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Charles was Part of His Plan for Me

By: Tiffany Beck

When asked to write an article about the concerns of being a Kennedy's Disease (KD) carrier, I found it difficult to write without addressing some controversial issues. What follows only begins to scratch the surface of my feelings and experiences as a carrier of Kennedy's Disease.

In the spring of 2005, I found out that I was a carrier of Kennedy's Disease. The first thing that came to mind was my future family ... what were my options? I always wanted kids; however, I grew up watching my grandfather slowly fade with Kennedy's Disease. The National Institute of Health, as well as, many organizations offer genetic counseling for those who want to learn more about your chances of having a child with Kennedy's Disease and what steps may be taken to avoid having a child with KD. The genetic counselor explained to me that one of my two "X" chromosomes has a longer repetition of CAG. So, if the mother is a carrier like me, the following possibilities exist for her offspring:

- 25% chance that she will have a girl that will be a carrier of KD
- 25% chance that she will have a girl with no links to KD (*healthy genes*).

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The KDA Conference

Just around the corner



The 2010 conference and symposium will be here before you know it. This year's theme is "Celebrating Ten Years: Closer than Ever Before". The conference will officially begin on Wednesday, November 10 and run through Friday, November 12 at the Holiday Inn - Bayside, San Diego, CA. The proposed agenda, registration form, hotel block and other information can be found at [KDA Conference](#).

UPDATE: The Kennedy's Disease Assoc. Conference Speaker's line-up is complete! All speakers are in place and what a line-up we will have this year in San Diego! However, the unexpected takeaway will be the lifelong friends you will make while attending the conference. We hope to see you there! Follow this link to see the [agenda](#).

- **Diane Merry PhD**, Thomas Jefferson University, creator of the first KD Mouse Model will once again be joining us for our annual conference! I do not believe Dr. Merry has missed a conference yet! We look forward to seeing her again!
- **Albert R. La Spada, MD, PhD**, Chief of Genetics at UC San Diego and co-discoverer of the KDA Gene with Dr. K. Fischbeck, National Institutes of Health, will also be joining this already amazing line-up of esteemed KD researchers and doctors at the KDA Conference 2010.

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Charles - continued from page 1

- 25% chance that she will have a boy that will have KD
- 25% chance that she will have a boy that will have no links to KD (*healthy genes*)

Given these odds, I felt heartbroken and had to do a lot of soul searching and research. Every couple that is faced with the fact that they have Kennedy's Disease in their family will have to deal with this harsh reality.

The Genetic counselor explained that being a carrier of KD is not the end of the world and that I had options.

Preconception options include in vitro fertilization or IVF, egg donation, and pre-implantation genetic diagnosis or PGD. This is when doctors can test healthy eggs and select the embryos that are from the X chromosome not containing KD. These healthy eggs can then be used in the IVF process. If a woman would prefer to become pregnant naturally, there are **other options available** including testing a fetus to see if it has the defective gene. These include ultrasounds, chronic villus sampling (CVS), and amniocentesis. Ultrasounds can be used to determine the sex of the child eliminating the chances of the fetus developing KD. CVS and amniocentesis can determine gender, as well as, test the child for the lengthened fragment associated with KD. These tests then can be used throughout the pregnancy to examine the unborn child.

Another option all together was adoption. There are support groups for parents who are faced with these different options; however, everyone will have to make their decision based upon their own beliefs and thoughts.

In the summer of 2007, I found myself in the position of being unexpectedly pregnant. I was a thrilled yet concerned about my unborn child. I chose not to have any of the tests done on my baby during pregnancy due to the risks the tests presented. My husband and I also discussed that if the tests came back positive we would not terminate the pregnancy, so there was no reason to take these risks. In January 2008, I gave birth to our beautiful baby boy. Sometimes I feel guilty knowing that I might have passed along the KD gene; however, I would not have it any other way. During the last couple of years, I have come to the realization that you have to have faith in God and in science. Kennedy's Disease has only been recognized for less than 20 years. During this time, huge advancements have been made to understand the disease while looking for a treatment.

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- **Maria Pennuto**, PhD, post-doc at NIH, now at the Italian Institute of Technology in Genoa will also be attending the Conference in San Diego this year! She has found that having extra IGF1 genes seems to improve muscle strength and function in mice bred to have an SBMA-like disease. She has been awarded KDA grants totaling \$45,000.
- **Dr. Angelo Poletti**, from University of Milan, Italy, who researches Kennedy's Disease across the 'pond' will also be joining us for the dinner banquet where you can ask him questions face-to-face (along with other researchers) and he will join the private brainstorming session!
- **Udai Pandey**, PhD, Assistant Professor of Genetics at Louisiana State University Health Sciences Center will also be attending and presenting. He is also a past recipient of KDA Research Grants totaling \$50,000.
- **Heather Montie**, PhD (a KD carrier herself) will be presenting at the 2010 Conference in Nov.! She met dedicated KD researcher Dr. Diane Merry (T. Jefferson University), through the KDA years ago when she was a high school student (her father was diagnosed with KD). She decided she wanted to research KD back then and today she is working diligently on behalf of all KDRs! The KDA was pleased to award her a \$50,000 grant towards her research in 2008 to investigate the androgen receptor (AR). This is the protein that is altered in Kennedy's Disease.
- We just received confirmation that **Kenneth H. Fischbeck**, MD, Distinguished Investigator & Co-Founder of the KD gene, **Angela Kokkinis**, **Chris Grunseich**, researcher and **Alice Schindler**, Genetic Counselor from NIH (National Institutes of Health); will be attending the conference.
- **J. Paul Taylor**, MD, PhD - KD Researcher from St. Jude's Children's Research Hospital will be attending and presenting at the KDA Conference in November!

Should you have any questions, please contact us at infor@kennedysdisease.org. We hope to see you at the conference!

Charles - continued from page 2

Call me naïve, but I believe God gave me this child for a purpose; and I believe He will do what He can to protect my baby and myself from pain and suffering. God has a plan for each of us and I believe that having my baby, Charles, was part of His plan for me.

For more information about Genetic Counseling, please click on the following links: The [National Society of Genetic Counselors](#), the [National Institutes of Health](#), or the [Kennedy's Disease Association](#).

KDA Tissue Donation Guide

Update



Updated Guide: If it has been some time since you read the guide, or if you are considering donating tissue for Kennedy's Disease research, the guide was recently updated (May 04, 2010). You can download the latest version at

<http://www.kennedysdisease.org/tissuedonationprogram.html>.

Background: Doctors, researchers, and scientists have long recognized the benefit of human tissue to further their research. Kennedy's Disease research is no different. In response to this need, the Kennedy's Disease Association (KDA) has established a Tissue Donation Program for individuals with Kennedy's Disease who wish to donate their tissue. This program is similar to an organ donation program, but is different because of the formalities required and the specific use for the tissue.

Storage and Use: Tissue will be stored at a KDA approved tissue storage facility and will be provided for biochemical and genetic studies. Tissue samples and results of clinical testing will be made available to qualified scientists only after their research proposal is reviewed and approved by the KDA Scientific Review Board (SRB) and Board of Directors. The KDA is hopeful that these studies will lead to advances in the diagnosis, treatment, and eventual cure of Kennedy's Disease.

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Susanne's Corner

Notes and Thoughts from Susanne Waite



1. Beth DuVall completed her half marathon!

She pulled a muscle early in the race, but she said that her mother and KD'rs remained her inspiration to stay the course and cross the finish line. Beth raised over \$4,000 for the KDA through her sponsorships. Thank you to everyone who helped Beth meet her goals and a **HUGE THANK YOU** to Beth for her helping the KDA in this special way! For more information, check out [Beth's page](#) on Razoo.

2. Awareness Candles Support the KDA



Sabrina Basham (daughter of Bill Basham who lives with KD) has come up with an 'enlightening' idea! She makes soy based candles and has decided to sell 'Awareness' candles - one for Kennedy's Disease, one for Breast Cancer and one for the Humane Society. Order your's now - \$12! Proceeds go to the cause of your candle! For more information, check out [Sabrina's website](#) or her [Facebook page](#).

3. Shopping to Help the KDA

The KDA "[Proceeds Partners](#)" YTD have sent us checks for \$607.33 just because many of you shopped through a link from the KDA site! It did not cost a thing extra to anyone. Susanne's testimonial:

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Family Involvement: Since any donation decision affects others, we recommend that you discuss it with your family so that they will understand and support your decision. If you are interested in donating tissue for Kennedy's Disease research, please download the guide. It will outline the program and includes the forms needed to make a tissue donation. Questions should be emailed to info@kennedysdisease.org.

Important: Pre-planning is critical. If you wish to be a donor, certain paperwork must be signed and arrangements need to be made with the hospital and pathologist ahead of time. If these decisions are delayed until the actual time for donation, the chance of a successful donation are slim.

Running

For Those Who Can't

Thank God for Family and Friends!

They are the Angels who lift us up to our feet when our own wings have trouble remembering how to fly...

Our most recent fundraising angel is a member of my family! We are proud and thankful to our nephew, Dave Heraper, as he prepares for a half-marathon in November at Malibu, CA. This race has been tagged "Run to Paradise" and it will be paradise for the Kennedy's Disease Association (KDA) to receive donations we desperately need for research.

It is common for these types of races to pre-select local charities to receive proceeds. However, any participant may choose to run and self-promote for a specific charity of their choosing. If you have a family member or friend who loves competitive sports, please encourage them to participate and raise funds for KDA. Just notify us and we are happy to help.

Anyone wishing to sponsor Dave Heraper in the Malibu Marathon may send a check directly to the Kennedy's Disease Association. We are also setting up a Razoo donation page for those wishing to use their credit card for a donation. There will be a direct link on the KDA website as well as a Facebook promotion and direct link. Thank you in advance for your generous support.



Thank you to everyone running for those who can't!

"I shop Amazon, Giftbaskets.com, Flowerpetal.com and others through the KDA every time I am buying a gift for family, friends and clients (and, even gifts for myself!) Do not forget the holidays are coming up - avoid the crowds as I do - shop the KDA Proceed Partners and help find a cure! Let your friends and family know about this program also!"

5. Supporting the KDA through Van Bourgondien's Fundraising Program



To our garden and bulb lovers: whether your garden is a 1/2 acre or a container garden on a deck, the KDA is holding a fundraiser with [Van Bourgondien Dutch Bulbs](#). The company is giving 50% of each dollar spent to the KDA. This is perfect time to buy flower bulbs (Tulips, and the like). These bulbs are best planted in the fall before the first frost starts. For Tulips, if in a warm climate, they have to be "forced" in the refrigerator before planting. They require so many days below a certain temperature to bloom in the spring. I am also going to post this on the KDA and my Facebook pages to see what kind of sales we can generate. We have set a goal of \$2,000. Will you help us get there? Also, please share this information with your friends and family.

KDA e-Xpress

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Editor: Bruce Gaughran

Comments, suggestions, and questions should be sent to:

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Access to Medical Care

For Persons with Mobility Disabilities



This [web article](#) was published recently in MDA's Quest Magazine. The government publication it references is quite good. The Q&A section was very interesting because it explained the responsibilities of a medical facility and health care providers in regards to adhering to the requirements of the Americans with Disability Act (ADA). Most new facilities comply with these requirements, but many family practices are in older buildings and do not have adequate room or even the ability to make major modifications to their facility. Most family doctors' offices have a ramp today, but not every office provides reasonable access to all rooms. If you have problems with accessing your doctor's office, give a copy of this guide to your doctor in hopes he will consider these recommendations.

Below is the web article as well as a couple of sections from the publication itself.

“Improving accessibility of health care facilities”

The Department of Health and Human Services Office for Civil Rights, in conjunction with the Department of Justice's Civil Rights Division, has produced a free publication titled *Access to Medical Care for Persons with Mobility Disabilities*.

The 19-page document contains technical guidance for making health care facilities more accessible for patients with disabilities. Although geared toward health care providers, individuals with disabilities also are welcome to the publication, either for themselves or to give to their doctors.

Included in the publication are an extensive question-and-answer section; illustrations of accessible medical equipment and examination rooms; and an overview of ADA (Americans with Disability Act) requirements.

Title III of the ADA prohibits discrimination on the basis of disability by private hospitals, physicians' offices, clinics and other health care providers. The Rehabilitation Act of 1973 prohibits such discrimination by health care providers that receive federal financial assistance.

To download a copy of the publication from the ADA website, go to http://www.ada.gov/medcare_ta.htm.

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The Younger Generation Needs to Step Up

Bruce Gaughran



I believe the KDA needs some younger people to step up (*become more active in the association*). My concern stems from the fact that the average age of the Kennedy's Disease Association (KDA) board of directors is getting up there. Half of the board members are in the sixties and until two weeks ago the youngest member was in the mid-forties. Furthermore, the number of active committee members has declined over the last few years.

I know I am probably overreacting (*as my parents did when the Beatles were introduced to America on the Ed Sullivan show*). I recognize, however, that with the average age of the board members increasing and our physical capabilities declining that we need some younger blood and a more diverse board (*ethnically as well as gender*). Board members have also had to increase their involvement in committee activities because participation from volunteers has dwindled. *Note: Just because I mentioned the younger generation in my header, that does not mean we will turn down more mature individuals who are willing to help.*

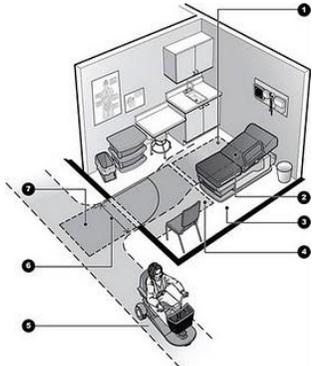
I know that one of the best moves I ever made was joining the KDA. Ranking right up there with 'joining' was when I became a board member. I feel blessed to be able to work with this great team. Everyone contributes in his or her own way. The diversity of opinion and level of involvement varies, but the enthusiasm and support never wavers.

Since the KDA is an all-volunteer organization, we also have a need for people to serve on its various committees. The current committees are:

- **Fund Raising** – This is still the lifeblood of the KDA. Without donations and other financial support, we cannot support research and education.
- **Public Relations** – The KDA does not receive enough press. We need people who can write press releases and help with the newsletters.
- **Conference** – This team plans and coordinates our educational conferences and symposiums.

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To be sent a copy, call the ADA Information Line at (800) 514-0301."



The publication begins by stating, "Both Title II and Title III of the ADA and Section 504 require that medical care providers provide individuals with disabilities:

- full and equal access to their health care services and facilities; and
- reasonable modifications to policies, practices, and procedures when necessary to make health care services fully available to individuals with disabilities, unless the modifications would fundamentally alter the nature of the services (i.e. alter the essential nature of the services)."



The last section of the publication discusses Staff Training. "A critical, but often overlooked component to ensuring success is adequate and ongoing training of medical practitioners and staff. Purchasing accessible medical equipment will not provide access if no one knows how to operate it. Staff must also know which examination and procedure rooms are accessible and where portable accessible medical equipment is stored. Whenever new equipment to provide accessible care is received, staff should be immediately trained on its proper use and maintenance. New staff should receive training as soon as they come on the job and all staff should undergo periodic refresher training during each year.

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- Education and Research – We are always in need of people to search the internet for current research and other articles/resources that could benefit those living with Kennedy's Disease.
- Support Groups (Subgroups include Carriers, Family Members and Friends, Wives and Significant Others, Support Forums, and Individuals with Symptoms) – These subgroups support those living with Kennedy's Disease. These volunteers respond to forum inquiries, host chat rooms, and provide support to those in need. I am certain this committee could do so much more to help if they had more volunteers.

Below are some general questions concerning the board and committee positions. If you have any specific questions or concerns, please do not hesitate to ask.

What committees would I serve on or chair? It depends upon your interest and skill set. You are not assigned to a committee; you choose one that you are interested in.

How much time will my committee involvement take up? It depends on many factors. If you are on the conference planning committee, it takes some weekly involvement (about 1-2 hours a week) for about three months and then some occasional involvement for about two months. Some committees like fund raising take more time (*perhaps up to 2-to-4 hours a month*). Time involvement is based more around the desires of the person serving.

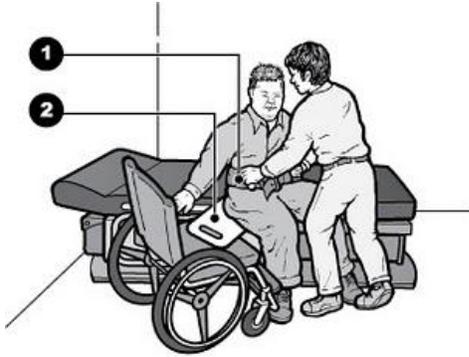
Is any travel involved? No travel is involved to serve on any committee.

What other roles might I be involved in? That depends upon the circumstance and your desired level of involvement. For example, a couple of years ago we decided to move our data off-site to a server with remote backup. This took a small amount of time, but nothing substantial. We have two people that quarterly audit the financial transactions/processes of our treasurer.

I am working full time. I am not certain how much time I can afford to give. Is that okay? Several board and committee members are working full time. They are not expected to volunteer a lot of time to perform regular KDA work, but they can still serve on committees and work on projects.

What are you looking for in board and committee members? As mentioned above, we are looking for diversity of opinion, fresh ideas and people willing to help the KDA become a more viable organization.

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Finally, training staff to properly assist with transfers and lifts, and to use positioning aids correctly will minimize the chance of injury for both patients and staff. Staff should be instructed to ask patients with disabilities if they need help before providing assistance and, if they do, how best they can help. People with mobility disabilities are not all the same - they use mobility devices of different types, sizes and weight, transfer in different ways, and have varying levels of physical ability. Make sure that staff know, especially if they are unsure, that it is not only permissible, but encouraged, to ask questions. Understanding what assistance, if any, is needed and how to provide it, will go a long way toward providing safe and accessible health care for people with mobility disabilities."

If reasonable access is, or potentially could be, an issue for you, download the PDF or call the number provided for a hard copy. Being disabled is difficult enough. We should not be further handicapped by less than adequate medical facilities.

Facebook

Online Social Networking

What began in 2004 as a network strictly for Harvard college students, followed by other Ivy League schools, has now become a most popular site for anyone with a valid email address. Facebook members may choose their personal levels of security to prevent strangers from accessing their personal information. Many users prefer to only publish what is already public information or (my personal rule of thumb) anything they wouldn't mind seeing on the front page of tomorrow's newspaper.

With 500 million members, as of July 2010, the value of connections for a non-profit like the Kennedy's Disease Association is priceless. Many of our members already use Facebook and we are encouraging everyone to consider doing the same. It's not just for students anymore! There is real marketing benefit in being able to send a message to all of your Facebook friends and their friends...and their friends...

[Visit the KDA](#) on Facebook.

Is there any compensation or benefits? I will not get into all the wonderful benefits (*no compensation and rarely a thank you*). Personally speaking, the rewards have been wonderful and I still feel it is a great opportunity and one that I thoroughly enjoy.

If I have not sold you yet on this challenging and exciting opportunity, I probably never will. Nevertheless, I had to try. I look forward to hearing from you.

Sean Blasko

Joins KDA Board of Directors



Sean Blasko became aware of Kennedy's Disease when his Uncle, Mike Goynes, was diagnosed in 1995. Sean first became involved with the KDA when he competed in an Ironman triathlon in Australia to raise money for KDA research. He then joined the fundraising committee and continued to hold annual fundraisers. Sean joined the KDA Board of Directors in 2010.

Sean was born in San Antonio, Texas and later moved to Austin where he received his B.S. in Mechanical Engineering in 2001 from the University of Texas. Sean later attended University of Texas School of Law where he received his J.D. in 2008. Sean currently lives in San Antonio and his legal practice is focused on tax, estate planning, and intellectual property law.

When you have a chance, please welcome Sean Blasko to the KDA team.

