

New York Genome Center Receives New York State Approval for Clinical Constitutional Whole Genome Sequencing Testing

Test offers the most complete package of genomic data available for patients with undiagnosed diseases, as well as patients seeking predispositional genetic testing

NEW YORK, NY (February 3, 2020) The New York Genome Center (NYGC) has received approval from the New York State Department of Health to offer clinical constitutional whole genome sequencing (WGS) testing in its Clinical Laboratory Evaluation Program-certified laboratory.

The WGS test provides clinicians with the most complete package of genomic data available for patients with undiagnosed diseases, as well as for patients seeking predispositional genetic testing, i.e., a systematic early detection or exclusion of a hereditary disease, a genetic predisposition to a disease, or to determine whether they carry a predisposition which may produce a hereditary disease in their children.

Since acquiring conditional approval for the WGS test a few years ago, the NYGC has developed new tools and an updated pipeline that further enhance its capabilities. The test is now performed using Illumina NovaSeq sequencers, allowing for greater speed and sample volumes than when run on the previous HiSeq X platform. Sequencing reads are now mapped to the latest hg38 reference human genome build, considered to be the most accurately sequenced genome build to date. These updates provide an opportunity to uncover novel genetic etiologies that were previously not about to be identified by older reference genome builds.

NYGC scientists have also updated the test's analysis workflow to include discovery of more genetic variant types. In addition to the single nucleotide variants, insertions/deletions, copy number variants, and variants in the mitochondrial genome, the test examines repeat expansions in selected genes, and copy number analysis in survival motor neuron genes to identify spinal muscular atrophy disease/carrier status. The test also deploys the novel software tool Expansion Hunter, developed in a collaboration with Illumina and NYGC and a global team of research partners, that can genotype repeats at the locus of interest, even if the expanded repeat is larger than the read length. The team's report on this innovative test tool was published by *Genome Research* in 2017.

Led by Vaidehi Jobanputra, PhD, Director of Molecular Diagnostics, NYGC's Clinical Lab is licensed to perform clinical testing in all fifty (50) of the United States and the District of Columbia. The lab's Whole Genome and Transcriptome Sequencing (WGTS) test for cancer patients, developed by NYGC scientists, received New York Department of Health approval in 2018. For more information, visit the NYGC Clinical Lab website at <https://www.nygenome.org/clinical-products/nygc-diagnostic-tests/>.

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About the New York Genome Center

The New York Genome Center (NYGC) is an independent, nonprofit academic research institution focused on furthering genomic research that leads to scientific advances and new insights and therapies for patients with neurodegenerative disease, neuropsychiatric disease, and cancer. Leveraging our strengths in whole genome sequencing, genomic analysis, and development of new genomic tools, the NYGC serves as a nexus for collaboration in disease-focused genomic research for the New York medical and academic communities and beyond.

NYGC harnesses the expertise and builds on the combined strengths of our faculty, staff scientists, member institutions, scientific working groups, affiliate members, and industry partners to advance genomic discovery. Central to our scientific mission is an outstanding faculty who lead independent research labs based at the NYGC, and hold joint tenure-track appointments with one of our member institutions.

Institutional founding members of the NYGC are: Cold Spring Harbor Laboratory, Columbia University, Albert Einstein College of Medicine, The Jackson Laboratory, Memorial Sloan Kettering Cancer Center, Icahn School of Medicine at Mount Sinai, New York-Presbyterian Hospital, New York University, Northwell Health, The Rockefeller University, Stony Brook University, and Weill Cornell Medicine. Institutional associate members are: American Museum of Natural History, Georgetown Lombardi Comprehensive Cancer Center, Hackensack Meridian Health, Hospital for Special Surgery, The New York Stem Cell Foundation, Princeton University, and Roswell Park Cancer Institute.

For more information on the NYGC, please visit: <http://www.nygenome.org>.

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