Addressing a patient-controlled approach for genomic data sharing

To the Editor: We appreciate the recent statement in Genetics in Medicine "Laboratory and Clinical Genomic Data Sharing Is Crucial to Improving Genetic Health Care: A Position Statement of the American College of Medical Genetics and Genomics". We support the position of ACMG and strongly agree that sharing of genomic information is becoming increasingly important for the care of individual patients and understanding of disease pathways. In response to this statement, we want to elaborate on a specific mechanism for which data sharing can be considered: a patient-controlled approach. In addition, we want to discuss several challenges that must be addressed to achieve patient sharing of genomic data.

The patient-controlled approach has shown its promise although more research and discussion is needed. A recent study, and several others, revealed that research participants strongly support obtaining their genetic test results, expressing a desire of ownership of their genetic information, mostly for ease in sharing with health providers or family members, as well as a strong belief in patient empowerment. Currently, there are several initiatives (e.g., GenomeConnect, My Research Legacy by the American Heart Association, and numerous National Institutes of Health registries) that invite patients to share biomedical and genetic data for research and health purposes. Additionally, the National Institutes of Health and Office of the National Coordinator for Health Information Technology recently launched Sync 4 Science, a pilot program that aims to give patients an easy way to share their health data with researchers in support of the goal of the All of Us Research Program. Consistent with the US Department of Health and Human Services’ vision in health information technology, there is an increased emphasis on empowering patients to control and share their healthcare data with providers and/or researchers.

There are several reasons that patients may wish to access and share their genetic information, many of which were defined recently in a personal communication when the American Civil Liberties Union filed a complaint pursuant to the Health Insurance Portability and Accountability Act with the US Department of Health and Human Services. Specifically, four patients were denied their requests for all of their genetic sequencing information from a well-known supplier of genetic testing. Rehm, in her letter of support, defends the right of patients to access their genomic data for several incentives, one of which is that patients may wish to advance research via data sharing of their own. This American Civil Liberties Union complaint highlights the current lack of guidelines and standards regarding patients’ access and rights to their own genetic data and also highlights the desire of individuals to understand their healthcare data and become more active partners in their health.

Genetic information about an individual inherently poses risk about privacy and confidentiality. For example, the potential predictive power of certain genetic variants in a person poses a confidentiality risk from unwanted use or disclosure of health status. To prevent discrimination on the basis of this type of genetic information, US Congress passed the Genetic Information Nondiscrimination Act of 2008. Additional efforts, as we advocate here, towards development of a patient-controlled approach for genomic data sharing would enable individuals to set control over their own sharing, privacy, and consent preferences. If patients could control data sharing, many of the ethical and regulatory issues concerning sharing of genomic data could be directly addressed or alleviated.

Regardless of patient- or institute-controlled approaches, the sheer volume and complexity of genomic data can also be a barrier to sharing. The majority of Clinical Laboratory Improvement Amendments–certified laboratories (e.g., Baylor Miraca Genetics Laboratories) providing clinical genome sequencing services and even companies that offer direct-to-consumer genetic testing (e.g., 23andMe and Genos) do provide raw sequence data upon request in the form of .fastq, .bam, and/or .vcf files via either secure online transfer or portable hard drives. However, many labs restrict raw data requests to only the ordering clinician and not patients, making it nearly impossible for a patient to access and share their own genetic data either with other healthcare providers at unaffiliated institutions or into public databases. Besides, transfer of large files online may be technologically challenging for some patients, while shipping of hard drives can be burdensome and costly (approximately 150–200 USD) to a patient. It is imperative that we change the system to technologically accommodate efficient and secure storage and transport of genomic data. We support the Office of the National Coordinator roadmap that encourages those involved in health information technology to contribute to development of a defined, shared roadmap leveraging health information technology interoperability to ultimately protect and advance healthcare for all.

There is a massive potential in research and clinical settings to leverage genomic data to advance human health overall. This potential, however, comes with many challenges. Efforts towards developing a patient-controlled approach for sharing of personal genome data will undoubtedly contribute to research and clinical initiatives. Ultimately, this type of approach will benefit the sake of patient engagement and will
promote the following: (i) data sharing across health organizations for clinical care purposes and/or (ii) contribution of data to public databases for research purposes.

DISCLOSURE
The authors declare no conflict of interest.

Katherine E. Miller, PhD and Simon M. Lin, MD, MBA
Research Information Solutions and Innovation, The Research Institute at Nationwide Children’s Hospital, Columbus, Ohio, USA. Correspondence: Simon M. Lin (Simon.Lin@nationwidechildrens.org)

REFERENCES