How a more strategic use of diagnostics can help you advance genomic medicine

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The future of precision medicine is here

The flood of genetic diagnostics that have become available promise significant advances in personalized medicine. Yet finding sustainable ways to unleash its power will require a system-level approach. An approach that adapts to the rapidly evolving field of genetics and genomics. 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.

External lab collaborations are the likely solution, but what is the ideal mix and number of labs to engage? And how can that help drive 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

Cancer, in particular, shows the opportunity at hand:

> Among patients with any of the 4 most common malignancies in the United States, Black patients are consistently more likely to die than white patients, with the exception of women with lung cancer.[1] Black women have lower incidence rates of breast cancer yet have a 40% higher mortality rate. Prostate and colorectal cancers are more often diagnosed among Black individuals, and Black men have prostate cancer mortality rates that are more than double that of their white counterparts. Genetics and genomics research should be perceived as a tool within the armamentarium needed to address the mortality gap between Black and white patients with cancer.[2]

Given the fact that at least 1/3 of health systems are using 3 or more vendors for genetic testing,[3] quality and accessibility are likely to be sacrificed in the effort to piece together a comprehensive test menu from smaller, specialty genetic testing labs. After all, different labs will have different quality standards—and smaller labs are unlikely to offer near universal payer access, which is an important consideration for equity.

So, how can health systems balance these important concerns? The answer could be simpler than you think.

Barriers to bringing genetics into the exam room[4]

1. **Limited expertise within healthcare systems** – the use of genetics in healthcare requires specialized knowledge most physicians do not have.
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 efficacy.

Single-source outsourcing may be your solution

The complexity of genetic testing offers an opening to consolidate diagnostics with larger reference labs, rather than a series of smaller specialists, that can offer the required stability and scale your health system may need. After all, it's easy to see value in reducing operational complexity through consolidation—especially when coupled with the likelihood of reducing costs due to economies of scale and reduced oversight from hospital personnel. Potential gains include

- A high-quality, comprehensive genetic test menu
- Help identifying appropriate tests
- Assistance interpreting results
- Access to genetic counselors
- Streamlined ordering mechanisms
- Simplification of revenue cycle management

Even though some may question whether larger reference labs are as nimble or cutting edge as specialty labs (an opinion that may hold merit concerning some reference labs, but certainly not all), the potential to drive cost efficiencies and build long-term, stable relationships holds value because it can help you reach your goal of controlling growth-related expenditures. If you can get the same quality results with easier management—and potentially at a lower cost—why wouldn't you? It's 1 way to help ensure genetic testing becomes more available to all.

Finally, academic collaborations may exist that can help advance the clinical utility and equity of diagnostics. In effect, working with them can also help move your system toward its overall goals.

A roadmap for building an affordable, scalable approach

1. 1 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.[5]

1 Insist on 1 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 1 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 1 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 caregivers 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,[6] 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 with

- An extensive history of advancing genetic testing through ongoing innovation, and quickly makes breakthroughs available for diagnosis and treatment of genetic and rare diseases
- 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
- 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)
- 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
- 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 making demonstrable efforts to increase accessibility for testing and broaden payer access, including

- Recognizing the importance of making genetic testing affordable for patients and manageable for your health system
- In-network with all leading healthcare payers, to help patients get the access they expect
- Robust 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.[7]

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.

Quest Diagnostics® offers 700+ genetic tests across the entire continuum, 40+ years of advanced genetic testing experience, expert clinical support with 650+ MDs and PhDs, and 50 genetic counselors and biostatisticians. We also have near-universal in-network patient access with 90%+ US health plans and payers, making us uniquely qualified to be your genetics testing partner.

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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[2] Balogun OD, Olopade OI. Addressing health disparities in cancer with genomics. *Nat Rev Genet* 2021;22:621-622. doi:10.1038/s41576-021-00390-44
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[7] Morley TJ, Han L, Castro VM, et al. Phenotypic signatures in clinical data enable systematic identification of patients for genetic testing. *Nat Med*. 2021;27:1097–1104. doi:10.1038/s41591-021-01356-z

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