



Beyond clinical utility: The multiple values of DTC genetics



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ABSTRACT

One point of consensus in the otherwise very controversial discussion about the benefits and dangers of DTC genetics in the health domain is the lack of substantial clinical utility. At the same time, both the empirical and conceptual literature indicate that health-related DTC tests can have value and utility outside of the clinic. We argue that a broader and multi-faceted conceptualization of utility and value would enrich the ethical and social discussion of DTC testing in several ways: First, looking at ways in which DTC testing can have personal and social value for users – in the form of entertainment, learning, or a way to relate to others – can help to explain why people still take DTC tests, and will, further down the line, foster a more nuanced understanding of secondary and tertiary uses of DTC test results (which could very well unearth new ethical and regulatory challenges). Second, considering the economic value and broader utility of DTC testing foregrounds wider social and political aspects than have been dominant in the ethical and regulatory debates surrounding DTC genetics so far. These wider political aspects include the profound power asymmetries that characterize the collection and use of personal genetic data in many contexts.

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1. Introduction

Internet-based companies offering genetic testing services directly to consumers have been surrounded by controversy from the start. While some authors have celebrated the arrival of genome-wide tests on the online market as marking a revolution in patient empowerment, others have raised concerns about consumers receiving genetic risk information without medical advice. The main concerns have focused on the negative psychological effects that genetic or genomic risk-susceptibility data with low predictive value could have on test-takers (e.g. by causing stress or anxiety), the potential adverse impact of these tests on the broader healthcare services in terms of unnecessary requests for screening and diagnostic services, and the possible privacy violations of sensitive and personal data (for an overview, see [Caulfield and McGuire, 2012](#)). Risks are seen as particularly high for people without adequate genetic literacy ([Offit, 2008](#); [McGuire and Burke, 2008](#); [Leighton et al., 2012](#)).

Moreover, some social scientists and ethicists consider direct-to-consumer (DTC) genetics as the epitome of a particularly individualist or consumerist approach to healthcare (e.g., [Hunter et al., 2008](#); [McGuire and Burke, 2008](#); [Harvey, 2010](#)). Donna Dickenson recently

referred to DTC genetic tests as one of the most problematic instantiations of personalized medicine – a new model of healthcare directly linked to the private sector and dictated by narcissism and the ideal of personal choice ([Dickenson, 2013](#)).

In the context of health, the DTC genetics market has not, as some enthusiasts had predicted, become a large and lucrative industry. The DTC genetics market continues to be dominated by genetic ancestry testing; health-related testing forms a very small niche within this market ([Wright and Gregory-Jones, 2010](#); [Petroni, 2014](#)). But contrary to some predictions, health-related DTC testing has not disappeared either. One of the pioneers of human genome-wide tests, the California-based company 23andMe, celebrated its one-millionth customer last year. "Just fifty years ago", an e-mail sent out to customers in June 2015 read, "doctors were reluctant to tell their patients if they had cancer. The world is different today".¹ (Note, however, that the figure of one million includes customers who bought the test purely out of interest in their genetic ancestry; it also includes some number of people—perhaps many thousands—who received 23andMe's service for free). Moreover, new DTC testing services are emerging, such as personal microbiome analysis, offering users a genetic analysis of, for example, their gut bacteria. Providers of such services include both for-profit ([uBiome.com](#)) and non-profit (American Gut Project, British Gut Projects) organizations,

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with the latter pursuing the aim of establishing an open-access database of microbial gut data.²

In recent years, a number of empirical studies have investigated the motivations, attitudes, and experiences of the general public and actual users of DTC tests. Findings from these studies paint a rather complex and partly contradictory picture (for a recent systematic overview of this literature, see Covolo et al., 2015). The main reasons for favorable attitudes towards these tests both among actual users and members of the public who had not taken a test have been their presumed medical importance and the potential for this information to prompt users to adopt a healthier lifestyle (McBride et al., 2009; Bloss et al., 2010; Cherkas et al., 2010; McGuire et al., 2009; Su et al., 2011; Savard et al., 2014). At the same time, both qualitative and quantitative studies of actual users of DTC genetics show no evidence for changes in anxiety levels, psychological health, diet, exercise or use of screening tests among DTC genetics users (Bloss et al., 2011, 2013; McGowan et al., 2010). Despite the absence of evidence for health-related behavior change, or psychological changes, however, most surveyed customers of DTC tests have been satisfied with the test experience (Bloss et al., 2010, 2011, 2013; McGowan et al., 2010).³ Why is this the case? If we accept that DTC genetic tests have little or no clinical utility – understood, in the narrow sense of the word, as the ability of a test to prevent or ameliorate adverse health outcomes such as mortality, morbidity, or disability through the adoption of efficacious treatments conditioned on test results (Grosse and Khoury, 2006; see also Khoury, 2003; Foster et al., 2009)⁴ – why do health-related DTC tests still exist? What utility and value do these tests have for test-takers, and for other actors?

2. DTC testing: Utility beyond the clinic

Let us first take a look specifically at what empirical studies say about why people take DTC tests. McGowan and colleagues interviewed early adopters of genome-wide DTC testing; they found that the two most common reasons for taking the test were to obtain health-related information and to learn about genetic risk factors (McGowan et al., 2010, 269). Another important motivation – not surprising in the group that this study focused on – was the desire to be on the vanguard of adopting new technologies. Other studies that analyzed the complexity and wide variety of practices around personalized genetic information showed

that what users “get out of” DTC genetic testing has little, of anything, to do with clinical decision making. Respondents in empirical studies referred to the role of curiosity and fascination with technological innovations and genetics; interest in participating in biomedical research (Su et al., 2011; Vayena et al., 2014); pride or professional interest in being on the vanguard as early adopters of a new technology (McGowan et al., 2010); interest in experimenting with biosocial relationships or a more proactive patient-role (Ducournau and Beaudevin, 2011; Ducournau et al., 2011); or the “fun factor”: a broad concept which encompasses the satisfaction of either taking part in promising and potentially useful research, or being part of a “cool” innovation (Vayena et al., 2012).

In all of these surveys it is difficult to distinguish between motivations to take a DTC test and the utility/value that DTC testing has for users. This is because when people are asked *after* having taken the test on why they took the test in the first place, there is no way of ascertaining on how they themselves separate motivations from utility; in light of the narrative structure of autobiographical memories it seems likely that in recalling motivations/expectations and ex-post utility of PGT, these notions of testing shade into one another.⁵

Both in terms of reported motivations to undergo testing and reported uses and thoughts about test results, another distinction that cannot easily be upheld is that between the personal and social domains. In a study carried out with volunteers in the British Twins cohort, for example, roughly 80% of those who said they were interested in taking a DTC test (5%–50% depending on the price of the service) said they wanted to do so to be able to convey risk information to their children (Cherkas et al., 2010). It is impossible, here, to describe such a (hypothetical) use of DTC test results purely as either a “personal” or “social” use – it is both. In studies with actual DTC genetics users, findings from Vayena et al.’s (2012) study in particular show the importance of curiosity, entertainment and enjoyment for people when they use test results to find genetic relatives online or in conversations with friends and family. In this survey of university students in Switzerland, the potential contribution to biomedical research was listed as the most important motivation for having these tests (Vayena et al., 2014). In addition, a qualitative study of online DTC test takers done in France found that all respondents had visited the dedicated blogs and forum run by the testing companies at least once. In some cases, they also used these platforms to share their own personal genetic information and establish, in the words of the authors, new forms of “biosociality” (Ducournau and Beaudevin, 2011; Ducournau et al., 2011).

Such findings undermine the portrayal of DTC genetic test takers as self-centered narcissists. The taking of DTC tests is typically not the solipsistic activity of an individual person, but something that is done with, or with reference to, family members, significant others, friends, or even society as a whole. In this sense, genomic information is personal and social at the same time: it is personal, but for *more than one person* (Laurie, 2001; Taylor, 2012; Widdows, 2013; Prainsack, in preparation). Reading one’s “personalized” health report can be an entertaining and interesting activity that does not serve the purpose of obtaining actionable health information or exploring one’s *genomic self*, but it also can be used to socialize on or offline, or to share data and information for social, research or philanthropic purposes.

Another possible use of personal genomic information obtained from DTC tests is the process of identity making. This aspect has been widely discussed within social science scholarship on DTC genetics (for an overview see Fishman and McGowan, 2014). In the early days of online genome-wide tests, journalists, biologists and other early adopters reported their experiences as genetic test-takers in the press, academic journals, and books (e.g., Duncan, 2009; Pinker, 2009; Angrist, 2010;

² These projects’ websites strike a very different tone than DTC services that promise personalized genetic risk profiles; the Gut Projects’ rhetoric revolves more around building a publicly accessible database to improve our understanding of the relationship between health and gut bacteria. Rather than receiving risk profiles, users see graphic representations of how their gut bacteria compare to other people whose bacteria have been analyzed. Although it remains to be seen whether users take their results as an incentive to eat healthier, more diverse diets, at this point, the question of what utility and value these tests have is still entirely open.

³ The Scripps Genomic Health Initiative is the first large-scale, longitudinal survey with several thousand voluntary participants who purchased a genomewide DTC test at a discounted rate from the former U.S.-based DTC company, Navigenics. The study sought to understand the behavioral and psychosocial impact of these data. Participants were surveyed for the first time when they decided to take the test, as well as three and twelve months after receiving the results. Out of the 3600 respondents who answered the initial survey, 1300 also completed both the three and twelve month follow-ups. The short-term follow-up survey after three months showed no measurable changes in anxiety, psychological health, diet, exercise or use of screening tests among these actual personal genomics users (Bloss et al., 2011). These findings contrast with those from hypothetical users (see, e.g., McBride et al., 2009; Cherkas et al., 2010; McGuire et al., 2009) and challenge some of the main assumptions in the discussion of DTC genetics: Here, test-results do not seem to have detrimentally affected test-takers; nor did results seem to have led to positive lifestyle changes. The twelve-month follow-up survey corroborated the trend found in results from the three-month survey and also indicated that customers felt rather satisfied with the test results, despite the lack of behavioral changes (Bloss et al., 2013).

⁴ There is no universally accepted definition of clinical utility. Besides narrow definitions of the term that focus on the impact that an action has on concrete health outcomes, other definitions of clinical utility comprise much wider ranges of usefulness, sometimes even including societal value (for an excellent overview see Grosse and Khoury, 2006). In this paper we use the term “clinical utility” to refer to a test’s ability to affect health outcomes (via informing treatment choice, etc.), as we consider this the most specific definition with the highest analytic value.

⁵ In fact, the Scripps Genomic Initiative was a longitudinal study that included an interview before the test and two follow-ups. However, the questionnaire collected information basically about users’ concerns and not their motivations of undergoing susceptibility genetic tests (Bloss et al., 2010, 2011, 2013).

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