Epileptic encephalopathy/ severe delay panels

GOSH (45 genes)

Name	Inheritance	
ADSL	AR	Adenylosuccinase deficiency
ALG13	AD	CDG 1s; now, new EIEE
ARHGEF9	XR	EIEE 8
ARX	XR	EIEE 1
ATP1A3	AD	Alternating hemi child/ CAPOS
ATRX	XR	Alpha thalassemia X-linked MR
CDKL5	X	EIEE 2
CHD2	AD	Childhood onset EE
CHRNA4	AD	ADNFLE
CHRNB2	AD	ADNFLE
CNTNAP2	AR	Pitt Hopkins like 1/ CDFE
EHMT1	AD	Kleefstra
FOXG1	AD	Congenital Rett
GABRB3	AD	Adjacent to UBE3A!
GRIN2A	AD	LKS/CSWS/FESD
GRIN2B	AD	West syndrome
KCNQ2	AD	EIEE 7
KCNT1	AD	EIEE 14 (MMPSI-~50%)
KIAA1279	AR	Goldberg-Shprintzen?why
LGI1	AD	ADPEAF
MAGI2	AD	Larger WBS deletion?
MBD5	AD	2q23.1 deletions
MECP2	X	Rett
MEF2C	AD	5q14.3 del/ point mutations
NRXN1	AR	Pitt Hopkins like 2/ Hz ASD+MR
PCDH19	XD	EIEE 9
PLCB1	AR	EIEE 12
PNKP	AR	EIEE 10
POLG	AR	Mitochondrial
PRRT2	AD	PKD, BFIS
SCN1A	AD	EIEE 6/Dravet
SCN2A	AD	EIEE 11
SCN8A	AD	EIEE 13
SLC16A2	XR	Allan-Herndon-Dudley
SLC25A22	AR	EIEE 3
SLC2A1	AR	GLUT1DS
SLC9A6	XR	XL Angelman (Christianson)
SPTAN1	AD	EIEE 5
STXBP1	AD	EIEE 4
SYNGAP1	AD	New DDD gene
TBC1D24	AR	DOORS/Epi/ EIEE 16
TCF4	AD	Pitt Hopkins
UBE2A	XR	XLMR syndromic
UBE3A	AD (imprinted)	Angelman
ZEB2	AD	Mowat Wilson

Cardiff (31 genes)

Gene	
ALDH7A1	Pyridoxine dependent
ARHGEF9	
ARX	
BTD	Biotinidase
CDKL5	
CNTNAP2	
FOXG1	
GABRG2	GEFS+
GLUD1	HH???
KCNQ2	
MAPK10	2 cases EE
MECP2	
MEF2C	
NRXN1	
PCDH19	
PLCB1	
PNKP	
PNPO	Pyridoxal phosphate
POLG	
SCN1A	
SCN2A	
SCN9A	GEFS+
SLC25A22	
SLC2A1	
SLC9A6	
SPTAN1	
SRGAP2	2 OMIM Tx
STXBP1	
TCF4	
UBE3A	
ZEB2	

Posted 150214, Abijit Dixit