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***Direct-to-Consumer Genetic Testing: Evolving Challenges***

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I. INTRODUCTION

In today's society, consumers have seemingly developed the expectation of countless informational resources at their fingertips. The evolution of the internet and the availability of a staggering amount of information have turned the average consumer into an active researcher, deciphering the answers to life's questions one web page at a time. A consequence of this conduct has been an inability to guarantee the reliability of information obtainable through the internet. The availability of often inaccurate information has found its way into our healthcare system as many turn to the internet for medical advice or assistance with self-diagnosis. This new trend in consumer orientation with respect to healthcare has recently become "particularly evident in the proliferation of direct-to-consumer (DTC) advertising for health-related products."<sup>1</sup> A popular example

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<sup>1</sup> Sarah E. Gollust et al., *Limitations of Direct-to-Consumer Advertising for Clinical Genetic Testing*, 288 J. AM. MED. ASS'N. 1762, 1762 (2002).

of this advertising is present in pharmaceutical advertisements in print and television commercials.<sup>2</sup>

In addition to pharmaceuticals, DTC advertised health-related products have included testing for traditional genetic conditions.<sup>3</sup> Such traditional conditions have included hereditary colon cancer syndromes or factor V Leiden deficiencies, among others.<sup>4</sup> Recently, there has been a substantial shift in the sophistication and availability of DTC genetic tests that has prompted the interest of state and federal regulatory bodies as well as genetic research organizations.<sup>5</sup> The interest in DTC genetic testing has centered on both a potential need for regulation and the potential issues that may prevent efficient regulation.

This article will examine the potential impact that DTC genetic testing may have on the quality of care that consumers receive, and focus on inaccurate expectations that can result from a lack of genetic counseling accompanied with consumer concerns regarding their genetic privacy. Part II of this article will establish the current uses and prevalence of DTC genetic testing. Part III will assess some of the risks and benefits that coincide with the implementation of DTC genetic testing. Again, a particular focus will be paid to concerns with respect to ill-informed consumers and the potential for inaccurate or unrealistic expectations concerning genetic testing benefits and its effect on consumer's quality of care. Part IV will explore issues with the government's limited role and

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<sup>2</sup> *Id.*

<sup>3</sup> Greg Feero, *Keep an Eye on Direct-to-Consumer Testing: Genomic Medicine*, 38 FAM. PRAC. NEWS 46, 46 (2008).

<sup>4</sup> *Id.*

<sup>5</sup> *Id.*

arguably ineffective regulation of DTC genetic testing and advertising. Finally, part V will discuss the implications of implementing stricter genetic counseling requirements with an emphasis on the role of primary care physicians in promoting consumer education and preventing consumer misconception.

## II. USES AND PREVALENCE OF DTC GENETIC TESTING

Our understanding of the human genome has exponentially increased following the completion of the Human Genome Project in 2003.<sup>6</sup> In fact, the Human Genome Project has resulted in the discovery of more than 1,800 disease genes and the potential identification of additional disease causing genes.<sup>7</sup> Though the exact number has been changing, there are currently more than 1,000 genetic tests that correlate to human diseases or conditions on the market today.<sup>8</sup> These clinical genetic tests include: carrier gene testing, prenatal testing, newborn screening, pharmacogenomic testing, diagnostic testing, and predictive testing.<sup>9</sup> Additionally, DTC tests have been created for multifactorial behavioral disorders<sup>10</sup> as well as nutrigenomic testing, which involve the testing of genetic variants and combining those results with a consumer's diet and lifestyle.<sup>11</sup>

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<sup>6</sup> Cynthia Marietta & Amy L. McGuire, *Direct-to-Consumer Genetic Testing: Is It the Practice of Medicine?*, 37 J. L. MED. ETHICS 369, 369 (2009).

<sup>7</sup> *Id.*

<sup>8</sup> *Id.*

<sup>9</sup> Gollust et al., *supra* note 1.

<sup>10</sup> See generally Alex Wilde et al., *Public Interest in Predictive Genetic Testing, Including Direct-to-Consumer Testing, for Susceptibility to Major Depression: Preliminary Findings*, 18 EUR. J. HUM. GENETICS 47, 48 (2010).

<sup>11</sup> Katrina A.B. Goddard et al., *Public Awareness and Use of Direct-to-Consumer Genetic Tests: Results from 3 State Population-Based Surveys, 2006*, 99 AM. J. PUB. HEALTH 442, 443 (2009).

Further, genetic testing can be used “at any stage in the developmental lifecycle.”<sup>12</sup>

Genetic tests are ordered for a variety of reasons including: “(1) to identify carriers of genetic disease; (2) to test embryos, fetuses, and newborns for disease-causing genetic abnormalities; (3) to establish clinical diagnoses or prognoses and inform clinical care; (4) to determine whether there is increased risk of developing a disease in the future; or (5) to predict response to a medication.”<sup>13</sup> Basically, DTC genetic testing “provides consumers with access to their genetic information without involving a doctor in the process.”<sup>14</sup> Genetic tests analyze DNA, RNA, chromosomes, or gene products contained in one of several different patient submitted samples.<sup>15</sup> Currently, approximately thirty-five DTC genetic companies reach consumers via the internet.<sup>16</sup>

### III. RISKS AND BENEFITS OF DTC GENETIC TESTING

There have been several disputes as to whether the alleged benefits of DTC genetic testing outweigh potential risks that it may impose. Specifically, the American College of Medicine Genetics issued a policy statement in 2004 opposing DTC genetic testing with the stance that genetic testing should only be provided through “appropriately qualified” healthcare professionals.<sup>17</sup> Further, the American College of Medicine Genetics stated that those healthcare

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<sup>12</sup> Gail H. Javitt et al., *Direct-to-Consumer Genetic Tests, Government Oversight, and the First Amendment: What the Government Can (and Can't) Do to Protect the Public's Health*, 57 OKLA. L. REV. 251, 257 (2004).

<sup>13</sup> *Id.*

<sup>14</sup> Marietta & McGuire, *supra* note 6.

<sup>15</sup> Javitt et al., *supra* note 12, at 257.

<sup>16</sup> Marietta & McGuire, *supra* note 6.

<sup>17</sup> *Id.* at 371.

professionals would be responsible for pre-test and post-test counseling involving the medical significance of the test results or follow-up.<sup>18</sup> The opposition to DTC genetic testing focuses on the potential vulnerability of “at home” genetic test consumers and the negative impact on the quality of care that those consumers may receive.<sup>19</sup> DTC genetic testing allegedly: “(1) fails to adequately explain complex genetic information; (2) is misleading in its failure to disclose the risks and limitations of testing; (3) allows tests without established clinical validity or utility to be promoted; and (4) does not include the counseling needed to put test results in proper context.”<sup>20</sup> Finally, DTC genetic testing also fosters consumer concerns with respect to privacy risks with potential discrimination that could result from determined genetic susceptibility to disease.<sup>21</sup>

Due to the complexity of genetic testing, ambiguity of test results, and challenges in interpretation, DTC genetic testing has inherent limitations.<sup>22</sup> For example, the presence of a specific gene sequence that has been associated with a particular disease would yield a positive test for that disease.<sup>23</sup> This positive test is limited because it only represents some probability of developing the identified disease and is far from deterministic.<sup>24</sup> In addition, if a variance is discovered on a genetic test and has not been associated with a disorder in other people, it can be

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<sup>18</sup> *Id.*

<sup>19</sup> *Id.*

<sup>20</sup> Javitt et al., *supra* note 12, at 253.

<sup>21</sup> Marietta & McGuire, *supra* note 6, at 370.

<sup>22</sup> *Id.*

<sup>23</sup> Javitt et al., *supra* note 12, at 260.

<sup>24</sup> *Id.*

very difficult to determine “whether it is a harmless polymorphism or a disease-causing mutation.”<sup>25</sup>

Further, the lack of genetic counseling in DTC genetic testing can have far reaching effects. With potentially inappropriate expectations derived from DTC genetic testing, primary care physicians could be required to expend precious time and resources to meet patients’ expectations, while simultaneously reducing time spent on other types of patient care and negatively impacting the quality of care the patient may receive.<sup>26</sup> Further, it may be very difficult to completely “modify consumers’ inaccurate expectations” potentially compounding patient care concerns.<sup>27</sup>

It is also very possible that physicians may not have the necessary skills to analyze the specifics of genetic inheritance, to accurately calculate genetic risks, or communicate genetic information that the patient has obtained in a nondirective way.<sup>28</sup> Further complicating what may be a primary care physician’s “suboptimal knowledge” of genetics, advertisements for genetic services in medical journals, mailers, and DTC genetic test kits may be a physician’s primary source of information about many genetic tests.<sup>29</sup> Informational brochures alone may not contain enough information to properly prepare physicians for patient questions.<sup>30</sup> The inadequacy of these possibilities indicate that primary care

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<sup>25</sup> Marietta & McGuire, *supra* note 6, at 370.

<sup>26</sup> Wylie Burke & Amy McGuire, *An Unwelcome Side Effect of Direct-to-Consumer Personal Genome Testing*, 300 J. AM. MED. ASS’N 2669, 2669 (2008).

<sup>27</sup> Gollust et al., *supra* note 1, at 1765.

<sup>28</sup> *Id.*

<sup>29</sup> *Id.*

<sup>30</sup> *Id.*

physicians may not be prepared for the potential flood of patients that have already received genetic testing or may seek it.<sup>31</sup> A physician's professional obligation to educate patients concerning DTC genetic test results, which may have a questionable value, could detract from more pertinent health related issues and negatively impact the patient's quality of care.<sup>32</sup>

Another realistic concern pertains to the security of DTC genetic test results.<sup>33</sup> Consumer concern relating to fears of privacy risks include the potential discrimination that could result from determined genetic susceptibility to disease.<sup>34</sup> Concerns relating to the security of results carry their own weight, but these concerns can be compounded with the ever-changing ownership of battling corporations responsible for genetic data they have collected.<sup>35</sup> Significant differences in company privacy policies that exist in DTC companies, coupled with issues that can arise regarding ownership of data collected following a company buyout or bankruptcy, present a complex problem concerning security.<sup>36</sup> These security concerns may impact consumer choices to obtain genetic testing while a breach of consumer security would negatively impact the effected consumer's quality of care.

After considering the potential risks, it is important to note a few benefits that supporters of DTC genetic testing have advanced. Proponents of DTC genetic testing believe that its implementation will result in an increase in

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<sup>31</sup> *Id.*

<sup>32</sup> Burke & McGuire, *supra* note 27.

<sup>33</sup> Wilde et al., *supra* note 10, at 49.

<sup>34</sup> Marietta & McGuire *supra* note 6, at 370.

<sup>35</sup> *Id.*

<sup>36</sup> *Id.*

personal responsibility among consumers for their health by making risk-assessment available to them.<sup>37</sup> Further, the absence of a licensed physician is argued to increase the utilization of DTC genetic testing.<sup>38</sup> It has even been suggested that DTC genetic testing advertisements may have an educational effect by promoting awareness of disease in community groups.<sup>39</sup> Despite recognized benefits of DTC genetic testing, the prevalence of consumer quality of care concerns shared by multiple organizations and agencies substantiate the need for oversight.<sup>40</sup>

#### IV. PROBLEMS WITH GOVERNMENT REGULATION OF DTC TESTING

With the variety of DTC genetic tests available, and information tested for, it can be difficult to determine whether test results are being provided for recreational purposes or whether the information is intended for medical diagnosis.<sup>41</sup> This difficulty in differentiation raises concerns about what services are actually being rendered. Some genetic tests strictly determine genetic information such as ancestry, while others test for single hereditary disorders, such as Huntington disease.<sup>42</sup> When genetic tests are conducted on a non-symptomatic patient, the results are solely predictive, not diagnostic.<sup>43</sup> But the difficult question becomes: when does genetic risk assessment information for

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<sup>37</sup> *Id.*

<sup>38</sup> *Id.*

<sup>39</sup> Gollust et al., *supra* note 1, at 1763.

<sup>40</sup> Marietta & McGuire, *supra* note 6, at 371.

<sup>41</sup> *Id.* at 369.

<sup>42</sup> *Id.* at 369-70.

<sup>43</sup> Javitt et al., *supra* note 12, at 260.

disorders constitute the practice of medicine?<sup>44</sup> If determined, when and how should this medical practice be regulated? How do we treat companies that provide DTC genetic testing and claim that results are for “educational purposes?”<sup>45</sup>

The United States government has the legal authority to regulate or limit consumer access to products and services that impact consumer health.<sup>46</sup> Regulatory supervision falls within the reach of the Food and Drug Administration (FDA), the Center for Medicare & Medicaid Services (CMS), and the Federal Trade Commission (FTC).<sup>47</sup> The FDA and CMS regulatory powers relate to the sale of products and services, including “development, testing, production, and distribution.”<sup>48</sup> The FTC’s oversight pertains to the advertising of commercial products and services.<sup>49</sup> Due to the ambiguity concerning the legal status of different types of genetic testing and how they are utilized, along with an arguably “underzealous exercise over available authority by CMS,” insufficient oversight has resulted.<sup>50</sup> In fact, genetic testing regulation and these regulating bodies have been described as ambiguous and insufficient.<sup>51</sup> The “cracks” between these regulations and their promulgation have seemingly allowed many of these DTC tests to slip past appropriate regulation.<sup>52</sup>

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<sup>44</sup> Marietta & McGuire, *supra* note 6, at 370.

<sup>45</sup> *Id.*

<sup>46</sup> Javitt et al., *supra* note 12, at 253.

<sup>47</sup> *Id.* at 268.

<sup>48</sup> *Id.*

<sup>49</sup> *Id.*

<sup>50</sup> *Id.*

<sup>51</sup> *Id.* at 253-54.

<sup>52</sup> *Id.* at 254.

A specific example of governmental issues with the regulation of direct-to-consumer marketing pertains to the doctrine of commercial speech.<sup>53</sup> Under the First Amendment to the U.S. Constitution, both state and federal governments are constrained from suppressing speech.<sup>54</sup> This general prohibition has been applied to commercial business and advertising resulting in the commercial speech doctrine.<sup>55</sup> One of the more recent and pertinent applications of the commercial speech doctrine by the Supreme Court was in the 2002 case of *Thompson v. Western States Medical Center*.<sup>56</sup> In *Thompson*, the Supreme Court determined the governmental interest in prohibiting pharmacists from advertising compounded drugs to be insufficient where the actual impact of the advertising was minimal and struck down the FDA Modernization Act of 1997.<sup>57</sup>

With the strict view taken by the Supreme Court in *Thompson*, it is likely that any restriction that the government or governmental agencies attempt to place on the advertising of DTC genetic testing would be subject to the assessment of actual impact of the advertising on the consumer and the governmental interest in regulating it.<sup>58</sup> Further, a court may be apprehensive to accept arguments that advertising restrictions are necessary due to consumer ignorance or difficulty in the consumer's ability to make decisions.<sup>59</sup> This essentially divests the

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<sup>53</sup> See generally *Thompson v. W. States Med. Ctr.*, 535 U.S. 357 (2002).

<sup>54</sup> Javitt et al., *supra* note 12, at 287.

<sup>55</sup> *Id.* at 289.

<sup>56</sup> See generally *Thompson*, 535 U.S. at 357-90.

<sup>57</sup> *Id.*

<sup>58</sup> Javitt et al., *supra* note 12, at 300.

<sup>59</sup> *Id.* at 300-01.

government's ability to regulate the advertisement and marketing of DTC genetic testing and further complicates effective regulation.

#### V. PRIMARY CARE PHYSICIAN'S ROLE IN POTENTIAL SOLUTIONS TO PERCEIVED RISKS OF DTC GENETIC TESTING

The limitations on the role the government can play in the effective regulation of DTC genetic testing encourages us to look to other avenues to protect consumers. This search for alternatives appears to lead to primary care physicians who may be best equipped to counsel patients with DTC genetic testing results.<sup>60</sup> Physicians will continue to play a role in both the education of consumers as well as ensuring a sufficient quality of care when interacting with those consumers after they have obtained DTC genetic tests.<sup>61</sup> Because “patient...decisions [are often based] on unconscious associations and assumptions rather than conscious deliberation” physicians must focus on patient counseling with respect to their choice to obtain DTC genetic testing.<sup>62</sup> It is important for physicians to utilize techniques during patient counseling such as “elicit[ing] underlying motivations and correct[ing] false assumptions.”<sup>63</sup> This can be done by focusing on the fact that DTC personal genetic tests currently offered are not supported with any data pertaining to the outcomes of testing.<sup>64</sup> It is possible that this approach, through education of patients who have went the route of DTC genetic testing, will positively impact the quality of care received while encouraging those patients to perpetuate a more accurate understanding of

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<sup>60</sup> Burke & McGuire, *supra* note 26, at 2669.

<sup>61</sup> *Id.*

<sup>62</sup> *Id.* at 2671.

<sup>63</sup> *Id.*

<sup>64</sup> *Id.*

risks, benefits, and limitations of DTC genetic testing to family, friends, and consumers on a broad scale.

## VI. CONCLUSION

It is clear that many obstacles to appropriate regulation of DTC genetic testing exist. Equally clear is the growing availability and versatility of genetic testing. With little dispute as to the benefits of genetic testing and the insight that it can provide under optimal circumstances, there is little doubt that its implementation will continue and likely increase. With more research, we can better understand the risks and benefits to consumers while ensuring a sufficient quality of care and effectively controlling the application of DTC genetic testing with insightful physicians and more effective legislative regulation.