

TABLE 1
Available Canine Genetic Tests

Table 1 is limited to testing laboratories in the United States and is intended as a guide and not a complete reference.

Disease*	Causative Gene	Breeds Affected	Inheritance Pattern	Availability of Test**
Arrhythmogenic right ventricular cardiomyopathy ¹	STRN	Boxer	Autosomal dominant— incomplete penetrance	NCSU
Canine leukocyte adhesion deficiency ²	ITGB2	Irish setter	Autosomal recessive	OptiGen***
Canine multifocal retinopathy [†]	VMD2	Mastiff, Great Pyrenees, Coton de Tulear	Autosomal recessive	OptiGen
Centronuclear myopathy ³	PTPLA	Labrador retriever	Autosomal recessive	AGI***, DDC, Vet- Gen
Cerebellar ataxia [†]	Not stated	American Staffordshire terrier, American pit bull terrier	Autosomal recessive	OptiGen
Cobalamin malabsorption [†]	Not stated	Australian shepherd, giant schnauzer	Not stated	UPenn
Collie eye anomaly, choroidal hypoplasia ⁴	NHEJ1	Australian shepherd, border collie, Lan- cashire heeler, rough collie, Shetland sheep- dog, smooth collie	Autosomal recessive	OptiGen
Cone degeneration ⁵	CNGB3	German shorthaired pointer, Alaskan mala- mute	Autosomal recessive	OptiGen
Cone-rod dystrophy 2 [†]	Not stated	American pit bull terrier	Autosomal recessive	OptiGen
Cone-rod dystrophy 3 ^{6,7}	ADAM9	Glen of Imaal terrier	Autosomal recessive	OptiGen***
Congenital hypothyroidism with goiter ^{8,9}	TPO	Toy fox terrier	Autosomal recessive	MSU, UPenn
Congenital stationary night blindness ¹⁰	REP65	Briard	Autosomal recessive	OptiGen***
Copper toxicosis ¹¹	COMMD1	Bedlington terrier	Autosomal recessive	VetGen
Cyclic neutropenia ¹²	AP3B1	Collie	Autosomal recessive	AGI***, VetGen
Cystinuria ¹³	SLC3A1	Newfoundland, Labrador retriever	Autosomal recessive	AGI***, DDC, OptiGen***, UPenn, VetGen
Degenerative myelopathy ¹⁴	SOD1	Too many to list (all breeds can be tested)	Autosomal recessive	AGI***, DDC, OFA, UMCCVM
Dilated cardiomyopathy [†]	PDK4	Doberman pinscher	Autosomal dominant	NCSU
Dominant progressive retinal atrophy ¹⁵	RHO	Old English mastiff, bullmastiff	Autosomal dominant	OptiGen
Exercise-induced collapse ¹⁶	DNM1	Labrador retriever, Boykin spaniel, Ches- apeake Bay retriever, curly coated retriever, German wirehaired pointer, Pembroke Welsh corgi	Autosomal recessive	AGI***, DDC, UMinn

TABLE 1, continued

Disease*	Causative Gene	Breeds Affected	Inheritance Pattern	Availability of Test**
Factor VII deficiency ¹⁷	CFVII	Beagle, Airedale terrier, Alaskan klee kai, giant schnauzer, Scottish deerhound	Autosomal recessive	UPenn, VetGen
Familial nephropathy ¹⁸	COL4A4	English cocker spaniel	Autosomal recessive	OptiGen
Fucosidosis ¹⁹	FUCA	English springer spaniel	Autosomal recessive	AGI***, UPenn
Glanzmann's thombasthenia ²⁰	ITGA2B	Otterhound, Great Pyrenees	Autosomal recessive	Auburn
Globoid cell leukodystrophy ²¹	GALC	Cairn terrier, West Highland white terrier	Autosomal recessive	JMC***
Glycogen storage disease type IIIa ²²	AGL	Curly coated retriever	Autosomal recessive	MSU
Hemophilia B ²³	F9 (coagulation factor IX)	Bull terrier, Lhasa apso	X-linked recessive	VetGen
Hereditary nephritis ²⁴	COL4A5	Samoyed	X-linked dominant	VetGen
Hyperuricosuria ²⁵	SLC2A9	Dalmatian, black Russian terrier, beagle (all breeds can be tested)	Autosomal recessive	UCDavis
Ichthyosis ²⁶	KRT10	Norfolk terrier, golden retriever	Autosomal recessive	MSU, OptiGen
Juvenile hereditary/early onset cataracts ^{27,28}	HSF4	Boston terrier, Staffordshire bull terrier, French bulldog	Autosomal recessive	AGI***, Vet Gen
Juvenile hereditary cataracts ²⁹	HSF4	Australian shepherd	Autosomal dominant-incomplete penetrance	AGI***
Juvenile dilated cardiomyopathy [†]	Unknown	Portuguese water dog	Autosomal recessive	UPenn
L-2-hydroxyglutaric aciduria ³⁰	L2HGDH	Staffordshire bull terrier	Autosomal recessive	UMCCVM
Leonberger polyneuropathy [†]	LPN1	Leonberger, Saint Bernard	Not stated	UMinn
Musladin-Lueke syndrome ³¹	ADAMTSL2	Beagle	Autosomal recessive	UCDavis
Mucopolysaccharidosis VII ³²	GUSB	German shepherd	Autosomal recessive	UPenn
Mucopolysaccharidosis IIIB [†]	NAGLU	Schipperke	Autosomal recessive	UPenn
Mucopolysaccharidosis VI [†]	ARSB	Miniature pinscher	Autosomal recessive	UPenn
Multidrug sensitivity ³³	MDR1	Too many to list (all breeds can be tested)	Autosomal recessive	NCSU
Myotonia congenita ³⁴	CLC-1	Miniature schnauzer	Autosomal recessive	UPenn
Narcolepsy ³⁵	HCRT2	Dachshund, Doberman pinscher, Labrador retriever	Autosomal recessive	OptiGen***
Neonatal encephalopathy ³⁶	ATF2	Standard poodle	Autosomal recessive	OFA, VetGen, UMCCVM
Neuronal ceroid lipofuscinosis ³⁷⁻⁴²	CTSD, TPP1, CLN5, ATP13A2	American bulldog, English setter, dachshund, border collie, Tibetan terrier	Autosomal recessive	UMCCVM, VetGen, AGI***, OptiGen***

TABLE 1, continued

Disease*	Causative Gene	Breeds Affected	Inheritance Pattern	Availability of Test**
Phosphofructokinase deficiency ⁴³	M-PFK	English springer spaniel, American and English cocker spaniel, whippet	Autosomal recessive	AGI***, DDC, OptiGen***, UMCCVM, UPenn, VetGen
Polyneuropathy ⁴⁴	NDRG1	Greyhound	Autosomal recessive	OptiGen
Primary hyperparathyroidism [†]	Not stated	Keeshond	Autosomal dominant	Cornell***
Primary lens luxation ⁴⁵	ADAMTS17	Too many to list	Autosomal recessive	OFA, UMCCVM
Progressive retinal atrophy ^{46,47} (RPGRIPI)	RPGRIPI	Miniature longhaired dachshund	Autosomal recessive-incomplete penetrance	AGI***, VetGen, UMCCVM
Progressive retinal atrophy [†] (PDEB)	PDEB	Cardigan Welsh corgi, Irish setter	Autosomal recessive	VetGen
Progressive retinal atrophy ⁴⁸ (PRCD)	PRCD	Too many to list	Autosomal recessive	OptiGen
Progressive retinal atrophy ⁴⁹ (PDE6A)	PDE6A	Cardigan Welsh corgi	Autosomal recessive	OptiGen***
Progressive retinal atrophy ^{50,51} (PDE6B)	PDE6B	Irish setter, sloughi	Autosomal recessive	OptiGen***
Progressive retinal atrophy type A [†]	Not stated	Miniature schnauzer	Autosomal dominant-incomplete penetrance	OptiGen
Progressive retinal atrophy X-linked ⁵²	RPGR	Samoyed, Siberian husky	X-linked recessive	OptiGen
Pug dog encephalitis ⁵³	Unknown	Pug	Autosomal recessive	UCDavis
Pyruvate dehydrogenase phosphatase deficiency ⁵⁴	PDP1	Clumber spaniel, Sussex spaniel	Autosomal recessive	UMCCVM, VetGen
Pyruvate kinase deficiency ^{55,56}	R-PK	West Highland white terrier, basenji, beagle, Cairn terrier, Laborador retriever, pug	Autosomal recessive	DDC, UPenn, OptiGen, VetGen
Renal cystadenocarcinoma nodular dermatofibrosis ⁵⁷	BHD	German shepherd	Autosomal dominant	VetGen
Retinal dysplasia/oculoskeletal dysplasia ⁵⁸	COL9A	Labrador retriever (Col9A3), Samoyed (Col9A2)	Autosomal recessive	OptiGen
Rod-cone dysplasia type 2 ⁵⁹	RD3	Collie	Autosomal recessive	OptiGen
Severe combined immune-deficiency ⁶⁰	IL-2RG	Basset hound, Welsh corgi	X-linked recessive	UPenn
Thrombopathia ⁶¹	RASGRP2	Basset hound	Autosomal recessive	Auburn
von Willebrand disease type I ⁶²	vWF	Bernese mountain dog, Coton de Tulear, Doberman pinscher, Drentsche patrijshond, German pinscher, Kerry blue terrier, papillon, Pembroke Welsh corgi, poodle, stabyhoun	Autosomal recessive with incomplete penetrance	VetGen

TABLE 1, continued

Disease*	Causative Gene	Breeds Affected	Inheritance Pattern	Availability of Test**
von Willebrand disease type II ⁶³	vWF	Deutsch Drahthaar, German shorthaired pointer, German wire-haired pointer, German longhaired pointer, collie, pointer	Autosomal recessive	VetGen
von Willebrand disease type III ⁶⁴	vWF	Scottish terrier, Shetland sheepdog, kooikehondje	Autosomal recessive	VetGen

*References listed are mutations published in scientific journals for the disease listed. Mutation listed may not correlate with actual test run by provider.

**See Abbreviation Key.

***Testing requires a blood sample. All other tests require buccal cheek swabs.

†No scientific publication available through PubMed.

Abbreviation Key

- **AGI** Animal Genetics, 1336 Timberland Road, Tallahassee, FL 32312 (www.animalgenetics.us).
- **Auburn** Auburn University, College of Veterinary Medicine, Auburn, AL 36849 (www.vetmed.auburn.edu/faculty/pathobiology-faculty/boudreaux).
- **Cornell** Cornell University, College of Veterinary Medicine, Goldstein Molecular and Genetics Laboratory, Ithaca, NY 14853 (www.vet.cornell.edu/labs/goldstein).
- **DDC** DNA Diagnostics Center Veterinary, One DDC Way, Fairfield, OH 45014 (www.vetdnacenter.com).
- **JMC** Thomas Jefferson University, David Wenger, PhD, Jefferson Medical College, Department of Neurology, 1020 Locust St. JAH394, Philadelphia, PA 19107.
- **MSU** Michigan State University, College of Veterinary Medicine, Laboratory of Comparative Medical Genetics, East Lansing, MI 48824 (www.mmg.msu.edu/fyfe.html).
- **NCSU** North Carolina State University, College of Veterinary Medicine, Veterinary Cardiac Genetics Laboratory, Raleigh, NC 27606 (<http://www.cvm.ncsu.edu/vhc/csds/vcg/index.html>).
- **OFA** Orthopedic Foundation for Animals, 2300 E. Nifong Blvd, Columbia, MO 65201 (www.offa.org/dnatesting).
- **OptiGen** OptiGen, 767 Warren Road, Suite 300, Ithaca, NY 14850 (www.optigen.com).
- **UCDavis** University of California Davis, School of Veterinary Medicine, Veterinary Genetics Laboratory, P.O. Box 1102, Davis, CA 95617 (www.vgl.ucdavis.edu).
- **UMCCVM** University of Missouri, College of Veterinary Medicine, Columbia, MO 65211 (www.caninegeneticdisorders.net).
- **UMinn** University of Minnesota, College of Veterinary Medicine, Veterinary Diagnostic Laboratory, St. Paul, MN 55108 (www.vdl.umn.edu/ourservices/canineneuromuscular).
- **UPenn** University of Pennsylvania, School of Veterinary Medicine, PennGen Laboratories, Philadelphia, PA 19104 (<http://research.vet.upenn.edu/penngen>).
- **VetGen** Veterinary Genetic Services, 3728 Plaza Drive, Suite 1, Ann Arbor, MI 48108 (www.vetgen.com).

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