# bionano<sup>°</sup> genomics

## Bionano Publishes Method for Identifying Genes Likely to Cause Neurological Diseases Based on a Model Developed by Lineagen to Improve Interpretation of Variants of Unknown Significance

### February 22, 2021

### Proprietary scoring algorithm may help identify novel genes for nervous system disorders; applications include patient testing and eventually discovery research with Saphyr

SAN DIEGO, Feb. 22, 2021 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO) announced the publication of a study by its Lineagen team that describes the first genome-wide model to assess and score nearly all protein-coding genes based on their likelihood to cause central nervous system (CNS) disease. The resulting list of high scoring genes enhances Lineagen's clinical interpretation capabilities allowing their genetic counsellors to better interpret variants of unknown significance (VUS), which can improve diagnostic abilities and provide for more precise counseling to patients and families. The gene scoring model can also be integrated into Bionano's analysis software for Saphyr data, allowing scientists and clinicians to quickly identify and filter structural variants based on their increased risk of causing CNS diseases.

The model, called the Neurogenic Systematic Correlation of Omics-Related Evidence, or NeuroSCORE, combines data from five public and proprietary genomics databases containing evidence on whether genes are highly expressed in the adult or developing human brain, have high rates of new genetic variation in people with CNS diseases and have few variants in individuals without neurological disorders. Armed with NeuroSCORE, scientists could discover novel genes not previously associated with CNS diseases, potentially enabling clinicians to more accurately diagnose patients with previously uncharacterized neurodevelopmental disease. These advances may make it possible to better study the complex molecular mechanisms for conditions such as autism spectrum disorder and other disorders of childhood development, which are often caused by a combination of genetic variants affecting multiple genes and pathways.

Erik Holmlin, PhD, CEO of Bionano Genomics commented: "In the twenty years since the first human genome was sequenced, scientists have made countless discoveries about the function and mechanisms of the 20,000 human genes and the proteins they encode. Despite this progress, many thousands of genes are poorly characterized and their function unknown. At the same time, a large number of patients with genetic disease don't receive a molecular diagnosis even after several genetic analyses because many of the identified variants are of unknown significance to the disease. Now, our Lineagen team can better interpret these VUS using our proprietary NeuroSCORE algorithm to diagnose more patients and provide more conclusive answers to families of patients with neurodevelopmental disease. When combined with the power of optical genome mapping with Saphyr, we believe neuroscientists could detect all structural variants genome wide and score them to make new discoveries. The NeuroSCORE publication is a great example of why Bionano acquired Lineagen last year – so that Lineagen's extensive experience and understanding of genetic diseases, including neurodevelopmental disease, can accelerate advancement of applications for Saphyr."

The publication is available on bioRxiv at https://www.biorxiv.org/content/10.1101/2021.02.04.429640v1

### **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 research use only 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. Bionano provides 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. Forwardlooking statements include statements regarding our intentions, beliefs, projections, outlook, analyses or current expectations concerning, among other things: the potential for NeuroSCORE to enable discovery of novel genes and improve diagnostic capabilities for patients with genetic disease. including neurodevelopment disease; our beliefs regarding the potential benefits of integrating NeuroSCORE with Bionano's existing Saphyr technology; the contribution of expertise and resources from Lineagen to the advancement of current and future applications for our Saphyr system; and the execution of 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.

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Source: Bionano Genomics