Veterinary Animal Hospital

Swab code: 41201051408125
Swab activated on 1/16/2022
Results completed on 2/7/2022
Report accessed on 3/6/2022
Ordered by Dr. John Smith

vetsupport@embarkvet.com
1-855-203-8271

Patient Information

Oliver
0 yrs 6 mths - M
EMR 8675309

Joe Kelley
example@embarkvet.com
555-555-5555

Genetic Results Summary

Breed Results
64.0% Australian Shepherd
26.0% Golden Retriever
10.0% Chow Chow

Genetic Age: 8 human years
Predicted Adult Weight: 59 lbs

Increased Risks
1 increased risk

Notable Risks
1 notable risk

Clear Results
216 variants not detected

Increased Risk

MDR1 Drug Sensitivity

Oliver has two copies of this codominant variant in the ABCB1 gene. Dogs that inherit two abnormal copies (homozygous) will produce no normal p-glycoprotein and will be most strongly affected. Dogs that inherit only one abnormal copy of the ABCB1 gene (heterozygous) can show some effects though they will be less severely impacted because some normal p-glycoprotein will still be produced.

You can learn more about penetrance and care for Oliver below or email vetsupport@embarkvet.com should you desire to speak with a genetic counselor.

Notable Result

Ichthyosis, ICH1

Oliver has one copy of this variant in the PNPLA1 gene. Because this variant is inherited in an autosomal recessive manner (meaning dogs need two copies of the variant to develop the disease), Oliver is unlikely to develop Ichthyosis, ICH1 due to this variant.

While Oliver is not at risk for developing ICH1, he can pass this variant on to the next generation. If Oliver is intended for breeding, please genotype any potential mates. You can email vetsupport@embarkvet.com to discuss with a genetic counselor how the genotype results should be applied to a breeding program.

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Increased Risk

MDR1 Drug Sensitivity

Variant Info

ABCB1
Codominant inheritance
2 copies of the variant

How to interpret this result

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What is MDR1 Drug Sensitivity?

P-glycoprotein (P-gp), encoded by the ABCB1 gene (formerly known as the MDR1 gene, and the condition is still referred to as Multidrug Resistance 1), is a membrane transport protein in the ATP-binding cassette superfamily. P-gp is normally expressed in various mammalian tissues including apical (luminal) membranes of epithelial cells lining the lower gastrointestinal tract, brain capillary endothelial cells, biliary canalicular cells, brush border of renal proximal tubules, placenta, and testes. P-gp limits drug absorption in the gastrointestinal tract and promotes drug elimination in the liver, kidneys, and intestine. Furthermore, P-gp restricts drug uptake into cells and tissues, in particular their permeation across the blood-brain barrier. Taken altogether, P-gp has an important protective function for the organism by eliminating potentially toxic compounds from the body and preventing their entry into the brain and organs of reproduction.

Because of the predominant role of P-gp in drug disposition, mutation of the ABCB1 gene alters the pharmacokinetic properties of P-gp transported drugs, leading to enhanced oral bioavailability and reduced drug elimination through the liver, kidneys, and gut. Moreover, the brain penetration of P-gp transported drugs is increased and in many cases provokes neurological toxicity.

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Age of Onset of Clinical Signs or Symptoms

MDR1 often presents in young adulthood, only because this is most commonly when a dog is first exposed to a problem drug like high dose ivermectin or acepromazine.

Clinical Signs

Symptoms arise after a dog has received an MDR1 problem drug or dosage and can range from vomiting and diarrhea to lethargy, seizures, or coma.

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Penetrance and Additional Impact on Phenotype

Interestingly, research indicates that all dogs with this variant in ABCB1 are descendants of a dog that lived in Great Britain before the genetic isolation of breeds by registry (ca. 1873). Dogs that inherit two abnormal copies (homozygous) will produce no normal p-glycoprotein and will be most strongly affected. Dogs that inherit only one abnormal copy of the MDR1 gene (heterozygous) can show some effects though they will be less severely impacted because some normal p-glycoprotein will still be produced.

Of note, several commonly used drugs can inhibit P-glycoprotein function, even in animals with normal ABCB1 gene structure. Consequently, veterinarians may encounter dogs and cats with intrinsic (genetically mediated) P-glycoprotein dysfunction, as well as with extrinsic, or acquired, P-glycoprotein dysfunction (animals receiving a drug that inhibits P-glycoprotein function). In ABCB1 wild-type (normal) dogs, ketoconazole and spinosad are most often associated with severe adverse effects because of their ability to inhibit P-glycoprotein function.

Approximate frequency for select breeds (from WSU):

- Australian Shepherd 50%
- Australian Shepherd Mini 50%
- Chinook 25%
- Collie 70%
- English Shepherd 15%
- German Shepherd Dog 10%
- Long-haired Whippet 50%
- McNab 30%
- Old English Sheepdog 5%
- Shetland Sheepdog 15%
- Silken Windhound 30%

Follow-up Diagnostics to Consider

This is usually a retroactive diagnosis after a dog has an adverse reaction to a problem drug--however, genetic testing could help avoid a first reaction altogether.

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Treatment and Management Options

- Drugs that have been documented to cause problems in dogs with the ABCB1 variant include (from WSU):
- Macrocyclic lactones (including such drugs as ivermectin, milbemycin, moxidectin, and selamectin) - Route of application and dosage is crucial for the safety of treatment with macrocyclic lactones. Whereas all available macrocyclic lactones can safely be administered to ABCB1 mutant dogs at doses usually used for heartworm prevention, these dogs will experience neurological toxicity following a high dose regimen which has historically been used for demodectic mange treatment. **All FDA-approved heartworm prevention products licensed in the United States have been tested and found to be safe in dogs with the MDR1 variant.** (For study results, see label indications for specific trademark products.)
- ABCB1 heterozygote dogs can be regarded as having an intermediate macrocyclic lactone-sensitive phenotype. Currently, there is no specific and safe antidote available for the treatment of macrocyclic lactone-induced toxicosis. Therefore, treatment is solely based on symptomatic and supportive care. Care should also be taken to minimize non-direct exposure to these drugs (e.g. environmental or large-animal treatment).
- Loperamide (ImodiumTM) - At doses used to treat diarrhea, this drug will cause neurological toxicity in dogs with the MDR1 variant. This drug should be avoided in all dogs with the MDR1 variant.
- Acepromazine - Dose reductions are required for dogs with one or two copies of the MDR1 variant.
- Butorphanol - Dose reductions are required for dogs with one or two copies of the MDR1 variant.
- Chemotherapy Agents (vincristine, vinblastine, doxorubicin, paclitaxel)- Dose reductions are required for dogs with one or two copies of the MDR1 variant in order to avoid severe toxicity.
- Apomorphine - Dose reductions are required for dogs with one or two copies of the MDR1 variant, as it can cause central nervous system depression at standard doses.

More Information

Additional information regarding drugs that are known to be transported by the human or rodent forms of the protein encoded by the MDR1 gene with or without additional research in dogs can be found at https://vcpl.vetmed.wsu.edu/problem-drugs. Recommended dosage adjustments from WSU can be found at https://www.cliniciansbrief.com/article/how-should-i-treat-dogs-cats-mdr1-mutation.

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References

Neff MW, Robertson KR, Wong AK, et al. Breed distribution and history of canine mdr1-1Delta, a pharmacogenetic mutation that marks the emergence of breeds from the collie lineage. Proc Natl Acad Sci U S A. 2004;101(32):11725-11730. doi:10.1073/pnas.0402374101

Deshpande D, Hill KE, Mealey KL, Chambers JP, Gieseg MA. The Effect of the Canine ABCB1-1Δ Mutation on Sedation after Intravenous Administration of Acepromazine. J Vet Intern Med. 2016;30(2):636-641. doi:10.1111/jvim.13827

Geyer J, Janko C. Treatment of MDR1 mutant dogs with macrocyclic lactones. Curr Pharm Biotechnol. 2012;13(6):969-986. doi:10.2174/138920112800399301

Mealey KL. Canine ABCB1 and macrocyclic lactones: heartworm prevention and pharmacogenetics. Vet Parasitol. 2008;158(3):215-222. doi:10.1016/j.vetpar.2008.09.009

Mealey KL, Bentjen SA, Gay JM, Cantor GH. Ivermectin sensitivity in collies is associated with a deletion mutation of the mdr1 gene. Pharmacogenetics. 2001;11(8):727-733. doi:10.1097/00008571-200111000-00012

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Notable Results

Ichthyosis, ICH1

How to interpret this result

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While Oliver is not at risk for developing ICH1, he can pass this variant on to the next generation. If Oliver is intended for breeding, please genotype any potential mates. You can email vetsupport@embarkvet.com to discuss with a genetic counselor how the genotype results should be applied to a breeding program.

Variant Info

PNPLA1 Exon 8
Recessive inheritance
1 copy of the variant

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What is Ichthyosis, ICH1?

As the largest organ in the body, skin protects the body from infection, allergens, pollutants, and UV light, and it plays a vital role in preventing dehydration. Any disorder that impairs skin anatomy or function or causes injury to the skin can lead to systemic illness.

Disorders of cornification (DOCs) are divided into primary and secondary causes. In primary cornification disorders, the excessive scaling is due to a direct defect in the formation of the outer skin layer (stratum corneum). The stratum corneum consists of overlapping layers of anucleate keratinocytes (corneocytes) encased in bilayers of lipid. This layer maintains the water content of the body by restricting water movement into and out of the skin. Secondary disorders are those where excessive scaling develops as a result of another condition (parasites, cancer, endocrinopathies).

Ichthyosis can be epidermolytic (EI) or nonepidermolytic (NI), which is determined based on the microscopic appearance of the skin. Dogs affected with epidermolytic ichthyosis have multiple regions of pigmented scale with alopecia (hair loss) and roughening of the skin. Nonepidermolytic ichthyosis, which can cause skin lesions and secondary inflammation, has been documented to affect Golden Retrievers and is caused by a variant in the PNPLA1 gene. PNPLA1 has a role in glycerophospholipid metabolism. This condition may also be referred to as ICH1.

Age of Onset of Clinical Signs or Symptoms

Typically, clinical signs develop in puppies but the disease tends to worsen with age. Golden Retrievers are typically diagnosed at less than one year of age; however, adult-onset cases are not uncommon. Severe hypermelanosis associated with rough and hyperpigmented skin on the ventrum may be noted by breeders as early as three to six weeks of age and could therefore be considered as an early cutaneous sign, often visible before the occurrence of the scaling.

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Clinical Signs

Ichthyosis may clinically present like many other things, including: allergies or a cutaneous drug reaction, parasites, infection, exposure to excessive UV light, endocrinopathies (Cushing's disease, hypothyroidism), autoimmune disease, epidermolysis bullosa, lethal acrodermatitis, vitamin and mineral deficiencies, sebaceous gland abnormalities, primary seborrhea, cancer, and dermatomyositis.

Ichthyosis is derived from the Greek root "ichthy," meaning fish, and was so named due to the visible scales on the skin. Ichthyotic dogs typically have large, greasy flakes of dandruff, but aren't itchy. The scales of skin can get so thick that they crack and cause uncomfortable fissures.

Affected dogs develop generalized scaling, initially with small to large whitish scales (often referred to as "snowflake-like") and progressively with blackish scales. Scales are typically distributed over most areas of the body: the lateral and ventral regions of the neck, trunk, rump, and dorsum and ventrum folds but do not appear on the head or extremities. Physical manifestations may wax and wane, and some dogs develop secondary bacterial skin infections that may confound a diagnosis.

Follow-up Diagnostics to Consider

For dogs showing signs of a skin disorder, the first step in diagnosing ichthyosis (and other DOCs) is for a veterinarian to examine the characteristic lesions. The veterinarian may perform blood work (complete blood count and serum chemistry), a skin scrape, skin cytology, dermatophyte (ringworm) culture, skin biopsy, +/- a urinalysis or specific endocrine testing. Genetic testing can also be done to confirm—or rule out—an inherited condition.

Primary disorders are generally diagnosed by ruling out all secondary causes, clinical presentation and/or age of onset, or skin biopsy.

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Treatment and Management Options

- There is no cure for Ichthyosis, ICH1.
- The treatments of choice are topical therapies such as specialized shampoos, moisturizing rinses, agents to remove excessive scale or to restore the skin barrier and thus prevent water loss, and topical medications to address secondary infections.
- Therapy must be tailored to the individual patient, and care should be taken not to damage or irritate the skin.
- Some dogs may benefit from oral essential fatty acid (EFA) supplementation or oral medications to treat infections.
- A novel topical therapy is under investigation to reinstate the corneocyte lipid envelope (CLE) in different forms of ichthyosis.

More Information

This form of Golden Retriever ichthyosis is generally considered "mild" although the severity can be dog-dependent. Of note, a new condition, named ICH2, has been reported in Golden Retrievers. ICH2 is a more severe form of Ichthyosis than ICH1. At this time, testing for ICH2 can only be done through the University of Pennsylvania.

References

Grall A, Guaguere E, Planchais S, et al. PNPLA1 mutations cause autosomal recessive congenital ichthyosis in golden retriever dogs and humans. Nat Genet. 2012;44(2):140-147. Published 2012 Jan 15. doi:10.1038/ng.1056

Mauldin EA. Canine ichthyosis and related disorders of cornification. Vet Clin North Am Small Anim Pract. 2013 Jan;43(1):89-97. doi: 10.1016/j.cvsm.2012.09.005. PMID: 23182326; PMCID: PMC3529142.

Guaguere E, Bensignor E, Kury S, et al. Clinical, histopathological and genetic data of ichthyosis in the golden retriever: a prospective study. J Small Anim Pract. 2009;50(5):227-235. doi:10.1111/j.1748-5827.2009.00730.x

Mauldin EA, Credille KM, Dunstan RW, Casal ML. The clinical and morphologic features of nonepidermolytic ichthyosis in the golden retriever. Vet Pathol. 2008 Mar;45(2):174-80. doi: 10.1354/vp.45-2-174. PMID: 18424829; PMCID: PMC3334879.

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Results Summary

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Auditory (1)

	Gene	Copies	Results
Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS	MYO7A	0	Clear

Cardiac (4)

Dilated Cardiomyopathy	Gene	Copies	Results
Dilated Cardiomyopathy, DCM1 - Doberman Pinscher Variant 1	PDK4	0	Clear
Dilated Cardiomyopathy, DCM2 - Doberman Pinscher Variant 2	TTN	0	Clear

Other	Gene	Copies	Results
Cardiomyopathy and Juvenile Mortality - Shepherd Variant	YARS2	0	Clear
Long QT Syndrome - English Springer Spaniel Variant	KCNQ1	0	Clear

Endocrine (3)

Hypothyroidism	Gene	Copies	Results
Congenital Dyshormonogenic Hypothyroidism with Goiter - Shih Tzu Variant	SLC5A5	0	Clear
Congenital Hypothyroidism - Tenterfield Terrier Variant	TPO	0	Clear
Congenital Hypothyroidism - Rat, Toy, and Hairless Terrier Variant	TPO	0	Clear

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Gastrointestinal (4)

Gastroenteropathy	Gene	Copies	Results
Lundehund Syndrome	LEPREL1	0	Clear

Malabsorptive Disorder	Gene	Copies	Results
Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption - Border Collie Variant	CUBN Exon 53	0	Clear
Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption - Beagle Variant	CUBN Exon 8	0	Clear
Inherited Selected Cobalamin Malabsorption with Proteinuria - Komondor Variant	CUBN	0	Clear

Hematologic (32)

Coagulopathy	Gene	Copies	Results
Bernard-Soulier Syndrome, BSS - Cocker Spaniel Variant	GP9	0	Clear
Congenital Macrothrombocytopenia - Cairn and Norfolk Terrier Variant	TUBB1 Exon 1	0	Clear
Factor IX Deficiency, Hemophilia B - Terrier Variant	F9 Exon 7	0	Clear
Factor IX Deficiency, Hemophilia B - Rhodesian Ridgeback Variant	F9 Exon 7	0	Clear
Factor VII Deficiency	F7 Exon 5	0	Clear
Factor VIII Deficiency, Hemophilia A - Boxer Variant	F8 Exon 10	0	Clear
Factor VIII Deficiency, Hemophilia A - German Shepherd Variant 1	F8 Exon 11	0	Clear
Factor VIII Deficiency, Hemophilia A - German Shepherd Variant 2	F8 Exon 1	0	Clear
Glanzmann's Thrombasthenia Type I - Great Pyrenees Variant	ITGA2B Exon 13	0	Clear

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	Gene	Copies	Results
Glanzmann's Thrombasthenia Type I - Otterhound Variant	ITGA2B Exon 12	0	Clear
May-Hegglin Anomaly - Pug Variant	MYH9	0	Clear
P2Y12 Receptor Platelet Disorder - Greater Swiss Mountain Dog Variant	P2Y12	0	Clear
Platelet Factor X Receptor Deficiency, Scott Syndrome - German Shepherd Dog Variant	TMEM16F	0	Clear
Prekallikrein Deficiency - Shih Tzu Variant	KLKB1 Exon 8	0	Clear
Thrombopathia - Basset Hound Variant	RASGRP1 Exon 5	0	Clear
Thrombopathia - Landseer Variant	RASGRP1 Exon 8	0	Clear
Thrombopathia - American Eskimo Dog Variant	RASGRP1 Exon 5	0	Clear
Von Willebrand Disease Type I, Type I vWD	VWF	0	Clear
Von Willebrand Disease Type II, Type II vWD - Pointer Variant	VWF	0	Clear
Von Willebrand Disease Type III, Type III vWD - Terrier Variant	VWF Exon 4	0	Clear
Von Willebrand Disease Type III, Type III vWD - Shetland Sheepdog Variant	VWF Exon 7	0	Clear

Red Blood Cell Abnormality	Gene	Copies	Results
Canine Elliptocytosis - Labrador Retriever Variant	SPTB Exon 30	0	Clear
Methemoglobinemia - Pomeranian Variant	CYB5R3	0	Clear
Pyruvate Kinase Deficiency - Basenji Variant	PKLR Exon 5	0	Clear
Pyruvate Kinase Deficiency - Labrador Retriever Variant	PKLR Exon 7	0	Clear

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	Gene	Copies	Results
Pyruvate Kinase Deficiency - Pug Variant	PKLR Exon 7	0	Clear
Pyruvate Kinase Deficiency - Beagle Variant	PKLR Exon 7	0	Clear
Pyruvate Kinase Deficiency - Terrier Variant	PKLR Exon 10	0	Clear

White Blood Cell Abnormality	Gene	Copies	Results
Canine Leukocyte Adhesion Deficiency Type I, CLAD I - Setter Variant	ITGB2 Exon 3	0	Clear
Canine Leukocyte Adhesion Deficiency Type III, CLAD III - German Shepherd Variant	FERMT3	0	Clear
Trapped Neutrophil Syndrome, TNS	VPS13B Exon 19	0	Clear

Other	Gene	Copies	Results
Ligneous Membranitis, LM - Scottish Terrier Variant	PLG	0	Clear

Immunologic (6)

	Gene	Copies	Results
Complement 3 Deficiency, C3 Deficiency - Brittany Variant	C3	0	Clear
Severe Combined Immunodeficiency, SCID - Terrier Variant	PRKDC	0	Clear
Severe Combined Immunodeficiency, SCID - Wetterhoun Variant	RAG1	0	Clear
Shar-Pei Autoinflammatory Disease, SPAID, Shar-Pei Fever	MTBP	0	Clear
X-linked Severe Combined Immunodeficiency, X-SCID - Basset Hound Variant	IL2RG Exon 1	0	Clear
X-linked Severe Combined Immunodeficiency, X-SCID - Corgi Variant	IL2RG	0	Clear

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Integument (18)

Collagen Abnormality	Gene	Copies	Results
Dystrophic Epidermolysis Bullosa - Golden Retriever Variant	COL7A1 Exon 68	0	Clear
Dystrophic Epidermolysis Bullosa - Central Asian Shepherd Dog Variant	COL7A1	0	Clear
Ehlers Danlos - Doberman Pinscher Variant	ADAMTS2	0	Clear
Musladin-Lueke Syndrome, MLS - Beagle Variant	ADAMTSL2 Exon 7	0	Clear

Keratin Abnormality	Gene	Copies	Results
Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatosis, Dry Eye Curly Coat Syndrome, CKCSID - Cavalier King Charles Spaniel Variant	FAM83H	0	Clear
Focal Non-Epidermolytic Palmoplantar Keratoderma, Pachyonychia Congenita - Dogue de Bordeaux Variant	KRT16 Exon 6	0	Clear
Hereditary Footpad Hyperkeratosis - Terrier and Kromfohrlander Variant	FAM83G	0	Clear
Hereditary Footpad Hyperkeratosis - Rottweiler Variant	DSG1	0	Clear
Hereditary Nasal Parakeratosis, HNPK - Labrador Retriever Variant	SUV39H2	0	Clear
Ichthyosis - Great Dane Variant	SLC27A4	0	Clear
Ichthyosis - American Bulldog Variant	NIPAL4 Exon 6	0	Clear
Ichthyosis, Epidermolytic Hyperkeratosis - Terrier Variant	KRT10 Intron 5	0	Clear
Ichthyosis, ICH1 - Golden Retriever Variant	PNPLA1 Exon 8	1	Notable

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Other	Gene	Copies	Results
Bald Thigh Syndrome - Greyhound Variant	IGFBP5	0	Clear
Ectodermal Dysplasia, Skin Fragility Syndrome - Chesapeake Bay Retriever Variant	PKP1 Intron 1	0	Clear
Lethal Acrodermatitis, LAD - Bull Terrier Variant	MKLN1	0	Clear
Oculocutaneous Albinism, OCA - Pekingese Variant	SLC45A2	0	Clear
X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia, XHED - German Shepherd Dog Variant	EDA	0	Clear

Metabolic (33)

Enzyme Deficiency	Gene	Copies	Results
Hypocatalasia, Acatalasemia - Beagle Variant	CAT	0	Clear
L-2-Hydroxyglutaricaciduria, L2HGA - Staffordshire Bull Terrier Variant	L2HGDH	0	Clear
Pyruvate Dehydrogenase Deficiency - Spaniel Variant	PDP1	0	Clear

Storage Disease	Gene	Copies	Results
Canine Fucosidosis - English Springer Spaniel Variant	FUCA1	0	Clear
GM1 Gangliosidosis - Shiba Inu Variant	GLB1 Exon 15	0	Clear
GM1 Gangliosidosis - Alaskan Husky Variant	GLB1 Exon 15	0	Clear
GM1 Gangliosidosis - Portuguese Water Dog Variant	GLB1 Exon 2	0	Clear
GM2 Gangliosidosis - Poodle Variant	HEXB Exon 3	0	Clear

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	Gene	Copies	Results
GM2 Gangliosidosis - Japanese Chin Variant	HEXA	0	Clear
Globoid Cell Leukodystrophy, Krabbe Disease - Terrier Variant	GALC Exon 5	0	Clear
Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA - Maltese Variant	G6PC	0	Clear
Glycogen Storage Disease Type II, Pompe's Disease, GSD II - Finnish and Swedish Lapphund, Lapponian Herder Variant	GAA Exon 15	0	Clear
Glycogen Storage Disease Type IIIA, GSD IIIA - Curly Coated Retriever Variant	AGL GDE	0	Clear
Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency - Whippet and English Springer Spaniel Variant	PFKM Exon 21	0	Clear
Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency - Wachtelhund Variant	PFKM Exon 8	0	Clear
Lagotto Storage Disease	ATG4D Exon 10	0	Clear
Late-Onset Neuronal Ceroid Lipofuscinosis, NCL 12 - Australian Cattle Dog Variant	ATP13A2	0	Clear
Mucopolysaccharidosis IIIB, Sanfilippo Syndrome Type B, MPS IIIB - Schipperke Variant	NAGLU	0	Clear
Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA - Dachshund Variant	SGSH	0	Clear
Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA - New Zealand Huntaway Variant	SGSH	0	Clear
Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII - Terrier Brasileiro Variant	GUSB	0	Clear
Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII - German Shepherd Variant	GUSB	0	Clear
Neuronal Ceroid Lipofuscinosis 1, NCL 1 - Dachshund Variant 1	PPT1 Exon 8	0	Clear
Neuronal Ceroid Lipofuscinosis 10, NCL 10 - American Bulldog Variant	CTSD Exon 5	0	Clear

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	Gene	Copies	Results
Neuronal Ceroid Lipofuscinosis 2, NCL 2 - Dachshund Variant 2	TPP1 Exon 4	0	Clear
Neuronal Ceroid Lipofuscinosis 5, NCL 5 - Border Collie Variant	CLN5 Exon 4	0	Clear
Neuronal Ceroid Lipofuscinosis 5, NCL 5 - Golden Retriever Variant	CLN5 Exon 4	0	Clear
Neuronal Ceroid Lipofuscinosis 6, NCL 6 - Australian Shepherd Variant	CLN6 Exon 7	0	Clear
Neuronal Ceroid Lipofuscinosis 7, NCL 7 - Chihuahua and Chinese Crested Variant	MFSD8	0	Clear
Neuronal Ceroid Lipofuscinosis 8, NCL 8 - English Setter Variant	CLN8 Exon 2	0	Clear
Neuronal Ceroid Lipofuscinosis 8, NCL 8 - Australian Shepherd Variant	CLN8	0	Clear
Neuronal Ceroid Lipofuscinosis 8, NCL 8 - Saluki Variant	CLN8	0	Clear
Neuronal Ceroid Lipofuscinosis, Cerebellar Ataxia, NCL4A - American Staffordshire Terrier Variant	ARSG Exon 2	0	Clear

Muscular (13)

Movement Disorder

	Gene	Copies	Results
Myotonia Congenita - Miniature Schnauzer Variant	CLCN1 Exon 7	0	Clear
Myotonia Congenita - Australian Cattle Dog Variant	CLCN1 Exon 23	0	Clear

Muscular Dystrophy

	Gene	Copies	Results
Limb Girdle Muscular Dystrophy - Boston Terrier Variant	SGCD	0	Clear
Muscular Dystrophy - Cavalier King Charles Spaniel Variant 1	DMD	0	Clear

Oliver

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	Gene	Copies	Results
Muscular Dystrophy - Golden Retriever Variant	DMD	0	Clear
Ullrich-like Congenital Muscular Dystrophy - Labrador Retriever Variant	COL6A3	0	Clear

Myopathy

	Gene	Copies	Results
Centronuclear Myopathy, CNM - Labrador Retriever Variant	PTPLA	0	Clear
Exercise-Induced Collapse, EIC	DNM1	0	Clear
Inflammatory Myopathy - Dutch Shepherd Variant	SLC25A12	0	Clear
Inherited Myopathy of Great Danes	BIN1	0	Clear
Myotubular Myopathy 1, X-linked Myotubular Myopathy, XL-MTM - Labrador Retriever Variant	MTM1 Exon 7	0	Clear
Nemaline Myopathy - American Bulldog Variant	NEB	0	Clear

Other

	Gene	Copies	Results
Myostatin Deficiency, Bully Whippet Syndrome	MSTN	0	Clear

Neurologic (32)

Brain or Seizure Disorder

	Gene	Copies	Results
Alaskan Husky Encephalopathy, Subacute Necrotizing Encephalomyelopathy	SLC19A3 Exon 2	0	Clear
Alexander Disease - Labrador Retriever Variant	GFAP Exon 4	0	Clear
Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy - Lagotto Romagnolo Variant	LGI2 Exon 8	0	Clear

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	Gene	Copies	Results
Cerebellar Abiotrophy, Neonatal Cerebellar Cortical Degeneration, NCCD - Beagle Variant	SPTBN2	0	Clear
Cerebellar Hypoplasia - Eurasier Variant	VLDLR	0	Clear
Hereditary Ataxia, Cerebellar Degeneration - Old English Sheepdog and Gordon Setter Variant	RAB24 Exon 1	0	Clear
Neonatal Encephalopathy with Seizures, NEWS - Poodle Variant	ATF2	0	Clear
Progressive Early-Onset Cerebellar Ataxia - Finnish Hound Variant	SEL1L	0	Clear
Spinocerebellar Ataxia with Myokymia and/or Seizures - Terrier Variant 2	KCNJ10	0	Clear
Spinocerebellar Ataxia, Late-Onset Ataxia, LoSCA - Terrier Variant 1	CAPN1	0	Clear
Spongy Degeneration with Cerebellar Ataxia 1, SDCA1, SeSAME/EAST Syndrome - Shepherd Variant 1	KCNJ10	0	Clear
Spongy Degeneration with Cerebellar Ataxia 2, SDCA2 - Shepherd Variant 2	ATP1B2	0	Clear

Movement Disorder	Gene	Copies	Results
Degenerative Myelopathy, DM	SOD1A	0	Clear
Hypomyelination and Tremors - Weimaraner Variant	FNIP2	0	Clear
Juvenile Myoclonic Epilepsy - Rhodesian Ridgeback Variant	DIRAS1	0	Clear
Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD - Kerry Blue Terrier Variant	SERAC1	0	Clear

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	Gene	Copies	Results
Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD - Chinese Crested Variant	SERAC1	0	Clear
Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome - English Springer Spaniel Variant	PLP1	0	Clear

Narcolepsy	Gene	Copies	Results
Narcolepsy - Doberman Pinscher Variant	HCRTR2	0	Clear
Narcolepsy - Labrador Retriever Variant	HCRTR2	0	Clear
Narcolepsy - Dachshund Variant	HCRTR2	0	Clear

Neurodegenerative Disorder	Gene	Copies	Results
Fetal-Onset Neonatal Neuroaxonal Dystrophy - Giant Schnauzer Variant	MFN2	0	Clear
Neuroaxonal Dystrophy, NAD - Spanish Water Dog Variant	TECPR2	0	Clear
Neuroaxonal Dystrophy, NAD - Rottweiler Variant	VPS11	0	Clear

Neuropathy	Gene	Copies	Results
Alaskan Malamute Polyneuropathy, AMPN	NDRG1	0	Clear
Demyelinating Polyneuropathy - Miniature Schnauzer Variant	SBF2/MTRM13	0	Clear
Juvenile Laryngeal Paralysis and Polyneuropathy, Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation, POANV - Rottweiler Variant	RAB3GAP1	0	Clear
Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1, LPN1	ARHGEF10 Exon 17	0	Clear

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	Gene	Copies	Results
Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 2, LPN2	GJA9	0	Clear
Laryngeal Paralysis - Miniature Bull Terrier Variant	RAPGEF6	0	Clear

Sensory Neuropathy

	Gene	Copies	Results
Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS - Spaniel and Pointer Variant	GDNF-AS	0	Clear
Sensory Neuropathy - Border Collie Variant	FAM134B	0	Clear

Neuromuscular (7)

Junctionopathy

	Gene	Copies	Results
Congenital Myasthenic Syndrome, CMS - Old Danish Pointing Dog Variant	CHAT Exon 6	0	Clear
Congenital Myasthenic Syndrome, CMS - Labrador Retriever Variant	COLQ Exon 14	0	Clear
Congenital Myasthenic Syndrome, CMS - Jack Russell Terrier Variant	CHRNE	0	Clear
Congenital Myasthenic Syndrome, CMS - Golden Retriever Variant	COLQ	0	Clear
Myasthenia Gravis-Like Syndrome - Heideterrier Variant	CHRNE	0	Clear

Movement Disorder

	Gene	Copies	Results
Episodic Falling Syndrome - Cavalier King Charles Spaniel Variant	BCAN Exons 1-4	0	Clear
Paroxysmal Dyskinesia, PxD - Soft Coated Wheaten Terrier Variant	PIGN	0	Clear

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Ophthalmologic (31)

Glaucoma	Gene	Copies	Results
Goniodysgenesis and Glaucoma, Pectinate Ligament Dysplasia, PLD - Border Collie Variant	OLFML3	0	Clear
Primary Open Angle Glaucoma - Norwegian Elkhound Variant	ADAMTS10	0	Clear
Primary Open Angle Glaucoma - Beagle Variant	ADAMTS10	0	Clear
Primary Open Angle Glaucoma - Basset Fauve de Bretagne Variant	ADAMTS17	0	Clear
Primary Open Angle Glaucoma and Primary Lens Luxation - Chinese Shar-Pei Variant	ADAMTS17	0	Clear

Iris or Lens	Gene	Copies	Results
Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts - Australian Shepherd Variant	HSF4	0	Clear
Primary Lens Luxation	ADAMTS17	0	Clear

Retinopathy	Gene	Copies	Results
Achromatopsia - German Shepherd Variant	CNGA3 Exon 7	0	Clear
Achromatopsia - Labrador Retriever Variant	CNGA3 Exon 7	0	Clear
Autosomal Dominant Progressive Retinal Atrophy - English Mastiff and Bullmastiff Variant	RHO Exon 1	0	Clear
Canine Multifocal Retinopathy, cmr1	BEST1/VMD2 Exon 2	0	Clear
Canine Multifocal Retinopathy, cmr2 - Coton de Tulear Variant	BEST1/VMD2 Exon 5	0	Clear
Canine Multifocal Retinopathy, cmr3 - Finnish and Swedish Lapphund, Lapponian Herder Variant	BEST1/VMD2 Exon 10	0	Clear

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	Gene	Copies	Results
Collie Eye Anomaly, Choroidal Hypoplasia, CEA	NHEJ1 Intron 4	0	Clear
Congenital Stationary Night Blindness - Briard Variant	RPE65	0	Clear
Congenital Stationary Night Blindness - Beagle Variant	LRIT3	0	Clear
Day Blindness, Cone Degeneration, Achromatopsia - Alaskan Malamute Variant	CNGB3	0	Clear
Day Blindness, Cone Degeneration, Achromatopsia - German Shorthaired Pointer Variant	CNGB3 Exon 6	0	Clear
Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1	SLC4A3 Exon 16	0	Clear
Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2	TTC8 Exon 8	0	Clear
Macular Corneal Dystrophy, MCD - Labrador Retriever Variant	CHST6	0	Clear
Progressive Retinal Atrophy - Basenji Variant	SAG	0	Clear
Progressive Retinal Atrophy, CNGA - Shetland Sheepdog Variant	CNGA1 Exon 9	0	Clear
Progressive Retinal Atrophy, Cone-Rod Dystrophy 1, crd1 - American Staffordshire Terrier Variant	PDE6B	0	Clear
Progressive Retinal Atrophy, Cone-Rod Dystrophy 4, crd4/cord1	RPGRIP1 Exon 2	0	Clear
Progressive Retinal Atrophy, PRA1 - Papillon Variant	CNGB1	0	Clear
Progressive Retinal Atrophy, PRA3 - Tibetan Spaniel and Terrier Variant	FAM161A	0	Clear
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration, prcd	PRCD Exon 1	0	Clear
Progressive Retinal Atrophy, Rod-Cone Dysplasia 1, rcd1 - Irish Setter Variant	PDE6B Exon 21	0	Clear

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	Gene	Copies	Results
Progressive Retinal Atrophy, Rod-Cone Dysplasia 3, rcd3 - Corgi Variant	PDE6A	0	Clear
X-Linked Progressive Retinal Atrophy 1, XL-PRA1 - Samoyed and Husky Variant	RPGR Exon 15	0	Clear

Oral Cavity (4)

Developmental Disorder	Gene	Copies	Results
Cleft Lip and/or Cleft Palate - Nova Scotia Duck Tolling Retriever Variant	ADAMTS20	0	Clear

Tooth Structure Defect	Gene	Copies	Results
Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia - Italian Greyhound Variant	ENAM	0	Clear
Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia - Parson Russell Terrier Variant	ENAM	0	Clear
Raine Syndrome, Canine Dental Hypomineralization Syndrome - Border Collie Variant	FAM20C	0	Clear

Personalized Medicine (3)

	Gene	Copies	Results
Alanine Aminotransferase Activity	GPT	0	Clear
MDR1 Drug Sensitivity	ABCB1	2	At risk
Malignant Hyperthermia	RYR1	0	Clear

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Pulmonary (4)

	Gene	Copies	Results
Neonatal Interstitial Lung Disease - Airedale Terrier Variant	LAMP3	0	Clear
Primary Ciliary Dyskinesia, PCD - Old English Sheepdog Variant	CCDC39	0	Clear
Primary Ciliary Dyskinesia, PCD - Alaskan Malamute Variant	NME5	0	Clear
Recurrent Inflammatory Pulmonary Disease, RIPD - Rough Collie Variant	AKNA	0	Clear

Skeletal (9)

Chondrodystrophy

	Gene	Copies	Results
Chondrodystrophy - Norwegian Elkhound and Karelian Bear Dog Variant	ITGA10	0	Clear
Oculoskeletal Dysplasia 2, Dwarfism-Retinal Dysplasia 2, drd2, OSD2 - Samoyed Variant	COL9A2 5' UTR	0	Clear
Osteochondrodysplasia, Skeletal Dwarfism - Poodle Variant	SLC13A1	0	Clear
Skeletal Dysplasia 2, SD2 - Labrador Retriever Variant	COL11A2	0	Clear

Decreased Bone Strength

	Gene	Copies	Results
Hereditary Vitamin D-Resistant Rickets - Pomeranian Variant	VDR Exon 4	0	Clear
Osteogenesis Imperfecta, Brittle Bone Disease - Beagle Variant	COL1A2	0	Clear
Osteogenesis Imperfecta, Brittle Bone Disease - Dachshund Variant	SERPINH1 Exon 5	0	Clear
Osteogenesis Imperfecta, Brittle Bone Disease - Golden Retriever Variant	COL1A1 Exon 18	0	Clear

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Other	Gene	Copies	Results
Craniomandibular Osteopathy, CMO - Terrier and Australian Shepherd Variant	SLC37A2 Exon 15	0	Clear

Urogenital (14)

Nephropathy	Gene	Copies	Results
Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN - English Springer Spaniel Variant	COL4A4 Exon 30	0	Clear
Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN - Cocker Spaniel Variant	COL4A4 Exon 3	0	Clear
Fanconi Syndrome - Basenji Variant	FAN1	0	Clear
Polycystic Kidney Disease, PKD - Bull Terrier Variant	PKD1 Exon 29	0	Clear
Protein Losing Nephropathy, PLN - Soft Coated Wheaten and Airedale Terrier Variant	NPHS1	0	Clear
X-Linked Hereditary Nephropathy, XLHN - Samoyed Variant 2	COL4A5 Exon 35	0	Clear

Urolithiasis	Gene	Copies	Results
2,8-Dihydroxyadenine Urolithiasis, 2,8-DHA Urolithiasis - American Indian Dog Variant	APRT Exon 3	0	Clear
Cystinuria Type I-A - Newfoundland Variant	SLC3A1 Exon 2	0	Clear
Cystinuria Type II-A - Australian Cattle Dog Variant	SLC3A1 Exon 6	0	Clear
Cystinuria Type II-B - Miniature Pinscher Variant	SLC7A9 Exon 9	0	Clear
Hyperuricosuria and Hyperuricemia or Urolithiasis, HUU	SLC2A9 Exon 5	0	Clear
Primary Hyperoxaluria - Coton de Tulear Variant	AGXT Exon 2	0	Clear

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Other	Gene	Copies	Results
Persistent Mullerian Duct Syndrome, PMDS - Miniature and Standard Schnauzer Variant	AMHR2	0	Clear
Renal Cystadenocarcinoma and Nodular Dermatofibrosis, RCND - German Shepherd Dog Variant	FLCN	0	Clear