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Article

High-Resolution Chromosome Ideogram Representation of Currently Recognized Genes for Autism Spectrum Disorders

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Abstract: Recently, autism-related research has focused on the identification of various genes and disturbed pathways causing the genetically heterogeneous group of autism spectrum disorders (ASD). The list of autism-related genes has significantly increased due to better awareness with advances in genetic technology and expanding searchable genomic databases. We compiled a master list of known and clinically relevant autism spectrum disorder genes identified with supporting evidence from peer-reviewed medical literature sources by searching key words related to autism and genetics and from authoritative autism-related public access websites, such as the Simons Foundation Autism Research Institute autism genomic database dedicated to gene discovery and characterization. Our list consists of 792 genes arranged in alphabetical order in tabular form with gene symbols placed on high-resolution human chromosome ideograms, thereby enabling clinical and laboratory geneticists and genetic counsellors to access convenient visual images of the location and distribution of ASD genes. Meaningful correlations of the observed phenotype in patients with suspected/confirmed ASD gene(s) at the chromosome region or breakpoint band site can be made to inform diagnosis and gene-based personalized care and provide genetic counselling for families.

Keywords: high-resolution chromosome ideograms; autism; genetic evidence; autism spectrum disorders (ASD); ASD genes

1. Introduction

Classical autism or autistic disorder is common, with developmental difficulties noted by three years of age. It belongs to a group of heterogeneous conditions known as autism spectrum disorders (ASDs) with significant impairments in verbal and non-verbal communication and social interactions with restricted repetitive behaviors, specifically in movements and interests [1–3]. Other symptoms include lack of eye contact or focus, sleep disturbances and tactile defensiveness beginning at an early age. Several validated rating scales are used at a young age to help establish the diagnosis, including the autism diagnostic observation schedule (ADOS) and the autism diagnostic interview-revised (ADI-R) supported by pertinent medical history and clinical findings [4–6]. ASD affects about 1% of children in the general U.S. population with a 4:1 male to female ratio, usually without congenital anomalies or growth retardation [7,8].

Autism was first used as a term by Kanner in 1943 when describing a group of children lacking the ability to establish interpersonal contact and communication [9]. About one-fourth of children with autism are diagnosed by 2–3 years of age and show regression of skills in about 30% of cases. About 60% of ASD subjects show intellectual disabilities at a young age [10,11]. When comparing the prevalence of health disorders involving the central nervous system, autism ranks higher than epilepsy (6.5 cases per 1000), brain paralysis or dementia (2.5 cases/1000 for each) and Parkinson disease (two cases per 1000); genetic factors are related to many of these disorders [12,13]. Autism also occurs more commonly than congenital malformations in the general population, but dysmorphic findings are present in about 25% of children with autism. Microcephaly is seen in about 10% of cases, but macrocephaly is documented with larger frontal and smaller occipital lobes in about 20% of children with autism. Those with autism and extreme macrocephaly are at a greater risk to have *PTEN* tumor suppressor gene mutations [14], while another autism-related gene (*CHD8*) can also lead to macrocephaly and autism [15].

Autism is due to a wide range of genetic abnormalities, as well as non-genetic causes, including the environment, environmental and gene interaction (epigenetics) and metabolic disturbances (e.g., mitochondrial dysfunction), with the recurrence risk dependent on the family history and presence or absence of dysmorphic features. Candidate genes for ASD are identified by different means, including cytogenetic abnormalities (*i.e.*, translocations at chromosome breakpoints or deletions (e.g., the 22q11.2 deletion) indicating the location or loss of specific genes) in individuals with ASD along with overlapping linkage and functional data related to the clinical presentation, with certain chromosome regions identified by genetic linkage using DNA markers that co-inherit with the specific phenotype [16,17]. A representative example for such an occurrence is the proto-oncogene (MET) involved in pathways related to neuronal development [18] and found to be linked to the chromosome 7q31 band, where this gene is located. Decreased activity of the gene promoter was recognized when specific single nucleotide polymorphisms (SNPs) were present in this region by linkage studies.

However, genetic linkage studies have received only limited success in the study of the genetics of autism. On the other hand, chromosomal microarray analysis using DNA probes disturbed across the genome can be used to detect chromosomal abnormalities at >100-times smaller than seen in high-resolution chromosome studies. Microarray studies have also become the first tier of genetic testing for this patient population and are recommended for all ASD patients [19]. Greater than 20% of studied patients with microarray analysis are found to have submicroscopic deletions or duplications in the genome containing genes that play a role in causing autism [20,21]. Identification of causative mutations is important to guide treatment selection and to manage medical co-morbidities, such as risks for seizures, developmental regression or for cancer (e.g., the *PTEN* gene).

Routine cytogenetic studies have shown abnormalities of chromosomes 2, 3, 4, 5, 7, 8, 11, 13, 15, 16, 17, 19, 22 and X, including deletions, duplications, translocations and inversions involving specific chromosome regions where known or candidate genes for ASD are located [22]. These studies further support the role of genetic factors in the causation of this common neurodevelopment disorder. Specifically, cytogenetic abnormalities involving the 15q11–q13 region are found in at least 1% of individuals with ASD and include *CYFIP1*, *GABRB3* and *UBE3A* genes in this chromosome region [23] and most recently the 15q11.2 BP1-BP2 microdeletion (Burnside-Butler) syndrome [24]. DNA copy number changes have also shown recurrent small deletions or duplications of the chromosome 16p11.2 band using microarray analysis [25,26] and the chromosome 15q13.2–q13.3 region [27], whereas copy number changes are noted throughout the genome in individuals with ASD, indicating the presence of multiple candidate genes on every human chromosome. These copy number changes are more often of the deletion type.

For idiopathic or non-syndromic autism, the empirical risk for siblings to be similarly affected is between 2% and 8% with an average of 4% [28]. In multiplex families having two or more affected children with autism, the recurrence risk may be as high as 25%, but generally ranges from 13% [29] to 19% [30] if due to single-gene disturbances as the cause, a major focus of this illustrative review. Advances in genetic technology beyond linkage or cytogenetic analysis of affected families with ASD or other complex disorders have led to genome-wide association studies (GWAS) involving hundreds of affected and control individuals by analyzing the distribution and clustering of hundreds and thousands of SNPs that have successfully been searched for candidate genes. The first GWAS for ASD was undertaken by Lauritsen *et al.* in 2006 [31] using 600 DNA markers in an isolated population of affected individuals from the Faroe Islands. They found an association of the chromosome 3p25.3 band, and later, other investigators studied more subjects with larger collections of genotyped markers and found several chromosome bands and regions ascertained when specific SNPs were over-represented in the ASD subjects, including 5p14.1, 5p15 and 16p13–p21 [32–37]. The studies implicated several gene families, including the cadherin family, encoding proteins for neuronal cell adhesion, while other genes (e.g., *SEMA5A*) were implicated in axonal guidance with lower gene expression levels in brain specimens from individuals with ASD [33], reviewed by Holt and Monaco [17]. Since that time, several additional studies searching for clinically relevant and known genes for ASD have identified a new collection of ASD genes [38–53].

The ability to identify an increased number of SNPs with advanced genetic platforms and extensive approaches using bioinformatics have led to improved access and a more thorough analysis. This has led to comparing genotyping data from GWAS and DNA copy number variants (CNVs) with the

identification of structural genetic defects, such as submicroscopic deletions or duplications of the genome, which was not possible a few years ago. Separate studies using array comparative genomic hybridization or microarray analysis to investigate those individuals with ASD continue to yield useful data in identifying candidate genes for ASD in affected individuals [20,21,54]. The yield for microarray analysis is reported to be approximately 20% for identifying deletions or duplications at sites where known or candidate ASD genes are present. The use of more advanced technology, such as next-generation sequencing (whole genome or exome) will yield additional valuable information on the location and description of lesions of genes contributing to ASD with increasing evidence for specific and recurring mutations of single genes involved with neurodevelopment and function, leading to potential therapeutic discoveries and interventions.

Autism is frequent in single-gene conditions, such as fragile X syndrome, tuberous sclerosis, Rett syndrome or neurofibromatosis, but single-gene disorders as a whole account for less than 20% of all cases; therefore, most individuals with ASD are non-syndromic. The heritability of ASD, which takes into consideration the extent of genetic factors contributing to autism, is estimated to be as high as 90% [55]; hence the relevance and continued importance of investigating the role of genetics in the causation of ASD and expanded diagnostic testing to inform and guide treatment for individuals with identifiable genetic disturbances.

A current list of clinically relevant and known candidate genes for ASD is needed for diagnostic testing and genetic counselling purposes in the clinical setting. Historically, a previous list of known or candidate genes showing an association with ASD was reported in 2011 by Holt and Monaco [17] with the placement of 175 genes on chromosome ideograms. A much greater number of validated genes are now recognized as playing a pivotal role in ASD, warranting an updated, revised summary. We will utilize high-resolution chromosome ideograms (850 band level) to plot the location of genes now recognized by searching the literature and website information as playing a documented role in ASD. In tabular form, we will list the individual gene symbol, expanded name or description and chromosome location.

2. Results and Discussion

The diagnostic approach for an individual with ASD should include a clinical genetics evaluation with interviews of parents and health caregivers for the collection and overview of historical problems, a three-generation family pedigree, recording of developmental milestones and description of atypical behaviors along with medical and surgical procedures and a current list of medications and ongoing treatments. Laboratory tests should include lead, thyroid function, lactate and pyruvate levels in order to assess metabolic and mitochondrial functions that may be impacted by an underlying genetic disturbance along with cholesterol and urine collection for organic acid levels. Brain imaging and electroencephalogram patterns should be reviewed, if available. In addition, the ADI-R and ADOS instruments are used to test the diagnosis of ASD.

To further increase the diagnostic yield in individuals with ASD presenting for genetic service, Schaefer *et al.* [19] proposed and utilized a three-tier approach to include a genetic work-up by a clinical geneticist with expertise in dysmorphology to identify known syndromes with or without dysmorphic features (e.g., birth marks), growth anomalies (e.g., microcephaly, macrocephaly and

short stature), viral titers (e.g., rubella) and metabolic screening (urine for organic acids and mucopolysaccharides, plasma lactate and amino acid levels). DNA testing for fragile X syndrome and Rett syndrome in females and males is also available, along with chromosomal and DNA microarrays to examine structural DNA lesions in those with a sporadic form of autism and the use of SNP arrays to examine for regions of homozygosity or uniparental disomy, whereby both members of a chromosome pair come from one parent [56]. Exome sequencing is now available particularly to those affected subjects with a positive family history of autism (multiplex families), if other diagnostic tests are uninformative. *PTEN* gene mutation screening would be indicated in those patients with extreme macrocephaly (head size > 2 SD) [14], if not previously done, and a review of brain MRI results. Serum and urine uric acid levels and assays for adenylate succinase deficiency should be done to include biochemical genetic studies and mitochondrial genome screening and function [57] if the above testing protocols are not diagnostic. Up to one in five children with ASD show findings of mitochondrial dysfunction [57], and a detailed genetic work-up will significantly increase the yield for the diagnosis of ASD, leading to a better understanding of causation, treatment and more accurate genetic counselling for those presenting for genetic services [20,21,54].

Advances made in genetic technology and bioinformatics have led to vastly improved genetic testing options for application in the clinical setting in patients presenting for genetic services [54]. Significant discoveries have been made with the recognition of genetic defects in the causation of ASD using microarray technology and, now, next generation sequencing. This technology has flourished with a combination of DNA probes used for both copy number variation and SNPs being required to identify segmental deletions and duplications in the genome and regions of homozygosity for the determination of identical by descent for the calculation of inbreeding coefficients or consanguinity status along with uniparental disomy of individual chromosomes [56].

Next generation exome DNA sequencing and RNA sequencing allows for discoveries of disease-causing genes and regulatory sequences required for normal function. Identifying and characterizing molecular signatures for novel or disturbed gene or exon expression and disease-specific profiles and patterns with expression heat maps have led to the recognition of interconnected disturbed gene pathways in many diseases, including a growing body of genetic evidence for autism and other psychiatric or aberrant behavioral disorders [54].

The position for each known or candidate gene for ASD susceptibility is plotted on high-resolution chromosome ideograms (850 band level), as shown in Figure 1 below. We have included gene symbols and expanded names along with the chromosome band location in Table 1 for the 792 genes recognized as playing a role in ASD.

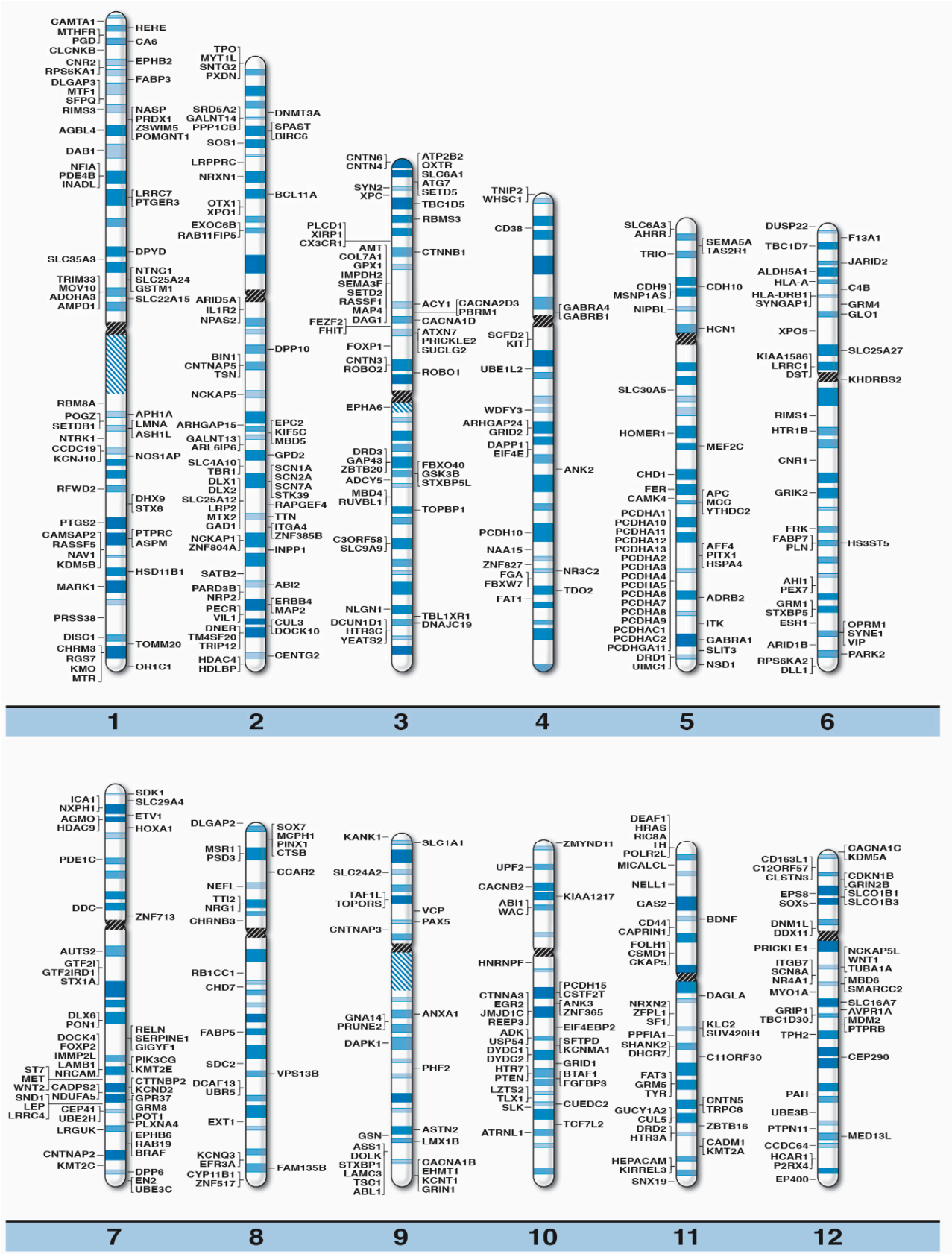

Figure 1. *Cont.*

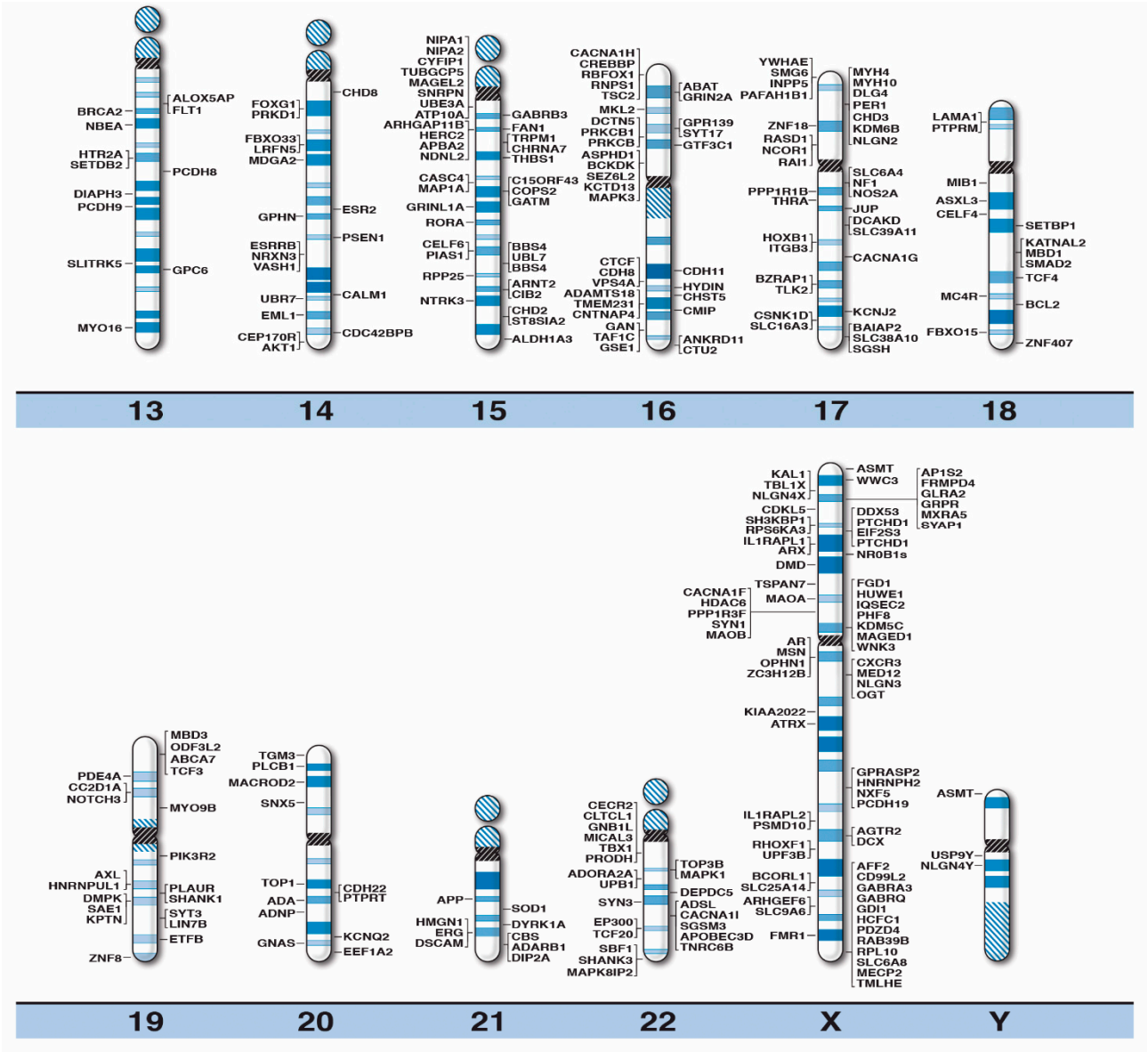

Figure 1. High-resolution human chromosome ideograms (850 band level) with the ASD gene symbol placed at the chromosomal band location. The centromere area, highlighted in black, separates the upper short "p" arm and lower long "q" arm for each chromosome. The gene symbols are arranged in alphabetical order with the expanded name and chromosome band position listed in Table 1.

Table 1. Recognized genes for autism spectrum disorders (ASD) and their chromosome locations.

Gene Symbol	Gene Name	Location
ABAT	4-aminobutyrate aminotransferase	16p13.2
ABCA7	ATP-binding cassette, sub-family A (ABC1), member 7	19p13.3
ABI1	Abl-interactor 1	10p12.1
ABI2	Abl-interactor 2	2q33.2
ABL1	C-Abl oncogene 1, non-receptor tyrosine kinase	9q34.12
ACY1	Aminoacylase 1	3p21.2
ADA	Adenosine deaminase	20q13.12
ADAMTS18	A disintegrin-like and metalloproteinase with thrombospondin type 1 motif, 18	16q23.1
ADARB1	Adenosine deaminase, RNA-specific, B1	21q22.3
ADCY5	Adenylate cyclase 5	3q21.1
ADK	Adenosine kinase	10q22.2
ADNP	Activity-dependent neuroprotector homeobox	20q13.13
ADORA2A	Adenosine A2A receptor	22q11.23
ADORA3	Adenosine A3 receptor	1p13.2
ADRB2	Adrenergic, β 2 receptor	5q32
ADSL	Adenylosuccinate lyase	22q13.1
AFF2	AF4/fragile X mental retardation 2 (FMR2) family, member 2	Xq28
AFF4	AF4/fragile X mental retardation 2 (FMR2) family, member 4	5q31.1
AGBL4	ATP/GTP binding protein-like 4	1p33
AGMO	Alkylglycerol monooxygenase	7p21.1
AGTR2	Angiotensin II receptor, type 2	Xq23
AHI1	Abelson helper integration site 1	6q23.3
AHRR	Aryl hydrocarbon receptor repressor	5p15.33
AKT1	v-Akt murine thymoma viral oncogene homolog 1	14q32.33
ALDH1A3	Aldehyde dehydrogenase 1 family, member A3	15q26.3
ALDH5A1	Aldehyde dehydrogenase 5 family, member A1	6p22.3
ALOX5AP	Arachidonate 5-lipoxygenase-activating protein	13q12.3
AMPD1	Adenosine monophosphate deaminase 1	1p13.2
AMT	Aminomethyltransferase	3p21.31
ANK2	Ankyrin 2	4q25
ANK3	Ankyrin 3	10q21.2
ANKRD11	Ankyrin repeat domain 11	16q24.3
ANXA1	Annexin A1	9q21.13
AP1S2	Adaptor-related protein complex 1, sigma 2 subunit	Xp22.2
APBA2	Amyloid β precursor protein-binding, family A, member 2	15q13.1
APC	Adenomatosis polyposis coli	5q22.2
APH1A	APH1A γ secretase subunit	1q21.2
APOBEC3D	Apolipoprotein B mRNA editing enzyme, catalytic polypeptide-like 3D	22q13.1
APP	Amyloid β precursor protein	21q21.3
AR	Androgen receptor	Xq12
ARHGAP11B	Rho GTPase activating protein 11B	15q13.2
ARHGAP15	Rho GTPase activating protein 15	2q22.2

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
ARHGAP24	Rho GTPase activating protein 24	4q22.1
ARHGEF6	RAC/CDC42 guanine nucleotide exchange factor (GEF) 6	Xq26.3
ARID1B	AT rich interactive domain 1B (SWI1-like)	6q25.3
ARID5A	AT rich interactive domain 5A (MRF1-like)	2q11.2
ARL6IP6	ADP-ribosylation-like factor 6 interacting protein 6	2q23.3
ARNT2	Aryl-hydrocarbon receptor nuclear translocator 2	15q25.1
ARX	Aristaless related homeobox	Xp21.3
ASH1L	Ash1 (absent, small, or homeotic)-like (Drosophila)	1q22
ASMT	Acetylserotonin *O*-methyltransferase, X-chromosomal	Xp22.33
ASMT	Acetylserotonin *O*-methyltransferase, Y-chromosomal	Yp11.32
ASPHD1	Aspartate β-hydroxylase domain containing 1	16p11.2
ASPM	Asp (abnormal spindle) homolog, microcephaly associated	1q31.3
ASS1	Argininosuccinate synthetase	9q34.1
ASTN2	Astrotactin 2	9q33.1
ASXL3	Additional sex combs-like 3	18q12.1
ATG7	Autophagy related 7	3p25.3
ATP10A	ATPase, Class V, type 10A	15q11.2
ATP2B2	ATPase, Ca++ transporting, plasma membrane 2	3p25.3
ATRNL1	Attractin-like 1	10q25.3
ATRX	α thalassemia/mental retardation syndrome X-linked	Xq21.1
ATXN7	Ataxin 7	3p14.1
AUTS2	Autism susceptibility candidate 2	7q11.22
AVPR1A	Arginine vasopressin receptor 1A	12q14.2
AXL	AXL receptor tyrosine kinase	19q13.2
BAIAP2	BAI1-associated protein 2	17q25.3
BBS4	Bardet-Biedl syndrome 4	15q24.1
BCKDK	Branched chain ketoacid dehydrogenase kinase	16p11.2
BCL11A	B-Cell CLL/lymphoma 11A (zinc finger protein)	2p16.1
BCL2	B-cell CLL/lymphoma 2	18q21.33
BCORL1	Bc16 co-repressor-like 1	Xq26.1
BDNF	Brain-derived neurotrophic factor	11p14.1
BIN1	Bridging integrator 1	2q14.3
BIRC6	Baculoviral IAP repeat containing 6	2p22.3
BRAF	v-Raf murine sarcoma viral oncogene homolog B	7q34
BRCA2	Breast cancer 2, early onset	13q13.1
BTAF1	RNA polymerase II, B-TFIID transcription factor-associated, 170 kDa (Mot1 homolog, *S. cerevisiae*)	10q23.32
BZRAP1	Benzodiazepine receptor (peripheral) associated protein 1	17q23.2
C11ORF30	Chromosome 11 open reading frame 30	11q13.5
C12ORF57	Chromosome 12 open reading frame 57	12p13.31
C15ORF43	Chromosome 15 open reading frame 43	15q21.1
C3ORF58	Chromosome 3 open reading frame 58	3q24
C4B	Complement component 4B	6p21.33

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
CA6	Carbonic anhydrase VI	1p36.2
CACNA1B	Calcium channel, voltage-dependent, N type, α 1B subunit	9q34.3
CACNA1C	Calcium channel, voltage-dependent, L type, α 1C subunit	12p13.33
CACNA1D	Calcium channel, voltage-dependent, L type, α 1D subunit	3p14.3
CACNA1F	Calcium channel, voltage-dependent, α 1F subunit	Xp11.23
CACNA1G	Calcium channel, voltage-dependent, T type, α 1G subunit	17q21.33
CACNA1H	Calcium channel, voltage-dependent, α 1H subunit	16p13.3
CACNA1I	Calcium channel, voltage-dependent, T type, α 1I subunit	22q13.1
CACNA2D3	Calcium channel, voltage-dependent, α 2/δ subunit 3	3p21.1
CACNB2	Calcium channel, voltage-dependent, β 2 subunit	10p12.33
CADM1	Cell adhesion molecule 1	11q23.3
CADPS2	Ca++-dependent activator protein for secretion 2	7q31.32
CALM1	Calmodulin 1 (phosphorylase kinase, δ)	14q32.11
CAMK4	Calcium/calmodulin-dependent protein kinase	5q22.1
CAMSAP2	Calmodulin regulated spectrin-associated protein family, member 2	1q32.1
CAMTA1	Calmodulin binding transcription activator 1	1p36.31
CAPRIN1	Cell cycle associated protein 1	11p13
CASC4	Cancer susceptibility candidate 4	15q15.3
CBS	Cystathionine β-synthase	21q22.3
CCAR2	Cell cycle and apoptosis regulator 2	8p21.3
CC2D1A	Coiled-coil and C2 domain-containing 1A	19p13.12
CCDC19	Coiled-coil domain-containing protein 19	1q23.2
CCDC64	Coiled-coil domain-containing 64	12q24.23
CD38	CD38 molecule	4p15.32
CD44	CD44 molecule	11p13
CD163L1	CD163 molecule-like 1	12p13.31
CD99L2	CD99 molecule-like 2	Xq28
CDC42BPB	CDC42 binding protein kinase β (DMPK-like)	14q32.32
CDH10	Cadherin 10, type 2	5p14.2
CDH22	Cadherin-like 22	20q13.1
CDH8	Cadherin 8, type 2	16q22.1
CDH9	Cadherin 9, type 2	5p14.1
CDH11	Cadherin 11, type 2	16q21
CDKL5	Cyclin-dependent kinase-like 5	Xp22.13
CDKN1B	Cyclin-dependent kinase inhibitor 1B	12p13.1
CECR2	Cat eye syndrome chromosome region, candidate 2	22q11.21
CELF4	CUGBP, Elav-like family, member 4	18q12.2
CELF6	CUGBP, Elav-like family, member 6	15q23
CENTG2	Centaurin γ-2	2q37.2
CEP170R	Centrosomal protein 170B	14q32.33
CEP290	Centrosomal protein 290 kDa	12q21.32
CEP41	Centrosomal protein 41 kDa	7q32.2
CHD1	Chromodomain helicase DNA binding protein 1	5q21.1

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
CHD2	Chromodomain helicase DNA binding protein 2	15q26.1
CHD3	Chromodomain helicase DNA binding protein 3	17p13.1
CHD7	Chromodomain helicase DNA binding protein 7	8q12.2
CHD8	Chromodomain helicase DNA binding protein 8	14q11.2
CHRM3	Cholinergic receptor, muscarinic 3	1q43
CHRNA7	Cholinergic receptor, neuronal nicotinic, α 7	15q13.3
CHRNB3	Cholinergic receptor, neuronal nicotinic, β 3	8p11.21
CHST5	Carbohydrate sulfotransferase 5	16q22.3
CIB2	Calcium and integrin binding family member 2	15q25.1
CKAP5	Cytoskeleton associated protein 5	11p11.2
CLCNKB	Chloride channel voltage-sensitive kidney, B	1p36.13
CLSTN3	Calsyntenin 3	12p13.31
CLTCL1	Clathrin, heavy chain-like 1	22q11.21
CMIP	c-MAF inducing protein	16q23.2
CNR1	Cannabinoid receptor 1	6q15
CNR2	Cannabinoid receptor 2	1p36.11
CNTN3	Contactin 3	3p12.3
CNTN4	Contactin 4	3p26.3
CNTN5	Contactin 5	11q22.1
CNTN6	Contactin 6	3p26.3
CNTNAP2	Contactin associated protein-like 2	7q35
CNTNAP3	Contactin associated protein-like 3	9p13.1
CNTNAP4	Contactin associated protein-like 4	16q23.1
CNTNAP5	Contactin associated protein-like 5	2q14.3
COL7A1	Collagen, type VII, α 1	3p21.31
COPS2	Thyroid hormone receptor interactor 15	15q21.1
CREBBP	CREB binding protein	16p13.3
CSMD1	Cytoskeleton associated protein 5	11p11.2
CSNK1D	Casein kinase 1, δ	17q25
CSTF2T	Cleavage stimulation factor, 3' pre-RNA, subunit 2, 64 kDa, tau	10q21.1
CTCF	CCCTC-binding factor	16q22.1
CTNNA3	Catenin (cadherin-associated protein), α 3	10q21.3
CTNNB1	Catenin (cadherin-associated protein), β 1, 88 kDa	3p22.1
CTSB	Cathepsin B	8p23.1
CTTNBP2	Cortactin binding protein 2	7q31.31
CTU2	Cytosolic thiouridylase subunit 2 homolog (S. pombe)	16q24.3
CUEDC2	CUE domain containing 2	10q24.32
CUL5	Cullin 5	11q22.3
CUL3	Cullin 3	2q36.2
CX3CR1	Chemokine (C-X3-C motif) receptor 1	3p22.2
CXCR3	Chemokine, CXC motif, receptor 3	Xq13.1
CYFIP1	Cytoplasmic FMRP interacting protein 1	15q11.2
CYP11B1	Cytochrome P450, subfamily XIB, polypeptide 1	8q24.3

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
DAB1	Disabled homolog 1	1p32.2
DAG1	Dystroglycan 1 (dystrophin-associated glycoprotein 1)	3p21.31
DAGLA	Diacylglycerol lipase, α	11q12.2
DAPK1	Death-associated protein kinase 1	9q21.33
DAPP1	Dual adaptor of phosphotyrosine and 3-phosphoinositides 1	4q23
DCAF13	DDB1 and CUL4 associated factor 13	8q22.3
DCAKD	Dephospho-CoA kinase domain-containing protein	17q21.31
DCTN5	Dynactin 5	16p12.2
DCUN1D1	DCN1, domain containing protein 1	3q27.1
DCX	Doublecortin	Xq23
DDC	DOPA decarboxylase	7p12.1
DDX11	DEAD (Asp-Glu-Ala-Asp)/H box 11	12p11.21
DDX53	DEAD (Asp-Glu-Ala-Asp) box polypeptide 53	Xp22.11
DEAF1	DEAF1 transcription factor	11p15.5
DEPDC5	DEP domain containing 3 protein 5	22q12.2
DHCR7	7-dehydrocholesterol reductase	11q13.4
DHX9	DEAH (Asp-Glu-Ala-His) box helicase 9	1q25.3
DIAPH3	Diaphanous, Drosophila, homolog 3	13q21.2
DIP2A	DIP2 disco-interacting protein 2 homolog A (Drosophila)	21q22.3
DISC1	Disrupted in schizophrenia 1	1q42.2
DLG4	Discs, large, Drosophila, homolog 4	17p13.1
DLGAP2	Discs, large- associated protein 2	8p23.3
DLGAP3	Discs, large- associated protein 3	1p34.3
DLL1	δ-like 1 (Drosophila)	6q27
DLX1	Distal-less homeobox 1	2q31.1
DLX2	Distal-less homeobox 2	2q31.1
DLX6	Distal-less homeobox 6	7q21.3
DMD	Dystrophin	Xp21.1
DMPK	Dystrophia myotonica-protein kinase	19q13.32
DNAJC19	DNAJ Hsp40 homolog, subfamily C, member 19	3q26.33
DNER	δ- and notch-like epidermal growth factor-related receptor	2q36.3
DNM1L	Dynamin 1-like	12p11.21
DNMT3A	DNA (cytosine-5)-methyltransferase 3 α	2p23.3
DOCK4	Dedicator of cytokinesis 4	7q31.1
DOCK10	Dedicator of cytokinesis 10	2q36.2
DOLK	Dolichol kinase	9q34.1
DPP10	Dipeptidyl peptidase 10	2q14.1
DPP6	Dipeptidyl peptidase 6	7q36.2
DPYD	Dihydropyrimidine dehydrogenase	1p21.3
DRD1	Dopamine receptor D1	5q35.2
DRD2	Dopamine receptor D2	11q23.2
DRD3	Dopamine receptor D3	3q13.31
DSCAM	Down syndrome cell adhesion molecule	21q22.2

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
DST	Dystonin	6p12.1
DUSP22	Dual specificity phosphatase 22	6p25.3
DYDC1	DPY30 domain containing 1	10q23.1
DYDC2	DPY30 domain containing 2	10q23.1
DYRK1A	Dual-specificity tyrosine-phosphorylation-regulated kinase 1A	21q22.13
EEF1A2	Eukaryotic translation elongation factor 1 α 2	20q13.33
EFR3A	EFR3 homolog A (*S. cerevisiae*)	8q24.22
EGR2	Early growth response 2	10q21.3
EHMT1	Euchromatic histone methyltransferase 1	9q34.3
EIF2S3	Eukaryotic translation initiation factor 2, subunit 3 γ	Xp22.11
EIF4E	Eukaryotic translation initiation factor 4E	4q23
EIF4EBP2	Eukaryotic translation initiation factor 4E binding protein 2	10q22.1
EML1	Echinoderm microtubule associated protein like 1	14q32.2
EN2	Engrailed 2	7q36.3
EP300	E1A binding protein p300	22q13.2
EP400	E1A binding protein p400	12q24.33
EPC2	Enhancer of polycomb, Drosophila homolog of 2	2q23.1
EPHA6	Ephrin receptor A6	3q11.2
EPHB2	Ephrin receptor B2	1p36.12
EPHB6	Ephrin receptor B6	7q34
EPS8	Epidermal growth factor receptor pathway substrate 8	12p12.3
ERBB4	v-ERB-A avian erythroblastic leukemia viral oncogene homolog 4	2q34
ERG	v-ETS avian erythroblastosis virus E26 oncogene homolog	21q22.2
ESR1	Estrogen receptor 1	6q25.1
ESR2	Estrogen receptor 2	14q23.2
ESRRB	Estrogen-related receptor β	14q24.3
ETFB	Electron-transfer-flavoprotein, β polypeptide	19q13.41
ETV1	Ets variant 1	7p21.2
EXOC6B	Exocyst complex component 6B	2p13.2
EXT1	Exostosin 1	8q24.11
F13A1	Factor XIII, A1 subunit	6p25.1
FABP3	Fatty acid binding protein 3, muscle and heart (mammary-derived growth inhibitor)	1p35.2
FABP5	Fatty acid binding protein 5	8q21.13
FABP7	Fatty acid binding protein 7	6q22.31
FAM135B	Family with sequence similarity 135, member B	8q24.23
FAN1	FANCD2/FANCI-associated nuclease 1	15q13.2
FAT1	FAT tumor suppressor, Drosophila homolog of, 1	4q35.2
FAT3	FAT tumor suppressor, Drosophila homolog of , 3	11q14.3
FBXO15	F-box protein 15	18q22.3
FBXO33	F-box protein 33	14q21.1
FBXO40	F-box protein 40	3q13.33
FBXW7	F-box and WD repeat domain containing 7, E3 ubiquitin protein	4q31.3
FER	FPS/FES related tyrosine kinase	5q21.3

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
FEZF2	FEZ family zinc finger 2	3p14.2
FGA	Fibrinogen, A α polypeptide	4q31.3
FGD1	FYVE, Rho GEF and PH domain containing 1	Xp11.22
FGFBP3	Fibroblast growth factor binding protein 3	10q23.32
FHIT	Fragile histidine triad	3p14.2
FLT1	c-FMS-related tyrosine kinase 1	13q12.3
FMR1	Fragile X mental retardation 1 (FMR1)	Xq27.3
FOLH1	Folate hydrolase 1	11p11.2
FOXG1	Forkhead box G1	14q12
FOXP1	Forkhead box P1	3p13
FOXP2	Forkhead box P2	7q31.1
FRK	FYN-related kinase	6q22.1
FRMPD4	FERM and PDZ domain containing protein 4	Xp22.2
GABRA1	γ-aminobutyric acid A receptor, α 1	5q34
GABRA3	γ-aminobutyric acid receptor, α 3	Xq28
GABRA4	γ-aminobutyric acid receptor, α 4	4p12
GABRB1	γ-aminobutyric acid receptor, β 1	4p12
GABRB3	γ-aminobutyric acid receptor, β 3	15q12
GABRQ	γ-aminobutyric acid receptor, θ	Xq28
GAD1	Glutamate decarboxylase 1 (brain, 67 kDa)	2q31.1
GALNT13	UDP-*N*-acetyl-α-D-galactosamine:polypeptide *N*-acetylgalactosaminyl-transferase 13	2q23.3
GALNT14	UDP-*N*-acetyl-α-D-galactosamine:polypeptide *N*-acetylgalactosaminyl-transferase 14	2p23.1
GAN	Gigaxonin	16q24.1
GAP43	Growth associated protein 43	3q13.31
GAS2	Growth arrest-specific 2	11p14.3
GATM	Glycine amidinotransferase (L-arginine:glycine amidinotransferase)	15q21.1
GDI1	GDP dissociation inhibitor 1	Xq28
GIGYF1	GRB10 interacting GYF protein 1	7q22.1
GLO1	Glyoxalase I	6p21.2
GLRA2	Glycine receptor, α 2 subunit	Xp22.2
GNA14	Guanine nucleotide-binding protein, α 14	9q21.2
GNAS	Guanine nucleotide-binding protein, α-stimulating activity polypeptide I complex locus	20q13.32
GNB1L	Guanine nucleotide-binding protein, β 1-like	22q11.21
GPC6	Glypican 6	13q31.3
GPD2	Glycerol-3-phosphate dehydrogenase 2	2q24.1
GPHN	Gephyrin	14q23.3
GPR139	G protein-coupled receptor 139	16p12.3
GPR37	G protein-coupled receptor 37	7q31.33
GPRASP2	G protein-coupled receptor associated sorting protein 2	Xq22.1
GPX1	Glutathione peroxidase 1	3p21.31

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
GRID1	Glutamate receptor, ionotropic, δ 1	10q23.2
GRID2	Glutamate receptor, ionotropic, δ 2	4q22.1
GRIK2	Glutamate receptor, ionotropic, kainate 2	6q16.3
GRIN1	Glutamate receptor, ionotropic, *N*-methyl D-aspartate 1	9q34.3
GRIN2A	Glutamate receptor, ionotropic, *N*-methyl D-aspartate 2A	16p13.2
GRIN2B	Glutamate receptor, ionotropic, *N*-methyl D-aspartate 2B	12p13.1
GRINL1A	GRINL1A complex locus 1	15q21.3
GRIP1	Glutamate receptor interacting protein 1	12q14.3
GRM1	Glutamate receptor, metabotropic 1	6q24.3
GRM4	Glutamate receptor, metabotropic 4	6p21.31
GRM5	Glutamate receptor, metabotropic 5	11q14.3
GRM8	Glutamate receptor, metabotropic 8	7q31.33
GRPR	Gastrin-releasing peptide receptor	Xp22.2
GSE1	Gse1 coiled-coil protein	16q24.1
GSK3B	Glycogen synthase kinase 3 β	3q13.33
GSN	Gelsolin	9q33.2
GSTM1	Glutathione *S*-transferase M1	1p13.3
GTF2I	General transcription factor III	7q11.23
GTF2IRD1	GTF2I repeat domain containing 1	7q11.23
GTF3C1	General transcription factor IIIC, polypeptide 1, α	16p12.1
GUCY1A2	Guanylate cyclase 1, soluble, α 2	11q22.3
HCAR1	Hydroxycarboxylic acid receptor 1/G protein-coupled receptor 81	12q24.31
HCFC1	Host cell factor C1	Xq28
HCN1	Hyperpolarization activated cyclic nucleotide-gated potassium channel 1	5p12
HDAC4	Histone deacetylase 4	2q37.3
HDAC6	Histone deacetylase 6	Xp11.23
HDAC9	Histone deacetylase 9	7p21.1
HDLBP	High density lipoprotein binding protein	2q37.3
HEPACAM	Hepatic and glial cell adhesion molecule	11q24.2
HERC2	HECT domain and RCC1-like domain 2	15q13.1
HLA-A	Major histocompatibility complex, class I, A	6p22.1
HLA-DRB1	Major histocompatibility complex, class II, DR β 1	6p21.32
HMGN1	High mobility group nucleosome binding domain 1	21q22.2
HNRNPF	Heterogeneous nuclear ribonucleoprotein F	10q11.21
HNRNPH2	Heterogeneous nuclear ribonucleoprotein H2	Xq22.1
HNRNPUL1	Heterogeneous nuclear ribonucleoprotein U-like 1	19q13.2
HOMER1	Homer, Drosophila, homolog 1 of 1	5q14.1
HOXA1	Homeobox A1	7p15.3
HOXB1	Homeobox B1	17q21.32
HRAS	v-HA-RAS Harvey rat sarcoma viral oncogene homolog	11p15.5
HS3ST5	Heparan sulfate 3-*O*-sulfotransferase 5	6q22.31
HSD11B1	11-β-hydroxysteroid dehydrogenase type 1	1q32.2
HSPA4	Heat shock 70 kDa protein 4	5q31.1

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
HTR1B	5-hydroxytryptamine receptor 1B	6q14.1
HTR2A	5-hydroxytryptamine receptor 2A	13q14.2
HTR3A	5-hydroxytryptamine receptor 3A	11q23.2
HTR3C	5-hydroxytryptamine receptor 3, family member C	3q27.1
HTR7	5-hydroxytryptamine receptor 7	10q23.31
HUWE1	HECT, UBA and WWE domain containing 1, E3 ubiquitin protein ligase	Xp11.22
HYDIN	Hydrocephalus-inducing, mouse, homolog of	16q22.2
ICA1	Islet cell autoantigen 1	7p21.3
IL1R2	Interleukin 1 receptor, type II	2q11.2
IL1RAPL1	Interleukin 1 receptor accessory protein-like 1	Xp21.3
IL1RAPL2	Interleukin 1 receptor accessory protein-like 2	Xq22.3
IMMP2L	Inner mitochondrial membrane peptidase, subunit 2, *S. cerevisiae*, homolog of	7q31.1
IMPDH2	Inosine-5-prime monophosphate dehydrogenase 2	3p21.31
INADL	Inactivation no after-potential D-like	1p31.3
INPP1	Inositol polyphosphate-1-phosphatase	2q32.2
INPP5	Inositol polyphosphate-5-phosphatase	17p13.3
IQSEC2	IQ motif and Sec7 domain 2	Xp11.22
ITGA4	Integrin, α 4	2q31.3
ITGB3	Integrin, β 3	17q21.32
ITGB7	Integrin, β 7	12q13.13
ITK	IL20 inducible t-cell kinase	5q33.3
JARID2	Jumonji, AT rich interactive domain 2	6p22.3
JMJD1C	Jumonji domain containing 1C	10q21.3
JUP	Junction plakoglobin	17q21.2
KAL1	Kallmann syndrome interval 1	Xp22.31
KANK1	KN motif and ankyrin repeat domains 1	9p24.3
KATNAL2	Katanin p60 subunit A-like 2	18q21.1
KCND2	Potassium voltage-gated channel, Shal-related subfamily, member 2	7q31.31
KCNJ2	Potassium inwardly-rectifying channel, subfamily J, member 2	17q24.3
KCNJ10	Potassium inwardly-rectifying channel, subfamily J, member 10	1q23.2
KCNMA1	Potassium large conductance calcium-activated channel, subfamily M, α member 1	10q22.3
KCNQ2	Potassium voltage-gated channel, KQT-like subfamily, member 2	20q13.3
KCNQ3	Potassium voltage-gated channel, KQT-like subfamily, member 3	8q24.22
KCNT1	Potassium channel, subfamily T, member 1	9q34.3
KCTD13	Potassium channel tetramerization domain containing protein 13	16p11.2
KDM5A	Lysine (K)-specific demethylase 5A	12p13.33
KDM5B	Lysine (K)-specific demethylase 5B	1q32.1
KDM5C	Lysine (K)-specific demethylase 5C	Xp11.22
KDM6B	Lysine (K)-specific demethylase 6B	17p13.1
KHDRBS2	KH domain containing, RNA binding, signal transduction associated protein 2	6q11.1
KIAA1217	Sickle tail protein homolog	10p12.31

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
KIAA1586	KIAA1586	6p12.1
KIAA2022	KIAA2022	Xq13.3
KIF5C	Kinesin family member 5C	2q23.1
KIRREL3	Kin of IRRE like 3	11q24.2
KIT	v-KIT Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog	4q12
KLC2	Kinesin light chain 2	11q13.2
KMO	Kynurenine 3-monooxygenase	1q43
KMT2A	Lysine (K)-specific methyltransferase 2A	11q23.3
KMT2C	Lysine (K)-specific methyltransferase 2C	7q36.1
KMT2E	Lysine (K)-specific methyltransferase 2E	7q22.3
KPTN	Kaptin (actin binding protein)	19q13.32
LAMA1	Laminin, α 1	18p11.23
LAMB1	Laminin, β 1	7q31.1
LAMC3	Laminin, γ 3	9q34.1
LEP	Leptin	7q32.1
LIN7B	Lin-7 homolog B (*C. elegans*)	19q13.33
LMNA	Lamin A/C	1q22
LMX1B	LIM homeobox transcription factor 1, β	9q33.3
LRFN5	Leucine-rich repeats and fibronectin type III domain containing 5	14q21.1
LRGUK	Leucine-rich repeats and guanylate kinase domain containing	7q33
LRP2	Low density lipoprotein receptor-related protein 2	2q31.1
LRPPRC	Leucine-rich PPR motif containing protein	2p21
LRRC1	Leucine-rich repeat-containing protein 1	6p12.1
LRRC4	Leucine-rich repeat-containing protein 4	7q32.1
LRRC7	Leucine-rich repeat-containing protein 7	1p31.1
LZTS2	Leucine zipper, putative tumor suppressor 2	10q24.31
MACROD2	Macro domain containing 2	20p12.1
MAGED1	Melanoma antigen family D, 1	Xp11.22
MAGEL2	MAGE-like 2	15q11.2
MAOA	Monoamine oxidase A	Xp11.3
MAOB	Monoamine oxidase B	Xp11.23
MAP1A	Microtubule-associated protein 1A	15q15.3
MAP2	Microtubule-associated protein (MAP) 2	2q34
MAP4	Microtubule-associated protein (MAP) 4	3p21.31
MAPK1	Mitogen-activated protein kinase 1	22q11.22
MAPK3	Mitogen-activated protein kinase 3	16p11.2
MAPK8IP2	Mitogen-activated protein kinase 8 interacting protein 2	22q13.33
MARK1	MAP/microtubule affinity-regulating kinase 1	1q41
MBD1	Methyl-CpG binding domain protein 1	18q21.1
MBD3	Methyl-CpG binding domain protein 3	19p13.3
MBD4	Methyl-CpG binding domain protein 4	3q21.3
MBD5	Methyl-CpG binding domain protein 5	2q23.1
MBD6	Methyl-CpG binding domain protein 6	12q13.2

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
MC4R	Melanocortin 4 receptor	18q21.32
MCC	Mutated in colorectal cancers	5q22.2
MCPH1	Microcephalin 1	8p23.1
MDGA2	Mephrin, A5 antigen, protein tyrosine phosphatase mu (MAM) domain containing glycosylphosphatidylinositol anchor 2	14q21.3
MDM2	MDM2 oncogene, E3 ubiquitin protein ligase	12q15
MECP2	Methyl CpG binding protein 2	Xq28
MED12	Mediator complex subunit 12	Xq13.1
MED13L	Mediator complex subunit 13-like	12q24.21
MEF2C	MADS box transcription myocyte enhancer factor 2, polypeptide C	5q14.3
MET	Met proto-oncogene	7q31.2
MIB1	Mind bomb E3 ubiquitin protein ligase 1	18q11.2
MICAL3	Microtubule-associated monooxygenase, calponin and lim domains-containing, 3	22q11.21
MICALCL	MICAL *C*-terminus-like protein	11p15.3
MKL2	Myocardin-like 2	16p13.12
MOV10	Moloney leukemia virus 10, mouse, homolog of	1p13.2
MSN	Moesin	Xq12
MSNP1AS	Moesin pseudogene 1 antisense	5p14.1
MSR1	Macrophage scavenger receptor	8p22
MTF1	Metal-regulatory transcription factor 1	1p34.3
MTHFR	5-10-methylene-tetrahydrofolate reductase	1p36.22
MTR	5-methyltetrahydrofolate-homocysteine *S*-methyltransferase	1q43
MTX2	Metaxin 2	2q31.1
MXRA5	Matrix-remodelling associated 5	Xp22.2
MYH4	Myosin, heavy chain 4, skeletal muscle	17p13.1
MYH10	Myosin, heavy chain 10, non-muscle	17p13.1
MYO16	Myosin XVI	13q33.3
MYO1A	Myosin IA	12q13.3
MYO9B	Myosin IXB	19p13.11
MYT1L	Myelin transcription factor 1-like	2p25.3
NAA15	*N*(α)-acetyltransferase 15, NatA auxiliary subunit	4q31.1
NASP	Nuclear autoantigenic sperm protein (histone-binding)	1p34.1
NAV1	Neuron navigator 1	1q32.1
NBEA	Neurobeachin	13q13.3
NCKAP1	NCK-associated protein 1	2q32.1
NCKAP5	NCK-associated protein 5	2q21.2
NCKAP5L	NCK-associated protein 5-like	12q13.12
NCOR1	Nuclear receptor corepressor 1	17p11.2
NDNL2	Necdin-like gene 2	15q13.1
NDUFA5	NADH-ubiquinone oxidoreductase 1 α subcomplex, 5	7q31.32
NEFL	Neurofilament protein, light polypeptide	8p21.2
NELL1	NEL-like 1	11p15.1
NF1	Neurofibromin 1	17q11.2

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
NFIA	Nuclear factor I/A	1p31.3
NIPA1	Non imprinted gene in Prader-Willi/Angelman syndrome chromosomal region 1	15q11.2
NIPA2	Non imprinted gene in Prader-Willi/Angelman syndrome chromosomal region 2	15q11.2
NIPBL	Nipped-B-like	5p13.2
NLGN1	Neuroligin 1	3q26.31
NLGN2	Neuroligin 2	17p13.1
NLGN3	Neuroligin 3	Xq13.1
NLGN4X	Neuroligin 4, X-linked	Xp22.31
NLGN4Y	Neuroligin 4, Y-linked	Yq11.221
NOS1AP	Nitric oxide synthase 1 (neuronal) adaptor protein	1q23.3
NOS2A	Nitric oxide synthase 2A	17q11.2
NOTCH3	Notch 3	19p13.12
NPAS2	Neuronal PAS domain protein 2	2q11.2
NR0B1	Nuclear receptor subfamily 0, group B, member 1	Xp21.2
NR3C2	Nuclear receptor subfamily 3, group C, member 2	4q31.23
NR4A1	Nuclear receptor subfamily 4, group A, member 1	12q13.13
NRCAM	Neuronal cell adhesion molecule	7q31.1
NRG1	Neuregulin 1	8p12
NRP2	Neuropilin 2	2q33.3
NRXN1	Neurexin I	2p16.3
NRXN2	Neurexin II	11q13.1
NRXN3	Neurexin III	14q24.3
NSD1	Nuclear receptor-binding Sa-var, enhancer of zeste, and trithorax domain protein 1	5q35.3
NTNG1	Netrin G1	1p13.3
NTRK1	Neurotrophic tyrosine kinase, receptor, type 1	1q23.1
NTRK3	Neurotrophic tyrosine kinase, receptor, type 3	15q25.3
NXF5	Nuclear RNA export factor 5	Xq22.1
NXPH1	Neurexophilin 1	7p21.3
ODF3L2	Outer dense fiber of sperm tails 3-like 2	19p13.3
OGT	O-linked *N*-acetylglucosamine transferase	Xq13.1
OPHN1	Oligophrenin 1	Xq12
OPRM1	Opioid receptor, mu 1	6q25.2
OR1C1	Olfactory receptor, family 1, subfamily C, member 1	1q44
OTX1	Orthodenticle Drosophila, homolog of	2p15
OXTR	Oxytocin receptor	3p25.3
P2RX4	Purinergic receptor P2X, ligand-gated ion channel, 4	12q24.31
PAFAH1B1	Platelet-activating factor acetylhydrolase 1B, regulatory subunit 1	17p13.3
PAH	Phenylalanine hydroxylase	12q23.2
PARD3B	PAR-3 family cell polarity regulator β	2q33.3
PARK2	Parkin	6q26
PAX5	Paired box 5	9p13.2
PBRM1	Polybromo 1	3p21.1
PCDH10	Protocadherin 10	4q28.3

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
PCDH15	Protocadherin 15	10q21.1
PCDH19	Protocadherin 19	Xq22.1
PCDH8	Protocadherin 8	13q14.3
PCDH9	Protocadherin 9	13q21.32
PCDHA1	Protocadherin α 1	5q31.3
PCDHA10	Protocadherin α 10	5q31.3
PCDHA11	Protocadherin α 11	5q31.3
PCDHA12	Protocadherin α 12	5q31.3
PCDHA13	Protocadherin α 13	5q31.3
PCDHA2	Protocadherin α 2	5q31.3
PCDHA3	Protocadherin α 3	5q31.3
PCDHA4	Protocadherin α 4	5q31.3
PCDHA5	Protocadherin α 5	5q31.3
PCDHA6	Protocadherin α 6	5q31.3
PCDHA7	Protocadherin α 7	5q31.3
PCDHA8	Protocadherin α 8	5q31.3
PCDHA9	Protocadherin α 9	5q31.3
PCDHAC1	Protocadherin α subfamily C, member 1	5q31.3
PCDHAC2	Protocadherin α subfamily C, member 2	5q31.3
PCDHGA11	Protocadherin γ subfamily A, member 11	5q31.3
PDE1C	Phosphodiesterase 1C	7p14.3
PDE4A	Phosphodiesterase 4A, cAMP-specific	19p13.2
PDE4B	Phosphodiesterase 4B, cAMP-specific	1p31.3
PDZD4	PDZ domain containing 4	Xq28
PECR	Peroxisomal trans-2-enoyl-CoA reductase	2q35
PER1	Period, Drosophila, homolog of	17p13.1
PEX7	Peroxisomal biogenesis factor 7	6q23.3
PGD	Phosphogluconate dehydrogenase	1p36.22
PHF2	PHD finger protein 2	9q22.31
PHF8	PHD finger protein 8	Xp11.22
PIAS1	Protein inhibitor of activated STAT, 1	15q23
PIK3CG	Phosphatidylinositol-3-kinase, catalytic, γ	7q22.3
PIK3R2	Phosphatidylinositol-3-kinase, regulatory subunit 2	19q13.11
PINX1	PIN2 interacting protein 1	8p23.1
PITX1	Paired-like homeodomain transcription factor 1	5q31.1
PLAUR	Plasminogen activator receptor, urokinase-type	19q13.31
PLCB1	Phospholipase C, β 1	20p12.3
PLCD1	Phospholipase C, δ 1	3p22.2
PLN	Phospholamban	6q22.31
PLXNA4	Plexin A4	7q32.3
POGZ	POGO transposable element with ZNF domain	1q21.3
POLR2L	Polymerase (RNA) II (DNA directed) polypeptide L, 7.6 kDa	11p15.5
POMGNT1	Protein *O*-mannose β-1, 2-*N*-acetylglucosaminyl-transferase	1p34.1

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
PON1	Paraoxonase 1	7q21.3
POT1	Protection of telomeres 1	7q31.33
PPFIA1	Protein tyrosine phosphatase, receptor type, F polypeptide, interacting protein, α 1	11q13.3
PPP1CB	Protein phosphatase 1, catalytic subunit, β isozyme	2p23.2
PPP1R1B	Protein phosphatase 1, regulatory (inhibitor) subunit 1B	17q12
PPP1R3F	Protein phosphatase 1, regulatory (inhibitor) subunit 3F	Xp11.23
PRODH	Proline dehydrogenase (oxidase) 1	22q11.21
PRICKLE1	Prickle, Drosophila, homolog of, 1	12q12
PRICKLE2	Prickle, Drosophila, homolog of, 2	3p14.1
PRKCB	Protein kinase C, β	16p12.2
PRKCB1	Protein kinase C, β-1	16p12.2
PRKD1	Protein kinase D1	14q12
PRDX1	Peroxiredoxin 1	1p34.1
PRSS38	Protease, serine, 38	1q42.13
PRUNE2	Prune, Drosophila, homolog of, 2	9q21.2
PSD3	Pleckstrin and Sec7 domains-containing protein 3	8p22
PSEN1	Presenilin 1	14q24.2
PSMD10	Proteasome 26S subunit, non-ATPase, 10	Xq22.3
PTCHD1	Patched domain containing protein 1	Xp22.11
PTEN	Phosphatase and tensin homolog	10q23.31
PTGER3	Prostaglandin E receptor 3, EP3 subtype	1p31.1
PTGS2	Prostaglandin-endoperoxide synthase 2	1q31.1
PTPN11	Protein tyrosine phosphatase, non-receptor type 11	12q24.13
PTPRB	Protein tyrosine phosphatase, receptor type, B	12q15
PTPRC	Protein tyrosine phosphatase, receptor type, C	1q31.3
PTPRM	Protein tyrosine phosphatase, receptor type, M	18p11.23
PTPRT	Protein tyrosine phosphatase, receptor type, T	20q13.11
PXDN	Peroxidasin, Drosophila homolog of	2p25.3
RAB11FIP5	RAB11 family-interacting protein 5	2p13.2
RAB19	RAB19, member RAS oncogene family	7q34
RAB39B	RAS-associated protein RAB39B	Xq28
RAI1	Retinoic acid induced gene 1	17p11.2
RAPGEF4	Rap guanine nucleotide exchange factor	2q31.1
RASD1	RAS protein, dexamethasone-induced, 1	17p11.2
RASSF1	RAS association (ralGDS/AF-6) domain family member 1	3p21.31
RASSF5	RAS association domain family protein 5	1q32.1
RB1CC1	RB1-inducible coiled-coil 1	8q11.23
RBFOX1	RNA binding protein FOX-1, *C. elegans*, homolog of, 1	16p13.3
RBM8A	RNA binding motif protein 8A	1q21.1
RBMS3	RNA binding motif protein, single stranded interacting, 3	3p24.1
REEP3	Receptor expression-enhancing protein 3	10q21.3

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
RELN	Reelin	7q22.1
RERE	RE-repeats encoding gene	1p36.23
RFWD2	Ring finger and WD repeat domains-containing protein 2	1q25.2
RGS7	Regulator of G protein signaling 7	1q43
RHOXF1	RHOX homeobox family, member 1	Xq24
RIC8A	RIC8 guanine nucleotide exchange factor A	11p15.5
RIMS1	Regulating synaptic membrane exocytosis 1	6q13
RIMS3	Protein regulating synaptic membrane exocytosis 3	1p34.2
RNPS1	RNA binding protein S1	16p13.3
ROBO1	Roundabout, Drosophila, homolog of, 1	3p12.2
ROBO2	Roundabout, Drosophila, homolog of, 2	3p12.3
RORA	RAR-related orphan receptor A	15q22.2
RPL10	Ribosomal protein L10	Xq28
RPP25	Ribonuclease P/MRP 25 kDa subunit	15q24.2
RPS6KA1	Ribosomal protein S6 kinase, 90 kDa, polypeptide 1	1p36.11
RPS6KA2	Ribosomal protein S6 kinase, 90 kDa, polypeptide 2	6q27
RPS6KA3	Ribosomal protein S6 kinase, 90 kDa, polypeptide 3	Xp22.12
RUVBL1	RuvB-E. coli, homolog-like 1	3q21.3
SAE1	SUMO1 activating enzyme, subunit 1	19q13.32
SATB2	Special AT-rich sequence-binding protein 2	2q33.1
SBF1	SET binding factor 1	22q13.33
SCFD2	Sec1 family domain containing 2	4q12
SCN1A	Sodium channel, neuronal, type I, α subunit	2q24.3
SCN2A	Sodium channel, voltage-gated, type II, α subunit	2q24.3
SCN7A	Sodium channel, voltage-gated, type VII, α subunit	2q24.3
SCN8A	Sodium channel, voltage-gated, type VIII, α subunit	12q13.13
SDC2	Syndecan 2	8q22.1
SDK1	Sidekick cell adhesion molecule 1	7p22.2
SEMA3F	Sema domain, immunoglobulin domain (Ig), short basic domain, secreted, (semaphorin) 3F	3p21.31
SEMA5A	Semaphorin 5A	5p15.31
SERPINE1	Serpin peptidase inhibitor, clade E (nexin, plasminogen activator inhibitor type 1), member 1	7q22.1
SETBP1	SET binding protein 1	18q12.3
SETD2	SET domain containing protein 2	3p21.31
SETD5	SET domain containing protein 5	3p25.3
SETDB1	SET domain, bifurcated, 1	1q21.3
SETDB2	SET domain, bifurcated, 2	13q14.2
SEZ6L2	Seizure related 6 homolog (mouse)-like 2	16p11.2
SF1	Splicing factor 1	11q13.1
SFPQ	Splicing factor proline/glutamine-rich	1p34.3
SFTPD	Surfactant, pulmonary-associated protein D	10q22.3

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
SGSH	*N*-sulfoglucosamine sulfohydrolase	17q25.3
SGSM3	Small G protein signaling modulator 3	22q13.1
SH3KBP1	SH3-domain kinase binding protein 1	Xp22.12
SHANK1	SH3 and multiple ankyrin repeat domains 1	19q13.3
SHANK2	SH3 and multiple ankyrin repeat domains 2	11q13.4
SHANK3	SH3 and multiple ankyrin repeat domains 3	22q13.33
SLC16A3	Solute carrier family 16 (monocarboxylic acid transporter), member 3	17q25
SLC16A7	Solute carrier family 16 (monocarboxylic acid transporter), member 7	12q14.1
SLC1A1	Solute carrier family 1 (neuronal/epithelial high affinity glutamate transporter), member 1	9p24.2
SLC22A15	Solute carrier family 22, (organic cation transporter), member 15	1p13.1
SLC24A2	Solute carrier family 24 (sodium/potassium/calcium exchanger), member 2	9p22.1
SLC25A12	Solute carrier family 25 (mitochondrial carrier, Aralar), member 12	2q31.1
SLC25A14	Solute carrier family 25 (mitochondrial carrier, brain), member 14	Xq26.1
SLC25A24	Solute carrier family 25 (mitochondrial carrier, phosphate carrier), member 24	1p13.3
SLC25A27	Solute carrier family 25, member 27	6p12.3
SLC29A4	Solute carrier family 29 (equilibrative nucleoside transporter), member 4	7p22.1
SLC30A5	Solute carrier family 30 (zinc transporter), member 5	5q13.1
SLC35A3	Solute carrier family 35 (UDP-*N*-acetylglucosamine transporter), member 3	1p21.2
SLC38A10	Solute carrier family 38, member 10	17q25.3
SLC39A11	Solute carrier family 39 (metal ion transporter), member 11	17q21.31
SLC4A10	Solute carrier family 4 (sodium bicarbonate transporter-like), member 10	2q24.2
SLC6A1	Solute carrier family 6 (neurotransmitter transporter), member 1	3p25.3
SLC6A3	Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	5p15.33
SLC6A4	Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	17q11.2
SLC6A8	Solute carrier family 6 (neurotransmitter transporter, creatine), member 8	Xq28
SLC9A6	Solute carrier family 9 (sodium/hydrogen exchanger), member 6	Xq26.3
SLC9A9	Solute carrier family 9 (sodium/hydrogen exchanger), member 9	3q24
SLCO1B1	Solute carrier organic anion transporter family, member 1B1	12p12.2
SLCO1B3	Solute carrier organic anion transporter family, member 1B3	12p12.2
SLIT3	Slit, Drosophila, homolog of, 3	5q35.1
SLITRK5	SLIT and NTRK-like family, member 5	13q31.2
SLK	STE20-like kinase	10q24.33
SMAD2	SMAD family member 2	18q21.1
SMARCC2	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily C, member 2	12q13.2
SMG6	SMG 6, *C. elegans*, homolog of	17p13.3
SND1	EBNA2 coactivator p100	7q32.1
SNRPN	Small nuclear ribonucleoprotein polypeptide N	15q11.2
SNTG2	Syntrophin, γ 2	2p25.3
SNX19	Sorting nexin 19	11q25
SNX5	Sorting nexin 5	20p11.23

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
SOD1	Superoxide dismutase 1, soluble	21q22.11
SOS1	Son of sevenless (SOS), Drosophila, homolog 1	2p22.1
SOX5	SRY (sex determining region Y)-box 5	12p12.1
SOX7	SRY (sex determining region Y)-box 7	8p23.1
SPAST	Spastin	2p22.3
SRD5A2	Steroid-5-α-reductase, 2	2p23.1
ST7	Suppressor of tumorigenicity 7	7q31.2
ST8SIA2	ST8 α-*N*-acetyl-neuraminide α-2,8-sialyltransferase 2	15q26.1
STK39	Serine/threonine protein kinase 39	2q24.3
STX6	Syntaxin 6	1q25.3
STX1A	Syntaxin 1A	7q11.23
STXBP1	Syntaxin-binding protein 1	9q34.1
STXBP5	Syntaxin-binding protein 5	6q24.3
STXBP5L	Syntaxin-binding protein 5-like	3q13.33
SUCLG2	Succinate-CoA ligase, GDP-forming, β subunit	3p14.1
SUV420H1	Suppressor of variegation 4–20, Drosophila, homolog of, 1	11q13.2
SYAP1	Synapse associated protein 1	Xp22.2
SYN1	Synapsin 1	Xp11.23
SYN2	Synapsin II	3p25.2
SYN3	Synapsin III	22q12.3
SYNE1	Spectrin repeat containing nuclear envelope 1	6q25.2
SYNGAP1	Synaptic RAS-GTPase-activating protein 1	6p21.32
SYT17	Synaptotagmin XVII	16p12.3
SYT3	Synaptotagmin III	19q13.33
TAF1C	TATA box-binding protein-associated factor 1C	16q24.1
TAF1L	TATA box-binding protein-associated factor 1-like	9p21.1
TAS2R1	Taste receptor, type 2, member 1	5p15.31
TBC1D30	TBC1 domain family, member 30	12q14.3
TBC1D5	TBC1 domain family, member 5	3p24.3
TBC1D7	TBC1 domain family, member 7	6p24
TBL1X	Transducin-β-like 1, X-linked	Xp22.31
TBL1XR1	Transducin-β-like 1 receptor 1	3q26.32
TBR1	T-box, brain, 1	2q24.2
TBX1	T-box 1	22q11.21
TCF3	Transcription factor 3	19p13.3
TCF4	Transcription factor 4	18q21.2
TCF20	Transcription factor 20 (AR1)	22q13.2
TCF7L2	Transcription factor 7-like 2 (t-cell specific, HMG-box)	10q25.2
TDO2	Tryptophan 2,3-dioxygenase	4q32.1
TGM3	Transglutaminase 3	20p13
TH	Tyrosine hydroxylase	11p15.5
THBS1	Thrombospondin 1	15q14

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
THRA	Thyroid hormone receptor, α-1	17q21.1
TLK2	Tousled-like kinase 2	17q23.2
TLX1	T-cell leukemia homeobox 1	10q24.31
TM4SF20	Transmembrane 4 L6 family, member 20	2q36.3
TMEM231	Transmembrane protein 231	16q23.1
TMLHE	Epsilon-trimethyllysine hydroxylase	Xq28
TNIP2	TNFAIP3 interacting protein 2	4p16.3
TNRC6B	Trinucleotide repeat containing 6B	22q13.1
TOMM20	MAS20P, *S. cerevisiae*, homolog of	1q42.3
TOP1	Topoisomerase, DNA, I	20q12
TOP3B	Topoisomerase, DNA, III, β	22q11.22
TOPBP1	Topoisomerase (DNA) II-binding protein 1	3q22.1
TOPORS	Topoisomerase I-binding, arginine/serine-rich, E3 ubiquitin protein ligase	9p21.1
TPH2	Tryptophan hydroxylase 2	12q21.1
TPO	Thyroid peroxidase	2p25.3
TRIM33	Tripartite motif containing protein 33	1p13.2
TRIO	Trio Rho guanine nucleotide exchange factor	5p15.2
TRIP12	Thyroid hormone receptor interactor 12	2q36.3
TRPC6	Transient receptor potential cation channel, subfamily C, member 6	11q22.1
TRPM1	Transient receptor potential cation channel, subfamily M, member 1	15q13.3
TSC1	Tuberous sclerosis 1	9q34.1
TSC2	Tuberous sclerosis 2	16p13.3
TSN	Translin	2q14.3
TSPAN7	Tetraspanin 7	Xp11.4
TTI2	TELO2-interacting protein 2	8p12
TTN	Titin	2q31.2
TUBA1A	Tubulin, α-1A	12q13.12
TUBGCP5	Tubulin-γ complex-associated protein 5	15q11.2
TYR	Tyrosinase	11q14.3
UBE1L2	Ubiquitin-activating enzyme, E1-like 2	4q13.2
UBE2H	Ubiquitin-conjugating enzyme E2H	7q32.2
UBE3A	Ubiquitin protein ligase E3A	15q11.2
UBE3B	Ubiquitin protein ligase E3B	12q24.11
UBE3C	Ubiquitin protein ligase E3C	7q36.3
UBL7	Ubiquitin-like 7	15q24.1
UBR5	Ubiquitin protein ligase E3 component *N*-recognin 5	8q22.3
UBR7	Ubiquitin protein ligase E3 component *N*-recognin 7	14q32.12
UIMC1	Ubiquitin interaction motif containing 1	5q35.2
UPB1	Ureidopropionase, β 1	22q11.23
UPF2	UPF2, yeast, homolog of	10p14
UPF3B	UPF3, yeast, homolog of, B	Xq24

Table 1. *Cont.*

Gene Symbol	Gene Name	Location
USP54	Ubiquitin specific peptidase 54	10q22.2
USP9Y	Ubiquitin specific protease 9, Y-chromosome	Yq11.21
VASH1	Vasohibin 1	14q24.3
VCP	Valosin containing protein	9p13.3
VIL1	Villin 1	2q35
VIP	Vasoactive intestinal peptide (VIP)	6q25.2
VPS13B	Vacuolar protein sorting 13, yeast, homolog of, B	8q22.2
VPS4A	Vacuolar protein sorting 4 homolog A (*S. cerevisiae*)	16q22.1
WAC	WW domain containing adaptor with coiled-coil	10p12.1
WDFY3	WD repeat and FYVE domain containing 3	4q21.23
WHSC1	Wolf-Hirschhorn syndrome candidate 1	4p16.3
WNK3	Protein kinase lysine deficient 3	Xp11.22
WNT1	Wingless-type MMTV integration site family, member 1	12q13.12
WNT2	Wingless-type MMTV integration site family, member 2	7q31.2
WWC3	WWC family member 3	Xp22.32
XIRP1	Cardiomyopathy-associated protein 1	3p22.2
XPC	Xeroderma pigmentosum complementation group C	3p25.1
XPO1	Exportin 1	2p15
XPO5	Exportin 5	6p21.1
YEATS2	YEATS domain containing 2	3q27.1
YTHDC2	YTH domain containing 2	5q22.2
YWHAE	Tyrosine 3-monooxygenase, tryptophan 5-monooxygenase activation protein, epsilon isoform	17p13.3
ZBTB16	Zinc finger- and BTB domain-containing protein 16	11q23.1
ZBTB20	Zinc finger- and BTB domain-containing protein 20	3q13.31
ZC3H12B	Zinc finger CCCH domain-containing protein 12B	Xq12
ZFPL1	Zinc finger protein-like 1	11q13.1
ZMYND11	Zinc finger, MYND-type containing 11	10p15.3
ZNF18	Zinc finger protein 18	17p12
ZNF365	Zinc finger protein 365	10q21.2
ZNF385B	Zinc finger protein 385B	2q31.3
ZNF407	Zinc finger protein 407	18q23
ZNF517	Zinc finger protein 517	8q24.3
ZNF8	Zinc finger protein 8	19q13.43
ZNF713	Zinc finger protein 713	7p11.2
ZNF804A	Zinc finger protein 804A	2q32.1
ZNF827	Zinc finger protein 827	4q31.22
ZSWIM5	Zinc finger, SWIM-type containing 5	1p34.1

3. Experimental Section

We used computer-based internet websites and PubMed (https://www.ncbi.nlm.nih.gov/pubmed) to search key words for genetics and autism. This included the integrated catalogue of human genetic studies related to autism found at the Simons Foundation Autism Research Initiative (SFARI) website

(https://gene.sfari.org), which currently lists 667 genes reported as of 25 February 2015. This public access initiative is an ongoing curated collection of clinically proven ASD genes supported by clinical and autism experts, medical geneticists and laboratory specialists in the study of autism. This site includes gene description and evidence of support for causation with cited literature reports. We examined peer-reviewed articles found in the medical literature following our search for genetic evidence (*i.e.*, gene variants, mutations or disturbed gene function) and the involvement of genetics playing a role in autism. Sources included whole-genome sequencing of ASD families randomly selected with at least one unaffected sibling [40] or gene expression profiles in ASD [39] along with other informative websites (e.g., Online Mendelian Inheritance in Man, www.OMIM.org). We then compiled the list of genes from these major sources for a total of 792 genes, whereby at least one mechanism was involved for each gene that could lead to ASD, a heterogeneous condition involving many genes; as our report is focused on the compilation of ASD genes from peer-reviewed research articles and authoritative computer website genomic databases for autism and not necessarily related to causal relationships between the individual gene and ASD. Those genes recognized, to date, as playing a role in ASD susceptibility and causation generally appear to impact chromatin remodeling, metabolism, mRNA translation, cell adhesion and synaptic function [39].

SFARI is a publicly available manually curated web-based searchable site of human genes with links to ASD and includes genes in catalogue form based on five categories—genetic association, syndromic, rare single-gene variant and functional and multi-genetic copy number variation—supported by cited research publications for each. Additional literature sources in our study consisted of both primary research articles and reviews summarizing genetic evidence. Many of the listed genes were identified in multiple research studies and widely reported in literature reviews, data repositories and/or computer genomic-based websites for autism (e.g., SFARI). A large number of genes showed a varied relationship to autism and neurodevelopment, but the mass of the literature surveyed limits the reliability of our relative strength estimates for the ASD and gene associations. The gene would be included if cited and recognized in peer-reviewed publications (e.g., PubMed) with supportive genetic evidence (e.g., genetic linkage, GWAS, functional gene expression patterns, informative SNPs, CNVs or identified gene mutations). Other supporting genetic evidence can be found at Simons Foundation Autism Research Initiative (SFARI) at https://sfari.org/sfari-initiatives/simons-simplex-collection, the National Institutes of Health (NIH) at https://www.ncbi.nlm.nih.gov/gap, the Online Inheritance in Man (OMIM) at www.omim.org or Genecards at https://www.genecards.org.

4. Conclusions

Readily available tissue sources, such as peripheral blood, established lymphoblastoid cell lines and saliva, hold promise for more advances in ASD by enabling the identification of new genes and a better understanding of the causation and disease mechanisms to further stimulate research with the hope to discover new treatment modalities impacted by the recognition of known disease-causing or candidate genes for ASD. We illustrated the master list of clinically relevant and known ASD genes in our summary by plotting individual genes on high-resolution chromosome ideograms and generated a tabular form to increase the awareness required for genetic testing and counselling purposes for family members presenting for genetic services. Creating a master list of genes related to ASD is a complicated

process; new genes are continually identified, but not all genes are equally important or certain to be causative. Additional research is needed to further investigate the causal relationships between the specific gene and ASD. The authors encourage the use of this collection of known and clinically relevant candidate genes for ASD in their evaluation of patients and families presenting for genetic testing options and for accurate genetic counselling.

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Author Contributions

Merlin G. Butler conceived of the study, reviewed data from ASD gene literature reports and wrote the manuscript; Syed K. Rafi obtained and reviewed articles pertaining to ASD genes and summarized the master gene list; and Ann M. Manzardo contributed to gene data review and interpretation, contributed to the content of the manuscript and reviewed the literature.

Conflicts of Interest

The authors declare no conflict of interest.

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