

Introduction

With the exponential growth of research and clinical practice related to genetic testing and personalized medicine, there is a need for genetics experts to guide providers into this new frontier. The more than 4,000 certified genetic counselors (GCs)¹ in the U.S. apply specialized education in genetics and counseling to identify appropriate testing and support interpretation of laboratory results to help in the development of an effective care plan. These masters-level, board-certified healthcare professionals provide the knowledge to support providers and members in making informed choices about genetic conditions.



Historically, the profession has been divided into two groups: clinical GCs based in hospitals and clinics and laboratory-based GCs. Today, a new role is unfolding for GCs in which they serve health plans in optimizing genetic test utilization management and offering providers highly-skilled genetics expertise in test selection and results interpretation—the two phases of the genetic testing cycle that are most often executed in error.² Understanding the complexity of genetic testing, many health plans are incorporating genetic counseling services from multiple sources to help ensure appropriate test utilization following established evidence based guidelines.



Employing these services can help control costs and provide members with the right diagnostic test progression for their specific medical circumstances, leading to better clinical outcomes.

To this end, Quest Diagnostics offers health plans the expertise of its team of more than 30 board-certified GCs who specialize in women's health, oncology, neurology, and cardiovascular disease. The Quest GC program was founded in 1987 and has expanded to encompass many

GC specialty areas that align with the growth in genetic testing research and clinical practice adoption during the intervening three decades. Additionally, Quest's 1.866.GENE.INFO call center was created in 2000 to further facilitate consultations. Trained to understand the relevance and clinical evidence of genetic tests and to translate complex genetic information into useful information, the Quest Diagnostics GC staff can help guide health plans' provider networks in the appropriate use of genetic testing for better health management.

The value of laboratory GC interventions

The role of the GC can be described in five phases: an intake phase, an initial contact phase, the encounter phase, the summary phase, and a follow-up phase.³ The laboratory-based GC's primary role is to act as the liaison between clinical and laboratory personnel. They answer complex questions from ordering providers regarding appropriate test selection, interpretation of results, and follow-up on abnormal results. They educate laboratory staff on clinical issues of diagnosis and ongoing testing for the effective management of disease. A lab-based genetic counseling team's systematic review of orders can help strengthen provider support, achieve more cost effective care, and improve member satisfaction.

Improving member satisfaction

A key priority for health plans is improving and maintaining high levels of member satisfaction, both in experience and clinical outcomes. Research indicates that improving member engagement can strengthen a health plan's financial performance by improving member retention, reducing administrative costs, and optimizing cost-of-care.⁴ The incorporation of genetic counseling services helps ensure that the right member receives the right test at the right time. Genetic counselors focus on test appropriateness, guided by established clinical guidelines, with the intent of gaining the most clinically actionable results possible for the member. In this way, genetic counseling interventions help to make certain that testing is relevant for an individual's condition or medical history, and that inappropriate testing does not lead to unneeded follow-up testing or treatment.

Recent advances in DNA sequencing technology have sometimes yielded minimal cost differences between gene-specific genetic tests versus panel tests that incorporate a much broader read of the member's DNA.⁵

However, panel testing does not guarantee better results, is not appropriate in all cases, and may present a conundrum for providers.

**Andi Ybarra, MS, CGC,
Genetic Counselor**

“Members appreciate the health and financial benefits of not having an unnecessary laboratory test.

When a provider contacts a Quest Diagnostics genetic counselor for assistance with ordering the correct test, we apply knowledge of the current clinical guidelines based on the patient’s personal and family history. It is a win for everyone involved—the provider, the patient and the health plan.”



When using a panel test, indiscriminate results may surface, such as an unexpected mutation, wholly independent of the testing intent. Consider the identification of variants of uncertain significance (VUS), which signal a mutation of no known clinical ramification and can be found more frequently with broader testing. From a genetic counseling standpoint, it might be an undue burden on the member to layer on psychosocial concern to his or her current diagnostic efforts. Moreover, VUS results may trigger unnecessary further testing, and could lead to additional out-of-pocket expenses for the member, as well as potential testing denials. By partnering with providers, genetic counselors can help avoid these member harms, ultimately assisting with overall member satisfaction.

Supporting providers in appropriate test ordering and results interpretation

Health plans have a clinical and financial responsibility to manage the proper use of genetic testing. To do so, some health plans are instituting programs to offer providers decision support when ordering specific genetic tests. For example, a pre-authorization is common for BRCA1 and BRCA2 tests. This process allows health plans to assess whether the test is indicated per current clinical guidelines, based on the member’s personal and family medical history. When an alternative testing plan is indicated, feedback from a genetic counseling team to the provider can facilitate an accurate diagnosis in less time and with decreased cost. In some cases, a genetic counselor may initiate consultation with an ordering provider, targeting frequently misordered tests.

For example, the genetic test for cystic fibrosis is commonly misordered. The recommendation from the American Congress of Obstetricians and Gynecologists (ACOG) is to begin diagnosis with a screening test. However, many providers mistakenly order gene deletion/duplication testing or a sequencing test.⁶ With only 1% of people having cystic fibrosis consistent with deletion/duplication, 99% of members tested would be missed by this method of screening. Moreover, selecting an inappropriate test adds significant, unnecessary expense to the screening process. This case is an example of how genetic counselors are able to provide needed educational support to providers to guide appropriate test selection for improved outcomes and lower costs.

**Cathi Rubin Franklin, MS, CGC,
Manager, Genomics Client Services**

“My personal expertise is hemoglobinopathies, which are very complex and many providers are not comfortable with either test selection or interpretation—even some hematologists.

I get calls all the time from clients—genetic counselors, primary care providers, OBGYNs, hematologists—asking me to consult and recommend the most appropriate testing for their patient. They are always extremely appreciative of the time I take to help them. I have many large practices that have me on ‘speed dial’ for these cases. How does this add value? It means that the clinician is getting the best information so that she or he can offer the best testing for their patient. It means that the patient is getting the test that is best for them and won’t need to worry about having to follow up with more testing or face additional costs because the most appropriate test wasn’t ordered in the first place.”



Secondly, GCs provide important assistance in the collection and review of member and family medical information. A family medical history is a diagnostic tool in its own right and can help identify people with a higher-than-usual risk of having common disorders, such as heart disease, certain cancers, and diabetes.

Lastly, the complex nature and volume of the genetic testing results requires professional interpretation to translate the clinical meaning and potential benefit to members.⁷ While providers believe genetic testing to be important to their practice, few report that they are comfortable ordering or interpreting tests.

“The vast majority of providers are very appreciative that Quest GCs take the time to call them to make sure the patient is getting the right test. Many providers are unsure about which test does what—especially when there are several for a particular disorder. Take alpha globin sequencing, alpha globin common mutations, and alpha globin dosage: all are alpha globin tests but each has a very different clinical utility, which can be extremely important. Even those providers who initially are put off by our calling, come around when we explain why we are calling and why we are recommending a more appropriate test. Those providers are usually very appreciative, especially when they find out that our recommendations are often for less expensive options that provide the appropriate testing for the patient.”

-Cathi Rubin Franklin, MS, CGC, Manager, Genomics Client Services

Moreover, genetic testing continues to experience rapid changes, but most providers do not receive significant training in genetics after the completion of medical school.⁸

In self-assessment, providers rate their knowledge of genetic testing as fair to poor.⁹ In fact, studies show that the majority of errors related to the genetic test cycle occur in pre- and post-analytic phases of laboratory testing when providers are selecting tests and applying results.¹⁰ GCs are uniquely positioned to support providers in the post-analytic processes of testing by aiding in the interpretation of test results.¹¹ Lab-based GCs are able to contact ordering providers to explain complex results, answer questions for providers who contact the laboratory, and assist providers in determining when additional testing may be helpful to further clarify a member’s diagnosis.

“Despite the importance of genetic testing, physicians typically do not always receive enough genetics education in medical school and it is difficult to maintain up-to-date information in a field changing daily. For this reason, they can be uncomfortable interpreting the results of genetic tests. Quest Diagnostics genetic counselors help providers confidently discuss results and the appropriate future health management steps.”

-Andi Ybarra, MS, CGC, Genetic Counselor II

Achieving cost savings and improved test utilization management

GC services can proactively support cost savings and reduce administrative burden to health plans. Leveraging utilization management protocols, a genetic counseling team can flag test codes that are known to have a high

misorder rate. The genetic counselor can then initiate appropriate, proactive follow-up with the provider to confirm, modify, or cancel the testing. A study published in 2014¹² demonstrated that approximately 26 percent of complex molecular test orders at a national reference laboratory over a 21-month period required modification. Test changes were classified as misorders (61%), improvements (34%), or other (5%). The cost-savings to the referring institutions from the cancellation of misordered tests alone averaged \$48,000 per month, totaling almost \$1.2 million over the course of the study. The average cost savings per misordered test was \$792. Supporting these findings, Priority Health, a Michigan-based health plan, estimated that 30% of genetic tests were ordered incorrectly. The plan increased interaction between GCs, members, and providers to ensure that the proper genetic tests were ordered, yielding a savings estimated at \$7.2 million in one year.¹³ GC intervention that reduces inappropriate testing may also decrease the administrative burden created by denials, appeals, customer service complaints, and questions from providers and members.

**Steven Keiles, MS, LCGC,
Senior Director, Genetic Counselor Organization**

“Genetic counseling is a service that we provide to all clinicians which really adds value to their practice.

This helps them ensure they are only ordering appropriate testing and are less likely to miss something in their patient population. Additionally, from a health plan perspective, our services can help minimize denials based on medical necessity, as well as the cost in employee time and resources, helping to lower the healthcare cost burden for everyone.”



GCs play an important role in industry-wide collaborations that seek to ensure appropriate test utilization. For example, Quest is a member of Pediatric Laboratory Utilization Guidance Services (PLUGS), an initiative at Seattle Children’s Hospital that is helping hospital laboratories and providers to decrease unnecessary laboratory testing, which ultimately reduces spending. Specific to genetic testing, PLUGS has collaborated to develop a genetic test utilization management solution for children’s hospitals and pediatric practices. The collaboration combines the hospital’s PLUGS utilization management (UM) program and its team of genetic testing experts with data tools

that can help genetic testing costs.¹⁴ PLUGS members who have implemented active utilization management programs save at least 10% on their total expense for tests sent to outside reference laboratories.¹⁵ Moreover, through the collaboration process, the member organizations involved in PLUGS guide the creation of national consensus policies related to pediatric laboratory testing.

In recognition of the importance of these collaboration efforts, the Quest Diagnostics GC team members contribute to industry education through the creation of study abstracts, posters, professional presentations, and peer-reviewed publications. They also dedicate time to professional advocacy groups such as the National Society of Genetic Counselors and the American Board of Genetic Counseling as well as providing mentoring for students and education for the public. Combined, these efforts help to build consensus for genetic testing best practices and help to establish the body of literature needed to develop tomorrow's guidelines.

Improved test utilization management through GC intervention

GCs backed by robust utilization management technology are able to help health plans seeking to control the ballooning cost of genetic and molecular testing.

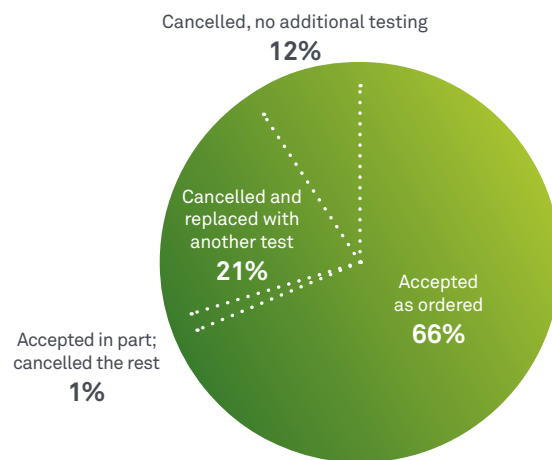
Quest Diagnostics performed an analysis of more than 3,600 genetic testing orders facilitated by GC verification in 2016. Quest has set up a system that flags commonly misordered genetic tests, allowing GCs to contact the ordering provider to verify that the ordered testing is appropriate. If the testing is not appropriate for the member, the GC may recommend alternative testing options or recommend against completing a genetic test. Ensuring that appropriate testing is performed may offer greater diagnostic value and/or significantly reduce cost.

Of the test orders verified with the ordering provider by a GC, only 59% of all genetic test orders were maintained as ordered. After GC review, 29% of all tests were cancelled and changed to a more appropriate test. Following consultation with a GC, 11% of orders were deemed inappropriate and cancelled outright with no additional testing ordered, and less than 1% of orders were partially cancelled. Looking at a subset of this testing, oncology-specific tests, provider test selection was modestly more

appropriate, with 66% of orders accepted as written, 21% replaced and 12% cancelled without additional testing. (Figure 1) In comparison, GC intervention impacted half of all non-oncology test orders. (Figure 2)

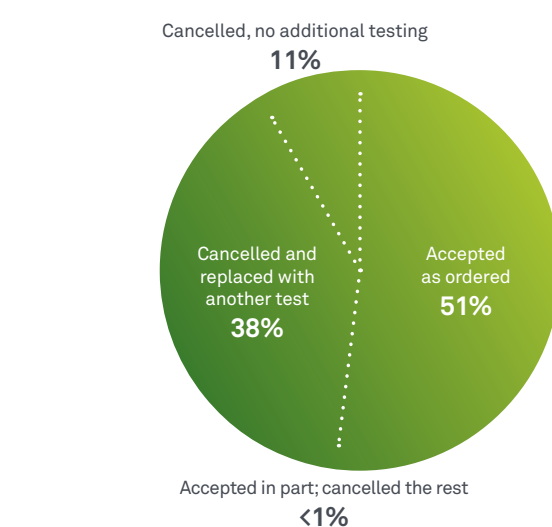
This process of verification of frequently misordered tests demonstrates the unique opportunity for lab-based GCs to help reduce unnecessary testing, guide appropriate testing, and prevent potential expenses for members and administrative burden for health plans.

Figure 1. Impact of GC intervention for oncology-related genetic tests (n=1,941)



Source: Quest Diagnostics, 2016

Figure 2. Impact of GC intervention for non-oncology, molecular genetic tests (n=1,672)



Source: Quest Diagnostics, 2016

Conclusion

The role of highly-trained GCs has expanded in recent years well beyond the clinical setting to include vital roles that help health plans deliver optimal clinical outcomes, reduce costs, and improve member satisfaction. In support of the health plan community, qualified Quest Diagnostics genetics professionals are available to help review member medical history, assess the appropriate

use of genetic testing, and guide providers in proper ordering and interpreting of tests results. The value of this role is evidenced by a large-scale study of GC-assisted genetic testing orders in which nearly half of all orders were amended through GC intervention. In this way, GC-facilitated collaboration between the ordering provider, laboratory, and health plan can optimize the clinical advantages of genetic testing for members while controlling the cost of care for the payer.

Learn more about Quest's network of genetic counselor support

Visit QuestDiagnostics.com/Genetics, contact 1.866.GENE.INFO, or get in touch with a Quest Diagnostics Health Plan representative by emailing HealthPlans@QuestDiagnostics.com.

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