Lack of racial diversity in DNA databases spurs errors identifying disease-causing genes

The Exome Aggregation Consortium, or ExAC, is a simple idea. It combines sequences for the protein-coding region of the genome…allowing scientists to compare them and understand how variable they are. But the resource is having tremendous impacts in biomedical research.

Many disease-association studies…have identified mutations as pathogenic…but it's possible that they weren't looking hard enough, or in the right populations.

In August this year, [geneticist Daniel] MacArthur's group published its analysis of ExAC data in Nature, revealing that many mutations thought to be harmful are probably not…

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[Hugh] Watkins, [geneticist at the University of Oxford, UK,] checked the ExAC database for information on genes that have been associated with these heart conditions…about 60 genes had been implicated as harboring pathogenic mutations…[but his] analysis revealed that 40 of these probably bear no link.

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Exac is quietly becoming a standard tool in medical genetics.

ExAC has also driven home a point that…other researchers have made repeatedly: that failing to include people from Asian, African, Latino and other non-European ancestries is holding back understanding of how genes influence disease by limiting the view of human genetic diversity.

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion and analysis. Read full, original post: Failing to include people from Asian, African, Latino and other non-European ancestries in DNA databases is causing errors in identification of disease genes