



# KDA e-Xpress

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## 2006 KDA Conference and Symposium

*“We now have a Reason for Hope”*

Attended by 47 associates and 15 doctors and researchers, this October's three-day event in Atlanta, GA generated much positive feedback.

The Doctors' panel discussion gave us certain insights into our disease and highlighted current understandings and research focus. The breakout sessions (men with KD and spouses/carriers) generated open, frank, and thought-provoking discussions. The meeting breaks, lunches, and formal dinner provided many opportunities to renew friendships, make new friends, and have one-on-one discussions with doctors and researchers. Diane Merry, Ph.D. and three of her post-doctoral researchers from Jefferson University gave a wonderful presentation on 'life inside a research lab'.

The feedback from several doctors and researchers was very positive concerning recent findings, the NIH clinical trial, and the current focus of their research.

*Article continued on page 2*

## KDA Committees and Volunteers

The KDA is fortunate to have several volunteers supporting the organization. If you are interested in helping the KDA in any of the areas listed below, please contact us at [info@kennedysdisease.org](mailto:info@kennedysdisease.org).

### COMMITTEE MEMBERS

### BOARD LIAISON

#### FUNDRAISING

T. J. Noack  
Tiffany Beck-Ortner  
Sean Michael Goynes

Ron Wiker - P  
Murray Williams

#### SUPPORT GROUPS

**Carriers**  
Mary Goynes

John Coakley - P  
Mike Goynes

#### **Family Members and Friends**

Seeking someone interested

#### **Individuals Exhibiting Symptoms**

Paul Buck  
Rich Smith

#### **Wives and Significant Others**

Paula Goynes  
Jeanne DeSchamp

#### CONFERENCES

Len Janicki  
Bud Goodnight  
T.J. Noack  
Jerry Brown  
Susanne Waite

Murray Williams - P  
Paul DeSchamp

#### PUBLIC RELATIONS

Bruce Gaughran - P  
Terry Waite  
Paul DeSchamp

#### EDUCATION & RESEARCH

Ed Meyertholen - P  
Bruce Gaughran

#### VOLUNTEER SUPPORT

Mike Goynes - P

*P = Primary Responsible Board Member*

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## CONFERENCE HIGHLIGHTS

**2007 Conference** - In light of the feedback provided by both KDA associates and doctors/researchers, a 2007 conference is currently being planned for early November in Las Vegas.

**Research Grants** - Several doctors spoke on the need for financial support from organizations like the KDA because of substantial cuts in federal funding. They commented that research is expensive and the funding is substantially less than in past years.

**Research Facilities** - There are currently 200+ labs working on poly-glutamine related disorders. Approximately 10-20 labs are working on Kennedy's Disease research. Many more labs are working on motor neuron diseases in general.

**National Institute of Health Clinical Trial** - The trial began in April of 2006. The drug being tested is Dutasteride (product name of Avodart). This drug is metabolized by the liver and has a six month half-life. There are currently 50 patients in this trial with 25 taking a placebo. The purpose of the current phase (Phase II) of the trial is to determine the safety of drug, its effectiveness in treating KD, and chronicle the history of the patients. Although the trial will run for 2½ years, if after one year the drug is proven safe and effective, they could open it up to all patients. If after one year the drug is proven safe and effective, they could open it up to all patients.

**Japanese Trial (Chemical Castration)** - Phase 2 study of leuprolide has gone well. The study was placebo-controlled for one year, with a 2 year follow-up period during which every patient received treatment. There was an indication of benefit in swallowing. Other outcome measures including quantitative muscle testing showed no change with placebo or leuprolide. A multicenter phase 3 placebo-controlled study in Japan with a focus on swallowing function is planned. They are hopeful that this will confirm a beneficial effect of the treatment on at least one aspect of the disease.

**Protein Accumulation May Cause KD** - The most accepted hypothesis for the cause of KD involves the accumulation of protein fragments in the nucleus of nerve cell. The accumulation of these proteins is believed to be the result of the inability of cells to properly degrade (breakdown and remove) the altered form of the androgen receptor protein (AR) that is found in all KD patients. Researchers are looking for ways to help the cells get rid of these mutant proteins. The disease and the accumulation of the AR in the nucleus are dependent on the presence of hormones testosterone and its close cousin, dihydrotestosterone (DHT) which bind to the AR - no hormone, no accumulation, and no disease (at least in mice!). Since the AR binds to DHT better than it binds testosterone and that DHT levels are high in nerve cells, researchers believe that reducing that amount of DHT may delay (and hopefully stop) the appearance of disease

symptoms. This is the theoretical basis of the clinical trial - dutasteride reduces the levels of DHT in cells. In addition, doctors do not recommend that patients take testosterone supplements.

**Another interesting Finding** - There may be some influence by the muscle cells themselves on the progression of the disease. It had generally been thought that the muscle atrophy was due to the loss of the motor neurons; this may be a bit too simple. Two lines of evidence indicate that the muscle cells may also be affected independent of the motor neuron cell death. It is not clear how this may influence the disease nor does it necessarily mean that the motor neuron death is not the primary cause of muscle atrophy.

## Two Research Grants Funded

Thanks to the generous donations of so many this year, the KDA, board of directors has approved the funding of \$50,000 in research grants this fall.

**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 SBMA, targeting the poly Q-expanded mutant AR. Dr. Chang proposes to develop a new therapeutic concept by targeting poly Q-AR via change conformation of poly Q-AR that allows poly Q-AR to become more vulnerable to be degraded. 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. He will use genetic approaches to: 1) characterize the role of the HDAC6 in SBMA pathogenesis, 2) identify the domains of HDAC6 that are required for suppression of SBMA degeneration, and 3) test the hypothesis that HDAC6-mediated suppression of SBMA pathogenesis is dependent upon autophagy. The long-term goal of the project is to contribute to the development of therapeutic interventions for SBMA.

Applications were accepted through August of 2006. The Scientific Review Board with the assistance of other researchers recommended the finalists in October and full grant proposals were reviewed in November.

The awardees in the past are J. Paul Taylor (twice), MD, PhD, Department of Neurology, University of Pennsylvania and Andrew Lieberman, MD, Department of Pathology, University of Michigan, who received grants of \$25,000 each. Their work has been reported on regularly and is aiding in the attempts to find a treatment or cure for Kennedy's Disease.

**The KDA thanks all researchers who have dedicated themselves to this cause and our Associates who continue to support the funding of these grants.**

## KDA Board of Directors Additions

The board of directors welcomes two new members. **Mike Goynes** worked at NASA in Houston repairing the Space Shuttle simulator computers and then moved to the Southeast where he has lived for the last 33 years. Mike's career has covered all aspects of electronics and computer hardware and software. Mike currently lives in Florida with his wife, Paula, who is also active in the KDA.

**Ed Meyertholen** teaches Cell and Molecular Biology, Genetics and Physiology at Austin Community College in Austin, TX. In 2005, Ed was awarded a sabbatical leave and was able to do research on Kennedy's Disease in Dr. Diane Merry's lab at Thomas Jefferson University in Philadelphia. Ed is also a member of the Scientific Advisory Board of the KDA. Ed currently lives in Texas with his wife, Pat.

## The 2006-2007 Board of Directors

Executive Director and Treasurer - Terry Waite

President - Bruce Gaughran

Secretary - J. Murray Williams

1st Vice President - John Coakley, Sr.

2nd Vice President - Ron Wiker

3rd Vice President - Paul DeSchamp

Member at Large - Mike Goynes

Member at Large - Ed Meyertholen

## KDA Tissue Donation Program

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) is pleased to announce the creation of a Tissue Donation Program for individuals with Kennedy's Disease who wish to donate their tissue. With the support of the University of Michigan Medical Center's Tissue Bank, individuals will be able to donate tissue to the KDA. All donated tissue will only be used for Kennedy's Disease research.

Additional information on this program can be found at <http://www.kennedysdisease.org/tissuedonationprogram.htm>

## 'Giving Thanks' Campaign

*"Without you, there is not KDA"*

Over the last twelve months, because of the generosity of supporters like you, the Kennedy's Disease Association (KDA) awarded \$75,000 in research grants. These grants provide critical funding to help researchers focus their attention on finding a treatment or cure for Kennedy's Disease.

Of the \$67,242 in donations received by the KDA this past year, 100% was invested directly into the Kennedy's Disease research effort.

Unfortunately, the federal government has reduced the funding available for medical research. As a result, research projects for Kennedy's Disease, along with other lesser known, lower public profile illnesses are being seriously under-funded.

Research is crucial to finding treatments and a cure for this disease that has affected all of our lives. Because of this, the **KDA's goal for 2007 is to award \$100,000 in research grants**. Yes, it is an aggressive goal, but not an unreasonable one based upon the generosity you have shown this past year.

If we all continue to work together, there is hope that someday soon, we will have a treatment or cure for Kennedy's Disease.

## Other KDA News

### 2005 ANNUAL REPORT

The KDA Annual Report for 2005 can be downloaded from our web site by clicking on the following link: <http://www.kennedysdisease.org/2005-Annual-Report.pdf>

### HOLIDAY CARDS

It is getting to be that time of year when greetings will be exchanged with family and friends. Please consider purchasing your cards from the KDA this year. There are 10 cards and envelopes for \$8.00 + \$2.00 shipping, 20 cards/envelopes for \$14 + \$2.50 shipping, or 30 cards /envelopes for \$20 + \$3.00 shipping.

### COOKBOOKS

The KDA has a wonderful cookbook, "Recipes from around the World." It contains 300 recipes including many contributed from KDA Associates. The books are \$12.00 when shipped within the US. If the books are to be shipped outside the US, please email the KDA for possible additional costs.

## **KDA Supports Gordon Research Conference on CAG Triplet Repeat Diseases**

The KDA has donated \$3,000 to the Gordon Research Conference scheduled for the spring of 2007. The Gordon Research Conference is an international meeting that brings together groups of 100-150 scientists to discuss topics in specific research areas. The Gordon conference requires that the attendees be a mix of senior scientists, junior scientists, and trainees. The format also requires presentation of unpublished data and encourages a lot of audience participation and discussion. Every two years there has been a Gordon conference on CAG expansion diseases. These Gordon conferences have probably the greatest concentration of Kennedy's Disease researchers and its research was featured in presentations in 2001 by Dr. Kurt Fischbeck and 2003 by Dr. Gen Sobue.

Diane Merry, Ph.D., the chairperson for the conference wrote, "We want to thank the Kennedy's Disease Association ... for its generous contribution of \$3,000 to help defray the costs of the Conference. We will of course recognize the Kennedy's Disease Association as one of the sponsors for this conference and we will provide an informal summary of the conference after it is complete." We will ensure that "the funds are designated for use for Kennedy's Disease researcher(s).

Again, thank you for all your help." We "... are extremely grateful to the KDA for its support of this Conference."

## **KDA Forum**

*"Have you visited the forum recently?"*

The KDA forum is alive and well. There are many topics posted covering a variety of subjects. The benefits of this forum service include:

- No advertising
- Email notification of message replies

Visit the forum on our web site.

[http://www.kennedysdisease.org/disc\\_forums.html](http://www.kennedysdisease.org/disc_forums.html)

## **KDA Chat Room**

Have you noticed some of the guests and topics covered recently: The Biology of Kennedy's Disease, Current Research with Dr. Lieberman, and The NIH Clinical Trials with Alison La Pean. Other guests included Dr. Taylor and Dr. Di Prospero. If you missed these chats, you can still read the transcripts by visiting our web site.

[http://www.kennedysdisease.org/disc\\_transcripts.html](http://www.kennedysdisease.org/disc_transcripts.html)

Watch for the 2007 winter and spring schedule of guests and topics.

### **KDA e-Xpress**

**A publication of the  
Kennedy's Disease Association.**

Editor: Bruce Gaughran

Comments, suggestions, and questions should be sent to:  
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### **The Objectives of the Kennedy's Disease Association**

- Sharing information about Kennedy's Disease with those who seek it
- Creating a support system for those living with Kennedy's Disease
- Increasing public awareness of Kennedy's Disease and its effect upon families
- Increasing awareness of Kennedy's Disease in the medical community
- Raising funds, with an aggressive target to earmark at least 70% of every dollar donated to be used for Kennedy's Disease research