

Optimal Selection™

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LABORATORIES

KTBR 0531 3

Sarah Blue Ridge, Maine Coon

Registered Name: Sarah Blue Ridge

Owner: Traci Heppner

Call Name: Nymphadora

Country: United States

Breed: Maine Coon

Testing date: 2020/3/25

Gender: Female

Test results - Known disorders in the breed

Disorder	Type	Mode of Inheritance	Result
Cystinuria; SCL7A9 mutation: c.881A>T	Metabolic Disorders	Autosomal Recessive	Clear
Erythrocyte Pyruvate Kinase (PK) Deficiency	Blood Disorders	Autosomal Recessive	Clear
Factor XII Deficiency	Blood Disorders	Autosomal Recessive	Clear
Hypertrophic Cardiomyopathy, MYBPC3 mutation: A31P found in Maine Coon	Cardiac Disorders	Autosomal Dominant (Incomplete Penetrance)	Clear
Spinal Muscular Atrophy (SMA)/Spinal Muscular Dystrophy	Muscular Disorders	Autosomal Recessive	Clear

Test results - New potential disorders in the breed

Disorder	Type	Mode of Inheritance	Result
Blood-brain Barrier Dysfunction, MDR1 / ABCB1	Pharmacogenetics	Autosomal Recessive	Clear

On behalf of Genoscooper Laboratories,



SIGNATURE

Jonas Donner, PhD, Head of Research and Development
at Genoscooper Laboratories

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Test results - Traits - page 1

Blood Type

Trait	Genotype	Description
Blood Type (3 variants)	N/N	Cat has blood type A.

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Test results - Traits - page 2

Coat Color

Trait	Genotype	Description
Color Locus B: Chocolate and Cinnamon (2 variants)	B/B	The cat produces black pigment.
Color Locus C: Pointed Coloration and Albinism (3 variants)	C/C	This cat has full color with no color restrictions.
Color Locus A: Agouti and Charcoal (2 variants)	a/a	The cat is likely to have non-agouti (solid) coat color.
any FERV1 insertion in the <i>KIT</i> gene	N/N	The cat has no copy of the FERV1 insertion in the <i>KIT</i> gene. The cat's coat color is not affected by White spotting or Dominant White mutations.
<i>MLPH</i> T83del (d allele)	D/D	The cat does not have color dilution.
<i>MC1R</i> c.250G>A (e allele)	E/E	The cat does not have e allele for Amber color found in Norwegian Forest Cat.
<i>KIT</i> c.1035_1036delinsCA	N/N	The cat does not have <i>KIT</i> mutation associated with gloving pattern in Birman cats.
<i>MC1R</i> (e ^r allele)	-/-	The cat does not have e ^r allele for Russet color found in Burmese.

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Test results - Traits - page 3

Coat Type

Trait	Genotype	Description
Long Hair (4 variants)	M3/M3	The cat is likely to have long hair.
<i>LPAR6</i> c.250_253_delTTTG	N/N	The cat does not have Cornish Rex curly coat.
<i>KRT71</i> c.445-1C	N/N	The cat does not have Selkirk Rex curly coat.

Morphology

Trait	Genotype	Description
Short tail, T-box mutations (3 variants)	N/N	The cat does not have any of the tested bobtail mutations originally found in Manx.
Polydactyly (3 variants)	N/N	The cat does not have any of the tested mutations causing extra digits.
<i>HES7</i> c.T5C	T/T	The cat has no bobtail mutation originally found in Japanese Bobtail.

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Test results - Additional disorders found in other breeds - page 1

Blood Disorders

Disorder	Mode of Inheritance	Result
Hemophilia B, mutation F9: c.1014C>T	X-linked Recessive	Clear
Hemophilia B, mutation F9: c.247G>A	X-linked Recessive	Clear

Cardiac Disorders

Disorder	Mode of Inheritance	Result
Hypertrophic Cardiomyopathy, MYBPC3 mutation: c.2460C>T found in Ragdoll	Autosomal Dominant (Incomplete Penetrance)	Clear

Endocrine Disorders

Disorder	Mode of Inheritance	Result
Congenital Adrenal Hyperplasia	Autosomal Recessive	Clear

Immunologic Disorders

Disorder	Mode of Inheritance	Result
Autoimmune Lymphoproliferative Syndrome	Autosomal Recessive	Clear
Congenital Hypotrichosis with Short Life Expectancy	Autosomal Recessive	Clear

Test results - Additional disorders found in other breeds - page 2

Metabolic Disorders

Disorder	Mode of Inheritance	Result
Acute Intermittent Porphyria	Autosomal Dominant	Clear
Acute Intermittent Porphyria; HMBS mutation: c.107_110delACAG	Autosomal Dominant	Clear
Acute Intermittent Porphyria; HMBS mutation: c.826-1G>A	Autosomal Dominant	Clear
Acute Intermittent Porphyria; HMBS mutation: c.844delGAG	Autosomal Dominant	Clear
Chylomicronemia, Lipoprotein Lipase Deficiency	Autosomal Recessive	Clear
Congenital Erythropoietic Porphyria, mutation UROS: c.331G>A	Autosomal Recessive	Clear
Cystinuria; SCL3A1 mutation	Autosomal Recessive	Clear
Cystinuria; SCL7A9 mutation: c.1175C>T	Autosomal Recessive	Clear
Cystinuria; SCL7A9 mutation: c.706G>A	Autosomal Recessive	Clear
Dihydropyrimidinuria	Autosomal Recessive	Clear
Mucopolysaccharidosis Type I	Autosomal Recessive	Clear
Mucopolysaccharidosis Type VI (MPS VI), Typical Form	Autosomal Recessive	Clear
Mucopolysaccharidosis Type VII, mutation GUSB: c.1074G>A	Autosomal Recessive	Clear
Mucopolysaccharidosis VII; GUSB mutation C1424T	Autosomal Recessive	Clear
Vitamin D-Dependent Rickets (VDDR-1A); CYP27B mutation: c.G637T	Autosomal Recessive	Clear

Muscular Disorders

Disorder	Mode of Inheritance	Result
Congenital Myasthenic Syndrome (CMS)	Autosomal Recessive	Clear
Myotonia Congenita	Autosomal Recessive	Clear
Periodic Hypokalemic Polymyopathy, Burmese Hypokalemia, or Familial Episodic Hypokalaemic Polymyopathy	Autosomal Recessive	Clear

Test results - Additional disorders found in other breeds - page 3

Neurologic Disorders

Disorder	Mode of Inheritance	Result
Feline GM1 Gangliosidosis	Autosomal Recessive	Clear
GM2 Gangliosidosis, Domestic Shorthair mutation HEXB: c.1467_1491inv	Autosomal Recessive	Clear
GM2 Gangliosidosis, Japanese Domestic mutation HEXB: c.667C>T	Autosomal Recessive	Clear
GM2 Gangliosidosis; Domestic Shorthair GM2A Mutation	Autosomal Recessive	Clear
Niemann-Pick C2, NPC Disease, Sphingomyelinosis NPC2 Mutation	Autosomal Recessive	Clear
Niemann-Pick C1, NPC Disease, Sphingomyelinosis NPC1 Mutation	Autosomal Recessive	Clear

Neuromuscular Disorders

Disorder	Mode of Inheritance	Result
Glycogen Storage Disease Type IV	Autosomal Recessive	Clear

Ocular Disorders

Disorder	Mode of Inheritance	Result
Bengal Progressive Atrophy	Autosomal Recessive	Clear
Retinal Dystrophy (rdAc)	Autosomal Recessive	Clear

Renal Disorders

Disorder	Mode of Inheritance	Result
Hyperoxaluria	Autosomal Recessive	Clear
Polycystic Kidney Disease	Autosomal Dominant	Clear

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APPENDIX

Explanation of the results of the tested disorders

Autosomal recessive inheritance (ARI)

Clear - A cat carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - A cat carries one copy of the tested mutation. Carriers typically have a normal, healthy appearance but pass on the mutation to approximately 50% of their offspring.

At risk - A cat carries two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

Autosomal dominant inheritance (ADI)

Clear - A cat carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

At risk - A cat carries one or two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

X-linked recessive inheritance (X-linked)

Clear - A cat carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - Female carriers typically have a normal, healthy appearance but carry one copy of the tested mutation on one of their X chromosomes. As males only have one X chromosome, there are no male carriers.

At risk - Female cats at risk carry two mutated copies of the tested mutation. Males carry one copy of the tested mutation on their single X chromosome. Cats at risk are at high or increased risk of developing the disease/condition.

Please note that the descriptions above are generalized based on typically observed inheritance patterns. When obtaining a 'carrier' or 'at risk' test result, always refer to the corresponding online test documentation for more detailed information on the condition and any exceptions.

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Terms and Conditions

MyCatDNA™ Genetic Breeding Analysis is a proprietary process designed and intended to be used on purebred cats solely to 1) Help quantify the genetic compatibility of potential breeding pairs and 2) To identify specific alleles or DNA mutations that are associated with certain inherited diseases or traits. No other purpose is authorized or permitted. It is not intended to diagnose diseases or predict behavior in any particular cat.

Upon receipt of your cat's DNA sample, Genoscoper Laboratories will analyze your cat's DNA to determine chromosomal similarities and differences in the genetic profile of a potential sire and dam and provide a match analysis. Your cat's DNA will also be analyzed for the presence of specific alleles that are associated with inherited conditions identified as occurring in your cat's breed. Genoscoper Laboratories' testing procedures are designed to provide reliable and accurate results, but are not guaranteed. By submitting your cat's sample(s) for MyCatDNA™ analysis it is understood that you agree that the sample(s), analysis, results and related information may be used confidentially by Genoscoper Laboratories in conjunction with other samples to increase the understanding of the breed's genetic structure, as well as for internal, research and development, or statistical purposes and may be shared with third parties for these purposes.

Samples may be disposed of or stored at Genoscoper Laboratories' option and will not be returned. Please view the full Mars Privacy Policy here: <http://www.mars.com/global/policies/privacy/pp-english.aspx>. It is also understood that future releases of the MyCatDNA™ test may refine results as more information is obtained regarding the breed structure and/or if new genetic markers are included.

MyCatDNA™ genetic assessments for individual cats and potential mates will be available online to the person(s) who registered the sample. A cat's results, photo and other information may be shared by the owner with other individuals whom they choose or transferred to a new owner if the cat changes ownership. The content of such online services 1) may be altered due to changes, additions, or removals of a cat's information in the MyCatDNA™ database or due to changes in technical or other design of such services and 2) includes information about third parties and other Genoscoper Laboratories clients' cats, which Genoscoper Laboratories is not responsible or liable for. Genoscoper Laboratories has right to terminate access to online services one year from the purchase date, unless a longer period has been agreed upon.

You agree to Genoscoper Laboratories instructions related to ordering process, payment, sampling and sample delivery. You also certify that the animal described in your order is the same animal whose sample is submitted for analysis, and that all information is accurate. You warrant that you are entitled to obtain and supply samples to Genoscoper Laboratories.

In the unlikely event that it is not possible to provide an analysis (for example due to an insufficient DNA sample) or that an error in the analysis occurs, liability by Genoscoper Laboratories or related companies and individuals is disclaimed and damages in any event are limited to the payment actually received by Genoscoper Laboratories for the specified analysis at issue. Genoscoper Laboratories' study of the complexities of the feline genome is ongoing with the goal of continuing to provide the most advanced and complete analysis possible.

Genoscoper Laboratories reserves the right to use any third party of its choice to undertake the testing, analysis or laboratory services for the analysis.