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PlainSEEK v1.1 Variant list

Gene/Region (GRCh37)	Variant Nomenclature	Associated disorder	Interpretation	Evidence
chr2:44102515-44102516	ABCG8(NM_022437.3):c.1720G>A, p.Gly574Arg	Sitosterolemia 1 (MIM#: 210250)	Pathogenic	ClinvarID:4968; PMIDs: 11099417:15996216; 25073796
chr1:76198408-76198409	ACADM(NM_000016.4): c.199T>C, p.Tyr67His	Acyl-CoA dehydrogenase, medium chain, deficiency of (MCAD deficiency) (MIM#: 201450)	Pathogenic	ClinvarID:3597; PMID: 11349232; 11409868; 15832312; 26947917; 20301597
chr1:76226845-76226846	ACADM(NM_000016.4):c.985A>G, p.Lys329Glu	Acyl-CoA dehydrogenase, medium chain, deficiency of (MCAD deficiency) (MIM#: 201450)	Pathogenic	ClinvarID:3586 PMID: 11349232; 11409868; 15832312 ; 26947917; 23574375; 20301597
chr20:43251679-43251680	ADA(NM_000022.2):c.646G>A, p.Gly216Arg	Severe combined immunodeficiency due to ADA deficiency (SCID) (MIM#: 102700)	Pathogenic	ClinvarID:1968; PMID: 1680289; 26255240
chr1:154560600-154560601	ADAR(NM_001111.4):c.3019G>A, p.Gly1007Arg	Aicardi-Goutieres syndrome 6 (MIM#: 615010)	Pathogenic	ClinvarID:39458; PMID: 15955093; 23001123; 20301648
chr1:33478879-33478880	AK2(NM_001625.3):c.622T>C, p.Ser208Pro	Reticular dysgenesis (MIM#: 267500)	Likely Pathogenic	ClinvarID:1001459; PMID: 31673062
chr11:108121752-108121753	ATM(NM_000051.3): c.1564_1565delGA, p.Glu522IlefsTer43	Ataxia-telangiectasia (MIM#: 208900)	Pathogenic	ClinvarID:127340; PMID: 9463314; 16266405; 9443866; 9887333; 20301790
chr11:108183150-108183151	ATM(NM_000051.3):c.5932G>T, p.Glu1978Ter	Ataxia-telangiectasia (MIM#: 208900)	Pathogenic	ClinvarID:127414; PMID: 16266405; 9443866; 9887333; 20301790
chr11:108188100-108188101	ATM(NM_000051.3):c.6200C>A, p.Ala2067Asp	Ataxia-telangiectasia (MIM#: 208900)	Pathogenic	ClinvarID:39749; PMID: 9887333; 25077176; 22345219; 20301790
chr18:55362419-55362420	ATP8B1(NM_005603.6):c.923G>T, p.Gly308Val	Cholestasis, progressive familial intrahepatic 1 (Byler disease) (MIM#: 211600)	Pathogenic	ClinvarID:7262; PMID: 9500542; 20301474
chr12:58020614-58020615	B4GALNT1(NM_001478.4):c.1514G>A, p.Arg505His	Spastic paraplegia 26, autosomal recessive (MIM#: 609195)	Pathogenic	ClinvarID:1074269; PMID: 24103911
chr11:66293651-66293652	BBS1(NM_024649.4):c.1169T>G, p.Met390Arg	Bardet-Biedl syndrome 1 (MIM#: 209900)	Pathogenic	ClinvarID:12143; PMID: 12118255; 22940089; 23143442
chr19:41930486-41930487	BCKDHA(NM_000709.3):c.1312T>A, p.Tyr438Asn	Maple syrup urine disease, type Ia (MIM#: 248600)	Pathogenic	ClinvarID:100009; PMID: 20301495; 12888983; 1867199
chr7:2583387-2583388	BRAT1(NM_152743.3):c.638dupA, p.Val214GlyfsTer189	Rigidity and multifocal seizure syndrome, lethal neonatal (MIM#: 614498)	Pathogenic	ClinvarID:31199; PMID: 22279524
chr3:15686692-15686693	BTD(NM_001323582.1):c.1270G>C, p.Asp424His	Biotinidase deficiency (MIM#: 253260)	Pathogenic	ClinvarID:1900; PMID: 9654207; 11668630; 9232193; 20301497

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chr3:15686730-15686731	BTD(NM_001323582.1):c.1308A>C, p.Gln436His	Biotinidase deficiency (MIM#: 253260)	Pathogenic	ClinvarID:1902; PMID: 20301497; 9232193; 11668630
chr3:15686821-15686822	BTD(NM_001323582.1):c.1399T>C, p.Trp467Arg	Biotinidase deficiency (MIM#: 253260)	Pathogenic	ClinvarID:25091; PMID: 20301497; 10801060
chr15:42703123-42703124	CAPN3(NM_000070.2):c.2306G>A, p.Arg769Gln	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly (MIM#: 236500)	Pathogenic	ClinvarID:17613; PMID: 7720071; 9246005; 20301490
chr10:95266795-95266796	CEP55(NM_018131.4):c.514dupA, p.Ile172AsnfsTer17	Ciliary dyskinesia, primary, 38 (MIM#: 618063)	Pathogenic	ClinvarID:562113; PMID: 30622327
chrX:47487524-47487525	CFP(NM_001145252.1):c.379T>G, p.Cys127Gly	Properdin deficiency (MIM#: 312060)	Likely Pathogenic	Clinvar:2785216 PMID: 31028937 (Crowgey) PP3-Strong; PM2; PP1(personal communication)
chr7:117292930-117292931	CFTR(NM_000492.3):c.3909C>G>A, p.Asn1303Lys	Cystic fibrosis (MIM#: 219700)	Pathogenic	ClinvarID:7136
chr7:117199644-117199645	CFTR(NM_000492.3):c.1521_1523delCTT, p.Phe508del	Cystic fibrosis (MIM#: 219700)	Pathogenic	ClinvarID:7105
chr7:117251796-117251797	CFTR(NM_000492.3):c.3302T>A, p.Met1101Lys	Cystic fibrosis (MIM#: 219700)	Pathogenic	ClinvarID:39516
chr7:117282540-117282541	CFTR(NM_000492.3):c.3773dupT, p.Leu1258fsTer7	Cystic fibrosis (MIM#: 219700)	Pathogenic	ClinvarID:38726
chr10:73768086-73768087	CHST3(NM_004273.5):c.1298C>T, p.Pro433Leu	Spondyloepiphyseal dysplasia with congenital joint dislocations (MIM#: 143095)	Likely Pathogenic	rs965883985; PMID: 31028937 (Crowgey); PP3 – Strong; PM2; PP1 (personal communication)
chr2:97464924-97464925	CNNM4(NM_020184.3):c.1813C>T, p.Arg605Ter	Jalili syndrome (MIM#: 217080)	Pathogenic	ClinvarID:2084124; PMID: 29322253; 19200525
chr7:148080972-148080973	CNTNAP2(NM_014141.5):c.3709delG, p.Asp1237fsTer17	Pitt-Hopkins like syndrome 1 (MIM#: 610042)	Pathogenic	ClinvarID:5490; PMID: 16571880
chr7:94049562-94049563	COL1A2(NM_000089.3):c.2098G>T, p.Gly700Cys	Osteogenesis imperfecta, type I (MIM#: 166200)	Pathogenic	ClinvarID:641929; PMID: 19594296 (also known as p.Gly610Cys in literature)
chr12:94243828-94243829	CRADD(NM_003805.3):c.382G>C, p.Gly128Arg	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly (MIM#: 614499)	Pathogenic	ClinvarID:30360; PMID: 27773430; 22279524
chrX:37664328-37664329	CYBB(NM_000397.3):c.1222G>A, p.Gly408Arg	Chronic granulomatous disease (MIM#: 306400)	Pathogenic	ClinvarID:68376 PMID: 9585602
chrX:37665659-37665660	CYBB(NM_000397.3):c.1335C>A, p.Cys445Ter	Chronic granulomatous disease (MIM#: 306400)	Pathogenic	PMID: 11162142; PMID: 31028937
chr8:143999147-143999148	CYP11B2(NM_000498.3): c.104_109delTGCTGCinsG, p.Val35GlyfsTer3	Hypoaldosteronism, congenital, due to CMO I deficiency (corticosterone methyloxidase type I deficiency) (MIM#: 203400)	Pathogenic	ClinvarID:16878

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chr4:108868556-108868557	CYP2U1(NM_183075.3): c.1152_1153delAG, p.Arg384SerfsTer54	Spastic paraplegia 56 (MIM#: 615030)	Likely Pathogenic	ClinvarID:582218
chr8:145541968-145541969	DGAT1(NM_012079.5):c.629_631delCCT, p.Ser210del	Diarrhea 7, protein-losing enteropathy type (MIM#: 615863)	Pathogenic	ClinvarID:548013; PMID: 29604290; 31778854
chr7:819733-819734	DNAAF5(NM_017802.3):c.2384T>C, p.Leu795Pro	Ciliary dyskenesia, primary, 18 (MIM#: 614874)	Pathogenic	ClinvarID:39684; PMID: 23040496
chr5:13753398-13753399	DNAH5(NM_001369.2):c.10815delT, p.Pro3606HisfsTer23	Ciliary dyskinesia, primary, 3 (MIM#: 608644)	Pathogenic	ClinvarID:65636; PMID: 23477994; 19357118
chr5:13865783-13865784	DNAH5(NM_001369.3):c.4348C>T, p.Gln1450Ter	Ciliary dyskinesia, primary, 3 (MIM#: 608644)	Pathogenic	ClinvarID:208992; PMID: 23477994; 16627867
chr9:34459051-34459052	DNAI1(NM_012144.3):c.48+2dupT	Ciliary dyskinesia, primary, 1 (MIM#: 244400)	Pathogenic	ClinvarID:5604; PMID: 10577904; 16858015; 11231901
chr6:7562985-7562986	DSP(NM_004415.4):c.699G>A, p.Trp233Ter	Arrhythmogenic right ventricular dysplasia 8; Cardiomyopathy, dilated, with woolly hair and keratoderma (MIM#: 607450; 605676; 615821)	Pathogenic	ClinvarID: 44946; PMID: 16917092
chr13:78475315-78475316	EDNRB(NM_001122659.2):c.828G>T, p.Trp276Cys	Hirschsprung disease, susceptibility to, 2 (MIM#: 600155)	Pathogenic	ClinvarID:16633; PMID: 8001158; 10964697
chr10:50681521-50681522	ERCC6(NM_000124.3):c.2709+1G>T	Cockayne syndrome, type B (MIM#: 133540)	Pathogenic	ClinvarID: 847342; PMID: 2359970
chr4:5795448-5795449	EVC(NM_153717.2):c.1886+5G>T	Ellis-van Creveld syndrome (MIM#: 225500)	Pathogenic	ClinvarID:5338; PMID: 10700162; 10700184
chr4:187206813-187206814	F11(NM_000128.3):c.1327C>T, p.Arg443Cys	Factor XI deficiency (MIM#: 612416)	Likely Pathogenic	ClinvarID:1324363; PMID: 16835901
chrX:138643868-138643869	F9(NM_000133.3):c.1025C>T, p.Thr342Met	Hemophilia B (MIM#: 306900)	Pathogenic	ClinvarID:10607; PMID: 1864609; 22639855
chr10:123279673-123279674	FGFR2(NM_000141.3):c.758C>G, p.Pro253Arg	Jackson-Weiss syndrome (MIM#: 123150)	Pathogenic	ClinvarID:13273; PMID: 7668257; 31145570
chr1:213032154-213032155	FLVCR1(NM_014053.3):c.361A>G, p.Asn121Asp	Ataxia, posterior column, with retinitis pigmentosa (MIM#: 609033)	Pathogenic	ClinvarID:18418; PMID: 21070897; 22279524
chr17:41063407-41063408	G6PC1(NM_000151.4):c.1039C>T, p.Gln347Ter	Glycogen storage disease Ia (MIM#: 232200)	Pathogenic	ClinvarID:12000; PMID: 8182131; 20301489
chr9:34648166-34648167	GALT(NM_000155.3):c.563A>G, p.Gln188Arg	Galactosemia (MIM#: 230400)	Pathogenic	ClinvarID:3614; PMID: 1897530; 20301691
chr9:34649441-34649442	GALT(NM_000155.3):c.940A>G, p.Asn314Asp	Galactosemia (MIM#: 230400)	Other-Possible Duarte	ClinvarID:3613; PMID: 8198125; 9012409; 20301691
chr19:13010299-13010300	GCDH(NM_000159.3):c.1262C>T, p.Ala421Val	Glutaric aciduria, type I (MIM#: 231670)	Pathogenic	ClinvarID:2082; PMID: 8900227; 17478444

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chr13:20763491-20763492	GJB2(NM_004004.5):c.229T>C, p.Trp77Arg	Deafness, autosomal recessive 1A (non-syndromic) (MIM#: 220290)	Pathogenic	ClinvarID:17003; PMID: 9328482; 20301449
chr13:20763685-20763686	GJB2(NM_004004.5):c.35delG, p.Gly12ValfsTer2	Deafness, autosomal recessive 1A (non-syndromic) (MIM#: 220290)	Pathogenic	ClinvarID:17004; PMID: 9328482; 20301449
chr1:228345661-228345662	GJC2(NM_020435.3):c.203A>G, p.Tyr68Cys	Leukodystrophy, hypomyelinating, 2 (MIM#: 608804)	Likely Pathogenic	ClinvarID:1472061;
chr3:33093386-33093387	GLB1(NM_000404.3):c.902C>T, p.Ala301Val	GM1-gangliosidosis, type I (MIM#: 230500)	Pathogenic	ClinvarID:381567; PMID: 16941474; 20175788
chr12:102147247-102147248	GNPTAB(NM_024312.4): c.3503_3504delTC, p.Leu1168GlnfsTer5	Mucolipidosis II alpha/beta (MIM#: 252500)	Pathogenic	ClinvarID:2771; PMID: 20301728; 16465621
chr5:140054360-140054361	HARS1(NM_002109.4):c.1361A>C, p.Tyr454Ser	Usher syndrome type 3B (MIM#: 614504)	Pathogenic	ClinvarID:29756; PMID: 22279524
chr1:119958076-119958077	HSD3B2(NM_000198.3):c.35G>A, p.Gly12Glu	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency (MIM#: 201810)	Pathogenic	ClinvarID:1513071; PMID: 26079780
chr3:50357477-50357478	HYAL2(NM_003773.4):c.443A>G, p.Lys148Arg	Orofacial clefting (MIM#: N/A)	Pathogenic	ClinvarID:1065389; PMID: 28081210
chr5:35857080-35857081	IL7R(NM_002185.3):c.2T>G, p.Met1?	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type (MIM#: 608971)	Likely Pathogenic	ClinvarID:948034; PMID: 31028937
chr20:33001601-33001602	ITCH(NM_031483.6):c.394dupA, p.Ile132AsnfsTer9	Autoimmune disease, multisystem, with facial dysmorphism (MIM#: 613385)	Pathogenic	ClinvarID:4391; PMID: 20170897
chr7:150648583-150648584	KCNH2(NM_000238.4):c.1897A>C, p.Asn633His	Long QT syndrome 2 (MIM#: 613688)	Likely Pathogenic	rs199472960; PMID: 16155735; PMID: 18593567; PMID: 31028937
chr11:2549221-2549222	KCNQ1(NM_000218.2):c.451_452delCT, p.Leu151GlyfsTer133	Long QT syndrome 1 (Jervell and Lange-Nielsen syndrome 1) (MIM#: 192500)	Pathogenic	ClinvarID:53045 PMID: 10077519
chr11:2592620-2592621	KCNQ1(NM_000218.2):c.671C>T, p.Thr224Met	Long QT syndrome 1 (Jervell and Lange-Nielsen syndrome 1) (MIM#: 192500)	Pathogenic	ClinvarID:67096 PMID: 33141630
chr19:47983174-47983175	KPTN(NM_007059.2): c.714_731dupTCTGCAGATGTGGTCGGT, p.Met241_Gln246dup	Mental retardation, autosomal recessive 41 (MIM#: 615637)	Pathogenic	ClinvarID:100680 PMID: 24239382
chr19:47983130-47983131	KPTN(NM_007059.2):c.776C>A, p.Ser259Ter	Mental retardation, autosomal recessive 41 (MIM#: 615637)	Pathogenic	ClinvarID:100679 PMID: 24239382
chrX:153136334-153136335	L1CAM(NM_001278116.1):c.604G>A, p.Asp202Asn	MASA syndrome; Hydrocephalus (MIM#: 303350; 307000)	Pathogenic	ClinvarID:689728 PMID: 25644381
chr3:49169567-49169568	LAMB2(NM_002292.3):c.440A>G, p.His147Arg	Pierson syndrome (MIM#: 609049)	Likely Pathogenic	ClinvarID:29774 PMID: 21236492
chr1:156104247-156104248	LMNA(NM_170707.3):c.568C>T, p.Arg190Trp	Cardiomyopathy, dilated, 1A (MIM#: 115200)	Pathogenic	ClinvarID:66908 PMID: 11897440
chr19:5694556-5694557	LONP1(NM_004793.3):c.2161C>G, p.Arg721Gly	CODAS syndrome (MIM#: 600373)	Likely Pathogenic	ClinvarID:1367756 PMID: 25574826; 34228963

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chr11:68153992-68153993	LRP5(NM_002335.2):c.1225A>G, p.Thr409Ala	Osteoporosis-pseudoglioma syndrome (MIM#: 259770)	Pathogenic	ClinvarID:843927 PMID: 18602879
chr11:68154042-68154043	LRP5(NM_002335.2):c.1275G>A, p.Trp425Ter	Osteoporosis-pseudoglioma syndrome (MIM#: 259770)	Pathogenic	ClinvarID:1459979 PMID: 18602879
chr20:10393912-10393913, chr20:10393438-10393439	MKKS(NM_170784.2): c.[250C>T;724G>T], p.[His84Tyr;Ala242Ser]	McKusick-Kaufman syndrome (MIM#: 236700)	Pathogenic – Haplotype	ClinvarID:478890, ClinvarID:167303 PMID: 10802661; 16104012
chr1:45973215-45973216	MMACHC(NM_015506.2):c.271dupA, p.Arg91LysfsTer14	Methylmalonic aciduria and homocystinuria, cblC type (MIM#: 277400)	Pathogenic	ClinvarID:1421 PMID: 16311595; 20610126
chr15:56739084-56739085	MNS1(NM_018365.2): c.407_410delAAAG, p.Glu136GlyfsTer16	Heterotaxy, visceral, 9, autosomal, with male infertility (MIM#: 618948)	Pathogenic	ClinvarID:635005 PMID: 31534215
chr7:40172767-40172768	MPLKIP(NM_138701.3):c.430A>G, p.Met144Val	Trichothiodystrophy 4, nonphotosensitive (MIM#: 234050)	Pathogenic	ClinvarID:1844 PMID: 4847854; 15645389
chr1:11854822-11854823	MTHFR(NM_005957.4):c.1129C>T, p.Arg377Cys	Homocystinuria due to MTHFR deficiency (MIM#: 236250)	Pathogenic	ClinvarID:3528 PMID: 17409006
chr10:30602854-30602855	MTPAP(NM_018109.3):c.1432A>G, p.Asn478Asp	Spastic ataxia 4, autosomal recessive (MIM#: 613672)	Pathogenic	ClinvarID:18391 PMID: 20970105; 25008111
chr12:110034364-110034365	MVK(NM_000431.3):c.1174G>A, p.Ala392Thr	Mevalonic aciduria (MIM#: 610377)	Likely Pathogenic	ClinvarID: 1039865 PMID: 31028937
chr12:110029079-110029080	MVK(NM_000431.3):c.803T>C, p.Ile268Thr	Mevalonic aciduria (MIM#: 610377)	Pathogenic	ClinvarID: 11932 PMID: 10369261; 11313769
chr11:47354742-47354743	MYBPC3(NM_000256.3):c.3330+2T>G	Severe neonatal hypertrophic cardiomyopathy/ Hypertrophic cardiomyopathy 4; Dilated cardiomyopathy 1MM; Left ventricular noncompaction 10 (MIM#: 115197; 615396)	Pathogenic	ClinvarID:8621 PMID: 25031304
chr3:47033133-47033134	NBEAL2(NM_015175.2):c.881C>G, p.Ser294Ter	Gray platelet syndrome (MIM#: 139090)	Pathogenic	ClinvarID:31117 PMID: 21765412
chr11:67377980-67377981	NDUFV1(NM_007103.3):c.640G>A, p.Glu214Lys	Mitochondrial complex I deficiency, nuclear type 4 (MIM#: 618225)	Pathogenic	ClinvarID:14059 PMID: 11349233
chr3:132420271-132420272	NPHP3(NM_153240.4):c.1628+2T>A	Nephronophthisis 3 (MIM#: 604387)	Likely Pathogenic	ClinvarID:1067254
chr3:132415641-132415642	NPHP3(NM_153240.4):c.2104C>T, p.Arg702Ter	Nephronophthisis 3 (MIM#: 604387)	Pathogenic	ClinvarID:2640
chr19:36337055-36337056	NPHS1(NM_004646.3):c.1481delC, p.Ser494CysfsTer55	Nephrotic syndrome, type 1 (MIM#: 256300)	Pathogenic	ClinvarID:56441 PMID: 10577936
chr19:36322580-36322581	NPHS1(NM_004646.3):c.3250delG, p.Val1084SerfsTer59	Nephrotic syndrome, type 1 (MIM#: 256300)	Pathogenic	ClinvarID:56496 PMID: 10577936
chr1:179530461-179530462	NPHS2(NM_014625.3):c.413G>A, p.Arg138Gln	Nephrotic syndrome, type 2 (MIM#: 600995)	Pathogenic	ClinvarID:5360 PMID: 24227627
chr16:167343-167344	NPRL3(NM_001077350.2):c.349delG, p.Glu117LysfsTer5	Epilepsy, familial focal, with variable foci 3 (MIM#: 617118)	Pathogenic	ClinvarID:861113 PMID: 28726809
chrX:38260562-38260563	OTC(NM_000531.5):c.422G>A, p.Arg141Gln	Ornithine transcarbamylase deficiency (MIM#: 311250)	Pathogenic	ClinvarID:10987 PMID: 8985493

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chr12:103237567-103237568	PAH(NM_000277.1):c.1066-11G>A	Phenylketonuria (MIM#: 261600)	Pathogenic	ClinvarID:607 PMID: 12888983
chr12:103234176-103234177	PAH(NM_000277.1):c.1315+1G>A	Phenylketonuria (MIM#: 261600)	Pathogenic	ClinvarID:576 PMID: 29560316
chr12:103288578-103288579	PAH(NM_000277.1):c.284_286delTCA, p.Ile95del	Phenylketonuria (MIM#: 261600)	Pathogenic	ClinvarID:604 PMID: 12655544
chr12:103246652-103246653	PAH(NM_000277.1):c.782G>A, p.Arg261Gln	Phenylketonuria (MIM#: 261600)	Pathogenic	ClinvarID:582 PMID: 29560316
chr20:3888870-3888871	PANK2(NM_153638.3): c.930_936delTGTCTTT, p.Phe311ProfsTer16	Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP syndrome) (MIM#: 607236)	Pathogenic	ClinvarID:4547 PMID: 11479594
chr3:136048853-136048854	PCCB(NM_000532.4):c.1606A>G, p.Asn536Asp	Propionicacidemia (MIM#: 606054)	Pathogenic	ClinvarID:38881 PMID: 12757933, 12888983
chr20:5096117-5096118	PCNA(NM_182649.1):c.683G>T, p.Ser228Ile	Ataxia-telangiectasia-like disorder 2 (MIM#: 615919)	Likely Pathogenic	ClinvarID:143043 PMID: 24911150
chr19:33902602-33902603	PEPD(NM_000285.3):c.793C>T, p.Arg265Ter	Prolidase deficiency (MIM#: 170100)	Pathogenic	ClinvarID:215 PMID: 16470701
chr14:51379745-51379746	PYGL(NM_002863.4):c.1620+1G>A	Glycogen storage disease VI (MIM#: 232700)	Pathogenic	ClinvarID:11996 PMID: 9536091
chr11:36597827-36597828	RAG1(NM_000448.2):c.2974A>G, p.Lys992Glu	Severe combined immunodeficiency, B cell-negative (MIM#: 601457)	Pathogenic	ClinvarID:372488 PMID: 18442948
chr9:35657944-35657945	RMRP(NR_003051.3):n.71A>G	Cartilage-hair hypoplasia (MIM#: 250250)	Pathogenic	ClinvarID:14208 PMID: 8034306, 16838329
chr13:51519580-51519581	RNASEH2B(NM_024570.3):c.529G>A, p.Ala177Thr	Aicardi-Goutieres syndrome 2 (MIM#: 610181)	Pathogenic	ClinvarID:1262 PMID: 17846997
chr2:122288505-122288506	RNU4ATAC(NR_023343.1):n.51G>A Note: CLASP1:c.196-605C>T in Clinvar	Microcephalic osteodysplastic primordial dwarfism, type I (MIM#: 210710)	Pathogenic	ClinvarID:30178 PMID: 21474760, 21474761, 23794361
chr20:35532653-35532654	SAMHD1(NM_015474.3):c.1411-2A>G	Aicardi-Goutieres syndrome 5 (MIM#: 612952)	Pathogenic	ClinvarID:126407 PMID: 21402907
chr20:35563512-35563513	SAMHD1(NM_015474.3):c.428G>A, p.Arg143His	Aicardi-Goutieres syndrome 5 (MIM#: 612952)	Pathogenic	ClinvarID:126411 PMID: 19525956
chr15:76998254-76998255	SCAPER(NM_020843.2):c.2236dupA, p.Ile746AsnfsTer6	Intellectual developmental disorder and retinitis pigmentosa (MIM#: 618195)	Likely Pathogenic	ClinvarID:800544 PMID: 31192531
chr19:35524544-35524545	SCN1B(NM_001037.4):c.350G>A, p.Gly117Asp	Generalized epilepsy with febrile seizures plus, type 1 (MIM#: 604233)	Likely Pathogenic	ClinvarID: 568700 PMID: 28726809
chr4:52896001-52896002	SGCB(NM_000232.4):c.271C>T, p.Arg91Cys	Muscular dystrophy, limb-girdle, autosomal recessive 4 (MIM#: 604286)	Pathogenic	ClinvarID:499193 PMID: 21480868
chr4:52895064-52895065	SGCB(NM_000232.4):c.452C>G, p.Thr151Arg	Muscular dystrophy, limb-girdle, autosomal recessive 4 (MIM#: 604286)	Pathogenic	ClinvarID:8712 PMID: 8968749, 9565988
chr16:56919274-56919275	SLC12A3(NM_001126108.1):c.1924C>G, p.Arg642Gly	Gitelman syndrome (MIM#: 263800)	Pathogenic	ClinvarID:397523 PMID: 10561140
chr6:74354305-74354306	SLC17A5(NM_012434.4):c.115C>T, p.Arg39Cys	Salla disease (MIM#: 604369)	Pathogenic	ClinvarID:5615 PMID: 16158439

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chr17:73274345-73274346	SLC25A19(NM_001126121.1):c.530G>C, p.Gly177Ala	Microcephaly, Amish type (MIM#: 607196)	Pathogenic	ClinvarID:4269 PMID: 12185364
chr4:186066327-186066328	SLC25A4(NM_001151.3):c.523delC, p.Gln175ArgfsTer38	Mitochondrial DNA depletion syndrome 12B (hypertrophic cardiomyopathy) (MIM#: 615418)	Pathogenic	ClinvarID:215174 PMID: 23401503
chr2:44528267-44528268	SLC3A1(NM_000341.3):c.1136+2T>C	Cystinuria (MIM#: 220100)	Likely Pathogenic	ClinvarID:2057421 PMID: 31028937 PVS1; PM2
chr2:44539745-44539746	SLC3A1(NM_000341.3):c.1354C>T, p.Arg452Trp	Cystinuria (MIM#: 220100)	Pathogenic	ClinvarID:336205 PMID: 9186880
chr17:42328845-42328846	SLC4A1(NM_000342.3):c.2422C>T, p.Arg808Cys	Spherocytosis, type 4 (MIM#: 612653)	Likely Pathogenic	ClinvarID:1330507
chr22:32500779-32500780	SLC5A1(NM_000343.3):c.1673G>A, p.Arg558His	Glucose/galactose malabsorption (MIM#: 606824)	Likely Pathogenic – Haplotype	ClinvarID:127077 PMID: 20486940
chr5:1409229-1409230	SLC6A3(NM_001044.4): c.1408_1409delinsAG,p.Tyr470Ser	Parkinsonism-dystonia, infantile, 1 (MIM#: 613135)	Likely Pathogenic	ClinvarID:1404765 PMID: 31028937 PM2; PP1-supporting
chr19:33333131-33333132	SLC7A9(NM_014270.4):c.1166C>T, p.Thr389Met	Cystinuria (MIM#: 220100)	Pathogenic	ClinVarID: 2138267 PMID: 31028937; PMID: 25109415; PM2; PP3-moderate; PP2 PP1 – personal communication
chr13:86369403-86369404	SLITRK6(NM_032229.2):c.1240C>T, p.Gln414Ter	Deafness and myopia (MIM#: 221200)	Pathogenic	ClinvarID:88860 PMID: 23946138
chr1:38003442-38003443	SNIP1(NM_024700.3):c.1097A>G, p.Glu366Gly	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures (MIM#: 614501)	Pathogenic	ClinvarID:30717 PMID: 34570759
chr13:36903552-36903553	SPG20/SPART(NM_015087.4): c.1110delA, p.Lys370AsnfsTer30	Troyer syndrome (MIM#: 275900)	Pathogenic	ClinvarID:3457 PMID: 18413476
chr1:158590222-158590223	SPTA1(NM_003126.2): c.6154delG, p.Ala2052LeufsTer4	Elliptocytosis-2 (MIM#: 130600)	Likely Pathogenic	ClinvarID: 1723630 PMID: 31028937
chr2:86071664-86071665	ST3GAL5(NM_003896.3):c.862C>T, p.Arg288Ter	Salt and pepper developmental regression syndrome (GM3 synthase deficiency) (MIM#: 609056)	Pathogenic	ClinvarID:5556 PMID: 30691927
chr7:40498795-40498796	SUGCT(NM_001193313.2):c.985C>T, p.Arg329Trp	Glutaric aciduria III (MIM#: 231690)	Other-biochemical pseudo-deficiency	ClinvarID:1849
chr11:67814961-67814962	TCIRG1(NM_006019.3):c.1228G>A, p.Gly410Arg	Osteopetrosis, autosomal recessive 1 (MIM#: 259700)	Pathogenic	ClinvarID:2137172
chr9:71831282-71831283	TJP2(NM_004817.3):c.143T>C,p.Val48Ala	Hypercholanemia, familial 1 (MIM#: 607748)	Likely Pathogenic	ClinvarID:2907 PMID: 12704386
chr1:165737436-165737437	TMCO1(NM_019026.4):c.139_140delAG, p.Ser47insTer	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome (MIM#: 213980)	Pathogenic	ClinvarID:420165 PMID: 30556256, 20018682

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chr9:132576340-132576341	TOR1A(NM_000113.2):c.907_909delGAG, p.Glu303del	Dystonia-1, torsion (MIM#: 128100)	Pathogenic	ClinvarID:5180 PMID: 9288096
chr17:73518381-73518382	TSEN54(NM_207346.2):c.1220C>T, p.Pro407Leu	Pontocerebellar hypoplasia	Likely Pathogenic	ClinvarID:1403387
chr14:81422193-81422194	TSHR(NM_000369.2):c.170T>C, p.Leu57Pro	Hyperthyroidism, familial gestational; Hyperthyroidism, nonautoimmune; Hypothyroidism, congenital, nongoiterous, 1 (MIM#: 603370; 609152; 275200)	Likely Pathogenic	ClinvarID:1303548
chr6:116600536-116600537	TSPYL1(NM_003309.3):c.457dupG, p.Glu153GlyfsTer17	Sudden infant death with dysgenesis of the testes syndrome (MIM#: 608800)	Pathogenic	PMID: 15273283; PMID: 31028937
chr22:50656166-50656167	TUBGCP6(NM_020461.3):c.5458T>G, p.Ter1820Gly	Microcephaly and chorioretinopathy, autosomal recessive, 1 (MIM#: 251270)	Likely Pathogenic	ClinvarID:30809 PMID: 22279524
chr11:88911695-88911696, chr11:89017960-89017961	TYR(NM_000372.4): c.[575C>A;1205G>A], p.[Ser192Tyr; Arg402Gln]	Albinism, oculocutaneous, type IB (MIM#: 606952)	Pathogenic-haplotype	ClinvarID:3778, ClinvarID:3779 PMID: 35027574
chr11:88911875-88911876	TYR(NM_000372.4):c.755T>G, p.Met252Arg	Albinism, oculocutaneous, type IB (MIM#: 606952)	Likely Pathogenic	ClinvarID: 1301877 PMID: 35027574
chr2:234669154-234669155	UGT1A1(NM_000463.2):c.222C>A, p.Tyr74Ter	Crigler-Najjar syndrome, type I (MIM#: 218800)	Pathogenic	ClinvarID:437210 PMID: 31553814
chr8:100830700-100830701, chr8:100836058-100836059	VPS13B(NM_017890.4): c.[8459T>C; 9260dupT], p.[Ile2820Thr;Leu3087PhefsTer20]	Cohen syndrome (MIM#: 216550)	Pathogenic – haplotype	ClinvarID:2826 ClinvarID:2827 PMID: 15211651
chr12:6128463-6128464	VWF(NM_000552.4):c.4120C>T, p.Arg1374Cys	von Willebrand disease, type 1 (MIM#: 193400)	Pathogenic	ClinvarID:100329 PMID: 11150026
chr15:85186949-85186950	WDR73(NM_032856.2):c.888delT, p.Phe296LeufsTer26	Galloway-Mowat syndrome 1 (MIM#: 251300)	Pathogenic	ClinvarID:225244 PMID: 26070982
chr2:219747051-219747052	WNT10A(NM_025216.2):c.283G>A, p.Glu95Lys	Odontoonychodermal dysplasia (MIM#: 257980)	Pathogenic	ClinvarID:68843 PMID: 23401279
chr1:33272093-33272094	YARS1(NM_003680.3):c.499C>A, p.Pro167Thr	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2 (MIM#: 619418)	Pathogenic	ClinvarID:662296 PMID: 30304524
chr1:40723995-40723996	ZMPSTE24(NM_005857.4):c.54dupT, Ile19TyrfsTer28	Restrictive dermopathy 1 (MIM#: 275210)	Pathogenic	ClinvarID:30584 PMID: 16297189