

Table S1. An alphabetical list of Mendelian traits/disorders for which a causal mutation has been reported, 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.

Trait/disorder	OMIA number	Species	Mutated gene symbol	Mutated gene name
Abortion and stillbirth due to mutation in MIMT1	001565	cattle	MIMT1	MER1 repeat containing imprinted transcript 1 (non-protein coding)
Abortion due to haplotype HH1	000001	cattle	APAF1	apoptotic peptidase activating factor 1
Abortion due to haplotype JH1	001697	cattle	CWC15	CWC15 spliceosome-associated protein homolog (<i>S. cerevisiae</i>)
Abortion due to haplotype MH2	001828	cattle	SLC37A2	solute carrier family 37 (glycerol-3-phosphate transporter), member 2
Achromatopsia (cone degeneration, hemeralopia), AMAL	001365	dog	CNGB3	cyclic nucleotide gated channel beta 3
Achromatopsia (cone degeneration, hemeralopia), GSPT	001676	dog	CNGB3	cyclic nucleotide gated channel beta 3
Achromatopsia-2	001481	sheep	CNGA3	cyclic nucleotide gated channel alpha 3
Acrodermatitis enteropathica	000593	cattle	SLC39A4	solute carrier family 39 (zinc transporter), member 4
Adrenal hyperplasia, congenital	000017	rabbit	CYP11A1	cytochrome P450, family 11, subfamily A, polypeptide 1
Adrenal hyperplasia, congenital, due to 11-beta hydroxylase deficiency	001661	cat	CYP11B1	cytochrome P450, family 11, subfamily B, polypeptide 1
Alopecia, colour mutant	000031	dog	MLPH	melanophilin
Analphalipoproteinaemia	000042	chicken	ABCA1	ATP-binding cassette, sub-family A (ABC1), member 1
Androgen insensitivity syndrome (AIS)	000991	horse	AR	androgen receptor
Anhidrotic ectodermal dysplasia	000543	cattle, dog	EDA	ectodysplasin A
Arachnomelia, BTA23	001541	cattle	MOCS1	molybdenum cofactor synthesis 1
Arachnomelia, BTA5	000059	cattle	SUOX	sulfite oxidase
Arrhythmogenic right ventricular cardiomyopathy	000878	dog	STRN	striatin, calmodulin binding protein
Ataxia, cerebellar	000078	dog	GRM1	glutamate receptor, metabotropic 1
Ataxia, cerebellar, progressive early-onset	001692	dog	SEL1L	sel-1 suppressor of lin-12-like (<i>C. elegans</i>)

Atrichia with papular lesions	001348	Rhesus monkey	HR	hairless homolog (mouse)
Autosomal dominant PRA	001346	dog	RHO	rhodopsin
Axonopathy	001106	cattle	MFN2	mitofusin 2
Barring	000102	chicken	CDKN2B	cyclin-dependent kinase inhibitor 2B (p15, inhibits CDK4)
Beta-lactoglobulin, aberrant low expression	001437	cattle	PAEP	progesterone-associated endometrial protein
Bleeding disorder due to P2RY12 defect	001564	dog	P2RY12	purinergic receptor P2Y, G-protein coupled, 12
Blood group system AB	000119	cat	CMAH	cytidine monophospho-N-acetylneuraminic acid hydroxylase
Blood group system ABO	001089	pig, Japanese macaque, chimpanzee, Rhesus monkey, crab-eating macaque, agile gibbon, common gibbon, siamang	GGTA1	glycoprotein, alpha-galactosyltransferase 1
Brachycephaly	001551	dog	BMP3	bone morphogenetic protein 3
Brachyspina	000151	cattle	FANCI	Fanconi anemia, complementation group I
C3 deficiency	000155	dog	C3	complement component 3

Cardiomyopathy and woolly haircoat syndrome	000161	cattle	PPP1R13L	protein phosphatase 1, regulatory subunit 13 like
Cardiomyopathy, dilated	000162	cattle, dog, turkey, golden hamster	OPA3	optic atrophy 3 (autosomal recessive, with chorea and spastic paraplegia)
Cardiomyopathy, hypertrophic	000515	cat, golden hamster	MYBPC3	myosin binding protein C, cardiac
Casein, alpha-S1, reduced concentration	000852	goat	CSN1S1	casein alpha s1
Casein, alpha-S2, absence	001623	goat	CSN1S2	casein alpha s2
Casein, beta, absence of	001424	goat	CSN2	casein beta
Cataract, early onset	001758	dog	HSF4	heat shock transcription factor 4
Cerebellar abiotrophy	000175	dog, horse	SPTBN2	spectrin, beta, non-erythrocytic 2
Chediak-Higashi syndrome	000185	cattle, American mink	LYST	lysosomal trafficking regulator
Chondrodysplasia	000187	dog, cattle	FGF4	fibroblast growth factor 4
Chondrodysplasia, Spider lamb	001703	sheep	FGFR3	fibroblast growth factor receptor 3
Chondrodysplasia, Texel	001400	sheep	SLC13A1	solute carrier family 13 (sodium/sulfate symporters), member 1
Ciliary dyskinesia, primary	001540	dog	CCDC39	coiled-coil domain containing 39
Citrullinaemia	000194	cattle	ASS1	argininosuccinate synthase 1
Coat colour, Himalayan	001497	American mink	TYR	tyrosinase (oculocutaneous albinism IA)

Coat colour, agouti

000201 horse, ASIP agouti signaling protein
cattle, red
fox,
sheep,
cat,
rabbit,
dog,
North
American
deer
mouse,
leopard,
Asiatic
golden cat

Coat colour, albinism

000202 cat, cattle, TYR tyrosinase (oculocutaneous albinism IA)
rabbit,
water
buffalo,
Japanese
medaka, ,
American
mink,
humpback
whale,
ferret

Coat colour, albinism, oculocutaneous type IV

001821 Japanese SLC45A2 solute carrier family 45, member 2
medaka

Coat colour, appaloosa

001341 horse TRPM1 transient receptor potential cation channel,
subfamily M, member 1

Coat colour, brown	001249	cattle, dog, cat, sheep, pig	TYRP1	tyrosinase-related protein 1
Coat colour, champagne	001263	horse	SLC36A1	solute carrier family 36 (proton/amino acid symporter), member 1
Coat colour, colour-sided	001576	cattle	KIT	v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog
Coat colour, cream dilution	001344	horse	SLC45A2	solute carrier family 45, member 2
Coat colour, dilute	000206	cat	MLPH	melanophilin
Coat colour, dilution	001545	cattle	PMEL	premelanosome protein
Coat colour, dominant black	001416	dog, gray wolf, coyote	CBD103	beta-defensin 103
Coat colour, dominant white	000209	pig, cattle, horse	KIT	v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog

Coat colour, extension

001199 cattle, MC1R melanocortin 1 receptor (alpha melanocyte
dog, stimulating hormone receptor)
horse, red
fox, pig,
sheep,
jaguar,
jaguarundi
,
American
black
bear,
woolly
mammoth
, cat,
rabbit,
Arctic fox,
rock
pocket
mouse,
oldfield
mouse,
gray
squirrel, ,
little
striped
whiptail,
water

Coat colour, golden

001433 zebrafish slc24a5 solute carrier family 24, member 5

Coat colour, grey/gray

001356 horse STX17 syntaxin 17

Coat colour, grizzle

001495 dog MC1R melanocortin 1 receptor (alpha melanocyte
stimulating hormone receptor)

Coat colour, harlequin	001454	dog	PSMB7	proteasome (prosome, macropain) subunit, beta type, 7
Coat colour, king	001721	cheetah	Taqpep	transmembrane aminopeptidase Q
Coat colour, macchiato	001688	horse	MITF	microphthalmia-associated transcription factor
Coat colour, melanistic mask	001590	dog	MC1R	melanocortin 1 receptor (alpha melanocyte stimulating hormone receptor)
Coat colour, merle	000211	dog	PMEL	premelanosome protein
Coat colour, patch	001743	pig	KIT	v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog
Coat colour, roan	001216	cattle, pig	KITLG	KIT ligand
Coat colour, silver	001438	horse, zebrafish	PMEL17	melanocyte protein 17 precursor
Coat colour, tabby	001429	cat	Taqpep	transmembrane aminopeptidase Q
Coat colour, white belt, due to KIT	001745	pig	KIT	v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog
Coat colour, white spotting	000214	dog, cattle, horse	MITF	microphthalmia-associated transcription factor
Coat colour, white spotting, due to KIT	001737	dog	KIT	v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog
Collie eye anomaly	000218	dog	NHEJ1	nonhomologous end-joining factor 1
Colorectal hamartomatous polyposis and ganglioneuromatosis	001515	dog	PTEN	phosphatase and tensin homolog
Complex vertebral malformation	001340	cattle	SLC35A3	solute carrier family 35 (UDP-N-acetylglucosamine (UDP-GlcNAc) transporter), member A3
Cone-rod dystrophy 3	001520	dog	ADAM9	ADAM metallopeptidase domain 9
Cone-rod dystrophy 4	001432	dog	RPGRIP1	retinitis pigmentosa GTPase regulator interacting protein 1
Cone-rod dystrophy, Standard Wire-haired Dachshund	001455	dog	NPHP4	nephronophthisis 4

Congenital keratoconjunctivitis sicca and ichthyosiform dermatosis	001683	dog	FAM83H	family with sequence similarity 83, member H
Congenital muscular dystonia 1	001450	cattle	ATP2A1	ATPase, Ca ⁺⁺ transporting, cardiac muscle, fast twitch 1
Congenital muscular dystonia 2	001451	cattle	SLC6A5	solute carrier family 6 (neurotransmitter transporter, glycine), member 5
Curly coat	000245	dog	KRT71	keratin 71
Curly coat, Devon rex	001581	cat	KRT71	keratin 71
Cystic fibrosis	001794	pig, ferret	CFTR	cystic fibrosis transmembrane conductance regulator (ATP-binding cassette sub-family C, member 7)
Cystinuria, type A	000256	dog	SLC3A1	solute carrier family 3 (cystine, dibasic and neutral amino acid transporters, activator of cystine, dibasic and neutral amino acid transport), member 1
Deafness	000259	pig	MITF	microphthalmia-associated transcription factor
Deficiency of uridine monophosphate synthase	000262	cattle	UMPS	uridine monophosphate synthetase
Deficient acetaminophen glucuronidation	001586	cat	UGT1A6	UDP-glucuronosyltransferase 1A6
Degenerative myelopathy	000263	dog	SOD1	superoxide dismutase 1, soluble
Dermoid sinus	000272	dog	FGF4	fibroblast growth factor 4
Dihydropyrimidinase deficiency	001776	cat	DPYS	dihydropyrimidinase
Dominant white with bilateral deafness	001680	cattle	MITF	microphthalmia-associated transcription factor
Dwarfism, Angus	001485	cattle	PRKG2	protein kinase, cGMP-dependent, type II
Dwarfism, Dexter	001271	cattle	ACAN	aggrecan
Dwarfism, Schmid metaphyseal chondrodysplasia	001718	pig	COL10A1	collagen, type X, alpha 1
Dwarfism, growth-hormone deficiency	001473	cattle	GH1	growth hormone 1
Dwarfism, pituitary	000307	dog	LHX3	LIM homeobox 3
Dwarfism, proportionate, with inflammatory lesions	001686	cattle	RNF11	ring finger protein 11
Dwarfism, sex-linked	000309	chicken	SPP2	secreted phosphoprotein 2, 24kDa

Ear size	001579	pig	PPARD	peroxisome proliferator-activated receptor delta
Early retinal degeneration	001297	dog	STK38L	serine/threonine kinase 38 like
Ectodermal dysplasia	000323	dog	FOXI3	forkhead box I3
Ehlers-Danlos syndrome	000327	horse	PPIB	peptidylprolyl isomerase B (cyclophilin B)
Ehlers-Danlos syndrome, Holstein variant	001716	cattle	EPYC	epiphycan
Ehlers-Danlos syndrome, type VII (Dermatosparaxis)	000328	cattle, sheep	ADAMTS2	ADAM metalloproteinase with thrombospondin type 1 motif, 2
Elliptocytosis	001318	dog	SPTB	spectrin, beta, erythrocytic
Epidermolysis bullosa	000340	cattle	KRT5	keratin 5
Epidermolysis bullosa, dystrophic	000341	dog, cattle	COL7A1	collagen, type VII, alpha 1
Epidermolysis bullosa, junctionalis, LAMA3	001677	dog, horse	LAMA3	laminin, alpha 3
Epidermolysis bullosa, junctionalis, LAMC2	001678	sheep, horse	LAMC2	laminin, gamma 2
Epilepsy	000344	chicken	LOC430486	similar to Ca ²⁺ regulator SV2A
Epilepsy, benign familial juvenile	001596	dog	LGI2	leucine-rich repeat LGI family, member 2
Episodic falling	001592	dog	BCAN	brevican
Exercise-induced collapse	001466	dog	DNM1	dynamamin 1
Factor VII deficiency	000361	dog	F7	coagulation factor VII (serum prothrombin conversion accelerator)
Factor XI deficiency	000363	cattle, dog	F11	coagulation factor XI
Feather colour, albinism	000369	chicken	TYR	tyrosinase (oculocutaneous albinism IA)
Feather colour, albinism, sex-linked, imperfect	000370	chicken, Japanese quail	SLC45A2	solute carrier family 45, member 2

Feather colour, bronze	001494	turkey	MC1R	melanocortin 1 receptor (alpha melanocyte stimulating hormone receptor)
Feather colour, dark brown	001569	chicken	SOX10	SRY (sex determining region Y)-box 10
Feather colour, dominant white	000373	chicken	PMEL	premelanosome protein
Feather colour, extended black	000374	chicken, red- footed booby, Bananaqui t, helmeted guineafow l, gyrfalcon, Lesser snow goose, , , brown booby	MC1R	melanocortin 1 receptor (alpha melanocyte stimulating hormone receptor)
Feather colour, extended brown	001435	Japanese quail	MC1R	melanocortin 1 receptor (alpha melanocyte stimulating hormone receptor)
Feather colour, lavender	001445	chicken, Japanese quail	MLPH	melanophilin
Feather colour, panda/dotted white	000375	Japanese quail	EDNRB2	endothelin receptor B subtype 2
Feather colour, recessive white	001252	chicken	TYR	tyrosinase (oculocutaneous albinism IA)

Feather colour, roux	001322	Japanese quail	TYRP1	tyrosinase-related protein 1
Feather colour, silver	000915	chicken, Japanese quail	SLC45A2	solute carrier family 45, member 2
Feather colour, yellow	001602	Japanese quail	ASIP	agouti signaling protein
Feathering, Z-linked	000380	chicken	PRLR	prolactin receptor
Fecundity, Belclare, FecX(B)	001719	sheep	BMP15	bone morphogenetic protein 15
Fecundity, Booroola, FecB(B)	000383	sheep	BMPR1B	bone morphogenetic protein receptor, type IB
Fecundity, Embrapa, FecG(E)	001559	sheep	GDF9	growth differentiation factor 9
Fecundity, Galway, FecX(G)	000384	sheep	BMP15	bone morphogenetic protein 15
Fecundity, Hanna, FecX(H)	001350	sheep	BMP15	bone morphogenetic protein 15
Fecundity, High fertility, FecG(H)	001801	sheep	GDF9	growth differentiation factor 9
Fecundity, Inverdale, FecX(I)	000386	sheep	BMP15	bone morphogenetic protein 15
Fecundity, Lacaune, FecX(L)	001357	sheep	BMP15	bone morphogenetic protein 15
Fecundity, Norwegian White Sheep	001763	sheep	GDF9	growth differentiation factor 9
Fecundity, Rasa Aragonesa, FecX(R)	001800	sheep	BMP15	bone morphogenetic protein 15
Fecundity, Thoka, FecG(T)	000385	sheep	GDF9	growth differentiation factor 9
Fecundity, small tail Han	001423	sheep	BMPR1B	bone morphogenetic protein receptor, type IB
Feet, white	001580	cat	KIT	v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog
Foal immunodeficiency syndrome	001578	horse	SLC5A3	solute carrier family 5 (sodium/myo-inositol cotransporter), member 3
Frizzle	000394	chicken	KRT75	keratin 75
Fucosidosis, alpha	000396	dog	FUCA1	fucosidase, alpha-L- 1, tissue
Furnishings (moustache and eyebrows)	001531	dog	RSPO2	R-spondin 2
Gaitedness	001715	horse	LOC100147177	doublesex- and mab-3-related transcription factor 3-like
Gallbladder mucoceles	001524	dog	ATCB4	ATP-binding cassette, sub-family B (MDR/TAP), member 4

Gangliosidosis, GM1	000402	cat, dog, sheep	GLB1	galactosidase, beta 1
Gangliosidosis, GM2, GM2A deficiency	001427	cat	GM2A	GM2 ganglioside activator
Gangliosidosis, GM2, type I	001461	American flamingo, sheep	HEXA	Hexosaminidase A (alpha polypeptide)
Gangliosidosis, GM2, type II	001462	cat, dog	HEXB	hexosaminidase B (beta polypeptide)
Gastrointestinal stromal tumor	001516	dog	KIT	v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog
Gaucher disease, type I	000405	sheep	GBA	glucosidase, beta, acid
Generalized PRA	001521	dog	CCDC66	coiled-coil domain containing 66
Glaucoma	000411	dog	ADAMTS10	ADAM metalloproteinase with thrombospondin type 1 motif, 10
Glycogen storage disease II	000419	cattle	GAA	glucosidase, alpha; acid
Glycogen storage disease IIIa	001577	dog	AGL	amylo-alpha-1, 6-glucosidase, 4-alpha- glucanotransferase
Glycogen storage disease IV	000420	cat, horse	GBE1	glucan (1,4-alpha-), branching enzyme 1
Glycogen storage disease Ia	000418	dog	G6PC	glucose-6-phosphatase, catalytic subunit
Glycogen storage disease V	001139	cattle, sheep	PYGM	phosphorylase, glycogen, muscle
Glycogen storage disease VII	000421	dog	PFKM	phosphofructokinase, muscle
Goitre, familial	000424	cattle, goat	TG	thyroglobulin
Golden Retriever PRA 1	001572	dog	SLC4A3	solute carrier family 4, anion exchanger, member 3
Haemophilia A	000437	dog, sheep, cattle	F8	coagulation factor VIII, procoagulant component
Haemophilia B	000438	cat, dog	F9	coagulation factor IX
Hair length	000439	cat, dog	FGF5	fibroblast growth factor 5

Henny feathering	000452	chicken	CYP19A1	cytochrome P450, family 19, subfamily A, polypeptide 1
Hypercholesterolaemia	000499	pig	LDLR	low density lipoprotein receptor
Hyperekplexia (Startle disease)	001594	dog	SLC6A5	solute carrier family 6 (neurotransmitter transporter, glycine), member 5
Hyperkeratosis, epidermolytic	001415	dog	KRT10	keratin 10
Hyperlipidaemia	001160	rabbit	LDLR	low density lipoprotein receptor
Hyperlipoproteinaemia	001210	cat	LPL	lipoprotein lipase
Hypocatalasia	001138	dog	CAT	catalase
Hypokalaemic periodic paralysis	001759	cat	WNK4	WNK lysine deficient protein kinase 4
Hypophosphatemic rickets, autosomal recessive, 1	001542	sheep	DMP1	dentin matrix acidic phosphoprotein 1
Hypopigmentation syndrome, lethal	001765	sheep	ETBR	endothelin B receptor
Hypothyroidism	000536	cat, dog	TPO	thyroid peroxidase
Hypotrichosis	000540	cattle, sheep	HEPHL1	hephaestin-like 1
Hypotrichosis with coat-colour dilution	001544	cattle	PMEL	premelanosome protein
Hypotrichosis, with whiskers short and curled	001583	cat	KRT71	keratin 71
Hypotrophic axonopathy	000545	Japanese quail	NEFL	neurofilament, light polypeptide
Ichthyosis	000546	dog	TGM1	transglutaminase 1 (K polypeptide epidermal type I, protein-glutamine-gamma-glutamyltransferase)
Ichthyosis congenita	000547	cattle	ABCA12	ATP-binding cassette, sub-family A (ABC1), member 12
Ichthyosis, Golden Retriever	001588	dog	PNPLA1	patatin-like phospholipase domain containing 1
Improper coat	001498	dog	RSPO2	R-spondin 2
Intestinal cobalamin malabsorption due to AMN mutation	000565	dog	AMN	amniotically expressed nuclear (mouse)

Krabbe disease	000578	dog, Rhesus monkey	GALC	galactosylceramidase
L-2-hydroxyglutaricacidemia	001371	dog	L2HGDH	L-2-hydroxyglutarate dehydrogenase
Lavender foal syndrome	001501	horse	MYO5A	myosin VA (heavy chain 12, myosin)
Leber congenital amaurosis (congenital stationary night blindness)	001222	dog	RPE65	retinal pigment epithelium-specific protein 65kDa
Lens luxation	000588	dog	ADAMTS17	ADAM metalloproteinase with thrombospondin type 1 motif, 17
Lethal multi-organ developmental dysplasia	001722	cattle	KDM2B	lysine (K)-specific demethylase 2B
Leucodystrophy	001130	dog	CYTB	cytochrome b
Leukemia, chronic monocytic	001573	dog	BCR	breakpoint cluster region
Leukocyte adhesion deficiency, type I	000595	cattle, dog	ITGB2	integrin, beta 2 (complement component 3 receptor 3 and 4 subunit)
Leukocyte adhesion deficiency, type III	001525	dog	FERMT3	fermitin family member 3
Malignant hyperthermia	000621	dog, horse, pig	RYR1	ryanodine receptor 1 (skeletal)
Mannosidosis, alpha	000625	cattle, cat, guinea pig	MAN2B1	mannosidase, alpha, class 2B, member 1
Mannosidosis, beta	000626	cattle, goat	MANBA	mannosidase, beta A, lysosomal
Maple syrup urine disease	000627	cattle	BCKDHA	branched chain keto acid dehydrogenase E1, alpha polypeptide
Marfan syndrome	000628	cattle	FBN1	fibrillin 1
May-Hegglin anomaly	001608	dog	MYH9	myosin, heavy chain 9, non-muscle
Meat quality (Rendement Napole)	001085	pig	PRKAG3	protein kinase, AMP-activated, gamma 3 non-catalytic subunit
Megacolon	000629	horse	EDNRB	endothelin receptor type B
Membranoproliferative glomerulonephritis type II	000636	pig	CFH	complement factor H

Metabolizer of a cognitive enhancer	001405	dog	CYP1A2	cytochrome P450, family 1, subfamily A, polypeptide 2
Microphthalmia	000649	sheep	PITX3	paired-like homeodomain 3
Motor neuron disease, lower	000662	sheep	AGTPBP1	ATP/GTP binding protein 1
Mucopolidosis II	001248	cat	GNTPAB	N-acetylglucosamine-1-phosphate transferase, alpha and beta subunits
Mucopolysaccharidosis I	000664	dog, cat	IDUA	iduronidase, alpha-L-
Mucopolysaccharidosis IIIA	001309	dog	SGSH	N-sulfoglucosamine sulfohydrolase
Mucopolysaccharidosis IIIB	001342	emu, dog, cattle	NAGLU	N-acetylglucosaminidase, alpha
Mucopolysaccharidosis IIID	000665	goat	GNS	glucosamine (N-acetyl)-6-sulfatase
Mucopolysaccharidosis VI	000666	cat, dog	ARSB	arylsulfatase B
Mucopolysaccharidosis VII	000667	cat, dog	GUSB	glucuronidase, beta
Multidrug resistance 1	001402	dog	ABCB1	ATP-binding cassette, sub-family B (MDR/TAP), member 1
Multifocal retinopathy 1	001444	dog	BEST1	bestrophin 1
Multifocal retinopathy 2	001553	dog	BEST1	bestrophin 1
Multifocal retinopathy 3	001554	dog	BEST1	bestrophin 1
Multiple ocular defects	000733	cattle, horse	WFDC1	WAP four-disulfide core domain 1
Muscular dystrophy	000679	chicken	WWP1	WW domain containing E3 ubiquitin protein ligase 1
Muscular dystrophy, Duchenne type	001081	cat, dog	DMD	Duchenne muscular dystrophy
Muscular hypertrophy (double muscling)	000683	cattle, pig, dog	MSTN	myostatin
Muscular hypertrophy (double muscling), Callipyge	001354	sheep	DLK1	delta-like 1 homolog (Drosophila)
Muscular hypertrophy (double muscling), Texel	001426	sheep	MSTN	myostatin
Musladin-Lueke syndrome	001509	dog	ADAMTSL2	ADAMTS-like 2
Myasthenic syndrome, congenital	000685	dog, cattle	CHAT	choline O-acetyltransferase
Myoclonus	000689	cattle	GLRA1	glycine receptor, alpha 1
Myoclonus epilepsy of Lafora	000690	dog	NHLRC1	NHL repeat containing 1

Myopathy of the diaphragmatic muscles	001319	cattle	HSPA1A	heat shock 70kDa protein 1A
Myopathy, centronuclear	001374	dog	PTPLA	protein tyrosine phosphatase-like (proline instead of catalytic arginine), member A
Myotonia	000698	goat, dog, horse	CLCN1	chloride channel, voltage-sensitive 1
Myotubular myopathy 1	001508	dog	MTM1	myotubularin 1
Naked neck	000701	chicken	GDF7	growth differentiation factor 7
Nanomelia	000702	chicken	ACAN	aggrecan
Narcolepsy	000703	dog	HCRTR2	hypocretin (orexin) receptor 2
Neonatal encephalopathy with seizures	001471	dog	ATF2	activating transcription factor 2
Nephritis, X-linked	001112	dog	COL4A5	collagen, type IV, alpha 5
Nephropathy	000710	dog	COL4A4	collagen, type IV, alpha 4
Neuroaxonal dystrophy	000715	dog	MFN2	mitofusin 2
Neuronal ceroid lipofuscinosis, 1	001504	dog	PPT1	palmitoyl-protein thioesterase 1
Neuronal ceroid lipofuscinosis, 10	001505	dog, sheep	CTSD	cathepsin D
Neuronal ceroid lipofuscinosis, 2	001472	dog	TPP1	tripeptidyl peptidase I
Neuronal ceroid lipofuscinosis, 4A	001503	dog	ARSG	arylsulfatase G
Neuronal ceroid lipofuscinosis, 5	001482	sheep, dog, cattle	CLN5	ceroid-lipofuscinosis, neuronal 5
Neuronal ceroid lipofuscinosis, 6	001443	sheep, dog	CLN6	ceroid-lipofuscinosis, neuronal 6, late infantile, variant
Neuronal ceroid lipofuscinosis, 8	001506	dog	CLN8	ceroid-lipofuscinosis, neuronal 8 (epilepsy, progressive with mental retardation)
Neuronal ceroid lipofuscinosis, adult-onset, Tibetan Terrier	001552	dog	ATP13A2	ATPase type 13A2
Neutropenia, cyclic	000248	dog	AP3B1	adaptor-related protein complex 3, beta 1 subunit
Niemann-Pick disease, type C	000725	cat	NPC1	Niemann-Pick disease, type C1

Night blindness, congenital stationary	001486	horse	TRPM1	transient receptor potential cation channel, subfamily M, member 1
Non-shivering thermiogenesis, absence of	001436	pig	UCP1	uncoupling protein 1
Oculoskeletal dysplasia 1	001522	dog	COL9A3	collagen, type IX, alpha 3
Oculoskeletal dysplasia 2	001523	dog	COL9A2	collagen, type IX, alpha 2
Oligozeugodactyly	001612	chicken	LMBR1	limb region 1 homolog (mouse)
Osteochondrodysplasia	001315	dog	SLC13A1	solute carrier family 13 (sodium/sulfate symporters), member 1
Osteogenesis imperfecta	000754	dog	COL1A2	collagen, type I, alpha 2
Osteogenesis imperfecta_Dachshund	001483	dog	SERPINH1	serpin peptidase inhibitor, clade H (heat shock protein 47), member 1, (collagen binding protein 1)
Osteopetrosis	000755	cattle, Japanese quail	SLC4A2	solute carrier family 4, anion exchanger, member 2 (erythrocyte membrane protein band 3-like 1)
Ovotesticular DSD (Disorder of Sexual Development)	001230	horse, cattle	SRY	sex determining region Y
Pancreatitis, hereditary	001403	dog	SPINK1	serine peptidase inhibitor, Kazal type 1
Pea comb	000782	chicken	SOX5	SRY (sex determining region Y)-box 5
Periodic Fever Syndrome	001561	dog	HAS2	hyaluronan synthase 2
Periodic paralysis II	000785	horse	SCN4A	sodium channel, voltage-gated, type IV, alpha subunit
Persistent Mullerian duct syndrome	000791	dog	AMHR2	anti-Mullerian hormone receptor, type II
Polled and multisystemic syndrome	001736	cattle	ZEB2	zinc finger E-box binding homeobox 2
Polled/Horns	000483	cattle, goat, sheep	POLLED	not yet named
Polycystic kidney disease	000807	cat, dog	PKD1	polycystic kidney disease 1 (autosomal dominant)
Polycythemia	000809	dog	JAK2	Janus kinase 2
Polydactyly	000810	cat, chicken	SHH	sonic hedgehog

Polyneuropathy	001292	dog	NDRG1	N-myc downstream regulated 1
Polysaccharide storage myopathy/Exertional rhabdomyolysis	001158	horse	GYS1	glycogen synthase 1 (muscle)
Porphyria cutanea tarda	001176	sheep	urod	uroporphyrinogen decarboxylase
Porphyria, acute intermittent	001493	cat	HMBS	hydroxymethylbilane synthase
Porphyria, congenital erythropoietic	001175	cat	UROS	uroporphyrinogen III synthase
Prekallikrein deficiency	000819	dog	KLKB1	kallikrein B, plasma (Fletcher factor) 1
Primary hyperoxaluria type I (Oxalosis I)	001672	dog	AGXT	alanine-glyoxylate aminotransferase
Primary hyperoxaluria type II (Oxalosis II)	000821	cat	GRHPR	glyoxylate reductase/hydroxypyruvate reductase
Progressive rod-cone degeneration	001298	dog	PRCD	progressive rod-cone degeneration
Protoporphyrin	000836	cattle	FECH	ferrochelatase
Pseudomyotonia, congenital	001464	cattle	ATP2A1	ATPase, Ca ⁺⁺ transporting, cardiac muscle, fast twitch 1
Pyruvate dehydrogenase deficiency	001406	dog	PDP1	pyruvate dehydrogenase phosphatase catalytic subunit 1
Pyruvate kinase deficiency of erythrocyte	000844	dog, cat	PKLR	pyruvate kinase, liver and RBC
Reduced scale-3	001695	Japanese medaka	edar	ectodysplasin-A receptor
Renal cystadenocarcinoma and nodular dermatofibrosis	001335	dog	FLCN	folliculin
Renal dysplasia	001135	cattle	CLDN16	claudin 16
Resistance to avian sarcoma and leukosis viruses, subgroup A	001299	chicken	CD320	CD320 molecule
Resistance to avian sarcoma and leukosis viruses, subgroup B	001302	chicken	TNFRSF10B	tumor necrosis factor receptor superfamily, member 10b
Resistance to avian sarcoma and leukosis viruses, subgroup C	001622	chicken	BTN1A1	butyrophilin, subfamily 1, member A1
Resistance to grass carp reovirus	001725	grass carp	CiRIG-I	ctenopharyngodon idella retinoic acid-inducible gene I
Resistance to myxovirus	001534	chicken	MX1	myxovirus (influenza virus) resistance 1, interferon-inducible protein p78 (mouse)

Resistance to oedema disease (F18 receptor)	000862	pig	FUT1	fucosyltransferase 1 (galactoside 2-alpha-L-fucosyltransferase, H blood group)
Resistance/susceptibility to lentivirus	001694	sheep	TMEM154	transmembrane protein 154
Restricted ovulator	000865	chicken	VLDLR	very low density lipoprotein receptor
Retinal degeneration I	001245	chicken	GC1	photoreceptor guanylate cyclase 1
Retinal degeneration II	001244	cat	CEP290	centrosomal protein 290kDa
Retinal degeneration, fluoroquinolone-induced	001707	cat	ABCG2	ATP-binding cassette, sub-family G (WHITE), member 2
Retinal dysplasia and degeneration	001366	chicken	MPDZ	multiple PDZ domain protein
Retinopathy globe enlarged	001368	chicken	GNB3	guanine nucleotide binding protein (G protein), beta polypeptide 3
Rex coat	001566	rabbit	LIPH	lipase, member H
Riboflavinuria	000876	chicken	RTBDN	retbindin
Rod-cone dysplasia	000881	cat	CRX	cone-rod homeobox
Rod-cone dysplasia 1	000882	dog	PDE6B	phosphodiesterase 6B, cGMP-specific, rod, beta
Rod-cone dysplasia 1a	001669	dog	PDE6B	phosphodiesterase 6B, cGMP-specific, rod, beta
Rod-cone dysplasia 2	001260	dog	RD3	retinal degeneration 3
Rod-cone dysplasia 3	001314	dog	PDE6A	phosphodiesterase 6A, cGMP-specific, rod, alpha
Rose comb	000884	chicken	MNR2	homeodomain protein
Scaleless	000889	chicken	FGF20	fibroblast growth factor 20
Scurs, type 2	001593	cattle	TWIST1	twist homolog 1 (Drosophila)
Sensory ataxic neuropathy	001467	dog	tRNA-Tyr	tRNA
Severe combined immunodeficiency disease, X-linked	000899	dog	IL2RG	interleukin 2 receptor, gamma
Severe combined immunodeficiency disease, autosomal	000220	horse, dog	DNAPK	DNA-dependent protein kinase catalytic subunit
Severe combined immunodeficiency disease, autosomal, T cell-negative, B cell-negative, NK cell-positive	001574	dog	RAG1	recombination activating gene 1

Sex determination	001728	torafugu	Amhr2	anti-Mullerian hormone type 2 receptor
Silky/Silkie pigmentation (Fibromelanosis)	001671	chicken	EDN3	endothelin 3
Skin colour, yellow	001449	chicken	BCO2	beta-carotene oxygenase 2
Sperm, short tail	001334	pig	SPEF2	sperm flagellar 2
Spermatogenic arrest	001673	pig	TEX14	testis expressed 14
Spherocytosis	001228	cattle, zebrafish	SLC4A1	solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego blood group)
Spinal dysmyelination	001247	cattle	SPAST	spastin
Spinal muscular atrophy	000939	cattle, cat	KDSR	3-ketodihydrosphingosine reductase
Spongiform encephalopathy	000944	sheep	PRNP	prion protein
Stress syndrome	001685	pig	DMD	dystrophin
Sweet taste, lack of	001617	cat, tiger, cheetah	TAS1R2	taste receptor, type 1, member 2 pseudogene
Syndactyly (mule foot)	000963	cattle	LRP4	low density lipoprotein receptor-related protein 4
Tail, crooked	001452	cattle	MRC2	mannose receptor, C type 2
Tail, short	000975	dog	T	T, brachyury homolog (mouse)
Talpid-3	000979	chicken	KIAA0586	KIAA0586
Thrombasthenia	001000	dog, horse	ITGA2B	integrin, alpha 2b (platelet glycoprotein IIb of IIb/IIIa complex, antigen CD41)
Thrombocytopaenia	001001	dog	TUBB1	tubulin, beta 1 class VI
Thrombopathia	001003	dog, cattle	RASGRP1	RAS guanyl releasing protein 1 (calcium and DAG-regulated)
Trapped Neutrophil Syndrome	001428	dog	VPS13B	vacuolar protein sorting 13 homolog B (yeast)
Tremor, X-linked	000770	rabbit, dog	PLP1	proteolipid protein 1

Tremor, high-frequency	001200	pig	MYH7	myosin, heavy chain 7, cardiac muscle, beta
Trimethylaminuria	001360	cattle, chicken	FMO3	flavin containing monooxygenase 3
Urolithiasis	001033	dog	SLC2A9	solute carrier family 2 (facilitated glucose transporter), member 9
Vitamin D-deficiency rickets, type I	000837	pig, cat	CYP27B1	25-hydroxyvitamin D3 1alpha-hydroxylase
Vitamin D-deficiency rickets, type II	001431	dog	VDR	vitamin D (1,25- dihydroxyvitamin D3) receptor
Von Willebrand disease II	001339	dog	VWF	von Willebrand factor
Von Willebrand disease III	001058	dog	VWF	von Willebrand factor
Waardenburg syndrome, type IIA	001401	golden hamster	MITF	microphthalmia-associated transcription factor
Wilson disease	001071	dog	COMMD1	copper metabolism (Murr1) domain containing 1
X-linked PRA 1	000831	dog	RPGR	retinitis pigmentosa GTPase regulator
X-linked PRA 2	001518	dog	RPGR	retinitis pigmentosa GTPase regulator
XY Ovotesticular DSD (Disorder of Sexual Development)	001709	Japanese medaka	DMRT1	doublesex and mab-3 related transcription factor 1
Xanthinuria, type II	001819	cattle	MOCOS	molybdenum cofactor sulfurase
Yellow fat	001079	sheep, cattle	BCO2	beta-carotene oxygenase 2