Quest Diagnostics™

From precision diagnostics to precision medicine to **precision health**

Leveraging diagnostic technology and innovation beyond treatment—toward precision health

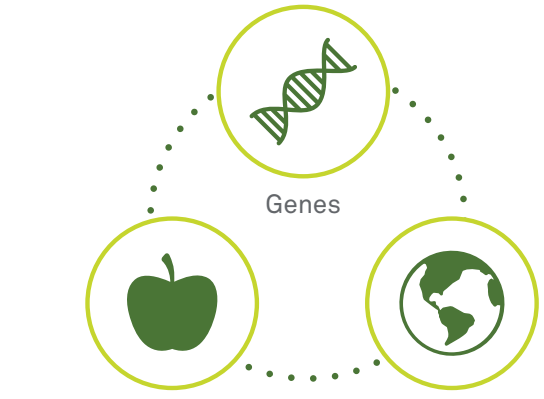
More than just the latest industry buzzword, precision medicine may very well be the future of healthcare—and health. The National Institutes of Health (NIH) defines precision medicine as "an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person."[1]

Though it has become more mainstream in the past few years, precision medicine—or the concepts behind it—has been around for decades.[2] What we now call precision medicine has evolved, from the NIH's Human Genome Project to diagnosing and treating diseases to, eventually, disease prevention, or what many now refer to as precision health.[3]

This evolution, or revolution, requires broader, big-picture thinking and loftier goals. Beyond genome sequencing—generally thought to be synonymous with precision medicine—a wide array of solutions, services, and technologies will have a role to play, including advanced therapies, mobile health, eHealth, patient engagement efforts, big data, and analytics.[2]

Advancing the industry from precision medicine to precision health will also require dedicated efforts from a number of key players. Scientists, policymakers, and healthcare leaders are all needed to lead the charge. They must work together to conduct research, establish new systems and infrastructures, and share data and information. As a result, they will provide clinicians with the science, data, and tools they need to prevent disease, improve health outcomes, and promote healthy living for all patients.

Advancing precision medicine



Leaders
- Scientists
- Policymakers
- Healthcare leaders
- Clinicians

Solutions/Services
- Advanced therapies
- Mobile health
- Patient engagement
- Big data

Technologies
- eHealth
- Analytics

Precision medicine: early origins

Despite its recent media attention, the application of genetics to medicine is not new. As early as 1902, British physician Sir Archibald Garrod suggested that genetic factors direct chemical transformations in humans and underlie individual variability.[4] And the term "pharmacogenetics" was first used in 1959 by German geneticist Friedrich Vogel, who had seen a relationship between adverse drug reactions and genetically determined variation.[4]

A few years prior, in 1953, the double-helical structure of DNA was discovered, leading to the development of the field of molecular biology.[3] However, it took many years for scientists to turn their attention toward genomics, and in the U.S., it wasn't until the 1980s that the "outlandish idea of sequencing the human genome began to create a stir."[3]

It created such a stir that the National Research Council (NRC) established its Committee on Mapping and Sequencing the Human Genome and issued its report of the same name in 1988—launching the far-reaching, industry- and world-changing, 13-year Human Genome Project.

The results of this project laid the foundation for what we now know as precision medicine. By essentially mapping the entire human genome, the project led to rapid advances in the diagnosis and treatment of cancer, and genomic analyses began to provide answers or new therapeutic approaches to challenging clinical problems.[5]

Precision medicine as we now know it

After the completion of the Human Genome Project, the NRC realized that large-scale sequencing of individual human genomes was going to become practical and established a committee to develop a new report.[3] Published in 2011, *Toward Precision Medicine* coined the term "precision medicine" and initiated more mainstream use and understanding of the term.[3]

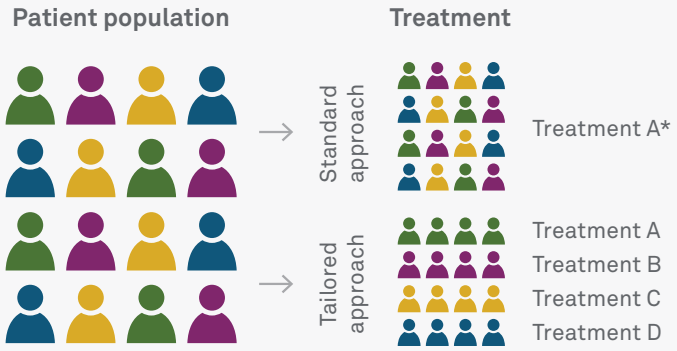
In 2015, President Obama launched the Precision Medicine Initiative, a research effort to revolutionize how the U.S. improves health and treats disease.[6] Participating healthcare provider organizations include regional and national medical centers (such as Cedars-Sinai Medical Center [CA], Columbia University Medical Center [NY], Partners HealthCare System [MA], and Banner Health [AZ]), community health centers (such as Cherokee Health Systems [TN] and Eau Claire Cooperative Health Center [SC]), and medical centers operated by the U.S. Department of Veterans Affairs.[7]

The initiative includes the following objectives:[6]

- More and better treatments for cancer—via the National Cancer Moonshot
- Creation of a voluntary national research cohort—the All of Us Research Program
- Commitment to protecting privacy
- Regulatory modernization
- Public-private partnerships

As a result of these combined efforts, academic medical centers, community hospitals, and health systems have used precision diagnostics and precision medicine to improve patient health in a variety of ways.

- **Pharmacogenomics**—enables clinicians to find the right treatment for the right patient at the right time; studies have shown that the use of combinatorial pharmacogenomic testing can reduce costs and increase patient adherence[8]
- **Companion diagnostics**—these genetic tests enable targeted drug therapies, assessing the likely utility or effectiveness of the treatment[9]
- **Cancer treatment and genomics**—advances in genomics have facilitated research on the biology of cancer cells, leading to the discovery of potential diagnostic markers and therapeutic targets, while a new generation of clinical trials has emerged to translate basic discoveries into new diagnostic tests and targeted therapies[10]
- **Molecular diagnostics**—molecular testing can now be used to identify and diagnose diseases from Lyme to HIV infections to tuberculosis
- **The study of microbiomes**—and their connection to complex diseases, like obesity, vascular disease, and autism[11]
- **Noninvasive prenatal testing**—allows for first-trimester screening, via blood draw, for chromosomal conditions



*Treatment A is effective in 20% of target population; 80% is waste

Pharmacogenomics: a foundation for precision medicine[12]

Pharmacogenomics is a good use case for precision medicine because the pathways of drug disposition and response are usually known, variation within the genes involved is common, and genetic testing is possible.[13]

None of these advancements in precision medicine would have been possible without the technology to support them. To bring massive amounts of genomic data closer to the point of care and make it actionable, for example, many health systems are prioritizing the development of sophisticated bioinformatics infrastructures and partnering with leading technology solutions companies, from Google to IBM.

Every day, new solutions are being developed. IBM Watson Genomics from Quest Diagnostics, for example, combines state-of-the-art tumor sequencing with powerful cancer data analysis. This can quickly uncover highly personalized cancer treatment options—including options that previously may have remained unidentified.

A vision for the future: precision health

As the field has evolved from one that's genome-based to one that's more healthcare and healthcare systems-based, healthcare leaders are looking ahead to what's next.

Beyond precision diagnostics and precision medicine is what many are calling precision health. Stanford Medicine defines precision health as a "focus on helping individuals thrive based on all the factors that are unique to their lives, from their genetics to their environment."[14] Instead of curing disease after the fact, the goal of precision health is to prevent disease before it even appears.

For example, as genetic testing becomes more affordable and widespread, all patients with elevated cholesterol levels could be screened for a common genetic disorder of cholesterol metabolism—giving clinicians a head start on treating affected patients before they suffer complications.[15] While focusing on preventive care and wellness can provide obvious benefits for patients and their health, it can also help reduce healthcare costs.

Eric Dishman, director of the All of Us Research Program at the NIH, explains that for the initiative to be successful—to truly move in the direction of precision health—precision engagement is also essential. He's hired a chief engagement officer to ensure that his research cohort includes people from all walks of life. And he argues that, at some point, "precision primary care will probably be the name of the game."[16]

How health systems can advance precision health

Academic medical centers, community hospitals, and health systems possess the infrastructure, research capabilities, and capital to advance precision health. Doing so also requires a shift in thinking. Like it or not, clinicians and other healthcare leaders must (1) increasingly become genomic medicine and data specialists, assessing large pools of information through the lens of the individual patient, while (2) continuing to form intimate bonds with patients to collect the kind of information that can't be gleaned from a diagnostics test (e.g., patient fears, pains, needs).[14]

Health systems must equip physicians with the right technologies, tools, and information. They can do this by partnering with other organizations to fill gaps in genomics offerings and develop and improve bioinformatics infrastructures to integrate genomic data into clinical practice. Other considerations include changes to the medical curricula, specialist training, and continuing education for current physicians.[17]

To gather the best, most useful data, academic medical centers, in particular, must enroll more Americans from diverse racial, gender, age, and environmental categories in clinical trials. And facilitating and maximizing collaboration and data sharing between academic medical centers, health systems, scientific institutes, biotech companies, and pharmaceutical companies are paramount.

Conclusion

From diagnostics to medicine to health to engagement, precision medicine has come a long way—and will continue to evolve. Dr. Geoffrey Ginsburg, founding director of the Center for Applied Genomics and Precision Medicine at Duke University Medical Center, says, "Precision medicine is a team sport. [It] will spawn forums and encourage participation. Don't be afraid to be part of that dialogue."[2]

In other words, precision medicine is a collective pursuit, leveraging the varying strengths of those who offer infrastructure, capital, and testing capabilities. Collaborations between healthcare entities is necessary for ongoing developments, advancements, and innovation.

In the short term, precision medicine has the power to help clinicians arrive at more targeted therapy, sooner. This translates into improved efficiencies for health systems and better outcomes for patients. And while today precision medicine is helping clinicians realize new therapies and provide hope for generations of patients to come, in the future it may be able to keep more people healthy.

Quest Diagnostics can support health systems in all aspects of **precision medicine**

Quest offers unparalleled scale and a unique value proposition geared for precision medicine, including the following services and solutions:

- Test development, including end-to-end, next-generation sequencing solutions
- Assay feasibility
- Proof of concept and validation
- Regulatory strategy, application development, and submission
- Market access strategies
- Commercial launch

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