Genes Involved in X-Linked Intellectual Disability by Order of Discovery (revised January 2020)

Year	Gene Name	Gene Symbol	Clinical Description	Function	How Discovered
1983	HYPOXANTHINE GUANINE PHOSPHORIBOSYLTRANSFERASE 1	*HPRT*	Lesch-Nyhan	Enzyme	Met-Fu
1983	PHOSPHOGLYCERATE KINASE 1	*PGK1*	Phosphoglycerokinase Deficiency	Enzyme	Met-Fu
1985	PROTEOLIPID PROTEIN 1	*PLP1*	Pelizaeus-Merzbacher, Spastic Paraplegia 2	Myelination	Mol-Fu
1986	ORNITHINE CARBAMOYLTRANSFERASE	*OTC*	Ornithine Transcarbamoylase Deficiency	Enzyme	Met-Fu
1987	DYSTROPHIN	*DMD*	Duchenne Muscular Dystrophy, IDX85	Structure of skeletal muscle membrane	Chr-rea
1989	PYRUVATE DEHYDROGENASE, ALPHA-1	*PDHA1*	Pyruvate Dehydrogenase Deficiency	Enzyme	Met-Fu
1990	IDURONATE 2-SULFATASE	*IDS*	Hunter	Lysosomal enzyme	Met-Fu
1991	FRAGILE X MENTAL RETARDATION 1	*FMR1*	Fragile X	RNA-binding protein, gene regulation	Chr-rea, L-can
1992	L1 CELL ADHESION MOLECULE	*L1CAM*	Hydrocephaly-MASA, Spastic Paraplegia 1, XL-ACC	Neuronal migration, cell adhesion	L-can
1992	NDP GENE	*NDP*	Norrie	Neuroectodermal cell interaction	Chr-rea
1992	OCRL GENE	*OCRL1*	Lowe	Enzyme	Chr-rea
1993	ATP-BINDING CASSETTE, SUBFAMILY D, MEMBER 1	*ABCD1 (ALDP)*	Adrenoleukodystrophy	Peroxisomal transport protein	L-can
1993	ATPase, Cu(2+)-TRANSPORTING, ALPHA POLYPEPTIDE	*ATP7A*	Menkes, Occipital Horn	Copper transport	Chr-rea
1993	MONOAMINE OXIDASE A	*MAOA* *	Monoamine Oxidase A Deficiency	Enzyme	L-can
1995	ATR-X GENE; X-LINKED NUCLEAR PROTEIN GENE, HELICASE 2, X-LINKED	*ATRX (XNP, XH2)*	Alpha-Thalassemia Intellectual Disability, Carpenter-Waziri, Chudley-Lowry, Holmes-Gang, XLID-Hypotonic Facies, XLID-Spastic Paraplegia, XLID-Arch Fingerprints-Hypotonia	Transcription factor, helicase activities	L-can
1996	AF4/FMR2 FAMILY, MEMBER 2; FRAGILE SITE, FOLIC ACID TYPE, RARE, FRA(X)(q28) E, INCLUDED; FRAXE, INCLUDED	*AFF2 (FMR2)*	Fragile XE	Unknown	Chr-rea
1996	FYVE, RhoGEF, AND PH DOMAIN-CONTAINING PROTEIN 1; FACIOGENITAL DYSPLASIA	*FGD1 (FGDY)*	Aarskog-Scott	Guanine nucleotide exchange factor	Chr-rea
1996	GLYCEROL KINASE	*GK*	Glycerol Kinase Deficiency	Metabolism, glycerol uptake	Met-Fu
1996	GLYPICAN 3	*GPC3*	Simpson-Golabi-Behmel	Cell adhesion, motility	L-can
1996	MYOTUBULARIN	*MTM1*	Myotubular Myopathy	Tyrosine phosphatise	L-can
1996	TRANSLOCASE OF INNER MITOCHONDRIAL MEMBRANE 8, YEAST, HOMOLOG OF, A; DEAFNESS/DYSTONIA PEPTIDE	*TIMM8A (DDP)*	Mohr-Tranebjaerg, Jensen	Transcription factor	L-can
1997	RAB GDP-DISSOCIATION INHIBITOR 1	*GDI1*	IDX41, IDX48	Stabilizes GDP bound conformations	L-can
1997	MIDLINE 1	*MID1*	Telecanthus-Hypospadias, Opitz G/BBB	Zinc finger gene	L-can, Chr-rea
1997	RIBOSOMAL PROTEIN S6 KINASE, 90-KD, 3; RIBOSOMAL S6 KINASE 2;	*RPS6KA3 (RSK2)*	Coffin-Lowry, IDX19	Kinase signaling pathway	L-can
1998	CALCIUM/CALMODULIN-DEPENDENT SERINE PROTEIN KINASE	*CASK*	XLID-Nystagmus-Seizures	Synaptic functions	Mol-Fu
1998	DOUBLECORTIN	*DCX*	Lissencephaly, X-linked	Neuronal migration	Chr-rea (del)
1998	DYSKERIN	*DKC1*	Dyskeratosis Congenita	Cell cycle and nucleolar functions	L-can
1998	FILAMIN A	*FLNA (FLN1)*	Periventricular Heterotopias, OPD I, OPD II	Actin-binding protein	L-can
1998	OLIGOPHRENIN 1	*OPHN1*	IDX60	GTPase activating protein	L-can
1998	p21 PROTEIN-ACTIVATED KINASE 3	*PAK3*	IDX30, IDX47	Rac/Cdc 42 effector	Chr-rea
1999	INTERLEUKIN 1 RECEPTOR ACCESSORY PROTEIN-LIKE 1	*IL1RAPL1*	IDX10, IDX21, IDX34	Unknown	Chr-rea
2000	RHO GUANINE NUCLEOTIDE EXCHANGE FACTOR 6	*ARHGEF6 * (a-PIX)*	IDX46	Effector of the rho GTPases	Chr-rea
2000	LYSOSOME-ASSOCIATED MEMBRANE PROTEIN 2	*LAMP2*	Danon Cardiomyopathy	Membrane, lysosome	L-can

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2000	NF-KAPPA-B ESSENTIAL MODULATOR	*NEMO (IKBKG)*	Incontinentia Pigmenti	Activates the transcription factor NF-KB	L-can
2000	TRANSMEMBRANE 4 SUPERFAMILY, MEMBER 2	*TM4SF2*	IDX58	Interacts with integrins	Chr-rea
2001	METHYL-CpG-BINDING PROTEIN 2	*MECP2*	Rett, IDX16, IDX64, IDX79	Binds methylated CpGs	L-can
2001	OFD1 GENE; OROFACIODIGITAL SYNDROME 1	*OFD1*	Oral-Facial-Digital I	Unknown	L-can
2001	SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER, CREATINE), MEMBER 8	*SLC6A8*	XLID with Seizures	Creatine transporter	Met-Fu
2002	FATTY ACID CoA LIGASE, LONG CHAIN 4	*ACSL4 (FACL4)*	IDX63, IDX68	Fatty acid CoA ligase 4	Chr-rea (del)
2002	ANGIOTENSIN II RECEPTOR, TYPE 2	*AGTR2* *	X-linked Optic Atrophy, IDX88	Angiotensin II receptor	Chr-rea
2002	ARISTALESS-RELATED HOMEOBOX, X-LINKED	*ARX*	Hydranencephaly, Partington, Proud, West, Lissencephaly and Abnormal Genitalia, X-linked, IDX29, IDX32, IDX33, IDX36, IDX43, IDX54, IDX76	Neuronal migration	Chr-rea (del)
2002	CYCLIN-DEPENDENT KINASE-LIKE 5; SERINE/THREONINE PROTEIN KINASE 9	*CDKL5 (STK9)*	Rett-Like Seizures-Hypotonia	Unknown	Chr-rea
2002	KRUPPEL-LIKE FACTOR 8	*KLF8 * (ZNF741)*	IDX		Chr-rea
2002	PHD FINGER PROTEIN 6	*PHF6*	Börjeson-Forssman-Lehmann	Unknown	L-can
2002	SRY-BOX 3	*SOX3*	XLID-Growth Hormone Deficiency	Pituitary function, transcription factor	Chr-rea, L-can
2003	IMMUNOGLOBULIN-BINDING PROTEIN 1	*IGBP1 *	Graham Coloboma		L-can
2003	NHS GENE; NANCE-HORAN SYNDROME GENE	*NHS*	Nance-Horan	-	L-can
2003	NEUROLIGIN 3	*NLGN3 *	Autism	Cell adhesion	L-can
2003	NEUROLIGIN 4	*NLGN4*	Autism	Cell adhesion	L-can
2003	POLYGLUTAMINE-BINDING PROTEIN 1	*PQBP1*	Renpenning, Sutherland-Haan, Hamel Cerebro-Palato-Cardiac, Golabi-Ito-Hall, Porteous, IDX2, IDX55	Polyglutamine binding, regulates transcription	L-can
2003	SPERMINE SYNTHASE	*SMS*	Snyder-Robinson	Synthesis of spermine	L-can
2003	ZINC FINGER PROTEIN 41	*ZNF41 *	IDX89	Zinger finger	Chr-rea
2003	ZINC FINGER PROTEIN 81	*ZNF81 *	IDX45	Zinc finger	Chr-rea
2004	RHO GUANINE NUCLEOTIDE EXCHANGE FACTOR 9	*ARHGEF9*	XLID-Hypotonia-Seizures, XLID-Macrocephaly Macroorchidism	Regulation of Rho protein signal transduction	Chr-rea
2004	ATPase, H+ TRANSPORTING, LYSOSOMAL, ACCESSORY PROTEIN 2; RENIN RECEPTOR	*ATP6AP2 * (ATP6A8-9)*	XLID-Infantile Epilepsy, gait disturbance, Parkinsonian manifestations	Renin receptor, transmembrane protein essential for ATPase involved in lysosomal function	L-can
2004	BCL6 COREPRESSOR	*BCOR*	Lenz Microphthalmia (1 type)	Histone/protein deacetylation	L-can
2004	DISCS LARGE, DROSOPHILA, HOMOLOG OF, 3; NEUROENDOCRINE DLG	*DLG3*	IDX8, IDX90	NMDA-receptor, mediated signaling, synaptic plasticity	X seq
2004	FTSJ HOMOLOG 1	*FTSJ1*	IDX9, IDX44	Methylase	L-can
2004	LYSINE-SPECIFIC DEMETHYLASE 5C; JUMONJI, AT-RICH INTERACTIVE DOMAIN 1C	*KDM5C (JARID1C, SMX)*	IDX13	Regulates transcription, chromatin remodelling	L-can
2004	NEURITE EXTENSION AND MIGRATION FACTOR; KIAA2022 PROTEIN	*KIAA2022 * (NEXMIF)*	Cantagrel Spastic Paraplegia	DNA synthesis, DNA polymerase activity	Chr-rea
2004	PHD FINGER PROTEIN 8	*PHF8*	XLID-Cleft Lip-Cleft Palate	Regulates transcription, binds DNA	L-can
2004	SHROOM FAMILY MEMBER 4; K1AA1202 PROTEIN	*SHROOM4 * (KIAA1202)*	Stoccos dos Santos	Roles in cellular architecture, neurulation, and ion channel function	Chr-rea
2004	SOLUTE CARRIER FAMILY 16 (MONOCARBOXYLIC ACID TRANSPORTER), MEMBER 2	*SLC16A2 (MCT8)*	Allan-Herndon-Dudley, IDX22	T3 receptor	L-can
2004	SYNAPSIN I	*SYN1*	Epilepsy-Macrocephaly, IDX50	Synaptic vesicle protein	L-can

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2005	ZINC FINGER DHHC DOMAIN-CONTAINING PROTEIN 15	*ZDHHC15* *	IDX91		Chr-rea
2006	ADAPTOR-RELATED PROTEIN COMPLEX 1, SIGMA-2 SUBUNIT	*AP1S2*	Turner XLID, Hydrocephaly-Basal Ganglia Calcification, IDX59	Assembly of endocytic vesicles	X-seq
2006	FANCB GENE; FANCONI ANEMIA-ASSOCIATED POLYPEPTIDE, 95-KD	*FANCB*	VACTERL-Hydrocephaly	DNA repair	Mol-Fu
2006	HOLOCYTOCHROME C SYNTHASE	*HCCS*	MIDAS	Energy production, cytochrome homolyase	Chr-rea (del)
2006	STRUCTURAL MAINTENANCE OF CHROMOSOMES 1A	*SMC1A/ SMC1L1*	Cornelia de Lange, X-linked	Cell cycle, mitotic spindle organization and biogenesis, chromosome segregation	Mol-Fu
2006	SUSHI REPEAT-CONTAINING PROTEIN, X-LINKED, 2	*SRPX2* *	XLID-Rolandic Seizures	Signal transduction, growth factor 2	Mol-Fu
2006	UBIQUITIN-CONJUGATING ENZYME E2A	*UBE2A*	XLID-Nail Dystrophy-Seizures	Ubiquitin cycle, ubiquitin-protein ligase	L-can
2006	ZINC FINGER PROTEIN 674	*ZNF674* *	XLID-Retinal Dystrophy-Short Stature and IDX92	Transcription regulation	Chr-rea (del)
2007	BROMODOMAIN- AND WD REPEAT-CONTAINING PROTEIN 3	*BRWD3*	XLID-Macrocephaly-Large Ears, IDX93	Transcription factor	X-seq
2007	CULLIN 4B	*CUL4B*	XLID-Hypogonadism-Tremor	Cell cycle, ubiquitin cycle, E3 ubiquitin ligase	X-seq
2007	GLUTAMATE RECEPTOR, IONOTROPIC, AMPA 3	*GRIA3*	Chiyonobu XLID, IDX94	Signal transduction, ion transport, glutamate signaling pathway	Chr-rea, Exp-Arr, X-seq
2007	17-BETA-HYDROXYSTEROID DEHYDROGENASE X	*HSD17B10 (HADH2)*	XLID-Choreoathetosis	Lipid metabolism	L-can
2007	MEDIATOR COMPLEX SUBUNIT 12NOTE: MEDIATOR COMPLEX SUBUNIT 12	*MED12 (HOPA)*	Opitz FG, Lujan, IDX67	Transcription regulation, RNA polymerase II transcription mediator activity, ligand-dependent nuclear receptor transcription coactivator activity, vitamin D receptor and thyroid hormone receptor binding	L-can
2007	NADH-UBIQUINONE OXIDOREDUCTASE 1 ALPHA SUBCOMPLEX, 1	*NDUFA1*	Mitochondrial Complex 1 Deficiency	Energy production, oxidoreductase activity	Mol-Fu
2007	NUCLEAR RNA EXPORT FACTOR 5	*NXF5* *	XLID-Short Stature-Muscle Wasting	mRNA processing, mRNA export from nucleus	Chr-rea
2007	PORCUPINE, DROSOPHILA, HOMOLOG	*PORCN*	Goltz	Wnt receptor signaling pathway, acyltransferase activity, integral to membrane of endoplasmic reticulum	Chr-rea (del)
2007	PHOSPHORIBOSYLPYROPHOSPHATE SYNTHETASE I	*PRPS1*	Arts, PRPS1 Superactivity	Ribonucleotide monophosphate biosynthesis	L-can
2007	RIBOSOMAL PROTEIN L10	*RPL10* *	Autism	Protein synthesis, ribosomal protein	X-seq
2007	UPF3, YEAST, HOMOLOG OF, B	*UPF3B*	Lujan/FG Phenotype, IDX62	mRNA catabolism, nonsense-mediated decay	X-seq
2007	ZINC FINGER DHHC DOMAIN-CONTAINING PROTEIN 9	*ZDHHC9*	XLID-Macrocephaly-Marfanoid Habitus		X-seq
2008	HECT, UBA, AND WWE DOMAINS-CONTAINING PROTEIN 1	*HUWE1*	XLID-Macrocephaly, Juberg-Marsidi-Brooks, IDX17, IDX31	Ubiquitin-protein ligase, mRNA transport	M-CGH
2008	PROTOCADHERIN 19	*PCDH19*	Epilepsy-Intellectual Disability Limited to Females		L-can
2008	SOLUTE CARRIER FAMILY 9, MEMBER 6	*SLC9A6*	Christianson, X-linked Angelman-like	Sodium-hydrogen antiporter activity, lysosome organization and biogenesis, regulation of endosome volume	L-can
2009	MAGNESIUM TRANSPORTER 1	*MAGT1* *	IDX95	Magnesium transporter with N-glycosylation sites and putative phosphorylation sites	L-can, X-seq
2009	MEMBRANE-BOUND TRANSCRIPTION FACTOR PROTEASE, SITE 2	*MBTPS2*	Ichthyosis Follicularis, Atrichia, Photophobia (IFAP)	Protease activity, activates signaling proteins	L-can
2009	NAD(P)H STEROID DEHYDROGENASE-LIKE PROTEIN	*NSDHL*	CK (microcephaly, pachygyria, facial dysmorphism, seizures), also in CHILD syndrome	Sterol metabolism	L-can
2009	SYNAPTOPHYSIN	*SYP*	IDX96	Membrane protein of small synaptic vesicles	X-seq
2009	ZINC FINGER PROTEIN 711	*ZNF711*	IDX97	Binds to a subset of PHF8 target genes	X-seq

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2010	IQ MOTIF- AND SEC7 DOMAIN-CONTAINING PROTEIN 2	*IQSEC2*	IDX1, IDX18, and other Nonsyndromal XLID	Regulation of vesicular transport and organelle structure	X-seq
2010	PATCHED DOMAIN-CONTAINING PROTEIN 1	*PTCHD1*	Autism-XLID	Transmembrane protein related to hedgehog receptors	M-CGH
2010	RAS-ASSOCIATED PROTEIN	*RAB39B*	XLID-Macrocephaly-Seizures - Autism, Waisman-Laxova, IDX72	Formation and maintenance of synapse	L-can
2011	CHLORIDE CHANNEL 4	*CLCN4*	IDX15, IDX49	Chloride transport	X-seq
2011	CONNECTOR ENHANCER OF KSR 2	*CNKSR2* *	XLID-Microcephaly-Seizures	Stimulates MAPK signalling	M-CGH
2011	EUKARYOTIC TRANSLATION INITIATION FACTOR 2, SUBUNIT 3	*EIF2S3*	MEHMO	Initiates translation	X-seq
2011	HOST CELL FACTOR C1	*HCFC1* *	IDX3	Cell proliferation	X-seq
2011	HISTONE DEACETYLASE 6	*HDAC6*	Chassaing-Lacombe Chondrodysplasia	Tubulin deacetylase	L-can
2011	HISTONE DEACETYLASE 8	*HDAC8*	Cornelia de Lange, X-linked	Chromatin cohesion	Mol-Fu, X-seq
2011	LAS1-LIKE RIBOSOME BIOGENESIS FACTOR	*LAS1L*	Wilson-Turner	Nucleolar protein, cell proliferation and ribosome biogenesis	X-seq
2011	N-ALPHA-ACETYLTRANSFERASE 10, NatA CATALYTIC SUBUNIT	*NAA10* *	N-Alpha-Acetyltransferase Deficiency	N-terminal acetylation	X-seq
2011	RAS-ASSOCIATED PROTEIN RAB40A-LIKE	*RAB40AL* *	Martin-Probst (?) Questioned: Hum Mutat 35:1171, 2014	Ras-like GTPase protein	X-seq
2011	RNA-BINDING MOTIF PROTEIN 10	*RBM10*	TARP	RNA-binding	X-seq
2011	THO COMPLEX, SUBUNIT 2	*THOC2*	IDX12, IDX35	mRNA transcription or export	X-seq
2012	APOPTOSIS-INDUCING FACTOR, MITOCHONDRIA-ASSOCIATED, 1	*AIFM1*	Charcot-Marie-Tooth disease, Cowchock variant and XLID-spondyloepimetaphyseal dysplasia	Induces apoptosis	X-seq
2012	ALG13, S. CEREVISIAE, HOMOLOG	*ALG13*	CDG1s	Glycosylation	Ex
2012	COILED-COIL DOMAIN-CONTAINING PROTEIN 22	*CCDC22* *	Cardiofacioskeletal (like 3C/Ritscher-Schinzel)	Unknown	X-seq
2012	CHLORIDE INTRACELLULAR CHANNEL 2	*CLIC2* *	XLID-Cardiomegaly-Seizures	Regulating ryanodine receptor channel activity	X-seq
2012	EMOPAMIL-BINDING PROTEIN	*EBP*	Variable manifestations but XLID-Aggression in one family	Enzyme in cholesterol metabolism	Met-Fu
2012	LYSINE-SPECIFIC DEMETHYLASE 6A	*KDM6A*	Kabuki syndrome 2	Histone demethylase and methyltransferase	Chr-rea
2012	PHOSPHATIDYLINOSITOL GLYCAN ANCHOR BIOSYNTHESIS CLASS A PROTEIN	*PIGA*	XLID-Brain iron accumulation	Enzyme, signal transduction pathway, adhesion molecules	X-seq
2012	EPSILON-TRIMETHYLLYSINE HYDROXYLASE	*TMLHE*	Autism-ID	Enzyme in carnitine synthesis	X-seq
2013	B-CELL RECEPTOR-ASSOCIATED PROTEIN 31	*BCAP31*	XLID-microcephaly-dystonia	ER & golgi structure and metabolism	X-seq
2013	SIGNAL SEQUENCE RECEPTOR, DELTA	*SSR4*	XLID-glycosylation defect	Glycosylation	Ex
2013	WD REPEAT-CONTAINING PROTEIN 45	*WDR45*	Neurodegeneration with brain iron accumulation-XLID	Autophagy and other cellular functions	Ex
2013	ZINC FINGER C4H2 DOMAIN-CONTAINING PROTEIN	*ZC4H2*	Wieacker-Wolff, Miles-Carpenter	Axon guidance	Ex, Chr-rea, M-CGH
2014	HIGH MOBILITY GROUP BOX 3	*HMGB3*	Microphthalmia 13	DNA replication, transcription, nucleosome assembly	Ex
2014	KINESIN FAMILY MEMBER 4A	*KIF4A*	IDX100	Moves proteins along microtubule	Ex
2014	MIDLINE 2	*MID2*	IDX101	Enzyme, ubiquity ligase E3 microtubule stabilization	L-can, Ex
2014	UBIQUITIN-SPECIFIC PROTEASE 9, X-LINKED	*USP9X*	IDX99	Neuronal migration growth	X-seq
2014	ZINC FINGER, MYM-TYPE 3	*ZMYM3* *	XLID-aortic stenosis-hypospadias	Component of histone deacetylase-containing multiple protein complexes	X-seq
2015	DEAD/H BOX 3, X-LINKED	*DDX3X*	Variable cognitive, neurologic and nonneurologic manifestations, IDX102	RNA helicase	Ex
2015	FERM AND PDZ DOMAINS-CONTAINING PROTEIN 4	*FRMPD4* *	XLID-Aphasia-seizures, IDX104	Regulates spine morphogenesis	X-seq
2015	KELCH-LIKE 15	*KLHL15*	Mild to moderate cognitive impairment, facial dysmorphism, IDX103	Uncertain	X-seq
2015	MALE-SPECIFIC LETHAL 3, DROSOPHILA, HOMOLOG	*MSL3*	MSL3-Related XLID	Transcription regulation, chromatin modifier	Ex

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2015	NON-POU DOMAIN-CONTAINING OCTAMER-BINDING PROTEIN	*NONO*	Mircsof-Langouët	Regulation of transcription (activation, repression, splicing, pre-mRNA processing, RNA transport)	Ex
2015	RNA-BINDING MOTIF PROTEIN, X CHROMOSOME	*RBMX*	Shashi	RNA binding	Ex
2015	RING FINGER PROTEIN, LIM DOMAIN-INTERACTING	*RLIM (RNF12)*	IDX with variable microcephaly, behaviour abnormalities and other manifestation, IDX61	Enzyme, ubiquity ligase E3	X-seq
2015	RING FINGER PROTEIN 113A	*RNF113A*	Trichothiodystrophy 5	Gene regulation, DNA repair	Ex
2015	TAF1 RNA POLYMERASE II, TATA BOX-BINDING PROTEIN-ASSOCIATED FACTOR, 250-KD	*TAF1*	XLID-Craniofacial-Caudal, Abidi	Key role in initiating transcription	Gen-seq
2015	UBIQUITIN-SPECIFIC PROTEASE 27, X-LINKED	*USP27X*	Variable cognitive and speech impairment and behavioural abnormalities, IDX105	Peptidase	X-seq
2016	EF-HAND DOMAIN (C-TERMINAL)-CONTAINING PROTEIN 2	*EFHC2*	IDX74	Calcium binding and other unknown functions	L-can
2016	GRIP1-ASSOCIATED PROTEIN 1	*GRASP1*	XLID-Short Stature-Spasticity	Synaptic function and neuronal connectivity	X-seq
2016	HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN H2	*HNRNPH2*	Bain XLID, female limited	Nuclear localization	Ex
2016	SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL CARRIER, ADENINE NUCLEOTIDE TRANSLOCATOR), MEMBER A5	*SLC25A5*	IDX70	Mitochondrial exchange of ADP/ATP	M-CGH
2016	STROMAL ANTIGEN 2	*STAG2*	STAG2-Related XLID	Chromatin cohesion	M-CGH
2017	G-PATCH DOMAIN AND KOW MOTIFS	*GPKOW*	XLID-Microcephaly-early lethality	Spliceosome component	X-seq
2017	O-LINKED N-ACETYLGLUCOSAMINE TRANSFERASE	*OGT*	XLID-Faciogenital	Post translational modification of nucleocytoplasmic proteins	Ex
2018	CHROMOSOME X OPEN READING FRAME 56	*CXorf56*	Dysmorphic faces, long face, prominent chin, mild ID, behavior problems	Dendritic neuronal protein	Gen-seq
2018	FAMILY WITH SEQUENCE SIMILARITY 50, MEMBER A	*FAM50A*	Short stature, ocular problems, seizures, small hands, feet, DD, mild ID, Armfield syndrome	Spliceosome protein	X-seq, L-can
2018	ZINC FINGER PROTEIN 92	*ZNF92*	Hypotonia, behavioral, mitochrondrial issues, DD, ID	Mitochrondrial functions	Ex, L-can
2018	SOLUTE CARRIER FAMILY 9, MEMBER 7	*SLC9A7*	IDX (hypotonia, muscle weakness, brisk reflexes	Cation exchange factor	X-seq
2018	HEPARAN SULFATE 6-O-SULFOTRANSFERASE 2	*HS6ST2*	ID, seizures, myopia, retinopathy	Sulfotransferase of heparan sulfate proteoglycans	Ex
2019	POLYMERASE, DNA, ALPHA-1; POLA1	*POLA1*	ID, growth impairment, hypogonadism	Subunit of polymerase alpha-primase	Ex, L-can

* Association of *ZNF674, RAB40AL, ZMYM3, MAGT1* and XLID considered uncertain

Chr-rea = chromosome rearrangement
Ex = exome sequencing
Gen-seq = genome sequencing
Exp-Arr = expression array
L-can = linkage and candidate gene testing
M-CGH = array-comparative genomic hybridization
Met-Fu = exploitation of metabolic alteration
Mol-Fu = exploitation of molecular finding
X-seq = sequencing of exons plus