


Dilated cardiomyopathy (RBM20)

Client Name:	Angelique van Greuning (ANG003)	Report No:	ZO2022/4399/20220729/#29030
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Profile:	DG2022/34844	Species:	Canis lupus familiaris / Canine / Dog
Name:	Zia van Greuning	Microchip #:	953010002929661
Breed:	Dobermann		
Test:	[RBM20] Dilated cardiomyopathy (RBM20)		
Results:	c.2472_2493del	WT/WT	CLEAR

Sample Type: Whole Blood (EDTA)	Extraction Method: DNA Extraction: D4069	Test Type: Genetic Health
<p>[RBM20] Dilated cardiomyopathy (RBM20)</p> <p>Hereditary dilated cardiomyopathy (DCM) is a myocardial disease that can result in the development of congestive heart failure and sudden death. DCM is known to affect Doberman Pinscher, Dobermann, Great Dane, Boxer, Cocker Spaniel and Schnauzers.</p> <p>The presence and/or absence of 22bp deletion in RBM20 was tested.</p> <p>Mutant RBM20 is inherited in an autosomal recessive manner and requires two mutant alleles to present with symptoms.</p> <p>References: Wyles et al 2016. Modeling structural and functional deficiencies of RBM20 familial dilated cardiomyopathy using human induced pluripotent stem cells. Human Molecular Genetics 25(2): pp254-265</p>		

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The DNA profile is based on a preliminary marker panel that is subject to modification pending additional genetic information.

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