Title: Bietti Crystalline Dystrophy *GeneReview* Table 3
Authors: Okialda KA, Stover NB, Weleber RG, Kelly EJ
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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. *CYP4V2* Pathologic Allelic Variants

DNA Nucleotide Change (Alias [1])	Protein Amino Acid Change	Reference Sequences
c.367C>G	L22V	NM_207352.3 NP_997235.3
c.424T>A	W44R	
c.485G>A	G61S	
c.327+1G>A (IVS2+1G>A)	See footnote 5	
c.541G>T	E79D	
c.557C>T	R85C	
c.587G>A	G95R	
c.636T>C	I111T	
c.639T>G	L112X	
c.671A>G	M123V	
c.704G>T	G134X	
c.823T>G	L173W	
c.959T>C	Y219H	
c.1037G>A	W244X	
c.1062_1063	dupA	
c.1065A>G	H254R	
c.800-1_8delTCATACAG_800_808delGTCATCGCGinsGC [2] (IVS6-8 del/insGC)	See footnote 2	
c.1262C>T	R320X	
c.1275A>T	D324V	
c.1278C>T	T325I	
c.1296A>C	H331P	
c.1324G>A	W340X	
c.1325T>C	S341P	
c.1091-1A>G (ISV8-2A>G)	See footnote 4	
c.1461K>T	K386T	
c.1473G>A	R390H	
c.1491C>T	P396L	
c.1502C>T	R400C	
c.1503G>A	R400H	
c.1226-6delTGACAG_1226_1235delCAGGTTACAG [3] (IVS9-6del)	See footnote 3	
c.1652C>T	Q450X	
c.1749C>A	S482X	
c.1827G>A	R508H	
c.1830C>T	P509L	

See Quick Reference for an explanation of nomenclature. *GeneReviews* follows the standard naming conventions of the Human Genome Variation Society (www.hgvs.org).

1. Variant designation that does not conform to current naming conventions
2. Skipping of exon 7, which encodes 62 amino acids [Wada et al 2005]
3. Skipping of exon 10, which encodes 60 amino acids [Shan et al 2005]
4. Skipping of exon 9, which encodes 62 amino acids
5. Skipping of exon 2, resulting in a frameshift in which the exon 1 sequence is followed by four novel amino acids and a premature terminating [Li et al 2004].

References

Li A, Jiao X, Munier FL, Schorderet DF, Yao W, Iwata F, Hayakawa M, Kanai A, Shy Chen M, Alan Lewis R, Heckenlively J, Weleber RG, Traboulsi EI, Zhang Q, Xiao X, Kaiser-Kupfer M, Sergeev YV, Hejtmancik JF (2004) Bietti crystalline corneoretinal dystrophy is caused by mutations in the novel gene CYP4V2. Am J Hum Genet. 74:817-26.

Shan M, Dong B, Zhao X, Wang J, Li G, Yang Y, Li Y (2005) Novel mutations in the CYP4V2 gene associated with Bietti crystalline corneoretinal dystrophy. Mol Vis. 11:738-43.

Wada Y, Itabashi T, Sato H, Kawamura M, Tada A, Tamai M (2005) Screening for mutations in CYP4V2 gene in Japanese patients with Bietti's crystalline corneoretinal dystrophy. Am J Ophthalmol. 139:894-9.