MDA Muscular Dystrophy Association

# Spinal Bulbar Muscular Atrophy (Kennedy's Disease)

**Spinal bulbar muscular atrophy (SBMA)**, also known as Kennedy's Disease, is a rare X-linked genetic disorder that primarily affects motor neurons.

It is a rare disorder that is estimated to affect approximately 1 in 40,000 to 1 in 50,000 males worldwide. The exact number of people affected can be difficult to determine due to underdiagnosis or misdiagnosis, as symptoms often begin later in life and may be mistaken for other conditions, such as amyotrophic lateral sclerosis (ALS), in early stages.

SBMA is caused by a **mutation in the androgen receptor (AR) gene on the X chromosome**. The mutation results in a specific DNA abnormality referred to as a CAG repeat. This abnormal expansion in the DNA leads to the production of a defective protein that accumulates and causes neurodegeneration.

This disease **primarily affects males**. **Females can be carriers and may exhibit mild symptoms** due to the random inactivation of the second X chromosome.

SBMA leads to **muscle weakness and wasting, starting with the bulbar muscles** (facial muscles, speech, swallowing, possibly breathing) and eventually progressing to other muscle groups.

**Symptoms usually begin in adulthood**, and the disease is characterized by progressive motor and sensory deficits.

Diagnosis of SBMA typically involves a combination of clinical evaluation, family history, and genetic testing. A blood test can confirm the diagnosis by checking for a CAG repeat expansion.

Other forms of motor neuron disease, such as ALS, progressive muscular atrophy (PMA), pseudobulbar affect (PBA), and primary lateral sclerosis (PLS), are not the same as SBMA, as they often affect different areas of the body and the underlying mechanisms are distinct.

# What Are the Signs and Symptoms of SBMA?

**Skeleton and muscle:**
- Progressive muscle weakness, especially in the arms, legs, and face
- Muscle atrophy (wasting) over time
- Fasciculations (muscle twitching) often noticed in the muscles of the chin
- Scoliosis (curved spine) in some cases
- Reduced deep tendon reflexes

**Speech and swallowing:**
- Difficulty speaking (dysarthria)
- Difficulty swallowing (dysphagia)

**Breathing:**
- Respiratory problems, including shortness of breath

**Cognition:**
- Cognitive changes, though generally mild compared to motor symptoms

**Endocrine:**
- Hormonal changes, including testicular atrophy and gynecomastia (enlarged breasts in men)

**Systemic:**
- Fatigue
- Mild sensory changes (e.g., numbness, tingling)



To learn more about SBMA and MDA's programs, visit **MDA.org** or contact the MDA Resource Center at **833-ASK-MDA1 (275-6321)** or **ResourceCenter@mdausa.org**.

# What Should I Know About SBMA?

1. Ongoing research is exploring potential treatments aimed at slowing or halting disease progression.

2. Clinical trials are investigating new drugs that target the androgen receptor or the underlying genetic mutations. Researchers are also working to improve diagnostic techniques and develop biomarkers to measure disease progression more effectively. Check out **clinicaltrials.gov** for more information on specific clinical trials.

3. Notable trials are underway globally, including studies on drug therapies, gene therapy, and methods to mitigate muscle degeneration.

4. Treatment focuses on symptom management and improving quality of life. This may include physical therapy to maintain mobility, speech therapy for swallowing difficulties, and respiratory care. Medications to manage pain, muscle spasms, and other symptoms are commonly used. Endocrine therapies, such as testosterone modulation, may also be considered to help manage hormone imbalances, although this has NOT been established in clinical trials.

# How Is SBMA treated?

There is currently no cure for SBMA as it is a genetic condition, but treatments aim to manage symptoms and restore function.

**Androgen receptor modulators (ARMs) or Androgen deprivation therapy (ADT)** may be used to manage some symptoms.

**Physical therapy and occupational therapy** may help manage mobility issues.

Clinical trials are investigating potential therapies, such as beta 2 agonists, which may address metabolic abnormalities. Research is also exploring drugs that modify insulin resistance related to SBMA.

Ongoing research in natural history studies and measuring disease progression may help refine treatment strategies.

A multidisciplinary team approach to treating SBMA is a valuable resource in anticipating changes in symptoms and improving quality of life. Connect with your local MDA Care Center at **mda.org/CareCenters**.

# MDA Glossary

### Androgen receptor (AR) gene
A gene that helps the body respond to male hormones like testosterone. Changes in this gene can affect how cells grow and work.

### Bulbar muscles
The group of muscles in the head and neck responsible for swallowing, speaking, and chewing

### Dysarthria
Difficulty speaking or forming words

### Dysphagia
Difficulty swallowing

### Fasciculations
Muscle twitching

### Gene mutation
A flaw in the DNA code

### Gynecomastia
The growth of breast tissue in males, which can happen because of hormone changes or imbalances

### Muscle atrophy
When muscles shrink or get weaker over time, often because they are not being used or because of a medical condition

### Scoliosis
An abnormal sideways curvature in the spine that occurs when weakened muscles are unable to hold the spine straight

### Testicular atrophy
When the testicles shrink and may not work as well, which can affect hormone levels and fertility

### X-linked genetic disorder
A condition passed down through the X chromosome, which means it usually affects males more often than females

This resource was developed with the expertise and knowledge of Jeffrey Rosenfeld, MD, PhD