Preparing for Personalized Medicine in Massachusetts

Enhancing Genetics Education for Patients and Health Professionals

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September 2013
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Acknowledgements

We would like to acknowledge the tremendous support from the Institute of Politics, particularly Laura Simolaris, the staff adviser of the Policy Program and Policy Program Directors Valentina Perez and David Mazza.

Additionally, we would like to thank Luciana Herman, Lecturer in Public Policy at the Harvard Kennedy School, Dr. Sean Palfrey, MD, Clinical Professor of Pediatrics and Public Health at Boston University, and Ravi Parikh, MD/MPP Candidate at Harvard University for their guidance and comments on the drafts of this paper. We would also like to thank Dr. Steve S. Han, MD, PhD of the Massachusetts General Hospital, Dr. Brian Skotko MD, MPP Co-director of the Down Syndrome Program at Massachusetts General Hospital, and Dr. Anthony John Iafrate, MD, Director of Molecular Pathology at Massachusetts General Hospital for their insights on genetic testing in the clinical realm.

The opinions expressed herein are those of the student authors and do not reflect the views of the Institute of Politics, clinicians and reviewers acknowledged here.
Executive Summary

Since the completion of the Human Genome Project in 2003, a veritable scientific army has devoted itself to deciphering how DNA influences biological processes and identifying the genetic variants that give rise to disease. The combination of this emerging breadth of knowledge with commercially available DNA sequencing technology has led to the rise of genome-wide analysis and genetic tests as powerful clinical tools for both diagnosing and customizing treatments.

However, several obstacles prevent the widespread use of genetic in clinical practice. Few providers cover clinical genetic testing procedures and while the cost of genetic tests have decreased, the costs of interpretation services have not.\textsuperscript{1,2} There is evidence that some physicians do not feel prepared to be cast into the position of interpreting and using these tests to inform their clinical decisions.\textsuperscript{3} Physicians often do not need to participate in genetics education and training to fulfill state-mandated continuing medical education requirements, which means that most exposure to genetics occurs in medical school or residency-training programs. Additionally, patients might be misinformed about the limitations as well as social and legal complexities of genetic testing. Furthermore, the benefits obtained from genetic analysis must be coupled with concern over the patients’ privacy: who can access these analyses, what protections patients have, and how clinicians should obtain informed consent.

The state of Massachusetts, home of the Whitehead Institute which originally sequenced over a third of the human genome in the early 2000’s, has a decade-long record of leadership in accruing knowledge about the human genome. But very little progress has been made in using genetic testing to improve health outcomes. Currently, there is neither a clear agenda nor funding to broadly expand educational initiatives in genetics for both health professionals and patients. Creating a broad genetics educational infrastructure as well as establishing clearer privacy rights will allow Massachusetts to not only set a higher standard for health care but also create collaborations amongst its cutting-edge biotechnology industry and strong network of medical institutions.

Given the rapid rate at which personalized genetics has advanced, it is imperative that the Massachusetts state legislature begin to formulate policies and regulations to address the various barriers and concerns surrounding its full integration into medicine.

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Methodology

A group of Harvard undergraduates and a research fellow at Boston Children’s Hospital conducted a literature review about educational issues related to genetic testing using PubMed and Google Scholar. Certain time parameters were established for the literature review. For our findings about privacy concerns, we examined only articles published after the passage of the Genetic Nondiscrimination Act of 2008. Additionally, we interviewed and consulted several physicians in the Boston area. In this paper we will use the term “genetic testing” to broadly refer to not only tests for single gene Mendelian traits, but also more complex tests frequently referred to as “genomic testing” which is consistent with the often interchangeable usage in the literature.
The Promise of Personalized Medicine

Personalized genetic testing has not only presented a new avenue for understanding the roots of previously unexplainable illnesses, but also revealed their immense diversity and widespread prevalence in the human population. From the early detection of genetic disorders in the embryo to the characterization of patient tumors for the development of personalized cancer therapeutics, genetic testing is well on its way to becoming an integral part of medical practice.

Additionally, personalized medicine has three clear advantages for the future of Massachusetts. The first benefit entails continuing the long-standing tradition of Massachusetts acting as a leader in innovative health care. Broad educational initiatives can help usher in the widespread use of genetics-based diagnosis and treatment. Encouraging the proper use of genetic testing in the clinic favors preventative care, a key goal of health care reform.

Secondly, as the cost of genetic tests decrease over time, the use of these tests will result in significant cost savings in treatment. Being able to prescribe drugs specifically designed to combat mutations that lead to disease will not only improve therapeutic outcomes but also prevent costly complications. For example, Warfarin, a drug used to treat blood clots, can require extremely varied dosage depending on the patient. The incorrect dosage can result in side effects such as severe bleeding. However, a genetic test can provide an indication of the appropriate dosage of warfarin prior to treatment of the patient. By preventing serious adverse events, providers will save millions of dollars annually.4

Thirdly, investing in genetics education for both patients and health professionals is a crucial opportunity for Massachusetts to bolster its strong biotechnology base and tap into the roughly $25 billion genetic testing industry that is projected to grow over the next 10 years.5 However, currently the uncertainty regarding widespread adoption of clinical genetic testing limits biotechnology investments in this area.6 Enhancing physician education can work to overcome investor concerns. Additionally, there is vast potential for new growth not only in developing novel tests, but in offering interpretation services for the data generated from DNA sequencing.7

In addition to these substantive benefits, several current events have pushed public discussion about personalized medicine to new heights, making legislative action on this topic appropriate. In June 2013, the Supreme Court ruled on Association for Molecular Pathology v. Myriad Genetics, determining that genes cannot be directly patented, opening up the market for competitors to develop tests for mutant BRCA1/2 genes which are linked to breast cancer. The coincidence of this ruling with the high profile New York Times op-ed written by actress Angelina Jolie on her voluntary mastectomy after learning that she inherited a mutant BRCA1 gene has given great momentum to the issue of genetic testing. The strong public interest in genetics and medicine suggests that an educational initiative promoting the widespread use of genetic tests in the clinic is particularly timely.
Preparing Physicians for Personalized Care in Massachusetts

The future of health care is one in which the use of genetic testing in the clinic will be the standard-of-care. According to Tarini et al., common genetic mutations that modestly increase risk of developing a disease, called low penetrance mutations, will be incorporated into routine assessments.\(^8\) Indications of the increasing importance of genetic testing in primary care, in particular, have already been established. For example, the BRCA test, which searches for mutations of the BRCA1/2 genes that confer a high risk of developing breast cancer, is expanding beyond the traditional purview of oncologists. The Affordable Care Act has taken the first step in catalyzing the spread of personalized medicine into the primary care clinic by defining BRCA testing as a preventative service.\(^9\) As a result, women with a history of the mutation in their family will be able to receive both the test and genetic counseling sessions at no cost and will likely be seeking this test in a primary care setting.\(^10\)

However, many primary care physicians report that they are not prepared to take on the new role of interpreting genetic tests and using them to inform clinical decisions. Importantly, primary care physicians will be on the frontlines as genetic advances develop. In fact, the need for increased genetics education for physicians is clear and has been established by numerous studies across the nation.\(^11\) Many physicians openly avow their lack of knowledge and comfort in integrating genetic testing into their practice.\(^12\) At the same time, some physicians are establishing practices heavily based on medical genetics. This gap in knowledge should be filled so that all physicians feel comfortable with a basic threshold of genetics knowledge.

**Lack of Physician Education**

Primary care physicians face difficulties in effectively using genetic tests in their practice. Primary care physicians agree that genetic testing will become more common in the next 5 years, yet only 22% feel that their training is sufficient to work with patients who have had genetic testing.\(^13\) Many primary care physicians have little experience in genetic testing, and thus lack confidence in ordering tests.\(^14\) In a survey of over 10,000 US physicians, nearly 4000 of whom identified as primary care providers, 97.6% physicians agreed that the response of specific drugs was influenced by genetics. However, only 10.3% felt competently educated about pharmacogenomic testing, which can determine whether a patient has the genetic variants that promote a better response to a drug.\(^15\)

Pediatricians similarly face challenges and difficulties in interpreting genetic information and communicating the information to patients.\(^16\) Skills in communication are becoming increasingly more important, especially in the clinical application of genetic testing. A survey-based study conducted on physicians who ordered chromosomal microarray analysis (CMA) between May 2008 and March 2011 displayed the lack of understanding of CMA and genetic testing.\(^17\) The physicians displayed difficulties in providing adequate pre-testing counseling and disclosing incidental and uncertain findings post-testing. In particular, the study showed that pediatricians and pediatric specialists especially struggled and reported a need for more education about CMA.\(^18\) Pediatricians have expressed concerns for residency programs to adapt to the changes in medicine and prepare physicians for communicating genetic information through understanding, simplifying, and explaining information in an accessible manner.\(^19\)
However, the need for enhanced training in genetics is not confined to primary care physicians. Even within subspecialties like oncology, which routinely use genetic testing as a clinical tool, additional physician education resources are necessary. When physicians or counselors were not specifically trained in genetics test interpretation, adverse consequences included misdiagnoses impacting medical liability, increased medical procedural costs, and mistrust in the physician-patient relationship.\(^{20}\) However, the impact of physician education in the field of oncology extends beyond malpractice and misdiagnosis. In a 2011 study published in *Genetics in Medicine*, researchers assessing physician education about genetic testing in cancer care discovered that oncologists unfamiliar with genetic testing were less likely to recommend adequate medical treatment and strategies when patients had BRCA1-positive test results.\(^{21}\) Thus, enhanced physician education is needed not only for reducing misdiagnosis, but also for prescribing adequate and effective treatment.

**Barriers to Physician Education**

Given that over 70% of physician associations consider genetics education an important responsibility, it is strange that a consistent lack of education about genetic testing still persists.\(^{22}\) We have identified systemic barriers to physician education that impede the widespread adoption of genetics knowledge.

At the crux of this dichotomy between perceived importance and participation in genetics education are competing demands in curricula and funding that deprioritize the integration of genetics within continuing medical education initiatives. Currently, the lack of genetic education in physician curricula nationally is partially due to a paucity of qualified faculty and a lack of applied genetic training. In Massachusetts, unlike other states, this lack of training cannot be attributed to medical school curricula. Out of the four medical schools in Massachusetts (Harvard, Boston University, Tufts, and University of Massachusetts), all have required genetics classes during the first two years of medical school.

However, this in-classroom teaching of genetics does not translate to practice years later. One study, by comparing scores from a basic genetics test in first years to a case-based test of genetics in a clinical setting during the third year of medical school, showed that medical students have a very low retention of genetics knowledge and display an inability to apply their knowledge to clinical situations.\(^{23}\) This gap points to a need for genetics education to be comprehensively taught in an applied setting through the clinical years of medical school, residency, and beyond as part of continuing medical education programs.

**Recommendations for Physician Education**

In order to ensure that physicians are properly trained to take advantage of scientific advances that could improve patient care, the state of Massachusetts should take the following actions to promote physician education about clinical genetic testing.
Establish Grants for Innovative Training Initiatives

Massachusetts should provide grants for pilot programs to enhance the integration of genetics education into the clinical years of medical school and residency. Programs may include:

(i) formal education pilots in the third and fourth years of medical school or residency training

(ii) informal peer-to-peer learning initiatives

(iii) the development of online training modules

These grants are not intended to fund programs for medical geneticists, but rather provide for broader education to non-specialists in genetics. Programs receiving funding must engage in specific training about informed consent for patients and the social implications of genetic testing. These programs will be evaluated over a period of three years and the findings ought to be revisited to determine effectiveness.

Integrate Genetics into Continuing Medical Education Requirements

Massachusetts should require all physicians to take Continuing Medical Education (CME) credits on the use of clinical genetic tests. The curriculum covered by these CME classes should include but are not limited to covering the following: scientific background and recent advances, clinical decision making support, application to specific specialties, communication for informed consent, and social implications of genetic testing. One hundred hours of continuing medical education are currently required for physicians by the state of Massachusetts to complete before renewing medical licenses biennially. Certain hours are allocated to specific subjects such as pain management. In other states such as Florida, Rhode Island, and Nevada, more defined subjects such as bioterrorism and HIV/AIDS management are state CME requirements. Therefore, there is a precedent for states to specify that CME requirements include important topics, such as genetics education.

Creating a Team: Using Genetic Counselors and Nurses

As the use of genetic testing expands, health professionals across a variety of disciplines will need to be prepared to work together to deal with genetic diseases.

The services of genetic counselors will be especially crucial because of a lack of physicians who are trained in genetics or interpretation services to meet the demand. Genetic counselors can act independently or as part of inter-disciplinary teams to address patients’ needs. A Vanderbilt study of a pilot genetic counseling clinic staffed by genetic counselors and advised by medical geneticists with M.D.’s found that genetic counselors were able to effectively address the needs of over 80% of patients in a timely manner without having to consult the doctors. In the study, primary care providers most frequently utilized the referrals to genetic counselors. Patient satisfaction surveys showed that patient’s trusted in the provider’s knowledge about the subject and approved of their receptiveness to answering questions.
Additionally, genetic counselors play an important supportive role. In a study of patients tested for BRCA mutations, it was found that greater time spent with genetic professionals during counseling was associated with better cancer risk management strategies among patients. The causality of this relationship cannot be established, as more proactive patients may be likely to both spend more time in these informative sessions as well as adopt the best risk management strategies. Nevertheless, the study’s results are a testament to the importance of genetic counselors in explaining the results of genetic tests.

According to the online registry of licensed genetic counselors in Massachusetts, which is available through the Massachusetts Executive Office of Health and Human Services, there are 163 licensed genetic counselors in the state. This translates to one genetic counselor for roughly 40,525 people in the state of Massachusetts. To put this number into perspective, there is one primary care physician for every 758 individuals in Massachusetts. Additionally, an estimated one in ten patients seen by a primary care physician has a genetic component to their disorder. The combination of these figures suggests that there is a deficiency in the number of available genetic counselors in Massachusetts. Exacerbating the sparseness of genetic counselors is the fact that they are primarily concentrated in urban, academic medical institutions.

In addition to genetic counselors, nurses are uniquely positioned to act as key contributors of genetic and genomic medicine because of their training in effective communication skills, ability to access comprehensive family histories, and scientific expertise in biology. As the most trusted health care provider in the United States, a nurse can act as crucial gatherer of information for patients and can access details about family history that physicians may not have time to gather. Yet, genetics is severely underemphasized in nurse training; in 2005, only 30% of nursing programs included genetics or genomics in their curriculums. Additionally, few nurses pursue advanced training in genetics. According to the Genetic Nursing Credentialing Commission’s online registry, there was only one Advanced Practice Nurse in Genetics (APNG) in Massachusetts as of 2007 and no genetic clinical nurses (GCN).

Creating a team of health professionals trained in genetics from physicians to genetic counselors and nurses will create multiple entry points for the integration of genetic testing into medicine. Figure 1 shows the referral network between genetic counselors, provider teams of physicians and nurses, and medical geneticists who have an M.D. and have completed residency training in genetics. Non-specialist physicians and nurses can directly provide care for patients whose disorders have genetic components. However, those who are constrained by time or feel inadequately prepared to deal with genetics-related issues can refer to genetic counselors for common cases and directly to medical geneticists for especially complex cases. Similarly, genetic counselors dealing with complex cases can refer to medical geneticists as well. However, in order for this network of referrals to be effective, there must be sufficient numbers of trained genetic counselors, nurses, and medical geneticists.
Recommendations for Genetic Counselors and Nurses

Create Accelerated Programs for Genetic Counseling

Since genetic tests will play a bigger role in future medical practice, it will be necessary to increase collaboration between physicians and genetic counselors. Because there are not enough genetic counselors to meet the demand for such services, we propose that accelerated training modules be made available to license more genetic counselors. Potentially, an undergraduate education could be paired with a certification program and licensing exam to meet the need for genetic counselors. Similar programs exist in other fields, such as the Harvard Undergraduate Teacher Education Program for students interested in pursuing teaching. Undergraduates can enroll in coursework taken in the last year of college which prepares students to become certified teachers in the state of Massachusetts.

Massachusetts should change its licensing requirements for genetic counselors to allow the licensing of graduates of accelerated genetic counseling programs. However, in order for this recommendation to become realized, coordination must take place between the American Board of Genetic Counseling (ABGC), individual universities, and the Massachusetts state licensing board. The board requires all genetic counselors to be certified by either the ABGC or American Board of Medical Genetics (ABMG) which require a minimum of a master’s degree in genetics for certification. Therefore, in order for the creation of accelerated genetic counseling programs, a variety of actors must work together.

Expand Nursing Core Competencies

The Massachusetts Department of Higher Education should update the Nursing Core Competencies to include advanced training in genetics as a knowledge component of Patient Centered Care. This will set a statewide standard for increased focus on genetics and integration of simple, yet crucial techniques in the clinic such as obtaining detailed family histories for all nursing programs run with support from the state of Massachusetts. In the long term, greater exposure to genetics and genomics early in training may incentivize nurses to attain advanced degrees in genetics certified through the Genetics Nursing Credentialing Commission. Additionally, accelerated nursing programs that combine advanced training in genetics could be considered in the future similar to the proposed program for genetic counseling.

Educating Patients: A Holistic Approach

Several areas of patient education about genetic testing merit concern. With proper education, patients should be able to understand the function of a genetic test and the reasons that it may be used. Patients should also be aware of their rights--privacy, confidentiality, use of data for research, and non-discriminatory measures. Lastly, patients should understand the results of a genetic test and how it can impact their current lifestyle. The disparity in patient education of genetic testing could also be of potential concern because certain population groups are more educated about genetic testing and may seek out these tests more than other groups that would have equally benefited from genetic tests. Particular populations that are more likely to be better educated about genetic testing are urban populations, aged 50-74 populations, and college graduates with access to internet and a regular source of healthcare, or a prior cancer diagnosis,
demonstrated greater knowledge about genetic testing.\textsuperscript{38} Though this disparity may inherently exist due to socioeconomic status, location, and other factors, healthcare professionals should be prepared to educate people of all backgrounds to ensure equal access to genetic tests.

\textit{Informed Consent}

One of the primary vehicles of patient education about genetic testing is through informed consent. Informed consent is the legal process through which medical practitioners educate and inform the patient of the potential benefits and risks associated with a procedure. Due to ambiguous test results and a high risk of misinterpretation, informed consent plays a crucial role in both genetic testing and whole genome sequencing. For example, research has shown that cancer patients who attend genetic counseling sessions afterwards overestimate their personal cancer risk.\textsuperscript{39} In the current law governing informed consent for genetic tests in Massachusetts, Chapter 111, Section 70g clearly defines “prior written consent” to include a patient’s understanding of the nature and reliability of genetic diagnostic tests. Currently, informed consent is required for all non-diagnostic genetic testing. The law requires the following: through conversations with a medical professional, that the patient be made aware of the disease for which they are testing for, purpose of the test, reliability of positive and negative results, level of certainty of positive result related to disease, available genetic counselors, and persons who can see the results of the test.

Importantly, the text excludes a crucial part of whole genome sequencing: secondary or incidental findings. Due to the powerful and comprehensive nature of whole-genome sequencing, results can reveal information about diseases other than the one originally being investigated. Patients need to be ready to receive information regarding other disorders or even family relationships.\textsuperscript{40}

\textbf{Recommendations for Patient Education}

\textit{Improve Genetics Curriculum in the High School Classroom}

It is imperative to provide holistic genetic education to the public so that when they become patients undergoing genetic testing, they are more equipped to fully understand the implications of the test and the interpretation of the results. In order to achieve this broad understanding, schools must first pinpoint broader concepts in genetics which need to be emphasized in the science curriculum. In a study conducted by Dougherty et al., educational experts rated the competency of teaching 12 core genetics concepts by state.\textsuperscript{41} The following 8 core concepts are rated ‘Inadequate’ in Massachusetts according to this study.

- Genes exist in different forms called alleles.
- Polygenic (or complex) traits (e.g., height, blood glucose) often show continuous variation within populations and are less predictable than single-gene traits.
- Polygenic traits are influenced by multiple genes and their products.
- Virtually all cells within an organism contain the same genetic information.
- Different genes are turned on and off at specific times to form different types of cells and to influence the way different cells function.
- The functions of genes and their products can be affected by the environment and other genes at one or many steps involved in producing a trait.
- Only mutations in the DNA of sex cells will be passed on to offspring. Mutations in somatic cells will be passed on to descendant cells.
- One harmful effect of mutations is genetic disease. Some genetic diseases are inherited (e.g., Tay-Sachs), and others develop during life (e.g., cancer). These concepts should be emphasized in a re-evaluated genetics curriculum taught to high school students. In addition to this broader goal of improving genetics education, targeted teaching about genetic testing should also be implemented. Similar teaching is already occurring on a small scale via PgEd, an educational initiative started by Harvard Medical School, which seeks to engage students in genetics and examine social, ethical, and legal implications of genetic testing. PgEd should be used as a model to integrate a higher level of genetics education into Massachusetts public high schools.

Establish Online Resources for Patients

To improve access to genetic information and services, a central web-based portal should be created. This portal should include information about genetic testing, disease management, support groups, clinical services, hotline numbers, a directory of qualified genetic service providers, and a breakdown of legal rights similar to a genetics portal created by Michigan. The portal should also include fact sheets translated into other languages including but not limited to Spanish, Mandarin, and Haitian Creole. Once such a website is established, it will require little maintenance cost and will be able to provide easily accessible information to patients. In addition, a central genetics resource center can be established to make information dissemination more efficient.

In order to enhance patient education about genetic testing, interactive computer programs may be useful as a supplement to genetic counselors. Green, et al. shows that interactive computer programs can offer different advantages in patient education about genetic testing for breast cancer susceptibility. The interactive computer program was preferred for helping patients learn at their own pace, avoid embarrassment, make good use of time, explain genes, and remain private. Such patient decision aids may be eligible for a secondary source of funding from the Affordable Care Act as well. A provision included in the ACA, which has not yet been actively pursued by the Secretary of the Department of Health and Human Services, is to provide grants for the development and evaluation of patient decision aids for treatment options. However it is unclear whether educational material for genetic testing would qualify for these grants and further investigation is necessary to determine if any federal funding is applicable.

Expand Informed Consent

The current law requiring “prior written consent” for all non-diagnostic testing should be further specified to include discussion about incidental findings in genetic testing and whole-genome sequencing. By structuring the ideal physician-patient conversation, we will be able to ensure that all patients are properly educated about genetic tests.

However, a broad mandate for written informed consent should not be extended to all diagnostic tests. Requiring written informed consent in all cases might thwart patient care. In 2011, the Massachusetts Society of Pathologists expressed their opposition to broad informed consent requirements for all diagnostic genetic testing proposed in bill S. 1080. Firstly, the need to conduct genetic testing may be realized only after the specimen has arrived at the laboratory. Having to acquire written informed consent post-receipt of the specimen would be lengthy and
bureaucratic, potentially leading to degradation of the specimen. Secondly, a specimen found during surgery may require immediate testing, and informed consent cannot be feasibly obtained from the patient at that time.

Yet, the extension of informed consent to further tests would increase patient education. Thus, it would be best to evaluate the adoption of written informed consent for diagnostic tests in clinical specialties independently and carefully. For example, prenatal testing appears to be an area in which written informed consent could be required across the board. However, further investigation and deliberation is necessary before any broad measures could be adopted across all specialties.

**Concerning Privacy: Combating Misinformation**

The fact that genetic information is unique to an individual, renders it more valuable than the results of non-genetic tests, and thus concern over the privacy of genetic information is much more salient than for general medical information. Concerns about genetic discrimination are especially prominent. However, in 2008 the federal government partially remedied this problem by passing the Genetic Information Nondiscrimination Act (GINA) which prevents discrimination for health insurance and employment based on genetic information. GINA failed to cover long term, life, and disability insurance as well as excluded veterans and Native Americans. However Massachusetts partially closed these loopholes by prohibiting insurance companies from requiring genetic tests. Additionally, in 2011, a bill (S.1080) coined the “Genetic Bill of Rights” was introduced in the Massachusetts state legislature that would entirely close these gaps in GINA. Since its introduction, the bill failed to move past its committee and has yet to be re-introduced in the current cycle.

*Impact of Patient Privacy Concerns*

Given the existing protections in place, it seems logically unlikely that privacy concerns or fear of genetic discrimination would factor into patients’ decision to undergo genetic testing. However, numerous studies indicate otherwise. Many clinicians are unaware of GINA and the protections it offers or fail to explicitly inform their patients about GINA. This deficit on the part of clinicians is detrimental to patients. In a study by Lowstuter, et al., 75% of physicians and medical professionals surveyed stated that concerns about privacy prevented patients from undergoing testing. The study also revealed that more than 60% of respondents were uneducated about GINA and their state’s laws protecting patients’ from discrimination by health care companies based on results of genetic testing. A study conducted by Laedtke, et al., which shows that 54.5% of the qualified physician respondents did not know about the existence or implications and protections provided by GINA, also indicates that physicians are woefully unaware of the laws protecting patients’ privacy.

*Impact of Privacy Concerns in Massachusetts*

Even four years after the passage of the Genetic Information Nondiscrimination Act of 2008, more than half of the public is still concerned about genetic discrimination. Such misconceptions exist in Massachusetts. A study conducted at the Dana Farber Cancer Institute in
conjunction with Massachusetts General Hospital found that 44% of respondents voiced concerns about discrimination in regards to insurance and employment when questioned about genetic testing and full-genome sequencing. This is indicative of an overall lack of knowledge about existing protections against genetic discrimination and a failure of health care professionals to inform their patients about the policy protections in place.

Patients’ fears that the results of their genetic tests may be used against them by insurance companies or employers negatively impact many of their decisions to undergo possibly lifesaving testing. Physicians must be educated about the protections provided by GINA and Massachusetts state law so that they can educate their patients and ensure that privacy concerns do not deter patients from undergoing genetic testing.

**Recommendations for Privacy Concerns**

*Facilitate Informed Discussions on Genetic Discrimination*

Due to widespread misconceptions about potential genetic discrimination among patients, physicians should be required to integrate a discussion about privacy concerns and legislative protections in place, such as GINA, as a part of informed consent. This should resolve any hesitancy to receive tests based on fears of social discrimination, since knowledge about GINA has empirically decreased patient concerns about engaging in genetic testing. As a result, physicians must remain up-to-date on the legislative landscape of privacy protections on both state and federal levels. Educational briefs on current genetic privacy policy should be available on the online patient resource portal recommended above.

*Completely close the GINA gap*

Current Massachusetts law already contains broad anti-discrimination policies for genetic information in the areas of life, long term, and disability insurance, which are the three broad areas that the Genetic Information Nondisclosure Act of 2008 failed to address. However, life and long-term care insurers are allowed to use voluntarily submitted genetic information to set terms for insurance policies. This exception should be removed, as proposed by S. 1080 (Massachusetts Genetic Bill of Rights) in order to ensure that genetic information can never influence insurance premiums.

**Challenges and Limitations**

In order for Massachusetts to create a population of health professionals and patients that are prepared to use clinical genetic tests, a broad range of actors will have to cooperate and prioritize genetics education initiatives. Foremost is the wide array of higher educational institutions and hospitals that have the greatest ability to directly engage practitioners and implement education initiatives. Also, professional organizations can create significant change and drive the adoption of greater interest in genetics. Opening channels of communication and reconciling competing interests will be an important undertaking in the coming years.

Additionally, questions of equal access will have to be monitored and evaluated in order to ensure that patient education can be disseminated across language barriers and varying geographic areas outside of urban centers. In particular, informed consent may be challenging when patients present with limited health literacy and/or language barriers.
Lastly, critics of an expansive vision of personalized medicine warn that extensive training is unnecessary since the current dearth of information about how genetic variants impact disease make it impractical to use genetic tests to inform clinical decisions. Many members of the research community have raised numerous concerns about rushing newly emerging discoveries into the clinic immediately. However, waiting to begin education on genetics until new therapies have emerged will greatly delay the adoption of clinical genetic tests for diagnostic purposes in the clinic. Training a workforce capable of adeptly translating genetic information into clinical decisions is vital now. It is imperative that an educational infrastructure for genetics is established immediately so that health professionals can take advantage of not only current knowledge, but also scientific advances as they emerge in the near future.

**Next Steps**

A budgetary analysis on the cost of funding physician education initiatives must be conducted along with an investigation of any federal funding sources that can be appropriated at the state level or by individual medical schools, hospitals, or institutions. Further study should also explore the funding possibilities provided by industry, while ensuring that the financial motives of industry do not unduly bias the curriculum of CME courses. Industry support can act as both an opportunity to advance widespread dissemination of current technologies and a threat to the separation of financial interests driving medicine.

Figure 2 shows the relationship between various proposed policies in this paper to achieve a minimally acceptable level of genetics education among physicians and patients. A key component of achieving an informed patient population is to ensure that health professionals are properly trained in genetics and communication strategies to explain important concepts and protections to patients.

![Hierarchy of education levels and relationships between intervening educational initiatives for physicians and patients. Orange boxes represent proposed recommendations.](image)

**Fig. 2** Hierarchy of education levels and relationships between intervening educational initiatives for physicians and patients. Orange boxes represent proposed recommendations.
Conclusion

Massachusetts lies poised to cement its already well regarded position as a leader in health care innovation by becoming a leader in personalized medicine. By prioritizing the training of health professionals across disciplines in genetics, Massachusetts can have a workforce prepared to deliver a higher quality of healthcare that puts into practice the scientific advances of the 21st century. Additionally, investing in genetics education for both patients and health professionals represents another opportunity for Massachusetts to bolster its strong biotechnology base. The trajectory of genetic testing has already been set. It is only a question of whether or not Massachusetts is ready to seize the leadership reigns in ushering in personalized medicine and taking advantage of the genetics knowledge that was developed a decade ago in its research centers.
Appendices

SWOT Analysis

**Key Opportunities**
- Strengthens biotech industry and medical practices
  - Can increase the use of genetic testing, thus prompting more business for genetic and diagnostics labs
  - Can lead to more research in the field of genetics
  - Massachusetts becomes even more of a medical hub
- Reduces health care coast in the long term
  - Allows for personalized medicine and better targeted treatments—less time and less use of treatments that are not beneficial
  - Encourages screening
  - Predictive of chronic diseases, and may encourage healthier lifestyles
- Advances knowledge
  - Translation from lab to clinic
- Provides productive infrastructure
  - By creating an infrastructure of education for physicians, genetic counselors, and nurses to better understand and interpret genetic tests, providers will be more efficient and able to provide the best care to patients as medical therapies increasingly rely on these tests.
Charts

Fig. 1: Genetics Referral Network

Fig 2: Relationship between various educational initiatives
**Glossary of Terms**

BRCA1: A gene that has been shown to correlate with an increased risk of breast cancer when mutated.

Chromosomal Microarray Analysis: A type of genetic test that can be used to diagnose many different genetic disorders. This test analyses all of a person’s chromosomes simultaneously.

Chromosome: A structure made of DNA and proteins. Each person has a total of 46 chromosomes.

DNA Sequencing: The practice of taking a biological sample from a patient and using it to determine the patient’s individual genome.

DNA: Deoxyribonucleic Acid, which is the molecule that forms genetic material.

Genetic Counselor: A medical provider who is trained to interpret the results of a genetic test and explain those results to the patient.

Genetic Discrimination: Any discrimination that occurs purely in response to a person’s genetic information. Examples include employer discrimination against workers with a certain genetic attribute or discrimination by insurance companies based on the possibility of a future medical condition.

Genetic Non-Discrimination Act of 2008 (GINA): A federal bill that was signed into law on May 21, 2008. This bill protected people from discrimination in health insurance and employment based on their genetic information.

Genetic Testing: Using DNA sequencing to diagnose a patient with a disease or screen for the future likelihood of having a disease.

Genetics: The study of how genetic material, particularly DNA, and people’s genetic make-up interacts with their health.

Genome: The entirety of a person’s genetic material.

Genomics: The study of genomes, focusing on how genomes are similar and different among different members of the population.

Informed Consent: The legal process through which a medical practitioner informs the patient of the potential risks and benefits of a procedure. Informed consent often (but not always) involves written documentation.

Massachusetts Genetic Bill of Rights: A bill (S. 1080) proposed by the Massachusetts state legislature that has not yet been passed into law. If passed, the bill would greatly expand on the informed consent rules governing genetic testing. It also looks to increase patients’ guaranteed privacy rights to their genetic information.

Oncology: The study and treatment of cancer.

Personalized Medicine: Medical care that is based on a person’s individual genetics, which could potentially allow treatment to be much more effective.

Residency Programs: Training programs for medical school graduates while they complete their residency on the way to becoming certified physicians.
28 Ibid


Ibid


