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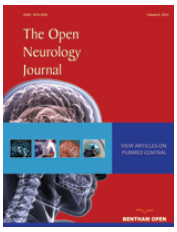

Functional and Genomic Features of Human Genes Mutated in Neuropsychiatric Disorders

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Table S1. List of genes known to harbor mutations for NPD.

GENE SYMBOL	GENE NAME	ENSEMBL ID	DISORDERS	PMID
ABCB7	ATP-BINDING CASSETTE, SUB-FAMILY B (MDR/TAP), MEMBER 7	ENSG00000131269	X-linked sideroblastic anemia and ataxia	10196363
ABCD1	ATP-BINDING CASSETTE, SUB-FAMILY D (ALD), MEMBER 1	ENSG00000101986	X-linked adrenoleukodystrophy	8441467
ABHD12	ABHYDROLASE DOMAIN CONTAINING 12	ENSG00000100997	Polyneuropathy, hearing loss and ataxia	20797687
ACSL4	ACYL-COA SYNTHETASE LONG-CHAIN FAMILY MEMBER 4	ENSG00000068366	nonspecific X-linked mental retardation	11889465
ADNP	activity-dependent neuroprotector homeobox	ENSG00000101126	autism spectrum disorder and intellectual disability	24531329
AGTR2	ANGIOTENSIN II RECEPTOR, TYPE 2	ENSG00000180772	X-linked mental retardation	12089445
AHI1	ABELSON HELPER INTEGRATION SITE 1	ENSG00000135541	Joubert syndrome	15467982
AIMP1	AMINOACYL TRNA SYNTHETASE COMPLEX-INTERACTING MULTIFUNCTIONAL PROTEIN 1	ENSG00000164022	Pelizaeus-Merzbacher-like Disease	21092922
ALDH7A1	ALDEHYDE DEHYDROGENASE 7 FAMILY, MEMBER A1	ENSG00000164904	pyridoxine-dependent seizures	16491085
ALS2	AMYOTROPHIC LATERAL SCLEROSIS 2 (JUVENILE)	ENSG00000003393	Infantile-onset ascending hereditary spastic paralysis	12145748
ALS2	AMYOTROPHIC LATERAL SCLEROSIS 2 (JUVENILE)	ENSG00000003393	recessive amyotrophic lateral sclerosis	11586297
ANG	ANGIOGENIN, RIBONUCLEASE, RNASE A FAMILY, 5	ENSG00000214274	amyotrophic lateral sclerosis	16501576
ANO10	ANOCTAMIN 10	ENSG00000160746	Autosomal-Recessive Cerebellar Ataxia.	21092923
ANO5	ANOCTAMIN 5	ENSG00000171714	proximal limb-girdle muscular dystrophy and distal myopathy	20096397
AP1S2	ADAPTOR-RELATED PROTEIN COMPLEX 1, SIGMA 2 SUBUNIT	ENSG00000182287	X-linked mental retardation	17186471
AP4B1	adaptor-related protein complex 4, beta 1 subunit	ENSG00000134262	severe autosomal-recessive intellectual disability, progressive spastic paraplegia	21620353
AP4E1	adaptor-related protein complex 4, epsilon 1 subunit	ENSG00000081014	severe autosomal-recessive intellectual disability, progressive spastic paraplegia	21620353
AP4M1	ADAPTOR-RELATED PROTEIN COMPLEX 4, MU 1 SUBUNIT	ENSG00000221838	congenital spastic tetraplegia	19559397

(Table S1) contd.....

GENE SYMBOL	GENE NAME	ENSEMBL ID	DISORDERS	PMID
AP4S1	adaptor-related protein complex 4, sigma 1 subunit	ENSG00000100478	severe autosomal-recessive intellectual disability, progressive spastic paraplegia	21620353
APP	AMYLOID BETA (A4) PRECURSOR PROTEIN	ENSG00000142192	familial Alzheimer's disease	1671712
APTX	APRATAXIN	ENSG00000137074	ataxia-ocular apraxia 1	11586300
ARID1A	AT rich interactive domain 1A (SWI-like)	ENSG00000117713	Coffin-Siris syndrome.	22426308
ARID1B	AT rich interactive domain 1B (SWI1-like)	ENSG00000049618	intellectual disability	22405089
ARX	ARISTALESS RELATED HOMEOBOX	ENSG00000004848	X-linked mental retardation	11889467
ASNS	asparagine synthetase (glutamine-hydrolyzing)	ENSG00000070669	congenital microcephaly, intellectual disability, progressive cerebral atrophy, and intractable seizure	24139043
ASPM	HYPOTHETICAL PROTEIN FLJ10517	ENSG00000066279	autosomal recessive primary microcephaly	12355089
ATCAY	ATAXIA, CEREBELLAR, CAYMAN TYPE (CAYTAXIN)	ENSG00000167654	Cayman ataxia	14556008
ATL1	atlastin GTPase 1	ENSG00000198513	autosomal dominant hereditary spastic paraplegia	11685207
ATM	ATAXIA TELANGIECTASIA MUTATED	ENSG00000149311	ataxia telangiectasia	7792600
ATP13A2	ATPASE TYPE 13A2	ENSG00000159363	autosomal recessive early-onset parkinsonism with pyramidal degeneration and dementia	16964263
ATP1A3	ATPASE, NA+/K+ TRANSPORTING, ALPHA 3 POLYPEPTIDE	ENSG00000105409	rapid-onset dystonia parkinsonism	15260953
ATP1A3	ATPase, Na+/K+ transporting, alpha 3 polypeptide	ENSG00000105409	Alternating hemiplegia of childhood	22842232
ATP7A	ATPASE, CU++ TRANSPORTING, ALPHA POLYPEPTIDE	ENSG00000165240	X-linked distal hereditary motor neuropathy.	20170900
ATR	ATAXIA TELANGIECTASIA AND RAD3 RELATED	ENSG00000175054	Seckel syndrome	12640452
ATRX	ALPHA THALASSEMIA/MENTAL RETARDATION SYNDROME X-LINKED	ENSG00000085224	X-linked mental retardation with alpha-thalassemia	7697714
B4GALNT1	beta-1,4-N-acetyl-galactosaminyl transferase 1	ENSG00000135454	recessive hereditary spastic paraplegia	23746551
BCKDK	branched chain ketoacid dehydrogenase kinase	ENSG00000103507	autism, epilepsy, and intellectual disability	22956686
BRWD3	BROMODOMAIN AND WD REPEAT DOMAIN CONTAINING 3	ENSG00000165288	X-linked mental retardation associated with macrocephaly	17668385
C10orf2	CHROMOSOME 10 OPEN READING FRAME 2	ENSG00000107815	autosomal dominant progressive external ophthalmoplegia	11431692
c12orf57	chromosome 12 open reading frame 57	ENSG00000111678	Recessive corpus callosum hypoplasia	23453666
C20orf54	CHROMOSOME 20 OPEN READING FRAME 54	ENSG00000101276	progressive ponto-bulbar palsy and bilateral sensorineural deafness	20206331
CA8	CARBONIC ANHYDRASE VIII	ENSG00000178538	ataxia and mild mental retardation	19461874
CABP2	calcium binding protein 2	ENSG00000167791	autosomal-recessive hearing impairment.	22981119
CACNA1A	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA 1A SUBUNIT	ENSG00000141837	Familial hemiplegic migraine/episodic ataxia type-2	8898206
CACNA1C	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, L TYPE, ALPHA 1C SUBUNIT	ENSG00000151067	Timothy syndrome	15454078
CACNA1H	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, ALPHA 1H SUBUNIT	ENSG00000196557	childhood absence epilepsy	12891677
CACNA1H	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, T TYPE, ALPHA 1H SUBUNIT	ENSG00000196557	autism spectrum disorders	16754686
CACNB4	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, BETA 4 SUBUNIT	ENSG00000182389	idiopathic generalized epilepsy and episodic ataxia	10762541
CADM1	CELL ADHESION MOLECULE 1	ENSG00000182985	Autism Spectrum Disorder	18957284
CASK	CALCIUM/CALMODULIN-DEPENDENT SERINE PROTEIN KINASE	ENSG00000147044	microcephaly and severe mental retardation	19165920
CC2D2A	KIAA1345 PROTEIN	ENSG00000048342	autosomal-recessive mental retardation with retinitis pigmentosa	18387594

(Table S2) contd.....

GENE SYMBOL	GENE NAME	ENSEMBL ID	DISORDERS	PMID
CCND2	cyclin D2	ENSG00000118971	megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome	24705253
CCT5	CHAPERONIN CONTAINING TCP1, SUBUNIT 5 (EPSILON)	ENSG00000150753	autosomal recessive mutilating sensory neuropathy with spastic paraplegia	16399879
CDH15	CADHERIN 15, M-CADHERIN (MYOTUBULE)	ENSG00000129910	intellectual disability	19012874
CDKL5	CYCLIN-DEPENDENT KINASE-LIKE 5	ENSG00000008086	X-linked West syndrome	12736870
CEP135	centrosomal protein 135kDa	ENSG00000174799	Autosomal-recessive primary microcephaly	22521416
CEP152	CENTROSOMAL PROTEIN 152KDA	ENSG00000103995	Primary microcephaly	20598275
CHCHD10	coiled-coil-helix-coiled-coil-helix domain containing 10	ENSG00000250479	frontotemporal dementia and amyotrophic lateral sclerosis	24934289
CHD2	chromodomain helicase DNA binding protein 2	ENSG00000173575	fever-sensitive myoclonic epileptic encephalopathy	24207121
CHD2	chromodomain helicase DNA binding protein 2	ENSG00000173575	epileptic encephalopathy	23708187
CHD7	chromodomain helicase DNA binding protein 7	ENSG00000171316	CHARGE syndrome	15300250
CHD8	chromodomain helicase DNA binding protein 8	ENSG00000100888	Autism spectrum disorder	24998929
CHMP1A	charged multivesicular body protein 1A	ENSG00000131165	pontocerebellar hypoplasia and microcephaly	23023333
CHMP2B	CHROMATIN MODIFYING PROTEIN 2B	ENSG00000083937	autosomal dominant frontotemporal dementia	16041373
CHRNA2	CHOLINERGIC RECEPTOR, NICOTINIC, ALPHA 2 (NEURONAL)	ENSG00000120903	familial epilepsy with nocturnal wandering and ictal fear	16826524
CHRNA4	CHOLINERGIC RECEPTOR, NICOTINIC, ALPHA 4	ENSG00000101204	autosomal dominant nocturnal frontal lobe epilepsy	7550350
CHRNB2	CHOLINERGIC RECEPTOR, NICOTINIC, BETA 2 (NEURONAL)	ENSG00000160716	autosomal dominant nocturnal frontal lobe epilepsy	11062464
CLCN2	CHLORIDE CHANNEL 2	ENSG00000114859	idiopathic generalized epilepsies	12612585
CLN8	CEROID-LIPOFUSCINOSIS, NEURONAL 8	ENSG00000182372	Progressive epilepsy with mental retardation	10508524
CNTNAP2	CONTACTIN ASSOCIATED PROTEIN-LIKE 2	ENSG00000174469	Recessive symptomatic focal epilepsy	16571880
CNTNAP2	CONTACTIN ASSOCIATED PROTEIN-LIKE 2	ENSG00000174469	Autosomal-Recessive Pitt-Hopkins-like Mental Retardation	19896112
CNTNAP2	CONTACTIN ASSOCIATED PROTEIN-LIKE 2	ENSG00000174469	cortical dysplasia–focal epilepsy	16571880
COL4A2	collagen, type IV, alpha 2	ENSG00000134871	intracerebral hemorrhage	22209247
CRBN	CEREBLON	ENSG00000113851	mild nonsyndromic mental retardation	15557513
CREBBP	CREB BINDING PROTEIN	ENSG00000005339	Rubinstein-Taybi syndrome	7630403
CSNK1D	casein kinase 1, delta	ENSG00000141551	familial advanced sleep phase syndrome	15800623
CSNK1D	casein kinase 1, delta	ENSG00000141551	Migraine with aura	23636092
CUL4B	CULLIN 4B	ENSG00000158290	X-linked mental retardation	17273978
DCC	DELETED IN COLORECTAL CARCINOMA	ENSG00000187323	isolated congenital mirror movements	20431009
DCHS1	dachsous cadherin-related 1	ENSG00000166341	recessive syndrome periventricular neuronal heterotopia	24056717
DCTN1	DYNACTIN 1 (P150, GLUED HOMOLOG, DROSOPHILA)	ENSG00000204843	lower motor neuron disease	12627231
DDHD2	DDHD domain containing 2	ENSG00000085788	early-onset spastic paraplegia and intellectual disability	23176823
DEPDC5	DEP domain containing 5	ENSG00000100150	Autosomal dominant familial focal epilepsy with variable foci	23542697
DFNB59	DEAFNESS, AUTOSOMAL RECESSIVE 59	ENSG00000204311	autosomal recessive auditory neuropathy	16804542
DIABLO	diablo, IAP-binding mitochondrial protein	ENSG00000184047	dominant progressive nonsyndromic hearing loss	21722859
DISC1	KIAA0457 PROTEIN	ENSG00000162946	schizophrenia	10814723
DLG3	DISCS, LARGE HOMOLOG 3 (NEUROENDOCRINE-DLG, DROSOPHILA)	ENSG00000082458	nonsyndromic X-linked mental retardation	15185169
DNAJC3	DnaJ (Hsp40) homolog, subfamily C, member 3	ENSG00000102580	Multisystemic Neurodegeneration	25466870

(Table S2) contd.....

GENE SYMBOL	GENE NAME	ENSEMBL ID	DISORDERS	PMID
DNAJC5	DnaJ (Hsp40) homolog, subfamily C, member 5	ENSG00000101152	Autosomal-Dominant Adult-Onset Neuronal Ceroid Lipofuscinosis	21820099
DRD2	DOPAMINE RECEPTOR D2	ENSG00000149295	myoclonus dystonia	10220438
DST	dystonin	ENSG00000151914	Hereditary sensory autonomic neuropathy	22522446
DYM	DYMECLIN	ENSG00000141627	Dyggve-Melchior-Clausen disease	12491225
DYNC1H1	dynein, cytoplasmic 1, heavy chain 1	ENSG00000197102	Dominant Axonal Charcot-Marie-Tooth Disease	21820100
DYNC1H1	dynein, cytoplasmic 1, heavy chain 1	ENSG00000197102	malformations of cortical development and microcephaly	23603762
EFHC1	HYPOTHETICAL PROTEIN FLJ10466	ENSG00000096093	Juvenile myoclonic epilepsy	15258581
EGR2	EARLY GROWTH RESPONSE 2 (KROX-20 HOMOLOG, DROSOPHILA)	ENSG00000122877	congenital hypomyelinating neuropathy/family with Charcot-Marie-Tooth type 1	9537424
EHMT1	euchromatic histone-lysine N-methyltransferase 1	ENSG00000181090	Kleefstra syndrome	16826528
EIF2B2	EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 2 BETA, 39KDA	ENSG00000119718	Leukoencephalopathy with vanishing white matter	11704758
EIF2B5	EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 5 EPSILON, 82KDA	ENSG00000145191	Leukoencephalopathy with vanishing white matter	11704758
ELOVL5	ELOVL fatty acid elongase 5	ENSG00000012660	Spinocerebellar Ataxia 38	25065913
EMG1	EMG1 NUCLEOLAR PROTEIN HOMOLOG (S. CEREVISIAE)	ENSG00000126749	Bowen-Conradi syndrome	19463982
EP300	E1A BINDING PROTEIN P300	ENSG00000100393	Rubinstein-Taybi syndrome	15706485
EPM2A	EPILEPSY, PROGRESSIVE MYOCLONUS TYPE 2A, LAFORA DISEASE (LAFORIN)	ENSG00000112425	progressive myoclonus epilepsy	9771710
ERBB4	v-erb-b2 avian erythroblastic leukemia viral oncogene homolog 4	ENSG00000178568	late-onset autosomal-dominant Amyotrophic Lateral Sclerosis	24119685
EXOSC3	exosome component 3	ENSG00000107371	pontocerebellar hypoplasia type 1	22544365
EZH2	enhancer of zeste 2 polycomb repressive complex 2 subunit	ENSG00000106462	Weaver syndrome	22177091
FA2H	FATTY ACID 2-HYDROXYLASE	ENSG00000103089	leukodystrophy with spastic paraparesis and dystonia	19068277
FAM134B	FAMILY WITH SEQUENCE SIMILARITY 134, MEMBER B	ENSG00000154153	Hereditary sensory and autonomic neuropathy type II	19838196
FAT4	FAT atypical cadherin 4	ENSG00000196159	recessive syndrome periventricular neuronal heterotopia	24056717
FGD1	FYVE, RHOGEF AND PH DOMAIN CONTAINING 1 (FACIOGENITAL DYSPLASIA)	ENSG00000102302	Non-syndromic X-linked mental retardation	11940089
FGF14	FIBROBLAST GROWTH FACTOR 14	ENSG00000102466	autosomal dominant cerebellar ataxia	12489043
FMN2	formin 2	ENSG00000155816	Nonsyndromic Autosomal-Recessive Intellectual Disability	25480035
FMR1	FRAGILE X MENTAL RETARDATION 1	ENSG00000102081	fragile X mental retardation	8490650
FOXP1	FORKHEAD BOX P1	ENSG00000114861	intellectual disability, autism and language impairment	20950788
FOXP2	TRINUCLEOTIDE REPEAT CONTAINING 10	ENSG00000128573	severe developmental verbal dyspraxia	11586359
FTSJ1	FTSJ HOMOLOG 1 (E. COLI)	ENSG00000068438	Nonsyndromic X-linked mental retardation	15162322
FUS	FUSION (INVOLVED IN T(12;16) IN MALIGNANT LIPOSARCOMA)	ENSG00000089280	Familial Amyotrophic Lateral Sclerosis	1925162
FXN	FRATAXIN	ENSG00000165060	Friedreich's ataxia	8596916
GABRG2	GAMMA-AMINOBUTYRIC ACID (GABA) A RECEPTOR, GAMMA 2	ENSG00000113327	Generalized epilepsy with febrile seizures plus	11326274
GAN	GIANT AXONAL NEUROPATHY (GIGAXONIN)	ENSG00000261609	Giant axonal neuropathy	11062483
GARS	GLYCYL-TRNA SYNTHETASE	ENSG00000106105	Charcot-Marie-Tooth disease type 2D/distal spinal muscular atrophy type V	12690580
GCH1	GTP CYCLOHYDROLASE 1 (DOPA-RESPONSIVE DYSTONIA)	ENSG00000131979	dopa responsive dystonia	7874165
GDAP1	GANGLIOSIDE-INDUCED DIFFERENTIATION-ASSOCIATED PROTEIN 1	ENSG00000104381	Charcot Marie Tooth 4A	11743579
GDI1	GDP DISSOCIATION INHIBITOR 1	ENSG00000203879	X-linked non-specific mental retardation	9620768

(Table S2) contd.....

GENE SYMBOL	GENE NAME	ENSEMBL ID	DISORDERS	PMID
GJB1	GAP JUNCTION PROTEIN, BETA 1, 32KDA	ENSG00000169562	X-linked Charcot-Marie-Tooth disease	8266101
GMPPA	GDP-mannose pyrophosphorylase A	ENSG00000144591	Intellectual Disability and Autonomic Dysfunction	24035193
GNB4	guanine nucleotide binding protein (G protein), beta polypeptide 4	ENSG00000114450	dominant intermediate Charcot-Marie-Tooth disease	23434117
GOSR2	golgi SNAP receptor complex member 2	ENSG00000108433	progressive myoclonus epilepsy with early ataxia	21549339
GRIA3	GLUTAMATE RECEPTOR, IONOTROPHIC, AMPA 3	ENSG00000125675	X-linked mental retardation	17989220
GRIK2	GLUTAMATE RECEPTOR, IONOTROPIC, KAINATE 2	ENSG00000164418	autosomal recessive mental retardation	17847003
GRIN2A	GLUTAMATE RECEPTOR, IONOTROPIC, N-METHYL D-ASPARTATE 2A	ENSG00000183454	idiopathic epilepsy and mental retardation	20890276
GRIN2A	glutamate receptor, ionotropic, N-methyl D-aspartate 2A	ENSG00000183454	Epilepsy-aphasia syndrome	23933818
GRIN2A	glutamate receptor, ionotropic, N-methyl D-aspartate 2A	ENSG00000183454	Idiopathic focal epilepsy with rolandic spikes	23933819
GRIN2B	GLUTAMATE RECEPTOR, IONOTROPIC, N-METHYL D-ASPARTATE 2B	ENSG00000273079	idiopathic epilepsy and mental retardation	20890276
GRM6	GLUTAMATE RECEPTOR, METABOTROPIC 6	ENSG00000113262	Night blindness	15781871
GRN	GRANULIN	ENSG00000030582	tau-negative frontotemporal dementia	16862116
GRXCR1	GLUTAREDOXIN, CYSTEINE RICH 1	ENSG00000215203	sensorineural autosomal-recessive nonsyndromic hearing impairment	20137778
HCN1	hyperpolarization activated cyclic nucleotide-gated potassium channel 1	ENSG00000164588	early infantile epileptic encephalopathy	24747641
HDAC4	HISTONE DEACETYLASE 4	ENSG00000068024	Brachydactyly mental retardation syndrome	20691407
HGF	HEPATOCYTE GROWTH FACTOR	ENSG00000019991	autosomal-recessive, nonsyndromic hearing loss	19576567
HIP1	HUNTINGTIN INTERACTING PROTEIN 1	ENSG00000127946	epilepsy and learning difficulties	21109226
HNRNPA1	heterogeneous nuclear ribonucleoprotein A1	ENSG00000135486	multisystem proteinopathy	23455423
HNRNPA2B1	heterogeneous nuclear ribonucleoprotein A2/B1	ENSG00000122566	multisystem proteinopathy	23455423
HSD17B10	HYDROXYSTEROID (17-BETA) DEHYDROGENASE 10	ENSG00000072506	X-linked mental retardation, choreoathetosis, and abnormal behavior.	17236142
HSPB1	HEAT SHOCK 27KDA PROTEIN 1	ENSG00000106211	autosomal dominant axonal Charcot-Marie-Tooth disease/distal hereditary motor neuropathy	15122254
HSPB8	HEAT SHOCK 22KDA PROTEIN 8	ENSG00000152137	distal hereditary motor neuropathy type II	15122253
HSPD1	HEAT SHOCK 60KDA PROTEIN 1 (CHAPERONIN)	ENSG00000144381	autosomal dominant spastic paraplegia	11898127
HTT	HUNTINGTIN (HUNTINGTON DISEASE)	ENSG00000197386	Huntington's disease	8458085
HUWE1	HECT, UBA and WWE domain containing 1, E3 ubiquitin protein ligase	ENSG00000086758	X-linked mental retardation	18252223
IGHMBP2	IMMUNOGLOBULIN MU BINDING PROTEIN 2	ENSG00000132740	spinal muscular atrophy with respiratory distress type 1	11528396
IGHMBP2	immunoglobulin mu binding protein 2	ENSG00000132740	Charcot-Marie Tooth Disease Type 2	25439726
IL1RAPL1	INTERLEUKIN 1 RECEPTOR ACCESSORY PROTEIN-LIKE 1	ENSG00000169306	X-linked mental retardation	10471494
IQSEC2	IQ MOTIF AND SEC7 DOMAIN 2	ENSG00000124313	nonsyndromic intellectual disability	20473311
ITM2B	INTEGRAL MEMBRANE PROTEIN 2B	ENSG00000136156	familial British dementia	10391242
JAM3	JUNCTIONAL ADHESION MOLECULE 3	ENSG00000166086	hemorrhagic destruction of the brain	21109224
KANSL1	KAT8 regulatory NSL complex subunit 1	ENSG00000120071	17q21.31 microdeletion syndrome	22544363
KAT6B	K(lysine) acetyltransferase 6B	ENSG00000156650	Say-Barber-Biesecker-Young-Simpson syndrome	22077973
KCNA1	POTASSIUM VOLTAGE-GATED CHANNEL, SHAKER-RELATED SUBFAMILY, MEMBER 1	ENSG00000111262	Episodic ataxia/myokymia syndrome	7842011

(Table S2) contd.....

GENE SYMBOL	GENE NAME	ENSEMBL ID	DISORDERS	PMID
KCNC1	potassium voltage-gated channel, Shaw-related subfamily, member 1	ENSG00000129159	progressive myoclonus epilepsy	25401298
KCNC3	POTASSIUM VOLTAGE-GATED CHANNEL, SHAW-RELATED SUBFAMILY, MEMBER 3	ENSG00000131398	autosomal dominant spinocerebellar ataxia	16501573
KCNJ18	POTASSIUM INWARDLY-RECTIFYING CHANNEL, SUBFAMILY J, MEMBER 12	ENSG00000184185	thyrotoxic hypokalemic periodic paralysis	20074522
KCNK18	POTASSIUM CHANNEL, SUBFAMILY K, MEMBER 18	ENSG00000186795	familial migraine with aura	20871611
KCNQ2	POTASSIUM VOLTAGE-GATED CHANNEL, KQT-LIKE SUBFAMILY, MEMBER 2	ENSG00000075043	benign familial neonatal convulsions	9425895
KCNQ3	POTASSIUM VOLTAGE-GATED CHANNEL, KQT-LIKE SUBFAMILY, MEMBER 3	ENSG00000184156	benign familial neonatal convulsions	9425900
KDM5C	lysine (K)-specific demethylase 5C	ENSG00000126012	X-linked mental retardation	15586325
KIAA1279	KIAA1279	ENSG00000198954	Goldberg-Shprintzen syndrome	15883926
KIF2A	kinesin heavy chain member 2A	ENSG00000068796	malformations of cortical development and microcephaly	23603762
KIF5A	KINESIN FAMILY MEMBER 5A	ENSG00000155980	hereditary spastic paraplegia	12355402
KIF5C	kinesin family member 5C	ENSG00000168280	malformations of cortical development and microcephaly	23603762
KIRREL3	KIN OF IRRE LIKE 3 (DROSOPHILA)	ENSG00000149571	intellectual disability	19012874
KMT2A	lysine (K)-specific methyltransferase 2A	ENSG00000118058	Wiedemann-Steiner syndrome	22795537
KMT2C	lysine (K)-specific methyltransferase 2C	ENSG00000055609	Kleefstra syndrome phenotypic spectrum	22726846
KMT2D	lysine (K)-specific methyltransferase 2D	ENSG00000167548	Kabuki syndrome	20711175
L1CAM	L1 CELL ADHESION MOLECULE	ENSG00000198910	X-linked hydrocephalus	1303258
L1CAM	L1 CELL ADHESION MOLECULE	ENSG00000198910	X-linked spastic paraplegia/MASA syndrome	7920659
LAMA1	laminin, alpha 1	ENSG00000101680	Cerebellar Dysplasia and Cysts with and without Retinal Dystrophy	25105227
LGI1	LEUCINE-RICH, GLIOMA INACTIVATED 1	ENSG00000108231	autosomal-dominant partial epilepsy with auditory features	11810107
LMNA	LAMIN A/C	ENSG00000160789	autosomal dominant Emery-Dreifuss muscular dystrophy	10080180
LRRK2	LEUCINE-RICH REPEAT KINASE 2	ENSG00000188906	autosomal-dominant, late-onset parkinsonism	15541309
LRTOMT	LEUCINE RICH TRANSMEMBRANE AND 0-METHYLTRANSFERASE DOMAIN CONTAINING	ENSG00000184154	profound nonsyndromic hearing loss	18953341
MAGEL2	MAGE-like 2	ENSG00000254585	Prader-Willi phenotypes and autism	24076603
MAPT	MICROTUBULE-ASSOCIATED PROTEIN TAU	ENSG00000186868	frontotemporal dementia and parkinsonism	9641683
MBD5	methyl-CpG binding domain protein 5	ENSG00000204406	Kleefstra syndrome phenotypic spectrum	22726846
MECP2	METHYL CPG BINDING PROTEIN 2 (RETT SYNDROME)	ENSG00000169057	Rett syndrome	10508514
MED12	mediator complex subunit 12	ENSG00000184634	Opitz-Kaveggia syndrome	17334363
MED17	MEDIATOR COMPLEX SUBUNIT 17	ENSG00000042429	Infantile cerebral and cerebellar atrophy	20950787
MED23	mediator complex subunit 23	ENSG00000112282	nonsyndromic autosomal recessive intellectual disability	21868677
MFN2	MITOFUSIN 2	ENSG00000116688	Charcot-Marie-Tooth neuropathy type 2A	15064763
MPZ	MYELIN PROTEIN ZERO (CHARCOT-MARIE-TOOTH NEUROPATHY 1B)	ENSG00000158887	Charcot-Marie-Tooth neuropathy type 1B	7693129
MRE11A	MRE11 MEIOTIC RECOMBINATION 11 HOMOLOG A (S. CEREVISIAE)	ENSG00000020922	ataxia-telangiectasia-like disorder	10612394
NAIP	BACULOVIRAL IAP REPEAT-CONTAINING 1	ENSG00000249437	spinal muscular atrophy	7813013
NALCN	sodium leak channel, non-selective	ENSG00000102452	Autosomal-Recessive Syndrome with Severe Hypotonia, Speech Impairment and Cognitive Delay	24075186
NDRG1	N-MYC DOWNSTREAM REGULATED GENE 1	ENSG00000104419	hereditary motor and sensory neuropathy-Lom	10831399
NEFH	NEUROFILAMENT, HEAVY POLYPEPTIDE 200KDA	ENSG00000100285	amyotrophic lateral sclerosis	7849698
NF1	NEUROFIBROMIN 1	ENSG00000196712	neurofibromatosis type 1	1694727

(Table S2) contd.....

GENE SYMBOL	GENE NAME	ENSEMBL ID	DISORDERS	PMID
NIPA1	NON IMPRINTED IN PRADER-WILLI/ANGELMAN SYNDROME 1	ENSG00000170113	autosomal dominant hereditary spastic paraplegia	14508710
NIPBL	NIPPED-B HOMOLOG (DROSOPHILA)	ENSG00000164190	Cornelia de Lange syndrome	15146186
NLGN3	NEUROLIGIN 3	ENSG00000196338	autism spectrum disorders	12669065
NLGN4X	neuroligin 4, X-linked	ENSG00000146938	autism spectrum disorders	12669065
NMNAT1	nicotinamide nucleotide adenylyltransferase 1	ENSG00000173614	Leber congenital amaurosis	22842227
NOP56	NOP56 ribonucleoprotein	ENSG00000101361	Spinocerebellar Ataxia 36	21683323
NOTCH3	NOTCH HOMOLOG 3 (DROSOPHILA)	ENSG00000074181	cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy	8878478
NR1I3	nuclear receptor subfamily 1, group I, member 3	ENSG00000143257	Kleefstra syndrome phenotypic spectrum	22726846
NR2F1	nuclear receptor subfamily 2, group F, member 1	ENSG00000175745	optic nerve atrophy with intellectual disability.	24462372
NRXN1	NEUREXIN 1	ENSG00000179915	Autosomal-Recessive Pitt-Hopkins-like Mental Retardation	19896112
NSUN2	NOP2/Sun RNA methyltransferase family, member 2	ENSG00000037474	autosomal-recessive intellectual disability	22541562
NTRK1	NEUROTROPHIC TYROSINE KINASE, RECEPTOR, TYPE 1	ENSG00000198400	Congenital insensitivity to pain with anhidrosis	8696348
NTRK2	NEUROTROPHIC TYROSINE KINASE, RECEPTOR, TYPE 2	ENSG00000148053	severe obesity and developmental delay	15494731
PACS1	phosphofurin acidic cluster sorting protein 1	ENSG00000175115	syndromic intellectual disability	23159249
PARK2	PARKINSON DISEASE (AUTOSOMAL RECESSIVE, JUVENILE) 2, PARKIN	ENSG00000185345	autosomal recessive juvenile parkinsonism	9560156
PARK7	PARKINSON DISEASE (AUTOSOMAL RECESSIVE, EARLY ONSET) 7	ENSG00000116288	autosomal recessive early-onset parkinsonism	12446870
PCDH19	PROTOCADHERIN 19	ENSG00000165194	Epilepsy and mental retardation limited to females	18469813
PDE4B	PHOSPHODIESTERASE 4B, CAMP-SPECIFIC	ENSG00000184588	schizophrenia	16293762
PDE8B	phosphodiesterase 8B	ENSG00000113231	Autosomal-dominant striatal degeneration	20085714
PDYN	PRODYNORPHIN	ENSG00000101327	spinocerebellar ataxia type 23	21035104
PER2	period circadian clock 2	ENSG00000132326	Familial advanced sleep phase syndrome	11232563
PFN1	profilin 1	ENSG00000108518	familial amyotrophic lateral sclerosis	22801503
PHF21A	PHD finger protein 21A	ENSG00000135365	Intellectual disability	22770980
PHF6	PHD FINGER PROTEIN 6	ENSG00000156531	Börjeson-Forssman-Lehmann syndrome	12415272
PIGO	phosphatidylinositol glycan anchor biosynthesis, class O	ENSG00000165282	Hyperphosphatasia with mental retardation syndrome	22683086
PINK1	PTEN INDUCED PUTATIVE KINASE 1	ENSG00000158828	Hereditary early-onset Parkinson's disease	15087508
PLEC	PLECTIN 1, INTERMEDIATE FILAMENT BINDING PROTEIN 500KDA	ENSG00000178209	Autosomal-Recessive Limb-Girdle Muscular Dystrophy	21109228
PLP1	PROTEOLIPID PROTEIN 1	ENSG00000123560	Pelizaeus-Merzbacher disease	2773936
PMP22	PERIPHERAL MYELIN PROTEIN 22	ENSG00000109099	Charcot-Marie-Tooth disease type 1A	1303281
PNKP	POLYNUCLEOTIDE KINASE 3'-PHOSPHATASE	ENSG00000039650	microcephaly and intractable seizures	20118933
POLG	POLYMERASE (DNA DIRECTED), GAMMA	ENSG00000140521	progressive external ophthalmoplegia	11431686
PQBP1	POLYGLUTAMINE BINDING PROTEIN 1	ENSG00000102103	X-linked mental retardation	14634649
PRICKLE1	PRICKLE HOMOLOG 1 (DROSOPHILA)	ENSG00000139174	autosomal-recessive progressive myoclonus epilepsy-ataxia syndrome	18976727
PRKCG	PROTEIN KINASE C, GAMMA	ENSG00000126583	dominant nonepisodic cerebellar ataxia	12644968
PRKRA	PROTEIN KINASE, INTERFERON-INDUCIBLE ds-RNA DEPENDENT ACTIVATOR	ENSG00000180228	young-onset dystonia-parkinsonism disorder	18243799
PRNP	PRION PROTEIN	ENSG00000171867	Gerstmann-Straeussler syndrome	2564168
PRPH	PERIPHERIN	ENSG00000135406	amyotrophic lateral sclerosis	15446584
PRPS1	PHOSPHORIBOSYL PYROPHOSPHATE SYNTHETASE 1	ENSG00000147224	nonsyndromic X-linked sensorineural deafness	20021999

(Table S2) contd.....

GENE SYMBOL	GENE NAME	ENSEMBL ID	DISORDERS	PMID
PRSS12	PROTEASE, SERINE, 12 (NEUROTRYPSIN, MOTOPSIN)	ENSG00000164099	autosomal recessive nonsyndromic mental retardation	12459588
PSEN1	PRESENILIN 1 (ALZHEIMER DISEASE 3)	ENSG00000080815	familial frontotemporal dementia	11895378
PSEN1	PRESENILIN 1 (ALZHEIMER DISEASE 3)	ENSG00000080815	familial Alzheimer's disease	7596406
PSEN2	PRESENILIN 2 (ALZHEIMER DISEASE 4)	ENSG00000143801	Familial Alzheimer's disease	7651536
PTPRQ	PROTEIN TYROSINE PHOSPHATASE, RECEPTOR TYPE, Q	ENSG00000139304	autosomal-recessive nonsyndromic hearing impairment	20346435
PURA	purine-rich element binding protein A	ENSG00000185129	Profound Neonatal Hypotonia, Seizures, and Encephalopathy	25439098
RAB39B	RAB39B, member RAS oncogene family	ENSG00000155961	X-Linked Intellectual Disability and Early-Onset Parkinson Disease	25434005
RAI1	RETINOIC ACID INDUCED 1	ENSG00000108557	Smith-Magenis syndrome	12652298
REEP1	RECEPTOR ACCESSORY PROTEIN 1	ENSG00000068615	hereditary spastic paraplegia type 31	16826527
REEP1	receptor accessory protein 1	ENSG00000068615	distal hereditary motor neuropathy type V	22703882
RELN	REELIN	ENSG00000189056	Autosomal recessive lissencephaly with cerebellar hypoplasia	10973257
ROGDI	rogdi homolog (Drosophila)	ENSG00000067836	Kohlschütter-Tönz Syndrome	22424600
RTTN	rotatin	ENSG00000176225	bilateral diffuse polymicrogyria	22939636
SACS	SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (SACSIN)	ENSG00000151835	Autosomal recessive spastic ataxia	10655055
SCN11A	sodium channel, voltage-gated, type XI, alpha subunit	ENSG00000168356	congenital inability to experience pain	24036948
SCN1A	SODIUM CHANNEL, VOLTAGE-GATED, TYPE I, ALPHA	ENSG00000144285	generalized epilepsy with febrile seizures plus	10742094
SCN1B	SODIUM CHANNEL, VOLTAGE-GATED, TYPE I, BETA	ENSG00000105711	generalized epilepsy with febrile seizures plus	9697698
SCN8A	sodium channel, voltage gated, type VIII, alpha subunit	ENSG00000196876	Infantile Epileptic Encephalopathy	22365152
SDHA	SUCCINATE DEHYDROGENASE COMPLEX, SUBUNIT A, FLAVOPROTEIN (FP)	ENSG00000073578	Leigh syndrome	7550341
SDHAF1	SUCCINATE DEHYDROGENASE COMPLEX ASSEMBLY FACTOR 1	ENSG00000205138	succinate dehydrogenase-defective infantile leukoencephalopathy	19465911
SERPINB6	SERPIN PEPTIDASE INHIBITOR, CLADE B (OVALBUMIN), MEMBER 6	ENSG00000124570	autosomal-recessive nonsyndromic sensorineural hearing loss	20451170
SERPINI1	SERPIN PEPTIDASE INHIBITOR, CLADE I (NEUROSERPIN), MEMBER 1	ENSG00000163536	autosomal dominantly inherited dementia	10517635
SETD5	SET domain containing 5	ENSG00000168137	Intellectual Disability	24680889
SETX	SENATAXIN	ENSG00000107290	Ataxia-ocular apraxia 2	14770181
SETX	SENATAXIN	ENSG00000107290	juvenile amyotrophic lateral sclerosis	15106121
SGCE	SARCOGLYCAN, EPSILON	ENSG00000127990	myoclonus-dystonia syndrome	11528394
SHANK2	SH3 AND MULTIPLE ANKYRIN REPEAT DOMAINS 2	ENSG00000162105	autism spectrum disorder and mental retardation.	20473310
SHANK3	SH3 AND MULTIPLE ANKYRIN REPEAT DOMAINS 3	ENSG00000251322	autism spectrum disorders	17173049
SHANK3	SH3 AND MULTIPLE ANKYRIN REPEAT DOMAINS 3	ENSG00000251322	schizophrenia	20385823
SHH	SONIC HEDGEHOG HOMOLOG (DROSOPHILA)	ENSG00000164690	autosomal dominant holoprosencephaly	8896572
SLC12A6	SOLUTE CARRIER FAMILY 12 (POTASSIUM/CHLORIDE TRANSPORTERS), MEMBER 6	ENSG00000140199	Peripheral neuropathy associated with agenesis of the corpus callosum	12368912
SLC24A1	SOLUTE CARRIER FAMILY 24 (SODIUM/POTASSIUM/CALCIUM EXCHANGER), MEMBER 1	ENSG00000074621	autosomal-recessive congenital stationary night blindness	20850105

(Table S2) contd.....

GENE SYMBOL	GENE NAME	ENSEMBL ID	DISORDERS	PMID
SLC6A3	SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER, DOPAMINE), MEMBER 3	ENSG00000142319	infantile parkinsonism-dystonia	19478460
SLC6A8	SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER, CREATINE), MEMBER 8	ENSG00000130821	X-Linked Mental Retardation with Seizures	11898126
SLC9A6	SOLUTE CARRIER FAMILY 9 (SODIUM/HYDROGEN EXCHANGER), MEMBER 6	ENSG00000198689	X-linked mental retardation (XLMR) syndrome	18342287
SLITRK1	SLIT AND NTRK-LIKE FAMILY, MEMBER 1	ENSG00000178235	Tourette's syndrome	16224024
SMARCA2	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 2	ENSG00000080503	Nicolaides-Baraitser syndrome	22366787
SMARCA4	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 4	ENSG00000127616	Coffin-Siris syndrome	22426308
SMARCB1	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily b, member 1	ENSG00000099956	Kleefstra syndrome phenotypic spectrum	22726846
SMARCB1	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily b, member 1	ENSG00000099956	Coffin-Siris syndrome	22426308
SMPX	small muscle protein, X-linked	ENSG00000091482	X-chromosomal hearing impairment	21549336
SNCA	SYNUCLEIN, ALPHA (NON A4 COMPONENT OF AMYLOID PRECURSOR)	ENSG00000145335	autosomal-dominant parkinsonism	9197268
SNCB	SYNUCLEIN, BETA	ENSG00000074317	dementia with Lewy bodies	15365127
SNX14	sorting nexin 14	ENSG00000135317	Cerebellar Ataxia and Intellectual Disability Syndrome	25439728
SOBP	SINE OCULIS BINDING PROTEIN HOMOLOG (DROSOPHILA)	ENSG00000112320	autosomal-recessive mental retardation	21035105
SOD1	SUPEROXIDE DISMUTASE 1, SOLUBLE (AMYOTROPHIC LATERAL SCLEROSIS 1 (ADULT))	ENSG00000142168	autosomal dominant familial amyotrophic lateral sclerosis	8446170
SOX3	SRY (SEX DETERMINING REGION Y)-BOX 3	ENSG00000134595	X-linked mental retardation with growth hormone deficiency	12428212
SPAST	SPASTIN	ENSG00000021574	Autosomal dominant hereditary spastic paraplegia	10610178
SPG7	CELL MATRIX ADHESION REGULATOR	ENSG00000197912	Hereditary spastic paraplegia	9635427
SPTLC2	SERINE PALMITOYLTRANSFERASE, LONG CHAIN BASE SUBUNIT 2	ENSG00000100596	Hereditary sensory and autonomic neuropathy type I	20920666
STAMBP	STAM binding protein	ENSG00000124356	Microcephaly-capillary malformation	23542699
STX1B	syntaxin 1B	ENSG00000099365	fever-associated epilepsy syndromes.	25362483
STXBP1	SYNTAXIN BINDING PROTEIN 1	ENSG00000136854	Early infantile epileptic encephalopathy with suppression-burst	18469812
SYN1	synapsin I	ENSG00000008056	autism	21441247
SYN1	synapsin I	ENSG00000008056	partial epilepsy	21441247
SYNGAP1	synaptic Ras GTPase activating protein 1	ENSG00000197283	epileptic encephalopathy	23708187
TARDBP	TAR DNA BINDING PROTEIN	ENSG00000120948	familial and sporadic amyotrophic lateral sclerosis	18309045
TBC1D24	TBC1 DOMAIN FAMILY, MEMBER 24	ENSG00000162065	familial infantile myoclonic epilepsy	20727515
TECPR2	tectonin beta-propeller repeat containing 2	ENSG00000196663	autosomal-recessive hereditary spastic paraparesis	23176824
TH	TYROSINE HYDROXYLASE	ENSG00000180176	Segawa's syndrome	7814018
THAP1	THAP DOMAIN CONTAINING, APOPTOSIS ASSOCIATED PROTEIN 1	ENSG00000131931	primary torsion dystonia	19182804
TOR1A	TORSIN FAMILY 1, MEMBER A (TORSIN A)	ENSG00000136827	Early-onset torsion dystonia	9288096
TPRN	CHROMOSOME 9 OPEN READING FRAME 75	ENSG00000176058	nonsyndromic deafness	20170899

(Table S1) contd.....

GENE SYMBOL	GENE NAME	ENSEMBL ID	DISORDERS	PMID
TRAPPC9	TRAFFICKING PROTEIN PARTICLE COMPLEX 9	ENSG00000167632	nonsyndromic autosomal-recessive mental retardation	20004765
TRPM1	TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY M, MEMBER 1	ENSG00000134160	autosomal-recessive complete congenital stationary night blindness	19896113
TRPV4	TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY V, MEMBER 4	ENSG00000111199	Spinal muscular atrophies	20037588
TUBA8	TUBULIN, ALPHA 8	ENSG00000183785	polymicrogyria with optic nerve hypoplasia	19896110
TUBB2B	TUBULIN, BETA 2B	ENSG00000137285	asymmetrical polymicrogyria	19465910
TUBB3	TUBULIN, BETA 3	ENSG00000198211	complex polyneuropathies	20074521
TUBB4A	tubulin, beta 4A class IVa	ENSG00000104833	Hypomyelination with atrophy of the basal ganglia and cerebellum	23582646
TUBG1	tubulin, gamma 1	ENSG00000131462	malformations of cortical development and microcephaly	23603762
UBE2A	UBIQUITIN-CONJUGATING ENZYME E2A (RAD6 HOMOLOG)	ENSG00000077721	X-linked mental retardation	16909393
UBR1	UBIQUITIN PROTEIN LIGASE E3 COMPONENT N-RECOGNIN 1	ENSG00000159459	Johanson-Blizzard syndrome	16311597
UPF3B	UPF3 REGULATOR OF NONSENSE TRANSCRIPTS HOMOLOG B (YEAST)	ENSG00000125351	syndromic and nonsyndromic mental retardation	17704778
VANGL1	VANG-LIKE 1 (VAN GOGH, DROSOPHILA)	ENSG00000173218	neural-tube defects	17409324
VAPB	VAMP (VESICLE-ASSOCIATED MEMBRANE PROTEIN)-ASSOCIATED PROTEIN B AND C	ENSG00000124164	atypical amyotrophic lateral sclerosis	15372378
VCP	VALOSIN-CONTAINING PROTEIN	ENSG00000165280	Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia	15034582
VLDLR	VERY LOW DENSITY LIPOPROTEIN RECEPTOR	ENSG00000147852	autosomal recessive cerebellar hypoplasia with cerebral gyral simplification	16080122
VPS13B	vacuolar protein sorting 13 homolog B (yeast)	ENSG00000132549	Cohen syndrome	12730828
VPS35	vacuolar protein sorting 35 homolog (S. cerevisiae)	ENSG00000069329	late-onset Parkinson disease	21763483
VPS37A	vacuolar protein sorting 37 homolog A (S. cerevisiae)	ENSG00000155975	autosomal recessive complex hereditary spastic paraparesis	22717650
WDR45	WD repeat domain 45	ENSG00000196998	Static encephalopathy of childhood with neurodegeneration in adulthood	23435086
WDR62	WD REPEAT DOMAIN 62	ENSG00000075702	microcephaly	20890278
WNK1	WNK lysine deficient protein kinase 1	ENSG00000060237	hereditary sensory and autonomic neuropathy type II	15060842
ZC4H2	zinc finger, C4H2 domain containing	ENSG00000126970	Arthrogryposis Multiplex Congenita and Intellectual Disability	23623388
ZDHHC9	ZINC FINGER, DHHC-TYPE CONTAINING 9	ENSG00000188706	X-linked mental retardation	17436253

Table S2. Functional clustering of NPD genes.

Gene Group 1	**Enrichment Score: 11**
1	VANGL1 vang-like 1 (van gogh, Drosophila)
2	ABHD12 abhydrolase domain containing 12
3	REEP1 receptor accessory protein 1
4	NIPA1 non imprinted in Prader-Willi/Angelman syndrome 1
5	FAM134B family with sequence similarity 134, member B
Gene Group 2	**Enrichment Score: 8.13**
1	VPS35 hypothetical protein LOC100133770; vacuolar protein sorting 35 homolog (S. cerevisiae)
2	VPS37A vacuolar protein sorting 37 homolog A (S. cerevisiae)
3	CHMP2B chromatin modifying protein 2B
4	VPS13B vacuolar protein sorting 13 homolog B (yeast)
Gene Group 3	**Enrichment Score: 7.05**

(Table S2) contd.....

1	KCNQ3 potassium voltage-gated channel, KQT-like subfamily, member 3
2	KCNQ2 potassium voltage-gated channel, KQT-like subfamily, member 2
3	TRPM1 transient receptor potential cation channel, subfamily M, member 1
4	CACNA1C hypothetical protein LOC100131098; calcium channel, voltage-dependent, L type, alpha 1C subunit
5	CACNA1A calcium channel, voltage-dependent, P/Q type, alpha 1A subunit
6	SLC6A8 solute carrier family 6 (neurotransmitter transporter, creatine), member 8
7	SLC12A6 solute carrier family 12 (potassium/chloride transporters), member 6
8	KCNJ18 similar to hkir2.2x; similar to inward rectifying K+ channel negative regulator Kir2.2v; potassium inwardly-rectifying channel, subfamily J, member 12
9	CLCN2 chloride channel 2
10	KCNC3 potassium voltage-gated channel, Shaw-related subfamily, member 3
11	ANO5 anoctamin 5
12	CHRNA2 cholinergic receptor, nicotinic, alpha 2 (neuronal)
13	ANO10 anoctamin 10
14	SCN1A sodium channel, voltage-gated, type I, alpha subunit
15	SLC24A1 solute carrier family 24 (sodium/potassium/calcium exchanger), member 1
16	SCN1B sodium channel, voltage-gated, type I, beta
17	HCN1 hyperpolarization activated cyclic nucleotide-gated potassium channel 1
18	GABRG2 gamma-aminobutyric acid (GABA) A receptor, gamma 2
19	CACNB4 calcium channel, voltage-dependent, beta 4 subunit
20	STX1B syntaxin 1B
21	CACNA1H calcium channel, voltage-dependent, T type, alpha 1H subunit
22	NALCN sodium leak channel, non-selective
23	KCNC1 potassium voltage-gated channel, Shaw-related subfamily, member 1
24	SLC9A6 solute carrier family 9 (sodium/hydrogen exchanger), member 6
25	SCN8A sodium channel, voltage gated, type VIII, alpha subunit
26	KCNK18 potassium channel, subfamily K, member 18
27	SCN11A sodium channel, voltage-gated, type XI, alpha subunit
28	TRPV4 transient receptor potential cation channel, subfamily V, member 4
Gene Group 4	**Enrichment Score: 6.15**
1	GRIN2A glutamate receptor, ionotropic, N-methyl D-aspartate 2A
2	GRIA3 glutamate receptor, ionotrophic, AMPA 3
3	GRIN2B glutamate receptor, ionotropic, N-methyl D-aspartate 2B
4	CHRNA2 cholinergic receptor, nicotinic, alpha 2 (neuronal)
5	GRIK2 glutamate receptor, ionotropic, kainate 2
6	GABRG2 gamma-aminobutyric acid (GABA) A receptor, gamma 2
7	CHRNB2 cholinergic receptor, nicotinic, beta 2 (neuronal)
8	CHRNA4 cholinergic receptor, nicotinic, alpha 4
Gene Group 5	**Enrichment Score: 5.01**
1	PRKCG protein kinase C, gamma
2	CSNK1D casein kinase 1, delta
3	CDKL5 cyclin-dependent kinase-like 5
4	WNK1 WNK lysine deficient protein kinase 1; hypothetical LOC100132369
5	BCKDK branched chain ketoacid dehydrogenase kinase
6	PINK1 PTEN induced putative kinase 1
7	CASK calcium/calmodulin-dependent serine protein kinase (MAGUK family)
8	LRRK2 leucine-rich repeat kinase 2
Gene Group 6 Enrichment Score: 4.46	
1	PHF6 PHD finger protein 6
2	KDM5C lysine (K)-specific demethylase 5C
3	PHF21A PHD finger protein 21A
4	THAP1 THAP domain containing, apoptosis associated protein 1
5	RAI1 retinoic acid induced 1
Gene Group 7	**Enrichment Score: 4.23**
1	CHD8 chromodomain helicase DNA binding protein 8

(Table S2) contd.....

2	SETX senataxin
3	IGHMBP2 immunoglobulin mu binding protein 2
4	SMARCA2 SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 2
5	ATRX alpha thalassemia/mental retardation syndrome X-linked (RAD54 homolog, S. cerevisiae)
6	CHD7 chromodomain helicase DNA binding protein 7
7	CHD2 chromodomain helicase DNA binding protein 2
8	SMARCA4 SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 4
Gene Group 8	**Enrichment Score: 3.92**
1	KIF2A kinesin heavy chain member 2A
2	CEP135 centrosomal protein 135kDa
3	DISC1 disrupted in schizophrenia 1
4	CEP152 centrosomal protein 152kDa
Gene Group 9	**Enrichment Score: 3.87 RG**
1	KIF5A kinesin family member 5A
2	KIF2A kinesin heavy chain member 2A
3	DYNC1H1 dynein, cytoplasmic 1, heavy chain 1
4	KIF5C kinesin family member 5C
Gene Group 10	**Enrichment Score: 3.3**
1	HNRNPA2B1 heterogeneous nuclear ribonucleoprotein A2/B1
2	FUS fusion (involved in t(12;16) in malignant liposarcoma)
3	TARDBP TAR DNA binding protein
4	HNRNPA1 heterogeneous nuclear ribonucleoprotein A1-like 3; similar to heterogeneous nuclear ribonucleoprotein A1; heterogeneous nuclear ribonucleoprotein A1 pseudogene 2; heterogeneous nuclear ribonucleoprotein A1; heterogeneous nuclear ribonucleoprotein A1 pseudogene
5	UPF3B UPF3 regulator of nonsense transcripts homolog B (yeast)
6	FMR1 fragile X mental retardation 1
Gene Group 11	**Enrichment Score: 2.69**
1	TUBB2B tubulin, beta 2B
2	TUBB3 tubulin, beta 3; melanocortin 1 receptor (alpha melanocyte stimulating hormone receptor)
3	TUBA8 tubulin, alpha 8
4	TUBG1 tubulin, gamma 1; similar to Tubulin, gamma 1
Gene Group 12	**Enrichment Score: 2.23**
1	AP4M1 adaptor-related protein complex 4, mu 1 subunit
2	PACS1 phosphofurin acidic cluster sorting protein 1
3	AP4S1 adaptor-related protein complex 4, sigma 1 subunit
4	AP4E1 adaptor-related protein complex 4, epsilon 1 subunit
5	GOSR2 golgi SNAP receptor complex member 2
6	AP4B1 adaptor-related protein complex 4, beta 1 subunit
7	AP1S2 adaptor-related protein complex 1, sigma 2 subunit pseudogene; adaptor-related protein complex 1, sigma 2 subunit
Gene Group 13	**Enrichment Score: 2.09**
1	EXOSC3 exosome component 3
2	NSUN2 NOL1/NOP2/Sun domain family, member 2
3	NOP56 NOP56 ribonucleoprotein homolog (yeast)
4	EMG1 EMG1 nucleolar protein homolog (S. cerevisiae)
Gene Group 14	**Enrichment Score: 2.01**
1	MED12 mediator complex subunit 12
2	SMARCB1 SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily b, member 1
3	SMARCA2 SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 2
4	ARID1B AT rich interactive domain 1B (SWI1-like)
5	PQBP1 polyglutamine binding protein 1
6	ARID1A AT rich interactive domain 1A (SWI-like)
7	MED17 mediator complex subunit 17
8	MED23 mediator complex subunit 23
Gene Group 15	**Enrichment Score: 1.41**
1	FOXP2 forkhead box P2

(Table S2) contd.....

	2 PHF21A PHD finger protein 21A
	3 NR2F1 nuclear receptor subfamily 2, group F, member 1
4	MECP2 methyl CpG binding protein 2 (Rett syndrome)
5	FOXP1 forkhead box P1
6	PURA purine-rich element binding protein A
Gene Group 16	**Enrichment Score: 1.19**
1	AHI1 Abelson helper integration site 1
2	WDR45 WD repeat domain 45
3	BRWD3 bromodomain and WD repeat domain containing 3
4	WDR62 WD repeat domain 62
5	GNB4 guanine nucleotide binding protein (G protein), beta polypeptide 4
Gene Group 17	**Enrichment Score: 1.19**
1	PCDH19 protocadherin 19
2	CNTNAP2 contactin associated protein-like 2
3	NLGN4X neuroligin 4, X-linked
4	SGCE sarcoglycan, epsilon
5	CDH15 cadherin 15, type 1, M-cadherin (myotubule)
6	NRXN1 neurexin 1
7	JAM3 junctional adhesion molecule 3
8	IL1RAPL1 interleukin 1 receptor accessory protein-like 1
9	KIRREL3 kin of IRRE like 3 (Drosophila)
10	FAT4 FAT tumor suppressor homolog 4 (Drosophila)
11	DCHS1 dachsous 1 (Drosophila)
12	ANO5 anoctamin 5
13	SCN1B sodium channel, voltage-gated, type I, beta

Table S3. List of NPD genes known to be druggable.

ABCB7	CACNA1H	FUS	KCNQ2	PINK1	SLC6A3
ABCD1	CASK	GABRG2	KCNQ3	PRKCG	SLC6A8
AGTR2	CDKL5	GRIA3	LRRK2	PRPS1	SLC9A6
ALDH7A1	CHRNA2	GRIK2	NLGN3	PRSS12	SOD1
ATM	CHRNA4	GRIN2A	NLGN4X	PTPRQ	TH
ATP1A3	CHRNB2	GRIN2B	NR1I3	SCN11A	TRPM1
ATR	CLCN2	GRM6	NR2F1	SCN1A	TRPV4
BCKDK	CSNK1D	HCN1	NTRK1	SCN8A	TUBA8
CA8	DRD2	HGF	NTRK2	SERPINB6	TUBB3
CACNA1A	EPM2A	KCNC1	PDE4B	SERPINI1	TUBG1
CACNA1C	ERBB4	KCNC3	PDE8B	SLC12A6	WNK1

Table S4. Characterization of multi-species conserved sequences of NPDs genes considered as less conserved in Hominoides species.

	GENE	**Chromosome**	**% Identity Chimpanzee***	**% Identity gorilla***	**% Identity Orangutan***	**% Identity gibbon***	**Pairwise % Identity in Hominoides (protein)**	**% identical site (protein)**
1	REEP1	2	92	77	80	91	55,1	13,5
2	SDHA	5	99	22	22	46	53,1	13,6
3	NRXN1	2	Deletion	Deletion	74	25	61,6	25,8
4	FMN2	1	60	62	80	91	60,5	31
5	SMARCA2	9	Deletion	90	97	80	67,8	41,1
6	SLC6A8	X	Deletion	?	80	82	69,4	46**
7	MAGEL2	15	Deletion	53	Deletion	53	79,1	51,6
8	ASPM	1	57	98	72	95	76,1	52,1
9	ARX	X	Deletion	91,9	53	36	76,2	53,4

(Table S4) contd.....

	GENE	Chromosome	% Identity Chimpanzee*	% Identity gorilla*	% Identity Orangutan*	% Identity gibbon*	Pairwise % Identity in Hominoides (protein)	% identical site (protein)
10	SLC24A1	15	99	83	81	63	72	53,4
11	SCN1B	19	59	56	59	74	75,9	54,6
12	SPG7	16	64	95	98	93	82,5	58,2
13	DISC1	1	85	86	79	23	79	58,4
14	NMNAT1	1	86	86	76	89	80,1	60,8
15	ALDH7A1	5	Deletion	Deletion	93	98	84	63
16	ATP13A2	1	91	86	98	16	82,8	63,3
17	DST	6	67	96	96	92	78,6	63,7
18	PARK2	6	99	Deletion	71	62	81,3	63,7
19	IL1RAPL1	X	63	63	100	63	86,4	66,1
20	SHANK2	11	98	62	68	79	83,1	69,8**
21	KIF2A	5	61	78	89	97	88,2	73,1
22	CACNA1H	16	Deletion	96	83	Deletion	79,9	73,4
23	TPRN	9	74	82	68	69	90,4	73,6**
24	NAIP	5	99	55	96	96	86,9	73,8
25	CACNA1A	19	91	75	78	95	86,9	74,8
26	CADM1	11	85	89	89	85	90,6	79,2
27	ARID1B	6	83	79	76	66	92,7	85,5**
28	KIF5C	2	90	90	95	81	99,8	99,3**

*Data from Ensembl database; **gene with unique protein structure

Table S5. Coincidence of NPD genes with fragile region on human X chromosome. * Coordinates of fragile regions of human X chromosome, proposed by Prada and Laissue (20).

GENE	Chromosome	Gene Start (bp) hg38	Gene End (bp) hg38	Gene Size (bp)	Band	Fragile region*
NLGN4X	X	5840637	6228863	388226	p22.31	FR 3
AP1S2	X	15825806	15855014	29208	p22.2	
CDKL5	X	18425583	1865,629	228046	p22.13	
SMPX	X	21705972	21758163	52191	p22.12	
ARX	X	25003694	25015948	12254	p21.3	
IL1RAPL1	X	28587399	29956723	1369324	p21.3	
CASK	X	41514934	41923463	408529	p11.4	
SYN1	X	47571898	47619853	47955	p11.23	
FTSJ1	X	48476021	48486364	10343	p11.23	FR9
PQBP1	X	48897912	48903143	5231	p11.23	
WDR45	X	49074433	49101170	26737	p11.23	
KDM5C	X	53191321	53225422	34101	p11.22	FR11
IQSEC2	X	53232876	53321324	88448	p11.22	
HSD17B10	X	53431258	53434373	3115	p11.22	
HUWE1	X	53532096	53686729	154633	p11.22	
FGD1	X	54445454	54496166	50712	p11.22	
ZC4H2	X	64915802	65034713	118911	q11.2	FR14
DLG3	X	70444861	70505490	60629	q13.1	
MED12	X	71118556	71142454	23898	q13.1	
NLGN3	X	71144831	71171201	26370	q13.1	
GJB1	X	71215194	71225516	10322	q13.1	
ABCB7	X	75053172	75156732	103560	q13.3	
ATRX	X	77504878	77786269	281391	q21.1	
ATP7A	X	77910656	78050395	139739	q21.1	
BRWD3	X	80670854	80809688	138834	q21.1	FR19
PCDH19	X	100291644	100410273	118629	q22.1	
PLP1	X	103773718	103792619	18901	q22.2	
PRPS1	X	107628424	107651026	22602	q22.3	FR25

(Table S5) contd.....

GENE	Chromosome	Gene Start (bp) hg38	Gene End (bp) hg38	Gene Size (bp)	Band	Fragile region*
ACSL4	X	109624244	109733403	109159	q23	
AGTR2	X	116170722	116174972	4250	q23	FR27
UBE2A	X	119574538	119584418	9880	q24	
UPF3B	X	119834022	119852998	18976	q24	
CUL4B	X	120524609	120575794	51185	q24	
GRIA3	X	123184153	123490915	306762	q25	
ZDHHC9	X	129803288	129843909	40621	q26.1	
PHF6	X	134373253	134428790	55537	q26.2	
SLC9A6	X	135985439	136047264	61825	q26.3	
SOX3	X	140502987	140505503	2516	q27.1	
FMR1	X	147911951	147951125	39174	q27.3	
SLC6A8	X	153688099	153696593	8494	q28	
ABCD1	X	153724868	153744762	19894	q28	FR29
L1CAM	X	153861514	153909223	47709	q28	
MECP2	X	154021573	154097755	76182	q28	
GDI1	X	154436920	154443467	6547	q28	
RAB39B	X	155258241	155264589	6348	q28	FR30

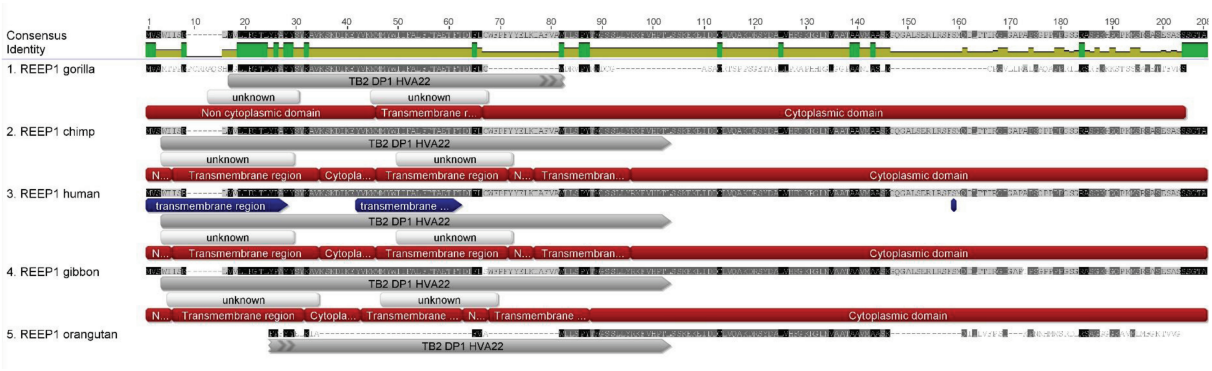

Fig. (S1). Alignment of the REEP1 orthologous genes. Gorilla, chimpanzee, Human, Gibbon and Orangutan REEP1 proteins were aligned using the MUSCLE program. Blue, grey and red bars show the protein domain predicted by InterProscan database (http://www.ebi.ac.uk/interpro/). Green bars show the consensus identity by Geneious software.

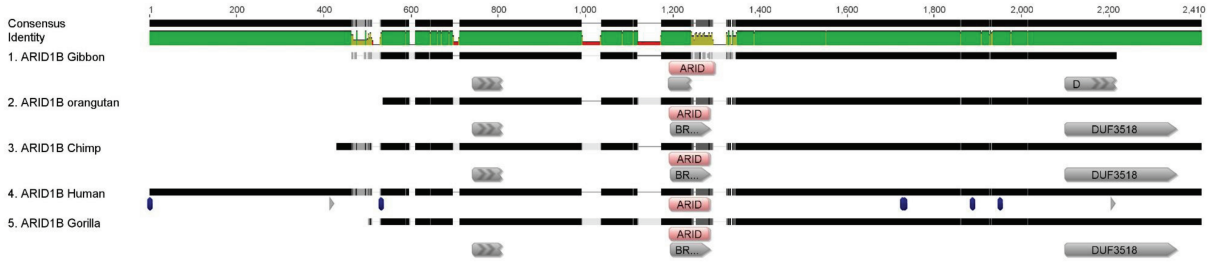

Fig. (S2). Alignment of the REEP1 orthologous genes. Gorilla, chimpanzee, Human, Gibbon and Orangutan REEP1 proteins were aligned using the MUSCLE program. Blue, grey and red bars show the protein domain predicted by InterProscan database (http://www.ebi.ac.uk/interpro/). Green bars show the consensus identity by Geneious software.