

**Call Name:** Jamie

**Breed:** American Cocker Spaniel

**Gender:** Female

**Owner:** Gary Sinclair

**Country:** United States

**Testing date:** 2020/1/23

## Test results - Known disorders in the breed

Disorder	Type	Mode of Inheritance	Result
Degenerative Myelopathy, (DM; SOD1A)	Neurological Disorders	Autosomal Recessive (Incomplete Penetrance)	Clear
Exercise-Induced Collapse, (EIC)	Neuromuscular Disorders	Autosomal Recessive (Incomplete Penetrance)	Clear
Hereditary Phosphofructokinase (PFK) Deficiency	Blood Disorders	Autosomal Recessive	Clear

## Test results - New potential disorders in the breed

Disorder	Type	Mode of Inheritance	Result
Skeletal Dysplasia 2, (SD2)	Skeletal Disorders	Autosomal Recessive	Clear

On behalf of Genoscoper Laboratories,

  
SIGNATURE

Jonas Donner, PhD, Head of Research and Development  
at Genoscoper Laboratories

# Optimal Selection™

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Laboratories

BR10 859

Jamie, American Cocker Spaniel

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## Test results for pharmacogenetics

Disorder	Mode of Inheritance	Result
Multi-Drug Resistance 1, (MDR1) or Ivermectin Sensitivity	Autosomal Dominant	Clear

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## Test results - Traits - page 1

### Coat Type

Trait	Genotype	Description
Coat Length	I/I	The dog is genetically long-haired.
Furnishings / Improper Coat in Portuguese Water Dogs (marker test)	GG/CC	The dog is not genetically likely to express furnishings.
<i>KRT71</i> c.451C>T (p.Arg151Trp)	C/C	The dog does not carry any copies of the tested allele causing curly coat. The dog most likely has non-curly hair.
<i>MC5R</i> c.237A>T	T/T	The dog has two copies of the allele associated with low shedding. The dog is likely average or low shedder.
<i>SGK3</i> (p.Val96Glyfs)	I/I	The dog does not carry the tested hairlessness allele of the American Hairless Terrier.
<i>SGK3</i> c.137_138insT (p.Glu47Glyfs)	D/D	The dog does not carry the tested hairlessness allele of the Scottish Deerhound.

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## Test results - Traits - page 2

### Coat Color

Trait	Genotype	Description
Color Locus E - Extensions	e/e	The dog has recessive red coat color.
Color Locus B - Brown	B/B	The dog is not likely to have brown pigment.
Color Locus K - Dominant Black	KB/ky    KB/kbr    kbr/ky    kbr/kbr	The dog is genetically dominant black or brindle.
Color Locus A - Agouti	at/at	The dog has genetically tan points or saddle tan pattern.
Color Locus S - Piebald or extreme white spotting	S/S	The dog is likely to have solid coat color with minimal white.
Color Locus H - Harlequin	h/h	The dog doesn't have harlequin pattern.
Dilution (d <sup>2</sup> allele)	G/G	The dog does not carry any copies of the rare d <sup>2</sup> allele associated with dilution in Chow Chow, Sloughi and Thai Ridgeback.
Merle (M allele)	m/m	The dog is genetically non-merle and does not carry a <i>SILV</i> gene SINE insertion.
Saddle Tan ( <i>RALY</i> gene dupl.)	-/-	The dog may have saddle tan pattern if it has also tan point genotype at the A locus.
Albinism (caL-allele)	C/C	The dog does not carry the tested mutation for albinism.

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## Test results - Traits - page 3

### Body Size

Trait	Genotype	Description
<i>IGF1</i> (chr15:41221438)	A/A	The dog is homozygous for the derived allele typically associated with small body mass.
<i>IGF1R</i> c.611G>A (p.Arg204His)	G/G	The dog carries two ancestral alleles typically found in larger-sized breeds.
<i>ACSL4</i> chrX:82919525C>T	C/C	The dog doesn't have the allele associated with large skeletal size and heavy muscling with considerable back fat thickness.
<i>IGSF1</i> p.Asp768Glu	A/C	The dog has one copy of the allele associated with heavy muscling.
<i>IRS4</i> chrX:82296039	G/G	The dog doesn't have the allele associated with large body size.
<i>FGF4</i> insertion	D/D	The dog is homozygous for the ancient allele. The dog is likely to have legs of normal length.
<i>STC2</i> (chr4:39182836)	A/T	The dog carries one copy of the allele associated with reduced body size and one copy of the allele associated with no size reduction.
Body size, GHR1 gene variant E191K	A/G	The dog carries one ancestral allele and one derived allele.
<i>GHR2</i> (p.Pro177Leu)	C/C	The dog has two copies of the ancestral allele associated with larger body size.
<i>HMGA2</i>	G/G	The dog has two copies of the ancestral allele associated with larger body size.

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## Test results - Traits - page 4

### Morphology

Trait	Genotype	Description
<i>BMP3</i> c.1344C>A (p.Phe448Leu)	C/C	The dog does not carry the tested allele typically associated with shortened head (brachycephaly). The dog is more likely to have an elongated head (dolichocephaly).
chr10:11072007	C/C	The dog carries two copies of an allele typically associated with floppy ears. The dog is more likely to have floppy than pricked ears.
<i>T</i> c.189C>G (p.Ile63Met)	C/C	The dog does not carry the tested bobtail-causing genetic variant. The dog is most likely long-tailed.
<i>EPAS1</i> (p.Gly305Ser)	G/G	The dog does not carry the tested variant associated with adaptation to high altitudes.
<i>LIMBR1</i> DC-1	G/G	The dog does not carry the tested allele associated with hind dewclaws in Asian breeds. The dog is not likely to have hind dewclaws.
<i>LIMBR1</i> DC-2	G/G	The dog does not carry the tested allele associated with hind dewclaws in western breeds. The dog is likely not to have hind dewclaws.
<i>AXL4</i>	D/D	The dog does not have the tested allele typically associated with blue eyes in Siberian Huskies. The dog is likely to have brown eyes.

On behalf of Genoscoper Laboratories,



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## Test results - Additional disorders found in other breeds - page 1

### Blood Disorders - page 1

Disorder	Mode of Inheritance	Result
Bleeding disorder due to P2RY12 defect	Autosomal Recessive	Clear
Canine Cyclic Neutropenia, Cyclic Hematopoiesis, Grey Collie Syndrome, (CN)	Autosomal Recessive	Clear
Canine Leukocyte Adhesion Deficiency (CLAD), type III	Autosomal Recessive	Clear
Canine Scott Syndrome, (CSS)	Autosomal Recessive	Clear
Factor IX Deficiency or Hemophilia B; mutation Gly379Glu	X-linked Recessive	Clear
Factor IX Deficiency or Hemophilia B; mutation originally found in Airedale Terrier	X-linked Recessive	Clear
Factor IX Deficiency or Hemophilia B; mutation originally found in Lhasa Apso	X-linked Recessive	Clear
Factor VII Deficiency	Autosomal Recessive	Clear
Factor VIII Deficiency or Hemophilia A; mutation originally found in Boxer	X-linked Recessive	Clear
Factor VIII Deficiency or Hemophilia A; mutation originally found in German Shepherd Dog	X-linked Recessive	Clear
Factor VIII Deficiency or Hemophilia A; mutation originally found in Havanese	X-linked Recessive	Clear
Factor VIII Deficiency or Hemophilia A; mutation originally found in Old English Sheepdog	X-linked Recessive	Clear
Factor VIII Deficiency or Hemophilia A; p.Cys548Tyr mutation originally found in German Shepherd	X-linked Recessive	Clear
Factor XI Deficiency	Autosomal Dominant (Incomplete Penetrance)	Clear
Familial Congenital Methemoglobinemia; mutation originally found in Pomeranian	Autosomal Recessive	Clear
Glanzmann Thrombasthenia Type I, (GT); mutation originally found in Pyrenean Mountain Dog	Autosomal Recessive	Clear
Glanzmann Thrombasthenia Type I, (GT); mutation originally found in mixed breed dogs	Autosomal Recessive	Clear
Hereditary Elliptocytosis		Clear
Macrothrombocytopenia; disease-linked variant originally found in Norfolk and Cairn Terrier	Autosomal Recessive	Clear
May-Hegglin Anomaly (MHA)	Autosomal Dominant	Clear
Prekallikrein Deficiency	Autosomal Recessive	Clear

## Test results - Additional disorders found in other breeds - page 2

### Blood Disorders - page 2

Disorder	Mode of Inheritance	Result
Pyruvate Kinase Deficiency; mutation originally found in Basenji	Autosomal Recessive	Clear
Pyruvate Kinase Deficiency; mutation originally found in Beagle	Autosomal Recessive	Clear
Pyruvate Kinase Deficiency; mutation originally found in Pug	Autosomal Recessive	Clear
Pyruvate Kinase Deficiency; mutation originally found in West Highland White Terrier	Autosomal Recessive	Clear
Trapped Neutrophil Syndrome, (TNS)	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 1	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 2	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 3; mutation originally found in Kooikerhondje	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 3; mutation originally found in Scottish Terrier	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 3; mutation originally found in Shetland Sheepdog	Autosomal Recessive	Clear



## Test results - Additional disorders found in other breeds - page 3

### Ocular Disorders - page 1

Disorder	Mode of Inheritance	Result
Canine Multifocal Retinopathy 1, (CMR1); mutation originally found in Mastiff-related breeds	Autosomal Recessive	Clear
Canine Multifocal Retinopathy 2, (CMR2); mutation originally found in Coton de Tulear	Autosomal Recessive	Clear
Canine Multifocal Retinopathy 3, (CMR3); mutation originally found in Lapponian Herder	Autosomal Recessive	Clear
Cone Degeneration, (CD) or Achromatopsia; mutation originally found in Alaskan Malamute	Autosomal Recessive	Clear
Cone Degeneration, (CD) or Achromatopsia; mutation originally found in German Shepherd Dog	Autosomal Recessive	Clear
Cone Degeneration, (CD) or Achromatopsia; mutation originally found in German Shorthaired Pointer	Autosomal Recessive	Clear
Cone-Rod Dystrophy 1, (crd1); mutation originally found in American Staffordshire Terrier	Autosomal Recessive	Clear
Cone-Rod Dystrophy 2, (crd2); mutation originally found in American Pit Bull Terrier	Autosomal Recessive	Clear
Cone-Rod Dystrophy, (cord1-PRA / crd4)	Autosomal Recessive (Incomplete Penetrance)	Clear
Cone-Rod Dystrophy, Standard Wirehaired Dachshund, (crd SWD)	Autosomal Recessive	Clear
Congenital Eye Disease; mutation originally found in Irish Soft-Coated Wheaten Terrier	Autosomal Recessive	Clear
Dominant Progressive Retinal Atrophy, (DPRA)	Autosomal Dominant	Clear
Early Onset PRA (EOPRA); mutation originally found in Portuguese Water Dog	Autosomal Recessive	Clear
Early Retinal Degeneration, (erd); mutation originally found in Norwegian Elkhound	Autosomal Recessive	Clear
Generalized Progressive Retinal Atrophy	Autosomal Recessive	Clear
Golden Retriever Progressive Retinal Atrophy 1, (GR_PRA 1)	Autosomal Recessive	Clear
Goniodysgenesis and glaucoma; mutation originally found in Border Collie	Autosomal Recessive	Clear
Italian Greyhound Progressive Retinal Atrophy 1 (IG-PRA1)	Autosomal Recessive	Clear
Primary Hereditary Cataract, (PHC); mutation originally found in Australian Shepherd	Autosomal Dominant (Incomplete Penetrance)	Clear
Primary Lens Luxation, (PLL)	Autosomal Recessive	Clear
Primary Open Angle Glaucoma, (POAG); mutation originally found in Basset Fauve de Bretagne	Autosomal Recessive	Clear

## Test results - Additional disorders found in other breeds - page 4

### Ocular Disorders - page 2

Disorder	Mode of Inheritance	Result
Primary Open Angle Glaucoma, (POAG); mutation originally found in Beagle	Autosomal Recessive	Clear
Primary Open Angle Glaucoma, (POAG); mutation originally found in Norwegian Elkhound	Autosomal Recessive	Clear
Primary Open Angle Glaucoma, (POAG); mutation originally found in Petit Basset Griffon Vendéen	Autosomal Recessive	Clear
Primary lens luxation (PLL) and glaucoma; mutation originally found in Shar Pei	Autosomal Recessive	Clear
Progressive Retinal Atrophy (PRA4); mutation originally found in Lhasa Apso	Autosomal Recessive	Clear
Progressive Retinal Atrophy Type III, (PRA type III); mutation originally found in Tibetan Spaniel and Tibetan Terrier	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (CNGA1-PRA); mutation originally found in Shetland Sheepdog	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (PAP1_PRA); mutation originally found in Papillon and Phalene	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (PRA); mutation originally found in Basenji	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (PRA); mutation originally found in Swedish Vallhund	Autosomal Recessive	Clear
Rod-Cone Dysplasia 1, (rcd1); mutation originally found in Irish Setter	Autosomal Recessive	Clear
Rod-Cone Dysplasia 1a, (rcd1a); mutation originally found in Sloughi	Autosomal Recessive	Clear
Rod-Cone Dysplasia 3, (rcd3)	Autosomal Recessive	Clear
X-Linked Progressive Retinal Atrophy 1, (XLPR1)	X-linked Recessive	Clear
X-Linked Progressive Retinal Atrophy 2, (XLPR2; Type A PRA)	X-linked Recessive	Clear

### Cardiac Disorders

Disorder	Mode of Inheritance	Result
Dilated Cardiomyopathy, (DCM); mutation originally found in Schnauzer	Autosomal Recessive	Clear
QT Syndrome	Autosomal Dominant	Clear

## Test results - Additional disorders found in other breeds - page 5

### Endocrine Disorders

Disorder	Mode of Inheritance	Result
Congenital Dyshormonogenic Hypothyroidism with Goiter; mutation originally found in Shih Tzu	Autosomal Recessive	Clear
Congenital Hypothyroidism; mutation originally found in Tenterfield Terrier	Autosomal Recessive	Clear
Congenital Hypothyroidism; mutation originally found in Toy Fox and Rat Terrier	Autosomal Recessive	Clear

### Immunological Disorders

Disorder	Mode of Inheritance	Result
Autosomal Recessive Severe Combined Immunodeficiency, (ARSCID)	Autosomal Recessive	Clear
Complement 3 (C3) Deficiency	Autosomal Recessive	Clear
Myeloperoxidase Deficiency	Autosomal Recessive	Clear
Severe Combined Immunodeficiency in Frisian Water Dogs, (SCID)	Autosomal Recessive	Clear
X-Linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Basset Hound	X-linked Recessive	Clear
X-Linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Cardigan Welsh Corgi	X-linked Recessive	Clear

## Test results - Additional disorders found in other breeds - page 6

### Renal Disorders

Disorder	Mode of Inheritance	Result
2,8-Dihydroxyadenine (2,8-DHA) urolithiasis	Autosomal Recessive	Clear
Cystic Renal Dysplasia and Hepatic Fibrosis; mutation originally found in Norwich Terrier	Autosomal Recessive	Clear
Cystinuria Type I-A; mutation originally found in Newfoundland Dog	Autosomal Recessive	Clear
Cystinuria Type II-A; mutation originally found in Australian Cattle Dog	Autosomal Dominant	Clear
Fanconi Syndrome	Autosomal Recessive	Clear
Hyperuricosuria, (HUU)	Autosomal Recessive	Clear
Polycystic Kidney Disease in Bull Terriers, (BTPKD)	Autosomal Dominant	Clear
Primary Hyperoxaluria, (PH); mutation originally found in Coton de Tulear	Autosomal Recessive	Clear
Protein Losing Nephropathy, (PLN); NPHS1 gene variant		Clear
Renal Cystadenocarcinoma and Nodular Dermatofibrosis, (RCND)	Autosomal Dominant	Clear
X-Linked Hereditary Nephropathy, (XLHN)	X-linked Recessive	Clear
X-Linked Hereditary Nephropathy, (XLHN); mutation originally found in Navasota Dog	X-linked Recessive	Clear
Xanthinuria, Type 1a; mutation originally found in mixed breed dogs	Autosomal Recessive	Clear
Xanthinuria, Type 2a; mutation originally found in Toy Manchester Terrier	Autosomal Recessive	Clear
Xanthinuria, Type 2b; mutation originally found in Cavalier King Charles Spaniel and English Cocker Spaniel	Autosomal Recessive	Clear

## Test results - Additional disorders found in other breeds - page 7

### Metabolic Disorders

Disorder	Mode of Inheritance	Result
Glycogen Storage Disease Type II or Pompe's Disease, (GSD II)	Autosomal Recessive	Clear
Glycogen Storage Disease Type IIIa, (GSD IIIa)	Autosomal Recessive	Clear
Glycogen Storage Disease Type Ia, (GSD Ia)	Autosomal Recessive	Clear
Hypocatalasia or Acatalasemia	Autosomal Recessive	Clear
Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS); mutation originally found in Beagle	Autosomal Recessive	Clear
Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS); mutation originally found in Border Collie	Autosomal Recessive	Clear
Mucopolysaccharidosis Type IIIA, (MPS IIIA); mutation originally found in Dachshund	Autosomal Recessive	Clear
Mucopolysaccharidosis Type IIIA, (MPS IIIA); mutation originally found in New Zealand Huntaway	Autosomal Recessive	Clear
Mucopolysaccharidosis Type VII, (MPS VII); mutation originally found in Brazilian Terrier	Autosomal Recessive	Clear
Mucopolysaccharidosis Type VII, (MPS VII); mutation originally found in German Shepherd	Autosomal Recessive	Clear
Pyruvate Dehydrogenase Phosphatase 1 (PDP1) Deficiency	Autosomal Recessive	Clear

## Test results - Additional disorders found in other breeds - page 8

### Muscular Disorders

Disorder	Mode of Inheritance	Result
Cavalier King Charles Spaniel Muscular Dystrophy, (CKCS-MD)	X-linked Recessive	Clear
Centronuclear Myopathy, (CNM); mutation originally found in Great Dane	Autosomal Recessive	Clear
Centronuclear Myopathy, (CNM); mutation originally found in Labrador Retriever	Autosomal Recessive	Clear
Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Golden Retriever	X-linked Recessive	Clear
Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Norfolk Terrier	X-linked Recessive	Clear
Muscular Dystrophy, Ullrich-type; mutation originally found in Landseer	Autosomal Recessive	Clear
Muscular Hypertrophy (Double Muscling)	Autosomal Recessive	Clear
Myotonia Congenita; mutation originally found in Australian Cattle Dog	Autosomal Recessive	Clear
Myotubular Myopathy; mutation originally found in Rottweiler	X-linked Recessive	Clear
Nemaline Myopathy; mutation originally found in American Bulldog	Autosomal Recessive	Clear
X-Linked Myotubular Myopathy	X-linked Recessive	Clear

## Test results - Additional disorders found in other breeds - page 9

### Neurological Disorders - page 1

Disorder	Mode of Inheritance	Result
Acral Mutilation Syndrome, (AMS)	Autosomal Recessive	Clear
Alaskan Husky Encephalopathy, (AHE)	Autosomal Recessive	Clear
Alexander Disease (AxD); mutation originally found in Labrador Retriever	Autosomal Dominant	Clear
Bandera's Neonatal Ataxia, (BNAt)	Autosomal Recessive	Clear
Benign Familial Juvenile Epilepsy or Remitting Focal Epilepsy	Autosomal Recessive	Clear
Cerebellar Cortical Degeneration, (CCD); mutation originally found in Vizsla	Autosomal Recessive	Clear
Cerebral Dysfunction; mutation originally found in Friesian Stabyhoun	Autosomal Recessive	Clear
Dandy-Walker-Like Malformation (DWLM); mutation originally found in Eurasier	Autosomal Recessive	Clear
Early-Onset Progressive Polyneuropathy; mutation originally found in Alaskan Malamute	Autosomal Recessive	Clear
Fetal Onset Neuroaxonal Dystrophy, (FNAD)	Autosomal Recessive	Clear
Hereditary Ataxia or Cerebellar Ataxia; mutation originally found in Old English Sheepdog and Gordon Setter	Autosomal Recessive	Clear
Hereditary Ataxia; mutation originally found in in Norwegian Buhund	Autosomal Recessive	Clear
Hyperekplexia or Startle Disease	Autosomal Recessive	Clear
Hypomyelination; mutation originally found in Weimaraner	Autosomal Recessive	Clear
Juvenile Myoclonic Epilepsy, (JME); mutation originally found in Rhodesian Ridgeback	Autosomal Recessive	Clear
Juvenile encephalopathy; mutation originally found in Parson Russell Terrier	Autosomal Recessive	Clear
L-2-Hydroxyglutaric aciduria, (L2HGA); mutation originally found in Staffordshire Bull Terrier	Autosomal Recessive	Clear
L-2-Hydroxyglutaric aciduria, (L2HGA); mutation originally found in West Highland White Terrier	Autosomal Recessive	Clear
Lagotto Storage Disease, (LSD)	Autosomal Recessive	Clear
Neonatal Cerebellar Cortical Degeneration or Cerebellar Abiotrophy, (NCCD)	Autosomal Recessive	Clear
Neonatal Encephalopathy with Seizures, (NEWS)	Autosomal Recessive	Clear

## Test results - Additional disorders found in other breeds - page 10

### Neurological Disorders - page 2

Disorder	Mode of Inheritance	Result
Neuroaxonal Dystrophy (NAD); mutation originally found in Rottweiler	Autosomal Recessive	Clear
Neuroaxonal Dystrophy (NAD); mutation originally found in Spanish Water Dog	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 1, (NCL1); mutation originally found in Dachshund	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 10, (NCL10); mutation originally found in American Bulldog	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in Alpine Dachsbracke	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in Australian Shepherd	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in English Setter	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis, (NCL7); mutation originally found in Chinese Crested Dog and Chihuahua	Autosomal Recessive	Clear
Polyneuropathy with ocular abnormalities and neuronal vacuolation, (POANV); mutation originally found in Black Russian Terrier	Autosomal Recessive	Clear
Progressive Early-Onset Cerebellar Ataxia; mutation originally found in Finnish Hound	Autosomal Recessive	Clear
Sensory Neuropathy; mutation originally found in Border Collie	Autosomal Recessive	Clear
Shaking Puppy Spongiform LeucoEncephaloMyelopathy, (SLEM); mutation originally found in Border Terrier	Autosomal Recessive	Clear
Spinocerebellar Ataxia with Myokymia and/or Seizures (SCA)	Autosomal Recessive	Clear
Spinocerebellar Ataxia/ Late-Onset Ataxia (SCA, LOA)	Autosomal Recessive	Clear
Spongy Degeneration with Cerebellar Ataxia, (SDCA1); mutation originally found in Belgian Shepherd Dog	Autosomal Recessive	Clear
Spongy Degeneration with Cerebellar Ataxia, (SDCA2); mutation originally found in Belgian Shepherd Dog	Autosomal Recessive	Clear
X-Linked Tremors; mutation originally found in English Springer Spaniel	X-linked Recessive	Clear



## Test results - Additional disorders found in other breeds - page 11

### Neuromuscular Disorders

Disorder	Mode of Inheritance	Result
Congenital Myasthenic Syndrome (CMS); mutation originally found in Labrador Retriever	Autosomal Recessive	Clear
Congenital Myasthenic Syndrome, (CMS); mutation originally found in Jack Russell Terrier	Autosomal Recessive	Clear
Congenital Myasthenic Syndrome, (CMS); mutation originally found in Old Danish Pointing Dog	Autosomal Recessive	Clear
Episodic Falling, (EF)	Autosomal Recessive	Clear
GM1 Gangliosidosis; mutation originally found in Portuguese Water Dog	Autosomal Recessive	Clear
GM2 Gangliosidosis, mutation originally found in Japanese Chin	Autosomal Recessive	Clear
GM2 Gangliosidosis; mutation originally found in Toy Poodle	Autosomal Recessive	Clear
Globoid Cell Leukodystrophy or Krabbe Disease, (GLD); mutation originally found in Irish Setter	Autosomal Recessive	Clear
Globoid Cell Leukodystrophy or Krabbe Disease, (GLD); mutation originally found in Terriers	Autosomal Recessive	Clear
Paroxysmal Dyskinesia, (PxD); mutation originally found in Irish Soft Coated Wheaten Terrier	Autosomal Recessive	Clear

## Test results - Additional disorders found in other breeds - page 12

### Skeletal Disorders

Disorder	Mode of Inheritance	Result
Chondrodysplasia; mutation originally found in Norwegian Elkhound and Karelian Bear Dog	Autosomal Recessive	Clear
Cleft Palate; Cleft Lip and Palate with Syndactyly; ADAMTS20 gene mutation originally found in Nova Scotia Duck Tolling Retriever	Autosomal Recessive	Clear
Cleft Palate; DLX6 gene mutation originally found in Nova Scotia Duck Tolling Retriever	Autosomal Recessive	Clear
Craniomandibular Osteopathy, (CMO); mutation associated with terrier breeds	Autosomal Dominant (Incomplete Penetrance)	Clear
Hereditary Vitamin D-Resistant Rickets, (HVDRR)	Autosomal Recessive	Clear
Osteochondrodysplasia; mutation originally found in Miniature Poodle	Autosomal Recessive	Clear
Osteochondromatosis; mutation originally found in American Staffordshire Terrier	Autosomal Dominant	Clear
Osteogenesis Imperfecta, (OI); mutation originally found in Beagle	Autosomal Dominant	Clear
Osteogenesis Imperfecta, (OI); mutation originally found in Dachshund	Autosomal Recessive	Clear
Skeletal Disease (Hypophosphatasia); mutation originally found in Karelian Bear Dog	Autosomal Recessive	Clear
Skeletal Dysplasia 2, (SD2)	Autosomal Recessive	Clear
Spondylocostal Dysostosis	Autosomal Recessive	Clear
Van den Ende-Gupta Syndrome, (VDEGS)	Autosomal Recessive	Clear

## Test results - Additional disorders found in other breeds - page 13

### Dermal Disorders

Disorder	Mode of Inheritance	Result
Dystrophic Epidermolysis Bullosa; mutation originally found in Central Asian Ovcharka	Autosomal Recessive	Clear
Dystrophic Epidermolysis Bullosa; mutation originally found in Golden Retriever	Autosomal Recessive	Clear
Epidermolytic Hyperkeratosis	Autosomal Recessive	Clear
Focal Non-Epidermolytic Palmoplantar Keratoderma, (FNEPPK); mutation originally found in Dogue de Bordeaux	Autosomal Recessive	Clear
Hereditary Footpad Hyperkeratosis, (HFH)	Autosomal Recessive	Clear
Hereditary Nasal Parakeratosis, (HNPK); mutation originally found in Greyhound	Autosomal Recessive	Clear
Ichthyosis; mutation originally found in American Bulldog	Autosomal Recessive	Clear
Ichthyosis; mutation originally found in Great Dane	Autosomal Recessive	Clear
Lamellar Ichthyosis, (LI)	Autosomal Recessive	Clear
Lethal Acrodermatitis, (LAD); mutation originally found in in Bull Terrier and Miniature Bull Terrier	Autosomal Recessive	Clear
Ligneous Membranitis	Autosomal Recessive	Clear
Musladin-Lueke syndrome, (MLS)	Autosomal Recessive	Clear
X-Linked Ectodermal Dysplasia, (XHED)	X-linked Recessive	Clear

## Test results - Additional disorders found in other breeds - page 14

### Other Disorders

Disorder	Mode of Inheritance	Result
Acute Respiratory Distress Syndrome, (ARDS); mutation originally found in Dalmatian	Autosomal Recessive	Clear
Amelogenesis Imperfecta, (AI)	Autosomal Recessive	Clear
Amelogenesis Imperfecta, (AI); mutation originally found in Parson Russell Terrier	Autosomal Recessive	Clear
Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, (CKCSID)	Autosomal Recessive	Clear
Dental Hypomineralization; mutation originally found in Border Collie	Autosomal Recessive	Clear
Narcolepsy; mutation originally found in Dachshund	Autosomal Recessive	Clear
Narcolepsy; mutation originally found in Labrador Retriever	Autosomal Recessive	Clear
Persistent Müllerian Duct Syndrome, (PMDS); mutation originally found in Miniature Schnauzer	Autosomal Recessive	Clear
Primary Ciliary Dyskinesia, (PCD)	Autosomal Recessive	Clear

## APPENDIX

### Explanation of the results of the tested disorders

#### Autosomal recessive inheritance (ARI)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - A dog carries one copy of the tested mutation. Carriers typically have a normal, healthy appearance but pass on the mutation to approximately 50% of their offspring.

At risk - A dog carries two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

#### Autosomal dominant inheritance (ADI)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

At risk - A dog carries one or two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

#### X-linked recessive inheritance (X-linked)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - Female carriers typically have a normal, healthy appearance but carry one copy of the tested mutation on one of their X chromosomes. As males only have one X chromosome, there are no male carriers.

At risk - Female dogs at risk carry two mutated copies of the tested mutation. Males carry one copy of the tested mutation on their single X chromosome. Dogs at risk are at high or increased risk of developing the disease/condition.

Please note that the descriptions above are generalized based on typically observed inheritance patterns. When obtaining a 'carrier' or 'at risk' test result, always refer to the corresponding online test documentation for more detailed information on the condition and any exceptions.

## OPTIMAL SELECTION™ DNA TEST TERMS AND CONDITIONS

Optimal Selection™ Genetic Breeding Analysis is a proprietary process designed and intended to be used on purebred dogs solely to 1) Help quantify the genetic compatibility of potential breeding pairs and 2) To identify specific alleles or DNA mutations that are associated with certain inherited diseases or traits. No other purpose is authorized or permitted. It is not intended to diagnose diseases or predict behavior in any particular dog.

Upon receipt of your dog's DNA sample, Wisdom Health will analyze your dog's DNA to determine chromosomal similarities and differences in the genetic profile of a potential sire and dam and provide a match analysis. Your dog's DNA will also be analyzed for the presence of specific alleles that are associated with inherited conditions identified as occurring in your dog's breed. Wisdom Health's testing procedures are designed to provide reliable and accurate results, but are not guaranteed. By submitting your dog's sample(s) for Optimal Selection™ analysis it is understood that you agree that the sample(s), analysis, results and related information may be used confidentially by Mars in conjunction with other samples to increase the understanding of the breed's genetic structure, as well as for internal, research and development, or statistical purposes and may be shared with third parties for these purposes.

Samples may be disposed of or stored at Wisdom Health's option and will not be returned. Please view the full Mars Privacy Policy here: <http://www.mars.com/global/policies/privacy/pp-english.aspx> It is also understood that future releases of the Optimal Selection™ test may refine results as more information is obtained regarding the breed structure and/or if new genetic markers are included.

Optimal Selection™ genetic assessments for individual dogs and potential mates will be available online to the person(s) who registered the sample. A dog's results, photo and other information may be shared by the owner with other individuals whom they choose or transferred to a new owner if the dog changes ownership. The content of such online services 1) may be altered due to changes, additions, or removals of a dog's information in the Optimal Selection™ database or due to changes in technical or other design of such services and 2) includes information about third parties and other Wisdom Health clients' dogs, which Wisdom Health is not responsible or liable for. Wisdom Health has right to terminate access to online services one year from the purchase date, unless a longer period has been agreed upon.

You agree to Wisdom Health instructions related to ordering process, payment, sampling and sample delivery. You also certify that the animal described in your order is the same animal whose sample is submitted for analysis, and that all information is accurate. You warrant that you are entitled to obtain and supply samples to Wisdom Health.

In the unlikely event that it is not possible to provide an analysis (for example due to an insufficient DNA sample) or that an error in the analysis occurs, liability by Wisdom Health or related companies and individuals is disclaimed and damages in any event are limited to the payment actually received by Wisdom Health for the specified analysis at issue. Wisdom Health's study of the complexities of the canine genome is ongoing with the goal of continuing to provide the most advanced and complete analysis possible.

Wisdom Health reserves the right to use any third party of its choice to undertake the testing, analysis or laboratory services for the analysis.