

## Article

### Arrhythmogenic Right Ventricular Cardiomyopathy Type 5

### Is a Fully Penetrant, Lethal Arrhythmic Disorder

### Caused by a Missense Mutation in the *TMEM43* Gene

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**Table S1. Sequencing Variants Identified through Mutation Screening**

Locus	Accession Number	Primer Exon Name	Variation Name		Amino Acid Change	Genomic Number of Variant	Samples	BB04-66		WP05-218		CH04-81		GT04-141		GP05-219		MH05-244		MS06-114	
								Family	AR1		AR15		AR1		AR8		AR15		AR1		AR2
			Clinical Status	Affected			Affected		Affected		Affected		Unaffected		Unaffected		Unaffected				
			Chromosome	1			2	1	2	1	2	1	2	1	2	1	2	1	2	1	2
IQSEC1	NM_014869	14c	3'UTR A>G	2892+1060 A>G		12914829		A	A	nd	nd	A	A	A	A	A	A	nd	nd	A	G
IQSEC1	NM_014869	14c	3'UTR C>T	2892+1343 C>T		12914546		T	T	T	T	T	T	T	T	T	T	T	T	T	T
IQSEC1	NM_014869	9	IVS9+60 T>C	2358+60 T>C		12929868		C	C	nd	nd	C	C	C	C	C	C	C	C	C	C
IQSEC1	NM_014869	8	IVS8+73C>T	2232+73 C>T		12931531		T	T	T	C	T	T	T	T	C	T	C	T	C	T
Marker	<i>D3S3610</i>			Start 12980656				246	246	256	242	nd	nd	246	242	256	242	246	242	nd	nd
IQSEC1	NM_014869	1	IVS1+73C>G	65+73 C>G		12983814		nd	nd	nd	nd	C	C	C	G	C	C	C	C	C	C
Marker	<i>D3S2403</i>			Start 13147397				251	279	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
NUP210	NM_024923	40d	3'UTR T>C	5664+1195T>C		13332986		C	C	C	T	C	C	C	T	C	T	C	T	C	T
NUP210	NM_024923	40c	3'UTR G>A	5664+1051G>A		13333130		A	A	A	G	A	A	A	G	A	G	A	G	A	G
NUP210	NM_024923	40c	3'UTR A>C	5664+1010 A>C		13333171		C	C	C	A	C	C	C	A	C	A	C	A	C	A
NUP210	NM_024923	40c	3'UTR C>T	5664+977 C>T		13333204		T	T	T	C	T	T	T	C	T	C	T	C	T	C
NUP210	NM_024923	40b	3'UTR A>T	5664+365 A>T		13333816		A	A	A	A	A	A	A	T	A	A	A	A	A	A
NUP210	NM_024923	38&39	IVS39+59 G>A	5563+59 G>A		13335513		A	A	A	G	A	A	A	G	A	G	A	G	A	G
NUP210	NM_024923	38&39	IVS39+9 C>G	5563+9 C>G		13335563		C	G	C	G	C	G	C	G	C	C	C	C	G	C
NUP210	NM_024923	37	IVS37+25 G>A	5383+25 G>A		13336238		A	A	A	G	A	A	A	G	A	G	A	G	A	G
NUP210	NM_024923	37	5339 G>A	5339 G>A	V>M	13336287		A	A	A	G	A	A	A	G	A	G	A	G	A	G

NUP210	NM_024923	37	5225 T>C	5225 T>C	L>S	13336391		C	C	C	C	C	C	C	C	C	C	C	T	C	T
NUP210	NM_024923	34	4701 C>A	4701 C>A	T>T	13339876		A	A	A	G	A	A	A	G	A	G	A	G	A	G
NUP210	NM_024923	33	4582 C>T	4582 C>T	R>W	13342357		C	C	C	C	C	C	C	T	C	C	C	C	C	C
NUP210	NM_024923	33	4533 G>A	4533 G>A	S>S	13342406		A	A	A	G	A	A	A	G	A	G	A	G	A	G
NUP210	NM_024923	32	4332C>T	4332C>T	C>C	13343892		C	C	C	T	C	C	C	T	C	T	C	T	C	T
NUP210	NM_024923	26	3489 G>A	3489 G>A	E>E	13354400		G	G	G	A	G	G	G	A	G	A	G	A	G	A
NUP210	NM_024923	26	IVS25 -35 C>T	3472 -35 C>T		13354452		C	C	C	C	C	C	C	T	C	T	C	T	C	C
NUP210	NM_024923	22&23	3048 T>C	3048 T>C	F>F	13358540		T	T	T	C	T	T	T	C	T	C	T	C	T	C
NUP210	NM_024923	22&23	IVS21 -26 C>T	2965 -26 C>T		13358649		C	C	C	C	C	C	C	C	C	T	C	T	C	C
NUP210	NM_024923	21	IVS21 +143 C>T	2964 +143 C>T		13359532		C	C	C	T	C	C	C	C	C	C	C	C	C	C
NUP210	NM_024923	17	2461 C>G	2461 C>G	P>A	13370475		C	C	C	C	C	C	C	G	C	G	C	G	C	C
NUP210	NM_024923	17	2357 G>T	2357 G>T	R>L	13370579		G	G	G	T	G	G	G	T	G	T	G	T	G	T
NUP210	NM_024923	16	2264 C>T	2264 C>T	A>V	13374786		C	C	C	T	C	C	C	C	C	C	C	C	T	C
NUP210	NM_024923	16	IVS15 -53 A>G	2155 -53 A>G		13374948		A	A	A	G	A	A	A	A	A	A	A	A	G	A
NUP210	NM_024923	14	1822 A>G	1822 A>G	I>V	13382556		A	A	A	G	A	A	A	G	A	G	A	G	A	G
NUP210	NM_024923	9	IVS8 -64 ins	1046 -64 ins		13394126		wt	wt	wt	ins	wt	wt	wt	wt	wt	wt	wt	wt	ins	wt
NUP210	NM_024923	8	IVS8 +29 G>T	1045 +29 G>T		13395383		G	T	G	G	G	G	G	G	G	G	G	G	G	T
NUP210	NM_024923	7	IVS7 +39 G>A	976 +39 G>A		13396024		G	G	G	G	G	G	G	G	G	A	G	G	G	G
NUP210	NM_024923	7	889 G>A	889 G>A	A>T	13396150		G	G	G	A	G	G	G	A	G	A	G	A	G	A
NUP210	NM_024923	7	IVS6 -98 A>G	818 -98 A>G		13396320		A	A	A	G	A	A	A	G	A	G	A	G	A	G
NUP210	NM_024923	6	IVS5 -91 C>A	685 -91 C>A		13402998		C	C	C	A	C	C	C	C	C	C	C	C	A	C
NUP210	NM_024923	3	IVS3 +27 T>C	436 +27 T>C		13413830		T	T	T	C	T	T	T	C	T	C	T	C	T	C
NUP210	NM_024923	3	IVS2 -17 T>C	305 -17 T>C		13414005		T	T	T	C	T	T	T	T	T	T	T	T	T	T
NUP210	NM_024923	1	IVS1+130G>A	166 +130G>A		13436430		G	G	G	A	G	G	G	G	G	G	G	G	G	G
HDAC11	NM_024827	2	IVS2 +79 C>T	151 +79 C>T		13497973		C	C	C	C	C	T	C	C	C	C	C	C	T	C
HDAC11	NM_024827	3	IVS3 +24 G>T	252 +24 G>T		13500088		G	G	G	G	G	G	G	G	G	G	G	G	G	T
<b>HDAC11</b>	<b>NM_024827</b>	<b>4</b>	<b>IVS4 +19 insG</b>	<b>369 +19 insG</b>		<b>13513371</b>		ins	wt	ins	ins	ins	wt	ins	wt	wt	wt	wt	wt	wt	wt
HDAC11	NM_024827	7	IVS6 -97 C>T	490 -97 C>T		13518274		C	C	C	C	C	C	C	T	C	C	C	C	C	C
HDAC11	NM_024827	8	IVS7 -74 G>A	553 -74 G>A		13519310		G	G	G	G	G	G	G	A	G	G	G	G	G	G
FBLN2	DA856838	EST 4	del GT			13586337		del	del	del	wt	del	wt	del	wt	wt	wt	wt	wt	wt	wt
FBLN2	DA856838	EST 4	G>A			13586424		G	G	G	G	G	G	G	G	A	G	G	G	G	G
FBLN2	NM_001004019	Ex2C #2	del**			13587794		del	del	del	del	del	del	del	del	del	del	del	del	del	del
FBLN2	NM_001004019	Ex2C #2	1081 A>G			13587937		A	A	A	A	A	A	A	A	A	G	A	A	A	A

FBLN2	BC111426	EST1	G>A			13627967		G	G	G	A	G	A	G	G	G	G	G	G	G	G
FBLN2	BC111426	EST1	A>G			13628401		A	A	A	G	A	G	A	A	A	G	A	A	A	G
Marker	<i>D3S1516</i>			Start 13628628				347	359	351	359	347	351	347	335	351	343	351	363	nd	nd
FBLN2	NM_001004019	5	IVS5 +18 C>T			13630683		C	C	C	C	C	C	C	C	C	T	C	C	C	C
FBLN2	BF368851	EST 6	C>G			13637585		G	G	G	G	G	G	G	G	G	G	G	C	G	C
FBLN2	NM_001004019	11	IVS11 +68 T>G			13644538		G	G	G	G	G	G	G	G	G	G	G	T	G	T
FBLN2	NM_001004019	12	2646 A>G			13645482		G	G	G	G	G	G	G	G	G	A	G	A	G	A
FBLN2	NM_001004019	12	2701 A>G			13645537		G	G	G	G	G	G	G	G	G	A	G	A	G	A
FBLN2	NM_001004019	13	2826 T>C			13645777		C	C	C	C	C	C	C	C	C	C	C	T	C	T
FBLN2	NM_001004019	13	IVS13 +45 T>G	2842 +45 T>G		13645838		G	G	G	G	G	G	G	G	T	G	T	G	T	G
FBLN2	NM_001004019	15	IVS15 +77G>C	3085 +77 G>C		13647393		G	C	G	C	G	C	G	C	G	G	G	C	G	C
FBLN2	W70042	EST 2	A>T			13649190		T	A	T	A	T	A	T	A	A	A	T	A	A	A
FBLN2	NM_001004019	Ex 17	IVS17 +105 G>T	3338 +105 G>T		13653174		G	G	G	T	G	T	G	G	T	T	G	T*	G	T*
FBLN2	NM_001004019	18a	IVS17 -18 G>A	3339 -18 G>A		13654045		G	G	G	A	G	G	G	G	A	A	G	G	G	G
FBLN2	NM_001004019	18a	3480 G>A			13654204		G	G	G	A	G	G	G	G	A	A	G	G	G	G
FBLN2	NM_001004019	18a	3612 C>T			13654336		C	C	C	T	C	C	C	C	T	T	C	C	C	C
FBLN2	NM_001004019	18b-3'UTR	G>A			13654616		G	A	G	A	G	G	nd	nd	G	G	G	A	G	A
FBLN2	NM_001004019	18b-3'UTR	T>A			13654689		T	A	T	T	T	T	nd	nd	T	T	T	T	T	A
FBLN2	NM_001004019	18b-3'UTR	C>A			13654696		C	A	C	C	C	C	nd	nd	C	C	C	C	C	A?
FBLN2	NM_001004019	18b-3'UTR	C>A			13654709		C	A	C	C	C	A	nd	nd	C	C	C	C	C	A
FBLN2	NM_001004019	18b-3'UTR	C>T			13654830		C	T	C	T	C	T	nd	nd	T	T	C	C	C	T
FBLN2	NM_001004019	18b-3'UTR	del TA			13654852		wt	del	wt	wt	wt	del	nd	nd	wt	wt	wt	wt	wt	del
FBLN2	NM_001004019	18b-3'UTR	T>A			13654856		T	T	T	A	T	T	nd	nd	A	A	T	T	T	T
Marker	<i>D3S3608</i>			Start 13670236				165	165	165	165	165	175	165	165	165	165	165	165	nd	nd
WNT7A	NM_004625	4a#1	IVS3 -37 T>C	571 -37T>C		13835958		T	T	T	C	T	T	T	T	T	T	T	C	T	T
Marker	<i>D3S2385</i>			Start 13853945				146	142	nd	nd	nd	nd	146	146	nd	nd	nd	nd	nd	nd
WNT7A	NM_004625	3	459 T>C	459 T>C	S>S	13871141		C	T	C	C	C	T	C	T	C	T	T	T	T	T
WNT7A	NM_004625	3	315 G>A	315 G>A	A>A	13871285		A	G	A	A	A	G	A	G	A	G	G	G	G	G

WNT7A	NM_004625	2	IVS2 +37 C>A	298 +37 C>A		13891408		C	A	C	C	C	C	nd	nd	nd	nd	C	A	C	A
WNT7A	AA405144	EST1-Ex2	A>G			13892334		A	G	A	G	A	A	G	G	G	G	A	G	A	G
WNT7A	AA405144	EST1-Ex1	delG			13893611		wt	wt	wt	delG	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt
WNT7A	NM_004625	1b	5'UTR C>A	1 -164 C>A		13896478		C	C	C	A	C	C	C	C	C	C	C	C	C	C
Marker	D3S3602		Start 13900968					113	119	113	111	113	113	123	119	11	119	10	nd	nd	nd
Marker	D3S1585		Start 13916682					118	126	118	118	118	126	126	126	12	118	13	nd	nd	nd
TPRXL	AK092426	Ex2	IVS1 -9 ins/delTTT	ATTT microsatellite		14079871		8	6	8	9	8	8	6	6	6	8	6	8	7	7
TPRXL	AK092426	Ex2	IVS2 +81 A>G	5' UTR		14080090		A	G	A	A	A	A	G	G	G	G	G	G	nd	nd
CHCHD4	NM_144636	BC033775 Ex 3	3'UTR A>C			14129232		A	A	A	A	A	A	C	C	C	A	C	C	C	C
CHCHD4	NM_144636	BC033775 Ex 2	IVS3 +36 G>C	160 +36 G>C		14132891		G	G	G	G	G	G	G	G	G	C	G	G	G	G
CHCHD4	NM_144636	BC033775 Ex 2	IVS3 +13 T>C	160 +13 T>C		14132914		T	T	T	T	T	T	T	T	T	C	T	T	T	T
CHCHD4	NM_144636	BC033775 Ex 1	5'UTR A>G			14141281		G	G	G	G	G	G	G	G	G	G	G	A	G	G
TMEM43	NM_024334	4	IVS3 -106 C>T	298 -106 C>T		14147975		C	C	C	C	C	C	T	C	T	C	C	C	C	C
TMEM43	NM_024334	5&6	IVS5 +51 T>C	442 +51 T>C		14149147		T	T	T	T	T	T	nd	nd	T	T	C	C	C	C
TMEM43	NM_024334	7	IVS6 -90 G>A	513 -90 G>A		14150150		G	G	G	G	G	G	G	G	G	A	G	G	G	G
TMEM43	NM_024334	7	536 T>C		M>T	14150263		T	T	T	T	T	T	C	C	C	T	T	T	C	C
TMEM43	NM_024334	8&9	IVS8 +55 G>A	705 +55 G>A		14151447		G	G	G	G	G	G	A	A	A	G	G	G	A	A
TMEM43	NM_024334	10	IVS9 -56 G>A	781 -56 G>A		14152252		G	G	G	G	G	G	A	A	A	G	G	G	A	A
TMEM43	NM_024334	11	IVS10 -47 C>T	883 -47 C>T		14155634		C	C	C	C	C	C	C	C	C	C	T	C	T	T
<b>TMEM43</b>	<b>NM_024334</b>	<b>12a</b>	<b>1073 C&gt;T</b>		<b>S&gt;L</b>	<b>14158166</b>		T	C	T	C	T	C	C	C	C	C	C	C	C	C
<b>TMEM43</b>	<b>NM_024334</b>	<b>12a</b>	<b>3' UTR T&gt;C</b>	<b>c.1203 +115 T&gt;C</b>		<b>14158411</b>		T	T	T	T	T	T	C	C	C	C	C	C	C	C
TMEM43	NM_024334	12b	3' UTR C>T			14158573		C	C	C	C	C	T	C	C	C	C	C	C	C	C
TMEM43	NM_024334	12b	3' UTR A>G			14158759		A	A	A	A	A	A	A	A	A	A	G	A	G	G
TMEM43	NM_024334	12c	3' UTR A>C			14159075		A	A	A	A	A	A	A	A	A	A	C	A	C	C
TMEM43	NM_024334	12d	3' UTR T>C			14159670		T	T	T	T	T	T	T	T	T	T	C	T	C	C
TMEM43	NM_024334	12d	3' UTR T>C			14159720		T	T	T	T	T	T	T	T	T	T	C	T	C	C
TMEM43	NM_024334	12e	IVS12 +58	1203 +1942		14160264		12	11	12	11	12	11	12	11	13	14	11	13	12	14



SLC6A6	NM_003043	1	del 5'UTR	1 - 40823 del		14419313		del	del	del	del	del	del	del	del	del	del	del	del	del	del
SLC6A6	NM_003043	1	del 5'UTR	1 - 40813 del		14419323		del	del	del	del	del	del	del	del	del	del	del	del	del	del
SLC6A6	NM_003043	1	del 5'UTR	1 - 40805 del		14419331		del	del	del	del	del	del	del	del	del	del	del	del	del	del
SLC6A6	NM_003043	1	del 5'UTR	1 - 40802 del		14419334		del	del	del	del	del	del	del	del	del	del	del	del	del	del
<b>SLC6A6</b>	<b>NM_003043</b>	<b>2</b>	<b>G&gt;A 5'UTR</b>	<b>1 - 27420</b>		<b>14432716</b>		A	G	A	G	A	G	A	G	G	G	G	G	G	G
SLC6A6	BC038790	mRNA 1a	C>G 5'UTR			14435728		G	G	G	C	G	G	G	G	G	C	G	G	C	C
SLC6A6	BC038790	mRNA 1b	C>T 5'UTR			14435919		T	T	nd	nd	T	T	T	C	C	C	T	T	C	C
SLC6A6	BC111489	5	IVS5+46 G>T	599+46 G>T		14464374		G	G	G	G	G	T	G	T	G	G	T	G	T	T
<b>SLC6A6</b>	<b>BC111489</b>	<b>5</b>	<b>IVS5+370 A&gt;G</b>	<b>c.599+370 A&gt;G</b>		<b>14464698</b>		G	G	G	A	G	A	G	A	A	A	A	A	A	A
SLC6A6	AK023516-Ex1a	mRNA 2	IV6-1745 G>A	733-1745 G>A		14481283		A	A	A	G	A	A	A	A	G	G	A	G	A	A
SLC6A6	AK023516-Ex1a	mRNA 2	IV6-1541 T>C	733-1541 T>C		14481487		C	C	C	T	C	C	C	C	T	T	C	T	C	T
SLC6A6	AK023516-Ex1a	mRNA 2	IV6-1521 C>A	733-1521 C>A		14481507		A	A	A	C	A	A	A	A	C	C	A	C	A	C
SLC6A6	AK023516-Ex1a	mRNA 2	IV6-1392 G>A	733-1392 G>A		14481638		A	A	A	G	A	A	A	A	G	G	A	G	A	G
SLC6A6	AK023516-Ex1a	mRNA 2	IV6-1363 G>A	733-1363 G>A		14481665		A	A	A	G	A	A	A	A	G	G	A	G	A	G
<b>SLC6A6</b>	<b>AK023516-Ex1b</b>	<b>mRNA 2</b>	<b>IV6-1226 A&gt;G</b>	<b>733-1226 A&gt;G</b>		<b>14481802</b>		G	G	G	A	G	A	G	A	A	A	A	A	nd	nd
SLC6A6	AK023516-Ex1c	mRNA 2	IV6-280 del A	733-280 del A		14482800		del	wt	del	wt	del	wt	del	wt	del	wt	del	wt	nd	nd
SLC6A6	NM_003043	15a	3'UTR del			14501629		del	wt	del	wt	del	wt	del	wt	del	wt	del	wt	del	wt
SLC6A6	NM_003043	15b	3'UTR G>A			14502367		A	A	A	A	A	A	A	A	A	A	A	A	A	A
SLC6A6	NM_003043	15c	3'UTR C>G			14502940		C	C	C	G	C	C	C	C	C	C	C	C	C	C
SLC6A6	NM_003043	15c	3'UTR C>G			14502943		G	G	G	G	G	G	G	G	G	G	G	G	G	G
SLC6A6	NM_003043	15c	3'UTR T>C			14503008		C	C	C	C	C	T	C	C	C	C	C	C	C	C
SLC6A6	NM_003043	15c	3'UTR T>C			14503032		C	T	C	T	C	T	C	T	C	T	C	T	T	T
SLC6A6	NM_003043	15c	3'UTR del			14503092		wt	del	wt	del	wt	del	wt	del	wt	del	wt	del	wt	wt
SLC6A6	NM_003043	15d	3'UTR C>T			14503260		T	T	T	T	T	C	T	T	T	T	T	T	T	T
SLC6A6	NM_003043	15e	3'UTR G>T			14503788		G	G	G	G	G	T	G	G	G	G	G	G	G	G
SLC6A6	NM_003043	15g	3'UTR del			14505435		wt	del	wt	del	wt	del	wt	del	wt	del	wt	del	wt	del
SLC6A6	NM_003043	15h	3' UTR A>G			14505725		A	A	A	A	A	A	A	A	A	A	A	G	A	A
GRIP2	NM_001080423	23	IVS22 -5 del	2974 -5 del		14513077		wt	wt	wt	del	wt	wt	nd	nd	wt	wt	wt	wt	wt	wt
GRIP2	NM_001080423	22	IVS22+20A>G	2973+20A>G		14520058		A	A	A	A	A	A	A	A	A	G	A	A	A	A
GRIP2	NM_001080423	21	2808 C>T		T>T	14522186		C	C	C	C	C	C	C	T	C	C	C	C	C	C

GRIP2	NM_001080423	20	IVS20 +101 C>T C>T	2692 +101 C>T		14523212		C	C	C	T	C	C	C	C	C	nd	nd	nd	nd		
GRIP2	NM_001080423	20	IVS19 -6 T>C	2513 -6 T>C		14523498		T	T	T	C	T	T	T	T	T	T	C	nd	nd		
GRIP2	NM_001080423	18	2256 T>C		S>S	14526448		T	T	T	C	T	T	T	C	T	T	nd	nd	T	C	
GRIP2	NM_001080423	16	IVS15 -7 T>C	2015 -7 T>C		14527998		T	C	T	C	T	T	T	C	T	C	C	C	C	C	
GRIP2	NM_001080423	14	1890 A>G		A>A	14530220		A	A	A	G	A	A	A	G	A	A	nd	nd	A	G	
GRIP2	NM_001080423	14	IVS13 -49 A>G	1787 -49 A>G		14530371		A	A	A	G	A	A	nd	nd	nd	nd	nd	nd	A	G	
GRIP2	NM_001080423	13	IVS13 +42 ins AACT	1787 +42 ins AACT		14530762		wt	wt	wt	wt	wt	wt	wt	ins	wt	wt	nd	nd	wt	wt	
GRIP2	NM_001080423	13	1776 T>C		S>S	14530815		T	T	T	T	T	T	T	C	T	T	nd	nd	T	T	
GRIP2	NM_001080423	13	IVS12 -35 G>A	1601 -35 G>A		14531025		G	G	G	G	G	G	G	A	G	G	nd	nd	G	G	
GRIP2	NM_001080423	10	1309 ins			14536633		ins	ins	ins	ins	ins	ins	ins	ins	ins	ins	nd	nd	ins	ins	
GRIP2	NM_001080423	8	894 T>C		S>S	14538262		T	T	T	C	T	T	T	T	T	T	nd	nd	T	C	
Marker	D3S3595			Start 14617332				265	265	265	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd	
C3orf19	NM_016474	2	IVS2+122 T>A	147+122 T>A		14671163		nd	nd	nd	nd	T	A	nd	nd	nd	nd	nd	T	A	T	A
C3orf19	NM_016474	3	IVS3+45C>A	248+45 C>A		14672185		C	C	C	C	C	C	C	C	C	C	C	C	C	C	A
C3orf19	NM_016474	3	IVS3+182 A>G	248+182 A>G		14672322		G	G	G	G	G	G	G	G	G	G	G	G	A	G	A
C3orf19	NM_016474	5	IVS4-55C>A	308 -55C>A		14677986		A	A	A	A	nd	nd	A	A	A	A	nd	nd	A	A	A
C3orf19	NM_016474	5	IVS5+83C>G & T	485+83C>G & T		14678301		C	C	C	C	C	C	C	C	C	C	C	C	T	C	G
C3orf19	NM_016474	5	IVS5+190 delTTACAATA	485+190 delTTACAATA		14678409		del	del	del	del	del	del	del	del	del	del	del	del	del	del	del
C3orf19	NM_016474	6	IVS6+139 insTTGA	581+139 insGATT		14681773		wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	ins	ins	ins	ins
C3orf19	NM_016474	6	IVS6+140 A>G	581 +140 A>G		14681774		G	G	G	G	G	G	G	G	G	G	G	G	A	G	A
C3orf19	NM_016474	6	IVS6+141 insTTGG	581+141 insTTGG		14681775		wt	ins	wt	ins	wt	ins	wt	ins	ins	ins	ins	wt	wt	wt	wt
C3orf19	NM_016474	8	IVS7 -13 A>G	724 -13 A>G		14683922		G	G	G	G	G	G	G	G	G	G	G	G	G	G	G
C3orf19	NM_016474	8	IVS8 +33 G>C	819 +33 G>C		14684062		G	G	G	G	G	G	G	G	G	C	G	G	G	G	C
C3orf19	NM_016474	11C	3'UTR G>A	1404+825 G>A		14688530		A	A	A	A	A	A	A	A	A	A	A	A	A	A	A
C3orf20	NM_032137	2	5'UTR C>T	405-505 C>T		14698720		C	C	C	C	C	C	C	C	C	T	C	C	C	C	T
C3orf20	NM_032137	2	5'UTR A>G	405-498 A>G		14698728		A	A	A	A	A	A	A	G	A	A	A	A	A	A	A
C3orf20	NM_032137	2	5'UTR C>A	405-471 C>A		14698754		A	A	A	A	A	A	A	A	A	A	A	A	A	A	A
C3orf20	NM_032137	3	125 G>A	125G>A	G>D	14699349		G	G	G	G	G	G	G	G	G	G	G	G	G	G	A
C3orf20	NM_032137	3	193 G>A	193 G>A	D>N	14699417		G	G	G	G	G	G	G	G	G	G	G	G	A	G	A
C3orf20	NM_032137	7	892 G>A	892 G>A	A>T	14720861		A	A	A	G	A	G	A	A	A	G	A	G	A	A	A
C3orf20	NM_032137	8	1219 G>A	1219 A>G	I>V	14730576		A	G	A	G	A	G	A	A	A	G	A	G	A	A	A
C3orf20	NM_032137	8	1264 G>C	1264 C>G	L>V	14730621		G	G	G	C	G	C	G	G	G	C	G	C	G	C	G

C3orf20	NM_032137	Ex9	IVS8 -66 A>G	1314-66 A>G		14731734		G	G	G	G	G	G	G	G	G	G	G	G	G	G
C3orf20	NM_032137	11	IVS11 +3 A>G	1690+3A>G		14743538		A	A	A	G	A	G	A	A	A	G	A	G	A	A
C3orf20	NM_032137	11	IVS11 +23 G>A	1690+23G>A		14743558		A	A	A	G	A	G	A	A	A	G	A	G	A	A
C3orf20	NM_032137	13	2039 G>A	2039 G>A	R>H	14773980		G	A	G	G	G	A	G	G	G	G	G	G	G	G
C3orf20	NM_032137	14	2334 G>A	2334 G>A	V>V	14776491		G	A	G	G	G	A	G	G	G	G	G	G	G	G
C3orf20	NM_032137	14	IVS14 +45 G>A	2352+45 G>A		14776554		G	A	G	A	G	A	G	A	G	A	A	A	G	A
C3orf20	NM_032137	16	IVS16 +117G>C	2630+117G>C		14788839		C	C	C	G	C	C	C	C	C	G	C	C	C	G
<b>FGD5</b>	<b>NM_152536</b>	<b>1c</b>	<b>934 G&gt;A</b>	<b>934 G&gt;A</b>	<b>V&gt;M</b>	<b>14837239</b>		A	G	A	G	A	G	A	G	G	G	G	G	G	G
<b>FGD5</b>	<b>NM_152536</b>	<b>5&amp;6</b>	<b>IVS5 +22 G&gt;A</b>	<b>2186+22 G&gt;A</b>		<b>14914202</b>		G	A	G	A	G	A	G	A	A	A	A	A	A	A
<b>FGD5</b>	<b>NM_152536</b>	<b>5&amp;6</b>	<b>IVS5 -82 G&gt;A</b>	<b>2187 -82 G&gt;A</b>		<b>14914368</b>		G	A	G	A	G	A	G	A	A	A	A	A	A	A
<b>FGD5</b>	<b>NM_152536</b>	<b>5&amp;6</b>	<b>2220 G&gt;T</b>	<b>2220 G&gt;T</b>	<b>L&gt;L</b>	<b>14914483</b>		G	T	G	T	G	T	G	T	T	T	T	T	T	T
FGD5	NM_152536	8	IVS8 +64 T>C	2482+64 T>C		14917028		T	T	T	C	T	T	T	T	C	C	T	C	C	C
<b>FGD5</b>	<b>NM_152536</b>	<b>10</b>	<b>IVS10 +50 C&gt;T</b>	<b>2613+50 C&gt;T</b>		<b>14924272</b>		C	T	C	T	C	T	C	T	T	T	T	T	T	T
FGD5	NM_152536	15	3072 C>T	3072 C>T	H>H	14939047		C	T	C	C	C	T	C	C	C	C	C	C	C	C
<b>FGD5</b>	<b>NM_152536</b>	<b>16</b>	<b>IVS15 -74 G&gt;A</b>	<b>3085-74 G&gt;A</b>		<b>14939483</b>		A	G	A	G	A	G	A	G	G	G	G	G	G	G
FGD5	NM_152536	17	IVS16-49 C>T	3215-49 C>T		14940470		T	T	T	T	T	T	T	T	T	T	C	T	C	T
FGD5	NM_152536	17	IVS16-8 A>G	3215-8 A>G		14940511		G	G	G	G	G	G	G	G	G	G	G	G	G	G
<b>NR2C2</b>	<b>NM_003298</b>	<b>8</b>	<b>IVS8+70 G&gt;A</b>	<b>855+70 G&gt;A</b>		<b>15040789</b>		A	G	A	G	A	A	A	A	G	G	G	G	G	G
NR2C2	NM_003298	12	IVS11-34 C>T	1290-34 C>T		15051147		C	C	C	C	C	C	C	C	T	C	C	C	C	T
NR2C2	NM_003298	14c	1635 G>A	1635 G>A	Q/Q	15055700		G	G	G	G	G	G	G	A	G	G	G	G	G	A
NR2C2	NM_003298	15b	3'UTR A>G			15059873		A	A	A	A	A	A	A	A	A	G	A	A	A	A
<b>NR2C2</b>	<b>NM_003298</b>	<b>15b</b>	<b>3'UTR T&gt;A</b>			<b>15059884</b>		A	T	A	T	A	A	A	T	T	T	T	T	T	T
NR2C2	NM_003298	15e	3'UTR C>T			15061256		C	C	C	C	C	C	C	T	C	C	C	C	C	T
<b>NR2C2</b>	<b>NM_003298</b>	<b>15g</b>	<b>3'UTR insGATA</b>			<b>15062484</b>		ins	wt	ins	wt	ins	isn	ins	wt	wt	wt	ins	wt	wt	wt
NR2C2	NM_003298	15k	3'UTR C>G			15064207		C	C	C	C	C	G	C	C	C	C	C	C	C	C
NR2C2	NM_003298	15k	3'UTR G>A			15064452		G	G	G	G	G	G	G	G	G	G	G	G	G	A
MRPS25	NM_022497	4f	3' UTR A>G	522 +2704A>G		15066248		A	A	A	A	A	A	A	A	A	A	A	A	A	G
MRPS25	NM_022497	4f	3' UTR T>C	522 +2700T>C		15066252		C	T	C	T	C	C	C	T	T	T	C	T	T	T
MRPS25	NM_022497	4d	3' UTR T>C	522 +1875T>C		15067077		T	T	T	T	T	T	T	C	T	T	T	T	T	C
<b>MRPS25</b>	<b>NM_022497</b>	<b>4c</b>	<b>3' UTR G&gt;A</b>	<b>522+1059G&gt;A</b>		<b>15067893</b>		A	G	A	G	A	G	A	G	G	G	G	G	G	G
MRPS25	NM_022497	4a	3' UTR A>T	522+192A>T		15068761		A	A	A	A	A	A	A	T	A	A	A	A	A	A
MRPS25	NM_022497	2	IVS2 +75 C>T	241+75 C>T		15075805		nd	nd	C	T	C	T	C	T	C	C	C	T	C	C



ZFYVE2 0	NM_022340	14g	3'UTR T>C			15088127		C	T	C	T	C	C	C	C	T	T	C	T	C	T	
ZFYVE2 0	NM_022340	14g	3'UTR T>A			15088307		T	T	T	T	T	T	T	T	T	T	T	T	T	A	
ZFYVE2 0	NM_022340	14g	3'UTR G>A			15088523		A	G	A	G	A	A	A	A	G	G	A	G	A	G	
ZFYVE2 0	NM_022340	14e	3'UTR insA / AA ??			15089241		wt	wt	wt	wt	wt	ins	nd	nd	wt	wt	wt	wt	wt	ins	
ZFYVE2 0	NM_022340	14c	3'UTR C>A			15090274		A	C	A	C	A	A	A	A	C	C	A	C	A	C	
ZFYVE2 0	NM_022340	14b	1734 C>G		S>S	15090914		C	C	C	C	C	C	C	C	C	C	C	C	C	G	
ZFYVE2 0	NM_022340	6	IVS5 -55 delTC	290 -55 delTC		15102527		del	wt	del	wt	del	del	del	del	wt	wt	del	wt	del	wt	
CAPN7	NM_014296	1	5'UTR A>G			15222800		G	G	nd	nd	nd	nd	nd	nd	G	A	G	G	G	G	
CAPN7	NM_014296	6	IVS5-160A>G	639-160A>G		15239826		A	A	A	A	A	G	A	A	A	A	A	A	A	A	
<b>CAPN7</b>	<b>NM_014296</b>	<b>11</b>	<b>IVS11+68 C&gt;T</b>	<b>1286 +68 C&gt;T</b>		<b>15250552</b>		<b>T</b>	<b>C</b>	<b>T</b>	<b>C</b>	<b>T</b>	<b>C</b>	<b>T</b>	<b>C</b>	<b>C</b>	<b>C</b>	<b>C</b>	<b>C</b>	<b>C</b>	<b>C</b>	
CAPN7	NM_014296	12	IVS12+150 G>A	1407 +150G>A		15251806		G	A	G	A	G	A	G	A	G	A	G	A	G	A	
<b>CAPN7</b>	<b>NM_014296</b>	<b>13&amp;14</b>	<b>IVS12-28 T&gt;C</b>	<b>1408 -28 T&gt;C</b>		<b>15256956</b>		<b>C</b>	<b>T</b>	<b>C</b>	<b>T</b>	<b>C</b>	<b>T</b>	<b>C</b>	<b>T</b>	<b>T</b>	<b>T</b>	<b>T</b>	<b>T</b>	<b>T</b>	<b>nd</b>	<b>nd</b>
CAPN7	NM_014296	19	IVS18-78 del	2074 -78del		15263760		wt	del	wt	del	wt	del	wt	del	wt	del	wt	del	wt	del	wt
SH3BP5	NM_004844	9b	3'UTR del	1368+498 del		15272099		nd	nd	wt	del	nd	nd	wt	del	wt	del	wt	del	wt	del	wt
SH3BP5	NM_004844	9a	3'UTR G>A	1368+23 G>A		15272574		G	G	G	G	G	G	G	G	G	A	G	G	G	G	
SH3BP5	NM_004844	8	961 G>A		V>M	15273553		G	G	G	G	G	G	G	G	G	G	G	G	A	G	G
SH3BP5	NM_004844	6	IVS5-53 C>T	627-53 C>T		15276367		T	C	nd	nd	T	T	T	C	T	C	T	T	T	T	
SH3BP5	NM_004844	6	IVS5-79 G>C	627-79G>C		15276393		G	C	nd	nd	G	C	G	C	G	C	G	C	G	C	
SH3BP5	NM_004844	4	390 T>C		R>R	15286329		C	T	C	C	C	C	C	C	T	T	C	T	C	T	
SH3BP5	NM_004844	4	IVS3-80 C>A	331-80 C>A		15286468		A	A	A	A	A	A	A	A	A	A	A	A	A	A	
Marker	<i>D3S3613</i>			Start 15336926				193	197	193	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd	
SH3BP5	NM_001018009	1	5'UTR A>G	1-71626 A>G		15357874		A	A	A	A	A	A	A	A	A	A	A	A	G	A	A

Two hundred and forty sequencing variants were identified in the 20 *ARVD5* candidate genes. Nineteen variants were found exclusively in subjects who were clinically affected (bold). The *ARVD5* ancestral haplotype is in the left column of affected individuals. Markers used by Ahmad et al. (1998) are in italics. Note that the subjects screened were not the same subjects used to narrow the region (Figure 3).