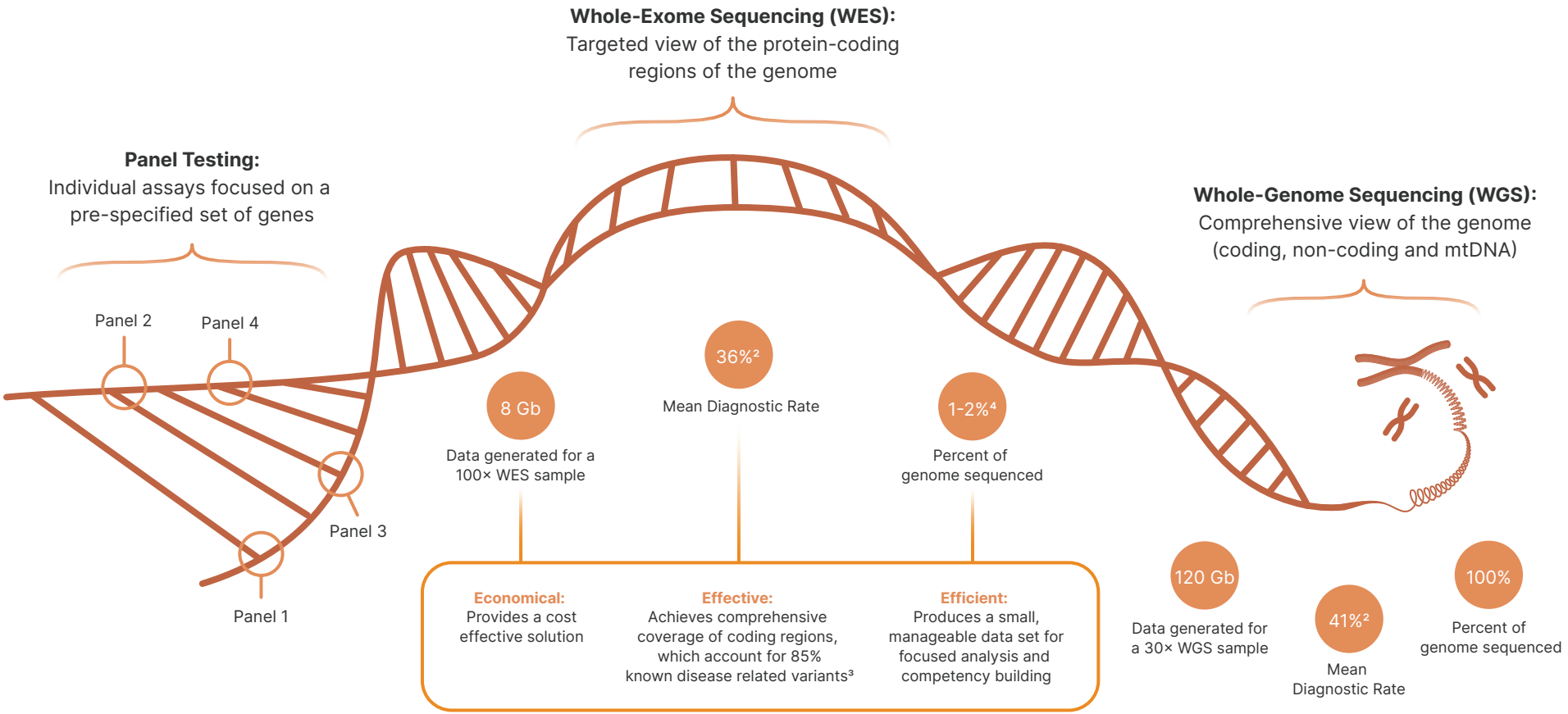
Your bridge to

Discover complete workflow offerings and support for whole-exome sequencing.

Anxhela Gustafson, PhD
Scientist
Genomics Institute at Shriners Children's

from panels to new frontiers of the genome

Exome sequencing is an effective, economical, and efficient approach when whole-genome sequencing is not accessible.

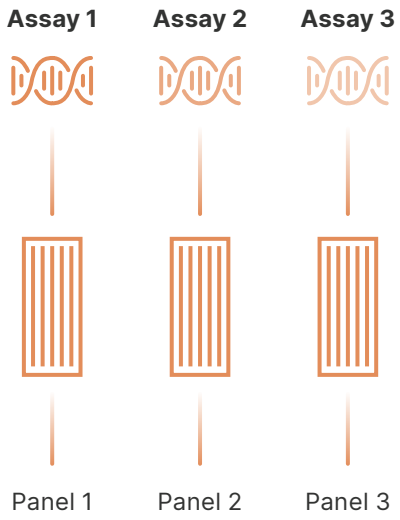


Single platform for panel designs

Approximately 250 new genes with disease-causing variation are added to the literature annually.[1] This rapid rate of genetic discovery can render current gene panels outdated and incomplete. The subsequent need to update multiple panels on a regular basis can be both labor-intensive and costly. By adopting a whole exome backbone, labs can deliver versatile and comprehensive virtual panels, with simplified workflows and decreased sequencing costs.

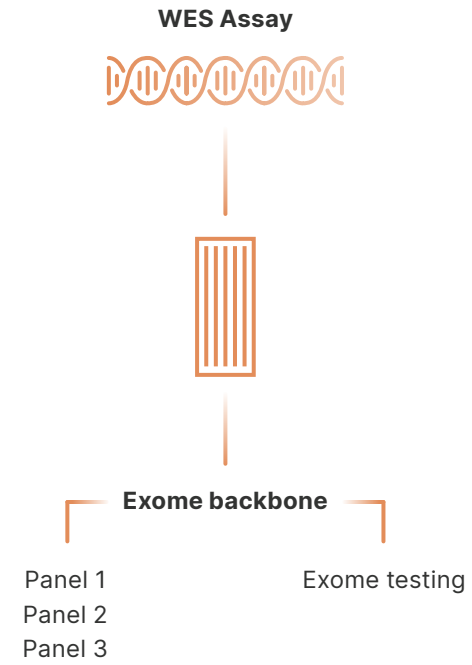
Panel testing

- Limited test menus
- Complicated multi assay panel workflows
- Costly and labor-intensive panel update workflow
- Finite ability to discover new genotype-phenotype associations
- Limited re-analysis options



Whole-exome sequencing

- Optimized lab efficiency via assay consolidation
- Single wet lab assay validation, frequent query
- Simplified workflow for panel updates
- Enhanced ability for new discoveries
- Immediate reflex analysis capabilities



All the parts and support from a single partner

Library Preparation

Illumina DNA Prep with Exome 2.5 Plus Enrichment
Combines on-bead tagmentation with built-in library normalization and hybridization enrichment allowing for:

- High level performance with only 50M PE reads
- ~90% Padded Read Enrichment and ~98% Uniformity of Coverage
- ~99% SNV Precision and ~97% SNV recall

Illumina library prep automation protocols are compatible with Beckman Coulter, Eppendorf, Hamilton, PerkinElmer, Tecan, and more. Exome 2.5 Plus may require minor modifications.

Features:

- Flexible and direct DNA input
- Fast, efficient, and reproducible enzyme tagmentation
- Comprehensive exome panel content
- >90% coverage of targets across clinically relevant databases
- Rapid turnaround time from sample to enriched, sequence-ready libraries

Sequencing

NextSeq™ 550 :
Cost-effective benchtop analyzer with:

- ~120 Gb output range
- 400M maximum single-end reads
- 2×150 bp maximum read length
- 5-16 exomes per run*

NextSeq 550 Dx may be used in RUO mode

NextSeq 1000/2000:
Easy-to-use cartridge-based platform with:

- ~360 Gb maximum output range
- 1.2B maximum single-end reads
- 2×300 bp maximum read length
- 4-48 exomes per run*

NovaSeq™ 6000:
Automation- and configuration-friendly system built for deep and broad coverage with:

- 6 Tb maximum output range
- 20B maximum single-end reads
- 2×250 bp maximum read length
- 24-500 exomes per run*

NovaSeq 6000 Dx may be used in RUO mode

NovaSeq X series:
Powerful system for ultra-high-throughput projects:

- 8 Tb† -16 Tb‡ maximum output range
- 26B† - 52b‡ single-end reads
- 2×150 bp maximum read length
- 40-1500 exomes per run*

*depending on flow cell type and desired read depth
† NovaSeq X System
‡ NovaSeq X Plus System
Note: Suggested read-length for WES is 2×101

Data Analysis

Illumina DRAGEN™ Enrichment Pipeline
Accurate, efficient secondary analysis solution for comprehensive variant calling, including SNV, CNV, and SV

Emedgene
Enable high efficiency tertiary analysis for germline research WES, with the support of explainable AI (XAI) and user-configured automation to maximize scale potential

WES workflows with Illumina Genomics Architecture (IGA)

IGA is a standardized, modular, and flexible framework that streamlines the integration and deployment of NGS by implementing automation-compatible, sample-to-answer workflows through a series of hardware and software.

Hardware

Automated liquid-handling platform

- **NextSeq 1000/2000 System**
- **NovaSeq 6000 System**

Library preparation → **Sequencing** → **Data/variant analysis**

Software

Clarity LIMS (Laboratory Information Management System) software:

- Orchestrates between the user, liquid handler, sequencing system, and data analysis platform
- Supports WES/WGS workflows

DRAGEN secondary analysis:
Provides rapid, automated primary (BCL to FASTQ conversion, or FASTQ generation) and secondary (mapping, alignment, variant calling) analyses

BaseSpace Sequence Hub:
Delivers data from sequencing run to Illumina Connected Analytics

Illumina Connected Analytics:
Represents a comprehensive, security-first, cloud-based platform for data storage, management, and analysis; interfaces directly with DRAGEN secondary analysis

For more information, visit: https://support-docs.illumina.com/SHARE/IlluminaGenomicsArchitecture/Content/SHARE/FrontPages/IGA.htm

Branch out with whole-exome sequencing

Trust Illumina to be your single partner for providing:

- Reagents, instrumentation, and software for data analysis and interpretation
- Fast, easy-to-use, robust assays for high-quality, reproducible results
- World class support for every element of your workflow

Learn more about the power of whole-exome sequencing here:

Scan QR code for more information

https://www.illumina.com/areas-of-interest/genetic-disease/rare-disease-genomics/targeted-rare-disease-seq.html

References

1. Seaby EG, Rehm HL, O'Donnell-Luria A. Strategies to Uplift Novel Mendelian Gene Discovery for Improved Clinical Outcomes. *Front Genet*. 2021;12. doi:10.3389/fgene.2021.674295.

2. Clark MM, Stark Z, Farnaes L, et al. Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases. *NPJ Genom Med*. 2018;3(1). doi:10.1038/s41525-018-0053-8 2.

3. van Dijk EL, Auger H, Jaszczyszyn Y, Thermes C. Ten years of next-generation sequencing technology. *Trends Genet*. 2014;30:418-426. doi: 10.1016/j.tig.2014.07.001.

4. Warr A, Robert C, Hume D, Archibald A, Deeb N, Watson M. Exome Sequencing: Current and Future Perspectives. *G3: Genes|Genomes|Genetics*. 2015;5(8):1543-1550. doi:10.1534/g3.115.018564.

illumina®

1.800.809.4566 toll-free (US) | +1.858.202.4566 tel
techsupport@illumina.com | www.illumina.com

M-GL-01485 v2.0