# GENETIC ANALYSIS REPORT

Orivet Genetic Pet Care
Breed Specific Medicine

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A.B.N. 8 722 516 58 99

## OWNER'S DETAILS

**Luke Erb**
5121 Perth Line 89
Gowanstown, On N0G 1Y0

## ANIMAL'S DETAILS

**Registered Name:** Arrowhead Noir Jekia
**Pet Name:** Jekia
**Breed:** Australian Cobberdog
**Date of Birth / Age:**

**Registration No:** 7589
**Microchip No:** 952000000978546
**Sex:** Female
**Colour:** Not Supplied By Client

## COLLECTION DETAILS

**Case Number:** 16-072765
**Collected By:** Luke Erb

**Date of Test:** 02/09/16
**Approved Coll. Mthd.:**

***Sample with Lab ID Number 16-072765 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 Arrowhead Noir Jekia

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

16-072765

**RESULTS REVIEWED AND CONFIRMED BY:**

Dr. Noam Pik BVs MDSV

George Sofronidis BSc (Hons)

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| **Collected By:** | Luke Erb | **Approved Coll. Mthd.:** | |

***Sample with Lab ID Number 16-072765 was received at Orivet Genetics, DNA was extracted and analysed with the following results reported:***

**DISEASE(S):**

EXERCISE INDUCED COLLAPSE **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
DEGENERATIVE MYELOPATHY **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
AUTOSOMAL HEREDITARY RECESSIVE NEPHROPATHY **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CENTRONUCLEAR MYOPATHY **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PROGRESSIVE RETINAL ATROPHY - CORD1/RCD-4 **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CYSTINURIA **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
GLOBOID CELL LEUKODYSTROPHY/KRABBE'S DISEASE **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MUCOPOLYSACCHARIDOSIS **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MYOTUBULAR MYOPATHY X LINKED **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
NARCOLEPSY **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
NEONATAL ENCEPHALOPATHY **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PHOSPHOFRUCTOKINASE (PFK) DEFICIENCY **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PYRUVATE KINASE (PK) DEFICIENCY **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PROGRESSIVE ROD CONE DEGENERATION - PRA **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
VON WILLEBRAND'S DISEASE TYPE I **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
GANGLIOSIDOSIS - TYPE 1 & 2 **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MALIGNANT HYPERTHERMIA **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
HEREDITARY NASAL PARAKERATOSIS (DRY NOSE) **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
SKELETAL DYSPLASIA 2 (COL11A2) **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
ELLIPTOCYTOSIS (B-SPECTRIN) **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**

**TRAIT(S):**

LONG HAIR GENE (PHENOTYPE) **- POSITIVE - SHOWING THE PHENOTYPE**
A-LOCUS AGOUTI **- at/at - TAN POINTS-TAN POINTS MAY BE BRINDLED (SEE K LOCUS)**
DILUTE MLPH GENE (BLUE/GREY) **- DD - NO COPY OF MLPH-D ALLELE**
K-LOCUS (DOM BLACK/WILD TYPE) **- KK - DOMINANT BLACK WILL NOT BE BRINDLED or EXPRESS AGOUTI**
EM-LOCUS MELANISTIC MASK ALLELE **- $E^m$ E - NO MASKING (Em) EXTENSION ALLELE**
E (EXTENSION) LOCUS MC1R **- ee - YELLOW, GOLDEN, CREAM, WHITE or APRICOT**
SPOTTING LOCUS (W GENE) **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
FOLLICULAR DYSPLASIA - COLOUR DILUTION ALOPECIA **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
BLACK HAIR FOLLICULAR DYSPLASIA **- NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
BROWN (GLNT331STOP) STOP CODON **($b^s$) - BB - DOES NOT CARRY BROWN or CHOCOLATE**
BROWN (345DELPRO) DELETION **($b^d$) - BB - DOES NOT CARRY BROWN or CHOCOLATE**
BROWN (SER41CYS) INSERTION CODON **($b^c$) - BB - DOES NOT CARRY BROWN or CHOCOLATE**

16-072765

**RESULTS REVIEWED AND CONFIRMED BY:**

Dr. Noam Pik BVs MDSV George Sofronidis BSc (Hons)

*The terms below are provided to help clarify certain results phrases on your genetic report. The phrases below are those as reported by Orivet and may vary from one laboratory to the other.*

**NORMAL/CLEAR/NEGATIVE - NO VARIANT DETECTED**

No presence of the variant (mutation) has been detected. The animal is clear of the disease and will not pass on any disease-causing mutation.

**CARRIER - ONE COPY OF THE VARIANT DETECTED**

This is also referred to as HETEROZYGOUS. One copy of the normal gene and copy of the affected (mutant) gene has been detected. The animal 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/POSITIVE FOR THE VARIANT**

Two copies of the disease gene variant (mutation) have been detected also referred to as HOMOZYGOUS for the variant. The animal may show symptoms (affected) associated with the disease. Appropriate treatment should be pursued by consulting a Veterinarian.

**AFFECTED – HETEROZYGOUS ONE COPY (AUTOSOMAL DOM)**

Also referred to as POSITIVE ONE COPY or POSITIVE HETEROZYGOUS. This result is associated with a disease that has a dominant mode of inheritance. One copy of the normal gene (wild type) and affected (mutant) gene is present. Appropriate treatment should be pursued by consulting a Veterinarian. This result can still be used to produce a clear offspring.

**AFFECTED – HOMOZYGOUS TWO COPIES (AUTOSOMAL DOM)**

Also referred to as POSITIVE HOMOZYGOUS. Two copies of the disease gene variant (mutant) have been detected and the animal may show symptoms associated with the disease. Please Note: This disease has dominant mode of inheritance so if mated to a clear animal ALL offspring with be AFFECTED – HETEROZYGOUS ONE COPY.

**NORMAL BY PARENTAGE HISTORY**

The sample submitted has had its parentage verified by DNA. By interrogating the DNA profiles of the Dam, Sire and Offspring 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.

**NORMAL BY PEDIGREE**

The sample submitted has had its parentage verified by Pedigree. The pedigree has been provided and details (genetic testing reports) of the parents have been included. Parentage could not be determined via DNA profile as no sample was submitted.

**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. This 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). This profile contains no disease or trait information and is simply a unique DNA signature for that animal.

**INDETERMINABLE**

The sample submitted has failed to give a conclusive result. This result is mainly due to the sample failing to "cluster" or result in the current grouping. This will be repeated and looked at manually; if a result cannot be determined, a recollection may be requested.

**PARENTAGE VERIFICATION**

**QUALIFIES/CONFIRMED or DOES NOT QUALIFY/EXCLUDED**

Parentage is determined by examining the markers on the DNA profile. A result is generated and stated for all DNA parentage requests. Parentage confirmation reports can only be generated if a DNA profile has been carried out for Dam, Offspring and possible Sire/s.

**PENDING**

Results for this test are still being processed. Some tests are run independently and are reported at a later date. When completed, the result will be emailed.

**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 (NO)**

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 (a genetically determined characteristic). Traits are a visual phenotype that range from colour to hair length, and 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.

**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 not a 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 continues to increase and we see some 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, history (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 that temperament 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.