Global Alliance for Genomics & Health
Collaborate. Innovate. Accelerate.

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U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES
OFFICE FOR CIVIL RIGHTS
200 Independence Avenue, SW, Suite 515F, HHH Building
Washington, DC 20201

To whom it may concern,

We are writing in support of the administrative complaint submitted by patients seeking access to their *BRCA1* and *BRCA2* genetic information. As leaders of the Global Alliance for Genomics and Health (GA4GH), we believe that variant aggregation and patient access to genomic and clinical data are critical steps in the effort to improve human health through genomic medicine.

The GA4GH consists of more than 400 organizational members and more than 600 individual members from around the globe who are working together to establish a common framework of harmonized approaches to enable effective and responsible sharing of genomic and clinical data. We work to illustrate the value of data sharing in real world contexts by catalyzing demonstration projects such as the BRCA Challenge, whose specific aim is to advance understanding of the genetic basis of breast and other cancers by pooling data on *BRCA1/2* genetic variants from around the world. Improved understanding of genetic variation in these genes has the potential to improve patient diagnoses and prevention of disease.

Founded in part on the principle of respect for the data sharing and privacy preferences of participants, GA4GH takes a humans rights approach to the issue of patient engagement: as stated in our *Framework for Responsible Sharing of Genomic and Health Related Data*, Article 27 of the 1948 *Universal Declaration of Human Rights* guarantees the rights of every individual in the world "to share in scientific advancement and its benefits" (including to freely engage in responsible scientific inquiry). For this reason, we believe that patients have a human right to access their own data in order to contribute to any research that would benefit from those data.

As such, we are committed to ensuring that patient access to genomic and health related data align with the goals of the BRCA Challenge. Patients who obtain their data from Myriad can immediately support research by sharing their information directly with the BRCA Exchange, the web portal built by the BRCA Challenge. Several other private labs that offer BRCA testing are already reflexively sharing variant-level data with public databases such as ClinVar, consistent with patient consent. The BRCA Challenge has aggregated those data, and is also currently collaborating with Ambry, Invitae, Counsyl,

GeneDx, Color Genomics, and others, to pilot the aggregation of some case-level data in a secure computing environment for improved pathogenicity classification. Data from Myriad remain a gaping void in the world's knowledge about *BRCA*-related cancers.

For precision medicine to realize its promise of transforming human health, researchers and clinicians must have access to large-scale datasets on the order of many millions of genomic and clinical cases. Millions of data points are needed to achieve statistically robust evidence linking genomic information with phenotypes, diagnoses, and treatment responses. Furthermore, data sets must contain information across all levels of pathogenicity, from benign to deleterious in order to reveal differences in health and disease. No single institution is capable of compiling such volumes on its own, so data aggregation is imperative.

All variant calls and raw sequencing files are used in assessing a patient's hereditary risk for BRCA-related cancers, so these data should be considered part of a patient's Designated Record Set (DRS). Access to these data is critical for the global community to learn from clinical genomic research and to achieve better health outcomes as a result. For this reason, the GA4GH recommends that OCR clarify that the DRS explicitly includes variant calls and raw sequencing files, giving patients the right under HIPAA to routinely access them and thus contribute to research and improved health.

Best regards,

Thomas Hudson
Chair, Steering Committee
Global Alliance for Genomics and Health

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