Spinal and Bulbar Muscular Atrophy (SBMA; Kennedy Disease)

Record Status
This is a bibliographic record of a published health technology assessment. No evaluation of the quality of this assessment has been made for the HTA database.

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Authors' objectives
Spinal and bulbar muscular atrophy (SBMA), also known as Kennedy disease, is a progressive, adult-onset neuromuscular disorder characterized by degeneration of lower motor neurons, particularly, the anterior horn cells of the spinal cord and the bulbar nerves of the brainstem. The prevalence of SBMA is estimated to be 1 in 40,000; however, some believe the condition may be underdiagnosed. Due to the X-linked nature of the disorder (i.e., the gene is located on the X chromosome), SBMA occurs almost exclusively in males. However, female carriers may exhibit mild or subclinical features of SBMA. Patients with SBMA present with a variable combination of weakness and atrophy of the facial, bulbar, and limb muscles; sensory disturbances; and endocrine symptoms. The muscle weakness associated with SBMA typically manifests in the third to sixth decade of life and most often involves the proximal limb muscles. It is slowly progressive, causes difficulties with walking and climbing stairs, and may lead to wheelchair dependence in the later stages of disease. Deep tendon reflexes are generally decreased or absent in SBMA patients, and fasciculations (muscle twitches), muscle cramps, and tremor are common. Bulbar involvement typically includes difficulty with swallowing (dysphagia) and speaking (dysarthria), both of which result from weakness and atrophy of the tongue, jaw, and throat muscles. Symptoms related to mild androgen insensitivity are common among affected males and include gynecomastia (breast development), testicular atrophy, and impaired fertility. The progression of SBMA is typically slow, and life span is not usually significantly reduced. However, early death from respiratory problems or aspiration pneumonia may occur. SBMA is caused by the expansion of a CAG trinucleotide repeat in the androgen receptor (AR) gene located on the X chromosome at bands q11 to q12. This repeat is located in the N-terminal transactivation domain of the gene and is translated into a polyglutamine tract within the AR protein. In unaffected individuals, the number of CAG repeats in the AR gene is between 11 and 35. In contrast, SBMA patients generally carry more than 38 CAG repeats. Alleles containing 36 or 37 CAG repeats are considered reduced-penetrance alleles and may be associated with either a normal or SBMA phenotype.

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