



8110 0007 5259

Berkano Big Bang, Maine Coon

**Registered Name:** Berkano Big Bang

**Owner:** Zdeněk Soukup

**Call Name:** Berkano Big Bang of Eldarion

**Country:** Czech Republic

**Registration ID:** SBT 082417032

**Testing date:** 2018/8/15


**Microchip:** 985113000455371

**Breed:** Maine Coon

**Gender:** Male

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 found in Maine Coon	Cardiac Disorders	Autosomal Dominant (Incomplete Penetrance)	 At risk (heterozygote)
Cystinuria; SCL7A9 mutation: c.881A>T	Metabolic Disorders	Autosomal Recessive	Clear
Erythrocyte Pyruvate Kinase (PK) Deficiency	Blood Disorders	Autosomal Recessive	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
Factor XII Deficiency	Blood 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 or AB

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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 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/g	The cat carries <i>KIT</i> mutation associated with gloving pattern in Birman breed. This variant is also found in other breeds of cats.
<i>MC1R</i> (er allele)	-/-	The cat does not have er 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/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 Manx mutations.
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 Japanese Bobtail mutation.

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

### Blood Disorders

Disorder	Mode of Inheritance	Result
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 found in Ragdoll	Autosomal Dominant	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



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## 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

### Pharmacogenetics

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



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

### 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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