



# Johns Hopkins to Open New Institute Combining Genomics Capabilities, Expertise

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NEW YORK (GenomeWeb) – Johns Hopkins is combining its research and clinical genomics expertise and capabilities into one institution, Johns Hopkins Genomics.

The new center will eventually be located in a new building in the East Baltimore BioPark, adjacent to the medical school, and will open next July and will have CLIA certification and serve both research and clinical customers.

Johns Hopkins Genomics is a joint effort between the McKusick-Nathans Institute of Genetic Medicine and the Department of Pathology. It will bring two major research labs — the National Institutes of Health-funded Center for Inherited Disease Research and Johns Hopkins' Genetic Resources Core Facility — and three clinical genomics labs — the Molecular Pathology Lab, the DNA Diagnostics Lab, and the Clinical Genomics Lab — under one roof.

The center will have about 200 staff members and offer a range of both research and clinical services. Research services will include genotyping, all types of sequencing applications like RNA-seq, microbiome sequencing, ChIP-seq, targeted panels, exome and whole-genome sequencing.

In addition, the center will offer sample processing, including live cell immortalization and a CAP-accredited biorepository. The center will also have digital PCR and qPCR assays, cell line authentication, and single-cell genomics capabilities.

On the clinical side, it will offer clinical exome sequencing, custom targeted sequencing for Mendelian variants and somatic mutations, phenotype-based gene panels for inherited disorders, somatic cancer gene panels, and clinical cytogenetics services.

The center's equipment will come primarily from the Genetics Resources Core Facility, which includes three Illumina HiSeq 2500s, two MiSeqs, and one Thermo Fisher Ion Proton machine. The center will also have PCR and Sanger sequencing equipment, including two of Thermo Fisher's QuantStudio 12K Flex machines, one QuantStudio 3D instrument, one TaqMan 7900HT, and two 3730XL machines. The DNA Diagnostic Laboratory and the Molecular Pathology Lab will also supply some instruments and additional equipment will be leased as needed.

David Valle, director of Johns Hopkins' Institute of Genetic Medicine, told GenomeWeb that the university decided to form the new entity in order to bring the various laboratories and investigators doing genomics under one roof "for reasons of intellectual cross fertilization as well as economies of scale."

Of note, Valle said Johns Hopkins has significant analytic expertise, which has largely been focused on the research side of genomics. However, moving both the research and clinical genomics groups under one roof will enable researchers to tap into that analytic expertise for the clinical arena. "That will be a

great step forward for us," he said.

For instance, Steven Salzberg's computational biology lab at the Institute of Genetic Medicine has developed novel methods for analysis of DNA and RNA sequences, as well as methods for genome assembly, Valle said. "And, we're very excited about bringing this analytic expertise to bear on the clinical groups."

The new center will offer research and clinical services for both internal researchers and patients as well as external ones.

Recently, the Clinical Genome Laboratory developed a clinical exome test, which will be migrated to the new facility. The test runs on the HiSeq 2500 and uses Agilent SureSelect for exome capture.

Valle said that the primary use of the exome test would be to diagnose unknown diseases. But, he said that in some cases, the laboratory has already used the test for critically ill newborns. In those cases, the laboratory makes use of the 2500's rapid run mode.

In addition, Valle said he anticipates using exome sequencing for an increasingly broad range of conditions, including in the cancer space. "Clinical exome sequencing is moving into mainstream medicine, so I imagine it will be under high demand."

Valle said that it frequently makes more economic sense to do an exome test as opposed to single-gene or small panels. For instance, in unknown disorders where the phenotype is complex, physicians often get caught up in a cycle of ordering one gene or set of genes, and when the test is negative, they go back to the drawing board to determine the next gene or panel to order. The process is time consuming and costs quickly build up, making "exome sequencing quite a bargain," Valle said.

Nonetheless, insurance companies are often reluctant to pay for tests, although groups are increasingly building evidence that exome sequencing can be useful and cost effective. Professional organizations, like the American College of Medical Genetics and Genomics, are also getting behind NGS-based clinical testing. The ACMG [issued a statement](#) earlier this year, broadening its definition of clinical utility.

Johns Hopkins Genomics will bill insurance companies for its exome and other NGS-based clinical tests, Valle said. Pricing for the clinical exome is still being determined, but Valle said that it would be "cost competitive."

Currently, Johns Hopkins' core facility offers research-based exome sequencing for \$1,325.

Aside from a clinical exome, the center will offer a range of gene panels for both inherited disease and oncology applications. Oncology tests are typically done for diagnosis, for instance, in the case where the tumor is of unknown primary origin, or to determine the appropriate treatment.

Increasingly, clinical genomics is moving away from PCR-based single-gene tests, Valle said. And, while the new institution will still have those capabilities, he said that he thinks "PCR-based tests will be pretty quickly phased out. Although, there may be certain targeted mutations that would still be done on PCR-based sequencing."

Initially, Valle said he expects most of center's work will be research focused, but that clinical testing would make up an increasing percentage as the laboratory establishes its clinical pipeline and as clinical sequencing becomes more widely adopted.

Valle anticipates about a few hundred patients in the first year of operations, jumping to thousands in subsequent years. "We want to be a little bit careful as we ramp up the pipeline to make sure all the bumps are out," he said.