Feline genetic disease mutations screened by DNA microarray, according to Anderson et al. (2022). In the study, 50 disease mutation sites were looked at in a total sample size of over 11,000 purebred and domestic cats. Diseases where disease associated variants were detected are highlighted in yellow.

DISEASE-ASSOCIATED VARIANTS

Acute Intermittent Porphyria (Variant 1)
Acute Intermittent Porphyria (Variant 2)
Acute Intermittent Porphyria (Variant 3)
Acute Intermittent Porphyria (Variant 4)
Acute Intermittent Porphyria (Variant 5)
Autoimmune Lymphoproliferative Syndrome
Burmese Head Defect (Discovered in the Burmese)
Congenital Adrenal Hyperplasia
Congenital Erythropoietic Porphyria c.331G>A
Congenital Erythropoietic Porphyria c.140T>C
Congenital Myasthenic Syndrome (Discovered in the Devon Rex and Sphynx)
Cystinuria Type 1A
Cystinuria Type B (Variant 1)
Cystinuria Type B (Variant 2)
Cystinuria Type B (Variant 3)
Dihydropyrimidinase Deficiency
Earfold and Osteochondrodysplasia (Discovered in the Scottish Fold)
Factor XII Deficiency (Variant 1)
Factor XII Deficiency (Variant 2)
Familial Episodic Hypokalemic Polymyopathy (Discovered in the Burmese)
Glutaric Aciduria Type II
Glycogen Storage Disease (Discovered in the Norwegian Forest Cat)
GM1 Gangliosidosis
GM2 Gangliosidosis
GM2 Gangliosidosis Type II (Discovered in Domestic Shorthair cats)
GM2 Gangliosidosis Type II (Discovered in Japanese domestic cats)
GM2 Gangliosidosis Type II (Discovered in the Burmese)
Hemophilia B (Variant 1)
Hemophilia B (Variant 2)
Hyperoxaluria Type II
Hypertrophic Cardiomyopathy (Discovered in the Maine Coon)
Hypertrophic Cardiomyopathy (Discovered in the Ragdoll)
Hypotrichosis (Discovered in the Birman)
Lipoprotein Lipase Deficiency
MDR1 Medication Sensitivity
Mucopolysaccharidosis Type I
Mucopolysaccharidosis Type VI

Mucopolysaccharidosis Type VI Modifier
Mucopolysaccharidosis Type VII (Variant 1)
Mucopolysaccharidosis Type VII (Variant 2)
Myotonia Congenita
Polycystic Kidney Disease (PKD)
Progressive Retinal Atrophy (Discovered in Abyssinian)
Progressive Retinal Atrophy (Discovered in the Bengal)
Progressive Retinal Atrophy (Discovered in the Persian)
Pyruvate Kinase Deficiency
Sphingomyelinosis (Variant 1)
Sphingomyelinosis (Variant 2)
Spinal Muscular Atrophy (Discovered in the Maine Coon)
Vitamin D-Dependent Rickets