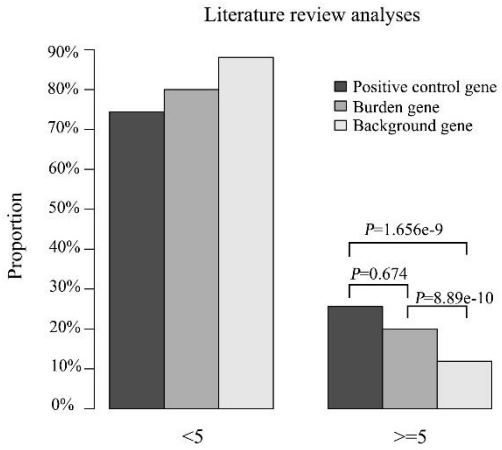
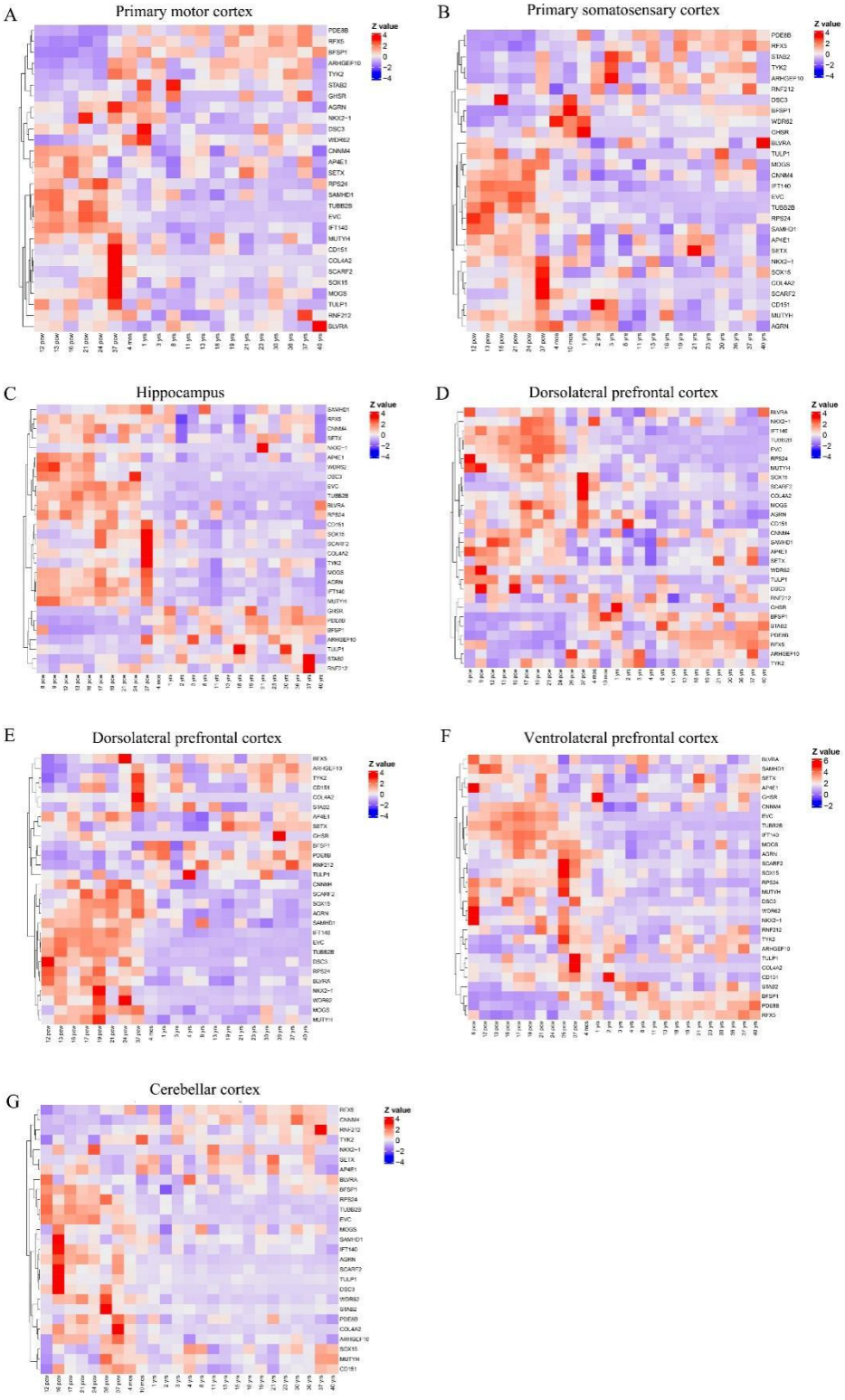


Supplementary Figure 1: Literature review of positive control gene, burden gene, and background gene. We calculated the percentage of genes with more than 5 brain related studies in each group (Positive control gene, burden gene, and background gene). The results of Fisher exact test were presented.



Supplementary Figure 2: Expression map of burden genes during brain development in different brain regions. The relative expression level of burden genes in motor related brain regions in different human development stage (GW12 to 40 years old).

Supplementary Table 1: Types of VEP/ANNOVAR annotated variants and classification.

Mutation type of ANNOVAR annotated variant	Mutation type of VEP annotated variant	Impact	Score	Mutation Type Group
splicing	splice acceptor variant	High	2	PTV
	splice donor variant	High	2	PTV
stopgain	stop gained	High	2	PTV
frameshift insertion		High	2	PTV
frameshift deletion	frameshift variant	High	2	PTV
frameshift block substitution		High	2	PTV
	start lost	High	2	PTV
stoploss	stop lost	High	2	PTV
nonsynonymous SNV	missense variant	Moderate	1	MIS
nonframeshift deletion	inframe deletion	Moderate	1	MIS
nonframeshift insertion	inframe insertion	Moderate	1	MIS
synonymous SNV	synonymous variant	Low	0	SYN
	splice region variant	Low	0	NON
	start retained variant	Low	0	NON
intronic	stop retained variant	Low	0	NON
	intron variant	Modifier	0	NON
ncRNA_exonic	non coding transcript exon variant	Modifier	0	NON
UTR3	3 prime UTR variant	Modifier	0	NON
UTR5	5 prime UTR variant	Modifier	0	NON
upstream	upstream_gene_variant	Modifier	0	NON
downstream	downstream_gene_variant	Modifier	0	NON
intergenic	intergenic_variant	Modifier	0	NON

Supplementary Table 2: Neurodevelopment and genetic characteristics of NDD patients.

Features	Patients without WBDV (*n* = 213)	Patients with WBDV (*n* = 40)	*P* value
Motor development delay	106 (49.8)	30 (75)	0.003*
Speech/language development delay	150 (70.4)	28 (70)	1.000
Intellectual disability	17 (8.0)	7 (18)	0.076

Data are presented as n (%).NDD: Neurodevelopmental delay; WBDV: Whole-brain deviation volume.

Supplementary Table 3: Genetic burden test results.

Gene	Fisher_PTVp	Fisher_MISp
COL4A2	0.0183909	0.180291
MOGS	0.0183909	0.65079
SCARF2	0.0183909	1
RFX5	0.1376147	0.013929
MUTYH	0.6410066	0.0484
PDE8B	1	0.005474
SAMHD1	1	0.008638
EVC	1	0.013929
WDR62	1	0.013929
STAB2	1	0.013994
BFSP1	1	0.018391
BLVRA	1	0.018391
BPNT2	1	0.018391
SOX15	1	0.018391
TULP1	1	0.018391
ARHGEF10	1	0.019568
CD151	1	0.019568
DSC3	1	0.019568
NKX2-1	1	0.019568
TUBB2B	1	0.022645
AGRN	1	0.027323
IFT140	1	0.029458
KMT2D	1	0.029853
CNNM4	1	0.035474
GHSR	1	0.035474
RNF212	1	0.035474
SETX	1	0.045466
AP4E1	1	0.0484

TYK2	1	0.0484
RPS24	1	0.049261

Supplementary Table 4: Micro-macro gene list.

Gene	OMIM disease	Micro/Macro
ABCC9	Cantu syndrome	macrocephaly risk gene
ACAN	Spondyloepimetaphyseal dysplasia, aggrecan type	macrocephaly risk gene
ADK	Hypermethioninemia due to adenosine kinase deficiency	macrocephaly risk gene
AKT1	Proteus syndrome	macrocephaly risk gene
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2	macrocephaly risk gene
AMER1	Osteopathia striata with cranial sclerosis	macrocephaly risk gene
ANKH	Craniometaphyseal dysplasia, autosomal dominant	macrocephaly risk gene
APC2	Sotos syndrome 3	macrocephaly risk gene
ARSB	Mucopolysaccharidosis, type vi	macrocephaly risk gene
ASXL2	Shashi-pena syndrome	macrocephaly risk gene
B4GALT1	Congenital disorder of glycosylation, type iid	macrocephaly risk gene
B4GALT7	Ehlers-danlos syndrome, spondylodysplastic type, 1	macrocephaly risk gene
BGN	Meester-loeys syndrome	macrocephaly risk gene
BRAF	Cardiofaciocutaneous syndrome 1	macrocephaly risk gene
BRWD3	Mental retardation, x-linked 93	macrocephaly risk gene
CCND2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3	macrocephaly risk gene
CDKN1C	Intrauterine growth retardation, metaphyseal dysplasia, adrenal hypoplasia congenita, and genital anomalies	macrocephaly risk gene
CEP120	Short-rib thoracic dysplasia 13 with or without polydactyly	macrocephaly risk gene
CHD1	Pilarowski-bjornsson syndrome	macrocephaly risk gene
CHD4	Sifrim-hitz-weiss syndrome	macrocephaly risk gene
CHD8	Autism, susceptibility to, 18	macrocephaly risk gene
COL2A1	Platyspondylic lethal skeletal dysplasia, torrance type	macrocephaly risk gene
CUL4B	Mental retardation, x-linked, syndromic, cabezas type	macrocephaly risk gene
CWC27	Retinitis pigmentosa with or without skeletal anomalies	macrocephaly risk gene
D2HGDH	D-2-hydroxyglutaric aciduria 1	macrocephaly risk gene
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 9	macrocephaly risk gene
DVL1	Robinow syndrome, autosomal dominant 2	macrocephaly risk gene
DVL3	Robinow syndrome, autosomal dominant 3	macrocephaly risk gene
EED	Cohen-gibson syndrome	macrocephaly risk gene
EIF2B1	Leukoencephalopathy with vanishing white matter	macrocephaly risk gene
EIF2B2	Leukoencephalopathy with vanishing white matter	macrocephaly risk gene
EIF2B3	Leukoencephalopathy with vanishing white matter	macrocephaly risk gene
EIF2B4	Leukoencephalopathy with vanishing white matter	macrocephaly risk gene
EIF2B5	Leukoencephalopathy with vanishing white matter	macrocephaly risk gene
EML1	Band heterotopia	macrocephaly risk gene
ERF	Craniosynostosis 4	macrocephaly risk gene
ETFA	Multiple acyl-coa dehydrogenase deficiency	macrocephaly risk gene

ETFB	Multiple acyl-coa dehydrogenase deficiency	macrocephaly risk gene
ETFDH	Multiple acyl-coa dehydrogenase deficiency	macrocephaly risk gene
EZH2	Weaver syndrome	macrocephaly risk gene
FAM111A	Kenny-caffey syndrome, type 2	macrocephaly risk gene
FH	Fumarase deficiency	macrocephaly risk gene
FIBP	Thauvin-robinet-faivre syndrome	macrocephaly risk gene
FMR1	Fragile x syndrome	macrocephaly risk gene
GCDH	Glutaric acidemia i	macrocephaly risk gene
GFAP	Alexander disease	macrocephaly risk gene
GLI3	Greig cephalopolysyndactyly syndrome	macrocephaly risk gene
GNAI3	Auriculocondylar syndrome 1	macrocephaly risk gene
GNAQ	Sturge-weber syndrome	macrocephaly risk gene
GPC3	Simpson-golabi-behmel syndrome, type 1	macrocephaly risk gene
GPC4	Keipert syndrome	macrocephaly risk gene
GRIA3	Intellectual developmental disorder, x-linked, syndromic, wu type	macrocephaly risk gene
GUSB	Mucopolysaccharidosis, type vii	macrocephaly risk gene
HDAC6	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia	macrocephaly risk gene
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2a	macrocephaly risk gene
HERC1	Macrocephaly, dysmorphic facies, and psychomotor retardation	macrocephaly risk gene
HEXB	Sandhoff disease	macrocephaly risk gene
HRAS	Costello syndrome	macrocephaly risk gene
HSD17B4	D-bifunctional protein deficiency	macrocephaly risk gene
IDS	Mucopolysaccharidosis, type ii	macrocephaly risk gene
IDUA	Hurler syndrome	macrocephaly risk gene
IFT43	Cranioectodermal dysplasia 3	macrocephaly risk gene
IFT81	Short-rib thoracic dysplasia 19 with or without polydactyly	macrocephaly risk gene
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma, and micrognathia	macrocephaly risk gene
IGF2	Silver-russell syndrome 3	macrocephaly risk gene
INPP5E	Joubert syndrome 1	macrocephaly risk gene
INPPL1	Opsismodysplasia	macrocephaly risk gene
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 7	macrocephaly risk gene
ITCH	Autoimmune disease, multisystem, with facial dysmorphism	macrocephaly risk gene
KDM1A	Cleft palate, psychomotor retardation, and distinctive facial features	macrocephaly risk gene
KIF7	Acrocallosal syndrome	macrocephaly risk gene
KPTN	Mental retardation, autosomal recessive 41	macrocephaly risk gene
KRAS	Oculoectodermal syndrome	macrocephaly risk gene
LAMB1	Lissencephaly 5	macrocephaly risk gene
LBR	Greenberg dysplasia	macrocephaly risk gene
LRP2	Donnai-barrow syndrome	macrocephaly risk gene
MAB21L2	Microphthalmia/coloboma and skeletal dysplasia syndrome	macrocephaly risk gene
MAN2B1	Mannosidosis, alpha b, lysosomal	macrocephaly risk gene

MAP2K2	Cardiofaciocutaneous syndrome 4	macrocephaly risk gene
MED12	Opitz-kaveggia syndrome	macrocephaly risk gene
MITF	Coloboma, osteopetrosis, microphthalmia, macrocephaly, albinism, and deafness	macrocephaly risk gene
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts 1	macrocephaly risk gene
MPDZ	Hydrocephalus, congenital, 2, with or without brain or eye anomalies	macrocephaly risk gene
MSL3	Basilicata-akhtar syndrome	macrocephaly risk gene
MTOR	Smith-kingsmore syndrome	macrocephaly risk gene
MYH8	Arthrogryposis, distal, type 7	macrocephaly risk gene
NF1	Watson syndrome	macrocephaly risk gene
NFIA	Brain malformations with or without urinary tract defects	macrocephaly risk gene
NFIB	Macrocephaly, acquired, with impaired intellectual development	macrocephaly risk gene
NFIX	Sotos syndrome 2	macrocephaly risk gene
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia	macrocephaly risk gene
NONO	Mental retardation, x-linked, syndromic 34	macrocephaly risk gene
NRAS	Noonan syndrome 6	macrocephaly risk gene
NSD1	Sotos syndrome 1	macrocephaly risk gene
NXN	Robinow syndrome, autosomal recessive 2	macrocephaly risk gene
OBSL1	Three M syndrome 2	macrocephaly risk gene
OPHN1	Mental retardation, x-linked, with cerebellar hypoplasia and distinctive facial appearance	macrocephaly risk gene
PAK1	Intellectual developmental disorder with macrocephaly, seizures, and speech delay	macrocephaly risk gene
PAM16	Spondylometaphyseal dysplasia, megarbane-dagher-melki type	macrocephaly risk gene
PDSS1	Coenzyme q10 deficiency, primary, 2	macrocephaly risk gene
PEX1	Peroxisome biogenesis disorder 1a (zellweger)	macrocephaly risk gene
PIGM	Glycosylphosphatidylinositol biosynthesis defect 1	macrocephaly risk gene
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1	macrocephaly risk gene
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3	macrocephaly risk gene
PIK3CA	Megalencephaly-capillary malformation-polymicrogyria syndrome	macrocephaly risk gene
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1	macrocephaly risk gene
PKDCC	Rhizomelic limb shortening with dysmorphic features	macrocephaly risk gene
PLCB4	Auriculocondylar syndrome 2	macrocephaly risk gene
PLG	Plasminogen deficiency, type i	macrocephaly risk gene
POLR3A	Wiedemann-rautenstrauch syndrome	macrocephaly risk gene
POP1	Anauxetic dysplasia 2	macrocephaly risk gene
PPP1CB	Noonan syndrome-like disorder with loose anagen hair 2	macrocephaly risk gene
PPP2R5D	Mental retardation, autosomal dominant 35	macrocephaly risk gene
PTCH1	Holoprosencephaly 7	macrocephaly risk gene
PTEN	Macrocephaly/autism syndrome	macrocephaly risk gene
RAB39B	Waisman syndrome	macrocephaly risk gene
RAF1	Noonan syndrome 5	macrocephaly risk gene
RIT1	Noonan syndrome 8	macrocephaly risk gene

RNF125	Tenorio syndrome	macrocephaly risk gene
ROR2	Robinow syndrome, autosomal recessive 1	macrocephaly risk gene
RRAS2	Noonan syndrome 12	macrocephaly risk gene
SEC23A	Craniolenticulosutural dysplasia	macrocephaly risk gene
SEC23B	Cowden syndrome 7	macrocephaly risk gene
SEC24D	Cole-carpenter syndrome 2	macrocephaly risk gene
SERPINH1	Osteogenesis imperfecta, type x	macrocephaly risk gene
SETD2	Luscan-lumish syndrome	macrocephaly risk gene
SHANK3	Phelan-mcdermid syndrome	macrocephaly risk gene
SHOC2	Noonan syndrome-like disorder with loose anagen hair 1	macrocephaly risk gene
SNX10	Osteopetrosis, autosomal recessive 8	macrocephaly risk gene
SNX14	Spinocerebellar ataxia, autosomal recessive 20	macrocephaly risk gene
SOS1	Noonan syndrome 4	macrocephaly risk gene
SOST	Craniodiaphyseal dysplasia, autosomal dominant	macrocephaly risk gene
SOX9	Campomelic dysplasia	macrocephaly risk gene
SPINT2	Diarrhea 3, secretory sodium, congenital, with or without other congenital anomalies	macrocephaly risk gene
SPRED1	Legius syndrome	macrocephaly risk gene
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy	macrocephaly risk gene
SUZ12	Imagawa-matsumoto syndrome	macrocephaly risk gene
SYN1	Epilepsy, x-linked, with variable learning disabilities and behavior disorders	macrocephaly risk gene
TBC1D7	Macrocephaly/megalencephaly syndrome, autosomal recessive	macrocephaly risk gene
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	macrocephaly risk gene
TBL1X	Hypothyroidism, congenital, nongoitrous, 8	macrocephaly risk gene
TCIRG1	Osteopetrosis, autosomal recessive 1	macrocephaly risk gene
THRA	Hypothyroidism, congenital, nongoitrous, 6	macrocephaly risk gene
TMEM216	Joubert syndrome 2	macrocephaly risk gene
TONSL	Spondyloepimetaphyseal dysplasia, sponastrime type	macrocephaly risk gene
TRIP11	Odontochondrodysplasia	macrocephaly risk gene
TRIP12	Clark-baraitser syndrome	macrocephaly risk gene
UPF3B	Mental retardation, x-linked, syndromic 14	macrocephaly risk gene
WDR60	Short-rib thoracic dysplasia 8 with or without polydactyly	macrocephaly risk gene
WNT5A	Robinow syndrome, autosomal dominant 1	macrocephaly risk gene
ZBTB20	Primrose syndrome	macrocephaly risk gene
ZBTB42	Lethal congenital contracture syndrome 6	macrocephaly risk gene
ZDHHC9	Intellectual developmental disorder, x-linked, syndromic, raymond type	macrocephaly risk gene
ZNF469	Brittle cornea syndrome 1	macrocephaly risk gene
AAAS	Achalasia-addisonianism-alacrimia syndrome	microcephaly risk gene
AARS1	Epileptic encephalopathy, early infantile, 29	microcephaly risk gene
ABCA2	Intellectual developmental disorder with poor growth and with or without seizures or ataxia	microcephaly risk gene

ACADSB	2-methylbutyrylglycinuria	microcephaly risk gene
ACBD5	Retinal dystrophy with leukodystrophy,	microcephaly risk gene
ACE	Renal tubular dysgenesis	microcephaly risk gene
ACO2	Infantile cerebellar-retinal degeneration	microcephaly risk gene
ACSL4	Mental retardation, X-linked 63	microcephaly risk gene
ACTB	Baraitser-Winter syndrome 1	microcephaly risk gene
ACTL6B	Intellectual developmental disorder with severe speech and ambulation defects	microcephaly risk gene
ADAR	Dyschromatosis symmetrica hereditaria	microcephaly risk gene
ADARB1	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures	microcephaly risk gene
ADAT3	Mental retardation, autosomal recessive 36	microcephaly risk gene
ADD3	Cerebral palsy, spastic quadriplegic, 3	microcephaly risk gene
ADSL	Adenylosuccinase deficiency	microcephaly risk gene
AGA	Aspartylglucosaminuria	microcephaly risk gene
AGT	Renal tubular dysgenesis	microcephaly risk gene
AGTPBP1	Neurodegeneration, childhood-onset, with cerebellar atrophy	microcephaly risk gene
AGTR1	Renal tubular dysgenesis	microcephaly risk gene
AIMP1	Leukodystrophy, hypomyelinating, 3	microcephaly risk gene
AIMP2	Leukodystrophy, hypomyelinating, 17	microcephaly risk gene
ALDH18A1	Cutis laxa, autosomal recessive, type IIIA	microcephaly risk gene
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency	microcephaly risk gene
ALG1	Congenital disorder of glycosylation, type Ik	microcephaly risk gene
ALG12	Congenital disorder of glycosylation, type Ig	microcephaly risk gene
ALG13	Epileptic encephalopathy, early infantile, 36	microcephaly risk gene
ALG3	Congenital disorder of glycosylation, type Id	microcephaly risk gene
ALG9	Gillessen-Kaesbach-Nishimura syndrome	microcephaly risk gene
AMPD2	Pontocerebellar hypoplasia, type 9	microcephaly risk gene
ANKLE2	Microcephaly 16, primary, autosomal recessive	microcephaly risk gene
ANKRD11	KBG syndrome	microcephaly risk gene
AP3B1	Hermansky-Pudlak syndrome 2	microcephaly risk gene
AP3B2	Epileptic encephalopathy, early infantile, 48	microcephaly risk gene
AP4B1	Spastic paraplegia 47, autosomal recessive	microcephaly risk gene
AP4E1	Stuttering, familial persistent, 1	microcephaly risk gene
AP4M1	Spastic paraplegia 50, autosomal recessive	microcephaly risk gene
AP4S1	Spastic paraplegia 52, autosomal recessive	microcephaly risk gene
ARCN1	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay	microcephaly risk gene
ARF1	Periventricular nodular heterotopia 8	microcephaly risk gene
ARFGEF2	Periventricular heterotopia with microcephaly	microcephaly risk gene
ARHGAP31	Adams-Oliver syndrome 1	microcephaly risk gene
ARSL	Chondrodysplasia punctata, X-linked recessive	microcephaly risk gene
ARV1	Epileptic encephalopathy, early infantile, 38	microcephaly risk gene
ARX	Epileptic encephalopathy, early infantile, 1	microcephaly risk gene

ASNS	Asparagine synthetase deficiency	microcephaly risk gene
ASPM	Microcephaly 5, primary, autosomal recessive	microcephaly risk gene
ASXL1	Bohring-Opitz syndrome	microcephaly risk gene
ASXL3	Bainbridge-Ropers syndrome	microcephaly risk gene
ATP6V0A2	Wrinkly skin syndrome	microcephaly risk gene
ATP6V1A	Epileptic encephalopathy, infantile or early childhood, 3	microcephaly risk gene
ATP7A	Occipital horn syndrome	microcephaly risk gene
ATR	Seckel syndrome 1	microcephaly risk gene
ATRX	Alpha-thalassemia/mental retardation syndrome	microcephaly risk gene
AUTS2	Mental retardation, autosomal dominant 26	microcephaly risk gene
BCAP31	Deafness, dystonia, and cerebral hypomyelination	microcephaly risk gene
BCL11A	Dias-Logan syndrome	microcephaly risk gene
BCOR	Microphthalmia, syndromic 2	microcephaly risk gene
BLM	Bloom syndrome	microcephaly risk gene
BPTF	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies	microcephaly risk gene
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal	microcephaly risk gene
BRCA1	Fanconi anemia, complementation group S	microcephaly risk gene
BRCA2	Fanconi anemia, complementation group D1	microcephaly risk gene
BRF1	Cerebellofaciodental syndrome	microcephaly risk gene
BRPF1	Intellectual developmental disorder with dysmorphic facies and ptosis	microcephaly risk gene
BUB1B	Mosaic variegated aneuploidy syndrome 1	microcephaly risk gene
C2CD3	Orofaciodigital syndrome XIV	microcephaly risk gene
CACNA1B	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements	microcephaly risk gene
CACNA1G	Spinocerebellar ataxia 42	microcephaly risk gene
CARS1	Microcephaly, developmental delay, and brittle hair syndrome	microcephaly risk gene
CARS2	Combined oxidative phosphorylation deficiency 27	microcephaly risk gene
CASK	Mental retardation, with or without nystagmus	microcephaly risk gene
CCDC47	Trichohepatoneurodevelopmental syndrome	microcephaly risk gene
CD96	C syndrome	microcephaly risk gene
CDC42	Takenouchi-Kosaki syndrome	microcephaly risk gene
CDC45	Meier-Gorlin syndrome 7	microcephaly risk gene
CDK13	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder	microcephaly risk gene
CDK5RAP2	Microcephaly 3, primary, autosomal recessive	microcephaly risk gene
CDKL5	Epileptic encephalopathy, early infantile, 2	microcephaly risk gene
CDT1	Meier-Gorlin syndrome 4	microcephaly risk gene
CENPF	Stromme syndrome	microcephaly risk gene
CENPJ	Microcephaly 6, primary, autosomal recessive	microcephaly risk gene
CEP135	Microcephaly 8, primary, autosomal recessive	microcephaly risk gene
CEP152	Microcephaly 9, primary, autosomal recessive	microcephaly risk gene
CEP290	Leber congenital amaurosis 10	microcephaly risk gene
CEP57	Mosaic variegated aneuploidy syndrome 2	microcephaly risk gene

CERT1	Mental retardation, autosomal dominant 34	microcephaly risk gene
CHAMP1	Mental retardation, autosomal dominant 40	microcephaly risk gene
CHD7	Hypogonadotropic hypogonadism 5 with or without anosmia	microcephaly risk gene
CHKB	Muscular dystrophy, congenital, megaconial type	microcephaly risk gene
CHMP1A	Pontocerebellar hypoplasia, type 8	microcephaly risk gene
CIT	Microcephaly 17, primary, autosomal recessive	microcephaly risk gene
CKAP2L	Filippi syndrome	microcephaly risk gene
CLP1	Pontocerebellar hypoplasia, type 10	microcephaly risk gene
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia	microcephaly risk gene
CLPP	Perrault syndrome 3	microcephaly risk gene
CLTC	Mental retardation, autosomal dominant 56	microcephaly risk gene
CNNM2	Hypomagnesemia 6, renal	microcephaly risk gene
CNOT1	Holoprosencephaly 12, with or without pancreatic agenesis	microcephaly risk gene
CNTNAP1	Lethal congenital contracture syndrome 7	microcephaly risk gene
COASY	Neurodegeneration with brain iron accumulation 6	microcephaly risk gene
COG1	Congenital disorder of glycosylation, type Iig	microcephaly risk gene
COG4	Saul-Wilson syndrome	microcephaly risk gene
COG5	Congenital disorder of glycosylation, type Iii	microcephaly risk gene
COG6	Congenital disorder of glycosylation, type Iil	microcephaly risk gene
COG7	Congenital disorder of glycosylation, type Iie	microcephaly risk gene
COQ9	Coenzyme Q10 deficiency, primary, 5	microcephaly risk gene
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2,	microcephaly risk gene
COX7B	Linear skin defects with multiple congenital anomalies 2,	microcephaly risk gene
CPT2	CPT II deficiency, myopathic, stress-induced,	microcephaly risk gene
CREBBP	Rubinstein-Taybi syndrome 1,	microcephaly risk gene
CRIPT	Short stature with microcephaly and distinctive facies,	microcephaly risk gene
CSNK2A1	Okur-Chung neurodevelopmental syndrome,	microcephaly risk gene
CTCF	Mental retardation, autosomal dominant 21,	microcephaly risk gene
CTNNA2	Cortical dysplasia, complex, with other brain malformations 9,	microcephaly risk gene
CTNNB1	Neurodevelopmental disorder with spastic diplegia and visual defects,	microcephaly risk gene
CTSD	Ceroid lipofuscinosis, neuronal, 10,	microcephaly risk gene
CTU2	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome,	microcephaly risk gene
CYB5R3	Methemoglobinemia, type I,	microcephaly risk gene
CYFIP2	Epileptic encephalopathy, early infantile, 65,	microcephaly risk gene
DCPS	Al-Raqad syndrome,	microcephaly risk gene
DDX11	Warsaw breakage syndrome,	microcephaly risk gene
DEAF1	Vulto-van Silfout-de Vries syndrome,	microcephaly risk gene
DEGS1	Leukodystrophy, hypomyelinating, 18,	microcephaly risk gene
DENND5A	Epileptic encephalopathy, early infantile, 49,	microcephaly risk gene
DGUOK	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4,	microcephaly risk gene

DHCR7	Smith-Lemli-Opitz syndrome,	microcephaly risk gene
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency,	microcephaly risk gene
DHTKD1	2-aminoadipic 2-oxoadipic aciduria,	microcephaly risk gene
DHX37	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies,	microcephaly risk gene
DIAPH1	Seizures, cortical blindness, microcephaly syndrome,	microcephaly risk gene
DKC1	Dyskeratosis congenita, X-linked,	microcephaly risk gene
DLAT	Pyruvate dehydrogenase E2 deficiency,	microcephaly risk gene
DLD	Dihydrolipoamide dehydrogenase deficiency,	microcephaly risk gene
DMXL2	Epileptic encephalopathy, early infantile, 81,	microcephaly risk gene
DNAJC21	Bone marrow failure syndrome 3,	microcephaly risk gene
DNM1L	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1,	microcephaly risk gene
DNMT3A	Heyn-Sproul-Jackson syndrome,	microcephaly risk gene
DOLK	Congenital disorder of glycosylation, type Im,	microcephaly risk gene
DONSON	Microcephaly-micromelia syndrome,	microcephaly risk gene
DPAGT1	Congenital disorder of glycosylation, type Ij,	microcephaly risk gene
DPM1	Congenital disorder of glycosylation, type Ie,	microcephaly risk gene
DPM2	Congenital disorder of glycosylation, type Iu,	microcephaly risk gene
DPYD	Dihydropyrimidine dehydrogenase deficiency,	microcephaly risk gene
DSTYK	Congenital anomalies of kidney and urinary tract 1,	microcephaly risk gene
DYM	Smith-McCort dysplasia,	microcephaly risk gene
DYNC1H1	Mental retardation, autosomal dominant 13,	microcephaly risk gene
DYNC1I2	Neurodevelopmental disorder with microcephaly and structural brain anomalies,	microcephaly risk gene
DYRK1A	Mental retardation, autosomal dominant 7,	microcephaly risk gene
EBF3	Hypotonia, ataxia, and delayed development syndrome,	microcephaly risk gene
EEF1A2	Epileptic encephalopathy, early infantile, 33,	microcephaly risk gene
EFEMP2	Cutis laxa, autosomal recessive, type IB,	microcephaly risk gene
EFL1	Shwachman-Diamond syndrome 2,	microcephaly risk gene
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type,	microcephaly risk gene
EHMT1	Kleefstra syndrome 1,	microcephaly risk gene
EIF2AK3	Wolcott-Rallison syndrome,	microcephaly risk gene
EIF2S3	MEHMO syndrome,	microcephaly risk gene
ELAC2	Combined oxidative phosphorylation deficiency 17,	microcephaly risk gene
ELOVL4	Spinocerebellar ataxia 34,	microcephaly risk gene
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation,	microcephaly risk gene
EMG1	Bowen-Conradi syndrome,	microcephaly risk gene
ENTPD1	Spastic paraplegia 64, autosomal recessive,	microcephaly risk gene
EP300	Rubinstein-Taybi syndrome 2,	microcephaly risk gene
EPG5	Vici syndrome,	microcephaly risk gene
EPRS1	Leukodystrophy, hypomyelinating, 15,	microcephaly risk gene
ERCC1	Cerebrooculofacioskeletal syndrome 4,	microcephaly risk gene
ERCC2	Trichothiodystrophy 1, photosensitive,	microcephaly risk gene

ERCC3	Xeroderma pigmentosum, group B,	microcephaly risk gene
ERCC4	Xeroderma pigmentosum, type F/Cockayne syndrome,	microcephaly risk gene
ERCC5	Xeroderma pigmentosum, group G/Cockayne syndrome,	microcephaly risk gene
ERCC6	Cerebrooculofacioskeletal syndrome 1,	microcephaly risk gene
ERCC6L2	Bone marrow failure syndrome 2,	microcephaly risk gene
ERCC8	Cockayne syndrome, type A,	microcephaly risk gene
ESCO2	Roberts syndrome,	microcephaly risk gene
EXOSC3	Pontocerebellar hypoplasia, type 1B,	microcephaly risk gene
EXOSC9	Pontocerebellar hypoplasia, type 1D,	microcephaly risk gene
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities,	microcephaly risk gene
FAM20C	Raine syndrome,	microcephaly risk gene
FANCA	Fanconi anemia, complementation group A,	microcephaly risk gene
FANCC	Fanconi anemia, complementation group C,	microcephaly risk gene
FANCD2	Fanconi anemia, complementation group D2,	microcephaly risk gene
FANCE	Fanconi anemia, complementation group E,	microcephaly risk gene
FANCI	Fanconi anemia, complementation group I,	microcephaly risk gene
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder,	microcephaly risk gene
FARS2	Spastic paraplegia 77, autosomal recessive,	microcephaly risk gene
FARSB	Rajab interstitial lung disease with brain calcifications,	microcephaly risk gene
FBLN5	Macular degeneration, age-related, 3,	microcephaly risk gene
FBXL3	Intellectual developmental disorder with short stature, facial anomalies, and speech defects,	microcephaly risk gene
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type),	microcephaly risk gene
FGF12	Epileptic encephalopathy, early infantile, 47,	microcephaly risk gene
FGFR1	Pfeiffer syndrome,	microcephaly risk gene
FIG4	Yunis-Varon syndrome,	microcephaly risk gene
FKRP	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5,	microcephaly risk gene
FLVCR2	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome,	microcephaly risk gene
FOXG1	Rett syndrome, congenital variant,	microcephaly risk gene
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19,	microcephaly risk gene
FRAS1	Fraser syndrome 1,	microcephaly risk gene
FREM1	Manitoba oculotrichoanal syndrome,	microcephaly risk gene
FTO	Growth retardation, developmental delay, facial dysmorphism,	microcephaly risk gene
FUT8	Congenital disorder of glycosylation with defective fucosylation 1,	microcephaly risk gene
G6PC3	Dursun syndrome,	microcephaly risk gene
GABBR2	Neurodevelopmental disorder with poor language and loss of hand skills,	microcephaly risk gene
GABRA2	Epileptic encephalopathy, early infantile, 78,	microcephaly risk gene
GABRA5	Epileptic encephalopathy, early infantile, 79,	microcephaly risk gene
GABRB2	Epileptic encephalopathy, infantile or early childhood, 2,	microcephaly risk gene
GALNT2	Congenital disorder of glycosylation, type IIt,	microcephaly risk gene
GATA6	Pancreatic agenesis and congenital heart defects,	microcephaly risk gene

GBA	Gaucher disease, type III,	microcephaly risk gene
GEMIN4	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities,	microcephaly risk gene
GFM1	Combined oxidative phosphorylation deficiency 1,	microcephaly risk gene
GFM2	Combined oxidative phosphorylation deficiency 39,	microcephaly risk gene
GINS1	Immunodeficiency 55,	microcephaly risk gene
GLE1	Congenital arthrogryposis with anterior horn cell disease,	microcephaly risk gene
GLI2	Culler-Jones syndrome,	microcephaly risk gene
GLYCTK	D-glyceric aciduria,	microcephaly risk gene
GMNN	Meier-Gorlin syndrome 6,	microcephaly risk gene
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14,	microcephaly risk gene
GNAO1	Epileptic encephalopathy, early infantile, 17,	microcephaly risk gene
GNPAT	Rhizomelic chondrodysplasia punctata, type 2,	microcephaly risk gene
GORAB	Geroderma osteodysplasticum,	microcephaly risk gene
GOT2	Epileptic encephalopathy, early infantile, 82,	microcephaly risk gene
GPT2	Mental retardation, autosomal recessive 49,	microcephaly risk gene
GRIA4	Neurodevelopmental disorder with or without seizures and gait abnormalities,	microcephaly risk gene
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive,	microcephaly risk gene
GRIN2B	Epileptic encephalopathy, early infantile, 27,	microcephaly risk gene
GRIN2D	Epileptic encephalopathy, early infantile, 46,	microcephaly risk gene
GRM7	Neurodevelopmental disorder with seizures, hypotonia, and brain imaging abnormalities,	microcephaly risk gene
GSX2	Diencephalic-mesencephalic junction dysplasia syndrome 2,	microcephaly risk gene
GTF2E2	Trichothiodystrophy 6, nonphotosensitive,	microcephaly risk gene
GTPBP2	Jaberi-Elahi syndrome,	microcephaly risk gene
HAAO	Vertebral, cardiac, renal, and limb defects syndrome 1,	microcephaly risk gene
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures,	microcephaly risk gene
HCCS	Linear skin defects with multiple congenital anomalies 1,	microcephaly risk gene
HCFC1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type),	microcephaly risk gene
HIVEP2	Mental retardation, autosomal dominant 43,	microcephaly risk gene
HMGCL	HMG-CoA lyase deficiency,	microcephaly risk gene
HNRNPH2	Mental retardation, X-linked, syndromic, Bain type,	microcephaly risk gene
HSPD1	Spastic paraplegia 13, autosomal dominant,	microcephaly risk gene
HTRA2	3-methylglutaconic aciduria, type VIII,	microcephaly risk gene
IARS1	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy,	microcephaly risk gene
IBA57	Multiple mitochondrial dysfunctions syndrome 3,	microcephaly risk gene
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome,	microcephaly risk gene
IFIH1	Aicardi-Goutieres syndrome 7,	microcephaly risk gene

IFT140	Retinitis pigmentosa 80,	microcephaly risk gene
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency,	microcephaly risk gene
IGF1R	Insulin-like growth factor I, resistance to,	microcephaly risk gene
IKBKG	Immunodeficiency 33,	microcephaly risk gene
INPP5K	Muscular dystrophy, congenital, with cataracts and intellectual disability,	microcephaly risk gene
IQSEC2	Mental retardation, X-linked 1/78,	microcephaly risk gene
ISCA1	Multiple mitochondrial dysfunctions syndrome 5,	microcephaly risk gene
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital,	microcephaly risk gene
ITPA	Epileptic encephalopathy, early infantile, 35,	microcephaly risk gene
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts,	microcephaly risk gene
KAT6A	Arboleda-Tham syndrome,	microcephaly risk gene
KAT6B	SBBYSS syndrome,	microcephaly risk gene
KATNB1	Lissencephaly 6, with microcephaly,	microcephaly risk gene
KCNA4	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum,	microcephaly risk gene
KCNJ2	Short QT syndrome 3,	microcephaly risk gene
KCNJ6	Keppen-Lubinsky syndrome,	microcephaly risk gene
KCNT1	Epilepsy, nocturnal frontal lobe, 5,	microcephaly risk gene
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions,	microcephaly risk gene
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation,	microcephaly risk gene
KIF14	Microcephaly 20, primary, autosomal recessive,	microcephaly risk gene
KIF1A	NESCAV syndrome,	microcephaly risk gene
KIF2A	Cortical dysplasia, complex, with other brain malformations 3,	microcephaly risk gene
KIF5A	Myoclonus, intractable, neonatal,	microcephaly risk gene
KIF5C	Cortical dysplasia, complex, with other brain malformations 2,	microcephaly risk gene
KIFBP	Goldberg-Shprintzen megacolon syndrome,	microcephaly risk gene
KMT2B	Dystonia 28, childhood-onset,	microcephaly risk gene
KMT2C	Kleefstra syndrome 2,	microcephaly risk gene
KMT2D	Kabuki syndrome 1,	microcephaly risk gene
KNL1	Microcephaly 4, primary, autosomal recessive,	microcephaly risk gene
KYNU	Vertebral, cardiac, renal, and limb defects syndrome 2,	microcephaly risk gene
LAGE3	Galloway-Mowat syndrome 2, X-linked,	microcephaly risk gene
LARP7	Alazami syndrome,	microcephaly risk gene
LIAS	Hyperglycinemia, lactic acidosis, and seizures,	microcephaly risk gene
LIG4	LIG4 syndrome,	microcephaly risk gene
LINGO1	Mental retardation, autosomal recessive 64,	microcephaly risk gene
LINS1	Mental retardation, autosomal recessive 27,	microcephaly risk gene
LIPT2	Encephalopathy, neonatal severe, with lactic acidosis and brain	microcephaly risk gene

	abnormalities,	
LRP5	van Buchem disease, type 2,	microcephaly risk gene
LSS	Alopecia-mental retardation syndrome 4,	microcephaly risk gene
MAB21L1	Cerebellar, ocular, craniofacial, and genital syndrome,	microcephaly risk gene
MACF1	Lissencephaly 9 with complex brainstem malformation,	microcephaly risk gene
MAP1B	Periventricular nodular heterotopia 9,	microcephaly risk gene
MAPRE2	Symmetric circumferential skin creases, congenital, 2,	microcephaly risk gene
MASP1	3MC syndrome 1,	microcephaly risk gene
MBD5	Mental retardation, autosomal dominant 1,	microcephaly risk gene
MBTPS2	IFAP syndrome with or without BRESHECK syndrome,	microcephaly risk gene
MCM4	Immunodeficiency 54,	microcephaly risk gene
MCOLN1	Mucolipidosis IV,	microcephaly risk gene
MCPH1	Microcephaly 1, primary, autosomal recessive,	microcephaly risk gene
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy,	microcephaly risk gene
MED25	Basel-Vanagait-Smirin-Yosef syndrome,	microcephaly risk gene
MESD	Osteogenesis imperfecta, type XX,	microcephaly risk gene
METTL5	Intellectual developmental disorder, autosomal recessive 72,	microcephaly risk gene
MFF	Encephalopathy due to defective mitochondrial and peroxisomal fission 2,	microcephaly risk gene
MFSD2A	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain imaging abnormalities,	microcephaly risk gene
MGME1	Mitochondrial DNA depletion syndrome 11,	microcephaly risk gene
MICOS13	Combined oxidative phosphorylation deficiency 37,	microcephaly risk gene
MICU1	Myopathy with extrapyramidal signs,	microcephaly risk gene
MIPEP	Combined oxidative phosphorylation deficiency 31,	microcephaly risk gene
MIR17HG	Feingold syndrome 2,	microcephaly risk gene
MKS1	Bardet-Biedl syndrome 13,	microcephaly risk gene
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type,	microcephaly risk gene
MPC1	Mitochondrial pyruvate carrier deficiency,	microcephaly risk gene
MPDU1	Congenital disorder of glycosylation, type If,	microcephaly risk gene
MPLKIP	Trichothiodystrophy 4, nonphotosensitive,	microcephaly risk gene
MRPS22	Combined oxidative phosphorylation deficiency 5,	microcephaly risk gene
MRPS34	Combined oxidative phosphorylation deficiency 32,	microcephaly risk gene
MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis,	microcephaly risk gene
MTFMT	Combined oxidative phosphorylation deficiency 15,	microcephaly risk gene
MTHFR	Homocystinuria due to MTHFR deficiency,	microcephaly risk gene
MTHFS	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination,	microcephaly risk gene
MVK	Hyper-IgD syndrome,	microcephaly risk gene
MYCN	Feingold syndrome 1,	microcephaly risk gene
MYH3	Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B,	microcephaly risk gene
MYO18B	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism,	microcephaly risk gene
NAA10	Ogden syndrome,	microcephaly risk gene

NACC1	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination,	microcephaly risk gene
NADK2	2,4-dienoyl-CoA reductase deficiency,	microcephaly risk gene
NALCN	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1,	microcephaly risk gene
NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type,	microcephaly risk gene
NARS2	Combined oxidative phosphorylation deficiency 24,	microcephaly risk gene
NBN	Leukemia, acute lymphoblastic,	microcephaly risk gene
NCAPD3	Microcephaly 22, primary, autosomal recessive,	microcephaly risk gene
NCAPG2	Khan-Khan-Katsanis syndrome,	microcephaly risk gene
NDE1	Lissencephaly 4 (with microcephaly),	microcephaly risk gene
NDUFA11	Mitochondrial complex I deficiency, nuclear type 14,	microcephaly risk gene
NDUFS1	Mitochondrial complex I deficiency, nuclear type 5,	microcephaly risk gene
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1,	microcephaly risk gene
NDUFV2	Mitochondrial complex I deficiency, nuclear type 7,	microcephaly risk gene
NEPRO	Anauxetic dysplasia 3,	microcephaly risk gene
NEXMIF	Mental retardation, X-linked 98,	microcephaly risk gene
NGLY1	Congenital disorder of deglycosylation,	microcephaly risk gene
NIPBL	Cornelia de Lange syndrome 1,	microcephaly risk gene
NOP10	Dyskeratosis congenita, autosomal recessive 1,	microcephaly risk gene
NSDHL	CHILD syndrome,	microcephaly risk gene
NSMCE2	Seckel syndrome 10,	microcephaly risk gene
NSUN2	Mental retardation, autosomal recessive 5,	microcephaly risk gene
NTNG2	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia,	microcephaly risk gene
NTRK2	Obesity, hyperphagia, and developmental delay,	microcephaly risk gene
NUP107	Galloway-Mowat syndrome 7,	microcephaly risk gene
NUP188	Sandestig-Stefanova syndrome,	microcephaly risk gene
OCLN	Pseudo-TORCH syndrome 1,	microcephaly risk gene
OGT	Mental retardation, X-linked 106,	microcephaly risk gene
ORC1	Meier-Gorlin syndrome 1,	microcephaly risk gene
ORC4	Meier-Gorlin syndrome 2,	microcephaly risk gene
ORC6	Meier-Gorlin syndrome 3,	microcephaly risk gene
OSGEP	Galloway-Mowat syndrome 3,	microcephaly risk gene
OSTM1	Osteopetrosis, autosomal recessive 5,	microcephaly risk gene
OTUD6B	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies,	microcephaly risk gene
PAFAH1B1	Subcortical laminar heterotopia,	microcephaly risk gene
PAH	Phenylketonuria,	microcephaly risk gene
PAK3	Mental retardation, X-linked 30/47,	microcephaly risk gene
PARN	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4,	microcephaly risk gene
PARS2	Epileptic encephalopathy, early infantile, 75,	microcephaly risk gene
PCDH12	Diencephalic-mesencephalic junction dysplasia syndrome 1,	microcephaly risk gene
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II,	microcephaly risk gene

PDHA1	Pyruvate dehydrogenase E1-alpha deficiency,	microcephaly risk gene
PDHX	Lacticacidemia due to PDX1 deficiency,	microcephaly risk gene
PEX5	Peroxisome biogenesis disorder 2B,	microcephaly risk gene
PEX7	Peroxisome biogenesis disorder 9B,	microcephaly risk gene
PGAP2	Hyperphosphatasia with mental retardation syndrome 3,	microcephaly risk gene
PGAP3	Hyperphosphatasia with mental retardation syndrome 4,	microcephaly risk gene
PHF6	Borjeson-Forssman-Lehmann syndrome,	microcephaly risk gene
PHGDH	Neu-Laxova syndrome 1,	microcephaly risk gene
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2,	microcephaly risk gene
PIGO	Hyperphosphatasia with mental retardation syndrome 2,	microcephaly risk gene
PIGS	Glycosylphosphatidylinositol biosynthesis defect 18,	microcephaly risk gene
PIGY	Hyperphosphatasia with mental retardation syndrome 6,	microcephaly risk gene
PISD	Liberfarb syndrome,	microcephaly risk gene
PLAA	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies,	microcephaly risk gene
PLEKHG2	Leukodystrophy and acquired microcephaly with or without dystonia,	microcephaly risk gene
PLK4	Microcephaly and chorioretinopathy, autosomal recessive, 2,	microcephaly risk gene
PLP1	Pelizaeus-Merzbacher disease,	microcephaly risk gene
PLPBP	Epilepsy, early-onset, vitamin B6-dependent,	microcephaly risk gene
PMM2	Congenital disorder of glycosylation, type Ia,	microcephaly risk gene
PMPCB	Multiple mitochondrial dysfunctions syndrome 6,	microcephaly risk gene
PNKP	Microcephaly, seizures, and developmental delay,	microcephaly risk gene
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency,	microcephaly risk gene
POGZ	White-Sutton syndrome,	microcephaly risk gene
POLA1	Pigmentary disorder, reticulate, with systemic manifestations, X-linked,	microcephaly risk gene
POLH	Xeroderma pigmentosum, variant type,	microcephaly risk gene
POLR1A	Acrofacial dysostosis, Cincinnati type,	microcephaly risk gene
POLR2A	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities,	microcephaly risk gene
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3,	microcephaly risk gene
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12,	microcephaly risk gene
POMT1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1,	microcephaly risk gene
POMT2	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2,	microcephaly risk gene
POR	Disordered steroidogenesis due to cytochrome P450 oxidoreductase,	microcephaly risk gene
PORCN	Focal dermal hypoplasia,	microcephaly risk gene
PPP1R15B	Microcephaly, short stature, and impaired glucose metabolism 2,	microcephaly risk gene
PPP2CA	Neurodevelopmental disorder and language delay with or without structural brain abnormalities,	microcephaly risk gene
PPP2R1A	Mental retardation, autosomal dominant 36,	microcephaly risk gene
PPT1	Ceroid lipofuscinosis, neuronal, 1,	microcephaly risk gene
PQBP1	Renpenning syndrome,	microcephaly risk gene

PRDX1	Methylmalonic aciduria and homocystinuria, cblC type, digenic,	microcephaly risk gene
PRKD1	Congenital heart defects and ectodermal dysplasia,	microcephaly risk gene
PRKDC	Immunodeficiency 26, with or without neurologic abnormalities,	microcephaly risk gene
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures,	microcephaly risk gene
PRUNE1	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies,	microcephaly risk gene
PSAT1	Neu-Laxova syndrome 2,	microcephaly risk gene
PSPH	Phosphoserine phosphatase deficiency,	microcephaly risk gene
PTDSS1	Lenz-Majewski hyperostotic dwarfism,	microcephaly risk gene
PTF1A	Pancreatic and cerebellar agenesis,	microcephaly risk gene
PTPN23	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity,	microcephaly risk gene
PTRH2	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease,	microcephaly risk gene
PTS	Hyperphenylalaninemia, BH4-deficient, A,	microcephaly risk gene
PUF60	Verheij syndrome,	microcephaly risk gene
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1,	microcephaly risk gene
PUS3	Mental retardation, autosomal recessive 55,	microcephaly risk gene
PUS7	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature,	microcephaly risk gene
PYCR1	Cutis laxa, autosomal recessive, type IIIB,	microcephaly risk gene
PYCR2	Leukodystrophy, hypomyelinating, 10,	microcephaly risk gene
QARS1	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy,	microcephaly risk gene
QDPR	Hyperphenylalaninemia, BH4-deficient, C,	microcephaly risk gene
QRICH1	Ververi-Brady syndrome,	microcephaly risk gene
RAB11B	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter,	microcephaly risk gene
RAB18	Warburg micro syndrome 3,	microcephaly risk gene
RAB3GAP1	Warburg micro syndrome 1,	microcephaly risk gene
RAB3GAP2	Warburg micro syndrome 2,	microcephaly risk gene
RAD21	Cornelia de Lange syndrome 4,	microcephaly risk gene
RAD51	Mirror movements 2,	microcephaly risk gene
RARS1	Leukodystrophy, hypomyelinating, 9,	microcephaly risk gene
RARS2	Pontocerebellar hypoplasia, type 6,	microcephaly risk gene
RBBP8	Jawad syndrome,	microcephaly risk gene
RBPJ	Adams-Oliver syndrome 3,	microcephaly risk gene
RELN	Lissencephaly 2 (Norman-Roberts type),	microcephaly risk gene
REN	Renal tubular dysgenesis,	microcephaly risk gene
RFT1	Congenital disorder of glycosylation, type In,	microcephaly risk gene
RHOBTB2	Epileptic encephalopathy, early infantile, 64,	microcephaly risk gene
RIC1	CATIFA syndrome,	microcephaly risk gene
RLIM	Tonne-Kalscheuer syndrome,	microcephaly risk gene

RNASEH2A	Aicardi-Goutieres syndrome 4,	microcephaly risk gene
RNASEH2B	Aicardi-Goutieres syndrome 2,	microcephaly risk gene
RNASEH2C	Aicardi-Goutieres syndrome 3,	microcephaly risk gene
RNASET2	Leukoencephalopathy, cystic, without megalencephaly,	microcephaly risk gene
RNF113A	Trichothiodystrophy 5, nonphotosensitive,	microcephaly risk gene
RNF13	Epileptic encephalopathy, early infantile, 73,	microcephaly risk gene
RNF168	RIDDLE syndrome,	microcephaly risk gene
RNU4ATAC	Microcephalic osteodysplastic primordial dwarfism, type I,	microcephaly risk gene
RPL10	Mental retardation, X-linked, syndromic, 35,	microcephaly risk gene
RPS19	Diamond-Blackfan anemia 1,	microcephaly risk gene
RPS23	Brachycephaly, trichomegaly, and developmental delay,	microcephaly risk gene
RPS6KA3	Mental retardation, X-linked 19,	microcephaly risk gene
RSPRY1	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type,	microcephaly risk gene
RTEL1	Dyskeratosis congenita, autosomal recessive 5,	microcephaly risk gene
RTTN	Microcephaly, short stature, and polymicrogyria with seizures,	microcephaly risk gene
RUSC2	Mental retardation, autosomal recessive 61,	microcephaly risk gene
SALL1	Townes-Brocks syndrome 1,	microcephaly risk gene
SAMHD1	Aicardi-Goutieres syndrome 5,	microcephaly risk gene
SATB2	Glass syndrome,	microcephaly risk gene
SBF1	Charcot-Marie-Tooth disease, type 4B3,	microcephaly risk gene
SC5D	Lathosterolosis,	microcephaly risk gene
SCN1A	Febrile seizures, familial, 3A,	microcephaly risk gene
SCN2A	Episodic ataxia, type 9,	microcephaly risk gene
SCN3A	Epilepsy, familial focal, with variable foci 4,	microcephaly risk gene
SCN8A	Seizures, benign familial infantile, 5,	microcephaly risk gene
SCYL1	Spinocerebellar ataxia, autosomal recessive 21,	microcephaly risk gene
SCYL2	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum,	microcephaly risk gene
SELENOI	Spastic paraplegia 81, autosomal recessive,	microcephaly risk gene
SEPSECS	Pontocerebellar hypoplasia type 2D,	microcephaly risk gene
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome,	microcephaly risk gene
SF3B4	Acrofacial dysostosis 1, Nager type,	microcephaly risk gene
SGPL1	Nephrotic syndrome, type 14,	microcephaly risk gene
SHH	Schizencephaly,	microcephaly risk gene
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome,	microcephaly risk gene
SIL1	Marinesco-Sjogren syndrome,	microcephaly risk gene
SIN3A	Witteveen-Kolk syndrome,	microcephaly risk gene
SIX3	Holoprosencephaly 2,	microcephaly risk gene
SKI	Shprintzen-Goldberg syndrome,	microcephaly risk gene
SLC13A5	Epileptic encephalopathy, early infantile, 25,	microcephaly risk gene
SLC16A2	Allan-Herndon-Dudley syndrome,	microcephaly risk gene
SLC1A2	Epileptic encephalopathy, early infantile, 41,	microcephaly risk gene
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive	microcephaly risk gene

	microcephaly,	
SLC25A19	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type),	microcephaly risk gene
SLC25A22	Epileptic encephalopathy, early infantile, 3,	microcephaly risk gene
SLC25A24	Fontaine progeroid syndrome,	microcephaly risk gene
SLC2A1	Dystonia 9,	microcephaly risk gene
SLC35A1	Congenital disorder of glycosylation, type IIf,	microcephaly risk gene
SLC35C1	Congenital disorder of glycosylation, type IIc,	microcephaly risk gene
SLC39A14	Hypermanganesemia with dystonia 2,	microcephaly risk gene
SLC5A6	Neurodegeneration, infantile-onset, biotin-responsive,	microcephaly risk gene
SLC6A8	Cerebral creatine deficiency syndrome 1,	microcephaly risk gene
SLC6A9	Glycine encephalopathy with normal serum glycine,	microcephaly risk gene
SLC9A6	Mental retardation, X-linked syndromic, Christianson type,	microcephaly risk gene
SLX4	Fanconi anemia, complementation group P,	microcephaly risk gene
SMAD4	Polyposis, juvenile intestinal,	microcephaly risk gene
SMARCA2	Nicolaides-Baraitser syndrome,	microcephaly risk gene
SMARCA4	Coffin-Siris syndrome 4,	microcephaly risk gene
SMARCB1	Coffin-Siris syndrome 3,	microcephaly risk gene
SMARCE1	Coffin-Siris syndrome 5,	microcephaly risk gene
SMC1A	Cornelia de Lange syndrome 2,	microcephaly risk gene
SMC3	Cornelia de Lange syndrome 3,	microcephaly risk gene
SMG9	Heart and brain malformation syndrome,	microcephaly risk gene
SMO	Pallister-Hall-like syndrome,	microcephaly risk gene
SMPD4	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies,	microcephaly risk gene
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome,	microcephaly risk gene
SNRPB	Cerebrocostomandibular syndrome,	microcephaly risk gene
SOX11	Coffin-Siris syndrome 9,	microcephaly risk gene
SOX2	Microphthalmia, syndromic 3,	microcephaly risk gene
SOX4	Coffin-Siris syndrome 10,	microcephaly risk gene
SP110	Hepatic venoocclusive disease with immunodeficiency,	microcephaly risk gene
SPATA5	Epilepsy, hearing loss, and mental retardation syndrome,	microcephaly risk gene
SPOP	Nabais Sa-de Vries syndrome, type 1,	microcephaly risk gene
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency,	microcephaly risk gene
SPTAN1	Epileptic encephalopathy, early infantile, 5,	microcephaly risk gene
SSR4	Congenital disorder of glycosylation, type Iy,	microcephaly risk gene
ST3GAL5	Salt and pepper developmental regression syndrome,	microcephaly risk gene
STAG1	Mental retardation, autosomal dominant 47,	microcephaly risk gene
STAG2	Mullegama-Klein-Martinez syndrome,	microcephaly risk gene
STAMBP	Microcephaly-capillary malformation syndrome,	microcephaly risk gene
STIL	Microcephaly 7, primary, autosomal recessive,	microcephaly risk gene
STT3A	Congenital disorder of glycosylation, type Iw,	microcephaly risk gene
SVBP	Neurodevelopmental disorder with ataxia, hypotonia, and	microcephaly risk gene

	microcephaly,	
SYNGAP1	Mental retardation, autosomal dominant 5,	microcephaly risk gene
TAF1	Dystonia-Parkinsonism, X-linked,	microcephaly risk gene
TAF13	Mental retardation, autosomal recessive 60,	microcephaly risk gene
TAF2	Mental retardation, autosomal recessive 40,	microcephaly risk gene
TAF6	Alazami-Yuan syndrome,	microcephaly risk gene
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration,	microcephaly risk gene
TAPT1	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type,	microcephaly risk gene
TASP1	Suleiman-El-Hattab syndrome,	microcephaly risk gene
TBC1D20	Warburg micro syndrome 4,	microcephaly risk gene
TBC1D23	Pontocerebellar hypoplasia, type 11,	microcephaly risk gene
TBC1D24	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp,	microcephaly risk gene
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum,	microcephaly risk gene
TBCE	Kenny-Caffey syndrome, type 1,	microcephaly risk gene
TBL1XR1	Pierpont syndrome,	microcephaly risk gene
TBX1	Conotruncal anomaly face syndrome,	microcephaly risk gene
TCF4	Corneal dystrophy, Fuchs endothelial, 3,	microcephaly risk gene
TDP2	Spinocerebellar ataxia, autosomal recessive 23,	microcephaly risk gene
TECPR2	Spastic paraplegia 49, autosomal recessive,	microcephaly risk gene
TELO2	You-Hoover-Fong syndrome,	microcephaly risk gene
TET3	Beck-Fahrner syndrome,	microcephaly risk gene
TFAP2A	Branchiooculofacial syndrome,	microcephaly risk gene
THOC2	Mental retardation, X-linked 12/35,	microcephaly risk gene
THOC6	Beaulieu-Boycott-Innes syndrome,	microcephaly risk gene
TINF2	Revesz syndrome,	microcephaly risk gene
TLK2	Mental retardation, autosomal dominant 57,	microcephaly risk gene
TMEM165	Congenital disorder of glycosylation, type IIk,	microcephaly risk gene
TMEM260	Structural heart defects and renal anomalies syndrome,	microcephaly risk gene
TMTC3	Lissencephaly 8,	microcephaly risk gene
TMX2	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity,	microcephaly risk gene
TOE1	Pontocerebellar hypoplasia, type 7,	microcephaly risk gene
TOP3A	Microcephaly, growth restriction, and increased sister chromatid exchange 2,	microcephaly risk gene
TP53	Bone marrow failure syndrome 5,	microcephaly risk gene
TP53RK	Galloway-Mowat syndrome 4,	microcephaly risk gene
TPRKB	Galloway-Mowat syndrome 5,	microcephaly risk gene
TRAIP	Seckel syndrome 9,	microcephaly risk gene
TRAPPC11	Muscular dystrophy, limb-girdle, autosomal recessive 18,	microcephaly risk gene
TRAPPC12	Encephalopathy, progressive, early-onset, with brain atrophy and	microcephaly risk gene

	spasticity,	
TRAPPC2L	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis,	microcephaly risk gene
TRAPPC4	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy,	microcephaly risk gene
TRAPPC6B	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy,	microcephaly risk gene
TRAPPC9	Mental retardation, autosomal recessive 13,	microcephaly risk gene
TREX1	Vasculopathy, retinal, with cerebral leukodystrophy,	microcephaly risk gene
TRIP13	Mosaic variegated aneuploidy syndrome 3,	microcephaly risk gene
TRIT1	Combined oxidative phosphorylation deficiency 35,	microcephaly risk gene
TRMT1	Mental retardation, autosomal recessive 68,	microcephaly risk gene
TRMT10A	Microcephaly, short stature, and impaired glucose metabolism 1,	microcephaly risk gene
TRRAP	Developmental delay with or without dysmorphic facies and autism,	microcephaly risk gene
TSEN15	Pontocerebellar hypoplasia, type 2F,	microcephaly risk gene
TSEN2	Pontocerebellar hypoplasia type 2B,	microcephaly risk gene
TSEN54	Pontocerebellar hypoplasia type 4,	microcephaly risk gene
TTI2	Mental retardation, autosomal recessive 39,	microcephaly risk gene
TUBA1A	Lissencephaly 3,	microcephaly risk gene
TUBA8	Cortical dysplasia, complex, with other brain malformations 8,	microcephaly risk gene
TUBB	Symmetric circumferential skin creases, congenital, 1,	microcephaly risk gene
TUBB2B	Cortical dysplasia, complex, with other brain malformations 7,	microcephaly risk gene
TUBB3	Fibrosis of extraocular muscles, congenital, 3A,	microcephaly risk gene
TUBB4A	Leukodystrophy, hypomyelinating, 6,	microcephaly risk gene
TUBG1	Cortical dysplasia, complex, with other brain malformations 4,	microcephaly risk gene
TUBGCP2	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures,	microcephaly risk gene
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, 3,	microcephaly risk gene
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1,	microcephaly risk gene
TUFM	Combined oxidative phosphorylation deficiency 4,	microcephaly risk gene
UBA5	Epileptic encephalopathy, early infantile, 44,	microcephaly risk gene
UBE3A	Angelman syndrome,	microcephaly risk gene
UBE3B	Kaufman oculocerebrofacial syndrome,	microcephaly risk gene
UBR1	Johanson-Blizzard syndrome,	microcephaly risk gene
UFC1	Neurodevelopmental disorder with spasticity and poor growth,	microcephaly risk gene
UFM1	Leukodystrophy, hypomyelinating, 14,	microcephaly risk gene
UGP2	Epileptic encephalopathy, early infantile, 83,	microcephaly risk gene
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2,	microcephaly risk gene
UPB1	Beta-ureidopropionase deficiency,	microcephaly risk gene
VARS1	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy,	microcephaly risk gene
VARS2	Combined oxidative phosphorylation deficiency 20,	microcephaly risk gene
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2,	microcephaly risk gene

VPS11	Leukodystrophy, hypomyelinating, 12,	microcephaly risk gene
VPS13B	Cohen syndrome,	microcephaly risk gene
VPS13D	Spinocerebellar ataxia, autosomal recessive 4,	microcephaly risk gene
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1,	microcephaly risk gene
VPS51	Pontocerebellar hypoplasia, type 13,	microcephaly risk gene
VPS53	Pontocerebellar hypoplasia, type 2E,	microcephaly risk gene
WDR37	Neurooculocardiogenitourinary syndrome,	microcephaly risk gene
WDR4	Microcephaly, growth deficiency, seizures, and brain malformations,	microcephaly risk gene
WDR45B	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures,	microcephaly risk gene
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations,	microcephaly risk gene
WDR73	Galloway-Mowat syndrome 1,	microcephaly risk gene
WWOX	Spinocerebellar ataxia, autosomal recessive 12,	microcephaly risk gene
XPA	Xeroderma pigmentosum, group A,	microcephaly risk gene
XRCC4	Short stature, microcephaly, and endocrine dysfunction,	microcephaly risk gene
ZBTB11	Intellectual developmental disorder, autosomal recessive 69,	microcephaly risk gene
ZBTB16	Skeletal defects, genital hypoplasia, and mental retardation,	microcephaly risk gene
ZBTB18	Mental retardation, autosomal dominant 22,	microcephaly risk gene
ZC4H2	Wieacker-Wolff syndrome,	microcephaly risk gene
ZEB2	Mowat-Wilson syndrome,	microcephaly risk gene
ZIC1	Structural brain anomalies with impaired intellectual development and craniosynostosis,	microcephaly risk gene
ZNF148	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies,	microcephaly risk gene
ZNF335	Microcephaly 10, primary, autosomal recessive,	microcephaly risk gene
ZNHIT3	PEHO syndrome,	microcephaly risk gene
ZSWIM6	Acromelic frontonasal dysostosis,	microcephaly risk gene
DDX3X	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type,	microcephaly/macrocephaly risk gene
DHCR24	Desmosterolosis,	microcephaly/macrocephaly risk gene
DOCK6	Adams-Oliver syndrome 2,	microcephaly/macrocephaly risk gene
EXT2	Seizures, scoliosis, and macrocephaly syndrome,	microcephaly/macrocephaly risk gene
FGFR3	Muenke syndrome,	microcephaly/macrocephaly risk gene
GJA1	Erythrokeratodermia variabilis et progressiva 3,	microcephaly/macrocephaly risk gene
HUWE1	Mental retardation, X-linked syndromic, Turner type,	microcephaly/macrocephaly risk gene
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type,	microcephaly/macrocephaly risk gene

L1CAM	MASA syndrome,	microcephaly/macrocephaly risk gene
MECP2	Mental retardation, X-linked syndromic, Lubs type,	microcephaly/macrocephaly risk gene
MGAT2	Congenital disorder of glycosylation, type IIa,	microcephaly/macrocephaly risk gene
MOCS1	Molybdenum cofactor deficiency A,	microcephaly/macrocephaly risk gene
MOCS2	Molybdenum cofactor deficiency B,	microcephaly/macrocephaly risk gene
MYMK	Carey-Fineman-Ziter syndrome,	microcephaly/macrocephaly risk gene
OFD1	Orofaciodigital syndrome I,	microcephaly/macrocephaly risk gene
PCGF2	Turnpenny-Fry syndrome,	microcephaly/macrocephaly risk gene
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis,	microcephaly/macrocephaly risk gene
POLE	FILS syndrome,	microcephaly/macrocephaly risk gene
PPP1R12A	Genitourinary and/or/brain malformation syndrome,	microcephaly/macrocephaly risk gene
RAC1	Mental retardation, autosomal dominant 48	microcephaly/macrocephaly risk gene
SLC25A1	Myasthenic syndrome, congenital, 23, presynaptic	microcephaly/macrocephaly risk gene
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	microcephaly/macrocephaly risk gene
TRIO	Intellectual developmental disorder, autosomal dominant 44, with microcephaly	microcephaly/macrocephaly risk gene
XYLT1	Desbuquois dysplasia 2	microcephaly/macrocephaly risk gene