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Direct-to-Consumer Genomics and Research Ethics: Should a More Robust Informed Consent Process Be Included?

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An important issue raised in the article by Lee and Crawley (2009) is whether direct-to-consumer (DTC) genomics, specifically personal genome testing (PGT) should be considered research. The authors state that DTC genetic companies are creating new models of research where social networking tools will allow consumers to group themselves around particular diseases or conditions and participate directly in biomedical research. One hope is that such networking will speed the acquisition of knowledge about genomics.

The United States federal regulations define research as including systematic investigation aimed at producing generalizeable knowledge (Emanuel 2003, 41). When scientists examine an individual's DNA for the purpose of PGT, it could be assumed that they do it in a systematic manner, given the descriptions to consumers and results provided by companies. Not to conduct the analysis in a systematic manner could undermine the aims and reputation of the companies. The companies claim to base their tests on current scientific literature and single nucleotide polymorphisms (SNPs), or common genetic variants, identified in the field. They investigate either a wide range of SNPs or specific mutations associated with a disease, such as BRCA1 and BRCA2. Good research and scientific practice involves clear aims, protocols and interpretable results that are reproducible. Ideally it is peer reviewed and open to scrutiny at different stages in the process. Whether the specific genetic tests are reliable and valid is less clear because there is no uniform or comprehensive system of regulatory oversight to assess the analytic and clinical validity of genetic tests before they are offered to the public (Javitt and Hudson 2006; Wasson et al. 2006; Wasson 2008). Many DTC genetic tests are considered 'home brew', or manufactured in one laboratory, and fall outside the regulatory authorization of the Centers for Medicare and Medicaid Services (CMS), which administers the Clinical Laboratory Improvement Act (CLIA) amended in 1988. The tests may represent the most current state of the science, while still lacking in validity or clinical utility.

The second requirement in the federal definition of research is to produce or contribute to generalizable knowledge. Various DTC genetic testing companies explicitly state they will conduct internal research on the DNA that consumers provide with the aim of contributing to the advancement of science and medicine (23andme.com 2009; Navigenics 2009). They also include sections that address collaborations with third parties to undertake research, noting the de-linked nature of the data potentially shared with both non-profit and commercial collaborators. The content of these studies, present or future, is unknown and would need to be assessed at the time they are proposed.

It can be argued that DTC PGT, as described by companies offering these services do meet the definition of research, in as much as we know about their processes and

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procedures from the outside. They indicate systematic investigation of the analysis of individual level DNA and aggregate DNA data as well as the explicitly stated intention of companies to conduct research that contributes to generalizeable knowledge. Whether that intention is brought to fruition will need to be examined further.

If DTC PGT is categorized as research, a subsequent question is whether the current informed consent process is sufficient. Informed consent includes the key components of disclosure, understanding and capacity (or legal competence), and voluntariness (Beauchamp and Childress 2001, 79–80). To satisfy the first, companies would need to disclose the relevant information needed for a consumer to be informed of the elements of the test, testing process and potential results and impact on the individual and/or others, particularly given the nature of genetic information. If these tests are considered to be research with human subjects, then IRB requirements for informed consent would be applicable. The companies would need to disclose the potential risks and benefits of the tests, side effects, and alternatives.

DTC genetic testing companies provide a good deal of information on their websites about the types of testing, nature of results and potential for positive or negative reactions to them. Do companies disclose sufficient information about potential results, risks and benefits and their significance? Risks include false positives and false negatives, receiving unexpected and/or distressing results, potential for breaches in privacy and confidentiality. Benefits include receiving accurate results about health status or particular conditions, being able to learn about one's individual genetic make-up and potentially take preventive measures. The precise information given varies from company to company as all have different versions of an informed consent.

Do individuals who have PGT understand that companies will conduct internal research with their material? Do they fully understand the significance of sharing their genetic material with third parties, even if de-linked from personal account information? This could include non-profit or commercial researchers, or potentially insurance companies or actuaries. We allow research on a variety of human biological samples, without re-consenting the individual donors as long as those samples are de-identified and delinked from the individual donor. This is much more challenging to ensure with DNA, as individuals can be traced with a portion of their DNA.

The second element of consent is less straightforward to assess in this context. It is not clear how companies assess the capacity of consumers. They may assume that someone who is able to navigate the website, locate the consent and sign it and order a test has capacity. This may be a reasonable assumption, although there may be instances when it is not accurate, for example, a high functioning mentally ill person or someone with early stage dementia. What consumers understand about the nature of the testing process and implications of the results is also unclear. The websites are often written at an education level higher than the recommended sixth to eighth grade for informed consent. Consumers reading this information may or may not be able to make sense of the genetic information and its significance before having a test. As is widely recognized in a medical context, a signature on a consent document does not necessarily indicate the level of understanding that a person has about a procedure or intervention in the medical setting. This may also be true for PGT, particularly given the complex nature of genetic information and lack of clinical utility of many of the identified SNPs. Whether an individual consumer sufficiently understands the implications of the results they will receive and how that could impact their life, positively or negatively, is difficult to know. Companies may offer links to health professionals or genetic counselors, though many suggest that if consumers have questions they should discuss them with their physicians, who may or may not be able to answer them.

Of the three key elements of consent, perhaps voluntariness is the least likely to cause concern. Consumers who are ordering the tests are most likely doing so of their own free will. There is a small chance that an individual would attempt to send in a sample that belongs to someone else, but that individual would have to get sufficient blood or saliva from him/her, and the websites articulate the legal consequences of those actions. Parents might choose to do this for their children. They would have the legal authority to do so, although there may be ethical discussion about whether it is a good idea.

The companies conducting DTC PGT recognize that they conduct research, internally and potentially externally with third parties. The criteria for informed consent is potentially being met at a minimal level—where consumers read information, sign a document—but could be strengthened, particularly given the complex nature of genetic results and information. This could be done through the company websites, via email, a set of questions consumers are required to answer before the kits are sent out, or by telephone. It would not necessarily be onerous and could have the added benefit of enhancing the informed consent process and the consumer's understanding of the significance of the testing and results.

If these DTC personal genome tests are considered research, given that they involve human subjects, should they be reviewed by an Institutional Review Board? Perhaps the companies view the individual PGT not as research, though they may conduct internal or collaborative research with the results of that individual test. If that is the case, then subsequent proposed research projects should be reviewed by an IRB to ensure human subjects are protected. Some companies note that future external research will be reviewed by an advisory committee, which presumably is selected and paid by the company, while others do not specify. One of the challenges here may be that without a system for oversight of genetic testing, it is difficult (if not impossible) to require companies to follow certain procedures and processes. Consumers can choose to order a PGT or not,

without reference to a health professional. How consumers of DTC PGT view the pros and cons of this type of testing and the results is only beginning to be explored. PGT offered DTC is an expanding area that seems likely to increase in the tests offered and potential impact on individuals, families, health care and society. Wider debate in the research, medical, scientific, and bioethical communities is needed to explore appropriate ways forward critically and carefully. ■

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