

# Optimal Selection™

POWERED BY GENOSCOPE®  
Laboratories

KTBR 0632 6

Mister Mega J Ise Rose, Maine Coon

**Registered Name:** Mister Mega J Ise Rose

**Owner:** Olga Winn

**Call Name:** Rosa

**Country:** United States

**Breed:** Maine Coon

**Testing date:** 2020/8/20

**Gender:** Female

## Test results - Known disorders in the breed

| Disorder   | Type                | Mode of Inheritance                        | Result |
|--|---------------------|--|--------|
| Erythrocyte Pyruvate Kinase (PK) Deficiency                            | Blood Disorders     | Autosomal Recessive                        | Clear  |
| Cystinuria; SCL7A9 mutation: c.881A>T                                  | Metabolic 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 banded hair. The cat carries 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> (e <sup>r</sup> allele)                         | -/-      | The cat does not have e <sup>r</sup> allele for Russet color found in Burmese.   |

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**Gender:** Female

## Test results - Traits - page 3

### Coat Type

| Trait                             | Genotype | Description   |
|-----------------------------------|----------|---|
| Long Hair (4 variants)            | N/M3     | The cat carries one copy of the long hair mutations and is likely to have short hair. |
| <i>LPAR6</i><br>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<br>(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  |

## 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.