Title: Caveolinopathies *GeneReview* Table 2
Authors: Bruno C, Sotgia F, Gazzerro E, Minetti C, Lisanti MP
Date: September 2012
Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 2. *CAV3* Allelic Variants Reported to Have No Noticeable Phenotypic Effect

#	Nt change	Exon	Protein	Phenotype	Reference
1	-122C>T	1	-	-	dbSNP
2	-37G>A	1	-	-	Leiden Muscular Dystrophy pages
3	27C>T	1	-	-	dbSNP, Leiden Muscular Dystrophy pages
4	99C>T	1	-	-	Hayashi et al [2004], Fee et al [2004], dbSNP, Leiden Muscular Dystrophy pages
5	114+26G>A	1	-	-	dbSNP, Leiden Muscular Dystrophy pages
6	114+99T>C	1	-	-	dbSNP
7	115-89G>T	2	-	-	dbSNP
8	115-45_115-29del	2	-	-	Leiden Muscular Dystrophy pages
9	c.115-23G>C	2	-	-	Leiden Muscular Dystrophy pages
10	c.123T>C	2	-	-	dbSNP, Leiden Muscular Dystrophy pages
11	c.137C>A	2	A46E	-	Leiden Muscular Dystrophy pages
12	166G>A	2	G56S	-	McNally et al [1998], de Paula et al [2001], Reijneveld et al [2006]
13	168C>A	2	-	-	Hayashi et al [2004]
14	171G>A	2	-	-	de Paula et al [2001]
15	204C>A	2	-	-	Hayashi et al [2004]
16	216C>G	2	C72W	-	de Paula et al [2001], Leiden Muscular Dystrophy pages
17	277G>A	2	A93T	-	Kubisch et al [2005]
18	377G>A	2	R126H	-	de Paula et al [2001]
11	*543T>C	2	-	-	dbSNP
12	*645A>T	2	-	-	dbSNP
13	*740C>G	2	-	-	dbSNP
14	*783A>G	2	-	-	dbSNP
15	*811C>G	2	-	-	dbSNP
16	*932T>A	2	-	-	dbSNP
17	*937C>A	2	-	-	dbSNP

Taken from http://www.dmd.nl/cav3_home.html

References

de Paula F, Vainzof M, Bernardino AL, McNally E, Kunkel LM, Zatz M. Mutations in the caveolin-3 gene: When are they pathogenic? Am J Med Genet. 2001;99:303-7.

Fee DB, So YT, Barraza C, Figueroa KP, Pulst SM. Phenotypic variability associated with Arg26Gln mutation in caveolin3. *Muscle Nerve.* 2004;30:375-8.

Hayashi T, Arimura T, Ueda K, Shibata H, Hohda S, Takahashi M, Hori H, Koga Y, Oka N, Imaizumi T, Yasunami M, Kimura A. Identification and functional analysis of a caveolin-3 mutation associated with familial hypertrophic cardiomyopathy. Biochem Biophys Res Commun. 2004;313:178-84.

Kubisch C, Ketelsen UP, Goebel I, Omran H. Autosomal recessive rippling muscle disease with homozygous CAV3 mutations. Ann Neurol. 2005;57:303-4.

McNally EM, de Sa Moreira E, Duggan DJ, Bonnemann CG, Lisanti MP, Lidov HG, Vainzof M, Passos-Bueno MR, Hoffman EP, Zatz M, Kunkel LM. Caveolin-3 in muscular dystrophy. Hum Mol Genet. 1998;7:871-7.

Reijneveld JC, Ginjaar IB, Frankhuizen WS, Notermans NC. CAV3 gene mutation analysis in patients with idiopathic hyper-CK-emia. Muscle Nerve. 2006;34:656-8.