

Table S2 Genes (listed alphabetically by symbol) in which mutations have been shown to result in Mendelian traits/disorders in non-rodent animals, up to the end of 2012. Further details (including references) for each entry are available at <http://omia.angis.org.au>. An up-to-date version of this table is available from the "Download" tab on the OMIA home page.

Gene symbol	Gene name	Trait/disorder	Species	OMIA number
ABCA1	ATP-binding cassette, sub-family A (ABC1), member 1	Analphalipoproteinaemia	chicken	000042
ABCA12	ATP-binding cassette, sub-family A (ABC1), member 12	Ichthyosis congenita	cattle	000547
ABCB1	ATP-binding cassette, sub-family B (MDR/TAP), member 1	Multidrug resistance 1	dog	001402
ABCB4	ATP-binding cassette, sub-family B (MDR/TAP), member 4	Gallbladder mucoceles	dog	001524
ABCG2	ATP-binding cassette, sub-family G (WHITE), member 2	Retinal degeneration, fluoroquinolone-induced	cat	001707
ABO		Blood group system ABO	Japanese macaque	001089
		Blood group system ABO	chimpanzee	001089
		Blood group system ABO	Rhesus monkey	001089
		Blood group system ABO	crab-eating macaque	001089
		Blood group system ABO	agile gibbon	001089
		Blood group system ABO	common gibbon	001089
		Blood group system ABO	siamang	001089
ACAN	aggrecan	Nanomelia Dwarfism, Dexter	chicken cattle	000702 001271
ADAM9	ADAM metallopeptidase domain 9	Cone-rod dystrophy 3	dog	001520
ADAMTS10	ADAM metallopeptidase with thrombospondin type 1 motif, 10	Glaucoma	dog	000411
ADAMTS17	ADAM metallopeptidase with thrombospondin type 1 motif, 17	Lens luxation	dog	000588
ADAMTS2	ADAM metallopeptidase with thrombospondin type 1 motif, 2	Ehlers-Danlos syndrome, type VII (Dermatosparaxis)	cattle	000328
		Ehlers-Danlos syndrome, type VII (Dermatosparaxis)	sheep	000328
ADAMTSL2	ADAMTS-like 2	Musladin-Lueke syndrome	dog	001509
AGL	amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase	Glycogen storage disease IIIa	dog	001577
AGTPBP1	ATP/GTP binding protein 1	Motor neuron disease, lower	sheep	000662
AGXT	alanine-glyoxylate aminotransferase	Primary hyperoxaluria type I (Oxalosis I)	dog	001672
AMHR2	anti-Mullerian hormone receptor, type II	Persistent Mullerian duct syndrome	dog	000791
AMN	amniotless homolog (mouse)	Intestinal cobalamin malabsorption due to AMN mutation	dog	000565

AP3B1	adaptor-related protein complex 3, beta 1 subunit	Neutropenia, cyclic	dog	000248
APAF1	apoptotic peptidase activating factor 1	Abortion due to haplotype HH1	cattle	000001
AR	androgen receptor	Androgen insensitivity syndrome (AIS)	horse	000991
ARSB	arylsulfatase B	Mucopolysaccharidosis VI	cat	000666
		Mucopolysaccharidosis VI	dog	000666
ARSG	arylsulfatase G	Neuronal ceroid lipofuscinosis, 4A	dog	001503
ASIP	agouti signaling protein	Coat colour, agouti	horse	000201
		Coat colour, agouti	cattle	000201
		Coat colour, agouti	red fox	000201
		Coat colour, agouti	sheep	000201
		Coat colour, agouti	cat	000201
		Coat colour, agouti	rabbit	000201
		Coat colour, agouti	dog	000201
		Feather colour, yellow	Japanese quail	001602
		Coat colour, agouti	North American deer mouse	000201
		Coat colour, agouti	leopard	000201
		Coat colour, agouti	Asiatic golden cat	000201
ASS1	argininosuccinate synthase 1	Citrullinaemia	cattle	000194
ATF2	activating transcription factor 2	Neonatal encephalopathy with seizures	dog	001471
ATP13A2	ATPase type 13A2	Neuronal ceroid lipofuscinosis, adult-onset, Tibetan Terrier	dog	001552
ATP2A1	ATPase, Ca ⁺⁺ transporting, cardiac muscle, fast twitch 1	Congenital muscular dystonia 1	cattle	001450
		Pseudomyotonia, congenital	cattle	001464
Amhr2		Sex determination	torafugu	001728
BCAN	brevican	Episodic falling	dog	001592
BCKDHA	branched chain keto acid dehydrogenase E1, alpha polypeptide	Maple syrup urine disease	cattle	000627
BCO2	beta-carotene oxygenase 2	Yellow fat	sheep	001079
		Skin colour, yellow	chicken	001449
		Yellow fat	cattle	001079
BCR	breakpoint cluster region	Leukemia, chronic monocytic	dog	001573
BEST1	bestrophin 1	Multifocal retinopathy 1	dog	001444
		Multifocal retinopathy 2	dog	001553
		Multifocal retinopathy 3	dog	001554
BMP15	bone morphogenetic protein 15	Fecundity, Galway, FecX(G)	sheep	000384
		Fecundity, Inverdale, FecX(I)	sheep	000386
		Fecundity, Hanna, FecX(H)	sheep	001350
		Fecundity, Lacaune, FecX(L)	sheep	001357
		Fecundity, Belclare, FecX(B)	sheep	001719
		Fecundity, Rasa Aragonesa, FecX(R)	sheep	001800
BMP3	bone morphogenetic protein 3	Brachycephaly	dog	001551

BMPR1B	bone morphogenetic protein receptor, type IB	Fecundity, Booroola, FecB(B)	sheep	000383
		Fecundity, small tail Han	sheep	001423
BTN1A1	butyrophilin, subfamily 1, member A1	Resistance to avian sarcoma and leukosis viruses, subgroup C	chicken	001622
C3	complement component 3	C3 deficiency	dog	000155
CAT	catalase	Hypocatalasia	dog	001138
CBD103	beta-defensin 103	Coat colour, dominant black	dog	001416
		Coat colour, dominant black	gray wolf	001416
		Coat colour, dominant black	coyote	001416
CCDC39	coiled-coil domain containing 39	Ciliary dyskinesia, primary	dog	001540
CCDC66	coiled-coil domain containing 66	Generalized PRA	dog	001521
CD320	CD320 molecule	Resistance to avian sarcoma and leukosis viruses, subgroup A	chicken	001299
CDKN2B	cyclin-dependent kinase inhibitor 2B (p15, inhibits CDK4)	Barring	chicken	000102
CEP290	centrosomal protein 290kDa	Retinal degeneration II	cat	001244
CFH	complement factor H	Membranoproliferative glomerulonephritis type II	pig	000636
CFTR	cystic fibrosis transmembrane conductance regulator (ATP-binding cassette sub-family C, member 7)	Cystic fibrosis	pig	001794
		Cystic fibrosis	ferret	001794
CHAT	choline O-acetyltransferase	Myasthenic syndrome, congenital	dog	000685
CHRNE	cholinergic receptor, nicotinic, epsilon (muscle)	Myasthenic syndrome, congenital	cattle	000685
CLCN1	chloride channel, voltage-sensitive 1	Myotonia	goat	000698
		Myotonia	dog	000698
		Myotonia	horse	000698
CLDN16	claudin 16	Renal dysplasia	cattle	001135
CLN5	ceroid-lipofuscinosis, neuronal 5	Neuronal ceroid lipofuscinosis, 5	sheep	001482
		Neuronal ceroid lipofuscinosis, 5	dog	001482
		Neuronal ceroid lipofuscinosis, 5	cattle	001482
CLN6	ceroid-lipofuscinosis, neuronal 6, late infantile, variant	Neuronal ceroid lipofuscinosis, 6	sheep	001443
		Neuronal ceroid lipofuscinosis, 6	dog	001443
CLN8	ceroid-lipofuscinosis, neuronal 8 (epilepsy, progressive with mental retardation)	Neuronal ceroid lipofuscinosis, 8	dog	001506

CMAH	cytidine monophospho-N-acetylneuraminic acid hydroxylase	Blood group system AB	cat	000119
CNGA3	cyclic nucleotide gated channel alpha 3	Achromatopsia-2	sheep	001481
CNGB3	cyclic nucleotide gated channel beta 3	Achromatopsia (cone degeneration, hemeralopia), AMAL	dog	001365
		Achromatopsia (cone degeneration, hemeralopia), GSPT	dog	001676
COL10A1	collagen, type X, alpha 1	Dwarfism, Schmid metaphyseal chondrodysplasia	pig	001718
COL1A2	collagen, type I, alpha 2	Osteogenesis imperfecta	dog	000754
COL4A4	collagen, type IV, alpha 4	Nephropathy	dog	000710
COL4A5	collagen, type IV, alpha 5	Nephritis, X-linked	dog	001112
COL7A1	collagen, type VII, alpha 1	Epidermolysis bullosa, dystrophic	dog	000341
		Epidermolysis bullosa, dystrophic	cattle	000341
COL9A2	collagen, type IX, alpha 2	Oculoskeletal dysplasia 2	dog	001523
COL9A3	collagen, type IX, alpha 3	Oculoskeletal dysplasia 1	dog	001522
COMMD1	copper metabolism (Murr1) domain containing 1	Wilson disease	dog	001071
CRX	cone-rod homeobox	Rod-cone dysplasia	cat	000881
CSN1S1	casein alpha s1	Casein, alpha-S1, reduced concentration	goat	000852
CSN1S2	casein alpha s2	Casein, alpha-S2, absence	goat	001623
CSN2	casein beta	Casein, beta, absence of	goat	001424
CTSD	cathepsin D	Neuronal ceroid lipofuscinosis, 10	dog	001505
		Neuronal ceroid lipofuscinosis, 10	sheep	001505
CWC15	CWC15 spliceosome-associated protein homolog (<i>S. cerevisiae</i>)	Abortion due to haplotype JH1	cattle	001697
CYP11A1	cytochrome P450, family 11, subfamily A, polypeptide 1	Adrenal hyperplasia, congenital	rabbit	000017
CYP11B1	cytochrome P450, family 11, subfamily B, polypeptide 1	Adrenal hyperplasia, congenital, due to 11-beta hydroxylase deficiency	cat	001661
CYP19A1	cytochrome P450, family 19, subfamily A, polypeptide 1	Henny feathering	chicken	000452
CYP1A2	cytochrome P450, family 1, subfamily A, polypeptide 2	Metabolizer of a cognitive enhancer	dog	001405
CYP27B1	25-hydroxyvitamin D3 1alpha-hydroxylase	Vitamin D-deficiency rickets, type I	pig	000837
		Vitamin D-deficiency rickets, type I	cat	000837
CYTB	cytochrome b	Leucodystrophy	dog	001130
ClRIG-I	ctenopharyngodon idella retinoic acid-inducible gene I	Resistance to grass carp reovirus	grass carp	001725
DLK1	delta-like 1 homolog (<i>Drosophila</i>)	Muscular hypertrophy (double muscling), Callipyge	sheep	001354

DMD	Duchenne muscular dystrophy	Muscular dystrophy, Duchenne type	cat	001081
		Muscular dystrophy, Duchenne type	dog	001081
		Stress syndrome	pig	001685
DMP1	dentin matrix acidic phosphoprotein 1	Hypophosphatemic rickets, autosomal recessive, 1	sheep	001542
DMRT1	doublesex and mab-3 related transcription factor 1	XY Ovotesticular DSD (Disorder of Sexual Development)	Japanese medaka	001709
DNAPK	DNA-dependent protein kinase catalytic subunit	Severe combined immunodeficiency disease, autosomal	horse	000220
DNM1	dynamain 1	Exercise-induced collapse	dog	001466
DPYS	dihydropyrimidinase	Dihydropyrimidinase deficiency	cat	001776
EDA	ectodysplasin A	Anhidrotic ectodermal dysplasia	cattle	000543
		Anhidrotic ectodermal dysplasia	dog	000543
EDN3	endothelin 3	Silky/Silkie pigmentation (Fibromelanosis)	chicken	001671
EDNRB	endothelin receptor type B	Megacolon	horse	000629
EDNRB2	endothelin receptor B subtype 2	Feather colour, panda/dotted white	Japanese quail	000375
EPYC	epiphycan	Ehlers-Danlos syndrome, Holstein variant	cattle	001716
ETBR	endothelin B receptor	Hypopigmentation syndrome, lethal	sheep	001765
EVC2	Ellis van Creveld syndrome 2	Chondrodysplasia	cattle	000187
F11	coagulation factor XI	Factor XI deficiency	cattle	000363
		Factor XI deficiency	dog	000363
F7	coagulation factor VII (serum prothrombin conversion accelerator)	Factor VII deficiency	dog	000361
F8	coagulation factor VIII, procoagulant component	Haemophilia A	dog	000437
		Haemophilia A	sheep	000437
		Haemophilia A	cattle	000437
F9	coagulation factor IX	Haemophilia B	cat	000438
		Haemophilia B	dog	000438
FAM83H	family with sequence similarity 83, member H	Congenital keratoconjunctivitis sicca and ichthyosiform dermatosis	dog	001683
FANCI	Fanconi anemia, complementation group I	Brachyspina	cattle	000151
FBN1	fibrillin 1	Marfan syndrome	cattle	000628
FECH	ferrochelatase	Protoporphyrria	cattle	000836
FERMT3	fermitin family member 3	Leukocyte adhesion deficiency, type III	dog	001525
FGF20	fibroblast growth factor 20	Scaleless	chicken	000889
FGF4	fibroblast growth factor 4	Chondrodysplasia	dog	000187

FGF5	fibroblast growth factor 5	Dermoid sinus	dog	000272
		Hair length	cat	000439
FGFR3	fibroblast growth factor receptor 3	Hair length	dog	000439
		Chondrodysplasia, Spider lamb	sheep	001703
FLCN	folliculin	Renal cystadenocarcinoma and nodular dermatofibrosis	dog	001335
FMO3	flavin containing monooxygenase 3	Trimethylaminuria	cattle	001360
FMO6P	flavin containing monooxygenase 6 pseudogene	Trimethylaminuria	chicken	001360
FOXI3	forkhead box I3	Ectodermal dysplasia	dog	000323
FUCA1	fucosidase, alpha-L- 1, tissue	Fucosidosis, alpha	dog	000396
FUT1	fucosyltransferase 1 (galactoside 2-alpha-L-fucosyltransferase, H blood group)	Resistance to oedema disease (F18 receptor)	pig	000862
G6PC	glucose-6-phosphatase, catalytic subunit	Glycogen storage disease Ia	dog	000418
GAA	glucosidase, alpha; acid	Glycogen storage disease II	cattle	000419
GALC	galactosylceramidase	Krabbe disease	dog	000578
		Krabbe disease	Rhesus monkey	000578
GBA	glucosidase, beta, acid	Gaucher disease, type I	sheep	000405
GBE1	glucan (1,4-alpha-), branching enzyme 1	Glycogen storage disease IV	cat	000420
		Glycogen storage disease IV	horse	000420
GC1	photoreceptor guanylate cyclase 1	Retinal degeneration I	chicken	001245
GDF7	growth differentiation factor 7	Naked neck	chicken	000701
GDF9	growth differentiation factor 9	Fecundity, Thoka, FecG(T)	sheep	000385
		Fecundity, Embrapa, FecG(E)	sheep	001559
		Fecundity, Norwegian White Sheep	sheep	001763
		Fecundity, High fertility, FecG(H)	sheep	001801
GGTA1	glycoprotein, alpha-galactosyltransferase 1	Blood group system ABO	pig	001089
GH1	growth hormone 1	Dwarfism, growth-hormone deficiency	cattle	001473
GLB1	galactosidase, beta 1	Gangliosidosis, GM1	cat	000402
		Gangliosidosis, GM1	dog	000402
		Gangliosidosis, GM1	sheep	000402
GLRA1	glycine receptor, alpha 1	Myoclonus	cattle	000689
GM2A	GM2 ganglioside activator	Gangliosidosis, GM2, GM2A deficiency	cat	001427
GNB3	guanine nucleotide binding protein (G protein), beta polypeptide 3	Retinopathy globe enlarged	chicken	001368
GNTTAB	N-acetylglucosamine-1-phosphate transferase, alpha and beta subunits	Mucopolipidosis II	cat	001248
GNS	glucosamine (N-acetyl)-6-sulfatase	Mucopolysaccharidosis IIID	goat	000665
GRHPR	glyoxylate reductase/hydroxypyruvate reductase	Primary hyperoxaluria type II (Oxalosis II)	cat	000821
GRM1	glutamate receptor, metabotropic 1	Ataxia, cerebellar	dog	000078
GUSB	glucuronidase, beta	Mucopolysaccharidosis VII	cat	000667
		Mucopolysaccharidosis VII	dog	000667

GYS1	glycogen synthase 1 (muscle)	Polysaccharide storage myopathy/Exertional rhabdomyolysis	horse	001158
HAS2	hyaluronan synthase 2	Periodic Fever Syndrome	dog	001561
HCRTR2	hypocretin (orexin) receptor 2	Narcolepsy	dog	000703
HEPHL1	hephaestin-like 1	Hypotrichosis	cattle	000540
HEXA	Hexosaminidase A (alpha polypeptide)	Gangliosidosis, GM2, type I	American flamingo	001461
HEXB	hexosaminidase B (beta polypeptide)	Gangliosidosis, GM2, type I	sheep	001461
		Gangliosidosis, GM2, type II	cat	001462
		Gangliosidosis, GM2, type II	dog	001462
HMBS	hydroxymethylbilane synthase	Porphyria, acute intermittent	cat	001493
HR	hairless homolog (mouse)	Hypotrichosis	sheep	000540
		Atrichia with papular lesions	Rhesus monkey	001348
HSF4	heat shock transcription factor 4	Cataract, early onset	dog	001758
HSPA1A	heat shock 70kDa protein 1A	Myopathy of the diaphragmatic muscles	cattle	001319
IDUA	iduronidase, alpha-L-	Mucopolysaccharidosis I	dog	000664
		Mucopolysaccharidosis I	cat	000664
IL2RG	interleukin 2 receptor, gamma	Severe combined immunodeficiency disease, X-linked	dog	000899
ITGA2B	integrin, alpha 2b (platelet glycoprotein IIb of IIb/IIIa complex, antigen CD41)	Thrombasthenia	dog	001000
		Thrombasthenia	horse	001000
ITGB2	integrin, beta 2 (complement component 3 receptor 3 and 4 subunit)	Leukocyte adhesion deficiency, type I	cattle	000595
		Leukocyte adhesion deficiency, type I	dog	000595
JAK2	Janus kinase 2	Polycythemia	dog	000809
KDM2B	lysine (K)-specific demethylase 2B	Lethal multi-organ developmental dysplasia	cattle	001722
KDSR	3-ketodihydrosphingosine reductase	Spinal muscular atrophy	cattle	000939
KIAA0586	KIAA0586	Talpid-3	chicken	000979
KIT	v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog	Coat colour, dominant white	pig	000209
		Coat colour, dominant white	cattle	000209
		Coat colour, dominant white	horse	000209
		Coat colour, roan	pig	001216
		Gastrointestinal stromal tumor	dog	001516
		Coat colour, colour-sided	cattle	001576
		Feet, white	cat	001580
		Coat colour, white spotting, due to KIT	dog	001737
		Coat colour, patch	pig	001743
		Coat colour, white belt, due to KIT	pig	001745

KITLG	KIT ligand	Coat colour, roan	cattle	001216
KLKB1	kallikrein B, plasma (Fletcher factor) 1	Prekallikrein deficiency	dog	000819
KRT10	keratin 10	Hyperkeratosis, epidermolytic	dog	001415
KRT5	keratin 5	Epidermolysis bullosa	cattle	000340
KRT71	keratin 71	Curly coat	dog	000245
		Curly coat, Devon rex	cat	001581
		Hypotrichosis, with whiskers short and curled	cat	001583
KRT75	keratin 75	Frizzle	chicken	000394
L2HGDH	L-2-hydroxyglutarate dehydrogenase	L-2-hydroxyglutaricacidemia	dog	001371
LAMA3	laminin, alpha 3	Epidermolysis bullosa, junctionalis, LAMA3	dog	001677
		Epidermolysis bullosa, junctionalis, LAMA3	horse	001677
LAMC2	laminin, gamma 2	Epidermolysis bullosa, junctionalis, LAMC2	sheep	001678
		Epidermolysis bullosa, junctionalis, LAMC2	horse	001678
LDLR	low density lipoprotein receptor	Hypercholesterolaemia	pig	000499
		Hyperlipidaemia	rabbit	001160
LGI2	leucine-rich repeat LGI family, member 2	Epilepsy, benign familial juvenile	dog	001596
LHX3	LIM homeobox 3	Dwarfism, pituitary	dog	000307
LIPH	lipase, member H	Rex coat	rabbit	001566
LIX1	Lix1 homolog (chicken)	Spinal muscular atrophy	cat	000939
LMBR1	limb region 1 homolog (mouse)	Oligozeugodactyly	chicken	001612
LOC100147177	doublesex- and mab-3-related transcription factor 3-like	Gaitedness	horse	001715
LOC430486	similar to Ca ²⁺ regulator SV2A	Epilepsy	chicken	000344
LPL	lipoprotein lipase	Hyperlipoproteinaemia	cat	001210
LRP4	low density lipoprotein receptor-related protein 4	Syndactyly (mule foot)	cattle	000963
LYST	lysosomal trafficking regulator	Chediak-Higashi syndrome	cattle	000185
		Chediak-Higashi syndrome	American mink	000185
MAN2B1	mannosidase, alpha, class 2B, member 1	Mannosidosis, alpha	cattle	000625
		Mannosidosis, alpha	cat	000625
MANBA	mannosidase, beta A, lysosomal	Mannosidosis, beta	cattle	000626
		Mannosidosis, beta	goat	000626
MC1R	melanocortin 1 receptor (alpha melanocyte stimulating hormone receptor)	Feather colour, extended black	chicken	000374
		Coat colour, extension	cattle	001199
		Coat colour, extension	dog	001199
		Coat colour, extension	horse	001199
		Coat colour, extension	red fox	001199
		Coat colour, extension	pig	001199
		Coat colour, extension	sheep	001199
		Coat colour, extension	jaguar	001199
		Coat colour, extension	jaguarundi	001199

		Coat colour, extension	American black bear	001199
		Feather colour, extended brown	Japanese quail	001435
		Coat colour, extension	woolly mammoth	001199
		Feather colour, extended black	red-footed booby	000374
		Feather colour, extended black	Bananaquit	000374
		Coat colour, extension	cat	001199
		Coat colour, extension	rabbit	001199
		Coat colour, extension	Arctic fox	001199
		Coat colour, extension	gray squirrel	001199
		Coat colour, extension	little striped whiptail	001199
		Coat colour, extension	water buffalo	001199
		Feather colour, bronze	turkey	001494
		Coat colour, grizzle	dog	001495
		Coat colour, extension	yak	001199
		Coat colour, melanistic mask	dog	001590
		Feather colour, extended black	helmeted guineafowl	000374
		Feather colour, extended black	gyrfalcon	000374
		Feather colour, extended black	Lesser snow goose	000374
		Feather colour, extended black		000374
		Feather colour, extended black		000374
		Feather colour, extended black	brown booby	000374
MFN2	mitofusin 2	Axonopathy	cattle	001106
		Neuroaxonal dystrophy	dog	000715
MIMT1	MER1 repeat containing imprinted transcript 1 (non-protein coding)	Abortion and stillbirth due to mutation in MIMT1	cattle	001565
MITF	microphthalmia-associated transcription factor	Feather colour, silver	Japanese quail	000915
		Osteopetrosis	Japanese quail	000755
		Waardenburg syndrome, type IIA	golden hamster	001401
		Coat colour, white spotting	dog	000214
		Coat colour, white spotting	cattle	000214
		Dominant white with bilateral deafness	cattle	001680
		Coat colour, white spotting	horse	000214
		Coat colour, macchiato	horse	001688

MLPH	melanophilin	Deafness	pig	000259
		Alopecia, colour mutant	dog	000031
		Coat colour, dilute	cat	000206
		Feather colour, lavender	chicken	001445
		Feather colour, lavender	Japanese quail	001445
MNR2	homeodomain protein	Rose comb	chicken	000884
MOCOS	molybdenum cofactor sulfurase	Xanthinuria, type II	cattle	001819
MOCS1	molybdenum cofactor synthesis 1	Arachnomelia, BTA23	cattle	001541
MPDZ	multiple PDZ domain protein	Retinal dysplasia and degeneration	chicken	001366
MRC2	mannose receptor, C type 2	Tail, crooked	cattle	001452
MSTN	myostatin	Muscular hypertrophy (double muscling)	cattle	000683
		Muscular hypertrophy (double muscling)	pig	000683
		Muscular hypertrophy (double muscling), Texel	sheep	001426
		Muscular hypertrophy (double muscling)	dog	000683
MTM1	myotubularin 1	Myotubular myopathy 1	dog	001508
MUTYH	mutY homolog (E. coli)	Cerebellar abiotrophy	horse	000175
MX1	myxovirus (influenza virus) resistance 1, interferon-inducible protein p78 (mouse)	Resistance to myxovirus	chicken	001534
MYBPC3	myosin binding protein C, cardiac	Cardiomyopathy, hypertrophic	cat	000515
MYH7	myosin, heavy chain 7, cardiac muscle, beta	Tremor, high-frequency	pig	001200
MYH9	myosin, heavy chain 9, non-muscle	May-Hegglin anomaly	dog	001608
MYO5A	myosin VA (heavy chain 12, myosin)	Lavender foal syndrome	horse	001501
Man2b1	mannosidase 2, alpha B1	Mannosidosis, alpha	guinea pig	000625
Mc1r		Coat colour, extension	rock pocket mouse	001199
		Coat colour, extension	oldfield mouse	001199
		Coat colour, extension		001199
		Coat colour, extension		001199
NAGLU	N-acetylglucosaminidase, alpha	Mucopolysaccharidosis IIIB	emu	001342
		Mucopolysaccharidosis IIIB	dog	001342
		Mucopolysaccharidosis IIIB	cattle	001342
NDRG1	N-myc downstream regulated 1	Polyneuropathy	dog	001292
NEFL	neurofilament, light polypeptide	Hypotrophic axonopathy	Japanese quail	000545
NHEJ1	nonhomologous end-joining factor 1	Collie eye anomaly	dog	000218
NHLRC1	NHL repeat containing 1	Myoclonus epilepsy of Lafora	dog	000690
NPC1	Niemann-Pick disease, type C1	Niemann-Pick disease, type C	cat	000725
NPHP4	nephronophthisis 4	Cone-rod dystrophy, Standard	dog	001455
		Wire-haired Dachshund		
OCA2		Coat colour, albinism		000202
OPA3	optic atrophy 3 (autosomal recessive, with chorea and spastic paraplegia)	Cardiomyopathy, dilated	cattle	000162

P2RY12	purinergic receptor P2Y, G-protein coupled, 12	Bleeding disorder due to P2RY12 defect	dog	001564
PAEP	progestagen-associated endometrial protein	Beta-lactoglobulin, aberrant low expression	cattle	001437
PDE6A	phosphodiesterase 6A, cGMP-specific, rod, alpha	Rod-cone dysplasia 3	dog	001314
PDE6B	phosphodiesterase 6B, cGMP-specific, rod, beta	Rod-cone dysplasia 1	dog	000882
		Rod-cone dysplasia 1a	dog	001669
PK4	pyruvate dehydrogenase kinase, isozyme 4	Cardiomyopathy, dilated	dog	000162
PDP1	pyruvate dehydrogenase phosphatase catalytic subunit 1	Pyruvate dehydrogenase deficiency	dog	001406
PFKM	phosphofructokinase, muscle	Glycogen storage disease VII	dog	000421
PISRT1	polled intersex syndrome regulated transcript 1	Polled/Horns	goat	000483
PITX3	paired-like homeodomain 3	Microphthalmia	sheep	000649
PKD1	polycystic kidney disease 1 (autosomal dominant)	Polycystic kidney disease	cat	000807
		Polycystic kidney disease	dog	000807
PKLR	pyruvate kinase, liver and RBC	Pyruvate kinase deficiency of erythrocyte	dog	000844
		Pyruvate kinase deficiency of erythrocyte	cat	000844
PLP1	proteolipid protein 1	Tremor, X-linked	rabbit	000770
		Tremor, X-linked	dog	000770
PMEL	premelanosome protein	Coat colour, merle	dog	000211
		Feather colour, dominant white	chicken	000373
		Hypotrichosis with coat-colour dilution	cattle	001544
		Coat colour, dilution	cattle	001545
PMEL17	melanocyte protein 17 precursor	Coat colour, silver	horse	001438
		Multiple ocular defects	horse	000733
PNPLA1	patatin-like phospholipase domain containing 1	Ichthyosis, Golden Retriever	dog	001588
POLLED	not yet named	Polled/Horns	cattle	000483
PPARD	peroxisome proliferator-activated receptor delta	Ear size	pig	001579
PPIB	peptidylprolyl isomerase B (cyclophilin B)	Ehlers-Danlos syndrome	horse	000327
PPP1R13L	protein phosphatase 1, regulatory subunit 13 like	Cardiomyopathy and woolly haircoat syndrome	cattle	000161
PPT1	palmitoyl-protein thioesterase 1	Neuronal ceroid lipofuscinosis, 1	dog	001504
PRCD	progressive rod-cone degeneration	Progressive rod-cone degeneration	dog	001298
PRKAG3	protein kinase, AMP-activated, gamma 3 non-catalytic subunit	Meat quality (Rendement Napole)	pig	001085
PRKDC	protein kinase, DNA-activated, catalytic polypeptide	Severe combined immunodeficiency disease, autosomal	dog	000220

PRKG2	protein kinase, cGMP-dependent, type II	Dwarfism, Angus	cattle	001485
PRLR	prolactin receptor	Feathering, Z-linked	chicken	000380
PRNP	prion protein	Spongiform encephalopathy	sheep	000944
PSMB7	proteasome (prosome, macropain) subunit, beta type, 7	Coat colour, harlequin	dog	001454
PTEN	phosphatase and tensin homolog	Colorectal hamartomatous polyposis and ganglioneuromatosis	dog	001515
PTPLA	protein tyrosine phosphatase-like (proline instead of catalytic arginine), member A	Myopathy, centronuclear	dog	001374
PYGM	phosphorylase, glycogen, muscle	Glycogen storage disease V	cattle	001139
		Glycogen storage disease V	sheep	001139
RAG1	recombination activating gene 1	Severe combined immunodeficiency disease, autosomal, T cell-negative, B cell-negative, NK cell-positive	dog	001574
RASGRP1	RAS guanyl releasing protein 1 (calcium and DAG-regulated)	Thrombopathia	dog	001003
RASGRP2	RAS guanyl releasing protein 2 (calcium and DAG-regulated)	Thrombopathia	cattle	001003
RD3	retinal degeneration 3	Rod-cone dysplasia 2	dog	001260
RHO	rhodopsin	Autosomal dominant PRA	dog	001346
RNF11	ring finger protein 11	Dwarfism, proportionate, with inflammatory lesions	cattle	001686
RPE65	retinal pigment epithelium-specific protein 65kDa	Leber congenital amaurosis (congenital stationary night blindness)	dog	001222
RPGR	retinitis pigmentosa GTPase regulator	X-linked PRA 1	dog	000831
		X-linked PRA 2	dog	001518
RPGRIP1	retinitis pigmentosa GTPase regulator interacting protein 1	Cone-rod dystrophy 4	dog	001432
RSPO2	R-spondin 2	Improper coat	dog	001498
		Furnishings (moustache and eyebrows)	dog	001531
RTBDN	retbindin	Riboflavinuria	chicken	000876
RXFP2	relaxin/insulin-like family peptide receptor 2	Polled/Horns	sheep	000483
RYR1	ryanodine receptor 1 (skeletal)	Malignant hyperthermia	dog	000621
		Malignant hyperthermia	horse	000621
		Malignant hyperthermia	pig	000621
SCN4A	sodium channel, voltage-gated, type IV, alpha subunit	Periodic paralysis II	horse	000785
SEL1L	sel-1 suppressor of lin-12-like (C. elegans)	Ataxia, cerebellar, progressive early-onset	dog	001692
SERPINH1	serpin peptidase inhibitor, clade H (heat shock protein 47), member 1, (collagen binding protein 1)	Osteogenesis imperfecta_Dachshund	dog	001483
SGCD		Cardiomyopathy, hypertrophic	golden hamster	000515

		Cardiomyopathy, dilated	golden hamster	000162
SGSH	N-sulfoglucosamine sulfohydrolase	Mucopolysaccharidosis IIIA	dog	001309
SHH	sonic hedgehog	Polydactyly	cat	000810
		Polydactyly	chicken	000810
SLC13A1	solute carrier family 13 (sodium/sulfate symporters), member 1	Chondrodysplasia, Texel	sheep	001400
		Osteochondrodysplasia	dog	001315
SLC2A9	solute carrier family 2 (facilitated glucose transporter), member 9	Urolithiasis	dog	001033
SLC35A3	solute carrier family 35 (UDP-N-acetylglucosamine (UDP-GlcNAc) transporter), member A3	Complex vertebral malformation	cattle	001340
SLC36A1	solute carrier family 36 (proton/amino acid symporter), member 1	Coat colour, champagne	horse	001263
SLC37A2	solute carrier family 37 (glycerol-3-phosphate transporter), member 2	Abortion due to haplotype MH2	cattle	001828
SLC39A4	solute carrier family 39 (zinc transporter), member 4	Acrodermatitis enteropathica	cattle	000593
SLC3A1	solute carrier family 3 (cystine, dibasic and neutral amino acid transporters, activator of cystine, dibasic and neutral amino acid transport), member 1	Cystinuria, type A	dog	000256
SLC45A2	solute carrier family 45, member 2	Feather colour, albinism, sex-linked, imperfect	chicken	000370
		Feather colour, silver	chicken	000915
		Coat colour, cream dilution	horse	001344
		Feather colour, albinism, sex-linked, imperfect	Japanese quail	000370
SLC4A1	solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego blood group)	Spherocytosis	cattle	001228
SLC4A2	solute carrier family 4, anion exchanger, member 2 (erythrocyte membrane protein band 3-like 1)	Osteopetrosis	cattle	000755
SLC4A3	solute carrier family 4, anion exchanger, member 3	Golden Retriever PRA 1	dog	001572
SLC5A3	solute carrier family 5 (sodium/myo-inositol cotransporter), member 3	Foal immunodeficiency syndrome	horse	001578
SLC6A5	solute carrier family 6 (neurotransmitter transporter, glycine), member 5	Congenital muscular dystonia 2	cattle	001451
		Hyperekplexia (Startle disease)	dog	001594
SOD1	superoxide dismutase 1, soluble	Degenerative myelopathy	dog	000263

SOX10	SRY (sex determining region Y)-box 10	Feather colour, dark brown	chicken	001569
SOX5	SRY (sex determining region Y)-box 5	Pea comb	chicken	000782
SPAST	spastin	Spinal dysmyelination	cattle	001247
SPEF2	sperm flagellar 2	Sperm, short tail	pig	001334
SPINK1	serine peptidase inhibitor, Kazal type 1	Pancreatitis, hereditary	dog	001403
SPP2	secreted phosphoprotein 2, 24kDa	Dwarfism, sex-linked	chicken	000309
SPTB	spectrin, beta, erythrocytic	Elliptocytosis	dog	001318
SPTBN2	spectrin, beta, non-erythrocytic 2	Cerebellar abiotrophy	dog	000175
SRY	sex determining region Y	Ovotesticular DSD (Disorder of Sexual Development)	horse	001230
		Ovotesticular DSD (Disorder of Sexual Development)	cattle	001230
STK38L	serine/threonine kinase 38 like	Early retinal degeneration	dog	001297
STRN	striatin, calmodulin binding protein	Arrhythmogenic right ventricular cardiomyopathy	dog	000878
STX17	syntaxin 17	Coat colour, grey/gray	horse	001356
SUOX	sulfite oxidase	Arachnomelia, BTA5	cattle	000059
T	T, brachyury homolog (mouse)	Tail, short	dog	000975
TAS1R2	taste receptor, type 1, member 2 pseudogene	Sweet taste, lack of	cat	001617
		Sweet taste, lack of	tiger	001617
		Sweet taste, lack of	cheetah	001617
TEX14	testis expressed 14	Spermatogenic arrest	pig	001673
TG	thyroglobulin	Goitre, familial	cattle	000424
		Goitre, familial	goat	000424
TGM1	transglutaminase 1 (K polypeptide epidermal type I, protein-glutamine-gamma-glutamyltransferase)	Ichthyosis	dog	000546
TMEM154	transmembrane protein 154	Resistance/susceptibility to lentivirus	sheep	001694
TNFRSF10B	tumor necrosis factor receptor superfamily, member 10b	Resistance to avian sarcoma and leukosis viruses, subgroup B	chicken	001302
TNNT2	troponin T type 2 (cardiac)	Cardiomyopathy, dilated	turkey	000162
TPO	thyroid peroxidase	Hypothyroidism	cat	000536
		Hypothyroidism	dog	000536
TPP1	tripeptidyl peptidase I	Neuronal ceroid lipofuscinosis, 2	dog	001472
TRPM1	transient receptor potential cation channel, subfamily M, member 1	Coat colour, appaloosa	horse	001341
		Night blindness, congenital stationary	horse	001486
TUBB1	tubulin, beta 1 class VI	Thrombocytopaenia	dog	001001
TWIST1	twist homolog 1 (Drosophila)	Scurs, type 2	cattle	001593
TYR	tyrosinase (oculocutaneous albinism IA)	Coat colour, albinism	cat	000202
		Coat colour, albinism	cattle	000202
		Coat colour, albinism	rabbit	000202
		Feather colour, albinism	chicken	000369
		Coat colour, albinism	water buffalo	000202
		Feather colour, recessive white	chicken	001252
		Coat colour, albinism	American mink	000202

		Coat colour, Himalayan	American mink	001497
		Coat colour, albinism	humpback whale	000202
		Coat colour, albinism	ferret	000202
TYRP1	tyrosinase-related protein 1	Coat colour, brown	cattle	001249
		Feather colour, roux	Japanese quail	001322
		Coat colour, brown	dog	001249
		Coat colour, brown	cat	001249
		Coat colour, brown	sheep	001249
		Coat colour, brown	pig	001249
Taqpep	transmembrane aminopeptidase Q	Coat colour, tabby	cat	001429
		Coat colour, king	cheetah	001721
UCP1	uncoupling protein 1	Non-shivering thermiogenesis, absence of	pig	001436
UGT1A6	UDP-glucuronosyltransferase 1A6	Deficient acetaminophen glucuronidation	cat	001586
UMPS	uridine monophosphate synthetase	Deficiency of uridine monophosphate synthase	cattle	000262
UROS	uroporphyrinogen III synthase	Porphyria, congenital erythropoietic	cat	001175
VDR	vitamin D (1,25- dihydroxyvitamin D3) receptor	Vitamin D-deficiency rickets, type II	dog	001431
VLDLR	very low density lipoprotein receptor	Restricted ovulator	chicken	000865
VPS13B	vacuolar protein sorting 13 homolog B (yeast)	Trapped Neutrophil Syndrome	dog	001428
VWF	von Willebrand factor	Von Willebrand disease III	dog	001058
		Von Willebrand disease II	dog	001339
WFDC1	WAP four-disulfide core domain 1	Multiple ocular defects	cattle	000733
WNK4	WNK lysine deficient protein kinase 4	Hypokalaemic periodic paralysis	cat	001759
WWP1	WW domain containing E3 ubiquitin protein ligase 1	Muscular dystrophy	chicken	000679
ZEB2	zinc finger E-box binding homeobox 2	Polled and multisystemic syndrome	cattle	001736
edar	ectodysplasin-A receptor	Reduced scale-3	Japanese medaka	001695
pmelb	premelanosome protein b	Coat colour, silver	zebrafish	001438
slc24a5	solute carrier family 24, member 5	Coat colour, golden	zebrafish	001433
slc45a2	membrane-associated transporter protein B	Coat colour, albinism, oculocutaneous type IV	Japanese medaka	001821
sptb	spectrin, beta, erythrocytic	Spherocytosis	zebrafish	001228
tRNA-Tyr	tRNA	Sensory ataxic neuropathy	dog	001467
tyr	tyrosinase	Coat colour, albinism	Japanese medaka	000202
urod	uroporphyrinogen decarboxylase	Porphyria cutanea tarda	sheep	001176