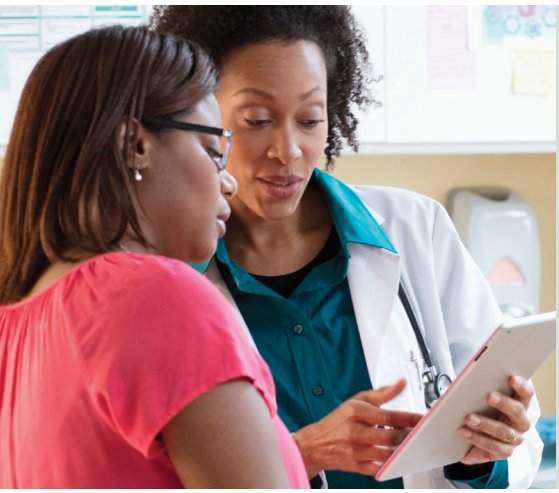
Answering the rising demand for genetic screening throughout the reproductive journey

Identifying solutions to friction points in health systems



Technology advancements and heightened public awareness of genetic disorders have combined to increase demand for genetic screening, in particular for people thinking about starting families.

This white paper explores some of the barriers health systems may face in responding to this surge in demand and where you can find solutions.

The current state of genetic screening

Genetic screening is a powerful tool that can help facilitate better patient outcomes and has become more accessible due, in part, to increased funding by the government and private sector to support the growing demand.[1]

According to one report, 30,000 individuals are screened for genetic disorders every year[2] and, among healthy people, up to 15% had a finding indicating an increased risk for a treatable or preventable disease.[3]

Insights from genetic screening can be particularly critical throughout the reproductive journey

Birth defects affect 1 in 33 babies born in the US each year.[4] While not all are related to genetics, this statistic is a factor in the high demand for prenatal screening. In fact, data show that up to 50% of pregnant women in the US receive noninvasive prenatal screening (NIPS).[5]

Access to NIPS has improved significantly, and the American College of Obstetricians and Gynecologists (ACOG) has recognized how critical genetic testing is in preparing for and managing a successful pregnancy for all women. In 2017, the organization issued recommendations for ethnic-specific, pan-ethnic, or expanded carrier screening before and during pregnancy for all women, regardless of ethnicity or family history.[6] Despite this, African American and Hispanic American women and other women from underserved groups are often not empowered to receive quality care, and without equitable access to genetic healthcare, health disparities will continue to persist.

Indeed, for many health systems, providing broader access to genetic screening can be challenging due to already overburdened systems, workflows, and genetics resources. Identifying those points of friction is imperative so health systems can complement, supplement, and/or consolidate resources to enhance efficiency and better support patients seeking genetic screening throughout the reproductive journey.

Genetic carrier screening

Every person carries several genetic variants, which usually do not result in symptoms of a genetic disease. But the children of 2 people who are carriers for the same genetic condition have an increased risk of inheriting both variants and exhibiting signs and symptoms of the genetic disorder.[7] Carrier screening can lead to the diagnosis of selected hereditary disorders in 2% to 3% of the population.[8]

cfDNA-based noninvasive prenatal screening (NIPS)

Extensive data published in peer-reviewed literature establish cfDNA-based NIPS as a powerful screening tool for fetal chromosomal aneuploidies,[9] including trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome), and trisomy 13 (Patau syndrome).

Points of friction within health systems

Even before the paradigm shift to mainstream genetic screening, health systems were straining to meet the goals of improving patient experiences and population health while reducing cost. The increased demand for genetic screening throughout the reproductive journey has illuminated common challenges in 3 distinct areas within health systems.

Inefficient systems and workflows

Despite healthcare spending in the US continuing to rise, reaching $4.5 trillion in 2022,[10] there are still significant challenges in improving health outcomes. Currently, wasteful use of resources accounts for up to a quarter of healthcare spending globally.[11]

Case in point: a study revealed that 20% of adults reported tests being ordered that had already been done in the past 2 years,[12] which potentially increases clinical risks and leads to wasted time and money spent complying with medical requests. In the US, measures to eliminate wasteful administrative spending alone could save up to $265 billion.[13]

A major step toward addressing these inefficiencies starts with enhanced use of electronic health records (EHRs). In fact, nearly $600K was recently issued by the AMA to study how EHRs can be used more effectively to reduce stress and burnout, improve workflows, boost value-based care, enhance patient outcomes, and reduce expenses.[14]

This effort will be significant, especially for health systems that are not fully integrating genetic screening into EHRs due to the complexity of testing information, referral recommendations, coordination of communication between providers and specialists, and post-test care pathways for patients identified with hereditary risk.[15] This integration requires a special expertise that isn't always inherent at health systems.

Inadequate genetics resources

A national survey found that 68% of participating organizations had many vacancies for geneticists and genetic counselors.[16] These shortages, combined with the large volume of genetic tests available, contribute to uneasiness about test ordering, interpreting results, and selecting appropriate follow-up diagnostic tests. Misinterpretation of genetic results can have significant consequences, including incorrect diagnoses, unnecessary treatments and interventions, and increased stress on patients and their families.[17]

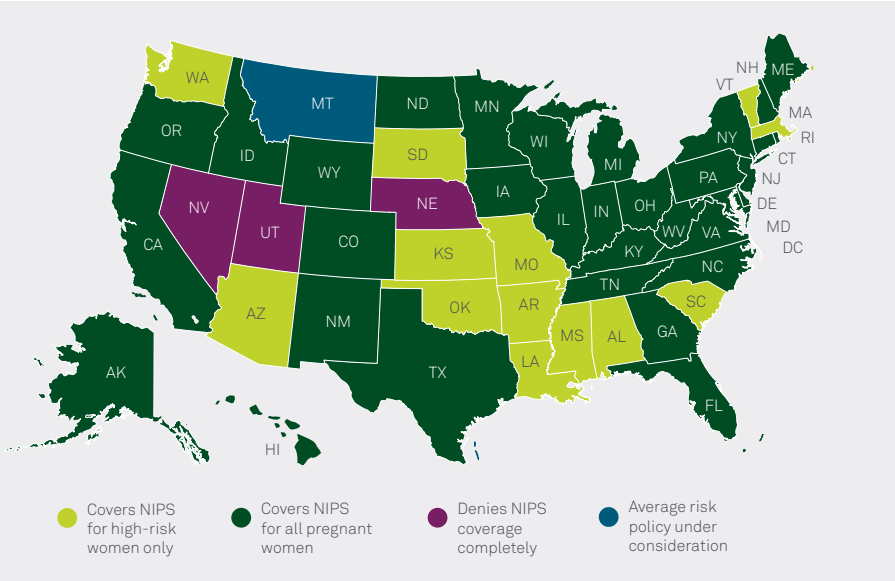
These data points paint the inauspicious picture well:

- There are **over 175,000 genetic tests on the market** with approximately **10 new tests added daily,**[18] along with **complicated coding policies,** and thousands of pages of medical policy to interpret—all making it difficult for a health system to stay current
- A recent survey showed that 83% of respondents reported that they were aware of **at least 1 example of misinterpretation of a genetic testing result** during their career[17]

Inconsistent insurance coverage and cost

Although cost has decreased, out-of-pocket costs of genetic testing can range from $100 to $2,000 depending on the type of test that is ordered and how much is covered by insurance.[15] While most state Medicaid programs cover NIPS for all pregnant women, there are some that only cover testing for high-risk women and some that do not cover it at all.[9]

Navigating coverage and preauthorization requirements is a significant administrative burden—in fact, an AMA survey reports that physicians complete an average of 41 prior authorizations per week, translating to almost 2 business days of physician and staff time.[19]



Reducing friction may come down to consolidating testing and supplementing resources with a specialized lab

These 3 common areas of friction in health systems are not trivial to resolve. However, a specialized lab focused on women's and reproductive health can help fill in gaps throughout a health system, especially for those that may be affected by the predicted shortfall of 10 million health workers by 2030.[20]

This checklist can help guide health systems in consolidating reproductive genetic screening while simultaneously reaping the benefits of a wide breadth of testing, high-quality test results, and specialized services to help navigate coverage and cost.

☑ Complete genetic testing solutions throughout the reproductive journey

- Whole patient care: testing by trimester, carrier screening, cfDNA NIPS, fetal diagnostics
- Comprehensive routine and advanced genetic testing across methodologies, technologies, and clinical applications, including cytogenetics, molecular genetics, biochemical genetics, and infectious agents

☑ Connectivity and data expertise

- Integration of complex EHR systems for visibility of all test orders, including genetic testing, to all providers
- Lab management and stewardship to help decrease unnecessary test orders and associated downstream care and support data-driven decisions about test over- and underutilization

☑ Availability of genetic experts

- Genomic scientist specialists, geneticists, and biostatisticians available for peer-to-peer consultation on test selection and results interpretation
- Post-test patient consult with genomic scientist specialist

☑ Affordable and accessible testing

- Near-universal health plan coverage
- Financial programs to assist patients
- Specialized services to help with preauthorization
- Billing/price transparency and patient support to clarify cost of testing and help with insurance and billing questions

Conclusion

Mainstream access to genetic testing throughout the reproductive journey may be the new norm. Its rapid growth and rising demand are the result of many factors, including the clinical need for better tools to predict, diagnose, treat, and monitor disease.

Working with a lab specialized in women's and reproductive health is imperative as demand for and importance of genetic screening increases. But it is crucial to work with a lab that also has a broad genetics test portfolio and the expertise to help guide test selection and interpretation.

With 67% of all US hospitals system-affiliated,[21] it's safe to say health systems are at the forefront of patient care. Identifying and removing barriers to genetic screening before and during pregnancy will be essential to improving patient experiences and delivering equitable population health, while reducing the per capita costs of healthcare.

Quest Diagnostics

Quest Diagnostics Women's and Reproductive Health offers specialized women's and reproductive health testing and services, advanced genetic screening, and expert clinical and medical insights to support health systems and their providers and patients.

Visit **ChooseQuestPrenatal.com** to learn how we can help remove barriers to genetic screening throughout the reproductive journey.

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