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Variant Report

Type to Filter

Curation

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Rare mutations

Below are rare variants (frequency less than 1%) that were submitted to ClinVar. The data presented has no guarantees of reporting accuracy. Heterozygous variants are reported as yellow and homozygous variants as red. A red or yellow variant does not necessarily mean one has or carries a condition or disease. This is for research and educational purposes only.

Gene: NF1
Variant: c.3732T>A (p.Val1244=)
rsID: rs756653022
Ref Allele: T
Alt Allele: A
Freq: 0.0008% rare
CADD: 10.47

ClinVar Submissions (4)

- Hereditary cancer-predisposing syndrome
- Neurofibromatosis, type 1, Autosomal dominant
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

TA

Gene: PDGFRB
Variant: c.164C>T (p.Ser55Leu)
rsID: rs147952898
Ref Allele: G
Alt Allele: A
Freq: 0.0024% rare
CADD: 14.74

ClinVar Submissions (1)

- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: SYNE1
Variant: c.9974G>A (p.Ser3325Asn)
rsID: rs746689638
Ref Allele: C
Alt Allele: T
Freq: 0.0032% rare
CADD: 20.7

ClinVar Submissions (1)

- not provided

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: COL5A1
Variant: c.2947G>A (p.Glu983Lys)
rsID: rs146348246
Ref Allele: G
Alt Allele: A
Freq: 0.0032% rare
CADD: 34

ClinVar Submissions (1)

- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: SPTAN1
Variant: c.6762+11G>A
rsID: rs775553506
Ref Allele: G
Alt Allele: A
Freq: 0.0048% rare
CADD: 0.111

ClinVar Submissions (1)

- not specified

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: CTC1
Variant: c.3247G>A (p.Glu1083Lys)
rsID: rs201885165
Ref Allele: C
Alt Allele: T
Freq: 0.0048% rare
CADD: 24.4

ClinVar Submissions (1)

- Dyskeratosis Congenita, Recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: COL18A1
Variant: c.2103C>T (p.Gly701=)
rsID: rs377652187
Ref Allele: C
Alt Allele: T
Freq: 0.0048% rare
CADD: 7.988

ClinVar Submissions (1)

- not specified

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: ANKH
Variant: c.*6205G>A
rsID: rs886060065
Ref Allele: C
Alt Allele: T
Freq: 0.008% rare
CADD: 0.289

ClinVar Submissions (1)

- Chondrocalcinosis
- Craniometaphyseal dysplasia, Autosomal dominant

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

Gene: CNGA3
Variant: c.*1240C>T
rsID: rs886056495
Ref Allele: C
Alt Allele: T
Freq: 0.0088% rare
CADD: 0.792

ClinVar Submissions (1)

- Achromatopsia

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: LRP2
Variant: c.10906C>T (p.Arg3636Trp)
rsID: rs747833963
Ref Allele: G
Alt Allele: A
Freq: 0.0088% rare
CADD: 22.5

ClinVar Submissions (1)

- Donnai Barrow syndrome, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: GRIN2B
Variant: c.*293G>A
rsID: rs199764468
Ref Allele: C
Alt Allele: T
Freq: 0.0104% rare
CADD: 7.507

ClinVar Submissions (1)

- Intellectual Disability, Dominant

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: SYNE1
Variant: c.15202A>G (p.Lys5068Glu)
rsID: rs139805184
Ref Allele: T
Alt Allele: C
Freq: 0.0143% rare
CADD: 15.9

ClinVar Submissions (2)

- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

TC

Gene: IMPAD1
Variant: c.*5142G>T
rsID: rs777098357
Ref Allele: C
Alt Allele: A
Freq: 0.0143% rare
CADD: 0.189

ClinVar Submissions (1)

- Chondrodysplasia

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CA

Gene: SUCLA2
Variant: c.1076A>C
(p.Glu359Ala)
rsID: rs150996983
Ref Allele: T
Alt Allele: G
Freq: 0.0175% rare
CADD: 23.8

ClinVar Submissions (1)

- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

TG

Gene: CACNA2D2
Variant: c.2295C>T
(p.Asn765=)
rsID: rs139125287
Ref Allele: G
Alt Allele: A
Freq: 0.0231% rare
CADD: 22.7

ClinVar Submissions (1)

- Early infantile epileptic encephalopathy

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: UROS
Variant: c.217T>C
(p.Cys73Arg)
rsID: rs121908012
Ref Allele: A
Alt Allele: G
Freq: 0.0247% rare
CADD: 23.6

ClinVar Submissions (4)

- Congenital erythropoietic porphyria, Autosomal recessive
- not provided

Clinically Significant Pathogenic

LitVar SNPedia dbSNP UniProt OMIM OpenSNP PubMed (10)

Hetero

AG

Gene: PEX16
Variant: c.877C>T
(p.Arg293Cys)
rsID: rs544053792
Ref Allele: G
Alt Allele: A
Freq: 0.0271% rare
CADD: 26.6

ClinVar Submissions (1)

- not provided

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: ARFGEF2
Variant: c.*141G>A
rsID: rs200664908
Ref Allele: G
Alt Allele: A
Freq: 0.0334% rare
CADD: 0.756

ClinVar Submissions (1)

- Periventricular Heterotopia

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: ADAMTS17
Variant: c.*2690G>A
rsID: rs566216613
Ref Allele: C
Alt Allele: T
Freq: 0.0414% rare

ClinVar Submissions (1)

- Weill-Marchesani-like syndrome, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

CADD: 3.482

Gene: RP1L1
Variant: c.954C>T
(p.Asp318=)
rsID: rs200317816
Ref Allele: G
Alt Allele: A
Freq: 0.043% rare
CADD: 1.004

ClinVar Submissions (1)

- Occult macular dystrophy, Autosomal dominant

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: IGF1R
Variant: c.-43T>C
rsID: rs768733704
Ref Allele: T
Alt Allele: C
Freq: 0.0518% rare
CADD: 20.4

ClinVar Submissions (1)

- Insulin-like growth factor 1 resistance to, Autosomal dominant

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Homo

CC

Gene: ALMS1
Variant: c.11356A>G
(p.Ile3786Val)
rsID: rs201819880
Ref Allele: A
Alt Allele: G
Freq: 0.0542% rare
CADD: 22.9

ClinVar Submissions (2)

- Alstrom syndrome, Autosomal recessive
- not provided

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

AG

Gene: MYOM1
Variant: c.4903G>A
(p.Gly1635Ser)
rsID: rs200773357
Ref Allele: C
Alt Allele: T
Freq: 0.0557% rare
CADD: 13.28

ClinVar Submissions (1)

- Hypertrophic cardiomyopathy

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: COL1A1
Variant: c.1461+13G>T
rsID: rs371161009
Ref Allele: C
Alt Allele: A
Freq: 0.0589% rare
CADD: 7.086

ClinVar Submissions (1)

- not specified

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CA

Gene: COL6A1
Variant: c.1350G>A
(p.Pro450=)
rsID: rs144887329
Ref Allele: G
Alt Allele: A
Freq: 0.0597% rare
CADD: 0.683

ClinVar Submissions (3)

- Bethlem myopathy 1
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: NHS
Variant: c.3866G>T
(p.Gly1289Val)
rsID: rs41304731
Ref Allele: G
Alt Allele: T

ClinVar Submissions (3)

- Nance-Horan syndrome, X-linked dominant
- not specified

Conflicting/Uncertain

Homo

TT

Freq: 0.0661% **rare**

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (2)

Gene: TRAPPC11
Variant: c.2147C>G (p.Ala716Gly)
rsID: rs143990563
Ref Allele: C
Alt Allele: G
Freq: 0.0685% **rare**
CADD: 20.7

ClinVar Submissions (3)

- Limb-girdle muscular dystrophy, type 2S
- not specified

Clinically Significant **Conflicting/Uncertain**

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CG

Gene: BRCA1
Variant: c.441+36_441+50delCTTTTCTTTTTTTTT
rsID: rs1168113546
Ref Allele: AAAAAAAAAGAAAAG
Alt Allele: A
Freq: 0.0749% **rare**

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

0|1

Gene: LHX3
Variant: c.944G>C (p.Arg315Pro)
rsID: rs201591640
Ref Allele: C
Alt Allele: G
Freq: 0.0765% **rare**
CADD: 21.9

ClinVar Submissions (2)

- Combined Pituitary Hormone Deficiency, Recessive
- not provided

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CG

Gene: SERAC1
Variant: c.*16A>G
rsID: rs199625765
Ref Allele: T
Alt Allele: C
Freq: 0.0788% **rare**
CADD: 0.901

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

TC

Gene: SELENON
Variant: c.*1985A>T
rsID: rs535082643
Ref Allele: A
Alt Allele: T
Freq: 0.0876% **rare**
CADD: 1.072

ClinVar Submissions (1)

- SEPN1-Related Disorders

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

AT

Gene: FLNC
Variant: c.7947C>T (p.Phe2649=)
rsID: rs368849358
Ref Allele: C
Alt Allele: T
Freq: 0.09% rare
CADD: 4.876

ClinVar Submissions (4)

- Cardiomyopathy, familial hypertrophic, 26, Autosomal dominant
- Cardiomyopathy, familial hypertrophic, 26, Autosomal dominant
- Dilated Cardiomyopathy, Dominant
- Hypertrophic cardiomyopathy
- Myofibrillar myopathy, filamin C-related, Autosomal dominant
- Myofibrillar myopathy, filamin C-related, Autosomal dominant
- Myopathy, distal, 4, Autosomal dominant
- Myopathy, distal, 4, Autosomal dominant
- Primary dilated cardiomyopathy
- not specified

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

CT

Gene: CACNA2D2
Variant: c.2995C>T (p.Pro999Ser)
rsID: rs146587089
Ref Allele: G
Alt Allele: A
Freq: 0.0964% rare
CADD: 16.93

ClinVar Submissions (1)

- Early infantile epileptic encephalopathy

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: SYNJ1
Variant: c.2237T>C (p.Ile746Thr)
rsID: rs147929290
Ref Allele: A
Alt Allele: G
Freq: 0.0964% rare
CADD: 27.2

ClinVar Submissions (1)

- Epileptic encephalopathy, early infantile, 53, Autosomal recessive
- Parkinson disease 20, early-onset, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

AG

Gene: TSPAN12
Variant: c.*334A>T
rsID: rs545129654
Ref Allele: T
Alt Allele: A
Freq: 0.0988% rare
CADD: 1.658

ClinVar Submissions (1)

- Familial exudative vitreoretinopathy

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

TA

Gene: DOCK3
Variant: c.5020A>T (p.Met1674Leu)
rsID: rs142515812
Ref Allele: A
Alt Allele: T
Freq: 0.1043% rare
CADD: 15.67

ClinVar Submissions (1)

- not provided

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

AT

Gene: RPE65
Variant: c.394G>A (p.Ala132Thr)
rsID: rs61752878
Ref Allele: C
Alt Allele: T
Freq: 0.1075% rare
CADD: 20.8

ClinVar Submissions (4)

- Leber congenital amaurosis 2, Autosomal recessive
- Retinitis pigmentosa 20, Autosomal recessive
- Retinitis pigmentosa 20, Autosomal recessive
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP UniProt OMIM OpenSNP PubMed (3)

Hetero

CT

Gene: LAMC2
Variant: c.3206C>T (p.Thr1069Met)
rsID: rs139043074
Ref Allele: C
Alt Allele: T
Freq: 0.1075% rare
CADD: 0.164

ClinVar Submissions (4)

- Epidermolysis bullosa, junctional
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: CACNA2D2
Variant: c.3402A>C (p.Gln1134His)
rsID: rs150284749
Ref Allele: T
Alt Allele: G
Freq: 0.1075% rare
CADD: 23.2

ClinVar Submissions (1)

- Early infantile epileptic encephalopathy

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

TG

Gene: SMC1A
Variant: c.*2609A>C
rsID: rs192734396
Ref Allele: T
Alt Allele: G
Freq: 0.1075% rare

ClinVar Submissions (1)

- Cornelia de Lange Syndrome

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Homo

GG

Gene: MARVELD2
Variant: c.1407C>T (p.Tyr469=)
rsID: rs61736168
Ref Allele: C
Alt Allele: T
Freq: 0.1107% rare
CADD: 9.384

ClinVar Submissions (4)

- Nonsyndromic Hearing Loss, Recessive
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

CT

Gene: GCK
Variant: c.208+11G>A
rsID: rs77440690
Ref Allele: C
Alt Allele: T
Freq: 0.1226% rare
CADD: 1.039

ClinVar Submissions (4)

- Maturity onset diabetes mellitus in young, Autosomal dominant
- Maturity-onset diabetes of the young, type 2
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (8)

Hetero

CT

Gene: INF2
Variant: c.3134G>A (p.Arg1045Gln)

ClinVar Submissions (2)

- Charcot-Marie-Tooth disease, dominant intermediate E, Autosomal dominant
- Focal segmental glomerulosclerosis

Hetero

rsID: rs200369827
Ref Allele: G
Alt Allele: A
Freq: 0.1226% rare
CADD: 0.005

- Focal segmental glomerulosclerosis 5

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

GA

Gene: FBN2
Variant: c.8364+7A>T
rsID: rs185052980
Ref Allele: T
Alt Allele: A
Freq: 0.1282% rare
CADD: 0.51

ClinVar Submissions (9)

- Congenital contractural arachnodactyly, Autosomal dominant
- Connective tissue disorder
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

TA

Gene: GLI3
Variant: c.680-25C>T
rsID: rs55872291
Ref Allele: G
Alt Allele: A
Freq: 0.1306% rare
CADD: 0.169

ClinVar Submissions (1)

- not specified

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

GA

Gene: MYO5B
Variant: c.3828C>T
(p.Leu1276=)
rsID: rs201760142
Ref Allele: G
Alt Allele: A
Freq: 0.1402% rare
CADD: 0.203

ClinVar Submissions (1)

- Diarrhea with Microvillus Atrophy

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: SLC34A3
Variant: c.1585A>T
(p.Ile529Phe)
rsID: rs140639805
Ref Allele: A
Alt Allele: T
Freq: 0.1537% rare
CADD: 0.678

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

AT

Gene: RAB3GAP1
Variant: c.*404C>T
rsID: rs149483456
Ref Allele: C
Alt Allele: T
Freq: 0.1649% rare
CADD: 6.6

ClinVar Submissions (1)

- Warburg micro syndrome

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: MMP9
Variant: c.886G>A
(p.Gly296Ser)
rsID: rs144098289
Ref Allele: G
Alt Allele: A
Freq: 0.1776% rare
CADD: 25.7

ClinVar Submissions (1)

- not provided

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: COL4A2

ClinVar Submissions (1)

Hetero

Variant: c.3326G>A
(p.Arg1109Gln)
rsID: rs184812559
Ref Allele: G
Alt Allele: A
Freq: 0.1872% rare
CADD: 3.403

- Porencephalic cyst

Likely benign

LitVar SNPedia dbSNP UniProt OpenSNP PubMed

GA

Gene: PCNT
Variant: c.3748C>T
(p.Arg1250Trp)
rsID: rs117987006
Ref Allele: C
Alt Allele: T
Freq: 0.1872% rare
CADD: 20.7

ClinVar Submissions (4)

- Microcephalic Osteodysplastic Primordial Dwarfism
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

CT

Gene: RYR2
Variant: c.2760G>A
(p.Glu920=)
rsID: rs186181155
Ref Allele: G
Alt Allele: A
Freq: 0.2015% rare
CADD: 4.369

ClinVar Submissions (5)

- Arrhythmogenic right ventricular cardiomyopathy
- Cardiovascular phenotype
- Catecholaminergic polymorphic ventricular tachycardia, Autosomal dominant
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

GA

Gene: FTCD
Variant: c.643C>T
(p.Arg215Cys)
rsID: rs149667449
Ref Allele: G
Alt Allele: A
Freq: 0.2071% rare
CADD: 24.3

ClinVar Submissions (1)

- GLUTAMATE FORMIMINOTRANSFERASE DEFICIENCY, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: ESCO2
Variant: c.1094G>A
(p.Arg365Lys)
rsID: rs144288263
Ref Allele: G
Alt Allele: A
Freq: 0.2094% rare
CADD: 15.62

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: HMCN1
Variant: c.15256+4A>T
rsID: rs184102616
Ref Allele: A
Alt Allele: T
Freq: 0.223% rare
CADD: 10.13

ClinVar Submissions (1)

- Macular degeneration

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

AT

Gene: TBC1D24
Variant: c.1482G>A
(p.Ala494=)
rsID: rs201059992
Ref Allele: G
Alt Allele: A

ClinVar Submissions (6)

- Caused by mutation in the TBC1 domain family, member 24
- Deafness, autosomal dominant 65, Autosomal dominant
- Epileptic encephalopathy, early infantile, 1, X-linked recessive
- Seizures
- not specified

Hetero

GA

Freq: 0.223% rare
CADD: 0.014

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Gene: F12;SLC34A1
Variant: c.*179G>A
rsID: rs141664220
Ref Allele: G
Alt Allele: A
Freq: 0.2238% rare
CADD: 0.432

ClinVar Submissions (1)

- Factor XII deficiency disease, Autosomal recessive
- Hereditary Angioedema
- Hypophosphatemic Nephrolithiasis/Osteoporosis

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: DOLK
Variant: c.631C>T
(p.Arg211Cys)
rsID: rs145310298
Ref Allele: G
Alt Allele: A
Freq: 0.2246% rare
CADD: 23.1

ClinVar Submissions (5)

- Congenital disorder of glycosylation
- Congenital disorder of glycosylation type 1M, Autosomal recessive
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (2)

Hetero

GA

Gene: HFM1
Variant: c.2308G>A
(p.Asp770Asn)
rsID: rs143399622
Ref Allele: C
Alt Allele: T
Freq: 0.2278% rare
CADD: 25.7

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

CT

Gene: TTN
Variant: c.54218G>A
(p.Arg18073Gln)
rsID: rs199895260
Ref Allele: C
Alt Allele: T
Freq: 0.2278% rare
CADD: 25.9

ClinVar Submissions (7)

- Cardiovascular phenotype
- Dilated Cardiomyopathy, Dominant
- Dilated cardiomyopathy 1G, Autosomal dominant
- Distal myopathy Markesbery-Griggs type, Autosomal dominant
- Hereditary myopathy with early respiratory failure, Autosomal dominant
- Hypertrophic cardiomyopathy
- Limb-Girdle Muscular Dystrophy, Recessive
- Limb-girdle muscular dystrophy, type 2J, Autosomal recessive
- Myopathy, early-onset, with fatal cardiomyopathy, Autosomal recessive
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (2)

Hetero

CT

Gene: WDR35
Variant: c.355C>T
(p.Arg119Cys)
rsID: rs140308808
Ref Allele: G
Alt Allele: A
Freq: 0.2302% rare
CADD: 24.9

ClinVar Submissions (4)

- Cranioectodermal dysplasia
- Short Rib Polydactyly Syndrome
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

GA

Gene: ACVR2B
Variant: c.*8812C>T
rsID: rs569525244
Ref Allele: C
Alt Allele: T
Freq: 0.2333% rare
CADD: 16.16

ClinVar Submissions (1)

- Heterotaxia

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: SLC34A1
Variant: c.398C>T
(p.Ala133Val)
rsID: rs148976897
Ref Allele: C
Alt Allele: T
Freq: 0.2365% rare
CADD: 26.3

ClinVar Submissions (2)

- Hypophosphatemic Nephrolithiasis/Osteoporosis
- Nephrocalcinosis

Clinically Significant Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

CT

Gene: TTN
Variant: c.90590C>G
(p.Ala30197Gly)
rsID: rs72648273
Ref Allele: G
Alt Allele: C
Freq: 0.2381% rare
CADD: 27.4

ClinVar Submissions (9)

- Cardiovascular phenotype
- Dilated Cardiomyopathy, Dominant
- Dilated cardiomyopathy 1G, Autosomal dominant
- Distal myopathy Markesbery-Griggs type, Autosomal dominant
- Hereditary myopathy with early respiratory failure, Autosomal dominant
- Hypertrophic cardiomyopathy
- Limb-Girdle Muscular Dystrophy, Recessive
- Limb-girdle muscular dystrophy, type 2J, Autosomal recessive
- Myopathy, early-onset, with fatal cardiomyopathy, Autosomal recessive
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (3)

Hetero

GC

Gene: TTN
Variant: c.16160G>A
(p.Cys5387Tyr)
rsID: rs72648913
Ref Allele: C
Alt Allele: T
Freq: 0.2461% rare
CADD: 18.68

ClinVar Submissions (4)

- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed (2)

Hetero

CT

Gene: ATXN10
Variant: c.321G>A
(p.Thr107=)
rsID: rs61733598
Ref Allele: G
Alt Allele: A
Freq: 0.2501% rare
CADD: 0.04

ClinVar Submissions (1)

- not specified

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: CDH23
Variant: c.1307G>A (p.Ser436Asn)
rsID: rs111033369
Ref Allele: G
Alt Allele: A
Freq: 0.2525% rare
CADD: 24.9

ClinVar Submissions (5)

- Nonsyndromic Hearing Loss, Recessive
- Retinitis pigmentosa-deafness syndrome
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (3)

Hetero

GA

Gene: SCN9A
Variant: c.*124A>G
rsID: rs201137748
Ref Allele: T
Alt Allele: C
Freq: 0.2676% rare
CADD: 6.784

ClinVar Submissions (1)

- Congenital Indifference to Pain
- Familial Febrile Seizures
- Generalized epilepsy with febrile seizures plus
- Inherited Erythromelalgia
- Paroxysmal extreme pain disorder, Autosomal dominant
- Severe myoclonic epilepsy in infancy, Autosomal dominant
- Small fiber neuropathy

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

TC

Gene: ADAMTS2
Variant: c.*2854T>C
rsID: rs11740156
Ref Allele: A
Alt Allele: G
Freq: 0.27% rare
CADD: 2.967

ClinVar Submissions (1)

- Ehlers-Danlos syndrome, type vii, autosomal recessive, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

AG

Gene: SERPINE1
Variant: c.*950C>T
rsID: rs536144892
Ref Allele: C
Alt Allele: T
Freq: 0.2779% rare
CADD: 1.78

ClinVar Submissions (1)

- Plasminogen activator inhibitor type 1 deficiency, Autosomal dominant

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

Gene: PKD1
Variant: c.8087T>G (p.Leu2696Arg)
rsID: rs201238819
Ref Allele: A
Alt Allele: C
Freq: 0.2803% rare
CADD: 13

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed (5)

Hetero

AC

Gene: RPS6KA3
Variant: c.213A>G (p.Leu71=)
rsID: rs56338023
Ref Allele: T
Alt Allele: C
Freq: 0.2827% rare

ClinVar Submissions (3)

- History of neurodevelopmental disorder
- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Homo

CC

Gene: SHANK3
Variant: c.4960C>A

ClinVar Submissions (2)

Hetero

(p.Pro1654Thr)
rsID: rs749130556
Ref Allele: C
Alt Allele: A
Freq: 0.2843% rare
CADD: 17.26

- History of neurodevelopmental disorder
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed (1)

CA

Gene: ZNF469
Variant: c.2841G>A
(p.Arg947=)
rsID: rs150435442
Ref Allele: G
Alt Allele: A
Freq: 0.2851% rare
CADD: 8.822

ClinVar Submissions (2)

Hetero

- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

GA

Gene: FLNC
Variant: c.2078A>C
(p.Asp693Ala)
rsID: rs34972246
Ref Allele: A
Alt Allele: C
Freq: 0.2899% rare
CADD: 24.3

ClinVar Submissions (4)

Hetero

- Cardiomyopathy, familial hypertrophic, 26, Autosomal dominant
- Dilated Cardiomyopathy, Dominant
- Myofibrillar myopathy, filamin C-related, Autosomal dominant
- Myopathy, distal, 4, Autosomal dominant
- not specified

Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

AC

Gene: SUOX
Variant: c.629C>T
(p.Pro210Leu)
rsID: rs141735896
Ref Allele: C
Alt Allele: T
Freq: 0.3058% rare
CADD: 25.9

ClinVar Submissions (1)

Hetero

- Sulfite oxidase deficiency, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

CT

Gene: DNAH1
Variant: c.5853G>A
(p.Val1951=)
rsID: rs80128076
Ref Allele: G
Alt Allele: A
Freq: 0.3098% rare
CADD: 12.3

ClinVar Submissions (1)

Hetero

- Ciliary dyskinesia, primary, 37, Autosomal recessive
- SPERMATOGENIC FAILURE 18, Autosomal recessive

Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

GA

Gene: NTRK1
Variant: c.16C>T
(p.Arg6Trp)
rsID: rs201472270
Ref Allele: C
Alt Allele: T
Freq: 0.317% rare
CADD: 23.4

ClinVar Submissions (7)

Hetero

- Familial medullary thyroid carcinoma, Autosomal dominant
- Hereditary insensitivity to pain with anhidrosis, Autosomal recessive
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP UniProt OMIM OpenSNP PubMed

CT

Gene: CBL
Variant: c.*1918C>T
rsID: rs528450894
Ref Allele: C
Alt Allele: T
Freq: 0.3353% rare
CADD: 2.361

ClinVar Submissions (1)

- Noonan-Like Syndrome Disorder

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: IL10RB
Variant: c.-72C>A
rsID: rs45607743
Ref Allele: C
Alt Allele: A
Freq: 0.3361% rare
CADD: 8.23

ClinVar Submissions (1)

- Inflammatory bowel disease

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CA

Gene: MUTYH
Variant: c.1187G>A
(p.Gly396Asp)
rsID: rs36053993
Ref Allele: C
Alt Allele: T
Freq: 0.3393% rare
CADD: 29.4

ClinVar Submissions (31)

- Carcinoma of colon
- Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas
- Endometrial carcinoma
- Hereditary cancer-predisposing syndrome
- MUTYH-associated polyposis
- MYH-associated polyposis, Autosomal dominant
- MYH-associated polyposis, Autosomal dominant
- MYH-associated polyposis, Autosomal dominant
- Neoplasm of stomach
- Neoplasm of stomach
- Pilomatrixoma, Autosomal dominant
- Small intestine carcinoid
- not provided
- not specified

Expert Reviewed Clinically Significant Pathogenic/Likely pathogenic

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (40)

Hetero

CT

Gene: HOMER2
Variant: c.797A>G
(p.Glu266Gly)
rsID: rs79448007
Ref Allele: T
Alt Allele: C
Freq: 0.3424% rare
CADD: 34

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

TC

Gene: NEUROD1
Variant: c.*78C>T
rsID: rs41270211
Ref Allele: G
Alt Allele: A
Freq: 0.3432% rare
CADD: 19.4

ClinVar Submissions (1)

- Maturity onset diabetes mellitus in young, Autosomal dominant

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: COL4A4
Variant: c.2630G>A
(p.Arg877Gln)
rsID: rs150979437
Ref Allele: C
Alt Allele: T

ClinVar Submissions (3)

- Alport syndrome
- not specified

Conflicting/Uncertain

Hetero

CT

Freq: 0.344% rare
CADD: 0.305

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Gene: ADAMTS2
Variant: c.*1639C>T
rsID: rs138971097
Ref Allele: G
Alt Allele: A
Freq: 0.3512% rare
CADD: 3.537

ClinVar Submissions (1)

- Ehlers-Danlos syndrome, type vii, autosomal recessive, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: RGR
Variant: c.744+5A>G
rsID: rs143720091
Ref Allele: A
Alt Allele: G
Freq: 0.3512% rare
CADD: 0.125

ClinVar Submissions (2)

- Retinitis Pigmentosa, Recessive
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

AG

Gene: LAMA2
Variant: c.1634T>A
(p.Leu545Gln)
rsID: rs118083923
Ref Allele: T
Alt Allele: A
Freq: 0.3608% rare
CADD: 26.4

ClinVar Submissions (6)

- Laminin alpha 2-related dystrophy
- not provided
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP UniProt OpenSNP PubMed (2)

Hetero

TA

Gene: APC
Variant: c.-18-871T>C
rsID: rs115198624
Ref Allele: T
Alt Allele: C
Freq: 0.3703% rare
CADD: 1.588

ClinVar Submissions (1)

- Hereditary cancer-predisposing syndrome

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

TC

Gene: MCPH1
Variant: c.477A>T
(p.Ser159=)
rsID: rs41313948
Ref Allele: A
Alt Allele: T
Freq: 0.3743% rare
CADD: 1.172

ClinVar Submissions (4)

- Primary Microcephaly, Recessive
- Primary autosomal recessive microcephaly 1, Autosomal recessive
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

AT

Gene: INSR
Variant: c.*501T>G
rsID: rs562228680
Ref Allele: A
Alt Allele: C
Freq: 0.3791% rare
CADD: 1.371

ClinVar Submissions (1)

- Insulin-resistant diabetes mellitus AND acanthosis nigricans
- Leprechaunism syndrome, Autosomal recessive
- Pineal hyperplasia AND diabetes mellitus syndrome, Autosomal recessive

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

AC

Gene: SEPT9
Variant: c.1520-10C>T
rsID: rs192537441
Ref Allele: C
Alt Allele: T
Freq: 0.3831% rare
CADD: 0.762

ClinVar Submissions (1)

- Hereditary Neuralgic Amyotrophy (HNA)

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: TBC1D4
Variant: c.2254A>G
(p.Thr752Ala)
rsID: rs149821147
Ref Allele: T
Alt Allele: C
Freq: 0.3878% rare
CADD: 9.77

ClinVar Submissions (1)

- not specified

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

TC

Gene: BRCA2
Variant: c.9256+3032G>A
rsID: rs150042254
Ref Allele: G
Alt Allele: A
Freq: 0.3894% rare
CADD: 3.459

ClinVar Submissions (1)

- Breast-ovarian cancer, familial 2, Autosomal dominant

Expert Reviewed Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: ARSI
Variant: c.87C>T
(p.Ala29=)
rsID: rs200012526
Ref Allele: G
Alt Allele: A
Freq: 0.3998% rare
CADD: 10.62

ClinVar Submissions (1)

- Spastic paraplegia

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: ARHGEF28
Variant: c.1862G>A
(p.Arg621Gln)
rsID: rs115243197
Ref Allele: G
Alt Allele: A
Freq: 0.4149% rare
CADD: 33

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

GA

Gene: TTN
Variant: c.99083C>T
(p.Thr33028Ile)
rsID: rs55842557
Ref Allele: G
Alt Allele: A
Freq: 0.4205% rare
CADD: 22.7

ClinVar Submissions (8)

- Cardiovascular phenotype
- Dilated Cardiomyopathy, Dominant
- Dilated cardiomyopathy 1G, Autosomal dominant
- Distal myopathy Markesbery-Griggs type, Autosomal dominant
- Hereditary myopathy with early respiratory failure, Autosomal dominant
- Hypertrophic cardiomyopathy
- Limb-Girdle Muscular Dystrophy, Recessive
- Limb-girdle muscular dystrophy, type 2J, Autosomal recessive
- Myopathy, early-onset, with fatal cardiomyopathy, Autosomal recessive
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (3)

Hetero

GA

Gene: TTN
Variant: c.99846A>C
(p.Thr33282=)
rsID: rs56375087
Ref Allele: T
Alt Allele: G
Freq: 0.4205% rare
CADD: 14.79

ClinVar Submissions (7)

- Cardiovascular phenotype
- Dilated Cardiomyopathy, Dominant
- Dilated cardiomyopathy 1G, Autosomal dominant
- Distal myopathy Markesbery-Griggs type, Autosomal dominant
- Hereditary myopathy with early respiratory failure, Autosomal dominant
- Hypertrophic cardiomyopathy
- Limb-Girdle Muscular Dystrophy, Recessive
- Limb-girdle muscular dystrophy, type 2J, Autosomal recessive
- Myopathy, early-onset, with fatal cardiomyopathy, Autosomal recessive
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (2)

Hetero

TG

Gene: EP300
Variant: c.2091T>G
(p.Ser697Arg)
rsID: rs61756764
Ref Allele: T
Alt Allele: G
Freq: 0.4213% rare
CADD: 25.5

ClinVar Submissions (4)

- Carcinoma of colon
- Rubinstein-Taybi syndrome 1, Autosomal dominant
- Rubinstein-Taybi syndrome 2, Autosomal dominant
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

TG

Gene: ADGRV1
Variant: c.5830G>A
(p.Asp1944Asn)
rsID: rs41302834
Ref Allele: G
Alt Allele: A
Freq: 0.4229% rare
CADD: 27.2

ClinVar Submissions (4)

- not provided
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (2)

Hetero

GA

Gene: TECPR2
Variant: c.3275C>T
(p.Ser1092Leu)
rsID: rs72700618
Ref Allele: C
Alt Allele: T
Freq: 0.4237% rare
CADD: 27.7

ClinVar Submissions (2)

- Spastic paraplegia 49, autosomal recessive, Autosomal recessive
- not specified

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

Gene: GLDC
Variant: c.1707+8G>A
rsID: rs144666843
Ref Allele: C
Alt Allele: T
Freq: 0.4253% rare
CADD: 6.698

ClinVar Submissions (2)

- Non-ketotic hyperglycinemia, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

Gene: TTN
Variant: c.4034G>A
(p.Gly1345Asp)
rsID: rs36021856
Ref Allele: C
Alt Allele: T
Freq: 0.4261% rare

ClinVar Submissions (8)

- Cardiovascular phenotype
- Dilated Cardiomyopathy, Dominant
- Dilated cardiomyopathy 1G, Autosomal dominant
- Distal myopathy Markesbery-Griggs type, Autosomal dominant
- Hereditary myopathy with early respiratory failure, Autosomal dominant

Hetero

CT

CADD: 24.5

- Hypertrophic cardiomyopathy
- Limb-Girdle Muscular Dystrophy, Recessive
- Limb-girdle muscular dystrophy, type 2J, Autosomal recessive
- Myopathy, early-onset, with fatal cardiomyopathy, Autosomal recessive
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP UniProt OMIM OpenSNP PubMed (3)

Gene: FGFR3
Variant: c.1150T>C (p.Phe384Leu)
rsID: rs17881656
Ref Allele: T
Alt Allele: C
Freq: 0.4261% rare
CADD: 16.82

ClinVar Submissions (8)

- Craniosynostosis
- not provided
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP UniProt OpenSNP PubMed (3)

Hetero

TC

Gene: LRPPRC
Variant: c.*343C>T
rsID: rs144519599
Ref Allele: G
Alt Allele: A
Freq: 0.4269% rare
CADD: 4.816

ClinVar Submissions (1)

- Leigh syndrome, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: MED13L
Variant: c.1863T>C (p.Ile621=)
rsID: rs61748071
Ref Allele: A
Alt Allele: G
Freq: 0.4316% rare
CADD: 10.4

ClinVar Submissions (1)

- Transposition of the great arteries, dextro-looped 1, Autosomal dominant

Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

AG

Gene: FUS
Variant: c.*1992A>C
rsID: rs146490489
Ref Allele: A
Alt Allele: C
Freq: 0.4364% rare
CADD: 1.491

ClinVar Submissions (1)

- Amyotrophic Lateral Sclerosis, Dominant

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

AC

Gene: CDKN1B
Variant: c.-202C>T
rsID: rs183710253
Ref Allele: C
Alt Allele: T
Freq: 0.438% rare
CADD: 9.568

ClinVar Submissions (1)

- Multiple endocrine neoplasia

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

CT

Gene: CERKL
Variant: c.-63C>T
rsID: rs140924460
Ref Allele: G
Alt Allele: A
Freq: 0.4388% rare
CADD: 4.385

ClinVar Submissions (1)

- Retinitis Pigmentosa, Recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: ANO3
Variant: c.164C>T
(p.Ser55Phe)
rsID: rs61746297
Ref Allele: C
Alt Allele: T
Freq: 0.4539% rare
CADD: 24.3

ClinVar Submissions (1)

- Dystonia

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: INVS
Variant: c.725C>T
(p.Ser242Leu)
rsID: rs2491097
Ref Allele: C
Alt Allele: T
Freq: 0.4715% rare
CADD: 26.4

ClinVar Submissions (3)

- Nephronophthisis
- not specified

Benign

LitVar SNPedia dbSNP UniProt OpenSNP PubMed (2)

Hetero

CT

Gene: TTC37
Variant: c.4187A>G
(p.Asn1396Ser)
rsID: rs116690692
Ref Allele: T
Alt Allele: C
Freq: 0.4723% rare
CADD: 14.17

ClinVar Submissions (2)

- not provided
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

TC

Gene: MYH2
Variant: c.5780G>A
(p.Arg1927Gln)
rsID: rs34161789
Ref Allele: C
Alt Allele: T
Freq: 0.481% rare
CADD: 29.4

ClinVar Submissions (4)

- Inclusion body myopathy 3, Autosomal dominant
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP UniProt OMIM OpenSNP PubMed (1)

Hetero

CT

Gene: FAT4
Variant: c.6219A>G
(p.Gln2073=)
rsID: rs35355603
Ref Allele: A
Alt Allele: G
Freq: 0.485% rare
CADD: 5.689

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

AG

Gene: RAD50
Variant: c.572C>T
(p.Thr191Ile)
rsID: rs2230017
Ref Allele: C
Alt Allele: T
Freq: 0.493% rare
CADD: 23.9

ClinVar Submissions (7)

- Hereditary cancer-predisposing syndrome
- Nijmegen breakage syndrome-like disorder, Autosomal recessive
- not provided
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP UniProt OMIM OpenSNP PubMed (2)

Hetero

CT

Gene: POLG
Variant: c.3198G>A
(p.Thr1066=)

ClinVar Submissions (10)

- POLG-Related Spectrum Disorders
- Progressive sclerosing poliodystrophy, Autosomal recessive

Hetero

rsID: rs61752780
Ref Allele: C
Alt Allele: T
Freq: 0.5025% rare
CADD: 8.288

- Seizures
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

CT

Gene: CNGA3
Variant: c.1569C>T (p.Asn523=)
rsID: rs61752503
Ref Allele: C
Alt Allele: T
Freq: 0.5033% rare
CADD: 0.751

ClinVar Submissions (1)

- not specified

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: FBN1
Variant: c.*1580G>A
rsID: rs17352989
Ref Allele: C
Alt Allele: T
Freq: 0.5041% rare
CADD: 0.338

ClinVar Submissions (1)

- Acromicric dysplasia, Autosomal dominant
- Ectopia lentis
- Geleophysic dysplasia
- MASS syndrome, Autosomal dominant
- Marfan syndrome, Autosomal dominant
- Stiff skin syndrome, Autosomal dominant
- Thoracic aortic aneurysm and aortic dissection
- Weill-Marchesani syndrome

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

Gene: LPL
Variant: c.*949G>A
rsID: rs11570893
Ref Allele: G
Alt Allele: A
Freq: 0.5073% rare
CADD: 0.412

ClinVar Submissions (1)

- Hyperlipoproteinemia, type I, Autosomal recessive

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: CUBN
Variant: c.9340G>A (p.Gly3114Ser)
rsID: rs117035284
Ref Allele: C
Alt Allele: T
Freq: 0.532% rare
CADD: 24.8

ClinVar Submissions (2)

- Megaloblastic anemia due to inborn errors of metabolism, Autosomal recessive
- not specified

Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

Gene: FOXP2
Variant: c.*3199G>A
rsID: rs149254099
Ref Allele: G
Alt Allele: A
Freq: 0.5352% rare
CADD: 12.25

ClinVar Submissions (1)

- Speech-language disorder 1, Autosomal dominant

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: TTC21B
Variant: c.2322+3A>G
rsID: rs79037278
Ref Allele: T

ClinVar Submissions (5)

- Jeune thoracic dystrophy
- Nephronophthisis

Hetero

Alt Allele: C
Freq: 0.5519% rare
CADD: 12.36

- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

TC

Gene: APC
Variant: c.-30378C>T
rsID: rs138386816
Ref Allele: C
Alt Allele: T
Freq: 0.5614% rare
CADD: 20.2

ClinVar Submissions (2)

- Familial adenomatous polyposis 1
- not specified

Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

Gene: GPD1L
Variant: c.*1506C>T
rsID: rs140250795
Ref Allele: C
Alt Allele: T
Freq: 0.563% rare
CADD: 0.829

ClinVar Submissions (1)

- Brugada syndrome

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: PDHX
Variant: c.976G>C
(p.Val326Leu)
rsID: rs35560997
Ref Allele: G
Alt Allele: C
Freq: 0.5662% rare
CADD: 27.6

ClinVar Submissions (4)

- Pyruvate dehydrogenase complex deficiency
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

GC

Gene: CEP290
Variant: c.5237G>A
(p.Arg1746Gln)
rsID: rs61941020
Ref Allele: C
Alt Allele: T
Freq: 0.567% rare
CADD: 22.5

ClinVar Submissions (6)

- Bardet-Biedl syndrome
- Joubert syndrome
- Joubert syndrome
- Leber congenital amaurosis
- Meckel-Gruber syndrome
- Meckel-Gruber syndrome
- Nephronophthisis
- Renal dysplasia and retinal aplasia
- not provided
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

CT

Gene: LAMA2
Variant: c.4470C>T
(p.Asp1490=)
rsID: rs35089085
Ref Allele: C
Alt Allele: T
Freq: 0.571% rare
CADD: 0.354

ClinVar Submissions (4)

- Laminin alpha 2-related dystrophy
- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed (2)

Hetero

CT

Gene: LAMA2
Variant: c.4437-5T>A
rsID: rs41285288
Ref Allele: T

ClinVar Submissions (7)

- Laminin alpha 2-related dystrophy
- not provided

Hetero

Alt Allele: A
Freq: 0.5853% rare
CADD: 11.92

- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (2)

TA

Gene: HBB
Variant: c.*132+221G>A
rsID: rs112988270
Ref Allele: C
Alt Allele: T
Freq: 0.5869% rare
CADD: 1.018

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

CT

Gene: ADAM17
Variant: c.844-5T>C
rsID: rs201461814
Ref Allele: A
Alt Allele: G
Freq: 0.5917% rare
CADD: 18.25

ClinVar Submissions (1)

- Inflammatory skin and bowel disease, neonatal 1, Autosomal recessive

Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

AG

Gene: LEP
Variant: c.*150G>A
rsID: rs28954115
Ref Allele: G
Alt Allele: A
Freq: 0.5917% rare
CADD: 7.456

ClinVar Submissions (1)

- Leptin deficiency or dysfunction, Autosomal recessive
- Monogenic Non-Syndromic Obesity

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: AGRN
Variant: c.3404A>G
(p.Gln1135Arg)
rsID: rs142416636
Ref Allele: A
Alt Allele: G
Freq: 0.5989% rare
CADD: 24.3

ClinVar Submissions (3)

- Myasthenic syndrome, congenital, 8, Autosomal recessive
- not specified

Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

AG

Gene: MYO7A
Variant: c.4461C>T
(p.Asn1487=)
rsID: rs56174006
Ref Allele: C
Alt Allele: T
Freq: 0.6021% rare
CADD: 0.047

ClinVar Submissions (4)

- Nonsyndromic Hearing Loss, Dominant
- Nonsyndromic Hearing Loss, Recessive
- Retinitis pigmentosa-deafness syndrome
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

CT

Gene: CFTR
Variant: c.1365G>T
(p.Ala455=)
rsID: rs79074685
Ref Allele: G
Alt Allele: T
Freq: 0.6124% rare
CADD: 1.971

ClinVar Submissions (2)

- Cystic fibrosis, Autosomal recessive
- not provided

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

GT

Gene: DYSF

ClinVar Submissions (6)

Hetero

Variant: c.3534C>T
(p.Ile1178=)
rsID: rs79899601
Ref Allele: C
Alt Allele: T
Freq: 0.6188% rare
CADD: 3.607

- Dysferlinopathy
- Limb-Girdle Muscular Dystrophy, Recessive
- Miyoshi myopathy
- not provided
- not specified

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed (1)

CT

Gene: ACTN4
Variant: c.1998G>A
(p.Gln666=)
rsID: rs145474119
Ref Allele: G
Alt Allele: A
Freq: 0.6299% rare
CADD: 13.02

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

GA

Gene: CFTR
Variant: c.3285A>T
(p.Thr1095=)
rsID: rs1800118
Ref Allele: A
Alt Allele: T
Freq: 0.6307% rare
CADD: 5.449

ClinVar Submissions (8)

- Cystic fibrosis, Autosomal recessive
- not provided
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (2)

Hetero

AT

Gene: FMN1
Variant: c.993A>G
(p.Lys331=)
rsID: rs74655292
Ref Allele: T
Alt Allele: C
Freq: 0.6403% rare
CADD: 12.58

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

TC

Gene: ATM
Variant: c.*44A>G
rsID: rs55900855
Ref Allele: A
Alt Allele: G
Freq: 0.6467% rare
CADD: 4.754

ClinVar Submissions (1)

- Ataxia-telangiectasia syndrome, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

AG

Gene: ATM
Variant: c.*236C>T
rsID: rs3092834
Ref Allele: C
Alt Allele: T
Freq: 0.6491% rare
CADD: 0.267

ClinVar Submissions (1)

- Ataxia-telangiectasia syndrome, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (2)

Hetero

CT

Gene: ATM
Variant: c.6235G>A
(p.Val2079Ile)
rsID: rs1800060
Ref Allele: G
Alt Allele: A
Freq: 0.6578% rare

ClinVar Submissions (12)

- Ataxia-telangiectasia syndrome, Autosomal recessive
- Hereditary cancer-predisposing syndrome
- not provided
- not specified

Hetero

GA

CADD: 0.3

Low clinical importance, Uncertain benign — Probably benign.

Benign/Likely benign

LitVar SNPedia dbSNP UniProt OMIM OpenSNP PubMed (2)

Gene: GFAP
Variant: c.738G>A
(p.Ala246=)
rsID: rs147404772
Ref Allele: C
Alt Allele: T
Freq: 0.6753% rare
CADD: 6.001

ClinVar Submissions (2)

- Alexander Disease, Autosomal dominant
- not provided

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

Gene: PLAU
Variant: c.-25C>T
rsID: rs2227579
Ref Allele: C
Alt Allele: T
Freq: 0.6769% rare
CADD: 10.03

ClinVar Submissions (1)

- Quebec platelet disorder, Autosomal dominant

Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

CT

Gene: RAB3GAP1
Variant: c.-53C>G
rsID: rs78560065
Ref Allele: C
Alt Allele: G
Freq: 0.6841% rare
CADD: 5.233

ClinVar Submissions (2)

- Warburg micro syndrome
- not specified

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CG

Gene: PHKA2
Variant: c.2077A>G
(p.Ile693Val)
rsID: rs143732206
Ref Allele: T
Alt Allele: C
Freq: 0.6865% rare

ClinVar Submissions (4)

- Glycogen storage disease type IXa1, X-linked recessive
- not provided
- not specified

Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Homo

CC

Gene: APC
Variant: c.645+61C>T
rsID: rs56328836
Ref Allele: C
Alt Allele: T
Freq: 0.6921% rare
CADD: 6.823

ClinVar Submissions (1)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: APC
Variant: c.1743+959C>T
rsID: rs56218335
Ref Allele: C
Alt Allele: T
Freq: 0.6929% rare
CADD: 4.534

ClinVar Submissions (1)

- Hereditary cancer-predisposing syndrome

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: DEAF1
Variant: c.1634C>G
(p.Ala545Gly)

ClinVar Submissions (2)

- not provided

Hetero

rsID: rs34114147
Ref Allele: G
Alt Allele: C
Freq: 0.696% rare
CADD: 17.38

- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

GC

Gene: CUL7
Variant: c.4463T>C (p.Leu1488Pro)
rsID: rs41274912
Ref Allele: A
Alt Allele: G
Freq: 0.7% rare
CADD: 22.4

ClinVar Submissions (1)

- Three M syndrome

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

AG

Gene: BBS10
Variant: c.1631A>G (p.Asn544Ser)
rsID: rs34737974
Ref Allele: T
Alt Allele: C
Freq: 0.7% rare
CADD: 3.342

ClinVar Submissions (5)

- Bardet-Biedl syndrome
- not provided
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

TC

Gene: GAN
Variant: c.*2584A>G
rsID: rs117621048
Ref Allele: A
Alt Allele: G
Freq: 0.7064% rare
CADD: 12.84

ClinVar Submissions (1)

- Giant axonal neuropathy, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

AG

Gene: CIITA
Variant: c.*103T>C
rsID: rs45617532
Ref Allele: T
Alt Allele: C
Freq: 0.7112% rare
CADD: 2.195

ClinVar Submissions (1)

- Bare lymphocyte syndrome 2, Autosomal dominant

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

TC

Gene: HPS3
Variant: c.*170C>T
rsID: rs182666670
Ref Allele: C
Alt Allele: T
Freq: 0.7175% rare
CADD: 0.995

ClinVar Submissions (1)

- Hermansky-Pudlak syndrome

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: NOTCH3
Variant: c.1725G>A (p.Thr575=)
rsID: rs79926127
Ref Allele: C
Alt Allele: T
Freq: 0.7255% rare
CADD: 2.39

ClinVar Submissions (3)

- Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy, Autosomal dominant
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

CT

Gene: F2
Variant: c.813C>T

ClinVar Submissions (1)

Hetero

(p.Gly271=)
rsID: rs5899
Ref Allele: C
Alt Allele: T
Freq: 0.7311% rare
CADD: 7.396

- Hereditary factor II deficiency disease
- Venous thrombosis

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

CT

Gene: MC1R
Variant: c.-356A>G
rsID: rs76337330
Ref Allele: A
Alt Allele: G
Freq: 0.7335% rare
CADD: 9.053

ClinVar Submissions (1)

- Malignant Melanoma Susceptibility

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

AG

Gene: RPGR
Variant: c.1367A>G
(p.Gln456Arg)
rsID: rs144635565
Ref Allele: T
Alt Allele: C
Freq: 0.7486% rare

ClinVar Submissions (4)

- Ciliary dyskinesia
- not provided
- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed (2)

Homo

CC

Gene: PITPNM3
Variant: c.699C>T
(p.Val233=)
rsID: rs149964592
Ref Allele: G
Alt Allele: A
Freq: 0.7502% rare
CADD: 0.049

ClinVar Submissions (2)

- Cone-Rod Dystrophy, Dominant
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

GA

Gene: RYR3
Variant: c.13365G>A
(p.Glu4455=)
rsID: rs118177681
Ref Allele: G
Alt Allele: A
Freq: 0.7645% rare
CADD: 14.11

ClinVar Submissions (1)

- Epileptic encephalopathy

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: APC
Variant: c.-19+968C>T
rsID: rs149097203
Ref Allele: C
Alt Allele: T
Freq: 0.7669% rare
CADD: 18.01

ClinVar Submissions (1)

- Hereditary cancer-predisposing syndrome

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: SLC26A3
Variant: c.357C>A
(p.Phe119Leu)
rsID: rs73419912
Ref Allele: G
Alt Allele: T
Freq: 0.7669% rare
CADD: 0.001

ClinVar Submissions (2)

- not provided
- not specified

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GT

Gene: COL9A3

ClinVar Submissions (3)

Hetero

Variant: c.546C>T
(p.Pro182=)
rsID: rs150148851
Ref Allele: C
Alt Allele: T
Freq: 0.7741% rare
CADD: 0.654

- Multiple Epiphyseal Dysplasia, Dominant
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

CT

Gene: SNAP29
Variant: c.*2783C>T
rsID: rs142566638
Ref Allele: C
Alt Allele: T
Freq: 0.7789% rare
CADD: 0.336

ClinVar Submissions (1)

- Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

Gene: SNAP29
Variant: c.*2791T>C
rsID: rs187911143
Ref Allele: T
Alt Allele: C
Freq: 0.7789% rare
CADD: 2.587

ClinVar Submissions (1)

- Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, Autosomal recessive

Conflicting/Uncertain

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

TC

Gene: DYNC1H1
Variant: c.2511A>G
(p.Ala837=)
rsID: rs17512054
Ref Allele: A
Alt Allele: G
Freq: 0.7884% rare
CADD: 8.797

ClinVar Submissions (4)

- Charcot-Marie-Tooth disease, axonal, type 2O, Autosomal dominant
- Charcot-Marie-Tooth disease, type 2
- History of neurodevelopmental disorder
- Intellectual Disability, Dominant
- Spinocerebellar Ataxia, Dominant
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

AG

Gene: SDHB
Variant: c.487T>C
(p.Ser163Pro)
rsID: rs33927012
Ref Allele: A
Alt Allele: G
Freq: 0.79% rare
CADD: 20.7

ClinVar Submissions (17)

- Cowden syndrome, Autosomal dominant
- Gastrointestinal stroma tumor, Autosomal dominant
- Hereditary cancer-predisposing syndrome
- Paragangliomas 4, Autosomal dominant
- Paragangliomas 4, Autosomal dominant
- Pheochromocytoma, Autosomal dominant
- Pheochromocytoma, Autosomal dominant
- not provided
- not specified

Low clinical importance, Uncertain benign — We evaluate as uncertain but presumed benign. One report linked this variant to Cowden-like syndromes, but the significance of their findings is unclear and other reports treat this as a non-pathological variation.

Conflicting/Uncertain

LitVar SNPedia dbSNP UniProt OMIM OpenSNP PubMed (18)

Hetero

AG

Gene: AKR1D1
Variant: c.*1139G>C
rsID: rs566511122

ClinVar Submissions (1)

- Congenital Bile Acid Synthesis Defect

Hetero

Ref Allele: G
Alt Allele: C
Freq: 0.798% rare
CADD: 1.892

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

GC

Gene: EXT1
Variant: c.-776T>C
rsID: rs555849430
Ref Allele: A
Alt Allele: G
Freq: 0.7988% rare
CADD: 18.59

ClinVar Submissions (1)

- Hereditary Multiple Osteochondromatosis

Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

AG

Gene: MYH8
Variant: c.3686T>C
(p.Met1229Thr)
rsID: rs35962914
Ref Allele: A
Alt Allele: G
Freq: 0.8075% rare
CADD: 22.3

ClinVar Submissions (2)

- not specified

Benign/Likely benign

LitVar SNPedia dbSNP UniProt OpenSNP PubMed (1)

Hetero

AG

Gene: RUNX2
Variant: c.1531G>A
(p.Gly511Ser)
rsID: rs11498198
Ref Allele: G
Alt Allele: A
Freq: 0.8115% rare
CADD: 26.5

ClinVar Submissions (4)

- Cleidocranial dysostosis, Autosomal dominant
- not provided
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP UniProt OMIM OpenSNP PubMed

Hetero

GA

Gene: ATP1A3
Variant: c.2487G>A
(p.Pro829=)
rsID: rs45606534
Ref Allele: C
Alt Allele: T
Freq: 0.8115% rare
CADD: 3.095

ClinVar Submissions (3)

- Alternating hemiplegia of childhood
- Dystonia 12, Autosomal dominant
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

Gene: VWF
Variant: c.7887+12T>C
rsID: rs55687637
Ref Allele: A
Alt Allele: G
Freq: 0.8402% rare
CADD: 7.573

ClinVar Submissions (2)

- not specified
- von Willebrand disorder

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

AG

Gene: PLEC
Variant: c.12441G>A
(p.Pro4147=)
rsID: rs146781600
Ref Allele: C
Alt Allele: T
Freq: 0.8466% rare
CADD: 0.057

ClinVar Submissions (3)

- Epidermolysa bullosa simplex and limb girdle muscular dystrophy, Autosomal recessive
- Epidermolysis bullosa simplex with nail dystrophy, Autosomal recessive
- Epidermolysis bullosa simplex with pyloric atresia, Autosomal recessive
- Epidermolysis bullosa simplex, Ogna type, Autosomal dominant
- Limb-girdle muscular dystrophy, type 2Q, Autosomal recessive
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

CT

Gene: SCN8A
Variant: c.3076C>T (p.Arg1026Cys)
rsID: rs117217073
Ref Allele: C
Alt Allele: T
Freq: 0.8505% rare
CADD: 24.5

ClinVar Submissions (6)

- Early Infantile Epileptic Encephalopathy, Autosomal Dominant
- Early infantile epileptic encephalopathy
- History of neurodevelopmental disorder
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

CT

Gene: ZFYVE26
Variant: c.1844C>T (p.Ser615Phe)
rsID: rs117228915
Ref Allele: G
Alt Allele: A
Freq: 0.8736% rare
CADD: 25.8

ClinVar Submissions (3)

- Spastic paraplegia
- not provided
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: TPP1
Variant: c.1044C>T (p.Ala348=)
rsID: rs35706972
Ref Allele: G
Alt Allele: A
Freq: 0.9119% rare
CADD: 0.259

ClinVar Submissions (6)

- Neuronal ceroid lipofuscinosis, Autosomal dominant
- Seizures
- not provided
- not specified

Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (1)

Hetero

GA

Gene: GAN
Variant: c.46C>T (p.Leu16=)
rsID: rs77470936
Ref Allele: C
Alt Allele: T
Freq: 0.9119% rare
CADD: 22.5

ClinVar Submissions (3)

- Giant axonal neuropathy, Autosomal recessive
- not provided

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

CT

Gene: EPG5
Variant: c.6516C>T (p.Tyr2172=)
rsID: rs114665741
Ref Allele: G
Alt Allele: A
Freq: 0.9557% rare
CADD: 4.022

ClinVar Submissions (1)

- Vici syndrome, Autosomal recessive

Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed

Hetero

GA

Gene: RTTN
Variant: c.5060C>G (p.Ser1687Cys)
rsID: rs34717557
Ref Allele: G
Alt Allele: C
Freq: 0.97% rare
CADD: 25.7

ClinVar Submissions (2)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

GC

Gene: RTTN
Variant: c.1008-4G>T

ClinVar Submissions (2)

Hetero

rsID: rs151203272
Ref Allele: C
Alt Allele: A
Freq: 0.9708% rare
CADD: 1.989

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

CA

Gene: RTTN
Variant: c.1842A>G
(p.Glu614=)
rsID: rs140245773
Ref Allele: T
Alt Allele: C
Freq: 0.974% rare
CADD: 15.09

ClinVar Submissions (2)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

TC

Gene: RTTN
Variant: c.1665C>A
(p.Asn555Lys)
rsID: rs34353615
Ref Allele: G
Alt Allele: T
Freq: 0.974% rare
CADD: 23.8

ClinVar Submissions (2)

- not specified

Benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

GT

Gene: PAX9
Variant: c.516G>A
(p.Lys172=)
rsID: rs61734510
Ref Allele: G
Alt Allele: A
Freq: 0.9835% rare
CADD: 22.4

ClinVar Submissions (2)

- Partial congenital absence of teeth
- Selective tooth agenesis

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

GA

Gene: SNTA1
Variant: c.828G>A
(p.Lys276=)
rsID: rs35938843
Ref Allele: C
Alt Allele: T
Freq: 0.9859% rare
CADD: 8.164

ClinVar Submissions (3)

- Cardiovascular phenotype
- Long QT syndrome
- Romano-Ward syndrome

Conflicting/Uncertain

LitVar SNPedia dbSNP OpenSNP PubMed

Hetero

CT

Gene: TERT
Variant: c.2097C>T
(p.Ala699=)
rsID: rs33963617
Ref Allele: G
Alt Allele: A
Freq: 0.9867% rare
CADD: 2.788

ClinVar Submissions (6)

- Aplastic anemia
- Dyskeratosis Congenita, Recessive
- Dyskeratosis congenita autosomal dominant, Autosomal dominant
- Dyskeratosis congenita, autosomal dominant, 2, Autosomal dominant
- Idiopathic fibrosing alveolitis, chronic form, Autosomal dominant
- Idiopathic fibrosing alveolitis, chronic form, Autosomal dominant
- not specified

Benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (3)

Hetero

GA

Gene: POMT1
Variant: c.2203C>T
(p.Arg735Cys)
rsID: rs147266709

ClinVar Submissions (5)

- Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B1, Autosomal recessive
- Limb-girdle muscular dystrophy-dystroglycanopathy, type C1, Autosomal recessive

Hetero

CT

Ref Allele: C
Alt Allele: T
Freq: 0.9867% **rare**
CADD: 28.1

- Walker-Warburg congenital muscular dystrophy
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OMIM OpenSNP PubMed (3)

Gene: MYH14
Variant: c.2718C>T (p.Arg906=)
rsID: rs138987081
Ref Allele: C
Alt Allele: T
Freq: 0.9891% **rare**
CADD: 7.805

ClinVar Submissions (5)

- Nonsyndromic Hearing Loss, Dominant
- not provided
- not specified

Benign/Likely benign

LitVar SNPedia dbSNP OpenSNP PubMed (1)

Hetero

CT