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Genomics and Health Equity Research: Current Challenges and Opportunities

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Health disparities are caused by several factors including genetics. African Americans and Hispanic populations have a higher rate of all major chronic health conditions, exemplified by a disproportionate burden of COVID-19 disease and death among minorities. With the mapping of the human genome in the early 2000s, and speed sequencing technology, the amount of new genetic information has significantly increased over the last 15 years. Today, genetic information is routinely used for diagnosis, prognosis, and treatment. However, the lack of large-scale genetic sequencing in diverse populations results in health disparities. Approximately 80% of the genetic information used in GWAS studies originate from people of European ancestry, and significantly less from other diverse populations, with only 2% originating from the African ancestry population. Review of clinically relevant sites ascertained from annotations in ClinVar revealed that approximately 50% of observations comes from people of European ancestry. Data from the UK Biobank showed that for many traits the accuracy of the Polygenic Risk Scores (PRS) declined when applying to diverse populations with people of African ancestry having only 25% accuracy compared to people of European ancestry. Therefore, increasing diversity in genomic databases is critical to ensure that one does not exacerbate health disparities by generating an inaccurate PRS. Moreover, ancestral genetic variants influence drug pharmacokinetics and substance use. Thus, to fill the gaps in genomic diversity we need to build biobanks and perform large-scale sequencing of all diverse human populations. A more inclusive genomics research will lead to equitable and precision health care.