

Title: Aterial Tortuosity Syndrome *GeneReview* Table 3 – Reported *SLC2A10* Mutations
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 Note: The following information is provided by the authors and has not been reviewed by *GeneReviews* staff.

Table 3. Reported *SLC2A10* Mutations in Families with Arterial Tortuosity Syndrome

Family	Origin	Consanguinity	DNA Nucleotide Change (ref seq NM_030777.3)	Protein Amino Acid Change (ref seq NP_110404.1)	Reference
1	Moroccan	Yes	c.510G>A	p.(Trp170Ter)	Coucke et al 2006
2	Moroccan	Yes	c.510G>A	p.(Trp170Ter)	Coucke et al 2006
3	Moroccan	Yes	c.961delG	p.(Val321fs)	Coucke et al 2006
4	Italy	Yes	c.1334delG	p.(Gly445fs)	Coucke et al 2006
5	Middle-East	Yes	c.243C>G	p.(Ser81Arg)	Coucke et al 2006
6	Middle-East	Yes	c.243C>G	p.(Ser81Arg)	Coucke et al 2006
7	Italy	No	c.1309G>A c.1330C>T	p.(Glu437Lys) p.Arg444ter	Drera et al 2007b
8	Belgium	Yes	c.685C>T	p.(Arg229Ter)	Callewaert et al 2008
9	American (Germany)	No	c.1276G>T c.1334delG	p.(Gly426Trp) p.(Gly445fs)	Callewaert et al 2008
10	Poland	No	c.730_733delCTAA c.1309G>A	p.(Leu244fs) p.(Glu437Lys)	Callewaert et al 2008
11	Turkey	Yes	c.737G>A	p.(Gly246Glu)	Callewaert et al 2008
12	USA (Poland/ Sicily)	No	c.1309G>A c.1411+480_c.1547+ 299del	p.(Glu437Lys) p.(Gly471_Arg515delTerfs)	Callewaert et al 2008
13	France	No	c.394C>T	p.(Arg132Trp)	Callewaert et al 2008
14	Spain	No	c.692G>A c.1334delG	p.(Arg231Gln) p.(Gly445fs)	Callewaert et al 2008
15	The Netherlands	No	c.394C>T c.1276G>T	p.(Arg132Trp) p.(Gly426Trp)	Callewaert et al 2008
16	UK	No	c.1334delG c.1411+480_c.1547+ 299del	p.(Gly445fs) p.(Gly471_Arg515delXfs)	Callewaert et al 2008
17	Sudan	Yes	c.425G>T	p.(Gly142Val)	Callewaert et al 2008

Family	Origin	Consanguinity	DNA Nucleotide Change (ref seq NM_030777.3)	Protein Amino Acid Change (ref seq NP_110404.1)	Reference
18	The Netherlands	No	c.394C>T c.1276G>T	p.(Arg132Trp) p.(Gly426Trp)	Callewaert et al 2008
19	France	No	c.1334G>A	p.(Gly445Glu)	Callewaert et al 2008
20-30 (same ancestry)	Middle-East (Qatar)	Yes	c.243C>G	p.(Ser81Arg)	Faiyaz-UI-Haque et al 2008
31	Middle-East	Yes	c.313C>T	p.(Arg105Cys)	Faiyaz-UI-Haque et al 2009
32	Middle-East	Yes	c.243C>G	p.(Ser81Arg)	Faiyaz-UI-Haque et al 2009
33	Middle-East	Yes	c.756C>A	p.(Cys252Ter)	Zaidi et al 2009
34	NR	Likely	c.685C>T	p.(Arg229Ter)	Allen et al 2009
35	Italian	No	c.685C>T c.756C>A	p.(Arg229Ter) p.(Cys252Ter)	Ritelli et al 2009
36	Italian	No	c.691C>T c.1334delG	p.(Arg231Trp) p.(Gly445fs)	Ritelli et al 2009
37	Italian	Yes	c.1411+1G>A	p.(Val430_I470del)	Castori et al 2012
38	NR	Yes	c.510G>A	p.(Trp170Ter)	Moceri et al 2013
39	Japan	No	c.417T>A c.692G>A	p.(Tyr139Ter) p.(Arg231Gln)	Takahashi et al 2013

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