GENOMIA
genetic laboratory

Result certificate #012345

Screening for hereditary diseases and appearance traits of the cat

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic
Sample:
Sample: 08-12346
Date received: 01.01.2008
Sample type: buccal swab

Information provided by the customer
Name: Madame Théophile DEMO
Breed: Persian cat
Date of birth: 31.12.1909
Reg. number: (CZ)ABCD EF 123/45/XYZ
Microchip: 123 456 789 012 345
Sex: female
Date of sampling: 01.01.2008
The identity of the animal has been checked.

Appearance

Name	Abbr.	Gene	Mutation	Copies	Result
Locus A		ASIP	c.123_124del	0	no effect
Golden Coat colour (discovered in Siberian cats)	Sunshine vwb^SIB	CORIN	c.2383C>T	0	no effect
Golden Coat colour (discovered in Siberian cats)	Extreme sunshine vwb^eSIB	CORIN	c.839G>A	0	no effect
Golden Coat colour (discovered in British Shorthair cats)	Copper BSH	CORIN	c.2425C>T	0	no effect
Coat colour, ticked (discovered in Abyssinian cats)	Ti^CK	DKK4	c.53C>T	0	no effect
Coat colour, ticked (discovered in Abyssinian cats)	Ti^A	DKK4	c.188G>A	0	no effect
Long Hair		FGF5	c.356_357insT	0	no effect
Long Hair		FGF5	c.406C>T	0	no effect
Long Hair		FGF5	c.475A>C	0	no effect
Long Hair		FGF5	c.474del	0	no effect
Hypotrichosis, with short life expectancy		FOXN1	c.1030_1033del	0	no effect
White Feet, gloving (discovered in Birma cats)	g	KIT	c.1035_1036delinsCA	0	no effect
Hypotrichosis (discovered in Sphynxs)		KRT71	c.816+1G>A	0	no effect
Curly coat (discovered in Selkirk rex)		KRT71	c.445-1G>C	0	no effect
Curly coat (discovered in Ural Rex)	urx	LIPH	c.477_483delinsC	0	no effect
Curly/woolly coat (discovered in Cornish Rex and German Rex)	r	LPAR6	c.250_253del	0	no effect
Coat colour, Blotched tabby	Ta^b2	LVRN	c.176C>A	0	no effect
Coat colour, Blotched tabby	Ta^b1	LVRN	c.682G>A	0	no effect
Coat colour, Blotched tabby	Ta^b3	LVRN	c.2522G>A	0	no effect
Red-brown colouration (discovered in Burmese cats)	er	MC1R	c.440_442del	0	no effect
Amber Coat colour (discovered in Norwegian Forest Cats)		MC1R	c.250G>A	0	no effect
Copal coat colour (discovered in Kurilian Bobtails)		MC1R	c.640_669del	0	no effect

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Locus D	d	MLPH	c.83del	0	no effect
Coat/skin colour, Burmese coat pattern	c^b	TYR	g.45907839C>A	1	may affect
Coat/skin colour, oculocutaneous albinism type I, complete albinism	c	TYR	c.939del	0	no effect
Coat/skin colour, Siamese coat pattern	c^s	TYR	c.904G>A	0	no effect
Coat/skin colour, oculocutaneous albinism type I	c^2	TYR	c.1204C>T	0	no effect
Locus B, chocolate allele	b	TYRP1	c.8C/G	0	no effect
Locus B, cinnamon allele	bl	TYRP1	c.298C/T	2	cinnamon

Autosomal dominant disorders

Name	Abbr.	Gene	Mutation	Copies	Result
Frontonasal dysplasia (discovered in Burmese cats)		ALX1	c.497_508del	0	clear
Porphyria, acute intermittent (discovered in Siamese cats)	AIP	HMBS	g.16544592_16544594del	0	clear
Porphyria, acute intermittent	AIP	HMBS	g.16542541C>T	0	clear
Porphyria, acute intermittent	AIP	HMBS	g.16544575G>A	0	clear
Porphyria, acute intermittent	AIP	HMBS	g.16540928_16540931del	0	clear
Hypertrophic Cardiomyopathy (discovered in Ragdolls)	HCM	MYBPC3	c.2453C>T	0	clear
Hypertrophic Cardiomyopathy (discovered in Domestic Shorthair cats)	HCM	MYH7	c.5647G>A	0	clear
Polycystic kidney disease (discovered in Persian cats)	PKD	PKD1	c.10063C>A	0	clear
Polydactyly	Pd^Hw	SHH	g.169532844T>C	0	clear
Polydactyly	Pd^UK1	SHH	g.169532842T>A	0	clear
Polydactyly	Pd^UK2	SHH	g.169533066C>G	0	clear
Ears, folded (discovered in Scottish Fold cats)	SFOCD	TRPV4	c.1024G>T	0	clear

Autosomal recessive disorders

Name	Abbr.	Gene	Mutation	Copies	Result
Hypothyroidism Congenital (discovered in Domestic Shorthair cats)		A445T / TPO	c.1333G>A	0	clear
Multidrug resistance 1	MDR1	ABCB1	c.1930_1931del	0	clear
Leber Congenital Amaurosis (discovered in Persian cats)	Pd-PRA	AIPL1	c.577C>T	0	clear
Hypertrophic Cardiomyopathy (discovered in Sphynxs)	HCM4	ALMS1	c.7384G>C	0	clear
Mucopolysaccharidosis VI (discovered in Domestic Shorthair and Siamese cats)	MPS VI	ARSB	c.1427T>C	0	clear
Mucopolysaccharidosis VI (discovered in Siamese cats)	MPS VI	ARSB	c.1558G>A	0	clear
Retinal degeneration	PRA-rdAc	CEP290	c.7584+9T>G	0	clear
Myotonia		CLCN1	c.1930+1G>T	0	clear
Muscular dystrophy-dystroglycanopathy (discovered in Devon Rex, Sphynx)		COLQ	c.1190G>A	0	clear
Congenital Adrenal Hyperplasia	CAH	CYP11B1	c.1151G>A	0	clear
Vitamin D-deficiency rickets, type I (discovered in Siamese cats)		CYP27B1	c.637G>T	0	clear
Dihydropyrimidinase deficiency	DHP def	DPYS	c.1303G>A	0	clear
Factor XI deficiency (discovered in Maine Coon cats)		F11	c.1546G>A	0	clear
Factor XII deficiency		F12	c.1321del	0	clear

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Autoimmune lymphoproliferative syndrome (discovered in British Shorthair cats)	ALPS	FASLG	c.418dup	0	clear
Gangliosidosis, GM1	GM1	GLB1	c.1448G>C	0	clear
Gangliosidosis, AB variant	GM2	GM2A	c.516_519del	0	clear
Primary hyperoxaluria type II	PH2	GRHPR	c.507-1G>A	0	clear
Mucopolysaccharidosis VII (discovered in Domestic Shorthair cats)	MPS VII	GUSB	c.1423T>G	0	clear
Mucopolysaccharidosis VII (discovered in Domestic Shorthair cats)	MPS VII	GUSB	c.1426C>T	0	clear
Mucopolysaccharidosis VII (discovered in Domestic Shorthair cats)	MPS VII	GUSB	c.1051G>A	0	clear
Gangliosidosis type 2 (discovered in Domestic Shorthair cats)	GM2	HEXB	c.1467_1491inv	0	clear
Gangliosidosis type 2 (discovered in Burmese cats)	GM2	HEXB	c.1244-8_1250del	0	clear
Gangliosidosis type 2 (discovered in Japanese Domestic cats)	GM2	HEXB	c.667C>T	0	clear
Gangliosidosis type 2 (discovered in Korat)	GM2	HEXB	c.39del	0	clear
Mucopolysaccharidosis I (discovered in Domestic Shorthair cats)	MPS I	IDUA	c.1042_1044del	0	clear
Progressive Retinal Atrophy (discovered in Bengal cats)	PRA	KIF3B	c.1000G>A	0	clear
Hyperlipoproteinaemia		LPL	c.1315G>A	0	clear
Primary Congenita Glaucoma 3 (discovered in Siamese cats)		LTBP2	c.1449_1452dup	0	clear
Mannosidosis, alpha (discovered in Persian cats)		MAN2B1	c.1749_1752del	0	clear
Hypertrophic Cardiomyopathy (discovered in Maine Coon cats)	HCM	MYBPC3	c.91G>C	0	clear
Niemann-Pick disease, type C1/C2 (discovered in Domestic Shorthaired cats)		NPC1	c.1322A>C	0	clear
Niemann-Pick disease, type C1/C2 (discovered in Domestic Shorthaired cats)		NPC1	c.2864G>C	0	clear
Pyruvate kinase deficiency of erythrocyte	PK def	PKLR	c.693+304G>A	0	clear
Acrodermatitis enteropathica (discovered in Turkish Van)		SLC39A4	c.1057G>C	0	clear
Cystinuria, type I – A (discovered in Domestic Shorthair cats)		SLC3A1	c.1342C>T	0	clear
Porphyria, congenital erythropoietic	CEP	UROS	g.83482394G>A	0	clear
Porphyria, congenital erythropoietic	CEP	UROS	g.83467845C>T	0	clear
Hypokalaemic periodic paralysis (discovered in Burmese cats)	BHP	WNK4	c.2899C⊠T	0	clear

Association genetic tests

Name	Abbr.	Gene	Mutation	Copies	Result
Genetic Blood Group		CMAH			A/b
Cystinuria, type B		SLC7A9	c.706G>A	0	no effect
Cystinuria, type B (discovered in Maine Coon cats)		SLC7A9	c.881T>A	0	no effect
Cystinuria, type B		SLC7A9	c.1175C>T	0	no effect
Locus B, chocolate allele	b	TYRP1	c.1261+5G>A	0	no effect
Feline Infectious Peritonitis Resistance	428T	fIFNG	c.428T>C	0	no effect

X-linked hereditary disorders

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Name	Abbr.	Gene	Mutation	Copies	Result
Haemophilia B (discovered in Domestic Longhaired cats)		F9	c.383G>A	0	no effect
Haemophilia B		F9	c.1150C>T	0	no effect

Markers without description

Name	Abbr.	Gene	Mutation	Copies	Result
MC1-CLCN2					TTCAGGGT
MC1-CLCN3					G/G

Explanation

Interpretation of the results can be found on the website https://www.genomia.cz/en/veterinarni/kocky/ on the pages of the respective examinations.

The mutations listed are annotated according to the Felis_catus_9.0 reference sequence.

Recessive inheritance: the trait (disease) becomes apparent if the individual inherits it from both parents (2 copies); carriers of the trait (disease) are asymptomatic but pass the causal mutation on to the next generation (1 copy).

Dominant inheritance: it is sufficient for an individual to inherit the trait (disease) from one parent (1 copy).

X-linked recessive inheritance: in males, 1 copy of the mutated gene is enough to cause the disease; in females, 2 copies of the mutated gene are needed to cause the disease.

The results of the association genetic tests indicate the predisposition to the trait. This is not a detection of a causal mutation.

Method: SOPAgriseq_feline, MPS

Date of issue: 06.01.2008
Date of testing: 12.06.2008 - 06.01.2008
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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