

BASIC SCIENCE FOR CLINICIANS

# Genetics of BAG3: A Paradigm for Developing Precision Therapies for Dilated Cardiomyopathies

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**ABSTRACT:** Nonischemic dilated cardiomyopathy is a common form of heart muscle disease in which genetic factors play a critical etiological role. In this regard, both rare disease-causing mutations and common disease-susceptible variants, in the Bcl-2-associated athanogene 3 (*BAG3*) gene have been reported, highlighting the critical role of *BAG3* in cardiomyocytes and in the development of dilated cardiomyopathy. The phenotypic effects of the *BAG3* mutations help investigators understand the structure and function of the *BAG3* gene. Indeed, we report herein that all of the known pathogenic/likely pathogenic variants affect at least 1 of 3 protein functional domains, ie, the WW domain, the second IPV (Ile-Pro-Val) domain, or the BAG domain, whereas none of the missense nontruncating pathogenic/likely pathogenic variants affect the proline-rich repeat (PXXP) domain. A common variant, p.Cys151Arg, associated with reduced susceptibility to dilated cardiomyopathy demonstrated a significant difference in allele frequencies among diverse human populations, suggesting evolutionary selective pressure. As *BAG3*-related therapies for heart failure move from the laboratory to the clinic, the ability to provide precision medicine will depend in large part on having a thorough understanding of the potential effects of both common and uncommon genetic variants on these target proteins. The current review article provides a roadmap that investigators can utilize to determine the potential interactions between a patient's genotype, their phenotype, and their response to therapeutic interventions with both gene delivery and small molecules.

**Key Words:** *BAG3* ■ dilated cardiomyopathy ■ genetics ■ heart failure ■ *INPP5F* ■ precision medicine

Cardiomyopathies are a heterogeneous group of diseases of cardiac muscle<sup>1</sup> that include dilated, hypertrophic, restrictive, constrictive and arrhythmogenic right ventricular cardiomyopathies.<sup>2</sup> Dilated cardiomyopathies (DCMs) have been historically separated into those that are secondary to ischemic heart disease (ischemic DCM) and those that are nonischemic, with ischemic DCM being somewhat more common. Genetic variants were first shown to be etiologic in the development of hypertrophic cardiomyopathy<sup>3,4</sup> and novel therapies have been developed, using gene therapy<sup>5</sup> or small molecules.<sup>6</sup> By contrast, the cause of DCM has remained more obscure. Recent studies have shown convincingly that both heritable and sporadic

genetic variants play a pivotal role in the development of many forms of DCM, the most common being loss of function variants in *Titin*, the largest protein found in humans. The *Titin* myofilament is one of the 3 major myofilaments found in the cardiac sarcomere (in addition to actin and myosin) and is critical for restoring the ability of the cardiac sarcomere to maintain optimal function of the left ventricle during diastole.<sup>7</sup> Truncating variants of *Titin* are the most common genetic variants, found in as many as 25% of patients with both heritable and sporadic forms of DCM, as well as in a significant number of patients with peripartum cardiomyopathy.<sup>8,9</sup> The discovery of genetic mutations causing DCM has enhanced our knowledge regarding the pathobiology

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## Nonstandard Abbreviations and Acronyms

<b>ACMG</b>	American College of Medical Genetics and Genomics
<b>B/LB</b>	benign/likely benign
<b>BAG3</b>	Bcl-2-associated athanogene 3
<b>CAPZB</b>	capping actin protein of muscle Z-line subunit beta
<b>DCM</b>	dilated cardiomyopathy
<b>eQTL</b>	expression quantitative trait locus
<b>gnomAD</b>	Genome Aggregation Database
<b>GTE<sub>x</sub></b>	Genotype-Tissue Expression
<b>Hsc70</b>	heat shock cognate
<b>Hsp70</b>	70 kilo-dalton heat shock protein
<b>IPV</b>	Ile-Pro-Val
<b>LAMP-2</b>	lysosomal associated membrane protein 2
<b>LTCC</b>	L-type voltage-gated calcium channel Cav1.2
<b>LD</b>	linkage disequilibrium
<b>NF-<math>\kappa</math>B</b>	nuclear factor kappa B
<b>P/LP</b>	pathogenic/likely pathogenic
<b>PDZGEF2</b>	PDZ domain containing guanine nucleotide exchange factor 2
<b>PPCM</b>	peripartum cardiomyopathy
<b>PXXP</b>	proline-rich repeat
<b>SH3</b>	Src homology 3
<b>sHsps</b>	small heat shock proteins
<b>SIFT</b>	Sorting Intolerant From Tolerant
<b>VUS</b>	variant of uncertain significance

of the disease and has provided an opportunity for more precise diagnosis, better risk prediction, and potential for improved treatment outcomes using both gene therapy and small molecules.<sup>10</sup>

In addition to *Titin*, genetic variants in >30 genes have been reported to be associated with the development of DCMs. To date, genes related to DCM identified in patients or demonstrated in animal models have greatly expanded the biomolecular knowledge involving the pathogenesis of DCMs. They also highlight important therapeutic targets. Examples of pathogenic genetic variants have been found in the genes encoding  $\beta$ -adrenergic receptors ( $\beta$ 1<sup>11</sup> and  $\beta$ 2<sup>12</sup>), the calcium voltage-gated channel subunit alpha1 C gene encoding the L-type voltage-gated calcium channel Ca<sub>v</sub>1.2 (LTCC),<sup>13</sup> and the mitochondrial calcium uniporter gene.<sup>14</sup> One of the most interesting of this group of DCM-associated genes is Bcl-2-associated athanogene-3 (*BAG3*). *BAG3* encodes a multifunctional protein that regulates or facilitates numerous biological processes in the heart and other tissues

including stabilization of the sarcomere,<sup>15</sup> maintaining the integrity of the nuclear envelope,<sup>16</sup> coupling the  $\beta$ -adrenergic receptor and the L-type Ca<sup>2+</sup> channel, activating autophagy and inhibiting apoptosis,<sup>17</sup> and modulating the cardiac inflammasome.<sup>18</sup> However, we know far more about the biology of less common proteins than we do about BAG-3. The work detailed in this review was undertaken to bring order to the large number of reports detailing both informative and non-informative genetic variants in *BAG3*, so as to better understand how these variants alter the cardiomyopathy phenotype.

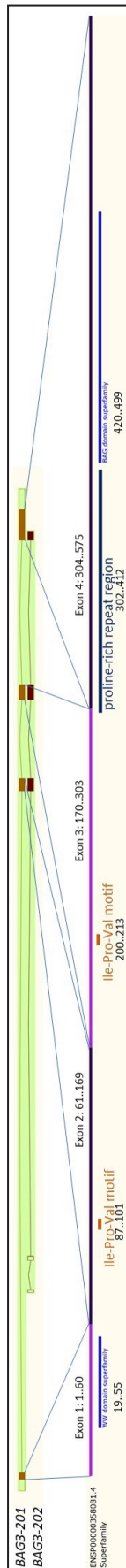
## BAG3 GENE

*BAG3* at Chr10q26.11 encodes a cochaperone, also known as Bcl-2-associated athanogene 3, which was first recognized for its ability to bind to Bcl-2 and effect an antiapoptotic function. The gene has ubiquitous expression in human tissues, with the highest level in heart, skeletal muscle, and the central nervous system (<https://www.ncbi.nlm.nih.gov/gene/9531#gene-expression>). High levels of *BAG3* have also been found in many cancers with perhaps the highest levels being found in pancreatic tumors.<sup>19</sup> The encoded protein, BAG3, comprises 575 aa and includes 4 known functional domains (National Center for Biotechnology Information reference NP\_004272.2, [Figure](#)).<sup>20,21</sup> Initially, BAG3 was found to serve as a cochaperone with the Hsp70 (70 kilo-dalton heat shock protein) at the Z-disc in striated muscle.<sup>22</sup> Molecular chaperones, eg, Hsps, are functionally related proteins assisting protein folding under both physiological and stressful conditions.<sup>23</sup> Hsp70s are the most ubiquitous members of the family of Hsps, and are found in all major subcellular compartments<sup>24</sup> where they are involved in a wide range of protein folding processes, including the folding of newly synthesized proteins and refolding of misfolded and aggregated proteins.<sup>25</sup>

## BAG3 Structure

The BAG3 protein comprises 4 types of protein functional domains ([Figure](#)), as delineated below:

1. The WW (Trp-Trp) domain seen in a number of unrelated proteins has known function in protein binding and signal transduction processes.<sup>26</sup> The WW domain binds with the PPDY motif of the PDZ domain containing guanine nucleotide exchange factor 2 (PDZGEF2), which induces activation of Small GTPase Rap1 and promotes integrin-mediated cell adhesion.<sup>27</sup> The role of BAG3 in cell adhesion mediated by the WW domain is critical for cell vitality and its antiapoptotic function.<sup>28</sup> One small nontruncating indel



**Figure. The Bcl-2-associated athanogene 3 (BAG3) gene model.**

The gene model was based on the annotation of the Ensemble project and the studies by Fuchs et al<sup>20</sup> and Doong et al<sup>34</sup> and the review by McCollum et al.<sup>21</sup> The encoded protein contains 4 types of protein domains, including 1 WW (Trp-Trp) domain (19–55 aa), 2 IPV (Ile-Pro-Val) motifs (87–101, 200–213 aa), 1 proline-rich repeat (PXXP) region (302–412 aa), and 1 BAG domain (420–499 aa).

2. Two IPV (Ile-Pro-Val) motifs bind with the molecular chaperones, sHsps (small heat shock proteins) HspB8/HspB6, and allow BAG3 to function as a scaffolding protein, linking Hsp70 to HspB8/HspB6 to form a ternary complex.<sup>30</sup> BAG3 facilitates the ability of sHsps to bind selectively with misfolded proteins and facilitates their refolding or degradation, the latter being mediated by Hsp70.<sup>31</sup> The importance of IPV in cardiomyopathies has been highlighted by disease-causing (classified as pathogenic/likely pathogenic [P/LP]) point mutations at the Pro209 position in the second IPV motif of BAG3.<sup>32</sup> The mutant protein of the toxic gain-of-function mutation p.Pro209Leu in BAG3 forms aggregates with the wild-type protein and causes BAG3 insufficiency.<sup>33</sup>
3. The proline-rich repeat (PXXP) region binds to SH3 (Src homology 3) motifs and mediates protein–protein interaction, eg, with phospholipase C-γ.<sup>34</sup> The activity of BAG3 is regulated by its phosphorylation status.<sup>35</sup> The binding of BAG3 with phospholipase C-γ enables the regulation of the activity of phosphorylated BAG3.<sup>34</sup> However, the role of PXXP in DCM is unclear. To date, 22 truncating mutations and 1 splicing mutation are reported in the PXXP region by the ClinVar database (<https://www.ncbi.nlm.nih.gov/clinvar/>, accessed by Aug 25, 2022). In contrast, there are no missense nontruncating P/LP variants related to DCM reported in the PXXP region. Moreover, two of the benign/likely benign (B/LB) variants (p.Pro407Leu and p.Pro380Ser) in the PXXP region recorded by the ClinVar database do not support the functional importance of this domain in DCM. Taken together, the pathogenicity of truncating/splicing variants are likely caused by lack of the BAG domain at the protein level.
4. The BAG domain mediates the binding of BAG3 with Hsp70. The BAG domain contains 3 antiparallel α-helices, while the second and third α-helices interact with the ATP-binding pocket of Hsp70 or the Hsc70 (heat shock cognate 70).<sup>36</sup> Hsp70 proteins have 3 major functional domains: an N-terminal ATPase domain, a substrate binding domain, and a C-terminal domain.<sup>37</sup> The interaction between BAG-3 and Hsp70 is mediated by the binding of the BAG domain of BAG3 and the N-terminal ATPase domain of Hsp70.<sup>36</sup> In addition, the BAG domain binds to the Bcl-2 Homology 4 domain of Bcl-2 to inhibit mitochondrial-dependent (intrinsic) apoptosis.<sup>38</sup> Bcl-2 promotes cell survival by inhibiting cleavage (activation) of the caspases, which are essential mediators of apoptosis, eg, apoptotic p.Ile30\_Asp31delinsAsnHis in the WW domain has been reported as likely pathogenic in DCM.<sup>29</sup>

protease activating factor 1.<sup>39</sup> Bcl-2 also inhibits the mitochondrial release of cytochrome c, which, when bound to apoptotic protease activating factor 1, activates the initiator executioner caspase-9 and the subsequent activation of the executioner caspases-3 and -7, which is the last step in the pathway leading to apoptosis.<sup>40</sup>

## Functions of BAG3 in Cardiomyocytes

BAG3 has been shown to play critical roles in cardiomyocytes, with effects mediated by Hsp70<sup>22</sup> and Bcl-2<sup>38</sup> actions. These critical functions include:

1. Maintaining sarcomere integrity: Under acute cellular stress, expression of HspB8/HspB6, Hsp70, and BAG3 are upregulated.<sup>35,41</sup> After the BAG3-sHsps (HspB8/HspB6) complex binds selectively with misfolded proteins, Hsp70 displaces sHsps in the complex.<sup>31</sup> The actin capping protein CapZ, a heterodimer with  $\alpha$  and  $\beta$  subunits,<sup>42</sup> binds the barbed ends of actin at sarcomeric Z-lines, and regulates thin filament assembly.<sup>42</sup> The  $\beta$ -subunit at Z-line is the  $\beta$ 1 isoform encoded by the *CAPZB* (capping actin protein of muscle Z-line subunit beta) gene.<sup>43</sup> At the Z-disc, BAG3 promotes the association between Hsc70 and CapZ $\beta$ 1 and stabilizes the structural integrity of the sarcomere during mechanical stress.<sup>44</sup> *Bag3* knockdown mice are shown to be susceptible to myofibrillar degeneration, and myofibrillar disarray has also been reported in humans with *BAG3* haploinsufficiency.<sup>44</sup>
2. Regulating macroautophagy: The Hsp70-BAG3 complex mediates the retrograde transport of misfolded proteins along microtubules to the microtubule-based inclusion bodies, known as aggresomes, at the microtubule organizing center.<sup>35,45</sup> Subsequently, BAG3 interacts directly with the macroautophagy receptor protein p62 (also known as sequestosome-1) and induces selective macroautophagy while lysosomes uptake and degrade protein aggregates.<sup>35,46</sup> BAG3-mediated selective macroautophagy plays critical roles in the cellular protein quality control system.<sup>35</sup>
3. Antiapoptosis: With BAG3 as an athanogene, the Hsp70-BAG3 complex protects antiapoptotic Bcl-2 proteins from proteasomal degradation and activates the antiapoptotic nuclear factor kappa B (NF- $\kappa$ B) pathway.<sup>47</sup> The activation of the NF- $\kappa$ B pathway consequently promotes the expression of Bcl-2.<sup>48</sup>
4.  $\beta$ -Adrenergic receptor/LTCC regulation: LTCC triggers intracellular  $Ca^{2+}$  release during excitation-contraction coupling.<sup>49</sup> The  $\beta$ -adrenergic/cAMP signaling pathway is the major mediator of LTCC

activation.<sup>50</sup> A previous study identified that BAG3 coimmunoprecipitated with  $\beta$ 1-adrenergic receptor and LTCC.<sup>51</sup> Haploinsufficiency of *BAG3* in mice results in  $\beta$ -adrenergic insensitivity of LTCC.<sup>52</sup>

5. Mitochondrial function: Mitochondrial quality control in cardiomyocytes plays a critical role in the progression of cardiovascular diseases.<sup>53</sup> Critical roles of *BAG3* in cardiomyocytes involve mitochondrial quality control by regulating both endogenous and exogenous expression of Parkin, a major regulator of mitophagy in mitochondrial homeostasis control.<sup>54</sup>

## PATHOGENESIS AND CLINICAL PRESENTATIONS OF GENETIC VARIANTS OF BAG3

### Dilated Cardiomyopathy

Genetic mutations of *BAG3* have been demonstrated to affect both the myocardium and the skeletal muscle leading to DCM and myofibrillar myopathy, respectively.<sup>55</sup> It has been observed that impaired function of *BAG3* by genetic mutation causes disrupted Z-disc and enhanced sensitivity to apoptosis in cardiomyocytes and skeletal muscles.<sup>22</sup> Disrupted interaction between *BAG3* and HSP70 leading to decreased levels of sHsps,<sup>56</sup> impaired *BAG3*-mediated sarcomeric protein turnover leading to reduced myofilament maximal force-generating capacity,<sup>15</sup> and cardiomyocyte accumulation of the preamyloid oligomers and activation of p38 signaling.<sup>57</sup> The phenotypic effects of *BAG3* mutations may be mediated by either haploinsufficiency<sup>57</sup> or a dominant negative mechanism.<sup>58</sup> Impaired selective macroautophagy has been highlighted as being critical in the pathogenesis in DCM and myofibrillar myopathy. In addition to the effects mediated by *BAG3*, the role of selective macroautophagy has also been emphasized by the previous finding that primary deficiency of the lysosomal structural protein LAMP-2 (lysosomal associated membrane protein 2) attributable to genetic variants causes X-linked vacuolar cardiomyopathy and myofibrillar myopathy (Danon disease).<sup>59</sup>

According to a large study of 2538 patients with DCM by Mazzarotto et al, truncating variants of *BAG3* were identified in 0.3% of DCM cases.<sup>60</sup> Clinically, *BAG3* mutations present with diverse cardiovascular phenotypes,<sup>61</sup> and with variable age of onset from 18 years<sup>61</sup> to 64 years.<sup>62</sup> According to the large sample study of 129 individuals with *BAG3* mutations (86% are truncating) by Domínguez et al, 68.4% patients had DCM, and the mean age of diagnosis was 36.9 years with an early age of onset in men ( $\approx$ 6 years in average).<sup>29</sup> ECG abnormalities were observed with the diagnosis and progression of DCM, including prolonged QRS duration, as well as

negative T waves in 34.0% patients.<sup>29</sup> Echocardiogram may disclose decreased left ventricular (LV) ejection fraction, increased LV end-diastolic diameter, and decreased score of tricuspid annular plane systolic excursion.<sup>29</sup> LV noncompaction has also been observed in patients with *BAG3* mutations.<sup>63</sup> Clinical end points, including cardiac death, heart transplant, LV assist device, aborted sudden cardiac death, and serious ventricular arrhythmia, were observed in 30.1% patients with the incidence of 5.1% per year.<sup>29</sup> It was also observed that acute-onset DCM may be triggered by infection in patients with *BAG3* mutations.<sup>64</sup> Patients with DCM who have *BAG3* mutations are less responsive to treatment than those with other genetic forms of DCM, while only 2.9% of patients with DCM at first evaluation have normalized LV ejection fraction during follow-up.<sup>29</sup> In addition to disease-causing mutations, some genetic variants in *BAG3* identified in individuals of African ancestry that were not causative of disease were associated with a negative outcome in patients with DCM.<sup>65</sup>

## Myocarditis

To date, a causal link between viral myocarditis and the development of DCM has also been recognized.<sup>66,67</sup> The critical roles of *BAG3* in cardiomyocytes imply its involvement in viral insults of cardiomyocytes, eg, in COVID-19 infection and influenza. *BAG3* mutations have been shown to result in dysregulation of mitochondrial dynamics and activation of p38 signaling in inflammatory response and apoptosis.<sup>57</sup> Correlation between *BAG3* mutations and myocarditis has been identified by recent studies.<sup>68,69</sup> These previous studies implied the possible correlation between *BAG3* mutation and myocarditis caused by COVID-19 infection, which warrants further investigation.

## Peripartum Cardiomyopathy

Peripartum cardiomyopathy (PPCM) is a rare life-threatening cardiomyopathy of unknown cause, which develops cardiac failure in the last month of pregnancy or within 5 months after delivery without history of a previous heart disease.<sup>70</sup> PPCM can be the initial manifestation of familial DCM.<sup>71</sup> Shared genetic predisposition in PPCM and DCM has been demonstrated in previous studies, including *BAG3* mutation.<sup>8,9</sup>

## Glycogen Storage Diseases

Autophagy mediates critical cytoprotective effects.<sup>72</sup> Defective autophagy causes the accumulation of aggregated/misfolded proteins, damaged organelles, and abnormal glycogen storage.<sup>73</sup> Myopathy may be caused by abnormal glycogen storage.<sup>74</sup> As discussed above, *BAG3* plays a critical role in regulating autophagy. Glycogen accumulations have been observed in a

**Table 1. Clinical Significance of Different Types of Variants**

Coding variant type	Clinical significance	Count (%)
Deletion	P/LP	7 (63.64)
	VUS	4 (36.36)
Nonsense (truncating)	P/LP	31 (93.94)
	VUS	1 (3.03)
	Conflicting interpretations of pathogenicity	1 (3.03)
Frame-shift (truncating)	P/LP	28 (84.85)
	VUS	5 (15.15)
Missense (nontruncating)	B/LB	9 (2.56)
	P/LP	6 (1.70)
	VUS	308 (87.50)
	Conflicting interpretations of pathogenicity	28 (7.95)

B/LB indicates benign/likely benign; P/LP, pathogenic/likely pathogenic; and VUS, variant of uncertain significance.

patient with myofibrillar myopathy and polyneuropathy caused by the *BAG3* mutation p.Pro209Gln.<sup>75</sup>

## Oncogenesis and Cancer Chemotherapy

In addition to the roles of *BAG3* in heart and DCM, *BAG3* has been demonstrated to play a role in tumorigenesis by mediating the effects of Hsp70 on Src signaling in cancer initiation and progression.<sup>76,77</sup> Bortezomib was the first proteasome inhibitor approved for anticancer therapy.<sup>78</sup> Bortezomib upregulates the level of *BAG3*, while *BAG3* gene silencing sensitizes leukemic cells to Bortezomib-induced apoptosis and enhances the therapeutic potency of Bortezomib.<sup>79</sup> *BAG3* may also downmodulate apoptotic response to tumor necrosis factor-related apoptosis-inducing ligand in cancer cells.<sup>80</sup> Targeting at *BAG3* with a small-molecule inhibitor YM-1 to block binding of Hsp70 to *BAG3* has been shown to suppress tumor growth in mice, highlighting *Bag3* as an appealing anticancer target.<sup>76</sup> Thus far, however, no genetic association has been identified between common variants of *BAG3* and cancers.

## RARE MUTATIONS AND PHENOTYPIC EFFECTS

To date, a number of disease-related variants in *BAG3* have been reported (Table S1).<sup>8,29,60,62,81,82</sup> Among these variants, 596 genetic variants in the *BAG3* gene related to cardiomyopathy or myofibrillar myopathy are archived in the ClinVar database (accessed April 6, 2022). Among these variants, 429 are annotated as coding variants, including 11 deletions, 33 nonsense (truncating) variants, 33 frame-shift (truncating) variants, and 351 missense (nontruncating) variants

(Table 1). Besides these coding variants, a splicing mutation (truncating) NM\_004281.4(BAG3):c.508-2A>G has been classified as pathogenic.<sup>29</sup> In addition to the above variants, 29 gross copy number variations encompassing *BAG3* have been classified as P/LP in ClinVar, with 23 (79.31%) P/LP gross copy number variations as copy number gain (Table S1). However, none of these copy number gain copy number variations are reported to be associated with DCM or myofibrillar myopathy. In contrast to the duplications or gain-of-function variants of *BAG3*, all P/LP variants related to DCM are loss-of-function variants.

## Deletions

Among the 11 deletions reported in ClinVar, 7 (63.64%) are predicted as P/LP and 4 (36.36%) as variant of uncertain significance (VUS). For the 7 P/LP deletions, 6 result in gross deletions of the coding region and 1 other deletion (21bp deletion, NM\_004281.4[BAG3]:c.508-14\_514del) causes damage of a splice acceptor site. For the 4 VUS deletions, one is an in-frame deletion (NC\_000010.11:g.[?\_119669831]\_[119670197\_?])del, missing exon 2 of *BAG3* with the first IPV motif), while the other 3 are single amino acid deletions. Two of the 3 single amino acid deletions involve the WW domain and the PXXP domain, respectively. Nonetheless, no deletion in the *BAG3* coding region was classified as B/LB.

In addition to the ClinVar records, other deletions related to DCM have been reported in the literature. Two gross deletions are predicted as pathogenic by the American College of Medical Genetics and Genomics (ACMG) guidelines, both resulting in loss of the critical BAG domain, ie, c.508\_1728del causing deletion of exons 3 and 4, as reported in DCM<sup>29</sup>; c.1070\_1660del causing deletion of aa.357–533 in exon 4.<sup>81</sup> In addition, a small nontruncating indel c.89\_91delTCGinsACC (p.Ile30\_Asp31delinsAsnHis), which locates at the WW domain, was reported in DCM and predicted as LP.<sup>29</sup> Importantly, all of the gross deletions involving the BAG domain identified to date are classified as pathogenic or LP. Potential importance of the WW domain in DCM is also supported by the reported small nontruncating LP deletion p.Ile30\_Asp31delinsAsnHis.<sup>29</sup>

## Nonsense Truncating Variants

Among the 33 nonsense truncating variants (producing a premature termination codon) registered in ClinVar, 31(93.94%) variants are classified as P/LP.<sup>9,29,62,81,83</sup> Only 1 (3.03%) nonsense variant p.Arg473Ter (with the BAG domain partially truncated, by missing 27 aa at the C-terminus of the BAG domain) is classified as having conflicting status of pathogenicity, and 1 (3.03%) nonsense variant p.Gln511Ter is classified as VUS without involving any of the protein functional domains.

Without exception, all of the P/LP nonsense variants are located upstream of the conflicting and VUS variants. The first P/LP nonsense truncating variant is p.Trp26Ter, which may fail to express any protein from the variant allele because of increased mRNA decay,<sup>84</sup> with resultant haploinsufficiency. The last P/LP nonsense truncating variant that has been identified is p.Tyr451Ter. Its placement at amino acid 451 justifies classifying any newly identified nonsense truncating variant at an earlier amino acid as P/LP.

## Frame-Shift Truncating Variants

Among the 33 frame-shift truncating variants recorded in ClinVar, 28 (84.85%) variants are classified as P/LP<sup>29,62,85</sup> and 5 (15.15%) are classified as VUS. The first frame-shift variant is p.Ser14fs and the last is p.Ala474fs. The distribution of the P/LP frame-shift variants are consistent with the distribution of nonsense variants as described above. In addition to the ClinVar recorded frame-shift variants, another frame-shift variant p.Glu57Lysfs\*154 Pathogenic has been reported to be associated with DCM and is predicted to be pathologic by ACMG criteria.<sup>29</sup> Without exception, all of the P/LP truncating variants reside upstream of the VUS variants in amino acid position. The last P/LP frame-shift variant is p.Ala474fs, which is consistent with the distribution of nonsense/nonframe-shift truncating variants. All VUS frame-shift variants occur after amino acid position 507, which is posterior to the BAG domain.

## Missense (Nontruncating) Variants

Compared with the deletions and truncating variants, missense variants causing a single amino acid substitution in *BAG3* are less deleterious. For the majority of the reported missense variants, functional classification has been VUS (308 [87.50%]) or with conflicting information regarding pathogenicity (28 [7.95%]) (Table S2). Among the 351 missense variants, only 6 (1.70%) are classified as P/LP. All 6 P/LP variants are located in a functional domain, ie, the amino acid 209 position at the second IPV domain, and amino acid positions at the BAG domain.<sup>29,32,86</sup> Interestingly, the missense variant p.Pro209Leu was initially identified as having an association with myofibrillar myopathy and restrictive cardiomyopathy or hypertrophic cardiomyopathy,<sup>86–88</sup> while later studies also reported the phenotype of DCM in relation with p.Pro209Leu.<sup>32,88</sup> In Zebrafish models, p.Pro209Leu was demonstrated as a toxic gain-of-function mutation.<sup>33</sup> Loss of *BAG3* may result in myofibrillar disintegration, while the mutant protein of p.Pro209Leu can rescue the myofibrillar disintegration phenotype.<sup>33</sup> However, the mutant protein may cause *BAG3* insufficiency as a result of forming aggregates with the wild-type protein as a dominant-negative effect.<sup>33</sup>

In contrast to the P/LP missense variants, two of the B/LB variants (p.Pro407Leu and p.Pro380Ser) are located in the PXXP region, while both have relatively common allele frequencies in the general population, ie, p.Pro407Leu MAF=0.126 and p.Pro380Ser MAF=0.003 according to the Genome Aggregation Database (gnomAD, version 2.1.1).<sup>89</sup> The amino acid substitutions caused by the 2 variants are significant with Grantham biochemical distances of 98 and 74, respectively.<sup>90</sup> These two B/LB variants thus undermine the functional importance of the PXXP region in DCM. Nonetheless, the co-occurrence of 2 missense variants—p.Pro63Ala (rs144041999) and p.Pro380Ser (rs144692954)—on the same chromosome was identified in individuals of African ancestry with cardiomyopathy, and shown to have dominant-negative effect in mouse model.<sup>65,91</sup> These 2 variants are most common in the African population with a frequency of 0.011 and in high linkage disequilibrium (LD) with  $r^2=1$  (<https://ldlink.nci.nih.gov/>). The p.Pro63Ala is near the WW domain and classified as benign by ClinVar. Further study on a larger sample of patients with cardiomyopathy and African ancestry may help to establish the causal relationship of the haplotype with cardiomyopathy.

Allele frequencies of the variants are highly informative for the differentiation of the P/LP variants versus the B/LB variants. Without exception, all 6 P/LP variants are absent from controls in the exome sequencing data of gnomAD<sup>89</sup> (ACMG Moderate Pathogenic Evidence Level: PM2). In contrast, 8 of the 9 B/LB variants are relatively common in the gnomAD (ACMG Benign Evidence Levels: benign stand alone BA1 or strong BS1). The other B/LB variant p.Glu553Asp is classified as likely benign with multiple lines of benign computational evidence (ACMG Evidence BP4).<sup>22</sup>

## COMMON VARIANTS OF BAG3

### Nonsynonymous coding variant

#### p.Cys151Arg

Common variants from a number of genetic loci have been reported in association with DCM. In a study by Villard et al,<sup>92</sup> a common nonsynonymous coding variant, rs2234962 at BAG3 (p.Cys151Arg), was reported in association with DCM, while the minor allele (allele C encoding Arg151) is protective (odds ratio, 0.64 [95% CI, 0.55–0.74]). The association of rs2234962 and DCM was consequently replicated in independent studies.<sup>93,94</sup> In the genome-wide association meta-analysis comprising 47 309 cases and 930 014 controls of European ancestry, Shah et al identified the strongest association signal with DCM from the intronic SNP (single-nucleotide polymorphism) rs17617337.<sup>94</sup> The SNP rs17617337 is in high LD with

the nonsynonymous rs2234962 ( $r^2=0.99$ ), thus representing the same association signal.<sup>94</sup> One study also showed that rs2234962 was associated with LV end-diastolic diameter in both patients with DCM and those with ischemic heart failure.<sup>95</sup> In the general population, rs2234962 is associated with LV functional and structural parameters, including LV end-diastolic volume,<sup>96</sup> LV end-systolic volume,<sup>96</sup> and LV ejection fraction.<sup>96,97</sup> In addition, a recent study showed that rs2234962 is associated with elevated levels of serum troponin, a marker of cardiomyocyte damage.<sup>98</sup>

The nonsynonymous p.Cys151Arg variant, located in between the 2 IPV motifs, is classified as B/LB by both ClinVar and the ACMG criteria, mainly because of its common allele frequency (Table S2). Bioinformatic approaches predict p.Cys151Arg with conflicting results, ie, damaging by SIFT (Sorting Intolerant From Tolerant), probably damaging by Polyphen-HumDiv (correlated with evolutionary conservation), but benign by Polyphen-HumVar (correlated with allele frequency). In addition to the protective effect, the position of p.Cys151Arg without involving a functional domain supports a benign classification, while all P/LP mutations of BAG3 (disease-causing) involve a functional domain. Significant differences in allele frequencies of p.Cys151Arg is seen among different racial populations. For example the putatively protective allele Arg151 is extremely rare in East Asian populations (<https://www.ncbi.nlm.nih.gov/snp/rs2234962>), which suggests evolutionary selective pressure related to the pleiotropic function of BAG3. No expression quantitative trait loci (eQTLs) tagged by rs2234962 in heart or other tissues have been observed by the Genotype-Tissue Expression (GTEx) project (<https://www.gtexportal.org/>). Concerning the molecular mechanisms underlying the genetic association of p.Cys151Arg, a recent study by Perez-Bermejo et al showed that p.Cys151Arg enhanced the recruitment of factors critical to the maintenance of myofibril integrity and improved response to proteotoxic stress in cardiomyocytes.<sup>99</sup>

### eQTLs of BAG3

According to GTEx project data, a number of SNPs have been identified as having an association with the expression levels of BAG3 in different tissues (Table 2). As shown by the harmonized data in the MR-Base platform (<https://www.mrbase.org/>), 5 eQTL SNPs are also associated with primary cardiomyopathies by the FinnGen biobank genome-wide association study (id:finn-b-I9\_CARDMPRI, [https://gwas.mrcieu.ac.uk/datasets/finn-b-I9\\_CARDMPRI/](https://gwas.mrcieu.ac.uk/datasets/finn-b-I9_CARDMPRI/); <https://www.finnngen.fi/>), which includes 2221 cases and 156 711 controls. The eQTL effects are seen in human tissues, including the tibial artery, sun-exposed skin in the lower leg, the sigmoid colon, skeletal muscle, and subcutaneous

**Table 2. The eQTLs of BAG3 and Their Associations With Cardiomyopathies**

SNP	Expression of BAG3	pval.eQTL	pval.cardiomyopathy	effect_allele.eQTL	beta.eQTL	beta.cardiomyopathy	EAF	r2 with p.Cys151Arg
rs196334	Artery tibial	7.29E-06	2.85E-08	T	-0.176	-0.177	0.546	0.139
rs196336	Skin sun-exposed lower leg	3.98E-19	2.98E-08	C	-0.346	-0.178	0.546	0.139
rs196298	Colon sigmoid	2.74E-06	8.62E-07	A	-0.316	-0.178	0.264	0.063
rs171910	Muscle skeletal	2.00E-06	5.01E-03	G	-0.224	-0.101	0.738	0.076
rs17099080	Adipose subcutaneous	1.06E-05	7.24E-03	C	0.224	0.098	0.253	0.034
rs11384142	Heart atrial appendage	1.25E-06	0.103	T	-1.084	-0.266	0.010	0.016
rs11598594	Skin not sun exposed suprapubic	1.01E-05	0.851	C	-0.799	-0.038	0.006	0.005
rs3887073	Brain anterior cingulate cortex BA24	4.32E-07	0.938	T	0.398	-0.004	0.103	0.005

BAG3 indicates Bcl-2-associated athanogene 3; EAF, effect allele frequency; eQTL, expression quantitative trait locus; and SNP, single nucleotide polymorphism.

adipose tissue. These eQTL SNPs have low LD  $r^2$  values with the nonsynonymous coding variant p.Cys151Arg (Figure S1, plotted by the NCI LDmatrix Tool, <https://analysis-tools.cancer.gov/LDlink/?tab=ldmatrix>). Two SNPs, rs196334 and rs196336, are in tight LD with  $r^2=1$  in the European population, representing the same genetic signal. No eQTL is identified in the left ventricle of the heart. Interestingly, these eQTLs are associated with upregulated expression of BAG3 in noncardiac tissues, but are associated with increased risk of cardiomyopathies. Mechanistic and phenotypic study of these eQTLs and their roles in cardiomyopathies is warranted as they represent a unique opportunity to study the potential therapeutic as well as the possible detrimental effects of manipulating BAG3 expression.

## CONCLUSIONS

Genetic analysis, including most importantly the phenotypic effect analysis of DNA variants, has been a successful approach to understand gene structure and function. A number of mutations in the BAG3 gene have been reported in relation to DCM causality, highlighting the critical role of BAG3 in cardiomyocytes. The observed phenotypic effects of the BAG3 mutations have provided new knowledge about essential functional domains. For instance, all of the truncating mutations before the 451th amino acid position in the BAG domain are P/LP variants, which indicates the essential/functional length of the BAG3 protein. All of the P/LP deletions involve the WW domain or the BAG domain, and all of the P/LP nsSNPs involve the second IPV domain or the BAG domain, highlighting the importance of these 3 protein domains in the function of BAG3 in the heart, whereas B/LB variants undermine the importance of the PXXP region.

Because of its critical role in cardiomyocytes in DCM, BAG3 is a clear major candidate gene in secondary DCM, including but not limited to peripartum DCM and severe cardiotoxicity of the proteasome inhibitor Bortezomib for cancer chemotherapy. A correlation between BAG3 mutations and myocarditis that was identified in a recent study<sup>68</sup> implies that BAG3 may also be involved in viral insults of cardiomyocytes. In the era of precision medicine and genomic medicine, genetic information acquired by DNA sequencing may help to identify high-risk individuals, with the goal of preventing adverse events related to BAG3. Patients with a positive family history of DCM and presence of P/LP genetic variants of the BAG3 gene, warrant extra attention for the incidence of secondary DCM-like PPCM, cardiotoxicity of cancer chemotherapy, or myocarditis in viral infections. In contrast, decreased susceptibility of secondary heart failure or viral myocarditis might be expected in patients with the protective allele of BAG3. These possibilities warrant further research.



In contrast to its protective roles in cardiomyocytes, *BAG3* has been identified as favoring tumor growth and antiapoptotic effects in cancerous cells.<sup>76,79</sup> This has led to *BAG3* being considered as a target of anticancer therapy, particularly in pancreatic cancer and heme-malignancies.<sup>76</sup> This conundrum can be obviated by developing therapeutic strategies in heart failure with reduced ejection fraction that do not cause incorporation of the exogenous *BAG3* into the native genome.<sup>100</sup> Additional studies are warranted to dissect the relationship between PPCM and *BAG3*.

## ARTICLE INFORMATION

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Dr Feldman is the founder of Renovacor, Inc., a publicly traded biotechnology company that is developing gene therapy for treatment of patients with cardiovascular and central nervous system diseases attributable to genetic variants in *BAG3*. He holds equity in the company and receives consultation fees, and is a member of the scientific advisory board of the company and serves as the Chief Scientific Advisor. Although he holds a grant from the company that funds the research in his laboratory in part, the work presented here was not funded by Renovacor. The remaining authors have no disclosures to report.

### Supplemental Material

Tables S1–S2  
Figure S1

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# **SUPPLEMENTAL MATERIAL**

**Table S1. Reported disease-related variants in BAG3**

GRCh37	NM_004281.4	Protein	Clinical significance (ClinVar)	HGMD_C1	Condition(s)	Notes (PMID)	Coding Variant type	Domain
121411253	c.72dup	p.Gly25fs	Pathogenic Pathogenic/Likely	DM	not provided   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		Frame-shift (truncating)	
121411264	c.77G>A	p.Trp26Ter	pathogenic	DM	not provided   Primary dilated cardiomyopathy		Nonsense (truncating)	
121411265	c.78G>A	p.Trp26Ter	Pathogenic	DM?	not provided		Nonsense (truncating)	
121411275	c.88A>G	p.Ile30Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		Missense (non-truncating)	WW domain
121411295	c.108G>A	p.Trp36Ter	Pathogenic	DM	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided		Nonsense (truncating)	
121429393	c.211C>T	p.Arg71Trp	Conflicting interpretations of pathogenicity	DM	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided		Missense (non-truncating)	
121429412	c.230C>T	p.Pro77Leu	Conflicting interpretations of pathogenicity	DM?	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not specified   not provided   Cardiomyopathy		Missense (non-truncating)	
121429431	c.249C>A	p.His83Gln	Conflicting interpretations of pathogenicity	DM	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   Primary dilated cardiomyopathy   not provided   not specified   Myofibrillar myopathy, BAG3-related		Missense (non-truncating)	
121429450	c.268C>T	p.Arg90Ter	Pathogenic	DM	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided		Nonsense (truncating)	
121429462	c.280A>T	p.Ile94Phe	Conflicting interpretations of pathogenicity	DM	Cardiovascular phenotype   not specified   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided   Primary dilated cardiomyopathy   Myofibrillar myopathy, BAG3-related		Missense (non-truncating)	IPV motif
121429499	c.317G>A	p.Arg106Gln	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not specified		Missense (non-truncating)	
121429508	c.329dup	p.His112fs	Pathogenic	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		Frame-shift (truncating)	
121429511	c.329C>T	p.Pro110Leu	Uncertain significance	NA	not provided   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		Missense (non-truncating)	

121429525	c.343C>T	p.Pro115Ser	Uncertain significance	DM	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429543	c.361C>T	p.Arg121Ter	Pathogenic	DM	not provided	Nonsense (truncating)
121429544	c.362G>A	p.Arg121Gln	Uncertain significance	DM?	not specified	Missense (non-truncating)
121429549	c.367C>T	p.Arg123Ter	Pathogenic/Likely pathogenic	DM	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Cardiovascular phenotype Primary dilated cardiomyopathy not provided	Nonsense (truncating)
121429550	c.368G>A	p.Arg123Gln	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)
121429562	c.381_382dup	p.Ala128fs	Pathogenic	DM	not provided	(truncating)
121429598	c.416G>A	p.Arg139Gln	Uncertain significance	DM?	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121429639	c.457C>T	p.Gln153Ter	Pathogenic	DM	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Nonsense (truncating)
121429645	c.463G>A	p.Ala155Thr	Conflicting interpretations of pathogenicity	DM?	Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Hypertrophic cardiomyopathy	Missense (non-truncating)
121429647	c.465A>G	p.Ala155=	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related not specified not provided Dilated cardiomyopathy 1HH Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121429647	c.468GGC[4]	p.Ala160dup	Benign/Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided not specified Myofibrillar Myopathy, Dominant Dilated Cardiomyopathy, Dominant	
121429649	c.467C>A	p.Ala156Glu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)

121429649	c.467C>G	p.Ala156Gly	Conflicting interpretations of pathogenicity	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121429649	c.467C>T	p.Ala156Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121429663	c.481C>T	p.Gln161Ter	Pathogenic	DM?	Cardiovascular phenotype	Nonsense (truncating)	
121429667	c.485C>T	p.Pro162Leu	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121431765	c.508-2A>G		Pathogenic	NA	not provided		
121431768	c.509G>A	p.Arg170Gln	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	
121431789	c.531dup	p.Asp178Ter	Pathogenic	DM	not provided	Nonsense (truncating)	
121431881	c.622A>C	p.Ile208Leu	Uncertain significance	DM?	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	IPV motif
121431884	c.625C>G	p.Pro209Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	IPV motif
121431884	c.625C>T	p.Pro209Ser	Pathogenic/Likely pathogenic	DM	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Abnormality of the musculature not provided	Missense (non-truncating)	IPV motif
121431885	c.626C>A	p.Pro209Gln	Pathogenic	DM	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Peripheral neuropathy Myofibrillar myopathy, BAG3-related Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	IPV motif
121431885	c.626C>T	p.Pro209Leu	Pathogenic	DM	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121431904	c.645C>A	p.Asn215Lys	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121431904	c.645C>T	p.Asn215=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified		
121431911	c.652C>T	p.Arg218Trp	Uncertain significance	DM	Dilated cardiomyopathy 1HH Familial isolated arrhythmogenic right ventricular dysplasia Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Cardiovascular phenotype	Missense (non-truncating)	
121431958	c.699C>A	p.Tyr233Ter	Pathogenic	DM	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Nonsense (truncating)	



121432002	c.743A>G	p.His248Arg	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)
121432010	c.751C>T	p.Gln251Ter	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Myofibrillar	Nonsense (truncating)
121432031	c.772C>T	p.Arg258Trp	Benign/Likely benign	DM?	myopathy Cardiovascular phenotype Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified not	Missense (non-truncating)
121432043	c.784G>A	p.Ala262Thr	Uncertain significance	DM	provided	Missense (non-truncating)
121432044	c.785C>T	p.Ala262Val	Conflicting interpretations of pathogenicity	DM?	Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified not provided Dilated cardiomyopathy 1A	Missense (non-truncating)
121432080	c.821C>A	p.Ser274Ter	Pathogenic	DM	not provided not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Nonsense (truncating)
121432080	c.821C>T	p.Ser274Leu	Conflicting interpretations of pathogenicity	NA	Primary familial dilated cardiomyopathy not provided	Missense (non-truncating)
121432095	c.836C>A	p.Ser279Ter	Likely pathogenic	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Nonsense (truncating)
121432095	c.836C>T	p.Ser279Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121432100	c.841G>A	p.Ala281Thr	Uncertain significance	DM?	Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Missense (non-truncating)
121432151	c.892G>A	p.Val298Met	Uncertain significance	DM?	not specified Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Myofibrillar Myopathy, Dominant Dilated Cardiomyopathy, Dominant Cardiovascular phenotype	Missense (non-truncating)
121433902	BAG3, EX4DEL		Pathogenic	DM	Dilated cardiomyopathy 1HH	Deletion
121435976	c.910C>T	p.Gln304Ter	Pathogenic	DM	not provided	Nonsense (truncating)

121435991	c.925C>G	p.Arg309Gly	Uncertain significance	NA	not provided	Missense (non-truncating)	PXXP region
121435991	c.925C>T	p.Arg309Ter	Pathogenic	DM	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Primary familial dilated cardiomyopathy Primary dilated cardiomyopathy not provided	Nonsense (truncating)	
121436009	c.943T>A	p.Ser315Thr	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	PXXP region
121436012	c.946C>T	p.Gln316Ter	Pathogenic/Likely pathogenic	DM	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Nonsense (truncating)	
121436027	c.961C>T	p.Pro321Ser	Uncertain significance	DM?	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436030	c.964G>T	p.Glu322Ter	Pathogenic	DM	not provided	Nonsense (truncating)	
121436081	c.1015A>G	p.Ile339Val	Uncertain significance	DM	not specified	Missense (non-truncating)	PXXP region
121436084	c.1018C>T	p.Gln340Ter	Pathogenic	DM	not provided	Nonsense (truncating)	
121436153	c.1087G>T	p.Glu363Ter	Pathogenic	DM	not provided	Nonsense (truncating)	
121436153	c.1088_1708c	p.Glu363_Pro	Likely pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Deletion	
121436204	c.1138C>T	p.Pro380Ser	Benign/Likely benign	DM	Myofibrillar myopathy, BAG3-related not provided not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Cardiovascular phenotype Atrial fibrillation	Missense (non-truncating)	PXXP region
121436267	c.1201G>A	p.Ala401Thr	Uncertain significance	NA	not provided	Missense (non-truncating)	PXXP region
121436280	c.1214C>T	p.Ala405Val	Conflicting interpretations of pathogenicity	DM?	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436306	c.1240G>A	p.Glu414Lys	Conflicting interpretations of pathogenicity	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121436306	c.1240G>T	p.Glu414Ter	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Nonsense (truncating)	
121436309	c.1243G>T	p.Ala415Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	

121436309	c.1245_1246c	p.Pro416fs	Pathogenic	DM	not provided	(truncating)	
121436365	c.1299G>A	p.Gln433=	Likely benign	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		
121436365	c.1300_1309c	p.Gln437fs	Likely pathogenic	DM	not provided	(truncating)	
121436382	c.1316_1317c	p.Val439fs	Pathogenic	DM	not provided	(truncating)	
121436419	c.1353C>A	p.Tyr451Ter	Pathogenic	DM	not provided   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Nonsense (truncating)	
121436429	c.1363G>A	p.Glu455Lys	Pathogenic	DM	not provided   Primary dilated cardiomyopathy   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436451	c.1385T>C	p.Leu462Pro	Likely pathogenic	DM	Dilated cardiomyopathy 1HH   not provided	Missense (non-truncating)	BAG domain
121436474	c.1408C>T	p.Pro470Ser	Likely pathogenic	DM	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436477	c.1411G>A	p.Glu471Lys	Uncertain significance	DM	Cardiovascular phenotype   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436483	c.1417C>T	p.Arg473Ter	Conflicting interpretations of pathogenicity	DM	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided   Dilated cardiomyopathy 1A	Nonsense (truncating)	
121436483	c.1418dup	p.Ala474fs	Likely pathogenic	NA	Dilated cardiomyopathy 1HH	(truncating)	
121436489	c.1423G>A	p.Asp475Asn	Conflicting interpretations of pathogenicity	DM?	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   not specified	Missense (non-truncating)	BAG domain
121436496	c.1430G>A	p.Arg477His	Uncertain significance	DM	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436624	c.1558C>T	p.Pro520Ser	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121430894			Uncertain significance	NA	See cases		
121429353			Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		
121429353			Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		
121431747			Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		
121435956			Pathogenic	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Deletion	
121429343			Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Deletion	

121435966			Pathogenic	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Deletion	
121436993	c.*199A>C		Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121437012	c.*218G>C		Benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided		
121437032	c.*240GTT[1]		Benign	NA	Myofibrillar Myopathy, Dominant Dilated Cardiomyopathy, Dominant not provided		
121437061	c.*267C>A		Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121437098	c.*306_*307del		Benign	NA	Myofibrillar Myopathy, Dominant Dilated Cardiomyopathy, Dominant not provided	Deletion	
121437113	c.*319C>T		Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436825	c.*31C>T		Benign/Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121437193	c.*399G>T		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121437196	c.*402T>C		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121437249	c.*455G>A		Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121437263	c.*469A>G		Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121437329	c.*535A>G		Benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided		
121411286	c.100_107del	p.Thr34fs	Likely pathogenic	NA	Primary dilated cardiomyopathy not provided	Frame-shift (truncating)	
121436068	c.1002T>G	p.Pro334=	Benign	NA	not provided not specified Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436077	c.1011C>T	p.Ile337=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436078	c.1012C>T	p.Pro338Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region

121436083	c.1017T>A	p.Ile339=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121411288	c.101C>T	p.Thr34Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	WW domain
121436086	c.1020A>G	p.Gln340=	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436093	c.1027C>T	p.Arg343Cys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	PXXP region
121436092	c.1027del	p.Arg343fs	Pathogenic	DM	not provided	(truncating)	
121436094	c.1028G>A	p.Arg343His	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436094	c.1028G>C	p.Arg343Pro	Uncertain significance	NA	not provided	Missense (non-truncating)	PXXP region
121436094	c.1028G>T	p.Arg343Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	PXXP region
121436095	c.1029C>A	p.Arg343=	Conflicting interpretations of pathogenicity	NA	Dilated cardiomyopathy 1HH Cardiovascular phenotype not specified Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Myofibrillar myopathy, BAG3-related		
121411289	c.102C>G	p.Thr34=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436096	c.1031_1032c	p.Lys344fs	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)	
121436098	c.1032A>T	p.Lys344Asn	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436099	c.1034_1038c	p.Glu345fs	Pathogenic	NA	Cardiovascular phenotype	(truncating)	
121436100	c.1034A>T	p.Glu345Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436101	c.1035G>A	p.Glu345=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436101	c.1035G>C	p.Glu345Asp	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Missense (non-truncating)	PXXP region
121411290	c.103G>A	p.Gly35Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
121411290	c.103G>T	p.Gly35Cys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain

121436113	c.1047A>G	p.Lys349=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436116	c.1050T>G	p.Pro350=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided		
121436121	c.1055C>G	p.Ser352Cys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436123	c.1057C>T	p.Gln353Ter	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Nonsense (truncating)	
121436121	c.1057del	p.Gln353fs	Pathogenic	DM	not provided	(truncating)	
121436124	c.1058A>G	p.Gln353Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121411292	c.105C>G	p.Gly35=	Likely benign	NA	not specified		
121436129	c.1063C>T	p.Pro355Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436130	c.1064C>T	p.Pro355Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436129	c.1067del	p.Pro356fs	Pathogenic/Likely pathogenic	NA	Primary dilated cardiomyopathy Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)	
121436144	c.1078G>C	p.Glu360Gln	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436149	c.1083G>A	p.Lys361=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided		
121436149	c.1083G>C	p.Lys361Asn	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436151	c.1085T>C	p.Val362Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436152	c.1086A>T	p.Val362=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436154	c.1088A>C	p.Glu363Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Missense (non-truncating)	PXXP region
121436162	c.1096G>A	p.Val366Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436162	c.1096G>C	p.Val366Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121411296	c.109C>T	p.Pro37Ser	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	WW domain
121436166	c.1100C>G	p.Pro367Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	PXXP region

121436171	c.1105G>C	p.Ala369Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436175	c.1109C>T	p.Pro370Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436184	c.1118G>A	p.Cys373Tyr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Primary familial dilated cardiomyopathy not specified	Missense (non-truncating)	PXXP region
121436184	c.1118G>C	p.Cys373Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121411298	c.111C>G	p.Pro37=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121411298	c.111C>T	p.Pro37=	Likely benign	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified		
121436187	c.1121C>T	p.Pro374Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436191	c.1125T>C	p.Pro375=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436192	c.1126C>T	p.Pro376Ser	Uncertain significance	NA	not provided	Missense (non-truncating)	PXXP region
121436193	c.1127C>A	p.Pro376His	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436194	c.1128C>T	p.Pro376=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121411298	c.112TTC[1]	p.Phe39del	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Deletion	
121436199	c.1133C>G	p.Pro378Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436201	c.1135G>A	p.Gly379Ser	Uncertain significance	NA	not specified	Missense (non-truncating)	PXXP region
121436201	c.1136del	p.Gly379fs	Pathogenic	DM	not provided	(truncating)	
121436205	c.1139C>T	p.Pro380Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436206	c.1140T>G	p.Pro380=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436207	c.1141T>C	p.Ser381Pro	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436209	c.1143T>C	p.Ser381=	Likely benign	NA	not provided		
121411301	c.114C>T	p.Phe38=	Likely benign	NA	not specified		

121436218	c.1152C>T	p.Pro384=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436214	c.1153_1160c	p.Ser385fs	Pathogenic	DM	not provided	(truncating)	
121436218	c.1155_1157c	p.Ser386dup	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436222	c.1156T>G	p.Ser386Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436223	c.1157C>A	p.Ser386Tyr	Uncertain significance	NA	Cardiovascular phenotype Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436225	c.1159C>T	p.Pro387Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436226	c.1160C>T	p.Pro387Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436227	c.1161C>T	p.Pro387=	Likely benign	NA	not provided		
121436222	c.1161dup	p.Lys388fs	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)	
121436229	c.1163A>G	p.Lys388Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436232	c.1166G>A	p.Ser389Asn	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436234	c.1168G>A	p.Val390Met	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436235	c.1169T>C	p.Val390Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121411303	c.116T>C	p.Phe39Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
121436238	c.1172C>T	p.Ala391Val	Uncertain significance	NA	not provided	Missense (non-truncating)	PXXP region
121436240	c.1174A>G	p.Thr392Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436245	c.1179A>G	p.Glu393=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121411304	c.117C>T	p.Phe39=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436248	c.1182G>A	p.Glu394=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436245	c.1183_1184c	p.Arg395fs	Pathogenic	DM	not provided	(truncating)	
121436250	c.1184G>A	p.Arg395Lys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region



121436253	c.1187C>T	p.Ala396Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436265	c.1199C>T	p.Thr400Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436268	c.1202C>G	p.Ala401Gly	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436268	c.1202C>T	p.Ala401Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436268	c.1205del	p.Pro402fs	Pathogenic	DM	not provided	(truncating)	
121436276	c.1210G>A	p.Glu404Lys	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436279	c.1213G>T	p.Ala405Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436281	c.1215T>C	p.Ala405=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436283	c.1217C>T	p.Thr406Ile	Uncertain significance	NA	not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436286	c.1220C>T	p.Pro407Leu	Benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Myofibrillar myopathy, BAG3-related Cardiovascular phenotype	Missense (non-truncating)	PXXP region
121411309	c.122A>G	p.Asp41Gly	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
121436297	c.1231G>A	p.Gly411Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436296	c.1232_1234c	p.Gly411del	Uncertain significance	DM?	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Deletion	
121436307	c.1241A>G	p.Glu414Gly	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121436308	c.1242G>C	p.Glu414Asp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121436310	c.1244C>A	p.Ala415Asp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121436315	c.1249C>T	p.Pro417Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121436316	c.1250C>A	p.Pro417Gln	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121436317	c.1251A>G	p.Pro417=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided		

121436322	c.1257dup	p.Pro420fs	Pathogenic/Likely pathogenic	NA	not provided	Frame-shift (truncating)	
121436325	c.1259C>T	p.Pro420Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	BAG domain
121411312	c.125A>G	p.His42Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
121436328	c.1262G>A	p.Gly421Glu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Cardiovascular phenotype	Missense (non-truncating)	BAG domain
121436330	c.1264G>A	p.Val422Met	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436332	c.1267_1276c	p.Leu423fs	Pathogenic	DM	Inborn genetic diseases not provided	(truncating)	
121436340	c.1274T>A	p.Val425Glu	Uncertain significance	NA	not specified	Missense (non-truncating)	BAG domain
121436347	c.1281C>T	p.Ala427=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436352	c.1286T>C	p.Leu429Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436354	c.1288G>A	p.Glu430Lys	Uncertain significance	NA	Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436355	c.1289A>T	p.Glu430Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436357	c.1291A>G	p.Lys431Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Missense (non-truncating)	BAG domain
121436356	c.1292dup	p.Val432fs	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Cardiovascular phenotype	Frame-shift (truncating)	
121436362	c.1296_1297i	p.Gln433Ter	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Nonsense (truncating)	
121436362	c.1296A>G	p.Val432=	Benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Cardiovascular phenotype		
121436363	c.1297C>T	p.Gln433Ter	Likely pathogenic	NA	Primary dilated cardiomyopathy	Nonsense (truncating)	
121436364	c.1298A>G	p.Gln433Arg	Uncertain significance	NA	Myofibrillar Myopathy, Dominant Dilated Cardiomyopathy, Dominant	Missense (non-truncating)	BAG domain

121411316	c.129C>T	p.Asn43=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436368	c.1302G>A	p.Gly434=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436373	c.1307A>G	p.Glu436Gly	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436378	c.1312G>T	p.Ala438Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	BAG domain
121436384	c.1318G>A	p.Asp440Asn	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436387	c.1321A>G	p.Asn441Asp	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436389	c.1323C>A	p.Asn441Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436389	c.1323C>G	p.Asn441Lys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436389	c.1326dup	p.Glu443Ter	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Nonsense (truncating)	
121411320	c.133C>A	p.Arg45Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	WW domain
121411320	c.133C>T	p.Arg45Cys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Missense (non-truncating)	WW domain
121436410	c.1344C>T	p.Asp448=	Likely benign	NA	not specified		
121436410	c.1344del	p.Asp448fs	Pathogenic	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Frame-shift (truncating)	
121436411	c.1345A>T	p.Lys449Ter	Likely pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Nonsense (truncating)	
121436414	c.1348A>T	p.Lys450Ter	Likely pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Nonsense (truncating)	
121436420	c.1354C>T	p.Leu452=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified not provided		
121436428	c.1362C>G	p.Ile454Met	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436428	c.1362C>T	p.Ile454=	Likely benign	NA	not specified Cardiovascular phenotype Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided		

121436435	c.1369T>C	p.Tyr457His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121411323	c.136A>T	p.Thr46Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
121436438	c.1372T>C	p.Leu458=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121411325	c.138C>T	p.Thr46=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided		
121436460	c.1394T>C	p.Leu465Pro	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436462	c.1396G>A	p.Asp466Asn	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436464	c.1398T>C	p.Asp466=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436473	c.1407C>A	p.Asp469Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436475	c.1409C>T	p.Pro470Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436476	c.1410C>T	p.Pro470=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified not provided		
121436473	c.1410del	p.Glu471fs	Likely pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)	
121436480	c.1414G>A	p.Gly472Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436484	c.1418G>A	p.Arg473Gln	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436486	c.1420G>A	p.Ala474Thr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436488	c.1422C>T	p.Ala474=	Likely benign	NA	not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436491	c.1425T>C	p.Asp475=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436494	c.1428G>A	p.Val476=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436495	c.1429C>T	p.Arg477Cys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121411329	c.142A>C	p.Thr48Pro	Uncertain significance	NA	1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	WW domain

121436502	c.1436C>T	p.Ala479Val	Conflicting interpretations of pathogenicity	NA	not specified not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121411330	c.143C>A	p.Thr48Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
121436506	c.1440G>A	p.Arg480=	Likely benign	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436512	c.1446C>T	p.Asp482=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided		
121436513	c.1447G>A	p.Gly483Ser	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified	Missense (non-truncating)	BAG domain
121436517	c.1451T>A	p.Val484Asp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436518	c.1452C>A	p.Val484=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436519	c.1453A>G	p.Arg485Gly	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121436521	c.1455G>A	p.Arg485=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436535	c.1469T>C	p.Ile490Thr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	BAG domain
121411333	c.146G>A	p.Trp49Ter	Pathogenic	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Nonsense (truncating)	
121436536	c.1470C>T	p.Ile490=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided		
121436543	c.1477A>G	p.Lys493Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436557	c.1491A>G	p.Lys497=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436562	c.1496T>C	p.Ile499Thr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	BAG domain
121436565	c.1499A>T	p.Asp500Val	Uncertain significance	NA	not provided	Missense (non-truncating)	
121436569	c.1503C>A	p.Val501=	Benign/Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Cardiovascular phenotype not provided not specified		

121436569	c.1503C>T	p.Val501=	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121436570	c.1504C>G	p.Pro502Ala	Uncertain significance	NA	not provided	Missense (non-truncating)
121436584	c.1519del	p.Val507fs	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Frame-shift (truncating)
121436589	c.1523A>G	p.Tyr508Cys	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Hypertrophic cardiomyopathy	Missense (non-truncating)
121436593	c.1527A>G	p.Glu509=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121436594	c.1528C>T	p.Leu510Phe	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436597	c.1531C>T	p.Gln511Ter	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Nonsense (truncating)
121436600	c.1534C>T	p.Pro512Ser	Likely benign	NA	not provided	Missense (non-truncating)
121436607	c.1541A>G	p.Asn514Ser	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436620	c.1554T>A	p.Asp518Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121411342	c.155C>G	p.Pro52Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436629	c.1563G>A	p.Leu521=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121436636	c.1570A>G	p.Ile524Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436636	c.1570A>T	p.Ile524Phe	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436637	c.1571T>C	p.Ile524Thr	Uncertain significance	NA	Myofibrillar Myopathy, Dominant Dilated Cardiomyopathy, Dominant	Missense (non-truncating)
121436640	c.1574T>C	p.Met525Thr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436644	c.1578G>T	p.Glu526Asp	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436648	c.1582G>C	p.Gly528Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436652	c.1586C>G	p.Ala529Gly	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436652	c.1586C>T	p.Ala529Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)

WW domain

121436653	c.1587C>G	p.Ala529=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121436653	c.1587C>T	p.Ala529=	Conflicting interpretations of pathogenicity	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified not provided Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121436654	c.1588G>A	p.Val530Met	Conflicting interpretations of pathogenicity	NA	Dilated cardiomyopathy 1HH not provided Myofibrillar myopathy, BAG3-related not specified Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436659	c.1593A>G	p.Ala531=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436664	c.1598A>C	p.Asp533Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436665	c.1599C>A	p.Asp533Glu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436667	c.1601A>G	p.Lys534Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436667	c.1601A>T	p.Lys534Met	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121411347	c.160G>C	p.Val54Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating) WW domain
121436689	c.1623T>A	p.Asn541Lys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436690	c.1624G>T	p.Ala542Ser	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436691	c.1625C>T	p.Ala542Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)
121436694	c.1628_1633c	p.Glu543_Pro	Uncertain significance	NA	not provided	Missense (non-truncating)
121436696	c.1630G>A	p.Asp544Asn	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436696	c.1630G>C	p.Asp544His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)
121436694	c.1632_1658c	p.Asp544_Prc	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	

121436698	c.1632del	p.His546fs	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Frame-shift (truncating)	
121436700	c.1634C>G	p.Pro545Arg	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Primary dilated cardiomyopathy	Missense (non-truncating)	
121436703	c.1637A>T	p.His546Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121436705	c.1639A>C	p.Thr547Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Missense (non-truncating)	
121436707	c.1641A>G	p.Thr547=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436708	c.1642G>C	p.Glu548Gln	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121436715	c.1649A>G	p.Gln550Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121411351	c.164C>T	p.Pro55Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
121436717	c.1651C>A	p.Gln551Lys	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121436718	c.1652A>G	p.Gln551Arg	Uncertain significance	NA	not provided	Missense (non-truncating)	
121436725	c.1659A>T	p.Glu553Asp	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Cardiovascular phenotype	Missense (non-truncating)	
121411350	c.165del	p.Ser56fs	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)	
121436728	c.1663_1666c	p.Ala556fs	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)	
121436730	c.1664C>A	p.Thr555Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121436733	c.1667C>A	p.Ala556Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	
121436733	c.1667C>T	p.Ala556Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121436735	c.1669G>A	p.Ala557Thr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121436739	c.1673C>T	p.Ala558Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	



121436740	c.1674G>A	p.Ala558=	Conflicting interpretations of pathogenicity	NA	not specified Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121411354	c.167C>G	p.Ser56Cys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436751	c.1685C>T	p.Pro562Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436753	c.1687A>C	p.Ser563Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436759	c.1693A>G	p.Met565Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436764	c.1698A>G	p.Thr566=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121436760	c.1699_1702c	p.Thr568fs	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)
121436768	c.1702A>C	p.Thr568Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436768	c.1702A>G	p.Thr568Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436769	c.1703C>G	p.Thr568Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121436782	c.1717dup	p.Ala573fs	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Frame-shift (truncating)
121436786	c.1720G>T	p.Ala574Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436790	c.1724C>G	p.Pro575Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436790	c.1724C>T	p.Pro575Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121436791	c.1725G>A	p.Pro575=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121411358	c.173del	p.Gly58fs	Pathogenic	DM	not provided	(truncating)
121411360	c.173G>T	p.Gly58Val	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121411366	c.179A>G	p.Lys60Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121411377	c.180+10G>C		Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121411380	c.180+13C>T		Likely benign	NA	not specified	

121411631	c.180+264G>A		Benign	NA	not provided	
121411636	c.180+269C>G		Benign	NA	not provided	
121411370	c.180+3G>A		Uncertain significance	NA	not provided	
121411410	c.180+43C>T		Likely benign	NA	not provided not specified	
121411372	c.180+5G>C		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121429256	c.181-107A>G		Benign	NA	not provided	
121429353	c.181-10T>C		Likely benign	NA	not specified	
121429348	c.181-15C>T		Conflicting interpretations of pathogenicity	NA	not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121429072	c.181-291A>G		Benign	NA	not provided	
121429357	c.181-6T>C		Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121429354	c.181-9T>A		Benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided not specified	
121429363	c.181G>A	p.Glu61Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429367	c.185C>T	p.Thr62Ile	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429369	c.187C>G	p.Pro63Ala	Benign	NA	not provided Cardiovascular phenotype Atrial fibrillation Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified	Missense (non-truncating)
121429373	c.191C>T	p.Ser64Phe	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429382	c.200A>G	p.Asn67Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified	Missense (non-truncating)
121429386	c.204C>G	p.Gly68=	Likely benign	NA	not specified	
121429385	c.206dup	p.Ser70fs	Pathogenic	NA	not provided	(truncating)
121429391	c.209C>T	p.Ser70Phe	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121429394	c.212G>A	p.Arg71Gln	Benign/Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Dilated cardiomyopathy 1HH not specified Cardiovascular phenotype Myofibrillar myopathy, BAG3-related	Missense (non-truncating)

121429394	c.212G>T	p.Arg71Leu	Conflicting interpretations of pathogenicity	NA	not provided   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   not specified	Missense (non-truncating)	
121429413	c.231G>A	p.Pro77=	Benign/Likely benign	NA	not provided   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		
121429415	c.233C>T	p.Pro78Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121429421	c.239G>T	p.Arg80Met	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121429432	c.252_258dup	p.Pro87fs	Pathogenic Pathogenic/Likely pathogenic	NA	Primary dilated cardiomyopathy not provided   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	(truncating)	
121429440	c.258C>G	p.Tyr86Ter	pathogenic	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Nonsense (truncating)	
121429443	c.261C>T	p.Pro87=	Likely benign	NA	Cardiovascular phenotype   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		
121429444	c.262C>T	p.Gln88Ter	Pathogenic	NA	cardiomyopathy 1HH	Nonsense (truncating)	
121429446	c.264G>A	p.Gln88=	Likely benign	NA	not specified		
121429448	c.266T>A	p.Leu89His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121429454	c.272C>T	p.Pro91Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121429458	c.277dup	p.Tyr93fs	Pathogenic	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Frame-shift (truncating)	
121429459	c.277T>C	p.Tyr93His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121429465	c.283C>G	p.Pro95Ala	Uncertain significance	NA	not specified	Missense (non-truncating)	IPV motif
121429466	c.284C>T	p.Pro95Leu	Uncertain significance	NA	Primary familial hypertrophic cardiomyopathy Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121429468	c.286A>C	p.Ile96Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	IPV motif
121429471	c.289C>T	p.Pro97Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121429481	c.299A>G	p.His100Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121429487	c.305G>A	p.Gly102Asp	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	

121429488	c.306C>T	p.Gly102=	Conflicting interpretations of pathogenicity	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	
121429489	c.307G>A	p.Ala103Thr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429497	c.315C>G	p.Asn105Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429498	c.316C>T	p.Arg106Trp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429500	c.318G>T	p.Arg106=	Likely benign	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121429502	c.320A>C	p.Gln107Pro	Uncertain significance	NA	not provided	Missense (non-truncating)
121429504	c.322G>A	p.Val108Met	Uncertain significance	NA	Primary dilated cardiomyopathy Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429504	c.322G>C	p.Val108Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429504	c.322G>T	p.Val108Leu	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121429507	c.325C>T	p.His109Tyr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429512	c.331_332del	p.Phe111fs	Pathogenic	DM	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Frame-shift (truncating)
121429514	c.332T>C	p.Phe111Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429518	c.336T>C	p.His112=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	
121429519	c.337G>A	p.Val113Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121429521	c.339C>T	p.Val113=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	
121429521	c.339del	p.Tyr114fs	Likely pathogenic	NA	not provided	(truncating)
121429522	c.340T>C	p.Tyr114His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429523	c.341A>G	p.Tyr114Cys	Uncertain significance	NA	not provided	Missense (non-truncating)
121429530	c.348G>C	p.Gln116His	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)

121429530	c.350dup	p.Gly118fs	Likely pathogenic	NA	not provided	(truncating)
121429524	c.351_352ins	p.Gly118fs	Pathogenic	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)
121429537	c.355A>G	p.Met119Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121429541	c.359A>T	p.Gln120Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429554	c.372T>G	p.Thr124=	Likely benign	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	
121429557	c.375G>T	p.Glu125Asp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429559	c.377C>T	p.Ala126Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121429560	c.378G>A	p.Ala126=	Likely benign	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	
121429563	c.381A>T	p.Ala127=	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	
121429568	c.386C>T	p.Ala129Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   not provided   not specified	Missense (non-truncating)
121429569	c.387G>A	p.Ala129=	Likely benign	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not specified	
121429573	c.391C>G	p.Pro131Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121429574	c.392C>T	p.Pro131Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429582	c.400T>A	p.Ser134Thr	Conflicting interpretations of pathogenicity	NA	not specified   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121429585	c.403C>T	p.Gln135Ter	Pathogenic	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Nonsense (truncating)
121429590	c.408_414del	p.Pro137fs	Pathogenic	DM	not provided	(truncating)

121429597	c.415C>T	p.Arg139Trp	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Missense (non-truncating)
121429600	c.418G>A	p.Gly140Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)
121429602	c.420C>T	p.Gly140=	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121429611	c.431_445dup	p.Thr144_Asp	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121429614	c.432C>A	p.Thr144=	Likely benign	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121429627	c.445A>G	p.Lys149Glu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429631	c.449_450del	p.Gln150fs	Pathogenic	DM	not provided	(truncating)
121429633	c.451_452inv	p.Cys151His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)
121429633	c.451T>C	p.Cys151Arg	Benign/Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Cardiovascular phenotype	Missense (non-truncating)
121429634	c.452G>A	p.Cys151Tyr	Uncertain significance	NA	not provided	Missense (non-truncating)
121429642	c.460G>A	p.Val154Met	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)
121429642	c.460G>C	p.Val154Leu	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429643	c.461T>C	p.Val154Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429643	c.462_464dup	p.Ala160dup	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121429650	c.468G>A	p.Ala156=	Likely benign	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121429652	c.470C>T	p.Ala157Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)

121429653	c.471G>A	p.Ala157=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified	
121429655	c.473C>T	p.Ala158Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429656	c.474G>A	p.Ala158=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121429662	c.480C>T	p.Ala160=	Likely benign	NA	not specified	
121429665	c.483G>A	p.Gln161=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided not specified	
121429666	c.484C>A	p.Pro162Thr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121429666	c.484C>T	p.Pro162Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429669	c.487C>T	p.Pro163Ser	Uncertain significance	NA	Primary familial dilated cardiomyopathy Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121429666	c.488del	p.Pro163fs	Pathogenic	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Frame-shift (truncating)
121429676	c.494C>T	p.Ser165Phe	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121429680	c.498C>T	p.His166=	Benign/Likely benign	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121429681	c.499G>A	p.Gly167Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Cardiovascular phenotype	Missense (non-truncating)
121429683	c.501A>G	p.Gly167=	Likely benign	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121429684	c.502C>T	p.Pro168Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121429842	c.507+153A>G		Benign	NA	not provided	
121429706	c.507+17C>T		Benign	NA	not provided	
121429707	c.507+18T>C		Benign	NA	not specified	
121429747	c.507+58C>T		Likely benign	NA	not provided	
121429762	c.507+73C>G		Benign	NA	not provided	
121429696	c.507+7A>G		Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	

121429689	c.507G>C	p.Glu169Asp	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Primary familial dilated cardiomyopathy	Missense (non-truncating)
121431766	c.508_509del	p.Arg170fs	Pathogenic	DM	not provided	(truncating)
121431750	c.508-14_514del		Likely pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Deletion
121431764	c.508-3C>G		Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121431761	c.508-4_519dup		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121431762	c.508-4dup		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121431767	c.508C>T	p.Arg170Trp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)
121431771	c.512C>A	p.Ser171Tyr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121431771	c.512C>T	p.Ser171Phe	Uncertain significance	NA	not provided	Missense (non-truncating)
121431773	c.514C>T	p.Gln172Ter	Pathogenic	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Nonsense (truncating)
121431774	c.515A>G	p.Gln172Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121431779	c.520C>T	p.Pro174Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121431783	c.524C>T	p.Ala175Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121431787	c.528C>T	p.Ala176=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	
121431787	c.530_531del	p.Ala176_Ser	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Nonsense (truncating)
121431793	c.534C>G	p.Asp178Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121431796	c.537C>A	p.Cys179Ter	Pathogenic	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Nonsense (truncating)
121431801	c.542C>T	p.Ser181Phe	Uncertain significance	NA	Cardiovascular phenotype Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121431804	c.545C>T	p.Ser182Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)



121431806	c.547T>C	p.Ser183Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Primary familial dilated cardiomyopathy	Missense (non-truncating)
121431808	c.549C>G	p.Ser183=	Conflicting interpretations of pathogenicity	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Primary familial dilated cardiomyopathy	
121431806	c.549CTC[1]	p.Ser185del	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Deletion Missense (non-truncating)
121431810	c.551C>T	p.Ser184Phe	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Dilated cardiomyopathy 1HH Primary familial hypertrophic cardiomyopathy Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121431813	c.554C>T	p.Ser185Leu	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121431814	c.555G>A	p.Ser185=	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121431815	c.556G>C	p.Ala186Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121431816	c.557C>T	p.Ala186Val	Uncertain significance	NA	Dilated Cardiomyopathy, Dominant Myofibrillar Myopathy, Dominant Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121431823	c.564G>T	p.Leu188=	Conflicting interpretations of pathogenicity	NA	Dilated cardiomyopathy 1A	(truncating)
121431827	c.568_577del	p.Ser190fs	Likely pathogenic	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121431828	c.569C>T	p.Ser190Phe	Uncertain significance	NA		
121431832	c.573C>T	p.Ser191=	Likely benign	NA		
121431833	c.574G>A	p.Gly192Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)
121431833	c.574G>T	p.Gly192Cys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121431835	c.576C>T	p.Gly192=	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	

121431837	c.578G>A	p.Arg193Lys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121431839	c.580del	p.Ser194fs	Likely pathogenic	NA	not provided	(truncating)	
121431841	c.582C>G	p.Ser194Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121431844	c.585C>T	p.Ser195=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121431857	c.598C>T	p.Gln200Ter	Pathogenic	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Nonsense (truncating)	
121431859	c.600G>C	p.Gln200His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121431864	c.605C>T	p.Pro202Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	IPV motif
121431865	c.606G>A	p.Pro202=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121431865	c.606G>T	p.Pro202=	Benign/Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified not provided Myofibrillar myopathy, BAG3-related		
121431866	c.607C>A	p.Arg203=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121431866	c.607C>T	p.Arg203Trp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121431865	c.607dup	p.Arg203fs	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Frame-shift (truncating)	
121431867	c.608G>A	p.Arg203Gln	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Missense (non-truncating)	IPV motif
121431867	c.608G>C	p.Arg203Pro	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121431869	c.610G>A	p.Gly204Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121431871	c.612G>A	p.Gly204=	Benign/Likely benign	NA	not specified not provided		
121431872	c.613del	p.Tyr205fs	Likely pathogenic	NA	not provided	(truncating)	

121431875	c.616A>C	p.Ile206Leu	Conflicting interpretations of pathogenicity	NA	Premature ventricular contraction   Cardiomyopathy   Cardiovascular phenotype   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   not provided   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	IPV motif
121431875	c.616A>G	p.Ile206Val	Uncertain significance	NA	not provided	Missense (non-truncating)	IPV motif
121411247	c.62_72del	p.Pro21fs	Pathogenic	DM	not provided	(truncating)	
121431882	c.623T>C	p.Ile208Thr	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	IPV motif
121431886	c.627G>A	p.Pro209=	Conflicting interpretations of pathogenicity	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   not specified		
121431886	c.627G>C	p.Pro209=	Likely benign	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		
121431887	c.628G>T	p.Val210Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	IPV motif
121411249	c.62C>G	p.Pro21Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	WW domain
121431893	c.634C>A	p.His212Asn	Uncertain significance	NA	Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121431895	c.636C>T	p.His212=	Likely benign	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   not provided		
121431896	c.637G>A	p.Glu213Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	IPV motif
121431899	c.640C>T	p.Gln214Ter	Pathogenic	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Nonsense (truncating)	
121431905	c.646G>A	p.Val216Ile	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121431905	c.646G>T	p.Val216Phe	Uncertain significance	NA	not provided	Missense (non-truncating)	
121431909	c.652del	p.Arg218fs	Pathogenic	DM	Dilated cardiomyopathy 1HH   not provided	Frame-shift (truncating)	
121431912	c.653G>A	p.Arg218Gln	Conflicting interpretations of pathogenicity	NA	Cardiovascular phenotype   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not specified   not provided   Left ventricular noncompaction	Missense (non-truncating)	
121431912	c.654del	p.Pro219fs	Pathogenic	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)	

121431914	c.655C>T	p.Pro219Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121431917	c.658G>A	p.Ala220Thr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121431927	c.668C>G	p.Pro223Arg	Uncertain significance	NA	Primary familial hypertrophic cardiomyopathy Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121431927	c.668C>T	p.Pro223Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121431928	c.670dup	p.Ser224fs	Likely pathogenic	NA	Dilated cardiomyopathy 1HH	(truncating)	
121431932	c.673T>C	p.Phe225Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121431936	c.677A>G	p.His226Arg	Uncertain significance	NA	not provided	Missense (non-truncating)	
121431938	c.679C>G	p.Gln227Glu	Conflicting interpretations of pathogenicity	NA	Cardiovascular phenotype not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121411254	c.67C>T	p.Pro23Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	WW domain
121411255	c.68C>T	p.Pro23Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
121431951	c.692C>T	p.Thr231Met	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121431952	c.693G>A	p.Thr231=	Conflicting interpretations of pathogenicity	NA	not provided not specified Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121431953	c.694C>G	p.His232Asp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121431954	c.695A>C	p.His232Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121431954	c.695A>T	p.His232Leu	Uncertain significance	NA	not specified	Missense (non-truncating)	
121431957	c.698A>G	p.Tyr233Cys	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121431963	c.704C>T	p.Ala235Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	

121431964	c.705G>A	p.Ala235=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified not provided		
121431964	c.705G>T	p.Ala235=	Likely benign	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121431968	c.709C>T	p.Gln237Ter	Pathogenic	NA	not provided	Nonsense (truncating)	
121411257	c.70C>T	p.Pro24Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
121431969	c.710A>G	p.Gln237Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121431970	c.711G>C	p.Gln237His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121431973	c.714G>A	p.Gly238=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121431974	c.715G>A	p.Glu239Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	
121431974	c.715G>T	p.Glu239Ter	Pathogenic	NA	not provided	Nonsense (truncating)	
121411258	c.71C>G	p.Pro24Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
121431984	c.727del	p.His243fs	Pathogenic	DM	not provided	(truncating)	
121411259	c.72C>A	p.Pro24=	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121411254	c.72del	p.Gly25fs	Pathogenic/Likely pathogenic	NA	not provided Primary dilated cardiomyopathy Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)	
121431989	c.730C>T	p.Gln244Ter	Pathogenic/Likely pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Primary dilated cardiomyopathy	Nonsense (truncating)	
121431996	c.737T>C	p.Val246Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121432009	c.751del	p.Gln251fs	Pathogenic	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Frame-shift (truncating)	
121432011	c.752del	p.Gln251fs	Pathogenic	DM	not provided	(truncating)	
121432020	c.761A>G	p.Asp254Gly	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121432024	c.765G>A	p.Trp255Ter	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Nonsense (truncating)	
121432025	c.766G>T	p.Glu256Ter	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Nonsense (truncating)	

121411263	c.76T>G	p.Trp26Gly	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
121432029	c.770C>T	p.Pro257Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121432030	c.771C>T	p.Pro257=	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified not provided		
121432032	c.773G>A	p.Arg258Gln	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121432034	c.775C>T	p.Pro259Ser	Uncertain significance	NA	Elevated circulating creatine kinase concentration	Missense (non-truncating)	
121432035	c.776C>T	p.Pro259Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121432040	c.781C>T	p.Arg261Trp	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified	Missense (non-truncating)	
121432041	c.782G>A	p.Arg261Gln	Conflicting interpretations of pathogenicity	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121432041	c.782G>C	p.Arg261Pro	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121432045	c.786G>A	p.Ala262=	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121432047	c.788C>T	p.Ala263Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	
121432048	c.789A>T	p.Ala263=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121432053	c.794C>T	p.Pro265Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Missense (non-truncating)	
121432054	c.795G>A	p.Pro265=	Likely benign	NA	Cardiovascular phenotype Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121432060	c.801G>T	p.Arg267Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121432061	c.802T>C	p.Ser268Pro	Conflicting interpretations of pathogenicity	NA	not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121432070	c.811C>T	p.Gln271Ter	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Nonsense (truncating)	
121432074	c.815G>T	p.Gly272Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	

121432077	c.818C>T	p.Ala273Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121432078	c.819A>G	p.Ala273=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121432081	c.822G>A	p.Ser274=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	
121432081	c.822G>C	p.Ser274=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121432081	c.822G>T	p.Ser274=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121432083	c.824del	p.Ser275fs	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)
121432085	c.826C>T	p.Arg276Trp	Uncertain significance	NA	not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	Missense (non-truncating)
121432086	c.827G>A	p.Arg276Gln	Conflicting interpretations of pathogenicity	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified	Missense (non-truncating)
121432087	c.828G>A	p.Arg276=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	
121432089	c.830A>T	p.Glu277Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121432093	c.834C>G	p.Gly278=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121432097	c.838C>T	p.Pro280Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121432103	c.844A>G	p.Arg282Gly	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121432113	c.854C>T	p.Thr285Met	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)
121432110	c.855_859dup	p.Leu287fs	Pathogenic/Likely pathogenic	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)
121432114	c.855G>A	p.Thr285=	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided not specified Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121432117	c.858A>G	p.Pro286=	Likely benign	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121432124	c.865T>G	p.Ser289Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)

121432129	c.870C>A	p.Pro290=	Likely benign	NA	not provided   Cardiovascular phenotype not specified   Cardiovascular phenotype   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	
121432129	c.870C>G	p.Pro290=	Benign/Likely benign	NA	not provided   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not specified	Missense (non-truncating)
121432131	c.872C>T	p.Ser291Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121432134	c.875C>G	p.Pro292Arg	Uncertain significance	NA		Missense (non-truncating)
121432139	c.880C>A	p.Arg294Ser	Uncertain significance	NA	not provided	Missense (non-truncating)
121432139	c.880C>T	p.Arg294Cys	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121432140	c.881G>A	p.Arg294His	Uncertain significance	NA	not provided   Primary dilated cardiomyopathy   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)
121432145	c.886C>T	p.His296Tyr	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121432147	c.888C>A	p.His296Gln	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	Missense (non-truncating)
121432147	c.888C>T	p.His296=	Conflicting interpretations of pathogenicity	NA	not specified   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   not provided   Cardiovascular phenotype   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	
121432145	c.889_890del	p.Thr297fs	Pathogenic	NA	not provided	(truncating)
121432148	c.889A>G	p.Thr297Ala	Uncertain significance	NA	not provided	Missense (non-truncating)
121432149	c.890C>T	p.Thr297Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121432150	c.891C>T	p.Thr297=	Likely benign	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   not specified	
121432155	c.896T>A	p.Val299Asp	Uncertain significance	NA	not provided	Missense (non-truncating)
121432156	c.897C>T	p.Val299=	Likely benign	NA	not provided	
121432161	c.903del	p.Arg301fs	Pathogenic	DM	not provided	(truncating)
121432164	c.905C>T	p.Pro302Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121432277	c.909+109C>T		Likely benign	NA	not provided	
121432184	c.909+16C>T		Likely benign	NA	not provided	

PXXP region



121432185	c.909+17G>A		Benign	NA	not specified		
121432359	c.909+191C>T		Likely benign	NA	not provided		
121432361	c.909+193G>A		Likely benign	NA	not provided		
121432169	c.909+1G>A		Likely pathogenic	NA	Primary dilated cardiomyopathy		
121432371	c.909+203G>A		Benign	NA	not provided		
121432440	c.909+272C>A		Benign	NA	not provided		
121432208	c.909+40A>G		Likely benign	NA	not provided		
121432172	c.909+4C>A		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121432172	c.909+4C>G		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121432172	c.909+4C>T		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121432168	c.909G>C	p.Gln303His	Uncertain significance	NA	not provided	Missense (non-truncating)	PXXP region
121435966	c.910-10C>G		Uncertain significance	NA	not specified		
121435955	c.910-21A>C		Benign	NA	not provided not specified Myofibrillar myopathy, BAG3-related		
121435718	c.910-258A>C		Likely benign	NA	not provided		
121435974	c.910-2A>C		Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121435979	c.915del	p.Pro305_Me	Pathogenic	DM	not provided	Nonsense (truncating)	
121435983	c.917T>C	p.Met306Thr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121435992	c.926G>A	p.Arg309Gln	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436010	c.944C>T	p.Ser315Phe	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436013	c.947A>G	p.Gln316Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified	Missense (non-truncating)	PXXP region
121436015	c.949C>T	p.Pro317Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436023	c.957C>G	p.Asn319Lys	Uncertain significance	NA	not provided	Missense (non-truncating)	PXXP region
121436033	c.967A>G	p.Ser323Gly	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436034	c.968G>A	p.Ser323Asn	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436032	c.969_972del	p.Lys324fs	Likely pathogenic	NA	Primary familial dilated cardiomyopathy	(truncating)	

121411283	c.96G>A	p.Pro32=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121411283	c.96G>T	p.Pro32=	Benign/Likely benign	NA	not specified not provided Cardiovascular phenotype Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436039	c.973C>G	p.Pro325Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436039	c.974del	p.Pro325fs	Pathogenic	DM	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Frame-shift (truncating)	
121436041	c.975A>T	p.Pro325=	Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436046	c.980C>G	p.Pro327Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436049	c.983T>C	p.Val328Ala	Uncertain significance	NA	Cardiovascular phenotype Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436050	c.984T>G	p.Val328=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436054	c.988C>T	p.Pro330Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	PXXP region
121436055	c.989C>T	p.Pro330Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	PXXP region
121436063	c.997C>T	p.Pro333Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	Missense (non-truncating)	PXXP region
121411286	c.99G>C	p.Gln33His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain
100027			Pathogenic	NA	See cases		
100027			Pathogenic	NA	See cases		
73232			Pathogenic	NA	not provided		
93297			Pathogenic	NA	See cases		
98087			Pathogenic	NA	not provided		
42347406			drug response	NA	Poly (ADP-Ribose) polymerase inhibitor response		
93283493			Pathogenic	NA	See cases		
94346520			Pathogenic	NA	not provided		
100780957			Pathogenic	NA	See cases		
103288313			Pathogenic	NA	See cases		

104633712	Pathogenic	NA	See cases	
105613040	Pathogenic	NA	not provided	
106003533	Pathogenic	NA	See cases	
108455687	Pathogenic	NA	not specified	
110022170	Pathogenic	NA	not provided	
114544537	Pathogenic	NA	not provided	
			Abnormality of the face Premature birth Patent foramen ovale Astigmatism Congenital ocular coloboma	
118891670	Uncertain significance	NA		
117019650	Pathogenic	NA	not specified	
117024753	Pathogenic	NA	Chromosome 10q26 deletion syndrome	
118247181	Pathogenic	NA	not provided	
			Intellectual disability Delayed speech and language development	
121410291	Likely benign	NA		
120782191	Uncertain significance	NA	not specified	
119996339	Pathogenic	NA	not provided	
121269222	Pathogenic	NA	not provided	
101953972	Pathogenic	NA	See cases	
102360249	Pathogenic	NA	See cases	
108685061	Pathogenic	NA	See cases	
109862345	Pathogenic	NA	See cases	
114460945	Likely pathogenic	NA	See cases	
113072857	Pathogenic	NA	See cases	
121032524	Pathogenic	NA	See cases	
119626076	Pathogenic	NA	See cases	
			Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121410882	Uncertain significance	NA		
			Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121411168	Uncertain significance	NA		
			Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121411168	Uncertain significance	NA		
			Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121411178	Uncertain significance	NA		
			Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121411178	Pathogenic	NA		Deletion

121411178			Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121411188			Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Deletion
121410869			Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121410829	c.-359C>T		Benign	NA	not provided	
121410670	c.-518C>T		Benign	NA	not provided	
121410645	c.-543T>C		Benign	NA	not provided	
121411060	c.-128G>T		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121411200	c.13A>G	p.Thr5Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified Dilated cardiomyopathy 1HH	Missense (non-truncating)
121411033	c.-155C>G		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121411171	c.-17G>A		Conflicting interpretations of pathogenicity	NA	Dilated cardiomyopathy 1HH not provided not specified Myofibrillar myopathy, BAG3-related	
121411170	c.-18G>A		Conflicting interpretations of pathogenicity	NA	not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121410980	c.-208C>T		Likely benign	NA	not provided	
121411207	c.20C>A	p.Ser7Ter	Likely pathogenic	NA	not provided	Nonsense (truncating)
121411209	c.22C>T	p.Pro8Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)
121411212	c.25A>G	p.Met9Val	Benign/Likely benign	NA	not provided Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified	Missense (non-truncating)
121410917	c.-271C>T		Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121410904	c.-284G>C		Conflicting interpretations of pathogenicity	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	
121410903	c.-285C>A		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	
121410903	c.-285C>T		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	

121411215	c.28A>G	p.Met10Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121410895	c.-293T>A		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121410894	c.-294A>C		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121411186	c.-2G>T		Uncertain significance	NA	Dilated cardiomyopathy 1HH		
121411189	c.2T>G	p.Met1Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121411217	c.30G>A	p.Met10Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121411217	c.30G>T	p.Met10Ile	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121411219	c.32A>T	p.Gln11Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121411221	c.34G>A	p.Val12Met	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121411223	c.36G>A	p.Val12=	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121411223	c.38_39dup	p.Ser14fs	Pathogenic	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Frame-shift (truncating)	
121411228	c.41C>T	p.Ser14Phe	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121411233	c.46A>G	p.Asn16Asp	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	Missense (non-truncating)	
121411236	c.49G>C	p.Gly17Arg	Uncertain significance	NA	not specified not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121411184	c.-4C>T		Conflicting interpretations of pathogenicity	NA	cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121411138	c.-50C>T		Benign	NA	not specified		
121411237	c.50G>C	p.Gly17Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	
121411242	c.55C>A	p.Arg19Ser	Uncertain significance	NA	not specified	Missense (non-truncating)	WW domain
121411242	c.55C>T	p.Arg19Cys	Uncertain significance	NA	Primary dilated cardiomyopathy Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	Missense (non-truncating)	WW domain

121411245	c.58G>A	p.Asp20Asn	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		Missense (non-truncating)	WW domain
121411192	c.5G>A	p.Ser2Asn	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		Missense (non-truncating)	
121411194	c.7G>T	p.Ala3Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		Missense (non-truncating)	
121411102	c.-86G>A		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related			
121411101	c.-87G>T		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related			
121411093	c.-95G>A		Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related			
119302758	119302758 - 124813305_Sii		Likely pathogenic	NA	not provided			
121411275	c.89_91delTC	p.Ile30_Asp3:	NA	DM	Cardiomyopathy, dilated		Indel (non-truncating)	WW domain
121429508	c.326A>G	p.His109Arg	NA	DM	Cardiomyopathy, dilated		Missense (non-truncating)	
121429647	c.474_476dup	p.Ala160dup	NA	DM	Left_ventricular_non_compaction			
121431768	c.508_1728del	Deletion of ex	NA	NA	Cardiomyopathy, dilated	30442290	Deletion	
121436382	c.1316T>A	p.Val439Glu	NA	DM?	Cardiomyopathy, dilated		Missense (non-truncating)	BAG domain
121411324	c.137C>T	p.Thr46Ile	NA	DM?	Noncompaction, left_ventricular		Missense (non-truncating)	WW domain
121411355	c.169_171del	p.Glu57Lysfs*	NA	DM	Cardiomyopathy, dilated		(truncating)	
121429609	c.427G>C	p.Glu143Gln	NA	DM?	Developmental_disorder		Missense (non-truncating)	
121429612	c.430A>G	p.Thr144Ala	NA	DM?	Myofibrillar_myopathy		Missense (non-truncating)	
121431355	SVLEN=-1779	Deletion	NA	DM	Cardiomyopathy, dilated		Deletion	
121432038	c.781_795del	p.Arg261_Pro	NA	DM	Myofibrillar_myopathy		Deletion	
121432062	c.803C>A	p.Ser268*	NA	DM	Sudden_cardiac_arrest		Nonsense (truncating)	
121432142	c.883G>A	p.Val295Met	NA	DM?	Cardiomyopathy, hypertrophic		Missense (non-truncating)	
121436136	c.1070_1660c	Deletion	NA	NA	Cardiomyopathy, dilated	29961767	Deletion	
121436408	c.1342G>A	p.Asp448Asn	NA	DM?	Brody_syndrome		Missense (non-truncating)	BAG domain
121436468	c.1402G>A	p.Val468Met	NA	DM	Cardiomyopathy, dilated		Missense (non-truncating)	BAG domain
121436505	c.1444_1445c	p.Asp482Argf	NA	DM?	Noncompaction, left_ventricular		(truncating)	
121436582	c.1516C>T	p.Gln506*	NA	DM?	Motor_&_sensory_neuropathy		Nonsense (truncating)	

121436723	c.1657G>A	p.Glu553Lys	NA	DM?	Cardiomyopathy, tako-tsubo	Missense (non-truncating)
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**Table S2. Missense(non-truncating) variants in BAG3**

GRCh37	NM_004281.4 Protein	Clinical significance (ClinVar)	HGMD_C1	Condition(s)	Domain	GnomAD Freq(all populations)
121411189	c.2T>G	p.Met1Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	.
121411192	c.5G>A	p.Ser2Asn	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	.
121411194	c.7G>T	p.Ala3Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	.
121411200	c.13A>G	p.Thr5Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not specified   Dilated cardiomyopathy 1HH	.
121411209	c.22C>T	p.Pro8Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related not provided   Cardiovascular phenotype   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   not specified	.
121411212	c.25A>G	p.Met9Val	Benign/Likely benign	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	0.0015
121411215	c.28A>G	p.Met10Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	.
121411217	c.30G>A	p.Met10Ile	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	.
121411217	c.30G>T	p.Met10Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	.
121411219	c.32A>T	p.Gln11Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	.
121411221	c.34G>A	p.Val12Met	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	3.25E-05
121411228	c.41C>T	p.Ser14Phe	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	.
121411233	c.46A>G	p.Asn16Asp	Uncertain significance	NA	not specified   not provided   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	3.25E-05
121411236	c.49G>C	p.Gly17Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	.
121411237	c.50G>C	p.Gly17Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	.

121411242	c.55C>T	p.Arg19Cys	Uncertain significance	NA	Primary dilated cardiomyopathy Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	0.0001
121411245	c.58G>A	p.Asp20Asn	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	WW domain	
121411249	c.62C>G	p.Pro21Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	WW domain	
121411254	c.67C>T	p.Pro23Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	WW domain	3.25E-05
121411255	c.68C>T	p.Pro23Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	
121411257	c.70C>T	p.Pro24Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	
121411258	c.71C>G	p.Pro24Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	
121411263	c.76T>G	p.Trp26Gly	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	
121411275	c.88A>G	p.Ile30Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	WW domain	3.25E-05
121411286	c.99G>C	p.Gln33His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	
121411288	c.101C>T	p.Thr34Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	WW domain	
121411290	c.103G>A	p.Gly35Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	
121411290	c.103G>T	p.Gly35Cys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	
121411296	c.109C>T	p.Pro37Ser	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	WW domain	
121411303	c.116T>C	p.Phe39Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	
121411309	c.122A>G	p.Asp41Gly	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	
121411312	c.125A>G	p.His42Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	
121411320	c.133C>A	p.Arg45Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	WW domain	
121411320	c.133C>T	p.Arg45Cys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	WW domain	6.51E-05



121411323	c.136A>T	p.Thr46Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Dilated cardiomyopathy	WW domain .	
121411329	c.142A>C	p.Thr48Pro	Uncertain significance	NA	1HH Myofibrillar myopathy, BAG3-related	WW domain .	
121411330	c.143C>A	p.Thr48Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain .	
121411342	c.155C>G	p.Pro52Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	WW domain .	
121411347	c.160G>C	p.Val54Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain .	
121411351	c.164C>T	p.Pro55Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	WW domain	3.26E-05
121411354	c.167C>G	p.Ser56Cys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Dilated cardiomyopathy		9.78E-05
121411360	c.173G>T	p.Gly58Val	Uncertain significance	NA	1HH Myofibrillar myopathy, BAG3-related	.	
121411366	c.179A>G	p.Lys60Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		3.27E-05
121429363	c.181G>A	p.Glu61Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		3.23E-05
121429367	c.185C>T	p.Thr62Ile	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Cardiovascular	.	
121429369	c.187C>G	p.Pro63Ala	Benign	NA	phenotype Atrial fibrillation Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified		0.003
121429373	c.191C>T	p.Ser64Phe	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121429382	c.200A>G	p.Asn67Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified	.	
121429391	c.209C>T	p.Ser70Phe	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121429393	c.211C>T	p.Arg71Trp	Conflicting interpretations of pathogenicity	DM	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Primary dilated cardiomyopathy Dilated cardiomyopathy 1HH not provided		0.0002

121429394	c.212G>A	p.Arg71Gln	Benign/Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Dilated cardiomyopathy 1HH not specified Cardiovascular phenotype Myofibrillar myopathy, BAG3-related			0.0241
121429394	c.212G>T	p.Arg71Leu	Conflicting interpretations of pathogenicity	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified			6.47E-05
121429412	c.230C>T	p.Pro77Leu	Conflicting interpretations of pathogenicity	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified not provided Cardiomyopathy			0.0002
121429415	c.233C>T	p.Pro78Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.		
121429421	c.239G>T	p.Arg80Met	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.		
121429431	c.249C>A	p.His83Gln	Conflicting interpretations of pathogenicity	DM	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Primary dilated cardiomyopathy not provided not specified Myofibrillar myopathy, BAG3-related			0.0029
121429448	c.266T>A	p.Leu89His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	IPV motif	.	
121429454	c.272C>T	p.Pro91Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	IPV motif	.	
121429459	c.277T>C	p.Tyr93His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Cardiovascular phenotype not specified Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Primary dilated cardiomyopathy Myofibrillar myopathy, BAG3-related	IPV motif		3.23E-05
121429462	c.280A>T	p.Ile94Phe	Conflicting interpretations of pathogenicity	DM	Myofibrillar myopathy, BAG3-related	IPV motif		0.0006
121429466	c.284C>T	p.Pro95Leu	Uncertain significance	NA	Primary familial hypertrophic cardiomyopathy Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	IPV motif	.	
121429468	c.286A>C	p.Ile96Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	IPV motif	.	
121429471	c.289C>T	p.Pro97Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	IPV motif	.	
121429481	c.299A>G	p.His100Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	IPV motif	.	

121429487	c.305G>A	p.Gly102Asp	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121429489	c.307G>A	p.Ala103Thr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121429497	c.315C>G	p.Asn105Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121429498	c.316C>T	p.Arg106Trp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	3.23E-05
121429499	c.317G>A	p.Arg106Gln	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified	.	
121429504	c.322G>A	p.Val108Met	Uncertain significance	NA	Primary dilated cardiomyopathy Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	3.23E-05
121429504	c.322G>C	p.Val108Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121429504	c.322G>T	p.Val108Leu	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121429507	c.325C>T	p.His109Tyr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121429511	c.329C>T	p.Pro110Leu	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121429514	c.332T>C	p.Phe111Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121429519	c.337G>A	p.Val113Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121429522	c.340T>C	p.Tyr114His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121429525	c.343C>T	p.Pro115Ser	Uncertain significance	DM	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121429530	c.348G>C	p.Gln116His	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121429537	c.355A>G	p.Met119Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121429541	c.359A>T	p.Gln120Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121429550	c.368G>A	p.Arg123Gln	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	.	

121429557	c.375G>T	p.Glu125Asp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.
121429559	c.377C>T	p.Ala126Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.
121429568	c.386C>T	p.Ala129Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided not specified	6.47E-05
121429573	c.391C>G	p.Pro131Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.
121429574	c.392C>T	p.Pro131Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.
121429582	c.400T>A	p.Ser134Thr	Conflicting interpretations of pathogenicity	NA	not specified Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	9.71E-05
121429597	c.415C>T	p.Arg139Trp	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	9.70E-05
121429598	c.416G>A	p.Arg139Gln	Uncertain significance	DM?	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.
121429600	c.418G>A	p.Gly140Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	.
121429627	c.445A>G	p.Lys149Glu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.
121429633	c.451T>C	p.Cys151Arg	Benign/Likely benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Cardiovascular phenotype	0.1475
121429642	c.460G>A	p.Val154Met	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	.
121429642	c.460G>C	p.Val154Leu	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	3.23E-05
121429643	c.461T>C	p.Val154Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.

121429645	c.463G>A	p.Ala155Thr	Conflicting interpretations of pathogenicity	DM?	Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Hypertrophic cardiomyopathy Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	0.0019
121429649	c.467C>G	p.Ala156Gly	Conflicting interpretations of pathogenicity	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	9.71E-05
121429649	c.467C>A	p.Ala156Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.
121429649	c.467C>T	p.Ala156Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	3.24E-05
121429652	c.470C>T	p.Ala157Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	6.47E-05
121429655	c.473C>T	p.Ala158Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.
121429666	c.484C>A	p.Pro162Thr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.
121429666	c.484C>T	p.Pro162Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.
121429667	c.485C>T	p.Pro162Leu	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.
121429669	c.487C>T	p.Pro163Ser	Uncertain significance	NA	Primary familial dilated cardiomyopathy Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	3.24E-05
121429676	c.494C>T	p.Ser165Phe	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Cardiovascular phenotype	.
121429681	c.499G>A	p.Gly167Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.
121429684	c.502C>T	p.Pro168Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Primary familial dilated cardiomyopathy	.
121429689	c.507G>C	p.Glu169Asp	Uncertain significance	NA		.

121431767	c.508C>T	p.Arg170Trp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided	6.47E-05
121431768	c.509G>A	p.Arg170Gln	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided	.
121431771	c.512C>A	p.Ser171Tyr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	.
121431774	c.515A>G	p.Gln172Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	.
121431779	c.520C>T	p.Pro174Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	.
121431783	c.524C>T	p.Ala175Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	.
121431793	c.534C>G	p.Asp178Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	.
121431801	c.542C>T	p.Ser181Phe	Uncertain significance	NA	Cardiovascular phenotype   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	.
121431804	c.545C>T	p.Ser182Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided	.
121431806	c.547T>C	p.Ser183Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	.
121431810	c.551C>T	p.Ser184Phe	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	.
121431813	c.554C>T	p.Ser185Leu	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided   Dilated cardiomyopathy 1HH   Primary familial hypertrophic cardiomyopathy   Myofibrillar myopathy, BAG3-related	6.47E-05
121431815	c.556G>C	p.Ala186Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	.
121431816	c.557C>T	p.Ala186Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	.
121431833	c.574G>A	p.Gly192Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not provided	0
121431833	c.574G>T	p.Gly192Cys	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related	.

121431837	c.578G>A	p.Arg193Lys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		3.23E-05
121431841	c.582C>G	p.Ser194Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121431859	c.600G>C	p.Gln200His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	IPV motif .	
121431864	c.605C>T	p.Pro202Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	IPV motif .	
121431866	c.607C>T	p.Arg203Trp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	IPV motif .	
121431867	c.608G>A	p.Arg203Gln	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	IPV motif	3.24E-05
121431867	c.608G>C	p.Arg203Pro	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	IPV motif	3.24E-05
121431869	c.610G>A	p.Gly204Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	IPV motif .	
121431875	c.616A>C	p.Ile206Leu	Conflicting interpretations of pathogenicity	NA	Premature ventricular contraction Cardiomyopathy Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	IPV motif	6.47E-05
121431881	c.622A>C	p.Ile208Leu	Uncertain significance	DM?	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	IPV motif .	
121431882	c.623T>C	p.Ile208Thr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	IPV motif .	
121431884	c.625C>T	p.Pro209Ser	Pathogenic/Likely pathogenic	DM	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Abnormality of the musculature not provided	IPV motif .	
121431884	c.625C>G	p.Pro209Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	IPV motif .	
121431885	c.626C>A	p.Pro209Gln	Pathogenic	DM	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	IPV motif .	
121431885	c.626C>T	p.Pro209Leu	Pathogenic	DM	Peripheral neuropathy Myofibrillar myopathy, BAG3-related Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	IPV motif .	
121431887	c.628G>T	p.Val210Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	IPV motif .	

121431893	c.634C>A	p.His212Asn	Uncertain significance	NA	Dilated cardiomyopathy 1HH	IPV motif	.	
121431896	c.637G>A	p.Glu213Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH	IPV motif	.	
121431904	c.645C>A	p.Asn215Lys	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		.	
121431905	c.646G>A	p.Val216Ile	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		.	3.23E-05
121431911	c.652C>T	p.Arg218Trp	Uncertain significance	DM	Dilated cardiomyopathy 1HH   Familial isolated arrhythmogenic right ventricular dysplasia   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related   not provided   Cardiovascular phenotype		.	
121431912	c.653G>A	p.Arg218Gln	Conflicting interpretations of pathogenicity	NA	Cardiovascular phenotype   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH   not specified   not provided   Left ventricular noncompaction Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		.	9.70E-05
121431914	c.655C>T	p.Pro219Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH		.	
121431917	c.658G>A	p.Ala220Thr	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		.	
121431927	c.668C>G	p.Pro223Arg	Uncertain significance	NA	Primary familial hypertrophic cardiomyopathy   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		.	
121431927	c.668C>T	p.Pro223Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		.	
121431932	c.673T>C	p.Phe225Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		.	
121431938	c.679C>G	p.Gln227Glu	Conflicting interpretations of pathogenicity	NA	Cardiovascular phenotype   not provided   Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		.	6.46E-05
121431951	c.692C>T	p.Thr231Met	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		.	
121431953	c.694C>G	p.His232Asp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		.	
121431954	c.695A>C	p.His232Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH   Myofibrillar myopathy, BAG3-related		.	
121431957	c.698A>G	p.Tyr233Cys	Uncertain significance	NA	not provided   Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		.	
121431963	c.704C>T	p.Ala235Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related   Dilated cardiomyopathy 1HH		.	



121431969	c.710A>G	p.Gln237Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121431970	c.711G>C	p.Gln237His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121431974	c.715G>A	p.Glu239Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	.	
121431996	c.737T>C	p.Val246Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121432002	c.743A>G	p.His248Arg	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	.	
121432020	c.761A>G	p.Asp254Gly	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121432029	c.770C>T	p.Pro257Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	3.23E-05
					not specified not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Myofibrillar		
121432031	c.772C>T	p.Arg258Trp	Benign/Likely benign	DM?	myopathy Cardiovascular phenotype		0.0007
121432032	c.773G>A	p.Arg258Gln	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121432035	c.776C>T	p.Pro259Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121432040	c.781C>T	p.Arg261Trp	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified	.	3.23E-05
121432041	c.782G>A	p.Arg261Gln	Conflicting interpretations of pathogenicity	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	3.23E-05
121432041	c.782G>C	p.Arg261Pro	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121432043	c.784G>A	p.Ala262Thr	Uncertain significance	DM	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified not provided	.	
121432044	c.785C>T	p.Ala262Val	Conflicting interpretations of pathogenicity	DM?	Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified not provided Dilated cardiomyopathy 1A	.	3.23E-05

121432047	c.788C>T	p.Ala263Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	.
121432053	c.794C>T	p.Pro265Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	.
121432060	c.801G>T	p.Arg267Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.
121432061	c.802T>C	p.Ser268Pro	Conflicting interpretations of pathogenicity	NA	not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.
121432074	c.815G>T	p.Gly272Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.
121432077	c.818C>T	p.Ala273Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.
121432080	c.821C>T	p.Ser274Leu	Conflicting interpretations of pathogenicity	NA	not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	0.0004
121432085	c.826C>T	p.Arg276Trp	Uncertain significance	NA	not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	.
121432086	c.827G>A	p.Arg276Gln	Conflicting interpretations of pathogenicity	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified	9.70E-05
121432089	c.830A>T	p.Glu277Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	3.24E-05
121432095	c.836C>T	p.Ser279Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.
121432097	c.838C>T	p.Pro280Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.
121432100	c.841G>A	p.Ala281Thr	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.
121432103	c.844A>G	p.Arg282Gly	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.
121432113	c.854C>T	p.Thr285Met	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	3.23E-05
121432124	c.865T>G	p.Ser289Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.
121432131	c.872C>T	p.Ser291Leu	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified	3.23E-05

121432134	c.875C>G	p.Pro292Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121432139	c.880C>T	p.Arg294Cys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121432140	c.881G>A	p.Arg294His	Uncertain significance	NA	not provided Primary dilated cardiomyopathy Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121432145	c.886C>T	p.His296Tyr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121432147	c.888C>A	p.His296Gln	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121432149	c.890C>T	p.Thr297Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121432151	c.892G>A	p.Val298Met	Uncertain significance	DM?	Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided		3.23E-05
121432157	c.898G>A	p.Asp300Asn	Benign/Likely benign	DM?	not specified Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Myofibrillar Myopathy, Dominant Dilated Cardiomyopathy, Dominant Cardiovascular phenotype		0.0003
121432164	c.905C>T	p.Pro302Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121435983	c.917T>C	p.Met306Thr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region .	
121435992	c.926G>A	p.Arg309Gln	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	6.47E-05
121436009	c.943T>A	p.Ser315Thr	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	PXXP region	9.70E-05
121436010	c.944C>T	p.Ser315Phe	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436013	c.947A>G	p.Gln316Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified	PXXP region .	
121436015	c.949C>T	p.Pro317Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436027	c.961C>T	p.Pro321Ser	Uncertain significance	DM?	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	6.46E-05
121436033	c.967A>G	p.Ser323Gly	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	

121436034	c.968G>A	p.Ser323Asn	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436039	c.973C>G	p.Pro325Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436046	c.980C>G	p.Pro327Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436049	c.983T>C	p.Val328Ala	Uncertain significance	NA	Cardiovascular phenotype Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region .	
121436054	c.988C>T	p.Pro330Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	3.23E-05
121436055	c.989C>T	p.Pro330Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436063	c.997C>T	p.Pro333Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	PXXP region	3.23E-05
121436078	c.1012C>T	p.Pro338Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region .	
121436093	c.1027C>T	p.Arg343Cys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	PXXP region	6.46E-05
121436094	c.1028G>A	p.Arg343His	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region .	
121436094	c.1028G>T	p.Arg343Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	PXXP region .	
121436098	c.1032A>T	p.Lys344Asn	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region .	
121436100	c.1034A>T	p.Glu345Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436101	c.1035G>C	p.Glu345Asp	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	PXXP region .	
121436121	c.1055C>G	p.Ser352Cys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region .	
121436124	c.1058A>G	p.Gln353Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	3.23E-05
121436129	c.1063C>T	p.Pro355Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region .	
121436130	c.1064C>T	p.Pro355Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	6.46E-05
121436144	c.1078G>C	p.Glu360Gln	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	

121436149	c.1083G>C	p.Lys361Asn	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436151	c.1085T>C	p.Val362Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	3.24E-05
121436154	c.1088A>C	p.Glu363Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	PXXP region .	
121436162	c.1096G>A	p.Val366Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436162	c.1096G>C	p.Val366Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	3.23E-05
121436166	c.1100C>G	p.Pro367Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	PXXP region	3.23E-05
121436171	c.1105G>C	p.Ala369Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436175	c.1109C>T	p.Pro370Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436184	c.1118G>A	p.Cys373Tyr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided Primary familial dilated cardiomyopathy not specified	PXXP region .	
121436184	c.1118G>C	p.Cys373Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436187	c.1121C>T	p.Pro374Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436193	c.1127C>A	p.Pro376His	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436199	c.1133C>G	p.Pro378Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region .	
121436204	c.1138C>T	p.Pro380Ser	Benign/Likely benign	DM	Myofibrillar myopathy, BAG3-related not provided not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Cardiovascular phenotype Atrial fibrillation	PXXP region	0.003
121436205	c.1139C>T	p.Pro380Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436207	c.1141T>C	p.Ser381Pro	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region .	
121436222	c.1156T>G	p.Ser386Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	

121436223	c.1157C>A	p.Ser386Tyr	Uncertain significance	NA	Cardiovascular phenotype Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	
121436225	c.1159C>T	p.Pro387Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	
121436226	c.1160C>T	p.Pro387Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	
121436229	c.1163A>G	p.Lys388Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	
121436232	c.1166G>A	p.Ser389Asn	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	0.0002
121436234	c.1168G>A	p.Val390Met	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	
121436235	c.1169T>C	p.Val390Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	
121436240	c.1174A>G	p.Thr392Ala	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	
121436250	c.1184G>A	p.Arg395Lys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	
121436253	c.1187C>T	p.Ala396Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	
121436265	c.1199C>T	p.Thr400Ile	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	
121436268	c.1202C>G	p.Ala401Gly	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	
121436268	c.1202C>T	p.Ala401Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	
121436276	c.1210G>A	p.Glu404Lys	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	
121436279	c.1213G>T	p.Ala405Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	
121436280	c.1214C>T	p.Ala405Val	Conflicting interpretations of pathogenicity	DM?	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	PXXP region	3.23E-05
121436283	c.1217C>T	p.Thr406Ile	Uncertain significance	NA	not specified Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region	6.46E-05
121436286	c.1220C>T	p.Pro407Leu	Benign	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified Dilated cardiomyopathy 1HH not provided Myofibrillar myopathy, BAG3-related Cardiovascular phenotype	PXXP region	0.1263

121436297	c.1231G>A	p.Gly411Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	PXXP region .	
121436306	c.1240G>A	p.Glu414Lys	Conflicting interpretations of pathogenicity	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		9.69E-05
121436307	c.1241A>G	p.Glu414Gly	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436308	c.1242G>C	p.Glu414Asp	Uncertain significance	NA	Dilated cardiomyopathy 1HH	.	
121436309	c.1243G>T	p.Ala415Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121436310	c.1244C>A	p.Ala415Asp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436315	c.1249C>T	p.Pro417Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121436316	c.1250C>A	p.Pro417Gln	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121436325	c.1259C>T	p.Pro420Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	BAG domain .	
121436328	c.1262G>A	p.Gly421Glu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Cardiovascular phenotype	BAG domain .	
121436330	c.1264G>A	p.Val422Met	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain	6.46E-05
121436352	c.1286T>C	p.Leu429Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain .	
121436354	c.1288G>A	p.Glu430Lys	Uncertain significance	NA	Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain .	
121436355	c.1289A>T	p.Glu430Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain .	
121436357	c.1291A>G	p.Lys431Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	BAG domain	6.46E-05
121436364	c.1298A>G	p.Gln433Arg	Uncertain significance	NA	Myofibrillar Myopathy, Dominant Dilated Cardiomyopathy, Dominant	BAG domain .	
121436373	c.1307A>G	p.Glu436Gly	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain .	

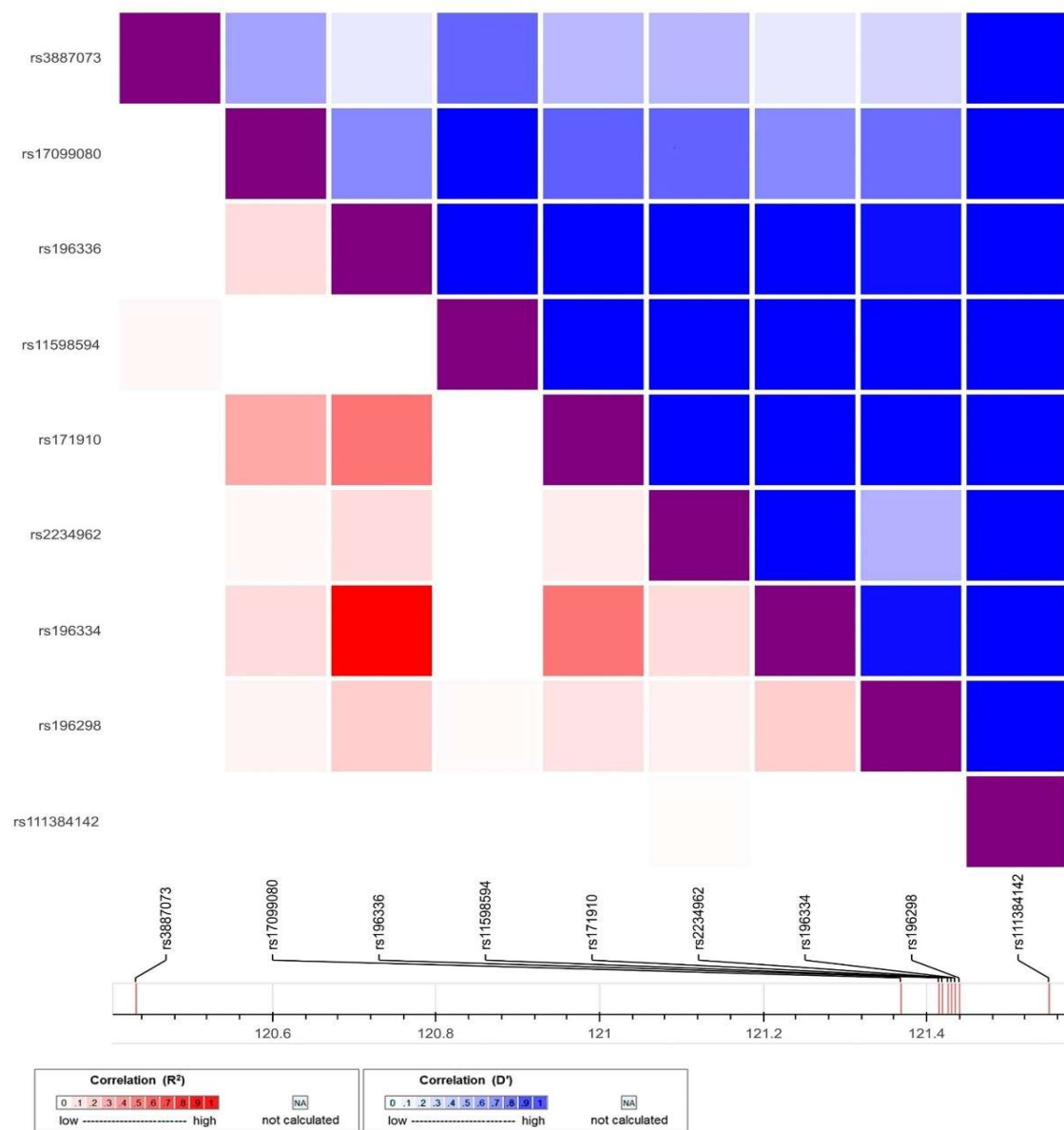
121436378	c.1312G>T	p.Ala438Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	BAG domain .	
121436384	c.1318G>A	p.Asp440Asn	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain	3.23E-05
121436387	c.1321A>G	p.Asn441Asp	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain	3.23E-05
121436389	c.1323C>A	p.Asn441Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain .	
121436389	c.1323C>G	p.Asn441Lys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain .	
121436428	c.1362C>G	p.Ile454Met	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain .	
121436429	c.1363G>A	p.Glu455Lys	Pathogenic	DM	not provided Primary dilated cardiomyopathy Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain .	
121436435	c.1369T>C	p.Tyr457His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain .	
121436451	c.1385T>C	p.Leu462Pro	Likely pathogenic	DM	Dilated cardiomyopathy 1HH not provided	BAG domain .	
121436460	c.1394T>C	p.Leu465Pro	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain .	
121436462	c.1396G>A	p.Asp466Asn	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain .	
121436473	c.1407C>A	p.Asp469Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain .	
121436474	c.1408C>T	p.Pro470Ser	Likely pathogenic	DM	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain .	
121436475	c.1409C>T	p.Pro470Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain .	
121436477	c.1411G>A	p.Glu471Lys	Uncertain significance	DM	Cardiovascular phenotype Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain .	
121436480	c.1414G>A	p.Gly472Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain .	
121436484	c.1418G>A	p.Arg473Gln	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain .	
121436486	c.1420G>A	p.Ala474Thr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain .	



121436489	c.1423G>A	p.Asp475Asn	Conflicting interpretations of pathogenicity	DM?	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not specified	BAG domain	9.68E-05
121436495	c.1429C>T	p.Arg477Cys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related	BAG domain	
121436496	c.1430G>A	p.Arg477His	Uncertain significance	DM	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain	
121436502	c.1436C>T	p.Ala479Val	Conflicting interpretations of pathogenicity	NA	not specified not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain	0.0009
121436513	c.1447G>A	p.Gly483Ser	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not specified	BAG domain	3.23E-05
121436517	c.1451T>A	p.Val484Asp	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain	
121436519	c.1453A>G	p.Arg485Gly	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain	
121436535	c.1469T>C	p.Ile490Thr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	BAG domain	
121436543	c.1477A>G	p.Lys493Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain	
121436562	c.1496T>C	p.Ile499Thr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	BAG domain	
121436589	c.1523A>G	p.Tyr508Cys	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Hypertrophic cardiomyopathy		
121436594	c.1528C>T	p.Leu510Phe	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436607	c.1541A>G	p.Asn514Ser	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436620	c.1554T>A	p.Asp518Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		3.23E-05
121436624	c.1558C>T	p.Pro520Ser	Uncertain significance	DM?	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH		
121436636	c.1570A>G	p.Ile524Val	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436636	c.1570A>T	p.Ile524Phe	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related		
121436637	c.1571T>C	p.Ile524Thr	Uncertain significance	NA	Myofibrillar Myopathy, Dominant Dilated Cardiomyopathy, Dominant		

121436640	c.1574T>C	p.Met525Thr	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121436644	c.1578G>T	p.Glu526Asp	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121436648	c.1582G>C	p.Gly528Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	3.23E-05
121436652	c.1586C>G	p.Ala529Gly	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	3.23E-05
121436652	c.1586C>T	p.Ala529Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436654	c.1588G>A	p.Val530Met	Conflicting interpretations of pathogenicity	NA	Dilated cardiomyopathy 1HH not provided Myofibrillar myopathy, BAG3-related not specified Cardiovascular phenotype Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	0.0003
121436664	c.1598A>C	p.Asp533Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436665	c.1599C>A	p.Asp533Glu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436667	c.1601A>G	p.Lys534Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436667	c.1601A>T	p.Lys534Met	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436689	c.1623T>A	p.Asn541Lys	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121436690	c.1624G>T	p.Ala542Ser	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121436691	c.1625C>T	p.Ala542Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	.	3.23E-05
121436696	c.1630G>A	p.Asp544Asn	Uncertain significance	NA	not provided Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436696	c.1630G>C	p.Asp544His	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	.	
121436700	c.1634C>G	p.Pro545Arg	Conflicting interpretations of pathogenicity	NA	Myofibrillar myopathy, BAG3-related not provided Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Primary dilated cardiomyopathy	.	6.46E-05
121436703	c.1637A>T	p.His546Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	

121436705	c.1639A>C	p.Thr547Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related not provided	.	
121436708	c.1642G>C	p.Glu548Gln	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	3.23E-05
121436715	c.1649A>G	p.Gln550Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436717	c.1651C>A	p.Gln551Lys	Uncertain significance	NA	not provided Dilated cardiomyopathy 1HH	.	3.23E-05
121436725	c.1659A>T	p.Glu553Asp	Likely benign	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH Cardiovascular phenotype	.	
121436730	c.1664C>A	p.Thr555Lys	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436733	c.1667C>A	p.Ala556Glu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH not provided	.	
121436733	c.1667C>T	p.Ala556Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436735	c.1669G>A	p.Ala557Thr	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436739	c.1673C>T	p.Ala558Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436751	c.1685C>T	p.Pro562Leu	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121436753	c.1687A>C	p.Ser563Arg	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121436759	c.1693A>G	p.Met565Val	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436768	c.1702A>C	p.Thr568Pro	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121436768	c.1702A>G	p.Thr568Ala	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436769	c.1703C>G	p.Thr568Ser	Uncertain significance	NA	Dilated cardiomyopathy 1HH Myofibrillar myopathy, BAG3-related	.	
121436786	c.1720G>T	p.Ala574Ser	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436790	c.1724C>G	p.Pro575Arg	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	
121436790	c.1724C>T	p.Pro575Leu	Uncertain significance	NA	Myofibrillar myopathy, BAG3-related Dilated cardiomyopathy 1HH	.	



**Figure S1.** Linkage disequilibrium (LD) of the eQTL SNPs with the nonsynonymous coding variant p.Cys151Arg (rs2234962).