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‘No Marine deserted on the battlefield’: two surviving spouses join forces to speed the defeat of Huntington's disease

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At Risk for Huntington's Disease

HD is a genetically caused brain disorder that causes uncontrollable bodily movements and robs people's ability to walk, talk, eat, and think. The final result is a slow, ugly death. Children of parents with HD have a 50-50 chance of inheriting the disease. There is no cure or treatment.

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'No Marine deserted on the battlefield': two surviving spouses join forces to speed the defeat of Huntington's disease

After the deadly, untreatable Huntington's disease claimed their spouses, Jonathan Monkemeyer and Jane Mervar – once strangers, now close – decided to devote their lives to finding ways to speed the search for effective remedies and making the case for the importance of juvenile HD (JHD) in the process.

Without at first knowing the cause of his wife Sheryl's strange illness, Jonathan quit his job in the early 2000s to become her full-time caregiver until she died from HD in 2009 at 46.

"It's the thing you have to do," Jonathan, an accomplished electrical engineer, said in a recent phone interview from his home in suburban Philadelphia. "You really don't have a choice in our country. We did a lot of nice things, which was good. We did peaceful things like traveling to local gardens. She spent a lot of time with our son."

Sheryl died at home. "I didn't expect her to die," Jonathan said. "I thought we would get the cure in time. The doctor said she had five years. But she fell and got hurt. She couldn't sit. I had to hold her. Her weight went from 109 pounds to 89 pounds within four weeks. She died of a heart attack, which is like starving to death."

Caring for Sheryl depleted the family's life savings, Jonathan added. "I'm heating with wood right now," he said. "I'm not using heating oil."

The couple's son Jonathan, now 14, has a 50-50 chance of having inherited the HD gene from his mother. (Usually only adults can decide whether to be tested for the gene, and most choose not to do so. Children can be tested if they already show symptoms.)

A parallel story

Halfway across the country in the village of L'Anse in the Upper Peninsula of Michigan, Jane faced her own difficult odyssey to decipher the disease afflicting her family. She lost not only her 49-year-old husband Karl, but also her 13-year-old daughter Karli Mukka to HD, both in early 2010. (Jane gave her daughters her maiden name.)

"Karl was a wonderful, ambitious, intelligent man," Jane said at the start of an exhausting and emotional four-hour interview. "He had very strong family values. He could always make me laugh."

However, she recounted, gradually "he started to change. Nobody could explain to me what was going on."

HD Links

Huntington's Disease Society of America
International Huntington Association
Huntington's Disease Drug Works
Huntington's Disease Lighthouse
Hereditary Disease Foundation
Huntington's Disease Advocacy Center
Thomas Cellini Huntington's Foundation
HDSA Orange County (CA) Affiliate
HD Free with PGD!
Stanford HOPES
Earth Source CoQ10, Inc.

HD Blogs and Individuals

Chris Furbee: Huntingtons Dance
Angela F.: Surviving Huntington's?
Heather's Huntington's Disease Page

Karl Mervar and daughter Karli Mukka, both victims of HD (family photo)

Like many HD patients, Karl became angry and aggressive, threatening his family with violence.

"Karl held us hostage with his guns," Jane said, recalling the dangers she, Karli, and her three other daughters faced as Karl's behavior became increasingly irrational. "There were a lot of scary, scary times. We had a safe room in the house. We'd go in the room and push the bed up against the door. The girls knew this routine. Then I would try to play with him or try to distract him.

"The darn thing is, I knew we were everything left in the world that meant anything to him."

JHD ravaged Karli's body, displacing the organs in her chest cavity and forcing her spine to the far side. Because of the disease's uncontrollable movements, Karli had chewed off half of her tongue by the time she died, Jane said.

A nurse suggested that Jane give Karli morphine and "let her go." She declined the advice.

"It was a hard spot to be in," Jane said. "I talked to Karli and asked her if she was ready to go on baby Jesus's lap. She said no. She died of natural causes."

Today Jane just gets by financially, thanks to Social Security benefits, as she cares full-time for her two other daughters with Karl, 22-year-old Erica Mukka and 20-year-old Jacey Mukka. Like Karli, both have JHD. Karisa Mukka, a 26-year-old daughter from a previous relationship, lives nearby.

Partners in love and advocacy

In June 2010, still in mourning for their lost loved ones, Facebook friends Jonathan and Jane struck up a lively conversation while sitting next to each other at the 25th convention of the Huntington's Disease Society of America (HDSA) in Raleigh, NC. After the convention, they spoke daily for at least a couple of hours. Jonathan visited L'Anse, and shortly thereafter Jane and Jacey stayed nine days with the Monkemeyers.

Their friendship led Jonathan and Jane into a romantic, long-distance relationship.

"They're an incredible family," Jonathan said of Jane and her daughters. "Their value system is not about themselves."

"I was in a pretty low place," Jane recalled. "I had lots of grief after Karli and Karl died. I wanted a reprieve from caregiving – just wanted to be dead. I'd be laughing after I finished talking to Jonathan. I think he saved me."

Their relationship and support for each other's families also became a partnership in advocacy for HD patients.

Bridging the gaps

To achieve their goals, Jonathan and Jane are politely but firmly challenging bureaucratic inertia.

Supporting himself and his son with his son's Social Security survivor benefits, Jonathan dedicates himself full-time to HD advocacy. He has developed a deep understanding of HD science. By his account, he has so far skimmed through more than 10,000 scientific articles related to the disease.

Applying an engineering approach to the problem of developing treatments, Jonathan developed a website, HDCircle.org, currently offline, on which he has posted information about HD researchers from around the world, links to HD organizations, and reviews of potential HD treatments. He plans to reactivate the site soon.

He also created a Facebook discussion page, Hereditary Disease Circle, with the goal of finding connections between HD and other neurological conditions such as Parkinson's disease, multiple sclerosis, posttraumatic stress disorder, and amyotrophic lateral sclerosis, also known as Lou Gehrig's disease.

In addition, he attends relevant meetings and conferences in order to network with people from other disease communities. He seeks to bring their best results and ideas to bear on HD research.

As Jonathan explained, he aims to create "synergies" and "bridge the gaps" among HD researchers and the various key organizations such as HDSA, the Hereditary Disease Foundation (HDF), the CHDI FoundationInc., the multi-million-dollar, non-profit virtual biotech formed solely to seek HD treatments, and the National Institutes of Health (NIH).

As one example, Jonathan said he has spoken personally with NIH Director Dr. Francis Collins, one of the pioneers in the search for the HD gene in the 1980s and 1990s, more than a dozen times, including last week in Washington, D.C.

NIH Director Francis Collins (left) and Jonathan Monkemeyer (personal photo)

"Essentially, engineers design things hierarchically," Jonathan explained. "I created a website, which is a blueprint for how the system works.

"We're the only disease without a gene therapy. There have been 1,000 gene therapy clinical trials. But we as a community don't seem to be organized enough. There's something in our organizational structure. By their very nature of having a job description, when you're within an organization, your function is to be in the organization. Everybody gets stuck in a silo of what they're doing. With so many scientists and stakeholders in the field of HD research, moving forward gets stymied by committee and the sense of urgency gets tuned out."

If he held a position within one of the organizations, "I'd have a boss to report to," Jonathan continued. "As an outsider, without a job, and asking questions as an advocate, it gives you the position to help steer people in the right direction towards what needs to be done. I have the greatest freedom, not being employed in an organization. I can talk to anybody I want to."

Sometimes Jonathan feels as if he's "walking on egg shells, because I'm not a researcher," he said. "You tell people very nicely and very artfully. We don't tear down institutions. We build them up."

He summed up his approach as "doing what needs to be done to drive innovation that will bring a therapeutic to our community."

What might work

Significantly, Jonathan's efforts include canvassing the research community for the latest discoveries and techniques that could translate into therapies for HD. He seeks to brainstorm about new developments, as well as previous ones, in his conversations with scientists.

Rather than simply await for the multi-million-dollar pharmaceutical efforts to bring results, advocates must actively participate in the search for treatments, perhaps even trying drugs and substances approved for other purposes in their own off-label studies, seeking advice from researchers on dosing, and having people reporting their observations via a website, Jonathan suggested.

He cited the example of Hannah's Hope Fund (HHF), whose advocates teamed with researchers in a low-cost effort to develop gene therapy for a rare genetic condition known as giant axonal neuropathy. HHF has met

with the federal Food and Drug Administration (FDA) and, if a safety study goes as planned, could start a clinical trial this year.

In the drug-discovery system in America, the profit motive "stymies innovation and responsibility" towards the patients, Jonathan observed. Rather than producing strong leaders like a Jonas Salk, who developed the vaccine for polio, the system today fosters a climate of "let's make everybody happy."

Jonathan also pointed to the new partnership between the NIH and the Milken Institute/Faster Cures, which seeks to increase collaboration among the government, foundations, universities, and the pharmaceutical industry in order to cure more diseases and do it faster. This initiative includes the creation of a new NIH program, the National Center for AdvancingTranslational Sciences.

Advocates for rare diseases like HD need "to get involved in every single aspect" of the search for treatments.

"AIDS advocates made the FDA bend," he noted. "We the patients have a moral incentive. Our voice counts and makes the difference."

An epiphany about JHD

Like other JHD advocates, Jane and Jonathan have strived to increase the attention to JHD by researchers, HD organizations, and the government.

In September 2010, Jonathan and Jane met with Dr. Steve Groft, the director of the Office of Rare Diseases Research at the NIH, to press the case for greater support for HD research.

For Jonathan and Jane, the meeting was a kind of epiphany. Pointing to the different emphases and the existence of different organizations in the field of diabetes and juvenile diabetes, Groft helped them see the key role that JHD research could play in the overall HD effort.

Dr. Steve Groft and Jane Mervar. In the middle is Max the Turtle, Karli's stuffed animal companion that is now a mascot of the JHDKids initiative (family photo).

"The meeting was phenomenal," Jane said. Jonathan said she came away with a greater sense of "we need to do something."

"Everybody was so resistant to acknowledge the juvenile population," Jane said. "It's just like some big political game. Nobody was playing that game for our children, so we were screaming: *we need a cure, we need a cure, we need a cure!*"

"You need a piece of legislation to get JHD funded, and then the NIH would fund it," Jonathan explained, pointing to one of the roadblocks facing the efforts to understand and treat JHD.

Both Jonathan and Jane observed that JHD research lags far behind other HD research, and, because of ethical concerns and the need to avoid mixing juvenile and adult research data, children aren't included in clinical trials.

Jane tried but failed to sign up Karli for a trial for ACR-16, seen as a potentially promising HD remedy. Jane described the formal response she received as "too bad for you, you have Juvenile HD." "I was devastated," she said.

Karli also took the supplement creatine, currently under study for HD and taken by many in the community. "It took us almost a year to get two doctors to follow Karli when she was on creatine and to get a guideline on dosage," Jane recalled. "There are a lot of families that are just slipping through the cracks."

Researchers have also lacked a so-called "natural history study" of JHD – a study to follow a group of patients over an extended period to better understand the condition and support the development of treatments.

Jane and Jonathan's advocacy has included pressing the HD organizations to pay greater attention to JHD, they said. Thanks to pressure from JHD families, last year HDSA agreed to the creation of a new fundraising effort specifically for JHD, Jane explained.

"Great things can be created from hard situations," Jane observed.

Jonathan and Jane at the White House after their meeting at the NIH Office of Rare Diseases and Research (family photo)

Raising the profile of JHD

Jane, her daughters, and other JHD families swung into action, joining other grassroots JHD initiatives in the effort to raise awareness and funds for research.

Jacey and Erica started JHDKids.com. With a computer and video equipment provided by the Make-a-Wish Foundation, Jacey has made a series of short films, including one about Karl and Karli titled *The Real Huntington's Disease*, which has had more than 220,000 views on YouTube. (Watch the video below.)

The JHDKids initiative is seeking funds specifically to support the JHD research of Dr. Jane Paulsen, the co-director of the HDSA Center of Excellence at the University of Iowa, and project partner Dr. Martha Nance, the director of the HDSA Center of Excellence in Minneapolis.

Both researchers work on a volunteer basis, with all funds raised going solely to research.

In the words of the JHDKids site, JHD differs significantly from adult onset HD in several ways. "The most significant difference is that in JHD the disease occurs before the brain is fully developed," says a statement on the site from the researchers. "This accounts for the wide variation at one age from JHD to another age. Maturation and neurodegeneration occur at the same time in JHD."

Drs. Paulsen and Nance began following JHD patients at the annual HDSA conventions in order to carry out the natural history study.

"Now, with the awareness and fundraising getting more widespread, they're able to bring our families into Iowa," Jane explained. "We're planning on going this spring. That will be the first time we're going to Iowa."

One big family

Jonathan and Jane have talked of bringing their families together in one place.

For now, however, they will continue their hours-long daily conversations from their respective abodes. Jonathan's location on the East Coast facilitates his access to the corridors of scientific and medical power, while Jane wants to respect Jacey's wish to die in the same place as her sister and father.

The distance does not diminish their commitment to each other's families, nor to the larger cause.

Jonathan helps Jane manage her caregiving crises. "He's always learning something new," she said. "He's a very faithful, spiritual person. He's just a very good man."

Erica and Jacey's doctors wouldn't predict how long they might live, although JHD patients typically die in their 20s or 30s, if not sooner, like Karli.

Jane knows that an effective, life-saving treatment might not come in time to save them. She concentrates on providing them with the healthiest, happiest life possible. Having worked as a nurse's aide in a nursing home and seen Karl go through his final decline in such a home, she hopes to keep her girls at home as long as possible.

"Jacey has a big phobia that if she can't see me, she'll die," Jane said. "We can't calm that down. She likes to watch movies. She likes to work on the website. She likes to see all the kids with JHD. She likes to come up with new ideas for designing the website."

At 19, Erica had married her high school sweetheart, but the marriage lasted only 11 months. She is currently dating another man. "She wants him to learn how to do her makeup and coordinate her clothing," said Jane, who has legal guardianship over both daughters.

She obtained a court order to obtain permission for a tubal ligation for Erica.

"I cried with her," Jane said. "It was just a real painful process to go through. She said, 'If I had a baby and got sicker, and what if my baby's like Karli?'"

"Fear for my son is certainly a reason," said Jonathan of his commitment to the HD cause, noting that, so far, his son has showed no HD symptoms. "That's a personal, selfish motivation. Why is my son's life more important than someone else's life? I know Jacey and Erica and everybody else that's dying from this."

He added, "I'm doing this full time, and as far as I'm concerned, there's nothing more important I can do with my life. It's knowing everybody in the community and knowing the suffering and the damage it causes to families. You don't leave a marine in the battlefield. It's just wrong to walk away. I can't stop doing it. This is my life experience."

His experiences as Sheryl's caregiver have deepened his feelings about others facing the same plight.

"When I see HD in somebody else, the empathy is much more intense and overwhelming," he confided. "To me, we're all a big family. That's why I can't walk away."

Posted by Gene Veritas at 4:10 PM

Labels: advocacy , CHDI , clinical trial , cure , gene therapy , HDF , HDSA , hereditary , Huntington's disease , juvenile Huntington's , NIH , nursing home , pharmaceutical , research , science , symptoms , treatments

9 comments:

Unknown said...

This comment has been removed by the author.

11:36 AM, February 12, 2013

Unknown said...

Gene, are you now advising your readers to take off label drugs and seek advice from researchers for dosing? As you wrote from your interview with Mr. Monkeymeyer...
"Rather than simply await for the multi-million-dollar pharmaceutical efforts to bring results, advocates must actively participate in the search for treatments, perhaps even trying drugs and substances approved for other purposes in their own off-label studies, seeking advice from researchers on dosing, and having people reporting their observations via a website, Jonathan suggested."
**Not only is this illegal for a researcher to advise on dosing [this should only be done by a licensed physician], but no researcher will jeopardize a human being or their license and livelihood. This could potentially harm or even kill someone. Gene Veritas I'm really surprised that you would write this ill-advised content in your blog.

12:29 PM, February 12, 2013

Anonymous said...
Just a sign of the helplessness and desperation a person with HD or a family member feels watching their loved ones suffer with no treatment or cure in sight.

5:45 PM, February 12, 2013

Anonymous said...
I get it what melisa said and I feel the same way. I am a christian woman and watch every day how this hd is but we have to be safe and see our doctor!! I had left a group on facebook because of to much going on and to much nonsence Blessed all who are sufferring AMEN!

8:11 AM, February 13, 2013

Anonymous said...
Gene, your articles are always so beautifully written. This one hits home even more as I know the people involved. Jonathon and Jane serve selflessly to move the therapy and cure forward. Jonathon's opportunity and ability to brainstorm with scientists is so good for the cause. This allows him to take a step back and look outside the box. The research community is usually driven by profit. These two are working for real people and living on a shoe string to propel their ideas forward. I appreciate Jane and Jonathon's efforts from the bottom of my heart. Thank you again Gene for you way with words.

10:19 AM, February 13, 2013

Anonymous said...
I am afraid that I am worried about the advice about the other drugs and substances for our people. I think it is unsafe and it is giving people bad advice without a doctor involved.

I also removed myself from their group and their drama like the other person it is just so sad but we cant be around it anymore.

Irene

2:28 PM, February 15, 2013

Anonymous said...

Thanks Gene, for highlighting Jane's story. I've heard it befoe and it has brought tears to my eyes several times. She is strong and brave, and I'm pleased to hear that she has found someone to share her journey with. May their work to bring light to this disease go well.

3:53 PM, February 17, 2013

Anonymous said...

It was a wonderful blog.. and Melissa.. your comment is really dissapointing. You would think you would want to help expose morw research.. i couldnt find where anyone was saying to take a suplemnet.

Pretty sad when one advcote attacks another.. you may not agree with john or Gene but to attack dose nothig to help.

Tara Hansen

6:38 PM, February 12, 2014

Anonymous said...

Huntingdont's disease can't get a cure until there is either a discovery that would come from another field that works out how to switch off, or greatly slow down the impact of the cag repeats.

Too few persons from afflicted families are prepared to actively do their part in getting tested early in life to provide sufficient data re numbers carrying the gene, potential for researchers to be able to analyse the varying courses if the disease, be in a position to almost eliminate any JHD cases and a majority of future adult onset cases of HD.There will be always a small number of HD gene positive persons while unsymptomatic who had utterly no notion of the possibility of their status. and hence passing it on through no information of a parent carrier. The cases of obscured extra late familial onset could be a source, though if HD was actually made truly widely recognisable as a condition, it's symptoms etc.,it would increase their younger generations members to check out for the condition.Too much familial underplaying or even partial efforts of concealment of HD, shame, fear of others more widely knowing of the condition impacts on elimination. Secondly the fear of supposed insurance, employment,plus justifying remaining ignorant as it's too upsetting to know if positive need to be countered. Few end up collecting great insurance plus employment ends not by gene positive status

but by diease onset. Finally the understandable but selfish desire to have a spouse and kids so push it into the denial optimism basket where due to low public knowledge spouses ignore the hints of the familial disease they heard hit some relatives.The rule ought to be if an at risk person thinks the prospect of finding out is unbearablely awful, it's obviously too awful a disease to transmit by having children aside by now pre-implantation selectiveness with no info to diulge gene status or earlier to have used donor gametes or adoption or no kids.Too many think I'd rather I was born while unsymptomatic but at risk, ought to be good enough for next generation, then devestated and guilt ridden once symptomatic or have a child with JHD.

More if the money raised by HD organisations needs to go into altering widespread awareness, bringing the disease out of the closet, prevention of it's transmission, working on highly effective treatments for symptoms after onset, securing quality services to care for persons comfortably, prompt income assistance when low incomes need benefits. It's inexcusable that a large proportion of JHD cases who need need have occured with identifying testing having been available from the early 90's.Or young persons being at risk.

I did a contract for a HD organisation 16 years ago, the same mantra of they'll find a cure, we need one with heads in the sand, few testing their status, yet not much has changed at all, especially in effective symptom managed treatments which is where for the ones to become symptomatic the best hopes could lie to reduce the horror and terror of the ineitable progression. Until then all efforts ought to be in preventing anyone you can be born at risk. If they do however find a cure or close enough to by effective slowing, then those at risk can tumble their dice of hopeful chances.

What's interesting is that in all that fund raising no HD organisation does divulge the actual money and man power of research efforts that can be evaluated in numerous directly likely to be effective results or if they are ever shared with other researchers in parrallel genetic fields. I've the greatest sympathy for those dealing with the disease, admiration for so many dealing corageously and tirelessly with the impact of this condition. Our engineers concerns were great to be raised. The HD organisations though wonderful in much of the work and support they deliver have gotten too entrenched, dogmatic, dominant in forcing unquestioning compliance by other professional bodies working alongside of them causing a stagnation impeding progress that's possible.

6:36 PM, October 13, 2014

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