Paw Print Genetics
A division of Genetic Veterinary Sciences, Inc.

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Laboratory Report

Laboratory #:	464204	**Call Name:**	Spot
Order #:	209367	**Registered Name:**	-
Ordered By:	Stephen Stoltzfus	**Breed:**	Toy Poodle
Ordered:	Aug. 27, 2024	**Sex:**	Male
Received:	Sept. 3, 2024	**DOB:**	May 2020
Reported:	Sept. 25, 2024	**Registration #:**	-

Results:

Disease	Gene	Genotype	Interpretation
Chondrodystrophy with Intervertebral Disc Disease Risk Factor (CDDY with IVDD)	*CFA12 FGF4*	M/M	Increased IVDD Risk Associated with CDDY
Congenital Methemoglobinemia	*CYB5R3*	WT/WT	Normal (Clear)
Degenerative Myelopathy (Common Variant)	*SOD1*	WT/WT	Normal (Clear)
Ehlers-Danlos Syndrome (Poodle Type, Variants 1 and 2)	*TNXB*	WT/WT	Normal (Clear)
GM2 Gangliosidosis (Poodle Type)	*HEXB*	WT/WT	Normal (Clear)
Hereditary Cataracts	*HSF4*	WT/WT	Normal (Clear)
Multidrug Resistance 1	*ABCB1*	WT/WT	Normal (Clear)
Neonatal Encephalopathy with Seizures	*ATF2*	WT/WT	Normal (Clear)
Osteochondrodysplasia	*SLC13A1*	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration	*PRCD*	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Rod-Cone Dysplasia 4	*C2orf71*	WT/WT	Normal (Clear)
Von Willebrand Disease I	*VWF*	WT/WT	Normal (Clear)

WT, wild type (normal); M, mutant; Y, Y chromosome (male)

Interpretation:

Molecular genetic analysis was performed for 13 specific mutations reported to be associated with disease in dogs. We identified two normal copies of the DNA sequences in 11 mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 11 mutations.

Molecular genetic analysis was performed for a specific mutation reported to be associated with Chondrodystrophy with Intervertebral Disc Disease Risk Factor (CDDY with IVDD) in dogs. We identified two mutant copies of the DNA sequences for *CFA12 FGF4* associated with CDDY and IVDD risk. Thus, this dog is affected with CDDY and is at increased risk of developing an early-onset form of IVDD known as Hansen's Type I IVDD.

Recommendations:

Chondrodystrophy with an increased risk to develop early-onset Intervertebral Disc Disease (CDDY and IVDD Risk) is inherited in an autosomal dominant fashion. Dogs which inherit one or two copies of the associated *FGF4* mutation in chromosome 12 (*CFA12 FGF4*) display CDDY, which is defined as shortening of long bones in the limbs

and premature degeneration and calcification of intervertebral discs. Based on this and the fact that this dog has inherited two copies of this mutation, this dog is affected with CDDY and is at increased risk of developing an early-onset form of IVDD known as Hansen's Type I IVDD. However, IVDD associated with this mutation displays incomplete penetrance, meaning that even though dogs with the mutation display premature degradation of intervertebral discs, not every dog with this mutation will develop clinical signs of IVDD. Hansen's Type I IVDD is marked by herniation of intervertebral disc material compressing the spinal cord causing a variety of clinical signs like severe pain (secondary to acute inflammation and hemorrhage in the spinal canal), neurological dysfunction of variable severity and possible permanent paralysis. Dogs related to this dog have an increased risk to be affected by or carry the mutated gene. Additional testing for this mutation is indicated for related dogs.

Paw Print Genetics® has genetic counseling available to you at no additional charge to answer any questions about these test results, their implications and potential outcomes in breeding this dog.