Urban Animal Report

Simba, the Labrador got a Trace Kit Test from Urban Animal

Here's what his final report looks like!

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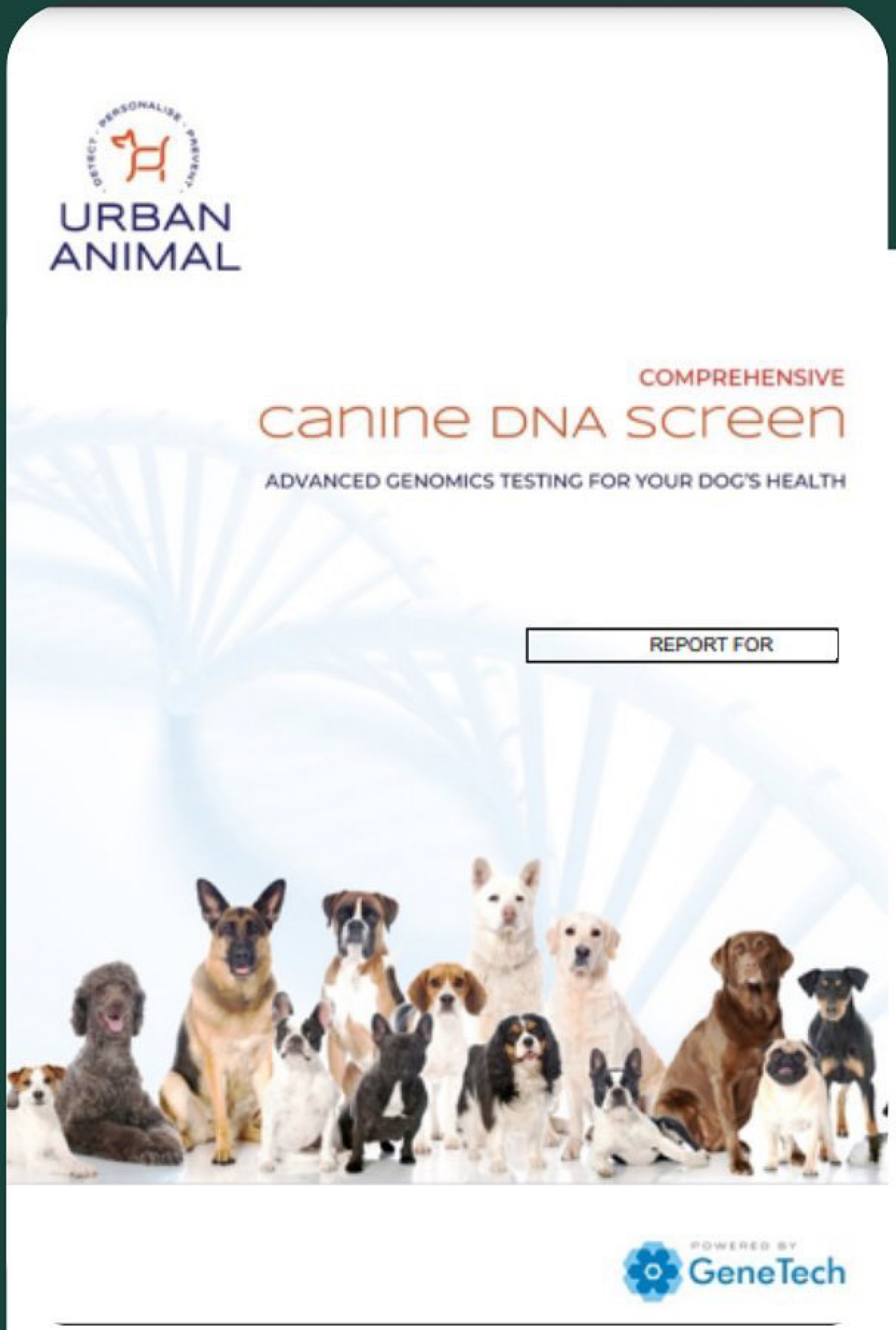

Overview of the Report

The Summary of Simba's results show 1 disorder, 3 carrier conditions and 4 traits which are explained in detail on each individual page

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Canine Traits and Disorders

Report for

Basic Details

Breed:	**Labrador**	Sex :		Age :	
Parents:		Weight:		Height:	
Known Health Conditions :		**None**			

Sample Details

Reference Id:	**IonCode_0791**	Collected:	**20/11/2022**	Sample Type:	**Swab**
Lab Ref Id:	**BSCTD22K7**	Received:	**22/11/2022**	Reported:	**20/12/2022**

Reference Details

Referring Vet:		Clinic's Name:	
Clinic Address:			
Owner name:		Email Address:	
Owner Address:			

Genomics Test Details

Panel:			
Laboratory:	**GeneTech**	Technology:	
Machine:		LibPrep Solution:	
Panel size:		Marker Coverage:	

SUMMARY OF RESULTS

Disorders	1
Carriers	3
Traits	4

DISORDERS IDENTIFIED
Menkes Disease

CARRIER STATUS IDENTIFIED
Achromatopsia-2
Cone Rod Dystrophy 3
Osteogenesis Imperfecta SERPINH1 related

TRAITS IDENTIFIED
Coat colour, dominant black
Coat color grizzle, coat color extension
Coat color Agouti
Coat color extension

Trait/ Disease details

For each disease/ trait identified, this individual page gives details about the condition, the symptoms, the onset period and also provides management strategies

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Neurological
Menkes Disease

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DISEASE CONDITION

Marker	**AGSCTD030**	GeneName	**ATPase, Cu transporting a-polypeptide**		Gene	**ATP7A**
Category	**Neurological**	Condition	**Menkes Disease**		Inheritance	**X-linked recessive**
Chr#:	**CHR X**	Genotype	Reference: **C**	Var Found: **T**	OMIA #	**000640**

About the Condition

Menkes disease, involving ATP7A, is a neurodegenerative disorder of copper deficiency. It is characterized by cerebral and cerebellar degeneration, connective tissue abnormalities, coarse hair and a failure to thrive. Symptoms are directly related to the dysfunction of copper dependent enzymes and there is a high variation in the severity of symptoms and if left untreated, may result in severe neurologic defects. The diagnosis is usually confirmed by analysing copper level in the urine.

Onset and Prognosis

This occurs at an early age and can be lethal if it is not treated. Phenotypic variability is reported.

Dog Breeds

Menkes disease is seen in Labrador Retriever, Doberman Pinscher, American Cocker Spaniel, Keeshond, Skye Terrier and West Highland White Terrier

Genetics and Inheritance

ATP7A gene is responsible for production of the ATPase enzyme that regulates copper levels in the body. The missense mutation causes decrease in copper levels and results to Menkes disease. The condition is inherited in an X-linked recessive pattern and affects only male dogs. Female dogs are unaffected carriers.

Management

1. Frequent copper analysis followed by early treatment can alleviate the neurological symptoms.
2. Avoid using affected dogs and unaffected carrier females in breeding programs

References

1.Wu, X., den Boer, E.R., Vos-Loohuis, M., Steenbeek, F.G.V., Monroe, G.R., Nijman, I.J., Leegwater, P.A.J., Fieten, H.: **Investigation of genetic modifiers of copper toxicosis in Labrador Retrievers**Life (Basel) 10:, 2020.

2.Pindar, S., Ramirez, C: **Predicting copper toxicosis: relationship between the ATP7A and ATP7B gene mutations and hepatic copper quantification in dogs**Hum Genet :, 2019.

3.Fieten, H., Gill, Y., Martin, A.J., Concilli, M., Dirksen, K., van Steenbeek, F.G., Spee, B., van den Ingh, T.S., Martens, E.C., Festa, P., Chesi, G., van de Sluis, B., Houwen, R.H., Watson, A.L., Aulchenko, Y.S., Hodgkinson, V.L., Zhu, S., Petris, M.J., Polishchuk, R.S., Leegwater, P.A., Rothuizen, J.: **The Menkes and Wilson disease genes counteract in copper toxicosis in Labrador retrievers: a new canine model for copper-metabolism disorders**Dis Model Mech 9:25-38, 2016.

Prevention

Now that we have found the genetic predispositions in advance, you can consult our expert vets and start taking the necessary steps to ensure a healthier future for Simba.

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