Giving Thanks to the Huntington's Disease Family Community for Advancing Research

At this time of giving, we want to give thanks to you, the Huntington's disease (HD) community, for all you have done for HD research, bringing us closer to medicines which will one day slow or halt HD.

By HDBuzz Editors | December 03, 2024 | Edited by HDBuzz Editors

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In the spirit of gratitude, this Giving Tuesday, the HDBuzz editorial team would like to reflect on the unique and transformative role the Huntington's disease (HD) family community plays in advancing scientific research. From their resilience in the face of this extremely challenging condition, to their extraordinary contributions to scientific studies, the HD family community continues to be an essential partner in driving progress toward treatments and, one day soon, a treatmentcure.

Participation in Research: A Commitment to Progress

One of the most inspiring aspects of the HD family community is their unwavering commitment to participating in research. Clinical trials, observational studies, and other data collection efforts all rely heavily on the active involvement of individuals with HD, their families, and at-risk individuals. These studies demand time, effort, and emotional resilience, yet the HD community consistently rises to the occasion, understanding that their participation is key to scientific progress.

The Huntington's disease family community has forged successful partnerships with researchers, clinicians, policy makers and other stakeholders, pushing research forward.

This has been a long-standing theme in HD research. The Venezuela project began in 1979, a critical project in the history of HD research, enabled by the contributions of families from communities in this region. Their participation laid the groundwork for identifying the genetic mutation that causes HD, revolutionising the field and enabling genetic testing and targeted research.

Selfless contributions

Programs like Enroll-HD, a global observational study, owe much of their success to the dedication of HD families. More than 350 projects interrogating Enroll-HD data have been conducted, and more than 150 peer-reviewed manuscripts detailing their finds have been published - a huge achievement!

Additionally, genome-wide association studies (also called GWAS) have been made possible through the donation of DNA samples from HD families to a variety of different registries. GWAS look to try and find genetic modifiers of disease; other genetic factors which track with symptoms starting earlier or later in life than might be predicted based on CAG number alone. Many of these newly identified modifiers from GWAS of people with HD are now top priority targets for drug discovery by academic and industry researchers alike.

Biomarker identification and tracking studies, such as HD-Clarity and iNFLuence-HD, rely on the collection of cerebrospinal fluid (CSF) samples and blood samples to identify measurable indicators of disease progression. Similarly, many stem cell and brain cell studies, which transform donated skin cell samples into disease models, have been a direct result of the generosity of HD families.

More and more HD research studies are using donated human brain samples. This tracks with the advent of many fancy new techniques that allow researchers to examine post-mortem brains at a cell-by-cell level, increasing the amount of information gathered from these precious samples by orders of magnitude. The findings that come from those studies get us closer to understanding HD in people, and closer to a treatment.

Sharing Personal Stories: Humanising the Science

The HD family community has been instrumental in humanising the science behind the disease. By sharing personal stories through blogs, conference presentations, and other outlets, they provide researchers, policymakers, and the general public with a window into the real-life impact of HD. These narratives inspire scientists to work harder, inform the design of patient-centred clinical trials, and help ensure that new therapies address the true needs of those affected.

Projects like My HD Story and POWER-HD further enrich the research landscape by collecting HD family member-reported outcomes, details of peoples real-world lived experiences, and other vital data. Moreover, these stories foster empathy and understanding, breaking down barriers of stigma and isolation. They serve as a reminder that behind every dataset and lab experiment are real people - families who are fighting to ensure a better future for their loved ones and others.

The Power of Advocacy and Awareness

Making our voices heard is critical! Change makers need to listen to the real-world experiences of people living with HD and of those participating in clinical trials.

The HD community has been a relentless force in advocating for increased funding, research opportunities, more social supports, and greater public awareness. Many of the local HD organisations around the world are driven by families and caregivers, and have worked tirelessly to bring HD into the spotlight. These efforts have not only fostered public understanding but have also paved the way for governments and private entities to prioritise HD research initiatives. Without their advocacy, many breakthroughs in funding and resources would not have been possible.

Notably, HD advocates have brought their voices to regulatory bodies like theFDA, ensuring that the patient perspective is central in evaluating new therapies. This advocacy has been crucial in shaping guidelines for drug development and approval processes, making them more responsive to the needs of the HD community.

A Call for Continued Partnership

As we give thanks for the HD family community, it is also a call to action for researchers, policymakers, and society at large to continue prioritising their voices and contributions. Building on this partnership means:

- Designing studies that respect and prioritise the needs of participants.
- Ensuring that research findings are communicated back to the community in a timely and accessible manner.
- Advocating for continued investment in HD research and community resources.

Looking Ahead with Gratitude

Progress in HD research would not be possible without the courage, generosity, and perseverance of the HD family community. They are the heart and soul of the fight against Huntington's disease, and their contributions illuminate the path toward a future free of this devastating condition.

As we reflect on all that has been achieved, let us reaffirm our gratitude to this remarkable community and recommit to working together toward our shared vision of hope, healing, and discovery. So this Giving Tuesday, we encourage everyone from the HD community, particularly those from HD families, to look inward, acknowledge your contributions, and give yourself some gratitude.

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

GLOSSARY

CSF A clear fluid produced by the brain, which surrounds and supports the brain and spinal cord.

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

biomarker a test of any kind - including blood tests, thinking tests and brain scans - that can measure or predict the progression of a disease like HD. Biomarkers may make clinical trials of new drugs quicker and more reliable.

genome the name given to all the genes that contain the complete instructions for making a person or other organism

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