A clear path to early treatment

Through cost-effective, actionable genetic diagnostics

Blueprint Genetics

Accessible genetic diagnostics

In recent decades, genetic testing has helped bring a diagnosis to millions of people with inherited diseases and their affected family members. With a conclusive diagnosis, clinicians can start the right course of treatment for their patients, or recommend the necessary lifestyle changes to ensure their patients live as comfortably and as long as possible. However, the time and expense needed to provide results through traditional genetic testing methods has meant that genetic diagnostics are often used as a last resort. Now, a revolution in genetic testing technology means that patients can get their results in a fraction of the time—and at a fraction of the cost.

Comprehensive genetic knowledge

Identifying rare genetic disorders using conventional diagnostics is a process of trial and error that often yields no definitive answers.

Inherited disorders are found in approximately 5% of the world's population, or around 350 million people. To date, over 7,500 different genetic diseases have been identified, and 3,500 have a known molecular genetic background, with this number growing at an ever-increasing pace.

Caring for these disorders accounts for approximately 17%
of healthcare costs. Their impact on the lives of patients and their families is indisputable, while they also present some unique challenges for health care professionals. Although the number of these diseases is sizeable, many are so rare that they're unknown to the vast majority of clinicians. Identifying them using conventional diagnostics is a process of trial and error that often yields no definitive answers.

Anything we can do to improve and speed up diagnosis not only allows us to plan the best and most cost-effective treatments, but also plays an important part in patient comfort and long-term prognosis. This is where genetic testing can truly make a difference.

Why do genetic testing?

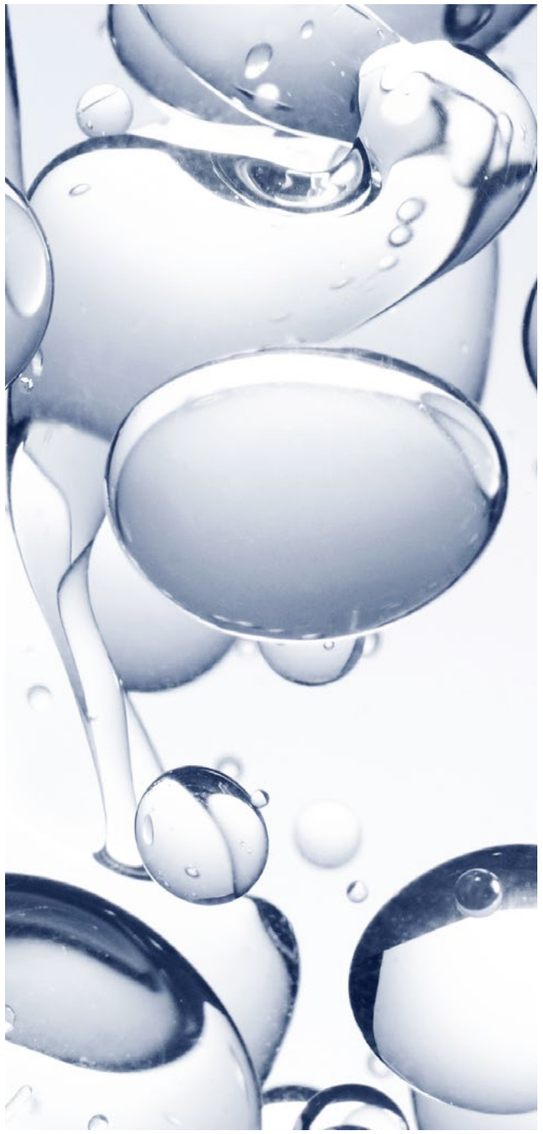
When a mutation is identified, you can make solid, accurate recommendations for your patient's treatment and clinical monitoring, as well as advise them on lifestyle choices to improve their quality
of life and longevity.

As a clinician, genetic testing can help you:

- Diagnose diseases and/or their severity
- Improve your patient's care by identifying the specific gene mutations that cause an already diagnosed disease
- Personalize your patient's care by choosing the best treatments
- Identify mutations which may pose a risk for developing a disease, or could be passed on to children

When a mutation is identified, you can make solid, accurate recommendations for your patient's treatment and clinical monitoring, as well as advise them on lifestyle choices to improve their quality of life and longevity. Close relatives of affected patients can also be screened to determine if they are at risk of developing the disease, allowing you to plan pre-emptive measures to mitigate their risks, and perhaps even prevent the onset of the disease entirely.

Moreover, identifying the specific gene mutation can have direct significance for the drugs used to treat the disease. In the case of mutations that inhibit the production of essential enzymes, for example, choosing medicines that compensate for the lack of these enzymes is critical. Drug dosages of any medication can also be tailored for an individual patient's metabolism, as the technology exists to sequence genes for this purpose as well.



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The drawbacks of traditional genetic testing

Traditional genetic testing requires time-consuming manual lab work, meaning a long delay before the results are available.

Genetic testing is becoming a standard diagnostic procedure in several fields of medicine. But because it has traditionally been slow and expensive, with a lot of negative results, it is typically chosen as a secondary diagnostic tool. Traditional genetic testing requires intensive manual lab work, which stacks high labor costs on top of the already expensive equipment needed. The time-consuming process also means a long delay before the results are available, and more time needed to refamiliarize yourself with each individual case.

Many of these intensive laboratory processes are based on Sanger sequencing technology that can only analyze one gene at a time. This means you must first make an educated guess as to which gene you should order a test for, then, after waiting for the result, the process must be started again if the results for the mutation are negative.

In addition, the answers provided through traditional genetic testing are generally reported to you as simply positive or negative. Because these diseases are so rare, interpreting the results and searching for the right treatment may be taxing on your time and resources. Compounding this, the field of genetics is advancing incredibly quickly and the amount of known genetic information is ever-expanding, making it difficult for most clinicians to keep-up-to-date with genetic diseases.

Comprehensive, rapid, and high-quality genetic diagnostics solutions

Blueprint Genetics provides high-coverage panels, high-resolution CNV detection, and comprehensive coverage of clinically relevant noncoding variants

Current state-of-the-art next-generation sequencing (NGS) technology enables rapid and cost-effective diagnostics of rare diseases. Many times genetic testing is the only solution to avoid a never ending diagnostic odyssey.

About 2% of the genome is actually encoding genes and only a subset of genes are known to be medically relevant. Blueprint Genetics' sequencing platform focuses on that portion of the genome that is clinically relevant, reducing the complexity of the sequencing process.

High-quality exome capture is performed using an in-house-designed WES platform (xGen Exome Research Panel with custom-designed capture probes, IDT) and the Illumina NovaSeq sequencing system to obtain deep and uniform sequencing data from coding exons, exon-intron boundary, and over 1,500 selected noncoding deep intronic variants. Our careful oligo design, validated laboratory process and quality control, clinical-grade sequencing coverage, and proprietary bioinformatic pipeline enable high quality for detecting single-nucleotide variants (SNVs), insertions and deletions (indels), and copy number aberrations. With improved sequencing coverage and sensitivity to detect clinically relevant mutations, we can provide higher diagnostic yield.

High-quality NGS sequencing:

- Mean sequencing coverage 174x, and ≥99.4% of base pairs (bp) covered at ≥20x
- 99.9% accuracy, 99.7% sensitivity, and 99.9% specificity for detecting SNVs
- 97% sensitivity for 1-50 bp indels

Blueprint Genetics uses custom whole exome sequencing assay and Illumina NovaSeq technology to offer you over 220 tests covering all medical specialties, and to deliver results within 28 days.

Making sense of it all

Of course, faster results still need to be supplied with enough context to be understood. Clinicians have a need for high-quality analysis and interpretation of results. Once sequencing is complete, the process of genetic data analysis begins. This requires an intelligent computing infrastructure with state-of-the-art algorithms for quality control, alignment with high-quality reads with a human reference genome, and variant calling.

To aid the process of variant interpretation, the sequencing results are matched against a comprehensive database of disease-related mutations —collected and curated in-house by a team of geneticists. A groundbreaking bioinformatics pipeline compiles information from more than 2,500 scientific publications and publicly available databases.

More than just results

A quicker and more confident diagnosis, less time and money wasted cycling through various diagnostic screening methods, and, ultimately, faster paths to treatment for your patients.

A team of geneticists and clinicians prepares a comprehensive clinical statement from the sequence analysis, assisted by our Clinical Interpretation Platform. The platform allows our team to instantly query millions of genetic and medical sources. The result is actionable clinical statements from our genetic and medical experts, complete with the patient history, all the relevant test findings, interpretation of the results, and gene-specific information and research citations.

This not only benefits specialized clinicians, it also makes genetic testing more accessible for general practitioners as well, allowing more time for patient treatment and care, and less time chasing and understanding results.

What may have previously taken months or even years to diagnose using traditional diagnostic tools and genetic testing methods now takes just 3 weeks. This means a quicker and more confident diagnosis, less time and money wasted cycling through various diagnostic screening methods, and, ultimately, faster paths to treatment for your patients.

A better life for millions

Early diagnosis through genetic testing means lives saved, as well as improved quality of life for hundreds of millions of people around the world.

This is an exciting time in genetic research, with each new year bringing us colossal growth in the amount of genetic information known to humankind. We are at the dawn of a new era of genetic testing, one that is faster and cheaper, that enables higher sequencing performance and earlier diagnosis of more diseases, while at the same time driving more personalized patient care. The faster turnaround time, lower cost, and actionable clinical statements made possible by innovative methods used at Blueprint Genetics make genetic testing more accessible earlier in the patient care journey. These methods save time and money, but most importantly, provide the necessary information to start the right treatments as quickly as possible. Early diagnosis means lives saved, as well as improved quality of life for hundreds of millions of people around the world.

Blueprint Genetics provides the most cost-effective, rapid, and actionable genetic diagnostics to clinicians available today. For more information about how our

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.

Our process makes genetic testing more accessible earlier in the patient care journey.

Blueprint Genetics

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