

Full name:	Epilepsy, seizures (196 genes) - IPG
Version number:	Version 3
Laboratory:	<u>Centre de Génétique-Institut de Pathologie et de Génétique (IPG)</u>
Created:	04 Jul 2019 - 09:32
Changed:	15 Sep 2023 - 09:45

Related Diseases

- Adenylosuccinate lyase deficiency
- Baraitser-Winter cerebrofrontofacial syndrome
- Fragile X syndrome
- Non-specific early-onset epileptic encephalopathy

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>AARS1</u>	100.00	1	NM_001605.3
<u>ACTB</u>	100.00	1	NM_001101.5
<u>ADSL</u>	100.00	1	NM_000026.4
<u>AKT3</u>	100.00	1	NM_005465.7
<u>ALDH7A1</u>	100.00	1	NM_001182.5
<u>ALG13</u>	98.77	1	NM_001099922.3
<u>ANKRD11</u>	99.99	1	NM_013275.6
<u>ARHGEF9</u>	100.00	1	NM_001353921.2

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<u>ARID1B</u>	98.38	1	NM_001374828.1
<u>ARX</u>	96.96	1	NM_139058.3
<u>ASXL3</u>	100.00	1	NM_030632.3
<u>ATN1</u>	100.00	1	NM_001940.4
<u>ATP1A2</u>	100.00	1	NM_000702.4
<u>ATP1A3</u>	100.00	1	NM_152296.5
<u>ATP7A</u>	100.00	1	NM_000052.7
<u>ATRX</u>	100.00	1	NM_000489.5
<u>ATXN2</u>	100.00	1	NM_001372574.1
<u>BRAT1</u>	100.00	1	NM_152743.4
<u>BTD</u>	100.00	1	NM_001370658.1
<u>CACNA1A</u>	100.00	1	NM_023035.3
<u>CACNA1D</u>	100.00	1	NM_001128840.3
<u>CACNA1E</u>	100.00	1	NM_001205293.3
<u>CACNA2D2</u>	100.00	1	NM_001174051.3
<u>CAD</u>	100.00	1	NM_004341.5
<u>CAMK2A</u>	100.00	1	NM_015981.4
<u>CAMK2B</u>	100.00	1	NM_001220.5
<u>CASK</u>	100.00	1	NM_003688.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CDKL5</u>	100.00	1	NM_001323289.2
<u>CERT1</u>	100.00	1	NM_001130105.1
<u>CHD2</u>	100.00	1	NM_001271.4
<u>CHRNA2</u>	100.00	1	NM_000742.4
<u>CHRNA4</u>	100.00	1	NM_000744.6
<u>CHRNA2</u>	100.00	1	NM_000748.3
<u>CLN3</u>	100.00	1	NM_001042432.2
<u>CLN5</u>	100.00	1	NM_006493.4
<u>CLN6</u>	100.00	1	NM_017882.3
<u>CLTC</u>	100.00	1	NM_004859.4
<u>CNTNAP2</u>	100.00	1	NM_014141.6
<u>COL4A1</u>	100.00	1	NM_001845.6
<u>CSNK2B</u>	100.00	1	NM_001320.7
<u>CSTB</u>	100.00	1	NM_000100.4
<u>CTSA</u>	100.00	1	NM_000308.4
<u>CTSD</u>	100.00	1	NM_001909.5
<u>CTSF</u>	100.00	1	NM_003793.4
<u>CUL4B</u>	100.00	1	NM_001079872.2
<u>CYFIP2</u>	100.00	1	NM_001037333.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>DCX</u>	100.00	1	NM_001195553.2
<u>DDC</u>	100.00	1	NM_001082971.2
<u>DDX3X</u>	100.00	1	NM_001356.4
<u>DEAF1</u>	100.00	1	NM_021008.4
<u>DEPDC5</u>	100.00	1	NM_001242896.3
<u>DHDDS</u>	100.00	1	NM_205861.3
<u>DNAJC5</u>	100.00	1	NM_025219.3
<u>DNM1</u>	100.00	1	NM_004408.4
<u>DOCK7</u>	100.00	1	NM_001367561.1
<u>DYNC1H1</u>	100.00	1	NM_001376.5
<u>DYRK1A</u>	100.00	1	NM_001347721.2
<u>EEF1A2</u>	100.00	1	NM_001958.5
<u>EHMT1</u>	100.00	1	NM_024757.5
<u>EPM2A</u>	100.00	1	NM_005670.4
<u>FGF12</u>	100.00	1	NM_021032.4
<u>FLNA</u>	100.00	1	NM_001110556.2
<u>FMR1</u>	100.00	1	NM_002024.6
<u>FOLR1</u>	100.00	1	NM_016729.3
<u>FOXG1</u>	99.86	1	NM_005249.5

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<u>FOXP1</u>	100.00	1	NM_032682.6
<u>GABBR2</u>	100.00	1	NM_005458.8
<u>GABRA1</u>	100.00	1	NM_001127644.2
<u>GABRA2</u>	100.00	1	NM_001330690.1
<u>GABRA3</u>	100.00	1	NM_000808.4
<u>GABRA5</u>	100.00	1	NM_000810.4
<u>GABRB1</u>	100.00	1	NM_000812.4
<u>GABRB2</u>	100.00	1	NM_001371727.1
<u>GABRB3</u>	100.00	1	NM_000814.6
<u>GABRD</u>	100.00	1	NM_000815.5
<u>GABRG2</u>	92.05	1	NM_198903.2
<u>GAD1</u>	100.00	1	NM_000817.3
<u>GAD2</u>	100.00	1	NM_001134366.2
<u>GAMT</u>	100.00	1	NM_000156.6
<u>GATM</u>	100.00	1	NM_001482.3
<u>GBA1</u>	98.97	1	NM_000157.4
<u>GLDC</u>	100.00	1	NM_000170.3
<u>GLI3</u>	100.00	1	NM_000168.6
<u>GNAO1</u>	100.00	1	NM_020988.3

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<u>GNB1</u>	100.00	1	NM_002074.5
<u>GOSR2</u>	100.00	1	NM_004287.5
<u>GPAA1</u>	100.00	1	NM_003801.4
<u>GRIA3</u>	100.00	1	NM_007325.5
<u>GRIN1</u>	100.00	1	NM_007327.4
<u>GRIN2A</u>	100.00	1	NM_001134407.3
<u>GRIN2B</u>	100.00	1	XM_011520629.2
<u>GRIN2D</u>	100.00	1	NM_000836.2
<u>H3-3A</u>	100.00	1	NM_002107.7
<u>H3-3B</u>	100.00	1	NM_005324.5
<u>HECW2</u>	100.00	1	NM_001348768.2
<u>HIVEP2</u>	100.00	1	NM_006734.4
<u>HLCS</u>	100.00	1	NM_000411.8
<u>HNRNPU</u>	100.00	1	NM_031844.3
<u>HUWE1</u>	100.00	1	NM_031407.7
<u>IQSEC2</u>	99.37	1	NM_001111125.3
<u>JMJD1C</u>	100.00	1	NM_032776.3
<u>KANSL1</u>	100.00	1	NM_001193466.2
<u>KCNA1</u>	100.00	1	NM_000217.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>KCNA2</u>	100.00	1	NM_004974.4
<u>KCNB1</u>	100.00	1	NM_004975.4
<u>KCNC1</u>	100.00	1	NM_001112741.2
<u>KCNH1</u>	100.00	1	NM_172362.3
<u>KCNJ10</u>	100.00	1	NM_002241.5
<u>KCNK4</u>	100.00	1	NM_033310.3
<u>KCNQ2</u>	100.00	1	NM_172107.4
<u>KCNQ3</u>	100.00	1	NM_004519.4
<u>KCNT1</u>	100.00	1	NM_020822.3
<u>KCNT2</u>	100.00	1	NM_198503.5
<u>KCTD7</u>	100.00	1	NM_153033.4
<u>KDM5C</u>	100.00	1	NM_004187.5
<u>KDM6A</u>	100.00	1	NM_001291415.2
<u>KIF1A</u>	100.00	1	NM_001244008.1
<u>KMT2D</u>	100.00	1	NM_003482.3
<u>KMT2E</u>	100.00	1	NM_182931.3
<u>KPTN</u>	100.00	1	NM_007059.4
<u>LGI1</u>	100.00	1	NM_005097.4
<u>LRP1</u>	100.00	1	NM_002332.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>MAGI2</u>	100.00	1	NM_012301.4
<u>MBD5</u>	100.00	1	NM_018328.4
<u>MECP2</u>	100.00	1	NM_001110792.2+NM_004992.3
<u>MED12</u>	100.00	1	NM_005120.3
<u>MED13L</u>	100.00	1	NM_015335.4
<u>MEF2C</u>	100.00	1	NM_002397.5
<u>MFSD8</u>	100.00	1	NM_001371596.2
<u>MICAL1</u>	100.00	1	NM_022765.4
<u>MID1</u>	100.00	1	NM_000381.4
<u>MOCS1</u>	100.00	1	NM_001358530.2
<u>MTHFR</u>	100.00	1	NM_005957.5
<u>MTOR</u>	100.00	1	NM_004958.4
<u>NEDD4L</u>	100.00	1	NM_001144967.3
<u>NEU1</u>	100.00	1	NM_000434.4
<u>NEXMIF</u>	100.00	1	NM_001008537.3
<u>NF1</u>	100.00	1	NM_001042492.3
<u>NFIA</u>	99.78	1	NM_001134673.4
<u>NHLRC1</u>	100.00	1	NM_198586.3
<u>NLGN3</u>	100.00	1	NM_181303.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>NPRL2</u>	100.00	1	NM_006545.5
<u>NPRL3</u>	100.00	1	NM_001077350.3
<u>NRXN1</u>	100.00	1	NM_001330078.2
<u>NTRK2</u>	100.00	1	XM_017014751.2
<u>NUS1</u>	100.00	1	NM_138459.5
<u>OPHN1</u>	100.00	1	NM_002547.3
<u>PACS1</u>	100.00	1	NM_018026.4
<u>PACS2</u>	100.00	1	NM_001100913.3
<u>PAFAH1B1</u>	100.00	1	NM_000430.4
<u>PANK2</u>	100.00	1	NM_153638.3
<u>PCDH19</u>	100.00	1	NM_001184880.2
<u>PHGDH</u>	100.00	1	NM_006623.4
<u>PIGA</u>	100.00	1	NM_002641.3
<u>PIGO</u>	100.00	1	NM_032634.4
<u>PIGQ</u>	100.00	1	NM_004204.5
<u>PIGS</u>	100.00	1	NM_033198.4
<u>PIGT</u>	100.00	1	NM_015937.6
<u>PIGU</u>	100.00	1	NM_080476.4
<u>PIGV</u>	100.00	1	NM_017837.4

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<u>PIK3CA</u>	100.00	1	NM_006218.4
<u>PIK3R2</u>	100.00	1	NM_005027.4
<u>PLCB1</u>	100.00	1	NM_015192.4
<u>PLPBP</u>	100.00	1	NM_007198.4
<u>PLXNA1</u>	100.00	1	NM_032242.3
<u>PNKP</u>	100.00	1	NM_007254.4
<u>PNPO</u>	100.00	1	NM_018129.4
<u>POLG</u>	100.00	1	NM_001126131.2
<u>PPP2R1A</u>	100.00	1	NM_014225.6
<u>PPP2R5D</u>	100.00	1	NM_006245.4
<u>PPT1</u>	100.00	1	NM_000310.4
<u>PRICKLE1</u>	100.00	1	NM_153026.3
<u>PRIMA1</u>	100.00	1	NM_178013.4
<u>PRRT2</u>	100.00	1	NM_145239.3
<u>PSAT1</u>	100.00	1	NM_058179.4
<u>PTEN</u>	100.00	1	NM_000314.8
<u>PURA</u>	100.00	1	NM_005859.5
<u>QARS1</u>	100.00	1	NM_005051.3
<u>RELN</u>	100.00	1	NM_005045.4

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<u>RLIM</u>	100.00	1	NM_016120.4
<u>SCAF4</u>	100.00	1	NM_020706.2
<u>SCARB2</u>	100.00	1	NM_005506.4
<u>SCN1A</u>	100.00	1	NM_001165963.3
<u>SCN1B</u>	100.00	1	NM_001037.5+NM_199037.5
<u>SCN2A</u>	100.00	1	NM_001040142.2
<u>SCN3A</u>	100.00	1	NM_006922.4
<u>SCN8A</u>	100.00	1	NM_001330260.2
<u>SCN9A</u>	100.00	1	NM_001365536.1
<u>SERPINI1</u>	100.00	1	NM_001122752.1
<u>SETBP1</u>	99.99	1	NM_015559.3
<u>SETD5</u>	100.00	1	NM_001080517.3
<u>SLC12A5</u>	100.00	1	NM_020708.5
<u>SLC19A3</u>	100.00	1	NM_025243.4
<u>SLC1A2</u>	100.00	1	NM_004171.4
<u>SLC25A22</u>	100.00	1	NM_001191061.2
<u>SLC2A1</u>	100.00	1	NM_006516.3
<u>SLC35A2</u>	100.00	1	NM_005660.3
<u>SLC6A1</u>	100.00	1	NM_003042.4

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<u>SLC6A11</u>	100.00	1	NM_014229.3
<u>SLC6A8</u>	100.00	1	NM_005629.4
<u>SLC9A6</u>	100.00	1	NM_001042537.1
<u>SMARCA2</u>	100.00	1	NM_003070.5
<u>SMC1A</u>	100.00	1	NM_006306.4
<u>SMS</u>	100.00	1	NM_004595.5
<u>SNAP25</u>	100.00	1	NM_130811.4
<u>SPTAN1</u>	100.00	1	NM_001130438.3
<u>ST3GAL3</u>	100.00	1	NM_006279.5
<u>STX1B</u>	100.00	1	NM_052874.5
<u>STXBP1</u>	100.00	1	NM_001032221.6
<u>SYN1</u>	100.00	1	NM_006950.3
<u>SYNGAP1</u>	100.00	1	NM_006772.3
<u>SZT2</u>	100.00	1	NM_001365999.1
<u>TBC1D24</u>	100.00	1	NM_001199107.2
<u>TBL1XR1</u>	100.00	1	NM_024665.7
<u>TCF4</u>	100.00	1	NM_001083962.2
<u>TNK2</u>	100.00	1	XM_005269270.3
<u>TPP1</u>	100.00	1	NM_000391.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>TRPM6</u>	100.00	1	NM_017662.5
<u>TRRAP</u>	100.00	1	NM_001244580.1
<u>TSC1</u>	100.00	1	NM_000368.5
<u>TSC2</u>	100.00	1	NM_000548.5
<u>TUBA1A</u>	100.00	1	NM_006009.4
<u>TWINK</u>	100.00	1	NM_021830.5
<u>UBE3A</u>	100.00	1	NM_130839.5
<u>UGP2</u>	100.00	1	NM_006759.3
<u>WASF1</u>	100.00	1	NM_003931.3
<u>WDR45</u>	100.00	1	NM_001029896.2
<u>WWOX</u>	100.00	1	NM_016373.4
<u>ZEB2</u>	100.00	1	NM_014795.4