



8110 0133 8342
NO*Arctic Sapphire Sansa Stark, Ragdoll

Registered Name: NO*Arctic Sapphire Sansa Stark

Owner: Siril Malene Isaksen

Call Name: NO*Arctic Sapphire Sansa Stark

Country: Norway

Registration ID: (NO) NRR LO 185931

Testing date: 2019/9/15

Microchip: 578098100606806

Breed: Ragdoll

Gender: Female

Cat's identity verified from microchip or tattoo by veterinarian or other authorized person during sample taking: **No**

Test results - Known disorders in the breed

Disorder	Type	Mode of Inheritance	Result
Hypertrophic Cardiomyopathy, MYBPC3 mutation: c.2460C>T found in Ragdoll	Cardiac Disorders	Autosomal Dominant (Incomplete Penetrance)	Clear
Polycystic Kidney Disease	Renal Disorders	Autosomal Dominant	Clear

Test results - New potential disorders in the breed

Disorder	Type	Mode of Inheritance	Result
Retinal Dystrophy (rdAc)	Ocular Disorders	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)	cs/cs	The cat is likely to have Siamese type color point restriction.
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	Ws/Ws II DW/Ws II DW/DW	The cat has two copies of the <i>KIT</i> gene with FERV1 insertion. The cat with pattern of white spotting has two copies of the White spotting insert. The all white cat has either one Dominant White insert and one White spotting insert or two Dominant White inserts.
<i>MLPH</i> T83del (d allele)	D/d	The coat color is not diluted; the cat carries one copy of the dilute gene.
<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)	M1/M4	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
Erythrocyte Pyruvate Kinase (PK) Deficiency	Autosomal Recessive	Clear
Factor XII Deficiency	Autosomal Recessive	Clear
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: A31P found in Maine Coon	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



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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
Cystinuria; SCL7A9 mutation: c.881A>T	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



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

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
Spinal Muscular Atrophy (SMA)/Spinal Muscular Dystrophy	Autosomal Recessive	Clear

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



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Test results - Additional disorders found in other breeds - page 4

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



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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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