MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Epilepsy/Seizure Genetic Panels

Reference transcripts based on build GRCh37 (hg19) interrogated by Epilepsy/Seizure Genetic Panels

Epilepsy Expanded Panel	
Gene	**GenBank Accession Number**
ABAT	NM_020686
ACY1	NM_000666
ADGRG1	NM_005682
ADSL	NM_000026
AFG3L2	NM_006796
ALDH7A1	NM_001182
ALG13	NM_001099922
AMT	NM_000481
ARFGEF2	NM_006420
ARHGEF9	NM_015185, NM_001173479
ARX	NM_139058
ASAH1	NM_177924
ATP13A2	NM_022089
ATP1A2	NM_000702
ATP6AP2	NM_005765
ATRX	NM_000489
BCKDK	NM_005881
BOLA3	NM_212552
CACNA1A	NM_001127221
CACNA2D2	NM_006030
CASK	NM_003688
CDKL5	NM_003159
CERS1	NM_021267
CHD2	NM_001271
CHRNA2	NM_000742
CHRNA4	NM_000744
CHRNB2	NM_000748
CLCN4	NM_001830
CLN3	NM_001042432
CLN5	NM_006493
CLN6	NM_017882
CLN8	NM_018941
CNTNAP2	NM_014141
COG7	NM_153603
COG8	NM_032382
COL18A1	ENST00000400337
COL4A1	NM_001845
COQ9	NM_020312

Epilepsy Expanded Panel	
Gene	**GenBank Accession Number**
CPA6	NM_020361
CPT2	NM_000098
CRH	NM_000756
CSTB	NM_000100
CTSD	NM_001909
CTSF	NM_003793
CUL4B	NM_003588
D2HGDH	NM_152783
DCX	NM_178153
DEPDC5	NM_001242896
DNAJC5	NM_025219
DNM1	NM_004408
DOCK7	NM_001271999
DYRK1A	NM_001396
EEF1A2	NM_001958
EPM2A	NM_005670
FARS2	NM_006567
FASTKD2	NM_014929
FGD1	NM_004463
FGFR3	NM_000142
FH	NM_000143
FKRP	NM_024301
FKTN	NM_001079802
FLNA	NM_001456
FOLR1	NM_016725
FOXG1	NM_005249
GABRA1	NM_000806
GABRB2	NM_021911
GABRB3	NM_000814
GABRD	NM_000815
GABRG2	NM_000816
GAMT	NM_000156
GATM	NM_001482
GCK	NM_000162
GFM1	NM_024996
GLUL	NM_002065
GNAO1	NM_020988
GOSR2	NM_004287
GPC3	NM_004484

Epilepsy Expanded Panel	
Gene	**GenBank Accession Number**
GRIA3	NM_000828
GRIN1	NM_007327
GRIN2A	NM_000833
GRIN2B	NM_000834
GRN	NM_002087
HCFC1	NM_005334
HCN1	NM_021072
HSD17B10	NM_004493
IBA57	NM_001010867
IER3IP1	NM_016097
KANSL1	NM_001193466
KCNB1	NM_004975
KCNC1	NM_001112741
KCNH5	NM_139318
KCNJ10	NM_002241
KCNQ2	NM_172107
KCNQ3	NM_004519
KCNT1	NM_020822
KCTD7	NM_153033
KDM5C	NM_004187
LAMA2	NM_000426
LARGE1	NM_004737
LGI1	NM_005097
MBD5	NM_018328
MECP2	NM_004992, NM_001110792
MEF2C	NM_002397
MFSD8	NM_152778
MOCS1	NM_005943, NM_001075098
MOCS2	NM_176806, NM_004531
MRPL12	NM_002949
NECAP1	NM_015509
NEU1	NM_000434
NHLRC1	NM_198586
NOTCH3	NM_000435
NPRL2	NM_006545
*NPRL3**	NM_001077350
NR2F1	NM_005654
NRXN1	NM_001135659
OCLN	NM_002538
OFD1	NM_003611
OPHN1	NM_002547

Epilepsy Expanded Panel	
Gene	**GenBank Accession Number**
PAFAH1B1	NM_000430
PAK3	NM_002578
PCDH19	NM_001184880
PDSS2	NM_020381
PEX7	NM_000288
PHF6	NM_032458
PHGDH	NM_006623
PIGA	NM_002641
PIGO	NM_032634
PIGV	NM_017837
PLCB1	NM_015192
PLP1	NM_000533
PNKP	NM_007254
PNPO	NM_018129
POLG	NM_002693
POMGNT1	NM_017739
POMT1	NM_007171
POMT2	NM_013382
PPT1	NM_000310
PQBP1	NM_005710
PRICKLE1	NM_153026
PRRT2	NM_145239
PURA	NM_005859
QARS	NM_005051
RAB39B	NM_171998
RAB3GAP1	NM_012233
RELN	NM_005045
RMND1	NM_017909
ROGDI	NM_024589
SCARB2	NM_005506
SCN1A	NM_001165963
SCN1B	NM_001037
SCN2A	NM_021007
SCN8A	NM_014191
SCN9A	NM_002977
SERPINI1	NM_005025
SETBP1	NM_015559
SIK1	NM_173354
SLC13A5	NM_177550
SLC19A3	NM_025243
SLC25A22	NM_024698
SLC2A1	NM_006516
SLC35A2	NM_001042498
SLC6A8	NM_005629

Epilepsy Expanded Panel	
Gene	**GenBank Accession Number**
SLC9A6	NM_006359, NM_001042537
SMC1A	NM_006306
SMS	NM_004595
SNAP29	NM_004782
SPR	NM_003124
SPTAN1	NM_001130438
SRPX2	NM_014467
ST3GAL3	NM_006279
ST3GAL5	NM_003896
STX1B	NM_052874
STXBP1	NM_003165
SYNGAP1	NM_006772
SYP	NM_003179
SZT2	NM_015284
TBC1D24	NM_001199107
TCF4	NM_001083962
TPP1	NM_000391
TSC1	NM_000368
TSC2	NM_000548
TUBA1A	NM_006009
TUBA8	NM_018943
TUBB2B	NM_178012
TWNK	NM_021830
UBE3A	NM_130838
VARS2	NM_001167734
VLDLR	NM_003383
WDR45	NM_007075
WDR62	NM_001083961
WWOX	NM_016373
ZEB2	NM_014795

*Reference transcript based on build GRCh38 (hg38) for the NPRL3 gene.

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes. Additionally, NGS is used to test for the presence of large deletions and/or duplications.

There are regions of the OCLN gene that cannot be effectively amplified and sequenced due to regions of homology.

There are regions of the *AFLG3L2*, *CERS1*, *COL18A1*, *COL4A1*, *CTSF*, *GABRD*, *KANSL1*, *MOCS1*, *NOTCH3*, *OCLN*, *PEX7*, *SCN1B*, and *SYNGAP1* genes that cannot be effectively analyzed for the presence of large deletions and/or duplications as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

Multiplex Ligation-Dependent Probe Amplification (MLPA), PCR, and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Encephalopathy with Seizures Panel	
Gene	**GenBank Accession Number**
ABAT	NM_020686
ACY1	NM_000666
ADSL	NM_000026
ALDH7A1	NM_001182
AMT	NM_000481
ARFGEF2	NM_006420
ARHGEF9	NM_015185, NM_001173479
ARX	NM_139058
ATP6AP2	NM_005765
ATRX	NM_000489
BCKDK	NM_005881
BOLA3	NM_212552
CACNA2D2	NM_006030
CASK	NM_003688
CDKL5	NM_003159
CHD2	NM_001271
CLCN4	NM_001830
CLN3	NM_001042432
CLN5	NM_006493
CLN6	NM_017882
CLN8	NM_018941
CNTNAP2	NM_014141
COG7	NM_153603
COG8	NM_032382
COQ9	NM_020312
CSTB	NM_000100
CTSD	NM_001909
CTSF	NM_003793
CUL4B	NM_003588
D2HGDH	NM_152783
DCX	NM_178153
DNM1	NM_004408
DOCK7	NM_001271999
EEF1A2	NM_001958
EPM2A	NM_005670
FARS2	NM_006567
FASTKD2	NM_014929
FGD1	NM_004463
FH	NM_000143
FOLR1	NM_016725
FOXG1	NM_005249
GABRA1	NM_000806
GABRB3	NM_000814

Encephalopathy with Seizures Panel	
Gene	**GenBank Accession Number**
GABRG2	NM_000816
GAMT	NM_000156
GCK	NM_000162
GFM1	NM_024996
GLUL	NM_002065
GNAO1	NM_020988
GOSR2	NM_004287
GPC3	NM_004484
GRIA3	NM_000828
GRIN1	NM_007327
GRIN2A	NM_000833
GRIN2B	NM_000834
HCFC1	NM_005334
HCN1	NM_021072
HSD17B10	NM_004493
IBA57	NM_001010867
IER3IP1	NM_016097
KCNB1	NM_004975
KCNH5	NM_139318
KCNJ10	NM_002241
KCNQ2	NM_172107
KCNQ3	NM_004519
KCNT1	NM_020822
KCTD7	NM_153033
KDM5C	NM_004187
MBD5	NM_018328
MECP2	NM_004992, NM_001110792
MEF2C	NM_002397
MFSD8	NM_152778
MOCS1	NM_005943, NM_001075098
MOCS2	NM_176806, NM_004531
MRPL12	NM_002949
NECAP1	NM_015509
NHLRC1	NM_198586
NRXN1	NM_001135659
OCLN	NM_002538
OFD1	NM_003611
OPHN1	NM_002547
PAK3	NM_002578
PCDH19	NM_001184880
PDSS2	NM_020381

Encephalopathy with Seizures Panel	
Gene	**GenBank Accession Number**
PHF6	NM_032458
PHGDH	NM_006623
PIGO	NM_032634
PLCB1	NM_015192
PLP1	NM_000533
PNKP	NM_007254
PNPO	NM_018129
POLG	NM_002693
PPT1	NM_000310
PQBP1	NM_005710
PURA	NM_005859
RAB39B	NM_171998
RMND1	NM_017909
ROGDI	NM_024589
SCARB2	NM_005506
SCN1A	NM_001165963
SCN1B	NM_001037
SCN2A	NM_021007
SCN8A	NM_014191
SCN9A	NM_002977
SERPINI1	NM_005025
SLC13A5	NM_177550
SLC19A3	NM_025243
SLC25A22	NM_024698
SLC2A1	NM_006516
SLC35A2	NM_001042498
SLC9A6	NM_006359, NM_001042537
SMC1A	NM_006306
SMS	NM_004595
SPR	NM_003124
SPTAN1	NM_001130438
SRPX2	NM_014467
ST3GAL3	NM_006279
ST3GAL5	NM_003896
STXBP1	NM_003165
SYNGAP1	NM_006772
SYP	NM_003179
SZT2	NM_015284
TBC1D24	NM_001199107
TPP1	NM_000391
TSC1	NM_000368
TSC2	NM_000548
TWNK	NM_021830

Encephalopathy with Seizures Panel	
Gene	**GenBank Accession Number**
VARS2	NM_001167734
WWOX	NM_016373

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes. Additionally, NGS is used to test for the presence of large deletions and/or duplications.

There are regions of the *OCLN* gene that cannot be effectively amplified and sequenced due to regions of homology.

There are regions of the *CTSF*, *MOCS1*, *OCLN*, *SCN1B*, and *SYNGAP1* genes that cannot be effectively analyzed for the presence of large deletions and/or duplications as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

Multiplex Ligation-Dependent Probe Amplification (MLPA), PCR, and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Early Epileptic Encephalopathy Panel	
Gene	**GenBank Accession Number**
ACY1	NM_000666
ADSL	NM_000026
ALDH7A1	NM_001182
ALG13	NM_001099922
ARFGEF2	NM_006420
ARHGEF9	NM_015185, NM_001173479
ARX	NM_139058
ATP6AP2	NM_005765
BCKDK	NM_005881
CACNA1A	NM_001127221
CDKL5	NM_003159
CHD2	NM_001271
CLN3	NM_001042432
CLN5	NM_006493
CLN6	NM_017882
CLN8	NM_018941
CNTNAP2	NM_014141
CSTB	NM_000100
CTSD	NM_001909
CTSF	NM_003793
DNM1	NM_004408
DYRK1A	NM_001396
EEF1A2	NM_001958
EPM2A	NM_005670
FARS2	NM_006567
FOLR1	NM_016725
FOXG1	NM_005249
GABRA1	NM_000806
GABRB2	NM_021911
GABRB3	NM_000814
GABRG2	NM_000816
GAMT	NM_000156
GATM	NM_001482
GOSR2	NM_004287
GRIN1	NM_007327
GRIN2A	NM_000833
GRIN2B	NM_000834
KANSL1	NM_001193466
KCNB1	NM_004975
KCNJ10	NM_002241
KCNQ2	NM_172107
KCNQ3	NM_004519
KCNT1	NM_020822

Early Epileptic Encephalopathy Panel	
Gene	**GenBank Accession Number**
KCTD7	NM_153033
MBD5	NM_018328
MECP2	NM_004992, NM_001110792
MEF2C	NM_002397
MFSD8	NM_152778
NHLRC1	NM_198586
NR2F1	NM_005654
NRXN1	NM_001135659
PCDH19	NM_001184880
PIGA	NM_002641
PIGO	NM_032634
PIGV	NM_017837
PLCB1	NM_015192
PNKP	NM_007254
PNPO	NM_018129
POLG	NM_002693
PPT1	NM_000310
PRRT2	NM_145239
QARS	NM_005051
ROGDI	NM_024589
SCARB2	NM_005506
SCN1A	NM_001165963
SCN1B	NM_001037
SCN2A	NM_021007
SCN8A	NM_014191
SCN9A	NM_002977
SETBP1	NM_015559
SIK1	NM_173354
SLC19A3	NM_025243
SLC25A22	NM_024698
SLC2A1	NM_006516
SLC6A8	NM_005629
SLC9A6	NM_006359, NM_001042537
SPTAN1	NM_001130438
ST3GAL3	NM_006279
ST3GAL5	NM_003896
STXBP1	NM_003165
SZT2	NM_015284
TBC1D24	NM_001199107
TCF4	NM_001083962
TPP1	NM_000391
TSC1	NM_000368

Early Epileptic Encephalopathy Panel	
Gene	**GenBank Accession Number**
TSC2	NM_000548
UBE3A	NM_130838
WDR45	NM_007075
WWOX	NM_016373
ZEB2	NM_014795

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes. Additionally, NGS is used to test for the presence of large deletions and/or duplications.

There are regions of the *CTSF*, *KANSL1*, and *SCN1B* genes that cannot be effectively analyzed for the presence of large deletions and/or duplications as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

Multiplex Ligation-Dependent Probe Amplification (*MLPA*), *PCR*, and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Neuronal Migration Disorders Panel	
Gene	**GenBank Accession Number**
ADGRG1	NM_005682
ARFGEF2	NM_006420
ARX	NM_139058
COL18A1	ENST00000400337
COL4A1	NM_001845
CPT2	NM_000098
DCX	NM_178153
DEPDC5	NM_001242896
FGFR3	NM_000142
FKRP	NM_024301
FKTN	NM_001079802
FLNA	NM_001456
LAMA2	NM_000426
LARGE1	NM_004737
PAFAH1B1	NM_000430
PEX7	NM_000288
POMGNT1	NM_017739
POMT1	NM_007171
POMT2	NM_013382
PQBP1	NM_005710
RAB3GAP1	NM_012233
RELN	NM_005045
SNAP29	NM_004782
SRPX2	NM_014467
TUBA1A	NM_006009
TUBA8	NM_018943
TUBB2B	NM_178012
VLDLR	NM_003383
WDR62	NM_001083961

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes. Additionally, NGS is used to test for the presence of large deletions and/or duplications.

There are regions of the *COL18A1*, *COL4A1*, and *PEX7* genes that cannot be effectively analyzed for the presence of large deletions and/or duplications as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

PCR and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Progressive Myoclonic Epilepsy Panel	
Gene	**GenBank Accession Number**
AFG3L2	NM_006796
ASAH1	NM_177924
ATP13A2	NM_022089
CERS1	NM_021267
CLN3	NM_001042432
CLN5	NM_006493
CLN6	NM_017882
CLN8	NM_018941
CSTB	NM_000100
CTSD	NM_001909
CTSF	NM_003793
DNAJC5	NM_025219
EPM2A	NM_005670
FOLR1	NM_016725
GOSR2	NM_004287
GRN	NM_002087
KCNC1	NM_001112741
KCTD7	NM_153033
MFSD8	NM_152778
NEU1	NM_000434
NHLRC1	NM_198586
PPT1	NM_000310
PRICKLE1	NM_153026
SCARB2	NM_005506
SERPINI1	NM_005025
TBC1D24	NM_001199107
TPP1	NM_000391

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes. Additionally, NGS is used to test for the presence of large deletions and/or duplications.

There are regions of the *AFLG3L2*, *CERS1*, and *CTSF* genes that cannot be effectively analyzed for the presence of large deletions and/or duplications as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

PCR and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Infantile Spasms Panel	
Gene	**GenBank Accession Number**
ALDH7A1	NM_001182
ARFGEF2	NM_006420
ARX	NM_139058
CDKL5	NM_003159
COL4A1	NM_001845
FOXG1	NM_005249
GABRB3	NM_000814
GRIN2A	NM_000833
MEF2C	NM_002397
PNPO	NM_018129
SCN2A	NM_021007
SLC25A22	NM_024698
SLC2A1	NM_006516
SPTAN1	NM_001130438
STXBP1	NM_003165
TSC1	NM_000368
TSC2	NM_000548

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes. Additionally, NGS is used to test for the presence of large deletions and/or duplications.

There are regions of the *COL4A1* gene that cannot be effectively analyzed for the presence of large deletions and/or duplications as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

PCR and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Focal Epilepsy Panel	
Gene	**GenBank Accession Number**
CHRNA2	NM_000742
CHRNA4	NM_000744
CHRNB2	NM_000748
CNTNAP2	NM_014141
CRH	NM_000756
DEPDC5	NM_001242896
FLNA	NM_001456
GRIN2A	NM_000833
KCNT1	NM_020822
LGI1	NM_005097
NPRL2	NM_006545
*NPRL3**	NM_001077350
PRRT2	NM_145239
SCN1A	NM_001165963
SCN1B	NM_001037
SRPX2	NM_014467

*Reference transcript based on build GRCh38 (hg38) for the NPRL3 gene.

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes. Additionally, NGS is used to test for the presence of large deletions and/or duplications.

There are regions of the *SCN1B* gene that cannot be effectively analyzed for the presence of large deletions and/or duplications as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

PCR and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Febrile Seizure Panel	
Gene	**GenBank Accession Number**
CPA6	NM_020361
GABRD	NM_000815
GABRG2	NM_000816
PCDH19	NM_001184880
SCN1A	NM_001165963
SCN1B	NM_001037
SCN2A	NM_021007
SCN9A	NM_002977
STX1B	NM_052874

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes. Additionally, NGS is used to test for the presence of large deletions and/or duplications.

There are regions in *GABRD* and *SCN1B* that cannot be effectively analyzed for the presence of large deletions and/or duplications as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

PCR and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Epilepsy with Migraine Panel	
Gene	**GenBank Accession Number**
ATP1A2	NM_000702
CACNA1A	NM_001127221
NOTCH3	NM_000435
POLG	NM_002693
PRRT2	NM_145239
SCN1A	NM_001165963
SLC2A1	NM_006516

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes. Additionally, NGS is used to test for the presence of large deletions and/or duplications.

There are regions in *NOTCH3* that cannot be effectively analyzed for the presence of large deletions and/or duplications as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

PCR and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Tuberous Sclerosis Panel	
Gene	**GenBank Accession Number**
TSC1	NM_000368
TSC2	NM_000548

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes. Additionally, NGS is used to test for the presence of large deletions and/or duplications.

PCR and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)