An innovative approach to genetic testing for improved patient care

2017

Blueprint Genetics

Blueprint Genetics is changing diagnostics by providing fast, affordable and comprehensive genetic knowledge

Who we are

Blueprint Genetics is a genetic knowledge company that provides comprehensive genetic diagnostics for rare inherited diseases. Our proprietary technologies deliver enhanced quality and performance with lower cost and significantly faster lead time.

We believe that transparency in genetic diagnostics is essential when selecting an optimal genetic test for a patient. Therefore, we offer full access to our methodology, sensitivity, and performance, as well to analytic validations of our testing platform. Blueprint Genetics represents the most transparent genetic diagnostic laboratory in the market.

We have invested in rigorous analytic validation of our diagnostic tests. We deliver diagnostic solutions that are improving the clinical standards of precision medicine and enriching patient lives.

Our mission

Our mission is to support healthcare professionals around the world in providing the best care for patients and families with rare inherited diseases.

Our promise

We continually strive to develop and provide clinicians, and their patients, with the most inclusive tools and resources for genetic diagnostics. By removing access barriers for clinicians, we create possibilities for targeted treatment and individualized care for patients.

Clinically actionable test design and services

Maximizing diagnostic yield
Blueprint Genetics' diagnostic panels have been carefully compiled to maximize their impact in clinical practice. Our sequencing analysis covers all coding exons based on the widest possible gene models.

Rapid turnaround time
Test results are provided in 21 days to facilitate effective clinical workflow.

Easy ordering
Test requisitions can be sent quickly through our online portal, Nucleus, or by mail.

Wide variety of accepted sample types
Our tests have been validated for a variety of specimen types, including blood, saliva, and DNA.

High-quality sequencing
Our analysis uncovers both smaller and larger changes in DNA. SNPs and small INDELs are detected with our proprietary targeted sequencing method, OS-Seq™, developed at Stanford University and published in Nature.

Analysis and interpretation
We use our own proprietary automated bioinformatics process, including the IBM Watson powered CLINT platform, to quickly and reliably produce clinically relevant information from sequencing data.

Clinical statement
Comprehensive clinical statements produced by our experts support the diagnosis of the patient, and help physicians to make more confident decisions. The reports fulfill all requirements provided in the ISO 15189, CAP, and CLIA standards.

Variant classification and follow-up service
Our variant classification scheme closely follows industry standards (ACMG 2015). Our follow-up report service ensures that all re- classified variants will be reported.

Commitment to the genetic diagnostics community
We are dedicated to advancing and improving the field of genetic diagnostics through transparency and public variant sharing.

Our test menu

Sequencing and Del/Dup (CNV) panels

Our Sequence Analysis is based on the proprietary targeted sequencing method OS-Seq™. Our analysis pipeline has been validated for detection of single nucleotide variants (SNVs), insertions and deletions (INDELs), and copy number variations (CNVs). Our offering includes over 200 panels in the following disease categories.

Cardiology
Dermatology
Ear, Nose & Throat
Endocrinology
Gastroenterology
Hematology
Hereditary Cancer
Immunology
Malformations
Metabolic Disorders
Nephrology
Neurology
Ophthalmology
Pulmonolgy

Whole Exome Sequencing

Whole Exome Sequencing (WES) provides an effective discovery approach to diagnostics in a large variety of complex genetic disorders. WES is combined with a highly optimized data analysis pipeline and applied for efficient detection of SNVs and INDELs.

Whole Exome
Whole Exome Family
Whole Exome Plus
Whole Exome Family Plus

Results in 6 to 9 weeks.
*(*available soon in the U.S*)*

Family Member Testing

Direct Sanger sequencing of PCR amplicons is applied to confirm SNVs and INDELs as well as for screening of disease-causing mutations in family members.

Results in 21 days

Quality metrics and acceptance for clinical testing

Analysis technology	Quality metric	Acceptance criteria	Actual*
Sequencing panel	*Test sample coverage (>15x)* *Reference sample sensitivity*	*>98%* *>98%*	*99.4%* *99.3%*
Sequencing panel	*Test sample coverage (>15x)*	*>98%*	*99.3%*
Whole Exome Sequencing	*Test sample coverage (average)*	*>100x*	*158x*

** 3 month average*

How to order

Providing you with actionable genetic knowledge

1

Order in one of three ways:

Online: Nucleus portal at nucleus.blueprintgenetics.com

PDF: download the form from our website, then fill it in and mail it to us.

Sample collection kit: order a specimen kit from our website. It includes a sample tube, requisition form, and prepaid return shipping.

2

Sample shipping

Blueprint Genetics / Samples
1250 Missouri Street, #208
San Francisco, CA 94107
United States

3

Analysis and interpretation

Our process takes 21 days (10 days express) and includes:

- DNA sequencing
- Advanced bioinformatics analysis
- Comprehensive clinical interpretation

4

Rapid access to results

- Login to Nucleus to review your results online.
- On request, we can also deliver your results by mail or fax.

Nucleus: Easy ordering & networking platform

Order & follow-up

Nucleus is designed to be as simple and user-friendly as possible, and offers support along the way. You can place an order easily by filling in the requisition form and uploading clinical documents. By logging in, you can follow the progress of your order. The progress bar clearly shows the current stage of every order, which makes dealing with multiple orders very easy. The notification bar will alert you whenever results are ready, updated, or if there is a new suggested connection.

Read & share results

We will send you a notification when results are available. Results can easily be viewed online, printed, or shared in a safe and secure way with a colleague treating the same patient.

Support

We will guide you through the whole diagnostic process. In Nucleus, you can browse frequently asked questions by topic. You can also reach our Clinical Genetics Support team when specialized expertise in genetic diagnostics is needed.

nucleus.blueprintgenetics.com

Connect with clinicians

Nucleus offers a unique possibility for clinicians to become connected with their colleagues from all over the world, that have patients with matching variants.

A connection is suggested each time a patient has a rare genetic variant classified as a VUS (Variants of uncertain significance) or Likely Pathogenic. With this service we want to provide the possibility to share knowledge and phenotypes regarding rare inherited diseases.

Each suggested connection can be either accepted or rejected. When both clinicians have accepted the suggestion, you will gain access to the contact information you have chosen to share.

We are here to support you

Customer Support

Our global Customer Support team can help you answer questions on a variety of topics:

- Placing orders
- Usage of Nucleus
- Billing
- Ordering sample collection kits
- Qrdering options
- Sample requirements
- Status of your patients' analyses

For support please contact:

Europe, Middle East, Australia, Asia:
support@blueprintgenetics.com
+358 40 2511 372

North and South America:
support.us@blueprintgenetics.com
+1 650 452 9340

Our website www.blueprintgenetics.com also offers a wide range of FAQ's.

Clinical Genetics Support

Our expert team of geneticists, physicians, clinical consultants, and genetic councelors is available for:

- Discussions on selecting the optimal diagnostic tools for your patient.
- Questions concerning clinical statements
- Discussions on variant classification
- Assistance in genetic counselling of your patient and their family
- Advice on how to improve service and quality of diagnostics.
- Tailor-made service options for your specific clinical needs

For support please contact:

genetics.support@blueprintgenetics.com.

Our result interpretation team consists of internationally recognized experts in the field of human genetics and clinical medicine. The average number of peer reviewed publications per team member is 55. This level of experience guarantees an unmatched quality of interpretation. Similarly, Blueprint Genetics has a team of clinical consultants with expertise covering all medical specialties. The clinical consultation team consists of 4 professors and 10 assistant professors from top universities. All consultants are MD, PhD and have specialist degree in their medical field of expertise. Get to know the team at blueprintgenetics.com

Testing with Blueprint

Cost-efficiency with high quality

Our innovative pipeline – from easy ordering to unique sequencing technologies and top- quality medical statements – enables affordable genetic testing without ever compromising the quality.

Transparency

We offer our customers access to quality data in every single patient analysis

Comprehensive and flexible diagnostics

Our platform has demonstrated its ability to detect a variety of different mutations, including SNVs, INDELs, and CNVs. In addition, our panels can be modified based on individual needs.

Clinical genetics support today and in the future

With our team of geneticists, physicians, and clinical consultants, we offer highly specialized expertize in genetic diagnostics. Furthermore, we continuously update existing results and send follow-up reports when new data leads to variant re-classification.

Blueprint Genetics

Read more about us, our services, and customer support at blueprintgenetics.com. Contact us with any questions, we're here to help!

Join the conversation
#GeneticKnowledge

www.blueprintgenetics.com