Hopes and Dreams
Huntington's Disease Society of America - Illinois Chapter Quarterly Newsletter

President's Message

Dear HD Families,

I hope all of you are having a great 2022. As I'm sure you would agree, it is nice to move into a level of normalcy following a challenging 2021 due to the pandemic.

The Illinois board is committed to supporting the HD community and making 2022 a very successful year. We have numerous activities planned for 2022. We are very excited to celebrate the 18th Annual Team Hope Walk at the Naperville Riverwalk on May 15th. I want to thank Larry Haigh and Karen Bennett for their leadership in organizing this fantastic event that Dave and Susie Hodgson started. I look forward to seeing many of you at this HDSA IL hallmark event. We have other great events planned for the summer and early fall. In addition, planning is in place for the Team Hope Galesburg Walk on 9/11 organized by Sarah Cozad and always a ton-of-fun Baggo tournament on 8/6, organized by Charlotte Rybarczyk. Thank you, Sarah, and Charlotte, for leading these great fundraisers. We are also in the planning stages of a Food Truck fundraising event in the city. This should be a lot of fun. Stay tuned for more details. Finally, our rock star social worker, Emily Zivin, is planning an Education/Wellness Day in September. More information will be forthcoming in the next month.

I'm also delighted to see the 37th Annual HDSA Convention is in full force this year in Atlanta, GA, from 6/9 – 6/11. I am very much looking forward to attending the convention this year. I attended the convention every year from 2007 to 2017 and learned so much about HD and was truly inspired by all the HD families I interacted with at these past conventions. I also have the good fortune to attend Leadership Day on 6/4. I look forward to the opportunity and interacting and learning from other HD chapter leaders.

The HD Illinois Chapter members will continue to support and serve the HD community to the best of their abilities in 2022. Don't hesitate to get in touch with members of the board or me if you have ideas or require any support.

I look forward to seeing you at many great 2022 summer events!

Arvind Sreedharan
President, HDSA Illinois Chapter

Huntington's Disease Society of America
Illinois CHAPTER

Family is everything

TEAM HOPE

TIME TO REGISTER

National Team Hope Sponsors

Genentech
A Member of the Roche Group

teva

Sunday • May 15, 2022

Naperville Riverwalk Grand Pavilion

912 Honorary Sindt Memorial Ct., Naperville, IL

Register your team today! Start your fundraising page!
https://www.hdsa.org/thwnaperville

Annual Huntington's Disease TEAM HOPE - Walk For the CURE

You can help the Huntington's Disease Society of American find hope for HD families, and provide help to the 41,000 Americans with HD and the 200,000 who are at risk.

Register online at the URL below or the QR code to the left! **REGISTER BY APRIL 5TH** to guarantee your t-shirt. Registration to walk is $30 for adults and $20 for children 12 and under. Early registration is encouraged. During registration you can join a team or create a team of your own 'participant page' and begin fundraising! We hope to see you there!

TEAM HOPE WALK

REGISTER YOUR TEAM TODAY!
https:/www.hdsa.org/thwnaperville

WALK DAY SCHEDULE

9:30am Registration
10:30am Walk

Sunday • May 15, 2022

HOT DOG LUNCH, LIVE DJ AND FAMILY FUN!

Danny Bega MD, MSCI
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Division of Movement Disorders
Northwestern HD Clinic
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When to Consider a Feeding Tube in Huntington's disease Care

By Dr. Danielle Larson

For individuals with Huntington's disease (HD) and their loved ones, the decision to use a feeding tube can seem overwhelming and complicated, and therefore the decision can be difficult to make. The goal of this article is to explain why individuals with HD may need a feeding tube, discuss how it can help with symptoms, and review how a feeding tube is placed and used.

There are two main reasons why individuals with HD may need a feeding tube: 1) weight loss, and 2) difficulty swallowing. HD can cause weight loss because individuals with HD need more calories, which can be due to having a lot of extra movements (chorea) and/or a higher metabolism. When someone with HD cannot eat as many calories as their body needs, they lose weight. HD can cause difficulty swallowing, which makes eating more calories difficult. Additionally, when swallowing food or water is difficult and causes coughing or choking, small particles of food or water can be inhaled into the lungs and cause an infection, called Aspiration Pneumonia. Aspiration Pneumonia can be dangerous and require treatment in the hospital.

By placing a feeding tube in the stomach, individuals are given nutrition through the tube, called "tube feeds." These extra calories help prevent weight loss. When swallowing is too difficult and there is a high risk for Aspiration Pneumonia, the decision can be made to use only the tube for feeding, water, and medications, and to not eat, drink or take any medications by mouth. In this way, the feeding tube helps to keep the individual with HD safe.

A feeding tube, technically called a gastrostomy tube, is placed in the stomach by a straight-forward and safe procedure. The procedure is done by someone with experience in gastrostomy tube placement, and typically patients can go home the same day of the procedure. During the procedure, a gastrostomy tube is placed through the abdominal wall to form a connection to the stomach. The medical team in charge of placing the gastrostomy tube teaches the patient and their care partners how to use and care for the tube day to day.

It is a good idea to discuss one's feelings about having a feeding tube placed for nutrition support or safety early on in one's disease course. Discussing this decision with loved ones early ensures that the individual with HD can fully understand the reasons a feeding tube might be necessary and communicate their wishes regarding this option with their loved ones.

Huntington's Disease Parity Act

By Emily Zivin, Illinois Chapter Social Worker

HDSA is still advocating for the HDSA Parity Act, which will waive Medicare's two-year waiting period for people with Huntington's disease, and we need everyone to lobby our senators and congressional staff to become co-sponsors. It is time to take action because two years is too long! If people need help with language for their emails to our senators and congressional staff, feel free to reach out to Emily Zivin, HDSA IL Chapter Social Worker directly at EZivin@hdsa.org.

Illinois Chapter Home News...The News-Gazette: Diagnosed with a terminal illness, Mahomet woman opts to 'go big'

By Dave Hinton

MAHOMET — If former novelist Nicole Wellman included all the twists and turns of her life in one of her books, readers might roll their eyes, thinking it's all too much.

- Former single mother with six children, including triplets.
- Supported her large brood by herself for many years by writing numerous books and working in public relations.
- Lost more than 100 pounds through fitness training and long-distance running.
- Has been diagnosed with a terminal illness.
- Will launch a State Farm insurance agency in Mahomet beginning next month.

"It may seem an odd time to launch a new business, but I'm going to go big or go home for as long as I can," the 50-year-old Wellman said.
Now married to Jim, whom she wed "a couple of years ago," they have 10 children between them.

The terminal-illness diagnosis came two years ago. Nicole Wellman discovered in her family genes Huntington's Disease — a neurodegenerative disease whose symptoms are those of ALS, Parkinson's and Alzheimer's. She was tested for the disease, and the prognosis was not good.

"It's a beast of a diagnosis," Wellman said. None of the symptoms have yet developed. But she knows they will.

"I have had no problems at all," she said. "I can't imagine where I'd be if I hadn't made those changes. The diet and exercise is a goal of staving off the symptoms" for as long as possible.

Wellman is determined to help as many people who have the disease as she can, including running in marathons and other long-distance races to raise money for the Huntington's Disease Society of America. Six years ago, she ran her first 5K race. She has also done some triathlons and a half-ironman, which includes a 1.2-mile swim, 56-mile bike ride and a half-marathon. She will run in the Chicago Marathon again this year.

Wellman is one determined person. She wrote 26 young-adult fiction and Christian-based parenting books but had to leave that behind. The publishing industry changed due to the advent of e-books and self-publishing. Wellman then did marketing work for a dental organization that served 1,600 dental offices.

From there, State Farm recruited her to become an agent. "Coming new into insurance and launching a business, it's a lot to learn," Wellman said. "But State Farm has been incredible with all the support. This is their 100-year anniversary, so this is a fun year to jump on board." The new office is due to open April 1 at 1204 E. Oak St., Suite 4, on Mahomet's east side.

Join us on June 9-11th for the 37th Annual HDSA Convention at the Marriott Marquis in Atlanta, Georgia! Following COVID-19 safety recommendations, we will be capping the number of in-person attendees and providing mandatory safety protocols. Can't make it to Atlanta? No Problem! Every session throughout the HDSA Convention will be livestreamed – FOR FREE!

Register at: http://hdsa.org/about-hdsa/annual-convention/

News from Northwestern Medicine
HDSA Center of Excellence

Northwestern Medicine HDSA Center of Excellence
Virtual Patient and Family Education Series 2022

Saturday, April 9th at 10 am via zoom
Asymptomatic Gene-Positive
Please join Seth Rotberg as he shares his HD journey. This will be an interactive session for individuals who are asymptomatic gene-positive.
Register in advance for the meeting:
https://northwestern.zoom.us/meeting/register/tJcsdu2uqDouGdUlJEu5eatDkT-HH455iUoH
*General HD support group to follow education session

Saturday, August 13th (via zoom or in-person TBD)
Couples Retreat
Please join Emily Zivin and she provides an interactive education session for couples to talk about their HD journey together with other couples in the community
Register in advance for this meeting:
https://northwestern.zoom.us/meeting/register/tJYlc-gsqDkuH9fUN4UUJ4jAHjLI7FHw0g8w
*General HD support group after education session

Saturday November 12th at 10 am via zoom (Date subject to change)
Clinical Research Update
Dr. Danny Bega
Register in advance for this meeting:
https://northwestern.zoom.us/meeting/register/tJUvde6vrz8tHNB-iwdeFGrlQxTkcU6d47wy
*Caregiver support group to follow education session

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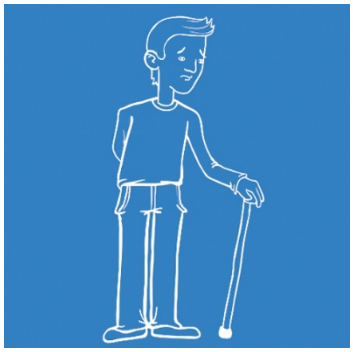
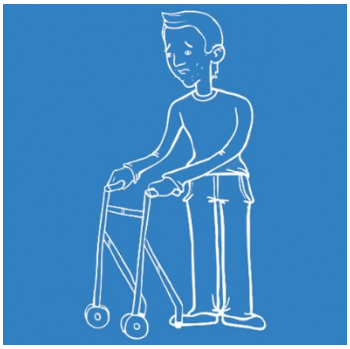


EARLY STAGE

In early stage HD, individuals are largely functional and may continue to work, drive, handle money, and live independently. Symptoms may include minor involuntary movements, subtle loss of coordination, difficulty thinking through complex problems, and perhaps some depression, irritability, or disinhibition.



MIDDLE STAGE

In middle stage HD, individuals lose the ability to work or drive and may no longer be able to manage their own finances or perform their own household chores, but will be able to eat, dress, and attend to personal hygiene with assistance. Chorea may be prominent, and people with HD have increasing difficulty with voluntary motor tasks. There may be problems with swallowing, balance, falls, and weight loss. Problem solving becomes more difficult because individuals cannot sequence, organize, or prioritize information.

LATE STAGE

In late stage HD, individuals require assistance in all activities of daily living. Although they are often nonverbal and bedridden in the end stages, it is important to note that people with HD seem to retain some comprehension. Chorea may be severe, but more often it is replaced by rigidity, dystonia, and bradykinesia. Psychiatric symptoms may occur at any point in the course of the disease but are harder to recognize and treat late in the disease because of communication difficulties.

SAVE THE DATE

HDSA Illinois Chapter Wellness Event

Saturday, October 1st from 10am to 2pm at Harper College

The chapter will be hosting this ½ day event to bring the local HD community back together again.

More details to come.

Memorials and Tributes

In Memory of Joe Etchingham from Patricia Casey & Patrick John Joyce, Michael Kinnavy, John P. & Kathleen Ruth Thornton, Karen Lewis, Michael J. & Susan Wandschneider, Thomas & Therese Connolly, Sandra L. & Steven T. Chronis, and Garden on the Run LLC.

In Memory of Judy Cuccinotto from Alice A. DePaul, Rosemary Holland, April L. & Angelo W. Bernar, Margaret A. & Robert Kiely, and Shana Helmholdt.

In Memory of Ina Mae Lindgren from Cindy & Paul Beherns.

In Memory of Terry Bruno from Teresa M Srajer.

In Tribute to Rick Haljean from Leonard & Sharon Ann Daddono.

MEDICAL RESEARCH CORNER

**Study recruitment is impacted by Covid-19 and will resume as soon as possible.

NORTHWESTERN MEDICINE HDSA CENTER OF EXCELLENCE

Sage HD Clinical Trial - To start recruiting soon

A Randomized, Placebo-Controlled, Double-Blind Study to Evaluate the Effect of SAGE-718 on Cognitive Function in Participants with Huntington's Disease. The primary purpose of this study is to evaluate the effect of SAGE-718 oral capsules on cognitive performance and functioning in participants with premanifest or early manifest HD. This study requires up to 136 days of study participation. If you are interested in learning more about the study and how to get involved, please reach out to study coordinator ZsaZsa Brown at 312-503-4121.

Development of the Virtual Unified Huntington's Disease Rating Scale (vUHDRS) - To start recruiting soon

The purpose of this study is to assess the reliability of virtually administered UHDRS compared to the in-person administration of the UHDRS to establish the use of the vUHDRS for clinical trial and regulatory purposes. This study will require up to 6 weeks of study duration. If you're interested in learning more about the study or how to get involved, please contact Destiny Gomez at 312-503-2778 or destiny.gomez@northwestern.edu**.**

MEANING INTERVENTION FOR NEWLY DIAGNOSED WITH HUNTINGTON'S DISEASE (MIND-HD) - COMING SOON

We invite people with Huntington's disease (HD) to participate in a study called "MIND-HD," funded by our recently awarded Huntington's disease Society of America Center of Excellence Pilot Grant. Our study involves two phases and will launch soon.

The first consists of optimizing health-related quality of life intervention, called "Meaning-Centered Psychotherapy (MCP)." MCP, continuously funded by the National Institutes of Health for over two decades, improves spiritual well-being in the advanced cancer population. However, MCP has yet to undergo testing for other non-cancer folks. Therefore, the optimization process will entail the study investigators leading focus groups and obtaining feedback from caregivers, family members, clinicians, and people with HD. The second phase will determine the feasibility and acceptability of the refined intervention to people with prodromal and early-stage HD. Dr. Danny Bega, MD, MSCI, and Dr. Leonard L. Sokol, MD, will lead the study at Northwestern.

Eligibility Criteria:

- Caregiver of a person with the HD genetic mutation
- Family member or friend of someone with the HD genetic mutation
- A person with the HD genetic mutation, in the prodromal or early stage of the disease
- Age 18 or older
- Have Internet access with a microphone and/or video camera
- Able to converse in English

You May Be Asked to Participate in One or More of the Following:

- Enroll in one to two, one-hour focus group sessions with other members of the HD community, discussing how the intervention can be best refined to the HD community
- Complete seven one-hour video conference technology sessions between you and a MIND-HD interventionist
- Submit answers to a brief survey and undergo post-intervention interviews to further refine the intervention for additional study

When will the recruit start?

- Coming soon. Those who are interested in obtaining additional information for the future should email Leonard.Sokol@NM.org. Dr. Sokol will provide further details soon.

Telemedicine for Huntington's Clinical Care
Individuals with Huntington's disease are invited to participate in the study "TeleHD" to determine the feasibility and value of telemedicine visits for HD patients and their care partners. This research study is conducted by Dr. Danielle Larson and Dr. Danny Bega.

Who is Eligible?

- Have a diagnosis of Huntington's Disease
- Ages 18 to 70
- Have a computer, laptop, tablet or phone with a camera, microphone, and internet access
- Fluent in English

What will you be asked to do?

- Complete two telemedicine visits (by camera at home) in addition to your two regular in-person Huntington's Clinic visits over a 6-9 month time period.
- During the visits, a neurologic exam will be performed, and you will complete two cognitive tests. The telemedicine visits will likely take less than 30 minutes.
- After each clinic visit, you will be asked to record the time and travel burden of your visit.
- After all of the visits, you will be asked to complete a survey about your satisfaction with telemedicine visits.

Northwestern Movement Disorders Center Biorepository
The Movement Disorders Center (MDC) Biorepository is a registry aimed to collect biologic and clinical information, such as blood and tissue samples, and family and medical histories from patients diagnosed with a movement disorder. The purpose of studying materials from the registry is to identify factors that either cause these neurologic conditions or increase one's risk for developing them. Samples collected for this biorepository include a blood sample (or a saliva sample) and a skin biopsy. Participants may choose to donate one or both samples.

KINECT-HD: Recruitment complete
This is a study for a new treatment for chorea associated with Huntington's disease. If you have chorea that is not currently being treated we need you. The study is of a medication called Valbenazine to treat chorea and is being conducted by the Huntington Study Group and Neurocrine Biosciences. The study involves 9 visits and will last 18 weeks. There is the opportunity to stay on the drug after the first part of the study is over. Participants will be randomly selected to receive the drug or placebo at first. We are very excited to participate as one of several sites around the country. If you or someone you know is interested in taking part in KINECT-HD, please contact our study coordinator ZsaZsa Brown at 312-503-4121 or email zsazsa.brown@northwestern.edu.

KINECT - HD 2 Study: Recruitment complete
Northwestern Medicine will be participating in an open-label extension study of Kinect-HD. The purpose of this study is to continue to gather safety and efficacy data on Valbenizine for the treatment of Huntington's chorea, while also providing study subjects who participated in Kinect-HD continued access to the study drug. In this open-label study, all subjects are given Valbenazine, even if they received placebo during Kinect-HD. Kinect-HD 2 is open to research subjects who completed participation in Kinect-HD. For more information on Kinect-HD 2 contact Zsa Zsa Brown at 312-503-4121 or zsazsabrown@northwestern.edu.

Hi-DEF Scale Study: Recruitment complete
Individuals with Huntington's disease are invited to participate in the Hi-DEF Scale Study. The purpose of this study is to learn more about impact of Huntington's disease on cognition and everyday functioning. The study involves a one-time commitment that lasts about 2.5-3 hours. Participants will be asked to complete some online questionnaires and two online cognitive tests. Once finished, the participant will be compensated for their time. If you're interested in learning more about the study or how to get involved, please contact Destiny Gomez at 312-503-2778 or destiny.gomez@northwestern.edu.

PROOF-HD - Recruitment complete
Northwestern is excited to be participating in the PROOF-HD Study. This is a phase 3, randomized, placebo-controlled study evaluating the efficacy and safety of an oral drug called Pridopidine in patients with early-stage Huntington's disease. The objective is to see if Pridopidine can *slow down functional decline* in Huntington's disease when compared to a placebo pill. If you are interested in learning more about the study and how to get involved, please reach out to study coordinator ZsaZsa Brown at 312-503-4121.

HDSA CENTER OF EXCELLENCE AT RUSH UNIVERSITY

Uniqure, a gene therapy study for Huntington's disease

Rush University Medical Center is excited to be participating in a new gene therapy trial for Huntington's disease, sponsored by Uniqure. The therapy is called AMT-130 and will hopefully slow the progression of HD by lowering the level of huntingtin protein in the brain. "Gene therapy" works by targeting genetic abnormalities that contribute to us getting sick. Administration of the therapy involves a small incision in the skull through which AMT-130 is delivered to the brain. Researchers are looking for people aged 25 to 65, with at least 40 CAG repeats in their huntingtin gene, and specific brain structure that will be assessed by MRI. Eligible participants will be randomized to receive the real treatment or a "sham" surgery involving a small mark made on the skin without making an actual incision. Study duration is approximately 5 years, during which time participants will complete physical assessments, treatment dosing, lumbar punctures, blood draws, and MRIs. Assessments and treatment will be completed across multiple sites. If you or someone you know would like to take part in the Uniqure study, please reach out to Jacob Hawkins at 312-563-5563, or email Jacob_Hawkins@rush.edu. We anticipate being ready to enroll patients in the next few months.

KINECT-HD, a phase three drug trial of Valbenazine for Huntington's chorea

Rush University Medical Center is recruiting participants for a clinical trial evaluating a drug called Valbenazine for the treatment of chorea. Valbenazine is already an FDA approved medication for another type of movement disorder that causes involuntary movements called tardive dyskinesia. The study is sponsored by the Huntington Study Group and Neurocrine Bioscience. Researchers are looking for people aged 18 to 75 with motor manifest Huntington's disease to be randomized to receive Valbenazine or placebo for 18 weeks. Participants will come to Rush for 9 research visits to take surveys, complete physical exams, and have their blood drawn. If you or someone you know would like to take part in KINECT-HD, please contact Jacob Hawkins at 312-563-5563 or email Jacob_Hawkins@rush.edu.

KINECT-HD 2, an open label rollover study for continuing Valbenazine administration for the treatment of chorea associated with Huntington disease

Rush University Medical Center is excited to participate in the open label extension study of Kinect-HD, a clinical trial of Valbenazine for the treatment of Huntington disease chorea. The purpose of this "rollover" study is to gather more safety and efficacy data on Valbenazine. Valbenazine is an FDA approved medication used to treat another type of disorder that causes involuntary movements called tardive dyskinesia. In this open label study, all subjects will be given real Valbenazine for up to two years. Kinect-HD2 is now open to all qualifying patients, not just those who participated in Kinect-HD. Researchers are looking for people aged 18-75 with motor manifest Huntington's disease. Participants will come to Rush to take surveys, complete physical exams, and have their blood drawn. The study is sponsored by the Huntington Study Group and Neurocrine Bioscience. If you or someone you know would like to take part in Kinect-HD2, please contact Jacob Hawkins at 312-563-5563 or email him at Jacob_Hawkins@rush.edu.

ENROLL-HD, a prospective registry study in a global Huntington's disease cohort

Researchers at Rush University Medical Center are looking for patients affected by Huntington's disease and their first-degree blood relatives to take part in an ongoing observational study. The data gathered in ENROLL-HD will be used to help doctors and scientists learn more about Huntington's disease and hopefully develop new treatments. Participation involves an annual visit conducted in the Rush Section of Movement Disorders at Rush University, where participants will complete surveys, cognitive tasks, family histories, and a blood draw.
in ENROLL-HD, please contact Jacob Hawkins at 312-563-5563 or email Jacob_Hawkins@rush.edu.

Cortical Control of Balance and Walking in HD

A neuroimaging study investigating brain activation during balance and walking under single-task and multitask conditions in people with Huntington's disease. We are looking for individuals with a clinical diagnosis of HD, 30 years of age and older, who can stand and walk unassisted. Participation requires one, 3.5-hour visit to *Rush* University Medical Center. This study is actively recruiting both healthy control and HD participants. Please contact Nicollette Purcell (Nicollette_L_Purcell@rush.edu) if you are interested in participating and would like additional information.

Optimization of Telegenetic Counseling for Huntington's Disease

A neuroimaging study investigating brain activity during balance and walking under single-task and multitask conditions in people with Huntington's disease. We are looking for individuals with a clinical diagnosis of HD ($\geq$40 repeats), 30 years of age and older, who can stand and walk unassisted. A study visit requires participants to come to Rush University Medical Center to perform cognitive assessments and walking and balance tasks while wearing a portable neuroimaging cap, followed by an MRI at the nearby University of Illinois-Chicago. Testing can be completed in one visit or split into two shorter visits. This study is actively recruiting both healthy control and HD participants. Individuals will be compensated for their participation. Please contact Nicollette Purcell (Nicollette_L_Purcell@rush.edu) if you, or someone you know, are interested in participating and would like additional information.

News from Our Illinois Chapter Social Worker Emily Zivin, LCSW

Huntington's Disease Society of America
Tel: 630-443-9876 or E-mail: ezivin@hdsa.org

Caring for the Caregiver

Coping With Caregiver Burnout

A caregiver is a person who helps another with their personal and medical needs. When a family member is a caregiver, this person tends to be unpaid. Typical caregiving activates include performing medical tasks, prepping meals, running errands, bathing and much more.

Being a caregiver can be both rewarding and exhausting. This role can be emotionally, mentally and physically draining. Because caregiving takes up so much time, caregivers often experience emotional and financial burden and tend to have limited social lives.

Caregiver burnout occurs when the burden and stress of caregiving becomes overwhelming and negatively impacts the life and health of the caregiver. Almost everyone who is a caregiver experiences caregiver burnout. Signs of caregiver burnout includes increased anxiety, avoidance of friends and family, depression, exhaustion, feelings of loss of control, irritability, lack of energy, loss of interest in personal life and neglecting personal health and needs. When these signs/symptoms arrive, it is clear the caregiver burnout is impacting both the physical and emotional wellbeing of the caregiver.

It's important to be aware of the warning signs. There are a number of things you can do to take care of yourself, stay healthy, and prevent burnout.

Some tools and tips that can help with caregiver burnout include:

- **Get support**: Talk about what you are feeling. Reach to your friends, families and mental health professionals.
- **Ask for help**: Let your family, friends and community know what you need from them.
- **Be truthful to yourself**: Set realistic expectations about what you can and cannot do. If you need help, delegate responsibilities to others. Remember to say no if you are feeling overburdened.
- **Connect with other caregivers**: HDSA offers virtual caregiver support groups as well as local HD support groups.
- **Take breaks**: Breaks can help relieve stress and restore energy.
- **Be social**: Continue to meet with friends and do things outside of your house. Look for hobbies that make you feel good. Activities should be something that gives you a break from caregiving and makes you feel happy.
- **Take care of your own health**.
- **Eat a healthy diet**.
- **Exercise** is a great way to reduce stress and increase energy.
- **Rest** is important to your well well-being.
- **Consider respite care**: Respite is typically a fee for service and can include an in home care provider or a short term stay at a nursing home facility.

HDBUZZ

Huntington's disease research news.
In plain language. Written by scientists.
For the global HD community.

A spoonful of branaplam helps the huntingtin go down

Branaplam was originally designed to treat spinal muscular atrophy, but a new paper outlines how it could hold promise for treating Huntington's. This oral drug lowers huntingtin protein and will now be tested in a study called VIBRANT-HD.

By Dr Rachel Harding,Edited by Dr Jeff Carroll - March 16, 2022

Scientists at Novartis and The Children's Hospital of Philadelphia have recently published a paper detailing how the drug branaplam, originally developed for the neurological disease spinal muscular atrophy (SMA), could be repurposed to treat Huntington's disease. Branaplam can lower levels of the huntingtin protein and is now being tested in the clinic in a phase IIb study, VIBRANT-HD.

Huntingtin-lowering therapies are being pursued by lots of companies

Despite setbacks with some recent clinical trials, many experts in the field agree that huntingtin-lowering remains an attractive strategy for treating HD. Every person with HD has an expansion in their huntingtin gene which means they will make an expanded form of the huntingtin protein. This expanded form of the protein seems to be toxic and is thought to cause to the signs and symptoms of HD. If we can reduce the amount of this toxic form of the protein, researchers hope we might slow or stop the progression of HD.

Lots of companies are working on huntingtin-lowering therapies, racing to see if their drug will help slow or halt symptoms for people with HD.

Lots of companies are testing huntingtin-lowering drugs in the clinic, including Roche, Wave Life Sciences, and uniQure, all of whom are using slightly different approaches to target the genetic message which is made into the huntingtin protein. The drugs they have developed cannot easily spread through the body, so they are given to patients through spinal tap or direct injection into the brain. While this means the drug can get to the parts of the body most badly affected by HD, these procedures are demanding for patients and very expensive. These are also not treatment options which could be trivially rolled out to the global community of people with HD due to healthcare access issues and prohibitive costs.

Repurposing an SMA drug to try to treat HD

What scientists call "small molecule therapies" are an attractive option to treat diseases affecting the brain. This type of drug can often be formulated so it can be taken orally as a pill or syrup, which is much easier for patients, and these drugs have a better likelihood of crossing from the bloodstream into the brain so patients can avoid onerous procedures. For a long time, it was a pipedream for many folks in the HD community that a small molecule huntingtin-lowering therapy could ever be made and then, two independent companies did just that! Very similar drugs developed by both Novartis and PTC Therapeutics can lower huntingtin – we recently wrote about a paper which describes the PTC drug on HDBuzz. Now we have more details about the Novartis drug, called branaplam.

Branaplam targets machinery in our cells which processes genetic messages, called splicing machinery. Each genetic message can be thought of like a story book, and when the story is over, the final part of the message reads the genetic equivalent of "the End" to tell the cell that the sequence for that message is complete. Drugs like branaplam rejig the pages of the story book so "The End" is read before it makes sense. When this happens, the cell will destroy the message and won't make the associated protein, similar to how you might get rid of a book that had a premature ending which made no sense.

"For a long time, it was a pipedream for many folks in the HD community that a small molecule huntingtin-lowering therapy could ever be made and then, two independent companies did just that! *"*

Branaplam was originally developed for a fatal childhood disorder called SMA because it also changes the levels of a protein called SMN2, which underlies that disease. Scientists at Novartis discovered that branaplam also changed the levels of the huntingtin protein so switched gears to test if branaplam would be a good treatment for people with HD and have now published their findings which we'll digest for you here.

Working out how branaplam lowers levels of the huntingtin protein

First, the research team treated cells in a dish with branaplam and looked at how the genetic messages in the cells were affected. They found that a signature in the huntingtin genetic message, which is normally chopped out by the splicing machinery, called a pseudoexon, was kept in the message molecule in branaplam treated cells. The scientists went on to show that this reduced the amount of the huntingtin genetic message because keeping in the pseudoexon genetic code, targets the huntingtin message to the trash bin of the cell. When the branaplam treated cells were no longer treated with the drug, this effect was reversed, and the levels of the huntingtin message bounced back to normal.

Whilst changes to the huntingtin message are a good sign, what we are really interested in is the levels of the huntingtin protein. The team measured huntingtin protein levels when different amounts of branaplam was dosed in cells in a dish and showed that the more drug was given, the more the level of huntingtin protein was lowered. The team next tested if this finding held true for cells in a dish derived from people with HD i.e., folks who have the Huntington's disease mutation. They showed that the levels of huntingtin message and protein were also reduced by branaplam in these cells too.

Insights from branaplam in HD animal models and SMA patients

Next, the scientists went on to see how branaplam performed in a mouse model of HD. Mice were given different oral doses of branaplam and then the levels of the huntingtin message were measured in different areas of the brain. In four different brain regions, they showed that the level of the huntingtin message including the pseudoexon was increased the more drug that was administered. This was matched by a decrease in the levels of the huntingtin protein. The scientists found that if mice were no longer treated with branaplam, the effect was reversed, and huntingtin levels bounced back.

Branaplam targets machinery in our cells which processes genetic messages, called splicing machinery.

Lowering the levels of huntingtin is all well and good, but what the research team really wanted to know is if this improved symptoms in the HD mouse model. Next, they tested the motor skills of the HD mice who had been treated with branaplam and compared them to HD mice which hadn't be treated as well as regular lab mice. The scientists suggest that the branaplam treated mice are more like the regular mice, but the presented data is fairly limited.

The team finally looked at the levels of the huntingtin message in blood from branaplam treated SMA infant patients. Patients in the open-label extension of the SMA branaplam trial received weekly doses of branaplam for over 2 years. After over 900 days, a sustained decrease in the levels of the huntingtin message in these blood samples could still be seen, showing ~40% decrease at this timepoint in the study. The Novartis team believes this indicates that the drug was having the desired effect over a long period of time in people.

"The next step for branaplam is a phase IIb study called VIBRANT-HD; this will be the first time branaplam is tested in adults with HD *"*

What's next for branaplam?

We recently heard from scientists at Novartis at the recent CHDI therapeutics meeting who gave us updates on their branaplam program. Dr Beth Borowsky gave us details of a now completed phase I study, where the drug was tested for the first time in adults to figure out a safe amount and frequency of dosing. As branaplam was originally developed to treat SMA in infants, figuring out a safe dose for adult patients is an important first step.

The next step for branaplam is a phase IIb study called VIBRANT-HD. This will be the first time branaplam is tested in adults with HD and this study will work out what dose of the drug needs to be administered to lower huntingtin. Branaplam will be given as an oral liquid, like cough medicine, that people in the trial will drink once a week. Different patients will be given different doses of branaplam so Novartis can work out what dose will work best for a second phase of the trial. Lots of different clinical measurements will be collected from participants in the trial, including levels of various biomarkers, like huntingtin and neurofilament. Recruitment for this trial is underway and hopefully we'll hear updates on how the trial is proceeding soon.

SUPPORT groups

We invite all those diagnosed with Huntington's Disease, their families, caregivers, and individuals who are at risk to attend our Support Group meetings. Meetings provide a supportive environment where participants can share concerns, challenges, and successes. In addition, participants can lend emotional support to one another and lessen feelings of isolation. Meetings are always free to attend, and all locations are accessible. Your involvement is important for our support groups! At a meeting you might learn about a community resource, discover a new research study, or hear from a guest speaker. Please consider joining us! For further information about any of the support groups, please contact Emily Zivin at 630.443.9876 or email at ezivin@hdsa.org.

Cancellations may occur in the case of inclement weather. We will attempt to notify everyone with advanced notice by email. If you are concerned that a meeting may be cancelled, please contact Emily Zivin at 630.443.9876 to confirm.

Illinois HDSA Chapter Virtual Support Group

3rd Tuesday of Every Month (7:00pm)
Register in advance for this meeting:
https://hdsa-org.zoom.us/meeting/register/tJEufumvpjosHtGoWGckkn3GCqwnTf8Fttn6
Questions? Contact Charlotte Rybarczyk at charlotte82963@gmail.com

MUNSTER, IN

2nd Tuesday of Even Months (7:00 – 8:30pm)
2022 Meetings: Contact Cindy Rogers for specific dates/format
Southside Christian Church, 1000 Broadmoor Avenue
Contact: Cindy Rogers (219-836-2369); clrogers111@comcast.net or Monica at 219-616-1393

Illinois HDSA Chapter Caregiver Support Group

April/June/August/October/December (7:00pm)
Contact Emily Zivin for exact dates
Winnetka Library, Community Room, lower level
768 Oak Street, Winnetka
Questions? Contact Emily Zivin at emily.zivin@northwestern.edu

Northwestern Medicine Virtual Support Group

2nd Saturday of Every Month (10:00 – 11:30am)
Odd months - Caregiver Support Group
Even months - General HD Support Group
Register in advance for this meeting:
https://northwestern.zoom.us/meeting/register/tJIqf-2vqT8qE9KddIKdiOkVyTijeHFvyhYL
Questions? Contact Emily Zivin at emily.zivin@northwestern.edu

Rush University Medical Center Group

4th Saturday of Every Month (10:30am – Noon)
For more information and Zoom details please reach out to the following support group leader:
Sarah Strait, RN (312-563-2900); sarah_strait@rush.edu

Meeting Guidelines - We read the guidelines before each meeting to remind us that we are all responsible for following and committing to the group standards, which are in place to keep this group a safe place to share.

Share the airtime - Everyone who wishes to share has an opportunity to do so. No one person should monopolize the group time.

One person speaks at a time - Each person should be allowed to speak free from interruptions and side conversations.

What is said here stays here - This is the essential principle of confidentiality and MUST be respected by all participants.

Differences of opinion are OK - We are ALL entitled to our own point of view.

We are all equal - We accept cultural, linguistic, social, and racial differences and promote their acceptance.

Use "I" language - It's important to use "I" language because you are talking about yourself and not a vague person or group of people.

The use of "I" helps avoid someone feeling like they are being attacked - Examples include: "I feel like you handled that difficult situation the best that you could have" "I had good experiences with antidepressant meds in my family"

It's OK not to share - People do not have to share if they do not wish to.

Its everyone's responsibility to make the group a safe place to share We respect confidentiality, treat each other with respect and kindness, and show compassion.

SAVE THIS DATE!

May 15th	HDSA IL Chapter Team Hope Walk – Naperville, IL
August 6th	HDSA IL Chapter Baggo Tournament – Rolling Meadows, IL
August 27th	HDSA IL Chapter Food Truck Event – Chicago, IL
September 10th	HDSA IL Chapter Team Hope Walk – Central Illinois
October 1st	HDSA IL Chapter Wellness Event – Harper College
December 4th	Celebration of Hope Brunch – Ivy Room, Chicago, IL

https://hdsa.org/il

SPRING 2022