

**Table 2. Mutations of the *MITF* gene characterized in Waardenburg syndrome and the related Tietz syndrome.**

Exon	cDNA*	ARN/protein	In original paper	Inheritance	Phenotype	Remarks	Origin	Reference
Intron 1M	c.33+1G>A	splice defect	GT→AG donor site	familial	WS2	Both families are possibly related	South Australia	[Tassabehji et al., 1994]
	c.33+1G>A	splice defect	nt 153+1 G→A	familial	WS2			[Tassabehji et al., 1995]
Exon 4	<b>c.411delG</b>	<b>p.Met138TrpfsX17</b>		<b>?</b>	<b>WS2 + osteochondromas</b>			<b>This paper</b>
Intron 4	c.442-2A>C	splice defect	AG→CG acceptor site	familial	WS2		Japan	[Tassabehji et al., 1994]
	c.442-1G>C	r.442_484del43; p.Leu148ValfsX20	IVS4-1G→C	familial	WS2			[Monma et al., 2008]
Exon 6	c.579_580delGT	p.Glu193AspfsX2	2bp del (GT) at nt 699	familial	WS2		USA / Ireland	[Lautenschlager et al., 1996]
	c.630C>G	p.Asn210Lys	G to C at nt 600; Asn210Lys	familial	Tietz syndrome			[Smith et al., 2000]
Intron 6	<b>c.634+1G&gt;A</b>	<b>splice defect</b>		<b>familial</b>	<b>WS2</b>			<b>This paper</b>
Exon 7	c.639delA	p.Glu213AspfsX8	c.639delA	familial	WS2	Carried by two different haplotypes	China	[Chen et al., 2008]
	c.640C>T	p.Arg214X	C760→T	familial	WS2		[Nobukuni et al., 1996]	
	c.640C>T	p.Arg214X	C760→T	familial	WS2		India	[Lalwani et al., 1998]
	<b>c.647G&gt;A</b>	<b>p.Arg216Lys</b>		<b>de novo</b>	<b>WS2</b>		<b>This paper</b>	
	c.?	p.Arg217del	del (R217)	familial	Tietz syndrome			[Tassabehji et al., 1995]
	c.?	p.Arg217del	del (R217)	de novo	Tietz syndrome			[Izumi et al., 2008]
	<b>c.649_651delAGA</b>	<b>p.Arg217del</b>	<b>del p.R217</b>	<b>familial</b>	<b>WS2/Tietz syndrome</b>	+ ocular albinism and a missense variation of the <i>TYRP1</i> gene in the propositus		<b>[Chiang et al., 2009]</b>
	<b>c.649_651delAGA</b>	<b>p.Arg217del</b>		<b>familial</b>	<b>Tietz syndrome</b>			<b>This paper</b>
	<b>c.671T&gt;G</b>	<b>p.Ile224Ser</b>		<b>familial</b>	<b>WS2</b>			<b>This paper</b>
Intron 7	<b>c.710+1G&gt;A</b>	<b>splice defect</b>		<b>sporadic(?)</b>	<b>WS2</b>	<b>In silico analysis predicts a possible loss of splice site</b>		<b>This paper</b>
	<b>c.710+5G&gt;T</b>	<b>splice defect ?</b>		<b>sporadic</b>	<b>WS2</b>		<b>This paper</b>	
Exon 8	c.748T>C	p.Ser250Pro	S250P	familial	WS		Togo	[Tassabehji et al., 1995]
	c.?	p.Tyr253Cys	Y253C	?	WS2			[Read and Newton, 1997]
	c.775C>T	p.Arg259X	C895→T	familial	WS2			[Nobukuni et al., 1996]
	<b>c.775C&gt;T</b>	<b>p.Arg259X</b>		<b>familial</b>	<b>WS2</b>			<b>This paper</b>
	<b>c.806delA</b>	<b>p.Asn269ThrfsX43</b>		<b>?</b>	<b>WS2</b>			<b>This paper</b>
	c.824delA	p.Glu275GlyfsX37	delA at nt 944 (codon 275)	familial	WS2	+ ocular albinism and a functionally significant polymorphism of the <i>TYR</i> gene		[Morell et al., 1997]
	c.?	p.Asn278Asp	N278D	sporadic(?)	WS2	Inherited from an unaffected father		[Tassabehji et al., 1995]
Exon 9	c.892T>C	p.Ser298Pro	S298P	familial	WS2			[Tassabehji et al., 1995]
Non synonymous non pathogenic variants:								
Exon 5	<b>c.483A&gt;T</b>	<b>p.Gln161His</b>		<b>familial</b>	<b>WS2</b>	<b>Does not cosegregate with the disease</b>		<b>This paper</b>

Exon 6	c.608G>A	p.Arg203Lys	R203K	familial	WS2	Does not cosegregate with the disease	[Tassabehji et al., 1995]
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\*cDNA nucleotide numbering with +1 as the A of the initiation codon in the reference sequence NM\_000248.2 corresponding to the M (melanocytic) isoform of microphthalmia.