Standardize NGS Panel with a focused, economical workflow

Clinical Exome Sequencing (CES)

Background

Clinical Exome Sequencing (CES) focuses on genes with clear clinical relevance. This focused approach optimizes sequencing costs while maintaining high diagnostic yields for carrier screening and disease diagnosis.

We have designed a standardized, economical CES workflow that utilizes a single set of reagents and protocols across all applications. This unified approach enables laboratories to seamlessly expand their service menus while ensuring operational consistency.

Economics

Provides the broad coverage needed for general conditions at a fraction of the cost of WES

One Procotol, All Panel Tests

Streamlines operations by using the same laboratory setup, reagents, and bioinformatics pipeline for all CES-based tests

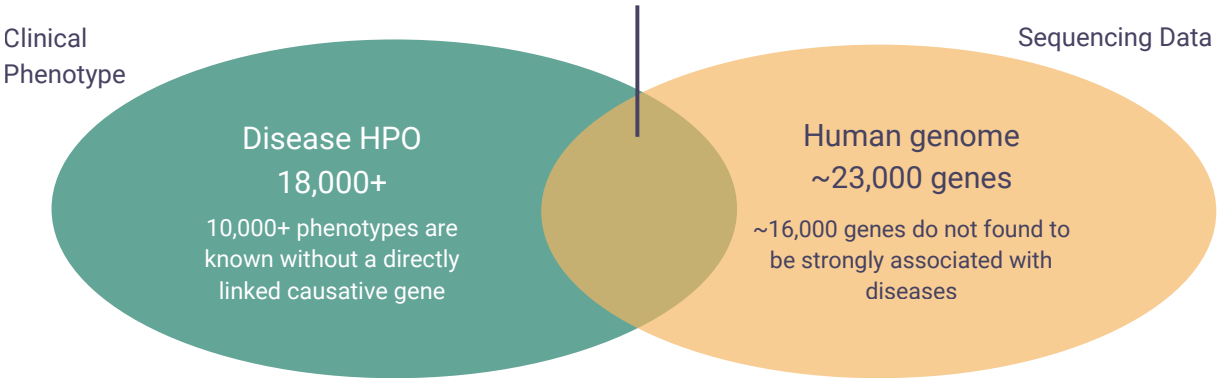
Seamless Scalability

Easily add new panels or modify existing services using our standardized, scalable framework

CES Range

Focuses on definitive disease-associated genes

7,000+ phenotypes associated with 4,000+ genes (OMIM)



Day 1
- gDNA extraction
- Pre-capture library
- Overnight hybrid capture

Day 2
- Post-capture library
- Pooling
- Library QC

Day 3
- Sequencing
- WESisi Data analysis

Day 4
- Variant interpretation and reporting

Day 5
- Report review
- Report issuing

CES Capture Technology

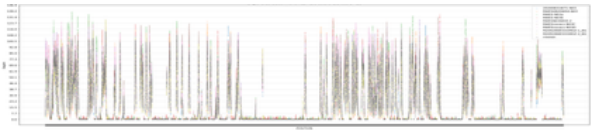
eWES™ Probe Design

Focus on genes with clear relationship to the clinical phenotype and supported by sufficient evidence, thereby reducing the complexity of interpretation and enhancing cost efficiency.

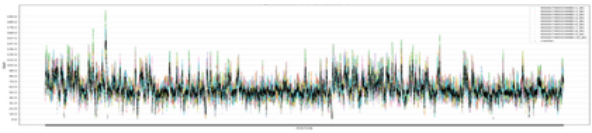
Number of genes	Targeted region size (bp)	Probe targeted region size (bp)	Total number of probe	Type of probe	Number of probe
4,773	13,060,162	16,127,338	161,434	1x Capture probe	155,941
				2x Capture probe (*DMD* gene)	2,307
				1x Capture probe (10-50bp pathogenic InDels)	5,493

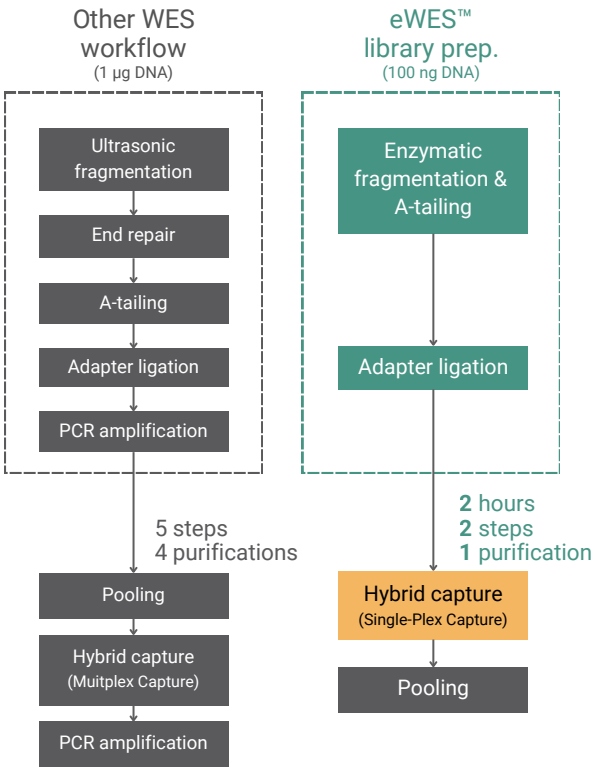
Unlike standard probes that struggle with difficult regions, eWES™ probes are optimized for capture efficiency in some complex areas.

DMD coverage (before optimization)



DMD coverage (after optimzation)



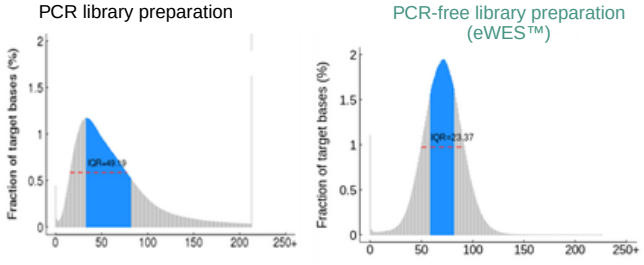


PCR-free eWES™ Library Preperation

Utilizing our PCR-free library preparation based on eWES™ technology can reduce the biases and base mismatches associated with PCR amplification significantly.

This approach enhances the uniformity of coverage and increases the sensitivity and specificity of detection, optimizing test performance.

Sequencing depth distribution



Smaller Interquartile Range (IQR) values indicates more uniform fragment lengths, leading to improved coverage consistency, reduced sequencing bias, and enhanced accuracy in variant calling

Parameters	Description
Technology	Targeted sequencing, paired-end
Platform	illumina or Salus SBS sequencing system
Sample type	gDNA from blood, CVS, amniotic fluid, cord blood/tissue, product of conception, etc.
Targets	SNVs, InDels (<50bp), and exon-CNVs in exons and splice sites ±5 bp of 4,773 genes
Performance	>99% sensitivity and precision for SNVs, reduced for InDels and CNVs

Appendix: Reference CES Panels from Berry Genomics Clinical Labs

(Fully adaptable for in-house laboratory integration, and large-scale custom projects)

Expanded Carrier Screening (ECS)

Single-gene disorders with a recessive inheritance pattern often have a high carrier frequency. Carriers typically do not exhibit symptoms but can pass pathogenic mutations to their offspring. SecureCari™ ECS screens for a comprehensive panel of clinically relevant genes simultaneously, facilitating informed family planning and decision-making.

- Flexible Scope: Multiple standard panels and customization options are available to meet specific needs

Panel	Reporting Scope
21-Gene Panel	Detects 4,032 P/ LP variants across 21 genes
51-Gene Panel	Detects 7,343 P/ LP variants across 51 genes
129-Gene Panel	Detects 11,280 P/ LP variants in 129 genes associated with 155 X-linked and autosomal recessive disorders
1766-Gene Panel	Detects variants across 1,766 genes associated with 1,784 autosomal recessive and X-linked recessive disorders (This panel is available only for couple-based testing. We report P/ LP variants only when both partners carry variants in the same gene, or when the female partner carries an X-linked variant. Single-carrier findings for autosomal recessive conditions and VUS are not disclosed in the final report.)

Newborn Genetic Screening

While biochemical newborn screening (NBS) has been widely implemented in most countries, it faces inherent limitations, including false-positives and an inability to detect conditions lacking definitive biochemical markers in the neonatal stage. NBSure™ Newborn Genetic Screening complements traditional biochemical assays by utilizing NGS technology. It significantly expands the screening scope to include relatively high-incidence conditions that are clinically actionable in early childhood, allowing for timely management and treatment.

- Expanded Scope: Screens for conditions undetectable by biochemical markers (e.g., SCID, SMA)
- High Specificity: Reduces false-positives caused by transient physiological factors
- Late-Onset Detection: Identifies variants associated with conditions presenting later

Panel	Reporting Scope
Jaundice Panel	Detects P/ LP variants in 82 genes associated with single-gene disorders presenting with a jaundice phenotype
Inherited Metabolic Disorders Panel	Detects P/ LP variants in 57 genes associated with 72 inherited metabolic diseases
358-Gene Panel	Detects P/ LP variants across 358 genes associated with 411 single-gene disorders
737-Gene Panel	Detects P/ LP variants across 737 genes associated with 1,287 single-gene disorders

Note: Carrier status for autosomal recessive and X-linked recessive conditions is not reported.

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