If you have been recently diagnosed with:

Spinal and Bulbar Muscular Atrophy – (SBMA) – Kennedy's Disease

What is SBMA-Kennedy's Disease?

Kennedy's Disease is a rare inherited neuromuscular condition. It's also known as:

- X-linked motor neuron disease
- X-linked recessive bulbospinal neuropathy or
- X-linked spinal and bulbar atrophy

It's an adult onset, form of SMA that mainly affects men. It's estimated that 1 in 40,000 people have the genetic fault that causes it and, because it's relatively rare, Kennedy's Disease is often initially misdiagnosed or goes undiagnosed for years.

What are the symptoms and effects?

Kennedy's Disease causes progressive weakening and wasting of the muscles, particularly in the arms and legs. This is due to the degeneration of lower motor neurons within the spinal cord and brainstem. It also causes hormonal changes. Symptoms and effects of Kennedy's Disease are described in more detail in the information published by the MND Associations and Kennedy's Disease UK (see over).

Which gene is affected?

Some of the genes that cause rare forms of SMA are associated with more than one condition, so please be aware that the website links suggested might provide information that is not just about SMA.

The affected gene is:

- The ***Androgen receptor (AR) gene***, on the X sex chromosome[1].

Further information can be found at: **www.ghr.nlm.nih.gov/gene/AR**

Inheritance pattern

Kennedy's Disease has an **X-linked Recessive** inheritance pattern.

> To find out more about this, please see our information sheet, '**The inheritance patterns of some rarer forms of SMA'.**

Support and resources

Kennedy's Disease UK:
Phone: 01604 250505
Website: www.kd-uk.com

MND Association:
Website: www.mndassociation.org (search for Kennedy's Disease)

Version: 1.1
Author: SMA UK Information Production Team
Published: August 2018
Last updated: March 2019
Next full review due: August 2021

References

1. La Spada *et al.* (1991) 'Androgen receptor gene mutations in X-linked spinal and bulbar muscular atrophy', *Nature,* 352, pp. 77–79.

We are grateful to the writers and reviewers who assist us in our information production. A list of who this includes may be viewed on our website: www.smauk.org.uk/our-writers-and-reviewers-panel or requested from supportservices@smauk.org.uk

If you have any feedback about this information, please do let us know at
supportservices@smauk.org.uk