Supplementary information S1 (table) | **Clathrin-mediated endocytosis and human diseases**

Disease category	Protein	Gene	Disease*	Alterations**	References
Cancer	Clathrin heavy chain	*CLTC*	IMT	CLTC-ALK fusion	Bridge JA et al Am J Pathol 2001 159, 411
			Large B-cell lymphoma	CLTC-ALK fusion	De Paepe P et al. Blood 2003, 102, 2638
			Paedriatric renal carcinoma	CLTC-TFE3 fusion	Argani P et al. Oncogene 2003, 22, 5374
			Renal cancer	somatic mutation	Dalgliesh GL et al Nature 2010 463, 360
			Breast cancer	somatic mutations	Kan Z. et al. Nature 2010, 466, 869
	EPS15	*EPS15*	AML	somatic mutations, EPS15-MLL fusion	Bernard OA, et al. Oncogene 1994, 9, 1039
			Lung cancer	somatic mutations	Kan Z. et al. Nature 2010, 466, 869
	CALM	*PICALM*	ALL and AML	PICALM-AF10, -MLL fusions, som. mutations	Dreyling MH et al. PNAS, 1996, 93, 4804
	Dab2	*DAB2*	Ovarian cancer	Downregulated	Mok SC et al. Oncogene 1998, 16, 2381
			Prostate cancer	Downregulated	Tseng CP et al. Endocrinology 1998, 139, 3542
			Bladder cancer	Downregulated	Karam JA et al. Clin Cancer Res 2007, 13, 4400
			Breast cancer	Downregulated	Fazili Z et al. Oncogene 1999, 18, 3104
			Oesophageal cancer	Downregulated	Anupam K et al. World J Gastroenterol 2006, 12, 6041
			Colorectal carcinomas	Downregulated	Kleef J et al. Dis. Colon Rectum. 2002, 45, 1242
	Endophilin	*SH3GL1*	AML	EEN-MLL fusion, somatic mutations	So CW et al PNAS 1997, 94, 2563
	HIP1	*HIP1*	CMML	HIP1-PDGFRB fusion, somatic mutations	Ross TS et al. Blood 1998, 91, 4419
			Lung cancer	somatic mutations	Kan Z. et al. Nature 2010, 466, 869
			Breast cancer	somatic mutations	Kan Z. et al. Nature 2010, 466, 869
	HIP1R	*HIP1R*	Colon cancer	overexpressed	Scanian MJ et al. Cancer Res. 2002, 62, 4041
			CLL	overexpressed	Porpaczy E et al. Eur J Clin Invest 2009, 39, 568
	Cortactin	*CTTN*	Primary breast carcinomas	Amplification, protein overexpression	Schuuring E. et al. Oncogene, 1992, 7, 355
	βArrestin1	*ARRB1*	Breast cancer	somatic mutations	Kan Z. et al. Nature 2010, 466, 869
	Numb	*NUMB*	Breast cancer	Downregulated	Pece S. et al. J Cell Biol 2004, 167, 215
Psychiatric diseases	Amphiphysin	*BIN1*	Schizophrenia	increased expression	English JA et al. Proteomics 2009, 9, 3368
	Endophilin	*SH3GL1*	Schizophrenia	decreased expression	Prabakaran S et al. Mol Psychiatry, 9, 2004
	Dynamin	*DNM1*	Schizophrenia	increased expression	Prabakaran S et al. Mol Psychiatry, 2004, 9, 684 Clark D et al Mol Psychiatry 2006, 11, 459 Pennington K et al Mol Psychiatry, 2007, 13, 1102
	Synaptojanin	*SYNJ1*	Bipolar disorder	mutations	Saito T et al. Mol Psychiatry 2001, 6, 387
Neuropathies & Myopathies	Amphiphysin	*BIN1*	CNM	mutations	Nicot AS et al. Nat Genet. 2007, 39, 1134
	Dynamin	*DNM2*	CNM	mutations	Bitoum M et a; Nat Genet 2005, 37, 1207
			CMT	mutations	Zuchner S et al. Nat Genet 2005, 37, 289
Neurodegenerative diseases	CALM	*PICALM*	Alzheimer	SNPs	Harold D et al Nat Genet 2009, 41, 1088
	Amphiphysin	*BIN1*	Alzheimer	SNPs	Seshadri S et al. JAMA 2010, 303, 1832
Metabolic syndromes	ARH	*LDLRAP1*	Autosomal recessive hypercholesterolaemia	mutations	Garcia CK, et al Science 2001, 292, 1394
	SGIP1	*SGIP1*	Obesity	SNPs	Cummings N et al Int J Obes 2011, *in press*
	SHIP2	*INPP1L*	Type2 diabetes	deletion inducing increased expression	Clement S. et al. Nature 2001 409, 92
			Hypertension	mutations	Kaisaki PJ et al., Diabetes 2004, 53, 1900
			Central obesity	mutations	Kaisaki PJ et al., Diabetes 2004, 53, 1900
			Dyslipidaemia	mutations	Kaisaki PJ et al., Diabetes 2004, 53, 1900
Genetic syndromes	OCRL	*OCRL1*	Lowe Syndrome	X-linked, eye, nervous system, kidney disease	Attre O et al Nature 1992 358, 239
			Dent Syndrome	X-linked, renal only form of Lowe syndrome	Hoopes RR et al. Am J Hum Genet 2005, 76, 260
	Synaptojanin	*SYNJ1*	Down Syndrome	gene on Chrs 21 - increased expression	Arai Y et al. Brain Development 2002, 24, 67
	Intersectin	*ITSN1*	Down Syndrome	gene on Chrs 21 - increased expression	Pucharcos C. et al Eur J Hum Genet. 1999, 7, 704
	Dyrk1A	*DYRK1A*	Down Syndrome	gene on Chrs 21 - increased expression	Guimera J et al. Genomics 1999, 57, 407

* IMT, Inflammatory Myoblastic Tumour; AML, Acute Myeloid Leukaemia; ALL, Acute Lymphoblastic Leukaemia; CMML, Chronic Myelomonocytic Leukaemia; CLL, Chronic Lymphocytic Leukaemia; CNM, autosomal dominant centronuclear myopathy; CMT, dominant intermediate Charcot-Marie-Tooth disease

** alterations in human diseases