

Title: Table 3. Mutations in *VCP* Identified in 39 Families, from IBMPFD *GeneReview*

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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. Mutations in *VCP* Identified in 39 Families

#	Amino Acid	Base Change (ORF)	<u>Exon</u>	Domain	Number of Families	References
1	R92H	284G>A	3	N terminus	1	Kaleem et al [2007]
2	R93C	277C>T	3	N terminus	4	Fanganiello et al [2011], Guyant-Marechal et al [2006], Hubbers et al [2007], Krause et al [2007]
3	R95G	283C>G	3	N terminus	2	Watts et al [2004]
4	R95C	283C>T	3	N terminus	1	Kimonis et al [2008]
5	P137L	410C>T	4	N terminus	1	Stojkovic et al [2009]
6	R155C	463C>T	5	N terminus	5	Gidaro et al [2008], Guyant-Marechal et al [2006], Hubbers et al [2007], Schroder et al [2005], Watts et al [2004]
7	R155H	464G>A	5	N terminus	8	Hubbers et al [2007], Watts et al [2004]
8	R155P	464G>C	5	N terminus	1	Watts et al [2004]
9	R155S	463C>A	5	N terminus	1	Vesa et al [2009]
10	R155L	N/A	5	N terminus	2	Kumar et al [2010], Stojkovic et al [2009]
11	G157R	469 G>C	5	N terminus	1	Djamshidian et al [2009]
12	R159H	476G>A	5	N terminus	2	Haubenberger et al [2005]
13	R159C	476G>A	5	N terminus	2	Bersano et al [2007], Spina et al [2008]
14	R191Q	572G>A	5	Linker 1	1	Johnson et al [2010], Spina et al [2008], Watts et al [2004]
15	L198W	593T>G	6	Linker 1	2	Watts et al [2007], Kumar et al [2010]
16	I206F	828A>T	6	Linker 1	1	[81]

#	Amino Acid	Base Change (ORF)	Exon	Domain	Number of Families	References
17	A232E	695C>A	6	Junction (L1-D1)	1	Watts et al [2004]
18	T262A	N/A	7	AAA D1	1	Spina et al [2008]
19	N387H	1159A>C	10	AAA D1	1	Watts et al [2007]
20	A439S	N/A	11	Linker 2	1	Stojkovic et al [2009]

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