

Supplementary Materials (Suppl. Tables 1-3)

Supplementary Table 1. Citations for justification of ratings for each high-risk autism gene.

Aut Gene	Rating	Refs for Rating	Refs for Rating	Function in Neurite or Synapse
<i>ACSL4</i>	3	Cho (2012)	Zhang et al. (2009)	Meloni et al., (2009)
<i>ADNP</i>	3	Helsmoortel et al. (2014)		Pacual & Guerri (2007)
<i>ADSL</i>	2	Leivo et al. (2005)	Ng et al. (2009)	
<i>AFF2</i>	3	Wittwer et al. (2001)		Gu & Nelson (2003)
<i>AGTR2</i>	3	Li et al. (2007)	Park & Zambidis (2009)	Maul et al. (2008)
<i>AHI1</i>	3	Weng et al. (2013)		Doering et al. (2008)
<i>ALDH5A1</i>	3	Tozuka et al. (2005)		Vardya et al. (2010)
<i>ALDH7A1</i>	2	Moreb (2008)	Mills et al. (2010)	
<i>APIS2</i>	2	Montpetit et al. (2008)	Kametaka et al. (2012)	Boehm & Bonifacino (2002)
<i>ARHGEF6</i>	2	Förster (2014)		Nodé-Langlois et al. (2006)
<i>ARID1B</i>	3	GeneCards (2014a)		Vogel-Ciernia et al. (2013)
<i>ARX</i>	3	Friocourt et al. (2008)		Yoshihara et al. (2005)
<i>ASMT</i>	2	Baka et al. (2010)		Melke et al. (2008)
<i>ATPI3A4</i>	2	Vallipuram et al. (2010)		
<i>ATRX</i>	3	Bérubé et al. (2005)	Bérubé et al. (2002)	Shioda et al. (2011)
<i>AUTS2</i>	3	Oksenberg et al. (2013)		
<i>AVPR1A</i>	1	GeneCards (2014b)		Pagani et al. (2014)
<i>BCL2</i>	3	Zhang et al. (1996)		Rolland & Conradt (2010)
<i>BDNF</i>	3	Ahmed et al. (1995)		An et al. (2008)
<i>BRAF</i>	3	Raabe et al. (2011)	Maddodi et al. (2010)	Pfeiffer et al. (2013)
<i>BTD</i>	2	Valenciano et al. (2002)		Pindolia et al. (2012)
<i>C4B</i>	2	Shinjyo et al. (2009)		
<i>CACNA1C</i>	3	Paşca et al. (2011)		Moosmang et al. (2005)
<i>CACNA1E</i>	2	Gu & Spitzer (1997)		
<i>CACNA1F</i>	2	Gu & Spitzer (1997)		Thibault et al. (2001)
<i>CACNA1H</i>	2	Gu & Spitzer (1997)		Chemin et al. (2002)
<i>CADMI</i>	2	Ito et al. (2011)	Pietri et al. (2008)	Hagiyama et al. (2009)
<i>CASK</i>	2	Najm et al. (2008)	Pietri et al. (2008)	Wang et al. (2008)
<i>CDH10</i>	1	Takeichi (1991)		Barber et al. (2011)
<i>CDH9</i>	1	Takeichi (1991)		Williams et al. (2011)
<i>CDKL5</i>	3	Valli et al. (2012)		Zhu et al. (2013b)

<i>CEP290</i>	2	Senocak et al. (2010)		Coppieters et al. (2010)
<i>CHD7</i>	3	Layman et al. (2009)		Melicharek et al. (2010)
<i>CHD8</i>	3	Nishiyama et al. (2012)	Otero et al. (2004)	
<i>CHRNA4</i>	2	Mohapel et al. (2005)		Didier et al. (1995)
<i>CHRNA2</i>	2	Mohapel et al. (2005)		Sun et al. (2008)
<i>CNTN4</i>	2	Hansford et al. (2003)		Mercati et al. (2013)
<i>CNTNAP2</i>	3	Peñagarikano et al. (2011)	Strauss et al. (2006)	Fujita et al. (2012)
<i>COMT</i>	2	Rowe et al. (1993)	Laifenfeld et al. (2002)	Forero et al. (2006)
<i>CREBBP</i>	3	Sharma et al. (2010)		Sharma et al. (2010)
<i>CYFIP1</i>	3	Napoli et al. (2008)	Castrén et al. (2005)	Napoli et al. (2008)
<i>DBH</i>	2	Rowe et al. (1993)	Laifenfeld et al. (2002)	Gustafson & Moore (1987)
<i>DCX</i>	3	Brown et al. (2003)	Jin et al. (2010)	Shmueli et al. (2001)
<i>DHCR7</i>	3	Yu & Patel (2005)	Whitehead & Cobo (2008)	Yu et al. (2005)
<i>DISC1</i>	3	Mao et al. (2009)		Kamiya et al. (2006)
<i>DMD</i>	3	Deng et al. (2009)		Montañez et al. (2004)
<i>DMPK</i>	1	Marteyn et al. (2011)	Tang et al. (2012)	Schulz et al. (2003)
<i>DNAH5</i>	1	Olbrich et al. (2002)		
<i>DRD3</i>	2	Rowe et al. (1993)		Andersen & Sonntag (2014)
<i>DRD4</i>	2	Rowe et al. (1993)		Kwon et al. (2008)
<i>DYRK1A</i>	3	Yabut et al. (2010)		Martinez de Lagran et al. (2012)
<i>EHMT1</i>	2	GeneCards (2014c)	Thambirajah et al. (2012)	Kramer et al. (2011)
<i>EIF4E</i>	3	GeneCards (2014d)		Hashimoto & Ishima (2010)
<i>EN2</i>	2	Sarnat et al. (2002)	Cosgaya et al. (1998)	Brunet et al. (2005)
<i>FBXO33</i>	1	Guo et al. (2014)		
<i>FEZF1</i>	3	Shimizu et al. (2010)		Watanabe et al. (2009)
<i>FEZF2</i>	3	Shimizu et al. (2010)		Rouaux & Arlotta (2010)
<i>FGD1</i>	3	Pasteris & Gorski (1999)	Cappello et al. (2006)	
<i>FGFR2</i>	3	Maric et al. (2007)		Schüller et al. (2008)
<i>FHIT</i>	2	Huber & Weiske (2008)		
<i>FMR1</i>	3	Castrén et al. (2005)	Moro et al. (2006)	Tucker et al. (2006)
<i>FOXG1</i>	3	Hanashima et al. (2004)		Polleux et al. (2007)
<i>FOXP1</i>	2	Rousso et al. (2012)		Palmesino et al. (2010)
<i>FOXP2</i>	3	Rousso et al. (2012)		Vernes et al. (2011)
<i>FTSJ1</i>	2	Freude et al. (2004)	Luo et al. (2010)	
<i>GABRA4</i>	3	Tozuka et al. (2005)	-	Duveau et al. (2011)
<i>GABRA5</i>	3	Tozuka et al. (2005)	-	Duveau et al. (2011)
<i>GABRB1</i>	3	Tozuka et al. (2005)	-	Duveau et al. (2011)
<i>GABRB3</i>	3	Tozuka et al. (2005)	-	Duveau et al. (2011)

<i>GABRG3</i>	3	Tozuka et al. (2005)		Duveau et al. (2011)
<i>GAD1</i>	3	Tozuka et al. (2005)		Chattopadhyaya et al. (2007)
<i>GAMT</i>	3	Ducray et al. (2007)		Braissant et al. (2001)
<i>GATM</i>	3	Ducray et al. (2007)		Braissant et al. (2001)
<i>GRIA3</i>	3	Suzuki et al. (2006)	Maric et al. (2000)	Sprengel (2006)
<i>GRIK2</i>	3	Joo et al. (2007)		Motazacker et al. (2007)
<i>GRIN2B</i>	3	Joo et al. (2007)		Dimassi et al. (2013)
<i>GRIN3B</i>	3	Joo et al. (2007)		Hensen et al. (2010)
<i>GRIP1</i>	3	Joo et al. (2007)		Hoogenraad et al. (2005)
<i>GRPR</i>	3	Walton et al. (2014)		Walton et al. (2014)
<i>GSE1</i>	0			
<i>GSTM1</i>	2	Richter & Kass (1991)	Salim et al. (2006)	Sobczak et al. (2014)
<i>GUCY2D</i>	0			
<i>HLA-DRB1</i>	0			
<i>HOXA1</i>	3	Martinez-Ceballos & Gudas (2008)		del Toro et al. (2001)
<i>HRAS</i>	3	Paquin et al. (2009)		Fivaz et al. (2008)
<i>IGF2</i>	3	Bracko et al. (2012)		Schmeisser et al. (2012)
<i>IL1RAPL1</i>	2	Gambino et al. (2007)	Deisseroth et al. (2004)	Valnegri et al. (2011)
<i>IMMP2L</i>	2	Xiao et al. (2013)	Maestrini et al. (2010)	Fogel et al. (2012)
<i>IQSEC2</i>	2	GeneCards (2014e)		Shoubridge et al. (2010)
<i>ITGB3</i>	3	Ma et al. (2010)	Azmitia (2001)	Whyte (2014)
<i>KCND2</i>	3	Schaarschmidt et al. (2009)		Kim & Hoffman (2012)
<i>KCNMA1</i>	2	Zhang et al. (2014)		Hebert et al. (2014)
<i>KDM5C</i>	3	Poeta et al. (2013)		Rosenfelder (2009)
<i>KIAA0100</i>	2	Bain et al. (2000)		
<i>KIAA2022</i>	2	Magome et al. (2013)		Van Maldergem et al. (2013)
<i>KRAS</i>	2	Uhrbom et al. (2002)		Storner & Zhu (2011)
<i>L1CAM</i>	3	Mikulak et al. (2012)		Schäfer & Frotscher (2012)
<i>L2HGDH</i>	1	Krell et al. (2011)		
<i>LAMP2</i>	1	Yasin et al. (2013)		
<i>LRFN5</i>	2	GeneCards (2014f)		Mah et al. (2010)
<i>LRP1</i>	3	Zilberberg et al. (2004)	Otero et al. (2004)	Yoon et al. (2013)
<i>MACROD2</i>	2	Corda & Di Girolamo (2003)	Sharifi et al. (2013)	
<i>MAOA</i>	2	Rowe et al. (1993)		Lee et al. (2005)
<i>MAP2K1</i>	3	Pang et al. (1995)	Jori et al. (2005)	GeneCards (2014g)
<i>MAPK1</i>	3	Qiu & Green (1992)		Sakai et al. (1999)

<i>MAPK3</i>	3	Qiu & Green (1992)		Song et al. (2005)
<i>MARK1</i>	2	Moroni et al. (2006)		Wu et al. (2011)
<i>MBD5</i>	2	Talkowski et al. (2011)		Camarena et al. (2014)
<i>MECP2</i>	3	Peddada et al. (2006)		Degano et al. (2009)
<i>MED12</i>	3	Wang et al. (2009)		Wang et al. (2009)
<i>MEF2C</i>	3	Li et al. (2008)		Barbosa et al. (2008)
<i>MEGF11</i>	1	Kay et al. (2012)		
<i>MET</i>	3	Kato et al. (2004)		Maina et al. (1997)
<i>MIDI</i>	2	Mnayer et al. (2006)	Dal Zotto et al. (1998)	Lu et al. (2013)
<i>MKKS</i>	2	Kim et al. (2005)		
<i>MSNPIAS</i>	2	Kerin et al. (2012)	Zhu et al. (2013c)	Paglini et al. (1998)
<i>NDP</i>	3	McNeill et al. (2013)		
<i>NFI</i>	3	Nishi et al. (1991)		Patrakitkomjom et al. (2008)
<i>NFIX</i>	3	Heng et al. (2014)		Wang et al. (2010)
<i>NHS</i>	2	Sharma et al. (2009)		
<i>NIPA1</i>	2	Tsang et al. (2009)	Ueberham & Arendt (2013)	Chen & Cheng (2009)
<i>NIPBL</i>	2	Pistocchi et al. (2013)		
<i>NLGN3</i>	1	Shi et al. (2013)		Budreck & Scheiffele (2007)
<i>NLGN4X</i>	3	Shi et al. (2013)		Shi et al. (2013)
<i>NPHP1</i>	2	Jauregui et al. (2008)	Chizhikov et al. (2007)	Eley et al. (2008)
<i>NRXN1</i>	3	Zeng et al. (2013)		Nussbaum et al. (2008)
<i>NRXN2</i>	1	Zeng et al. (2013)		Craig & Kang (2007)
<i>NSD1</i>	3	Berdasco et al. (2009)		
<i>NTF3</i>	3	Fariñas et al. (1996)	Ghosh & Greenberg (1995)	Dijkhuizen et al. (1997)
<i>NTF4</i>	3	Shen et al. (2010)		Cohen et al. (1994)
<i>OCRL</i>	3	Ramirez et al. (2012)		Ramirez et al. (2012)
<i>OPHN1</i>	2	Rocas et al. (2013)		Fauchereau et al. (2003)
<i>OTC</i>	2	Sautin et al. (2007)		Qureshi et al. (1998)
<i>OTX1</i>	3	Frantz et al. (1994)		Weimann et al. (1999)
<i>OXTR</i>	3	Bakos et al. (2013)	Jafarzadeh et al. (2014)	Sala et al. (2011)
<i>PAFAH1B1</i>	3	Pawlisz et al. (2008)		Hirotsune et al. (1998)
<i>PAH</i>	3	Liss & Grümer (1966)	Hörster et al. (2006)	Li et al. (2010)
<i>PCDH19</i>	3	Biswas (2012)	Gao et al. (2001)	Garrett (2009)
<i>PHF6</i>	3	Zhang et al. (2013)	Wang et al. (2013a)	Zhang et al. (2013)
<i>PHF8</i>	3	Qiu et al. (2010)		Asensio-Juan et al. (2012)
<i>PITX1</i>	2	Heldring et al. (2012)		
<i>POGZ</i>	2	GeneCards (2014h)		
<i>POMGNT1</i>	2	Hehr et al. (2007)	Verrotti et al. (2010)	Abbott et al. (2006)
<i>POMT1</i>	2	Beltrán-Valero de Bernabé et al. (2002)	Verrotti et al. (2010)	Judas et al. (2009)

<i>PQBPI</i>	2	Li et al. (2013)	Sheen et al. (2010)	Wang et al. (2013b)
<i>PRKCB</i>	1	NCBI (2014)		
<i>PRKX</i>	3	Blaschke et al. (2000)		Blaschke et al. (2000)
<i>PRSSI2</i>	1	Wolfer et al. (2001)		Mitsui et al. (2013)
<i>PTCHD1</i>	3	Ericson et al. (1997)	Schneider et al. (2001)	So et al. (2006)
<i>PTEN</i>	3	Lachyankar et al. (2000)		Lachyankar et al. (2000)
<i>PTPN11</i>	3	Gauthier et al. (2007)		Rosário et al. (2007)
<i>RAB39B</i>	2	Giannandrea et al. (2010)		Giannandrea et al. (2010)
<i>RAI1</i>	2	Seranski et al. (2001)		
<i>RAPGEF4</i>	3	Shi et al. (2006)		Penzes et al. (2011)
<i>RBFOX1</i>	3	Fogel et al. (2012)		Gehman et al. (2011)
<i>RELN</i>	3	Kim et al. (2002)	Tabata & Nakajima (2002)	Nichols & Olson (2010)
<i>RIMS3</i>	0			Kumar et al. (2010)
<i>RNF135</i>	2	Yoshiga et al. (2013)	Douglas et al. (2007)	
<i>RPE65</i>	2	Métrailler et al. (2013)		Koenekoop (2003)
<i>RPGRIP1L</i>	3	Spassky et al. (2008)		
<i>RPL10</i>	2	Dugani et al. (2010)		Chiocchetti et al. (2011)
<i>RPS6KA3</i>	3	Dugani et al. (2010)		Ammar et al. (2013)
<i>SATB2</i>	3	Gyorgy et al. (2008)	Savarese et al. (2009)	Zhang et al. (2012)
<i>SBF1</i>	2	Chaerkady et al. (2011)		Firestein & Cleary (2001)
<i>SCN1A</i>	2	Okamura & Shidara (1990)	Barber et al. (2011)	Reid et al. (2014)
<i>SCN2A</i>	1	Okamura & Shidara (1990)		
<i>SEMA5A</i>	3	Zhu et al. (2013a)		Schwamborn et al. (2004)
<i>SEZ6L2</i>	1	Anderson et al. (2012)		Kumar et al. (2008)
<i>SGSH</i>	3	Nurcombe et al. (1993)	Israsena et al. (2004)	Hantas-Ambroise et al. (1987)
<i>SHANK2</i>	2	Kim et al. (2009)		Grabrucker et al. (2011)
<i>SHANK3</i>	2	Schuetz et al. (2004)	Wang et al. (2014)	Grabrucker et al. (2011)
<i>SLC22A9</i>	1	Volk (2014)		
<i>SLC25A12</i>	2	Richter & Kass (1991)	Legagnol-Bestel et al. (2008)	Legagnol-Bestel et al. (2008)
<i>SLC6A4</i>	3	Page et al. (2009)		Daubert & Condron (2010)
<i>SLC6A8</i>	3	Ducray et al. (2007)		Braissant et al. (2001)
<i>SLC9A6</i>	2	Zhang-James et al. (2011)	Bergmann et al. (1991)	Ouyang et al. (2013)
<i>SLCO1C1</i>	3	Schlosser et al. (2002)	Honegger & Lenoir (1980)	Leonard & Farwell (1997)
<i>SMC1A</i>	2	Horsfield et al. (2007)	Frank & Tsai (2009)	
<i>ST8SIA2</i>	3	Amoureux et al. (2000)	Satin & Kitajima (2013)	Lee et al. (2011)
<i>SUV420H1</i>	3	Völkel & Angrand (2007)	Yu et al. (2009)	
<i>SYN1</i>	2	Horsfield et al. (2007)	Cai et al. (2003)	Paonessa et al. (2013)
<i>TBR1</i>	3	Bedogni et al. (2010)		Huang et al. (2014)
<i>TBX1</i>	3	Raft et al. (2004)		Forstner et al. (2013)

<i>TRIO</i>	2	Seipel et al. (1999)		Estrach et al. (2002)
<i>TSC1</i>	3	Meikle et al. (2007)	Sun et al. (2010)	Florice et al. (2007)
<i>TSC2</i>	3	Soucek et al. (1998)		Nie et al. (2010)
<i>TTN</i>	1	Qi et al. (2008)		
<i>TUBA1A</i>	3	Fallet-Bianco et al. (2008)	Poirier et al. (2007)	Veldman et al. (2010)
<i>TUBGCP5</i>	2	Fallet-Bianco et al. (2008)		Baas & Joshi (1992)
<i>UBE3A</i>	3	Mardirossian et al. (2009)	Mishra et al. (2009)	Greer et al. (2010)
<i>UPF3B</i>	3	Jolly et al. (2013)		Jolly et al. (2013)
<i>UPP2</i>	2	GeneCards (2014j)	Silei et al. (2000)	Rooslid et al. (2011)
<i>VIP</i>	3	Pincus et al. (1990)		White et al. (2000)
<i>VPS13B</i>	2	Budisteanu et al. (2010)	Baker (2013)	
<i>YWHAE</i>	2	Toyo-oka et al. (2003)		Toyo-oka et al. (2003)
<i>ZNF674</i>	0			
<i>ZNF81</i>	0			

References for Supplementary Table 1

Abbott, K. L., Troupe, K., Lee, I., and Pierce, M. (2006). Integrin-dependent neuroblastoma cell adhesion and migration on laminin is regulated by expression levels of two enzymes in the O-mannosyl-linked glycosylation pathway, PomGnT1 and GnT-Vb. *Exp. Cell Res.* *312*, 2837-2850.

Acosta, R., Montañez, C., Fuentes-Mera, L., Gonzalez, E., Gómez, P., Quintero-Mora, L., Mornet, D., Alvarez-Salas, L. M., and Cisneros, B. (2004). Dystrophin Dp71 is required for neurite outgrowth in PC12 cells. *Exp. Cell Res.* *296*, 265-275.

Ahmed, S., Reynolds, B. A., and Weiss, S. (1995). BDNF enhances the differentiation but not the survival of CNS stem cell-derived neuronal precursors. *J. Neurosci.* *15*, 5765-5778.

Ammar, M. R., Humeau, Y., Hanauer, A., Nieswandt, B., Bader, M. F., and Vitale, N. (2013). The Coffin-Lowry syndrome-associated protein RSK2 regulates neurite outgrowth through phosphorylation of phospholipase D1 (PLD1) and synthesis of phosphatidic acid. *J. Neurosci.* *33*, 19470-19479.

Amoureux, M. C., Cunningham, B. A., Edelman, G. M., and Crossin, K. L. (2000). N-CAM binding inhibits the proliferation of hippocampal progenitor cells and promotes their differentiation to a neuronal phenotype. *J. Neurosci.* *20*, 3631-3640.

An, J. J., Gharami, K., Liao, G. Y., Woo, N. H., Lau, A. G., Vanevski, F., Torre, E. R., Jones, K. R., Feng, Y., Lu, B., et al. (2008). Distinct role of long 3' UTR BDNF mRNA in spine morphology and synaptic plasticity in hippocampal neurons. *Cell* *134*, 175-187.

Andersen, S. L., and Sonntag, K. C. (2014). Juvenile methylphenidate reduces prefrontal cortex plasticity via D3 receptor and BDNF in adulthood. *Front. Synaptic Neurosci.* 6, 1.

Anderson, G. R., Galfin, T., Xu, W., Aoto, J., Malenka, R. C., and Südhof, T. C. (2012). Candidate autism gene screen identifies critical role for cell-adhesion molecule CASPR2 in dendritic arborization and spine development. *Proc. Natl. Acad. Sci. USA* 109, 18120-18125.

Asensio-Juan, E., Gallego, C., Martínez-Balbás, M. A. (2012). The histone demethylase PHF8 is essential for cytoskeleton dynamics. *Nucleic Acids Res.* 40, 9429-9440.

Azmitia, E. C. (2001). Modern views on an ancient chemical: serotonin effects on cell proliferation, maturation, and apoptosis. *Brain Res. Bull.* 56, 413-424.

Baas, P. W., and Joshi, H. C. (1992). Gamma-tubulin distribution in the neuron: implications for the origins of neuritic microtubules. *JCB* 119, 171-178.

Bain, G., Mansergh, F. C., Wride, M. A., Hance, J. E., Isogawa, A., Rancourt, S. L., Ray, W. J., Yoshimura, Y., Tsuzuki, T., Gottlieb, D. I., et al. (2000). ES cell neural differentiation reveals a substantial number of novel ESTs. *Funct. Integr. Genomics* 1, 127-139.

Baka, M., Uyanikgil, Y., Ateş, U., and Kültürsay, N. (2010). Investigation of maternal melatonin effect on the hippocampal formation of newborn rat model of intrauterine cortical dysplasia. *Childs Nerv. Syst.* 26, 1575-1581.

Baker, K. (2013). Syndromes not yet proven to be ciliopathies. In *Ciliopathies: A Reference for Clinicians*. T. D. Kenny and P. L. Beales, eds. (Oxford: Oxford University Press), pp-262-274.?

Bakos, J., Strbak, V., Paulikova, H., Krajnakova, L., Lestanova, Z., Bacova, Z. (2013). Oxytocin receptor ligands induce changes in cytoskeleton in neuroblastoma cells. *J. Mol. Neurosci.* 50, 462-468.

Barba, C., Parrini, E., Coras, R., Galuppi, A., Craiu, D., Kluger, G., Parmeggiani, A., Pieper, T., Schmitt-Mechelke, T., Striano, P., et al. (2014). Co-occurring malformations of cortical development and SCN1A gene mutations. *Epilepsia* 55, 1009-1019.

Barber, J. C., Huang, S., Bateman, M. S., and Collins, A. L. (2011). Transmitted deletions of medial 5p and learning difficulties; does the cadherin cluster only become penetrant when flanking genes are deleted? *Am. J. Med. Genet. A.* 155A, 2807-2815.

Barbosa, A. C., Kim, M. S., Ertunc, M., Adachi, M., Nelson, E. D., McAnally, J., Richardson, J. A., Kavalali, E. T., Monteggia, L. M., Bassel-Duby, R., et al. (2008). MEF2C, a transcription factor that facilitates learning and memory by negative regulation of synapse numbers and function. *Proc. Natl. Acad. Sci. USA* 105, 9391-9396.

Bedogni, F., Hodge, R. D., Elsen, G. E., Nelson, B. R., Daza, R. A., Beyer, R. P., Bammler, T. K., Rubenstein, J. L., and Hevner, R. F. (2010). Tbr1 regulates regional and laminar identity of postmitotic neurons in developing neocortex. *Proc. Natl. Acad. Sci. USA* *107*, 13129-13134.

Beltrán-Valero de Bernabé, D., Currier, S., Steinbrecher, A., Celli, J., van Beusekom, E., van der Zwaag, B., Kayserili, H., Merlini, L., Chitayat, D., Dobyns, W. B., (2002). Mutations in the O-mannosyltransferase gene POMT1 give rise to the severe neuronal migration disorder Walker-Warburg syndrome. *Am. J., Hum. Genet.* *71*, 1033-1043.

Berdasco, M., Ropero, S., Setien, F., Fraga, M. F., Lapunzina, P., Losson, R., Alaminos, M., Cheung, N. K., Rahman, N., and Esteller, M. (2009). Epigenetic inactivation of the Sotos overgrowth syndrome gene histone methyltransferase NSD1 in human neuroblastoma and glioma. *Proc. Natl. Acad. Sci. USA* *106*, 21830-21835.

Bergmann, M., Lahr, G., Mayerhoffer, A., and Gratzl, M. (1991). Expression of synaptophysin during the prenatal development of the rat spinal cord: correlation with basic differentiation processes of neurons. *Neuroscience* *42*, 569-582.

Bérubé, N. G., Jagla, M., Smeenk, C., De Repentigny, Y., Kothary, R., and Picketts, D. J. (2002). Neurodevelopmental defects resulting from ATRX overexpression in transgenic mice. *Hum. Mol. Genet.* *11*, 253-261.

Bérubé, N. G., Mangelsdorf, M., Jagla, M., Vanderluit, J., Garrick, D., Gibbons, R. J., Higgs, D. R., Slack, R. S., and Picketts, D. J. (2005). The chromatin-remodeling protein ATRX is critical for neuronal survival during corticogenesis. *J. Clin. Invest.* *115*, 258-267.

Biswas, S. (2012). Role of protocadherins in the zebrafish neural development. Diss. Defense. (The Ohio State University.) Accessed on 8/28/2014 from <http://gradworks.umi.com/35/35/3535233.html>

Blaschke, R. J., Monaghan, A. P., Bock, D., and Rappold, G. A. (2000). A novel murine PKA-related protein kinase involved in neuronal differentiation. *Genomics* *64*, 187-194.

Boehm, M., and Bonifacino, J. S. (2002). Genetic analyses of adaptin function from yeast to mammals. *Gene* *286*, 175-186.

Bracko, O., Singer, T., Aigner, S., Knoblock, M., Winner, B., Ray, J., Clemenson, G. D., Jr., Suh, H., Couillard-Despres, S., Aigner, L., Gage, F. H., et al. (2012). Gene expression profiling of neural stem cells and their neuronal progeny reveals IGF2 as a regulator of adult hippocampal neurogenesis. *J. Neurosci.* *32*, 3376-3387.

- Braissant, O., Henry, H., Loup, M., Eilers, B. and Bachmann, C. (2001). Endogenous synthesis and transport of creatine in the rat brain: an in situ hybridization study. *Brain Res. Mol. Brain Res.* 86, 193-201.
- Brown, J. P., Couillard, Després, S., Cooper-Kuhn, C. M., Winkler, J., Aigner, L. and Kuhn, H. G. (2003). Transient expression of doublecortin during adult neurogenesis. *J. Comp. Neurol.* 467, 1-10.
- Brunet, I., Weinl, C., Piper, M., Trembleau, A., Volovitch, M., Harris, W., Prochiantz, A., and Holt, C. (2005). The transcription factor Engrailed-2 guides retinal axons. *Nature* 438, 94-98.
- Budisteanu, M., Barca, D., Chirieac, S. M., and Magureanu, S. (2010). Cohen syndrome – a rare genetic cause of hypotonia in children. *Maedica* 5, 56-61.
- Budreck, E. C., and Scheiffele, P. (2007). Neuroligin-3 is a neuronal adhesion protein at GABAergic and glutamatergic synapses. *Eur. J., Neurosci.* 26, 1738-1748.
- Cai, X., Dong, F., Edelmann, R. E., and Makaroff, C. A. (2003). The *Arabidopsis* SYN1 cohesin protein is required for sister chromatid arm cohesion and homologous chromosome pairing. *J. Cell Sci.* 116, 2999-3007.
- Camarena, V., Cao, L., Abad, C., Abrams, A., Toledo, Y., Araki, K., Araki, M., Walz, K., and Young, J. I. (2014). Disruption of Mbd5 in mice causes neuronal functional deficits and neurobehavioral abnormalities consistent with 2q23.1 microdeletion syndrome. *EMBO Mol. Med.* 6, 1003-1015.
- Cappello, S., Attardo, A., Wu, X., Iwasato, T., Itohara, S., Wilsch-Bräuninger, M., Eilken, H. M., Rieger, M. A., Schroeder, T. T., Huttner, W. B., et al. (2006). The Rho-GTPase cdc42 regulates neural progenitor fate at the apical surface. *Nat. Neurosci.* 9, 1099-1107.
- Castrén, M., Tervonen, T., Kärkkäinen, V., Heinonen, S., Castrén, E., Larsson, K. Bakker, C. E., Oostra, B. A., and Akerman, K. (2005). Altered differentiation of neural stem cells in fragile X syndrome. *Proc. Natl. Acad. Sci. USA* 102, 17834-17839.
- Chaerkady, R., Letzen, B., Renuse, S., Sahasrabuddhe, N. A., Kumar, P., All, A. H., Thakor, N. V., Delanghe, B., Gearhart, J. D., Pandey, A., et al. (2011). Quantitative temporal proteomic analysis of human embryonic stem cell differentiation into oligodendrocyte progenitor cells. *Proteomics* 11, 4007-4020.
- Chattopadhyaya, B., Di, Cristo, G., Wu, C. Z., Knott, G., Kuhlman, S., Fu, Y., Palmiter, R. D., and Huang, Z. J. (2007). GAD67-mediated GABA synthesis and signaling regulate inhibitory synaptic innervation in the visual cortex. *Neuron* 54, 889-903.

Chemin, J., Nargeot, J., and Lory, P. (2002). Neuronal T-type alpha 1H calcium channels induce neuritogenesis and expression of high-voltage-activated calcium channels in the NG108-15 cell line. *J. Neurosci.* 22, 6856-6862.

Chen, S. Y., and Cheng, H. J. (2009). Functions of axon guidance molecules in synapse formation. *Curr. Opin. Neurobiol.* 19, 471-478.

Chiocchetti, A., Pakalapati, G., Duketis, E., Wieman, S., Poustka, A., Poustka, F., and Klauck, S. M. (2011). Mutation and expression analyses of the ribosomal protein gene RPL10 in an extended German sample of patients with autism spectrum disorder. *Am. J. Med. Genet.* 155A, 1472-1475.

Chizhikov, V. V., Davenport, J., Zhang, Q., Shih, E. K., Cabello, O. A., Fuchs, J. L., Yoder, B. K., and Millen, K. J. (2007). Cilia proteins control cerebellar morphogenesis by promoting expansion of the granule progenitor pool. *J. Neurosci.* 27, 9780-9789.

Cho, Y. Y. (2012). A novel role of brain-type ACS4 isotype in neuronal differentiation. *Biochem. Biophys. Res. Commun.* 419, 505-510.

Cohen, A., Bray, G. M., and Aguayo, A. J. (1994). Neurotrophin-4/5 (NT-4/5) increases adult rat retinal ganglion cell survival and neurite outgrowth in vitro. *J. Neurobiol.* 25, 953-959.

Coppieters, F., Lefever, S., Leroy, B. P., and De Baere, E. (2010). CEP290, a gene with many faces: mutation overview and presentation of CEP290base. *Hum. Mutat.* 31, 1097-1108.

Corda, D., and Di Girolamo, M. (2003). Functional aspects of protein mono-ADP-ribosylation. *EMBO J.* 22, 1953-1958.

Cosgaya, J. M., Aranda, A., Cruces, J., and Martín-Blanco, E. (1998). Neuronal differentiation of PC12 cells induced by engrailed homeodomain is DNA-binding specific and independent of MAP kinases. *J. Cell Sci.* 111, 2377-2384.

Craig, A. M., and Kang, Y. (2007). Neurexin-neuroligin signaling in synapse development. *Curr. Opin. Neurobiol.* 17, 43-52.

Dal Zotto, L., Quaderia, N. A., Elliott, R., Lingerfelter, P. A., Carrel, L., Valsecchi, V., Montini, E., Yen, C. H., Chapman, V., Kalcheva, I., et al. (1998). The mouse Mid1 gene: implications for the pathogenesis of Opitz syndrome and the evolution of the mammalian pseudoautosomal region. *Hum. Mol. Genet.* 7, 489-499.

Daubert, E. A., and Condrón, B. G. (2010). Serotonin: a regulator of neuronal morphology and circuitry. *Trends Neurosci.* 33, 424-434.

Degano, A. L., Pasterkamp, R. J., and Ronnett, G. V. (2009). MeCP2 deficiency disrupts axonal guidance, fasciculation, and targeting by altering Semaphorin 3F function. *Mol. Cell Neurosci.* *42*, 243-254.

Deisseroth, K., Singla, S., Toda, H., Monje, M., Palmer, T. D., and Malenka, R. C. (2004). Excitation-neurogenesis coupling in adult neural stem/progenitor cells. *Neuron* *42*, 535-552.

del Toro, E. D., Borday, V., Davenne, M., Neun, R., Rijli, F. M., and Champagnat, J. (2001). Generation of a novel functional neuronal circuit in *Hoxa1* mutant mice. *J. Neurosci.* *21*, 5637-5642.

Deng, B., Glanzman, D., and Tidball, J. G. (2009). Nitric oxide generated by muscle corrects defects in hippocampal neurogenesis and neural differentiation caused by muscular dystrophy. *J. Physiol.* *587*, 1769-1778.

Didier, M., Bix, G., Berman, S. A., and Bursztajn, S. (1995). Expression of the alpha 4 neuronal nicotinic acetylcholine receptor subunit in the developing mouse hippocampus. *Int. J. Dev. Neurosci.* *13*, 703-713.

Dijkhuizen, P. A., Hermens, W. T., Teunis, M. A., and Verhaagen, J. (1997). Adenoviral vector-directed expression of neurotrophin-3 in rat dorsal root ganglion explants results in a robust neurite outgrowth response. *J. Neurobiol.* *33*, 172-184.

Dimassi, S., Andrieux, J., Labalme, A., Lesca, G., Cordier, M. P., Boute, O., Neut, D., Edery, P., Sanlaville, D., and Schluth-Bolard, C. (2013). Interstitial 12p13.1 deletion involving *GRIN2B* in three patients with intellectual disability. *Am. J. Med. Genet.* *161*, 2564-2569.

Doering, J. E., Kane, K., Hsiao, Y. C., Yao, C., Shi, B., Slowik, A. D., Dhagat, B., Scott, D. D., Ault, J. G., Page-McCaw, P. S., and Ferland, R. J. (2008). Species differences in the expression of *Ahi1*, a protein implicated in the neurodevelopmental disorder Joubert syndrome, with preferential accumulation to stigmoid bodies. *J. Comp. Neurol.* *511*, 238-256.

Douglas, J., Cilliers, D., Coleman, K., Tatton-Brown, K., Barker, K., Bernhard, B., Burn, J., Huson, S., Josifova, D., et al. (2007). Mutations in *RNF135*, a gene within the NF1 microdeletion region, cause phenotypic abnormalities including overgrowth. *Nat. Genet.* *39*, 963-965.

Ducray, A. D., Schläppi, J. A., Qualls, R., Andres, R. H., Seiler, R. W., Schlattner, U., Wallimann, T., and Widmer, H. R. (2007). Creatine treatment promotes differentiation of GABA-ergic neuronal precursors in cultured fetal rat spinal cord. *J. Neurosci. Res.* *85*, 1863-1875.

- Dugani, C. B., Paquin, A., Kaplan, D. R., and Miller, F. D. (2010). Coffin-Lowry syndrome: a role for RSK2 in mammalian neurogenesis. *Dev. Biol.* *347*, 348-359.
- Duveau, V., Laustela, S., Barth, L., Gianolini, F., Vogt, K. E., Keist, R., Chandra, D., Homanics, G. E., Rudolph, U., and Fritschy, J. M. (2011). Spatiotemporal specificity of GABA_A receptor-mediated regulation of adult hippocampal neurogenesis. *Eur. J. Neurosci.* *34*, 362-373.
- Eley, L., Moolchhala, S. H., Simms, R., Hildebrandt, F., and Sayer, J. A. (2008). Nephrocystin-1 interacts directly with Ack1 and is expressed in human collecting duct. *Biochem. Biophys. Res. Commun.* *371*, 877-882. *Cell* *90*, 169-180.
- Ericson, J., Rashbass, P., Schedl, A., Brenner-Morton, S., Kawakami, A., van Heyningen, V., Jessell, T. M., and Briscoe, J. (1997). Pax6 controls progenitor cell identity and neuronal fate in response to graded Shh signaling.
- Estrach, S., Schmidt, S., Diriong, S., Penna, A., Blangy, A., Fort, P., and Debant, A. (2002). The human Rho-GEF trio and its target GTPase RhoG are involved in the NGF pathway, leading to neurite outgrowth. *Curr. Biol.* *12*, 307-312.
- Fallet-Bianco, C., Loeuillet, L., Poirier, K., Loget, P., Chapon, F., Pasquier, L., Saillour, Y., Beldjord, C., Chelly, J., and Francis, F. (2008). Neuropathological phenotype of a distinct form of lissencephaly associated with mutations in TUBA1A. *Brain* *131*, 2304-2320.
- Fariñas, I., Yoshida, C. K., Backus, C., and Reichardt, L. F. (1996). Lack of neurotrophin-3 results in death of spinal sensory neurons and premature differentiation of their precursors. *Neuron* *17*, 1065-1078.
- Fauchereau, F., Herbrand, U., Chafey, P., Eberth, A., Koulakoff, A., Vinet, M. C., Admadian, M. R., Chelly, J., and Billuart, P. (2003). The RhoGAP activity of OPHN1, a new F-actin-binding protein, is negatively controlled by its amino-terminal domain. *Mol. Cell Neurosci.* *23*, 574-586.
- Fivaz, M., Bandara, S., Inoue, T., and Meyer, T. (2008). Robust neuronal symmetry breaking by Ras-triggered local positive feedback. *Curr. Biol.* *18*, 44-50.
- Firestein, R., and Cleary, M. L. (2001). Pseudo-phosphatase Sbf1 contains an N-terminal GEF homology domain that modulates its growth regulatory properties. *J. Cell. Sci.* *114*, 2921-2927.
- Florice, F., Higaki, K., Maki, H., Nanba, E., Ninomiya, H., and Ohno, K. (2007). Antisense suppression of TSC1 gene product, hamartin, enhances neurite outgrowth in NGF-treated PC12h cells. *Brain Dev.* *29*, 502-529.

Fogel, B. L., Wexler, E., Wahnich, A., Friedrich, T., Vijayendran, C., Gao, F., Parikshak, N., Konopka, G., and Geschwind, D. H. (2012). RBFOX1 regulates both splicing and transcriptional networks in human neuronal development. *Hum. Mol. Genet.* *21*, 4171-4186.

Forero, D. A., Benítez, B., Arboleda, G., Yunis, J. J., Pardo, R., and Arboleda, H. (2006). Analysis of functional polymorphisms in three synaptic plasticity-related genes (BDNF, COMT and UCHL1) in Alzheimer's disease in Colombia. *Neurosci. Res.* *55*, 334-341.

Förster, E. (2014). Reelin, neuronal polarity and process orientation of cortical neurons. *Neuroscience* *269*, 102-111.

Forstner, A. J., Degenhardt, F., Schratt, G., and Nöthen, M. M. (2013). MicroRNAs as the cause of schizophrenia in 22q11.2 deletion carriers, and possible implications for idiopathic disease: a mini-review. *Front. Mol. Neurosci.* *6*, 47.

Frank, C. L., and Tsai, L. H. (2009). Alternative functions of core cell cycle regulators in neuronal migration, neuronal maturation, and synaptic plasticity. *Neuron* *62*, 312-326.

Frantz, G. D., Weimann, J. M., Levin, M. E., and McConnell, S. K. (1994). Otx1 and Otx2 define layers and regions in developing cerebral cortex and cerebellum. *J. Neurosci.* *14*, 5725-5740.

Freude, K., Hoffman, K., Jensen, L. R., Delatycki, M. B., des Portes, V., Moser, B., Hamel, B., van Bokhoven, H., Moraine, C., Fryns, J. P., et al. (2004). Mutations in the FTSJ1 gene coding for a novel S-adenosylmethionine-binding protein cause nonsyndromic X-linked mental retardation. *Am. J. Hum. Genet.* *75*, 305-309.

Friocourt, G., Kanatani, S., Tabata, H., Yozu, M., Takahashi, T., Antypa, M., Raguénès, O., Chelly, J., Férec, C., Nakajima, K., et al. (2008). Cell-autonomous roles of ARX in cell proliferation and neuronal migration during corticogenesis. *J. Neurosci.* *28*, 5794-5805.

Fujita, E., Tanabe, Y., Momoi, M. Y., and Momoi, T. (2012). Cntnap2 expression in the cerebellum of Foxp2(R552H) mice, with a mutation related to speech-language disorder. *Neurosci. Lett.* *506*, 277-280.

Gambino, F., Pavlowsky, A., Béglé, A., Dupont, J. L., Bahi, N., Courjaret, R., Gardette, R., Hadjkacem, H., Skala, H., Poulain, B., et al. (2007). IL1-receptor accessory protein-like 1 (IL1RAPL1), a protein involved in cognitive functions, regulates N-types Ca²⁺-channel and neurite elongation. *Proc. Natl. Acad. Sci. USA* *104*, 9063-9068.

Gao, X., Bian, W., Yang, J., Tang, K., Kitani, H., Atsumi, T., and King, N. (2001). A role of N-cadherin in neuronal differentiation of embryonic carcinoma P19 cells. *Biochem. Biophys. Res. Commun.* *284*, 1098-1103.

Garrett, A. (2009). Control of synaptogenesis and dendritic arborization by the Y-protocadherin family of adhesion molecules. Diss. Defense. (The University of Iowa). Accessed on 8/28/2014 from <http://ir.uiowa.edu/etd/362/>

Gauthier, A. S., Furstoss, O., Araki, T., Chan, R., Neel, B. G., Kaplan, D. R., and Miller F. D. (2007). Control of CNS cell-date decisions by SHP-2 and its dysregulation in Noonan syndrome. *Neuron* 54, 245-262.

Gehman, L. T., Stoilov, P., Maguire, J., Damianov, A., Lin, C. H., Shiue, L., Ares, M., Jr., Mody, I., and Black, D. L. (2011). The splicing regulator Rbfox1 (A2BP1) controls neuronal excitation in the mammalian brain. *Nat. Genet.* 43, 706-711.

GeneCards. (2014a). AT Rich Interactive Domain 1B (SWI1-Like). Accessed on 8/26/2014 from <http://www.genecards.org/cgi-bin/carddisp.pl?gene=ARID1B&search=ARID1B>

GeneCards. (2014b). Arginine Vasopressin Receptor 1A. Accessed on 8/26/2014 from <http://www.genecards.org/cgi-bin/carddisp.pl?gene=AVPR1A&search=AVPR1A>

Genecards. (2014c). Euchromatic Histone-Lysine N-Methyltransferase 1. Accessed on 8/27/2014 from <http://www.genecards.org/cgi-bin/carddisp.pl?gene=EHMT1&search=EHMT1>

GeneCards. (2014d). Eukaryotic Translation Initiation Factor 4E. Accessed on 8/27/2014 from <http://www.genecards.org/cgi-bin/carddisp.pl?gene=EIF4E&search=EIF4E>

GeneCards. (2014e). IQ Motif and Sec7 Domain 2. Accessed on 8/27/2014 from <http://www.genecards.org/cgi-bin/carddisp.pl?gene=IQSEC2&search=IQSEC2>

GeneCards. (2014f). Leucine Rich Repeat and Fibronectin Type III Domain Containing... Accessed on 8/27/2014 from <http://www.genecards.org/cgi-bin/carddisp.pl?gene=LRFN5&search=LRFN5>

GeneCards. (2014g). Mitogen-activated Protein Kinase Kinase 1. Accessed on 8/27/2014 from <http://www.genecards.org/cgi-bin/carddisp.pl?gene=MAP2K1&search=5d645617d4626ea19e99af87bce79f87>

GeneCards. (2014h). Pogo Transposable Element with ZNF Domain. Accessed on 8/28/2014 from <http://www.genecards.org/cgi-bin/carddisp.pl?gene=POGZ&search=POGZ>

GeneCards. (2014j). Uridine Phosphorylase 2. Accessed on 9/6/2014 from <http://www.genecards.org/cgi-bin/carddisp.pl?gene=UPP2&search=3665af89ad8056ac5436a59892fd395b>

- Ghosh, A., and Greenberg, M. E (1995). Distinct roles for bFGF and NT-3 in the regulation of cortical neurogenesis. *Neuron* *15*, 89-103.
- Giannandrea, M., Bianchi, V., Mignogna, M. L., Sirri, A., Carrabino, S., D'Elia, E., Vecellio, M., Russo, S., Cogliati, F., Larizza, L., et al. (2010). Mutations in the small GTPase gene RAB39B are responsible for X-linked mental retardation associated with autism, epilepsy, and macrocephaly. *Am. J. Hum. Genet.* *86*, 185-195.
- Grabrucker, A. M., Knight, M. J., Proepper, C., Bockman, J., Joubert, M., Rowan, M., Nienhaus, G. U., Garner, C. C., Bowie, J. U., Kreutz, M. R., et al. (2011). Concerted action of zinc and ProSAP/Shank in synaptogenesis and synapse maturation. *EMBO J.* *30*, 569-581.
- Greer, P. L., Hanayama, R., Bloodgood, B. L., Mardinly, A. R., Lipton, D. M., Flavell, S. W., Kim, T.-K., Griffith, E. C., Waldon, Z., Maehr, R., et al. (2010). The Angelman syndrome protein Ube3A regulates synapse development by ubiquitinating Arc. *Cell* *140*, 704-716.
- Gu, Y., and Nelson, D. L. (2003). FMR2 function: insight from a mouse knockout model. *Cytogenet. Genome Res.* *100*, 129-139.
- Gu, X., and Spitzer, N. C. (1997). Breaking the code: regulation of neuronal differentiation by spontaneous calcium transients. *Dev. Neurosci.* *19*, 33-41.
- Guo, J., North, B. J., Tron, A. E., Inuzuka, H., and Wei, W. (2014). The role of FBXO subfamily of F-box proteins in tumorigenesis. In *SCF and APC E3 Ubiquitin Ligases in Tumorigenesis*. H. Inuzuka and W. Wei, eds. (Springer International Publishing), pp. 73-87.
- Gustafson, E. L., and Moore, R. Y. (1987). Noradrenaline neuron plasticity in developing rat brain: effects of neonatal 6-hydroxydopamine demonstrated by dopamine-beta-hydroxylase immunocytochemistry. *Brain Res.* *465*, 143-155.
- Gyorgy, A. B., Szemes, M., de Juan Romero, C., Tarabykin, V., and Agoston, D. V. (2008). SATB2 interacts with chromatin-remodeling molecules in differentiating cortical neurons. *Eur. J. Neurosci.* *27*, 865-873.
- Hagiyama, M., Ichiyangi, N., Kimura, K. B., Murakami, Y., and Ito, A. (2009). Expression of a soluble isoform of cell adhesion molecule 1 in the brain and its involvement in directional neurite outgrowth. *Am. J. Pathol.* *174*, 2278-2289.
- Hanashima, C., Li, S. C., Shen, L., Lai, E., and Fishell, G. (2004). Foxg1 suppresses early cortical cell fate. *Science* *303*, 56-59.

- Hansford, L. M., Smith, S. A., Haber, M., Norris, M. D., Cheung, B., and Marshall, G. M. (2003). Cloning and characterization of the human neural cell adhesion molecule, CNTN4 (alias BIG-2). *Cytogenet. Genome Res.* *101*, 17-23.
- Hantaz-Ambroise, D., Vigny, M., and Koenig, J. (1987). Heparan sulfate proteoglycan and laminin mediate two different types of neurite outgrowth. *J. Neurosci.* *7*, 2293-2304.
- Hashimoto, K., and Ishima, T. (2010). A novel target of action of minocycline in NGF-induced neurite outgrowth in PC12 cells: translation initiation factor eIF4AI. *PLoS One* *5*, e15430.
- Hebert, B., Pietropaolo, S., Mème, S., Laudier, B., Laugeray, A., Doisne, N., Quartier, A., Lefeuvre, S., Got, L., Cahard, D., et al. (2014). Rescue of fragile X syndrome phenotypes in Fmr1 KO mice by the small-molecule PAK inhibitor FRAX486. *Orphanet. J., Rare Dis.* *9*, 124.
- Hehr, U., Uyanik, G., Gross, C., Walter, M. C., Bohring, A., Cohen, M., Oehl-Jaschkowitz, B., Bird, L. M., Shamdeen, G. M., Bogdahn, U., et al. (2007). Novel POMGnT1 mutations define broader phenotypic spectrum of muscle-eye-brain disease. *Neurogenetics* *8*, 279-288.
- Heldring, N., Joseph, B., Hermanson, O., and Kiousi, C. (2012). Pitx2 expression promotes p21 expression and cell cycle exit in neural stem cells. *CNS Neurol. Disord. Drug Targets* *11*, 884-892.
- Helsmoortel, C., Vulto-van Silfhout, A. T., Coe, B. P., Vandeweyer, G., Rooms, L., van den Ende, J., Schuurs-Hoeijmakers, J. H., Marcelis, C. L., Willemsen, M. H., Vissers, L. E., et al. (2014). A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. *Nat. Genet.* *46*, 380-384.
- Heng, Y. H., McLeay, R. C., Harvey, T. J., Smith, A. G., Barry, G., Cato, K., Plachez, C., Little, E., Mason, S., Dixon, C., et al. (2014). NFIX regulates neural progenitor cell differentiation during hippocampal morphogenesis. *Cereb. Cortex* *24*, 261-279.
- Henson, M. A., Roberts, A. C., Pérez-Otaño, I., and Philpot, B. D. (2010). Influence of the NR3A subunit on NMDA receptor functions. *Prog. Neurobiol.* *91*, 23-37.
- Hirotsune, S., Fleck, M. W., Gambello, M. J., Bix, G. J., Chen, A., Clark, G. D., Ledbetter, D. H., McBain, C. J., and Wynshaw-Boris, A. (1998). Graded reduction of Pafah1b1 (Lis1) activity results in neuronal migration defects and early embryonic lethality. *Nat. Genet.* *19*, 333-339.
- Honegger, P., and Lenoir, D. (1980). Triiodothyronine enhancement of neuronal differentiation in aggregating fetal rat brain cells cultured in a chemically defined medium. *Brain Res.* *199*, 425-434.

Hoogenraad, C. C., Milstein, A. D., Ethell, I. M., Henkemeyer, M., and Sheng, M. (2005). GRIP1 controls dendrite morphogenesis by regulating EphB receptor trafficking. *Nat. Neurosci.* 8, 906-915.

Horsfield, J. A., Anagnostou, S. H., Hu, J. K., Cho, K. H., Geisler, R., Lieschke, G., Crosier, K. E., and Crosier, P. S. (2007). Cohesin-dependent regulation of Runx genes. *Development* 134, 2639-2649.

Hörster, F., Schwab, M. A., Sauer, S. W., Pietz, J., Hoffman, G. F., Okun, J. G., Kölker, S., and Kins, S. (2006). Phenylalanine reduces synaptic density in mixed cortical cultures from mice. *Pediatr. Res.* 59, 544-548.

Huang, T. N., Chuang, H. C., Chou, W. H., Chen, C. Y., Wang, H. F., Chou, S. J., and Hsueh, Y. P. (2014). Tbr1 haploinsufficiency impairs amygdalar axonal projections and results in cognitive abnormality. *Nat. Neurosci.* 17, 240-247.

Huber, O., and Weiske, J. (2008). Beta-catenin takes a HIT. *Cell Cycle* 7, 1326-1331.

Israsena, N., Hu, M., Fu, W., Kan, L., and Kessler, J. A. (2004). The presence of FGF2 signaling determines whether beta-catenin exerts effects on proliferation or neuronal differentiation of neural stem cells. *Dev Biol.* 268, 220-231.

Ito, T., Williams-Nate, Y., Iwai, M., Tsuboi, Y., Hagiyaama, M., Ito, A., Sakurai-Yageta, M., and Murakami, Y. (2011). Transcriptional regulation of the CADM1 gene by retinoic acid during the neural differentiation of murine embryonal carcinoma P19 cells. *Genes Cells* 16, 791-802.

Jafarzadeh, N., Javeri, A., Khaleghi, M., and Taha, M. F. (2014). Oxytocin improves proliferation and neural differentiation of adipose tissue-derived stem cells. *Neurosci. Lett.* 564, 105-110.

Jauregui, A. R., Nguyen, K. C., Hall, D. H. and Barr, M. M. (2008). The *Caenorhabditis elegans* nephrocystins act as global modifiers of cilium structure. *J. Cell Biol.* 180, 973-988.

Jin, K., Wang, X., Xie, L., Mao, X. O., and Greenberg, D. A. (2010). Transgenic ablation of doublecortin-expressing cells suppresses adult neurogenesis and worsens stroke outcome in mice. *Proc. Natl. Acad. Sci. USA* 107, 7993-7998.

Jolly, L. A., Homan, C. C., Jacob, R., Barry, S., and Gecz, J. (2013). The *UPF3B* gene, implicated in intellectual disability, autism, ADHD and childhood onset schizophrenia regulates neural progenitor cell behaviour and neuronal outgrowth. *Hum. Mol. Genet.* 22, 4673-4687.

Joo, J. Y., Kim, B. W., Lee, J. S., Park, J. Y., Kim, S., Yun, Y. J., Lee, S. H., Lee, S. H., Rhim, H., and Son, H. (2007). Activation of NDMA receptors increases proliferation and differentiation of hippocampal neural progenitor cells. *J. Cell Sci.* *120*, 1358-1370.

Jori, F. P., Napolitano, M. A., Melone, M. A., Cipollaro, M., Cascino, A., Altucci, L., Peluso, G., Giordano, A., and Galderisi, U. (2005). Molecular pathways involved in neural in vitro differentiation of marrow stromal stem cells. *J. Cell. Biochem.* *94*, 645-655.

Judas, M., Sedmak, G., Rados, M., Sarnavka, V., Fumić, K., Willer, T., Gross, C., Hehr, U., Strahl, S., Cuk, M., et al. (2009). POMT1-associated Walker-Warburg syndrome: a disorder of dendritic development of neocortical neurons. *Neuropediatrics* *40*, 6-14.

Kametaka, S., Kametaka, A., Yonekura, S., Haruta, M., Takenoshita, S., Goto, S., and Waguri, S. (2012). AP-1 clathrin adaptor and CG8538/Aftiphilin are involved in Notch signaling during eye development in *Drosophila melanogaster*. *J. Cell. Sci.* *125*, 634-648.

Kamiya, A., Tomoda, T., Chang, J., Takaki, M., Zhan, C., Morita, M., Cascio, M. B., Elashvili, S., Koizum, H., Takanezawa, Y., et al. (2006). DISC1-NDEL1/NUDEL protein interaction, an essential component for neurite outgrowth, is modulated by genetic variations of DISC1. *Hum. Mol. Genet.* *15*, 3313-3323.

Kato, M., Yoshimura, S., Kohuzawa, J., Kitajima, H., Kaku, Y., Iwama, T., Shinoda, J., Kunisada, T., and Sakai, N. (2004). Hepatocyte growth factor promotes neuronal differentiation of neural stem cells derived from embryonic stem cells. *Neuroreport* *15*, 5-8.

Kay, J. N., Chu, M. W., and Sanes, J. R. (2012). MEGF10 and MEGF11 mediate homotypic interactions required for mosaic spacing of retinal neurons. *Nature* *483*, 465-469.

Kerin, T., Ramanathan, A., Rivas, K., Grepo, N., Coetzee, G. A., and Campbell, D. B. (2012). A noncoding RNA antisense to moesin at 5p14.1 in autism. *Sci. Transl. Med.* *4*, 128ra40.

Kim, E., and Hoffman, D. A. (2012). Dynamic regulation of synaptic maturation state by voltage-gated A-type K⁺ channels in CA1 hippocampal pyramidal neurons. *J. Neurosci.* *32*, 14427-14432.

Kim, J. H., Kim, J. H., Yang, E., Park, J. H., Yu, Y. S., and Kim, K. W. (2009). Shank2 expression coincides with neuronal differentiation in the developing retina. *Exp. Mol. Med.* *41*, 236-242.

Kim, H. M., Qu, T., Kriho, V., Lacor, P., Smalheiser, N., Pappas, G. D., Guidotti, A., Costa, E., and Sugaya, K. (2002). Reelin function in neural stem cell biology. *Proc. Natl. Acad. Sci. USA* *99*, 4020-4025.

Kim, J. C., Ou, Y. Y., Badano, J. L., Esmail, M. A., Leitch, C. C., Friedrich, E., Beales, P. L., Archibald, J. M., Katsanis, N., Rattner, J. B., et al. (2005). MKKS/BBS6, a divergent chaperonin-like protein linked to the obesity disorder Bardet-Biedl syndrome, is a novel centrosomal component required for cytokinesis. *J. Cell Sci.* *118*, 1007-1020.

Koenekoop, R. K. (2003). Abnormal retinal architecture in a 33-week-old fetus with LCA and a homozygous C330Y mutation in RPE65. *Ophthalmic Genet.* *24*, 125-126.

Kramer, J. M., Kochinke, K., Oortveld, M. A., Marks, H., Kramer, D., de Jong, E. K., Asztalos, Z., Westwood, J. T., Stunnenberg, H. G., Sokolowski, M. B., et al. (2011). Epigenetic regulation of learning and memory by *Drosophila* EHMT/G9a. *PLoS Biol.* *9*, e1000569.

Krell, D., Assoku, M., Galloway, M., Mulholland, P., Tomlinson, I., and Bardella, C. (2011). Screen for IDH1, IDH2, IDH3, D2HGDH and L2HGDH mutations in glioblastoma. *PLoS One* *6*, e19868.

Kumar, R. A., KaraMohamed, S., Sudi, J., Conrad, D. F., Brune, C., Badner, J. A., Gilliam, T. C., Nowak, N. J., Cook, E. H., Jr., Dobyns, W. B., et al. (2008). Recurrent 16p11.2 microdeletions in autism. *Hum. Mol. Genet.* *17*, 628-638.

Kumar, R. A., Sudi, J., Babatz, T. D., Brune, C. W., Oswald, D., Yen, M., Nowak, N. J., Cook, E. H., Christian, S. L., and Dobyns, W. B. (2010). A de novo 1p34.2 microdeletion identifies the synaptic vesicle gene RIMS3 as a novel candidate for autism. *J. Med. Genet.* *47*, 81-90.

Kwon, O. B., Paredes, D., Gonzalez, C. M., Neddens, J., Hernandez, L., Vullhorst, D., and Buonanno, A. (2008). Neuregulin-1 regulates LTP at CA1 hippocampal synapses through activation of dopamine D4 receptors. *Proc. Natl. Acad. Sci. USA* *105*, 15587-15592.

Lachyankar, M. B., Sultana, N., Schonhoff, C. M., Mitra, P., Poluha, W., Lambert, S., Quesenberry, P. J., Litofsky, N. S., Recht, L. D., Nabi, R., et al. (2000). A role for nuclear PTEN in neuronal differentiation. *J. Neurosci.* *20*, 1404-1413.

Laifenfeld, D., Klein, E., and Ben-Schachar, D. (2002). Norepinephrine alters the expression of genes involved in neuronal sprouting and differentiation: relevance for major depression and antidepressant mechanisms. *J. Neurochem.* *83*, 1054-1064.

Layman, W. S., McEwen, D. P., Beyer, L. A., Lalani, S. R., Fernbach, S. D., Oh, E., Swaroop, A., Hegg, C. C., Raphael, Y., Martens, J. R., et al. (2009). Defects in neural stem cell proliferation and olfaction in *Chd7* deficient mice indicate a mechanism for hyposmia in human CHARGE syndrome. *Hum. Mol. Genet.* *18*, 1909-1923.

Lee, M. T., Chen, C. H., Lee, C. S., Chen, C. C., Chong, M. Y., Ouyang, W. C., Chiu, N. Y., Chuo, L. J., Chen, C. Y., Tan, H. K., et al. (2011). Genome-wide association study of bipolar I disorder in the Han Chinese population. *Mol. Psychiatry* 16, 548-556.

Lee, L. J., Lo, F. S., and Erzurumlu, R. S. (2005). NMDA receptor-dependent regulation of axonal and dendritic branching. *J. Neurosci.* 25, 2304-2311.

Leivo, I., Jee, K. J., Heikinheimo, K., Laine, M., Ollila, J., Nagy, B., and Knuutila, S. (2005). Characterization of gene expression in major types of salivary gland carcinomas with epithelial differentiation. *Cancer Genet. Cytogenet.* 156, 104-113.

Leonard, J. L., and Farwell, A. P. (1997). Thyroid hormone-regulated actin polymerization in brain. *Thyroid* 7, 147-151.

Lepagnol-Bestel, A. M., Maussion, G., Boda, B., Cardona, A., Iwayama, Y., Delezoide, A. L., Moalic, J. M., Muller, D., Dean, B., Yoshikawa, T., et al. (2008). SLC25A12 expression is associated with neurite outgrowth and is upregulated in the prefrontal cortex of autistic subjects. *Mol. Psychiatry* 13, 385-397.

Li, D., Gu, X., Lu, L., and Liang, L. (2010). Effects of phenylalanine on the survival and neurite outgrowth of rat cortical neurons in primary cultures: possible involvement of brain-derived neurotrophic factor. *Mol. Cell Biochem.* 339, 1-7.

Li, C., Ito, H., Fujita, K., Shiwaku, H., Qi, Y., Tagawa, K., Tamura, T., and Okazawa, H. (2013). Sox2 transcriptionally regulates PQBP1, an intellectual disability-microcephaly causative gene, in neural stem progenitor cells. *PLoS One* 8, e68627.

Li, J. M., Mogi, M., Tsukuda, K., Tomochika, H., Iwanami, J., Min, L. J., Nahmias, C., Iwai, and M., Horiuchi, M. (2007). Angiotensin II-induced neural differentiation via angiotensin II type 2 (AT2) receptor-MMS2 cascade involving interaction between AT2 receptor-interacting protein and Src homology 2 domain-containing protein-tyrosine phosphatase 1. *Mol. Endocrinol.* 21, 499-511.

Li, H., Radford, J. C., Ragusa, M. J., Shea, K. L., McKercher, S. R., Zaremba J. D., Soussou, W., Nie, Z., Kang, Y. J., Nakanishi, N., et al. (2008). Transcription factor MEF2C influences neural stem/progenitor cell differentiation and maturation in vivo. *Proc. Natl. Acad. Sci. USA* 105, 9397-9402.

Liss, L., and Grümer, H. D. (1966). Effect of L-phenylalanine on central nervous system elements in tissue culture. *J. Neurol. Neurosurg. Psychiatry* 29, 371-374.

Lu, T. Chen, R., Cox, T. C., Moldrich, R. X., Kurniawan, N., Tan, G., Perry, J. K., Ashworth, A., Barlett, P. F., Xu, L., et al. (2013). X-linked microtubule-associated protein, Mid1, regulates axon development. *Proc. Natl. Acad. Sci. USA* 110, 19131-19136.

Luo, Y., Shan, G., Gui, W., Smrt, R. D., Johnson, E. B., Li, X., Pfeiffer, R. L., Szulwach, K. E., Duan, R., Barkho, B. Z., et al. (2010). Fragile X mental retardation protein regulates proliferation and differentiation of adult neural stem/progenitor cells. *PLoS Genet.* 6, e1000898.

Ma, D. Q., Rabionet, R., Konidari, I., Jaworski, J., Cukier, H. N., Wright, H. H., Abramson, R. K., Gilbert, J. R., Cuccaro, M. L., Pericak-Vance, M. A., et al. (2010). Association and gene-gene interaction of SLC6A4 and ITGB3 in autism. *Am J. Med. Genet. B Neuropsychiatr. Genet.* 153B, 477-483.

Maddodi, N., Bhat, K. M., Devi, S., Zhang, S. C., and Setaluri, V. (2010). Oncogenic BRAFV600E induces expression of neuronal differentiation marker MAP2 in melanoma cells by promoter demethylation and down-regulation of transcription repressor HES1. *J. Biol. Chem.* 285, 242-254.

Maestrini, E., Pagnamenta, A. T., Lamb, J. A., Bacchelli, E., Sykes, N. H., Sousa, I., Toma, C., Barnby, G., Butler, H., Winchester, L., et al. (2010). High-density SNP association study and copy number variation analysis of the AUTS1 and AUTS5 loci implicate the IMMP2L-DOCK4 gene region in autism susceptibility. *Mol. Psychiatry* 15, 954-968.

Magome, T., Hattori, T., Taniguchi, M., Ishikawa, T., Miyata, S., Yamada, K., Takamura, H., Matsuzaki, Ito, A., Tohyama, M., et al. (2013). XLMR protein related to neurite extension (Xpn/KIAA2022) regulates cell-cell and cell-matrix adhesion and migration. *Neurochem. Int.* 63, 561-569.

Mah, W., Ko, J., Nam, J., Han, K., Chung, W. S., and Kim, E. (2010). Selected SALM (synaptic adhesion-like molecule) family proteins regulate synapse formation. *J. Neurosci.* 30, 5559-5568.

Maina, F., Hilton, M. C., Ponzetto, C., Davies, A. M., and Klein, R. (1997). Met receptor signaling is required for sensory nerve development and HGF promotes axonal growth and survival of sensory neurons. *Genes Dev.* 11, 3341-3350.

Mao, Y., Ge, X., Frank, C. L., Madison, J. M., Koehler, A. N., Doud, M. K., Tassa, C., Berry, E. M., Soda, T., Singh, K. K., et al. (2009). Disrupted in schizophrenia 1 regulates neuronal progenitor proliferation via modulation of GSK3beta/beta-catenin signaling. *Cell* 136, 1017-1031.

Mardirossian, S., Rampon, C., Salvert, D., Fort, P., and Sarda, N. (2009). Impaired hippocampal plasticity and altered neurogenesis in adult *Ube3a* maternal deficient mouse model for Angelman syndrome. *Exp. Neurol.* 220, 341-348.

Maric, D., Fiorio Pla, A., Chang, Y. H., and Barker, J. L. (2007). Self-renewing and differentiating properties of cortical neural stem cells are selectively regulated by basic

fibroblast growth factor (FGF) signaling via specific FGF receptors. *J. Neurosci.* 27, 1836-1852.

Maric, D., Liu, Q. Y., Grant, G. M., Andreadis, J. D., Hu, Q., Chang, Y. H., Barker, J. L., Joseph, J., Stenger, D. A., and Ma, W. (2000). Functional ionotropic glutamate receptors emerge during terminal cell division and early neuronal differentiation of rat neuroepithelial cells. *J. Neurosci. Res.* 61, 652-662.

Marteyn, A., Maury, Y., Gauthier, M. M., Lecuyer, C., Vernet, R., Denis, J. A., Pietu, G., Peschanski, M., and Martinat, C. (2011). Mutant human embryonic stem cells reveal neurite and synapse formation defects in type 1 myotonic dystrophy. *Cell Stem Cell* 8, 434-444.

Martinez-Ceballos, E., and Gudas, L. J. (2008). Hoxa1 is required for the retinoic acid-induced differentiation of embryonic stem cells into neurons. *J. Neurosci. Res.* 86, 2809-2819.

Martinez de Lagran, M., Benavides-Piccione, R., Ballesteros-Yañez, I., Calvo, M., Morales, M., Fillat, C., Defelipe, J., Ramakers, G. J., and Dierssen, M. (2012). Dyrk1A influences neuronal morphogenesis through regulation of cytoskeletal dynamics in mammalian cortical neurons. *Cereb. Cortex* 22, 2867-2877.

Maul, B., von Bohlen und Halbach, O., Becker, A., Sterner-Kock, A., Voigt, J. P., Siems, W. E., Grecksch, G., and Walther, T. (2008). Impaired spatial memory and altered dendritic spine morphology in angiotensin II receptor-deficient mice. *J. Mol. Med. (Berl)*. 86, 563-571.

McNeill, B., Mazerolle, C., Bassett, E. A., Mears, A. J., Ringuette, R., Lagali, P., Picketts, D. J., Paes, K., Rice, D., and Wallace, V. A. (2013). Hedgehog regulates Norrie disease protein to drive neural progenitor self-renewal. *Hum. Mol. Genet.* 22, 1005-1016.

Meikle, L., Talos, D. M., Onda, H., Pollizzi, K., Rotenberg, A., Sahin, M., Jensen, F. E., and Kwiatkowski, D. J. (2007). A mouse model of tuberous sclerosis: neuronal loss of Tsc1 causes dysplastic and ectopic neurons, reduced myelination, seizure activity, and limited survival. *J. Neurosci.* 27, 5546-5558.

Melicharek, D. J., Ramirez, L. C., Singh, S., Thompson, R., and Marenda, D. R. (2010). Kismet/CHD7 regulates axon morphology, memory and locomotion in *Drosophila* model of CHARGE syndrome. *Hum. Mol. Genet.* 19, 4253-4264.

Melke, J., Goubrain Botros, H., Chaste, P., Betancur, C., Nygren, G., Anckarsäter, H., Rastam, M., Ståhlberg, O., Gilberg, I. C., Delorme, R., et al. (2008). Abnormal melatonin synthesis in autism spectrum disorders. *Mol. Psychiatry* 13, 90-98.

Meloni, I., Parri, V., De Filippis, R., Ariani, F., Artuso, R., Bruttini, M., Katzaki, E., Longo, I., Mari, F., Bellan, C., et al. (2009). The XLMR gene ACSL4 plays a role in dendritic spine architecture. *Neuroscience* 159, 657-669.

Mercati, O., Danckaert, A., André-Leroux, G., Bellinzoni, M., Gouder, L., Watanade, K., Shimoda, Y., Grailhe, R., De Chaumont, F., Bourgeron, T., et al. (2013). *Contactin 4, -5 and -6 differentially regulate neuritogenesis while they display identical PTPRG binding sites.* *Biol. Open* 2, 324-334.

Métrailler, S., Emery, M., Schorderet, D. F., Cottet, S., and Roduit, R. (2013). ERK1/2 pathway is activated in degenerated Rpe65-deficient mice. *Exp. Eye Res.* 116, 86-95.

Mikulak, J., Negrini, S., Klajn, A., D'Alessandro, R., Mavilio, D., and Meldolesi, J. (2012). Dual REST-dependence of L1CAM: from gene expression to alternative splicing governed by Nova2 in neural cells. *J. Neurochem.* 120, 699-709.

Mills, P. B., Footitt, E. J., Mills, K. A., Tuschl, K., Aylett, S., Varadkar, S., Hemingway, C., Marlow, N., Rennie, J., Baxter, P., et al. (2010). Genotypic and phenotypic spectrum of pyridoxine-dependent epilepsy (ALDH7A1 deficiency). *Brain* 133, 2148-2159.

Mishra, A., Godavarthi, S. K., and Jana, N. R. (2009). *UBE3A/E6-AP regulates cell proliferation by promoting proteasomal degradation of p27.* *Neurobiol. Dis.* 36, 26-34.

Mitsui, S., Hidaka, C., Furihata, M., Osako, Y., and Yuri, K. (2013). A mental retardation gene, motopsin/prss12, modulates cell morphology by interaction with seizure-related gene 6. *Biochem. Biophys. Res. Commun.* 436, 638-644.

Mohapel, P., Leanza, G., Kokaia, M., and Lindvall, O. (2005). Forebrain acetylcholine regulates adult hippocampal neurogenesis and learning. *Neurobiol. Aging* 26, 939-946.

Montpetit, A., Côté, S., Brustein, E., Drouin, C. A., Lapointe, L., Boudreau, M., Meloche, C., Drouin, R., Hudson, T. J., Drapeau, P., et al. (2008). Disruption of AP1S1, causing a novel neurocutaneous syndrome, perturbs development of the skin and spinal cord. *PLoS Genet.* 4, e10000296.

Moosmang, S., Haider, N., Klugbauer, N., Adelsberger, H., Langweiser, N., Müller, J., Stiess, M., Marais, E., Schulla, V., Lacinova, L., et al. (2005). Role of hippocampal Cav1.2 Ca²⁺ channels in NMDA receptor-independent synaptic plasticity and spatial memory. *J. Neurosci.* 25, 9883-9892.

Moreb, J. S. (2008). Aldehyde dehydrogenase as a marker for stem cells. *Curr. Stem Cell Res. T.* 3, 237-246.

Moro, F., Pisano, T., Bernardina, B. D., Polli, R., Murgia, A., Zoccante, L., Darra, F., Battaglia, A., Pramparo, T., Zuffardi, O., et al. (2006). Periventricular heterotopia in fragile X syndrome. *Neurology* 67, 713-715.

Moroni, R. F., De Biasi, S., Colapietro, P., Larizza, L., and Beghini, A. (2006). Distinct expression pattern of microtubule-associated protein/microtubule affinity-regulating kinase 4 in differentiated neurons. *Neuroscience* *143*, 83-94.

Motazacker, M. M., Rost, B. R., Hucho, T., Garshasbi, M., Kahrizi, K., Ullman, R., Abedini, S. S., Nieh, S. E., Amini, S. H., Goswami, C., et al. (2007). A defect in the ionotropic glutamate receptor 6 gene (GRIK2) is associated with autosomal recessive mental retardation. *Am. J. Hum. Genet.* *81*, 792-798.

Mnayer, L., Khuri, S., Merheby, H. A., Meroni, G., and Elsas, L. J. (2006). A structure-function study of MID1 mutations associated with a mild Opitz phenotype. *Mol. Genet. Metab.* *8*, 198-203.

Najm, J., Horn, D., Wimplinger, I., Golden, J. A., Chizhikov, V. V., Sudi, J., Christian, S. L., Ullmann, R., Kuechler, A., Haas, C. A., et al. (2008). Mutations of CASK cause an X-linked brain malformation phenotype with microcephaly and hypoplasia of the brainstem and cerebellum. *Nat. Genet.* *40*, 1065-1067.

Napoli, I., Mercaldo, V., Boyl, P. P., Eleuteri, B., Zalfa, F., De Rubeis, S., Di Marino, D., Mohr, E., Massimi, M., Falconi, M., et al. (2008). The fragile X syndrome protein represses activity-dependent translation through CYFIP1, a new 4E-BP. *Cell* *134*, 1042-1054.

NCBI. (2014). PRKCB protein kinase C, beta [*Homo sapiens* (human)]. Accessed on 8/28/2014 from <http://www.ncbi.nlm.nih.gov/gene/5579>

Ng, A., Uribe, R. A., Yieh, L., Nuckels, R., and Gross, J. M. (2009). Zebrafish mutations in gart and paics identify crucial roles for de novo purine synthesis in vertebrate pigmentation and ocular development. *Development* *136*, 2601-2611.

Nichols, A. J., and Olson, E. C. (2010). Reelin promotes neuronal orientation and dendritogenesis during preplate splitting. *Cereb. Cortex* *20*, 2213-2223.

Nie, D., Di Nardo, A., Han, J. M., Baharanyi, H., Kramvis, I., Huynh, T., Dabora, S., Codeluppi, S., Pandolfi, P. P., Pasquale, E. B., et al. (2010). *Nat. Neurosci.* *13*, 163-172.

Nishi, T., Lee, P. S., Oka, K., Levin, V. A., Tanase, S., Morino, Y., and Saya, H. (1991). Differential expression of two types of the neurofibromatosis type 1 (NF1) gene transcripts related to neuronal differentiation. *Oncogene* *6*, 1555-1559.

Nishiyama, M., Skoultchi, A. I., and Nakayama, K. I. (2012). Histone H1 recruitment by CHD8 is essential for suppression of the Wnt— β -catenin signaling pathway. *Mol. Cell. Biol.* *32*, 501-512.

Nodé-Langlois, R., Muller, D., and Boda, B. (2006). Sequential implication of the mental retardation proteins ARHGEF6 and PAK3 in spine morphogenesis. *J. Cell Sci.* *119*, 4986-4993.

Nurcombe, V., Ford, M. D., Wildschut, J. A., and Barlett, P. F. (1993). Developmental regulation of neural responses to FGF-1 and FGF-2 by heparin sulfate proteoglycan. *Science* *260*, 103-106.

Nussbaum, J., Xu, Q., Payne, T. J., Ma, J. Z., Huang, W., Gelernter, J., and Li, M. D. (2008). Significant association of the neurexin-1 gene (NRXN1) with nicotine dependence in European- and African-American smokers. *Hum. Mol. Genet.* *17*, 1569-1577.

Okamura, Y., and Shidara, M. (1990). Changes in sodium channels during neural differentiation in the isolated blastomere of the ascidian embryo. *J. Physiol.* *431*, 39-74.

Oksenberg, N., Stevison, L., Wall, J. D., and Ahituv, N. (2013). Function and regulation of AUTS2, a gene implicated in autism and human evolution. *PLoS Genet.* *9*, e1003221.

Olbrich, H., Häffner, K., Kispert, A., Völkel, A., Volz, A., Sasmaz, G., Reinhardt, R., Hennig, S., Lehrach, H., Konietzko, N., et al. (2002). Mutations in DNAH5 cause primary ciliary dyskinesia and randomization of left-right asymmetry. *Nat. Genet.* *30*, 143-144.

Otero, J. J., Fu, W., Kan, L., Cuadra, A. E., and Kessler, J. A. (2004). Beta-catenin signaling is required for neural differentiation of embryonic stem cells. *Development* *131*, 3545-3557.

Ouyang, Q., Lizarraga, S. B., Schmidt, M., Yang, U., Gong, J., Ellisor, D., Kauer, J. A., and Morrow, E. M. (2013). Christianson syndrome protein NHE6 modulates TrkB endosomal signaling required for neuronal circuit development. *Neuron* *80*, 97-112.

Pagani, J. H., Zhao, M., Cui, Z., Williams Avram, S. K., Caruana, D. A., Dudek, S. M., and Young, W. S. (2014). Role of the vasopressin 1b receptor in rodent aggressive behavior and synaptic plasticity in hippocampal area CA2. *Mol. Psychiatry*, doi: 10.1038/mp.2014.47.

Page, D. T., Kutti, O. J., Prestia, C., and Sur, M. (2009). Haploinsufficiency for Pten and serotonin transporter cooperatively influences brain size and social behavior. *Proc. Natl. Acad. Sci. USA* *106*, 1989-1994.

Paglini, G., Kunda, P., Quiroga, S., Kosik, K., and Cáceres, A. (1998). Suppression of radixin and moesin alters growth cone morphology, motility, and process formation in primary cultured neurons. *J. Cell Biol.* *143*, 443-455.

Palmesino, E., Rousso, D. L., Kao, T. J., Klar, A., Laufer, E., Uemura, O., Okamoto, H., Novitch, B. G., and Kania, A. (2010). Foxp1 and Ihx1 coordinate motor neuron migration with axon trajectory choice by gating Reeling signaling. *PLoS Biol.* 8, e1000446.

Pang, L., Sawada, T., Decker, S. J., and Saltiel, A. R. (1995). Inhibition of MAP kinase kinase blocks the differentiation of PC-12 cells induced by nerve growth factor. *J. Biol. Chem.* 270, 13585-13588.

Paonessa, F., Latifi, S., Scarongella, H., Cesca, F., and Benfenati, F. (2013). Specificity protein 1 (Sp1)-dependent activation of the synapsin I gene (SYN1) is modulated by RE1-silencing transcription factor (REST) and 5'-cytosine-phosphoguanine (CpG) methylation. *J. Biol. Chem.* 288, 3227-3239.

Paquin, A., Hordo, C., Kaplan, D. R., and Miller, F. D. (2009). Costello syndrome H-Ras alleles regulate cortical development. *Dev. Biol.* 330, 440-451.

Park, T. S., and Zambidis, E. T. (2009). A role for the renin-angiotensin system in hematopoiesis. *Haematologica* 94, 745-747.

Paşca, S. P., Portmann, T., Voineagu, I., Yazawa, M., Shcheglovitov, A., Paşca, A. M., Cord, B., Palmer, T. D., Chikahisa, S., Nishino, S., et al. (2011). Using iPSC-derived neurons to uncover cellular phenotypes associated with Timothy syndrome. *Nat. Med.* 17, 1657-1662.

Pascual, M., and Guerri, C. (2007). The peptide NAP promotes neuronal growth and differentiation through extracellular signal-regulated protein kinase and Akt pathways, and protects neurons co-cultured with astrocytes damaged by ethanol. *J. Neurochem.* 103, 557-568.

Pasteris, N. G., and Gorski, J. L. (1999). Isolation, characterization, and mapping of the mouse and human Fgd2 genes, faciogenital dysplasia (FGD1; Aarskog syndrome) gene homologues. *Genomics* 60, 57-66.

Patrakitkomjom, S., Kobayashi, D., Morikawa, T., Wilson, M. M., Tsubota, N., Irie, A., Ozawa, T., Aoki, M., Arimura, N., Kaibuchi, K., et al. (2008). Neurofibromatosis type 1 (NF1) tumor suppressor, neurofibromin, regulates the neuronal differentiation of PC12 cells via its associating protein, CRMP-2. *J. Biol. Chem.* 283, 9399-9413.

Pawlisz, A. S., Mutch, C., Wynshaw-Boris, A., Chenn, A., Walsh, C. A., and Feng, Y. (2008). Lis1-Nde1-dependent neuronal fate control determines cerebral cortical size and lamination. *Hum. Mol. Genet.* 17, 2441-2455.

Peddada, S., Yasui, D. H., and LaSalle, J. M. (2006). Inhibitors of differentiation (ID1, ID2, ID3 and ID4) genes are neuronal targets of MeCP2 that are elevated in Rett syndrome. *Hum. Mol. Genet.* 15, 2003-2014.

Peñagarikano, O., Abrahams, B. S., Herman, E. I., Winden, K. D., Gdalyahu, A., Dong, H., Sonnenblick, L. I., Gruver, R., Almajano, J., Bragin, A., et al. (2011). Absence of CNTNAP2 leads to epilepsy, neuronal migration abnormalities, and core autism-related deficits. *Cell* 147, 235-246.

Penzes, P., Woolfrey, K. M., and Srivastava, D. P. (2011). Epac2-mediated dendritic spine remodeling: implications for disease. *Mol. Cell. Neurosci.* 46, 368-380.

Pfeiffer, V., Götz, R., Xiang, C., Camarero, G., Braun, A., Zhang, Y., Blum, R., Heinsen, H., Nieswandt, B., and Rapp, U. R. (2013). Ablation of BRAF impairs neuronal differentiation in the postnatal hippocampus and cerebellum. *PLoS One* 8, e58259.

Pietri, T., Easley-Neal, C., Wilson, C., and Washbourne, P. (2008). Six cadm/SynCAM genes are expressed in the nervous system of developing zebrafish. *Dev. Dyn.* 237, 233-246.

Pincus, D. W., Dicicco-Bloom, E. M., and Black, I. B. (1990). Vasoactive intestinal peptide regulates mitosis, differentiation and survival of cultured sympathetic neuroblasts. *Nature* 343, 564-567.

Pindolia, K., Chen, J., Cardwell, C., Cui, X., Chopp, M., and Wolf, B. (2012). Neurological deficits in mice with profound biotinidase deficiency are associated with demyelination and axonal degeneration. *Neurobiol. Dis.* 47, 428-435.

Pistocchi, A., Fazio, G., Cereda, A., Ferrari, L., Bettini, L. R., Messina, G., Cotelli, F., Biondi, A., Selicorni, A., and Massa, V. (2013). Cornelia de Lange syndrome: NIPBL haploinsufficiency downregulates canonical Wnt pathway in zebrafish embryos and patients' fibroblasts. *Cell Death Dis.* 4, e866.

Poeta, L., Fusco, F., Drongitis, D., Shoubridge, C., Manganelli, G., Filosa, S., Paciolla, M., Courtney, M., Collombat, P., Lioi, M. B., et al. (2013). A regulatory path associated with X-linked intellectual disability and epilepsy links KDM5C to the polyalanine expansions in ARX. *Am. J. Hum. Genet.* 92, 114-125.

Poirier, K., Keays, D. A., Francis, F., Saillour, Y., Bahi, N., Manouvrier, S., Fallet-Bianco, C., Pasquier, L., Toutain, A., Tuy, F. P., et al. (2007). Large spectrum of lissencephaly and pachygyria phenotypes resulting from de novo missense mutations in tubulin alpha 1A (TUBA1A). *Hum. Mutat.* 28, 1055-1064.

Polleux, F., Ince-Dunn, G., and Ghosh, A. (2007). Transcriptional regulation of vertebrate axon guidance and synapse formation. *Nat. Rev. Neurosci.* 8, 331-340.

Qi, J., Chi, L., Labeit, S., and Banes, A. J. (2008). *Am. J. Physiol. Cell. Physiol.* 295, C975-985.

Qiu, M. S., and Green, S. H. (1992). PC12 cell neuronal differentiation is associated with prolonged p21ras activity and consequent prolonged ERK activity. *Neuron* 9, 705-717.

Qiu, J., Shi, G., Jia, Y., Li, J., Wu, M., Li, J., Dong, S., and Wong, J. (2010). The X-linked mental retardation gene PHF8 is a histone demethylase involved in neuronal differentiation. *Cell Res.* 20, 908-918.

Qureshi, K., Rao, K. V., and Qureshi, I. A. (1998). Differential inhibition by hyperammonemia of the electron transport chain enzymes in synaptosomes and non-synaptic mitochondria in ornithine transcarbamylase-deficient spf-mice: restoration by acetyl-L-carnitine. *Neurochem. Res.* 23, 855-861.

Raabe, E. H., Lim, K. S., Kim, J. M., Meeker, A., Mao, X. G., Nikkhah, G., Maciaczyk, J., Kahlert, U., Jain, D., Bar, E., et al. (2011). BRAF activation induces transformation and then senescence in human neural stem cells: a pilocytic astrocytoma model. *Clin. Cancer Res.* 17, 3590-3599.

Raft, S., Nowotschin, S., Liao, J., and Morrow, B. E. (2004). Suppression of neural fate and control of inner ear morphogenesis by Tbx1. *Development* 131, 1801-1812.

Ramirez, I. B., Pietka, G., Jones, D. R., Divecha, N., Alia, A., Baraban, S. C., Hurlstone, A. F., and Lowe, M. (2012). Impaired neural development in a zebrafish model for Lowe syndrome. *Hum. Mol. Genet.* 21, 1744-1759.

Reid, C. A., Leaw, B., Richards, K. L., Richardson, R., Wimmer, V., Yu, C., Hill-Yardin, E. L., Lerche, H., Scheffer, I. E., et al. (2014). Reduced dendritic arborization and hyperexcitability of pyramidal neurons in a Scn1b-based model of Dravet syndrome. *Brain* 137, 1701-1715.

Richter, C., and Kass, G. E. (1991). Oxidative stress in mitochondria: its relationship to cellular Ca²⁺ homeostasis, cell death, proliferation, and differentiation. *Chem. Bio. Interact.* 77, 1-23.

Rocas, D., Alix, E., Michel, J., Cordier, M. P., Labalme, A., Guilbert, H., Till, M., Schluth-Bolard, C., de Haas, P., Massardiet, J., Portes, V. D., et al. (2013). Neuropathological features in a female fetus with OPHN1 deletion and cerebellar hypoplasia. *Eur. J. Med. Genet.* 56, 270-273.

Rolland, S. G., and Conradt, B. (2010). New role of the BCL2 family of proteins in the regulation of mitochondrial dynamics. *Curr. Opin. Biol.* 852, 852-858.

Rooslid, T., Castronovo, S., Villosio, A., Ziemba, A., and Pizzorno, G. (2011). A novel structural mechanism for redox regulation of uridine phosphorylase 2 activity. *J. Struct. Biol.* 176, 229-237.

- Rosário, M., Franke, R., Bednarski, C., and Birchmeier, W. (2007). The neurite outgrowth multiadaptor RhoGAP, NOMA-GAP, regulates neurite extension through SHP2 and Cdc42. *J. Cell Biol.* *178*, 503-516.
- Rosenfelder, L. M. (2009). Effects of silencing the mental retardation gene *Jarid1c* on neuronal differentiation of pluripotent P19 cells. PhD diss., Worcester Polytechnic Institute.
- Rouaux, C., and Arlotta, P. (2010). *Fezf2* directs the differentiation of corticofugal neurons from striatal progenitors in vivo. *Nat. Neurosci.* *13*, 1345-1347.
- Rouso, D. L., Pearson, C. A., Gaber, Z. B., Miquelajauregui, A., Li, S., Portera-Cailliau, C., Morrissey, E. E., and Novitch, B. G. (2012). Foxp-mediated suppression of N-cadherin regulates neuroepithelial character and progenitor maintenance in the CNS. *Neuron* *74*, 314-330.
- Rowe, S. J., Messenger, N. J., and Warner, A. E. (1993). The role of noradrenaline in the differentiation of amphibian embryonic neurons. *Development* *119*, 1343-1357.
- Sakai, T., Furuyama, T., Ohoka, Y., Miyazaki, N., Fujioka, S., Sugimoto, H., Amasaki, M., Hattori, S., and Matsuya, T. (1999). Mouse semaphoring H induces PC12 cell neurite outgrowth activating Ras-mitogen-activated protein kinase signaling pathway via Ca(2+) influx. *J. Biol. Chem.* *274*, 29666-29671.
- Sala, M., Braidà, D., Lentini, D., Busnelli, M., Bulgheroni, E., Capurro, V., Finardi, A., Donzelli, A., Pattini, L., Rubino, T., et al. (2011). *Biol. Psychiatry* *69*, 875-882.
- Salim, K., Kehoe, L., Minkoff, M. S., Bilsland, J. G., Munoz-Sanjuan, I., and Guest, P. C. (2006). Identification of differentiating neural progenitor cell markers using shotgun isobaric tagging mass spectrometry. *Stem Cells Dev.* *15*, 461-470.
- Sarnat, H. B., Benjamin, D. R., Siebert, J. R., Kletter, G. B., and Cheyette, S. R. (2002). Agenesis of the mesencephalon and metencephalon with cerebellar hypoplasia: putative mutation in the *EN2* gene—report of 2 cases in early infancy. *Pediatr. Dev. Pathol.* *5*, 54-68.
- Satin, C., and Kitajima, K. (2013). Impact of structural aberrancy of polysialic acid and its synthetic enzyme ST8SIA2 in schizophrenia. *Front. Cell Neurosci.* *7*, 61.
- Sautin, Y. Y., Nakagawa, T., Zharikov, S., and Johnson, R. J. (2007). Adverse effects of the classic antioxidant uric acid in adipocytes: NADPH oxidase-mediated oxidative/nitrosative stress. *Am. J. Physiol. Cell Physiol.* *293*, C584-596.
- Savarese, F., Dávila, A., Nechanitzky, R., De La Rossa-Velazquez, I., Pereira, C. F., Engelka, R., Takahashi, K., Jenuwein, T., Kohwi-Shigematsu, T., Fisher, A. G., et al.

(2009). *Satb1* and *Satb2* regulate embryonic stem cell differentiation and *Nanog* expression. *Genes Dev.* *23*, 2625-2638.

Schaarschmidt, G., Wegner, F., Schwarz, S. C., Schmidt, H., and Schwarz, J. (2009). Characterization of voltage-gated potassium channels in human neural progenitor cells. *PLoS One* *4*, e6168.

Schäfer, M. K., and Frotscher, M. (2012). Role of L1CAM for axon sprouting and branching. *Cell Tissue Res.* *349*, 39-48.

Schlosser, G., Koyano-Nakagawa, N., and Kintner, C. (2002). Thyroid hormone promotes neurogenesis in the *Xenopus* spinal cord. *Dev. Dyn.* *225*, 485-498.

Schmeisser, M. J., Baumann, B., Johannsen, S., Vindedal, G. F., Jensen, V., Hvalby, Ø. C., Sprengel, R., Seither, J., Magbool, A., Magnutzki, A., et al. (2012). *IκB* kinase/nuclear factor κ B-dependent insulin-like growth factor 2 (*Igf2*) expression regulates synapse formation and spine maturation via *Igf2* receptor signaling. *J. Neurosci.* *32*, 5688-5703.

Schneider, R. A., Hu, D., Rubenstein, J. L. R., Maden, M., and Helms, J. A. (2001). Local retinoid signaling coordinates forebrain and facial morphogenesis by maintaining *FGF8* and *SHH*. *Development* *128*, 2755-2767.

Schuetz, G., Rosário, M., Grimm, J., Boeckers, T. M., Gundelfinger, E. D., and Birchmeier, W. (2004). The neuronal scaffold protein *Shank3* mediates signaling and biological function of the receptor tyrosine kinase *Ret* in epithelial cells. *J. Cell. Biol.* *167*, 945-952.

Schüller, A. C., Ahmed, Z., and Ladbury, J. E. (2008). Extracellular point mutations in *FGFR2* result in elevated *ERK1/2* activation and perturbation of neuronal differentiation. *Biochem. J.* *410*, 205-211.

Schulz, P. E., McIntosh, A. D., Kasten, M. R., Wieringa, B., and Epstein, H. F. (2003). A role for myotonic dystrophy protein kinase in synaptic plasticity. *J. Neurophysiol.* *89*, 1177-1186.

Schwamborn, J. C., Fiore, R., Bagnard, D., Kappler, J., Kalschmidt, C., and Püschel, A. W. (2004). Semaphorin 3A stimulates neurite extension and regulates gene expression in PC12 cells. *J. Biol. Chem.* *279*, 30923-30926.

Seipel, K., Medley, Q. G., Kedersha, N. L., Zhang, X. A., O'Brien, S. P., Serra-Pages, C., Hemler, M. E., and Streuli, M. (1999). Trio amino-terminal guanine nucleotide exchange factor domain expression promotes actin cytoskeleton reorganization, cell migration and anchorage-independent cell growth. *J. Cell. Sci.* *112*, 1825-1834.

- Senocak, E. U., Oğuz, K. K., Haliloğlu, G., Topçu, M., and Cila, A. (2010). Structural abnormalities of the brain other than molar tooth sign in Joubert syndrome-related disorders. *Diagn. Interv. Radiol.* 16, 3-6.
- Seranski, P., Hoff, C., Radelof, U., Hennig, S., Reinhardt, R., Schwartz, C. E., Heiss, N. S., and Poustka, A. (2001). RAI1 is a novel polyglutamine encoding gene that is deleted in Smith-Magenis syndrome patients. *Gene* 270, 69-76.
- Sharifi, R., Morra, R., Appel, C. D., Tallis, M., Chioza, B., Jankevicius, G., Simpson, M. A., Matic, I., Ozkan, E., Golia, B., et al. (2013). Deficiency of terminal ADP-ribose protein glycohydrolase TARG1/C6orf130 in neurodegenerative disease. *EMBO J.* 32, 1225-1237.
- Sharma, N., Jadhav, S. P. and Bapat, S. A. (2010). CREBBP re-arrangements affect protein function and lead to aberrant neuronal differentiation. *Differentiation* 79, 218-231.
- Sharma, S., Koh, K. S., Collin, C., Dave, A., McMellon, A., Sugiyama, Y., McAvoy, J. W., Voss, A. K., Géczy, J., and Craig, J. E. (2009). NHS-A isoform of the NHS gene is a novel interactor of ZO-1. *Exp. Cell. Res.* 315, 2358-2372.
- Sheen, V. L., Torres, A. R., Du, X., Barry, B., Walsh, C. A., and Kimonis, V. E. (2010). Mutation in PQBP1 is associated with periventricular heterotopia. *Am. J. Med. Genet.* 152A, 2888-2889.
- Shen, Y., Inoue, N., and Heese, K. (2010). Neurotrophin-4 (ntf4) mediates neurogenesis in mouse embryonic neural stem cells through the inhibition of the signal transducer and activator of transcription-3 (stat3) and the modulation of the activity of protein kinase B. *Cell Mol. Neurobiol.* 30, 909-916.
- Shi, L., Chang, X., Zhang, P., Coda, M. P., Lu, W., and Wang, K. (2013). The functional genetic link of NLGN4X knockdown and neurodevelopment in neural stem cells. *Hum. Mol. Genet.* 22, 3749-3760.
- Shi, G.-X., Rehmann, H., and Andres, D. A. (2006). A novel cyclic AMP-dependent Epac-Rit signaling pathway contributes to PACAP38-mediated neuronal differentiation. *Mol. Cell. Biol.* 26, 9136-9147.
- Shimizu, T., Nakazawa, M., Kani, S., Bae, Y. K., Shimizu, T., Kageyama, R., and Hibi, M. (2010). Zinc finger genes Fezf1 and Fezf2 control neuronal differentiation by repressing Hes5 expression in the forebrain. *Development* 137, 1875-1885.
- Shinjyo, N., Ståhlberg, A., Dragunow, M., Pekny, M., and Pekna, M. (2009). Complement-derived anaphylatoxin C3a regulates in vitro differentiation and migration of neural progenitor cells. *Stem Cells* 27, 2824-2832.

Shioda, N., Beppu, H., Fukuda, T., Li, E., Kitjima, I., and Fukunaga, K. (2011). Aberrant calcium/calmodulin-dependent protein kinase II (CaMKII) activity is associated with abnormal dendritic spine morphology in the ATRX mutant mouse brain. *J. Neurosci.* *31*, 346-358.

Shmueli, O., Gdalyahu, A., Sorokina, K., Nevo, E., Avivi, A., and Reiner, O. (2001). *Hum. Mol. Genet.* *10*, 1061-1070.

Shoubridge, C., Walikonis, R. S., Gécz, J., and Harvey, R. J. (2010). Subtle functional defects in the Arf-specific guanine nucleotide exchange factor IQSEC2 cause non-syndromic X-linked intellectual disability. *Small GTPases* *1*, 98-103.

Silei, V., Politi, V., and Lauro, G. M. (2000). Uridine induces differentiation in human neuroblastoma cells via Protein Kinase C epsilon. *J. Neurosci. Res.* *61*, 206-211.

So, P.-L., Yip, P. K., Bunting, S., Wong, L.-F., Mazarakis, N. D., Hall, S., McMahon, S., Maden, M., and Corcoran, J. P. T. (2006). Interactions between retinoic acid, nerve growth factor and sonic hedgehog signaling in neurite outgrowth. *Dev. Biol.* *298*, 167-175.

Sobczak, M., Boczek, T., Kowalski, A., Wiktorska, M., Niewiarowska, J., and Zylinska, L. (2014). Downregulation of microsomal glutathione-S-transferase 1 modulates protective mechanisms in differentiated PC12 cells. *J. Physiol. Biochem.* *70*, 375-383.

Song, J. H., Wang, C. X., Song, D. K., Wang, P., Shuaib, A., and Hao, C. (2005). Interferon gamma induces neurite outgrowth by up-regulation of p35 neuron-specific cyclin-dependent kinase 5 activator via activation of ERK1/2 pathway. *J. Biol. Chem.* *280*, 12896-12901.

Soucek, T., Hölzl, G., Bernaschek, G., and Hengstschlager, M. (1998). A role of the tuberous sclerosis gene-2 product during neuronal differentiation. *Oncogene* *16*, 2197-2204.

Spassky, N., Han, Y. G., Aguilar, A., Strehl, L., Besse, L., Laclef, C., Ros, M. R., Garcia-Verdugo, J. M., and Alvarez-Buylla, A. (2008). Primary cilia are required for cerebellar development and Shh-dependent expansion of progenitor pool. *Dev. Biol.* *317*, 246-259.

Sprengel, R. (2006). Role of AMPA receptors in synaptic plasticity. *Cell Tissue Res.* *326*, 447-455.

Stornetta, R. L., and Zhu, J. J. (2011). Ras and Rap signaling in synaptic plasticity and mental disorders. *Neuroscient* *17*, 54-78.

Strauss, K. A., Puffenberger, E. G., Huentelman, M. J., Gottlieb, S., Dobrin, S. E., Parod, J. M., Stephan, D. A., and Morton, D. H. (2006). Recessive symptomatic focal epilepsy and mutant contactin-associated protein-like 2. *N. Engl. J. Med.* *354*, 1370-1377.

- Sun, P., Quan, Z., Zhang, B., Wu, T., and Xi, R. (2010). TSC1/2 tumour suppressor complex maintains *Drosophila* germline stem cells by preventing differentiation. *Development* *137*, 2461-2469.
- Sun, C., Warland, D. K., Ballesteros, J. M., van der List, D., and Chalupa, L. M. (2008). Retinal waves in mice lacking the beta2 subunit of the nicotinic acetylcholine receptor. *Proc. Natl. Acad. Sci. USA* *105*, 13638-13643.
- Suzuki, M., Nelson, A. D., Eickstaedt, J. B., Wallace, K., Wright, L. S., and Svendsen, C. N. (2006). Glutamate enhances proliferation and neurogenesis in human neural progenitor cell cultures derived from the fetal cortex. *Eur. J. Neurosci.* *24*, 645-653.
- Tabata, H., and Nakajima, K. (2002). Neurons tend to stop migration and differentiate along the cortical internal plexiform zones in the Reelin signal-deficient mice. *J. Neurosci. Res.* *69*, 723-730.
- Takeichi, M. (1991). Cadherin cell adhesion receptors as a morphogenetic regulator. *Science* *251*, 1451-1455.
- Talkowski, M. E., Mullegama, S. V., Rosenfeld, J. A., van Bon, B. W., Shen, Y., Repnikova, E. A., Gastier-Foster, J., Thrush, D. L., Kathiresan, S., Ruderfer, D. M. (2011). Assessment of 2q23.1 microdeletion syndrome implicates MBD5 as a single causal locus of intellectual disability, epilepsy, and autism spectrum disorder. *Am. J. Hum. Genet.* *89*, 551-563.
- Tang, Z. Z., Yarotsky, V., Wei, L., Sobczak, K., Nakamori, M., Eichinger, K., Moxley, R. T., Dirksen, R. T., and Thornton, C. A. (2012). Muscle weakness is myotonic dystrophy associated with misregulated splicing and altered gating of Ca(V)1.1 calcium channel. *Hum. Mol. Genet.* *21*, 1312-1324.
- Thambirajah, A. A., Ng, M. K., Frehlick, L. J., Li, A., Serpa, J. J., Petrotchenko, E. V., Silva-Moreno, B., Missiaen, K. K., Borchers, C. H., Adam Hall, J., et al. (2012). MeCP2 binds to nucleosome free (linker DNA) regions and to H3K9/H3K27 methylated nucleosomes in the brain. *Nucleic Acids Res.* *40*, 2884-2897.
- Thibault, O., Hadley, R., and Landfield, P. W. (2001). Elevated postsynaptic [Ca²⁺]_i and L-type calcium channel activity in aged hippocampal neurons: relationship to impaired synaptic plasticity. *J. Neurosci.* *21*, 9744-9756.
- Toyo-oka, K., Shionoya, A., Gambello, M. J., Cardoso, C., Leventer, R., Ward, H. L., Ayala, R., Tsai, L.-H., Dobyns, W., Ledbetter, D., et al. (2003). 14-3-3-ε is important for neuronal migration by binding to NUDEL: a molecular explanation for Miller-Dieker syndrome. *Nat. Genet.* *34*, 274-285.

Tozuka, Y., Fukuda, S., Namba, T., Seki, T., and Hisatsune, T. (2005). GABAergic excitation promotes neuronal differentiation in adult hippocampal progenitor cells. *Neuron* 47, 803-815.

Tsang, H. T., Edwards, T. L., Wang, X., Connell, J. W., Davies, R. J., Durrington, H. J., O’Kane, C. J., Luzio, J. P., and Reid, E. (2009). The hereditary spastic paraplegia proteins NIPA1, spastin and spartin are inhibitors of mammalian BMP signaling. *Hum. Mol. Genet.* 18, 3805-3821.

Tucker, B., Richards, R. I., and Lardelli, M. (2006). Contribution of mGluR and Fmr1 functional pathways to neurite morphogenesis, craniofacial development and fragile X syndrome. *Hum. Mol. Genet.* 15, 3446-3458.

Ueberham, U., and Arendt, T. (2013). The role of Smad proteins for development, differentiation and dedifferentiation of neurons. Accessed on 8/28/2014 from <http://www.intechopen.com/books/trends-in-cell-signaling-pathways-in-neuronal-fate-decision/the-role-of-smad-proteins-for-development-differentiation-and-dedifferentiation-of-neurons>

Uhrbom, L., Dai, C., Celestino, J. C., Rosenblum, M. K., Fuller, G. N., and Holland, E. C. (2002). Ink4a-Arf loss cooperates with KRas activation in astrocytes and neural progenitors to generate glioblastomas of various morphologies depending on activated Akt. *Cancer Res.* 62, 5551-5558.

Valenciano, A. I., Mayordomo, R., de La Rosa, E. J., and Hallböök, F. (2002). Biotin decreases retinal apoptosis and induces eye malformations in the early chick embryo. *Neuroreport* 13, 297-299.

Valli, E., Trazzi, S., Fuchs, C., Erriquez, D., Bartesaghi, R., Perini, G., and Ciani, E. (2012). CDKL5, a novel MYCN-repressed gene, blocks cell cycle and promotes differentiation of neuronal cells. *Biochim. Biophys. Acta* 1819, 1173-1185.

Vallipuram, J., Grenville, J., and Crawford, D. A. (2010). The E646D-ATP13A4 mutation associated with autism reveals a defect in calcium regulation. *Cell. Mol. Neurobiol.* 30, 233-246.

Valnegri, P., Montrasio, C., Brambilla, D., Ko, J., Passafaro, and Sala, C. (2011). The X-linked intellectual disability protein IL1RAPL1 regulates excitatory synapse formation by binding PTP δ and RhoGAP2. *Hum. Mol. Genet.* 20, 4797-4809.

Van Maldergem, L., Hou, Q., Kalscheuer, V. M., Rio, M., Doco-Fenzy, M., Medeira, A., de Brouwer, A. P., Cabrol, C., Haas, S. A., Cacciagli, P., et al. (2013). Loss of function of KIAA2022 causes mild to severe intellectual disability with an autism spectrum disorder and impairs neurite outgrowth. *Hum. Mol. Genet.* 22, 3306-3314.

- Vardya, I., Drasbek, K. R., Gibson, K. M., and Jensen, K. (2010). Plasticity of postsynaptic, but not presynaptic, GABA_B receptors in SSADH deficient mice. *Exp. Neurol.* 225, 114-122.
- Veldman, M. B., Bembien, M. A., and Goldman, D. (2010). Tuba1a gene expression is regulated by KLF6/7 and is necessary for CNS development and regeneration in zebrafish. *Mol. Cell. Neurosci.* 43, 370-383.
- Vernes, S. C., Oliver, P. L., Spiteri, E., Lockstone, H. E., Puliyadi, R., Taylor, J. M., Ho, J., Mombereau, C., Brewer, A., Lowy, E., et al. (2011). *PLoS Genet.* 7, e1002145.
- Verrotti, A., Spalice, A., Ursitti, F., Papetti, L., Mariani, R., Castronovo, A., Mastrangelo, M., and Iannetti, P. (2010). New trends in neuronal migration disorders. *Eur. J. Paediatr. Neurol.* 14, 1-12.
- Vogel-Ciernia, A., Matheos, D. P., Barrett, R. M., Kramár, E. A., Azzawi, S., Chen, Y., Magnan, C. N., Zeller, M., Sylvain, A., Haetig, J., et al. (2013). The neuron-specific chromatin regulatory subunit BAF53b is necessary for synaptic plasticity and memory. *Nat. Neurosci.* 16, 552-561. *Wiley Interdis. Rev. Membr. Transp. Sig.* 3, 1-13.
- Volk, C. (2014). OCTs, OATs, and OCTNs: structure and function of the polyspecific organic ion transporters of the SLC22 family.
- Völkel, P., and Angrand, P. O. (2007). The control of histone lysine methylation in epigenetic regulation. *Biochimie* 89, 1-20.
- Walton, N. M., de Koning, A., Xie, X., Shin, R., Chen, Q., Miyake, S., Tajinda, K., Gross, A. K., Kogan, J. H., Heusner, C. L., et al. (2014). Gastrin-releasing peptide contributes to the regulation of adult hippocampal neurogenesis and neuronal development. *Stem Cells* 32, 2454-2466.
- Wang, W., Crandall, J. E., Litwack, E. D., Gronostajski, R. M., and Kilpatrick, D. L. (2010). Targets of the nuclear factor I regulon involved in early and late development of postmitotic cerebellar granule neurons. *J. Neurosci. Res.* 88, 258-265.
- Wang, J., Leung, J. W., Gong, Z., Feng, L., Shi, X., and Chen, J. (2013a). PHF6 regulates cell cycle progression by suppressing ribosomal RNA synthesis. *J. Biol. Chem.* 288, 3174-3183.
- Wang, Q., Moore, M. J., Adelmant, G., Marto, J. A., and Silver, P. A. (2013b). PQBP1, a factor linked to intellectual disability, affects alternative splicing associated with neurite outgrowth. *Genes Dev.* 27, 615-626.
- Wang, P. Y., Seabold, G. K., and Wenthold, R. J. (2008). Synaptic adhesion-like molecules (SALMs) promote neurite outgrowth. *Mol. Cell. Neurosci.* 39, 83-94.

- Wang, X., Xu, Q., Bey, A. L., Lee, Y., and Jiang, Y. H. (2014). Transcriptional and functional complexity of Shank3 provides a molecular framework to understand the phenotypic heterogeneity of SHANK3 causing autism and Shank3 mutant mice. *Mol. Autism* 5, 30.
- Wang, X., Yang, N., Uno, E., Roeder, R. G., and Guo, S. (2006). A subunit of the mediator complex regulates vertebrate neuronal development. *Proc. Natl. Acad. Sci. USA* 103, 17284-17289.
- Watanabe, Y., Inoue, K., Okuyama-Yamamoto, A., Nakai, N., Nakatani, J., Nibu, K., Sato, N., Iiboshi, Y., Yusa, K., Kondoh, G., et al. (2009). Fezf1 is required for penetration of the basal lamina by olfactory axons to promote olfactory development. *J. Comp. Neurol.* 515, 565-584.
- Weimann, J. M., Zhang, Y. A., Levin, M. E., Devine, W. P., Brûlet, W. P., and McConnell, S. K. (1999). Cortical neurons require Oxt1 for the refinement of exuberant axonal projections to subcortical targets. *Neuron* 24, 819-831.
- Weng, L., Lin, Y. F., Li, A. L., Wang, C. E., Yan, S., Sun, M., Gaertig, M. A., Mitha, N., Kosaka, J., Wakabayashi, T., et al. (2013). Loss of Ahi1 affects early development by impairing BM88/Cend1-mediated neuronal differentiation. *J. Neurosci.* 33, 8172-8184.
- White, D. M., Walker, S., Brenneman, D. E., and Gozes, I. (2000). CREB contributes to the increased neurite outgrowth of sensory neurons induced by vasoactive intestinal polypeptide and activity-dependent neurotrophic factor. *Brain Res.* 868, 31-38.
- Whitehead, J., and Cobo, E. (2008). Stopping a trial early in oncology: for patients or for industry? *Ann. Oncol.* 19, 1512-1513.
- Whyte, A. J. (2014). Genetic and functional interactions between Itgb3 and Slc6a4 in mouse brain. Hosted by Vanderbilt University. Accessed on 8/27/2014 from <http://etd.library.vanderbilt.edu/available/etd-03212013-135948/>
- Williams, M. E., Wilke, S. A., Daggett, A., Davis, E., Otto, S., Ravi, D., Ripley, B., Bushong, E. A., Ellisman, M. H., Klein, G., et al. (2011). Cadherin-9 regulates synapse-specific differentiation in the developing hippocampus. *Neuron* 71, 640-655.
- Wittwer, F., van der Straten, A., Keleman, K., Dickson, B. J., and Hafen, E. (2001). Lilliputian: an AF4/FMR2-related protein that controls cell identity and cell growth. *Development* 128, 791-800.
- Wolfer, D. P., Lang, R., Cinelli, P., Madani, R., and Sonderegger, P. (2001). Multiple roles of neurotropsin in tissue morphogenesis and nervous system development suggested by the mRNA expression pattern. *Mol. Cell. Neurosci.* 18, 407-433.

Wu, P. R., Tsai, P. I., Chen, G. C., Chou, H. J., Huang, Y. P., Chen, Y. H., Lin, M. Y., Kimchi, A., Chien, C. T., and Chen, R. H. (2011). DAPK activates MARK1/2 to regulate microtubule assembly, neuronal differentiation, and tau toxicity. *Cell Death Differ.* *18*, 1507-1520.

Xiao, Y., Peng, Y., Wan, J., Tang, G., Chen, Y., Tang, J., Ye, W. C., Ip, N. Y., and Shi, L. (2013). The atypical guanine nucleotide exchange factor Dock4 regulates neurite differentiation through modulation of Rac1 GTPase and actin dynamics. *J. Biol. Chem.* *288*, 20034-20045.

Yabut, O., Domagauer, J., and D'Arcangelo, G. (2010). Dyrk1A overexpression inhibits proliferation and induces premature neuronal differentiation of neural progenitor cells. *J. Neurosci.* *30*, 4004-4014.

Yasin, S. A., Ali, A. M., Tata, M., Picker, S. R., Anderson, G. W., Latimer-Bowman, E., Nicholson, S. L., Harkness, W., Cross, J. H., Paine, S. M., et al. (2013). mTOR-dependent abnormalities in autophagy characterize human malformations of cortical development: evidence from focal cortical dysplasia and tuberous sclerosis. *Acta Neuropathol.* *126*, 207-218.

Yoon, C., Van Niekerk, E. A., Henry, K., Ishikawa, T., Orita, S., Tuszynski, M. H., and Campana, W. M. (2013). Low-density lipoprotein receptor-related protein 1 (LRP1)-dependent cell signaling promotes axonal regeneration. *J. Biol. Chem.* *288*, 26557-26568.

Yoshiga, D., Taketomi, T., Muratsu, D., Onimura, T., Shiraishi, H., Sansada, T., Kobayashi, T., and Nakamura, S. (2013). RNF135, a RING finger protein, negatively regulates RAS/MAPK signaling. IADR General Session. Accessed on 8/29/2014 from <https://iadr.confex.com/iadr/2010barce/webprogram/Paper136847.html>

Yoshihara, S., Omichi, K., Yanazawa, M., Kitamura, K., and Yoshihara, Y. (2005). *Arx* homeobox gene is essential for development of mouse olfactory system. *Development* *132*, 751-762.

Yu, I. T., Park, J. K., Kim, S. H., Lee, J. S., Kim, Y. S., and Son, H. (2009). Valproic acid promotes neuronal differentiation by induction of proneural factors in association with H4 acetylation. *Neuropharmacology* *56*, 472-480.

Yu, H., and Patel, S. B. (2005). Recent insights into the Smith-Lemli-Opitz syndrome. *Clin. Genet.* *68*, 383-391.

Yu, H., Wessels, A., Tint, G. S., and Patel, S. B. (2005). Partial rescue of neonatal lethality of *Dhcr7* null mice by a nestin promoter-driven DHCR7 transgene expression. *Brain Res. Dev. Brain Res.* *156*, 46-60.

Zeng, L., Zhang, P., Shi, L., Yamamoto, V., Lu, W., and Wang, K. (2013). Functional impacts of NRXN1 knockdown on neurodevelopment in stem cell models. *PLoS One* 8, e59685.

Zhang, Y., Chen, D., and Wang, Z. (2009). Analyses of mental dysfunction-related ACSL4 in drosophila reveal its requirement for Dpp/BMP production and visual wiring in the brain. *Hum. Mol. Genet.* 18, 3894-3905.

Zhang, C., Mejia, L. A., Huang, J., Valnegri, P., Bennett, E. J., Anckar, J., Jahani-Asl, A., Gallardo, G., Ikeuchi, Y., Yamada, T., et al. (2013). The X-linked intellectual disability protein PHF6 associates with the PAF1 complex and regulates neuronal migration in the mammalian brain. *Neuron* 78, 986-993.

Zhang, L., Song, N. N., Chen, J. Y., Huang, Y., Li, H., and Ding, Y. Q. (2012). Satb2 is required for dendritic arborization and soma spacing in mouse cerebral cortex. *Cereb. Cortex* 22, 1510-1519.

Zhang, K. Z., Westberg, J. A., Hölttä, E., and Andersson, L. C. (1996). BCL2 regulates neural differentiation. *Proc. Natl. Acad. Sci. USA* 93, 4504-4508.

Zhang, Y. Y., Yue, J., Che, H., Sun, H. Y., Tse, H. F., and Li, G. R. (2014). BKCa and hEag1 channels regulate cell proliferation and differentiation in human bone marrow-derived mesenchymal stem cells. *J. Cell Physiol.* 229, 202-212.

Zhang-James, Y., DasBanerjee, T., Sagvolden, T., Middleton, F. A., and Faraone S. V. (2011). SLC9A9 mutations, gene expression, and protein-protein interactions in rat models of attention-deficit/hyperactivity disorder. *Am. J. Med. Genet. B Neuropsychiatr. Genet.* 156B, 835-843.

Zhu, B., Chen, C., Xue, G., Moyzis, R. K., Dong, Q., Chen, C., He, Q., Lei, X., Wang, Y., et al. (2013a). The SEMA5A gene is associated with hippocampal volume, and their interaction is associated with performance on Raven's Progressive Matrices. *Neuroimage* 88C, 181-187.

Zhu, Y. C., Li, D., Wang, L., Lu, B., Zheng, J., Zhao, S. L., Zeng, R., and Xiong, Z. Q. (2013b). Palmitoylation-dependent CDKL5-PSD-95 interaction regulates synaptic targeting of CDKL5 and dendritic spine development. *Proc. Natl. Acad. Sci. USA* 110, 9118-9123.

Zhu, X., Morales, F. C., Agarwal, N. K., Dogruluk, T., Gagea, M., Georgescu, M. M. (2013c). Moesin is a glioma progression marker that induces proliferation and Wnt/beta-catenin pathway activation via interaction with CD44. *Cancer Res.* 73, 1142-1155.

Zilberberg, A., Yaniv, A., and Gazit, A. (2004). The low density lipoprotein receptor-1, LRP1, interacts with the human frizzled-1 (HFz1) and down-regulates the canonical Wnt signaling pathway. *J. Biol. Chem.* 279, 17535-17542.

Supplementary Table 2. Gene indications in epilepsy and schizophrenia for each high-risk autism gene. Available citations for epilepsy indicate a positive “hit” in the literature. “Yes/No” indicates whether a gene is contained within the *SzGene* Database.

Aut Gene	Indicated in Epilepsy	Contained in SzGene Database
<i>ACSL4</i>		Yes
<i>ADNP</i>	Cosgrave et al. (2008)	No
<i>ADSL</i>	Marie et al. (1999)	Yes
<i>AFF2</i>	Coffee et al. (2008)	No
<i>AGTR2</i>	Ylisaukko-oja et al. (2004)	No
<i>AHI1</i>	Dixon-Salazar	Yes
<i>ALDH5A1</i>	Knerr et al. (2007)	Yes
<i>ALDH7A1</i>	Plecko et al. (2007)	No
<i>APIS2</i>	Tarpey et al. (2006)	No
<i>ARHGEF6</i>		No
<i>ARID1B</i>		No
<i>ARX</i>	Kato et al. (2007)	No
<i>ASMT</i>		No
<i>ATPI3A4</i>		No
<i>ATRX</i>	Akahoshi et al. (2005)	No
<i>AUTS2</i>	Mefford et al. (2010)	No
<i>AVPR1A</i>		No
<i>BCL2</i>		No
<i>BDNF</i>	Scharfman et al. (2002)	Yes
<i>BRAF</i>	Adachi et al. (2012)	No
<i>BTBD</i>	Salbert et al. (1993)	No
<i>C4B</i>		Yes
<i>CACNA1C</i>		Yes
<i>CACNA1E</i>	Rijkers et al. (2010)	No
<i>CACNA1F</i>		Yes
<i>CACNA1H</i>	Chen et al. (2003)	No
<i>CADMI</i>		No
<i>CASK</i>	Saito et al. (2012)	No
<i>CDH10</i>		No
<i>CDH9</i>		No
<i>CDKL5</i>	Archer et al. (2006)	No
<i>CEP290</i>	Şenocak et al. (2010)	No
<i>CHD7</i>	Lalani et al. (2009)	No
<i>CHD8</i>		No
<i>CHRNA4</i>	Hirose et al. (1999)	Yes

<i>CHRNA2</i>	Phillips et al. (2001)	Yes
<i>CNTN4</i>	Roohi et al. (2009)	No
<i>CNTNAP2</i>	Mefford et al. (2010)	No
<i>COMT</i>	Doyle & Sellinger (1980)	Yes
<i>CREBBP</i>	Seltzer & Paciorkowski (2014)	No
<i>CYFIP1</i>		No
<i>DBH</i>	Browning et al. (1989)	Yes
<i>DCX</i>	Kerjan et al. (2009)	No
<i>DHCR7</i>	Johnson (1975)	No
<i>DISC1</i>		Yes
<i>DMD</i>	De Sarro et al. (2004)	No
<i>DMPK</i>		No
<i>DNAH5</i>		No
<i>DRD3</i>		Yes
<i>DRD4</i>	Rubenstein et al. (2001)	Yes
<i>DYRK1A</i>	Courcet et al. (2012)	No
<i>EHMT1</i>	Kleefstra et al. (2006)	No
<i>EIF4E</i>		No
<i>EN2</i>	Tripathi et al. (2008)	No
<i>FBXO33</i>	Flood et al. (2004)	No
<i>FEZF1</i>		No
<i>FEZF2</i>	Lodato et al. (2011)	No
<i>FGD1</i>		No
<i>FGFR2</i>		Yes
<i>FHIT</i>		No
<i>FMR1</i>	Musemeci et al. (2007)	No
<i>FOXL1</i>	Brunetti-Pierri et al. (2011)	No
<i>FOXP1</i>	Carr et al. (2010)	No
<i>FOXP2</i>	Turner et al. (2013)	Yes
<i>FTSJ1</i>	Takano et al. (2008)	No
<i>GABRA4</i>	Roberts et al. (2005)	No
<i>GABRA5</i>	Di Rocco et al. (2013)	Yes
<i>GABRB1</i>		No
<i>GABRB3</i>	DeLorey & Olsen (1999)	No
<i>GABRG3</i>	Pelc et al. (2008)	No
<i>GAD1</i>	Darrah et al. (2013)	Yes
<i>GAMT</i>	Mercimek-Mahmutoglu et al. (2006)	No
<i>GATM</i>	Stöckler et al. (2014)	No
<i>GRIA3</i>	Bonnet et al. (2012)	Yes
<i>GRIK2</i>	Mulle et al. (1998)	Yes
<i>GRIN2B</i>	Lemke et al. (2014)	Yes

<i>GRIN3B</i>		No
<i>GRIP1</i>		Yes
<i>GRPR</i>		No
<i>GSE1</i>		No
<i>GSTM1</i>	Liu & Tsai (2002)	Yes
<i>GUCY2D</i>		No
<i>HLA-DRB1</i>	Jain et al. (1999)	No
<i>HOXA1</i>		No
<i>HRAS</i>	Della Marca et al. (2011)	No
<i>IGF2</i>	Dikkes et al. (2007)	Yes
<i>IL1RAPL1</i>	Dinopoulos et al. (2014)	No
<i>IMMP2L</i>		No
<i>IQSEC2</i>	Gandomi et al. (2014)	No
<i>ITGB3</i>		No
<i>KCND2</i>	Lee et al. (2014)	No
<i>KCNMA1</i>	Ermolinsky et al. (2008)	No
<i>KDM5C</i>	Poeta et al. (2013)	No
<i>KIAA0100</i>		No
<i>KIAA2022</i>	Cantagrel et al. (2004)	No
<i>KRAS</i>	Adachi et al. (2012)	No
<i>L1CAM</i>	Maruta et al. (1996)	Yes
<i>L2HGDH</i>	Vilarinho et al. (2010)	No
<i>LAMP2</i>		No
<i>LRFN5</i>	de Bruijn et al. (2010)	No
<i>LRP1</i>		No
<i>MACROD2</i>		No
<i>MAOA</i>	Teskey et al. (2004)	Yes
<i>MAP2K1</i>	Nateri et al. (2007)	No
<i>MAPK1</i>	Nateri et al. (2007)	No
<i>MAPK3</i>		No
<i>MARK1</i>		No
<i>MBD5</i>	Talkowski et al. (2011)	No
<i>MECP2</i>	Jian et al. (2007)	No
<i>MED12</i>	Van Buggenhout & Fryns (2006)	Yes
<i>MEF2C</i>	Nowakowska et al. (2010)	No
<i>MEGF11</i>		No
<i>MET</i>	Bae et al (2010)	Yes
<i>MIDI1</i>		No
<i>MKKS</i>		No
<i>MSNPIAS</i>		No
<i>NDP</i>	Lev et al. (2007)	No

<i>NFI</i>	Kulkantrakorn & Geller (1998)	No
<i>NFIX</i>	Yoneda et al. (2012)	No
<i>NHS</i>		No
<i>NIPAI</i>	Svenstrup et al. (2011)	No
<i>NIPBL</i>	Pavlidis et al. (2014)	No
<i>NLGN3</i>		No
<i>NLGN4X</i>		No
<i>NPHP1</i>	Bartnik et al. (2012)	No
<i>NRXN1</i>	Harrison et al. (2011)	Yes
<i>NRXN2</i>		No
<i>NSD1</i>	Tatton-Brown & Nazneen (2004)	No
<i>NTF3</i>	Risbud et al. (2011)	Yes
<i>NTF4</i>		No
<i>OCRL</i>	Ramirez et al. (2012)	No
<i>OPHN1</i>	Santos-Rebouças et al. (2014)	No
<i>OTC</i>	Bogdanovic et al. (2000)	No
<i>OTX1</i>	Acampora et al. (1996)	No
<i>OXTR</i>	Sala et al. (2011)	Yes
<i>PAFAH1B1</i>	Greenwood et al. (2009)	Yes
<i>PAH</i>	Martynyuk et al. (2007)	Yes
<i>PCDH19</i>	Depienne et al. (20009)	No
<i>PHF6</i>	Lower et al. (2002)	No
<i>PHF8</i>		No
<i>PITX1</i>		No
<i>POGZ</i>		No
<i>POMGNT1</i>	Teber et al. (2008)	No
<i>POMT1</i>	Mochida (2008)	No
<i>PQBP1</i>	Stevenson et al. (1998)	No
<i>PRKCB</i>	Szepetowski & Monaco (1998)	No
<i>PRKX</i>		No
<i>PRSS12</i>		No
<i>PTCHD1</i>		No
<i>PTEN</i>	Backman et al. (2001)	Yes
<i>PTPN11</i>	de Jong et al. (2011)	No
<i>RAB39B</i>	Giannandrea et al. (2010)	No
<i>RAI1</i>	Goldman (2006)	No
<i>RAPGEF4</i>	Zhao et al. (2013)	No
<i>RBFOX1</i>	Lal et a. (2013)	No
<i>RELN</i>	Gong et al. (2007)	Yes
<i>RIMS3</i>		No
<i>RNF135</i>		No

<i>RPE65</i>		No
<i>RPGRIPI1</i>	Şenocak et al. (2010)	Yes
<i>RPL10</i>		No
<i>RPS6KA3</i>	Matsumoto et al. (2013)	No
<i>SATB2</i>	Leoyklang et al. (2007)	No
<i>SBF1</i>		No
<i>SCN1A</i>	Claes et al. (2001)	No
<i>SCN2A</i>	Herlenius et al. (2007)	No
<i>SEMA5A</i>	Yang et al. (2005)	No
<i>SEZ6L2</i>	Guerra (2011)	No
<i>SGSH</i>	OMIM (2014)	No
<i>SHANK2</i>		No
<i>SHANK3</i>	Han et al. (2013)	Yes
<i>SLC22A9</i>		No
<i>SLC25A12</i>	Falk et al. (2014)	Yes
<i>SLC6A4</i>	Hrvoje et al. (2010)	Yes
<i>SLC6A8</i>	Fons et al. (2009)	No
<i>SLC9A6</i>	Gilfillan et al. (2008)	No
<i>SLCO1C1</i>		No
<i>SMC1A</i>	Baquero-Montoya et al. (2014)	No
<i>ST8SIA2</i>	Kamien et al. (2014)	Yes
<i>SUV420H1</i>		No
<i>SYN1</i>	Fassio et al. (2011)	No
<i>TBR1</i>	Traylor et al. (2012)	No
<i>TBX1</i>		Yes
<i>TRIO</i>		No
<i>TSC1</i>	Meikle et al. (2007)	No
<i>TSC2</i>	Chu-Shore et al. (2009)	Yes
<i>TTN</i>		No
<i>TUBA1A</i>	Sohal et al. (2012)	Yes
<i>TUBGCP5</i>		No
<i>UBE3A</i>	Minassian et al. (1998)	No
<i>UPF3B</i>		No
<i>UPP2</i>		No
<i>VIP</i>	Marksteiner et al. (1989)	No
<i>VPS13B</i>		No
<i>YWHAE</i>		Yes
<i>ZNF674</i>		No
<i>ZNF81</i>		No

References for Supplementary Table 2

Acampora, D., Mazan, S., Avantaggiato, V., Barone, P., Tuorto, F., Lallemand, Y., Brûlet, P., and Simeone, A. (1996). Epilepsy and brain abnormalities in mice lacking the *Otx1* gene. *Nat. Genet.* 14, 218-222.

Adachi, M., Abe, Y., Aoki, Y., and Matsubara, Y. (2012). Epilepsy in RAS/MAPK syndrome: two cases of cardio-facio-cutaneous syndrome with epileptic encephalopathy and a literature review. *Seizure* 21, 55-60.

Akahoshi, K., Ohashi, H., Hattori, Y., Saitoh, S., Fukushima, Y., and Wada, T. (2005). A woman with 46,XX,dup(16)(p13.11-p13.3) and the ATR-X phenotype. *Am. J. Med. Genet. Part A* 132A, 414-418.

Archer, H. L., Evans, J., Edwards, S., Colley, J., Newbury-Ecob, R., O'Callaghan, F., Huyton, M., O'Regan, M., Tolmie, J., et al. (2006). *J. Med. Genet.* 43, 729-734.

Backman, S. A., Stambolic, V., Suzuki, A., Haight, J., Elia, A., Pretorius, J., Tsao, M.-S., Shannon, P., Bolon, B., Ivy, G. O., et al. (2001). Deletion of *Pten* in mouse brain causes seizures, ataxia and defects in soma size resembling Lhermitte-Duclos disease. *Nat. Genet.* 29, 396-403.

Bae, M. H., Bissonette, G. B., Mars, W. M., Michalopoulos, G. K., Achim, C. L., Depireux, D. A., and Powell, E. M. (2010). Hepatocyte growth factor (HGF) modulates GABAergic inhibition and seizure susceptibility. *Exp. Neurol.* 221, 129-135.

Baquero-Montoya, C., Gil-Rodríguez, M. C., Teresa-Rodrigo, M. E., Hernández-Marcos, M., Bueno-Lozano, G., Bueno-Martínez, I., Remeseiro, S., Fernández-Hernández, R., Bassecourt-Serra, M., et al. (2014). Could a patient with *SMC1A* duplication be classified as a human cohesinopathy? *Clinical Genet.* 85, 446-451.

Bartnik, M., Szczepanik, E., Derwińska, K., Wiśniowiecka-Kowalik, B., Gambin, T., Sykulski, M., Ziemkiewicz, K., Kędzio, M., Gos, M., Hoffman-Zacharska, D., et al. (2012). Application of array comparative genomic hybridization in 102 patients with epilepsy and additional neurodevelopmental disorders. *Am. J. Med. Genet. B Neuropsychiatr. Genet.* 159B, 760-771.

Bogdanovic, M. D., Kidd, D., Briddon, A., Duncan, J. S., and Land, J. M. (2000). Late onset heterozygous ornithine transcarbamylase deficiency mimicking complex partial status epilepticus. *J. Neurol. Neurosurg. Psychiatry* 69, 813-815.

Bonnet, C., Masurel-Paulet, A., Khan, Béri-Dexheimer, Callier, P., Mugneret, F., Philippe, C., Thauvin-Robinet, C., Faivre, L., et al. (2012). Exploring the potential role of disease-causing mutation in a gene desert: duplication of noncoding elements 5' of *GRIA3* is associated with *GRIA3* silencing and X-linked intellectual disability. *Hum. Mutat.* 33, 355-358.

Browning, R. A., Wade, D. R., Marcinczyk, M., Long, G. L., and Jobe, P. C. (1989). Regional brain abnormalities in norepinephrine uptake and dopamine beta-hydroxylase activity in the genetically epilepsy-prone rat. *J. Pharmacol. Exp. Ther.* *249*, 229-235.

Brunetti-Pierri, N., Paciokowski, A. R., Ciccone, R., Mina, E. D., Bonaglia, M. C., Borgatti, R., Schaaf, C. P., Sutton, V. R., Xia, Z., Jelluma, N., et al. (2011). Duplications of *FOXG1* in 14q12 are associated with developmental epilepsy, mental retardation, and severe speech impairment. *Euro. J. Hum. Genet.* *19*, 102-107.

Cantagrel, V., Lossi, A. M., Boulanger, S., Depetris, D., Mattei, M. G., Gecz, J., Schwartz, C. E., Van Maldergem, L., and Villard, L. (2004). Disruption of a new X linked gene highly expressed in brain in a family with two mentally retarded males. *J. Med. Genet.* *41*, 736-742.

Carr, C. W., Moreno-De-Luca, D., Parker, C., Zimmerman, H. H., Ledbetter, N., Martin, C. L., Dobyns, W. B., and Abdul-Rahman, O. A. (2010). Chiari I malformation, delayed gross motor skills, severe speech delay, and epileptiform discharges in a child with *FOXP1* haploinsufficiency. *Euro. J. Hum. Genet.* *18*, 1216-1220.

Chen, Y., Lu, J., Pan, H., Zhang, Y., Wu, H., Xu, K., Liu, X., Jiang, Y., Bao, X., Yao, Z., et al. (2003). Association between genetic variation of *CACNA1H* and childhood absence epilepsy. *Ann. Neurol.* *54*, 239-243.

Chu-Shore, C., J., Major, P., Montenegro, M., and Thiele, E. (2009). Cyst-like tubers are associated with TSC2 and epilepsy in tuberous sclerosis complex. *Neurology* *72*, 1165-1169.

Claes, L., Del-Favero, D., Ceulemans, B., Lagae, L., Van Broeckhoven, C., and De Jonghe, P. (2001). De novo mutations in the sodium-channel gene *SCN1A* cause severe myoclonic epilepsy of infancy. *Am. J. Hum. Genet.* *68*, 1327-1332.

Coffee, B., Ikeda, M., Budimirovic, D. B., Hjelm, L. N., Kaufman, W. E., and Warren, S. T. (2008). Mosaic *FMR1* deletion causes fragile X syndrome and can lead to molecular misdiagnosis: a case report and review of the literature. *Am. J. Med. Genet.* *146A*, 1358-1367.

Cosgrave, A. S., McKay, J. S., Bubb, V., Morris, R., Quinn, J. P., and Thippeswamy T. (2008). Regulation of activity-dependent neuroprotective protein (ADNP) by the NO-cGMP pathway in the hippocampus during kainic acid-induced seizure. *Neurobiol. Dis.* *30*, 281-292.

Courcet, J.-B., Faivre, L., Malzac, P., Masurel-Paulet, A., Lopez, E., Callier, P., Lambert, L., Lemesle, M., Thevenon, J., Gigot, N., et al. (2012). The *DYRK1A* gene is a cause of syndromic intellectual disability with severe microcephaly and epilepsy. *J. Med. Genet.* *49*, 731-736.

- Darrah, S. D., Miller, M. A., Ren, D., Hoh, N. Z., Scanlon, J. M., Conley, Y. P., and Wagner, A. K. (2013). *Epilep. Res.* *103*, 180-194.
- de Bruijn, D. R., van Dijk, A. H., Pfundt, R., Hoischen, A., Merkx, G. F., Gradek, G. A., Lybæk, H., Stray-Pedersen, A., Brunner, H. G., and Houge, G. (2010). Severe progressive autism associated with two de novo changes: a 2.6-Mb 2q31.1 deletion and a balanced t(14;21)(q21.1;p11.2) translocation with long-range epigenetic silencing of LRFN5 expression. *Mol. Syndromol.* *1*, 46-57.
- de Jong, M., Schieving, J., and Goraj, B. (2011). Remarkable intra-cerebral lesions on MRI in a patient with Noonan syndrome. *Europ. J. Radiol. Extra* *78*, e17-e19.
- Della Marca, G., Leoni, C., Dittoni, S., Battaglia, D., Losurdo, A., Testani, E., Colicchio, S., Gnani, V., Gambardella, M. L., Alfieri, P., et al. (2011). Increased sleep spindle activity in patients with Costello syndrome (HRAS gene mutation). *J. Clin. Neurophysiol.* *28*, 314-318.
- DeLorey, T. M., and Olsen, R. W. (1999). GABA and epileptogenesis: comparing gabrb3 gene-deficient mice with Angelman syndrome in man. *Epilep. Res.* *36*, 123-132.
- Depienne, C., Bouteiller, D., Keren, B., Cheuret, E., Poirier, K., Trouillard, O., Benyahia, B., Quelin, C., Carpentier, W., Julia, S., et al. (2009). Sporadic infantile epileptic encephalopathy caused by mutations in *PCDH19* resembles Dravet syndrome but mainly affects females. *PLoS Genet.* *5*, e1000381.
- De Sarro, G., Ibbadu, G. F., Marra, R., Rotiroti, D., Loiacono, A., Di Paola, E. D., and Russo, E. (2004). Seizure susceptibility to various convulsant stimuli in dystrophin-deficient *mdx* mice. *Neurosci. Res.* *50*, 37-44.
- Dikkes, P., Jaffe, D. B., Guo, W. H., Chao, C., Hemond, P., Yoon, K., Zurakowski, D., and Lopezh, M. F. (2007). *Brain Res.* *1175*, 85-95.
- Dinopoulos, A., Stefanou, M. I., Attilakos, A., Tsirouda, M., and Papaevangelou, V. (2014). A case of startle epilepsy associated with IL1RAPL1 gene deletion. *Pediatr. Neurol.* *51*, 271-274.
- Dixon-Salazar, T., Silhavy, J. L., Marsh, S. E., Louie, C. M., Scott, L. C., Gururaj, A., Al-Gazali, L., Al-Tawari, A., Kayserili, H., Sztriha, L., et al. (2004). Mutations in the *AHII* gene, encoding Jouberting, cause Joubert syndrome with cortical polymicrogyria. *Am. J. Hum. Genet.* *75*, 979-987.
- Di Rocco, A., Loggini, A., Di Rocco, M., Di Rocco, P., Rossi, R. P., Gimelli, G., and Bazil, C. (2013). Paradoxical worsening of seizure activity with pregabalin in an adult with isodicentric 15 (IDIC-15) syndrome involving duplications of the *GABRB3*, *GABRA5* and *GABRG3* genes. *BMC Neurol.* *13*, 43.

- Doyle, R. L., and Sellinger, O. Z. (1980). Differences in activity in cerebral methyltransferases and monoamine oxidases between audiogenic seizure susceptible and resistant mice and deermice. *Pharm. Biochem. Beh.* *13*, 589-591.
- Ermolinsky, B., Arshadmansab, M. F., Pacheco Otalora, L. F., Zarei, M. M., and Garrido-Sanabria, E. R. (2008). Deficit of *Kcnma1* mRNA expression in the dentate gyrus of epileptic rats. *Neuroreport* *19*, 1291-1294.
- Falk, M. J., Li, D., Gai, X., McCormick, E., Place, E., Lasorsa, F. M., Otieno, F. G., Hou, C., Kim, C. E., et al. (2014). *AGC1* deficiency causes infantile epilepsy, abnormal myelination, and reduced *N*-acetylaspartate. *JIMD Reports*, doi: 10.1007/8904_2013_287
- Fassio, A., Patry, L., Congia, S., Onofri, F., Piton, A., Gauthier, J. Pozzi, D., Messa, M., Defranchi, E., Fadda, M., et al. (2011). *SYN1* loss-of-function mutations in autism and partial epilepsy cause impaired synaptic function. *Hum. Mol. Genet.* *20*, 2297-2307.
- Flood, W. D., Moyer, R. W., Tsykin, A., Sutherland, G. R., and Koblar, S. A. (2004). *Nxf* and *Fbxo33*: novel seizure-responsive genes in mice. *Euro. J. Neurosci.* *20*, 1819-1826.
- Fons, C., Sempere, Á., Sanmarti, F. X., Arias, Á, Póo, Pineda, M., Ribes, A., Merinero, B., Vilaseca, M. A., Salomons, G. S., et al. (2009). Epilepsy spectrum in cerebral creatine transporter deficiency. *Epilepsia* *50*, 2168-2170.
- Gandomi, S. K., Farwell Gonzalez, K. D., Parra, M., Shahmirzadi, L., Mancuso, J., Pinchurin, P., Temme, R., Dugan, S., Zeng, W., and Tang, S. (2014). Diagnostic exome sequencing identifies two novel *IQSEC2* mutations associated with X-linked intellectual disability with seizures: implications for genetic counseling and clinical diagnosis. *J. Genet. Couns.* *23*, 289-298.
- Giannandrea, M., Bianchi, V., Mignogna, M. L., Sirri, A., Carrabino, S., D'Elia, E., Vecellio, M., Russo, S., Cogliati, F., Larizza, L. Ropers, H.-H., et al. (2010). Mutations in the small GTPase gene *RAB39B* are responsible for X-linked mental retardation associated with autism, epilepsy, and macrocephaly. *Am. J. Hum. Genet.* *86*, 185-195.
- Gilfillan, G. D., Selmer, K. K., Roxrud, I., Smith, R., Kyllerman, M., Eiklid, K., Kroken, M., Mattingsdal, M., Egeland, T., Stenmark, H., et al. (2008). *SLC9A6* mutations caused X-linked mental retardation, microcephaly, epilepsy, and ataxia, a phenotype mimicking Angelman syndrome. *Am. J. Hum. Genet.* *82*, 1003-1010.
- Gong, C., Wang, T.-W., Huang, H. S., and Parent, J. M. (2007). Reelin regulates neuronal progenitor migration in intact and epileptic hippocampus. *J. Neurosci.* *27*, 1803-1811.
- Goldman, A. M. (2006). Topical review: epilepsy and chromosomal rearrangements in Smith-Magenis syndrome [del(17)(p11.2p11.2)]. *J. Child Neurol.* *21*, 93-98.

- Greenwood, J. S. F., Wang, Y., Estrada, R. C., Ackerman, L., Ohara, P. T., and Baraban, S. C. (2009). Seizures, enhanced excitation, and increased vesicle number of *Lis1* mutant mice. *Ann. Neurol.* 66, 644-653.
- Guerra, D. J. (2011). The molecular genetics of autism spectrum disorders: genomic mechanisms, neuroimmunopathology, and clinical implications. *Autism Res. Treat.* 2011, 398636.
- Han, K., Holder, J. L., Schaaf, C. P., Lu, H., Chen, H., Kang, H., Tang, J., Wu, Z., Hao, S., Cheung, S. W., et al. (2013). *SHANK3* overexpression causes manic-like behavior with unique pharmacogenetic properties. *Nature* 503, 72-77.
- Harrison, V., Connell, L., Hayesmoore, J., McParland, J., Pike, M. G., and Blair, E. (2011). Compound heterozygous deletion of *NRXN1* causing severe developmental delay with early onset epilepsy in two sisters. *Am. J. Med. Genet. A* 155A, 2826-2831.
- Herlenius, E., Heron, S. E., Grinton, B. E., Keay, D., Scheffer, I. E., Mulley, J. C., and Berkovic, S. (2007). *SCN2A* mutations and benign familial neonatal-infantile seizures: the phenotypic spectrum. *Epilepsia* 48, 1138-1142.
- Hirose, S., Iwata, H., Akiyoshi, H., Kobayashi, K., Ito, M., Wada, K., Kaneko, S., and Mitsundome, A. (1999). A novel mutation of *CHRNA4* responsible for autosomal dominant nocturnal frontal lobe epilepsy. *Neurology* 53, 1749-1753.
- Hrvoje, H., Jasminka, S., Lipa, C.-S., Vida, D., and Branimir, J. (2010). Association of serotonin transporter promoter (5-HTTLPR) and intron 2 (VNTR-2) polymorphisms with treatment response in temporal lobe epilepsy. *Epilepsy Res.* 91, 35-38.
- Jain, S., Padma, M. V., Kanga, U., Mehra, N. K., Puri, A., and Maheshwari, M. C. (1999). Family studies and human leukocyte antigen class II typing in Indian probands with seizures in association with single small enhancing computer tomography lesions. *Epilepsia* 40, 232-238.
- Jian, L., Nagarajan, L., de Klerk, N., Ravine, D., Christodoulou, J., and Leonard, H. (2007). Seizures in Rett syndrome: an overview from a one-year calendar study. *Eur. J. Paediatr. Neurol.* 11, 310-317.
- Johnson, V. P. (1975). Smith-Lemli-Opitz syndrome: review and report of two affected siblings. *Euro. J. Pediatr.* 119, 221-234.
- Kamien, B., Harraway, J., Lundie, B., Smallhorne, L., Gibbs, V., Heath, A., and Fillerton, J. M. (2014). Characterization of a 520 kb deletion on chromosome 15q26.1 including *ST8SLA2* in a patient with behavioral disturbance, autism spectrum disorder, and epilepsy. *Am. J. Hum. Genet.* 164A, 782-788.

- Kato, M., Saitoh, S., Kamei, A., Shiraishi, H., Ueda, Y., Akasaka, M., Tohyama, J., Akasaka, N., and Hayasaka, K. (2007). A longer polyalanine expansion mutation in the *ARX* gene causes early infantile epileptic encephalopathy with suppression-burst pattern (Ohtahara syndrome). *Am. J. Hum. Genet.* 81, 361-366.
- Kerjan, G., Koizumi, H., Han, E. B., Dubé, C. M., Djakovic, S. N., Patrick, G. N., Baram, T. Z., Heinemann, S. F., and Gleeson, J. G. (2009). Mice lacking doublecortin and doublecortin-like kinase 2 display altered hippocampal neuronal maturation and spontaneous seizures. *Proc. Natl. Acad. Sci. USA* 106, 6766-6771.
- Kleefstra, T., Brunner, H. G., Amiel, J., Oudakker, A. R., Nillesen, W. M., Magee, A., Geneviève, D., Cormier-Daire, V., van Esch, H., Fryns, J.-P., et al. (2006). Loss-of-function mutations in *Euchromatin Histone Methyltransferase 1 (EHMT1)* cause the 9q34 subtelomeric deletion syndrome. *Am. J. Hum. Genet.* 79, 370-377.
- Knerr, I., Pearl, P. L., Bottiglieri, T., Snead, O. C., Jakobs, C., and Gibson, K. M. (2007). Therapeutic concepts in succinate semialdehyde dehydrogenase (SSADH; ALDH5a1) deficiency (γ -hydroxybutyric aciduria). Hypotheses evolved from 25 years of patient evaluation, studies in *Aldh5a1*^{-/-} mice and characterization of γ -hydroxybutyric acid pharmacology. *J. Inher. Metab. Dis.* 30, 279-294.
- Kulkantrakorn, K., and Geller, T. J. (1998). Seizures in neurofibromatosis 1. *Pediatr. Neurol.* 19, 347-350.
- Lal, D., Reinthaler, E. M., Altmüller, J., Toliat, M. R., Thiele, H., Nürnberg, P., Lerche, H., Hahn, A., Möller, R. S., Muhle, H., et al. (2013). *RBFOX1* and *RBFOX3* mutations in Rolandic epilepsy. *PLoS One* 8, e73323.
- Lalani, S. R., and Belmont, J. W. (2009). CHARGE syndrome. In *Encyclopedia of Molecular Mechanisms of Disease*. F. Lang, ed. (Berlin: Springer), pp. 312-313.
- Lee, H., Lin, M. C., Kornblum, H. I., Papazian, D. M., and Nelson, S. F. (2014). Exome sequencing identifies de novo gain of function missense mutation in *KCND2* in identical twins with autism and seizures that slows potassium channel inactivation. *Hum. Mol. Genet.* 23, 3481-3489.
- Lemke, J. R., Hendrickx, R., Geider, K., Laube, B., Schwake, M., Harvey, R. J., James, V. M., MBIol, A. P., Neidhardt, J., et al. (2014). *GRIN2B* mutations in west syndrome and intellectual disability with focal epilepsy. *Ann. Neurol.* 75, 147-154.
- Leoyklang, P., Suphapeetiporn, K., Siriwan, P., Desudchit, T., Chaowanapanja, P., Gahl, W. A., and Shotelersuk, V. (2007). Heterozygous nonsense mutation *SATB2* associated with cleft palate, osteoporosis, and cognitive defects. *Hum. Mutat.* 28, 732-738.
- Lev, D., Weigl, Hasan, M., Gak, E., Davidovich, M., Vinkler, C., Leshinsky-Silver, E., Lerman-Sagie, T., and Waternberg, N. (2007). A novel missense mutation in the *NDP*

gene in a child with Norrie disease and severe neurological involvement including infantile spasms. *Am. J. Med. Genet. A* 143A, 921-924.

Liu, C.-S., and Tsai, C.-S. (2002). Enhanced lipid peroxidation in epileptics with null genotype of glutathione *S*-transferase M1 and intractable seizure. *Japan. J. Pharmacol.* 90, 291-294.

Lodato, S., Rouaux, C., Quast, K. B., Jantrachotechatchawan, C., Studer, M., Hensch, T. K., and Arlotta, P. (2011). Excitatory projection neuron subtypes control the distribution of local inhibitory interneurons in the cerebral cortex. *Neuron* 69, 763-779.

Lower, K. M., Turner, G., Kerr, B. A., Mathews, K. D., Shaw, M. A., Gedeon, Á. K., Schelley, S., Hoyme, H. E., White, S. M., Delatycki, M. B., et al. (2002). Mutations in *PHF6* are associated with Börjeson-Forssman-Lehmann syndrome. *Nat. Genet.* 32, 661-665.

Marie, S., Cuppens, H., Heuterspreute, M., Jaspers, M., Tola, E. Z., Gu, X. X., Leguis, E., Vincent M.-F., Jaeken, J., Cassiman, J.-J., et al. (1999). Mutation analysis in adenylosuccinate lyase deficiency: eight novel mutations in the re-evaluated full ADSL coding sequence. *Hum. Mutat.* 13, 197-202.

Marksteiner, J., Sperk, G., and Maas, D. (1989). Differential increases in brain levels of neuropeptide Y and vasoactive intestinal polypeptide after kainic acid-induced seizures in the rat. *Naunyn Schmiedebergs Arch. Pharmacol.* 339, 173-177.

Martynyuk, A. E., Ucar, D. A., Yang, D. D., Norman, W. M., Carney, P. R., Dennis, D. M., and Laipis, P. J. (2007). Epilepsy in phenylketonuria: a complex dependence on serum phenylalanine levels. *Epilepsia* 48, 1143-1150.

Maruta, K., Ohi, T., Ohdo, S., Takechi, T., Sakuragawa, N., and Matsukura, S. (1996). [A family with X-linked hydrocephalus resulting from mutations in the neural cell adhesion molecule L1]. *Rinsho Shinkeigaku* 36, 462-466.

Matsumoto, A., Kuwajima, M., Miyake, K., Kojima, K., Nakashima, N., Jimbo, E. F., Kubota, T., Momoi, M. Y., and Yamagata, T. (2013). An Xp22.12 microduplication including *RPS6KA3* identified in a family with variably affected intellectual and behavioral disabilities. *J. Hum. Genet.* 58, 755-757.

Mefford, H. C., Muhle, H., Ostertag, P., von Spiczak, S., Buysse, K., Baker, C., Franke, A., Malafosse, A., Genton, P., Thomas, P., et al. (2010). Genome-wide copy number variation in epilepsy: novel susceptibility loci in idiopathic generalized and focal epilepsies. *PLoS Genet.* 6, e1000962.

Meikle, L., Talos, D. M., Onda, H., Pollizzi, K., Rotenberg, A., Sahin, M., Jensen, F. E., and Kwiatkowski, D. J. (2007). A mouse model of tuberous sclerosis: neuronal loss of

Tsc1 causes dysplastic and ectopic neurons, reduced myelination, seizure activity, and limited survival. *J. Neurosci.* 27, 5546-5558.

Mercimek-Mahmutoglu, S., Stoeckler-Ipsiroglu, S., Adami, A., Appleton, R., Araújo, H. C., Duran, M., Ensenauer, R., Fernandez-Alvarez, E., Garcia, P., Grolik, C., et al. (2006). GAMT deficiency. *Neurology* 67, 480-484.

Minassian, B., Delorey, T. M., Olsen, R. W., Philppart, M., Bronstein, Y., Zhang, Q., Guerrini, R., van Ness, Paul, Livet, M. O., et al. (1998). Angelman syndrome: correlations between epilepsy phenotypes and genotypes. *Ann. Neurol.* 43, 485-493.

Mochida, G. H. (2008). Genetic malformations of the cerebral cortex and seizure susceptibility during infancy and childhood. In *Biology of Seizure Susceptibility in Developing Brain*. T. Takahashi and Y. Fukuyama, eds. (Escher: John Libbey Eurotext Ltd.), pp. 175-184.

Mulle, C., Sailer, A., Pérez-Otaño, I., Dickinson-Anson, H., Castillo, P. E., Bureau, H., Maron, C., Gage, F. H., Mann, et al. (1998). Altered synaptic physiology and reduced susceptibility to kainite-induced seizures in GluR6-deficient mice. *Nature* 392, 601-605.

Musemeci, S. A., Calabrese, G., Bonaccorso, C. M., D'Antoni, S., Brouwer, J. R., Bakker, C. E., Elia, M., Ferri, R., Nelson, D. L., Oostra, B. A., et al. (2007). Audiogenic seizure susceptibility is reduced in fragile X knockout mice after introduction of *FMR1* transgenes. *Exper. Neurol.* 203, 233-240.

Nateri, A. S., Raivich, G., Gebhardt, C., Da Costa, C., Naumann, H., Vreugdenhil, M., Makwana, M., Brandner, S., Adams, R. H., Jefferys, J. G., et al. (2007). ERK activation causes epilepsy by stimulating NMDA receptor activity. *EMBO J.* 26, 4891-4901.

Nowakowska, B. A., Obersztyn, E., Szymańska, K., Bekiesińska-Figatowska, M., Xia, Z., Ricks, C. B., Bocian, E., Stockton, D. W., Szczaluba, K., Nawara, M., et al. (2010). Severe mental retardation, seizures, and hypotonia due to deletions of MEF2C. *Am. J. Med. Genet. B Neuropsychiatr. Genet.* 153B, 1042-1051.

Online Mendelian Inheritance in Man (OMIM). (2014). Mucopolysaccharidosis, type IIIA; MPS3A. Accessed on 9/9/2014 from <http://www.omim.org/entry/252900>

Pavlidis, E., Cantalup, G., Bianchi, S., Piccolo, B., and Pisani, F. (2014). Epileptic features in Cornelia de Lange syndrome: case report and literature review. *Brain Dev.* Doi:10.1016/j.braindev.2013.12.008.

Pelc, K., Boyd, S. G., Cheron, G., and Dan, B. (2008). Epilepsy in Angelman syndrome. *Seizure.* 17, 211-217.

Phillips, H. A., Favre, I., Kirkpatrick, M., Zuberia, S. M., Goudie, D., Heron, S. E., Scheffer, I. E., Sutherland, G. R., Berkovic, S. F., Bertrand, D., et al. (2001). *CHRNA2* is

the second acetylcholine receptor subunit associated with autosomal dominant nocturnal frontal lobe epilepsy. *Am. J. Hum. Genet.* 68, 225-231.

Plecko, B., Paul, K., Paschke, E., Stoeckler-Ipsiroglu, S., Struys, E., Jakobs, C., Hartmann, H., Luecke, T., di Capua, M., Korenke, C., et al. (2007). Biochemical and molecular characterization of 18 patients with pyridoxine-dependent epilepsy and mutations of the antiquitin (*ALDH7A1*) gene. *Hum. Mutat.* 28, 19-26.

Poeta, L., Fusco, F., Drongitis, D., Shoubbridge, C., Manganelli, G., Filosa, S., Paciolla, M., Courtney, M., Collombat, P., Lioi, M. B., et al. (2013). A regulatory path associated with X-linked intellectual disability and epilepsy links KDM5C to the polyalanine expansions in ARX. *Am. J. Hum. Genet.* 92, 114-125.

Ramirez, I. B.-R., Pietka, G., Jones, D. R., Divecha, N., Alia, A., Baraban, S. C., Hurlstone, A. F., and Lowe, M. (2012). *Hum. Mol. Genet.* 21, 1744-1759.

Rijkers, K., Mescheriakova, J., Majoie, M., Lemmens, E., van Wijk, X., Philippens, M., Van Kranen-Mastenbroek, V., Schijns, O., Vles, J., and Hoogland, G. (2010). Polymorphisms in *CACNA1E* and *Camk2d* are associated with seizure susceptibility of Sprague-Dawley rats. *Epilep. Res.* 91, 28-34.

Risbud, R. M., Lee, C., and Porter, B. E. (2011). Neurotrophin-3 mRNA a putative target of miR21 following status epilepticus. *Brain Res.* 1424, 53-59.

Roberts, D. S., Raol, Y. H., Bandyopadhyay, S., Lund, I. V., Budreck, E. C., Passini, M. J., Wolfe, J. H., Brooks-Kayal, A. R., and Russek, S. J. (2005). Egr3 stimulation of *GABRA4* promoter activity as a mechanism for seizure-induced up-regulation of GABA_A receptor $\alpha 4$ subunit expression. *Proc. Natl. Acad. Sci. USA* 102, 11894-11899.

Roohi, J., Montagna, C., Tegay, D. H., Palmer, L. E., DeVincent, C., Pomeroy, J. C., Christian, S. L., Nowak, N., and Hatchwell, E. (2009). Disruption of contactin 4 in three subjects with autism spectrum disorder. *J. Med. Genet.* 46, 176-182.

Rubenstein, M., Cepeda, C., Hurst, R. S., Flores-Hernandez, J., Ariano, M. A., Falzone, T. L., Kozell, L. B., Meshul, C. K., Bunzow, J. R., Low, M. J., et al. (2001). Dopamine D₄ receptor-deficient mice display cortical hyperexcitability. *J. Neurosci.* 21, 3756-3763.

Saitsu, H., Kato, M., Osaka, H., Moriyama, N., Horita, H., Nishiyama, K., Yoneda, Y., Kondo, Y., Tsurusaki, Y., Doi, H., et al. (2012). *CASK* aberrations in male patients with Ohtahara syndrome and cerebellar hypoplasia. *Epilepsia* 53, 1441-1449.

Sala, M., Braidà, D., Lentini, D., Busnelli, M., Bulgheroni, E., Capurro, V., Finardi, A., Donzelli, A., Pattini, L., Rubino, L., et al. (2011). *Biol. Psychiatry* 69, 875-882.

Salbert, B. A., Pellock, J. M., and Wolf, B. (1993). Characterization of seizures associated with biotinidase deficiency. *Neurology* 43, 1351-1355.

Santos-Rebouças, C. B., Belet, S., de Almeida, L. G., Ribeiro, M. G., Medina-Acosta, E., Bahia, P. R. V., da Silva, A. F. A., dos Santos, F. L., de Lacerda, G. C. B., Pimentel, M. M., G., et al. (2014). A novel in-frame deletion affecting the BAR domain of *OPHN1* in a family with intellectual disability and hippocampal alterations. *Euro. J. Hum. Genet.* 22, 644-651.

Scharfman, H. E., Goodman, J. H., Sollas, A. L., and Croll, S. D. (2002). Spontaneous limbic seizures after intrahippocampal infusion of brain-derived neurotrophic factor. *Exp. Neurol.* 174, 201-214.

Seltzer, L. E., and Paciorkowski, A. R. (2014). Genetic disorders associated with postnatal microcephaly. *Am. J. Med. Genet.* 166, 140-155.

Şenocak, E. U., Oğuz, K. K., Haliloğlu, G., Topçu, M., and Cila, A. (2010). Structural abnormalities of the brain other than molar tooth sign in Joubert syndrome-related disorders. *Diagn. Interv. Radiol.* 16, 3-6.

Sohal, A. P. S., Montgomery, T., Mitra, D., and Ramesh, V. (2012). *TUBA1A* mutation-associated lissencephaly: case report and review of the literature. *Pediatr. Neurol.* 46, 127-131.

Stevenson, R. E., Arena, J. F., Ouzts, E., Gibson, A., Shokeir, M. H. K., Vnencak-Jones, C., Lubs, H. A., May, M., and Schwartz, C. E. (1998). Renpenning syndrome maps to Xp11. *Am. J. Hum. Genet.* 62, 1092-1101.

Stöckler, S., Braissant, O., and Schulze, A. (2014). Creatine disorders. In *Physician's Guide to the Diagnosis, Treatment, and Follow-up of Inherited Metabolic Diseases*.

Svenstrup, K., Møller, R. S., Christensen, J., Budtz-Jørgensen, E., Gilling, M., and Nielsen, J. E. (2011). *Eur. J. Neurol.* 18, 1197-1199.

Szepietowski, P., and Monaco, A. P. (1998). Recent progress in the genetics of human epilepsies. *Neurogenetics* 1, 153-163.

Takano, K., Nakagawam E., Inoue, K., Kamada, F., Kure, S., and Goto, Y. (2008). A loss-of-function mutation in the *FTSJ1* gene causes nonsyndromic X-linked mental retardation in a Japanese family. *Am. J. Med. Genet Part B Neuropsych. Genet.* 147B, 479-484.

Talkowski, M. E., Mullegama, S. V., Rosenfeld, J. A., van Bon, B. W., Shen, Y., Repnikova, E. A., Gastier-Foster, J., Thrush, D. L., Kathiresan, S., Ruderfer, D. M., et al. (2011). Assessment of 2q23.1 microdeletion syndrome implicates *MBD5* as a single causal locus of intellectual disability, epilepsy, and autism spectrum disorder. *Am. J. Hum. Genet.* 89, 551-563.

Tarpey, P. S., Stevens, C., Teague, J., Edkins, S., O'Meara, S., Avis, T., Barthorpe, S., Buck, G., Butler, A., Cole, J., et al. (2006). Mutations in the gene encoding the sigma 2 subunit of the adaptor protein 1 complex, *APIS2*, cause X-linked mental retardation. *Am. J. Hum. Genet.* 79, 1119-1124.

Tatton-Brown, K., and Nazneen, R. (2004). Clinical features of NSD1-positive Sotos syndrome. *Clin. Dysmorph.* 13, 199-204.

Teber, S., Sezer, T., Kafalı, M., Manzini, C., Yüksel, B. K., Tekin, M., Fitöz, S., Walsh, C. A., and Deda, G. (2008). Severe muscle-eye-brain disease is associated with a homozygous mutation in the *POMGnT1* gene. *Europ. J. Paediatr. Neurol.* 12, 133-136.

Teskey, G. C., Radford, K. S., Seif, I., and Dyck, R. H. (2004). MAO(A) knockout mice are more susceptible to seizures but show reduced epileptogenesis. *Epilepsy Res.* 59, 25-34.

Traylor, R. N., Dobyns, Rosenfeld, J. A., Wheeler, P., Spence, J. E., Bandholz, A. M., Bawle, E. V., Carmany, E. P., Powell, C. M., et al. (2012). Investigation of *TBR1* hemizyosity: four individuals with 2q24 microdeletions. *Mol. Syndromol.* 3, 102-112.

Tripathi, P. P., Sgadò, P., Scali, M., Viaggi, C., Casarosa, S., Simon, H. H., Vaglini, F., Corsini, G. U., and Bozzi, Y. (2008). Increased susceptibility to kainic acid-induced seizures in *Engrailed-2* knockout mice. *Neuroscience* 159, 842-849.

Turner, S. J., Hildebrand, M. S., Block, S., Damiano, J., Fahey, M., Reilly, S., Bahlo, M., Scheffer, I. E., and Morgan, A. T. (2013). Small intragenic deletion in *FOXP2* associated with childhood apraxia of speech and dysarthria. *Am. J. Med. Genet. A* 161A, 2321-2326.

Van Buggenhout, G., and Fryns, J. P. (2006). Lujan-Fryns syndrome (mental retardation, X-linked, marfanoid habitus). *Orphanet J. Rare Dis.* 1, 26.

Vilarinho, L., Tafulo, S., Sibilio, M., Kok, F., Fontana, F., Diogo, L., Venâncio, M., Ferreira, M., Nogueira, C., Valongo, C., et al. (2010). Identification of novel L2HGDH gene mutations and update of the pathological spectrum. *J. Hum. Genet.* 55, 55-58.

Yang, J., Houk, B., Hauser, K. F., Luo, Y., Smith, G., Schauwecker, E., and Barnes, G. N. (2005). Genetic background regulates semaphorin gene expression and epileptogenesis in mouse brain after kainic acid status epilepticus. *Neuroscience* 131, 853-869.

Ylisaukko-oja, T., Rehnström, K., Vanhala, R., Tengström, Lähdetie, J., Järvelä, I. (2003). Identification of two *AGTR2* mutations in male patients with non-syndromic mental retardation. *Hum. Genet.* 114, 211-213.

Yoneda, Y., Saitsu, H., Touyama, M., Makita, Y., Miyamoto, A., Hamada, K., Kurotaki, N., Tomita, H., Nishiyama, K., Tsurusaki, Y., et al. (2012). Missense mutations in the

DNA-binding/dimerization domain of NFIX cause Sotos-like features. *J. Hum. Genet.* 57, 207-211.

Zhao, K., Wen, R., Wang, X., Pei, L., Shang, Y., Bazan, N., Zhu, L.-Q., Tian, Q., and Lu, Y. (2013). EPAC inhibition of SUR1 receptor increases glutamate release and seizure vulnerability. *J. Neurosci.* 33, 8861-8865.

Supplementary Table 3. General gene product function for each high-risk autism gene.

Aut Gene	Function
<i>ACSL4</i>	fatty acid metabolism; lipid metabolism
<i>ADNP</i>	transcription factor; transcription regulation
<i>ADSL</i>	purine biosynthesis
<i>AFF2</i>	mRNA processing; mRNA splicing
<i>AGTR2</i>	cell membrane; membrane; angiotensin II receptor
<i>AHII</i>	cilium biogenesis/degradation
<i>ALDH5A1</i>	mitochondrion; GABA degradation
<i>ALDH7A1</i>	mitochondrion; detoxification
<i>APIS2</i>	protein transport; transport
<i>ARHGEF6</i>	GTPase; guanine nucleotide exchange factor
<i>ARID1B</i>	transcription regulation; chromatin remodeling
<i>ARX</i>	transcription factor; transcription regulation
<i>ASMT</i>	melatonin biosynthesis
<i>ATP13A4</i>	membrane; cation-transporting ATPase activity
<i>ATRX</i>	DNA damage; DNA repair; transcription regulation; chromatin remodeling; ATPase
<i>AUTS2</i>	unknown
<i>AVPR1A</i>	vasopressin receptor
<i>BCL2</i>	apoptosis; mitochondrion
<i>BDNF</i>	growth factor
<i>BRAF</i>	kinase; transferase; intracellular signal transduction
<i>BTBD</i>	hydrolase; biotin metabolism
<i>C4B</i>	immune regulation; complement pathway
<i>CACNA1C</i>	calcium transport; ion transport
<i>CACNA1E</i>	calcium transport; ion transport
<i>CACNA1F</i>	calcium transport; ion transport
<i>CACNA1H</i>	calcium transport; ion transport
<i>CADM1</i>	cell adhesion
<i>CASK</i>	scaffolding protein; ion channel trafficking; cytoskeleton; cell adhesion; calcium-dependent
<i>CDH10</i>	cell adhesion; calcium-dependent
<i>CDH9</i>	cell adhesion; calcium-dependent
<i>CDKL5</i>	kinase; methylation regulation
<i>CEP290</i>	cilium biogenesis/degradation; protein transport
<i>CHD7</i>	rRNA processing; transcription regulation; chromatin remodeling
<i>CHD8</i>	transcription regulation; chromatin remodeling
<i>CHRNA4</i>	acetylcholine receptor; ion transport
<i>CHRNA2</i>	acetylcholine receptor; ion transport; sodium and potassium transport
<i>CNTN4</i>	cell adhesion
<i>CNTNAP2</i>	cell adhesion

<i>COMT</i>	catecholamine metabolism; neurotransmitter degradation
<i>CREBBP</i>	histone acetylation; transcription regulation
<i>CYFIP1</i>	translation regulation; cytoskeleton
<i>DBH</i>	catecholamine biosynthesis
<i>DCX</i>	cytoskeleton
<i>DHCR7</i>	lipid biosynthesis/metabolism
<i>DISC1</i>	intracellular signaling; canonical Wnt pathway
<i>DMD</i>	cytoskeleton; ligand for dystroglycan
<i>DMPK</i>	kinase; transferase
<i>DNAH5</i>	cytoskeleton; maintenance of cilia integrity; ATPase
<i>DRD3</i>	dopamine receptor; G-protein coupled receptor
<i>DRD4</i>	dopamine receptor; G-protein coupled receptor
<i>DYRK1A</i>	kinase; transferase; nuclear signaling
<i>EHMT1</i>	histone methyltransferase; chromatin remodeling
<i>EIF4E</i>	translation regulation
<i>EN2</i>	transcription factor; transcription regulation
<i>FBXO33</i>	ubiquitination regulation; proteasomal degradation
<i>FEZF1</i>	transcription factor; transcription regulation
<i>FEZF2</i>	transcription factor; transcription regulation
<i>FGD1</i>	GTPase; cytoskeletal regulation
<i>FGFR2</i>	growth factor receptor
<i>FHIT</i>	FGF receptor; receptor kinase/transferase
<i>FMR1</i>	translation regulation; mRNA transport
<i>FOXG1</i>	transcription factor; transcription regulation
<i>FOXP1</i>	transcription factor; transcription regulation
<i>FOXP2</i>	transcription factor; transcription regulation
<i>FTSJ1</i>	tRNA processing; translation regulation; methyltransferase
<i>GABRA4</i>	GABA receptor subunit; ion transport; chloride regulation
<i>GABRA5</i>	GABA receptor subunit; ion transport; chloride regulation
<i>GABRB1</i>	GABA receptor subunit; ion transport; chloride regulation
<i>GABRB3</i>	GABA receptor subunit; ion transport; chloride regulation
<i>GABRG3</i>	GABA receptor subunit; ion transport; chloride regulation
<i>GAD1</i>	neurotransmitter biosynthesis
<i>GAMT</i>	creatine metabolism; methyltransferase
<i>GATM</i>	creatine metabolism; methyltransferase
<i>GRIA3</i>	glutamate receptor; ion transport; calcium regulation
<i>GRIK2</i>	glutamate receptor; ion transport
<i>GRIN2B</i>	glutamate receptor; ion transport; calcium regulation
<i>GRIN3B</i>	glutamate receptor; ion transport; calcium regulation
<i>GRIP1</i>	glutamate receptor interacting protein; scaffold protein; intracellular signal transduction
<i>GRPR</i>	gastrin releasing peptide receptor; g-protein coupled receptor

<i>GSE1</i>	unknown
<i>GSTM1</i>	transferase; glutathione conjugation; detoxification
<i>GUCY2D</i>	cGMP biosynthesis
<i>HLA-DRB1</i>	immune regulation
<i>HOXA1</i>	transcription factor; transcription regulation
<i>HRAS</i>	GTPase; intracellular signal transduction
<i>IGF2</i>	growth factor
<i>IL1RAPL1</i>	interleukin receptor; calcium regulation
<i>IMMP2L</i>	hydrolase; protease; mitochondrion
<i>IQSEC2</i>	guanine nucleotide exchange factor; GTPase regulation
<i>ITGB3</i>	cell adhesion; multi-ligand receptor; calcium-dependent
<i>KCND2</i>	voltage-gated potassium channel; ion transport; calcium activated
<i>KCNMA1</i>	voltage-gated potassium channel; ion transport; calcium activated
<i>KDM5C</i>	histone demethylase; chromatin remodeling; transcription regulation
<i>KIAA0100</i>	unknown
<i>KIAA2022</i>	cytoskeletal regulation; cell adhesion regulation
<i>KRAS</i>	GTPase
<i>L1CAM</i>	cell-cell adhesion/cytoskeleton
<i>L2HGDH</i>	L-2-hydroxyglutarate dehydrogenase; mitochondrion
<i>LAMP2</i>	lysosome regulation; extracellular signaling transduction; intracellular signaling transduction; detox
<i>LRFN5</i>	cell adhesion
<i>LRP1</i>	endocytic receptor; endocytosis & phagocytosis regulation; lipid metabolism
<i>MACROD2</i>	glutamate regulation; ADP ribosylation
<i>MAOA</i>	catecholamine metabolism; neurotransmitter degradation; mitochondrion
<i>MAP2K1</i>	kinase; transferase; intracellular signal transduction
<i>MAPK1</i>	kinase; transferase; intracellular signal transduction
<i>MAPK3</i>	kinase; transferase; intracellular signal transduction
<i>MARK1</i>	kinase; cytoskeleton; cell polarity
<i>MBD5</i>	chromatin remodeling
<i>MECP2</i>	methylation; chromatin remodeling; transcription regulation
<i>MED12</i>	transcription regulation
<i>MEF2C</i>	transcription regulation
<i>MEGF11</i>	cell adhesion
<i>MET</i>	hepatocyte growth factor receptor; intracellular signal transduction
<i>MID1</i>	ubiquitination regulation; cytoskeleton
<i>MKKS</i>	chaperone; cilium biogenesis; vesicular transport
<i>MSNPIAS</i>	regulation of moesin; cytoskeletal regulation
<i>NDP</i>	ligand; Wnt activation
<i>NF1</i>	GTPase; intracellular signal transduction
<i>NFIX</i>	transcription factor; transcription regulation
<i>NHS</i>	cytoskeleton

<i>NIPA1</i>	magnesium transport; ion transport
<i>NIPBL</i>	enhancer regulation; chromatin remodeling; chromatid cohesion
<i>NLGN3</i>	cell adhesion
<i>NLGN4X</i>	cell adhesion
<i>NPHP1</i>	cytoskeleton; cell polarity; cilium biogenesis/degradation
<i>NRXN1</i>	cell adhesion; calcium regulation
<i>NRXN2</i>	cell adhesion; calcium regulation
<i>NSD1</i>	histone methyltransferase; chromatin remodeling; transcription regulation
<i>NTF3</i>	growth factor
<i>NTF4</i>	growth factor
<i>OCRL</i>	cilium biogenesis/degradation; hydrolase; cytoskeleton; lysosome regulation
<i>OPHN1</i>	GTPase; cytoskeleton; intracellular signal transduction
<i>OTC</i>	transferase; mitochondrion; amino acid biosynthesis; arginine biosynthesis; urea cycle
<i>OTX1</i>	transcription factor; transcription regulation
<i>OXTR</i>	oxytocin receptor; g-protein coupled receptor; calcium-dependent
<i>PAFAH1B1</i>	GTPase regulation; cytoskeleton
<i>PAH</i>	phenylalanine catabolism; monooxygenase; oxidoreductase
<i>PCDH19</i>	cell adhesion; calcium-dependent
<i>PHF6</i>	transcription regulation
<i>PHF8</i>	histone regulation; histone lysine demethylase
<i>PITX1</i>	transcription factor; transcription regulation
<i>POGZ</i>	cytoskeleton; cell cycle progression; chromatid adhesion; kinetochore assembly
<i>POMGNT1</i>	O-mannosyl glycosylation; glycosyltransferase
<i>POMT1</i>	O-mannosyltransferase; glycosyltransferase
<i>PQBP1</i>	transcription factor; transcription regulation
<i>PRKCB</i>	kinase; intracellular signal transduction; calcium-dependent
<i>PRKX</i>	kinase; intracellular signal transduction
<i>PRSS12</i>	hydrolase; protease; cytoskeleton
<i>PTCHD1</i>	Shh receptor; intracellular signal transduction
<i>PTEN</i>	phosphatase; hydrolase; intracellular signal transduction; tumor suppressor
<i>PTPN11</i>	phosphatase; hydrolase; intracellular signal transduction
<i>RAB39B</i>	GTPase; vesicular regulation
<i>RAI1</i>	transcription regulation; chromatin remodeling
<i>RAPGEF4</i>	guanine nucleotide exchange factor; GTPase regulation; exocytosis regulation
<i>RBFOX1</i>	mRNA processing; alternative splicing regulation
<i>RELN</i>	cytoskeleton; cell adhesion; calcium-sensitive
<i>RIMS3</i>	cell adhesion; exocytosis regulation
<i>RNF135</i>	ubiquitination regulation; innate immunity
<i>RPE65</i>	hydrolase; isomerase
<i>RPGRIP1L</i>	cytoskeleton; cell adhesion; cilium regulation
<i>RPL10</i>	translation regulation

<i>RPS6KA3</i>	kinase; transferase; intracellular signal transduction
<i>SATB2</i>	transcription regulation; chromatin remodeling
<i>SBF1</i>	pseudophosphatase; guanine nucleotide exchange factor; GTPase
<i>SCN1A</i>	sodium transport; ion transport
<i>SCN2A</i>	sodium transport; ion transport
<i>SEMA5A</i>	cytoskeleton
<i>SEZ6L2</i>	endoplasmic reticulum regulation
<i>SGSH</i>	hydrolase; heparan sulfate degradation; lysosomal regulation
<i>SHANK2</i>	cell adhesion
<i>SHANK3</i>	cell adhesion
<i>SLC22A9</i>	transport
<i>SLC25A12</i>	glutamate-aspartate exchange; mitochondrion; transport; calcium-dependent
<i>SLC6A4</i>	serotonin transport; sodium- and chloride-dependent
<i>SLC6A8</i>	creatine regulation; creatine transport; sodium- & chloride-dependent
<i>SLC9A6</i>	sodium-hydrogen exchange; endosome regulation
<i>SLCO1C1</i>	anion transport; thyroid hormone transport; estradiol transport
<i>SMC1A</i>	cytoskeleton; chromatide cohesion; cell cycle progression; DNA repair
<i>ST8SIA2</i>	sialic acid transfer to oligosaccharides & glycoproteins; polysialic acid production; cell adhesion
<i>SUV420H1</i>	histone methyltransferase; chromatin remodeling; transcription regulation
<i>SYN1</i>	vesicular regulation; cytoskeleton; neurotransmitter release
<i>TBR1</i>	transcription factor; transcription regulation
<i>TBX1</i>	transcription factor; transcription regulation
<i>TRIO</i>	guanine nucleotide exchange factor; GTPase regulation; kinase; transferase; cytoskeleton regulation
<i>TSC1</i>	tumor suppressor; intracellular signal transduction
<i>TSC2</i>	tumor suppressor; intracellular signal transduction; cytoskeletal-mediated transport; GTPase
<i>TTN</i>	cytoskeleton; chromosome condensation; chromosome segregation
<i>TUBA1A</i>	cytoskeleton; chromosome segregation
<i>TUBGCP5</i>	cytoskeleton; centromere formation
<i>UBE3A</i>	ubiquitination regulation
<i>UPF3B</i>	mRNA export; mRNA surveillance; translation regulation
<i>UPP2</i>	glycosyltransferase; production of carbon & energy sources; nucleotide synthesis; detoxification
<i>VIP</i>	ligand; hormone
<i>VPS13B</i>	vesicular regulation
<i>YWHAE</i>	adaptor protein; intracellular signal transduction
<i>ZNF674</i>	transcription factor; transcription regulation
<i>ZNF81</i>	transcription factor; transcription regulation