Free the Data: The End of Genetic Data as Trade Secrets

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FOR PATIENTS AND THEIR CLINICIANS, broad access to information about DNA sequence variation is critical. The information is essential to interpreting the clinical significance of genetic variants, but it is not always readily available. At the present time, genetic test providers have the ability to create private databases containing information they have collected through testing and to restrict access to this vital information.

A case study of *BRCA1/2* testing by Myriad Genetics reveals the implications of creating a private database of clinical information and the importance of open access of this information. Until the June 2013 U.S. Supreme Court ruling that invalidated patents on isolated, naturally occurring human DNA, Myriad Genetics provided the only commercially available test for *BRCA1/2* diagnostic testing in countries where patents were filed (for example, the United States). Myriad has tested over 1 million people and has gained valuable experience and information.

A particularly significant consequence of being the sole provider of the *BRCA1/2* diagnostic tests is that Myriad has collected a vast number of the genetic variants associated with the *BRCA1* and *BRCA2* genes, along with the accompanying interpretation of each variant. If this testing becomes available elsewhere, this information will not be available to researchers, clinicians, and patients.

Some *BRCA1/2* test results have a clear-cut interpretation. Perhaps the most clear-cut is a negative result upon testing a relative of someone with a known mutation. A negative result could be uncertain unless the negative result occurs in a person with a known mutation in the family and that mutation was not detected. A positive result, or detection of a known deleterious mutation, is also clear-cut when that mutation has been disclosed. However, some variants are particularly difficult to interpret because the effect of the variation is not yet known. These *BRCA* variations are aptly named “variants of unknown significance” (VUS).

Myriad’s patent exclusivity on the *BRCA1/2* genes allowed the company to be the sole distributors of the genetic test, and thus it has been the largest collector of any clinically discovered VUS. Through the information gained from this collection, Myriad reports it was able to reduce the frequency of reporting a VUS. Furthermore, when a

patient receives a test result with a VUS, Myriad offers free testing to the family members in an effort to gain more information and help determine the clinical significance of the variant. The ability to include additional data on the VUS and expand the proprietary database is a clear incentive for Myriad to offer free testing to the family members of the patient with the VUS. Patients also benefit from this practice.

However, other providers of the *BRCA1/2* tests will face unnecessary challenges due to the lack of transparency of data. The practice of privatizing clinical data obstructs the scientific community from independently verifying the data’s clinical significance and accuracy.

The ideal course of action to mitigate this problem would be to require every laboratory that tests for *BRCA1/2* to register each test in the National Center for Biotechnology Information (NCBI), National Institutes of Health, Genetic Testing Registry, and deposit all *BRCA1/2* variants in the NCBI ClinVar database.

Another powerful way of liberating the *BRCA1/2* variants is for the very people who have been tested to “free the data” and independently submit their *BRCA1/2* report to directly contribute to an open-access database. Current projects that have undertaken this effort include the Sharing Clinical Reports Project (www.sharingclinicalreports.org), where physicians are encouraged to submit their patients’ de-identified report to be uploaded to the ClinVar database, and the complementary Free the Data! Campaign (www.free-the-data.org), where the individuals are empowered to share the information themselves. The Sharing Clinical Reports Project avoids violating the Health Insurance Portability and Accountability Act by the de-identification of the patient’s personal information, but the Free the Data! Campaign goes one step further in individual autonomy by enabling tested individuals to share their variant in ClinVar and to set their own sharing, privacy, and data access preferences. They can also share phenotypic information, thus providing researchers with information to help advance an understanding of these variants.

BRCA1/2 is just an example of the clinical trade secret issue. Allowing public access to variants and associated phenotypes on all genes is essential for the accelerated

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understanding of health and disease that everyone desires. Private databases thwart progress and slow understanding of the contribution of genetics to health and disease. Laboratories need to share clinical variants for the public good; it cannot be held as a proprietary interest. Individuals need to exert their influence and “free the data” to let the data speak.

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