Orivet Genetic Pet Care

Case Study

Orivet Genetic Pet Care DNA Screen Leads to Early Diagnosis of Polycystic Kidney Disease in a Domestic Short Hair Cat

Patient Condition and History

Tom was adopted from the local shelter at the age of 8 weeks. His parents were told he was a Domestic Shorthair (DSH). They really wanted to find out more about his genetic makeup and any health risks he may have. They decided to order the Orivet Genetic Risk Analysis and a Basic DNA Diseases Screen.

Tom's Basic Genetic Diseases DNA Screen (attached) revealed he was Affected (carrying one copy of the dominant mutation) for Polycystic Kidney Disease (PKD).

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

At 10 months of age of age, and as part of Tom's wellness plan, his parents consented to going ahead with the recommended sonographic (ultrasound) examination of his kidneys and abdomen. Unfortunately, this study revealed very small cysts present on Tom's kidneys and also the liver, confirming PKD diagnosis.

Tom's parents had discussed managing his condition with his Veterinarian and a Veterinary Internal Medicine Specialist. They have instigated a specific preventative plan and continue monitoring disease progression and early detection of any clinical signs of developing renal failure. This in turn will lead to more effective management of his condition including dietary modification and the use of medication. His parents were trained to look out for specific signs that may help early detection.

GENETIC ANALYSIS REPORT

Orivet Genetic Pet Care
Breed Specific Medicine

address P.O. Box 110
St Kilda 3182 VIC
phone +61 3 9534 1544
fax +61 3 9525 3550
email info@orivet.com.au
website www.orivet.com.au
A.B.N. 87225165899

OWNER'S DETAILS

Annie Faye A. Singson
123 Suares Drive
Davao City

ANIMAL'S DETAILS

Registered Name:	Singson (PH45637)	**Registration No:**	
Pet Name:	Tom	**Microchip No:**	9567890000023
Breed:	Ragdoll	**Sex:**	Male
Date of Birth / Age:	01/03/15	**Colour:**	Tabby Point

COLLECTION DETAILS

Case Number:	16-136172	**Date of Test:**	29/02/16
Collected by:	Dr Noam Pik	**Approved Coll. Mthd.:**	Yes

Sample with Lab ID Number 16-136172 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 - **AFFECTED - HETEROZYGOUS ONE COPY (AUTOSOMAL DOM)**
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 – **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
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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TOM'S ALGORITHMIC RISK ANALYSIS OF COMMON HERITABLE DISEASES

MOST RELEVANT DISEASES	OTHER LESS COMMON CONDITIONS
1. Chronic Kidney Disease *	1. Upper Respiratory Tract Infection ("Cat Flu")
2. Feline Lower Urinary Tract Disease (FLUTD)	2. Lymphoma
3. Hyperthyroidism	3. Megacolon
4. Chronic Bronchial Disease (Feline Asthma)	4. Eosinophilic Granuloma Complex
5. Osteoarthritis	5. Anxiety And Behaviour Related Disorders
6. Hypertrophic Cardiomyopathy (HCM)	6. Feline Infectious Peritonitis (FIP)
7. Feline Diabetes Mellitus	7. Squamous Cell Carcinoma (SCC)
8. Inherited Deafness	8. Feline Acne
	9. Geriatric Dementia
	10. Food Intolerance And Allergies
	11. Hepatic Lipidosis
	12. Pancreatitis And Triaditis
	13. Pleural Effusion
	14. Atopy
	15. Feline Immunodeficiency Virus (FIV)
	16. Feline Leukaemia Virus (FeLV)

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

*** Tested for the PKD renal mutation only.**

Discussion

Identifying genetic heritable conditions and analysing data for heritable disease predisposition has proven very useful for Tom.

PKD is thought to be the most prevalent inherited disease in cats. PKD is transmitted as an autosomal dominant trait. This means that an affected cat inherits the disease from only one affected parent. There are no "carriers" as such; however the disease has variable expression, meaning some cats can be severely affected, while others may have very mild or occasionally even no clinical signs of disease.

Most cats develop kidney failure between 3 - 10 years of age, with the average age being 7 years. Severely affected cats may develop kidney failure earlier than this, while some cats may have small cysts in their kidneys that never become large enough to cause renal failure.

Ultrasound screening of kidneys is recommended at 10 months of age for maximum sensitivity. This examination can document the size of renal cysts, and repeat scanning can also document disease progression. Occasionally cats may have cysts in other locations, such as the liver, as well as the kidneys.

Conclusions

Using Orivet Genetic Pet Care screening and risk assessment tools was an inexpensive way to identify and manage the future health risks for Tom. Although there is no cure for PKD, early diagnosis and treatment can prolong survival and significantly improve quality of life. Both his parents and her Veterinarians have the comfort of knowing they are doing everything possible to give Tom 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 Tom'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".