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Lack of Diversity in Genomics Research

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Precision medicine is a movement that seeks to serve a patient's individual medical needs with a tailored level of treatment. Such a practice requires an essential framework of genetic information to allow doctors to make accurate treatment decisions. Genome-wide association studies (GWAS) provide massive amounts of information for databases, such as the GWAS Catalog; however, there is an apparent lack of diversity in the genomics data available to researchers and drug-developers. The majority of participants in GWAS are of European descent. As of January 2019, 78 percent of genetics contributed by GWAS have been of European descent; while, only 22 percent are minorities (Yeager 2019). Stemming from a history of discrimination and unfair logistics, the lack of diversity in genomic databases undercuts the accessibility and function of Precision Medicine, and this issue needs to be resolved.

After the completion of the human genome project in 2003, the ability to conduct GWAS and create a database of genetic information became possible and affordable. Over the past two decades, the data on human genetics has grown. Based on this growth, one would assume our knowledge of treatments based on the unique genetic makeup patients would be vast and precise by now. At times, however, the proclamations of Precision Medicine overlook the history of genetics— the percentage of European genetics in the GWAS Catalog remains the majority, even if they are not the majority by global population (Popejoy 2016). To understand how the lack of diversity in genetic databases developed, one must look at the questionable ethical history, medical assumptions, and objective logistics of genetic studies.

Genetic research has had its share of ethically turbulent moments. As discussed by journalist Chou, the conversation between the scientific community and the public often struggles to distinguish between race and ethnicity of participants in genome studies (Chou 2017). The social construction of race is false, based on external phenotypes like skin color, and not backed by science because significant genetic variation exists even within the historical "five races"—African, Asian, European, Native American, and Oceanian— that have been used as a standard of categorization (Chou 2017). The genetic isolation of populations within the human species results in countless variations in phenotype and contributes to general ethnic differences and tendencies, which can greatly impact a patient's health and lifestyle from a medical perspective. Thus, using ethnicity to minimize variables when categorizing data experimentally can be useful, but the variation within ethnicity remains and makes constructing GWAS difficult (Chou 2017).

Another ethical factor, besides the issue of distinguishing race from ethnicity when gathering patient samples, is the blatant harm genetics research has inflicted on uninformed populations. One of the most striking examples concerns the Havasupai tribe in Arizona (Santos 2008). The Havasupai tribe agreed to a genetic study proposed by Arizona State University in the 1990s and were under the impression that the researchers would report the results and provide insight into the high frequency of type-2 diabetes in the tribe (Santos 2008). However, the Havasupai never received a report from the study; in fact, the cell lines produced from the tribe's samples were distributed among other universities, where studies deviating from the original agreement were conducted without the Havasupai's consent (Santos 2008). Consequently, the Havasupai filed lawsuits on the grounds of "exploitation and violation of civil rights" (Santos 2008). Numerous incidents similar to this have historically occurred in minority populations; a well-known example being the exploitation of cells from Henrietta Lacks, also known as HeLa cells. With such an unethical history of interaction with research endeavors, minorities commonly refuse to participate in genetic research studies, which contributes to the lack of diversity in GWAS (Santos 2008).

Along with ethical issues, genetics research has also been conducted with certain assumptions in mind that simply do not hold true. As Adebowale Adeyemo, deputy director of the Center for Research on Genomics and Global Health at the National Human Genome Research Institute, highlights in an interview, there have been assumptions that destitute populations and minority populations do not need genomic testing because infectious illnesses are a more prevalent and pressing concern than genetic diseases in such populations. However, over the years it has been proven that genetic diseases, such as heart disease and other chronic diseases, are also a significant concern in poor populations and minority populations just as in affluent or majority populations (Mullin 2016).

Moreover, the genomes of many minorities and indigenous populations are more difficult to decode because of the numerous diverse communities within each minority and indigenous population. This complexity appears to deter researchers because of the assumptions that the complexity will not provide accurate data to produce treatments (Santos 2008). For those few who do take on the challenge, the difficulty comes with great reward in discovering mutations and variants that often prove useful for medicine (Santos 2008). The unique qualities of minority and indigenous genomes provide greater knowledge on how genetics influence drug interactions during treatment and other health issues like diabetes (Popejoy 2016). Therefore, concerning medical care, minority patients require just as much genetic data as the European patients, if not more because of the great diversity within the genomes of minority and indigenous populations that can provide great benefit to treatment and drug development.

While ethics and assumptions show how the research community has been wrong in its execution of genetic studies, some aspects of the logistics of GWAS can hinder the ability to achieve diversity in studies. The primary logistical issue connects to historical points already mentioned. If people do not wish to contribute their genetic information due to historical oppression, discrimination, and unethical practice under false assumptions, then it proves extremely difficult to gather data on such populations today. The inability to attain samples from minorities slows progress towards diversifying the GWAS Catalog and other genetic databases, despite efforts by programs like All Of Us, which was created by the NIH in 2018 to develop a database of at least a million diverse patient genomes (Yeager 2019).

Due to how long the databases have been filled with European majority, another logistical issue stems from the analytical models used to process the data produced by these massive GWAS. Analytical methods called ancestry metrics are used to process genomic data found in GWAS databases, but these metrics do not properly analyze an individual of a diverse background (Bustamante 2011). As a result, our current methodology of analysis likely produces a significant amount of error for genomes that carry more variants and deviate from the more homogenous populations that the metrics are based upon (Bustamante 2011).

The history of genetics research is riddled with ethical and logistical issues that have contributed to the present lack of diversity in GWAS. Incidents such as sampling bias based on supposed "race" and violation of minority patients' consent rights have caused logical issues for present day GWAS that seek to gather diverse genomes but cannot due to a lack of trust in research. Thankfully, several programs, similar to All Of Us, have been commissioned in recent years to create a better relationship between the public and researchers. While the past cannot be changed, the way in which future GWAS are constructed and conducted can be adjusted to achieve a genetic database that properly represents and provides Precision Medicine for the entire human species.

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