Diverse group says ‘We are HDSA’

Volunteers share story of involvement at HDSA National Convention

A blogger, an advocate and a lumberjill.

What do they have in common? They’re all volunteers for the HDSA, outspoken ambassadors for the HD community and members of the larger HD family that met in Las Vegas June 8-10 for the 2012 HDSA National Convention.

The three told their stories of HD involvement during the convention’s opening ceremonies. Ken Serbin of San Diego told how he found the courage to use his real name as a speaker and popular HD blogger so he could have a bigger impact. Ted Krull of Orange County, Calif., talked about making the most out of life with his daughter Emily, who was diagnosed with HD at age 18. Emily’s spirit and desire to educate people about HD inspired Krull to continue to be an advocate in the HD community after Emily passed away in 2009.

And Shana Martin of Madison, Wis., a world-champion lumberjill athlete and prominent spokeswoman for HDSA, talked about her mother, who has been suffering from HD for 30 years. She described her HDSA family, who always understands what she is going through or why she is feeling scared. HDSA gave Martin both a life purpose in volunteering with the HDSA National Youth Alliance and Wisconsin chapter, she said, and the family she values so much.

“Not many people are blessed enough to have a purpose in life,” Martin said. “And that is what I got with Huntington’s disease and you guys in my family. I was surrounded by people who understood me… and I was no longer alone.”

Almost 900 HD family members came to Las Vegas earlier this month to learn, share, celebrate, remember and gain hope. Staff from the UI HDSA COE were also there to spread the word about ongoing research studies, conduct on-site research and meet HD family members.

HDSA CEO Louise Vetter spoke on June 8 primarily about the HDSA’s strategic plan for 2012-2016. The plan includes a new HDSA research initiative and continued growth of the society, including new COEs, hiring more social workers and involving new volunteers.

In the past year, too many family members and friends have succumbed to HD, Vetter said. The race against time continues, with the desperate need to save the next person.

“HD is not easy. It touches and tears at our families,” she said. “But we unite at convention and as communities, dedicated to improving the lives of people with HD and their families.”

UI HDSA COE Co-director and PREDICT-HD Principal Investigator Jane Paulsen, Ph.D., took part in the clinical research showcase panel on June 9 and talked about the need to use encouraging HD research findings to affect people in their daily lives.

Paulsen talked about several potential new components of PREDICT-HD, including an examination of how one’s environment and exposure to toxins may play a role in HD. Researchers also think there is key information relating to HD progression in the cerebrospinal fluid, which she anticipates will be collected as part of the study in the near future.

During the research keynote address on June 9, Sarah Tabrizi, M.D., FRCP, Ph.D., professor of clinical neurology at University College London and principal investigator of the TRACK-HD study, talked about what she called the explosion of HD research in the past two decades and some new research objectives. One method currently being researched would involve inhibition of Sirtuin-1 that would prevent the mutated protein in HD from being expressed, or, as Tabrizi said, it would be “taken to the garbage disposal.”

Looking forward, Tabrizi suggested the ultimate treatment for HD will likely be a combination of treatments working together to target different parts of the brain, using multiple methods like gene silencing and neuron redevelopment.

“Think of the world as one big laboratory, where everyone works together toward a treatment,” Tabrizi said. “It is our only chance for achieving success.”

UI HDSA COE staff Sean Thompson, Nancy Downing and Pat Ryan contributed to this article.
HDSA recognizes Paulsen’s JHD contributions

By Sean Thompson
HIND-Sight Editor

For her contribution to the understanding of Juvenile HD, UI HDSA COE Co-director Dr. Jane Paulsen was named winner of the 2012 HDSA Juvenile Huntington’s Disease Award.

Paulsen received the award in Las Vegas on June 9 at the HDSA National Convention. She was recognized along with Dr. Martha Nance, director of the HDSA Center of Excellence at Hennepin County Medical Center in Minneapolis and a collaborator with Paulsen on multiple HD research endeavors.

As the principal investigator of the Juvenile Huntington’s Disease (JHD) Natural History Study and JHD Qualitative Study, Paulsen has long recognized the need to learn more about this form of HD that differs from typical adult-onset HD and occurs in fewer than 10 percent of individuals with HD. Paulsen has collaborated with the European Huntington’s Disease Network’s JHD Working Group on new JHD initiatives. She also gives talks and presentations on JHD at various HD events around the world, most recently at HDSA National Convention in June.

“Dr. Paulsen’s JHD studies are incredibly important bases for much of the work underway to better understand juvenile onset Huntington’s disease, and the development of clinical guidelines and procedures to improve the quality of life for everyone affected,” said HDSA CEO Louise Vetter.

Vetter said Paulsen’s work on JHD is also the reason she and Nance were chosen to co-chair a new JHD initiative started by HDSA in conjunction with the National Institutes of Health. The initiative is aimed at developing a comprehensive, united program to further JHD research and improve clinical care.

“We are grateful Dr. Paulsen has agreed to co-lead this initiative,” Vetter said. “The knowledge and experience she brings will help recruit the best possible collaborators and ensure that real progress is made, and more help can be provided to families battling JHD on a daily basis.”

Paulsen is also a member of the JHD Initiative, an organization started by JHD family members to raise awareness and funds for JHD research projects. Additionally, Paulsen has educated patients’ family physicians and teachers on this rare and devastating disease.

According to the HDSA, children with JHD can begin to suffer from symptoms such as stiffness in the legs and arms, decline in cognitive function, behavioral disturbances and seizures anywhere between infancy and 20 years of age.

There is no cure for this fatal condition, but JHD research continues. See the infographic above for information on how JHD families can get involved with research at the UI HDSA COE.

Creatine study continues at UI

When most people think of creatine, bench presses and curls are probably among the first things to come to mind.

But creatine isn’t just a nutritional supplement for strength training. It’s been shown to reduce brain shrinkage in HD mouse models and reduce levels of a possible HD biomarker in the blood in humans. It’s being studied further in the ongoing CREST-E clinical trial, which is enrolling participants at the UI HDSA COE.

High doses of pharmaceutical-grade creatine (or a placebo) are given to participants to see if it can slow HD-related functional decline in daily activities and to make sure such high doses are well tolerated.

See right for more information about CREST-E and other studies you can participate in at the UI HDSA COE.

HD studies currently enrolling at the UI

- **PREDICT-HD:** For pre-symptomatic, gene-positive individuals. Contact Stephen Cross, predict-hd@uiowa.edu, 319-384-1008
- **CREST-E:** For symptomatic, diagnosed individuals. Contact Jacky Walker, jacky-walker@uiowa.edu, 319-353-4357
- **JHD Natural History:** For those who have shown features of Juvenile HD before age 20. Contact Michelle Harreld, michelle-harreld@uiowa.edu, 319-384-1174
- **Kids HD:** For those ages 6-18 who have a parent or grandparent with HD. Contact Joy Goins, kids-hd@uiowa.edu, 319-467-5519

![photos on this page courtesy HDSA](L to R) **HD Chair Don Barr, Dr. Martha Nance, Dr. Jane Paulsen and Louise Vetter.**

Paulsen and Nance received the 2012 HDSA JHD Award at the HDSA National Convention.

A victory for HD families
Listing for HD, JHD to speed up Social Security benefit decisions

Those suffering from the devastating effects of HD and Juvenile HD seeking federal disability benefits will soon receive decisions within days instead of months or years.

That’s according to officials with the Social Security Administration (SSA), which recently added both diseases to the SSA’s Compassionate Allowances List. According to an announcement from SSA CEO Louise Vetter, the Compassionate Allowance program puts disability benefit decisions on a fast track for those with the most serious disabilities.

The SSA website says this identification of serious disabilities is meant to provide disability benefits quickly to “applicants whose medical conditions are so serious that their conditions obviously meet disability standards.”

“This is an important victory for our families, and HDSDA is proud to have led this advocacy effort,” Vetter said in the announcement.

So far, the Compassionate Allowances program has been effective, says SSA Commissioner Michael J. Astrue. In a SSA press release, Astrue says nearly 61,000 people have been approved for disability benefits quicker because of the program in the past fiscal year. When the program began in 2008, there were only 50 diseases on the list; that number now tops 165.

JHD’s addition to the list is effective as of Aug. 11, and adult symptomatic HD will be added by the end of the year, according to the SSA.

The announcement for symptomatic HD was made the day before what would have been the 100th birthday for folk legend Woody Guthrie, who suffered and died from HD. Astrue said in the press release it was a fitting time for the announcement.

The HDSDA is excited about this victory for HD families, but is not ready to rest its legislative advocacy efforts. HDSDA Manager of Program Services and Advocacy Jane Kogan said in an email that the Compassionate Allowance program does not fix the two year waiting period for Medicare benefits, nor does it update SSA’s outdated guidelines for HD, which were written 30 years ago and only take into account the physical impairments of HD. These inaccurate criteria can be another source for delays and often denial of benefits, according to HDSDA. UI HDSDA COE Co-director Jane Paulsen and Neuropsychologist Megan Smith are involved in the effort to update the criteria.

Kogan said the HD community needs to redouble efforts to support the Huntington’s Disease Parity Act of 2011, which would update the HD guidelines and waive the two year waiting period. Kogan urges people to contact members of their congressional delegation who need to redouble efforts to support the HD community.

For more information on the UI HDSDA Center of Excellence, visit our website at:
www.uihealthcare.com/depts/huntingtonsdisease/

Become a fan on Facebook!
Search for:
U. of Iowa Huntington

Jeff’s Litany
By Jeff Sullivan

(Editor's Note: The following poem was submitted by a reader for inclusion in HIND-Sight)

Huntington’s Disease.
Emotionally edgy.
Controls my life.
Takes away slowly.

I’m glad to be alive.
Have Mental Illness.
Mood disorder with bipolar.
Emotions. Shaky.

Origins of HD.
Parts of Europe, Germany; Grandpa.
Image having roots in HD hell.
Brings out anger.

I am glad to be surviving.
HD tantrums are scary.
Have bipolar tantrums as well.
Have fallen eight times onto floors.

Unbalanced; frustrating.
Unsteady; body.
Cane helps keep me upright.
Unbalanced, means, tipsy.

Mother alive at 78.
For her HD created a lifetime in hell.
Now home care respite givers come.
Mom is the angel with the smile.

Caretaking dad.
Bad back and knees.
Needs painkillers,
Works free, hear no complaints.

Team Hope Walk – Sept. 15
Walk, run and fun for hope

Whether you come to walk, run, bid on prizes, eat or just enjoy a morning of family fun, you’ll be helping raise money for a great cause at the HDSDA Iowa Chapter Team Hope Walk on Sept. 15.

The walk takes place in Ankeny at Des Moines Area Community College (DMACC) campus at Building 7. Same-day registration for the adult 5K run/walk and one-mile fun run for kids is at 8:30 a.m., with the walk/run beginning at 9 a.m.

Registration for adults is $20 for adults and $25 for same-day registration. Kids are $15 or $20 on the day of the walk. All participants receive a free T-shirt.

HDSDA Iowa Chapter President Lori Wesack said the event will also include a silent auction, food and a bouncy house for kids. Last year, the event raised over $10,000 for the HDSDA, and she’s hoping for an even bigger event this year.

For more information, to donate to the silent auction or volunteer, email hdsaiowachapter@gmail.com.
Study shows simple procedure can map fetus genome


For the first time, researchers have determined virtually the entire genome of a fetus using only a blood sample from the pregnant woman and a saliva specimen from the father.

The accomplishment heralds an era in which parents might find it easier to know the complete DNA blueprint of a child months before it is born. That would allow thousands of genetic diseases to be detected prenatally.

The paper, published June 6 in the journal Science Translational Medicine, was written by genome scientists at the University of Washington. They took advantage of new high-speed DNA sequencing and some statistical and computational acrobatics to deduce the DNA sequence of the fetus with about 98 percent accuracy. The process is not practical, affordable or accurate enough for [clinical or routine] use now, experts said.

It is already possible to determine the DNA sequence of a fetus by testing the placental tissue. But these procedures are invasive and carry a risk of inducing a miscarriage.

The technique described in the paper would not require complete cells from the fetus and would make such DNA testing easier and less risky. The genome was determined from blood samples taken 18-and-a-half weeks into the pregnancy, although the researchers said the technique could probably be applied in the first trimester.

The technique takes advantage of the discovery in the 1990s that fragments of DNA from the fetus can be found in a pregnant woman's blood plasma, probably the result of [the normal process of] fetal cells dying and breaking apart.

Such information would allow detection of so-called Mendelian disorders, like cystic fibrosis, Tay-Sachs disease and Marfan syndrome, which are caused by mutations in a single gene.

The ability to sequence an entire fetal genome is likely to raise numerous issues. Use of the approach could lead to an increase in abortions because some parents might terminate the pregnancy if the fetus was found to have a genetic disease.

Moreover, a full fetal genome sequence would turn up numerous mutations for which information is lacking as to whether they cause disease, posing a dilemma for expectant parents and their doctors.

“There's definitely plenty of room for improvement,” Professor Jay Shendure, associate professor of genome sciences at the University of Washington, who supervised the research team, said. But, he added, “This is not science fiction anymore.”