2024 HDBuzz Prize: Thinking beyond therapies - it’s time to consider racial disparity in HD care and research

We're proud to announce Dr. Zanna Voysey as a 2024 HDBuzz Prize winner! She shares a new study that details racial disparities in HD care and research. Addressing this could improve the lives of people living with HD in the here-and-now.

By Dr Zanna Voysey | October 28, 2024 | Edited by Dr Sarah Hernandez

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Huntington’s disease (HD) is a progressive brain disease that typically starts to show symptoms between the ages of 30 to 50, when people are in the prime of life. It’s also heritable, meaning anyone who has a parent with HD has a 50% chance of getting it. HD has historically been thought of as more common among those of White ancestry, but new data challenges this, suggesting comparable rates in Black individuals. Racial and ethnic health inequalities are well documented in North America, with Black and Latino individuals found to be less likely to receive neurological care, even when socioeconomic and insurance payer factors are controlled for. How, then, does this affect HD gene carriers?

Delayed diagnosis for Black people living with HD in North America

Until now, questions around racial health disparities in HD have received little attention, but a new study from Adys Mendizabal and colleagues from UCLA has begun to address this.

Adys and her team explored racial disparities in HD care by looking at data from over 4,000 North American HD gene carriers in the ENROLL-HD database.

A new study suggests that Black people living with Huntington's disease are diagnosed on average one year later than White people living with HD.

ENROLL-HD is the world's largest observational study for HD families, where people living with HD from all over the world are asked questions by their neurologist during clinic visits with the option to donate blood samples. No treatments are tested - ENROLL-HD purely aims to give researchers a better understanding of HD. It's a collaboration between HD families, clinicians, and researchers.

Adys and her team used the ENROLL-HD database to see how much time there was between when a gene carrier's family first noted symptoms, to when a diagnosis of HD was made. They found that, on average, the time to receiving a diagnosis was one year longer for Black compared to White individuals in North America. This fits with findings from other studies, showing that Black individuals are typically at a more advanced stage of HD when they enter ENROLL-HD compared to White individuals.

A likely underestimate

What's more, the realities are likely worse. In the study, almost 90% of participants were White, while 3.4% were Latino, and 2.3% were Black. These proportions don't marry up with what clinicians typically report seeing in their clinics, suggesting racial and ethnic minority groups are less frequently recruited to ENROLL-HD.

This highlights a second issue facing HD gene carriers in North America: that of under-representation in research studies. Indeed, the fact that differences weren't seen for other racial and ethnic groups in the study is likely because there were too few such participants in the study at all.

It's also important to consider that ENROLL-HD participants as a whole are a somewhat skewed reflection of the HD community. This means the figures from the UCLA study are likely an underestimate, and the real-world racial and ethnic disparities in HD care are probably even greater. This is because ENROLL-HD only takes place at HD Centers of Excellence, in urban and academic-affiliated settings.

> **"They found that, on average, the time to receiving a diagnosis was one year longer for Black compared to White individuals in North America. "**

HD Centers of Excellence provide multidisciplinary care for HD families, with neurologists, psychiatrists, social workers, therapists, counselors, and other professionals who have experience working with HD families. Centers of Excellence are, in part, supported by the Huntington's Disease Society of America.

Factors at play

It’s important to note that Mendizabal and colleagues’ study wasn’t designed to drill down on exactly what is driving this delay in diagnosis, so currently this is speculative.

The researchers couldn’t tell, for example, if it was because Black HD gene carriers delay going to doctors, or have greater delays in accessing specialist care. However, they point out evidence from other disease groups, where Black individuals have been found to be more frequently misdiagnosed, and less frequently referred for genetic testing.

Likewise, they highlight the possibility of the interaction with other sociodemographic factors, for example employment status, but had too few participants in their study to be able to explore this meaningfully. They also cite studies raising concerns of racial disparities in the stigmatization of genetic conditions.

From an ENROLL-HD recruitment perspective, they raise the possibility of unequal access to HD Centers of Excellence, a potential lack of culturally similar research staff, and data from other diseases suggesting that racial and ethnic minority groups are less likely to be invited to take part in research.

Racial and ethnic minority groups are likely under-represented in the ENROLL-HD clinical database, meaning racial disparities in Huntington’s disease are probably underestimated.

Importantly, they also point to decades of unethical experimentation in Black, Latino, and Native American communities in the United States, which has promoted community mistrust of research.

Pointers for the research community

One of the key takeaways from this study is that we need more research to really uncover the drivers behind these differences, so we can figure out how best to address them.

This is particularly important since advanced imaging scans are now being brought into HD diagnosis and stage classification – which has the potential to widen the gap for underserved populations.

In the meantime though, modifications to ENROLL-HD could help address this area by:

1. Expanding racial and ethnic grouping options, to more accurately capture diversity among participants. For example, multiracial or multiethnic individuals currently have no categorisation option besides ‘Other’.
2. Adopting a standardized recruitment approach that promotes more balanced racial and ethnic representation within the study.
3. Increasing the numbers of ENROLL-HD sites in under-represented areas, such as Latin America, Australasia, Asia, and Africa.

> **“In the absence of a treatment breakthrough, findings like these remind us to think outside the box and remember that there are other ways that we can seek to improve the lives of those living with HD in the here-and-now. ”**

A silver lining

While the results of this study raise concern, we can also see them as a positive. In the absence of a treatment breakthrough, findings like these remind us to think outside the box and remember that there are other ways that we can seek to improve the lives of those living with HD in the here-and-now.

This study also shows the value of the time, energy, and effort that HD families put into participating in ENROLL-HD. This study is just one example of the many that use the data collected from ENROLL-HD to advance our understanding of HD, and hopefully get us to a treatment sooner.

To date, ENROLL-HD has 21,669 participants from 155 clinical sites across 23 countries. This is fantastic participation from the patient community! But this study reminds us that it’s vital that everyone from the HD community is represented, including historically marginalized communities. If you would like to learn more about ENROLL-HD or join the study, you can do so here.

The authors have no conflicts of interest to declare. For more information about our disclosure policy see our FAQ...

GLOSSARY

observational A study in which measurements are made in human volunteers but no experimental drug or treatment is given