Hôpitaux Universitaires La Pitié Salpêtrière-Charles Foix
Unité Fonctionnelle de Neurogénétique Moléculaire et Cellulaire
Département de Génétique
Centre de Génétique Moléculaire et Chromosomique **(http://www.cgmc-psl.fr)**
Pôle de Biologie Médicale et Pathologie

Bâtiment 6 rue la Peyronnie
Secteur Pitié
47/83, boulevard de l'Hôpital
75651 PARIS cedex 13

Liste des gènes étudiés en diagnostic dans le panel "épilepsies monogéniques"

(en bleu : gènes étudiés dans le cadre des encéphalopathies épileptiques; en jaune : gènes étudiés dans le cadre des épilepsies familiales; en vert : gènes étudiés dans les 2 cas)

Gène	Locus	Pathologie OMIM	Transcrit de référence (NM)
ADSL	22q13.1	Adenylosuccinase deficiency	NM_000026
ALDH7A1	5q23.2	Epilepsy, pyridoxine-dependent	NM_001182
ALG13	Xq23	Congenital disorder of glycosylation, type Is	NM_001099922
ARHGEF9	Xq11.1-q11.2	Epileptic encephalopathy, early infantile, 8	NM_015185
ARX	Xp21.3	Epileptic encephalopathy, early infantile, 1	NM_139058
ATP1A3	19q13.2	Alternating hemiplegia of childhood 2 / CAPOS syndrome	NM_001256214.1
BTD	3p25.1	Biotinidase deficiency	NM_001281723
CASK	Xp11.4	FG syndrome 4 , Mental retardation and microcephaly with pontine and cerebellar hypoplasia, Mental retardation, with or without nystagmus	NM_003688
CDKL5	Xp22.13	Epileptic encephalopathy, early infantile, 2	NM_003159
CHD2	15q26.1	Epileptic encephalopathy, childhood-onset	NM_001271
CHRNA4	20q13.33	Epilepsy, nocturnal frontal lobe, 1	NM_000744
CHRNB2	1q21.3	Epilepsy, nocturnal frontal lobe, 3	NM_000748
CPA6	8q13.2	Epilepsy, familial temporal lobe, 5	NM_020361
CSTB	21q22.3	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	NM_000100
DEPDC5	22q12.2-q12.3	Epilepsy, familial focal, with variable foci	NM_001242896
DYRK1A	21q22.13	Mental retardation, autosomal dominant 7	NM_001396
EPM2A	6q24.3	Epilepsy, progressive myoclonic 2A (Lafora)	NM_005670
FOLR1	11q13.4	Neurodegeneration due to cerebral folate transport deficiency	NM_016725
FOXG1	14q12	Rett syndrome, congenital variant	NM_005249
GABRA1	5q34	Epileptic encephalopathy, early infantile, 19	NM_000806
GABRB3	15q12	Epileptic encephalopathy	NM_000814
GABRG2	5q34	Epilepsy, generalized, with febrile seizures plus, type 3	NM_198903
GNAO1	16q12.2	Epileptic encephalopathy, early infantile, 17	NM_020988
GOSR2	17q21.32	Epilepsy, progressive myoclonic 6	NM_054022
GRIN1	9q34.3	Mental retardation, autosomal dominant 8	NM_001185090
GRIN2A	16p13.2	Epilepsy, focal, with speech disorder and with or without mental retardation	NM_001134407
GRIN2B	12p13.1	Mental retardation, autosomal dominant 6	NM_000834
HCN1	5p12	Epileptic encephalopathy, early infantile, 24	NM_021072
IQSEC2	Xp11.22	Mental retardation, X-linked 1	NM_001111125
KCNC1	11p15.1	Epilepsy, progressive myoclonic 7	NM_001112741
KCNH1	1q32.2	Temple-Baraitser syndrome	NM_172362
KCNJ10	1q23.2	SESAME syndrome	NM_002241
KCNQ2	20q13.33	Epileptic encephalopathy, early infantile, 7 / Myokymia / Seizures, benign neonatal, 1	NM_172107
KCNQ3	8q24.22	Seizures, benign neonatal, type 2	NM_004519
KCNT1	9q34.3	Epileptic encephalopathy, early infantile, 14 / Epilepsy, nocturnal frontal lobe, 5	NM_020822
KCTD7	7q11.21	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions	NM_153033
LGI1	10q23.33	Epilepsy, familial temporal lobe, 1	NM_005097
MBD5	2q23.1	Mental retardation, autosomal dominant 1	NM_018328
MECP2	Xq28	Rett syndrome, Encephalopathy, neonatal severe	NM_004992
MEF2C	5q14.3	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations	NM_001193347
NEDD4L	18q21.31	pas de pathologie OMIM	NM_001144967
NHLRC1	6p22.3	Epilepsy, progressive myoclonic 2B (Lafora)	NM_198586
PCDH19	Xq22.1	Epileptic encephalopathy, early infantile, 9 = epilepsy and mental retardation restricted to females	NM_001184880
PIGA	Xp22.2	Multiple congenital anomalies-hypotonia-seizures syndrome 2	NM_002641
PIGN	18q21.33	Multiple congenital anomalies-hypotonia-seizures syndrome 1	NM_176787
PIGO	9p13.3	Hyperphosphatasia with mental retardation syndrome 2	NM_032634.3
PNKP	19q13.33	Microcephaly, seizures, and developmental delay	NM_007254
PNPO	17q21.32	Pyridoxamine 5'-phosphate oxidase deficiency	NM_018129
POLG	15q26.1	Mitochondrial DNA depletion syndrome 4A (Alpers type)	NM_001126131
PRICKLE1	12q12	Epilepsy, progressive myoclonic 1B	NM_153026
PRRT2	16p11.2	Convulsions, familial infantile, with paroxysmal choreoathetosis / Episodic kinesigenic dyskinesia 1 / Seizures, benign familial infantile, 2	NM_001256443
SCARB2	4q21.1	Epilepsy, progressive myoclonic 4, with or without renal failure	NM_005506
SCN1A	2q24.3	Dravet syndrome / Epilepsy, generalized, with febrile seizures plus, type 2	NM_001165963
SCN1B	19q13.12	Epilepsy, generalized, with febrile seizures plus, type 1	NM_001037
SCN2A	2q24.3	Epileptic encephalopathy, early infantile, 11 , Seizures, benign familial infantile, 3	NM_021007
SCN8A	12q13.13	Epileptic encephalopathy, early infantile, 13	NM_014191
SLC2A1	1p34.2	GLUT1 deficiency syndrome 1	NM_006516
SLC6A1	3p25.3	Myoclonic-atonic epilepsy	NM_003042.3
SLC9A6	Xq26.3	Mental retardation, X-linked syndromic, Christianson type	NM_001042537
SLC13A5	17p13.1	Epileptic encephalopathy, early infantile, 25	NM_177550
SLC25A22	11p15.5	Epileptic encephalopathy, early infantile, 3	NM_001191061
SLC35A2	Xp11.23	Congenital disorder of glycosylation, type IIm	NM_005660
SPTAN1	9q34.11	Epileptic encephalopathy, early infantile, 5	NM_001130438
STAMBP	2p13.1	Microcephaly-capillary malformation syndrome	NM_006463
STX1B	16p11.2	Generalized epilepsy with febrile seizures plus, type 9	NM_052874
STXBP1	9q34.11	Epileptic encephalopathy, early infantile, 4	NM_003165
SYNGAP1	6p21.32	Mental retardation, autosomal dominant 5	NM_006772
TBC1D24	16p13.3	Deafness , autosomal recessive 86/DOOR syndrome/Epileptic encephalopathy, early infantile, 16/Myoclonic epilepsy, infantile, familial	NM_001199107
TCF4	18q21.2	Pitt-Hopkins syndrome	NM_001243226
UBE3A	15q11.2	Angelman syndrome	NM_000462
WWOX	16q21-q23	Epileptic encephalopathy, early infantile, 28	NM_016373