



## Liste des gènes analysés dans le cadre du Panel épilepsie (PAGEM)

Symbole du gène	Nom du gène	Chromosome	Transmission
<b>ADSL</b>	Adenylosuccinate lyase	chr22	AR
<b>ALDH4A1</b>	Aldehyde dehydrogenase 4 family, member A1	chr1	AR
<b>ALDH7A1</b>	Aldehyde dehydrogenase 7 family, member A1	chr5	AR
<b>ALG13</b>	Alg13, S. cerevisiae, homolog of	chrX	XLD
<b>AP3B2</b>	Adaptor-related protein complex 3, beta-2 subunit	chr15	AR
<b>ARHGEF9</b>	Rho guanine nucleotide exchange factor 9	chrX	XLR
<b>ARX</b>	Aristaless-related homeobox, X-linked	chrX	XLR
<b>ATIC</b>	AICAR formyltransferase / IMP cyclohydrolase	chr2	AR
<b>ATP1A3</b>	ATPase, Na+K+ transporting, alpha-3 subunit	chr19	AD
<b>BRAT1</b>	BRCA1-associated ATM activator 1	chr7	AR
<b>BTBD</b>	Biotinidase	chr3	AR
<b>CACNA1A</b>	Calcium channel, voltage-dependent, P/Q type, alpha-1A subunit	chr19	AD
<b>CAD</b>	CAD trifunctional protein of pyrimidine biosynthesis	chr2	AR
<b>CASK</b>	Calcium/calmodulin-dependent serine protein kinase	chrX	XLD
<b>CDKL5</b>	Cyclin-dependent kinase-like 5	chrX	XLD
<b>CHD2</b>	Chromodomain helicase DNA binding protein 2	chr15	AD
<b>CHRNA2</b>	Cholinergic receptor, nicotinic, alpha-2 subunit	chr8	AD
<b>CHRNA4</b>	Cholinergic receptor, nicotinic, alpha-4 subunit	chr20	AD
<b>CHRNB2</b>	Cholinergic receptor, nicotinic, beta-2 subunit	chr1	.
<b>CPA6</b>	Carboxypeptidase A6	chr8	AD/AR
<b>CSTB</b>	Cystatin B	chr21	AR
<b>DEPDC5</b>	DEP domain-containing protein 5	chr22	AD
<b>DNM1</b>	Dynamin 1	chr9	AD
<b>DYRK1A</b>	Dual specificity tyrosine-phosphorylation regulated kinase 1A	chr21	AD
<b>EEF1A2</b>	Eukaryotic translation elongation factor-1, alpha-2 subunit	chr20	AD
<b>EPM2A</b>	Laforin	chr6	AR
<b>FARS2</b>	Phenylalanyl-tRNA synthetase 2, mitochondrial	chr6	AR
<b>FGF12</b>	Fibroblast growth factor 12	chr3	AD
<b>FOLR1</b>	Folate receptor-1, adult	chr11	AR
<b>FOXP1</b>	Forkhead box G1B	chr14	AD
<b>FRRS1L</b>	Ferric-chelate reductase 1-like	chr9	AR
<b>GABBR2</b>	Gamma-aminobutyric acid B receptor 2	chr9	AD
<b>GABRA1</b>	Gamma-aminobutyric acid (GABA) A receptor, alpha-1 subunit	chr5	AD
<b>GABRB1</b>	Gamma-aminobutyric acid (GABA) A receptor, beta-1 subunit	chr4	AD
<b>GABRB2</b>	Gamma-aminobutyric acid (GABA) A receptor, beta-2 subunit	chr5	AD
<b>GABRB3</b>	Gamma-aminobutyric acid (GABA) A receptor, beta-3 subunit	chr15	AD
<b>GABRG2</b>	Gamma-aminobutyric acid (GABA) A receptor, gamma-2 subunit	chr5	AD
<b>GNAO1</b>	Guanine nucleotide-binding protein (G protein), alpha-activating activity	chr16	AD
<b>GOSR2</b>	Golgi snap receptor complex member 2	chr17	AR
<b>GPHN</b>	Gephyrin	chr14	AR
<b>GRIN1</b>	Glutamate receptor, ionotropic, N-methyl D-aspartate 1	chr9	AD/AR
<b>GRIN2A</b>	Glutamate receptor, ionotropic, N-methyl D-aspartate 2A	chr16	AD
<b>GRIN2B</b>	Glutamate receptor, ionotropic, N-methyl D-aspartate 2B	chr12	AD
<b>GRIN2D</b>	Glutamate receptor, ionotropic, N-methyl-D-aspartate 2D	chr19	AD
<b>HCN1</b>	Hyperpolarization-activated cyclic nucleotide-gated potassium channel 1	chr5	AD
<b>HDAC4</b>	Histone deacetylase 4	chr2	.
<b>HNRNPU</b>	Heterogeneous nuclear ribonucleoprotein U	chr1	AD
<b>IQSEC2</b>	IQ motif- and Sec7 domain-containing protein 2	chrX	XLD
<b>KCNA2</b>	Potassium channel, voltage-gated, Shaker-related subfamily, member 2	chr1	AD
<b>KCNB1</b>	Potassium voltage-gated channel, Shab-related subfamily, member 1	chr20	AD
<b>KCNC1</b>	Potassium voltage-gated channel, Shaw-related subfamily, member 1	chr11	AD
<b>KCNH1</b>	Potassium voltage-gated channel, subfamily H, member 1	chr1	AD
<b>KCNJ10</b>	Potassium inwardly-rectifying channel, subfamily J, member 10	chr1	AR
<b>KCNQ2</b>	Potassium voltage-gated channel, KQT-like subfamily, member 2	chr20	AD
<b>KCNQ3</b>	Potassium voltage-gated channel, KQT-like subfamily, member 3	chr8	AD
<b>KCNT1</b>	Potassium channel, subfamily T, member 1	chr9	AD
<b>KCTD7</b>	Potassium channel tetramerization domain containing 7	chr7	AR
<b>LG1</b>	Leucine-rich gene, glioma-inactivated, 1	chr10	AD

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Symbole du gène	Nom du gène	Chromosome	Transmission
	<i>(début de la liste sur la Page 1/2)</i>		
<b>MBD5</b>	Methyl-CpG-binding domain protein 5	chr2	AD
<b>MECP2</b>	Methyl-CpG-binding protein 2	chrX	XLD/XLR
<b>MEF2C</b>	MADS box transcription enhancer factor 2, polypeptide C	chr5	AD
<b>MOCS1</b>	Molybdenum cofactor synthesis 1	chr6	AR
<b>MOCS2</b>	Molybdenum cofactor synthesis 2	chr5	AR
<b>MTOR</b>	Mechanistic target of rapamycin	chr1	AD
<b>NEDD4L</b>	Ubiquitin protein ligase NEDD4-like	chr18	AD
<b>NHLRC1</b>	NHL repeat containing E3 ubiquitin protein ligase 1	chr6	AR
<b>NPRL2</b>	Npr2, <i>S. cerevisiae</i> , homolog of	chr3	AD
<b>NPRL3</b>	Nitrogen permease regulator-like 3	chr16	AD
<b>NR2F1</b>	Nuclear receptor subfamily 2, group F, member 1	chr5	AD
<b>PACS2</b>	Phosphofurin acidic cluster sorting protein 2	chr14	AD
<b>PCDH19</b>	Protocadherin 19	chrX	XL
<b>PIGA</b>	Phosphatidylinositol glycan anchor biosynthesis, class A protein	chrX	XLR
<b>PIGG</b>	Phosphatidylinositol glycan anchor biosynthesis, class G protein	chr4	AR
<b>PIGN</b>	Phosphatidylinositol glycan anchor biosynthesis, class N protein	chr18	AR
<b>PIGO</b>	Phosphatidylinositol glycan anchor biosynthesis, class O protein	chr9	AR
<b>PIGQ</b>	Phosphatidylinositol glycan anchor biosynthesis class Q protein	chr16	.
<b>PLCB1</b>	Phospholipase C, beta-1	chr20	AR
<b>PNKP</b>	Polynucleotide kinase 3' phosphatase	chr19	AR
<b>PNPO</b>	Pyridoxamine 5'-phosphate oxidase	chr17	AR
<b>POLG</b>	DNA polymerase gamma, catalytic subunit	chr15	AD/AR
<b>PRDM8</b>	PR/SET domain 8	chr4	AR
<b>PRICKLE1</b>	Prickle-like 1	chr12	AR
<b>PRRT2</b>	Proline-rich transmembrane protein 2	chr16	AD
<b>QARS</b>	Glutamyl-tRNA synthetase	chr3	AR
<b>RORB</b>	RAR-related orphan receptor B	chr9	.
<b>SCARB2</b>	Scavenger receptor class B, member 2	chr4	AR
<b>SCN1A</b>	Sodium channel, voltage-gated, type I, alpha subunit	chr2	AD
<b>SCN1B</b>	Sodium channel, voltage-gated, type I, beta subunit	chr19	AD/AR
<b>SCN2A</b>	Sodium channel, voltage-gated, type II, alpha subunit	chr2	AD
<b>SCN8A</b>	Sodium channel, voltage gated, type VIII, alpha subunit	chr12	AD
<b>SIK1</b>	Salt-inducible kinase 1	chr21	AD
<b>SLC12A5</b>	Solute carrier family 12 (potassium-chloride transporter), member 5	chr20	AD/AR
<b>SLC13A5</b>	Solute carrier family 13 (sodium-dependent citrate transporter), member 5	chr17	AR
<b>SLC19A3</b>	Solute carrier family 19 (folate transporter), member 3	chr2	AR
<b>SLC1A2</b>	Solute carrier family 1 (glial high affinity glutamate transporter), member 2	chr11	AD
<b>SLC25A22</b>	Solute carrier family 25 (mitochondrial carrier, glutamate), member 22	chr11	AR
<b>SLC2A1</b>	Solute carrier family 2 (facilitated glucose transporter), member 1	chr1	AD/AR
<b>SLC35A2</b>	Solute carrier family 35 (UDP-galactose transporter), member 2	chrX	XLD
<b>SLC6A1</b>	Solute carrier family 6 (neurotransmitter transporter, GABA), member 1	chr3	AD
<b>SLC9A6</b>	Solute carrier family 9 (sodium/hydrogen exchanger), member 6	chrX	XLD
<b>SPTAN1</b>	Spectrin, alpha, nonerythrocytic 1	chr9	AD
<b>ST3GAL3</b>	ST3 beta-galactoside alpha-2,3-sialyltransferase 3	chr1	AR
<b>ST3GAL5</b>	ST3 beta-galactoside alpha-2,3-sialyltransferase 5	chr2	.
<b>STAMBP</b>	STAM binding protein	chr2	AR
<b>STX1B</b>	Syntaxin 1B	chr16	AD
<b>STXBP1</b>	Syntaxin-binding protein 1	chr9	AD
<b>SYN1</b>	Synapsin 1	chrX	XLD/XLR
<b>SYNGAP1</b>	Synaptic Ras GTPase activating protein 1	chr6	AD
<b>SYNJ1</b>	Synaptojanin 1	chr21	AR
<b>SZT2</b>	Seizure threshold 2, mouse, homolog of	chr1	AR
<b>TBC1D24</b>	TBC1 domain family, member 24	chr16	AD/AR
<b>TCF4</b>	Transcription factor 4	chr18	AD
<b>UBA5</b>	Ubiquitin-like modifier activating enzyme 5	chr3	AR
<b>UBE3A</b>	Ubiquitin protein ligase E3A	chr15	.
<b>WDR45</b>	WD repeat-containing protein 45	chrX	XLD
<b>WWOX</b>	WW domain-containing oxidoreductase	chr16	AR