Kennedy's disease

**Summary**

- Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly the arms and legs.
- Kennedy's disease is caused by a genetic mutation of the androgen receptor gene on the X chromosome.
- There is no cure, and treatment can only ease some of the symptoms.

Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly the arms and legs. Other major symptoms include severe cramps and problems with speech and swallowing. The disease progresses slowly, and life expectancy is usually normal. Kennedy’s disease is also known as X-linked spinal bulbar muscular atrophy (SBMA). There is no cure yet, and treatment can only ease some of the symptoms.

In most cases, females who inherit the gene are carriers, while men who inherit the gene develop the symptoms. (Rarely, an affected woman may develop symptoms too.)

**Symptoms of Kennedy's disease**

The many symptoms of Kennedy’s disease generally first appear when the person is aged between 30 and 50 years and can include:

- swallowing difficulties
- speech difficulties
- hand tremors
- shaky muscles when holding certain postures
- muscle weakness
- muscle wastage
- muscle twitches
- muscle cramps and spasms
- enlarged calf muscles due to constant cramping
- localised areas of numbness
- absent reflexes, such as the foot remaining still when the tendon beneath the kneecap is tapped
- enlarged breast tissue (gynaecomastia)
- impotence
- low sperm count
- shrunken testicles
- reduced sex drive.

**Kennedy's disease destroys motor nerves**

The brain controls the movement of muscles with nerves. The command to move travels from the upper motor neurons on the brain’s surface, down through the spinal cord and into the lower motor neurons. Every muscle is connected to between 50 and 200 lower motor neurones, and each neurone subdivides into many tiny branches. The end of each branch is called a pre-synaptic terminal. The electrical command to move the muscle is translated...
at the pre-synaptic terminal into a chemical called acetylcholine, which prompts special receptors in the muscle tissue to contract.

Kennedy's disease causes the loss of motor neurone cells, including those in the brain stem that supply the bulbar muscles. These muscles control the throat, which is why swallowing, breathing and speech problems are common symptoms of Kennedy's disease.

This inherited disorder affects males

Humans have 46 paired chromosomes, with two sex chromosomes that decide gender and 44 chromosomes that dictate other factors. The mother's egg always provides a female sex chromosome known as 'X', while the father's sperm contributes either an 'X' (female) or 'Y' (male) chromosome. A pairing of 'XX' sex chromosomes means the baby is a girl, while 'XY' means the baby is a boy. Kennedy's disease is caused by a genetic mutation of the androgen receptor gene on the X chromosome.

Since the disease is recessive, the presence of the normal gene on the other X chromosome means that girls don't develop the symptoms of disease, because the normal gene overrides the mutated one. However, these girls will be carriers, and each son of a carrier mother has a 50 per cent chance of inheriting the gene and developing the disease. This is because boys only receive one X chromosome and, if that has the mutated gene, they will one day develop the symptoms of Kennedy's disease.

The androgen receptor gene

The androgen receptor gene helps to control the activity of the male sex hormones (androgens). This explains the androgen insensitivity symptoms such as breast enlargement, impotence and low sperm count. However, it is not known why this gene mutation causes the other symptoms of Kennedy's disease.

Diagnosis of Kennedy's disease

Since Kennedy's disease is rare, it can be misdiagnosed. It is sometimes mistaken for the more common motor neurone disease (amyotrophic lateral sclerosis), a progressive nervous system disease characterised by the breaking down of neurones in the spinal cord and brain.

Kennedy's disease can be diagnosed using a number of tests including:

- **Blood test to check for elevated serum creatine kinase (CPK)** – people with Kennedy's disease usually have a greater than average amount of this particular enzyme circulating in their blood.
- **Genetic tests using a blood sample** – this test can check whether the Kennedy's disease gene is present, even if the person is asymptomatic or a carrier. Genetic counselling is important.

Treatment for Kennedy's disease

There is no cure for Kennedy's disease, because medical science doesn't know how to regenerate muscle neurones. Treatment aims to ease some of the symptoms and can include:

- medications to reduce muscle cramps and tremors
- plenty of rest and avoidance of exhaustion
- healthy, balanced diet
- gentle and regular aerobic exercise
- regular stretching to help reduce muscle cramping
- pain management
- speech therapy
- occupational therapy
Where to get help

- Your **GP (doctor)**
- **Motor Neurone Disease Association** Tel. 1800 806 632
- **Victorian Clinical Genetic Services** Tel. 1300 118 247