We Are HDSA!

Huntington's Disease Society of America

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Social Security Administration Announces Compassionate Allowance Designation for Juvenile Onset Huntington's Disease

Time to qualify for SSD benefits reduced from months to days

By Jane Kogan

On April 11, Social Security Commissioner, Michael J. Astrue, announced that the Social Security Administration (SSA) was adding Juvenile Onset Huntington's Disease (JHD) as a Compassionate Allowance (CAL) condition. CAL cases receive expedited processing within the context of the existing disability determination process. This means that an individual diagnosed with JHD can now get their Disability application approved much more quickly – sometimes within days.

Medical conditions on the Compassionate Allowances list are so devastating that they qualify for Social Security benefits based on minimal objective medical information. Currently, only 130 conditions are included in SSA's Compassionate Allowances list. The Juvenile Onset Huntington's Disease Compassionate Allowance Status will begin in August of 2012.

This is an important victory for families facing Juvenile Onset Huntington's Disease and HDSA is proud to have led this advocacy effort. HDSA has maintained a dialogue with SSA since 2004 to streamline the Disability process for HD. Since the inception of the Compassionate Allowances program in 2007, HDSA has been advocating for a CAL designation for HD through letters, testimony at hearings, and face-to-face meetings. Here is a brief history of HDSA's outreach to the Social Security Administration with regard to Compassionate Allowances:

December, 2007: HDSA submitted written testimony for the Compassionate Allowance hearing on Rare Diseases.

October, 2008: SSA released a list of conditions included in their Compassionate Allowances program. HD was not included on this list. HDSA contacted SSA in writing and phone inquiring why HD was not included.

November, 2009: HDSA submitted testimony to the House Ways and Means Social Security Subcommittee hearing on clearing the disability backlog.

July, 2010: HDSA submitted a statement for the record at a Senate Health, Education, Labor & Pensions (HELP) Committee hearing on treating rare and neglected pediatric diseases.

July, 2011: SSA added diseases to the CAL program. HD was not included on this list. HDSA wrote a letter urging SSA to add HD to the list of CAL conditions.

Most recently, last November, HDSA met with Commissioner Astrue and SSA senior officials to advocate for the inclusion of HD and Juvenile onset HD in CAL. We will continue our dialogue with SSA until adult-onset Huntington's disease is added as a CAL condition.

HDSA has a Disability toolkit, located on the HDSA website at **www.hdsa.org/disability** that includes three training webinars, detailed information about the disability process, application strategies, and answers to the most frequently asked questions about applying for Disability with Huntington's disease.

Did you know that some individuals with adult-onset HD may already qualify for CAL?

Go to **www.hdsa.org/disability** to learn more about the Compassionate Allowance designation for Mixed Dementia.

If your loved one has been denied Social Security Disability (SSDI or SSI), HDSA may be able to help: Please contact Jane Kogan at **jkogan@hdsa.org** if your loved one fits the SSA eligibility criteria for Social Security Disability, but the application was denied by Disability Determination Services.

HDSA Clinical Trials Diplomat Program:

The HD gene was found because hundreds of HD family members participated in research. Now there is a critical need for volunteers to participate in clinical and observational studies so researchers can learn if treatments are safe and effective. HDSA Clinical Trials Diplomats are HD family members who can share a personal perspective on clinical trials participation. This summer, invite an HDSA Diplomat to speak at your event or your support group meeting! For more information about the Diplomat program or to learn about scheduling a Diplomat for your support group, contact Seth J. Meyer at **smeyer@hdsa.org.**

A Physician's Guide:

Thanks to a generous educational grant from Lundbeck, HDSA is able to provide a complimentary copy of *A Physician's Guide* to every HD Family when they send or call the HDSA national office with the name, address, and phone number of both the neurologist and primary care provider seen by your loved one with HD.

Upon receipt, HDSA will send one free copy of the all new *A Physician's Guide* to you as well as a copy to your neurologist and primary care provider.

Call Anita Mark-Paul at **800-345-4372 ext. 219** or e-mail Anita at **amarkpaul@hdsa.org** to recieve your free copy!

Researchers Meet in Palm Springs to Discuss Advances in HD Pipeline

By Fred Taubman, Director of Marketing Development and Communications
(Adapted from The Marker, Spring 2012. For the full article, please visit ***www.hdsa.org****.)*

The theme of the recent CHDI Annual Conference could have been "The Big Picture." There were many big ideas discussed; demonstrations of promising movements of several targeted approaches progressing through the drug discovery pipeline, discussions of the utilization of multiple approaches for a successful HD onset-slowing/preventing regimen, and the introduction of Systems Biology – looking at the total big picture – to the HD world.

More than 250 scientists from around the world, representing academic institutions, hospitals, big pharmaceutical and biotech companies, gathered in Palm Springs for four days of symposia, workshops, and networking aimed at producing collaborations that would accelerate the advance of HD science and the progress of potential therapies for those affected by HD.

Session One

We know that you will find the huntingtin protein in almost every cell of the human body. Therefore, a relatively new approach to medicine, Systems Biology, attempts to understand the disease complexity by examining and collecting data from all of the dynamic biological networks and molecular interactions in the body. This can involve assembling huge data sets of biological information, such as DNA, RNA, proteins, and integrating this data into models of health and disease. Lee Hood, MD, PhD, is the inventor of Systems Biology and explained how this approach would work in HD drug discovery, including the stratification of disease into various subtypes and the creation of new blood markers that will allow the evaluation of the effect of potential drug therapies on the different organs in the body.

Jim Gusella's, PhD, presentation focused on work he and his colleagues are pursuing with another large body of data; genetic data gathered from thousands of participants in PREDICT and REGISTRY studies. While it's common knowledge that HD is caused by a mutant huntingtin protein, the age of motor onset and the severity of symptoms are obviously modified by other factors. That's why two people with the same CAG repeat may become symptomatic at ages ten or twenty years apart. Gusella is attempting to identify these genetic modifiers and then determine how they affect the pathogenic processes, such as gene expression, microRNA expressions, chromatin modification, protein expression, or other processes – with the hope that the findings yield both therapeutic targets and markers to measure their effectiveness.

Session Two

Session two of the conference focused on progress in the areas of translational and post-translational regulation of huntingtin – how the huntingtin protein may change its function or structure by combining with other biochemical function groups. The objective of most of the work on post-translational modification of huntingtin revolves around identifying the modification that would affect the toxicity of the protein to negate its effects.

Dmitri Krainc, MD, PhD, reported on his lab's work utilizing acetylation to promote cellular clearance of mutant huntingtin in the autophagy process. He explained how acetylation can also affect phosphorylation, and likely has an impact on metabolism, protein stability and aging, mRNA function, splicing and other processes.

Session Three

Session three featured what may be the most provocative and promising approach to HD therapeutics – silencing or lowering/eliminating the production of the mutant huntingtin protein that causes the disease. The HD community has been hearing about this work for several years now, and significant progress has been made moving this therapeutic targeting down the pipeline toward human trials.

Beverly Davidson, PhD, presented the work her team at the University of Iowa has done using siRNA (small interfering RNA) and mRNA (messenger RNA) to achieve significant huntingtin-lowering results. It appeared that a 60% reduction in mutant

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Researchers meet **Discuss HD Pipeline** (continued)

HDSA Video Contest: Congratulations Kathleen Samulski!

Congratulations to Kathleen for producing the winning video! Watch Kathleen's video and the other submissions to the HDSA Advocacy Video Contest at **www.hdsa.org/advvideo.**

Advocacy:

Did you know that HDSA has an Advocacy Toolkit? Go to **www.hdsa.org/advocacy** to download resources to help you advocate for the Huntington's Disease Parity Act (S. 648/H.R. 718)

Upcoming HDSA Webinars

June:

HDSA Caregiver's Corner: Law Enforcement Toolkit II: Educating Law Enforcement

July:

HDSA Lunch & Learn: Preparing for the Unknown

To register for the above webinars, please contact Jane Kogan at **jkogan@hdsa.org**. You can view past webinars on our website at **www.hdsa.org** under "Caregiver's Corner" and "Lunch and Learn" in the "Living with HD" section.

HDSA E-mail List:

Want to get more information about HDSA events and activities? Visit the HDSA website at **www.hdsa.org** to sign up!

huntingtin created positive results in mice. Davidson also described work done to develop refined miRNA (micro RNA) that would eliminate potential off-targeting (binding with the wrong proteins). Testing of these treatments in non-human primates suggest that human clinical trials are in the foreseeable future.

Isis Pharmaceuticals has been working on a gene silencing approach that utilizes anti-sense oligonucleotides (ASOs are single strands of DNA that are designed to bind to a specific protein) as the delivery mechanism. This is placed in the cerebro-spinal fluid through an intrathecal pump. Isis is also investigating a second approach, targeting single nucleotide polymorphisms, and a third approach that targets the CAG repeat sequence itself.

Bill Kaemmerer, PhD, gave an update on the other major gene silencing initiative, the collaboration between his company, Alnylam, and Medronics. They are using siRNA to suppress both mutant and wild-type huntingtin.

Session Four

To allow attendees to review the 90+ posters, and speak with their presenters, Session Four of this year's CHDI Conference was dedicated to just that. A committee also judges the posters and selects the top three to make a formal presentation to the entire meeting. Jeff Carroll, PhD, was announced as the Poster Award Winner in the closing ceremonies.

One of the most interesting elements of the CHDI meeting is the featured speaker who closes day two of the conference. This year's speaker was Ann Graybiel, PhD, from MIT. Dr. Graybiel is a widely honored and respected scientist who was introduced as a "mentor" and an "inspiration" by many of the assembled HD scientists. She spoke about her work with cortico-basal ganglia networks and how the field of systems neuroscience could be of benefit to the HD field. The basal ganglia – nuclei at the base of the cerebrum – are deeply involved in a large range of function, including motor symptoms, as they connect with the frontal executive areas and prefrontal cortex.

Session Five & Six

Session Five addressed small molecule drug discovery for HD. Christopher Schmidt, PhD, spoke about Pfizer's approach, utilizing phosphodiesterase (PDE) inhibitors to affect striatal gene transcription. PDEs are enzymes that can break bonds involved in the structure of the DNA "backbone." PDE inhibitors have been shown to prevent or reverse elements of cortico-striatal dysfunctioning in transgenic models of HD.

Sarah Tabrizi, MD, PhD, presented the findings of TRACK-HD and announced the beginning of Track-On HD, a follow up study. Track-On HD will continue the work from TRACK and further refine these pre-manifest biomarkers, as well as hopefully identity the re-routing of signaling in the brain to compensate for early striatal loss.

This type of measurable pre-manifest evidence from the TRACK study was part of the reason for the next presentation by Mark Guttman, MD. His was a call to change the view of HD diagnosis, from a single event to a cascade of events. With the recognition that many people with HD may present cognitive or psychological symptoms years before motor symptom onset, the traditional diagnosis of HD from motor symptoms alone has become a point of debate among researchers, clinicians, and HD family members.

Robert Pacifi closed the conference with a reminder that the key to success, as demonstrated by Maestro Roger Nierenberg in the opening session, was listening carefully to your colleagues, and taking the act of collaboration very seriously. Many of the Conference presentations had demonstrated the unique collaborative nature of the HD scientific community, which were amplified, and will be expanded upon from the interactions at CHDI in 2012.

For more information and updates on research and clinical trials, please visit the HDSA website at **www.hdsa.org/research**.

HDSA Information

New Family Guide Series Publication: Talking With Kids about HD

HDSA now has a Family Guide Series publication that will help parents start a conversation about HD in the family. You can order the publication by contacting Anita Mark-Paul at **amarkpaul@hdsa.org** or by phone at **800-345-HDSA, extension 219.**

Share Your Feedback:

Would you like to receive *We Are HDSA!* via e-mail? Is there an article you would like to see in *We Are HDSA!*? Is there a topic you would like to see discussed? Please send all feedback concerning *We Are HDSA!* to Seth J. Meyer at **smeyer@hdsa.org** with the subject line "We Are HDSA!"

New Support Group Application:

Are you interested in starting a support group in your area? Take the first step by filling out the support group application, found at **www.hdsa.org/sgapp.** If you have any questions about starting a support group or support groups in your area, please contact Seth J. Meyer at **smeyer@hdsa.org.**

Live Videostreaming of the 27th HDSA Annual Convention:

Thanks to a generous grant from Lundbeck, HDSA will be providing live videostreaming of the Opening Ceremony, the Research Keynote, and Clinical Trials Showcase, and a selections of workshops so you can participate from your hope, or organize your own "satellite convention." To view the live sessions, point your browser to **www.ustream.tv/channel/hdsa2012**. Videos of these sessions will be available on the HDSA national website June 25, 2012. For more information, please visit **www.hdsa.org/convention**.

Youth Support Growing Around the World!

By B.J. Viau

National Youth Alliance (NYA)

What is the NYA?

The NYA (National Youth Alliance), which formed over 10 years ago as a youth gathering, has turned into a network of young people in the U.S. that provides support and guidance for each other. There are over 400 members of the NYA around the country and the group is managed by a small committee of volunteers.

What does the NYA do?

The NYA strives to offer support and education to young people while encouraging them to get involved in their local HD community. The group meets annually at the HDSA Annual Convention for a weekend of youth activities including a full day dedicated to young people on the Thursday before the convention. NYA provides scholarships to attend the HDSA Convention and, in 2012, there were over 30 scholarships awarded.

How do you stay up to date with the NYA?

The volunteer committee does its best to keep everyone updated about NYA activities through its website, **hdsa.org/nya**, and through a Facebook group that you can find by searching "NYA Huntington's Disease". To be an official member of the NYA, you can sign up on the NYA section of the HDSA website. For more information about the NYA, please contact Mynelly Perez, Senior Manager of Communications and Event Marketing, at **mperez@hdsa.org**. The NYA is open to all young people who are interested in meeting other young people affected by HD.

Huntington's Disease Youth Organization (HDYO)

What is HDYO?

HDYO (Huntington's Disease Youth Organization) is a new organization created in 2012 to specifically support young people around the world who are affected by HD. HDYO has received financial support from many international HD associations, including HDSA, to create and publish youth specific support opportunities. HDYO is run by a group of young people from around the world who are all from affected HD families.

What does HDYO do?

HDYO is used as another tool to help support, educate and motivate young people through the tough times HD brings. The launch of **www.HDYO.org** is the first of many tools to launch and offer this support. HDYO does not take over other youth groups like the NYA, but looks to partner with and supplement these groups to provide more support where it is needed. For more information on how HDYO can help you in your community, please email **info@hdyo.org**.

What is available on the HDYO website?

The website has specific sections geared towards children, teens, young adults, parents, Juvenile HD, and professionals. Sections on the website have been written by young people and edited by HD professionals to provide a safe and effective way to discuss certain issues. Topics such as caring for a parent, talking to friends, genetic testing, relationships or talking to children are all available in English and will soon be translated into 10+ languages for an international audience. Also on the site are videos, research opportunities and a forum to connect with others. HDYO will continue to update the website and add new content as it is requested.

If you are a young person from an HD Family, a parent of affected children or a health care professional, please check out these two groups and help spread the word to others where support is needed. Young people are the future faces of HD and we want to make sure we support them when they are young, educate them while they grow up and motivate them to make a difference in the battle against Huntington's disease!