Getting to better outcomes through streamlined genetic testing

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No matter how you look at it, genetic testing has arrived.

> 2022 is an inflection point in the development of diagnostics and treatments for rare genetic diseases.[1]

As you think about the role genetic testing will play in your health system, consider these statistics. First, laboratory tests guide as much as 70% of medical decisions.[2] Second, in a recent study by Harvard Medical School of genetic versus environmental causes of disease, nearly 40% of diseases studied had a genetic component.[3] Together, these 2 findings help explain the explosive growth of genetic testing in recent years. But how can a system already facing spiraling costs and complexity stretch to accept genetic testing as a mainstream occurrence? Is it possible to contain the expense? Will the tests be accessible for all? And how will healthcare systems ensure these complex diagnostics are accurately ordered, interpreted, and applied to treatment or prevention approaches?

Finding sustainable ways to unleash the power of genetic testing—allowing it to usher in the era of personalized medicine—will require a system-level approach. An approach that adapts to the rapidly evolving field of genetics. That helps health systems simplify the application of genomic medicine at scale, without creating barriers for underserved patients. And that meets the needs and expectations of physicians ordering the tests.

Genetic testing is critical to saving lives—and money

Breast cancer is one of the most common cancers in women, a leading cause of death, and a major source of expense in the healthcare industry. The current standard of care only covers genetic screening for women diagnosed with breast cancer who meet specific family history and clinical requirements. Yet research has shown that these requirements fail to include about half of people with a "high risk" genetic variation. In the same study, genetic testing for *BRCA1, BRCA2,* and *PALB2* mutations in all cases of breast cancer would be cost-effective in terms of cost per quality-adjusted year of life and, more importantly, could prevent more than 10,000 cases of breast and ovarian cancers, and over 3,000 deaths per year in the US and UK combined.[6]

Over 75,000 genetic tests exist on the market, magnifying a host of issues with regulation, prior authorization, and health equity.[4]
– American Medical Association

Medicare payments for genetic testing quadrupled in just 3 years.[5]

A roadmap for building an affordable, scalable approach

1. One complete solution | 2. New forms of care | 3. Better affordability

With the cost and effort to deliver the full breadth of specialized genetics testing outstripping most health systems' lab budgets and capabilities, it's understandable that specialty providers have sprung up to provide a growing set of testing options. Yet niche players lead to variable quality, confusion around who to contact for which tests, and increased oversight from health system managers. None of which support your goals of increased quality and reduced costs. So, how should you go about finding the right genetic testing partners? Use these thoughts as a guide.

> There has been rapid growth in the number of new genetic tests entering the market, with about 10 new tests appearing daily.[7]

1 Insist on one complete genetics solution to deliver high-quality care

While you could assemble a series of specialist testing labs to flesh out a complete genetic testing menu, a better approach would be to partner with one lab capable of handling most—or all—of your needs now and in the future. Working with a single, top-tier lab will standardize quality and simplify ordering, reviewing, and paying for diagnostics with one optimized process. The best labs will offer:

Broad and deep reach:

- Routine and advanced genetic tests
- Regional genomic centers of excellence
- Scale, quality, and access of large national reference laboratory partners
- Specialization and customization of best-in-class specialty genetic labs

Comprehensive testing across methodologies, technologies, and clinical applications including:

- Cytogenetics
- Molecular genetics
- Biochemical genetics
- Infectious agents

Support for your clinicians with offerings like:

- Screening, diagnostics, and therapeutics
- Continuity of follow-up testing with a same-director review
- IT solutions for true clinical data integration

2 Look for genetic expertise to help advise your clinicians and innovate new forms of care

When genetics are part of trying to deliver quality care, it's not enough to send samples to a lab and get results back. According to the National Institutes of Health (NIH), there are over 7,000 rare diseases, many of which are genetic in origin,[8] so sometimes there may not be an answer in the results. That's when you need a lab with broad and deep genetic expertise to work with your healthcare professionals to determine what they're dealing with. Look for a lab that:

- Has an extensive history of advancing genetic testing through ongoing innovation, and quickly makes breakthroughs available for diagnosis and treatment of genetic and rare diseases
- Has a large team of industry-leading experts including MDs and PhDs, genetic counselors, and biostatisticians to support clinicians with test selection and interpretation, consult on clinical policy and clinical claims reviews, and review your approach to genetic testing
- Has genetics experts who conduct ongoing collaboration with the nation's most advanced academic research institutions, publish extensively, and participate and provide leadership in professional medical societies such as the National Society of Genetic Counselors (NSGC) and the American Society of Human Genetics (ASHG)
- Has experts who can consult with physicians to ensure care decisions are based on detailed information and how gene variants revealed by testing might impact their patients
- Offers dedicated and accessible support options, including a dedicated phone line, to provide genetic testing support for your clinicians on their schedule

3 Insist on affordability to increase the impact of genetics across your entire patient population

High-quality healthcare should not depend on the patient's zip code or income. So, it's important to work with a lab that is making demonstrable efforts to increase accessibility for testing and broaden payer access. You need a lab that:

- Recognizes the importance of making genetic testing affordable for patients and manageable for your health system
- Is in-network with all leading healthcare payers, to help patients get the access they expect
- Has a patient assistance program to make clinical laboratory services available to patients who are underinsured, uninsured, or who cannot afford to pay for testing

To test or not to test: look to electronic health records (EHR)?

Criteria for who should be tested can be applied inconsistently, leading to treatment delays and inequity in healthcare outcomes. To explore a possible solution, a recent study developed a model using diagnostic billing information from EHR data to determine its predictive abilities. For 2,000+ patients who received chromosomal microarray (CMA) testing and a control population of 9,000, the model achieved greater than 90% accuracy versus actual testing in identifying those with potentially pathogenic copy number variants based on their CMA results.[10]

What to look for in a genetic testing partner may depend on your role

Deciding who to trust with your genetic diagnostics can impact everything from the health of patients to the health of your hospital system. Accordingly, it's not a decision made lightly—or by just one person. That makes it important to understand the similar and competing influences on key stakeholders throughout the organization so you can settle on an approach that works for everyone. In gathering your team for a system-level approach, consider including people from these disciplines to account for their needs:

Senior administration is always on the lookout for ways to strengthen both patient and hospital outcomes. Diagnostics can play a role in many of their key concerns:

- Balancing hospital performance with the cost of care
- Reducing operational complexity
- Building long-term strategic relationships
- Equity and accessibility of care

Hospital administrators care that 2/3 of lab requests may have questionable significance.[11]

Lab directors have multiple competing drivers, but first among them is the desire to solve their clinicians' diagnostic needs. A smart decision here can solve many of their pain points:

- New levels of complexity from modern diagnostics
- Large test menus
- Increasingly complex analysis of results
- Enhanced clinician support needs
- Pressure to cut costs from their process

Among the most consistently cited reasons for disparities in access to testing are challenges with healthcare provider readiness and clinical workflow.[12]

– American Society of Human Genetics Economic Impact Report

Geneticists care deeply about proving the value of this evolving, complex specialty. They will be interested in decisions that:

- Formalize a standardized approach to genetic testing
- Make testing more accessible and actionable
- Provide access to peers for help selecting appropriate tests and interpreting results
- Increase the amount of data available for clinical decisions

Nearly half of all NIH research funding specifies some connection with genetics and genomics.[13]

Barriers to bringing genetics into the exam room[14]

1. **Limited expertise within healthcare systems** – the use of genetics in healthcare requires specialized knowledge.
2. **Lack of genetic counselors** – genetic counselors engage patients before testing and after results have been received, providing detailed and nuanced information required for many tests—as well as supporting clinicians in decisions about genetic testing and understanding results.
3. **New workflows required** – clinical decision support software for genetics should alert healthcare providers when genetic testing is appropriate based on patient information, provide a list of tests with an explanation of why one might be preferred over another, and share results in clear, easily understandable language.
4. **Coverage for genetic testing** – the US exhibits patchwork coverage for genetic testing, with some tests being covered under specific circumstances while many are not covered at all. The major reason cited by payers for not covering genetic testing is a lack of evidence of clinical utility.

Genetic diagnostics should be more readily available

While many agree on the power of genetic testing, there is no shared framework yet for how those tests should be administered or paid for. That's why it's best to start with a sensible, streamlined approach that maximizes efficiencies and outcomes while minimizing confusion and cost.

Large reference labs make sense because they offer unique benefits, such as the staff and expertise to innovate new tests and methodologies. They also feature the ability to simplify ordering and interpretation of tests and control costs through deep ties to the healthcare industry. And finally, they offer programs that can reduce payment concerns for underserved patient populations.

To find additional resources or learn about how Quest Diagnostics® can help you build a state-of-the-art genetics testing program, please visit QuestDiagnostics.com/genetic-tests.

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