Orivet Genetic Pet Care

Case Study

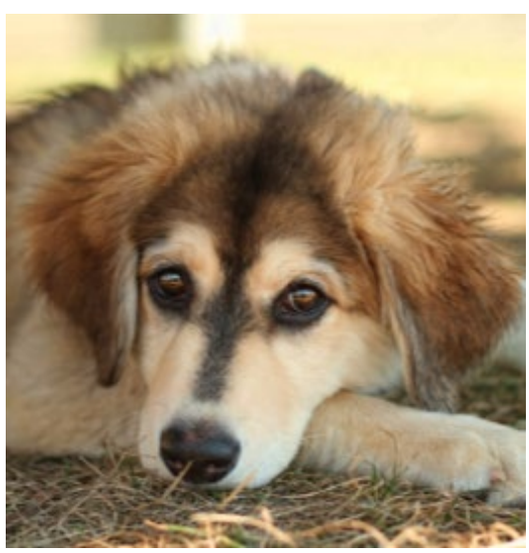
Orivet Genetic Pet Care Screen Saves Puppy From Bleeding in Surgery

Patient Condition and History

The owners of Jackie, a four-month-old female mixedbreed puppy decided to find out about the breeds present in Jackie and to screen her DNA for genetic diseases in order to understand any health risks she may have. The Breed Identification DNA test revealed the Golden Retriever (50%), Samoyed (25%) and Poodle (25%) Breeds as her main ancestral breeds. The Comprehensive Genetic Disease Screen (attached) revealed she was carrying two mutated copies of the von Willebrand's Type 1 disease gene which can lead to clotting problems. Her Genetic Risk Analysis (attached) revealed an increased risk to several other conditions and a wellness plan was put in place to be on the look out for these.his Veterinarian. They are considering surgical correction at a later stage, but in the meantime are supporting him by strict management of his weight and a modified nutritional plan with cautious use of anti-inflammatory medications as needed.

Four weeks later, and just prior to her de-sexing operation, the owner reported to the Veterinarian that they had noticed Jackie was bleeding from her mouth when a milk tooth fell out.

The Veterinarian, now aware of the von Willebrand's Disease genetic predisposition, decided to perform a buccal mucosal bleeding time test ahead of her de-sexing surgery, which was scheduled for the following month. This test revealed a severely prolonged clotting time, which would have put Jackie at a much higher risk during this routine surgery.



Jackie was given a special plasma transfusion rich with von Willebrand's clotting factor prior to surgery and great care was taken during surgery with double ligation of her uterine stumps. She made an uneventful recovery.

Patient Genetic Risk Analysis (see copies below)

- Breed Identification DNA Test
- Orivet comprehensive genetic disease screen for mixed breed dogs
- Orivet Genetic Health Analysis Risk Report

GENETIC ANALYSIS REPORT

Orivet Genetic Pet Care
Breed Specific Medicine

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OWNER'S DETAILS

Erlyn A. Aragones
P. O. BOX 110
Kilda, VI 3182

ANIMAL'S DETAILS

Registered Name:	Aragones - A38909	**Registration No:**	Pending
Pet Name:	Jackie	**Microchip No:**	345261679000000
Breed:	Mixed Breed Dog	**Sex:**	Female
Date of Birth / Age:	22/10/15	**Colour:**	Black Brindle

COLLECTION DETAILS

Case Number:	16-135899	**Date of Test:**	03/02/16
Collected by:	George Sofronidis	**Approved Coll. Mthd.:**	Yes

Sample with Lab ID Number 16-135899 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)**
PROGRESSIVE RETINAL ATROPHY-RCD3 - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
AUTOSOMAL HEREDITARY RECESSIVE NEPHROPATHY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED**
ARRYTHMOGENIC RIGHT VENTRICULAR CARDIO - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED**
CANINE HYPERURICOSURIA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CANINE LEUKOCYTE ADHESION DEFICIENCY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CANINE MULTIFOCAL RETINOPATHY 1 - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CENTRONUCLEAR MYOPATHY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CONGENITAL HYPOTHYROIDISM - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
COPPER TOXICOSIS - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CEREBELLA ATAXIA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CONE-ROD DYSTROPHY 1 - PRA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CYSTINURIA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
FUCOSIDOSIS - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
FACTOR VII DEFICIENCY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
GLOBOID CELL LEUKODYSTROPHY / KRABBE'S DISEASE - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
GM1 - GANGLIOSIDOSIS - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
HEREDITARY CATARACT (DOMINANT) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
HEREDITARY CATARACT (JUVENILE) - **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)**
MUSCULAR DYSTROPHY X-LINKED (MDX) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MUCOPOLYSACHARIDOSIS - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MYOTUBULAR MYOPATHY X LINKED - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
NARCOLEPSY - **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 - DACHSHUND - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
NCL- ENGLISH SETTER - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
NEONATAL ENCEPHALOPATHY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PYRUVATE DEHYDROGENASE PHOSPHATASE 1 DEF - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PHOSPHOFRUCTOKINASE (PFK) DEFICIENCY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PYRUVATE KINASE (PK) DEFICENCY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
POLYNEUROPATHY / NEUROPATHY (NDGR1) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PROGRESSIVE RETINAL ATROPHY-RCD1 - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PROGRESSIVE RETINAL ATROPHY-RCD1A - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
VON WILLEBRAND'S DISEASE TYPE I - **AFFECTED / POSITIVE FOR THE VARIANT**
VON WILLEBRAND'S DISEASE TYPE III - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
X LINKED PRA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MYOTONIA CONGENITA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
DILATED CARDIOMYOPATHY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
TRAPPED NEUTROPHIL SYNDROME (TNS) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
CATALASE DEFICIENCY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
DRY EYE CURLY COAT SYNDROME (TNS) - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
POMPES DISEASE - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
EPISODIC FALLING SYNDROME - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
RETINAL DYSPLASIA / OSD - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
COBALAMIN MALABSORPTION CUBILIN DEFICIENCY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
OSTEOGENESIS IMPERFECTA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
GENERALISED PRA 1 AND 2 - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
MALIGNANT HYPERTHERMIA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
HEREDITARY MULTIFOCAL RENAL CYSTADENOCARCINOMA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
PREKALLIKREIN DEFICIENCY - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**
TYPE A PRA - **NORMAL / CLEAR / NEGATIVE (NO VARIANT DETECTED)**

16-135899

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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JACKIE'S ALGORITHMIC RISK ANALYSIS OF COMMON HERITABLE DISEASES

10 MOST COMMON AND SIGNIFICANT DISEASE	OTHER LESS COMMON CONDITIONS
1. Cancer	1. Cardiomyopathy
2. Hip Dysplasia.	2. Elbow Dysplasia
3. Hypothyroidism	3. Distichiasis. (Extra Eyelashes)
4. Osteochondrosis	4. Keratoconjunctivitis Sicca (KCS, Dry Eye).
5. Progressive Retinal Atrophy, Generalised	5. Retinal Dysplasia
6. Pancreatitis.	6. Diabetes Mellitus.
7. Gastric Dilatation Volvulus (GDV, "Bloat")	7. Glaucoma
8. Luxating patella	8. Hyperadrenocorticism (Cushing's Disease)
9. Cataract (Cloudiness of the Eye Lens)	9. Entropion
10. Epilepsy (Idiopathic, Primary or Inherited)	10. Inherited Deafness
	11. Hypoadrenocorticism (Addison's Disease)
	12. Haemophilia A (Factor VIII Deficiency)
	13. Corneal Dystrophy
	14. Panosteitis
	15. Von Willebrand's Disease (type I)

Jackie's DNA was tested for these diseases.

Discussion

Identifying breed makeup, screening for genetic heritable conditions and analysing data for heritable disease predisposition has proven very useful for Jackie. von Willebrand's Disease is a known autosomal recessive clotting disorder of at least 2 of her ancestral parent breeds. The knowledge obtained has alerted Jackie's Veterinarian to an increased risk of bleeding (amongst others). Further clinical evidence has prompted her Veterinarian to act, confirming a clinical diagnosis. This in turn instigated precautionary measures to prevent a possible serious and expensive complications leading to a potentially devastating outcome from a routine surgical procedure.

Conclusions

Using Orivet Genetic Pet Care screening and risk assessment tools was an inexpensive way to identify and manage the future health risks for Jackie. Both owner and Veterinarian have the comfort of knowing they have done everything possible to give Jackie 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 Jackie'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".