

# GENETIC ANALYSIS REPORT



**Breed Specific  
Medicine**

## OWNER'S DETAILS

**Katalin Bodo**

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## ANIMAL'S DETAILS

**Registered Name:** Moonlight Yenzo Yoda

**Pet Name:** Yoda

**Breed:** Birman

**Registration No:** 102316 02

**Microchip No:** 900118000083237

**Sex:** Male

## COLLECTION DETAILS

**Case Number:** 14-098775

**Collected By:** Katalin Bodo

**Date of Test:** 03/12/14

**Approved Coll. Mthd.:** Yes

*Sample with Lab ID Number 14-098775 was received at Orivet Genetics, DNA was extracted and analysed with the following results reported:*

**DISEASE(S):** PYRUVATE KINASE (PK) DEFICIENCY (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
MUCOPOLYSACCHARADOSIS (**INDETERMINABLE - RESULT OBTAINED IS INCONCLUSIVE**)  
POLYCYSTIC KIDNEY DISEASE (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
NEIMANN-PICK DISEASE TYPE C (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
FAMILIAL EPISODIC HYPOKALEMIC POLYMYOPATHY (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
HYPERTROPHIC CARDIOMYOPATHY - MAINE COON (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
HYPERTROPHIC CARDIOMYOPATHY - RAGDOLL (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
PROGRESSIVE RETINAL ATROPHY (PRA) CEP 290 (PRA-RDC) (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
PROGRESSIVE RETINAL ATROPHY (PRA) CRX (PRA-RDY) (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
SPINAL MUSCULAR ATROPHY (SMA) - MAINE COON (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
GLYCOGEN STORAGE DISEASE TYPE IV (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
GANGLIOSIDOSI- GM2 (**NORMAL / CLEAR - NO MUTATION DETECTED**)



**RESULTS REVIEWED AND CONFIRMED BY:**

  
Dr. Noam Pik BVs MDSV

  
George Sofronidis BSc (Hons)

## AN OVERVIEW OF GENETIC TESTING - GLOSSARY OF TERMS

The terms below are provided to help clarify certain items on your genetic reports. The genetic results/terms are those as reported by Orivet.

**NORMAL/CLEAR - NO MUTATION DETECTED** - No presence of the mutation (wild type) is detected. The animal is clear of disease, will not pass on any disease-causing mutation.

**CARRIER/ HETEROZYGOUS - ONE COPY DETECTED** - One copy of the normal gene (wild type) and affected (mutant) gene is present, will not exhibit disease symptoms or develop the disease. Consideration needs to be taken if breeding this animal- if breeding with another carrier or affected or unknown then it may produce an affected offspring.

**AFFECTED HETEROZYGOUS (ONE COPY)** - One copy of the normal gene (wild type) and affected (mutant) gene is present, yet due to the dominant mode of inheritance of the disease the animal may show symptoms (affected). Appropriate treatment should be pursued by consulting a veterinarian.

**AFFECTED/ POSITIVE - TWO COPIES** - Two copies of the disease gene (mutant) are present, the animal may show symptoms (affected) associated with the disease. Appropriate treatment should be pursued by consulting a veterinarian.

**NORMAL BY PARENTAGE HISTORY** - The sample submitted has had its parentage confirmed- by pedigree or DNA. By definition, this information together with the history submitted for the parents excludes this animal from having this disease. The controls run confirm that the dog is **NORMAL** for the disease requested.

**NO RESULTS AVAILABLE** - Insufficient information has been provided to provide a result for this test. Sire and Dam information and/or sample may be required. This result is mostly associated with tests that have a patent/license and therefore certain restrictions apply. Please contact the laboratory to discuss

**DNA PROFILE** - Also known as a DNA fingerprint is unique for the animal. No animal shares the same DNA profile. An individual's DNA profile is inherited from both parents and can be used for verifying parentage (pedigrees). The nomenclature **CSNP** identifies the single nucleotide polymorphism (SNP) at a particular site on the chromosome with each number representing a different site.

**FAIL** - The sample submitted has failed to give a conclusive result. Failures are due mainly to quality/quantity of DNA. We strongly advise that another sample be re-collected and submitted. To minimize bacterial contamination you should allow the swab to air dry (stand up) for at least 3 minutes prior to placing them back into the original swab packaging.

**PARENTAGE CONFIRMATION** - A separate parentage report is generated and emailed for any parentage request. Parentage confirmation report can only be generated if a DNA profile has been carried out for dam, offspring and possible offspring.

**PENDING** - Result for this test is still being processed. When completed, the result will be emailed. Certain tests are run on different chips which can lead to results being uploaded and completed separately.

**INDETERMINABLE** - The samples submitted has failed to give a conclusive result, this result may need to be determined via a manual process. If you have submitted a swab sample you may need to recollect and resubmit a blood sample to enable a conclusive result for the test.

**APPROVED COLLECTION METHOD (YES)** - the sample submitted for testing HAS met the requirements recommended by member bodies for the DNA collection process. The animal has been identified via its microchip number (Positive ID) and collected by a Veterinarian or Approved Collection Agent.

**APPROVED COLLECTION METHOD (BLANK)** - the sample submitted for testing HAS NOT met the requirements recommended by member bodies for the DNA collection process.

**TRAIT** - A feature that an animal is born with (genetically determined characteristic). Traits are visual phenotype that range from colour to hair length, also includes certain features such as tail length. If an individual is **AFFECTED** for a trait then it will show that characteristic eg. **AFFECTED** for the B (brown) LOCUS or bb will be brown/chocolate.

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**CLARIFICATION OF GENETIC TESTING** The goal of genetic testing is to provide breeders with relevant information to improve breeding practices in the interest of animal health. However, genetic inheritance is no simple process, and may be complicated by several factors. Below is some information to help clarify these factors.

**1)** Some diseases may demonstrate signs of what geneticists call "genetic heterogeneity". This is a term to describe an apparently single condition that may be caused by more than one mutation/and or gene.

**2)** It is possible that there exists more than one disease that presents in a similar fashion and segregates in a single breed. These conditions- although phenotypically similar- may be caused by separate mutations/ and or genes.

**3)** It is possible that the disease affecting your breed may be what geneticists call an "oligogenic disease". This is a term to describe the existence of additional genes that may modify the action of a dominant gene associated with a disease. These modifier genes may for example give rise to a variable age of onset for a particular condition, or affect the penetrance of a particular mutation such that some animals may never develop the condition.

The range of hereditary diseases continue to increase and we see some of that are relatively benign and others that can cause severe and/or fatal disease. Diagnosis of any disease should be based on pedigree history, clinical signs, his (incidence) of the disease and the specific genetic test for the disease. Penetrance of a disease will always vary not only from breed to breed but within a breed, and will vary with different diseases. Factors that influence penetrance are genetics, nutrition and environment. Although genetic testing should be a priority for breeders we strongly recommend the temperant and phenotype also be considered when breeding.

**Orivet Genetic Pet Care aims to frequently update breeders with the latest research from the scientific literature. If breeders have any questions regarding a particular condition, please contact us on (03) 9534 1544 and we will be happy to work with you to answer any relevant questions.**

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