High AutoCaSc value suggest that the gene-variant combination is more plausible (applies only for cases of NDD and for some cases of neurological disorders). If the number of candidates in one family is high, the plausibility of each single gene decreases a bit. If you have questions or are interested in cooperation, contact rami.aboujamra@medizin.uni-leipzig.de

HGNC symbol	Variant 1	Variant 2	Auto CaSc	# candidates in family	Disease group / leading symptom	HPO-Main-Terms	Zygosity	Origin	Family history
SPTAN1	NM_001130438.2: c.2612del p.(Lys871Serfs*5)		13.4	4	NDD + Epilepsy	Intellectual disability,Global developmental delay, Motor delay, Developmental regression	het	de novo	no
STX1A	NM_004603.3: c.284-1G>A p.?		12.9	1	NDD	severe ID, decreased fetal movements, muscular hypotonia	homo	paternal & maternal	2m, cousins I°
HSPD1	NM_002156.4: c.1394_1406del p.(Ile465Lysfs*9)		12.8	1	Neuro	Hypogonadotrophic hypogonadism, Tall stature, Psychosis, Depression, Psychotic episodes, Dementia, Overgrowth, Neurodegeneration, Bipolar affective disorder, Brain atrophy	het	de novo	no
PIK3CA	NM_006218.3:c.1A>G p.0?		12.7	1	NDD	Mental retardation, Excessive tallness, Macrocephaly	het	de novo	no
RORB	Del (NM_006914.3): 9:76768311-77229880		12.7	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	het	de novo	no
ANK2	NM_001148.4:c.1288-1G>A p.?		12.4	3	NDD + Epilepsy	neonatal epileptic encephalopathy	het	de novo	no
KMT2E	NM_018682.3: c.3554C>G p.(Ser1185*)		12.4	1	NDD	Intellectual disability, Seizures, EEG with spike-wave complexes, EEG with continuous slow activity,	het	de novo	no
GLS	NM_001256310.1: c.695dup p.(Asp232Glufs*2)		12.4	1	NDD + Epilepsy	Seizures, Status epilepticus, Infantile onset, Infantile spasms, Epileptic encephalopathy	homo	paternal & maternal	yes, one affected sister, consanguinity
UNC13A	NM_001080421.2: c.3215+1G>C p.?		12.3	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	het	de novo	no

CUX1	NM_001202543.1: c.3783_3784dup p.(Leu1262Argfs*10)	12.1	1	NDD	Macrocephaly, Umbilical hernia, Chronic constipation, Inguinal hernia, Delayed speech and language development, mild global developmental delay	het	de novo	no
DLGAP1	NM_004746.3:c.101 8C>T p.(Arg340*)	11.8	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech	het	de novo	no
RYR2	NM_001035.3: c.6202C>T p.(Arg2068*)	11.5	2	NDD + epilepsy	(+) Dravet syndrome,(+) Seizure,(+) Myoclonic seizure,(+) Myoclonic absence seizure,(+) Global developmental delay,(+) Intellectual disability	het	de novo	Cousin of the father with epilepsy
KDM5A	NM_001042603.2: c.4048C>T p.(Arg1350*)	11.5	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	het	de novo	no
TANC2	NM_025185.3: c.4405del p.(Arg1469Glyfs*6)	11.4	1	NDD + Epilepsy	Seizures, Global developmental delay, Encephalopathy, Epileptic encephalopathy	het	de novo	no
ZNF804A	NM_194250.1: c.1049del p.(Gly350Valfs*7)	11.2	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	homo	maternal & unknown	yes, two siblings with intellectual impairment
CLCN3	NM_173872.3: c.336_339del p.(Lys112Asnfs*6)	11.1	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	homo	paternal & maternal	yes, two similar affected brothers, one deceased
PABPC1	NM_002568.3: c.1691A>G p.(Glu564Gly)	11.0	1	NDD + Epilepsy	global developmental delay, seizures, visual impairment, bicuspid aortic valve	het	de novo	no

CACNA1B	NM_000718.3: c.1442C>T p.(Ala481Val)		11.0	2	Epilepsy	Seizures, Global developmental delay, Hypoglycorrhachia, Hyperglycorrhachia	het	de novo	no
MAPK8IP3	NM_001040439.1: c.1556G>A p.(Arg519Gln)		10.9	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormality of body weight, Increased body weight,	het	de novo	yes,paternal uncle went to special school
PUM2	NM_015317.2:c.2216del p.(His739Leufs*10)		10.9	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	het	unknown	no
GLS	NM_001256310.1: c.815G>A p.(Arg272Lys)	NM_001256310.1:c.241C>T p.(Gln81*)	10.7	1	NDD + Epilepsy	Microcephaly, Seizures, Status epilepticus, CNS demyelination, EEG with burst suppression, Peripheral demyelination, Epileptic encephalopathy	comphet	paternal & maternal	yes, one affected sister
ACTL6B	NM_016188.4: c.1027G>A p.(Gly343Arg)		10.7	1	NDD	Muscular hypotonia, Abnormality of mouth shape, Stereotypical hand wringing, Microcephaly, Global developmental delay	het	de novo	no
CACNA1C	NM_199460.3: c.496T>C p.(Phe166Leu)		10.7	2	NDD + Epilepsy	epilepsy with absences and generalized tonic-clonic seizures, severe intellectual disability with autistic traits, low blood pressure, obstipation, normal MRI 2008	het	de novo	yes, 2/5 brothers with ID and epilepsy
TAB2	NM_015093.5:c.1448del p.(Pro483Leufs*16)		10.6	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	het	de novo	no
NRX2	NM_015080.3: c.4484del p.(Phe1495Serfs*71)		10.6	1	NDD	Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Abnormal facial shape, Intellectual disability, moderate	het	unknown	no

ATP1A3	NM_001256214.1: c.6+3A>G p.?	10.5	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	homo	paternal & maternal	yes, sister is also affected
NKTR	NM_005385.4: c.3076del p.(Glu1026Argfs*26)	10.4	1	Epilepsy	Myoclonic spasms, Seizure, EEG abnormality	het	de novo	a fathers cousins two children with epilepsy in childhood
NSD2	NM_001042424.2: c.3295G>A p.(Glu1099Lys)	10.3	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	het	de novo	no
ZFYVE26	NM_015346.3: c.5779T>A p.(Tyr1927Asn)	10.3	1	NDD	Global developmental delay, Absent speech, Proportionate short stature, Short stature	het	de novo	no
JPH4	NM_001146028.1: c.953_956del p.(Gly318Alafs*53)	10.2	1	NDD	Microcephaly, Autism, Intellectual disability, Muscular hypotonia, Global developmental delay	het	de novo	yes, CAVE: halfbrother affected and not sequenced. XR is thus
ASIC1	NM_001095.3: c.363-2A>G p.?	10.2	3	NDD + Epilepsy	Seizures, Abnormal social behavior, Epileptic encephalopathy	het	de novo	no
TNRC18	NM_001080495.2: c.7518dup p.(Ala2507Argfs*44)	10.2	1	NDD	Microcephaly, Intellectual disability, Global developmental delay, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay	het	de novo	no
ARRB2	NM_001257328.1: c.684+1G>C p.?	10.2	1	Epilepsy	autism-spectre disorder, focal- onset epilepsy	het	de novo	no
PHLPP1	NM_194449.3: c.3756-2A>G p.?	10.2	3	Epilepsie	therapy-resistant epilepsy	het	de novo	no

DDX42	NM_007372.3: c.221+1G>A p.?	10.2	1	NDD + Epilepsy	Epilepsy, optic atrophy, diabetes insipidus and hypothyroidism	het	de novo	no
KDM5B	NM_006618.4: c.1286T>G p.(Ile429Ser)	10.1	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	het	de novo	no
GRIA4	NM_000829.3: c.2090G>C p.(Arg697Pro)	10.1	1	NDD	Delayed speech and language development, Intellectual disability, Poor speech	het	de novo	no
EIF5B	NM_015904.3: c.3607C>T p.(Gln1203*)	10.1	1	NDD	Macrocephaly, Autism, Intellectual disability, Absent speech, Intellectual disability, severe	het	de novo	no
EGR3	NM_001199880.1: c.477C>A p.(Tyr159*)	10.1	1	NDD	Intellectual disability, learning disability	het	de novo	no
GABBR1	NM_001470.3:c.1190C>T p.(Ala397Val)	10.1	2	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Global developmental delay, Motor delay, Encopresis	het	de novo	no
TRAP1	NM_016292.2: c.1941-1G>A p.?	10.0	1	NDD	moderate ID, mental deterioration, autism, self-mutilation, muscular hypotonia, nystagmus, leukodystrophy	homo	paternal & maternal	1m, cousins 2°
ETV5	NM_004454.2: c.232+1G>A p.?	10.0	4	NDD	global developmental delay, intellectual disability, generalized hypotonia,	het	de novo	no
WDFY3	NM_014991.4: c.749A>G p.(Asn250Ser)	10.0	2	Epilepsy	seizures, peripheral axonal neuropathy, motor delay, gait disturbance, EEG with focal epilepti-form discharges	het	de novo	yes, father with autistic features, father and three siblings with ADHD, one brother with Rolandic epilepsy

TOB1	NM_001243877.1: c.888_907delTAACC TCAGTCCTCTCCAGTi nsGGG p.(Leu296Leufs*4)	9.9	1	NDD	Cerebral calcification, Seizures, Congenital cataract, Autistic behavior, Obesity, Global developmental delay	het	de novo	no
PAK1	NM_001128620.1: c.1409T>G p.(Leu470Arg)	9.9	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	het	de novo	no
POLR3A	NM_007055.3: c.1096A>G p.(Arg366Gly)	9.9	1	Epilepsy	Seizures, Focal impaired awareness seizure, Hypothalamic hamartoma, Focal-onset seizure, Epileptic spasms, Langerhans cell histiocytosis	het	de novo	no
ANKRD17	NM_032217.4: c.5360_5363del p.(Gln1787Argfs*5)	9.9	1	NDD	Coarse facial features, Muscular hypotonia, Global developmental delay, Motor delay, Hypertonia, Pes planus, Gait ataxia, Limb hypertonia, Muscular hypotonia of the trunk	het	de novo	no
ANKRD17	NM_032217.4: c.3751_3754del p.(Arg1252Thrfs*6)	9.9	1	NDD	Cryptorchidism, Microcephaly, Strabismus, Hypermetropia, Behavioral abnormality, Stereotypy, Global developmental delay, Small for gestational age, Short stature	het	de novo	no
ZMYM2	NM_001190964.2: c.2338C>T p.(Arg780*)	9.9	1	Epilepsy	Seizures, Focal seizures, Generalized myoclonic seizures, Focal myoclonic seizures, Segmental myoclonic seizures, Falls, Frequent falls	het	de novo	no
AKAP13	NM_006738.5: c.742C>T p.(Arg248*)	9.9	1	NDD + Epilepsy	Seizures, Global developmental delay, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Generalized tonic seizures, Epileptic encephalopathy	het	de novo	no
KIF5C	NM_004522.2: c.2385dup p.(Gln796Alafs*19)	9.9	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	het	unknown	yes, older brother with epilepsy until the age of 3, grandfather (paternal) with epilepsy in childhood

EFHC1	NM_018100.3: c.323del p.(Pro108Leufs*13)	9.9	3	NDD	Microcephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe	homo	paternal & maternal	no
RFX3	NM_001282116.1: c.115C>T p.(Gln39*)	9.8		NDD	(+) Global developmental delay,(+) Poor coordination,(+) Large for gestational age,(+) Overgrowth,(+) Tall stature,(+) Ataxia,(+) Muscular hypotonia,(+) Behavioral abnormality	het	de novo	no
ZNF384	NM_001135734.2: c.459del p.(Gly154Alafs*15)	9.8	2	NDD	(+) Global developmental delay,(+) Scotoma,(+) Intellectual disability, mild,(+) Intellectual disability, borderline,(+) Myopia,(+) Depressivity,(+) Anxiety,(+) Motor delay,(+) Retinal atrophy	het	de novo	no
RASGRP1	NM_001128602.1: c.1487C>G p.(Ser496*)	9.8	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures	het	de novo	no
MRTFA	NM_001318139.2: c.800del p.(Lys267Argfs*2)	9.8	3	Epilepsy	Suspected vitamin-B6-dependent epilepsy, Generalized-onset epileptic spasm, Atypical absence seizure, EEG abnormality	het	de novo	no
EZH2	NM_001203247.1: c.2197G>A p.(Ala733Thr)	9.8	3	NDD	Microcephaly, Delayed speech and language development, Global developmental delay, Abnormal facial shape, Severe short stature, Short stature, Intellectual disability	het	de novo	no
H3-3A	NM_002107.4: c.250C>G p.(Arg84Gly)	9.8	2	NDD + Epilepsy	Stereotypy, Delayed speech and language development, Global developmental delay, Motor delay, Delayed gross motor development, EEG abnormality, Delayed fine motor development	het	de novo	no
BAZ1B	NM_032408.3: c.461G>A p.(Gly154Asp)	9.7	1	Epilepsy	absence epilepsy, EEG abnormality	het	de novo	yes, mother with absence epilepsy, maternal grandfather with epilepsy
RASA2	NM_006506.3: c.1591-2A>G p.?	9.7		NDD	(+) Periventricular leukomalacia,(+) Global developmental delay,(+) Cerebral palsy,(+) Elevated hepatic transaminase,(+) Muscular hypotonia,(+) Small for gestational age	het	de novo	no

CCAR2	NM_021174.5: c.2484C>A p.Tyr828*	9.7	2	NDD	moderate ID, small for gestational age, short stature	homo	paternal & maternal	2fm, >cousins I°
FRYL	NM_015030.1: c.1224del p.(Lys409Argfs*15)	9.7	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	het	de novo	no
SMURF1	NM_020429.2:c.139 0C>T p.(Gln464*)	9.6	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	het	de novo	no
MADD	NM_001135943.1: c.1037T>C p.(Leu346Pro)	9.6	3	NDD	global developmental delay, absent speech, gait disturbance, EEG abnormality, decreased body weight	homo	paternal & maternal	yes, one affected brother
GNAI1	NM_002069.5: c.143C>A p.(Thr48Lys)	9.5	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, Cognitive impairment	het	de novo	no
PUM1	NM_001020658.1: c.3439C>T p.(Arg1147Trp)	9.5	2	NDD	Global developmental delay, Microcephaly, Cryptorchidism, Ptosis, Short stature, Short phalanx of finger, Frontal hirsutism, Arachnoid cyst	het	de novo	no

CACNB4	NM_000726.3: c.848C>T p.(Ser283Leu)		9.5	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures	het	de novo	no
SPART	NM_001142294.1:c.1655T>G p.(Val552Gly)	NM_001142294.1:c.848C>T p.(Ser283Phe)	9.4	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology	comphet	paternal & maternal	yes, maternal aunt and paternal uncle with seizures
NINL	NM_025176.4: c.277+2T>C p.?		9.4	4	NDD	Intellectual disability, Global developmental delay	homo	paternal & maternal	no
BDP1	NM_018429.2: c.6847G>T p.(Glu2283*)		9.4	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	homo	paternal & maternal	no
GALNT2	NM_004481.4: c.865C>T p.Gln289*		9.4	1	NDD + Epilepsy	very severe ID, seizures, autism, aggressive behavior, feeding problems in infancy, short stature, constipation, strabismus, inguinal hernia	homo	paternal & maternal	yes, (two or three affected siblings, parents cousins I°, however possibly different phenotypes)
ICE2	NM_024611.5: c.2764G>T p.Gly922*		9.4	1	NDD + Epilepsy	mild ID, deafness, febrile seizures, EEG abnormalities, atrial septal defect	homo	paternal & maternal	1f, cousins 1°

KDM2A	NM_012308.2: c.956G>A p.(Arg319Gln)		9.4	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 dysphagia	het	de novo	no
MAPKAPK2	NM_004759.4: c.445C>T p.(Arg149*)		9.3	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	het	de novo	no
DOCK3	NM_004947.4: c.1175G>A p.(Arg392Gln)	NM_004947.4:c.3740T>C p.(Met1247Thr)	9.3	1	NDD + Epilepsy	Seizures, Global developmental delay	comphet	paternal & maternal	yes, brother is also affected
CPLX1	NM_006651.3: c.250dup p.(Ala84Glyfs*256)		9.3	3	NDD + Epilepsy	Global developmental delay and obsessive-compulsive behavior, seizures	het	unknown	no
INIP	NM_021218.2: c.266delC p.Ala89fs		9.2	1	NDD + Epilepsy	mild ID, febrile seizures, recurrent infections, carious teeth, microcephaly, muscular hypotonia, ataxia, myopia	homo	paternal & maternal	1m, cousins 2°
MINPP1	NM_004897.4: c.75_94del p.(Leu27Argfs*39)		9.2	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Ataxia, Global developmental delay, Gait ataxia, Olivopontocerebellar atrophy, Short stature, Pontocerebellar atrophy, Olivopontocerebellar hypoplasia, Cognitive impairment	homo	paternal & maternal	yes, brother is also affected

EVI5L	NM_001159944.2: c.841del p.(Leu281fs)	9.2	2	NDD + Epilepsy	Pallister Hall syndrome with hand and foot polydactyly, cleft palate, seizures, global developmental delay, 4-5 finger syndactyly, hamartoma	het	de novo	no
PAPOLG	NM_022894.3: c.533C>G p.(Ser178*)	9.2	4	Epilepsy	Seizures, Generalized-onset seizure	het	de novo	no
DPYSL2	NM_001197293.3: c.1859C>T p.(Pro620Leu)	9.1	2	NDD	Cognitive impairment, Global developmental delay, Tall stature, Obesity	het	unknown	no
C16orf70	NM_001320540.1: c.1050+1G>A p.?	9.1	2	NDD	Global developmental delay, short stature, talipes equinovarus	het	de novo	no
PRPF6	NM_012469.3: c.67C>T p.(Arg23Trp)	9.1	1	NDD	Visual impairment, Intellectual disability, Growth delay, Mildly reduced visual acuity, Feeding difficulties	het	de novo	no
SLC44A1	NM_080546.4: c.377_380delGTGA p.Ser126fs	9.1	1	NDD	mild ID, macrocephaly, acanthosis nigricans, accessory mamilla, muscular hypotonia, frontotemporal cerebral atrophy	homo	paternal & maternal	1m, cousins I°
MADD	NM_001135943.1: c.2284G>T p.(Ala762Ser)	9.0	3	NDD + Epilepsy	epileptic encephalopathy, therapy-resistant seizures, pain in legs	homo	paternal & maternal	yes, sister also affected, 1 sister died in the age of 6 months
ABCA2	NM_001606.4: c.2261T>C p.(Phe754Ser)	9.0	3	Epilepsy	epilepsy, febrile seizures	het	de novo	no
PIKFYVE	NM_015040.3: c.1319A>G p.(Gln440Arg)	9.0	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe	homo	paternal & maternal	yes, brother similary affected
ZMYM2	NM_001190964.2: c.2881G>C p.(Glu961Gln)	9.0	2	NDD + Epilepsy	Seizures, Global developmental delay, Episodic ataxia	het	de novo	no
MBNL3	NM_018388.3: c.279delT p.Ala94fs	9.0	1	NDD	moderate ID, autism	homo	paternal & maternal	1m, cousins 2°
SKOR2	NM_001278063.1: c.2752+1G>T p.?	9.0	3	NDD	Short stature, microcephaly, mild intellectual disability, hyperopia	homo	unknown	yes, similary affected sister

CHD1L	NM_004284.4: c.1175G>A p.Arg392His	9.0	1	NDD	mild ID, microcephaly, muscular hypotonia, rigidity, ataxia, intention tremor, hypopigmented macules, EEG abnormalities	homo	paternal & maternal	1f, consanguinity
SF3A3	NM_006802.3: c.1408C>T p.(Arg470*)	8.9	3	Epilepsy	epilepsy with febrile seizures	het	de novo	no
UBR5	NM_015902.5: c.3682C>T p.(Pro1228Ser)	8.9	1	NDD + Epilepsy	Epilepsy associated with fever or infection, tonic-clonic seizures, mild mental retardation, macrocephaly and sleep EEG with sharp slow waves	het	de novo	yes, Son of maternal cousin with epilepsy, uncle with with febrile seizures and maternal macrocephaly
MAGI2	NM_012301.3: c.3780C>A p.Asp1260Glu	8.9	1	NDD	mild ID, hypermetropia	homo	paternal & maternal	3fm, >cousins I°
GDF11	NM_005811.4: c.955dup p.(Thr319Asnfs*5)	8.9	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	het	de novo	yes, sister with absence seizures, may be the same phenotype, so be careful evaluating this variant

CNP	NM_033133.4: c.44A>G p.(Lys15Arg)	8.9	1	NDD	schwere Intelligenzminderung, spricht nur Einzelworte, eingeschränktes Sprachverständnis, körperlich gesund	homo	paternal & maternal	yes, Parents are healthy and consanguin (Cosuin/Cousin e 1,Grades); Sister Esraa: severe intelligence reduction, speech better than Soaad, but muscle weakness, sitting in wheelchair, strabismus, microcephaly; Sister Tabarak (b., 27,09,2015) motor and speech development delay
HCN2	NM_001194.3: c.1120A>C p.(Met374Leu)	8.9	1	NDD + Epilepsy	Microcephaly, delayed speech and language development, intellectual disability, global developmental delay, motor delay, generalized-onset seizure, epileptic spasms, cognitive	het	de novo	no
HSPA8	NM_006597.5: c.98A>G p.(Gln33Arg)	8.9	1	NDD + Epilepsy	seizures, focal seizures, myoclonic seizures	het	de novo	no
NRXN3	NM_001330195.1: c.115C>T p.(Arg39Cys)	8.9	3	NDD	Global developmental delay and severe motor delay, cerebral palsy (GMFCS level 5), hypoplasia of the corpus callosum	homo	paternal & maternal	no
CASP9	NM_001229.4: c.868+5G>C p.?	8.8	2	NDD	Cataract, Peters anomaly, Autism, Global developmental delay	homo	paternal & maternal	yes, one affected brother

TAOK1	NM_020791.2: c.332C>T p.(Ser111Phe)	8.8	1	NDD	Dysmorphic syndrome, cleft lip and palate, failure to thrive, macrocephaly, muscular hypotonia, developmental delay	het	de novo	yes, Father has similar facial dysmorphisms and limitation of joint mobility
AGO2	NM_001164623.1: c.602G>T p.(Gly201Val)	8.8	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	het	de novo	no
DDB1	NM_001923.4: c.563G>A p.(Arg188Gln)	8.8	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 waves	het	de novo	no
ATR	NM_001184.4: c.2419G>A p.(Gly807Arg)	8.8	3	NDD + epilepsy	Global developmental delay, Microcephaly, Seizures	homo	paternal & maternal	no
ATP2B1	NM_001001323.1: c.1376A>G p.(His459Arg)	8.8	3	NDD + Epilepsy	Autism, Intellectual disability, Seizures, Global developmental delay, Poor speech, Focal seizures	het	de novo	no
WARS1	NM_173701.1: c.397C>T p.(Arg133Cys)	8.7	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	homo	paternal & maternal	yes, brother similary affected, highly consanguine family
DOCK4	NM_014705.3:c.2945C>T p.(Thr982Ile)	8.7	1	Epilepsy	tonic-clonic seizures, intelligence impairment, tremor, ataxia	het	de novo	no

BSN	NM_003458.3: c.11163C>G p.(Ser3721Arg)	8.7	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	het	de novo	no
CACNA2D1	NM_000722.3: c.1514C>T p.Thr505Ile	8.7	1	NDD	severe ID, muscular hypotonia, stereotypical motor behaviors, inguinal hernia, omphalocele	homo	paternal & maternal	1m, consanguinity
NPTN	NM_012428.3:c.1025C>T p.(Pro342Leu)	8.7	2	NDD	Autism, Delayed speech and language development, Intellectual disability, Global developmental delay, Diarrhea, Macrocephaly, Partial Epilepsy	het	de novo	no
SENP3	NM_015670.5: c.713C>A p.(Ser238*)	8.7	3	NDD + Epilepsy	epilepsy with absences and generalized tonic-clonic seizures, severe intellectual disability with autistic traits, low blood pressure, obstipation, normal MRI 2008	het	de novo	yes, 2/5 brothers with ID and epilepsy
PRICKLE1	NM_153026.2:c.128A>G p.(Glu43Gly)	8.7	1	NDD + Epilepsy	Global developmental delay with a decreased and autistic spectrum disorder characteristics, attends a special school, MRI and EEG inconspicuous	het	de novo	no
CSMD1	Del (NM_033225.5): 8: 4277425-4277637	8.7	1	NDD + Epilepsy	Psychosis, Dementia, Intellectual disability, Seizures, Intellectual disability, mild, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Intellectual disability, moderate, Intellectual disability, borderline, Focal seizures, EEG with focal epileptiform discharges	het	unknown	mother with schizophrenia
USF3	NM_001009899.3: c.1750C>T p.(Gln584*)	8.6	1	NDD	muscular hypotonia, developmental delay, normal cMRI, left retinal coloboma	het	de novo	no
ENO2	NM_001975.2: c.710C>T p.Thr237Met	8.6	1	NDD	mild ID, small for gestational age, short stature, microcephaly	homo	paternal & maternal	2fm, cousins I°
PRKCA	NM_002737.2: c.64C>T p.(Arg22Cys)	8.6	4	NDD	Tall stature, Strabismus, Esotropia, Precocious puberty, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Curly hair, Scoliosis	het	de novo	no

GRIK5	NM_001301030.1:c.818C>A p.(Ser273Tyr)	NM_001301030.1:c.1745G>A p.(Arg582His)	8.6	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	comphet	paternal & maternal	yes, mother with epilepsy, sister with spina bifida/hydrocephalus + no NDD
SRSF11	NM_004768.3:c.1178del p.(Arg393Hisfs*26)		8.6	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	het	unknown	no
ZNF143	NM_003442.5:c.44_45delAG p.Glu15ValfsTer25		8.6	1	NDD	NDD	homo	paternal & maternal	N/A
COPS2	NM_001143887.1:c.37G>A p.(Glu13Lys)		8.6	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	het	de novo	yes, sister with absence seizures, may be the same phenotype, so be careful evaluating this variant
NDST1	NM_001543.4:c.2468G>A p.(Gly823Glu)		8.6	1	Epilepsy	Focal seizures with cyanosis, sec. generalizing, EEG highly pathological, so far no cMRI examination has been carried out	het	de novo	no

NRDE2	NM_017970.3: c.441del p.(Arg148Alafs*11)	8.5	1	NDD	Intellectual disability, seizures, global developmental delay, encephalopathy infantile spasms	homo	paternal & maternal	yes, two similarily affected siblings
SEC23IP	NM_007190.3: c.2101G>T p.Glu701*	8.5	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	homo	paternal & maternal	1m, cousins 1°
PSMB10	NM_002801.4: c.56+1G>A p.?	8.5		NDD	(+) Global developmental delay,(+) Intellectual disability, borderline,(+) Intellectual disability, mild,(+) Short stature,(+) Microcephaly,(+) Bird-like facies	homo	unknown	no
GNAI1	NM_002069.5:c.430 C>T p.(Arg144*)	8.5	1	NDD	Intellectual disability, Muscular hypotonia Global developmental delay, Bilateral ptosis, Ventricular septal defect, Abnormal facial shape, Migraine, Neurodevelopmental delay, Neurodevelopmental abnormality	het	unknown	yes, mother and father with ID
TKT	NM_001135055.2: c.1751T>C p.(Val584Ala)	8.5	3	NDD	Global developmental delay, Motor delay	het	de novo	no
BMP4	NM_001202.5:c.172 G>C p.(Glu58Gln)	8.4	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia	homo	paternal & maternal	no
APBA1	NM_001163.3:c.521 T>C p.(Leu174Pro)	8.4	2	NDD + Epilepsy	Epileptic encephalopathy, intrauterine hydrocephalus (gestational week 24), epilepsy	het	de novo	no
RORB	NM_006914.3:c.235 +1_235+2insT p.?	8.4	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	het	unknown	yes, Father with epilepsy and already deceased, sister with epilepsy and ID
ZCCHC14	NM_015144.2: c.52C>T p.(Gln18*)	8.4	1	NDD	motor delay, proximal muscle weakness, makrozephalia, epicanthus med., frontal blossing	het	de novo	Father OFC>97 P

SRRT	NM_015908.5: c.437C>T p.(Pro146Leu)	8.4	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	het	de novo	no
EIF2S2	NM_003908.4:c.692 G>A p.(Arg231His)	8.4	2	NDD	Inguinal hernia, Intellectual disability, Global developmental delay, Small for gestational age, Penile hypospadias, Short stature, Decreased body weight	het	de novo	no
PSMC5	NM_002805.5: c.587del p.(Lys196Argfs*29)	8.4	2	NDD	Microcephaly, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Scoliosis, Short stature, Cognitive impairment	het	maternal	yes, Mother with similar symptoms and epilepsy, maternal grandfather with epilepsy, two maternal uncles with epilepsy
KDM6B	NM_001080424.1: c.1130C>T p.(Ala377Val)	8.4	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	homo	paternal & maternal	no
TRIM9	NM_015163.5: c.1117G>A p.(Val373Met)	8.3	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	het	de novo	no
ATP6AP1	NM_001183.5:c.3G> T p.0?	8.3	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart	hemi	unknown	no

GRIN2C	NM_000835.4: deletion exons 2 and 3 p.?		8.3	1	NDD	Epicanthus, Hypertelorism, Behavioral abnormality, Precocious puberty, Intellectual disability	het	unknown	N/A
MKRN1	NM_013446.4: c.340C>T p.(Gln114*)		8.3	1	NDD	Intellectual disability, mild, Global developmental delay, Mild global developmental delay, Short stature, Talipes equinovarus	het	de novo	no
PTBP1	NM_002819.4: c.144A>T p.(Lys48Asn)		8.3	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	het	de novo	no
NCOA1	NM_003743.4:c.345 7C>T p.(Gln1153*)		8.3	2	NDD	Intellectual disability, Global developmental delay, Obesity, Abnormal heart morphology, Truncal obesity, Kyphosis, Attention deficit hyperactivity disorder	het	unknown	no
UNC13C	NM_001080534.2: c.283C>T p.(Arg95*)		8.3	1	NDD	Global developmental delay, microcephaly, Ehlers-Danlos-Syndrom (CHST1 positive)	homo	paternal & maternal	consanguinity
CNTNAP4	NM_033401.3: c.3353G>C p.(Gly1118Ala)		8.3	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures	het	de novo	no
AP3B2	NM_001278512.1: c.2879A>G p.(Asn960Ser)	NM_001278512.1: c.2662G>A p.(Glu888Lys)	8.3	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	comphet	paternal & maternal	no
VPS54	NM_016516.2: c.701C>T p.(Ala234Val)		8.2	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe	homo	paternal & maternal	yes, brother similary affected

SMURF2	NM_022739.3: c.1921A>G p.Thr641Ala	8.2	2	NDD	mild ID, muscular hypotonia, microcephaly, hypospadias, megalocornea, cerebral atrophy	homo	paternal & maternal	2m, twice cousins I°
PRKCE	NM_005400.2:c.118 6C>T p.(Arg396Trp)	8.2	1	Epilepsy	Seizures, Focal-onset seizure	het	de novo	no
SPTBN5	NM_016642.3: c.5680G>T p.(Glu1894*)	8.2	2	NDD	intellectual disability	homo	paternal & maternal	consanguinity
FAM234B	NM_020853.1: c.1009C>T p.Gln337*	8.2	1	NDD + Epilepsy	mild ID, seizures, obesity, delayed puberty	homo	paternal & maternal	1f, cousins 2°
ZBTB34	NM_001099270.1:c. 18del p.(Phe6Leufs*14)	8.2	1	NDD + Epilepsy	Delayed speech and language development, Global developmental delay, Focal-onset seizure, Childhood onset	het	de novo	no
SLC4A7	NM_001321103.1: c.249_252del p.(Lys83Asnfs*62)	8.2	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	homo	paternal & maternal	no
ARL14EP	NM_152316.2: c.707G>A p.(Arg236His)	8.2	2	NDD + Epilepsy	Intellectual disability, Absent speech, Hyperreflexia, Generalized tonic-clonic seizures, Absence seizure, EEG abnormality, Excessive salivation, Brain atrophy, Epileptic encephalopathy	het	de novo	no
ARHGEF6	NM_004840.2:c.257 A>C p.Ap86Ala	8.2	1	NDD	NDD	hemi	unknown	N/A
TANC2	NM_025185.3: c.3397G>T p.(Gly1133*)	8.2	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	het	unknown	no

KLHDC9	NM_152366.4: c.886+1G>C p.?	8.2	1	NDD + Epilepsy	Seizures, Global developmental delay, Generalized hypotonia, Abnormality of the cerebellum, Abnormal facial shape, Generalized seizures	homo	paternal & maternal	yes, Sister with similar symptoms, parents are healthy and cousins (1st degree), mother had recurrent abortion
ATP6V0A1	NM_001130020.1: c.53A>T p.(Gln18Leu)	8.1	1	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	het	de novo	no
SPEN	NM_015001.2: c.3968T>G p.(Met1323Arg)	8.1	3	NDD + Epilepsy	mild global developmental delay, seizures, heterotopia, oral cleft, tall stature, obesity	het	de novo	no
STAM	NM_003473.3: c.119G>C p.(Arg40Pro)	8.1	1	NDD	Short stature, Ataxia, Cataract, Microphthalmia, Microcephaly, Nystagmus, Global developmental delay	het	de novo	N/A
PTOV1	NM_017432.4:c.842 dup p.(Pro282Alafs*79)	8.1	1	NDD + Epilepsy	Intellectual disability, Seizure, Global developmental delay, bilateral toni-clonic seizure, atonic seizure	het	de novo	no
RORB	NM_006914.3:c.777 G>A p.(Trp259*)	8.1	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe, Epileptic spasms	het	unknown	no
CLSTN1	NM_001009566.1: c.1844C>T p.(Thr615Met)	8.1	2	NDD	Cataract, Peters anomaly, Autism, Global developmental delay	homo	paternal & maternal	yes, one affected brother

POLD1	NM_001308632.1: c.1657G>A p.(Val553Ile)	8.1	1	NDD	Global developmental delay	homo	paternal & maternal	yes, brother is also affected
CHRD	NM_001304472.1:c. 2491C>G p.(Pro831Ala)	8.0	3	NDD	Microcephaly, Intellectual disability, Ataxia, Intellectual disability, mild, Global developmental delay, Spastic gait, Gait ataxia, Intention tremor, Spastic ataxia	het	de novo	no
SPEN	NM_015001.2: c.8092A>G p.(Asn2698Asp)	8.0	2	NDD	Microcephaly, Underdeveloped nasal alae, Strabismus, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Postnatal microcephaly	het	de novo	no
LMX1A	NM_001174069.1: c.517dup p.(Ser173Lysfs*15)	8.0	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	het	unknown	no
CMIP	NM_198390.2: c.42del, p.(Gln15Argfs*36)	8.0	3	NDD + Epilepsy	Intellectual disability, seizures	het	paternal	yes, brother simialry affected, father with epilepsy
ASXL2	NM_018263.4:c.189 4C>G p.(His632Asp)	8.0	2	NDD	Seizures, Generalized tonic-clonic seizures, Myoclonic atonic seizures, Epileptic encephalopathy	het	de novo	no
GTF3C3	NM_012086.4: c.1436A>G p.Tyr479Cys	8.0	1	NDD + Epilepsy	mild ID, seizures, recurrent infections, constipation, abnormalities of the face, postaxial hexadactyly, ataxia, radioulnar synostosis, ventricular septal defect, EEG abnormalities	homo	paternal & maternal	2f, cousins 1°
TRMT1	NM_001136035.2: c.1964G>A p.(Gly655Glu)	8.0	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	het	de novo	no
NLRP5	NM_153447.4: c.1846_1849del p.(Lys616Glyfs*17)	8.0	2	NDD + epilepsy	(+) Dravet syndrome,(+) Seizure,(+) Myoclonic seizure,(+) Myoclonic absence seizure,(+) Global developmental delay,(+) Intellectual disability	homo	paternal & maternal	Cousin of the father with epilepsy

ATP6AP2	NM_005765.3: c.858G>A p.(Ala286=)		8.0		NDD	(+) Moderate global developmental delay,(+) Muscular hypotonia,(+) Dysgenesis of the hippocampus,(+) Aggressive behavior,(+) Impulsivity,(+) Low frustration tolerance,(+) Pes planus,(+) Synophrys,(-) Seizure,(-) Ataxia	het	de novo	no
DOCK7	NM_001271999.1: c.2977T>C p.(Trp993Arg)	NM_001271999.1:c. 708del p.(Phe236Leufs*13)	8.0	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	comphet	paternal & maternal	no
EPHB2	NM_001309193.1: c.2858T>C p.(Ile953Thr)		8.0	1	NDD + Epilepsy	epilepsy, global developmental delay, dysmorphic facial features	het	de novo	no
CCDC66	NM_001141947.1:c. 847_848del p.(Glu283Serfs*3)		8.0	3	NDD + Epilepsy	Global developmental delay with delayed speech, astatic attacks, absence epilepsy and EEG abnormalities	homo	paternal & maternal	no
ARHGEF7	NM_001113511.2: c.17A>C p.(Gln6Pro)		7.9	3	NDD	global developmental delay, intellectual disability	het	de novo	no
RHEB	NM_005614.3: c.47C>T p.(Ser16Phe)		7.9	1	NDD + Epilepsy	Tall stature, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, normal MRI	het	de novo	no
PPFIA1	NM_003626.3: c.1070A>G p.His357Arg		7.9	1	NDD	very severe ID, muscular hypotonia, spasticity, resting tremor, abnormality of the thorax, seizures, cerebral atrophy	homo	paternal & maternal	2m, >cousins I°
REST	NM_005612.4:c.222 7G>A p.(Glu743Lys)		7.9	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Pes planus, Focal tonic seizure, Cognitive impairment	homo	paternal & maternal	no
RASAL2	NM_004841.3: c.433G>T p.(Glu145*)		7.9	3	NDD	Global developmental delay, Motor delay	het	de novo	no
SLITRK2	NM_001144003.2: c.2485G>T p.(Glu829*)		7.9	3	NDD	Severe global developmental delay, sleep disturbance, behavioral abnormalities, flat face	hemi	maternal	no

AVPR1A	NM_000706.4: c.164T>A p.(Ile55Asn)	7.9	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	homo	paternal & maternal	no
CTBP2	NM_022802.2: c.1192dup p.(Arg398Profs*68)	7.9	3	NDD + Epilepsy	Intellectual disability, epilepsy with generalized tonic-clonic seizures, short attention span	het	maternal	yes, Father with epilepsy, mother with epilepsy and intellectual disability
POLL	NM_001174084.1:c. 1255C>T p.(Arg419*)	7.8	5	NDD + Epilepsy	Intellectual disability, Seizure	homo	paternal & maternal	no
ZIK1	NM_001010879.3: c.924del p.(Ser308Serfs*203)	7.8	3	NDD + Epilepsy	Intellectual disability, Generalized seizures, Febrile seizures, Focal seizures	homo	paternal & maternal	no
RNF13	NM_007282.4:c.(409+1_410-1)_(500+1_501-1)del p.?	7.8	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	het	de novo	no
CNOT1	NM_001265612.1: c.6727A>G p.(Met2243Val)	7.8	1	NDD	Intellectual disability, Global developmental delay	homo	paternal & maternal	no
KCNN2	NM_021614.3: c.1082A>G p.(Tyr361Cys)	7.8	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	het	de novo	no
ADD1	NM_176801.2: c.849del p.(Val284Phefs*2)	7.8		NDD + epilepsy	+) Moderate global developmental delay,(+) Seizure,(+) Abnormality of the face,(+) Mitral valve prolapse,(+) Mitral regurgitation	het	het	no

SLC5A7	NM_021815.4: c.178+1G>C p.?		7.8	1	Neuro	Ataxia, spastic paraplegia, muscle weakness, hyperreflexia, pes cavus, myalgia, limb muscle weakness, paraplegia	het	maternal	yes, mother affected
CHURC1	NM_145165.3: c.349_350insG p.(Leu117Argfs*15)	NM_145165.3:c.400del p.(Arg134Aspfs*3)	7.8	2	NDD + Epilepsy	Tall stature, Macrocephaly, Delayed speech and language development, Enuresis, Seizures, Global developmental delay, Obesity, Rett syndrome	comphet	paternal & maternal	no
ATP2C2	NM_001286527.2: c.2636A>G p.Asp879Gly		7.8	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	homo	paternal & maternal	2fm, cousins I°
SLC32A1	NM_080552.2: c.787G>A p.(Val263Met)		7.8	2	NDD + Epilepsy	Intellectual disability, Seizures, Generalized myoclonic seizures, Infantile onset	het	de novo	no
TENM3	NM_001080477.3: c.2221G>A p.(Glu741Lys)		7.8	1	NDD	Autism, Autistic behavior, Intellectual disability, Global developmental delay, Intellectual disability, moderate, Intellectual disability, severe	het	de novo	no
CASKIN1	NM_020764.3: c.4103G>A p.(Ser1368Asn)		7.8	3	NDD	global developmental delay, absent speech, gait disturbance, EEG abnormality, decreased body weight	homo	paternal & maternal	yes, one affected brother
DGCR2	NM_005137.3: c.998T>C p.(Leu333Pro)		7.8	2	NDD	Cognitive impairment, Global developmental delay, Tall stature, Obesity	het	unknown	no
LRRK2	NM_198578.3: c.3634C>T p.(Pro1212Ser)	NM_198578.3:c.137C>T p.(Thr46Met)	7.8	3	NDD	Behavioral abnormality, Intellectual disability, Gait ataxia	comphet	paternal & maternal	no
MCM7	NM_001278595.1: c.1147A>C p.(Met383Leu)		7.7	1	NDD + Epilepsy	Intellectual disability, Seizures, IGlobal developmental delay, Infantile onset, epileptic encephalopathy	het	de novo	no

MDGA2	NM_001113498.2: c.794T>A p.(Val265Asp)	7.7	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	het	de novo	yes, Father with similar symptom, grandmother (paternal): mental retardation; Grandmother and great-grandmother maternal: Leukemia in old age
CLMN	NM_024734.3: c.730C>T p.Arg244*	7.7	1	NDD	moderate ID, muscular hypotonia, gait disturbance, EEG abnormalities, cerebral atrophy	homo	paternal & maternal	1m, cousins I°
PSMB4	NM_002796.2: c.226G>A p.(Gly76Ser)	7.7	2	NDD + Epilepsy	Intellectual disability, Absent speech, Hyperreflexia, Generalized tonic-clonic seizures, Absence seizure, EEG abnormality, Excessive salivation, Brain atrophy, Epileptic encephalopathy	het	de novo	no
ABCB5	NM_001163941.1: c.2867_2867+1del p.(Ile956Lysfs*43)	7.7		NDD	(+) Mild global developmental delay,(+) Muscular hypotonia	het	de novo	no
XPOT	NM_007235.6: c.1516_1517del p.(Val506Cysfs*2)	7.7		NDD	(+) Cerebral vasculitis,(+) Ischemic stroke,(+) Moyamoya disease,(+) Leukoencephalopathy	het	unknown	no
STT3B	NM_178862.2: c.777+4A>C p.?	7.7	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	het	de novo	no
GABRG1	NM_173536.3: c.487A>G p.(Thr163Ala)	7.7	2	NDD	Strabismus, Autism, Ataxia, Specific learning disability, Gait ataxia, Language impairment, Pain insensitivity, Abnormality of movement, Motor tics, Dyskinesia, Exodeviation	het	de novo	no

CSNK1A1	NM_001025105.2: c.686G>A p.(Arg229Gln)		7.7	3	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia	het	de novo	no
CSMD1	NM_033225.5: c.3641T>C p.(Leu1214Pro)		7.7	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	het	de novo	no
GCN1	NM_006836.1: c.7082G>A p.(Arg2361Gln)		7.7	3	NDD	Seizures, Hypoglycemia, Myopathy, Focal seizures, Ichthyosis, EEG with focal epileptiform discharges	het	de novo	no
HSP90AA1	NM_001017963.2: c.626G>A p.(Arg209Gln)		7.7	3	NDD	Global developmental delay and severe motor delay, cerebral palsy (GMFCS level 5), hypoplasia of the corpus callosum	homo	paternal & maternal	no
APOLD1	NM_001130415.1: c.755_756del p.(Glu252Valfs*20)		7.6	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	homo	paternal & maternal	no
NARS1	NM_004539.3: c.1600C>T p.(Arg534*)		7.6	2	NDD + Epilepsy	GDD, ataxia, muscular hypotonia, microcephaly, EEG abnormalities, brain volume reduction	het	de novo	no
CELSR2	NM_001408.2: c.4706C>T p.(Pro1569Leu)	NM_001408.2:c.8629G>A p.(Gly2877Ser)	7.6	3	NDD	global developmental delay, absent speech, gait disturbance, EEG abnormality, decreased body weight	comphet	paternal & maternal	yes, one brother deceased at age of three with similar symptoms, three unaffected
FGF2	NM_002006.4: c.498C>G p.(Tyr166*)		7.6	1	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	het	unknown	no

CACNG3	NM_006539.3:c.437-1G>A p.?	7.6	1	NDD	Microcephaly, Intellectual disability, Global developmental delay, Intellectual disability, moderate, Mild microcephaly, Decreased head circumference	het	unknown	no
TNK2	NM_001010938.1: c.278T>G p.(Leu93Arg)	7.6	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	het	de novo	no
PUM2	NM_015317.1: c.1595G>A p.(Ser532Asn)	7.6	1	NDD + Epilepsy	Seizures, Global developmental delay, Hypsarrhythmia,	het	de novo	no
DLG5	NM_004747.3:c.453 dup p.(Gln152Serfs*26)	7.6	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	het	unknown	no
GCC2	NM_181453.3: c.3982C>T p.His1328Tyr	7.6	1	NDD	ID, short stature, elbow contractures, wrist contractures, axillar pterygium, abnormalities of the face, deafness, abnormality of thrombocytes	homo	paternal & maternal	2m, cousins I°
EIF4A2	NM_001967.3: c.109_111delGAT p.Asp37del	7.6	1	NDD	mild ID, muscular hypotonia, tremor	homo	paternal & maternal	1f, cousins I° once removed
WDFY3	NM_014991.4: c.6820T>C p.(Ser2274Pro)	7.6	1	NDD	ID, Jesem with microcephaly and psychosis	het, in multiple	unknown	both siblings affected, parents are consanguineous (no information regarding phenotype of the parents)
DUT	XM_005254212.1: c.218T>C p.(Val73Ala)	7.6	1	NDD + Epilepsy	Retrognathia, Myoclonus, EEG abnormality, Infantile encephalopathy, Epileptic encephalopathy	het	de novo	no

PIGC	NM_002642.3: c.422C>T p.(Thr141Ile)	NM_002642.3:c.138 C>A p.(Tyr46*)	7.6	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	comphet	paternal & maternal	no
EIF3B	NM_001037283.1: c.2120G>A p.(Arg707Gln)		7.6	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	het	de novo	no
CEP76	NM_024899.3: c.302T>C p.Ile101Thr		7.6	1	NDD	moderate ID, muscular hypotonia, short stature, microcephaly	homo	paternal & maternal	1m, >cousins I°
EP400	NM_015409.4:c.427 7+1G>T p.?		7.5	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	het	unknown	no
GLRA2	NM_002063.4: c.1334G>A p.(Arg445Gln)		7.5		NDD + epilepsy	(+) Tonic seizure,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Intellectual disability,(+) Global developmental delay,(+) Cognitive impairment	hemi	maternal	no

TFE3	NM_006521.5: c.566A>G p.(Tyr189Cys)	7.5	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	hemi	de novo	no
LRRC7	NM_001330635.1:c. 2143C>T p.(Gln715*)	7.5	1	NDD	Intellectual disability, Global developmental delay,Overweight	het	unknown	yes, brother with HSMN and ID
SIPA1L1	NM_015556.2:c.332 1_3322del p.(Arg1107Serfs*11)	7.5	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	het	unknown	no
FRY	NM_023037.2: c.4688G>C p.(Ser1563Thr)	7.5	1	NDD	global developmental delay, intellectual disability, epileptic seizures, microcephaly, Dandy-Walker malformation, Polymicrogyria, syndactyly, partial duplication of thumb phalanx	het	de novo	no
POU3F3	NM_006236.2: c.1220G>T p.(Arg407Leu)	7.5	1	NDD	GDD (first words with 27mo, first steps with >30mo), microcephaly, EEG abnormalities, borad-based gait, strabism, myopia, facial dysmorphism	het	unknown	no
RADIL	NM_018059.4: c.1450C>T p.(Gln484*)	7.5	1	NDD	recurrent hypoglycemia, microcephaly, hypopituritism	homo	paternal & maternal	no

KPNA1	NM_002264.3:c.1015G>A p.(Ala339Thr)		7.4	1	NDD	Global developmental delay and speech delay, microcephaly	het	de novo	no
APLNR	NM_005161.4:c.952C>T p.(Gln318*)		7.4	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	het	de novo	no
CAPN9	NM_006615.2: c.1591G>A p.(Ala531Thr)	NM_006615.2:c.1273-1_1287del p.(Cys425Glufs*262)	7.4	1	NDD	Global developmental delay, Motor delay, Polyneuropathy, Hip dysplasia, Coxa valga, Kyphosis	comphet	paternal & maternal	yes, father with HMSN1A
MDN1	NM_014611.3: c.11732G>C p.(Ser3911Thr)		7.4	1	NDD	Global developmental delay, Delayed gross motor development, Macrocephaly, Patent foramen ovale	het	de novo	grandmother and great-grandmother maternally with breast cancer
AMZ2	NM_001033569.1: c.25C>T p.Gln9*		7.4	2	NDD	mild ID, muscular hypotonia, microcephaly, hypospadias, megalocornea, cerebral atrophy	homo	paternal & maternal	2m, twice cousins I°
PHACTR1	NM_001242648.2: c.1156G>A p.(Glu386Lys)		7.4	2	NDD	Global developmental delay, Intellectual disability, mild	het	de novo	no
CANX	NM_001024649.1: c.143A>T p.(Asp48Val)	NM_001024649.1: c.1102G>A p.(Val368Ile)	7.4	3	Epilepsie	therapy-resistant epilepsy	comphet	paternal & maternal	no

FAT2	NM_001447.2: c.9524T>C p.(Leu3175Pro)	7.4	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	homo	paternal & maternal	yes, sister similary affected
GABRA3	NM_000808.3: c.931+5G>A p.?	7.3	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	hemi	maternal	no
RHOQ	NM_012249.3: c.359G>A p.(Gly120Glu)	7.3	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	het	de novo	no
PTPRN2	Del (NM_002847.4): 7:157873875-158384503	7.3	1	NDD	Behavioral abnormality, Autism, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Intellectual disability, moderate, Poor speech, Intellectual disability, borderline	het	de novo	no
BBX	NM_001142568.2: c.2524C>G p.(Arg842Gly)	7.3	2	NDD	mild ID, facial dysmorphisms, Missing anterior teeth in the lower jaw, Vierfingerfurche, dry skin, narrow mouth, behavioral abnormalities	homo	paternal & maternal	yes, Mother and sister also with missing front teeth, brother of mother with ID
MBD2	NM_003927.4: c.107G>T p.(Gly36Val)	7.3	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	het	de novo	no

ABCA2	NM_001606.4:c.801_802delTGinsGT p.(Val268Phe)		7.3	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)	homo	paternal & maternal	no
ARFGEF3	NM_020340.4: c.5123+2T>C p.?		7.3	2	NDD	Strabismus, Autism, Ataxia, Specific learning disability, Gait ataxia, Language impairment, Pain insensitivity, Abnormality of movement, Motor tics, Dyskinesia, Exodeviation	het	maternal	no
SF3A1	NM_005877.5: NM_005877.5:c.310G>A p.(Gly104Arg)		7.3	1	Epilepsy	Seizures, Global developmental delay, Abnormality of movement, Epileptic encephalopathy	het	de novo	yes, maternal grandmother with epilepsy
PRDX2	NM_005809.5: c.153C>A p.(Cys51*)		7.3	1	Epilepsy	Seizures, absent septum pellucidum, paroxysmal dyskinesia, dyskinesia	het	de novo	no
VPS4A	NM_013245.2: c.291T>G p.(Ser97Arg)		7.3	2	NDD + Epilepsy	Intellectual disability, Seizures, Epileptic encephalopathy	het	de novo	no
DISP1	NM_032890.3: c.1357A>C p.(Met453Leu)	NM_032890.3:c.3233G>A p.(Arg1078His)	7.3	2	NDD	Cleft palate, Panhypopituitarism, Intellectual disability, Patent ductus arteriosus, Facial cleft, Scoliosis, Short stature, Median cleft lip and palate	comphet	paternal & maternal	no
OGDHL	NM_018245.2: c.2606G>A p.Arg869Gln		7.2	2	NDD	moderate ID, small for gestational age, short stature	homo	paternal & maternal	2fm, >cousins I°
CDK12	NM_016507.4: c.4237C>T p.(His1413Tyr)		7.2	3	NDD + epilepsy	Global developmental delay, Microcephaly, Seizures	homo	paternal & maternal	no
TRPC5	NM_012471.2: c.280G>A p.(Val94Met)		7.2		NDD	(+) Global developmental delay,(+) Hyperactivity,(+) Delayed speech and language development,(+) Hypertelorism,(+) Depressed nasal ridge,(+) Low-set ears,(+) Muscular hypotonia, lateral fallende Lidachsen	hemi	maternal	no

UBR2	NM_015255.2:c.2462+2T>C p.?		7.2	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	het	unknown	no
L3MBTL1	NM_015478.6: c.478T>A p.(Ser160Thr)		7.2	3	NDD	Agitation, Aggressive behavior, Delayed speech and language development, Intellectual disability	homo	paternal & maternal	yes, affected cousins, unclear if the same phenotype, consanguinity
LPIN2	NM_014646.2: c.2537A>G p.(Asn846Ser)		7.2	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	het	de novo	no
NARS1	NM_004539.3: c.1067A>C p.(Asp356Ala)	NM_004539.3:c.1564C>T p.(Arg522*)	7.2	2	NDD + Epilepsy	Global developmental delay, febrile seizures, muscular hypotonia	comphet	paternal & maternal	no
GRAMD1B	NM_001286563.1: c.586C>T p.Arg196Trp		7.2	1	NDD	moderate ID	homo	paternal & maternal	2f, >cousins I°
BDH1	NM_004051.4: c.668G>A p.Arg223His		7.2	1	NDD + Epilepsy	very severe ID, seizures, muscular hypotonia, limb hypertonia, spasticity, short stature, microcephaly, leukodystrophy	homo	paternal & maternal	2fm, >cousins I°
GRIN3B	NM_138690.1: c.1090_1091del p.(Met364Valfs*5)	NM_138690.1:c.1936A>G p.(Met646Val)	7.2	1	NDD	Intellectual disability, Abnormal facial shape, Myoclonus	comphet	paternal & maternal	no
HACL1	NM_012260.3: c.1246C>G p.His416Asp		7.2	1	NDD	severe ID, muscular hypotonia, low-set ears, bifid uvula, cryptorchidism, aplasia cutis congenita, unilateral renal agenesis, cardiac malformation, increased creatine kinase	homo	paternal & maternal	2m, cousins I°

BHLHE40	NM_003670.2: c.62A>G p.(Glu21Gly)		7.2	2	NDD	Hydrocephalus, Coloboma, Iris coloboma, Global developmental delay, Aqueductal stenosis, Lens coloboma, Patent foramen ovale, Absent septum pellucidum, Abnormality of the cerebral white matter, Cerebral white matter atrophy, Cerebral white matter hypoplasia, Hypoplasia of the corpus callosum	het	de novo	no
DNAJC10	NM_001271581.1: c.1671+1G>T p.?		7.2	3	NDD + Epilepsy	Refractory epilepsy, epileptic encephalopathy, psychomotor retardation, microcephaly, short stature, facial dysmorphisms, nephrocalcinosis	het	de novo	no
SLC25A6	NM_001636.3: c.239G>A p.(Arg80His)		7.2	2	NDD	(+) Global developmental delay,(+) Scotoma,(+) Intellectual disability, mild,(+) Intellectual disability, borderline,(+) Myopia,(+) Depressivity,(+) Anxiety,(+) Motor delay,(+) Retinal atrophy	het	de novo	no
PTPRT	NM_133170.4: c.3039+1G>A p.?		7.1		NDD	(+) Cerebral vasculitis,(+) Ischemic stroke,(+) Moyamoya disease,(+) Leukoencephalopathy	het	unknown	no
RAB5B	NM_001252036.1: c.115C>G p.(Arg39Gly)		7.1	3	NDD + Epilepsy	Hydrocephalus, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Aqueductal stenosis	het	de novo	no
GTPBP2	NM_019096.3: c.1191C>A p.(Asn397Lys)		7.1	1	NDD	Tall stature, Macrocephaly, Retrognathia, High forehead, Low-set ears, Global developmental delay	het	de novo	no
LRRIQ3	NM_001105659.1: c.968C>A p.Ser323*		7.1	2	NDD	mild ID	homo	paternal & maternal	2f, >cousins I°
GPR161	NM_001267609.1: c.1550dup p.(Gly518Argfs*44)		7.1	2	NDD	Hypertelorism, Low-set ears, Brachydactyly, Intellectual disability, Global developmental delay, Hypoplasia of the corpus callosum, Elevated serum creatinine, Moderate global developmental delay	het	de novo	no
DIS3	NM_001128226.1: c.1486A>G p.(Arg496Gly)	NM_001128226.1:c.2785T>C p.(*929Glnext*14)	7.1	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormality of body weight, Increased body weight,	comphet	paternal & maternal	yes, paternal uncle went to special school

KDM2B	NM_032590.4: c.2345C>T p.(Ser782Leu)		7.1	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 hypotonia	homo	paternal & maternal	no
EIF2AK2	NM_001135651.2: c.1210T>C p.(Tyr404His)		7.1	1	Epilepsy	West syndrome with salaam spasms, hypsarrythmia in EEG, age-appropriate development	het	de novo	no
RXRB	NM_001270401.1: c.1091C>T p.Pro364Leu		7.1	1	NDD	very severe ID, short stature, microcephaly	homo	paternal & maternal	2f, cousins I°
RBFOX1	NM_001308117.1:c.445C>G p.(Pro149Ala)	NM_001142333.1:c.1069G>C p.(Ala357Pro)	7.1	2	NDD	global muscular hypotension with axial weakness, facial dysmorphia, indicated high palate, broad neck, muscle relief decreasing distally, hypersalivation, no secure free sitting	comphet	paternal & maternal	no
OPCML	NM_001012393.2: c.175del p.(Val59Trpfs*4)		7.1	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 impairment	het	de novo	no
FNDC3A	NM_001079673.1: c.1186G>A p.Asp396Asn		7.1	1	NDD + Epilepsy	severe ID, seizures, muscular hypotonia, short stature	homo	paternal & maternal	2m, >cousins I°
PLXNB3	NM_001163257.1: c.4343C>A p.(Thr1448Asn)		7.1	2	NDD	Hydrocephalus, Intellectual disability, hypotonia, Global developmental delay, Atria septal defect, Patent ductus arteriosus,Transposition of the great arteries with ventricular septal defect	het	de novo	no

ARHGEF28	NM_001080479.2:c.548T>G p.(Leu183Trp)		7.0	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	het	de novo	no
SV2C	NM_014979.3: c.533G>C p.Ser178Thr		7.0	1	NDD	moderate ID, microcephaly, short stature	homo	paternal & maternal	2m, cousins I°
KALRN	NM_001024660.4: c.3534G>T p.(Arg1178Ser)	NM_001024660.4: c.5176+21733A>G p.(=)	7.0	2	Epilepsy	(+) Myoclonic seizure,(+) EEG with spike-wave complexes, suspected focal cortical dysplasia frontal right	comphet	paternal & maternal	no
ARFGEF3	NM_020340.4: c.787G>A p.(Ala263Thr)		7.0	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	het	de novo	no
CHKA	NM_001277.2: c.1021T>C p.(Phe341Leu)	NM_001277.2:c.14dup p.(Cys6Leufs*19)	7.0	1	NDD + Epilepsy	severe psychomotor retardation, central movement disorder with preference for right-sided extremities, epilepsy with epileptic spasms, microcephaly, tendency to self-harm	comphet	paternal & maternal	no
MIA3	NM_198551.2: c.3981+3A>G p.?		7.0	3	NDD	Agitation, Aggressive behavior, Delayed speech and language development, Intellectual disability	homo	paternal & maternal	yes, affected cousins, unclear if the same phenotype, consanguinity
DUX4L4	NM_001177376.2: c.880C>T p.(Gln294*)		7.0	1	NDD	Microcephaly, Intellectual disability, Global developmental delay, Short stature	het	de novo	no
EIF3B	NM_001037283.1: c.28C>A p.(Pro10Thr)		7.0	1	Epilepsy	Absence seizures, EEG abnormality, Febrile seizures, Eyelid myoclonias, Childhood onset	het	de novo	no
NOP58	NM_015934.4: c.1018C>G p.(Leu340Val)		7.0	1	NDD + Epilepsy	Autism, Intellectual disability, Status epilepticus, Focal-onset seizure, Hippocampal atrophy	het	de novo	no

TTC28	NM_001145418.1: c.4501T>C p.(Trp1501Arg)		7.0	2	NDD	Cryptorchidism, Hypospadias, Narrow mouth, Microcephaly, Synophrys, Intellectual disability, Global developmental delay, Highly arched eyebrow, Penile hypospadias, Short stature, Decreased body weight, Microtia, Feeding difficulties, Scrotal hypospadias,	het	de novo	no
SLC4A2	NM_003040.3:c.2507T>C p.(Ile836Thr)		7.0	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	het	de novo	no
TRIM47	NM_033452.2: c.433C>T p.(Leu145Phe)		7.0	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	het	de novo	no
DOC2B	NM_003585.4: c.898G>A p.(Gly300Ser)		6.9	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures, infantile onset	het	de novo	no
RPTOR	NM_020761.2: c.3533G>A p.(Arg1178His)	NM_020761.2:c.503A>G p.(Asn168Ser)	6.9	5	Epilepsy	Seizures, Generalized myoclonic seizures	comphet	paternal & maternal	no
ADIPOR1	NM_001290553.1: c.644T>C p.Leu215Pro		6.9	1	NDD	very severe ID, EEG abnormalities, microcephaly	homo	paternal & maternal	3m, >cousins I°
HYDIN	NM_001270974.2: c.6271A>C p.(Ile2091Leu)		6.9	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech	het	de novo	no
ACTR5	NM_024855.3: c.958G>T p.(Asp320Tyr)		6.9	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	homo	paternal & maternal	no
RGMA	NM_001166283.1: c.748G>C p.(Ala250Pro)		6.9	2	NDD	Spasticity, Global developmental delay, Motor delay, Cerebral palsy, Abnormality of movement, Dyskinesia	het	de novo	no

TRAK2	NM_015049.2: c.1210G>A p.(Val404Ile)		6.9	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	het	de novo	no
HEATR1	NM_018072.5: c.3949-26_3954del p.(Asp1317Valfs*827)		6.9	3	NDD + Epilepsy	Global developmental delay and obsessive-compulsive behavior, seizures	het	unknown	no
KALRN	NM_001024660.3: c.5980C>G p.(Leu1994Val)	NM_001024660.3:c.2171C>T p.(Ser724Leu)	6.9	3	NDD	Hypothyroidism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Short stature, Abnormal social behavior	comphet	paternal & maternal	no
AP1G1	XM_005255821.1: c.468G>A p.(=)		6.9	1	NDD + Epilepsy	Seizures, Epileptic encephalopathy	het	de novo	N/A
WTAP	NM_001270531.1: c.463A>G p.(Lys155Glu)		6.9	1	NDD	Microcephaly, Delayed speech and language development, Hyperactivity, Global developmental delay, dystrophy, Receptive language delay	het	de novo	no
PTGDR2	NM_004778.2: c.924C>A p.(Cys308*)		6.9	3	NDD + Epilepsy	epileptic encephalopathy, therapy-resistant seizures, pain in legs	homo	paternal & maternal	yes, sister also affected, 1 sister died in the age of 6 months
EPHA4	NM_004438.4: c.2105T>C p.(Met702Thr)		6.9	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	het	unknown	no

LRRK1	NM_024652.5:c.5615C>G p.(Ser1872Cys)		6.9	4	Epilepsy	Episodic ataxia, EEG abnormality	het	de novo	yes, maternal aunt with hemiplegic migraine, mother with migraine, paternal grandfather with migraine and apoplex (40yrs)
TNPO3	NM_012470.3: c.2541dup p.(Tyr848Leufs*8)		6.9	3	NDD	Renal agenesis, Abnormal cornea morphology, Aniridia, Microphthalmia, Global developmental delay	het	de novo	no
DOHH	NM_001145165.1: c.446C>G p.(Pro149Arg)	NM_001145165.1:c.224T>G p.(Val75Gly)	6.8	1	NDD + Epilepsy	Global developmental delay, Epilepsy since the age of 3 with tonic-clonic seizures, EEG abnormalities, pain insensitivity	comphet	paternal & maternal	yes, Brother with global developmental delay and muscular hypotonia, maternal aunt with IUFD (Edwards-syndrome), paternal niece with global developmental delay, paternal aunt with induced abortion (Edwards-syndrome)

SPHK2	NM_001204159.2:c.1774del p.(His592Thrfs*19)		6.8	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	het	de novo	no
PLCG1	NM_002660.2: c.3196C>G p.(Pro1066Ala)		6.8	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	het	unknown	no
PKP4	NM_001005476.2: c.744_745del p.(Val250Aspfs*110)		6.8	1	NDD	Microcephaly, Global developmental delay, Global brain atrophy	het	de novo	no
MFAP1	NM_005926.2: c.88T>C p.(Ser30Pro)		6.8	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	het	de novo	no
SLIT1	NM_003061.2: c.4378C>T p.(Arg1460Trp)		6.8	1	NDD	Intellectual disability, movement disorder	homo	paternal & maternal	no
PMM1	NM_002676.2:c.416C>T p.(Ser139Leu)		6.8	2	NDD + Epilepsy	Epileptic encephalopathy, Seizure since the age of 11	het	de novo	no
SYNJ2	NM_003898.3: c.107C>A p.(Ala36Asp)		6.8	1	Epilepsy	Restlessness, Intellectual disability, Hypsarrhythmia, Epileptic spasms, Infantile spasms, I	het	de novo	no
DMAP1	NM_001034023.1: c.581G>A p.(Arg194Gln)	NM_001034023.1:c.670C>T p.(Arg224*)	6.8	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormal facial shape, Cognitive impairment	comphet	paternal & maternal	no
TMEM151A	NM_153266.3:c.76-1G>C p.?		6.8	3	NDD + Epilepsy	Intellectual disability, Seizures, Status epilepticus	het	de novo	no
RNF213	NM_001256071.2:c.9611A>G p.(His3204Arg)		6.8	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia	het	de novo	no

GEMIN4	NM_015721.2: c.1580A>G p.(Asn527Ser)	NM_015721.2:c.1415_1416del p.(Pro472Argfs*23)	6.8	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	comphet	paternal & maternal	no
ATP13A5	NM_198505.2:c.1949A>G p.(Tyr650Cys)	NM_198505.2:c.1241dup p.(Tyr415Leufs*72)	6.8	1	NDD	epileps, ID, cerebral palsy, EEG and MRI abnormlities	comphet	unknown	yes, Brother with intellectual disability and epilepsy deceased with 24 years
MDN1	NM_014611.2:c.13276C>G p.(Leu4426Val)		6.8	2	NDD + Epilepsy	Microcephaly, Seizure, Dystonia, Cerebral palsy, Abnormality of movement, Epileptic encephalopathy	het	de novo	no
FUNDC1	NM_173794.3: c.154A>G p.(Thr52Ala)		6.8	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	hemi	maternal	no
PYROXD2	NM_032709.2:c.1062+2T>G p.?		6.8	5	NDD + Epilepsy	Intellectual disability, Seizure	homo	paternal & maternal	no
EXD3	NM_017820.4: c.859G>A p.(Asp287Asn)	NM_017820.4:c.1831-2A>G p.?	6.7	2	Epilepsy	seizures, peripheral axonal neuropathy, motor delay, gait disturbance, EEG with focal epilepti-form discharges	comphet	paternal & maternal	yes, father with autistic features, father and three siblings with ADHD, one brother with Rolandic epilepsy
SNX27	NM_030918.5: c.913G>A p.(Ala305Thr)	NM_030918.5:c.69_71dup p.(Gly25dup)	6.7	1	NDD	Microcephaly, Hirsutism, Intellectual disability, Global developmental delay, Short stature	comphet	paternal & maternal	no

NLE1	NM_018096.4: c.593A>G p.(His198Arg)	6.7	3	Epilepsy	epilepsy, febrile seizures	het	de novo	no
RORB	NM_006914.3:c.208 T>C p.(Cys70Arg)	6.7	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 encephalopathy	het	maternal	yes, 2/5 brothers with epilepsy and intellectual disability, 2 maternal cousins with epilepsy, paternal aunt with intellectual disability, several paternal relatives with intellectual disability but no epilepsy
BARX2	NM_003658.4: c.386G>A p.(Arg129Gln)	6.7	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia	homo	paternal & maternal	yes,affected monozygotic triplet, parents konsaguin
SGF29	NM_138414.2: c.733T>C p.(Tyr245His)	6.7	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	het	de novo	no
OS9	NM_006812.3: c.1181A>T p.(Glu394Val)	6.7	2	NDD + Epilepsy	Macrocephaly, Seizures, Global developmental delay, Epileptic encephalopathy	homo	paternal & maternal	no

PDE4DIP	NM_001198834.3:c.3733G>A p.(Ala1245Thr)	NM_001198834.3:c.1229_1231delAATinsTAG p.(Glu410_Leu411delinsValVal)	6.7	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology	comphet	paternal & maternal	yes, maternal aunt and paternal uncle with seizures
RFX7	NM_022841.5 : c.3083C>T p.(Pro1028Leu)		6.7	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	het	de novo	no
BSN	NM_003458.3:c.9919A>G p.(Ser3307Gly)		6.7	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	homo	paternal & maternal	no
HDAC4	NM_006037.3: c.1663G>A p.(Gly555Ser)		6.7	3	NDD	Autism, Intellectual disability, Global developmental delay	het	paternal	no
NPTX1	NM_002522.3: c.970G>A p.(Gly324Arg)		6.7	2	NDD	Spastic tetraparesis, Optic atrophy, Periventricular leukomalacia, Microcephaly, Global developmental delay	het	de novo	no
VPS51	NM_013265.3:c.1777A>G p.(Lys593Glu)		6.7	3	Epilepsy	Epilepsy with generalized tonic-clonic seizures, ED 10/2019, microcephaly	het	de novo	no
STC1	NM_003155.2: c.693_697del p.(Glu232Glyfs*12)		6.7	1	NDD	mild global developmental delay, expressive speech disorder, obesity since age three years	het	de novo	no

ORC3	NM_181837.2: c.419A>G p.(Asp140Gly)	6.7	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	homo	paternal & maternal	yes, sister is also affected
TENM2	NM_001122679.1:c. 7970C>T p.(Thr2657Met)	6.7	3	NDD	Global developmental delay, intellectual impairment, absent speech	het	de novo	no
PLEKHB1	NM_021200.2: c.164A>C p.(His55Pro)	6.7	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia	homo	paternal & maternal	yes,affected monozygotic triplet, parents konsaguin
FNDC3A	NM_001079673.2: c.760+1G>T p.?	6.7	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	het	unknown	no
LRIG3	NM_153377.4: c.979G>A p.(Asp327Asn)	6.7	1	NDD	Global developmental delay, Absent speech, Myelomeningocele	het	de novo	no
SLC16A10	NM_018593.4: c.626G>A p.(Gly209Asp)	6.6	2	NDD + Epilepsy	Microcephaly, Behavioral abnormality, Seizures, Global developmental delay, Absence seizure, Generalized-onset seizure, Myoclonic atonic seizures	het	de novo	no
BCAS1	NM_003657.3: c.1720C>T p.(Pro574Ser)	6.6	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe	homo	paternal & maternal	yes, brother similary affected
PACS1	NM_018026.4: c.445-17_445-7del p.?	6.6	3	Epilepsy	Suspected vitamin-B6-dependent epilepsy, Generalized-onset epileptic spasm, Atypical absence seizure, EEG abnormality	het	de novo	no

SEMA5B	NM_001256347.1: c.499C>A p.(Pro167Thr)		6.6	2	NDD	Hydrocephalus, Coloboma, Iris coloboma, Global developmental delay, Aqueductal stenosis, Lens coloboma, Patent foramen ovale, Absent septum pellucidum, Abnormality of the cerebral white matter, Cerebral white matter atrophy, Cerebral white matter hypoplasia, Hypoplasia of the corpus callosum	het	de novo	no
ITSN1	NM_003024.2:c.1690T>C p.(Ser564Pro)		6.6	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	het	unknown	no
MTCH1	NM_001271641.1:c.2T>A p.0?		6.6	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure	homo	paternal & maternal	no
ZMYM4	NM_005095.2: c.1300A>G p.(Thr434Ala)		6.6	1	Epilepsy	Seizures, Global developmental delay, Generalized tonic-clonic seizures with focal onset, Focal seizures, Epileptic encephalopathy	het	de novo	no
ZNF449	NM_152695.5:c.961A>T p.(Lys321*)		6.6	1	Epilepsy	Hypothyroidism, Primary hypothyroidism, Congenital hypothyroidism, Seizure, Generalized-onset seizure, Atonic seizure, Focal emotional seizure with laughing, Clonic seizure	hemi	de novo	no
GEMIN5	NM_015465.4: c.3340C>G p.(Leu1114Val)	NM_015465.4:c.2504A>G p.(Lys835Arg)	6.6	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	comphet	paternal & maternal	yes, sister has variant also and since she only had a few seizures and no NDD
DZIP3	NM_014648.3: c.209C>T p.(Pro70Leu)		6.6	1	NDD	Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Poor speech, Cognitive impairment	het	de novo	yes, paternal nice with ID
HMG20A	NM_001304504.1: c.694C>G p.Arg232Gly		6.6	1	NDD + Epilepsy	moderate ID, seizures	homo	paternal & maternal	5fm, cousins I°

ADAM11	NM_002390.5: c.98G>T p.(Trp33Leu)		6.6	1	NDD	Strabismus, Hypermetropia, Delayed speech and language development, Intellectual disability, Seizures, Global developmental delay, Absent speech, Absence seizures, Febrile seizures, Receptive language delay	het	de novo	no
E2F4	NM_001950.3: c.947_958del p.(Ser316_Ser319del)		6.6	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	het	de novo	no
PKD1	NM_001009944.2: c.11396C>T p.(Ala3799Val)	NM_001009944.2:c.8593C>T p.(Arg2865Trp)	6.6	1	NDD + Epilepsy	Enuresis, Seizures, Global developmental delay, Specific learning disability, Absence seizure, Generalized-onset seizure	comphet	paternal & maternal	no
ARMCX1	NM_016608.1: c.520dup p.(Arg174Profs*3)		6.6	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	hemi	maternal	no
TTC28	NM_001145418.1: c.3020A>G p.(Tyr1007Cys)		6.6	1	NDD	Tall stature, Macrocephaly, Autistic behavior, Delayed speech and language development, Intellectual disability, Global developmental delay, Obesity, Abnormal social behavior	het	de novo	yes, mother with intellectual impairment
RAPGEF3	NM_001098531.2: c.2312A>C p.(His771Pro)		6.6	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormal facial shape, Cognitive impairment	het	de novo	no
CHD6	NM_032221.4: c.1678C>A p.(Gln560Lys)	NM_032221.4:c.2224A>G p.(Arg742Gly)	6.6	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech	comphet	paternal & maternal	no
C9ORF114	NM_016390.3: c.1058C>T p.Thr353Met		6.6	1	NDD + Epilepsy	profound ID, seizures, microcephaly, short stature, limb hypertonia, bruxism	homo	paternal & maternal	2m, >cousins I°
SLC25A5	NM_001152.4: c.616A>G p.(Lys206Glu)		6.6	2	NDD	Mild global developmental delay, Cleft palate, Hearing impairment, Ventricular septal defect, Patent ductus arteriosus, Pulmonic stenosis, Hypospadias, Enlarged vestibular aqueduct, Pes	hemi	maternal	no
HSPH1	NM_006644.4: c.515del p.(Asn172Metfs*3)		6.5		NDD + epilepsy	(+) Seizure,(+) Global developmental delay,(+) Stereotypical hand wringing,(+) Muscular hypotonia	het	unknown	no

FEN1	NM_004111.5: c.140G>A p.(Arg47His)	6.5	1	Epilepsy	Seizures, Focal impaired awareness seizure, Spherocytosis, Arrhythmia	homo	paternal & maternal	no
SKIDA1	NM_207371.3: c.2600C>T p.Ala867Val	6.5	1	NDD	severe ID, small for gestational age, strabismus, short stature	homo	paternal & maternal	3fm, cousins I°
USP19	NM_001199161.1:c. 2012C>A p.(Ser671Tyr)	6.5	4	Epilepsy	Episodic ataxia, EEG abnormality	het	de novo	yes, maternal aunt with hemiplegic migraine, mother with migraine, paternal grandfather with migraine and apoplex (40yrs)
CDH20	NM_031891.3: c.958G>C p.(Asp320His)	6.5	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	het	de novo	no
SPSB1	NM_025106.3: c.572T>C p.(Ile191Thr)	6.5	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	het	de novo	no
SF3A1	NM_005877.5:c.709 C>T p.(Arg237*)	6.5	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	het	maternal	no
FAM214B	NM_001317991.1: c.588del p.(Ile196Metfs*115)	6.5	2	NDD	Intellectual disability	het	paternal	yes, Father and other male relatives are also affected

POLR1B	NM_001137604.1: c.2893G>A p.(Val965Ile)		6.5	3	NDD	Seizures, Pachygyria, Delayed CNS myelination, Heterotopia, Periventricular gray matter heterotopia, Intracranial cystic lesion, Abnormality of brain morphology	het	de novo	no
TRPV1	NM_018727.5:c.896 C>T p.(Thr299Met)		6.5	5	NDD + Epilepsy	Intellectual disability, Seizure	homo	paternal & maternal	no
UNC5D	NM_080872.3: c.977A>G p.(His326Arg)		6.5	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 hypotonia	homo	paternal & maternal	no
FAM168B	NM_001009993.2: c.452G>A p.(Gly151Glu)		6.5	3	NDD + Epilepsy	Seizures, Abnormal social behavior, Epileptic encephalopathy	het	de novo	no
ZC3H4	NM_015168.1: c.54_71dup p.(Pro19_Pro24dup)		6.5	1	NDD + Epilepsy	global developmental delay, faocal epilepsy, truncal ataxia	het	de novo	no
PAM	NM_001319943.1: c.1670C>G p.(Ser557Trp)		6.5	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	homo	paternal & maternal	no
CASP9	NM_001229.4: c.631-6T>C p.?	NM_001229.4:c.710 A>C p.(His237Pro)	6.5	3	NDD	Renal agenesis, Abnormal cornea morphology, Aniridia, Microphthalmia, Global developmental delay	comphet	paternal & maternal	no
FYTTD1	NM_032288.6: c.755G>C p.(Arg252Pro)		6.5	1	NDD	Microcephaly, Nystagmus, Impaired social interactions, Intellectual disability, Muscular hypotonia, Global developmental delay, EEG abnormality	het	de novo	no
PSMB6	NM_002798.2:c.238 T>C p.(Ser80Pro)		6.5	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	homo	paternal & maternal	no

ZNF81	NM_007137.3: c.476A>G p.(Lys159Arg)	6.5	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	hemi	maternal	no
SMARCA1	NM_003069.4: c.34G>A p.(Val12Met)	6.5	4	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay	hemi	maternal	no
DNAJC7	NM_001144766.2: c.941C>T p.(Ala314Val)	6.4	1	NDD + Epilepsy	Seizures, Global developmental delay, Generalized seizures, Hypsarrhythmia, Epileptic spasms	het	de novo	no
TSPAN18	NM_130783.4: c.275T>C p.Leu92Pro	6.4	1	NDD	severe ID, deafness	homo	paternal & maternal	2m, cousins I°
SV2B	NM_001167580.2: c.895C>T p.(Gln299*)	6.4	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	het	unknown	yes, maternal grandmother with epilepsy
AHCYL2	NM_015328.3: c.304_330dup p.(Gly102_Gly110dup)	6.4	2	NDD + Epilepsy	Delayed speech and language development, Seizures, Focal seizures, Multifocal epileptiform discharges, EEG with focal epileptiform discharges, Epileptic encephalopathy	het	de novo	no
FRMD5	NM_032892.5: c.1045A>C p.(Ser349Arg)	6.4	1	NDD	Global developmental delay, Expressive language delay, Receptive language delay, Gait ataxia, Muscular hypotonia, Microcephaly, Cerebral atrophy, Abnormality of eye movement	het	de novo	no
ANKFN1	NM_153228.2: c.1052A>G p.(Asn351Ser)	6.4	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures, infantile onset	het	de novo	no
ATP8B2	NM_020452.3: c.1745G>A p.(Arg582Gln)	6.4	2	NDD	Intellectual disability, Intellectual disability, mild, Intellectual disability, moderate, Increased body weight, Increased adipose tissue	het	de novo	no

BIRC6	NM_016252.3:c.10735A>G p.(Met3579Val)	6.4	2	NDD + Epilepsy	Microcephaly, Visual impairment, Intellectual disability, Seizures, Global developmental delay, Motor delay, Encephalopathy, Generalized tonic-clonic seizures	homo	paternal & maternal	no
UNC13A	NM_001080421.2: c.2786G>A p.(Gly929Glu)	6.4	3	NDD	Agitation, Aggressive behavior, Delayed speech and language development, Intellectual disability	het	unknown	yes, affected cousins, unclear if the same phenotype, consanguinity
FAM200A	NM_145111.3:c.1702C>T p.(Gln568*)	6.4	5	NDD + Epilepsy	Intellectual disability, Seizure	homo	paternal & maternal	no
RNF103-CHMP3	NM_001198954.1:c.307G>A p.(Val103Met)	6.4	3	NDD + Epilepsy	Global developmental delay with delayed speech, astatic attacks, absence epilepsy and EEG abnormalities	het	de novo	no
RGL1	NM_015149.4: c.737C>G p.(Ser246Cys)	6.4	2	NDD + Epilepsy	Tall stature, Macrocephaly, Delayed speech and language development, Enuresis, Seizures, Global developmental delay, Obesity, Rett syndrome	het	de novo	no
USP13	NM_003940.2: c.2498+1G>A p.?	6.4	1	NDD	Renal dysplasia, Polycystic kidney dysplasia, Synophrys, Global developmental delay	het	de novo	no
KCND2	NM_012281.2:c.1003A>G p.(Ile335Val)	6.3	1	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	het	unknown	no
FRYL	NM_015030.1: c.3851T>G p.(Leu1284Arg)	6.3	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	homo	paternal & maternal	no

TMEM151B	NM_001137560.1: c.1319T>A p.(Val440Asp)	6.3	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	het	de novo	no
LAMA5	NM_005560.4:c.5408C>T p.(Ser1803Phe)	6.3	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Pes planus, Focal tonic seizure, Cognitive impairment	homo	paternal & maternal	no
DOCK1	NM_001380.4: c.4546A>G p.(Ser1516Gly)	6.3	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 hypotonia	homo	paternal & maternal	no
IGF2R	NM_000876.3:c.1312G>A p.(Ala438Thr) NM_000876.3:c.5506G>A p.(Val1836Ile)	6.3	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart	comphet	unknown	no
SLITRK2	NM_001144003.2:c.265G>T p.(Val89Leu)	6.3	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	het	de novo	no
NRCAM	NM_001193582.1: c.3362C>G p.(Pro1121Arg)	6.3	1	NDD + epilepsy	Hypospadias, Microcephaly, Atypical absence seizure, Bilateral tonic-clonic seizure, Intellectual disability, Premature birth, Patent ductus arteriosus, Hearing impairment	het	unknown	yes, grandfather hat ms with epilepsy, the son of the grandfathers sister has intellectual disabillity and abnormal gait
VGF	NM_003378.3: c.1318G>A p.(Glu440Lys)	6.3	4	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity	homo	paternal & maternal	no

NFATC1	NM_001278669.1:c.2249_2251delCCTinsTCG p.(Pro750_Cys751delinsLeuGly)		6.3	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	het	de novo	no
BTN2A2	NM_001197237.1: c.386G>A p.Cys129Tyr		6.3	1	NDD	very severe ID, muscular hypotonia, constipation	homo	paternal & maternal	2m, cousins 1° once removed
KALRN	NM_001024660.4: c.4026-8T>C p.?	NM_001024660.4:c.5369A>G p.(Gln1790Arg)	6.3	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	comphet	paternal & maternal	no
XDH	NM_000379.3: c.2559G>C p.(Lys853Asn)		6.3	4	Epilepsy	Seizures, Generalized-onset seizure	het	de novo	no
MACF1	NM_012090.5: c.1531C>T p.(Arg511Cys)	NM_012090.5:c.3465G>A p.(=)	6.3	1	NDD + Epilepsy	global developmental delay, seizures,	comphet	paternal & maternal	yes, father had also seizures until the age of 17 years
BDP1	NM_018429.2:c.4813A>G p.(Arg1605Gly)		6.3	3	NDD + Epilepsy	Intellectual disability, Seizures, Status epilepticus	het	de novo	no
GIPC1	NM_005716.3: c.718C>T p.(Arg240*)		6.3	1	NDD	Intellectual disability, V,a, epilepsy, failure to thrive, short stature, microcephaly, hypotonia, obstipation, strabismus, not able to walk, no language	het	de novo	no
DBF4B	NM_145663.2: NM_145663.2:c.902G>T p.(Cys301Phe)		6.3	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe	homo	paternal & maternal	yes, brother is also affected
MYO5B	NM_001080467.2: c.1624C>T p.(Arg542Cys)		6.3	2	NDD + epilepsy	mild global developmental delay, febrile seizure (within the age range of 3 months to 6 years)	het	de novo	yes, father with fever-associated seizures in

SLC25A43	NM_145305.2: c.224C>T p.(Ala75Val)		6.3	2	NDD	Cryptorchidism, Microcephaly, Global developmental delay, Motor delay, Growth delay, Intrauterine growth retardation	hemi	maternal	no
SETD1B	NM_015048.1: c.3074G>A p.(Arg1025Gln)	NM_015048.1:c.4354C>T p.(Arg1452Cys)	6.3	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech	comphet	paternal & maternal	no
MRO	NM_001127176.1: c.550T>A p.(Phe184Ile)		6.3	1	NDD + Epilepsy	Absent speech, Obesity, Intellectual disability, severe, Epilepsy	homo	paternal & maternal	yes, brother with similar symptomes, but without epilepsy
MCIDAS	NM_001190787.1: c.487C>T p.(Arg163Trp)		6.2	1	NDD + Epilepsy	Stereotypy, Delayed speech and language development, Global developmental delay, Motor delay, Delayed gross motor development, EEG abnormality, Delayed fine motor development	het	de novo	no
PPRC1	NM_015062.4: c.1825C>T p.Pro609Ser		6.2	1	NDD + Epilepsy	severe ID, seizures, cerebral atrophy, leukodystrophy, macular degeneration, abnormality of the retina	homo	paternal & maternal	2f, cousins I°
UTP18	NM_016001.2: c.1503+1G>A p.?		6.2	2	NDD + Epilepsy	Epilepsy (post- brain haemorrhage condition), intelligence impairment, autism, seizures, premature birth	het	de novo	no
FBP2	NM_003837.3: c.128A>G p.(Lys43Arg)		6.2	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	het	de novo	no
NUCB2	NM_005013.2: c.88_91del p.(Asp30Argfs*15)		6.2	3	NDD + Epilepsy	Intellectual disability, Seizures, Ataxia	het	de novo	no
PARD6A	NM_016948.2: c.934C>T p.Arg312*		6.2	1	NDD	mild ID, stereotypical motor behaviors, muscular hypotonia, strabismus, EEG abnormalities	het	de novo	1m, cousins 2° once removed
KCTD16	NM_020768.3: c.1231T>C p.(Phe411Leu)		6.2	3	NDD + Epilepsy	Therapy-resistant epilepsy since the age of two, Epileptic encephalopathy	het	de novo	no

ASTN2	NM_014010.4: c.1013A>G p.(Lys338Arg)	NM_014010.4:c.872 A>T p.(Asp291Val)	6.2	1	NDD + Epilepsy	Ptosis, Seizures, Epileptic encephalopathy	comphet	paternal & maternal	no
TMEM132D	NM_133448.2: c.1489A>G p.Lys497Glu		6.2	2	NDD	mild ID	homo	paternal & maternal	2f, >cousins I°
DOCK7	NM_001271999.1: c.2932C>T p.(Arg978Cys)	NM_001271999.1:c. 2464G>T p.(Ala822Ser)	6.2	1	NDD	Global developmental delay, postnatal growth retardation, facial dysmorphisms with ante-verted nares, smooth philtrum, narrow upper lip and dysplastic ears	comphet	paternal & maternal	no
MTMR9	NM_015458.3: c.220A>G p.(Lys74Glu)		6.2	1	NDD	Developmental delay, club foot, short stature, microcephaly, deafness	het	de novo	no
ANKRD30B	NM_001145029.1: c.1795G>T p.(Glu599*)		6.2	1	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay	homo	paternal & maternal	no
TANK	NM_001199135.1:c. 1012T>C p.(Tyr338His)		6.2	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	het	de novo	no
TMEM94	NM_001321148.1: c.2906G>A p.(Arg969Gln)	NM_001321148.1:c. 2978T>C p.(Met993Thr)	6.2	1	NDD + Epilepsy	Seizures, Global developmental delay, Focal seizures, Retinoblastoma	comphet	paternal & maternal	no
PDE4DIP	NM_001198834.3: c.5842A>G p.(Lys1948Glu)	NM_001198834.3:c. 4063C>T p.(Arg1355*)	6.2	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Cerebellar vermis atrophy, Cognitive impairment	comphet	paternal & maternal	no

PDE4DIP	NM_001198834.3:c.6862A>C p.(Lys2288Gln)	NM_001198834.3:c.6043A>G p.(Ile2015Val)	6.2	1	NDD	Intellectual disability, Global developmental delay, Motor delay, Failure to thrive, Increased serum lactate, Infantile muscular hypotonia, Delayed myelination, Alaninuria	comphet	paternal & maternal	yes, Father with metabolic disorder, mother with epilepsy, thyroid disease, and suspected diabetes mellitus; maternal uncle with motor retardation
ADARB2	NM_018702.3:c.1570G>A p.(Glu524Lys)	NM_018702.3:c.914G>A p.(Ser305Asn)	6.2	2	NDD	Microcephaly, Hearing impairment, Autism, Intellectual disability, Spasticity, Global developmental delay, Cerebral calcification	comphet	paternal & maternal	no
OSBPL9	NM_148909.3:c.413_422del p.(Ser138Ilefs*16)		6.2	1	Epilepsy	Seizure, Generalized non-motor (absence) seizure, Generalized myoclonic seizure, Atypical absence seizure, Eyelid myoclonia seizure, Myoclonic seizure	het	unknown	no
GPM6B	NM_001001994.2: c.22A>G p.(Ile8Val)		6.2	3	NDD + Epilepsy	epileptic encephalopathy, therapy-resistant seizures, pain in legs	hemi	maternal	yes, sister also affected, 1 sister died in the age of 6 months
MTR	NM_000254.2:c.2812A>G p.(Ser938Gly)		6.2	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	het	unknown	no
NIT1	NM_001185092.1: c.244_256del p.(Phe83Hisfs*63)	NM_001185092.1:c.302T>C p.(Leu101Pro)	6.2	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	comphet	paternal & maternal	no

LENG8	NM_052925.3: c.2147G>A p.Arg716Gln		6.2	1	NDD	severe ID, mental deterioration, sleep disturbances, behavioral abnormality, hyperpigmented macules, EEG abnormalities	homo	paternal & maternal	2m, cousins I°
RSRC2	NM_023012.5: c.1271T>G p.(Phe424Cys)		6.1	1	NDD	Global developmental delay, Microcephaly, Agenesis of corpus callosum, Failure to thrive, Growth delay, EEG abnormality, Abnormal cry	het	de novo	no
PLXNA3	NM_017514.5: c.1015C>G p.(Leu339Val)		6.1	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	hemi	maternal	no
ZBTB21	NM_001098402.2: c.2088del p.(Lys696Asnfs*5)		6.1		NDD + epilepsy	(+) Seizure,(+) Global developmental delay,(+) Stereotypical hand wringing,(+) Muscular hypotonia	het	unknown	no
SEZ6L2	NM_001243332.1: c.1084G>A p.(Val362Met)	NM_001243332.1:c.85C>T p.(Pro29Ser)	6.1	1	Epilepsy	Seizures, status epilepticus, focal-onset seizure, EEG with spike-wave complexes, epilepsy not completely under control, cognitive deficiency, intellectual disability	comphet	paternal & maternal	no
POLR2A	NM_000937.4: c.4808G>A p.(Arg1603His)	NM_000937.4:c.778G>A p.(Val260Met)	6.1	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures	comphet	paternal & maternal	no
NCOA2	NM_006540.2: c.1454T>C p.(Met485Thr)	NM_006540.2:c.3509T>C p.(Met1170Thr)	6.1	1	NDD	Intellectual disability, Seizures, Encephalopathy, Cerebral atrophy, Intellectual disability, profound, EEG abnormality, Intellectual disability, severe, Cognitive impairment	comphet	paternal & maternal	no
FMNL3	NM_175736.4: c.2575C>T p.(Arg859Trp)		6.1	3	NDD	Short stature, microcephaly, mild intellectual disability, hyperopia	homo	unknown	yes, similary affected sister
IRAK1	NM_001025242.1: c.609T>G p.(Cys203Trp)		6.1	2	NDD + Epilepsy	Intellectual disability, Seizures, Generalized myoclonic seizures, Infantile onset	het	de novo	no
SCN11A	NM_014139.2: c.95C>T p.(Ala32Val)	NM_014139.2:c.2821G>A p.(Glu941Lys)	6.1	4	Epilepsy	Seizures, Generalized-onset seizure	comphet	paternal & maternal	no

ASIC1	NM_020039.3:c.1116T>A p.(Tyr372*)		6.1	1	NDD + Epilepsy	Behavioral abnormality, Autistic behavior, Delayed speech and language development, Seizure, Pyloric stenosis, Attention deficit hyperactivity disorder	het	unknown	no
CELSR3	NM_001407.2: c.8254T>C p.(Phe2752Leu)	NM_001407.2:c.79T>C p.(Phe27Leu)	6.1	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)	comphet	paternal & maternal	no
GRIN3B	NM_138690.1: c.1811C>T p.(Thr604Met)	NM_138690.1:c.2114A>C p.(Tyr705Ser)	6.1	1	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Hypsarrhythmia, Infantile onset,	comphet	paternal & maternal	N/A
NUSAP1	NM_016359.4:c.1213C>T p.(Gln405*)		6.1	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology	het	de novo	yes, maternal aunt and paternal uncle with seizures
NCOR2	NM_001077261.3: c.7241C>T p.(Ala2414Val)	NM_001077261.3:c.1520_1528dup p.(Gln507_Gln509dup)	6.1	1	NDD + Epilepsy	Balkenagenesie, Polymikrogyrie, Plexuszysten, retinale Auffälligkeiten	comphet	paternal & maternal	no
HMGXB3	NM_014983.2: c.2026C>T p.(Pro676Ser)		6.1	1	NDD	Delayed speech and language development, Intellectual disability, Global developmental delay, Expressive language delay	het	de novo	no
DBP	NM_001352.4: c.511G>T p.(Ala171Ser)		6.1	2	NDD	Global developmental delay, Intellectual disability, mild	het	de novo	no

CRYBG1	NM_001624.3: c.4489G>A p.(Val1497Ile)	6.1	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 hypermetropia	het	de novo	no
GRK3	NM_005160.3: c.916G>T p.(Glu306*)	6.1	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	het	maternal	yes, Mother and maternal half-sister have intellectual disability
GPKOW	NM_015698.5: c.511A>G p.(Met171Val)	6.1	1	NDD + Epilepsy	ID, focal epilepsy, motor delay, speech delay, autism, behavioral abnormalities	hemi	maternal	yes, paternal half brother with learning difficulties, maternal half sister with epilepsy in puberty, maternal cousin with ID

HCK	NM_002110.3: c.1547C>A p.(Thr516Lys)	6.1	2	NDD + Epilepsy	Pallister Hall syndrome with hand and foot polydactyly, cleft palate, seizures, global developmental delay, 4-5 finger syndactyly, hamartoma	het	de novo	no
MOXD1	NM_015529.3: c.350A>G p.(His117Arg)	6.0	3	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	homo	paternal & maternal	yes, affected uncle het
PCSK1N	NM_013271.4: c.491T>C p.(Leu164Pro)	6.0	1	Epilepsy	familiar epilepsy, speech delay, ADHS	hemi, in multiple	maternal	yes, mother with myoclonus, several family members died due to fever cramps (maternal grand-grandmother, two maternal uncles and one maternal aunt), several family members with epi-leptic seizures (one maternal aunt and three brothers)
CHMP7	NM_152272.3: c.214C>A p.(Leu72Met)	6.0	3	NDD + Epilepsy	Autism, Seizures, Global developmental delay, Motor delay, Absent speech, Epileptic encephalopathy	het	de novo	N/A
ATP13A4	NM_032279.3:c.826 G>A p.(Glu276Lys)	6.0	2	NDD	Long palpebral fissure, Prominent fingertip pads, Intellectual disability, Large fleshy ears	homo	paternal & maternal	no

GRIN3B	NM_138690.2: c.2114A>G p.(Tyr705Cys)	NM_138690.2:c.2314G>A p.(Gly772Ser)	6.0	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	comphet	paternal & maternal	no
DIP2A	NM_015151.3: c.410C>T p.(Ser137Leu)	NM_015151.3:c.2476G>A p.(Ala826Thr)	6.0	2	NDD	Intellectual disability, Intellectual disability, mild, Intellectual disability, moderate, Increased body weight, Increased adipose tissue	comphet	paternal & maternal	no
ATP2B3	NM_001001344.2:c.3530C>T p.(Pro1177Leu)		6.0	2	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Global developmental delay, Motor delay, Encopresis	hemi	maternal	no
CASKIN1	NM_020764.3 : c.3091C>T p.(Arg1031Cys)		6.0	1	NDD	Speech delay, mild intellectual disability, non-verbal IQ of 57, no prominent facial dysmorphism, stereotypic behavior	homo	paternal & maternal	consanguinity
CFAP74	NM_001304360.1: c.3409del p.(Gln1137Argfs*37)		6.0	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	het	de novo	no
KIRREL2	NM_032123.6: c.1275del p.(Pro425Profs*41)		6.0	1	Epilepsy	Seizures, Generalized tonic-clonic seizures, Absence seizures, Generalized myoclonic seizures, Episodic vomiting, Epileptic spasms, Myoclonic atonic seizures, Epileptic encephalopathy	het	paternal	yes, father affected
HSD17B6	NM_003725.3: c.440G>A p.(Ser147Asn)		6.0	4	Epilepsy	Seizures, Generalized-onset seizure	het	de novo	no
FLYWCH1	NM_001308068.1: c.2112-3T>G p.?	NM_001308068.1:c.1111A>T p.(Ser371Cys)	6.0	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	comphet	paternal & maternal	yes, maternal uncle with epilepsy and ID

PPP1R37	NM_019121.1: c.509C>T p.(Ser170Phe)		6.0	2	NDD	Bilateral cryptorchidism, Short stature, Epileptic encephalopathy, Microcephaly	het	de novo	no
FAT1	NM_005245.3: c.11017G>C p.(Val3673Leu)	NM_005245.3:c.6079C>T p.(Arg2027Cys)	6.0	2	NDD + Epilepsy	Seizures, Global developmental delay, Generalized-onset seizure, Periventricular leukomalacia	comphet	paternal & maternal	no
MRPL15	NM_014175.3: c.743C>T p.(Thr248Ile)		6.0	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 hypotonia	homo	paternal & maternal	no
AFDN	NM_001207008.1: c.436A>G p.(Lys146Glu)		6.0	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	het	paternal	yes, father affected
TENM1	NM_001163278.1: c.5977A>T p.(Thr1993Ser)		6.0	1	NDD	Delayed speech and language development, Intellectual disability, Global developmental delay	hemi	maternal	no
ARHGEF10L	NM_018125.3: c.354_355delCCinsTT p.(Arg119Trp)		6.0	1	Epilepsy	Seizures, Ataxia, Spasticity, Focal clonic seizures, Myoclonic spasms, Generalized dystonia, Focal-onset seizure, Focal myoclonic seizures	homo	paternal & maternal	yes, Father possibly affected (no further information)
DRG1	NM_004147.3: c.43-1G>T p.?		5.9	3	NDD + Epilepsy	Autism, Intellectual disability, Seizures, Global developmental delay, Poor speech, Focal seizures	het	de novo	no
PCDH11X	NM_032968.4: c.1688A>G p.(Gln563Arg)		5.9		Epilepsie	(+) 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	hemi	maternal	no
SETD1B	NM_015048.1: c.1743C>G p.(Asp581Glu)	NM_015048.1:c.2999G>A p.(Arg1000Gln)	5.9	2	NDD	Macrocephaly, hypoplasia of the corpus callosum, suspected developmental delay, right hemiparesis	comphet	paternal & maternal	no

SCRN1	NM_001145514.1: c.1106A>G p.(Lys369Arg)		5.9	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)	homo	paternal & maternal	no
PCLO	NM_033026.5 : c.13206G>T p.(Gln4402His)	NM_033026.5:c.1297G>A p.(Ala433Thr)	5.9	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 hypotonia	comphet	paternal & maternal	no
RAB11FIP2	NM_001330167.1: c.1334T>C p.(Met445Thr)		5.9	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	het	de novo	no
DHX36	NM_020865.2: c.800_802del p.(Ile267del)		5.9	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	het	de novo	no
BAIAP3	NM_001199096.1: c.892G>T p.(Gly298Trp)		5.9	2	NDD	Global developmental delay, Microcephaly, Cryptorchidism, Ptosis, Short stature, Short phalanx of finger, Frontal hirsutism, Arachnoid cyst	het	de novo	no
ARSH	NM_001011719.1: c.339A>G p.(Ile113Met)		5.9	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	het	de novo	no

LUC7L	NM_018032.3: c.614G>A p.(Arg205His)		5.9	2	NDD + Epilepsy	Seizures, Global developmental delay	het	de novo	no
NRIP1	NM_003489.3: c.2077G>T p.(Gly693Cys)		5.9	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 impairment	het	de novo	no
GET4	NM_015949.2: c.491A>G p.(Tyr164Cys)		5.9	3	NDD	hydrocephalus, intellectual disability, spastic paraplegia, global developmental delay, agenesis of corpus callosum, spastic paraparesis, intellectual disability, severe, cerebral palsy	homo	paternal & maternal	no
CASZ1	NM_001079843.2: c.4004G>A p.(Arg1335His)		5.9	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia	homo	paternal & maternal	yes,affected monozygotic triplet, parents konsaguin
ZZEF1	NM_015113.3: c.1580C>T p.(Pro527Leu)		5.9	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Specific learning disability, Absence seizure, Generalized-onset seizure, Intellectual disability, borderline, Attention deficit hyperactivity disorder	het	de novo	no
LRCH3	NM_032773.3: c.761A>G p.Gln254Arg		5.8	1	NDD + Epilepsy	severe ID, seizures, muscular hypotonia, cardiac malformation, cerebral atrophy	homo	paternal & maternal	3fm, consanguinity
DNAH3	NM_017539.2: c.7420A>T p.(Lys2474*)	NM_017539.2: c.5287G>A p.(Val1763Met)	5.8		NDD	(+) Global developmental delay,(+) Delayed speech and language development,(+) Autistic behavior,(+) Hearing impairment,(+) Developmental regression	comphet	paternal & maternal	no
ADAMTSL1	NM_001040272.5: c.1316A>G p.(Lys439Arg)		5.8	1	NDD + Epilepsy	Global developmental delay, dystonic movements, abnormal EEG, epilepsy, microcephaly, clinodactyly of the 5th finger, pectus excavatum	het	de novo	no
SCN7A	NM_002976.3:c.2932A>G p.(Ile978Val)		5.8	3	Epilepsy	Epilepsy with generalized tonic-clonic seizures, ED 10/2019, microcephaly	homo	maternal	no
CD200	NM_001004196.3: c.161C>A p.(Thr54Lys)		5.8	3	NDD + Epilepsy	Refractory epilepsy, epileptic encephalopathy, psychomotor retardation, microcephaly, short stature, facial dysmorphisms, nephrocalcinosis	het	de novo	no

CLASP1	NM_015282.2:c.4142T>A p.(Phe1381Tyr)		5.8	NDD	Cardio	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart	het	unknown	no
CENPI	NM_006733.3: c.652C>T p.(Arg218Cys)		5.8	3	Epilepsie	atypic absence seizure, strartle-induced seizure, attention deficit hyperactivity disorder, seizure	hemi	maternal	no
PTPRS	NM_002850.3: c.4810G>A p.(Ala1604Thr)	NM_002850.3: c.4453G>A p.(Ala1485Thr)	5.8		NDD	(+) Short stature,(+) Global developmental delay,(+) Intellectual disability,(+) Microcephaly	comphet	paternal & maternal	no
GIGYF1	NM_022574.4: c.1701G>T p.(Gln567His)		5.8	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Status epilepticus, Intellectual disability, moderate, Intellectual disability, borderline, Intellectual disability, severe, Cognitive impairment, Epileptic encephalopathy	het	de novo	no
CHD5	NM_015557.2: c.776C>G p.(Ser259Cys)	NM_015557.2:c.3650C>T p.(Thr1217Ile)	5.8	2	NDD	Delayed speech and language development, Intellectual disability	comphet	paternal & maternal	no
RNF10	NM_001330474.1: c.850C>T p.(His284Tyr)		5.8	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 hypotonia	homo	paternal & maternal	no
KCNG4	NM_172347.2:c.1022C>T p.(Ala341Val)		5.8	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure	homo	paternal & maternal	no
URB2	NM_014777.2: c.1949del p.(Gly650Valfs*2)		5.8	2	NDD + Epilepsy	Seizures, Myoclonic absences, developmental delay	het	de novo	yes, mother with epilepsy in childhood
PSD3	NM_015310.3: c.3092A>G p.(Glu1031Gly)	NM_015310.3: c.2929-3C>T p.?	5.8	2	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Focal seizures, Intellectual disability, severe	comphet	paternal & maternal	no
MYOF	NM_013451.3:c.3511C>T p.(Arg1171Trp)	NM_013451.3:c.4268del p.(Pro1423Hisfs*21)	5.7	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia	comphet	paternal & maternal	no

CAPS2	NM_032606.3: c.525+1G>A p.?		5.7	1	NDD + Epilepsy	mental retardation, epilepsy, no speech development, deafness, short stature	het	de novo	no
TIAM2	NM_012454.3: c.4679_4681dup p.(Asn1560_Leu1561insHis)		5.7	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia	homo	paternal & maternal	yes,affected monozygotic triplet, parents konsaguin
LCTL	NM_207338.3: c.692_693dup		5.7	2	NDD + Epilepsy	epileptic encephalopathy, seizures	het	de novo	no
CTSB	NM_001908.3: c.444C>T p.(=)		5.7	4	NDD	Intellectual disability, Global developmental delay	homo	paternal & maternal	no
DNAJC27	NM_016544.2: c.422del p.(His141Leufs*4)		5.7	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	het	maternal	yes, Mother and maternal half-sister have intellectual disability
NCOA7	NM_001122842.2: c.2660+2T>A p.?		5.7	1	NDD	Behavioral abnormality, Delayed speech and language development, Global developmental delay	het	de novo	no
LAMA5	NM_005560.4: c.10753G>T p.(Asp3585Tyr)	NM_005560.4:c.1390G>A p.(Gly464Ser)	5.7	1	NDD + Epilepsy	Abnormality of the head, Microcephaly, Seizures, Postnatal microcephaly, Loss of consciousness, Atonic seizures	comphet	paternal & maternal	no
TMEM147	NM_032635.3: c.344+5G>A p.?		5.7	1	NDD	very severe ID, impaired vision, joint contractures	homo	paternal & maternal	3fm, >cousins III°
ABCC12	NM_033226.2: c.796G>A p.(Gly266Arg)	NM_033226.2:c.442del p.(Ile148Serfs*20)	5.7	3	NDD + Epilepsy	Therapy-resistant epilepsy since the age of two, Epileptic encephalopathy	comphet	paternal & maternal	no
GRINA	NM_000837.1: c.967-6C>T p.?		5.7	2	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,	homo	paternal & maternal	yes, one affected brother, but unclear if the same phenotype, consanguinity
WDR13	NM_001347217.2: c.194G>A p.(Arg65His)		5.7	1	NDD	Global developmental delay, EEG abnormality, Carious teeth, Finger clinodactyly, Decreased head circumference, Intellectual disability	hemi	unknown	no

SNX6	NM_021249.4: c.586C>T p.(Arg196*)		5.6	2	NDD	mild ID, facial dysmorphisms, Missing anterior teeth in the lower jaw, simian crease, dry skin, narrow mouth, behavioral abnormalities	homo	paternal & maternal	yes, Mother and sister also with missing front teeth, brother of mother with ID
NOL4	NM_003787.4:c.1A>C p.0?		5.6	1	NDD	Microcephaly, Aggressive behavior, Intellectual disability, Intellectual disability, mild, Abnormal aggressive, impulsive or violent behavior	het	maternal	no
ITPKA	NM_002220.2: c.1093G>A p.(Gly365Arg)		5.6	2	NDD + Epilepsy	Macrocephaly, Seizures, Global developmental delay, Epileptic encephalopathy	homo	paternal & maternal	no
SUPV3L1	NM_003171.4: c.1931G>A p.(Arg644Gln)	NM_003171.4: c.2358C>G p.(Asp786Glu)	5.6		NDD+epilepsy	(+) Global developmental delay,(+) Focal-onset seizure,(+) Abnormality of the nasal alae,(+) Poor eye contact	comphet	paternal & maternal	N/A
FAM214B	NM_001317991.1: c.1012C>G p.(Pro338Ala)		5.6	3	Epilepsie	(+) Focal tonic seizure,(+) EEG with focal sharp waves,(+) Nocturnal seizures,(-) Brain imaging abnormality	homo	paternal & maternal	maternal grandmother with epilepsy, café au lait spots
TMEM232	NM_001039763.3:c.884A>G p.(Gln295Arg)		5.6	1	Epilepsy	epilepsy, movement disorder, syncope, myoclonia, pathological waking EEG, cystic, malformation of the right kidney	het	de novo	no
ALDH8A1	NM_022568.3: c.160G>T p.(Ala54Ser)		5.6	1	NDD	Macrocephaly, Global developmental delay, Hepatosplenomegaly, Hypertriglyceridemia, Hepatomegaly, Recurrent infections	het	de novo	no
FAT1	NM_005245.3: c.2137A>G p.(Ile713Val)	NM_005245.3:c.9440T>G p.(Val3147Gly)	5.6	2	NDD + Epilepsy	Seizures, Status epilepticus, Hypsarrhythmia,	comphet	paternal & maternal	no
RHBDF1	NM_022450.3: c.1082G>A p.(Arg361His)		5.6	3	NDD	global developmental delay, absent speech, muscular hypotonia, austism spectrum disorder, microcephaly	het	de novo	no
EXOC4	NM_001037126.1: c.472-6T>C p.?	NM_001037126.1:c.860C>T p.(Ala287Val)	5.6	1	NDD + Epilepsy	early onset epilepsy, mild developmental delay, decreased glucose in liquor, behavarioal abnormalities	comphet	paternal & maternal	N/A

MED22	NM_133640.4: c.397_399del p.(Glu133del)		5.6	2	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,	homo	paternal & maternal	yes, one affected brother, but unclear if the same phenotype, consanguinity
FAM199X	NM_207318.3: c.961T>A p.(Ser321Thr)		5.6	1	Epilepsy	Aggressive behavior, Delayed speech and language development, Seizures, Global developmental delay, Absent speech, EEG abnormality, Focal impaired awareness seizure, Focal-onset seizure	hemi	maternal	no
DBN1	NM_080881.2:c.1452C>G p.(Asn484Lys)	NM_080881.2:c.1663_1664delTCinsCT p.(Pro555Leu)	5.6	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Pes planus, Focal tonic seizure, Cognitive impairment	comphet	paternal & maternal	no
CSTF2	NM_001306206.1: c.724G>A p.(Ala242Thr)		5.6	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	hemi	maternal	yes, two similarly affected brothers
STX4	NM_004604.4: c.118_120del p.(Glu40del)		5.6	3	Epilepsie	(+) Focal tonic seizure,(+) EEG with focal sharp waves,(+) Nocturnal seizures,(-) Brain imaging abnormality	homo	paternal & maternal	maternal grandmother with epilepsy, café au lait spots
HS6ST2	NM_001077188.1: c.853T>G p.(Trp285Gly)		5.6	1	NDD	global developmental delay, focal epilepsy, absent speech, Delayed gross motor development, Tetraparesis, Facial palsy	hemi	maternal	no
BOK	NM_032515.4: c.356C>T p.(Thr119Met)		5.6	1	NDD	Microcephaly, Underdeveloped nasal alae, Strabismus, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Postnatal microcephaly	het	de novo	no

AASDH	NM_181806.3: c.2908-5_2908-4insGTT p.?	NM_181806.3:c.3220dup p.(Leu1074Profs*10)	5.6	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 dysphagia	comphet	paternal & maternal	no
RHBDL1	NM_001318733.1: c.1127C>A p.(Ala376Glu)		5.6	1	Epilepsie	Focal-onset seizure, Seizure, Encephalopathy, Focal cortical dysplasia	het	de novo	one sibling with trisomy 21, one sibling with malformation of thumb, parents are healthy
PRDX4	NM_006406.1: c.724G>A p.(Gly242Arg)		5.5	2	NDD + Epilepsy	Seizures, Global developmental delay	hemi	maternal	no
KCTD18	NM_001321547.1: c.875C>T p.Ser292Leu		5.5	1	NDD	moderate ID, short stature, microcephaly, dislocated hips	homo	paternal & maternal	2f, >cousins I°
PIK3AP1	NM_152309.3: c.601A>T p.(Lys201*)		5.5	1	NDD + epilepsy	Epileptic encephalopathy, Cognitive impairment, Microcephaly, Short stature, Febrile seizure	het	unknown	no
SVEP1	NM_153366.4: c.6371T>C p.(Ile2124Thr)		5.5	3	Epilepsie	atypic absence seizure, strartle-induced seizure, attention deficit hyperactivity disorder, seizure	homo	paternal & maternal	no
SRRM4	NM_194286.3: c.1295C>T p.(Ser432Phe)	NM_194286.3: c.1172G>A p.(Arg391His)	5.5	3	Epilepsie	therapy-resistant epilepsy	comphet	paternal & maternal	no
INTS1	NM_001080453.2: c.6248T>G p.(Phe2083Cys)	NM_001080453.2:c.5272A>G p.(Ile1758Val)	5.5	2	NDD + Epilepsy	GDD, ataxia, muscular hypotonia, microcephaly, EEG abnormalities, brain volume reduction	comphet	paternal & maternal	no

USP34	NM_014709.4: c.7561G>C p.(Val2521Leu)	NM_014709.4: c.4229C>T p.(Ala1410Val)	5.5		NDD	(+) Intellectual disability,(+) Hyperactivity,(+) Autistic behavior	comphet	paternal & maternal	no
RENBP	NM_002910.5: c.695G>A p.(Gly232Glu)		5.5	1	Epilepsy	Epilepsy, susceptibility to fall	hemi	maternal	no
USP20	NM_001008563.4: c.582del p.(Lys194Asnfs*46)		5.5	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	het	de novo	yes, sister is possibly affected
ANKFY1	NM_001257999.1: c.1966G>A p.(Ala656Thr)		5.5	3	NDD + Epilepsy	Autism, Seizures, Global developmental delay, Motor delay, Absent speech, Epileptic encephalopathy	homo	paternal & maternal	N/A
KCND1	NM_004979.5: c.343G>A p.(Asp115Asn)		5.5	1	NDD + Epilepsy	Epilepsy with absences and eyelid myoclonias, normal cMRI, EEG abnormalities, IQ 85 (low normal), speech delay, obstipation	hemi	de novo	no
SP9	NM_001145250.1: c.1133A>G p.(Glu378Gly)		5.5	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	het	de novo	no
PCNX2	NM_014801.3: c.3846C>A p.(Asp1282Glu)		5.5	2	NDD	Visual loss, Optic nerve hypoplasia, Global developmental delay, Hypoglycemia, Abnormality of optic chiasm morphology	het	de novo	no
ZFYVE16	NM_014733.4: c.2570C>T p.(Pro857Leu)		5.5	2	NDD + Epilepsy	Global developmental delay, febrile seizures, muscular hypotonia	homo	paternal & maternal	no
HELZ2	NM_001037335.2: c.6691+4C>T p.?		5.5	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	homo	paternal & maternal	no

MAST3	NM_015016.2: c.3367C>T p.(Arg1123*)		5.5		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	het	unknown	no
GLRA4	NM_001024452.2:c. 39_41del p.(Leu14del)		5.5	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	hemi	unknown	no
ZNF219	NM_001101672.1: c.673_678del p.(Ala225_Pro226del)		5.5	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	het	de novo	no
INO80	NM_017553.2: c.1294G>A p.(Gly432Arg)		5.5	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	het	unknown	no
FHOD3	NM_001281740.2:c. 1836-2A>G p.?		5.5	1	NDD + Epilepsy	Entwicklungsstörung, Epilepsie (Absencen, Grand-mal-Anfälle)	het	unknown	no
CNTN6	NM_014461.3: c.275A>T p.(Asp92Val)	NM_014461.3:c.2573G>A p.(Ser858Asn)	5.5	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay	comphet	paternal & maternal	no

NOVA2	NM_002516.3: c.1267G>C p.(Gly423Arg)		5.5	2	NDD	Microcephaly, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Scoliosis, Short stature, Cognitive impairment	het	maternal	yes, Mother with similar symptoms and epilepsy, maternal grandfather with epilepsy, two maternal uncles with epilepsy
PHACTR3	NM_001199505.1: c.17G>T p.(Gly6Val)		5.5	1	NDD	Intellectual disability, Global developmental delay	het	de novo	no
SEL1L	NM_001244984.1: c.149C>T p.(Thr50Ile)		5.5	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	het	de novo	no
NUP188	NM_015354.2:c.17G>C p.(Gly6Ala)	NM_015354.2:c.2917C>T p.(Arg973*)	5.5	3	NDD + Epilepsy	Intellectual disability, Seizures, Status epilepticus	comphet	paternal & maternal	no
ZCRB1	NM_033114.3: c.78G>C p.(Leu26Phe)		5.4	3	NDD + Epilepsy	Refractory epilepsy, epileptic encephalopathy, psychomotor retardation, microcephaly, short stature, facial dysmorphisms, nephrocalcinosis	het	de novo	no
BHLHE41	NM_030762.2: c.1222G>C p.(Ala408Pro)		5.4	1	NDD + Epilepsy	Seizures, Febrile seizures, Childhood onset, Epileptic encephalopathy	het	de novo	yes, Mother: Epilepsy at the age of 3-6 years
POU3F2	NM_005604.4: c.664C>T p.(Pro222Ser)		5.4		Neuro	Leukodystrophy, Leukoencephalopathy, Attention deficit hyperactivity disorder, Neurological speech impairment, Neonatal asphyxia, Gait disturbance	het	paternal	father with suspected leukencephalopathy and neurogenic bladder dysfunction
BTAF1	NM_003972.3: c.2662G>A p.(Glu888Lys)		5.4		NDD + epilepsy	(+) Global developmental delay,(+) Absent speech,(+) Seizure,(+) Intellectual disability	het	het	no

MMS22L	NM_198468.2: c.2679+1G>A p.?	NM_198468.2:c.268 A>G p.(Arg90Gly)	5.4	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech	comphet	paternal & maternal	no
LAMA5	NM_005560.4:c.863 2G>A p.(Val2878Ile)	NM_005560.4:c.657 8G>A p.(Arg2193His)	5.4	2	Epilepsy	Seizure, Status epilepticus, EEG abnormality, Focal impaired awareness seizure, Focal-onset seizure, EEG with focal spike waves	comphet	paternal & maternal	no
FAT3	NM_001008781.2: c.763C>G p.(His255Asp)	NM_001008781.2: c.11140A>G p.(Lys3714Glu)	5.4		NDD + epilepsy	Atypical absence seizure, Myoclonic seizure, Epileptic encephalopathy, Myoclonus, EEG abnormality, Hyperammonemia, Abnormal vitamin B12 level, normal development	comphet	paternal & maternal	no
TACC2	NM_206862.3: c.65_66insCCTC p.(Gln23Leufs*22)	NM_206862.3:c.780 1C>T p.(Pro2601Ser)	5.4	3	NDD	hydrocephalus, intellectual disability, spastic paraplegia, global developmental delay, agenesis of corpus callosum, spastic paraparesis, intellectual disability, severe, cerebral palsy	comphet	paternal & maternal	no
DLC1	NM_182643.2:c.609 A>C p.(Lys203Asn)		5.4	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia	homo	paternal & maternal	no
WDR3	NM_006784.2: c.989+1G>C p.?	NM_006784.2:c.155 5T>A p.(Ser519Thr)	5.4	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay	comphet	paternal & maternal	no
ZFHX3	NM_006885.3: c.5449G>T p.(Val1817Leu)	NM_006885.3: c.2321C>T p.(Ala774Val)	5.4	1	NDD + epilepsy		comphet	paternal & maternal	no
CENPV	NM_181716.2: c.75_92del p.(Ala26_Ala31del)		5.4	3	NDD + Epilepsy	mild global developmental delay, seizures, heterotopia, oral cleft, tall stature, obesity	het	de novo	no
VPS52	NM_022553.5: c.10G>A p.(Ala4Thr)	NM_022553.5:c.92C >T p.(Ala31Val)	5.4	3	NDD + Epilepsy	neonatal epileptic encephalopathy	comphet	paternal & maternal	no
KCTD8	NM_198353.2: c.82G>C p.(Ala28Pro)		5.4	1	NDD	Regressive global developmental delay with intellectual disability, attention deficit disorder, dysplasia of the corpus callosum, obesity grade 1	het	de novo	no

NAV2	NM_001244963.1: c.2486C>T p.(Pro829Leu)	NM_001244963.1:c.7137+3G>A p.?	5.4	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	comphet	paternal & maternal	no
DMRT3	NM_021240.3: c.917C>T p.(Ala306Val)		5.4	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	het	de novo	no
ACAD10	NM_001136538.1: c.1670C>G p.(Pro557Arg)	NM_001136538.1:c.3230A>G p.(His1077Arg)	5.4	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	comphet	paternal & maternal	yes, Father with similar symptom, grandmother (paternal): mental retardation; Grandmother and great-grandmother maternal: Leukemia in old age
ANKDD1A	NM_182703.5: c.1470G>C p.(Arg490Ser)		5.4	1	NDD	(+) Delayed speech and language development,(+) Diminished ability to concentrate,(+) Cognitive impairment,(+) Hearing impairment	het	de novo	two cousins of the mother with autism spectrum disorder
H2BC4	NM_003526.2: c.154G>T p.(Asp52Tyr)		5.4	2	NDD	Spastic tetraparesis, Optic atrophy, Periventricular leukomalacia, Microcephaly, Global developmental delay	het	de novo	no
ZNF692	NM_001136036.2: c.70C>G p.(Gln24Glu)		5.4	2	NDD + Epilepsy	Seizures, Global developmental delay, Generalized-onset seizure, Periventricular leukomalacia	het	de novo	no

NUMBL	NM_004756.4:c.1193C>A p.(Pro398His)		5.4	2	NDD	Long palpebral fissure, Prominent fingertip pads, Intellectual disability, Large fleshy ears	homo	paternal & maternal	no
SEZ6L2	NM_001243332.1: c.910A>G p.(Thr304Ala)		5.4	3	NDD	Autism (Asperger), Autistic behavior, Depressivity, Macrocephaly	het	maternal	sister is similarly affected (global DD, autism, macrocephaly, muscular hypotonia)
NIF3L1	NM_001136039.2:c.860C>T p.(Thr287Ile)		5.4	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia	homo	paternal & maternal	no
CHD5	NM_015557.2: c.5003-5G>A p.?	NM_015557.2:c.5249C>T p.(Thr1750Met)	5.3	3	NDD	Autism, Intellectual disability, Global developmental delay	comphet	paternal & maternal	no
FREM3	NM_001168235.1: c.728del p.(Glu243Glyfs*25)	NM_001168235.1:c.5401C>T p.(Leu1801Phe)	5.3	2	Epilepsy	Seizures, Encephalopathy, Focal seizures, Encephalitis	comphet	paternal & maternal	no
CSMD1	NM_033225.5: c.7327A>G p.(Ile2443Val)	NM_033225.5:c.8444A>C p.(Glu2815Ala)	5.3	3	NDD + Epilepsy	mild global developmental delay, seizures, heterotopia, oral cleft, tall stature, obesity	comphet	paternal & maternal	no
ZC3H3	NM_015117.2:c.1595C>T p.(Thr532Ile)		5.3	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	het	de novo	no
TMEM199	NM_152464.2: c.5C>T p.(Ala2Val)		5.3	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 impairment	het	de novo	no
ATP6V0A1	NM_001130020.1: c.2222G>A p.(Arg741Gln)		5.3	2	NDD + epilepsy	(+) Seizure,(+) Large for gestational age,(+) Microcephaly,(+) Global developmental delay,(+) Muscular hypotonia	het	de novo	no

NR2F6	NM_005234.3:		5.3	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 septal aneurysm	het	de novo	no
DHRS7	NM_016029.3: c.475A>G p.(Ile159Val)		5.3	2	NDD + Epilepsy	epilepsy with Lennox-Gastaut syndrome and complex retardation	het	de novo	no
PTP4A1	NM_003463.4: c.8G>A p.(Arg3Gln)		5.3	1	NDD	mental retardation, autism	het	de novo	no
UNC79	NM_020818.4: c.3857-691A>G p.(=)	NM_020818.4:c.1547C>T p.(Ser516Leu)	5.3	2	NDD	Cleft palate, Panhypopituitarism, Intellectual disability, Patent ductus arteriosus, Facial cleft, Scoliosis, Short stature, Median cleft lip and palate	comphet	paternal & maternal	no
LAMA5	NM_005560.4: c.6659G>T p.(Arg2220Leu)	NM_005560.4:c.1246C>G p.(Pro416Ala)	5.3	2	NDD + Epilepsy	Seizures, Global developmental delay, Episodic ataxia	comphet	paternal & maternal	no
TANC1	NM_001145909.1: c.10G>C p.(Ala4Pro)	NM_001145909.1:c.1007G>A p.(Arg336Gln)	5.3	3	NDD	global developmental delay, absent speech, muscular hypotonia, austism spectrum disorder, microcephaly	comphet	paternal & maternal	no
ELMOD2	NM_153702.3: c.580C>T p.(Arg194Cys)		5.3	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	het	de novo	no
MAP3K15	NM_001001671.3: c.2037dup p.(Ile680Hisfs*9)		5.3	5	Epilepsy	Seizures, Generalized myoclonic seizures	hemi	maternal	no

TMEM94	NM_001321148.1:c.1976A>C p.(Gln659Pro)		5.3	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart	het	unknown	no
PAPSS1	NM_005443.4: c.1672G>A p.(Val558Ile)		5.3	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Cerebellar vermis atrophy, Cognitive impairment	homo	paternal & maternal	no
ZBTB45	NM_001316978.1:c.655G>A p.(Asp219Asn)		5.3	2	NDD	Inguinal hernia, Intellectual disability, Global developmental delay, Small for gestational age, Penile hypospadias, Short stature, Decreased body weight	het	de novo	no
PON1	NM_000446.5: c.717G>C p.(Glu239Asp)		5.3	4	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay	het	de novo	no
ZNF319	NM_020807.2:c.654_655del p.(Arg219Alafs*2)		5.3	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	het	unknown	no
ZFYVE9	NM_004799.3: c.3220C>A p.(Leu1074Met)	NM_004799.3: c.4124A>T p.(Tyr1375Phe)	5.3	2	NDD + epilepsy	Neonatal hypoglycemia, Seizure, Global developmental delay	comphet	paternal & maternal	no
GEMIN5	NM_015465.4: c.1627A>G p.(Ser543Gly)	NM_015465.4:c.851G>A p.(Arg284His)	5.3	2	NDD	Cryptorchidism, Microcephaly, Global developmental delay, Motor delay, Growth delay, Intrauterine growth retardation	comphet	paternal & maternal	no
ATP2B4	NM_001001396.2: c.2819A>G p.(Lys940Arg)		5.3	3	NDD + Epilepsy	Specific learning disability, Absence seizures, Cortical dysplasia, EEG with continuous slow activity, Seizures	homo	paternal & maternal	no
SPTB	NM_001024858.2: c.610G>A p.(Asp204Asn)	NM_001024858.2:c.5063A>G p.(Asn1688Ser)	5.2	1	NDD	Global developmental delay, Leukopenia, Leukemia, Acute lymphoblastic leukemia	comphet	paternal & maternal	no
ZC3H12B	NM_001010888.3: c.899A>G p.(Asn300Ser)		5.2	3	NDD + Epilepsy	Intellectual disability, Seizures, Ataxia	hemi	maternal	no

ZNF319	NM_020807.2: c.1294G>C p.(Val432Leu)		5.2	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	het	de novo	no
TNR	NM_003285.2: c.3659C>T p.(Ser1220Phe)	NM_003285.2:c.496 A>G p.(Thr166Ala)	5.2	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	comphet	paternal & maternal	no
ZFP91	NM_053023.4: c.172C>T p.(Arg58Trp)		5.2	3	NDD + Epilepsy	neonatal epileptic encephalopathy	het	de novo	no
MARVELD3	NM_001017967.3: c.1168G>A p.(Gly390Ser)		5.2	1	NDD	Autistic behavior, Intellectual disability, Global developmental delay, Obesity, Polyphagia, Developmental stagnation, Retractile testis, Cognitive impairment	het	de novo	no
ZDHHC2	NM_016353.5: c.47_52del p.(Arg16_Val17del)		5.2	2	Epilepsy	(+) Myoclonic seizure,(+) EEG with spike-wave complexes, suspected focal cortical dysplasia frontal right	het	de novo	no
NCAPH	NM_001281710.1: c.563-4T>G p.?	NM_001281710.1:c. 667G>A p.(Glu223Lys)	5.2	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 impairment	comphet	paternal & maternal	no
IQCH	NM_001031715.2: c.2552T>C p.(Leu851Pro)		5.2	1	NDD	GDD, bland-white-garland-syndrome, facial dysmorphisms, cleft palate, sudden cardiac arrest at the age of 3 months, hemi spastic	het	de novo	no
ARHGAP4	NM_001666.4: c.301C>T p.(His101Tyr)		5.2	3	NDD + Epilepsy	Hydrocephalus, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Aqueductal stenosis	hemi	maternal	no
TTC3	NM_001320703.1:c. 3970G>A p.(Glu1324Lys)		5.2	1	NDD	Abnormality of the kidney, Global developmental delay, Hip dysplasia, Short stature	het	de novo	no

CDH13	NM_001220488.1:c.2228G>A p.(Arg743His)	NM_001220488.1:c.1505C>T p.(Ser502Phe)	5.2	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech	comphet	paternal & maternal	no
GABRE	NM_004961.3: c.41T>C p.(Leu14Ser)		5.2	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	hemi	maternal	no
PCDHA11	NM_018902.4: c.88C>T p.(Gln30*)		5.2	2	NDD + Epilepsy	epilepsy with Lennox-Gastaut syndrome and complex retardation	het	de novo	no
KLHL17	NM_198317.2: c.1568C>T p.(Ala523Val)		5.2	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures	homo	paternal & maternal	no
SLC29A1	NM_001078174.1: c.766+5G>A p.?	NM_001078174.1:c.1357C>T p.(Arg453Trp)	5.2	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 encephalopathy	comphet	paternal & maternal	no
NUSAP1	NM_001243142.1: c.808C>T p.(Arg270Cys)		5.2	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	homo	paternal & maternal	no
DNAJC17	NM_018163.2: c.273G>T p.(Glu91Asp)		5.2	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	het	de novo	no

ANO4	NM_178826.3: c.868G>A p.(Ala290Thr)	5.2	2	Epilepsy	Seizures, Focal seizures, Multifocal epileptiform discharges, EEG with focal epileptiform discharges, Polymorphic focal epileptiform discharges, EEG with focal spikes	homo	paternal & maternal	no
ATP8B4	NM_024837.3: c.2698-2A>G p.?	5.2	2	NDD + epilepsy	mild global developmental delay, febrile seizure (within the age range of 3 months to 6 years)	het	de novo	yes, father with fever-associated seizures in
RNF144A	NM_014746.4:c.428 G>C p.(Cys143Ser)	5.2	3	Epilepsy	Epilepsy with generalized tonic-clonic seizures, ED 10/2019, microcephaly	homo	maternal	no
AQP6	NM_001652.3: c.146C>T p.(Pro49Leu)	5.2	2	NDD	Bilateral cryptorchidism, Short stature, Epileptic encephalopathy, Microcephaly	het	de novo	no
UTP11	NM_016037.3: c.230A>G p.(Asp77Gly)	5.2	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	het	de novo	no
ANKRD6	NM_001242809.1: NM_001242809.1:c. 1667C>T p.(Pro556Leu)	5.1	1	NDD	Dandy-Walker malformation, Omphalocele, Occipital encephalocele, Meningocele	het	de novo	no
SLITRK4	NM_001184749.2: c.2435T>C p.(Phe812Ser)	5.1	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	hemi	maternal	yes, half-brother with similar symptomes, maternal uncle of 2. Degree (cousin of mother) with behavior abnormality

FAT3	NM_001008781.2: c.1367C>T p.(Ala456Val)	NM_001008781.2:c.11012G>T p.(Arg3671Leu)	5.1	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	comphet	paternal & maternal	no
SRRM4	NM_194286.3: c.560G>A p.(Arg187His)	NM_194286.3:c.140C>T p.(Pro47Leu)	5.1	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	comphet	paternal & maternal	no
SF3B2	NM_006842.2: c.76G>A p.(Ala26Thr)		5.1	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 hypotonia	homo	paternal & maternal	no
BTBD6	NM_033271.2:c.223C>T p.(Leu75Phe)	NM_033271.2:c.835G>A p.(Ala279Thr)	5.1	1	NDD	Developmental disorder	comphet	paternal & maternal	no
CSMD3	NM_052900.2: c.9581A>C p.(Gln3194Pro)	NM_052900.2:c.7073G>A p.(Arg2358Gln)	5.1	1	Epilepsy	one tonic spasm, developmental delay, 20-30 headdrops per day, hypsarrythmia	comphet	paternal & maternal	no
FAT2	NM_001447.2:c.5000T>G p.(Val1667Gly)		5.1	3	NDD	Global developmental delay, intellectual impairment, absent speech	homo	paternal & maternal	no
GUCY2F	NM_001522.2: c.1445C>G p.(Ser482Cys)		5.1	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Ataxia, Global developmental delay, Gait ataxia, Olivopontocerebellar atrophy, Short stature, Pontocerebellar atrophy, Olivopontocerebellar hypoplasia, Cognitive impairment	homo	paternal & maternal	yes, brother similary affected
SLIT3	NM_003062.3: c.2818C>T p.(Arg940Cys)		5.1	3	NDD + Epilepsy	Intellectual disability, epilepsy with generalized tonic-clonic seizures, short attention span	het	maternal	yes, Father with epilepsy, mother with epilepsy and intellectual disability

MYO10	NM_012334.3: c.5014A>G p.(Thr1672Ala)	NM_012334.3: c.2534A>G p.(Glu845Gly)	5.1	1	NDD	Intellectual disability, Microcephaly, Narrow mouth, Cleft soft palate, Short stature	comphet	paternal & maternal	no
HSPB7	NM_014424.4: c.202C>T p.(Arg68Cys)		5.1	3	NDD	Global developmental delay, Motor delay	het	de novo	no
MORC4	NM_024657.4: c.1382A>G p.(Tyr461Cys)		5.1	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	hemi	maternal	yes, two brothers similary affected, one deceased
TENM2	NM_001122679.1: c.4082A>G p.(Tyr1361Cys)	NM_001122679.1:c.7924G>A p.(Val2642Met)	5.0	2	NDD	Hypertelorism, Low-set ears, Brachydactyly, Intellectual disability, Global developmental delay, Hypoplasia of the corpus callosum, Elevated serum creatinine, Moderate global developmental delay	comphet	paternal & maternal	no
ALS2CL	NM_147129.5: c.1109+5G>A p.?		5.0	3	Epilepsie	atypic absence seizure, strartle-induced seizure, attention deficit hyperactivity disorder, seizure	het	de novo	no
ADGRD2	NM_001161808.1: c.1068C>A p.(Cys356*)		5.0		NDD	(+) Global developmental delay,(+) Motor delay,(+) Neonatal asphyxia,(+) Neonatal seizure,(+) Hypertonia,(+) Dysphagia,(+) Tongue fasciculations,(+) Microcephaly,(+) Infantile encephalopathy	het	de novo	brother with neonatal seizures, no developmental delay; sister with cleft lip and neonatal seizures; maternal aunt with epilepsy since the age of 18
IL1RAPL2	NM_017416.1: c.187A>G p.(Asn63Asp)		5.0	3	NDD	Muscular hypotonia, motor development delay, speech delay, stereotypic movements, suspected autism	hemi	maternal	no
PKN3	NM_013355.4: c.137A>C p.(Asp46Ala)		5.0	1	Epilepsy	Generalisierte Epilepsie mit febrilen Anfällen seit dem 3. LJ	het	de novo	no

HNRNPM	NM_005968.4:c.23C>T p.(Ala8Val)	5.0	2	Epilepsy	generalized epilepsy with nocturnal tonic-clonic seizures (onset in the 2nd year of life), mild intellectual impairment	homo	paternal & maternal	no
ZNF888	XM_005259451.1: c.1525_1550del p.(Leu509Trpfs*8)	5.0	3	NDD	Coloboma, Iris coloboma, mild Intellectual disability, mild Global developmental delay	het	de novo	no
MROH2B	NM_173489.4: c.3685del p.(Asp1229Thrfs*15)	5.0	1	Epilepsy	Seizures, Encephalopathy, Absence seizure, Generalized-onset seizure	het	de novo	no
XYLB	NM_005108.3:c.1101G>A p.(Met367Ile)	5.0	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	het	de novo	no
SLC10A3	NM_001142391.1: c.1160C>T p.(Thr387Met)	5.0	3	NDD	Autism, Intellectual disability, Global developmental delay	hemi	maternal	no
SPHK2	NM_001204158.2: c.1534G>T p.(Val512Leu)	5.0	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)	homo	paternal & maternal	no
GAL3ST3	NM_033036.2: c.39G>C p.(Lys13Asn)	5.0	3	NDD + Epilepsy	seizures, focal seizures	het	de novo	no

PTK2B	NM_004103.4: c.1057C>T p.(Arg353Trp)		5.0	1	Epilepsy	Absence seizures, familiar	het	paternal	yes, Familiar epilepsy; Index with daily absence-epilepsy, father with fever cramps in childhood, sister with fever cramps, paternal grand-uncle with absence-epilepsy
PHC2	NM_198040.2:c.604 G>A p.(Ala202Thr)		5.0	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	het	unknown	no
PKHD1L1	NM_177531.4:c.519 4C>T p.(Pro1732Ser)	NM_177531.4:c.800 5C>T p.(Gln2669*)	5.0	2	Epilepsy	Seizure, Status epilepticus, EEG abnormality, Focal impaired awareness seizure, Focal-onset seizure, EEG with focal spike waves	comphet	paternal & maternal	no
ZKSCAN3	NM_001242894.1: c.253A>T p.(Ile85Phe)		5.0	3	NDD	Hypothyroidism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Short stature, Abnormal social behavior	het	de novo	no
SNF8	NM_007241.3: c.572G>A p.(Gly191Asp)	NM_007241.3:c.236 C>T p.(Pro79Leu)	5.0	2	NDD	Global developmental delay, Microcephaly, Agenesis of corpus callosum, Failure to thrive, Growth delay, EEG abnormality, Abnormal cry	Comphet	paternal & maternal	no
GBP5	NM_001134486.2: c.154T>C p.(Ser52Pro)	NM_001134486.2:c. 502_505dup p.(Ser169*)	5.0	2	NDD	Hydrocephalus, Intellectual disability, hypotonia, Global developmental delay, Atria septal defect, Patent ductus arteriosus,Transposition of the great arteries with ventricular septal defect	comphet	paternal & maternal	no

HMG20A	NM_001304504.1:c.237+5G>T p.?		5.0	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	homo	paternal & maternal	no
AATK	NM_001080395.2: c.2915G>T p.(Gly972Val)		4.9	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	homo	paternal & maternal	no
KLHL6	NM_130446.2: c.1061C>A p.(Pro354Gln)		4.9	2	NDD + Epilepsy	epileptic encephalopathy, seizures	homo	paternal & maternal	no
APLN	NM_014499.3: c.416T>C p.(Phe139Ser)		4.9	1	NDD	developmental delay, speech delay, motor delay, aggressive behaviour, selfharming behaviour, no ID (IQ98)	hemi	maternal	yes, father with ADHS as a child
GABRE	NM_004961.3: c.319G>T p.(Gly107Cys)		4.9	2	NDD	Macrocephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Intellectual disability, severe, Cognitive impairment	hemi	maternal	no
EPHB3	NM_004443.4: c.1711G>A p.(Val571Met)		4.9	2	NDD	Intellectual disability, Seizures (onset at age of 14 years), Sleep disturbance, Hypotonic cerebral palsy	homo	paternal & maternal	no
FOXO4	NM_005938.3: c.43A>T		4.9	3	NDD	global developmental delay, absent speech, muscular hypotonia, austism spectrum disorder,	hemi	maternal	no
SFXN3	NM_030971.3: c.785G>A p.(Arg262His)	NM_030971.3:c.640del p.(Ala214Glnfs*9)	4.9	3	NDD	Hypothyroidism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Short stature, Abnormal social behavior	comphet	paternal & maternal	no
MYRIP	NM_001284423.1: c.383G>A p.(Arg128His)	NM_001284423.1:c.86G>A p.(Arg29His)	4.9	3	NDD	Severe global developmental delay, sleep disturbance, behavioral abnormalities, flat face	comphet	paternal & maternal	no

SCUBE2	NM_001170690.1: c.68C>T p.(Pro23Leu)		4.9	2	NDD + Epilepsy	global developmental delay, seizures, hypoplasia of the corpus callosum	het	de novo	N/A
MAGEA10	NM_001011543.2: c.125C>T p.(Thr42Ile)		4.9	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	hemi	maternal	yes, Father with similar symptom, grandmother (paternal): mental retardation; Grandmother and great-grandmother maternal: Leukemia in old age
LRFN4	NM_024036.5: c.473G>C, p.(Arg158Pro)	NM_024036.5: c.853C>T, p.(Arg285Cys)	4.9	2	NDD	schwere Entwicklungsverzögerung, spastische Cerebralparese, Dystrophie, MRT unauf-fällig, EEG auffällig	comphet	paternal & maternal	no
RNF44	NM_014901.4: c.802-8T>G p.?		4.9	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	het	de novo	no
C11ORF95	NM_001144936.1: c.1592T>C p.(Val531Ala)		4.9	2	NDD + Epilepsy	global developmental delay, seizures, hypoplasia of the corpus callosum	homo	paternal & maternal	N/A
TAF5	NM_006951.4: c.479C>T p.(Ala160Val)		4.9	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Cerebellar vermis atrophy, Cognitive impairment	homo	paternal & maternal	no
CCDC136	NM_022742.4: c.1018C>T p.(Arg340Trp)	NM_022742.4: c.1079G>A p.(Ser360Asn)	4.9		NDD + epilepsy	(+) Intellectual disability,(+) Arthrogryposis multiplex congenita,(+) Polymicrogyria,(+) Seizure	comphet	paternal & maternal	no
RHOT2	NM_138769.2: c.586T>G p.(Ser196Ala)	NM_138769.2:c.1201C>T p.(Arg401Cys)	4.9	1	NDD + Epilepsy	spastic tetraparesis, generalized tonic-clonic seizures, microcephaly, polymicrogyria, periventricular gliosis and cysts, global	comphet	paternal & maternal	no

UBE3C	NM_014671.2: c.485G>C p.(Ser162Thr)	NM_014671.2:c.871G>A p.(Val291Ile)	4.9	3	NDD + Epilepsy	Intellectual disability, Generalized seizures, Febrile seizures, Focal seizures	comphet	paternal & maternal	no
AKAP17A	NM_005088.2: c.1328T>C p.(Leu443Pro)		4.9	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	het	de novo	no
TEX44	NM_152614.2: c.1146C>G p.(His382Gln)		4.9	1	NDD	mild global developmental delay, delayed speech and language development	het	de novo	no
FAM171A1	NM_001010924.1: c.2435C>T p.(Ala812Val)		4.8	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	homo	paternal & maternal	no
TLK2	NM_001112707.1: c.667A>T p.(Met223Leu)		4.8	3	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	het, in multiple	unknown	yes, brother with suspected epilepsy, father with febrile seizures in childhood, uncle (paternal) epilepsy and autism, cousin (paternal) with febrile seizures, uncle (maternal) possibly febrile seizures
FHDC1	NM_033393.2: c.568C>T p.(Arg190Trp)		4.8	1	NDD	Hypertension, Intellectual disability,mild, Obesity, Abnormality of the pulmonary valve, I Hyperlipidemia, Childhood-onset truncal obesity	het	de novo	yes, mother was in a learn special school

RPS6KA6	NM_001330512.1: c.2113-3T>G p.?		4.8	2	Epilepsy	nocturnal focal seizures, onset on the age of 4, microcephaly, developmental delay, intelligence impairment (IQ=81), ADHS	hemi	maternal	no
CAND2	NM_001162499.1: c.2591C>T p.(Ala864Val)		4.8	4	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay	het	de novo	no
ZNF664	NM_001204298.1: c.691G>A p.(Glu231Lys)		4.8	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	het	de novo	no
MORF4L2	NM_001142418.1: c.287A>G p.(Gln96Arg)		4.8	3	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormality of the caudate nucleus, Infantile onset, Attention deficit hyperactivity disorder	hemi	maternal	no
ZNF182	NM_001178099.1:c.181A>G p.(Ser61Gly)		4.8	1	Epilepsy	Seizure, Nocturnal seizures	hemi	maternal	no
ABCB10	NM_012089.2:c.833_838del p.(Asp278_Thr279del)		4.8	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	het	de novo	no
CRYBG3	NM_153605.3:c.8492G>A p.(Arg2831His)		4.8	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure	homo	paternal & maternal	no
TENM1	NM_001163278.1: c.757A>G p.(Asn253Asp)		4.8	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	hemi	maternal	yes, Mother and maternal half-sister have intellectual disability
DGKQ	NM_001347.3: c.1736A>T p.(His579Leu)	NM_001347.3:c.1408C>T p.(Arg470Trp)	4.8	2	Epilepsy	Seizures, Global developmental delay, Hypoglycorrhachia, Hyperglycorrhachia	comphet	paternal & maternal	no

STARD7	NM_020151.3: c.64C>T p.(Leu22Phe)		4.8	1	Epilepsy	Seizures, Generalized tonic-clonic seizures, Absence seizures	het	de novo	no
GRPR	NM_005314.2: c.923G>A p.(Arg308His)		4.8	2	NDD + Epilepsy	Seizures, Global developmental delay, Epileptic encephalopathy	hemi	maternal	no
OGDHL	NM_001143996.1: c.489G>C p.(Trp163Cys)	NM_001143996.1:c.1315C>T p.(Arg439Cys)	4.7	2	NDD + Epilepsy	Seizures, Myoclonic absences, developmental delay	comphet	paternal & maternal	yes, mother with epilepsy in childhood
BTBD3	NM_181443.3:c.1502C>A p.(Pro501Gln)		4.7	2	NDD	Intellectual disability, Global developmental delay, Obesity, Abnormal heart morphology, Truncal obesity, Kyphosis, Attention deficit hyperactivity disorder	het	unknown	yes, parents with adipositas, brother similary affected
PSMB3	NM_002795.2: c.424T>C p.(Cys142Arg)		4.7	1	NDD	Trismus, Arthrogryposis multiplex congenita, Vesicoureteral reflux, Abnormality of the kidney, abnormal facial shape, Global developmental delay	het	de novo	no
DAAM2	NM_001201427.1:c.1339C>G p.(Gln447Glu)	NM_001201427.1:c.1745C>A p.(Pro582His)	4.7	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech	comphet	paternal & maternal	no
PRDM13	NM_021620.3: c.994G>A p.(Gly332Arg)		4.7	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	homo	paternal & maternal	yes, sister similary affected
GRM2	NM_000839.3: c.2462C>T p.(Pro821Leu)		4.7	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	het	unknown	no
SMCR8	NM_144775.2: c.2404C>T p.(Arg802Cys)		4.7	2	NDD	Microcephaly, Epicanthus, Intellectual disability, Global developmental delay, Plagiocephaly, Abnormal facial shape, Wide nasal base	het	de novo	no

SDK1	NM_152744.3: c.1295G>C p.(Gly432Ala)	NM_152744.3:c.3802C>T p.(Arg1268Trp)	4.7	3	NDD + Epilepsy	seizures, focal seizures	comphet	paternal & maternal	no
UNC5A	NM_133369.2:c.578C>A p.(Ser193Tyr)	NM_133369.2:NM_133369.2:c.267C>G p.(Ile89Met)	4.7	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	comphet	paternal & maternal	no
SLC25A35	NM_001320870.1: c.194G>A p.(Gly65Asp)		4.7	3	ndd	Microcephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe	homo	paternal & maternal	no
MTCL1	NM_015210.3: c.604A>G p.(Thr202Ala)	NM_015210.3:c.1607T>C p.(Ile536Thr)	4.7	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	comphet	paternal & maternal	no
MPP3	NM_001330233.1: c.742C>T p.(Arg248Cys)		4.7	1	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity	homo	paternal & maternal	no
NLRX1	NM_024618.2: c.428C>T p.(Pro143Leu)		4.7	1	NDD	Ptosis, Muscular hypotonia, Global developmental delay, Abnormal facial shape, Short stature, Feeding difficulties, Thick hair	het	de novo	no
FAM13B	NM_016603.3: c.2203G>A p.(Val735Ile)	NM_016603.3:c.110A>G p.(Glu37Gly)	4.7	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	comphet	paternal & maternal	no
SBNO2	NM_001100122.1:c.1960G>T p.(Val654Leu)		4.7	2	NDD + Epilepsy	Epileptic encephalopathy, intrauterine hydrocephalus (gestational week 24), epilepsy	het	de novo	no

COL19A1	NM_001858.5: c.1843G>A p.(Gly615Ser)	4.7	3	ndd	Microcephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe	homo	paternal & maternal	no
KCNK7	NM_033347.1: c.681C>G p.(His227Gln)	4.7	1	Epilepsy	Seizures, Generalized tonic-clonic seizures, Generalized myoclonic seizures	het	de novo	no
MAGED1	NM_001005332.1: c.640A>G p.(Thr214Ala)	4.7	1	NDD	Early onset autism	hemi	maternal	no
ZNF182	NM_001178099.1: c.1319C>T p.(Thr440Met)	4.7	4	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity	hemi	maternal	no
RNF167	NM_015528.2:c.793 C>G p.(Arg265Gly)	4.6	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	homo	paternal & maternal	no
CAMTA2	NM_001171166.1: c.2639A>G p.(Asp880Gly)	4.6	2	NDD + Epilepsy	Seizures, Status epilepticus, Hypsarrhythmia,	homo	paternal & maternal	no
TOPAZ1	NM_001145030.1: c.481A>T p.(Ser161Cys)	4.6		Epilepsy	Focal-onset seizure, Focal sensory seizure	het	de novo	yes, Father and Uncle with same sypmtoms in childhood
PCDHA9	NM_031857.1: c.1134_1135delCGinsTT p.(Ala379Ser)	4.6	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 hypotonia	homo	paternal & maternal	no
PLXNA1	NM_032242.3: c.475T>C p.(Tyr159His)	4.6	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,	het	unknown	no

TNMD	NM_022144.2: c.145G>T p.(Gly49Trp)		4.6	3	NDD	Microcephaly, Delayed speech and language development, Global developmental delay, Abnormal facial shape, Severe short stature, Short stature, Intellectual disability	het	de novo	no
ACTR1A	NM_005736.3: c.715G>C p.(Ala239Pro)		4.6		NDD + epilepsy	Generalized-onset motor seizure, Spastic tetraplegia, Intellectual disability, severe, Cataract, Pes planus	het	unknown	no
RGPD8	NM_001164463.1:c.3225G>T p.(Gln1075His)		4.6	3	NDD + Epilepsy	Global developmental delay with delayed speech, astatic attacks, absence epilepsy and EEG abnormalities	het	de novo	no
PRMT9	NM_138364.3:c.1144C>A p.(Gln382Lys)		4.6	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Pes planus, Focal tonic seizure, Cognitive impairment	homo	paternal & maternal	no
WWP2	NM_001270453.1: c.491A>C p.(Glu164Ala)	NM_001270453.1:c.166G>C p.(Ala56Pro)	4.6	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	comphet	paternal & maternal	no
DCDC1	NM_181807.3: c.515G>A p.(Arg172Lys)		4.6	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	het	de novo	no

C1orf228	NM_001145636.1: c.979C>T p.(Arg327Cys)		4.6	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	het	de novo	no
PNCK	NM_001135740.1: c.643C>G p.(Leu215Val)		4.6	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	hemi	maternal	Cousin on the father's side with Down syndrome, Grandfather on the father's side with unexpected death at the age of 45
KANK4	NM_181712.4: c.1849C>T p.(Gln617*)		4.6	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	het	de novo	no
HIST1H3H	NM_003536.2: c.397G>T p.(Gly133Cys)		4.6	1	NDD + Epilepsy	Global developmental delay, Hypsarrhythmia, Inability to walk, Epileptic spasms, Infantile spasms	het	de novo	no
SLC2A8	NM_014580.4: c.1150G>A p.(Gly384Ser)	NM_014580.4:c.1239C>G p.(Cys413Trp)	4.5	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, Cognitive impairment	comphet	paternal & maternal	no
FAT3	NM_001008781.2: c.3669+7G>A p.?	NM_001008781.2:c.12922G>C p.(Asp4308His)	4.5	3	NDD + Epilepsy	Autism, Seizures, Global developmental delay, Motor delay, Absent speech, Epileptic encephalopathy	comphet	paternal & maternal	N/A

NEURL4	NM_032442.2:c.4345C>G p.(Pro1449Ala)	NM_032442.2:c.2944G>A p.(Glu982Lys)	4.5	2	Epilepsy	Seizures, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Atonic seizures, Focal tonic seizures, Myoclonic atonic seizures	comphet	paternal & maternal	no
SLC23A1	NM_152685.3:c.1105A>G p.(Ile369Val)	NM_152685.3:c.1063C>A p.(Pro355Thr)	4.5	3	NDD	Renal agenesis, Abnormal cornea morphology, Aniridia, Microphthalmia, Global developmental delay	comphet	paternal & maternal	no
ZNF280D	NM_001288588.1:c.2532C>G p.(His844Gln)		4.5	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	het	de novo	no
DYNC1I1	NM_004411.4:c.1421C>G p.(Ala474Gly)		4.5	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	het	unknown	no
CXorf21	NM_025159.2:c.774A>C p.(Arg258Ser)		4.5	2	NDD + Epilepsy	Leukenzephalopathie, keine Sprache, muskuläre Hypertonie, schwere motorische und sprachlich-mentale Entwicklungsverzögerung, therapierefraktäre Krampfanfälle	hemi	maternal	no
H2AC8	NM_021052.2:c.107G>A p.(Arg36His)		4.5		NDD	(+) Arachnoid cyst,(+) Headache,(+) Hallucinations,(+) Visual hallucinations,(+) Auditory hallucinations,(+) Delayed speech and language development,(+) Global developmental delay,(+) Intellectual disability,(+) Obesity	het	de novo	Mother with arterial hypertension, obesity, chronic bronchitis, in 8 th graduated from school, grandmother and grandfather from the sid of the mother with arterial
TNN	NM_022093.1:c.1949A>T p.(Tyr650Phe)	NM_022093.1:c.2852T>G p.(Val951Gly)	4.5	1	NDD + Epilepsy	infantile spams since 6 months of age, conspicuous odor, crying phases, failure to thrive	comphet	paternal & maternal	no

MYO9B	NM_001130065.1: c.248C>T p.(Ser83Leu)	NM_001130065.1:c.5020G>A p.(Val1674Met)	4.5	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	comphet	paternal & maternal	no
TENM2	NM_001122679.1: c.3881C>G p.(Ser1294Cys)		4.5	1	NDD + Epilepsy	atonic-astatic seizures and mild intellectual disability	het	unknown	yes, brother with infantile autism
TMEM121B	NM_031890.3: c.254T>C p.(Val85Ala)		4.5	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	het	de novo	no
CA5B	NM_007220.3: c.352_354dup p.(Gly118dup)		4.4	4	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity	hemi	maternal	no
PCSK6	NM_002570.4: c.412C>A p.(Leu138Ile)	NM_002570.4: c.2232A>T p.(Arg744Ser)	4.4	2	NDD	Mild global developmental delay, Cleft palate, Hearing impairment, Ventricular septal defect, Patent ductus arteriosus, Pulmonic stenosis, Hypospadias, Enlarged vestibular aqueduct, Pes	comphet	paternal & maternal	no
BIRC6	NM_016252.3: c.8570C>G p.(Ser2857Cys)	NM_016252.3:c.12796G>A p.(Val4266Met)	4.4	1	NDD + Epilepsy	Delayed speech and language development, Seizures, Familial predisposition, Poor speech, Infantile onset, Infantile spasms	comphet	paternal & maternal	no
TMEM143	NM_018273.3: c.1022T>C p.(Met341Thr)		4.4	3	Epilepsie	(+) Focal tonic seizure,(+) EEG with focal sharp waves,(+) Nocturnal seizures,(-) Brain imaging abnormality	het	de novo	maternal grandmother with epilepsy, café au lait spots
TTBK1	NM_032538.2: c.3116_3118del p.(Thr1039del)		4.4	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	het	paternal	yes,father affected

PPM1L	NM_139245.3:c.237 G>C p.(Glu79Asp)		4.4	2	NDD + Epilepsy	Microcephaly, Visual impairment, Intellectual disability, Seizures, Global developmental delay, Motor delay, Encephalopathy, Generalized tonic-clonic seizures	homo	paternal & maternal	no
TCP11	NM_001093728.2: c.1440T>A p.(Phe480Leu)		4.4	3	NDD	Microcephaly, Delayed speech and language development, Global developmental delay, Abnormal facial shape, Severe short stature, Short stature, Intellectual disability	het	de novo	no
SEMA4C	NM_017789.4: c.2077_2078delGAinsTT p.(Glu693Leu)	NM_017789.4: c.517+3G>A p.?	4.4		Epilepsie	At the age of 7-8 months tonic stiffnesses for a few seconds every few weeks, later on big-ger seizures, MRI without findings, no motor delay, increased levels of serum lactate, glutaric aciduria	comphet	paternal & maternal	no
DSCAML1	NM_020693.3:c.132 2C>T p.(Ser441Phe)		4.4	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	het	maternal	yes, brother similary affected
ZDHHC14	NM_024630.2: c.1441G>A p.(Gly481Ser)		4.4	3	NDD	Behavioral abnormality, Intellectual disability, Gait ataxia	het	de novo	no
NOMO1	NM_014287.4: c.2173G>A p.(Gly725Ser)		4.4	2	Epilepsie	(+) Focal-onset seizure,(+) Brain imaging abnormality	homo	paternal & maternal	
HOOK2	NM_001100176.1: c.1718-6C>T p.?		4.4	2	NDD	intellectual disability	homo	paternal & maternal	consanguinity
MDN1	NM_014611.2: c.2965-3T>C p.?	NM_014611.2:c.9524A>C p.(His3175Pro)	4.4	4	NDD	global developmental delay, intellectual disability, generalized hypotonia,	comphet	paternal & maternal	no
TAAR2	NM_001033080.1: c.113G>T p.(Arg38Ile)		4.4	3	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormality of the caudate nucleus, Infantile onset, Attention deficit hyperactivity disorder	het	de novo	no
LRCH3	NM_032773.3:c.256 C>T p.(Arg86Trp)		4.4	5	NDD + Epilepsy	Intellectual disability, Seizure	homo	paternal & maternal	no

DGKZ	NM_001199266.1: c.132_134del p.(Ser45del)	NM_001199266.1:c.16G>C p.(Gly6Arg)	4.4	3	NDD + Epilepsy	Autism, Intellectual disability, Seizures, Global developmental delay, Poor speech, Focal seizures	comphet	paternal & maternal	no
WDR59	NM_030581.3: c.2326G>T p.(Val776Leu)	NM_030581.3:Del Exons 19 - 25	4.4	3	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	comphet	paternal & maternal	no
MEGF11	NM_032445.2: c.3080T>G p.(Leu1027Arg)	NM_032445.2:c.254G>C p.(Arg85Thr)	4.4	1	Epilepsy	Seizures, Prolonged QT interval, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Recurrent infections, Focal seizures	comphet	paternal & maternal	no
RXRB	NM_001270401.1: c.1472C>A p.(Ala491Asp)		4.3	2	NDD	Intellectual disability, severe, Severe global developmental delay, Seizure, Focal-onset seizure, Motor seizure, Cerebral palsy (GMFCS V), Microcephaly, Feeding difficulties no speech	het	unknown	no
NKX3-1	NM_006167.3:c.491C>T p.(Thr164Met)	NM_006167.3:c.113G>A p.(Gly38Asp)	4.3	1	NDD + Epilepsy	Intelligenzminderung, Epilepsie, Schwerhörigkeit	comphet	paternal & maternal	no
DNAH14	NM_001373.1: c.13384G>A p.(Ala4462Thr)	NM_001373.1:c.6100C>T p.(Arg2034*)	4.3	2	NDD	Visual loss, Optic nerve hypoplasia, Global developmental delay, Hypoglycemia, Abnormality of optic chiasm morphology	comphet	paternal & maternal	no
PRRG1	NM_000950.2: c.331C>T p.(Arg111Cys)		4.3	3	NDD	Autism, Delayed speech and language development, Global developmental delay, Motor delay, Generalized hypotonia, Poor speech, Abnormality of movement	hemi	maternal	no
MARCH6	NM_005885.3: c.1108T>C p.(Tyr370His)	NM_005885.3:c.1897-3C>T p.?	4.3	1	NDD + Epilepsy	global development delay, seizures, microcephaly, autism, single transverse palmar crease, broad palm, abnormal fracial shape	comphet	paternal & maternal	no
DENND4B	NM_014856.2: c.319G>A p.(Val107Met)	NM_014856.2:c.941G>A p.(Ser314Asn)	4.3	2	NDD	Delayed speech and language development, Intellectual disability	comphet	paternal & maternal	no
MTMR3	NM_021090.3: c.848A>G p.(Asn283Ser)	NM_021090.3:c.1088G>A p.(Arg363Gln)	4.3	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	comphet	paternal & maternal	no

MYRIP	NM_001284423.1: c.1525G>A p.(Asp509Asn)	NM_001284423.1:c.2419C>T p.(Pro807Ser)	4.3	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	comphet	paternal & maternal	no
PLXNA1	NM_032242.3: c.2690G>A p.(Arg897His)	NM_032242.3:c.1045G>C p.(Val349Leu)	4.3	2	Epilepsy	Seizures, Encephalopathy, Focal seizures, Encephalitis	comphet	paternal & maternal	no
ZNF341	NM_032819.4: c.2260C>T p.(Arg754Cys)		4.3	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Specific learning disability, Absence seizure, Generalized-onset seizure, Intellectual disability, borderline, Attention deficit hyperactivity disorder	het	de novo	no
HELZ2	NM_001037335.2:c.7693C>T p.(Arg2565Cys)	NM_001037335.2:c.1750C>T p.(Arg584Trp)	4.3	3	NDD	Microcephaly, Intellectual disability, Ataxia, Intellectual disability, mild, Global developmental delay, Spastic gait, Gait ataxia, Intention tremor, Spastic ataxia	comphet	paternal & maternal	no
DNHD1	NM_144666.2:c.7549C>T p.(Arg2517Cys)	NM_144666.2:c.2104-4T>A p.?	4.2	2	NDD + Epilepsy	Microcephaly, Seizure, Dystonia, Cerebral palsy, Abnormality of movement, Epileptic encephalopathy	comphet	paternal & maternal	no
HIST1H4B	NM_003544.2: c.158A>G p.(Glu53Gly)		4.2	3	NDD	Seizures, Pachygyria, Delayed CNS myelination, Heterotopia, Periventricular gray matter heterotopia, Intracranial cystic lesion, Abnormality of brain morphology	het	de novo	no
PCID2	NM_001258212.1:c.835G>A p.(Asp279Asn)	NM_001258212.1:c.35A>G p.(Gln12Arg)	4.2	2	NDD	global muscular hypotension with axial weakness, facial dysmorphia, indicated high palate, broad neck, muscle relief decreasing distally, hypersalivation, no secure free sitting	comphet	paternal & maternal	no
TDRD9	NM_153046.2:c.2273C>T p.(Pro758Leu)		4.2	2	NDD	Autism, Hypertrichosis, Intellectual disability, Global developmental delay, Absent speech, Mutism	homo	paternal & maternal	no
DEPTOR	NM_022783.2: c.496A>G p.(Met166Val)	NM_022783.2:c.426-5C>T p.?	4.2	3	Epilepsy	Seizures, Hypoglycemia, Myopathy, Focal seizures, Ichthyosis, EEG with focal epileptiform discharges	comphet	paternal & maternal	no

URB1	NM_014825.2:c.5312A>T p.(Tyr1771Phe)	NM_014825.2:c.3362G>A p.(Arg1121His)	4.2	3	NDD	Microcephaly, Intellectual disability, Ataxia, Intellectual disability, mild, Global developmental delay, Spastic gait, Gait ataxia, Intention tremor, Spastic ataxia	comphet	paternal & maternal	no
EIF5B	NM_015904.3:c.3090-13_3090-7del p.?	NM_015904.3:c.2081G>A p.(Arg694Gln)	4.2	1	NDD	Epicanthus, Hypertelorism, Behavioral abnormality, Precocious puberty, Intellectual disability	comphet	paternal & maternal	no
STARD9	NM_020759.2: c.12652C>T p.(His4218Tyr)	NM_020759.2:c.13445C>T p.(Ser4482Phe)	4.2	1	NDD + Epilepsy	mild ID, generalized epilepsy	comphet	paternal & maternal	yes, one paternal uncle and one paternal aunt has ID
TANC1	NM_001145909.1: c.2395G>A p.(Asp799Asn)		4.2	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay	het	unknown	no
WWC3	NM_015691.3: c.2935C>T p.(Arg979Trp)		4.2	3	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	hemi	maternal	no
SMYD5	NM_006062.3: c.100A>G p.(Lys34Glu)	NM_006062.3: c.833G>A p.(Arg278His)	4.2	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	comphet	paternal & maternal	no
INPP5F	NM_014937.3: c.3172_3174del p.(Ser1058del)	NM_014937.3:c.3144_3149del p.(Leu1049_Glu1050del)	4.1	1	NDD + Epilepsy	Global developmental delay, Epileptic spasms	comphet	paternal & maternal	no
SSPO	NM_198455.2: c.14254G>A p.(Val4752Ile)		4.1	3	NDD	Autism, Delayed speech and language development, Global developmental delay, Motor delay, Generalized hypotonia, Poor speech, Abnormality of movement	het	de novo	no

ANKS1A	NM_015245.2:c.2269C>T p.(Arg757Cys)		4.1	4	Epilepsy	Episodic ataxia, EEG abnormality	het	maternal	yes, maternal aunt with hemiplegic migraine, mother with migraine, paternal grandfather with migraine and apoplex (40yrs)
ZNF503	NM_032772.4: c.69_71dup p.(Gly27dup)	NM_032772.4:c.1105G>T p.(Gly369Cys)	4.1	3	NDD + Epilepsy	seizures, focal seizures	comphet	paternal & maternal	no
ZNF611	NM_001161499.1:c.1904C>T p.(Ser635Leu)		4.1	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology	homo	paternal & maternal	yes, maternal aunt and paternal uncle with seizures
LRCH2	NM_020871.3: c.2141A>G p.(Asn714Ser)		4.1	3	NDD + Epilepsy	Therapy-resistant epilepsy since the age of two, Epileptic encephalopathy	hemi	maternal	no
CBLL2	NM_152577.3: c.701A>G p.(Lys234Arg)		4.1	3	NDD + Epilepsy	Hydrocephalus, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Aqueductal stenosis	hemi	maternal	no
GRIK3	NM_000831.3:c.1531-5T>G p.?		4.1	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	het	unknown	yes, nephew with epilepsy and heart malformation
GCNA	NM_052957.4: c.673C>A p.(Pro225Thr)		4.1	2	NDD + Epilepsy	Epilepsy (post- brain haemorrhage condition), intelligence impairment, autism, seizures, premature birth	het	de novo	no
UTP14A	NM_006649.3:c.124A>G p.(Lys42Glu)		4.1	2	NDD + Epilepsy	Epileptic encephalopathy, Seizure since the age of 11	hemi	maternal	no

SPATA8	NM_173499.4: c.28C>T p.(Gln10*)	4.1	2	Epilepsy	nocturnal focal seizures, onset on the age of 4, microcephaly, developmental delay, intelligence impairment (IQ=81), ADHS	het	de novo	no
ARHGAP6	NM_013427.2: c.1586T>C p.(Val529Ala)	4.0	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	hemi	maternal	no
HOXD4	NM_014621.2: c.111C>A p.(Tyr37*)	4.0	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	het	unknown	yes, maternal grandmother with epilepsy
CLCC1	NM_001048210.2: c.1324C>T p.(Leu442Phe)	4.0	3	NDD + Epilepsy	Intellectual disability, epilepsy with generalized tonic-clonic seizures, short attention span	het	maternal	yes, Father with epilepsy, mother with epilepsy and intellectual disability
EIF4ENIF1	NM_001164501.1:c. 1588C>T p.(Leu530Phe)	4.0	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart	het	unknown	no
SOX7	NM_031439.3:c.723 G>A p.(Pro241=)	4.0	2	NDD	Microcephaly, Hearing impairment, Autism, Intellectual disability, Spasticity, Global developmental delay, Cerebral calcification	het	de novo	no

PPP3CC	NM_001243975.1:c.323G>A p.(Arg108His)		4.0	4	Epilepsy	Episodic ataxia, EEG abnormality	het	maternal	yes, maternal aunt with hemiplegic migraine, mother with migraine, paternal grandfather with migraine and apoplex (40yrs)
MED14	NM_004229.3: c.3657T>G p.(His1219Gln)		4.0	1	NDD + Epilepsy	Autistic behavior, Intellectual disability, Seizures, Global developmental delay, Intellectual disability, severe, Severe global developmental delay, Epileptic encephalopathy	hemi	maternal	no
ANXA6	NM_001155.4: c.1670C>T p.(Pro557Leu)	NM_001155.4:c.319-6_319-5delCCinsTG p.?	4.0	2	NDD	Spasticity, Global developmental delay, Motor delay, Cerebral palsy, Abnormality of movement, Dyskinesia	comphet	paternal & maternal	no
TAF7L	NM_001168474.1: c.1100A>G p.(Gln367Arg)		4.0	2	Epilepsy	Seizures, Focal seizures, Multifocal epileptiform discharges, EEG with focal epileptiform discharges, Polymorphic focal epileptiform discharges, EEG with focal spikes	hemi	maternal	no
TEX13C	NM_001195272.1: c.1745A>G p.(Glu582Gly)		4.0	2	NDD	Cryptorchidism, Hypospadias, Narrow mouth, Microcephaly, Synophrys, Intellectual disability, Global developmental delay, Highly arched eyebrow, Penile hypospadias, Short stature, Decreased body weight, Microtia, Feeding difficulties, Scrotal hypospadias,	hemi	maternal	no
KIR3DL3	NM_153443.4:c.1053G>A p.(Lys351=)		4.0	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology	homo	paternal & maternal	yes, maternal aunt and paternal uncle with seizures
STARD8	NM_001142503.2: c.2248C>A p.(Leu750Ile)		4.0	1	Epilepsy	EEG with burst suppression, Epileptic encephalopathy, Global developmental delay, Intellectual disability, Seizure	hemi	maternal	no

GIPR	NM_000164.3: c.784C>G p.(Leu262Val)	NM_000164.3:c.393 G>T p.(Arg131Ser)	4.0	1	NDD	Absent speech, Obesity, Intellectual disability, severe	comphet	paternal & maternal	yes, brother with similar symptomes, but without epilepsy
ZBTB45	NM_001316978.2: c.976G>A p.(Gly326Arg)		4.0	2	Epilepsie	(+) Focal-onset seizure,(+) Brain imaging abnormality	homo	paternal & maternal	N / A
RSRC2	NM_023012.5: c.603-8T>C p.?		4.0	3	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	het	de novo	no
PGBD2	NM_170725.2: c.607A>C p.(Thr203Pro)		4.0	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	het	de novo	no
HAGH	NM_005326.4: c.355_357dup p.(Ser119dup)	NM_005326.4:c.578 A>G p.(Tyr193Cys)	4.0	5	Epilepsy	Seizures, Generalized myoclonic seizures	comphet	paternal & maternal	no
COL20A1	NM_020882.2: NM_020882.2:c.361 4-8C>T p.?		3.9	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)	het	de novo	no
KIAA1107	NM_015237.3:c.299 C>T p.(Thr100Ile)		3.9	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure	homo	paternal & maternal	no
SRPX	NM_001170750.1: c.1270A>T p.(Thr424Ser)		3.9	4	NDD	Intellectual disability, Global developmental delay	hemi	maternal	no

HEPH	NM_138737.4: c.812_814del p.(Pro271del)		3.9	3	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia	hemi	maternal	no
CD99L2	NM_001242614.1: c.541G>C p.(Gly181Arg)		3.9	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	hemi	maternal	no
TAB3	NM_152787.3: c.1952A>G p.(Gln651Arg)		3.9	2	NDD + Epilepsy	Intellectual disability, Seizures, Epileptic encephalopathy	hemi	maternal	no
ZNF761	NM_001008401.3: c.2085_2086del p.(Cys695Trpfs*5)		3.9	5	Epilepsy	Seizures, Generalized myoclonic seizures	het	de novo	no
DNAH17	NM_173628.3: c.11678-7_11678-3del p.?	NM_173628.3:c.9998C>T p.(Ser3333Leu)	3.9	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	comphet	paternal & maternal	no
C5orf64	NM_173667.3: c.37T>C p.(Leu13=)		3.9	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 encephalopathy	het	de novo	no

STARD9	NM_020759.2: c.1649A>G p.(Asn550Ser)	NM_020759.2: c.10380C>G p.(His3460Gln)	3.9	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	comphet	paternal & maternal	no
NIF3L1	NM_001136039.2: c.131C>T p.(Ser44Leu)	NM_001136039.2:c.347C>G p.(Ala116Gly)	3.8	3	NDD	Behavioral abnormality, Intellectual disability, Gait ataxia	comphet	paternal & maternal	no
WBP1	NM_012477.3: c.25G>A p.(Gly9Ser)		3.8	1	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Atonic seizures	het	de novo	no
STPG2	NM_174952.2: c.1128T>A p.(Ser376Arg)	NM_174952.2:c.431G>A p.(Gly144Asp)	3.8	2	NDD + Epilepsy	Delayed speech and language development, Seizures, Focal seizures, Multifocal epileptiform discharges, EEG with focal epileptiform discharges, Epileptic encephalopathy	comphet	paternal & maternal	no
DUSP16	NM_030640.2: c.1091C>A p.(Pro364His)	NM_030640.2:c.183C>G p.(Asp61Glu)	3.8	1	NDD + Epilepsy	Microcephaly, Delayed speech and language development, Seizures, Global developmental delay, Recurrent infections, Infantile onset, Postnatal microcephaly	comphet	paternal & maternal	no
MAB21L4	NM_001085437.2: c.755A>G p.(Tyr252Cys)		3.8	1	NDD	Abnormality of dental enamel, Autistic behavior, Delayed speech and language development, Global developmental delay, Motor delay, Sleep disturbance, Poor coordination	het	de novo	no
LCN15	NM_203347.1: c.399C>A p.(Ser133Arg)		3.8	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	homo	paternal & maternal	no
ZNF331	NM_001079906.1: c.281G>A p.(Arg94His)		3.8	3	NDD + Epilepsy	Intellectual disability, Generalized seizures, Febrile seizures, Focal seizures	homo	paternal & maternal	no

POU2F1	NM_002697.4: c.318G>C p.(Gln106His)		3.8	3	NDD	Autism (Asperger), Autistic behavior, Depressivity, Macrocephaly	het	paternal	sister is similarly affected (global DD, autism, macrocephaly, muscular hypotonia)
POU4F2	NM_004575.2: c.417C>A p.(Asp139Glu)	NM_004575.2:c.180_200del p.(Gly62_Gly68del)	3.8	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures, infantile onset	comphet	paternal & maternal	no
DUSP9	NM_001318503.1:c.745G>A p.(Asp249Asn)		3.8	2	Epilepsy	generalized epilepsy with nocturnal tonic-clonic seizures (onset in the 2nd year of life), mild intellectual impairment	hemi	maternal	no
HIVEP1	NM_002114.3: c.4588T>C p.(Ser1530Pro)	NM_002114.3: c.1916T>C p.(Val639Ala)	3.8		NDD	(+) Global developmental delay,(+) Hyperactivity,(+) Delayed speech and language development,(+) Hypertelorism,(+) Depressed nasal ridge,(+) Low-set ears,(+) Muscular hypotonia, lateral fallende Lidachsen	comphet	paternal & maternal	no
MICAL1	NM_001159291.1: c.571+1G>T p.?	NM_001159291.1:c.2724-8C>T p.?	3.8	3	NDD + Epilepsy	Specific learning disability, Absence seizures, Cortical dysplasia, EEG with continuous slow activity, Seizures	comphet	paternal & maternal	no
CREB5	NM_182898.3:c.302T>A p.(Met101Lys)		3.7	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	het	unknown	no
NIPAL3	NM_020448.4: c.205G>A p.(Ala69Thr)	NM_020448.4:c.163-8G>A p.?	3.7	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	comphet	paternal & maternal	no

KANSL2	NM_017822.3: c.880C>T p.(His294Tyr)		3.7	1	NDD	Intellectual disability, Intellectual disability, moderate, Intellectual disability, severe	het	unknown	yes, two affected maternal uncles, mother and maternal grandmother with mild intellectual disability
GRAMD1C	NM_017577.4: c.168C>A p.(Ser56Arg)	NM_017577.4: c.557A>G p.(Glu186Gly)	3.7	2	Epilepsy	(+) Complex febrile seizure,(+) Simple febrile seizure,(+) Seizure,(-) Motor delay,(-) Intellectual disability	comphet	paternal & maternal	febrile seizures on the side of the mother in childhood: half sister, mother herself, brother of grandfather; mother of grandfather on the side of the father with epilepsy after 50. year of life
STARD9	NM_020759.2: c.4693A>G p.(Ser1565Gly)	NM_020759.2: c.5795A>G p.(Asn1932Ser)	3.7	2	Epilepsy	(+) Complex febrile seizure,(+) Simple febrile seizure,(+) Seizure,(-) Motor delay,(-) Intellectual disability	comphet	paternal & maternal	febrile seizures on the side of the mother in childhood: half sister, mother herself, brother of grandfather; mother of grandfather on the side of the father with epilepsy after 50. year of life

PTPN12	NM_002835.3: c.89G>A p.(Arg30Gln)		3.7	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	homo	paternal & maternal	no
SLC18B1	NM_052831.3: c.821G>T p.(Gly274Val)	NM_052831.3: c.654T>A p.(Asn218Lys)	3.7	3	NDD + epilepsy	Global developmental delay, Microcephaly, Seizures	comphet	paternal & maternal	no
SIGLEC9	NM_001198558.1:c.682G>A p.(Val228Ile)		3.7	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech	het	de novo	no
RASSF10	NM_001080521.2: c.816C>G p.(Tyr272*)		3.7	2	NDD	Cleft palate, Hydrocephalus, Microcephaly, Retinopathy, Intellectual disability, Global developmental delay, Pes planus, Short stature, Pes valgus, Cognitive impairment, Cleft hard palate	het	de novo	no
DNHD1	NM_144666.2: c.2758A>G p.(Ser920Gly)	NM_144666.2:c.2546G>A p.(Arg849Gln)	3.7	3	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia	comphet	paternal & maternal	no
TCEAL3	NM_001006933.1:c.585C>G p.(His195Gln)		3.7	1	NDD	Global developmental delay, Gait ataxia, Infantile muscular hypotonia	hemi	maternal	no
STMN3	NM_015894.3: c.19+8C>A p.?		3.7	1	NDD + Epilepsy	moderate ID, focal epilepsy, brain atrophy, stair fall	het	de novo	no
DGKZ	NM_001199266.1: c.3227C>G p.(Thr1076Arg)	NM_001199266.1:c.3326A>G p.(Gln1109Arg)	3.7	1	Epilepsy	Epileptic encephalopathy, Seizures, Failure to thrive, Hypoplasia of the corpus callosum, Hypsarrhythmia, Infantile onset, muscular hypotonia,	comphet	paternal & maternal	no
FBN3	NM_032447.4: c.6521C>T p.(Pro2174Leu)	NM_032447.4: c.4847G>A p.(Cys1616Tyr)	3.7		NDD + epilepsy	(+) Seizure,(+) Large for gestational age,(+) Microcephaly,(+) Global developmental delay,(+) Muscular hypotonia	comphet	paternal & maternal	no
NEU4	NM_001167599.2: c.1396T>C p.(Cys466Arg)	NM_001167599.2:c.407G>A p.(Arg136His)	3.6	2	Epilepsy	Focal impaired awareness seizures since 10/2016, EEG abnormality, delayed speech and language development, aggressive behavior	comphet	paternal & maternal	no
KLHDC4	NM_017566.3:c.908T>C p.(Met303Thr)	NM_017566.3:c.529C>T p.(Arg177Trp)	3.6	1	NDD + Epilepsy	Neurodevelopmental delay, Global developmental delay, Infantile spasms, Seizure, Epileptic spasm, Abnormal nervous system physiology, Neonatal seizure	comphet	paternal & maternal	no

DNHD1	NM_144666.2: c.3410G>A p.(Arg1137Gln)	NM_144666.2:c.2450A>C p.(His817Pro)	3.6	2	NDD	Retinal dystrophy, Microphthalmia, Delayed speech and language development, Global developmental delay, Poor speech, Vitreoretinopathy, Congenital blindness	comphet	paternal & maternal	no
SEMA3B	NM_001005914.2: c.952C>T p.(His318Tyr)	NM_001005914.2:c.728T>C p.(Phe243Ser)	3.6	3	NDD	global developmental delay, intellectual disability	comphet	paternal & maternal	no
SPATA31A3	NM_001083124.1: c.3206C>T p.(Ser1069Phe)		3.6	3	NDD + Epilepsy	Specific learning disability, Absence seizures, Cortical dysplasia, EEG with continuous slow activity, Seizures	het	de novo	no
TACC2	NM_206862.3: c.1407G>C p.(Glu469Asp)	NM_206862.3:c.1242G>C p.(Glu414Asp)	3.6	2	NDD	Macrocephaly, hypoplasia of the corpus callosum, suspected developmental delay,	comphet	paternal & maternal	no
PHRF1	NM_020901.3: c.1451+3G>A p.?	NM_020901.3:c.3544A>G p.(Lys1182Glu)	3.6	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	comphet	paternal & maternal	no
HMCN2	NM_001291815.1: c.4444G>T p.(Gly1482*)	NM_001291815.1:c.489+1G>C p.?	3.6	5	Epilepsy	Seizures, Generalized myoclonic seizures	comphet	paternal & maternal	no
H1-10	NM_006026.3: c.80C>T p.(Ser27Leu)		3.5	2	NDD	Retinal dystrophy, Microphthalmia, Delayed speech and language development, Global developmental delay, Poor speech, Vitreoretinopathy, Congenital blindness	homo	paternal & maternal	no
RGL4	NM_153615.1: c.101C>T p.(Thr34Met)		3.5	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 encephalopathy	het	de novo	no

ZSCAN10	NM_032805.2: c.1436C>A p.(Ser479Tyr)	NM_032805.2:c.2245G>T p.(Ala749Ser)	3.5	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	comphet	paternal & maternal	yes, maternal uncle with epilepsy and ID
B4GALNT3	NM_173593.3: c.1798G>A p.(Glu600Lys)	NM_173593.3:c.1640C>T p.(Pro547Leu)	3.5	4	NDD	Intellectual disability, Global developmental delay	comphet	paternal & maternal	no
PRRG3	NM_024082.3: c.572C>T p.(Pro191Leu)		3.5	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	hemi	maternal	no
CAST	het CAST deletion 1 point exon 16		3.5	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 dysphagia	het	de novo	no
MAP7D1	NM_018067.4: c.1225G>T p.(Ala409Ser)		3.5	1	Epilepsy	Infantile febrile seizures and tonic-clonic seizures with aura, despite current treatment with valproate, seizures continue	homo	paternal & maternal	no
AATF	NM_012138.3: c.695-3C>A p.?		3.5	3	NDD	Severe global developmental delay, sleep disturbance, behavioral abnormalities, flat face	het	de novo	no

CARMIL1	XM_005249221.1: c.3617C>T p.(Ser1206Leu)	XM_005249221.1:c.2659G>A p.(Glu887Lys)	3.5	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	comphet	paternal & maternal	no
DPY19L4	NM_181787.2: c.1256C>T p.(Ser419Phe)	NM_181787.2: c.1870C>T p.(Arg624*)	3.5	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	comphet	paternal & maternal	no
MCTP2	NM_018349.3:c.409G>A p.(Gly137Ser)	NM_018349.3:c.1889C>T p.(Pro630Leu)	3.5	2	NDD	Global developmental delay, macrocephaly and makrosomia, muscular hypotonia and ischemic stroke at four months of age	comphet	paternal & maternal	no
TMEM63A	NM_014698.2:c.1423T>C p.(Phe475Leu)		3.5	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	het	unknown	no
WNK2	NM_006648.3:c.5229G>T p.(Lys1743Asn)		3.5	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	het	unknown	no
SLC35B3	NM_001142540.1: c.1135C>T p.(Pro379Ser)	NM_001142540.1:c.1069G>C p.(Gly357Arg)	3.5	3	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormality of the caudate nucleus, Infantile onset, Attention deficit hyperactivity disorder	comphet	paternal & maternal	no
ARCM3	NM_173081.4: c.1346G>A p.(Arg449His)		3.4	2	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Focal seizures, Intellectual disability, severe	homo	paternal & maternal	no

LCN1	NM_001252618.1: c.305A>G p.(His102Arg)		3.4	1	NDD	Tall stature, delayed speech and language development, neuroblastoma	het	de novo	no
DACH2	NM_053281.3: c.1519G>T p.(Val507Phe)		3.4	2	NDD	Macrocephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Intellectual disability, severe, Cognitive impairment	hemi	maternal	no
GPKOW	NM_015698.4: c.1334G>A p.(Arg445Gln)		3.4	1	NDD	Autism, Global developmental delay	hemi	maternal	no
CFAP54	XM_001715090.5: c.2257A>G p.(Met753Val)	XM_001715090.5:c. 2057G>A p.(Arg686Lys)	3.4	3	NDD	Coloboma, Iris coloboma, mild Intellectual disability, mild Global developmental delay	comphet	paternal & maternal	no
TCP11	NM_001093728.2: c.256A>G p.(Lys86Glu)		3.4	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 hypotonia	homo	paternal & maternal	no
SSBP2	NM_001256732.2:c. 566C>T p.(Pro189Leu)		3.4	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	het	unknown	no
ZHX1	NM_001017926.2: c.179A>G p.(Asn60Ser)	NM_001017926.2:c. 962C>T p.(Ala321Val)	3.4	2	Epilepsy	Hearing impairment, Visual impairment, Nystagmus, Seizures, Abnormality of the cerebrospinal fluid, Epileptic spasms, Abnormal CSF glucose level	comphet	paternal & maternal	no
ZSWIM5	NM_020883.1: c.2369G>A p.(Arg790His)	NM_020883.1:c.191 5C>T p.(Pro639Ser)	3.4	1	NDD + Epilepsy	Microcephaly, Hearing impairment, Myopia, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Febrile seizures	comphet	paternal & maternal	yes, mother ID, 2 uncles & grandmother ms ID

CDCA2	NM_152562.3: c.922A>G p.(Arg308Gly)	NM_152562.3:c.1634C>T p.(Thr545Ile)	3.4	3	NDD	hydrocephalus, intellectual disability, spastic paraplegia, global developmental delay, agenesis of corpus callosum, spastic paraparesis, intellectual disability, severe, cerebral palsy	comphet	paternal & maternal	no
SRCIN1	NM_025248.2:c.40C>T p.(Pro14Ser)		3.4	3	NDD	Global developmental delay, intellectual impairment, absent speech	homo	paternal & maternal	no
ANKRD33B	NM_001164440.1: c.784G>A p.(Glu262Lys)	NM_001164440.1:c.1421A>C p.(Glu474Ala)	3.3	1	NDD	Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, moderate, Intellectual disability, severe	comphet	paternal & maternal	no
ARL13A	NM_001162491.1: c.349G>C p.(Asp117His)		3.3	1	NDD	Intellectual disability, Global developmental delay, Hemiplegia/hemiparesis	hemi	maternal	no
FRMPD3	XM_042978.8: c.3538C>T p.(Arg1180Trp)		3.3	2	NDD	Microcephaly, Epicanthus, Intellectual disability, Global developmental delay, Plagiocephaly, Abnormal facial shape, Wide nasal base	hemi	maternal	no
ENOX2	NM_006375.2: c.148A>G p.(Met50Val)		3.3	2	NDD	Microcephaly, Underdeveloped nasal alae, Strabismus, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Postnatal microcephaly	hemi	maternal	no
GPATCH2	NM_018040.3: c.1167-8C>T p.?		3.3	2	NDD	Cleft palate, Hydrocephalus, Microcephaly, Retinopathy, Intellectual disability, Global developmental delay, Pes planus, Short stature, Pes valgus, Cognitive impairment, Cleft hard palate	het	de novo	no
STARD9	NM_020759.2: c.4624C>A p.(Leu1542Met)	NM_020759.2:c.1655G>T p.(Arg552Leu)	3.3	2	NDD	Seizures, Generalized tonic-clonic seizures, Myoclonic atonic seizures, Epileptic encephalopathy	comphet	paternal & maternal	no
FAT3	NM_001008781.2: c.12282-7G>A p.?	NM_001008781.2:c.10369-3C>T p.?	3.2	2	NDD + Epilepsy	Seizures, Global developmental delay, Epileptic encephalopathy	comphet	paternal & maternal	no
LANCL3	NM_001170331.2: c.1037G>A p.(Ser346Asn)		3.2	2	NDD + epilepsy	Neonatal hypoglycemia, Seizure, Global developmental delay	hemi	maternal	no
ARAP2	NM_015230.3: c.4037G>A p.(Arg1346Gln)	NM_015230.3:c.2257G>C p.(Glu753Gln)	3.2	1	NDD	N/A	comphet	paternal & maternal	no

ZNF12	NM_006956.2: c.670T>C p.(Ser224Pro)	NM_006956.2:c.1438G>A p.(Val480Ile)	3.1	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures	comphet	paternal & maternal	no
GNL3L	NM_001184819.1: c.884T>A p.(Leu295Gln)		3.1	1	NDD	Global developmental delay with delayed speech and language development and a suspected autism spectre disorder, makrosomia	hemi	maternal	no
CASS4	NM_001164114.1: c.1576G>A p.(Val526Ile)	NM_001164114.1:c.1421G>T p.(Arg474Leu)	3.1	4	NDD	global developmental delay, intellectual disability, generalized hypotonia,	comphet	paternal & maternal	no
PRSS41	NM_001135086.1: c.30_41dup p.(Leu11_Ala14dup)		3.0	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	homo	paternal & maternal	no
BAHCC1	NM_001080519.2: c.4691+5C>G		3.0	3	NDD	Seizures, Pachygyria, Delayed CNS myelination, Heterotopia, Periventricular gray matter heterotopia, Intracranial cystic lesion, Abnormality of brain morphology	het	de novo	no
GABRA2	NM_000807.3: c.438del p.(Arg147Glufs*12)		Growth, Skeletal	1	Growth, Skeletal	short stature, muscular hypotonia, micropenis, acromely, hydronephrosis, congenital GH-deficiency, hypogonadism	het	de novo	no
AHCTF1	NM_015446.4:c.5018_5019insGG p.(Ile1673Metfs*4)		Growth, Skeletal	3	Growth, Skeletal	Panhypopituitarism, Proportionate short stature, Short stature, Anterior pituitary hypoplasia, Ectopic posterior pituitary, Posterior pituitary hypoplasia	het	de novo	no
FBXW7	NM_033632.3: c.23_24del p.(Val8Glyfs*14)		Other	1	Other	(+) Brain neoplasm,(+) Ewing sarcoma	het	unknown	no
SLC30A5	NM_022902.4: c.832_836del p.(Ile278Phefs*33)		Fehlbildung	3	Fehlbildung	Cardiomyopathy, Hydrops fetalis, Noncompaction cardiomyopath	homo	paternal & maternal	yes, three affected siblings

KCNK9	NM_001282534.1: c.391C>T p.(Arg131Cys)		Muscle	1	Muscle	muscular hypotonia, tongue fasciculation, motor developmental delay	het	de novo	no
SRCIN1	NM_025248.2: p.(Gly415Cys)		Fehlbildung	1	Fehlbildung	Oral cleft, Congenital diaphragmatic hernia, Dextrocardia, Cerebellar cyst, Branchial cyst, Bone cyst	het	de novo	yes, habitual abortions, mother mit lip and cleft palate & epilepsy
SLTM	NM_001013843.2: c.2595G>A p.(Trp865*)		Neuro	1	Neuro	Muscular hypotonia, Hypometric saccades, Chorea, Mild conductive hearing impairment, Constipation, Scapular winging, Gait ataxia, Truncal ataxia, Motor delay	het	de novo	no
DST	NM_001144769.2: c.11762G>A p.(Arg3921Gln)	NM_001144769.2: c.227C>A p.(Ala76Glu)	Metabolism	1	Metabolism	hypotrophes Frühgeborenes, Mikrozephalie, Leberversagen, Cholestase, Herzinsuffizienz, Gastrointestinale Blutung, Hypoglykämie, intraventrikuläre Blutung	comphet	paternal & maternal	yes, brother died under same clinical phenotype, parents healthy
SHANK1	NM_016148.3: c.5324G>T p.(Gly1775Val)		Fehlbildung	3	Fehlbildung	Meningo- or exencephalocele occipital, right radius aplasia with shortened ulna, death in utero	homo	paternal & maternal	no
SRGAP3	NM_014850.3: c.2227+6_2227+9del p.?		Fehlbildung	1	Fehlbildung	Premature birth, Esophageal atresia, Spina bifida, Total anomalous pulmonary venous return	het	de novo	no
C7orf26	NM_024067.3:c.575_576insT p.(Ser193Ilefs*3)		Immunology	2	Immunology	Abnormal facial shape, Alopecia areata, Recurrent infections, Antinuclear antibody positivity, Paroxysmal nocturnal hemoglobinuria, Prolonged neonatal jaundice	het	de novo	no
TMEM35B	NM_001195156.1: c.289+2del p.?		Other		Other	+) Elevated serum alanine aminotransferase,(+) Elevated serum aspartate aminotransferase,(+) Abnormality of the liver,(+) Splenomegaly,(-) Wilson disease,(-) Niemann-Pick disease type D	homo	unknown	sister also affected with same variant homo
RNF20	NM_019592.6:c.2783G>A p.(Arg928His)		Metabolism	2	Metabolism	Obesity	het	de novo	no

KDR	NM_002253.3: c.3161_3162insAA p.(Tyr1054*)	Cardio	1	Cardio	Abnormal aortic morphology, Abdominal aortic aneurysm, Descending thoracic aorta aneurysm, Cerebral arterial thrombosis	het	unknown	no
GRIA1	NM_000827.3: c.81C>A p.(=)	Other	3	Other	Seizures, Hypoglycemia, Myopathy, Focal seizures, Ichthyosis, EEG with focal epileptiform discharges	homo	paternal & maternal	no
HIRA	NM_003325.4: c.194A>G p.(Gln65Arg)	Fehlbildung	2	Fehlbildung	Non-midline cleft lip and palate	het	maternal	yes, mother and aunt with bilateral cleft lip and palate
ACTN2	NM_001103.3:c.1108-2A>T p.?	Muscle	1	Muscle	Tetraparesis and muscle weakness since age of 51 years	het	paternal	no
HECTD1	NM_015382.3:c.6068G>T p.(Gly2023Val)	Metabolism	2	Metabolism	Obesity	homo	paternal & maternal	no
FADS1	NM_013402.4: c.247G>T p.(Ala83Ser)	Other	2	Other	Anemia, Fever, Recurrent fever, Refractory anemia	het	de novo	no
PLCB3	NM_000932.2: c.1792G>C p.(Glu598Gln)	Fehlbildung	1	Fehlbildung	Failure to thrive, Growth delay, Omphalocele, Double outlet right ventricle	het	de novo	no
STRN3	NM_001083893.2: c.542+2T>G p.?	Growth, Skeletal	2	Growth, Skeletal	Short stature (151cm), Short for gestational age	het	paternal	yes, father is also affected by short stature

LAMP2	NM_001122606.1:c.731C>G p.(Thr244Ser)	Metabolism	2	Metabolism	Myalgia, Hyperlipoproteinemia, Increased erythrocyte protoporphyrin concentration, Angioedema	hemi	maternal	yes, maternal halfsister with developmental delay and muscular disorder, brother with autism, other brother possibly with mitochondrial disorder, mother with hyperlipidemia, migraines and pubertas retarda
POLR2C	NM_032940.2:c.109del p.(Val37Serfs*8)	Immunology	2	Immunology	Abnormal facial shape, Alopecia areata, Recurrent infections, Antinuclear antibody positivity, Paroxysmal nocturnal hemoglobinuria, Prolonged neonatal jaundice	het	de novo	no
INTS7	NM_015434.3: NM_015434.3:c.2240G>T p.(Arg747Ile)	Fehlbildung	1	Fehlbildung	Microcephaly, Intrauterine growth retardation, Abnormal facial shape, Basal ganglia calcification, Cerebral calcification, Congenital intracerebral calcification	het	de novo	no
COG6	NM_020751.2: c.1209T>G p.(Ile403Met)	Connective Tissue	2	Connective Tissue	Syncope, Joint hypermobility, Recurrent fractures, Chronic pain, Dysesthesia	het	de novo	no
ADCY7	NM_001114.5: c.2866C>T p.(Arg956Trp)	Neuro	3	Neuro	Multifocal cerebral white matter abnormalities, Leukoencephalopathy, Migraine, Abnormal cerebellum morphology, Gait disturbance, Gait imbalance	het	de novo	no

LIMD1	NM_014240.3: c.1669C>T p.(His557Tyr)	NM_014240.3: c.1532C>T p.(Ala511Val)	Connective Tissue	3	Connective Tissue	Recurrent fractures, Patellar dislocation, Recurrent infections, Migraine, Asthma	comphet	paternal & maternal	yes, brother similarly affected, Mother with arterial tutuosity, migraine and fractures of the feet.
USP4	NM_003363.3: c.1748A>G p.(Tyr583Cys)		Metabolism	2	Metabolism	Myalgia, Hyperlipoproteinemia, Increased erythrocyte protoporphyrin concentration, Angioedema	homo	paternal & maternal	yes, maternal halfsister with developmental delay and muscular disorder, brother with autism, other brother possibly with mitochondrial disorder mother with hyperlipidemia, migraines and pubertas retarda
TPR	NM_003292.2: c.1038A>G p.(Ile346Met)	NM_003292.2:c.2380T>A p.(Ser794Thr)	Other	2	Other	Anemia, Fever, Recurrent fever, Refractory anemia	comphet	paternal & maternal	no
ARL8B	NM_018184.2: c.286A>T p.(Ile96Leu)		Neuro	2	Neuro	Abnormality of the corpus callosum, Agenesis of corpus callosum, Talipes equinovarus, Polymicrogyria, Myelomeningocele, Brainstem dysplasia, Dysplastic corpus callosum, Periventricular gray matter heterotopia	het	de novo	no
PKD1	NM_001009944.2: c.12436G>A p.(Val4146Ile)	NM_001009944.2:c.9718G>A p.(Ala3240Thr)	Metabolism	3	Metabolism	Obesity, Increased adipose tissue, Glioma, Class III obesity, Overweight, Brain neoplasm	comphet	paternal & maternal	no

RYR3	NM_001036.4:c.2770A>G p.(Thr924Ala)	NM_001036.4:c.11246-5C>G p.?	Neuro	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	comphet	paternal & maternal	no
RIC8B	NM_001330145.1:c.399G>C p.(Gln133His)		Neuro	3	Neuro	Sudden spastic of lower extremitiesa and bowel incontinence at the age of 43 years	het	de novo	no
HEPHL1	NM_001098672.1:c.1097G>A p.(Cys366Tyr)		Connective Tissue	2	Connective Tissue	Syncope, Joint hypermobility, Recurrent fractures, Chronic pain, Dysesthesia	het	de novo	no
CX3CR1	NM_001171174.1:c.756del p.(Cys253Alafs*12)		Neuro	2	Neuro	Familial predisposition, Migraine, EEG abnormality, Episodic hemiplegia, Left hemiplegia	het	maternal	yes, Mother with hemiplegic migraine
USP8	NM_001128610.2:c.2658+2_2658+3insAAGA p.?	NM_001128610.2:c.2371A>G p.(Ile791Val)	Muscle	1	Muscle	Spasticity, Intention tremor, Vertigo, Dyskinesia	comphet	paternal & maternal	no
PODN	NM_001199080.2:c.559-1G>C p.?		Growth, Skeletal	2	Growth, Skeletal	Joint hypermobility, Asymmetry of the thorax, Scoliosis	het	de novo	no
MRM3	NM_018146.3:c.173C>G p.(Pro58Arg)		Metabolism	3	Metabolism	Ketotic hypoglycemia	het	de novo	no
GPX4	NM_001039848.3:c.587+5G>A p.?	NM_001039848.3:c.475G>T p.(Gly159Cys)	Neuro	2	Neuro	Gait disturbance, Dystonia, Progressive spastic paraplegia, Paraplegia, Leg dystonia	comphet	paternal & maternal	no
PACSIN3	NM_001184974.1:c.604-3C>G p.?		Metabolism	3	Metabolism	Ketotic hypoglycemia	het	de novo	no
SLC25A14	NM_001282197.1:c.124G>C p.(Val42Leu)		Neuro	3	Neuro	Sudden spastic of lower extremitiesa and bowel incontinence at the age of 43 years	hemi	maternal	no

FAM199X	NM_207318.4: c.932T>G p.(Met311Arg)		Connective Tissue	3	Connective Tissue	Recurrent fractures, Patellar dislocation, Recurrent infections, Migraine, Asthma	het	de novo	yes, brother similarly affected, Mother with arterial tutuosity, migraine and fractures of the feet.
TBC1D9B	NM_198868.2: c.583G>T p.(Ala195Ser)		Neuro	1	Neuro	Abnormality of the optic nerve, Optic atrophy, Polyneuropathy, Encephalopathy, Leukoencephalopathy, Leukodystrophy, Tetraplegia	het	de novo	no
CLTCL1	NM_007098.3: c.1820A>G p.(His607Arg)	NM_007098.3:c.2791A>G p.(Ile931Val)	Cardio	1	Cardio	Motor delay, Ventricular septal defect, Tracheal stenosis, Abnormal trachea morphology, Pulmonary artery atresia, Pulmonary valve atresia, Abnormality of cardiovascular system morphology	comphet	paternal & maternal	no
MST1	NM_020998.3: c.1603C>G p.(Arg535Gly)		Neuro	2	Neuro	Migraine, Migraine with aura, Migraine without aura, Cortical dysplasia, Frontoparietal cortical dysplasia	het	de novo	no
SLC38A1	NM_001278390.1: c.529A>G p.(Ile177Val)		Neuro	1	Neuro	Seizure, Tremor, Hand tremor, Nevus, Focal-onset seizure, Abnormality of brain morphology	het	de novo	no
GORAB	NM_152281.2: c.383T>C p.(Ile128Thr)		Growth, Skeletal	2	Growth, Skeletal	Joint hypermobility, Asymmetry of the thorax, Scoliosis	het	de novo	no
AKAP13	NM_006738.5: c.914A>G p.(Gln305Arg)	NM_006738.5:c.8228A>C p.(Lys2743Thr)	Neuro	2	Neuro	Migraine, Migraine with aura, Migraine without aura, Cortical dysplasia, Frontoparietal cortical dysplasia	comphet	paternal & maternal	no
PARPBP	NM_001319988.1:c.62G>T p.(Arg21Leu)		Metabolism	2	Metabolism	Hypertension, Irregular menstruation, Obesity, Hyperuricemia, Red hair, Narcolepsy, Increased blood pressure, Adipositas since the age of five to eight years	het	de novo	no

CCNT2	NM_058241.3: c.370-3_370-2insA p.?		Immunology	1	Immunology	Increased circulating IgG4 level, Neutropenia, Papule, Folliculitis, Lymphangitis, Pustule, Immunodeficiency	het	de novo	no
PPM1G	NM_177983.2: c.1579T>C p.(Ser527Pro)		Metabolism	1	Metabolism	GDD, adipositas, unbalanced glucosetolerance, massive hyperinsulinamia, muscular hypotonia,	homo	paternal & maternal	consanguinity
ANXA3	NM_005139.2:c.541-2A>G p.?		Neuro	1	Neuro	+) Sleep disturbance, Restless legs, Insomnia	het	paternal	yes, father and paternal aunt similary affected
EMC9	NM_016049.3: c.158A>T p.(His53Leu)		Fehlbildung	2	Fehlbildung	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	het	de novo	no
SYT3	NM_001160328.1: c.401C>G p.(Ala134Gly)		Fehlbildung	3	Fehlbildung	Meningo- or exencephalocele occipital, right radius aplasia with shortened ulna, death in utero	homo	paternal & maternal	no
TMEM181	NM_020823.1:c.448del p.(Ser150Profs*18)	NM_020823.1:c.1781C>T p.(Pro594Leu)	Fehlbildung	1	Fehlbildung	Growth delay, Intrauterine growth retardation	comphet	paternal & maternal	no
ZNF449	NM_152695.5: c.1394G>A p.(Cys465Tyr)		Growth, Skeletal	2	Growth, Skeletal	Growth delay, short stature, intrauterine growth retardation, Silver-Russell-like appearance	het	de novo	yes, brother affected so rather XR/AR

ARFGEF3	NM_020340.4: c.421-4A>G p.?	NM_020340.4:c.2003C>T p.(Ala668Val)	Neuro	2	Neuro	Abnormality of the corpus callosum, Agenesis of corpus callosum, Talipes equinovarus, Polymicrogyria, Myelomeningocele, Brainstem dysplasia, Dysplastic corpus callosum, Periventricular gray matter heterotopia	comphet	paternal & maternal	no
TMEM92	NM_001168215.1:c.212G>C p.(Cys71Ser)		Fehlbildung	1	Fehlbildung	inguinal hernia, cryptorchidism, hypospadias, renal cyst, hemangioma, bicuspid aortic valve, coarctation of aorta, anal atresia, choroid plexus cyst, abnormal vertebral morphology, multiple renal cysts	het	de novo	no
GPSM3	NM_001276501.1: c.318G>C p.(Gln106His)		Fehlbildung	2	Fehlbildung	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	het	de novo	no
TBCCD1	NM_001134415.1:c.1392T>G p.(Cys464Trp)		Metabolism	3	Metabolism	Ketotic hypoglycemia	het	de novo	no
SNX8	NM_013321.3: c.922C>T p.(Gln308*)		Growth, Skeletal	2	Growth, Skeletal	Growth delay, short stature, intrauterine growth retardation, Silver-Russell-like appearance	het	de novo	yes, brother affected so rather XR/AR
C6orf136	NM_001161376.1:c.478G>T p.(Ala160Ser)	NM_001161376.1:c.430C>T p.(Arg144Trp)	Metabolism	1	Metabolism	Hypoglycemia, Neonatal hypoglycemia	comphet	paternal & maternal	yes, brother with similar symptoms
TTC28	NM_001145418.1: c.5009A>T p.(His1670Leu)	NM_001145418.1: c.4237G>A p.(Gly1413Ser)	Growth, Skeletal	1	Growth, Skeletal	Trigonocephaly, Abnormality of calvarial morphology	comphet	paternal & maternal	no

RPS6KC1	NM_012424.4: c.2633G>A p.(Gly878Glu)		Fehlbildung	3	Fehlbildung	Meningo- or exencephalocele occipital, right radius aplasia with shortened ulna, death in utero	homo	paternal & maternal	no
ATG9A	NM_001077198.3: c.2398C>T p.(His800Tyr)		Growth, Skeletal	2	Growth, Skeletal	Short stature (151cm), Short for gestational age	het	paternal	yes, father is also affected by short stature
ERN2	NM_033266.3: c.2489C>T p.(Pro830Leu)		Metabolism	1	Metabolism	GDD, adipositas, unbalanced glucosetolerance, massive hyperinsulinamia, muscular hypotonia,	homo	paternal & maternal	consanguinity
PLEKHM3	NM_001080475.2:c.2219G>A p.(Arg740Lys)		Neuro	2	Neuro	Gait disturbance, Dystonia, Progressive spastic paraplegia, Paraplegia, Leg dystonia	het	de novo	no
FAM131A	NM_144635.4:c.838C>G p.(Leu280Val)		Muscle	1	Muscle	neuromuscular scoliosis, muscular hypotonia, intermittent exotropia	homo	paternal & maternal	no
PASK	NM_001252120.1: c.307G>A p.(Gly103Ser)	NM_001252120.1:c.2713C>T p.(Arg905Trp)	Neuro	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	comphet	paternal & maternal	no
TTLL4	NM_014640.5: c.2401C>G p.(Leu801Val)	NM_014640.5: c.2692G>A p.(Glu898Lys)	Neuro	3	Neuro	Multifocal cerebral white matter abnormalities, Leukoencephalopathy, Migraine, Abnormal cerebellum morphology, Gait disturbance, Gait imbalance	comphet	paternal & maternal	no
RAB11FIP4	NM_032932.5:c.1562G>A p.(Gly521Asp)		Immunology	2	Immunology	recurrent purulent abscess of the groin	het	maternal	yes, mother and maternal grandmother also affected by abscesses
SDR42E1	NM_145168.2: c.4G>A p.(Asp2Asn)		Metabolism	3	Metabolism	Obesity, Increased adipose tissue, Glioma, Class III obesity, Overweight, Brain neoplasm	homo	paternal & maternal	no

KLHL29	NM_052920.2: c.797C>T p.(Pro266Leu)		Neuro		Neuro	Behavioral abnormality, Frontotemporal dementia	het	de novo	no
FBN3	NM_032447.4: c.7780G>A p.(Val2594Ile)	NM_032447.4:c.1135C>T p.(Arg379*)	Metabolism	3	Metabolism	Obesity, Increased adipose tissue, Glioma, Class III obesity, Overweight, Brain neoplasm	comphet	paternal & maternal	no
RASGEF1A	NM_001282862.1: c.346-3del p.?		Fehlbildung	1	Fehlbildung	Meningocele, Hypoplastic nasal bridge, Increased nuchal translucency, Short fetal femur length	het	de novo	no
SFRP5	NM_003015.3:c.872T>C p.(Met291Thr)	NM_003015.3:c.861_863del p.(Lys287del)	Metabolism	2	Metabolism	Hypertension, Irregular menstruation, Obesity, Hyperuricemia, Red hair, Narcolepsy, Increased blood pressure, Adipositas since the age of five to eight years	comphet	paternal & maternal	no
COL20A1	NM_020882.2: c.3467G>T p.(Gly1156Val)	NM_020882.2:c.807C>A p.(Asn269Lys)	NDD	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay	comphet	paternal & maternal	no
TDRD6	NM_001010870.2:c.1895A>G p.(His632Arg)	NM_001010870.2:c.2566G>A p.(Asp856Asn)	Growth, Skeletal	3	Growth, Skeletal	Panhypopituitarism, Proportionate short stature, Short stature, Anterior pituitary hypoplasia, Ectopic posterior pituitary, Posterior pituitary hypoplasia	comphet	paternal & maternal	no
TRPC7	NM_020389.2: c.1577A>G p.(Tyr526Cys)		Neuro	3	Neuro	Sudden spastic of lower extremitiesa and bowel incontinence at the age of 43 years	homo	paternal & maternal	no
CRIM1	NM_016441.2: c.2867C>T p.(Ala956Val)	NM_016441.2:c.1658+4C>T p.?	Neuro	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	comphet	paternal & maternal	no
FBN3	NM_032447.5: c.6184G>A p.(Ala2062Thr)	NM_032447.5: c.4370A>G p.(Asn1457Ser)	Neuro	3	Neuro	Multifocal cerebral white matter abnormalities, Leukoencephalopathy, Migraine, Abnormal cerebellum morphology, Gait disturbance, Gait imbalance	comphet	paternal & maternal	no

EFHC2	NM_025184.3:c.975A>C p.(Leu325=)		Growth, Skeletal	3	Growth, Skeletal	Panhypopituitarism, Proportionate short stature, Short stature, Anterior pituitary hypoplasia, Ectopic posterior pituitary, Posterior pituitary hypoplasia	hemi	maternal	no
TEC	NM_003215.2:c.1526G>T p.(Gly509Val)		Immunology	2	Immunology	recurrent purulent abscess of the groin	het	maternal	yes, mother and maternal grandmother also affected by abscesses
RGMB	NM_001012761.3:c.863C>T p.(Thr288Ile)		Fehlbildung	2	Fehlbildung	Non-midline cleft lip and palate	het	maternal	yes, mother and aunt with bilateral cleft lip and palate
ARHGEF38	NM_001242729.2:c.1363_1365delACGinsGCA p.(Thr455Ala)	NM_001242729.2:c.2122G>A p.(Asp708Asn)	Metabolism	1	Metabolism	Diabetes insipidus, Central diabetes insipidus, Panhypopituitarism, Short stature, Proportionate short stature	comphet	paternal & maternal	no
GIT2	NM_057169.4:c.699T>G p.(Tyr233*)		Growth, Skeletal	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	het	de novo	N/A
LOXL4	NM_032211.6:c.396C>A p.(Cys132*)		Growth, Skeletal	1	Growth, Skeletal	(+) Small for gestational age,(+) Mild short stature,(+) Attention deficit hyperactivity disorder,(+) Delayed skeletal maturation,(+) Intrauterine growth retardation,(+) Mild intrauterine growth retardation	het	unknown	yes, mother also short
ARHGAP35	NM_004491.4:c.597_600del p.(Thr200Serfs*18)		Immunology	1	Immunology	Splenomegaly, Lymphopenia, recurrent infections, immunodeficiency, decreased circulating IGA, increased circulating IgM level	het	de novo	no

KIF5B	NM_004521.3: c.135_136dup p.(Tyr46Phefs*67)		Fehlbildung	1	Fehlbildung	Macrodactyly, Upper limb asymmetry, Hemihypertrophy of upper limb, Hyperextensible thumb	het	unknown	no
TMEM61	NM_182532.2: c.101G>C p.(Cys34Ser)	NM_182532.2: c.583G>A p.(Ala195Thr)	Wachstum, Skelett	2	Wachstum, Skelett	Hypoterlorism, Trigonocephaly	comphet	paternal & maternal	no
NIN	NM_020921.3: c.4760A>C p.(Gln1587Pro)	NM_020921.3: c.446C>T p.(Thr149Met)	Wachstum, Skelett	2	Wachstum, Skelett	Hypoterlorism, Trigonocephaly	comphet	paternal & maternal	no
TRHDE	NM_013381.2: c.1050_1052delTGTinsGGG p.(Val351Gly)		Wachstum, Skelett		Wachstum, Skelett	+) Arthrogryposis multiplex congenita,(+) Plagiocephaly,(+) Congenital finger flexion contractures,(+) Wrist flexion contracture,(+) Elbow flexion contracture,(+) Shoulder flexion contracture,(+) Adducted thumb,(+) Respiratory failure	het	de novo	Grandmother on the side of the father with clubfoot
GIGYF1	NM_022574.4: c.1778A>T p.(Asp593Val)		Wachstum, Skelett		Wachstum, Skelett	(+) Cleft soft palate,(+) Cleft hard palate	het	de novo	familary connective tissue weakness
MAP3K6	NM_004672.4: c.3789-5C>T p.?	NM_004672.4: c.1733T>A p.(Val578Asp)	Wachstum, Skelett		Wachstum, Skelett	(+) Cleft soft palate,(+) Cleft hard palate	comphet	paternal & maternal	familary connective tissue weakness