epiSEEK® Comprehensive Sequence Analysis of Epilepsy and Seizure Disorders (471 genes)

Koolen-de Vries syndrome	KANSL1
Angelman/Angelman-like/Pitt-Hopkins syndromes	CNTNAP2, MBD5, NRXN1, SLC9A6, TCF4, UBE3A
Autosomal Dominant Focal Epilepsies	CHRNA2, CHRNA4, CHRNB2, LGI1
Benign familial infantile seizures (BFIS)	PRRT2
Benign familial neonatal seizures (BFNS)	KCNQ2, KCNQ3
Benign familial neonatal-infantile seizures (BFNIS)	SCN2A
Cerebral folate deficiency	FOLR1
Creatine deficiency syndromes	GAMT, GATM, SLC6A8
EAST/SeSAME syndrome	KCNJ10
Familial infantile myoclonic epilepsy (FIME)	TBC1D24
Generalized epilepsy with febrile seizures plus (GEFS+)	GABRG2, SCN1A, SCN1B, SCN2A
Hyperekplexia	ARHGEF9, GLRA1, GLRB, GPHN, SLC6A5
Joubert Syndrome	AHI1, ARL13B, CC2D2A, CEP290, INPP5E, NPHP1, OFD1, RPGRIP1L, TMEM216, TMEM67
Juvenile Myoclonic Epilepsy (JME)	BRD2, CACNB4, EFHC1, GABRA1
Migraine	ATP1A2, BRAF, CACNA1A, NOTCH3, POLG, SCN1A, SLC2A1
Neuronal Ceroid Lipofuscinoses (NCL)	CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, KCTD7, MFSD8, PPT1, TPP1
Peroxisomal Biogenesis Disorders	PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26
Progressive Myoclonic Epilepsy	ASAH1, CSTB, EPM2A, FOLR1, GOSR2, KCTD7, NHLRC1, PRICKLE1, SCARB2
Ras/MAPK pathway dysregulation (Costello, Noonan, LEOPARD, Legius, Cardio-facio-cutaneous, other)	BRAF, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, SOS1, SPRED1
Rett/atypical Rett syndromes	CDKL5, FOXG1, MBD5, MECP2, MEF2C

Brain or Nervous System Malformations

AHI1, ARFGEF2, ARL13B, ASPM, ATR, BRAF, BUB1B, C12orf57, CASK, CBL, CC2D2A, CCDC88C, CDK5RAP2, CDON, CENPJ, CEP152, CEP290, COL18A1, COL4A1, CPT2, DCLK2, DCX, ELP4, EMX2, EOMES, FGF8, FGFR3, FKRP, FKTN, FLNA, GLI2, GLI3, GPR56, KAT6B, LAMA2, LARGE, LIG4, MCPH1, MED17, MLC1, NHEJ1, NPHP1, OFD1, PAFAH1B1, PAX6, PCNT, PEX7, PIK3CA, PIK3R2, PLA2G6, PNKP, POMGNT1, POMT1, POMT2, PQBP1, PTCH1, RAB3GAP1, RARS2, RELN, SERPINI1, SHH, SHOC2, SIX3, SLC25A19,SNAP25, SNAP29, SRPX2, STIL, STRADA, TGIF1, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB2B, VANGL1, VRK1, WDR62, ZIC2

Congenital Disorder of Glycosylation

ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, B4GALT1, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DOLK, DPAGT1, DPM1, DPM3, MAGT1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PMM2, RFT1, SLC35A1, SLC35A2, SLC35C1, SRD5A3, TMEM165, TUSC3

Drug Metabolism and Cannabinoid Receptors and Pathways

AKT3, CNR1, CNR2, CYP2C19, CYP2C9, CYP3A4, CYP3A5, DAGLA, FAAH, GPR55, MGLL, MTOR

Early onset Epileptic Encephalopathies or Infantile Spasms

ALDH7A1, ARHGEF9, ARX, ATP6AP2, CDKL5, CNTNAP2, FH, FOXG1, GABRG2, GRIN2A, GRIN2B, HNRNPU, KCNQ2, KCNT1, LIAS, MAGI2, MEF2C, NEDD4L, NRXN1, PCDH19, PLCB1, PNPO, POLG, PRRT2, SCN1A, SCN1B, SCN2A, SCN2B, SCN8A, SCN9A, SLC25A22, SLC2A1, SLC6A4, SLC9A6, SPTAN1, ST3GAL3, STXBP1, TCF4, TSC1, TSC2, ZEB2

Epilepsy in intellectual disability and behavioral disorders

AGTR2, ARHGEF9, ARX, ATP6AP2, ATP6V0A2, ATP7A, ATRX, CASK, CDKL5, CHRNA7, CNTNAP2, CUL4B, DCX, DLGAP2, FGD1, GLRA1, GPC3, GRIA3, GRIN2A, GRIN2B, HSD17B10, IQSEC2, KCNJ10, KDM5C, MAGT1, MECP2, MED12, OFD1, OPHN1, PAK3, PCDH19, PHF6, PIGV, PLP1, PQBP1, RAB39B, SLC16A2, SLC9A6, SMC1A, SMPD1, SMS, SRPX2, SYN1, SYNGAP1, SYP

GABA Receptors

GABBR1, GABBR2, GABRA2, GABRA3, GABRA4, GABRA5, GABRA6, GABRB1, GABRB2, GABRE, GABRG1, GABRG3, GABRP, GABRQ, GABRR1, GABRR2, GABRR3

Generalized/Myoclonic/Absence Epilepsies/Febrile Seizures

ALDH7A1, ATN1, BRAT1, CACNA1A, CACNA1H, CACNB4, CASR, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CNTN2, CSTB, DEPDC5, EFHC1 ,EFHC2, EPM2A, GABRA, GABRB3, GABRD, GABRG2, GOSR2, GPR98, GRIN1, GRIN2A, GRIN2B, JRK, KCNMA1, KCNQ2, KCNQ3, KCTD7, MBD5, ME2, NHLRC1, PCDH19, PRICKLE1, PRICKLE2, PRRT2, SCARB2, SCN10A, SCN11A, SCN1A, SCN1B, SCN2A, SCN3A, SCN3B, SCN4A, SCN4B, SCN5A, SCN7A, SCN8A, SCN9A, SLC2A1, TBC1D24

Mitochondrial Dysfunction

ADCK3, APTX, BCS1L, C12orf65, COQ2, COQ9, COX10, COX15, DLD, LRPPRC, NDUFA2, NDUFAF6, NDUFS1, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, OPA1, PC, PDHA1, PDHX, PDSS1, PDSS2, POLG, SCO2, SDHA, SURF1, TACO1, VDAC1

Protein and Carbohydrate Metabolism

ABCC8, ACY1, ADSL, AGA, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATPAF2, BTD, CTSA, DPYD, ETFA, ETFB, ETFDH, FH, FOLR1, FUCA1, GCDH, GCSH, GLDC, GNE, HPD, HYAL1, L2HGDH, MOCS1, MOCS2, NEU1, PANK2, PGK1, PNPO, PRODH, QDPR, SLC17A5, SLC25A15, SLC46A1, SUMF1, SUOX

Selected inborn errors of metabolism

ABAT, ACOX1, ATIC, ATP5A1, ATP7A, BCKDHA, BCKDHB, C12orf65, DBT, DDC, DHCR7, GLUD1, GLUL, HSD17B10, HSD17B4, KCNJ11, MGME1, MMACHC, MTHFR, MTR, MTRR, NDUFA1, PHGDH, PSAT1, SLC19A3, SUCLA2, TMEM70, VDAC1

Storage diseases and organelle dysfunction including lysosomal storage disorders and leukodystrophies

ARSA, ARSB, ASPA, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FUCA1, GALC, GALNS, GFAP, GLB1, GNE, GNPTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, IDS, IDUA, MCOLN1, MLC1, NAGLU, NOTCH3, NPC1, NPC2, PLP1, PSAP, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SDHA, SGSH, SMPD1, SUMF1, TREX1

Syndromic disorders with epilepsy and others

ABCC2, ASPA, ATP1A2, ATP1A3, ATP2A2, BCKDK, CCL2, CLCNKA, CLCNKB, CNTNAP2, CPT1A, DYRK1A, FGD1, FLVCR2, FOXH1, GJD2, GUSB, HCN1, HCN2, HCN3, HCN4, HERC2, IDH2, KCNA1, KCNAB1, KCNJ1, KCNV2, KIAA1279, KMT2D, LBR, LGI1, MAPK10, NDE1, NIPBL, NODAL, RAI1, RBFOX1, RNASEH2C, RTTN, SETBP1, SGCE, SLC1A3, SLC4A10, SMC1A, SMC3, SNIP1, ST3GAL5, TBX1, TRPM6, VPS13A, VPS13B, ZEB2