

THE marker

Huntington's Disease Society of America

HDSA Marches on Washington



20
YEARS

In 1993, scientists discover the gene that causes HD. This issue celebrates discoveries and advancements in research and care.



Huntington's Disease
Society of America



From the Desk of

Louise Vetter

HDSA Chief Executive Officer

In April 2013, the Huntington's disease community commemorated an historic milestone – the 20th anniversary of the discovery of the gene that causes HD. As many of you know, 1993 capped a decade in which HD researchers searched for the elusive gene site using the marker they had identified in 1983.

To observe this important date in HD history, HDSA sought to bring our research and advocacy arms together for a two day event in Washington, DC that featured both a Research Symposium and an Advocacy Day on the Hill.

On April 3, HDSA and Senator Kirsten Gillibrand (our HD champion in the U.S. Senate) hosted a Research Symposium that featured Francis S. Collins, MD, PhD, Director of the National Institutes of Health, and former Director of the Human Genome Project, speaking on the impact of genomics in advancing science. Joining him were three of our illustrious “gene hunters” – HD researchers who were an integral part of the collaborative that found the gene in 1993 – Jim Gusella, PhD, Marcy MacDonald, PhD, and Nancy Wexler, PhD. Each told a part of the HD story from the actual search to the present constructs in HD research to what lay on the horizon for investigators worldwide. More

than 100 HDSA advocates attended the Symposium which was held in the Hart Senate Office Building. A special thanks to Senator Debbie Stabenow (MI) for securing the space which had a breathtaking view of the U.S. Capitol dome.

Thanks to a generous unrestricted educational grant from Lundbeck, HDSA was able to live stream the Symposium to HD homes across the country. For those who missed the broadcast, or who would like to view the Symposium again, you can find it on the HDSA website at hdsa.org/research/hd-gene-symposium-20-years. It was a truly wonderful event, and I encourage all of you to share the link to the archived file with your family, friends and healthcare professionals.

At twilight, advocates marched to the Capitol and lit blue glowing “candles” to symbolize those for whom they would be speaking the next day during Capitol Hill visits. Thanks to our Washington, DC based advocacy firm, more than 111 Congressional visits were scheduled for the following day.

Thursday April 4 was an action packed day beginning with a half day of training on how to effectively advocate for HD.

The process is easy and HDSA is here to help you take that first step. If you are interested in joining our efforts to

change the outdated guidelines used by the Social Security Administration to determine disability for a person with HD AND if you want the two year waiting period for Medicare benefits to be waived for a person with HD, then contact Jane Kogan, HDSA Advocacy Manager at jkogan@hdsa.org.

After firing up our 100 advocates, they were sent forth to meet with their respective Congressional representatives in order to secure their members commitment to co-sponsor the Huntington's Disease Parity Act (HR1015/S723).

Throughout the afternoon, advocates traveled between Congressional office buildings using the skills they had learned that morning to change a “maybe” to a “YES”. Advocacy Day 2013 was a huge success but our advocates still have much to do – as do we all. The Huntington's Disease Parity Act cannot become a reality without the help of every member of the HD community. I urge you to lend your voice to HD and our advocacy efforts by sending an email or making a phone call to your Congressional representatives.

Together, we **CAN** make a difference. ■

Steve Seekins

Chair, HDSA Board of Trustees

Welcome to the latest issue of *The Marker*. We have so much to share with you in this issue! From new programs and services for our families to re-introduction of the Huntington's Disease Parity Act in the 113th Congress, to a Research Symposium that commemorated the 20th Anniversary of the Discovery of the HD gene to the 28th Annual HDSA Convention, there is so much to offer in the following pages.

This is my first welcome as the Chair of the HDSA Board of Trustees and I am excited to hold this position. I have been active in Huntington's Disease and HDSA for over a decade, first as a member of the Los Angeles Chapter and then as a national trustee involved in nominations and governance and the new Strategic Plan (online@hdsa.org).

This will also be my first HDSA Annual Convention as Chair and I cannot tell you how much I am looking forward to seeing all of you in Jacksonville, FL. The 28th Annual HDSA Convention will be held from June 21-23 at the Hyatt Regency Hotel in downtown Jacksonville. It's located along the banks of the St. John's River and when you need a little downtime from the hustle and bustle of the Convention,

you can re-charge by strolling along the river walk.

The Convention Program Planning Committee has been hard at work creating a vibrant and comprehensive educational program that provides vital information for every group in our community - from caregivers to those at-risk to youth and those symptomatic. We are very pleased to offer two keynote speakers this year – on Friday following the Opening Ceremony and on Saturday morning. You can read more about the Convention program on page 17.

We are also very pleased to announce that this year your Convention registration fees also include TWO meals – a luncheon on Friday which features an awards ceremony for our outstanding chapter and affiliate volunteers and a breakfast on Saturday which launches our second day of Convention activities. Also included is our ever popular Saturday evening Gala and National Awards Dinner that features the traditional candle lighting ceremony.

The HDSA Activity Center for People with HD will also be open for those who have HD and would like to spend an hour or more while their caregiver

attends a workshop or two. The HDSA Activity Center for People with HD will feature arts and crafts, yoga, music, games, and so much more. Look for the location in the Convention program and be sure to check it out.


The HDSA Exhibit Hall will also undergo a transformation this year. Rather than opening on Friday morning, we are offering our Convention attendees the chance to tour the hall and meet our vendors on THURSDAY afternoon. The first 50 folks in the Exhibit Hall will get a free t-shirt and will be entered in a raffle, so be sure to head on over on Thursday after 2:00 p.m.

The National Youth Alliance will be holding their annual Silent Auction on Friday. This is the NYAs largest fund raising event and the proceeds go to fund scholarships for youth to attend the HDSA Convention. Please stop by the NYA Silent Auction and be as generous as possible.

In closing this inaugural message to you, I wish you safe travels if you are journeying to Jacksonville and a heartfelt thanks for all you do for HD and HDSA. ■

Table of Contents

HDSA Research.....	2-9	Advocacy.....	13-16	Ways to Give.....	18-20
New at HDSA	10-12	Community	17, 21, 26-27	Living with HD.....	22-25



Venice Says “Benvenuti” to HD Researchers from Around the World as the Host of the 8th Annual CHDI Therapeutics Conference

Every four years, the CHDI Therapeutics Conference takes their show on the road. Instead of meeting in Palm Springs, this year the CHDI Foundation, a global private foundation funding HD science, hosted nearly 300 HD researchers from academia, non-profit, biotech, pharma and government in Venice, Italy for four days of intensive scientific sessions. This was the highest attended conference in the eight year history of the event. While there are many HD relevant meetings, this is the only conference dedicated to providing a forum for HD “drug hunters,” pharmaceutical and biotech companies working in HD, to share ideas and discuss their progress in the push to develop effective therapies for HD.

On the same day the Conference began, exciting news quickly traveled to the attendees. On April 8th, a formal announcement was made saying a

large partnering deal between Isis Pharmaceuticals and Roche had been struck. In the deal, Roche agreed to pay Isis \$30 million to develop its huntingtin antisense oligonucleotides (ASOs) for HD. If the Isis ASOs successfully meet their clinical trial milestones, the deal includes an additional \$362 million in payments to Isis.

The large Phase III clinical studies that are required for regulatory approval can cost hundreds of millions of dollars. This makes the identification of a development partner essential for small companies like Isis. Besides the financial aspect of the deal, the partnership is also very significant because now another major pharmaceutical company also sees HD as a viable business strategy. Roche also has important technological and personnel resources at its disposal that it will be bringing to bear on HD.

Just like in 2012, the CHDI Conference was kicked off by a session dedicated to systems biology, a holistic study of an organism, which is viewed as an integrated and interacting network of genes, proteins and biochemical reactions, not individual components. CHDI’s Chief Scientific Officer Robert Pacifici highlighted the importance of a systems biology approach to better understand HD, because new technologies are now available that generate an immense amount of HD data. He described that these data sets are simply too large to be consumed and interpreted by conventional means. In addition, despite the identification of the huntingtin gene 20 years ago, scientists are finding that HD is a much more complex disease than originally suspected. Huntingtin has many biological functions. These functions are all part of different biological pathways and networks that simply do not work in

isolation. For these reasons, the HD research community is committed to building a new “map” of HD. The end results should be better therapeutic intervention points, better biomarkers and a more complete understanding of the most appropriate animal models to use in the development of HD drugs.

The afternoon session on day 1 addressed the hypothesis that HD is a developmental disease. Elena Cattaneo (University of Milan) discussed the evolution of the polyglutamine repeat in huntingtin. She also presented data using neural stem cells that further suggests that the huntingtin protein is essential for early brain development. Peg Nopoulos (University of Iowa) presented an update on the Kids-HD research program that hopes to understand if there are behavioral and developmental alterations in at-risk, gene expanded children and at-risk, non-gene expanded children aged 6-18. Dr. Nopoulos has observed loss of cerebral cortex starting as early as 7 years of age. These anatomical changes are correlated to behavioral changes in the children. However, it was discussed that more robust statistical analyses must be performed to confirm the significance of these changes.

Dr. Ali Brivanlou (Rockefeller University) presented data from human HD embryonic stem cells (hESCs). Using these cells Dr. Brivanlou has identified four naturally occurring huntingtin mRNA isoforms. The significance of these findings will be further tested, but they could result in new huntingtin proteins with altered functions. In addition, he explained that hESCs with 48 CAG repeats

showed significant decreases in levels of bioenergetic molecules (ATP, ADP, and AMP) and aerobic glycolysis. This could provide further clues to the early biological systems that become dysfunctional early in the course of HD.

Day 2 focused solely on the best and most validated target for HD, mutant huntingtin. We heard from experts on the role of mutant huntingtin in HD and received updates from Phillip Gregory of Sangamo Biosciences and David Corey of UT Southwestern Medical Center on novel approaches to target the huntingtin DNA and RNA respectively. Sangamo's technology uses zinc finger proteins to selectively target the CAG region of the mutant huntingtin DNA sequence. In Venice,

For people with HD and their families, day 3 contained some of the more exciting results. The morning session was devoted to hearing about CHDI late stage therapeutic programs. Pfizer, working in collaboration with CHDI, reported new HD animal data using their PDE10A inhibitor (MP-10). Pfizer's data suggest that their PDE10 inhibitors preferentially affect the indirect pathway in the basal ganglia, the portion of the brain that is highly susceptible to degeneration in HD. The basal ganglia is critical for the normal control of many biological functions such as cognition and motor control. This pathway is thought to be preferentially effected in HD. Inhibition of this enzyme, which is predominantly localized to the



they reported their latest data showing they have effectively lowered huntingtin levels by around 50% in a mouse model of HD. They will now study the effects of their ZFPs when expressed over time using viruses to deliver the intervention.

striatum appears to normalize the way the synapses fire in HD mice and rats. Planning is now underway to access the safety and tolerability of MP-10 in people with early HD for 28 days. In addition to safety, readouts using fMRI, as well as behavioral and motor tasks, will be assessed. Pfizer does not expect

Continued on page 10

Overview of Stem Cells in HD Research and Development

George Yohrling, PhD; HDSA Director of Medical and Scientific Affairs

One of the main goals of the five year HDSA Strategic Plan is to support HD research and communicate its impact. In an effort to improve our research communication activities, HDSA recently launched a new HD Research Webinar Series. These monthly webinars will provide a regular forum for persons with HD, families and caregivers to hear updates on the latest research findings directly from HD scientists.

Stem cells are an area of research that has received a lot of attention lately from both researchers and the community. To address this, HDSA invited Dr. Jamshid Arjomand, from CHDI Foundation, to be the first presenter in the research webinar series. Dr. Arjomand is the Director of Basic Research at CHDI where he oversees a large portfolio of HD stem cell-related research activities. Dr. Arjomand presented on the use of stem cells in HD research and therapeutics.

Stem cells are unspecialized cells capable of continuous renewal and have the capacity to become any cell type or organ. This characteristic is referred to as pluripotency. There are several varieties of stem cells which were briefly discussed. One type of stem cell that holds great promise is the induced pluripotent stem cell (iPSCs). These cells were described by Yamanaka et al in 2006 when his group discovered a way to reprogram adult skin into “pluripotent” stem cells. This “revolutionary” finding resulted in Dr. Yamanaka being awarded the 2012 Nobel Prize for Physiology and Medicine.

Several different approaches to stem cell transplantation therapies were highlighted by Dr. Arjomand. The approach currently being tested, in small clinical studies, involves fetal brain transplants, in which small dissections of the developing fetal brain(s) are implanted into the regions

most affected in HD, with the hope that the fetal graft will replace the dying HD striatal neurons. The largest such study is the ongoing clinical trial called Multicentric Intracerebral Grafting in Huntington’s Disease (MIG-HD) being performed in France. The primary completion date of this trial is slated for mid-2013.

Mesenchymal stem cells (MSCs) are another experimental option and their potential is only now being realized through basic mouse and rat studies. MSCs are predominantly derived from bone marrow and seem to have a much more limited differentiation capacity and are thus referred to as “multipotent.” MSCs can be transplanted directly, or genetically engineered, to express and secrete important growth factors, such as brain-derived neurotrophic factor (BDNF), a peptide that is commonly decreased during the course of HD. Several different animal

studies injected MSCs directly into the brain of HD rodent models and demonstrated potential in mediating some aspects of neuronal repair. Another approach capitalizes on the endogenous adult stem cells found in each of our brains. Although only tested in mice, the strategy is to coerce

differentiated into striatal neurons and implanted into the brains of the same person with HD. However, this strategy is, at this time, only theoretical, and many technical hurdles still need to be resolved before any clinical implementation could be considered.

Stem Cell Research has created a website (www.closerlookatstemcells.org) that allows the public to better evaluate the claims made by some clinics offering treatment or soliciting trial participants. It also details 10 important points all people should know about stem cell treatments.

“Stem cells are an area of research that has received a lot of attention lately from both researchers and the community.”

these cells to multiply, migrate and mature into striatal medium spiny neurons. However, these new neurons would still carry the mutation that is responsible for HD, so the long-term benefits of such an approach would need to be evaluated carefully.

Finally, iPSCs can be created from a person with HD and the expanded huntingtin mutation can be corrected in a cell culture dish. In theory, these corrected cells could then be

While all very exciting possibilities, there are many questions that must be carefully addressed to ensure that the stem cell treatment is safe and effective. Animal studies to understand the “what, where and how” of the different technologies must be conducted. These unknowns do not stop some from advertising stem cells as a miracle treatment for a wide array of diseases. To address this troubling trend, the International Society for

Finally, the potential use of stem cells in HD research and development was discussed. A large consortium of stem cell/HD researchers was formed in 2010 to spearhead this effort. The group is primarily funded by NINDS and CHDI, but other organizations, such as HDSA and HDF, are also financially supporting these research efforts. The goal of the HD iPSC Consortium is not to develop a cell therapy for HD, but instead is focused on developing better and more relevant neuronal models of Huntington's disease for basic research, target identification & validation, and drug discovery efforts. The Consortium will also develop a repository of well characterized HD stem cells and protocols that will be made available to the entire HD research community.

If you are interested in viewing this presentation, this webinar was recorded and is now archived on the HDSA website at www.hdsa.org/researchwebinar. If you would like more information on stem cells, the NIH has created a helpful online resource at: <http://stemcells.nih.gov/info/basics/pages/default.aspx> ■



Prana Biotechnology publishes positive results for PBT2 in animal models of HD

Researchers from Prana Biotechnology and UCSF have recently published data on the small molecule, PBT2, in two different animal models of Huntington's disease (Cherny et al). PBT2 is a novel compound that is known to cross the blood-brain barrier, appears safe to humans, and has demonstrated neuroprotective effects in various brain disease models. The purported mechanism of action of PBT2 is to act as a chaperone of metals such as iron and zinc. Both iron and zinc are thought to play an important role in the aggregation of proteins, such as huntingtin. PBT2 is thought to sequester these metals away from the mutant huntingtin protein and prevent the formation of toxic, oligomeric (multi-subunit) forms of huntingtin.

In the newly published paper, Cherny et al tested PBT2 in both a worm and mouse model of HD. While PBT2 delayed the paralysis commonly observed in the HD worm, it is interesting to note that PBT2 had no apparent effect on the aggregation of the short polyglutamine-containing protein that is expressed in their worm. This suggests that the worm model used here is not ideal for understanding the impact PBT2 has on the different forms of huntingtin.

More striking results were seen when the researchers administered PBT2 to a commonly used mouse model of HD (R6/2 mouse). The R6/2 mice express just a short fragment (3%) of the total human huntingtin protein. When dosed, beginning at just three weeks

of age, PBT2 had a significant impact on motor behavior, body weight, brain weight and survival. The HD mice treated with PBT2 lived 26% longer than the same HD mice treated with a vehicle control.

While these results are certainly encouraging, they are not entirely unexpected. Previous work from Nguyen et al in 2005 showed that clioquinol, a compound very similar to PBT2 in terms of chemical structure and mechanism of action, improved survival in the R6/2 mouse by 20% and had a significant effect on huntingtin aggregation.

Based on these results, as well as previous data suggesting PBT2 may positively impact cognition (thinking)

in Alzheimer's disease, Prana set out to discover if PBT2 will have similar effects in persons with HD. PBT2 is currently being tested in a Phase II clinical study called Reach2HD for people with early to mid-stage HD in Australia and the United States. The Reach2HD trial is now fully recruited. The HD research community will be anxiously awaiting the results from this pivotal study. ■

References:

Nguyen T, Hamby A, Massa SM. Clioquinol down-regulates mutant huntingtin expression in vitro and mitigates pathology in a Huntington's disease mouse model. *Proc Natl Acad Sci U S A*. 2005 Aug 16; 102(33):11840-5.

Cherny RA, Ayton S, Finkelstein DI, Bush AI, McColl G, Massa SM. PBT2 reduces toxicity in a *C. elegans* model of polyQ aggregation and extends lifespan, reduces striatal atrophy and improves motor performance in the R6/2 mouse model of Huntington's disease. *J Huntington's Disease* 2012, 211-219.



Huntingtin Antisense Oligonucleotides (ASOs) Continue to Show Promise as Potential Disease Modifying Therapy for HD

George Yohrling, PhD; HDSA Director of Medical and Scientific Affairs

On March 19th, Drs. Holly Kordasiewicz and Kristina Lemonidis from Isis Pharmaceuticals presented an update to the community on the use of antisense oligonucleotides (ASOs) to treat HD. Isis Pharmaceuticals is a biotechnology company focused on RNA targeting therapeutics. RNA is the message that encodes the synthesis of all proteins. Isis has pioneered antisense technology, and as a result, created a drug discovery and development company with nearly 30 drugs currently in development. They plan to add 3-5 new compounds each year. It is their goal that an ASO candidate to target huntingtin will be identified by the end of 2013.

Antisense therapy is a form of treatment being pursued for many different disorders such as diabetes, cancer, ALS and now HD. The ASOs currently being tested in animal

models of HD are small, single-stranded molecules composed of between 18-20 nucleic acids. The ASOs can be designed to bind to almost any region of RNA. When the ASO binds to the RNA, the RNA is targeted for degradation. The end result of effective ASO binding to RNA is that less protein will be made. This is exactly what Isis has shown in rodent models of HD. When the ASO directed against the huntingtin RNA is administered to HD mouse models, the levels of huntingtin mRNA and protein decrease. In addition to decreases in huntingtin levels, they also observed improvements in motor activity and coordination, lessened anxiety, even a slowing of the loss of brain mass. Another exciting observation is that following a short two week dosing, the huntingtin levels in the mice remain significantly suppressed for up to 12

weeks. This suggests that intermittent treatment may be possible in persons with HD. This is important because the ASOs do not enter the brain via the most common routes of drug administration (oral, intravenous). As of today, intrathecal (spinal cord) injections must be used to bathe the brain in ASOs.

Despite the promising pre-clinical results, testing of anti-huntingtin ASOs in humans will likely not begin until late 2014. Even then, those studies will be Phase I clinical trials in a small number of people (<30) to assess the safety of the drug and to better understand how to dose the drug in Phase II and III clinical trials. While the initiation of these trials cannot come soon enough, this novel approach to silence the huntingtin gene holds great promise. ■

HD Research Webinar Series

In an effort to improve our research communication activities, HDSA recently launched a new HD Research Webinar Series. The goal of these monthly webinars is to provide a regular forum for persons with HD, families and caregivers to hear updates on the latest research findings directly from HD scientists. The series kicked off on February 27, 2013 with a

presentation by Dr. Jamshid Arjomand (CHDI Foundation) on Stem Cells in HD Research and Therapeutics.

Each webinar is approximately 35-45 minutes with a 15-25 minute question and answer period at the end. The entire series of Research Webinars is archived in the "Research" section of the HDSA national website. There you

can find details for any scheduled webinar. For more information, or to recommend a topic for a future Research Webinar, please contact George Yohrling, PhD at gyohrling@hdsa.org. ■

HD Human Biology Research Project

HDSA is proud to announce that in March 2013 a new HD research program was launched to provide financial support for investigator-initiated research directed to better understand the biology of Huntington's disease (HD) as it occurs in humans. We expect that the research will directly impact the lives of persons with HD or help guide future drug development for HD.

HDSA is interested in sponsoring innovative lines of enquiry of human HD data that will shed light on the most proximal biological events caused by the mutant huntingtin gene. All research proposals must incorporate a patient-centric focus in collaboration with an HDSA Center

of Excellence. The HDSA Scientific Advisory Board (SAB) has called internationally for research proposals which specifically address any of the objectives listed below:

- 1. Biomarker discovery/development:**
Reliable and robust readouts to assess efficacy of huntingtin lowering strategies in the clinic are of the utmost importance to the HD community.
- 2. Pathway/Mechanism Studies** using analysis of high-throughput genomic or proteomic data generated from human HD biological samples to uncover proximal biological pathways affected in human HD.

- 3. Outcomes research** to provide evidence about which clinical interventions work best for which types of persons with HD and under what circumstances.

- 4. Analysis of existing clinical trial material.**

- 5. Research to improve of quality of life of persons with HD.**

Awards will be made by the SAB in September 2013 so this important research can commence in the fall. Details about the funded projects will be available on www.hdsa.org so those interested can participate in the research, if applicable. ■

Donald A. King Summer Research Fellowships Awarded

HDSA believes there is an important need to continue to train the next-generation of scientists with research expertise in neurodegenerative disorders, especially Huntington's disease. The goal of the Donald A. King Summer Research Fellowship program is just that - to attract the brightest young scientists into the field of Huntington's disease research all while facilitating meaningful HD research to clarify the biological mechanisms underlying HD pathology.

The Donald A. King Summer Research Fellowships will sponsor HD

investigations that can be conducted over an approximate 10-week summer period. Fellowship recipients, working under the supervision of senior HD scientists, will undertake a project that will be helpful in future HD research while also nurturing a continuing interest in HD.

This year, HDSA received many outstanding proposals from students around the country. Unfortunately, only two recipients could be selected for 2013. We are pleased to announce that the winners of the Donald King Summer Research Fellowship are

Jenny Lin (MIT) and Jolene Luther (University of Iowa).

Jenny Lin is currently a student at MIT, but will be working with Drs. William Yang and Steve Horvath at UCLA on a project entitled "A systems biology approach to analyze Huntington's disease". Jolene Luther is currently a student at the University of Iowa. This summer she will be working with Drs. Jane Paulsen and Hans Johnson at the University of Iowa on a project called "MRI T2 hypointensities as a biomarker in prodromal HD." ■

Clinical Trials Readiness

HDSA has a number of initiatives to help educate HD families about current clinical and observational studies, and to raise awareness about the importance of participating in clinical research. Here are a few ways that YOU can be involved:

HDTrials.org

HDTrials.org is a Huntington's disease clinical trial notification site created by members of the HD Community and managed by HDSA. Registration is simple: provide an email address (which may be anonymous or a pseudonym) and your zip code. When new clinical trials or studies for HD are approved, you will be notified by email about the study and the sites near the zip code you provided. You can register with multiple zip codes if you would like to notify other family members about trials in their area as well. Please register so you will learn about new opportunities to participate in clinical trials and studies as soon as they are available.

NEW! Clinical Trials Locator

A new feature on the HDSA website. To locate clinical studies and trials that are recruiting in your area, go to www.hdsa.org/clinicaltrials.

Clinical Trials Diplomat Program

HDSA Clinical Trials Diplomats are trained volunteers who believe that clinical trials are important to advancing research, treatments, and a cure for HD. Diplomats are usually people who have participated in a clinical or observational study, who are trained by HDSA to provide information and answer questions about clinical trials in small group settings such as support group meetings. To learn more about the Clinical Trials Diplomats program, schedule a Diplomat to speak in your community or to become

a Diplomat, please contact Jane Kogan at jkogan@hdsa.org or by phone at extension 226.

Clinical Trials Presentation and Resources

HDSA has developed two standard presentations for clinical trials: A professional presentation that is given by an HD researcher, clinician, or healthcare professional involved in clinical trials and a Diplomat presentation, which is given by a trained volunteer. HDSA has also developed a pamphlet, entitled *A Brief Guide to the Clinical Trial Process* that gives a detailed overview and covers the most frequently asked questions. Each presentation includes take home material about local clinical and observational studies. ■

New at HDSA: Research Ambassadors & Brain Bank

Research Ambassadors!

Family Members Talking About Research to Other Families

One of the goals in the 2013 Program of Work is to recruit HDSA Research Ambassadors from each HDSA Chapter and Affiliate. These volunteers will work alongside Nancy Rhodes (nrhodes@hdsa.org) and Dr. George Yohrling (gyohrling@hdsa.org), and will be trained to speak about HDSA research efforts at local educational and community events.

The training will include a 3-5 minute research power point presentation, as well as handouts to answer questions that the Research Ambassadors will bring with them to each event. Please reach out to your regional staff contact if you have a Chapter or Affiliate volunteer who is interested in taking on this exciting role. This is a great

opportunity to keep abreast of all HDSA research updates! We are proud to report that we have 12 Research Ambassadors as of March 15th, 2013!

Brain Bank

Making the gift of a brain from a person with HD to research is a wonderful way to contribute to advancing treatments. Scientists from the nation's top research and medical centers request tissue for their investigations. Though one brain can provide many samples, to validate research it is often necessary to supply a variety of brain specimens. At the current time, there is only

one brain bank that accepts HD brains from across the country. If you or your family is interested in learning more about brain donations, please contact the Harvard Brain Tissue Resource Center at 800-BRAIN BANK (800-272-4622) or visit them at www.brainbank.mclean.org. It is strongly advised that families start this process early so brain donations can be accepted. ■



CHDI Conference Continued from page 3

the clinical study in people with HD to begin until 2014.

Three other HD drug programs were highlighted by CHDI scientists. The KMO inhibitor program was presented by Laci Mrzljak (CHDI). CHDI has a potent, peripherally acting KMO inhibitor called CHDI-340246. They believe the drug may work by altering the balance of toxic chemicals in the brain formed by the metabolism of the amino acid, tryptophan. However, to date, the compound showed little, if any, beneficial effects upon chronic

dosing in two different HD mouse models (R6/2 and Q175).

Efforts are also well underway at CHDI to identify small molecule inhibitors of the histone deacetylase (HDAC) class IIa family of enzymes. In this family of proteins, HDAC4 is the most promising. Genetic knock down of HDAC4 in multiple HD mouse models has been shown to positively benefit the mice. This suggests that inhibition of HDAC4 activity may provide a therapeutic benefit to people with HD. Additional studies are required to determine if

targeting the catalytic site of HDAC4 is a viable approach for HD.

Finally, Jonathan Bard (CHDI) presented work on another collaboration with Pfizer to develop trkB agonists as a neuroprotective strategy for HD. The hope is that activation of the trkB receptor will increase brain-derived neurotrophic factor (BDNF), an important protein that is made in the cortex and is lost in HD. The Pfizer collaboration centers around a selective trkB antibody called 38B8. Animal testing of this

Continued on page 12

HDSA Commemorates 20th Anniversary of Discovery of HD Gene

On April 3, Senator Kirsten Gillibrand and HDSA hosted a Research Symposium that commemorated the 20th Anniversary of the discovery of the gene that causes HD with the US Capitol dome serving as the backdrop. The Symposium was the first noteworthy event in a two day advocacy experience that brought advocates from 30 states not only to hear from four of the original “gene hunters” but also to bring their voices to Capitol Hill to advocate for the Huntington’s Disease Parity Act (HR1015/S723).

The Research Symposium was web cast live to HD families across the country thanks to a generous unrestricted educational grant from Lundbeck. The Symposium is now available on the HDSA website for additional viewing.

Francis Collins, MD, PhD, current Director of the National Institutes of Health, served as the keynote speaker and he set the tone for the Symposium by highlighting the significance of the gene discovery and how it led, in turn, to the successful completion of the mapping of the human genome in 2003 which changed the direction of medical research. Indeed, this year marks several significant dates in HD history as it was 30 years ago that the first marker for HD was found by James Gusella, PhD in his lab at Mass General. Twenty years ago, the gene that causes HD was found on the short arm of chromosome 4 by a research collaborative led by Nancy Wexler, PhD which included Drs. Jim Gusella and Marcy MacDonald.

And just a decade ago, the world heralded the completion of the Human Genome Project. Many may recall Dr. Francis Crick, who with James Watson discovered DNA in 1953, coming via satellite to join scientists in Washington as the announcement was made. Indeed these parallels in history were thought provoking and lent an even more historic flavor to the day.

Following Francis Collins was Nancy Wexler, PhD who is credited with assembling the research collaborative that discovered the location of the HD gene. Dr. Wexler gave a poignant talk on how the many families in Venezuela and the United States and Canada contributed their genetic material in a worldwide effort to find the answers to the HD puzzle.

James Gusella followed Dr. Wexler and spoke about what science had learned about HD and genetics over the past 20 years. He talked about the work done in studying the expansion of the gene and how it was determined that only one copy of the mutation was necessary to trigger the disease. Even though a person may have two mutated alleles (one from each parent) only one abnormal copy actually was needed to cause HD. He also touched on how the huntingtin protein is essential to life and while it is not known what the role of wild

(normal) huntingtin may be, scientists do know that the absence of huntingtin leads to death.

Marcy MacDonald completed the Symposium by talking about the future in HD. She spoke about the need to move beyond looking for single targets in treating HD. The current model is expensive and has not proven fruitful. She suggested that researchers may need to consider new



Pictured L-R: James Gusella, PhD; Marcy MacDonald, PhD; Francis Collins, MD, PhD; Louise Vetter; Nancy Wexler, PhD

tools and a new approach that looks at the whole disease process and for interventions that can be introduced before disease onset. She spoke of the challenges in working on HD and the unique nature of the disease as it affects every person differently. She concluded by emphasizing the need to move in new directions in order to effect real change.

A brisk question and answer session completed the Symposium. HDSA would like to thank Senator Debbie Stabenow (MI) and her staff for securing the Symposium venue in the Hart Senate Office Building. ■

HDSA On Capitol Hill

April 4, 2013 – HDSA Advocacy Day actually began at dusk on April 3 when advocates – almost 100 strong – marched to the steps to the U.S. Capitol to represent their loved ones who could not make the trip and those who no longer had a voice to give to HD. After an afternoon spent commemorating the 20th Anniversary of the Discovery of the HD Gene, this symbolic walk to our nation's Capitol was a great way to launch Advocacy Day and announce their intent to make their voices heard by their Congressional representatives.

Thus on a beautiful spring day, HDSA advocates from 30 states and the District of Columbia descended on Capitol Hill to talk with their legislators about the Huntington's Disease Parity Act (HR1015/S723) and to ask for their co-sponsorship. You can read a first person account of Advocacy Day 2013 on page 14.

HDSA kicked off Advocacy Day on the Hill with an intense three hour training session for advocates that was conducted by HDSA's DC based advocacy firm and HDSA's Advocacy Manager, Jane Kogan. Training included tips on how to effectively "make your ask," role playing and break out groups. Advocates were encouraged to practice telling their story beforehand so they were better able to effectively deliver their personal message to their member of Congress. Each advocate received custom tailored leave behind materials for their legislators that explained the need for the Huntington's Disease Parity Act and included essential information such as the estimated number of persons affected by HD in each state.

Every advocate visited multiple Congressional offices over the course of the day and, though most met with their Congressional members health aide or staff person, each was given a warm welcome and encouraged to educate the staffer about HD and why the legislation was so important. As a result of Advocacy Day on the Hill 2013, more than 15 co-sponsors were added to the Huntington's Disease Parity Act thus bringing our total to 26.

But much more needs to be done, not only by those who participated in Advocacy Day on the Hill, but by all members of the HD community. Please see page 13 to see how you can join the Huntington's Disease Parity Act movement. Together, we can make a real difference in the lives of those affected by HD. ■

CHDI Conference Continued from page 10

antibody by direct injection into the brain is planned in both HD mouse and rat models. A secondary approach to elevate BDNF levels is also being pursued by CHDI. They hope to use viruses (adeno-associated viruses) as carriers of the BDNF gene and inject them directly into brains to enhance BDNF in people with HD.

The conference concluded with a session on HD clinical biomarkers. Beth Borowsky (CHDI), who is also a member of the HDSA Scientific Advisory Board, presented important work on a proposed disease-state biomarker for HD called 8OHdG. An earlier paper reported that 8OHdG, a marker of DNA damage, was increased in the blood of people with HD. A

thorough attempt was made to confirm this finding in new blood samples of people with HD. They reported that there are no differences in 8OHdG at any stage of HD. Unfortunately, this suggests that 8OHdG will not be a useful disease-state biomarker.

Tiago Mestre (University of Toronto) presented an update on Enroll-HD and the desire to use it as a platform to facilitate clinical research. Finally, Ken Marek (Institute for Neurodegenerative Disorders) presented exciting clinical data using the PET imaging tracer MNI659 to monitor changes in PDE10A levels in people with HD. While the sample number is low, the preliminary data indicate that binding of the PDE10A PET ligand decrease

while the UHDRS motor scores increase, and therefore could be an important biomarker.

The drug development path from scientific idea to FDA approval is long. While it is recognized that these approvals simply cannot come quickly enough, the HD community should be more optimistic than ever. Years of painstaking research is now moving us closer and closer to the initiation of clinical trials that may alter the course of Huntington's disease. While much work remains, rest assured that progress IS being made by an army of dedicated and passionate HD researchers from around the globe. ■

HDSA Works to Re-Introduce the Huntington's Disease Parity Act of 2013



Overview: The Huntington's Disease Parity Act will compel the Social Security Administration (SSA) to revise the outdated and medically inaccurate guidelines that it uses to determine disability for HD. It would also waive the two year Medicare waiting period for individuals who are disabled by HD. Taken together, these two provisions will make it much easier for people who are disabled by HD to receive Social Security Disability Insurance (SSDI) and Medicare benefits.

As the 112th Congress adjourned at the end of 2012, the Huntington's Disease Parity Act had 143 co-sponsors in the House and 12 in the Senate.

When the new 113th Congress convened on January 3, 2013, it meant that the Huntington's Disease Parity Act had to be re-introduced and the co-sponsors that had formerly signed onto the bill have to now be asked to co-sponsor the bill (with its new House and Senate numbers) once again.

To complicate matters our two champions in the House, Bob Filner (D-CA) and Brian Bilbray (R-CA), did not return to Congress and thus HDSA had to seek two new champions to introduce our bill. We are pleased to announce that in March, Adam Kinzinger (R-IL) and Bob Pascrell

“...these two provisions will make it much easier for people who are disabled by HD to receive Social Security Disability Insurance (SSDI) and Medicare benefits.”

(D-NJ) introduced the Huntington's Disease Parity Act in the US House of Representatives as HR 1015. Thus far the bill has garnered co-sponsorship from 26 House legislators.

In April, Senator Kirsten Gillibrand (D-NY) re-introduced the Huntington's Disease Parity Act in the Senate as S723.

If you have not as yet had a moment to thank Representatives Kinzinger and Pascrell and Senator Gillibrand please go to www.hdsa.org/advocacy to send a thank you to our Congressional champions.

As members of the HD community, the Huntington's Disease Parity Act is something that we can all work toward. Everyone can write a letter, send an email and advocate for this important piece of legislation. To learn more about the bill, download resources or get involved, go to www.hdsa.org/advocacy. For more information please contact Jane Kogan at jkogan@hdsa.org or by phone at extension 226. ■

An Advocate's Story

By: Brenda Porter

I was more than a little hesitant when HDSA reached out to me earlier this year to invite me to attend the HDSA Symposium and Legislative Advocacy Day in Washington, DC. I'm just a mom from Illinois; most days, I feel lucky if I can put something resembling dinner on the table for my daughter. Traveling to Washington, DC to meet with Congressional staff, and persuade them to co-sponsor a bill, was not on my radar. I was familiar with the Huntington's Disease Parity Act and

Symposium, which provided a great overview of the past, present and future of HD research. Then, we took a group picture on the Capitol steps and had a nice dinner.

Advocacy Day started early for everyone, with a comprehensive training provided by HDSA. They reviewed what the legislation would do, the key points to mention in our meetings, and even did a mock interview so we would understand what to expect. We then broke

The first two meetings with Mark Kirk and Richard Durbin's office went smoothly. I was part of a group, and I let others do most of the talking. My last meeting provided me with the opportunity to speak on my own and I met with Margie Almanza, Congressman Schock's Legislative Assistant. Margie listened to my story and said that she is going to reach out to the Ways & Means leadership and let them know that they are hearing from constituents on the bill. Margie

“My first day in DC was a bit of a whirlwind ... We attended the Research Symposium, which provided a great overview of the past, present and future of HD research.”

had already been active in sending emails through the HDSA E-Advocacy system, www.hdsa.org/takeaction, which is a great, super easy way to reach out to your members of Congress.

My New Year's resolution was to be a stronger advocate for HD, so how could I not attend this important two day event? I was scared, I was nervous, I wasn't entirely sure I could meet with anyone and make any kind of sense, but armed with my brother's advice “you can't make it worse than it already is,” I headed to DC.

My first day in DC was a bit of a whirlwind, as I'm sure it was for everyone. We attended the Research

into smaller groups, and I met the people I would attend meetings with throughout the day to review what we had learned and continue to role play. I was in a group with six other wonderful people from Illinois. Armed with packets of information, I left the hotel to attend meetings with staff of my two Senators, Mark Kirk and Richard Durbin. My last meeting was with my Representative, Aaron Schock's office. The meeting with Congressman Schock's office was particularly important because he is a member of the House Ways & Means Committee, which is in charge of Social Security matters.

also said that each time someone reaches out to her, she will be letting the Committee leadership know. I have spent the past week contacting friends and family: asking them to call, email, or write a letter to Aaron Shock's office in support of the HD Parity Act, and I will continue to do what I can to support this important bill. I am honored to have had the opportunity to raise awareness for HD and I look forward to the day when the Parity Act is passed! ■



Advocacy

HDSA Advocacy works to improve the lives of HD families by supporting and promoting legislation and policy initiatives that would benefit the HD community. By working together and speaking in one voice, HDSA advocates have been able to help advance legislation such as the Genetic Information Nondiscrimination Act (GINA) and the Patient Protection and Affordable Care Act. Most recently, HDSA Advocates contributed to the Social Security naming Juvenile Onset HD and adult onset HD as conditions that qualify for their Compassionate Allowance (CAL) Initiative.

HDSA Advocacy is active on Facebook as well as Twitter. The HDSA Advocacy Facebook group now has over 3000 advocates and the main HDSA Facebook page has more than 8000 friends. Most recently, in a 2 week period, over 1000 people shared an HDSA petition to thank the 2 House Champions who introduced the Huntington's Disease Parity Act of 2013 (H.R. 1015). Be sure to add us to your social media network, friend us on Facebook, and follow us on Twitter—just search HDSA to find us and join in the conversation!

HDSA E-Advocacy Center & Advocacy Toolkit

Huntington's Disease Parity Act resources, including copies of the House and Senate bills, background information, and costs, are available for download on the HDSA advocacy website, www.hdsa.org/advocacy. Be a voice for HD by becoming an E-Advocate by going to www.hdsa.org/join and receive email alerts when your voice

can make a crucial difference. HDSA gives you the tools you need to participate!

Huntington's Disease Parity Act of 2013

For more about this important piece of legislation, please see page 13.

Essential Health Benefits

Essential Health Benefits (EHB's) are a collection of health care services that insurance policies, offered through State Health Insurance Marketplaces are required to provide by 2014. States are currently in the process of developing or implementing their health insurance marketplaces, either as a state-based initiative or in partnership with the Federal Government. States that have opted not to create an insurance marketplace will automatically be a part of the Federal marketplace. HDSA is asking advocates to take action to advocate that EHBs promote meaningful mental

health parity, address the coverage gaps that exist in small group plans, and mitigate the potential disparities in coverage among various state packages. Go to www.hdsa.org/ehb or contact Jane Kogan at jkogan@hdsa.org to learn more.

Disability Application Toolkit

Want to learn about the process of applying for Disability for HD? Visit the HDSA online Disability toolkit, found at www.hdsa.org/disability.

The new section contains an overview of the criteria, the process, frequently asked questions, Disability webinars and more. If you have been denied Disability for medical reasons, and are currently in the appeals process, please contact Jane Kogan at jkogan@hdsa.org or at extension 226.

Continued on page 16

Advocacy Continued from page 15

Genetic Information Nondiscrimination Act Resources

HDSA's Genetic Information Nondiscrimination Act (GINA) Toolkit can be found on www.hdsa.org/gina. It features frequently asked questions about what GINA does, GINA and the workplace, GINA and health insurance, case examples, and more! For more information, please contact Jane Kogan at jkogan@hdsa.org or at extension 226.

Coming Soon: Voices of HD

Each month, HDSA will profile an individual who is making a difference by participating in HDSA Advocacy efforts. Learn more at www.hdsa.org/voices and contact Jane Kogan at jkogan@hdsa.org or by phone at extension 226 to be profiled as a part of the section.

Know Your Rights Webinars

HDSA is developing a section of the website to help the community navigate the regulations, laws, and policy that impact individuals affected by HD and their families. These resources will be available on the HDSA website in the fall of 2013.

Workplace Accommodation Information & Resources

HDSA is currently working with the Job Accommodation Network, and surveying the HD community to learn more about workplace accommodation needs for Huntington's disease. Go to <http://www.surveymonkey.com/s/HDSAAcomm> or contact Jane Kogan to share your feedback. ■



HDSA 28th Annual Convention

The 28th Annual HDSA Convention is just around the corner so be sure to register today! This year, HDSA will be visiting sunny Jacksonville FL located just south of the Georgia border on the Atlantic Ocean. The Hyatt Regency Jacksonville is easily accessible from all parts of Florida, Georgia, Alabama and points north. So make it a point to join us for Convention.

This year, our program is better than ever. For the first time, HDSA is providing two keynote addresses – one on Friday morning immediately following the Opening Ceremony and the second on Saturday morning. We have two exceptional speakers: Dr. Marc Holder from the University of British Columbia who will talk with us about happiness and well being in HD, and Dr. Michael Hayden also lately from the University of British Columbia who will discuss strides in HD research.

Also new this year is the HDSA Chapter Awards Luncheon which will take place from 12:30 to 2:00 p.m. on Friday. This luncheon is included in your registration fee so please be sure to join us as we recognize outstanding volunteers from HDSA chapters and affiliates across the country.

Saturday morning, HDSA offers all Convention attendees breakfast to start an exciting day of research and workshops focused on care. The breakfast is also part of your 2013 Convention registration fees.

Friday afternoon workshops will begin at 2:15 p.m. and will be 75 minutes each. Saturday workshops will begin at 1:30 and will be 60 minutes each. Sessions will include topics for those

new to HD, caregivers, youth, young adults, parents of kids with JHD, field leadership and so much more. The Convention program can be found at www.hdsa.org/convention.

Florida residents are cordially invited to an Advocacy Meet and Greet that will be held on Friday, June 21 from 5:30 – 6:30 p.m. HDSA will be inviting members of the US Congress hailing from Florida to meet their constituents. Advocacy training will be held prior to the Meet and Greet so those new to advocacy can learn how to ask their representatives to co-sponsor the Huntington's Disease Parity Act (HR1015/S723). See page 13 for more information on this important piece of legislation.

Completing a full day of activities on Friday will be the NYA Talent Show. This annual event features talented youth and young adults who perform to raise money for scholarships to the next year's HDSA Convention. The Talent Show begins at 7:00 p.m. and the first 100 people in the door will receive a FREE tee shirt and FREE raffle tickets for a 2014 Convention package (2 convention registrations and hotel accommodations). In addition, the NYA offers a day long Silent Auction on Friday in the HDSA Exhibit Hall. The proceeds from this activity also help to fund scholarships for youth. Please be sure to stop by the HDSA Exhibit Hall and make a bid on any one of the outstanding auction items.

To ensure Convention attendees have plenty of time to visit the various vendors in the Exhibit Hall, HDSA is offering an "early bird" opening on Thursday June 20 at 2:00 p.m. The first 100 folks to stop by the Exhibit Hall will



receive a free tee shirt and there will be two FREE raffles for gift certificates to dinner to use on Thursday or Friday night. Raffle winners will be announced on Thursday at 7:00 p.m.

Saturday evening, HDSA welcomes all to the Annual Gala and National Awards Dinner which includes the ever popular candle lighting. The gala is also included in your Convention registration fee. If you would like additional members of your family to attend the gala, tickets are just \$85/person. See information about Convention registration on page 20.

Finally, HDSA is offering a Second Annual Clinical Research Initiative Day which provides family members with the opportunity to participate in small scale studies, surveys, focus groups and other types of research studies on Sunday at the Hyatt Regency Hotel. A list of studies can be found on the HDSA website. Please be sure to pre-register for the studies that are of interest to you. You can also register during the HDSA Convention at the investigators tables in the Exhibit Hall. ■

Ways to Give

There are many ways for you to make a contribution to help HDSA improve the lives of people with Huntington's disease and their families.

- **Make a one-time or recurring Donation or a Tribute/Memorial Gift to honor a friend or relative or the memory of a loved one:** Please visit our website, www.hdsa.org and click on the “Donate” icon at the top of the page. This will take you to a secure page where you can make a **direct online donation** to HDSA.

Or you can use the donation envelope located in this issue.

- **Establish a Family Fund:** Join with friends and relatives and pool your resources to honor your family or remember a loved one and make your donated dollars work harder than you could individually.

For information on how to establish a Family Fund please call 1-800-345-HDSA (4372), extension 235

- **Work Place Giving Matching Gifts:** Your employer or organization may be part of the **HDSA Program**, which can double your donation.

A list of participants is available on our website. Click on the “Donate” icon at the top of the HDSA website, www.hdsa.org, and select “**Matching Gifts**” on the left side of the page to view a

list of companies that participate in our matching gift program. If your employer is not part of this program, we would be happy to help enroll your company or organization.

United Way/Community Health Charities/Combined Federal Campaign:

Giving at work through payroll deductions to support HDSA is simple, and there are many convenient ways to contribute. Check to see if your employer participates in any of these workplace giving programs.

- **Become a Corporate Partner:** Help HDSA in its efforts to improve the lives of people with HD and their families by either making a general donation or donating to support a specific program or service.

Join an event: participate or become a sponsor of the hundreds of HDSA events around the country, such as our Team Hope Walks or Celebration of Hope Galas.

Workplace Giving: encourage employee giving through payroll deductions and show your employees that you support their philanthropic efforts by contributing a company match of their gift.

- **Donate your Vehicle:** call toll free 888-HDSA-151/888-437-2151 or e-mail, at your convenience, donations@charitableautoresources.com to speak to an HDSA Vehicle Donation Representative. Our representative will schedule a pickup that's convenient for you, and provide you with confirmation of your donation.

Or visit our website, www.hdsa.org and click on the “**How to Help**” icon – select “**Other Ways to Give**” on the left side of the page - select “**Vehicle Donation,**” which will take you to a secure page where you can choose to make an **online vehicle donation** to HDSA.

- **HDSA Shop:** Please visit the HDSA Shop by visiting our website, www.hdsa.org. Click on the “**How to Help**” icon and select “**HDSA Shop**” on the left side of the page. This will take you to a secure page where you can purchase merchandise such as a Care2Cure Bracelet or Necklace, amaryllis plant or a golf polo shirt. By purchasing merchandise you are making a difference – and helping us build awareness at the same time. ■

Simple Ways to Support HDSA that can also Benefit You and Your Family

Include HDSA in Your Estate Plans

- Retirement Plans: Designate HDSA to receive part or all what remains of your retirement plan after your death.
- Insurance policies: Designate HDSA to receive all or part of the proceeds of a life insurance policy.
- A Charitable Bequest: Add a charitable bequest for HDSA to your estate plan for a specific amount, a specific property, or for a percentage of the estate.
- Life Income Gifts: Life income gifts such as charitable gift annuities, charitable remainder trusts and charitable lead trusts are creative way to provide income for and your family, reduce estate taxes and contribute to HDSA.
- For information on making a planned gift to HDSA please call 1-800-345-HDSA (4372), extension 235

Donate Appreciated Stock Or Mutual Funds

- Donating stock or mutual funds is an easy way to support HDSA and earn a charitable tax deduction for the full fair market value of the gift while you lower your capital gains taxes.

For information on how to make a stock or mutual fund donation please call 1-800-345-HDSA (4372), extension 235

Donate Through Your IRA

- If you are 70 ½ or older you can make a charitable donation of up to \$100,000 per year from your IRA accounts to HDSA as part of Congress's re-authorized of the American Taxpayer Relief Act without first incurring income taxes on the withdrawal.

If you would like to make an IRA rollover gift for 2013, you need only direct your IRA plan provider to make the distribution to HDSA.

Please call 1-800-345-HDSA (4372), extension 235 for more information or to receive a sample letter of instruction to send to your plan provider.



It's Time for the BMW Sweepstakes

Win a 2013 BMW 128i Coupe or \$25,000 cash

The Huntington's Disease Society of America is pleased to offer an exciting way to support HDSA's efforts to improve the lives of people affected by Huntington's disease and their families – the BMW Sweepstakes!

Tickets are \$100 each. For every two tickets you buy at \$100 each, you will receive a third ticket free. The sweepstakes is limited to just 2,500 tickets so your chances of winning have never been better.

Go online at www.hdsa.org/bmwsweeps to purchase your tickets today! You can also buy tickets at the 28th Annual HDSA Convention. The drawing will be held on June 22nd during the HDSA Annual Awards Dinner and Gala.

Don't wait! Tickets are limited. Act today. You need not be present to WIN!

BMW disclaimer

Official Rules: No purchase obligation or test drive necessary. No portion of sweepstakes ticket purchase is tax-deductible. No responsibility is assumed for lost, late or non-delivered mail. Winners will be selected in a random drawing to be conducted on June 22, 2013. All prizes must be redeemed by September 30, 2013. Sweepstakes open only to licensed drivers who are 21 years of age or older and are residents of the United States (except Puerto Rico). Employees of the Huntington's Disease Society of America and employees of BMW of North America Inc., their retailers, advertising, print and promotion agencies and members of their immediate families are not eligible. Winners will be notified by phone and/or mail. Odds of winning are determined by the number of eligible entries received. Taxes are the sole responsibility of winners. Sweepstakes is subject to all federal, state and local laws and regulations and is void wherever prohibited by law. Entry and acceptance of prize offered constitutes permission to use winner's name, photograph, or other likeness for the purpose of promotion on behalf of the Huntington's Disease Society of America, Inc. unless prohibited by law.



GRAND PRIZE:
2013 BMW 128i Sedan
OR **\$25,000 CASH**

2nd Prize: \$5,000 cash
3rd Prize: \$2,500 cash
4th Prize: \$750 cash
5th Prize: \$500 cash

Convention Registration Information

REGISTRATION TYPE	FULL MAY 1 - MAY 30, 2013	LATE OR ON-SITE JUNE 1 - & LATER 2013
Adult	\$220	\$280
Family (2 or more adults) per person	\$190	\$230
NYA	\$130	\$155
Non-NYA (children 18 and younger)	\$130	\$155
DAY REGISTRATIONS (Gala not included)		
Friday	\$130	\$155
Saturday	\$130	\$155
Convention Gala	\$85	\$100

For more information about the HDSA 28th Annual Convention, please visit our website at: www.hdsa.org/convention or call 800-345-HDSA (4372)

HDSA In Your Community

Join an HDSA Chapter or Affiliate!

Looking to connect with volunteers and make new friends in your community? Volunteer with one of HDSA's 51 Chapters and Affiliates across the country! By volunteering with a Chapter or Affiliate you will connect with other people just like you who support the HDSA mission, and can learn from, support, motivate, and educate one another.

Joining an existing Chapter or Affiliate will add value to the work volunteers have already begun in their communities. Your support and dedication will enhance the impact and relationships that many volunteers have spent years building through cultivation and networking. Join the board, head up a committee, sign up for a Team Hope Walk or another existing event, join educational or advocacy activities—the possibilities are endless! Becoming a part of your local Chapter or Affiliate gives you a voice in your community and beyond.

You are given an opportunity to speak about HDSA to the media, to share your inspirations and to create a more impactful network.

If you live in an area without HDSA resources, consider creating a new HDSA Affiliate! Starting a new

Affiliate within our organization is an exciting and rewarding adventure. You will meet others who share your views and passions, develop new friendships, and foster a nurturing community. Building a new Affiliate takes energy, passion, dedication, and commitment. HDSA looks for community leaders who possess these virtues, as well as leadership and organizational skills. We can assure you that the work and time you place into this endeavor is nothing short of rewarding. We are looking for Volunteer Leaders who are willing to bring change and support to their area. If you feel you are a good candidate for a leadership role, reach out to HDSA today and we will provide the outreach and tools to help make your Affiliate dream a reality.

100 Team Hope Walks and Growing!

The Team Hope Walk Program is our largest national grassroots event. Thousands of families, friends, co-workers, neighbors and communities walk together each year to support

HDSA's fight to improve the lives of people affected by HD and their families. Currently, Team Hope takes place in more than 100 cities nationwide. Since its inception in 2007, Team Hope has raised close to \$4 million, thanks to the support and tireless commitment from walkers just like you. From becoming a national or local sponsor to forming a team to volunteering, you too can help make a difference in so many lives. Join an existing Team Hope Walk or coordinate a new one in your community, and help us provide help for today, hope for tomorrow one step at a time.

For more information on volunteering with a Chapter or Affiliate, starting a new Affiliate, or joining TEAM HOPE, please contact Nancy Rhodes, Director of Field Development and Operations, at (212) 242-1968 x234 or via e-mail at nrhodes@hdsa.org. ■



Communications Strategies for Those with Huntington's Disease

By Jeff Searle, MS, Department of Hearing and Speech, Kansas University Medical Center

Huntington's disease affects many skills which are necessary for effective communication. Disturbances of speech production, or the mechanics of speaking, include:

- impaired respiration for speaking
- hoarse and strained vocal quality
- inappropriate loudness and rate
- imprecise articulation

Cognitive language skills, or the thought process we need in order to formulate what we want to say, are also affected by the disease. Difficulties include such things as:

- initiating conversations
- spontaneity in communication
- putting thoughts into words
- a reduced number of words available to the person
- limited ability to respond within a conversation
- understanding complex information
- slow response time
- impaired skills in reading and writing, ranging from physical to comprehension difficulties.

Persons with HD also have problems:

- learning new information and new skills

- reduced short term memory skills
- poor attention and concentration abilities
- lack of ability to organize, reason and problem solve.

Because Huntington's disease is a degenerative brain disorder, these difficulties occur randomly as the disease progresses. This unpredictability can be extremely frustrating for the person. For example, they may make a request clearly at one moment but have tremendous difficulty articulating the same request only a few seconds later.

As a caregiver, it is important to understand that the person has no control over this; they are not being stubborn or making a conscious choice not to remember or repeat a task. It is the degenerative unpredictable nature of Huntington's disease that is the root of the problem.

Strategies for Communication

The principal guideline in achieving effective communication is for the listener to consciously **accept the responsibility** for the conversation exchange. Because of the many deficits which frequently occur, the person with HD often needs to rely on external cues and guidance from the listener to achieve communication. It

is also important to realize that while you accept the responsibility for the exchange, you must not control the conversation.

When working with a person with Huntington's disease who has communication difficulties, it is recommended that you speak with a speech/language pathologist to help you implement, monitor and adjust the following strategies:

- Speak Slowly
- Repeat or Re-phrase the message
- Simplify the Message-Offer Cues
- Try to Gain Topic Knowledge
- Spell Part of the Message
- Use Yes/No Questions
- Refocus Person on Topic
- Monitor Behavior and Provide Feedback

Some additional strategies are:

- Provide a Predictable Environment
- Allow Adequate Time for Communication
- Reduce Distractions
- Use Communication Aids
- Monitor Listening and Attending Ability
- Provide and Ask for Feedback



“It is also important to realize that while you accept the responsibility for the exchange, you must not control the conversation.”

Strategies for Communication During Advanced Stages of Disease

An individual in the advanced stages of Huntington's disease may well be non-verbal, yet interacting with another human being may be the only connection the person has with his/her environment. One of the biggest mistakes you can make is to assume that because a person has lost the ability to speak they have also lost the ability to understand. We know that persons in the later stages of the disease have a high

degree of comprehension of ordinary conversation, therefore, maintaining communication at any level during this stage is extremely important. The following strategies will help you in your efforts:

- Get to Know the Person
- Keep the Person Informed
- Be Innovative & Perceptive
- Establish a Routine for Care
- Recognize and Use Gestures and Facial Expressions as Communication Tools

• Inform Others of Your Strategies

Individuals in the advanced stages of the HD still have much to share. It is the caregivers' responsibility to see that non-verbal persons remain in touch with their environment and those around them. It can make an enormous difference to that person's quality of life if they feel connected and affirmed as an individual. ■

Educational Resources for Families

HDSA provides education to HD families, as well as professionals that work with individuals affected by HD, through the HDSA Annual Convention and field-based educational events, as well as publications, programs, and webinars. Most resources, including presentations from past HDSA Conventions, are available on the HDSA website, www.hdsa.org.

A Physician's Guide to the Management of Huntington's Disease: Third Edition

One of the most important resources for the HD community, *A Physician's Guide* was updated in 2011. Thanks to a generous educational grant from Lundbeck, HDSA has been able to provide a complimentary copy of *A Physician's Guide* to every HD family as well as to the primary care physician and neurologist of their loved one with HD. You can now download a copy of *A Physician's Guide* by going to www.hdsa.org/publications. Call Anita Mark-Paul at 800-345-4372 extension 219 or email Anita at amarkpaul@hdsa.org for your copy of the Guide.

Law Enforcement Education Toolkits: Resources to educate law enforcement & caregivers

Caregivers and social workers have asked for a resource that they can use to educate local law enforcement about HD, and to learn more about how interactions with law enforcement can be managed. To address this need, HDSA has developed a tool kit for local law enforcement personnel and first responders that provides an understanding of HD, reasons why a person may behave seemingly inappropriately or aggressively, and ways to help foster a better understanding in the community and avert disease related confrontations.

Additionally, HDSA has developed a toolkit for caregivers that focuses on working with law enforcement and includes resources that can be customized to provide local emergency contact information. To order a copy of the Toolkits, please contact Anita Mark Paul at amarkpaul@hdsa.org. For more information, or to request multiple Law Enforcement toolkits for a training session, please contact Jane Kogan, Education & Advocacy Manager at jkogan@hdsa.org or at 800-345-4372 extension 226.

Updated & New Family Guide Series Publications

HDSA's *Family Guide Series* provides a comprehensive overview of the causes, symptoms, applicable treatments, and care currently available for people with HD and their families as they navigate the path to understanding and managing HD. Here are the newest publications in the series:

- ***Talking with Kids about Huntington's Disease:*** This pamphlet provides an introduction to talking about Huntington's disease with youth, teen and young adults.
- ***Caregivers' Guide to Communicating with Healthcare Providers:*** This pamphlet provides an overview of how caregivers can present what they see and experience while caregiving to their loved one's doctors in an effective way.

- ***Physical & Occupational Therapy for Huntington's Disease:***

This pamphlet provides helpful information and resources for people in all stages of HD and was updated to include new trends in HD care.

- ***Nutrition & Huntington's Disease:*** This pamphlet discusses the nutritional needs in early, middle and late stage HD and provides hints for increasing calorie intake and recipes.

All of the *Family Guide Series* titles are available for download at www.hdsa.org/publications. Contact Anita Mark-Paul at amarkpaul@hdsa.org or at extension 219 to order printed copies of these publications.

Coming Soon: Advance Directives & HD Family Guide:

This pamphlet, developed with funding from the Bess Spiva Timmons Foundation, will offer individuals with HD, their caregivers, and individuals who are at-risk insights about advanced care planning. The pamphlet will be available in June of 2013. Contact Anita Mark Paul at amarkpaul@hdsa.org or at extension 219 to pre-order.

Coming Soon: Talking with Kids about HD Handbook

This handbook, developed with funding from the American Legion Child Welfare Foundation, will help guide parents and adults through the issues and challenges that come

with trying to explain Huntington's disease to a child or young adult who does not have HD, and who is at-risk of inheriting the disease. The handbook will also provide examples of resources that exist to help parents navigate the path of discussing HD with their children. The Handbook will be available in June of 2013. Contact Anita Mark Paul at amarkpaul@hdsa.org or at extension 219 to pre-order.

Spanish Language Translations of HDSA Family Guide Series & Fast Facts

Many of HDSA's *Family Guide Series* publications, as well as *Fast Facts About HD*, are available in Spanish. To download a publication, please go to www.hdsa.org/publications. Contact Anita Mark-Paul at amarkpaul@hdsa.org or at extension 219 to order printed copies of these publications. Translation of HDSA publications was made possible by a generous gift from Lundbeck.

JHD Resources

HDSA has created several publications that can assist parents, healthcare professionals and school administrators to cope with the ever changing needs of the young person affected by Juvenile Onset HD. Unless otherwise specified, publications are available for download, and one print copy is available free of charge. The following publications about Juvenile Onset HD are available from HDSA:

- **Family Guide Series to Juvenile onset HD:** this 20 page brochure provides an overview of the juvenile version of HD.
- **The Juvenile HD Handbook:** this 87 page guide for families and caregivers was revised in 2007 and can assist in the diagnosis and care of a child affected by JHD.
- **The Juvenile HD Resource Directory:** this 78 page state specific guide also contains a CD loaded with additional resources for families.

- **Juvenile Huntington's Disease and the School Experience:** Education and the Child Affected by Juvenile Onset HD is a 150 page handbook designed to educate school administrators, teachers and classmates about JHD. A Resource CD is included at the back. Note: this publication is only available in print.

You can download these publications at www.hdsa.org/publications. Contact Anita Mark-Paul at amarkpaul@hdsa.org or at extension 219 to order printed copies of these publications.

Speaker's Bureau

HDSA's Speakers Bureau provides interesting and qualified speakers on a wide range of topics for educational events at HDSA chapters, affiliates, regions and support groups. For more information, or to learn about scheduling a speaker at your event, please contact Jane Kogan at jkogan@hdsa.org or at extension 226.

Caregiver's Corner and Lunch and Learn Web Based Workshops

HDSA's Caregiver's Corner webinars are web-based workshops and seminars that are geared to topics of interest for caregivers. Lunch & Learn webinars cover topics that are not strictly germane to HD caregivers. Each webinar is 60 minutes with a presentation and question/answer session. The entire series of Caregiver's Corner and Lunch and Learn webinars are archived on the HDSA national web site. Go to www.hdsa.org/ccorner or www.hdsa.org/ll to browse through the archived library. ■



HDSA Community Resources

HDSA website: www.hdsa.org

The HDSA national website continues to evolve to serve the needs of everyone affected by HD. In addition to expanded lists of resources (searchable by state) and enlarged research, clinical trials, advocacy and community resource sections, the HDSA Forum, constituency extranets and new sections such as “HD and HDSA in the News” have been added.

Chapter and Affiliate Websites

This year, HDSA created new websites for all 51 Chapters and Affiliates. These sites focus on the work being done in the Chapter/Affiliate’s area, and provide lists of local resources (medical and non-medical) and other information of importance to the HD community. To find your local chapter/affiliate website, go to www.hdsa.org and click on “Find HDSA in Your Community.” Then click on your state.

HDSA Helpline

HDSA is here to help you. Do you have a question about HD? Do you need information about local services that HDSA provides? Call Seth Meyer, LMSW at the new HDSA Helpline number: 888-HDSA-506. The HDSA helpline is manned during business hours (EST). You can also email Seth at smeyer@hdsa.org to be connected with your local support groups, social workers, and resources that are appropriate for you and your loved one. If you are calling for anything other than assistance with resources and referrals please call HDSA’s general toll free number at 800-345-HDSA (4372).

HDSA Forum

To supplement the 170+ support groups HDSA hosts throughout the US, we’ve added an online Forum to the national website. The Forum is a place for people to share their thoughts, concerns, best practices and offer support to other members of the HD community. Participants can start new topic threads and comment on existing ones. It’s a great venue for those who want to share their experiences online, and for others seeking guidance or reassurance from someone who has experienced HD. The HDSA Forum is open to anyone through a simple registration process on the site.

We Are HDSA!

We Are HDSA! has proved to be widely popular, providing information on a variety of topics including caregiver issues, family planning, clinical trials, and advocacy. Don’t miss an issue. Sign up to receive We Are HDSA! via email by contacting Seth Meyer at smeyer@hdsa.org. Also remember, all issues of We Are HDSA! can also be found on the website.

National Youth Alliance (NYA)

At the 2000 HDSA Annual Convention a number of young people from HD families gathered to discuss the unique issues created by growing up in a family with HD. At this meeting the HDSA National Youth Alliance (NYA) was formed. The NYA has grown to more than 200 members who meet virtually throughout the year, and in person at the HDSA Annual Convention.

Since 2006, the group has met on NYA Day (the Thursday before the HDSA Convention begins) as a special day for youth and young adults. Through annual NYA fundraising efforts, more than 30 NYA members are able to attend both NYA Day and the HDSA Convention each year. The NYA provides youth and young adults (ages 9-29) with support as well as the knowledge they need as they face the challenges of HD. If you are interested in joining the NYA, contact Mynelly Perez at mperez@hdsa.org.

HD Buzz

Research into the genetic, molecular and biological causes of Huntington’s disease, and the quest for identifying therapies is extremely complicated. To foster better understanding of this cutting edge scientific work by the families who make up the HD community, Drs. Jeff Carroll and Ed Wild created “HDBuzz.net.” On HDBuzz, leading HD scientists explain their work in basic language. Topics range from new discoveries in the basic science of HD, to developments of potential new therapies and breakthroughs in other diseases that may be applicable to HD. HDSA has been a supporter of HDBuzz since its inception and features all HDBuzz articles on the national HDSA website. ■

The Role of an HDSA Social Worker

Living with HD can be difficult. It can take a lot of time and resources to find appropriate care and support. To help, HDSA has 40 social workers around the country available to guide you along the HD path. Our social workers understand Huntington's disease and the special needs of people with HD and their families. HDSA social workers are available to answer questions and point you in the right direction, whether you have HD, your loved one has HD, you are an at-risk family member or you are a professional working with HD.

Below are just a few of the subjects HDSA social workers can help with:

- Provide information about Long Term Care facilities
- Present in service trainings for facilities and professional organizations
- Facilitate support groups
- Manage a local HDSA Helpline
- Take part in national workgroups
- Identify and expand local resources
- Work with local agencies to help build knowledge of HD
- Collaborate with clinics and other agencies as needed
- Assist in planning education days



HDSA social workers are an invaluable resource for people with HD, families, and professionals. To find your local HDSA social worker, please visit www.hdsa.org/sw or contact the HDSA National Helpline via phone at (888) HDSA-506 or via e-mail at hdsainfo@hdsa.org ■

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The appearance of advertising, or the mention of commercial products available for sale in articles published in this publication, is not an HDSA, Inc. guarantee or endorsement of the product or the claims made by the manufacturer. Statements and opinions expressed in articles are not necessarily those of HDSA, Inc.

HDSA, Inc is a national not-for-profit organization founded in 1986 to help individuals with Huntington's disease and their families.

The Society is a member of the National Health Council, the International Huntington Association, the National Organization of Rare Diseases, Community Health Charities and the National Voluntary Health Agencies.

The Huntington's Disease Society of America meets all nine standards of the National Charities Information Bureau.

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HD Milestones

- 1953:** James Watson and Francis Crick are credited with identifying the double helix of DNA
- 1967:** Woody Guthrie succumbs to complications associated with HD
- 1968:** Marjorie Guthrie forms the Committee to Combat HD (precursor of HDSA)
- 1977:** Congressional Commission to Control HD and its consequences releases its findings. First funding for HD research and care
- 1983:** Marker for HD found by James Gusella, PhD in lab at Mass General
- 1983:** Marjorie Guthrie dies just a few months before discovery of marker is announced
- 1993:** Gene that causes HD found on short arm of chromosome 4 by HD Research Collaborative
- 2003:** Francis Collins, MD, PhD announces completion of mapping of human genome at ceremony in Washington DC.
- 2010:** Huntington's Disease Parity Act introduced in 111th Congress
- 2012:** Both adult and juvenile onset HD are added to Social Security Administration list of compassionate allowances
- 2013:** 20th Anniversary commemorating discovery of HD gene held in Washington, DC
10th Anniversary of mapping of human genome
50th Anniversary of discovery of DNA by Watson and Crick

HDSA in Washington

April 3 & 4, 2013



Louise Vetter



Nancy Wexler, PhD



Marcy MacDonald, PhD



Francis Collins, MD, PhD



James Gusella, PhD



Advocacy Training



Attending the Research Symposium



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