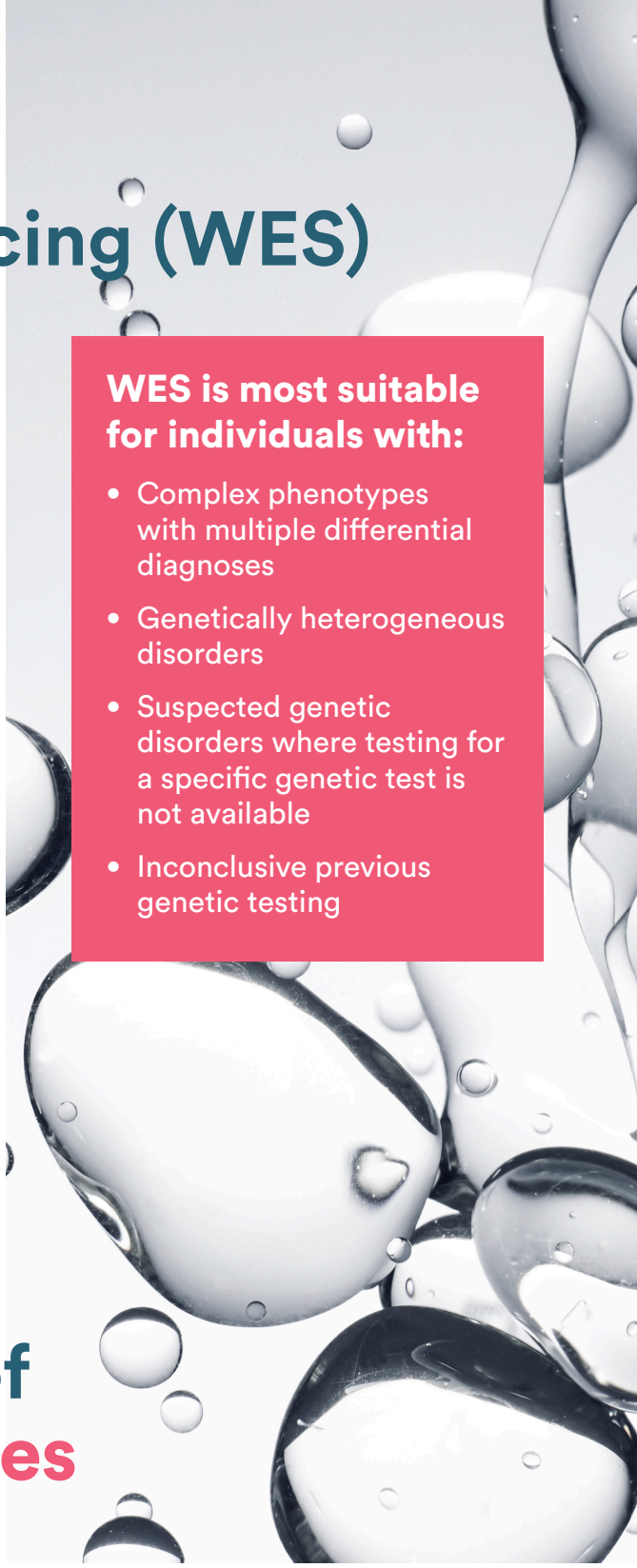
Blueprint Genetics' Whole Exome Sequencing (WES)

Blueprint Genetics offers a high-quality, innovative, and transparent WES diagnostic approach for healthcare professionals.

- Highly uniform sequencing depth across all protein-coding genes and selected non-coding regions
- Mean sequencing coverage of 174x, and ≥99.4% of base pairs (bp) covered at ≥20x
- 99.7% sensitivity and >99.99% specificity for SNV detection and 97% sensitivity and >99.99% specificity for indel detection (up to 220 bp)
- Best-in-class sensitivity for detecting copy number variations (CNV), with 93% sensitivity to detect 1 exon del/dup and >99% sensitivity to detect 5 exon del/dup
- Our clinical WES is tailored to improve coverage for challenging genes (e.g. *PKD1, RPGR* [ORF 15], *GBA*) and identify ~1,200 clinically relevant non-coding variants



WES is most suitable for individuals with:

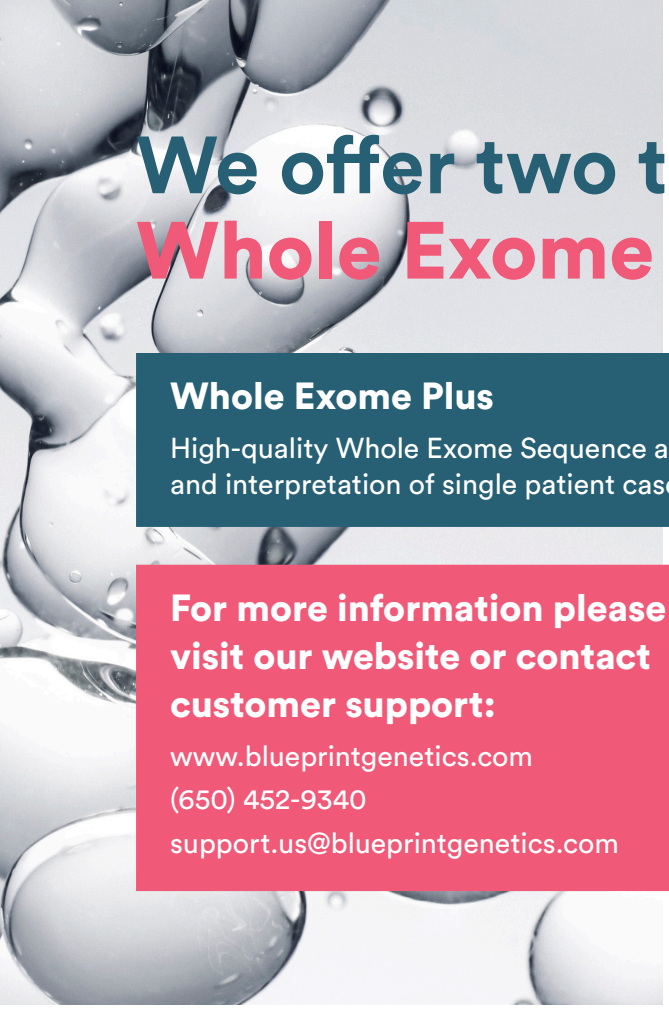
- Complex phenotypes with multiple differential diagnoses
- Genetically heterogeneous disorders
- Suspected genetic disorders where testing for a specific genetic test is not available
- Inconclusive previous genetic testing

Maximized coverage of clinically relevant genes

Join the conversation #GeneticKnowledge

www.blueprintgenetics.com

Blueprint Genetics



We offer two types of Whole Exome Sequencing

Blueprint Genetics

Whole Exome Plus

High-quality Whole Exome Sequence analysis and interpretation of single patient cases.

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 in WES 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

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

1. Sample

We accept blood, saliva, and isolated DNA samples. Simplified test requisition through our secure portal, Nucleus, or by paper.

2. Sequencing

High-quality exome capture technology and NGS methods to obtain deep and uniform, clinical-grade WES data.

3. Analysis and interpretation

Proprietary automated bioinformatics processes to rapidly and reliably produce clinically relevant information from the sequencing data.

4. Clinical statement

A team of geneticists and specialized clinicians interpret the results utilizing information from the latest publications and databases to produce a comprehensive clinical statement. Through our online portal, clinicians have full transparent access to their individual patients' performance and quality data. Data return in various formats (BAM, FASTQ, and VCF) available for a fee upon request.