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ORIGINAL ARTICLE

Novel genotype–phenotype associations demonstrated by high-throughput sequencing in patients with hypertrophic cardiomyopathy

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ABSTRACT

Objective A predictable relation between genotype and disease expression is needed in order to use genetic testing for clinical decision-making in hypertrophic cardiomyopathy (HCM). The primary aims of this study were to examine the phenotypes associated with sarcomere protein (SP) gene mutations and test the hypothesis that variation in non-sarcomere genes modifies the phenotype.

Methods Unrelated and consecutive patients were clinically evaluated and prospectively followed in a specialist clinic. High-throughput sequencing was used to analyse 41 genes implicated in inherited cardiac conditions. Variants in SP and non-SP genes were tested for associations with phenotype and survival.

Results 874 patients (49.6±15.4 years, 67.8% men) were studied; likely disease-causing SP gene variants were detected in 383 (43.8%). Patients with SP variants were characterised by younger age and higher prevalence of family history of HCM, family history of sudden cardiac death, asymmetric septal hypertrophy, greater maximum LV wall thickness (all p values<0.0005) and an increased incidence of cardiovascular death (p=0.012). Similar associations were observed for individual SP genes. Patients with *ANK2* variants had greater maximum wall thickness (p=0.0005). Associations at a lower level of significance were demonstrated with variation in other non-SP genes.

Conclusions Patients with HCM caused by rare SP variants differ with respect to age at presentation, family history of the disease, morphology and survival from patients without SP variants. Novel associations for SP genes are reported and, for the first time, we demonstrate possible influence of variation in non-SP genes associated with other forms of cardiomyopathy and arrhythmia syndromes on the clinical phenotype of HCM.

INTRODUCTION

Hypertrophic cardiomyopathy (HCM) is a common autosomal dominant genetic trait associated with sudden cardiac death (SCD) and progressive heart failure.[1 2] Patients are routinely offered genetic testing in order to provide them with information about the likely impact of disease on their lives and facilitate lifestyle and medical interventions that improve prognosis.[2 3] However, for this strategy to succeed, there must be a predictable relation between specific genotypes and disease expression.

In around 50% of cases, HCM is caused by mutations in genes coding for sarcomere or sarcomere-related genes.[4] So far, the most commonly reported genotype–phenotype associations are those that relate to the presence or absence of sarcomere protein (SP) gene mutations rather than mutations in specific genes.[5 6] A number of studies have suggested that some mutations are associated with reduced survival, but these findings are inconsistent and fail to account for the often dramatic variation in clinical phenotypes seen in individuals with the same genetic variant.[w1–w7 7 w8 8 w9 9 w10 w11 10 w12 11 w13 w14 12 13 w15–w18 14 w19 15 16]

Several studies have examined the role of common genetic variation on the expression of sarcomere mutations using genome-wide association studies or a candidate gene approach, but most have failed to show any major effect on disease expression.[w20–w22 17 18] HCM cases (as well as controls) also carry rare variants in genes coding for desmosomal, ion-channel and other proteins implicated in inherited heart disease[19] but their relevance to disease expression is unknown.

The hypothesis of this study is that rare variants in sarcomere genes and also in non-sarcomere genes implicated in other forms of inherited cardiac disorders (for which sequence data are available in our study) modify the clinical characteristics and severity of HCM.

METHODS

Study population and design, clinical evaluation and sample collection

The study was approved by the University College London (UCL)/UCL Hospitals (UCLH) Joint Research Ethics Committee. Before enrolment, all patients provided written informed consent and received genetic counselling in accordance with international guidelines.[3]

An observational, retrospective, longitudinal cohort study design was used. The study population comprised unrelated and consecutively evaluated patients with HCM referred to the Inherited Cardiovascular Disease Unit at The Heart Hospital, UCLH, London, UK. Clinical evaluation included a personal and family history, physical examination,

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12 lead ECG, echocardiography, symptom limited upright exercise testing with simultaneous respiratory gas analysis (cardiopulmonary exercise test) and ambulatory ECG monitoring as previously described.[20] HCM was diagnosed in probands when the maximum left ventricular (LV) wall thickness (MLVWT) on 2D echocardiography measured 15 mm or more in at least one myocardial segment or when MLVWT exceeded 2 SDs corrected for age, size and gender in the absence of other diseases that could explain the hypertrophy.[21] In individuals with unequivocal disease in a first degree relative, diagnosis was made using extended familial criteria for HCM.[22] Ethnicity was self-reported and classified using a modified National Health Service ethnic categorisation. Patients were evaluated every 6–12 months or earlier if there was a clinical event. Initial evaluation and follow-up data were collected prospectively and registered in a relational database. The definitions of severe LV hypertrophy, family history of SCD, syncope, non-sustained ventricular tachycardia (NSVT) and abnormal blood pressure response were as previously described.[23]

Targeted gene enrichment and high-throughput sequencing

Blood samples were collected at initial evaluation and DNA was isolated from peripheral blood lymphocytes using standard methods. The sequencing methodology has been reported in detail previously.[19] In summary, the protocol was designed to screen 2.1 Mbp of genomic DNA sequence per patient, covering coding, intronic and selected regulatory regions of 20 genes known to be associated with HCM and dilated cardiomyopathy (*MYH7, MYBPC3, TNNT2, TNNI3, MYL2, MYL3, ACTC1, TPM1, TNNC1, MYH6, CSRP3, DES, TCAP, PDLIM3, PLN, LDB3, LMNA, VCL, RBM20* and *TTN*), 10 genes implicated in arrhythmia syndromes/ion-channel disease (*RYR2, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, ANK2, CASQ2, CAV3* and *KCNJ2*), seven genes associated with arrhythmogenic right ventricular cardiomyopathy (*PKP2, DSC2, DSG2, JUP, DSP, TMEM43* and *TGFß3*) and a further four candidate genes (*GJA1, PLEC, PNN* and *PKP4*) which were not analysed in this work.[19] Analysis of titin (*TTN*) variants and their effect on phenotype is ongoing and will be reported in a separate paper.

Bioinformatic analysis

Paired-end reads were aligned using Novoalign software V.2.7.19 on the human reference genome build hg19. Duplicate reads were flagged using the Picard MarkDuplicate tool. Our calling strategy followed closely the Genome Analysis Toolkit (GATK) best practices as of January 2014. Briefly, following BAM file compression using the GATK ReduceReads module,[24] multisample calling was performed on all probands jointly with a set of 1492 unrelated whole exomes (UCL-exome consortium) using the GATK Unified Genotyper.[24] After GATK variant recalibration (separately for SNPs and indels), calls were annotated using the ANNOVAR software (with the Ensembl gene definitions).[w23] For all association tests, we filtered variants for the GATK recalibration PASS filter.

Candidate variants for further analysis were defined using frequency and predicted functional effect. For the functional filter, exonic non-synonymous, loss-of-function and splice-site variants were included. Sequence data were filtered using a minor allele frequency threshold of ≤0.2% based on the NHLBI exome variant server data (computed through the ANNOVAR annotations). To provide a more accurate estimate of variant frequency in controls that is not affected by potential differences in calling strategy in the NHLBI dataset, we randomly selected 25% of the 1492 UCL-exome samples as an 'external control set' and removed variants that appeared more than twice in these 372 'external controls'. These samples were only used to define a variant frequency and not included in the subsequent association test, to avoid a previously noted statistical issue, where variant frequency is defined in the same set that is used for case control testing.[w24] Variants present in the dbSNP build 137 database[w25] and published in the literature were identified. In silico prediction of pathogenicity for novel missense variants was performed using Polyphen2, SIFT and Condel.[w26 w27 25] A variant was predicted to be pathogenic if classified as 'damaging' by SIFT and simultaneously 'possibly' or 'probably damaging' by Polyphen2, or if predicted to be damaging by Condel.

Summary statistics for genotype–phenotype associations

R (V.3.0.0) and SPSS (V.22.0.0.0, IBM Corp.) were used for the analyses. Clinical phenotype data are presented as frequency (and percentage) for non-continuous variables and mean±SD or median and IQR for continuous variables where appropriate. Normally distributed continuous variables were compared using unpaired two-tailed Student's t test. Multiple groups were compared using analysis of variance. Categorical variables were compared using χ^2 or Fisher exact tests. When appropriate, non-parametric tests were used.

Group comparisons were made for the prevalence and severity of each phenotypic trait (at baseline and final follow-up) in patients with and without a rare variant in one or more of the eight most common SP genes (*MYH7, MYBPC3, TNNI3, TNNT2, MYL2, MYL3, ACTC1* and *TPM1*). We also compared the prevalence and severity of each phenotypic trait in patients carrying only one versus more than one variant in SP genes. The same comparisons were made for the presence and absence of rare variation in non-SP genes in the whole cohort and in the subgroup of individuals with a disease-causing SP gene mutation.

Multiple testing correction strategy

For each trait of interest we tested the effect of variants in eight SP genes and 28 non-SP genes. Therefore, a nominal p value of 0.05 was not appropriate. In addition, the Bonferroni correction for the number of phenotypes multiplied by the number of genes is too stringent because it tests the global null of no association between any pair of gene/trait. We therefore took an intermediate approach, correcting the analysis of each phenotype for the number of gene tests. For SP genes, we performed nine tests (one per SP gene, plus an additional test for all SP mutations combined). Therefore, we used $p<0.0056$, which is 0.05/9. For non-SP genes, we corrected for 28 tests, which translates into $p<0.0018=0.05/28$. Data on associations that did not fulfil these thresholds but met a nominal p value of <0.05 are presented in the Results section and online supplementary files.

Survival analysis

Definition of endpoints in the survival analyses was as previously described.[23] Survival from cardiovascular death (a composite of SCD and death from heart failure or stroke) and SCD or equivalent (appropriate implantable cardioverter-defibrillator (ICD) shock) was modelled using Kaplan–Meier analysis and log-rank test from the first clinical evaluation at The Heart Hospital and from birth.

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Table 1 Demographic and clinical characteristics of the study cohort

	Frequency (percentage) or mean±SD (range) or median (IQR)
Demographics	
Age at initial evaluation (years)	49.6±15.4 (6–87)
Male	590/874 (67.8%)
Ethnicity	
Caucasian	622 (71.2%)
Indian and other Asian	68 (7.8%)
African/Caribbean	39 (4.5%)
Chinese	6 (0.7%)
Other	20 (2.3%)
Not reported	119 (13.6%)
Presentation	
Family history of HCM	226/853 (26.5%)
Family history of SCD	182/872 (20.9%)
NYHA class III or IV	100/850 (11.8%)
Syncope	140/856 (16.4%)
Chest pain	205/854 (24.0%)
Initial ECG	
Atrial fibrillation	43/874 (4.9%)
PR interval (ms)	174.8±32.4 (108–320)
QRS duration (ms)	101.0±25.5 (64–238)
Initial CPEX	
SBP rest (mm Hg)	128.8±21.0 (80–210)
SBP response to exercise (mmHg)	48.5±24.2 (-5–150)
Abnormal SBP response to exercise	92/662 (13.9%)
Initial echocardiography	
Maximal LV wall thickness (mm)	18.5±4.4 (9–38)
Severe LVH (≥30 mm)	17/601 (2.8%)
Right ventricular hypertrophy (>5 mm)	184/864 (21.3%)
Asymmetric septal hypertrophy pattern	643/850 (75.6%)
Left atrial diameter (mm)	44.0±7.5 (18–90)
LV end-diastolic diameter (mm)	45.9±5.9 (29–65)
LV dilatation (>55 mm)	38/851 (4.5%)
LV end-systolic diameter (mm)	28.5±5.6 (9–50)
Fractional shortening (%)	38.3±8.2 (16–70)
Systolic dysfunction (≤25% FS)	29/829 (3.5%)
E wave deceleration time (ms)	221.0 (184–268)
Mitral regurgitation—moderate/severe	163/851 (19.2%)
Peak LVOT gradient (mm Hg)	12.0 (4.0–60.0)
LVOT gradient >30 mm Hg	328/812 (40.4%)
NSVT—Holter	127/566 (22.4%)
Follow-up	
New-onset atrial fibrillation	216/874 (24.7%)
Implantable cardioverter-defibrillator	177/874 (20.3%)
Myectomy	130/874 (14.9%)
Alcohol septal ablation	46/874 (5.3%)
Myectomy and/or alcohol septal ablation and/or pacemaker implantation for LVOT gradient reduction	182/874 (20.8%)
Cardiovascular death	25/874 (2.9%)
SCD	16/874 (1.8%)

Total N=874.
CPEX, cardiopulmonary exercise test; FS, fractional shortening; HCM, hypertrophic cardiomyopathy; LVH, LV hypertrophy; LVOT, LV outflow tract; NSVT, non-sustained ventricular tachycardia; NYHA, New York Heart Association; SBP, systolic blood pressure; SCD, sudden cardiac death.

RESULTS

Study population

In all, 874 unrelated and consecutive patients with HCM were studied. Mean follow-up time was 4.8±3.5 years (0–16.8 years). Table 1 summarises the demographic and clinical characteristics of the patients at initial evaluation and their outcomes.

SP genes variants

Overall, 383 patients (43.8%) had 265 distinct rare (minor allele frequency ≤0.2%) variants in one or more the eight SP genes most commonly associated with HCM *(MYH7*, *MYBPC3*, *TNNT2*, *TNNI3*, *MYL2*, *MYL3*, *ACTC1* and *TPM1)* (table 2 and see online supplementary table S1). A total of 142 (53.5%) of

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Table 2 Prevalence of rare variants (minor allele frequency ≤0.2%) in the eight main sarcomere genes

Gene	Number of cases	Percentage of sarcomere-positive individuals (N=383)	Percentage of the total cohort (N=874)
ACTC1	3	0.8	0.3
MYBPC3	191	49.9	21.9
MYH7	99	25.9	11.3
MYL2	6	1.6	0.7
MYL3	4	1.0	0.5
TNNI3	15	3.9	1.7
TNNT2	20	5.2	2.3
TPM1	8	2.1	0.9
Multiple	37	9.7	4.2
Total	383	100	43.8

The number and proportion of individuals for each individual gene excludes patients carrying more than one variant who are grouped under 'multiple'.

these rare variants were published previously as disease-causing mutations; 44 (16.6%) were novel missense variants predicted in silico to be pathogenic and 40 (15%) were novel potential loss-of-function variants. In all, 37 patients (4.2%) carried multiple candidate variants in these eight SP genes.

Non-SP gene variants

In all, 114 distinct rare desmosomal protein gene variants were present in 122 (14.0%) patients; 192 rare ion-channel disease gene variants were present in 196 patients (22.4%). A total of 29 (25.4%) of the desmosomal variants and 38 (19.8%) of the ion-channel variants were published previously. A further 74 (24.2%) of these non-sarcomere variants were novel missense variants predicted in silico to be pathogenic and 20 (6.5%) were potential loss-of-function variants. In all, 122 patients (43.0% of 284) with these non-SP variants also carried a SP variant.

Genotype–phenotype associations

Genotype–phenotype associations significant at the defined stringent thresholds are summarised in figures 1–3 and table 3. A complete list of p values significant at $p<0.05$ for all pairs of traits/genes is provided in table 4 for non-SP genes and online supplementary table S2 for SP and related genes. Online supplementary table S3 summarises the associations for non-SP genes presented in tables 3 and 4, analysed within the subcohort of sarcomere-positive individuals only.

Effect of mutations in sarcomere genes

Patients with at least one variant in one of the eight main sarcomere genes were younger at diagnosis and had a higher frequency of a family history of HCM or SCD compared with those without sarcomere variants. Patients with SP mutations were more likely to have asymmetric septal hypertrophy than apical or concentric patterns and had greater MLVWT. The prevalence of male sex was lower in sarcomere-positive individuals (62.4% vs 72.0%, p=0.00213); these individuals were also more likely to have an ICD implanted. Patients with sarcomere mutations had a lower resting systolic blood pressure (SBP) (123.1±19.2 vs 133.7±21.3 mm Hg, $p=1.54\times10^{-9}$) and a lower SBP response to exercise (44.1±21.5 vs 52.2±26.9 mm Hg, $p=7.61\times10^{-5}$).

Similar and additional associations were observed when individual SP genes were considered (table 3 and see online supplementary table S2).

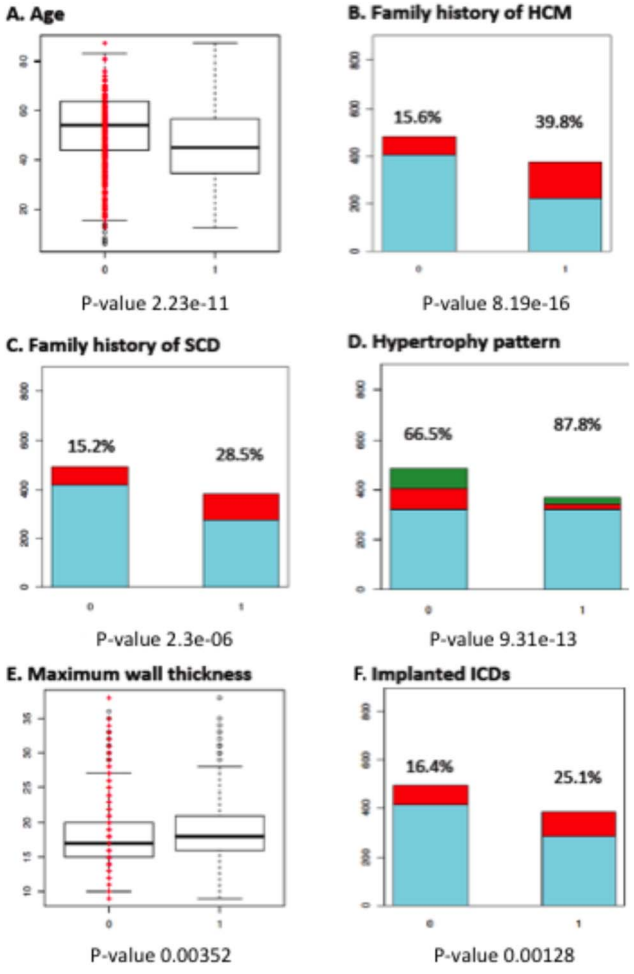

Figure 1 Comparison between sarcomere gene mutation-positive and -negative patients. (A) Age at initial evaluation (45.78±14.65 vs 53.05 ±14.94 years). (B) Family history of hypertrophic cardiomyopathy (HCM). (C) Family history of sudden cardiac death (SCD). (D) Hypertrophy pattern. (E) Maximum wall thickness (18.83±4.42 vs 18.12 ±4.08 mm). (F) Implanted implantable cardioverter-defibrillators (ICDs). Key: 0: sarcomere-negative; 1: sarcomere-positive. For B, C, E: red colour and percentages indicate the individuals with the trait within each genotype; for D light blue—asymmetric septal hypertrophy; red—apical hypertrophy; green—concentric hypertrophy.

The proportion of cardiovascular deaths during follow-up was higher in patients with at least one variant in one of the eight main SP genes. The same was true for sudden death/ICD discharge (figure 2 and 3).

Patients with multiple SP gene variants

Patients who carried more than one sarcomere variant had an increased prevalence of syncope when compared with individuals with only one sarcomere variant (35.1% vs 16.6%; 13/37 vs 56/337, p=0.012). SBP response to exercise was lower in individuals with multiple sarcomere variants compared with a single variant (36.5±21.9 vs 45.1±21.2 mm Hg, p=0.012) and there was a higher proportion of patients with an abnormal blood pressure response to exercise (10/29 vs 39/276; 34.5% vs 14.1%, p=0.010).

Associations with rare variants in desmosomal and ion-channel genes

A total of 71 patients carried rare *ANK2* variants (of these, 36 also carried SP variants). At a significance threshold of

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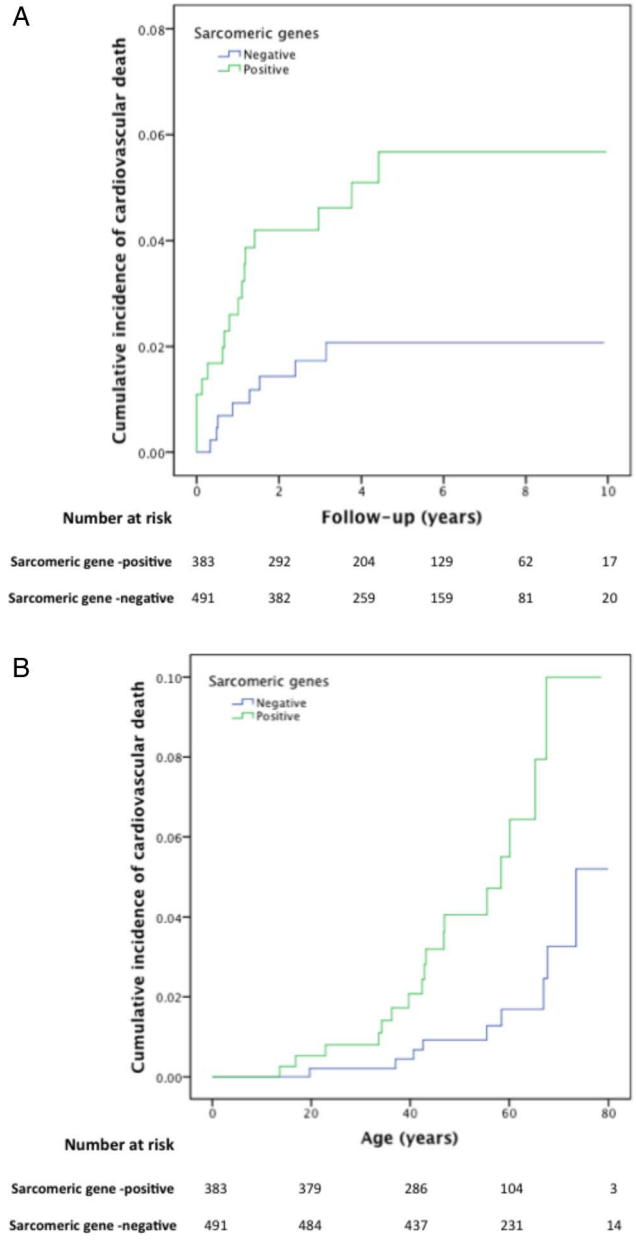

Figure 2 Kaplan–Meier cumulative incidence curves for cardiovascular death (see Methods section), comparing sarcomere-positive and sarcomere-negative individuals, modelled for (A): follow-up from first evaluation (years), log-rank test p value=0.012 (HR 2.81; 95% CI 1.21 to 6.51) and (B): time from birth (years), log-rank test p value=0.001 (HR 3.99; 95% CI 1.71 to 9.36). The Y axis values indicate proportions.

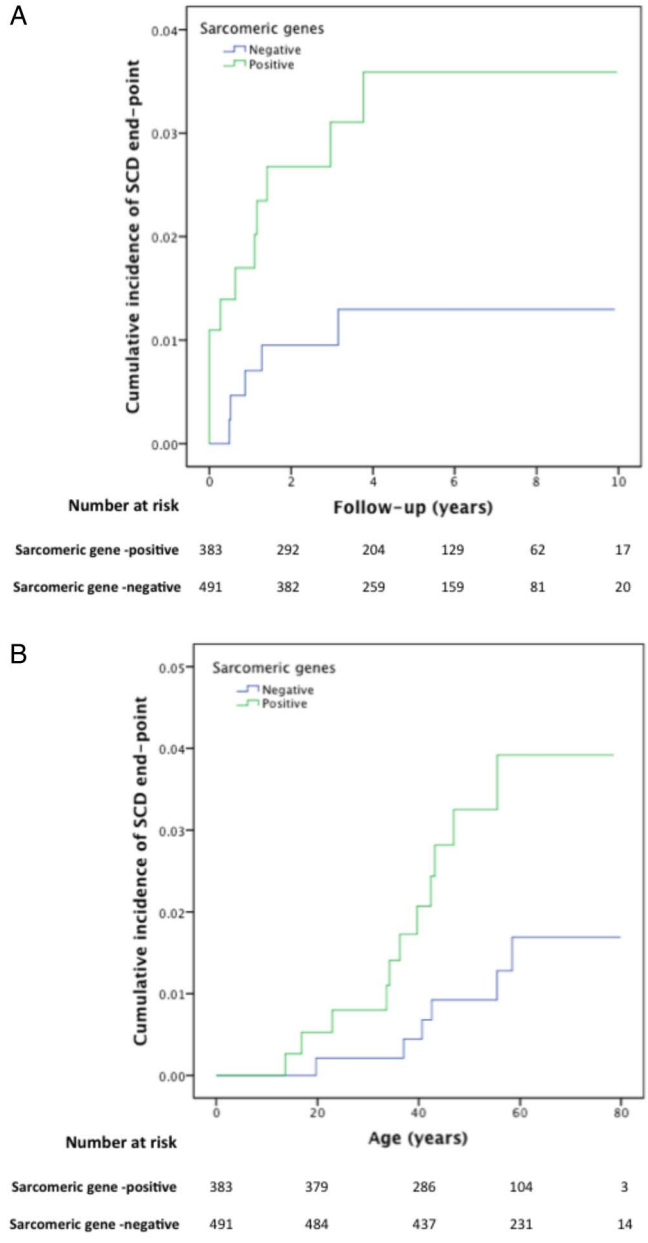

Figure 3 Kaplan–Meier cumulative incidence curves for sudden cardiac death/aborted sudden cardiac death, comparing sarcomere-positive and sarcomere-negative individuals, modelled for (A): follow-up from first evaluation (years), log-rank test p value=0.039 (HR 2.89; 95% CI 1.01 to 8.33) and (B): time from birth (years), log-rank test p value=0.028 (HR 3.44; 95% CI 1.19 to 9.92) . The Y axis values indicate proportions.

$p<0.0018$, the proportion of patients with an MLVWT ≥30 mm was greater in carriers of an *ANK2* rare variant (table 3). This association was still present when restraining the analyses to the subcohort of sarcomere-positive individuals only (see online supplementary table S3).

Additional genotype–phenotype correlations were identified at a less stringent $p<0.05$. These are listed in table 4, and include an increased mean MLVWT in *ANK2* variant carriers.

DISCUSSION

In this study of a large consecutive cohort of HCM probands screened with high-throughput sequencing, we have detected a class effect of SP gene variants on the HCM phenotype and identified novel associations with mutations in individual SP genes. We also demonstrated evidence of an association between non-SP genes and disease expression that could explain some of the characteristic clinical heterogeneity of HCM.

Influence of sarcomeric variation on phenotype

The presence of any sarcomere variant was associated with an asymmetric septal hypertrophy pattern, younger age at presentation, family history of HCM and SCD and female gender. This study also shows that patients with SP variants had higher cardiovascular and sudden death-related mortality during follow-up. Patients with more than one SP variant had more

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Table 3 Genotype–phenotype associations for individual sarcomeric and related protein genes and non-sarcomere protein (SP) genes meeting the predefined statistical thresholds for multiple testing.

Phenotype	Gene	Frequency or mean±SD Rare variant present	Frequency or mean±SD Rare variant absent	p Value
Age at initial evaluation (years)	*MYBPC3*	45.5±14.4	51.0±15.5	8.97×10^{-6}
	MYH7	43.9±15.4	50.5±15.2	1.91×10^{-5}
Family history of HCM	*MYBPC3*	40.4% (86/213)	21.9% (140/640)	2.7×10^{-7}
	MYH7	47.4% (54/114)	23.4% (173/740)	3.06×10^{-7}
Family history of SCD	*MYBPC3*	28.7% (62/216)	18.3% (120/656)	0.001
	MYH7	31.6% (37/117)	19.2% (145/775)	0.003
ASH pattern	*MYBPC3*	88.0% (184/209)	71.6% (459/641)	3.75×10^{-6}
	MYH7	89.2% (99/111)	73.6% (544/739)	0.001
MLVWT (mm)	*MYBPC3*	19.4±4.7	18.2±4.2	0.0005
MLVWT ≥30 mm	*ANK2*	12.5% (6/48)	2% (11/553)	0.0005
LV end-diastolic diameter (mm)	*MYBPC3*	44.8±5.5	46.3±6.0	0.00089
LV end-systolic diameter (mm)	*MYBPC3*	27.4±6.0	28.8±5.4	0.005
Right ventricular hypertrophy	*TNNI3*	50% (10/20)	21.6% (174/806)	0.004
SBP response to exercise (mmHg)	*MYH7*	36.6±19.9	50.2±24.4	5.49×10^{-6}
Abnormal SBP response to exercise	*TNNT2*	40.9% (9/22)	13.0% (83/640)	0.002
Myectomy and/or alcohol septal ablation and/or pacemaker implantation for gradient reduction	*MYBPC3* (splicing variants)	43.1% (22/51)	20.0% (160/799)	3.0×10^{-4}
LVOTO (>30 mm Hg)	*TNNI3*	10.0% (2/20)	41.2% (326/792)	0.005

p Values reflect the comparison for proportions or means between the group of patients with versus the group of patients without a rare variant in a given gene (p value thresholds of <0.0056 for SP genes and <0.0018 for non-SP genes).
ASH, asymmetric septal hypertrophy; SBP, systolic blood pressure; HCM, hypertrophic cardiomyopathy; LVOTO, LV outflow tract obstruction; MLVWT, maximum LV wall thickness; SCD, sudden cardiac death.

SCD risk markers, consistent with the suggestion in previously published series of a gene dose effect.[13 26–28] However, the low number of outcome events during follow-up may have biased the survival analysis and precluded an analysis of other associations, including the effect of carrying multiple compared with single variants. The survival from birth is provided for comparison with the published literature but also introduces an inherent survivor bias. With regard to individual SP genes, we demonstrate a number of novel associations that provide evidence for mutation specific effects on clinical phenotype and prognosis.

Modifier effect of non-sarcomere variants

The data in this study suggest that rare *ANK2* variants are associated with severe LV hypertrophy. *ANK2*, or *ankyrin B*, stabilises membrane ion-channels in cardiomyocytes and mutations in the gene cause long QT syndrome 4, ventricular arrhythmias and sinus node disease.[29 30] We are unaware of any link between *ANK2* expression and changes in LV morphology, but as ankyrins interact with proteins that influence calcium homeostasis and ß-adrenergic signalling, it is conceivable that they eventually affect the cellular phenotype that results from a primary SP gene variant. The strength of the statistical association (p=0.0005) exceeds the requirement of a Bonferroni correction for the number of tested genes (36 independent tests), but further replication in independent cohorts will be necessary to confirm these results.

In addition to the association with *ANK2* variation, we detected a number of associations at lower statistical significance with variation in other non-SP genes. Patients with *SCN5A* rare variants were more likely to have left atria enlargement at their last evaluation. A link between *SCN5A* disruption and TGF-β_1-mediated fibrosis has recently been demonstrated in a murine model of sinus node disease[w28] and it is possible that *SCN5A* variants influence the pro-fibrotic milieu associated with SP mutations. SCN5A rare variation was also associated with a higher proportion of LV outflow tract obstruction. Individuals with *PLN* rare variants were more likely to have NSVT, which is interesting considering the recently described arrhythmogenic risk of a founder *PLN* mutation.[w29] As for the association with *ANK2*, replication of these findings is required.

Table 4 Genotype–phenotype associations for non-sarcomeric protein genes not meeting the predefined statistical thresholds for multiple testing

Phenotype	Gene	Frequency or mean±SD—Variant present	Frequency or mean±SD—Variant absent	p Value
LA diameter at last follow-up (mm)	*SCN5A*	46.9±5.4	44.3±7.5	0.04
LVOTO (>30 mm Hg)	*SCN5A*	57.5% (23/40)	39.5% (305/772)	0.03
	Ion-channel	49.2% (89/181)	37.9% (239/631)	0.006
MLVWT (mm)	*ANK2*	19.7±5.6	18.4±4.2	0.02
E/e′ ratio	*CASQ2*	16.4±7.1	11.4±5.9	0.02
NSVT	*PLN*	100% (3/3)	22.1% (124/562)	0.01

p Values reflect the comparison for proportions or means between patients with and without a rare variant in a given gene.
E to é ratio, ratio between the maximal velocity of the E wave from the pulsed wave Doppler of the transmitral flow and the maximal velocity of the e′ wave of tissue Doppler at the mitral annulus; LA, left atria; LVOTO, LV outflow tract obstruction; MLVWT, maximal LV wall thickness; NSVT, non-sustained ventricular tachycardia.

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Clinical implications

If genetic variation is to become a clinically relevant biomarker, it is essential that there is a clear understanding of genotype–phenotype relationships. The associations between sarcomere gene variants and the broad phenotype examined in this study contribute to this understanding and, if confirmed in other populations, could inform the counselling of patients and relatives who are contemplating predictive genetic testing. The demonstration that non-sarcomeric variants may influence disease expression is an illustration of the complexity that underlies the biology of this disease. New models that incorporate a broad genetic profile and deep clinical phenotyping are necessary to test the role of mutation analysis in prognostic models.

Key messages

What is known on this subject?

In up to 50% of cases, hypertrophic cardiomyopathy is caused by mutations in genes coding for sarcomere or sarcomere-related genes, but the often dramatic variation in clinical phenotypes caused by the same or similar mutations remains largely unexplained.

What might this study add?

This study presents novel genotype–phenotype associations in a large cohort of 874 patients using high-throughput genetic sequencing. We describe a strong class effect for sarcomeric protein variants on clinical presentation, LV morphology and survival. For the first time, a modifier effect of rare variants in non-sarcomeric genes associated with other forms of cardiomyopathy and arrhythmia syndromes is demonstrated.

How might this impact on clinical practice?

These are novel findings which suggest new and testable insights on the biology and pathophysiology of the disease that might eventually have important clinical implications for counselling of patients and risk prediction models.

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Contributors LRL: conception and design of the study, analysis and interpretation of data, drafting of the manuscript, and final approval of the manuscript submitted; PS, WJM, PME and VP: conception and design of the study, analysis and interpretation of data, revising the manuscript critically for important intellectual content, and final approval of the manuscript submitted; OPG, MH, CO'M, HCT, CD, SJ and LM: analysis and interpretation of data, revising the manuscript critically for important intellectual content, and final approval of the manuscript submitted.

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Competing interests LM is a shareholder of Health in Code SL.

Ethics approval University College London (UCL)/UCL Hospitals (UCLH) Joint Research Ethics Committee.

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Supplementary table 1. List of distinct rare variants (MAF<0.2%; see methods). All variants were detected in heterozygozity. Type of variant: D: variant reported on dbSNP137; L: novel predicted loss-of-function (nonsense, frameshift or splice site) variant; N: novel non-frameshift or missense variant not predicted *in silico* to be pathogenic; P: variant reported in the literature as disease-associated; S: novel missense variant predicted *in silico* to be pathogenic. MAF: minor allele frequency.

Gene	Genomic signature	Transcript / Amino acid change	dbSNP137 ID	Type of variant	MAF- cases
ACTC1	15_35084398_G_A	ENSG00000159251:ENST00000290378:exon5:c.C701T:p.S234F		N	0,00057
ACTC1	15_35085539_T_C	ENSG00000159251:ENST00000290378:exon3:c.A361G:p.M121V		S	0,00058
ACTC1	15_35085599_C_T	ENSG00000159251:ENST00000290378:exon3:c.G301A:p.E101K	rs193922680	D;P	0,00057
ANK2	4_113970940_G_A	ENSG00000145362:ENST00000264366:exon1:c.G56A:p.S19N,ENSG00000145362:ENST00000357077:exon1:c.G56A:p.S19N,ENSG00000145362:ENST00000394537:exon1:c.G56A:p.S19N,ENSG00000145362:ENST00000504454:exon1:c.G56A:p.S19N		N	0,00057
ANK2	4_114067089_T_A	ENSG00000145362:ENST00000511380:exon1:c.T65A:p.M22K	rs116338686	D	0,00115
ANK2	4_114072285_T_C	ENST00000504454:exon2:c.129+5T>C		L	0,00057
ANK2	4_114158192_C_T	ENSG00000145362:ENST00000264366:exon6:c.C533T:p.A178V,ENSG00000145362:ENST00000357077:exon6:c.C533T:p.A178V,ENSG00000145362:ENST00000394537:exon6:c.C533T:p.A178V,ENSG00000145362:ENST00000515034:exon6:c.C128T:p.A43V,ENSG00000145362:ENST00000503271:exon7:c.C470T:p.A157V,ENSG00000145362:ENST00000503423:exon7:c.C470T:p.A157V,ENSG00000145362:ENST00000504454:exon7:c.C578T:p.A193V,ENSG00000145362:ENST00000506722:exon7:c.C470T:p.A157V		N	0,00058
ANK2	4_114195614_C_G	ENSG00000145362:ENST00000264366:exon15:c.C1492G:p.Q498E,ENSG00000145362:ENST00000357077:exon15:c.C1492G:p.Q498E,ENSG00000145362:ENST00000394537:exon15:c.C1492G:p.Q498E,ENSG00000145362:ENST00000503271:exon16:c.C1429G:p.Q477E,ENSG00000145362:ENST00000503423:exon16:c.C1429G:p.Q477E,ENSG00000145362:ENST00000504454:exon16:c.C1537G:p.Q513E,ENSG00000145362:ENST00000506722:exon16:c.C1429G:p.Q477E		S	0,00057
ANK2	4_114199026_T_C	ENSG00000145362:ENST00000264366:exon16:c.T1717C:p.Y573H,ENSG00000145362:ENST00000357077:exon16:c.T1717C:p.Y573H,ENSG00000145362:ENST00000394537:exon16:c.T1717C:p.Y573H,ENSG00000145362:ENST00000503271:exon17:c.T1654C:p.Y552H,ENSG00000145362:ENST00000503423:exon17:c.T1654C:p.Y552H,ENSG00000145362:ENST00000504454:exon17:c.T1762C:p.Y588H,ENSG00000145362:ENST00000506722:exon17:c.T1654C:p.Y552H		S	0,00057

ANK2	4_114208841_T_A	ENSG00000145362:ENST00000264366:exon19:c.T2160A:p.D720E,ENSG00000145362:ENST00000357077:exon19:c.T2160A:p.D720E,ENSG00000145362:ENST00000394537:exon19:c.T2160A:p.D720E,ENSG00000145362:ENST00000503423:exon19:c.T1998A:p.D666E,ENSG00000145362:ENST00000503271:exon20:c.T2097A:p.D699E,ENSG00000145362:ENST00000504454:exon20:c.T2205A:p.D735E,ENSG00000145362:ENST00000506722:exon20:c.T2097A:p.D699E		N	0,00057
ANK2	4_114239695_C_G	ENSG00000145362:ENST00000509550:exon6:c.C446G:p.A149G,ENSG00000145362:ENST00000264366:exon26:c.C2819G:p.A940G,ENSG00000145362:ENST00000357077:exon26:c.C2819G:p.A940G,ENSG00000145362:ENST00000394537:exon26:c.C2819G:p.A940G,ENSG00000145362:ENST00000503423:exon26:c.C2657G:p.A886G,ENSG00000145362:ENST00000503271:exon27:c.C2756G:p.A919G,ENSG00000145362:ENST00000504454:exon27:c.C2864G:p.A955G,ENSG00000145362:ENST00000506722:exon27:c.C2756G:p.A919G		S	0,00115
ANK2	4_114244911_G_A	ENST00000506722:exon28:-4G>A	rs139641776	D; L	0,00057
ANK2	4_114244926_C_A	ENSG00000145362:ENST00000506722:exon28:c.C2849A:p.P950Q		N	0,00114
ANK2	4_114251533_G_A	ENSG00000145362:ENST00000509550:exon7:c.G659A:p.R220H,ENSG00000145362:ENST00000264366:exon27:c.G3032A:p.R1011H,ENSG00000145362:ENST00000357077:exon27:c.G3032A:p.R1011H,ENSG00000145362:ENST00000394537:exon27:c.G3032A:p.R1011H,ENSG00000145362:ENST00000503423:exon27:c.G2870A:p.R957H,ENSG00000145362:ENST00000503271:exon28:c.G2969A:p.R990H,ENSG00000145362:ENST00000504454:exon28:c.G3077A:p.R1026H,ENSG00000145362:ENST00000506722:exon29:c.G3005A:p.R1002H		S	0,00058
ANK2	4_114251575_G_C	ENSG00000145362:ENST00000509550:exon7:c.G701C:p.G234A,ENSG00000145362:ENST00000264366:exon27:c.G3074C:p.G1025A,ENSG00000145362:ENST00000357077:exon27:c.G3074C:p.G1025A,ENSG00000145362:ENST00000394537:exon27:c.G3074C:p.G1025A,ENSG00000145362:ENST00000503423:exon27:c.G2912C:p.G971A,ENSG00000145362:ENST00000503271:exon28:c.G3011C:p.G1004A,ENSG00000145362:ENST00000504454:exon28:c.G3119C:p.G1040A,ENSG00000145362:ENST00000506722:exon29:c.G3047C:p.G1016A		S	0,00058
ANK2	4_114263066_T_G	ENSG00000145362:ENST00000510275:exon1:c.T72G:p.D24E,ENSG00000145362:ENST00000509550:exon12:c.T1644G:p.D548E,ENSG00000145362:ENST00000264366:exon32:c.T4017G:p.D1339E,ENSG00000145362:ENST00000503423:exon32:c.T3855G:p.D1285E,ENSG00000145362:ENST00000357077:exon33:c.T4116G:p.D1372E,ENSG00000145362:ENST00000394537:exon33:c.T4116G:p.D1372E,ENSG00000145362:ENST00000504454:exon34:c.T4161G:p.D1387E,ENSG00000145362:ENST00000506722:exon35:c.T4089G:p.D1363E		S	0,00057

ANK2	4_114267117_C_T	ENSG00000145362:ENST00000510275:exon3:c.C266T:p.T89M,ENSG00000145362:ENST00000509550:exon14:c.C1838T:p.T613M,ENSG00000145362:ENST00000264366:exon34:c.C4211T:p.T1404M,ENSG00000145362:ENST00000503423:exon34:c.C4049T:p.T1350M,ENSG00000145362:ENST00000357077:exon35:c.C4310T:p.T1437M,ENSG00000145362:ENST00000394537:exon35:c.C4310T:p.T1437M,ENSG00000145362:ENST00000504454:exon36:c.C4355T:p.T1452M,ENSG00000145362:ENST00000506722:exon37:c.C4283T:p.T1428M	rs142534126	D;S	0,00057
ANK2	4_114267122_G_T	ENSG00000145362:ENST00000510275:exon3:c.G271T:p.G91C,ENSG00000145362:ENST00000509550:exon14:c.G1843T:p.G615C,ENSG00000145362:ENST00000264366:exon34:c.G4216T:p.G1406C,ENSG00000145362:ENST00000503423:exon34:c.G4054T:p.G1352C,ENSG00000145362:ENST00000357077:exon35:c.G4315T:p.G1439C,ENSG00000145362:ENST00000394537:exon35:c.G4315T:p.G1439C,ENSG00000145362:ENST00000504454:exon36:c.G4360T:p.G1454C,ENSG00000145362:ENST00000506722:exon37:c.G4288T:p.G1430C	rs34591340	D;P	0,00057
ANK2	4_114267176_A_G	ENSG00000145362:ENST00000510275:exon3:c.A325G:p.K109E,ENSG00000145362:ENST00000509550:exon14:c.A1897G:p.K633E,ENSG00000145362:ENST00000264366:exon34:c.A4270G:p.K1424E,ENSG00000145362:ENST00000503423:exon34:c.A4108G:p.K1370E,ENSG00000145362:ENST00000357077:exon35:c.A4369G:p.K1457E,ENSG00000145362:ENST00000394537:exon35:c.A4369G:p.K1457E,ENSG00000145362:ENST00000504454:exon36:c.A4414G:p.K1472E,ENSG00000145362:ENST00000506722:exon37:c.A4342G:p.K1448E		N	0,00057
ANK2	4_114269433_A_G	ENSG00000145362:ENST00000510275:exon4:c.A329G:p.E110G,ENSG00000145362:ENST00000509550:exon15:c.A1901G:p.E634G,ENSG00000145362:ENST00000264366:exon35:c.A4274G:p.E1425G,ENSG00000145362:ENST00000503423:exon35:c.A4112G:p.E1371G,ENSG00000145362:ENST00000357077:exon36:c.A4373G:p.E1458G,ENSG00000145362:ENST00000394537:exon36:c.A4373G:p.E1458G,ENSG00000145362:ENST00000504454:exon37:c.A4418G:p.E1473G,ENSG00000145362:ENST00000506722:exon38:c.A4346G:p.E1449G	rs72544141	D;P	0,00172
ANK2	4_114274519_G_A	ENSG00000145362:ENST00000264366:exon37:c.G4646A:p.R1549Q,ENSG00000145362:ENST00000503423:exon37:c.G4484A:p.R1495Q,ENSG00000145362:ENST00000357077:exon38:c.G4745A:p.R1582Q,ENSG00000145362:ENST00000504454:exon39:c.G4790A:p.R1597Q	rs138842207	D	0,00286
ANK2	4_114274546_A_G	ENSG00000145362:ENST00000264366:exon37:c.A4673G:p.E1558G,ENSG00000145362:ENST00000503423:exon37:c.A4511G:p.E1504G,ENSG00000145362:ENST00000357077:exon38:c.A4772G:p.E1591G,ENSG00000145362:ENST00000504454:exon39:c.A4817G:p.E1606G		S	0,00057
ANK2	4_114274806_G_A	ENSG00000145362:ENST00000264366:exon37:c.G4933A:p.E1645K,ENSG00000145362:ENST00000503423:exon37:c.G4771A:p.E1591K,ENSG00000145362:ENST00000357077:exon38:c.G5032A:p.E1678K,ENSG00000145362:ENST00000504454:exon39:c.G5077A:p.E1693K		N	0,00057
ANK2	4_114275005_C_A	ENSG00000145362:ENST00000264366:exon37:c.C5132A:p.A1711D,ENSG00000145362:ENST00000357077:exon38:c.C5231A:p.A1744D	rs147706514	D	0,00057
ANK2	4_114275005_C_T	ENSG00000145362:ENST00000264366:exon37:c.C5132T:p.A1711V,ENSG00000145362:ENST00000357077:exon38:c.C5231T:p.A1744V		N	0,00057

ANK2	4_114275096_G_T	ENSG00000145362:ENST00000264366:exon37:c.G5223T:p.Q1741H,ENSG00000145362:ENST00000357077:exon38:c.G5322T:p.Q1774H		S	0,00057
ANK2	4_114275262_C_T	ENSG00000145362:ENST00000264366:exon37:c.C5389T:p.H1797Y,ENSG00000145362:ENST00000357077:exon38:c.C5488T:p.H1830Y		N	0,00057
ANK2	4_114275344_T_A	ENSG00000145362:ENST00000264366:exon37:c.T5471A:p.V1824E,ENSG00000145362:ENST00000357077:exon38:c.T5570A:p.V1857E	rs141212932	D	0,00057
ANK2	4_114275425_C_T	ENSG00000145362:ENST00000264366:exon37:c.C5552T:p.S1851L,ENSG00000145362:ENST00000357077:exon38:c.C5651T:p.S1884L	rs150737736	D	0,00057
ANK2	4_114275688_C_A	ENSG00000145362:ENST00000264366:exon37:c.C5815A:p.Q1939K,ENSG00000145362:ENST00000357077:exon38:c.C5914A:p.Q1972K		N	0,00057
ANK2	4_114275852_A_T	ENSG00000145362:ENST00000264366:exon37:c.A5979T:p.K1993N,ENSG00000145362:ENST00000357077:exon38:c.A6078T:p.K2026N		N	0,00115
ANK2	4_114275950_C_T	ENSG00000145362:ENST00000264366:exon37:c.C6077T:p.T2026M,ENSG00000145362:ENST00000357077:exon38:c.C6176T:p.T2059M	rs200765866	D;S	0,00058
ANK2	4_114275980_G_A	ENSG00000145362:ENST00000264366:exon37:c.G6107A:p.R2036H,ENSG00000145362:ENST00000357077:exon38:c.G6206A:p.R2069H	rs149645600	D;S	0,00058
ANK2	4_114276279_G_A	ENSG00000145362:ENST00000264366:exon37:c.G6406A:p.V2136I,ENSG00000145362:ENST00000357077:exon38:c.G6505A:p.V2169I	rs149292242	D;S	0,00057
ANK2	4_114276360_G_A	ENSG00000145362:ENST00000264366:exon37:c.G6487A:p.G2163S,ENSG00000145362:ENST00000357077:exon38:c.G6586A:p.G2196S		N	0,00057
ANK2	4_114276657_A_G	ENSG00000145362:ENST00000264366:exon37:c.A6784G:p.T2262A,ENSG00000145362:ENST00000357077:exon38:c.A6883G:p.T2295A		N	0,00058
ANK2	4_114276879_G_T	ENSG00000145362:ENST00000264366:exon37:c.G7006T:p.V2336F,ENSG00000145362:ENST00000357077:exon38:c.G7105T:p.V2369F		N	0,00130
ANK2	4_114276891_A_G	ENSG00000145362:ENST00000264366:exon37:c.A7018G:p.T2340A,ENSG00000145362:ENST00000357077:exon38:c.A7117G:p.T2373A	rs184514058	D	0,00066
ANK2	4_114276922_C_T	ENSG00000145362:ENST00000264366:exon37:c.C7049T:p.P2350L,ENSG00000145362:ENST00000357077:exon38:c.C7148T:p.P2383L	rs35960628	D	0,00066
ANK2	4_114276957_A_C	ENSG00000145362:ENST00000264366:exon37:c.A7084C:p.T2362P,ENSG00000145362:ENST00000357077:exon38:c.A7183C:p.T2395P	rs201693280	D	0,00133
ANK2	4_114277041_G_A	ENSG00000145362:ENST00000264366:exon37:c.G7168A:p.A2390T,ENSG00000145362:ENST00000357077:exon38:c.G7267A:p.A2423T	rs3733616	D	0,00062
ANK2	4_114277108_A_G	ENSG00000145362:ENST00000264366:exon37:c.A7235G:p.D2412G,ENSG00000145362:ENST00000357077:exon38:c.A7334G:p.D2445G		S	0,00059
ANK2	4_114277642_C_G	ENSG00000145362:ENST00000264366:exon37:c.C7769G:p.S2590C,ENSG00000145362:ENST00000357077:exon38:c.C7868G:p.S2623C	rs116253689	D	0,00057

ANK2	4_114277871_G_A	ENSG00000145362:ENST00000264366:exon37:c.G7998A:p.M2666I,ENSG00000145362:ENST00000357077:exon38:c.G8097A:p.M2699I	rs148904454	D	0,00058
ANK2	4_114278014_G_A	ENSG00000145362:ENST00000264366:exon37:c.G8141A:p.R2714H,ENSG00000145362:ENST00000357077:exon38:c.G8240A:p.R2747H	rs142137451	D	0,00057
ANK2	4_114278542_A_G	ENSG00000145362:ENST00000264366:exon37:c.A8669G:p.Q2890R,ENSG00000145362:ENST00000357077:exon38:c.A8768G:p.Q2923R		S	0,00057
ANK2	4_114278701_C_G	ENSG00000145362:ENST00000264366:exon37:c.C8828G:p.S2943C,ENSG00000145362:ENST00000357077:exon38:c.C8927G:p.S2976C		S	0,00057
ANK2	4_114278820_G_A	ENSG00000145362:ENST00000505342:exon1:c.G76A:p.E26K,ENSG00000145362:ENST00000264366:exon37:c.G8947A:p.E2983K,ENSG00000145362:ENST00000357077:exon38:c.G9046A:p.E3016K	rs149963885	D	0,00057
ANK2	4_114279054_C_G	ENSG00000145362:ENST00000505342:exon1:c.C310G:p.P104A,ENSG00000145362:ENST00000264366:exon37:c.C9181G:p.P3061A,ENSG00000145362:ENST00000357077:exon38:c.C9280G:p.P3094A		S	0,00115
ANK2	4_114279615_C_G	ENSG00000145362:ENST00000505342:exon1:c.C871G:p.Q291E,ENSG00000145362:ENST00000264366:exon37:c.C9742G:p.Q3248E,ENSG00000145362:ENST00000357077:exon38:c.C9841G:p.Q3281E		N	0,00057
ANK2	4_114279646_C_G	ENSG00000145362:ENST00000505342:exon1:c.C902G:p.P301R,ENSG00000145362:ENST00000264366:exon37:c.C9773G:p.P3258R,ENSG00000145362:ENST00000357077:exon38:c.C9872G:p.P3291R		N	0,00057
ANK2	4_114279892_C_T	ENSG00000145362:ENST00000505342:exon1:c.C1148T:p.A383V,ENSG00000145362:ENST00000264366:exon37:c.C10019T:p.A3340V,ENSG00000145362:ENST00000357077:exon38:c.C10118T:p.A3373V		N	0,00059
ANK2	4_114280441_A_G	ENSG00000145362:ENST00000505342:exon1:c.A1697G:p.H566R,ENSG00000145362:ENST00000264366:exon37:c.A10568G:p.H3523R,ENSG00000145362:ENST00000357077:exon38:c.A10667G:p.H3556R		S	0,00057
ANK2	4_114281999_C_T	ENSG00000145362:ENST00000505342:exon2:c.C1732T:p.R578W,ENSG00000145362:ENST00000510275:exon6:c.C403T:p.R135W,ENSG00000145362:ENST00000509550:exon17:c.C1975T:p.R659W,ENSG00000145362:ENST00000264366:exon38:c.C10603T:p.R3535W,ENSG00000145362:ENST00000394537:exon38:c.C4447T:p.R1483W,ENSG00000145362:ENST00000357077:exon39:c.C10702T:p.R3568W,ENSG00000145362:ENST00000506722:exon40:c.C4420T:p.R1474W	rs72556376	D;P	0,00057
ANK2	4_114282005_G_A	ENSG00000145362:ENST00000505342:exon2:c.G1738A:p.E580K,ENSG00000145362:ENST00000510275:exon6:c.G409A:p.E137K,ENSG00000145362:ENST00000509550:exon17:c.G1981A:p.E661K,ENSG00000145362:ENST00000264366:exon38:c.G10609A:p.E3537K,ENSG00000145362:ENST00000394537:exon38:c.G4453A:p.E1485K,ENSG00000145362:ENST00000357077:exon39:c.G10708A:p.E3570K,ENSG00000145362:ENST00000506722:exon40:c.G4426A:p.E1476K	rs180843436	D;P	0,00057

ANK2	4_114290677_A_G	ENSG00000145362:ENST00000505342:exon6:c.A2356G:p.T786A,ENSG00000145362:ENST00000510275:exon10:c.A1027G:p.T343A,ENSG00000145362:ENST00000509550:exon21:c.A2599G:p.T867A,ENSG00000145362:ENST00000264366:exon42:c.A11227G:p.T3743A,ENSG00000145362:ENST00000394537:exon42:c.A5071G:p.T1691A,ENSG00000145362:ENST00000357077:exon43:c.A11326G:p.T3776A,ENSG00000145362:ENST00000506722:exon44:c.A5044G:p.T1682A		S	0,00057
ANK2	4_114290837_C_T	ENSG00000145362:ENST00000505342:exon6:c.C2516T:p.P839L,ENSG00000145362:ENST00000510275:exon10:c.C1187T:p.P396L,ENSG00000145362:ENST00000509550:exon21:c.C2759T:p.P920L,ENSG00000145362:ENST00000264366:exon42:c.C11387T:p.P3796L,ENSG00000145362:ENST00000394537:exon42:c.C5231T:p.P1744L,ENSG00000145362:ENST00000357077:exon43:c.C11486T:p.P3829L,ENSG00000145362:ENST00000506722:exon44:c.C5204T:p.P1735L		N	0,00058
ANK2	4_114290844_G_T	ENSG00000145362:ENST00000505342:exon6:c.G2523T:p.E841D,ENSG00000145362:ENST00000510275:exon10:c.G1194T:p.E398D,ENSG00000145362:ENST00000509550:exon21:c.G2766T:p.E922D,ENSG00000145362:ENST00000264366:exon42:c.G11394T:p.E3798D,ENSG00000145362:ENST00000394537:exon42:c.G5238T:p.E1746D,ENSG00000145362:ENST00000357077:exon43:c.G11493T:p.E3831D,ENSG00000145362:ENST00000506722:exon44:c.G5211T:p.E1737D	rs144046572	D;S	0,00058
ANK2	4_114290875_C_T	ENSG00000145362:ENST00000505342:exon6:c.C2554T:p.R852W,ENSG00000145362:ENST00000510275:exon10:c.C1225T:p.R409W,ENSG00000145362:ENST00000509550:exon21:c.C2797T:p.R933W,ENSG00000145362:ENST00000264366:exon42:c.C11425T:p.R3809W,ENSG00000145362:ENST00000394537:exon42:c.C5269T:p.R1757W,ENSG00000145362:ENST00000357077:exon43:c.C11524T:p.R3842W,ENSG00000145362:ENST00000506722:exon44:c.C5242T:p.R1748W	rs139797180	D;S	0,00057
ANK2	4_114293765_G_A	ENSG00000145362:ENST00000510275:exon11:c.G1388A:p.R463Q		N	0,00057
ANK2	4_114294276_A_G	ENSG00000145362:ENST00000505342:exon7:c.A2671G:p.T891A,ENSG00000145362:ENST00000510275:exon12:c.A1435G:p.T479A,ENSG00000145362:ENST00000509550:exon22:c.A2914G:p.T972A,ENSG00000145362:ENST00000264366:exon43:c.A11542G:p.T3848A,ENSG00000145362:ENST00000394537:exon43:c.A5386G:p.T1796A,ENSG00000145362:ENST00000357077:exon44:c.A11641G:p.T3881A,ENSG00000145362:ENST00000506722:exon45:c.A5359G:p.T1787A		N	0,00057
ANK2	4_114294462_C_T	ENSG00000145362:ENST00000505342:exon8:c.C2746T:p.R916W,ENSG00000145362:ENST00000510275:exon13:c.C1510T:p.R504W,ENSG00000145362:ENST00000509550:exon23:c.C2989T:p.R997W,ENSG00000145362:ENST00000264366:exon44:c.C11617T:p.R3873W,ENSG00000145362:ENST00000394537:exon44:c.C5461T:p.R1821W,ENSG00000145362:ENST00000357077:exon45:c.C11716T:p.R3906W,ENSG00000145362:ENST00000506722:exon46:c.C5434T:p.R1812W	rs121912706	D;P	0,00172
ANK2	4_114294488_G_C	ENSG00000145362:ENST00000505342:exon8:c.G2772C:p.E924D,ENSG00000145362:ENST00000510275:exon13:c.G1536C:p.E512D,ENSG00000145362:ENST00000509550:exon23:c.G3015C:p.E1005D,ENSG00000145362:ENST00000264366:exon44:c.G11643C:p.E3881D,ENSG00000145362:ENST00000394537:exon44:c.G5487C:p.E1829D,ENSG00000145362:ENST00000357077:exon45:c.G11742C:p.E3914D,ENSG00000145362:ENST00000506722:exon46:c.G5460C:p.E1820D		S	0,00057

ANK2	4_114295915_A_G	ENST00000514960:exon19:c.2702-5A>G,ENST00000505342:exon9:c.2890-5A>G,ENST00000506344:exon2:c.134-5A>G		L	0,00057
ANK2	4_114296105_A_G	ENST00000514960:exon19:c.2884+3A>G,ENST00000505342:exon9:c.3072+3A>G,ENST00000506344:exon2:c.316+3A>G		L	0,00057
ANK2	4_114299338_G_A	ENST00000506344:exon3:c.408+1G>A,ENST00000514167:exon2:c.183+1G>A	rs180914830	D;L	0,00057
CASQ2	1_116243897_C_A	ENSG00000118729:ENST00000456138:exon9:c.G952T:p.D318Y,ENSG00000118729:ENST00000261448:exon11:c.G1165T:p.D389Y		N	0,00061
CASQ2	1_116243928_ATC_-	ENSG00000118729:ENST00000456138:exon9:c.919_921del:p.307_307del,ENSG00000118729:ENST00000261448:exon11:c.1132_1134del:p.378_378del		N	0,00060
CASQ2	1_116247824_C_T	ENSG00000118729:ENST00000456138:exon7:c.G715A:p.D239N,ENSG00000118729:ENST00000261448:exon9:c.G928A:p.D310N	rs141314684	D	0,00057
CASQ2	1_116269737_T_C	ENSG00000118729:ENST00000456138:exon4:c.A400G:p.K134E,ENSG00000118729:ENST00000261448:exon6:c.A613G:p.K205E		S	0,00057
CASQ2	1_116275587_C_G	ENSG00000118729:ENST00000456138:exon3:c.G328C:p.A110P,ENSG00000118729:ENST00000261448:exon5:c.G541C:p.A181P		N	0,00057
CASQ2	1_116283431_C_T	ENSG00000118729:ENST00000261448:exon3:c.G338A:p.S113N	rs199750975	D	0,00057
CASQ2	1_116287470_C_T	ENSG00000118729:ENST00000261448:exon2:c.G298A:p.A100T,ENSG00000118729:ENST00000456138:exon2:c.G298A:p.A100T		N	0,00057
CASQ2	1_116310926_T_C	ENST00000261448:exon2:c.234+3A>G,ENST00000456138:exon2:c.234+3A>G		L	0,00057
CAV3	3_8775602_G_A	ENSG00000182533:ENST00000343849:exon1:c.G40A:p.V14I,ENSG00000182533:ENST00000397368:exon1:c.G40A:p.V14I	rs121909281	D	0,00115
CAV3	3_8787374_G_A	ENSG00000182533:ENST00000343849:exon2:c.G277A:p.A93T,ENSG00000182533:ENST00000397368:exon2:c.G277A:p.A93T	rs28936686	D;P	0,00063
CSRP3	11_19204270_G_A	ENSG00000129170:ENST00000265968:exon6:c.C532T:p.P178S,ENSG00000129170:ENST00000533783:exon7:c.C532T:p.P178S		S	0,00057
CSRP3	11_19206558_C_T	ENSG00000129170:ENST00000265968:exon5:c.G449A:p.C150Y,ENSG00000129170:ENST00000533783:exon6:c.G449A:p.C150Y		S	0,00172
CSRP3	11_19206593_C_A	ENST00000265968:exon6:c.415-1G>T,ENST00000533783:exon7:c.415-1G>T		L	0,00115
CSRP3	11_19209682_C_G	ENST00000265968:exon4:c.281+1G>C,ENST00000533783:exon5:c.281+1G>C		L	0,00057
CSRP3	11_19209752_A_G	ENSG00000129170:ENST00000265968:exon3:c.T212C:p.I71T,ENSG00000129170:ENST00000533783:exon4:c.T212C:p.I71T		N	0,00057
CSRP3	11_19209773_C_T	ENSG00000129170:ENST00000265968:exon3:c.G191A:p.R64H,ENSG00000129170:ENST00000533783:exon4:c.G191A:p.R64H		S	0,00057

CSRP3	11_19209833_A_G	ENSG00000129170:ENST00000265968:exon3:c.T131C:p.L44P,ENSG00000129170:ENST00000533783:exon4:c.T131C:p.L44P	rs104894205	D;P	0,00172
CSRP3	11_19213968_A_G	ENSG00000129170:ENST00000265968:exon2:c.T28C:p.C10R,ENSG00000129170:ENST00000533783:exon3:c.T28C:p.C10R		S	0,00057
CSRP3	11_19213974_C_T	ENSG00000129170:ENST00000265968:exon2:c.G22A:p.A8T,ENSG00000129170:ENST00000533783:exon3:c.G22A:p.A8T	rs45531937	D	0,00057
DES	2_220284854_A_G	ENSG00000175084:ENST00000373960:exon2:c.A616G:p.N206D		N	0,00058
DES	2_220285382_GG_-			L	0,00059
DES	2_220288540_G_A	ENSG00000175084:ENST00000373960:exon7:c.G1286A:p.R429Q	rs200580581	D	0,00059
DSC2	18_28648063_C_T	ENSG00000134755:ENST00000280904:exon16:c.G2624A:p.R875Q		N	0,00057
DSC2	18_28648870_C_T	ENSG00000134755:ENST00000251081:exon15:c.G2498A:p.R833H,ENSG00000134755:ENST00000280904:exon15:c.G2498A:p.R833H		P	0,00058
DSC2	18_28648998_TCC_-	ENSG00000134755:ENST00000251081:exon15:c.2368_2370del:p.790_790del,ENSG00000134755:ENST00000280904:exon15:c.2368_2370del:p.790_790del		N	0,00058
DSC2	18_28649002_CCT_-	ENSG00000134755:ENST00000251081:exon15:c.2364_2366del:p.788_789del,ENSG00000134755:ENST00000280904:exon15:c.2364_2366del:p.788_789del		P	0,00058
DSC2	18_28649033_C_T	ENSG00000134755:ENST00000251081:exon15:c.G2335A:p.G779R,ENSG00000134755:ENST00000280904:exon15:c.G2335A:p.G779R	rs139290300	D;S	0,00058
DSC2	18_28649081_C_T	ENSG00000134755:ENST00000251081:exon15:c.G2287A:p.A763T,ENSG00000134755:ENST00000280904:exon15:c.G2287A:p.A763T		N	0,00057
DSC2	18_28650748_A_C	ENSG00000134755:ENST00000251081:exon14:c.T2194G:p.L732V,ENSG00000134755:ENST00000280904:exon14:c.T2194G:p.L732V	rs151024019	D;P	0,00458
DSC2	18_28651640_C_T	ENSG00000134755:ENST00000251081:exon13:c.G2056A:p.G686S,ENSG00000134755:ENST00000280904:exon13:c.G2056A:p.G686S		N	0,00057
DSC2	18_28654750_G_A	ENSG00000134755:ENST00000251081:exon12:c.C1787T:p.A596V,ENSG00000134755:ENST00000280904:exon12:c.C1787T:p.A596V	rs148185335	P	0,00115
DSC2	18_28654874_C_A	ENST00000251081:exon13:c.1664-1G>T,ENST00000280904:exon13:c.1664-1G>T		L	0,00058
DSC2	18_28662896_G_A	ENSG00000134755:ENST00000251081:exon8:c.C1073T:p.T358I,ENSG00000134755:ENST00000280904:exon8:c.C1073T:p.T358I	rs139399951	D	0,00057
DSC2	18_28666574_C_T	ENSG00000134755:ENST00000251081:exon7:c.G907A:p.V303M,ENSG00000134755:ENST00000280904:exon7:c.G907A:p.V303M	rs145560678	D;P	0,00229
DSC2	18_28666624_C_A	ENSG00000134755:ENST00000251081:exon7:c.G857T:p.G286V,ENSG00000134755:ENST00000280904:exon7:c.G857T:p.G286V	rs199682735	D	0,00057

DSC2	18_28666627_A_G	ENSG00000134755:ENST00000251081:exon7:c.T854C:p.I285T,ENSG00000134755:ENST00000280904:exon7:c.T854C:p.I285T	rs199918720	D	0,00057
DSC2	18_28666657_G_A	ENSG00000134755:ENST00000251081:exon7:c.C824T:p.T275M,ENSG00000134755:ENST00000280904:exon7:c.C824T:p.T275M		P	0,00057
DSC2	18_28671095_G_A	ENSG00000134755:ENST00000251081:exon4:c.C370T:p.H124Y,ENSG00000134755:ENST00000280904:exon4:c.C370T:p.H124Y		N	0,00057
DSC2	18_28672152_G_A	ENSG00000134755:ENST00000251081:exon3:c.C266T:p.S89L,ENSG00000134755:ENST00000280904:exon3:c.C266T:p.S89L	rs141379407	D;P	0,00057
DSC2	18_28672246_A_C	ENSG00000134755:ENST00000251081:exon3:c.T172G:p.F58V,ENSG00000134755:ENST00000280904:exon3:c.T172G:p.F58V	rs138749562	D;P	0,00057
DSC2	18_28681901_C_T	ENSG00000134755:ENST00000251081:exon1:c.G34A:p.G12R,ENSG00000134755:ENST00000280904:exon1:c.G34A:p.G12R		N	0,00127
DSC2	18_28681932_C_A	ENSG00000134755:ENST00000251081:exon1:c.G3T:p.M1I,ENSG00000134755:ENST00000280904:exon1:c.G3T:p.M1I		S	0,00153
DSG2	18_29099805_C_A	ENSG00000046604:ENST00000261590:exon3:c.C121A:p.H41N,ENSG00000046604:ENST00000585206:exon3:c.C121A:p.H41N	rs201499704	D;P	0,00114
DSG2	18_29101120_G_A	ENSG00000046604:ENST00000261590:exon5:c.G437A:p.R146H,ENSG00000046604:ENST00000585206:exon5:c.G437A:p.R146H		P	0,00058
DSG2	18_29102067_A_G	ENSG00000046604:ENST00000261590:exon6:c.A545G:p.N182S,ENSG00000046604:ENST00000585206:exon6:c.A545G:p.N182S		N	0,00057
DSG2	18_29104717_A_G	ENSG00000046604:ENST00000261590:exon8:c.A880G:p.K294E		P	0,00057
DSG2	18_29104798_T_A	ENSG00000046604:ENST00000261590:exon8:c.T961A:p.F321I	rs201040643	D;P	0,00057
DSG2	18_29104840_A_G	ENSG00000046604:ENST00000261590:exon8:c.A1003G:p.T335A	rs191564916	D;P	0,00057
DSG2	18_29110986_-_C	ENSG00000046604:ENST00000261590:exon9:c.1051_1052insC:p.S351fs		L	0,00057
DSG2	18_29110986_A_C	ENSG00000046604:ENST00000261590:exon9:c.A1051C:p.S351R		N	0,00057
DSG2	18_29115255_G_A	ENSG00000046604:ENST00000261590:exon10:c.G1303A:p.D435N		P	0,00057
DSG2	18_29115328_A_G	ENSG00000046604:ENST00000261590:exon10:c.A1376G:p.Y459C		S	0,00057
DSG2	18_29122591_A_G	ENSG00000046604:ENST00000261590:exon14:c.A2110G:p.I704V	rs141388237	D	0,00174
DSG2	18_29122696_A_T	ENSG00000046604:ENST00000261590:exon14:c.A2215T:p.I739F		N	0,00059
DSG2	18_29126099_C_T	ENSG00000046604:ENST00000261590:exon15:c.C2750T:p.A917V		P	0,00057
DSG2	18_29126255_C_T	ENSG00000046604:ENST00000261590:exon15:c.C2906T:p.A969V		S	0,00057
DSG2	18_29126389_G_A	ENSG00000046604:ENST00000261590:exon15:c.G3040A:p.V1014I	rs200830807	D;P	0,00057

DSG2	18_29126426_G_A	ENSG00000046604:ENST00000261590:exon15:c.G3077A:p.S1026N		S	0,00057
DSG2	18_29126458_A_G	ENSG00000046604:ENST00000261590:exon15:c.A3109G:p.N1037D		N	0,00057
DSG2	18_29126475_G_C	ENSG00000046604:ENST00000261590:exon15:c.G3126C:p.Q1042H		S	0,00057
DSG2	18_29126516_C_T	ENSG00000046604:ENST00000261590:exon15:c.C3167T:p.T1056I		P	0,00057
DSP	6_7562969_A_G	ENSG00000096696:ENST00000379802:exon5:c.A682G:p.I228V,ENSG00000096696:ENST00000418664:exon5:c.A682G:p.I228V		N	0,00058
DSP	6_7562975_G_A	ENSG00000096696:ENST00000379802:exon5:c.G688A:p.D230N,ENSG00000096696:ENST00000418664:exon5:c.G688A:p.D230N	rs147315869	D;P	0,00172
DSP	6_7564006_A_T	ENSG00000096696:ENST00000379802:exon6:c.A764T:p.Y255F,ENSG00000096696:ENST00000418664:exon6:c.A764T:p.Y255F		S	0,00057
DSP	6_7565758_G_T	ENST00000379802:exon7:c.939+5G>T,ENST00000418664:exon7:c.939+5G>T		L	0,00058
DSP	6_7569444_G_A	ENSG00000096696:ENST00000379802:exon12:c.G1445A:p.C482Y,ENSG00000096696:ENST00000418664:exon12:c.G1445A:p.C482Y		S	0,00058
DSP	6_7569482_G_A	ENSG00000096696:ENST00000379802:exon12:c.G1483A:p.V495M,ENSG00000096696:ENST00000418664:exon12:c.G1483A:p.V495M		N	0,00058
DSP	6_7570791_G_A	ENSG00000096696:ENST00000379802:exon13:c.G1696A:p.A566T,ENSG00000096696:ENST00000418664:exon13:c.G1696A:p.A566T	rs148147581	D;P	0,00059
DSP	6_7571692_A_G	ENSG00000096696:ENST00000379802:exon14:c.A1778G:p.N593S,ENSG00000096696:ENST00000418664:exon14:c.A1778G:p.N593S	rs34239595	D	0,00057
DSP	6_7574937_A_G	ENSG00000096696:ENST00000379802:exon17:c.A2345G:p.D782G,ENSG00000096696:ENST00000418664:exon17:c.A2345G:p.D782G		S	0,00057
DSP	6_7575014_C_T	ENSG00000096696:ENST00000379802:exon17:c.C2422T:p.R808C,ENSG00000096696:ENST00000418664:exon17:c.C2422T:p.R808C	rs150339369	D;S	0,00172
DSP	6_7576669_C_T	ENSG00000096696:ENST00000379802:exon19:c.C2773T:p.R925W,ENSG00000096696:ENST00000418664:exon19:c.C2773T:p.R925W	rs145933612	D;S	0,00057
DSP	6_7578770_T_C	ENSG00000096696:ENST00000379802:exon22:c.T3059C:p.M1020T,ENSG00000096696:ENST00000418664:exon22:c.T3059C:p.M1020T		N	0,00057
DSP	6_7580129_A_G	ENSG00000096696:ENST00000379802:exon23:c.A3706G:p.R1236G		S	0,00115
DSP	6_7580528_G_A	ENSG00000096696:ENST00000379802:exon23:c.G4105A:p.E1369K	rs141805096	D	0,00058
DSP	6_7580810_G_A	ENSG00000096696:ENST00000379802:exon23:c.G4387A:p.V1463I		N	0,00058
DSP	6_7580912_C_T	ENSG00000096696:ENST00000379802:exon23:c.C4489T:p.R1497W	rs148041814	D	0,00057
DSP	6_7580913_G_A	ENSG00000096696:ENST00000379802:exon23:c.G4490A:p.R1497Q		N	0,00057

DSP	6_7580981_A_T	ENSG00000096696:ENST00000379802:exon23:c.A4558T:p.S1520C		S	0,00058
DSP	6_7581198_A_G	ENSG00000096696:ENST00000379802:exon23:c.A4775G:p.K1592R	rs200421954	D;P	0,00120
DSP	6_7581420_G_A	ENSG00000096696:ENST00000379802:exon23:c.G4997A:p.R1666Q		N	0,00058
DSP	6_7581543_A_G	ENSG00000096696:ENST00000379802:exon23:c.A5120G:p.Q1707R		S	0,00057
DSP	6_7581601_C_A	ENSG00000096696:ENST00000379802:exon23:c.C5178A:p.N1726K	rs147415451	D;P	0,00057
DSP	6_7581644_G_A	ENSG00000096696:ENST00000379802:exon23:c.G5221A:p.A1741T		N	0,00057
DSP	6_7581786_A_G	ENSG00000096696:ENST00000379802:exon23:c.A5363G:p.Q1788R	rs139673146	D	0,00057
DSP	6_7583299_A_G	ENSG00000096696:ENST00000379802:exon24:c.A5804G:p.Y1935C,ENSG00000096696:ENST00000418664:exon24:c.A4007G:p.Y1336C		N	0,00058
DSP	6_7583347_G_A	ENSG00000096696:ENST00000379802:exon24:c.G5852A:p.R1951Q,ENSG00000096696:ENST00000418664:exon24:c.G4055A:p.R1352Q		N	0,00058
DSP	6_7583376_G_A	ENSG00000096696:ENST00000379802:exon24:c.G5881A:p.V1961I,ENSG00000096696:ENST00000418664:exon24:c.G4084A:p.V1362I		N	0,00058
DSP	6_7584376_C_G	ENSG00000096696:ENST00000379802:exon24:c.C6881G:p.A2294G,ENSG00000096696:ENST00000418664:exon24:c.C5084G:p.A1695G	rs147000526	D;P	0,00115
DSP	6_7584845_G_C	ENSG00000096696:ENST00000379802:exon24:c.G7350C:p.K2450N,ENSG00000096696:ENST00000418664:exon24:c.G5553C:p.K1851N		S	0,00057
DSP	6_7584922_A_T	ENSG00000096696:ENST00000379802:exon24:c.A7427T:p.E2476V,ENSG00000096696:ENST00000418664:exon24:c.A5630T:p.E1877V		S	0,00057
DSP	6_7585127_T_G	ENSG00000096696:ENST00000379802:exon24:c.T7632G:p.F2544L,ENSG00000096696:ENST00000418664:exon24:c.T5835G:p.F1945L		N	0,00057
DSP	6_7585233_G_T	ENSG00000096696:ENST00000379802:exon24:c.G7738T:p.D2580Y,ENSG00000096696:ENST00000418664:exon24:c.G5941T:p.D1981Y		N	0,00057
DSP	6_7585612_A_T	ENSG00000096696:ENST00000379802:exon24:c.A8117T:p.K2706M,ENSG00000096696:ENST00000418664:exon24:c.A6320T:p.K2107M		N	0,00059
DSP	6_7585686_T_C	ENSG00000096696:ENST00000379802:exon24:c.T8191C:p.Y2731H,ENSG00000096696:ENST00000418664:exon24:c.T6394C:p.Y2132H	rs201397978	D	0,00240
DSP	6_7585774_C_T	ENSG00000096696:ENST00000379802:exon24:c.C8279T:p.A2760V,ENSG00000096696:ENST00000418664:exon24:c.C6482T:p.A2161V		N	0,00058
DSP	6_7585921_C_T	ENSG00000096696:ENST00000379802:exon24:c.C8426T:p.S2809L,ENSG00000096696:ENST00000418664:exon24:c.C6629T:p.S2210L		N	0,00058
DSP	6_7585962_C_G	ENSG00000096696:ENST00000379802:exon24:c.C8467G:p.P2823A,ENSG00000096696:ENST00000418664:exon24:c.C6670G:p.P2224A	rs142717240	D;S	0,00115

DSP	6_7586019_C_T	ENSG00000096696:ENST00000379802:exon24:c.C8524T:p.R2842C,ENSG00000096696:ENST00000418664:exon24:c.C6727T:p.R2243C	rs144850908	D	0,00058
JUP	17_39912055_C_T	ENSG00000173801:ENST00000310706:exon14:c.G2179A:p.D727N,ENSG00000173801:ENST00000393930:exon14:c.G2179A:p.D727N,ENSG00000173801:ENST00000393931:exon14:c.G2179A:p.D727N		N	0,00093
JUP	17_39912444_T_C	ENSG00000173801:ENST00000310706:exon13:c.A2069G:p.N690S,ENSG00000173801:ENST00000393930:exon13:c.A2069G:p.N690S,ENSG00000173801:ENST00000393931:exon13:c.A2069G:p.N690S	rs147628503	D	0,00070
JUP	17_39919360_C_T	ENSG00000173801:ENST00000310706:exon8:c.G1372A:p.A458T,ENSG00000173801:ENST00000393930:exon8:c.G1372A:p.A458T,ENSG00000173801:ENST00000393931:exon8:c.G1372A:p.A458T	rs139559495	D	0,00080
JUP	17_39919408_T_A	ENSG00000173801:ENST00000310706:exon8:c.A1324T:p.I442F,ENSG00000173801:ENST00000393930:exon8:c.A1324T:p.I442F,ENSG00000173801:ENST00000393931:exon8:c.A1324T:p.I442F	rs142213474	D;S	0,00064
JUP	17_39921027_G_C	ENSG00000173801:ENST00000310706:exon7:c.C1096G:p.P366A,ENSG00000173801:ENST00000393930:exon7:c.C1096G:p.P366A,ENSG00000173801:ENST00000393931:exon7:c.C1096G:p.P366A		N	0,00070
JUP	17_39925401_C_T	ENSG00000173801:ENST00000540235:exon3:c.G527A:p.R176Q,ENSG00000173801:ENST00000310706:exon4:c.G527A:p.R176Q,ENSG00000173801:ENST00000393930:exon4:c.G527A:p.R176Q,ENSG00000173801:ENST00000393931:exon4:c.G527A:p.R176Q,ENSG00000173801:ENST00000420370:exon4:c.G527A:p.R176Q,ENSG00000173801:ENST00000437187:exon4:c.G527A:p.R176Q,ENSG00000173801:ENST00000449889:exon4:c.G527A:p.R176Q,ENSG00000173801:ENST00000424457:exon5:c.G527A:p.R176Q	rs144171604	D	0,00107
JUP	17_39925704_G_C	ENSG00000173801:ENST00000540235:exon2:c.C434G:p.P145R,ENSG00000173801:ENST00000310706:exon3:c.C434G:p.P145R,ENSG00000173801:ENST00000393930:exon3:c.C434G:p.P145R,ENSG00000173801:ENST00000393931:exon3:c.C434G:p.P145R,ENSG00000173801:ENST00000420370:exon3:c.C434G:p.P145R,ENSG00000173801:ENST00000437187:exon3:c.C434G:p.P145R,ENSG00000173801:ENST00000449889:exon3:c.C434G:p.P145R,ENSG00000173801:ENST00000424457:exon4:c.C434G:p.P145R		S	0,00085
KCNE1	21_35821619_G_A	ENSG00000180509:ENST00000416357:exon2:c.C314T:p.S105L,ENSG00000180509:ENST00000337385:exon3:c.C314T:p.S105L,ENSG00000180509:ENST00000399284:exon3:c.C314T:p.S105L,ENSG00000180509:ENST00000399289:exon3:c.C314T:p.S105L,ENSG00000180509:ENST00000432085:exon3:c.C314T:p.S105L,ENSG00000180509:ENST00000399286:exon4:c.C314T:p.S105L		N	0,00057
KCNE1	21_35821827_G_A	ENSG00000180509:ENST00000416357:exon2:c.C106T:p.R36C,ENSG00000180509:ENST00000337385:exon3:c.C106T:p.R36C,ENSG00000180509:ENST00000399284:exon3:c.C106T:p.R36C,ENSG00000180509:ENST00000399289:exon3:c.C106T:p.R36C,ENSG00000180509:ENST00000432085:exon3:c.C106T:p.R36C,ENSG00000180509:ENST00000399286:exon4:c.C106T:p.R36C		N	0,00058

KCNE1	21_35821850_G_A	ENSG00000180509:ENST00000416357:exon2:c.C83T:p.S28L,ENSG00000180509:ENST00000337385:exon3:c.C83T:p.S28L,ENSG00000180509:ENST00000399284:exon3:c.C83T:p.S28L,ENSG00000180509:ENST00000399289:exon3:c.C83T:p.S28L,ENSG00000180509:ENST00000432085:exon3:c.C83T:p.S28L,ENSG00000180509:ENST00000399286:exon4:c.C83T:p.S28L	rs199473350	D;P	0,00058
KCNE1	21_35821916_G_T	ENSG00000180509:ENST00000416357:exon2:c.C17A:p.T6N,ENSG00000180509:ENST00000337385:exon3:c.C17A:p.T6N,ENSG00000180509:ENST00000399284:exon3:c.C17A:p.T6N,ENSG00000180509:ENST00000399289:exon3:c.C17A:p.T6N,ENSG00000180509:ENST00000432085:exon3:c.C17A:p.T6N,ENSG00000180509:ENST00000399286:exon4:c.C17A:p.T6N		N	0,00058
KCNE2	21_35736455_G_A			L	0,00057
KCNE2	21_35742806_C_T	ENSG00000159197:ENST00000290310:exon2:c.C29T:p.T10M	rs199473648	D;P	0,00057
KCNE2	21_35742817_G_A	ENSG00000159197:ENST00000290310:exon2:c.G40A:p.V14I	rs142153692	D;P	0,00115
KCNE2	21_35742938_T_C	ENSG00000159197:ENST00000290310:exon2:c.T161C:p.M54T	rs74315447	D;P	0,00057
KCNE2	21_35742947_T_C	ENSG00000159197:ENST00000290310:exon2:c.T170C:p.I57T	rs74315448	D;P	0,00057
KCNE2	21_35743134_C_A	ENSG00000159197:ENST00000290310:exon2:c.C357A:p.F119L	rs139202426	D	0,00058
KCNH2	7_150642517_G_A	ENSG00000055118:ENST00000330883:exon11:c.C2396T:p.P799L,ENSG00000055118:ENST00000392968:exon13:c.C3128T:p.P1043L,ENSG00000055118:ENST00000262186:exon15:c.C3416T:p.P1139L		N	0,00093
KCNH2	7_150644131_C_T	ENSG00000055118:ENST00000330883:exon10:c.G2144A:p.R715Q,ENSG00000055118:ENST00000392968:exon12:c.G2876A:p.R959Q,ENSG00000055118:ENST00000262186:exon14:c.G3164A:p.R1055Q	rs41307270	D;S	0,00063
KCNH2	7_150644718_T_C	ENSG00000055118:ENST00000330883:exon8:c.A1921G:p.S641G,ENSG00000055118:ENST00000392968:exon10:c.A2653G:p.S885G,ENSG00000055118:ENST00000262186:exon12:c.A2941G:p.S981G	rs76649554	D	0,00111
KCNH2	7_150645607_C_T	ENSG00000055118:ENST00000330883:exon7:c.G1597A:p.G533S,ENSG00000055118:ENST00000392968:exon9:c.G2329A:p.G777S,ENSG00000055118:ENST00000262186:exon11:c.G2617A:p.G873S	rs41314354	D	0,00117
KCNH2	7_150647078_G_A	ENSG00000055118:ENST00000430723:exon9:c.C2576T:p.T859M	rs41314366	D;S	0,00059
KCNH2	7_150647144_C_A	ENSG00000055118:ENST00000430723:exon9:c.G2510T:p.G837V		N	0,00060
KCNH2	7_150647353_GT_-	ENSG00000055118:ENST00000330883:exon5:c.1280_1281del:p.427_427del,ENSG00000055118:ENST00000392968:exon7:c.2012_2013del:p.671_671del,ENSG00000055118:ENST00000262186:exon9:c.2300_2301del:p.767_767del,ENSG00000055118:ENST00000430723:exon9:c.2300_2301del:p.767_767del		N	0,00073
KCNH2	7_150654468_G_A	ENSG00000055118:ENST00000392968:exon3:c.C751T:p.P251S,ENSG00000055118:ENST00000262186:exon5:c.C1039T:p.P347S,ENSG00000055118:ENST00000430723:exon5:c.C1039T:p.P347S	rs138776684	D;P	0,00118
KCNH2	7_150655425_T_C	ENSG00000055118:ENST00000392968:exon2:c.A350G:p.D117G,ENSG00000055118:ENST00000262186:exon4:c.A638G:p.D213G,ENSG00000055118:ENST00000430723:exon4:c.A638G:p.D213G		N	0,00148

KCNH2	7_150655495_CGCCCGCGC_-	ENSG00000055118:ENST00000392968:exon2:c.272_280del:p.91_94del,ENSG00000055118:ENST00000262186:exon4:c.560_568del:p.187_190del,ENSG00000055118:ENST00000430723:exon4:c.560_568del:p.187_190del		P	0,01220
KCNH2	7_150655510_CGCCGCCCG_-	ENSG00000055118:ENST00000392968:exon2:c.257_265del:p.86_89del,ENSG00000055118:ENST00000262186:exon4:c.545_553del:p.182_185del,ENSG00000055118:ENST00000430723:exon4:c.545_553del:p.182_185del		N	0,01124
KCNH2	7_150655521_C_T	ENSG00000055118:ENST00000392968:exon2:c.G254A:p.R85Q,ENSG00000055118:ENST00000262186:exon4:c.G542A:p.R181Q,ENSG00000055118:ENST00000430723:exon4:c.G542A:p.R181Q	rs41308954	D	0,01099
KCNH2	7_150655537_G_A	ENSG00000055118:ENST00000392968:exon2:c.C238T:p.R80W,ENSG00000055118:ENST00000262186:exon4:c.C526T:p.R176W,ENSG00000055118:ENST00000430723:exon4:c.C526T:p.R176W	rs36210422	D;S	0,00543
KCNH2	7_150656690_G_A	ENSG00000055118:ENST00000392968:exon1:c.C154T:p.R52W,ENSG00000055118:ENST00000262186:exon3:c.C442T:p.R148W,ENSG00000055118:ENST00000430723:exon3:c.C442T:p.R148W	rs139544114	D	0,00140
KCNH2	7_150674983_G_A	ENSG00000055118:ENST00000262186:exon1:c.C19T:p.H7Y,ENSG00000055118:ENST00000430723:exon1:c.C19T:p.H7Y		S	0,00175
KCNJ2	17_68172133_A_G	ENSG00000123700:ENST00000243457:exon2:c.A953G:p.N318S,ENSG00000123700:ENST00000535240:exon2:c.A953G:p.N318S		N	0,00057
KCNQ1	11_2466480_-_CGCGCCCAT	ENSG00000053918:ENST00000155840:exon1:c.152_153insCGCGCCCAT:p.Y51delinsYAPI		N	0,01376
KCNQ1	11_2466545_C_A	ENSG00000053918:ENST00000155840:exon1:c.C217A:p.P73T	rs199472676	D;P	0,00110
KCNQ1	11_2542757_C_T	ENSG00000053918:ENST00000380776:exon2:c.C133T:p.R45C	rs80269976	D	0,00058
KCNQ1	11_2542793_G_A	ENSG00000053918:ENST00000380776:exon2:c.G169A:p.D57N	rs10400212	D	0,00059
KCNQ1	11_2549229_C_T	ENSG00000053918:ENST00000155840:exon2:c.C458T:p.T153M,ENSG00000053918:ENST00000335475:exon2:c.C77T:p.T26M,ENSG00000053918:ENST00000496887:exon3:c.C197T:p.T66M	rs143709408	D;P	0,00068
KCNQ1	11_2591921_C_T	ENSG00000053918:ENST00000155840:exon3:c.C541T:p.R181C,ENSG00000053918:ENST00000335475:exon3:c.C160T:p.R54C,ENSG00000053918:ENST00000496887:exon4:c.C280T:p.R94C	rs199473395	D;P	0,00094
KCNQ1	11_2594092_T_G	ENSG00000053918:ENST00000155840:exon6:c.T797G:p.L266R,ENSG00000053918:ENST00000335475:exon6:c.T416G:p.L139R,ENSG00000053918:ENST00000496887:exon7:c.T536G:p.L179R		S	0,00062
KCNQ1	11_2604687_A_T	ENSG00000053918:ENST00000155840:exon7:c.A944T:p.Y315F,ENSG00000053918:ENST00000335475:exon7:c.A563T:p.Y188F	rs74462309	D;P	0,00074
KCNQ1	11_2608850_G_T	ENSG00000053918:ENST00000155840:exon9:c.G1179T:p.K393N,ENSG00000053918:ENST00000335475:exon9:c.G798T:p.K266N	rs12720457	D	0,00158
KCNQ1	11_2790079_G_A	ENSG00000053918:ENST00000155840:exon12:c.G1520A:p.R507Q,ENSG00000053918:ENST00000335475:exon12:c.G1139A:p.R380Q		N	0,00058
KCNQ1	11_2797186_G_A	ENST00000155840:exon13:c.1591-4G>A,ENST00000335475:exon13:c.1210-4G>A		L	0,00085

KCNQ1	11_2869087_G_A	ENSG00000053918:ENST00000155840:exon16:c.G1885A:p.G629S,ENSG00000053918:ENST00000335475:exon16:c.G1504A:p.G502S		N	0,00097
KCNQ1	11_2869105_G_A	ENSG00000053918:ENST00000155840:exon16:c.G1903A:p.G635R,ENSG00000053918:ENST00000335475:exon16:c.G1522A:p.G508R	rs199473484	D;P	0,00097
KCNQ1	11_2869127_G_T	ENSG00000053918:ENST00000155840:exon16:c.G1925T:p.C642F,ENSG00000053918:ENST00000335475:exon16:c.G1544T:p.C515F		N	0,00096
LDB3	10_88428332_G_C			L	0,00070
LDB3	10_88439193_G_A	ENSG00000122367:ENST00000263066:exon2:c.G163A:p.V55I,ENSG00000122367:ENST00000310944:exon2:c.G163A:p.V55I,ENSG00000122367:ENST00000361373:exon2:c.G163A:p.V55I,ENSG00000122367:ENST00000372056:exon2:c.G163A:p.V55I,ENSG00000122367:ENST00000372066:exon2:c.G163A:p.V55I,ENSG00000122367:ENST00000542786:exon2:c.G163A:p.V55I,ENSG00000122367:ENST00000352360:exon3:c.G163A:p.V55I,ENSG00000122367:ENST00000429277:exon3:c.G163A:p.V55I,ENSG00000122367:ENST00000458213:exon3:c.G163A:p.V55I	rs3740343	D;P	0,00115
LDB3	10_88441437_C_T	ENSG00000122367:ENST00000310944:exon4:c.C566T:p.S189L,ENSG00000122367:ENST00000361373:exon4:c.C566T:p.S189L,ENSG00000122367:ENST00000372056:exon4:c.C566T:p.S189L,ENSG00000122367:ENST00000542786:exon4:c.C566T:p.S189L,ENSG00000122367:ENST00000429277:exon5:c.C566T:p.S189L	rs45487699	D;P	0,00072
LDB3	10_88441535_G_A	ENSG00000122367:ENST00000310944:exon4:c.G664A:p.A222T,ENSG00000122367:ENST00000361373:exon4:c.G664A:p.A222T,ENSG00000122367:ENST00000372056:exon4:c.G664A:p.A222T,ENSG00000122367:ENST00000542786:exon4:c.G664A:p.A222T,ENSG00000122367:ENST00000429277:exon5:c.G664A:p.A222T	rs139922045	D	0,00066
LDB3	10_88446940_G_C	ENSG00000122367:ENST00000263066:exon5:c.G459C:p.Q153H,ENSG00000122367:ENST00000372056:exon5:c.G804C:p.Q268H,ENSG00000122367:ENST00000372066:exon5:c.G459C:p.Q153H,ENSG00000122367:ENST00000429277:exon6:c.G804C:p.Q268H,ENSG00000122367:ENST00000458213:exon6:c.G459C:p.Q153H		N	0,00067
LDB3	10_88446992_G_A	ENSG00000122367:ENST00000263066:exon5:c.G511A:p.A171T,ENSG00000122367:ENST00000372056:exon5:c.G856A:p.A286T,ENSG00000122367:ENST00000372066:exon5:c.G511A:p.A171T,ENSG00000122367:ENST00000429277:exon6:c.G856A:p.A286T,ENSG00000122367:ENST00000458213:exon6:c.G511A:p.A171T		N	0,00066
LDB3	10_88466427_G_A	ENSG00000122367:ENST00000361373:exon7:c.G1036A:p.A346T	rs201968775	D	0,00094
LDB3	10_88466440_C_T	ENSG00000122367:ENST00000361373:exon7:c.C1049T:p.T350I	rs200796750	D;P	0,00092
LDB3	10_88469687_G_A	ENSG00000122367:ENST00000352360:exon5:c.G340A:p.A114T,ENSG00000122367:ENST00000263066:exon8:c.G781A:p.A261T,ENSG00000122367:ENST00000361373:exon8:c.G1111A:p.A371T,ENSG00000122367:ENST00000429277:exon9:c.G1126A:p.A376T,ENSG00000122367:ENST00000458213:exon9:c.G781A:p.A261T	rs45539535	D;P	0,00143

LDB3	10_88476312_G_A	ENSG00000122367:ENST00000352360:exon6:c.G689A:p.R230H,ENSG00000122367:ENST00000263066:exon9:c.G1130A:p.R377H,ENSG00000122367:ENST00000361373:exon9:c.G1460A:p.R487H,ENSG00000122367:ENST00000429277:exon10:c.G1475A:p.R492H,ENSG00000122367:ENST00000458213:exon10:c.G1130A:p.R377H	rs146265188	D;S	0,00088
LDB3	10_88476339_T_C	ENSG00000122367:ENST00000352360:exon6:c.T716C:p.F239S,ENSG00000122367:ENST00000263066:exon9:c.T1157C:p.F386S,ENSG00000122367:ENST00000361373:exon9:c.T1487C:p.F496S,ENSG00000122367:ENST00000429277:exon10:c.T1502C:p.F501S,ENSG00000122367:ENST00000458213:exon10:c.T1157C:p.F386S	rs147072071	D	0,00083
LDB3	10_88476446_G_C	ENSG00000122367:ENST00000352360:exon6:c.G823C:p.A275P,ENSG00000122367:ENST00000263066:exon9:c.G1264C:p.A422P,ENSG00000122367:ENST00000361373:exon9:c.G1594C:p.A532P,ENSG00000122367:ENST00000429277:exon10:c.G1609C:p.A537P,ENSG00000122367:ENST00000458213:exon10:c.G1264C:p.A422P	rs143764931	D;S	0,00258
LDB3	10_88476524_A_G	ENSG00000122367:ENST00000352360:exon6:c.A901G:p.I301V,ENSG00000122367:ENST00000263066:exon9:c.A1342G:p.I448V,ENSG00000122367:ENST00000361373:exon9:c.A1672G:p.I558V,ENSG00000122367:ENST00000429277:exon10:c.A1687G:p.I563V,ENSG00000122367:ENST00000458213:exon10:c.A1342G:p.I448V		P	0,00079
LMNA	1_156096679_G_A	ENSG00000160789:ENST00000392353:exon1:c.G86A:p.R29K,ENSG00000160789:ENST00000368297:exon2:c.G86A:p.R29K,ENSG00000160789:ENST00000515459:exon2:c.G86A:p.R29K		N	0,00123
LMNA	1_156096738_C_T			L	0,00067
LMNA	1_156105051_C_T	ENSG00000160789:ENST00000347559:exon5:c.C884T:p.S295L,ENSG00000160789:ENST00000361308:exon5:c.C884T:p.S295L,ENSG00000160789:ENST00000368299:exon5:c.C884T:p.S295L,ENSG00000160789:ENST00000368300:exon5:c.C884T:p.S295L,ENSG00000160789:ENST00000392353:exon5:c.C641T:p.S214L,ENSG00000160789:ENST00000448611:exon5:c.C548T:p.S183L,ENSG00000160789:ENST00000368297:exon6:c.C641T:p.S214L,ENSG00000160789:ENST00000473598:exon6:c.C587T:p.S196L,ENSG00000160789:ENST00000368301:exon8:c.C884T:p.S295L		N	0,00081
LMNA	1_156105062_A_G	ENSG00000160789:ENST00000347559:exon5:c.A895G:p.I299V,ENSG00000160789:ENST00000361308:exon5:c.A895G:p.I299V,ENSG00000160789:ENST00000368299:exon5:c.A895G:p.I299V,ENSG00000160789:ENST00000368300:exon5:c.A895G:p.I299V,ENSG00000160789:ENST00000392353:exon5:c.A652G:p.I218V,ENSG00000160789:ENST00000448611:exon5:c.A559G:p.I187V,ENSG00000160789:ENST00000368297:exon6:c.A652G:p.I218V,ENSG00000160789:ENST00000473598:exon6:c.A598G:p.I200V,ENSG00000160789:ENST00000368301:exon8:c.A895G:p.I299V	rs150924946	D	0,00083

LMNA	1_156105708_C_T	ENSG00000160789:ENST00000347559:exon6:c.C953T:p.A318V,ENSG00000160789:ENST00000361308:exon6:c.C953T:p.A318V,ENSG00000160789:ENST00000368299:exon6:c.C953T:p.A318V,ENSG00000160789:ENST00000368300:exon6:c.C953T:p.A318V,ENSG00000160789:ENST00000392353:exon6:c.C710T:p.A237V,ENSG00000160789:ENST00000448611:exon6:c.C617T:p.A206V,ENSG00000160789:ENST00000368297:exon7:c.C710T:p.A237V,ENSG00000160789:ENST00000473598:exon7:c.C656T:p.A219V,ENSG00000160789:ENST00000368301:exon9:c.C953T:p.A318V		N	0,00063
LMNA	1_156106078_G_T	ENSG00000160789:ENST00000508500:exon2:c.G109T:p.G37C,ENSG00000160789:ENST00000347559:exon7:c.G1231T:p.G411C,ENSG00000160789:ENST00000361308:exon7:c.G1231T:p.G411C,ENSG00000160789:ENST00000368299:exon7:c.G1231T:p.G411C,ENSG00000160789:ENST00000368300:exon7:c.G1231T:p.G411C,ENSG00000160789:ENST00000392353:exon7:c.G988T:p.G330C,ENSG00000160789:ENST00000448611:exon7:c.G895T:p.G299C,ENSG00000160789:ENST00000368297:exon8:c.G988T:p.G330C,ENSG00000160789:ENST00000473598:exon8:c.G934T:p.G312C,ENSG00000160789:ENST00000368301:exon10:c.G1231T:p.G411C		N	0,00076
LMNA	1_156108453_A_C	ENSG00000160789:ENST00000508500:exon5:c.A661C:p.S221R,ENSG00000160789:ENST00000347559:exon10:c.A1783C:p.S595R,ENSG00000160789:ENST00000368300:exon11:c.A1873C:p.S625R,ENSG00000160789:ENST00000448611:exon11:c.A1537C:p.S513R,ENSG00000160789:ENST00000473598:exon12:c.A1576C:p.S526R		N	0,00076
LMNA	1_156108454_G_C	ENSG00000160789:ENST00000508500:exon5:c.G662C:p.S221T,ENSG00000160789:ENST00000347559:exon10:c.G1784C:p.S595T,ENSG00000160789:ENST00000368300:exon11:c.G1874C:p.S625T,ENSG00000160789:ENST00000448611:exon11:c.G1538C:p.S513T,ENSG00000160789:ENST00000473598:exon12:c.G1577C:p.S526T		N	0,00076
LMNA	1_156108510_C_T	ENSG00000160789:ENST00000508500:exon5:c.C718T:p.R240C,ENSG00000160789:ENST00000347559:exon10:c.C1840T:p.R614C,ENSG00000160789:ENST00000368300:exon11:c.C1930T:p.R644C,ENSG00000160789:ENST00000448611:exon11:c.C1594T:p.R532C,ENSG00000160789:ENST00000473598:exon12:c.C1633T:p.R545C	rs142000963	D;S	0,00151
MYBPC3	11_47353626_G_A	ENSG00000134571:ENST00000256993:exon31:c.C3808T:p.R1270X,ENSG00000134571:ENST00000399249:exon32:c.C3811T:p.R1271X,ENSG00000134571:ENST00000545968:exon33:c.C3811T:p.R1271X		P	0,00061
MYBPC3	11_47353639_G_C	ENSG00000134571:ENST00000256993:exon31:c.C3795G:p.C1265W,ENSG00000134571:ENST00000399249:exon32:c.C3798G:p.C1266W,ENSG00000134571:ENST00000545968:exon33:c.C3798G:p.C1266W		S	0,00060
MYBPC3	11_47353666_G_T	ENSG00000134571:ENST00000256993:exon31:c.C3768A:p.N1256K,ENSG00000134571:ENST00000399249:exon32:c.C3771A:p.N1257K,ENSG00000134571:ENST00000545968:exon33:c.C3771A:p.N1257K		S	0,00237
MYBPC3	11_47353674_C_T	ENSG00000134571:ENST00000256993:exon31:c.G3760A:p.A1254T,ENSG00000134571:ENST00000399249:exon32:c.G3763A:p.A1255T,ENSG00000134571:ENST00000545968:exon33:c.G3763A:p.A1255T		P	0,00117

MYBPC3	11_47353685_T_C	ENSG00000134571:ENST00000256993:exon31:c.A3749G:p.Y1250C,ENSG00000134571:ENST00000399249:exon32:c.A3752G:p.Y1251C,ENSG00000134571:ENST00000545968:exon33:c.A3752G:p.Y1251C		P	0,00059
MYBPC3	11_47353686_A_G	ENSG00000134571:ENST00000256993:exon31:c.T3748C:p.Y1250H,ENSG00000134571:ENST00000399249:exon32:c.T3751C:p.Y1251H,ENSG00000134571:ENST00000545968:exon33:c.T3751C:p.Y1251H		S	0,00058
MYBPC3	11_47353709_G_C	ENSG00000134571:ENST00000256993:exon31:c.C3725G:p.P1242R,ENSG00000134571:ENST00000399249:exon32:c.C3728G:p.P1243R,ENSG00000134571:ENST00000545968:exon33:c.C3728G:p.P1243R		S	0,00058
MYBPC3	11_47353740_G_A	ENSG00000134571:ENST00000256993:exon31:c.C3694T:p.Q1232X,ENSG00000134571:ENST00000399249:exon32:c.C3697T:p.Q1233X,ENSG00000134571:ENST00000545968:exon33:c.C3697T:p.Q1233X		P	0,00058
MYBPC3	11_47353755_G_A	ENSG00000134571:ENST00000256993:exon31:c.C3679T:p.R1227C,ENSG00000134571:ENST00000399249:exon32:c.C3682T:p.R1228C,ENSG00000134571:ENST00000545968:exon33:c.C3682T:p.R1228C	rs201312636	D;S	0,00057
MYBPC3	11_47354116_C_T	ENST00000256993:exon31:c.3624+1G>A,ENST00000399249:exon32:c.3627+1G>A,ENST00000545968:exon33:c.3627+1G>A		P	0,00059
MYBPC3	11_47354123_G_-	ENSG00000134571:ENST00000256993:exon30:c.3618delC:p.S1206fs,ENSG00000134571:ENST00000399249:exon31:c.3621delC:p.S1207fs,ENSG00000134571:ENST00000545968:exon32:c.3621delC:p.S1207fs		L	0,00059
MYBPC3	11_47354139_CAGCAGAGCA_-	ENSG00000134571:ENST00000256993:exon30:c.3593_3602del:p.1198_1201del,ENSG00000134571:ENST00000399249:exon31:c.3596_3605del:p.1199_1202del,ENSG00000134571:ENST00000545968:exon32:c.3596_3605del:p.1199_1202del		L	0,00059
MYBPC3	11_47354151_GCAGTGTAGCCC_-	ENSG00000134571:ENST00000256993:exon30:c.3579_3590del:p.1193_1197del,ENSG00000134571:ENST00000399249:exon31:c.3582_3593del:p.1194_1198del,ENSG00000134571:ENST00000545968:exon32:c.3582_3593del:p.1194_1198del		N	0,00118
MYBPC3	11_47354172_G_A	ENSG00000134571:ENST00000256993:exon30:c.C3569T:p.S1190L,ENSG00000134571:ENST00000399249:exon31:c.C3572T:p.S1191L,ENSG00000134571:ENST00000545968:exon32:c.C3572T:p.S1191L		P	0,00060
MYBPC3	11_47354209_C_T	ENSG00000134571:ENST00000256993:exon30:c.G3532A:p.E1178K,ENSG00000134571:ENST00000399249:exon31:c.G3535A:p.E1179K,ENSG00000134571:ENST00000545968:exon32:c.G3535A:p.E1179K	rs199669878	D;P	0,00059
MYBPC3	11_47354256_G_C	ENST00000256993:exon31:c.3488-3C>G,ENST00000399249:exon32:c.3491-3C>G,ENST00000545968:exon33:c.3491-3C>G		L	0,00059
MYBPC3	11_47354389_CCTTGGTGGTGG_-	ENSG00000134571:ENST00000256993:exon29:c.3452_3463del:p.1151_1155del,ENSG00000134571:ENST00000399249:exon30:c.3455_3466del:p.1152_1156del,ENSG00000134571:ENST00000545968:exon31:c.3455_3466del:p.1152_1156del		N	0,00059

MYBPC3	11_47354451_TAG_-	ENSG00000134571:ENST00000256993:exon29:c.3399_3401del:p.1133_1134del,ENSG00000134571:ENST00000399249:exon30:c.3402_3404del:p.1134_1135del,ENSG00000134571:ENST00000545968:exon31:c.3402_3404del:p.1134_1135del		N	0,00059
MYBPC3	11_47354471_C_G	ENSG00000134571:ENST00000256993:exon29:c.G3381C:p.E1127D,ENSG00000134571:ENST00000399249:exon30:c.G3384C:p.E1128D,ENSG00000134571:ENST00000545968:exon31:c.G3384C:p.E1128D		N	0,00059
MYBPC3	11_47354522_-_CAC	ENSG00000134571:ENST00000256993:exon29:c.3330_3331insGTG:p.E1110delinsEW,ENSG00000134571:ENST00000399249:exon30:c.3333_3334insGTG:p.E1111delinsEW,ENSG00000134571:ENST00000545968:exon31:c.3333_3334insGTG:p.E1111delinsEW		N	0,00062
MYBPC3	11_47354740_C_G	ENST00000256993:exon29:c.3327+5G>C,ENST00000399249:exon30:c.3330+5G>C,ENST00000545968:exon31:c.3330+5G>C		P	0,00348
MYBPC3	11_47354749_G_A	ENSG00000134571:ENST00000256993:exon28:c.C3323T:p.T1108I,ENSG00000134571:ENST00000399249:exon29:c.C3326T:p.T1109I,ENSG00000134571:ENST00000545968:exon30:c.C3326T:p.T1109I		P	0,00138
MYBPC3	11_47354781_C_T	ENSG00000134571:ENST00000256993:exon28:c.G3291A:p.W1097X,ENSG00000134571:ENST00000399249:exon29:c.G3294A:p.W1098X,ENSG00000134571:ENST00000545968:exon30:c.G3294A:p.W1098X		P	0,00071
MYBPC3	11_47354818_C_T	ENSG00000134571:ENST00000256993:exon28:c.G3254A:p.W1085X,ENSG00000134571:ENST00000399249:exon29:c.G3257A:p.W1086X,ENSG00000134571:ENST00000545968:exon30:c.G3257A:p.W1086X		L	0,00075
MYBPC3	11_47354848_-_A	ENSG00000134571:ENST00000256993:exon28:c.3224_3225insT:p.D1075fs,ENSG00000134571:ENST00000399249:exon29:c.3227_3228insT:p.D1076fs,ENSG00000134571:ENST00000545968:exon30:c.3227_3228insT:p.D1076fs		P	0,00378
MYBPC3	11_47355103_C_T	ENST00000256993:exon28:c.3187+5G>A,ENST00000399249:exon29:c.3190+5G>A,ENST00000545968:exon30:c.3190+5G>A		P	0,00134
MYBPC3	11_47355191_C_T	ENSG00000134571:ENST00000256993:exon27:c.G3104A:p.R1035H,ENSG00000134571:ENST00000399249:exon28:c.G3107A:p.R1036H,ENSG00000134571:ENST00000545968:exon29:c.G3107A:p.R1036H		N	0,00062
MYBPC3	11_47355233_C_G	ENSG00000134571:ENST00000256993:exon27:c.G3062C:p.R1021P,ENSG00000134571:ENST00000399249:exon28:c.G3065C:p.R1022P,ENSG00000134571:ENST00000545968:exon29:c.G3065C:p.R1022P		P	0,00063
MYBPC3	11_47355249_C_T	ENSG00000134571:ENST00000256993:exon27:c.G3046A:p.E1016K,ENSG00000134571:ENST00000399249:exon28:c.G3049A:p.E1017K,ENSG00000134571:ENST00000545968:exon29:c.G3049A:p.E1017K		P	0,00064
MYBPC3	11_47355294_G_A	ENSG00000134571:ENST00000256993:exon27:c.C3001T:p.R1001W,ENSG00000134571:ENST00000399249:exon28:c.C3004T:p.R1002W,ENSG00000134571:ENST00000545968:exon29:c.C3004T:p.R1002W	rs3729799	D	0,00065

MYBPC3	11_47355304_C_T	ENST00000256993:exon28:c.2992-1G>A,ENST00000399249:exon29:c.2995-1G>A,ENST00000545968:exon30:c.2995-1G>A		L	0,00066
MYBPC3	11_47355514_T_A	ENSG00000134571:ENST00000256993:exon26:c.A2950T:p.K984X,ENSG00000134571:ENST00000399249:exon27:c.A2953T:p.K985X,ENSG00000134571:ENST00000545968:exon28:c.A2953T:p.K985X		L	0,00068
MYBPC3	11_47356588_C_A	ENST00000256993:exon26:c.2902+5G>T,ENST00000399249:exon27:c.2905+5G>T,ENST00000545968:exon28:c.2905+5G>T		L	0,00074
MYBPC3	11_47356592_C_T	ENST00000256993:exon26:c.2902+1G>A,ENST00000399249:exon27:c.2905+1G>A,ENST00000545968:exon28:c.2905+1G>A		P	0,00147
MYBPC3	11_47356633_AG_-	ENSG00000134571:ENST00000256993:exon25:c.2861_2862del:p.954_954del,ENSG00000134571:ENST00000399249:exon26:c.2864_2865del:p.955_955del,ENSG00000134571:ENST00000545968:exon27:c.2864_2865del:p.955_955del		L	0,00156
MYBPC3	11_47356671_G_A	ENSG00000134571:ENST00000256993:exon25:c.C2824T:p.R942X,ENSG00000134571:ENST00000399249:exon26:c.C2827T:p.R943X,ENSG00000134571:ENST00000545968:exon27:c.C2827T:p.R943X		P	0,00312
MYBPC3	11_47356710_G_C	ENSG00000134571:ENST00000256993:exon25:c.C2785G:p.L929V,ENSG00000134571:ENST00000399249:exon26:c.C2788G:p.L930V,ENSG00000134571:ENST00000545968:exon27:c.C2788G:p.L930V		N	0,00082
MYBPC3	11_47357425_CA_-			P	0,00061
MYBPC3	11_47357437_G_T	ENSG00000134571:ENST00000256993:exon24:c.C2725A:p.P909T,ENSG00000134571:ENST00000399249:exon25:c.C2728A:p.P910T,ENSG00000134571:ENST00000545968:exon26:c.C2728A:p.P910T		P	0,00060
MYBPC3	11_47357547_G_T	ENSG00000134571:ENST00000256993:exon24:c.C2615A:p.P872H,ENSG00000134571:ENST00000399249:exon25:c.C2618A:p.P873H,ENSG00000134571:ENST00000545968:exon26:c.C2618A:p.P873H		P	0,00063
MYBPC3	11_47357555_G_-	ENSG00000134571:ENST00000256993:exon24:c.2607delC:p.P869fs,ENSG00000134571:ENST00000399249:exon25:c.2610delC:p.P870fs,ENSG00000134571:ENST00000545968:exon26:c.2610delC:p.P870fs		P	0,00252
MYBPC3	11_47357560_G_T	ENSG00000134571:ENST00000256993:exon24:c.C2602A:p.P868T,ENSG00000134571:ENST00000399249:exon25:c.C2605A:p.P869T,ENSG00000134571:ENST00000545968:exon26:c.C2605A:p.P869T		P	0,00188
MYBPC3	11_47357560_GA_GGA	ENSG00000134571:ENST00000256993:exon24:c.2601_2602TCC,ENSG00000134571:ENST00000399249:exon25:c.2604_2605TCC,ENSG00000134571:ENST00000545968:exon26:c.2604_2605TCC		N	0,00063
MYBPC3	11_47357561_A_-	ENSG00000134571:ENST00000256993:exon24:c.2601delT:p.G867fs,ENSG00000134571:ENST00000399249:exon25:c.2604delT:p.G868fs,ENSG00000134571:ENST00000545968:exon26:c.2604delT:p.G868fs		P	0,00253
MYBPC3	11_47357563_C_G	ENST00000256993:exon25:c.2600-1G>C,ENST00000399249:exon26:c.2603-1G>C,ENST00000545968:exon27:c.2603-1G>C,ENST00000544791:exon27:c.2535-1G>C		L	0,00062
MYBPC3	11_47357564_T_C	ENST00000256993:exon25:c.2600-2A>G,ENST00000399249:exon26:c.2603-2A>G,ENST00000545968:exon27:c.2603-2A>G,ENST00000544791:exon27:c.2535-2A>G		L	0,00125

MYBPC3	11_47358987_C_-	ENSG00000134571:ENST00000256993:exon23:c.2554delG:p.G852fs,ENSG00000134571:ENST00000399249:exon24:c.2557delG:p.G853fs,ENSG00000134571:ENST00000545968:exon25:c.2557delG:p.G853fs		P	0,00080
MYBPC3	11_47358999_C_-	ENSG00000134571:ENST00000256993:exon23:c.2542delG:p.V848fs,ENSG00000134571:ENST00000399249:exon24:c.2545delG:p.V849fs,ENSG00000134571:ENST00000545968:exon25:c.2545delG:p.V849fs		L	0,00152
MYBPC3	11_47359000_C_-	ENSG00000134571:ENST00000256993:exon23:c.2541delG:p.A847fs,ENSG00000134571:ENST00000399249:exon24:c.2544delG:p.A848fs,ENSG00000134571:ENST00000545968:exon25:c.2544delG:p.A848fs		P	0,00151
MYBPC3	11_47359020_-_A	ENSG00000134571:ENST00000256993:exon23:c.2521_2522insT:p.Y841fs,ENSG00000134571:ENST00000399249:exon24:c.2524_2525insT:p.Y842fs,ENSG00000134571:ENST00000545968:exon25:c.2524_2525insT:p.Y842fs		L	0,00072
MYBPC3	11_47359032_-_C	ENSG00000134571:ENST00000256993:exon23:c.2509_2510insG:p.E837fs,ENSG00000134571:ENST00000399249:exon24:c.2512_2513insG:p.E838fs,ENSG00000134571:ENST00000545968:exon25:c.2512_2513insG:p.E838fs		L	0,00071
MYBPC3	11_47359047_C_T	ENSG00000134571:ENST00000256993:exon23:c.G2494A:p.A832T,ENSG00000134571:ENST00000399249:exon24:c.G2497A:p.A833T,ENSG00000134571:ENST00000545968:exon25:c.G2497A:p.A833T	rs199865688	D;P	0,00138
MYBPC3	11_47359085_C_T	ENSG00000134571:ENST00000256993:exon23:c.G2456A:p.R819Q,ENSG00000134571:ENST00000399249:exon24:c.G2459A:p.R820Q,ENSG00000134571:ENST00000545968:exon25:c.G2459A:p.R820Q	rs2856655	D;P	0,00142
MYBPC3	11_47359095_G_A	ENSG00000134571:ENST00000256993:exon23:c.C2446T:p.R816W,ENSG00000134571:ENST00000399249:exon24:c.C2449T:p.R817W,ENSG00000134571:ENST00000545968:exon25:c.C2449T:p.R817W		S	0,00071
MYBPC3	11_47359109_T_C	ENSG00000134571:ENST00000256993:exon23:c.A2432G:p.K811R,ENSG00000134571:ENST00000399249:exon24:c.A2435G:p.K812R,ENSG00000134571:ENST00000545968:exon25:c.A2435G:p.K812R		N	0,00072
MYBPC3	11_47359115_C_T	ENSG00000134571:ENST00000256993:exon23:c.G2426A:p.R809H,ENSG00000134571:ENST00000399249:exon24:c.G2429A:p.R810H,ENSG00000134571:ENST00000545968:exon25:c.G2429A:p.R810H		P	0,00218
MYBPC3	11_47359116_G_A	ENSG00000134571:ENST00000256993:exon23:c.C2425T:p.R809C,ENSG00000134571:ENST00000399249:exon24:c.C2428T:p.R810C,ENSG00000134571:ENST00000545968:exon25:c.C2428T:p.R810C		S	0,00074
MYBPC3	11_47359251_C_G	ENSG00000134571:ENST00000256993:exon22:c.G2400C:p.Q800H,ENSG00000134571:ENST00000399249:exon23:c.G2403C:p.Q801H,ENSG00000134571:ENST00000544791:exon24:c.G2403C:p.Q801H,ENSG00000134571:ENST00000545968:exon24:c.G2403C:p.Q801H		N	0,00073

MYBPC3	11_47359280_-_C	ENSG00000134571:ENST00000256993:exon22:c.2371_2372insG:p.W791fs,ENSG00000134571:ENST00000399249:exon23:c.2374_2375insG:p.W792fs,ENSG00000134571:ENST00000544791:exon24:c.2374_2375insG:p.W792fs,ENSG00000134571:ENST00000545968:exon24:c.2374_2375insG:p.W792fs		P	0,00645
MYBPC3	11_47359281_-_C	ENSG00000134571:ENST00000256993:exon22:c.2370_2371insG:p.Q790fs,ENSG00000134571:ENST00000399249:exon23:c.2373_2374insG:p.Q791fs,ENSG00000134571:ENST00000544791:exon24:c.2373_2374insG:p.Q791fs,ENSG00000134571:ENST00000545968:exon24:c.2373_2374insG:p.Q791fs		P	0,00646
MYBPC3	11_47359347_T_C	ENST00000256993:exon23:c.2306-2A>G,ENST00000399249:exon24:c.2309-2A>G,ENST00000545968:exon25:c.2309-2A>G,ENST00000544791:exon25:c.2309-2A>G		P	0,00064
MYBPC3	11_47360071_C_T	ENSG00000134571:ENST00000256993:exon21:c.G2305A:p.D769N,ENSG00000134571:ENST00000399249:exon22:c.G2308A:p.D770N,ENSG00000134571:ENST00000544791:exon23:c.G2308A:p.D770N,ENSG00000134571:ENST00000545968:exon23:c.G2308A:p.D770N	rs36211723	D;P	0,00064
MYBPC3	11_47360104_C_T	ENSG00000134571:ENST00000256993:exon21:c.G2272A:p.E758K,ENSG00000134571:ENST00000399249:exon22:c.G2275A:p.E759K,ENSG00000134571:ENST00000544791:exon23:c.G2275A:p.E759K,ENSG00000134571:ENST00000545968:exon23:c.G2275A:p.E759K		S	0,00062
MYBPC3	11_47360110_C_T	ENSG00000134571:ENST00000256993:exon21:c.G2266A:p.V756M,ENSG00000134571:ENST00000399249:exon22:c.G2269A:p.V757M,ENSG00000134571:ENST00000544791:exon23:c.G2269A:p.V757M,ENSG00000134571:ENST00000545968:exon23:c.G2269A:p.V757M		P	0,00061
MYBPC3	11_47360200_C_T	ENSG00000134571:ENST00000256993:exon21:c.G2176A:p.V726M,ENSG00000134571:ENST00000399249:exon22:c.G2179A:p.V727M,ENSG00000134571:ENST00000544791:exon23:c.G2179A:p.V727M,ENSG00000134571:ENST00000545968:exon23:c.G2179A:p.V727M		S	0,00066
MYBPC3	11_47360235_G_A	ENST00000256993:exon22:c.2146-5C>T,ENST00000399249:exon23:c.2149-5C>T,ENST00000545968:exon24:c.2149-5C>T,ENST00000544791:exon24:c.2149-5C>T	rs36211722	D;P	0,00139
MYBPC3	11_47360927_G_-	ENSG00000134571:ENST00000256993:exon20:c.2093delC:p.P698fs,ENSG00000134571:ENST00000399249:exon21:c.2096delC:p.P699fs,ENSG00000134571:ENST00000544791:exon22:c.2096delC:p.P699fs,ENSG00000134571:ENST00000545968:exon22:c.2096delC:p.P699fs		P	0,00173
MYBPC3	11_47360930_G_-	ENSG00000134571:ENST00000256993:exon20:c.2090delC:p.A697fs,ENSG00000134571:ENST00000399249:exon21:c.2093delC:p.A698fs,ENSG00000134571:ENST00000544791:exon22:c.2093delC:p.A698fs,ENSG00000134571:ENST00000545968:exon22:c.2093delC:p.A698fs		P	0,00173
MYBPC3	11_47361309_G_A	ENSG00000134571:ENST00000256993:exon19:c.C1957T:p.R653C,ENSG00000134571:ENST00000399249:exon20:c.C1960T:p.R654C,ENSG00000134571:ENST00000544791:exon21:c.C1960T:p.R654C,ENSG00000134571:ENST00000545968:exon21:c.C1960T:p.R654C		P	0,00059
MYBPC3	11_47361343_T_C	ENST00000256993:exon20:c.1925-2A>G,ENST00000399249:exon21:c.1928-2A>G,ENST00000545968:exon22:c.1928-2A>G,ENST00000544791:exon22:c.1928-2A>G		P	0,00061

MYBPC3	11_47362731_C_T	ENSG00000134571:ENST00000256993:exon17:c.G1852A:p.E618K,ENSG00000134571:ENST00000399249:exon18:c.G1855A:p.E619K,ENSG00000134571:ENST00000544791:exon19:c.G1855A:p.E619K,ENSG00000134571:ENST00000545968:exon19:c.G1855A:p.E619K	rs200352299	D;P	0,00065
MYBPC3	11_47363546_C_T	ENSG00000134571:ENST00000256993:exon16:c.G1783A:p.G595R,ENSG00000134571:ENST00000399249:exon17:c.G1786A:p.G596R,ENSG00000134571:ENST00000544791:exon18:c.G1786A:p.G596R,ENSG00000134571:ENST00000545968:exon18:c.G1786A:p.G596R	rs199728019	D;P	0,00116
MYBPC3	11_47363567_G_A	ENSG00000134571:ENST00000256993:exon16:c.C1762T:p.R588C,ENSG00000134571:ENST00000399249:exon17:c.C1765T:p.R589C,ENSG00000134571:ENST00000544791:exon18:c.C1765T:p.R589C,ENSG00000134571:ENST00000545968:exon18:c.C1765T:p.R589C		S	0,00058
MYBPC3	11_47363704_T_-	ENSG00000134571:ENST00000256993:exon16:c.1625delA:p.K542fs,ENSG00000134571:ENST00000399249:exon17:c.1628delA:p.K543fs,ENSG00000134571:ENST00000544791:exon18:c.1628delA:p.K543fs,ENSG00000134571:ENST00000545968:exon18:c.1628delA:p.K543fs		L	0,00058
MYBPC3	11_47364125_T_A	ENST00000256993:exon16:c.1621+4A>T,ENST00000399249:exon17:c.1624+4A>T,ENST00000545968:exon18:c.1624+4A>T,ENST00000544791:exon18:c.1624+4A>T		P	0,01346
MYBPC3	11_47364129_C_A	ENSG00000134571:ENST00000256993:exon15:c.G1621T:p.E541X,ENSG00000134571:ENST00000399249:exon16:c.G1624T:p.E542X,ENSG00000134571:ENST00000544791:exon17:c.G1624T:p.E542X,ENSG00000134571:ENST00000545968:exon17:c.G1624T:p.E542X		L	0,00090
MYBPC3	11_47364129_C_G	ENSG00000134571:ENST00000256993:exon15:c.G1621C:p.E541Q,ENSG00000134571:ENST00000399249:exon16:c.G1624C:p.E542Q,ENSG00000134571:ENST00000544791:exon17:c.G1624C:p.E542Q,ENSG00000134571:ENST00000545968:exon17:c.G1624C:p.E542Q	rs121909374	D;P	0,00269
MYBPC3	11_47364162_C_G	ENSG00000134571:ENST00000256993:exon15:c.G1588C:p.G530R,ENSG00000134571:ENST00000399249:exon16:c.G1591C:p.G531R,ENSG00000134571:ENST00000544791:exon17:c.G1591C:p.G531R,ENSG00000134571:ENST00000545968:exon17:c.G1591C:p.G531R		P	0,00083
MYBPC3	11_47364173_A_G	ENSG00000134571:ENST00000256993:exon15:c.T1577C:p.L526P,ENSG00000134571:ENST00000399249:exon16:c.T1580C:p.L527P,ENSG00000134571:ENST00000544791:exon17:c.T1580C:p.L527P,ENSG00000134571:ENST00000545968:exon17:c.T1580C:p.L527P		P	0,00078
MYBPC3	11_47364180_A_G	ENSG00000134571:ENST00000256993:exon15:c.T1570C:p.Y524H,ENSG00000134571:ENST00000399249:exon16:c.T1573C:p.Y525H,ENSG00000134571:ENST00000544791:exon17:c.T1573C:p.Y525H,ENSG00000134571:ENST00000545968:exon17:c.T1573C:p.Y525H		S	0,00076
MYBPC3	11_47364204_C_G	ENSG00000134571:ENST00000256993:exon15:c.G1546C:p.A516P,ENSG00000134571:ENST00000399249:exon16:c.G1549C:p.A517P,ENSG00000134571:ENST00000544791:exon17:c.G1549C:p.A517P,ENSG00000134571:ENST00000545968:exon17:c.G1549C:p.A517P		N	0,00071
MYBPC3	11_47364249_G_A	ENSG00000134571:ENST00000256993:exon15:c.C1501T:p.R501W,ENSG00000134571:ENST00000399249:exon16:c.C1504T:p.R502W,ENSG00000134571:ENST00000544791:exon17:c.C1504T:p.R502W,ENSG00000134571:ENST00000545968:exon17:c.C1504T:p.R502W		P	0,01038

MYBPC3	11_47364269_C_T	ENSG00000134571:ENST00000256993:exon15:c.G1481A:p.R494Q,ENSG00000134571:ENST00000399249:exon16:c.G1484A:p.R495Q,ENSG00000134571:ENST00000544791:exon17:c.G1484A:p.R495Q,ENSG00000134571:ENST00000545968:exon17:c.G1484A:p.R495Q	rs200411226	D;P	0,00319
MYBPC3	11_47364270_G_A	ENSG00000134571:ENST00000256993:exon15:c.C1480T:p.R494W,ENSG00000134571:ENST00000399249:exon16:c.C1483T:p.R495W,ENSG00000134571:ENST00000544791:exon17:c.C1483T:p.R495W,ENSG00000134571:ENST00000545968:exon17:c.C1483T:p.R495W		P	0,00064
MYBPC3	11_47364270_G_C	ENSG00000134571:ENST00000256993:exon15:c.C1480G:p.R494G,ENSG00000134571:ENST00000399249:exon16:c.C1483G:p.R495G,ENSG00000134571:ENST00000544791:exon17:c.C1483G:p.R495G,ENSG00000134571:ENST00000545968:exon17:c.C1483G:p.R495G		N	0,00256
MYBPC3	11_47364282_C_T	ENSG00000134571:ENST00000256993:exon15:c.G1468A:p.V490M,ENSG00000134571:ENST00000399249:exon16:c.G1471A:p.V491M,ENSG00000134571:ENST00000544791:exon17:c.G1471A:p.V491M,ENSG00000134571:ENST00000545968:exon17:c.G1471A:p.V491M		P	0,00064
MYBPC3	11_47364376_C_G	ENST00000256993:exon15:c.1454+5G>C,ENST00000399249:exon16:c.1457+5G>C,ENST00000545968:exon17:c.1457+5G>C,ENST00000544791:exon17:c.1457+5G>C		L	0,00060
MYBPC3	11_47364377_T_C	ENST00000256993:exon15:c.1454+4A>G,ENST00000399249:exon16:c.1457+4A>G,ENST00000545968:exon17:c.1457+4A>G,ENST00000544791:exon17:c.1457+4A>G		L	0,00120
MYBPC3	11_47364393_G_A	ENSG00000134571:ENST00000256993:exon14:c.C1442T:p.A481V,ENSG00000134571:ENST00000399249:exon15:c.C1445T:p.A482V,ENSG00000134571:ENST00000544791:exon16:c.C1445T:p.A482V,ENSG00000134571:ENST00000545968:exon16:c.C1445T:p.A482V		S	0,00060
MYBPC3	11_47364479_A_-	ENSG00000134571:ENST00000256993:exon14:c.1356delT:p.P452fs,ENSG00000134571:ENST00000399249:exon15:c.1359delT:p.P453fs,ENSG00000134571:ENST00000544791:exon16:c.1359delT:p.P453fs,ENSG00000134571:ENST00000545968:exon16:c.1359delT:p.P453fs		L	0,00061
MYBPC3	11_47364620_G_A	ENSG00000134571:ENST00000256993:exon13:c.C1300T:p.Q434X,ENSG00000134571:ENST00000399249:exon14:c.C1303T:p.Q435X,ENSG00000134571:ENST00000544791:exon15:c.C1303T:p.Q435X,ENSG00000134571:ENST00000545968:exon15:c.C1303T:p.Q435X		L	0,00063
MYBPC3	11_47364621_G_C	ENSG00000134571:ENST00000256993:exon13:c.C1299G:p.Y433X,ENSG00000134571:ENST00000399249:exon14:c.C1302G:p.Y434X,ENSG00000134571:ENST00000544791:exon15:c.C1302G:p.Y434X,ENSG00000134571:ENST00000545968:exon15:c.C1302G:p.Y434X		L	0,00063
MYBPC3	11_47364621_G_T	ENSG00000134571:ENST00000256993:exon13:c.C1299A:p.Y433X,ENSG00000134571:ENST00000399249:exon14:c.C1302A:p.Y434X,ENSG00000134571:ENST00000544791:exon15:c.C1302A:p.Y434X,ENSG00000134571:ENST00000545968:exon15:c.C1302A:p.Y434X		L	0,00063
MYBPC3	11_47364650_G_A	ENSG00000134571:ENST00000256993:exon13:c.C1270T:p.Q424X,ENSG00000134571:ENST00000399249:exon14:c.C1273T:p.Q425X,ENSG00000134571:ENST00000544791:exon15:c.C1273T:p.Q425X,ENSG00000134571:ENST00000545968:exon15:c.C1273T:p.Q425X		P	0,00063
MYBPC3	11_47364677_C_T	ENSG00000134571:ENST00000256993:exon13:c.G1243A:p.G415S,ENSG00000134571:ENST00000399249:exon14:c.G1246A:p.G416S,ENSG00000134571:ENST00000544791:exon15:c.G1246A:p.G416S,ENSG00000134571:ENST00000545968:exon15:c.G1246A:p.G416S		P	0,00066

MYBPC3	11_47364686_-_A	ENSG00000134571:ENST00000256993:exon13:c.1234_1235insT:p.E412_S413delinsX,ENSG00000134571:ENST00000399249:exon14:c.1237_1238insT:p.E413_S414delinsX,ENSG00000134571:ENST00000544791:exon15:c.1237_1238insT:p.E413_S414delinsX,ENSG00000134571:ENST00000545968:exon15:c.1237_1238insT:p.E413_S414delinsX		L	0,00137
MYBPC3	11_47364689_-_A	ENSG00000134571:ENST00000256993:exon13:c.1231_1232insT:p.F411fs,ENSG00000134571:ENST00000399249:exon14:c.1234_1235insT:p.F412fs,ENSG00000134571:ENST00000544791:exon15:c.1234_1235insT:p.F412fs,ENSG00000134571:ENST00000545968:exon15:c.1234_1235insT:p.F412fs		P	0,00137
MYBPC3	11_47364698_T_C	ENST00000256993:exon14:c.1224-2A>G,ENST00000399249:exon15:c.1227-2A>G,ENST00000545968:exon16:c.1227-2A>G,ENST00000544791:exon16:c.1227-2A>G		L	0,00070
MYBPC3	11_47365143_C_T	ENSG00000134571:ENST00000256993:exon12:c.G1123A:p.V375M,ENSG00000134571:ENST00000399249:exon12:c.G1123A:p.V375M,ENSG00000134571:ENST00000544791:exon13:c.G1123A:p.V375M,ENSG00000134571:ENST00000545968:exon13:c.G1123A:p.V375M		S	0,00095
MYBPC3	11_47367757_C_A	ENST00000256993:exon12:c.1090+1G>T,ENST00000399249:exon12:c.1090+1G>T,ENST00000545968:exon13:c.1090+1G>T,ENST00000544791:exon13:c.1090+1G>T		P	0,00231
MYBPC3	11_47367757_C_T	ENST00000256993:exon12:c.1090+1G>A,ENST00000399249:exon12:c.1090+1G>A,ENST00000545968:exon13:c.1090+1G>A,ENST00000544791:exon13:c.1090+1G>A		P	0,00058
MYBPC3	11_47367764_T_C	ENSG00000134571:ENST00000256993:exon11:c.A1084G:p.S362G,ENSG00000134571:ENST00000399249:exon11:c.A1084G:p.S362G,ENSG00000134571:ENST00000544791:exon12:c.A1084G:p.S362G,ENSG00000134571:ENST00000545968:exon12:c.A1084G:p.S362G		S	0,00058
MYBPC3	11_47367776_C_T	ENSG00000134571:ENST00000256993:exon11:c.G1072A:p.D358N,ENSG00000134571:ENST00000399249:exon11:c.G1072A:p.D358N,ENSG00000134571:ENST00000544791:exon12:c.G1072A:p.D358N,ENSG00000134571:ENST00000545968:exon12:c.G1072A:p.D358N		N	0,00058
MYBPC3	11_47367822_GACGCCGT_-	ENSG00000134571:ENST00000256993:exon11:c.1019_1026del:p.340_342del,ENSG00000134571:ENST00000399249:exon11:c.1019_1026del:p.340_342del,ENSG00000134571:ENST00000544791:exon12:c.1019_1026del:p.340_342del,ENSG00000134571:ENST00000545968:exon12:c.1019_1026del:p.340_342del		L	0,00058
MYBPC3	11_47367848_C_T	ENSG00000134571:ENST00000256993:exon11:c.G1000A:p.E334K,ENSG00000134571:ENST00000399249:exon11:c.G1000A:p.E334K,ENSG00000134571:ENST00000544791:exon12:c.G1000A:p.E334K,ENSG00000134571:ENST00000545968:exon12:c.G1000A:p.E334K		P	0,00061
MYBPC3	11_47367849_G_C	ENSG00000134571:ENST00000256993:exon11:c.C999G:p.Y333X,ENSG00000134571:ENST00000399249:exon11:c.C999G:p.Y333X,ENSG00000134571:ENST00000544791:exon12:c.C999G:p.Y333X,ENSG00000134571:ENST00000545968:exon12:c.C999G:p.Y333X		L	0,00061
MYBPC3	11_47367887_C_T	ENSG00000134571:ENST00000256993:exon11:c.G961A:p.V321M,ENSG00000134571:ENST00000399249:exon11:c.G961A:p.V321M,ENSG00000134571:ENST00000544791:exon12:c.G961A:p.V321M,ENSG00000134571:ENST00000545968:exon12:c.G961A:p.V321M	rs200119454	D;P	0,00187
MYBPC3	11_47367923_T_C	ENST00000256993:exon12:c.927-2A>G,ENST00000399249:exon12:c.927-2A>G,ENST00000545968:exon13:c.927-2A>G,ENST00000544791:exon13:c.927-2A>G		P	0,00132

MYBPC3	11_47368187_C_T	ENSG00000134571:ENST00000256993:exon10:c.G917A:p.R306Q,ENSG00000134571:ENST00000399249:exon10:c.G917A:p.R306Q,ENSG00000134571:ENST00000544791:exon11:c.G917A:p.R306Q,ENSG00000134571:ENST00000545968:exon11:c.G917A:p.R306Q		N	0,00059
MYBPC3	11_47368202_C_T	ENST00000256993:exon11:c.906-4G>A,ENST00000399249:exon11:c.906-4G>A		L	0,00060
MYBPC3	11_47369023_G_A	ENSG00000134571:ENST00000256993:exon9:c.C859T:p.H287Y,ENSG00000134571:ENST00000399249:exon9:c.C859T:p.H287Y,ENSG00000134571:ENST00000544791:exon9:c.C859T:p.H287Y,ENSG00000134571:ENST00000545968:exon9:c.C859T:p.H287Y		P	0,00066
MYBPC3	11_47369406_A_G	ENST00000256993:exon8:c.821+2T>C,ENST00000399249:exon8:c.821+2T>C,ENST00000545968:exon8:c.821+2T>C,ENST00000544791:exon8:c.821+2T>C		P	0,00087
MYBPC3	11_47369975_C_T	ENSG00000134571:ENST00000256993:exon6:c.G772A:p.E258K,ENSG00000134571:ENST00000399249:exon6:c.G772A:p.E258K,ENSG00000134571:ENST00000544791:exon6:c.G772A:p.E258K,ENSG00000134571:ENST00000545968:exon6:c.G772A:p.E258K		P	0,00616
MYBPC3	11_47370037_T_G	ENSG00000134571:ENST00000256993:exon6:c.A710C:p.Y237S,ENSG00000134571:ENST00000399249:exon6:c.A710C:p.Y237S,ENSG00000134571:ENST00000544791:exon6:c.A710C:p.Y237S,ENSG00000134571:ENST00000545968:exon6:c.A710C:p.Y237S		P	0,00140
MYBPC3	11_47370092_C_G	ENSG00000134571:ENST00000256993:exon6:c.G655C:p.V219L,ENSG00000134571:ENST00000399249:exon6:c.G655C:p.V219L,ENSG00000134571:ENST00000544791:exon6:c.G655C:p.V219L,ENSG00000134571:ENST00000545968:exon6:c.G655C:p.V219L		P	0,00442
MYBPC3	11_47371330_T_C	ENSG00000134571:ENST00000256993:exon5:c.A649G:p.S217G,ENSG00000134571:ENST00000399249:exon5:c.A649G:p.S217G,ENSG00000134571:ENST00000544791:exon5:c.A649G:p.S217G,ENSG00000134571:ENST00000545968:exon5:c.A649G:p.S217G	rs138753870	D;P	0,00067
MYBPC3	11_47371366_G_A	ENSG00000134571:ENST00000256993:exon5:c.C613T:p.Q205X,ENSG00000134571:ENST00000399249:exon5:c.C613T:p.Q205X,ENSG00000134571:ENST00000544791:exon5:c.C613T:p.Q205X,ENSG00000134571:ENST00000545968:exon5:c.C613T:p.Q205X		L	0,00068
MYBPC3	11_47371390_CCCATTTG_-	ENSG00000134571:ENST00000256993:exon5:c.582_589del:p.194_197del,ENSG00000134571:ENST00000399249:exon5:c.582_589del:p.194_197del,ENSG00000134571:ENST00000544791:exon5:c.582_589del:p.194_197del,ENSG00000134571:ENST00000545968:exon5:c.582_589del:p.194_197del		L	0,00071
MYBPC3	11_47371560_C_T	ENST00000256993:exon5:c.505+5G>A,ENST00000399249:exon5:c.505+5G>A,ENST00000545968:exon5:c.505+5G>A,ENST00000544791:exon5:c.505+5G>A		L	0,00096
MYBPC3	11_47372107_CA_-	ENSG00000134571:ENST00000256993:exon3:c.351_352del:p.117_118del,ENSG00000134571:ENST00000399249:exon3:c.351_352del:p.117_118del,ENSG00000134571:ENST00000544791:exon3:c.351_352del:p.117_118del,ENSG00000134571:ENST00000545968:exon3:c.351_352del:p.117_118del		L	0,00182
MYBPC3	11_47372859_C_T	ENSG00000134571:ENST00000256993:exon2:c.G223A:p.D75N,ENSG00000134571:ENST00000399249:exon2:c.G223A:p.D75N,ENSG00000134571:ENST00000544791:exon2:c.G223A:p.D75N,ENSG00000134571:ENST00000545968:exon2:c.G223A:p.D75N		P	0,00082

MYBPC3	11_47372866_-_CC	ENSG00000134571:ENST00000256993:exon2:c.216_217insGG:p.G72fs,ENSG00000134571:ENST00000399249:exon2:c.216_217insGG:p.G72fs,ENSG00000134571:ENST00000544791:exon2:c.216_217insGG:p.G72fs,ENSG00000134571:ENST00000545968:exon2:c.216_217insGG:p.G72fs		L	0,00081
MYBPC3	11_47372898_TGCCCTCTGTG_-	ENSG00000134571:ENST00000256993:exon2:c.174_184del:p.58_62del,ENSG00000134571:ENST00000399249:exon2:c.174_184del:p.58_62del,ENSG00000134571:ENST00000544791:exon2:c.174_184del:p.58_62del,ENSG00000134571:ENST00000545968:exon2:c.174_184del:p.58_62del		P	0,00319
MYBPC3	11_47372956_C_T	ENSG00000134571:ENST00000256993:exon2:c.G126A:p.W42X,ENSG00000134571:ENST00000399249:exon2:c.G126A:p.W42X,ENSG00000134571:ENST00000544791:exon2:c.G126A:p.W42X,ENSG00000134571:ENST00000545968:exon2:c.G126A:p.W42X		L	0,00078
MYBPC3	11_47372961_-_G	ENSG00000134571:ENST00000256993:exon2:c.121_122insC:p.R41fs,ENSG00000134571:ENST00000399249:exon2:c.121_122insC:p.R41fs,ENSG00000134571:ENST00000544791:exon2:c.121_122insC:p.R41fs,ENSG00000134571:ENST00000545968:exon2:c.121_122insC:p.R41fs		L	0,00077
MYBPC3	11_47374186_C_G	ENSG00000134571:ENST00000256993:exon1:c.G13C:p.G5R,ENSG00000134571:ENST00000399249:exon1:c.G13C:p.G5R,ENSG00000134571:ENST00000544791:exon1:c.G13C:p.G5R,ENSG00000134571:ENST00000545968:exon1:c.G13C:p.G5R	rs201278114	D;P	0,00117
MYH6	14_23852451_G_T	ENSG00000197616:ENST00000356287:exon36:c.C5644A:p.R1882S,ENSG00000197616:ENST00000405093:exon37:c.C5644A:p.R1882S		S	0,00058
MYH6	14_23852501_C_T	ENSG00000197616:ENST00000356287:exon36:c.G5594A:p.R1865Q,ENSG00000197616:ENST00000405093:exon37:c.G5594A:p.R1865Q	rs138720701	D;P	0,00059
MYH6	14_23852520_C_T	ENSG00000197616:ENST00000356287:exon36:c.G5575A:p.D1859N,ENSG00000197616:ENST00000405093:exon37:c.G5575A:p.D1859N		S	0,00069
MYH6	14_23853739_C_T	ENSG00000197616:ENST00000356287:exon35:c.G5477A:p.G1826D,ENSG00000197616:ENST00000405093:exon36:c.G5477A:p.G1826D	rs200260229	D	0,00119
MYH6	14_23853740_C_T	ENSG00000197616:ENST00000356287:exon35:c.G5476A:p.G1826S,ENSG00000197616:ENST00000405093:exon36:c.G5476A:p.G1826S	rs202141059	D	0,00119
MYH6	14_23853806_G_T	ENSG00000197616:ENST00000356287:exon35:c.C5410A:p.Q1804K,ENSG00000197616:ENST00000405093:exon36:c.C5410A:p.Q1804K	rs144571463	D	0,00060
MYH6	14_23854220_TCT_-	ENSG00000197616:ENST00000356287:exon34:c.5192_5194del:p.1731_1732del,ENSG00000197616:ENST00000405093:exon35:c.5192_5194del:p.1731_1732del		N	0,00057
MYH6	14_23855136_C_A	ENST00000405093:exon35:c.5163+1G>T,ENST00000356287:exon34:c.5163+1G>T		L	0,00058
MYH6	14_23855160_G_A	ENSG00000197616:ENST00000356287:exon33:c.C5140T:p.R1714W,ENSG00000197616:ENST00000405093:exon34:c.C5140T:p.R1714W	rs140651265	D;S	0,00058
MYH6	14_23855228_C_T	ENSG00000197616:ENST00000356287:exon33:c.G5072A:p.R1691H,ENSG00000197616:ENST00000405093:exon34:c.G5072A:p.R1691H		S	0,00060
MYH6	14_23855609_T_C	ENSG00000197616:ENST00000356287:exon32:c.A4874G:p.N1625S,ENSG00000197616:ENST00000405093:exon33:c.A4874G:p.N1625S		S	0,00058

MYH6	14_23855779_G_T	ENSG00000197616:ENST00000356287:exon32:c.C4704A:p.N1568K,ENSG00000197616:ENST00000405093:exon33:c.C4704A:p.N1568K	rs149771264	D;S	0,00058
MYH6	14_23856786_C_G	ENSG00000197616:ENST00000356287:exon31:c.G4602C:p.Q1534H,ENSG00000197616:ENST00000405093:exon32:c.G4602C:p.Q1534H	rs199600772	D;S	0,00057
MYH6	14_23856987_C_T	ENSG00000197616:ENST00000356287:exon30:c.G4505A:p.R1502Q,ENSG00000197616:ENST00000405093:exon31:c.G4505A:p.R1502Q	rs199936506	D	0,00115
MYH6	14_23857371_A_T	ENSG00000197616:ENST00000356287:exon29:c.T4352A:p.F1451Y,ENSG00000197616:ENST00000405093:exon30:c.T4352A:p.F1451Y		N	0,00058
MYH6	14_23857395_G_T	ENSG00000197616:ENST00000356287:exon29:c.C4328A:p.A1443D,ENSG00000197616:ENST00000405093:exon30:c.C4328A:p.A1443D		P	0,00058
MYH6	14_23857530_C_T	ENSG00000197616:ENST00000356287:exon29:c.G4193A:p.R1398Q,ENSG00000197616:ENST00000405093:exon30:c.G4193A:p.R1398Q	rs150815925	D	0,00058
MYH6	14_23858107_G_A	ENSG00000197616:ENST00000356287:exon28:c.C4136T:p.T1379M,ENSG00000197616:ENST00000405093:exon29:c.C4136T:p.T1379M	rs145611185	D;P	0,00058
MYH6	14_23858161_C_T	ENSG00000197616:ENST00000356287:exon28:c.G4082A:p.R1361H,ENSG00000197616:ENST00000405093:exon29:c.G4082A:p.R1361H		S	0,00058
MYH6	14_23858648_G_A	ENSG00000197616:ENST00000356287:exon27:c.C3932T:p.T1311I,ENSG00000197616:ENST00000405093:exon28:c.C3932T:p.T1311I		S	0,00058
MYH6	14_23858687_G_A	ENSG00000197616:ENST00000356287:exon27:c.C3893T:p.A1298V,ENSG00000197616:ENST00000405093:exon28:c.C3893T:p.A1298V		N	0,00058
MYH6	14_23858720_C_T	ENSG00000197616:ENST00000356287:exon27:c.G3860A:p.G1287E,ENSG00000197616:ENST00000405093:exon28:c.G3860A:p.G1287E		N	0,00058
MYH6	14_23859370_C_T	ENSG00000197616:ENST00000356287:exon25:c.G3628A:p.D1210N,ENSG00000197616:ENST00000405093:exon26:c.G3628A:p.D1210N		S	0,00065
MYH6	14_23859484_C_T	ENSG00000197616:ENST00000356287:exon25:c.G3514A:p.E1172K,ENSG00000197616:ENST00000405093:exon26:c.G3514A:p.E1172K		S	0,00092
MYH6	14_23859571_G_A	ENSG00000197616:ENST00000356287:exon25:c.C3427T:p.R1143W,ENSG00000197616:ENST00000405093:exon26:c.C3427T:p.R1143W		S	0,00093
MYH6	14_23859652_G_T	ENSG00000197616:ENST00000356287:exon25:c.C3346A:p.R1116S,ENSG00000197616:ENST00000405093:exon26:c.C3346A:p.R1116S		P	0,00080
MYH6	14_23861814_T_A	ENSG00000197616:ENST00000356287:exon24:c.A3299T:p.Q1100L,ENSG00000197616:ENST00000405093:exon25:c.A3299T:p.Q1100L		S	0,00057
MYH6	14_23862208_C_T	ENSG00000197616:ENST00000356287:exon23:c.G3164A:p.R1055Q,ENSG00000197616:ENST00000405093:exon24:c.G3164A:p.R1055Q		N	0,00057
MYH6	14_23862646_C_A	ENSG00000197616:ENST00000356287:exon22:c.G3010T:p.A1004S,ENSG00000197616:ENST00000405093:exon23:c.G3010T:p.A1004S	rs143978652	D;P	0,00057

MYH6	14_23862870_C_T	ENST00000405093:exon23:c.2928+5G>A,ENST00000356287:exon22:c.2928+5G>A	rs28730772	D;L	0,00400
MYH6	14_23863087_G_A	ENSG00000197616:ENST00000356287:exon21:c.C2716T:p.R906C,ENSG00000197616:ENST00000405093:exon22:c.C2716T:p.R906C	rs143928061	D;S	0,00057
MYH6	14_23863348_G_A	ENSG00000197616:ENST00000356287:exon20:c.C2614T:p.R872C,ENSG00000197616:ENST00000405093:exon21:c.C2614T:p.R872C	rs201193346	D;S	0,00058
MYH6	14_23863384_G_A	ENSG00000197616:ENST00000356287:exon20:c.C2578T:p.R860C,ENSG00000197616:ENST00000405093:exon21:c.C2578T:p.R860C		S	0,00057
MYH6	14_23865497_G_A	ENSG00000197616:ENST00000356287:exon19:c.C2425T:p.R809C,ENSG00000197616:ENST00000405093:exon20:c.C2425T:p.R809C		S	0,00057
MYH6	14_23865569_G_A	ENSG00000197616:ENST00000356287:exon19:c.C2353T:p.R785C,ENSG00000197616:ENST00000405093:exon20:c.C2353T:p.R785C		S	0,00058
MYH6	14_23866275_G_T	ENSG00000197616:ENST00000356287:exon17:c.C2065A:p.P689T,ENSG00000197616:ENST00000405093:exon18:c.C2065A:p.P689T		N	0,00060
MYH6	14_23868065_T_G	ENSG00000197616:ENST00000356287:exon14:c.A1763C:p.D588A,ENSG00000197616:ENST00000405093:exon15:c.A1763C:p.D588A	rs142992009	D	0,00057
MYH6	14_23869462_C_T	ENST00000405093:exon15:c.1581+3G>A,ENST00000356287:exon14:c.1581+3G>A		L	0,00057
MYH6	14_23869967_C_A	ENSG00000197616:ENST00000356287:exon12:c.G1361T:p.R454L,ENSG00000197616:ENST00000405093:exon13:c.G1361T:p.R454L		S	0,00058
MYH6	14_23870001_G_A	ENSG00000197616:ENST00000356287:exon12:c.C1327T:p.R443C,ENSG00000197616:ENST00000405093:exon13:c.C1327T:p.R443C	rs182373896	D;S	0,00058
MYH6	14_23870012_C_T	ENSG00000197616:ENST00000356287:exon12:c.G1316A:p.W439X,ENSG00000197616:ENST00000405093:exon13:c.G1316A:p.W439X		L	0,00058
MYH6	14_23870034_C_G	ENSG00000197616:ENST00000356287:exon12:c.G1294C:p.V432L,ENSG00000197616:ENST00000405093:exon13:c.G1294C:p.V432L		N	0,00057
MYH6	14_23870151_A_C	ENSG00000197616:ENST00000356287:exon12:c.T1177G:p.S393A,ENSG00000197616:ENST00000405093:exon13:c.T1177G:p.S393A	rs199877580	D	0,00057
MYH6	14_23871758_C_G	ENSG00000197616:ENST00000356287:exon11:c.G1056C:p.K352N,ENSG00000197616:ENST00000405093:exon12:c.G1056C:p.K352N		S	0,00058
MYH6	14_23871766_C_T	ENSG00000197616:ENST00000356287:exon11:c.G1048A:p.V350I,ENSG00000197616:ENST00000405093:exon12:c.G1048A:p.V350I	rs200260629	D	0,00058
MYH6	14_23872631_A_T	ENSG00000197616:ENST00000356287:exon9:c.T824A:p.I275N,ENSG00000197616:ENST00000405093:exon10:c.T824A:p.I275N	rs201327273	D;P	0,00120
MYH6	14_23874309_G_A	ENSG00000197616:ENST00000356287:exon5:c.C530T:p.T177M,ENSG00000197616:ENST00000405093:exon6:c.C530T:p.T177M		S	0,00058
MYH6	14_23874525_C_T	ENSG00000197616:ENST00000356287:exon4:c.G409A:p.E137K,ENSG00000197616:ENST00000405093:exon5:c.G409A:p.E137K		N	0,00061

MYH6	14_23874889_C_T	ENSG00000197616:ENST00000356287:exon3:c.G292A:p.E98K,ENSG00000197616:ENST00000405093:exon4:c.G292A:p.E98K	rs140596256	D	0,00059
MYH6	14_23874936_G_A	ENSG00000197616:ENST00000356287:exon3:c.C245T:p.P82L,ENSG00000197616:ENST00000405093:exon4:c.C245T:p.P82L		S	0,00058
MYH6	14_23876352_C_G	ENSG00000197616:ENST00000356287:exon2:c.G81C:p.Q27H,ENSG00000197616:ENST00000405093:exon3:c.G81C:p.Q27H		N	0,00058
MYH6	14_23876363_G_T	ENSG00000197616:ENST00000356287:exon2:c.C70A:p.L24I,ENSG00000197616:ENST00000405093:exon3:c.C70A:p.L24I		N	0,00115
MYH6	14_23876785_C_T			L	0,00059
MYH7	14_23882979_T_A	ENSG00000092054:ENST00000355349:exon39:c.A5779T:p.I1927F		P	0,00058
MYH7	14_23883068_C_T	ENSG00000092054:ENST00000355349:exon39:c.G5690A:p.R1897H		S	0,00058
MYH7	14_23883217_G_A	ENSG00000092054:ENST00000355349:exon38:c.C5654T:p.A1885V		N	0,00059
MYH7	14_23884200_G_A	ENST00000355349:exon38:c.5559+4C>T		L	0,00058
MYH7	14_23884421_C_T	ENSG00000092054:ENST00000355349:exon37:c.G5342A:p.R1781H		P	0,00058
MYH7	14_23884469_A_T	ENSG00000092054:ENST00000355349:exon37:c.T5294A:p.M1765K		S	0,00058
MYH7	14_23884860_C_T	ENSG00000092054:ENST00000355349:exon35:c.G5135A:p.R1712Q	rs193922390	D;P	0,00063
MYH7	14_23885349_C_T	ENSG00000092054:ENST00000355349:exon34:c.G4817A:p.R1606H		N	0,00058
MYH7	14_23885502_T_C	ENSG00000092054:ENST00000355349:exon34:c.A4664G:p.E1555G		P	0,00058
MYH7	14_23886806_C_T	ENSG00000092054:ENST00000355349:exon31:c.G4259A:p.R1420Q		P	0,00058
MYH7	14_23886815_G_A	ENSG00000092054:ENST00000355349:exon31:c.C4250T:p.T1417I		S	0,00058
MYH7	14_23887522_C_T	ENSG00000092054:ENST00000355349:exon30:c.G4066A:p.E1356K		P	0,00296
MYH7	14_23887607_G_T	ENSG00000092054:ENST00000355349:exon30:c.C3981A:p.N1327K	rs141764279	D	0,00058
MYH7	14_23888775_T_C	ENSG00000092054:ENST00000355349:exon28:c.A3770G:p.N1257S		N	0,00058
MYH7	14_23889158_C_T	ENSG00000092054:ENST00000355349:exon27:c.G3622A:p.D1208N		S	0,00062
MYH7	14_23889187_T_C	ENSG00000092054:ENST00000355349:exon27:c.A3593G:p.D1198G		S	0,00070
MYH7	14_23892910_A_G	ENSG00000092054:ENST00000355349:exon24:c.T2945C:p.M982T	rs145532615	D;P	0,00057
MYH7	14_23893148_C_G	ENSG00000092054:ENST00000355349:exon23:c.G2890C:p.V964L	rs45496496	D;P	0,00344
MYH7	14_23893192_T_A	ENSG00000092054:ENST00000355349:exon23:c.A2846T:p.E949V		S	0,00057

MYH7	14_23893246_T_C	ENSG00000092054:ENST00000355349:exon23:c.A2792G:p.E931G		S	0,00057
MYH7	14_23893255_T_C	ENSG00000092054:ENST00000355349:exon23:c.A2783G:p.D928G		S	0,00057
MYH7	14_23893268_C_G	ENSG00000092054:ENST00000355349:exon23:c.G2770C:p.E924Q		P	0,00057
MYH7	14_23893268_C_T	ENSG00000092054:ENST00000355349:exon23:c.G2770A:p.E924K	rs121913628	D;P	0,00057
MYH7	14_23893316_G_C	ENSG00000092054:ENST00000355349:exon23:c.C2722G:p.L908V	rs121913631	D;P	0,00057
MYH7	14_23893321_T_C	ENSG00000092054:ENST00000355349:exon23:c.A2717G:p.D906G		P	0,00286
MYH7	14_23893357_T_C	ENSG00000092054:ENST00000355349:exon23:c.A2681G:p.E894G		P	0,00172
MYH7	14_23893988_T_G	ENSG00000092054:ENST00000355349:exon22:c.A2669C:p.Q890P		S	0,00060
MYH7	14_23894048_C_T	ENSG00000092054:ENST00000355349:exon22:c.G2609A:p.R870H	rs36211715	D;P	0,00293
MYH7	14_23894051_C_G	ENSG00000092054:ENST00000355349:exon22:c.G2606C:p.R869P		N	0,00117
MYH7	14_23894051_C_T	ENSG00000092054:ENST00000355349:exon22:c.G2606A:p.R869H	rs202141173	D;P	0,00058
MYH7	14_23894052_G_A	ENSG00000092054:ENST00000355349:exon22:c.C2605T:p.R869C		P	0,00117
MYH7	14_23894052_G_T	ENSG00000092054:ENST00000355349:exon22:c.C2605A:p.R869S		N	0,00058
MYH7	14_23894084_C_T	ENSG00000092054:ENST00000355349:exon22:c.G2573A:p.R858H	rs2856897	P	0,00058
MYH7	14_23894118_T_C	ENSG00000092054:ENST00000355349:exon22:c.A2539G:p.K847E		P	0,00116
MYH7	14_23894118_TCT_-	ENSG00000092054:ENST00000355349:exon22:c.2537_2539del:p.846_847del		N	0,00058
MYH7	14_23894123_C_T	ENSG00000092054:ENST00000355349:exon22:c.G2534A:p.R845K		N	0,00058
MYH7	14_23894193_T_A	ENSG00000092054:ENST00000355349:exon22:c.A2464T:p.M822L		P	0,00058
MYH7	14_23894525_C_T	ENSG00000092054:ENST00000355349:exon21:c.G2389A:p.A797T	rs3218716	D;P	0,00172
MYH7	14_23894539_G_T	ENSG00000092054:ENST00000355349:exon21:c.C2375A:p.S792Y		N	0,00058
MYH7	14_23894554_C_T	ENSG00000092054:ENST00000355349:exon21:c.G2360A:p.R787H		P	0,00173
MYH7	14_23894566_C_T	ENSG00000092054:ENST00000355349:exon21:c.G2348A:p.R783H		P	0,00058
MYH7	14_23894969_C_A	ENSG00000092054:ENST00000355349:exon20:c.G2221T:p.G741W		P	0,00057
MYH7	14_23894969_C_G	ENSG00000092054:ENST00000355349:exon20:c.G2221C:p.G741R	rs121913632	D;P	0,00057
MYH7	14_23895023_G_A	ENSG00000092054:ENST00000355349:exon20:c.C2167T:p.R723C	rs121913630	D;P	0,00230
MYH7	14_23895023_G_C	ENSG00000092054:ENST00000355349:exon20:c.C2167G:p.R723G		P	0,00057

MYH7	14_23895179_C_T	ENSG00000092054:ENST00000355349:exon19:c.G2156A:p.R719Q	rs121913641	D;P	0,00118
MYH7	14_23895180_G_A	ENSG00000092054:ENST00000355349:exon19:c.C2155T:p.R719W	rs121913637	D;P	0,00059
MYH7	14_23895243_C_G	ENSG00000092054:ENST00000355349:exon19:c.G2092C:p.V698L		N	0,00065
MYH7	14_23895254_C_T	ENSG00000092054:ENST00000355349:exon19:c.G2081A:p.R694H		P	0,00065
MYH7	14_23896042_C_T	ENSG00000092054:ENST00000355349:exon18:c.G1988A:p.R663H		P	0,00287
MYH7	14_23896043_G_A	ENSG00000092054:ENST00000355349:exon18:c.C1987T:p.R663C		P	0,00172
MYH7	14_23896812_A_T	ENSG00000092054:ENST00000355349:exon16:c.T1870A:p.Y624N		P	0,00057
MYH7	14_23896866_C_T	ENSG00000092054:ENST00000355349:exon16:c.G1816A:p.V606M	rs121913627	D;P	0,00229
MYH7	14_23897796_C_A	ENSG00000092054:ENST00000355349:exon15:c.G1491T:p.E497D		P	0,00057
MYH7	14_23897840_C_T	ENSG00000092054:ENST00000355349:exon15:c.G1447A:p.E483K	rs121913651	D;P	0,00057
MYH7	14_23897854_A_T	ENSG00000092054:ENST00000355349:exon15:c.T1433A:p.I478N		S	0,00057
MYH7	14_23898213_C_A	ENSG00000092054:ENST00000355349:exon14:c.G1358T:p.R453L		S	0,00057
MYH7	14_23898240_T_C	ENSG00000092054:ENST00000355349:exon14:c.A1331G:p.N444S		P	0,00057
MYH7	14_23898292_G_T	ENSG00000092054:ENST00000355349:exon14:c.C1279A:p.L427M		P	0,00230
MYH7	14_23898464_C_T	ENSG00000092054:ENST00000355349:exon13:c.G1231A:p.V411I		P	0,00057
MYH7	14_23898487_C_T	ENSG00000092054:ENST00000355349:exon13:c.G1208A:p.R403Q	rs121913624	D;P	0,00057
MYH7	14_23898488_G_A	ENSG00000092054:ENST00000355349:exon13:c.C1207T:p.R403W	rs3218714	D;P	0,00172
MYH7	14_23898551_C_A	ENSG00000092054:ENST00000355349:exon13:c.G1144T:p.D382Y		P	0,00115
MYH7	14_23899059_C_A	ENSG00000092054:ENST00000355349:exon12:c.G1063T:p.A355S		N	0,00115
MYH7	14_23899059_C_T	ENSG00000092054:ENST00000355349:exon12:c.G1063A:p.A355T		P	0,00229
MYH7	14_23899071_T_C	ENSG00000092054:ENST00000355349:exon12:c.A1051G:p.K351E		P	0,00115
MYH7	14_23899101_A_G	ENSG00000092054:ENST00000355349:exon12:c.T1021C:p.F341L		S	0,00057
MYH7	14_23899864_G_T	ENSG00000092054:ENST00000355349:exon11:c.C904A:p.L302M		S	0,00057
MYH7	14_23900635_A_G	ENSG00000092054:ENST00000355349:exon9:c.T788C:p.I263T		P	0,00057
MYH7	14_23900656_C_T	ENSG00000092054:ENST00000355349:exon9:c.G767A:p.G256E	rs121913633	D;P	0,00057
MYH7	14_23900677_C_T	ENSG00000092054:ENST00000355349:exon9:c.G746A:p.R249Q	rs3218713	D;P	0,00057

MYH7	14_23900811_C_T	ENSG00000092054:ENST00000355349:exon8:c.G715A:p.D239N		P	0,00058
MYH7	14_23901712_C_T	ENSG00000092054:ENST00000355349:exon6:c.G506A:p.R169K		N	0,00058
MYH7	14_23901922_C_T	ENSG00000092054:ENST00000355349:exon5:c.G428A:p.R143Q		P	0,00059
MYH7	14_23902293_G_T	ENSG00000092054:ENST00000355349:exon4:c.C345A:p.Y115X		L	0,00058
MYH7	14_23902340_C_T	ENSG00000092054:ENST00000355349:exon4:c.G298A:p.A100T		S	0,00058
MYH7	14_23902892_C_T	ENSG00000092054:ENST00000355349:exon3:c.G50A:p.R17H		S	0,00058
MYH7	14_23903456_G_A		rs45566639	D;L	0,00401
MYL2	12_111350901_T_G	ENSG00000111245:ENST00000548438:exon5:c.A359C:p.E120A,ENSG00000111245:ENST00000228841:exon6:c.A401C:p.E134A	rs143139258	D;S	0,00115
MYL2	12_111350926_G_C	ENSG00000111245:ENST00000548438:exon5:c.C334G:p.Q112E,ENSG00000111245:ENST00000550439:exon5:c.C319G:p.Q107E,ENSG00000111245:ENST00000228841:exon6:c.C376G:p.Q126E		S	0,00058
MYL2	12_111352004_C_G	ENSG00000111245:ENST00000548438:exon3:c.G218C:p.G73A,ENSG00000111245:ENST00000550439:exon3:c.G203C:p.G68A,ENSG00000111245:ENST00000228841:exon4:c.G260C:p.G87A		S	0,00058
MYL2	12_111352091_C_T	ENSG00000111245:ENST00000548438:exon3:c.G131A:p.R44Q,ENSG00000111245:ENST00000550439:exon3:c.G116A:p.R39Q,ENSG00000111245:ENST00000228841:exon4:c.G173A:p.R58Q	rs104894369	D;P	0,00057
MYL2	12_111353547_G_T	ENSG00000111245:ENST00000550439:exon2:c.C84A:p.N28K,ENSG00000111245:ENST00000228841:exon3:c.C141A:p.N47K	rs199474808	D;P	0,00057
MYL2	12_111356937_C_T	ENSG00000111245:ENST00000550439:exon1:c.G7A:p.E3K,ENSG00000111245:ENST00000228841:exon2:c.G64A:p.E22K,ENSG00000111245:ENST00000548438:exon2:c.G64A:p.E22K	rs104894368	D;P	0,00057
MYL2	12_111356964_C_T	ENSG00000111245:ENST00000228841:exon2:c.G37A:p.A13T,ENSG00000111245:ENST00000548438:exon2:c.G37A:p.A13T	rs104894363	D;P	0,00115
MYL3	3_46899903_T_C	ENSG00000160808:ENST00000292327:exon5:c.A530G:p.E177G,ENSG00000160808:ENST00000395869:exon5:c.A530G:p.E177G	rs193922391	D;S	0,00058
MYL3	3_46900970_G_A	ENSG00000160808:ENST00000292327:exon4:c.C476T:p.T159M,ENSG00000160808:ENST00000395869:exon4:c.C476T:p.T159M		S	0,00058
MYL3	3_46900986_G_A	ENSG00000160808:ENST00000292327:exon4:c.C460T:p.R154C,ENSG00000160808:ENST00000395869:exon4:c.C460T:p.R154C	rs143852164	D;S	0,00115
MYL3	3_46901009_C_T	ENSG00000160808:ENST00000292327:exon4:c.G437A:p.G146D,ENSG00000160808:ENST00000395869:exon4:c.G437A:p.G146D		S	0,00058
MYL3	3_46902285_C_T	ENSG00000160808:ENST00000292327:exon3:c.G188A:p.R63H,ENSG00000160808:ENST00000395869:exon3:c.G188A:p.R63H	rs139354105	D;S	0,00058
PDLIM3	4_186423516_T_C	ENSG00000154553:ENST00000284771:exon7:c.A883G:p.T295A,ENSG00000154553:ENST00000284770:exon8:c.A1027G:p.T343A		N	0,00172

PDLIM3	4_186423579_C_T	ENSG00000154553:ENST00000284771:exon7:c.G820A:p.D274N,ENSG00000154553:ENST00000284770:exon8:c.G964A:p.D322N		S	0,00057
PDLIM3	4_186427754_C_T	ENSG00000154553:ENST00000284771:exon5:c.G571A:p.D191N,ENSG00000154553:ENST00000284770:exon6:c.G715A:p.D239N	rs142143310	D	0,00058
PDLIM3	4_186427786_G_A	ENSG00000154553:ENST00000284771:exon5:c.C539T:p.P180L,ENSG00000154553:ENST00000284770:exon6:c.C683T:p.P228L	rs201185673	D	0,00115
PDLIM3	4_186429568_C_A	ENSG00000154553:ENST00000284770:exon5:c.G547T:p.V183L		N	0,00057
PDLIM3	4_186435447_A_G	ENSG00000154553:ENST00000284767:exon5:c.T563C:p.L188P		N	0,00057
PDLIM3	4_186435483_G_C	ENSG00000154553:ENST00000284770:exon4:c.C339G:p.N113K,ENSG00000154553:ENST00000284767:exon5:c.C527G:p.T176S		N	0,00057
PDLIM3	4_186444600_C_T	ENSG00000154553:ENST00000505886:exon2:c.G94A:p.G32R,ENSG00000154553:ENST00000512293:exon2:c.G94A:p.G32R	rs200354645	D	0,00115
PDLIM3	4_186446250_T_C	ENSG00000154553:ENST00000284767:exon2:c.A169G:p.T57A,ENSG00000154553:ENST00000284770:exon2:c.A169G:p.T57A,ENSG00000154553:ENST00000284771:exon2:c.A169G:p.T57A	rs142951316	D	0,00115
PKP2	12_32949101_G_T	ENSG00000057294:ENST00000340811:exon11:c.C2299A:p.R767S,ENSG00000057294:ENST00000070846:exon12:c.C2431A:p.R811S	rs139734328	D	0,00058
PKP2	12_32955491_C_G	ENST00000340811:exon11:c.2014-1G>C,ENST00000070846:exon12:c.2146-1G>C	rs193922674	D;P	0,00057
PKP2	12_32975431_A_C	ENSG00000057294:ENST00000340811:exon8:c.T1809G:p.C603W,ENSG00000057294:ENST00000070846:exon9:c.T1941G:p.C647W	rs149392678	D;S	0,00114
PKP2	12_32994007_C_-	ENSG00000057294:ENST00000340811:exon6:c.1511delG:p.G504fs,ENSG00000057294:ENST00000070846:exon7:c.1643delG:p.G548fs		P	0,00057
PKP2	12_32994100_T_C	ENSG00000057294:ENST00000340811:exon6:c.A1418G:p.N473S,ENSG00000057294:ENST00000070846:exon7:c.A1550G:p.N517S	rs144536197	D	0,00115
PKP2	12_32996206_C_T	ENSG00000057294:ENST00000070846:exon6:c.G1420A:p.A474T	rs138538072	D	0,00193
PKP2	12_33021936_C_T	ENSG00000057294:ENST00000070846:exon4:c.G1095A:p.M365I,ENSG00000057294:ENST00000340811:exon4:c.G1095A:p.M365I		N	0,00058
PKP2	12_33021997_C_G	ENST00000340811:exon5:c.1035-1G>C,ENST00000070846:exon5:c.1035-1G>C		L	0,00057
PKP2	12_33030850_C_A	ENSG00000057294:ENST00000070846:exon3:c.G964T:p.G322C,ENSG00000057294:ENST00000340811:exon3:c.G964T:p.G322C		S	0,00064
PKP2	12_33031039_C_A	ENSG00000057294:ENST00000070846:exon3:c.G775T:p.E259X,ENSG00000057294:ENST00000340811:exon3:c.G775T:p.E259X		P	0,00062
PKP2	12_33031321_C_T	ENSG00000057294:ENST00000070846:exon3:c.G493A:p.D165N,ENSG00000057294:ENST00000340811:exon3:c.G493A:p.D165N		S	0,00118

PKP2	12_33031329_G_A	ENSG00000057294:ENST00000070846:exon3:c.C485T:p.T162M,ENSG00000057294:ENST00000340811:exon3:c.C485T:p.T162M		N	0,00059
PKP2	12_33031468_T_C	ENSG00000057294:ENST00000070846:exon3:c.A346G:p.T116A,ENSG00000057294:ENST00000340811:exon3:c.A346G:p.T116A		N	0,00057
PKP2	12_33031883_G_C	ENSG00000057294:ENST00000070846:exon2:c.C307G:p.P103A,ENSG00000057294:ENST00000340811:exon2:c.C307G:p.P103A		N	0,00057
PKP2	12_33031883_G_T	ENSG00000057294:ENST00000070846:exon2:c.C307A:p.P103T,ENSG00000057294:ENST00000340811:exon2:c.C307A:p.P103T	rs139215336	D	0,00057
PKP2	12_33031888_C_T	ENSG00000057294:ENST00000070846:exon2:c.G302A:p.R101H,ENSG00000057294:ENST00000340811:exon2:c.G302A:p.R101H	rs149542398	D;P	0,00115
PKP2	12_33031931_C_G	ENSG00000057294:ENST00000070846:exon2:c.G259C:p.V87L,ENSG00000057294:ENST00000340811:exon2:c.G259C:p.V87L		P	0,00057
PKP2	12_33049492_C_A	ENSG00000057294:ENST00000070846:exon1:c.G174T:p.E58D,ENSG00000057294:ENST00000340811:exon1:c.G174T:p.E58D	rs146708884	D;P	0,00771
PKP4	2_159313728_A_C			L	0,00993
PKP4	2_159459593_A_G	ENSG00000144283:ENST00000389757:exon4:c.A257G:p.K86R,ENSG00000144283:ENST00000389759:exon4:c.A257G:p.K86R,ENSG00000144283:ENST00000421462:exon4:c.A257G:p.K86R,ENSG00000144283:ENST00000426248:exon4:c.A257G:p.K86R	rs144452632	D	0,00057
PKP4	2_159459601_C_T	ENSG00000144283:ENST00000389757:exon4:c.C265T:p.P89S,ENSG00000144283:ENST00000389759:exon4:c.C265T:p.P89S,ENSG00000144283:ENST00000421462:exon4:c.C265T:p.P89S,ENSG00000144283:ENST00000426248:exon4:c.C265T:p.P89S	rs141436976	D	0,00057
PKP4	2_159477550_A_G	ENSG00000144283:ENST00000389757:exon5:c.A329G:p.Q110R,ENSG00000144283:ENST00000389759:exon5:c.A329G:p.Q110R,ENSG00000144283:ENST00000421462:exon5:c.A329G:p.Q110R,ENSG00000144283:ENST00000426248:exon5:c.A329G:p.Q110R	rs148019751	D	0,00057
PKP4	2_159477598_A_G	ENSG00000144283:ENST00000389757:exon5:c.A377G:p.Y126C,ENSG00000144283:ENST00000389759:exon5:c.A377G:p.Y126C,ENSG00000144283:ENST00000421462:exon5:c.A377G:p.Y126C,ENSG00000144283:ENST00000426248:exon5:c.A377G:p.Y126C		S	0,00114
PKP4	2_159481523_G_C	ENSG00000144283:ENST00000389757:exon7:c.G737C:p.G246A,ENSG00000144283:ENST00000389759:exon7:c.G737C:p.G246A		N	0,00058
PKP4	2_159481526_C_T	ENSG00000144283:ENST00000389757:exon7:c.C740T:p.S247F,ENSG00000144283:ENST00000389759:exon7:c.C740T:p.S247F		N	0,00058
PKP4	2_159481840_C_A	ENSG00000144283:ENST00000389757:exon7:c.C1054A:p.P352T,ENSG00000144283:ENST00000389759:exon7:c.C1054A:p.P352T		N	0,00058
PKP4	2_159481864_G_T	ENSG00000144283:ENST00000389757:exon7:c.G1078T:p.D360Y,ENSG00000144283:ENST00000389759:exon7:c.G1078T:p.D360Y	rs147567711	D;S	0,00058

PKP4	2_159481865_A_G	ENSG00000144283:ENST00000389757:exon7:c.A1079G:p.D360G,ENSG00000144283:ENST00000389759:exon7:c.A1079G:p.D360G	rs147809285	D	0,00058
PKP4	2_159497153_G_A	ENSG00000144283:ENST00000389757:exon10:c.G1577A:p.R526H,ENSG00000144283:ENST00000389759:exon10:c.G1577A:p.R526H	rs144253065	D;S	0,00057
PKP4	2_159499094_G_A	ENSG00000144283:ENST00000389757:exon11:c.G1792A:p.V598I,ENSG00000144283:ENST00000389759:exon11:c.G1792A:p.V598I		N	0,00057
PKP4	2_159499181_A_G	ENSG00000144283:ENST00000389757:exon11:c.A1879G:p.I627V,ENSG00000144283:ENST00000389759:exon11:c.A1879G:p.I627V		N	0,00057
PKP4	2_159517919_A_G	ENSG00000144283:ENST00000389757:exon13:c.A2168G:p.Y723C,ENSG00000144283:ENST00000389759:exon13:c.A2168G:p.Y723C	rs150425961	D;S	0,00057
PKP4	2_159519410_C_T	ENSG00000144283:ENST00000389757:exon14:c.C2213T:p.T738M,ENSG00000144283:ENST00000389759:exon14:c.C2213T:p.T738M	rs201257617	D;S	0,00058
PKP4	2_159519557_G_A	ENSG00000144283:ENST00000389757:exon14:c.G2360A:p.G787E,ENSG00000144283:ENST00000389759:exon14:c.G2360A:p.G787E		S	0,00057
PKP4	2_159519581_G_A	ENSG00000144283:ENST00000389757:exon14:c.G2384A:p.R795K,ENSG00000144283:ENST00000389759:exon14:c.G2384A:p.R795K	rs139221917	D	0,00115
PKP4	2_159519846_G_T	ENSG00000144283:ENST00000389757:exon15:c.G2466T:p.W822C,ENSG00000144283:ENST00000389759:exon15:c.G2466T:p.W822C		S	0,00058
PKP4	2_159523058_G_A	ENSG00000144283:ENST00000389757:exon16:c.G2711A:p.R904H,ENSG00000144283:ENST00000389759:exon16:c.G2711A:p.R904H		S	0,00057
PKP4	2_159530233_C_T	ENSG00000144283:ENST00000389757:exon18:c.C2969T:p.T990I,ENSG00000144283:ENST00000389759:exon18:c.C2969T:p.T990I	rs190318071	D	0,00057
PKP4	2_159530257_G_A	ENSG00000144283:ENST00000389757:exon18:c.G2993A:p.R998Q,ENSG00000144283:ENST00000389759:exon18:c.G2993A:p.R998Q		S	0,00057
PKP4	2_159536972_C_G	ENSG00000144283:ENST00000389757:exon21:c.C3233G:p.S1078C,ENSG00000144283:ENST00000389759:exon22:c.C3362G:p.S1121C		S	0,00114
PKP4	2_159537050_G_A	ENSG00000144283:ENST00000389757:exon21:c.G3311A:p.R1104Q,ENSG00000144283:ENST00000389759:exon22:c.G3440A:p.R1147Q		N	0,00057
PLN	6_118879986_A_T			L	0,00057
PLN	6_118880110_G_A	ENSG00000198523:ENST00000357525:exon2:c.G26A:p.R9H		S	0,00057
PLN	6_118880137_T_C	ENSG00000198523:ENST00000357525:exon2:c.T53C:p.I18T		N	0,00057
PLN	6_118880145_-_CT	ENSG00000198523:ENST00000357525:exon2:c.61_62insCT:p.P21fs		L	0,00057
PLN	6_118880157_C_T	ENSG00000198523:ENST00000357525:exon2:c.C73T:p.R25C		N	0,00057
PLN	6_118880158_G_A	ENSG00000198523:ENST00000357525:exon2:c.G74A:p.R25H		N	0,00057

PLN	6_118880236_T_C	ENSG00000198523:ENST00000357525:exon2:c.T152C:p.L51P		S	0,00057
PNN	14_39646638_A_G	ENSG00000100941:ENST00000216832:exon4:c.A277G:p.R93G,ENSG00000100941:ENST00000553331:exon4:c.A277G:p.R93G,ENSG00000100941:ENST00000556530:exon4:c.A277G:p.R93G		N	0,00058
PNN	14_39649827_C_T	ENSG00000100941:ENST00000216832:exon9:c.C914T:p.A305V	rs140529795	D	0,00057
PNN	14_39650139_A_G	ENSG00000100941:ENST00000216832:exon9:c.A1226G:p.N409S	rs143201887	D	0,00057
PNN	14_39650169_A_G	ENSG00000100941:ENST00000216832:exon9:c.A1256G:p.N419S	rs145173115	D	0,00057
PNN	14_39650261_A_G	ENSG00000100941:ENST00000216832:exon9:c.A1348G:p.S450G	rs145827544	D	0,00057
PNN	14_39650320_A_T	ENSG00000100941:ENST00000216832:exon9:c.A1407T:p.Q469H	rs148193612	D	0,00058
PNN	14_39650334_CTCAAC_-	ENSG00000100941:ENST00000216832:exon9:c.1421_1426del:p.474_476del		N	0,00058
PNN	14_39650338_ACCTCA_-	ENSG00000100941:ENST00000216832:exon9:c.1425_1430del:p.475_477del		N	0,00058
PNN	14_39650559_A_C	ENSG00000100941:ENST00000216832:exon9:c.A1646C:p.H549P		N	0,00057
PNN	14_39650883_C_G	ENSG00000100941:ENST00000216832:exon9:c.C1970G:p.T657S		N	0,00057
RBM20	10_112404334_-_GCA	ENSG00000203867:ENST00000369519:exon1:c.122_123insGCA:p.M41delinsMQ		N	0,01980
RBM20	10_112404342_-_AGC	ENSG00000203867:ENST00000369519:exon1:c.130_131insAGC:p.P44delinsQP		N	0,01754
RBM20	10_112541047_G_T	ENSG00000203867:ENST00000369519:exon2:c.G680T:p.G227V	rs202238753	D	0,00058
RBM20	10_112541290_T_C	ENSG00000203867:ENST00000369519:exon2:c.T923C:p.V308A		N	0,00059
RBM20	10_112541394_C_T	ENSG00000203867:ENST00000369519:exon2:c.C1027T:p.H343Y	rs112226602	D	0,00058
RBM20	10_112541424_G_A	ENSG00000203867:ENST00000369519:exon2:c.G1057A:p.E353K		S	0,00058
RBM20	10_112541460_G_A	ENSG00000203867:ENST00000369519:exon2:c.G1093A:p.G365R	rs201047984	D;S	0,00058
RBM20	10_112541502_G_A	ENSG00000203867:ENST00000369519:exon2:c.G1135A:p.G379R	rs199842148	D	0,00057
RBM20	10_112541511_G_T	ENSG00000203867:ENST00000369519:exon2:c.G1144T:p.A382S		N	0,00057
RBM20	10_112541568_G_A	ENSG00000203867:ENST00000369519:exon2:c.G1201A:p.D401N		S	0,00057
RBM20	10_112541632_T_C	ENSG00000203867:ENST00000369519:exon2:c.T1265C:p.F422S		S	0,00057
RBM20	10_112544571_C_T	ENSG00000203867:ENST00000369519:exon5:c.C1451T:p.T484I	rs116442272	D	0,00057
RBM20	10_112557371_G_A	ENSG00000203867:ENST00000369519:exon6:c.G1633A:p.V545I		N	0,00057
RBM20	10_112559642_G_A	ENSG00000203867:ENST00000369519:exon7:c.G1766A:p.R589Q		N	0,00115
RBM20	10_112572113_C_T	ENSG00000203867:ENST00000369519:exon9:c.C1958T:p.T653I		S	0,00060

RBM20	10_112572169_G_A	ENSG00000203867:ENST00000369519:exon9:c.G2014A:p.G672S		N	0,00061
RBM20	10_112572271_C_A	ENSG00000203867:ENST00000369519:exon9:c.C2116A:p.P706T		N	0,00059
RBM20	10_112572356_G_A	ENSG00000203867:ENST00000369519:exon9:c.G2201A:p.R734Q		S	0,00059
RBM20	10_112572368_C_T	ENSG00000203867:ENST00000369519:exon9:c.C2213T:p.P738L		N	0,00059
RBM20	10_112572394_C_T	ENSG00000203867:ENST00000369519:exon9:c.C2239T:p.H747Y		S	0,00059
RBM20	10_112572419_G_A	ENSG00000203867:ENST00000369519:exon9:c.G2264A:p.R755H		S	0,00058
RBM20	10_112572473_A_G	ENSG00000203867:ENST00000369519:exon9:c.A2318G:p.K773R	rs181769913	D	0,00058
RBM20	10_112572488_C_T	ENSG00000203867:ENST00000369519:exon9:c.C2333T:p.A778V		N	0,00058
RBM20	10_112581039_G_A	ENSG00000203867:ENST00000369519:exon11:c.G2662A:p.D888N	rs201370621	D;P	0,00344
RBM20	10_112581264_A_G	ENSG00000203867:ENST00000369519:exon11:c.A2887G:p.K963E		N	0,00057
RBM20	10_112581282_G_A	ENSG00000203867:ENST00000369519:exon11:c.G2905A:p.V969I		N	0,00057
RBM20	10_112581381_C_G	ENSG00000203867:ENST00000369519:exon11:c.C3004G:p.L1002V		N	0,00115
RBM20	10_112581391_A_G	ENSG00000203867:ENST00000369519:exon11:c.A3014G:p.D1005G		S	0,00057
RBM20	10_112581400_G_A	ENSG00000203867:ENST00000369519:exon11:c.G3023A:p.R1008Q		N	0,00115
RBM20	10_112581424_G_C	ENSG00000203867:ENST00000369519:exon11:c.G3047C:p.G1016A		S	0,00057
RBM20	10_112581492_C_T	ENSG00000203867:ENST00000369519:exon11:c.C3115T:p.P1039S		N	0,00057
RBM20	10_112581642_C_G	ENSG00000203867:ENST00000369519:exon11:c.C3265G:p.P1089A	rs147356378	D	0,00058
RBM20	10_112581643_C_G	ENSG00000203867:ENST00000369519:exon11:c.C3266G:p.P1089R		N	0,00057
RYR2	1_237538086_G_A	ENSG00000198626:ENST00000542537:exon6:c.G406A:p.D136N,ENSG00000198626:ENST00000366574:exon7:c.G454A:p.D152N,ENSG00000198626:ENST00000360064:exon8:c.G448A:p.D150N		N	0,00058
RYR2	1_237608773_A_G	ENSG00000198626:ENST00000542537:exon13:c.A1195G:p.T399A,ENSG00000198626:ENST00000366574:exon14:c.A1243G:p.T415A,ENSG00000198626:ENST00000360064:exon15:c.A1237G:p.T413A		N	0,00057
RYR2	1_237619933_C_G	ENSG00000198626:ENST00000542537:exon15:c.C1462G:p.R488G,ENSG00000198626:ENST00000366574:exon16:c.C1510G:p.R504G,ENSG00000198626:ENST00000360064:exon17:c.C1504G:p.R502G		S	0,00057
RYR2	1_237619982_G_T	ENSG00000198626:ENST00000542537:exon15:c.G1511T:p.R504L,ENSG00000198626:ENST00000366574:exon16:c.G1559T:p.R520L,ENSG00000198626:ENST00000360064:exon17:c.G1553T:p.R518L		N	0,00057
RYR2	1_237659825_T_G	ENSG00000198626:ENST00000542537:exon19:c.T1928G:p.I643S,ENSG00000198626:ENST00000366574:exon20:c.T1976G:p.I659S,ENSG00000198626:ENST00000360064:exon21:c.T1970G:p.I657S		S	0,00057

RYR2	1_237659929_C_T	ENSG00000198626:ENST00000542537:exon19:c.C2032T:p.R678X,ENSG00000198626:ENST00000366574:exon20:c.C2080T:p.R694X,ENSG00000198626:ENST00000360064:exon21:c.C2074T:p.R692X		L	0,00057
RYR2	1_237659953_G_A	ENSG00000198626:ENST00000542537:exon19:c.G2056A:p.G686R,ENSG00000198626:ENST00000366574:exon20:c.G2104A:p.G702R,ENSG00000198626:ENST00000360064:exon21:c.G2098A:p.G700R		S	0,00057
RYR2	1_237660057_G_A	ENST00000366574:exon20:c.2203+5G>A,ENST00000360064:exon21:c.2197+5G>A,ENST00000542537:exon19:c.2155+5G>A		L	0,00057
RYR2	1_237664074_G_A	ENSG00000198626:ENST00000542537:exon20:c.G2219A:p.S740N,ENSG00000198626:ENST00000366574:exon21:c.G2267A:p.S756N,ENSG00000198626:ENST00000360064:exon22:c.G2261A:p.S754N	rs193922623	D;S	0,00057
RYR2	1_237664112_C_G	ENSG00000198626:ENST00000542537:exon20:c.C2257G:p.R753G,ENSG00000198626:ENST00000366574:exon21:c.C2305G:p.R769G,ENSG00000198626:ENST00000360064:exon22:c.C2299G:p.R767G		S	0,00057
RYR2	1_237664149_A_G	ENSG00000198626:ENST00000542537:exon20:c.A2294G:p.N765S,ENSG00000198626:ENST00000366574:exon21:c.A2342G:p.N781S,ENSG00000198626:ENST00000360064:exon22:c.A2336G:p.N779S		S	0,00057
RYR2	1_237666593_C_T	ENSG00000198626:ENST00000542537:exon21:c.C2353T:p.R785C,ENSG00000198626:ENST00000366574:exon22:c.C2401T:p.R801C,ENSG00000198626:ENST00000360064:exon23:c.C2395T:p.R799C		S	0,00057
RYR2	1_237666731_A_G	ENSG00000198626:ENST00000542537:exon21:c.A2491G:p.T831A,ENSG00000198626:ENST00000366574:exon22:c.A2539G:p.T847A,ENSG00000198626:ENST00000360064:exon23:c.A2533G:p.T845A		N	0,00057
RYR2	1_237670112_C_G	ENSG00000198626:ENST00000542537:exon22:c.C2668G:p.P890A,ENSG00000198626:ENST00000366574:exon23:c.C2716G:p.P906A,ENSG00000198626:ENST00000360064:exon24:c.C2710G:p.P904A		S	0,00057
RYR2	1_237693732_T_C	ENSG00000198626:ENST00000542537:exon24:c.T2780C:p.L927S,ENSG00000198626:ENST00000366574:exon25:c.T2828C:p.L943S,ENSG00000198626:ENST00000360064:exon26:c.T2822C:p.L941S		S	0,00058
RYR2	1_237711759_G_T	ENSG00000198626:ENST00000542537:exon25:c.G2887T:p.A963S,ENSG00000198626:ENST00000366574:exon26:c.G2935T:p.A979S,ENSG00000198626:ENST00000360064:exon27:c.G2929T:p.A977S	rs202015519	D	0,00057
RYR2	1_237711861_C_T	ENSG00000198626:ENST00000542537:exon25:c.C2989T:p.R997W,ENSG00000198626:ENST00000366574:exon26:c.C3037T:p.R1013W,ENSG00000198626:ENST00000360064:exon27:c.C3031T:p.R1011W		N	0,00057
RYR2	1_237711862_G_A	ENSG00000198626:ENST00000542537:exon25:c.G2990A:p.R997Q,ENSG00000198626:ENST00000366574:exon26:c.G3038A:p.R1013Q,ENSG00000198626:ENST00000360064:exon27:c.G3032A:p.R1011Q	rs149514924	D;P	0,00115
RYR2	1_237713928_C_T	ENSG00000198626:ENST00000542537:exon26:c.C3103T:p.R1035C,ENSG00000198626:ENST00000366574:exon27:c.C3151T:p.R1051C,ENSG00000198626:ENST00000360064:exon28:c.C3145T:p.R1049C		N	0,00057

RYR2	1_237713940_C_T	ENSG00000198626:ENST00000542537:exon26:c.C3115T:p.R1039C,ENSG00000198626:ENST00000366574:exon27:c.C3163T:p.R1055C,ENSG00000198626:ENST00000360064:exon28:c.C3157T:p.R1053C		S	0,00057
RYR2	1_237730008_G_A	ENSG00000198626:ENST00000542537:exon27:c.G3308A:p.R1103H,ENSG00000198626:ENST00000366574:exon28:c.G3356A:p.R1119H,ENSG00000198626:ENST00000360064:exon29:c.G3350A:p.R1117H	rs201312753	D;S	0,00116
RYR2	1_237732502_G_C	ENSG00000198626:ENST00000542537:exon28:c.G3433C:p.V1145L,ENSG00000198626:ENST00000366574:exon29:c.G3481C:p.V1161L,ENSG00000198626:ENST00000360064:exon30:c.G3475C:p.V1159L		S	0,00057
RYR2	1_237754201_G_C	ENSG00000198626:ENST00000542537:exon30:c.G4021C:p.D1341H,ENSG00000198626:ENST00000366574:exon31:c.G4069C:p.D1357H,ENSG00000198626:ENST00000360064:exon32:c.G4063C:p.D1355H	rs193922626	D	0,00114
RYR2	1_237758826_T_C	ENSG00000198626:ENST00000542537:exon33:c.T4417C:p.C1473R,ENSG00000198626:ENST00000366574:exon34:c.T4465C:p.C1489R,ENSG00000198626:ENST00000360064:exon35:c.T4459C:p.C1487R	rs200450676	D;S	0,00058
RYR2	1_237777530_G_C	ENSG00000198626:ENST00000542537:exon36:c.G5054C:p.G1685A,ENSG00000198626:ENST00000366574:exon37:c.G5102C:p.G1701A,ENSG00000198626:ENST00000360064:exon38:c.G5096C:p.G1699A		S	0,00057
RYR2	1_237777694_T_G	ENSG00000198626:ENST00000542537:exon36:c.T5218G:p.S1740A,ENSG00000198626:ENST00000366574:exon37:c.T5266G:p.S1756A,ENSG00000198626:ENST00000360064:exon38:c.T5260G:p.S1754A		S	0,00057
RYR2	1_237777722_C_G	ENSG00000198626:ENST00000542537:exon36:c.C5246G:p.S1749C,ENSG00000198626:ENST00000366574:exon37:c.C5294G:p.S1765C,ENSG00000198626:ENST00000360064:exon38:c.C5288G:p.S1763C		S	0,00057
RYR2	1_237778090_C_T	ENSG00000198626:ENST00000542537:exon36:c.C5614T:p.R1872W,ENSG00000198626:ENST00000366574:exon37:c.C5662T:p.R1888W,ENSG00000198626:ENST00000360064:exon38:c.C5656T:p.R1886W		N	0,00058
RYR2	1_237778108_C_A	ENSG00000198626:ENST00000542537:exon36:c.C5632A:p.L1878I,ENSG00000198626:ENST00000366574:exon37:c.C5680A:p.L1894I,ENSG00000198626:ENST00000360064:exon38:c.C5674A:p.L1892I		S	0,00057
RYR2	1_237787064_G_C	ENST00000366574:exon39:c.5917-1G>C,ENST00000360064:exon40:c.5911-1G>C,ENST00000542537:exon38:c.5869-1G>C		L	0,00057
RYR2	1_237794736_G_A	ENSG00000198626:ENST00000542537:exon41:c.G6402A:p.M2134I,ENSG00000198626:ENST00000366574:exon42:c.G6450A:p.M2150I,ENSG00000198626:ENST00000360064:exon43:c.G6444A:p.M2148I		N	0,00057

RYR2	1_237813288_G_A	ENSG00000198626:ENST00000542537:exon49:c.G7576A:p.A2526T,ENSG00000198626:ENST00000366574:exon50:c.G7624A:p.A2542T,ENSG00000198626:ENST00000360064:exon51:c.G7618A:p.A2540T		N	0,00057
RYR2	1_237814727_A_G	ENSG00000198626:ENST00000542537:exon50:c.A7702G:p.M2568V,ENSG00000198626:ENST00000366574:exon51:c.A7750G:p.M2584V,ENSG00000198626:ENST00000360064:exon52:c.A7744G:p.M2582V		N	0,00057
RYR2	1_237821276_T_C	ENSG00000198626:ENST00000542537:exon53:c.T8114C:p.I2705T,ENSG00000198626:ENST00000366574:exon54:c.T8162C:p.I2721T,ENSG00000198626:ENST00000360064:exon55:c.T8156C:p.I2719T	rs201500134	D	0,00230
RYR2	1_237823350_G_T	ENSG00000198626:ENST00000542537:exon54:c.G8226T:p.K2742N,ENSG00000198626:ENST00000366574:exon55:c.G8274T:p.K2758N,ENSG00000198626:ENST00000360064:exon56:c.G8268T:p.K2756N		S	0,00057
RYR2	1_237868518_G_T	ENSG00000198626:ENST00000540213:exon5:c.G440T:p.R147L,ENSG00000198626:ENST00000542288:exon6:c.G320T:p.R107L,ENSG00000198626:ENST00000542537:exon66:c.G9407T:p.R3136L,ENSG00000198626:ENST00000366574:exon67:c.G9455T:p.R3152L,ENSG00000198626:ENST00000360064:exon68:c.G9449T:p.R3150L		N	0,00057
RYR2	1_237868623_A_G	ENSG00000198626:ENST00000540213:exon5:c.A545G:p.K182R,ENSG00000198626:ENST00000542288:exon6:c.A425G:p.K142R,ENSG00000198626:ENST00000542537:exon66:c.A9512G:p.K3171R,ENSG00000198626:ENST00000366574:exon67:c.A9560G:p.K3187R,ENSG00000198626:ENST00000360064:exon68:c.A9554G:p.K3185R	rs184218219	D;S	0,00057
RYR2	1_237868632_G_A	ENSG00000198626:ENST00000540213:exon5:c.G554A:p.R185Q,ENSG00000198626:ENST00000542288:exon6:c.G434A:p.R145Q,ENSG00000198626:ENST00000542537:exon66:c.G9521A:p.R3174Q,ENSG00000198626:ENST00000366574:exon67:c.G9569A:p.R3190Q,ENSG00000198626:ENST00000360064:exon68:c.G9563A:p.R3188Q		S	0,00057
RYR2	1_237875041_T_C	ENST00000366574:exon71:c.10231-4T>C,ENST00000360064:exon72:c.10225-4T>C,ENST00000542537:exon70:c.10183-4T>C,ENST00000542288:exon10:c.1096-4T>C,ENST00000540213:exon9:c.1216-4T>C	rs117180147	D;L	0,00058
RYR2	1_237886554_C_G	ENSG00000198626:ENST00000542288:exon13:c.C1546G:p.L516V,ENSG00000198626:ENST00000542537:exon73:c.C10633G:p.L3545V,ENSG00000198626:ENST00000366574:exon74:c.C10681G:p.L3561V,ENSG00000198626:ENST00000360064:exon75:c.C10675G:p.L3559V		N	0,00057
RYR2	1_237947785_T_A	ENSG00000198626:ENST00000542288:exon30:c.T3695A:p.M1232K,ENSG00000198626:ENST00000542537:exon89:c.T12725A:p.M4242K,ENSG00000198626:ENST00000366574:exon90:c.T12773A:p.M4258K,ENSG00000198626:ENST00000360064:exon92:c.T12791A:p.M4264K		N	0,00058
RYR2	1_237948105_G_A	ENSG00000198626:ENST00000542288:exon30:c.G4015A:p.D1339N,ENSG00000198626:ENST00000542537:exon89:c.G13045A:p.D4349N,ENSG00000198626:ENST00000366574:exon90:c.G13093A:p.D4365N,ENSG00000198626:ENST00000360064:exon92:c.G13111A:p.D4371N		N	0,00058

RYR2	1_237948273_-_TAAT	ENST00000366574:exon90:c.13260+1->TAAT,ENST00000360064:exon92:c.13278+1->TAAT,ENST00000542537:exon89:c.13212+1->TAAT,ENST00000542288:exon30:c.4182+1->TAAT		L	0,00057
RYR2	1_237961442_C_G	ENSG00000198626:ENST00000536033:exon5:c.C361G:p.P121A,ENSG00000198626:ENST00000542537:exon96:c.C14014G:p.P4672A,ENSG00000198626:ENST00000366574:exon97:c.C14062G:p.P4688A,ENSG00000198626:ENST00000360064:exon99:c.C14080G:p.P4694A		N	0,00057
RYR2	1_237969434_T_C	ENST00000366574:exon99:c.14152-3T>C,ENST00000360064:exon101:c.14170-3T>C,ENST00000542537:exon98:c.14104-3T>C,ENST00000536033:exon7:c.451-3T>C		L	0,00058
SCN5A	3_38591847_G_C	ENSG00000183873:ENST00000414099:exon26:c.C5962G:p.P1988A,ENSG00000183873:ENST00000449557:exon26:c.C5854G:p.P1952A,ENSG00000183873:ENST00000450102:exon26:c.C5854G:p.P1952A,ENSG00000183873:ENST00000423572:exon27:c.C6013G:p.P2005A,ENSG00000183873:ENST00000425664:exon27:c.C5962G:p.P1988A,ENSG00000183873:ENST00000451551:exon27:c.C5854G:p.P1952A,ENSG00000183873:ENST00000455624:exon27:c.C5917G:p.P1973A,ENSG00000183873:ENST00000333535:exon28:c.C6016G:p.P2006A,ENSG00000183873:ENST00000413689:exon28:c.C6016G:p.P2006A,ENSG00000183873:ENST00000443581:exon28:c.C6013G:p.P2005A	rs45489199	D	0,00060
SCN5A	3_38591895_C_G	ENSG00000183873:ENST00000414099:exon26:c.G5914C:p.V1972L,ENSG00000183873:ENST00000449557:exon26:c.G5806C:p.V1936L,ENSG00000183873:ENST00000450102:exon26:c.G5806C:p.V1936L,ENSG00000183873:ENST00000423572:exon27:c.G5965C:p.V1989L,ENSG00000183873:ENST00000425664:exon27:c.G5914C:p.V1972L,ENSG00000183873:ENST00000451551:exon27:c.G5806C:p.V1936L,ENSG00000183873:ENST00000455624:exon27:c.G5869C:p.V1957L,ENSG00000183873:ENST00000333535:exon28:c.G5968C:p.V1990L,ENSG00000183873:ENST00000413689:exon28:c.G5968C:p.V1990L,ENSG00000183873:ENST00000443581:exon28:c.G5965C:p.V1989L		N	0,00060
SCN5A	3_38591978_G_A	ENSG00000183873:ENST00000414099:exon26:c.C5831T:p.P1944L,ENSG00000183873:ENST00000449557:exon26:c.C5723T:p.P1908L,ENSG00000183873:ENST00000450102:exon26:c.C5723T:p.P1908L,ENSG00000183873:ENST00000423572:exon27:c.C5882T:p.P1961L,ENSG00000183873:ENST00000425664:exon27:c.C5831T:p.P1944L,ENSG00000183873:ENST00000451551:exon27:c.C5723T:p.P1908L,ENSG00000183873:ENST00000455624:exon27:c.C5786T:p.P1929L,ENSG00000183873:ENST00000333535:exon28:c.C5885T:p.P1962L,ENSG00000183873:ENST00000413689:exon28:c.C5885T:p.P1962L,ENSG00000183873:ENST00000443581:exon28:c.C5882T:p.P1961L	rs199473638	D;P	0,00061
SCN5A	3_38592120_G_A	ENSG00000183873:ENST00000414099:exon26:c.C5689T:p.H1897Y,ENSG00000183873:ENST00000449557:exon26:c.C5581T:p.H1861Y,ENSG00000183873:ENST00000450102:exon26:c.C5581T:p.H1861Y,ENSG00000183873:ENST00000423572:exon27:c.C5740T:p.H1914Y,ENSG00000183873:ENST00000425664:exon27:c.C5689T:p.H1897Y,ENSG00000183873:ENST00000451551:exon27:c.C5581T:p.H1861Y,ENSG00000183873:ENST00000455624:exon27:c.C5644T:p.H1882Y,ENSG00000183873:ENST00000333535:exon28:c.C5743T:p.H1915Y,ENSG00000183873:ENST00000413689:exon28:c.C5743T:p.H1915Y,ENSG00000183873:ENST00000443581:exon28:c.C5740T:p.H1914Y		N	0,00058

SCN5A	3_38592408_C_T	ENSG00000183873:ENST00000414099:exon26:c.G5401A:p.D1801N,ENSG00000183873:ENST00000449557:exon26:c.G5293A:p.D1765N,ENSG00000183873:ENST00000450102:exon26:c.G5293A:p.D1765N,ENSG00000183873:ENST00000423572:exon27:c.G5452A:p.D1818N,ENSG00000183873:ENST00000425664:exon27:c.G5401A:p.D1801N,ENSG00000183873:ENST00000451551:exon27:c.G5293A:p.D1765N,ENSG00000183873:ENST00000455624:exon27:c.G5356A:p.D1786N,ENSG00000183873:ENST00000333535:exon28:c.G5455A:p.D1819N,ENSG00000183873:ENST00000413689:exon28:c.G5455A:p.D1819N,ENSG00000183873:ENST00000443581:exon28:c.G5452A:p.D1818N	rs137854619	D;P	0,00058
SCN5A	3_38592503_C_T	ENSG00000183873:ENST00000414099:exon26:c.G5306A:p.S1769N,ENSG00000183873:ENST00000449557:exon26:c.G5198A:p.S1733N,ENSG00000183873:ENST00000450102:exon26:c.G5198A:p.S1733N,ENSG00000183873:ENST00000423572:exon27:c.G5357A:p.S1786N,ENSG00000183873:ENST00000425664:exon27:c.G5306A:p.S1769N,ENSG00000183873:ENST00000451551:exon27:c.G5198A:p.S1733N,ENSG00000183873:ENST00000455624:exon27:c.G5261A:p.S1754N,ENSG00000183873:ENST00000333535:exon28:c.G5360A:p.S1787N,ENSG00000183873:ENST00000413689:exon28:c.G5360A:p.S1787N,ENSG00000183873:ENST00000443581:exon28:c.G5357A:p.S1786N	rs199473316	D;P	0,00172
SCN5A	3_38592527_G_A	ENSG00000183873:ENST00000414099:exon26:c.C5282T:p.T1761M,ENSG00000183873:ENST00000449557:exon26:c.C5174T:p.T1725M,ENSG00000183873:ENST00000450102:exon26:c.C5174T:p.T1725M,ENSG00000183873:ENST00000423572:exon27:c.C5333T:p.T1778M,ENSG00000183873:ENST00000425664:exon27:c.C5282T:p.T1761M,ENSG00000183873:ENST00000451551:exon27:c.C5174T:p.T1725M,ENSG00000183873:ENST00000455624:exon27:c.C5237T:p.T1746M,ENSG00000183873:ENST00000333535:exon28:c.C5336T:p.T1779M,ENSG00000183873:ENST00000413689:exon28:c.C5336T:p.T1779M,ENSG00000183873:ENST00000443581:exon28:c.C5333T:p.T1778M	rs199473634	D;S	0,00058
SCN5A	3_38597928_G_A	ENST00000414099:exon24:c.4383+4C>T,ENST00000425664:exon25:c.4383+4C>T,ENST00000443581:exon26:c.4434+4C>T,ENST00000451551:exon25:c.4275+4C>T,ENST00000413689:exon26:c.4437+4C>T,ENST00000423572:exon25:c.4434+4C>T,ENST00000333535:exon26:c.4437+4C>T,ENST00000455624:exon25:c.4434+4C>T,ENST00000450102:exon24:c.4275+4C>T,ENST00000449557:exon24:c.4275+4C>T		L	0,00066
SCN5A	3_38598027_T_G	ENSG00000183873:ENST00000414099:exon23:c.A4288C:p.I1430L,ENSG00000183873:ENST00000449557:exon23:c.A4180C:p.I1394L,ENSG00000183873:ENST00000450102:exon23:c.A4180C:p.I1394L,ENSG00000183873:ENST00000423572:exon24:c.A4339C:p.I1447L,ENSG00000183873:ENST00000425664:exon24:c.A4288C:p.I1430L,ENSG00000183873:ENST00000451551:exon24:c.A4180C:p.I1394L,ENSG00000183873:ENST00000455624:exon24:c.A4339C:p.I1447L,ENSG00000183873:ENST00000333535:exon25:c.A4342C:p.I1448L,ENSG00000183873:ENST00000413689:exon25:c.A4342C:p.I1448L,ENSG00000183873:ENST00000443581:exon25:c.A4339C:p.I1447L	rs199473250	D	0,00059

SCN5A	3_38603947_G_A	ENSG00000183873:ENST00000449557:exon20:c.C3760T:p.L1254F,ENSG00000183873:ENST00000450102:exon20:c.C3760T:p.L1254F,ENSG00000183873:ENST00000414099:exon21:c.C3922T:p.L1308F,ENSG00000183873:ENST00000423572:exon21:c.C3919T:p.L1307F,ENSG00000183873:ENST00000451551:exon21:c.C3760T:p.L1254F,ENSG00000183873:ENST00000455624:exon21:c.C3919T:p.L1307F,ENSG00000183873:ENST00000333535:exon22:c.C3922T:p.L1308F,ENSG00000183873:ENST00000413689:exon22:c.C3922T:p.L1308F,ENSG00000183873:ENST00000425664:exon22:c.C3922T:p.L1308F,ENSG00000183873:ENST00000443581:exon22:c.C3919T:p.L1307F	rs41313031	D;P	0,00079
SCN5A	3_38603958_G_A	ENSG00000183873:ENST00000449557:exon20:c.C3749T:p.T1250M,ENSG00000183873:ENST00000450102:exon20:c.C3749T:p.T1250M,ENSG00000183873:ENST00000414099:exon21:c.C3911T:p.T1304M,ENSG00000183873:ENST00000423572:exon21:c.C3908T:p.T1303M,ENSG00000183873:ENST00000451551:exon21:c.C3749T:p.T1250M,ENSG00000183873:ENST00000455624:exon21:c.C3908T:p.T1303M,ENSG00000183873:ENST00000333535:exon22:c.C3911T:p.T1304M,ENSG00000183873:ENST00000413689:exon22:c.C3911T:p.T1304M,ENSG00000183873:ENST00000425664:exon22:c.C3911T:p.T1304M,ENSG00000183873:ENST00000443581:exon22:c.C3908T:p.T1303M	rs199473603	D;P	0,00153
SCN5A	3_38603991_A_G	ENSG00000183873:ENST00000449557:exon20:c.T3716C:p.F1239S,ENSG00000183873:ENST00000450102:exon20:c.T3716C:p.F1239S,ENSG00000183873:ENST00000414099:exon21:c.T3878C:p.F1293S,ENSG00000183873:ENST00000423572:exon21:c.T3875C:p.F1292S,ENSG00000183873:ENST00000451551:exon21:c.T3716C:p.F1239S,ENSG00000183873:ENST00000455624:exon21:c.T3875C:p.F1292S,ENSG00000183873:ENST00000333535:exon22:c.T3878C:p.F1293S,ENSG00000183873:ENST00000413689:exon22:c.T3878C:p.F1293S,ENSG00000183873:ENST00000425664:exon22:c.T3878C:p.F1293S,ENSG00000183873:ENST00000443581:exon22:c.T3875C:p.F1292S	rs41311127	D;P	0,00076
SCN5A	3_38607905_C_T	ENSG00000183873:ENST00000449557:exon19:c.G3673A:p.V1225I,ENSG00000183873:ENST00000450102:exon19:c.G3673A:p.V1225I,ENSG00000183873:ENST00000414099:exon20:c.G3835A:p.V1279I,ENSG00000183873:ENST00000423572:exon20:c.G3832A:p.V1278I,ENSG00000183873:ENST00000451551:exon20:c.G3673A:p.V1225I,ENSG00000183873:ENST00000455624:exon20:c.G3832A:p.V1278I,ENSG00000183873:ENST00000333535:exon21:c.G3835A:p.V1279I,ENSG00000183873:ENST00000413689:exon21:c.G3835A:p.V1279I,ENSG00000183873:ENST00000425664:exon21:c.G3835A:p.V1279I,ENSG00000183873:ENST00000443581:exon21:c.G3832A:p.V1278I	rs199473341	D;P	0,00059
SCN5A	3_38608022_C_G	ENSG00000183873:ENST00000449557:exon19:c.G3556C:p.E1186Q,ENSG00000183873:ENST00000450102:exon19:c.G3556C:p.E1186Q,ENSG00000183873:ENST00000414099:exon20:c.G3718C:p.E1240Q,ENSG00000183873:ENST00000423572:exon20:c.G3715C:p.E1239Q,ENSG00000183873:ENST00000451551:exon20:c.G3556C:p.E1186Q,ENSG00000183873:ENST00000455624:exon20:c.G3715C:p.E1239Q,ENSG00000183873:ENST00000333535:exon21:c.G3718C:p.E1240Q,ENSG00000183873:ENST00000413689:exon21:c.G3718C:p.E1240Q,ENSG00000183873:ENST00000425664:exon21:c.G3718C:p.E1240Q,ENSG00000183873:ENST00000443581:exon21:c.G3715C:p.E1239Q	rs199473211	D;P	0,00058

SCN5A	3_38608058_A_G	ENSG00000183873:ENST00000449557:exon19:c.T3520C:p.Y1174H,ENSG00000183873:ENST00000450102:exon19:c.T3520C:p.Y1174H,ENSG00000183873:ENST00000414099:exon20:c.T3682C:p.Y1228H,ENSG00000183873:ENST00000423572:exon20:c.T3679C:p.Y1227H,ENSG00000183873:ENST00000451551:exon20:c.T3520C:p.Y1174H,ENSG00000183873:ENST00000455624:exon20:c.T3679C:p.Y1227H,ENSG00000183873:ENST00000333535:exon21:c.T3682C:p.Y1228H,ENSG00000183873:ENST00000413689:exon21:c.T3682C:p.Y1228H,ENSG00000183873:ENST00000425664:exon21:c.T3682C:p.Y1228H,ENSG00000183873:ENST00000443581:exon21:c.T3679C:p.Y1227H	rs199473205	D;P	0,00059
SCN5A	3_38622640_A_G	ENSG00000183873:ENST00000414099:exon16:c.T3010C:p.C1004R,ENSG00000183873:ENST00000423572:exon16:c.T3010C:p.C1004R,ENSG00000183873:ENST00000449557:exon16:c.T3010C:p.C1004R,ENSG00000183873:ENST00000450102:exon16:c.T3010C:p.C1004R,ENSG00000183873:ENST00000455624:exon16:c.T3010C:p.C1004R,ENSG00000183873:ENST00000333535:exon17:c.T3010C:p.C1004R,ENSG00000183873:ENST00000413689:exon17:c.T3010C:p.C1004R,ENSG00000183873:ENST00000425664:exon17:c.T3010C:p.C1004R,ENSG00000183873:ENST00000443581:exon17:c.T3010C:p.C1004R,ENSG00000183873:ENST00000451551:exon17:c.T3010C:p.C1004R	rs199473183	D;P	0,00157
SCN5A	3_38622706_A_G	ENSG00000183873:ENST00000414099:exon16:c.T2944C:p.C982R,ENSG00000183873:ENST00000423572:exon16:c.T2944C:p.C982R,ENSG00000183873:ENST00000449557:exon16:c.T2944C:p.C982R,ENSG00000183873:ENST00000450102:exon16:c.T2944C:p.C982R,ENSG00000183873:ENST00000455624:exon16:c.T2944C:p.C982R,ENSG00000183873:ENST00000333535:exon17:c.T2944C:p.C982R,ENSG00000183873:ENST00000413689:exon17:c.T2944C:p.C982R,ENSG00000183873:ENST00000425664:exon17:c.T2944C:p.C982R,ENSG00000183873:ENST00000443581:exon17:c.T2944C:p.C982R,ENSG00000183873:ENST00000451551:exon17:c.T2944C:p.C982R	rs199473182	D;P	0,00080
SCN5A	3_38627537_G_T	ENST00000414099:exon16:c.2437-5C>A,ENST00000425664:exon17:c.2437-5C>A,ENST00000443581:exon17:c.2437-5C>A,ENST00000451551:exon17:c.2437-5C>A,ENST00000413689:exon17:c.2437-5C>A,ENST00000423572:exon16:c.2437-5C>A,ENST00000333535:exon17:c.2437-5C>A,ENST00000455624:exon16:c.2437-5C>A,ENST00000450102:exon16:c.2437-5C>A,ENST00000449557:exon16:c.2437-5C>A	rs72549411	D;L	0,00174
SCN5A	3_38628928_C_A	ENSG00000183873:ENST00000414099:exon14:c.G2399T:p.R800L,ENSG00000183873:ENST00000423572:exon14:c.G2399T:p.R800L,ENSG00000183873:ENST00000449557:exon14:c.G2399T:p.R800L,ENSG00000183873:ENST00000450102:exon14:c.G2399T:p.R800L,ENSG00000183873:ENST00000455624:exon14:c.G2399T:p.R800L,ENSG00000183873:ENST00000333535:exon15:c.G2399T:p.R800L,ENSG00000183873:ENST00000413689:exon15:c.G2399T:p.R800L,ENSG00000183873:ENST00000425664:exon15:c.G2399T:p.R800L,ENSG00000183873:ENST00000443581:exon15:c.G2399T:p.R800L,ENSG00000183873:ENST00000451551:exon15:c.G2399T:p.R800L		N	0,00070

SCN5A	3_38639408_G_T	ENSG00000183873:ENST00000414099:exon13:c.C2074A:p.Q692K,ENSG00000183873:ENST00000423572:exon13:c.C2074A:p.Q692K,ENSG00000183873:ENST00000449557:exon13:c.C2074A:p.Q692K,ENSG00000183873:ENST00000450102:exon13:c.C2074A:p.Q692K,ENSG00000183873:ENST00000455624:exon13:c.C2074A:p.Q692K,ENSG00000183873:ENST00000333535:exon14:c.C2074A:p.Q692K,ENSG00000183873:ENST00000413689:exon14:c.C2074A:p.Q692K,ENSG00000183873:ENST00000425664:exon14:c.C2074A:p.Q692K,ENSG00000183873:ENST00000443581:exon14:c.C2074A:p.Q692K,ENSG00000183873:ENST00000451551:exon14:c.C2074A:p.Q692K	rs45553235	D;P	0,00124
SCN5A	3_38639443_C_T	ENSG00000183873:ENST00000414099:exon13:c.G2039A:p.R680H,ENSG00000183873:ENST00000423572:exon13:c.G2039A:p.R680H,ENSG00000183873:ENST00000449557:exon13:c.G2039A:p.R680H,ENSG00000183873:ENST00000450102:exon13:c.G2039A:p.R680H,ENSG00000183873:ENST00000455624:exon13:c.G2039A:p.R680H,ENSG00000183873:ENST00000333535:exon14:c.G2039A:p.R680H,ENSG00000183873:ENST00000413689:exon14:c.G2039A:p.R680H,ENSG00000183873:ENST00000425664:exon14:c.G2039A:p.R680H,ENSG00000183873:ENST00000443581:exon14:c.G2039A:p.R680H,ENSG00000183873:ENST00000451551:exon14:c.G2039A:p.R680H	rs199473142	D;P	0,00061
SCN5A	3_38640418_C_T	ENSG00000183873:ENST00000414099:exon12:c.G2014A:p.A672T,ENSG00000183873:ENST00000423572:exon12:c.G2014A:p.A672T,ENSG00000183873:ENST00000449557:exon12:c.G2014A:p.A672T,ENSG00000183873:ENST00000450102:exon12:c.G2014A:p.A672T,ENSG00000183873:ENST00000455624:exon12:c.G2014A:p.A672T,ENSG00000183873:ENST00000333535:exon13:c.G2014A:p.A672T,ENSG00000183873:ENST00000413689:exon13:c.G2014A:p.A672T,ENSG00000183873:ENST00000425664:exon13:c.G2014A:p.A672T,ENSG00000183873:ENST00000443581:exon13:c.G2014A:p.A672T,ENSG00000183873:ENST00000451551:exon13:c.G2014A:p.A672T	rs199473140	D;P	0,00062
SCN5A	3_38640450_C_T	ENSG00000183873:ENST00000414099:exon12:c.G1982A:p.R661Q,ENSG00000183873:ENST00000423572:exon12:c.G1982A:p.R661Q,ENSG00000183873:ENST00000449557:exon12:c.G1982A:p.R661Q,ENSG00000183873:ENST00000450102:exon12:c.G1982A:p.R661Q,ENSG00000183873:ENST00000455624:exon12:c.G1982A:p.R661Q,ENSG00000183873:ENST00000333535:exon13:c.G1982A:p.R661Q,ENSG00000183873:ENST00000413689:exon13:c.G1982A:p.R661Q,ENSG00000183873:ENST00000425664:exon13:c.G1982A:p.R661Q,ENSG00000183873:ENST00000443581:exon13:c.G1982A:p.R661Q,ENSG00000183873:ENST00000451551:exon13:c.G1982A:p.R661Q		N	0,00067
SCN5A	3_38645332_GAGGGC_-	ENSG00000183873:ENST00000414099:exon11:c.1756_1761del:p.586_587del,ENSG00000183873:ENST00000423572:exon11:c.1756_1761del:p.586_587del,ENSG00000183873:ENST00000449557:exon11:c.1756_1761del:p.586_587del,ENSG00000183873:ENST00000450102:exon11:c.1756_1761del:p.586_587del,ENSG00000183873:ENST00000455624:exon11:c.1756_1761del:p.586_587del,ENSG00000183873:ENST00000333535:exon12:c.1756_1761del:p.586_587del,ENSG00000183873:ENST00000413689:exon12:c.1756_1761del:p.586_587del,ENSG00000183873:ENST00000425664:exon12:c.1756_1761del:p.586_587del,ENSG00000183873:ENST00000443581:exon12:c.1756_1761del:p.586_587del,ENSG00000183873:ENST00000451551:exon12:c.1756_1761del:p.586_587del		N	0,00062

SCN5A	3_38645333_AGGGCG_-	ENSG00000183873:ENST00000414099:exon11:c.1755_1760del:p.585_587del,ENSG00000183873:ENST00000423572:exon11:c.1755_1760del:p.585_587del,ENSG00000183873:ENST00000449557:exon11:c.1755_1760del:p.585_587del,ENSG00000183873:ENST00000450102:exon11:c.1755_1760del:p.585_587del,ENSG00000183873:ENST00000455624:exon11:c.1755_1760del:p.585_587del,ENSG00000183873:ENST00000333535:exon12:c.1755_1760del:p.585_587del,ENSG00000183873:ENST00000413689:exon12:c.1755_1760del:p.585_587del,ENSG00000183873:ENST00000425664:exon12:c.1755_1760del:p.585_587del,ENSG00000183873:ENST00000443581:exon12:c.1755_1760del:p.585_587del,ENSG00000183873:ENST00000451551:exon12:c.1755_1760del:p.585_587del		P	0,00062
SCN5A	3_38645438_C_G	ENSG00000183873:ENST00000414099:exon11:c.G1655C:p.G552A,ENSG00000183873:ENST00000423572:exon11:c.G1655C:p.G552A,ENSG00000183873:ENST00000449557:exon11:c.G1655C:p.G552A,ENSG00000183873:ENST00000450102:exon11:c.G1655C:p.G552A,ENSG00000183873:ENST00000455624:exon11:c.G1655C:p.G552A,ENSG00000183873:ENST00000333535:exon12:c.G1655C:p.G552A,ENSG00000183873:ENST00000413689:exon12:c.G1655C:p.G552A,ENSG00000183873:ENST00000425664:exon12:c.G1655C:p.G552A,ENSG00000183873:ENST00000443581:exon12:c.G1655C:p.G552A,ENSG00000183873:ENST00000451551:exon12:c.G1655C:p.G552A		N	0,00061
SCN5A	3_38646398_G_C	ENSG00000183873:ENST00000414099:exon10:c.C1340G:p.A447G,ENSG00000183873:ENST00000423572:exon10:c.C1340G:p.A447G,ENSG00000183873:ENST00000449557:exon10:c.C1340G:p.A447G,ENSG00000183873:ENST00000450102:exon10:c.C1340G:p.A447G,ENSG00000183873:ENST00000455624:exon10:c.C1340G:p.A447G,ENSG00000183873:ENST00000333535:exon11:c.C1340G:p.A447G,ENSG00000183873:ENST00000413689:exon11:c.C1340G:p.A447G,ENSG00000183873:ENST00000425664:exon11:c.C1340G:p.A447G,ENSG00000183873:ENST00000443581:exon11:c.C1340G:p.A447G,ENSG00000183873:ENST00000451551:exon11:c.C1340G:p.A447G	rs199473113	D	0,00057
SCN5A	3_38647444_C_T	ENSG00000183873:ENST00000414099:exon9:c.G1336A:p.E446K,ENSG00000183873:ENST00000423572:exon9:c.G1336A:p.E446K,ENSG00000183873:ENST00000449557:exon9:c.G1336A:p.E446K,ENSG00000183873:ENST00000450102:exon9:c.G1336A:p.E446K,ENSG00000183873:ENST00000455624:exon9:c.G1336A:p.E446K,ENSG00000183873:ENST00000333535:exon10:c.G1336A:p.E446K,ENSG00000183873:ENST00000413689:exon10:c.G1336A:p.E446K,ENSG00000183873:ENST00000425664:exon10:c.G1336A:p.E446K,ENSG00000183873:ENST00000443581:exon10:c.G1336A:p.E446K,ENSG00000183873:ENST00000451551:exon10:c.G1336A:p.E446K	rs199473339	D;P	0,00115
SCN5A	3_38655278_G_A	ENSG00000183873:ENST00000423572:exon5:c.C659T:p.T220I,ENSG00000183873:ENST00000449557:exon5:c.C659T:p.T220I,ENSG00000183873:ENST00000333535:exon6:c.C659T:p.T220I,ENSG00000183873:ENST00000443581:exon6:c.C659T:p.T220I	rs45620037	D;P	0,00116
SCN5A	3_38655318_T_C	ENSG00000183873:ENST00000423572:exon5:c.A619G:p.T207A,ENSG00000183873:ENST00000449557:exon5:c.A619G:p.T207A,ENSG00000183873:ENST00000333535:exon6:c.A619G:p.T207A,ENSG00000183873:ENST00000443581:exon6:c.A619G:p.T207A		N	0,00058

SCN5A	3_38662449_C_T	ENSG00000183873:ENST00000414099:exon4:c.G496A:p.A166T,ENSG00000183873:ENST00000423572:exon4:c.G496A:p.A166T,ENSG00000183873:ENST00000449557:exon4:c.G496A:p.A166T,ENSG00000183873:ENST00000450102:exon4:c.G496A:p.A166T,ENSG00000183873:ENST00000455624:exon4:c.G496A:p.A166T,ENSG00000183873:ENST00000333535:exon5:c.G496A:p.A166T,ENSG00000183873:ENST00000413689:exon5:c.G496A:p.A166T,ENSG00000183873:ENST00000425664:exon5:c.G496A:p.A166T,ENSG00000183873:ENST00000443581:exon5:c.G496A:p.A166T,ENSG00000183873:ENST00000451551:exon5:c.G496A:p.A166T	rs201232332	D;S	0,00060
SCN5A	3_38671821_C_G	ENSG00000183873:ENST00000414099:exon2:c.G373C:p.V125L,ENSG00000183873:ENST00000423572:exon2:c.G373C:p.V125L,ENSG00000183873:ENST00000449557:exon2:c.G373C:p.V125L,ENSG00000183873:ENST00000450102:exon2:c.G373C:p.V125L,ENSG00000183873:ENST00000455624:exon2:c.G373C:p.V125L,ENSG00000183873:ENST00000333535:exon3:c.G373C:p.V125L,ENSG00000183873:ENST00000413689:exon3:c.G373C:p.V125L,ENSG00000183873:ENST00000425664:exon3:c.G373C:p.V125L,ENSG00000183873:ENST00000443581:exon3:c.G373C:p.V125L,ENSG00000183873:ENST00000451551:exon3:c.G373C:p.V125L	rs199473059	D;P	0,00115
SCN5A	3_38674747_G_A	ENSG00000183873:ENST00000414099:exon1:c.C52T:p.R18W,ENSG00000183873:ENST00000423572:exon1:c.C52T:p.R18W,ENSG00000183873:ENST00000449557:exon1:c.C52T:p.R18W,ENSG00000183873:ENST00000450102:exon1:c.C52T:p.R18W,ENSG00000183873:ENST00000455624:exon1:c.C52T:p.R18W,ENSG00000183873:ENST00000327956:exon2:c.C52T:p.R18W,ENSG00000183873:ENST00000333535:exon2:c.C52T:p.R18W,ENSG00000183873:ENST00000413689:exon2:c.C52T:p.R18W,ENSG00000183873:ENST00000425664:exon2:c.C52T:p.R18W,ENSG00000183873:ENST00000443581:exon2:c.C52T:p.R18W,ENSG00000183873:ENST00000451551:exon2:c.C52T:p.R18W	rs199473044	D;P	0,00128
TCAP	17_37821616_G_A	ENSG00000173991:ENST00000309889:exon1:c.G4A:p.A2T,ENSG00000173991:ENST00000578283:exon1:c.G4A:p.A2T		N	0,00062
TCAP	17_37821644_C_T	ENSG00000173991:ENST00000309889:exon1:c.C32T:p.S11L,ENSG00000173991:ENST00000578283:exon1:c.C32T:p.S11L	rs45495192	D	0,00126
TCAP	17_37821645_GGA_-	ENSG00000173991:ENST00000309889:exon1:c.33_35del:p.11_12del,ENSG00000173991:ENST00000578283:exon1:c.33_35del:p.11_12del		N	0,00063
TCAP	17_37821649_GAG_-	ENSG00000173991:ENST00000309889:exon1:c.37_39del:p.13_13del,ENSG00000173991:ENST00000578283:exon1:c.37_39del:p.13_13del		P	0,00063
TCAP	17_37822175_G_T	ENSG00000173991:ENST00000309889:exon2:c.G317T:p.R106L,ENSG00000173991:ENST00000578283:exon3:c.G245T:p.R82L		N	0,00092
TCAP	17_37822195_C_T	ENSG00000173991:ENST00000309889:exon2:c.C337T:p.L113F,ENSG00000173991:ENST00000578283:exon3:c.C265T:p.L89F		N	0,00091
TCAP	17_37822211_C_T	ENSG00000173991:ENST00000309889:exon2:c.C353T:p.A118V,ENSG00000173991:ENST00000578283:exon3:c.C281T:p.A94V	rs143233087	D	0,00090
TGFB3	14_76429772_C_G	ENSG00000119699:ENST00000238682:exon5:c.G813C:p.K271N,ENSG00000119699:ENST00000556285:exon5:c.G813C:p.K271N	rs147601018	D	0,00058

TGFB3	14_76437523_C_T	ENSG00000119699:ENST00000238682:exon3:c.G592A:p.E198K,ENSG00000119699:ENST00000556285:exon3:c.G592A:p.E198K		N	0,00058
TGFB3	14_76437535_G_A	ENSG00000119699:ENST00000238682:exon3:c.C580T:p.R194W,ENSG00000119699:ENST00000556285:exon3:c.C580T:p.R194W		S	0,00058
TGFB3	14_76438059_C_T	ENSG00000119699:ENST00000238682:exon2:c.G355A:p.E119K,ENSG00000119699:ENST00000556285:exon2:c.G355A:p.E119K		N	0,00059
TGFB3	14_76446944_G_A	ENSG00000119699:ENST00000238682:exon1:c.C293T:p.S98L,ENSG00000119699:ENST00000556285:exon1:c.C293T:p.S98L	rs142047577	D	0,00059
TMEM43	3_14170982_G_A	ENSG00000170876:ENST00000306077:exon2:c.G83A:p.R28Q		S	0,00058
TMEM43	3_14170990_G_A	ENSG00000170876:ENST00000306077:exon2:c.G91A:p.E31K		N	0,00058
TMEM43	3_14172439_G_A	ENSG00000170876:ENST00000306077:exon3:c.G280A:p.A94T		N	0,00088
TMEM43	3_14175244_T_C	ENSG00000170876:ENST00000306077:exon7:c.T518C:p.M173T		S	0,00093
TMEM43	3_14176287_G_A	ENSG00000170876:ENST00000306077:exon8:c.G601A:p.D201N	rs138182276	D	0,00063
TMEM43	3_14177355_A_T	ENSG00000170876:ENST00000306077:exon10:c.A829T:p.T277S		N	0,00059
TNNC1	3_52485431_T_C	ENSG00000114854:ENST00000232975:exon5:c.A430G:p.N144D		N	0,00060
TNNC1	3_52485805_C_T	ENSG00000114854:ENST00000496590:exon3:c.G140A:p.G47E,ENSG00000114854:ENST00000232975:exon4:c.G272A:p.G91E		S	0,00057
TNNC1	3_52485879_G_A	ENST00000232975:exon5:c.203-5C>T,ENST00000496590:exon4:c.71-5C>T	rs142519988	D;L	0,00058
TNNC1	3_52486194_C_T	ENSG00000114854:ENST00000232975:exon3:c.G130A:p.V44M		S	0,00063
TNNI3	19_55663243_G_C	ENSG00000129991:ENST00000588882:exon5:c.C517G:p.L173V,ENSG00000129991:ENST00000344887:exon8:c.C592G:p.L198V		P	0,00057
TNNI3	19_55663249_C_T	ENSG00000129991:ENST00000588882:exon5:c.G511A:p.D171N,ENSG00000129991:ENST00000344887:exon8:c.G586A:p.D196N	rs104894727	D;P	0,00115
TNNI3	19_55663278_C_T	ENSG00000129991:ENST00000588882:exon5:c.G482A:p.R161Q,ENSG00000129991:ENST00000344887:exon8:c.G557A:p.R186Q		P	0,00057
TNNI3	19_55665421_C_T	ENSG00000129991:ENST00000588882:exon4:c.G451A:p.V151M,ENSG00000129991:ENST00000344887:exon7:c.G526A:p.V176M		S	0,00115
TNNI3	19_55665436_C_T	ENSG00000129991:ENST00000588882:exon4:c.G436A:p.A146T,ENSG00000129991:ENST00000344887:exon7:c.G511A:p.A171T	rs121917761	D;P	0,00057
TNNI3	19_55665462_C_T	ENSG00000129991:ENST00000588882:exon4:c.G410A:p.R137Q,ENSG00000129991:ENST00000344887:exon7:c.G485A:p.R162Q		P	0,00173
TNNI3	19_55665463_G_A	ENSG00000129991:ENST00000588882:exon4:c.C409T:p.R137W,ENSG00000129991:ENST00000344887:exon7:c.C484T:p.R162W		P	0,00115

TNNI3	19_55665477_G_A	ENSG00000129991:ENST00000588882:exon4:c.C395T:p.A132V,ENSG00000129991:ENST00000344887:exon7:c.C470T:p.A157V		P	0,00057
TNNI3	19_55665514_G_A	ENSG00000129991:ENST00000588882:exon4:c.C358T:p.R120W,ENSG00000129991:ENST00000344887:exon7:c.C433T:p.R145W	rs104894724	D;P	0,00229
TNNI3	19_55665525_C_T	ENSG00000129991:ENST00000588882:exon4:c.G347A:p.R116Q,ENSG00000129991:ENST00000344887:exon7:c.G422A:p.R141Q		P	0,00057
TNNI3	19_55667616_G_A	ENSG00000129991:ENST00000588882:exon2:c.C160T:p.R54C,ENSG00000129991:ENST00000344887:exon5:c.C235T:p.R79C	rs3729712	D	0,00082
TNNI3	19_55668007_T_A	ENSG00000129991:ENST00000586858:exon1:c.A39T:p.K13N,ENSG00000129991:ENST00000588882:exon1:c.A39T:p.K13N,ENSG00000129991:ENST00000344887:exon4:c.A114T:p.K38N		S	0,00062
TNNI3	19_55668029_G_T	ENSG00000129991:ENST00000586858:exon1:c.C17A:p.S6Y,ENSG00000129991:ENST00000588882:exon1:c.C17A:p.S6Y	rs139150276	D	0,00245
TNNT2	1_201328345_C_T	ENSG00000118194:ENST00000360372:exon14:c.G845A:p.W282X,ENSG00000118194:ENST00000367315:exon14:c.G851A:p.W284X,ENSG00000118194:ENST00000236918:exon15:c.G875A:p.W292X,ENSG00000118194:ENST00000367317:exon15:c.G860A:p.W287X,ENSG00000118194:ENST00000367320:exon15:c.G761A:p.W254X,ENSG00000118194:ENST00000367322:exon15:c.G851A:p.W284X,ENSG00000118194:ENST00000421663:exon15:c.G869A:p.W290X,ENSG00000118194:ENST00000367318:exon16:c.G860A:p.W287X,ENSG00000118194:ENST00000458432:exon16:c.G887A:p.W296X,ENSG00000118194:ENST00000509001:exon16:c.G860A:p.W287X		P	0,00061
TNNT2	1_201328348_C_T	ENSG00000118194:ENST00000360372:exon14:c.G842A:p.R281H,ENSG00000118194:ENST00000367315:exon14:c.G848A:p.R283H,ENSG00000118194:ENST00000236918:exon15:c.G872A:p.R291H,ENSG00000118194:ENST00000367317:exon15:c.G857A:p.R286H,ENSG00000118194:ENST00000367320:exon15:c.G758A:p.R253H,ENSG00000118194:ENST00000367322:exon15:c.G848A:p.R283H,ENSG00000118194:ENST00000421663:exon15:c.G866A:p.R289H,ENSG00000118194:ENST00000367318:exon16:c.G857A:p.R286H,ENSG00000118194:ENST00000458432:exon16:c.G884A:p.R295H,ENSG00000118194:ENST00000509001:exon16:c.G857A:p.R286H	rs141121678	D;P	0,00122
TNNT2	1_201328372_C_G	ENSG00000118194:ENST00000360372:exon14:c.G818C:p.R273P,ENSG00000118194:ENST00000367315:exon14:c.G824C:p.R275P,ENSG00000118194:ENST00000236918:exon15:c.G848C:p.R283P,ENSG00000118194:ENST00000367317:exon15:c.G833C:p.R278P,ENSG00000118194:ENST00000367320:exon15:c.G734C:p.R245P,ENSG00000118194:ENST00000367322:exon15:c.G824C:p.R275P,ENSG00000118194:ENST00000421663:exon15:c.G842C:p.R281P,ENSG00000118194:ENST00000367318:exon16:c.G833C:p.R278P,ENSG00000118194:ENST00000458432:exon16:c.G860C:p.R287P,ENSG00000118194:ENST00000509001:exon16:c.G833C:p.R278P		N	0,00062

TNNT2	1_201328373_G_A	ENSG00000118194:ENST00000360372:exon14:c.C817T:p.R273C,ENSG00000118194:ENST00000367315:exon14:c.C823T:p.R275C,ENSG00000118194:ENST00000236918:exon15:c.C847T:p.R283C,ENSG00000118194:ENST00000367317:exon15:c.C832T:p.R278C,ENSG00000118194:ENST00000367320:exon15:c.C733T:p.R245C,ENSG00000118194:ENST00000367322:exon15:c.C823T:p.R275C,ENSG00000118194:ENST00000421663:exon15:c.C841T:p.R281C,ENSG00000118194:ENST00000367318:exon16:c.C832T:p.R278C,ENSG00000118194:ENST00000458432:exon16:c.C859T:p.R287C,ENSG00000118194:ENST00000509001:exon16:c.C832T:p.R278C	rs121964857	D;P	0,00498
TNNT2	1_201328386_G_T	ENST00000367322:exon16:c.813-3C>A,ENST00000367318:exon17:c.822-3C>A,ENST00000360372:exon15:c.807-3C>A,ENST00000367315:exon15:c.813-3C>A,ENST00000367317:exon16:c.822-3C>A,ENST00000236918:exon16:c.837-3C>A,ENST00000421663:exon16:c.831-3C>A,ENST00000458432:exon17:c.849-3C>A,ENST00000367320:exon16:c.723-3C>A,ENST00000509001:exon17:c.822-3C>A,ENST00000438742:exon16:c.804-3C>A		L	0,00062
TNNT2	1_201328760_T_A	ENSG00000118194:ENST00000360372:exon13:c.A797T:p.N266I,ENSG00000118194:ENST00000367315:exon13:c.A803T:p.N268I,ENSG00000118194:ENST00000236918:exon14:c.A827T:p.N276I,ENSG00000118194:ENST00000367317:exon14:c.A812T:p.N271I,ENSG00000118194:ENST00000367320:exon14:c.A713T:p.N238I,ENSG00000118194:ENST00000367322:exon14:c.A803T:p.N268I,ENSG00000118194:ENST00000421663:exon14:c.A821T:p.N274I,ENSG00000118194:ENST00000438742:exon14:c.A794T:p.N265I,ENSG00000118194:ENST00000367318:exon15:c.A812T:p.N271I,ENSG00000118194:ENST00000458432:exon15:c.A839T:p.N280I,ENSG00000118194:ENST00000509001:exon15:c.A812T:p.N271I		P	0,00059
TNNT2	1_201328787_T_C	ENSG00000118194:ENST00000360372:exon13:c.A770G:p.N257S,ENSG00000118194:ENST00000367315:exon13:c.A776G:p.N259S,ENSG00000118194:ENST00000236918:exon14:c.A800G:p.N267S,ENSG00000118194:ENST00000367317:exon14:c.A785G:p.N262S,ENSG00000118194:ENST00000367320:exon14:c.A686G:p.N229S,ENSG00000118194:ENST00000367322:exon14:c.A776G:p.N259S,ENSG00000118194:ENST00000421663:exon14:c.A794G:p.N265S,ENSG00000118194:ENST00000438742:exon14:c.A767G:p.N256S,ENSG00000118194:ENST00000367318:exon15:c.A785G:p.N262S,ENSG00000118194:ENST00000458432:exon15:c.A812G:p.N271S,ENSG00000118194:ENST00000509001:exon15:c.A785G:p.N262S		P	0,00117
TNNT2	1_201331150_T_C	ENSG00000118194:ENST00000360372:exon11:c.A565G:p.T189A,ENSG00000118194:ENST00000367315:exon11:c.A571G:p.T191A,ENSG00000118194:ENST00000236918:exon12:c.A595G:p.T199A,ENSG00000118194:ENST00000367317:exon12:c.A580G:p.T194A,ENSG00000118194:ENST00000367320:exon12:c.A481G:p.T161A,ENSG00000118194:ENST00000367322:exon12:c.A571G:p.T191A,ENSG00000118194:ENST00000421663:exon12:c.A589G:p.T197A,ENSG00000118194:ENST00000438742:exon12:c.A562G:p.T188A,ENSG00000118194:ENST00000367318:exon13:c.A580G:p.T194A,ENSG00000118194:ENST00000458432:exon13:c.A607G:p.T203A,ENSG00000118194:ENST00000509001:exon13:c.A580G:p.T194A		N	0,00058

TNNT2	1_201332459_A_C	ENSG00000118194:ENST00000360372:exon9:c.T520G:p.S174A,ENSG00000118194:ENST00000236918:exon10:c.T550G:p.S184A,ENSG00000118194:ENST00000367315:exon10:c.T535G:p.S179A,ENSG00000118194:ENST00000367317:exon10:c.T535G:p.S179A,ENSG00000118194:ENST00000438742:exon10:c.T520G:p.S174A,ENSG00000118194:ENST00000367318:exon11:c.T535G:p.S179A,ENSG00000118194:ENST00000367320:exon11:c.T445G:p.S149A,ENSG00000118194:ENST00000367322:exon11:c.T535G:p.S179A,ENSG00000118194:ENST00000421663:exon11:c.T541G:p.S181A,ENSG00000118194:ENST00000509001:exon11:c.T535G:p.S179A,ENSG00000118194:ENST00000458432:exon12:c.T571G:p.S191A		N	0,00057
TNNT2	1_201332505_CTC_-	ENSG00000118194:ENST00000360372:exon9:c.472_474del:p.158_158del,ENSG00000118194:ENST00000236918:exon10:c.502_504del:p.168_168del,ENSG00000118194:ENST00000367315:exon10:c.487_489del:p.163_163del,ENSG00000118194:ENST00000367317:exon10:c.487_489del:p.163_163del,ENSG00000118194:ENST00000438742:exon10:c.472_474del:p.158_158del,ENSG00000118194:ENST00000367318:exon11:c.487_489del:p.163_163del,ENSG00000118194:ENST00000367320:exon11:c.397_399del:p.133_133del,ENSG00000118194:ENST00000367322:exon11:c.487_489del:p.163_163del,ENSG00000118194:ENST00000421663:exon11:c.493_495del:p.165_165del,ENSG00000118194:ENST00000509001:exon11:c.487_489del:p.163_163del,ENSG00000118194:ENST00000455702:exon12:c.517_519del:p.173_173del,ENSG00000118194:ENST00000458432:exon12:c.523_525del:p.175_175del		P	0,00172
TNNT2	1_201332514_CTC_-	ENSG00000118194:ENST00000360372:exon9:c.463_465del:p.155_155del,ENSG00000118194:ENST00000236918:exon10:c.493_495del:p.165_165del,ENSG00000118194:ENST00000367315:exon10:c.478_480del:p.160_160del,ENSG00000118194:ENST00000367317:exon10:c.478_480del:p.160_160del,ENSG00000118194:ENST00000438742:exon10:c.463_465del:p.155_155del,ENSG00000118194:ENST00000367318:exon11:c.478_480del:p.160_160del,ENSG00000118194:ENST00000367320:exon11:c.388_390del:p.130_130del,ENSG00000118194:ENST00000367322:exon11:c.478_480del:p.160_160del,ENSG00000118194:ENST00000421663:exon11:c.484_486del:p.162_162del,ENSG00000118194:ENST00000509001:exon11:c.478_480del:p.160_160del,ENSG00000118194:ENST00000455702:exon12:c.508_510del:p.170_170del,ENSG00000118194:ENST00000458432:exon12:c.514_516del:p.172_172del		P	0,00172
TNNT2	1_201334389_G_A	ENSG00000118194:ENST00000360372:exon7:c.C296T:p.A99V,ENSG00000118194:ENST00000236918:exon8:c.C326T:p.A109V,ENSG00000118194:ENST00000367315:exon8:c.C311T:p.A104V,ENSG00000118194:ENST00000367317:exon8:c.C311T:p.A104V,ENSG00000118194:ENST00000438742:exon8:c.C296T:p.A99V,ENSG00000118194:ENST00000367318:exon9:c.C311T:p.A104V,ENSG00000118194:ENST00000367322:exon9:c.C311T:p.A104V,ENSG00000118194:ENST00000421663:exon9:c.C317T:p.A106V,ENSG00000118194:ENST00000422165:exon9:c.C326T:p.A109V,ENSG00000118194:ENST00000509001:exon9:c.C311T:p.A104V,ENSG00000118194:ENST00000455702:exon10:c.C341T:p.A114V,ENSG00000118194:ENST00000458432:exon10:c.C347T:p.A116V		P	0,00174

TNNT2	1_201334419_C_T	ENSG00000118194:ENST00000360372:exon7:c.G266A:p.R89H,ENSG00000118194:ENST00000236918:exon8:c.G296A:p.R99H,ENSG00000118194:ENST00000367315:exon8:c.G281A:p.R94H,ENSG00000118194:ENST00000367317:exon8:c.G281A:p.R94H,ENSG00000118194:ENST00000438742:exon8:c.G266A:p.R89H,ENSG00000118194:ENST00000367318:exon9:c.G281A:p.R94H,ENSG00000118194:ENST00000367322:exon9:c.G281A:p.R94H,ENSG00000118194:ENST00000412633:exon9:c.G278A:p.R93H,ENSG00000118194:ENST00000421663:exon9:c.G287A:p.R96H,ENSG00000118194:ENST00000422165:exon9:c.G296A:p.R99H,ENSG00000118194:ENST00000509001:exon9:c.G281A:p.R94H,ENSG00000118194:ENST00000455702:exon10:c.G311A:p.R104H,ENSG00000118194:ENST00000458432:exon10:c.G317A:p.R106H		P	0,00059
TNNT2	1_201334425_C_T	ENSG00000118194:ENST00000360372:exon7:c.G260A:p.R87Q,ENSG00000118194:ENST00000236918:exon8:c.G290A:p.R97Q,ENSG00000118194:ENST00000367315:exon8:c.G275A:p.R92Q,ENSG00000118194:ENST00000367317:exon8:c.G275A:p.R92Q,ENSG00000118194:ENST00000438742:exon8:c.G260A:p.R87Q,ENSG00000118194:ENST00000367318:exon9:c.G275A:p.R92Q,ENSG00000118194:ENST00000367322:exon9:c.G275A:p.R92Q,ENSG00000118194:ENST00000412633:exon9:c.G272A:p.R91Q,ENSG00000118194:ENST00000421663:exon9:c.G281A:p.R94Q,ENSG00000118194:ENST00000422165:exon9:c.G290A:p.R97Q,ENSG00000118194:ENST00000509001:exon9:c.G275A:p.R92Q,ENSG00000118194:ENST00000455702:exon10:c.G305A:p.R102Q,ENSG00000118194:ENST00000458432:exon10:c.G311A:p.R104Q	rs121964856	D;P	0,00059
TNNT2	1_201334426_G_A	ENSG00000118194:ENST00000360372:exon7:c.C259T:p.R87W,ENSG00000118194:ENST00000236918:exon8:c.C289T:p.R97W,ENSG00000118194:ENST00000367315:exon8:c.C274T:p.R92W,ENSG00000118194:ENST00000367317:exon8:c.C274T:p.R92W,ENSG00000118194:ENST00000438742:exon8:c.C259T:p.R87W,ENSG00000118194:ENST00000367318:exon9:c.C274T:p.R92W,ENSG00000118194:ENST00000367322:exon9:c.C274T:p.R92W,ENSG00000118194:ENST00000412633:exon9:c.C271T:p.R91W,ENSG00000118194:ENST00000421663:exon9:c.C280T:p.R94W,ENSG00000118194:ENST00000422165:exon9:c.C289T:p.R97W,ENSG00000118194:ENST00000509001:exon9:c.C274T:p.R92W,ENSG00000118194:ENST00000455702:exon10:c.C304T:p.R102W,ENSG00000118194:ENST00000458432:exon10:c.C310T:p.R104W		P	0,00118
TNNT2	1_201334766_A_T	ENSG00000118194:ENST00000360372:exon6:c.T221A:p.I74N,ENSG00000118194:ENST00000236918:exon7:c.T251A:p.I84N,ENSG00000118194:ENST00000367315:exon7:c.T236A:p.I79N,ENSG00000118194:ENST00000367317:exon7:c.T236A:p.I79N,ENSG00000118194:ENST00000438742:exon7:c.T221A:p.I74N,ENSG00000118194:ENST00000367318:exon8:c.T236A:p.I79N,ENSG00000118194:ENST00000367322:exon8:c.T236A:p.I79N,ENSG00000118194:ENST00000412633:exon8:c.T233A:p.I78N,ENSG00000118194:ENST00000421663:exon8:c.T242A:p.I81N,ENSG00000118194:ENST00000422165:exon8:c.T251A:p.I84N,ENSG00000118194:ENST00000509001:exon8:c.T236A:p.I79N,ENSG00000118194:ENST00000367320:exon9:c.T263A:p.I88N,ENSG00000118194:ENST00000455702:exon9:c.T266A:p.I89N,ENSG00000118194:ENST00000458432:exon9:c.T272A:p.I91N	rs121964855	D;P	0,00058

TNNT2	1_201337340_G_A	ENSG00000118194:ENST00000360372:exon3:c.C68T:p.A23V,ENSG00000118194:ENST00000236918:exon4:c.C98T:p.A33V,ENSG00000118194:ENST00000367315:exon4:c.C83T:p.A28V,ENSG00000118194:ENST00000367317:exon4:c.C83T:p.A28V,ENSG00000118194:ENST00000438742:exon4:c.C68T:p.A23V,ENSG00000118194:ENST00000367318:exon5:c.C83T:p.A28V,ENSG00000118194:ENST00000367322:exon5:c.C83T:p.A28V,ENSG00000118194:ENST00000412633:exon5:c.C83T:p.A28V,ENSG00000118194:ENST00000421663:exon5:c.C89T:p.A30V,ENSG00000118194:ENST00000422165:exon5:c.C98T:p.A33V,ENSG00000118194:ENST00000509001:exon5:c.C83T:p.A28V,ENSG00000118194:ENST00000367320:exon6:c.C113T:p.A38V,ENSG00000118194:ENST00000455702:exon6:c.C113T:p.A38V,ENSG00000118194:ENST00000458432:exon6:c.C119T:p.A40V	rs200754249	D;P	0,00058
TNNT2	1_201337541_G_A	ENST00000360372:exon3:c.52+5C>T		L	0,00058
TNNT2	1_201338976_G_-	ENST00000236918:exon4:c.52-2C>-,ENST00000458432:exon6:c.74-3C>-,ENST00000367320:exon6:c.68-3C>-,ENST00000455702:exon6:c.68-3C>-,ENST00000422165:exon5:c.53-3C>-	rs200153031	D	0,00184
TNNT2	1_201338976_G_A	ENST00000236918:exon4:c.52-2C>T,ENST00000458432:exon6:c.74-3C>T,ENST00000367320:exon6:c.68-3C>T,ENST00000455702:exon6:c.68-3C>T,ENST00000422165:exon5:c.53-3C>T		N	0,00122
TNNT2	1_201338978_G_A	ENST00000236918:exon4:c.52-4C>T,ENST00000458432:exon6:c.74-5C>T,ENST00000367320:exon6:c.68-5C>T,ENST00000455702:exon6:c.68-5C>T,ENST00000422165:exon5:c.53-5C>T		L	0,00061
TNNT2	1_201338979_G_-	ENST00000236918:exon4:c.52-5C>-		L	0,00183
TPM1	15_63335110_G_A	ENSG00000140416:ENST00000267996:exon1:c.G82A:p.D28N,ENSG00000140416:ENST00000288398:exon1:c.G82A:p.D28N,ENSG00000140416:ENST00000357980:exon1:c.G82A:p.D28N,ENSG00000140416:ENST00000358278:exon1:c.G82A:p.D28N,ENSG00000140416:ENST00000403994:exon1:c.G82A:p.D28N,ENSG00000140416:ENST00000558347:exon1:c.G82A:p.D28N,ENSG00000140416:ENST00000559397:exon1:c.G82A:p.D28N,ENSG00000140416:ENST00000559556:exon1:c.G82A:p.D28N,ENSG00000140416:ENST00000560445:exon1:c.G82A:p.D28N,ENSG00000140416:ENST00000561425:exon1:c.G82A:p.D28N		N	0,00187
TPM1	15_63335138_A_C	ENSG00000140416:ENST00000267996:exon1:c.A110C:p.K37T,ENSG00000140416:ENST00000288398:exon1:c.A110C:p.K37T,ENSG00000140416:ENST00000357980:exon1:c.A110C:p.K37T,ENSG00000140416:ENST00000358278:exon1:c.A110C:p.K37T,ENSG00000140416:ENST00000403994:exon1:c.A110C:p.K37T,ENSG00000140416:ENST00000558347:exon1:c.A110C:p.K37T,ENSG00000140416:ENST00000559397:exon1:c.A110C:p.K37T,ENSG00000140416:ENST00000559556:exon1:c.A110C:p.K37T,ENSG00000140416:ENST00000560445:exon1:c.A110C:p.K37T,ENSG00000140416:ENST00000561425:exon1:c.A110C:p.K37T		N	0,00094

TPM1	15_63336299_C_T	ENSG00000140416:ENST00000288398:exon2:c.C188T:p.A63V,ENSG00000140416:ENST00000358278:exon2:c.C188T:p.A63V,ENSG00000140416:ENST00000403994:exon2:c.C188T:p.A63V,ENSG00000140416:ENST00000558347:exon2:c.C188T:p.A63V,ENSG00000140416:ENST00000559556:exon2:c.C188T:p.A63V,ENSG00000140416:ENST00000357980:exon3:c.C314T:p.A105V	rs199476306	D;P	0,00057
TPM1	15_63351757_T_C	ENST00000288398:exon4:c.375-5T>C,ENST00000358278:exon4:c.375-5T>C,ENST00000403994:exon4:c.375-5T>C,ENST00000357980:exon5:c.501-5T>C,ENST00000267996:exon4:c.375-5T>C,ENST00000559397:exon4:c.375-5T>C,ENST00000559556:exon4:c.375-5T>C,ENST00000560970:exon4:c.317-5T>C,ENST00000561266:exon3:c.191-5T>C,ENST00000559831:exon3:c.147-5T>C,ENST00000334895:exon3:c.267-5T>C,ENST00000404484:exon3:c.267-5T>C,ENST00000558544:exon2:c.133-5T>C,ENST00000560959:exon3:c.267-5T>C,ENST00000317516:exon3:c.267-5T>C,ENST00000559281:exon3:c.267-5T>C,ENST00000561395:exon2:c.114-5T>C		P	0,00057
TPM1	15_63353123_C_T	ENSG00000140416:ENST00000558264:exon3:c.C170T:p.A57V,ENSG00000140416:ENST00000317516:exon4:c.C440T:p.A147V,ENSG00000140416:ENST00000334895:exon4:c.C440T:p.A147V,ENSG00000140416:ENST00000404484:exon4:c.C440T:p.A147V,ENSG00000140416:ENST00000559281:exon4:c.C440T:p.A147V,ENSG00000140416:ENST00000559831:exon4:c.C320T:p.A107V,ENSG00000140416:ENST00000560615:exon4:c.C170T:p.A57V,ENSG00000140416:ENST00000560959:exon4:c.C440T:p.A147V,ENSG00000140416:ENST00000267996:exon5:c.C548T:p.A183V,ENSG00000140416:ENST00000288398:exon5:c.C548T:p.A183V,ENSG00000140416:ENST00000358278:exon5:c.C548T:p.A183V,ENSG00000140416:ENST00000403994:exon5:c.C548T:p.A183V,ENSG00000140416:ENST00000559397:exon5:c.C548T:p.A183V,ENSG00000140416:ENST00000559556:exon5:c.C548T:p.A183V,ENSG00000140416:ENST00000357980:exon6:c.C674T:p.A225V		S	0,00058
TPM1	15_63353922_G_A	ENSG00000140416:ENST00000561395:exon3:c.G200A:p.R67Q,ENSG00000140416:ENST00000334895:exon5:c.G466A:p.E156K,ENSG00000140416:ENST00000558264:exon5:c.G272A:p.R91Q,ENSG00000140416:ENST00000559281:exon5:c.G466A:p.E156K,ENSG00000140416:ENST00000267996:exon6:c.G574A:p.E192K,ENSG00000140416:ENST00000403994:exon6:c.G574A:p.E192K,ENSG00000140416:ENST00000559556:exon6:c.G574A:p.E192K,ENSG00000140416:ENST00000559831:exon6:c.G422A:p.R141Q,ENSG00000140416:ENST00000357980:exon7:c.G700A:p.E234K	rs199476315	D;P	0,00115
TPM1	15_63354818_T_G	ENSG00000140416:ENST00000317516:exon7:c.T638G:p.L213W,ENSG00000140416:ENST00000334895:exon7:c.T638G:p.L213W,ENSG00000140416:ENST00000404484:exon7:c.T638G:p.L213W,ENSG00000140416:ENST00000559281:exon7:c.T638G:p.L213W,ENSG00000140416:ENST00000560959:exon7:c.T638G:p.L213W,ENSG00000140416:ENST00000267996:exon8:c.T746G:p.L249W,ENSG00000140416:ENST00000288398:exon8:c.T746G:p.L249W,ENSG00000140416:ENST00000358278:exon8:c.T746G:p.L249W,ENSG00000140416:ENST00000403994:exon8:c.T746G:p.L249W,ENSG00000140416:ENST00000559397:exon8:c.T746G:p.L249W,ENSG00000140416:ENST00000559556:exon8:c.T746G:p.L249W,ENSG00000140416:ENST00000357980:exon9:c.T872G:p.L291W		S	0,00057

TPM1	15_63354833_A_G	ENSG00000140416:ENST00000317516:exon7:c.A653G:p.D218G,ENSG00000140416:ENST00000334895:exon7:c.A653G:p.D218G,ENSG00000140416:ENST00000404484:exon7:c.A653G:p.D218G,ENSG00000140416:ENST00000559281:exon7:c.A653G:p.D218G,ENSG00000140416:ENST00000560959:exon7:c.A653G:p.D218G,ENSG00000140416:ENST00000267996:exon8:c.A761G:p.D254G,ENSG00000140416:ENST00000288398:exon8:c.A761G:p.D254G,ENSG00000140416:ENST00000358278:exon8:c.A761G:p.D254G,ENSG00000140416:ENST00000403994:exon8:c.A761G:p.D254G,ENSG00000140416:ENST00000559397:exon8:c.A761G:p.D254G,ENSG00000140416:ENST00000559556:exon8:c.A761G:p.D254G,ENSG00000140416:ENST00000357980:exon9:c.A887G:p.D296G		S	0,00057
TPM1	15_63354907_A_G	ENSG00000140416:ENST00000560959:exon7:c.A727G:p.I243V		N	0,00057
TPM1	15_63354938_CGCT_-	ENSG00000140416:ENST00000560959:exon7:c.758_761del:p.253_254del		L	0,00114
TPM1	15_63354943_C_T	ENSG00000140416:ENST00000560959:exon7:c.C763T:p.L255F	rs56054026	D	0,00114
TPM1	15_63356281_A_G	ENSG00000140416:ENST00000559281:exon8:c.A683G:p.K228R,ENSG00000140416:ENST00000288398:exon9:c.A791G:p.K264R,ENSG00000140416:ENST00000403994:exon9:c.A791G:p.K264R		S	0,00057
TPM1	15_63358166_A_G	ENSG00000140416:ENST00000560445:exon3:c.A193G:p.I65V		N	0,00057
VCL	10_75832551_G_A	ENSG00000035403:ENST00000211998:exon5:c.G563A:p.R188Q,ENSG00000035403:ENST00000372755:exon5:c.G563A:p.R188Q		N	0,00057
VCL	10_75832578_C_T	ENSG00000035403:ENST00000211998:exon5:c.C590T:p.T197I,ENSG00000035403:ENST00000372755:exon5:c.C590T:p.T197I	rs189242810	D;S	0,00057
VCL	10_75834522_C_T	ENSG00000035403:ENST00000211998:exon6:c.C644T:p.T215I,ENSG00000035403:ENST00000372755:exon6:c.C644T:p.T215I		N	0,00057
VCL	10_75849088_A_G	ENSG00000035403:ENST00000436396:exon2:c.A173G:p.K58R,ENSG00000035403:ENST00000211998:exon9:c.A1157G:p.K386R,ENSG00000035403:ENST00000372755:exon9:c.A1157G:p.K386R	rs200342284	D	0,00115
VCL	10_75849097_A_G	ENSG00000035403:ENST00000436396:exon2:c.A182G:p.D61G,ENSG00000035403:ENST00000211998:exon9:c.A1166G:p.D389G,ENSG00000035403:ENST00000372755:exon9:c.A1166G:p.D389G		N	0,00115
VCL	10_75849776_T_C	ENST00000211998:exon10:c.1177-5T>C,ENST00000372755:exon10:c.1177-5T>C,ENST00000436396:exon3:c.193-5T>C		L	0,00057
VCL	10_75849841_G_A	ENSG00000035403:ENST00000436396:exon3:c.G253A:p.A85T,ENSG00000035403:ENST00000211998:exon10:c.G1237A:p.A413T,ENSG00000035403:ENST00000372755:exon10:c.G1237A:p.A413T	rs146278697	D	0,00057
VCL	10_75855425_A_C	ENSG00000035403:ENST00000436396:exon5:c.A571C:p.I191L,ENSG00000035403:ENST00000211998:exon12:c.A1555C:p.I519L,ENSG00000035403:ENST00000372755:exon12:c.A1555C:p.I519L	rs141033098	D;S	0,00173
VCL	10_75855491_C_G	ENSG00000035403:ENST00000436396:exon5:c.C637G:p.L213V,ENSG00000035403:ENST00000211998:exon12:c.C1621G:p.L541V,ENSG00000035403:ENST00000372755:exon12:c.C1621G:p.L541V		N	0,00058
VCL	10_75857050_T_C	ENSG00000035403:ENST00000436396:exon6:c.T848C:p.V283A,ENSG00000035403:ENST00000211998:exon13:c.T1832C:p.V611A,ENSG00000035403:ENST00000372755:exon13:c.T1832C:p.V611A		N	0,00057

VCL	10_75857059_C_T	ENSG00000035403:ENST00000436396:exon6:c.C857T:p.T286M,ENSG00000035403:ENST00000211998:exon13:c.C1841T:p.T614M,ENSG00000035403:ENST00000372755:exon13:c.C1841T:p.T614M		S	0,00057
VCL	10_75860740_A_G	ENSG00000035403:ENST00000436396:exon7:c.A923G:p.H308R,ENSG00000035403:ENST00000211998:exon14:c.A1907G:p.H636R,ENSG00000035403:ENST00000372755:exon14:c.A1907G:p.H636R	rs71579374	D	0,00057
VCL	10_75860750_G_T	ENSG00000035403:ENST00000436396:exon7:c.G933T:p.K311N,ENSG00000035403:ENST00000211998:exon14:c.G1917T:p.K639N,ENSG00000035403:ENST00000372755:exon14:c.G1917T:p.K639N		S	0,00057
VCL	10_75860773_C_T	ENSG00000035403:ENST00000436396:exon7:c.C956T:p.A319V,ENSG00000035403:ENST00000211998:exon14:c.C1940T:p.A647V,ENSG00000035403:ENST00000372755:exon14:c.C1940T:p.A647V		N	0,00057
VCL	10_75860806_T_C	ENSG00000035403:ENST00000436396:exon7:c.T989C:p.V330A,ENSG00000035403:ENST00000211998:exon14:c.T1973C:p.V658A,ENSG00000035403:ENST00000372755:exon14:c.T1973C:p.V658A		N	0,00172
VCL	10_75871695_TAGGTG_-	ENSG00000035403:ENST00000436396:exon12:c.1790_1795del:p.597_599del,ENSG00000035403:ENST00000211998:exon19:c.2774_2779del:p.925_927del		N	0,00057
VCL	10_75871722_C_T	ENSG00000035403:ENST00000436396:exon12:c.C1817T:p.A606V,ENSG00000035403:ENST00000211998:exon19:c.C2801T:p.A934V	rs16931179	D;P	0,00057
VCL	10_75871748_C_G	ENSG00000035403:ENST00000436396:exon12:c.C1843G:p.P615A,ENSG00000035403:ENST00000211998:exon19:c.C2827G:p.P943A	rs71579375	D;P	0,00057
VCL	10_75871782_TGT_-	ENSG00000035403:ENST00000436396:exon12:c.1877_1879del:p.626_627del,ENSG00000035403:ENST00000211998:exon19:c.2861_2863del:p.954_955del		P	0,00057
VCL	10_75873951_A_G	ENSG00000035403:ENST00000417648:exon5:c.A538G:p.I180V,ENSG00000035403:ENST00000436396:exon13:c.A1975G:p.I659V,ENSG00000035403:ENST00000372755:exon19:c.A2755G:p.I919V,ENSG00000035403:ENST00000211998:exon20:c.A2959G:p.I987V		N	0,00057

ACTC1: actin, alpha, cardiac muscle 1; *ANK2*: ankyrin 2, neuronal; *CASQ2*: calsequestrin 2 cardiac muscle; *CAV3*: caveolin 3; *CSRP3*: cysteine and glycine-rich protein 3 cardiac LIM protein; *DES*: desmin; *DSC2*: desmocollin 2; *DSG2*: desmoglein 2; *DSP*: desmoplakin; *JUP*: junction plakoglobin; *KCNE1*: potassium voltage-gated channel, Isk-related family, member 1; *KCNE2*: potassium voltage-gated channel, Isk-related family, member 2; *KCNH2*: potassium voltage-gated channel, subfamily H eag-related, member 2; *KCNJ2*: potassium inwardly-rectifying channel, subfamily J, member 2; *KCNQ1*: potassium voltage-gated channel, KQT-like subfamily, member 1; *LDB3*: LIM domain binding 3; *LMNA*: lamin A/C; *MYBPC3*: myosin binding protein C, cardiac; *MYH6*: myosin, heavy chain 6, cardiac muscle, alpha; *MYH7*: myosin, heavy chain 7, cardiac muscle, beta; *MYL2*: myosin, light chain 2, regulatory, cardiac, slow; *MYL3*: myosin, light chain 3, alkali; *PDLIM3*: PDZ and LIM domain 3; *PKP2*: plakophilin 2; *PLN*, phospholamban; *RBM20*: RNA binding motif protein 20; *RYR2*: ryanodine receptor 2 cardiac; *SCN5A*: sodium channel, voltage-gated, type V, alpha subunit; *TCAP*: titin-cap; *TGFbeta3*: transforming growth factor, beta 3; *TMEM43*: transmembrane protein 43; *TNNC1*: troponin C type 1 slow; *TNNI3*: troponin I type 3 cardiac; *TNNT2*: troponin T type 2 cardiac; *TPM1*: tropomyosin 1 alpha; VCL: vinculin.

Supplementary table 2. Genotype-phenotype associations for individual sarcomere protein and related genes, with P-values <0.05. P-values reflect the comparison for proportions or means, between patients with and without a rare variant in a given gene.

Phenotype	Gene	Frequency or mean±standard deviation - Rare variant present	Frequency or mean±standard deviation – Rare variant absent	P-value
Age at initial evaluation (years)	*MYBPC3*	45.5±14.4	51.0±15.5	$8.97x10^{-6}$
	MYH7	43.9±15.4	50.5±15.2	$1.91x10^{-5}$
	TNNT2	43.5±14.7	49.8±15.4	0.031
	CSRP3	56.7±8.1	49.5±15.5	0.006
	VCL	43.2±17.6	49.8±15.3	0.0361
Family history of HCM	*MYBPC3*	40.4% (86/213)	21.9% (140/640)	$2.7x10^{-7}$
	MYH7	47.4% (54/114)	23.4% (173/740)	$3.06x10^{-7}$
LA diameter (mm)	*RBM20*	40.9±6.8	44.2±7.5	0.00239
MLVWT (mm)	*MYBPC3*	19.4±4.7	18.2±4.2	0.0005
LV end-diastolic diameter (mm)	*MYBPC3*	44.8±5.5	46.3±6.0	0.00089
	RBM20	43.6±5.2	46.1±6.0	0.00431
LV end-systolic diameter (mm)	*MYBPC3*	27.4±6.0	28.8±5.4	0.00512
Fractional shortening (%)	*MYBPC3*	39.7±8.9	38.2±8.8	0.026
LV systolic dysfunction	*TNNI3*	15.8% (3/19)	3.2% (26/810)	0.020
ASH pattern	*MYBPC3*	88.0% (184/209)	71.6% (459/641)	$3.75x10^{-6}$
	MYH7	89.2% (99/111)	73.6% (544/739)	0.00104
	MYH6	81.2% (56/69)	75.2% (587/781)	0.025
Moderate-severe mitral regurgitation	*MYL2*	55.6% (5/9)	19.4% (158/814)	0.041
Right ventricular hypertrophy	*TNNI3*	50% (10/20)	21.6% (174/806)	0.004
LVOTO (>30 mmHg)	*TNNI3*	10% (2/20)	41.2% (326/792)	0.00465
LVOT gradient (mmHg)	*TNNI3*	13.1±26.6	34.8±41.3	0.002
Family history of SCD	*MYBPC3*	28.7% (62/216)	18.3% (120/656)	0.001
	MYH7	31.6% (37/117)	19.2% (145/775)	0.003
	TPM1	62.5% (5/8)	20.5% (177/864)	0.012
BP response to exercise (mmHg)	MYH7	36.6±19.9	50.2±24.4	5.49×10^{-6}
BP response to exercise ≤ 20 mmHg	*MYH7*	22.5% (20/89)	12.6% (72/573)	0.020
	TNNT2	40.9% (9/22)	13.0% (83/640)	0.002
Syncope	*MYH7*	22.8% (26/114)	15.4% (114/742)	0.045

Implanted cardioverter-defibrillator	*MYH7*	28.2% (33/117)	19.2% (145/756)	0.027
Myectomy and/or alcohol septal ablation and/or pacemaker implantation for gradient reduction	*MYBPC3* (splicing variants)	43.1% (22/51)	20.0% (160/799)	3.0×10^{-4}

HCM: hypertrophic cardiomyopathy; NYHA: New York Heart Association; LA: left atria; MLVWT: maximum left ventricular wall thickness; LV: left ventricular; ASH: asymmetric septal hypertrophy; LVOT: left ventricular outflow tract; LVOTO: left ventricular outflow tract obstruction; SCD: sudden cardiac death; BP: blood pressure;

Supplementary table 3. Genotype-phenotype associations for individual non-sarcomere protein genes, when only analysing the sub-cohort of sarcomere-positive individuals. P-values reflect the comparison for proportions or means, between the group of patients with versus the group of patients without a rare variant in a given non-sarcomere protein gene.

Phenotype	**Gene**	**Frequency or mean±standard deviation - Variant present**	**Frequency or mean±standard deviation - Variant absent**	**P-value**
LA diameter at last follow-up (mm)	*SCN5A*	47.4±4.8	44.1±7.8	0.109
LVOTO (>30 mmHg)	*SCN5A*	50.0% (9/18)	36.0% (122/339)	0.315
	Ion-channel	43.6% (34/78)	34.8% (97/279)	0.184
MLVWT (mm)	*ANK2*	19.9±6.2	18.9±4.5	0.377
MLVWT ≥30mm	*ANK2*	16% (4/25)	2.8% (7/251)	0.007
E/e' ratio	*CASQ2*	17.5±7.7	10.6±5.2	0.004
NSVT	*PLN*	100% (1/1)	24.7% (61/247)	0.083

LA: left atria; LVOTO: left ventricular outflow tract obstruction; MLVWT: maximal left ventricular wall thickness; E/e´ratio: ratio between the maximal velocity of the E wave from the pulsed wave Doppler of the transmitral flow and the maximal velocity of the e' wave of tissue Doppler at the mitral annulus; NSVT: non-sustained ventricular tachycardia.