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Researchers turn to policy to tackle health disparities in an age of personalized medicine

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A new paper from researchers from Tufts University and colleagues addresses how increased support for minority-focused research, community-based participatory research, and studies of gene-environment interactions may improve science's understanding of chronic diseases across races and ethnicities. The paper, published today in *Health Affairs*, outlines policy efforts needed to ensure the advancement of genetic applications in healthcare in ways that reduce existing disparities.

Focusing on breast cancer and chronic kidney disease, the authors show that while genetic and molecular knowledge has grown and been used to fight these diseases in the last decade, significant racial and ethnic health disparities persist and hinder universal progress. The policy recommendations they propose are:

- Increasing the enrollment of non-white participants in research on complex diseases.
- Community-based participatory research to promote genetic literacy and encourage more volunteers to become involved in research.
- Funding research on the interactions of genes and the environment.
- Educating healthcare providers as well as patients about the risks, benefits and limitations of genetic research.

"We need to collect more data from groups for whom we currently have

insufficient information so that we can improve care for all individuals. If we don't expand our efforts, the quality and effectiveness of genetic research and services will be limited in ways that can perpetuate health disparities. The Precision Medicine Initiative for one will be a big step forward in this endeavor and is to be commended for including community-based health provider organizations in its network to attract volunteers. Increasing diversity in research and testing will help maximize the possibilities of precision medicine," said senior author José M. Ordovás, Ph.D., director of the Nutrition and Genomics Laboratory at the Jean Mayer USDA Human Nutrition Research Center on Aging at Tufts University.

The authors identify three key areas where their recommendations would make improvements: incomplete genetic databases, inadequate treatment options, and insufficiently understood disease mechanisms.

People with non-European ancestries are underrepresented in genetic databases, limiting the ability to apply genetic knowledge to reduce disease in these groups. While researchers know that hereditary breast cancer is linked to mutations in the BRCA1 or BRCA2 genes, the "normal" genetic sequence for these genes was determined based on women of European and Ashkenazi Jewish descent. Studies show that African-American and Hispanic women are much less likely than white women to receive genetic counseling or testing for hereditary breast cancer; the absence of this data perpetuates an incomplete genetic database on which clinical decisions about treating breast cancer rely.

For many diseases, clinical advances in treatment have developed based on new knowledge of genetic markers. Studies show that non-Hispanic black women tend to be diagnosed with more advanced sporadic breast cancer (occurring without family history) compared to white women. Many potential genetic markers might explain the racial and ethnic disparity in tumor aggressiveness in sporadic cancer. However, the small

number of tumor samples from non-European women offers inadequate details about patient and tumor characteristics and restricts the use of genetic knowledge in clinical treatments for all individuals.

Similarly, treatments tailored to individuals with high-risk genotypes for certain diseases have not yet been identified because the molecular mechanisms behind the diseases are unknown as well. Researchers know that African-Americans are disproportionately affected by variants in the APOL1 gene which can increase a person's risk of kidney disease by up to seven times. Due to insufficient understanding of the molecular mechanisms by which the gene increases disease risk, effective treatments elude clinicians.

"Ultimately, we want the knowledge gained from a reduction in health disparities to lead to an increase in treatments for people who are most at risk. If we understand the aggressive breast cancer subtype that more frequently affects black women, we might be able to expand treatment options. We want to look at environmental factors as well as tumor biology to know how they contribute to the disease, and how we might then attack it," said first author Caren E. Smith, D.V.M., a scientist in the Nutrition and Genomics Laboratory at the Jean Mayer USDA Human Nutrition Research Center on Aging at Tufts University.

More information: Smith, C.E., Fullerton S.M., Dookeran K.A., Hampel H., Tin A., Maruthur N.M., Schisler J.C., Henderson .JA., Tucker K.L., and Ordovas J.M., "Using genetic technologies to reduce, rather than widen, health disparities," *Health Affairs*. 2016; 35(8):1367-1373. DOI: 10.1377/hlthaff.2015.1476

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