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Online Genomes

Problematizing the disruptiveness of direct-to-consumer genetic tests

Abstract

Direct-to-social consumer genetic tests (DTCGT) provide basically individualized risk profiles for common diseases through social web techniques. Academic debates predominantly denounced them as a potentially harmful commercialization of genetics and hinged on questions of how laypeople would (mis)understand and (mis)use this information. DTCGT has also stimulated a heterogeneous set of methods and theoretical perspectives in social sciences, which have drawn on but also complicated a purely medical understanding of these devices.

The latter consider DTCGT as disruptive, but for a variety of reasons. The notion of *disruptiveness* is so used here as a conceptual map intends to point out to the different analytical approaches and issues related to DTCGT. Accordingly, three corpora are identified. The first one conceives disruption as the medical impact on the prospective patients. The second one proposes a biopolitical interpretation, by framing DTCGT as vectors of new governmentality based on charging individuals of the responsibility of disease prediction and prevention. The third one scrutinizes users' engagements with data and infrastructures at the intersection of informatics and genomics and locates DTCGT within the broader process of blossoming health digitalization. Accordingly, disruptive issues at stake concern mostly the political and economic aspects related to data sharing and crowdsourcing.

Key-words

Personal genomics/genetics
Direct-to-consumer genetic tests
Biomedical impact
Biopolitics
Digital biosociality
Digital health

Introduction: A disruptive innovation?

In the early 20th century, the technical standardization of thermometers made it possible for patients to take their own temperatures without the help of a doctor. This provoked an outcry among physicians, who complained about the “unauthorized appropriation” of medical competence and the possibility that patients could “control or even master’ the physicians by acquiring medical knowledge” (Hess, 2005: 119). A century later, the commercialization of another diagnostic device – predictive genetic tests for common disease – outside of the medical setting has raised very similar issues. Clinicians, life scientists and health regulators have denounced *direct-to-consumer genetic tests* (DTCGT)

for health purposes as a premature and potentially harmful transposition of genetic research into the market and questioned how laypeople would understand and use this information. This parallel calls for a reconsideration of the novelty of healthcare consumer genetics. To what extent are DTCGT new in comparison to (DTC) thermometers? This typically historical question acquires sociological pertinence as it resonates with the pivotal role that information plays in contemporary societies. The concept of “disruption” (Bower & Christensen, 1995) as the keyword of successful innovation, especially in the field of information technologies, point to the multi-layered effects that information may trigger. In the contemporary information age, a service (eg AirBnB) is disruptive if it is able to reconfigure conventional practice by mobilizing a technological-mediated cooperation between users and providers. Disruptiveness combines heterogeneous socio-technical features: free access to information, disintermediation of experts, digital platforms coordinating the connected actors, and a more efficient, spontaneous and transparent vision of social life. In this regard, it is interesting that DTC genetics is usually praised or criticized as a highly *disruptive* innovation, while ascribing different meanings to disruptiveness. The question then becomes, in what sense are DTCGT disruptive?

Before approaching this question, it is important to outline some details about DTCGT for health purposes. This industry began in the mid-1990s with small companies offering tests for a few genetic variants related to rare diseases or nutrient metabolism. These first attempts to commercialize genetics went nearly unnoticed. The matter changed dramatically in 2007, when multiple ambitious companies moved in the same period to offer innovative services, taking full advantage of recent developments in both genotyping techniques, particularly the next-generation sequencing, and new participatory tools of the Internet, the *social web*. This *second wave* of consumer personal genomics was inaugurated by three companies, tellingly dubbed the “Big Three”¹: 23andMe and Navigenics, two Californian start-ups funded by the biotech and IT industries, including Google and Genentech, and DECODEme, the commercial arm of the biotech company known for having sequenced a relevant portion of the entire, albeit small, nation of Iceland (DECODE)².

The Big Three offer consumers the possibility of obtaining several hundred thousand pieces of genetic data at an affordable price³. More specifically, these data predominantly concern an individual’s susceptibility to multifactorial, common conditions such as certain cancers and cardiovascular or degenerative diseases as well as responses to drugs (*pharmacogenomics*). The tests also provide *recreational* information about either ancestry or certain physical traits (hair and eye color, and even earwax type!), or proclivity to certain behaviors (addiction to smoking) or activities (endurance

¹ For a vivid sketch of the birth of personal genomics, see Angrist (2010).

² Navigenics and DECODEme have discontinued consumer services.

³ At the very beginning, the price ranged from \$2,500 (Navigenics) to \$399 (23andMe); it dropped to \$99 in 2012.

vs. sprinter)⁴. Finally, social web technologies make it possible to *access* users' personal data and to explore, browse, analyze and comment on them.

Companies market these services as revolutionary ways to empower patients and personalize healthcare through direct access to personal genomic data. Media outlets have covered DTCGT extensively, largely buying into the producers' rhetoric and reporting on many examples of individuals having their own genomes sequenced (O'Riordan, 2010b). However, as mentioned above, some regulators, policy advisors, clinicians and scientists have spoken out to denounce the drawbacks and dangers of DTCGT, such as potential violations of personal data privacy, the unreliability of data and their limited clinical validity and utility given that the only actions people can take to avoid developing diseases consist of fairly ordinary behaviors, such as being physically active or not smoking (eg Hunter, Khoury, and Drazen 2008). The most hotly contested issue is the lack of professional mediation in communicating complex susceptibility data, which might either falsely reassure customers or cause unwarranted anxiety (eg Lancet 2008). DTCGT companies have played strategically with the ambivalent nature of their data, whether recreational or medical, to avoid the kind of regulatory restrictions applied to genetic diagnostic devices, particularly the mediation of a physician in delivering such information (Curnutte & Testa, 2012). However, both proponents and critics share the same imaginary of DTCGT as medical devices that rethink "personalized medicine" through individualized risk profiles based on genetics (Tutton, 2014).

To return to our research question, the debate surrounding regulatory and bioethical issues frames DTCGT disruption in purely biomedical terms, particularly centered on the bioethical and regulatory debates. Consumer personal genomics has stimulated not only this *normative* debate but also a wide-ranging collection of methods and theoretical perspectives in the social sciences, thereby both drawing on and complicating a purely medical vision of DTCGT. What all these approaches have in common is the tendency to consider DTCGT a disruptive innovation, albeit for different reasons. The doctors' disintermediation goes along with practices and values related to new technological affordances of accessing and managing health-related information.

DTCGT disruptiveness can be thus associated with a new healthcare model, processes of individual empowerment or self-discipline, socio-technical practices based on personal bio-data, participation to biomedical research, and economic extraction of value. It is then possible to identify three major interpretations of disruptiveness corresponding to three underlying assumptions of practices around and technologies of information. Each of them resonates with a broad sociological framework, namely rational action theory, governmentality studies and ecological approaches to digital practices and infrastructures. These *underlying sociologies of information* and *meanings of disruptiveness* are here used to map the social sciences approaches to DTCGT – and provide a cartography that could be extended to the broader turn towards self-tracking practices and the digitalization and datafication of health (eg Lupton 2014).

⁴ Navigenics only provided health-related information.

The first line of research assumes that individual actors will use information in a social vacuum to attain a goal, and it shares with the normative debate a conception of disruption understood in terms of biomedical effects on prospective users. Specifically, it comprises surveys that investigate people's awareness, motivations, and attitudes regarding DTCGT aimed at bringing empirical evidence to the bioethical debate. The second line of inquiry understands personal genetic information in terms of its influence on personhood and subjectivity by focusing on the politics of medical development towards susceptibility and risk prediction. Drawing on Foucauldian theories of governmentality and biopolitics, these studies focus mainly on personal genomics discourses as vectors of broader trends in medicine (the personalization, prevention and prediction in healthcare) and politics (the individualization of risk management). The third one elaborates an ecological approach to information by taking into serious consideration the on-line and off-line environments where personal genetic data circulate and become meaningful. This literature, including several theoretical approaches, in particular science and technology studies (STS), internet studies and the socio-anthropology of health, analyzes users' practices and narratives with data and infrastructures at the intersection of informatics and genomics. The resulting *genomic digital ecologies* are analyzed as instantiations of the digitalization of health and experimentation with new practices and techniques surrounding health-related personal data. In this framework, the political and economic analysis of *sharing* genetic data as a new form of research participation and value creation will be granted particular relevance.

DTCGT: awareness, motivations and impact

In the immediate aftermath of the new generation of DTCGT, a number of empirical studies have been carried out to assess and predict the current and future impact of these tests⁵. They investigate people's awareness and adoption of and motivations and attitudes towards this presumably incipient and disruptive phenomenon in order to provide empirical evidence of the issues raised by the bioethical and regulatory debate. Both of them share a rationalistic interpretation of the subject and a cybernetic conception of communication, according to which information is used to attain a goal, and, accordingly, expect that genetic risk assessment will trigger innovative and personalized healthcare. As mainly physicians or bioethicists conduct this research, this corpus may be described as *sociology without sociologists*. Due probably to the difficulty of reaching the customers of a private service, the first studies are conducted on *hypothetical situations*, i.e. addressing several groups of people, considering them as *potential* or *experimental* test-takers – a feature which also clearly indicates researchers' perception that predicting the scope of the supposedly inexorable spread of DTCGT is a pressing task.

⁵ In 2012, a literature review of these studies counted more than 300 papers on this topic (McGuire et al., 2009).

A first group of studies address *potential users*, be they the general public of a single country⁶ or specific groups deemed more inclined to consumer genetics, such as people genetically *at risk* of various forms of cancer (Gray et al., 2009; Perez et al., 2011) or frequent users of the internet (McGuire, Diaz, Wang, & Hilsenbeck, 2009). In spite of the low rates of awareness and adoption,⁷ these studies “expect that the proportion of people who are aware of and use these tests will continue to increase” (Ortiz et al., 2011). Accordingly, one of their main concerns is to identify *potentially* marginalized socio-demographic segments of the population (Ortiz et al., 2011; Rahm et al., 2012) and, thus, to fulfill “the promise of genomics to improve personal and population health” (Kolor et al. 2012: 866). Another important goal is to explore users’ (*hypothetical*) motivations for potentially undergoing these tests (Cherkas, Harris, Levinson, Spector, & Prainsack, 2010; Gray et al., 2009) and/or their (equally *hypothetical*) attitudes, expectations and understandings of them (Leighton, Valverde, & Bernhardt, 2011; McGuire & Burke, 2008; Perez et al., 2011; Wilde, Meiser, Mitchell, & Schofield, 2010; Wilde, Meiser, Mitchell, Hadzi-Pavlovic, & Schofield, 2011). Those who are interested in pursuing DTCGT basically consider them to be *medical devices*. Among US social networkers, for example, “many respondents (34%) consider the information obtained from a PGT [*personal genomic testing*] to be a medical diagnosis and anticipate that it will influence their future healthcare decisions” (McGuire et al., 2009: 8). In general, these studies bring empirical evidence to the bioethical concerns, in particular *worrying* about overestimation of the clinical significance of DTCGT (McGuire et al., 2009), exposure to risk factors (Gray et al., 2009) or potential genetic stigma (Wilde et al., 2011), or *misunderstanding* due to the general public’s (or even physicians’) inadequate numerical literacy and difficulty in correctly understanding susceptibility risk factors for complex diseases (Leighton et al., 2011).

Other studies devise more creative research grounded in *experimental users* to “address research questions that more closely pertain to the ‘real world’ interface between the... DTC genetic testing industry and the public” (Bloss et al., 2010: 557). Some of them ask participants questions before offering real genetic testing to the general public (Gollust et al., 2011; McBride et al., 2009; Sanderson, O’Neill, Bastian, Bepler, & McBride, 2009) or to groups at risk, such as healthy people with a familial history of lung cancer (O’Neill et al., 2008; Sanderson et al., 2009). In keeping with previous research, such studies find that respondents are mainly interested in using these tests as motivational tools to adopt healthier behaviors. These intentions often do not translate into concrete action, however. The most striking results are from a longitudinal survey of several thousand *experimental users*. While the results before administering the test were similar to those discussed so far (Bloss et al., 2010), follow-up surveys after three (Bloss, Schork, & Topol, 2011) and twelve months (Bloss, Wineinger, Darst, Schork, & Topol,

⁶ The US (Kolor et al., 2012; Leighton et al., 2011), Australia (Wilde et al., 2010, 2011), the UK (Cherkas et al., 2010) or Puerto Rico (Ortiz et al., 2011).

⁷ In the US, for example, about one third of the general population knew about DTCGT and only 1 % used them (Wilde et al., 2011). Among specific groups, such as networkers, the percentage was a bit higher, with 60% aware of them and 6% using them (Wilde et al. 2011)

2013) show no measurable influence on test-takers in terms of anxiety, use of screening tests among test-takers, or positive lifestyle changes. Paradoxically, despite the lack of measurable effects, a large portion of the participants perceived the test to be of some kind of “personal utility” (Bloss et al., 2013), a new concept which seems to replace and compensate for the lack of clinical utility as a motivation to get tested.

Studies on *actual users* confirm these findings. According to the first important qualitative study addressing “early adopters” (McGowan, Fishman, & Lambrix, 2010), *actual users* get these tests mainly for health purposes. However, the users discover the limitations of the tests and the majority of them do not make changes in their behavior for health reasons. Another important motivation, as also confirmed by Su, Howard, & Borry (2011), is curiosity. While findings from other studies give different results, a recent review essay shows that, on balance, DTCGT have been neither as harmful as feared by critics nor as empowering as promised by proponents (see eg Covolo et al. 2015).

This literature analyzes DTCGT in different social settings, and, when they come to the real practices of the users, they conclude that they have little effect on modifying behavior, for better or worse. On the contrary, what emerges ultimately is that genetic data provide multiple forms of personal and social value, far “beyond clinical utility” (Turrini & Prainsack, 2016).

Moral pioneers or genetic entrepreneurs? The politics of genetic susceptibility

Other social approaches contextualize DTCGT within more longstanding and politically inflected debates on genetics, particularly in relation to the influential concept of “geneticization” (Lippman, 1992). This notion refers to the tendency to define human diseases or other differences exclusively through genetic makeup. In this regard, DTCGT have been understood as vectors of geneticization through a dual operation: they take advantage of DNA’s ability to represent a molecular substitute for each individual’s nature, and, simultaneously, they leave it up to users to discover themselves, thus presenting identification as a creative process of “individual self-determination” (Nordgren & Juengst, 2009).

Foucauldian analysis has focused on the political aspects of this ambivalent process, by framing it as an instantiation of self-discipline resulting from interactions between the genetic discourse on vitality, morbidity, and mortality and strategies for governing life. The notion of “governmentality” formulated in Foucault’s later work (2008) is often used to describe how contemporary neoliberal power is exercised non-coercively, through the construction of autonomous, free, and responsible subjects willing and able to choose for and govern themselves. Personal genetic information, particularly in relation to the predictive capacity of risk, becomes a tool of neoliberal governmentality, which deploys its disruptiveness by extending the medical gaze on optimizing health rather than healing diseases (Rose, 2007a). Genetic risk thus extends the medical gaze to healthy people, considering them “asymptomatic patients” suffering from future diseases that must be treated in the present with preventive measures. Studies on DTCGT cast them as a laboratory of broader political trends giving rise to new techniques and strategies

through which individuals can *manage their own genes*. DTCGT are not only diagnostic devices involved in medical practice, they are also “technologies of the self” operating in a regime of *molecular biopolitics*. The same conceptual analysis leads to two radically different takes on this power, as some emphasize the emancipatory aspects while others focus on the disciplinary ones.

Some scholars argue that the possibility to genetically decipher themselves, that is, to decipher one’s own biological constitution at the molecular level, offers opportunities for bottom-up claims-making and political empowerment. By introducing the notion of “biological citizenship”, Rose and Novas (2005) highlight how the molecular perspective opens up new spaces of political intervention. Individuals mobilize individually or collectively to seek information about their condition and take responsibility for their own health without resignation and independently of medical paternalism and state mediation. DTCGT users are thus examples of “moral pioneers” (Rapp, 1999) who experience the possibility of deciphering life at the molecular level and take responsibility for this new knowledge. Also, in this view, biological classifications in individuals, families, lineages, communities, population, and races become democratic tools deployed first-hand by the interested parties according to logics that are “no longer inspired by the dream of the taking charge of the lives of each in the name of the destiny of all” (Rose 2007b, 62). Tellingly, when Rose analyzes DTCGT, he addresses predisposition to disease together with ancestry, framing both as strategic fields of agency open to being appropriated from below and deployed for concrete actions (Rose, 2008).

Other studies fully embrace this conceptual framing but with the opposite political valence. They cast the agency of patients/citizens as purely individual and economically interested; rather than pioneers, they are described as entrepreneurs. In her studies on nutrigenomics, Harvey (2009, 130) speaks of “genetic entrepreneurs” as figures who employ susceptibility “to create a future that maximizes their ‘vital capital’ by ensuring the optimal functioning of their unique genome”. Accordingly, people pursue health and wellness preservation and optimization not as a citizenship-based claim for patient empowerment but rather as an ethical subjection to the purchase of goods, such as genetic information about risk (Harvey, 2010). Likewise, Ducournau and colleagues (2011; 2013) in their analysis of commercial testing websites and users’ practices situate online personal genomics as part of a broader political shift towards an “individualized biopolitics”, i.e., the management of life through delegation to individuals. The very meaning of citizens’ making a donation to medicine, classically understood as an altruistic act aimed at confirming the civic bond between citizens, is reconceptualized in individualistic and economical terms (Tutton & Prainsack, 2011). These perspectives parallel arguments critiquing the personalistic direction medicine has taken and the deleterious effects this may have on the collectively-oriented organization of healthcare (McGuire & Burke, 2008; Dickenson, 2013).

Both these takes on DTCGT, in spite of the differences of their political evaluations, tend to reason in terms of the opposition between empowerment and self-discipline. Information is used monolithically in support of either one political process or the other.

This tendency makes it more difficult to acknowledge the ecological complexities of the heterogeneous networks constructed on the basis of genome scans.

Users' engagement with data, platforms and narratives: Contextualizing DTCGT in biodigital ecologies

The literature discussed thus far is focused on the biomedical aspects of personal genomics, addressing either its impact on single prospective patients or the broader and long-term political consequences of individualized genetic profiling. However, they do not contextualize information within the digital ecologies developing around genetic data that, in hindsight, have played a crucial role in shaping the development of this market. Online personal genomics has not grown into the large and lucrative industry it was expected to become. It also continues to be dominated by genetic ancestry and relatedness, with health-related services forming a small and volatile, albeit very lively, niche industry. New companies have popped up in many countries, even outside of North America and Europe, particularly in Asia. Their services, which include either tests or simply the interpretation of data produced elsewhere, are increasingly specialized in sectors such as physical activity, nutrigenomics, common diseases, dating, reproductive services, and so forth. More health-oriented companies such as Navigenics and DECODEme have discontinued consumer services, whereas 23andMe, which bet on the social web, plays a dominant role with more than 2 millions customers tested as of April 2017 (Herper, 2017).

Research in STS, internet studies and the socio-anthropology of biomedicine have analyzed customers' practices, companies' discourses and other online platforms where this biodigital information circulates. Users' engagement with genetic data and online platforms has been redefining our understanding of the body, health, and disease in a historical context in which new cultures of healthism are developing outside of clinics and strictly medical discourse (Turrini, 2015). Apparently irrelevant aspects, such as filling up the genetic test kit bought online with saliva, are interpreted as a culturally relevant instance of converting the body into information (see fig.1).⁸ According to this literature, it would be a mistake to read this practice as a sort of disembodiment or disappearance of the body; rather, it integrates biology into information systems (O'Riordan, 2011). Drawing on Castells, Levina (2010) imagines DTCGT as a way of extending our biology and DNA to the "network society" whose members are expected and encouraged to engage in "continuous and constant sharing of one self with others" (Levina 2010: 1). For O'Riordan (2010, 2013), DTCGT convert bodies into digital texts of which users are at the same time the content and the reader. These "digital media artifacts" address "digital genome publics" and construct new media cultures based on genomics. The second wave of DTCGT companies market their products as a matter of "big data" (Saukko, 2017). The clinical significance of this information is less important than its "abundance," both

⁸ Many other studies have also underlined the cultural meanings of the gesture of spitting as the conversion of a bodily excretion into data (Harris, Kelly, & Wyatt, 2014; Harris et al., 2016, 2012; Kragh-Furbo & Tutton, 2017; O'Riordan, 2010; Pálsson, 2009).

quantitative and semantic. Thanks to the possibility of accessing “raw data,” personal genome scans lend themselves to digital circuits of potentially infinite signification and interpretation through the simple employment of user-friendly software freely available on the web.

----insertfig.1----

Viewed through these analytical perspectives, any clear distinction between medical and non-medical blurs. What comes to the fore is the complexity of the relationships taking form around genetic data, including more playful and unforeseen uses. In their analysis of the videos that DTCGT users have posted on YouTube, Harris and colleagues (2014) coin the term “autobiologies” to grasp this sense of hybridity and playfulness. Like illness narratives, these videos are based on health monitoring; like “stories of consumption and experimentation”, they consist in reading and commenting on new kinds of health-related data through the various informational sources (databases, graphics, advice) available on the web. Spitting into the “spittoon”, although perceived as “gross”, is filmed as an interesting part of their experience, whereas the reliability of results about disease outcomes is not given careful consideration, for these tests are used not for diagnostic purposes but as a way of thinking about other topics, especially family history. In a similar vein, Ruckenstein (2017) emphasizes the creativity through which users “keep data alive” through multiple and unforeseen online/offline uses and purposes. She argues that test takers willingly position themselves as “data subjects”, an observation which clearly locates the disruptiveness of DTCGT as part of the turn towards the access, production and/or use of personal health-related data.

Through a closer analysis of genomic digital ecologies, the question of users’ lack of relationship with health professionals also takes on more complexity. Scholars find that there are different socio-technical architectures between companies (Parthasarathy, 2010), especially given that these services have been integrated to different extents into the field of medical expertise to respond to the varying requirements of regulatory agencies. Rather than a lack of mediation, what has occurred is a *reconfiguration of genetic expertise* in new temporal-spatial locations on the internet (Harris, Kelly, & Wyatt, 2013; Prainsack & Vayena, 2013).

In general, these approaches have emphasized a kind of relationship around genetic data that recasts the notion of “biosociality” (Rabinow, 1992) in a digital environment. In its original formulation, biosociality referred to the phenomenon of groups focused on patient advocacy and health activism taking shape around shared genetic factors. In the case of personal genomics, the phenomenon is instead one of *digital biosociality*, created when healthy people browse, download, upload, retrieve, pool, and share huge amounts of dispersed, personal digital data for a heterogeneous set of reasons, often personal or familial, often without any clear distinction between ancestry and health. The political issue that emerges here is how the technological affordance of the web to interact and share may imply an obligation to do it dictated by social, coercive

mechanisms. The disruptiveness thus shifts to the political and economic issues at stake in *participation*, one of the hallmarks of digital cultures.

Citizen science, labor and searching for the unicorn: the politics and economy of participation

While there has been a heated discussion about the potential risks to people who receive DTCGT, few studies have analyzed how participation in biomedical research is conceived of and practiced (Lee & Crawley, 2009). And yet 23andMe, the current global leader in this industry, has integrated participation into its business plan and asks customers to donate their genetic data and to complement them with clinical data. These strategies form part of a broader tendency in biomedicine to take advantage of social web tools to extract data from spontaneously constituted cohorts (Allison, 2009; Swan, 2012). Crowdsourcing personal genomic data mobilizes the crowds' *size* rather than its *wisdom*, and yet it is presented as a revolutionary opportunity for consumers to contribute to biomedical research and the collective good.

“Our research arm, 23andWe, gives customers the opportunity to leverage their data by contributing it to studies of genetics... [and this] can produce revolutionary findings that will benefit us all”. This quotation from 23andMe website clearly shows the conflation between market and politics, as customers are imagined as the active authors of a change in favor of the collective good. According to Prainsack (2011: 155) this hybridization “fits into the bigger picture of citizen science”, the movement seeking to make scientific expertise more open by involving laypeople in research design, funding or data collection and analysis. 23andMe has reinvented participation by drawing on practices originating in free software digital cultures (such as data sharing) as well as market strategies such as fidelity programs based on exchanging personal data for small rewards or trust in the company (Prainsack, 2011). This firm has been able to align for-profit aims, such as filing a number of patent applications, with a seemingly genuine commitment to facilitating research, particularly by allowing members access to their own raw data (Prainsack, 2014). Still, important issues about the quality of data being produced and, most importantly, the power asymmetry between the company and its users remain.

Critical perspectives on participation have analyzed the economic implications of users' engagements. Freely donated genetic personal data have been understood as a specific form of “free labor” that digital industry is able to extract through the pleasurable and voluntary activities of digital technology users while they are online (Terranova, 2000). Citizen science is thus basically interpreted as a new frontier of social web economic accumulation (Levina, 2010) which takes form through the compulsion to interact, to be connected (O’Riordan, 2011). The gift implied in this case goes beyond information to also entail biological substance, which is the key to accessing the body. It may also be interpreted, therefore, as a form of “clinical labor” (Cooper & Waldby, 2014) through which bodily parts produce both scientific and economic value in the circuits of the bioeconomy (Harris, Wyatt, & Kelly, 2012; Palsson, 2014). In this context, racial differences are an important issue at stake; several initiatives of 23andMe to recruit Afro-

American clearly show to what extent data on minorities are at the same time scientifically interesting, economically profitable, and politically legitimizing (Merz, 2016). By instantiating the conflation of production and consumption (*prosumption*), these scholars argue, participation in DTCGT constitutes a form of profitable extraction concealed by the rhetoric of patient empowerment, open-access and sharing, the economic value of which has not been acknowledged by users.

Participation has also been integrated into the business plan of 23andMe. It is a pillar of a “two-sided data-banking market model” (Stoeklé et al., 2016) whose core business entails not simply selling inexpensive genetic tests for ancestry or health, but rather establishing a high-value genetic database for research. Moreover, the company’s commitment to citizens and the democratization of science are part and parcel of a rhetoric designed to attract venture capital investment. Combining economic value and moral worth is a key strategy for presenting an innovation as disruptive, and this has actually played a crucial role in making 23andMe the only “unicorn” (a successful start-up, in financial jargon) in the field of personal genomics (Hogarth, 2017).

The disruptive force of participation crosses citizen science and consumption economic process of extraction of personal health-related data and the ideals of genomics and citizens’ empowerment. It is also important to recall that crowdsourcing personal genetic data has spread beyond the market, as a wide range of non-proprietary user-generated databases has adopted this practice. These initiatives are committed to highlighting the scientific and economic importance of health-related personal information and fostering new participatory practices through personal genetic (and clinical) data sharing and crowdsourcing (Riso et al., 2017).

Conclusions

DTCGT are *socio-technical innovations* which, by leveraging the convergence between genetics and informatics, are blurring the boundaries between: doctors and patients; experts and laypeople; patients, healthy people, and research participants; and knowledge production and economic exploitation. It is no coincidence that the disruptiveness of these devices has been at the heart of intense and heterogeneous debates in ethics, regulation, and the social sciences. The latter, in particular, have deployed a wide set of analytical perspectives and issues to problematize the meaning of disruptiveness as related to their implicit sociology of information.

Three groups have been identified and associated to three broad sociological frameworks, rational actor theory, governmentality studies and ecological approaches to health-related data and platforms. Taken together, they delineate a research trajectory that sets off from explorations of the biomedical impact of DTCGT on prospective and actual patients and goes on to explore the political processes involved in the spread of genetic susceptibility and, finally, users’ practices on and around genetic data, especially but not exclusively on online platforms. While earlier research centered on bioethical and regulatory concerns, this newer research trajectory highlights the ways in which user engagement with data and platforms has shaped the development of this industry.

This classification does not intend to be hierarchical. Its aim is rather to underscore the differences developed in the debate and to take advantage of them for future analysis of DTC genomics and analogous personal health-related data production and circulation on the web. It is impossible to predict which sense of disruptiveness among those described will prevail in personal genomics. Future genetic tests might have a considerable biomedical impact, as foreseen by the first group of studies. Personalization, commercialization and digitalization may also lead people to develop an enterprising relationship to our health and bodies, and this could have negative effects on public healthcare services, as the second group seems to suggest. At any rate, it is important to crack open the *black box* of biodigital practices, technologies and environments in which this health-related information is produced, interpreted and collected. The first lines of inquiry display a rather simplistic comprehension of the internet, as they understand it as either a means of further stratifying and marginalizing innovative biomedical practices or a mere accelerator of existing political dynamics. A more detailed scrutiny reveals how *genome digital ecologies*, i.e. the flows of data (online and offline) and platforms, play an active role in reconfiguring the notions of self, body, health, wellness and disease through now constellations of consumers, patients, citizens, medical professionals, researchers, and private companies.

References

- Allison, M. (2009). Can web 2.0 reboot clinical trials? *Nature Biotechnology*, 27(10), 895–903.
- Angrist, M. (2010). *Here Is a Human Being: At the Dawn of Personal Genomics*. New York: HarperCollins.
- Bloss, C. S., Ornowski, L., Silver, E., Cargill, M., Vanier, V., Schork, N. J., & Topol, E. J. (2010). Consumer perceptions of direct-to-consumer personalized genomic risk assessments. *Genetics in Medicine*, 12(9), 556–566. <http://doi.org/10.1097/GIM.0b013e3181eb51c6>
- Bloss, C. S., Schork, N. J., & Topol, E. J. (2011). Effect of Direct-to-Consumer Genomewide Profiling to Assess Disease Risk. *The New England Journal of Medicine*, 364(6), 524–34. <http://doi.org/10.1056/NEJMoa1011893>
- Bloss, C. S., Wineinger, N. E., Darst, B. F., Schork, N. J., & Topol, E. J. (2013). Impact of direct-to-consumer genomic testing at long term follow-up. *Journal of Medical Genetics*, 50(6), 393–400. <http://doi.org/10.1136/jmedgenet-2012-101207>
- Bower, J. L., & Christensen, C. M. (1995). Disruptive technologies: catching the wave. *Harvard Business Review*, (Jan-Feb), 43–53. *Harvard Business Review*, (Jan-Feb), 43–53.
- Cherkas, L. F., Harris, J. M., Levinson, E., Spector, T. D., & Prainsack, B. (2010). A Survey of UK Public Interest in Internet-Based Personal Genome Testing. *PloS One*, 5(10), e13473. <http://doi.org/10.1371/journal.pone.0013473>
- Cooper, M., & Waldby, C. (2014). *Clinical Labor: Tissue Donors and Research Subjects in the Global Bioeconomy*. Durham and London: Duke University Press.
- Covolo, L., Rubinelli, S., Ceretti, E., & Gelatti, U. (2015). Internet-Based Direct-to-

- Consumer Genetic Testing : A Systematic Review. *Journal of Medical Internet Research*, 17(12), e279. <http://doi.org/10.2196/jmir.4378>
- Curnutte, M., & Testa, G. (2012). Consuming genomes: Scientific and social innovation in direct-to-consumer genetic testing. *New Genetics and Society*, 31(2), 159–181. <http://doi.org/10.1080/14636778.2012.662032>
- Dickenson, D. (2013). *Me Medicine vs. We Medicine: Reclaiming Biotechnology for the Common Good*. New York and Chichester, UK: Columbia University Press.
- Ducournau, P., & Beaudevin, C. (2011). Génétique en ligne. Déterritorialisation des régulations de santé publique et formes de développement commercial. *Anthropologie et Santé. Revue Internationale Francophone D'anthropologie de La Santé*, 3, 1–18.
- Ducournau, P., Gourraud, P.-A., Rial-Sebbag, E., Cambon-Thomsen, A., & Bulle, A. (2013). Direct-to-consumer health genetic testing services: What commercial strategies for which socio-ethical issues? *Health Sociology Review*, 22(1), 75–87.
- Foucault, M. (2008). *The birth of biopolitics : lectures at the Collège de France, 1978-79*. New York: Palgrave Macmillan.
- Gollust, S. E., Gordon, E. S., Zayac, C., Griffin, G., Christman, M. F., Pyeritz, R. E., ... Bernhardt, B. a. (2011). Motivations and perceptions of early adopters of personalized genomics: Perspectives from research participants. *Public Health Genomics*, 15(1), 22–30. <http://doi.org/10.1159/000327296>
- Gray, S. W., O'Grady, C., Karp, L., Smith, D., Schwartz, J. S., Hornik, R. C., & Armstrong, K. (2009). Risk information exposure and direct-to-consumer genetic testing for BRCA mutations among women with a personal or family history of breast or ovarian cancer. *Cancer Epidemiology Biomarkers and Prevention*, 18(4), 1303–1311. <http://doi.org/10.1158/1055-9965.EPI-08-0825>
- Harris, A., Kelly, S. E., & Wyatt, S. (2013). Counseling customers: Emerging roles for genetic counselors in the direct-to-consumer genetic testing market. *Journal of Genetic Counseling*, 22(2), 277–288. <http://doi.org/10.1007/s10897-012-9548-0>
- Harris, A., Kelly, S. E., & Wyatt, S. (2014). Autobiologies on YouTube: Narratives of Direct-to-Consumer Genetic Testing. *New Genetics and Society*, 33(1), 60–78. <http://doi.org/10.1080/14636778.2014.884456>
- Harris, A., Kelly, S. E., & Wyatt, S. (2016). *Cybergenetics: Health Genetics and New Media*. London and New York: Routledge.
- Harris, A., Wyatt, S., & Kelly, S. E. (2012). The Gift of Spit (and the Obligation To Return It): How consumers of online genetic testing services participate in research. *Information, Communication & Society*, 16(2), 236–257. <http://doi.org/10.1080/1369118X.2012.701656>
- Harvey, A. (2009). From genetic risk to post-genomic uncertainties: Nutrigenomics and the birth of the genetic entrepreneur. *New Genetics and Society*, 28(2), 119–137. <http://doi.org/10.1080/14636770902901447>
- Harvey, A. (2010). Genetic risks and healthy choices: Creating citizen-consumers of genetic services through empowerment and facilitation. *Sociology of Health and Illness*, 32(3), 365–381. <http://doi.org/10.1111/j.1467-9566.2009.01202.x>
- Herper, M. (2017). 23andMe Rides Again: FDA Clears Genetic Tests To Predict Disease Risk. *Forbes*.
- Hess, V. (2005). Standardizing body temperature: Quantification in hospitals and daily life, 1850-1900. In G. Jorland, A. Opinel, & G. Weisz (Eds.), *Body Counts: Medical Quantification in Historical and Sociological Perspectives* (pp. 109–126). Montreal-Kingston: McGill-Queen's University Press.

- Hogarth, S. (2017). Valley of the unicorns: consumer genomics, venture capital and digital disruption. *New Genetics and Society*, 36(3), 250–272.
<http://doi.org/10.1080/14636778.2017.1352469>
- Hunter, D. J., Khoury, M. J., & Drazen, J. M. (2008). Letting the genome out of the bottle: Will we get our wish? *New England Journal of Medicine*, 358(2), 105–107.
<http://doi.org/10.1056/NEJMp1415160>
- Kolor, K., Duquette, D., Zlot, A., Foland, J., Anderson, B., Giles, R., ... Khoury, M. J. (2012). Public awareness and use of direct-to-consumer personal genomic tests from four state population-based surveys, and implications for clinical and public health practice. *Genetics in Medicine*, 14(10), 860–867.
<http://doi.org/10.1038/gim.2012.67>
- Kragh-Furbo, M., & Tutton, R. (2017). Spitting images: remaking saliva as a promissory substance. *New Genetics and Society*, 36(2), 159–185.
<http://doi.org/10.1080/14636778.2017.1320943>
- Lancet. (2008). Direct-to-consumer genetic tests: flawed and unethical. *The Lancet Oncology*, 9(12), 1113. [http://doi.org/10.1016/S1470-2045\(08\)70288-2](http://doi.org/10.1016/S1470-2045(08)70288-2)
- Lee, S. S. J., & Crawley, L. (2009). Research 2.0: Social networking and direct-to-consumer (DTC) genomics. *American Journal of Bioethics*, 9(6–7), 35–44.
<http://doi.org/10.1080/15265160902874452>
- Leighton, J. W., Valverde, K., & Bernhardt, B. a. (2011). The general public's understanding and perception of direct-to-consumer genetic test results. *Public Health Genomics*, 15(1), 11–21. <http://doi.org/10.1159/000327159>
- Levina, M. (2010). Googling Your Genes: Personal Genomics and the Discourse of Citizen Bioscience in the Network Age. *Journal of Science Communication*, 9(1), 1–8.
- Lippman, A. (1992). Led (astray) by genetic maps: The cartography of the human genome and health care. *Social Science & Medicine*, 35(12), 1469–1476.
[http://doi.org/10.1016/0277-9536\(92\)90049-V](http://doi.org/10.1016/0277-9536(92)90049-V)
- Lupton, D. (2014). Critical Perspectives on Digital Health Technologies. *Social Compass*, 12(8), 1344–1359. <http://doi.org/10.1111/soc4.12226> Critical
- McBride, C. M., Alford, S. H., Reid, R. J., Larson, E. B., Baxevanis, A. D., & Brody, L. C. (2009). Characteristics of users of online personalized genomic risk assessments: implications for physician-patient interactions. *Genetics in Medicine*, 11(8), 582–587. <http://doi.org/10.1097/GIM.0b013e3181b22c3a>
- McGowan, M. L., Fishman, J. R., & Lambrix, M. A. (2010). Personal genomics and individual identities: motivations and moral imperatives of early users. *New Genetics and Society*, 29(3), 261–290.
<http://doi.org/10.1080/14636778.2010.507485>
- McGuire, A. L., & Burke, W. (2008). An Unwelcome Side Effect of Direct-to-Consumer: Raiding the Medical Commons. *JAMA: The Journal of the American Medical Association*, 300(22), 13–15.
- McGuire, A. L., Diaz, C. M., Wang, T., & Hilsenbeck, S. G. (2009). Social networkers' attitudes toward direct-to-consumer personal genome testing. *The American Journal of Bioethics: AJOB*, 9(6–7), 3–10.
<http://doi.org/10.1080/15265160902928209>
- Merz, S. (2016). “Health and ancestry start here”: Race and presumption in direct-to-consumer genetic testing services. *Ephemera*, 16(3), 119–140.
- Nordgren, a., & Juengst, E. T. (2009). Can genomics tell me who I am? Essentialistic rhetoric in direct-to-consumer DNA testing. *New Genetics and Society*, 28(2), 157–172. <http://doi.org/10.1080/14636770902901595>

- O'Neill, S. C., White, D. B., Sanderson, S. C., Lipkus, I. M., Bepler, G., Bastian, L. a, & McBride, C. M. (2008). The feasibility of online genetic testing for lung cancer susceptibility: uptake of a web-based protocol and decision outcomes. *Genetics in Medicine*, 10(2), 121–130. <http://doi.org/10.1097/GIM.0b013e31815f8e06>
- O'Riordan, K. (2010a). *The Genome Incorporated: Constructing Biodigital Identity*. Farnham (UK) & Burlington: Ashgate.
- O'Riordan, K. (2010b). Writing biodigital life: Personal genomes and digital media. *Biography*, 34(1), 119–131. <http://doi.org/10.1353/bio.2011.0001>
- O'Riordan, K. (2011). Revisiting digital technologies: Envisioning biodigital bodies. *Communications*, 36(3), 291–312. <http://doi.org/10.1515/comm.2011.015>
- O'Riordan, K. (2013). Biodigital Publics: Personal Genomes as Digital Media Artefacts. *Science as Culture*, 22(4), 516–539. <http://doi.org/10.1080/14636778.2013.764069>
- Ortiz, A. P., López, M., Flores, L. T., Soto-Salgado, M., Finney Rutten, L. J., Serrano-Rodriguez, R. a, ... Tortolero-Luna, G. (2011). Awareness of Direct-to-Consumer Genetic Tests and Use of Genetic Tests Among Puerto Rican Adults, 2009. *Preventing Chronic Disease*, 8(5), A110. <http://doi.org/A110> [pii]
- Palsson, G. (2014). LabouringMe, LabouringUs. In B. Prainsack, S. Schicktanz, & G. Werner-Felmayer (Eds.), *Genetics as Social Practice: Transdisciplinary Views on Science and Culture* (pp. 165–180). Farnha: Ashgate.
- Pálsson, G. (2009). Spitting Image. *Anthropology Now*, 1(3), 12–22.
- Parthasarathy, S. (2010). Assessing the social impact of direct-to-consumer genetic testing: Understanding sociotechnical architectures. *Genetics in Medicine : Official Journal of the American College of Medical Genetics*, 12(9), 544–7. <http://doi.org/10.1097/GIM.0b013e3181e71c70>
- Perez, G. K., Cruess, D. G., Cruess, S., Brewer, M., Stroop, J., Schwartz, R., & Greenstein, R. (2011). Attitudes toward direct-to-consumer advertisements and online genetic testing among high-risk women participating in a hereditary cancer clinic. *Journal of Health Communication*, 16(6), 607–628. <http://doi.org/10.1080/10810730.2011.551993>
- Prainsack, B. (2011). Voting with their mice: personal genome testing and the “participatory turn” in disease research. *Accountability in Research*, 18(3), 132–47. <http://doi.org/10.1080/08989621.2011.575032>
- Prainsack, B. (2014). Understanding participation: the “citizen science” of genetics. In B. Prainsack, S. Schicktanz, & G. Werner-Felmayer (Eds.), *Genetics as Social Practice: Transdisciplinary Views on Science and Culture* (pp. 147–164). Farnham: Ashgate.
- Prainsack, B., & Vayena, E. (2013). Beyond the clinic: “direct-to-consumer” genomic profiling services and pharmacogenomics. *Pharmacogenomics*, 14(4), 403–12. <http://doi.org/10.2217/pgs.13.10>
- Rabinow, P. (1992). Artificiality and Enlightenment: From Sociobiology to Biosociality. In J. Crary & S. Kwinter (Eds.), *Zone 6: Incorporations* (Zone, pp. 234–252). New York: Zone.
- Rahm, A. K., Feigelson, H. S., Wagner, N., Le, A. Q., Halterman, E., Cornish, N., & Dearing, J. W. (2012). Perception of direct-to-consumer genetic testing and direct-to-consumer advertising of genetic tests among members of a large managed care organization. *Journal of Genetic Counseling*, 21(3), 448–461. <http://doi.org/10.1007/s10897-011-9477-3>
- Rapp, R. (1999). *Testing women, testing the fetus : the social impact of amniocentesis in America*. New York and London: Routledge.

- Riso, B., Tupasela, A., Vears, D. F., Felzmann, H., Cockbain, J., Loi, M., ... Rakic, V. (2017). Ethical sharing of health data in online platforms – which values should be considered? *Life Sciences, Society and Policy*, 13(1), 12. <http://doi.org/10.1186/s40504-017-0060-z>
- Rose, N. (2007a). Genomic susceptibility as an emergent form of life? Genetic testing, identity, and the remit of medicine. In R. V. Burri & J. Dumit (Eds.), *Biomedicine as culture: Instrumental practices, technoscientific knowledge, and new modes of life*. New York-London: Routledge.
- Rose, N. (2007b). *The Politics of Life Itself: Biomedicine, Power, and Subjectivity in the Twenty-First Century*. Princeton: Princeton University Press.
- Rose, N. (2008). Race, risk and medicine in the age of “your own personal genome.” *BioSocieties*, 3, 423–439. <http://doi.org/10.1017/S1745855208006339>
- Rose, N., & Novas, C. (2005). Biological Citizenship. In A. Ong & S. J. Collier (Eds.), *Global Assemblages* (pp. 439–463). Oxford: Blackwell. <http://doi.org/10.1002/9780470696569.ch23>
- Ruckenstein, M. (2017). Keeping data alive: talking DTC genetic testing. *Information Communication and Society*, 20(7), 1024–1039. <http://doi.org/10.1080/1369118X.2016.1203975>
- Sanderson, S. C., O’Neill, S. C., Bastian, L. A., Bepler, G., & McBride, C. M. (2009). What can interest tell us about uptake of genetic testing? intention and behavior amongst smokers related to patients with lung cancer. *Public Health Genomics*, 13(2), 116–124. <http://doi.org/10.1159/000226595>
- Saukko, P. (2017). Shifting metaphors in direct-to-consumer genetic testing: from genes as information to genes as big data. *New Genetics and Society*, 36(3), 296–313. <http://doi.org/10.1080/14636778.2017.1354691>
- Stoeklé, H.-C., Mamzer-Bruneel, M.-F., Vogt, G., & Hervé, C. (2016). 23andMe: a new two-sided data-banking market model. *BMC Medical Ethics*, 17(19). <http://doi.org/10.1186/s12910-016-0101-9>
- Su, Y., Howard, H. C., & Borry, P. (2011). Users’ motivations to purchase direct-to-consumer genome-wide testing: an exploratory study of personal stories. *Journal of Community Genetics*, 2(3), 135–46. <http://doi.org/10.1007/s12687-011-0048-y>
- Swan, M. (2012). Crowdsourced health research studies: An important emerging complement to clinical trials in the public health research ecosystem. *Journal of Medical Internet Research*, 14(2), 186–198. <http://doi.org/10.2196/jmir.1988>
- Terranova, T. (2000). Free Labor: Producing Culture for the Digital Economy. *Social Text*, 18(2), 33–58. http://doi.org/10.1215/01642472-18-2_63-33
- Turrini, M. (2015). A genealogy of “healthism.” *Eä - Journal of Medical Humanities & Social Studies of Science and Technology*, 7(1), 11–27.
- Turrini, M., & Prainsack, B. (2016). Beyond clinical utility: The multiple values of DTC genetics. *Applied and Translational Genomics*, 8, 4–8. <http://doi.org/10.1016/j.atg.2016.01.008>
- Tutton, R. (2014). *Genomics and the reimagining of personalized medicine*. Farnham, UK: Ashgate.
- Wilde, A., Meiser, B., Mitchell, P. B., & Schofield, P. R. (2010). Public interest in predictive genetic testing, including direct-to-consumer testing, for susceptibility to major depression: preliminary findings. *European Journal of Human Genetics*, 18(1), 47–51. <http://doi.org/10.1038/ejhg.2009.138>
- Wilde, A., Meiser, B., Mitchell, P., Hadzi-Pavlovic, D., & Schofield, P. (2011). Community interest in predictive genetic testing for susceptibility to major depressive disorder

in a large national sample. *Psychological Medicine*, 41(8), 1605–1613.
<http://doi.org/10.1017/S0033291710002394>