chr	start	gene	geneFullName	omimPhenotype	hgvs_c	hgvs_p	acmg	maf	rsId	dbSnp...	zyg...	quality	reads
17	73836194	UNC13D	unc-13 homolog D (C. elegans)	Hemophagocytic lymphohistiocytosis, familial, 3	NM_199242.2:c.859-3C>A		3	0.0001		0.0	het.	99.0	18 / 35
17	73825066	UNC13D	unc-13 homolog D (C. elegans)	Hemophagocytic lymphohistiocytosis, familial, 3	NM_199242.2:c.2955-2A>G		2	0.0001		0.0	het.	99.0	16 / 31
1	24180963	FUCA1 TRNAN-GUU	fucosidase, alpha-L- 1, tissue TRNAN-GUU	Fucosidosis	NM_000147.4:c.856C>T	NP_000138.2:p.Gln286Ter	2	0.0001		0.0	het.	99.0	24 / 41
17	4836804	GP1BA	glycoprotein Ib (platelet), alpha polypeptide	Bernard-Soulier syndrome, type A1 (recessive); {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}; Bernard-Soulier syndrome, type A2 (dominant); von Willebrand disease, platelet-type	NM_000173.5:c.905G>A	NP_000164.5:p.Gly302Asp	3	0.0002	rs185903059	9.183E-4	het.	99.0	27 / 50
17	4837070	GP1BA	glycoprotein Ib (platelet), alpha polypeptide	Bernard-Soulier syndrome, type A1 (recessive); {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}; Bernard-Soulier syndrome, type A2 (dominant); von Willebrand disease, platelet-type	NM_000173.5:c.1171C>T	NP_000164.5:p.Pro391Ser	3	0.0009	rs147455264	0.005969	het.	99.0	17 / 30
1	91843691	HFM1 TRNAN-GUU	HFM1, ATP-dependent DNA helicase homolog (S. cerevisiae) TRNAN-GUU	Premature ovarian failure 9	NM_001017975.3:c.1286T>G	NP_001017975.3:p.Val429Gly	3	0.0002		0.0	het.	99.0	18 / 47
9	2645735	VLDLR	very low density lipoprotein receptor	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	NM_003383.3:c.1474G>A NM_001018056.1:c.1474G>A	NP_003374.3:p.Ala492Thr NP_001018066.1:p.Ala492Thr	3	0.0001	rs141757226	0.0	het.	99.0	27 / 52
16	79633085	MAF	v-maf avian musculoaponeurotic fibrosarcoma oncogene homolog	Cataract 21, multiple types	NM_005360.4:c.715G>A NM_001031804.2:c.715G>A	NP_005351.2:p.Ala239Thr NP_001026974.1:p.Ala239Thr	3	0.0002		0.0	het.	13.0	5 / 6
16	79633099	MAF	v-maf avian musculoaponeurotic fibrosarcoma oncogene homolog	Cataract 21, multiple types	NM_005360.4:c.690_701del NM_001031804.2:c.690_701del	NP_005351.2:p.Gly235_Gly238del NP_001026974.1:p.Gly235_Gly238del	3	0.0000		0.0	het.	74.0	2 / 4
2	179542447	TTN	titin	Cardiomyopathy, familial hypertrophic, 9; Cardiomyopathy, dilated, 1G; Tibial muscular dystrophy, tardive; Muscular dystrophy, limb-girdle, type 2J; Myopathy, proximal, with early respiratory muscle involvement; Myopathy, early-onset, with fatal cardiomyopathy	NM_133432.3:c.13658-35403G>T NM_001267550.1:c.34192G>T NM_001256850.1:c.33241G>T NM_003319.4:c.13283-35403G>T NM_133437.3:c.13859-35403G>T NM_133378.4:c.30460G>T	NP_001254479.1:p.Val11398Phe NP_001243779.1:p.Val11081Phe NP_596869.4:p.Val10154Phe	3	0.0002		0.0	het.	99.0	27 / 62
1	47716921	STIL TRNAN-GUU	SCL/TAL1 interrupting locus TRNAN-GUU	Microcephaly 7, primary, autosomal recessive	NM_003035.2:c.3751A>G NM_001048166.1:c.3754A>G	NP_001041631.1:p.Ile1252Val NP_003026.2:p.Ile1251Val	3	0.0002	rs142210835	0.0	het.	99.0	19 / 45
9	2635542	VLDLR	very low density lipoprotein receptor	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	NM_001018056.1:c.172_173del NM_003383.3:c.172_173del	NP_001018066.1:p.Val59Ter NP_003374.3:p.Val59Ter	2	0.0001		0.0	het.	99.0	23 / 37
1	74671377	FPGT FPGT-TNNI3K TNNI3K TRNAN-GUU	FPGT-TNNI3K readthrough fucose-1-phosphate guanylyltransferase TNNI3 interacting kinase TRNAN-GUU	?Cardiac conduction disease with or without dilated cardiomyopathy	NM_001112808.2:c.382+4283A>C NM_001199328.2:c.923A>C NM_001199329.2:c.*319A>C NM_001199327.1:c.382+4283A>C NM_003838.4:c.1685A>C	NP_001186257.2:p.Lys308Thr NP_003829.3:p.Lys562Thr	3	0.0002		0.0	het.	99.0	20 / 46
1	53715098	LRP8 TRNAN-GUU	low density lipoprotein receptor-related protein 8, apolipoprotein e receptor TRNAN-GUU	{Myocardial infarction, susceptibility to}	NM_017522.4:c.2064+1264A>C NM_004631.4:c.2807A>C NM_033300.3:c.2297A>C NM_001018054.2:c.2676+1264A>C	NP_150643.2:p.Gln766Pro NP_004622.2:p.Gln936Pro	3	0.0002		0.0	het.	99.0	21 / 34
1	43893302	SZT2 TRNAN-GUU	seizure threshold 2 homolog (mouse) TRNAN-GUU	Epileptic encephalopathy, early infantile, 18	NM_015284.3:c.3529C>T	NP_056099.3:p.Arg1177Trp	3	0.0002		0.0	het.	99.0	19 / 36
1	240371443	FMN2	formin 2	Mental retardation, autosomal recessive 47	NM_020066.4:c.3331_3363del	NP_064450.3:p.Pro1112_Pro1122del	3	0.0005		0.0	het.	87.0	3 / 4
1	240370975	FMN2	formin 2	Mental retardation, autosomal recessive 47	NM_020066.4:c.2863C>G	NP_064450.3:p.Pro955Ala	3	0.0000		0.0	het.	59.0	8 / 23
22	50660214	TUBGCP6	tubulin, gamma complex associated protein 6	Microcephaly and chorioretinopathy, autosomal recessive, 1	NM_020461.3:c.2574G>C	NP_065194.2:p.Trp858Cys	3	0.0005	rs147989796	0.004591	hom.	99.0	40 / 40
1	110884168	RBM15 TRNAN-GUU	RNA binding motif protein 15 TRNAN-GUU	Megakaryoblastic leukemia, acute	NM_001201545.1:c.2141G>T NM_022768.4:c.2141G>T	NP_001188474.1:p.Gly714Val NP_073605.4:p.Gly714Val	3	0.0007	rs182719265	4.591E-4	het.	99.0	17 / 38
1	32669882	CCDC28B TRNAN-GUU	coiled-coil domain containing 28B TRNAN-GUU	{Bardet-Biedl syndrome 1, modifier of}	NM_024296.3:c.427G>A	NP_077272.2:p.Glu143Lys	3	0.0002		0.0	het.	99.0	17 / 31
1	53793511	LRP8 TRNAN-GUU	low density lipoprotein receptor-related protein 8, apolipoprotein e receptor TRNAN-GUU	{Myocardial infarction, susceptibility to}	NM_033300.3:c.71_73dupTGC NM_017522.4:c.71_73dupTGC NM_001018054.2:c.71_73dupTGC NM_004631.4:c.71_73dupTGC	NP_004622.2:p.Leu24dup NP_150643.2:p.Leu24dup NP_059992.3:p.Leu24dup NP_001018064.1:p.Leu24dup	3	0.0009		0.0	het.	99.0	19 / 29
1	44071942	PTPRF TRNAN-GUU	protein tyrosine phosphatase, receptor type, F TRNAN-GUU	?Breasts and/or nipples, aplasia or hypoplasia of, 2	NM_130440.2:c.3488_3490del NM_002840.3:c.3515_3517del	NP_002831.2:p.Gln1173del NP_569707.2:p.Gln1164del	3	0.0002		0.0	het.	99.0	12 / 24
2	179440424	TTN TTN-AS1	titin TTN antisense RNA 1	Cardiomyopathy, familial hypertrophic, 9; Cardiomyopathy, dilated, 1G; Tibial muscular dystrophy, tardive; Muscular dystrophy, limb-girdle, type 2J; Myopathy, proximal, with early respiratory muscle involvement; Myopathy, early-onset, with fatal cardiomyopathy	NM_001267550.1:c.70435C>T NM_001256850.1:c.65512C>T NM_133432.3:c.43615C>T NR_038272.1:n.2044-6875G>A NM_003319.4:c.43240C>T NM_133378.4:c.62731C>T NM_133437.3:c.43816C>T NR_038271.1:n.596+4248G>A	NP_597676.3:p.Arg14539Trp NP_597681.3:p.Arg14606Trp NP_596869.4:p.Arg20911Trp NP_003310.4:p.Arg14414Trp NP_001254479.1:p.Arg23479Trp NP_001243779.1:p.Arg21838Trp	3	0.0002		0.0	het.	99.0	21 / 31