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**Test Report**

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**Analysis of the genetic predisposition to disease in cats  
(CombiBreed Genetic Diseases)**

Name: Marcepanella Arkiza\*PL  
Breed: Maine Coon  
Lab-no.: KE201800009      Identification: 616093900537311  
Gender: female      Date of birth: 2017-03-31

Test	Inheritance	Result
Bloodtyping AB (DNA test)	recessive	N/N
Dihydropyrimidinase Deficiency	recessive	Normal
Gangliosidosis, GM2, GM2A	recessive	Normal
Gangliosidosis, GM2, Typ II - 1	recessive	Normal
Gangliosidosis, GM2, Typ II - 2	recessive	Normal
Glycogentoxicosis, GSD Typ IV	recessive	Normal
Hämophilia B - 1	X-chromosomal	Normal
Hämophilia B - 2	X-chromosomal	Normal
Hyperlipoproteinaemia	recessive	Normal
Hypertrophic Cardio Myopathy 1 (HCM1)	dominant (s. comment)	Normal
Hypertrophic Cardio Myopathy 3 (HCM3)	dominant (s. comment)	Normal
Hypothyroidism	recessive	Normal
Mucopolysaccharidosis I	recessive	Normal
Mucopolysaccharidosis VI	recessive	Normal
Mucopolysaccharidosis VII	recessive	Normal
Niemann-Pick Typ C	recessive	Normal

Test	Inheritance	Result
Congenital Adrenal Hyperplasia	recessive	Normal
Periodic Paralysis (WNK4 - Hypokalemia)	recessive	Normal
Polycystic Kidney Disease (PKD)	dominant (s. comment)	Normal
Primary Hyperoxaluria II	recessive	Normal
Progressive Retinal Atrophy (rdAc-PRA)	recessive	Normal
Pyruvatkinase Deficiency (PKDef)	recessive	Carrier
Spinale Muscular Atrophy (SMA)	recessive	Normal
Vitamin D-deficiency Rickets Typ I	recessive	Normal

**Note:** The analysis was performed in a partner laboratory.

**Comment:** Hypertrophic Cardiomyopathy 1 (HCM1): The test is based on the analysis of a mutation in the gene MYBPC3. The disease causing mutation was described for Maine Coons but may be relevant for other breeds, too. It might be possible that unaffected cats will become ill due to other, yet undescribed, mutations.  
Hypertrophic Cardiomyopathy 3 (HCM3): The test is based on the analysis of a mutation in the gene MYBPC3. The disease causing mutation was described for Ragdolls but may be relevant for other breeds, too. It might be possible that unaffected cats will become ill due to other, yet undescribed, mutations.  
Polycystic Kidney Disease (PKD): The test is based on the analysis of a mutation in the gene ADPKD1. It might be possible that unaffected cats will become ill due to other, yet undescribed, mutations.

Rheinbach, 2018-02-13

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( ) Dr. Jansen, Managing Director  
 ( ) Dr. van Haeringen, Managing Director  
 ( ) Dr. Weber, Proxy

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## Legend

### Recessive:

**Normal:** The animal does not carry the mutation. In case of use in breeding, this animal will not spread the predisposition into the population.

**Carrier:** The animal is heterozygous for the predisposition. In case of use in breeding, on average 50% of the offspring will inherit the mutated allele. Carriers of the predisposition will not become ill.

**Affected:** The animal has two mutated alleles. In case of use in breeding, all offspring will inherit the mutated allele. Affected animals will become ill.

### Dominant:

**Normal:** The animal does not carry the mutation. In case of use in breeding, this animal will not spread the predisposition into the population.

**Carrier:** The animal is heterozygous for the predisposition. In case of use in breeding, on average 50% of the offspring will inherit the mutated allele. Carriers of the predisposition will become ill.

**Affected:** The animal has two mutated alleles. In case of use in breeding, all offspring will inherit the mutated allele. Affected animals will become ill.

### X-chromosomal:

**Normal:** The female animal does not carry the mutation. In case of use in breeding, this animal will not spread the predisposition into the population.

Male animal carries a healthy copy of the X-chromosome and will not become ill.

**Carrier:** The female animal is heterozygous for the predisposition. In case of use in breeding, on average 50% of the offspring will inherit the mutated allele. Female animals will not become ill. Male animals have only one copy of the X-chromosome. Therefore, carriers of the mutation will become ill.

**Affected:** The female animal has two mutated alleles and will become ill. In case of use in breeding, all offspring will inherit the mutated allele.

Male animals have only one copy of the X-chromosome. Therefore, carriers of the mutation will become ill.

### Multifactorial:

**Normal:** The animal does not carry the mutation. In case of use in breeding, this animal will not spread the predisposition into the population.

**Carrier:** The animal is heterozygous for the predisposition. In case of use in breeding, on average 50% of the offspring will inherit the mutated (undesirable) allele.

**Affected:** The animal has two mutated alleles. In case of use in breeding, all offspring will inherit the mutated (undesirable) allele.