



8110 0007 6083

Joshuatree Cherry Berry of Eldarion, Maine Coon

Registered Name: Joshuatree Cherry Berry of Eldarion

Owner: Zdeněk Soukup

Call Name: Joshuatree Cherry Berry of Eldarion

Country: Czech Republic

Registration ID: SBT 080717008

Testing date: 2018/8/15

Microchip: 941000021208341

Breed: Maine Coon

Gender: Female

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 |
|---|---------------------|--|--------|
| Cystinuria; SCL7A9 mutation: c.881A>T | Metabolic Disorders | Autosomal Recessive | Clear |
| Erythrocyte Pyruvate Kinase (PK) Deficiency | Blood Disorders | Autosomal Recessive | Clear |
| Hypertrophic Cardiomyopathy 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 |
| 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 banded hair. The cat carries solid coat color. |
| any FERV1 insertion in the <i>KIT</i> gene | N/Ws II N/DW | The cat has one FERV1 insertion in the <i>KIT</i> 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. |
| <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) | M4/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 |



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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 |
| 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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