

Table 4. Mutations of the *EDN3* gene characterized in Waardenburg syndrome.

Exon	cDNA*	RNA/protein	In original paper	Inheritance	Phenotype	Remarks	Origin	Reference
Exon 2	c.163G>T	p.Glu55X	E55X	familial	WS4 in homozygotes	Consanguineous family		[Bidaud et al., 1997]
	c.262_263delinsT	p.Ala88SerfsX121	262 GC→T	sporadic	WS4 in homozygotes	Unrelated parents		[Edery et al., 1996]
	c.262_263delinsT	p.Ala88SerfsX121		?	WS4 in homozygotes	No consanguinity	Bosnia	This paper
	c.286C>T	p.Arg96Cys		sporadic	WS4 in homozygotes	No consanguinity		This paper
	c.293C>A	p.Thr98Lys		sporadic	WS4 in homozygotes	Consanguineous families; this variant has been excluded as a frequent polymorphism in a control population of India	India to Caribbean via Madagascar	This paper
	c.293C>A	p.Thr98Lys		familial	WS4 in homozygotes; ±depigmentations in heterozygotes		India	This paper
	c.293C>A	p.Thr98Lys		sporadic	WS4 in compound heterozygote	Patient carries a p.Cys173Arg mutation on the second allele.	India	This paper
	c.335A>G	p.His112Arg		familial	WS4 in homozygotes; depigmentations in heterozygotes	Consanguineous family		[Vinuela et al., 2009]
Exon 3	c.380A>G	p.Tyr127Cys	Y127C	familial	depigmentations ± deafness in heterozygotes		India	[Pingault et al., 2002]
	c.476G>T	p.Cys159Phe		familial	WS4 in homozygotes; depigmentations ± deafness in heterozygotes	Consanguineous family	Pakistan	[Hofstra et al., 1996]
	c.476G>T	p.Cys159Phe		familial	WS4 in homozygotes	Consanguineous family	Pakistan	This paper
	c.476G>T	p.Cys159Phe		familial	WS4 in homozygotes	Consanguineous family	Pakistan	This paper
	c.507C>A	p.Cys169X	C169X	familial	WS4 in heterozygotes		Yugoslavia	[Pingault et al., 2001]
	c.517T>C	p.Cys173Arg		sporadic	WS4 in compound heterozygote	Patient carries a p.Thr98Lys mutation on the second allele.	India	This paper
Non synonymous polymorphisms as reported in literature and in dbSNP (when confirmed in at least one control population):								
Exon 1	c.49G>A	p.Ala17Thr	A17T			rs11570255 in dbSNP frequency 0.007		[Garcia-Barcelo et al., 2004]
Exon 4	c.565dupA	p.Thr189AsnfsX10				rs11570344 in dbSNP frequency 0.006		
Exon 5	c.670G>A	p.Ala224Thr	A224T			rs11570351 in dbSNP frequency 0.006		

*cDNA nucleotide numbering with +1 as the A of the initiation codon in the reference sequence NM_000114.2.