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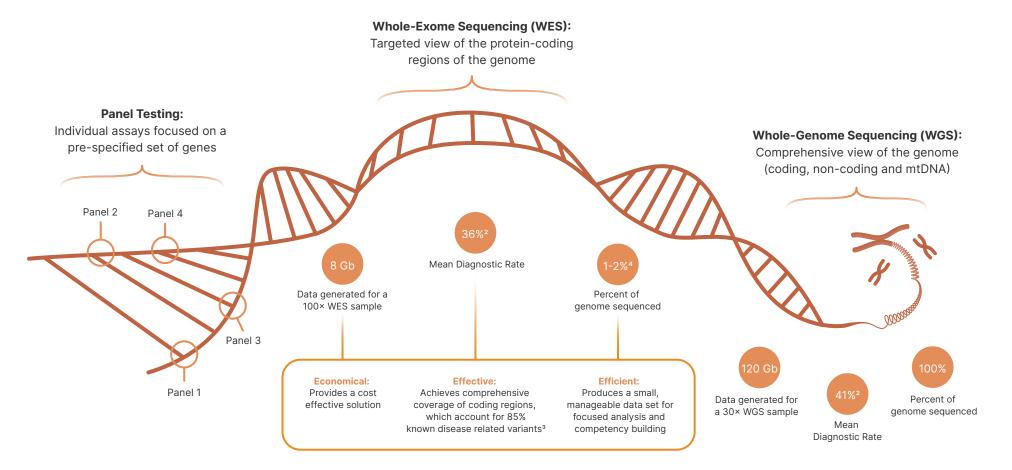
Discover complete workflow offerings and support for whole-exome sequencing.

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

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from panels to new frontiers of the genome

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



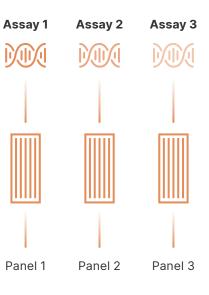
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Approximately 250 new genes with disease-causing variation are added to the literature annually.¹ This rapid rate of genetic discovery can render current gene panels outdated and incomplete. The subsequent need to update multiple panels on a regular 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.

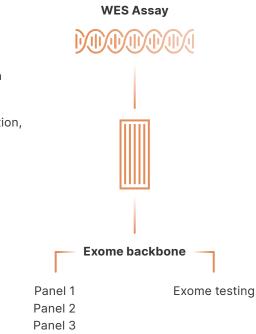
Panel testing

- 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

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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

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



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



Sequencing

NextSeg 1000/2000:

Easy-to-use cartridge-based platform with:

rtner

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



Illumina DRAGEN™ Enrichment Pipeline

Accurate, efficient secondary analysis solution for comprehensive variant calling, including SNV, CNV, and SV



NovaSeq X series:

Powerful system for ultra-high-throughput projects:

- 8 Tbt -16 Tbt maximum output range
- 26Bt 52bt 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



Emedgene

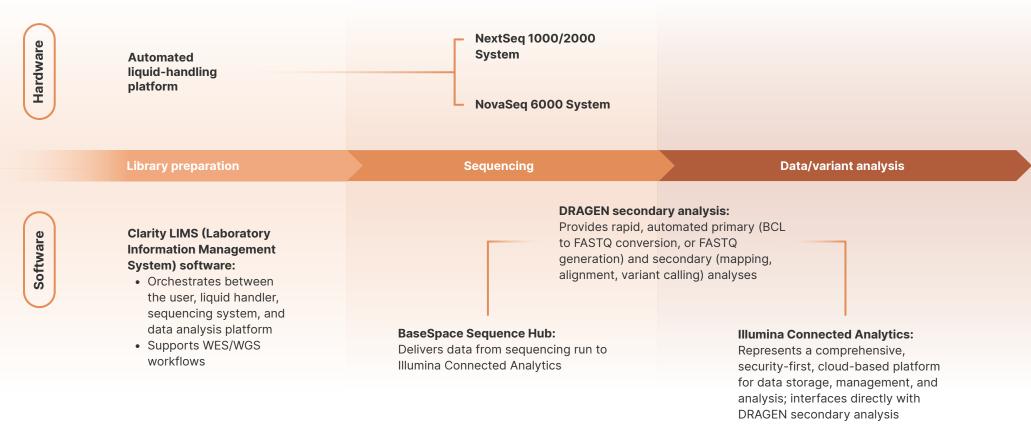
Enable high efficiency tertiary analysis for germline research WES, with the support of explainable AI (XAI) and user-configured automation to maximize scale potential

Data Analysis

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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.



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

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- Fast, easy-to-use, robust assays for high-quality, reproducible results
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