

# CANINE GENETIC DISEASE TESTS

The following tables are a compilation of tests for genetic diseases that are available at various laboratories throughout the world. It is up to date as of January 2010 and has been compiled from the following websites:

- [www.vetgen.com](http://www.vetgen.com)
- [www.optigen.com](http://www.optigen.com)
- [www.gtq.com.au](http://www.gtq.com.au)
- [research.vet.upenn.edu](http://research.vet.upenn.edu)



There are other tests relating to parentage, coat colour, coat length and breed which have not been included but information is available either from the laboratory or the websites mentioned above.

Samples required (blood, cheek swabs, urine) will differ with the tests and the laboratories, so please contact NZVP to get the appropriate sample. Prices will depend on exchange rates and courier costs. A current cost can be obtained from us.

The tests are grouped by Breed in Table 1 and by Genetic Disease in Table 2.

BREED	GENETIC DISEASE
Alaskan Klee Kai	Factor VII
American Bulldog	Neuronal Ceroid Lipofuscinosis
American Cocker Spaniel	prcd – PRA, Phosphofructokinase Deficiency (PFK)
American Eskimo Dog	prcd - PRA
American Hairless Terrier	Hairlessness
American Staffordshire Terrier	Cerebellar Ataxia
Aust. Stumpy Tail Cattle Dog	prcd - PRA
Australian Cattle Dog	prcd - PRA
Australian Shepherds	Ivermectin sensitivity MDR1, Collie Eye Anomaly, prcd – PRA, cobalamin malabsorption
Basenji	Pyruvate kinase deficiency, Fanconi syndrome,
Basset Hound	Methylmalonic aciduria, Severe combined immune deficiency (SCID)
Beagle	Cobalamin malabsorption, Factor VII Deficiency, PK Deficiency, Musladin-Lueke Syndrome
Bedlington Terrier	Copper toxicosis
Bernese Mountain Dog	von Willebrands Disease Type I
Black Russian Terrier	Hyperuricosuria
Boston Terrier	Hereditary cataracts
Border Collies	Collie Eye Anomaly, Neuronal Ceroid Lipofuscinosis, Cobalamin malabsorption
British Bulldog	Neuronal Ceroid Lipofuscinosis
Briards	Congenital Stationary Night Blindness
Bulldog	Hyperuricosuria
Bull Mastiff	Dominant PRA, Multifocal retinopathy
Bull Terrier	Factor IX (Haemophilia B)
Cairn Terrier	PK Deficiency, Globoid Cell Leukodystrophy (GCL) / Krabbe's Disease
Cardigan Welsh Corgi	rcd3 – PRA, Severe Combined Immunodeficiency (SCID)

**Table 1 cont'd.**

<b>BREED</b>	<b>GENETIC DISEASE</b>
Chesapeake Bay Retriever	prcd - PRA
Chihuahua	PK deficiency
Chinese Crested	prcd - PRA
Clumber Spaniel	Pyruvate Dehydrogenase Phosphatase Deficiency (PDP)
Cocker Spaniel	Mitochondrial myopathy, Phosphofructokinase deficiency (PFK)
Collie	Collie Eye Anomaly, Cyclic neutropenia (Gray Collie syndrome)
Collie (Rough)	Ivermectin sensitivity MDR1, Collie Eye Anomaly, rcd2- PRA
Coton de Tulear	Multi focal retinopathy, von Willebrands Disease Type I
Dalmatian	Hyperuricosuria
Daschund	Narcolepsy, PK deficiency
Deutsch Drahthaar	von Willebrands Disease Type II
Doberman	von Willebrands Disease Type I, Narcolepsy
Dogue de Bordeaux	Multi focal retinopathy
Drentsche Patrijshond	von Willebrands Disease Type I
English Cocker Spaniel	prcd – PRA, Autosomal Hereditary Recessive Nephropathy (FN)
English Setter	Neuronal Ceroid Lipofuscinosis
English Springer Spaniel	Fucosidosis, Phosphofructokinase Deficiency (PFK)
Entlebucher Mountain Dog	prcd – PRA
Eskimo Toy	PK deficiency
Finnish Lapphund	prcd – PRA
Fox Terrier (Toy)	Congenital hypothyroidism
French Bulldog	Hereditary Cataracts
German Pinscher	von Willebrands Disease Type I
German Shepherd	MPS VII, Renal cystadenocarcinoma nodular dermatofibrosis (RCND)
German Shorthaired Pointer	Cone Degeneration, von Willebrands Disease Type II
German Wirehaired Pointer	von Willebrands Disease Type II
Giant Schnauzer	Cobalamin malabsorption
Golden Retriever	prcd - PRA
Great Pyrenees	Multi focal retinopathy
Huntaway	MPS IIIA
Irish Red & White Setter	rcd1 - PRA
Irish Setter	rcd1 – PRA, Canine Leucocyte Adhesion Deficiency (CLAD)
Karelian Bear Dog	prcd - PRA
Kerry Blue Terrier	Factor XI Deficiency, von Willebrands Disease Type I
Komondor	Cobalamin malabsorption
Kooikerhondje	von Willebrands Disease Type III
Kuvazs	prcd - PRA
Labrador Retriever	prcd – PRA, Retinal dysplasia/OSD, Narcolepsy, CentroNuclear Myopathy, cystinuria
Lancashire Heelers	Collie Eye Anomaly
Lapponian Herders	prcd - PRA
Lhasa Apso	Factor IX (Haemophilia B), Renal Dysplasia
Manchester Terrier	von Willebrands Disease Type I
Mastiff (English)	Dominant PRA, Multi focal retinopathy
Miniature Pinscher	MPS VI

**Table 1 cont'd.**

<b>BREED</b>	<b>GENETIC DISEASE</b>
Miniature Poodle	prcd – PRA
Miniature Schnauzer	Type A – PRA, MPS VI, Myotonia congenita
Newfoundland	Cystinuria
Norwegian Elkhound	prcd – PRA, Fanconi syndrome, Renal glucosuria
Nova Scotia Duck Tolling Retriever	prcd – PRA, Collie Eye Anomaly
Old English Sheep Dog	Ivermectin sensitivity MDR1, Mitochondrial myopathy
Papillon	von Willebrands Disease Type I
Pembroke Welsh Corgi	Severe Combined Immunodeficiency (SCID)
Plott Hound	MPS I
Pointer	von Willebrands Disease Type II
Poodle (Standard)	von Willebrands Disease Type I, Neonatal Encephalopathy
Poodle (Min & Toy)	prcd – PRA
Portuguese Water Dog	prcd – PRA, GM-1, Juvenile dilated cardiomyopathy (JDCM), hypersarcosinemia
Rottweiler	MPS I
Samoyed	X-Linked PRA, Retinal dysplasia/OSD (dwarfism), Hereditary nephropathy
Schipperke	MPS IIIB
Scottish Deerhound	Factor VII Deficiency
Scottish Terrier	von Willebrands Disease Type III
Setters (Irish, Red & White)	CLAD, rcd1-PRA
Shar Pei	Methylmalonic aciduria
Shetland Sheepdog	Ivermectin sensitivity MDR1, Collie Eye Anomaly, Von Willebrands Disease Type III
Shih Tzu	Renal Dysplasia
Siberian Husky	X-Linked PRA
Silky Terrier	prcd – PRA
Sloughi	rcd1a - PRA
Smooth Collies	Collie Eye Anomaly, rcd2 - PRA
Soft Coated Wheaten Terrier	Renal Dysplasia
Spanish Water Dog	prcd – PRA
Stabyhoun	von Willebrands Disease Type I
Staffordshire Bull Terrier	Hereditary Cataracts, L-2-hydroxyglutaric aciduria (L2-HGA)
Swedish Lapphund	prcd – PRA
Sussex Spaniel	Pyruvate Dehydrogenase Phosphatase Deficiency (PDP)
Tibetan Spaniel	Primary hyperoxaluria
Welsh Corgi	MPS VI, Severe Combined Immune Deficiency (SCID), von Willebrands Disease Type I
West Highland White Terrier	Pyruvate Kinase (PK) deficiency, Globoid Cell Leukodystrophy (GCL) / Krabbe's Disease
Whippet	Phosphofructokinase (PFK) deficiency
Whippet (Longhaired)	Collie Eye Anomaly
Wire Haired Dachshund	MPS IIIA
Yorkshire Terrier	prcd – PRA

**Table 2: Tests By Genetic Disease**

<b>GENETIC DISEASE</b>	<b>BREED</b>
Autosomal Recessive Hereditary Nephropathy	English Cocker Spaniel
Canine Leucocyte Adhesion Deficiency (CLAD)	Irish Setter
CentroNuclear Myopathy (CNM)	Labrador Retriever
Cerebellar Ataxia	American Staffordshire Terrier
Cobalamin malabsorption	Australian Shepherd, Beagle, Border Collie, Giant Schnauzer, Komondor
Collie Eye Anomaly/ Choroidal Hypoplasia (CE/CH)	Border Collie, Collie (Smooth/Rough), Lancashire Heelers, Shetland Sheepdog, Australian Shepherd, Nova Scotia Duck Tolling Retriever, Long Haired Whippet
Cone Degeneration (CD)	German Shorthaired Pointer
Congenital Stationary Night Blindness (CSNB)	Briard
Copper Toxicosis	Bedlington Terrier
Cystinuria	Newfoundland, Labrador Retriever
Factor VII Deficiency	Beagle, Scottish Deerhound, Alaskan Klee Kai
Factor IX Deficiency (Haemophilia B)	Bull Terrier, Lhasa Apso
Factor XI Deficiency	Kerry Blue Terrier
Fanconi Syndrome	Basenji, Norwegian Elkhound
Fucosidosis	English Springer Spaniel
Globoid Cell Leukodystrophy (GCL) / Krabbe's Disease	Cairn Terrier, West Highland White Terrier
Grey Collie Syndrome (canine cyclic neutropenia)	Collies – Rough & Smooth
GM1 - Gangliosidosis	Portuguese Water Dog
Hairlessness in Terriers	American Hairless Terriers
Hereditary Cataract (HC)	Staffordshire Bull Terrier, Boston Terrier, French Bulldog
Hereditary Nephropathy (ARHN)	English Cocker Spaniel, Samoyed (Dominant form)
Hypersarcosinemia	Portuguese Water Dog
Hyperuricosuria	All dogs but most common in Dalmatian, Bulldog, Black Russian Terrier
Hypothyroidism (congenital)	Toy Fox Terrier (and other breeds)
Ivermectin Sensitivity MDR1	Collie (Rough), Shetland Sheepdog, Australian Shepherd, Old English Sheep Dog
Juvenile Dilated Cardiomyopathy (JDCM)	Portuguese Water Dog
L-2-hydroxyglutaric aciduria (L2-HGA)	Staffordshire Bull Terrier
Methylmalonic Aciduria	Shar Pei, Basset Hound
Mitochondrial myopathy	Cocker Spaniel, Old English Sheepdog
Mucopolysaccharidosis (MPS I)	Plott Hound, Rottweiler
MPS IIIA	Wire Haired Dachshund, New Zealand Huntaway
MPS IIIB	Schipperke
MPS VI	Miniature Pinscher, Miniature Schnauzer, Welsh Corgi
MPS VII	German Shepherd
Multifocal Retinopathy	Bull Mastiff, English Mastiff, Coton De Tulear, Dogue de Bordeaux, Great Pyrenees
Musladin-Lueke Syndrome	Beagle

**Table 2 cont'd.**

<b>GENETIC DISEASE</b>	<b>BREED</b>
Myotonia Congenita	Miniature Schnauzer
Narcolepsy (NARC)	Doberman, Labrador Retriever, Dachshund
Neonatal Encephalopathy (NEN)	Standard Poodle
Neuronal Ceroid Lipofuscinosis (NCL)	Border Collie, American & British Bulldogs, English Setter
Phosphofructokinase (PFK) Deficiency	American Cocker Spaniel, Cocker Spaniel, English Springer Spaniel, Whippet
Primary hyperoxaluria	Tibetan Spaniel
Progressive Retinal Atrophy (PRA Dominant)	Bull Mastiff, English Mastiff
Progressive Retinal Atrophy (PRA Type A)	Miniature Schnauzer
Progressive Retinal Atrophy (PRA X-linked)	Samoyed, Siberian Husky
Progressive Retinal Atrophy (PRA prcd)	Australian Cattle Dog, Labrador Retriever, Golden Retriever, Australian Stumpy Tail Cattle dog, Portuguese Water Dog, Poodle (Miniature & Toy), Chinese Crested, Hungarian Kuvasz, Nova Scotia Duck Tolling Retriever, English Cocker Spaniel, Swedish Lapphund, Finnish Lapphund, American Cocker Spaniel, Chesapeake Bay Retriever, American Eskimo Dog, Australian Shepherd, Entlebucher Mountain Dog, Karelian Bear Dog, Lapponian Herders, Norwegian Elkhound, Silky Terriers, Spanish Water Dog, Yorkshire Terrier
Progressive Retinal Atrophy (PRA rcd1)	Irish Setter, Irish Red & White Setter
Progressive Retinal Atrophy (PRA rcd1a)	Sloughi
Progressive Retinal Atrophy (PRA rcd 2)	Collies
Progressive Retinal Atrophy (PRA rcd 3)	Cardigan Welsh Corgi
Pyruvate Dehydrogenase Phosphatase 1 Deficiency (PDP)	Clumber Spaniel, Sussex Spaniel
Pyruvate Kinase Deficiency (PK)	Beagle, Cairn Terrier, Chihuahua, Dachshund, Basenji, West Highland White Terrier, Eskimo Toy
Renal cystadenocarcinoma nodular dermatofibrosis (RCND)	German Shepherd
Renal Dysplasia (RD)	Lhasa Apso, Shih Tzu, Soft Coated Wheaten Terrier
Renal Glucosuria	Norwegian Elkhound
Retinal Dysplasia/OSD (dwarfism)	Labrador Retriever, Samoyed
Severe Combined Immunodeficiency (SCID)	Basset Hound, Cardigan & Pembroke Welsh Corgi
Von Willebrands Disease (vWD) Type I	Bernese Mountain Dog, Coton de Tulear, Doberman Pinscher, Drentsche Patrijshond, German Pinscher, Kerry Blue Terrier, Manchester Terrier, Papillon, Pembroke Welsh Corgi, Standard Poodle, Stabyhoun
vWD Type II	German Shorthaired Pointer, German Wirehaired Pointer, Pointer, Deutsch Drahthaar
vWD Type III	Scottish Terrier, Shetland Sheepdog, Kooikerhondje