Orivet
Genetic Pet Care

Case Study

Orivet Genetic Pet Care DNA Screen Detects Hypertrophic Cardiomyopathy in a Mixed-Breed Cat

Patient Condition and History

Misty was adopted from the animal rescue at the age of 10 weeks. Her parents were told she was a Ragdoll, but no pedigree certificates were included. They really wanted to find out more about her genetic makeup and any health risks. They decided to order the Orivet Genetic Risk Analysis and a Basic DNA Disease Screen.

Misty's Basic Genetic Disease DNA Screen (attached) revealed she was Affected (carrying one copy of the dominant mutation) for Hypertrophic Cardiomyopathy.

Based on her DNA test, breed, age, weight and sex, a Genetic Risk Analysis was performed (attached) which also flagged an increased risk for Cardiomyopathy as well as several other conditions. A Lifetime wellness plan was put in place to be on the look out for these.

At one year of age, and as part of Misty's wellness plan, her parents consented to going ahead with the recommended Echocardiography (ultrasound) of her heart exam. This study confirmed normal heart function (contractility and fractional ejection were within normal limits), however measurements detected a slight increase in the thickness of her heart wall.

Misty's parents had discussed managing her condition with her Veterinarian and a specialist Veterinary Cardiologist. They have instigated specific preventative plan to continue monitoring her risk allowing for early detection of any clinical deterioration, which in turn will lead to more effective management of her condition. Her parents were trained to look out for specific signs that may help early detection.

GENETIC ANALYSIS REPORT

Orivet Genetic Pet Care
Breed Specific Medicine

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OWNER'S DETAILS

Mellissa Price
41 Octavia Street
St Kilda, VI 3182

ANIMAL'S DETAILS

Registered Name: Price (3452RT)
Pet Name: Misty
Breed: Ragdoll
Date of Birth / Age: 21/12/15

Registration No:
Microchip No: 94560000002341
Sex: Female
Colour: Tortoise Shell

COLLECTION DETAILS

Case Number: 16-136070
Collected by: Dr Noam Pik

Date of Test: 29/02/16
Approved Coll. Mthd.: Yes

Sample with Lab ID Number 16-136070 was received at Orivet Genetics, DNA was extracted and analysed with the following results reported:

DISEASE(S):
PYRUVATE KINASE (PK) DEFICIENCY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
POLYCYSTIC KIDNEY DISEASE - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
NEIMANN-PICK DISEASE TYPE C - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
FAMILIAL EPISODIC HYPOKALEAMIC POLYMYOPATHY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
HYPERTROPHIC CARDIOMYOPATHY - MAINE COON - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
HYPERTROPHIC CARDIOMYOPATHY - RAGDOLL - **AFFECTED - HETEROZYGOUS ONE COPY (AUTOSOMAL DOM)**
PROGRESSIVE RETINAL ATROPHY (PRA-RDC) CEP 290 - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PROGRESSIVE RETINAL ATROPHY (PRA-RDY) CRX - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
SPINAL MUSCULAR ATROPHY (SMA) - MAINE COON - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
GLYCOGEN STORAGE DISEASE TYPE IV - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MUCOPOLYSACCHARIDOSIS TYPE I (DSH) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MUCOPOLYSACCHARIDOSIS TYPE VI (SIAMESE) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MUCOPOLYSACCHARIDOSIS TYPE VII (DSH) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MYOTONIA CONGENITA (CLCN1) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
LIPOPROTEIN DEFICIENCY (LPL) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
HYPEROXALURIA (GRPHPR) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
ACUTE INTERMITENT PORPHYRIA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MYOPATHY (DEVON REX COLQ) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
NEURODEGENERATIVE LYSOSOMAL STORAGE - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
BURMESE GANGLIOSIDOSIS (HEXB) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CYSTINURIA (SLC3A1) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**

16-136070

RESULTS REVIEWED AND CONFIRMED BY:

Dr. Noam Pik BVs MDSV

George Sofronidis BSc (Hons)

GENETIC RISK ANALYSIS REPORT

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MISTY'S ALGORITHMIC RISK ANALYSIS OF COMMON HERITABLE DISEASES

MOST RELEVANT DISEASES	OTHER LESS COMMON CONDITIONS
1. Hypertrophic Cardiomyopathy (HCM)	1. Gingivitis
2. Urolithiasis	2. FIP (Feline Infectious Peritonitis)
3. Mucopolysaccharidosis VI	3. Possible Predisposition To Feline Pancreatitis.
4. Polycystic Kidney Disease	

As part of the Basic Genetic Disease Screen, Mitsy's DNA was tested for these diseases.

Discussion

Identifying genetic heritable conditions and analysing data for heritable disease predisposition has proven very useful for Misty. The HCM mutations appear to be dominant genes, but with variable penetrance. This means some cats can develop signs of severe disease at an earlier age while some may not develop signs of disease until late in life. Large scale studies estimate that the Ragdoll breed has a prevalence of HCM of approximately 30%.Cardiomyopathy and is possibly with influenced by other (modifier) genes and environmental factors.

Conclusions

Using Orivet Genetic Pet Care screening and risk assessment tools was an inexpensive way to identify and manage the future health risks for Misty. Although there is no cure for HCM, both her parents and her Veterinarians have the comfort of knowing they are doing everything possible to give Misty the best chance to live a long and healthy life.

A lifetime plan was implemented to manage risk and ensure prevention, early diagnosis and more effective management of Misty's specific health needs.

Dr. Noam Pik BVSc, BVMS, MBA, IVAS, MACVS (Emergency and Critical Care) and Orivet's Chief Veterinarian, noted, "Every pet is a truly unique individual. Orivet provide valuable knowledge that allows tailoring of the wellness health plan, improving medical outcomes and saving owners significant costs in the long run".