Case Study

Orivet Genetic Pet Care Helps Early Diagnosis & Better Management of KCS in a Cavalier King Charles Cross

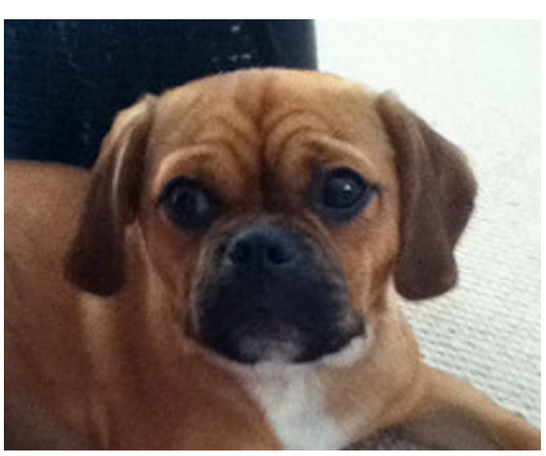
Patient Condition and History

Oscar, a four-year-old male mixed-breed dog underwent Breed Identification and Genetic Risk Analysis DNA testing to help identify and plan for any future health concerns he may have.

The Breed Identification DNA test revealed that Oscar's breed makeup was Pug (25%) and Cavalier King Charles Spaniel (75%) Breeds. Based on the breeds detected, his age weight and sex, his Genetic Risk Analysis (attached) revealed an increased risk of Kerato-Conjunctivitis Sicca (KCS) or Dry Eye as well as several other conditions. A wellness plan was put in place to be on the look out for these.

His Basic DNA Disease Screen screened for 37 genetic conditions and identified he was Affected for the KCS gene mutation in CKCSs and a Carrier for Degenerative Myelopathy.

The owner had commented that Oscar's eyes were often red, irritated and had a purulent discharge. As part of his new wellness plan, Oscar had a simple Schirmer tear test performed, revealing a significantly reduced tear production at 9 mm / minute (normal 25-42 mm/min). A diagnosis of KCS was made and medical treatment was initiated promptly. Oscar's eyes cleared up quickly and remained in perfect health to the date of this report.



Patient Genetic Risk Analysis (see copies below)

- Breed Identification DNA Test
- Orivet Genetic Health Analysis Risk Report
- Orivet Basic Mixed Breed Genetic Disease Screen

GENETIC ANALYSIS REPORT

Orivet Genetic Pet Care
Breed Specific Medicine

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A.B.N. 87225165899

OWNER'S DETAILS

George Sofronidis
Orivet Genetic Pet Care
St Kilda, VI 3182

ANIMAL'S DETAILS

Registered Name:	Sofronidis (P12345A)	**Registration No:**	
Pet Name:	Oscar	**Microchip No:**	956005670000123
Breed:	Cavalier Cross	**Sex:**	Male
Date of Birth / Age:	26/02/12	**Colour:**	Red & White

COLLECTION DETAILS

Case Number:	16-135764	**Date of Test:**	25/02/16
Collected by:	Orivet Research and Devel	**Approved Coll. Mthd.:**	Yes

Sample with Lab ID Number 16-135764 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 - **CARRIER (ONE COPY OF VARIANT DETECTED)**
AUTOSOMAL HEREDITARY RECESSIVE NEPHROPATHY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
ARRYTHMOGENIC RIGHT VENTRICULAR CARDIO - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
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 - **AFFECTED / POSITIVE FOR THE VARIANT**
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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OSCAR'S ALGORITHMIC RISK ANALYSIS OF COMMON HERITABLE DISEASES

10 MOST RELEVANT DISEASES	OTHER LESS COMMON CONDITIONS
1. Hip Dysplasia	1. Diabetes Mellitus
2. Brachycephalic Syndrome	2. Exocrine Pancreatic Insufficiency (EPI)
3. Progressive Retinal Atrophy, Generalised	3. Inherited Deafness
4. Exposure keratopathy (Corneal Ulcers)	4. Entropion
5. Luxating patella	5. Otitis Externa (Ear Infections)
6. Cataract (Cloudiness of the Eye Lens)	6. Urolithiasis
7. Skin fold dermatitis	7. Hemivertebra
8. Keratoconjunctivitis Sicca (KCS, Dry Eye)	8. Vitreous Degeneration
9. Retinal Dysplasia	9. Corneal Dystrophy
10. Distichiasis (Extra Eyelashes)	10. Syringomyelia and Chiari-like malformation
	11. Primary Secretory Otitis Media
	12. Demodectic Mange
	13. Organic Aciduria (L-2-HGA)
	14. Ichthyosis
	15. Hereditary Myopathy

Oscar's DNA was tested for these diseases.

Discussion

Identifying the breed makeup and analysing data for increased risk for heritable diseases has proven very useful for Oscar. Although Dry Eye is not a life-threatening condition, it is definitely an irritating disease that can lead to more serious infections and more severe eye disease. Treatment is effective.

The knowledge obtained in Oscar's report has helped communicate the risk to his owners and further discussions lead the Veterinarian to confirm the clinical diagnostic and initiate a therapeutic action. This in turn will help maintain Oscar's healthy eyes and prevent possible complications that could affect Oscar's quality of life.

Conclusions

Using Orivet Genetic Pet Care screening and risk assessment tools was an inexpensive way to identify and manage a potential health risks for Oscar. Both owner and Veterinarian have the comfort of knowing they have done everything possible to give Oscar 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 Oscar'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. We provide valuable knowledge that allows tailoring of the wellness health plan to the individual, improving medical outcomes and saving owners significant costs in the long run".