

Canine Genetic Health Certificate™

Call Name:	Harper	Laboratory #:	465836
Registered Name:	Lone Pine's Harper At Pleasant Woods	Registration #:	DN72167702
Breed:	Miniature American Shepherd	Microchip #:	956000014385661
Sex:	Female	Certificate Date:	Oct. 29, 2024
DOB:	June 2022		

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

Disease	Gene	Genotype	Interpretation
Chondrodystrophy with Intervertebral Disc Disease Risk Factor (CDDY with IVDD)	<i>CFA12 FGF4</i>	WT/WT	Normal (Clear) - No CDDY or Increased IVDD Risk
Coagulation Factor VII Deficiency	<i>F7</i>	WT/WT	Normal (Clear)
Collie Eye Anomaly	<i>NHEJ1</i>	WT/WT	Normal (Clear)
Cone Degeneration	<i>CNGB3</i>	WT/WT	Normal (Clear)
Cranio-mandibular Osteopathy	<i>SLC37A2</i>	WT/WT	Normal (Clear)
Degenerative Myelopathy (Common Variant)	<i>SOD1</i>	WT/WT	Normal (Clear)
Exercise-Induced Collapse	<i>DNM1</i>	WT/WT	Normal (Clear)
Hereditary Ataxia (Australian Shepherd Type)	<i>PNPLA8</i>	WT/WT	Normal (Clear)
Hereditary Cataracts (Australian Shepherd Type)	<i>HSF4</i>	WT/WT	Normal (Clear)
Hyperuricosuria	<i>SLC2A9</i>	WT/WT	Normal (Clear)
Intestinal Cobalamin Malabsorption (Australian Shepherd Type)	<i>AMN</i>	WT/WT	Normal (Clear)
Intestinal Cobalamin Malabsorption (Border Collie Type)	<i>CUBN</i>	WT/WT	Normal (Clear)

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

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Disease	Gene	Genotype	Interpretation
Junctional Epidermolysis Bullosa (Australian Shepherd Type)	<i>LAMB3</i>	WT/WT	Normal (Clear)
Multidrug Resistance 1	<i>ABCB1</i>	WT/WT	Normal (Clear)
Multifocal Retinopathy 1	<i>BEST1</i>	WT/M	Carrier
Neuronal Ceroid Lipofuscinosis 6	<i>CLN6</i>	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 8 (Australian Shepherd Type)	<i>CLN8</i>	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration	<i>PRCD</i>	WT/WT	Normal (Clear)
Von Willebrand Disease I	<i>VWF</i>	WT/WT	Normal (Clear)

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Coat Color and Trait Certificate

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

Coat Color/Trait Test	Gene	Genotype	Interpretation
Chondrodysplasia (CDPA)	<i>CFA18 FGF4</i>	cd/cd	No Leg Shortening Associated with CDPA

Interpretation:

Two genetic mutations are associated with shortened legs in dogs. Both mutations consist of copied sections (duplication) of the canine *FGF4* gene (called an *FGF4*-retrogene) that have been inserted into two aberrant locations in the genome; one in chromosome 12 (*CFA12 FGF4*; associated with CDDY and IVDD risk) and one in chromosome 18 (*CFA18 FGF4*; associated with chondrodysplasia [CDPA], but not associated with IVDD). Appropriate breeding decisions regarding dogs which have inherited the *CFA12 FGF4* mutation (WT/M or M/M) need to address both the potential loss of genetic diversity in a population which would occur if dogs with this mutation were prohibited from breeding as well as the loss of the short-legged appearance that is a defining physical characteristic for some breeds. In breeds which inherit both mutations, breeders may use genetic testing results to selectively breed for the CDPA (*CFA18 FGF4*) mutation while breeding away from the CDDY and IVDD risk (*CFA12 FGF4*) mutation to reduce IVDD risk and retain the short-legged appearance. However, the frequency of each mutation varies between breeds and, in some cases, may not be conducive to such a breeding strategy. For example, breeds with extreme limb shortening (e.g. Basset hound, Dachshund, Corgi) typically develop their appearance due to inheritance of both the *CFA12 FGF4* and *CFA18 FGF4* mutations. In addition, depending on the breed, offspring born without either the *CFA12 FGF4* or *CFA18 FGF4* mutations may display longer limbs than cohorts and, therefore, not meet specific breed standards.

This dog carries two copies of the **cd** allele which does not result in leg shortening. However, the actual leg length of the dog is a result of a combination of factors including the mutation associated with CDDY and IVDD risk (*CFA12 FGF4*) as well as variants in other genes. This dog will pass one copy of **cd** to 100% of its offspring.

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.

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