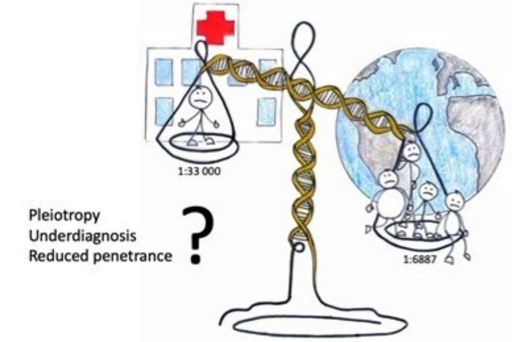
Kennedy's Disease Association

OUR MISSION

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

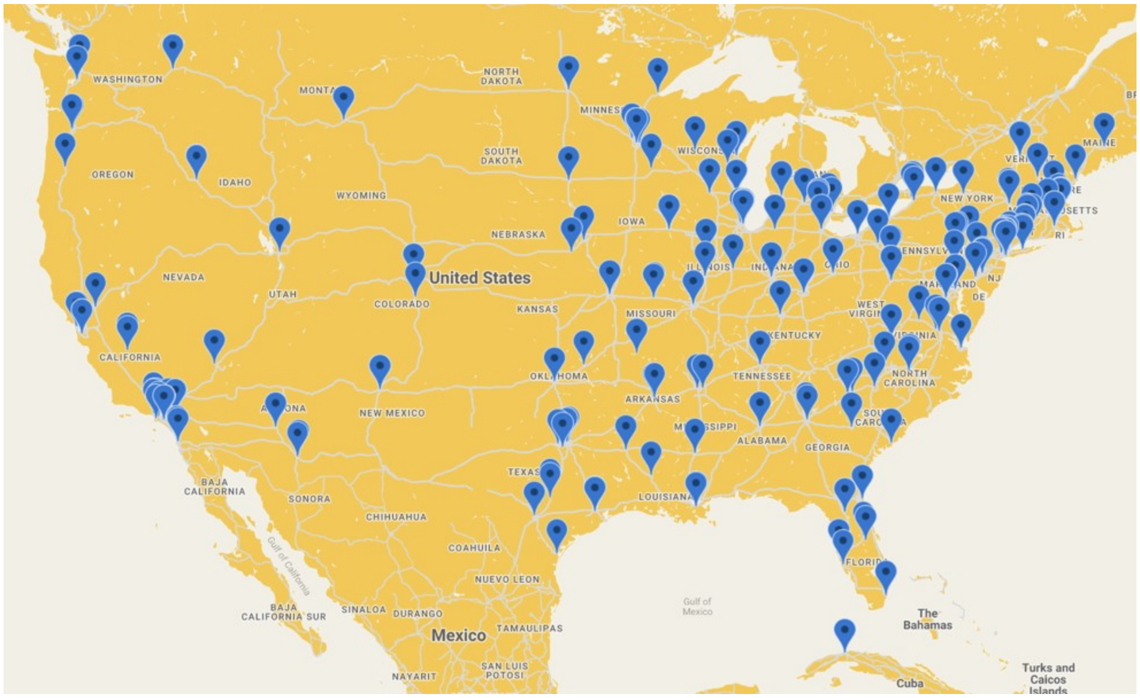


Kennedy's Disease gene mutation more common than previously thought - University College of London Study reveals

Researchers at the University College of London (UCL) recently published a groundbreaking paper about the prevalence of the gene mutation causing Kennedy's disease (SBMA). Kennedy's disease (KD), as Newsletter readers know, is an adult-onset, genetic, neuromuscular disease, caused by a genetic mutation of the Androgen Receptor (AR) gene. Disease results when there are more than 37 CAG expansion repeats in the AR gene. Previous literature reported that KD affected about 1:30,000 males worldwide. The prevalence of the gene mutation itself in the general population remained a mystery. The UCL paper, based on study of over 70,000 individuals from several genomic repositories including the 100,000 Genome Project, postulates that the prevalence of the gene mutation in the general population to be 1:3182 people, not all of whom would experience symptoms. UCL also estimates the disease prevalence in males to be 1:6,887, more than four times the previously reported level of KD. These striking results suggest that Kennedy's disease is underdiagnosed or -- as many KD patients will tell you -- misdiagnosed due to lack of knowledge about the disease. The UCL findings should help overcome this knowledge gap and lead to greater interest in the field of neuromuscular research and development of effective therapies to treat this devastating disease. The UCL study can be found at the following website: Unexpected frequency of the pathogenic AR CAG repeat expansion in the general population | Brain | Oxford Academic (oup.com)

MDA Care Centers

The Muscular Dystrophy Association (MDA) maintains a network of over 150 Care Centers throughout the United States (see map below). These centers are located in leading hospitals and health care institutions, typically in neurology or neuroscience departments. Physicians at MDA centers are familiar with Kennedy’s Disease and other neuromuscular diseases, can assist in diagnosis, and connect you with a genetic counselor to coordinate genetic testing and answer questions. Once you're diagnosed with Kennedy's Disease or Spinal-Bulbar Muscular Atrophy (SBMA), the center staff will often schedule follow-up visits with a neurologist, physical and occupational therapists, to help manage symptoms and obtain assistive devices / mobility aids. MDA does not provide financial support, but can provide suggestions on alternative funding sources and local service providers.



A Readers Story

Kennedy's Disease: A Child's Perspective

My dad was in his early thirties when he received the initial verdict from his doctors: myasthenia gravis. It turned out to be just the first in a series of diagnoses: ALS, then muscular dystrophy, then finally, "We don't exactly know." He died at age 67 without ever hearing the term Kennedy's Disease. The only reason I know he had KD is that now my cousin has also been diagnosed with it. Mystery solved.

My dad was obviously different from all the other dads I knew growing up. I have very early memories of going to dinner with family friends, falling asleep on the drive home, and then not understanding why Dad wouldn't carry me in from the car. He'd always wake me, and I'd have to walk half-asleep up to bed. Though how could he possibly carry me, when it took him ages just to climb the single flight of stairs to our apartment (holding on to the walls because there were no handrails)?

As I grew older, I realized my father didn't have the physical strength to do many of the things "normal" dads did. And when he did do something that I'm sure my friends took for granted with their own dads, the moment would be so precious I knew I'd never forget it. Like the time he took me on a rollercoaster. I can still hear him laughing with joy, even though he knew he'd make a spectacle of himself at the end of the ride when people had to help him out of the car. He hated being the object of curious or pitiful looks. And yet I knew he'd put himself in that situation for me, and I loved him for it.

So despite Dad's challenges, we did have fun together, albeit sporadically, and I treasured every one of those atypical moments we shared. As time passed, he began to avoid social situations whenever possible. He never wanted to be the centre of attention, as if feeling his disability made him unworthy of people's concern. Looking back now, as a scholar dedicated to exploring and researching ableism, I can see that Dad internalized the disability stigma and tragically called himself an "invalid." But in my youthful ignorance, I failed to ask him so many crucial, existential questions. And more importantly, I missed the chance to show him he was anything but an invalid. To me, he was perfect.

By Vera Dolan, PhD

A KD daughter and an academic researcher interested in the sociocultural lived experiences of persons with disabilities.

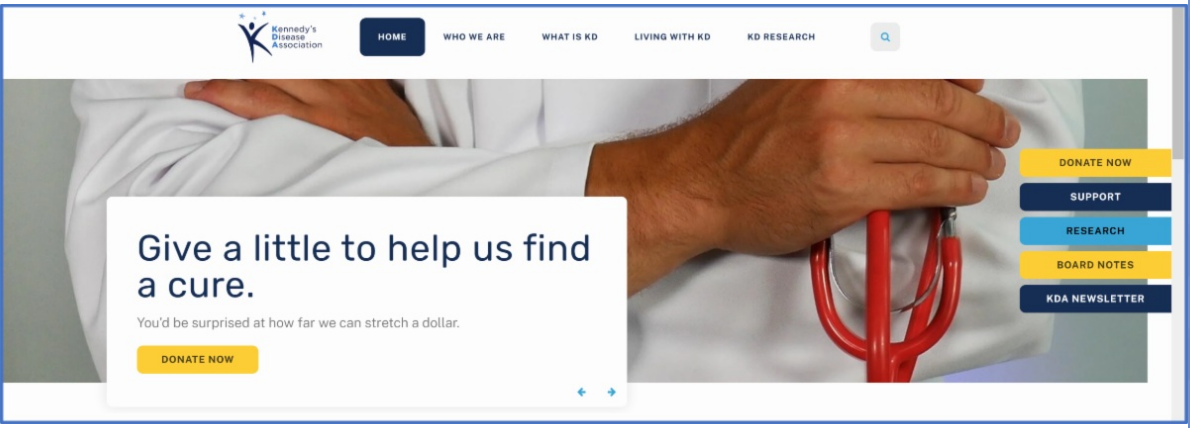
To share your experiences, submit an article to our Newsletter Editor

KD Support Group Zoom Chat

KDA is hosting a Zoom chat for men to share experiences and just catch up with old friends. Chats are typically held the 2nd Saturday of each month at 12:00 Eastern time, but schedules can vary due to holidays. If you would like an invite to the next call, please email

MensZoomChat@kennedysdisease.org

New KDA Website



Give a little to help us find a cure.

You'd be surprised at how far we can stretch a dollar.

KDA has a new, improved, and updated website! The new site features streamlined displays that are easier to read, topics that are easier to find, updated information on Living with KD and KD Research, an Events calendar, notes from Board of Directors meetings, and more. You'll find the site at the same location: **Home : Kennedy's Disease Association (kennedysdisease.org)**. Many thanks to KDA Board member Ronald Moffett for establishing the new site.

Take a look at the new KDA website. We hope you find it a continuing source of helpful information in our common efforts to live with and manage KD and find a cure.

Help Find a Treatment or Cure

The Kennedy's Disease Association's mission is to inform, support, educate, fund research and find a cure for Kennedy's Disease. KDA has awarded grants and fellowships to researchers in the United States, Canada, Britain, Italy, and Japan to support both basic research and clinical trials on the causes and potential treatments for for Kennedy's Disease. To donate, go to

DONATE NOW

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