

Ethical perspectives of direct-to-consumer genetic testing


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Introduction

Genomics, the study of entire genetic sequence of human body, is a relatively new discipline in public health. Owing to its inherent individual-centric, personal approach it is surrounded by controversies and ethical dilemmas in the domain of public health. The controversies revolve around the idea that the theory and practice of genomics contradict the tenet, organization and overall philosophy of public health. After decades of research, complete human genome sequence was published in Science in 2009 (Cook-Deegan & Heaney, 2010). The sequencing itself was claimed as a marvelous achievement and propounded to ensure further advancement of public health and medical science.

Genetic screening/testing- the outcome of better understanding of human genome, has raised several questions which were never asked before, the criticism which was never raised before and the dilemmas that were never felt before (Knoppers, Avard, & Howard, 2010). The approaches of genetics were never tried before the way they are being done in the name of direct-to-consumer (DTC) genetic testing. This aspect of genetic testing has raised some additional questions related to marketing and business approach of genomics together with the overall controversies of genetics itself.

In DTC genetic testing, a consumer directly orders a particular type of genetic test by sending specimen via mail to a laboratory or company, which in turn creates their genetic profiles according to the request and then sends it back to the consumer. People choose DTC for three purposes: 1. To gain information about their ancestry 2. To know their susceptibility towards certain diseases or risk factors 3. Just for the curiosity on their genetic makeup (Su, 2013). After testing, the company sends the result to the consumer via mail or internet sources. The process completes without the presence of physician or genetic counselor. This approach is entirely

different from provider-user interaction model. However, some companies have developed a model that the consumer should have consulted a physician to be able to order a test (Howard & Borry, 2012). Otherwise internet-based information on websites of the testing company is the main source of information for the consumer.

Dilemmas related to DTC genetic tests are not independent of the overall controversies of genetic testing. It has been able to draw the concern of health workers, policy makers, administrators and all the stakeholders of health care systems. This paper aims to discuss the ethical perspectives of direct-to-consumer genetic testing.

Ethical dilemmas of DTC genetic testing

First dilemma starts with the quality assurance. Obtaining precise result and making a valid interpretation of the results are huge challenges in the absence of involvement of genetic experts and counselors. A study conducted among 2100 American adults throughout the country showed that more than half (62%) said that physicians should be involved in the explanation of genetic results. This fact challenges the legitimacy of practice of DTC without the involvement of a physician or any health worker for that matter (Almeling & Gadarian, 2013). On the flip side, supervision by the physicians for the purpose of counseling and interpretation of the genetic information has been shadowed by the research findings which show that there is limitation of knowledge among physicians regarding genetic information (Bloss, Darst, Topol, & Schork, 2011; Goldsmith, Jackson, O'Connor, & Skirton, 2013).

Validity and reliability of genetic testing should be highly ensured while marketing the technology, so the message communicated should be warranted. Tests must be standardized in relation to the other types of tests available. DTC genetic testing should be regulated within the

framework of pharmaceutical regulations and quality control mechanisms. Pre and post marketing quality testing should be done to ensure safety and accuracy of the test (Bowen, Battuello, & Raats, 2005).

Genetic experts claim that it is difficult to conduct large RCTs to ensure validity because of practical difficulties in matching (Bloss et al., 2011). They blame public health researchers for overemphasizing the evidence of socio-economic determinants and neglecting the susceptibility predisposed by genetic characteristics (Palotie, Widen, & Ripatti, 2013). Systematic reviews show that even the education and experience of health workers and physicians is not sufficient to ensure the quality of tests and precise interpretation of the test results (Goldsmith et al., 2013). This signifies the limitation of DTC in ensuring the validity and reliability of tests as well as precise interpretation of result.

Second dilemma is related to information communication. In the facility-based approach of health care delivery, the experts provide adequate information through counseling which enables people to make informed choices. Needy people can seek background information about the test, procedure of the test and they can make a plan for the next step after obtaining the result. This is practically impossible in DTC.

Advocates of the DTC genetic tests claim that the shift from classical physician-driven clinical setting to the consumer-driven market will empower people by providing the knowledge on their susceptibility towards particular diseases and their risk factors. It will help them make some decision for better health and happiness (Kaye, 2008). On the other side, genetic testing is a specialized domain of medicine and we can't make an assumption about the minimum educational qualification of people which enables them to take an informed decision. It is less

likely that people will be able to answer all the queries through internet and that the decision made will be conscious. Even if they made decision of ordering the test, they may have no idea of what they will do with the 'result data'. It may be inappropriate to expect the correct interpretation of test results by an ordinary consumer as correctly as the experts (Arnos, 2008).

Third ethical question represents the confidentiality of the genetic information. In the absence of regulation and legal framework to protect the privacy of consumer, there is always a risk of unauthorized use of information by the company or any third party (Tong, 2013). There are dilemmas about the ownership of genetic data i.e. is the information about genetic test the property of individuals or the company? People have suffered from various problems associated with violation of privacy. Some of the frequent examples include getting their claims rejected by the insurance companies, discrimination in employment and professional life (Geelen, Horstman, Marcelis, Doevendans, & Van Hoyweghen, 2012).

Some people might prefer DTC because they don't need to directly deal with the health workers and they might assume that this is more confidential. In this case it might serve the patients' privacy purpose but there is no evidence to say that DTC is more confidential than the classical physician prescribed test. There are also some fundamental questions. What will happen to the information if the company goes bankrupt or sold? Are the DTC data completely safe, private and confidential (Marietta & McGuire, 2009)?

Although genomics is a personalized medicine, it is not entirely personal. Results of DTC genetic testing of an individual may also be significant to his/her family members. Hiding or sharing the test result is an ethical concern. If DTC result is not communicated properly with the interpretation of the risk or safety, it may affect family communication, family response and

family relations too (Peterson, 2005; Tong, 2013). Within family there may be individuals who don't care or don't want to know the genetic predictions.

Fourth ethical concern is related with the price of the test and related services. Cost of a DTC genetic test ranges from 100 to several thousand USD depending on the test (Bloss et al., 2011). The market of DTC was assumed to be over 1 billion dollar per year in 2009 and expected to increase by 20-30 percent annually. It is thus clear that the cost of the service is a major challenge for countries to adopt DTC as a part of public health services. However, scientists say that the cost sequencing of the genome is dramatically decreasing over time and it will be a minute fraction of the health care expenditure in future (Palotie et al., 2013). It seems that test companies are the sole beneficiaries of profit generated from the test trade and tests are beyond the reach of ordinary people.

Fifth important ethical issue is consequence of the test and user safety. There is only limited application of the genetic test to address common health problems because the etiology originates from the complex interaction of genetic, social and environmental factors. Since the benefits of genetic tests are not clear and the cost is very high, the claim that genetic tests will improve the health of people doesn't seem credible. As a privately-offered service it instead has the prospect of masking the health inequalities together with social determinants of health. (Saukko, 2013).

Scientific reviews have shown that there is no sufficient evidence to say that the genetic profile will be helpful in detecting the risk factors of common diseases. The synthesis of studies shows that a company's claim that they are able to provide the nutrition and lifestyle modification recommendation on the basis of the algorithms of user's genetic profile is a hunch; they don't

have sufficient evidence to do or even to say that (Janssens et al., 2008). This means the claims made by the testing companies are not scientifically evident, are over-exaggerated and false to promote the sale of DTC genetic testing (Mutch, Wahli, & Williamson, 2005).

In 2008, a survey was conducted among 312 participants of Member of the National Society of Genetic Counselors, US. The findings revealed that about 90% of the participant genetic counselors believed that DTC possess additional risks of miscommunication of the risk and misinterpretation of the results because of the limited presence of the experts in the whole procedure of test initiation, testing and result interpretation (Hock et al., 2011). False result and inappropriate interpretation may lead to increased risk of stress or false sense of security to the consumers.

Should direct-to-consumer genetic testing be more regulated?

Sakkuko P. 2012 has compiled some facts about the marketing strategies adopted by major DTC genetic testing companies. Emphasizing the individual health right, specifically- right to know, the companies capitalize on the "anti-paternalistic sentiment". The companies throw the emotional assurance of being more empowered at their consumers. The companies are also providing false assurance in their websites like: providing nutrient supplements that could correct the genetic damage, linking the consumers to the experts for further discussion and additional interpretation, glamorizing the potential benefit of the genetic tests which are not evident (Saukko, 2013). This shows that there is a clear fraud being committed by the companies, they are stealing consumer's money and they are compromising their rights. This loophole in the regulation must be addressed by legal framework.

Different approaches are being tried for regulating the genetic testing around the world. In the U.S., genetic testing is under the lenses of Food and Drug Administration (FDA). In Germany and France, the presence of physician is mandatory while performing genetic test which means that DTC is not allowed there. On the other hand, DTC genetic testing is permitted in the UK and the debate about the classification of genetic testing technology/toolkits according to the anticipated risks/ harm is a topic of debate (Saukko, 2013).

Different theories state different points regarding regulations and state intervention in the area of DTC genetic testing. Liberal market theory says that DTC genetic testing should be allowed freely, without any restriction and regulation because they do not inflict any direct harm to the consumers(they are not drugs).Conservatives or consequentialists say that no test should be allowed without securing their utility and clinical support by the experts like physicans and counselors because there are harms associated with it, harms of psychological distress of false risk or false assurance of being out of the risk. Democratic pluralists say that the tests should be allowed to operate after ensuring the safety of the test and validity and reliability of the technology claimed by the companies (Wright, Hall, & Zimmern, 2011).

The legal regulation of the genetic research has been viewed from two perspectives- individual right, and research and development perspective. Research institutions and companies are saying that regulating research and development will halt the progress and invite unnecessary complications. They are of the opinion that regulating research is unjust for scientific development (Arnos, 2008). However, safety and protection of the rights of the consumer must be the top priority here.

Ethical dilemmas discussed above are valid and they should be addressed through sensible interventions. Neither liberalism nor extreme paternalism (prohibiting the people from taking the tests) can address those ethical dilemmas. Stewardship approach with intervention level, recommended by the Nuffield Council on Bioethics, 2007 may be appropriate to address both the liberal and paternalism side of state involvement in genetic testing (Krebs, 2008; Nuffield Council on Bioethics, 2007). State needs to ensure the environment where the test companies are socially responsible towards people's right by regulating the companies and DTC genetic test without violating people's right of deciding to access safe and scientifically sound DTC genetic tests (Calman, 2009).

Conclusions

Sequencing of DNA codes has been completed and a large benefit to public health is anticipated. Scientists say that even they don't know what they have invented and how big the future implications will be (Morris, 2013). Since there is no or very limited involvement of health professional in planning, performing and interpreting the result of the test, it will be difficult for ordinary people to make decision based on their test results. Even though it is voluntary, it is less likely to represent informed consent.

Genetic information obtained from the unregulated procedure, with less reliable technology and such information without precise interpretation can mislead the consumer by, for example, changing the dose of a drug without consultation. Since it is profit-driven and almost unregulated, there is a greater risk for consumers being robbed or harmed. Also it is hard to deny the possibility of overusing the test. As shown by the studies, validity, reliability and predictability of such tests is not satisfactory and the claims made by the companies on internet are not real. Inaction from the state and/or victim-blaming is not justifiable. Consumer choices

and preferences are influenced by availability of tests, how they are promoted by the companies, and how the price is valued and distributed(Nuffield Council on Bioethics, 2007). While DTC may show benefits in future, the risk of being harmed or violation of confidentiality is still high in the absence of regulations to make the companies legally obliged for the wellbeing of people. Therefore an immediate step from the nation states is needed to protect the people from being harmed, to protect privacy and confidentiality of genetic information and to ensure the quality and transference of DTC genetic tests.

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