devSEEK® Panel – Gene List by Gene

Gene	Disease association	Category
ABCD1	Adrenoleukodystrophy	Other
ACSL4	Intellectual disability, X-linked 63	Intellectual Disability
ADAT3	Intellectual disability, autosomal recessive 36	Intellectual Disability
ADNP	Helsmoortel-van der Aa syndrome	Syndromic Intellectual Disability
AFF2	Intellectual disability, X-linked, FRAXE type	Intellectual Disability
AGTR2	X-linked non-syndromic intellectual disability	Intellectual Disability
AIFM1	Combined oxidative phosphorylation deficiency 6	Mitochondrial Rspiratory Chain Dysfunction
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency	Intellectual Disability, Seizures, Neurologic Disease
ALG6	Congenital disorder of glycosylation type Ic	Syndromic Intellectual Disability
AMT	Glycine encephalopathy	Intellectual Disability, Seizures, Neurologic Disease
ANK3	Intellectual disability, autosomal recessive 37	Intellectual Disability
AP1S2	Intellectual disability, X-linked syndromic, Fried type	Intellectual Disability
AP4B1	Spastic paraplegia 47, atuosomal recessive	Spastic paraplegia
AP4E1	Spastic paraplegia 51, atuosomal recessive	Spastic paraplegia
AP4M1	Spastic paraplegia 50, atuosomal recessive	Spastic paraplegia
AP4S1	Spastic paraplegia 52, atuosomal recessive	Spastic paraplegia
ARFGEF2	periventricular hereotopia & microcephaly	Intellectual Disability
ARHGEF6	Intellectual disability, X-linked 46	Intellectual Disability
ARHGEF9	Epileptic encephalopathy, early infantile, 8	Epilepsy
ARID1A	Intellectual disability, autosomal dominant 14	Intellectual Disability
ARID1B	Intellectual disability, autosomal dominant 12 (Coffin-Siris syndrome)	Intellectual Disability
ARX	Epileptic encephalopathy, early infantile, 1; Proud Syndrome; X-linked lissencephaly with abnormal genitalia	Brain or Nervous System Malformations
ATP6AP2	Intellectual disability, X-linked, syndromic, Hedera type	Intellectual Disability
ATP7A	Menkes disease	Intellectual Disability, Seizures, Neurologic Disease
ATRX	Alpha thalassemia X-linked intellectual disability syndrome; Intellectual disability-hyptonic facies syndrome, x-linked	Intellectual Disability
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency	Selected inborn errors of metabolism
BCOR	Lenz Microphthalmia Syndrome; OCULOFACIOCARDIODENTAL SYNDROME (OFCD)	Syndromic Intellectual Disability
BRAF	Cardiofaciocutaneous syndrome; LEOPARD sydrome; Noonan syndrome	Ras/MAPK Pathway-related Disorder
BRWD3	Intellectual disability, X-linked 93	Intellectual Disability
C12orf57	Temtamy Syndrome	Syndromic Intellectual Disability
CA8	Cerebellar ataxia and Intellectual disability with or without quadrupedal locomotion 3	Intellectual Disability
CACNA1C	Timothy syndrome	Syndromic Intellectual Disability
CASK	FG syndrome 4; Intellectual disability and microcephaly with pontine and cerebellar hypoplasia	Brain or Nervous System Malformations
CBL	Noonan syndrome-like disorder	Ras/MAPK Pathway-related Disorder
CC2D1A	Intellectual disability, autosomal recessive 3	Intellectual Disability
CCDC22	Associated with intellectual disability	Intellectual Disability
CDH15	Intellectual disability, autosomal dominant 3	Intellectual Disability
CDKL5	Angleman Syndrome; epileptic encephalopathy, early infantile, 2	Rett/atypical Rett syndromes
CHD7	CHARGE syndrome	Syndromic Intellectual Disability
CHD8	Autism, susceptibility to, 18	Autism Spectrum Disorder
CLCN4	candidate for X-linked intellectual disability	Intellectual Disability

Gene	Disease association	Category
CLIC2	Intellectual disability, X-linked, syndromic 32	Intellectual Disability
CLIP1	Autosomal recessive non-syndromic intellectual disability	Intellectual Disability
CNKSR2	Undetermined early-onset epileptic encephalopathy	Intellectual Disability, Seizures, Neurologic Disease
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome; pitt-hopkins syndrome	Angelman/Angelman-like/Pitt-Hopkins syndromes
CRBN	Intellectual disability, autosomal recessive 2	Intellectual Disability
CREBBP	Rubinstein-Taybi syndrome	Rubinstein-Taybi syndrome
CTCF	Intellectual disability, autosomal dominant 21	Intellectual Disability
CTNNB1	Intellectual disability, autosomal dominant 19	Intellectual Disability
CUL4B	Intellectual disability, X-linked, syndromic 15(Cabezas type)	Intellectual Disability
D2HGDH	D-2-hydroxyglutaric aciduria	Hydroxyglutaric acidurias
DCX	epilepsy, Intellectual disability, subcortical band heterotopia ("double cortex" syndrome) in females and lissencephaly ("smooth brain" syndrome) in males	Brain or Nervous System Malformations
DDHD2	Spastic paraplegia 54, autosomal recessive	Spastic paraplegia
DEAF1	Intellectual disability, autosomal dominant 24	Intellectual Disability
DHCR7	Smith-Lemli-Opitz syndrome	Syndromic Intellectual Disability
DKC1	Dyskeratosis congenita, X-linked	Syndromic Intellectual Disability
DLG3	Intellectual disability, X-linked 90	Intellectual Disability
DMD	Becker muscular dystrophy; Duchenne muscular dystrophy	Syndromic Intellectual Disability
DNMT3A	Tatton-Brown-Rahman syndrome	Syndromic Intellectual Disability
DYNC1H1	AD intellectual disability 12; Charcot-Marie-Tooth disease, axonal, type 20; spinal muscular atrophy	Syndromic Intellectual Disability
DYRK1A	Intellectual disability, autosomal dominant 7	Intellectual Disability
EBP	MEND syndrome	Syndromic Intellectual Disability
EHMT1	Kleefstra syndrome	Kleefstra syndrome
ERLIN2	Spastic paraplegia 18, autosomal recessive	Spastic paraplegia
EZR	Autosomal recessive non-syndromic intellectual disability	Intellectual Disability
FAAH2	candidate for X-linked intellectual disability	Intellectual Disability
FANCB	Fanconi anemia, complementation group b	Syndromic Intellectual Disability
FGD1	AARSKOG-SCOTT SYNDROME (faciogenital dysplasia) and X-linked Intellectual disability, syndromatic 16	Syndromic Intellectual Disability
FGF8	Hypogonadotropic hypogonadism 6 with or without anosmia	Autism association
FLNA	FG syndrome; Heterotopia, periventricular	Brain or Nervous System Malformations
FMN2	Intellectual disability, autosomal recessive 47	Intellectual Disability
FMR1* **	Fragile X syndrome and FMR1-related disorders	Syndromic Intellectual Disability
FOLR1	Neurodegeneration due to cerebral folate transport deficiency	Syndromic Intellectual Disability
FOXG1	Rett syndrome, congenital variant	Rett/atypical Rett syndromes
FOXP1	Intellectual disability with language impairment and autistic features	Autism susceptibility
FOXP2	Speech-language disorder-1	Other, DD, ID, ASD related disorders
FRMPD4	Associated with intellectual disability	Intellectual Disability
FTSJ1	Intellectual disability, X-linked 9	Intellectual Disability
GABRB3	Epilepsy, childhood absence, susceptibility to, 5	Epilepsy
GATAD2B	autosomal dominant intellectual disability 18	Intellectual Disability
GDI1	X-linked intellectual disability 41	Intellectual Disability
GJB1	Charcot-Marie-Tooth disease, X-linked dominant, 1	Peripheral Neuropathy
GK	Glycerol kinase deficiency	Intellectual Disability, Seizures, Neurologic Disease
GPC3	Simpson-Golabi-Behmel syndrome, type 1	Syndromic Intellectual Disabiity
GRIA3	Intellectual disability, X-linked 94	Intellectual Disability
GRIK2	Intellectual disability, autosomal recessive, 6	Intellectual Disability
GRIN2A	Epilepsy, focal, with speech disorder and with or without Intellectual disability	Epilepsy
GRIN2B	Intellectual disability, autosomal dominant 6	Intellectual Disability
GSPT2	candidate for X-linked intellectual disability	Intellectual Disability
HCCS	Microphthalmia, syndromic 7	Syndromic Intellectual Disability
HCFC1	Intellectual disability, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	Intellectual Disability
HDAC8	Cornelia de Lange syndrome 5; Wilson-Turner syndrome	Cornelia de Lange syndrome
HOXA1	Athabaskan brainstem dysgenesis syndrome; Bosley-Salih-Alorainy syndrome	Syndromic Intellectual Disability

Gene	Disease association	Category
HPRT1	Lesch-Nyhan syndrome	Selected inborn errors of metabolism
HRAS	Costello syndrome (Congenital myopathy with excess of muscle spindles)	Ras/MAPK Pathway-related Disorder
HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency; Intellectual disability, X-linked syndromic 10	Other, DD, ID, ASD related disorders
HUWE1	Intellectual disability, X-linked syndromic, Turner type	Intellectual Disability
IDS	Mucopolysaccharidosis type ii	Mucopolysaccharidosis
IGBP1	Corpus callosum, agenesis of, with Intellectual disability, ocular coloboma and micrognathia	Brain or Nervous System Malformations
IL1RAPL1	Intellectual disability, X-linked 21/34	Intellectual Disability
IQSEC2	Intellectual disability, X-linked 1	Intellectual Disability
KCNJ10	SESAME syndrome	Syndromic Intellectual Disability
KDM5C	Intellectual disability, X-linked, syndromic, Claes-Jensen type	Intellectual Disability
KIAA2022	Intellectual disability, X-linked	Intellectual Disability
KIF1A	Intellectual disability, autosomal dominant 9; Spastic paraplegia 30, autosomal recessive	Intellectual Disability
KIRREL3	Intellectual disability, autosomal dominant 4	Intellectual Disability
KLF8	Associated with intellectual disability	Intellectual Disability
KMT2D	Kabuki Syndrome	Syndromic Intellectual Disability
KMTD2	Kabuki syndrome	Syndromic Intellectual Disability
KPTN	Intellectual disability, autosomal recessive 41	Intellectual Disability
KRAS	Noonan syndrome; Cardiofaciocutaneous syndrome	Ras/MAPK Pathway-related Disorder
L1CAM	Corpus callosum, partial agenesis of; CRASH/ MASAsyndrome	Brain or Nervous System Malformations
L2HGDH	L-2-hydroxyglutaric aciduria	Hydroxyglutaric acidurias
LAMC3	Cortical malformations, occipital	Brain/Nervous System Malformation
LAMP2	Danon disease	Cardiomyopathy/Myopathy
LAS1L	Spinal muscular atrophy with respiratory distress type 2	Motor Neuron Disease
LINS	Intellectual disability, autosomal recessive 27	Intellectual Disability
MAN1B1	Intellectual disability, autosomal recessive 15	Intellectual Disability
MAOA	Brunner syndrome	Syndromic Intellectual Disability
MAP2K1	Cardiofaciocutaneous syndrome	Ras/MAPK Pathway-related Disorder
MAP2K2	Cardiofaciocutaneous syndrome	Ras/MAPK Pathway-related Disorder
MBD5	Intellectual disability, autosomal dominant 1 (MRD1)	Rett/atypical Rett syndromes
MBTPS2	IFAP syndrome with or without Bresheck syndrome	Syndromic Intellectual Disability
MECP2	Encephalopathy, neonatal severe; Rett syndrome	Rett/atypical Rett syndromes
MED12	Lujan-Fryns syndrome; Ohdo syndrome, X-linked; Opitz-Kaveggia syndrome	Syndromic Intellectual Disability
MED23	Intellectual disability, autosomal recessive 18 (MRT18)	Intellectual Disability
MEF2C	Rett/atypical Rett syndromes	Rett/atypical Rett syndromes
METTL23	Intellectual disability, autosomal recessive 44	Intellectual Disability
MID1	Opitz GBBB syndrome, type I	Syndromic Intellectual Disability
NAA10	Microphthalmia, syndrome 1; N-terminal acetyltransferase deficiency	Other, DD, ID, ASD related disorders
NDP	Exudative vitreoretinopathy 2, X-linked	Neurodegenerative Disorder
NDST1	Intellectual disability, autosomal recessive 46	Intellectual Disability
NDUFA1	Mitochondrial complex i deficiency	Mitochondrial Disease
NF1	Neurofibramatosis-Noonan syndrome; Watson syndrome	Ras/MAPK Pathway-related Disorder
NHS	Nance-Horan syndrome	Syndromic Intellectual Disability
NIPBL	Cornelia de Lange syndrome 1	Cornelia de Lange syndrome
NLGN3	Asperger syndrome susceptibility, X-linked 1; Autism susceptibility, X-linked 1	Autism susceptibility
NLGN4X	Asperger syndrome susceptibility, X-linked 2; Autism susceptibility, X-linked 2; Intellectual disability, X-linked	Autism susceptibility
NRAS	Noonan syndrome	Ras/MAPK Pathway-related Disorder
NRXN1	Pitt-Hopkins-like syndrome 2	Angelman/Angelman-like/Pitt-Hopkins syndromes
NSD1	Beckwith-Wiedemann syndrome; Sotos syndrome 1	Syndromic Intellectual Disability
NSDHL	CHILD syndrome; CK syndrome	Syndromic Intellectual Disability
NSUN2	Intellectual disability, autosomal recessive 5	Intellectual Disability
OCRL	Dent disease 2; Lowe syndrome	Syndromic Intellectual Disability

Gene	Disease association	Category
OFD1	Joubert syndrome 10; Oral-facial-digital syndrome 1; Simpson-Golabi-Behmel syndrome, type 2	Syndromic Intellectual Disability
OPHN1	Intellectual disability, X-linked, with cerebellar hypoplasia and distinctive facial appearance	Intellectual Disability
OTC	Ornithine transcarbamylase deficiency, hyperammonemia due to	IEM
PACS1	Intellectual disability, autosomal dominant 17	Intellectual Disability
PAFAH1B1	Lissencephaly 1; Subcortical laminar heterotopia	Brain or Nervous System Malformations
PAK3	Intellectual disability, X-linked 30/47	Intellectual Disability
PCDH19	Epileptic encephalopathy, early infantile, 9	Epilepsy
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	Other, DD, ID, ASD related disorders
PDHA1	Leigh syndrome, X-linked; Pyruvate dehydrogenase E1-alpha deficiency	Syndromic Intellectual Disability
PGAP1	Intellectual disability, autosomal recessive 42	Intellectual Disability
PGK1	Phosphoglycerate kinase 1 deficiency	Other
PHF6	Borjeson-Forssman-Lehmann syndrome	Syndromic Intellectual Disability
PHF8	Intellectual disability syndrome, X-linked, Siderius type	Intellectual Disability
PLP1	Pelizaeus-Merzbacher disease; Spastic paraplegia 2, X-linked	Spastic paraplegia
PNKP	Epileptic encephalopathy, early infantile, 10	Epilepsy
PORCN	Focal dermal hypoplasia	Syndromic Intellectual Disability
PQBP1	Renpenning syndrome	Syndromic Intellectual Disability
PRPS1	Arts syndrome; Charcot-Marie-Tooth disease, X-linked recessive, 5	Syndromic Intellectual Disability
PRSS12	Intellectual disability, autosomal recessive 1	Intellectual Disability
PTCHD1	Autism susceptibility	Autism susceptibility
PTEN	Bannayan-Riley-Ruvalcaba syndrome; Cowden syndrome 1; Macrocephaly/autism syndrome	Syndromic Intellectual Disability
PTPN11	LEOPARD syndrome; Noonan syndrome	Ras/MAPK Pathway-related Disorder
PURA	Intellectual disability, autosomal dominant 31	Intellectual Disability
RAB39B	Intellectual disability, X-linked 72	Intellectual Disability
RAB40AL	Martin-Probst X-linked deafness-intellectual disability syndrome	Syndromic Intellectual Disability
RAD21	Cornelia de Lange syndrome 4	Cornelia de Lange syndrome
RAF1	Noonan syndrome; LEOPARD syndrome	Ras/MAPK Pathway-related Disorder
RAI1	Smith-Magenis syndrome	Syndromic Intellectual Disability
RBM10	Tarp syndrome	Syndromic Intellectual Disability
RELN	Lissencephaly 2 (Norman-Roberts type)	Brain or Nervous System Malformations
RPL10	Autism, susceptibility to, X-linked 5	Autism susceptibility
RPS6KA3	Coffin-Lowry syndrome; Intellectual disability, X-linked 19	Syndromic Intellectual Disability
SCN1A	Dravet syndrome; Epilepsy, generalized, with febrile seizures plus, type 2 / Febrile seizures, familial, 3A	Epilepsy
SCN2A	Epileptic encephalopathy, early infantile, 11; Seizures, benign familial infantile, 3	Epilepsy
SETBP1	Intellectual disability, autosomal dominant 29	Intellectual Disability
SETD2	Luscan-Lumish syndrome	Syndromic Intellectual Disability
SETD5	Intellectual disability, autosomal dominant 23	Intellectual Disability
SHANK2	Autism susceptibility 17	Autism susceptibility
SHANK3	Phelan-McDermid syndrome	Syndromic Intellectual Disability
SHOC2	Noonan syndrome-like disorder	Ras/MAPK Pathway-related Disorder
SLC16A2	Allan-Herndon-Dudley syndrome	syndromic Intellectual Disability
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria	Hydroxyglutaric acidurias
SLC2A1	Epilepsy, idiopathic generalized, suscpetibility to, 12; GLUT1 deficiency syndrome 1; GLUT1 deficiency syndrome 2	Syndromic Intellectual Disability
SLC6A17	Intellectual disability, autosomal recessive 48	Intellectual Disability
SLC6A8	Cerebral creatine deficiency syndrome 1	Syndromic Intellectual Disability
SLC9A6	Intellectual disability, X-linked syndromic, Christianson type	Intellectual Disability
SLC9A9	Autism susceptibility	Autism susceptibility
SMARCB1	Intellectual disability, autosomal dominant 15	Syndromic Intellectual Disability
SMC1A	Cornelia de Lange syndrome 2	Cornelia de Lange syndrome
SMC3	Cornelia de Lange syndrome 3	Cornelia de Lange syndrome
SMS	Intellectual disability, X-linked, Snyder-Robinson type	Intellectual Disability
SOS1	Noonan syndrome	Ras/MAPK Pathway-related Disorder

Gene	Disease association	Category
SOX11	Coffin-siris syndrome	Syndromic Intellectual Disability
SOX3	Intellectual disability, X-linked, with panhypopituitarismIntellectual disability, X-linked, with isolated growth hormone deficiency,included	Intellectual Disability
SPRED1	Legius syndrome	Ras/MAPK Pathway-related Disorder
SRPX2	Rolandic epilepsy, Intellectual disability, and speech dyspraxia	Intellectual Disability
ST3GAL3	Epileptic encephalopathy, early infantile, 15; Intellectual disability, autosomal recessive 12	Epilepsy
STXBP1	Epileptic encephalopathy, early infantile, 4	Epilepsy
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	Epilepsy
SYNGAP1	Intellectual disability, autosomal dominant 5	Intellectual Disability
SYP	Intellectual disability, X-linked 96	Intellectual Disability
TAF2	Intellectual disability, autosomal recessive 40	Intellectual Disability
TBL1XR1	Pierpont syndrome	Syndromic Intellectual Disability
TBR1	Autism spectrum disorder	Autism Spectrum Disorder
TBX1	DiGeorge syndrome; Tetralogy of Fallot; Velocardiofacial syndrome	Syndromic Intellectual Disability
TCF4	Pitt-Hopkins syndrome	Angelman/Angelman-like/Pitt-Hopkins syndromes
TECR	Intellectual disability, autosomal recessive 14	Intellectual Disability
THOC2	Intellectual disability, X-linked 12/35	Intellectual Disability
TIMM8A	Opticoacoustic nerve atrophy with dementia	Neurodegenerative Disorder
TMEM231	Joubert syndrome 20	Ciliopathy
TMLHE	Epsilon-trimethyllysine hydroxylase deficiency	Autism Spectrum Disorder
TRAPPC9	Intellectual disability, autosomal recessive 13	Intellectual Disability
TSC1	Tuberous sclerosis-1	Tuberous sclerosis
TSC2	Tuberous sclerosis-2	Tuberous sclerosis
TSPAN7	Intellectual disability, X-linked 58	Intellectual Disability
TTI2	Intellectual disability, autosomal recessive 39	Intellectual Disability
TUBA1A	Lissencephaly 3	Brain or Nervous System Malformations
TUSC3	Intellectual disability, autosomal recessive 7	Intellectual Disability
UBE2A	Intellectual disability, X-linked syndromic, Nascimento-type	Intellectual Disability
UBE3A	Angelman syndrome	Angelman/Angelman-like/Pitt-Hopkins syndromes
UPF3B	Intellectual disability, X-linked, syndromic 14	Intellectual Disability
USP9X	Intellectual disability, X-linked 99	Intellectual Disability
VLDLR	Cerebellar hypoplasia and Intellectual disability with or without quadrupedal locomotion 1	Brain or Nervous System Malformations
VPS13B	Cohen syndrome	Syndromic Intellectual Disability
WDR13	candidate for X-linked intellectual disability	Intellectual Disability
ZC4H2	Wieacker-Wolff syndrome	Syndromic Intellectual Disability
ZDHHC15	Intellectual disability, X-linked 91	Intellectual Disability
ZDHHC9	Intellectual disability, X-linked syndromic, Raymond type	Intellectual Disability
ZEB2	Mowat-Wilson syndrome	Syndromic Intellectual Disability
ZMYND11	Intellectual disability, autosomal dominant 30	Intellectual Disability
ZNF41	X-linked non-syndromic intellectual disability	Intellectual Disability
ZNF674	X-linked non-syndromic intellectual disability	Intellectual Disability
ZNF711	Intellectual disability, X-linked 97	Intellectual Disability
ZNF81	Intellectual disability, X-linked 45	Intellectual Disability

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Gene	Disease association	Category
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome; pitt-hopkins syndrome	Angelman/Angelman-like/Pitt-Hopkins syndromes
NRXN1	Pitt-Hopkins-like syndrome 2	Angelman/Angelman-like/Pitt-Hopkins syndromes
TCF4	Pitt-Hopkins syndrome	Angelman/Angelman-like/Pitt-Hopkins syndromes
UBE3A	Angelman syndrome	Angelman/Angelman-like/Pitt-Hopkins syndromes
FGF8	Hypogonadotropic hypogonadism 6 with or without anosmia	Autism association
CHD8	Autism, susceptibility to, 18	Autism Spectrum Disorder
TBR1	Autism spectrum disorder	Autism Spectrum Disorder
TMLHE	Epsilon-trimethyllysine hydroxylase deficiency	Autism Spectrum Disorder
FOXP1	Intellectual disability with language impairment and autistic features	Autism susceptibility
NLGN3	Asperger syndrome susceptibility, X-linked 1; Autism susceptibility, X-linked 1	Autism susceptibility
NLGN4X	Asperger syndrome susceptibility, X-linked 2; Autism susceptibility, X-linked 2; Intellectual disability, X-linked	Autism susceptibility
PTCHD1	Autism susceptibility	Autism susceptibility
RPL10	Autism, susceptibility to, X-linked 5	Autism susceptibility
SHANK2	Autism susceptibility 17	Autism susceptibility
SLC9A9	Autism susceptibility	Autism susceptibility
ARX	Epileptic encephalopathy, early infantile, 1; Proud Syndrome; X-linked lissencephaly with abnormal genitalia	Brain or Nervous System Malformations
CASK	FG syndrome 4; Intellectual disability and microcephaly with pontine and cerebellar hypoplasia	Brain or Nervous System Malformations
DCX	epilepsy, Intellectual disability, subcortical band heterotopia ("double cortex" syndrome) in females and lissencephaly ("smooth brain" syndrome) in males	Brain or Nervous System Malformations
FLNA	FG syndrome; Heterotopia, periventricular	Brain or Nervous System Malformations
IGBP1	Corpus callosum, agenesis of, with Intellectual disability, ocular coloboma and micrognathia	Brain or Nervous System Malformations
L1CAM	Corpus callosum, partial agenesis of; CRASH/ MASAsyndrome	Brain or Nervous System Malformations
PAFAH1B1	Lissencephaly 1; Subcortical laminar heterotopia	Brain or Nervous System Malformations
RELN	Lissencephaly 2 (Norman-Roberts type)	Brain or Nervous System Malformations
TUBA1A	Lissencephaly 3	Brain or Nervous System Malformations
VLDLR	Cerebellar hypoplasia and Intellectual disability with or without quadrupedal locomotion 1	Brain or Nervous System Malformations
LAMC3	Cortical malformations, occipital	Brain/Nervous System Malformation
LAMP2	Danon disease	Cardiomyopathy/Myopathy
TMEM231	Joubert syndrome 20	Ciliopathy
HDAC8	Cornelia de Lange syndrome 5; Wilson-Turner syndrome	Cornelia de Lange syndrome
NIPBL	Cornelia de Lange syndrome 1	Cornelia de Lange syndrome
RAD21	Cornelia de Lange syndrome 4	Cornelia de Lange syndrome
SMC1A	Cornelia de Lange syndrome 2	Cornelia de Lange syndrome
SMC3	Cornelia de Lange syndrome 3	Cornelia de Lange syndrome
ARHGEF9	Epileptic encephalopathy, early infantile, 8	Epilepsy
GABRB3	Epilepsy, childhood absence, susceptibility to, 5	Epilepsy
GRIN2A	Epilepsy, focal, with speech disorder and with or without Intellectual disability	Epilepsy
PCDH19	Epileptic encephalopathy, early infantile, 9	Epilepsy
PNKP	Epileptic encephalopathy, early infantile, 10	Epilepsy
SCN1A	Dravet syndrome; Epilepsy, generalized, with febrile seizures plus, type 2 / Febrile seizures, familial, 3A	Epilepsy
SCN2A	Epileptic encephalopathy, early infantile, 11; Seizures, benign familial infantile, 3	Epilepsy
ST3GAL3	Epileptic encephalopathy, early infantile, 15; Intellectual disability, autosomal recessive 12	Epilepsy
STXBP1	Epileptic encephalopathy, early infantile, 4	Epilepsy
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	Epilepsy

Gene	Disease association	Category
D2HGDH	D-2-hydroxyglutaric aciduria	Hydroxyglutaric acidurias
L2HGDH	L-2-hydroxyglutaric aciduria	Hydroxyglutaric acidurias
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria	Hydroxyglutaric acidurias
OTC	Ornithine transcarbamylase deficiency, hyperammonemia due to	IEM
ACSL4	Intellectual disability, X-linked 63	Intellectual Disability
ADAT3	Intellectual disability, autosomal recessive 36	Intellectual Disability
AFF2	Intellectual disability, X-linked, FRAXE type	Intellectual Disability
AGTR2	X-linked non-syndromic intellectual disability	Intellectual Disability
ANK3	Intellectual disability, autosomal recessive 37	Intellectual Disability
AP1S2	Intellectual disability, X-linked syndromic, Fried type	Intellectual Disability
ARFGEF2	periventricular hereotopia & microcephaly	Intellectual Disability
ARHGEF6	Intellectual disability, X-linked 46	Intellectual Disability
ARID1A	Intellectual disability, autosomal dominant 14	Intellectual Disability
ARID1B	Intellectual disability, autosomal dominant 12 (Coffin-Siris syndrome)	Intellectual Disability
ATP6AP2	Intellectual disability, X-linked, syndromic, Hedera type	Intellectual Disability
ATRX	Alpha thalassemia X-linked intellectual disability syndrome; Intellectual disability-hyptonic facies syndrome, x-linked	Intellectual Disability
BRWD3	Intellectual disability, X-linked 93	Intellectual Disability
CA8	Cerebellar ataxia and Intellectual disability with or without quadrupedal locomotion 3	Intellectual Disability
CC2D1A	Intellectual disability, autosomal recessive 3	Intellectual Disability
CCDC22	Associated with intellectual disability	Intellectual Disability
CDH15	Intellectual disability, autosomal dominant 3	Intellectual Disability
CLCN4	candidate for X-linked intellectual disability	Intellectual Disability
CLIC2	Intellectual disability, X-linked, syndromic 32	Intellectual Disability
CLIP1	Autosomal recessive non-syndromic intellectual disability	Intellectual Disability
CRBN	Intellectual disability, autosomal recessive 2	Intellectual Disability
CTCF	Intellectual disability, autosomal dominant 21	Intellectual Disability
CTNNB1	Intellectual disability, autosomal dominant 19	Intellectual Disability
CUL4B	Intellectual disability, X-linked, syndromic 15(Cabezas type)	Intellectual Disability
DEAF1	Intellectual disability, autosomal dominant 24	Intellectual Disability
DLG3	Intellectual disability, X-linked 90	Intellectual Disability
DYRK1A	Intellectual disability, autosomal dominant 7	Intellectual Disability
EZR	Autosomal recessive non-syndromic intellectual disability	Intellectual Disability
FAAH2	candidate for X-linked intellectual disability	Intellectual Disability
FMN2	Intellectual disability, autosomal recessive 47	Intellectual Disability
FRMPD4	Associated with intellectual disability	Intellectual Disability
FTSJ1	Intellectual disability, X-linked 9	Intellectual Disability
GATAD2B	autosomal dominant intellectual disability 18	Intellectual Disability
GDI1	X-linked intellectual disability 41	Intellectual Disability
GRIA3	Intellectual disability, X-linked 94	Intellectual Disability
GRIK2	Intellectual disability, autosomal recessive, 6	Intellectual Disability
GRIN2B	Intellectual disability, autosomal dominant 6	Intellectual Disability
GSPT2	candidate for X-linked intellectual disability	Intellectual Disability
HCFC1	Intellectual disability, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	Intellectual Disability
HUWE1	Intellectual disability, X-linked syndromic, Turner type	Intellectual Disability
IL1RAPL1	Intellectual disability, X-linked 21/34	Intellectual Disability
IQSEC2	Intellectual disability, X-linked 1	Intellectual Disability
KDM5C	Intellectual disability, X-linked, syndromic, Claes-Jensen type	Intellectual Disability
KIAA2022	Intellectual disability, X-linked	Intellectual Disability
KIF1A	Intellectual disability, autosomal dominant 9; Spastic paraplegia 30, autosomal recessive	Intellectual Disability
KIRREL3	Intellectual disability, autosomal dominant 4	Intellectual Disability
KLF8	Associated with intellectual disability	Intellectual Disability
KPTN	Intellectual disability, autosomal recessive 41	Intellectual Disability
LINS	Intellectual disability, autosomal recessive 27	Intellectual Disability
MAN1B1	Intellectual disability, autosomal recessive 15	Intellectual Disability

Gene	Disease association	Category
MED23	Intellectual disability, autosomal recessive 18 (MRT18)	Intellectual Disability
METTL23	Intellectual disability, autosomal recessive 44	Intellectual Disability
NDST1	Intellectual disability, autosomal recessive 46	Intellectual Disability
NSUN2	Intellectual disability, autosomal recessive 5	Intellectual Disability
OPHN1	Intellectual disability, X-linked, with cerebellar hypoplasia and distinctive facial appearance	Intellectual Disability
PACS1	Intellectual disability, autosomal dominant 17	Intellectual Disability
PAK3	Intellectual disability, X-linked 30/47	Intellectual Disability
PGAP1	Intellectual disability, autosomal recessive 42	Intellectual Disability
PHF8	Intellectual disability syndrome, X-linked, Siderius type	Intellectual Disability
PRSS12	Intellectual disability, autosomal recessive 1	Intellectual Disability
PURA	Intellectual disability, autosomal dominant 31	Intellectual Disability
RAB39B	Intellectual disability, X-linked 72	Intellectual Disability
SETBP1	Intellectual disability, autosomal dominant 29	Intellectual Disability
SETD5	Intellectual disability, autosomal dominant 23	Intellectual Disability
SLC6A17	Intellectual disability, autosomal recessive 48	Intellectual Disability
SLC9A6	Intellectual disability, X-linked syndromic, Christianson type	Intellectual Disability
SMS	Intellectual disability, X-linked, Snyder-Robinson type	Intellectual Disability
SOX3	Intellectual disability, X-linked, with panhypopituitarismIntellectual disability, X-linked, with isolated growth hormone deficiency,included	Intellectual Disability
SRPX2	Rolandic epilepsy, Intellectual disability, and speech dyspraxia	Intellectual Disability
SYNGAP1	Intellectual disability, autosomal dominant 5	Intellectual Disability
SYP	Intellectual disability, X-linked 96	Intellectual Disability
TAF2	Intellectual disability, autosomal recessive 40	Intellectual Disability
TECR	Intellectual disability, autosomal recessive 14	Intellectual Disability
THOC2	Intellectual disability, X-linked 12/35	Intellectual Disability
TRAPPC9	Intellectual disability, autosomal recessive 13	Intellectual Disability
TSPAN7	Intellectual disability, X-linked 58	Intellectual Disability
TTI2	Intellectual disability, autosomal recessive 39	Intellectual Disability
TUSC3	Intellectual disability, autosomal recessive 7	Intellectual Disability
UBE2A	Intellectual disability, X-linked syndromic, Nascimento-type	Intellectual Disability
UPF3B	Intellectual disability, X-linked, syndromic 14	Intellectual Disability
USP9X	Intellectual disability, X-linked 99	Intellectual Disability
WDR13	candidate for X-linked intellectual disability	Intellectual Disability
ZDHHC15	Intellectual disability, X-linked 91	Intellectual Disability
ZDHHC9	Intellectual disability, X-linked syndromic, Raymond type	Intellectual Disability
ZMYND11	Intellectual disability, autosomal dominant 30	Intellectual Disability
ZNF41	X-linked non-syndromic intellectual disability	Intellectual Disability
ZNF674	X-linked non-syndromic intellectual disability	Intellectual Disability
ZNF711	Intellectual disability, X-linked 97	Intellectual Disability
ZNF81	Intellectual disability, X-linked 45	Intellectual Disability
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency	Intellectual Disability, Seizures, Neurologic Disease
AMT	Glycine encephalopathy	Intellectual Disability, Seizures, Neurologic Disease
ATP7A	Menkes disease	Intellectual Disability, Seizures, Neurologic Disease
CNKSR2	Undetermined early-onset epileptic encephalopathy	Intellectual Disability, Seizures, Neurologic Disease
GK	Glycerol kinase deficiency	Intellectual Disability, Seizures, Neurologic Disease
EHMT1	Kleefstra syndrome	Kleefstra syndrome
NDUFA1	Mitochondrial complex i deficiency	Mitochondrial Disease
AIFM1	Combined oxidative phosphorylation deficiency 6	Mitochondrial Rspiratory Chain Dysfunction
LAS1L	Spinal muscular atrophy with respiratory distress type 2	Motor Neuron Disease
IDS	Mucopolysaccharidosis type ii	Mucopolysaccharidosis
NDP	Exudative vitreoretinopathy 2, X-linked	Neurodegenerative Disorder
TIMM8A	Opticoacoustic nerve atrophy with dementia	Neurodegenerative Disorder
ABCD1	Adrenoleukodystrophy	Other
PGK1	Phosphoglycerate kinase 1 deficiency	Other
FOXP2	Speech-language disorder-1	Other, DD, ID, ASD related disorders
HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency; Intellectual	Other, DD, ID, ASD related disorders

Gene	Disease association	Category
	disability, X-linked syndromic 10	
NAA10	Microphthalmia, syndrome 1; N-terminal acetyltransferase deficiency	Other, DD, ID, ASD related disorders
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	Other, DD, ID, ASD related disorders
GJB1	Charcot-Marie-Tooth disease, X-linked dominant, 1	Peripheral Neuropathy
BRAF	Cardiofaciocutaneous syndrome; LEOPARD sydrome; Noonan syndrome	Ras/MAPK Pathway-related Disorder
CBL	Noonan syndrome-like disorder	Ras/MAPK Pathway-related Disorder
HRAS	Costello syndrome (Congenital myopathy with excess of muscle spindles)	Ras/MAPK Pathway-related Disorder
KRAS	Noonan syndrome; Cardiofaciocutaneous syndrome	Ras/MAPK Pathway-related Disorder
MAP2K1	Cardiofaciocutaneous syndrome	Ras/MAPK Pathway-related Disorder
MAP2K2	Cardiofaciocutaneous syndrome	Ras/MAPK Pathway-related Disorder
NF1	Neurofibramatosis-Noonan syndrome; Watson syndrome	Ras/MAPK Pathway-related Disorder
NRAS	Noonan syndrome	Ras/MAPK Pathway-related Disorder
PTPN11	LEOPARD syndrome; Noonan syndrome	Ras/MAPK Pathway-related Disorder
RAF1	Noonan syndrome; LEOPARD syndrome	Ras/MAPK Pathway-related Disorder
SHOC2	Noonan syndrome-like disorder	Ras/MAPK Pathway-related Disorder
SOS1	Noonan syndrome	Ras/MAPK Pathway-related Disorder
SPRED1	Legius syndrome	Ras/MAPK Pathway-related Disorder
CDKL5	Angleman Syndrome; epileptic encephalopathy, early infantile, 2	Rett/atypical Rett syndromes
FOXG1	Rett syndrome, congenital variant	Rett/atypical Rett syndromes
MBD5	Intellectual disability, autosomal dominant 1 (MRD1)	Rett/atypical Rett syndromes
MECP2	Encephalopathy, neonatal severe; Rett syndrome	Rett/atypical Rett syndromes
MEF2C	Rett/atypical Rett syndromes	Rett/atypical Rett syndromes
CREBBP	Rubinstein-Taybi syndrome	Rubinstein-Taybi syndrome
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency	Selected inborn errors of metabolism
HPRT1	Lesch-Nyhan syndrome	Selected inborn errors of metabolism
AP4B1	Spastic paraplegia 47, atuosomal recessive	Spastic paraplegia
AP4E1	Spastic paraplegia 51, atuosomal recessive	Spastic paraplegia
AP4M1	Spastic paraplegia 50, atuosomal recessive	Spastic paraplegia
AP4S1	Spastic paraplegia 52, atuosomal recessive	Spastic paraplegia
DDHD2	Spastic paraplegia 54, autosomal recessive	Spastic paraplegia
ERLIN2	Spastic paraplegia 18, autosomal recessive	Spastic paraplegia
PLP1	Pelizaeus-Merzbacher disease; Spastic paraplegia 2, X-linked	Spastic paraplegia
GPC3	Simpson-Golabi-Behmel syndrome, type 1	Syndromic Intellectual Disabiity
ADNP	Helsmoortel-van der Aa syndrome	Syndromic Intellectual Disability
ALG6	Congenital disorder of glycosylation type Ic	Syndromic Intellectual Disability
BCOR	Lenz Microphthalmia Syndrome; OCULOFACIOCARDIODENTAL SYNDROME (OFCD)	Syndromic Intellectual Disability
C12orf57	Temtamy Syndrome	Syndromic Intellectual Disability
CACNA1C	Timothy syndrome	Syndromic Intellectual Disability
CHD7	CHARGE syndrome	Syndromic Intellectual Disability
DHCR7	Smith-Lemli-Opitz syndrome	Syndromic Intellectual Disability
DKC1	Dyskeratosis congenita, X-linked	Syndromic Intellectual Disability
DMD	Becker muscular dystrophy; Duchenne muscular dystrophy	Syndromic Intellectual Disability
DNMT3A	Tatton-Brown-Rahman syndrome	Syndromic Intellectual Disability
DYNC1H1	AD intellectual disability 12; Charcot-Marie-Tooth disease, axonal, type 20; spinal muscular atrophy	Syndromic Intellectual Disability
EBP	MEND syndrome	Syndromic Intellectual Disability
FANCB	Fanconi anemia, complementation group b	Syndromic Intellectual Disability
FGD1	AARSKOG-SCOTT SYNDROME (faciogenital dysplasia) and X-linked Intellectual disability, syndromatic 16	Syndromic Intellectual Disability
FMR1* **	Fragile X syndrome and FMR1-related disorders	Syndromic Intellectual Disability
FOLR1	Neurodegeneration due to cerebral folate transport deficiency	Syndromic Intellectual Disability
HCCS	Microphthalmia, syndromic 7	Syndromic Intellectual Disability
HOXA1	Athabaskan brainstem dysgenesis syndrome; Bosley-Salih-Alorainy syndrome	Syndromic Intellectual Disability
KCNJ10	SESAME syndrome	Syndromic Intellectual Disability

Gene	Disease association	Category
KMT2D	Kabuki Syndrome	Syndromic Intellectual Disability
KMTD2	Kabuki syndrome	Syndromic Intellectual Disability
MAOA	Brunner syndrome	Syndromic Intellectual Disability
MBTPS2	IFAP syndrome with or without Bresheck syndrome	Syndromic Intellectual Disability
MED12	Lujan-Fryns syndrome; Ohdo syndrome, X-linked; Opitz-Kaveggia syndrome	Syndromic Intellectual Disability
MID1	Opitz GBBB syndrome, type I	Syndromic Intellectual Disability
NHS	Nance-Horan syndrome	Syndromic Intellectual Disability
NSD1	Beckwith-Wiedemann syndrome; Sotos syndrome 1	Syndromic Intellectual Disability
NSDHL	CHILD syndrome; CK syndrome	Syndromic Intellectual Disability
OCRL	Dent disease 2; Lowe syndrome	Syndromic Intellectual Disability
OFD1	Joubert syndrome 10; Oral-facial-digital syndrome 1; Simpson-Golabi-Behmel syndrome, type 2	Syndromic Intellectual Disability
PDHA1	Leigh syndrome, X-linked; Pyruvate dehydrogenase E1-alpha deficiency	Syndromic Intellectual Disability
PHF6	Borjeson-Forssman-Lehmann syndrome	Syndromic Intellectual Disability
PORCN	Focal dermal hypoplasia	Syndromic Intellectual Disability
PQBP1	Renpenning syndrome	Syndromic Intellectual Disability
PRPS1	Arts syndrome; Charcot-Marie-Tooth disease, X-linked recessive, 5	Syndromic Intellectual Disability
PTEN	Bannayan-Riley-Ruvalcaba syndrome; Cowden syndrome 1; Macrocephaly/autism syndrome	Syndromic Intellectual Disability
RAB40AL	Martin-Probst X-linked deafness-intellectual disability syndrome	Syndromic Intellectual Disability
RAI1	Smith-Magenis syndrome	Syndromic Intellectual Disability
RBM10	Tarp syndrome	Syndromic Intellectual Disability
RPS6KA3	Coffin-Lowry syndrome; Intellectual disability, X-linked 19	Syndromic Intellectual Disability
SETD2	Luscan-Lumish syndrome	Syndromic Intellectual Disability
SHANK3	Phelan-McDermid syndrome	Syndromic Intellectual Disability
SLC16A2	Allan-Herndon-Dudley syndrome	syndromic Intellectual Disability
SLC2A1	Epilepsy, idiopathic generalized, suscpetibility to, 12; GLUT1 deficiency syndrome 1; GLUT1 deficiency syndrome 2	Syndromic Intellectual Disability
SLC6A8	Cerebral creatine deficiency syndrome 1	Syndromic Intellectual Disability
SMARCB1	Intellectual disability, autosomal dominant 15	Syndromic Intellectual Disability
SOX11	Coffin-siris syndrome	Syndromic Intellectual Disability
TBL1XR1	Pierpont syndrome	Syndromic Intellectual Disability
TBX1	DiGeorge syndrome; Tetralogy of Fallot; Velocardiofacial syndrome	Syndromic Intellectual Disability
VPS13B	Cohen syndrome	Syndromic Intellectual Disability
ZC4H2	Wieacker-Wolff syndrome	Syndromic Intellectual Disability
ZEB2	Mowat-Wilson syndrome	Syndromic Intellectual Disability
TSC1	Tuberous sclerosis-1	Tuberous sclerosis
TSC2	Tuberous sclerosis-2	Tuberous sclerosis