

Spinal and bulbar muscular atrophy

Description

Spinal and bulbar muscular atrophy, also known as Kennedy disease, is a disorder of specialized nerve cells that control muscle movement (motor neurons). These nerve cells originate in the spinal cord and the part of the brain that is connected to the spinal cord (the brainstem).

Spinal and bulbar muscular atrophy mainly affects males and is characterized by muscle weakness and wasting (atrophy) that usually begins in adulthood and worsens slowly over time. Muscle wasting in the arms and legs results in cramping; leg muscle weakness can also lead to difficulty walking and a tendency to fall. Certain muscles in the face and throat (bulbar muscles) are also affected, which causes progressive problems with swallowing and speech. Additionally, muscle twitches (fasciculations) are common. Some males with the disorder experience unusual breast development (gynecomastia) and may be unable to father a child (infertile).

Frequency

This condition affects fewer than 1 in 150,000 males and is very rare in females.

Causes

Spinal and bulbar muscular atrophy results from a particular type of mutation in the *AR* gene. This gene provides instructions for making a protein called an androgen receptor. This receptor attaches (binds) to a class of hormones called androgens, which are involved in male sexual development. Androgens and androgen receptors also have other important functions in both males and females, such as regulating hair growth and sex drive.

The *AR* gene mutation that causes spinal and bulbar muscular atrophy is the abnormal expansion of a DNA segment called a CAG triplet repeat. Normally, this DNA segment is repeated up to about 36 times. In people with spinal and bulbar muscular atrophy, the CAG segment is repeated at least 38 times, and it may be two or three times its usual length. Although the extended CAG region changes the structure of the androgen receptor, it is unclear how the altered protein disrupts nerve cells in the brain and spinal cord. Researchers believe that a fragment of the androgen receptor protein containing the CAG segment accumulates within these cells and interferes with normal cell

functions. The nerve cells gradually die, leading to the muscle weakness and wasting seen in this condition. People with a higher number of CAG repeats tend to develop signs and symptoms of spinal and bulbar muscular atrophy at an earlier age.

[Learn more about the gene associated with Spinal and bulbar muscular atrophy](#)

- AR

Inheritance

This condition is inherited in an X-linked pattern. A condition is considered X-linked if the mutated gene that causes the disorder is located on the X chromosome, one of the two sex chromosomes. In males (who have only one X chromosome), a mutation in the only copy of the gene in each cell causes the disorder. In most cases, males experience more severe symptoms of the disorder than females (who have two X chromosomes). Females with a mutation in one copy of the *AR* gene in each cell are typically unaffected.

A few females with mutations in both copies of the gene have had mild features related to the condition, including muscle cramps and occasional tremors. Researchers believe that the milder signs and symptoms in females may be related to lower androgen levels.

A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

Other Names for This Condition

- Bulbospinal muscular atrophy, X-linked
- KD
- Kennedy disease
- Kennedy spinal and bulbar muscular atrophy
- Kennedy's disease
- SBMA
- X-linked spinal and bulbar muscular atrophy

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Kennedy disease (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1839259/>)

Genetic and Rare Diseases Information Center

- Kennedy disease (<https://rarediseases.info.nih.gov/diseases/6818/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov (<https://clinicaltrials.gov/search?cond=%22Spinal and bulbar muscular atrophy%22>)

Catalog of Genes and Diseases from OMIM

- SPINAL AND BULBAR MUSCULAR ATROPHY, X-LINKED 1; SMAX1 (<https://omim.org/entry/313200>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28Muscular+Atrophy,+Spinal%5BMAJR%5D%29+AND+%28%28spinal+and+bulbar+muscular+atrophy%5BTIAB%5D%29+OR+%28x-linked+bulbospinal+muscular+atrophy%5BTIAB%5D%29+OR+%28kennedy+disease%5BTIAB%5D%29+OR+%28kennedy's+disease%5BTIAB%5D%29+OR+%28sbma%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

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