

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

You can also read this guide on our website at smauk.org.uk/eyjm where you can follow all the links we give to further information.

Who is this for

This information is for you if you have been recently diagnosed with SBMA, which is also commonly referred to as Kennedy’s Disease.

1. What is SBMA?

SBMA is a rare inherited neuromuscular condition. It is also sometimes known as:

- X-linked motor neuron disease
- X-linked recessive bulbospinal neuropathy
- X-linked SBMA

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

2. What are the Symptoms and Effects?

SBMA 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 SBMA are described in more detail in the information published by the MND Associations and Kennedy's Disease UK (see below under Support and Resources).

3. Which Gene is Affected?

The affected gene is:

- The **Androgen Receptor (AR) gene**, on the X sex chromosome¹⁻².

Further information about the [AR Gene >](#).

Some of the genes that cause rarer forms of SMA are associated with more than one condition, so please be aware that the provided website links may give information about disorders additional to SMA.

4. Inheritance Pattern

SBMA has an X-linked Recessive inheritance pattern.

To find out more: [The inheritance patterns of some rarer forms of SMA >](#)

5. Support and Resources

[Kennedy's Disease UK >](#)

- Phone: 01604 250505

- Phone: 0808 802 6262

6. References

1. La Spada et al. (1991) 'Androgen receptor gene mutations in X-linked spinal and bulbar muscular atrophy', *Nature*, 352, pp. 77–79.
2. <https://www.omim.org/entry/313200> (last accessed 22nd January 2025)



Version: 2

Author: SMA UK Information Production Team

Last reviewed: January 2025

Next Review: January 2028

Links last checked: January 2025

This publication, and its links, provides information. It is meant to support, not replace, clinical and professional care.

Find out more about [how we produce our information](#)

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

www.smauk.org.uk • office@smauk.org.uk • 01789 267520

Unit 9, Shottery Brook Office Park, Timothy's Bridge Road, Stratford-upon-Avon, CV37 9NR

Registered Charity Number: 1106815