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.

Muscular

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

Other Symptoms

• 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.

To learn more about Kennedy's Disease, visit:

www.kennedysdisease.org;

To find a contact in or closest to your province, visit:
http://www.kennedysdisease.org/disc_contacts_canada.html