Individual DNA samples and health information sold by 23andMe

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23andMe, a direct-to-consumer personal genetic testing company, and Genentech, a leading biotechnology company, have entered into a \$60 million deal to perform whole-genome sequencing (WGS) on DNA samples collected by 23andMe from patients with Parkinson disease, according to a story by Matthew Herper¹ of *Forbes* magazine.

23andMe has provided single-nucleotide polymorphism (SNP) testing to more than 800,000 people, including about 12,000 with Parkinson disease. Customers must agree to 23andMe's terms of service, a long and detailed legal document incorporated into their website that states the genetic information and self-reported health information of customers may be shared with "research partners," including commercial partners. 23andMe's customers are offered the opportunity to take part in additional research in which their SNP microarray results are correlated with the results of voluntary surveys of health status and history. These genome-wide association studies, which involve anonymized, aggregated data, have produced a number of interesting observations that have been reported in peer-reviewed scientific and medical journals.^{2,3}

The new deal with Genentech is concerning because it involves something very different: sequencing and analysis of the entire genomes of 23andMe customers. This additional testing will be done on patient DNA samples stored by 23andMe after completion of their SNP testing. We have four major concerns about the implications of this business deal for the patients who will be tested. The first is whether individuals will be offered the option of receiving results and appropriate genetic counselling regarding high-penetrance, Mendelian, Parkinson disease-related variants discovered through sequencing their DNA. There is a growing consensus that subjects should have the option of receiving information about disease-causing mutations discovered through genetic research studies, especially when the results are actionable and have high clinical importance.^{4,5}

A second issue is related to informed consent for WGS, which has raised many ethical concerns in other contexts because of incidental findings, privacy, autonomy, and implications for other (untested) family members.^{6,7} 23andMe has stated that they will request specific consent to use individual, rather than

aggregate, data in this research, but details of this consent process for WGS are not available. This issue is important because it is unlikely that customers who signed up for SNP testing realize the implications of the WGS and analysis that will now be performed on their samples. In clinical practice, genetic counselling is required before WGS or whole-exome sequencing to make certain that families understand all of the possible ramifications of the testing. In a research setting, informed consent for WGS should include discussions of these same concerns as potential risks of the testing.

The issue of incidental findings deserves particular consideration. Research studies and clinical sequencing programs vary in their approaches to incidental findings, and details regarding how 23andMe will handle incidental findings have not been announced in the company's public statements. However, many individuals express a desire to receive all WGS results, especially those that predict the onset of serious diseases that may be preventable with appropriate medical or lifestyle interventions.

A third issue relates to privacy and the possibility of unauthorized re-identification of an individual from "de-identified" DNA sequence data.⁸ This may be of particular concern to individuals who are at risk for conditions that are associated with psychiatric disorders or dementia, such as Parkinson disease.

A fourth issue that is of concern to the larger scientific community as well as to patients and their advocacy groups is whether the genomic and phenotypic data collected from this project will be made available to other researchers and to assist in the clinical diagnosis of other patients. Open sharing of these valuable data in clinical databases such as ClinVar and LOVD is likely to increase greatly their scientific and clinical value. By contrast, holding these data in a closed proprietary database will limit the ability of other researchers and clinicians to use the information gained and may not be in the best interest of affected patients or their families.⁹ An international code of conduct has been proposed to facilitate genomic and clinical data sharing for biomedical research.¹⁰

While the business press may laud a lucrative deal to sell patient samples and clinical information for proprietary research, we—as genetic clinicians concerned about the appropriate use of

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powerful genomic technologies in patients—need to ask some serious questions. Do the patients and family members who are participating in 23andMe's research understand the extent of the information that these studies will provide that may not be returned to them? Do the subjects know that their DNA is being used to profit others and that the data obtained may not be openly shared within the scientific and medical community? Is this research being done with appropriate genetic counseling and the full informed consent of the subjects?

DISCLOSURE

The authors declare no conflict of interest.

REFERENCES

1. Herper M. Surprise! With \$60 million Genentech deal, 23andMe has a business plan. <http://www.forbes.com/sites/matthewherper/2015/01/06/surprise-with-60-million-genentech-deal-23andme-has-a-business-plan>. Accessed 25 February 2015.
2. Do CB, Tung JY, Dorfman E, et al. Web-based genome-wide association study identifies two novel loci and a substantial genetic component for Parkinson's disease. *PLoS Genet* 2011;7:e1002141.
3. Eriksson N, Benton GM, Do CB, et al. Genetic variants associated with breast size also influence breast cancer risk. *BMC Med Genet* 2012;13:53.
4. Jarvik GP, Amendola LM, Berg JS, et al.; eMERGE Act-ROR Committee and CERC Committee; CSER Act-ROR Working Group. Return of genomic results to research participants: the floor, the ceiling, and the choices in between. *Am J Hum Genet* 2014;94:818–826.
5. Wolf SM. Return of individual research results and incidental findings: facing the challenges of translational science. *Annu Rev Genomics Hum Genet* 2013;14:557–577.
6. Biesecker LG, Green RC. Diagnostic clinical genome and exome sequencing. *N Engl J Med* 2014;370:2418–2425.
7. Burke W, Antommaria AH, Bennett R, et al. Recommendations for returning genomic incidental findings? We need to talk! *Genet Med* 2013;15:854–859.
8. Gymrek M, McGuire AL, Golan D, Halperin E, Erlich Y. Identifying personal genomes by surname inference. *Science* 2013;339:321–324.
9. Ormond KE, Cho MK. Translating personalized medicine using new genetic technologies in clinical practice: the ethical issues. *Per Med* 2014;11:211–222.
10. Knoppers BM, Harris JR, Budin-Ljøsne I, Dove ES. A human rights approach to an international code of conduct for genomic and clinical data sharing. *Hum Genet* 2014;133:895–903.