Illumina Genome Network

Delivering whole human genome sequencing services using the industry's most respected sequencing platforms.

Highlights

- **Most Widely Adopted and Published Technology**
  Cited in more than 5,400 publications
- **Uncompromising Data Quality**
  Highest sensitivity and specificity
- **Expert Sequencing Service Providers**
  CSPro® providers have extensive sequencing and data analysis experience
- **Cost-Effective Services with Fast Turnaround Times**
  Committed to providing industry-leading turnaround times
- **Simplified Data Analysis**
  Standard analysis included, with raw data provided in the most widely adopted formats

Whole-Genome Sequencing When You Need It

Next-generation sequencing systems, such as the Illumina HiSeq® and NextSeq™ systems, provide the throughput and data quality necessary to support a broad range of applications for human genomics research. For example, whole-genome sequencing can identify disease-associated genes, determine the differences and similarities between distinct human populations, or identify the gene variations that make each person genetically unique. Follow-on studies such as targeted resequencing, SNP discovery, structural variation analysis, and RNA-Seq can provide in-depth information regarding specific genes and mutations, and their impact on gene expression.

The Illumina Genome Network enables researchers to leverage human whole-genome sequencing—even with low sample volumes—without the time and expense of acquiring equipment and training personnel. It enables completion of whole-genome sequencing projects quickly and affordably, by providing access to Illumina FastTrack Services and Illumina Certified Service Providers (CSPro)† —organizations with expert sequencing teams using Illumina platforms.

What is the Illumina Genome Network?

The Illumina Genome Network is a group of leading institutions worldwide, with experience and expertise in Illumina technology, which offers sequencing services to individual researchers. Customers can connect with the Network provider of their choosing to create a service package that meets their individual needs.

Expert Genome Network Providers

Illumina hand picks its Genome Network providers to ensure that sequencing projects are completed to the highest-quality standards with rapid turnaround. All have proven their sequencing expertise with numerous publications and extensive training, enabling them to achieve Illumina CSPro status. Researchers benefit from their skill and vast amount of data analysis knowledge.

All Illumina Genome Network providers have multiple HiSeq systems, ensuring the capacity and scalability necessary to manage large projects. Current members include British Columbia Cancer Agency, the Broad Institute, Cold Spring Harbor Laboratory, HudsonAlpha Institute for Biotechnology, Illumina FastTrack Services, the Macrogen Genomic Medicine Institute, McGill University, New York Genome Center, Peking University, Science For Life, Takara Bio Inc., and the University of Washington Department of Genome Sciences. For the most current list of Illumina Genome Network providers, visit www.illumina.com/genomenetwork.

Proven Technology

Illumina sequencing platforms employ sequencing by synthesis (SBS) chemistry, the most widely used and proven next-generation sequencing chemistry with more than 5,400 publications to date. Whole-genome sequencing with SBS chemistry yields the highest-quality data and provides the most powerful ability to detect rare variants—all with the speed and reliability needed to move research forward.

High-Quality Data

Illumina sequencing platforms deliver the most accurate human genome at any coverage level. Illumina SBS chemistry provides the highest yield of error-free reads, and fewer false negatives and false positives than competing technologies. With Illumina sequencing platforms, researchers can count on exceptional data quality.

Sequence More Cost-Effectively, Quickly

Illumina has negotiated competitive prices with its Genome Network providers. The per genome cost will be at the same level as similar services that use less accepted technologies performed by less experienced users. In addition, the Illumina Genome Network is committed to providing industry-leading turnaround times.

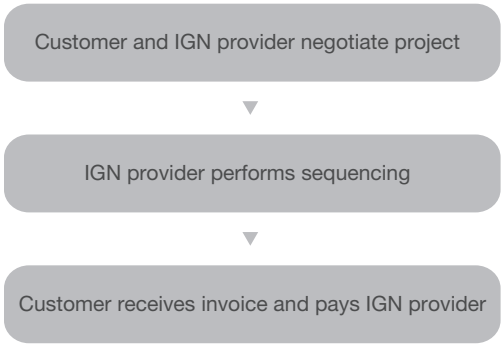
Analysis Assistance

Sequencing through the Illumina Genome Network provides more than a virtual instrument. Illumina Genome Network providers offer simplified data analysis solutions. Customers receive raw data in the most widely adopted formats with access to the broadest selection of commercial and open-source analysis tools in the industry. Standard analysis is included at no charge, with premium data analysis services also available.

Additionally, Illumina has partnered with leading data analysis and visualization companies, including Knome and Diagnomics, to provide optimized software tools for downstream filtering and interrogation of IGN data sets that will deliver biologically relevant and actionable results.

†The Illumina CSPro Program is a collaborative service partnership dedicated to ensuring delivery of the highest quality data available for genetic analysis applications. Learn more at www.illumina.com/cspro.

Figure 1: The Illumina Genome Network Process



Follow-On Study Support

The Illumina Genome Network offers researchers an affordable entry point to Illumina next-generation sequencing. Following the completion of a whole-genome sequencing project, Illumina Genome Network or CSPro providers can perform follow-on projects using Illumina SBS-based platforms, ensuring efficient analysis.

How Does it Work?

To make accessing Illumina sequencing technology as easy as possible, Illumina has committed to support each IGN provider as they take on and complete whole-genome sequencing projects. Customers work directly with each provider to execute their project—receiving high-quality data with rapid turnaround times (Figure 1).

What Does the Service Include?

Illumina Genome Network providers perform whole human genome sequencing at 30× average coverage, from short-insert paired-end reads (Table 1). Each provider can also offer genotyping with the HumanOmni2.5 BeadChip, providing enhanced confidence in sample validation and supporting the ability to quickly assess population level genome characteristics in large sample projects. For SNV and indel detection, data analysis is performed using commercially supported alignment and variant calling pipelines, such as Illumina Sequence Analysis and Comparison (ISAAC)[1] aligner, BWA[2]+GATK,[3] or other pipeline software, while ancillary software is used for CNV and SV detection. The delivered data set will include reads and compressed quality scores that reduce the data footprint and cost of storage without impacting quality of downstream analysis results.

Human Whole-Genome Sequencing Without Compromise

Reliable. Accurate. Affordable. Customers can trust their large-scale human whole-genome sequencing projects to Illumina Genome Network providers, with their proven ability to obtain results using the most widely adopted sequencing technology. Start today at www.illumina.com/genomenetwork.

Table 1: Illumina Genome Network Service Details

Sample Input Requirements
DNA input requirement: 500 ng–3 µg
Service Performance Specifications
Average autosomal fold coverage of 30× (higher coverage offered)
Percent of non-N reference coverage > 90% for non-cancer samples
At least 3,000,000 SNPs detected for non-cancer samples
Data Deliverables
Flexible alignment and variant calling pipelines.
Variants (Delivered in VCF and gVCF format)*
Single nucleotide polymorphisms (SNPs)
Indels (1–50 bp)
Copy number variation (losses and gains)
Structural Variants (SVs)
Inversion
Segmental duplications
Interchromosomal translocations
Intrachromosomal translocations
Large insertions (> 50 bp)
Large deletions (> 50 bp)
Reads
Aligned and unaligned reads in archival BAM format
Annotations*
dbSNP comparisons
Gene and functional impact annotations
Disease association
Allele frequencies
Genotyping array data (optional)
Summary report
Delivery
Electronic delivery provided by select providers. All results formatted for additional downstream analysis.
Premium data analysis packages available through select Illumina Genome Network software providers

* Variable per IGN provider.

References

1. Raczy C, Petrovski R, Saunders CT, Chorny I, Kruglyak S, et al. (2013) Isaac: ultra-fast whole-genome secondary analysis on Illumina sequencing platforms. Bioinformatics 29(16):2041–2043.
2. Burrows-Wheeler-Aligner (http://bio-bwa.sourceforge.net/).
3. Genome Analysis Toolkit (http://www.broadinstitute.org/gatk/).

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