The role of genetics in population health management

By Megan Czarniecki, MS, MA, CGC

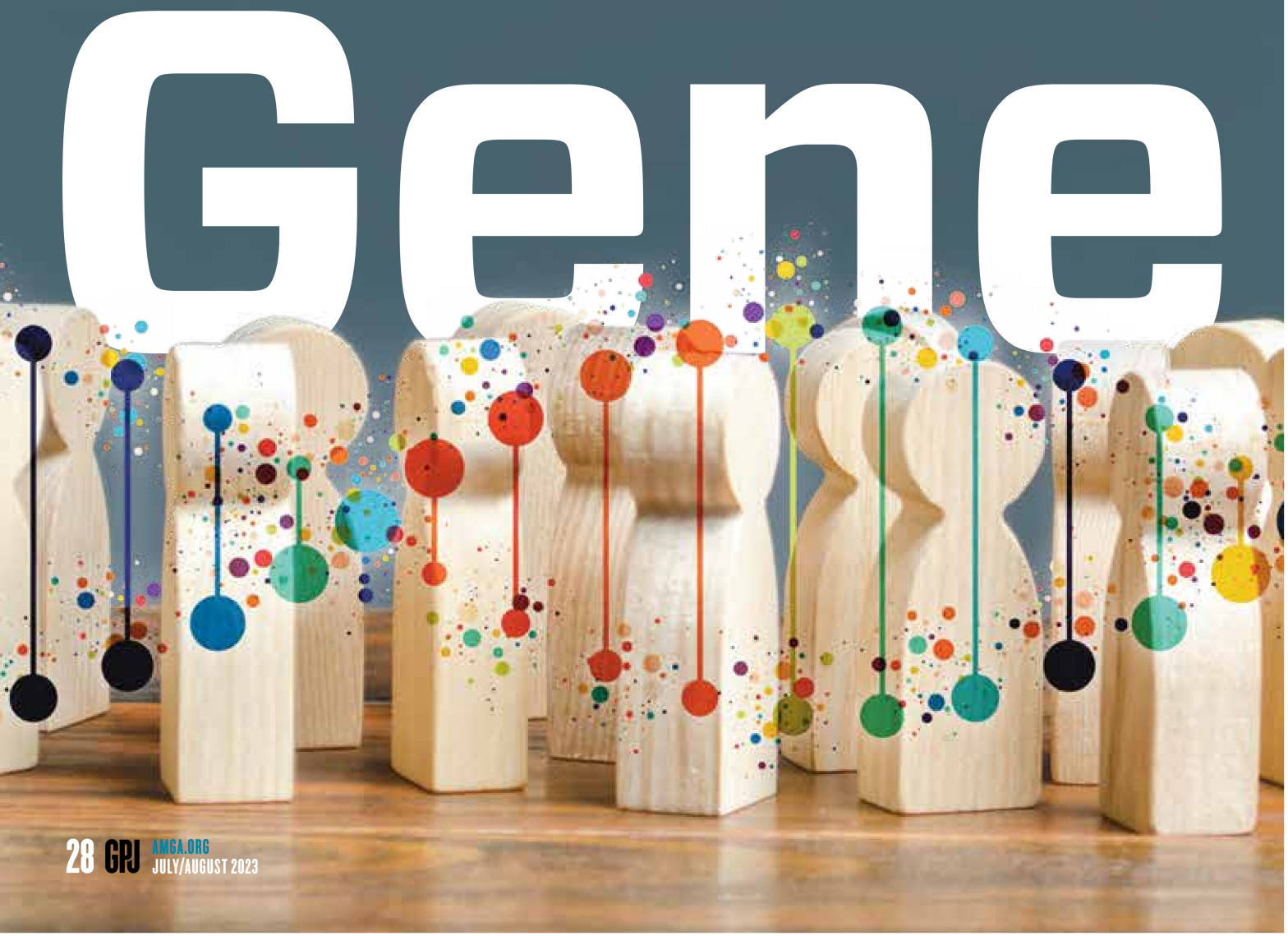
What comes to mind when you think about genetic medicine? Perhaps you conjure thoughts of a patient with cancer undergoing genetic testing to determine optimal treatment options. Or maybe you envision a young couple talking to an obstetrician about whether their future children might suffer from a rare disease that occurred in a relative.

Whatever scenario you imagine, it likely involves an interaction between an individual provider and an individual patient at high risk for a genetic disease. You probably aren't considering the role of genetics in the entire population—population health management and value-based care—but that's precisely what's looming on the horizon.

Genetic medicine holds enormous potential to drive true value for entire patient populations. That's in part because it enables a seeming paradox: personalized medicine at scale. To achieve that long-sought goal, however, the whole healthcare industry must adopt a different mindset and a different strategic approach to the way we think about genetic medicine.

The Current State

In the past 10 to 20 years, genetic medicine has mainly been focused on patients and families at high risk for genetic disorders. A classic example is that of a provider suggesting that a patient undergo genetic testing for BRCA1 and BRCA2 based on a personal or family history of breast cancer. In cases like these, we've made great strides in identifying patients who might benefit from genetic testing and targeted interventions.



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Today, providers' efforts are primarily concentrated on identifying such high-risk patients, while the majority of the population is deemed to have medium or low risk of genetic disorders and rarely offered the option of genetic testing. Based on this approach, the typical "use case" for genetic testing remains limited to the relatively small number of patients at high risk for specific diseases caused by single-gene mutations.

The identification of high-risk patients has also been left to healthcare providers who largely aren't sure where to turn for supportive genetics information and resources.

Increasingly, however, this narrow, individual-oriented view of genetic medicine is shifting—along with associated patient management strategies. Rather than only identifying small numbers of high-risk patients for relatively rare conditions, healthcare organizations are beginning to consider how to apply genetic insights to broader populations of low- and medium-risk patients—as well as to more common multifactorial clinical conditions such as cardiovascular disease, diabetes, and osteoporosis.

The desire to succeed in value-based care frameworks features prominently in this mindset shift.

Adopting a Value-Based Mindset

At their core, all value-based care models aim to achieve the same primary goals: better care and outcomes for individuals and patient populations at a lower per capita cost.[1] This requires healthcare organizations to look at the effectiveness of various interventions across populations and not just in individual patients. In other words, value-based care prioritizes ultimate outcomes at the population level.

The question then becomes: How can we move the needle on a population's overall health? Genetics plays a critical role in answering that question in two key ways:

1. **Risk stratification.** Combining genetic testing with analytics is a powerful way to stratify patient populations into low-, medium-, and high-risk groups. In value-based care, risk stratification is a huge driver of ultimate results because it enables organizations to target the appropriate interventions to those most likely to benefit from them. It helps ensure that care is correctly tailored to patients rather than delivered in a one-size-fits-all fashion.
2. **Patient engagement.** Genetic testing can also be a significant driver of individual behavior. Patients whose genetic

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tests indicate they may have a predisposition for heart disease, for example, might be more motivated to engage in heart-healthy activities. Even if genetic testing doesn't necessarily change the interventions recommended for a particular group of patients, it serves as a persuasive patient engagement and motivational tool. A glimpse into their genetic future may be just the kick-start some patients need to adopt a healthier lifestyle or better adhere to their medications.

In many ways, genetic testing enables the kind of proactive patient care that lies at the heart of value-based care and population health management. It calls upon healthcare organizations to flip the script on the classic "symptoms-diagnosis-treatment" progression. Traditionally, patient care starts when an individual reaches out to a provider for care—often for an existing condition. The provider then arrives at a diagnosis, potential interventions, and care plan.

By contrast, genetics-informed population health management starts with healthcare organizations evaluating which interventions and care plans for which patient groups or indications are most likely to keep their members the healthiest. Then, working backward from a desired outcome, they try to engage patients in the clinical programs best suited to their needs. In this way, healthcare organizations can achieve personalized medicine at scale.

The caveat, of course, is that such programs cannot be implemented physician-by-physician, patient-by-patient; they must be created at the health system or health plan level.

This doesn't change the time-honored dynamic of providers being responsible for identifying their patients' potential genetic risks and managing care appropriately. Rather, providers will need more support from health systems and health plans to successfully use genetic insights to improve population health.

A System-Level Approach

Genetics-informed population health management requires providers and healthcare organizations to expand their concept of who can benefit from genetic information. No longer should it be restricted to individual patients with a personal or family history of cancer or rare disease. There are genetic markers for a wide range of disease states, and more gene-based therapies, as well.

Genetics-informed population health programs tend to experience the best results when they involve multifactorial conditions—those influenced by both genes and the environment. For example, we know that smoking increases the odds of bladder cancer, as does a particular variant in the NAT2 gene.[2] Genetic tests could help stratify populations into low-, medium-, or high-risk groups based on their NAT2 gene profile and smoking status. Then, smoking cessation interventions could be appropriately tailored to help patients maximize healthy behaviors and minimize negative ones.

For such programs to succeed, support from provider institutions and health plans is crucial. This critical distinction separates the genetics programs of the past from those of the future.

Going forward, individual providers won't be able to conduct and apply genetic testing at the scale necessary to influence population health. Instead, health systems and health plans must commit to augmenting providers' workflows with relevant genetic information, including making it accessible to providers and patients at every point of care.

Genetic tests could help stratify populations into low-, medium-, or high-risk groups based on their gene profile.

Individual providers shouldn't need to know everything about genetics. Instead, genetic expertise should be made available within technology tools. Solutions that can be integrated into the electronic health record (EHR), for instance, can help providers interpret the results of a given genetic test or access "best practice" care recommendations.

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Genetics-informed population health programs will entail more technology, analytics, and patient engagement efforts than in the past. Yet those resources can alleviate the enormous pressure on individual providers to identify high-risk patients and keep abreast of genetic advances. A population-level program is not just about ordering the right genetic test at the right time; more importantly, it's focused on supporting patients and providers to ensure effective care delivery that creates the best possible outcomes.

Keeping Patients at the Center of Care

Patient education and engagement are other equally critical components of genetics-informed population health. Some large academic medical centers are already offering genetic testing to bigger swaths of the patient population based on family or medical history so that high-risk patients can have more informed care discussions with their providers. At the same time, the vast majority of patients who aren't high-risk can stay engaged through more tailored educational materials.

As patients move from health system to health system or health plan to health plan, patient empowerment will become the most important element of genetics-informed population health. We'll likely see programs develop as premium health plan offerings for employers, for example, because plans, employers, and patients all benefit from better outcomes at lower costs.

Indeed, for health plans, employers, and health systems, genetics can also become a "safety net" to expand access and identify potentially high-risk patients before risk manifests.

Historically, access to genetic testing and genetics-informed treatments has been limited to those patients fortunate enough to be able to visit leading disease specialists, typically at academic medical centers. Seldom is genetic information available when patients present to rural primary care practices or urgent care centers, for instance. Yet health plans and health systems that incorporate holistic, value-based contracts and leverage genetics-informed population health management programs stand to broaden access and engage patients in ways that reap tremendous clinical and financial benefits for all parties involved.

Genetics: The Gateway to Value

Genetic medicine has been on the periphery of routine patient care throughout the past decades, reserved for a small handful of patients with single-gene mutations that put them at high risk for devastating disease. Providers have been responsible for knowing when genetic testing could benefit their patients and shepherding them through the process.

Now, the industry is beginning to seek even loftier benefits by applying genetic medicine to broader patient populations. The days of individual providers trying to identify and manage small numbers of high-risk patients on their own are ending. On the horizon is an entirely new mindset aimed at leveraging genetic information to move the needle on outcomes for common, multifactorial conditions for entire populations.

The move from focusing on single, high-risk individuals to genetics-informed population health management requires health systems and health plans to adopt new strategies. More analytics, technology, and patient engagement will be necessary. But with the proper genetics expertise and resources, providers can deliver more effective, patient-centric care at the population level. Genetics thus becomes a crucial gateway to achieving all the tenets of value-based care. GPJ

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