Reducing Cost-of-Care in Next-Generation Genetic Sequencing

By

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Abstract

The cost of care in healthcare settings is rapidly rising, and it is increasingly difficult to decrease these costs without compromising quality and accessibility. This article investigates methods and potential opportunities for innovation of next-generation genetic sequencing in healthcare decisions and trajectories of care. Using various sources including medical encyclopedias and peer-reviewed journal articles, the author explores the ways in which genetic testing became popular in medicine and how genetic testing is currently being used. She uses relevant research to discuss the most likely trajectory of genetics within the field of medicine and its most cost-effective interventions. By streamlining the process of analyzing the samples and the journey of samples from a patient to specialty lab to provider, the author concludes that it is possible to reduce administrative, travel, and labor costs while expediting the process of receiving results—all of which increase quality and accessibility. Going this route also increases access by making the process more efficient. In sum, looking at the three factors of quality, cost, and access, the author analyzes this healthcare topic and offers potential innovations that satisfy a three-fold aim.

Keywords: quality, cost, price, access, genetics, triple aim

Background Research on Cost-of-Care for Genetic Testing

Oftentimes, there are costs to care well before disease treatment even begins. These costs are associated with the testing and initial appointments to determine the cause of the disease so that an appropriate treatment plan can be established and started. One of these types of costs is genetic testing, which is done for various reasons in the preliminary stages of diagnosis. Genetic tests are costly and can be the source of severe financial burdens both to insurance companies and to the patient responsible for co-payments, deductibles, and out-of-pocket costs. While next-generation genetic sequencing is a major technological advancement in medicine, there still exist hurdles in its practical implementation. To show how one might propose and implement innovations in the field of next-generation genetic sequencing, this article covers the history of genetics in medicine, outlines the costs associated with genetic testing, and delves into possible innovations to make genetic testing more affordable.

Literature Review

Providers and researchers have used genetic testing as a tool for decades. However, such testing has seen a dramatic increase in prevalence in the past ten years due to advances and the completion of the human genome project. Generally, physicians order genetic testing for two reasons: 1) to identify the genetic change associated with an existing disease; or 2) to identify a genetic change that suggests a risk for developing a disease in the future. For diagnosis, genetic testing is used to “diagnose a genetic condition if you or your child has symptoms.” For prevention, genetic testing can be used to determine if a genetic condition runs in a patient’s family before they have symptoms. Examples of how genetic tests are used are 1) to learn about the chance a current or future fetus will have a genetic condition, [or] 2) to understand and guide cancer prevention or a treatment plan (Genetic Testing 2022).

The three types of genetic tests performed increase in cost as the number of genes tested increases. The first type is a single gene test that, as the name suggests,
selects one gene and sequences it. This test provides the physician, or another provider, with information on a single gene. The second type of test is a panel test, which selects for and sequences an array of genes that fit into the same category or cluster. This type of test is used in screening for cancer risks and for other traits such as “low muscle tone, short stature, or epilepsy” (Genetic Testing 2022). The third type is a large-scale test. This test can either be in the form of exon sequencing— the sequencing of all genetic material that is expressed in DNA– or genome sequencing, which looks at the entire genome of a person. The important financial takeaway from these three levels of genetic tests is that as more genes are sequenced, the cost of sequencing increases. However, testing costs can vary greatly. A study by Lynce & Isaacs (2016) estimates that the cost of sequencing a panel, the second level of sequencing, can “range from $249 to $6,040, with most costing $1,500 to $6,040.” Clearly, the cost of genetic testing may pose a great financial burden and it is only the first step in finding the cause of the disease.

Who is Affected by Cost?

Because of its potential for widespread use, everyone is potentially affected by the high cost of genetic testing. As previously stated, genetic testing is used primarily in the diagnosis of disease as well as for research that may affect the future of human health. Researchers use next-generation sequencing to find drugs that can target specific genetic mutations in hopes of curing genetic conditions like cystic fibrosis and various forms of breast cancer. However, focusing more on the medical diagnosis use of genetic testing, 62% of providers in one survey said that their main barrier to genetic testing was its cost (Rahma et al. 2020). Cost impacts both the patient and the provider because, while the patient cannot receive the recommended testing for their condition, the provider also cannot diagnose the issue in the way they think is best. A conversation around cost involving more stakeholders and who is most impacted by the high cost is needed.

Insurance companies are the gatekeepers to patients’ having access to genetic testing because they decide which medical tests they will cover, and the prices patients will be charged. By following the chain, we can see that the decision-making level continues to trace back to the source. A doctor orders a specific test, patients are made aware of the portion of the cost for which they are responsible, or the insurer denies the test based on cost. In a survey of physicians, Rahma et al. (2020) found that “91.9% showed a positive attitude regarding availability of genetic testing” but identified the barriers as “the cost of testing (62%), followed by lack of training or education and insurance coverage (57.8% and 57.2%, respectively)” All of these factors increase the administrative burden, which, in turn, increases the cost of a test that a gatekeeper decides may not be approved. Overall, the issue of the cost of genetic testing is a major issue for multiple stakeholders.

Future Unintended Consequences of the Cost Barrier

Next-generation sequencing is a tool that has taken decades to develop. If we do not properly utilize it, then we as a society will suffer from preventable deaths and curable diseases. We need to take a long-term approach to the barrier of cost in next-generation sequencing.

Regarding shorter-term consequences, there are people today who are not receiving the proper medical care because of their inability to afford the expensive genetic test that would drive the proper route of care. Pinzón-Espinosa et al. (2022) discuss this issue in the field of mental health and found that “genetic testing may provide information to identify, classify, and discriminate between different stages of the disease or patient subtypes, thereby contributing to the objective of personalized patient care.” Sadly, if the cost of next-generation sequencing continues to be a barrier to its use, an unknown number (but likely a high percentage) of patients will go either undiagnosed, misdiagnosed, untreated, or mistreated.

The high cost of genetic testing will be a barrier to conducting trials that have the potential to cure disease and improve patient outcomes. Rine & Fagen (2015) discuss the lack of increased funding for scientific-medical research in the United States alongside the increased cost of this research. They examine the cost of genetic research in depth and how the high cost discourages organizations interested in conducting this research from applying for grants. In such cases, the cost of conducting the research will likely be estimated at more than a single grant would cover. Rine & Fagen (2015) state that “it takes a million dollars or more to set up a new junior faculty member at many research institutions” and proposes that cost be weighed against the ‘cost’ of not sustaining existing junior, mid-career, and senior faculty who suffer a funding gap.”
Reducing the Cost of Genetic Testing Will Serve the Greater Good

Based on the research discussed in this article, it follows that decreasing the cost of care for next-generation sequencing and, therefore, increasing its use would serve the greater good. Academic medical centers are beginning to increase the use of genetic testing by working with the government to cover these tests. Many hospitals accept Medicaid/Medicare to cover the cost of care for those who meet the federal requirements for each program’s enrollment. There are specific rules and coverage benefits from Medicare aimed at the target population of older people. Expanding Medicare benefits to include genetic testing would greatly improve health outcomes for these already at-risk patients.

The issue now lies within the purview of physicians, medical centers, and the government (acting as the insurer) to expand the benefits of Medicare. Lin et al. (2021) looked at academic medical centers and safety net clinics to determine the current methods for paying for genetic tests when ordered by a physician and found that most of the cost negotiation was actually happening between the medical facility and the laboratory performing the test. This phenomenon presents a flip side of the story to the proposed solution of involving policymakers to have the government cover more of the cost of testing. Lin et al. (2021) state that these medical sites “largely worked with laboratories that offer testing for little to no OOP [Out of Pocket] cost to patients, resulting in relatively few economic barriers to testing” and how they innovated on their own to deliver the best care to patients.

The inability of key stakeholders, including private insurance companies and policymakers acting as insurers for Medicaid, to resolve the issue of cost has forced innovation on the part of providers to deliver access to genetic testing for their patients. However, this innovation created by providers offers only a temporary solution because of the inability of private laboratories to provide “no or low OOP costs if panel testing is not covered (which is the case for most insurers), no-cost cascade testing for family members, and financial assistance programs for low-income patients” (Lin et al. 2021).

Therefore, the solution that the author of the current article proposes is that providers—i.e., physicians, administrative hospital staff, and organizations that provide care, implement their own genetic testing, collect the genetic samples, sequence them, and deliver the results all within the walls of the organization such as an academic medical center.

Society at large would also benefit from decreased cost and increased use of genetic testing because genetic testing would enrich the educational experience of students both in high school and in higher education. Obviously, not only would the increased delivery of genetic information enhance the learning opportunities of medical students and those in biology-focused higher education programs, but it would also open the minds of high school students by exposing them to diverse options in the science, technology, engineering, and math fields. These educational opportunities could mold today’s learners into tomorrow’s leaders in a meaningful way. Additionally, access to genetic databases can enhance the education of medical students and those in professional clinical studies enabling them to be better equipped to face genetic diagnoses in their patients (Kopel 2019).

An Innovation Plan to Address the Cost-of-Care Issue Based on Relevant Research

This section delves deeper into proposed solutions to the cost barrier of genetic sequencing, including expanding Medicare coverage of next-generation sequencing, making the technology cost less, and installing the technology in the healthcare facility instead of an off-site laboratory.

Previous and Current Innovations

The field of genetic sequencing has undergone a rapid expansion in the past two decades, with a significant increase in the last five years alone with the introduction of CRISPR technology being used in research and medicine. The demand for the use of this technology has forced industry leaders to consider methods of cost reduction to use CRISPR more broadly. The use of genetic testing and CRISPR has become a full industry that functions alongside the healthcare industry in a nuanced space, and the business world is taking notice. Vassey (2021) discusses the increase in funding for genetic testing in recent years, saying that “It is estimated that the global genetic testing market value accounted for nearly $13 billion in 2019 and is forecasted to nearly triple by 2026.” Vassey (2021) also considers the innovation of direct-to-consumer genetic tests such as 23andMe. This test offers 55+ health results using an at-home test that the consumer mails back to the company which sequences the sample in their laboratory. 23andMe “has raised nearly $800 million
in funding and was last valued at $2.5 billion” (Vassev 2021). The innovation of direct-to-consumer tests has greatly increased the access to genetic testing by more people for a lower cost but is not a complete solution, as these companies do not offer extensive testing of entire genomes.

Vassey (2021) explains that companies like Genesight and BiogeniQ have developed whole genome sequencing to fill the need for more complete genetic sequencing for doctors to accurately diagnose and treat genetic diseases. These companies and others are trying to not only offer whole genome sequencing but also create databases that are continuously updated so that the financial investment spent on this testing can stretch as much as possible by being useful in the future.

The Proposed Solution in Detail

The proposed solution to properly address the issue of the high cost of testing is three-fold. First, expand coverage for genetic testing so it can serve as a preventative tool for disease and decrease the cost of disease and illness down the line. Second, install genetic sequencing technology such as Ultima, a startup that has created technology to sequence genes for as little as $100, right in the healthcare facilities so that these organizations may process the genetic samples in the same administrative system in which they are collected. The third solution involves creating a results-sharing database. This action would stretch the dollars that are being spent on expensive genetic sequencing, ensure that no results are wasted, and that the data would be available for future research of treatments and cures.

A committee formed by the US Institute of Medicine said, “the insurance concept of what is reimbursable (so-called medically necessary) should be defined to include appropriate genetic testing and related education and counseling, and these genetics services be reimbursed under health insurance plans” (Andrews et al. 1994). This quotation articulates the first step in the solution of covering testing through Medicaid to expand the use of testing to more of the low-income population.

Adding in the second and third parts of the solution will put the technology, information, and, therefore, power in the hands of providers and patients. “Ultima is the first company to deliver a $100 sequence” (Furchgott 2022) and with this technology installed in hospitals where the doctors are ordering the tests, faster results will be delivered to providers and patients, thus improving health outcomes. As Furchgott (2022) puts it, “what can be applied at the bedside” is what matters. The final piece of the solution is using all this new information to compile a database of genetic information that can be used by providers and researchers alike to better treat patients and to also develop treatments and cures.

How This Solution Addresses the Issue of Cost

The solution described here addresses the issue from multiple angles and would not only fix current problems, but also ideally prevent future problems. Involving policymakers to address Medicaid’s lack of coverage for many genetic tests would allow for wider access. In this case, policymakers are acting as insurers to help other stakeholders such as providers and patients, to improve access to technology that leads to improved patient outcomes.

To incentivize the expanded coverage, healthcare organizations could install the next-generation sequencing equipment on-site and minimize outsourcing costs that come with administrative and logistical costs. An example of this would be the installation of the G4 system, which sequences genetic samples and automatically uploads the results to the Terra platform. Terra was “developed by the Broad Institute of MIT and Harvard, Microsoft, and Verily—for secondary and tertiary data analysis, storage, and sharing of G4-generated data” (G4 workflow 2022). These steps address the issue of funding these tests by reducing as well as creating a database to stretch the dollars spent, making the cost even more justified (increasing value).

Implementing This Plan

This plan can be implemented by lobbying Congress to introduce a bill that will expand Medicaid coverage for genetic testing. Simultaneously, there can be increased funding to private companies, crowdfunding, or grants to produce and install the technology in healthcare settings. This money would be funneled to Ultima, G4, and other companies that specialize in equipment to sequence genes for a lower cost. Singular Genomics, the parent company of G4, uses private investors for the development and manufacturing of its technology (Investors 2022). Ultima Genomics draws funding through companies acting as investors (About Ultima Genomics) while Element Biosciences draws funding from only a handful of venture capitalist firms (Element secures $15 million Series a financing 2022). Once these technologies are installed in hospitals, and policymakers and insurers have provided the coverage, providers can start ordering tests and contributing to the database of genetic information.
Long-Term Effects of the Proposed Plan

The long-term benefits of genetic testing have been clearly demonstrated by a wealth of research done in various clinical disciplines including mental health and psychiatric care, early detection of and treatment of cancer, screening and diagnosis of chronic hereditary conditions. The Pinzón-Espinosa et al. (2022) article about the use of genetic testing in psychiatric care puts it best: “New technologies in genetic testing provide important new information about the diagnosis, treatment, and prevention of diseases and are of great value for precision medicine.” A goal of genetic testing is to eventually make precision medicine not only widely available, but also the norm. Furchgott’s (2022) article on genetic testing technology states that “much more research is needed to reach the point where genome sequencing becomes a ubiquitous part of a checkup. But cheaper sequencing is a critical first step.” This solution will, in the long term, have a positive impact on each aspect of the triple aim, starting with cost. By decreasing the cost of genetic testing, access as well as the quality of care will also increase, especially for lower-income patients on Medicaid. By improving the care of the most at-risk groups in society, we are improving public health overall and lifting collective health in the long run.

CONCLUSION

Next-generation genetic sequencing represents an exciting chiasm of science and business as the prospect of personalized medicine becomes more possible. This intersection of all stakeholders in healthcare demonstrates the need for collaboration to realize the potential of advancements in care. Policymakers must expand coverage of genetic testing for Medicaid (and Medicare) patients, and insurers at large can increase their coverage of testing. Providers need to be thoughtful in which tests they are ordering, and patients need to work with their providers to make sure the care they receive is appropriate. Payers—the insurer, the patient, or another source, need to understand the benefits of genetic testing and work with the technologies offered to increase the number of tests being performed. Technology will continue to improve so long as long as stakeholders believe in its potential.

This article demonstrates the immense potential for improved quality of care that genetic testing offers and how that can become a reality in the US healthcare system through increased coverage of testing, new technologies that offer sequencing for a reduced cost, and the creation and maintenance of genetic information databases. Through this multi-faceted approach, genetic information can be in the hands of providers and patients and will improve health outcomes.

References


About the Author: Sarah Auletta is a student in the class of 2024 at Simmons University in Boston, Massachusetts, where she is double majoring in Public Health and Spanish and minoring in Healthcare Management. She presented the project on which this article is based at the 14th Annual Undergraduate Research Conference on Health and Society at Providence College. Sarah has worked at a variety of jobs including being the practice biller and grant administrator for her hometown, operations manager for an arts festival and coordinating logistics for Simmons’ orientation program. With a passion to make as many people feel better as possible, her love for the field of public health was born. She is excited to see where these interests take her both in academia and in practice.