Table 5 Uncommon mutations for inherited diseases of domestic cats*

Disease	OMIA entry link	Gene	Mutation†
11β-hydroxylase deficiency (congenital adrenal hypoplasia)[95]	001661-9685	*CYP11B1*	Exon 7 G>A
Dihydropyrimidinase deficiency[96]	001776-9685	*DPYS*	c.1303G>A
Factor XII deficiency[97]	000364-9685	*FXII*	c.1321delC
Fibrodysplasia ossificans progressiva	000388-9685	unpublished	unpublished
Gangliosidosis 1[98]	000402-9685	*GLB1*	c.1448G>C
Gangliosidosis 2[99,100]	001462-9685	*HEXB*	c.1467_1491inv c.667C>T
Gangliosidosis 2[74]	001427-9685	*GM2A*	c.390_393GGTC
Hemophilia B[101]	000438-9685	*F9*	c.247G>A c.1014C>T
Hyperoxaluria[102]	000821-9685	*GRHPR*	G>A I4 acceptor site
Hypothyroidism	000536-9685	unpublished	unpublished
Lipoprotein lipase deficiency[103]	001210-9685	*LPL*	c.1234G>A
Mucolipidosis II[104]	001248-9685	*GNPTAB*	c.2655C>T
Mannosidosis, alpha[105]	000625-9685	*LAMAN*	c.1748_1751delCCAG
Mucopolysaccharidosis I[106]	000664-9685	*IDUA*	c.1107_1109delCGA c.1108_1110GAC
Mucopolysaccharidosis VI[94]	000666-9685	*ARSB*	c.1427T>C
Mucopolysaccharidosis VI[93,107]	000666-9685	*ARSB*	c.1558G>A
Mucopolysaccharidosis VII[108]	000667-9685	*GUSB*	c.1052A>G
Muscular dystrophy[68]	001081-9685	*DMD*	900bp del M promoter – exon 1
Niemann–Pick C[109]	000725-9685	*NPC*	c.2864G>C
Polydactyly[17]	000810-9685	*SHH*	c.479A>G c.257G>C c.481A>T
Porphyria (congenital erythropoietic)[110]‡	001175-9685	*UROS*	c.140C>T c.331G>A
Porphyria (acute intermittent)[111]‡	001493-9685	*HMBS*	c.842_844delGAG c.189dupT c.250G>A c.445C>T
Vitamin D resistant rickets[112,113]	000837-9685	*CYP27B1*	c.223G>A c.731delG c.637G>T

*The presented conditions are not prevalent in breeds or populations but may have been established into research colonies. †Not all transcripts for a given gene may have been discovered or well documented in the cat; mutations presented as interpreted from original publication. ‡A variety of mutations have been identified, yet unpublished, for porphyrias in domestic cats. Contact PennGen at the University of Pennsylvania for additional information. 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