Kennedy's Disease
ASSOCIATION

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A public benefit, non-profit corporation
Designated 501(c)3

Together we will reach the Stars

It is our mission to inform, support, communicate, educate, research and find a cure for Kennedy's Disease.

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REACHING FOR THE STARS

The stars of hope for those affected by Kennedy's Disease may have seemed untouchable in the past. But with dedication, planning, expertise, and good fortune we are reaching those stars.

The Kennedy's Disease Association is bringing together the critical elements for that journey. We will get there. We are making the time, gathering the financial resources, amassing medical and technological expertise and helping coordinate the efforts of many dedicated individuals—affected individuals, researchers and physicians, supporters, donors, volunteers, educators, facilitators, fund raisers and others—to help us find the way.

WHAT IS KENNEDY'S DISEASE (SPINAL BULBAR MUSCULAR ATROPHY)?

Spinal Bulbar Muscular Atrophy (SBMA) is a rare X-linked adult onset neuromuscular disorder that is progressive. It is currently both non-treatable and incurable. It is commonly referred to as Kennedy's Disease (KD)—named after Dr. William R. Kennedy, a physician who conducted early research and determined the genetic nature of the disease.

Symptoms of KD become apparent when the patient's spinal and bulbar neurons have already begun to gradually degenerate and die. This initially causes twitching and cramping of the muscles. KD then gradually progresses into muscular weakness and atrophy (wasting) in the legs, trunk, arms, shoulders, neck and face. These weaknesses dramatically decrease the patient's capacity for walking, speech, and swallowing.

Symptoms usually begin to appear between the ages of 30 and 50-though earlier and later onsets have been recorded. Males who have inherited this gene are always affected by neuromuscular symptoms Females have also been know to have exhibited symptoms.

Life expectancy is noted to be near-normal to normal, as KD is not in itself fatal. However, late-term KD patients may have difficulty swallowing and may ingest food or liquids into the lungs. This may cause aspiration pneumonia, which needs to be properly treated immediately. Pneumonia and respiratory complications are especially serious for those affected with KD and can cause death.

It is estimated that 1 in 40,000 people worldwide are affected with KD. However, this number of incidence may be underestimated because those with KD are often misdiagnosed. A common misdiagnosis is Amyotrophic Lateral Sclerosis (aka: ALS or Lou Gehrig's Disease) as symptoms may appear similar. A DNA blood test for Kennedy's Disease is helping to change this fact.

Education

HOW IS IT CONTRACTED?

KD is a genetic disorder. The gene believed to be responsible for the KD trait is the androgen receptor gene, which is located on the X chromosome (Xq11-12). KD can be passed from generation to generation, if one of the parents has the KD trait.

Females with the trait are carriers, and their children each have a 50% chance of inheriting an affected X chromosome from their mother (regardless the gender of the child). Males with the trait never pass the trait to their sons (because they give them only their Y chromosome), and always pass the trait to their daughters (because they give them their affected X chromosome). Couples, known to have KD in their family and concerned about childbearing, may consider discussing the matter with a genetic counselor.

WHAT IS THE KENNEDY'S DISEASE ASSOCIATION (KDA)?

The Kennedy's Disease Association is a non-profit corporation established in August 2000. It has been granted 501(c)3 status by the Internal Revenue Service, meaning that all donated contributions are tax-deductible. Its staffing is 100% volunteer, all of whom are KD patients, carriers, spouses, caregivers, family members or friends.

The goals of the Kennedy's Disease Association include:

- **Sharing information about KD with those who seek it**
- **Creating a support system for those living with KD**
- **Increasing public awareness of KD and its effects upon families**
- **Increasing awareness of KD in the medical community**
- **Raising funds for KD, with an aggressive target to earmark 90% of every dollar donated to be contributed to KD research**

Support

Communication

Research

The KDA has established a comprehensive Web site to help share information worldwide in a timely, cost-efficient, effective way. Since KD is considered rare, the best way we can 'meet' is via the Internet.

The KDA Web site is located at:
www.kennedysdisease.org

Those without Internet access, or e-mail capabilities, are invited to contact the KDA by phone or mail at:

Kennedy's Disease Association, Inc.
P.O. Box 1105
Coarsegold, CA 93614
Telephone: 855-532-7762
International: +1-734-288-5580

HOW DO I JOIN?

At present, the KDA does not charge a fee for individuals to participate in KDA activities. Please provide us your name, address, phone number and e-mail address (if applicable), so that we can include you in our organization. If you have the ability, donations in any amount are requested so that you can help us conquer this disease.

HOW CAN I HELP SUPPORT THE KDA AND ITS MISSION?

- **Make a tax-deductible donation**
- **Volunteer, we can always use your help and expertise**
- **Remember the KDA in your estate planning, life insurance or will**
- **Organize a Fundraiser**

COMMON SYMPTOMS OF KENNEDY'S DISEASE:

An individual with Kennedy's Disease may experience some or all of the below signs and symptoms. (Note that there is significant variation in observed affect, age of onset and rate of progression.)

NEUROLOGIC:

BULBAR SIGNS
Problems with breathing, swallowing, talking and other functions of the throat.

DYSPHAGIA
Trouble swallowing.

INTENTION TREMOR
Typically hand or finger tremors when trying to do something.

ABNORMAL BABINSKI
Abnormal plantar response, ie., when the bottom of the foot is scraped, the toes fail to reflexively bend down.

LOWER MOTOR NEUROPATHY
Loss of lower motor nerve cells leads to weakness and wasting of the muscle.

PRIMARY SENSORY NEUROPATHY
Numbness over certain areas. Loss of sensation.

DECREASED OR ABSENT DEEP TENDON REFLEXES
When the knee is tapped with a medical hammer; there is no response.

MUSCULAR:

FASCICULATIONS
Twitching of small muscles without purposeful movement visible through the skin, and very noticeable in the perioral region.

CRAMPS
Large muscle spasms.

POSTURAL TREMOR
Shaky muscles with certain positions.

MUSCULAR ATROPHY
Wasting and shrinkage of muscles that occurs when the lower motor nerve does not stimulate the muscle adequately.

HYPERTROPHIED CALVES
Enlarged calves (even though weakened) caused by fatty infiltration of the muscle cells.

THORACIC:

GYNECOMASTIA
Enlargement of male breasts, usually occurring during puberty.

ENDROCRINE:

ANDROGEN DEFICIENCY
Loss of masculinizing effect.

ESTROGEN EXCESS
More of an apparent estrogen effect because of the loss of masculinizing effect.

GENITO-URINARY:

IMPOTENCE
Erectile dysfunction.

REDUCED FERTILITY
Low sperm count.

TESTICULAR ATROPHY
Testicles become smaller and less functional.

MISCELLANEOUS CHARACTERISTICS:

LATE APPARENT ONSET
Usually show symptoms late 30's or after.

SLOW PROGRESSION
Near-normal lifespan.

ASYMMETRY OF CLINICAL SIGNS
Muscles of one side may be more affected than the same muscles on the other side.

LABORATORY:

ELEVATED SERUM CREATINE KINASE
Elevation of CPK enzyme in the blood test. Can be confused with the enzyme released during a heart attack.

GENETIC BLOOD TEST
The Kennedy's Disease gene can be found in the blood by a genetic laboratory in both affected males and carrier females.

I want to help you reach the stars and support your efforts in helping those with Kennedy's Disease.

☐ I HAVE ENCLOSED $ _____ AS A DONATION

☐ PLEASE KEEP ME INFORMED ABOUT THE KDA

☐ OTHER

NAME: _____

ADDRESS: _____

CITY: _____ STATE: _____ ZIP: _____

COUNTRY: _____

PHONE: ( ) _____ E-MAIL: _____

Please detach and mail back