Title: Cerebrotendinous Xanthomatosis *GeneReview* Table 3
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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 3. Selected *CYP27A1* Allelic Variants

Variant Classification	DNA Nucleotide Change	Predicted Protein Change	References or Reference Sequences
Pathogenic	c.5_6insC	p.Met1fsTer178	Segev et al 1995
	c.10_11 ins10bp	p.Ala7fsTer	Szlago et al 2008
	c.73delG		Tian & Zhang 2011
	c.206delC		Chen et al 2011
	c.355delC	p.Met118fsTer	Leitersdorf et al 1994
		(p.Arg104Gln) [1]	Nozue et al 2010
	c.369_375delGTACCCA		Tian & Zhang 2011
	c.379C>T	p.Arg127Trp	Verrips et al 1999
	c.380G>A	p.Arg127Gln	Watts et al 1996
		p.Trp133Ter	Lee et al 2001
	c.409C>T	p.Arg137Trp	Nakashima et al 1994
	c.433 G>A	p.Gly145Arg	Gallus et al 2010
	c.434G>A	p.Gly145Glu	Lamon-Fava et al 2002
	c.435G>T	p.Gly145Gly (alternative splicing)	Chen et al 1998b
	c.446+1G>A		Verrips et al 2000
	c.475C>T	p.Gln159Ter	Verrips et al 1996
	c.525-526delG	p.Thr175fsTer	Verrips et al 1996
	c.583G>T	p.Glu195Ter	Wakamatsu et al 1999
	c.599C>T	p.Gln191Ter	Huang et al 2011
	c.646G>C	p.Ala216Pro	Garuti et al 1996
	c.647-1 G>T		Gallus et al 2010
	c.691C>T	p.Arg231Ter	Garuti et al 1997
	c.745C>T	p.Gln249Ter	Verrips et al 2000
	c.752 C>A	p.Ser2518	Gallus et al 2010
	c.776A>G	p.Lys259Arg	Verrips et al 2000
	c.779G>A	p.Trp260Ter	Verrips et al 2000
	c.808C>T	p.Arg270Ter	Ahmed et al 1997
	c.819delT	p.Leu272fsTer	Leitersdorf et al 1993
	c.844+1G>A	p.del exon 4	Verrips et al 1997
	c.845-1G>A		Leitersdorf et al 1993
	c.850A>T	p.Lys284Ter	Meiner et al 1994
	c.863delA	p.Glu288fs	Gallus et al 2010

	c.944_948del TGGCC	p.Leu314fsTer	Lamon-Fava et al 2002
	c.1016C>T	p.Thr339Met	Reshef et al 1994
	c.1017 G>C		Wallon et al 2010
	c.1061A>G	p.Asp354Gly	Verrips et al 2000a
	c.1146_1151delins	p.H382QfsTer26	Suh et al 2012
	c.1151C>T	p.Pro384Leu	Verrips et al 2000
	c.1180-1181del CT	p.frame shift>ptc	Lee et al 2001
	c.1183C>T	p.Arg395Cys	Cali et al 1991
	c.1183C>A	p.Arg395Ser	Chen et al 1998b
	c.1184G>A	p.Arg395His	Chen et al 1996, Chen et al 1998a
	c.1184+1 G>A		Garuti et al 1997
	c.1185-1G>T		Mak et al 2004
	c.1263+81_1596+?del		Garuti et al 1996, Garuti et al 1997
	c.1202C>G	p.Pro401Arg	Okuyama et al 1996
	c.1209C>G	p.Asn403Lys	Castelnovo et al 2003
	c.1213C>T	p.Arg405Trp	Verrips et al 2000
	c.1214G>A	p.Arg405Gln	Chen et al 1997
	c.1222G>T	p.Glu408Ter	Von Bahr et al 2005
	c.1238T>A	p.Val413Asp	Koyama et al 2012
	c.1263G>A		Garuti et al 1996
	c.1263+5G>T		Garuti et al 1997
	c.1264-1G>A		Garuti et al 1997
	c.1323C>T	p.Pro441Ser	Lee et al 2001
	c.1330-1333delTTCC	p.Ser443fsTer449	Schneider et al 2010
	c.1402C>T	p.Gln461Ter	Lee et al 2001
	c.1415G>C	p.Gly472Ala	Verrips et al 2000
	c.1420C>T	p.Arg474Trp	Kuriyama et al 1991
	c.1421G>A	p.Arg474Gln	Kuriyama et al 1991
	c.1435C>T	p.Arg479Cys	Cali et al 1991
	c.1435C>G	p.Arg479Gly	Guyant-Maréchal et al 2005
Benign	c.366A>C	p.Gly122Gly	Nakashima et al 1994
	c.438G>A	p.Pro146Pro	Lamon-Fava et al 2002
	c.852G>A	p.Lys284Lys	Leitersdorf et al 1993
	c.243G>A	p.Leu81Leu	NM_000784.3:c.243G>A NP_000775.1
	c.882G>A	p.Glu294Glu	NM_000784.3:c.882G>A NP_000775.1
	c.888A>G	p.Gln296Gln	NM_000784.3:c.888A>G

			NP_000775.1
	c.1374C>A	p.Pro458Pro	NM_000784.3:c.1374C>A NP_000775.1

1. Variant designation that does not conform to current naming conventions

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