

SUPPLEMENTAL MATERIAL

Supplemental Table S1. List of the genes selected to perform the custom array.

Gene Description (Functional classes)	Gene	trIDs*	Chr†	Segs (n)‡	Segs (bp) §	Transc (n) ¶
CARDIOMYOPATHY-ASSOCIATED						
actin, alpha 1, skeletal muscle	<i>ACTA1</i>	NM_001100	1	1	3852	1
cardiac muscle alpha actin 1 proprotein	<i>ACTC1</i>	NM_005159	15	2	8523	1
calreticulins	<i>CALR3</i>	NM_145046	19	7	8574	1
Ca-binding chaperons						
calsequestrin 2 (cardiac muscle)	<i>CASQ2</i>	NM_001232	1	11	13711	1
caveolin 3≠	<i>CAV3</i>	NM_001234,NM_033337	3	2	3423	2
cytochrome c oxidase assembly protein≠	<i>COX15</i>	NM_078470,NM_004376	10	8	15983	2
cysteine and glycine-rich protein 3≠	<i>CSRP3</i>	NM_003476,NM_001127656	11	6	7347	2
Desmin	<i>DES</i>	NM_001927	2	4	7255	1
frataxin≠	<i>FXN</i>	NM_000144,NM_181425	9	5	7275	2
junctophilin≠	<i>JPH2</i>	NM_020433,NM_175913	20	6	12735	2
lysosomal-associated membrane protein≠	<i>LAMP2</i>	NM_002294,NM_001122606,NM_013995	X	8	19044	3
LIM domain binding 3	<i>LDB3</i>	NM_001080115,NM_01080114,NM_001080116,NM_007078	10	13	21033	4
myosin binding protein C, cardiac	<i>MYBPC3</i>	NM_000256	11	7	19751	1
myosin, heavy chain 6, cardiac muscle, alpha	<i>MYH6</i>	NM_002471	14	8	24026	1
myosin, heavy chain 7, cardiac muscle, beta	<i>MYH7</i>	NM_000257	14	4	23206	1
slow cardiac myosin						
regulatory light chain 2	<i>MYL2</i>	NM_000432	12	5	6796	1
myosin, light chain 3, alkali; ventricular, skeletal, slow	<i>MYL3</i>	NM_000258	3	4	5307	1
myosin light chain	<i>MYLK2</i>	NM_033118	20	5	10877	1

kinase 2						
myosin VI	<i>MYO6</i>	NM_004999	6	28	40652	1
myozenin 2	<i>MYOZ2</i>	NM_016599	4	5	8124	1
NADH dehydrogenase (ubiquinone) flavoprotein 2	<i>NDUFV2</i>	NM_021074	18	7	7918	1
obscurin, cytoskeletal calmodulin≠	<i>OBSCN</i>	NM_052843,NM_0010 98623	1	23	82957	2
phospholamban	<i>PLN</i>	NM_002667	6	2	3716	1
AMP-activated protein kinase gamma2 subunit≠	<i>PRKAG2</i>	NM_024429,NM_0010 40633,NM_016203	7	16	20335	3
solute carrier family 25 sorcina≠	<i>SLC25A4</i> <i>SRI</i>	NM_001151 NM_198901,NM_0031 30	4 7	2 6	4750 10155	1 2
titin-cap (telethonin)	<i>TCAP</i>	NM_003673	17	1	2210	1
troponin C type 1	<i>TNNC1</i>	NM_003280	3	2	3474	1
troponin I, cardiac	<i>TNNI3</i>	NM_000363	19	3	5486	1
troponin T type 2, cardiac≠	<i>TNNT2</i>	NM_001001430,NM_0 00364,NM_001001431, NM_001001432 NM_001018005,NM_0 01018004,NM_0010180 08,NM_000366,NM_00 1018020,NM_00101800 7,NM_001018006 NM_003319,NM_1333 78,NM_133432,NM_13 3437,NM_133379	1	8	14014	4
tropomyosin 1 alpha chain≠	<i>TPM1</i>	NM_000366,NM_00 1018020,NM_00101800 7,NM_001018006 NM_003319,NM_1333 78,NM_133432,NM_13 3437,NM_133379	15	8	14192	7
titina≠	<i>TTN</i>	NM_003319,NM_1333 78,NM_133432,NM_13 3437,NM_133379	2	44	229584	5
vinculin isoform meta-VCL≠	<i>VCL</i>	NM_014000,NM_0033 73	10	18	25559	2
integrin, alpha 8	<i>ITGA8</i>	NM_003638	10	26	31822	1
cardiac ankyrin repeat protein	<i>CARP</i>	NM_014391	10	4	8118	1
v-raf-1 murine leukemia viral	<i>RAF1</i>	NM_002880	3	10	15534	1
oncogene homolog 1						

METABOLIC HCM

acid-alpha glucosidase	<i>GAA</i>	NM_000152	17	5	14287	1
amilo-1-6-glucosidase	<i>AGL</i>	NM_000642	1	19	31359	1
acid-beta glucosidase	<i>GBA</i>	NM_001005741	1	2	8923	1

ION CHANNELS

potassium voltage-gated channel,	<i>KCNQ1</i>	NM_181798,NM_0002 18	11	12	19308	2
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KQT-like≠						
potassium channel voltage-gated, subfamily H≠	<i>KCNH2</i>	NM_172056,NM_0002 38,NM_172057	7	7	15667	3
potassium channel voltage-gated ISK-related subfamily member 1≠	<i>KCNE1</i>	NM_000219,NM_0011 27670,NM_001127669, NM_001127668	21	4	8545	4
potassium channel voltage-gated ISK-related subfamily member 2	<i>KCNE2</i>	NM_172201	21	2	2805	1
sodium channel, voltage-gated, type V, alpha subunit≠	<i>SCN5A</i>	NM_198056,NM_0010 99405,NM_001099404, NM_000335 NM_001129830,NM_0 01129827,NM_0011298 29,NM_000719,NM_00 1129831,NM_00112983 9,NM_001129836,NM_ 001129833,NM_001129 832,NM_001129834,N M_001129835,NM_001 129837,NM_001129838 ,NM_001129840,NM_0 01129841,NM_0011298 42,NM_001129843,NM _001129844,NM_00112 9846,NM_199460	3	23	35206	4
calcium channel, voltage-dependent, L-type, alpha-1C subunit ≠	<i>CACNA1C</i>		12	37	58658	20
sodium channel, voltage-gated, type IV, beta subunit	<i>SCN4B</i>	NM_174934	11	4	9483	1
potassium channel, inwardly rectifying, subfamily J, member2	<i>KCNJ2</i>	NM_000891	17	2	7388	1
MEMBRANE CHANNELS						
solute carrier family 9, isoform A3	<i>SLC9A3</i>	NM_004174	5	8	14281	1
solute carrier family 9, isoform A2	<i>SLC9A2</i>	NM_003048	2	10	16646	1
solute carrier family 9, isoform A4	<i>SLC9A4</i>	NM_001011552	2	11	15714	1
ATPase Ca(2+)-transporting,	<i>ATP2A2</i>	NM_001681,NM_1706 65	12	10	23130	2

slow-twitch≠

solute carrier family 6, member 4	<i>SLC6A4</i> (<i>SLC6A4</i>)	NM_001045	17	9	15765	1
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GROWTH FACTORS

transforming growth factor, beta 1	<i>TGF-β</i> (<i>TGFB1</i>)	NM_000660	19	5	8407	1
fibroblast growth factor 1 (acidic) ≠	<i>FGFa</i> (<i>FGF1</i>)	NM_000800,NM_0331 37,NM_033136	5	4	6346	3
insulin-like growth factor 1≠	<i>IGF1</i>	NM_001111285,NM_0 01111284,NM_0011112 83,NM_000618	12	6	13820	4
early growth response 1	<i>EGR1</i>	NM_001964	5	1	4825	1
insulin like growth factor 2≠	<i>IGF2</i>	NM_001127598,NM_0 00612,NM_001007139	11	6	13161	3

FACTORS INVOLVED IN CARDIOMYOCYTE GROWTH AND CONTRACTILITY

beta1 adrenergic receptor 1	<i>ADRB1</i>	NM_000684	10	1	3863	1
angiotensin receptor 1≠	<i>AGTR1</i>	NM_004835,NM_0006 85,NM_032049,NM_00 9585,NM_031850	3	5	7576	5
endothelin 1	<i>EDN1</i>	NM_001955	6	4	6268	1
endothelin receptor, type A	<i>EDNRA</i>	NM_001957	4	7	11745	1
angiotensinogen	<i>AGT</i>	NM_000029	1	4	7451	1
endothelin receptor. Type B≠	<i>EDNRB</i>	NM_000115,NM_0039 91,NM_001122659	13	5	10584	3
Protein kinase C, alpha	<i>PRKCA</i>	NM_002737	17	15	25199	1

* trIDs: transcripts identification

†chr: chromosome

‡Segs (n): number of segments

§Segs (bp): length (bp) of the screened gene regions

¶Transc (n): number of transcripts captured

≠: more than one transcript was captured

Supplemental Videos legends:

Supplemental Video 1

Parasternal long axis view of III4 by echocardiography: enlarged LA, normal LV size and wall thickness, and mild pericardial effusion. Papillary muscle is easy to be muddled with posterior wall.

Supplemental Video 2

LV short axis view with mitral valve section of III4 by echocardiography: normal LV size and wall thickness.

Supplemental Video 3

LV short axis with papillary muscle section of III4 by echocardiography: normal LV size and wall thickness. Papillary muscle is easy to be muddled with posterior wall.

Supplemental Video 4

Reversed apical four-chamber view of III4 by echocardiography: significant biatrial enlargement, normal biventricular size. Papillary muscle can be differentiated from ventricular wall. Foggy blood flow from LA to LV indicates slow flow in cardiac diastolic phase.

Supplemental Video 5

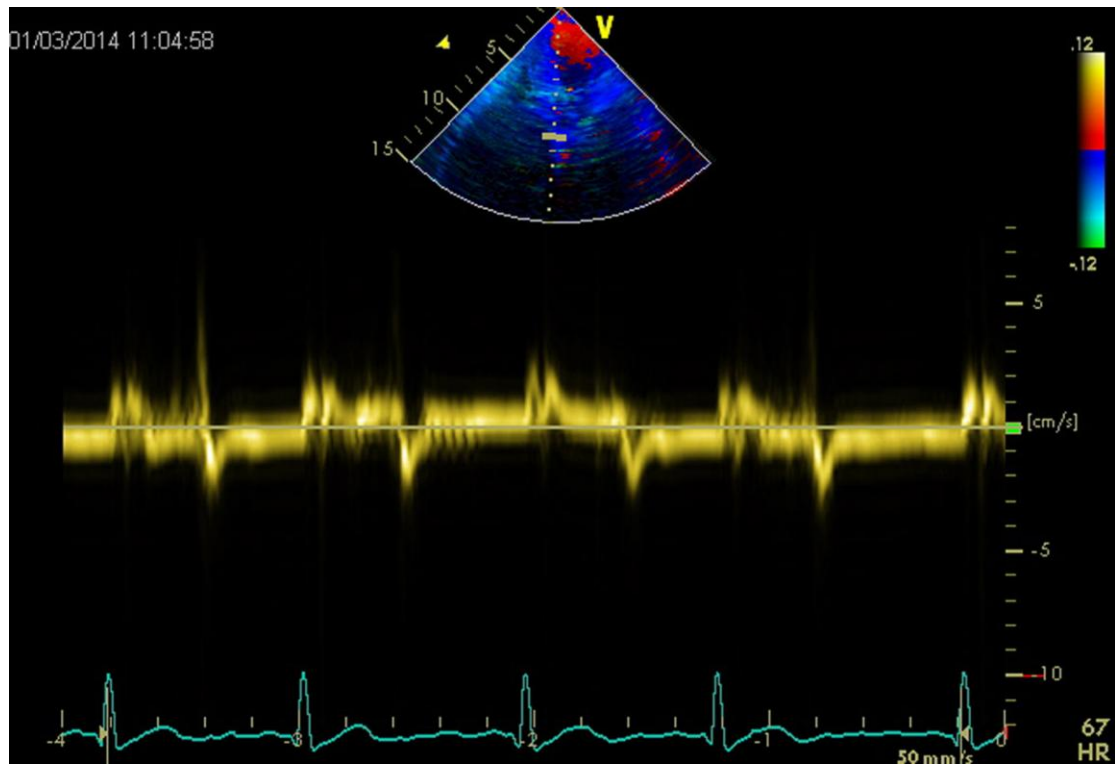
Animated movie of cardiac magnetic resonance of III6: enlargement of LA, normal LV size and wall thickness.

Supplemental Video 6

Long axis view of patient 4 by echocardiography: enlarged LA, normal LV size and wall thickness.

Supplemental Figure and legend:

Supplemental Figure S1



Supplemental Figure S1

Echocardiography of III4 shows reduced septal mitral annulus velocity (e') by Tissue Doppler Image.