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Prevention for those who can pay: insurance reimbursement of genetic-based preventive interventions in the liminal state between health and disease

Anya E.R. Prince*

Center for Genomics and Society (CGS), School of Medicine, University of North Carolina-Chapel Hill, NC, USA
Corresponding author. E-mail: anya_prince@med.unc.edu

ABSTRACT

Clinical use of genetic testing to predict adult onset conditions allows individuals to minimize or circumvent disease when preventive medical interventions are available. Recent policy recommendations and changes expand patient access to information about asymptomatic genetic conditions and create mechanisms for expanded insurance coverage for genetic tests. The American College of Medical Genetics and Genomics (ACMG) recommends that laboratories provide incidental findings of medically actionable genetic variants after whole genome sequencing. The Patient Protection and Affordable Care Act (ACA) established mechanisms to mandate

*Ms Prince received her JD and Masters of Public Policy at Georgetown University. Prior to her position as Post-Doctoral Research Fellow at CGS, she worked at the Cancer Legal Resource Center, where she focused on discrimination and insurance concerns for those with genetic predispositions to cancer. There, she assisted women with insurance appeals for coverage of preventive screenings and surgeries. In her current position, she continues to focus on the practical effects and applications of policy and law on individuals and their families through Gene-Screen. Gene-Screen is a project, funded by the National Human Genome Research Institute that investigates the ethical, legal, and social implications of genetic screening for medically actionable genetic variants in a healthy adult population. Her current research projects explore how genetic information complicates applications and claims of fraud in life insurance applications and whether genomic research results should be automatically placed in a participant's medical records.

coverage for genetic tests, such as BRCA. The ACA and ACMG, however, do not address insurance coverage for preventive interventions. These policies equate access to testing as access to prevention, without exploring the accessibility and affordability of interventions. In reality, insurance coverage for preventive interventions in asymptomatic adults is variable given the US health insurance system's focus on treatment. Health disparities will be exacerbated if only privileged segments of society can access preventive interventions, such as prophylactic surgeries, screenings, or medication. To ensure equitable access to interventions, federal or state legislatures should mandate insurance coverage for both predictive genetic testing and recommended follow-up interventions included in a list established by an expert panel or regulatory body.

KEYWORDS: ACA, ACMG, health disparity, insurance coverage, predictive genetic testing, prevention

INTRODUCTION

Genetic technology is lauded for its promise to prevent disease, yet predictive genetic testing in and of itself provides only information, not prevention. If there is a preventive power of genetic testing, it arises from the personal actions taken by the individual and the clinical actions taken in concert with a healthcare professional, given the information learned from the genome. In reality, any knowledge of one's genetic risks equates to the possibility of mitigating disease only when given adequate access to recommended medical interventions. For example, predictive genetic testing can indicate the need to increase cancer screening or undergo preventive surgery for those at high risk of certain conditions, such as hereditary non-polyposis colorectal cancer or hereditary breast and ovarian cancer (HBOC). Despite this, recent policy recommendations and changes, such as the American College of Medical Genetics and Genomics (ACMG) incidental findings recommendations and the Patient Protection and Affordable Care Act (ACA), conflate access to genetic testing with access to preventive services, without addressing issues of insurance coverage for the ensuing medical interventions. Unequal access to interventions threatens to increase health disparities and creates an unjust health system in society if only certain segments of our population may attempt to mitigate disease through preventive measures.

The example of genomic technologies challenges the entrenched dichotomy in the insurance realm of prevention versus treatment and introduces a new dimension to the recurring discourse. Health insurers primarily provide coverage for treatment more often than prevention, but the medical interventions for adult onset genetic conditions occupy a hazy space between these extremes. Continued advances in genomic technology will likely lead to more policy implementation, similar to the ACMG recommendations and the ACA, that provides individuals with the opportunity to learn about genetic risk for conditions for which they have no symptoms. Access to interventions for asymptomatic individuals following a predictive genetic test or genetic screening, even for those with insurance coverage and a regular source of care, may be much more limited than access to similar interventions after other preventive measures, such as cancer screening, or after diagnosis.

This paper focuses on predictive genetic testing in asymptomatic individuals that can lead to medical options to prevent adult onset genetic conditions and the changing state of insurance coverage for subsequent recommended interventions. While this is a very specific emphasis, recent policy focus on prevention provides a window of opportunity to address disparity concerns in this arena. As knowledge of the predictive value of genetic variants and available preventive measures grows, issues of insurance coverage for the interventions will become increasingly important. Part I provides a brief overview of genetic testing in the clinical setting, including a description of key terms and concepts. Part II examines insurance coverage of genetic testing prior to the ACA. Health insurers primarily provide coverage for treatment more often than prevention, but the medical interventions for adult onset genetic conditions occupy a hazy space between these extremes. Given the historical treatment focus of insurance and the desire to decrease health care costs through preventive, the ACA specifically addressed insurance coverage of preventive services. Part III of the paper discusses the ACA's expansion of insurance coverage for genetic testing, specifically examining BRCA testing coverage changes. Newly created policy mechanisms established with regard to the US Preventive Services Task Force (USPSTF) may allow for broader coverage of other predictive genetic testing and interventions. Part IV examines how the movement from genetic medicine to genomic medicine complicates the insurance system. Although the ACMG recommendations were focused on how laboratories should address incidental findings, they may represent the first incidences of preventive genomic testing. However, this section explores how patients may be harmed by learning of genetic predispositions that they cannot prevent due to lack of insurance coverage. Part V delves more deeply into the ethical concerns of health disparities and distributive justice that may arise from the expansion of access to genetic test results, without the concurrent expansion of access to insurance coverage for preventive measures. Finally, Part VI proposes policy recommendations that can minimize concerns over equal and complete access to prevention and intervention.

Increased clinical use of and growing insurance coverage for predictive genetic testing threaten to exacerbate health disparities across society absent reimbursement for associated interventions. Clinical use of predictive genetic testing is still nascent, thus providing an opportunity to learn from these early attempts at policy implementation. Access to interventions is of vital consideration for any future policy efforts to implement screening on a broader population level. To ensure equitable access to interventions, federal or state legislatures should mandate insurance coverage for both predictive genetic testing and recommended follow-up interventions included in a list established by an expert panel or regulatory body.

I. GENETIC TESTING, PREVENTION, AND THE US HEALTHCARE SYSTEM

A. Key terms and definitions

The landscape of genetic and genomic testing is complex and this paper is focused on a specific type of testing in a specific population, therefore this section will define these key categories used throughout the paper. This paper discusses both genetic and genomic technologies. Genetics is the study of individual genes and their effect on

diseases, such as Huntington's disease or cystic fibrosis. Genomics, a broader concept, is the study of an individual's entire genome and the examination of how different genes and the environment interact together. The paper specifically focuses on predictive genetic testing, rather than diagnostic, carrier status, or other genetic testing. Also, the paper is more narrowly concerned with predictive testing only for medically actionable conditions, those for which there are recommended medical interventions that aim to prevent the condition. While there may be medical interventions in pediatric or prenatal settings, the scope of the paper will focus on adult onset conditions. Any use of genetic testing in the paper refers to predictive testing for medically actionable adult onset disease, unless otherwise indicated.

The primary inquiry of the paper is insurance access to preventive interventions. Therefore, the population of interest is asymptomatic adults because individuals who have not developed any symptoms or detected biological changes are the population that would seek preventive interventions. Additionally, the term 'patient' is used to acknowledge interactions between clinicians and individuals. However, patient is not meant to indicate that the individual has symptoms or is in treatment. In this paper, asymptomatic individuals may also be patients to the extent that they have a relationship with a physician.

B. Prevention in the US healthcare system

The current model of insurance coverage in the US favors payment for treatment of diseases that have already developed over coverage for preventive services. Only a fraction of healthcare expenditures are spent on prevention,[1] thus financially incentivizing clinicians to also focus on sickness rather than prevention.[2] In this vein, the standard of care has incorporated genetics and genomics technologies when they generally relate to personalized treatment for diagnosed diseases, pharmocogenetics, or diagnostic inquiries, but their use for prevention has not yet widely become standard of care.[3]

Despite the rapid expansion in our knowledge regarding the 20,000–25,000 genes in the human body, the functions of the vast majority of genes and their variants are not well understood. At one end of the spectrum of possible genetic testing lies the interesting-but-harmless result, such as an ability to roll one's tongue; at the other end lies harmful conditions, such as Huntington's disease. Somewhere along the continuum falls susceptibility to disease, such as predisposition to colon cancer. The clinical utility of information about deleterious mutations associated with susceptibility varies due to the penetrance of the mutation—how likely a person with the mutation is to manifest symptoms—, the preventability of the condition that develops, and the validity of

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1 Carla Denly, *U.S. Ranks near Bottom among Industrialized Nations in Efficiency of Health Care Spending*, UCLA Newsroom, Dec. 12, 2013, http://newsroom.ucla.edu/portal/ucla/weak-u-s-health-care-system-ranks-249652.aspx (accessed March 3, 2015).

2 Abdulrahman El-Sayed, *Prevention vs. Treatment and the Perverse Incentives Inflating the Costs of Healthcare*, Huffington Post, Oct. 18, 2011, http://www.huffingtonpost.com/abdulrahman-m-elsayed/health-care-prevention_b_1015734.html (accessed March 3, 2015).

3 See eg Genetics and Public Policy Center, *Genetic Testing Practice Guidelines: Translating Genetic Discoveries into Clinical Care*, 27, 2008, http://www.dnapolicy.org/images/issuebriefpdfs/Professional_Guidelines_Issue_Brief.pdf (accessed March 3, 2015) (noting the broad range of clinical uses of genetic testing including disease diagnosis, disease management, and reproductive decision-making aid).

the test. Clinical validity describes how well a test identifies a clinical status and, in the context of genetic testing, is dependent in part on penetrance, whereas clinical utility depends upon which actionable medical steps the test provides for the patient.

Presently, asymptomatic individuals learn of their genetic risk through predictive genetic testing, such as testing for a specific gene or panel of genes through a healthcare professional or direct-to-consumer testing, or as a secondary finding of a clinical genomic test ordered for a different condition.[4] The preventive measures that clinicians would recommend in response to identification of a deleterious mutation generally fall into four varied categories—surveillance, preventive surgery, drug treatment, and lifestyle changes. Multiple interventions may be available for the same condition and interventions may be recommended simultaneously or in progression. As knowledge of genomics increases, these options for intervention could potentially expand to other areas, such as gene modification or environmental manipulation to influence epigenetics.

Public health literature references prevention by type—primary, secondary, and tertiary.[5] Primary prevention occurs before a disease manifests through symptoms or biological changes. A common example is vaccination to protect against certain infectious childhood diseases. Whereas primary prevention reduces both incidence and prevalence of a condition because it blocks an individual from getting a disease, secondary prevention occurs after biological changes have arisen in an individual but reduces disease severity by preventing progression or mortality. Cancer screenings are a common secondary prevention. They do not stop cancer occurrence, but can lead to early discovery and higher rates of remission. Tertiary prevention refers to procedures that are part of the treatment or management of disease that slow disease development and minimize morbidity or mortality; however, because tertiary prevention occurs in symptomatic patients, it is not a focus of this paper.

The concept of secondary prevention illustrates the fine line between prevention and treatment and its implication for insurance coverage. Secondary prevention, such as a mammography for asymptomatic individuals, does not prevent cancer development, but aims to decrease the morbidity and mortality associated with a cancer diagnosis by discovering any symptoms as early as possible. As soon as a secondary prevention measure detects an abnormality, an individual begins a diagnostic odyssey to determine a definitive clinical diagnosis and subsequent treatment, regardless of whether the biological changes are at very early stages.[6] Because detection of an abnormality triggers an abrupt shift from prevention to treatment in our current healthcare culture, we have no intermediary status for patients between these extremes. Indeed, our healthcare system does not have common or adequate language to describe this liminal state. The abrupt

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4 On Nov. 22, 2013, the FDA sent a cease and desist letter to the direct-to-consumer genetic testing company, 23andMe, which halted direct-to-consumer testing across the US. Therefore, although genetic testing may be offered through these companies in the future, direct-to-consumer testing is not currently widely available. See eg Patricia J. Zettler et al., *23andMe, the Food and Drug Administration, and the Future of Genetic Testing*, 174 JAMA Int. Med. 493, 493 (2014) (explaining that if direct-to-consumer companies, such as 23andMe, can show both analytic validity and clinical validity for offered tests, they may be able to resume offering tests directly to consumers).

5 See eg Robert S. Gordon, Jr., *An Operational Classification of Disease Prevention*, 98 Pub. Health Rep. 107, 107 (1983).

6 H. Gilbert Welch et al., Overdiagnosed: Making People Sick in the Pursuit of Health (2011).

shift to treatment leads to evidence and concerns of overdiagnosis and overtreatment of patients without symptoms or other medical concerns.[7]

A positive genetic test or a family history of illness may cause an individual, whether symptomatic or not, and his/her physician to go looking for biological changes at an earlier stage than they would have without this knowledge. In result, diagnosis and treatment may occur when the disease is at a very early stage.[8] Detected biological changes that do not rise to the threshold of diagnosis have been called 'predisease'.[9] However, despite its ambiguous state, our dichotomized healthcare system often forces predisease to be categorized as a disease needing treatment—threatening more overdiagnoses. The introduction of genetic testing likely expands this predisease state since biological changes can be detected at increasingly earlier moments, thus further complicating the line between prevention and treatment. Categorizing interventions as prevention or treatment based on an indistinct and shifting threshold has implications for insurance coverage and reimbursement. Asymptomatic individuals seeking insurance coverage for preventive interventions, such as cancer screenings or prophylactic surgery, are forced to straddle a variety of laws that silo their genetic condition into prevention or treatment categories.

The diversity of possible interventions for any individual gene mutation foreshadows difficulties that insurance companies face in evaluating the clinical utility of each intervention. Personalized treatment plans and prevention options create a burden on the insurance system to develop effective and appropriate coverage policies for each intervention. Due in part to these difficulties, insurance policies for genetic testing and preventive interventions have not been systematically or comprehensively completed, leaving a fragmented system with varied levels of reimbursement for preventive testing and interventions.[10]

II. THE STATE OF INSURANCE COVERAGE PRE-ACA

In the face of incomplete insurance company reimbursement policies, wide use of genetic and genomic testing is unlikely.[11] Typically, only small subsets of the population are having genetic testing for certain diseases, making it difficult for companies to gather data and determine the utility of broad application.[12] Insurance coverage of genetic testing varies greatly depending on the type of insurance and the genetic test. Generally, coverage decisions are influenced by whether the test is diagnostic, preventive, or informational and whether the test has both clinically validity and utility.[13]

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7 See *id.* at 134, 135.

8 *Id.*; See also Anya E.R. Prince & Benjamin E. Berkman, *When Does an Illness Begin: Genetic Discrimination and Disease Manifestation*, 40 J.L. MED. & ETHICS 655, 662 (2012).

9 Eg Anthony J. Viera, *Predisease: When Does It Make Sense?*, 33 EPIDEMIOLOGIC REV. 122 (2011).

10 See *infra* part II.C.

11 Teri. A. Manolio et al., *Implementing Genomic Medicine in the Clinic: The Future Is Here*, 15 GENET. MED. 258, 262 (2013) (noting that insurance reimbursement of the costs of genomic sequencing are a crucial component for increased use of the technology).

12 Muin. J. Khoury et al., *The Evidence Dilemma in Genomic Medicine*, 27 HEALTH AFF. 1600, 1604–05 (2008).

13 Wylie Burke, 60 *Clinical Validity and Clinical Utility of Genetic Tests*, CURR. PROTOCOLS HUM. GENET. 9:15.1, 9:15.3 (2009); U.S. Department of Health and Human Services, Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS), *Coverage and Reimbursement of Genetic Tests and Services* (2006), http://osp.od.nih.gov/sites/default/files/CR_report.pdf (accessed March 3, 2015); Michael D. Graf et al.,

The ACA made broad changes to insurance coverage for preventive care in both the private and public sector. This section of the paper will examine the state of insurance coverage for genetic tests and medical interventions prior to implementation of the ACA, and the next section will discuss how the ACA altered the lay of the land.

A. Medicare and genetic testing

Historically, Medicare has had only limited coverage for preventive services. Yet, because it is the largest healthcare reimbursement system in the US, many private insurers look to Medicare for guidance on reimbursement policies and cost.[14] Medicare only covers services that are 'reasonable and necessary for the diagnosis or treatment of illness or injury',[15] but the law does not specify what rises to the level of 'reasonable and necessary'.[16] Genetic testing done in an asymptomatic individual for preventive purposes flies directly in the face of Medicare's rules because the system explicitly excludes coverage for tests for screening purposes 'that are performed in the absence of signs, symptoms, complaints, or personal history of disease or injury', unless there is specific statutory authorization.[17]

These statutory exceptions to the treatment rule have developed over time as Congress has codified coverage for several preventive services, such as mammograms, colonoscopies, prostate cancer screenings, and diabetes screenings.[18] This system of ad hoc coverage creates an onerous process to add exceptions,[19] especially given the current political landscape in Congress surrounding changes to healthcare. One recent advocate for change was the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS). It argued for statutory authorization that would allow Centers for Medicare and Medicaid Services (CMS) to cover genetic testing and services that meet evidence standards, viewing the testing as potentially beneficial for the Medicare population.[20] Perhaps acknowledging the difficulty in securing Congressional changes to the Medicare law, the SACGHS also offered a regulatory recommendation where the Health and Human Services (HHS) Secretary interpret 'personal history' of a disease to include 'family history' of a disease.[21] This interpretation would open the door for reimbursement of genetic tests, albeit by equating family history of disease with disease for the individual. In practice, this suggestion would only make the indistinct line between prevention and disease blurrier.

Reimbursement for interventions and services related to genetic testing, such as genetic counseling, is also foreclosed upon in the Medicare system. Medicare generally limits coverage for genetic counseling both through the screening exception and by its characterization of genetic counselors, not recognizing them as healthcare

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Genetic Testing Insurance Coverage Trends: A Review of Publicly Available Policies from the Largest US Payers, 10 Personalized Med. 235, 235 (2013).

14 SACGHS, *supra* note 13, at 4; Note that the SACGHS refers to preventive genetic tests as pre-dispositional.

15 42 U.S.C. 42 § 1395y(a)(1) (2012).

16 Michael J. DeBoer, *Medicare Coverage Policy and Decision Making, Preventive Services, and Comparative Effectiveness Research before and after the Affordable Care Act*, 7 J. Health & Biomed. L. 493, 504–05 (2011).

17 Medicare Program, 66 Fed. Reg. 58,788, 58,813 (2001) (to be codified at 42 C.F.R. pt. 410).

18 42 U.S.C. 42 § 1395y(a)(1); DeBoer, *supra* note 16, at 505.

19 Cynthia E. Boyd, *Medicare: It's Time to Talk About Changing It*, 19 Annals Health L. 79, 82 (2009) (explaining that the system of creating statutory exceptions on a one by one basis is 'not an easy feat').

20 SACGHS, *supra* note 13, at 4, 5.

21 *Id.*

professionals who can be paid directly.[22] Additionally, prophylactic surgery and other preventive interventions are not covered by Medicare, because they fail to meet the requirement for services 'reasonable and necessary' for treatment.

B. Medicaid and genetic testing

Medicaid, a health insurance program for low-income Americans, is funded by both federal and state governments and is managed by the states. This state level management leads to variation in coverage levels across the nation. Most states cover genetic testing of some kind, although the number and type of test vary.[23] Several states take into consideration whether the genetic test is used for the purpose of diagnosis, although, unlike Medicare, there is no universal ban against coverage for tests on asymptomatic individuals. Medicaid coverage for genetic tests can vary, not just geographically, but also temporally, depending on fluctuations in state Medicaid funding.[24] Genetic testing that identifies mutations in *BRCA* 1 and *BRCA* 2, genes associated with an increased risk of breast and ovarian cancer, are two of the more widely covered genetic tests for adults in Medicaid. Thirty-two states offer reimbursement for testing, although the specific criteria for coverage can vary, and may not provide coverage for asymptomatic individuals.[25]

C. Private insurance and genetic testing

Private health insurance companies in the US vary in the number and kind of genetic tests that they cover. Information on coverage for genetic testing is elusive because not all companies provide publicly available coverage determinations and distinct health insurance policies offered by a company may have different criteria than any broad policy guidelines indicate.[26]

(1) Genetic testing

Private insurance companies cover some genetic testing for adult onset conditions, both for prevention and diagnosis, but broad reimbursement for these tests has not occurred. Although it is difficult to gather extensive information regarding private insurance coverage, the general coverage criteria in publicly available plan policies include whether the tests would directly influence disease treatment management, were for diagnostic purposes, or were a preventive measure for high-risk patients.[27] Plans explicitly excluded genetic testing for informational purposes, for population screening without a family history of the condition, and for minors tested for adult onset conditions.[28]

Overall uptake of insurance coverage for genetic testing has been relatively slow for three main reasons. First, the private insurance market generally looks toward Medicare for guidance on coverage and reimbursement levels, but Medicare does not pay

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22 *Id.* at 50.
23 *Id.* at 32, 33.
24 *Id.*
25 Facing Our Risk of Cancer Empowered (FORCE), *Insurance, Financial Assistance, Cost of Services*, http://www.facingourrisk.org/info_research/finding-health-care/financial-help/ (accessed March 2, 2015).
26 Graf et al., *supra* note 13, at 241; SACGHS, *supra* note 13, at 17.
27 Graf et al., *supra* note 13, at 237; SACGHS, *supra* note 13, at 17, 18.
28 *Id.*; SACGHS, *supra* note 13, at 18.

for genetic testing and services given its strict rules against payment for prevention and recommended follow-up.[29]

Second, lack of clear evidence showing measurable medical benefits of testing, or clinical utility, creates a major bottleneck for insurance reimbursement.[30] If systematic reviews of a medical condition create guidelines cautioning against genetic testing, motivation for additional research and funding diminishes sharply, leaving no opportunity to develop competing or complementary evidence regarding the condition, even for a specific subset of the population. Additionally, lack of insurance coverage for a test minimizes public uptake thus reducing available evidence.

Third, standards of cost-effectiveness are often not met because genetic tests screen for diseases that are rare and thus have low prevalence in society.[31] Insurance companies may be particularly loath to pay for genetic testing versus diagnostic genetic testing because they are less likely to see any potential cost savings that would occur due to enrollee attrition rates in insurance plans over time.[32]

Despite the slow uptake of coverage for genetic testing, private insurance companies have broader and more varied reimbursement for genetic testing and services than public insurance programs. Variation occurs not just in the types of genetic tests covered, but between the specific criteria that determine coverage.[33] These coverage determinations, often made ad hoc, sometimes inaccurately reflect scientific information regarding the genetic conditions.[34] In result, available clinically relevant genetic tests are excluded 'because "no evidence of effectiveness" does not necessarily imply "evidence of no effectiveness"'.[35] Thus, there is a gap between sufficient evidence for clinical use and evidence for insurance coverage.

(2) Recommended interventions

The variable insurance coverage for genetic tests creates a cascade of ad hoc coverage decisions for recommended interventions after individuals have been tested—creating an equally variable system of coverage for interventions. The remainder of this paper will not concentrate on the issue of barriers to genetic testing coverage, but will focus on what the reimbursement policies are, and should be, after those genetic tests are covered.

Most policy guidelines within the industry strongly encourage genetic testing to occur concurrently with genetic counseling.[36] Numerous studies indicate that participating in genetic counseling prior to and after genetic testing lowers the risk of many

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29 See *supra* part II.A.

30 National Human Genome Research Instiute (NHGRI), *Reimbursement Models to Promote Evidence Generation and Innovation for Genomic Tests. Workshop Summary* (2012), http://www.genome.gov/27552210 (accessed March 3, 2015); Khoury et al., *supra* note 12, at 1606.

31 SACGHS, *supra* note 13, at 21,23.

32 See eg James P. Evans, *Health Care in the Age of Genetic Medicine*, 298 J. Am. Med. Ass'n 2670, 2671 (2007); See also Marc S. Williams, *Can Genomics Deliver on the Promise of Improved Outcomes and Reduced Costs*, 11 Disease Mgmt. Health Outcomes 277, 281–82 (2003).

33 See eg Megan Latchaw et al., *Health Insurance Coverage of Genetic Services in Illinois*, 12 Genet. Med. 525 (2010).

34 *Id.* at 529.

35 Wolf H. Rogowski et al., *Criteria for Fairly Allocating Scarce Health-Care Resources to Genetic Tests: Which Matter Most?*, 22 Eur. J. Hum. Genet. 25, 27 (2013).

36 See *infra* part III.C.

psychological side effects of testing, including anxiety and depression.[37] Further, genetic counseling prior to genetic testing leads to better risk perception among patients, a decrease in individual uptake of genetic testing, and a lower occurrence of unnecessary genetic testing.[38] Despite the documented benefits of genetic counseling, reimbursement for genetic counseling is spotty. Insurance companies do not universally pay for counseling for a variety of reasons, including that genetic counselors are not licensed in every state, they are not recognized as preferred providers by all insurance companies, and counseling in general is not a covered benefit.[39]

Interventions after a positive genetic test can be costly, especially if the recommended course of action is a prophylactic surgery; therefore, health insurance companies have monetary incentives to deny reimbursement of preventive interventions.[40] By definition, genetic services thwart negative medical outcomes from occurring in the future. However, as with reimbursement for genetic testing, insurance companies do not have an incentive to pay for costly prevention such as prophylactic surgery when the average individual stays with his or her health insurance company for less than six years.[41] For example, if a 25-year-old woman elects to have a prophylactic mastectomy, her health insurance company may not reap any cost savings by paying for the expensive surgery because she would likely switch companies during the 10 to 40 years when she would have been likely to develop breast cancer.

Choosing the appropriate intervention is inherently an individualized decision based upon the associated risk, family planning concerns, and other socio-economic considerations. Coverage decisions for preventive medical interventions can be dependent upon what is 'medically necessary' for the individual.[42] For this reason, until coverage for an intervention has garnered enough evidence to show clinical utility for a broad swath of the population, individuals may need to resort to insurance appeals or litigation in order to secure coverage.

(3) Prevention/treatment dichotomy and insurance appeals

As discussed above, the line between prevention and treatment has been consistently pushed earlier and earlier in the manifestation of a disease.[43] The current system of insurance coverage, ignoring susceptibility and asymptomatic states of illness, incentivizes this movement. Take, for example, the development of hemochromatosis—a hereditary condition that causes excess iron to accumulate in the body.[44]

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37 See eg Arwen H. Pieterse et al., *Longer-Term Influence of Breast Cancer Genetic Counseling on Cognitions and Distress: Smaller Benefits for Affected Versus Unaffected Women*, 85 PATIENT EDUC. & COUNSELING 425, 430 (2011).

38 See eg Chris M.R. Smerecnik et al., *A Systematic Review of the Impact of Genetic Counseling on Risk Perception Accuracy*, 18 J. GENET. COUNSELING 217 (2009).

39 See eg Tabitha A. Harrison et al., *Billing for Medical Genetics and Genetic Counseling Services: A National Survey*, 19 J. GENET. COUNSELING 38, 40 (2010); Mark A. Rothstein & Sharona Hoffman, *Genetic Testing, Genetic Medicine, and Managed Care*, 34 WAKE FOREST L. REV. 849, 876 (1999).

40 Rothstein & Hoffman, *supra* note 39, at 878, 879.

41 Evans, *supra* note 32, at 2671; Williams, *supra* note 32, at 281, 282.

42 See eg Alexandra K. Glazier, *Genetic Predispositions, Prophylactic Treatments and Private Health Insurance: Nothing Is Better Than a Good Pair of Genes*, 23 AM. J.L. & MED. 45, 48 (1997).

43 See *supra* part I.B.

44 National Heart, Lung, and Blood Institute, *How Is Hemochromatosis Diagnosed?*, http://www.nhlbi.nih.gov/health/health-topics/topics/hemo/diagnosis (accessed March 3, 2015); Mayo Clinic

An individual with a family history of hemochromatosis can have genetic testing to determine if he/she has inherited the mutations that significantly increase his/her risk for the disease. Physicians can detect an elevated iron level through two biochemical tests—serum transferrin saturation and serum ferritin. These tests are performed in order to determine if an individual has begun to build up iron levels, which will eventually lead to organ damage, since not all individuals with hereditary hemochromatosis will develop symptoms. If iron levels are sufficiently elevated, it is recommended that the individual go through routine phlebotomy—basically removing the excess iron along with the blood. If more severe damage, such as liver disease or cirrhosis, has already occurred, a physician can set up a treatment plan to minimize the progress and damage of the disease by more frequent phlebotomies as well as other tests that monitor the extent and progression of organ damage. Thus, in the progression of hemochromatosis, there are at least four possible interventions that can occur in various combinations—genetic testing, iron tests, phlebotomy, and management of liver disease. Where along this continuum do the interventions cross the line from prevention to treatment? Most would agree that management of liver disease is clearly treatment, but are serum measurements of iron levels or removal of excess iron via phlebotomy, best categorized as treatment or as secondary prevention? To secure reimbursement, clinicians and patients may be forced to argue that they are doing treatment and appeal any denial of coverage.

Patients with private insurance have more options to appeal than patients with public plans, where Medicare's statutory ban on preventive services makes appeal impossible and Medicaid's bureaucratic requirements complicate appeals.[45] Private insurance companies can make coverage determinations for individual patients because insurance policies generally have a broad policy to cover 'medically necessary' services for their policyholders. If an intervention is denied, it is likely either because the insurer has broad coverage language exempting preventive services from coverage or because the insurance company argues that it is not medically necessary. Insurance appeals in the genetic testing arena often boil down to whether there is enough evidence that interventions associated with a positive result are medically necessary.[46]

Although appeals have been employed successfully by a number of individuals to ensure coverage for interventions, the system forces genetic services into several conflicting legal classifications simultaneously depending upon the context. The underlying subcontext to the medically necessary category is that it is necessary to address or treat a specific disease. This is much like Medicare's 'reasonable and necessary' clause. Arguing that a preventive service is medically necessary inherently implies that there is a genetic condition that constitutes a medically actionable disease.[47] However, the Genetic Information Nondiscrimination Act (GINA), which was passed in 2008 to ban genetic

Hemochromatosis: Tests & Diagnosis, http://www.mayoclinic.org/diseases-conditions/hemochromatosis/basics/tests-diagnosis/con-20023606 (accessed March 3, 2015).

45 See eg MaryBeth Musumeci, *A Guide to the Medicaid Appeals Process*, Kaiser Family Foundation, 2012, http://www.scdd.ca.gov/res/docs/pdf/Whats_New/KaiserGuidetoMedicaidAppealsProcess.pdf (accessed March 3, 2015).

46 See eg Glazier, *supra* note 42, at 48.

47 See eg Katskee v. Blue Cross Blue Shield of Neb., 515 N.W.2d 645 (Neb. 1994) (holding that having a genetic risk for breast and ovarian cancer constituted a disease for purposes of insurance and, therefore, prophylactic surgery was a medically necessary procedure); Glazier, *supra* note 42, at 49, 51.

discrimination in health insurance and employment, clarifies that genetic information, in the absence of symptoms, cannot be considered a pre-existing medical condition.[48] GINA did not address interventions for genetic conditions, either in the context of pre-existing conditions or treatment for disease. Patients and healthcare providers continue to argue that preventive interventions are necessary for a medically actionable disease post-GINA, but this requires legal acrobatics where genetic conditions are explicitly not disease in one context and explicitly disease in another.

(4) Insurance coverage variability: BRCA as a case example

HBOC caused by mutations in the genes *BRCA* 1 and *BRCA* 2 is one of the most widely known and studied genetic conditions, yet insurance companies offer varying levels of insurance coverage for genetic test and the recommended interventions. The ACA altered the landscape for private insurance coverage of BRCA testing, but these changes will be discussed in depth later in the paper.[49] This section lays out insurance coverage for BRCA testing and interventions prior to the ACA.

BRCA 1 and *BRCA* 2 were identified in 1994 and 1995, respectively.[50] Over ten years later, the USPSTF examined testing for these two genes and found sufficient evidence that it had clinical utility for women with a family history of breast and ovarian cancer.[51] The USPSTF is a panel of independent scientists that examines medical evidence and makes recommendations for the clinical use of preventive methods.[52] These recommendations came in 2005, after BRCA testing was already an established practice in the clinical setting.[53] The interventions available to individuals with a positive BRCA test range from screenings, such as mammography at an earlier age than the general population, to prophylactic surgery, such as a preventative mastectomy or oophorectomy.[54] Myriad Genetics held patents for the testing for *BRCA* 1 and *BRCA* 2 tests until 2012 when the Supreme Court invalidated these rights.[55] Therefore, prior to the ACA, individuals in the US could only get clinical testing through Myriad.

Prior to the ACA, insurance coverage for BRCA testing was by no means guaranteed across all insurance companies. Criteria for coverage levels were based upon personal history of cancer, family history, age, and ethnicity.[56] Similar to Medicare, some private insurance companies only covered tests if an individual was already diagnosed with cancer. Due to variable coverage, Myriad Genetics required patients to provide confirmation of insurance coverage prior to testing in order to mitigate lost profits from

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48 Genetic Information Nondiscrimination Act (GINA), Pub. L. No. 110–233, 122 Stat. 881(2008) (codified as amended in scattered sections of 26, 29, and 42 U.S.C).

49 See *infra* part III.C.

50 National Human Genome Research Institute (NHGRI), *Questions About the BRCA1 and BRCA2 Gene Study and Breast Cancer* (2012), https://www.genome.gov/10000940 (accessed March 3, 2015).

51 *Genetic Risk Assessment and BRCA Mutation Testing for Breast and Ovarian Cancer Susceptibility,* 143 Annals Int. Med. 355 (2005); Since 2005, the USPSTF has reexamined BRCA testing and republished their recommendations in 2013.

52 *About the USPSTF,* USPSTF, http://www.uspreventiveservicestaskforce.org/about.htm (accessed March 3, 2015).

53 Khoury et al., *supra* note 12, at 1604, 1605.

54 NHGRI, *supra* note 50.

55 Ass'n for Molecular Pathology, et al., v. Myriad Genetics, Inc., et al., *133 S.Ct. 2107* (2013).

56 See eg Grace Wang et al., *Eligibility Criteria in Private and Public Coverage Policies for BRCA Genetic Testing and Genetic Counseling*, 13 Genet. Med. 1045, 1046–47 (2011).

unreimbursed tests.[57] Those who were underinsured and could not afford to pay out of pocket were often unable to get access to the test. Indeed, cost, which ranges from several hundred dollars to test for a specific gene variant to several thousand dollars for the entire gene, remains one of the most cited reasons that an individual chooses not to get BRCA testing.[58] Myriad offered uninsured individuals financial assistance, but not those with insurance. The company has since closed this gap for the underinsured by expanding their patient assistance program to include individuals with insurance whose policies do not completely cover the costs of the test.[59]

Insurance coverage for genetic services following BRCA testing, such as genetic counseling, screening, and prophylactic surgery, is even less of an assurance than insurance coverage for the genetic test itself. Access to medical interventions is not only a question of access to insurance. Even among individuals with insurance, such as in Massachusetts, which has mandated health coverage, reimbursement for preventive care is not guaranteed.[60]

Access to screenings as a preventive measure following BRCA testing is often possible for women at older ages, but can be difficult for those at younger ages. For example, at the population level, women are recommended to get mammograms between ages 40 and 50. However, physicians recommend that women at high risk of breast cancer begin regular mammograms at the age of 10 years younger than their youngest relative was diagnosed with cancer.[61] Since *BRCA*-related cancers commonly occur at young ages, this can lead to recommended screenings as early as a woman's twenties.

Many states have legislation that mandates insurance coverage for mammograms; however, these laws generally only require coverage for women over 40 or 50 based on evidence of clinical utility for the general population.[62] Some states, such as Illinois, have an exception to the age requirement when there is a family history of cancer or other indication of high risk.[63] These state laws do not apply to all private insurance policies because self-funded employer sponsored plans—those which pay all healthcare costs of employees rather than premiums to an insurance company—are exempted from state law.[64] For those states that do not have exceptions to their age limits, insurance companies can opt not to cover mammograms for women of younger ages even if it is a medically recommended course of action. In these situations, a woman will likely

57 SACGHS, *supra* note 13, at 46 n. 56.

58 Dawn C. Allain et al., *Consumer Awareness and Attitudes About Insurance Discrimination Post Enactment of the Genetic Information Nondiscrimination Act*, 11 Familial Cancer 637, 640 (2012).

59 Myriad, *Myriad Expands Financial Assistance Program to Underinsured Patients*, GenomeWeb Daily News Jul. 15, 2013, http://www.genomeweb.com/clinical-genomics/myriad-expands-financial-assistance-program-underinsured-patients (accessed March 3, 2015).

60 Jeanne Erdmann, *Is Your State Legislature Waiting for You to Get Cancer?* Slate Medical Examiner Dec. 25, 2013.

61 See Jennifer K. Litton et al., *Earlier Age of Onset of BRCA Mutation-Related Cancers in Subsequent Generations*, 118 Cancer 321, 323 (2012).

62 American Cancer Society, *Paying for Breast Cancer Screening*, http://www.cancer.org/cancer/breastcancer/moreinformation/breastcancerearlydetection/breast-cancer-early-detection-paying-for-br-ca-screening (accessed March 3, 2015); Note—after the ACA, insurance coverage is also mandated through federal law, but also has an age limit of 40. The clinical validity of mammograms is a highly debated subject, but state and federal legislation tend to follow recommendations beginning at age 40.

63 *Id.*

64 Employee Retirement Income Security Act (ERISA), Pub. L. No. 93–406, 88 Stat. 829 (1974) (codified as amended in scattered sections of 5, 18 , 26 , 29 and 42 U.S.C.).

have to appeal and argue that the screening should be covered as a medically necessary procedure.

Prophylactic surgery, both mastectomy and oophorectomy, is covered by several insurance companies, but was, and still is, not a widespread practice. A 2000 study showed that explicit coverage for a prophylactic surgery among private health plans ranged from 24 to 43% depending on the type of surgery and the clinical circumstance.[65] Fourteen per cent of plans had an explicit policy of non-coverage for prophylactic mastectomy based on a strong family history and 10% non-coverage based on the identification of a *BRCA* mutation.[66] These numbers were similar for a prophylactic oophorectomy—15 and 10%, respectively.[67] The remaining plans made coverage determinations on a case-by-case basis. These ad hoc determinations could vary from patient to patient and could require an appeal to ensure coverage.[68] It is likely that more insurance companies would currently cover preventive surgery given the increased knowledge of the clinical utility of the surgery. The study, however, remains an important reminder of the variability of insurance coverage and the lengthy odyssey until insurance companies adopt broad coverage policies for preventive medical interventions already clinically in use.

The blurred boundaries between treatment and prevention in the case of *BRCA* put some women in a difficult 'Catch-22'. A number of insurance companies have denied women health insurance or charged higher premiums in part because they had undergone a prophylactic mastectomy. These insurance companies viewed the surgery itself, not *BRCA* status, as a pre-existing condition. Although there is a strong argument that this would be proxy genetic discrimination under GINA, these denials were never tested in court and are now a moot point since insurance companies can no longer take any pre-existing conditions into account due to the ACA.

Testing for *BRCA* is arguably among the most widely utilized and widely known genetic tests available. However, even this oft-studied genetic test continued to have gaps in insurance coverage prior to the ACA—and perhaps surprisingly, also after.[69] The variability of private insurance coverage for BRCA testing and its related interventions highlights the difficulties of translating genetic findings of clinical utility, not just to clinical care, but also to standard reimbursement policies. Insurance companies often do not agree on the clinical utility of a test, and there is a general unwillingness to accept the upfront costs of prevention without guaranteed cost savings in the future.

III. THE STATE OF INSURANCE COVERAGE POST-ACA

The ACA made numerous, substantial changes related to prevention services in the healthcare system in the US. Two broad changes in particular alter the realm of insurance reimbursement for genetic testing and interventions. First, private insurance companies must cover, free of cost to the patient, any prevention method that the USPSTF has reviewed and recommended. Second, all health insurance plans offered in the state private markets must cover ten essential health benefits, including preventive and

65 Henry M. Kuerer et al., *Current National Health Insurance Coverage Policies for Breast and Ovarian Cancer Prophylactic Surgery*, 7 Annals Surgical Oncology 325, 327–29 (2000).

66 *Id.*

67 *Id.*

68 *Id.*

69 See *infra* part III.C. for a discussion of coverage post-ACA.

wellness services. Although these changes present an opportunity for expanded coverage of genetic tests, their framework threatens to create disparate access to services if the law is not implemented with comprehensive access in mind.

A. USPSTF and the ACA

The ACA greatly expands access to preventive services by mandating private insurance companies to reimburse completely for select evidence-based services. Specifically, immunizations recommended by the Advisory Committee on Immunization Practices of the Centers for Disease Control and Prevention, preventive care and screenings discussed in guidelines of the Health Resources and Services Administration, and services rated as an A or a B by the USPSTF must be covered.[70] Beginning on September 23, 2010, insurance companies were required to cover these preventive services with no cost sharing, so the patient does not have to pay coinsurance, copay, or any amount of a deductible.[71]

The USPSTF makes recommendations regarding preventive measures related to genetic testing results and other screening measures. It was established in 1984 and has been associated with the Agency for Healthcare Research and Quality since 1995. The mission of the USPSTF, an independent panel of medical experts, is:

> assessing the benefits and harms of preventive services in people asymptomatic for the target condition, based on age, gender, and risk factors for disease and making recommendations about which preventive services should be incorporated routinely into primary care practice.[72]

When the task force reviews a service, they assign a grade—A, B, C, D, or I—depending on the recommendation.[73] Grades A and B are recommendations for the service because there is high certainty that the net benefit is substantial or moderate or because there is moderate certainty that the net benefit is substantial. If the USPSTF recommends against the service or if there is insufficient evidence to make a determination, the service is given a D or I rating, respectively. Grade C is currently given if 'the USPSTF recommends selectively offering or providing this service to individual patients based on professional judgment and patient preferences'—although the exact description for this rating level has changed over the past decade.[74]

The ACA's expansion of coverage for preventive services is a first step toward refocusing the US healthcare system toward prevention and has been hailed by many advocacy groups as a beneficial and important part of healthcare reform. However, in addition to the substantial benefits of the changes, there are at least two crucial, systemic gaps.

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70 Coverage of Preventive Health Services, 29 C.F.R. § 2590.715–2713 (2011).

71 Kaiser Family Foundation, *Preventive Services Covered by Private Health Plans under the Affordable Care Act*, (2011), http://kaiserfamilyfoundation.files.wordpress.com/2013/01/8219.pdf (accessed March 3, 2015).

72 U.S. Preventive Services Task Force, *Procedure Manual* (2011), http://www.uspreventiveservicestaskforce.org/uspstf08/methods/procmanual.htm (accessed March 3, 2015).

73 U.S. Preventive Services Task Force, *Grade Definitions*, http://www.uspreventiveservicestaskforce.org/uspstf/grades.htm (accessed March 3, 2015).

74 *Id.*

First, the ACA requirements regarding coverage of preventive care do not apply consistently across all health insurers. In the private market, mandated coverage of preventive services does not apply to grandfathered health insurance policies.[75] Grandfathered health plans are those that were established prior to March 23, 2010—the day that Congress passed the ACA—that have not made significant changes to benefits or consumer costs.[76] For group plans, the relevant date is when the plan was established, not when the individual first enrolled in a plan. For example, an employer sponsored plan that was established in 2005 may be grandfathered, even if the individual started working for the company in 2013. Most individuals in the private insurance market receive health insurance through an employer and a large percent of employer-sponsored plans are grandfathered.[77] Therefore, a significant portion of privately insured individuals does not have access to the free preventive services established by the ACA. However, this number is expected to decrease as plans lose their grandfathered status after making policy changes. Indeed, the current percentage of grandfathered plans is 26%, down from 56% in 2011.[78] A grandfathered status does not necessarily mean that the preventive services will not be covered by the health plans, but coverage is not guaranteed and the patient will be responsible for deductibles, copays, or coinsurances.

In addition to inconsistent application in private plans, the rules regarding preventive services also do not apply to public services such as Medicaid and Medicare. The ACA expanded the list of preventive services covered under Medicare and made several preventive services free of cost to the patients.[79] Similarly to the private market, preventive services rated by the USPSTF as an A or a B are generally covered by Medicare. However, the ACA gives the CMS discretion as to which USPSTF recommendations are included in coverage.[80] Thus, the final list of preventive services covered by Medicare is smaller than the private insurance list. Most notably absent is coverage for *BRCA* counseling and testing.[81]

Second, the framework and procedures of the USPSTF create gaps in how the preventive services recommendations are applied. Overall, the USPSTF is unabashedly clear in its limited scope and purpose—'the Task Force's scope is specific: its recommendations address primary or secondary preventive services targeting conditions that represent a substantial burden in the Unites States and that are provided in primary care settings or available through primary care practice'.[82] The USPSTF does not examine preventive measures that are part of treatment or management of an already manifested condition.[83]

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75 Coverage of Preventive Health Services, 29 C.F.R. § 2590.715–2713 (2011).

76 Preservation of Right to Maintain Existing Coverage, 29 C.F.R § 54.9815–1251T (2010).

77 The Kaiser Family Foundation and Health Research and Educational Trust, *Employer Health Benefits: 2014 Annual Survey,* 2014, http://kff.org/report-section/ehbs-2014-summary-of-findings/ (accessed March 3, 2015).

78 *Id.* at 255.

79 AARP Public Policy Institute, *Improvements to Medicare's Preventive Services Under Health Reform,* 2010, http://assets.aarp.org/rgcenter/ppi/health-care/fs180-preventive.pdf (accessed March 3, 2015).

80 PPACA § 1404, 42 U.S.C. § 1395m(n)(1)(A).

81 Centers for Medicare and Medicaid Services, *Preventive & Screening Services,* http://www.medicare.gov/coverage/preventive-and-screening-services.html (accessed March 3, 2015).

82 USPSTF, *supra* note 72, at § 1.3.

83 *Id.*

This focus—an emphasis on preventive services for asymptomatic individuals—creates complications when applied to clinical care and insurance coverage because it excludes individuals with past or current symptoms of disease. For example, if a woman in remission for breast cancer goes to her physician for an annual mammogram, the doctor is likely to code it as diagnostic in order to ensure that a recurrence of cancer may be found. This diagnostic exam is not covered as a preventive service under the ACA, even though society may conceptually think of it as a mammogram to prevent a recurrence of cancer.

There have also been complications applying USPSTF recommendations in the insurance setting even when the individual's situation appears to fall squarely within the recommended circumstance. For example, the USPSTF gives preventive colon cancer screenings for individuals between the ages 50 and 75 an A rating.[84] After implementation of the ACA, insurers began to cover colonoscopies free of cost to patients. However, it is standard of care in a preventive colonoscopy to remove polyps discovered during the screening and test to determine if they are cancerous. In these cases, several insurance companies argued that the polyp removal was therapeutic and therefore cost sharing was imposed.[85] This circular reasoning undermined the entire purpose of mandating payment for preventive colonoscopies. Removal of polyps before they become cancerous is the way to prevent colon cancer through secondary prevention. Preventive colonoscopies simply provide the opportunity for this. However, it was in these precise instances that the insurance companies were charging the patients. In February 2013, the federal government clarified that an insurance company cannot impose cost sharing in the event that a polyp is removed during a preventive colonoscopy.[86] Even after this clarification, patient's confusion remains regarding the frequency of colonoscopies and distrust that insurance companies will follow the guidance.[87]

B. Ten essential health benefits under the ACA

Under the 'individual mandate', the ACA requires most individuals in the US to purchase health insurance beginning in 2014.[88] To facilitate this, healthcare reform created online aggregate insurance marketplaces in each state, called exchanges, in order to help individuals choose insurance policies. Every plan sold through an exchange must offer essential health benefits in order to be considered comprehensive coverage.[89] The ACA delineated ten broad categories of essential health benefits and tasked the HHS Secretary to define these benefits in greater detail during the regulatory period. The ten categories are ambulatory patient services, emergency services,

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84 Ned Calonge et al., *Screening for Colorectal Cancer: US Preventive Services Task Force Recommendation Statement*, 149 Annals Int. Med. 627 (2008).

85 Karen Pollitz et al., *Coverage of Colonoscopies under the Affordable Care Act's Prevention Benefit*, Kaiser Family Foundation, American Cancer Society, and the National Colorectal Cancer Roundtable, Aug. 31, 2012.

86 U.S. Department of Labor, *FAQs About Affordable Care Act Implementation Part XII*, http://www.dol.gov/ebsa/faqs/faq-aca12.html#6 (accessed March 3, 2015); Michelle Andrews, *Questions About Colon Screening Coverage Still Vex Consumers*, Kaiser Health News, http://www.kaiserhealthnews.org/Features/Insuring-Your-Health/2013/041613-Michelle-Andrews-preventive-colonoscopy-costs.aspx (accessed March 3, 2015).

87 Andrews, *supra* note 86.

88 PPACA § 1501, 26 U.S.C. § 5000A.

89 PPACA § 1302, 42 U.S.C. § 18022.

hospitalization, maternity and newborn care, mental health and substance use disorder services, prescription drugs, rehabilitative and habilitative services, laboratory services, pediatric services, and, most importantly for the realm of genetic testing, preventive and wellness services.[90]

It was expected that HHS would establish a national standard for these essential health benefits, but the agency opted to punt the question to the states. Under HHS guidance, states can choose a benchmark plan that serves as a reference plan for the other insurance policies in the state.[91] This decision was widely criticized by health advocates because it creates bifurcated levels of coverage across the nation and codifies current inadequate insurance standards as acceptable coverage.[92] On the other hand, because the guidance minimized the changes that insurance plans would have had to adhere to under a broad national standard, health insurance companies applauded the state-by-state implementation.[93]

The establishment of the state-by-state benchmark plan also limits the ability to centralize standards for new preventive services. Given the healthcare industry's treatment focus, any chosen benchmark plan is unlikely to noticeably expand coverage for wellness and preventive services. Although the ACA broadened prevention coverage *without cost-sharing* through the USPSTF, the range of preventive services that should be covered by the insurance companies *with cost-sharing* is now dependent upon existing state mandates and the state's benchmark plan. Thus, the state of insurance coverage for genetic services, especially interventions, is unlikely to greatly alter from the situation prior to the ACA.

C. *BRCA* as a case example post-ACA

The USPSTF reviewed genetic testing for HBOC in both 2005 and 2013. When the panel first reviewed HBOC, it recommended 'women whose family history is associated with an increased risk for deleterious mutations in *BRCA*1 or *BRCA*2 genes be referred for genetic counseling and evaluation for BRCA testing'.[94] Subsequently, the 2013 recommendation statement made several key changes to the earlier version. These changes came after the implementation of the ACA and were likely responses to public concerns and insurance coverage ambiguities created by merging the USPSTF recommendations with insurance coverage mandates.[95] The narrow group of individuals to which the USPSTF guideline applies has been altered or clarified in several instances between the 2005 and 2013 recommendations. There are four specific ways that the prior USPSTF recommendations excluded individuals from consideration and most were readdressed in the 2013 guideline versions.

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90 PPACA § 1302, 42 U.S.C. § 18022(b)(1).

91 Center for Medicare and Medicaid Services, *Center for Consumer Information and Insurance Oversight*, Dec. 16, 2011.

92 Health Affairs and Robert Wood Johnson Foundation, *Health Policy Brief: Essential Health Benefits,* 2013, http://healthaffairs.org/healthpolicybriefs/brief_pdfs/healthpolicybrief_91.pdf (accessed March 3, 2015); See also Jennifer P. Ruger, *Fair Enough? Inviting Inequities in State Health Benefits*, 366 New Eng. J. Med. 681, 681–82 (2012).

93 Health Affairs and Robert Wood Johnson Foundation, *supra* note 92, at 3.

94 U.S. Preventive Services Task Force (USPSTF), *Genetic Risk Assessment and BRCA Mutation Testing for Breast and Ovarian Cancer Susceptibility: Recommendation Statement*, 143 Annals Int. Med. 355 (2005).

95 Virginia A. Moyer, *Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer in Women: US Preventive Services Task Force Recommendation Statement*, 160 Annals Int. Med. 271 (2014).

First, the recommendation statement is gender specific in both the 2005 and 2013 versions, and men are explicitly excluded from the evidence synthesis. Thus, insurance companies are not mandated to cover BRCA testing in men, and if they do, can require copays and coinsurance. Omission from guideline inclusion is dangerous if it is interpreted to mean that the procedure is not recommended for this population—for example that BRCA testing is never clinically valid in men. In reality, there may be sufficient evidence that testing is clinically relevant for men on an individual basis. The system created by the ACA may make it difficult for this population to navigate insurance coverage and appeals.

Second, the BRCA testing guidelines do not apply to women who have a diagnosis of breast or ovarian cancer. Examining evidence of interventions for diagnostic purposes is outside of the USPSTF scope.[96] While this has always been clear from the mission and delineated procedures of the panel, the 2013 recommendation statement made this unambiguously clear within the recommendation itself. The 2013 recommendations further suggested that women in remission who had not received BRCA testing as part of their care, but who have a family history, speak to their doctor about the possibility of testing.[97] However, this statement has no force and is only encouragement since the official scope of the review only includes asymptomatic individuals with no personal history of cancer.

Third, the panel completely reversed its position on whether the recommendations apply to women with a relative who has a *known* deleterious *BRCA* mutation. In 2005, these women were explicitly excluded from the recommendations.[98] The 2013 version, however, noted 'women who have 1 or more family members with a known potentially harmful mutation in the *BRCA1* or *BRCA2* genes should be offered genetic counseling and testing'.[99]

Fourth, the panel bifurcated the female population and gave a D grade for genetic testing for women whose family history is not associated with an increased risk for breast and ovarian cancer. Due to this recommendation, women with no family history of breast or ovarian cancer are unlikely to obtain insurance coverage for BRCA testing. Between 2005 and 2013 the USPSTF altered the threshold of family history needed to identify a woman as high risk. The guidelines in 2005 included very specific delineations for which familial patterns were associated with cancer risk. The recommendation statement specifically listed several combinations of number of diagnoses, age of onset, ethnicity, and cancer type as evidence of high risk.[100] However, by 2013, the USPSTF shied away from specifics and instead refer to several risk assessment tools that clinicians can use with their patients.[101] Although a generally extensive family history of breast cancer is still required to reach the recommendation thresholds, the change by the USPSTF shows an inclination to leave more room for interpretation in the hands of the clinicians. This will hopefully limit disputes between insurance companies and doctors over whether the family history is sufficient to warrant insurance reimbursement.

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96 See *supra* part III.A.

97 Moyer, *supra* note 95, at 3.

98 USPSTF, *supra* note 94, at 356.

99 Moyer, *supra* note 95, at 3.

100 USPSTF, *supra* note 94, at 356.

101 Moyer, *supra* note 95, at 3, 4.

The gray area of what insurance companies was required to cover for high-risk women was immediately apparent when the preventive service provisions of the ACA went into effect. The ensuing debate highlighted the inherent tension between the insurance groups trying to minimize expenditures and cover a minimal amount of services free of cost, versus advocacy groups trying to push for broad interpretation. The 2005 USPSTF guidelines did not explicitly state that genetic testing was recommended for high-risk women; therefore, insurance companies questioned whether they needed to pay for only the referral for genetic counseling, for the counseling itself, or for BRCA testing.[102] As in the case of colonoscopies, the federal government clarified that the USPSTF recommendation encompassed both genetic counseling and BRCA testing.[103] When the USPSTF reexamined *BRCA* in 2013, the recommendation grade remained at the B level, but the recommendation statement was expanded to provide more explicit guidance.

"The USPSTF recommends that primary care providers screen women who have family members with breast, ovarian, tubal, or peritoneal cancer with one of several screening tools designed to identify a family history that may be associated with an increased risk for potentially harmful mutations in breast cancer susceptibility genes (*BRCA1* or *BRCA2*). Women with positive screening results should receive genetic counseling and, if indicated after counseling, BRCA testing."[104]

The examples of insurance coverage for colonoscopies and BRCA testing post ACA mandates illustrate both the creative interpretations insurance companies make to limit expenditures and shift cost sharing back to the patient and the poor fit of USPSTF recommendations as insurance coverage guidelines—'because these recommendations, developed by panels of clinicians for use by other clinicians, were never intended to be used for coverage purposes, they lack the precision of standards developed expressly for the purpose of providing coverage of healthcare services'.[105]

Changes to the BRCA testing recommendations likely predict that future USPSTF procedures will be altered to more clearly provide guidance for insurance coverage to avoid these interpretive quandaries in the future.[106] The *BRCA* example also shows that the difficulty in identifying which services in the individual's prevention plan should be included. For instance, because of the way that the recommendation was originally written, genetic counseling is included. It could have been written so that the test was clearly included, but not counseling. The interventions and follow-up that are recommended after BRCA testing are not discussed in the USPSTF report in enough detail to be included as mandated insurance benefits.[107]

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[102] Blue Cross and Blue Shield Association, *Comments on Interim Final Rules for Group Health Plans and Health Insurance Issuers Relating to Coverage of Preventive Services Under PPACA*, 2010.

[103] U.S. Department of Labor, *supra* note 86, at Q6.

[104] Moyer, *supra* note 95, at 1.

[105] Blue Cross and Blue Shield Association, *supra* note 102, at 2.

[106] At the time of writing, the USPSTF was in the process of updating its procedures manual.

[107] See *infra* part VI.A.2. for further discussion.

IV. ACMG POLICY CHANGES TO GENETIC INFORMATION FOR PREVENTION

A. Genomic sequencing technology

Expansion of medical technology outpaces any social policy intended to inform its use. Thus, the coverage for BRCA testing as a preventive measure under the ACA operates within the complexity of both the health care system and the interpretation of USPSTF recommendations. A close look at genomic testing in the clinical setting makes clear the challenges for insurance reimbursement. Massive parallel sequencing technology, such as whole genome or whole exome sequencing, is utilized to identify sequence variants in an estimated 20,000–25,000 human genes, containing approximately three billion individual units called base pairs.[108] Whole genome sequencing is the process of reading and sequencing all of the base pairs in an individual's genome.[109] Whole exome sequencing is a targeted capture and sequencing of the parts of the genes that code for proteins—approximately 1% of the whole genome.[110] Mutations in the portion of genes that code for proteins are more likely to result in diseases and conditions of clinical significance than mutations elsewhere in the genome. Therefore, whole exome sequencing can be a cheaper way to capture information about disease, although there is a possibility of missing clinically important mutations due in part to the capture methods used in the process.[111]

Advances in medical technology and drastically reduced cost will soon make it cheaper to perform genomic sequencing than a series of single genetic tests. Scientists recently surpassed the threshold of sequencing an entire genome for $1000 in one day.[112] This herald's momentous progress since the Human Genome Project successfully sequenced the first human genome in 2003 for 2.7 billion dollars after thirteen years of commitment.[113] With this sign of progress comes the acknowledgement that interpretation of any sequence variants is still in its infancy. Further knowledge, of which genetic variants influence disease, is required before the potential of these technologies can be realized.

In the clinical setting, genomic sequencing is currently used most often for diagnosis, but will likely have implications for prevention as well, given the vast of amount of information potentially produced by sequencing methodologies. Within that data reside evidence relevant for diagnosis, as well as for unanticipated conditions, including about a patient's risk factors for other diseases.[114] As costs continue to plummet and scientific understanding of interpretation grows, clinicians may increasingly turn to genomic sequencing as a preventive measure.

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[108] Genetics Home Reference, *What is a Gene,* U.S. NATIONAL LIBRARY OF MEDICINE, 2014, http://ghr.nlm.nih.gov/handbook/basics/gene (accessed March 3, 2015).

[109] ACMG Board of Directors, *Points to Consider in the Clinical Application of Genomic Sequencing,* 14 GENET. MED. 759, 759 (2012).

[110] *Id.*

[111] *Id.*

[112] Paul Rincon, *Science Enters $1,000 Genome Era*, BBC, 2014, www.bbc.co.uk/news/science-environment-25751958 (accessed March 3, 2015).

[113] National Human Genome Research Institute, *The Human Genome Project Completion: Frequently Asked Questions*, 2010, http://www.genome.gov/11006943 (accessed March 3, 2015).

[114] See eg Isaac S. Kohane et al., *Taxonomizing, Sizing, and Overcoming the Incidentalome*, 14 GENET. MED. 399, 400 (2012).

B. ACMG and genomic sequencing

In a highly debated policy statement, the ACMG recommended that when undertaking whole genome and exome sequencing for clinical diagnostic purposes, laboratories should deliberately seek and return results to the ordering clinician for mutations in additional outlined genes that could provide a medical benefit to the patient when learned before the onset of symptoms. The ACMG list currently contains fifty-six gene mutations, which are associated with twenty-four conditions that meet the panel's criteria for clinical validity and utility, although the list will be amended as knowledge of genomics continues to advance.[115]

Under these recommendations, laboratories are responsible for actively seeking these medically actionable results. Classified as a type of incidental finding, these are 'incidental' only because they are 'alterations in genes that are not apparently relevant to a diagnostic indication for which the sequencing test was ordered'.[116] The ACMG felt that this subset of gene mutations was important to seek out because of their medical actionability. The ACMG recommendations originally directed the return of such findings, regardless of the preference of the patient to learn about the presence of these mutations and regardless of the age, medical condition, psychological state, or other considerations of the patient[117]; however, recent policy changes recommend offering patients an opportunity to opt out of having these medically actionable gene variants analysed.[118]

The genetic mutations included on the ACMG list were specifically chosen because of available preventive measures and association with long periods before symptoms develop. For example, *BRCA* 1 and *BRCA* 2 are among the genes included in part because individuals with deleterious mutations can take preventive measures such as screenings and preventive surgery. Another condition on the list, familial hypercholesterolemia, caused by a genetic mutation associated with high cholesterol levels and risk of coronary heart disease, is included because individuals can be placed on a medication regimen of statins to significantly lower the risks.

Regardless of the debate and ethics of divulging even medically actionable incidental findings, these disclosures are most likely to occur when an individual is asymptomatic *for the incidental conditions*, even though he or she is symptomatic, and underwent genomic testing, *for another clinical purpose*. These recommendations were not intended by the ACMG to become the official standard of care among laboratories and, by proxy, the ordering clinician;[119] however, commentators have noted that courts may use the recommendations as a tool to determine the standard of care in medical malpractice

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[115] Robert C. Green et al., *ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing*, 15 GENET. MED. 565, 569 (2013) (Note: since publication of the initial recommendation, the list of recommended genes has been modified, although the general principles remain).

[116] *Id.* at 566.

[117] *Id.* at 568.

[118] GenomeWeb Daily News, *Updated ACMG Recommendations to Allow Patient Opt out of Incidental Findings*, 2014, http://www.genomeweb.com/clinical-genomics/updated-acmg-recommendations-allow-patient-opt-out-incidental-findings (accessed March 3, 2015).

[119] Green et al., *supra* note 115, at 565 (noting that 'these recommendations are designed primarily as an educational resource for medical geneticists and other health-care providers to help them provide quality medical genetic services').

cases.[120] As laboratories begin to follow the ACMG guidelines, the disclosure of deleterious mutations in these fifty-six genes will become some of the first examples of genomic testing for prevention.

Insurance coverage for both the sequencing and any preventive interventions may be complicated. In its initial discussion of incidental findings, the ACMG noted, 'we do not know the implications that this may have on reimbursement for clinical sequencing'. Recommending the clinical reporting of incidental findings without consideration of insurance reimbursement for either the test or the interventions places the ACMG in a morally uncertain position because patients may be exposed to greater harms and society may be exposed to increased health disparities.[121] The ACMG concluded that population prevalence and possible interventions warranted prioritizing the duty to prevent harm over patient autonomy.[122] For a portion of patients, the reality of insurance coverage and costs will preclude them from accessing the suggested interventions, a situation that may lead to greater patient harm due to increased anxiety or other psychological concerns. Evidence based inquiries into the efficacy of genetic testing hinges on a delicate balance of harms and benefits. Making the assumption that all patients will have access to the interventions is likely to skew the data toward patient benefit without weighing the harms of learning about a genetic mutation for which one cannot access the preventive measures.

V. ETHICAL CONSIDERATIONS

The ACA, ACMG recommendations, and insurance reimbursement policies for genetic testing and follow-up care overall threaten to increase health disparities across the healthcare system. As genomic sequencing increasingly is used in clinical practice more patients will have information about preventable genetic risk. Financial considerations may allow only certain individuals to take effective action based on this knowledge.

Unfortunately, health disparities are rampant and entrenched in the US healthcare system and access to genetic testing is no exception.[123] For example, despite the recommendations of the USPSTF, BRCA testing was underutilized prior to the ACA, especially for racial minorities.[124] Genomic testing threatens to further stratify the population in very significant ways.[125] Disparate access to preventive interventions foreshadows a worrisome reality where certain segments of the population, who are able to pay

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120 John Conley, *The ACMG Screening Recommendations*, Genom. L. Rep., July 30, 2013, http://www.genomicslawreport.com/index.php/2013/07/30/the-amg-gene-screening-recommendations/ (accessed March 3, 2015).

121 Mark A. Rothstein, *The Case against Precipitous, Population-Wide, Whole-Genome Sequencing*, 40 J.L. Med. & Ethics 682, 685 (2012) (commenting that a 'revolution' in insurance reimbursement of genetic services is needed before proper individual care is received); Wylie Burke et al., *Recommendations for Returning Genomic Incidental Findings? We Need to Talk!*, 15 Genet. Med. 854, 857 (2013) (noting that additional costs associated with testing for 56 additional variants should not be generated without patient consent).

122 Green et al., *supra* note 115, at 568.

123 Muin J. Khoury, *Why We Can't Wait: A Public Health Approach to Health Disparities in Genomic Medicine*, Centers for Disease Control and Prevention: Genomics and Health Impact Blog, June 27, 2013, http://blogs.cdc.gov/genomics/2013/06/27/why-we-cant-wait/ (accessed March 3, 2015).

124 Douglas E. Levy et al., *Underutilization of BRCA1/2 Testing to Guide Breast Cancer Treatment: Black and Hispanic Women Particularly at Risk*, 13 Genet. Med. 349 (2011); Michael J. Hall & Olufunmilayo I. Olopade, *Disparities in Genetic Testing: Thinking Outside the BRCA Box*, 24 J. Clinical Oncology 2197 (2006).

125 SACGHS, *supra* note 13, at 55.

for services, are able to prevent the burden of certain diseases completely, leaving only those of lower socio-economic statuses to suffer.

Racial minorities, individuals with lower socio-economic status, and the uninsured are currently more likely to be diagnosed with later stage cancer and more likely to die from a cancer diagnosis.[126] These disparities will become even starker if only a privileged few are able to access preventive medical interventions after a positive genetic test. Insurance reimbursement for these preventive services will by no means completely eradicate disparate access. Lack of access to general physicians for referrals, out-of-pocket costs—even relatively small ones—, and low health literacy are only a few of the more general barriers that exist in the current system.[127] Some have argued that the rise of genetic technology mandates universal health coverage because of the threat to health disparities.[128] Short of drastic changes to the healthcare system, insurance coverage for preventive medical interventions is an option that helps to limit exacerbated health disparities associated with genomic testing.

The concerns of health disparities and insurance coverage for medical interventions raise broad questions of distributive justice. Policies that expand access to predictive genetic testing, but do not similarly address access to the medically recommended follow-up preventive interventions create an unjust distribution of health care services. I argue that there is a moral duty for policy makers, insurance companies, and advisory committees to consider access to, and reimbursement for, preventive interventions when making recommendations regarding coverage or access to predictive genetic tests.

Two of the principal theories of distributive justice in health care support this moral duty. First, Norman Daniels, under his Rawlsian theory of distributive justice, argues that a just health care system must provide individuals with a fair equality of opportunity.[129] Daniels maintains that individuals must have access to health care services that are 'needed to maintain, restore, or compensate' for the loss of normal species functioning[130] and emphasizes that this includes access both to treatment and prevention of disease, including genetic-related prevention.[131] Thus, failure to reimburse measures one can take to prevent a disease is unjust under this distributive justice theory.

Second, a luck egalitarianism model of distributive justice also supports social policies that guarantee reimbursement for preventive interventions. Under luck egalitarianism, it is unjust for an individual to have comparably worse health outcomes than others due to brute luck—ie due to situations out of one's control.[132] This holds true, whether the inequality in health arises from social factors or from natural factors, such as genetic risk.[133] Thus, luck egalitarianism would support access to predictive genetic testing for

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126 See eg National Cancer Institute, *Cancer Health Disparities*, http://www.cancer.gov/cancertopics/factsheet/disparities/cancer-health-disparities (accessed March 3, 2015); Cancer Action Network, Americian Cancer Society, *Cancer Disparities: A Chartbook*, 2009, http://action.acscan.org/site/DocServer/cancer-disparities-chartbook.pdf?docID=15341 (accessed March 3, 2015).

127 Cancer Action Network, Americian Cancer Society, *supra* note 126, at 2.

128 Evans, *supra* note 32, at 2670.

129 Norman Daniels, Just Health Care (1985); Ronald Bayer et al., In Search of Equity (1983).

130 Daniels, *supra* note 129, at 86.

131 *Id.* at 140,141; Dan W. Brock et al., From Chance to Choice: Genetics and Justice (2001) (noting that 'our analysis reinforces the case for a social obligation to provide genetic health services because of our reliance on an account of health care that places great importance on fair equality of opportunity'.).

132 Shlomi Segall, Health, Luck, and Justice (2009).

133 *Id.* at 109.

medically actionable conditions because it provides information about whether an individual is unlucky due to natural factors; But, the theory would also demand equal access to the preventive interventions so that those unlucky due to social factors have as equal an opportunity to prevent disease as those with the private means to do so.[134]

While both theories support a moral duty to address insurance reimbursement of preventive interventions following a genetic test, this claim raises several critiques and questions common to theories of distributive justice. First, there are limited health care resources in our society and expansion of coverage may lead to a reduction of benefits elsewhere in the system.[135] As our society begins to understand more about the association between genetic variants and disease, and preventive measures, the sheer volume of potential genetic services could overwhelm the health care and insurance system.[136] Second, requiring equality in society threatens to lead to a 'leveling down' approach—ie, one can make the less fortunate equal to the fortunate by either providing services to those worse off, or preventing the fortunate from accessing services.[137] Reducing the number of predictive genetic tests available across society overall would equalize concerns of access, but with perverse results. Prohibiting all individuals from undergoing predictive genetic or genomic testing is by no means a practical suggestion or sound policy.

Although these critiques and concerns are important discussions to have in the broader debate over how to incorporate genomic technologies into theories and practices of just health care, it is beyond the scope of this paper to tackle these larger questions. This paper addresses a relatively narrow piece of the distributive justice debate: What is the duty of policy-makers and advisory boards to consider the sequelae of interventions and medical procedures when these groups are considering access to genetic testing for a particular genetic variant? Although resource concerns may follow from mandating coverage for a broad range of services, I argue that the initial cost/benefit considerations regarding access to the genetic test must include attention to the follow-up interventions.

These considerations may end up saving resources in the future if the preventive services are more cost effective than the resultant disease. Mandating insurance reimbursement for interventions may also address the collective action problem where, due to the turnover in the insurance market, paying for even cost-effective measures may not be logical for one insurance company on its own. The following section discusses possible policy mechanisms available to increase the chances for equal opportunity in this area.

VI. POLICY MECHANISMS

As insurance companies consider reimbursement policies for predictive genetic tests and as advisory groups and state and federal governments weigh in on which genetic tests should be covered, these groups also must also consider coverage for the

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134 *Id.* at 121.

135 Amy Gutmann, *For and against Equal Access to Health Care*, in IN SEARCH OF EQUITY (Ronald Bayer, et al. eds., 1983), at 61.

136 This debate also raises interesting questions of whether theories of distributive justice demand coverage for genetic enhancements, a distinction that will likely be as difficult to make as the line between prevention and treatment. See *supra* part I.B.; SEGALL, *supra* note 132, at 122; BROCK et al., *supra* note 131, at 309.

137 Gutmann, *supra* note 135, at 56–59.

preventive services in order to minimize health disparities and ensure a just distribution of health care.

A. Policy options that fail to adequately address concerns of equitable access

(1) Leave it to the marketplace

There are scarce resources in the US healthcare system, and therefore rationing is an unavoidable necessity. Our healthcare system focuses on treatment, in part because those individuals have the greatest need as the threat to their health is imminent, not prospective. Expansion of insurance coverage for any number of procedures increases costs to an already strained system, thus threatening to further increase health insurance premiums for individuals. Mandating insurance coverage for prevention in order to minimize health disparities will be fruitless if it prices out of the market the very individuals the policy is aimed at protecting. Therefore, allowing decisions about coverage for preventive services to occur at the insurance company level may be an appropriate option under the argument that scarce resources should be rationed toward treatment, not prevention.

This argument is problematic because policymakers have already made the determination that BRCA testing should be covered by insurance. The changes of the ACA will expand the utilization and insurance coverage of certain genetic tests as the USPSTF continues to review genetic conditions. While there may be an argument that resources should focus on treatment, there is a moral argument that once a genetic test is reimbursed, the interventions must also be equitably reimbursed. Additionally, the ACA increased focus on preventive care to address concerns about healthcare costs and rationing since greater access to preventive services can help reduce health costs in the future. Although increased insurance costs may arise from coverage due to mandated reimbursement, overall the health insurance market may see cost savings as chronic diseases are prevented and the current collective action problem of paying for prevention is minimized.

Targeted reimbursement policies are also necessary in light of the ACMG incidental findings recommendations. The fifty-six genes on the ACMG list were chosen by the committee under the rationale that they were associated with conditions for which there are medically actionable preventive measures. Access to these measures is a necessary pre-requisite to meet the estimated clinical utility of the genetic information returned by laboratories. Lack of reimbursement and other barriers to access thus pull the rug out from underneath the basis for returning incidental findings to patients undergoing next generation sequencing.

Past experience, especially in the context of BRCA testing, has showed that insurance companies will not consistently cover expenses for intervention services even when they are clinically utilized or recommended. This trend will likely recur for other genetic tests that are discovered if a systemic change does not occur. Leaving questions of insurance coverage for interventions to the marketplace are unlikely to ensure adequate and just coverage or slow expanding health disparities.

(2) Coverage through USPSTF

The ACA requires that health insurance companies reimburse individuals without cost sharing for any preventive procedure that the USPSTF has rated an A or a B. Therefore,

if the USPSTF reviews all subsequent interventions when the panel evaluates a genetic test, recommended interventions could be covered. This occurred in a limited manner when the USPSTF reviewed BRCA testing because they included genetic counseling in the recommendation. However, other interventions, such as mammograms, MRIs, and prophylactic surgeries were not given recommendations.

It will be difficult for the USPSTF to comprehensively review all interventions given the structure of the panel and the confusion between what is considered prevention and treatment. The limited authority of the USPSTF prevents review of evidence for tertiary prevention.[138] For example, in the context of hemochromatosis, if the task force interprets iron tests or phlebotomy as treatment for clinical conditions, it would not be able to make a graded recommendation. However, if these interventions were considered secondary preventions that are meant for patients with biological alterations that have not led to symptoms, review is permissible. The determination of whether interventions are treatment or preventive could drastically vary from genetic condition to genetic condition, leading to disparate coverage across genetic conditions.

The cost sharing mechanisms for prevention under the ACA also weigh against a policy solution through recommendations from the USPSTF. The ACA mandates free preventive services under the rationale that even small copays and coinsurance amounts can preclude individuals from receiving essential preventive screenings.[139] However, the types of preventive services recommended currently by the USPSTF are counseling for chronic diseases, screenings, and preventive medication.[140] All of these are a far cry from the in-depth and expensive interventions following genetic testing such as prophylactic surgery. Mandating complete coverage for such expensive procedures would likely increase premium costs and likely be at odds with Congress' vision of free preventive services.

Additionally, preventive interventions without cost sharing create a perverse and inequitable system. For example, individuals who are diagnosed with colon cancer may need to undergo a colectomy as part of treatment. A prophylactic colectomy can also be recommended for some genetic conditions, such as familial adenomatous polyposis (FAP). If the USPSTF reviewed FAP and recommended a colectomy for those with a positive genetic test, it would create an inequitable system in which those with a cancer diagnosis would be faced with high out of pocket costs for the same procedure that asymptomatic individuals with FAP would obtain for free. Insurance coverage for genetic conditions and their associated interventions is essential to avoid a genetic underclass;[141] however, providing no cost sharing for extensive procedures threatens to make an underclass of those whose disease has no genetic cause.

Cost sharing for such expensive interventions will be prohibitive for some individuals. An ideal solution would ensure that all individuals have equal access to services, but unfortunately the US healthcare system does not provide this equity. Until the USA provides affordable and accessible insurance for all, there will remain some segment of

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138 USPSTF, *supra* note 72, at § 1.3.

139 See eg Roy C. Baron et al., *Client-Directed Interventions to Increase Community Access to Breast, Cervical, and Colorectal Cancer Screening: A Systematic Review*, 35 Am. J. Preventive Med. S56, S61–63 (2008).

140 USPSTF, *USPSTF A-Z Topic Guide*, http://www.uspreventiveservicestaskforce.org/uspstopics.htm (accessed March 3, 2015).

141 See Evans, *supra* note 32, at 2671.

the population that cannot access preventive interventions. Health disparities cannot be completely eradicated and equal access to healthcare cannot be completely guaranteed, but this can be mitigated by a base level of insurance coverage for interventions. The USPSTF recommendation system is not the best vehicle to ensure reimbursement because these procedures walk the fine line between prevention and can be too expensive to advocate for the complete removal of cost sharing.

B. Policy options that address concerns of equitable access to preventive services

(1) Coverage mandate through federal legislation or regulation

Federal legislation mandating insurance coverage for adult onset genetic testing for asymptomatic individuals and applicable interventions would ensure reimbursement at appropriate cost sharing levels. Legislatures have utilized this tried and true approach in other preventive arenas, from mandated coverage for cancer screenings[142] to coverage for specialty medical foods and supplements recommended for individuals with inherited metabolic disorders tested for through state newborn screening programs, including phenylketonuria (PKU).[143] This approach would not prevent individuals from accessing, or clinicians and laboratories from offering, new genetic tests on the free market, but would ensure that once broad coverage for the predictive genetic test occurred, the interventions would be equitably reimbursed.

Federal action can come in the form of legislation or changes to the regulatory system. As mentioned, in the regulations for the ACA, HHS gave each state the authority to establish what the essential health benefits are for its area. These regulations could be amended to create a federal baseline of preventive services that must be covered, including genetic services and interventions, without delegating this determination to the states.

(2) Coverage mandate through state legislation

Federal legislation is an appropriate, but currently unlikely, vehicle to ensure consistent access to given existing congressional unwillingness to pass healthcare legislation or amend the ACA. If neither federal legislation nor regulation addresses this issue, a state-by-state solution is a possible, albeit less desirable approach. A state-by-state solution creates an ad hoc and disparate system of coverage across the country, as illustrated by reimbursement for medical foods for individual with PKU. Additionally, self-funded health insurers are exempt from state law and therefore any mandated coverage would not be required for these companies.[144] If necessary, advocates should push for a state-by-state solution as an intermediate strategy that builds support and consensus until window for national legislation or regulation opens.

States can mandate coverage for genetic testing and recommended interventions either through broad legislation or as an essential health benefit under the mechanisms established by the ACA. All health insurance plans sold in a state exchange must mirror the chosen state benchmark plan, which must provide benefits for ten essential health

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142 See *supra* part II.C.4.

143 Susan A. Berry et al., *Insurance Coverage of Medical Foods for Treatment of Inherited Metabolic Disorders*, 15 GENET. MED. 978 (2013).

144 ERISA, Pub. L. No. 93–406.

benefits, including prevention and wellness.[145] The existing state benchmark plans vary in coverage policies for genetic tests. For example, Maine's plan only covers genetic testing and counseling on diagnosed patients if it will provide information for the treatment plan. Hawaii and Idaho specifically include genetic testing under diagnostic services and Iowa explicitly excludes genetic testing for informational purposes.[146] Coverage for genetic tests and interventions is not clearly defined in the chosen plans.

Currently, federal regulations allow states to define additional benefits that fall within the essential health benefit categories and to add additional categories of essential health benefits.[147] State legislation could add coverage for genetic testing and interventions to the preventive and wellness services benefit category. This strategy fails to ensure coverage for those individuals in large group health plans, as only individual and small group health plans are required to cover essential health benefits[148]; however large group plans are the most likely to have broader coverage in general. If federal regulation were altered in this area, it would include these same limitations of coverage, but would be beneficial in the policy consistency it would provide across the country.

(3) Responsive policy

Regardless of whether states or the federal government passes legislation broadly or through essential health benefits, the policy must be nimble enough to keep pace with medical developments in this area. Various methods of policy development are available for the establishment and modification of the list of covered tests and interventions. In the early years of newborn screening, state models for developing the list of screened conditions, included traditional legislative action, regulatory authority, reliance on a panel or advisory body, or a combination of these strategies.[149] These options are similarly available in the context of medical interventions following a genetic test and offer varying degrees of flexibility and responsiveness to medical developments. State legislatures should avoid a delineated list of mandated procedures because, once passed, legislation remains fairly stagnate and difficult to alter or amend the list apace of genetic advancement, as is the case with Medicare's prevention exceptions. A desirable balance of clarity and responsiveness can be achieved through either a regulatory or panel-based system.

A similar type of advisory panel has been recommended at the federal level to "offer advice on what genetic and genomic findings are sufficiently well understood, significant, and actionable to qualify for return."[150] This type of advisory board is a natural choice to make additional recommendations for insurance coverage because it would

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145 See *supra* part III.B.

146 Centers for Medicare and Medicaid Services, *Additional Information on Proposed State Essential Health Benefits Benchmark Plans,* http://www.cms.gov/CCIIO/Resources/Data-Resources/ehb.html (accessed March 3, 2015).

147 45 C.F.R. § 155.170(a).

148 42 U.S.C. § 300gg-6(a).

149 Jeffrey J. Stoddard & Philip M. Farrell, *State-to-State Variations in Newborn Screenings Policies*, 151 Archives Pediatric & Adolescent Med. 561, 562 (1997).

150 Susan M. Wolf et al., *Managing Incidental Findings and Research Results in Genomic Research Involving Biobanks and Archived Data Sets*, 14 Genet. Med. 361 (2012); Richard R. Fabsitz et al., *Ethical and Practical Guidelines for Reporting Genetic Research Results to Study Participants Updated Guidelines from a National Heart, Lung, and Blood Institute Working Group*, 3 Circulation Cardiovascular Genet. 574 (2010).

already be examining the interventions in order to determine which findings are sufficiently actionable. Thus, they can easily take into account the necessary insurance reimbursement to determine the cost/benefit analysis of which results to return. This weighing of costs and benefits can include not only the benefits and harms of coverage for the individual, but also consideration of the overall cost-effectiveness of the interventions. Further discussion is needed to determine the types of considerations that such an advisory panel should take into account. For example, what is an appropriate evidence threshold for both the test and the interventions to include them on the list and how likely of a benefit must there be for to meet criteria for inclusion?

4. Voluntary adoption by state benchmark plans

Although the marketplace is inadequate to ensure comprehensive coverage for genetic testing and interventions,[151] a market-based solution may be appropriate or necessary in states where the legislature will not mandate coverage. Advocacy groups could lobby the state benchmark plans to cover preventive genetic services and interventions. In the past, private insurance companies have failed to cover these services in part due to enrollee turnover.[152] However, once the state benchmark plan covers the services, all other plans in the exchange must cover substantially equal benefits, therefore the costs concerns will be minimized since those entering the plan are more likely to have undergone genetic testing and interventions paid for by a previous plan.[153] In this way, both the costs and the benefits will reach a multitude of plans, as opposed to the system prior to the ACA where the first adapter paid the steep costs of interventions such as surgery, but was not guaranteed to see the savings. Thus, adoption by the state benchmark plan efficiently addresses the collective action dilemma of the free market system. State or federal legislation can establish a system of continued review of medical advances through expert panels or advisory boards and is thus a desirable solution, but in the absence of legislature action, ad hoc coverage by state benchmark plans can help move society toward equitable coverage.

VII. CONCLUSION

Genomic technologies and the ability to test asymptomatic individuals for adult-onset conditions have created a liminal state, between cultural and bureaucratic concepts of health and illness. These individuals are not experiencing symptoms, but due to genetic or genomic testing, know that they are at increased risk of a genetic condition or disease. Testing thus creates a kind of iatrogenic condition: a no man's land at odds the US healthcare system that is rooted in treatment, not prevention.

Knowledge of genetic information allows the individual to take steps to mitigate risk through medical interventions such as surgery, screening, or medication, but these interventions have variable coverage in the health insurance marketplace. Barriers to access for these interventions threaten to exacerbate health disparities and undermine the basic policy rationale for advocating preventive testing in an asymptomatic population. Therefore, state or federal legislatures should establish advisory boards or regulatory authorities whose role is to create a dynamic list of interventions for asymptomatic

[151] See *supra* part VI.A.1.

[152] See *supra* part II.C.2.

[153] 45 C.F.R. § 156.115(a) (1) (i–iii).

individuals that must be covered by insurance companies when medically appropriate for the individual. Additional tests would still be available on the free market, but this panel could ensure that once a policy was in place or in consideration to mandate access to predictive genetic testing or information, the necessary interventions would also be equitably accessible. Without mandated coverage of interventions, the promise of genomic testing as a tool for prevention is blunted and a disparate healthcare system is further entrenched in our society.

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