Table 2 Inherited diseases of domestic cats for which a commercial DNA test is available

Disease/trait (Alleles) OMIA entry link	MOI*	Phenotype	Gene	Gene name	Mutation
AB blood type (*A*$^+$, *b*)[71] 000119-9685	AR	Determines type B	*CMAH*	*Cytidine monophospho-N-acetylneuraminic acid hydroxylase*	c.1del-53_70 c.139G>A
Craniofacial defect	AR	Craniofacial defect	unpublished	unpublished	unpublished
Gangliosidosis 1[72] 000402-9685	AR	Lipid storage disorder (GM1)	*GLB1*	*Galactosidase, beta 1*	c.1457G>C
Gangliosidosis 2[69,73] 01462-0985	AR	Lipid storage disorder (GM2)	*HEXB*	*Hexominidase B*	c.1356del-1_8 c.1356_1362delGTTCTCA c.39delC
Glycogen storage disease IV[74] 000420-9685	AR	Glycogen storage disorder	*GBE1*	*Glycogen branching enzyme 1*	IVS11+1552_IVS12-1339 del6.2kb ins334 bp
Hypertrophic cardiomyopathy[75,76] 000515-9685	AD	Cardiac disease (HCM)	*MYBPC*	*Myosin binding protein C*	c.93G>C c.2460C>T
Hypokalemia[77] 001759-9685	AR	Potassium deficiency (HK)	*WNK4*	*WNK lysine deficient protein kinase 4*	c.2899C>T
Progressive retinal atrophy[78] 001244-9685	AR	Late-onset blindness (rdAC)	*CEP290*	*Centrosomal protein 290kDa*	IVS50 + 9T>G
Progressive retinal atrophy[79] 000881-9685	AD	Early-onset blindness (rdy)	*CRX*	*Cone-rod homeobox*	c.546delC
Polycystic kidney disease[80] 000807-9685	AD	Kidney cysts (PKD)	*PKD1*	*Polycystin 1*	c.10063C>A
Pyruvate kinase deficiency[81] 000844-9685	AR	Hemopathy (PK deficiency)	*PKLR*	*Pyruvate kinase, liver, RBC*	c.693+304G>A
Spinal muscular atrophy[82] 000939-9685	AR	Muscular atrophy (SMA)	*LIX1-LNPEP*	*Limb expression 1 homolog – leucyl/cystinyl aminopeptidase*	Partial gene deletions

*Mode of inheritance of the non-wild type variant. Not all transcripts for a given gene may have been discovered or well documented in the cat; mutations presented as interpreted from original publication. '+' implies the wild type allele when known. In reference to the mutant allele, AD = autosomal dominant, AR = autosomal recessive. OMIA: Online Mendelian Inheritance in Animals (omia.angis.org.au) entries provide links to citations and clinical descriptions of the phenotypes and the diseases. Listed citations are for the causative variant discovery