**Table 1** The phenotypic traits of the domestic cat.

| Locus (alleles) | MOI‡ | Phenotype | Gene | Gene Name |
|---|---|---|---|---|
| *Agouti ($A^+$, a)*[62] | AR | Banded fur to solid | *ASIP* | *Agouti signaling protein* |
| *Brown ($B^+$, b, $b^l$)*[63,64] | AR | Brown, light brown color variants | *TYRP1* | *Tyrosinase related protein* |
| *Color ($C^+$, $C^b$, $C^s$, c)*[64-66] | AR | Burmese, Siamese color pattern, full albino | *TYR* | *Tyrosinase* |
| *Dilution ($D^+$, d)*[67] | AR | Black to grey / blue, Orange to cream | *MLPH* | *Melanophilin* |
| *Extension ($E^+$, e) – Amber*[68] | AR | Brown/red color variant | *MC1R* | *Melanocortin receptor 1* |
| *Fold (Fd, $fd^+$)* | AD | Ventral ear fold | *unknown* | *unpublished* |

| | | | | |
|---|---|---|---|---|
| *Gloves ($G^+$, g)* | AR | White feet | *KIT* | *unpublished* |
| *Hairless ($Hr^+$, hr))*[47] | AR | Atrichia | *KRT71* | *Keratin 71* |
| *Inhibitor (I, i)* | AD | Loss of pheomelanin | *unknown* | *unknown* |
| *Japanese Bobtail (J, $j^+$)* | AD | *Kinked tail* | *unknown* | *unknown* |
| *Kurl (K, $k^+$)* | AD | *Rostral curled pinnea* | *unknown* | *unknown* |
| *Long fur ($L^+$, l)*[23,69] | AR | *Long fur* | *FGF5* | *Fibroblast growth factor 3* |
| *Manx (M, $m^+$)* | AD | Absence/short tail | *TBOX* | *T - box* |
| *Orange ($O^+$, o)* | X linked | Change in pigment hue | *unknown* | *unknown* |
| *Polydactyla (Pd, $pd^+$)*[77] | AD | Extra toes | *SHH* | *Sonic hedgehog* |
| *Rexing ($R^+$, r)*[70] | AR | Curly hair coat – Cornish Rex | *LPAR6* | *Lysophosphatidic acid receptor 6* |
| *Rexing ($Re^+$, re)*[47] | AR | Curly hair coat – Devon Rex | *KRT71* | *Keratin 71* |

| | | | | |
|---|---|---|---|---|
| *Rexing ($R^S$, $r^{s+}$)[24]* | AD | Curly hair coat – Selkirk Rex | *KRT71* | *Keratin 71* |
| *Spotting (S, $s^+$)* | Co-D | Bicolor / van white | *unknown* | *unknown* |
| *Tabby[48] ($T^+$, $t^b$)* | AR | Blotched/classic pattern | *TAQPEP* | *Transmembrane aminopeptidase Q* |
| *Ticked ($T^a$, t)* | AD | No Tabby pattern | *unknown* | *unknown* |
| *White (W, $w^+$)* | AD | Loss of pigmentation | *unknown* | *unknown* |
| *Wide-band* | unknown | Length of pheomelanin band in hair | *unknown* | *unknown* |
| *Dwarfism* | AD | Shortening of long bones | *unknown* | *unknown* |
| *Peterbald* | AD | Hairless, brush coat | *unknown* | *unknown* |
| *LaPerm* | AD | Curly hair coat | *unknown* | *Unknown* |

‡ Mode of inheritance of the non-wild type variant.  A “+” implies the wild type allele. If reference to the mutant allele, AD implies autosomal dominant, AR implies autosomal recessive, co-D implies co-dominant.

**Table 2** Common commercialized DNA tests for diseases of domestic cats and breeds.

| Disease / Trait (alleles) | MOI‡ | Phenotype | Breeds | Gene | Mutation |
|---|---|---|---|---|---|
| AB Blood Type ($A^+$, b)[71] | AR | Determines Type B | All cats | *CMAH* | c.1del-53_70, c.139G>A |
| Craniofacial Defect | AR | Craniofacial Defect | Burmese | | Unpublished |
| Gangliosidosis 1[72] | AR | Lipid storage disorder | Korat, Siamese | *GBL1* | c.1457G>C |
| Gangliosidosis 2[73] | AR | Lipid storage disorder | Burmese | *HEXB* | c.1356del-1_8, c.1356_1362delGTTCTCA |
| Gangliosidosis 2[74] | AR | Lipid storage disorder | Korat | *HEXB* | c.39delC |
| Glycogen Storage Dis. IV[75] | AR | Glycogen storage disorder | Norwegian Forest | *GBE1* | IVS11+1552_IVS12-1339 del6.2kb ins334 bp |
| Hypertrophic Cardiomyopathy[60] | AD | Cardiac disease | Maine Coon | *MYBPC* | c.93G>C |

| | | | | | |
|---|---|---|---|---|---|
| Hypertrophic Cardiomyopathy[76] | AD | Cardiac Disease | Ragdoll | *MYBPC* | c.2460C>T |
| Hypokalemia[38] | AR | Potassium deficiency | Burmese | *WNK4* | c.2899C>T |
| Progressive Retinal Atropy[42] | AR | Late onset blindness | Abyssinian | *CEP290* | IVS50 + 9T>G |
| Progressive Retinal Atropy[41] | AD | Early onset blindness | Abyssinian | *CRX* | c.546delC |
| Polycystic Kidney Disease[78] | AD | Kidney cysts | Persian | *PKD1* | c.10063C>A |
| Pyruvate Kinase Def.[43] | AR | Hemopathy | Abyssinian | *PKLR* | c.693+304G>A |
| Spinal Muscular Atrophy[79] | AR | Muscular atrophy | Maine Coon | *LIX1-LNPEP* | 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. A "+" implies the wild type allele. If

reference to the mutant allele, AD implies autosomal dominant, AR implies autosomal recessive, co-D implies co-dominant.

**Table 3** Domestic cat DNA testing laboratories.

| Laboratory and Webpage | Region | Univ. Research Affiliate | ID | Cat Test† Disease | Color | Blood | Coat |
|---|---|---|---|---|---|---|---|
| Animal DNA Testing<br>www.animalsdna.com | Australia | | Yes | 8 | 4 | Yes | Long |
| Animal Health Trust<br>www.aht.org.uk | UK | Animal Health Trust | Yes | PKD | No | No | No |
| Antagene Immeuble Le Meltem<br>www.antagene.com | France | | Yes | 4 | Color | Yes | No |
| BioAxis DNA Research Centre Ltd.<br>www.dnares.in | India | | Yes | PKD | No | No | No |
| DNA Diagnostics Center<br>www.dnacenter.com | USA | | No | PKD | No | No | No |
| GENINDEXE<br>www.genindexe.com | France | | Yes | 7 | 5 | Yes | No |
| Genoscoper<br>www.genoscoper.com | Finland | | Yes | 7 | Yes | Yes | Long |
| Gribbles<br>www.gribblesvets.com | Australia | | No | PKD | No | No | No |
| IDEXX<br>www.idexx.ca | Canada | | No | PK def. | No | No | No |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| Laboklin<br>www.laboklin.de/ | Germany | | Yes | 9 | 5 | Yes | Long |
| Langford Veterinary Services<br>Langfordvets.co.uk | UK | Bristol | Yes | 10 | 8 | Yes | Long |
| PennGen<br>research.vet.upenn.edu/penngen‡ | USA | Pennsylvania | No | PK Def. GSD | No | No | No |
| PROGENUS S.A.<br>www.progenus.be | Belgium | | Yes | 7 | 6 | No | Long |
| Van Haeringen Laboratory<br>www.vhlgenetics.com | Netherlands | | Yes | 10 | 6 | Yes | Long |
| Veterinary Cardiac Genetics Lab<br>VCGL@vetmed.wsu.edu | USA | Washington St. | No | HCM | No | No | No |
| Veterinary Genetics Lab<br>www.vgl.ucdavis.edu | USA | California, Davis | Yes | 14 | All | Yes | All |
| VetGen<br>www.vetgen.com | USA | Michigan | Yes | No | Brown Dilute | No | Long |
| Vetogene<br>www.vetogene.com | Italy | Milan | Yes | 5 | No | Yes | Long |

† Tests reference to those listed in Tables 1- 2. If a laboratory offers only one or two test, those tests are listed. PKD and the HCMs are the most popular cat tests offerings. ‡ PennGen also offers tests for diseases in Table 5 that are not of concern to the cat breeds or cat population in general.