

# GENETIC ANALYSIS REPORT



## OWNER'S DETAILS

**Gerhard Bangen**  
Flottbeker Stieg 21  
Hamburg, 22607

Add: P.O. Box 110  
St Kilda 3182 VIC

Ph: +61 3 9534 1544  
Fax: +61 3 9525 3550

email: info@orivet.com.au  
website: www.orivet.com.au

A.B.N. 8 722 516 58 99

## ANIMAL'S DETAILS

**Registered Name:** Zimbert Von Flottbek  
**Pet Name:** Zimbert  
**Breed:** Bengalese

**Registration No:** L0382307  
**Microchip No:** 945000005020259  
**Sex:** Male

## COLLECTION DETAILS

**Case Number:** 15-068993  
**Collected By:** Gerhard Bangen

**Date of Test:** 29/04/15  
**Approved Coll. Mthd.:**

Sample with Lab ID Number 15-068993 was received at Orivet Genetics, DNA was extracted and analysed with the following results reported:

DNA PROFILE The DNA Profile below represents the genetic identification of Zimbert Von Flottbek

SNP01	SNP02	SNP03	SNP04	SNP05	SNP06	SNP07	SNP08	SNP09	SNP10	SNP11
	CC	GT	AA	CC	CC	CT	TT	CT	GG	AA
SNP12	SNP13	SNP14	SNP15	SNP16	SNP17	SNP18	SNP19	SNP20	SNP21	SNP22
CC	AA	GG	CT	AA	GA	GG	GG	CC	CC	AA
SNP23	SNP24	SNP25	SNP26	SNP27	SNP28	SNP29	SNP30	SNP31	SNP32	SNP33
GA	AA	CT	GG		AA	TT		GA	GG	
SNP34	SNP35	SNP36	SNP37	SNP38	SNP39	SNP40	SNP41	SNP42	SNP43	SNP44
	GG	GA	CT	CC	CC	GG	GA	TT	CT	GG
SNP45	SNP46	SNP47	SNP48	SNP49	SNP50	SNP51	SNP52	SNP53	SNP54	SNP55
	CT	TT	GA		GT	GG	GA	GG	CC	TT
SNP56	SNP57	SNP58	SNP59	SNP60	SNP61	SNP62	SNP63	SNP64	SNP65	SNP66
AG	CT	CT	CC	GG	CC	CC	CT	GG	GG	GG
SNP67	SNP68	SNP69	SNP70	SNP71	SNP72	SNP73	SNP74	SNP75	SNP76	SNP77
CT	CT	GG	AA	CC	AA	GG	GA	AA	GT	AA
SNP78	SNP79	SNP80	SNP81	SNP82	SNP83	SNP84	SNP85	SNP86	SNP87	SNP88
TT	GG	AG	CT	TT	CC	CC		CT	AA	TC



RESULTS REVIEWED AND CONFIRMED BY:

Dr. Noam Pik BVs MDSV

George Sofronidis BSc (Hons)

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**DISEASE(S):** PYRUVATE KINASE (PK) DEFICIENCY (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
MUCOPOLYSACCHARADOSIS (**NORMAL / CLEAR - NO MUTATION DETECTED**)  
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) (**INDETERMINABLE - RESULT OBTAINED IS INCONCLUSIVE**)  
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**)

**TRAIT(S):** ALBINISM (**ALB N / ALB N - NO ALBINO MUTATION DETECTED**)  
AMBER (**E/E - NO COPIES OF AMBER MUTATION DETECTED**)  
DILUTION (**D/D FULL COLOUR - DOES NOT HAVE DILUTE ALLELE**)  
BLOOD GROUP (**N/N = TYPE A or AB (non-b/non-b) CAN BE A/A A/AB or AB/AB**)  
CHOCOLATE AND CINAMMON (**B/B (FULL COLOUR - CAT DOES NOT CARRY BROWN OR CINNAMON)**)  
COLOURPOINT RESTRICTION (**c<sup>s</sup>/c<sup>s</sup> - SIAMESE**)  
AGOUTI (**A/a (OFFSPRING CAN BE AGOUTI / NON-AGOUTI DEPENDS ON MATING)**)  
LONG HAIR SHORHAIR (**N/N = NONE OF THE 4 LONG HAIR MUTATIONS DETECTED**)  
WHITE GLOVES (**N<sup>9</sup>/N<sup>9</sup> - NO GLOVE MUTATION DETECTED**)



15-068993

**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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Orivet  
Genetic Pet Care

**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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