



dr. van haeringen polygen bvba

a VHLGenetics company

Faber Bina  
rue Freddy Terwagne 30 C  
BE-4577 STREE-LES-HUY  
Customer number 80760

Analysis Certificate

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#### Animal data

Name: UA BLUESMAINE VELMA  
Date of birth: 28.12.2015  
Sexe: Female  
Reg. nr.: BE02000166323  
Chip number: 981098106264709  
Breed: Maine Coon

#### Sample data

VHP\_ID: K451683  
Test ID-nr: 1000001823 1  
Material: Swab - Copan

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#### K754 - Pyruvatekinase Def. - Date of test: 14.08.2017

Testresult: NORMAL

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#### K751 - GSD Type IV - Date of test: 11.08.2017

Testresult: NORMAL

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#### K793 - Bloodtyping (DNA) - Date of test: 11.08.2017

Testresult: genotype N/N

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#### K762 - rdAc-PRA - Date of test: 14.08.2017

Testresult: NORMAL

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#### K711 - PKD test - Date of test: 14.08.2017

Testresult: pkd1/pkd1

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#### K799 - HCM3 - Date of test: 14.08.2017

Testresult: NORMAL

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#### K725 - HCM1 Test - Date of test: 14.08.2017

Testresult: NORMAL

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K767 - SMA - Date of test: 11.08.2017

Testresult: NORMAL

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K504 - Periodic paralysis (WNK4-Hypokalemia) - Date of test: 14.08.2017

Testresult: NORMAL

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K597 - Congenital Adrenal Hyperplasia - Date of test: 14.08.2017

Testresult: NORMAL

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K598 - Dihydropyrimidinase Deficiency - Date of test: 14.08.2017

Testresult: NORMAL

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K599 - Hyperlipoproteinaemia - Date of test: 14.08.2017

Testresult: NORMAL

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K600 - Niemann-Pick C1 Disease - Date of test: 14.08.2017

Testresult: NORMAL

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K601 - Primary Hyperoxaluria II - Date of test: 14.08.2017

Testresult: NORMAL

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K640 - Gangliosidosis, GM2, type II - 1 - Date of test: 11.08.2017

Testresult: NORMAL

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K641 - Vitamin D-deficiency rickets, type I - Date of test: 14.08.2017

Testresult: NORMAL

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K646 - Gangliosidosis, GM2, GM2A - Date of test: 14.08.2017

Testresult: NORMAL

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K647 - Gangliosidosis, GM2, type II - 2 - Date of test: 14.08.2017

Testresult: NORMAL

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K649 - Hypothyroidism - Date of test: 14.08.2017

Testresult: NORMAL

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K650 - Mucopolysaccharidosis VII - Date of test: 14.08.2017

Testresult: NORMAL

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K651 - Mucopolysaccharidosis VI - Date of test: 14.08.2017

Testresult: NORMAL

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K656 - Haemophilia B - 1 - Date of test: 14.08.2017

Testresult: NORMAL

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K657 - Haemophilia B - 2 - Date of test: 14.08.2017

Testresult: NORMAL

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K386 - Mucopolysaccharidosis I - Date of test: 14.08.2017

Testresult: NORMAL

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W.A. van Haeringen, PhD  
Executive Director

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#### K754 - Pyruvatekinase Def.

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K751 - GSD Type IV

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K793 - Bloodtyping (DNA)

Information about the Bloodtyping (DNA):

genotype b/b: The cat carries two copies of the recessive b allele. Serologically the cat shows bloodgroup B.

genotype N/b: The cat carries one copy of the recessive b allele. Serologically the cat shows bloodgroup A or AB. The cat will pass the mutation onto its offspring with a probability of 50%.

genotype N/N: The cat is a non-carrier of the recessive b allele. Serologically the cat shows bloodgroup A or AB.

This test is validated in all breeds, except Ragdolls and Turkish Angora.

In a few percent of the cases, results are inconclusive and status of the B blood group cannot be determined. This is caused by a genetic variation that was not described in the publication.

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#### K762 - rdAc-PRA

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K711 - PKD test

Information about the PKD test:

Based on the results three groups of animals can be detected:

pkd1/pkd1: The cat is NO CARRIER, and has two healthy copies from the gene.

PKD1/pkd1: The cat is AFFECTED, and has one healthy and one defect copy from the gene.

PKD1/PKD1: The cat is AFFECTED, and has two defect copies from the gene.

The PKD test detects the presence of a mutation in the ADPKD1 gene (C->A mutation in exon 29), which is suggested to be responsible for Polycystic Kidney Disease (PKD) in several breeds. PKD of other genesis, especially caused by other unknown mutations cannot be excluded by this test.

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### K799 - HCM3

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will also become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

Information about the HCM3 test

The HCM3 test is based on the detection of a mutation in the MYBPC3 gene, which is suggested to cause hypertrophic cardiomyopathy (HCM) in Ragdoll cats. In Ragdolls the mutation which is suggested to cause HCM is like in Maine Coons in the MYBPC3-gene but in a different domain. HCM of other genesis caused by other mutations cannot be excluded by this test.

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### K725 - HCM1 Test

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will also become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

Information about the HCM1 test

The HCM1 test detects the mutation in the MYBPC gene (G->C mutation in exon 3) which is suggested to be responsible for hypertrophic cardiomyopathy in several cat breeds. HCM of other genesis especially caused by other mutation or other unknown mutations cannot be excluded by this test.

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### K767 - SMA

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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### K504 - Periodic paralysis (WNK4-Hypokalemia)

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K597 - Congenital Adrenal Hyperplasia

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K599 - Hyperlipoproteinaemia

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K600 - Niemann-Pick C1 Disease

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K601 - Primary Hyperoxaluria II

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K640 - Gangliosidosis, GM2, type II - 1

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K641 - Vitamin D-deficiency rickets, type I

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill

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due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K646 - Gangliosidosis, GM2, GM2A

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K647 - Gangliosidosis, GM2, type II - 2

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K649 - Hypothyroidism

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K650 - Mucopolysaccharidosis VII

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K651 - Mucopolysaccharidosis VI

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

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**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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#### K656 - Haemophilia B - 1

Explanation about the result for females:

**NORMAL:** The animal is free and has two healthy alleles. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

Explanation about the result for males:

**NORMAL:** The animal is free and has one healthy allele and the sex chromosome Y. It cannot spread the disease in the population.

**AFFECTED:** The animal is affected and has one mutant (disease) allele and the sex chromosome Y. When used in breeding, all male offspring will receive the sex chromosome Y. All female offspring will receive the mutant (disease) allele.

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#### K657 - Haemophilia B - 2

Explanation about the result for females:

**NORMAL:** The animal is free and has two healthy alleles. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

Explanation about the result for males:

**NORMAL:** The animal is free and has one healthy allele and the sex chromosome Y. It cannot spread the disease in the population.

**AFFECTED:** The animal is affected and has one mutant (disease) allele and the sex chromosome Y. When used in breeding, all male offspring will receive the sex chromosome Y. All female offspring will receive the mutant (disease) allele.

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#### K386 - Mucopolysaccharidosis I

Explanation about the result:

**NORMAL:** The animal is free and has two healthy alleles. When used in breeding, this animal will not become ill due to the disease. It cannot spread the disease in the population.

**CARRIER:** The animal is carrier and has one healthy and one mutant (disease) allele. When used in breeding, 50 percent of the offspring will receive the disease allele. Carriers will not become ill.

**AFFECTED:** The animal is affected and has two mutant (disease) alleles. When used in breeding, all offspring will receive the mutant allele from this animal. Affected animals will become ill.

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