

Kennedy's Disease

What is Kennedy's Disease?

- Kennedy's Disease (KD) is a rare neuromuscular disease and a form of adult-onset spinal muscular atrophy (SMA).
- Kennedy's Disease is the result of mutations in the Androgen Receptor gene on the X chromosome. There are small increases in one specific region of the gene. The larger the increases, the earlier the onset and the more pronounced the symptoms.
- It is also known as Spinal Bulbar Muscular Atrophy (SBMA).
- It affects the spinal and bulbar neurons causing muscle wasting (atrophy) predominantly in the legs, arms, face and throat.
- It is an inherited chromosomal X-linked recessive disease.
- Only males are fully affected.

How common is Kennedy's Disease?

- It is estimated that one in 40,000 people worldwide have KD.

Symptoms

Neurological

- Weakness of tongue and mouth muscles.
- Problems breathing (breathy speech pattern).
- Difficulty swallowing.
- Poor articulation.
- Changes in voice and speech (harsh or strained voice, decrease in range of pitch).
- Numbness.
- Hand tremors.
- Decreased or absent deep tendon reflexes.

Resources

www.kennedysdisease.org
www.mdausa.org

Muscular

- Twitching or muscles when at rest (Fasciculations).
- Cramps: large muscle spasms.
- Increased calf size due to cramps.
- Loss of muscle bulk.

Other

- Erectile dysfunction.
- Breast enlargement (Gynecomastia).
- Impotence.
- Decreased size and function of testicles.
- Low sperm count.

Other facts and characteristics

- Late onset: Symptoms usually develop in the late 30's or later.
- Progresses quite slowly. Those living with KD have a nearly normal lifespan.
- Females are rarely affected but can be carriers and experience a mild expression of the disease.

Diagnosis

- Many people are not diagnosed for years.
- Due to similarities in symptoms, many people are misdiagnosed with ALS.
- KD can be diagnosed easily with a simple DNA blood test.
- It is most often diagnosed between the ages of 20-50 years.

Treatment

- To date, KD is incurable and there are no treatments.
- Some people living with the disease do take prescribed medications to alleviate symptoms.
- Patients have reported that stretching exercises can help ease cramps and pain.

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