

General Information

Frequencies of Disease Alleles *

* These data have been published in [Anderson et al. en 2022](#) and mainly concerns cats from North America and Finland. The frequencies of disease alleles in cats from other regions of the world remain to be determined.

Frequency of the mutated allele in the following breeds:

Cystinuria Type B SLC7A9 c.881T>A	Usually observed in these breeds:	0.2%	Maine Coon
	Also found in other breeds (not observed before 2022) :	0.7% 0.02%	Siberian Maine Coon Polydactyl
Factor XII Deficiency F12 c.1321delC	Usually observed in these breeds:	9.9% 4.8% <i>Detected :</i>	Maine Coon Domestic Cat <i>Bengal, Siamese</i>
	Also found in other breeds (not observed before 2022) :	6.4% <i>Detected :</i>	Savannah <i>American SH, Balinese, Cymric, Highlander, Himalayan, Maine Coon Polydactyl, Manx, Minuet, Munchkin, Ragdoll, Tennessee Rex</i>
Factor XII Deficiency F12 c.1631G>C	Usually observed in these breeds:	23.3% 20.3% 10.6% 9.5%	Domestic Cat Bengal Maine Coon Siamese
	Also found in other breeds (not observed before 2022) :	47.0% 17.5% 15.8% 12.0% 9.7% 8.1% <i>Detected :</i>	Selkirk Rex Devon Rex Lykoi Ragdoll Scottish Fold Shorthair Sphynx <i>Balinese, Bombay, British Shorthair, Cashmere, Donskoy, Exotic SH, Highlander, Himalayan, Maine Coon Polydactyl, Minuet, Munchkin, Neva Masquerade, Munchkin , Oriental, Persan, Peterbald, Savannah, Selkirk Rex LH, Scottish Straight, Siberian, Tennessee Rex, Turkish Angora</i>
Gangliosidosis Type II (GM2-B) HEXB c.1244-8_1250del	Usually observed in these breeds:	0.9%	Burmese
Hypertrophic Cardiomyopathy (HCM-Maine Coon) MYBPC3 c.91G>C (A31P)	Usually observed in these breeds:	0.8%	Maine Coon
	Also found in other breeds (not observed before 2022) :	6.7% 0.3% <i>Detected :</i>	Maine Coon Polydactyl Domestic Cat <i>Pixiebob LH, Siberian</i>
Hypertrophic Cardiomyopathy (HCM-Ragdoll) MYBPC3 c.2460C>T (R820W)	Usually observed in these breeds:	3.0%	Ragdoll
	Also found in other breeds (not observed before 2022) :	0.2% <i>Detected :</i>	Domestic Cat <i>American Bobtail, Highlander, Munchkin, RagaMuffin</i>
Medication Sensitivity (MDR1) ABCB1 c.1930_1931delTC	Usually observed in these breeds:	0.8% 0.7% 0.0% 0.0%	Domestic Cat Siamese Ragdoll Russe
	Also found in other breeds (not observed before 2022) :	6.7% 5.4% 2.6% 0.9%	Maine Coon Polydactyl Maine Coon Balinese Turkish Angora



FELOME

54 Rue Molière
94200 Ivry-sur-Seine FRANCE

www.felome.fr
contact@felome.com

General Information

Frequencies of Disease Alleles *

* These data have been published in [Anderson et al. en 2022](#) and mainly concerns cats from North America and Finland. The frequencies of disease alleles in cats from other regions of the world remain to be determined.

Frequency of the mutated allele in the following breeds:

Polycystic Kidney Disease (PKD1) PKD1 c.10063C>A	Usually observed in these breeds:	2.6% 1.5% 0.2% 0.09% 0.0%	Scottish Fold Shorthair Exotic Shorthair Siberian Ragdoll Persan
	Also found in other breeds (not observed before 2022) :	1.6% 0.3%	Scottish Straight Maine Coon
Progressive Retinal Atrophy - Bengal (Type PRA-b) KIF3B c.1000G>A	Usually observed in these breeds:	18.9%	Bengal
	Also found in other breeds (not observed before 2022) :	Detected :	<i>Bengal Mix, Highlander, Highlander SH, Savannah</i>
Progressive Retinal Atrophy (Type PRA-rdAc) CEP290 IVS50+9T>G	Usually observed in these breeds:	47.0% 30.1% 29.2% 23.0% 19.7% 12.7% 10.7%	Peterbald Siamese Cornish Rex Oriental Shorthair Balinese Somali Abyssinian
	Also found in other breeds (not observed before 2022) :	33.3% 12.5% 5.9% 3.3% 3.1% 0.2% Detected :	Oriental Longhair Savannah Donskoy Havana Brown Devon Rex Domestic Cat <i>American SH, European SH, Highlander, Maine Coon, Manx, Pixiebob LH, Ragdoll, Scottish Fold SH, Sphynx, Tennessee Rex</i>
Pyruvate Kinase Deficiency (PKDef) PKLR c.693+304 G>A	Usually observed in these breeds:	23.5% 14.3% 14.1% 14.1% 11.2% 5.5% 3.3% 2.9% Detected :	Egyptian Mau Maine Coon Savannah Somali Bengal Abyssinian Norwegian Forest Cat Domestic Cat <i>LaPerm, Singapura</i>
	Also found in other breeds (not observed before 2022) :	26.3% 20.2% 6.7% 4.3% Detected :	Pixiebob Maine Coon Polydactyl Lykoi Neva Masquerade <i>Caracat, Chausie, European SH, Highlander, Highlander SH, Minuet LH, Munchkin, Pixiebob LH, Toyger</i>
Spinal Muscular Atrophy (SMA) LIX1 (& LNPEP) 140kb del exons 4-6	Usually observed in these breeds:	0.6%	Maine Coon
	Also found in other breeds (not observed before 2022) :	Detected :	<i>Highlander, Maine Coon Polydactyl</i>



FELOME

54 Rue Molière
94200 Ivry-sur-Seine FRANCE

www.felome.fr
contact@felome.com