



Kennedy's Disease Association Questions and Answers

TABLE OF CONTENTS

Section I: KENNEDY'S DISEASE

Q: What is Kennedy's Disease?	3
Q: What are some of the symptoms of Kennedy's Disease?	3
Q: When do you first start seeing signs of the disease?	3
Q: Who gets Kennedy's Disease?	3
Q: Is there a treatment or cure for Kennedy's Disease?	3
Q: What causes the symptoms?	3
Q: What causes the muscle cells to die?	3
Q: What causes the motor neurons to die?	4
Q: Why can't the cell recycle proteins?	4
Q: How is Kennedy's Disease diagnosed?	5
Q: Why is Kennedy's Disease so often misdiagnosed?	5
Q: Why don't we hear much about Kennedy's Disease?	5
Q: Can the disease be passed on to my children?	5
Q: One of my parents has the defective gene. I have or am considering having children. Should I be tested or should I have my children tested?.....	5
Q: I am considering having surgery. Is there any information about Kennedy's Disease that I should make my doctor or anesthesiologist aware of before hand?.....	5
Q: I have Kennedy's Disease. Should I carry a medical card in my wallet?.....	6
Q: Is there a brochure I can give my doctor on Kennedy's Disease?	6

Section II: KENNEDY'S DISEASE ASSOCIATION

Q: What is the Kennedy's Disease Association?	6
Q: Why was the KDA formed?.....	6
Q: What are the objectives of the KDA?	6
Q: What services does the KDA provide?	6
Q: How many members does the KDA have?	7
Q: How does the KDA operate?	7
Q: Who are the researchers on the Scientific Review Board?.....	7
Q: How is the KDA funded?	7
Q: Does the KDA help support Kennedy's Disease research?	7
Q: Besides supporting research, how are the donations used?.....	8
Q: How are research grants awarded?	8
Q: Who has received KDA research grants?	8
Q: How does the Tissue Donation Program work? Is the tissue ever sold?	9
Q: How do I join the KDA?	9
Q: What is the KDA chat room?	10
Q: Do you keep copies of past chats with doctors, researchers, and others?	10



Kennedy's Disease Association Questions and Answers

Q: Is there a place I can ask others questions or share my experiences? 10
Q: I heard that exercise was bad for a person with KDA. Is that true? 10
Q: I have Kennedy's Disease and am considering going on disability. I have heard that applying for and being awarded Social Security – Disability compensation is quite difficult. Where can I find help for this? 10
Q: What is the KDA memorial page all about? 11
Q: How do I donate to the KDA? 11
Q: How do I contact the KDA?..... 11



Kennedy's Disease Association Questions and Answers

Section I: KENNEDY'S DISEASE

Q: What is Kennedy's Disease?

A: Kennedy's Disease (also known as Spinal Bulbar Muscular Atrophy, SBMA, or Kennedy's Syndrome) is a rare and currently incurable and non-treatable X-linked recessive genetic progressive neuro-muscular disease. Both the spinal and bulbar neurons are affected causing muscle weakness and wasting (atrophy) throughout the body which is most noticeable in the extremities (legs/arms), it is also especially noticeable in the face and throat, and causes speech and swallowing difficulties, along with major muscle cramps as well as other symptoms.

Q: What are some of the symptoms of Kennedy's Disease?

A: See the following link: [KD Symptoms](#)

Q: When do you first start seeing signs of the disease?

A: Generally, symptoms begin to appear in the late 20's or early 30's. However, there have been cases where the symptoms showed up in the late teens or as late as in the 60's.

Q: Who gets Kennedy's Disease?

A: Kennedy's Disease is a genetic disease, passed on from generation to generation in a family. It is an X-linked recessive inherited gene. Generally males who inherit the gene exhibit symptoms, while females who inherit the gene are usually just carriers. Although, in rare instances, females may also exhibit symptoms.

Q: Is there a treatment or cure for Kennedy's Disease?

A: Currently there is no treatment or cure. Recent research, however, show some promising signs. There is currently a clinical drug trial taking place at the National Institute of Health in Bethesda, MD. Read more about the trial at this link: [Clinical Trial](#). Read about other research that is taking place at this link: [Research](#).

Q: What causes the symptoms?

A: The primary symptom of KD, muscle weakness, is due to the death of muscle cells. The loss of these cells means that there are fewer muscle cells to contract and thus the contraction of the muscle is weaker.

Q. What causes the muscle cells to die?

A. The direct cause of the muscle cell death is believed to be the death of the nerve cells that control the contraction of the muscle cells. These nerve cells are known as motor neurons. Motor neurons are the cells that connect the brain to the muscle cells. When you wish to contract a muscle to pick up a pencil, for example, your brain sends signals to the motor neurons that control those muscles. The motor neurons then convey the



Kennedy's Disease Association Questions and Answers

signal to the muscle cells and the muscle cells contract, performing the movement that you envisioned in your brain. In KD, the motor neurons die and so the connection from the brain to the muscle cells is broken. As a direct result of the loss of the motor neuron, the muscle cells die as well. Whenever muscle cells lose their motor neuron, they will usually die. It is generally thought that KD does not directly cause the muscle cells to die although there is some recent evidence that KD may result in some changes in the muscle.

Q. What causes the motor neurons to die?

This is the million dollar question. The straight answer is that we really do not know for sure. There is a lot of evidence that the motor neuron cell death may be due to the inability of the cell from KD patients to adequately recycle proteins and this results in the build up of old, trashy proteins. This build up of trash is believed to somehow be toxic to the cell.

Q. Why can't the cell recycle proteins?

KD is a genetic disease – this means that KD patients have a defective gene. The function of genes is to tell the cell how to make a specific protein and a cell can only make proteins for which there is a gene. For example, we all have a gene to tell our cells how to make hemoglobin, the protein that carries oxygen in our blood. Without that gene, our cells would not be able to make any hemoglobin. Since we need hemoglobin to live, such an individual would never even be born. However, occasionally, a gene for a specific protein may be changed so that the cells would make an altered form of the protein. This altered protein may still work, but possibly not as well. This is what happens in sickle cell anemia. The hemoglobin gene has been altered and the hemoglobin made does not work as well.

Since KD is a genetic disease, patients with KD must have an altered gene. The gene that is altered is the one that tells the cells how to make the protein known as the androgen receptor (AR). The normal function of the AR is to mediate the actions of testosterone. Without the AR, testosterone would have no effect on a cell. In men with KD, the AR that is made is altered. It still works but sometimes not as well. The current thinking is that the problem with those with KD, however, is that the altered AR cannot be removed by the cell. Typically, once used, the AR is destroyed by the cell. This is done by processes that help remove all proteins. Even worse, when the cell tries to remove the AR, the altered form of AR jams up these cellular processes preventing the cells from removing any proteins. Apparently, the nerve cell must be able to remove proteins to survive, so the overall effect of the jamming is to kill the cells. Much of the research on KD right now involves investigating ways to 'un-jam' these protein removal mechanisms and prevent the death of the cell.



Kennedy's Disease Association Questions and Answers

Q: How is Kennedy's Disease diagnosed?

A: Fortunately, there is a simple blood test today that checks a person's DNA for the defective chromosome. Almost any DNA testing laboratory can perform the test. A doctor or nurse can draw the blood and send it off to the laboratory. Test results are normally returned within three-to-six weeks.

Q: Why is Kennedy's Disease so often misdiagnosed?

A: Until the 1990's, there was no specific test for Kennedy's Disease. The most frequent misdiagnosis has been ALS (Lou Gehrig's Disease). The Muscular Dystrophy has posted an article written by Dr. Kenneth Fischbeck in this regard: [Mistaken Diagnosis? ALS and SBMA Can Be Confused](#). Other misdiagnoses that some individuals with Kennedy's Disease have received prior to Kennedy's Disease being determined have been: [Spinal Muscular Atrophy III - Kugelberg-Welander](#), [Glycogen Storage Disease](#), [Guillian's Barre](#), [Myasthenia Gravis](#), and [Multiple Sclerosis](#).

Q: Why don't we hear much about Kennedy's Disease?

A: Kennedy's Disease is considered a rare medical disorder. It is estimated that only 1 in 40,000 individuals worldwide have Kennedy's Disease. However, many go misdiagnosed or undiagnosed for years.

Q: Can the disease be passed on to my children?

A: If the mother is the only carrier of the defective gene: there is a 50 % chance of passing the affected gene on to male children (in which case, they would develop symptoms in adulthood). The chance of passing the defective gene on to female children is also 50% (in which case, they would be carriers of the gene, but probably never develop actual symptoms). If the father is the only carrier of the defective gene the chances of passing the defective gene to a male child are 0%. However, it is 100 % certain that the gene will be passed on to a female child, and she will be a carrier of the gene.

Q: One of my parents has the defective gene. I have or am considering having children. Should I be tested or should I have my children tested?

A: This is a question that is asked quite often. Some families have even considered prenatal testing. The KDA recommends that if you have this concern, you should consult with a genetic counselor.

Q: I am considering having surgery. Is there any information about Kennedy's Disease that I should make my doctor or anesthesiologist aware of before hand?

A. Yes, the KDA recommends that you print the information found on our web site and review it with your surgeon. The links and articles provide information and warnings that should be taken into account before surgery. You can review and print relevant information at the following link: [Surgery Information](#).



Kennedy's Disease Association Questions and Answers

Q: I have Kennedy's Disease. Should I carry a medical card in my wallet?

A: Yes. It would be quite helpful for an emergency team to know about your physical condition. You can print a medical card at the following link: [Medical Card](#).

The KDA also recommends that you have a medical history form on file at your doctor's office and your local hospital. When completed, the form is an excellent resource for medical professionals and could possibly save your life. It would also be good to carry a copy with you on trips. You can print a copy of the form at the following link: [Medical History Form](#).

Q: Is there a brochure I can give my doctor on Kennedy's Disease?

A: A comprehensive easy to read guide on Kennedy's Disease that explains what it is, how it is contracted, what the symptoms are, etc. is available. Click on these links to see the brochure: [KDA Brochure Front](#) [KDA Brochure Back](#)

Section II: KENNEDY'S DISEASE ASSOCIATION

Q: What is the Kennedy's Disease Association?

A: The Kennedy's Disease Association (KDA) is a totally volunteer non-profit corporation, incorporated in California on Monday, August 21, 2000. The KDA is recognized under United States of America Internal Revenue Code 501(c)3 as a publicly supported organization as described in sections 509(a)1 and 170(b)1(A)(vi).

Q: Why was the KDA formed?

A: Read the KDA Story at this link: [The History of the KDA](#).

Q: What are the objectives of the KDA?

A: The Kennedy's Disease Association has been formed with the following objectives in mind:

1. Financially support and promote medical research to find a cure for Kennedy's Disease
2. Improve awareness of Kennedy's Disease in the medical community
3. Create a support system for those living with Kennedy's Disease
4. Increase public awareness about Kennedy's Disease and its effects on families
5. Share information about Kennedy's Disease for those who seek it

Q: What services does the KDA provide?

A: Click on the following link: [KDA Services](#)



Kennedy's Disease Association

Questions and Answers

Q: How many members does the KDA have?

A: As of June, 2007, the KDA has 787 registered associates living in thirty-three countries. 595 of our associates carry the defective gene. The KDA also has 46 doctors and researchers on its distribution list.

Q: How does the KDA operate?

A: The KDA has a board of directors, elected officers, and a Scientific Review Board (SRB). All volunteer their services. The SRB is made up of four researchers and two other members representing the business and academic communities. The researchers are all involved in Kennedy's Disease research. Its roll is to review current research, provide direction for the funding of research grants, and be a resource to the board of directors.

Q: Who are the researchers on the Scientific Review Board?

A:

- Dr. Kenneth Fischbeck – Bethesda, MD - National Institute of Health. A leading researcher, professor and educator on the subject of Kennedy's Disease; a co-discoverer of the Spinal Bulbar Muscular Atrophy gene.
- Dr. Diane Merry – Philadelphia, PA - Thomas Jefferson University. Dedicates research team to Spinal Bulbar Muscular Atrophy.
- Dr. Al La Spada – Seattle, WA - University of Seattle. Co-discovered the Spinal Bulbar Muscular Atrophy gene and continues to research Kennedy's Disease.
- Dr. Andrew P. Lieberman – Ann Arbor, MI - University of Michigan. Researcher in the area of inherited neurodegenerative diseases. Director of the Neuropathology Core for the Michigan Alzheimer's Disease Research Center.

Q: How is the KDA funded?

A: The KDA is funded by donations and the sale of certain products including cook books, coffee mugs, calendars, etc. Locally sponsored fundraising events are especially helpful in supporting the KDA.

Q: Does the KDA help support Kennedy's Disease research?

A: Yes, in several ways. First, over the past four years, the KDA awarded \$125,000 in research grants. It also funds KDA conferences where researchers and those afflicted with the disease come together to share experiences, current research, and knowledge. The KDA financially supports certain Kennedy's Disease Research conferences. It also worked closely with the National Institute of Health (NIH) to provide preliminary information on the feasibility of the Kennedy's Disease clinical drug trial that is currently in progress. It supports several Kennedy's Disease research labs across the country by providing tissue samples when human cells are needed to support research and testing. Kennedy's Disease tissue is currently stored at the University of Michigan Tissue Bank.



Kennedy's Disease Association Questions and Answers

Q: Besides supporting research, how are the donations used?

A: Because the KDA is a totally volunteer organization, over the last three years 89¢ of every dollar went towards Kennedy's Disease Research Grants (68¢) and Education (21¢). During the same period, the KDA also spent \$30,000 on Kennedy's Disease education that included the mailing of 12,000 information packets to Neurologists, the funding of three KDA conferences, and financially supporting certain Kennedy's Disease Research conferences. The KDA only spent \$15,619 (11¢ of every dollar) on Operating Expenses. These expenses average approximately \$5,300 a year and include the maintenance of our KDA web site, our message forum, the twice a month chat rooms, office supplies, office equipment, postage, insurance premiums, banking fees, ISP charges, etc. To read more about where the money is spent see the following link: [Where Does the Money Go?](#)

Q: How are research grants awarded?

A: Because the KDA is relatively small and funding is limited, our focus in recent years has been to provide "seed-money" to post-doc and other young researchers who do not currently have the funding or credentials to receive funding from larger organizations such as the National Institute of Health or the MDA. This "seed-money" normally provides the researcher an opportunity to further his/her research while giving him/her time to apply for other grants. The Scientific Review Board reviews all applications. The reviewers focus on research projects that are specific to or could be used in finding a treatment or cure for Kennedy's Disease. The reviewers ask three or four of the applicants to submit full grant applications. The Scientific Review Board reviews the finalists and recommends to the Board of Directors which applicant(s) should receive research funding.

Q: Who has received KDA research grants?

A: In 2006, two research grants were funded:

- A \$25,000 grant was awarded to Chawnshang Chang Ph.D. from the University of Rochester. His research plans to develop a treatment regimen for Kennedy's Disease targeting the poly Q-expanded mutant AR. This concept may be a way to cure the disease.
- Another \$25,000 grant was awarded to Udai Bhan Pandey Ph.D. from the University of Pennsylvania. Dr. Pandey proposes to use molecular genetic approaches in *Drosophila* to characterize the mechanism of suppression by HDAC6. His long-term goal is to contribute to the development of therapeutic interventions for Kennedy's Disease.
- In 2005, one research grant was funded:
 - A \$25,000 emergency funding grant was awarded to J. Paul Taylor, MD, Ph.D. from the University of Pennsylvania. The grant helped support Dr. Taylor and his team's research using the *Drosophila melanogaster* (fruitfly) model system to investigate the molecular pathogenesis of Spinal and Bulbar Muscular Atrophy (aka Kennedy's Disease). In response to the KDA's grant, we received the



Kennedy's Disease Association Questions and Answers

following email from Dr. Taylor referencing the status of his current Kennedy's Disease research. *"... This (grant) could be a life saver. We have made great strides with our work; in fact, we have a manuscript on our Kennedy's Disease work that has received good reviews. This work was largely funded by (the) KDA and I have been anxiously waiting for this work to be accepted for publication before alerting you. I have also had two graduate students join my lab who are doing their Ph.D. thesis work on Kennedy's Disease.*

- In 2004, one research grant was funded:
 - A \$25,000 grant was awarded to Andrew Lieberman, MD, Ph.D., University of Michigan for development of the first ever Kennedy's Disease Knock-In Mouse Model. *"Thanks so much for all of your hard work on our behalf! It sure is inspirational for us to know that the KDA cares enough about this project to launch a difficult fund raising drive to support our work, and it's really gratifying to see that the generosity of the KDA membership made these efforts successful so quickly."* -- Dr. Andrew Lieberman
- In 2003, one research grant was funded:
 - A \$25,000 grant was awarded to J. Paul Taylor, MD, PhD, University of Pennsylvania for developing the *Drosophila melanogaster* (fruitfly) model system to investigate the molecular pathogenesis of Spinal and Bulbar Muscular Atrophy.

Q: How does the Tissue Donation Program work? Is the tissue ever sold?

A: 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 KDA has created a program for interested families to donate tissue for Kennedy's Disease research. The program is similar to an organ donation program, but is different because of the formalities required and the specific use for the tissue. Tissue is stored at a KDA approved tissue storage facility and is provided to qualified laboratories 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. **Tissue is never sold.** Tissue is only provided to qualified laboratories that are engaged in Kennedy's Disease research. You can read more about the Tissue Donation Program by clicking on the following link: [Tissue Donation](#).

Q: How do I join the KDA?

A: There are two ways to join. Go to the following link: [Join The KDA](#). Or, write or send an email to the KDA providing your name, address, age, phone number, email address, and advise us if you have KD or are a carrier. The email can be sent to: info@kennedysdisease.org. The mailing address is: Kennedy's Disease Association, Inc., P.O. Box 1105, Coarsegold, CA 93614-1105 (U.S.A.)



Kennedy's Disease Association Questions and Answers

Q: What is the KDA chat room?

A: The Kennedy's Disease chat room was designed to be a cost-effective way for anyone who would like to discuss living with KD. Chats are scheduled for the first and third Saturday of each month at 10:30 A.M. Eastern Time. You must register before joining in on a chat. You can register and also log on to a chat by clicking on the following link: [KDA Chat Room](#).

Q: Do you keep copies of past chats with doctors, researchers, and others?

A: If you missed a chat, you can read the transcripts at: [Chat Transcripts](#).

Q: Is there a place I can ask others questions or share my experiences?

A: Yes, the KDA has a forum. There are many topics posted covering a variety of subjects. The first time you visit the forum you must register. The forum is located at the following link: [KDA Forum](#). The benefits of this service include:

- No advertising/pop-ups.
- Email notification when someone has replied to your message.
- Improved ease of use, message handling and storage.
- Private messaging is a means for community members to talk to each other outside the realm of the public community. Members never have to know each other's email, so this is a more secure way of having a private conversation within a community. Multiple members can participate in a private message.

Q: I heard that exercise was bad for a person with KDA. Is that true?

A: No, in fact, several neurologists confirm that light and 'smart' exercising is good for your muscles and motor neurons because it stimulates them and keeps them functioning longer. Another benefit of exercise and stretching is that it can cause 'cell inhibition'. However, any type of activity that overly taxes your muscles could be detrimental to your condition. The key is to just do what the body feels comfortable doing and to never exceed your capabilities. The goal is to stimulate the healthy muscles and motor neurons without harming them. Remember to always consult with your doctor prior to beginning any exercise program. There is an exercise guide available at the following link: [Smart Exercise Guide](#).

Q: I have Kennedy's Disease and am considering going on disability. I have heard that applying for and being awarded Social Security – Disability compensation is quite difficult. Where can I find help for this?

A: The KDA has developed a guide for applying for Social Security – Disability (SS-D). Feedback from associates who have used the guide have been very positive. However, this information guide should not be used as the only source for preparing your Social Security - Disability application. It is recommended that you consult with the Social Security Administration, your neurologist, and if desired, a legal professional before starting the application process. In developing the guide, the Social Security Administration's website (<http://www.ssa.gov/disability/>) was used for much of the



Kennedy's Disease Association Questions and Answers

format and information provided within. For an overview of the process, click on the following link: [SS-D Tips](#). Click on the following link to read the SS-D guide: [SS-D Process Guide](#).

Q: What is the KDA memorial page all about?

A: The KDA memorial page honors those who have left us. If you have a loved one with Kennedy's Disease who has passed away and would like to have him/her added to the memorial page, e-mail the information to the KDA and, if possible, include a picture. If you do not have a picture on your computer to send via e-mail, you can mail it to us via regular US mail. The KDA can scan it and return the original to you.

Q: How do I donate to the KDA?

A: Here are 5 ways that you can help the KDA:

1. [Click here to make a credit card donation](#) through the [Secure Server](#) at "Network for Good". Network for good is a service that processes and routes credit card payments to non-profits. They do take 4.75% of the donation as a processing fee. To read more about Network for good and its policies [Click Here](#).
2. Send a check or money order tax-deductible donation directly to the KDA.
3. Make a stock transfer through www.ameritrade.com. Contact us at info@kennedysdisease.org to get more information regarding this type of transaction.
4. Remember the KDA in your will, living trust or memorial request.
5. Get involved with the Kennedy's Disease Association. Donate your time or services.

Q: How do I volunteer at the KDA?

A: Since we are an 'all volunteer' organization, we are always looking for volunteers for our committees such as Fundraising and Support Groups. If you are interested in volunteering, please contact the KDA using one of the methods listed below. Any service you can provide will be much appreciated.

Q: How do I contact the KDA?

A: You can call, mail, or email the KDA:

Kennedy's Disease Association, Inc.
P.O. Box 1105
Coarsegold, CA 93614-1105 (U.S.A.)

Phone Number: (559) 658-5950
Email Address: info@kennedysdisease.org