

Panel de 245 gènes impliqués dans les malformations du développement cortical et les épilepsies

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<u>non du gène</u>	<u>refseq</u>	<u>description</u>
ACTB	XM_005249819	actin, beta
ACTG1	NM_001199954	actin, gamma 1
ADRA2B	NM_000682	adrenoceptor alpha 2B
ADSL	NM_000026	adenylosuccinate lyase
AHI1	NM_001134831	Abelson helper integration site 1
AKT3	NM_181690	v-akt murine thymoma viral oncogene homolog 3
ALDH4A1	NM_170726	aldehyde dehydrogenase 4 family, member A1
ALDH7A1	XM_005271994	aldehyde dehydrogenase 7 family, member A1
ALG13	NM_001257231	ALG13, UDP-N-acetylglucosaminyltransferase subunit
ALG3	NM_005787	ALG3, alpha-1,3- mannosyltransferase
AP3B2	NM_001278511	adaptor-related protein complex 3, beta 2 subunit
ARFGEF2	XM_005260253	ADP-ribosylation factor guanine nucleotide-exchange factor 2 (brefeldin A-inhibited)
ARHGEF9	XM_005278084	Cdc42 guanine nucleotide exchange factor (GEF) 9
ARX	NM_139058	aristaless related homeobox
ASNS	NM_133436	asparagine synthetase (glutamine-hydrolyzing)
ASPM	NM_018136	asp (abnormal spindle) homolog, microcephaly associated (Drosophila)
ATIC	NM_004044	5-aminoimidazole-4-carboxamide ribonucleotide formyltransferase/IMP cyclohydrolase
ATP1A2	XM_005245210	ATPase, Na ⁺ /K ⁺ transporting, alpha 2 polypeptide
ATP1A3	XM_005278390	ATPase, Na ⁺ /K ⁺ transporting, alpha 3 polypeptide
ATP6V0A2	NM_012463	ATPase, H ⁺ transporting, lysosomal V0 subunit a2
ATP6V1A	XM_005247528	ATPase, H ⁺ transporting, lysosomal 70kDa, V1 subunit A
ATRX	XM_005262158	alpha thalassemia/mental retardation syndrome X-linked
B3GALNT2	NM_152490	beta-1,3-N-acetylgalactosaminyltransferase 2
B3GALNT2	NM_001277155	beta-1,3-N-acetylgalactosaminyltransferase 2
B3GNT1	NM_006876	UDP-GlcNAc:betaGal beta-1,3-N-acetylglucosaminyltransferase 1
BRAT1	NM_152743	BRCA1-associated ATM activator 1
BTD	NM_000060	biotinidase
CACNA1A	XM_005260068	calcium channel, voltage-dependent, P/Q type, alpha 1A subunit
CAD	NM_004341	carbamoyl-phosphate synthetase 2, aspartate transcarbamylase, and dihydroorotase
CASC5	NM_170589	cancer susceptibility candidate 5
CASK	NM_003688	calcium/calmodulin-dependent serine protein kinase (MAGUK family)
CCND2	NM_001759	cyclin D2
CDK5RAP2	NM_018249	CDK5 regulatory subunit associated protein 2
CDK6	NM_001259	cyclin-dependent kinase 6
CDKL5	NM_003159	cyclin dependent kinase like 5
CENPJ	NM_018451	centromere proteinJ
CEP135	XM_005265788	centrosomal protein 135kDa
CEP152	NM_001194998	centrosomal protein 152kDa
CEP63	NM_025180	centrosomal protein 63kDa
CHD2	XM_005254837	chromodomain helicase DNA binding protein 2
CHMP1A	NM_002768	charged multivesicular body protein 1A
CHRNA2	XM_005273397	cholinergic receptor, nicotinic, alpha 2 (neuronal)
CHRNA4	XM_005260191	cholinergic receptor, nicotinic, alpha 4 (neuronal)
CHRNB2	NM_000748	cholinergic receptor, nicotinic, beta 2 (neuronal)
CNTN2	XM_005245467; NM_005076	contactin 2 (axonal)
CNTNAP1	XM_005257748	contactin associated protein 1
CNTNAP2	NM_014141	contactin associated protein-like 2
COL18A1	XM_005261178	collagen, type XVIII, alpha 1
COL4A1	NM_001845	collagen, type IV, alpha 1

CPA6	NM_020361	carboxypeptidase A6
CRADD	NM_003805	CASP2 and RIPK1 domain containing adaptor with death domain
CSTB	NM_000100	cystatin B (stefin B)
CUL4B	NM_003588	cullin 4B
DCHS1	XM_005253207	dachsous cadherin-related 1
DCX	NM_000555	doublecortin
DDB1	XM_005273804; NM_001923	damage-specific DNA binding protein 1, 127kDa
DDB2	NM_000107	damage-specific DNA binding protein 2, 48kDa
DDX3X	XM_005272592	DEAD (Asp-Glu-Ala-Asp) box helicase 3, X-linked
DENND5A	XM_005252831	DENN/MADD domain containing 5A
DEPDC5	XM_005261866	DEP domain containing 5
DNM1	XM_005251769;NM_0 01005336	dynamamin 1
DOCK7	XM_005271292	dedicator of cytokinesis 7
DYNC1H1	NM_001376	dynein, cytoplasmic 1, heavy chain 1
DYRK1A	NM_001396	dual-specificity tyrosine-(Y)-phosphorylation regulated kinase 1A
EEF1A2	NM_001958	eukaryotic translation elongation factor 1 alpha 2
ELL	NM_006532	elongation factor RNA polymerase II
EML1	NM_001008707	echinoderm microtubule associated protein like 1
EPM2A	NM_005670	epilepsy, progressive myoclonus type 2A, Lafora disease (laforin)
ERCC1	NM_001983	excision repair cross-complementing rodent repair deficiency, complementation group 1 (includes overlapping antisense sequence)
ERCC2	XM_005258641	excision repair cross-complementing rodent repair deficiency, complementation group 2
ERCC3	NM_000122	excision repair cross-complementing rodent repair deficiency, complementation group 3
ERCC4	NM_005236	excision repair cross-complementing rodent repair deficiency, complementation group 4
ERCC5	NM_000123	excision repair cross-complementing rodent repair deficiency, complementation group 5
ERCC6	XM_005269595;XM_0 05269596;NM_000124	excision repair cross-complementing rodent repair deficiency, complementation group 6
ERCC8	NM_000082	excision repair cross-complementing rodent repair deficiency, complementation group 8
ERMARD	NM_001278531	ER membrane-associated RNA degradation
EXOSC3	NM_016042	exosome component 3
FARS2	XM_005248812	phenylalanyl-tRNA synthetase 2, mitochondrial
FAT4	XM_005263210	FAT atypical cadherin 4
FKRP	XM_005259253	fukutin related protein
FKTN	NM_001198963	fukutin
FLNA	NM_001110556	filamin A, alpha
FOLR1	NM_016725	folate receptor 1 (adult)
FOXP1	NM_005249	forkhead box G1
FRRS1L	NM_014334	ferric-chelate reductase 1-like
GABBR2	XM_005252316	gamma-aminobutyric acid (GABA) B receptor, 2
GABRA1	NM_000806	gamma-aminobutyric acid (GABA) A receptor, alpha 1
GABRB2	NM_000813	gamma-aminobutyric acid (GABA) A receptor, beta 2
GABRB3	NM_000814	gamma-aminobutyric acid (GABA) A receptor, beta 3
GABRG2	NM_198903	gamma-aminobutyric acid (GABA) A receptor, gamma 2
GNAO1	NM_020988	guanine nucleotide binding protein (G protein), alpha activating activity polypeptide O
GOSR2	NM_054022	golgi SNAP receptor complex member 2
GPHN	NM_001024218	gephyrin
GPR56	XM_005256252	G protein-coupled receptor 56
GRIN1	NM_001185090	glutamate receptor, ionotropic, N-methyl D-aspartate 1
GRIN2A	NM_001134407	glutamate receptor, ionotropic, N-methyl D-aspartate 2A
GRIN2B	XM_005253351	glutamate receptor, ionotropic, N-methyl D-aspartate 2B
GTF2E2	NM_002095	general transcription factor IIE, polypeptide 2, beta 34kDa
GTF2H5	NM_207118	general transcription factor IIH, polypeptide 5
HCN1	NM_021072	hyperpolarization activated cyclic nucleotide-gated potassium channel 1
HCN2	NM_001194	hyperpolarization activated cyclic nucleotide-gated potassium channel 2
HDAC4	XM_005264078	histone deacetylase 4

HNRNPU	NM_004501	heterogeneous nuclear ribonucleoprotein U (scaffold attachment factor A)
IQSEC2	NM_001111125	IQ motif and Sec7 domain 2
KATNB1	NM_005886	katanin p80 (WD repeat containing) subunit B 1
KCNB1	NM_004975	potassium voltage-gated channel, Shab-related subfamily, member 1
KCNC1	NM_001112741	potassium voltage-gated channel, Shaw-related subfamily, member 1
KCNH1	NM_172362	potassium voltage-gated channel, subfamily H (eag-related), member 1
KCNJ10	NM_002241	potassium inwardly-rectifying channel, subfamily J, member 10
KCNQ2	NM_172107	potassium voltage-gated channel, KQT-like subfamily, member 2
KCNQ3	XM_005250913	potassium voltage-gated channel, KQT-like subfamily, member 3
KCNT1	NM_020822	potassium channel, subfamily T, member 1
KCTD7	NM_153033	potassium channel tetramerization domain containing 7
KIAA1109	NM_015312	KIAA1109
KIAA1279	NM_015634	KIAA1279
KIF21B	NM_001252100	kinesin family member 21B
KIF2A	NM_001098511	kinesin heavy chain member 2A
KIF5C		kinesin family member 5C
LAMA1	NM_005559	laminin, alpha 1
LAMB1	NM_002291	laminin, beta 1
LAMC3	NM_006059	laminin, gamma 3
LARGE	XM_005261831	like-glycosyltransferase
LG1	NM_005097	leucine-rich, glioma inactivated 1
MAP1B	XM_005248507	microtubule-associated protein 1B
MAST1	XM_005259820	microtubule associated serine/threonine kinase 1
MBD5	XM_005263713;NM_018328	methyl-CpG binding domain protein 5
MCPH1	NM_024596	microcephalin 1
MECP2	XM_005277853	methyl CpG binding protein 2 (Rett syndrome)
MED12		mediator complex subunit 12
MED17	NM_004268	mediator complex subunit 17
MEF2C	XM_005248509;XM_005248508;XM_005248510;NM_001193347	myocyte enhancer factor 2C
MOCS2	NM_176806	molybdenum cofactor synthesis 2
MPLKIP	NM_138701	M-phase specific PLK1 interacting protein
MTOR	XM_005263441	mechanistic target of rapamycin (serine/threonine kinase)
NDE1	NM_001143979	nudE neurodevelopment protein 1
NEDD4L	NM_001144968	neural precursor cell expressed, developmentally down-regulated 4-like, E3 ubiquitin protein ligase
NFIX	NM_001271043	nuclear factor I/X (CCAAT-binding transcription factor)
NHLRC1	NM_198586	NHL repeat containing E3 ubiquitin protein ligase 1
NPRL2	XM_005264808	nitrogen permease regulator-like 2 (S. cerevisiae)
NPRL3	NM_001243247	nitrogen permease regulator-like 3 (S. cerevisiae)
NR2F1	XM_005272071	nuclear receptor subfamily 2, group F, member 1
NSD1	XM_005265961	nuclear receptor binding SET domain protein 1
NSDHL	NM_015922	NAD(P) dependent steroid dehydrogenase-like
OPHN1	NM_002547	oligophrenin 1
PAFAH1B1	NM_000430	platelet-activating factor acetylhydrolase 1b, regulatory subunit 1 (45kDa)
PCDH19	NM_001184880	protocadherin 19
PCNA	NM_002592	proliferating cell nuclear antigen
PDE2A	NM_001146209	phosphodiesterase 2A, cGMP-stimulated
PDHA1	NM_001173454	pyruvate dehydrogenase (lipoamide) alpha 1
PHC1	NM_004426	polyhomeotic homolog 1 (Drosophila)
PHF6	NM_032458	PHD finger protein 6
PIGA	NM_002641	phosphatidylinositol glycan anchor biosynthesis, class A
PIGG	NM_017733	phosphatidylinositol glycan anchor biosynthesis, class G
PIGN	XM_005266675	phosphatidylinositol glycan anchor biosynthesis, class N
PIGO	NM_001201484	phosphatidylinositol glycan anchor biosynthesis, class O

PIK3CA	NM_006218	phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit alpha
PIK3R2	NM_005027	phosphoinositide-3-kinase, regulatory subunit 2 (beta)
PLCB1	XM_005260681	phospholipase C, beta 1 (phosphoinositide-specific)
PNKP	NM_007254	polynucleotide kinase 3'-phosphatase
PNPO	NM_018129	pyridoxamine 5'-phosphate oxidase
POLG	NM_001126131	polymerase (DNA directed), gamma
POLH	XM_005249186	polymerase (DNA directed), eta
POMGNT1	NM_001243766	protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1,2-)
POMGNT2	NM_032806	protein O-linked mannose N-acetylglucosaminyltransferase 2 (beta 1,4-)
POMK	NM_032237	protein-O-mannose kinase
POMT1	XM_005272159	protein-O-mannosyltransferase 1
POMT2	NM_013382	protein-O-mannosyltransferase 2
PRDM8	XM_005263145	PR domain containing 8
PRICKLE1	NM_153026	prickle homolog 1 (Drosophila)
PRRT2	XM_005255054;NM_01256442	proline-rich transmembrane protein 2
PSAT1	XM_005251947	phosphoserine aminotransferase 1
QARS	NM_001272073	glutamyl-tRNA synthetase
RARS2	XM_005248735	arginyl-tRNA synthetase 2, mitochondrial
RELN	NM_005045	reelin
RNF113A	NM_006978	ring finger protein 113A
RNU4ATAC	NR_023343	RNA, U4atac small nuclear (U12-dependent splicing)
ROGDI	NM_024589	rogdi homolog (Drosophila)
RORB	XM_005252129	RAR-related orphan receptor B
RPS6KA3	XM_005274577	ribosomal protein S6 kinase, 90kDa, polypeptide 3
RTTN	NM_173630	rotatin
SASS6	XM_005270551	spindle assembly 6 homolog (C. elegans)
SCARB2	NM_005506	scavenger receptor class B, member 2
SCN1A		sodium channel, voltage-gated, type I, alpha subunit
SCN1B	XM_005259144	sodium channel, voltage-gated, type I, beta subunit
SCN2A	NM_021007	sodium channel, voltage-gated, type II, alpha subunit
SCN8A	NM_014191	sodium channel, voltage gated, type VIII, alpha subunit
SEPSECS	XM_005248167	Sep (O-phosphoserine) tRNA:Sec (selenocysteine) tRNA synthase
SIK1	NM_173354	salt-inducible kinase 1
SLC12A5	XM_005260470	solute carrier family 12 (potassium/chloride transporter), member 5
SLC12A6	NM_133647	solute carrier family 12 (potassium/chloride transporter), member 6
SLC13A5	NM_177550	solute carrier family 13 (sodium-dependent citrate transporter), member 5
SLC19A3	XM_005246874	solute carrier family 19 (thiamine transporter), member 3
SLC1A2	NM_001195728	solute carrier family 1 (glial high affinity glutamate transporter), member 2
SLC25A19	XM_005257561	solute carrier family 25 (mitochondrial thiamine pyrophosphate carrier), member 19
SLC25A22	NM_024698	solute carrier family 25 (mitochondrial carrier: glutamate), member 22
SLC2A1	NM_006516	solute carrier family 2 (facilitated glucose transporter), member 1
SLC35A2	NM_005660	solute carrier family 35 (UDP-galactose transporter), member A2
SLC6A1	XM_005265411;XM_005265410;NM_003042	solute carrier family 6 (neurotransmitter transporter), member 1
SLC9A6	NM_001042537	solute carrier family 9, subfamily A (NHE6, cation proton antiporter 6), member 6
SMARCAL1	XM_005246632	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a-like 1
SON	NM_138927	SON DNA binding protein
SPTAN1	XM_005252145	spectrin, alpha, non-erythrocytic 1
ST3GAL3	NM_174963	ST3 beta-galactoside alpha-2,3-sialyltransferase 3
ST3GAL5	NM_001042437	ST3 beta-galactoside alpha-2,3-sialyltransferase 5
STAG3	XM_005250116	stromal antigen 3
STAMPB	XM_005264090	STAM binding protein
STIL	XM_005271131	SCL/TAL1 interrupting locus
STX1B	NM_052874	syntaxin 1B
STXBP1	NM_003165	syntaxin binding protein 1

SYN1	NM_006950	synapsin I
SYNGAP1	XM_005249459	synaptic Ras GTPase activating protein 1
SYNJ1	XM_005261079	synaptojanin 1
SZT2	XM_005270688	seizure threshold 2 homolog (mouse)
TBC1D23	NM_001199198	TBC1 domain family, member 23
TBC1D24	NM_001199107	TBC1 domain family, member 24
TCF4	NM_001243226	transcription factor 4
TMEM5	NM_014254	transmembrane protein 5
TRAPPC9	NM_031466	trafficking protein particle complex 9
TSC1	XM_005272212	tuberous sclerosis 1
TSC2	XM_005255527	tuberous sclerosis 2
TSEN2	NM_025265	TSEN2 tRNA splicing endonuclease subunit
TSEN34	NM_024075	TSEN34 tRNA splicing endonuclease subunit
TSEN54	NM_207346	TSEN54 tRNA splicing endonuclease subunit
TUBA1A	NM_006009	tubulin, alpha 1a
TUBA8	NM_018943	tubulin, alpha 8
TUBB	NM_178014	tubulin, beta class I
TUBB2A	NM_001069	tubulin, beta 2A class IIa
TUBB2B	NM_178012	tubulin, beta 2B class IIb
TUBB3	NM_006086	Tubulin beta-3 chain
TUBG1	NM_001070	tubulin, gamma 1
TUBGCP4	NM_014444	tubulin, gamma complex associated protein 4
UBE3A	NM_130839	ubiquitin protein ligase E3A
UBE3B	NM_130466	ubiquitin protein ligase E3B
USP7	NM_003470	ubiquitin specific peptidase 7 (herpes virus-associated)
UVSSA	NM_020894	UV-stimulated scaffold protein A
VLDLR	NM_003383	very low density lipoprotein receptor
VRK1	NM_003384	vaccinia related kinase 1
WDR45	NM_007075	WD repeat domain 45
WDR62	NM_001083961	WD repeat domain 62
WDR73	NM_032856	WD repeat domain 73
WDR81	XM_005256454	WD repeat domain 81
WWOX	NM_016373	WW domain containing oxidoreductase
XPA	NM_000380	xeroderma pigmentosum, complementation group A
XPC	NM_004628	xeroderma pigmentosum, complementation group C
ZNF335	XM_005260507	zinc finger protein 335