Kennedy's Disease Association

OUR MISSION

inform, support, educate, fund research, and find a cure for Kennedy's Disease

KDA Research Funding for 2024

Since its founding in 2000, the Kennedy's Disease Association (KDA) has provided over $2 million in funding to help find a cure or treatment for Kennedy's Disease. This year, thanks to the continuing generosity of patients, families, and friends, we are providing $437,000 for KD research in three countries. This is the highest amount for any single year in KDA's history.

This year's KDA grant recipients are:

- **Xavier Salvatella Giralt**, IRB Barcelona, Spain (two-year award)
- **Masoud Shekarabi**, Thomas Jefferson University
- **Heather Montie**, Philadelphia College of Osteopathic Medicine
- **Davide Pareyson**, Besta Institute, Milan, Italy (two-year award)

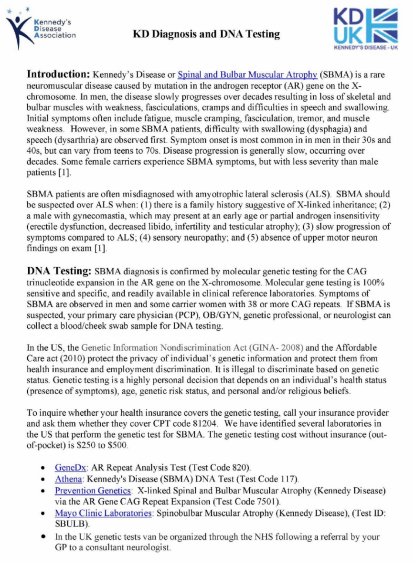
The recipient of the 2023 Waite-Griffin SBMA Fellowship is **Anastasia Gromova**, University of California, Irvine.

We would like to congratulate all the awardees and express our appreciation for their efforts to research and counter the effects of KD.

To maintain the momentum in supporting global KD research, KDA is launching a Fall fundraising campaign. Look for more information in the near future.

KD "*One Pager*" Released
Topic: Diagnosis and DNA Testing

The Kennedys Disease Association is developing a series of short Reference Guides (1-2 pages) on different aspects of KD including symptoms and management approaches. These documents provide information for patients as well as medical personnel who may not be familiar with KD. We encourage you to print copies to



KD Diagnosis and DNA Testing

Introduction: Kennedy's Disease or Spinal and Bulbar Muscular Atrophy (SBMA) is a rare neuromuscular disease caused by mutation in the androgen receptor (AR) gene on the X-chromosome. In men, the disease slowly progresses over decades resulting in loss of skeletal and bulbar muscles with weakness, fasciculations, cramps and difficulties in speech and swallowing. Initial symptoms often include fatigue, muscle cramping, fasciculation, tremor, and muscle weakness. However, in some SBMA patients, difficulty with swallowing (dysphagia) and speech (dysarthria) are observed first. Symptom onset is most common in in men in their 30s and 40s, but can vary from teens to 70s. Disease progression is generally slow, occurring over decades. Some female carriers experience SBMA symptoms, but with less severity than male patients [1].

SBMA patients are often misdiagnosed with amyotrophic lateral sclerosis (ALS). SBMA should be suspected over ALS when: (1) there is a family history suggestive of X-linked inheritance; (2) a male with gynecomastia, which may present at an early age or partial androgen insensitivity (erectile dysfunction, decreased libido, infertility and testicular atrophy); (3) slow progression of symptoms compared to ALS; (4) sensory neuropathy; and (5) absence of upper motor neuron findings on exam [1].

DNA Testing: SBMA diagnosis is confirmed by molecular genetic testing for the CAG trinucleotide expansion in the AR gene on the X-chromosome. Molecular gene testing is 100% sensitive and specific, and readily available in clinical reference laboratories. Symptoms of SBMA are observed in men and some carrier women with 38 or more CAG repeats. If SBMA is suspected, your primary care physician (PCP), OB/GYN, genetic professional, or neurologist can collect a blood/cheek swab sample for DNA testing.

In the US, the Genetic Information Nondiscrimination Act (GINA- 2008) and the Affordable Care act (2010) protect the privacy of individual's genetic information and protect them from health insurance and employment discrimination. It is illegal to discriminate based on genetic status. Genetic testing is a highly personal decision that depends on an individual's health status (presence of symptoms), age, genetic risk status, and personal and/or religious beliefs.

To inquire whether your health insurance covers the genetic testing, call your insurance provider and ask them whether they cover CPT code 81204. We have identified several laboratories in the US that perform the genetic test for SBMA. The genetic testing cost without insurance (out-of-pocket) is $250 to $500.

- GeneDx: AR Repeat Analysis Test (Test Code 820).
- Athena: Kennedy's Disease (SBMA) DNA Test (Test Code 117)
- Prevention Genetics: X-linked Spinal and Bulbar Muscular Atrophy (Kennedy Disease) via the AR Gene CAG Repeat Expansion (Test Code 7501).
- Mayo Clinic Laboratories: Spinobulbar Muscular Atrophy (Kennedy Disease), (Test ID: SBULB).
- In the UK genetic tests van be organized through the NHS following a referral by your GP to a consultant neurologist.

share with doctors, clinicians, and therapists.

The first reference guide is about "KD Diagnosis and DNA Testing" and is now available for download.

The guide provides a description of KD symptoms, how to distinguish KD from ALS, DNA testing procedures, links to four commercial labs that provide DNA testing, and some suggestions on next steps if you are diagnosed with KD.

Researcher Profile - Dr. Diane Merry

Dr. Merry is a Professor in the Department of Biochemistry and Molecular Biology and Vice Chair, Faculty Development and Engagement at Thomas Jefferson University in Philadelphia. She is also a member of Jefferson's Vickie & Jack Farber Institute for Neuroscience and Chair of its Scientific Advisory Board. Dr. Merry's research group uses novel cell and mouse models to understand how motor neurons and muscle cells respond to the accumulation of misfolded AR protein and to identify therapeutic opportunities in SBMA. Working with Dr. Merry, Dr. Anna Pluciennik received two KDA funded grants to: 1) better understand the polyQ expanded androgen receptor (AR) and other cellular processes and the role of the deubiquitinase USP7 in SBMA mouse models (in the second grant). This work culminated in a publication in early 2021 in nteractome and USP7 – two PhD students are working hard on USP7 to try to develop novel therapies that target this interaction.

The First KD Banbury Conference

The KDA Tries a New Approach to Researcher Collaboration

On September 10-12, 2023, the KDA held its first Banbury Conference. Researchers from around the world gathered at the Banbury Center to stimulate additional research into the mechanisms of SBMA pathology, and to identify potential therapeutic approaches.

Think Tank participants continue the conversation during breaks and meals.
(Cold Spring Harbor Laboratory / Constance Brukin.)

Banbury Center in New York is a gathering spot for scientific think tanks. "The meetings are

A key goal of the workshop was to invite fresh perspectives from research experts with a diversity of backgrounds and to encourage new collaborations among scientists leading to new areas of SBMA research and/or therapy development.

recognized internationally as being amongst the world's best discussion workshops for topics in molecular biology, molecular genetics, human genetics, neuroscience and science policy." (Cold Spring Harbor Laboratory / 2023)

Technical sessions covered topics including:

- Role of muscle, nerve and the neuromuscular junction in SBMA
- Modeling cell biology using Induced pluripotent stem cells (iPSCs)
- Innovative Next steps in SBMA Research

The conference produced so many new ideas that we are planning the Second KD Banbury Conference in 2024!

Funding for the workshop was provided by the 2023 KD Texas Golf Scramble. **Thank you!**

Conference Participants. (Cold Spring Harbor Laboratory / Constance Brukin.)

CoRDS Rare Disease Registry

Coordination of Rare Diseases at Sanford (CoRDS) is a free, nonprofit, international patient registry for all rare diseases, located at Sanford Research in Sioux Falls, SD. CoRDS compiles information on rare diseases, connects researchers with patients, and will notify you of emerging clinical trials. Since starting work with KDA, CoRDS has registered **385 men and women from 24 countries** who report carrying the KD gene.

On behalf of AnnJi Pharmaceutical Co., Ltd., the CoRDS registry contacted men with SBMA with the opportunity to enrolled in an ongoing clinical trial of AJ201, a new drug designed to promote degradation of the mutant androgen receptor (AR) protein responsible for Kennedy's Disease.

The reason CoRDS does the initial contact for drug trials is because the people in the registry own their data, including contact information. CoRDS does not share contact information with anyone.

CoRDs has recently updated their website and will be contacting individuals that have previously registered to provide a new password and update their information. If you have not already registered, we encourage you to register now. The KD patient registry was developed jointly with Sanford Research, the NIH and the KDA. This registry will facilitate KD/SBMA research and will be used to invite patients for clinical trials. Most important, a robust registry will provide a roadmap for investigators and will help them develop improved treatments and potentially a cure for this debilitating rare disease.

To register and/or update your information

Click Here

London International Kennedy Disease Conference

KD UK
KENNEDY'S DISEASE - UK

The first ever London International Kennedy Disease Conference is being held November 4-7, 2023. The first two days of the conference (4-5 November) are focused on people living with Kennedy Disease, carriers, carers and for people wishing to learn more about the disease. The venue for Days 1 and 2 is fully accessible, and you can also view presentations by remote access (internet). there is full disabled access. The second two days (November 6-7) are predominately aimed at researchers, clinicians and scientists. The venue for Days 3 and 4 is not fully accessible and will not be broadcast.

KDA members viewing the presentations online need to remember that the presentations start at 11 AM London time (6 AM in New York, 3 AM in California).

For more information and to register for in-person or online attendance, click the link below.

Click for more Information

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