Every candidate gene has an AutoCaSc value that suggests ist relevance. When high (e.g. >7, reddish), then it makes more sense than when it is low (<5, green colors). If the case is not developmental delay / epilepsy, it gets the value A, B and C.

HGNC_Symbol	Variant1FullName	Variant2FullName	Inheritance	AutoCaSc	Zygosity	Origin	Number_Candidates_In_Family	DiseaseGroup_Leading Symptome	HPO_Main_Terms
GLS	NM_001256310.1:c.695dupp.(Asp232Glufs*2)		homo	12,4	homo	maternal&paternal	1	NDD + Epilepsy	Seizures, Status epilepticus, Infantile onset, Infantile spasms, Epileptic encephalopathy
DGKZ	NM_001199266.1:c.3227C>Gp.(Thr1076Arg)	NM_001199266.1:c.3326A>Gp.(Gln1109Arg)	comphet	3,7	comphet	maternal&paternal	1	NDD + Epilepsy	Epileptic encephalopathy, Seizures, Failure to thrive, Hypoplasia of the corpus callosum, Hypsarrhythmia, Infantile onset, muscular hypotonia,
DUT	XM_005254212.1:c.218T>Cp.(Val73Ala)		de novo	7,6	het	de novo	1	NDD + Epilepsy	Retrognathia, Myoclonus, EEG abnormality, Infantile encephalopathy, Epileptic encephalopathy
GLS	NM_001256310.1:c.815G>Ap.(Arg272Lys)	NM_001256310.1:c.241C>Tp.(Gln81*)	comphet	10,7	comphet	maternal&paternal	1	NDD + Epilepsy	Microcephaly, Seizures, Status epilepticus, CNS demyelination, EEG with burst suppression, Peripheral demyelination, Epileptic encephalopathy
PLXNB3	NM_001163257.1:c.4343C>Ap.(Thr1448Asn)		de novo	7,1	het	de novo	2	NDD	Hydrocephalus, Intellectual disability, hypotonia, Global developmental delay, Atria septal defect, Patent ductus arteriosus,Transposition of the great arteries with ventricular septal defect
GBP5	NM_001134486.2:c.154T>Cp.(Ser52Pro)	NM_001134486.2:c.502_505dupp.(Ser169*)	comphet	5,0	comphet	maternal&paternal	2	NDD	Hydrocephalus, Intellectual disability, hypotonia, Global developmental delay, Atria septal defect, Patent ductus arteriosus,Transposition of the great arteries with ventricular septal defect
GRIN3B	NM_138690.1:c.1811C>Tp.(Thr604Met)	NM_138690.1:c.2114A>Cp.(Tyr705Ser)	comphet	6,1	comphet	maternal&paternal	1	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Hypsarrhythmia, Infantile
CLSTN1	NM_001009566.1:c.1844C>Tp.(Thr615Met)		homo	8,1	homo	maternal&paternal	2	NDD	Cataract, Peters anomaly, Autism, Global developmental delay
CASP9	NM_001229.4:c.868+5G>Cp.?		homo	8,8	homo	maternal&paternal	2	NDD	Cataract, Peters anomaly, Autism, Global developmental delay
PUM2	NM_015317.1:c.1595G>Ap.(Ser532Asn)		de novo	7,6	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Hypsarrhythmia,
CARMIL1	XM_005249221.1:c.3617C>Tp.(Ser1206Leu)	XM_005249221.1:c.2659G>Ap.(Glu887Lys)	comphet	3,5	comphet	maternal&paternal	1	NDD + Epilepsy	Microcephaly, Delayed puberty, Abnormality of skin pigmentation, Seizures, Agenesis of corpus callosum, Growth delay, Intellectual disability, Limb hypertonia, Scoliosis, Chorioretinal lacunae, Muscular hypotonia of the trunk, Infantile axial hypotonia, Infantile spasms, Small hand
SMCR8	NM_144775.2:c.2404C>Tp.(Arg802Cys)		de novo	4,7	het	de novo	2	NDD	Microcephaly, Epicanthus, Intellectual disability, Global developmental delay, Plagiocephaly, Abnormal facial shape, Wide nasal base
FRMPD3	XM_042978.8:c.3538C>Tp.(Arg1180Trp)		hemi	3,3	hemi	maternal	2	NDD	Microcephaly, Epicanthus, Intellectual disability, Global developmental delay, Plagiocephaly, Abnormal facial shape, Wide nasal base
PUM1	NM_001020658.1:c.3439C>Tp.(Arg1147Trp)		de novo	9,5	het	de novo	2	NDD	Global developmental delay, Microcephaly, Cryptorchidism, Ptosis, Short stature, Short phalanx of finger, Frontal hirsutism, Arachnoid cyst
BAIAP3	NM_001199096.1:c.892G>Tp.(Gly298Trp)		de novo	5,9	het	de novo	2	NDD	Global developmental delay, Microcephaly, Cryptorchidism, Ptosis, Short stature, Short phalanx of finger, Frontal hirsutism, Arachnoid cyst
PSMB3	NM_002795.2:c.424T>Cp.(Cys142Arg)		de novo	4,7	het	de novo	1	NDD	Trismus, Arthrogryposis multiplex congenita, Vesicoureteral reflux, Abnormality of the kidney, abnormal facial shape, Global developmental delay
VPS4A	NM_013245.2:c.291T>Gp.(Ser97Arg)		de novo	7,3	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Epileptic encephalopathy
TAB3	NM_152787.3:c.1952A>Gp.(Gln651Arg)		hemi	3,9	hemi	maternal	2	NDD + Epilepsy	Intellectual disability, Seizures, Epileptic encephalopathy
PPP1R37	NM_019121.1:c.509C>Tp.(Ser170Phe)		de novo	6,0	het	de novo	2	NDD	Bilateral cryptorchidism, Short stature, Epileptic encephalopathy, Microcephaly
AQP6	NM_001652.3:c.146C>Tp.(Pro49Leu)		de novo	5,2	het	de novo	2	NDD	Bilateral cryptorchidism, Short stature, Epileptic encephalopathy, Microcephaly
SLC32A1	NM_080552.2:c.787G>Ap.(Val263Met)		de novo	7,8	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Generalized myoclonic seizures, Infantile onset
IRAK1	NM_001025242.1:c.609T>Gp.(Cys203Trp)		de novo	6,1	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Generalized myoclonic seizures, Infantile onset
GRINA	NM_000837.1:c.967-6C>Tp.?		homo	5,7	homo	maternal&paternal	2	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
MED22	NM_133640.4:c.397_399delp.(Glu133del)		homo	5,6	homo	maternal&paternal	2	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
GTPBP2	NM_019096.3:c.1191C>Ap.(Asn397Lys)		de novo	7,1	het	de novo	1	NDD	Tall stature, Macrocephaly, Retrognathia, High forehead, Low-set ears, Global developmental delay
PABPC1	NM_002568.3:c.1691A>Gp.(Glu564Gly)		de novo	11,0	het	de novo	1	NDD + Epilepsy	global developmental delay, seizures, visual impairment, bicuspid aortic valve
NCOA2	NM_006540.2:c.1454T>Cp.(Met485Thr)	NM_006540.2:c.3509T>Cp.(Met1170Thr)	comphet	6,1	comphet	maternal&paternal	1	NDD	Intellectual disability, Seizures, Encephalopathy, Cerebral atrophy, Intellectual disability, profound, EEG abnormality, Intellectual disability, severe, Cognitive impairment
SPEN	NM_015001.2:c.8092A>Gp.(Asn2698Asp)		de novo	8,0	het	de novo	2	NDD	Microcephaly, Underdeveloped nasal alae, Strabismus, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Postnatal microcephaly
BOK	NM_032515.4:c.356C>Tp.(Thr119Met)		de novo	5,6	het	de novo	1	NDD	Microcephaly, Underdeveloped nasal alae, Strabismus, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Postnatal microcephaly
ENOX2	NM_006375.2:c.148A>Gp.(Met50Val)		hemi	3,3	hemi	maternal	2	NDD	Microcephaly, Underdeveloped nasal alae, Strabismus, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Postnatal microcephaly
CHD5	NM_015557.2:c.5003-5G>Ap.?	NM_015557.2:c.5249C>Tp.(Thr1750Met)	comphet	5,3	comphet	maternal&paternal	3	NDD	Autism, Intellectual disability, Global developmental delay
HDAC4	NM_006037.3:c.1663G>Ap.(Gly555Ser)		het	6,7	het	maternal	3	NDD	Autism, Intellectual disability, Global developmental delay
SLC10A3	NM_001142391.1:c.1160C>Tp.(Thr387Met)		hemi	5,0	hemi	maternal	3	NDD	Autism, Intellectual disability, Global developmental delay
WBP1	NM_012477.3:c.25G>Ap.(Gly9Ser)		de novo	3,8	het	de novo	1	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Atonic seizures
DUX4L4	NM_001177376.2:c.880C>Tp.(Gln294*)		de novo	7,0	het	de novo	1	NDD	Microcephaly, Intellectual disability, Global developmental delay, Short stature
TEX44	NM_152614.2:c.1146C>Gp.(His382Gln)		de novo	4,9	het	de novo	1	NDD	mild global developmental delay, delayed speech and language development

ASIC1	NM_001095.3:c.363-2A>Gp.?		de novo	10,2	het	de novo	3	NDD + Epilepsy	Seizures, Abnormal social behavior, Epileptic encephalopathy
FAM168B	NM_001009993.2:c.452G>Ap.(Gly151Glu)		de novo	6,5	het	de novo	3	NDD + Epilepsy	Seizures, Abnormal social behavior, Epileptic encephalopathy
SPEN	NM_015001.2:c.3968T>Gp.(Met1323Arg)		de novo	8,1	het	de novo	3	NDD + Epilepsy	mild global developmental delay, seizures, heterotopia, oral cleft, tall stature, obesity
CSMD1	NM_033225.5:c.7327A>Gp.(Ile2443Val)	NM_033225.5:c.8444A>Cp.(Glu2815Ala)	comphet	5,3	comphet	maternal&paternal	3	NDD + Epilepsy	mild global developmental delay, seizures, heterotopia, oral cleft, tall stature, obesity
CENPV	NM_181716.2:c.75_92delp.(Ala26_Ala31del)		de novo	5,4	het	de novo	3	NDD + Epilepsy	mild global developmental delay, seizures, heterotopia, oral cleft, tall stature, obesity
CACNB4	NM_000726.3:c.848C>Tp.(Ser283Leu)		de novo	9,5	het	de novo	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures
KLHL17	NM_198317.2:c.1568C>Tp.(Ala523Val)		homo	5,2	homo	maternal&paternal	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures
POLR2A	NM_000937.4:c.4808G>Ap.(Arg1603His)	NM_000937.4:c.778G>Ap.(Val260Met)	comphet	6,1	comphet	maternal&paternal	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures
ZNF12	NM_006956.2:c.670T>Cp.(Ser224Pro)	NM_006956.2:c.1438G>Ap.(Val480Ile)	comphet	3,1	comphet	maternal&paternal	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures
CASKIN1	NM_020764.3:c.4103G>Ap.(Ser1368Asn)		homo	7,8	homo	maternal&paternal	3	NDD	global developmental delay, absent speech, gait disturbance, EEG abnormality, decreased body weight
MADD	NM_001135943.1:c.1037T>Cp.(Leu346Pro)		homo	9,6	homo	maternal&paternal	3	NDD	global developmental delay, absent speech, gait disturbance, EEG abnormality, decreased body weight
CELSR2	NM_001408.2:c.4706C>Tp.(Pro1569Leu)	NM_001408.2:c.8629G>Ap.(Gly2877Ser)	comphet	7,6	comphet	maternal&paternal	3	NDD	global developmental delay, absent speech, gait disturbance, EEG abnormality, decreased body weight
FAT3	NM_001008781.2:c.3669+7G>Ap.?	NM_001008781.2:c.12922G>Cp.(Asp4308His)	comphet	4,5	comphet	maternal&paternal	3	NDD + Epilepsy	Autism, Seizures, Global developmental delay, Motor delay, Absent speech, Epileptic encephalopathy
CHMP7	NM_152272.3:c.214C>Ap.(Leu72Met)		de novo	6,0	het	de novo	3	NDD + Epilepsy	Autism, Seizures, Global developmental delay, Motor delay, Absent speech, Epileptic encephalopathy
ANKFY1	NM_001257999.1:c.1966G>Ap.(Ala656Thr)		homo	5,5	homo	maternal&paternal	3	NDD + Epilepsy	Autism, Seizures, Global developmental delay, Motor delay, Absent speech, Epileptic encephalopathy
ACTL6B	NM_016188.4:c.1027G>Ap.(Gly343Arg)		de novo	10,7	het	de novo	1	NDD	Muscular hypotonia, Abnormality of mouth shape, Stereotypical hand wringing, Microcephaly, Global developmental delay
LUC7L	NM_018032.3:c.614G>Ap.(Arg205His)		de novo	5,9	het	de novo	2	NDD + Epilepsy	Seizures, Global developmental delay
PRDX4	NM_006406.1:c.724G>Ap.(Gly242Arg)		hemi	5,5	hemi	maternal	2	NDD + Epilepsy	Seizures, Global developmental delay
GRIA4	NM_000829.3:c.2090G>Cp.(Arg697Pro)		de novo	10,1	het	de novo	1	NDD	Delayed speech and language development, Intellectual disability, Poor speech
MAPK8IP3	NM_001040439.1:c.1556G>Ap.(Arg519Gln)		de novo	10,9	het	de novo	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormality of body weight, Increased body weight,
DIS3	NM_001128226.1:c.1486A>Gp.(Arg496Gly)	NM_001128226.1:c.2785T>Cp.(*929Glnext*14)	comphet	7,1	comphet	maternal&paternal	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormality of body weight, Increased body weight,
CAMTA2	NM_001171166.1:c.2639A>Gp.(Asp880Gly)		homo	4,6	homo	maternal&paternal	2	NDD + Epilepsy	Seizures, Status epilepticus, Hypsarrhythmia,
FAT1	NM_005245.3:c.2137A>Gp.(Ile713Val)	NM_005245.3:c.9440T>Gp.(Val3147Gly)	comphet	5,6	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Status epilepticus, Hypsarrhythmia,
STAM	NM_003473.3:c.119G>Cp.(Arg40Pro)		de novo	8,1	het	de novo	1	NDD	Short stature, Ataxia, Cataract, Microphthalmia, Microcephaly, Nystagmus, Global developmental delay
GAL3ST3	NM_033036.2:c.39G>Cp.(Lys13Asn)		de novo	5,0	het	de novo	3	NDD + Epilepsy	seizures, focal seizures
SDK1	NM_152744.3:c.1295G>Cp.(Gly432Ala)	NM_152744.3:c.3802C>Tp.(Arg1268Trp)	comphet	4,7	comphet	maternal&paternal	3	NDD + Epilepsy	seizures, focal seizures
ZNF503	NM_032772.4:c.69_71dup, p.(Gly27dup)	NM_032772.4:c.1105G>Tp.(Gly369Cys)	comphet	4,1	comphet	maternal&paternal	3	NDD + Epilepsy	seizures, focal seizures
TOB1	.CCTCAGTCCTCTCCAGTinsGGGp.(Leu296Leufs*4)		de novo	9,9	het	de novo	1	NDD	Cerebral calcification, Seizures, Congenital cataract, Autistic behavior, Obesity, Global developmental delay
GPKOW	NM_015698.4:c.1334G>Ap.(Arg445Gln)		hemi	3,4	hemi	maternal	1	NDD	Autism, Global developmental delay
MACF1	NM_012090.5:c.1531C>Tp.(Arg511Cys)	NM_012090.5:c.3465G>Ap.(=)	comphet	6,3	comphet	maternal&paternal	1	NDD + Epilepsy	global developmental delay, seizures,
TAAR2	NM_001033080.1:c.113G>Tp.(Arg38Ile)		de novo	4,4	het	de novo	3	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormality of the caudate nucleus, Infantile onset, Attention deficit hyperactivity disorder
MORF4L2	NM_001142418.1:c.287A>Gp.(Gln96Arg)		hemi	4,8	hemi	maternal	3	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormality of the caudate nucleus, Infantile onset, Attention deficit hyperactivity disorder
SLC35B3	NM_001142540.1:c.1135C>Tp.(Pro379Ser)	NM_001142540.1:c.1069G>Cp.(Gly357Arg)	comphet	3,5	comphet	maternal&paternal	3	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormality of the caudate nucleus, Infantile onset, Attention deficit hyperactivity disorder
URB2	NM_014777.2:c.1949delp.(Gly650Valfs*2)		de novo	5,8	het	de novo	2	NDD + Epilepsy	Seizures, Myoclonic absences, developmental delay
OGDHL	NM_001143996.1:c.489G>Cp.(Trp163Cys)	NM_001143996.1:c.1315C>Tp.(Arg439Cys)	comphet	4,7	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Myoclonic absences, developmental delay
SNX27	NM_030918.5:c.913G>Ap.(Ala305Thr)	NM_030918.5:c.69_71dup, p.(Gly25dup)	comphet	6,7	comphet	maternal&paternal	1	NDD	Microcephaly, Hirsutism, Intellectual disability, Global developmental delay, Short stature
DOC2B	NM_003585.4:c.898G>Ap.(Gly300Ser)		de novo	6,9	het	de novo	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures, infantile onset
ANKFN1	NM_153228.2:c.1052A>Gp.(Asn351Ser)		de novo	6,4	het	de novo	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures, infantile onset
POU4F2	NM_004575.2:c.417C>Ap.(Asp139Glu)	NM_004575.2:c.180_200delp.(Gly62_Gly68del)	comphet	3,8	comphet	maternal&paternal	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures, infantile onset
C11ORF95	NM_001144936.1:c.1592T>Cp.(Val531Ala)		homo	4,9	homo	maternal&paternal	2	NDD + Epilepsy	global developmental delay, seizures, hypoplasia of the corpus callosum
SCUBE2	NM_001170690.1:c.68C>Tp.(Pro23Leu)		de novo	4,9	het	de novo	2	NDD + Epilepsy	global developmental delay, seizures, hypoplasia of the corpus callosum
NINL	NM_025176.4:c.277+2T>Cp.?		homo	9,4	homo	maternal&paternal	4	NDD	Intellectual disability, Global developmental delay
CTSB	NM_001908.3:c.444C>Tp.(=)		homo	5,7	homo	maternal&paternal	4	NDD	Intellectual disability, Global developmental delay
CNOT1	NM_001265612.1:c.6727A>Gp.(Met2243Val)		homo	7,8	homo	maternal&paternal	1	NDD	Intellectual disability, Global developmental delay
B4GALNT3	NM_173593.3:c.1798G>Ap.(Glu600Lys)	NM_173593.3:c.1640C>Tp.(Pro547Leu)	comphet	3,5	comphet	maternal&paternal	4	NDD	Intellectual disability, Global developmental delay
SRPX	NM_001170750.1:c.1270A>Tp.(Thr424Ser)		hemi	3,9	hemi	maternal	4	NDD	Intellectual disability, Global developmental delay
CUX1	202543.1:c.3783_3784dup, p.(Leu1262Argfs*10)		de novo	12,1	het	de novo	1	NDD	Macrocephaly, Umbilical hernia, Chronic constipation, Inguinal hernia, Delayed speech and language development, mild global developmental delay

KMT2E	NM_018682.3:c.3554C>Gp.(Ser1185*)		de novo	12,4	het	de novo	1	NDD	Intellectual disability, Seizures, EEG with spike-wave complexes, EEG with continuous slow activity,
NPTX1	NM_002522.3:c.970G>Ap.(Gly324Arg)		de novo	6,7	het	de novo	2	NDD	Spastic tetraparesis, Optic atrophy, Periventricular leukomalacia, Microcephaly, Global developmental delay
H2BC4	NM_003526.2:c.154G>Tp.(Asp52Tyr)		de novo	5,4	het	de novo	2	NDD	Spastic tetraparesis, Optic atrophy, Periventricular leukomalacia, Microcephaly, Global developmental delay
FRY	NM_023037.2:c.4688G>Cp.(Ser1563Thr)		de novo	7,5	het	de novo	1	NDD	global developmental delay, intellectual disability, epileptic seizures, microcephaly, Dandy-Walker malformation, Polymicrogyria, syndactyly, partial duplication of thumb phalanx
MICAL1	NM_001159291.1:c.571+1G>Tp.?	NM_001159291.1:c.2724-8C>Tp.?	comphet	3,8	comphet	maternal&paternal	3	NDD + Epilepsy	Specific learning disability, Absence seizures, Cortical dysplasia, EEG with continuous slow activity, Seizures
SPATA31A3	NM_001083124.1:c.3206C>Tp.(Ser1069Phe)		de novo	3,6	het	de novo	3	NDD + Epilepsy	Specific learning disability, Absence seizures, Cortical dysplasia, EEG with continuous slow activity, Seizures
ATP2B4	NM_001001396.2:c.2819A>Gp.(Lys940Arg)		homo	5,3	homo	maternal&paternal	3	NDD + Epilepsy	Specific learning disability, Absence seizures, Cortical dysplasia, EEG with continuous slow activity, Seizures
EGR3	NM_001199880.1:c.477C>Ap.(Tyr159*)		de novo	10,1	het	de novo	1	NDD	Intellectual disability, learning disability
FREM3	NM_001168235.1:c.728delp.(Glu243Glyfs*25)	NM_001168235.1:c.5401C>Tp.(Leu1801Phe)	comphet	5,3	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Encephalopathy, Focal seizures, Encephalitis
PLXNA1	NM_032242.3:c.2690G>Ap.(Arg897His)	NM_032242.3:c.1045G>Cp.(Val349Leu)	comphet	4,3	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Encephalopathy, Focal seizures, Encephalitis
SPTBN5	NM_016642.3:c.5680G>Tp.(Glu1894*)		homo	8,2	homo	maternal&paternal	2	NDD	intellectual disability
HOOK2	NM_001100176.1:c.1718-6C>Tp.?		homo	4,4	homo	maternal&paternal	2	NDD	intellectual disability
ZKSCAN3	NM_001242894.1:c.253A>Tp.(Ile85Phe)		de novo	5,0	het	de novo	3	NDD	Hypothyroidism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Short stature, Abnormal social behavior
KALRN	NM_001024660.3:c.5980C>Gp.(Leu1994Val)	NM_001024660.3:c.2171C>Tp.(Ser724Leu)	comphet	6,9	comphet	maternal&paternal	3	NDD	Hypothyroidism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Short stature, Abnormal social behavior
SFXN3	NM_030971.3:c.785G>Ap.(Arg262His)	NM_030971.3:c.640delp.(Ala214Glnfs*9)	comphet	4,9	comphet	maternal&paternal	3	NDD	Hypothyroidism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Short stature, Abnormal social behavior
AP1G1	XM_005255821.1:c.468G>Ap.(=)		de novo	6,9	het	de novo	1	NDD + Epilepsy	Seizures, Epileptic encephalopathy
NLRX1	NM_024618.2:c.428C>Tp.(Pro143Leu)		de novo	4,7	het	de novo	1	NDD	Ptosis, Muscular hypotonia, Global developmental delay, Abnormal facial shape, Short stature, Feeding difficulties, Thick hair
AGO2	NM_001164623.1:c.602G>Tp.(Gly201Val)		de novo	8,8	het	de novo	1	NDD + Epilepsy	Intellectual disability, Global developmental delay, Motor delay, Gait disturbance, Absent speech, Bicuspid aortic valve, Patent foramen ovale, Atrioventricular block, Intellectual disability, moderate, Poor speech, Obstructive sleep apnea, Short stature, Sleep apnea, Intellectual disability, severe, Epileptic spasms, Epileptic encephalopathy
L3MBTL1	NM_015478.6:c.478T>Ap.(Ser160Thr)		homo	7,2	homo	maternal&paternal	3	NDD	Agitation, Aggressive behavior, Delayed speech and language development, Intellectual disability
UNC13A	NM_001080421.2:c.2786G>Ap.(Gly929Glu)		de novo	8,4	het	de novo	3	NDD	Agitation, Aggressive behavior, Delayed speech and language development, Intellectual disability
MIA3	NM_198551.2:c.3981+3A>Gp.?		homo	7,0	homo	maternal&paternal	3	NDD	Agitation, Aggressive behavior, Delayed speech and language development, Intellectual disability
GCN1	NM_006836.1:c.7082G>Ap.(Arg2361Gln)		de novo	7,7	het	de novo	3	Neuro	Seizures, Hypoglycemia, Myopathy, Focal seizures, Ichthyosis, EEG with focal epileptiform discharges
GRIA1	NM_000827.3:c.81C>Ap.(=)		homo	8,2	homo	maternal&paternal	3	Neuro	Seizures, Hypoglycemia, Myopathy, Focal seizures, Ichthyosis, EEG with focal epileptiform discharges
DEPTOR	NM_022783.2:c.496A>Gp.(Met166Val)	NM_022783.2:c.426-5C>Tp.?	comphet	4,0	comphet	maternal&paternal	3	NDD + Epilepsy	Seizures, Hypoglycemia, Myopathy, Focal seizures, Ichthyosis, EEG with focal epileptiform discharges
FHDC1	NM_033393.2:c.568C>Tp.(Arg190Trp)		de novo	4,8	het	de novo	1	NDD	Hypertension, Intellectual disability,mild, Obesity, Abnormality of the pulmonary valve, I Hyperlipidemia, Childhood-onset truncal obesity
RASGRP1	NM_001128602.1:c.1487C>Gp.(Ser496*)		de novo	9,8	het	de novo	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures
CNTNAP4	NM_033401.3:c.3353G>Cp.(Gly1118Ala)		de novo	8,3	het	de novo	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures
MCM7	NM_001278595.1:c.1147A>Cp.(Met383Leu)		de novo	7,7	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizures, IGlobal developmental delay, Infantile onset, epileptic encephalopathy
ATP2B1	NM_001001323.1:c.1376A>Gp.(His459Arg)		de novo	8,8	het	de novo	3	NDD + Epilepsy	Autism, Intellectual disability, Seizures, Global developmental delay, Poor speech, Focal seizures
DRG1	NM_004147.3:c.43-1G>Tp.?		de novo	5,9	het	de novo	3	NDD + Epilepsy	Autism, Intellectual disability, Seizures, Global developmental delay, Poor speech, Focal seizures
DGKZ	NM_001199266.1:c.132_134delp.(Ser45del)	NM_001199266.1:c.16G>Cp.(Gly6Arg)	comphet	4,4	comphet	maternal&paternal	3	NDD + Epilepsy	Autism, Intellectual disability, Seizures, Global developmental delay, Poor speech, Focal seizures
TANC2	NM_025185.3:c.4405delp.(Arg1469Glyfs*6)		de novo	11,4	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Encephalopathy, Epileptic encephalopathy
ARHGEF7	NM_001113511.2:c.17A>Cp.(Gln6Pro)		de novo	7,9	het	de novo	3	NDD	global developmental delay, intellectual disability
SEMA3B	NM_001005914.2:c.952C>Tp.(His318Tyr)	NM_001005914.2:c.728T>Cp.(Phe243Ser)	comphet	3,6	comphet	maternal&paternal	3	NDD	global developmental delay, intellectual disability
ETV5	NM_004454.2:c.232+1G>Ap.?		de novo	10,0	het	de novo	4	NDD	global developmental delay, intellectual disability, generalized hypotonia,
MDN1	NM_014611.2:c.2965-3T>Cp.?	NM_014611.2:c.9524A>Cp.(His3175Pro)	comphet	4,4	comphet	maternal&paternal	4	NDD	global developmental delay, intellectual disability, generalized hypotonia,
CASS4	NM_001164114.1:c.1576G>Ap.(Val526Ile)	NM_001164114.1:c.1421G>Tp.(Arg474Leu)	comphet	3,1	comphet	maternal&paternal	4	NDD	global developmental delay, intellectual disability, generalized hypotonia,

WDFY3	NM_014991.4:c.749A>Gp.(Asn250Ser)		de novo	10,0	het	de novo	2	NDD + Epilepsy	seizures, peripheral axonal neuropathy, motor delay, gait disturbance, EEG with focal epilepti-form discharges
EXD3	NM_017820.4:c.859G>Ap.(Asp287Asn)	NM_017820.4:c.1831-2A>Gp.?	comphet	6,7	comphet	maternal&paternal	2	NDD + Epilepsy	seizures, peripheral axonal neuropathy, motor delay, gait disturbance, EEG with focal epilepti-form discharges
CFAP54	XM_001715090.5:c.2257A>Gp.(Met753Val)	XM_001715090.5:c.2057G>Ap.(Arg686Lys)	comphet	3,4	comphet	maternal&paternal	3	NDD	Coloboma, Iris coloboma, mild Intellectual disability, mild Global developmental delay
GRIN3B	M_138690.1:c.1090_1091delp.(Met364Valfs*5)	NM_138690.1:c.1936A>Gp.(Met646Val)	comphet	7,2	comphet	maternal&paternal	1	NDD	Intellectual disability, Abnormal facial shape, Myoclonus
EIF5B	NM_015904.3:c.3607C>Tp.(Gln1203*)		de novo	10,1	het	de novo	1	NDD	Macrocephaly, Autism, Intellectual disability, Absent speech, Intellectual disability, severe
PTP4A1	NM_003463.4:c.8G>Ap.(Arg3Gln)		de novo	5,3	het	de novo	1	NDD	mental retardation, autism
POLR1B	NM_001137604.1:c.2893G>Ap.(Val965Ile)		de novo	6,5	het	de novo	3	NDD	Seizures, Pachygyria, Delayed CNS myelination, Heterotopia, Periventricular gray matter heterotopia, Intracranial cystic lesion, Abnormality of brain morphology
HIST1H4B	NM_003544.2:c.158A>Gp.(Glu53Gly)		de novo	4,2	het	de novo	3	NDD	Seizures, Pachygyria, Delayed CNS myelination, Heterotopia, Periventricular gray matter heterotopia, Intracranial cystic lesion, Abnormality of brain morphology
BAHCC1	NM_001080519.2:c.4691+5C>G		de novo	3,0	het	de novo	3	NDD	Seizures, Pachygyria, Delayed CNS myelination, Heterotopia, Periventricular gray matter heterotopia, Intracranial cystic lesion, Abnormality of brain morphology
PAK1	NM_001128620.1:c.1409T>Gp.(Leu470Arg)		de novo	9,9	het	de novo	1	ndd	Macrocephaly, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Focal clonic seizures, Focal seizures, Intellectual disability, severe, Epileptic spasms, Focal motor seizures, Focal tonic seizures, Cognitive impairment
PHACTR1	NM_001242648.2:c.1156G>Ap.(Glu386Lys)		de novo	7,4	het	de novo	2	NDD	Global developmental delay, Intellectual disability, mild
DBP	NM_001352.4:c.511G>Tp.(Ala171Ser)		de novo	6,1	het	de novo	2	NDD	Global developmental delay, Intellectual disability, mild
STC1	NM_003155.2:c.693_697delp.(Glu232Glyfs*12)		de novo	6,7	het	de novo	1	NDD	mild global developmental delay, expressive speech disorder, obesity since age three years
KANK4	NM_181712.4:c.1849C>Tp.(Gln617*)		de novo	4,6	het	de novo	1	NDD	Retinal coloboma, Seizures, Intellectual disability, mild, Global developmental delay, Motor delay, Hypoplasia of the retina, Intracranial cystic lesion, Mild global developmental delay, Infantile spasms
LCTL	NM_207338.3:c.692_693dup		de novo	5,7	het	de novo	2	NDD + Epilepsy	epileptic encephalopathy, seizures
KLHL6	NM_130446.2:c.1061C>Ap.(Pro354Gln)		homo	4,9	homo	maternal&paternal	2	NDD + Epilepsy	epileptic encephalopathy, seizures
GRIN3B	NM_138690.2:c.2114A>Gp.(Tyr705Cys)	NM_138690.2:c.2314G>Ap.(Gly772Ser)	comphet	6,0	comphet	maternal&paternal	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Intellectual disability, progressive, Focal seizures, EEG with focal slow activity, Intellectual disability, severe, Focal motor seizures, EEG with focal epileptiform discharges, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Neurodevelopmental abnormality, Cognitive impairment
DNAJC7	NM_001144766.2:c.941C>Tp.(Ala314Val)		de novo	6,4	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Generalized seizures, Hypsarrhythmia, Epileptic spasms
KIRREL2	NM_032123.6:c.1275delp.(Pro425Profs*41)		het	6,0	het	paternal	1	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Absence seizures, Generalized myoclonic seizures, Episodic vomiting, Epileptic spasms, Myoclonic atonic seizures, Epileptic
EIF3B	NM_001037283.1:c.28C>Ap.(Pro10Thr)		de novo	7,0	het	de novo	1	NDD + Epilepsy	Absence seizures, EEG abnormality, Febrile seizures, Eyelid myoclonias, Childhood onset
HIST1H3H	NM_003536.2:c.397G>Tp.(Gly133Cys)		de novo	4,6	het	de novo	1	NDD + Epilepsy	Global developmental delay, Hypsarrhythmia, Inability to walk, Epileptic spasms, Infantile spasms
FBP2	NM_003837.3:c.128A>Gp.(Lys43Arg)		de novo	6,2	het	de novo	2	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Motor delay, Frontal bossing, Delayed gross motor development, Delayed fine motor development, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay
AASDH	NM_181806.3:c.2908-5_2908-4insGTTp.?	NM_181806.3:c.3220dup, p.(Leu1074Profs*10)	comphet	5,6	comphet	maternal&paternal	3	NDD + Epilepsy	Narrow mouth, Upslanted palpebral fissure, Delayed speech and language development, Intellectual disability, Global developmental delay, Pachygyria, Lissencephaly, Absent speech, Dysphagia, Polymicrogyria, Status epilepticus, Gliosis, Intellectual disability, moderate, Cerebellar malformation, Poor speech, Abnormality of the cerebral white matter, Excessive salivation, Focal white matter lesions, Focal seizures, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptic spasms, EEG with focal epileptiform discharges, Cerebral white matter atrophy, Cerebral white matter agenesis, Oral-pharyngeal
CAST	deletionexon16		de novo	9,0	het	de novo	3	NDD + Epilepsy	Narrow mouth, Upslanted palpebral fissure, Delayed speech and language development, Intellectual disability, Global developmental delay, Pachygyria, Lissencephaly, Absent speech, Dysphagia, Polymicrogyria, Status epilepticus, Gliosis, Intellectual disability, moderate, Cerebellar malformation, Poor speech, Abnormality of the cerebral white matter, Excessive salivation, Focal white matter lesions, Focal seizures, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptic spasms, EEG with focal epileptiform discharges, Cerebral white matter atrophy, Cerebral white matter agenesis, Oral-pharyngeal
E2F4	M_001950.3:c.947_958delp.(Ser316_Ser319del)		de novo	6,6	het	de novo	2	NDD	Cleft palate, Intellectual disability, Intellectual disability, mild, Global developmental delay, Absent speech, Atria septal defect, Abnormal facial shape, Intellectual disability, moderate, Short stature, Intellectual disability, severe
C1orf228	NM_001145636.1:c.979C>Tp.(Arg327Cys)		de novo	4,6	het	de novo	2	NDD	Cleft palate, Intellectual disability, Intellectual disability, mild, Global developmental delay, Absent speech, Atria septal defect, Abnormal facial shape, Intellectual disability, moderate, Short stature, Intellectual disability, severe

ZNF664	NM_001204298.1:c.691G>Ap.(Glu231Lys)		de novo	4,8	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Hypsarrhythmia, Epileptic spasms, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay, Cognitive impairment, Epileptic encephalopathy
NIT1	M_001185092.1:c.244_256delp.(Phe83Hisfs*63)	NM_001185092.1:c.302T>Cp.(Leu101Pro)	comphet	6,2	comphet	maternal&paternal	2	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Hypsarrhythmia, Epileptic spasms, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay, Cognitive impairment, Epileptic encephalopathy
LPIN2	NM_014646.2:c.2537A>Gp.(Asn846Ser)		de novo	7,2	het	de novo	3	NDD + Epilepsy	Nystagmus, Horizontal nystagmus, Seizures, Global developmental delay, Absent speech, Cardiomyopathy, Vacuolated lymphocytes, Abnormal facial shape, Gait ataxia, Absence seizures, EEG abnormality, Myoclonic atonic seizures, Epileptic encephalopathy
KDM6B	NM_001080424.1:c.1130C>Tp.(Ala377Val)		homo	8,4	homo	maternal&paternal	3	NDD + Epilepsy	Nystagmus, Horizontal nystagmus, Seizures, Global developmental delay, Absent speech, Cardiomyopathy, Vacuolated lymphocytes, Abnormal facial shape, Gait ataxia, Absence seizures, EEG abnormality, Myoclonic atonic seizures, Epileptic encephalopathy
MAPKAPK2	NM_004759.4:c.445C>Tp.(Arg149*)		de novo	9,3	het	de novo	1	NDD + Epilepsy	Cryptorchidism, Hypospadias, Microcephaly, Visual impairment, Visual field defect, Intellectual disability, Muscular hypotonia, Global developmental delay, Plagiocephaly, Oligohydramnios, Intellectual disability, severe, Epileptic spasms, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay
MOXD1	NM_015529.3:c.350A>Gp.(His117Arg)		homo	6,0	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Febrile seizures, Intellectual disability, borderline, Intellectual disability, severe, Focal tonic seizures
TLK2	NM_001112707.1:c.667A>Tp.(Met223Leu)		het	4,8	het	unknown	3	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Febrile seizures, Intellectual disability, borderline, Intellectual disability, severe, Focal tonic seizures
KDM5B	NM_006618.4:c.1286T>Gp.(Ile429Ser)		de novo	10,1	het	de novo	1	NDD + Epilepsy	Renal duplication, Hydrocephalus, Autism, Hypertrichosis, Intellectual disability, Seizures, Global developmental delay, Agenesis of corpus callosum, Abnormal facial shape, Intellectual disability, moderate, Impaired pain sensation, Intellectual disability, severe, Colpocephaly, Cognitive impairment, Septo-optic dysplasia
NIPAL3	NM_020448.4:c.205G>Ap.(Ala69Thr)	NM_020448.4:c.163-8G>Ap.?	comphet	3,7	comphet	maternal&paternal	2	NDD	Hearing impairment, Sensorineural hearing impairment, Delayed speech and language development, Precocious puberty, Muscular hypotonia, Global developmental delay, Absent speech, Poor speech, High-frequency hearing impairment, Muscular hypotonia of the trunk
ZIK1	NM_001010879.3:c.924delp.(Ser308Serfs*203)		homo	7,8	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Generalized seizures, Febrile seizures, Focal seizures
ZNF331	NM_001079906.1:c.281G>Ap.(Arg94His)		homo	3,8	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Generalized seizures, Febrile seizures, Focal seizures
UBE3C	NM_014671.2:c.485G>Cp.(Ser162Thr)	NM_014671.2:c.871G>Ap.(Val291Ile)	comphet	4,9	comphet	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Generalized seizures, Febrile seizures, Focal seizures
SGF29	NM_138414.2:c.733T>Cp.(Tyr245His)		de novo	6,7	het	de novo	1	NDD	Microcephaly, Abnormality of the outer ear, Protruding ear, Abnormality of the ear, Hypotelorism, Autistic behavior, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Talipes equinovarus, Hypoplasia of the corpus callosum, Intellectual disability, profound, Intellectual disability, moderate, Short stature, Intellectual disability, severe, Clinodactyly
HSPD1	NM_002156.4:c.1394_1406delp.(Ile465Lysfs*9)		de novo	12,8	het	de novo	1	Neuro	Hypogonadotrophic hypogonadism, Tall stature, Psychosis, Depression, Psychotic episodes, Dementia, Overgrowth, Neurodegeneration, Bipolar affective disorder, Brain atrophy
PLCB3	NM_000932.2:c.1792G>Cp.(Glu598Gln)		de novo	B	het	de novo	1	Malformations	Failure to thrive, Growth delay, Omphalocele, Double outlet right ventricle
STARD9	NM_020759.2:c.1649A>Gp.(Asn550Ser)	NM_020759.2:c.10380C>Gp.(His3460Gln)	comphet	3,9	comphet	maternal&paternal	1	NDD + Epilepsy	Global developmental delay, Absence seizures, Intellectual disability, moderate, Progressive truncal ataxia, Epileptic spasms, Myoclonic absences, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Infantile spasms
GABRA3	NM_000808.3:c.931+5G>Ap.?		hemi	7,3	hemi	maternal	2	NDD + Epilepsy	Microcephaly, Agitation, Intellectual disability, Intellectual disability, mild, Global developmental delay, Constipation, Intellectual disability, moderate, EEG abnormality, Intellectual disability, borderline, Attention deficit hyperactivity disorder, Epileptic spasms, Anteverted ears
ELMOD2	NM_153702.3:c.580C>Tp.(Arg194Cys)		de novo	5,3	het	de novo	2	NDD + Epilepsy	Microcephaly, Agitation, Intellectual disability, Intellectual disability, mild, Global developmental delay, Constipation, Intellectual disability, moderate, EEG abnormality, Intellectual disability, borderline, Attention deficit hyperactivity disorder, Epileptic spasms, Anteverted ears

NR2F6	NM_005234.3:c.1051G>Ap.(Gly351Arg)		de novo	5,49	het	de novo	1	NDD	Microcephaly, Global developmental delay, Generalized hypotonia, Neonatal hypotonia, Failure to thrive, Severe failure to thrive, Failure to thrive in infancy, Ventricular septal defect, Abnormal cardiac septum morphology, Overlapping toe, Neonatal onset, Short stature, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Abnormal ventricular septum morphology, Gerbode ventricular septal defect, Inlet ventricular septal defect, Muscular ventricular septal defect, Subarterial ventricular septal defect, Perimembranous ventricular septal defect, Restrictive ventricular septal defect, Abnormality of cardiovascular system morphology, Ventricular septal aneurysm, Muscular ventricular
TMEM199	NM_152464.2:c.5C>Tp.(Ala2Val)		de novo	5,3	het	de novo	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Abnormal facial shape, Status epilepticus, Intellectual disability, moderate, Infantile muscular hypotonia, Intellectual disability, severe, Epileptic spasms, Cognitive
NCAPH	NM_001281710.1:c.563-4T>Gp.?	NM_001281710.1:c.667G>Ap.(Glu223Lys)	comphet	5,2	comphet	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Abnormal facial shape, Status epilepticus, Intellectual disability, moderate, Infantile muscular hypotonia, Intellectual disability, severe, Epileptic spasms, Cognitive
DOK2	NM_003974.3:c.1007C>Ap.(Thr336Asn)	NM_003974.3:c.602G>Ap.(Arg201His)	comphet	C	comphet	maternal&paternal	1	Immunology	Hemolytic anemia, Fever, Abnormal thrombosis, Vasculitis, Intermittent thrombocytopenia, Congenital blindness, Colon perforation
UNC13C	NM_001080534.2:c.283C>Tp.(Arg95*)		homo	8,3	homo	maternal&paternal	1	NDD	Global developmental delay, microcephaly, Ehlers-Danlos-Syndrom (CHST1 positive)
NRIP1	NM_003489.3:c.2077G>Tp.(Gly693Cys)		de novo	5,9	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic-clonic seizures, Focal clonic seizures, Intellectual disability, moderate, Focal seizures with impairment of consciousness or awareness, Intellectual disability, borderline, Generalized tonic seizures, Symptomatic seizures, Focal tonic seizures, Cognitive
DDX42	NM_007372.3:c.221+1G>Ap.?		de novo	10,2	het	de novo	1	NDD + Epilepsy	Epilepsy, optic atrophy, diabetes insipidus and hypothyroidism
ZCRB1	NM_033114.3:c.78G>Cp.(Leu26Phe)		de novo	5,4	het	de novo	3	NDD + Epilepsy	Refractory epilepsy, epileptic encephalopathy, psychomotor retardation, microcephaly, short stature, facial dysmorphisms, nephrocalcinosis
DNAJC10	NM_001271581.1:c.1671+1G>Tp.?		de novo	7,2	het	de novo	3	NDD + Epilepsy	Refractory epilepsy, epileptic encephalopathy, psychomotor retardation, microcephaly, short stature, facial dysmorphisms, nephrocalcinosis
CD200	NM_001004196.3:c.161C>Ap.(Thr54Lys)		de novo	5,8	het	de novo	3	NDD + Epilepsy	Refractory epilepsy, epileptic encephalopathy, psychomotor retardation, microcephaly, short stature, facial dysmorphisms, nephrocalcinosis
SPHK2	NM_001204158.2:c.1534G>Tp.(Val512Leu)		homo	5,0	homo	maternal&paternal	1	NDD + Epilepsy	Perinatal cerebral infarction, global developmental delay, motor and speech delay, microcephaly, epilepsy, short stature, combined heart failure (DORV, VSD, ASD, valvular pulmonary stenosis)
CELSR3	NM_001407.2:c.8254T>Cp.(Phe2752Leu)	NM_001407.2:c.79T>Cp.(Phe27Leu)	comphet	6,1	comphet	maternal&paternal	1	NDD + Epilepsy	No language development, microcephaly (-2,2 SD), short stature (<3P), EEG abnormalities, epilepsy, delayed myelination in cMRT; Trio-Exom-Analysis unremarkable (05,12,2016)
GEMIN4	NM_015721.2:c.1580A>Gp.(Asn527Ser)	M_015721.2:c.1415_1416delp.(Pro472Argfs*23)	comphet	6,8	comphet	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Global developmental delay, Abnormal heart morphology, Ventricular septal defect, Status epilepticus, Intellectual disability, moderate, Short stature, Atrioventricular canal defect, Intellectual disability, borderline, Intellectual disability, severe, Epileptic spasms, Epileptiform EEG discharges, Abnormality of cardiovascular system morphology
ZNF319	NM_020807.2:c.1294G>Cp.(Val432Leu)		de novo	5,2	het	de novo	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Global developmental delay, Abnormal heart morphology, Ventricular septal defect, Status epilepticus, Intellectual disability, moderate, Short stature, Atrioventricular canal defect, Intellectual disability, borderline, Intellectual disability, severe, Epileptic spasms, Epileptiform EEG discharges, Abnormality of cardiovascular system morphology
RENBP	NM_002910.5:c.695G>Ap.(Gly232Glu)		hemi	5,5	hemi	maternal	1	NDD + Epilepsy	Epilepsy, susceptibility to fall
NARS1	NM_004539.3:c.1600C>Tp.(Arg534*)		de novo	7,6	het	de novo	2	NDD + Epilepsy	GDD, ataxia, muscular hypotonia, microcephaly, EEG abnormalities, brain volume reduction
INTS1	NM_001080453.2:c.6248T>Gp.(Phe2083Cys)	NM_001080453.2:c.5272A>Gp.(Ile1758Val)	comphet	5,5	comphet	maternal&paternal	2	NDD + Epilepsy	GDD, ataxia, muscular hypotonia, microcephaly, EEG abnormalities, brain volume reduction
TAOK1	NM_020791.2:c.332C>Tp.(Ser111Phe)		de novo	8,8	het	de novo	1	NDD	Dysmorphic syndrome, cleft lip and palate, failure to thrive, macrocephaly, muscular hypotonia, developmental delay
STARD9	NM_020759.2:c.12652C>Tp.(His4218Tyr)	NM_020759.2:c.13445C>Tp.(Ser4482Phe)	comphet	4,2	comphet	maternal&paternal	1	NDD + Epilepsy	mild ID, generalized epilepsy
IQCH	NM_001031715.2:c.2552T>Cp.(Leu851Pro)		de novo	5,2	het	de novo	1	NDD	GDD, bland-white-garland-syndrome, facial dysmorphisms, cleft palate, sudden cardiac arrest at the age of 3 months, hemi spastic
ZC3H4	NM_015168.1:c.54_71dup, p.(Pro19_Pro24dup)		de novo	6,5	het	de novo	1	NDD + Epilepsy	global developmental delay, faocal epilepsy, truncal ataxia
GIPC1	NM_005716.3:c.718C>Tp.(Arg240*)		de novo	6,3	het	de novo	1	NDD	Intellectual disability, V,a, epilepsy, failure to thrive, short stature, microcephaly, hypotonia, obstipation, strabismus, not able to walk, no language
EIF2AK2	NM_001135651.2:c.1210T>Cp.(Tyr404His)		de novo	7,1	het	de novo	1	NDD + Epilepsy	West syndrome with salaam spasms, hypsarrythmia in EEG, age-appropriate development
CSMD3	NM_052900.2:c.9581A>Cp.(Gln3194Pro)	NM_052900.2:c.7073G>Ap.(Arg2358Gln)	comphet	5,1	comphet	maternal&paternal	1	NDD + Epilepsy	one tonic spasm, developmental delay, 20-30 headdrops per day, hypsarrythmia
MTMR9	NM_015458.3:c.220A>Gp.(Lys74Glu)		de novo	6,2	het	de novo	1	NDD	Developmental delay, club foot, short stature, microcephaly, deafness
BBX	NM_001142568.2:c.2524C>Gp.(Arg842Gly)		homo	7,3	homo	maternal&paternal	2	NDD	mild ID, facial dysmorphisms, Missing anterior teeth in the lower jaw, Vierfingerfurche, dry skin, narrow mouth, behavioral abnormalities
SNX6	NM_021249.4:c.586C>Tp.(Arg196*)		homo	5,6	homo	maternal&paternal	2	NDD	mild ID, facial dysmorphisms, Missing anterior teeth in the lower jaw, simian crease, dry skin, narrow mouth, behavioral abnormalities
MADD	NM_001135943.1:c.2284G>Tp.(Ala762Ser)		homo	9,0	homo	maternal&paternal		NDD + Epilepsy	epileptic encephalopathy, therapy-resistant seizures, pain in legs
CAPS2	NM_032606.3:c.525+1G>Ap.?		de novo	5,7	het	de novo	1	NDD + Epilepsy	mental retardation, epilepsy, no speech development, deafness, short stature

APLN	NM_014499.3:c.416T>Cp.(Phe139Ser)		hemi	4,9	hemi	maternal	1	NDD	developmental delay, speech delay, motor delay, aggressive behaviour, selfharming behaviour, no ID (IQ98)
SLTM	NM_001013843.2:c.2595G>Ap.(Trp865*)		de novo	9,2	het	de novo	1	Neuro	Muscular hypotonia, Hypometric saccades, Chorea, Mild conductive hearing impairment, Constipation, Scapular winging, Gait ataxia, Truncal ataxia, Motor delay
POU3F3	NM_006236.2:c.1220G>Tp.(Arg407Leu)		het	7,5	het	unknown	1	NDD	GDD (first words with 27mo, first steps with >30mo), microcephaly, EEG abnormalities, borad-based gait, strabism, myopia, facial dysmorphism
PPM1G	NM_177983.2:c.1579T>Cp.(Ser527Pro)		homo	4,2	homo	maternal&paternal	1	Metabolism	GDD, adipositas, unbalanced glucosetolerance, massive hyperinsulinamia, muscular hypotonia,
ERN2	NM_033266.3:c.2489C>Tp.(Pro830Leu)		homo	4,4	homo	maternal&paternal	1	NDD	GDD, adipositas, unbalanced glucosetolerance, massive hyperinsulinamia, muscular hypotonia,
PCSK1N	NM_013271.4:c.491T>Cp.(Leu164Pro)		hemi	6,0	hemi	maternal	1	NDD + Epilepsy	familiar epilepsy, speech delay, ADHS
NCOR2	NM_001077261.3:c.7241C>Tp.(Ala2414Val)	7261.3:c.1520_1528dup, p.(Gln507_Gln509dup)	comphet	6,1	comphet	maternal&paternal	1	NDD + Epilepsy	Balkenagenesie, Polymikrogyrie, Plexuszysten, retinale Auffälligkeiten
PCDHA11	NM_018902.4:c.88C>Tp.(Gln30*)		de novo	5,2	het	de novo	2	NDD + Epilepsy	epilepsy with Lennox-Gastaut syndrome and complex retardation
DHRS7	NM_016029.3:c.475A>Gp.(Ile159Val)		de novo	5,3	het	de novo	2	NDD + Epilepsy	epilepsy with Lennox-Gastaut syndrome and complex retardation
EXOC4	NM_001037126.1:c.472-6T>Cp.?	NM_001037126.1:c.860C>Tp.(Ala287Val)	comphet	5,6	comphet	maternal&paternal	1	NDD + Epilepsy	early onset epilepsy, mild developmental delay, decreased glucose in liquor, behavarioal abnormalities
PKP4	001005476.2:c.744_745delp.(Val250Aspfs*110)		de novo	6,8	het	de novo	1	NDD	Microcephaly, Global developmental delay, Global brain atrophy
PTK2B	NM_004103.4:c.1057C>Tp.(Arg353Trp)		het	5,0	het	paternal	1	NDD + Epilepsy	Absence seizures, familiar
DST	NM_001144769.2:c.11762G>Ap.(Arg3921Gln)	NM_001144769.2:c.227C>Ap.(Ala76Glu)	comphet	A	comphet	maternal&paternal	1	Metabolism	hypotrophes Frühgeborenes, Mikrozephalie, Leberversagen, Cholestase, Herzinsuffizienz, Gastrointestinale Blutung, Hypoglykämie, intraventrikuläre Blutung
GABRA2	NM_000807.3:c.438delp.(Arg147Glufs*12)		de novo	A	het	de novo	1	Growth, Skeletal	short stature, muscular hypotonia, micropenis, acromely, hydronephrosis, congenital GH-deficiency, hypogonadism
RGL4	NM_153615.1:c.101C>Tp.(Thr34Met)		de novo	3,5	het	de novo	2	NDD + Epilepsy	Autism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Mental deterioration, Intellectual disability, profound, Generalized seizures, Intellectual disability, moderate, Intellectual disability, borderline, Focal seizures, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptic spasms, Epileptiform EEG discharges, EEG with focal epileptiform discharges, Polymorphic focal epileptiform discharges, EEG with generalized epileptiform discharges, Cognitive impairment, Epileptic
DOCK7	NM_001271999.1:c.2932C>Tp.(Arg978Cys)	NM_001271999.1:c.2464G>Tp.(Ala822Ser)	comphet	6,2	comphet	maternal&paternal	1	NDD	Global developmental delay, postnatal growth retardation, facial dysmorphisms with anteverted nares, smooth philtrum, narrow upper lip and dysplastic ears
STMN3	NM_015894.3:c.19+8C>Ap.?		de novo	3,7	het	de novo	1	NDD + Epilepsy	moderate ID, focal epilepsy, brain atrophy, stair fall
TRIM47	NM_033452.2:c.433C>Tp.(Leu145Phe)		de novo	7,0	het	de novo	2	NDD + Epilepsy	Structural focal epilepsy with secondary generalized tonic-clonic seizures, first seizures with 3 month, right frontal polymicrogyria, small visceral cranium, right convex thoracic scoliosis, moderate mental retardation with behavioral abnormalities, absent speech, latent left-sided pareses
DNAJC17	NM_018163.2:c.273G>Tp.(Glu91Asp)		de novo	5,2	het	de novo	1	NDD + Epilepsy	Hearing abnormality, Hearing impairment, Sensorineural hearing impairment, Strabismus, Psychosis, Osteoporosis, Intellectual disability, Seizures, Intellectual disability, mild, Mental deterioration, Generalized tonic-clonic seizures, EEG abnormality, Kyphosis, Type II diabetes mellitus, Intellectual disability, progressive, Intellectual disability, borderline, Severe hearing impairment, Cognitive impairment
ARAP2	NM_015230.3:c.4037G>Ap.(Arg1346Gln)	NM_015230.3:c.2257G>Cp.(Glu753Gln)	comphet	3,2	comphet	maternal&paternal	1	NDD	N/A
BHLHE41	NM_030762.2:c.1222G>Cp.(Ala408Pro)		de novo	5,4	het	de novo	1	NDD + Epilepsy	Seizures, Febrile seizures, Childhood onset, Epileptic encephalopathy
SLC29A1	NM_001078174.1:c.766+5G>Ap.?	NM_001078174.1:c.1357C>Tp.(Arg453Trp)	comphet	5,2	comphet	maternal&paternal	1	NDD + Epilepsy	Autism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Mental deterioration, Intellectual disability, profound, Generalized seizures, Intellectual disability, moderate, Intellectual disability, borderline, Focal seizures, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptic spasms, Epileptiform EEG discharges, EEG with focal epileptiform discharges, Polymorphic focal epileptiform discharges, EEG with generalized epileptiform discharges, Cognitive impairment, Epileptic
GPKOW	NM_015698.5:c.511A>Gp.(Met171Val)		hemi	6,1	hemi	maternal	1	NDD + Epilepsy	ID, focal epilepsy, motor delay, speech delay, autism, behavioral abnormalities
CNP	NM_033133.4:c.44A>Gp.(Lys15Arg)		homo	8,9	homo	maternal&paternal	1	NDD	schwere Intelligenzminderung, spricht nur Einzelworte, eingeschränktes Sprachverständnis, körperlich gesund
MBD2	NM_003927.4:c.107G>Tp.(Gly36Val)		de novo	7,3	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Focal seizures, Atonic seizures, Intellectual disability, severe, Epileptic spasms, Epileptic encephalopathy
MEGF11	NM_032445.2:c.3080T>Gp.(Leu1027Arg)	NM_032445.2:c.254G>Cp.(Arg85Thr)	comphet	4,4	comphet	maternal&paternal	1	NDD + Epilepsy	Seizures, Prolonged QT interval, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Recurrent infections, Focal seizures
ANKRD33B	NM_001164440.1:c.784G>Ap.(Glu262Lys)	NM_001164440.1:c.1421A>Cp.(Glu474Ala)	comphet	3,3	comphet	maternal&paternal	1	NDD	Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, moderate, Intellectual disability, severe
SEL1L	NM_001244984.1:c.149C>Tp.(Thr50Ile)		de novo	5,5	het	de novo	1	NDD + Epilepsy	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Intellectual disability, severe

NSD2	NM_001042424.2:c.3295G>Ap.(Glu1099Lys)		de novo	10,3	het	de novo	1	NDD	Cryptorchidism, Renal dysplasia, Phenotypic abnormality, Nephrocalcinosis, Delayed speech and language development, Global developmental delay, Motor delay, Cholestasis, Patent ductus arteriosus, Splenomegaly, Pyloric stenosis, Splenic cyst
TNRC18	M_001080495.2:c.7518dup, p.(Ala2507Argfs*44)		de novo	10,2	het	de novo	1	NDD	Microcephaly, Intellectual disability, Global developmental delay, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay
DOCK7	NM_001271999.1:c.2977T>Cp.(Trp993Arg)	NM_001271999.1:c.708delp.(Phe236Leufs*13)	comphet	8,0	comphet	maternal&paternal	3	NDD + Epilepsy	Microcephaly, Strabismus, Ptosis, Hypermetropia, Nystagmus, Behavioral abnormality, Autism, Stereotypy, Seizures, Global developmental delay, Absent speech, Abnormal facial shape, Hypoplasia of the corpus callosum, Intellectual disability, profound, Intellectual disability, moderate, Poor speech, Short stature, Intellectual disability, severe, Epileptic spasms, Epileptic encephalopathy
ZNF219	001101672.1:c.673_678delp.(Ala225_Pro226del)		de novo	5,5	het	de novo	3	NDD + Epilepsy	Microcephaly, Strabismus, Ptosis, Hypermetropia, Nystagmus, Behavioral abnormality, Autism, Stereotypy, Seizures, Global developmental delay, Absent speech, Abnormal facial shape, Hypoplasia of the corpus callosum, Intellectual disability, profound, Intellectual disability, moderate, Poor speech, Short stature, Intellectual disability, severe, Epileptic spasms, Epileptic encephalopathy
DCDC1	NM_181807.3:c.515G>Ap.(Arg172Lys)		de novo	4,6	het	de novo	3	NDD + Epilepsy	Microcephaly, Strabismus, Ptosis, Hypermetropia, Nystagmus, Behavioral abnormality, Autism, Stereotypy, Seizures, Global developmental delay, Absent speech, Abnormal facial shape, Hypoplasia of the corpus callosum, Intellectual disability, profound, Intellectual disability, moderate, Poor speech, Short stature, Intellectual disability, severe, Epileptic spasms, Epileptic encephalopathy
ZSWIM5	NM_020883.1:c.2369G>Ap.(Arg790His)	NM_020883.1:c.1915C>Tp.(Pro639Ser)	comphet	3,4	comphet	maternal&paternal	1	NDD + Epilepsy	Microcephaly, Hearing impairment, Myopia, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Febrile
APOLD1	1_001130415.1:c.755_756delp.(Glu252Valfs*20)		homo	7,6	homo	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Epileptic spasms, Seizures, Focal seizures with impairment of consciousness or awareness, Focal motor seizures, Abnormality of brain morphology, Neuroepithelial neoplasm
AVPR1A	NM_000706.4:c.164T>Ap.(Ile55Asn)		homo	7,9	homo	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Epileptic spasms, Seizures, Focal seizures with impairment of consciousness or awareness, Focal motor seizures, Abnormality of brain morphology, Neuroepithelial neoplasm
HELZ2	NM_001037335.2:c.6691+4C>Tp.?		homo	5,5	homo	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Epileptic spasms, Seizures, Focal seizures with impairment of consciousness or awareness, Focal motor seizures, Abnormality of brain morphology, Neuroepithelial neoplasm
NUSAP1	NM_001243142.1:c.808C>Tp.(Arg270Cys)		homo	5,2	homo	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Epileptic spasms, Seizures, Focal seizures with impairment of consciousness or awareness, Focal motor seizures, Abnormality of brain morphology, Neuroepithelial neoplasm
STARD7	NM_020151.3:c.64C>Tp.(Leu22Phe)		de novo	4,8	het	de novo	1	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Absence seizures
PIGC	NM_002642.3:c.422C>Tp.(Thr141Ile)	NM_002642.3:c.138C>Ap.(Tyr46*)	comphet	7,6	comphet	maternal&paternal	2	NDD + Epilepsy	Macrocephaly, Hypotelorism, Intellectual disability, Seizures, Intellectual disability, mild, Obesity, Large for gestational age, Dilated cardiomyopathy, Absence seizures, Focal seizures with impairment of consciousness or awareness, Diffuse cerebellar atrophy
PHRF1	NM_020901.3:c.1451+3G>Ap.?	NM_020901.3:c.3544A>Gp.(Lys1182Glu)	comphet	3,6	comphet	maternal&paternal	2	NDD + Epilepsy	Macrocephaly, Hypotelorism, Intellectual disability, Seizures, Intellectual disability, mild, Obesity, Large for gestational age, Dilated cardiomyopathy, Absence seizures, Focal seizures with impairment of consciousness or awareness, Diffuse cerebellar atrophy
SYNJ2	NM_003898.3:c.107C>Ap.(Ala36Asp)		de novo	6,8	het	de novo	1	NDD + Epilepsy	Restlessness, Intellectual disability, Hypsarrhythmia, Epileptic spasms, Infantile spasms, I
ANO4	NM_178826.3:c.868G>Ap.(Ala290Thr)		homo	5,2	homo	maternal&paternal	2	NDD + Epilepsy	Seizures, Focal seizures, Multifocal epileptiform discharges, EEG with focal epileptiform discharges, Polymorphic focal epileptiform discharges, EEG with focal spikes
TAF7L	NM_001168474.1:c.1100A>Gp.(Gln367Arg)		hemi	4,0	hemi	maternal	2	NDD + Epilepsy	Seizures, Focal seizures, Multifocal epileptiform discharges, EEG with focal epileptiform discharges, Polymorphic focal epileptiform discharges, EEG with focal spikes
ZC3H12B	NM_001010888.3:c.899A>Gp.(Asn300Ser)		hemi	5,2	hemi	maternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Ataxia
NUCB2	NM_005013.2:c.88_91delp.(Asp30Argfs*15)		de novo	6,2	het	de novo	3	NDD + Epilepsy	Intellectual disability, Seizures, Ataxia
NCOA7	NM_001122842.2:c.2660+2T>Ap.?		de novo	5,7	het	de novo	1	NDD	Behavioral abnormality, Delayed speech and language development, Global developmental delay
BSN	NM_003458.3:c.11163C>Gp.(Ser3721Arg)		de novo	8,7	het	de novo	1	NDD + Epilepsy	Seizures, Hemiplegia, Developmental stagnation, Epileptic encephalopathy, Global developmental delay, Abnormality of movement, Progressive extrapyramidal movement disorder, Reduced consciousness/confusion, Epileptiform EEG discharges
DNAH17	NM_173628.3:c.11678-7_11678-3delp.?	NM_173628.3:c.9998C>Tp.(Ser3333Leu)	comphet	3,9	comphet	maternal&paternal	1	NDD + Epilepsy	Seizures, Hemiplegia, Developmental stagnation, Epileptic encephalopathy, Global developmental delay, Abnormality of movement, Progressive extrapyramidal movement disorder, Reduced consciousness/confusion, Epileptiform EEG discharges
ASTN2	NM_014010.4:c.1013A>Gp.(Lys338Arg)	NM_014010.4:c.872A>Tp.(Asp291Val)	comphet	6,2	comphet	maternal&paternal	1	NDD + Epilepsy	Ptosis, Seizures, Epileptic encephalopathy
EZH2	NM_001203247.1:c.2197G>Ap.(Ala733Thr)		de novo	9,8	het	de novo	3	NDD	Microcephaly, Delayed speech and language development, Global developmental delay, Abnormal facial shape, Severe short stature, Short stature, Intellectual disability
TCP11	NM_001093728.2:c.1440T>Ap.(Phe480Leu)		de novo	4,4	het	de novo	3	NDD	Microcephaly, Delayed speech and language development, Global developmental delay, Abnormal facial shape, Severe short stature, Short stature, Intellectual disability
TNMD	NM_022144.2:c.145G>Tp.(Gly49Trp)		de novo	4,6	het	de novo	3	NDD	Microcephaly, Delayed speech and language development, Global developmental delay, Abnormal facial shape, Severe short stature, Short stature, Intellectual disability

FRYL	NM_015030.1:c.1224delp.(Lys409Argfs*15)		de novo	9,7	het	de novo	1	NDD	Abnormality of the dentition, Cleft palate, Cleft soft palate, Microcephaly, Dental crowding, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Abnormal facial shape, Mild short stature, Proportionate short stature, Short stature, Decreased body weight, Cleft hard palate, Abnormality of cardiovascular system morphology
ZMYM2	NM_001190964.2:c.2338C>Tp.(Arg780*)		de novo	9,9	het	de novo	1	NDD + Epilepsy	Seizures, Focal seizures, Generalized myoclonic seizures, Focal myoclonic seizures, Segmental myoclonic seizures, Falls, Frequent falls
NRXN2	NM_015080.3:c.4484delp.(Phe1495Serfs*71)		het	10,6	het	unknown	1	NDD	Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Abnormal facial shape, Intellectual disability, moderate
SLIT1	NM_003061.2:c.4378C>Tp.(Arg1460Trp)		homo	6,8	homo	maternal&paternal	1	NDD	Intellectual disability, movement disorder
RFX7	NM_022841.5 :c.3083C>T p.(Pro1028Leu)		de novo	6,7	het	de novo	1	NDD + Epilepsy	Congenital cataract, Optic nerve hypoplasia, Delayed speech and language development, Intellectual disability, Seizures, Apnea, Generalized myoclonic seizures, Abnormality of the basal ganglia, Delayed CNS myelination, Sleep disturbance, Focal seizures with impairment of consciousness or awareness, Abnormality of brain morphology, Abnormal myelination, Delayed myelination, Infantile spasms, Abnormality of movement
KLHDC9	NM_152366.4:c.886+1G>Cp.?		homo	8,2	homo	maternal&paternal	1	NDD + Epilepsy	Seizures, Global developmental delay, Generalized hypotonia, Abnormality of the cerebellum, Abnormal facial shape, Generalized seizures
CASKIN1	NM_020764.3 :c.3091C>Tp.(Arg1031Cys)		homo	6,0	homo	maternal&paternal	1	NDD	Speech delay, mild intellectual disability, non-verbal IQ of 57, no prominent facial dysmorphism, stereotypic behavior
TMEM121B	NM_031890.3:c.254T>Cp.(Val85Ala)		de novo	4,5	het	de novo	2	NDD	Hydrocephalus, Hand polydactyly, Postaxial hand polydactyly, Megalencephaly, Foot polydactyly, Postaxial foot polydactyly, Polymicrogyria, Polymicrogyria, anterior to posterior gradient, Severe hydrocephalus, Unilateral polymicrogyria, Frontoparietal polymicrogyria, Polydactyly, Perisylvian polymicrogyria, Postaxial polydactyly
FAM13B	NM_016603.3:c.2203G>Ap.(Val735Ile)	NM_016603.3:c.110A>Gp.(Glu37Gly)	comphet	4,7	comphet	maternal&paternal	2	NDD	Hydrocephalus, Hand polydactyly, Postaxial hand polydactyly, Megalencephaly, Foot polydactyly, Postaxial foot polydactyly, Polymicrogyria, Polymicrogyria, anterior to posterior gradient, Severe hydrocephalus, Unilateral polymicrogyria, Frontoparietal polymicrogyria, Polydactyly, Perisylvian polymicrogyria, Postaxial polydactyly
MDGA2	NM_001113498.2:c.794T>Ap.(Val265Asp)		de novo	7,7	het	de novo	3	NDD	Aggressive behavior, Autistic behavior, Short attention span, Intellectual disability, mild, Global developmental delay, Specific learning disability, Leukemia, Acute leukemia, Short stature, Lymphoid leukemia, Chronic leukemia, Intellectual disability, borderline, Abnormal aggressive, impulsive or violent behavior, Attention deficit hyperactivity disorder, Myeloid leukemia
ACAD10	NM_001136538.1:c.1670C>Gp.(Pro557Arg)	NM_001136538.1:c.3230A>Gp.(His1077Arg)	comphet	5,4	comphet	maternal&paternal	3	NDD	Aggressive behavior, Autistic behavior, Short attention span, Intellectual disability, mild, Global developmental delay, Specific learning disability, Leukemia, Acute leukemia, Short stature, Lymphoid leukemia, Chronic leukemia, Intellectual disability, borderline, Abnormal aggressive, impulsive or violent behavior, Attention deficit hyperactivity disorder, Myeloid leukemia
MAGEA10	NM_001011543.2:c.125C>Tp.(Thr42Ile)		hemi	4,9	hemi	maternal	3	NDD	Aggressive behavior, Autistic behavior, Short attention span, Intellectual disability, mild, Global developmental delay, Specific learning disability, Leukemia, Acute leukemia, Short stature, Lymphoid leukemia, Chronic leukemia, Intellectual disability, borderline, Abnormal aggressive, impulsive or violent behavior, Attention deficit hyperactivity disorder, Myeloid leukemia
STPG2	NM_174952.2:c.1128T>Ap.(Ser376Arg)	NM_174952.2:c.431G>Ap.(Gly144Asp)	comphet	3,8	comphet	maternal&paternal	2	NDD + Epilepsy	Delayed speech and language development, Seizures, Focal seizures, Multifocal epileptiform discharges, EEG with focal epileptiform discharges, Epileptic encephalopathy
CACNA1B	NM_000718.3:c.1442C>Tp.(Ala481Val)		de novo	11,0	het	de novo	2	NDD + Epilepsy	Seizures, Global developmental delay, Hypoglycorrhachia, Hyperglycorrhachia
DGKQ	NM_001347.3:c.1736A>Tp.(His579Leu)	NM_001347.3:c.1408C>Tp.(Arg470Trp)	comphet	4,8	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Global developmental delay, Hypoglycorrhachia, Hyperglycorrhachia
TACC2	NM_206862.3:c.65_66insCCTCp.(Gln23Leufs*22)	NM_206862.3:c.7801C>Tp.(Pro2601Ser)	comphet	5,4	comphet	maternal&paternal	3	NDD	hydrocephalus, intellectual disability, spastic paraplegia, global developmental delay, agenesis of corpus callosum, spastic paraparesis, intellectual disability, severe, cerebral
GET4	NM_015949.2:c.491A>Gp.(Tyr164Cys)		homo	5,9	homo	maternal&paternal	3	NDD	hydrocephalus, intellectual disability, spastic paraplegia, global developmental delay, agenesis of corpus callosum, spastic paraparesis, intellectual disability, severe, cerebral
CDCA2	NM_152562.3:c.922A>Gp.(Arg308Gly)	NM_152562.3:c.1634C>Tp.(Thr545Ile)	comphet	3,4	comphet	maternal&paternal	3	NDD	hydrocephalus, intellectual disability, spastic paraplegia, global developmental delay, agenesis of corpus callosum, spastic paraparesis, intellectual disability, severe, cerebral
DZIP3	NM_014648.3:c.209C>Tp.(Pro70Leu)		de novo	6,6	het	de novo	1	NDD	Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Poor speech, Cognitive impairment
DDB1	NM_001923.4:c.563G>Ap.(Arg188Gln)		de novo	8,8	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Status epilepticus, Intellectual disability, severe, Epileptiform EEG discharges, EEG with focal sharp slow waves, EEG with generalized sharp slow waves, EEG with occipital sharp slow waves, EEG with parietal sharp slow waves, EEG with temporal sharp slow waves, EEG with frontal sharp slow waves, EEG with central sharp slow waves, EEG with occipital sharp waves, EEG with parietal sharp
MTCL1	NM_015210.3:c.604A>Gp.(Thr202Ala)	NM_015210.3:c.1607T>Cp.(Ile536Thr)	comphet	4,7	comphet	maternal&paternal	1	NDD + Epilepsy	Microcephaly, Hearing impairment, Intellectual disability, Seizures, Intellectual disability, mild, Spasticity, Global developmental delay, Polyhydramnios, Intellectual disability, profound, Intellectual disability, moderate, Respiratory failure, Short stature, Intellectual disability, progressive, Intellectual disability, borderline, Intellectual disability, severe, Gait imbalance, Gait disturbance, Abnormality of movement, Severe short stature

MPP3	NM_001330233.1:c.742C>Tp.(Arg248Cys)		homo	4,7	homo	maternal&paternal	1	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity
VGF	NM_003378.3:c.1318G>Ap.(Glu440Lys)		homo	6,3	homo	maternal&paternal	4	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity
ZNF182	NM_001178099.1:c.1319C>Tp.(Thr440Met)		hemi	4,7	hemi	maternal	4	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity
CA5B	NM_007220.3:c.352_354dup, p.(Gly118dup)		hemi	4,4	hemi	maternal	4	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity
KDM5A	NM_001042603.2:c.4048C>Tp.(Arg1350*)		de novo	11,5	het	de novo	1	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Focal clonic seizures, Focal seizures with impairment of consciousness or awareness, Intellectual disability, severe, Focal motor seizures, Focal tonic seizures
KCNN2	NM_021614.3:c.1082A>Gp.(Tyr361Cys)		de novo	7,8	het	de novo	2	NDD	Myopia, Nystagmus, Stereotypy, Delayed speech and language development, Intellectual disability, Motor delay, Absent speech, Abnormality of the foot, Intellectual disability, profound, Difficulty walking, Poor speech, Equinus calcaneus, Vertical nystagmus, Intellectual disability, severe, Severe global developmental delay, Pschomotor retardation
TNK2	NM_001010938.1:c.278T>Gp.(Leu93Arg)		de novo	7,6	het	de novo	2	NDD	Myopia, Nystagmus, Stereotypy, Delayed speech and language development, Intellectual disability, Motor delay, Absent speech, Abnormality of the foot, Intellectual disability, profound, Difficulty walking, Poor speech, Equinus calcaneus, Vertical nystagmus, Intellectual disability, severe, Severe global developmental delay, Pschomotor retardation
PSD3	NM_015310.3:c.3092A>Gp.(Glu1031Gly)	NM_015310.3:c.2929-3C>Tp.?	comphet	5,8	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Focal seizures, Intellectual disability, severe
ARMC3	NM_173081.4:c.1346G>Ap.(Arg449His)		homo	3,4	homo	maternal&paternal	2	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Focal seizures, Intellectual disability, severe
SRGAP3	NM_014850.3:c.2227+6_2227+9delp.?		de novo	C	het	de novo	1	Malformations	Premature birth, Esophageal atresia, Spina bifida, Total anomalous pulmonary venous
CSMD1	NM_033225.5:c.3641T>Cp.(Leu1214Pro)		de novo	7,7	het	de novo	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Intellectual disability, severe, Epileptiform EEG discharges, Neurodevelopmental delay, Epileptic encephalopathy, Myoclonic absences, EMG: myotonic discharges, Generalized tonic-clonic seizures
MFAP1	NM_005926.2:c.88T>Cp.(Ser30Pro)		de novo	6,8	het	de novo	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Intellectual disability, severe, Epileptiform EEG discharges, Neurodevelopmental delay, Epileptic encephalopathy, Myoclonic absences, EMG: myotonic discharges, Generalized tonic-clonic seizures
DPY19L4	NM_181787.2:c.1256C>Tp.(Ser419Phe)	NM_181787.2:c.1870C>Tp.(Arg624*)	comphet	3,5	comphet	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Intellectual disability, severe, Epileptiform EEG discharges, Neurodevelopmental delay, Epileptic encephalopathy, Myoclonic absences, EMG: myotonic discharges, Generalized tonic-clonic seizures
SPTAN1	NM_001130438.2:c.2612delp.(Lys871Serfs*5)		de novo	13,4	het	de novo	4	NDD + Epilepsy	Intellectual disability,Global developmental delay, Motor delay, Developmental regression
AP3B2	NM_001278512.1:c.2879A>Gp.(Asn960Ser)	NM_001278512.1:c.2662G>Ap.(Glu888Lys)	comphet	8,3	comphet	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Developmental regression, Intellectual disability, progressive, Intellectual disability, borderline, Intellectual disability, severe, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Delayed social development, Profound global developmental delay, Neurodevelopmental delay, Cognitive impairment
EIF3B	NM_001037283.1:c.2120G>Ap.(Arg707Gln)		de novo	7,6	het	de novo	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Developmental regression, Intellectual disability, progressive, Intellectual disability, borderline, Intellectual disability, severe, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Delayed social development, Profound global developmental delay, Neurodevelopmental delay, Cognitive impairment
PRRG3	NM_024082.3:c.572C>Tp.(Pro191Leu)		hemi	3,5	hemi	maternal	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Developmental regression, Intellectual disability, progressive, Intellectual disability, borderline, Intellectual disability, severe, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Delayed social development, Profound global developmental delay, Neurodevelopmental delay, Cognitive impairment
USP20	NM_001008563.4:c.582delp.(Lys194Asnfs*46)		de novo	5,5	het	de novo	3	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Expressive language delay, Delayed fine motor development, Intellectual disability, severe

FAM171A1	NM_001010924.1:c.2435C>Tp.(Ala812Val)		homo	4,8	homo	maternal&paternal	3	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Expressive language delay, Delayed fine motor development, Intellectual disability, severe
LCN15	NM_203347.1:c.399C>Ap.(Ser133Arg)		homo	3,8	homo	maternal&paternal	3	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Expressive language delay, Delayed fine motor development, Intellectual disability, severe
ADAM11	NM_002390.5:c.98G>Tp.(Trp33Leu)		de novo	6,6	het	de novo	1	NDD	Strabismus, Hypermetropia, Delayed speech and language development, Intellectual disability, Seizures, Global developmental delay, Absent speech, Absence seizures, Febrile seizures, Receptive language delay
GABRE	NM_004961.3:c.41T>Cp.(Leu14Ser)		hemi	5,2	hemi	maternal	1	NDD	Strabismus, Myopia, Autistic behavior, Anxiety, Hyperactivity, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Abnormal fear/anxiety-related behavior
HMGXB3	NM_014983.2:c.2026C>Tp.(Pro676Ser)		de novo	6,1	het	de novo	1	NDD	Delayed speech and language development, Intellectual disability, Global developmental delay, Expressive language delay
RAB11FIP2	NM_001330167.1:c.1334T>Cp.(Met445Thr)		de novo	5,9	het	de novo	2	NDD	kombinierte Entwicklungsverzögerung/Lernbehinderung (IQ=69), leichtes Übergewicht, faziale Dysmorphie, kurze Finger, Brachyzephalus, CA und FRAX unauffällig, Array: Dup1q31.1 mat, Dup11q14.1 mat
KDM2B	NM_032590.4:c.2345C>Tp.(Ser782Leu)		homo	7,1	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular
UNC5D	NM_080872.3: c.977A>Gp.(His326Arg)		homo	6,5	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular
RNF10	NM_001330474.1:c.850C>Tp.(His284Tyr)		homo	5,8	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular
PCLO	NM_033026.5 :c.13206G>T p.(Gln4402His)	NM_033026.5:c.1297G>Ap.(Ala433Thr)	comphet	5,9	comphet	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular
DOCK1	NM_001380.4:c.4546A>Gp.(Ser1516Gly)		homo	6,3	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular
SF3B2	NM_006842.2:c.76G>Ap.(Ala26Thr)		homo	5,1	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular
PCDHA9	_031857.1:c.1134_1135delCGinsTTp.(Ala379Ser)		homo	4,6	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular
MRPL15	NM_014175.3:c.743C>Tp.(Thr248Ile)		homo	6,0	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular
TCP11	NM_001093728.2:c.256A>Gp.(Lys86Glu)		homo	3,4	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular

TFE3	NM_006521.5:c.566A>Gp.(Tyr189Cys)		de novo	7,5	hemi	de novo	1	NDD + Epilepsy	Microcephaly, Myopia, Delayed speech and language development, Abnormality of the thumb, Intellectual disability, Seizures, Intellectual disability, mild, Spasticity, Global developmental delay, Mental deterioration, Motor delay, Absent speech, Hip dysplasia, Obesity, Small for gestational age, Short nail, Broad nail, Abnormal facial shape, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Intellectual disability, profound, Hepatomegaly, Intellectual disability, moderate, EEG abnormality, Poor speech, Mild short stature, Short stature, Increased body weight, Precocious puberty in males, Moderately short stature, Generalized tonic seizures, Intellectual disability, severe, Epileptic spasms, Myoclonic atonic seizures, Broad thumb, Cerebral palsy, Cognitive impairment
KDM2A	NM_012308.2:c.956G>Ap.(Arg319Gln)		de novo	9,4	het	de novo	3	NDD + Epilepsy	Narrow mouth, Upslanted palpebral fissure, Delayed speech and language development, Intellectual disability, Global developmental delay, Pachygyria, Lissencephaly, Absent speech, Dysphagia, Polymicrogyria, Status epilepticus, Gliosis, Intellectual disability, moderate, Cerebellar malformation, Poor speech, Abnormality of the cerebral white matter, Excessive salivation, Focal white matter lesions, Focal seizures, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptic spasms, EEG with focal epileptiform discharges, Cerebral white matter atrophy, Cerebral white matter agenesis, Oral-pharyngeal
MARCHF6	NM_005885.3:c.1108T>Cp.(Tyr370His)	NM_005885.3:c.1897-3C>Tp.?	comphet	4,3	comphet	maternal&paternal	1	NDD + Epilepsy	global development delay, seizures, microcephaly, autism, single transverse palmar crease, broad palm, abnormal fracial shape
RSRC2	NM_023012.5:c.603-8T>Cp.?		de novo	4,0	het	de novo	3	NDD + Epilepsy	Behavioral abnormality, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic-clonic seizures, Absence seizures, Generalized myoclonic seizures, Intellectual disability, moderate, Generalized tonic seizures, Atonic seizures, Cognitive impairment
WDR59	NM_030581.3:c.2326G>Tp.(Val776Leu)	NM_030581.3:DelExons19-25	comphet	4,4	comphet	maternal&paternal	3	NDD + Epilepsy	Behavioral abnormality, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic-clonic seizures, Absence seizures, Generalized myoclonic seizures, Intellectual disability, moderate, Generalized tonic seizures, Atonic seizures, Cognitive impairment
WWC3	NM_015691.3:c.2935C>Tp.(Arg979Trp)		hemi	4,2	hemi	maternal	3	NDD + Epilepsy	Behavioral abnormality, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic-clonic seizures, Absence seizures, Generalized myoclonic seizures, Intellectual disability, moderate, Generalized tonic seizures, Atonic seizures, Cognitive impairment
OPCML	NM_001012393.2:c.175delp.(Val59Trpfs*4)		de novo	7,1	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic-clonic seizures, Focal clonic seizures, Intellectual disability, moderate, Focal seizures with impairment of consciousness or awareness, Intellectual disability, borderline, Generalized tonic seizures, Symptomatic seizures, Focal tonic seizures, Cognitive
PRKCA	NM_002737.2:c.64C>Tp.(Arg22Cys)		de novo	8,6	het	de novo	4	NDD	Tall stature, Strabismus, Esotropia, Precocious puberty, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Curly hair, Scoliosis
SRRT	NM_015908.5:c.437C>Tp.(Pro146Leu)		de novo	8,4	het	de novo	4	NDD	Tall stature, Strabismus, Esotropia, Precocious puberty, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Motor delay, Curly hair, Woolly hair, Intellectual disability, moderate, Scoliosis, Infantile muscular hypotonia, Precocious puberty in females, Proportionate tall stature, Cognitive impairment
KALRN	NM_001024660.4:c.4026-8T>Cp.?	NM_001024660.4:c.5369A>Gp.(Gln1790Arg)	comphet	6,3	comphet	maternal&paternal	4	NDD	Tall stature, Strabismus, Esotropia, Precocious puberty, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Motor delay, Curly hair, Woolly hair, Intellectual disability, moderate, Scoliosis, Infantile muscular hypotonia, Precocious puberty in females, Proportionate tall stature, Cognitive impairment
TRMT1	NM_001136035.2:c.1964G>Ap.(Gly655Glu)		de novo	8,0	het	de novo	4	NDD	Tall stature, Strabismus, Esotropia, Precocious puberty, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Motor delay, Curly hair, Woolly hair, Intellectual disability, moderate, Scoliosis, Infantile muscular hypotonia, Precocious puberty in females, Proportionate tall stature, Cognitive impairment
GNAI1	NM_002069.5:c.143C>Ap.(Thr48Lys)		de novo	9,5	het	de novo	2	NDD + Epilepsy	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Absent speech, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Generalized seizures, Focal clonic seizures, Intellectual disability, moderate, Focal seizures with impairment of consciousness or awareness, Poor speech, Focal seizures, Intellectual disability, severe, Epileptic spasms, Focal motor seizures, Focal tonic seizures, Abnormality of movement,
SLC2A8	NM_014580.4:c.1150G>Ap.(Gly384Ser)	NM_014580.4:c.1239C>Gp.(Cys413Trp)	comphet	4,5	comphet	maternal&paternal	2	NDD + Epilepsy	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Absent speech, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Generalized seizures, Focal clonic seizures, Intellectual disability, moderate, Focal seizures with impairment of consciousness or awareness, Poor speech, Focal seizures, Intellectual disability, severe, Epileptic spasms, Focal motor seizures, Focal tonic seizures, Abnormality of movement,
TRAK2	NM_015049.2:c.1210G>Ap.(Val404Ile)		de novo	6,9	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Intellectual disability, profound, Intellectual disability, moderate, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptiform EEG discharges, Cognitive impairment, Epileptic encephalopathy

TENM1	NM_001163278.1:c.5977A>Tp.(Thr1993Ser)		hemi	6,0	hemi	maternal	1	NDD	Delayed speech and language development, Intellectual disability, Global developmental delay
ACTR5	NM_024855.3:c.958G>Tp.(Asp320Tyr)		homo	6,9	homo	maternal&paternal	3	NDD + Epilepsy	Delayed speech and language development, Seizures, Global developmental delay, Motor delay, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Febrile seizures, Postnatal microcephaly
TENM3	NM_001080477.3:c.2221G>Ap.(Glu741Lys)		de novo	7,8	het	de novo	1	NDD	Autism, Autistic behavior, Intellectual disability, Global developmental delay, Intellectual disability, severe, no speech
ZMYM4	NM_005095.2:c.1300A>Gp.(Thr434Ala)		de novo	6,6	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Generalized tonic-clonic seizures with focal onset, Focal seizures, Epileptic encephalopathy
STX1A	NM_004603.3:c.284-1G>Ap.?		homo	12,9	homo	maternal&paternal	1	NDD	severe ID, decreased fetal movements, muscular hypotonia
MAGI2	NM_012301.3:c.3780C>Ap.Asp1260Glu		homo	8,9	homo	maternal&paternal	1	NDD	mild ID, hypermetropia
TRAP1	NM_016292.2:c.1941-1G>Ap.?		homo	10,0	homo	maternal&paternal	1	NDD	moderate ID, mental deterioration, autism, self-mutilation, muscular hypotonia, nystagmus, leukodystrophy
CCAR2	NM_021174.5:c.2484C>Ap.Tyr828*		homo	9,7	homo	maternal&paternal	2	NDD	moderate ID, small for gestational age, short stature
CLMN	NM_024734.3:c.730C>Tp.Arg244*		homo	7,7	homo	maternal&paternal	1	NDD	moderate ID, muscular hypotonia, gait disturbance, EEG abnormalities, cerebral atrophy
ENO2	NM_001975.2:c.710C>Tp.Thr237Met		homo	8,6	homo	maternal&paternal	1	NDD	mild ID, small for gestational age, short stature, microcephaly
GALNT2	NM_004481.4:c.865C>Tp.Gln289*		homo	9,4	homo	maternal&paternal	1	NDD + Epilepsy	very severe ID, seizures, autism, aggressive behavior, feeding problems in infancy, short stature, constipation, strabismus, inguinal hernia
AMZ2	NM_001033569.1:c.25C>Tp.Gln9*		homo	7,4	homo	maternal&paternal	2	NDD	mild ID, muscular hypotonia, microcephaly, hypospadias, megalocornea, cerebral atrophy
SLC44A1	NM_080546.4:c.377_380delGTGAp.Ser126fs		homo	9,1	homo	maternal&paternal	1	NDD	mild ID, macrocephaly, acanthosis nigricans, accessory mamilla, muscular hypotonia, frontotemporal cerebral atrophy
ICE2	NM_024611.5:c.2764G>Tp.Gly922*		homo	9,4	homo	maternal&paternal	1	NDD + Epilepsy	mild ID, deafness, febrile seizures, EEG abnormalities, atrial septal defect
FAM234B	NM_020853.1:c.1009C>Tp.Gln337*		homo	8,2	homo	maternal&paternal	1	NDD + Epilepsy	mild ID, seizures, obesity, delayed puberty
SEC23IP	NM_007190.3:c.2101G>Tp.Glu701*		homo	8,5	homo	maternal&paternal	1	NDD	severe ID, feeding problems in infancy, microcephaly, non-midline cleft of the upper lip, 1-2 and 3-4 toe syndactyly, broad toes, mirror image dupliction of toes, craniosynostosis, scaphocephaly, hypoplastic corpus callosum, holoprosencephaly, lissencephaly, leukodystrophy, central diabetes insipidus
SV2C	NM_014979.3:c.533G>Cp.Ser178Thr		homo	7,0	homo	maternal&paternal	1	NDD	moderate ID, microcephaly, short stature
PPFIA1	NM_003626.3:c.1070A>Gp.His357Arg		homo	7,9	homo	maternal&paternal	1	NDD	very severe ID, muscular hypotonia, spasticity, resting tremor, abnormality of the thorax, seizures, cerebral atrophy
LRRIQ3	NM_001105659.1:c.968C>Ap.Ser323*		homo	7,1	homo	maternal&paternal	2	NDD	mild ID
INIP	NM_021218.2:c.266delCp.Ala89fs		homo	9,2	homo	maternal&paternal	1	NDD + Epilepsy	mild ID, febrile seizures, recurrent infections, carious teeth, microcephaly, muscular hypotonia, ataxia, myopia
GTF3C3	NM_012086.4:c.1436A>Gp.Tyr479Cys		homo	8,0	homo	maternal&paternal	1	NDD + Epilepsy	mild ID, seizures, recurrent infections, constipation, abnormalities of the face, postaxial hexadactyly, ataxia, radioulnar synostosis, ventricular septal defect, EEG abnormalities
MBNL3	NM_018388.3:c.279delTp.Ala94fs		homo	9,0	homo	maternal&paternal	1	NDD	moderate ID, autism
OGDHL	NM_018245.2:c.2606G>Ap.Arg869Gln		homo	7,2	homo	maternal&paternal	2	NDD	moderate ID, small for gestational age, short stature
CACNA2D1	NM_000722.3:c.1514C>Tp.Thr505Ile		homo	8,7	homo	maternal&paternal	1	NDD	severe ID, muscular hypotonia, stereotypical motor behaviors, inguinal hernia, omphalocele
TMEM132D	NM_133448.2:c.1489A>Gp.Lys497Glu		homo	6,2	homo	maternal&paternal	2	NDD	mild ID
HACL1	NM_012260.3:c.1246C>Gp.His416Asp		homo	7,2	homo	maternal&paternal	1	NDD	severe ID, muscular hypotonia, low-set ears, bifid uvula, cryptorchidism, aplasia cutis congenita, unilateral renal agenesis, cardiac malformation, increased creatine kinase
SPOUT1	NM_016390.3:c.1058C>Tp.Thr353Met		homo	6,6	homo	maternal&paternal	1	NDD + Epilepsy	profound ID, seizures, microcephaly, short stature, limb hypertonia, bruxism
SMURF2	NM_022739.3:c.1921A>Gp.Thr641Ala		homo	8,2	homo	maternal&paternal	2	NDD	mild ID, muscular hypotonia, microcephaly, hypospadias, megalocornea, cerebral atrophy
GRAMD1B	NM_001286563.1:c.586C>Tp.Arg196Trp		homo	7,2	homo	maternal&paternal	1	NDD	moderate ID
PPRC1	NM_015062.4:c.1825C>Tp.Pro609Ser		homo	6,2	homo	maternal&paternal	1	NDD + Epilepsy	severe ID, seizures, cerebral atrophy, leukodystrophy, macular degeneration, abnormality of the retina
BDH1	NM_004051.4:c.668G>Ap.Arg223His		homo	7,2	homo	maternal&paternal	1	NDD + Epilepsy	very severe ID, seizures, muscular hypotonia, limb hypertonia, spasticity, short stature, microcephaly, leukodystrophy
CHD1L	NM_004284.4:c.1175G>Ap.Arg392His		homo	9,0	homo	maternal&paternal	1	NDD	mild ID, microcephaly, muscular hypotonia, rigidity, ataxia, intention tremor, hypopigmented macules, EEG abnormalities
ATP2C2	NM_001286527.2:c.2636A>Gp.Asp879Gly		homo	7,8	homo	maternal&paternal	1	NDD	severe ID, muscular hypotonia of the trunk, spastic paraparesis, preaxial polydactyly, abnormality of muscle fibers, colpocephaly, cerebellar hypoplasia, hypoplasia of the corpus callosum
PARD6A	NM_016948.2:c.934C>Tp.Arg312*		de novo	6,2	het	de novo	1	NDD	mild ID, stereotypical motor behaviors, muscular hypotonia, strabismus, EEG abnormalities
HMG20A	NM_001304504.1:c.694C>Gp.Arg232Gly		homo	6,6	homo	maternal&paternal	1	NDD + Epilepsy	moderate ID, seizures
TSPAN18	NM_130783.4:c.275T>Cp.Leu92Pro		homo	6,4	homo	maternal&paternal	1	NDD	severe ID, deafness
CEP76	NM_024899.3:c.302T>Cp.Ile101Thr		homo	7,6	homo	maternal&paternal	1	NDD	moderate ID, muscular hypotonia, short stature, microcephaly
ADIPOR1	NM_001290553.1:c.644T>Cp.Leu215Pro		homo	6,9	homo	maternal&paternal	1	NDD	very severe ID, EEG abnormalities, microcephaly
GCC2	NM_181453.3:c.3982C>Tp.His1328Tyr		homo	7,6	homo	maternal&paternal	1	NDD	ID, short stature, elbow contractures, wrist contractures, axillar pterygium, abnormalities of the face, deafness, abnormality of thrombocytes
SKIDA1	NM_207371.3:c.2600C>Tp.Ala867Val		homo	6,5	homo	maternal&paternal	1	NDD	severe ID, small for gestational age, strabismus, short stature
LRCH3	NM_032773.3:c.761A>Gp.Gln254Arg		homo	5,8	homo	maternal&paternal	1	NDD + Epilepsy	severe ID, seizures, muscular hypotonia, cardiac malformation, cerebral atrophy
RXRB	NM_001270401.1:c.1091C>Tp.Pro364Leu		homo	7,1	homo	maternal&paternal	1	NDD	very severe ID, short stature, microcephaly

BTN2A2	NM_001197237.1:c.386G>Ap.Cys129Tyr		homo	6,3	homo	maternal&paternal	1	NDD	very severe ID, muscular hypotonia, constipation
TMEM147	NM_032635.3:c.344+5G>Ap.?		homo	5,7	homo	maternal&paternal	1	NDD	very severe ID, impaired vision, joint contractures
LENG8	NM_052925.3:c.2147G>Ap.Arg716Gln		homo	6,2	homo	maternal&paternal	1	NDD	severe ID, mental deterioration, sleep disturbances, behavioral abnormality, hyperpigmented macules, EEG abnormalities
FNDC3A	NM_001079673.1:c.1186G>Ap.Asp396Asn		homo	7,1	homo	maternal&paternal	1	NDD + Epilepsy	severe ID, seizures, muscular hypotonia, short stature
KCTD18	NM_001321547.1:c.875C>Tp.Ser292Leu		homo	5,5	homo	maternal&paternal	1	NDD	moderate ID, short stature, microcephaly, dislocated hips
EIF4A2	NM_001967.3:c.109_111delGATp.Asp37del		homo	7,6	homo	maternal&paternal	1	NDD	mild ID, muscular hypotonia, tremor
ATP6V0A1	NM_001130020.1:c.53A>Tp.(Gln18Leu)		de novo	8,1	het	de novo	1	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Absence seizures, Generalized myoclonic seizures, Status epilepticus, Focal clonic seizures, Focal seizures without impairment of consciousness or awareness, EEG abnormality, Focal seizures with impairment of consciousness or awareness, Hypsarrhythmia, Infantile onset, Focal seizures, Epileptic spasms, Symptomatic seizures, Typical absence seizures, Focal motor seizures, Focal tonic seizures, Infantile spasms, Epileptic encephalopathy, Multifocal seizures
RSRC2	NM_023012.5:c.1271T>Gp.(Phe424Cys)		de novo	6,1	het	de novo	1	NDD	Global developmental delay, Microcephaly, Agenesis of corpus callosum, Failure to thrive, Growth delay, EEG abnormality, Abnormal cry
SNF8	NM_007241.3:c.572G>Ap.(Gly191Asp)	NM_007241.3:c.236C>Tp.(Pro79Leu)	Comphet	5,0	Comphet	maternal&paternal	2	NDD	Global developmental delay, Microcephaly, Agenesis of corpus callosum, Failure to thrive, Growth delay, EEG abnormality, Abnormal cry
ARL13A	NM_001162491.1:c.349G>Cp.(Asp117His)		hemi	3,3	hemi	maternal	1	NDD	Intellectual disability, Global developmental delay, Hemiplegia/hemiparesis
TMEM94	NM_001321148.1:c.2906G>Ap.(Arg969Gln)	NM_001321148.1:c.2978T>Cp.(Met993Thr)	comphet	6,2	comphet	maternal&paternal	1	NDD + Epilepsy	Seizures, Global developmental delay, Focal seizures, Retinoblastoma
AFDN	NM_001207008.1:c.436A>Gp.(Lys146Glu)		het	6,0	het	paternal	2	NDD	Tall stature, Behavioral abnormality, Short attention span, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Attention deficit hyperactivity disorder, Cognitive impairment
TTBK1	NM_032538.2:c.3116_3118delp.(Thr1039del)		het	4,4	het	paternal	2	NDD	Tall stature, Behavioral abnormality, Short attention span, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Attention deficit hyperactivity disorder, Cognitive impairment
ARFGEF3	NM_020340.4:c.421-4A>Gp.?	NM_020340.4:c.2003C>Tp.(Ala668Val)	comphet	5,0	comphet	maternal&paternal	2	Neuro	Abnormality of the corpus callosum, Agenesis of corpus callosum, Talipes equinovarus, Polymicrogyria, Myelomeningocele, Brainstem dysplasia, Dysplastic corpus callosum, Periventricular gray matter heterotopia
COL19A1	NM_001858.5:c.1843G>Ap.(Gly615Ser)		homo	4,7	homo	maternal&paternal	3	ndd	Microcephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
SLC25A35	NM_001320870.1:c.194G>Ap.(Gly65Asp)		homo	4,7	homo	maternal&paternal	3	ndd	Microcephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
GRIK5	NM_001301030.1:c.818C>Ap.(Ser273Tyr)	NM_001301030.1:c.1745G>Ap.(Arg582His)	comphet	8,6	comphet	maternal&paternal	2	NDD + Epilepsy	Strabismus, Single umbilical artery, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Spastic tetraparesis, Absent speech, Generalized myoclonic seizures, Polymicrogyria, Tetraparesis, Intellectual disability, moderate, EEG abnormality, Sleep disturbance, Myoclonic spasms, Unilateral polymicrogyria, Frontoparietal polymicrogyria, Generalized tonic seizures, Epileptic spasms, Focal myoclonic seizures, EEG with generalized spikes, Perisylvian polymicrogyria, Tetraplegia/tetraparesis, Cognitive impairment, Maternal seizures, Abnormal eating behavior, Exodeviation, Segmental myoclonic seizures
GEMIN5	NM_015465.4:c.1627A>Gp.(Ser543Gly)	NM_015465.4:c.851G>Ap.(Arg284His)	comphet	5,3	comphet	maternal&paternal	2	NDD	Cryptorchidism, Microcephaly, Global developmental delay, Motor delay, Growth delay, Intrauterine growth retardation
SLC25A43	NM_145305.2:c.224C>Tp.(Ala75Val)		hemi	6,3	hemi	maternal	2	NDD	Cryptorchidism, Microcephaly, Global developmental delay, Motor delay, Growth delay, Intrauterine growth retardation
TRIM9	NM_015163.5:c.1117G>Ap.(Val373Met)		de novo	8,3	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizures, Muscular hypotonia, Global developmental delay, Mental deterioration, Pes cavus, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Generalized seizures,Leukodystrophy, Abnormality of the cerebral white matter, Infantile spasms
ZMYM2	NM_001190964.2:c.2881G>Cp.(Glu961Gln)		de novo	9,0	het	de novo	2	NDD + Epilepsy	Seizures, Global developmental delay, Episodic ataxia
LAMA5	NM_005560.4:c.6659G>Tp.(Arg2220Leu)	NM_005560.4:c.1246C>Gp.(Pro416Ala)	comphet	5,3	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Global developmental delay, Episodic ataxia
CDH20	NM_031891.3:c.958G>Cp.(Asp320His)		de novo	6,5	het	de novo	5	NDD + Epilepsy	Tall stature, Autism, Precocious puberty, Intellectual disability, Seizures, Global developmental delay, Focal seizures with impairment of consciousness or awareness, Focal seizures, Precocious puberty in males, Increased serum insulin-like growth factor 1
FUNDC1	NM_173794.3:c.154A>Gp.(Thr52Ala)		hemi	6,8	hemi	maternal	5	NDD + Epilepsy	Tall stature, Autism, Precocious puberty, Intellectual disability, Seizures, Global developmental delay, Focal seizures with impairment of consciousness or awareness, Focal seizures, Precocious puberty in males, Increased serum insulin-like growth factor 1
SPSB1	NM_025106.3:c.572T>Cp.(Ile191Thr)		de novo	6,5	het	de novo	5	NDD + Epilepsy	Tall stature, Autism, Precocious puberty, Intellectual disability, Seizures, Global developmental delay, Focal seizures with impairment of consciousness or awareness, Focal seizures, Precocious puberty in males, Increased serum insulin-like growth factor 1
PRSS41	_001135086.1:c.30_41dup, p.(Leu11_Ala14dup)		homo	3,0	homo	maternal&paternal	5	NDD + Epilepsy	Tall stature, Autism, Precocious puberty, Intellectual disability, Seizures, Global developmental delay, Focal seizures with impairment of consciousness or awareness, Focal seizures, Precocious puberty in males, Increased serum insulin-like growth factor 1

RNF44	NM_014901.4:c.802-8T>Gp.?		de novo	4,9	het	de novo	5	NDD + Epilepsy	Tall stature, Autism, Precocious puberty, Intellectual disability, Seizures, Global developmental delay, Focal seizures with impairment of consciousness or awareness, Focal seizures, Precocious puberty in males, Increased serum insulin-like growth factor 1
LAMA5	NM_005560.4:c.10753G>Tp.(Asp3585Tyr)	NM_005560.4:c.1390G>Ap.(Gly464Ser)	comphet	5,7	comphet	maternal&paternal	1	NDD + Epilepsy	Abnormality of the head, Microcephaly, Seizures, Postnatal microcephaly, Loss of consciousness, Atonic seizures
RORB			de novo	10,9	het	de novo	2	NDD	Hearing impairment, Hypermetropia, Nystagmus, Delayed speech and language development, Intellectual disability, Global developmental delay, Motor delay, Generalized tonic-clonic seizures, Short stature, Decreased body weight, Simple febrile seizures
CRYBG1	NM_001624.3:c.4489G>Ap.(Val1497Ile)		de novo	6,1	het	de novo	2	NDD	Hearing impairment, Prelingual sensorineural hearing impairment, Conductive hearing impairment, Sensorineural hearing impairment, Hypermetropia, Nystagmus, Horizontal nystagmus, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Growth delay, Generalized tonic-clonic seizures, Delayed gross motor development, Mild short stature, Proportionate short stature, Short stature, Abnormality of body weight, Decreased body weight, High hypermetropia, Moderate sensorineural hearing impairment, Bilateral conductive hearing impairment, Congenital sensorineural hearing impairment, Bilateral sensorineural hearing impairment, Severe sensorineural hearing impairment, Simple febrile seizures, Mild global developmental delay, Moderate global developmental delay, Severe hearing impairment, Cognitive impairment, Mild hypermetropia, Moderate hypermetropia, Latent
GRK3	NM_005160.3:c.916G>Tp.(Glu306*)		het	6,1	het	maternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Neurodevelopmental delay, Cognitive impairment, Epileptic encephalopathy
TENM1	NM_001163278.1:c.757A>Gp.(Asn253Asp)		het	4,8	het	maternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Neurodevelopmental delay, Cognitive impairment, Epileptic encephalopathy
DNAJC27	NM_016544.2:c.422delp.(His141Leufs*4)		het	5,7	het	maternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Neurodevelopmental delay, Cognitive impairment, Epileptic encephalopathy
MINPP1	NM_004897.4:c.75_94delp.(Leu27Argfs*39)		homo	9,2	homo	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Ataxia, Global developmental delay, Gait ataxia, Olivopontocerebellar atrophy, Short stature, Pontocerebellar atrophy, Olivopontocerebellar hypoplasia, Cognitive impairment
GUCY2F	NM_001522.2:c.1445C>Gp.(Ser482Cys)		homo	5,1	homo	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Ataxia, Global developmental delay, Gait ataxia, Olivopontocerebellar atrophy, Short stature, Pontocerebellar atrophy, Olivopontocerebellar hypoplasia, Cognitive impairment
ANKRD30B	NM_001145029.1:c.1795G>Tp.(Glu599*)		homo	6,2	homo	maternal&paternal	1	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay
UNC5A	NM_133369.2:c.578C>Ap.(Ser193Tyr)	NM_133369.2:c.267C>Gp.(Ile89Met)	comphet	4,7	comphet	maternal&paternal	1	NDD + Epilepsy	Hypermetropia, Intellectual disability, Seizures, Global developmental delay, Absence seizure, Intellectual disability, severe, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay, Cognitive impairment
PIKFYVE	NM_015040.3:c.1319A>Gp.(Gln440Arg)		homo	9,0	homo	maternal&paternal	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
VPS54	NM_016516.2:c.701C>Tp.(Ala234Val)		homo	8,2	homo	maternal&paternal	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
BCAS1	NM_003657.3:c.1720C>Tp.(Pro574Ser)		homo	6,6	homo	maternal&paternal	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
LRIG3	NM_153377.4:c.979G>Ap.(Asp327Asn)		de novo	6,7	het	de novo	1	NDD	Global developmental delay, Absent speech, Myelomeningocele
GDF11	NM_005811.4:c.955dup, p.(Thr319Asnfs*5)		de novo	8,9	het	de novo	3	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Absence seizure, Typical absence seizure, Early onset absence seizures
COPS2	NM_001143887.1:c.37G>Ap.(Glu13Lys)		de novo	8,6	het	de novo	3	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Absence seizure, Typical absence seizure, Early onset absence seizures
GEMIN5	NM_015465.4:c.3340C>Gp.(Leu1114Val)	NM_015465.4:c.2504A>Gp.(Lys835Arg)	comphet	6,6	comphet	maternal&paternal	3	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Absence seizure, Typical absence seizure, Early onset absence seizures
CD99L2	NM_001242614.1:c.541G>Cp.(Gly181Arg)		hemi	3,9	hemi	maternal	1	NDD + Epilepsy	Tall stature, Glaucoma, Growth hormone excess, Intellectual disability, Seizures, Global developmental delay, Obesity, Mitral regurgitation, Abnormal facial shape, Progeroid facial appearance, Focal-onset seizure
RHEB	NM_005614.3:c.47C>Tp.(Ser16Phe)		de novo	7,9	het	de novo	1	NDD + Epilepsy	Tall stature, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, normal MRI

PSMC5	NM_002805.5:c.587delp.(Lys196Argfs*29)		het	8,4	het	maternal	2	NDD	Microcephaly, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Scoliosis, Short stature, Cognitive impairment
NOVA2	NM_002516.3:c.1267G>Cp.(Gly423Arg)		het	5,5	het	maternal	2	NDD	Microcephaly, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Scoliosis, Short stature, Cognitive impairment
PTPRN2	Del(NM_002847.4)-7-157873875-158384503		de novo	6,7	het	de novo	1	NDD	Behavioral abnormality, Autism, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Intellectual disability, moderate, Poor speech, Intellectual disability, borderline
LCN1	NM_001252618.1:c.305A>Gp.(His102Arg)		de novo	3,4	het	de novo	1	NDD	Tall stature, delayed speech and language development, neuroblastoma
ORC3	NM_181837.2:c.419A>Gp.(Asp140Gly)		homo	6,7	homo	maternal&paternal	4	NDD	Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Severe muscular hypotonia, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Intellectual disability, severe, Central hypotonia, Cognitive impairment
SRRM4	NM_194286.3:c.560G>Ap.(Arg187His)	NM_194286.3:c.140C>Tp.(Pro47Leu)	comphet	5,1	comphet	maternal&paternal	1	NDD	Microcephaly, Brachydactyly, Syndactyly, Intellectual disability, Intellectual disability, mild, Motor delay, Hypertonia, Toe syndactyly, Intellectual disability, moderate, 2-3 toe syndactyly, Feeding difficulties, Cognitive impairment, Impaired feeding ability
ALDH8A1	NM_022568.3:c.160G>Tp.(Ala54Ser)		de novo	5,6	het	de novo	1	NDD	Macrocephaly, Global developmental delay, Hepatosplenomegaly, Hypertriglyceridemia, Hepatomegaly, Recurrent infections
FEN1	NM_004111.5:c.140G>Ap.(Arg47His)		homo	6,5	homo	maternal&paternal	1	NDD + Epilepsy	Seizures, Focal impaired awareness seizure, Spherocytosis, Arrhythmia
CX3CR1	NM_001171174.1:c.756delp.(Cys253Alafs*12)		het	6,0	het	maternal	2	Neuro	Familial predisposition, Migraine, EEG abnormality, Episodic hemiplegia, Left hemiplegia
TMEM151B	NM_001137560.1:c.1319T>Ap.(Val440Asp)		de novo	6,3	het	de novo	1	NDD + Epilepsy	Cleft soft palate, Hydrocephalus, Abnormality of the inner ear, Hearing impairment, Iris coloboma, Delayed speech and language development, Macular coloboma, Intellectual disability, Seizures, Global developmental delay, Agenesis of corpus callosum, Dandy-Walker malformation, Abnormal ear morphology
FAM214B	NM_001317991.1:c.588delp.(Ile196Metfs*115)		het	6,5	het	paternal	2	NDD	Intellectual disability
SENP3	NM_015670.5:c.713C>Ap.(Ser238*)		de novo	8,7	het	de novo	3	NDD + Epilepsy	epilepsy with absences and generalized tonic-clonic seizures, severe intellectual disability with autistic traits, low blood pressure, obstipation, normal MRI 2008
BDP1	NM_018429.2:c.6847G>Tp.(Glu2283*)		homo	9,4	homo	maternal&paternal	3	NDD + Epilepsy	Delayed speech and language development, Seizures, Global developmental delay, Motor delay, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Febrile seizures, Postnatal microcephaly, suspected myopia
CHD5	NM_015557.2:c.776C>Gp.(Ser259Cys)	NM_015557.2:c.3650C>Tp.(Thr1217Ile)	comphet	5,8	comphet	maternal&paternal	2	NDD	Delayed speech and language development, Intellectual disability
DENND4B	NM_014856.2:c.319G>Ap.(Val107Met)	NM_014856.2:c.941G>Ap.(Ser314Asn)	comphet	4,3	comphet	maternal&paternal	2	NDD	Delayed speech and language development, Intellectual disability
RHOT2	NM_138769.2:c.586T>Gp.(Ser196Ala)	NM_138769.2:c.1201C>Tp.(Arg401Cys)	comphet	4,9	comphet	maternal&paternal	1	NDD + Epilepsy	spastic tetraparesis, generalized tonic-clonic seizures, microcephaly, polymicrogyria, periventricular gliosis and cysts, global developmental delay
CAPN9	NM_006615.2:c.1591G>Ap.(Ala531Thr)	006615.2:c.1273-1_1287delp.(Cys425Glufs*262)	comphet	7,4	comphet	maternal&paternal	1	NDD	Global developmental delay, Motor delay, Polyneuropathy, Hip dysplasia, Coxa valga, Kyphosis
PTPN12	NM_002835.3:c.89G>Ap.(Arg30Gln)		homo	3,7	homo	maternal&paternal	3	NDD + Epilepsy	Delayed speech and language development, Seizures, Global developmental delay, Motor delay, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Febrile seizures, Postnatal microcephaly
ATP8B2	NM_020452.3:c.1745G>Ap.(Arg582Gln)		de novo	6,4	het	de novo	2	NDD	Intellectual disability, Intellectual disability, mild, Intellectual disability, moderate, Increased body weight, Increased adipose tissue
DIP2A	NM_015151.3:c.410C>Tp.(Ser137Leu)	NM_015151.3:c.2476G>Ap.(Ala826Thr)	comphet	6,0	comphet	maternal&paternal	2	NDD	Intellectual disability, Intellectual disability, mild, Intellectual disability, moderate, Increased body weight, Increased adipose tissue
AKAP13	NM_006738.5:c.742C>Tp.(Arg248*)		de novo	9,9	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Generalized tonic seizures, Epileptic encephalopathy
AKAP17A	NM_005088.2:c.1328T>Cp.(Leu443Pro)		de novo	4,9	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Generalized tonic-clonic seizures, Focal clonic seizures, Intellectual disability, moderate, Increased body weight, Focal-onset seizure, Increased adipose tissue, Generalized tonic seizures, Focal myoclonic seizures, Focal tonic seizures
UTP11	NM_016037.3:c.230A>Gp.(Asp77Gly)		de novo	5,2	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Generalized tonic-clonic seizures, Focal clonic seizures, Intellectual disability, moderate, Increased body weight, Focal-onset seizure, Increased adipose tissue, Generalized tonic seizures, Focal myoclonic seizures, Focal tonic seizures

GPSM3	NM_001276501.1:c.318G>Cp.(Gln106His)		de novo	4,7	het	de novo	2	Neuro	Microcephaly, Edema, Agenesis of corpus callosum, Abnormal cerebellum morphology, Cerebellar hypoplasia, Growth abnormality, Growth delay, Intrauterine growth retardation, Hypoplasia of the corpus callosum, Polymicrogyria, Abnormality of neuronal migration, Gray matter heterotopias, Gray matter heterotopia, Spontaneous abortion, Periventricular heterotopia, White matter neuronal heterotopia, Aplasia/Hypoplasia of the cerebellum, Fetal onset, Small cerebellar cortex
EMC9	NM_016049.3:c.158A>Tp.(His53Leu)		de novo	5,0	het	de novo	2	Neuro	Microcephaly, Edema, Agenesis of corpus callosum, Abnormal cerebellum morphology, Cerebellar hypoplasia, Growth abnormality, Growth delay, Intrauterine growth retardation, Hypoplasia of the corpus callosum, Polymicrogyria, Abnormality of neuronal migration, Gray matter heterotopias, Gray matter heterotopia, Spontaneous abortion, Periventricular heterotopia, White matter neuronal heterotopia, Aplasia/Hypoplasia of the cerebellum, Fetal onset, Small cerebellar cortex
SLC4A7	M_001321103.1:c.249_252delp.(Lys83Asnfs*62)		homo	8,2	homo	maternal&paternal	4	NDD	Adducted thumb, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Pyloric stenosis, Ventriculomegaly, Intellectual disability, moderate, Infantile muscular hypotonia, Feeding difficulties, Cognitive impairment, Impaired feeding ability
SCRN1	NM_001145514.1:c.1106A>Gp.(Lys369Arg)		homo	5,9	homo	maternal&paternal	4	NDD	Adducted thumb, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Pyloric stenosis, Ventriculomegaly, Intellectual disability, moderate, Infantile muscular hypotonia, Feeding difficulties, Cognitive impairment, Impaired feeding ability, VUS in COLQ (31.07.2019)
COL20A1	NM_020882.2:c.3614-8C>Tp.?		de novo	3,9	het	de novo	4	NDD	Adducted thumb, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Pyloric stenosis, Ventriculomegaly, Intellectual disability, moderate, Infantile muscular hypotonia, Feeding difficulties, Cognitive impairment, Impaired feeding ability, VUS in COLQ (31.07.2019)
WTAP	NM_001270531.1:c.463A>Gp.(Lys155Glu)		de novo	6,9	het	de novo	1	NDD	Microcephaly, Delayed speech and language development, Hyperactivity, Global developmental delay, dystrophy, Receptive language delay
GPR161	M_001267609.1:c.1550dup, p.(Gly518Argfs*44)		de novo	7,1	het	de novo	2	NDD	Hypertelorism, Low-set ears, Brachydactyly, Intellectual disability, Global developmental delay, Hypoplasia of the corpus callosum, Elevated serum creatinine, Moderate global developmental delay
TENM2	NM_001122679.1:c.4082A>Gp.(Tyr1361Cys)	NM_001122679.1:c.7924G>Ap.(Val2642Met)	comphet	5,0	comphet	maternal&paternal	2	NDD	Hypertelorism, Low-set ears, Brachydactyly, Intellectual disability, Global developmental delay, Hypoplasia of the corpus callosum, Elevated serum creatinine, Moderate global developmental delay
H3-3A	NM_002107.4:c.250C>Gp.(Arg84Gly)		de novo	9,8	het	de novo	2	NDD + Epilepsy	Stereotypy, Delayed speech and language development, Global developmental delay, Motor delay, Delayed gross motor development, EEG abnormality, Delayed fine motor
CHURC1	M_145165.3:c.349_350insGp.(Leu117Argfs*15)	NM_145165.3:c.400delp.(Arg134Aspfs*3)	comphet	7,8	comphet	maternal&paternal	2	NDD + Epilepsy	Tall stature, Macrocephaly, Delayed speech and language development, Enuresis, Seizures, Global developmental delay, Obesity, Rett syndrome
RGL1	NM_015149.4:c.737C>Gp.(Ser246Cys)		de novo	6,4	het	de novo	2	NDD + Epilepsy	Tall stature, Macrocephaly, Delayed speech and language development, Enuresis, Seizures, Global developmental delay, Obesity, Rett syndrome
USF3	NM_001009899.3:c.1750C>Tp.(Gln584*)		de novo	8,6	het	de novo	1	NDD	muscular hypotonia, developmental delay, normal cMRI, left retinal coloboma
KCND1	NM_004979.5:c.343G>Ap.(Asp115Asn)		de novo	5,5	hemi	de novo	1	NDD + Epilepsy	Epilepsy with absences and eyelid myoclonias, normal cMRI, EEG abnormalities, IQ 85 (low normal), speech delay, obstipation
EFHC1	NM_018100.3:c.323delp.(Pro108Leufs*13)		homo	9,9	homo	maternal&paternal	3	NDD	Microcephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
WWP2	NM_001270453.1:c.491A>Cp.(Glu164Ala)	NM_001270453.1:c.166G>Cp.(Ala56Pro)	comphet	4,6	comphet	maternal&paternal	2	NDD + Epilepsy	Strabismus, Single umbilical artery, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Spastic tetraparesis, Absent speech, Generalized myoclonic seizures, Polymicrogyria, Tetraparesis, Intellectual disability, moderate, EEG abnormality, Sleep disturbance, Myoclonic spasms, Unilateral polymicrogyria, Frontoparietal polymicrogyria, Generalized tonic seizures, Epileptic spasms, Focal myoclonic seizures, EEG with generalized spikes, Perisylvian polymicrogyria, Tetraplegia/tetraparesis, Cognitive impairment, Maternal seizures, Abnormal eating behavior, Exodeviation, Segmental myoclonic seizures
CTBP2	NM_022802.2:c.1192dup, p.(Arg398Profs*68)		het	7,9	het	maternal	3	NDD + Epilepsy	Intellectual disability, epilepsy with generalized tonic-clonic seizures, short attention span
SLIT3	NM_003062.3:c.2818C>Tp.(Arg940Cys)		het	5,1	het	maternal	3	NDD + Epilepsy	Intellectual disability, epilepsy with generalized tonic-clonic seizures, short attention span
CLCC1	NM_001048210.2:c.1324C>Tp.(Leu442Phe)		het	4,0	het	maternal	3	NDD + Epilepsy	Intellectual disability, epilepsy with generalized tonic-clonic seizures, short attention span
ABCA2	NM_001606.4:c.2261T>Cp.(Phe754Ser)		de novo	9,0	het	de novo	3	NDD + Epilepsy	epilepsy, febrile seizures
SF3A3	NM_006802.3:c.1408C>Tp.(Arg470*)		de novo	8,9	het	de novo	3	NDD + Epilepsy	epilepsy with febrile seizures
NLE1	NM_018096.4:c.593A>Gp.(His198Arg)		de novo	6,7	het	de novo	3	NDD + Epilepsy	epilepsy, febrile seizures
CHKA	NM_001277.2:c.1021T>Cp.(Phe341Leu)	NM_001277.2:c.14dup, p.(Cys6Leufs*19)	comphet	7,0	comphet	maternal&paternal	1	NDD + Epilepsy	severe psychomotor retardation, central movement disorder with preference for right-sided extremities, epilepsy with epileptic spasms, microcephaly, tendency to self-harm
ANKRD17	M_032217.4:c.3751_3754delp.(Arg1252Thrfs*6)		de novo	9,9	het	de novo	1	NDD	Cryptorchidism, Microcephaly, Strabismus, Hypermetropia, Behavioral abnormality, Stereotypy, Global developmental delay, Small for gestational age, Short stature

FRYL	NM_015030.1:c.3851T>Gp.(Leu1284Arg)		homo	6,3	homo	maternal&paternal	1	NDD	Cryptorchidism, Hydroureter, Cleft palate, Cleft soft palate, Global developmental delay, Absent septum pellucidum, Polyhydramnios, Premature birth, Abnormal facial shape, Ventriculomegaly, Severe short stature, Short stature, Frontal cortical atrophy, Temporal cortical atrophy, Bilateral cryptorchidism, Moderately short stature, Brain atrophy
ADAMTSL1	NM_001040272.5:c.1316A>Gp.(Lys439Arg)		de novo	5,8	het	de novo	1	NDD + Epilepsy	Global developmental delay, dystonic movements, abnormal EEG, epilepsy, microcephaly, clinodactyly of the 5th finger, pectus excavatum
STARD9	NM_020759.2:c.4624C>Ap.(Leu1542Met)	NM_020759.2:c.1655G>Tp.(Arg552Leu)	comphet	3,3	comphet	maternal&paternal	2	NDD	Seizures, Generalized tonic-clonic seizures, Myoclonic atonic seizures, Epileptic encephalopathy
CRIM1	NM_016441.2:c.2867C>Tp.(Ala956Val)	NM_016441.2:c.1658+4C>Tp.?	comphet	3,5	comphet	maternal&paternal	2	Neuro	Dystonia, Flexion contracture, Difficulty walking, Limb dystonia, Progressive inability to walk, Ankle flexion contracture, Loss of ability to walk in first decade, Inability to walk by childhood/adolescence, Loss of ability to walk, Generalized dystonia
PASK	NM_001252120.1:c.307G>Ap.(Gly103Ser)	NM_001252120.1:c.2713C>Tp.(Arg905Trp)	comphet	4,1	comphet	maternal&paternal	2	Neuro	Dystonia, Flexion contracture, Difficulty walking, Limb dystonia, Progressive inability to walk, Ankle flexion contracture, Loss of ability to walk in first decade, Inability to walk by childhood/adolescence, Loss of ability to walk, Generalized dystonia
KCNK9	NM_001282534.1:c.391C>Tp.(Arg131Cys)		de novo	9,5	het	de novo	1	NDD	muscular hypotonia, tongue fasciculation, motor developmental delay
RASGEF1A	NM_001282862.1:c.346-3delp.?		de novo	C	het	de novo	1	Malformations	Meningocele, Hypoplastic nasal bridge, Increased nuchal translucency, Short fetal femur length
ARL8B	NM_018184.2:c.286A>Tp.(Ile96Leu)		de novo	6,3	het	de novo	2	Neuro	Abnormality of the corpus callosum, Agenesis of corpus callosum, Talipes equinovarus, Polymicrogyria, Myelomeningocele, Brainstem dysplasia, Dysplastic corpus callosum, Periventricular gray matter heterotopia
ITPKA	NM_002220.2:c.1093G>Ap.(Gly365Arg)		homo	5,6	homo	maternal&paternal	2	NDD + Epilepsy	Macrocephaly, Seizures, Global developmental delay, Epileptic encephalopathy
OS9	NM_006812.3:c.1181A>Tp.(Glu394Val)		homo	6,7	homo	maternal&paternal	2	NDD + Epilepsy	Macrocephaly, Seizures, Global developmental delay, Epileptic encephalopathy
DMAP1	NM_001034023.1:c.581G>Ap.(Arg194Gln)	NM_001034023.1:c.670C>Tp.(Arg224*)	comphet	6,8	comphet	maternal&paternal	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormal facial shape, Cognitive impairment
RAPGEF3	NM_001098531.2:c.2312A>Cp.(His771Pro)		de novo	6,6	het	de novo	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormal facial shape, Cognitive impairment
STT3B	NM_178862.2:c.777+4A>Cp.?		de novo	7,7	het	de novo	3	NDD	Hypotelorism, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Single median maxillary incisor, Agenesis of permanent teeth, Abnormality of dental morphology, Reduced number of teeth, Intellectual disability, severe
ARSH	NM_001011719.1:c.339A>Gp.(Ile113Met)		de novo	5,9	het	de novo	3	NDD	Hypotelorism, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Single median maxillary incisor, Agenesis of permanent teeth, Abnormality of dental morphology, Reduced number of teeth, Intellectual disability, severe
KANSL2	NM_017822.3:c.880C>Tp.(His294Tyr)		het	3,7	het	unknown	1	NDD	Intellectual disability, Intellectual disability, moderate, Intellectual disability, severe
ZFP91	NM_053023.4:c.172C>Tp.(Arg58Trp)		de novo	5,2	het	de novo	3	NDD + Epilepsy	neonatal epileptic encephalopathy
VPS52	NM_022553.5:c.10G>Ap.(Ala4Thr)	NM_022553.5:c.92C>Tp.(Ala31Val)	comphet	5,4	comphet	maternal&paternal	3	NDD + Epilepsy	neonatal epileptic encephalopathy
EVI5L	NM_001159944.2:c.841delp.(Leu281fs)		de novo	9,2	het	de novo	2	NDD + Epilepsy	Pallister Hall syndrome with hand and foot polydactyly, cleft palate, seizures, global developmental delay, 4-5 finger syndactyly, hamartoma
HCK	NM_002110.3:c.1547C>Ap.(Thr516Lys)		de novo	6,1	het	de novo	2	NDD + Epilepsy	Pallister Hall syndrome with hand and foot polydactyly, cleft palate, seizures, global developmental delay, 4-5 finger syndactyly, hamartoma
ZFYVE16	NM_014733.4:c.2570C>Tp.(Pro857Leu)		homo	5,5	homo	maternal&paternal	2	NDD + Epilepsy	Global developmental delay, febrile seizures, muscular hypotonia
NARS1	NM_004539.3:c.1067A>Cp.(Asp356Ala)	NM_004539.3:c.1564C>Tp.(Arg522*)	comphet	7,2	comphet	maternal&paternal	2	NDD + Epilepsy	Global developmental delay, febrile seizures, muscular hypotonia
RHOQ	NM_012249.3:c.359G>Ap.(Gly120Glu)		de novo	7,3	het	de novo	1	NDD	Abnormality of the kidney (horseshoe kidney), Delayed speech and language development, Intellectual disability, Global developmental delay, Motor delay, Specific learning disability, Anal atresia, Intellectual disability, Jejunal atresia, Ileal atresia, Premature Abnormality of the pubic hair
SLITRK2	NM_001144003.2:c.2485G>Tp.(Glu829*)		hemi	7,9	hemi	maternal	3	NDD	Severe global developmental delay, sleep disturbance, behavioral abnormalities, flat face
MYRIP	NM_001284423.1:c.383G>Ap.(Arg128His)	NM_001284423.1:c.86G>Ap.(Arg29His)	comphet	4,9	comphet	maternal&paternal	3	NDD	Severe global developmental delay, sleep disturbance, behavioral abnormalities, flat face
AATF	NM_012138.3:c.695-3C>Ap.?		de novo	3,5	het	de novo	3	NDD	Severe global developmental delay, sleep disturbance, behavioral abnormalities, flat face
ZNF280D	NM_001288588.1:c.2532C>Gp.(His844Gln)		de novo	4,5	het	de novo	1	NDD + Epilepsy	Mild intellectual disability, hydrocephalus, abnormality of brain morphology including cortical dysplasia, focal impaired awareness seizure, generalized tonic-clonic seizures with focal onset
PCNX2	NM_014801.3:c.3846C>Ap.(Asp1282Glu)		de novo	5,5	het	de novo	2	NDD	Visual loss, Optic nerve hypoplasia, Global developmental delay, Hypoglycemia, Abnormality of optic chiasm morphology
DNAH14	NM_001373.1:c.13384G>Ap.(Ala4462Thr)	NM_001373.1:c.6100C>Tp.(Arg2034*)	comphet	4,3	comphet	maternal&paternal	2	NDD	Visual loss, Optic nerve hypoplasia, Global developmental delay, Hypoglycemia, Abnormality of optic chiasm morphology
FAM199X	NM_207318.3:c.961T>Ap.(Ser321Thr)		hemi	5,6	hemi	maternal	1	NDD + Epilepsy	Aggressive behavior, Delayed speech and language development, Seizures, Global developmental delay, Absent speech, EEG abnormality, Focal impaired awareness seizure, Focal-onset seizure
NEU4	NM_001167599.2:c.1396T>Cp.(Cys466Arg)	NM_001167599.2:c.407G>Ap.(Arg136His)	comphet	3,6	comphet	maternal&paternal	2	NDD + Epilepsy	Focal impaired awareness seizures since 10/2016, EEG abnormality, delayed speech and language development, aggressive behavior

ARL14EP	NM_152316.2:c.707G>Ap.(Arg236His)		de novo	8,2	het	de novo	2	NDD + Epilepsy	Intellectual disability, Absent speech, Hyperreflexia, Generalized tonic-clonic seizures, Absence seizure, EEG abnormality, Excessive salivation, Brain atrophy, Epileptic encephalopathy
PSMB4	NM_002796.2:c.226G>Ap.(Gly76Ser)		de novo	7,7	het	de novo	2	NDD + Epilepsy	Intellectual disability, Absent speech, Hyperreflexia, Generalized tonic-clonic seizures, Absence seizure, EEG abnormality, Excessive salivation, Brain atrophy, Epileptic encephalopathy
SHANK1	NM_016148.3:c.5324G>Tp.(Gly1775Val)		homo	B	homo	maternal&paternal	3	Malformations	Meningo- or exencephalocele occipital, right radius aplasia with shortened ulna, death in utero
SYT3	NM_001160328.1:c.401C>Gp.(Ala134Gly)		homo	C	homo	maternal&paternal	3	Malformations	Meningo- or exencephalocele occipital, right radius aplasia with shortened ulna, death in utero
RPS6KC1	NM_012424.4:c.2633G>Ap.(Gly878Glu)		homo	C	homo	maternal&paternal	3	Malformations	Meningo- or exencephalocele occipital, right radius aplasia with shortened ulna, death in utero
TACC2	NM_206862.3:c.1407G>Cp.(Glu469Asp)	NM_206862.3:c.1242G>Cp.(Glu414Asp)	comphet	3,6	comphet	maternal&paternal	2	NDD	Macrocephaly, hypoplasia of the corpus callosum, suspected developmental delay,
SETD1B	NM_015048.1:c.1743C>Gp.(Asp581Glu)	NM_015048.1:c.2999G>Ap.(Arg1000Gln)	comphet	5,9	comphet	maternal&paternal	2	NDD	Macrocephaly, hypoplasia of the corpus callosum, suspected developmental delay, right hemiparesis
DUSP16	NM_030640.2:c.1091C>Ap.(Pro364His)	NM_030640.2:c.183C>Gp.(Asp61Glu)	comphet	3,8	comphet	maternal&paternal	1	NDD + Epilepsy	Microcephaly, Delayed speech and language development, Seizures, Global developmental delay, Recurrent infections, Infantile onset, Postnatal microcephaly
MCIDAS	NM_001190787.1:c.487C>Tp.(Arg163Trp)		de novo	6,2	het	de novo	1	NDD + Epilepsy	Stereotypy, Delayed speech and language development, Global developmental delay, Motor delay, Delayed gross motor development, EEG abnormality, Delayed fine motor
POLD1	NM_001308632.1:c.1657G>Ap.(Val553Ile)		homo	8,1	homo	maternal&paternal	1	NDD	Global developmental delay
FAT2	NM_001447.2:c.9524T>Cp.(Leu3175Pro)		homo	7,4	homo	maternal&paternal	4	NDD	Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Severe muscular hypotonia, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Intellectual disability, severe, Central hypotonia, Cognitive impairment
PRDM13	NM_021620.3:c.994G>Ap.(Gly332Arg)		homo	4,7	homo	maternal&paternal	4	NDD	Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Severe muscular hypotonia, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Intellectual disability, severe, Central hypotonia, Cognitive impairment
RASSF10	NM_001080521.2:c.816C>Gp.(Tyr272*)		de novo	3,7	het	de novo	2	NDD	Cleft palate, Hydrocephalus, Microcephaly, Retinopathy, Intellectual disability, Global developmental delay, Pes planus, Short stature, Pes valgus, Cognitive impairment, Cleft hard palate
GPATCH2	NM_018040.3:c.1167-8C>Tp.?		de novo	3,3	het	de novo	2	NDD	Cleft palate, Hydrocephalus, Microcephaly, Retinopathy, Intellectual disability, Global developmental delay, Pes planus, Short stature, Pes valgus, Cognitive impairment, Cleft hard palate
ATP1A3	NM_001256214.1:c.6+3A>Gp.?		homo	10,5	homo	maternal&paternal	4	NDD	Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Severe muscular hypotonia, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Intellectual disability, severe, Central hypotonia, Cognitive impairment
CLTCL1	NM_007098.3:c.1820A>Gp.(His607Arg)	NM_007098.3:c.2791A>Gp.(Ile931Val)	comphet	C	comphet	maternal&paternal	1	Cardio	Motor delay, Ventricular septal defect, Tracheal stenosis, Abnormal trachea morphology, Pulmonary artery atresia, Pulmonary valve atresia, Abnormality of cardiovascular system morphology
RAB5B	NM_001252036.1:c.115C>Gp.(Arg39Gly)		de novo	7,1	het	de novo	3	NDD + Epilepsy	Hydrocephalus, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Aqueductal stenosis
ARHGAP4	NM_001666.4:c.301C>Tp.(His101Tyr)		hemi	5,2	hemi	maternal	3	NDD + Epilepsy	Hydrocephalus, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Aqueductal stenosis
CBLL2	NM_152577.3:c.701A>Gp.(Lys234Arg)		hemi	4,1	hemi	maternal	3	NDD + Epilepsy	Hydrocephalus, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Aqueductal stenosis
ANKRD17	vl_032217.4:c.5360_5363delp.(Gln1787Argfs*5)		de novo	9,9	het	de novo	1	NDD	Coarse facial features, Muscular hypotonia, Global developmental delay, Motor delay, Hypertonia, Pes planus, Gait ataxia, Limb hypertonia, Muscular hypotonia of the trunk
LRRK2	NM_198578.3:c.3634C>Tp.(Pro1212Ser)	NM_198578.3:c.137C>Tp.(Thr46Met)	comphet	7,8	comphet	maternal&paternal	3	NDD	Behavioral abnormality, Intellectual disability, Gait ataxia
ZDHHC14	NM_024630.2:c.1441G>Ap.(Gly481Ser)		de novo	4,4	het	de novo	3	NDD	Behavioral abnormality, Intellectual disability, Gait ataxia
NIF3L1	NM_001136039.2:c.131C>Tp.(Ser44Leu)	NM_001136039.2:c.347C>Gp.(Ala116Gly)	comphet	3,8	comphet	maternal&paternal	3	NDD	Behavioral abnormality, Intellectual disability, Gait ataxia
MAP3K15	NM_001001671.3:c.2037dup, p.(Ile680Hisfs*9)		hemi	5,3	hemi	maternal	5	NDD + Epilepsy	Seizures, Generalized myoclonic seizures
HMCN2	NM_001291815.1:c.4444G>Tp.(Gly1482*)	NM_001291815.1:c.489+1G>Cp.?	comphet	3,6	comphet	maternal&paternal	5	NDD + Epilepsy	Seizures, Generalized myoclonic seizures
RPTOR	NM_020761.2:c.3533G>Ap.(Arg1178His)	NM_020761.2:c.503A>Gp.(Asn168Ser)	comphet	6,9	comphet	maternal&paternal	5	NDD + Epilepsy	Seizures, Generalized myoclonic seizures
ZNF761	_001008401.3:c.2085_2086delp.(Cys695Trpfs*5)		de novo	3,9	het	de novo	5	NDD + Epilepsy	Seizures, Generalized myoclonic seizures
HAGH	NM_005326.4:c.355_357dup, p.(Ser119dup)	NM_005326.4:c.578A>Gp.(Tyr193Cys)	comphet	4,0	comphet	maternal&paternal	5	NDD + Epilepsy	Seizures, Generalized myoclonic seizures
SLC30A5	NM_022902.4:c.832_836delp.(Ile278Phefs*33)		homo	A	homo	maternal&paternal	3	Malformations	Cardiomyopathy, Hydrops fetalis, Noncompaction cardiomyopath
POLR3A	NM_007055.3:c.1096A>Gp.(Arg366Gly)		de novo	9,9	het	de novo	1	NDD + Epilepsy	Seizures, Focal impaired awareness seizure, Hypothalamic hamartoma, Focal-onset seizure, Epileptic spasms, Langerhans cell histiocytosis

CACNA1C	NM_199460.3:c.496T>Cp.(Phe166Leu)		de novo	10,7	het	de novo	2	NDD + Epilepsy	epilepsy with absences and generalized tonic-clonic seizures, severe intellectual disability with autistic traits, low blood pressure, obstipation, normal MRI 2008
PLCG1	NM_002660.2:c.3196C>Gp.(Pro1066Ala)		het	6,8	het	unknown	1	NDD + Epilepsy	Focal epilepsy and mild intellectual disability, depression, global developmental delay, EEG abnormality, short stature, Obesity, mild ataxia on physical examination, cerebellar atrophy
EPHA4	NM_004438.4:c.2105T>Cp.(Met702Thr)		het	6,9	het	unknown	1	NDD + Epilepsy	Sensorineural hearing impairment, Strabismus, Abnormality of the hand, Intellectual disability, Seizures, Intellectual disability, mild, Abnormality of neuronal migration, Gray matter heterotopias, Gray matter heterotopia, Intellectual disability, moderate, EEG abnormality, Kyphosis, Clinodactyly of the 5th finger, Periventricular heterotopia, Focal-onset seizure, Focal sensory seizure, EEG with focal epileptiform discharges, EEG with focal spikes, Abnormality of brain morphology
AATK	NM_001080395.2:c.2915G>Tp.(Gly972Val)		homo	4,9	homo	maternal&paternal	2	NDD	Delayed speech and language development, Global developmental delay, Ventriculomegaly, Developmental regression, Expressive language delay, Severe expressive language delay, Progressive language deterioration, Receptive language delay, Moderate expressive language delay, Mild expressive language delay, Mild receptive language delay, Moderate receptive language delay, Severe receptive language delay
ARHGAP6	NM_013427.2:c.1586T>Cp.(Val529Ala)		hemi	4,0	hemi	maternal	2	NDD	Delayed speech and language development, Global developmental delay, Ventriculomegaly, Developmental regression, Expressive language delay, Severe expressive language delay, Progressive language deterioration, Receptive language delay, Moderate expressive language delay, Mild expressive language delay, Mild receptive language delay, Moderate receptive language delay, Severe receptive language delay
UNC13A	NM_001080421.2:c.3215+1G>Cp.?		het	10,3	het	unknown	3	NDD	Single transverse palmar crease, Syndactyly, Intellectual disability, Obesity, Toe syndactyly, Status epilepticus, Abnormality of body weight, Bipolar affective disorder, Increased adipose tissue, Epileptic spasms, Schizophrenia, Mania
LMX1A	NM_001174069.1:c.517dup, p.(Ser173Lysfs*15)		het	8,0	het	unknown	3	NDD	Single transverse palmar crease, Syndactyly, Intellectual disability, Obesity, Toe syndactyly, Status epilepticus, Abnormality of body weight, Bipolar affective disorder, Increased adipose tissue, Epileptic spasms, Schizophrenia, Mania
INO80	NM_017553.2:c.1294G>Ap.(Gly432Arg)		het	5,5	het	unknown	3	NDD	Single transverse palmar crease, Syndactyly, Intellectual disability, Obesity, Toe syndactyly, Status epilepticus, Abnormality of body weight, Bipolar affective disorder, Increased adipose tissue, Epileptic spasms, Schizophrenia, Mania
TANC2	NM_025185.3:c.3397G>Tp.(Gly1133*)		het	8,2	het	unknown	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized myoclonic seizures, Status epilepticus, Intellectual disability, moderate, Epileptic spasms, Cognitive impairment, Epileptic encephalopathy
TANC1	NM_001145909.1:c.2395G>Ap.(Asp799Asn)		het	4,2	het	unknown	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay
COL20A1	NM_020882.2:c.3467G>Tp.(Gly1156Val)	NM_020882.2:c.807C>Ap.(Asn269Lys)	comphet	4,0	comphet	maternal&paternal	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay
C16orf70	NM_001320540.1:c.1050+1G>Ap.?		de novo	9,1	het	de novo	2	NDD	Global developmental delay, short stature, talipes equinovarus
EPHB2	NM_001309193.1:c.2858T>Cp.(Ile953Thr)		de novo	8,0	het	de novo	1	NDD + Epilepsy	epilepsy, global developmental delay, dysmorphic facial features
BIRC6	NM_016252.3:c.8570C>Gp.(Ser2857Cys)	NM_016252.3:c.12796G>Ap.(Val4266Met)	comphet	4,4	comphet	maternal&paternal	1	NDD + Epilepsy	Delayed speech and language development, Seizures, Familial predisposition, Poor speech, Infantile onset, Infantile spasms
WDR3	NM_006784.2:c.989+1G>Cp.?	NM_006784.2:c.1555T>Ap.(Ser519Thr)	comphet	5,4	comphet	maternal&paternal	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay
FGF2	NM_002006.4:c.498C>Gp.(Tyr166*)		het	7,6	het	unknown	1	NDD + Epilepsy	Hydrocephalus, Intellectual disability, Hemiparesis, Cerebellar atrophy, Focal clonic seizures, Febrile seizures, Hemiplegia/hemiparesis, Periventricular leukomalacia, Focal motor seizure, Focal autonomic seizure, Focal myoclonic seizures, Focal tonic seizures
CNTN6	NM_014461.3:c.275A>Tp.(Asp92Val)	NM_014461.3:c.2573G>Ap.(Ser858Asn)	comphet	5,5	comphet	maternal&paternal	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay
FAM131A	NM_144635.4:c.838C>Gp.(Leu280Val)		homo	B	homo	maternal&paternal	1	Muscle	neuromuscular scoliosis, muscular hypotonia, intermittent exotropia
TMEM92	NM_001168215.1:c.212G>Cp.(Cys71Ser)		de novo	B	het	de novo	1	Malformations	inguinal hernia, cryptorchidism, hypospadias, renal cyst, hemangioma, bicuspid aortic valve, coarctation of aorta, anal atresia, choroid plexus cyst, abnormal vertebral morphology, multiple renal cysts
NEURL4	NM_032442.2:c.4345C>Gp.(Pro1449Ala)	NM_032442.2:c.2944G>Ap.(Glu982Lys)	comphet	4,5	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Atonic seizures, Focal tonic seizures, Myoclonic atonic seizures
RORB	NM_006914.3:c.235+1_235+2insTp.?		het	8,4	het	unknown	2	NDD + Epilepsy	Male hypogonadism, Hypogonadotrophic hypogonadism, Hypogonadism, Thick upper lip vermilion, Delayed puberty, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Myoclonus, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Intellectual disability, moderate, Intellectual disability, borderline, Focal tonic seizures, Thick vermilion border
RORB	NM_006914.3:c.208T>Cp.(Cys70Arg)		het	6,7	het	maternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Mental deterioration, Absence seizure, Generalized myoclonic seizures, Status epilepticus, Intellectual disability, profound, Intellectual disability, moderate, Intellectual disability, severe, Epileptic spasms, Myoclonic absence, Cognitive impairment, Epileptic
XYLB	NM_005108.3:c.1101G>Ap.(Met367Ile)		de novo	5,0	het	de novo	1	NDD	Single transverse palmar crease, Intellectual disability, Muscular hypotonia, Global developmental delay, Gait disturbance, Absent speech, Supernumerary nipple, Lumbar scoliosis, Abnormal social behavior, Cognitive impairment, Self-injurious behavior
GABBR1	NM_001470.3:c.1190C>Tp.(Ala397Val)		de novo	10,1	het	de novo	2	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Global developmental delay, Motor delay, Encopresis

ATP2B3	NM_001001344.2:c.3530C>Tp.(Pro1177Leu)		hemi	6,0	hemi	maternal	2	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Global developmental delay, Motor delay, Encopresis
GRIK3	NM_000831.3:c.1531-5T>Gp.?		het	4,1	het	unknown	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic-clonic seizures, Intellectual disability, moderate, Focal-onset seizure, Pituitary hypothyroidism, Intellectual disability, severe, Focal tonic seizures, Arrhythmia, Abnormality of brain morphology, Abnormality of cardiovascular system morphology
BSN	NM_003458.3:c.9919A>Gp.(Ser3307Gly)		homo	6,7	homo	maternal&paternal	2	NDD + Epilepsy	Male hypogonadism, Hypogonadotrophic hypogonadism, Hypogonadism, Thick upper lip vermilion, Delayed puberty, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Myoclonus, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Intellectual disability, moderate, Intellectual disability, borderline, Focal tonic seizures, Thick vermilion border
PUM2	NM_015317.2:c.2216delp.(His739Leufs*10)		het	10,9	het	unknown	1	NDD + Epilepsy	Strabismus, Intellectual disability, Seizures, Global developmental delay, Generalized tonic-clonic seizures, Global brain atrophy, EEG abnormality, Developmental regression, Short stature, Brain atrophy, Cognitive impairment
HMG20A	NM_001304504.1:c.237+5G>Tp.?		homo	5,0	homo	maternal&paternal	1	NDD + Epilepsy	Macroglossia, Mandibular prognathia, Thick eyebrow, Intellectual disability, Seizures, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Kyphoscoliosis, Genu valgum, Excessive salivation, Protruding tongue, Intellectual disability, severe, Thick hair, Rigors
KIF5C	NM_004522.2:c.2385dup, p.(Gln796Alafs*19)		het	9,9	het	unknown	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Mental deterioration, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Focal clonic seizures, Intellectual disability, moderate, Focal impaired awareness seizure, Intellectual disability, borderline, Focal-onset seizure, Focal motor seizure, Focal tonic seizures, Generalized clonic seizures, Abnormal morphology of the hippocampus, Focal seizures, afebril
RNF13	007282.4:c.(409+1_410-1)_(500+1_501-1)delp.?		de novo	6,2	het	de novo	1	NDD + Epilepsy	Microcephaly, Seizures, Global developmental delay, Hepatosplenomegaly, Anemia, Hypoplasia of the corpus callosum, Polymicrogyria, Abnormality of midbrain morphology, Elevated hepatic transaminase, Elevated gamma-glutamyltransferase activity
RORB	NM_006914.3:c.777G>Ap.(Trp259*)		het	8,1	het	unknown	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe, Epileptic
PRKCE	NM_005400.2:c.1186C>Tp.(Arg396Trp)		de novo	8,2	het	de novo	1	NDD + Epilepsy	Seizures, Focal-onset seizure
SSBP2	NM_001256732.2:c.566C>Tp.(Pro189Leu)		het	3,4	het	unknown	3	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Dystonia, Gliosis, Focal clonic seizures, EEG abnormality, Poor speech, Focal-onset seizure, Dyslexia, Focal motor seizure, Focal tonic seizures, Abnormality of movement, Cognitive impairment
ZNF319	NM_020807.2:c.654_655delp.(Arg219Alafs*2)		het	5,3	het	unknown	3	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Dystonia, Gliosis, Focal clonic seizures, EEG abnormality, Poor speech, Focal-onset seizure, Dyslexia, Focal motor seizure, Focal tonic seizures, Abnormality of movement, Cognitive impairment
GLRA4	NM_001024452.2:c.39_41delp.(Leu14del)		hemi	5,5	hemi	unknown	2	NDD + Epilepsy	Thin upper lip vermilion, Turricephaly, Synophrys, Acne, Intellectual disability, Seizures, Mental deterioration, Spastic tetraparesis, Absent speech, Flexion contracture, Cerebral atrophy, Nail dysplasia, Focal clonic seizures, Tetraparesis, Spastic paraparesis, Paraparesis, Tetraplegia, Neonatal respiratory distress, High, narrow palate, Elbow flexion contracture, Limb joint contracture, Skeletal muscle atrophy, Limb muscle weakness, Short stature, Focal-onset seizure, Limb undergrowth, Paraplegia/paraparesis, Intellectual disability, severe, Focal motor seizure, Focal tonic seizures, Tetraplegia/tetraparesis
TAB2	NM_015093.5:c.1448delp.(Pro483Leufs*16)		de novo	10,6	het	de novo	3	NDD	Hypotelorism, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Single median maxillary incisor, Agenesis of permanent teeth, Abnormality of dental morphology, Reduced number of teeth, Intellectual disability, severe
SIPA1L1	_015556.2:c.3321_3322delp.(Arg1107Serfs*11)		het	7,5	het	unknown	2	NDD + Epilepsy	Macrotia, Myopia, Intellectual disability, Seizures, Intellectual disability, mild, Cerebellar atrophy, Tremor, Cerebral atrophy, Broad-based gait, Intellectual disability, profound, Intellectual disability, moderate, Hand tremor, Focal-onset seizure, Epileptic spasms, Interictal epileptiform activity, Limb tremor, Epileptic encephalopathy, Long ear
SRSF11	NM_004768.3:c.1178delp.(Arg393Hisfs*26)		het	8,6	het	unknown	2	NDD + Epilepsy	Macrotia, Myopia, Intellectual disability, Seizures, Intellectual disability, mild, Cerebellar atrophy, Tremor, Cerebral atrophy, Broad-based gait, Intellectual disability, profound, Intellectual disability, moderate, Hand tremor, Focal-onset seizure, Epileptic spasms, Interictal epileptiform activity, Limb tremor, Epileptic encephalopathy, Long ear
BDP1	NM_018429.2:c.4813A>Gp.(Arg1605Gly)		de novo	6,3	het	de novo	3	NDD + Epilepsy	Intellectual disability, Seizures, Status epilepticus
NUP188	NM_015354.2:c.17G>Cp.(Gly6Ala)	NM_015354.2:c.2917C>Tp.(Arg973*)	comphet	5,5	comphet	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Status epilepticus
TMEM151A	NM_153266.3:c.76-1G>Cp.?		de novo	6,8	het	de novo	3	NDD + Epilepsy	Intellectual disability, Seizures, Status epilepticus
ANK2	NM_001148.4:c.1288-1G>Ap.?		de novo	12,4	het	de novo	3	NDD + Epilepsy	benign epilepsy
NFATC1	51delCCTinsTCGp.(Pro750_Cys751delinsLeuGly)		de novo	6,3	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Abnormal cerebellum morphology, Abnormality of the basal ganglia, Increased serum lactate, Abnormality of midbrain morphology, Lacticaciduria, Epileptic spasms, Abnormality of brain morphology

ABCA2	M_001606.4:c.801_802delTGinsGTp.(Val268Phe)		homo	7,3	homo	maternal&paternal	4	NDD	Adducted thumb, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Pyloric stenosis, Ventriculomegaly, Intellectual disability, moderate, Infantile muscular hypotonia, Feeding difficulties, Cognitive impairment, Impaired feeding ability, VUS in COLQ (31.07.2019)
BIRC6	NM_016252.3:c.10735A>Gp.(Met3579Val)		homo	6,4	homo	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Visual impairment, Intellectual disability, Seizures, Global developmental delay, Motor delay, Encephalopathy, Generalized tonic-clonic seizures
PPM1L	NM_139245.3:c.237G>Cp.(Glu79Asp)		homo	4,4	homo	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Visual impairment, Intellectual disability, Seizures, Global developmental delay, Motor delay, Encephalopathy, Generalized tonic-clonic seizures
RGMA	NM_001166283.1:c.748G>Cp.(Ala250Pro)		de novo	6,9	het	de novo	2	NDD	Spasticity, Global developmental delay, Motor delay, Cerebral palsy, Abnormality of movement, Dyskinesia
ANXA6	NM_001155.4:c.1670C>Tp.(Pro557Leu)	NM_001155.4:c.319-6_319-5delCCinsTGp.?	comphet	4,0	comphet	maternal&paternal	2	NDD	Spasticity, Global developmental delay, Motor delay, Cerebral palsy, Abnormality of movement, Dyskinesia
NRDE2	NM_017970.3:c.441delp.(Arg148Alafs*11)		homo	8,5	homo	maternal&paternal	1	NDD	Intellectual disability, seizures, global developmental delay, encephalopathy infantile
INTS7	NM_015434.3:c.2240G>Tp.(Arg747Ile)		de novo	6,0	het	de novo	1	NDD	Microcephaly, Intrauterine growth retardation, Abnormal facial shape, Basal ganglia calcification, Cerebral calcification, Congenital intracerebral calcification
SF3A1	NM_005877.5:c.310G>Ap.(Gly104Arg)		de novo	7,3	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Abnormality of movement, Epileptic encephalopathy
SLC16A10	NM_018593.4:c.626G>Ap.(Gly209Asp)		de novo	6,6	het	de novo	2	NDD + Epilepsy	Microcephaly, Behavioral abnormality, Seizures, Global developmental delay, Absence seizure, Generalized-onset seizure, Myoclonic atonic seizures
MROH2B	NM_173489.4:c.3685delp.(Asp1229Thrfs*15)		de novo	5,0	het	de novo	1	NDD + Epilepsy	Seizures, Encephalopathy, Absence seizure, Generalized-onset seizure
PRDX2	NM_005809.5:c.153C>Ap.(Cys51*)		de novo	7,3	het	de novo	1	NDD + Epilepsy	Seizures, absent septum pellucidum, paroxysmal dyskinesia, dyskinesia
SLC5A7	NM_021815.4:c.178+1G>Cp.?		het	7,8	het	maternal	1	Neuro	Ataxia, spastic paraplegia, muscle weakness, hyperreflexia, pes cavus, myalgia, limb muscle weakness, paraplegia
ZNF341	NM_032819.4:c.2260C>Tp.(Arg754Cys)		de novo	4,3	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Specific learning disability, Absence seizure, Generalized-onset seizure, Intellectual disability, borderline, Attention deficit hyperactivity disorder
KCNK7	NM_033347.1:c.681C>Gp.(His227Gln)		de novo	4,7	het	de novo	1	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Generalized myoclonic seizures
ZZEF1	NM_015113.3:c.1580C>Tp.(Pro527Leu)		de novo	5,9	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Specific learning disability, Absence seizure, Generalized-onset seizure, Intellectual disability, borderline, Attention deficit hyperactivity disorder
MTMR3	NM_021090.3:c.848A>Gp.(Asn283Ser)	NM_021090.3:c.1088G>Ap.(Arg363Gln)	comphet	4,3	comphet	maternal&paternal	1	NDD + Epilepsy	Delayed speech and language development, Seizures, Global developmental delay, Focal impaired awareness seizure, Cortical dysplasia, Focal-onset seizure, Complex febrile seizures, Abnormal morphology of the hippocampus
INPP5F	NM_014937.3:c.3172_3174delp.(Ser1058del)	4937.3:c.3144_3149delp.(Leu1049_Glu1050del)	comphet	4,1	comphet	maternal&paternal	1	NDD + Epilepsy	Global developmental delay, Epileptic spasms
HCN2	NM_001194.3:c.1120A>Cp.(Met374Leu)		de novo	8,9	het	de novo	1	NDD + Epilepsy	Microcephaly, delayed speech and language development, intellectual disability, global developmental delay, motor delay, generalized-onset seizure, epileptic spasms, cognitive impairment
DHX36	NM_020865.2:c.800_802delp.(Ile267del)		de novo	5,9	het	de novo	1	NDD	Short attention span, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Delayed gross motor development, Attention deficit hyperactivity disorder, Delayed fine motor development
DOCK3	NM_004947.4:c.1175G>Ap.(Arg392Gln)	NM_004947.4:c.3740T>Cp.(Met1247Thr)	comphet	9,3	comphet	maternal&paternal	1	NDD + Epilepsy	Seizures, Global developmental delay
SEZ6L2	NM_001243332.1:c.1084G>Ap.(Val362Met)	NM_001243332.1:c.85C>Tp.(Pro29Ser)	comphet	6,1	comphet	maternal&paternal	1	NDD + Epilepsy	Seizures, status epilepticus, focal-onset seizure, EEG with spike-wave complexes, epilepsy not completely under control, cognitive deficiency, intellectual disability
NOP58	NM_015934.4:c.1018C>Gp.(Leu340Val)		de novo	7,0	het	de novo	1	NDD + Epilepsy	Autism, Intellectual disability, Status epilepticus, Focal-onset seizure, Hippocampal atrophy
SLITRK4	NM_001184749.2:c.2435T>Cp.(Phe812Ser)		hemi	5,1	hemi	maternal	1	NDD	Myopia, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Dysarthria, Global developmental delay, Delayed gross motor development, Intellectual disability, moderate, Delayed fine motor development, High myopia
PGBD2	NM_170725.2:c.607A>Cp.(Thr203Pro)		de novo	4,0	het	de novo	2	NDD	Autism, Autistic behavior, Intellectual disability, Global developmental delay, Situs inversus totalis, Abnormal facial shape, Asthma, Recurrent respiratory infections, Short stature, Respiratory tract infection
ZNF81	NM_007137.3:c.476A>Gp.(Lys159Arg)		hemi	6,5	hemi	maternal	2	NDD	Autism, Autistic behavior, Intellectual disability, Global developmental delay, Situs inversus totalis, Abnormal facial shape, Asthma, Recurrent respiratory infections, Short stature, Respiratory tract infection
ZFYVE26	NM_015346.3:c.5779T>Ap.(Tyr1927Asn)		de novo	10,3	het	de novo	1	NDD	Global developmental delay, Absent speech, Proportionate short stature, Short stature
FAT3	NM_001008781.2:c.1367C>Tp.(Ala456Val)	NM_001008781.2:c.11012G>Tp.(Arg3671Leu)	comphet	5,1	comphet	maternal&paternal	1	NDD + Epilepsy	strukturelle und therapierefraktäre Epilepsie (ESES/CSWS), zervikale Syringomyelie, Intelligenzminderung, Verhaltensauffälligkeiten, Z.n. IVH Grad IV (intraventrikuläre Hämorrhagie) in 2. Lebenswoche, cMRT-Auffälligkeiten
PKN3	NM_013355.4:c.137A>Cp.(Asp46Ala)		de novo	5,0	het	de novo	1	NDD + Epilepsy	Generalisierte Epilepsie mit febrilen Anfällen seit dem 3. LJ
GABRE	NM_004961.3:c.319G>Tp.(Gly107Cys)		hemi	4,9	hemi	maternal	2	NDD	Macrocephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Intellectual disability, severe, Cognitive impairment

DACH2	NM_053281.3:c.1519G>Tp.(Val507Phe)		hemi	3,4	hemi	maternal	2	NDD	Macrocephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Intellectual disability, severe, Cognitive impairment
ARRB2	NM_001257328.1:c.684+1G>Cp.?		de novo	10,2	het	de novo	1	NDD + Epilepsy	autism-spectre disorder, focal- onset epilepsy
DBF4B	NM_145663.2:c.902G>Tp.(Cys301Phe)		homo	6,3	homo	maternal&paternal	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
TBC1D9B	NM_198868.2:c.583G>Tp.(Ala195Ser)		de novo	5,6	het	de novo	1	Neuro	Abnormality of the optic nerve, Optic atrophy, Polyneuropathy, Encephalopathy, Leukoencephalopathy, Leukodystrophy, Tetraplegia
CASP9	NM_001229.4:c.631-6T>Cp.?	NM_001229.4:c.710A>Cp.(His237Pro)	comphet	6,5	comphet	maternal&paternal	3	NDD	Renal agenesis, Abnormal cornea morphology, Aniridia, Microphthalmia, Global developmental delay
TNPO3	NM_012470.3:c.2541dup, p.(Tyr848Leufs*8)		de novo	6,9	het	de novo	3	NDD	Renal agenesis, Abnormal cornea morphology, Aniridia, Microphthalmia, Global developmental delay
SLC23A1	NM_152685.3:c.1105A>Gp.(Ile369Val)	NM_152685.3:c.1063C>Ap.(Pro355Thr)	comphet	4,5	comphet	maternal&paternal	3	NDD	Renal agenesis, Abnormal cornea morphology, Aniridia, Microphthalmia, Global developmental delay
SMARCA1	NM_003069.4:c.34G>Ap.(Val12Met)		hemi	6,5	hemi	maternal	4	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay
PON1	NM_000446.5:c.717G>Cp.(Glu239Asp)		de novo	5,3	het	de novo	4	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay
CAND2	NM_001162499.1:c.2591C>Tp.(Ala864Val)		de novo	4,8	het	de novo	4	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay
MARVELD3	NM_001017967.3:c.1168G>Ap.(Gly390Ser)		de novo	5,2	het	de novo	1	NDD	Autistic behavior, Intellectual disability, Global developmental delay, Obesity, Polyphagia, Developmental stagnation, Retractile testis, Cognitive impairment
ANKRD6	NM_001242809.1:c.1667C>Tp.(Pro556Leu)		de novo	5,1	het	de novo	1	NDD	Dandy-Walker malformation, Omphalocele, Occipital encephalocele, Meningocele
CLCN3	NM_173872.3:c.336_339delp.(Lys112Asnfs*6)		homo	11,1	homo	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormal corpus callosum morphology, Agenesis of corpus callosum, Generalized tonic-clonic seizures, Hypoplasia of the corpus callosum, Generalized myoclonic seizures, Generalized-onset seizure, Atonic seizures, Epileptic spasms
MORC4	NM_024657.4:c.1382A>Gp.(Tyr461Cys)		hemi	5,1	hemi	maternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormal corpus callosum morphology, Agenesis of corpus callosum, Generalized tonic-clonic seizures, Hypoplasia of the corpus callosum, Generalized myoclonic seizures, Generalized-onset seizure, Atonic seizures, Epileptic spasms
PAM	NM_001319943.1:c.1670C>Gp.(Ser557Trp)		homo	6,5	homo	maternal&paternal	2	NDD	Strabismus, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Generalized hypotonia, Intellectual disability, moderate, Intellectual disability, severe
MYO9B	NM_001130065.1:c.248C>Tp.(Ser83Leu)	NM_001130065.1:c.5020G>Ap.(Val1674Met)	comphet	4,5	comphet	maternal&paternal	2	NDD	Strabismus, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Generalized hypotonia, Intellectual disability, moderate, Intellectual disability, severe
CSNK1A1	NM_001025105.2:c.686G>Ap.(Arg229Gln)		de novo	7,7	het	de novo	3	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia
HEPH	NM_138737.4:c.812_814delp.(Pro271del)		hemi	3,9	hemi	maternal	3	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia
DNHD1	NM_144666.2:c.2758A>Gp.(Ser920Gly)	NM_144666.2:c.2546G>Ap.(Arg849Gln)	comphet	3,7	comphet	maternal&paternal	3	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia
RADIL	NM_018059.4:c.1450C>Tp.(Gln484*)		homo	7,5	homo	maternal&paternal	1	NDD	recurrent hypoglycemia, microcephaly, hypopiturism
PHACTR3	NM_001199505.1:c.17G>Tp.(Gly6Val)		de novo	5,5	het	de novo	1	NDD	Intellectual disability, Global developmental delay
SP9	NM_001145250.1:c.1133A>Gp.(Glu378Gly)		de novo	5,5	het	de novo	1	NDD + Epilepsy	picanthus, Seizures, Global developmental delay, Abnormal facial shape, Generalized-onset seizure, Severe muscular hypotonia, Muscular hypotonia of the trunk, Infantile muscular hypotonia
PTBP1	NM_002819.4:c.144A>Tp.(Lys48Asn)		de novo	8,3	het	de novo	2	NDD	Cleft palate, Cleft soft palate, Thickened nuchal skin fold, Intellectual disability, Global developmental delay, Small for gestational age, Short stature, Cleft hard palate
TNR	NM_003285.2:c.3659C>Tp.(Ser1220Phe)	NM_003285.2:c.496A>Gp.(Thr166Ala)	comphet	5,2	comphet	maternal&paternal	2	NDD	Cleft palate, Cleft soft palate, Thickened nuchal skin fold, Intellectual disability, Global developmental delay, Small for gestational age, Short stature, Cleft hard palate
MAB21L4	NM_001085437.2:c.755A>Gp.(Tyr252Cys)		de novo	3,8	het	de novo	1	NDD	Abnormality of dental enamel, Autistic behavior, Delayed speech and language development, Global developmental delay, Motor delay, Sleep disturbance, Poor
NAV2	NM_001244963.1:c.2486C>Tp.(Pro829Leu)	NM_001244963.1:c.7137+3G>Ap.?	comphet	5,4	comphet	maternal&paternal	1	NDD	Astigmatism, Hypermetropia, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Dandy-Walker malformation, Cerebellar hypoplasia, Delayed gross motor development, Enlarged cisterna magna, Scoliosis, High hypermetropia, Intellectual disability, severe, Mild global developmental delay, Cognitive impairment, Hernia, Mild hypermetropia
MED14	NM_004229.3:c.3657T>Gp.(His1219Gln)		hemi	4,0	hemi	maternal	1	NDD + Epilepsy	Autistic behavior, Intellectual disability, Seizures, Global developmental delay, Intellectual disability, severe, Severe global developmental delay, Epileptic encephalopathy
MYRIP	NM_001284423.1:c.1525G>Ap.(Asp509Asn)	NM_001284423.1:c.2419C>Tp.(Pro807Ser)	comphet	4,3	comphet	maternal&paternal	1	NDD + Epilepsy	Hearing impairment, Delayed speech and language development, Atopic dermatitis, Intellectual disability, Seizures, Motor delay, Pachygyria, Lissencephaly, Bradykinesia, Dysdiadochokinesis, Orofacial dyskinesia, Poor speech, Scoliosis, Aspiration, Thoracic scoliosis, Thoracolumbar scoliosis, Lumbar scoliosis, Allergy

ZNF692	NM_001136036.2:c.70C>Gp.(Gln24Glu)		de novo	5,4	het	de novo	2	NDD + Epilepsy	Seizures, Global developmental delay, Generalized-onset seizure, Periventricular leukomalacia
FAT1	NM_005245.3:c.11017G>Cp.(Val3673Leu)	NM_005245.3:c.6079C>Tp.(Arg2027Cys)	comphet	6,0	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Global developmental delay, Generalized-onset seizure, Periventricular leukomalacia
PAPOLG	NM_022894.3:c.533C>Gp.(Ser178*)		de novo	9,2	het	de novo	4	NDD + Epilepsy	Seizures, Generalized-onset seizure
SCN11A	NM_014139.2:c.95C>Tp.(Ala32Val)	NM_014139.2:c.2821G>Ap.(Glu941Lys)	comphet	6,1	comphet	maternal&paternal	4	NDD + Epilepsy	Seizures, Generalized-onset seizure
HSD17B6	NM_003725.3:c.440G>Ap.(Ser147Asn)		de novo	6,0	het	de novo	4	NDD + Epilepsy	Seizures, Generalized-onset seizure
XDH	NM_000379.3:c.2559G>Cp.(Lys853Asn)		de novo	6,3	het	de novo	4	NDD + Epilepsy	Seizures, Generalized-onset seizure
FYTTD1	NM_032288.6:c.755G>Cp.(Arg252Pro)		de novo	6,5	het	de novo	1	NDD	Microcephaly, Nystagmus, Impaired social interactions, Intellectual disability, Muscular hypotonia, Global developmental delay, EEG abnormality
ARMCX1	NM_016608.1:c.520dup, p.(Arg174Profs*3)		hemi	6,6	hemi	maternal	3	NDD + Epilepsy	Autistic behavior, Delayed speech and language development, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Poor fine motor coordination, Delayed social development, Cognitive impairment
ARFGEF3	NM_020340.4:c.787G>Ap.(Ala263Thr)		de novo	7,0	het	de novo	3	NDD + Epilepsy	Autistic behavior, Delayed speech and language development, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Poor fine motor coordination, Delayed social development, Cognitive impairment
DMRT3	NM_021240.3:c.917C>Tp.(Ala306Val)		de novo	5,4	het	de novo	3	NDD + Epilepsy	Autistic behavior, Delayed speech and language development, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Poor fine motor coordination, Delayed social development, Cognitive impairment
CFAP74	NM_001304360.1:c.3409delp.(Gln1137Argfs*37)		de novo	6,0	het	de novo	1	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Global developmental delay, Absence seizure, Generalized-onset seizure, EEG abnormality, Developmental regression, Poor speech
H1-10	NM_006026.3:c.80C>Tp.(Ser27Leu)		homo	3,5	homo	maternal&paternal	2	NDD	Retinal dystrophy, Microphthalmia, Delayed speech and language development, Global developmental delay, Poor speech, Vitreoretinopathy, Congenital blindness
DNHD1	NM_144666.2:c.3410G>Ap.(Arg1137Gln)	NM_144666.2:c.2450A>Cp.(His817Pro)	comphet	3,6	comphet	maternal&paternal	2	NDD	Retinal dystrophy, Microphthalmia, Delayed speech and language development, Global developmental delay, Poor speech, Vitreoretinopathy, Congenital blindness
MRO	NM_001127176.1:c.550T>Ap.(Phe184Ile)		homo	6,3	homo	maternal&paternal	1	NDD + Epilepsy	Absent speech, Obesity, Intellectual disability, severe, Epilepsy
RIC8B	NM_001330145.1:c.399G>Cp.(Gln133His)		de novo	6,1	het	de novo	3	Neuro	Sudden spastic of lower extremitiesa and bowel incontinence at the age of 43 years
SLC25A14	NM_001282197.1:c.124G>Cp.(Val42Leu)		hemi	5,7	hemi	maternal	3	Neuro	Sudden spastic of lower extremitiesa and bowel incontinence at the age of 43 years
TRPC7	NM_020389.2:c.1577A>Gp.(Tyr526Cys)		homo	3,7	homo	maternal&paternal	3	Neuro	Sudden spastic of lower extremitiesa and bowel incontinence at the age of 43 years
PAPSS1	NM_005443.4:c.1672G>Ap.(Val558Ile)		homo	5,3	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Cerebellar vermis atrophy, Cognitive impairment
PDE4DIP	NM_001198834.3:c.5842A>Gp.(Lys1948Glu)	NM_001198834.3:c.4063C>Tp.(Arg1355*)	comphet	6,2	comphet	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Cerebellar vermis atrophy, Cognitive impairment
TAF5	NM_006951.4:c.479C>Tp.(Ala160Val)		homo	4,9	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Cerebellar vermis atrophy, Cognitive impairment
JPH4	M_001146028.1:c.953_956delp.(Gly318Alafs*53)		de novo	10,2	het	de novo	1	NDD	Microcephaly, Autism, Intellectual disability, Muscular hypotonia, Global developmental delay
HSPA8	NM_006597.5:c.98A>Gp.(Gln33Arg)		de novo	8,9	het	de novo	1	NDD + Epilepsy	seizures, focal seizures, myoclonic seizures
BAZ1B	NM_032408.3:c.461G>Ap.(Gly154Asp)		de novo	9,7	het	de novo	1	NDD + Epilepsy	absence epilepsy, EEG abnormality
ADARB2	NM_018702.3:c.1570G>Ap.(Glu524Lys)	NM_018702.3:c.914G>Ap.(Ser305Asn)	comphet	6,2	comphet	maternal&paternal	2	NDD	Microcephaly, Hearing impairment, Autism, Intellectual disability, Spasticity, Global developmental delay, Cerebral calcification
DISP1	NM_032890.3:c.1357A>Cp.(Met453Leu)	NM_032890.3:c.3233G>Ap.(Arg1078His)	comphet	7,3	comphet	maternal&paternal	2	NDD	Cleft palate, Panhypopituitarism, Intellectual disability, Patent ductus arteriosus, Facial cleft, Scoliosis, Short stature, Median cleft lip and palate
UNC79	NM_020818.4:c.3857-691A>Gp.(=)	NM_020818.4:c.1547C>Tp.(Ser516Leu)	comphet	5,3	comphet	maternal&paternal	2	NDD	Cleft palate, Panhypopituitarism, Intellectual disability, Patent ductus arteriosus, Facial cleft, Scoliosis, Short stature, Median cleft lip and palate
GABRG1	NM_173536.3:c.487A>Gp.(Thr163Ala)		de novo	7,7	het	de novo	2	NDD	Strabismus, Autism, Ataxia, Specific learning disability, Gait ataxia, Language impairment, Pain insensitivity, Abnormality of movement, Motor tics, Dyskinesia, Exodeviation
ARFGEF3	NM_020340.4:c.5123+2T>Cp.?		het	7,3	het	maternal	2	NDD	Strabismus, Autism, Ataxia, Specific learning disability, Gait ataxia, Language impairment, Pain insensitivity, Abnormality of movement, Motor tics, Dyskinesia, Exodeviation
ZHX1	NM_001017926.2:c.179A>Gp.(Asn60Ser)	NM_001017926.2:c.962C>Tp.(Ala321Val)	comphet	3,4	comphet	maternal&paternal	2	NDD + Epilepsy	Hearing impairment, Visual impairment, Nystagmus, Seizures, Abnormality of the cerebrospinal fluid, Epileptic spasms, Abnormal CSF glucose level
PRPF6	NM_012469.3:c.67C>Tp.(Arg23Trp)		de novo	9,1	het	de novo	1	NDD	Visual impairment, Intellectual disability, Growth delay, Mildly reduced visual acuity, Feeding difficulties
SOX7	NM_031439.3:c.723G>Ap.(Pro241=)		de novo	4,0	het	de novo	2	NDD	Microcephaly, Hearing impairment, Autism, Intellectual disability, Spasticity, Global developmental delay, Cerebral calcification
KCTD16	NM_020768.3:c.1231T>Cp.(Phe411Leu)		de novo	6,2	het	de novo	3	NDD + Epilepsy	Therapy-resistant epilepsy since the age of two, Epileptic encephalopathy
MST1	NM_020998.3:c.1603C>Gp.(Arg535Gly)		de novo	5,5	het	de novo	2	Neuro	Migraine, Migraine with aura, Migraine without aura, Cortical dysplasia, Frontoparietal cortical dysplasia
AKAP13	NM_006738.5:c.914A>Gp.(Gln305Arg)	NM_006738.5:c.8228A>Cp.(Lys2743Thr)	comphet	5,4	comphet	maternal&paternal	2	Neuro	Migraine, Migraine with aura, Migraine without aura, Cortical dysplasia, Frontoparietal cortical dysplasia
ABCC12	NM_033226.2:c.796G>Ap.(Gly266Arg)	NM_033226.2:c.442delp.(Ile148Serfs*20)	comphet	5,7	comphet	maternal&paternal	3	NDD + Epilepsy	Therapy-resistant epilepsy since the age of two, Epileptic encephalopathy
LRCH2	NM_020871.3:c.2141A>Gp.(Asn714Ser)		hemi	4,1	hemi	maternal	3	NDD + Epilepsy	Therapy-resistant epilepsy since the age of two, Epileptic encephalopathy

WARS1	NM_173701.1:c.397C>Tp.(Arg133Cys)		homo	8,7	homo	maternal&paternal	2	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Global developmental delay, Absent speech, Intellectual disability, profound, Intellectual disability, moderate, Poor speech, Inability to walk, Melanoma, Intellectual disability, severe
CSTF2	NM_001306206.1:c.724G>Ap.(Ala242Thr)		hemi	5,6	hemi	maternal	2	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Global developmental delay, Absent speech, Intellectual disability, profound, Intellectual disability, moderate, Poor speech, Inability to walk, Melanoma, Intellectual disability, severe
TANK	NM_001199135.1:c.1012T>Cp.(Tyr338His)		de novo	6,2	het	de novo	1	NDD + Epilepsy	Restlessness, Single transverse palmar crease, Seizures, Global developmental delay, Abnormal corpus callosum morphology, Abnormality of neuronal migration, Abnormality of the periventricular white matter, Infantile spasms
TTC28	NM_001145418.1:c.3020A>Gp.(Tyr1007Cys)		het	6,6	het	unknown	1	NDD	Tall stature, Macrocephaly, Autistic behavior, Delayed speech and language development, Intellectual disability, Global developmental delay, Obesity, Abnormal social behavior
TNN	NM_022093.1:c.1949A>Tp.(Tyr650Phe)	NM_022093.1:c.2852T>Gp.(Val951Gly)	comphet	4,5	comphet	maternal&paternal	1	NDD + Epilepsy	infantile spams since 6 months of age, conspicuous odor, crying phases, failure to thrive
TKT	NM_001135055.2:c.1751T>Cp.(Val584Ala)		de novo	8,5	het	de novo	3	NDD	Global developmental delay, Motor delay
RASAL2	NM_004841.3:c.433G>Tp.(Glu145*)		de novo	7,9	het	de novo	3	NDD	Global developmental delay, Motor delay
HSPB7	NM_014424.4:c.202C>Tp.(Arg68Cys)		de novo	5,1	het	de novo	3	NDD	Global developmental delay, Motor delay
GNL3L	NM_001184819.1:c.884T>Ap.(Leu295Gln)		hemi	3,1	hemi	maternal	1	NDD	Global developmental delay with delayed speech and language development and a suspected autism spectre disorder, makrosomia
SETD1B	NM_015048.1:c.3074G>Ap.(Arg1025Gln)	NM_015048.1:c.4354C>Tp.(Arg1452Cys)	comphet	6,3	comphet	maternal&paternal	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech
HYDIN	NM_001270974.2:c.6271A>Cp.(Ile2091Leu)		de novo	6,9	het	de novo	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech
MMS22L	NM_198468.2:c.2679+1G>Ap.?	NM_198468.2:c.268A>Gp.(Arg90Gly)	comphet	5,4	comphet	maternal&paternal	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech
CHD6	NM_032221.4:c.1678C>Ap.(Gln560Lys)	NM_032221.4:c.2224A>Gp.(Arg742Gly)	comphet	6,6	comphet	maternal&paternal	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech
ZNF804A	NM_194250.1:c.1049delp.(Gly350Valfs*7)		homo	11,2	homo	maternal&paternal	1	NDD	High palate, Aggressive behavior, Autistic behavior, Intellectual disability, Global developmental delay, Hepatosplenomegaly, Protuberant abdomen, Abnormal facial shape, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Low levels of vitamin D, Self-injurious behavior, Decreased serum iron
GIPR	NM_000164.3:c.784C>Gp.(Leu262Val)	NM_000164.3:c.393G>Tp.(Arg131Ser)	comphet	4,0	comphet	maternal&paternal	1	NDD	Absent speech, Obesity, Intellectual disability, severe
TENM2	NM_001122679.1:c.3881C>Gp.(Ser1294Cys)		het	4,5	het	unknown	1	NDD + Epilepsy	atonic-astatic seizures and mild intellectual disability
KCTD8	NM_198353.2:c.82G>Cp.(Ala28Pro)		de novo	5,4	het	de novo	1	NDD	Regressive global developmental delay with intellectual disability, attention deficit disorder, dysplasia of the corpus callosum, obesity grade 1
TIAM2	.3:c.4679_4681dup, p.(Asn1560_Leu1561insHis)		homo	5,7	homo	maternal&paternal	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia
CASZ1	NM_001079843.2:c.4004G>Ap.(Arg1335His)		homo	5,9	homo	maternal&paternal	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia
PLEKHB1	NM_021200.2:c.164A>Cp.(His55Pro)		homo	6,7	homo	maternal&paternal	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia
BARX2	NM_003658.4:c.386G>Ap.(Arg129Gln)		homo	6,7	homo	maternal&paternal	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia
SKOR2	NM_001278063.1:c.2752+1G>Tp.?		homo	9,0	homo	unknown	3	NDD	Short stature, microcephaly, mild intellectual disability, hyperopia
FMNL3	NM_175736.4:c.2575C>Tp.(Arg859Trp)		homo	6,1	homo	unknown	3	NDD	Short stature, microcephaly, mild intellectual disability, hyperopia
ARHGEF10L	M_018125.3:c.354_355delCCinsTTp.(Arg119Trp)		homo	6,0	homo	maternal&paternal	1	NDD + Epilepsy	Seizures, Ataxia, Spasticity, Focal clonic seizures, Myoclonic spasms, Generalized dystonia, Focal-onset seizure, Focal myoclonic seizures
SPTB	NM_001024858.2:c.610G>Ap.(Asp204Asn)	NM_001024858.2:c.5063A>Gp.(Asn1688Ser)	comphet	5,2	comphet	maternal&paternal	1	NDD	Global developmental delay, Leukopenia, Leukemia, Acute lymphoblastic leukemia
USP13	NM_003940.2:c.2498+1G>Ap.?		de novo	6,4	het	de novo	1	NDD	Renal dysplasia, Polycystic kidney dysplasia, Synophrys, Global developmental delay
SNX8	NM_013321.3:c.922C>Tp.(Gln308*)		de novo	B	het	de novo	2	Growth, Skeletal	Growth delay, short stature, intrauterine growth retardation, Silver-Russell-like appearance
ZNF449	NM_152695.5:c.1394G>Ap.(Cys465Tyr)		de novo	B	het	de novo	2	Growth, Skeletal	Growth delay, short stature, intrauterine growth retardation, Silver-Russell-like appearance
MAGED1	NM_001005332.1:c.640A>Gp.(Thr214Ala)		hemi	4,7	hemi	maternal	1	NDD	Early onset autism
SLC38A1	NM_001278390.1:c.529A>Gp.(Ile177Val)			5,5	het		1	Neuro	Seizure, Tremor, Hand tremor, Nevus, Focal-onset seizure, Abnormality of brain
ZSCAN10	NM_032805.2:c.1436C>Ap.(Ser479Tyr)	NM_032805.2:c.2245G>Tp.(Ala749Ser)	comphet	3,5	comphet	maternal&paternal	3	NDD + Epilepsy	Seizure, Global developmental delay, Gait ataxia, Bilateral tonic-clonic seizure, Unsteady gait, Focal-onset seizure, Cognitive impairment, Mild malformation of cortical development
FLYWCH1	NM_001308068.1:c.2112-3T>Gp.?	NM_001308068.1:c.1111A>Tp.(Ser371Cys)	comphet	6,0	comphet	maternal&paternal	3	NDD + Epilepsy	Seizure, Global developmental delay, Gait ataxia, Bilateral tonic-clonic seizure, Unsteady gait, Focal-onset seizure, Cognitive impairment, Mild malformation of cortical development
HEPHL1	NM_001098672.1:c.1097G>Ap.(Cys366Tyr)		de novo	A	het	de novo	2	Connective Tissue	Syncope, Joint hypermobility, Recurrent fractures, Chronic pain, Dysesthesia
COG6	NM_020751.2:c.1209T>Gp.(Ile403Met)		de novo	B	het	de novo	2	Connective Tissue	Syncope, Joint hypermobility, Recurrent fractures, Chronic pain, Dysesthesia
ZBTB34	NM_001099270.1:c.18delp.(Phe6Leufs*14)		de novo	8,2	het	de novo	1	NDD + Epilepsy	Delayed speech and language development, Global developmental delay, Focal-onset seizure, Childhood onset
PODN	NM_001199080.2:c.559-1G>Cp.?		de novo	B	het	de novo	2	Growth, Skeletal	Joint hypermobility, Asymmetry of the thorax, Scoliosis
GORAB	NM_152281.2:c.383T>Cp.(Ile128Thr)		de novo	C	het	de novo	2	Growth, Skeletal	Joint hypermobility, Asymmetry of the thorax, Scoliosis
GIT2	NM_057169.4:c.699T>Gp.(Tyr233*)		de novo	A	het	de novo	1	Growth, Skeletal	Failure to thrive, Small for gestational age, Short stature, Decreased body weight, Attention deficit hyperactivity disorder, Focal-onset seizure, Abnormal growth hormone level
NDST1	NM_001543.4:c.2468G>Ap.(Gly823Glu)		de novo	8,6	het	de novo	1	NDD + Epilepsy	Focal seizures with cyanosis, sec. generalizing, EEG highly pathological, so far no cMRI examination has been carried out
TTC3	NM_001320703.1:c.3970G>Ap.(Glu1324Lys)		de novo	5,2	het	de novo	1	NDD	Abnormality of the kidney, Global developmental delay, Hip dysplasia, Short stature

ASXL2	NM_018263.4:c.1894C>Gp.(His632Asp)		de novo	8,0	het	de novo	2	NDD	Seizures, Generalized tonic-clonic seizures, Myoclonic atonic seizures, Epileptic encephalopathy
TBCCD1	NM_001134415.1:c.1392T>Gp.(Cys464Trp)		de novo	B	het	de novo	3	Metabolism	Ketotic hypoglycemia
MRM3	NM_018146.3:c.173C>Gp.(Pro58Arg)		de novo	B	het	de novo	3	Metabolism	Ketotic hypoglycemia
PACSIN3	NM_001184974.1:c.604-3C>Gp.?		de novo	B	het	de novo	3	Metabolism	Ketotic hypoglycemia
MDN1	NM_014611.2:c.13276C>Gp.(Leu4426Val)		de novo	6,8	het	de novo	2	NDD + Epilepsy	Microcephaly, Seizure, Dystonia, Cerebral palsy, Abnormality of movement, Epileptic encephalopathy
MAP7D1	NM_018067.4:c.1225G>Tp.(Ala409Ser)		homo	3,5	homo	maternal&paternal	1	NDD + Epilepsy	Infantile febrile seizures and tonic-clonic seizures with aura, despite current treatment with valproate, seizures continue
CPLX1	NM_006651.3:c.250dup, p.(Ala84Glyfs*256)		het	9,3	het	unknown	3	NDD + Epilepsy	Global developmental delay and obsessive-compulsive behavior, seizures
HEATR1	3072.5:c.3949-26_3954delp.(Asp1317Valfs*827)		het	6,9	het	unknown	3	NDD + Epilepsy	Global developmental delay and obsessive-compulsive behavior, seizures
HS6ST2	NM_001077188.1:c.853T>Gp.(Trp285Gly)		hemi	5,6	hemi	maternal	1	NDD	global developmental delay, focal epilepsy, absent speech, Delayed gross motor development, Tetraparesis, Facial palsy
USP4	NM_003363.3:c.1748A>Gp.(Tyr583Cys)		homo	A	homo	maternal&paternal	2	Metabolism	Myalgia, Hyperlipoproteinemia, Increased erythrocyte protoporphyrin concentration, Angioedema
DNHD1	NM_144666.2:c.7549C>Tp.(Arg2517Cys)	NM_144666.2:c.2104-4T>Ap.?	comphet	4,2	comphet	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Seizure, Dystonia, Cerebral palsy, Abnormality of movement, Epileptic encephalopathy
FBN3	NM_032447.4:c.7780G>Ap.(Val2594Ile)	NM_032447.4:c.1135C>Tp.(Arg379*)	comphet	C	comphet	maternal&paternal	3	Metabolism	Obesity, Increased adipose tissue, Glioma, Class III obesity, Overweight, Brain neoplasm
SDR42E1	NM_145168.2:c.4G>Ap.(Asp2Asn)		homo	C	homo	maternal&paternal	3	Metabolism	Obesity, Increased adipose tissue, Glioma, Class III obesity, Overweight, Brain neoplasm
FADS1	NM_013402.4:c.247G>Tp.(Ala83Ser)		de novo	A	het	de novo	2	Other	Anemia, Fever, Recurrent fever, Refractory anemia
TPR	NM_003292.2:c.1038A>Gp.(Ile346Met)	NM_003292.2:c.2380T>Ap.(Ser794Thr)	comphet	C	comphet	maternal&paternal	2	Other	Anemia, Fever, Recurrent fever, Refractory anemia
ZNF449	NM_152695.5:c.961A>Tp.(Lys321*)		de novo	6,6	hemi	de novo	1	NDD + Epilepsy	Hypothyroidism, Primary hypothyroidism, Congenital hypothyroidism, Seizure, Generalized-onset seizure, Atonic seizure, Focal emotional seizure with laughing, Clonic seizure
DOHH	NM_001145165.1:c.446C>Gp.(Pro149Arg)	NM_001145165.1:c.224T>Gp.(Val75Gly)	comphet	6,8	comphet	maternal&paternal	1	NDD + Epilepsy	Global developmental delay, Epilepsy since the age of 3 with tonic-clonic seizures, EEG abnormalities, pain insensitivity
ABCB10	M_012089.2:c.833_838delp.(Asp278_Thr279del)		de novo	4,8	het	de novo	2	NDD	Renal duplication, Global developmental delay, Annular pancreas, Esophageal atresia, Duodenal atresia, Tracheoesophageal fistula, Short stature, Partially duplicated kidney, Anorectal anomaly, Duodenal stenosis, Rectovestibular fistula
DLGAP1	NM_004746.3:c.1018C>Tp.(Arg340*)		de novo	11,8	het	de novo	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech
DAAM2	NM_001201427.1:c.1339C>Gp.(Gln447Glu)	NM_001201427.1:c.1745C>Ap.(Pro582His)	comphet	4,7	comphet	maternal&paternal	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech
SIGLEC9	NM_001198558.1:c.682G>Ap.(Val228Ile)		de novo	3,7	het	de novo	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech
CDH13	NM_001220488.1:c.2228G>Ap.(Arg743His)	NM_001220488.1:c.1505C>Tp.(Ser502Phe)	comphet	5,2	comphet	maternal&paternal	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech
ASIC1	NM_020039.3:c.1116T>Ap.(Tyr372*)		het	6,1	het	unknown	1	NDD + Epilepsy	Behavioral abnormality, Autistic behavior, Delayed speech and language development, Seizure, Pyloric stenosis, Attention deficit hyperactivity disorder
PKHD1L1	NM_177531.4:c.5194C>Tp.(Pro1732Ser)	NM_177531.4:c.8005C>Tp.(Gln2669*)	comphet	5,0	comphet	maternal&paternal	2	NDD + Epilepsy	Seizure, Status epilepticus, EEG abnormality, Focal impaired awareness seizure, Focal-onset seizure, EEG with focal spike waves
LAMA5	NM_005560.4:c.8632G>Ap.(Val2878Ile)	NM_005560.4:c.6578G>Ap.(Arg2193His)	comphet	5,4	comphet	maternal&paternal	2	NDD + Epilepsy	Seizure, Status epilepticus, EEG abnormality, Focal impaired awareness seizure, Focal-onset seizure, EEG with focal spike waves
PRICKLE1	NM_153026.2:c.128A>Gp.(Glu43Gly)		de novo	8,7	het	de novo	1	NDD + Epilepsy	Global developmental delay with a decreased and autistic spectrum disorder characteristics, attends a special school, MRI and EEG inconspicuous
CCDC66	M_001141947.1:c.847_848delp.(Glu283Serfs*3)		homo	8,0	homo	maternal&paternal	3	NDD + Epilepsy	Global developmental delay with delayed speech, astatic attacks, absence epilepsy and EEG abnormalities
RNF103-CHMP3	NM_001198954.1:c.307G>Ap.(Val103Met)		de novo	6,4	het	de novo	3	NDD + Epilepsy	Global developmental delay with delayed speech, astatic attacks, absence epilepsy and EEG abnormalities
RGPD8	NM_001164463.1:c.3225G>Tp.(Gln1075His)		de novo	4,6	het	de novo	3	NDD + Epilepsy	Global developmental delay with delayed speech, astatic attacks, absence epilepsy and EEG abnormalities
NUMBL	NM_004756.4:c.1193C>Ap.(Pro398His)		homo	5,4	homo	maternal&paternal	2	NDD	Long palpebral fissure, Prominent fingertip pads, Intellectual disability, Large fleshy ears
ATP13A4	NM_032279.3:c.826G>Ap.(Glu276Lys)		homo	6,0	homo	maternal&paternal	2	NDD	Long palpebral fissure, Prominent fingertip pads, Intellectual disability, Large fleshy ears
UBR5	NM_015902.5:c.3682C>Tp.(Pro1228Ser)		de novo	8,9	het	de novo	1	NDD + Epilepsy	Epilepsy associated with fever or infection, tonic-clonic seizures, mild mental retardation, macrocephaly and sleep EEG with sharp slow waves
NPTN	NM_012428.3:c.1025C>Tp.(Pro342Leu)		de novo	8,7	het	de novo	2	NDD	Autism, Delayed speech and language development, Intellectual disability, Global developmental delay, Diarrhea, Macrocephaly, Partial Epilepsy
USP8	NM_001128610.2:c.2658+2_2658+3insAAGAp.?	NM_001128610.2:c.2371A>Gp.(Ile791Val)	comphet	5,9	comphet	maternal&paternal	1	neuro	Spasticity, Intention tremor, Vertigo, Dyskinesia
VPS51	NM_013265.3:c.1777A>Gp.(Lys593Glu)		de novo	6,7	het	de novo	3	NDD + Epilepsy	Epilepsy with generalized tonic-clonic seizures, ED 10/2019, microcephaly
RNF144A	NM_014746.4:c.428G>Cp.(Cys143Ser)		homo	5,2	homo	maternal	3	NDD + Epilepsy	Epilepsy with generalized tonic-clonic seizures, ED 10/2019, microcephaly
SCN7A	NM_002976.3:c.2932A>Gp.(Ile978Val)		homo	5,8	homo	maternal	3	NDD + Epilepsy	Epilepsy with generalized tonic-clonic seizures, ED 10/2019, microcephaly

UTP18	NM_016001.2:c.1503+1G>Ap.?		de novo	6,2	het	de novo	2	NDD + Epilepsy	Epilepsy (post- brain haemorrhage condition), intelligence impairment, autism, seizures, premature birth
GCNA	NM_052957.4:c.673C>Ap.(Pro225Thr)		de novo	4,1	het	de novo	2	NDD + Epilepsy	Epilepsy (post- brain haemorrhage condition), intelligence impairment, autism, seizures, premature birth
RYR3	NM_001036.4:c.2770A>Gp.(Thr924Ala)	NM_001036.4:c.11246-5C>Gp.?	comphet	6,1	comphet	maternal&paternal	1	Neuro	Torticollis, Ataxia, Dysarthria, Dystonia, Slurred speech, Gait ataxia, Limb ataxia, Truncal ataxia, Episodic ataxia, Apraxia, Limb dystonia, Focal dystonia, Gait apraxia, Oromandibular dystonia
MTCH1	NM_001271641.1:c.2T>Ap.0?		homo	6,6	homo	maternal&paternal	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure
KCNG4	NM_172347.2:c.1022C>Tp.(Ala341Val)		homo	5,8	homo	maternal&paternal	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure
KIAA1107	NM_015237.3:c.299C>Tp.(Thr100Ile)		homo	3,9	homo	maternal&paternal	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure
CRYBG3	NM_153605.3:c.8492G>Ap.(Arg2831His)		homo	4,8	homo	maternal&paternal	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure
PDE4DIP	NM_001198834.3:c.6862A>Cp.(Lys2288Gln)	NM_001198834.3:c.6043A>Gp.(Ile2015Val)	comphet	6,2	comphet	maternal&paternal	1	NDD	Intellectual disability, Global developmental delay, Motor delay, Failure to thrive, Increased serum lactate, Infantile muscular hypotonia, Delayed myelination, Alaninuria
TCEAL3	NM_001006933.1:c.585C>Gp.(His195Gln)		hemi	3,7	hemi	maternal	1	NDD	Global developmental delay, Gait ataxia, Infantile muscular hypotonia
PLEKHM3	NM_001080475.2:c.2219G>Ap.(Arg740Lys)		de novo	4,4	het	de novo	2	Neuro	Gait disturbance, Dystonia, Progressive spastic paraplegia, Paraplegia, Leg dystonia
GPX4	NM_001039848.3:c.587+5G>Ap.?	NM_001039848.3:c.475G>Tp.(Gly159Cys)	comphet	5,8	comphet	maternal&paternal	2	Neuro	Gait disturbance, Dystonia, Progressive spastic paraplegia, Paraplegia, Leg dystonia
KLHDC4	NM_017566.3:c.908T>Cp.(Met303Thr)	NM_017566.3:c.529C>Tp.(Arg177Trp)	comphet	3,6	comphet	maternal&paternal	1	NDD + Epilepsy	Neurodevelopmental delay, Global developmental delay, Infantile spasms, Seizure, Epileptic spasm, Abnormal nervous system physiology, Neonatal seizure
TDRD9	NM_153046.2:c.2273C>Tp.(Pro758Leu)		homo	4,2	homo	maternal&paternal	2	NDD	Autism, Hypertrichosis, Intellectual disability, Global developmental delay, Absent speech, Mutism
TEC	NM_003215.2:c.1526G>Tp.(Gly509Val)		het	C	het	maternal	2	Immunology	recurrent purulent abscess of the groin
RAB11FIP4	NM_032932.5:c.1562G>Ap.(Gly521Asp)		het	C	het	maternal	2	Immunology	recurrent purulent abscess of the groin
ITSN1	NM_003024.2:c.1690T>Cp.(Ser564Pro)		het	6,6	het	unknown	5	NDD + Epilepsy	Macrocephaly, Aggressive behavior, Delayed speech and language development, Enuresis, Intellectual disability, Seizure, Global developmental delay, Intellectual disability, moderate, Febrile seizure (within the age range of 3 months to 6 years), Polyphagia, Increased adipose tissue, Feeding difficulties, Cognitive impairment, Overweight
DYNC1I1	NM_004411.4:c.1421C>Gp.(Ala474Gly)		het	4,5	het	unknown	5	NDD + Epilepsy	Macrocephaly, Aggressive behavior, Delayed speech and language development, Enuresis, Intellectual disability, Seizure, Global developmental delay, Intellectual disability, moderate, Febrile seizure (within the age range of 3 months to 6 years), Polyphagia, Increased adipose tissue, Feeding difficulties, Cognitive impairment, Overweight
TMEM63A	NM_014698.2:c.1423T>Cp.(Phe475Leu)		het	3,5	het	unknown	5	NDD + Epilepsy	Macrocephaly, Aggressive behavior, Delayed speech and language development, Enuresis, Intellectual disability, Seizure, Global developmental delay, Intellectual disability, moderate, Febrile seizure (within the age range of 3 months to 6 years), Polyphagia, Increased adipose tissue, Feeding difficulties, Cognitive impairment, Overweight
MTR	NM_000254.2:c.2812A>Gp.(Ser938Gly)		het	6,2	het	unknown	5	NDD + Epilepsy	Macrocephaly, Aggressive behavior, Delayed speech and language development, Enuresis, Intellectual disability, Seizure, Global developmental delay, Intellectual disability, moderate, Febrile seizure (within the age range of 3 months to 6 years), Polyphagia, Increased adipose tissue, Feeding difficulties, Cognitive impairment, Overweight
HNRNPM	NM_005968.4:c.23C>Tp.(Ala8Val)		homo	5,0	homo	maternal&paternal	2	NDD + Epilepsy	generalized epilepsy with nocturnal tonic-clonic seizures (onset in the 2nd year of life), mild intellectual impairment
DUSP9	NM_001318503.1:c.745G>Ap.(Asp249Asn)		hemi	3,8	hemi	maternal	2	NDD + Epilepsy	generalized epilepsy with nocturnal tonic-clonic seizures (onset in the 2nd year of life), mild intellectual impairment
LRRC7	NM_001330635.1:c.2143C>Tp.(Gln715*)		het	7,5	het	unknown	1	NDD	Intellectual disability, Global developmental delay,Overweight
SLC4A2	NM_003040.3:c.2507T>Cp.(Ile836Thr)		de novo	7,0	het	de novo	1	NDD + Epilepsy	Global developmental delay with intelligence impairment and speech delay; epilepsy with tonic-clonic seizures and atypical absences (pseudo-Lennox); short stature; hypercholesterinemia
UTP14A	NM_006649.3:c.124A>Gp.(Lys42Glu)		hemi	4,1	hemi	maternal	2	NDD + Epilepsy	Epileptic encephalopathy, Seizure since the age of 11
SMURF1	NM_020429.2:c.1390C>Tp.(Gln464*)		de novo	9,6	het	de novo	2	NDD	Premature infant (32 weeks, 1600g), maldescensus testis bilateral, plagiocephalus, central motor coordination and movement disorder with dystonic movements, trunk muscular hypotension, delayed development, MRI: subependymal left heterotopia, steep tentorium, small posterior fossa, compressed 4th ventricle, flattened skull on the right
SPHK2	NM_001204159.2:c.1774delp.(His592Thrfs*19)		de novo	6,8	het	de novo	2	NDD	Premature infant (32 weeks, 1600g), maldescensus testis bilateral, plagiocephalus, central motor coordination and movement disorder with dystonic movements, trunk muscular hypotension, delayed development, MRI: subependymal left heterotopia, steep tentorium, small posterior fossa, compressed 4th ventricle, flattened skull on the right
SF3A1	NM_005877.5:c.709C>Tp.(Arg237*)		het	6,5	het	maternal	1	NDD	Neurodevelopmental delay, Bifid uvula, Global developmental delay, Atrial septal defect, Hypoglycemia, Abnormal blood glucose concentration, Mild global developmental delay, Moderate global developmental delay, Abnormal glycosylation
C7orf26	NM_024067.3:c.575_576insTp.(Ser193Ilefs*3)		de novo	A	het	de novo	2	Immunology	Abnormal facial shape, Alopecia areata, Recurrent infections, Antinuclear antibody positivity, Paroxysmal nocturnal hemoglobinuria, Prolonged neonatal jaundice
POLR2C	NM_032940.2:c.109delp.(Val37Serfs*8)		de novo	B	het	de novo	2	Immunology	Abnormal facial shape, Alopecia areata, Recurrent infections, Antinuclear antibody positivity, Paroxysmal nocturnal hemoglobinuria, Prolonged neonatal jaundice

PLXNA1	NM_032242.3:c.475T>Cp.(Tyr159His)		het	4,6	het	unknown	2	NDD + Epilepsy	Developemntal regression with intellectual impairment and behavioral problems; Refractory epilepsy with tonic-clonic seizures, myoclonic seizures, status epilepticus, absence epilepsy and febrile seizures with an onset at the age of 1.5 years,
ZNF182	NM_001178099.1:c.181A>Gp.(Ser61Gly)		hemi	4,8	hemi	maternal	1	NDD + Epilepsy	Seizure, Nocturnal seizures
AHCTF1	015446.4:c.5018_5019insGGp.(Ile1673Metfs*4)		de novo	A	het	de novo	3	Growth, Skeletal	Panhypopituitarism, Proportionate short stature, Short stature, Anterior pituitary hypoplasia, Ectopic posterior pituitary, Posterior pituitary hypoplasia
EFHC2	NM_025184.3:c.975A>Cp.(Leu325=)		hemi	C	hemi	maternal	3	Growth, Skeletal	Panhypopituitarism, Proportionate short stature, Short stature, Anterior pituitary hypoplasia, Ectopic posterior pituitary, Posterior pituitary hypoplasia
TDRD6	NM_001010870.2:c.1895A>Gp.(His632Arg)	NM_001010870.2:c.2566G>Ap.(Asp856Asn)	comphet	C	comphet	maternal&paternal	3	Growth, Skeletal	Panhypopituitarism, Proportionate short stature, Short stature, Anterior pituitary hypoplasia, Ectopic posterior pituitary, Posterior pituitary hypoplasia
SBNO2	NM_001100122.1:c.1960G>Tp.(Val654Leu)		de novo	4,7	het	de novo	2	NDD + Epilepsy	Epileptic encephalopathy, intrauterine hydrocephalus (gestational week 24), epilepsy
APBA1	NM_001163.3:c.521T>Cp.(Leu174Pro)		de novo	8,4	het	de novo	2	NDD + Epilepsy	Epileptic encephalopathy, intrauterine hydrocephalus (gestational week 24), epilepsy
NRXN3	NM_001330195.1:c.115C>Tp.(Arg39Cys)		homo	8,9	homo	maternal&paternal	3	NDD	Global developmental delay and severe motor delay, cerebral palsy (GMFCS level 5), hypoplasia of the corpus callosum
MCTP2	NM_018349.3:c.409G>Ap.(Gly137Ser)	NM_018349.3:c.1889C>Tp.(Pro630Leu)	comphet	3,5	comphet	maternal&paternal	2	NDD	Global developmental delay, macrocephaly and makrosomia, muscular hypotonia and ischemic stroke at four months of age
BMP4	NM_001202.5:c.172G>Cp.(Glu58Gln)		homo	8,4	homo	maternal&paternal	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia
NIF3L1	NM_001136039.2:c.860C>Tp.(Thr287Ile)		homo	5,4	homo	maternal&paternal	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia
MYOF	NM_013451.3:c.3511C>Tp.(Arg1171Trp)	NM_013451.3:c.4268delp.(Pro1423Hisfs*21)	comphet	5,7	comphet	maternal&paternal	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia
DLC1	NM_182643.2:c.609A>Cp.(Lys203Asn)		homo	5,4	homo	maternal&paternal	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia
RNF213	NM_001256071.2:c.9611A>Gp.(His3204Arg)		de novo	6,8	het	de novo	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia
PARPBP	NM_001319988.1:c.62G>Tp.(Arg21Leu)		de novo	B	het	de novo	2	Metabolism	Hypertension, Irregular menstruation, Obesity, Hyperuricemia, Red hair, Narcolepsy, Increased blood pressure, Adipositas since the age of five to eight years
SFRP5	NM_003015.3:c.872T>Cp.(Met291Thr)	NM_003015.3:c.861_863delp.(Lys287del)	comphet	C	comphet	maternal&paternal	2	Metabolism	Hypertension, Irregular menstruation, Obesity, Hyperuricemia, Red hair, Narcolepsy, Increased blood pressure, Adipositas since the age of five to eight years
C6orf136	NM_001161376.1:c.478G>Tp.(Ala160Ser)	NM_001161376.1:c.430C>Tp.(Arg144Trp)	comphet	B	comphet	maternal&paternal	1	Metabolism	Hypoglycemia, Neonatal hypoglycemia
SPART	NM_001142294.1:c.1655T>Gp.(Val552Gly)	NM_001142294.1:c.848C>Tp.(Ser283Phe)	comphet	9,4	comphet	maternal&paternal	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology
NUSAP1	NM_016359.4:c.1213C>Tp.(Gln405*)		de novo	6,1	het	de novo	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology
PDE4DIP	NM_001198834.3:c.3733G>Ap.(Ala1245Thr)	31delAATinsTAGp.(Glu410_Leu411delinsValVal)	comphet	6,7	comphet	maternal&paternal	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology
ZNF611	NM_001161499.1:c.1904C>Tp.(Ser635Leu)		homo	4,1	homo	maternal&paternal	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology
KIR3DL3	NM_153443.4:c.1053G>Ap.(Lys351=)		homo	4,0	homo	maternal&paternal	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology
TMEM181	NM_020823.1:c.448delp.(Ser150Profs*18)	NM_020823.1:c.1781C>Tp.(Pro594Leu)	comphet	B	comphet	maternal&paternal	1	Malformations	Growth delay, Intrauterine growth retardation
HSP90AA1	NM_001017963.2:c.626G>Ap.(Arg209Gln)		homo	7,7	homo	maternal&paternal	3	NDD	Global developmental delay and severe motor delay, cerebral palsy (GMFCS level 5), hypoplasia of the corpus callosum
FAM200A	NM_145111.3:c.1702C>Tp.(Gln568*)		homo	6,4	homo	maternal&paternal	5	NDD + Epilepsy	Intellectual disability, Seizure
POLL	NM_001174084.1:c.1255C>Tp.(Arg419*)		homo	7,8	homo	maternal&paternal	5	NDD + Epilepsy	Intellectual disability, Seizure
PYROXD2	NM_032709.2:c.1062+2T>Gp.?		homo	6,8	homo	maternal&paternal	5	NDD + Epilepsy	Intellectual disability, Seizure
TRPV1	NM_018727.5:c.896C>Tp.(Thr299Met)		homo	6,5	homo	maternal&paternal	5	NDD + Epilepsy	Intellectual disability, Seizure
LRCH3	NM_032773.3:c.256C>Tp.(Arg86Trp)		homo	4,4	homo	maternal&paternal	5	NDD + Epilepsy	Intellectual disability, Seizure
CHRD	NM_001304472.1:c.2491C>Gp.(Pro831Ala)		de novo	8,0	het	de novo	3	NDD	Microcephaly, Intellectual disability, Ataxia, Intellectual disability, mild, Global developmental delay, Spastic gait, Gait ataxia, Intention tremor, Spastic ataxia
URB1	NM_014825.2:c.5312A>Tp.(Tyr1771Phe)	NM_014825.2:c.3362G>Ap.(Arg1121His)	comphet	4,2	comphet	maternal&paternal	3	NDD	Microcephaly, Intellectual disability, Ataxia, Intellectual disability, mild, Global developmental delay, Spastic gait, Gait ataxia, Intention tremor, Spastic ataxia
HELZ2	NM_001037335.2:c.7693C>Tp.(Arg2565Cys)	NM_001037335.2:c.1750C>Tp.(Arg584Trp)	comphet	4,3	comphet	maternal&paternal	3	NDD	Microcephaly, Intellectual disability, Ataxia, Intellectual disability, mild, Global developmental delay, Spastic gait, Gait ataxia, Intention tremor, Spastic ataxia
TMEM94	NM_001321148.1:c.1976A>Cp.(Gln659Pro)		het	5,3	het	unknown	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart
CLASP1	NM_015282.2:c.4142T>Ap.(Phe1381Tyr)		het	5,8	het	unknown	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart
EIF4ENIF1	NM_001164501.1:c.1588C>Tp.(Leu530Phe)		het	4,0	het	unknown	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart
IGF2R	NM_000876.3:c.1312G>Ap.(Ala438Thr)	NM_000876.3:c.5506G>Ap.(Val1836Ile)	comphet?	6,3	comphet?	unknown	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart

BTBD6	NM_033271.2:c.223C>Tp.(Leu75Phe)	NM_033271.2:c.835G>Ap.(Ala279Thr)	comphet	5,1	comphet	maternal&paternal	1	NDD	Developmental disorder
ANXA3	NM_005139.2:c.541-2A>Gp.?		het	5,1	het	paternal	1	Neuro	+) Sleep disturbance, Restless legs, Insomnia
SLITRK2	NM_001144003.2:c.265G>Tp.(Val89Leu)		de novo	6,3	het	de novo	1	NDD + Epilepsy	Behavioral abnormality, Intellectual disability, Seizure, Global developmental delay, Short stature, Focal-onset seizure, Focal motor seizure, Generalized-onset motor seizure, Focal-onset epileptic spasm
PPP3CC	NM_001243975.1:c.323G>Ap.(Arg108His)		het	4,0	het	maternal	4	NDD + Epilepsy	Episodic ataxia, EEG abnormality
ANKS1A	NM_015245.2:c.2269C>Tp.(Arg757Cys)		het	4,1	het	maternal	4	NDD + Epilepsy	Episodic ataxia, EEG abnormality
RNF20	NM_019592.6:c.2783G>Ap.(Arg928His)		de novo	A	het	de novo	2	Metabolism	Obesity
HECTD1	NM_015382.3:c.6068G>Tp.(Gly2023Val)		homo	B	homo	maternal&paternal	2	Metabolism	Obesity
PTOV1	NM_017432.4:c.842dup, p.(Pro282Alafs*79)		de novo	8,1	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizure, Global developmental delay, bilateral toni-clonic seizure, atonic seizure
ACTN2	NM_001103.3:c.1108-2A>Tp.?		het	A	het	unknown	1	Muscle	Tetraparesis and muscle weakness since age of 51 years
CACNG3	NM_006539.3:c.437-1G>Ap.?		het	7,6	het	unknown	1	NDD	Microcephaly, Intellectual disability, Global developmental delay, Intellectual disability, moderate, Mild microcephaly, Decreased head circumference
LRRK1	NM_024652.5:c.5615C>Gp.(Ser1872Cys)		de novo	6,9	het	de novo	4	NDD + Epilepsy	Episodic ataxia, EEG abnormality
USP19	NM_001199161.1:c.2012C>Ap.(Ser671Tyr)		de novo	6,5	het	de novo	4	NDD + Epilepsy	Episodic ataxia, EEG abnormality
EP400	NM_015409.4:c.4277+1G>Tp.?		het	7,5	het	unknown	4	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Abnormal facial shape, Delayed gross motor development, Abnormal muscle tone, Infantile muscular hypotonia, Delayed fine motor development, Neurodevelopmental delay
WNK2	NM_006648.3:c.5229G>Tp.(Lys1743Asn)		het	3,5	het	unknown	4	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Abnormal facial shape, Delayed gross motor development, Abnormal muscle tone, Infantile muscular hypotonia, Delayed fine motor development, Neurodevelopmental delay
PHC2	NM_198040.2:c.604G>Ap.(Ala202Thr)		het	5,0	het	unknown	4	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Abnormal facial shape, Delayed gross motor development, Abnormal muscle tone, Infantile muscular hypotonia, Delayed fine motor development, Neurodevelopmental delay
CREB5	NM_182898.3:c.302T>Ap.(Met101Lys)		het	3,7	het	unknown	4	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Abnormal facial shape, Delayed gross motor development, Abnormal muscle tone, Infantile muscular hypotonia, Delayed fine motor development, Neurodevelopmental delay
ATP6AP1	NM_001183.5:c.3G>Tp.0?		hemi	8,3	hemi	unknown	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart
LAMP2	NM_001122606.1:c.731C>Gp.(Thr244Ser)		hemi	B	hemi	maternal	2	Metabolism	Myalgia, Hyperlipoproteinemia, Increased erythrocyte protoporphyrin concentration, Angioedema
PMM1	NM_002676.2:c.416C>Tp.(Ser139Leu)		de novo	6,8	het	de novo	2	NDD + Epilepsy	Epileptic encephalopathy, Seizure since the age of 11
EIF2S2	NM_003908.4:c.692G>Ap.(Arg231His)		de novo	8,4	het	de novo	2	NDD	Inguinal hernia, Intellectual disability, Global developmental delay, Small for gestational age, Penile hypospadias, Short stature, Decreased body weight
ZBTB45	NM_001316978.1:c.655G>Ap.(Asp219Asn)		de novo	5,3	het	de novo	2	NDD	Inguinal hernia, Intellectual disability, Global developmental delay, Small for gestational age, Penile hypospadias, Short stature, Decreased body weight
LAMA5	NM_005560.4:c.5408C>Tp.(Ser1803Phe)		homo	6,3	homo	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Pes planus, Focal tonic seizure, Cognitive impairment
REST	NM_005612.4:c.2227G>Ap.(Glu743Lys)		homo	7,9	homo	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Pes planus, Focal tonic seizure, Cognitive impairment
DBN1	NM_080881.2:c.1452C>Gp.(Asn484Lys)	080881.2:c.1663_1664delTCinsCTp.(Pro555Leu)	comphet	5,6	comphet	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Pes planus, Focal tonic seizure, Cognitive impairment
PRMT9	NM_138364.3:c.1144C>Ap.(Gln382Lys)		homo	4,6	homo	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Pes planus, Focal tonic seizure, Cognitive impairment
CXorf21	NM_025159.2:c.774A>Cp.(Arg258Ser)		hemi	4,5	hemi	maternal	2	NDD + Epilepsy	Leukenzephalopathie, keine Sprache, muskuläre Hypertonie, schwere motorische und sprachlich-mentale Entwicklungsverzögerung, therapierefraktäre Krampfanfälle
OSBPL9	NM_148909.3:c.413_422delp.(Ser138Ilefs*16)		het	6,2	het	unknown	1	NDD + Epilepsy	Seizure, Generalized non-motor (absence) seizure, Generalized myoclonic seizure, Atypical absence seizure, Eyelid myoclonia seizure, Myoclonic seizure
FHOD3	NM_001281740.2:c.1836-2A>Gp.?		het	5,5	het	unknown	1	NDD + Epilepsy	Entwicklungsstörung, Epilepsie (Absencen, Grand-mal-Anfälle)
ARHGEF28	NM_001080479.2:c.548T>Gp.(Leu183Trp)		de novo	7,0	het	de novo	1	NDD	Abnormality of body height, Tall stature, Epicanthus, Abnormality of the nasal bridge, Abnormal eyelid morphology, Widely spaced teeth, Behavioral abnormality, Restlessness, Low frustration tolerance, Delayed speech and language development, Global developmental delay, Large for gestational age, Abnormal facial shape, Increased body weight, Depressed nasal bridge
ASB13	NM_024701.3:c.404T>Gp.(Leu135Arg)		het	C	het	unknown	1	Immunology	Immunodeficiency, Autoimmunity, Decreased antibody level in blood, Combined immunodeficiency, Abnormal immunoglobulin level, Abnormality of immune system physiology

NOL4	NM_003787.4:c.1A>Cp.0?		het	5,6	het	maternal	1	NDD	Microcephaly, Aggressive behavior, Intellectual disability, Intellectual disability, mild, Abnormal aggressive, impulsive or violent behavior
TMEM232	NM_001039763.3:c.884A>Gp.(Gln295Arg)		de novo	5,6	het	de novo	1	NDD + Epilepsy	epilepsy, movement disorder, syncope, myoclonia, pathological waking EEG, cystic, malformation of the right kidney
NCOA1	NM_003743.4:c.3457C>Tp.(Gln1153*)		het	8,3	het	unknown	2	NDD	Intellectual disability, Global developmental delay, Obesity, Abnormal heart morphology, Truncal obesity, Kyphosis, Attention deficit hyperactivity disorder
ARHGAP35	NM_004491.4:c.597_600delp.(Thr200Serfs*18)		de novo	A	het	de novo	1	Immunology	Splenomegaly, Lymphopenia, recurrent infections, immunodeficiency, decreased circulating IGA, increased circulating IgM level
APLNR	NM_005161.4:c.952C>Tp.(Gln318*)		de novo	7,4	het	de novo	1	NDD	Coarse facial features, Hearing impairment, Delayed speech and language development, Intellectual disability, Global developmental delay, Absent speech, Failure to thrive, Premature birth, Atrial septal defect, Hypoglycemia, Abnormal facial shape, Expressive language delay, Decreased circulating IgA level,(-) Recurrent upper respiratory tract infections, Decreased body weight,(-) Intestinal obstruction, Decreased circulating IgG2 level, Respiratory tract infection, Psychomotor retardation, Decreased body mass index, Decreased circulating IgG subclass level, Decreased circulating IgG1 level
RBFOX1	NM_001308117.1:c.445C>Gp.(Pro149Ala)	NM_001142333.1:c.1069G>Cp.(Ala357Pro)	comphet	7,1	comphet	maternal&paternal	2	NDD	global muscular hypotension with axial weakness, facial dysmorphia, indicated high palate, broad neck, muscle relief decreasing distally, hypersalivation, no secure free sitting
PCID2	NM_001258212.1:c.835G>Ap.(Asp279Asn)	NM_001258212.1:c.35A>Gp.(Gln12Arg)	comphet	4,2	comphet	maternal&paternal	2	NDD	global muscular hypotension with axial weakness, facial dysmorphia, indicated high palate, broad neck, muscle relief decreasing distally, hypersalivation, no secure free sitting
KPNA1	NM_002264.3:c.1015G>Ap.(Ala339Thr)		de novo	7,4	het	de novo	1	NDD	Global developmental delay and speech delay, microcephaly
UBR2	NM_015255.2:c.2462+2T>Cp.?		het	7,2	het	unknown	1	NDD	Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Obesity, Expressive language delay, Abnormality of body weight, Increased body weight, Increased adipose tissue, Neurodevelopmental delay, Overweight
KCND2	NM_012281.2:c.1003A>Gp.(Ile335Val)		het	6,3	het	unknown	1	NDD + Epilepsy	Abnormality of vision, Visual impairment, Seizure, Ataxia, Motor delay, Vomiting, Migraine, Episodic ataxia, Episodic vomiting, Poor fine motor coordination, Focal-onset seizure, Delayed fine motor development, Fatigue, Episodic fatigue
GNAI1	NM_002069.5:c.430C>Tp.(Arg144*)		het	8,5	het	unknown	1	NDD	Intellectual disability, Muscular hypotonia Global developmental delay, Bilateral ptosis, Ventricular septal defect, Abnormal facial shape, Migraine, Neurodevelopmental delay, Neurodevelopmental abnormality
DLG5	NM_004747.3:c.453dupp.(Gln152Serfs*26)		het	7,6	het	unknown	1	NDD	Intellectual disability, Seizure, Intellectual disability, mild, Encephalopathy, Bilateral tonic-clonic seizure, Gliosis, Febrile seizure (within the age range of 3 months to 6 years), Aplasia/Hypoplasia involving the central nervous system, Abnormal nervous system physiology, Epileptic encephalopathy, Motor seizure
DSCAML1	NM_020693.3:c.1322C>Tp.(Ser441Phe)		het	4,4	het	maternal	1	NDD + Epilepsy	Intellectual disability, Seizure, Intellectual disability, mild, Pachygyria, Polymicrogyria, Generalized-onset seizure, Abnormal cortical gyration, Focal-onset seizure, Bilateral perisylvian polymicrogyria, Focal polymicrogyria
PSMB6	NM_002798.2:c.238T>Cp.(Ser80Pro)		homo	6,5	homo	maternal&paternal	3	NDD	globale Entwicklungsverzögerung (greifen mit 4,5 Jahren, freies Laufen mit 6 Jahren, erste Worte mit 6 Jahren), Trinkschwäche, Atemaussetzer, Mikrozephalie, chronische Diarrhoe, Dystrophie
ZC3H3	NM_015117.2:c.1595C>Tp.(Thr532Ile)		de novo	5,3	het	de novo	3	NDD	globale Entwicklungsverzögerung (greifen mit 4,5 Jahren, freies Laufen mit 6 Jahren, erste Worte mit 6 Jahren), Trinkschwäche, Atemaussetzer, Mikrozephalie, chronische Diarrhoe, Dystrophie
RNF167	NM_015528.2:c.793C>Gp.(Arg265Gly)		homo	4,6	homo	maternal&paternal	3	NDD	globale Entwicklungsverzögerung (greifen mit 4,5 Jahren, freies Laufen mit 6 Jahren, erste Worte mit 6 Jahren), Trinkschwäche, Atemaussetzer, Mikrozephalie, chronische Diarrhoe, Dystrophie
NKX3-1	NM_006167.3:c.491C>Tp.(Thr164Met)	NM_006167.3:c.113G>Ap.(Gly38Asp)	comphet	4,3	comphet	maternal&paternal	1	NDD + Epilepsy	Intelligenzminderung, Epilepsie, Schwerhörigkeit
TENM2	NM_001122679.1:c.7970C>Tp.(Thr2657Met)		de novo	6,7	het	de novo	3	NDD	Global developmental delay, intellectual impairment, absent speech
SRCIN1	NM_025248.2:c.40C>Tp.(Pro14Ser)		homo	3,4	homo	maternal&paternal	3	NDD	Global developmental delay, intellectual impairment, absent speech
FAT2	NM_001447.2:c.5000T>Gp.(Val1667Gly)		homo	5,1	homo	maternal&paternal	3	NDD	Global developmental delay, intellectual impairment, absent speech
BTBD3	NM_181443.3:c.1502C>Ap.(Pro501Gln)		het	4,7	het	unknown	2	NDD	Intellectual disability, Global developmental delay, Obesity, Abnormal heart morphology, Truncal obesity, Kyphosis, Attention deficit hyperactivity disorder
ATP13A5	NM_198505.2:c.1949A>Gp.(Tyr650Cys)	NM_198505.2:c.1241dup, p.(Tyr415Leufs*72)	comphet?	6,8	comphet?	unknown	1	NDD	epileps, ID, cerebral palsy, EEG and MRI abnormlities
DOCK4	NM_014705.3:c.2945C>Tp.(Thr982Ile)		de novo	8,7	het	de novo	1	NDD + Epilepsy	tonic-clonic seizures, intelligence impairment, tremor, ataxia
RPS6KA6	NM_001330512.1:c.2113-3T>Gp.?		hemi	4,8	hemi	maternal	2	NDD + Epilepsy	nocturnal focal seizures, onset on the age of 4, microcephaly, developmental delay, intelligence impairment (IQ=81), ADHS
SPATA8	NM_173499.4:c.28C>Tp.(Gln10*)		de novo	4,1	het	de novo	2	NDD + Epilepsy	nocturnal focal seizures, onset on the age of 4, microcephaly, developmental delay, intelligence impairment (IQ=81), ADHS
TANC1	NM_001145909.1:c.10G>Cp.(Ala4Pro)	NM_001145909.1:c.1007G>Ap.(Arg336Gln)	comphet	5,3	comphet	maternal&paternal	3	NDD	global developmental delay, absent speech, muscular hypotonia, austism spectrum disorder, microcephaly
RHBDF1	NM_022450.3:c.1082G>Ap.(Arg361His)		de novo	5,6	het	de novo	3	NDD	global developmental delay, absent speech, muscular hypotonia, austism spectrum disorder, microcephaly
FOXO4	NM_005938.3:c.43A>Tp.(Ile15Phe)		hemi	4,9	hemi	maternal	3	NDD	global developmental delay, absent speech, muscular hypotonia, austism spectrum disorder, microcephaly

CMIP	NM_198390.2:c.42del,p.(Gln15Argfs*36)		het	8,0	het	paternal	3	NDD + Epilepsy	Intellectual disability, seizures
MKRN1	NM_013446.4:c.340C>Tp.(Gln114*)		de novo	8,3	het	de novo	1	NDD	Intellectual disability, mild, Global developmental delay, Mild global developmental delay, Short stature, Talipes equinovarus
SLC25A5	NM_001152.4:c.616A>Gp.(Lys206Glu)		hemi	6,6	hemi	maternal	2	NDD	Mild global developmental delay, Cleft palate, Hearing impairment, Ventricular septal defect, Patent ductus arteriosus, Pulmonic stenosis, Hypospadias, Enlarged vestibular aqueduct, Pes cavus, Scoliosis
PCSK6	NM_002570.4:c.412C>Ap.(Leu138Ile)	NM_002570.4:c.2232A>Tp.(Arg744Ser)	comphet	4,4	comphet	maternal&paternal	2	NDD	Mild global developmental delay, Cleft palate, Hearing impairment, Ventricular septal defect, Patent ductus arteriosus, Pulmonic stenosis, Hypospadias, Enlarged vestibular aqueduct, Pes cavus, Scoliosis
RXRB	NM_001270401.1:c.1472C>Ap.(Ala491Asp)		het	4,3	het	unknown	2	NDD	Intellectual disability, severe, Severe global developmental delay, Seizure, Focal-onset seizure, Motor seizure, Cerebral palsy (GMFCS V), Microcephaly, Feeding difficulties no
WDR13	NM_001347217.2:c.194G>Ap.(Arg65His)		hemi	5,7	hemi	unknown	1	NDD + Epilepsy	Global developmental delay, EEG abnormality, Carious teeth, Finger clinodactyly, Decreased head circumference, Intellectual disability
EPHB3	NM_004443.4:c.1711G>Ap.(Val571Met)		homo	4,9	homo	maternal&paternal	2	NDD	Intellectual disability, Seizures (onset at age of 14 years), Sleep disturbance, Hypotonic cerebral palsy
SV2B	NM_001167580.2:c.895C>Tp.(Gln299*)		het	6,4	het	unknown	2	NDD + Epilepsy	Seizure, Focal impaired awareness motor seizure, Generalized-onset seizure, Generalized-onset motor seizure, Bilateral tonic-clonic seizure with generalized onset,(+) Subcortical band heterotopia, Neurodevelopmental abnormality, Specific learning disability Intellectual disability, mild
HOXD4	NM_014621.2:c.111C>Ap.(Tyr37*)		het	4,0	het	unknown	2	NDD + Epilepsy	Seizure, Focal impaired awareness motor seizure, Generalized-onset seizure, Generalized-onset motor seizure, Bilateral tonic-clonic seizure with generalized onset,(+) Subcortical band heterotopia, Neurodevelopmental abnormality, Specific learning disability Intellectual disability, mild
PIK3AP1	NM_152309.3:c.601A>Tp.(Lys201*)		het	5,5	het	unknown	1	NDD + Epilepsy	Epileptic encephalopathy, Cognitive impairment, Microcephaly, Short stature, Febrile
GRM2	NM_000839.3:c.2462C>Tp.(Pro821Leu)		het	4,7	het	unknown	1	NDD + Epilepsy	Atypical absence seizure, Multifocal seizures, Focal impaired awareness seizure, Bilateral tonic-clonic seizure with focal onset, Intellectual disability, mild, Intellectual disability, moderate, Ventricular extrasystoles
TTC28	NM_001145418.1:c.5009A>Tp.(His1670Leu)	NM_001145418.1:c.4237G>Ap.(Gly1413Ser)	comphet	C	comphet	maternal&paternal	1	Growth, Skeletal	Trigonocephaly, Abnormality of calvarial morphology
FNDC3A	NM_001079673.2:c.760+1G>Tp.?		het	6,7	het	unknown	1	NDD + Epilepsy	+) Moderate global developmental delay,(+) Intellectual disability, mild,(+) Aggressive behavior,(+) Pes valgus,(+) Abnormality of the palmar creases,(+) Brachydactyly,(+) Long face,(+) Frontal bossing,(+) Hypertelorism,(+) Epicanthus,(+) Sparse lateral eyebrow,(+) Hypoplastic philtrum,(+) Focal-onset seizure,(+) Cerebral white matter hypoplasia
STRN3	NM_001083893.2:c.542+2T>Gp.?		het	B	het	paternal	2	Growth, Skeletal	Short stature (151cm), Short for gestational age
ATG9A	NM_001077198.3:c.2398C>Tp.(His800Tyr)		het	C	het	paternal	2	Growth, Skeletal	Short stature (151cm), Short for gestational age
ZNF143	NM_003442.5:c.44_45delAGp.Glu15ValfsTer25		homo	8,6	homo	maternal&paternal	1	NDD	NDD
ARHGEF6	NM_004840.2:c.257A>Cp.Ap86Ala		hemi	8,2	hemi	unknown	1	NDD	NDD
FRMD5	NM_032892.5:c.1045A>Cp.(Ser349Arg)		de novo	6,4	het	de novo	1	NDD	Global developmental delay, Expressive language delay, Receptive language delay, Gait ataxia, Muscular hypotonia, Microcephaly, Cerebral atrophy, Abnormality of eye movement
CCNT2	NM_058241.3:c.370-3_370-2insAp.?		de novo	C	het	de novo	1	Immunology	Increased circulating IgG4 level, Neutropenia, Papule, Folliculitis, Lymphangitis, Pustule, Immunodeficiency
MRTFA	NM_001318139.2:c.800delp.(Lys267Argfs*2)		de novo	9,8	het	de novo	3	NDD + Epilepsy	Suspected vitamin-B6-dependent epilepsy, Generalized-onset epileptic spasm, Atypical absence seizure, EEG abnormality
LRFN4	NM_024036.5:c.473G>C,p.(Arg158Pro)	NM_024036.5:c.853C>T,p.(Arg285Cys)	comphet	4,9	comphet	maternal&paternal	2	NDD	schwere Entwicklungsverzögerung, spastische Cerebralparese, Dystrophie, MRT unauf-fällig, EEG auffällig
PACS1	NM_018026.4:c.445-17_445-7delp.?		de novo	6,6	het	de novo	3	NDD + Epilepsy	Suspected vitamin-B6-dependent epilepsy, Generalized-onset epileptic spasm, Atypical absence seizure, EEG abnormality
BTAF1	NM_003972.3:c.2662G>Ap.(Glu888Lys)		het	5,0	het	unknown		NDD + Epilepsy	(+) Global developmental delay,(+) Absent speech,(+) Seizure,(+) Intellectual disability
ADCY7	NM_001114.5:c.2866C>Tp.(Arg956Trp)		de novo	6,6	het	de novo	3	Neuro	Multifocal cerebral white matter abnormalities, Leukoencephalopathy, Migraine, Abnormal cerebellum morphology, Gait disturbance, Gait imbalance
TTLL4	NM_014640.5:c.2401C>Gp.(Leu801Val)	NM_014640.5:c.2692G>Ap.(Glu898Lys)	comphet	4,1	comphet	maternal&paternal	3	Neuro	Multifocal cerebral white matter abnormalities, Leukoencephalopathy, Migraine, Abnormal cerebellum morphology, Gait disturbance, Gait imbalance
FBN3	NM_032447.5:c.6184G>Ap.(Ala2062Thr)	NM_032447.5:c.4370A>Gp.(Asn1457Ser)	comphet	3,5	comphet	maternal&paternal	3	Neuro	Multifocal cerebral white matter abnormalities, Leukoencephalopathy, Migraine, Abnormal cerebellum morphology, Gait disturbance, Gait imbalance
HIRA	NM_003325.4:c.194A>Gp.(Gln65Arg)		het	C	het	maternal	2	Malformations	Non-midline cleft lip and palate
RGMB	NM_001012761.3:c.863C>Tp.(Thr288Ile)		het	C	het	maternal	2	Malformations	Non-midline cleft lip and palate
STARD8	NM_001142503.2:c.2248C>Ap.(Leu750Ile)		hemi	4,0	hemi	maternal	1	NDD + Epilepsy	EEG with burst suppression, Epileptic encephalopathy, Global developmental delay, Intellectual disability, Seizure
KDR	NM_002253.3:c.3161_3162insAAp.(Tyr1054*)		het	B	het	unknown	1	Cardio	Abnormal aortic morphology, Abdominal aortic aneurysm, Descending thoracic aorta aneurysm, Cerebral arterial thrombosis
FAM199X	NM_207318.4:c.932T>Gp.(Met311Arg)		de novo	A	hemi	de novo	2	Connective Tissue	Recurrent fractures, Patellar dislocation, Recurrent infections, Migraine, Asthma
LIMD1	NM_014240.3:c.1669C>Tp.(His557Tyr)	NM_014240.3:c.1532C>Tp.(Ala511Val)	comphet	A	comphet	maternal&paternal	2	Connective Tissue	Recurrent fractures, Patellar dislocation, Recurrent infections, Migraine, Asthma
FBXW7	NM_033632.3:c.23_24delp.(Val8Glyfs*14)		het	B	het	unknown	1	Other	(+) Brain neoplasm,(+) Ewing sarcoma
ATR	NM_001184.4:c.2419G>Ap.(Gly807Arg)		homo	8,8	homo	maternal&paternal	3	NDD + Epilepsy	Global developmental delay, Microcephaly, Seizures

CDK12	NM_016507.4:c.4237C>Tp.(His1413Tyr)		homo	7,2	homo	maternal&paternal	3	NDD + Epilepsy	Global developmental delay, Microcephaly, Seizures
SLC18B1	NM_052831.3:c.821G>Tp.(Gly274Val)	NM_052831.3:c.654T>Ap.(Asn218Lys)	comphet	3,7	comphet	maternal&paternal	3	NDD + Epilepsy	Global developmental delay, Microcephaly, Seizures
ZFYVE9	NM_004799.3:c.3220C>Ap.(Leu1074Met)	NM_004799.3:c.4124A>Tp.(Tyr1375Phe)	comphet	5,3	comphet	maternal&paternal	2	NDD + Epilepsy	Neonatal hypoglycemia, Seizure, Global developmental delay
LANCL3	NM_001170331.2:c.1037G>Ap.(Ser346Asn)		hemi	3,2	hemi	maternal	2	NDD + Epilepsy	Neonatal hypoglycemia, Seizure, Global developmental delay
LOXL4	NM_032211.6:c.396C>Ap.(Cys132*)		het	C	het	unknown	1	Growth, Skeletal	(+) Small for gestational age,(+) Mild short stature,(+) Attention deficit hyperactivity disorder,(+) Delayed skeletal maturation,(+) Intrauterine growth retardation,(+) Mild intrauterine growth retardation
NKTR	NM_005385.4:c.3076delp.(Glu1026Argfs*26)		de novo	10,4	het	de novo	1	NDD + Epilepsy	Myoclonic spasms, Seizure, EEG abnormality
DPYSL2	ENST00000311151.5:c.1544C>T p.Pro515Leu		ad_inherited	7,2	ad_inherited	unknown	2	NDD	Cognitive impairment, Global developmental delay, Tall stature, Obesity
DGCR2	ENST00000263196.7:c.998T>C p.Leu333Pro		het	4,6	het	unknown	2	NDD	Cognitive impairment, Global developmental delay, Tall stature, Obesity
KIF5B	NM_004521.3:c.135_136dupp.(Tyr46Phefs*67)		het	B	het	unknown	1	Malformations	Macrodactyly, Upper limb asymmetry, Hemihypertrophy of upper limb, Hyperextensible thumb
NRCAM	NM_001193582.1:c.3362C>Gp.(Pro1121Arg)		het	6,3	het	unknown	1	NDD + Epilepsy	Hypospadias, Microcephaly, Atypical absence seizure, Bilateral tonic-clonic seizure, Intellectual disability, Premature birth, Patent ductus arteriosus, Hearing impairment
PSMB10	NM_002801.4:c.56+1G>Ap.?		homo	8,5	homo	unknown	1	NDD	(+) Global developmental delay,(+) Intellectual disability, borderline,(+) Intellectual disability, mild,(+) Short stature,(+) Microcephaly,(+) Bird-like facies
TOPAZ1	NM_001145030.1:c.481A>Tp.(Ser161Cys)		de novo	4,6	het	de novo	1	NDD + Epilepsy	Focal-onset seizure, Focal sensory seizure
ARHGEF38	2729.2:c.1363_1365delACGinsGCAp.(Thr455Ala)	NM_001242729.2:c.2122G>Ap.(Asp708Asn)	comphet	C	comphet	maternal&paternal	1	Metabolism	Diabetes insipidus, Central diabetes insipidus, Panhypopituitarism, Short stature, Proportionate short stature
ATP8B4	NM_024837.3:c.2698-2A>Gp.?		de novo	5,2	het	de novo	2	NDD + Epilepsy	mild global developmental delay, febrile seizure (within the age range of 3 months to 6
MYO5B	NM_001080467.2:c.1624C>Tp.(Arg542Cys)		de novo	6,3	het	de novo	2	NDD + Epilepsy	mild global developmental delay, febrile seizure (within the age range of 3 months to 6
PTPRT	NM_133170.4:c.3039+1G>Ap.?		het	B	het	unknown	2	Cardio	(+) Cerebral vasculitis,(+) Ischemic stroke,(+) Moyamoya disease,(+) Leukoencephalopathy
XPOT	NM_007235.6:c.1516_1517delp.(Val506Cysfs*2)		het	7,7	het	unknown	2	Neuro	(+) Cerebral vasculitis,(+) Ischemic stroke,(+) Moyamoya disease,(+) Leukoencephalopathy
TMEM35B	NM_001195156.1:c.289+2delp.?		homo	A	homo	unknown		Other	+) Elevated serum alanine aminotransferase,(+) Elevated serum aspartate aminotransferase,(+) Abnormality of the liver,(+) Splenomegaly,(-) Wilson disease,(-) Niemann-Pick disease type D
HSPH1	NM_006644.4:c.515delp.(Asn172Metfs*3)		het	6,5	het	unknown	2	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Stereotypical hand wringing,(+) Muscular hypotonia
ZBTB21	NM_001098402.2:c.2088delp.(Lys696Asnfs*5)		het	6,1	het	unknown	2	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Stereotypical hand wringing,(+) Muscular hypotonia
SVEP1	NM_153366.4:c.6371T>Cp.(Ile2124Thr)		homo	5,5	homo	maternal&paternal	3	NDD + Epilepsy	atypic absence seizure, strartle-induced seizure, attention deficit hyperactivity disorder, seizure
ALS2CL	NM_147129.5:c.1109+5G>Ap.?		de novo	5,0	het	de novo	3	NDD + Epilepsy	atypic absence seizure, strartle-induced seizure, attention deficit hyperactivity disorder, seizure
CENPI	NM_006733.3:c.652C>Tp.(Arg218Cys)		hemi	5,8	hemi	maternal	3	NDD + Epilepsy	atypic absence seizure, strartle-induced seizure, attention deficit hyperactivity disorder, seizure
FAT3	NM_001008781.2:c.763C>Gp.(His255Asp)	NM_001008781.2:c.11140A>Gp.(Lys3714Glu)	comphet	5,4	comphet	maternal&paternal		NDD + Epilepsy	Atypical absence seizure, Myoclonic seizure, Epileptic encephalopathy, Myoclonus, EEG abnormality, Hyperammonemia, Abnormal vitamin B12 level, normal development
USP34	NM_014709.4:c.7561G>Cp.(Val2521Leu)	NM_014709.4:c.4229C>Tp.(Ala1410Val)	comphet	5,5	comphet	maternal&paternal		NDD	(+) Intellectual disability,(+) Hyperactivity,(+) Autistic behavior
ANKDD1A	NM_182703.5:c.1470G>Cp.(Arg490Ser)		de novo	5,4	het	de novo	1	NDD	(+) Delayed speech and language development,(+) Diminished ability to concentrate,(+) Cognitive impairment,(+) Hearing impairment
KLHL29	NM_052920.2:c.797C>Tp.(Pro266Leu)		de novo	4,1	het	de novo		Neuro	Behavioral abnormality, Frontotemporal dementia
ACTR1A	NM_005736.3:c.715G>Cp.(Ala239Pro)		het	4,6	het	unknown		NDD + Epilepsy	Generalized-onset motor seizure, Spastic tetraplegia, Intellectual disability, severe, Cataract, Pes planus
ZCCHC14	NM_015144.2:c.52C>Tp.(Gln18*)		de novo	A	het	de novo	1	NDD	motor delay, proximal muscle weakness, makrozephalia, epicanthus med., frontal blossing
SEZ6L2	NM_001243332.1:c.910A>Gp.(Thr304Ala)		het	5,4	het	maternal	3	NDD	Autism (Asperger), Autistic behavior, Depressivity, Macrocephaly
POU2F1	NM_002697.4:c.318G>Cp.(Gln106His)		het	3,8	het	paternal	3	NDD	Autism (Asperger), Autistic behavior, Depressivity, Macrocephaly
SEMA4C	017789.4:c.2077_2078delGAinsTTp.(Glu693Leu)	NM_017789.4:c.517+3G>Ap.?	comphet	4,4	comphet	maternal&paternal	1	NDD + Epilepsy	At the age of 7-8 months tonic stiffnesses for a few seconds every few weeks, later on bigger seizures, MRI without findings, no motor delay, increased levels of serum lactate, glutaric aciduria
GLRA2	NM_002063.4:c.1334G>Ap.(Arg445Gln)		hemi	7,5	hemi	maternal	1	NDD + Epilepsy	(+) Tonic seizure,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Intellectual disability,(+) Global developmental delay,(+) Cognitive impairment
POU3F2	NM_005604.4:c.664C>Tp.(Pro222Ser)		het	5,4	het	paternal		Neuro	Leukodystrophy, Leukoencephalopathy, Attention deficit hyperactivity disorder, Neurological speech impairment, Neonatal asphyxia, Gait disturbance
MAST3	NM_015016.2:c.3367C>Tp.(Arg1123*)		het	5,5	het	unknown		NDD + Epilepsy	Abnormal morphology of the limbic system,Seizure, Focal-onset seizure, Focal impaired awareness motor seizure, Bilateral tonic-clonic seizure with focal onset, Global developmental delay, Mild global developmental delay, Intellectual disability, Intellectual disability, mild, EEG with focal slow activity
PHLPP1	NM_194449.3:c.3756-2A>Gp.?		de novo	10,2	het	de novo	3	NDD + Epilepsy	therapy-resistant epilepsy
SRRM4	NM_194286.3:c.1295C>Tp.(Ser432Phe)	NM_194286.3:c.1172G>Ap.(Arg391His)	comphet	5,5	comphet	maternal&paternal	3	NDD + Epilepsy	therapy-resistant epilepsy
CANX	NM_001024649.1:c.143A>Tp.(Asp48Val)	NM_001024649.1:c.1102G>Ap.(Val368Ile)	comphet	7,4	comphet	maternal&paternal	3	NDD + Epilepsy	therapy-resistant epilepsy

H2AC8	NM_021052.2:c.107G>Ap.(Arg36His)		de novo	4,5	het	de novo	1	NDD	(+) Arachnoid cyst,(+) Headache,(+) Hallucinations,(+) Visual hallucinations,(+) Auditory hallucinations,(+) Delayed speech and language development,(+) Global developmental delay,(+) Intellectual disability,(+) Obesity
TMEM61	NM_182532.2:c.101G>Cp.(Cys34Ser)	NM_182532.2:c.583G>Ap.(Ala195Thr)	comphet	C	comphet	maternal&paternal	2	Growth, skeletal	Hypoterlorism, Trigonocephaly
TRPC5	NM_012471.2:c.280G>Ap.(Val94Met)		hemi	7,2	hemi	maternal	2	NDD	(+) Global developmental delay,(+) Hyperactivity,(+) Delayed speech and language development,(+) Hypertelorism,(+) Depressed nasal ridge,(+) Low-set ears,(+) Muscular hypotonia, lateral fallende Lidachsen
HIVEP1	NM_002114.3:c.4588T>Cp.(Ser1530Pro)	NM_002114.3:c.1916T>Cp.(Val639Ala)	comphet	3,8	comphet	maternal&paternal	2	NDD	(+) Global developmental delay,(+) Hyperactivity,(+) Delayed speech and language development,(+) Hypertelorism,(+) Depressed nasal ridge,(+) Low-set ears,(+) Muscular hypotonia, lateral fallende Lidachsen
ZNF384	NM_001135734.2:c.459delp.(Gly154Alafs*15)		de novo	9,8	het	de novo	2	NDD	(+) Global developmental delay,(+) Scotoma,(+) Intellectual disability, mild,(+) Intellectual disability, borderline,(+) Myopia,(+) Depressivity,(+) Anxiety,(+) Motor delay,(+) Retinal atrophy
SLC25A6	NM_001636.3:c.239G>Ap.(Arg80His)		de novo	7,2	het	de novo	2	NDD	(+) Global developmental delay,(+) Scotoma,(+) Intellectual disability, mild,(+) Intellectual disability, borderline,(+) Myopia,(+) Depressivity,(+) Anxiety,(+) Motor delay,(+) Retinal atrophy
NIN	NM_020921.3:c.4760A>Cp.(Gln1587Pro)	NM_020921.3:c.446C>Tp.(Thr149Met)	comphet	C	comphet	maternal&paternal	2	Growth, skeletal	Hypoterlorism, Trigonocephaly
ZDHHC2	NM_016353.5:c.47_52delp.(Arg16_Val17del)		de novo	5,2	het	de novo	2	NDD + Epilepsy	(+) Myoclonic seizure,(+) EEG with spike-wave complexes, suspected focal cortical dysplasia frontal right
KALRN	NM_001024660.4:c.3534G>Tp.(Arg1178Ser)	NM_001024660.4:c.5176+21733A>Gp.(=)	comphet	7,0	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Myoclonic seizure,(+) EEG with spike-wave complexes, suspected focal cortical dysplasia frontal right
TRHDE	3381.2:c.1050_1052delTGTinsGGGp.(Val351Gly)		de novo	B	het	de novo	1	Growth, skeletal	+) Arthrogryposis multiplex congenita,(+) Plagiocephaly,(+) Congenital finger flexion contractures,(+) Wrist flexion contracture,(+) Elbow flexion contracture,(+) Shoulder flexion contracture,(+) Adducted thumb,(+) Respiratory failure
RFX3	NM_001282116.1:c.115C>Tp.(Gln39*)		de novo	9,8	het	de novo	1	NDD	(+) Global developmental delay,(+) Poor coordination,(+) Large for gestational age,(+) Overgrowth,(+) Tall stature,(+) Ataxia,(+) Muscular hypotonia,(+) Behavioral abnormality
PTPRS	NM_002850.3:c.4810G>Ap.(Ala1604Thr)	NM_002850.3:c.4453G>Ap.(Ala1485Thr)	comphet	5,8	comphet	maternal&paternal	1	NDD	(+) Short stature,(+) Global developmental delay,(+) Intellectual disability,(+) Microcephaly
ABCB5	)1163941.1:c.2867_2867+1delp.(Ile956Lysfs*43)		de novo	7,7	het	de novo	1	NDD	(+) Mild global developmental delay,(+) Muscular hypotonia
RASA2	NM_006506.3:c.1591-2A>Gp.?		de novo	9,7	het	de novo	1	NDD	(+) Periventricular leukomalacia,(+) Global developmental delay,(+) Cerebral palsy,(+) Elevated hepatic transaminase,(+) Muscular hypotonia,(+) Small for gestational age
GRAMD1C	NM_017577.4:c.168C>Ap.(Ser56Arg)	NM_017577.4:c.557A>Gp.(Glu186Gly)	comphet	3,7	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Complex febrile seizure,(+) Simple febrile seizure,(+) Seizure,(-) Motor delay,(-) Intellectual disability
STARD9	NM_020759.2:c.4693A>Gp.(Ser1565Gly)	NM_020759.2:c.5795A>Gp.(Asn1932Ser)	comphet	3,7	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Complex febrile seizure,(+) Simple febrile seizure,(+) Seizure,(-) Motor delay,(-) Intellectual disability
NLRP5	M_153447.4:c.1846_1849delp.(Lys616Glyfs*17)		homo	8,0	homo	maternal&paternal	2	NDD + Epilepsy	(+) Dravet syndrome,(+) Seizure,(+) Myoclonic seizure,(+) Myoclonic absence seizure,(+) Global developmental delay,(+) Intellectual disability
CCDC136	NM_022742.4:c.1018C>Tp.(Arg340Trp)	NM_022742.4:c.1079G>Ap.(Ser360Asn)	comphet	4,9	comphet	maternal&paternal	1	NDD + Epilepsy	(+) Intellectual disability,(+) Arthrogryposis multiplex congenita,(+) Polymicrogyria,(+)
MDN1	NM_014611.3:c.11732G>Cp.(Ser3911Thr)		de novo	7,4	het	de novo	1	NDD	Global developmental delay, Delayed gross motor development, Macrocephaly, Patent foramen ovale
SUPV3L1	NM_003171.4:c.1931G>Ap.(Arg644Gln)	NM_003171.4:c.2358C>Gp.(Asp786Glu)	comphet	5,6	comphet	maternal&paternal		NDD + Epilepsy	(+) Global developmental delay,(+) Focal-onset seizure,(+) Abnormality of the nasal alae,(+) Poor eye contact
RYR2	NM_001035.3:c.6202C>Tp.(Arg2068*)		de novo	11,5	het	de novo	2	NDD + Epilepsy	(+) Dravet syndrome,(+) Seizure,(+) Myoclonic seizure,(+) Myoclonic absence seizure,(+) Global developmental delay,(+) Intellectual disability
RHBDL1	NM_001318733.1:c.1127C>Ap.(Ala376Glu)		de novo	5,6	het	de novo	1	NDD + Epilepsy	Focal-onset seizure, Seizure, Encephalopathy, Focal cortical dysplasia
ATP6AP2	NM_005765.3:c.858G>Ap.(Ala286=)		de novo	8,0	het	de novo		NDD	(+) Moderate global developmental delay,(+) Muscular hypotonia,(+) Dysgenesis of the hippocampus,(+) Aggressive behavior,(+) Impulsivity,(+) Low frustration tolerance,(+) Pes planus,(+) Synophrys,(-) Seizure,(-) Ataxia
DNAH3	NM_017539.2:c.7420A>Tp.(Lys2474*)	NM_017539.2:c.5287G>Ap.(Val1763Met)	comphet	5,8	comphet	maternal&paternal	1	NDD	(+) Global developmental delay,(+) Delayed speech and language development,(+) Autistic behavior,(+) Hearing impairment,(+) Developmental regression
PCDH11X	NM_032968.4:c.1688A>Gp.(Gln563Arg)		hemi	5,9	hemi	maternal		NDD + Epilepsy	(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Short attention span,(+) Specific learning disability,(+) Generalized non-motor (absence) seizure,(+) Headache,(+) Recurrent infections
PNCK	NM_001135740.1:c.643C>Gp.(Leu215Val)		hemi	4,6	hemi	maternal	1	NDD	(+) Neurodevelopmental delay,(+) Mild expressive language delay,(+) Morphological central nervous system abnormality,(+) Hydromyelia,(+) Achilles tendon contracture,(+) Testicular torsion,(+) Syringomyelia,(+) Sleep disturbance,(+) Limited hip extension,(+) Spastic paraplegia,(+) Motor delay
ZBTB45	NM_001316978.2:c.976G>Ap.(Gly326Arg)		homo	4,0	homo	maternal&paternal	2	NDD + Epilepsy	(+) Focal-onset seizure,(+) Brain imaging abnormality
NOMO1	NM_014287.4:c.2173G>Ap.(Gly725Ser)		homo	4,4	homo	maternal&paternal	2	NDD + Epilepsy	(+) Focal-onset seizure,(+) Brain imaging abnormality
PLXNA3	NM_017514.5:c.1015C>Gp.(Leu339Val)		hemi	6,1	hemi	maternal	2	NDD + Epilepsy	(+) Infantile encephalopathy,(+) Microcephaly,(+) Short stature,(+) Muscular hypotonia,(+) Micropenis,(+) Global developmental delay,(+) Abnormal facial shape,(+) Cerebral ischemia,(+) Focal-onset seizure,(+) Epicanthus,(+) Decreased body weight,(+) Oxycephaly,(+) Hypospadias,(+) Cryptorchidism

SMYD5	NM_006062.3:c.100A>Gp.(Lys34Glu)	NM_006062.3:c.833G>Ap.(Arg278His)	comphet	4,2	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Infantile encephalopathy,(+) Microcephaly,(+) Short stature,(+) Muscular hypotonia,(+) Micropenis,(+) Global developmental delay,(+) Abnormal facial shape,(+) Cerebral ischemia,(+) Focal-onset seizure,(+) Epicanthus,(+) Decreased body weight,(+) Oxycephaly,(+) Hypospadias,(+) Cryptorchidism
GIGYF1	NM_022574.4:c.1778A>Tp.(Asp593Val)		de novo	B	het	de novo	2	Growth, skeletal	(+) Cleft soft palate,(+) Cleft hard palate
MAP3K6	NM_004672.4:c.3789-5C>Tp.?	NM_004672.4:c.1733T>Ap.(Val578Asp)	comphet	C	comphet	maternal&paternal	2	Growth, skeletal	(+) Cleft soft palate,(+) Cleft hard palate
TMEM143	NM_018273.3:c.1022T>Cp.(Met341Thr)		de novo	4,4	het	de novo	3	NDD + Epilepsy	(+) Focal tonic seizure,(+) EEG with focal sharp waves,(+) Nocturnal seizures,(-) Brain imaging abnormality
FAM214B	NM_001317991.1:c.1012C>Gp.(Pro338Ala)		homo	5,6	homo	maternal&paternal	3	NDD + Epilepsy	(+) Focal tonic seizure,(+) EEG with focal sharp waves,(+) Nocturnal seizures,(-) Brain imaging abnormality
STX4	NM_004604.4:c.118_120delp.(Glu40del)		homo	5,6	homo	maternal&paternal	3	NDD + Epilepsy	(+) Focal tonic seizure,(+) EEG with focal sharp waves,(+) Nocturnal seizures,(-) Brain imaging abnormality
SEMA5A	NM_003966.3:c.2123C>Tp.(Thr708Met)		homo	8,3	homo	maternal&paternal	6	NDD	(+) Severe global developmental delay,(+) Intellectual disability,(+) Feeding difficulties,(+) Muscular hypotonia
ADGRD2	NM_001161808.1:c.1068C>Ap.(Cys356*)		de novo	5,0	het	de novo	1	NDD	(+) Global developmental delay,(+) Motor delay,(+) Neonatal asphyxia,(+) Neonatal seizure,(+) Hypertonia,(+) Dysphagia,(+) Tongue fasciculations,(+) Microcephaly,(+) Infantile encephalopathy
ATP6V0A1	NM_001130020.1:c.2222G>Ap.(Arg741Gln)		het	5,3	het	unknown	1	NDD + Epilepsy	(+) Seizure,(+) Large for gestational age,(+) Microcephaly,(+) Global developmental delay,(+) Muscular hypotonia
AHNAK	NM_001620.2:c.11743G>Ap.(Asp3915Asn)		homo	6,4	homo	maternal&paternal	3	NDD	(+) Global developmental delay,(+) Motor delay,(+) Cleft palate,(+) Cleft lip,(+) Cerebellar hypoplasia
DHRS3	NM_004753.6:c.730G>Cp.(Glu244Gln)		homo	5,6	homo	maternal&paternal	3	NDD	(+) Global developmental delay,(+) Motor delay,(+) Cleft palate,(+) Cleft lip,(+) Cerebellar hypoplasia
TRPM2	NM_003307.3:c.2392G>Tp.(Val798Phe)		homo	5,6	homo	maternal&paternal	3	NDD	(+) Global developmental delay,(+) Motor delay,(+) Cleft palate,(+) Cleft lip,(+) Cerebellar hypoplasia
MAGEA10	NM_001011543.2:c.229G>Tp.(Asp77Tyr)		hemi	C	hemi	maternal	2	Growth, skeletal	(+) Trigonocephaly
OAS3	NM_006187.3:c.101G>Ap.(Gly34Asp)	NM_006187.3:c.1443C>Ap.(Asn481Lys)	comphet	C	comphet	maternal&paternal	1	Growth, skeletal	(+) Trigonocephaly
POLR3E	NM_018119.3:c.437A>Gp.(Asp146Gly)		de novo	A	het	de novo	2	Metabolism	(+) Low levels of vitamin A,(+) Low levels of vitamin D,(+) Leukopenia,(+) Thrombocytopenia,(+) Hepatosplenomegaly,(+) Portal vein thrombosis
TENM2	NM_001122679.1:c.3262A>Tp.(Ile1088Phe)	NM_001122679.1:c.6169C>Tp.(Arg2057Trp)	comphet	C	comphet	maternal&paternal	2	Metabolism	(+) Low levels of vitamin A,(+) Low levels of vitamin D,(+) Leukopenia,(+) Thrombocytopenia,(+) Hepatosplenomegaly,(+) Portal vein thrombosis
ZFHX3	NM_006885.3:c.5449G>Tp.(Val1817Leu)	NM_006885.3:c.2321C>Tp.(Ala774Val)	comphet	5,4	comphet	maternal&paternal	1	NDD + Epilepsy	
PTPRH	NM_002842.4:c.1324G>Ap.(Ala442Thr)	NM_002842.4:c.683G>Ap.(Trp228*)	comphet	B	comphet	maternal&paternal	1	Eye	(+) Optic neuropathy,(+) Amblyopia,(+) Nystagmus,(+) Strabismus,(+) Mixed astigmatism,(+) Protanomaly
BTBD18	ENST00000422652.1:c.1236dup, p.Arg413Ter		de novo	A	het	de novo	2	Malformations	Cleft palate, renal agnesia left
PLEKHB2	ENST00000409158.1:c.83C>T p.Ser28Leu		homo	C	homo	maternal&paternal	2	Malformations	(+) Cleft lip,(+) Cleft palate,(+) Unilateral renal agenesis
HDAC6	ENST00000334136.5:c.3248G>A p.Gly1083Asp		hemi	C	hemi	maternal	2	Growth, skeletal	Trigonocephaly, Abnormality of calvarial morphology
ZBTB12	ENST00000375527.2:c.583G>A p.Glu195Lys		de novo	5,0	het	de novo	2	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Intellectual disability
ADI1	ENST00000327435.6:c.214G>A p.Asp72Asn	ENST00000327435.6:c.166C>T p.Arg56Ter	comphet	4,9	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Intellectual disability
PPP2R5C	ENST00000422945.2:c.1341A>T p.Lys447Asn		het	5,5	het	unknown	1	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Hemimegalencephaly
FAM171A2	ST00000293443.7:c.1170del p.Glu391ArgfsTer67		homo	8,2	homo	maternal&paternal	2	NDD	(+) Intellectual disability,(+) Microcephaly
JMJD1C	ENST00000399262.2:c.1372G>A p.Glu458Lys		homo	7,6	homo	maternal&paternal	2	NDD	(+) Intellectual disability,(+) Microcephaly
RC3H2	ENST00000373670.1:c.382C>A p.Arg128Ser		ad_inherited	4,1	ad_inherited	unknown		NDD + Epilepsy	(+) Focal tonic seizure,(+) Focal myoclonic seizure,(+) Atypical absence seizure,(+) Intellectual disability, mild
PHF20	ENST00000374012.3:c.1300A>G p.Lys434Glu		het	3,6	het	unknown	2	NDD	(+) Microcephaly,(+) Plagiocephaly,(+) Ventricular septal defect,(+) Short palpebral fissure,(+) Smooth philtrum,(+) Thin upper lip vermilion,(+) Short stature,(+) Absent speech,(+) Motor delay
FAT3	ENST00000298047.6:c.5027A>G p.Tyr1676Cys	ENST00000298047.6:c.10393A>G p.Ile3465Val	comphet?	4,7	comphet?	unknown	2	NDD	(+) Microcephaly,(+) Plagiocephaly,(+) Ventricular septal defect,(+) Short palpebral fissure,(+) Smooth philtrum,(+) Thin upper lip vermilion,(+) Short stature,(+) Absent speech,(+) Motor delay
NEFM	ENST00000221166.5:c.446C>G p.Ala149Gly		de novo	7,1	het	de novo	1	NDD + Epilepsy	(+) Global developmental delay,(+) Intellectual disability,(+) Behavioral abnormality,(+) Short stature,(+) Focal motor seizure,(+) Focal-onset seizure,(+) Bilateral tonic-clonic seizure with focal onset
GABRE	ENST00000370328.3:c.1148A>G p.Asn383Ser		de novo	4,9	het	de novo	2	Epilepsy	Seizure, abnormality of metabolism, epileptic encephalopathy
AWAT1	ENST00000374521.3:c.273C>G p.Asp91Glu		hemi	3,2	hemi	maternal	3	Epilepsy	intellecutal disability, focal onset seizur, cortical dysplasia, brain atrophy
FAM171A1	ENST00000378116.4:c.364T>C p.Ser122Pro	ENST00000378116.4:c.1418A>G p.Glu473Gly	comphet	3,4	comphet	maternal&paternal	3	Epilepsy	intellecutal disability, focal onset seizur, cortical dysplasia, brain atrophy
ZNRF4	ENST00000222033.4:c.1135C>G p.His379Asp		de novo	4,5	het	de novo	3	Epilepsy	intellecutal disability, focal onset seizur, cortical dysplasia, brain atrophy
DCBLD1	ENST00000296955.8:c.1178G>A p.Arg393Gln		homo	4,8	homo	maternal&paternal	6	NDD	Severe global developmental delay, Feeding difficulties, Muscular hypotonia
NCOA7	ENST00000368357.3:c.1396G>A p.Ala466Thr		homo	3,3	homo	maternal&paternal	6	NDD	Severe global developmental delay, Feeding difficulties, Muscular hypotonia
SLC27A4	ENST00000300456.4:c.1462+5_1462+9del None		homo	4,9	homo	maternal&paternal	6	NDD	Severe global developmental delay, Feeding difficulties, Muscular hypotonia
MTUS2	ENST00000431530.3:c.2752C>T p.Arg918Trp		homo	4,3	homo	maternal&paternal	6	NDD	Severe global developmental delay, Feeding difficulties, Muscular hypotonia
STXBP4	ENST00000376352.2:c.866G>C p.Cys289Ser		homo	3,9	homo	maternal&paternal	6	NDD	Severe global developmental delay, Feeding difficulties, Muscular hypotonia
ANK2	ENST00000357077.4:c.10768G>T p.Glu3590Ter		de novo	11,9	het	de novo	1	Epilepsy	Focal myoclonic seizure

GRIPAP1	ENST00000376441.1:c.1007A>G p.Asn336Ser		hemi	5,9	hemi	maternal	2	NDD	(+) Intellectual disability,(+) Global developmental delay,(+) Abnormality of movement,(+) Dystonia,(+) Spasticity
H1FOO	ENST00000324382.2:c.863C>T p.Ala288Val		de novo	4,2	het	de novo	2	NDD	(+) Intellectual disability,(+) Global developmental delay,(+) Abnormality of movement,(+) Dystonia,(+) Spasticity
NKPD1	ENST00000317951.4:c.1076A>G p.Tyr359Cys		de novo	5,4	het	de novo	1	NDD	Caudal regression syndrome, Currarino Triad, Global developmental delay
HTR4	ENST00000360693.3:c.721C>T p.Gln241Ter		het	6,8	het		2	NDD + Epilepsy	Intellectual disability,(+) Atypical absence seizure,(+) Generalized tonic seizure,(+) Generalized-onset epileptic spasm,(+) Myoclonus,(+) Generalized atonic seizure,(+) Bilateral tonic-clonic seizure with generalized onset
NSD3	ENST00000317025.8:c.3725G>A p.Arg1242Gln		het	5,7	het		2	NDD + Epilepsy	Intellectual disability,(+) Atypical absence seizure,(+) Generalized tonic seizure,(+) Generalized-onset epileptic spasm,(+) Myoclonus,(+) Generalized atonic seizure,(+) Bilateral tonic-clonic seizure with generalized onset
ARHGEF2	ENST00000361247.4:c.355C>T p.Arg119Trp	ENST00000361247.4:c.415C>T p.Arg139Cys	comphet	7,1	comphet	maternal&paternal	1	NDD + muscle	(+) Muscular hypotonia, (+) Increased serum lactate, (+) Motor delay, (+) Strabismus, (+) Reduced visual acuity, (+) Visual impairment
SHANK1	ENST00000293441.1:c.4932C>G p.Asp1644Glu		ad_inherited	7,1	ad_inherited			NDD + Epilepsy	Typical absence seizure,(+) Myoclonic seizure,(+) Bilateral tonic-clonic seizure,(+) Intellectual disability, mild,(+) Intellectual disability, borderline,(+) EEG with spike-wave complexes (2.5-3.5 Hz)
NCKAP1	ST00000360982.2:c.3366_3369del p.Tyr1122Ter		de novo	11,7	het	de novo		NDD + Epilepsy	(+) Epicanthus,(+) Narrow face,(+) Anteverted nares,(+) High palate,(+) Global developmental delay,(+) Focal-onset seizure
AFF3	ENST00000356421.2:c.3181G>A p.Val1061Ile	ENST00000356421.2:c.3632G>A p.Arg1211Gln	comphet	4,9	comphet	maternal&paternal	1	NDD + Epilepsy	(+) Epileptic encephalopathy,(+) Agenesis of corpus callosum,(+) Abnormal cortical gyration, (+) Hypomyelination
NRXN3	ENST00000554719.1:c.2776C>T p.Arg926Cys		het	7,4	het	unknown		NDD + Epilepsy	(+) Generalized non-motor (absence) seizure,(+) Attention deficit hyperactivity disorder,(+) Talipes cavus equinovarus,(+) Global developmental delay,(+) Low-frequency hearing loss
CPSF4	ENST00000292476.5:c.655C>T p.Pro219Ser		de novo	7,0	het	de novo	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
PCDH1	ENST00000287008.3:c.3698G>A p.Arg1233His		homo	5,0	homo	maternal&paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
ADNP2	ENST00000262198.4:c.422T>G p.Ile141Ser		homo	6,0	homo	maternal&paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
PPFIBP1	NM_177444.3:c.1197+1G>A, p.?		homo	8,2	homo	maternal&paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
HTR3B	ENST00000260191.2:c.550G>A p.Asp184Asn		homo	5,3	homo	maternal&paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
ARHGEF12	ST00000397843.2:c.3460_3462del p.Asn1154del		homo	5,4	homo	maternal&paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
ASUN	ENST00000261191.7:c.341G>A p.Arg114Gln		homo	4,2	homo	maternal&paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
TNRC18	00430969.1:c.4261_4262delinsGG p.Leu1421Gly		homo	5,4	homo	maternal&paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
CDC25C	ENST00000323760.6:c.1129T>C p.Cys377Arg		homo	6,1	homo	maternal&paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
HEXIM2			homo	7,0	homo	maternal&paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
KANK1	ENST00000382303.1:c.3733G>A p.Gly1245Arg	ENST00000382303.1:c.1652G>A p.Cys551Tyr	comphet	6,3	comphet	maternal&paternal	3	NDD + Epilepsy	(+) Global developmental delay,(+) Infantile spasms,(+) Generalized-onset seizure,(+) Hearing impairment,(+) Epileptic encephalopathy
DRP2	ENST00000395209.3:c.575A>C p.Gln192Pro		hemi	4,6	hemi	maternal	3	NDD + Epilepsy	(+) Global developmental delay,(+) Infantile spasms,(+) Generalized-onset seizure,(+) Hearing impairment,(+) Epileptic encephalopathy
RNF113A	00000371442.2:c.265_270del p.Glu89_Glu90del		x_linked	5,0	x_linked	maternal	3	NDD + Epilepsy	(+) Global developmental delay,(+) Infantile spasms,(+) Generalized-onset seizure,(+) Hearing impairment,(+) Epileptic encephalopathy
TSSC1	ENST00000382125.4:c.514G>A p.Val172Met		de novo	5,2	het	de novo	3	Muscle	Motor delay, Muscular hypotonia, Skeletal muscle atrophy
NKTR	00232978.8:c.2511_2514del p.Gln838LysfsTer23		de novo	10,2	het	de novo	3	Muscle	Motor delay, Muscular hypotonia, Skeletal muscle atrophy
DRP2	ENST00000395209.3:c.2438C>T p.Ala813Val		hemi	4,9	hemi	maternal	3	Muscle	Motor delay, Muscular hypotonia, Skeletal muscle atrophy
KCNRG	ST00000312942.1:c.394dup, p.Thr132AsnfsTer3		homo	8,0	homo	maternal&paternal	2	NDD	(+) Global developmental delay,(+) Cognitive impairment,(+) Autism,(+) Autistic behavior
ERVMER34-1	ENST00000443173.1:c.936A>T p.Lys312Asn		de novo	B	het	de novo		Other	(+) Intrauterine growth retardation,(+) Oligohydramnios
CELSR3	ENST00000164024.4:c.5751+1G>C None		homo	11,2	homo	maternal&paternal		NDD + Epilepsy	(+) Focal-onset seizure,(+) Generalized-onset seizure,(+) Global developmental delay,(+) Dystonia,(+) Cerebral white matter agenesis,(+) Microcephaly
ITGAM	ENST00000544665.3:c.2923C>T p.Pro975Ser		de novo	6,5	het	de novo	1	NDD + Growth	Failure to thrive, Short stature, Feeding difficulties, Hepatomegaly, Atrial septal defect, Abdominal distention, Global developmental delay, Congenital microcephaly, Plagiocephaly, Dysmorphic facial features
ATG9A	0000409618.1:c.1282del p.Gln428SerfsTer13		het	6,2	het	unknown	1	Epilepsy	(+) Seizure,(+) Attention deficit hyperactivity disorder,(+) Intellectual disability, mild
ALDH3B2	ENST00000349015.3:c.505G>A p.Val169Ile	ENST00000349015.3:c.635G>A p.Arg212Gln	comphet	C	comphet	maternal&paternal	1	Cardio	Unbalanced atrioventricular canal defect, Anomalous pulmonary venous return, Congenital malformation of the great arteries, Bradycardia
NME4	ENST00000219479.2:c.1A>T p.Met1?		homo	7,78	homo	unknown		NDD + Epilepsy	Moderate intellectual disability, delayed speech and language development, absence seizure, focal impaired awareness motor seizure, bilateral tonic-clonic seizure with generalized onset, muscular hypotonia, joint laxity, abnormal facial shape, temporal lobe sclerosis right (Hippocampectomy 01/2005), hypogonadotropic hypogonadism
YWHAB	ENST00000372839.3:c.637T>C p.Tyr213His		de novo	7,2	het	de novo	1	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay
PRPF40A	ENST00000410080.1:c.84+2T>G None			6,15		unknown		Epilepsy	Generalized non-motor (absence) seizure
DNAH6	ENST00000389394.3:c.11360G>A p.Gly3787Asp			3,99		unknown		Epilepsy	(+) Generalized-onset seizure,(+) Focal motor seizure,(+) EEG abnormality,(+) Mild short stature,(+) Microcephaly,(+) Decreased glucose-6-phosphate dehydrogenase level in blood
ALS2CL	ENST00000318962.4:c.893C>T p.Ala298Val	ENST00000318962.4:c.2704G>A p.Glu902Lys	comphet	3,3	comphet	maternal&paternal	1	Epilepsy	(+) Myoclonic seizure,(+) Generalized myoclonic-tonic-clonic seizure,(+) Ataxia,(+) Suicidal ideation

WDFY4	ENST00000325239.5:c.3175+2del None			6,2		unknown		NDD	(+)Global developmental delay,(+) Delayed speech and language development,(+) Muscular hypotonia,(+) Anal atresia,(+) Perineal fistula,(+) Atrial septal defect,(+) Dextrocardia,(+) Hearing impairment,(+) Unilateral ptosis,(+) Posterior plagiocephaly,(+) Scoliosis,(+) Low-set ears,(+) Retrognathia,(+) Abnormality of the philtrum,(+) Bilateral single transverse palmar creases,(+) Abnormality of toe
GRIK3	ENST00000373091.3:c.176C>T p.Ala59Val			5,44		unknown		NDD	(+) Global developmental delay,(+) Ataxia,(+) Muscular hypotonia,(+) Macrocephaly,(+) Tall stature,(+) Obesity
CHD8	ENST00000399982.2:c.4418G>T p.Arg1473Leu			6,61		unknown		NDD + Epilepsy	(+) Tonic seizure,(+) Intellectual disability, severe,(+) Kyphoscoliosis,(+) Hyperlordosis,(+) Focal polymicrogyria,(+) Frontoparietal polymicrogyria,(+) Global brain atrophy,(+) EEG with focal epileptiform discharges,(+) Bilateral tonic-clonic seizure,(+) Absent speech
EHMT2	ENST00000375537.4:c.912_914del p.Glu323del	ENST00000375537.4:c.1509G>A p.Ala503=	comphet	6,33	comphet	maternal&paternal	1	NDD	(+) Intellectual disability,(+) Global developmental delay,(+) Microcephaly,(+) Behavioral abnormality,(+) 2-3 toe syndactyly
SAMD9L	NST00000318238.4:c.694del p.Arg232AlafsTer21		homo	A	homo	maternal&paternal	2	Immunology	(+)Decreased proportion of CD8-positive T cells,(+) Severe SARS-CoV-2 infection,(+) Failure to thrive secondary to recurrent infections,(+) Exocrine pancreatic insufficiency
TRIM24	ENST00000343526.4:c.1237T>A p.Trp413Arg		de novo	A	het	de novo	2	Immunology	(+)Decreased proportion of CD8-positive T cells,(+) Severe SARS-CoV-2 infection,(+) Failure to thrive secondary to recurrent infections,(+) Exocrine pancreatic insufficiency
FADS1	ENST00000350997.7:c.238G>A p.Asp80Asn		homo	B	homo	maternal&paternal	4	NDD	(+) Double outlet right ventricle,(+) Pulmonic stenosis,(+) Failure to thrive,(+) Frontal hirsutism,(+) Low-set ears,(+) Narrow face,(+) Hearing impairment
RCOR2	ENST00000301459.4:c.1376C>T p.Thr459Met		homo	B	homo	maternal&paternal	4	NDD	(+) Double outlet right ventricle,(+) Pulmonic stenosis,(+) Failure to thrive,(+) Frontal hirsutism,(+) Low-set ears,(+) Narrow face,(+) Hearing impairment
SRGAP1	ENST00000355086.3:c.1421A>G p.Glu474Gly	ENST00000355086.3:c.1217G>A p.Arg406His	comphet	4,8	comphet	maternal&paternal	1	Epilepsy	Generalized-onset seizure, Bilateral tonic-clonic seizure, Focal-onset seizure, EEG with spike-wave complexes
GAL3ST4	)360039.4:c.1207_1208insC p.Leu403ProfsTer10		homo	8,0	homo	maternal&paternal		NDD	(+) Profound global developmental delay,(+) Muscular hypotonia,(+) Abnormality of the Achilles tendon,(+) Abnormal foot morphology,(+) Increased lactate dehydrogenase level,(+) Increased serum lactate,(+) Delayed CNS myelination,(+) Hypoplasia of the corpus callosum,(+) Abnormal macular morphology,(-) Abnormal facial shape
PER1	ENST00000317276.4:c.694G>C p.Val232Leu	ENST00000317276.4:c.3373G>A p.Val1125Met	comphet	C	comphet	maternal&paternal	4	NDD	(+) Double outlet right ventricle,(+) Pulmonic stenosis,(+) Failure to thrive,(+) Frontal hirsutism,(+) Low-set ears,(+) Narrow face,(+) Hearing impairment
HECTD1	ENST00000399332.1:c.5140C>T p.Arg1714Cys	ENST00000399332.1:c.6725C>T p.Thr2242Met	comphet	C	comphet	maternal&paternal	4	NDD	(+) Double outlet right ventricle,(+) Pulmonic stenosis,(+) Failure to thrive,(+) Frontal hirsutism,(+) Low-set ears,(+) Narrow face,(+) Hearing impairment
TNRC18	ENST00000430969.1:c.690G>T p.Glu230Asp	ENST00000430969.1:c.5525C>T p.Ala1842Val	comphet	4,4	comphet	maternal&paternal	3	Neuro	Leukodystrophy, Leukoencephalopathy, Strabismus (normal development)
NCOR1	ENST00000268712.3:c.3360G>C p.Glu1120Asp	ENST00000268712.3:c.5240G>A p.Arg1747Gln	comphet	5,7	comphet	maternal&paternal	3	Neuro	Leukodystrophy, Leukoencephalopathy, Strabismus (normal development)
TMEM205	ENST00000354882.5:c.326G>A p.Arg109His		homo	3,8	homo	maternal&paternal	3	Neuro	Leukodystrophy, Leukoencephalopathy, Strabismus (normal development)
CROCC	ENST00000375541.5:c.5585G>A p.Arg1862Gln	ENST00000375541.5:c.736G>C p.Ala246Pro	comphet?	4,8	comphet?	unknown	4	NDD + Epilepsy	Bilateral tonic-clonic seizure with focal onset, Hypothyroidism, Hepatosplenomegaly, Intellectual disability, Global developmental delay, EEG abnormality, EEG with focal sharp waves, Cranial hyperostosis, Poor speech
KIAA1407	ENST00000295878.3:c.89A>C p.Lys30Thr	ST00000295878.3:c.1035dup, p.Lys346GlufsTer7	comphet?	4,3	comphet?	unknown	4	NDD + Epilepsy	Bilateral tonic-clonic seizure with focal onset, Hypothyroidism, Hepatosplenomegaly, Intellectual disability, Global developmental delay, EEG abnormality, EEG with focal sharp waves, Cranial hyperostosis, Poor speech
RBM19	ENST00000545145.2:c.520T>G p.Ser174Ala	ENST00000545145.2:c.1247A>G p.Glu416Gly	comphet?	4,6	comphet?	unknown	4	NDD + Epilepsy	Bilateral tonic-clonic seizure with focal onset, Hypothyroidism, Hepatosplenomegaly, Intellectual disability, Global developmental delay, EEG abnormality, EEG with focal sharp waves, Cranial hyperostosis, Poor speech
TRIM14	ENST00000341469.2:c.1104C>A p.Asp368Glu		ad_inherited	B	ad_inherited	maternal		Immunology	(+) Recurrent infections,(+) Sepsis,(+) Affected mother
SCAF8	IST00000367186.4:c.119dup, p.Leu41ProfsTer14		unknown	6,06	unknown	unknown		NDD + Epilepsy	(+) Intellectual disability, severe,(+) Severe global developmental delay,(+) Bilateral tonic-clonic seizure with focal onset,(+) Cataract,(+) Abnormality of the kidney,(+) EEG
PRKRIR	000260045.3:c.2274_2275delinsCT p.Glu759Ter		het	B	het	unknown		Muscle	Maligne Hyperthermie
TRANK1	ENST00000429976.2:c.4634A>G p.Asn1545Ser		homo	B	homo	maternal&paternal	4	Other	Precocious puberty, Tremor, Hypertrichosis, Hirsutism, Increased head circumference, Increased body weight, Acne
MAP7D1	ENST00000373151.2:c.2003A>C p.Glu668Ala		homo	B	homo	maternal&paternal	4	Other	Precocious puberty, Tremor, Hypertrichosis, Hirsutism, Increased head circumference, Increased body weight, Acne
NME6	ENST00000421967.1:c.548A>T p.His183Leu		homo	B	homo	maternal&paternal	4	Other	Precocious puberty, Tremor, Hypertrichosis, Hirsutism, Increased head circumference, Increased body weight, Acne
PHC3	ENST00000495893.2:c.959A>G p.His320Arg		homo	B	homo	maternal&paternal	4	Other	Precocious puberty, Tremor, Hypertrichosis, Hirsutism, Increased head circumference, Increased body weight, Acne
GPR124	ENST00000412232.2:c.1579C>T p.Leu527Phe		de novo	5,9	het	de novo	1	NDD + Epilepsy	Intellectual disability, moderate, Global developmental delay, Focal-onset seizure, Generalized-onset seizure, Abnormality of brain morphology in MRI , Muscle weakness of the right side of the body
TIMP1	ENST00000218388:c.224T>C p.Leu75Ser		hemi	4,33	hemi	maternal	2	NDD	Mental retardation
SEMA4B	ENST00000411539:c.1044-8C>T None	ENST00000411539:c.2320G>A p.Gly774Ser	comphet	3,78	comphet	maternal&paternal	2	NDD	Mental retardation
GOLGA2	IST00000421699:c.2414del p.Met805ArgfsTer18		het	8,8	het	unknown		NDD	Intellectual disability, Abnormal facial shape
ATP13A3	ENST00000439040.5:c.2638A>T p.(Met880Leu)		het	C	het	unknown		Growth, skeletal	(+) Mild short stature
SMARCA1	ENST00000371122:c.2402A>G p.Glu801Gly		hemi	7,67	hemi	unknown		NDD + Epilepsy	Intellectual disability, severe,(+) Severe global developmental delay,(+) EEG abnormality,(+) Generalized tonic seizure,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Status epilepticus,(+) Spastic tetraparesis,(+) Bilateral talipes equinovarus,(+) Pilomatrixoma

SPRED3	ENST00000338502:c.1210C>T p.Arg404Cys		de novo	5,4	het	de novo	2	NDD + Epilepsy	(+) Atonic seizure,(+) Generalized clonic seizure,(+) Generalized tonic seizure,(+) Intellectual disability, mild,(+) Gastroesophageal reflux,(+) Postnatal microcephaly
PIPOX	ENST00000323372.4:c.28G>T p.Ala10Ser	ENST00000323372.4:c.514G>A p.Gly172Arg	comphet	4,3	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Atonic seizure,(+) Generalized clonic seizure,(+) Generalized tonic seizure,(+) Intellectual disability, mild,(+) Gastroesophageal reflux,(+) Postnatal microcephaly
CCDC180	ENST00000375202:c.820C>T p.Arg274Ter	ENST00000375202:c.4179+5G>C None	comphet?	3,8	comphet?	unknown		NDD	Global developmental delay, Aggressive behavior
NSF	ENST00000398238:c.2218C>A p.Pro740Thr		het	6,09	het	unknown	1	NDD + Epilepsy	myoklonische Anfälle, komplexe Partialanfälle sekundärer Generalisierung, V.a. Absencen, schwere Intelligenzminderung, Entwicklungsstörung keine Kontaktaufnahme, Strabismus divergens, Nystagmus, Okulomotoriusparese, beginnende Cerebralparese, muskuläre Hypotonie, Optikusatrophie bei Netzhautdystrophie, komplexe Hirnfehlbildungen: Aphasie des Nucleus caudatus und Potamen rechts. Hypoplasie des Balkens. Polyvyrie. höhergradige Atrophie der linken Kleinhirnhemisphäre
ITPK1	0000267615:c.899_900insGA p.Gly301LysfsTer6		het	6,1	het	unknown		Epilepsy	fokale Epilepsie refraktär auf Levetiracetam und Valproat, bislang unauffällige Entwicklung
EIF5B	ENST00000289371:c.1360del p.Ile454TyrfsTer5		het	6,8	het	unknown	2	NDD + Epilepsy	(+) Intellectual disability, severe,(+) Stereotypical hand wringing,(+) Self-injurious behavior,(+) Obsessive-compulsive behavior,(+) Seizure,(+) Scoliosis
MARK2	ENST00000402010:c.1934+1G>A None		het	7,6	het	unknown	2	NDD + Epilepsy	(+) Intellectual disability, severe,(+) Stereotypical hand wringing,(+) Self-injurious behavior,(+) Obsessive-compulsive behavior,(+) Seizure,(+) Scoliosis
NRCAM	ENST00000379028:c.2738G>A p.Gly913Asp	ENST00000379028:c.2491C>A p.Pro831Thr	comphet?	7,76	comphet?	unknown		NDD + Epilepsy	(+) Intellectual disability,(+) Global developmental delay,(+) Seizure,(+) Motor delay,(+) EEG abnormality,(+) Poor coordination,(+) Delayed speech and language development,(+) Cafe-au-lait spot,(+) Autism
TBC1D7	ENST00000606214:c.728T>C p.Leu243Ser		homo	7,3	homo	maternal&paternal	2	NDD + Epilepsy	Entwicklungsverzögerung, atone Anfällle, sporadisches Lennox-Gastaut-Syndrom, Intelligenzminderung, ASD
STRAP	ENST00000419869:c.41C>T p.Thr14Met		de novo	7,01	het	de novo	2	NDD + Epilepsy	Entwicklungsverzögerung, atone Anfällle, sporadisches Lennox-Gastaut-Syndrom, Intelligenzminderung, ASD
NYAP1	NM_173564.4:c.2426T>G		de novo	6,2	het	de novo	2	NDD + Epilepsy	(+) Moderate global developmental delay,(+) Muscular hypotonia,(+) Molar tooth sign on MRI,(+) Developmental cataract,(+) Febrile seizure (within the age range of 3 months to 6 years)
ITSN1	ENST00000381318:c.3997T>C p.Cys1333Arg		het	C	het	unknown	1	Neuro	(+) Semantic dementia,(+) Frontotemporal dementia,(+) Tremor
SLITRK4	ENST00000381779:c.1282C>T p.Arg428Cys		hemi	4,83	hemi	maternal	2	NDD + Epilepsy	(+) Focal-onset seizure,(+) Dandy-Walker malformation,(+) Overgrowth,(+) Global developmental delay
MOSPD2	ENST00000380492:c.1427G>A p.Arg476His		hemi	3,9	hemi	maternal	2	NDD + Epilepsy	(+) Focal-onset seizure,(+) Dandy-Walker malformation,(+) Overgrowth,(+) Global developmental delay
GTF3A	ENST00000381140:c.55G>A p.Ala19Thr		de novo	5,35	het	de novo	2	NDD + Epilepsy	West-Syndrom, developmental delay, Neuroblastom
HCN2	0251287:c.2156_2164dup, p.Pro719_Pro721dup		het	C	het	unknown	1	Neuro	(+) Multifocal cerebral white matter abnormalities,(+) Abnormality of the periventricular white matter
NUDT21	ENST00000300291:c.187A>G p.Arg63Gly		unknown	3,9	unknown	unknown	1	NDD	(+) Global developmental delay,(+) Short stature,(+) Microcephaly,(+) Failure to thrive,(+) Short toe,(+) Abnormality of the face
AOX1	ENST00000374700:c.2024T>C p.Val675Ala	ENST00000374700:c.3478G>A p.Glu1160Lys	comphet?	3,62	comphet?	unknown	2	NDD + Epilepsy	West-Syndrom, developmental delay, Neuroblastom
EZH1	ENST00000428826:c.1691A>G p.Lys564Arg		het	C	het	maternal	3	Immunology	(+) Abnormal oral mucosa morphology,(+) Aphthous ulcer,(+) Recurrent aphthous stomatitis,(+) Abnormal blistering of the skin,(+) Oral mucosal blisters,(+) Genital blistering,(+) Arthralgia,(+) Nausea,(+) Episodic fatigue,(+) Vertigo
INTS2	ENST00000444766:c.650A>T p.Asn217Ile		het	C	het	maternal	3	Immunology	(+) Abnormal oral mucosa morphology,(+) Aphthous ulcer,(+) Recurrent aphthous stomatitis,(+) Abnormal blistering of the skin,(+) Oral mucosal blisters,(+) Genital blistering,(+) Arthralgia,(+) Nausea,(+) Episodic fatigue,(+) Vertigo
LMTK3	ENST00000270238:c.1460C>T p.Pro487Leu		het	C	het	maternal	3	Immunology	(+) Abnormal oral mucosa morphology,(+) Aphthous ulcer,(+) Recurrent aphthous stomatitis,(+) Abnormal blistering of the skin,(+) Oral mucosal blisters,(+) Genital blistering,(+) Arthralgia,(+) Nausea,(+) Episodic fatigue,(+) Vertigo
FAM184B	ENST00000265018:c.2750T>C p.Leu917Pro	ENST00000265018:c.1634G>T p.Gly545Val	comphet	3,54	comphet	maternal&paternal	2	Epilepsy	(+) EEG abnormality,(+) Generalized-onset seizure,(+) Autism,(+) Mild global developmental delay
TACC2	ENST00000369005:c.6763G>T p.Asp2255Tyr	ENST00000369005:c.7316G>A p.Arg2439Gln	comphet	3,73	comphet	maternal&paternal	2	Epilepsy	(+) EEG abnormality,(+) Generalized-onset seizure,(+) Autism,(+) Mild global developmental delay
PTPRD	ENST00000381196:c.3988G>A p.Gly1330Ser	ENST00000381196:c.1372G>A p.Asp458Asn	comphet	C	comphet	maternal&paternal	3	Malformations	hypotrophes Neugeborenes (Gewicht 5P, Länge 1P, Kopf 50P, 1z), Plagiozephalus DD Brachyzephalus, präaxiale Polydaktylie Typ 1 mit biphalangealem Daumen rechts, V.a. bikuspide Aortenklappe, Harntransprotströrung I-II° rechts und I° links, Neugeborenen-Hörscreening auffällig, Rektumstenose (Stoma), V.a. VACTERL-Assoziation (4/7 Symptomen), Körpermaße zur Vorstellung: Gewicht 52P, Größe 23P, Kopfumfang 10P
WDR5	ENST00000358625:c.620A>G p.Lys207Arg		de novo	B	het	de novo	3	Malformations	(+) Abnormality of the urinary system,(+) Brachycephaly,(+) Hearing abnormality,(+) Preaxial hand polydactyly,(+) Plagiocephaly,(+) Bicuspid aortic valve,(+) Anal stenosis hypotrophes Neugeborenes (Gewicht 5P, Länge 1P, Kopf 50P, 1z), Plagiozephalus DD Brachyzephalus, präaxiale Polydaktylie Typ 1 mit biphalangealem Daumen rechts, V.a. bikuspide Aortenklappe, Harntransprotströrung I-II° rechts und I° links, Neugeborenen-Hörscreening auffällig, Rektumstenose (Stoma), V.a. VACTERL-Assoziation (4/7 Symptomen), Körpermaße zur Vorstellung: Gewicht 52P, Größe 23P, Kopfumfang 10P
CKAP5	ENST00000529230:c.3056G>C p.Cys1019Ser		het	5,78	het	unknown	1	NDD	(+) Moderate global developmental delay,(+) Macrocephaly,(+) Muscular hypotonia,(+) Strabismus,(+) Midface retrusion,(+) Hand apraxia,(+) Large fontanelles

DST	ENST00000370788		de novo	8,5	het	de novo	2	NDD	(+) Moderate global developmental delay,(+) Muscular hypotonia,(+) Molar tooth sign on MRI,(+) Developmental cataract,(+) Febrile seizure (within the age range of 3 months to 6 years)
RC3H2	ENST00000373670:c.1A>G p.Met1?		het	5,7	het	unknown	1	Neuro	+) Generalized dystonia,(+) Intellectual disability,(+) Global developmental dela
PHF14	ENST00000403050:c.541del p.Lys182AsnfsTer19			5,3			1	Immunology	(+) Autoimmunity,(+) Hepatitis,(+) Recurrent fractures,(+) Allergy,(+) Abnormality of the face,(+) Unerupted tooth,(+) Recurrent infections
URGCP	NST00000453200:c.2192del p.Gln731ArgfsTer20		het	4,7	het	unknown	1	NDD + Epilepsy	(+) Intellectual disability, severe,(+) Severe global developmental delay,(+) Bilateral tonic-clonic seizure,(+) Abnormality of movement,(+) Short stature,(+) Failure to thrive,(+) Ventriculomegaly,(+) Hypoplasia of the corpus callosum,(+) Cerebellar hypoplasia,(+) Strabismus,(+) Microcephaly
PLCG1	ENST00000373272:c.1687C>T p.His563Tyr		het	4,45	het	unknown	1	NDD	(+) Joint hypermobility,(+) Pes cavus,(+) Poor gross motor coordination,(+) Delayed speech and language development,(+) Myopia,(+) Global developmental delay
SUDS3	ENST00000543473:c.557G>A p.Arg186Gln		de novo	5,83		de novo	1	NDD + Epilepsy	(+) Global developmental delay,(+) Motor delay,(+) Muscular hypotonia,(+) Frontal bossing,(+) Depressed nasal bridge,(+) Anteverted nares,(+) Hypertelorism,(+) Epicanthus,(+) Bifid uvula,(+) Sacral dimple,(+) Prominent fingertip pads,(+) Atopic dermatitis,(+) Hypermetropia,(+) Strabismus
KIF20B	ENST00000371728:c.2035_2037del p.Ile679del		homo	6,25	homo	maternal&paternal	3	Epilepsy	focal onset seizures
RXFP2	ENST00000298386:c.1594C>T p.Arg532Ter	ENST00000298386:c.1600G>A p.Gly534Arg	comphet	4,49	comphet	maternal&paternal	3	Epilepsy	focal onset seizures
LRRC3C	ENST00000377924:c.244C>T p.Arg82Cys	ENST00000377924:c.769C>T p.Arg257Trp	comphet	3,06	comphet	maternal&paternal	3	Epilepsy	focal onset seizures
G2E3	00000206595:c.419_420del p.Glu140ValfsTer12		het	5,1	het	unknown	1	NDD	(+) Global developmental delay,(+) Stereotypy,(+) Autism,(+) Poor speech,(+) Muscular hypotonia
AHNAK	ENST00000378024:c.15413C>T p.Ala5138Val	ENST00000378024:c.725C>T p.Ser242Leu	comphet	4,1	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Mild global developmental delay,(+) Delayed speech and language development,(+) Behavioral abnormality,(+) Delayed social development,(+) Abnormal facial shape,(+) Focal-onset seizure
FRMPD3	ENST00000276185:c.1379G>C p.Ser460Thr		hemi	3,3	hemi	maternal	2	NDD + Epilepsy	(+) Mild global developmental delay,(+) Delayed speech and language development,(+) Behavioral abnormality,(+) Delayed social development,(+) Abnormal facial shape,(+) Focal-onset seizure
UBE4B	ENST00000343090:c.2754G>A p.Met918Ile			4,4		unknown	1	Epilepsy	(+) Focal atonic seizure
EVPL	ENST00000301607:c.505G>C p.Gly169Arg		homo	3,9	homo	maternal&paternal	1	Epilepsy	Focal seizures since the age of ten, genetically proved Ehlers Danlos syndrome
PPP4C	ENST00000279387:c.214G>A p.Val72Ile		het	C	het	unknown	1	Immunology	(+) Immunodeficiency
REPS2	ENST00000357277:c.1930C>T p.Arg644Ter		hemi	6,9	hemi	maternal	4	NDD	Dystonia,(+) Intellectual disability,(+) Developmental regression
UBR4	ENST00000375254:c.5500A>G p.Ser1834Gly		de novo	9,3	het	de novo	4	NDD	Dystonia,(+) Intellectual disability,(+) Developmental regression
CCDC155	ENST00000447857:c.1214del p.Ile405ThrfsTer40		homo	8,08	homo	maternal&paternal	4	NDD	Dystonia,(+) Intellectual disability,(+) Developmental regression
NR2E1	ENST00000368986:c.1154T>C p.Ile385Thr		de novo	8,6	het	de novo	2	NDD	(+) Global developmental delay,(+) Intellectual disability, mild,(+) Sleep disturbance,(+) Short toe,(+) Periauricular sinus and cyst
RLF	00372771:c.5215_5216del p.Val1739LysfsTer12		de novo	9,8	het	de novo	2	NDD	(+) Global developmental delay,(+) Intellectual disability, mild,(+) Sleep disturbance,(+) Short toe,(+) Periauricular sinus and cyst
SCRIB	ENST00000356994:c.4896A>T p.Glu1632Asp	ENST00000356994:c.2224A>G p.Ile742Val	comphet	C	comphet	maternal&paternal	1	Growth, skeletal	(+) Scaphocephaly
SEC14L5	ENST00000251170:c.1368G>C p.Gln456His		de novo	B	het	de novo	1	Growth, skeletal	(+) Craniosynostosis,(+) Trigonocephaly, no neurodevelopmental delay
SPTBN5	ENST00000320955:c.9170G>A p.Arg3057Gln		homo	5,1	homo	maternal&paternal	5	Neuro	(-) Abnormality of brain morphology,(+) Lower limb spasticity
ZSCAN29	ENST00000396976:c.1298G>A p.Arg433Gln		homo	3,8	homo	maternal&paternal	5	Neuro	(-) Abnormality of brain morphology,(+) Lower limb spasticity
SSFA2	ENST00000431877:c.1060_1062del p.Ser354del		homo	4,4	homo	maternal&paternal	5	Neuro	(-) Abnormality of brain morphology,(+) Lower limb spasticity
ERBB2IP	ENST00000506030:c.472G>A p.Gly158Ser	ENST00000506030:c.2473C>T p.His825Tyr	comphet	5,6	comphet	maternal&paternal	5	Neuro	(-) Abnormality of brain morphology,(+) Lower limb spasticity
CORO1C	ENST00000261401:c.318+1G>C None		de novo	9,0	het	de novo	1	NDD + Epilepsy	(+) Seizure,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Mild global developmental delay,(+) EEG abnormality
CDC42BPB	ENST00000361246:c.4610C>G p.Pro1537Arg	ENST00000361246:c.1439A>G p.Asn480Ser	comphet	5,2	comphet	maternal&paternal	2	Epilepsy	(+) Focal-onset seizure,(+) Febrile seizure (within the age range of 3 months to 6 years),(-) Global developmental delay
PPP3CB	ENSP00000378306:c.2758T>C p.Cys920Arg		het	C	het	unknown	1	Growth, skeletal	(+) Fused cervical vertebrae,(+) Sprengel anomaly,(+) Scoliosis,(+) Short stature,(+) Abnormal facial
BIRC6	ENST00000421745:c.9946T>G p.Phe3316Val		het	B	het	unknown	1	Malformations	(+) Hypopituitarism,(+) Optic atrophy,(+) Septo-optic dysplasia,(+) Cerebellar hypoplasia,(+) Hypoplasia of the corpus callosum,(+) Hypoglycemia,(+) Patellar hypoplasia
PTBP3	ENST00000458258:c.207del p.Arg70GlufsTer15		het	4,7	het	unknown	1	NDD	(+) Mild global developmental delay,(+) Short stature
FAT3	ENST00000298047:c.4430C>T p.Thr1477Met	ENST00000298047:c.10819G>A p.Ala3607Thr	comphet	4,4	comphet	maternal&paternal	2	Epilepsy	(+) Focal-onset seizure,(+) Generalized-onset seizure,(+) Migraine with aura,(+) Scoliosis,(+) Patent foramen ovale,(+) Mitral valve prolapse,(+) Mitral regurgitation
KANK4	ENST00000317477:c.2587A>G p.Met863Val	ENST00000371153:c.1957C>T p.Arg653Cys	comphet	3,0	comphet	maternal&paternal	2	Epilepsy	(+) Focal-onset seizure,(+) Generalized-onset seizure,(+) Migraine with aura,(+) Scoliosis,(+) Patent foramen ovale,(+) Mitral valve prolapse,(+) Mitral regurgitation
MFSD9	ENST00000258436:c.391A>G p.Asn131Asp	1011840-2011844	homo	4,3	homo	unknown	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
ATOH8	ENST00000306279:c.124A>G p.Thr42Ala		homo	3,2	homo	unknown	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
PMEL	ENST00000449260:c.727C>T p.Gln243Ter		homo	7,3	homo	unknown	3	NDD + Epilepsy	(+) Autism,(+) Generalized tonic seizure,(+) Intellectual disability, severe,(+) Parietal cortical atrophy,(+) Occipital cortical atrophy,(+) Bilateral tonic-clonic seizure with generalized onset
ABHD14B	ENST00000483233:c.536G>A p.Arg179Gln	ENST00000483233:c.250A>G p.Ile84Val	comphet	3,1	comphet	maternal&paternal	3	NDD	(Global developmental delay),(+) Dysphagia,(+) Infantile muscular hypotonia

ROCK1	ENST00000399799:c.4019A>T p.Gln1340Leu		het	4,1	het	unknown	3	NDD + Epilepsy	(+) Autism,(+) Generalized tonic seizure,(+) Intellectual disability, severe,(+) Parietal cortical atrophy,(+) Occipital cortical atrophy,(+) Bilateral tonic-clonic seizure with generalized onset
RC3H2	ENST00000373670:c.2386A>G p.Thr796Ala	ENST00000373670:c.1124A>G p.Glu375Gly	comphet	3,6	comphet	maternal&paternal	3	NDD	(Global developmental delay),(+) Dysphagia,(+) Infantile muscular hypotonia
GABRE	ENST00000370328:c.572T>C p.Ile191Thr		hemi	5,3	hemi	maternal	3	NDD	(Global developmental delay),(+) Dysphagia,(+) Infantile muscular hypotonia
CHD9	ENST00000566029:c.7279A>T p.Ile2427Phe		het	3,9	het	unknown	2	NDD	(+) Delayed speech and language development,(+) Motor delay,(+) Severe expressive language delay
RRN3	ENST00000198767:c.1267A>G p.Lys423Glu		de novo	5,67	het	de novo	2	NDD	Inguinal hernia,(+) Retinal coloboma,(+) Microphthalmia,(+) Optic nerve hypoplasia,(+) Delayed speech and language development,(+) Pectus excavatum of inferior sternum,(+) Hypopigmented skin patches,(+) Hypotonia,(+) Intrauterine growth retardation,(+) Abnormal facial shape,(+) Congenital nystagmus,(+) Mild
PDZD4	000164640:c.1782_1784delinsAGG p.Glu595Gly		hemi	4,9	hemi	maternal	2	NDD	Inguinal hernia,(+) Retinal coloboma,(+) Microphthalmia,(+) Optic nerve hypoplasia,(+) Delayed speech and language development,(+) Pectus excavatum of inferior sternum,(+) Hypopigmented skin patches,(+) Hypotonia,(+) Intrauterine growth retardation,(+) Abnormal facial shape,(+) Congenital nystagmus,(+) Mild
UNC13C	ENST00000260323:c.422del p.Gln141ArgfsTer36		homo	9,9	homo	unknown	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
KIF27	ENST00000297814:c.1016G>A p.Arg339Gln		homo	6,0	homo	unknown	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
SEMA6B	ENST00000586582:c.199A>G p.Ile67Val		het	3,9	het	unknown	2	NDD + Epilepsy	(+) Microcephaly,(+) Optic atrophy,(+) Aggressive behavior,(+) Melanocytic nevus,(+) Hypotonia,(+) Spasticity,(+) Scoliosis,(+) Hypokalemia,(+) Pontocerebellar atrophy,(+) Focal tonic seizure,(+) Severe global developmental delay,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Dystonic gait,(+) Epileptic
SLC4A7	ENST00000295736:c.307G>A p.Asp103Asn		homo	5,3	homo	unknown	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
TIAM2	1783:c.2967_2968insGTGAC p.Leu990ValfsTer46	ENST00000461783:c.3935G>A p.Ser1312Asn	comphet?	5,5	comphet?	unknown	2	NDD + Epilepsy	(+) Pontocerebellar atrophy,(+) Focal tonic seizure,(+) Severe global developmental delay,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Dystonic gait,(+) Epileptic encephalopathy, (+) Microcephaly,(+) Optic atrophy,(+) Aggressive behavior,(+) Melanocytic nevus,(+) Hypotonia,(+) Spasticity,(+) Scoliosis,(+) Hypokalemia
PIAS2	ENST00000585916:c.376del p.Thr126LeufsTer23			5,55		unknown	1	NDD + Growth	(+) developmenal dealy (IQ 68) (+) puberty praecox (+) recurrent infections with
PRR32	ENST00000371125:c.751C>T p.Arg251Ter		homo	3,5	hom	unknown	8	NDD + Epilepsy	(+) Microcephaly,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Abnormal cardiac MRI
ZNF701	ENST00000540331:c.842del p.Phe281SerfsTer16		homo	4,0	hom	unknown	8	NDD + Epilepsy	(+) Microcephaly,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Abnormal cardiac MRI
UBC	ENST00000541272:c.277-14_502del		homo	7,2	hom	unknown	8	NDD + Epilepsy	(+) Microcephaly,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Abnormal cardiac MRI
LRP8	ENST00000306052:c.100G>T p.Ala34Ser		het	5,0	het	unknown	1	NDD	(+) Autism,(+) Delayed speech and language development,(+) Hyperactivity,(+) Intellectual disability, borderline
IMPDH2	ENST00000326739:c.687_689del p.Lys229del		de novo	7,1	het	de novo	1	Malformations	(-) Cleft palate,(+) Microcephaly,(+) Webbed neck,(+) Cutis laxa,(+) Dandy-Walker malformation,(+) Joint laxity,(+) Intrauterine growth retardation,(+) Small for gestational age,(+) Premature birth,(+) Ventricular septal defect,(+) Dextrocardia,(+) Hypoplasia of the corpus callosum,(+) Scoliosis,(+) Reduced subcutaneous adipose tissue,(+) Persistent patent ductus venosus,(+) Abnormality of the scalp hair,(+) Palpebral edema,(+) Long fingers
CAPZB	ENST00000375142:c.*11-2A>G None		het	7,0	het	unknown	1	NDD + Hypotonie	(+) Abnormality of upper lip,(+) Epicanthus,(+) High forehead,(+) Single transverse palmar crease,(+) Hypotonia,(+) Motor delay,(+) Failure to thrive
SUPT5H	ENST00000599117:c.2377C>T p.Leu793Phe		het	3,7	het	unknown	1	NDD + Epilepsy	(+) Abnormality of the face,(+) Hypotonia,(+) Focal-onset seizure,(+) Intellectual disability, severe,(+) Muscular ventricular septal defect
ZMYM2	ENST00000382869:c.2321A>G p.Gln774Arg		het	5,8	het	unknown	2	Epilepsy	(+) Typical absence seizure,(+) Bilateral tonic-clonic seizure with generalized onset
DOCK4	ENST00000437633:c.593G>C p.Ser198Thr		het	5,5	het	unknown	2	Epilepsy	(+) Typical absence seizure,(+) Bilateral tonic-clonic seizure with generalized onset
TLN1	ENST00000314888:c.580C>T p.Arg194Trp		het	4,55	het	unknown	1	NDD + Epilepsy	(+) Dolichocephaly,(+) Intellectual disability,(+) Global developmental delay,(+) Sagittal craniosynostosis,(+) Bilateral superior vena cava
WDR24	ENST00000293883:c.2005G>T p.Asp669Tyr		het	C	het	unknown	3	Neuro	(+) Spastic gait,(+) Babinski sign,(+) Impaired distal tactile sensation,(+) Abnormal pyramidal sign,(+) Multifocal hyperintensity of cerebral white matter on MRI
MYCBP2	ENST00000544440:c.7277A>G p.Gln2426Arg		het	C	het	unknown	3	Neuro	(+) Spastic gait,(+) Babinski sign,(+) Impaired distal tactile sensation,(+) Abnormal pyramidal sign,(+) Multifocal hyperintensity of cerebral white matter on MRI
NAA35	ENST00000361671:c.686A>G p.Gln229Arg		het	C	het	unknown	3	Neuro	(+) Spastic gait,(+) Babinski sign,(+) Impaired distal tactile sensation,(+) Abnormal pyramidal sign,(+) Multifocal hyperintensity of cerebral white matter on MRI
PLXNA4	ENST00000359827:c.1246T>A p.Ser416Thr		het	4,3	het	unknown	2	Epilepsy	(+) EEG abnormality,(+) Focal motor seizure,(+) Eating-induced seizure,(+) Somatosensory-induced seizure,(+) Generalized-onset motor seizure
KIAA1239	ENST00000309447:c.280G>A p.Asp94Asn		het	3,3	het	unknown	2	Epilepsy	(+) EEG abnormality,(+) Focal motor seizure,(+) Eating-induced seizure,(+) Somatosensory-induced seizure,(+) Generalized-onset motor seizure
RNF20	ENST00000389120:c.716T>C p.Leu239Pro		het	5,5	het	paternal	2	NDD + Epilepsy	(+) Seizure,(+) Abnormal facial shape,(+) Severe muscular hypotonia,(+) Severe global developmental delay
XPO1	ENST00000401558:c.431A>G p.Lys144Arg		het	5,4	het	paternal	2	NDD + Epilepsy	(+) Seizure,(+) Abnormal facial shape,(+) Severe muscular hypotonia,(+) Severe global developmental delay

SRRM2	ENST00000301740:c.5653C>T p.Arg1885Ter		het	7,02	het	unknown	1	NDD + Epilepsy	(+) Seizure (+) Motor Delay (+) Microcephaly
PSPC1	ENST00000338910:c.92C>T p.Pro31Leu		het	3,5	het	unknown	2	Epilepsy	(+) Myoclonic absence seizure
NFATC3	ENST00000346183:c.1774+1G>A None		het	6,2	het	unknown	2	Epilepsy	(+) Myoclonic absence seizure
WNK2	ST00000297954:c.3381del p.Lys1127AsnfsTer23		het	5,7	het	unknown	1	NDD	(+) Microcephaly,(+) Pectus excavatum,(+) Hypotonia,(+) Global developmental delay,(+) Pes planus,(+) Pes valgus
SRRM2	ENST00000301740:c.1585C>T p.Gln529Ter		het	8,4	het	unknown	1	NDD	(-) Abnormality of the face,(+) Behavioral abnormality,(+) Short attention span,(+) Low frustration tolerance,(+) Delayed speech and language development,(+) Global developmental delay,(+) Obesity
SCAF11	ENST00000369367:c.1146del p.Lys382AsnfsTer5		het	6,2	het	unknown	3	NDD	(+) Abnormality of the face,(+) Ataxia,(+) Global developmental delay,(+) EEG abnormality,(+) Poor speech
UNC79	ENST00000256339:c.3515T>C p.Met1172Thr		homo	7,3	homo	unknown	3	NDD	(+) Abnormality of the face,(+) Ataxia,(+) Global developmental delay,(+) EEG abnormality,(+) Poor speech
ATP13A1	ENST00000357324:c.2699C>T p.Pro900Leu		het	4,1	het	unknown	3	NDD	(+) Abnormality of the face,(+) Ataxia,(+) Global developmental delay,(+) EEG abnormality,(+) Poor speech
PCSK5	ENST00000545128:c.1024G>A p.Gly342Arg		het	4,8	het	unknown	2	NDD	(+) Hypotonia,(+) Global developmental delay,(+) Failure to thrive in infancy,(+) Slender build,(+) Intellectual disability, moderate,(+) Abnormality of dental morphology,(+) Mild intrauterine growth retardation,(+) Broad hallux,(+) Long toe,(+) Primary microcephaly,(+) Long fingers
TMEM132D	ENST00000422113:c.79+1G>A None		het	5,8	het	unknown	2	NDD	(+) Hypotonia,(+) Global developmental delay,(+) Failure to thrive in infancy,(+) Slender build,(+) Intellectual disability, moderate,(+) Abnormality of dental morphology,(+) Mild intrauterine growth retardation,(+) Broad hallux,(+) Long toe,(+) Primary microcephaly,(+) Long fingers
HSPA4	ENST00000304858:c.792dup p.Arg265ThrfsTer7		unknown	8,7	unknown	unknown		NDD	(+) Tall stature,(+) Autism,(+) Autistic behavior,(+) Hypotonia,(+) Global developmental delay
PHC2	ENST00000257118:c.383C>T p.Ser128Phe		het	4,9	het	unknown	2	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Global brain atrophy,(+) Secondary microcephaly
NPIPB5	ENST00000424340:c.1505C>T p.Pro502Leu		de novo	4,3	het	de novo	2	NDD	(+) Hypertelorism,(+) Abnormal eyebrow morphology,(+) Triphalangeal thumb,(+) Intellectual disability,(+) Global developmental delay
TRAPPC1	ENST00000303731:c.293A>C p.His98Pro	ENST00000303731:c.215A>G p.His72Arg	comphet?	6,3	comphet?	unknown	2	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Global brain atrophy,(+) Secondary microcephaly
UBR2	ENST000003728899: c.1532T>C p.Leu511Pro	ENST000003728899: c.5026G>A p.Val1676Ile	comphet	5,6	comphet	maternal&paternal	2	NDD	(+) Hypertelorism,(+) Abnormal eyebrow morphology,(+) Triphalangeal thumb,(+) Intellectual disability,(+) Global developmental delay
KDM1A	ENST00000400181:c.1894C>T p.Arg632Cys		de novo	9,65	het	de novo	1	NDD + Epilepsy	(+) Seizure,(+) Hypsarrhythmia,(+) Moderate global developmental delay,(+) Epileptic encephalopathy
CUL2	IST00000537177:c.1043dup p.Asn348LysfsTer21		het	7,0	het	unknown	3	NDD	(+) Epicanthus,(+) Round face,(+) Short philtrum,(+) Short chin,(+) Large hands,(+) Prominent fingertip pads,(+) Hypotonia,(+) Specific learning disability,(+) Joint hypermobility,(+) Obesity,(+) Pes planus,(+) Impaired continence,(+) Lip hyperpigmentation
TRIM9	ENST00000298355:c.386C>T p.Pro129Leu		het	5,0	het	unknown	3	NDD	(+) Epicanthus,(+) Round face,(+) Short philtrum,(+) Short chin,(+) Large hands,(+) Prominent fingertip pads,(+) Hypotonia,(+) Specific learning disability,(+) Joint hypermobility,(+) Obesity,(+) Pes planus,(+) Impaired continence,(+) Lip hyperpigmentation
PHF21A	ENST00000418153:c.882A>G p.Ile294Met		het	6,4	het	unknown	3	NDD	(+) Epicanthus,(+) Round face,(+) Short philtrum,(+) Short chin,(+) Large hands,(+) Prominent fingertip pads,(+) Hypotonia,(+) Specific learning disability,(+) Joint hypermobility,(+) Obesity,(+) Pes planus,(+) Impaired continence,(+) Lip hyperpigmentation
SRRM2	00301740:c.6774_6775del p.Arg2260AsnfsTer26		het	7,67	het	unknown	1	NDD	(+) Seizure,(+) Status epilepticus,(+) Complex febrile seizure
SPTBN1	ENST00000356805:c.2047T>C p.Phe683Leu		het	5,2	het	unknown	2	NDD	+) Autism,(+) Hypotonia,(+) Global developmental delay,(+) Absent speech
XIRP2	ENST00000409195:c.3288G>A p.Trp1096Ter	ENST00000409195:c.6515T>C p.Val2172Ala	comphet?	5,4	comphet?	unknown	2	NDD	+) Autism,(+) Hypotonia,(+) Global developmental delay,(+) Absent speech
STARD9	ENST00000290607:c.8609C>T p.Thr2870Ile		de novo	5,4	het	de novo	1	Malformations	(+) Intrauterine growth retardation,(+) Abnormality of ductus venosus blood flow,(+) Abnormality of umbilical vein blood flow
AGO1	ENST00000373204:c.86T>C p.Ile29Thr		het	6,0	het	unknown	3	NDD + Epilepsy	(+) Developmental stagnation at onset of seizures,(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal motor seizure
CADPS	ENST00000383710:c.2681A>G p.Gln894Arg		het	5,8	het	unknown	3	NDD + Epilepsy	(+) Developmental stagnation at onset of seizures,(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal motor seizure
MYCBP2	ENST00000544440:c.8456C>T p.Pro2819Leu		het	5,2	het	unknown	3	NDD + Epilepsy	(+) Developmental stagnation at onset of seizures,(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal motor seizure
RHOT1	T00000358365:c.517_538del p.Leu173ArgfsTer2		unknown	7,43	het	unknown	1	NDD + Epilepsy	(+) Delayed speech and language development,(+) Dystonia,(+) Migraine,(+) Hemiplegia,(+) Hemiplegia/hemiparesis
FLRT2	ENST00000330753:c.1102C>T p.Pro368Ser	ENST00000330753:c.1766G>A p.Cys589Tyr	comphet	B	comphet	maternal&paternal	2	Muscle	(+) Generalized hypotonia,(+) Motor axonal neuropathy,(+) Progressive distal muscle weakness
KDM4C	ENST00000381309:c.629+6T>G None		de novo	B	het	de novo	2	Muscle	(+) Generalized hypotonia,(+) Motor axonal neuropathy,(+) Progressive distal muscle weakness
KCND2	ENST00000331113:c.107A>G p.Lys36Arg		het	5,76	het	unknown		NDD + Epilepsy	(+) Hydrocephalus,(+) Intellectual disability,(+) Myoclonic seizure
ARCN1	ENST00000264028:c.134A>G p.Gln45Arg		het	6,7	het	unknown	1	NDD	(+) Global developmental delay
BAZ1A	ENST00000360310:c.1252A>G p.Thr418Ala		het	4,91	het	unknown	1	NDD	(+) Abnormality of the face,(+) Autism,(+) Seizure,(+) Mild global developmental

RIF1	ENST00000243326:c.7095+2T>C None		het	7,2	het	unknown	3	NDD	(+) Epicanthus,(+) Upslanted palpebral fissure,(+) Hypotelorism,(+) Hyperactivity,(+) Global developmental delay,(+) Absent speech,(+) Primary microcephaly
TLN1	ENST00000314888:c.6473C>T p.Ala2158Val		het	4,2	het	unknown	3	NDD	(+) Epicanthus,(+) Upslanted palpebral fissure,(+) Hypotelorism,(+) Hyperactivity,(+) Global developmental delay,(+) Absent speech,(+) Primary microcephaly
ACLY	000352035:c.1587_1596del p.Met529IlefsTer18	ENST00000352035:c.616+4A>T	comphet?	6,6	comphet?	unknown	3	NDD	(+) Epicanthus,(+) Upslanted palpebral fissure,(+) Hypotelorism,(+) Hyperactivity,(+) Global developmental delay,(+) Absent speech,(+) Primary microcephaly
PDCL	ENST00000259467:c.203G>A p.Arg68His		homo	5,57	homo	maternal&paternal	4	NDD + Epilepsy	(+) Microcephaly,(+) Cerebellar hypoplasia,(+) Cerebral atrophy,(+) Hypoplasia of the corpus callosum,(+) Status epilepticus,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Delayed myelination
FAM78A	ENST00000372271:c.496G>A p.Val166Ile		homo	4,16	homo	maternal&paternal	4	NDD + Epilepsy	(+) Microcephaly,(+) Cerebellar hypoplasia,(+) Cerebral atrophy,(+) Hypoplasia of the corpus callosum,(+) Status epilepticus,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Delayed myelination
DUSP8	ENST00000397374:c.36T>G p.Asp12Glu		homo	3,3	homo	maternal&paternal	4	NDD + Epilepsy	(+) Microcephaly,(+) Cerebellar hypoplasia,(+) Cerebral atrophy,(+) Hypoplasia of the corpus callosum,(+) Status epilepticus,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Delayed myelination
GRIK1	ENST00000399907:c.10G>T p.Gly4Cys		homo	7,77	homo	maternal&paternal	4	NDD + Epilepsy	(+) Microcephaly,(+) Cerebellar hypoplasia,(+) Cerebral atrophy,(+) Hypoplasia of the corpus callosum,(+) Status epilepticus,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Delayed myelination
ADGRB2	ENST00000373658:c.1203C>A p.Cys401Ter		het	5,89	het	unknown	1	NDD	(+) Behavioral abnormality,(+) Delayed speech and language development,(+) Severe global developmental delay
ITPR3	ENST00000374316:c.143C>G p.Pro48Arg	ENST00000447857:c.1214del p.Ile405ThrfsTer40	het	6,3	het	unknown	2	NDD	(+) Short attention span,(+) Dyslexia,(+) Abnormal social behavior,(+) Abnormal emotion/affect behavior
LPHN3	ENST00000514591:c.4292A>G p.His1431Arg		het	5,5	het	unknown	2	NDD	(+) Short attention span,(+) Dyslexia,(+) Abnormal social behavior,(+) Abnormal emotion/affect behavior
BZRAP1	ENST00000343736:c.5540G>A p.Ser1847Asn	ENST00000343736:c.4348G>T p.Gly1450Cys	comphet?	5,1	comphet?	unknown	1	NDD + Epilepsy	(+) Autism,(+) Delayed speech and language development,(+) Bilateral tonic-clonic seizure,(+) Mild global developmental delay
C1orf109	ENST00000358011:c.218T>A p.Leu73Gln		homo	5,0	homo	unknown	2	NDD	(+) Microcephaly,(+) Hypertonia,(+) Failure to thrive,(+) Increased serum lactate,(+) Opisthotonus,(+) Poor motor coordination,(+) Increased CSF lactate,(+) Hyperalaninemia,(+) Severe global developmental delay,(+) Abnormal visual fixation
USP21	ENST00000368002:c.709C>T p.Arg237Trp		homo	4,9	homo	unknown	2	NDD	(+) Microcephaly,(+) Hypertonia,(+) Failure to thrive,(+) Increased serum lactate,(+) Opisthotonus,(+) Poor motor coordination,(+) Increased CSF lactate,(+) Hyperalaninemia,(+) Severe global developmental delay,(+) Abnormal visual fixation
ARHGDIB	ENST00000228945:c.239C>T p.Pro80Leu		homo	4,22	homo	maternal&paternal	5	Neuro	(-) Abnormality of brain morphology,(+) Lower limb spasticity
NAP1L1	ENST00000261182:c.1058_1059+1dup		het	6,3	het	unknown	1	Epilepsy + ASD	(+) Tall stature,(+) Autistic behavior,(+) Short attention span,(+) Delayed speech and language development,(+) Generalized non-motor (absence) seizure,(+) Diminished ability to concentrate
HTR3E	ENST00000440596:c.1031T>C p.Leu344Pro		de novo	4,4	het	de novo	3	NDD	(+) Intellectual disability,(+) Cortical dysplasia,(+) Focal-onset seizure
XIRP2	ENST00000409195:c.5646G>A p.Trp1882Ter	ENST00000409043:c.*1158G>A p.Gly810Glu	comphet	5,1	comphet	maternal&paternal	3	NDD	(+) Intellectual disability,(+) Cortical dysplasia,(+) Focal-onset seizure
KCP	ENST00000476647:n.4653C>T None	ENST00000476647:n.1049+2T>G None	comphet	B	comphet	maternal&paternal	3	Malformations	hypotrophes Neugeborenes (Gewicht 5P, Länge 1P, Kopf 50P, 1z), Plagiozephalus DD Brachyzephalus, präaxiale Polydaktylie Typ 1 mit biphalangealem Daumen rechts, V.a. bikuspide Aortenklappe, Harntransprotströrung I-II° rechts und I° links, Neugeborenen-Hörscreening auffällig, Rektumstenose (Stoma), V.a. VACTERL-Assoziation (4/7 Symptomen), Körpermaße zur Vorstellung: Gewicht 52P, Größe 23P, Kopfumfang 10P
KCNG2	ENST00000316249:c.11G>A p.Trp4Ter		het	4,8	het	unknown	1	NDD	(+) Obsessive-compulsive behavior,(+) Global developmental delay,(+) Obesity,(+) Postural instability,(+) Sleep disturbance,(+) Highly arched eyebrow,(+) Polyphagia,(+) Poor fine motor coordination,(+) Dyslexia
CDC42BPG	ENST00000342711:c.1289G>A p.Ser430Asn		de novo	4,7	het	de novo	1	Epilepsy	(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal motor seizure,(+) Autonomic epileptic aura
TOP2B	ENST00000435706:c.3360A>T p.Gln1120His		het	5,4	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Episodic hemiplegia
PBRM1	ENST00000394830:c.233G>A p.Arg78Gln		het	**4,5**	het	unknown	1	NDD	(+) Autism,(+) Intellectual disability,(+) Seizure,(+) Scoliosis,(+) Severe global developmental delay
HDAC1	ENST00000373548:c.1322A>G p.Lys441Arg		het	**5,4**	het	unknown	1	NDD + Epilepsy	(+) Intellectual disability,(+) Focal-onset seizure,(+) Myoclonic absence seizure,(+) Moderate global developmental delay,(+) Mild malformation of cortical development
HUWE1	ENST00000342160:c.12115C>T p.Pro4039Ser		hemi	**B**	hemi	maternal	1	Malformations	(+) Renal insufficiency,(+) Aortic valve stenosis,(+) Respiratory insufficiency,(+) Hyperechogenic kidneys,(+) Elevated C-reactive protein level
PRKCB	ENST00000303531:c.1810G>C p.Asp604His		het	**5,0**	het	unknown	1	NDD	(+) Microcephaly,(+) Short stature,(+) Moderate global developmental delay
SIPA1L1	ENST00000555818:c.5402T>C p.Ile1801Thr		unknown	4,33	het	unknown	1	NDD + Epilepsy	(+) Cleft palate,(+) Seizure,(+) Ataxia,(+) Spasticity,(+) Short stature,(+) Severe global developmental delay,(+) Cleft lip
ANKRD28	ENST00000399451:c.3065C>G p.Pro1022Arg		de novo	**4,9**	het	de novo	4	NDD	(+) Wide mouth,(+) Coarse facial features,(+) Autism,(+) Intellectual disability,(+) Moderate global developmental delay
USP39	ENST00000323701:c.1498A>C p.Ile500Leu		de novo	**5,9**	het	de novo	4	NDD	(+) Wide mouth,(+) Coarse facial features,(+) Autism,(+) Intellectual disability,(+) Moderate global developmental delay
CAPN8	ENST00000366872:c.34C>T p.Arg12Trp		homo	**3,6**	homo	maternal&paternal	4	NDD	(+) Wide mouth,(+) Coarse facial features,(+) Autism,(+) Intellectual disability,(+) Moderate global developmental delay
SLC44A2	ENST00000335757:c.1060G>A p.Val354Met	ENST00000335757:c.1061T>C p.Val354Ala	comphet	**3,7**	comphet	maternal&paternal	4	NDD	(+) Wide mouth,(+) Coarse facial features,(+) Autism,(+) Intellectual disability,(+) Moderate global developmental delay

NCKAP1	ENST00000360982:c.1138G>T p.Ala380Ser		het	6,0	het	unknown	1	NDD + Epilepsy	(+) Microcephaly,(+) Behavioral abnormality,(+) Seizure,(+) Moderate global developmental delay,(+) Dissociative reaction
PITPNM2	ENST00000320201:c.643+2T>C None		het	5,94	het	unknown	1	NDD + Epilepsy	(+) Coarse facial features,(+) Aggressive behavior,(+) Seizure,(+) Obesity,(+) Moderate global developmental delay
ASTN1	ENST00000361833:c.3622C>T p.Arg1208Ter		de_novo	A	het	de novo	1	Neuro	(+) Depression,(+) Headache,(+) Progressive neurologic deterioration,(+) Nonprogressive cerebellar ataxia,(+) Anti-Yo antibody
EP400	ENST00000389561:c.2665C>T p.Gln889Ter		unknown	6,5	het	unknown	2	NDD	(+) Global developmental delay,(+) Agenesis of corpus callosum
ZBTB10	ENST00000430430:c.2203C>T p.Arg735Ter		unknown	5,0	het	unknown	2	NDD	(+) Global developmental delay,(+) Agenesis of corpus callosum
UBR2	ENST00000372899:c.4319G>A p.Gly1440Glu		de_novo	7,4	het	de novo	1	NDD	(+) Abnormal lip morphology,(+) Thick lower lip vermilion,(+) Open mouth,(+) Coarse facial features,(+) Intellectual disability,(+) Global developmental delay,(+) Abnormal facial shape,(+) Thick vermilion border
ST3GAL2	ENST00000393640:c.420del p.Tyr141ThrfsTer37		unknown	5,6	het	unknown	1	NDD + Epilepsy	(+) Seizure,(+) Neonatal hypoglycemia,(+) Generalized non-motor (absence) seizure,(-) EEG abnormality,(+) Proportionate short stature,(-) Abnormal cardiac MRI
CPSF3	ENST00000238112:c.1147C>A p.Pro383Thr		unknown	C	het	unknown	1	Immunology	(+) Episodic abdominal pain,(+) Periodic fever
ANXA11	ENST00000438331:c.1403A>G p.Asp468Gly		de_novo	4,86	het	de novo		NDD	(+) Behavioral abnormality,(+) Dementia,(+) Intellectual disability, mild,(+) Motor delay,(+) Neurological speech impairment,(+) Global brain atrophy,(+) Sleep disturbance,(+) Encephalitis,(+) Pica
MRPL42	ENST00000549982:c.143A>G p.Glu48Gly		de_novo	4,9	het	de novo		NDD	(+) Behavioral abnormality,(+) Dementia,(+) Intellectual disability, mild,(+) Motor delay,(+) Neurological speech impairment,(+) Global brain atrophy,(+) Sleep disturbance,(+) Encephalitis,(+) Pica
ARFGEF1	ENST00000262215:c.1028-2A>T None		unknown	7,9	het		1	NDD + Epilepsy	(+) Intellectual disability,(+) Focal-onset seizure
HSPA4	ENST00000304858:c.1450G>C p.Val484Leu		homo	7,8	homo	unknown	3	NDD	(+) Intellectual disability,(+) Hypotonia,(+) Mild global developmental delay,(+) Abnormal ear morphology
GPR84	ENST00000551809:c.895del p.Gln299SerfsTer19		homo	8,4	homo	unknown	3	NDD	(+) Intellectual disability,(+) Hypotonia,(+) Mild global developmental delay,(+) Abnormal ear morphology
MYO1A	NST00000442789:c.2827del p.Val943CysfsTer15		homo	8,6	homo	unknown	3	NDD	(+) Intellectual disability,(+) Hypotonia,(+) Mild global developmental delay,(+) Abnormal ear morphology
TMEM131L	ENST00000409959:c.1226G>A p.Trp409Ter		unknown	5,3	het		1	NDD	(+) Torticollis,(+) Nystagmus,(+) Behavioral abnormality,(+) Intellectual disability,(+) Global developmental delay,(+) Scoliosis,(+) Abducens palsy
AGAP2	ENST00000257897:c.52C>T p.Arg18Ter		unknown	7,1	het	unknown	1	neuro	+) Episodic ataxia
KCNG1	ENST00000371571:c.59C>T p.Ser20Leu		unknown	3,46	het	unknown	1	NDD + Epilepsy	(+) Epileptic encephalopathy
TLN2	00561311:c.4308_4309del p.Cys1436TrpfsTer17		unknown	6,3	het	unknown	1	NDD + Epilepsy	(+) Focal-onset seizure,(+) EEG with focal epileptiform discharges,(+) EEG with generalized epileptiform discharges,(+) Mild global developmental delay
MCMBP	ENST00000360003:c.1110A>G p.Ile370Met		de_novo	4,7	het	de novo		NDD	(+) Trigonocephaly,(+) Hypertelorism,(+) Upslanted palpebral fissure,(+) Autism,(+) Delayed speech and language development,(+) Hypotonia,(+) Clinodactyly of the 5th finger,(+) Moderate global developmental delay,(+) Epicanthus palpebralis
SYMPK	ENST00000245934:c.226-7_226-2del None		unknown	B	het	unknown	1	Muscle	(+) Motor delay,(+) Muscle weakness,(+) Lower limb muscle weakness,(+) Infantile muscular hypotonia
CHD1L	ENST00000369258:c.1086-2A>G None		unknown	6,7	het	unknown		Epilepsy	(+) Generalized non-motor (absence) seizure
DENR	00000280557:c.426_429del p.Glu143HisfsTer15		unknown	5,9	het	unknown	1	NDD + Epilepsy	(+) Open mouth,(+) Abnormality of the face,(+) Hypomimic face,(+) Intellectual disability,(+) Spastic diplegia,(+) Aphasia,(+) Focal-onset seizure,(+) Severe global developmental delay,(+) Happy demeanor
PTBP1	ENST00000356948:c.8+2T>G		unknown	8,3	het	unknown	1	Epilepsy	(+) Hydrocephalus,(+) Macrocephaly,(+) Headache,(+) Focal-onset seizure,(+) Episodic hemiplegia
DPP9	ENST00000262960:c.842G>C p.Arg281Pro		de_novo	A	het	de novo		Other	(+) Splenomegaly,(+) Pancytopenia,(+) Congenital thrombocytopenia,(+) Immunodeficiency,(+) Bone marrow hypocellularity,(+) Hemophagocytosis,(+)
PTPRN	ENST00000295718:c.1237A>G p.Thr413Ala		de_novo	5,8	het	de novo		NDD	(+) Epicanthus,(+) Depressed nasal ridge,(+) Upslanted palpebral fissure,(-) Intellectual disability,(+) Hypotonia,(+) Motor delay,(+) Expressive language delay,(+) Aplastic/hypoplastic toenail,(+) Oligodactyly,(+) Clinodactyly
WEE1	ENST00000450114:c.848G>A p.Arg283Lys		unknown	4,0	het	unknown	1	NDD	(+) Low-set, posteriorly rotated ears,(+) Abnormality of skin pigmentation,(+) Specific learning disability,(+) Mutism,(+) Intellectual disability, borderline,(+) Mild global developmental delay
LRRC37A2	ENST00000576629:c.4967C>G p.Pro1656Arg		de_novo	4,2	het	de novo	2	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
PLXND1	ENST00000324093:c.5657C>T p.Pro1886Leu	ENST00000324093:c.2668G>A p.Ala890Thr	comphet	5,6	comphet	maternal&paternal	2	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
ABLIM1	ENST00000277895:c.688G>A p.Gly230Arg		de_novo	6,9	het	de novo	2	Growth, skeletal	(+) Abnormal thumb morphology,(+) Preaxial hand polydactyly,(+) Vertebral segmentation defect,(+) Pilonidal sinus,(+) Muscular ventricular septal defect,(+) Perimembranous ventricular septal defect
MYO7B	ENST00000428314:c.2349C>G p.Phe783Leu	ENST00000428314:c.6250-1G>A None	comphet	4,2	comphet	maternal&paternal	2	Growth, skeletal	(+) Abnormal thumb morphology,(+) Preaxial hand polydactyly,(+) Vertebral segmentation defect,(+) Pilonidal sinus,(+) Muscular ventricular septal defect,(+) Perimembranous ventricular septal defect
FASTKD3	ENST00000264669:c.1634C>T p.Thr545Ile		de_novo	5,4	het	de novo	2	NDD + Epilepsy	(+) Hemangioma,(+) Seizure,(+) Global developmental delay,(+) Abnormal facial shape,(+) Spastic paraparesis,(+) Abnormality of brain morphology,(+) Cerebral palsy
TIMM8A	ENST00000372902:c.62A>G p.His21Arg		de_novo	7,27	het	de novo		NDD + Epilepsy	(+) Hemangioma,(+) Seizure,(+) Global developmental delay,(+) Abnormal facial shape,(+) Spastic paraparesis,(+) Abnormality of brain morphology,(+) Cerebral palsy

ARPC4	ENST00000397256:c.331C>T p.Arg111Cys		de_novo	6,4	het	de novo	1	NDD	(+) Microcephaly,(+) Hypotonia,(+) Global developmental delay
GSG1L	ENST00000447459:c.184A>G p.Asn62Asp		de_novo	4,6	het	de novo		NDD + Epilepsy	(+) Focal clonic seizure,(+) Dyslexia,(+) Mild global developmental delay,(+) Focal impaired awareness tonic seizure
DIP2C	ENST00000280886:c.2216C>T p.Ala739Val		unknown	4,8	het	unknown	1	NDD	Moderate global developmental delay
BTBD18	NST00000422652:c.1398del p.Tyr467MetfsTer45		unknown	5,4	het	unknown	1	other	Hypotonia,(+) Vocal cord paralysis,(+) Dyspnea
DHX8	ENST00000262415:c.1239A>T p.Lys413Asn		de_novo	B	het	de novo		Metabolism	(+) Inguinal hernia,(+) Jaundice,(+) Cholestasis,(+) Organic aciduria,(+) Hyperbilirubinemia,(+) Elevated circulating alanine aminotransferase concentration
PLXNC1	ENST00000258526:c.3505A>C p.Asn1169His		unknown	3,1	het	unknown	1	NDD	(+) Tall stature,(+) Polyuria,(+) Autism,(+) Hyperactivity,(+) Global developmental delay,(+) Obesity,(+) Polydipsia
TAOK2	ST00000308893:c.2811dup p.Cys938LeufsTer56		unknown	7,2	het	unknown	2	NDD	(+) Autism,(+) Delayed speech and language development,(+) Absent speech,(+) Sleep-wake cycle disturbance,(+) Toe walking
HMX3	ENST00000357878:c.1031C>A p.Ser344Ter		unknown	5,9	het	unknown	2	NDD	(+) Autism,(+) Delayed speech and language development,(+) Absent speech,(+) Sleep-wake cycle disturbance,(+) Toe walking
LRP8	ENST00000306052:c.497-1G>C None		unknown	8,5	het	unknown	1	NDD	(+) Autism,(+) Delayed speech and language development,(+) Developmental regression,(+) Mild global developmental delay
STAM	ENST00000377524:c.265del p.Ser89AlafsTer6		unknown	7,7	het	unknown	1	Epilepsy	(-) Intellectual disability,(+) Focal-onset seizure
RBBP7	NST00000380084:c.89_99del p.His30ProfsTer15		unknown	7,1	het	unknown	2	Epilepsy	(+)atypical absence seizure
KCNH5	ENST00000322893:c.523G>A p.Val175Ile		unknown	4,3	het	unknown	2	Epilepsy	(+)atypical absence seizure
ROCK2	ENST00000315872:c.1598-1G>A None		unknown	6,9	het	unknown	1	NDD	(+) Long philtrum,(+) Carious teeth,(+) Global developmental delay,(+) Abnormal facial shape,(+) Nephroblastoma,(+) Prominent forehead,(+) Abnormal eating behavior
NAP1L2	ENST00000373517:c.700G>T p.Glu234Ter		unknown	5,0	het	unknown	1	Epilepsy	(+) focal myoclonic seizure (+) generalzied tonic-clonic seizure with focal onset
MAGEB5	ENST00000602297:c.770dup p.Tyr257Ter		unknown	4,0	hom	unknown	1	Epilepsy	(+) Absence seizures
HDAC3	ENST00000305264:c.1076G>A p.Arg359His		unknown	5,7	het	unknown	2	NDD	(+) Autistic behavior,(+) Moderate global developmental delay
OTOP1	ENST00000296358:c.803A>G p.Tyr268Cys		homo	5,3	hom	unknown	2	NDD	(+) Autistic behavior,(+) Moderate global developmental delay
UNC13A	ST00000519716:c.1597-4_1597-3delinsAA None		de_novo	7,1	het	de novo	1	NDD + Epilepsy	(+) Hydrocephalus,(+) Decreased response to growth hormone stimuation test,(+) Seizure,(+) Cerebral hemorrhage,(+) Premature birth,(+) Intellectual disability, moderate,(+) Scoliosis,(+) Lymphoma,(+) Immunodeficiency,(+) Short stature,(+) Moderate global
LAMTOR1	ENST00000278671:c.3G>T p.Met1?		unknown	5,9	het	unknown	2	NDD	(+) Delayed puberty,(+) Obesity,(+) Moderate global developmental delay
SUSD4	ENST00000343846:c.26A>G p.Asn9Ser		de_novo	4,8	het	de novo	6	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
CWC22	ENST00000410053:c.1633C>T p.Arg545Ter		de_novo	7,9	het	de novo	6	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
PTPRN	ENST00000295718:c.2766C p.Ile922Met	ENST00000295718:c.2766C>G p.Ile922Met	comphet	4,1	comphet	maternal&paternal	6	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
KIAA0947	ENST00000296564:c.1718C>T p.Thr573Ile	ENST00000296564:c.6464A>G p.His2155Arg	comphet	3,6	comphet	maternal&paternal	6	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
CASKIN1	ENST00000343516:c.1709T>C p.Ile570Thr	ENST00000343516:c.246C>T p.Gly82=	comphet	5,5	comphet	maternal&paternal	6	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
TENT4A	ENST00000230859:c.398C>G p.(Ser133Cys)		homo	4,8	homo	unknown	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
RASSF10	ENST00000340901:c.899A>C p.(Glu300Ala)		homo	3,4	homo	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
KLHL36	ENST00000564996:c.169G>C p.Val57Leu		homo	4,2	homo	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
KIAA0100	ENST00000528896:c.5345G>A p.Gly1782Glu		homo	5,4	homo	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
CPD	ENST00000225719:c.691G>A p.Ala231Thr		homo	4,5	homo	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
SLFN13	ENST00000285013:c.2666C>A p.Ala889Glu		homo	4,4	homo	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
MICALL2	ENST00000297508:c.1336G>A p.Asp446Asn	ENST00000297508:c.1987C>T p.Arg663Cys	comphet	3,5	comphet	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
SF3A2	ENST00000221494:c.1354G>T p.Glu452Ter		unknown	5,0	het	unknown	1	Epilepsy	(+) Bilateral tonic-clonic seizure,(+) Generalized-onset seizure
LRP1B	ENST00000389484:c.7366G>A p.Val2456Ile		homo	5,9	hom	maternal&paternal	1	NDD + Epilepsy	(+) Hypermetropia,(+) Autism,(+) Intellectual disability,(+) Seizure
MAP4K4	ENST00000347699:c.123+2T>C None		unknown	7,3	het	unknown	2	NDD	(+) Delayed speech and language development,(+) Global developmental delay,(+) Motor tics,(+) Phonic tics
TFDP2	ENST00000489671:c.44_47del p.Val15GlufsTer4		unknown	5,2	het	unknown	2	NDD	(+) Delayed speech and language development,(+) Global developmental delay,(+) Motor tics,(+) Phonic tics
RBL2	ENST00000262133:c.3G>T p.Met1?		unknown	7,3	het	unknown	2	NDD	(+) Strabismus,(+) Autistic behavior,(+) Hypotonia,(+) High myopia,(+) Mild global developmental delay
BAI3	ENST00000370598:c.1516C>T p.Arg506Ter		unknown	6,0	het	unknown	1	NDD	(+) Intellectual disability,(+) Moderate global developmental delay, large ears, synophris, downslanted palprebal fissures
MINK1	ENST00000355280:c.3199C>T p.His1067Tyr		unknown	4,2	het	unknown	1	NDD + Epilepsy	(+) Psychosis,(+) Intellectual disability,(+) Focal tonic seizure,(+) Focal hyperkinetic seizure,(+) Focal cortical dysplasia
PDS5A	ENST00000303538:c.1231C>T p.Arg411Trp		de_novo	8,1	het	de novo	2	Growth, skeletal	(+) Retrognathia,(+) Epicanthus,(+) Protruding ear,(+) Hypotonia,(+) Short stature
GPHN	000478722:c.1332_1346del p.His445_Ser449del		unknown	6,4	het	unknown		NDD + Epilepsy	(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Mild global developmental delay,(+) Bilateral tonic-clonic seizure with generalized onset

PLXNB2	ENST00000449103:c.5455C>A p.Gln1819Lys		unknown	**5,1**	het	unknown		NDD + Epilepsy	(+) Microcephaly,(+) Abnormality of the face,(+) Behavioral abnormality,(+) Intellectual disability, mild
XPO7	ENST00000252512:c.1994G>A p.Arg665Gln		unknown	**3,6**	het	unknown	1	NDD + Epilepsy	(+) Intellectual disability,(+) Hemiplegia,(+) Elevated circulating creatine kinase concentration,(+) Severe global developmental delay,(+) Infantile spasms,(+) Eyelid laxity
ACTN1	ENST00000394419:c.1870C>T p.Arg624Ter		unknown	**6,6**	het	unknown	2	NDD	(+) Delayed puberty,(+) Obesity,(+) Moderate global developmental delay
TCF7L2	ENST00000543371:c.407C>T p.Ala136Val		de_novo	**8,5**	het	de novo	1	NDD + Epilepsy	(+) Hypotonia,(+) Motor delay,(+) Dystonia,(+) Generalized-onset seizure,(+) Severe global developmental delay
BTAF1	ENST00000265990:c.4437T>A p.Ser1479Arg		unknown	**4,7**	het	unknown		NDD + Epilepsy	(+) Intellectual disability, mild,(+) Bilateral tonic-clonic seizure,(+) Focal myoclonic seizure,(+) Mild global developmental delay
ZNF827	ENST00000379448:c.292C>T p.Gln98Ter		unknown	**5,6**	het	unknown	1	NDD	(+) Microcephaly,(+) Global developmental delay,(+) Short stature
MRP63	ENST00000309594:c.-5-2A>G None		homo	**8,9**	homo	unknown	2	Epilepsy	(+) Generalized-onset seizure
SMG1	ENST00000446231:c.5213A>T p.Asp1738Val		unknown	**5,8**	het	unknown	2	Epilepsy	(+) Generalized-onset seizure
CLUH	ENST00000570628:c.1654A>T p.Lys552Ter		unknown	**B**	het	unknwon	1	Metabolism	(+) Tall stature,(+) Precocious puberty,(+) Obesity,(+) Hypertriglyceridemia,(+) Accelerated skeletal maturation
SEMA3F	ENST00000002829:c.1093G>A p.Val365Met		de_novo	**7,6**	het	de_novo	4	Muscle	Motor delay, Muscular hypotonia, Skeletal muscle atrophy
ADCY9	ENST00000294016:c.2727C>G p.Tyr909Ter		unknown	**5,8**	het	unknown	1	NDD + Epilepsy	(+) Intellectual disability,(+) Seizure,(+) Dystonia,(+) Severe global developmental delay
RAB11FIP3	ENST00000262305:c.1116-2A>G None		unknown	**6,0**	het	unknown	1	Epilepsy	Generalized non-motor (absence) seizure
GPC1	ENST00000264039:c.1268+4G>A None		de_novo	**5,2**	het	de novo	3	NDD	Trigonocephaly, Epicanthus,Hypertelorism,Short chin, Retinal coloboma, Astigmatism, Hypermetropia, Iris coloboma, Motor delay, Patent foramen ovale, EEG abnormality, Depressed nasal bridge, Vertical nystagmus, Perimembranous ventricular septal defect, Anisometropia
SGK223	ST00000520004:c.3247del p.Gln1083ArgfsTer52		de_novo	**5,0**	het	de novo	3	NDD	Trigonocephaly, Epicanthus,Hypertelorism,Short chin, Retinal coloboma, Astigmatism, Hypermetropia, Iris coloboma, Motor delay, Patent foramen ovale, EEG abnormality, Depressed nasal bridge, Vertical nystagmus, Perimembranous ventricular septal defect, Anisometropia
CHAF1A	ENST00000301280:c.829G>T p.Glu277Ter		de_novo	**10,1**	het	de novo	3	NDD	Trigonocephaly, Epicanthus,Hypertelorism,Short chin, Retinal coloboma, Astigmatism, Hypermetropia, Iris coloboma, Motor delay, Patent foramen ovale, EEG abnormality, Depressed nasal bridge, Vertical nystagmus, Perimembranous ventricular septal defect, Anisometropia
TSC22D4	ENST00000300181:c.1A>G p.Met1?		unknown	**5,1**	het	unknown		NDD	(+) Abnormality of the face,(+) Intellectual disability,(+) Short stature,(+) Moderate global developmental delay,(+) Primary microcephaly
GPR115	ENST00000283303:c.1860del p.Phe620LeufsTer3		homo	**8,5**	homo	unknown	1	NDD + Epilepsy	(+) Hypothyroidism,(+) Seizure,(+) Intellectual disability, mild,(+) Global developmental delay,(+) Gliosis
PHF21B	ENST00000313237:c.53A>C p.Gln18Pro		de_novo	**5,6**	het	de novo	1	NDD	hypertelorism, pointed chin, behavioral disorder (no acceptance of rules, aggressive behaviour, urge to move), global developmental delay, poor speech (single words/2-word combinations), known heterozygous deletion in 10p15.3, incl. DIP2C-gene, inherited maternal; deletion in 16p12.2
TUBA1B	ENST00000336023:c.362G>A p.Arg121Gln		unknown	**4,9**	het	unknown	3	NDD + Epilepsy	(+) Intellectual disability,(+) Generalized non-motor (absence) seizure,(+) Moderate global developmental delay
CHD9	ENST00000566029:c.4967G>C p.Ser1656Thr		unknown	**4,4**	het	unknown	3	NDD + Epilepsy	(+) Intellectual disability,(+) Generalized non-motor (absence) seizure,(+) Moderate global developmental delay
XKR3	ENST00000331428:c.614T>A p.Leu205Ter		homo	**7,0**	het	unknown	3	NDD + Epilepsy	(+) Intellectual disability,(+) Generalized non-motor (absence) seizure,(+) Moderate global developmental delay
CUL2	ENST00000537177:c.571G>C p.Val191Leu		unknown	**4,4**	het	unknown	1	Epilepsy	Focal-onset seizure
DAPK1	ENST00000408954:c.2980G>A p.Asp994Asn		unknown	**3,7**	het	unknown		NDD	(+) Intellectual disability,(+) Obesity
TRA2B	ENST00000453386:c.151A>G p.Arg51Gly		unknown	**5,0**	het	unknown	1	NDD	(+) Psychosis,(+) Intellectual disability, mild
TRA2B	ENST00000453386:c.151A>G p.Arg51Gly		unknown	**5,0**	het	unknown	1	NDD	(+) Downslanted palpebral fissures,(+) Autism,(+) Global developmental delay
RALGPS1	ENST00000259351:c.1544C>A p.Pro515His		unknown	**3,2**	het	unknown		NDD + Epilepsy	(+) Aggressive behavior,(+) Focal clonic seizure,(+) Expressive language delay,(+) Focal tonic seizure,(+) Severe global developmental delay,(+) Focal atonic seizure,(+) Impulsivity
TRA2B	T00000453386:c.266_280del p.Asp90_Tyr94del		de_novo	**6,9**	het	de novo	1	NDD + Epilepsy	(+) Microcephaly,(+) Delayed speech and language development,(+) Hypotonia,(+) Status epilepticus,(+) Generalized tonic seizure,(+) Atonic seizure
UBE2Q1	ENST00000292211:c.946C>G p.Leu316Val		unknown	**3,3**	het	unknown		NDD + Epilepsy	(+) Intellectual disability, mild,(+) Generalized myoclonic-atonic seizure
CHD9	NST00000566029:c.7499_7501del p.Gly2500del		unknown	**3,5**	het	unknown	4	NDD	(+) Hearing impairment,(+) Obesity,(+) Mild global developmental delay
FBXL19	ENST00000380310:c.431G>C p.Arg144Pro		unknown	**3,0**	het	unknown	4	NDD	(+) Hearing impairment,(+) Obesity,(+) Mild global developmental delay
BRPF3	ENST00000357641:c.2228A>C p.Glu743Ala		unknown	**4,0**	het	unknown	4	NDD	(+) Hearing impairment,(+) Obesity,(+) Mild global developmental delay
GMPPB	NST00000321599:c.764_765delinsTT p.Thr255Ile		unknown	**4,3**	het	unknown	4	NDD	(+) Hearing impairment,(+) Obesity,(+) Mild global developmental delay
TNRC6A	00395799:c.4677_4680del p.Trp1559CysfsTer30		unknown	**7,5**	het	unknown	1	NDD	(+) Autism,(+) Impaired social interactions,(+) Obesity,(+) Moderate global developmental delay
SEC24A	ENST00000398844:c.1642A>G p.Thr548Ala		de_novo	**5,9**	het	de novo	2	NDD	microcephaly, congenital diaphragmatic hernia, pectus excavatum of inferior sternum, motor delay, failure to thrive in infancy, patent ductus arteriosus mild global developmental delay

CUL1	ENST00000325222:c.2137G>A p.Ala713Thr		de_novo	8,2	het	de novo	2	NDD	microcephaly, congenital diaphragmatic hernia, pectus excavatum of inferior sternum, motor delay, failure to thrive in infancy, patent ductus arteriosus, mild global developmental delay
CLOCK	ST00000309964:c.1599dup p.Thr534AspfsTer55		unknown	7,9	het	unknown	1	NDD	(-) Microcephaly,(+) Delayed speech and language development,(-) Seizure,(+) Global developmental delay,(+) Motor delay,(+) Muscular hypotonia of the trunk
ZNF611	ENST00000543227:c.1319C>T p.Ser440Phe		de_novo	3,5	het	de novo	1	NDD	Aggressive behavior, Global developmental delay, Developmental regression, Self-injurious behavior
RAB11A	ENST00000261890:c.-23A>G p.His112Arg		unknown	8,8	het	unknown	1	NDD + Epilepsy	(+) Coarse facial features,(+) Delayed speech and language development,(+) Intellectual disability, mild,(+) EEG abnormality,(+) Precocious puberty in females,(+) Delayed fine motor development,(+) Primary microcephaly
CT47B1	ENST00000371311:c.622C>T p.Pro208Ser		de_novo	4,2	het	de novo	1	NDD + Epilepsy	osteopenia, intellectual disability, seizure, global developmental delay
SNW1	ST00000261531:c.182_187del p.Gly61_Gly62del		de_novo	5,9	het	de novo	2	NDD + Epilepsy	microcephaly, visual impairment, delayed speech and language development, anemia, bilateral tonic-clonic seizure, abnormal cortical gyration, hip dislocation, thoracolumbar scoliosis, focal-onset seizure, intellectual disability, severe, cerebral palsy
ZNF768	ENST00000380412:c.1511A>G p.His504Arg		de_novo	5,4	het	de novo	2	NDD + Epilepsy	microcephaly, visual impairment, delayed speech and language development, anemia, bilateral tonic-clonic seizure, abnormal cortical gyration, hip dislocation, thoracolumbar scoliosis, focal-onset seizure, intellectual disability, severe, cerebral palsy
TNPO1	ENST00000337273:c.2438G>C p.Arg813Thr		unknown	3,5	het	unknown	1	Growth, skeletal	(+) Renal duplication,(+) Cleft palate,(+) Abnormality of the ribs,(+) Glandular hypospadias,(+) Atopic dermatitis,(+) Premature birth,(+) Neutropenia,(+) Scoliosis,(+) Cleft
WDR13	ENST00000218056:c.194G>A p.Arg65His		hemi	5,2	hemi	maternal	3	NDD	(+) Hearing impairment,(+) Abnormality of refraction,(+) Seizure,(+) Hypotonia,(+) Global developmental delay,(+) Motor delay,(+) Holoprosencephaly,(+) Failure to thrive,(+) Muscular dystrophy,(+) Abnormality of temperature regulation,(+) Secondary microcephaly,(+) Bilateral cryptorchidism
RBM10	ENST00000377604:c.308G>A p.Arg103Gln		hemi	7,0	hemi	maternal	3	NDD	(+) Hearing impairment,(+) Abnormality of refraction,(+) Seizure,(+) Hypotonia,(+) Global developmental delay,(+) Motor delay,(+) Holoprosencephaly,(+) Failure to thrive,(+) Muscular dystrophy,(+) Abnormality of temperature regulation,(+) Secondary microcephaly,(+) Bilateral cryptorchidism
CCAR2	ENST00000308511:c.2627G>C p.Arg876Pro		de_novo	6,1	het	de novo	3	NDD	(+) Hearing impairment,(+) Abnormality of refraction,(+) Seizure,(+) Hypotonia,(+) Global developmental delay,(+) Motor delay,(+) Holoprosencephaly,(+) Failure to thrive,(+) Muscular dystrophy,(+) Abnormality of temperature regulation,(+) Secondary microcephaly,(+) Bilateral cryptorchidism
DBN1	3_1334insGCCACGGAGATCC p.Ala445GlyfsTer13		unknown	7,9	het	unknown	1	NDD	(+) Obesity,(+) Intellectual disability, borderline
INTS6	ENST00000420668:c.498C>G p.Tyr166Ter		de_novo	9,9	het	de novo	1	NDD	(+) Global developmental delay,(+) Motor delay,(+) Agenesis of corpus callosum,(+) Morphological central nervous system abnormality,(+) Cerebellar dysplasia,(+) Muscular hypotonia of the trunk,(+) Schizencephaly,(+) Abnormal nervous system morphology,(+) Abnormal subarachnoid space morphology,(+) Interhemispheric cyst,(+) Paroxysmal tonic upgaze
TSPAN18	ENST00000340160:c.275T>C p.Leu92Pro		homo	5,2	homo	unknown	2	NDD	(+) Behavioral abnormality,(+) Global developmental delay,(+) Intellectual disability, borderline
NOVA2	ENST00000263257:c.571A>G p.Lys191Glu		unknown	4,8	het	unknown	2	NDD	(+) Behavioral abnormality,(+) Global developmental delay,(+) Intellectual disability, borderline
SLC17A7	ENST00000221485:c.170T>C p.Phe57Ser		unknown	7,5	het	unknown	1	Epilepsy + ataxia	(+) Generalized myoclonic seizure,(+) Episodic ataxia,(+) Generalized tonic seizure,(+) Generalized clonic seizure
NSD1	ENST00000347982:c.5468C>T p.Thr1823Met		unknown	B	het	unknown	1	Obesity	(+) Tall stature,(+) Precocious puberty,(+) Obesity,(+) Hypertriglyceridemia,(+) Accelerated skeletal maturation
DUSP26	ENST00000256261:c.56G>T p.Arg19Leu		de_novo	6,1	het	de novo	2	Epilepsy	epilepsy with focal and generalized components, microcephaly, MRI unremarkable, psychosomatic development unremarkable, seizure-free under Sultiam, EEG with rolando-focus and generalization tendency
B4GALNT4	ENST00000329962:c.2232C>G p.Asn744Lys		de_novo	5,3	het	de novo	2	Epilepsy	epilepsy with focal and generalized components, microcephaly, MRI unremarkable, psychosomatic development unremarkable, seizure-free under Sultiam, EEG with rolando-focus and generalization tendency
ARMCX4	ENST00000423738:c.2150A>G p.Gln717Arg		x_linked	4,0	hemi	unknown	1	NDD + Epilepsy	(+) Intellectual disability,(+) Seizure,(+) Global developmental delay
DENND1C	ENST00000381480:c.1241C>T p.Ala414Val		de_novo	B	het	de novo	1	Growth, skeletal	bei U3 auffällige Kopfform festgestellt, Sagittalnahtsynostose, keine neurologischen Auffälligkeiten
GPN1	ENST00000264718:c.982T>A p.Ser328Thr		de_novo	4,8	het	de novo	2	Epilepsy	bilateral tonic-clonic seizure with generalized onset
TNKS2	ENST00000371627:c.1901A>G p.Asp634Gly		de_novo	7,1	het	de novo	2	Epilepsy	bilateral tonic-clonic seizure with generalized onset
PITRM1	ENST00000224949:c.2263C>T p.(Arg755Trp)		de_novo	B	het	de novo	1	Cardio	(+) Dilated cardiomyopathy,(+) Abnormal left ventricle morphology,(+) Primum atrial septal defect,(+) Multiple muscular ventricular septal defects
DPP6	ENST00000332007:c.1075A>C p.Lys359Gln		de_novo	9,4	het	de novo	1	Epilepsy	(+) Abnormality of the pinna,(+) Generalized non-motor (absence) seizure,(+) Focal clonic seizure,(+) 2-3 toe syndactyly,(+) Focal tonic seizure
ZFP36	NST00000248673:c.708del p.Gly237AlafsTer129		unknown	5,8	het	unknown	1	Epilepsy	(+) Strabismus,(-) Global developmental delay,(+) Generalized non-motor (absence) seizure,(+) Status epilepticus,(+) Focal-onset seizure,(+) EEG with focal spikes,(+) EEG with focal spike waves

ITGB1	ENST00000302278:c.1844G>A p.Cys615Tyr		de_novo	**A**	het	de novo	1	Cardio	(+) Gliosis,(+) Cerebral ischemia,(+) Cerebral vasculitis,(+) Perivascular spaces,(+) Arterial stenosis
DAGLA	ENST00000257215:c.2613dup p.Ser872GlnfsTer6		de_novo	**A**	het	de novo	1	Neuro	abnormality of eye movement, ataxia
PLXNA2	ENST00000367033:c.2594C>T p.Thr865Met		homo	**8,0**	homo	unknown	1	NDD + Epilepsy	(+) Microcephaly,(+) Global developmental delay,(+) Encephalopathy,(+) Increased body weight,(+) Febrile status epilepticus