for Children, and Dr. Mark Ebbert from the Mayo Clinic will present findings from their work using the Saphyr system for structural genomic resolution. Analysis of chromosomal repeats, complex genomic haplotypes, and risk loci found in genetic disease will be highlighted by the speakers. Entitled *Resolving Structural Variants Across the Whole Genome to Power Your Next Discovery in Human Genetics*, the event will take place on Thurs. Oct 17, from 12:45 - 2:00pm at the Marriott Marquis, Houston, River Oaks, Level 3, and include a complimentary lunch.

Additional presentations featuring optical genome mapping:

High Throughput Analysis of Tandem Repeat Contraction Associated with Facioscapulohumeral Muscular Dystrophy (FSHD) by Optical Mapping

Presented by Jian Wang, Bionano Genomics

Wed. Oct. 16, 2 - 3pm PgmNr: 2535/W

Full Genome Analysis for Identification of Single Nucleotide and Structural Variants in Genes that Cause Developmental Delay

Presented by Hsiao-Jung Kao, Academia SINICA

Wed. Oct. 16, 2 - 3pm PgmNr: 2547/W

A Robust Benchmark for Germline Structural Variant Detection

Presented by Justin Zook, National Institute of Standards and Technology

Wed. Oct. 16, 2 - 3pm PgmNr: 1695/W

De Novo Genome Assembly and Phasing for Undiagnosed Conditions

Presented by Joseph Shieh, University of California, San Francisco

Wed. Oct. 16, 2 -3 pm PgmNr: 2529/W

Bionano Prep SP Isolates High Quality Ultra-high Molecular Weight (UHMW) Genomic DNA to Improve Research of Cancer and Undiagnosed Disorders

Presented by Henry Sadowski, Bionano Genomics

Wed. Oct. 16, 3 - 4pm PgmNr: 2598/W

nanotatoR: An Annotation Tool for Genomic Structural Variants

Presented by Surajit Bhattacharya, Children's National Medical Center

Wed. Oct. 16, 3 - 4pm PgmNr: 1506/W

Detection, Characterization, and Breakpoint Refinement of Balanced Rearrangements by Optical Mapping in Clinical Cases

Presented by Alex Hastie, Bionano Genomics + LabCorp

Thurs. Oct. 17, 2 - 3pm PgmNr: 2569/T

Genetic/epigenetic Diagnosis of Facioscapulohumeral Muscular Dystrophy (FSHD) via Optical Mapping

Presented by Yi-Wen Chen, Children's National Medical Center

Thurs. Oct. 17, 2 - 3pm PgmNr: 2533/T

Comprehensive Analysis of Structural Variants in Clinical Cancer Samples

Presented by Ernest Lam, Bionano Genomics

Thurs. Oct. 17, 3 - 4pm PgmNr: 1060/T

Advanced Structural Analysis of CDH Risk Loci with Optical Genome Mapping Technology

Presented by Mauro Longoni, Massachusetts General Hospital

Thurs. Oct. 17, 3 - 4pm PgmNr: 2578/T

Structural Variants Associated with GWAS SNPs Provide Mechanistic Explanation of Phenotypic Associations

Presented by Seth Berger, Children's National Medical Center
Thurs. Oct. 17, 3 - 4pm PgmNr: 2254/T

The Complete Linear Assembly and Methylation Map of Human Chromosome 8

Presented by Glennis Logsdon, University of Washington
Fri. Oct. 18, 1 - 2pm PgmNr: 1703/F

High Throughput High Molecular Weight DNA Extraction from Human Tissues for Long-read Sequencing

Presented by Kelvin Liu, Circulomics
Fri. Oct. 18, 1 - 2pm PgmNr: 1769/F

Optical Mapping for Chromosomal Abnormalities: A Pilot Feasibility Study for Clinical Use

Presented by Gokce Toruner, UT MD Anderson Cancer Center
Fri. Oct. 18, 1 - 2pm PgmNr: 2447/F

Comprehensive Detection of Germline and Somatic Structural Mutation in Cancer Genomes by Bionano Genomics Optical Mapping

Presented by Mark Ebbert, Mayo Clinic
Fri. Oct. 18, 2 - 3pm PgmNr: 1760/F

'Dark' and 'Camouflaged' Genes May Harbor Disease-relevant Variants that Long-read Sequencing Can Resolve

Presented by Andy Pang, Bionano Genomics
Fri. Oct. 18, 2 - 3pm PgmNr: 1814/F

Bionano Genomics "Sample to Answer" Workflow for Single Molecule Analysis of Variation in Genome Structure

Presented by Sven Bocklandt, Bionano Genomics
Fri. Oct. 18, 2 - 3pm PgmNr: 1838/F

Draft Assembly of an Armenian Genome

Presented by Hayk Barseghyan, Children's National Medical Center
Fri. Oct. 18, 2 - 3pm PgmNr: 2342/F

About Bionano Genomics

Bionano is a life sciences instrumentation company in the genome analysis space. Bionano develops and markets the Saphyr system, 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 digital cytogenetics, which is designed to be a more systematic, streamlined and industrialized form of traditional cytogenetics. The Saphyr system comprises an instrument, chip

consumables, reagents and a suite of data analysis tools. For more information, visit www.bionanogenomics.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, including among other things: the timing and content of the presentations identified in this press release; and the ability of genome mapping to perform comprehensive clinical analysis as well as conventional technologies. 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 that our sales, revenue, expense and other financial guidance may not be as expected, as well as risks and uncertainties associated with 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 key clinical studies to demonstrate the effectiveness of our products; 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, 2018 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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