Orivet Genetic Pet Care

Case Study

Orivet Genetic Pet Care DNA Screen Detects Cardiomyopathy in a Boxer-Cross

Patient Condition and History

Butch was adopted from the local shelter at the age of 8 months. His parents were told he was a Boxer cross, but really wanted to find out more about his genetic makeup and any health risks. They decided to order the Mixed Breed Identification DNA test along with the Genetic Risk Analysis and a Basic DNA Diseases Screen.

Butch's Basic Genetic Disease DNA Screen (attached) revealed that he was Affected (carrying one copy of the dominant mutation) for Arrhythmogenic Right Ventricular Cardiomyopathy, also knows as Boxer Cardiomyopathy.

The Breed Identification DNA test revealed that Butch's breed makeup was indeed Boxer (50%) with the English Mastiff making the other 50%. Based on the breeds detected, his 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 Butch's wellness plan, his parents consented to going ahead with the recommended five-minute ECG exam. Unfortunately this examination detected several Ventricular Premature (heart) Contractions (VPCs). After a short discussion with the Butch's Veterinarian, it was agreed to also perform an ultrasound Echocardiography of his heart. Butch's heart muscle appeared to be only slightly affected with very early disease changes being detectable. He remained clinically normal at this stage.

Butch's parents had discussed managing this condition with his Veterinarian and a specialist Veterinary Cardiologist. They have instigated specific preventative measures to avoid severe arrhythmias (heartbeat rhythm disturbances), which puts Butch at high risk of syncope (fainting) and sudden death. These included dietary supplementation with L-Carnitine and Omega-3 fatty acid in addition to antiarrthymic medications. He is going to be monitored more closely in the coming years.

GENETIC ANALYSIS REPORT

Orivet Genetic Pet Care
Breed Specific Medicine

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OWNER'S DETAILS

Ron Newland
6 Border Court
Lockyer Waters, QL 4311

ANIMAL'S DETAILS

Registered Name:	Newland (QL4567P)	**Registration No:**	
Pet Name:	Butch	**Microchip No:**	91800000345678
Breed:	Boxer mix	**Sex:**	Male
Date of Birth / Age:	03/09/14	**Colour:**	Tri-Colour

COLLECTION DETAILS

Case Number:	16-135866	**Date of Test:**	25/02/16
Collected by:	Dr Noam Pik	**Approved Coll. Mthd.:**	Yes

Sample with Lab ID Number 16-135866 was received at Orivet Genetics, DNA was extracted and analysed with the following results reported:

DISEASE(S):

PRIMARY LENS LUXATION - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
EXERCISE INDUCED COLLAPSE - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
COLLIE EYE ANOMALY/CHOROIDAL HYPOPLASIA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
DEGENERATIVE MYELOPATHY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
AUTOSOMAL HEREDITARY RECESSIVE NEPHROPATHY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
ARRYTHMOGENIC RIGHT VENTRICULAR CARDIO - **AFFECTED - HETEROZYGOUS ONE COPY (AUTOSOMAL DOM)**
CONE DEGENERATION - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CANINE HYPERURICOSURIA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CANINE MULTIFOCAL RETINOPATHY 1 - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CONE-ROD DYSTROPHY 1 - PRA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CYSTINURIA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
FACTOR VII DEFICIENCY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
HEREDITARY CATARACT - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
IVERMECTIN SENSITIVITY MDR1 - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
L2 HYDROXYGLUTARIC ACIDURIA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CEROID LIPOFUSCINOSIS AMERICAN BULLDOG - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
NCL- BORDER COLLIE - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
NCL- ENGLISH SETTER - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
NEONATAL ENCEPHALOPATHY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
POLYNEUROPATHY/NEUROPATHY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PROGRESSIVE RETINAL ATROPHY-RCD1A - **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)**
VON WILLEBRAND'S DISEASE TYPE III - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
DILATED CARDIOMYOPATHY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
TRAPPED NEUTROPHIL SYNDROME (TNS) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
DRY EYE CURLY COAT SYNDROME - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PROGRESSIVE RETINAL ATROPHY - RCD4 - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
COBALAMIN MALABSORPTION CUBILIN DEFICIENCY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
GENERALISED PRA 1 AND 2 - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PROGRESSIVE RETINAL ATROPHY - LATE ONSET - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
TYPE A PRA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
HEREDITARY NASAL PARAKERATOSIS (DRY NOSE) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
HAEMOPHILIA A (FACTOR VIII) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
ELLIPTOCYTOSIS (B-SPECTRIN) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**

RESULTS REVIEWED AND CONFIRMED BY:

Dr. Noam Pik BVs MDSV

George Sofronidis BSc (Hons)

GENETIC RISK ANALYSIS REPORT

Orivet Genetic Pet Care
Breed Specific Medicine

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BUTCH'S ALGORITHMIC RISK ANALYSIS OF COMMON HERITABLE DISEASES

10 MOST RELEVANT DISEASES	OTHER LESS COMMON CONDITIONS
1. Cancer	1. Progressive Retinal Atrophy, Generalised
2. Hip Dysplasia.	2. Elbow Dysplasia
3. Hypothyroidism	3. Distichiasis (Extra Eyelashes)
4. Osteochondrosis	4. Entropion
5. Cardiomyopathy	5. Ectropion
6. Exposure Keratopathy (Corneal Ulcers)	6. Hyperadrenocorticism (Cushing's Disease)
7. Gastric Dilatation & Volvulus (or "Bloat")	7. Pulmonic Stenosis
8. Skin fold dermatitis	8. Inherited Deafness
9. Intervertebral Disc Disease (IVDD)	9. Von Willebrand's Disease (type I)
10. Epilepsy (Idiopathic, Primary)	10. Subaortic Stenosis
	11. Cutaneous Asthenia (Ehlers-Danlos Syndrome)

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

Discussion

Identifying genetic heritable conditions and analysing data for heritable disease predisposition has proven very useful for Butch. Boxer Cardiomyopathy is an autosomal dominant disease with variable expression and is possibly influenced by other (modifier) genes and environmental factors. The variability in expression of the mutation means that some dogs may develop disease earlier than others – some dogs with the mutation may not develop serious clinical Cardiomyopathy at all. Unfortunately, there is a high incidence of severe arrhythmias with this form of Cardiomyopathy, and this may lead to episodes of fainting, exercise intolerance, or sometimes sudden death.

Studies have shown low levels of the amino acid L-carnitine in the heart muscle of some affected Boxers, and it is postulated that there may be an inherited defect leading to cardiac L-carnitine deficiency. Some dogs may respond to dietary supplementation. Omega-3 fatty acid supplementation has also been reported to aid in reducing the severity of arrhythmias, in addition to antiarrhythmic medications.

Conclusions

Using Orivet Genetic Pet Care screening and risk assessment tools was an inexpensive way to identify and manage the future health risks for Butch. Both his parents and his Veterinarians have the comfort of knowing they are doing everything possible to give Butch 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 Butch'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 provides valuable knowledge that allows tailoring of the wellness health plan, improving medical outcomes and saving owners significant costs in the long run".