

DNA Test Report

Owner Info

First Name

Mari-Claire

Last Name

Watkins

Pet Info

Registered Name

Ponsonby's Frankie

Date of Birth

8/31/2021

Nickname (Call Name)

Ponsonby's Frankie

Sample ID

PRYMTTZ

Sex

Female

Registration

N/A

Microchip ID

N/A

Tattoo ID

N/A

DNA Test Report

Ancestry Results

Sporting

100% Golden Retriever

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Genetic Diversity (Heterozygosity)

Ponsonby's Frankie's Percentage of Heterozygosity

38%

Ponsonby's Frankie's genome analysis shows higher than average genetic heterozygosity when compared with other Golden Retrievers.

Typical Range for Golden Retrievers

28 - 37%

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Health Conditions Tested

Genetic Condition	Gene	Risk Variant	Copies	Result
2,8-dihydroxyadenine (DHA) Urolithiasis	APRT	G>A	0	Clear
Acral Mutilation Syndrome	GDNF	C>T	0	Clear
Acute Respiratory Distress Syndrome	ANLN	C>T	0	Clear
Alaskan Husky Encephalopathy	SLC19A3	G>A	0	Clear
Alexander Disease	GFAP	G>A	0	Clear
Amelogenesis Imperfecta	ENAM	Deletion	0	Clear
Bandera's Neonatal Ataxia	GRM1	Insertion	0	Clear
Benign Familial Juvenile Epilepsy	LGI2	A>T	0	Clear
Canine Leukocyte Adhesion Deficiency (CLAD), type III	FERMT3	Insertion	0	Clear
Canine Multifocal Retinopathy 1	BEST1	C>T	0	Clear
Canine Multifocal Retinopathy 2	BEST1	G>A	0	Clear
Canine Multifocal Retinopathy 3	BEST1	Deletion	0	Clear
Canine Scott Syndrome	ANO6	G>A	0	Clear
Centronuclear Myopathy (Discovered in the Great Dane)	BIN1	A>G	0	Clear
Centronuclear Myopathy (Discovered in the Labrador Retriever)	PTPLA	Insertion	0	Clear
Cerebellar Ataxia	RAB24	A>C	0	Clear
Cerebellar Cortical Degeneration	SNX14	C>T	0	Clear
Cerebellar Hypoplasia	VLDLR	Deletion	0	Clear
Cerebral Dysfunction	SLC6A3	G>A	0	Clear
Chondrodysplasia	ITGA10	C>T	0	Clear
Cleft Lip & Palate with Syndactyly	ADAMTS20	Deletion	0	Clear
Cleft Palate	DLX6	C>A	0	Clear

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Health Conditions Tested (continued)

Genetic Condition	Gene	Risk Variant	Copies	Result
Complement 3 Deficiency	C3	Deletion	0	Clear
Cone Degeneration (Discovered in the Alaskan Malamute)	CNGB3	Deletion	0	Clear
Cone Degeneration (Discovered in the German Shepherd Dog)	CNGA3	C>T	0	Clear
Cone Degeneration (Discovered in the German Shorthaired Pointer)	CNGB3	G>A	0	Clear
Cone-Rod Dystrophy	NPHP4	Deletion	0	Clear
Cone-Rod Dystrophy 1	PDE6B	Deletion	0	Clear
Cone-Rod Dystrophy 2	IQCB1	Insertion	0	Clear
Congenital Dyshormonogenic Hypothyroidism with Goiter (Discovered in the Shih Tzu)	SLC5A5	G>A	0	Clear
Congenital Hypothyroidism (Discovered in the Tenterfield Terrier)	TPO	C>T	0	Clear
Congenital Hypothyroidism (Discovered in the Toy Fox and Rat Terrier)	TPO	C>T	0	Clear
Congenital Myasthenic Syndrome (Discovered in the Golden Retriever)	COLQ	G>A	0	Clear
Congenital Myasthenic Syndrome (Discovered in the Jack Russell Terrier)	CHRNE	Insertion	0	Clear
Congenital Myasthenic Syndrome (Discovered in the Labrador Retriever)	COLQ	T>C	0	Clear
Congenital Myasthenic Syndrome (Discovered in the Old Danish Pointer)	CHAT	G>A	0	Clear
Congenital Stationary Night Blindness (CSNB)	RPE65	A>T	0	Clear
Craniofacial Osteopathy	SLC37A2	C>T	0	Clear
Cystic Renal Dysplasia and Hepatic Fibrosis	INPP5E	G>A	0	Clear
Cystinuria Type I-A	SLC3A1	C>T	0	Clear
Cystinuria Type II-A	SLC3A1	Deletion	0	Clear
Deafness and Vestibular Dysfunction (Discovered in Doberman Pinscher)	PTPRQ	Insertion	0	Clear
Degenerative Myelopathy	SOD1	G>A	0	Clear
Demyelinating Neuropathy	SBF2	G>T	0	Clear

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Health Conditions Tested (continued)

Genetic Condition	Gene	Risk Variant	Copies	Result
Dental Hypomineralization	FAM20C	C>T	0	Clear
Dilated Cardiomyopathy (Discovered in the Schnauzer)	RBM20	Deletion	0	Clear
Dominant Progressive Retinal Atrophy	RHO	C>G	0	Clear
Dystrophic Epidermolysis Bullosa (Discovered in the Central Asian Ovcharka)	COL7A1	C>T	0	Clear
Dystrophic Epidermolysis Bullosa (Discovered in the Golden Retriever)	COL7A1	C>T	0	Clear
Early Retinal Degeneration (Discovered in the Norwegian Elkhound)	STK38L	Insertion	0	Clear
Early-onset PRA (Discovered in the Portuguese Water Dog)	CCDC66	Insertion	0	Clear
Early-Onset Progressive Polyneuropathy (Discovered in the Alaskan Malamute)	NDRG1	G>T	0	Clear
Early-Onset Progressive Polyneuropathy (Discovered in the Greyhound)	NDRG1	Deletion	0	Clear
Enamel Hypoplasia (Discovered in the Parson Russell Terrier)	ENAM	C>T	0	Clear
Epidermolytic Hyperkeratosis	KRT10	G>T	0	Clear
Episodic Falling Syndrome	BCAN	Insertion	0	Clear
Exercise-Induced Collapse	DNM1	G>T	0	Clear
Factor VII Deficiency	F7	G>A	0	Clear
Factor XI Deficiency	FXI	Insertion	0	Clear
Fanconi Syndrome	FAN1	Deletion	0	Clear
Fetal Onset Neuroaxonal Dystrophy	MFN2	G>C	0	Clear
Focal Non-Epidermolytic Palmoplantar Keratoderma	KRT16	G>C	0	Clear
Generalized Progressive Retinal Atrophy (Discovered in the Schapendoes)	CCDC66	Insertion	0	Clear
Glanzmann Thrombasthenia Type I	ITGA2B	C>T	0	Clear
Glanzmann Thrombasthenia Type I (Discovered in Great Pyrenees)	ITGA2B	C>G	0	Clear
Globoid Cell Leukodystrophy (Discovered in Terriers)	GALC	A>C	0	Clear

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Health Conditions Tested (continued)

Genetic Condition	Gene	Risk Variant	Copies	Result
Globoid Cell Leukodystrophy (Discovered in the Irish Setter)	GALC	A>T	0	Clear
Glycogen Storage Disease Type Ia	G6PC	G>C	0	Clear
Glycogen Storage Disease Type IIIa, (GSD IIIa)	AGL	Deletion	0	Clear
GM1 Gangliosidosis (Discovered in the Portuguese Water Dog)	GLB1	G>A	0	Clear
GM1 Gangliosidosis (Discovered in the Shiba)	GLB1	Deletion	0	Clear
GM2 Gangliosidosis (Discovered in the Japanese Chin)	HEXA	G>A	0	Clear
GM2 Gangliosidosis (Discovered in the Toy Poodle)	HEXB	Deletion	0	Clear
Hemophilia A (Discovered in Old English Sheepdog)	FVIII	C>T	0	Clear
Hemophilia A (Discovered in the Boxer)	FVIII	C>G	0	Clear
Hemophilia A (Discovered in the German Shepherd Dog - Variant 1)	FVIII	G>A	0	Clear
Hemophilia A (Discovered in the German Shepherd Dog - Variant 2)	FVIII	G>A	0	Clear
Hemophilia A (Discovered in the Havanese)	FVIII	Insertion	0	Clear
Hemophilia B	FIX	G>A	0	Clear
Hemophilia B (Discovered in the Airedale Terrier)	FIX	Insertion	0	Clear
Hemophilia B (Discovered in the Lhasa Apso)	FIX	Deletion	0	Clear
Hereditary Ataxia (Discovered in the Norwegian Buhund)	KCNIP4	T>C	0	Clear
Hereditary Elliptocytosis	SPTB	C>T	0	Clear
Hereditary Footpad Hyperkeratosis	FAM83G	G>C	0	Clear
Hereditary Nasal Parakeratosis (Discovered in the Greyhound)	SUV39H2	Deletion	0	Clear
Hereditary Nasal Parakeratosis (Discovered in the Labrador Retriever)	SUV39H2	A>C	0	Clear
Hereditary Vitamin D-Resistant Rickets Type II	VDR	Deletion	0	Clear
Hyperekplexia or Startle Disease	SLC6A5	G>T	0	Clear

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Health Conditions Tested (continued)

Genetic Condition	Gene	Risk Variant	Copies	Result
Hyperuricosuria	SLC2A9	G>T	0	Clear
Hypocatalasia	CAT	G>A	0	Clear
Hypomyelination	FNIP2	Deletion	0	Clear
Hypophosphatasia	Pending	T>G	0	Clear
Ichthyosis (Discovered in the American Bulldog)	NIPAL4	Deletion	0	Clear
Ichthyosis (Discovered in the Great Dane)	SLC27A4	G>A	0	Clear
Intestinal Cobalamin Malabsorption (Discovered in the Beagle)	CUBN	Deletion	0	Clear
Intestinal Cobalamin Malabsorption (Discovered in the Border Collie)	CUBN	Deletion	0	Clear
Intestinal Cobalamin Malabsorption (Discovered in the Komondor)	CUBN	G>A	0	Clear
Juvenile Encephalopathy (Discovered in the Parson Russell Terrier)	Pending	Deletion	0	Clear
Juvenile Laryngeal Paralysis and Polyneuropathy	RAB3GAP1	Deletion	0	Clear
Juvenile Myoclonic Epilepsy	DIRAS1	Deletion	0	Clear
L-2-Hydroxyglutaric Aciduria	L2HGDH	T>C	0	Clear
L-2-Hydroxyglutaric Aciduria (Discovered in the Westie)	Pending	Insertion	0	Clear
Lagotto Storage Disease	ATG4D	G>A	0	Clear
Lamellar Ichthyosis	TGM1	Insertion	0	Clear
Lethal Acrodermatitis (Discovered in the Bull Terrier)	MKLN1	A>C	0	Clear
Ligneous Membranitis	PLG	T>A	0	Clear
Lung Developmental Disease (Discovered in the Airedale Terrier)	LAMP3	C>T	0	Clear
Macrothrombocytopenia	TUBB1	G>A	0	Clear
May-Hegglin Anomaly	MYH9	G>A	0	Clear
MDR1 Medication Sensitivity	MDR1/ABCB1	Deletion	0	Clear

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Health Conditions Tested (continued)

Genetic Condition	Gene	Risk Variant	Copies	Result
Microphthalmia (Discovered in the Soft-Coated Wheaten Terrier)	RBP4	Deletion	0	Clear
Mucopolysaccharidosis Type IIIA (Discovered in the Dachshund)	SGSH	C>A	0	Clear
Mucopolysaccharidosis Type IIIA (Discovered in the New Zealand Huntaway)	SGSH	Insertion	0	Clear
Mucopolysaccharidosis Type VII (Discovered in the Brazilian Terrier)	GUSB	C>T	0	Clear
Mucopolysaccharidosis Type VII (Discovered in the German Shepherd Dog)	GUSB	G>A	0	Clear
Muscular Dystrophy (Discovered in the Cavalier King Charles Spaniel)	Dystrophin	G>T	0	Clear
Muscular Dystrophy (Discovered in the Golden Retriever)	Dystrophin	A>G	0	Clear
Muscular Dystrophy (Discovered in the Landseer)	COL6A1	G>T	0	Clear
Muscular Dystrophy (Discovered in the Norfolk Terrier)	Dystrophin	Deletion	0	Clear
Muscular Hypertrophy (Double Muscling)	MSTN	T>A	0	Clear
Musladin-Lueke Syndrome	ADAMTSL2	C>T	0	Clear
Myeloperoxidase Deficiency	MOP	C>T	0	Clear
Myotonia Congenita	CLCN1	Insertion	0	Clear
Myotonia Congenita (Discovered in the Labrador Retriever)	CLCN1	T>A	0	Clear
Myotonia Congenita (Discovered in the Miniature Schnauzer)	CLCN1	C>T	0	Clear
Myotubular Myopathy	MTM1	A>C	0	Clear
Narcolepsy (Discovered in the Dachshund)	HCRTR2	G>A	0	Clear
Narcolepsy (Discovered in the Labrador Retriever)	HCRTR2	G>A	0	Clear
Nemaline Myopathy	NEB	C>A	0	Clear
Neonatal Cerebellar Cortical Degeneration	SPTBN2	Deletion	0	Clear
Neonatal Encephalopathy with Seizures	ATF2	T>G	0	Clear
Neuroaxonal Dystrophy	TECPR2	C>T	0	Clear

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Health Conditions Tested (continued)

Genetic Condition	Gene	Risk Variant	Copies	Result
Neuroaxonal Dystrophy (Discovered in the Papillon)	PLA2G6	G>A	0	Clear
Neuroaxonal Dystrophy (Discovered in the Rottweiler)	VPS11	A>G	0	Clear
Neuronal Ceroid Lipofuscinosis 1	PPT1	Insertion	0	Clear
Neuronal Ceroid Lipofuscinosis 12 (Discovered in the Australian Cattle Dog)	ATP13A2	C>T	0	Clear
Neuronal Ceroid Lipofuscinosis 7	MFSD8	Deletion	0	Clear
Neuronal Ceroid Lipofuscinosis 8 (Discovered in the Alpine Dachsbracke)	CLN8	Deletion	0	Clear
Neuronal Ceroid Lipofuscinosis 8 (Discovered in the Australian Shepherd)	CLN8	G>A	0	Clear
Neuronal Ceroid Lipofuscinosis 8 (Discovered in the English Setter)	CLN8	T>C	0	Clear
Neuronal Ceroid Lipofuscinosis 8 (Discovered in the Saluki)	CLN8	Insertion	0	Clear
Obesity risk (POMC)	POMC	Deletion	0	Clear
Osteochondrodysplasia	SLC13A1	Deletion	0	Clear
Osteochondromatosis (Discovered in the American Staffordshire Terrier)	EXT2	C>A	0	Clear
Osteogenesis Imperfecta (Discovered in the Beagle)	COL1A2	C>T	0	Clear
Osteogenesis Imperfecta (Discovered in the Dachshund)	SERPINH1	T>C	0	Clear
P2RY12-associated Bleeding Disorder	P2RY12	Deletion	0	Clear
Paroxysmal Dyskinesia	PIGN	C>T	0	Clear
Persistent Müllerian Duct Syndrome	AMHR2	C>T	0	Clear
Phosphofruktokinase Deficiency	PFKM	G>A	0	Clear
Polycystic Kidney Disease	PKD1	G>A	0	Clear
Prekallikrein Deficiency	KLKB1	T>A	0	Clear
Primary Ciliary Dyskinesia	CCDC39	C>T	0	Clear
Primary Ciliary Dyskinesia (Discovered in the Alaskan Malamute)	NME5	Deletion	0	Clear

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Health Conditions Tested (continued)

Genetic Condition	Gene	Risk Variant	Copies	Result
Primary Lens Luxation	ADAMTS17	G>A	0	Clear
Primary Open Angle Glaucoma (Discovered in Basset Fauve de Bretagne)	ADAMTS17	G>A	0	Clear
Primary Open Angle Glaucoma (Discovered in Petit Basset Griffon Vendeen)	ADAMTS17	Insertion	0	Clear
Primary Open Angle Glaucoma and Lens Luxation (Discovered in Chinese Shar-Pei)	ADAMTS17	Deletion	0	Clear
Progressive Early-Onset Cerebellar Ataxia	SEL1L	T>C	0	Clear
Progressive Retinal Atrophy (Discovered in the Basenji)	SAG	T>C	0	Clear
Progressive Retinal Atrophy (Discovered in the Golden Retriever - GR-PRA1 variant)	SLC4A3	Insertion	0	Clear
Progressive Retinal Atrophy (Discovered in the Lhasa Apso)	IMPG2	Insertion	0	Clear
Progressive Retinal Atrophy (Discovered in the Papillon and Phalène)	CNGB1	Deletion	0	Clear
Progressive Retinal Atrophy (Discovered in the Shetland Sheepdog - BBS2 variant)	Pending	G>C	0	Clear
Progressive Retinal Atrophy (Discovered in the Shetland Sheepdog - CNGA1 variant)	CNGA1	Deletion	0	Clear
Progressive Retinal Atrophy (Discovered in the Swedish Vallhund)	MERTK	Insertion	0	Clear
Progressive Retinal Atrophy 1 (Discovered in the Italian Greyhound)	Pending	G>A	0	Clear
Progressive Retinal Atrophy Type III	FAM161A	Insertion	0	Clear
Protein Losing Nephropathy	NPHS1	G>A	0	Clear
Pyruvate Dehydrogenase Phosphatase 1 Deficiency	PDP1	C>T	0	Clear
Pyruvate Kinase Deficiency (Discovered in the Basenji)	PKLR	Deletion	0	Clear
Pyruvate Kinase Deficiency (Discovered in the Beagle)	PKLR	G>A	0	Clear
Pyruvate Kinase Deficiency (Discovered in the Pug)	PKLR	T>C	0	Clear
Pyruvate Kinase Deficiency (Discovered in the West Highland White Terrier)	PKLR	Insertion	0	Clear
QT Syndrome	KCNQ1	C>A	0	Clear

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Health Conditions Tested (continued)

Genetic Condition	Gene	Risk Variant	Copies	Result
Renal Cystadenocarcinoma and Nodular Dermatofibrosis	FLCN	A>G	0	Clear
Rod-Cone Dysplasia 1	PDE6B	G>A	0	Clear
Rod-Cone Dysplasia 1a	PDE6B	Insertion	0	Clear
Rod-Cone Dysplasia 3	PDE6A	Deletion	0	Clear
Sensory Ataxic Neuropathy	tRNATyr	Deletion	0	Clear
Sensory Neuropathy	FAM134B	Insertion	0	Clear
Severe Combined Immunodeficiency	PRKDC	G>T	0	Clear
Severe Combined Immunodeficiency (Discovered in Frisian Water Dogs)	RAG1	G>T	0	Clear
Shaking Puppy Syndrome (Discovered in the Border Terrier)	Pending	G>A	0	Clear
Skeletal Dysplasia 2	COL11A2	G>C	0	Clear
Spinocerebellar Ataxia (Late-Onset Ataxia)	CAPN1	G>A	0	Clear
Spinocerebellar Ataxia with Myokymia and/or Seizures	KCNJ10	C>G	0	Clear
Spondylocostal Dysostosis	HES7	Deletion	0	Clear
Spongy Degeneration with Cerebellar Ataxia (Discovered in Belgian Malinois - SDCA1)	KCNJ10	T>C	0	Clear
Spongy Degeneration with Cerebellar Ataxia (Discovered in Belgian Malinois - SDCA2)	ATP1B2	Insertion	0	Clear
Stargardt Disease (Discovered in the Labrador Retriever)	ABCA4	Insertion	0	Clear
Trapped Neutrophil Syndrome	VPS13B	Deletion	0	Clear
Van den Ende-Gupta Syndrome	SCARF2	Deletion	0	Clear
von Willebrand's Disease, type 1	VWF	G>A	0	Clear
von Willebrand's Disease, type 2	VWF	T>G	0	Clear
von Willebrand's Disease, type 3 (Discovered in the Kooiker Hound)	VWF	G>A	0	Clear

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Health Conditions Tested (continued)

Genetic Condition	Gene	Risk Variant	Copies	Result
von Willebrand's Disease, type 3 (Discovered in the Scottish Terrier)	VWF	Deletion	0	Clear
von Willebrand's Disease, type 3 (Discovered in the Shetland Sheepdog)	VWF	Deletion	0	Clear
X-Linked Ectodermal Dysplasia	EDA	G>A	0	Clear
X-Linked Hereditary Nephropathy (Discovered in the Navasota Dog)	COL4A5	Deletion	0	Clear
X-Linked Hereditary Nephropathy (Discovered in the Samoyed)	COL4A5	G>T	0	Clear
X-Linked Myotubular Myopathy	MTM1	C>A	0	Clear
X-Linked Progressive Retinal Atrophy 1	RPGR	Deletion	0	Clear
X-Linked Progressive Retinal Atrophy 2	RPGR	Deletion	0	Clear
X-Linked Severe Combined Immunodeficiency (Discovered in the Basset Hound)	IL2RG	Deletion	0	Clear
X-Linked Severe Combined Immunodeficiency (Discovered in the Cardigan Welsh Corgi)	IL2RG	Insertion	0	Clear
X-Linked Tremors	PLP1	A>C	0	Clear
Xanthinuria (Discovered in a mixed breed dog)	Pending	G>A	0	Clear
Xanthinuria (Discovered in the Cavalier King Charles Spaniel)	Pending	Deletion	0	Clear
Xanthinuria (Discovered in the Toy Manchester Terrier)	Pending	G>T	0	Clear

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Coat Color

Genetic Trait	Gene	Variant	Copies	Result
Fawn	ASIP	a ^y	0	No effect
Recessive Black	ASIP	a	0	No effect
Tan Points	ASIP	a ^t	2	Tan points possible
Dominant Black	CBD103	K ^B	2	Black possible
Mask	MC1R	E ^m	0	No effect
Recessive Red (Variant 1)	MC1R	e ¹	2	Cream to red coat
Widow's Peak (Discovered in the Afghan Hound and Saluki)	MC1R	E ^G	0	No effect
Chocolate (Variant 1)	TYRP1	b ^c	0	No effect
Chocolate (Variant 2)	TYRP1	b ^s	0	No effect

Coat Patterns

Genetic Trait	Gene	Variant	Copies	Result
Piebald	MITF	s ^p	0	No effect
Merle	PMEL	M	0	No effect
Harlequin	PSMB7	H	0	No effect
Saddle Tan	RALY	-	1	Saddle possible

Coat Length and Curl

Genetic Trait	Gene	Variant	Copies	Result
Long Hair (Variant 1)	FGF5	lh ¹	2	Long coat
Curly Coat	KRT71	C	0	No effect

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Hairlessness

Genetic Trait	Gene	Variant	Copies	Result
Hairlessness (Discovered in the Chinese Crested Dog) Linkage test	FOXI3	Hr ^{cc}	0	No effect
Hairlessness (Discovered in the American Hairless Terrier)	SGK3	hr ^{ah}	0	No effect
Hairlessness (Discovered in the Scottish Deerhound)	SKG3	hr ^{sd}	0	No effect

Shedding

Genetic Trait	Gene	Variant	Copies	Result
Reduced Shedding	MC5R	sd	1	Occasional shedder

More Coat Traits

Genetic Trait	Gene	Variant	Copies	Result
Hair Ridge	FGF3, FGF4, FGF19, ORAOV1	R	0	No effect
Furnishings	RSPO2	F	0	No effect
Albino	SLC45A2	c ^{al}	0	No effect

Head Shape

Genetic Trait	Gene	Variant	Copies	Result
Short Snout (Variant 2)	BMP3	-	0	No effect
Short Snout (Variant 1)	SMOC2	-	0	No effect

Eye Color

Genetic Trait	Gene	Variant	Copies	Result
Blue Eyes	ALX4	-	0	No effect

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Ears

Genetic Trait	Gene	Variant	Copies	Result
Floppy Ears	MSRB3	-	2	Floppy ears more likely

Extra Toes

Genetic Trait	Gene	Variant	Copies	Result
Hind Dewclaws (Discovered in Asian breeds)	LMBR1	DC-1	0	No effect
Hind Dewclaws (Discovered in Western breeds)	LMBR1	DC-2	0	No effect

More Body Features

Genetic Trait	Gene	Variant	Copies	Result
Back Muscle and Bulk	ACSL4	-	0	No effect
High Altitude Adaptation	EPAS1	-	0	No effect
Short Legs	FGF4	-	0	Medium to long legs
Short Tail	T-box	T	0	Full tail length likely