8 Reasons to Choose Blueprint Genetics

Maximized diagnostic potential with high-performing next generation sequencing (NGS) platform

All our diagnostic tests—regardless of whether you are ordering a single gene, a panel or WES—are performed on a high performing platform enabling flexibility. Comprehensive, analytic validation of our platform is available on our website.

Customizable NGS panels with easy expansion to WES

Expand to Whole Exome Sequencing from any panel or single gene test. Customize your panel by adding or removing genes at no additional cost.

Exceptional combination of coverage >99.4% at depth >20x

Mean sequencing coverage of >174x offers highly uniform sequencing depth across all protein-coding genes.

Comprehensive testing including non-coding variants

Our testing covers ~2,000 clinically relevant non-coding disease-causing variants.

High-resolution NGS-based CNV detection included in all tests

CNVs are an important disease mechanism that should be evaluated in all patients with a suspected inherited disorder. Our platform offers best-in-class sensitivity for detecting CNVs, with >92% sensitivity to detect single-exon level deletions and >99% sensitivity to detect 5 exon CNVs.

Clear and meaningful clinical report

Our interpretation team goes the extra mile to provide comprehensive variant classification for your patient.

Affordable pricing with patient-friendly billing solutions

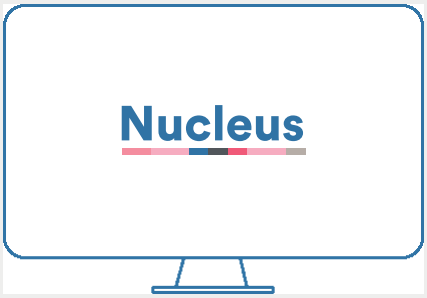
Our billing experts, clinical genetics support, and client services team are committed to helping you when you need them.

A wide range of additional services to support your patient's diagnostic journey

We offer additional services from Familial Variant Testing and Variant of Uncertain Significance (VUS) Clarification Service to Re-evaluation of WES variants.

Nucleus: Easy ordering platform

Place orders, track progress, and read results with our online ordering portal.



Sign in or register for Nucleus at nucleus.blueprintgenetics.com

Blueprint Genetics

Blueprint Genetics Products and Services

Diagnostic Panels

- Over 220 panels covering 15 different medical specialties.
- All panels are filtered from our high-quality sequencing assay.
- All panels include both Sequence and Deletion/Duplication (CNV) Analysis.

Whole Exome Sequencing

- Whole Exome Plus includes high-quality sequence analysis of single patient cases, coupled with Deletion/Duplication (CNV) analysis.
- Also available as Whole Exome Family Plus for index patient and parents or other family members.

Mitochondrial DNA testing

- Our high-quality mtDNA testing includes both sequencing and copy number variant (CNV) analysis for all the 37 mtDNA genes.
- The entire mitochondrial genome is included in a selection of our panels.

Open access testing for Inherited Retinal Degeneration (IRD)

- The My Retina Tracker Program provides individuals living in the United States with a clinical diagnosis of an IRD access to no-cost genetic testing and genetic counseling.
- This program was created in collaboration with the Foundation Fighting Blindness and InformedDNA.

Single Gene Tests

- ~3,900 single genes available for testing, all of which include sequencing and Deletion/Duplication (CNV) analysis.

Customization of all panels

- Advanced flexibility to customize any panel by adding up to 200 genes from a selection of over 4,000 clinically relevant genes at no additional cost.
- Ability to add the entire mitochondrial genome or individual mtDNA genes on any panel.
- Name and save your custom panels in our ordering platform, Nucleus.

Expand to Exome

- Expand to Whole Exome Sequencing is available from any single gene, panel, or customized panel in all of our 15 different medical specialties.

Variant Specific Testing

- Blueprint Genetics offers Familial Variant Testing for family members of an index patient or Targeted Variant Testing for confirmation of research findings or variant classification purposes.
- **Five Free Familial Variant Tests** service is available for blood relatives of a proband who had panel test performed at Blueprint Genetics. To ensure that your patient is eligible for the service, please visit **blueprintgenetics.com** for more information.

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.

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