Navigating the Complex Maze of Huntington’s Disease Research
HDSA Research in 2022: Innovation and Community

Message from Arik Johnson, PsyD
HDSA Chief Mission Officer

Research is a cornerstone of the Huntington’s Disease Society of America’s mission. In our commitment to improving the lives of all people affected by Huntington’s disease, HDSA both invests in science by directly funding research in the lab and connecting HD families to participate in the clinical research necessary to develop new potential treatments. This year’s issue of The Marker is filled with reports showcasing HDSA funded scientists and collective efforts to support the development of new therapeutics.

The world of HD science, especially as of late, is filled with ups and downs. However, for every clinical trial that ends, there are new trials beginning and even more happening in pre-clinical research. Those “failures” actually provide data that is incorporated into the broader HD science landscape and inspires further innovation. This leads to better understanding HD processes and unlocks opportunities for potential treatments to keep the field of HD science moving forward.

Research is also a fundamental element in all HDSA Mission programs and services. Our Educational Program features scientific updates at both local and national events, like the HDSA Annual Convention. HDSA Social Workers and Support Group Leaders regularly field questions from the community about studies and trials and connect them to HD TrialFinder.org. Our Disability Program and Advocacy efforts are guided by all that has been discovered and informed by the work happening today to increase access to care.

Research is truly the key to solving the mysteries of HD and unlocking methods of treating symptoms and the disease itself. Your ongoing support of HDSA’s research programs and the larger mission makes this important work possible. Thank you for your continued partnership in the fight against HD.

Please enjoy this edition of The Marker and cheers to the year ahead.

Message from Leora Fox, PhD
HDSA Assistant Director, Research & Patient Engagement

As the year draws to a close, I reflect upon the many opportunities HDSA had in 2022 to support fantastic Huntington’s disease science, share research news with the community, and amplify family voices in drug development. 2022 brought a diverse and expanded portfolio of scientific approaches, rooted in a commitment to human-centric discovery and career development in HD research. In this year’s edition of The Marker, we highlight current fellows’ exciting findings, as well as past award recipients who have developed into independent investigators.

Our direct research funding programs support passionate people advancing HD science in ways that will directly impact clinical trials, and who are becoming mentors of the next generation of HD researchers. The breadth of HDSA-supported work continues to expand, from innovative molecular applications, to understanding racial disparities in HD diagnosis, to rigorous clinical study of the benefits of dance, to improvements in palliative care. Our wide community reach, strong partnerships with industry, and the influence of HD family voices in drug development, are reflected in data surrounding the use of HDSA research resources like HD TrialFinder.

As in-person educational events and forums for scientific innovation began to resume in 2022, HDSA continued to be a trusted source of reliable information about HD science, information about opportunities to participate in research, hope in new directions, and support in the face of clinical setbacks.

I believe strongly that community engagement with research is a form of advocacy, and an understanding of progress should be accessible to all. Thank you for your support of HD research and care, and for reading The Marker.

A Focus on Human Biology and the HD Clinic

Although animal models have been instrumental to our understanding of HD biology, the most relevant scientific observations are those that are made in humans and human tissues. For this reason, HDSA has adopted a patient-centric research strategy, with the HD Human Biology Project as its cornerstone. Clinical Research Pilot Grants are also available to medical professionals associated with HDSA Centers of Excellence. To date, HDSA has committed more than $6.25 million dollars to support these programs.
Following review of applications by HDSA’s expert Scientific Advisory Board, six scientists were awarded the 2022 HDSA Human Biology Fellowship. These awards are open to people at any career stage, location, or institution, and provide up to $150,000 to support a two-year human-centric research project, from small human trials to the study of human cells in a dish. This year’s cohort includes researchers from Hungary, Spain, the United Kingdom, and the United States, and spans a diverse scientific range. We look forward to hearing about their progress in the months and years to come.

### Ines Bras, PhD
Postdoctoral Scholar
University of Central Florida

**Non-Invasive HD Biomarkers**

Dr. Bras has shown that huntingtin protein can be found in extracellular vesicles, small sacs that are shed from brain cells. Over the course of this project, she will work with mentor Dr. Amber Southwell to refine techniques for detecting these vesicles in blood donated by people with HD. This project could produce a new and less invasive biomarker — a potential way to track the effectiveness of huntingtin-lowering therapies over the duration of a clinical trial.

### Lea Danics, PhD
Postdoctoral Scientist
Semmelweis University (Hungary)

**Identifying Novel Therapeutic Targets in HD**

Under the mentorship of Dr. Karolina Pircs, Dr. Danics’ project will utilize a model system in which human skin cells donated by people with HD can be directly converted into a dish into brain cells, retaining properties of the donors in a way that better reflects the adult onset of HD. This cutting-edge system will enable Dr. Danics to investigate the biological pathways that control the breakdown of toxic huntingtin, which could be applied to drug development.

### Paul Dennis, MFA
Professor and Chair, Dance Department
Hunter College, CUNY

**Dance for Huntington’s Disease**

While regular exercise is often recommended for people with HD, there are no rigorous clinical studies examining the use of dance as an intervention to improve daily functioning and quality of life. Mr. Dennis has partnered with a team of HD-experienced physical therapy and neurology specialists for a clinical pilot to generate data supporting the safety and benefits of a non-pharmacological intervention for Huntington’s disease.

### Maria Rosario Fernandez-Fernandez, PhD
Senior Scientist
FINBA-ISPA (Spain)

**Restoring Protein-Building Machinery**

This work takes a new approach to exploring and correcting brain cell dysfunction in HD, aimed at the machinery that builds proteins from RNA recipes. Dr. Fernandez-Fernandez has found that this machinery is disorganized and tends to stall in HD. Her goal is to test existing chemical compounds with “stalling relief” properties in human cells, to restore their ability to produce proteins efficiently.

### Kilian Hett, PhD
Postdoctoral Research Fellow
Vanderbilt University

**Studying the Flow of Cerebrospinal Fluid**

Dr. Hett, an expert in neuroimaging analysis, will work under the direction of Dr. Daniel Claasen, Director of the HDSA Center of Excellence at Vanderbilt. He will use non-invasive MRI imaging to investigate circulation of the fluid that bathes the nervous system, known as cerebrospinal fluid (CSF). Understanding how CSF flow changes in HD has implications for improved spinal delivery of huntingtin-lowering drugs.

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**Supporting Career Development in HD**

Funding opportunities can be limited for young scientists wishing to study a rare disorder like Huntington’s disease, some of whom are themselves facing life with HD. HDSA is committed to career development in the HD field by introducing young minds to HD research, and supporting early post-doctoral scientists with a desire to make HD part of their career plan.

HDSA’s vision of ensuring that the pipeline of passionate HD scientists and clinicians remains full is shared by the Berman and Topper families. With generous commitments exceeding $2,640,000, HDSA has to date awarded 10 Berman-Topper HD Career Development Fellowships since the program’s inception in 2016. These extraordinary young scientists have exceeded expectations, self-organizing to form a supportive network where they collaborate and troubleshoot their work.

A highly competitive application process led to the selection of Dr. Sara Sameni of California’s Salk Institute for Biological Studies as the 2022 recipient of the Berman-Topper Fellowship. She has unique expertise in biomedical engineering and mathematics. Under the mentorship of Dr. Terry Sejnowski, she is applying novel computational techniques to HD cells to create personalized models that can predict disease course and treatment response in people with HD.

In her very first quarterly report, Sarah simulated what happens to important signaling molecules when cells get sick over the course of HD, and showed how some of these changes might be corrected with an existing drug.
2022 Donald A. King Summer Research Fellowships

In 2005, HDSA launched the Donald A. King Summer Research Fellowship with the goal of attracting bright young scientists to HD research. The program was established to honor Don King, a tireless advocate for HD families who served as HDSA’s Chairman of the Board from 1999 to 2003 before his sudden passing in 2004. In 2022, HDSA’s SAB selected five researchers to receive this fellowship, jumpstarting their research endeavors through the study of HD biology and pathology.

Anthony Ventimiglia
Ohio State University
Anthony was mentored by Dr. Richard Fishel, and studied protein complex MLH1-PMS1, which plays a role in repair of CAG repeat expansions in Huntington’s disease.

Katherine Vinski
University of Pittsburgh
Katherine studied how crosstalk between two protein pathways affects stress response in HD and how they might be regulated, under the mentorship of Dr. Diane Carlisle.

Carly Fisher
Ohio State University
Carly investigated the role of a DNA repair protein in regulating CAG repeat expansion in Huntington’s disease, under the mentorship of Dr. Richard Fishel.

Manasa Chillarige
University of Pittsburgh
Manasa worked with mentor Dr. Diane Carlisle to study whether activation of a specific protein pathway can protect brain cells from the negative effects of stress brought on by HD.

Carolina Gomez Casas
UMass Medical School
Carolina worked with Dr. Michael Brodsky to determine how a CRISPR-Cas9 system may be used to reduce CAG expansions in the mutated HD gene.

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In February, HDSA was proud to announce that fifty-five outstanding Huntington’s disease care facilities were awarded the designation of HDSA Centers of Excellence for 2022. In addition to the 55 grant funded clinics, ten regional partner sites were also named, ensuring expert HD care at 65 distinct medical facilities in 35 states across the nation. This year, $1,896,333 was awarded to the HDSA Centers of Excellence program.

The HDSA Centers of Excellence are multi-disciplinary teams with expertise in Huntington’s disease that share an exemplary commitment to providing comprehensive care for this complex, hereditary, neurodegenerative disease. They are also selected based on their ability to provide HD families with opportunities and referrals to participate in cutting-edge clinical research, including observational studies and interventional drug trials.

More information regarding HDSA Centers of Excellence and contact information can be found at HDSA.org/coe.
The 2022 HDSA Centers of Excellence Grantees

Albany Medical College (NY)  
Barrow Neurological Institute (AZ)  
Beth Israel Deaconess Medical Center (MA)  
Cleveland Clinic (OH)  
Columbia Health Sciences/NYS Psychiatric Institute (NY)  
Duke University (NC)  
Emory University (GA)  
MedStar Georgetown University Hospital (DC)  
Hennepin Health Care (MN)  
Henry Ford Hospital (MI)  
Indiana University  
Johns Hopkins University (MD)  
Massachusetts General Hospital Medical University of South Carolina  
Northwestern University (IL)  
Ochsner Health System (LA)  
Ohio State University  
Oregon Health & Science University*  
Rocky Mountain Movement Disorders Clinic (CO)  
Rowan University School of Medicine & Rutgers University RWJ Medical School (NJ)  
Rush University Medical Center (IL)  
University of Arkansas  
University at Buffalo (NY)  
University of California, Davis Medical Center  
• Partner Site: Kaiser Permanente (CA)  
University of California, Irvine  
University of California, Los Angeles  
University of California, San Diego  
University of California, San Francisco  
University of Cincinnati (OH)  
University of Colorado  
University of Florida  
University of Iowa  
University of Kansas Medical Center  
University of Louisville (KY)  
University of Miami (FL)  
University of Mississippi Medical Center  
University of Nebraska Medical Center  
University of Pennsylvania  
• Partner Site: St. Luke’s University Health Network, Bethlehem, PA*  
University of Pittsburgh Medical Center (PA)  
University of Rochester (NY)  
University of South Carolina School of Medicine  
University of South Florida  
• Partner Site: Central Florida Center for Huntington’s Disease, Winter Park and Orlando, FL*  
University of Texas Health Science Center at Houston, McGovern Medical School  
• Partner Site: Covenant Medical Group Neurology, Lubbock, TX  
• Partner Site: Texas Movement Disorders Specialist, PLLC  
• Partner Site: University of Texas Health San Antonio  
University of Utah  
University of Vermont, Frederick Binter Center for Parkinson’s Disease and Movement Disorders  
University of Virginia  
University of Washington (WA)  
• Partner Site: Selkirk Neurology, Spokane, WA*  
University of Wisconsin  
Vanderbilt University Medical Center (TN)  
• Partner Site: Cale Neuroscience Center, University of Tennessee Medical Center  
• Partner Site: University of Tennessee, Erlanger Medical Center  
Virginia Commonwealth University  
Washington University School of Medicine (MO)  

* Blue text indicates newly designated HDSA Center of Excellence or Partner Sites for 2022.
Keeping On the Pulse of HD Research

HDSA is committed to involving the HD community in research, by keeping abreast of scientific advancements, ensuring that news is conveyed with accuracy and clarity, and providing information about research opportunities. We strive to meet these goals by attending conferences and communicating closely with industry partners. We then help families to navigate scientific and clinical twists and turns by creating spaces to share news, and producing understandable research content.

**HD Research Conferences in 2022**

As in-person gatherings resumed throughout 2022, HD researchers from academia and industry came together at field-wide meetings to share findings and discuss progress. HDSA staff were present to hear research updates and strengthen HDSA’s partnerships at a variety of scientific conferences.

**FEBRUARY**
17th Annual HD Therapeutics Conference: A Forum for Drug Discovery & Development
Palm Springs, CA
Hosted by CHDI Foundation, industry and academic scientists met to discuss their findings

**JUNE**
HDSA’s 37th Annual Convention
Atlanta, GA
A variety of research programming was designed by HDSA for families, along with showcases of companies recruiting clinical studies

**AUGUST**
HDSA2022, the Hereditary Disease Foundation’s (HDF) 12th Milton Wexler Biennial Symposium
Cambridge, MA
This conference is devoted to academic and clinical research on HD and related rare diseases.

The editorial board of HDBuzz.

**SEPTEMBER**
European Huntington’s Disease Network Plenary Meeting
Bologna, Italy
This biannual plenary meeting highlights clinical progress from academic and industry researchers.

**NOVEMBER**
29th Annual Meeting of the Huntington Study Group
Tampa, FL
The yearly HSG meeting focuses on clinical progress and is directed at medical professionals and families.

**Hot Topics in HD Research**

**New Mouse Model Aids Researchers**

In early 2022, researchers at UCLA published a new mouse model for HD, called **BAC-CAG**, that mimics CAG-repeat expansion in humans more closely than previous mouse models of the disease. It could help us better understand how instability in the CAG repeat region of the huntingtin gene sets the course of HD in humans.

**HD Integrated Staging System — A New Classification System for HD**

In mid-2022, a worldwide team of HD researchers published a classification system for Huntington’s Disease known as the HD Integrated Staging System (HD-ISS). Among its many applications is to provide a framework for running drug trials in presymptomatic individuals who carry the HD gene.

Dr. Sarah Tabrizi presented the HD-ISS at HDSA’s Convention in June.

**Advancements In Zooming In On Single Cells**

2022 brought technological advances allowing researchers to zoom in on single cells and examine changes in hundreds of genetic messages and proteins simultaneously. This is allowing scientists like Harvard’s Dr. Steve McCarroll to investigate why certain brain cells are most vulnerable in HD.

**Enroll-HD Celebrates Ten Years**

This year brought the tenth anniversary of **Enroll-HD**, a worldwide observational study that follows people in HD families over time. This study and its thousands of participants continue to be a driving force for pharmaceutical intervention and improving our understanding of HD biology.

Photo by Matt Teuten, courtesy of the Hereditary Disease Foundation
2022 Developments In Clinical Research

There are currently dozens of companies working in the HD research space, developing drugs to treat HD from many different scientific angles. Numerous players are in the early phases of HD drug development; others are further along in their clinical investigations of treatments for HD. Here we provide some highlights to help you navigate the news around their progress in 2022.

**Annexon Biosciences** completed in 2022 a successful safety trial of ANX-005, a drug aimed at preserving the connections between brain cells. They announced in August that they are planning for a larger Phase 2 trial in people with Huntington’s disease.

**AskBio** announced in late August 2022 that they were approved to begin a Phase 1/2 trial for a novel HD gene therapy, BV-101. The trial will take place in France, and the drug aims to improve cholesterol metabolism in order to preserve the function of brain cells and lower huntingtin protein.

**LoQus23 Therapeutics** is focused on developing oral drugs to stop the expansion of CAG repeats. In May 2022 they announced an international collaboration to develop tools and therapies to monitor and slow the rate at which the repeat grows.

**Neurocrine Biosciences** reported successful control of HD chorea in the Phase 3 KINECT-HD trial of valenazaine. This is a once-daily pill to help with Huntington’s disease movement symptoms, and they are now applying to regulatory agencies for approval.

**Novartis** tested an oral huntingtin-lowering therapy, branaplam, which was originally developed for a different childhood disorder. The Phase 2 VIBRANT-HD trial was suspended in August 2022 due to concerns about peripheral neuropathy, and unfortunately, development of branaplam ended in December. Despite this disappointing conclusion, Novartis remains committed to learning as much as possible about the effects of branaplam and making that knowledge available to the HD community for the benefit of future drug development.

**Prilenia Therapeutics** is running the PROOF-HD Trial of pridopidine, a drug aimed at protecting brain cells to slow the progression of HD. They fully recruited this 600-person Phase 3 study in 2022, and will share the results in the second half of 2023.

**PTC Therapeutics** is also developing an oral huntingtin-lowering therapy, PTC-518, which was designed for HD. Their 1-year Phase 2 trial is called PIVOT-HD and began recruiting in mid-2022, with an expansion of eligibility criteria in November. It is currently moving forward in Australia and Europe in people with pre-motor and motor symptoms. PTC is seeking approval from the FDA to continue the trial as planned in the US.

**Roche** announced in November 2022 that a new trial of the spinally delivered huntingtin-lowering drug tominersen will begin recruiting early next year. After the Phase 3 GENERATION-HD1 trial was halted in 2021 due to concerns about long-term safety, Roche presented data during 2022 suggesting that younger people who began the trial with less severe symptoms may have benefited from tominersen. For this reason, they took a step back and will proceed with a Phase 2 trial, GENERATION-HD2. They also confirmed their commitment to HD by announcing that they have two additional huntingtin-lowering drugs in their pipeline, one spinal and one oral.

**Sage Therapeutics** began running the DIMENSION and SURVEYOR trials in people with HD to test SAGE-718, an oral drug aimed at preserving thinking abilities and day to day function. These trials began in mid-2022 and are recruiting participants at sites across the USA.

**Spark Therapeutics** is working on an innovative approach that aims to turn the brain’s support cells into new neurons. They have partnered with NeuExcell to develop a gene therapy for HD.

**uniQure** is conducting a small clinical trial (Phase 1/2) of the first gene therapy for HD (AMT-130), delivered to the brain via surgery. Promising safety and huntingtin-lowering was reported in June 2022 for participants who received a low dose; a pause was initiated in the high dose group in August, but surgeries resumed in November with new safety measures in place. More results are expected in mid-2023.

**Vaccinex** published a paper detailing the results of the SIGNAL trial of pepinemab for HD, which concluded in 2020. Although the study did not meet its primary endpoints, some data suggested potential cognitive benefits of the drug.

**Wave Life Sciences** is also testing a spinally delivered huntingtin-lowering therapy, WVE-003, in a Phase 1/2 trial in people with HD in Canada and Europe. Their focus is the harmful form of huntingtin, and they have a redesigned chemistry that will hopefully allow Wave to overcome the roadblocks they faced in 2021, when the PRECISION-HD trials were halted due to lack of huntingtin-lowering.
Supporting HD Research Participation and Advocacy

HDSA’s research communications, resources for participation in research, and global research advocacy programs seek to ensure that families can find hope in progress, make informed choices about participation, and have a voice in the drug development process. Our goal is to guide families through the HD media maze and provide accurate and up-to-date information.

HDSA Research Communications

- **Research Webinars**
  - HDSA Research Webinars give academic and industry scientists a direct forum to share their latest findings with families.

- **HDBuzz**
  - HDBuzz supports HDBuzz, a website devoted to clear and effective communication about HD research and clinical studies.

- **HDSA Online**
  - Research news and press releases are vetted and posted on HDSA’s website news feed and social media channels.

- **This Week in HD Research**
  - Kelly Andrew and Dr. Leora Fox provide a roundup of HD research activities in HDSA’s blog, This Week in HD Research.

HDSA-Vetted Survey Studies

Researchers, healthcare professionals, and students studying HD frequently need input from the HD community on different topics relevant to HD families. More than a dozen HDSA-vetted surveys were featured on our website in 2022, with opportunities for everyone in the community.

**HDSA’s HD Trialfinder**

HDSA’s HD Trialfinder (www.hdtrialfinder.org) is a website, call center, and clinical trials matching service created by HDSA in 2015 to help families learn about and participate in HD clinical trials. HD Trialfinder is powered and programmed by Carebox, which provides anonymous user data to HDSA. HDSA was selected by the Huntington Study Group to present a poster at their November 2022 conference on community use of HD Trialfinder. Below are the key findings from this analysis.

Usage of HD Trialfinder is even across HDSA’s main regional development areas, highlighting the reach of our resources and interest in clinical trials across the USA. Users are mainly creating profiles for themselves or a family member, and are most commonly seeking and viewing information about clinical studies. This finding can help HDSA to strengthen the messaging to clinical researchers that it is valuable to present opportunities in understandable language.

Site views and profiles on HD Trialfinder peaked in response to major clinical trial news, speaking to HDSA’s wide reach and strong partnerships with industry.

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**Huntington’s Disease Coalition for Patient Engagement (HD-COPE)**

Now expanding and in its fifth year, HD-COPE continues to provide family input to help industry researchers better understand HD and to guide their clinical plans. The worldwide group of volunteer advocates met virtually and in-person during 2022 with several pharmaceutical and biotech companies.

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**Unique Web Sessions Over Time**

![Unique Web Sessions Over Time Graph](image)
HDSA Award-Winning Scientists Continue Ground-Breaking Work

Former HDSA Human Biology Fellows Continue to Innovate
Several scientists published academic papers in 2022 detailing their findings supported by the Human Biology Fellowship.

Dr. Edith Pfister
2018 Human Biology Project Fellow, Dr. Edith Pfister, works on chemical engineering of genetic therapies for brain diseases. A recent publication in Nature Communications explored a novel siRNA drug design and showed that it could silence the harmful huntingtin message in a mouse model of HD.

Dr. Veronica Brito
2016 Human Biology Project award recipient, Dr. Veronica Brito, examines changes in RNA metabolism in HD and how they may damage brain tissue over the course of the disease. A study published in Cellular and Molecular Life Sciences this year looked at how this phenomenon affected learning and memory in HD mice, which could be a new treatment target for HD.

Dr. Ali Khoshnan
2017 Human Biology Project Fellow, Dr. Ali Khoshnan, works to understand how changes in gut microbiota affect HD symptoms and onset in fruit flies. A study recently published in Frontiers in Neuroscience shows that when HD fruit flies are exposed to certain bacteria, they experience more rapid onset of HD-like symptoms.

Dr. Amber Southwell
2015 Human Biology Fellow, Dr. Amber Southwell, became a professor during her fellowship and continues to run an independent HD laboratory at the University of Central Florida, where she has trained several Donald King Summer Research Fellows and is now mentoring 2022 Human Biology Project Fellow, Dr. Ines Bras.

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HDSA-Supported Scientists Blossom
HDSA’s Research Fellowships have supported awardees through important career transitions; these HD-committed scientists have gone on to start laboratories of their own and are training the next generation of HD researchers.

At the conclusion of her HDSA award period, 2019 Berman-Topper Fellow Dr. Lauren Byrne was named the recipient of a prestigious Medical Research award in the UK. Lauren’s HDSA fellowship supported her transition to an independent research lab at University College London, and her new award will devote unprecedented funding to the study of Juvenile HD.

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We are grateful to the HDSA Scientific Advisory Board (SAB) members who generously volunteer their time and talent to ensure that HDSA’s research programs are scientifically sound and maximally impactful for the HD community. The HDSA SAB is comprised of leading researchers and clinicians in HD and related fields, as well as HD community members with a passion for research. Additionally, the SAB advises HDSA management and its Board of Trustees on a range of issues influencing the scientific direction of the Society.
Huntington’s disease (HD) is a fatal genetic disorder that causes the progressive breakdown of nerve cells in the brain. It deteriorates a person’s physical and mental abilities often starting in their prime working years. Currently, there is no cure for Huntington’s disease.

HD is known as the quintessential family disease, because every child of a parent with HD has a 50/50 chance of carrying the faulty gene that causes Huntington’s disease. Today, there are approximately 41,000 symptomatic Americans and more than 200,000 individuals at-risk of inheriting the disease.

The Huntington’s Disease Society of America (HDSA) is the premier nonprofit organization dedicated to improving the lives of everyone affected by HD. From community services and education to advocacy and research, HDSA is the world’s leader in providing help for today and hope for tomorrow for people with HD and their families.

Across the United States HDSA supports 50 volunteer-led Chapters & Affiliates, 55 HDSA Centers of Excellence, more than 60 social workers and 80 support groups specifically for HD families.