Chapter 15
Cadherin-Related Diseases

Shinji Hirano and Keiko Imai-Okano

This is a list for quick reference of cadherins in human diseases.

Please note that many unproven candidates and possible involvements (some are marked by *) are also listed. In addition, this is not a complete list and there are many omissions of citations.

15.1 List of Cadherin-Related Diseases

G Mendelian genetic, *M* multifactorial, *E* epigenetic, *A* autoimmune, *I* infection, *P* Expression profile, *X* experimental

Cadherins/Gene type		Diseases	References
Classic cadherins			
E-cadherin (CDH1)	**G**	Gastric cancer (HDGC) (OMIM137215)	Guilford et al. (1998)
	G	Lobular breast cancer (OMIM114480)	Masciari et al. (2007)
	G	Endometrial carcinoma (OMIM608089)	Risinger et al. (1994)
	G	Ovarian cancer (OMIM167000)	Risinger et al. (1994)
	M	Prostate cancer (OMIM176807)	Jonsson et al. (2004)
	M	Ulcerative colitis (UC)	Barrett et al. (2009)
	M	Asthma	Ierodiakonou et al. (2011)
	M	*Crohn disease (OMIM266600)	Elding et al. (2011)

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S. Hirano (✉) • K. Imai-Okano
Department of Cell Biology, Kansai Medical University, 2-5-1 Shinmachi, Hirakata, Osaka 573-1010, Japan
e-mail: s-hirano@hirakata.kmu.ac.jp; imaikei@hirakata.kmu.ac.jp

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Cadherins/Gene type		Diseases	References
	I	Candidiasis	Phan et al. (2007)
	I	Listeria infection	Mengaud et al. (1996)
	A	Pemphigus Foliaceus	Flores et al. (2012)
N-cadherin (CDH2)	**I**	Candidiasis	Phan et al. (2007)
P-cadherin (CDH3)	**G**	Hypotrichosis with juvenile macular dystrophy HJMD (OMIM601553)	Sprecher et al. (2001)
	G	EEM syndrome (OMIM225280)	Kjaer et al. (2005)
	M	*Crohn disease (OMIM266600)	Elding et al. (2011)
	M	Autism spectrum disorders (ASD)	O'Roak et al. (2012)
R-cadherin (CDH4)	**M**	Chronic kidney disease without diabetes mellius	Yoshida et al. (2010)
	M	Schizophrenia	Girard et al. (2011)
	E	*Colorectal and Gastric cancer	Miotto et al. (2004)
VE-cadherin (CDH5)	**M**	Central Serous Chorioretinopathy	Schubert et al. (2014)
	M	Autism spectrum disorders (ASD)	O'Roak et al. (2012)
	I	Infection by Leptospira	Evangelista et al. (2014)
	A	*Rheumatoid arthritis, Behçet's disease *Systemic lupus erythematosus	Bouillet et al. (2013)
K-cadherin (CDH6)	**M**	Autism spectrum disorders (ASD)	Butler et al. (2015)
	G	*Myopia 19	Ma et al. (2010)
CDH7	**M**	Bipolar disorders	Soronen et al. (2010)
	M	*Ectodermal dysplasia (OMIM 602401)	Tariq et al. (2008)
	M	Autism spectrum disorders (ASD)	O'Roak et al. (2012)
CDH8	**M**	Learning disabilities, autism	Pagnamenta et al. (2011)
CDH9	**M**	*Autism, ASD (OMIM209850)	Wang et al. (2009)
CDH10	**G**	*Myopia 19	Ma et al. (2010)
	M	*Autism, ASD (OMIM209850)	Wang et al. (2009)
CDH11	**E**	*Cancer metastasis	Carmona et al. (2012)
	M	Alcoholism	Johnson et al. (2006)
	M	Bipolar alcoholism	Lydall et al. (2011)
CDH12	**M**	Bipolar alcoholism	Lydall et al. (2011)
	M	Leptin level	Zhang et al. (2013)
	G	*Myopia 19	Ma et al. (2010)
	M	*Schizophrenia	Singh et al. (2010)
M-cadherin (CDH15)	**M**	Mental retardation (OMIM612580)	Bhalla et al. (2008)
	M	Autism	Willemsen et al. (2010)
CDH18	**M**	Metabolic syndrome	Zhang et al. (2013)
	M	Colorectal cancer	Venkatachalam et al. (2011)
	M	*Schizophrenia	Singh et al. (2010)
CDH19	**M**	*Ectodermal dysplasia (OMIM 602401)	Tariq et al. (2008)

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Cadherins/Gene type		Diseases	References
CDH20	**M**	Barrett's adenocarcinoma	Wiech et al. (2009)
PB-cadherin (CDH22)	**M**	Type2 diabetes	Bento et al. (2008)
	M	Kawasaki disease	Shendre et al. (2014)
Desmosomal cadherins			
desmoglein 1 (DSG1)	**G**	Palmoplantar Keratoderma (SPPK). (OMIM 148700)	Rickman et al. (1999)
	G	Congenital erythroderma with palmoplantar keratoderma, hypotrichosis, and hyper-IgE (EPKHE) (OMIM615508) = SAM syndrome	Samuelov et al. (2013)
	A	Pemphigus foliaceus	Eyre and Stanley (1987)
	A	Pemphigus vulgaris	Amagai et al. (1991)
	A	Paraneoplastic pemphigus	Amagai et al. (1998)
	I	Staphylococcal scalded-skin syndrome (SSSS)	Amagai et al. (2000)
desmoglein 2 (DSG2)	**G**	Arrhythmogenic right ventricular cardiomyopathy 10 (ARVC10) (OMIM 610193)	Pilichou et al. (2006)
	G	Cardiomyopathy, dilated, 1BB (CMD1BB) (OMIM 612877)	Posch et al. (2008)
	X	*Colon cancer progression	Kamekura et al. (2014)
desmoglein 3 (DSG3)	**A**	Pemphigus vulgaris	Amagai et al. (1999)
	A	Paraneoplastic pemphigus	Amagai et al. (1998)
	P	*Cancer progression	Brown and Wan (2015)
DSG4	**G**	Localized autosomal recessive hypotrichosis (LAH1) (Hypotrichosis 6 OMIM 607903)	Kljuic et al. (2003)
	G	Monilethrix-like congenital hypotrichosis	Shimomura et al. (2006)
	A	Pemphigus foliaceus	Nagasaka et al. (2004)
	A	Pemphigus vulgaris	Nagasaka et al. (2004)
desmocollin 2 (DSC2)	**G**	Arrhythmogenic right ventricular dysplasia-11 (ARVD11, OMIM 610476)	Syrris et al. (2006)
	G	ARVD with mild palmoplantar keratoderma and woolly hair (Naxos disease)	Simpson et al. (2009)
	X	*Cancer progression	Kolegraff et al. (2011)
desmocollin 3 (DSC3)	**G**	Hypotrichosis and recurrent skin vesicles (OMIM 613102)	Ayub et al. (2009)
	A	Pemphigus vulgaris	Mao et al. (2010)

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Cadherins/Gene type		Diseases	References
T-cadherin			
T-cadherin (CDH13)	**M**	Serum adiponectin level	Ling et al. (2009) and Jee et al. (2010)
	M	Metabolic syndrome	Fava et al. (2011)
	M	Hypertension	Org et al. (2009)
	M	Lung function in air pollution	Imboden et al. (2015)
	M	Height	Axenovich et al. (2009)
	M	*Hirschsprung disease	Carrasquillo et al. (2002)
	M E	Lung cancer	Brock et al. (2008)
	M	*Retinoblastomas	Gratias et al. (2007)
	M	Prostate cancer	Thomas et al. (2008)
	E	Non-small cell lung cancer (NSCLC)	Pesek et al. (2011)
	M	Alcoholism	Johnson et al. (2006) and Treutlein et al. (2009)
	M	d-Amphetamine response	Hart et al. (2012)
	M	Methamphetamine dependence	Uhl et al. (2008b)
	M	Addiction-related phenotypes	Uhl et al. (2008a)
	M	Autism, Autism spectrum disorders (ASD)	Chapman et al. (2011)
	M	Attention deficit hyperactivity disorder (ADHD)	Lasky-Su et al. (2008) and Lesch et al. (2008)
	M	Personality	Terracciano et al. (2010)
	M	Childhood asthma	Ding et al. (2013)
Protocadherin			
PCDH1	**M**	Bronchial hyperresponsiveness (BHR) (asthma-related traits)	Koppelman et al. (2009)
	M	Eczema	Koning et al. (2012)
PCDH cluster	**E**	*Wilms' tumor	Dallosso et al. (2009)
	E	*Colorectal cancer	Dallosso et al. (2012)
		*Cervical cancer	Wang et al. (2015)
PCDHA cluster	**M**	Schizophrenia and bipolar disorder	Lachman et al. (2008)
	M	Bipolar disorder	Pedrosa et al. (2008)
	M	Autism	Anitha et al. (2013)
PCDHA3	**M**	*Restless legs syndrome (RLS8) (OMIM 615197)	Weissbach et al. (2012)
PCDHB4	**M**	Autism spectrum disorders (ASD)	O'Roak et al. (2012)
	M	Microcephaly	Alazami et al. (2015)
PCDH7	**M**	Epilepsy	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies (2014)
	M	Body shape and cholesterol level	Surakka et al. (2011)

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Cadherins/Gene type		Diseases	References
	M	Survival in non-small cell lung cancer	Huang et al. (2009)
	M	Sleep duration	Ollila et al. (2014)
PCDH8	**M**	Autism spectrum disorders (ASD)	Butler et al. (2015)
	M	Cognition in the presence of type2 diabetes	Cox et al. (2014)
	E	*Renal cell carcinoma	Morris et al. (2011)
	P	*B-cell chronic lymphocytic leukemia	Mittal et al. (2007)
	E	*Bladder cancer	Lin et al. (2013)
	G?	*Retinoblastoma and mental retardation microdeletion syndrome	Caselli et al. (2007)
PCDH9	**M**	ASD	Marshall et al. (2008)
PCDH10	**E**	Testicular cancer	Cheung et al. (2010)
	M	Autism	Morrow et al. (2008)
PCDH11X	**M**	Developmental dyslexia	Veerappa et al. (2013)
	M	*Schizophrenia, cerebral asymmetry	Levchenko et al. (2014)
	M	*Tourette syndrome (OMIM 137580)	Melchior et al. (2013)
	M	Late-onset Alzheimer's disease	Carrasquillo et al. (2009)
PCDH11X, 11Y	**M**	Languate delay	Speevak and Farrell (2011)
PCDH12	**M**	Schizophrenia	Gregorio et al. (2009)
PCDH17	**E**	Prostate cancer	Lin et al. (2014)
	E	Bladder cancer	Wang et al. (2014)
	G?	*Retinoblastoma and mental retardation microdeletion syndrome	Caselli et al. (2007)
PCDH18	**M**	Autism	Morrow et al. (2008)
PCDH19	**G**	Early infantile epileptic encephalopathy-9 (EIEE9) = epilepsy and mental retardation restricted to females (EFMR) (OMIM 300088)	Dibbens et al. (2008)
		with Dvavet syndrome-like features	Depienne et al. (2009) and Depienne and LeGuern (2012)
		with autism and cognitive impairment	Depienne and LeGuern (2012) and Camacho et al. (2012)
PCDH20	**E**	Nasopharyngeal carcinoma	Chen et al. (2015)
	M	Sphingolipid metabolism	Demirkan et al. (2012)

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Cadherins/Gene type		Diseases	References
FAT			
FAT1	**M**	*Facioscapulohumeral dystrophy-like	Puppo et al. (2015)
	M	Autism spectrum disorders (ASDs)	Cukier et al. (2014)
	M	4q-syndrome	Sadeqzadeh et al. (2014)
	M	*Cervical cancer	Chung et al. (2015)
	P	Intrahepatic cholangiocarcinoma	Settakorn et al. (2005)
	G P	Oral cancer	Nakaya et al. (2007)
	P	*Breast cancer	Kwaepila et al. (2006)
	M P	Various carcinomas and tumors	Sadeqzadeh et al. (2014)
	M	Bipolar affective disorder (BPAD)	Blair et al. (2006)
FAT2	**M**	Autism	Butler et al. (2015)
	M	*Restless legs syndrome (RLS8) (OMIM 615197)	Weissbach et al. (2012)
	M	Pancreatic cancer	Tang et al. (2014)
	M	*Colorectal cancer	Xie et al. (2014)
FAT3	**P**	*Lung cancer	Rohrbeck and Borlak (2009)
FAT4	**M**	*Lung cancer	Berndt et al. (2011)
	M	Esophageal cancer	Du et al. (2013)
	M	Gastric cancer	Wadhwa et al. (2013)
	M	Melanoma	Nikolaev et al. (2012)
	P	Breast cancer	Qi et al. (2009)
	G	Van Maldergem syndrome-2 (VMLDS2)	Cappello et al. (2013)
	G	*Hennekam lymphangiectasia-lymphedema syndrome-2 (HKLLS2; OMIM616006) (allele of VMLDS2)	Alders et al. (2014)
Dachsous			
DCHS1	**G**	Van Maldergem syndrome-2 (VMLDS2)	Cappello et al. (2013)
DCHS2	**M**	Alzheimer's disease	Kamboh et al. (2012)
	M	Osteoporosis	Han et al. (2012)
7D-cadherin			
LI-cadherin (CDH17)	**M**	Hepatocellular carcinoma	Wang et al. (2006)
	P	Gastric cancer	Oue et al. (2004) and Lee et al. (2010)
	M	Colorectal carcinoma	Chen et al. (2012)
	P M	Various cancers	Weissbach et al. (2012)
	M	Hypertension	Zhu et al. (2015)

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Cadherins/Gene type		Diseases	References
Calsyntenin			
CLSTN1 Calsyntenin-1	**M**	Pancreatic cancer	Tang et al. (2014)
	E	Prostate, colon, breast cancer	Chung et al. (2015)
	M	Lung cancer	Langer et al. (2010)
CLSTN2 Calsyntenin-2	**M**	Alzheimer's disease (AD15) (OMIM611155)	Liu et al. (2007)
Inner ear cadherins			
CDH23	**G**	Usher syndrome ID (OMIM 601067)	Bolz et al. (2001) and Bork et al. (2001)
	G	Usher syndrome type ID/F (OMIM 601067)	Zheng et al. (2005)
	G	Autosomal recessive deafness 12 (DFNB12) (OMIM601386)	Bork et al. (2001) and Schultz et al. (2005)
	M	Personality	Terracciano et al. (2010)
	M	Chronic kidney disease (CKD)	Gorski et al. (2015)
	E	Alzheimer's disease	De Jager et al. (2014)
PCDH15	**G**	Usher syndrome IF (OMIM 602083)	Ahmed et al. (2001)
	G	Deafness, autosomal recessive 23 (DFNB23) (OMIM 609533)	Ahmed et al. (2003)
	M	Extrapulmonary tuberculosis	Oki et al. (2011)
	M	Late-onset Alzheimer's disease (LOAD)	Fallin et al. (2010)
	M	Familial combined hyperlipid-emia (FCHL)	Huertas-Vazquez et al. (2010)
	M	Retinal dystrophies	Coppieters et al. (2014)
	M	Antibody response to smallpox vaccine	Ovsyannikova et al. (2012)
PCDH21 (CDHR1)	**G**	Cone-rod dystrophy15 (OMIM613660)	Ostergaard et al. (2010)
		Retinitis pigmentosa 65	Henderson et al. (2010)
CDHR3 (CDH28)	**M**	Asthma	Bonnelykke et al. (2014)
CDH26	**M**	Asthma	Ferreira et al. (2009)
RET	**G**	Hirschsprung disease	Attie et al. (1995) and Angrist et al. (1995)
	G	Central hypoventilation syndrome (OMIM 209880)	Bolk et al. (1996)
	G	Multiple endocrine neoplasia, Type II (OMIM 171400, 162300)	Shirahama et al. (1998)
	G	Familial medullary thyroid carcinoma	Elisei et al. (2007)
	G	Pheochromocytoma (OMIM 171300)	Eng et al. (1995)
	G	Renal agenesis (OMIM 191830)	Skinner et al. (2008)

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Cadherins/Gene type		Diseases	References
	M	Vesicoureteral reflux (OMIM 193000)	Yang et al. (2008)
	G	Congential anomalies of the kidney or urinary tract (CAKUT)	Hwang et al. (2014)
	G	Various other cancers	Mulligan 2014)
CELSR1	**M**	Spina bifida	Lei et al. (2014)
	M	Ischemic stroke	Yamada et al. (2009)
	M	Hypertension	Ueyama et al. (2013)
CELSR2	**M**	*Serum lipid	Wallace et al. (2008), Kathiresan et al. (2008) and Samani et al. (2008)
	M	*Coronary heart disease	Ronald et al. (2009) and Kathiresan et al. (2009)
CELSR3	**E**	Oral squamous cell carcinoma	Khor et al. (2014)

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