

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 Protocol, All Panel Tests

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

Seamless Scalability

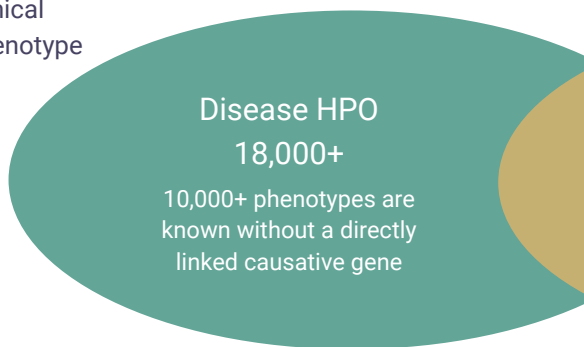
Easily add new panels or modify existing services using our standardized, scalable framework

CES Range

Focuses on definitive disease-associated genes

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

Clinical
Phenotype



Sequencing Data



- 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

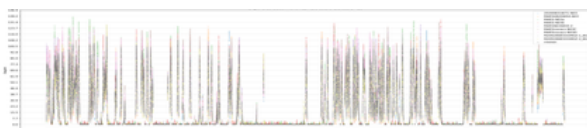
eWES™ Probe Design

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.

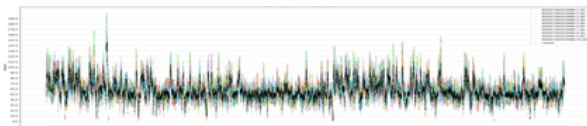
Number of genes	Targeted region size (bp)	Probe targeted region size (bp)	Total number of probe	Type of probe	Number of probe
4,773	13,060,162	16,127,338	161,434	1x Capture probe	155,941
				2x Capture probe (<i>DMD</i> gene)	2,307
				1x Capture probe (10-50bp pathogenic InDels)	5,493

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

DMD coverage (before optimization)



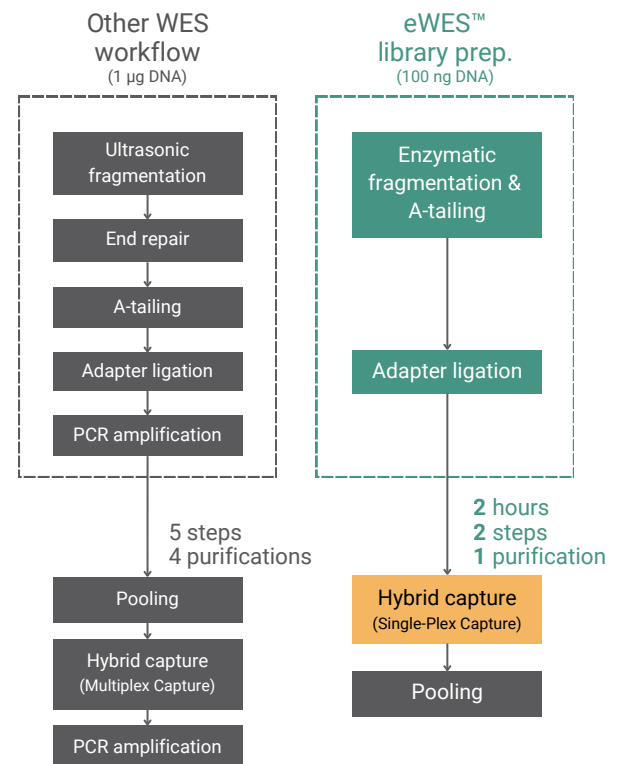
DMD coverage (after optimization)



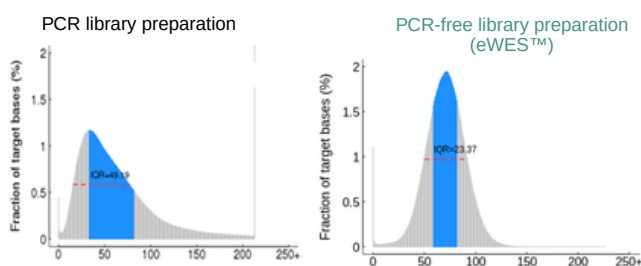
PCR-free eWES™ Library preparation

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

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



SecureCari™
Product Page

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 that are 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



NBSure™
Product Page



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