

Myofibrillar Myopathy: A Very Quick Review of Consumer DNA Testing

Jason Filley (genealogy@mfm4.org)

ABSTRACT

Providing DNA samples to anyone is a highly personal decision that may have privacy, legal, and financial considerations. None of this is a recommendation of any sort. This is just quick observation and thought experiment. Myofibrillar Myopathy (“MFM”) is an extremely rare neuromuscular disease. Most MFMs are late-onset, beginning in the fifth or sixth decade, and are characterized by severe muscle weakness, frequently accompanied with muscle atrophy, cardiomyopathy, and respiratory problems. Consumer DNA testing from genealogy services (Ancestry.com, 23andMe, FamilyTreeDNA, and MyHeritage) covers a wide spectrum of known MFM variants. Results can be manually reviewed or automated via online service, with Promethease being the most popular.

KEYWORDS Myofibrillar Myopathy; MFM

This paper covers Myofibrillar Myopathy only, though the same steps should be able to be replicated for other neuromuscular diseases (e.g., limb-girdle muscular dystrophy).

This is terse, but the primary impetus of this write-up is to point out things that may have escaped the notice of those already savvy or expert in DNA analysis.

Item: DNA testing companies already have databases of a subset of presumptively-positive MFM patients, and around 80% of users give consent for results to be used in health research. Researchers might partner and follow up with potential patients for higher quality DNA test verification and possible clinical referral.

Item: Since determining genetic distance between users is the *raison d'être* of genealogy services, it is trivial for them to determine relationship clusters per variant. Family histories taken at the doctor’s office are unlikely to include much beyond first cousins; genealogy DNA tests are reliable until about 5th cousins. It is not unheard of that “different families” have the same variant but are actually the same family inheriting the disease from a common founder. The odds of inherited vs. *de novo* mutations vary by gene. For

example, LDB3 mutations A147T and A165V tend to have long family histories, while DES is incredibly prone to mutation and occurs frequently in patients with little or no family history.

Item: Can this existing data be used to help affected people, while still respecting privacy, legal, and financial concerns (e.g., pre-existing condition impacts on life/health/nursing home/disability insurance)?

Item: Analysis services like Promethease need more rigorous quality control. Obvious false positives exist and needlessly scare people.

Item: Analysis services like Promethease and data services like SNPedia lack accurate and actionable information for the MFM variants. There is no ‘next action.’ The least-worst option at present would be to add an entry of “Join the ‘Myofibrillar Myopathy’ Facebook group and inquire there.”

Item: Whole Genome Sequencing is not covered here, but Nebula Genomics is currently offering WGS for US\$300.

Consumer DNA Test Coverage of MFM

Consumer DNA tests from the four most popular genealogy services (Ancestry.com, MyHeritage, 23andMe, and FamilyTreeDNA) test for a number of MFM variants, with Ancestry.com providing the broadest coverage.

Some of the most well-known variants are tested, including: LDB3^{A147T}, LDB3^{A165V}, LDB3^{R268C}, BAG3^{P209L}, CRYAB^{R120G}, FLNC^{W2710X}, MYOT^{S60C}, MYOT^{T57I}, and DES^{L345P}.

A ClinVar download of search for 'myofibrillar myopathy' results in 4426 total entries. (date retrieved: 11/12/2020) A few of the results are questionable, but negligible overall, so these results don't include any curating or pruning.

<https://www.ncbi.nlm.nih.gov/clinvar/?term=myofibrillar+myopathy>

Of those **4426** variants:

- **1144** variants are covered by at least one test. **888** of those are for TTN alone, leaving **256** covering other genes.
- **691** have a "Clinical significance" of "Pathogenic," "Likely Pathogenic" or "Conflicting interpretations of pathogenicity." "Conflicting interpretations of pathogenicity" includes "conflicts" where a mutation is pathogenic for MFM but is uncertain for cardiomyopathy or other condition.
- **386** have a "Clinical significance" of "Benign" or "Likely Benign."
- **67** have a "Clinical significance" of "Uncertain Significance."

Of the **1144** MFM variants – of any "Clinical significance" – tested by at least one of the four companies:

- Ancestry.com covers **1070** variants and covers **553** unique variants (variants that aren't covered by any of the other three companies).
- MyHeritage covers **496** variants but only covers **9** unique variants.
- FamilyTreeDNA covers **267** variants but only covers **4** unique variants.
- 23andMe covers **262** variants but only covers **2** unique variants.

Of the Pathogenic/Likely/Conflicting, **554** are for TTN, leaving **137** non-TTN.

The table at the end of this paper contains the list of consumer-DNA-tested variants considered to be 'Pathogenic,' 'Likely Pathogenic,' or 'Conflicting Interpretations of Pathogenicity.' Those considered 'Benign,' 'Likely Benign,' or 'Variant of Unknown Significance ('VOUS')' are not included.

DNA Test Result Processing

Consumer DNA tests are not performed to higher standards as one would get with a professional test as from, say, Invitae. At best, genealogy DNA tests provide a presumptive indicator of a subset of known variants; they're not comprehensive or 100% reliable. All consumer DNA providers expressly warn against using their results to make medical decisions.

Someone who has already taken a genealogy DNA test has nothing to lose by reviewing their existing results, though this is complicated and requires time and critical thinking. A lot of doctors will not even refer or review DNA tests with a patient and will instead refer the patient to a genetic counselor.

Someone with a family history of a known variant tested by consumer DNA could get the relevant DNA test, which doesn't require a doctor's order or referral. Whether consumer DNA tests are or will be linked to medical history and affect insurance is something each person should seriously contemplate before taking a DNA test or uploading existing results to an analysis service like Promethease.

Download Raw Data

To download the raw result data from a genealogy DNA test provider, consult their online help or perform an Internet search for instructions (e.g., "23andme raw data"). Results are a plain or compressed text file that is easily opened and searched by a text editor (e.g., Notepad).

```
rs398124238 5 137206447 I I
rs121908461 5 137206456 C C
rs121908457 5 137206504 C C
rs28937597 5 137206510 C C
rs121908458 5 137206519 C C
rs121908460 5 137206624 G G
rs7713839 5 137210651 T C
rs199789331 5 137213248 G G
rs151094883 5 137213294 G G
```

Figure 1 Line item for pathogenic MYOT^{Ser60Cys}, shown negative here, from AncestryDNA. A recent paper chronicled the diagnostic steps, including an EMG and muscle biopsy, for a patient with this mutation. AncestryDNA would have shown this for under \$100.

QA and Due Diligence Required

Reported variants aren't 100% reliable. It seems that some rsids are used by the genealogy DNA companies for other non-canonical use. Analysis services – Promethease is a popular one – don't always catch these mistakes.

Table 1 Sample mutations flagged by Promethease The CLCN1 mutation was correctly found and matches findings from a Lantern Project/PerkinElmer test. Other results flagged by Promethease are easily proven to be false positives: the homozygous indels are *prima facie* incorrect, due to non-matching phenotype (*three* homozygous BAG3 mutations?), and a quick check of the same rsids on OpenSNP show that the same indels are reported for almost all users. 99% of OpenSNP contributors report the same mutation but cannot all *all* have homozygous BAG3 mutations.

rsid	chr	position	allele1	allele2	variant	company
rs55960271	7	143048771	T	C	(CLCN1:R894X)	Ancestry.com
rs794728981	10	121436333	I	I	(BAG3:Leu423fs)	Ancestry.com + 23andMe
rs727505283	10	121411287	I	I	(BAG3:Thr34AlafsX21)	23andMe
rs794728981	10	121436333	I	I	(BAG3:Leu423fs)	23andMe
rs797044606	2	152352797	D	D	(NEB:Ser8196delinsTyrTer)	23andMe
rs121913648	14	23885315	I	I	(MYH7:Lys1617del)	23andMe
rs587780494	2	179639882	D	D	(TTN:Lys2186fs)	23andMe

rs60538473(I;I)

[more info](#)

1	Magnitude
(I;I)	23andme_genome_
Pathogenic	ClinVar Significance
DES	Genes
2	Chromosome
219418979	Position
6.5	Max Magnitude
2017-09-03	Rs Modified
plus	Stabilized
plus	Orientation

ClinVar Pathogenic not provided Myofibrillar myopathy 1

Figure 2 Pathogenic Desminopathy flagged by Promethease for a 23andMe upload, showing homozygous insertions in DES. A user not acquainted with MFM or having any reason to believe she has neuromuscular problems could easily be pointlessly alarmed by this. More than one has shown up concerned on the “Myofibrillar Myopathy” Facebook group for this exact reason.

opensnp.org/snps/rs60538473

SNPedia (0) PLoS (0) Mendeley (0) Personal Genome Project (0) Genome.gov (0) Other users (1) Comments (0)

Users who share this SNP:

User	Genotype
[User]	II
[User]	II
[User]	II
[User]	II
[User]	II
[User]	II
[User]	II

99% of user reports for rs60538473 on openSNP show homozygous insertions for this pathogenic DES mutation. This simply cannot be correct. This is an obvious false positive from 23andMe.

Figure 3 OpenSNP shows 99% of the listed 699 users with entries for rs60538473 as having the same homozygous insertion, which is impossible. This is a false positive. Similar items where the SNP shows a significant percentage of users with homozygous indels should be reviewed and weeded out.

Listing 1 Sample QA using R on OpenSNP data for rs121908333 (LDB3:A147T), rs121908334 (LDB3:A165V), and rs121908335 (LDB3:R268C). Open the resulting 'ldb3.csv' and filter in Excel or other tool.

```
library("rsnps")
setwd("C:/Users/You/opensnp")

rsids <- c('rs121908333', 'rs121908334', 'rs121908335')

for (rsid in rsids){
  print(rsid)
  x <- allgensnp(snp = rsid)
  write.table(x, "ldb3.csv", sep = ",", col.names = !file.exists("ldb3.csv"), append = T)
}
```

Listing 2 Data Collation to tab-delimited 'mfm_results.txt'

```
#Using these personal result files:
17.5M AncestryDNA.txt
19.4M MyHeritage_raw_dna_data.csv
15.3M familytreedna_37_Chrom_Autoso.csv
16.0M 23andme_genome.txt
 1.8M clinvar_result.txt (https://www.ncbi.nlm.nih.gov/clinvar/?term=myofibrillar+myopathy)

# remove the spurious trailing <TAB> on the first line of clinvar_result.txt
# Clean up raw results files from Ancestry.com, 23andMe, MyHeritage, and FamilyTreeDNA
# Remove commented lines and personal results

# ancestry
egrep -v "^#" AncestryDNA.txt | cut -f 1-3 >ancestry_dna.txt

# familytreedna
cut -d, -f 1-3 familytreedna_37_Chrom_Autoso.csv >familytreedna_dna.txt

# 23andme
echo -e "rsid\tchromosome\tposition" >23andme_dna.txt
egrep -v "^#" 23andme_genome.txt | cut -f 1-3 >>23andme_dna.txt

# myheritage
egrep -v "^#" MyHeritage_raw_dna_data.csv | tr -d "\"" | cut -d, -f 1-3 >myheritage_dna.txt

# Use sqlite to import text files and collate. Could be optimized, but this is sufficient for a POC
sqlite3 clinvarmfm
sqlite>
.mode tabs
.import clinvar_result.txt clinvarmfm
.import ancestry_dna.txt ancestrydna
.import 23andme_dna.txt twentythreeandme
.mode csv
.import familytreedna_dna.txt familytreedna
.import myheritage_dna.txt myheritage

CREATE VIEW allmfm AS
SELECT *,
  (SELECT COUNT(ancestrydna.rsid) FROM ancestrydna
   WHERE ancestrydna.rsid = clinvarmfm.'dbSNP ID') AS ancestrycom,
  (SELECT COUNT(myheritage.RSID) FROM myheritage
   WHERE myheritage.RSID = clinvarmfm.'dbSNP ID') AS mh,
  (SELECT COUNT(familytreedna.RSID) FROM familytreedna
   WHERE familytreedna.RSID = clinvarmfm.'dbSNP ID') AS ftdna,
  (SELECT COUNT(twentythreeandme.rsid) FROM twentythreeandme
   WHERE twentythreeandme.rsid = clinvarmfm.'dbSNP ID') AS twentythree
FROM clinvarmfm;

.headers on
.mode tabs
.output mfm_results.txt

SELECT *, (ancestrycom+mh+ftdna+twentythree) AS total FROM allmfm;

.quit
```

Table 2 Table of MFM variants tested by genealogy companies, using 2020 tests (“chips”). A few of the results are questionable, but negligible overall, so these results don’t include any curating or pruning. Includes only ‘Pathogenic,’ ‘Likely Pathogenic,’ and ‘Conflicting Interpretations of Pathogenicity.’ ‘Benign,’ ‘Likely Benign,’ and VOUS are not included. Table is sorted by ‘gene’ then ‘rsid.’ AD=AncestryDNA; MH=MyHeritage; FT=FamilyTreeDNA; 23=23andMe

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
BAG3	rs112929734		Cardiovascular phenotype; not specified; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related	Y	-	-	-
BAG3	rs117749531	E414*	Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related	Y	Y	-	-
BAG3	rs117749531	E414K	not specified; Dilated cardiomyopathy 1HH; not provided; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related	Y	Y	-	-
BAG3	rs121918312	P209Q	Myofibrillar myopathy, BAG3-related	Y	-	Y	Y
BAG3	rs121918312	P209L	Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not provided; Charcot-Marie-Tooth disease	Y	-	Y	Y
BAG3	rs138832242	D475N	Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not specified	-	-	Y	Y
BAG3	rs139399890		Dilated cardiomyopathy 1HH; not specified; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; Cardiovascular phenotype; Myofibrillar myopathy, BAG3-related	Y	-	-	-
BAG3	rs141355480	P77L	not specified; Cardiomyopathy; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not provided	Y	Y	-	-
BAG3	rs143919208	S274L	not provided; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not specified	Y	-	-	-
BAG3	rs144678100	V530M	not specified; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH; Dilated cardiomyopathy 1HH; Cardiovascular phenotype; Myofibrillar myopathy, BAG3-related	Y	Y	-	-
BAG3	rs144814361		Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related	-	-	-	Y
BAG3	rs145393807	I94F	not provided; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not specified; Cardiovascular phenotype; Primary dilated cardiomyopathy; Myofibrillar myopathy, BAG3-related	Y	Y	-	-
BAG3	rs147259596		Dilated cardiomyopathy 1HH; not specified; Myofibrillar myopathy, BAG3-related; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH	Y	Y	-	-
BAG3	rs149358702		Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not specified; Cardiovascular phenotype; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related	Y	-	-	-
BAG3	rs151331972	H83Q	not specified; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy; not provided	Y	-	-	-
BAG3	rs200212999		Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH; not provided; not specified	Y	-	-	-
BAG3	rs201638005	R218Q	Left ventricular noncompaction; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH; not specified; Cardiovascular phenotype; not provided	Y	-	-	-
BAG3	rs34656239	A479V	Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not specified; not provided; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related	Y	-	Y	Y
BAG3	rs35434411	R71L	Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not specified	Y	Y	Y	Y
BAG3	rs375257731	S134T	Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH; not specified; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH	-	Y	-	-
BAG3	rs376198104	V154M	not specified; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH	Y	-	-	-
BAG3	rs387906874	R71W	Dilated cardiomyopathy 1HH; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy	Y	Y	-	Y
BAG3	rs387906875	R123*	Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not provided; Primary dilated cardiomyopathy; Dilated cardiomyopathy 1HH; Cardiovascular phenotype	Y	-	Y	Y
BAG3	rs397516881	E455K	Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not provided; Primary dilated cardiomyopathy	Y	-	-	-
BAG3	rs397516882		Myofibrillar myopathy, BAG3-related; not specified; Dilated cardiomyopathy 1HH	Y	-	-	-
BAG3	rs397516883	A262V	Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; Cardiovascular phenotype; not specified; not provided; Dilated cardiomyopathy 1A	Y	-	-	-
BAG3	rs556465096	R139W	not specified; Myofibrillar myopathy, BAG3-related; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH; Dilated cardiomyopathy 1HH	Y	Y	-	-
BAG3	rs572038196	A156G	not specified; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related	Y	-	-	-
BAG3	rs727505109	P356fs	Primary dilated cardiomyopathy; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH	Y	-	-	-
BAG3	rs730880054	S185L	not provided; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH; Primary familial hypertrophic cardiomyopathy; Myofibrillar myopathy, BAG3-related; Dilated cardiomyopathy 1HH	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
BAG3	rs775151738		Dilated cardiomyopathy 1HH; Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not provided; Myofibrillar myopathy, BAG3-related; not specified	Y	-	-	-
BAG3	rs869248137	R309*	Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; Familial dilated cardiomyopathy; Primary dilated cardiomyopathy; not provided	Y	-	-	-
BAG3	rs876657634	Q244*	Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; Primary dilated cardiomyopathy	Y	-	-	-
BAG3	rs876661342	Y233*	Dilated cardiomyopathy 1HH; Myofibrillar myopathy, BAG3-related; not provided	Y	-	-	-
CD46	rs35366573	A353V, A338V, A324V	A323V, A309V, Myofibrillar myopathy, BAG3-related; Atypical hemolytic-uremic syndrome 2	Y	Y	Y	Y
CRYAB	rs104894201	R120G, R53G	Alpha-B crystallinopathy	Y	-	-	-
CRYAB	rs104894202	Q151*, Q84*	Alpha-B crystallinopathy	Y	-	-	-
CRYAB	rs149787233	P39L	Dilated cardiomyopathy 1II; Fatal infantile hypertonic myofibrillar myopathy; not specified; Cardiovascular phenotype; Cataract 16, multiple types; Alpha-B crystallinopathy	Y	-	-	-
CRYAB	rs150516929	G154S, G87S	Hypertrophic cardiomyopathy; Congestive heart failure; Myofibrillar Myopathy, Dominant; Cardiovascular phenotype; not specified; Dilated cardiomyopathy 1II; Cardiomyopathy; Primary familial hypertrophic cardiomyopathy; Cataract 16, multiple types; Congenital cataract; Fatal infantile hypertonic myofibrillar myopathy; not provided	Y	Y	Y	Y
CRYAB	rs2234704	P51L	Cardiovascular phenotype; Cardiomyopathy; Fatal infantile hypertonic myofibrillar myopathy; not specified; not provided; Dilated cardiomyopathy 1II; Cataract 16, multiple types; Alpha-B crystallinopathy	Y	Y	-	-
CRYAB	rs281865142	S48fs, S115fs	Alpha-B crystallinopathy; Dilated cardiomyopathy 1II; not provided	-	-	Y	Y
CRYAB	rs387907339	D109H, D42H	Alpha-B crystallinopathy	Y	-	-	-
DES	rs121913000	A360P	Myofibrillar myopathy 1	Y	-	-	-
DES	rs121913001	N393I	not provided; Myofibrillar myopathy 1	Y	-	Y	-
DES	rs121913002		Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided; Cardiovascular phenotype; not specified	-	Y	Y	-
DES	rs121913002	I451M	Neurogenic scapuloperoneal syndrome, Kaeser type; Dilated cardiomyopathy 1I; Myofibrillar myopathy 1; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided	-	Y	Y	-
DES	rs121913003	R406W	Myofibrillar myopathy 1; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided	Y	-	Y	-
DES	rs121913004	Q389P	Myofibrillar myopathy 1; not provided	Y	-	Y	-
DES	rs121913005	T442I	Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; Myofibrillar myopathy 1; not provided	Y	-	Y	-
DES	rs144261171	R212Q	not provided; Myofibrillar myopathy 1; Dilated cardiomyopathy 1I; Neurogenic scapuloperoneal syndrome, Kaeser type; not specified; Left ventricular noncompaction cardiomyopathy; Myofibrillar Myopathy, Dominant; Myofibrillar myopathy 1; Muscular dystrophy, limb-girdle, type 2R	Y	Y	Y	Y
DES	rs147327878	E262V	Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided; not specified	Y	Y	Y	Y
DES	rs148947510	D312A	Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not specified; Cardiovascular phenotype; not provided	Y	-	-	-
DES	rs150974575	R429*	Myofibrillar myopathy 1; Neuromuscular disease; Primary dilated cardiomyopathy; Myofibrillar myopathy 1; Muscular dystrophy, limb-girdle, type 2R; not provided	Y	-	Y	Y
DES	rs199972656		Myofibrillar Myopathy, Dominant; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided; Cardiomyopathy; Neurogenic scapuloperoneal syndrome, Kaeser type; Dilated cardiomyopathy 1I; Myofibrillar myopathy 1; not specified	Y	-	-	-
DES	rs200545412	G84S	Myofibrillar myopathy 1; Muscular dystrophy, limb-girdle, type 2R; not provided; not specified	Y	-	-	-
DES	rs267607482	N342D	Myofibrillar myopathy 1; not provided	Y	-	Y	Y
DES	rs267607483		not provided; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; Primary dilated cardiomyopathy; Myofibrillar myopathy 1	-	-	Y	-
DES	rs267607485	K449T	not provided; Myofibrillar myopathy	Y	-	-	-
DES	rs267607490	R454W	Primary dilated cardiomyopathy; Primary dilated cardiomyopathy; Neuromuscular disease; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided; Myofibrillar myopathy 1; Primary familial hypertrophic cardiomyopathy	Y	-	-	-
DES	rs267607495	S12F	Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; Primary dilated cardiomyopathy; Neuromuscular disease; not provided	Y	-	Y	Y
DES	rs267607499	N116S	Myofibrillar myopathy 1; not provided	Y	-	Y	Y
DES	rs34337334	D312N	not specified; not provided; Cardiomyopathy; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; Neurogenic scapuloperoneal syndrome, Kaeser type; Dilated cardiomyopathy 1I; Myofibrillar myopathy 1; Cardiovascular phenotype	Y	-	-	Y

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
DES	rs367961979	R222H	Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; Myofibrillar Myopathy, Dominant; not specified; Myofibrillar myopathy 1; Dilated cardiomyopathy 1I; Neurogenic scapuloperoneal syndrome, Kaeser type	-	Y	-	-
DES	rs370239228		not specified; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided; Neurogenic scapuloperoneal syndrome, Kaeser type; Dilated cardiomyopathy 1I; Myofibrillar myopathy 1	Y	-	-	-
DES	rs372825868	S57L	Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided; not specified; Neurogenic scapuloperoneal syndrome, Kaeser type; Dilated cardiomyopathy 1I; Myofibrillar myopathy 1; Cardiovascular phenotype	Y	-	-	-
DES	rs397516691		not provided; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not specified	Y	-	-	-
DES	rs397516694	R127P	not specified; Arrhythmogenic right ventricular cardiomyopathy; not provided; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1	Y	-	Y	Y
DES	rs397516698		Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1	Y	-	Y	-
DES	rs397516698		Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; Myofibrillar myopathy 1; not provided	Y	-	Y	-
DES	rs397516698		Myofibrillar myopathy 1	Y	-	Y	-
DES	rs398122940		Myofibrillar myopathy 1	Y	-	-	-
DES	rs41272699	A213V	Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; Myofibrillar myopathy; Neurogenic scapuloperoneal syndrome, Kaeser type; Myofibrillar myopathy 1; Dilated cardiomyopathy 1I; Myofibrillar Myopathy, Dominant; not specified; Cardiovascular phenotype; Congenital diaphragmatic hernia; Cardiomyopathy; not provided	Y	Y	Y	-
DES	rs57639980	L345P	Myofibrillar myopathy 1; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided	Y	-	Y	Y
DES	rs57694264	E401K	Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided	-	-	Y	Y
DES	rs578191306		Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not specified; Myofibrillar Myopathy, Dominant; not provided; Myofibrillar myopathy 1; Dilated cardiomyopathy 1I; Neurogenic scapuloperoneal syndrome, Kaeser type	Y	-	-	-
DES	rs57955682	L385P	not provided; Myofibrillar myopathy 1	Y	-	Y	Y
DES	rs57965306	R350P	Neurogenic scapuloperoneal syndrome, Kaeser type; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided	Y	-	Y	Y
DES	rs58687088	N366del	not provided; Myofibrillar myopathy 1	Y	-	Y	-
DES	rs58898021	A357P	not provided; Myofibrillar myopathy	Y	-	-	-
DES	rs59962885	A337P	not provided; Myofibrillar myopathy 1; Dilated cardiomyopathy 1I	-	-	Y	-
DES	rs60538473		not provided; Myofibrillar myopathy 1	-	-	Y	Y
DES	rs60798368	R16C	Myofibrillar myopathy; not provided	Y	-	-	-
DES	rs61130669	D399Y	Myofibrillar myopathy 1; not provided	Y	-	-	-
DES	rs61726467	E413*	Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1	Y	-	-	-
DES	rs61726467	E413K	Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided	Y	-	-	-
DES	rs62635763	P419S	Myofibrillar myopathy 1; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided	Y	-	Y	-
DES	rs62636491	S298L	Cardiomyopathy; Myofibrillar myopathy 1; Dilated cardiomyopathy 1I; Neurogenic scapuloperoneal syndrome, Kaeser type; not provided; Dilated cardiomyopathy; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1	Y	-	-	Y
DES	rs62636495	S13F	not provided; Primary dilated cardiomyopathy; Myofibrillar myopathy 1	-	-	Y	Y
DES	rs748323823		Cardiovascular phenotype; not provided; Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not specified	Y	-	Y	-
DES	rs794728992	Q99E	Muscular dystrophy, limb-girdle, type 2R; Myofibrillar myopathy 1; not provided	Y	-	Y	Y
DNAJB6	rs142974468	S321L, S206L	not specified; not provided; Limb-Girdle Muscular Dystrophy, Dominant; Myofibrillar Myopathy, Dominant; Limb-girdle muscular dystrophy, type 1E	Y	Y	-	-
EPHB1, CEP63, KY	rs377332009	Y135*, Y114*, Y119*, Y93*	Myopathy, myofibrillar, 7	Y	-	-	-
FLNC	rs149641783	R1341Q	Myopathy, distal, 4; Dilated Cardiomyopathy, Dominant; Cardiomyopathy, familial hypertrophic, 26; Myofibrillar myopathy, filamin C-related; not provided; not specified	Y	Y	-	-
FLNC	rs182845462	V523A	not provided; Cardiomyopathy, familial hypertrophic, 26; Dilated Cardiomyopathy, Dominant; Myofibrillar myopathy, filamin C-related; Myopathy, distal, 4	-	Y	Y	Y
FLNC	rs189525930	G507R	not provided; Cardiomyopathy, familial hypertrophic, 26; Myopathy, distal, 4; Myofibrillar myopathy, filamin C-related; Dilated Cardiomyopathy, Dominant	-	Y	Y	Y
FLNC	rs200215903	G896R	not provided; Myofibrillar myopathy, filamin C-related; Cardiomyopathy, familial hypertrophic, 26; Myopathy, distal, 4; Dilated Cardiomyopathy, Dominant	-	Y	Y	Y
FLNC	rs387906586	M251T	Myopathy, distal, 4	Y	-	-	-
FLNC	rs387906587	A193T	Myopathy, distal, 4; not provided; not specified	Y	-	-	-
FLNC	rs770606675	R650*	not provided; Myofibrillar myopathy, filamin C-related; Cardiomyopathy, familial hypertrophic, 26; Myopathy, distal, 4; Dilated Cardiomyopathy, Dominant	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23	
FLNC	rs781135153		not provided; Myofibrillar myopathy, filamin C-related; Cardiomyopathy, familial hypertrophic, 26; Myopathy, distal, 4; Dilated Cardiomyopathy, Dominant	Y	-	-	-	
FLNC, FLNC-AS1	rs121909518	W2710*, W2677*	Myofibrillar myopathy, filamin C-related; Dilated Cardiomyopathy, Dominant; Cardiomyopathy, familial hypertrophic, 26; Myopathy, distal, 4; Myofibrillar myopathy, filamin C-related; not provided	Y	-	-	-	
FLNC, FLNC-AS1	rs184018403	I1882V, I1849V	not provided; not specified; Myopathy, distal, 4; Cardiomyopathy, familial hypertrophic, 26; Myofibrillar myopathy, filamin C-related; Dilated Cardiomyopathy, Dominant	Y	Y	-	-	
GYG1	rs370652040		Polyglucosan body myopathy 2; Glycogen storage disease XV; Polyglucosan body myopathy 2; not provided; Glycogen storage disease XV	Y	Y	-	-	
GYG1	rs727502871	D163fs	Polyglucosan body myopathy 2; Glycogen storage disease XV; not provided	Y	-	-	-	
LDB3	rs138251566	T351A, T304A	Cardiovascular phenotype; Myofibrillar myopathy, ZASP-related; not provided; Hypertrophic cardiomyopathy; not specified; Dilated cardiomyopathy 1A; Cardiomyopathy; Primary dilated cardiomyopathy	Y	-	-	-	
LDB3	rs139922045	A222T	Dilated cardiomyopathy 1C; Myofibrillar myopathy, ZASP-related; not specified; Cardiovascular phenotype; not provided; Myofibrillar myopathy, ZASP-related	Y	-	-	-	
LDB3	rs140552419		not specified; Cardiovascular phenotype; Myofibrillar myopathy, ZASP-related; not provided	Y	Y	-	-	
LDB3	rs143764931	A532P, A485P, A469P	A537P, A422P	Myofibrillar myopathy, ZASP-related; Cardiomyopathy; not provided; not specified	-	Y	Y	Y
LDB3	rs144445130		Myofibrillar myopathy, ZASP-related; not provided; Cardiovascular phenotype; not specified	Y	-	-	-	
LDB3	rs200796750	T350I, T303I	Hypertrophic cardiomyopathy; Myofibrillar myopathy, ZASP-related; not specified	Y	Y	-	-	
LDB3	rs372331627	I558V, I448V, I511V, I495V, I563V	Arrhythmogenic right ventricular cardiomyopathy; Cardiomyopathy; Myofibrillar myopathy, ZASP-related; not specified; not provided	Y	-	-	-	
LDB3	rs375306400	S223L	not specified; not provided; Myofibrillar myopathy, ZASP-related	Y	-	-	-	
LDB3	rs45487699	S189L	not specified; Dilated cardiomyopathy 1A; Familial hypertrophic cardiomyopathy 24; Cardiovascular phenotype; not provided; Myofibrillar myopathy, ZASP-related; Dilated cardiomyopathy 1C	Y	Y	Y	Y	
LDB3	rs45521338	R218C, R333C	R265C	not specified; not provided; Myofibrillar myopathy, ZASP-related	Y	-	Y	Y
LDB3	rs45529531		not specified; Myofibrillar Myopathy, Dominant; Cardiomyopathy; Left ventricular noncompaction cardiomyopathy; Dilated Cardiomyopathy, Dominant; Cardiovascular phenotype; Myofibrillar myopathy, ZASP-related	Y	Y	-	-	
LDB3	rs45539535	A371T, A308T, A376T	A261T, A324T	not specified; Cardiovascular phenotype; not provided; Myofibrillar myopathy, ZASP-related; Cardiomyopathy	Y	-	-	-
LDB3	rs775180716	P243L, P358L	P290L	Myofibrillar myopathy	Y	-	-	-
LDB3, LOC110121486	rs121908333	A147T, A262T	Cardiomyopathy; Myofibrillar myopathy, ZASP-related; not provided	Y	-	-	-	
LDB3, LOC110121486	rs121908334	A165V, A280V	Myofibrillar myopathy; Myofibrillar myopathy, ZASP-related; Neuromuscular disease; not provided; Cardiomyopathy	Y	-	Y	Y	
LDB3, LOC110121486	rs121908338	D117N, D232N	not provided; Cardiovascular phenotype; Primary dilated cardiomyopathy; Dilated cardiomyopathy 1C; Myofibrillar myopathy, ZASP-related; Dilated cardiomyopathy; not specified	Y	Y	Y	Y	
LDB3, LOC110121486	rs371708921		Myofibrillar Myopathy, Dominant; Dilated Cardiomyopathy, Dominant; not specified; Left ventricular noncompaction cardiomyopathy; Myofibrillar myopathy, ZASP-related	Y	Y	-	-	
LDB3, LOC110121486	rs371708921		Myofibrillar myopathy, ZASP-related; Left ventricular noncompaction cardiomyopathy; Myofibrillar Myopathy, Dominant; Dilated Cardiomyopathy, Dominant; not specified	Y	Y	-	-	
LDB3, LOC110121486	rs373632943	A171T, A286T	Cardiovascular phenotype; Myofibrillar myopathy, ZASP-related; not specified	Y	-	-	-	
LDB3, LOC110121486	rs71473272		Myofibrillar Myopathy, Dominant; Dilated Cardiomyopathy, Dominant; Left ventricular noncompaction cardiomyopathy; Cardiovascular phenotype; Myofibrillar myopathy, ZASP-related; not specified	Y	Y	-	-	
LDB3, LOC110121486	rs754704023		Myofibrillar Myopathy, Dominant; Dilated Cardiomyopathy, Dominant; Left ventricular noncompaction cardiomyopathy; not specified; Myofibrillar myopathy, ZASP-related	Y	-	-	-	
MYOT	rs121908457	S55F	Myofibrillar myopathy 3; not provided; Progressive distal muscle weakness; Progressive proximal muscle weakness; Fatty replacement of skeletal muscle; Lower limb pain; Foot dorsiflexor weakness; Distal lower limb muscle weakness; Muscle fiber inclusion bodies; Distal amyotrophy; EMG: myopathic abnormalities; Urinary bladder sphincter dysfunction	Y	-	-	-	
MYOT	rs121908458	S60F	Myofibrillar myopathy 3; not provided	Y	-	Y	-	
MYOT	rs121908458	S60C	not provided; Myofibrillar myopathy 3; Myofibrillar myopathy	Y	-	Y	-	
MYOT	rs121908460	S95I	Myofibrillar myopathy 3	Y	-	-	-	

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
MYOT	rs140678912		not provided; not specified; Myofibrillar myopathy 3	Y	-	-	-
MYOT	rs145427063		Myofibrillar myopathy 3; not specified; not provided	Y	-	-	-
MYOT	rs150293853	R178H, R63H	Myofibrillar myopathy 3; Spheroid body myopathy; Limb-Girdle Muscular Dystrophy, Dominant; Myofibrillar Myopathy, Dominant; not specified	-	-	Y	Y
MYOT	rs28937597	T57I	Myofibrillar myopathy 3; not provided	Y	-	-	-
MYOT	rs78633961	D286E, D217E	Spheroid body myopathy; Limb-Girdle Muscular Dystrophy, Dominant; Myofibrillar Myopathy, Dominant; not provided	Y	-	-	-
PYROXD1	rs781565158	N155S, N84S	Myopathy, myofibrillar, 8	Y	-	-	-
RYR1	rs118192140	T4709M, T4704M	Central core myopathy; Minicore myopathy; Malignant hyperthermia, susceptibility to, 1; not provided; RYR1-Related Disorders; Congenital myopathy with fiber type disproportion; Central core myopathy; Minicore myopathy; Malignant hyperthermia, susceptibility to, 1	Y	-	Y	-
SQSTM1	rs796051870		Paget disease of bone 2, early-onset; Amyotrophic lateral sclerosis and/or frontotemporal dementia 1; Paget disease of bone 3; GNE myopathy	Y	-	-	-
TTN	rs10200398		Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs111671438	V10240F, V9923F	Hypertrophic cardiomyopathy; Cardiomyopathy; Long QT syndrome; Dilated cardiomyopathy 1G; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-
TTN	rs114626713	I7007F, I6690F	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G	-	Y	Y	Y
TTN	rs115744476		Dilated cardiomyopathy 1G; Cardiovascular phenotype; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not provided	Y	Y	Y	Y
TTN	rs115985443		Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs116572520	P6580S, P6263S	Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	Y	Y
TTN	rs117551279	R5819Q, R4892Q	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not provided; not specified; Tibial muscular dystrophy; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	Y	Y
TTN	rs11900987	N3026I, N2980I	Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype	Y	Y	Y	Y
TTN	rs138060032	R279W	not provided; Cardiomyopathy; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	Y	-
TTN	rs138076523	A7543T, A6616T	Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; not specified	-	-	Y	Y
TTN	rs138331646		Dilated cardiomyopathy 1G; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not specified; Cardiomyopathy; not provided	Y	Y	-	-
TTN	rs138560523		not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	-	-	-
TTN	rs138853909	R7223H, R6906H	Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Dilated cardiomyopathy 1G; not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	Y	Y

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN	rs139992576	N3171S, N3125S	Tibial muscular dystrophy; Cardiovascular phenotype; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure	-	-	Y	Y
TTN	rs140003804		Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not specified; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs140640738	E11522D, E10221D, E11148D	Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; not provided	Y	Y	Y	Y
TTN	rs141213991	A1827S, A1781S	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Cardiovascular phenotype; Dilated cardiomyopathy; Heart failure; not specified	Y	Y	-	Y
TTN	rs141258018	R3113H, R3067H	Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	Y	Y
TTN	rs141687561		Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified	Y	-	-	-
TTN	rs141768043		Dilated cardiomyopathy 1G; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs141856116	V8660I, V8343I, V7416I	Ventricular tachycardia; Cardiomyopathy; not specified; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not provided	Y	Y	Y	Y
TTN	rs142414432	A445T	not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs142460433		not specified; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs142951505	V1034M, V988M	Primary dilated cardiomyopathy; Limb-Girdle Muscular Dystrophy, Recessive; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not specified; Cardiovascular phenotype; not provided; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	Y	Y
TTN	rs143030869	A529T	Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs143035953	V2772M, V2726M	not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs143049740		Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy	Y	-	-	-
TTN	rs143368674		Cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Cardiovascular phenotype; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN	rs143845692		Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; not provided; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not specified	Y	Y	-	-
TTN	rs144032104	V7798M, V7481M, V6554M	Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	Y	Y
TTN	rs144135510	R1998H, R1952H	Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; not specified	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN	rs144716589		Cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs144848584	G14867R, G12299R, G13226R, G5994R, G5927R, G5802R	Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	-	-	Y	Y
TTN	rs144930507	T9357I, T9040I, T8113I	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not provided	Y	Y	Y	Y
TTN	rs145204073	I6321V, I5077V, I6004V	Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy	Y	Y	-	-
TTN	rs145211131		Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN	rs146057575	N8891S, N8574S, N7647S	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Hypertrophic cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not specified; not provided	Y	Y	Y	Y
TTN	rs146219199		Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Cardiomyopathy; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	-	-
TTN	rs146496197	R1890C, R1844C	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not specified; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Systolic heart failure; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN	rs146627500	I6725M, I6408M, I5481M	Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Dilated cardiomyopathy; not provided; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN	rs146690035	A570V	Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy	Y	Y	Y	Y
TTN	rs146970027	H2508R, H2462R	not specified; Tibial muscular dystrophy; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN	rs147314430	S3692T	Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Familial hypertrophic cardiomyopathy 9; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not specified; not provided	Y	Y	-	-
TTN	rs148094198		not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN	rs148140756	A10796V, A11723V, A13230V	Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN	rs148169214	R3363H, R3317H	Tibial muscular dystrophy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs149131555	V6286I, V5969I, V5042I	Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	Y	Y
TTN	rs149470241	S6779C, S6462C, S5535C	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; not specified	Y	Y	Y	Y
TTN	rs149523263		Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	Y	Y

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN	rs149523263	F7846L, F7529L F6602L,	not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not provided; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	Y	Y
TTN	rs149668487		Dilated cardiomyopathy 1G; not provided; Cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs149855485	R8497H, R8180H R7253H,	Tibial muscular dystrophy; not provided; Dilated cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	Y	Y
TTN	rs150544093		not specified; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN	rs150667217	V1202A, V1156A	Myopathy, early-onset, with fatal cardiomyopathy; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs150733188	V10865L, V10548L V9621L,	Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	Y	Y
TTN	rs151253841	P5334L	Dilated Cardiomyopathy, Dominant; Limb-Girdle Muscular Dystrophy, Recessive; Tibial muscular dystrophy; not specified; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Hypertrophic cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN	rs17354992	D15110N, D13469N, D6170N, D6237N D12542N, D6045N,	Dilated cardiomyopathy 1G; not specified; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Hypertrophic cardiomyopathy; Cardiovascular phenotype	Y	Y	Y	Y
TTN	rs17452588	S7726L, S7409L S6482L,	Cardiovascular phenotype; Hypertrophic cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	-	Y	Y	Y
TTN	rs181189778	S14564T, S12923T, S5691T, S5499T S11996T, S5624T,	Myopathy, early-onset, with fatal cardiomyopathy; Arrhythmogenic right ventricular cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Tibial muscular dystrophy; not specified; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	Y	Y
TTN	rs181375012	K1129R, K1083R	Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	-	-	Y	Y
TTN	rs181395238	R10985P, R10668P R9741P,	Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	-	-	Y	Y
TTN	rs183336802		Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs183482849	D7462E, D7145E D6218E,	Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure	-	Y	Y	Y
TTN	rs183499397	C7471Y, C8398Y C8715Y,	Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified	-	-	Y	Y
TTN	rs184078045	I13430T, I12503T, I6131T, I6198T I15071T, I6006T,	Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	-	-	Y	Y
TTN	rs184412722	N6275S, N5958S N5031S,	not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	Y	Y	Y
TTN	rs184713215	K11090R, K12017R, K4718R, K4785R K13658R, K4593R,	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	Y	Y

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN	rs184888200		not provided; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Primary dilated cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN	rs185962498	G4604E, G5531E	G5848E, Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not provided	Y	-	Y	Y
TTN	rs186404793	P13206S, P11699S	P10772S, Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	Y	Y
TTN	rs186699871		not provided; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN	rs187365142		Dilated cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not provided	Y	Y	-	-
TTN	rs187703540	R3388Q, R3342Q	Hypertrophic cardiomyopathy; not provided; Cardiovascular phenotype; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G	Y	Y	Y	Y
TTN	rs188234466	K8648E, K8331E	K7404E, not provided; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy	-	-	Y	Y
TTN	rs189966800	K11755T, K11381T	K10454T, Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype	Y	Y	Y	Y
TTN	rs190604150		Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs190636272	K5438R, K5121R	K4194R, Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Dilated cardiomyopathy 1G	Y	Y	Y	Y
TTN	rs192553687	Y5095C, Y6022C	Y6339C, Myopathy, early-onset, with fatal cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not specified	Y	-	-	-
TTN	rs192766485	P12193S, P13120S, P5888S, P5821S	P14761S, P5696S, Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Primary dilated cardiomyopathy; not provided; Cardiovascular phenotype; Familial hypertrophic cardiomyopathy 9; Tibial muscular dystrophy	Y	Y	Y	Y
TTN	rs199525540		Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs199557654	Y7706C, Y8633C	Y8950C, Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Primary familial hypertrophic cardiomyopathy; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified	Y	-	-	-
TTN	rs199848546		Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs199951296		not specified; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs200088963		Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Cardiomyopathy; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs200103997	V7074I, V8001I	V8318I, not specified; Tibial muscular dystrophy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23	
TTN	rs200118743	G6933A, G6616A	G5689A	Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Tibial muscular dystrophy; not provided; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs200187117			Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-
TTN	rs200213832	P10613A, P10296A	P9369A	not provided; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs200459347	P10586Q, P10269Q	P9342Q	Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not provided; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs200549353			Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy	Y	-	-	-
TTN	rs200593368	T10162M, T9845M	T8918M	Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Cardiovascular phenotype; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	-	-	-
TTN	rs200916144			Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN	rs200917885			not specified; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs201005813	G7696S, G8623S	G8940S	Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not provided; not specified	Y	-	-	-
TTN	rs201057307	A15200G, A13559G, A6135G, A6260G, A6327G	A12632G	not specified; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not provided; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype	Y	Y	-	-
TTN	rs201120871	P10022S, P10949S	P11266S	not provided; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs201155967			Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; not specified	Y	Y	-	-
TTN	rs201263441	D6249N, D5932N	D5005N	TTN-Related Disorders; not specified; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	Y	-	-
TTN	rs201284459	T2238M, T2192M		Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Dilated cardiomyopathy 1G; not provided; Limb-girdle muscular dystrophy, type 2J; Ventricular tachycardia; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-
TTN	rs201394117	A7122T, A6805T	A5878T	Cardiomyopathy; not specified; not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN	rs201523784	S8540F, S8223F	S7296F	Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Hypertrophic cardiomyopathy	Y	Y	-	-
TTN	rs201571580	V7078L, V8005L	V8322L	Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN	rs201611946			Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN	rs201625116			Dilated cardiomyopathy 1G; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23	
TTN	rs201653851	R6745C, R7672C	R7989C,	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN	rs201738153	P10853S, P10536S	P9609S,	Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; not provided; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs201857541	V15I		Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified	Y	-	-	-
TTN	rs201904848			not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs201944202	V13500F, V11859F, V4560F, V4627F	V10932F, V4435F,	Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	-	-
TTN	rs202014478	V11622I, V11248I	V10321I,	Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	-	-
TTN	rs202089818			not provided; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN	rs202160275	V8259I, V9186I	V9503I,	Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not specified; Cardiovascular phenotype; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN	rs34706299	V14110A, V12469A, V5170A, V11542A, V5045A, V5237A		Dilated cardiomyopathy 1G; Cardiovascular phenotype; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; mitochondrial; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN	rs367700246	T1733M, T1687M		Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs367826445	G6433R, G7360R	G7677R,	not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Cardiovascular phenotype; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs368101794			Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not specified	Y	-	-	-
TTN	rs368282893	V1099M, V1053M		Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	-	-	Y	Y
TTN	rs368548209			Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs369128249			Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy	Y	-	-	-
TTN	rs369265969			Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	-	-	-
TTN	rs369275615			not provided; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs369959066			Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Dilated cardiomyopathy 1G; not specified; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN	rs370080995		not specified; Dilated cardiomyopathy 1G; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs370597107		Cardiovascular phenotype; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs371496970	I6781M, I7708M	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Tibial muscular dystrophy	Y	Y	-	-
TTN	rs371826762	I8337V, I9264V	Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Supraventricular tachycardia; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; not provided; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs372382546		Myopathy, early-onset, with fatal cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN	rs372528823		Tibial muscular dystrophy; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN	rs372802604		Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy	Y	-	-	-
TTN	rs372997298		not specified; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy	Y	-	-	-
TTN	rs373366126	L3097P, L3051P	not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy	Y	-	-	-
TTN	rs373378672	D4845N, D4528N	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided	Y	-	-	-
TTN	rs373429851		Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided	Y	Y	-	-
TTN	rs373491468	R14526Q, R11958Q, R12885Q, R5461Q, R5653Q, R5586Q	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs373636988		Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Hypertrophic cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs374156904	R9516H, R8272H	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Cardiovascular phenotype; Cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs374620001	T2690I, T2644I	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs375209098	P7132S, P8059S	Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not provided	Y	-	-	-
TTN	rs375266859	R9744C, R9427C	Myopathy, early-onset, with fatal cardiomyopathy; Ichthyosis (disease); Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not specified; not provided; Cardiomyopathy; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Cardiac arrest; Sudden cardiac death	Y	-	-	-
TTN	rs375448572		Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN	rs375519815		not specified; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs376037792		Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy	Y	Y	-	-
TTN	rs376768790	V896I, V850I	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; not specified	Y	Y	-	-
TTN	rs376836503	L11160P, L10233P, L11534P	Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN	rs376874956	E11286K, E10042K, E10969K	not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Cardiomyopathy; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy	Y	-	-	-
TTN	rs377049518	E3276G, E3230G	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not provided	Y	-	-	-
TTN	rs397517521		Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Primary dilated cardiomyopathy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs397517540		Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not specified	Y	-	-	-
TTN	rs397517560		Hypertrophic cardiomyopathy; Cardiomyopathy; Tibial muscular dystrophy; Dilated Cardiomyopathy, Dominant; Limb-Girdle Muscular Dystrophy, Recessive; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs397517725	R2540H, R2494H	Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs45447891		Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Cardiovascular phenotype; not provided	Y	-	-	-
TTN	rs532102837	P10162T, P11089T, P11406T	Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiomyopathy; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided	Y	-	-	-
TTN	rs550471556		Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Tibial muscular dystrophy; not specified	Y	-	-	-
TTN	rs552951988	D6431Y, D7358Y, D7675Y	Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	Y	-	-
TTN	rs55914517	A1081S, A1035S	Myopathy, early-onset, with fatal cardiomyopathy; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs55914517	A1081T, A1035T	not provided; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; not specified; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Hypertrophic cardiomyopathy	Y	Y	-	-
TTN	rs55972547		Cardiovascular phenotype; Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN	rs56046320	R922H, R876H	Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs56812642		Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not specified; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs72647870	G1137R, G1091R	not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Hypertrophic cardiomyopathy; Cardiovascular phenotype	Y	Y	Y	Y
TTN	rs72647883		Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs72647885	A2980T, A2934T	Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not specified	Y	Y	Y	Y
TTN	rs72647894	R3120Q, R3074Q	Atrial fibrillation; not specified; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not provided; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	Y	Y
TTN	rs72648923	A4900T, A4583T	A3656T, Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Long QT syndrome; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	Y	Y
TTN	rs72648925	T4957S, T4640S	T3713S, not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; not provided	Y	Y	Y	Y
TTN	rs72648929	V5060L, V4743L	V3816L, not provided; Tibial muscular dystrophy; Cardiovascular phenotype; Supraventricular tachycardia; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J	-	Y	-	-
TTN	rs72648930	Q5188P, Q4871P	Q3944P, Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified	Y	Y	-	-
TTN	rs72648937	V5435M, V5118M	V4191M, not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Heart failure; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	Y	Y
TTN	rs72648940	D5516Y, D5199Y	D4272Y, not provided; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy; Arrhythmogenic right ventricular cardiomyopathy; Cardiomyopathy; Dilated cardiomyopathy 1G	Y	Y	Y	Y
TTN	rs72648942	Y5683C, Y5366C	Y4439C, Myopathy, early-onset, with fatal cardiomyopathy; Brugada syndrome; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Cardiovascular phenotype; not specified; not provided; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	Y	Y
TTN	rs72648943	L5742F, L5425F	L4498F, Hypertrophic cardiomyopathy; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs72648949	T6259S, T5942S	T5015S, Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy; Atrial fibrillation; Arrhythmogenic right ventricular cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; not specified; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Dilated cardiomyopathy 1G; Tibial muscular dystrophy	Y	Y	Y	Y
TTN	rs72648953	P6384T, P6067T	P5140T, not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	Y	Y

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23	
TTN	rs72648958	E6781K, E6464K	E5537K	Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-
TTN	rs72648964	G7058D, G6741D	G5814D	Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J	Y	Y	Y	Y
TTN	rs72648965			not specified; Primary familial hypertrophic cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-
TTN	rs72648969	R7545Q, R7248Q	R6301Q	Limb-girdle muscular dystrophy, type 2J; not specified; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs72648971			Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs72648973	D7768N, D7451N	D6524N	not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN	rs72648975			Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not specified; not provided	Y	Y	-	-
TTN	rs72648980	S8218G, S7901G	S6974G	Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Cardiovascular phenotype; not provided; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J	Y	Y	Y	Y
TTN	rs72648981	E8274K, E7957K	E7030K	not specified; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	Y	Y	Y
TTN	rs72648989	I7588V, I8515V	I8832V	Myopathy, myofibrillar, 9, with early respiratory failure; not provided; not specified; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	Y	Y	Y
TTN	rs72648997			Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; not provided; Tibial muscular dystrophy	Y	Y	-	-
TTN	rs72650008			Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy	Y	Y	-	-
TTN	rs72650019	V10467I, V10150I	V9223I	Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not provided; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs72650030	A10827V, A10510V	A9583V	not provided; not specified; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs72650031	P10875L, P10558L	P9631L	Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Ventricular tachycardia; Supraventricular tachycardia; Brugada syndrome; not provided; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	Y	Y
TTN	rs72650034	R11018Q, R10701Q	R9774Q	not provided; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Cardiovascular phenotype; EMG: slow motor conduction; Distal lower limb amyotrophy; Intrinsic hand muscle atrophy; Foot dorsiflexor weakness; Pes cavus	Y	Y	-	-
TTN	rs72650035	Y11020C, Y10703C	Y9776C	Tibial muscular dystrophy; Arrhythmogenic right ventricular cardiomyopathy; not specified; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not provided; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN	rs72650040	R11139T, R9895T, R10822T	Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	Y	Y
TTN	rs72650064	P10722T, P11649T, P13156T	not provided; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Supraventricular tachycardia; Cardiomyopathy; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Primary familial hypertrophic cardiomyopathy	Y	Y	-	-
TTN	rs72650077		not provided; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs72677223		not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN	rs72677225	K15136N, K12568N, K6263N, K13495N, K6196N, K6071N	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not specified; Cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	Y	Y
TTN	rs727503679	V3342I, V3296I	Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs73038337		Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	-	-	-
TTN	rs73973137	P10917L, P10600L, P9673L, P10600L	not specified; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiovascular phenotype	Y	Y	Y	Y
TTN	rs73973139	K4865E, K5792E, K6109E	not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; not provided	Y	-	-	-
TTN	rs74324101		Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Dilated cardiomyopathy 1G	Y	-	-	-
TTN	rs746912694		Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs747722195		Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy	Y	-	-	-
TTN	rs75031300	R2354H, R2308H	not provided; Cardiomyopathy; Tibial muscular dystrophy; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	-	-	Y	Y
TTN	rs755262343		Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy	Y	-	-	-
TTN	rs761666344		Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN	rs772821743		Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN	rs775954427		not specified; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN	rs780101457		Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN	rs781392140		Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN	rs876657615		not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy	Y	-	-	-
TTN, LOC10192705	rs36021856	G1345D, G1299D	Dilated cardiomyopathy 1G; not specified; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	Y	Y
TTN, LOC101927055	rs138506461	S1400T, S1354T	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	-	-	-
TTN, LOC101927055	rs145709534		Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy	Y	-	-	-
TTN, LOC101927055	rs151310601	F1466L, F1420L	not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiomyopathy; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Arrhythmogenic right ventricular cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, LOC101927055	rs72647876	R1441H, R1395H	Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	Y	Y	Y
TTN, TTN-AS1	rs111727915	R29574H, R27006H, R27933H, R20634H, R20509H, R20701H	Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Cardiomyopathy; Tibial muscular dystrophy; not provided; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, TTN-AS1	rs114026724	I30213T, I27645T, I28572T, I21340T, I21273T, I21148T	Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiovascular phenotype; not specified; Arrhythmogenic right ventricular cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs115813214	A15627V, A13059V, A13986V, A6562V, A6754V, A6687V	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	-	-	-
TTN, TTN-AS1	rs116142642	V18229M, V15661M, V16588M, V9164M, V9356M, V9289M	not specified; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs118079537	R24995H, R27563H, R25922H, R18498H, R18623H, R18690H	Tibial muscular dystrophy; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs138576504	L15913F, L13345F, L14272F, L6848F, L7040F, L6973F	Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype	Y	Y	-	-
TTN, TTN-AS1	rs139223781		Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, TTN-AS1	rs139506970	I27002V, I29570V, I27929V, I20505V, I20697V, I20630V	Cardiovascular phenotype; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs139789997		Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, TTN-AS1	rs139953862		not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, TTN-AS1	rs140018785	R21700H, R24268H, R22627H, R15395H, R15203H, R15328H	Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy; not specified	-	Y	-	-
TTN, TTN-AS1	rs140319117	R31523W, R34091W, R32450W, R25026W, R25151W, R25218W	not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Dilated cardiomyopathy 1G; not provided; Heart failure; Dilated cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs140350441		Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not provided; Cardiovascular phenotype	Y	Y	-	-
TTN, TTN-AS1	rs141624266	V29270I, V26702I, V27629I, V20205I, V20330I, V20397I	Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs142095604		Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Ventricular tachycardia; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; not provided; not specified; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, TTN-AS1	rs142525903	R31748C, R29180C, R30107C, R22683C, R22875C, R22808C	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; not provided; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	-	-
TTN, TTN-AS1	rs142874389	R24261Q, R21693Q, R22620Q, R15196Q, R15388Q, R15321Q	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not provided	Y	Y	-	-
TTN, TTN-AS1	rs143499441	I35218T, I32650T, I33577T, I26153T, I26345T, I26278T	Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not provided	Y	-	-	-
TTN, TTN-AS1	rs143975327	M26478R, M29046R, M27405R, M19981R, M20106R, M20173R	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, TTN-AS1	rs144963736	D34035N, D31467N, D32394N, D24970N, D25162N, D25095N	Dilated cardiomyopathy; Cardiomyopathy; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs146181116	S29465F, S26897F, S27824F, S20400F, S20592F, S20525F	Tibial muscular dystrophy; Hypertrophic cardiomyopathy; Arrhythmogenic right ventricular cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Cardiovascular phenotype; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs146400809	P10821L, P9577L, P10504L	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Abnormality of neuronal migration; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Hypertrophic cardiomyopathy; not specified	Y	Y	Y	Y
TTN, TTN-AS1	rs146767076	A24046V, A21478V, A22405V, A14981V, A15106V, A15173V	Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs146941600	I19342T, I21910T, I20269T, I12845T, I13037T, I12970T	Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; not provided; Cardiovascular phenotype; Dilated cardiomyopathy 1G; not specified	Y	-	-	-
TTN, TTN-AS1	rs147703145		Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs148067743	R27957T, R25389T, R26316T, R19017T, R19084T, R18892T	Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not provided; Brugada syndrome	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, AS1	rs148231754		Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Dilated cardiomyopathy 1G	Y	-	Y	Y
TTN, AS1	rs148617456	A30589T, A28021T, A28948T, A21524T, A21716T, A21649T	not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Cardiovascular phenotype	Y	-	-	-
TTN, AS1	rs148849567	R21916Q, R19348Q, R20275Q, R13043Q, R12851Q, R12976Q	Cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Cardiovascular phenotype	Y	-	-	-
TTN, AS1	rs149001703	N32797S, N30229S, N31156S, N23732S, N23857S, N23924S	Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Long QT syndrome; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not specified; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs149075285	I21083S, I23651S, I22010S, I14586S, I14778S, I14711S	not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs149391616	R31903Q, R34471Q, R32830Q, R25598Q, R25406Q, R25531Q	not provided; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, AS1	rs149763294	E24127K, E21559K, E22486K, E15062K, E15187K, E15254K	Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Cardiomyopathy; Tibial muscular dystrophy; not provided	Y	Y	-	-
TTN, AS1	rs150661999	V21597M, V19029M, V19956M, V12532M, V12657M, V12724M	Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Dilated cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy	Y	Y	-	-
TTN, AS1	rs180975448	S28434I, S31002I, S29361I, S21937I, S22062I, S22129I	Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Cardiovascular phenotype; not provided; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, AS1	rs182491843	T24390A, T21822A, T22749A, T15325A, T15450A, T15517A	Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype	Y	Y	-	-
TTN, AS1	rs182683829	V32906I, V31265I, V30338I, V23841I, V24033I, V23966I	Myopathy, early-onset, with fatal cardiomyopathy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not specified	Y	Y	-	-
TTN, AS1	rs183013408	G29364S, G26796S, G27723S, G20299S, G20491S, G20424S	Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; not provided	Y	-	-	-
TTN, AS1	rs183923129		not specified; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not provided	Y	Y	-	-
TTN, AS1	rs184078016	T28771I, T31339I, T29698I, T22274I, T22466I, T22399I	Tibial muscular dystrophy; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, AS1	rs184863287		Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not provided; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, AS1	rs185002960	R25120H, R27688H, R26047H, R18623H, R18748H, R18815H	Myopathy, early-onset, with fatal cardiomyopathy; not specified; Tibial muscular dystrophy; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-
TTN, AS1	rs185683410	A34751P, A32183P, A33110P, A25686P, A25878P, A25811P	not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, TTN-AS1	rs185887755	R26852C, R25211C, R17912C, R17979C, R17787C	Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not provided	Y	-	-	-
TTN, TTN-AS1	rs186234393	N29259S, N30900S, N21835S, N28332S, N21960S, N22027S	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Hypertrophic cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Dilated Cardiomyopathy, Dominant; Tibial muscular dystrophy; Limb-Girdle Muscular Dystrophy, Recessive; Cardiovascular phenotype; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs186563991		Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; not specified; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-
TTN, TTN-AS1	rs186681106	W25938S, W23370S, W24297S, W16873S, W16998S, W17065S	Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, TTN-AS1	rs187270666	R25730Q, R28298Q, R26657Q, R19233Q, R19425Q, R19358Q	Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs187868672	N24256D, N22615D, N21688D, N15191D, N15316D, N15383D	Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not provided; not specified; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, TTN-AS1	rs188063446		Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs188370772	T25346A, T27914A, T26273A, T18849A, T19041A, T18974A	not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs188917199	V34473I, V32832I, V25533I, V25600I, V25408I	Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not provided	Y	-	-	-
TTN, TTN-AS1	rs190058852		Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, TTN-AS1	rs190565627	E34789K, E32221K, E33148K, E25724K, E25916K, E25849K	Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	Y	-	-
TTN, TTN-AS1	rs190830121		Myopathy, myofibrillar, 9, with early respiratory failure; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs191539637	R14337H, R16905H, R15264H, R7840H, R8032H, R7965H	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not provided; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs192001910	T34431M, T31863M, T32790M, T25366M, T25491M, T25558M	not provided; Cardiovascular phenotype; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs192360370	R27694C, R25126C, R18754C, R26053C, R18821C, R18629C	Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs193022702		not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs199505541	E27823D, E30391D, E28750D, E21326D, E21451D, E21518D	Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, ASI	rs199506676	E20374K, E22942K, E21301K, E13877K, E14069K, E14002K	Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Primary dilated cardiomyopathy; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Familial hypertrophic cardiomyopathy 9; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN, ASI	rs199531140	E33129Q, E35697Q, E34056Q, E26632Q, E26757Q, E26824Q	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy	Y	-	-	-
TTN, ASI	rs199632397	E32991Q, E35559Q, E33918Q, E26494Q, E26619Q, E26686Q	Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Inborn genetic diseases; not provided; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Cardiomyopathy; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, ASI	rs199670463	G24745E, G27313E, G25672E, G18248E, G18373E, G18440E	not specified; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, ASI	rs199714102		Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy	Y	Y	-	-
TTN, ASI	rs199805060	T30188I, T32756I, T31115I, T23816I, T23691I, T23883I	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, ASI	rs199875474		Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, ASI	rs199895260	R20641Q, R18073Q, R19000Q, R11576Q, R11701Q, R11768Q	Hypertrophic cardiomyopathy; Cardiomyopathy; not specified; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Cardiovascular phenotype	Y	Y	-	-
TTN, ASI	rs199929362	G25036S, G27604S, G25963S, G18539S, G18664S, G18731S	Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Tibial muscular dystrophy	Y	-	-	-
TTN, ASI	rs200042932	G15463R, G12895R, G13822R, G6590R, G6523R, G6398R	Limb-girdle muscular dystrophy, type 2J; not provided; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN, ASI	rs200141081	R28327Q, R30895Q, R29254Q, R21830Q, R22022Q, R21955Q	Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, ASI	rs200144345	R21111K, R23679K, R22038K, R14614K, R14806K, R14739K	not provided; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy	Y	Y	-	-
TTN, ASI	rs200155485	R19348W, R21916W, R20275W, R12976W, R12851W, R13043W	not provided; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, ASI	rs200173934		Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Cardiovascular phenotype	Y	Y	-	-
TTN, ASI	rs200212521	E20241Q, E17673Q, E18600Q, E11176Q, E11301Q, E11368Q	Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, ASI	rs200237973	A34064T, A31496T, A24999T, A32423T, A25124T, A25191T	Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; not provided	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, TTN-AS1	rs200343420	V22300I, V19732I, V20659I, V13235I, V13360I, V13427I	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy	Y	-	-	-
TTN, TTN-AS1	rs200378865	R35043C, R32475C, R33402C, R25978C, R26103C, R26170C	Tibial muscular dystrophy; not specified; Cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, TTN-AS1	rs200406978	F24414L, F26982L, F25341L, F18109L, F17917L, F18042L	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; not specified; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, TTN-AS1	rs200410212		Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not specified; Cardiovascular phenotype; Dilated cardiomyopathy 1G; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN, TTN-AS1	rs200450022	S28088N, S25520N, S26447N, S19215N, S19148N, S19023N	Myopathy, early-onset, with fatal cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, TTN-AS1	rs200476500	V28368I, V30936I, V29295I, V21871I, V21996I, V22063I	Myopathy, myofibrillar, 9, with early respiratory failure; not specified; not provided; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs200497615	R35652Q, R33084Q, R34011Q, R26587Q, R26779Q, R26712Q	Tibial muscular dystrophy; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, TTN-AS1	rs200650668	S17270I, S14702I, S15629I, S8205I, S8397I, S8330I	Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Cardiomyopathy; Cardiovascular phenotype; not specified; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, TTN-AS1	rs200689750	T20858A, T18290A, T19217A, T11918A, T11793A, T11985A	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Limb-girdle muscular dystrophy, type 2J; not specified; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs200697681	D24730N, D27298N, D25657N, D18233N, D18358N, D18425N	Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not specified	Y	Y	-	-
TTN, TTN-AS1	rs200726948	R21118W, R18550W, R12178W, R12245W, R19477W, R12053W	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Cardiovascular phenotype; not specified; Cardiomyopathy; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs200775919	V27873A, V25305A, V18808A, V19000A, V18933A, V26232A	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Familial hypertrophic cardiomyopathy 9; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not provided	Y	-	-	-
TTN, TTN-AS1	rs200825430		Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype	Y	-	-	-
TTN, TTN-AS1	rs200843338	R28326Q, R25758Q, R26685Q, R19386Q, R19453Q, R19261Q	Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; Cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, TTN-AS1	rs200854704	G30541R, G27973R, G28900R, G21476R, G21601R, G21668R	Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs200968679	V15486L, V18054L, V16413L, V9114L, V8989L, V9181L	not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, AS1	rs200971254	R22231H, R19663H, R20590H, R13166H, R13291H, R13358H	Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, AS1	rs201019681	T30778R, T28210R, T29137R, T21905R, T21713R, T21838R	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not provided; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, AS1	rs201035511	P18273L, P15705L, P16632L, P9208L, P9400L, P9333L	Limb-girdle muscular dystrophy, type 2J; not specified; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, AS1	rs201043950	G23274D, G20706D, G21633D, G14209D, G14334D, G14401D	Inborn genetic diseases; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; not specified; Cardiovascular phenotype; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, AS1	rs201080904	R29945H, R32513H, R30872H, R23448H, R23573H, R23640H	not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Cardiovascular phenotype	Y	Y	-	-
TTN, AS1	rs201095164	D25558V, D22990V, D16493V, D16618V, D16685V, D23917V	Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, AS1	rs201158906	G27497R, G24929R, G25856R, G18432R, G18624R, G18557R	Cardiomyopathy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	-	-	-
TTN, AS1	rs201167216	L17812M, L20380M, L18739M, L11315M, L11440M, L11507M	Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; Cardiomyopathy; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, AS1	rs201213901	R17618C, R15050C, R15977C, R8678C, R8553C, R8745C	Cardiomyopathy; Dilated cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; not provided; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, AS1	rs201218828	E32357A, E34925A, E25985A, E33284A, E25860A, E26052A	Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Primary familial hypertrophic cardiomyopathy; not provided; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, AS1	rs201246720		Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not provided; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Tibial muscular dystrophy	Y	Y	-	-
TTN, AS1	rs201272728	Q25938K, Q28506K, Q26865K, Q19441K, Q19633K, Q19566K	Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs201285872		Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; not provided; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, AS1	rs201290358	V28895M, V26327M, V27254M, V19830M, V19955M, V20022M	Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; not provided; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, AS1	rs201348580	I25482V, I28050V, I26409V, I19110V, I18985V, I19177V	Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, AS1	rs201400267	A30681D, A28113D, A29040D, A21741D, A21616D, A21808D	Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; not provided; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, TTN-AS1	rs201412693	L15669P, L18237P, L16596P, L9364P, L9297P, L9172P	Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Hypertrophic cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not provided; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Tibial muscular dystrophy	Y	Y	-	-
TTN, TTN-AS1	rs201450276		Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	-	-	-
TTN, TTN-AS1	rs201489661	T24844K, T27412K, T25771K, T18347K, T18539K, T18472K	Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not provided; Dilated cardiomyopathy 1G; not specified	-	Y	-	-
TTN, TTN-AS1	rs201505306	R16841H, R19409H, R17768H, R10344H, R10469H, R10536H	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype; not specified; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs201512527	Q24965H, Q22397H, Q23324H, Q16025H, Q15900H, Q16092H	Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; not provided; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	Y	-	-
TTN, TTN-AS1	rs201554140	V33749M, V31181M, V32108M, V24684M, V24809M, V24876M	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not specified; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy	Y	-	-	-
TTN, TTN-AS1	rs201662134	P19277L, P21845L, P20204L, P12780L, P12905L, P12972L	Myopathy, myofibrillar, 9, with early respiratory failure; not provided; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs201687390	V26411I, V28979I, V27338I, V20106I, V19914I, V20039I	Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs201717871	V13125I, V15693I, V14052I, V6628I, V6753I, V6820I	not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not provided	Y	Y	-	-
TTN, TTN-AS1	rs201770959		not provided; Cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	Y	-	-
TTN, TTN-AS1	rs201804005	S21967Y, S24535Y, S22894Y, S15470Y, S15595Y, S15662Y	Myopathy, early-onset, with fatal cardiomyopathy; Myopathy; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified	Y	Y	-	-
TTN, TTN-AS1	rs201825412	R15620Q, R17261Q, R14693Q, R8321Q, R8388Q, R8196Q	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs201831707	Y29208F, Y26640F, Y27567F, Y20335F, Y20143F, Y20268F	Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	Y	-	-
TTN, TTN-AS1	rs201835888	S35869G, S33301G, S34228G, S26804G, S26996G, S26929G	Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs201840554	E19546Q, E16978Q, E17905Q, E10481Q, E10673Q, E10606Q	Dilated cardiomyopathy 1G; Cardiomyopathy; Ventricular fibrillation; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Cardiovascular phenotype; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs201896662	V27724G, V25156G, V26083G, V18784G, V18851G, V18659G	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; not specified; not provided; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs201958805	K27468Q, K24900Q, K25827Q, K18528Q, K18595Q, K18403Q	Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, AS1	rs202040332	S28469C, S25901C, S26828C, S19529C, S19596C, S19404C	not provided; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not specified	Y	Y	-	-
TTN, AS1	rs202064385	R32367C, R29799C, R30726C, R23302C, R23494C, R23427C	not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Cardiovascular phenotype; not specified	Y	Y	-	-
TTN, AS1	rs202094100	W16471C, W13903C, W14830C, W7531C, W7598C, W7406C	Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Familial hypertrophic cardiomyopathy 9; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not specified; Dilated cardiomyopathy 1G; Cardiomyopathy; Tibial muscular dystrophy; not provided; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	Y	-	-
TTN, AS1	rs202141158		Cardiovascular phenotype; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Tibial muscular dystrophy	Y	Y	-	-
TTN, AS1	rs202185465		Limb-girdle muscular dystrophy, type 2J; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, AS1	rs202240487	R21306H, R18738H, R19665H, R12366H, R12241H, R12433H	Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not provided; not specified; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, AS1	rs2288326	A31693D, A29125D, A30052D, A22628D, A22820D, A22753D	Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-
TTN, AS1	rs2303834		Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy	Y	-	-	-
TTN, AS1	rs281864930	K27023fs, K27090fs, K26898fs, K35963fs, K33395fs, K34322fs	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	Y	Y
TTN, AS1	rs34924609	V33889I, V31321I, V32248I, V24949I, V25016I, V24824I	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Supraventricular tachycardia; Hypertrophic cardiomyopathy; Tibial muscular dystrophy; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified	Y	Y	-	-
TTN, AS1	rs367622770		Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; not specified	Y	Y	-	-
TTN, AS1	rs367732133		Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; not specified	Y	Y	-	-
TTN, AS1	rs367908657		not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, AS1	rs367979582	P30313S, P32881S, P31240S, P23816S, P23941S, P24008S	Tibial muscular dystrophy; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs368243641		Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, AS1	rs368270588		not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs368686031		not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; not specified	Y	Y	-	-
TTN, AS1	rs368692510		Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, TTN-AS1	rs368716894	V19157I, V21725I, V20084I, V12852I, V12660I, V12785I	Dilated cardiomyopathy 1G; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs368759398		Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	Y	-	-
TTN, TTN-AS1	rs368867993	I20720M, I23288M, I21647M, I14415M, I14223M, I14348M	Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs368914555	R25012W, R22444W, R23371W, R16072W, R16139W, R15947W	not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs369626133		Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs369707906	R24000H, R25641H, R23073H, R16701H, R16576H, R16768H	Sudden cardiac arrest; Hypertrophic cardiomyopathy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated Cardiomyopathy, Dominant; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-Girdle Muscular Dystrophy, Recessive; Myopathy, early-onset, with fatal cardiomyopathy; not specified	Y	Y	-	-
TTN, TTN-AS1	rs369855092	L27429M, L29997M, L28356M, L21057M, L21124M, L20932M	Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs371075036		Dilated cardiomyopathy 1G; Cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	-	-
TTN, TTN-AS1	rs371081043	K19596I, K22164I, K20523I, K13099I, K13224I, K13291I	not specified; Tibial muscular dystrophy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs371344165		Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; not specified; not provided; Cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs371480338		not specified; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs371612136		not specified; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	-	-	-
TTN, TTN-AS1	rs371788070	V25193L, V27761L, V18696L, V18888L, V26120L, V18821L	Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, TTN-AS1	rs371802557		not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated Cardiomyopathy, Dominant; Limb-Girdle Muscular Dystrophy, Recessive; Hypertrophic cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs371943746	G13355R, G15923R, G14282R, G6858R, G6983R, G7050R	not provided; Inborn genetic diseases; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs372019333		Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy; Cardiomyopathy; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	-	-	-
TTN, TTN-AS1	rs372065796		Myopathy, early-onset, with fatal cardiomyopathy; not specified; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not provided; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, AS1	rs372382315		not provided; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, AS1	rs372635204		Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; not provided; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, AS1	rs3731746	T24053K, T26621K, T24980K, T17681K, T17556K, T17748K	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy	Y	-	-	-
TTN, AS1	rs3731753		Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype	Y	-	-	-
TTN, AS1	rs373206096		Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy; not provided	Y	-	-	-
TTN, AS1	rs373305248	D17380V, D14812V, D15739V, D8315V, D8440V, D8507V	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Tibial muscular dystrophy	Y	-	-	-
TTN, AS1	rs373316165		Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not provided; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, AS1	rs373514079		Familial hypertrophic cardiomyopathy 9; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; not specified	Y	-	-	-
TTN, AS1	rs373526624	R17836H, R15268H, R16195H, R8771H, R8963H, R8896H	not specified; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, AS1	rs373727636	I28331T, I30899T, I29258T, I21834T, I22026T, I21959T	not specified; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; not provided	Y	-	-	-
TTN, AS1	rs373853269		Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, AS1	rs373871146		Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not provided; Tibial muscular dystrophy; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, AS1	rs374058726	I17575V, I19216V, I16648V, I10151V, I10276V, I10343V	not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Inborn genetic diseases; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, AS1	rs374081262		Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified	Y	-	-	-
TTN, AS1	rs374168580		Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified	Y	Y	-	-
TTN, AS1	rs374612925		Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, AS1	rs374858668		Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not specified	Y	Y	-	-
TTN, AS1	rs374878689		not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not provided	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, TTN-AS1	rs375006117		Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Cardiovascular phenotype	Y	Y	-	-
TTN, TTN-AS1	rs375398118		Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	-	-	-
TTN, TTN-AS1	rs375538420		Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs375553630		Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs375565646		Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not provided; not specified; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs376273101	D13550G, D16118G, D14477G, D7245G, D7053G, D7178G	Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs376532382		Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified	Y	Y	-	-
TTN, TTN-AS1	rs376539252		Myopathy, myofibrillar, 9, with early respiratory failure; not provided; not specified; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs376810671		not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, TTN-AS1	rs376820301	R27839Q, R26198Q, R25271Q, R18774Q, R18899Q, R18966Q	Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Cardiovascular phenotype; Hypertrophic cardiomyopathy; not specified	Y	Y	-	-
TTN, TTN-AS1	rs376894729		Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs377220635		Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided	Y	-	-	-
TTN, TTN-AS1	rs377288086		Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not specified	Y	Y	-	-
TTN, TTN-AS1	rs377337528	A33039V, A35607V, A33966V, A26542V, A26667V, A26734V	Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not specified	Y	-	-	-
TTN, TTN-AS1	rs377506142	T24385M, T26953M, T25312M, T18013M, T18808M, T17888M	Limb-girdle muscular dystrophy, type 2J; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Cardiomyopathy; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy	Y	-	-	-
TTN, TTN-AS1	rs377618488		Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs377682563	V16958I, V19526I, V10461I, V10653I, V17885I, V10586I	Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy	Y	Y	-	-
TTN, TTN-AS1	rs3813249	S32503C, S35071C, S33430C, S26198C, S26131C, S26006C	Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, AS1	rs397517612	F17754L, F15186L, F16113L, F8689L, F8814L, F8881L	Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs397517620		Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Tibial muscular dystrophy	Y	-	Y	Y
TTN, AS1	rs397517627	A18981V, A16413V, A17340V, A10108V, A9916V, A10041V	not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, AS1	rs397517630	L19196V, L16628V, L17555V, L10256V, L10131V, L10323V	Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Ventricular tachycardia; Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Tibial muscular dystrophy	Y	-	-	-
TTN, AS1	rs397517636	V19476I, V16908I, V17835I, V10411I, V10536I, V10603I	Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; not specified	Y	Y	-	-
TTN, AS1	rs397517719	E26995K, E24427K, E25354K, E17930K, E18055K, E18122K	Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Cardiovascular phenotype	Y	-	-	-
TTN, AS1	rs397517730	E28232D, E25664D, E19292D, E19359D, E19167D, E26591D	Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided	Y	-	-	-
TTN, AS1	rs397517759	I31543M, I28975M, I22478M, I22670M, I22603M, I29902M	Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy	Y	-	-	-
TTN, AS1	rs397517771	G32581V, G30013V, G30940V, G23516V, G23641V, G23708V	not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs397517772	A32620V, A30052V, A23555V, A23747V, A23680V, A30979V	Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, AS1	rs397517781	G33277E, G30709E, G31636E, G24212E, G24404E, G24337E	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated Cardiomyopathy, Dominant; Limb-Girdle Muscular Dystrophy, Recessive; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Hypertrophic cardiomyopathy; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy	Y	Y	-	-
TTN, AS1	rs397517782		Cardiomyopathy; Hypertrophic cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Dilated Cardiomyopathy, Dominant; Limb-Girdle Muscular Dystrophy, Recessive; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs530507211	R24978H, R26619H, R24051H, R17554H, R17746H, R17679H	not provided; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, AS1	rs545443009	W33439R, W30871R, W31798R, W24499R, W24374R, W24566R	not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs553313488		not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs55634791	A27564V, A24996V, A25923V, A18691V, A18624V, A18499V	Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs55660660	I29929T, I32497T, I30856T, I23432T, I23624T, I23557T	Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; Dilated cardiomyopathy	Y	-	-	-
TTN, AS1	rs55663050	S16640T, S14072T, S14999T, S7575T, S7700T, S7767T	not provided; Dilated cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; not specified	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, TTN-AS1	rs55669553	R33964H, R31396H, R32323H, R24899H, R25024H, R25091H	not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Dilated cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs55696153		Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	Y	-	-
TTN, TTN-AS1	rs55704830	R32587P, R30019P, R30946P, R23522P, R23647P, R23714P	Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs55704830	R32587H, R30019H, R30946H, R23522H, R23714H, R23647H	Myopathy, myofibrillar, 9, with early respiratory failure; Inborn genetic diseases; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not specified; not provided	Y	Y	-	-
TTN, TTN-AS1	rs55725279	E35527V, E32959V, E33886V, E26462V, E26587V, E26654V	not specified; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	-	-	-
TTN, TTN-AS1	rs55734111	R18050C, R15482C, R16409C, R8985C, R9110C, R9177C	Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs55762754	E24609Q, E22041Q, E22968Q, E15669Q, E15736Q, E15544Q	not provided; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	-	-	-
TTN, TTN-AS1	rs55837610	I23902T, I21334T, I22261T, I14837T, I14962T, I15029T	Premature ventricular contraction; not provided; Cardiovascular phenotype; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs55838839		not provided; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs55842557	T35596I, T33028I, T33955I, T26656I, T26531I, T26723I	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	Y	Y	Y
TTN, TTN-AS1	rs55847238		Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy	Y	Y	-	-
TTN, TTN-AS1	rs55857909		Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Cardiovascular phenotype	Y	-	-	-
TTN, TTN-AS1	rs558830502	E24137D, E26705D, E25064D, E17832D, E17640D, E17765D	Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy	Y	-	-	-
TTN, TTN-AS1	rs55892928		Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs55938627		Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not specified; Cardiovascular phenotype; Cardiomyopathy; Tibial muscular dystrophy; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	-	Y	-	-
TTN, TTN-AS1	rs55945684	V34563A, V31995A, V32922A, V25690A, V25498A, V25623A	Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Hypertrophic cardiomyopathy; Cardiovascular phenotype; not specified	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, ASI	rs55980498	P23044S, P20476S, P21403S, P13979S, P14171S, P14104S	Atrial fibrillation; Hypertrophic cardiomyopathy; Brugada syndrome; Congestive heart failure; not specified; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not provided; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	Y	-	-
TTN, ASI	rs56044609	R26440H, R23872H, R24799H, R17567H, R17375H, R17500H	not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	-	-	-
TTN, ASI	rs56061641	C33331R, C30763R, C31690R, C24266R, C24458R, C24391R	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Hypertrophic cardiomyopathy	Y	Y	-	-
TTN, ASI	rs56169243		Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not provided; Tibial muscular dystrophy; not specified; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	Y	-	-
TTN, ASI	rs56173891	M31683V, M34251V, M32610V, M25186V, M25311V, M25378V	Hypertrophic cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-
TTN, ASI	rs56181243		Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; not specified; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, ASI	rs56201325	T24311A, T21743A, T22670A, T15246A, T15371A, T15438A	not specified; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, ASI	rs56245285		Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	-	Y	-	-
TTN, ASI	rs56264840	N27520K, N24952K, N25879K, N18455K, N18580K, N18647K	Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	-	-
TTN, ASI	rs562680371	E18191K, E20759K, E19118K, E11819K, E11694K, E11886K	not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN, ASI	rs56273463	R33738C, R31170C, R32097C, R24673C, R24865C, R24798C	Atrial fibrillation; Cardiomyopathy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, ASI	rs56341835	E25906K, E23338K, E24265K, E16841K, E16966K, E17033K	not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Tibial muscular dystrophy	Y	Y	-	-
TTN, ASI	rs56347248	I34199V, I31631V, I32558V, I25134V, I25259V, I25326V	Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiovascular phenotype	Y	Y	-	-
TTN, ASI	rs56372592	T23012K, T25580K, T23939K, T16515K, T16707K, T16640K	Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, ASI	rs56375087		Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, ASI	rs563969986	N18120S, N19761S, N17193S, N10696S, N10821S, N10888S	Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not specified	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, AS1	rs56399205	L21481P, L24049P, L22408P, L15176P, L15109P, L14984P	not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Ventricular tachycardia; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Cardiovascular phenotype; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, AS1	rs571093313	R22095W, R19527W, R13155W, R13222W, R20454W, R13030W	Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G	Y	-	-	Y
TTN, AS1	rs573415766	D22282G, D24850G, D23209G, D15977G, D15785G, D15910G	not provided; Tibial muscular dystrophy; not specified; Hypertrophic cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated Cardiomyopathy, Dominant; Limb-Girdle Muscular Dystrophy, Recessive; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, AS1	rs574660186	R22499*, R20858*, R19931*, R13626*, R13434*, R13559*	Primary dilated cardiomyopathy; Tibial muscular dystrophy; Familial hypertrophic cardiomyopathy 9; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided	Y	-	-	-
TTN, AS1	rs578191491	N27772I, N26131I, N25204I, N18832I, N18899I, N18707I	Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified	Y	-	-	-
TTN, AS1	rs587782987		not provided; Primary dilated cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, AS1	rs59148238	Y29293C, Y31861C, Y30220C, Y22921C, Y22796C, Y22988C	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; not provided; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, AS1	rs6706088		not specified; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN, AS1	rs72629785	R34455C, R31887C, R32814C, R25390C, R25515C, R25582C	not provided; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN, AS1	rs72629793	M35859T, M33291T, M34218T, M26986T, M26794T, M26919T	Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-
TTN, AS1	rs72646808	R17699H, R15131H, R16058H, R8634H, R8826H, R8759H	Dilated cardiomyopathy; Dilated cardiomyopathy 1G; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; not provided	Y	Y	-	-
TTN, AS1	rs72646823	K18518R, K15950R, K16877R, K9645R, K9453R, K9578R	Tibial muscular dystrophy; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; not provided; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype	Y	Y	-	-
TTN, AS1	rs72646842		Myopathy, early-onset, with fatal cardiomyopathy; not specified; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided	-	Y	-	-
TTN, AS1	rs72646848		Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Cardiovascular phenotype	Y	-	-	-
TTN, AS1	rs72646852		Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Cardiovascular phenotype	Y	-	-	-
TTN, AS1	rs72646853	A18579T, A21147T, A19506T, A12274T, A12207T, A12082T	Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not specified; not provided; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, ASI	rs72646855	I21197V, I18629V, I12257V, I19556V, I12132V, I12324V	Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Primary familial hypertrophic cardiomyopathy; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Cardiomyopathy; not provided; Tibial muscular dystrophy; Cardiovascular phenotype	Y	Y	-	-
TTN, ASI	rs72646859	R21392C, R18824C, R19751C, R12327C, R12452C, R12519C	Cardiomyopathy; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	-	-
TTN, ASI	rs72646864		Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, ASI	rs72646867		Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	Y	-	-
TTN, ASI	rs72646877		not specified; not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN, ASI	rs72646880	A22820P, A20252P, A21179P, A13755P, A13947P, A13880P	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified; not provided; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	Y	-	-
TTN, ASI	rs72646886		Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; not provided; Cardiomyopathy; not specified; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN, ASI	rs72646889		Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy	Y	-	-	-
TTN, ASI	rs72646890		Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	-	-	-
TTN, ASI	rs72646899	P24964L, P22396L, P23323L, P15899L, P16091L, P16024L	Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; not specified	Y	Y	-	-
TTN, ASI	rs72648205	G26298R, G23730R, G24657R, G17233R, G17425R, G17358R	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Premature ventricular contraction	Y	-	-	-
TTN, ASI	rs72648206	R26409H, R23841H, R24768H, R17344H, R17469H, R17536H	Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not provided; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, ASI	rs72648212		Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Cardiovascular phenotype; not provided; not specified; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, ASI	rs72648226		Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not provided; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, ASI	rs72648227	P29138T, P26570T, P27497T, P20073T, P20265T, P20198T	Cardiovascular phenotype; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J	Y	Y	-	-
TTN, ASI	rs72648237	V27228M, V29796M, V28155M, V20856M, V20731M, V20923M	not specified; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Hypertrophic cardiomyopathy; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, ASI	rs72648238	R29809Q, R27241Q, R28168Q, R20744Q, R20936Q, R20869Q	Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; not specified	Y	Y	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN, TTN-AS1	rs72648247	P30726S, P28158S, P29085S, P21661S, P21853S, P21786S	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated Cardiomyopathy, Dominant; Limb-Girdle Muscular Dystrophy, Recessive; Tibial muscular dystrophy; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Hypertrophic cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Supraventricular tachycardia; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; not provided; not specified	Y	Y	-	-
TTN, TTN-AS1	rs72648255		Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; not specified	Y	Y	-	-
TTN, TTN-AS1	rs72648259	I29189T, I31757T, I30116T, I22817T, I22692T, I22884T	Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; not provided	Y	-	-	-
TTN, TTN-AS1	rs72648263	A31885T, A29317T, A30244T, A22820T, A23012T, A22945T	Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not provided; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Ventricular fibrillation; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Tibial muscular dystrophy	Y	Y	-	-
TTN, TTN-AS1	rs72648266		Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not specified; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-
TTN, TTN-AS1	rs72648272	R32748C, R30180C, R31107C, R23683C, R23808C, R23875C	Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Dilated cardiomyopathy 1G; not provided; not specified; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure	Y	Y	-	-
TTN, TTN-AS1	rs72648273	A32765G, A30197G, A31124G, A23825G, A23892G, A23700G	Limb-girdle muscular dystrophy, type 2J; not provided; Tibial muscular dystrophy; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Hypertrophic cardiomyopathy	Y	Y	-	-
TTN, TTN-AS1	rs72648278	E33301K, E30733K, E31660K, E24236K, E24361K, E24428K	not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Hypertrophic cardiomyopathy; Cardiovascular phenotype; not provided	Y	-	-	-
TTN, TTN-AS1	rs72677232	V15750I, V13182I, V14109I, V6685I, V6810I, V6877I	Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy; Cardiovascular phenotype; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; not provided; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN, TTN-AS1	rs72677233	R15772Q, R13204Q, R14131Q, R6707Q, R6832Q, R6899Q	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Dilated cardiomyopathy; Atrial fibrillation; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; not specified	Y	Y	-	-
TTN, TTN-AS1	rs72677242	P16243S, P13675S, P14602S, P7303S, P7178S, P7370S	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Long QT syndrome; not provided; Cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; not specified	Y	Y	-	-
TTN, TTN-AS1	rs727503577	K19800N, K22368N, K13303N, K20727N, K13428N, K13495N	Tibial muscular dystrophy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	-	-	-
TTN, TTN-AS1	rs727503622	T13425N, T15993N, T14352N, T6928N, T7120N, T7053N	not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN, TTN-AS1	rs727504672	V28010I, V30578I, V28937I, V21513I, V21638I, V21705I	not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not specified; Tibial muscular dystrophy; Cardiomyopathy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, TTN-AS1	rs748292845		Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Myopathy, early-onset, with fatal cardiomyopathy; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, TTN-AS1	rs752204728	E15249A, E16890A, E7825A, E7950A, E14322A, E8017A	Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G	Y	-	-	-
TTN, TTN-AS1	rs753334568	P31732L, P29164L, P22792L, P22859L, P30091L, P22667L	Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; Myopathy, myofibrillar, 9, with early respiratory failure	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN-AS1, TTN	rs150430592	C30276G, C27708G, C28635G, C21211G, C21336G, C21403G	Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; Dilated cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Cardiovascular phenotype; not provided	Y	Y	-	-
TTN-AS1, TTN	rs181717727	G19020R, G21588R, G19947R, G12523R, G12648R, G12715R	Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy	Y	Y	-	-
TTN-AS1, TTN	rs182549226	D30534V, D27966V, D28893V, D21661V, D21469V, D21594V	Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not specified	Y	Y	-	-
TTN-AS1, TTN	rs191484894	S31766F, S29198F, S30125F, S22701F, S22826F, S22893F	not specified; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Cardiovascular phenotype; Dilated cardiomyopathy 1G; Dilated cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Myopathy, early-onset, with fatal cardiomyopathy	Y	Y	-	-
TTN-AS1, TTN	rs199615557	N17512K, N15871K, N8639K, N8447K, N8572K	Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; not provided	Y	-	-	-
TTN-AS1, TTN	rs200181804	F24187L, F26755L, F25114L, F17690L, F17882L, F17815L	Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided	Y	-	-	-
TTN-AS1, TTN	rs200463088		Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-
TTN-AS1, TTN	rs200732032	G19750S, G17182S, G18109S, G10685S, G10810S, G10877S	not provided; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN-AS1, TTN	rs200766837	K28700T, K31268T, K22328T, K22203T, K22395T, K29627T	Myopathy, early-onset, with fatal cardiomyopathy; Myopathy; not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN-AS1, TTN	rs200797552	E20245K, E22813K, E21172K, E13748K, E13873K, E13940K	not specified; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiovascular phenotype	Y	-	-	-
TTN-AS1, TTN	rs201562505	I24333V, I26901V, I25260V, I17836V, I17961V, I18028V	Dilated cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; TTN-Related Disorders; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; not specified; Cardiomyopathy; Myopathy, early-onset, with fatal cardiomyopathy; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN-AS1, TTN	rs368021072		Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; not provided; not specified; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN-AS1, TTN	rs368265962		not specified; not provided; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Dilated cardiomyopathy 1G	Y	-	-	-
TTN-AS1, TTN	rs372267046	R18625H, R21193H, R19552H, R12253H, R12320H, R12128H	not specified; Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN-AS1, TTN	rs374003257		not specified; not provided; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy	Y	-	-	-
TTN-AS1, TTN	rs377439315		not provided; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Myopathy, early-onset, with fatal cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiovascular phenotype; Dilated cardiomyopathy 1G; not specified	Y	Y	-	-
TTN-AS1, TTN	rs397517599	I16788T, I14220T, I15147T, I7915T, I7723T, I7848T	Tibial muscular dystrophy; not specified; Dilated cardiomyopathy 1G; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G	Y	-	-	-

Gene(s)	dbSNP ID	Protein change	Condition(s)	AD	MH	FT	23
TTN-AS1, TTN	rs55866005	V34854L, V32286L, V25981L, V33213L, V25789L, V25914L	Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; not provided; Cardiovascular phenotype; Myopathy, early-onset, with fatal cardiomyopathy; not specified; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Tibial muscular dystrophy; Dilated cardiomyopathy 1G	Y	Y	-	-
TTN-AS1, TTN	rs55886356	Q33922H, Q31354H, Q32281H, Q24982H, Q25049H, Q24857H	Limb-girdle muscular dystrophy, type 2J; not specified; Ventricular tachycardia; Cardiomyopathy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Cardiomyopathy; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; not provided; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	-	-
TTN-AS1, TTN	rs56130023	T21820I, T19252I, T20179I, T12755I, T12880I, T12947I	Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; not specified; not provided; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype	Y	Y	-	-
TTN-AS1, TTN	rs72646873	S22367P, S19799P, S20726P, S13302P, S13427P, S13494P	Dilated cardiomyopathy 1G; Tibial muscular dystrophy; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Myopathy, early-onset, with fatal cardiomyopathy; not provided; not specified; Cardiovascular phenotype; Cardiomyopathy; Brugada syndrome	Y	Y	-	-
TTN-AS1, TTN	rs72677231	R15731C, R13163C, R14090C, R6791C, R6666C, R6858C	Myopathy, myofibrillar, 9, with early respiratory failure; not specified; not provided; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Cardiomyopathy; Cardiovascular phenotype; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Tibial muscular dystrophy	Y	Y	-	-
TTN-AS1, TTN	rs72677243	I16318T, I13750T, I14677T, I7445T, I7253T, I7378T	Dilated cardiomyopathy 1G; not specified; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; Limb-girdle muscular dystrophy, type 2J; Cardiomyopathy; Myopathy, myofibrillar, 9, with early respiratory failure; Tibial muscular dystrophy; Myopathy, early-onset, with fatal cardiomyopathy; Cardiovascular phenotype; not provided	Y	Y	-	-
TTN-AS1, TTN	rs72677247		Tibial muscular dystrophy; Dilated cardiomyopathy 1G; Myopathy, myofibrillar, 9, with early respiratory failure; Limb-girdle muscular dystrophy, type 2J; Dilated cardiomyopathy 1G; not provided; not specified; Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J	Y	-	-	-
TTN-AS1, TTN	rs869320742	W31729C, W30088C, W29161C, W22664C, W22856C, W22789C	Myopathy, early-onset, with fatal cardiomyopathy; Limb-girdle muscular dystrophy, type 2J; Myopathy, myofibrillar, 9, with early respiratory failure; Dilated cardiomyopathy 1G; not provided; Tibial muscular dystrophy	Y	-	-	-