

**Table 5. Mutations of the *SOX10* gene characterized in Waardenburg syndrome.**

Exon #	cDNA*	RNA/protein	In original paper	Inheritance	Phenotype	Remarks	Origin	Reference
Exon 3	<b>c.50-73delinsGCCCGA CGCTAGGGCCCTAG</b>	<b>p.Ser17CysfsX7</b>		<b>?</b>	<b>WS4</b>			<b>This paper</b>
	c.126_127delGCinsCT	p.Arg43X	R43X ; 126GC>CT	de novo	WS4			[Pingault et al., 2002]
	c.169delG	p.Glu57SerfsX52	nt168delG	de novo	WS4		China	[Sham et al., 2001]
	c.249C>A	p.Tyr83X	Y83X (C→A at 249)	de novo	WS4			[Pingault et al., 1998]
	c.328_329delGC	p.Ala110LeufsX23	327delCG	familial	WS4			[Pingault et al., 2002]
	c.404G>C	p.Ser135Thr	S135T	sporadic	YDBHS (mild) / WS2			[Bondurand et al., 1999]
Intron 3	c.428+2T>G	Splice defect	428+2T-G	familial	WS4			[Touraine et al., 1998]
Exon 4	c.470C>T	p.Ala157Val		de novo	WS4			[Morin et al., 2008]
	c.477_482dupGCTCCG	p.Leu160_Arg161dup	482ins6 (GCTCCG)	de novo	WS4			[Pingault et al., 1998]
	c.506delC	p.Pro169ArgfsX117	p.Pro169fsX284	<b>?</b>	<b>WS2</b>		Japan	[Iso et al., 2008]
	<b>c.519C&gt;G</b>	<b>p.Tyr173X</b>		<b>familial</b>	<b>WS4 (no depigmentation)</b>	<b>Mother and brother have the mutation and present with isolated deafness</b>		<b>This paper</b>
	c.521A>C	p.Gln174Pro	Q174P	<b>?</b>	PCW; global hypopigmentation	+ visual impairment + inner ear abnormalities + aplasia of cochlear nerves and olfactory bulbs		[Barnett et al., 2009]
	c.565G>T	p.Glu189X	E189X (G→T at 565)	de novo	WS4			[Pingault et al., 1998]
	<b>?</b>	p.Tyr207X	Y207X	de novo	WS4			[Southard-Smith et al., 1999]
	<b>c.644_648delGGCAC</b>	<b>p.Arg215ProfsX64</b>		<b>de novo</b>	<b>WS4</b>	<b>+ inner ear abnormalities</b>		<b>This paper</b>
Intron 4	c.698-2A>C	Splice defect		de novo	PCW	+ inner ear abnormalities + autistic-like behaviour	Turkey	[Sznajer et al., 2008]
Exon 5	c.700C>T	p.Gln234X	Q234X	de novo	PCWH (po)	+ hypoventilation + inner ear abnormalities	Algeria	[Pingault et al., 2002]
	c.748C>T	p.Gln250X	Q250X	sporadic	PCWH	+ hypoventilation		[Inoue et al., 2002]
	c.752C>A	p.Ser251X	S251X	de novo	PCWH		France	[Touraine et al., 2000]
	c.780delG	p.Arg261AlafsX25	778delG	de novo	WS4		Japan	[Pingault et al., 2002]
	c.797delG	p.Gly266AlafsX20	795delG	de novo	demyelinating peripheral neuropathy, po, deafness			[Pingault et al., 2000]
	<b>c.811delA</b>	<b>p.Ile271SerfsX15</b>		<b>sporadic</b>	<b>WS4</b>	<b>+ inner ear abnormalities</b>		<b>This paper</b>
	c.847_848insT	p.His283LeufsX11	847insT, fr297X	de novo	PCWH			[Inoue et al., 2004]
	c.915delG	p.His306ThrfsX5			PCWH	+ inner ear abnormalities		[Vinuela et al., 2009]
	<b>c.921delA</b>	<b>p.Gly308AlafsX3</b>		<b>?</b>	<b>PCWH</b>	<b>+ inner ear abnormalities</b>		<b>This paper</b>
	c.938dupA	p.Tyr313X	938insA, Y313X	de novo	PCWH			[Inoue et al., 2004]
	c.939C>A	p.Tyr313X	Y313X	familial	PCWH	+ hypogonadism; suspected mosaicism in the mother (unaffected)	Germany	[Touraine et al., 2000]
	c.939C>A	p.Tyr313X	Y313X	sporadic	PCWH		France	[Touraine et al., 2000]
	c.939C>G	p.Tyr313X	939C→G, Y313X	de novo	PCWH	+ inner ear abnormalities	Japan	[Inoue et al., 2004]
	c.1037C>G <sup>§</sup>	p.Ser346X <sup>§</sup>	S384X	de novo	PCWH			[Verheij et al., 2006]
	<b>c.1047dupT</b>	<b>p.Val350CysfsX52</b>		<b>familial</b>	<b>WS4</b>			<b>This paper</b>
	c.1059delA	p.Ala354ProfsX3	1058delA	sporadic	WS4			[Touraine et al., 1998]

	c.1077_1078delGA	p.Glu359AspfsX42	1076delGA	familial	WS4		[Pingault et al., 1998]
	c.1090C>T	p.Gln364X	1090C→G, Q364X	de novo	PCWH		[Inoue et al., 2004]
	c.1114C>T	p.Gln372X	Q372X	de novo	PCWH (po)	+ inner ear abnormalities	[Pingault et al., 2002]
	?	p.Ser376X	376X	?	WS4		[Toki et al., 2003]
	?	p.Gln377X	Q377X	familial	PCWH	Suspected mosaicism in the father (unaffected)	[Southard-Smith et al., 1999]
	<b>c.1195_1196delCA</b>	<b>p.Gln399ValfsX2</b>		<b>de novo</b>	<b>WS4 (no dépigmentation)</b>	<b>+ inner ear abnormalities</b>	<b>This paper</b>
	c.1399T>A	p.*467LysextX86	X467K	de novo	PCWH	China	[Sham et al., 2001]
	c.1400_1411delAAAAGG GGGCCCT	p.*467CysextX82		de novo	PCWH	+ dystopia canthorum Japan	[Inoue et al., 1999]
	<b>c.1401A&gt;C</b>	<b>p.*467TyrextX86</b>		<b>de novo</b>	<b>PCWH</b>		<b>This paper</b>
Non synonymous non pathogenic variants:							
Exon 3	c.274G>C	p.Val92Leu			WS4	no functional effect in vitro; the patient also carries a full <i>SOX10</i> deletion that is responsible for the disease	[Bondurand et al., 2007]

\*cDNA nucleotide numbering with +1 as the A of the initiation codon in the reference sequence NM\_006941.3.

#Nomenclature of exons and introns as in [Pingault et al., 1998]. The 5'UTR structure of the gene is unknown and the number of exons is controversial.

§Confirmed by the author

PCWH = Peripheral and Central nervous system, Waardenburg syndrome, Hirschsprung disease; po = pseudo-obstruction; HD = Hirschsprung disease; YDBHS = Yemenite deaf blind hypopigmentation syndrome