8110 0121 2018

Estya JeNaty, Ragdoll

Registered Name: Estya JeNaty
Call Name: Estya
Microchip: 756098000039546
Breed: Ragdoll
Gender: Female

Owner: Natacha Wäckerling
Country: Switzerland
Testing date: 2020/1/15

Cat's identity verified from microchip or tattoo by veterinarian or other authorized person during sample taking: **Yes**

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 Genoscoper Laboratories,

SIGNATURE

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

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Breed: Ragdoll
Gender: Female

Owner: Natacha Wäckerling
Country: Switzerland
Testing date: 2020/1/15

Cat's identity verified from microchip or tattoo by veterinarian or other authorized person during sample taking: **Yes**

Test results - Traits - page 1

Blood Type

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

On behalf of Genoscoper Laboratories,

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Estya JeNaty, Ragdoll

Registered Name:	Estya JeNaty	**Owner:**	Natacha Wäckerling
Call Name:	Estya	**Country:**	Switzerland
Microchip:	756098000039546	**Testing date:**	2020/1/15
Breed:	Ragdoll		
Gender:	Female		

Cat's identity verified from microchip or tattoo by veterinarian or other authorized person during sample taking: **Yes**

Test results - Traits - page 2

Coat Color

Trait	Genotype	Description
Color Locus B: Chocolate and Cinnamon (2 variants)	b/b	The cat produces brown 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 *KIT* gene	N/Ws II N/DW	The cat has one FERV1 insertion in the KIT gene and one wild type allele. The cat with pattern of white spotting has one White spotting insert and all white cat has one Dominant White insert.
MLPH T83del (d allele)	D/d	The coat color is not diluted; the cat carries one copy of the dilute gene.
MC1R c.250G>A (e allele)	E/E	The cat does not have e allele for Amber color found in Norwegian Forest Cat.
KIT c.1035_1036delinsCA	N/g	The cat carries KIT mutation associated with gloving pattern in Birman breed. This variant is also found in other breeds of cats.
MC1R (e^r allele)	-/-	The cat does not have er allele for Russet color found in Burmese.

On behalf of Genoscoper Laboratories,

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

Registered Name:	Estya JeNaty	**Owner:**	Natacha Wäckerling
Call Name:	Estya	**Country:**	Switzerland
Microchip:	756098000039546	**Testing date:**	2020/1/15
Breed:	Ragdoll		
Gender:	Female		

Cat's identity verified from microchip or tattoo by veterinarian or other authorized person during sample taking: **Yes**

Test results - Traits - page 3

Coat Type

Trait	Genotype	Description
Long Hair (4 variants)	M4/M4	The cat is likely to have long hair.
LPAR6 c.250_253_delTTTG	N/N	The cat does not have Cornish Rex curly coat.
KRT71 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.
HES7 c.T5C	T/T	The cat has no bobtail mutation originally found in Japanese Bobtail.

On behalf of Genoscoper Laboratories,

Jonas Donner

SIGNATURE

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

8110 0121 2018

Estya JeNaty, Ragdoll

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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Estya JeNaty, Ragdoll

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

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

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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Estya JeNaty, Ragdoll

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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