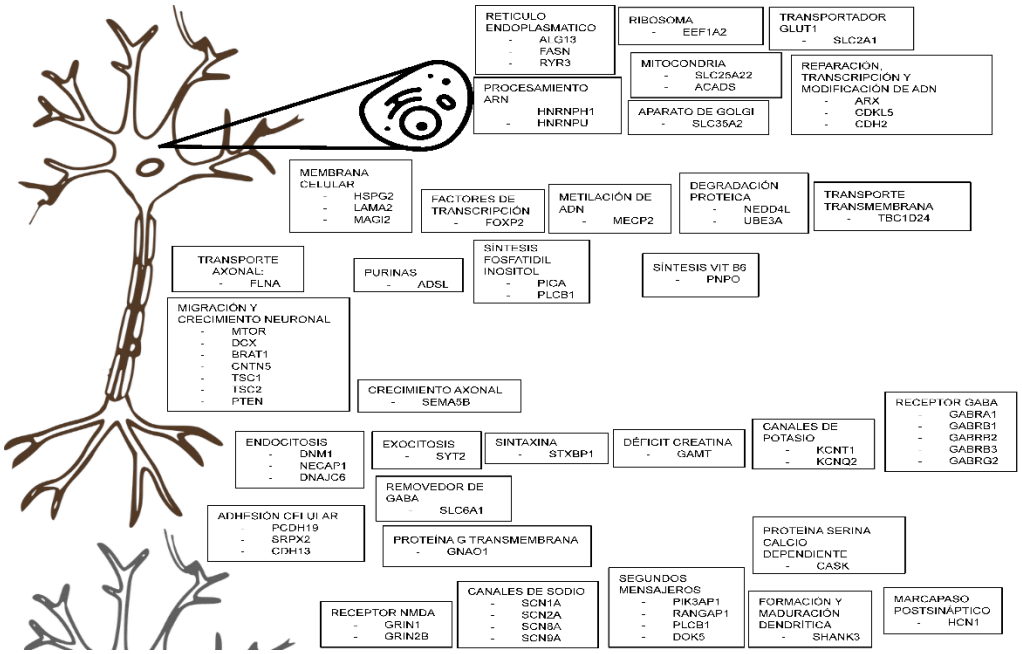
TABLA 1.- Genes envueltos y sus funciones en encefalopatías epilépticas genéticamente determinadas

Síndrome epiléptico	Genes asociados
Síndrome Ohtahara	ARX[1]; BRAT1[2]; CASK[3]; CDKL5 (STK9)[4]; GABRA1[5]; GNAO1[6]; KCNQ2[7]; NECAP1[8]; PIGA[9]; PLCB1[10]; PNPO[11]; SCN2A[12]; SLC25A22[13]; STXBP1[14]
Encefalopatía mioclónica temprana	GABRB2[15]; PIGA[9]; SLC25A22[13,16]
Síndrome de West	ACADS[4]; ADSL[17]; ALG13[17,18]; ARX[19]; CASK[17]; CDKL5 (STK9)[18,20]; DCX[18]; DNAJC6[18]; DNM1[21]; CNTN5[18]; EEF1A2[61]; EIF2C4[17]; FASN[21]; FLNA[18]; GABBR2[21]; GABRA1[18,22]; GABRB1[18]; GAMT[4]; GNAO1[21]; GRIN1[18]; GRIN2B[23]; HSPG2[17]; IL27RA[17]; LAMA2[17]; MAGI2[27]; MTOR[18]; MYO9B[17]; NEDD4L[18]; NPC1L1[17]; NR2F1[17]; PIK3AP1[21]; PNPO[17]; PTEN[18]; RANGAP1[21]; RYR3[21]; SCN1A[24]; SCN2A[25]; SCN8A[18]; SEMA5B[17]; SLC35A2[21]; SQSTM1[17]; STXBP1[17,26]; SVIL[17]; TSC1[18]; TSC2[18]; WDR45[17]
Epilepsia con convulsiones mioclónicas atónicas (astática) (Síndrome de Doose)	CDH2[28]; GABRB3[29]; GABRG2[29]; SCN1A[29]; SCN1B[29]; SCN2A[29]; SLC2A1[29]; SLC6A1[30,31]; STX1B[29,32,33]
Síndrome de Dravet	CDH2[34]; GABRA1[35]; GABRG2[36]; HCN1[37]; PCDH19[38]; SCN1A[39]; SCN1B[40]; SCN2A[41]; SCN9A[42,43]; STXBP1[35]
Estado de mal mioclónico en encefalopatías no progresivas	MECP2[44]; UBE3A[44]
Síndrome de Lennox-Gastaut	ALG13[18]; CDH2[18]; DCX[18]; DNAJC6[18]; DNM1[21]; FASN[21]; FLNA[18]; GABBR2[21]; GABRB3[22]; GRIN2B[18]; HNRNPH1[18]; HNRNPU[18]; IQSEC1[18]; IQSEC2[18]; MTOR[18]; RANGAP1[21]; RYR3[21]; SCN8A[18]; SLC35A2[21]; STXBP1[18]
Síndrome de Landau-Kleffner	FOXP2[45]; GRIN2A[46]; SLC26A1[47]; SRPX2[45,48]; SYT2[47]
Síndrome de punta-onda continua durante el sueño lento	ATX1[4,49]; CDH13[4,49]; DOK5[47]; FOXP2[45]; GRIN2A[46,50,51,52]; KCNQ2[53]; MDGA2[4,49]; SCN2A[62,63]; SHANK3[52]; SLC9A6[4,49]; SRPX2[4,45,48,49]
Convulsiones parciales migratorias malignas de la infancia	KCNT1[54]; PLCB1[55]; SCN1A[56]; SCN2A[57]; SCN8A[18,58]; SLC25A22[59]; STXBP1[18]; TBC1D24[60]
Epilepsia grave de la infancia con múltiples focos independientes	**No hay un gen específico que causa MISF debido a que normalmente evoluciona de otros síndromes**



Síndromes epilépticos y los genes asociados. A pesar de ser exhaustivo, el listado puede no incluir todos los genes actualmente informados ya que hay publicaciones frecuentes sobre el tema. Los genes se encuentran ordenados alfabéticamente y no por su incidencia.

El esquema muestra la zona celular donde los genes afectados tienen mayor efecto funcional
MISF: multiple independent spike foci (múltiple focos epileptógenos independientes)

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