Western bias in human genetic studies is 'both scientifically damaging and unfair'

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Sheko men taking part in a lactose tolerance test during a study by the Tishkoff Lab in Ethiopia. Credit: Tishkoff Lab

Despite efforts to include more diversity in research, people of European ancestry continue to be vastly overrepresented and ethnically diverse populations largely excluded from human genomics research, according to the authors of a commentary published March 21 in a special issue of the journal *Cell* on human genetics. This lack of diversity in studies has serious consequences for science and medicine.

For one thing, they say, the bias in the data limits scientists' understanding of the genetic and environmental factors influencing health and disease. It also limits the ability to make accurate predictions of a person's disease risk based on genetics and to develop new and potentially more effective treatment approaches.

"Leaving entire populations out of human genetic studies is both scientifically damaging and unfair," says co-author Sarah Tishkoff (@SarahTishkoff), a human evolutionary geneticist at the University of Pennsylvania. "We may be missing genetic variants that play an important role in health and disease across ethnically diverse populations, which may have deleterious consequences in terms of disease prevention and treatment."

Tishkoff and her colleagues, including Giorgio Sirugo, University of Pennsylvania, and Scott M. Williams, Case Western Reserve University School of Medicine, report that, as of 2018, individuals included in genome-wide association studies (GWAS) were 78% European, 10% Asian, 2% African, 1% Hispanic, and