A problem not yet manifest: gaps in insurance coverage of medical interventions after genetic testing

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INTRODUCTION

In the past decade, the field of genomics has rapidly changed and expanded.\textsuperscript{1} With these advancements also come new applications of genomics and genetics to clinical medicine. The information gathered from genetic testing and genome sequencing can reveal a great deal about not only an individual’s current health, but his/her future health as well.\textsuperscript{2} This rapid expansion of scientific and medical capacity is accompanied by rapid changes for law and policy making thoughtful regulation essential.

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\textsuperscript{1} See generally, Asude Alpman Durmaz et al., Evolution of Genetic Techniques: Past, Present, and Beyond, 2105 BIOMED. RES. INT’L 1 (2015).
\textsuperscript{2} Genetic Testing, GENETIC HOME REFERENCE, Sept. 7, 2015, http://ghr.nlm.nih.gov/handbook/testing?show=all#interpretingresults (accessed Sept. 13, 2015).

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The human genome includes many variations, most of which have no known significance. However, some variants can be the cause of important medical conditions, and for a subset of these, there are useful healthcare interventions, which can be deployed if the genetic variation is recognized. Sometimes, these genetic variations are the specific target of a genetic test. Sometimes, the variations are found incidentally, in a genetic test performed for other purposes, such as pharmacogenomics or pre-conception screening. In 2013, the American College of Genetic Medicine (ACMG) released recommendations specifying which of these incidental findings should be given to clinicians. The ACMG limited its recommendation to 56 genetic variations, which can result in approximately 24 genetic conditions. Most of these genetic conditions are very rare, with many of the available treatments limited to continued monitoring and increased surveillance for changes in symptomology and/or disease progression.

Anya E.R. Prince discusses these issues in her article, ‘Prevention for those who can pay: insurance reimbursement of genetic-based preventive interventions in the liminal state between health and disease.’ Prince worries that while many health insurance companies offer coverage for genetic testing, fewer offer coverage for prophylactic measures and treatments for the conditions said testing might reveal. Lack of such coverage would undermine the policy goals that motivated insurance coverage mandates in the first place, and may perpetuate health disparities. We advance Prince’s analysis of this issue by exploring actual coverage in the private health insurance market, which covers 64 per cent of Americans. In particular, we analyse the coverage of two genetic conditions—BRCA and catecholaminergic polymorphic ventricular tachycardia (CPVT)—by reviewing policy documents of commercial health insurance companies, including Cigna, Aetna, Blue Cross Blue Shield, and United Health One. We find that, while genetic testing and consequent treatment is not universally covered, many companies do offer a broad scope of coverage in this area. Prince argues that the Affordable Care Act (ACA) and ACMG tout preventative measures without considering the impact the information can have on individuals who are unable to afford the preventative next steps.

IMPACT OF THE ACA

The ACA sought to increase the number of individuals in the U.S.A. with adequate health insurance by creating employer and individual mandates, creating new mechanisms for affordable coverage (health care exchanges), and expanding eligibility for insurance under existing mechanisms (primarily Medicaid). In addition to covering

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3 Robert C. Green et al., ACMG Recommendations for Reporting Incidental Findings in Clinical Exome and Genome Sequencing, 15 GENET. MED. 4 (2013).
4 Id. at 570.
5 Id. at 577.
6 Anya E.R. Prince, Prevention for Those Who Can Pay: Insurance Reimbursement of Genetic-based Preventive Interventions in the Liminal State between Health and Disease, 2 J. L. BIOSCI. 365–395 (2015).
7 Jessica C. Smith & Carla Medalia, Health Insurance Coverage in the United States: 2013, U.S. DEP’T. COM., Sept. 2014, https://www.census.gov/content/dam/Census/library/publications/2014/demo/p60–250.pdf (accessed Sept. 14, 2015).
more people, the ACA tried to improve the quality of coverage, imposing caps on cost sharing and mandatory benefit packages. Preventative services are a cornerstone of the ACA, including immunizations, HIV screening, and well-woman visits.8

Section 2713 of the ACA requires non-grandfathered group plans as well as insurance offered in the individual or group market to provide coverage for evidence-based services that received a rating of A or B from the United States Preventative Task Force (USPSTF).9 The ACA instructs that when a recommendation applies to a high-risk population, the recommended service must be covered for individuals within that population if deemed appropriate by a clinician.10 Section 2713 also states that additional coverage for services beyond those provided by the USPSTF is not prohibited.11 These provisions apply to those genetic tests and subsequent treatments that the USPSTF deems both beneficial and substantially backed by available evidence.12 Thus, under the ACA, coverage for certain genetic tests and treatments has increased, although not all of the genetic conditions identified by the ACMG have received the requisite rating of an A or B by the USPSTF to mandate coverage.

INSURANCE COVERAGE

There are multiple ways in which a genetic test or subsequent treatment may not be subject to a legal coverage mandate. The plan may be grandfathered, and thus not subject to ACA coverage mandate. Or, the tests may not be recommended by the USPSTF. On the other hand, there are market forces, which shape private health insurance coverage, as well as more general legal principles, such as the contractual requirement for an insurer to cover medically necessary healthcare. Accordingly, we sought to understand whether the coverage gaps hypothesized by Prince actually exist in the real world of health insurance. To test this question, we selected two genetic conditions, which are medically actionable: BRCA and CPVT.13 The USPSTF has recommended BRCA testing for women with family histories, but it has not made a genetic screening recommendation with regard to CPVT.14

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8 Preventive Services Covered under the Affordable Care Act, HHS. gov, U.S. DEP’T HEALTH HUM. SERV, Sept. 27, 2012 [hereinafter Preventive Services], http://www.hhs.gov/healthcare/facts/factsheets/2010/07/preventive-services-list.html (accessed Sept. 13, 2015).

9 Affordable Care Act Implementation FAQs – Set 12, CMS. gov, CTR. FOR MEDICARE AND MEDICAID SERV., https://www.cms.gov/CCHIO/Resources/Fact-Sheets-and-FAQs/aca_implementation_faqs12.html (accessed Sept. 13, 2015).

10 Id.

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

12 See generally U.S. Preventive Services Task Force, Sept. 2014, http://www.uspreventiveservicestaskforce.org/ (accessed Sept. 13, 2015).

13 See Green, supra note 4, at 570 (listing both of these conditions).

14 See BRCA-Related Cancer: Risk Assessment, Genetic Counseling, and Genetic Testing, U.S. Preventative Services Task Force, Dec. 2013, http://www.uspreventiveservicestaskforce.org/Page/Document/Update-SummaryFinal/brica-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing (stating the recommendation); The Center for Consumer Information & Insurance Oversight, Centers for Medicare and Medicaid Services: Affordable Care Act Implementation FAQs - Set 12, CMS.GOV, available at https://www.cms.gov/CCHIO/Resources/Fact-Sheets-and-FAQs/aca_implementation_faqs12.html (interpreting the USPSTF recommendation to apply to the testing itself).
BRCA

Made famous in recent years by the actress Angelina Jolie, BRCA1 and BRCA2 are protein-producing genes that repair damaged DNA and help ensure the stability of a cell’s genetic material, which, in turn, suppresses tumour growth. Variations in these genes undermine the normal and necessary process of DNA repair, and hence, can cause genetic alterations to cells and lead to cancer. Some of these variants to the genes can be inherited and can increase the risk of both breast and ovarian cancers in women. According to the National Cancer Institute, BRCA variants are responsible for approximately 20–25 per cent of hereditary breast cancers, 5–10 per cent of all breast cancers, and about 15 per cent of ovarian cancers. Additionally, lifetime risk for developing breast cancer is as high as 90 per cent if a woman inherits a harmful BRCA1 or BRCA2 variation.

Prince writes that although variations to the BRCA1 and BRCA2 genes are one of the more widely studied genetic conditions, insurance companies provide varying levels of coverage for both genetic testing and recommended interventions. These interventions include breast cancer screening beginning at an earlier age since cancers linked to BRCA mutations often develop in women in their 20s and 30s as well as prophylactic mastectomy and/or oophorectomy. Prince goes on to state that, ‘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.’

To explore this potential problem, we examined the actual medical coverage policies of major private insurance companies. Cigna, for instance, offers BRCA1 and BRCA2 genetic testing for individuals with a known familial mutation, personal history of breast or other cancers, or a family history of breast, ovarian, or pancreatic cancer. Cigna does ‘not’, however, cover BRCA1 or BRCA2 genetic testing in the general population, in individuals with no personal history of breast or ovarian cancer, and individuals under the age of 18. Similarly, United Healthcare’s policy states that BRCA1 and BRCA2 testing is medically necessary (ie, covered under standard contractual coverage provisions) for customers including, but not limited to women with a personal history of breast cancer who were diagnosed at 45 or younger with or without a family history of cancer; women with a personal history of breast cancer diagnosed at 50 or younger with one close blood relative with breast cancer or unknown family history; women without

15 BRCA1 and BRCA 2: Cancer Risk and Genetic Testing. Nat’l. Cancer Inst., Apr. 1, 2015 [hereinafter Cancer Risk] http://www.cancer.gov/about-cancer/causes-prevention/genetics/brca-fact-sheet#q1 (accessed Sept. 13, 2015).
16 Id.
17 Id.
18 Id.
19 Id.
20 Prince, supra note 8, at 12.
21 Cancer Risk, supra note 16.
22 Prince, supra note 8, at 13.
23 Genetic Testing for Susceptibility to Breast and Ovarian Cancer (e.g. BRCA 1 & BRCA 2), CIGNA MEDICAL COVERAGE POLICY 1–5, Aug. 15, 2015 [hereinafter Susceptibility to Breast and Ovarian Cancer], https://cignaforhcp.cigna.com/public/content/pdf/coveragePolicies/medical/mm.0001_coverageposition -criteria_genetic_testing_for_breast_and_ovarian_cancer.pdf (accessed Sept. 13, 2015).
24 Id. at 7.
a personal history of breast or ovarian cancer with a known BRCA1 or BRCA2 mutation in the family; and, women with a personal history of ovarian cancer.  Remarkably, the coverage provided by these insurers is in some ways broader than that recommended by USPSTF.

Prince argues that, '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.' Our review of policy documents found that, not only do many of these companies offer coverage for screening, they also cover prophylactic mastectomies and oophorectomies for their clients. Aetna’s policy, for example, states that it considers prophylactic mastectomy medically necessary to reduce breast cancer in high-risk women including those with BRCA1 or BRCA2 mutations that have been confirmed by molecular susceptibility testing. The same policy covers prophylactic bilateral oophorectomy or salpingo-oophorectomy for women with BRCA1 or BRCA2 mutations. Other major insurance companies also cover prophylactic mastectomies and/or oophorectomies when it is considered medically necessary for customers who have a high risk of breast cancer. In nearly all of the policies looked at, definitions of high risk included a BRCA1 or BRCA2 mutation.

These findings should not be surprising. Such policies represent sound financial decisions for insurance companies, since ‘the strategy of prophylactic mastectomy...
and oophorectomy has the lowest overall cost for insurers—this is the least expensive method for managing the insured’s risk. This dynamic is unlike the economics behind much preventative care, which—contrary to much political rhetoric—is not cost effective, from a financial point of view, given the inability to target only that subset of individuals at the highest risk of developing a disease long into the future.

**CPVT**

CPVT causes arrhythmia, or abnormal heart rhythm, which can cause an overly fast and irregular heartbeat. This abnormal heartbeat can lead to light-headedness, dizziness, fainting, and even death. According to estimates, the condition affects approximately 1 in 10,000 people and can result when variants in one of two genes—RYR2 and CASQ2—occur. The variants, which cause disruption to the handling of calcium within the heart muscle cells (myocytes), are responsible for approximately 50 per cent and 1–2 per cent of all cases, respectively.

Aetna offers coverage of genetic testing for CPTV in children and young adults who have a first degree relative with a diagnosis of CPTV as well as for individuals who have structurally sound hearts but exhibit exercise and/or emotion induced PVT or ventricular fibrillation. United Healthcare’s most recently released guidelines also state that genetic tests for CPTV are medically necessary when certain criteria are met.

Following a positive genetic test, insurers do cover relevant interventions. For example, Cigna provides coverage for transvenous implantable cardioverter defibrillator (ICD) for genetic conditions including CPTV when medically necessary. ICDs are devices that are surgically implanted in order to continuously monitor the heart rate of the user and deliver a shock whenever an arrhythmia is detected in order to stop the arrhythmia. The devices, which reduce the risk of cardiac arrest, are useful in individuals who experience frequent arrhythmic events and are a beneficial therapy for people

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32 See generally, Victor R. Grann et al., Comparative Effectiveness of Screening and Prevention Strategies Among BRCA1/2-affected Mutation Carriers, 125 BREST CANCER RES. TREAT. 844 (2011).

33 See generally, Joshua T. Cohen et al., Does Preventive Care Save Money? Health Economics and the Presidential Candidates, 358 NEW. ENG. J. MED. 661 (2008).

34 Catecholaminergic Polymorphic Ventricular Tachycardia, Genetics Home Reference, Dec. 2009, http://ghr.nlm.nih.gov/condition/catecholaminergic-polymorphic-ventricular-tachycardia (accessed Sept. 13, 2015).

35 Id.

36 Id.

37 Id.

38 Genetic Testing, AETNA, May 6, 2015, http://www.aetna.com/cpb/medical/data/100_199/0140.html (accessed Sept. 13, 2015).

39 Policy Update Bulletin, UNITED HEALTHCARE 4, June 2015, https://www.unitedhealthcareonline.com/ccmcontent/ProviderII/UHC/en-US/Assets/ProviderStaticFiles/ProviderStaticFilesPdf/Tools%20and%20Resources/Policies%20and%20Protocols/Medical%20Policies/SV%20MPUBs/MMG%20Update%20Bulletin%20June%202015.pdf (accessed Sept. 13, 2015).

40 Wearable Cardioverter Defibrillator and Automatic External Defibrillator, CIGNA 3, Feb. 15, 2015, https://cignaforshcp.cigna.com/public/content/pdf/coveragePolicies/medical/mn_0431_coverageposition_criteria.wearable_cardioverter_defibrillator_and_aed.pdf (accessed Sept. 13, 2015).

41 Id. at 6.
with CPTV. Like Cigna, Aetna also covers ICDs in individuals with familial or inherited CPTV who have ‘syncope and/or documented sustained VT while receiving beta-blockers’. 

CAVEATS AND CONCLUSIONS

We explored merely two medically actionable genetic conditions (BRCA and CPTV), using a convenience sample of policy documents, available on the internet. We cannot say whether the insurance coverage for these two genetic variations is representative of the more than two dozen other medically actionable variations recommended by ACMG.

We also searched for explicit screening and treatment for these two conditions in policy guidance documents. Insurance policies are complex, lengthy documents with many caveats and exemptions. The actual terms of the contract, including coverage of ‘medically necessary’ care, and certain exclusions, will govern. In particular, nearly every insurer precludes coverage for treatment that is deemed ‘experimental’, although it is not always clear what constitutes experimental treatment or what degree of proof is needed to warrant coverage.

These prohibitions could block access to coverage for a number of these conditions, where evidence supporting the efficacy and safety of treatment is lacking.

Nonetheless, when looking at the companies’ own policy guidance documents, we were unable to document the problems that Prince hypothesized. We found that major insurers do cover not only a range of genetic tests but also treatment and surveillance for those conditions that can benefit from increased monitoring and proactive treatment.

Thus, our preliminary review of the evidence for these particular insurers, in this particular market, for these particular genetic variations, suggest that Prince’s worries about gaps in coverage may be more theoretical than real. Nonetheless, as the science and medicine evolves, these ethical and legal issues should continue to be monitored and resolved in order to ensure that barriers to needed healthcare do not arise.

42 Id.
43 Cardioverter-Defibrillators, AETNA, Aug. 28, 2015, http://www.aetna.com/cpb/medical/data/500_599/0585.html (accessed Sept. 13, 2015).
44 See eg Susceptibility to Breast and Ovarian Cancer, supra note 24; BRCA Testing, supra note 29.