Finding a Cure
Why is it so Difficult?

Article by Bruce Gaughran

Ed Meyertholen, a biology professor and KDA research interpretation guru, posted the following response to a KDA Forum question on why finding a treatment or cure for Kennedy’s and other neuromuscular diseases is so difficult. Ed did such a fine job of clarifying the problem I thought that others should have the opportunity to read his response.

Reference medical research on neuromuscular diseases, no one has yet determined exactly why the cells die in such diseases, much less how to stop the cell death. This is not due to lack of money, effort, organization or urgency, but because it is a very, very difficult problem. Each cell has thousands of machine-like molecules called proteins and each protein has a specific function in the cell. Of these thousands of proteins, it is not obvious which are important for keeping the cell working properly – or even how they keep the cell working properly.

Cont’d Page 2
**Dates:**
Wednesday, Thursday, & Friday
October 10, 11, & 12, 2012

**Location:**
Harrah's New Orleans
228 Poydras Street
New Orleans, LA 70130
(504) 533-6000

**Pre-Conference Gathering:**
Don’t miss the pre-conference get-together dinner on Tuesday evening, October 9. This will be a great opportunity to meet new friends and get reacquainted.

**Hotel Info:**
1. The AAA Four Diamond Award-Winning Harrah's Hotel is located in the heart of the world's most exhilarating city. This 26-story marvel, with 450 oversized luxurious rooms and suites, provides an unparalleled escape. Rooms feature deluxe beds, high-definition flat screen televisions and wireless Internet.
2. Enjoy spectacular views of the Mississippi River and New Orleans skyline. Located just steps from the French Quarter and Convention Center and a few blocks from the Superdome, there is no better place to stay in New Orleans than Harrah's New Orleans luxury hotel.
3. The room rate is $142 per day plus tax, and twelve handicap rooms are available. Blocks of rooms are reserved for October 09, 10, 11 & 12, but this rate is good for two days prior to and one day after the conference if you would like to extend your stay!

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**Cure, Cont'd from P1**

Within the brain alone, there are thousands if not millions of different types of cells, each with their own function and specific set of proteins. Kennedy's Disease (or Parkinson Disease or Huntington Disease or ALS etc.) all cause a small subset (and this subset is different in each disease) of these cells to die. It is not understood what specifically makes each cell type different or why only some neurons die in each of these diseases or, in fact, why any nerve cells die in these diseases. It is very difficult to find a cure for Kennedy's Disease when it is not even understood why the cells die.

One of the first steps to understanding why a cell dies in Kennedy's Disease is to understand how the cell works—thus understanding the role of each of the proteins in the cell.

We do know that in Kennedy's Disease, the defect is originally in the protein called the Androgen Receptor (AR) and somehow (no one knows how despite extensive research) this leads some specific nerves cells to die. To figure out the molecular cause, it is necessary to figure out all the possible interactions that AR can have and try to identify the one that causes the cells to die. The AR interacts with hundreds of other proteins. Which of these interactions is relevant for Kennedy's Disease? No one yet knows. This is akin to finding a specific hay strand in a haystack—it is a very difficult problem and, despite our wishes, it takes time to work this out.

There is another issue that needs to be understood. Suppose that someone develops a treatment for Kennedy's Disease tomorrow. What would this treatment do?

Most likely, it would prevent the nerve cells from dying. It is, however, extremely unlikely that this treatment would regenerate new nerve cells (this is technology that is quite futuristic).

Thus, those of us with Kennedy's Disease that has progressed will not have our symptoms reversed—we would just stop the current downward progression. While this would be a tremendous development, we would still have Kennedy's Disease symptoms.
The Kennedy's Disease Association (KDA) is planning to fund one or more research grant this fall to further the understanding of the pathological mechanisms of Kennedy's Disease. The projected funding for each grant will be up to $25,000.

Applications from junior investigators and from senior post-doctoral fellows are encouraged. Proposals must be received by Friday, June 22, 2012. The KDA will send a confirmation email within three days of receipt. Please send proposals to the following email or physical address:

Email: info@kennedysdisease.org

Scientific Review Board
Kennedy's Disease Association
PO Box 1105
Coarsegold, CA 93614

Schedule and Review Process:

- Friday, June 22 - Full Grant Proposals are due at the KDA and the review process begins.
- Proposals will be reviewed by the Scientific Review Board (SRB) of the KDA and, if required, a panel of reviewers not affiliated with the KDA.
- Monday, August 27 - SRB makes grant funding recommendations to the Board of Directors.
- Wednesday, September 12 - KDA awards grant(s).
- Monday, October 1 - Grant(s) are funded.

The general guidelines for the format of a proposal can be downloaded by clicking on this link: 2012 Research Grants

How Your Donations Are Used
Fiscal Years 2007 thru 2011

- Research Grants: $234,014 (79%)
- Education: $35,880 (12%)
- Op. Expenses: $25,779 (9%)

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KDA Research Grants
New Funding Opportunities for Kennedy’s Disease Research
I read the following abstract the other day and it immediately raised a flag. I have heard several stories about complications that resulted from a doctor or surgeon not knowing a patient has Kennedy’s Disease, or an anesthesiologist not being aware of the problems that might be caused by using the wrong anesthesia on a patient with a neurodegenerative disorder. 

Complications After Cardiovascular Surgery in a Case of Undiagnosed Spinal-Bulbar Muscular Atrophy (Kennedy Disease)

Stacey A. Skoretz, MSc, May-Sann Yee, MD, FRCPC and Rosemary Martino, MA, MSc, PhD

Abstract

Neurodegenerative diseases are often associated with life-threatening declines in respiratory and swallowing mechanisms. We report the case of a 70-year-old man who had postoperative dysphagia and respiratory failure that required reintubation after coronary artery bypass surgery. Impairment of the patient’s speech, swallowing, and respiratory mechanisms identified during postoperative clinical and instrumental examinations was suggestive of a neurodegenerative disease. Genetic testing confirmed a diagnosis of spinal-bulbar muscular atrophy (Kennedy disease). This case report aims to highlight increased morbidity in patients with undiagnosed neuromuscular disorders in the critical care setting and the benefits of vigilant postoperative monitoring and multidisciplinary involvement throughout the care of complex patients.

Over the last three years I have written several articles explaining the need to carry a medical information card and to be your own medical advocate (spokesperson). You also need someone who can be an advocate for you in case you are unable to express your feelings.

The key message to take away from this article is that there might come a time when you are unable to advise your surgeon of your medical condition, allergies, etc. Being prepared ahead of time with several backup options could save your life.

KDA Carriers – An Invitation to Share

Hi Friends,

It is tough to be a carrier, as most will agree; I’m sure for many reasons. I think it might be interesting for us to get a chance to talk and exchange contact information. I’ve been in touch with a few of our very own KD researchers and other carriers in the organization to get more informed on the topic. I am planning to attend the Conference this October and hopefully we can all meet together for a bit. If you are interested and planning to attend as well, please drop me a note at kdacarrier@swbell.net and we can make some plans. If you cannot attend this year, let me know your comments or questions, so that I might forward them to the experts and get back with you. Thanks for your support!

Mary.

You are not alone

The KDA’s mission is to inform, support, educate, fund research, and find a cure for Kennedy’s Disease.
I’ve often said that I have enough friends; how stupidly selfish of me. I am a much better man for having known Ron Wiker and Charlie Rannells.

We had met ten years ago at the Kennedy’s Disease Association Conference in Baltimore, one of the first ones, in 2002. None of us knew more than a few of the others at that time, and partly because of geography, (Ron in Lancaster, Charlie in Martinsburg) we bonded, getting together at my home in Westminster, MD or at their homes on different occasions. Each time was, no is, memorable.

With each of us having KD, the ability to openly discuss things was important; as we could share differently from the impersonal chats having met in person. Both men were devoted family men, loving husbands, fathers, and grandfathers; patient to a fault, but strong men in their own way.

Ron was ahead of his time in 2002 as he was the ‘inventor’ of the expanded waist pants now sold by every clothing store in the USA. He also had fashioned an easy-to-grasp pull for a man’s fly, and was the first to notice that almost every man with KD had a full head of hair. Charlie was a wealth of information, having been the voice of the US Department of Agriculture for years. We shared great times.

As time went on, each of them relied on a feeding tube for nourishment. That did not dampen their spirits; it was just what they had to do. Both men were down-to-earth spiritual, quiet in nature; the kind of a friend that you didn’t have to talk with every week; a conversation could be continued next week, month, or six months down the road without a break in continuity.

I spent a great hour with my friend Ron about a month ago, and it was a joy and blessing to be in the company of a great man who I had the privilege of knowing. I’m a better man today because of Ron and Charlie, and I miss them both.
When they attended the 2010 KDA Conference in San Diego Cliff & Liz Johnston, from Australia, did not know about the custom of the bringing an item to be auctioned off after dinner at the KDA Silent Auction. So they thought about it, and came up with a plan to host a couple at their home in Melbourne for 4 days and 5 nights – complete with airport transfers, accommodations, sightseeing in the Melbourne area, and meals!

When the bid started at $100 Paula and I were there! Someone bid it up but we kept upping it until finally, at $250, it was ours! Paula and I were VERY excited about the trip and stayed in touch with Liz & Cliff until we finally settled on a date in December (before it got too hot) to visit Down Under. We decided that it would be better to break the trip up into segments to ease jet-lag. So, in late November, we struck out to visit Len & Jeanne Janicki in Las Vegas for a few days before going to Honolulu and staying at Waikiki Beach for several days to enjoy the beach and try to catch some surfers on the north shore of Oahu at the Kamikaze Pipeline. Turned out we had just missed the big waves two days ago but still enjoyed the trip. We had acclimated to the time zone there sufficiently when we boarded the plane to Sydney and, 8 hours later, we were landing in Sydney and transferring to the domestic terminal to go to Melbourne. When we arrived in Melbourne Cliff and Liz were there to pick us up. They had just come from a dinner show and were in formal attire! Nice way to be greeted – we felt like VIPs from the start! They showed us their beautiful house that they had renovated from a row house from the 1800s. Our sleeping accommodations were perfect! They had the entire 4 days planned and reviewed the itinerary with us for our approval before going to bed that evening.

On day one we headed off to the Healesville Sanctuary, about 1 hour NE of Melbourne. There we saw most all of the wildlife that you could possibly see in Australia. From the Kangaroo and Platypus to the Koala, Echidna, Tasmanian Devil, and Wombat – everything Australian.

And then we had a great picnic lunch that Liz had prepared for us – it was so relaxing to enjoy a great meal in such peaceful surroundings. We loved it!
Day two was spent admiring the beautiful sights and art in downtown Melbourne. Even the streets and highways have been adorned with art—from road noise sound barriers that have been beautified to huge art sculptures that are placed throughout the city... art is everywhere. We were very lucky to have been in Melbourne during Tjukurrtnju: Origins of Western Desert Art Touring Exhibition at The Ian Potter Centre. We could have spent the entire day there looking at Aborigine artifacts—and not seen it all. Unfortunately, any type of photography was strictly forbidden in the Museum so the only memories we have are in our minds (and, of course, the Internet). Afterwards we went up in the Eureka building to see the Skydeck and go out on the Edge.

Paula actually found some gold—although it was only a couple of flakes weighing less than could be measured.

Later we visited the Ballarat Gold Museum and saw replicas of nuggets weighing thousands of ounces—where the expression "Mother Lode" came from.

When we got back to the Johnstons’ house we realized we were fortunate to have picked the same week to visit that their neighborhood had their annual ‘Block Party’—and it was in front of the Johnstons’ house! We met some really great people from their neighborhood and had some fantastic dishes that people brought to share, along with some of Cliff’s wine that is the best I’ve ever tasted.

The elevator we rode went up 88 stories in 37 seconds! It was an ear-popping experience! From the highest public observation deck in the Southern Hemisphere we were able to see every section of Melbourne and hundreds of miles to the Great Divide. We were VERY impressed! And, as if that wasn’t enough, Cliff & Liz outdid themselves that evening by serving a delicious leg of lamb that Cliff had grilled to perfection—complete with all the trimmings that Liz had prepared.

On day 4 we had the honor and privilege to meet Cliff & Liz’s grandson, Max. I refer to Max as the ‘miracle child’ because he is the only person alive that I know of who was conceived with KD and is now 100% cured. Cliff’s daughter, Kellie, is a carrier and when she and her husband, Chris, decided to have a child, they first consulted a doctor who explained PGD to them and suggested it was an alternative to passing the defective gene to their offspring. Max had just turned 5 and was as healthy and sharp as any child could be. This is truly an amazing procedure and their family would be happy to speak to anyone who is interested in learning more about it.

On day 3 we took off for Sovereign Hill—a living gold mining outdoor museum depicting life in Ballarat as it was in 1851–1861. We witnessed gold mining tools being made, rock-crushing machinery in operation, and even had the chance to pan for gold.

Later that same day we visited the Royal Botanic Gardens in Cranbourne. To see and learn about all of the native plants that were there would have taken days but we only had that afternoon so we limited our sightseeing to the most unusual species of flowers and plants. We covered acres of plants, trees, and flowers and ended up in the gift shop for some coffee before returning to the Johnstons’ house.
That evening we were treated to some delicious grilled Kangaroo fillet—it was very lean and tender.

On our last day in Melbourne we went shopping at Queen Victoria Market and bought Aussie shirts, hats, and a kangaroo skin. We drove around Melbourne one last time before saying goodbye to our good friends Cliff & Liz (and Polly, of course!) and boarding our plane. It was a trip we will never forget!

PS – Stay tuned! We’ll tell you all about Perth in the next newsletter.

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**Important Notice – Conference Updates!!!!!!**

We currently have 25 rooms blocked for Tuesday, Oct 9 and 30 rooms blocked for Wednesday & Thursday nights. We can increase this as needed but only IF the hotel has rooms available. If there is a chance you may be attending the KDA conference this year, you are requested to reserve your rooms ASAP so that if we need additional rooms we can increase the block before the property runs out of rooms – which we are guaranteed will happen because Harrah’s – New Orleans is a very popular hotel. If you need to cancel, we are allowed to decrease the block up until 30 days prior to the event but, after that, the KDA is responsible for any unused rooms in the block so we especially don’t want to err in that direction after September 9th.

When making your reservation, call toll-free at 800-223-7277 and be sure to tell the agent you are reserving a room for the **KDA Conference** under the **Group Code of $10KENN** in order to get the room rate listed above.

Along that same line, we are trying something new this year to help reduce Conference costs. We would like to offer a sponsor page on the last page (or back cover) of the conference agenda booklets that will be passed out to all attendees. This page will list companies who contribute $500 (or more) to the KDA to sponsor doctors’ and researchers’ rooms for the conference. The company logo (in color) and a short write-up about the sponsoring organization’s products or services will be included.

Without the doctors and researchers there would be little advantage to holding a conference, so the new sponsorship program should be a tremendous help to the KDA and allow us to provide even better grants to researchers. If you know of a company that would be willing to participate in this program please let us
Traveling Abroad With KD

Article by Paula Goynes

My husband, Mike, and I had a wonderful vacation in Australia last year. We spent ten days with two very special KD families. Liz and Cliff Johnston showed us all the wonderful sights in Melbourne and then we flew to Perth where Mike White’s family welcomed us into their home. Mike was working offshore, but Sharon and their two daughters, Hannah and Charlotte, made us feel like family immediately, even though we had never met before our arrival.

We had an absolutely marvelous time with both families and will never forget their hospitality. We hope to share the pictures and memories with many of you at this year’s KDA conference in New Orleans.

Our trip to Australia was a magnificent once-in-a-lifetime adventure for us, and we were very impressed with the wonderful facilities and benefits available to their disabled citizens.

Sometimes I get discouraged and frustrated with the way people with disabilities are treated in my country, but after our experience on our layover in Guangzhou, China, during our flight home, I am once again reminded that most of us are very lucky.

We found the airport in Guangzhou is definitely not accustomed or prepared to deal with disabled travelers. After much trouble and long delays just to disembark the airplane (including only one wheelchair for two disabled passengers), we finally reached the gate for our next flight. Then an airport official came and took me away from Mike. She said I must go with her and to take only my passport and boarding pass.

She kept talking about a “push cart,” but her English was poor and I do not know ANY Chinese so we had trouble communicating.

Mike and I decided she must be talking about the electric scooter we use when traveling and that it must have been damaged in transit, so I left everything else with Mike and followed the young woman, carrying only my passport and boarding pass, as instructed.

She had me RUN full speed all through the airport, backwards through security and customs and then directed me to give my passport to an official at a desk in a back room. Then she took me to the area where they apparently checked incoming baggage. She showed me the battery to Mike’s scooter and told me it was “very dangerous.” I tried to explain that we have travelled on many airlines and in several countries with the scooter and the battery is definitely not dangerous, but it did no good.

There was some discussion among officials before my passport was returned to me and I honestly thought they were going to arrest me for trying to put a “dangerous” item on the airplane. Then I was forced to run through the airport again, including customs and security, and then instructed to “stop run now” when the gate was in sight.

The gate officials and flight attendants were all angry with Mike because he would not board the plane without me, but he had no idea where I was or what was happening and was afraid they would take off without me. Of course, they had given our assigned seats away and we were sent to the very last seats at the back of the plane. No one offered to help as I struggled with our carry-on items while Mike used seat backs to help him manage the long walk to our seats.

The battery is still in China and when we retrieved the scooter in Los Angeles, it had been severely damaged, including dents and torn seat.

This article is not intended as a criticism of China as a country, but is submitted in an effort to help others with disabilities to foresee and, hopefully, avoid similar issues when traveling abroad.
For all the negativity applied to having Kennedy's Disease, occasionally research comes along reflecting something positive. MSN’s Health published the following report on April 11.

**Huntington's Disease Linked to Reduced Cancer Risk in Study**
Findings suggest shared genetic mechanism, researchers say -- Mary Elizabeth Dallas --

WEDNESDAY, April 11 (HealthDay News) -- People who have Huntington's disease are much less likely to develop cancer than people without the inherited disorder, according to a new study that suggests the diseases share a common genetic mechanism.

The Swedish researchers found that those with Huntington's had a 53 percent lower risk of being diagnosed with cancer compared to the general population.

Besides Huntington's disease, the lower cancer risk applies to the other eight rare neurodegenerative disorders known as polyglutamine (polyQ) diseases. Those diseases, which result in the progressive degeneration of neurons involved in motor control, include spinobulbar muscular atrophy (also known as Kennedy's disease); dentatorubral-pallidoluysian atrophy; and six types of spinocerebellar ataxia.

From 1969 through 2008, the researchers identified 1,510 patients with Huntington's disease; 471 people with spinobulbar muscular atrophy; and 3,425 with hereditary ataxia, a substitute for spinocerebellar ataxia. Cancer was diagnosed in 6 percent of the Huntington's patients, 7 percent of the spinobulbar muscular atrophy patients and 12 percent with hereditary ataxia.

The study, published online April 11 in The Lancet Oncology, determined those with spinobulbar muscular atrophy had a 35 percent lower risk of cancer, and patients with hereditary ataxia had a 23 percent lower risk. Before being diagnosed with a polyQ disease, the patient's risk of cancer was even lower, the researchers said.

"Our findings suggest a common mechanism in patients with polyQ diseases that protects against the development of cancer," Dr. Jianguang Ji and colleagues from Lund University and Skane University Hospital, in Sweden, wrote in a journal news release. "Future studies should investigate the specific biological mechanisms underlying the reduced cancer risk in patients with polyQ diseases," they concluded.

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