

CHAPTER OFFICERS

PRESIDENT – Arvind Sreedharan

703-599-6000 ~ avs2004@comcast.net

VICE PRESIDENT – OPEN

TREASURER – Andy Hucker

224-715-0729 ~ ahucker1549@comcast.net

SECRETARY – Jenny Placek

630-995-6440 ~ jenniferplacek09@gmail.com

BOARD MEMBERS

Karen Bennett – karben22@hotmail.com

847-212-1240

Mary Bos - mary_bos@att.net

630-830-5329

Sarah Cozad – cozinn.sc@gmail.com

309-299-0284

Debbie Cyr – dcyr0374@gmail.com

815-303-6550

Wayne Galasek – wgalasek@aol.com

708-289-1273

Taryn Reneau – TarynReneau@aol.com

310-621-9016

Charlotte Rybarczyk –

charlotte82963@gmail.com

847-528-7354

Ann Terry – ann_terry5@yahoo.com

312-339-9356

OTHER CHAPTER MEMBERS

Emily Zivin – Social Worker

630-443-9876 ezivin@hdsa.org

Camille Colletti – Regional Director

847-849-0680 ccolletti@hdsa.org

Maryann Moynihan – Newsletter Editor

708-955-3080 shamrock1959@gmail.com

Hopes & Dreams

is the official publication of the

Illinois Chapter of Huntington's Disease Society of America, Inc.,

P.O. Box 1454, Lake Villa, IL 60046

(630) 443-9876 ~ www.hdsa.org/il

This newsletter attempts to report items of interest relating to the individuals with Huntington's Disease, their families, healthcare professionals, and interested friends and supporters. HDSA and the Illinois Chapter do not provide medical advice, nor do they promote, endorse or recommend any product, therapy or institution. Please check all drugs, treatments, therapies and products with your physician. Statements and opinions expressed in articles are not necessarily those of HDSA, Inc. and the Illinois Chapter.

President's Message



Dear HD Families,

I hope the summer has treated you well and you were able to attend one of the many unique HD chapter events. Both Baggo tournaments this summer, one organized by Larry Haigh on 6/25 and another on 8/6 organized by Charlotte Rybarczyk, were hugely successful. There were fantastic turnouts for great fundraising at both events. Thank you to Charlotte and Larry for your efforts in coordinating these fun events.

We also had an excellent turnout at the "A Night at the Races for HD" event on 8/20, organized by Wayne Galasek. It was a beautiful night to watch some harness racing and support the HD IL Chapter. Thank you, Wayne, for putting on a creative event that I suspect could be an HD tradition.

In addition, we had a fun food truck Center of Excellence appreciation day on 8/27 at Lake Shore Park in the City. It was an opportunity to recognize members of the Rush and Northwestern COEs for all they do for the HD community on a beautiful summer afternoon.

Finally, special thanks to Sarah Cozad for her planning efforts for the Team Hope Galesburg Walk on 9/11. See some photos on page 9.

We have a wonderful regional event planned for the end of the year. The annual Celebration of Hope Event will take place on Sunday, December 4, 2022, at the Ivy Room in Chicago, IL. The Illinois HDSA Chapter and Upper Great Lakes Region takes pride in organizing this memorable event. This year we will be honoring BJ Viau. BJ has played a considerable role in the HD community on multiple fronts since his mom's diagnosis in 1995. He also co-founded the HDYO, an international non-profit to help young people impacted by HD.

For more information on this event, please see the link, <https://uppergreatlakes.hdsa.org/events/2022-chicago-celebration-of-hope>

I look forward to seeing you at this great event later this year.

Arvind Sreedharan
President, HDSA Illinois Chapter

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

Taking care of your mental health is essential for maintaining a happy life. Mental Health impacts how we think, feel and live our lives. There are many ways how we feel impacts our everyday living including:

- Relationships
- Physical health
- Emotional well being
- Reducing suicide rates
- Productivity and financial stability

It is important when facing difficult times, that individuals know about resources that can help.

HDSA offers 8 free counseling sessions a year for anyone impacted by HD. More information can be found here: <https://hdsa.org/find-help/community-social-support/hdsa-telehealth/>

HDSA offers free online support groups: <https://hdsa.org/find-help/community-social-support/hdsa-support-groups/>

HDSA Illinois Chapter offers in-person and online support groups: <https://illinois.hdsa.org/about/support-groups>

In addition to HDSA specific resources, you can also connect to a private therapist who can work with you on an individual basis.

Taking care of your mental health is very important. There are resources that can help. You are not alone.

Fifth Annual Baggo Tournament a Success!!

We are thankful for all of the sponsors, donors, participants, and volunteers who helped to make our fifth annual HDSA Illinois Chapter Baggo tournament a success! On Saturday, August 6th in Rolling Meadows, 19 teams played in a double elimination bracket vying to win custom Baggo games built by R&R Custom Cabinetmaking. The heat did not ruin the fun as everyone played and enjoyed an Italian Beef and hot dog lunch provided by Vienna Beef. A silent auction and split the pot raffle rounded out the festivities. Our winners this year were Michael Marchetti & Nick Kidrick (AKA The Hole Shockers). And, best of all, over \$12,000 was raised for our HD families in Illinois!



A HUGE THANK YOU TO OUR BAGGO SPONSORS:



JOE RYBARCZYK
KYLE & KRISTIN RYBARCZYK
NICK KEYZER LARRY HAIGH
DEBBIE & TIM CYR



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Huntington's Disease
Society of America



UPPER GREAT LAKES REGION

The Huntington's Disease Society of America
Upper Great Lakes Region
invite you to attend the 18th Annual

Celebration of Hope

Help for Today. Hope for Tomorrow.

Honoring BJ Viau

Sunday, December 4, 2022 at 1:00 p.m.

Brunch at The Ivy Room

12 E. Ohio Street, Chicago, IL
www.hdsa.org/coh-chicago



Huntington's Disease
Society of America



UPPER GREAT LAKES REGION

Celebration of Hope

Join us for Brunch, Silent and Live Auctions, Mimosas, and Live Music to benefit The Huntington's Disease Society of America

Sunday, December 4th at 1:00pm
The Ivy Room
12 E. Ohio Street, Chicago, IL 60611

**Register at www.hdsa.org/coh-chicago
Tickets \$100 Table of 8 \$750**

Contact Deb Boyd for more information or sponsorship opportunities
269-303-9119 dboyd@hdsa.org

The Celebration of Hope program recognizes the incredible work done across the network of fifty-four HDSA Centers of Excellence. These HD centers provide an elite multidisciplinary approach to Huntington's disease care and research. Two of these world-class facilities are located in Illinois at Rush University Medical Center and Northwestern University. Patients seen at Rush and Northwestern benefit from expert neurologists, psychiatrists, therapists, counselors and other professionals who have deep experience working with families affected by HD. These professionals work collaboratively to help families plan the best HD care program.

All contributions are tax deductible to the extent provided by law. HDSA operates under section 501(c)3 of the IRS code. We estimate the fair market value of one ticket to be \$60. The charitable contribution per ticket is \$40. HDSA EIN#: 13-3349872.

teva
National Sponsor



Danny Bega MD, MSCI
Associate Professor, Department of Neurology
Division of Movement Disorders
Northwestern HD Clinic
Galter Pavilion, 675 N. St. Clair, #20-100, Chicago, IL 60611
312-695-7950 (main)
312-695-5747 (fax)
hd@nm.org (email)
<http://huntingtons.nm.org>
<http://www.rush.edu/services/conditions/huntingtons-disease>

Improving Quality of Life in Huntington's Disease Through Exercise

By Sarah Brooker, MD, PhD; Sushma Kola, MD; Danny Bega, MD
Northwestern Medicine HD Center of Excellence

For people with Huntington's disease (HD), exercise is an important part of a healthy lifestyle and can significantly improve quality of life. The impacts of exercise are numerous and range from direct benefits on balance, strength, and endurance to more indirect benefits on level of independence, mood, and cognition. People living with HD often have difficulty with motor skills, and a regular fitness regimen can help address problems with walking, balance, and daily activities. Exercise can also improve fatigue, mood, and cardiovascular fitness in this population. The positive impact of exercise on cognition has also been well described. Several studies of people with HD, as well as people with other neurodegenerative disorders, have demonstrated a correlation between increased physical activity and improved intellectual function on tests of memory and other cognitive skills. Lastly, group exercise can be a great way to remain engaged in the community, develop and strengthen friendships and maintain a social network.

To reap the benefits of exercise, there is not one specific type of activity that people with HD must follow. Rather, the exercise plan must be individualized for each person based on their interests, lifestyle, and needs. Perhaps the most important aspect of choosing an exercise regimen is finding out which activities are most fun, enjoyable, and practical. An exercise plan that one is motivated to participate in and stick with long-term is the most likely to be beneficial. The key components of fitness are *balance, flexibility, strength, and endurance*. Balance is particularly important for people with HD since the movements caused by chorea lead to unpredictable shifts in center of gravity. Therefore, working on improving balance can help to prevent falls and improve the person's level of independence.

The specific exercises chosen should cater to the individual's abilities, with a focus on safety and being aware of one's limitations as they change over time. Exercise can be beneficial for people with HD across all disease stages. For people who are pre-symptomatic or have only very mild symptoms, a more rigorous exercise plan can be designed with regular activities that combine balance, strength-training, and aerobics. For people who are more symptomatic but can walk independently, a focus on maintaining independent gait is important. These individuals may incorporate a regular walking routine as well as activities that target balance, such as yoga, tai chi, or dance. For those who require assistive devices or help from others to walk, it is recommended that they have someone walk with them. And for those who have more difficulty walking, there are numerous exercises that can be done in a seated or recumbent position, including hand cycling, chair aerobics, or stretching. In addition to more formal exercises, active hobbies can also be very beneficial, such as gardening, crafting, and cooking.

When designing an optimal exercise plan, involving a multidisciplinary care team can be immensely useful to learn new exercises that are the most beneficial and safe. Therapy teams can help to individualize fitness plans and design exercises that are most suited to the patient's abilities and needs. Multidisciplinary therapy teams can include physical therapists, occupational therapists, speech therapists, exercise physiologists, and physical medicine and rehabilitation physicians. Together, these team members can assess each person's baseline ability level, address needs, and teach strategies for balance, fall avoidance, and optimizing independence and daily function.

Home Exercise Resources for Huntington's Disease

Chair-based Exercise Videos for HD:

<https://vimeo.com/289892489>

<https://vimeo.com/289893681>

Shana Verstegen's HD Workout Tips:

Balance

<https://www.youtube.com/watch?v=goA-WZZKJHc>

<https://www.youtube.com/watch?v=1KQoHjXfRIE&t=12s>

<https://www.youtube.com/watch?v=71g4Vq4UOUg>

Posture

<https://www.youtube.com/watch?v=Um2ZdRXx64E>

<https://www.youtube.com/watch?v=traWPM6V5SY>

<https://www.youtube.com/watch?v=RbmVoTU5MZg&list=PLLQmMRDsNEY1cLo>

Mobility

https://www.youtube.com/watch?v=W_AbU88Eg3w

<https://www.youtube.com/watch?v=7ShuCscAiBA>

<https://www.youtube.com/watch?v=I-UFQoFJr6Q>

Flexibility

<https://www.youtube.com/watch?v=Un2KaxF6GJI&list=PLLQmMRDsNEY1cLor6>

<https://www.youtube.com/watch?v=rJ6WSHJDZro&list=PLLQmMRDsNEY1cLor6>

<https://www.youtube.com/watch?v=nqHfswWgMWA&list=PLLQmMRDsNEY1cLo>

Cardio

<https://www.youtube.com/watch?v=E2QSPMdUEoA>

<https://www.youtube.com/watch?v=2JI20dWIYiY>

<https://www.youtube.com/watch?v=FW5h4IgyUM0&list=PLLQmMRDsNEY1cLor6>

<https://www.youtube.com/watch?v=RbXnenPoqDw&list=PLLQmMRDsNEY1cLor6>

<https://www.youtube.com/watch?v=Hp7yTGgUcHY&list=PLLQmMRDsNEY1cLor6>

Core

<https://www.youtube.com/watch?v=xwfNaKGYCBk&list=PLLQmMRDsNEY1cLor6>

<https://www.youtube.com/watch?v=gQe7nDEylzU&list=PLLQmMRDsNEY1cLor6>

<https://www.youtube.com/watch?v=W0D40DJzWjk&list=PLLQmMRDsNEY1cLor6>

https://www.youtube.com/watch?v=u_XCWHoHduo&list=PLLQmMRDsNEY1cLo

<https://www.youtube.com/watch?v=109->

<https://www.youtube.com/watch?v=hmRkjbzbaOtc&list=PLLQmMRDsNEY1cLor6>

Additional Exercise Resources:

Movement Disorders

<https://www.youtube.com/playlist?list=PLoi2wccX7y->

Silver Sneaker's Exercise Video

<https://drive.google.com/file/d/1a1zuMiWrklIWLAXIjJ3n5WBCDh8Et6PU/view>

Physical Therapy Class for Ataxia

<https://www.youtube.com/watch?v=Z7DGPR8DVwY&feature=youtu.be>

Memorials and Tributes

In Memory of Gus Phillipps from Art, Dean, & Irene Karamagianis; Mark Riske & Pamela Wieland; Peter & Demetra Baziotis; Collette Sarlas; Mary Ann & Mark Schwandt; James & Mary A Lande; and Demetrius & Lois Andressakis

News from HDSA Center of Excellence at Northwestern Medicine



Virtual Patient and Family Education Series 2022

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/joining/register/tJUvde6vrz8tHNB-iwdeFGriQxTkU6d47wy>

*Caregiver support group to follow education session

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.

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 open again

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 Valbenazine 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 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 (≥ 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.

Photos from Team Hope Galesburg Walk on September 11th





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

Serious side effects reported for some people treated with the huntingtin-lowering drug AMT-130, currently in clinical trials

After receiving a high dose of uniQure's gene therapy for Huntington's disease, a few patients experienced serious side effects, but are now recovering. HDBuzz explores what this means for the experimental huntingtin-lowering drug AMT-130.

By Dr Sarah Hernandez and Dr Rachel Harding, Edited by Dr Rachel Harding

Last month, we relayed positive news from uniQure's trial testing AMT-130, a gene therapy delivered via brain surgery to lower huntingtin (HTT). Data released by uniQure in June suggested [AMT-130 was safe](#) and well tolerated in the small group of people that were treated with a low dose of the drug. Now we're back to provide an update on findings from the group of people treated with a higher dose of AMT-130. This new set of data shows that the higher dose of the drug may be causing serious side effects. This doesn't necessarily mean AMT-130 doesn't work and won't move forward, but it does mean that we need to take a pause, really look into what the data are telling us, and work out a safe plan to move forward for people being treated with the drug.

Despite setbacks, HTT lowering is still considered an attractive strategy by many researchers

One advantage researcher that study Huntington's disease (HD) have is that we know exactly what causes HD - an expansion in the HTT gene. The expanded HTT gene produces an expanded HTT message that is then processed into an expanded form of the HTT protein that causes damage in brain cells. So, in theory, reducing the presence of that expanded HTT protein could alleviate the symptoms associated with HD because it directly targets the root cause of the disease. This means that despite recent setbacks for several clinical trials designed to lower HTT levels, HTT lowering is still considered an attractive strategy for HD therapeutics by many researchers.

There are several different ways researchers are trying to lower HTT. The first horse out of the gate in the HTT lowering race were antisense oligonucleotides (ASOs). These are short sequences that bind to a specific message which then cause it to be degraded. Without the message, no protein can be produced. So, while the gene remains intact, the protein is never made. This type of HTT-lowering technology is being explored by Roche with their drug tominersen that took a step back to find the right dose and patient population. Wave Life Sciences is also using ASOs to selectively lower the expanded copy of HTT with their ongoing Phase I/II trial for WVE-003, SELECT-HD.

Another way to lower HTT that's being tested in clinical trials is through "splice modulators". These are drugs that change how the genetic message is edited. Like a story, every gene has a beginning, middle, and end. The end is a specific sequence that tells molecules in the cell to stop reading the code for that gene. Splice modulators work by editing the message to move that ending code up, confusing the sequence of that gene. So rather than a beginning, middle, and end, the story is just a beginning and end. The cell recognizes that this makes no sense and stops producing that protein.

HDBuzz recently wrote about the splice modulator branaplam, being tested by Novartis in the VIBRANT-HD study, [for which dosing was suspended due to safety concerns](#). Another splice modulator, PTC-518, is being tested by PTC Therapeutics. Even though PTC-518 works in a similar way to branaplam, a head-to-head comparison of these drugs suggests they are actually quite different. So bad news for one doesn't necessarily mean there will be bad news for the other. We're still eagerly waiting for news about the PTC-518 trial!

AMT-130 is a one-shot gene therapy approach to lower HTT

"Scientists at uniQure believe that the higher dose of the drug will not necessarily lower HTT further in each cell, but that more drug will mean that more brain cells will have their levels of HTT lowered by the same amount. "

A third way to lower HTT is through gene therapy, which is the technology being used by uniQure with AMT-130. This drug works by using a harmless virus to deliver DNA instructions that will destroy the HTT message. The HTT gene still exists in its original form, but now the cell contains a new message that will prevent the production of the HTT protein. Because the cells infected with the harmless virus contain the genetic instructions, they can make the HTT lowering message all on their own. This means AMT-130 is a one-and-done approach - deliver the therapy through a single procedure, and the cells will

continue to make the instructions that allow them to lower HTT. This is both exciting and nerve-wracking. While it means only one treatment is necessary, it also means any changes are likely permanent.

To get AMT-130 directly where it's needed most - the brain - it's delivered using brain surgery. Because brain surgery is always risky, this trial was rolled out very slowly to be as careful as possible. After the first 2 surgeries were complete, the participants were watched to make sure there were no immediate negative effects. When everything went well, surgery for the rest of the study participants continued.

The trial testing AMT-130, HD-Gene-TRX1, is a Phase I/II designed to test safety and tolerability of the drug as well as find the right dose that will work for people with HD. Because one of the primary goals of this study was finding the right dose that will work best for people with HD, 2 groups were tested: a low dose group and a high dose group. Scientists at uniQure believe that the higher dose of the drug will not necessarily lower HTT further in each cell, but that more drug will mean that more brain cells will have their levels of HTT lowered by the same amount.

36 people in total were enrolled in uniQure's AMT-130 study: 10 that received an imitation surgery that will act as the control group, a critical part of any study, and 26 people in the treatment group. Of the 26 in the treatment group, 12 were treated with a low dose of AMT-130 and 14 were in the group for the high dose. So far, 12 of those 14 have undergone surgery.

In June we got an update from uniQure about people that were treated with the low dose of AMT-130 12 months after their surgeries, [which HDBuzz wrote about](#). In that group, the surgeries and drug were well tolerated with no major safety issues reported. uniQure shared that preliminary data indicating that HTT seemed to be lowered more in the group treated with AMT-130 than the control group. While this is exciting news because it means AMT-130 appears to be doing what we want it to do - lowering HTT - this was reported in a very small group of only 4 participants.

Some participants in the AMT-130 study have suffered serious side effects

In early August, uniQure made an announcement about participants from the high dose group in the AMT-130 study. Three participants (out of 14) from this arm of the study were found to have severe adverse reactions by an independent safety review committee. Two people that underwent surgery in Europe reported swelling and a third person, treated at a U.S. location, reported a severe headache and related symptoms shortly after surgery. While this is very upsetting and disappointing news, importantly, all three patients have either fully or substantially recovered and have now been released from the hospital.

What's next for AMT-130?

There are many theories as to why these patients suffered these side effects, including some form of immune response. However, there is no clear or definitive explanations just yet and we must wait for further information before jumping to conclusions.

While the safety review committee doesn't suspect the effects observed in the high-dose group of the trial are due to the drug itself, surgeries for the remaining 2 participants in this arm of the study have been halted for now. The low-dose arm is proceeding as planned and all trial participants - in both the low- and high-dose groups - will continue to be followed for the duration of the trial. uniQure still expect to report data from the trial according to the originally planned schedule and we will be hearing further updates from the company about this trial in early 2023.

What does this mean for HTT lowering as an approach to treat HD?

The HD community has received disappointing news from many of the HTT lowering trials now and it is easy to feel like perhaps this is not a good strategy to keep pursuing to try and treat people with HD. It is important to keep a few things in mind though as all is not lost just yet. All of these trials have suffered very different problems and we only really have theories for why they haven't panned out as we hoped, all of which might be unrelated to HTT lowering itself. All of these trials are also treating people with HD who are already showing symptoms and perhaps these folks are more vulnerable to potential side effects from these drugs. It's important to note that none of these trials have given us a definitive answer as to whether HTT lowering in people with HD will improve symptoms or change the course of the disease. As the uniQure trial continues, we hope that the next data release might shed some light on this important question.

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/tJEufumvpjosHtGoWGckn3GCqwnTf8Ftn6>

Questions? Contact Charlotte Rybarczyk at charlotte82963@gmail.com

Northwestern Medicine Virtual Support Group

2nd Thursday of Every Month (7pm via Zoom)

Odd months - Caregiver Support Group

Even months - General HD Support Group

Register in advance for this meeting:

<https://northwestern.zoom.us/meeting/register/tJlqf-2vqT8qE9KddkDiOkVyTijeHFvyhYL>

Questions? Contact Emily Zivin at emily.zivin@northwestern.edu

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); crogers111@comcast.net or Monica at 219-616-1393

Rush University Medical Center Virtual 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

Illinois HDSA Chapter Caregiver Support Group

June 29/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

*****IN PERSON*** LAKE COUNTY**

2nd Monday of Every Month (7:00 – 8:30pm)

Advocate Condell Medical Center, 801 Milwaukee Avenue,
West Tower, Libertyville, IL

Contact: Marilyn & Barry Kahn (847-975-2403);

marilynkahn1@gmail.com

(Call for additional information)

*******IN PERSON*******

NORTHWEST INDIANA HUNTINGTON'S AWARENESS, SUPPORT & HOPE

3rd Thursday of Every Month (6:00 – 7:00pm CST)

Methodist Hospital Southlake, 200 East 89th Avenue, Pavilion B,
1st Floor Conference Room, Merrillville, IN 46410

Contact: Amy Turner Ladow (Mobile: 610-241-2753);

nwiHDASH@gmail.com or amyturnerladow@gmail.com.



Here is the link to the NWI Facebook Meeting Event which has all the details in the body.

<https://www.facebook.com/events/1088870821982032>

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.

It's 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!

December 4th

Celebration of Hope Brunch – Ivy Room, Chicago, IL

<https://hdsa.org/il>

AUTUMN 2022

