MND Australia Fact Sheet

Kennedy's Disease

What you should know

- Kennedy's Disease refers to Spinal and Bulbar Muscular Atrophy (SBMA) and is a rare kind of inherited disease that affects the nerve cells in the human brain
- The disease causes the muscles of the face and limbs to become weaker and waste away
- Other major symptoms include enlargement of breasts, muscle cramps and spasms, hand tremors, impotence and speech and swallowing difficulties
- The symptoms can be managed and the disease is not usually life limiting, but there is no cure
- Age of onset is usually between 35 and 40 years and men are more commonly affected.

What is Kennedy's Disease?

Kennedy's Disease refers to Spinal and Bulbar Muscular Atrophy (SBMA) and is a rare type of inherited disease that affects the neurons (more commonly known as nerve cells) in the brain[1]. Kennedy's Disease is named after William R. Kennedy who reported seeing this disease in patients in 1968[1].

It is estimated that around 1 to 2 per 100,000 people worldwide has Kennedy's Disease, although the number may be higher due to difficulties with making a definitive diagnosis[1].

The disease is characterised by the degeneration of lower motor neurons of the brainstem and spinal cord[2]. This results in the muscles of the face and limbs becoming weaker and wasting away[3]. The loss of muscle strength eventually leads to weakness in muscles throughout the body which makes movement difficult, particularly in the legs and arms, and with speech and swallowing.

Kennedy's Disease more commonly affects men.

Other major symptoms of the disease include enlargement of breasts, muscle cramps and spasms, hand tremors, impotence and speech and swallowing difficulties[4]. The symptoms can be managed, but there is no cure[5].

Age of onset is usually between 35 and 40 years[1]. The disease generally progresses slowly[6] and does not usually impact life expectancy[7].

Genetic mutation and Kennedy's Disease

A mutation in the androgen receptor gene (AR) is responsible for the disease[8]. The androgen receptor gene is located on the X chromosome[9] and helps to control the activity of male sex hormones (androgens).

Why are men mostly affected?

Men who inherit the mutated androgen receptor gene develop the disease. The mutated gene is present on the X chromosome, which is one of two chromosomes that decide the sex of a human. Men have X and Y chromosomes[10].

Women have two X chromosomes[10]. Women who inherit the mutated gene also carry it on their X chromosome. The impact of the normal gene on the other X chromosome usually overrides the mutated gene so this makes it rare for a woman to develop the disease. Women who have inherited the mutated gene are carriers of the disease[1]. Each son of a woman who is a carrier has a 50% chance of inheriting the gene and developing the disease[1].

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Diagnosis

Kennedy's Disease is rare and can be misdiagnosed, often being mistaken for motor neurone disease (MND) as it shares a lot of similar symptoms. A thorough medical review is required to make an accurate diagnosis and includes assessment of the patient's family history, symptoms and rate of progression. Tests[11] used to confirm a diagnosis of Kennedy's Disease include:

- Genetic tests using a blood sample that looks for the faulty gene
- A blood test to check for elevated serum creatine kinase (CPK) as almost all patients with SBMA are found to have very high levels, , and
- Neurologist assessments for elevated CAG repeats (a segment of DNA).

Similarity and differences with MND

The main similarity between Kennedy's Disease and MND is the symptoms they display. Both diseases cause the weakening and wasting of muscles. While the wasting of muscles can make Kennedy's Disease easy to mistake for MND, there are a number of significant differences between the diseases.

Differences

	Kennedy's Disease	MND
Onset	Onset is usually between 35 and 40 years	Can affect adults at any age, but most people who develop MND are in their 50s or 60s
Cause	Inherit the faulty gene from mother	The causes for the majority of cases of MND remain unknown. However, about 5 to 10% of cases are caused by inheriting a mutated gene from a parent.
Incidence	Approximately 1-2 per 100,000	Approximately 8.7 per 100,000
Progress	Slow	Rapid
Life expectancy	Usually normal	Average survival time after diagnosis is 2.5 years
Gender	Men more commonly develop the disease	MND affects both men and women

How is Kennedy's Disease managed?

Currently, there is no cure for Kennedy's Disease. Treatment aims to reduce and manage the symptoms. A healthy lifestyle that includes a balanced diet, gentle and regular aerobic exercise, getting plenty of rest and avoidance of exhaustion plays a big part in daily management of the condition. Regular stretching to help reduce muscle cramping is also useful. Other interventions[11] include:

- medications to reduce muscle cramps and tremors
- pain management
- speech therapy
- occupational therapy
- physiotherapy
- dietary advice
- fall prevention

Where to get support

Talking with your General Practitioner (GP) is the best place to start if you have any suspicions that you may have Kennedy's Disease. You will usually be referred to a Neurologist for further review and testing. If you do have Kennedy's Disease your GP can help you access the health professionals you need as time goes on.

Female carriers of Kennedys Disease can receive genetic and fertility specialist advice to help reduce the chance of passing on the disease. MND state-based associations provide ongoing support through their Advisors.

A place to find support from others affected by Kennedy's Disease is via the private Facebook group *Kennedys Disease Downunder*.

More information

Your state MND Association or an MND clinic or service

Your General Practitioner or Neurologist

Kennedy's Disease Association: https://www.kennedysdisease.org/

To find out about motor neurone disease and other fact sheets in this series contact the MND Association in your state or territory ph. 1800 777 175 or visit www.mndaustralia.org.au

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