JMLA Journal of the Medical Library Association

Supplemental electronic content to
J Med Lib Assoc. Apr;101(2):DOI: http://dx.doi.org/10.3163/1536-5050.101.2.010
www.mlanet.org

Analysis of inconsistencies in terminology of spinal and bulbar muscular atrophy and its effect on retrieval of research

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Table 2
Frequency of all spinal and bulbar muscular atrophy (SBMA) name variants, 1968–2010

	Number of records	Percent of name variants*
spinal and bulbar muscular atrophy	306	38.8%
Kennedy's disease	161	20.4%
spinobulbar muscular atrophy	100	12.7%
Kennedy disease	77	9.8%
X-linked spinal and bulbar muscular atrophy	76	9.6%
bulbospinal muscular atrophy	51	6.5%
bulbospinal neuronopathy	49	6.2%
spinal bulbar muscular atrophy	42	5.3%
X-linked recessive bulbospinal neuronopathy	30	3.8%
Kennedy syndrome	23	2.9%
Spinal and bulbar muscular-atrophy	18	2.3%
Kennedy-alter-sung	16	2.0%
Kennedy's syndrome	16	2.0%
X-linked bulbospinal muscular atrophy	14	1.8%
X-linked bulbospinal neuronopathy	14	1.8%
X-linked recessive bulbospinal muscular atrophy	14	1.8%
x-linked spinal and bulbar muscular-atrophy	13	1.6%
X-linked spinobulbar muscular atrophy	13	1.6%
bulbo-spinal muscular atrophy	11	1.4%
bulbar and spinal muscular atrophy	10	1.3%
X-linked recessive spinal and bulbar muscular atrophy	9	1.1%
bulbar spinal muscular atrophy	8	1.0%
bulbospinal muscular-atrophy	7	0.9%
bulbo-spinal neuronopathy	6	0.8%
Kennedy-Alter-Sung syndrome	6	0.8%
Kennedy-alter-sung type	6	0.8%
Progressive proximal spinal and bulbar muscular atrophy	6	0.8%
progressive proximal spinal and bulbar muscular atrophy of late onset	6	0.8%
spinal and bulbar amyotrophy	6	0.8%
Spinal-bulbar muscular atrophy	6	0.8%
X-linked bulbo-spinal neuronopathy	6	0.8%
bulbar-spinal muscular atrophy	5	0.6%
bulbospinal amyotrophy	5	0.6%
bulbo-spinal amyotrophy	5	0.6%
bulbospinal muscular-atrophy of late onset	4	0.5%
progressive spinal and bulbar muscular atrophy	4	0.5%
X-linked bulbar and spinal muscular atrophy	4	0.5%
X-linked spinal and bulbar muscular atrophy of late onset	4	0.5%
bulbospinal atrophy	3	0.4%
bulbospinal muscle atrophy	3	0.4%
Kennedy-Alter-Sung disease	3	0.4%

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Kennedys disease	3	0.4%
Kennedy-type-bulbo-spinal amyotrophy	3	0.4%
spinal and bulbar Kennedy's amyotrophy	3	0.4%
spinal and bulbar muscle atrophy	3	0.4%
X-linked bulbo-spinal muscular atrophy	3	0.4%
X-linked recessive spinobulbar muscular atrophy	3	0.4%
familial bulbo-spinal muscular atrophy	2	0.3%
hereditary proximal spinal and bulbar motor neuron disease	2	0.3%
hereditary proximal spinal and bulbar motor neuron disease of late onset	2	0.3%
hereditary spinobulbar muscular atrophy	2	0.3%
Kennedy disease/spinal bulbar muscular atrophy	2	0.3%
Kennedy's spinal amyotrophy	2	0.3%
Kennedy's spinal and bulbar amyotrophy	2	0.3%
Kennedys-syndrome	2	0.3%
Kennedy-Stefanis	2	0.3%
Kennedy-syndrom	2	0.3%
Late-onset X-linked recessive spinal and bulbar muscular atrophy	2	0.3%
progressive bulbar and spinal muscular atrophy	2	0.3%
SBMA motor neuronopathy	2	0.3%
spinal/bulbar muscular atrophy	2	0.3%
X-linked bulbospinal amyotrophy	2	0.3%
X-linked bulbo-spinal neuronopathy (BSN) of late onset	2	0.3%
X-linked bulbo-spinal neuronopathy of late onset	2	0.3%
X-linked Kennedy's spinal and bulbar muscular atrophy	2	0.3%
X-linked recessive bulbar spinal muscular atrophy	2	0.3%
X-linked recessive bulbo-spinal muscular atrophy	2	0.3%
X-linked recessive bulbospinal neuropathy	2	0.3%
X-linked spinal muscular atrophy	2	0.3%
adult onset spinal/bulbar muscular atrophy	1	0.1%
adult onset X-linked recessive bulbo-spinal muscular atrophy	1	0.1%
adult sex-linked proximal hereditary motor neuropathy	1	0.1%
adult spinal and bulbar muscular atrophy	1	0.1%
adult spinal and bulbar muscular atrophy with X-linked recessive inheritance	1	0.1%
adult-onset bulbospinal neuronopathy	1	0.1%
adult-onset spinal and bulbar amyotrophy	1	0.1%
adult-type familial bulbospinal atrophy	1	0.1%
bulbar and spinal muscular atrophy of late onset	1	0.1%
bulbar and spinal muscular atrophy of X-linked recessive trait	1	0.1%
bulbar-spinal amyotrophy	1	0.1%
bulbar-spinal muscular atrophy (BSMA) of the Kennedy-Alter-Sung type	1	0.1%
bulbo spinal muscular atrophy	1	0.1%
bulbo spinal neuronopathy	1	0.1%
bulbo-spinal lower motor neuron disease	1	0.1%
bulbospinal muscular atrophy of late onset	1	0.1%
bulbospinal muscular disease	1	0.1%
bulbospinal muscular disease of the Kennedy-Alter-Sung type	1	0.1%
chromosome X-linked recessive bulbospinal neuronopathy	1	0.1%
chronic X-linked recessive spinal amyotrophy	1	0.1%
distal spinal bulbar muscular atrophy	1	0.1%

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early-onset and rapidly progressive X-linked spinal and bulbar muscular atrophy	1	0.1%
familial bulbar spinal muscular-atrophy	1	0.1%
familial bulbospinal atrophy	1	0.1%
familial bulbospinal muscular-atrophy	1	0.1%
Familial bulbospinal neuronopathy	1	0.1%
familial progressive bulbar and spinal muscular atrophy	1	0.1%
Familial progressive bulbar-spinal muscular atrophy	1	0.1%
Hereditary proximal spina; and bulbar motor neuron disease	1	0.1%
hereditary motor neuron disease: The proximal, adult, sex-linked form	1	0.1%
hereditary proximal spina; and bulbar motor neuron disease of late onset	1	0.1%
Idiopathic bulbo-spinal muscle atrophy	1	0.1%
Kennedy Alter-Sung syndrome	1	0.1%
Kennedy disease/syndrome	1	0.1%
Kennedy gene	1	0.1%
Kennedy syndrome—bulbo-spinal muscular atrophy	1	0.1%
Kennedy type of SMA	1	0.1%
Kennedy-Alter-Sung (KAS) disease	1	0.1%
Kennedy-alter-sung (kas) syndrome	1	0.1%
Kennedy-Alter-Sung syndrome-like proximal dominant neurogenic atrophy	1	0.1%
Kennedy-Alter-Sung type bulbo-spinal muscular atrophy	1	0.1%
Kennedy-Alter-Sung type spinal muscular atrophy	1	0.1%
Kennedy's bulbospinal atrophy	1	0.1%
Kennedy's medullo-spinal amyotrophy	1	0.1%
Kennedy's neuron disease	1	0.1%
Kennedys syndrome	1	0.1%
Kennedy's X-linked bulbospinal amyotrophy	1	0.1%
Kennedys-disease	1	0.1%
Kennedy-Stefanis chronic spinal amyotrophy	1	0.1%
Kennedy-Stefanis disease	1	0.1%
Kennedy-stefanis type	1	0.1%
Kennedy-Syndrome	1	0.1%
late onset progressive bulbar and spinal muscular atrophy	1	0.1%
Late onset x linked recessive spinal and bulbar muscular atrophy	1	0.1%
Late onset x-linked progressive spinal muscular atrophy	1	0.1%
late onset X-linked recessive bulbospinal neuronopathy	1	0.1%
late progressive recessive X-linked proximal spinal and bulbar amyotrophy	1	0.1%
late recessive X-linked Kennedy's spinal and bulbar amyotrophy	1	0.1%
late recessive x-linked spinal and bulbar kennedy's amyotrophy	1	0.1%
muscular atrophy, spinal and bulbar (SBMA), X-linked Kennedy type	1	0.1%
progressive bulbar-spinal amyotrophy	1	0.1%
Progressive bulbospinal amyotrophy	1	0.1%
progressive bulbospinal muscular atrophy of sex-linked recessive trait	1	0.1%
progressive spinal/bulbar muscular atrophy	1	0.1%
proximal spinal muscular atrophy	1	0.1%
Pseudomyopathic spinal amyotrophy	1	0.1%
recessive x-coupled kennedy spinal and bulbar amyotrophy	1	0.1%
recessive X-linked amyotrophic spinobulbar muscular atrophy	1	0.1%

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sex-linked proximal hereditary motor neuropathy	1	0.1%
sex-linked recessive bulbar spinal muscular atrophy	1	0.1%
spinal and bulb muscular atrophy	1	0.1%
spinal and bulbar (SBMA), X-linked Kennedy type	1	0.1%
spinal and bulbar muscular atrophy with X-linked recessive inheritance	1	0.1%
spinal and bulbar muscular dystrophy	1	0.1%
spinal and bulbular atrophy	1	0.1%
spinal bulbar and muscular atrophy	1	0.1%
spinal muscular atrophy (SMA) of Kennedy type	1	0.1%
spinal muscular atrophy of the Kennedy type	1	0.1%
spinal-and-bulbar-muscular-atrophy	1	0.1%
spino bulbar muscular atrophy	1	0.1%
spinobulbar amyotrophy	1	0.1%
spinobulbar and muscular atrophy	1	0.1%
spino-bulbar muscular atrophy	1	0.1%
spinobulbare muskelatrophie	1	0.1%
spinobulbomuscular atrophy	1	0.1%
type Kennedy	1	0.1%
x chromosomal bulbospinal neuropathy	1	0.1%
X chromosome associated progressive bulbospinal neuropathy	1	0.1%
x chromosome-linked bulbospinal neuronopathy	1	0.1%
X chromosome-linked spinal muscular atrophy	1	0.1%
X linked spinobulbar muscular atrophy	1	0.1%
X-BSMA with neuropathy	1	0.1%
x-chromosomal bulbospinal muscular atrophy	1	0.1%
X-chromosomal recessive bulbospinal neuronopathy	1	0.1%
x-chromosomal recessive spinobulbar muscular atrophy	1	0.1%
x-chromosomal recessive spinobulbar muscular atrophy (type Kennedy)	1	0.1%
X-chromosome linked bulbospinal muscular atrophy	1	0.1%
X-chromosome-linked adult bulbospinal neuronopathy	1	0.1%
X-linked adult-onset bulbospinal muscular atrophy	1	0.1%
X-linked adult-onset SBMA	1	0.1%
X-linked bulbar spinal muscular atrophy	1	0.1%
X-linked bulbospinal muscular atrophy Kennedy	1	0.1%
X-linked bulbospinal muscular-atrophy	1	0.1%
X-linked bulbospinal neuronopathy Kennedy	1	0.1%
X-linked bulbospinomuscular atrophy	1	0.1%
X-linked muscular atrophy	1	0.1%
X-linked muscular-atrophy	1	0.1%
x-linked progressive spinal muscular atrophy	1	0.1%
X-linked proximal spinal and bulbar amyotrophy	1	0.1%
X-linked recessive bulbar-spinal muscular atrophy	1	0.1%
X-linked recessive bulbospinal amyotrophy	1	0.1%
x-linked recessive bulbospinal muscular-atrophy	1	0.1%
x-linked recessive bulbo-spinal muscular-atrophy	1	0.1%
X-linked recessive bulbo-spinal neuronopathy	1	0.1%
X-linked recessive spinal amyotrophy	1	0.1%
x-linked recessive spinal and bulbar muscular-atrophy	1	0.1%
X-linked spinal and bulbal muscular atrophy	1	0.1%
X-linked spinal and bulbar atrophy of late onset	1	0.1%

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X-linked spinal and bulbar muscular-atrophy of late onset	1	0.1%
X-linked spinal bulbar and muscular atrophy	1	0.1%
x-linked spinal bulbar muscular-atrophy	1	0.1%
X-linked spino-bulbar muscular atrophy	1	0.1%
X-linked spinomuscular atrophy	1	0.1%
X-linked: spinobulbar muscular atrophy	1	0.1%
bulbar spinal neuronopathy	0	—
bulbar-spinal muscular-atrophy	0	—
bulbar-spinal neuronopathy	0	—
bulbo-SMA	0	—
Kennedy-disease	0	—
Kennedy's-disease	0	—
Kennedy's-syndrome	0	—
Kennedy-Stefanis syndrome	0	—
Spinobulbornuscular atrophy	0	—
x chromosomal bulbospinal neuronopathy	0	—
X-linked bulbospino muscular-atrophy	0	—
X-linked spinal and bulbar muschular atrophy	0	—
x-linked spinal bulbar muscular atrophy	0	—
Total records	1,352	
Total all SBMA variants (with duplicates) count for each year	788	
All SBMA variants (without duplicates) count for each year	756	

* Records may appear in more than one category. Percentage is the percentage of all name variants (788), not of unique records.