Original Article

Carrier Testing in Known Autosomal Recessive Intellectual Disability Genes in an Iranian Healthy Individual Using Exome Sequencing

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Abstract

Background: Intellectual Disability (ID) is one of the most common disabling impairments worldwide. Autosomal recessive ID (ARID), a genetically heterogeneous disorder, is more common in countries such as Iran where the rate of consanguineous marriages is high. Considering the social-economic burden of ARID in our country, it is crucial to find out whether couples who are cousins are carriers for disease causing mutations, in order to prevent the birth of an affected child.

Methods: Using exome sequencing, we screened known ARID genes in a normal individual to identify possible mutations in heterozygous form.

Results: We identified four protein coding alleles which possibly affect protein function, in different ID genes: *PMM2, RBM28, SLC19A3*, and *VPS13B.*

Conclusion: These findings can be used to prevent the birth of children with ARID by checking the other partner for possible disease causing variants.

Keywords: Carrier screening, consanguinity, exome, intellectual disability, Iran

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Introduction

Carrier screening has been performed since 1960 to identify healthy individuals who carry mutated genes and are at an increased risk of having affected children in a number of autosomal recessive (AR) disorders including: Wilson disease, cystic fibrosis, hemoglobinopathies, spinal muscular atrophy, and Tay-sachs disease. These screenings have substantially decreased the prevalence of specific AR disorders.[1–7] In countries with high rate of consanguine marriages, carrier couples and consequently AR disorders are more prevalent.[8] Carriers may have no family history of AR disorders and be unaware of their carrier status before the birth of their first affected child.

One of the most common disabling impairments around the world is intellectual disability (ID) which affects 1%–3% of western countries population and has more victims in underdeveloped countries as a result of malnutrition, poor health care, and parental consanguinity.[8] ID is a genetically heterogeneous disorder which imposes serious social and economic problems to the society, an affected individual suffers from intellectual and adaptive behavior limitations which limit their independence in both home and society. Taking care of an affected person is estimated to cost approximately US$1 million in their lifetime.[9–11] In Iran, 30%–39% of couples are consanguine, for whom screening the carrier status in ARID genes should be performed to prevent affected pregnancies.[8,12]

Since 2011, the concept of carrier detection has evolved by means of next-generation sequencing.[13] Using this method, a substantial number of genes could be checked simultaneously. In this study, we sequenced known ARID genes in a healthy Iranian individual using exome sequencing. Data analysis was performed to identify carrier status of our subject for potentially disease causing variants in ARID genes.

Materials and Methods

Subject and DNA sample sequencing

In this study, a 26-year-old Iranian female with no familial history of ARID was selected for carrier screening. The subject's health condition was evaluated thorough obtaining her medical record, drug use, and smoking habit which were all negative. She also had no history of autosomal recessive genetic disorders with childhood onset, prevalent diseases such as diabetes and hypertension, surgery, or medication. The subject had completed routine childhood vaccination.

Blood sample was taken from the subject, after obtaining written consent according to the guidelines of the Ethics Committee, University of Social Welfare and Rehabilitation Sciences, Tehran, Iran. DNA was extracted from blood sample by salting out.[14] The highly purified DNA sample underwent library preparation, and then exome enrichment using the Agilent SureSelect human Exome kit (v.4) (Agilent Technologies, Inc., Santa Clara, CA).

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Captured regions underwent paired-end sequencing, generating 2×100-bp reads, using Ilumina Hiseq2500 (Illumina Inc., San Diego, CA).

Candidate genes

Four hundred twenty eight genes previously reported in syndromic and/or non-syndromic ARID in public databases including PubMed (http://www.ncbi.nlm.nih.gov/pubmed), OMIM (http://www.omim.org/), and Genetics Home Reference (http://ghr.nlm.nih.gov/) were chosen for this study. All the ARID genes reported until June 2014, were included in this study with no preference upon gene mutation frequency. The reported mutations in these genes were mostly missense or small insertion/deletions, but large insertion/deletion mutations were reported, as well (Supplementary Table 1).

Data analysis

Alignment and variant calling

The sequencing reads were aligned to the hg19 built of the human reference genome, using the Burrows-Wheeler Aligner (BWA).[15] Picard 1.79 was used for generating and sorting BAM file, adding or replacing read groups to it, and generating a BAM index file (http://broadinstitute.github.io/picard/). Genome Analysis Toolkit (GATK) 2.8.1 was used for refining BAM file by filtering bases with low quality, realigning insertion/deletion sites, and recalibrating base quality scores.[16,17] The depth of coverage and coverage per gene were calculated for the refined BAM file using GATK Depth of Coverage tool including "Calculate Coverage over Genes" option. Then, single-nucleotide and insertion/deletions variants were called in ARID genes using GATK Unified Genotyper.

Annotation and variant filtering

In order to identify potentially disease causing variants for which our subject was carrier, the called variants in ARID genes were annotated and underwent various filtering steps.

First, the intronic (more than two base pairs from exon boundaries), synonymous, and homozygous variants were excluded using ANNOVAR software.[18] Since the alleles which underlie ARID in human populations typically have very rare frequencies, variants reported with minor allele frequency (MAF ≥ 0.01) in public databases including the 1000 genome project and ESP6500, were eliminated, using ANNOVAR software. Truncating variants (initial codon, nonsense, splicing variants, and frameshift insertions/deletions) were considered pathogenic. The effects of missense variants on protein function were predicted using *insilico* tools including SIFT, Polyphen2-HVAR, LRT, MutationTaster, MutationAssessor, FATHMM, Radial SVM, LR, GERP++ RS, and phyloP through dbNSFP software, as well as combined annotation dependent depletion (CADD) web server.[19–21] All variants with CADD scaled score < 10 -not included in 10% most deleterious variants- and all missense variants predicted to be neutral or benign by more than six algorithms were removed. The rest of variants were checked for quality, using Integrative Genomic Viewer (IGV) 2.3.19[22,23] and variants with poor mapping quality were excluded. The frequencies of remaining variants were additionally checked in an exome sequencing database of 285 Iranian ID samples and the variants with frequency ≥ 0.01 were excluded.

Final confirmation

To eliminate potential false positive variants, the final results were confirmed by means of Sanger sequencing using BIG Dye Terminators (Applied Biosystems 3130 Genetic Analyzer; Applied Biosystems, Foster City, CA). The sequencing primers were designed using Primer3Plus (http://www.bioinformatics.nl/cgi-bin/primer3plus/primer3plus.cgi/) and Oligoanalyzer 3.1 (http://eu.idtdna.com/analyzer/applications/oligoanalyzer/). The Sanger sequencing results were analyzed by CodonCode Aligner 5.0.1 (CodonCode Corp., Dedham, MA).

Results

An average of 99.32%, 98.23%, and 95.85% of bases were covered at 1X, 10X, and 20X, respectively, within the exome enrichment kit targeted regions. An average of 94.74%, 88.3%, and 85.46% of bases were covered at 1X, 10X, and 20X, respectively, within the ARID-related coding refseq exons. The average depth of coverage for all ARID genes was 102.24x (ranges between 0 and 323) (Supplementary Figure 1).

A total of 1,016 variants were identified, including 964 missense and 52 insertion/deletion variants. Out of 1,016 variants, a large fraction (1,012 variants) was filtered out in different filtering steps. Initially, intronic variants were excluded and 578 variants in exonic or splice site regions were kept, among which 349 variants were heterozygous. By removing synonymous variants, the total number of variants was reduced to 126, which were then checked for allele frequency. Eighteen variants with MAF ≤0.01, which were not reported in homozygous state, remained for function prediction step. Scaled CADD score for 12 variants was ≥10, nine of which were predicted to be damaging according to dbNSFP output. The quality checking of the nine variants revealed that five variants-Q418E in *SLC19A3* gene (OMIM#606152), K688N in *RBM28* gene (OMIM#612074), Q416H in *VPS13B* gene (OMIM#607817), V206L in *PC* gene (OMIM#608786), and N216D in *PMM2* gene (OMIM#601785) - were mapped with high quality (Table 1). Evaluating the frequency of these five variants in our database of 285 Iranian samples excluded the V206L variant in *PC* gene. This variant was identified in six Iranian individuals, which suggests that in the Iranian population the V206L in *PC* is not as rare as in other populations (Table 1). The filtering steps resulted in identifying four protein coding heterozygous changes which predicted to affect protein function in conserved regions of *PMM2*, *RBM28*, *SLC19A3*, and *VPS13B* genes (Figure 1A).To confirm these results, each mutated region was sequenced using a pair of primers (Table 1). Sanger sequencing results confirmed the presence of all four variants (Figure 1B).

Discussion

Before next generation sequencing (NGS) era, carrier-screening tests were mostly performed in individuals with a family history of genetic disorders following identification of a specific mutation in an affected member of the family. Population-based genetic screening was limited to disorders with a commonly seen mutation like Cystic Fibrosis, which is recommended to all pregnant women in the US,[24] or disorders commonly seen in a specific population like β-thalassemia in Iran. β-thalassemia is 2.6 times more prevalent in Iran than in Western countries. The screening of couples before getting married alongside with the implement

Table 1. Detected variants and their function predictions, frequencies, and Sanger sequencing primers.

Gene	Transcript	Variant Position	Reference allele >Alternative allele	Amino Acid Change	Minor Allele Frequency in ESP6500	CADD Scaled Score	dbNSF Predictions*	Number of Observed Variant(s) in 285 Iranian Individuals	Sanger Sequencing Forward Primer	Sanger Sequencing Reverse Primer
SLC19A3	NM_025243	2:228552944	G>C	Q418E	0.000	27.1	D,D,D,D,M,D, D,D,5.44,2.831	0	5'TGACACAAATACCCCTGTGG	5'AATGCTGACTGGCAAGTTGA
RBM28	NM_001166135	7:127953309	T>A	K688N	0.000	13.8	D,B,D,D,M,T, T,T,0.619,0.115	0	5'GCTTTCAGAGGGTCTGTTGC	5'ACCTGCTCGGACGATAATTG
PMM2	NM_000303	16:8941587	A>G	N216D	0.000	24.2	D,D,D,D,H,D, D,D,5.21,2.088	0	5'AGAAACTCTCCCTGCCCAGT	5'TCCCACGTTAGGAGAACAGC
VPS13B	NM_015243	8:100146901	G>T	Q416H	0.002	15.75	D,D,D,D,L,T,T, T,2.58,0.319	1 heterozygous	5'GCACCTGCTTTCCTGCTTAAT	5'TGTTAGAGGCTTTCCTTGTAGAATC
PC	NM_022172	11:66638540	C>A	V206L	0.002	19.04	D,B,D,D,L,D, D,D,4.99,2.327	6 heterozygous	-	-

* The predictions of following software are listed from left to right: SIFT, Polyphen2-HVAR, LRT, MutationTaster, MutationAssessor, FATHMM, Radial SVM, LR, GERP++ RS, and phyloP. GERP score is ranged from -12.36 to +6.18. Higher scores indicate more deleterious changes, and a site with a score of ≥ 2, is considered to be conserved. Positive phyloP scores are representative of conserved sites, and higher scores are more deleterious. D = damaging, P = possibly-damaging, B = benign, H = high, M = medium, L = low, T = tolerate.

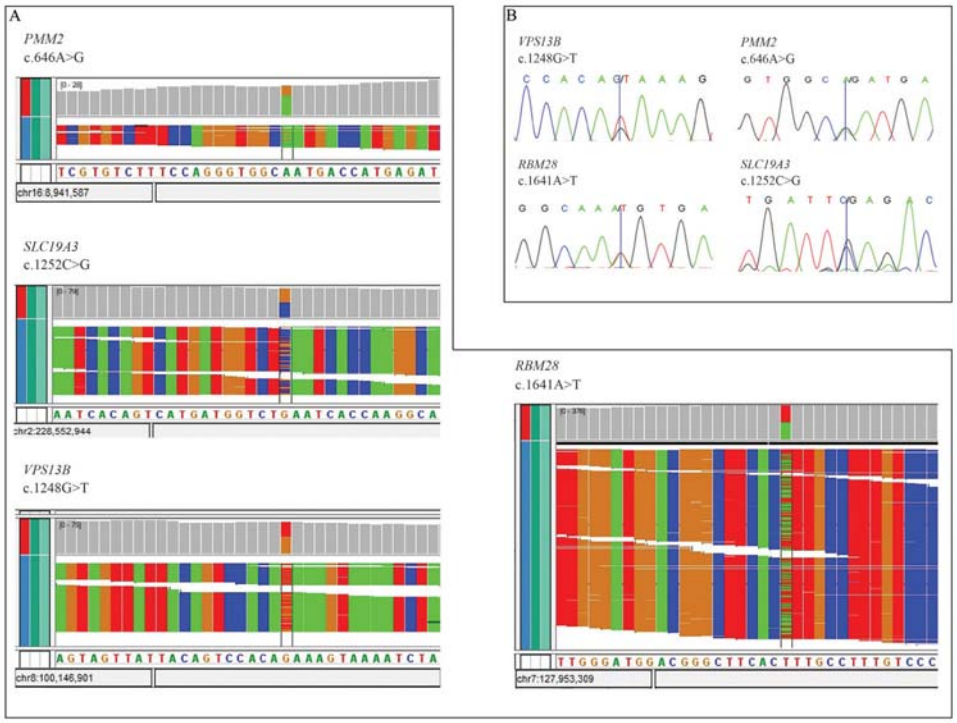

Figure 1. Images of four potentially disease causing variants **A)** Visualization of exome sequencing data through IGV software **B)** Sanger sequencing chromatograms.

of prevention programs has resulted in considerable reduction of this condition in Iranian population with 82.03% success rate.[25,26]

Many of the carriers are not aware of their genetic condition, and they usually find out about it after the birth of their first affected child. NGS technique has facilitated carrier screening in a large number of genes, including most clinically severe AR disorders genes which were not screened before,[13] with low cost. In 2011, Bell, *et al.* investigated carrier status for 448 sever recessive genetic diseases of childhood in 104 subjects using NGS technique.[13] The subjects were carriers for zero to seven disorders (2.8 disorders in average). In 2013, Lazarin, *et al.* performed carrier detection for 23,452 individuals from different ethnicities in 417 disease causing mutations of 108 recessive disorders.[27] The results suggested that 24% of subjects were carriers for at least one genetic disorder, while 5.2% were carriers for more than one disorder. They suggested that since being carrier for specific recessive disorders is not limited to a specific ethnicity, an identical thorough carrier test can be performed for different ethnicities. These studies have shown that, the carrier frequencies in not routinely screened diseases are high and carrier screening should be performed for these diseases, as well.[13,27] Later, Tabor, *et al.* analyzed exome sequencing data of 6517 individuals from African American or European American ethnicities (ESP6500) for pathogenic variants in 31 Mendelian disorders. By taking strict filtering steps, they showed that 45% of individuals carried at least one deleterious allele.[28]

In this study, we utilized NGS technology to identify the carrier status of a normal subject in ARID genes. Considering the fact that there is no specific gene(s) responsible for the major fraction of ARID cases,[12] all ARID genes collected from public databases were included in this study, regardless of their frequencies. Data analysis and filtering steps resulted in removing 99.6% of called variants in ARID genes. Four potentially disease causing alleles were identified in 4 out of 428 genes investigated in this study: *PMM2*, *SLC19A3*, *RBM28*, and *VPS13B*. These genes are involved in syndromic ID disorders.

Congenital disorder of glycosylation

PMM2 defects lead to congenital disorder of glycosylation, type Ia (CDG1A) (OMIM#212065) in different ethnicities. This disorder has three stages: infantile multisystem defects, late infantile and childhood ataxia-intellectual disability, and adult stable disability. In 2011, a comprehensive study on etiology of ID in 136 consanguineous Iranian families revealed *PMM2* missense mutation, p.Y106F, in a big family with three affected children who were diagnosed with CDG1A.[12]

Thiamine metabolism dysfunction syndrome 2, Wernicke's-like encephalopathy, and leigh syndrome

Missense or small insertion/deletion mutations in *SLC19A3* cause different symptoms and disorders, namely Thiamine metabolism dysfunction Syndrome 2 (THMD2) (OMIM#607483), Wernicke's-like Encephalopathy (OMIM#606152), and Leigh syndrome (OMIM#256000), in different individuals. The major symptoms of THMD2 are recurrent episodes of encephalopathy, generalized dystonia, and epilepsy.[29] The characteristic of Wernicke's-like Encephalopathy is status epilepticus which develops in the second decade of life.[30] Leigh syndrome is a severe neurodegenerative disorder with progressive psychomotor progression, and eventually leads to death.[31] In another neurological disorder, symptoms such as epileptic spasms in early infancy, severe psychomotor retardation, and progressive brain atrophy are reported in patients with homozygous mutations in *SLC19A3*.[32]

Alopecia, neurologic defects, and endocrinopathy

In 2008, a missense mutation, p.L351P, in *RBM28* gene was

identified in a consanguineous Palestinian Arab family with alopecia, neurologic defects, and endocrinopathy syndrome (ANES) (OMIM#612079). The five siblings of this family are the only reported cases of ANES. The main characteristics of ANES are skin involvement, neurological defects including moderate to severe ID, and endocrine system involvement.[33]

Cohen syndrome

Defected *VPS13B* is the only reported cause of Cohen syndrome (OMIM#216550), which adversely affects different parts of the body. The major features of syndrome are ID and typical Cohen syndrome facial characteristics.[34]

The carrier of the potentially disease causing changes in these genes may be at risk of giving birth to children with ARID syndrome. In order to prevent the birth of affected children, the other partner can be checked for these four genes.

In this study, we focused on the most frequent genetic disabling impairment and screened all the genes which were previously reported in AR form of this disorder. Generating a comprehensive gene list was the advantage of this study, in comparison with previous ones, that despite targeting a wide range of disorders, analysis were limited to the previously reported mutations or a portion of the genes reported in a specific heterogeneous disorder.[13,27]

Considering the high rate of AR disorders in our country, carrier screening in a consanguineous couple can be performed for other AR disorders, as well. Setting up a carrier detection panel for severe AR disorders in the Iranian population depends on further studies and identifying common AR disorders and their related genes in our country.

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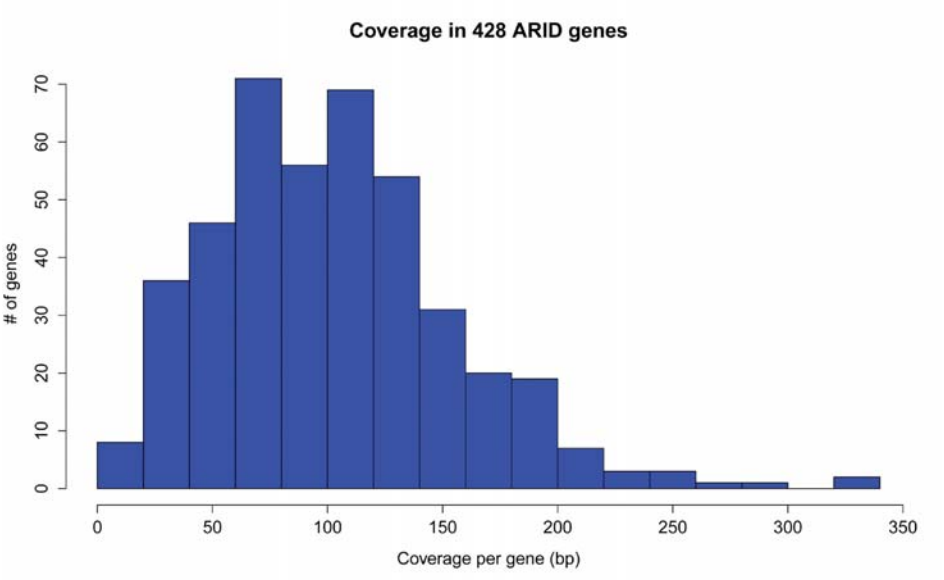

Supplementary Figure 1. Histogram of coverage in 428 ARID genes.

Supplementary Table 1. List of 428 ARID genes which were analyzed in this study.

Gene	Transcript	Syndromic/ Nonsyndromic	Reference URL address	Reference URL address	Reference URL address
AAAS	NM_001173466	S	Mol Genet Metab. 2007 Dec;92(4):359-63. Epub 2007 Oct 2	Nat Genet. 2000 Nov;26(3):332-5.	
AASS	NM_005763	S	Am J Hum Genet. 2000 Jun;66(6):1736-43. Epub 2000 Apr 20.		
ABCC8	NM_000352	S	http://ghr.nlm.nih.gov/gene/ABCC8, Kapoor et al., 2009	Nat Clin Pract Endocrinol Metab. 2009 Feb;5(2):101-12. doi: 10.1038/ncpendmet1046	
ABHD12	NM_015600	S	Am J Hum Genet. 2010 Sep 10;87(3):410-7. doi: 10.1016/j.ajhg.2010.08.002		
ABHD5	NM_016006	S	http://ghr.nlm.nih.gov/condition/chanarin-dorfman-syndrome	Am J Hum Genet. 2001 Nov;69(5):1002-12. Epub 2001 Oct 2.	
ACADSB	NM_001609	S	J Med Case Rep. 2007 Sep 20;1:98.		
ACBD6	NM_032360	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
ACOX1	NM_001185039	S	Am J Med Genet A. 2008 Jul 1;146A(13):1676-81. doi: 10.1002/ajmg.a.32298.	http://www.omim.org/entry/609751?search=acox1&highlight=acox1	

ADAMTS10	NM_030957	S	Am J Med Genet A. 2003 Dec 1;123A(2):204-7.	http://www.omim.org/entry/277600	
ADAT3	NM_138422	S	J Med Genet. 2013 Jul;50(7):425-30. doi: 10.1136/jmedgenet-2012-101378. Epub 2013 Apr 25		
ADK	NM_001202450	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
ADRA2B	NM_000682	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
ADSL	NM_000026	S	Am J Med Genet A. 2003 Jul 15;120A(2):185-90.		
AGA	NM_000027	S	J Child Neurol. 2014 Jan;29(1):36-42. doi: 10.1177/0883073812469049. Epub 2012 Dec 26	EMBO J. 1991 Jan;10(1):51-8.	
AGPAT2	NM_001012727	S	J Clin Endocrinol Metab. 2003 Oct;88(10):4840-7.	Nat Genet. 2002 May;31(1):21-3. Epub 2002 Apr 22.	
AGPS	NM_003659	S	Hum Mol Genet. 1998 May;7(5):847-53.	Clin Genet. 2005 Feb;67(2):107-33.	http://www.omim.org/entry/600121
AHCY	NM_000687	S	http://ghr.nlm.nih.gov/condition/hypermethioninemia	J Pediatr. 1990 Aug;117(2 Pt 1):220-6.	
AHI1	NM_001134830	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
ALDH18A1	NM_001017423	S	Eur J Hum Genet. 2008 Oct;16(10):1176-86. doi: 10.1038/ejhg.2008.91. Epub 2008 May 14.		
ALDH1A3	NM_000693	S	Clin Genet. 2013 Sep 11. doi: 10.1111/cge.12277. [Epub ahead of print]		
ALDH3A2	NM_001031806	S	Arch Neurol. 2006 Feb;63(2):278-80		
ALDH4A1	NM_001161504	S	Hum Mol Genet. 1998 Sep;7(9):1411-5.		
ALDH5A1	NM_170740	S	Neurology. 2012 Jul 3;79(1):47-54. doi: 10.1212/WNL.0b013e31825dcf71. Epub 2012 Jun 20.		
ALDOA	NM_000034	S	Am J Hematol. 1981 Dec;11(4):425-37.	http://www.omim.org/entry/103850?search=aldoa&highlight=aldoa	
ALS2	NM_020919	S	Ann Neurol. 2003 Jan;53(1):144-5.	http://www.omim.org/entry/607225?search=606352/%20607225&highlight=606352%20607225%20606352/	
ALX1	NM_006982	S	Am J Hum Genet. 2010 May 14;86(5):789-96. doi: 10.1016/j.ajhg.2010.04.002. Epub 2010 May 6.		
ALX4	NM_021926	S	Hum Mol Genet. 2009 Nov 15;18(22):4357-66. doi: 10.1093/hmg/ddp391. Epub 2009 Aug 19.		
AMT	NM_000481	S	Neurology. 2000 Feb 8;54(3):754-6.	http://www.omim.org/entry/605899	
ANKH	NM_054027	S	J Clin Endocrinol Metab. 2011 Jan;96(1):E189-98. doi: 10.1210/jc.2010-1539. Epub 2010 Oct 13.		

AP1S1	NM_001283	S	PLoS Genet. 2008 Dec;4(12):e1000296. doi: 10.1371/journal.pgen.1000296. Epub 2008 Dec 5.	
AP3B1	NM_003664	S	Blood. 2006 Jul 1;108(1):362-9. Epub 2006 Mar 14.	
AP4B1	NM_006594	S	Am J Hum Genet. 2011 Jun 10;88(6):788-95. doi: 10.1016/j.ajhg.2011.04.019. Epub 2011 May 27.	
AP4E1	NM_007347	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
AP4M1	NM_004722	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
AP4S1	NM_007077	S	Am J Hum Genet. 2011 Jun 10;88(6):788-95. doi: 10.1016/j.ajhg.2011.04.019. Epub 2011 May 27.	
ARFGEF2	NM_006420	S	Nat Genet. 2004 Jan;36(1):69-76. Epub 2003 Nov 30.	
ARG1	NM_000045	S	Hum Genet. 1995 Sep;96(3):255-60.	
ARL14EP (C11orf46)	NM_152316	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
ARL6 (BBS3)	NM_032146	S	Nat Genet. 2004 Sep;36(9):989-93. Epub 2004 Aug 15.	
ASCC3	NM_006828	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
ASCL1	NM_004316	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
ASL	NM_001024943	S	Hum Genet. 2002 Oct;111(4-5):350-9. Epub 2002 Aug 14	
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ASPM	NM_001206846	S	J Med Genet. 2005 Sep;42(9):725-9.	
ATIC	NM_004044	S	Am J Hum Genet. 2004 Jun;74(6):1276-81. Epub 2004 Apr 26.	
ATN1	NM_001007026	S	Neurology. 1995 Oct;45(10):1934-6.	
ATP13A2	NM_001141974	S	http://www.ncbi.nlm.nih.gov/books/NBK1428/	Parkinsonism Relat Disord. 2011 Feb;17(2):135-8. doi: 10.1016/j.parkreldis.2010.10.011. Epub 2010 Nov 20.
ATP6V0A2	NM_012463	S	Hum Genet. 2012 Nov;131(11):1761-73. doi: 10.1007/s00439-012-1197-8. Epub 2012 Jul 8.	Nat Genet. 2008 Jan;40(1):32-4. Epub 2007 Dec 23.
ATP8A2	NM_016529	S	Eur J Hum Genet. 2013 Mar;21(3):281-5. doi: 10.1038/ejhg.2012.170. Epub 2012 Aug 15.	Eur J Hum Genet. 2010 Dec;18(12):1360-3. doi: 10.1038/ejhg.2010.126. Epub 2010 Aug 4.
ATR	NM_001184	S	Nat Genet. 2003 Apr;33(4):497-501. Epub 2003 Mar 17	
AUH	NM_001698	S	http://ghr.nlm.nih.gov/condition/3-methylglutaconic-aciduria	
B3GALT6	NM_080605	S	Am J Hum Genet. 2013 Jun 6;92(6):935-45. doi: 10.1016/j.ajhg.2013.04.016. Epub 2013 May 9	
B3GALTL	NM_194318	S	Am J Hum Genet. 2006 Sep;79(3):562-6. Epub 2006 Jul 19.	

BBIP1 (BBS18)	NM_001195306	S	J Med Genet. 2014 Feb;51(2):132-6. doi: 10.1136/jmedgenet-2013-101785. Epub 2013 Sep 11.	http://www.ncbi.nlm.nih.gov/pubmed/20301537	
BBS1	NM_024649	S	J Med Genet. 2010 Apr;47(4):262-7. doi: 10.1136/jmg.2009.071365. Epub 2009 Sep 24.	Am J Hum Genet. 2003 May;72(5):1187-99. Epub 2003 Apr 3.	
BBS10	NM_024685	S	J Med Genet. 2010 Apr;47(4):262-7. doi: 10.1136/jmg.2009.071365. Epub 2009 Sep 24.	J Med Genet. 2010 Dec;47(12):848-52. doi: 10.1136/jmg.2010.079392. Epub 2010 Aug 30.	
BBS12	NM_152618	S	J Med Genet. 2010 Apr;47(4):262-7. doi: 10.1136/jmg.2009.071365. Epub 2009 Sep 24.	Am J Med Genet A. 2010 Oct;152A(10):2666-9. doi: 10.1002/ajmg.a.33650	
BBS2	NM_031885	S	J Med Genet. 2010 Apr;47(4):262-7. doi: 10.1136/jmg.2009.071365. Epub 2009 Sep 24.	Nat Genet. 2006 May;38(5):521-4. Epub 2006 Apr 2.	Eur J Hum Genet. 2006 Nov;14(11):1195-203. Epub 2006 Jul 5.
BBS4	NM_033028	S	J Med Genet. 2010 Apr;47(4):262-7. doi: 10.1136/jmg.2009.071365. Epub 2009 Sep 24.	J Med Genet. 2010 Apr;47(4):236-41. doi: 10.1136/jmg.2009.070755. Epub 2009 Oct 26.	Hum Mol Genet. 1995 Jan;4(1):9-13.
BBS5	NM_152384	S	J Med Genet. 2010 Apr;47(4):262-7. doi: 10.1136/jmg.2009.071365. Epub 2009 Sep 24.	Am J Hum Genet. 1999 Mar;64(3):900-4.	
BBS7	NM_018190	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
BBS9	NM_014451	S	http://www.ncbi.nlm.nih.gov/books/NBK1363/		
BCKDHA	NM_000709	S	J Inherit Metab Dis. 2007 Nov;30(6):903-9. Epub 2007 Oct 8.		
BCKDHB	NM_183050	S	J Inherit Metab Dis. 2007 Nov;30(6):903-9. Epub 2007 Oct 8.		
BCKDK	NM_005881	S	Science. 2012 Oct 19;338(6105):394-7. doi: 10.1126/science.1224631. Epub 2012 Sep 6.		
BIN1	NM_139348	S	Neurology. 2010 Feb 9;74(6):519-21. doi: 10.1212/WNL.0b013e3181cef7f9.	http://www.omim.org/entry/601248?search=601248&highlight=601248	
BOLA3	NM_212552	S	Brain. 2014 Feb;137(Pt 2):366-79. doi: 10.1093/brain/awt328. Epub 2013 Dec 11.		
BSCL2	NM_001130702	S	J Clin Endocrinol Metab. 2003 Oct;88(10):4840-7.	Nat Genet. 2002 May;31(1):21-3. Epub 2002 Apr 22.	
BUB1B	NM_001211	S	http://www.omim.org/entry/257300	Am J Med Genet A. 2006 Feb 15;140(4):358-67.	
C12orf57	NM_138425	NS/S	Am J Med Genet A. 2013 Jun;161A(6):1207-13. doi: 10.1002/ajmg.a.35850. Epub 2013 Apr 30		
C12orf65	NM_001194995	S	Eur J Hum Genet. 2014 Jan 15. doi: 10.1038/ejhg.2013.284. [Epub ahead of print]	Eur J Med Genet. 2013 Nov;56(11):599-602. doi: 10.1016/j.ejmg.2013.09.010. Epub 2013 Sep 28	
CA2	NM_000067	S	Hum Mutat. 1992;1(4):288-92.	Hum Mutat. 1997;9(5):383-7.	
CA5A	NM_001739	S	Am J Hum Genet. 2014 Mar 6;94(3):453-61. doi: 10.1016/j.ajhg.2014.01.006. Epub 2014 Feb 13.		
CA8	NM_004056	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		

CACNA1G	NM_198397	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
CAPN10	NM_023083	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
CASC5	NM_144508	S	Hum Mol Genet. 2012 Dec 15;21(24):5306-17. doi: 10.1093/hmg/dds386. Epub 2012 Sep 13.	
CASP2	NM_032982	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
CASR	NM_000388	S	Mol Biol Rep. 2012 Mar;39(3):2395-400. doi: 10.1007/s11033-011-0990-0. Epub 2011 Jun 12	
CBS	NM_000071	S	Thromb Haemost. 2000 Apr;83(4):554-8.	
CC2D1A	NM_017721	NS	J Med Genet. 2006 Mar;43(3):203-10. Epub 2005 Jul 20.	
CC2D2A	NM_001080522	S	Am J Hum Genet. 2008 Apr;82(4):1011-8. doi: 10.1016/j.ajhg.2008.01.021	
CCBE1	NM_133459	S	Nat Genet. 2009 Dec;41(12):1272-4. doi: 10.1038/ng.484.	
CCNA2	NM_001237	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
CD96	NM_005816	S	Am J Hum Genet. 2007 Oct;81(4):835-41. Epub 2007 Aug 27	
CDK5RAP2	NM_001011649	S	Nat Genet. 2005 Apr;37(4):353-5. Epub 2005 Mar 27.	
CDK6	NM_001259	S	Hum Mol Genet. 2013 Dec 20;22(25):5199-214. doi: 10.1093/hmg/ddt374. Epub 2013 Aug 4	
CENPJ	NM_018451	S	Nat Genet. 2005 Apr;37(4):353-5. Epub 2005 Mar 27.	J Hum Genet. 2006;51(9):760-4. Epub 2006 Aug 10
CEP135	NM_025009	S	Am J Hum Genet. 2012 May 4;90(5):871-8. doi: 10.1016/j.ajhg.2012.03.016. Epub 2012 Apr 19.	
CEP152	NM_001194998	S	Am J Hum Genet. 2010 Jul 9;87(1):40-51. doi: 10.1016/j.ajhg.2010.06.003	Nat Genet. 2011 Jan;43(1):23-6. doi: 10.1038/ng.725. Epub 2010 Dec 5.
CEP290 (BBS14)	NM_025114	S	Nat Genet. 2008 Apr;40(4):443-8. doi: 10.1038/ng.97. Epub 2008 Mar 9.	Nat Genet. 2006 Jun;38(6):674-81. Epub 2006 May 7.
CEP63	NM_025180	S	Nat Genet. 2011 Oct 9;43(11):1147-53. doi: 10.1038/ng.971.	
CHKB	NM_005198	S	Am J Hum Genet. 2011 Jun 10;88(6):845-51. doi: 10.1016/j.ajhg.2011.05.010.	
CHRNA7	NM_000746	S	Am J Med Genet A. 2014 Apr 3. doi: 10.1002/ajmg.a.36535. [Epub ahead of print]	
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CLN3	NM_001042432	S	http://www.ncbi.nlm.nih.gov/books/NBK1428/	Am J Hum Genet. 1997 Aug;61(2):310-6.	
CLN5	NM_006493	S	http://www.ncbi.nlm.nih.gov/books/NBK1428/	Neurology. 2010 Feb 16;74(7):565-71. doi: 10.1212/WNL.0b013e3181cff70d	
CLN6	NM_017882	S	http://www.ncbi.nlm.nih.gov/books/NBK1428/	Am J Hum Genet. 2011 May 13;88(5):566-73. doi: 10.1016/j.ajhg.2011.04.004. Epub 2011 May 5.	Am J Hum Genet. 2002 Feb;70(2):537-42. Epub 2001 Nov 27.
CLN8	NM_018941	S	Nat Genet. 1999 Oct;23(2):233-6.		
CNKSR1	NM_006314	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
CNNM2	NM_199076	S	PLoS Genet. 2014 Apr 3;10(4):e1004267. doi: 10.1371/journal.pgen.1004267. eCollection 2014.		
CNTNAP2	NM_014141	S	N Engl J Med. 2006 Mar 30;354(13):1370-7	Am J Hum Genet. 2009 Nov;85(5):655-66. doi: 10.1016/j.ajhg.2009.10.004. Epub 2009 Nov 5.	
COG5	NM_181733	S	Hum Mol Genet. 2009 Nov 15;18(22):4350-6. doi: 10.1093/hmg/ddp389. Epub 2009 Aug 18		
COL18A1	NM_130445	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
COQ5	NM_032314	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
COX10	NM_001303	S	Hum Mol Genet. 2000 May 1;9(8):1245-9.		
COX14 (C12ORF62)	NM_001257133	S	Am J Hum Genet. 2012 Jan 13;90(1):142-51. doi: 10.1016/j.ajhg.2011.11.027.		
COX15	NM_004376	S	J Med Genet. 2004 Jul;41(7):540-4.		
COX6B1	NM_001863	S	Am J Hum Genet. 2008 Jun;82(6):1281-9. doi: 10.1016/j.ajhg.2008.05.002. Epub 2008 May 22.		
CRADD	NM_003805	NS	PLoS One. 2012;7(1):e28936. doi: 10.1371/journal.pone.0028936. Epub 2012 Jan 17.		
CRBN	NM_001173482	NS	Neurology. 2004 Nov 23;63(10):1927-31.		
CTDP1	NM_004715	S	Nat Genet. 2003 Oct;35(2):185-9. Epub 2003 Sep 21		
CTSA	NM_000308	S	J Hum Genet. 2000;45(4):200-6.		
CTSD	NM_001909	S	http://www.ncbi.nlm.nih.gov/books/NBK1428/		
CYB5R3	NM_001129819	S	Blood. 1995 Apr 15;85(8):2254-62.		
CYP27A1	NM_000784	S	Am J Med Genet A. 2005 Dec 1;139A(2):114-7		

CYP2U1	NM_183075	S	J Neurol. 2014 Feb;261(2):373-81. doi: 10.1007/s00415-013-7206-6. Epub 2013 Dec 13.
D2HGDH	NM_152783	S	Am J Hum Genet. 2005 Feb;76(2):358-60. Epub 2004 Dec 17.
DAG1	NM_001177643	S	N Engl J Med. 2011 Mar 10;364(10):939-46. doi: 10.1056/NEJMoa1006939
DBT	NM_001918	S	J Inherit Metab Dis. 2007 Nov;30(6):903-9. Epub 2007 Oct 8.
DCAF17 (C2ORF37)	NM_001164821	S	Pediatr Dermatol. 2014 Jan;31(1):83-7. doi: 10.1111/pde.12219. Epub 2013 Sep 9
DDC (AADC)	NM_000790	S	Neurology. 2010 Jul 6;75(1):64-71. doi: 10.1212/WNL.0b013e3181e620ae. Epub 2010 May 26.
DDHD2	NM_001164234	S	J Neurol. 2014 Feb;261(2):373-81. doi: 10.1007/s00415-013-7206-6. Epub 2013 Dec 13.
DEAF1	NM_021008	S	Am J Med Genet A. 2014 Mar 25. doi: 10.1002/ajmg.a.36482. [Epub ahead of print]
DHCR7	NM_001163817	S	Proc Natl Acad Sci U S A. 1998 Jul 7;95(14):8181-6.
DNAJC19	NM_145261	S	J Med Genet. 2006 May;43(5):385-93. Epub 2005 Jul 31.
DNAJC6	NM_014787	S	Mol Genet Metab. 2012 Jul;106(3):345-50. doi: 10.1016/j.ymgme.2012.04.026. Epub 2012 May 10.
DNMT3B	NM_001207055	S	Nature. 1999 Nov 11;402(6758):187-91.
DPM1	NM_003859	S	J Clin Invest. 2000 Jan;105(2):191-8.
DPM2	NM_003863	S	Ann Neurol. 2012 Oct;72(4):550-8. doi: 10.1002/ana.23632.
DPYD (DPD)	NM_000110	S	Hum Genet. 2009 Jun;125(5-6):581-90. doi: 10.1007/s00439-009-0653-6. Epub 2009 Mar 19.
DUOX2 (THOX2)	NM_014080	S	N Engl J Med. 2002 Jul 11;347(2):95-102.
DYM	NM_017653	S	Am J Hum Genet. 2003 Feb;72(2):419-28. Epub 2002 Dec 16.
EDNRB	NM_001122659	S	J Med Genet. 1999 Jun;36(6):485-9
EEF1B2	NM_001037663	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.
EFEMP2 (FBLN4)	NM_016938	S	http://ghr.nlm.nih.gov/condition/cutis-laxa
EFTUD2	NM_001142605	S	Orphanet J Rare Dis. 2013 Jul 24;8:110. doi: 10.1186/1750-1172-8-110
EIF2AK3	NM_004836	S	Nat Genet. 2000 Aug;25(4):406-9

EIF2B1	NM_001414	S	J Hum Genet. 2009 Feb;54(2):74-7. doi: 10.1038/jhg.2008.10. Epub 2009 Jan 16.	
EIF2B2	NM_014239	S	Gene. 2012 Apr 1;496(2):141-3. doi: 10.1016/j.gene.2011.12.047. Epub 2012 Jan 17	
EIF2B3	NM_020365	S	J Hum Genet. 2009 Feb;54(2):74-7. doi: 10.1038/jhg.2008.10. Epub 2009 Jan 16.	
EIF2B4	NM_001034116	S	J Hum Genet. 2009 Feb;54(2):74-7. doi: 10.1038/jhg.2008.10. Epub 2009 Jan 16.	
EIF2B5	NM_003907	S	J Hum Genet. 2009 Feb;54(2):74-7. doi: 10.1038/jhg.2008.10. Epub 2009 Jan 16.	
ELOVL4	NM_022726	S	Am J Hum Genet. 2011 Dec 9;89(6):745-50. doi: 10.1016/j.ajhg.2011.10.011. Epub 2011 Nov 17.	
ELP2	NM_001242879	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
ENTPD1	NM_001098175	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
ERCC2	NM_000400	S	Hum Mol Genet. 2001 Oct 15;10(22):2539-47.	
ERCC3	NM_000122	S	http://ghr.nlm.nih.gov/gene/ERCC3	
ERCC5	NM_001204425	S	J Invest Dermatol. 2002 Feb;118(2):344-51	
ERCC6	NM_000124	S	Am J Hum Genet. 2000 Apr;66(4):1221-8. Epub 2000 Mar 15	
ERCC8	NM_000082	S	J Hum Genet. 2006;51(8):701-5. Epub 2006 Jul 25.	
ERLIN2	NM_007175	S	Neurogenetics. 2011 Nov;12(4):333-6. doi: 10.1007/s10048-011-0291-8. Epub 2011 Jul 28	Hum Mol Genet. 2011 May 15;20(10):1886-92. doi: 10.1093/hmg/ddr070. Epub 2011 Feb 17.
ESCO2	NM_001017420	S	Am J Hum Genet. 2005 Dec;77(6):1117-28. Epub 2005 Oct 31.	Hum Mol Genet. 2008 Jul 15;17(14):2172-80. doi: 10.1093/hmg/ddn116. Epub 2008 Apr 14.
EXOSC3	NM_016042	S	Neurogenetics. 2013 Nov;14(3-4):247-50. doi: 10.1007/s10048-013-0371-z. Epub 2013 Aug 24	
FAM126A	NM_032581	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
FASN	NM_004104	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
FASTKD2	NM_014929	S	Am J Hum Genet. 2008 Sep;83(3):415-23. doi: 10.1016/j.ajhg.2008.08.009. Epub 2008 Sep 4.	
FBLN5	NM_006329	S	http://ghr.nlm.nih.gov/condition/cutis-laxa	
FBXO31	NM_024735	NS	Hum Genet. 2014 Mar 13. [Epub ahead of print]	

FGFR3	NM_000142	S	Am J Hum Genet. 2006 Nov;79(5):935-41. Epub 2006 Sep 26.		
FKRP	NM_001039885	S	http://repositorium.sdum.uminho.pt/bitstream/1822/20437/1/Bessa%20C_InTech_B2012.pdf		
FKTN	NM_006731	S	Clin Genet. 2008 Feb;73(2):139-45. doi: 10.1111/j.1399-0004.2007.00936.x. Epub 2007 Dec 19		
FOLR1	NM_016729	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
FOXL2	NM_023067	S	Hum Genet. 2007 Mar;121(1):107-12. Epub 2006 Nov 7.		
FRY	NM_023037	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
FTCD	NM_006657	S	Hum Mutat. 2003 Jul;22(1):67-73.		
FUCA1	NM_000147	S	Am J Hum Genet. 1988 Nov;43(5):756-63.		
GAD1	NM_013445	S	BMC Neurol. 2004 Nov 30;4(1):20.		
GALE	NM_001127621	S	Mol Genet Metab. 1998 Jan;63(1):26-30.		
GALK1	NM_000154	S	Mol Genet Metab. 1998 Jan;63(1):26-30.		
GALT	NM_000155	S	Mol Genet Metab. 1998 Jan;63(1):26-30.		
GAMT	NM_138924	S	Neurology. 2006 Nov 14;67(9):1713-4.	http://www.omim.org/entry/601240?search=GAMT&highlight=gamt	
GATM	NM_001482	S	Mol Genet Metab. 2010 Oct-Nov;101(2-3):228-32. doi: 10.1016/j.ymgme.2010.06.021. Epub 2010 Jul 7.		
GBA2	NM_020944	S	J Neurol. 2014 Feb;261(2):373-81. doi: 10.1007/s00415-013-7206-6. Epub 2013 Dec 13.		
GCDH	NM_000159	S	http://www.omim.org/entry/231670,	http://www.ncbi.nlm.nih.gov/pubmed/8139602	
GCH1	NM_000161	S	PLoS One. 2014 Apr 4;9(4):e94100. doi: 10.1371/journal.pone.0094100. eCollection 2014.	http://ghr.nlm.nih.gov/condition/tetrahydrobiopterin-deficiency	Zhonghua Yi Xue Yi Chuan Xue Za Zhi. 2007 Apr;24(2):210-2
GCHFR	NM_005258	S	http://www.ncbi.nlm.nih.gov/pubmed/20301531		
GCSH	NM_004483	S	PLoS One. 2014 Apr 4;9(4):e94100. doi: 10.1371/journal.pone.0094100. eCollection 2014.		
GLB1	NM_001079811	S	http://www.ncbi.nlm.nih.gov/books/NBK164500/	http://www.ncbi.nlm.nih.gov/pubmed/24156116	
GLDC	NM_000170	S	http://www.ncbi.nlm.nih.gov/pubmed/20301531		
GLRA1	NM_000171	S	Pediatr Neurol. 2012 Feb;46(2):89-93. doi: 10.1016/j.pediatrneurol.2011.11.008.		

GLRX5	NM_016417	S	Brain. 2014 Feb;137(Pt 2):366-79. doi: 10.1093/brain/awt328. Epub 2013 Dec 11.	
GLYCTK	NM_145262	S	Hum Mutat. 2010 Dec;31(12):1280-5. doi: 10.1002/humu.21375. Epub 2010 Nov 9.	
GM2A	NM_000405	S	http://ghr.nlm.nih.gov/condition/gm2-gangliosidosis-ab-variant	
GNMT	NM_018960	S	http://ghr.nlm.nih.gov/condition/hypermethioninemia	
GNPAT	NM_014236	S	Hum Mol Genet. 1998 May;7(5):847-53.	Hum Mutat. 2012 Jan;33(1):189-97. doi: 10.1002/humu.21623. Epub 2011 Oct 31.
GNPTAB	NM_024312	S	http://ghr.nlm.nih.gov/condition/mucolipidosis-iii-alpha-beta	
GNPTG	NM_032520	S	J Clin Invest. 2000 Mar;105(5):673-81.	
GON4L	NM_001037533	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
GPR56	NM_001145770	S	Brain Dev. 2013 Aug 24. pii: S0387-7604(13)00236-2. doi: 10.1016/j.braindev.2013.07.015. [Epub ahead of print]	Am J Hum Genet. 2002 Apr;70(4):1028-33. Epub 2002 Feb 13.
GRIK2	NM_001166247	NS	Am J Hum Genet. 2007 Oct;81(4):792-8. Epub 2007 Aug 31.	
GRM1	NM_000838	S	Am J Hum Genet. 2012 Sep 7;91(3):553-64. doi: 10.1016/j.ajhg.2012.07.019. Epub 2012 Aug 16.	
GRN	NM_002087	S	http://www.ncbi.nlm.nih.gov/books/NBK1428/	
GSS	NM_000178	S	Nat Genet. 1996 Nov;14(3):361-5	http://ghr.nlm.nih.gov/condition/glutathione-synthetase-deficiency
GTF2H5	NM_207118	S	http://ghr.nlm.nih.gov/condition/trichothiodystrophy	
GUSB	NM_000181	S	Hum Mutat. 2009 Apr;30(4):511-9. doi: 10.1002/humu.20828.	
HAL	NM_002108	S	http://www.omim.org/entry/235800	
HAX1	NM_006118	S	Am J Med Genet A. 2010 Dec;152A(12):3157-63. doi: 10.1002/ajmg.a.33748.	
HEXA	NM_000520	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
HEXB	NM_000521	S	J Inherit Metab Dis. 2009 Dec;32 Suppl 1:S307-11. doi: 10.1007/s10545-009-1261-2. Epub 2009 Nov 4	

HIST1H4B	NM_003544	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
HIST3H3	NM_003493	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
HOXA1	NM_153620	S	Nat Genet. 2005 Oct;37(10):1035-7. Epub 2005 Sep 11.		
HPD	NM_002150	S	Hum Genet. 2000 Jun;106(6):654-62.		
HSD17B4	NM_000414	S	J Pediatr. 2001 Dec;139(6):865-7.	Am J Hum Genet. 2010 Aug 13;87(2):282-8. doi: 10.1016/j.ajhg.2010.07.007. Epub 2010 Jul 30.	
HSPD1	NM_002156	S	Am J Hum Genet. 2008 Jul;83(1):30-42. doi: 10.1016/j.ajhg.2008.05.016. Epub 2008 Jun 19.		
IDUA	NM_000203	S	Bunge et al. (1994), http://www.omim.org/entry/607014		
IFT122	NM_052989	S	Mol Genet Genomic Med. 2014 Mar;2(2):103-6. doi: 10.1002/mgg3.44. Epub 2013 Dec 10.		
IFT140	NM_014714	S	Am J Hum Genet. 2012 May 4;90(5):864-70. doi: 10.1016/j.ajhg.2012.03.006. Epub 2012 Apr 12.		
IGF1	NM_001111283	S	Nestle Nutr Inst Workshop Ser. 2013;71:43-55. doi: 10.1159/000342548. Epub 2013 Jan 22.	N Engl J Med. 1996 Oct 31;335(18):1363-7.	J Med Genet. 2003 Dec;40(12):913-7.
INPP4A	NM_001134224	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
INPP5E	NM_019892	S	Nat Genet. 2009 Sep;41(9):1027-31. doi: 10.1038/ng.427. Epub 2009 Aug 9.		
INSR	NM_000208	S	http://www.omim.org/entry/147670		
IRF6	NM_001206696	S	Clin Dysmorphol. 2007 Jul;16(3):163-6.		
IRX5	NM_005853	S	Nat Genet. 2012 May 13;44(6):709-13. doi: 10.1038/ng.2259.		
ISPD	NM_001101417	S	Am J Hum Genet. 2012 Dec 7;91(6):1135-43. doi: 10.1016/j.ajhg.2012.10.009.	Nat Genet. 2012 May;44(5):575-80. doi: 10.1038/ng.2252.	
KCNJ10	NM_002241	S	Proc Natl Acad Sci U S A. 2009 Apr 7;106(14):5842-7. doi: 10.1073/pnas.0901749106. Epub 2009 Mar 16.		
KCNJ11	NM_000525	S	http://ghr.nlm.nih.gov/condition/familial-hyperinsulinism		

KCTD7	NM_001166290	S	J Med Genet. 2012 Jun;49(6):391-9. doi: 10.1136/jmedgenet-2012-100859	Ann Neurol. 2007 Jun;61(6):579-86.
KDM5A	NM_001042603	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
KDM6B	NM_001080424	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
KIAA1033	NM_015275	S	http://www.ncbi.nlm.nih.gov/pubmed/21498477	
KIAA1279	NM_015634	S	Hum Mol Genet. 2013 Jun 15;22(12):2387-99. doi: 10.1093/hmg/ddt083. Epub 2013 Feb 19.	Am J Hum Genet. 2005 Jul;77(1):120-6. Epub 2005 May 9
KIF14	NM_014875	S	Clin Genet. 2013 Oct 15. doi: 10.1111/cge.12301. [Epub ahead of print]	
KIF1A	NM_004321	S	Genome Res. 2011 May;21(5):658-64. doi: 10.1101/gr.117143.110. Epub 2011 Apr 12	
KIF7	NM_198525	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
L2HGDH	NM_024884	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
LAMA1	NM_005559	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
LAMA2	NM_000426	S	Neurology. 2000 Oct 24;55(8):1128-34.	
LARGE	NM_004737	S	Eur J Hum Genet. 2011 Apr;19(4):452-7. doi: 10.1038/ejhg.2010.212. Epub 2011 Jan 19.	
LARP7	NM_016648	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
LEPR	NM_002303	S	Mol Genet Metab. 2012 Jul;106(3):345-50. doi: 10.1016/j.ymgme.2012.04.026. Epub 2012 May 10.	
LIAS	NM_006859	S	Brain. 2014 Feb;137(Pt 2):366-79. doi: 10.1093/brain/awt328. Epub 2013 Dec 11.	
LIG4	NM_002312	S	PLoS One. 2013;8(1):e54389. doi: 10.1371/journal.pone.0054389. Epub 2013 Jan 25	
LINS (LINS1)	NM_001040616	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
LRP5	NM_002335	S	Am J Med Genet A. 2010 Jan;152A(1):133-40. doi: 10.1002/ajmg.a.33177.	

LZTFL1 (BBS17)	NM_020347	S	Clin Genet. 2014 May;85(5):476-81. doi: 10.1111/cge.12198. Epub 2013 Jun 12.	http://www.ncbi.nlm.nih.gov/pubmed/20301537	
MAN1B1	NM_016219	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
MAN2B1 (LAMAN)	NM_000528	S	Am J Hum Genet. 1999 Jan;64(1):77-88	Hum Mol Genet. 1997 May;6(5):717-26.	
MANBA	NM_005908	S	Mol Genet Metab. 2002 Dec;77(4):282-90		
MAT1A	NM_000429	S	J Clin Invest. 1996 Aug 15;98(4):1021-7.	J Clin Invest. 1995 Oct;96(4):1943-7.	
MCEE	NM_032601	S	http://ghr.nlm.nih.gov/condition/methylmalonic-acidemia		
MCOLN1	NM_020533	S	Am J Med Genet A. 2009 Oct;149A(10):2290-5. doi: 10.1002/ajmg.a.33031.	Brain Dev. 2009 Oct;31(9):702-5. doi: 10.1016/j.braindev.2008.10.001. Epub 2008 Nov 8.	
MCPH1	NM_001172574	S	J Med Genet. 2010 Dec;47(12):823-8. doi: 10.1136/jmg.2009.076398. Epub 2010 Oct 26.	Hum Genet. 2006 Feb;118(6):708-15. Epub 2005 Nov 26.	
MED13L	NM_015335	NS/S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
MED23	NM_015979	NS	Science. 2011 Aug 26;333(6046):1161-3. doi: 10.1126/science.1206638.		
MEGF8	NM_001410	S	Am J Hum Genet. 2012 Nov 2;91(5):897-905. doi: 10.1016/j.ajhg.2012.08.027. Epub 2012 Oct 11.		
METTL23	NM_001080510	S	Hum Mol Genet. 2014 Feb 5. [Epub ahead of print]	Hum Mol Genet. 2014 Apr 1. [Epub ahead of print]	
MFSD8	NM_152778	S	Brain. 2009 Mar;132(Pt 3):810-9. doi: 10.1093/brain/awn366. Epub 2009 Feb 5.	Am J Hum Genet. 2007 Jul;81(1):136-46. Epub 2007 May 14	http://www.ncbi.nlm.nih.gov/books/NBK1428/
MGAT2	NM_002408	S	Am J Med Genet A. 2012 Jan;158A(1):245-6. doi: 10.1002/ajmg.a.34347. Epub 2011 Nov 21.	Am J Hum Genet. 1996 Oct;59(4):810-7.	
MGME1	NM_052865	S	Nat Genet. 2013 Feb;45(2):214-9. doi: 10.1038/ng.2501. Epub 2013 Jan 13.		
MKKS (BBS6)	NM_170784	S	Nat Genet. 2000 Sep;26(1):15-6	Nat Genet. 2000 Sep;26(1):67-70.	
MKS1 (BBS13)	NM_017777	S	Nat Genet. 2008 Apr;40(4):443-8. doi: 10.1038/ng.97. Epub 2008 Mar 9.	Nat Genet. 2006 Feb;38(2):155-7. Epub 2006 Jan 15.	
MLC1	NM_139202	S	Am J Hum Genet. 2011 Apr 8;88(4):422-32. doi: 10.1016/j.ajhg.2011.02.009. Epub 2011 Mar 17.		

MMAA	NM_172250	S	http://ghr.nlm.nih.gov/condition/methylmalonic-acidemia		
MMAB	NM_052845	S	http://ghr.nlm.nih.gov/condition/methylmalonic-acidemia		
MMADHC	NM_015702	S	N Engl J Med. 2008 Apr 3;358(14):1454-64. doi: 10.1056/NEJMoa072200.	http://ghr.nlm.nih.gov/condition/methylmalonic-acidemia	
MPLKIP	NM_138701	S	Am J Hum Genet. 2005 Mar;76(3):510-6. Epub 2005 Jan 11.		
MTHFR	NM_005957	S	Eur J Hum Genet. 1998 May-Jun;6(3):257-65.		
MTRR	NM_000254	S	J Inherit Metab Dis. 2002 Oct;25(6):461-76.		
MUT	NM_000255	S	J Clin Invest. 1992 Feb;89(2):385-91.	Hum Mutat. 2005 Feb;25(2):167-76.	
MVK	NM_000431	S	Pediatrics. 2003 Feb;111(2):258-61.		
MYH3	NM_002470	S	http://ghr.nlm.nih.gov/condition/freeman-sheldon-syndrome		
MYO5A	NM_000259	S	Nat Genet. 1997 Jul;16(3):289-92.		
NAGLU	NM_000263	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
NBN	NM_002485	S	http://www.ncbi.nlm.nih.gov/pubmed/9590180	http://www.omim.org/entry/251260	
NDE1	NM_001143979	S	Am J Hum Genet. 2011 May 13;88(5):523-35. doi: 10.1016/j.ajhg.2011.03.019. Epub 2011 Apr 28.		
NDST1	NM_001543	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
NEU1	NM_000434	S	Hum Mol Genet. 2000 Apr 12;9(7):1075-85	http://www.omim.org/entry/608272?search=neu1&highlight=neu1	
NIN	NM_020921	S	J Clin Endocrinol Metab. 2012 Nov;97(11):E2140-51. doi: 10.1210/jc.2012-2150. Epub 2012 Aug 29.		
NRXN1	NM_001135659	S	Am J Hum Genet. 2009 Nov;85(5):655-66. doi: 10.1016/j.ajhg.2009.10.004. Epub 2009 Nov 5	Am J Med Genet A. 2011 Nov;155A(11):2826-31. doi: 10.1002/ajmg.a.34255. Epub 2011 Sep 30.	
NSUN2	NM_001193455	NS	Am J Hum Genet. 2012 May 4;90(5):847-55. doi: 10.1016/j.ajhg.2012.03.021. Epub 2012 Apr 26.		
NTRK1	NM_001007792	S	Nat Genet. 1996 Aug;13(4):485-8.	Clin Genet. 2009 Mar;75(3):230-6. doi: 10.1111/j.1399-0004.2008.01143.x.	
NUP62	NM_012346	S	http://www.omim.org/entry/271930		
OAT	NM_000274	S	Mol Genet Metab. 2009 Jun;97(2):109-13. doi: 10.1016/j.ymgme.2008.12.010. Epub 2009 Mar 31	Valayannopoulos, et al., 2009	http://www.omim.org/entry/258870

PAH	NM_000277	S	PLoS One. 2014 Apr 4;9(4):e94100. doi: 10.1371/journal.pone.0094100. eCollection 2014.	Eisensmith and woo., 1992, http://www.omim.org/entry/261600
PARP1	NM_001618	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
PC	NM_022172	S	Hum Mutat. 2009 May;30(5):734-40. doi: 10.1002/humu.20908.	http://www.omim.org/entry/266150
PCBD1	NM_000281	S	PLoS One. 2014 Apr 4;9(4):e94100. doi: 10.1371/journal.pone.0094100. eCollection 2014.	
PCCA	NM_001178004	S	World J Pediatr. 2014 Feb;10(1):64-8. doi: 10.1007/s12519-014-0454-4. Epub 2014 Jan 25.	http://ghr.nlm.nih.gov/condition/propionic-acidemia
PCCB	NM_000532	S	World J Pediatr. 2014 Feb;10(1):64-8. doi: 10.1007/s12519-014-0454-4. Epub 2014 Jan 25.	http://ghr.nlm.nih.gov/condition/propionic-acidemia
PDHX	NM_001166158	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
PDSS1	NM_014317	S	J Clin Invest. 2007 Mar;117(3):765-72.	
PECR	NM_018441	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
PEPD	NM_000285	S	http://www.ncbi.nlm.nih.gov/pmc/articles/PMC295128/pdf/jcinvest00058-0045.pdf	
PEX1	NM_000466	S	Am J Med Genet A. 2004 May 1;126A(4):333-8.	
PEX11B	NM_003846	S	J Med Genet. 2012 May;49(5):307-13. doi: 10.1136/jmedgenet-2012-100778.	
PEX6	NM_000287	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
PEX7	NM_000288	S	Am J Hum Genet. 2002 Mar;70(3):612-24. Epub 2002 Jan 7.	Nat Genet. 1997 Apr;15(4):381-4.
PGAP2	NM_014489	S	Am J Hum Genet. 2013 Apr 4;92(4):575-83. doi: 10.1016/j.ajhg.2013.03.008.	
PGAP3	NM_033419	S	Am J Hum Genet. 2014 Feb 6;94(2):278-87. doi: 10.1016/j.ajhg.2013.12.012. Epub 2014 Jan 16.	
PGM3	NM_001199918	S	J Allergy Clin Immunol. 2014 Feb 28. pii: S0091-6749(14)00262-0. doi: 10.1016/j.jaci.2014.02.013. [Epub ahead of print]	
PIGL	NM_004278	S	Am J Hum Genet. 2012 Apr 6;90(4):685-8. doi: 10.1016/j.ajhg.2012.02.010. Epub 2012 Mar 22.	
PIGO	NM_032634	S	Am J Hum Genet. 2012 Jul 13;91(1):146-51. doi: 10.1016/j.ajhg.2012.05.004. Epub 2012 Jun 7.	

PIGV	NM_001202554	S	Nat Genet. 2010 Oct;42(10):827-9. doi: 10.1038/ng.653. Epub 2010 Aug 29	
PIGW	NM_178517	S	J Med Genet. 2014 Mar;51(3):203-7. doi: 10.1136/jmedgenet-2013-102156. Epub 2013 Dec 23.	
PITX3	NM_005029	S	Invest Ophthalmol Vis Sci. 2006 Apr;47(4):1274-80	
PLA2G6	NM_003560	S	Clin Genet. 2010 Nov;78(5):432-40. doi: 10.1111/j.1399-0004.2010.01417.x.	Neurology. 2008 Oct 28;71(18):1402-9. doi: 10.1212/01.wnl.0000327094.67726.28. Epub 2008 Sep 17.
PMM2	NM_000303	S	http://www.ncbi.nlm.nih.gov/pubmed/17307006	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.
PNKP	NM_007254	S	Nat Genet. 2010 Mar;42(3):245-9. doi: 10.1038/ng.526. Epub 2010 Jan 31.	Neurogenetics. 2013 Feb;14(1):43-51. doi: 10.1007/s10048-012-0351-8. Epub 2012 Dec 9.
PNP	NM_000270	S	http://ghr.nlm.nih.gov/condition/purine-nucleoside-phosphorylase-deficiency	
POLR3B	NM_018082	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
POMGNT1	NM_017739	S	Mol Genet Genomics. 2013 Aug;288(7-8):297-308. doi: 10.1007/s00438-013-0749-5. Epub 2013 May 21	Hum Mol Genet. 2003 Mar 1;12(5):527-34.
POMGNT2 (GTDC2)	NM_032806	S	Am J Hum Genet. 2012 Sep 7;91(3):541-7. doi: 10.1016/j.ajhg.2012.07.009.	
POMK	NM_032237	S	J Med Genet. 2014 Apr;51(4):275-82. doi: 10.1136/jmedgenet-2013-102236. Epub 2014 Feb 20.	
POMT1	NM_007171	S	Neurology. 2009 May 26;72(21):1802-9. doi: 10.1212/01.wnl.0000346518.68110.60. Epub 2009 Mar 18.	Brain. 2007 Oct;130(Pt 10):2725-35. Epub 2007 Sep 18.
POMT2	NM_013382	S	J Med Genet. 2005 Dec;42(12):907-12. Epub 2005 May 13.	
POR	NM_000941	S	http://ghr.nlm.nih.gov/condition/cytochrome-p450-oxidoreductase-deficiency	
POU1F1	NM_001122757	S	Am J Med Genet A. 2011 Sep;155A(9):2242-6. doi: 10.1002/ajmg.a.34136. Epub 2011 Aug 3.	
PPT1	NM_000306	S	http://www.ncbi.nlm.nih.gov/pubmed/9571187	
PRKCG	NM_002739	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
PRKRA	NM_003690	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
PRMT10	NM_138364.2	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	

PRODH	NM_001195226	S	http://www.ncbi.nlm.nih.gov/pubmed/18197084		
PROP1	NM_006261	S	http://www.ncbi.nlm.nih.gov/pubmed/18174732		
PRRT2	NM_145239	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
PRSS12	NM_003619	NS	Science. 2002 Nov 29;298(5599):1779-81.		
PTH	NM_000315	S	http://www.ncbi.nlm.nih.gov/pubmed/?term=garfield+karaplis	http://www.omim.org/entry/146200	
PTS	NM_000317	S	http://ghr.nlm.nih.gov/condition/tetrahydrobiopterin-deficiency	PLoS One. 2014 Apr 4;9(4):e94100. doi: 10.1371/journal.pone.0094100. eCollection 2014.	Zhonghua Yi Xue Yi Chuan Xue Za Zhi. 2007 Apr;24(2):210-2
PVRL1 (HVEC)	NM_203285	S	http://www.omim.org/entry/225060		
PYCR1	NM_006907	S	Nat Genet. 2009 Sep;41(9):1016-21. doi: 10.1038/ng.413. Epub 2009 Aug 2.	Am J Med Genet. 2001 Jul 1;101(3):213-20.	
QDPR	NM_000320	S	PLoS One. 2014 Apr 4;9(4):e94100. doi: 10.1371/journal.pone.0094100. eCollection 2014.	http://www.omim.org/entry/261630	http://www.ncbi.nlm.nih.gov/pubmed/10029353
RAB18	NM_021252	S	Biochem Soc Trans. 2012 Dec 1;40(6):1394-7. doi: 10.1042/BST20120169		
RAB23	NM_016277	S	Am J Hum Genet. 2007 Jun;80(6):1162-70. Epub 2007 Apr 18.		
RAB3GAP1	NM_001172435	S	Eur J Hum Genet. 2010 Oct;18(10):1100-6. doi: 10.1038/ejhg.2010.79. Epub 2010 May 26.	Hum Genet. 2011 Jan;129(1):45-50. doi: 10.1007/s00439-010-0896-2. Epub 2010 Oct 22.	
RAB3GAP2	NM_012414	S	Hum Genet. 2011 Jan;129(1):45-50. doi: 10.1007/s00439-010-0896-2. Epub 2010 Oct 22.		
RABL6 (C9orf86)	NM_001173989	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
RALGDS	NM_006266	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
RARS2	NM_020320	S	http://ghr.nlm.nih.gov/condition/pontocerebellar-hypoplasia		
RBBP8	NM_002894	S	http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3188555/		
RBM28	NM_001166135	S	Am J Hum Genet. 2008 May;82(5):1114-21. doi: 10.1016/j.ajhg.2008.03.014. Epub 2008 Apr 24		
RELN	NM_005045	S	Nat Genet. 2000 Sep;26(1):93-6.		

RGS7	NM_002924	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
RMRP	NR_003051	S	Am J Hum Genet. 2005 Nov;77(5):795-806. Epub 2005 Sep 29		
RNASEH2A	NM_006397	S	Nat Genet. 2006 Aug;38(8):910-6. Epub 2006 Jul 16	Am J Hum Genet. 2007 Oct;81(4):713-25. Epub 2007 Sep 4.	
RNASEH2B	NM_024570	S	Nat Genet. 2006 Aug;38(8):910-6. Epub 2006 Jul 16	Am J Hum Genet. 2007 Oct;81(4):713-25. Epub 2007 Sep 4.	
RNASEH2C	NM_032193	S	Nat Genet. 2006 Aug;38(8):910-6. Epub 2006 Jul 16	Am J Hum Genet. 2007 Oct;81(4):713-25. Epub 2007 Sep 4.	
ROGDI	NM_024589	S	Am J Hum Genet. 2012 Apr 6;90(4):708-14. doi: 10.1016/j.ajhg.2012.03.005.	Hum Mutat. 2013 Feb;34(2):296-300. doi: 10.1002/humu.22241. Epub 2012 Nov 27	Am J Hum Genet. 2012 Apr 6;90(4):701-7. doi: 10.1016/j.ajhg.2012.02.012. Epub 2012 Mar 15.
RPGRIP1L	NM_001127897	S	Clin Genet. 2008 Aug;74(2):164-70. doi: 10.1111/j.1399-0004.2008.01047.x. Epub 2008 Jun 28.	J Med Genet. 2010 Jan;47(1):8-21. doi: 10.1136/jmg.2009.067249. Epub 2009 Jul 1.	
RTTN	NM_173630	S	Am J Hum Genet. 2012 Sep 7;91(3):533-40. doi: 10.1016/j.ajhg.2012.07.008. Epub 2012 Aug 30.		
SACS	NM_014363	S	Neurology. 2006 Apr 11;66(7):1103-4.	Neurology. 2010 Sep 28;75(13):1181-8. doi: 10.1212/WNL.0b013e3181f4d86c.	
SALL1	NM_002968	S	J Pediatr. 2013 Mar;162(3):612-7. doi: 10.1016/j.jpeds.2012.08.042. Epub 2012 Oct 12		
SAMHD1	NM_015474	S	Eur J Hum Genet. 2011 Mar;19(3):287-92. doi: 10.1038/ejhg.2010.213. Epub 2010 Nov 24.		
SC5D	NM_006918	S	Am J Hum Genet. 2002 Oct;71(4):952-8. Epub 2002 Aug 20		
SCAPER	NM_020843	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
SCO1	NM_004589	S	http://ghr.nlm.nih.gov/condition/cytochrome-c-oxidase-deficiency	Am J Hum Genet. 2000 Nov;67(5):1104-9. Epub 2000 Sep 28.	
SCO2	NM_001169111	S	http://ghr.nlm.nih.gov/condition/cytochrome-c-oxidase-deficiency	Arch Neurol. 2002 May;59(5):862-5.	
SDCCAG8 (BBS16) (NPHP10)	NM_006642	S	Nat Genet. 2010 Oct;42(10):840-50. doi: 10.1038/ng.662. Epub 2010 Sep 12.		
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SLC12A3 (TSC)	NM_000339	S	Am J Nephrol. 2008;28(6):900-7. doi: 10.1159/000141932. Epub 2008 Jun 26.		

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SLC17A5	NM_012434	S	Nat Genet. 1999 Dec;23(4):462-5.		
SLC19A3	NM_025243	S	Am J Hum Genet. 2005 Jul;77(1):16-26. Epub 2005 May 3	Brain. 1998 Jul;121 (Pt 7):1267-79.	Arch Neurol. 2010 Jan;67(1):126-30. doi: 10.1001/archneurol.2009.293.
SLC25A15	NM_014252	S	Hum Mutat. 2009 May;30(5):741-8. doi: 10.1002/humu.20930.	Nat Genet. 1999 Jun;22(2):151-8	
SLC2A1	NM_006516	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
SLC31A1	NM_001859	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
SLC35C1	NM_001145265	S	Nat Genet. 2001 May;28(1):69-72.	Nat Genet. 2001 May;28(1):73-6.	
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SMPD1	NM_000543	S	Hum Mutat. 2009 Jul;30(7):1117-22. doi: 10.1002/humu.21018.		
SOBP	NM_018013	S	Am J Hum Genet. 2010 Nov 12;87(5):694-700. doi: 10.1016/j.ajhg.2010.10.005. Epub 2010 Oct 28.		
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SPR	NM_003124	S	Am J Hum Genet. 2001 Aug;69(2):269-77. Epub 2001 Jul 6.		
SRD5A3	NM_024592	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	Eur J Hum Genet. 2011 Jan;19(1):115-7. doi: 10.1038/ejhg.2010.132. Epub 2010 Aug 11	Cell. 2010 Jul 23;142(2):203-17. doi: 10.1016/j.cell.2010.06.001. Epub 2010 Jul 15.
ST3GAL3	NM_174963	NS	Am J Hum Genet. 2011 Sep 9;89(3):407-14. doi: 10.1016/j.ajhg.2011.08.008.		
STIL	NM_001048166	S	Am J Hum Genet. 2009 Feb;84(2):286-90. doi: 10.1016/j.ajhg.2009.01.017.		
SUCLA2	NM_003850	S	J Hum Genet. 2013 Aug;58(8):526-30. doi: 10.1038/jhg.2013.45. Epub 2013 Jun 13.	Am J Hum Genet. 2005 Jun;76(6):1081-6. Epub 2005 Apr 22.	
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SURF1	NM_003172	S	Am J Med Genet A. 2004 Jul 15;128A(2):195-8.	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
TAF2	NM_003184	S	Pediatr Neurol. 2013 Dec;49(6):411-416.e1. doi: 10.1016/j.pediatrneurol.2013.07.017. Epub 2013 Sep 29	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.	
TAT	NM_000353	S	Proc Natl Acad Sci U S A. 1992 Oct 1;89(19):9297-301.	Hum Genet. 1987 Dec;77(4):352-8	
TBC1D24	NM_020705	S	Am J Hum Genet. 2010 Sep 10;87(3):371-5. doi: 10.1016/j.ajhg.2010.08.001	J Med Genet. 2013 Mar;50(3):199-202. doi: 10.1136/jmedgenet-2012-101313. Epub 2013 Jan 23.	Lancet Neurol. 2014 Jan;13(1):44-58. doi: 10.1016/S1474-4422(13)70265-5. Epub 2013 Nov 29
TBC1D7	NM_016495	S	J Med Genet. 2013 Nov;50(11):740-4. doi: 10.1136/jmedgenet-2013-101680. Epub 2013 May 17.	Hum Mutat. 2014 Apr;35(4):447-51. doi: 10.1002/humu.22529.	
TBCE	NM_003193	S	Nat Genet. 2002 Nov;32(3):448-52. Epub 2002 Oct 21.		
TBR1	NM_006593	S	Am J Med Genet A. 2014 Jan 23. doi: 10.1002/ajmg.a.36363. [Epub ahead of print]	Nat Genet. 2007 Apr;39(4):454-6. Epub 2007 Mar 11	
TCIRG1	NM_006019	S	J Med Genet. 2003 Feb;40(2):115-21		
TECPR2	NM_001172631	S	Am J Hum Genet. 2012 Dec 7;91(6):1065-72. doi: 10.1016/j.ajhg.2012.09.015. Epub 2012 Nov 21		
TECR	NM_138501	NS	Hum Mol Genet. 2011 Apr 1;20(7):1285-9. doi: 10.1093/hmg/ddq569. Epub 2011 Jan 6.		
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TH	NM_000360	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
THOC6	NM_001142350	S	Orphanet J Rare Dis. 2013 Apr 26;8(1):62. [Epub ahead of print]		
TMCO1	NM_019026	S	Proc Natl Acad Sci U S A. 2010 Jan 5;107(1):258-63. doi: 10.1073/pnas.0908457107. Epub 2009 Dec 14.	Eur J Hum Genet. 2014 Jan 15. doi: 10.1038/ejhg.2013.291. [Epub ahead of print]	
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TMEM237	NM_152388	S	Am J Hum Genet. 2011 Dec 9;89(6):713-30. doi: 10.1016/j.ajhg.2011.11.005.		
TMEM67	NM_001142301	S	J Med Genet. 2009 Oct;46(10):663-70. doi: 10.1136/jmg.2009.066613. Epub 2009 Jun	J Clin Invest. 2011 Jul;121(7):2662-7. doi: 10.1172/JCI43639.	Nat Genet. 2008 Apr;40(4):443-8. doi: 10.1038/ng.97. Epub 2008 Mar 9.

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TPP1 (CLN2)	NM_000391	S	Clin Genet. 1998 Sep;54(3):234-8.	Am J Hum Genet. 1999 Jun;64(6):1511-23.	http://www.ncbi.nlm.nih.gov/books/NBK1428/
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TREX1	NM_016381	S	Am J Hum Genet. 2000 Jul;67(1):213-21. Epub 2000 May 25.		
TRIM32	NM_012210	S	Proc Natl Acad Sci U S A. 2006 Apr 18;103(16):6287-92. Epub 2006 Apr 10.	http://www.ncbi.nlm.nih.gov/pubmed/20301537	
TRMT1	NM_001142554	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
TSEN2	NM_025265	S	Nat Genet. 2008 Sep;40(9):1113-8. doi: 10.1038/ng.204.		
TSEN34	NM_001077446	S	Nat Genet. 2008 Sep;40(9):1113-8. doi: 10.1038/ng.204.		
TSEN54	NM_207346	S	Nat Genet. 2008 Sep;40(9):1113-8. doi: 10.1038/ng.204.		
TSHB	NM_000549	S	EMBO J. 1989 Aug;8(8):2291-6.		
TTC8 (BBS8)	NM_198309	S	J Med Genet. 2010 Apr;47(4):262-7. doi: 10.1136/jmg.2009.071365. Epub 2009 Sep 24.	Nature. 2003 Oct 9;425(6958):628-33. Epub 2003 Sep 21	
TTI2 (C8orf41)	NM_025115	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
TUBGCP6	NM_020461	S	PLoS One. 2012;7(1):e28936. doi: 10.1371/journal.pone.0028936. Epub 2012 Jan 17.		
TUSC3	NM_178234	NS	Am J Hum Genet. 2008 May;82(5):1158-64. doi: 10.1016/j.ajhg.2008.03.018. Epub 2008 May 1.	Am J Hum Genet. 2008 May;82(5):1150-7. doi: 10.1016/j.ajhg.2008.03.021. Epub 2008 May 1.	Am J Med Genet A. 2011 Aug;155A(8):1976-80. doi: 10.1002/ajmg.a.34077. Epub 2011 Jul 7.
UBE3B	NM_130466	S	Am J Hum Genet. 2012 Dec 7;91(6):998-1010. doi: 10.1016/j.ajhg.2012.10.011. Epub 2012 Nov 29.	200	
UBR1	NM_174916	S	Nat Genet. 2005 Dec;37(12):1345-50. Epub 2005 Nov 20.		
UBR7	NM_175748	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
UGT1A1	NM_000463	S	http://ghr.nlm.nih.gov/condition/crigler-najjar-syndrome		
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UROC1	NM_001165974	S	J Med Genet. 2009 Jun;46(6):407-11. doi: 10.1136/jmg.2008.060632. Epub 2009 Mar 19.		

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VPS13B (COH1)	NM_015243	S	Am J Med Genet A. 2008 Sep 1;146A(17):2221-6. doi: 10.1002/ajmg.a.32239.	Am J Hum Genet. 2003 Jun;72(6):1359-69. Epub 2003 May 2.	
VPS37A	NM_001145152	S	J Med Genet. 2012 Jul;49(7):462-72. doi: 10.1136/jmedgenet-2012-100742. Epub 2012 Jun 20.		
VRK1	NM_003384	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
WDPCP (C2orf86) (BBS15)	NM_015910	S	Science. 2010 Sep 10;329(5997):1337-40. doi: 10.1126/science.1191184. Epub 2010 Jul 29.		
WDR45B (WDR45L)	NM_019613	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
WDR62	NM_001083961	S	Bilguvar et al. (2010)	Nat Genet. 2010 Nov;42(11):1010-4. doi: 10.1038/ng.682. Epub 2010 Oct 3.	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.
WDR81	NM_001163673	S	Genome Res. 2011 Dec;21(12):1995-2003. doi: 10.1101/gr.126110.111. Epub 2011 Sep 1.		
WFS1	NM_001145853	S	Lancet. 1990 Sep 15;336(8716):667-9.	Am J Psychiatry. 1991 Jun;148(6):775-9.	
WWOX	NM_016373.2	S	Brain. 2014 Feb;137(Pt 2):411-9. doi: 10.1093/brain/awt338. Epub 2013 Dec 24.		
ZBTB16	NM_001018011	S	J Med Genet. 2008 Nov;45(11):731-7. doi: 10.1136/jmg.2008.059451. Epub 2008 Jul 8.		
ZBTB24	NM_001164313	S	Am J Med Genet A. 2012 Aug;158A(8):2043-6. doi: 10.1002/ajmg.a.35486. Epub 2012 Jul 11		
ZBTB40	NM_001083621	S	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
ZC3H14	NM_001160103	S	Proc Natl Acad Sci U S A. 2011 Jul 26;108(30):12390-5. doi: 10.1073/pnas.1107103108. Epub 2011 Jul 6.		
ZCCHC8	NM_017612	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
ZFYVE26 (SPG15)	NM_015346	S	Neurology. 2009 Oct 6;73(14):1111-9. doi: 10.1212/WNL.0b013e3181bacf59.	Am J Hum Genet. 2008 Apr;82(4):992-1002. doi: 10.1016/j.ajhg.2008.03.004.	
ZNF335	NM_022095	S	Cell. 2012 Nov 21;151(5):1097-112. doi: 10.1016/j.cell.2012.10.043		
ZNF526	NM_133444	NS	Nature. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423.		
ZNF592	NM_014630	S	Eur J Hum Genet. 2010 Oct;18(10):1107-13. doi: 10.1038/ejhg.2010.82. Epub 2010 Jun 9		