Blueprint Genetics' Whole Exome Sequencing (WES)

Blueprint Genetics' exome offers a high-quality, transparent option when choosing a diagnostic test for your patient

What is included?

- Consistently uniform sequencing depth across all protein-coding genes and selected non-coding regions
- Improved coverage of challenging genes (e.g. *PKD1*, *RPGR*-ORF 15, *GBA*)
- ~1,500 clinically relevant non-coding variants

Quality metrics

- Mean sequencing depth of 174x with ≥99.4% of base pairs (bp) covered at ≥20x
- 99.7% sensitivity and >99.99% specificity for SNV detection
- 97% sensitivity and >99.99% specificity for indel detection (up to 220 bp)
- Best-in-class sensitivity for detecting copy number variations (CNV), with >92% sensitivity to detect single-exon level deletions and >99% sensitivity to detect 5 exon CNVs

What does this mean for your patient?
A more comprehensive test.

WES is most suitable for individuals with:

- Complex phenotypes with multiple differential diagnoses
- Genetically heterogeneous disorders
- A genetic disorder is suspected, but a smaller, more focused panel is not available
- Inconclusive previous genetic testing

	Blueprint Genetics	Lab A	Lab B	Lab C
% coverage of target region	99.7% >20x **	95% >10x	97% >20x	97.5% >10x
# bp covered <20x (or <10x)*	60,000 bp <20x	1,500,000 bp <10x	900,000 bp <20x	750,000 bp <10x
# exons/genes covered <20x (or <10x)*	414 exons 45 genes	10,345 exons 1,119 genes	6,207 exons 672 genes	5,100 exons 550 genes

*Estimates intended for illustrative purposes

**% coverage of target region for production samples

We offer two types of Whole Exome Sequencing

Whole Exome Plus

High-quality Whole Exome Sequence analysis and interpretation for index patients.

Whole Exome Family Plus

High-quality Whole Exome Sequence analysis and interpretation of an index patient and parents (trio), or other family members. The trio approach improves the diagnostic rate by facilitating sequence variant analysis, segregation analysis, and identification of *de novo* variants.

For more information please visit our website or contact customer support:

www.blueprintgenetics.com
(650) 452-9340
support.us@blueprintgenetics.com

Expansion services

Any panel can be expanded to Whole Exome or Whole Exome Family Plus tests.

Blueprint Genetics' comprehensive diagnostic service provides a faster path to informed decisions about medical management and improved patient outcomes:

1. Sample

We accept blood, saliva, and isolated DNA samples. Ordering is simple; either through our online secure portal Nucleus or a paper requisition.

2. Sequencing

High-quality customized exome capture technology and NGS methods result in deep and uniform, clinical-grade WES data.

3. Analysis and interpretation

A proprietary, automated bioinformatics pipeline produces rapid and reliable clinically relevant information from the sequencing data. A genotype-first approach to the analysis increases the diagnostic yield by allowing for unexpected or atypical presentations of a disorder.

4. Clinical statement

A comprehensive clinical statement is generated by a team of geneticists and specialized clinicians who interpret the results using the latest publications and databases. Ordering clinicians have full visibility to their individual patient's test performance and quality metrics.

We are continuously developing our services and offering. We may amend service descriptions from time to time by posting new versions on our website. For up-to-date information, please visit blueprintgenetics.com.

Blueprint Genetics