Canine HealthCheck ✔

Demographic Information

Call Name	Ava	Owner	Jennifer Hoyt
Registered Name	Porter's Ava @ Schaffert's	Registration Number	ASDT-NE-1800186
Breed	Toy Australian Shepherd	Tattoo	
Sex	F	Microchip	
DOB	September 17, 2018	Laboratory #	AN-20-000281

These tests were developed and performed by Paw Print Genetics®, Spokane WA.

Explanation of Results

Normal	A 'Normal' result means that your dog does not have the mutation that causes the associated genetic disease.
Carrier	A 'Carrier' result indicates that your dog has inherited one copy of the mutation that has been reported to cause this genetic disease. Your dog may not be clinically affected by this mutation because two copies of the mutation are usually required to cause disease.
Carrier / At-Risk	A 'Carrier / At-Risk' result indicates that your dog inherited one copy of the mutation that has been reported to cause this genetic disease. Based on the mode of genetic inheritance for this particular disease, inheriting one mutant copy of the gene may result in the disease. Dogs with one copy of the mutation may have a milder phenotype as compared to dogs with two copies of this mutation.
At-Risk / Affected	An 'At-Risk / Affected' result indicates that your dog inherited one or two copies of the mutation that has been reported to cause this genetic disease. Based on the mode of genetic inheritance for this particular disease, inheriting one or two mutant copies of the gene may result in the disease.
No Result	'No Result' indicates that we were unable to obtain a genotype for your dog for this specific disease or trait and does not mean that your dog is a carrier or at-risk for this disease. There are a variety of reasons why a specific test may not provide a reportable result. Unique variations in the genetic code of some individuals may exist and cause certain regions of the genome to not perform properly with a specific test. In addition, suboptimal sampling of the dog's cheek cells could also result in poor sample performance due to inadequate cell counts, bacterial and fungal growth, or the presence of other test inhibitors. An acceptable level of tests with no results has been determined by Paw Print Genetics. Dogs with at least 90% of the test results are determined to be acceptable and reportable. If your dog has an unacceptable level of tests with no results, you will be contacted for a new sample to repeat the testing.

Please review our testing terms and disclaimers regarding your results.

WT: wild type (normal) M: mutant Y: Y chromosome (male)

Breed Profile

Disease Name	Genotype	Interpretation
Coagulation Factor VII Deficiency	WT/WT	Normal (Clear)
Collie Eye Anomaly	WT/WT	Normal (Clear)
Cone Degeneration	WT/WT	Normal (Clear)
Degenerative Myelopathy Common Variant	WT/WT	Normal (Clear)
Exercise-Induced Collapse	WT/WT	Normal (Clear)
Hereditary Cataracts Australian Shepherd Type	WT/WT	Normal (Clear)
Hyperuricosuria	WT/WT	Normal (Clear)
Multidrug Resistance 1	WT/WT	Normal (Clear)
Multifocal Retinopathy 1	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 6	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 8 Australian Shepherd Type	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration prcd	WT/WT	Normal (Clear)

WT: wild type (normal) M: mutant Y: Y chromosome (male)

Coat Colors & Traits

Trait Name	Genotype	Interpretation
A Locus Agouti	a^t/a^t	Tricolor, black and tan
B Locus Brown	B/B	Black coat, nose and foot pads
B Locus (Brown) - b^a	0	
B Locus (Brown) - b^c	0	
B Locus (Brown) - b^d	0	
B Locus (Brown) - b^s	0	
Brachycephaly	BR/BR	Likely medium to long muzzle
Chondrodysplasia CDPA	cd/cd	Likely typical leg length
Cu Locus Curly Hair	Cu/Cu	Straight coat
D Locus Dilute	D/D	Non dilute
D Locus (Dilute) - d^1	0	
D Locus (Dilute) - d^2	0	
E Locus Yellow/Red	E/E	Black
E^g Locus Grizzle, Afghan Hound Type	N/N	No grizzle
E^m Locus Melanistic Mask	E^m/E^m	Melanistic mask
H Locus Harlequin, Great Dane Type	h/h	No harlequin
K Locus Dominant Black	k^y/k^y	Agouti expression allowed
L Locus Long Hair/Fluffy	Lh/Lh	Longhaired
L Locus (Long Hair/Fluffy) - Lh^1	2	
L Locus (Long Hair/Fluffy) - Lh^2	0	
Polydactyly	pd/pd	Normal (typical) toes (likely no hind dewclaws)
SD Locus Shedding	sd/SD	Moderate shedding
Sex Determination	X/X	Female
T Locus Natural Bobtail	t/t	Normal tail

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Determinants of coat colors and traits are complex. Many of these variants are known and many of the genes screened in the Canine HealthCheck interact. In addition, not all the genetic factors that contribute to a dog's coat color and traits are known. Because of the complexities in gene-gene interactions, the coat colors and traits reported in your Canine HealthCheck results may vary from your dog's actual appearance. Individual differences in genes throughout the canine genome, not tested in this genetic screen, may also affect the final coat color or traits seen in your dog.

Diseases

Disease Name	Genotype	Interpretation
Alaskan Husky Encephalopathy	WT/WT	Normal (Clear)
Alaskan Malamute Polyneuropathy	WT/WT	Normal (Clear)
Amelogenesis Imperfecta	WT/WT	Normal (Clear)
Benign Familial Juvenile Epilepsy	WT/WT	Normal (Clear)
Canine Multiple System Degeneration	WT/WT	Normal (Clear)

Chinese Crested Type		
Canine Multiple System Degeneration Kerry Blue Terrier Type	WT/WT	Normal (Clear)
Cerebellar Ataxia Finnish Hound Type	WT/WT	Normal (Clear)
Chondrodysplasia Karelian Bear Dog and Norwegian Elkhound Type	WT/WT	Normal (Clear)
Coagulation Factor VII Deficiency	WT/WT	Normal (Clear)
Collie Eye Anomaly	WT/WT	Normal (Clear)
Complement 3 Deficiency	WT/WT	Normal (Clear)
Cone Degeneration	WT/WT	Normal (Clear)
Cone Degeneration German Shorthaired Pointer Type	WT/WT	Normal (Clear)
Congenital Myasthenic Syndrome Labrador Retriever Type	WT/WT	Normal (Clear)
Congenital Myasthenic Syndrome Old Danish Pointer Type	WT/WT	Normal (Clear)
Congenital Stationary Night Blindness	WT/WT	Normal (Clear)
Cyclic Neutropenia	WT/WT	Normal (Clear)
Cystinuria Australian Cattle Dog Type	WT/WT	Normal (Clear)
Cystinuria Labrador Retriever Type	WT/WT	Normal (Clear)
Cystinuria Miniature Pinscher Type	WT/WT	Normal (Clear)
Cystinuria Newfoundland Type	WT/WT	Normal (Clear)
Degenerative Myelopathy Common Variant	WT/WT	Normal (Clear)
Degenerative Myelopathy Early-Onset Risk Modifier Pembroke Welsh Corgi Type	M/M	Two Copy Carrier - Not associated with disease
Dilated Cardiomyopathy Doberman Pinscher Type Risk Factor, Variant 1	No Result	No Result
Dry Eye Curly Coat Syndrome	WT/WT	Normal (Clear)
Dystrophic Epidermolysis Bullosa	WT/WT	Normal (Clear)
Early Retinal Degeneration	WT/WT	Normal (Clear)
Ectodermal Dysplasia Chesapeake Bay Retriever Type	WT/WT	Normal (Clear)
Ectodermal Dysplasia, X-Linked Shepherd Type	WT/WT	X-Linked Female Normal
Elliptocytosis	WT/WT	Normal (Clear)
Epidermolytic Hyperkeratosis	WT/WT	Normal (Clear)
Episodic Falling Syndrome	WT/WT	Normal (Clear)
Exercise-Induced Collapse	WT/WT	Normal (Clear)
Familial Nephropathy Cocker Spaniel Type	WT/WT	Normal (Clear)
Familial Nephropathy English Springer Spaniel Type	WT/WT	Normal (Clear)
Gallbladder Mucoceles	WT/WT	Normal (Clear)
Glanzmann's Thrombasthenia Great Pyrenees Type	WT/WT	Normal (Clear)
Glanzmann's Thrombasthenia Otterhound Type	WT/WT	Normal (Clear)

Globoid Cell Leukodystrophy Irish Setter Type	WT/WT	Normal (Clear)
Globoid Cell Leukodystrophy Terrier Type	WT/WT	Normal (Clear)
Glycogen Storage Disease Ia	WT/WT	Normal (Clear)
Glycogen Storage Disease IIIa	WT/WT	Normal (Clear)
Glycogen Storage Disease VII Wachtelhund Type	WT/WT	Normal (Clear)
Glycogen Storage Disease VII, PFK Deficiency	WT/WT	Normal (Clear)
GM1 Gangliosidosis Alaskan Husky Type	WT/WT	Normal (Clear)
GM1 Gangliosidosis Portuguese Water Dog Type	WT/WT	Normal (Clear)
GM1 Gangliosidosis Shiba Inu Type	WT/WT	Normal (Clear)
GM2 Gangliosidosis Japanese Chin Type	WT/WT	Normal (Clear)
GM2 Gangliosidosis Poodle Type	WT/WT	Normal (Clear)
Greyhound Polyneuropathy	WT/WT	Normal (Clear)
Hemophilia A Boxer Type	WT/WT	X-Linked Female Normal
Hemophilia A German Shepherd Dog, Type 1	WT/WT	X-Linked Female Normal
Hemophilia A German Shepherd Dog, Type 2	WT/WT	X-Linked Female Normal
Hemophilia B Cairn Terrier Type	WT/WT	X-Linked Female Normal
Hemophilia B Lhasa Apso Type	WT/WT	X-Linked Female Normal
Hemophilia B Rhodesian Ridgeback Type	WT/WT	X-Linked Female Normal
Hereditary Cataracts	WT/WT	Normal (Clear)
Hereditary Cataracts Australian Shepherd Type	WT/WT	Normal (Clear)
Hereditary Footpad Hyperkeratosis Irish Terrier and Kromfohrländer Type	WT/WT	Normal (Clear)
Hereditary Nasal Parakeratosis	WT/WT	Normal (Clear)
Hereditary Nephritis Samoyed Type	WT/WT	X-Linked Female Normal
Hyperuricosuria	WT/WT	Normal (Clear)
Ichthyosis American Bulldog Type	WT/WT	Normal (Clear)
Ichthyosis Golden Retriever Type	WT/WT	Normal (Clear)
Inherited Myopathy of Great Danes	WT/WT	Normal (Clear)
Intestinal Cobalamin Malabsorption Beagle Type	WT/WT	Normal (Clear)
Intestinal Cobalamin Malabsorption Border Collie Type	WT/WT	Normal (Clear)
Juvenile Laryngeal Paralysis and Polyneuropathy	WT/WT	Normal (Clear)
Juvenile Myoclonic Epilepsy Rhodesian Ridgeback Type	WT/WT	Normal (Clear)
L-2-Hydroxyglutaric Aciduria Staffordshire Bull Terrier Type	WT/WT	Normal (Clear)

Late Onset Ataxia	WT/WT	Normal (Clear)
Leukocyte Adhesion Deficiency, Type I	WT/WT	Normal (Clear)
Leukocyte Adhesion Deficiency, Type III	WT/WT	Normal (Clear)
Ligneous Membranitis	WT/WT	Normal (Clear)
May-Hegglin Anomaly	WT/WT	Normal (Clear)
Mucopolysaccharidosis I	WT/WT	Normal (Clear)
Mucopolysaccharidosis IIIA Dachshund Type	WT/WT	Normal (Clear)
Mucopolysaccharidosis IIIA New Zealand Huntaway Type	WT/WT	Normal (Clear)
Mucopolysaccharidosis VII Shepherd Type	WT/WT	Normal (Clear)
Multidrug Resistance 1	WT/WT	Normal (Clear)
Multifocal Retinopathy 1	WT/WT	Normal (Clear)
Multifocal Retinopathy 2	WT/WT	Normal (Clear)
Multifocal Retinopathy 3	WT/WT	Normal (Clear)
Muscular Dystrophy Golden Retriever Type	WT/WT	X-Linked Female Normal
Musladin-Lueke Syndrome	WT/WT	Normal (Clear)
Myostatin Deficiency Whippet and Longhaired Whippet Type	WT/WT	Normal (Clear)
Myotonia Congenita Australian Cattle Dog Type	WT/WT	Normal (Clear)
Myotonia Congenita Schnauzer Type	WT/WT	Normal (Clear)
Myotubular Myopathy 1	WT/WT	X-Linked Female Normal
Narcolepsy Dachshund Type	WT/WT	Normal (Clear)
Narcolepsy Doberman Pinscher Type	WT/WT	Normal (Clear)
Narcolepsy Labrador Retriever Type	WT/WT	Normal (Clear)
Neonatal Cerebellar Cortical Degeneration	WT/WT	Normal (Clear)
Neonatal Encephalopathy with Seizures	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis Tibetan Terrier Type	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 1 Cane Corso Type	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 10	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 2	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 4A	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 5 Australian Cattle Dog/Border Collie Type	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 6	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 8 Australian Shepherd Type	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 8 Setter Type	WT/WT	Normal (Clear)
Osteogenesis Imperfecta Beagle Type	WT/WT	Normal (Clear)
Osteogenesis Imperfecta Dachshund Type	No Result	No Result

Osteogenesis Imperfecta Golden Retriever Type	WT/WT	Normal (Clear)
P2RY12 Receptor Platelet Disorder	WT/WT	Normal (Clear)
Pembroke Welsh Corgi Duchenne Muscular Dystrophy	WT/WT	X-Linked Female Normal
Persistent Müllerian Duct Syndrome	No Result	No Result
Polyneuropathy Leonberger and Saint Bernard Type	WT/WT	Normal (Clear)
Polyneuropathy Leonberger Type 2	WT/WT	Normal (Clear)
Pompe Disease	WT/WT	Normal (Clear)
Prekallikrein Deficiency	WT/WT	Normal (Clear)
Primary Ciliary Dyskinesia	WT/WT	Normal (Clear)
Primary Hyperoxaluria	WT/WT	Normal (Clear)
Primary Lens Luxation	WT/WT	Normal (Clear)
Primary Open Angle Glaucoma	WT/WT	Normal (Clear)
Progressive Retinal Atrophy Basenji Type	WT/WT	Normal (Clear)
Progressive Retinal Atrophy Bullmastiff/Mastiff Type	WT/WT	Normal (Clear)
Progressive Retinal Atrophy Irish Setter Type	WT/WT	Normal (Clear)
Progressive Retinal Atrophy Sloughi Type	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Cone-Rod Dystrophy	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Cone-Rod Dystrophy 1	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Cone-Rod Dystrophy 3	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Generalized	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Golden Retriever 1	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Golden Retriever 2	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, PRA1 Papillon Type	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration prcd	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Rod-Cone Dysplasia 3	WT/WT	Normal (Clear)
Pyruvate Dehydrogenase Deficiency	WT/WT	Normal (Clear)
Pyruvate Kinase Deficiency Basenji Type	WT/WT	Normal (Clear)
Pyruvate Kinase Deficiency Beagle Type	WT/WT	Normal (Clear)
Pyruvate Kinase Deficiency Labrador Retriever Type	WT/WT	Normal (Clear)
Pyruvate Kinase Deficiency Pug Type	WT/WT	Normal (Clear)
Pyruvate Kinase Deficiency Terrier Type	WT/WT	Normal (Clear)
Renal Cystadenocarcinoma and Nodular Dermatofibrosis	WT/WT	Normal (Clear)
Severe Combined Immunodeficiency Disease Terrier Type	WT/WT	Normal (Clear)
Severe Combined Immunodeficiency Disease Wetterhoun Type	WT/WT	Normal (Clear)
Severe Combined Immunodeficiency Disease, X-Linked Basset Hound Type	WT/WT	X-Linked Female Normal

Severe Combined Immunodeficiency Disease, X-Linked Corgi Type	WT/WT	X-Linked Female Normal
Shar-Pei Autoinflammatory Disease	WT/WT	Normal (Clear)
Skeletal Dysplasia 2	WT/WT	Normal (Clear)
Spinal Dysraphism	WT/WT	Normal (Clear)
Spinocerebellar Ataxia	WT/WT	Normal (Clear)
Startle Disease	WT/WT	Normal (Clear)
Thrombopathia American Eskimo Dog Type	WT/WT	Normal (Clear)
Thrombopathia Basset Hound Type	WT/WT	Normal (Clear)
Thrombopathia Newfoundland Type	WT/WT	Normal (Clear)
Trapped Neutrophil Syndrome	WT/WT	Normal (Clear)
Von Willebrand Disease I	WT/WT	Normal (Clear)
Von Willebrand Disease II	WT/WT	Normal (Clear)
Von Willebrand Disease III Kooikerhondje Type	WT/WT	Normal (Clear)
Von Willebrand Disease III Scottish Terrier Type	WT/WT	Normal (Clear)
Von Willebrand Disease III Shetland Sheepdog Type	WT/WT	Normal (Clear)

WT: wild type (normal) M: mutant Y: Y chromosome (male)

Helen F Smith, PhD

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

Coat Color and Trait Certificate

Call Name:	Ava	**Laboratory #:**	299432
Registered Name:	Porter's Ava @ Schaffert's	**Registration #:**	ASDT-NE-1800186
Breed:	Toy Australian Shepherd	**Certificate Date:**	April 19, 2022
Sex:	Female		
DOB:	Sept. 2018		

This canine's DNA showed the following genotype(s):

Coat Color/Trait Test	Gene	Genotype	Interpretation
M Locus (Merle)	*PMEL*	m/M	*See detailed interpretation

Interpretation:

M Locus Genotype: m/M^{239}

This dog carries one copy of the **m** (non-merle, wild-type) allele and one copy of the **M** (merle insertion variant) allele of the *PMEL* gene. This dog will pass on one copy of the **m** (non-merle, wild-type) allele to 50% of its offspring and one copy of the **M** (merle insertion variant) allele to 50% of its offspring. The approximate size of the M allele of this dog (+/- 1 base pair) is listed in superscript in the genotype. Merle is inherited in a dominant fashion, meaning that only one copy of an M allele is necessary for a dog to display some variation of the merle coat color/pattern, which is marked by random dilution of eumelanin (black pigment) leaving patches of normal coat color within areas of diluted pigmentation.

Specific sizes of the M allele have been associated with the potential to produce "classic" merle patterning or other M-associated coat color variations. Merle is most appropriately viewed as a spectrum of coat colors/patterns and the size of the variant M allele is associated with a coat color/pattern somewhere within that spectrum. Although some coat color/pattern variations have been associated with specific sizes of the M allele in certain breeds, referred to here as a 'bin', the size of the M allele does not guarantee a specific outcome. In general, dogs with M allele sizes between 200 – 246 base pairs (bp) have been associated with non-merle or minimal-merle coat colors/patterns and are often referred to as "cryptic" merle; M allele sizes between 247 – 264 bp have been associated with "atypical" or "diluted" coat colors/patterns; M allele sizes between 265 – 269 bp have been associated with the "classic" merle coat colors/patterns; and M allele sizes between 270 – 280 bp have been associated with a "tweed", "harlequin" or "patchwork" merle coat colors/patterns. Many exceptions to the coat color/pattern associations found in the various M allele bin sizes listed here have been identified. Therefore, care should be taken when correlating M allele sizes with anticipated coat color/pattern outcomes. These bin sizes should not be interpreted as having discrete boundaries but should be viewed as a range within which specific coat colors and patterns are likely. Variations in genetic background between breeds and in individual dogs within a breed may result in the identification of different coat colors/patterns not typically found in a given bin, especially when the size of an M allele is at the border between bins. Furthermore, due to the complex nature of the merle insertion variant and the limitations of currently available molecular technologies, precise sizing of the merle insertion variant is challenging. However, the sizing of the merle insertion variant in our laboratory has been validated to be accurate to within +/- 1 bp which, nevertheless, makes correlations between genotype and coat color/pattern of dogs close to the boundaries of a specific bin potentially problematic. In addition, the M allele bins defined here are only relevant to test results generated by Paw Print Genetics. The variable nature of the M gene variant and subtle differences in methodologies used by each laboratory precludes strict interlaboratory genotype comparisons. Therefore, in some cases, it may be prudent to test related dogs in a single laboratory if comparisons across related dogs or dogs within a breed are desired.