Title: Spinocerebellar Ataxia Type 14 *GeneReview* Table 2
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Table 2. SCA14 Genotypes and Phenotypes

Familial (F)/ Sporadic (S)	Predicted Amino Acid Changes	Nucleotide Changes	Exon	Mean Age of Onset (Range in yrs)	Gait Ataxia	Dysarthria	Abnormal Eye Movement	Myoclonus	Tremor / Chorea	Decreased Sense	Depression	Cognitive Deficits	Additional Signs	Tendon Reflexes	MRI Cerebellar Atrophy	Author
F	p.R26G	c.76a>g	1	37(15-68)	+	+	9/10	–	1/10	–	–	–	ptosis	↓/↑	+	Sailer et al 2012
F	p.R41P	c.122g>c	1	46(29-58)	+	+	–	–	–	–	–	–	–	nl	+	Chen et al 2005
F	p.G63V	c.188g>t	2	Childhood-50	+	+	+	–	–	–	–	–	Dystonia	↓	+	Nolte et al 2007b
F	p.C77S	c.229t>a	3	40 (26, 56)	+	+	+	–	–	+	+	–	–	N/A	+	Wieczorek et al 2007
S	p.K100-H101del	c.296-301del	4	38	+	+	+	–	–	–	+	–	–	↑	+	Chen et al 2005
F	p.H101Y	c.301c>t	4	31(10-50)	+	+	+	–	–	–	–	–	–	↓/↑	+	Chen et al 2003
F	p.H101Y	c.301c>t	4	adolescence	+	N/A	N/A	–	+	–	+	–	Psychosis	N/A	+	Nolte et al 2007a
F	p.H101Q	c.303c>g	4	32 (40-20)	+	+	+	–	–	–	–	–	–	N/A	+	Alonso et al 2004
F	p.C114Y	c.341g>a	4	28.8(17-37)	+	+	2/5	–	2/5	–	–	1/5	Plantar+	nl/↑	+	Klebe et al 2005
F	p.G118D	c.353g>a	4	38.3(21-59)	+	+	+	2/23	1/23	8/22	–	–	Dystonia	nl/↑/↓	+	van de Warrenburg et al 2003, Verbeek et al 2005, Visser et al 2007
F	p..S119P	c.355t>c	4	42(35-51)	+	+	–	–	–	+	+	–		↑	+	Chen et al 2003
F	p.S119F	c.356c>t	4	52.7(14-70)	+	–	–	–	–	–	–	1/5	Seizures	nl/↑	+	Hiramoto et al 2006
F	p.G123R	c.367g>a	4	48.3(40-60)	+	+/–	2/3	–	–	2/3	–	1/3	Deafness	N/A	+	Klebe et al 2005
F	p.G123E	c.368g>a	4	36(24-48)	+	+/–	–	1/2	1/2	1/2	–	2/2		N/A	+	Klebe et al 2005
F	p.Q127R	c.380a>g	4	28(12-42)	+	+	+	5/11	–	–	–	–		↓	+	Yabe et al 2003
S	p.G128D	c.383g>a	4	22, 44	+	+	+	–	–	–	–	–		nl	+	Chen et al 2003, Morita et al 2006
F	p.G128D	c.383g>a	4	32 (20-49)	+	+	+	–	+	–	+?	2/3	Dystonia Vertigo ↓vision	↑	+	Miura et al 2009
F	p.C131R	c.391t>c	4	childhood-50	+	+	–	–	+	–	–	–	Seizures?	N/A	+	Dalski et al 2006
F	p.C131Y	c.392g>a	4	adult (18, 50)	+	+	–	–	–	–	–	–	–	↑	+	Klebe et al 2005
F	p.V138E	c.413t>a	4	22(3-45)	+	+/–	+	1/7	2/7	2/7	–	–	Plantar+	nl/↑	+	Vlak et al 2006
F	p.H139G	c.47c>t	5	28.1(10-45)	+	+/–	+	–	1/13	6/13	–	3/13		nl/↑	+	Koht e al 2012
F	p.C150F	c.449,450gc>tt	5	31.3(19-44)	+	+/–	+	–	1/6	4/6	–	–		↑	+	Fahey et al 2005
F	p.G360S	c.1078g>a	10	36.5(20-53)	+	+/–	–	–	2/2	–	–	–	Rippling	N/A	+	Klebe et al 2005
F	p.S361G	c.1081a>g	10	28(5-60)	+	+	–	–	–	–	+	–		nl	+	Chen et al 2005

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F	p.F643L	c.1928t>c	18	34(child-60)	+	+/–	10/18	Facial myokymia	4/18	4/18	–	13/18		↑	+	Stevanin et al 2004
F	p.F643L	c.1928t>c	18	20	+	+	–	–	–	+	–	–		N/A	+	Klebe et al 2005
F	p.V692G	c.2075t>g	18	15	+	+	–	–	2/2	–	–	N/A	Plantar+	↑	+	Klebe et al 2005
F	p.M697I-ex13	c.2089-2192del	18	Childhood-60	+	N/A	N/A	1/3	+	N/A		–		↓	+	Asai et al 2009

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