Title: Caveolinopathies *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. *CAV3* Mutations and Associated Phenotypes

Nt Change	Exon	Protein	Position	Phenotype	Reference
77G>A	1	R26Q	N-terminal	Isolated HCK (s)	Carbone et al [2000]
„	„	„	„	RMD	Vorgerd et al [2001], Schara et al [2002]
„	„	„	„	LGMD-1	Figarella-Branger et al [2003]
„	„	„	„	DM	Tateyama et al [2002]
„	„	„	„	RMD + DM	Yabe et al [2002]
„	„	„	„	LGMD-1C + RMD	Fee et al [2004]
„	„	„	„	Isolated H-CK	Fulizio et al [2005]
81C>A	1	D27E	N-terminal	LGMD-1C + RMD + DM	Fisher et al [2003]
82C>A	1	P28T	N-terminal	RMD	Van der Bergh et al [2004]
83C>T	1	P28L	N-terminal	Isolated HCK (f)	Merlini et al [2002]
96C>G	1	N32K	N-terminal	LGMD-1C	Sugie et al [2004]
99C>G	1	N33K	N-terminal	DM+ HCK	Fulizio et al [2005]
IVS+2T>C	1	No protein	N-terminal	LGMD-1C	Muller et al [2006]
128T>A	2	V43E	N-terminal	LGMD-1C	Sugie et al [2004]
133G>A	2	A45T	N-terminal	LGMD-1C	Herrmann et al [2000]
“	“	“	“	RMD	Betz et al [2001]
“	“	“	“	LGMD-1C+ HCK	Fulizio et al [2005]
134C>T	2	A45V	N-terminal	RMD	Betz et al [2001]
136G>A	2	E46K	N-terminal	RMD	Ricardo et al [2005]
154A>G	2	S52G	Scaffolding domain	RMD	Dotti et al [2006]
169G>A	2	V57M	Scaffolding domain	Isolated HCK (f)	Alias et al [2004]
183C>A	2	S61R	Scaffolding domain	LGMD-1C+ HCK	Fulizio et al [2005]
187A>C	2	T63P	Scaffolding domain	LGMD-1C	Matsuda et al [2001]
188C>G	2	T63S	Scaffolding domain	HCM	Hayashi et al [2004]
186-194del	2	TFT63-65del	Scaffolding domain	LGMD-1C	Minetti et al [1998]

Nt Change	Exon	Protein	Position	Phenotype	Reference
233C>T	2	T78M	Membrane-spanning domain	LQTS	Reijneveld et al [2006], Vatta et al [2006]
257T>C	2	L85P*	Membrane-spanning domain	RMD	Kubish et al [2003]
277G>A	2	A92T*	Membrane-spanning domain	RMD + LGMD-1C	Kubish et al [2003]
290T>G		F96C	Membrane-spanning domain	LQTS	Vatta et al [2006]
290-293del	2	F96del	Membrane-spanning domain	LGMD-1C + RMD + HCK	Cagliani et al [2003]
314C>T	2	P104L	Membrane-spanning domain	LGMD-1C	Minetti et al [1998]
423C>G	2	S140R	C-terminal domain	LQTS	Vatta et al [2006]

Taken from http://www.hgmd.cf.ac.uk/ac/index.php

HCK, hyperCKemias (s) sporadic, (f) familiar

RMD, rippling muscle disease

LGMD, limb-girdle muscular dystrophy

DM, distal myopathy

HCM, hypertrophic cardiomyopathy

LQTS, long QT syndrome

*, homozygous mutation

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