

Known Genes/Mutations and available Genetic Testing

ADAM9 [1, 2]

Cone-Rod Dystrophy 3 (crd3)

Glen of Imaal Terrier (Optigen; Bochum University)

ADAMST10 [3, 4]

Primary open angle glaucoma (POAG)

Beagle (vetGen; Optigen; UCD)

Norwegian Elkhound (vetGen)

ADAMST17 [5]

Primary open angle glaucoma (POAG)

Basset Hound (vetGen; AHT)

Petit Basset Griffon Vandeun (AHT)

ADAMST17 [6, 7]

Primary Lens Luxation

American Eskimo Dog (Optigen; UCD)

Australian Cattle Dog (MU; vetGen; AHT; Optigen; UCD)

Australian Kelpie (vetGen)

Border Collie (vetGen)

Chinese Crested (MU; vetGen; AHT; Optigen; UCD)

Chinese Foo Dog (Optigen; UCD)

Jack Russell Terrier (vetGen; MU; AHT; Optigen; UCD)

Jagdterrier (vetGen; MU; AHT; Optigen; UCD)

Lakeland Terrier (vetGen; Optigen; UCD)

Lancashire Heeler (vetGen; MU; AHT; Optigen; UCD)

Lucas Terrier (Optigen; UCD)

Miniature Bull Terrier (vetGen; MU; AHT; Optigen; UCD)

Norwich Terrier (vetGen; Optigen; UCD)

Parson Russell Terrier (vetGen; MU; AHT; Optigen; UCD)

Patterdale Terrier (vetGen; MU; AHT; Optigen)

Rat Terrier (vetGen; MU; AHT; Optigen; UCD)

Sealyham Terrier (vetGen; MU; AHT; Optigen; UCD)

Teddy Roosevelt Terrier (Optigen)

Tenterfield Terrier (vetGen; MU; AHT; Optigen; UCD)

Tibetan Terrier (vetGen; MU; AHT; Optigen; UCD)

Toy Fox Terrier (vetGen; AHT; Optigen; UCD)

Volpino Italiano (vetGen; MU; AHT; Optigen; UCD)

Welsh Terrier (vetGen; MU; AHT; Optigen; UCD)

Wire-haired Fox Terrier (vetGen; AHT; Optigen; UCD)

Yorkshire Terrier (vetGen; AHT; Optigen; UCD)

ARSG [8]

Neuronal ceroid lipofuscinosis (NCL-A)

American Staffordshire Terrier (Optigen)

ATP13A2 [9]

Neuronal ceroid lipofuscinosis (NCL)

Tibetan Terrier (MU; AHT)

BEST1 (VMD2) [10-13]***Canine multifocal retinopathy type 1 (MCR1)***

American Bulldog (vetGen; Optigen)
Australian Shepherd (vetGen; Optigen)
Australian Shepherd Miniature (Optigen)
Australian Shepherd Toy (Optigen)
Boerboel (Optigen)
Bull Mastiff (vetGen; Optigen)
Cane Corso (vetGen; Optigen)
Dogue de Bordeaux (vetGen; Optigen)
English Bulldog (vetGen)
English Bull Mastiff (vetGen; Optigen)
French Bulldog (vetGen)
Great Pyrenees (vetGen; Optigen)
Miniature American Shepherd (Optigen)
Old English Mastiff (vetGen; Optigen)
Perro de Presna Canario (Optigen)

Canine multifocal retinopathy type 2 (CMR2)

Coton du Tulear (vetGen; Optigen)

Canine multifocal retinopathy type 3 (CMR3)

Lapponian Herder (Optigen)

C2orf71 [14, 15]***Late Onset Progressive Retinal Atrophy (rcd4-PRA)***

Australian Cattle Dog (MU; vetGen; Optigen)
English Setter (MU; vetGen; Optigen)
Gordon Setter (MU; vetGen; Optigen; ATH)
Irish Setter (MU; vetGen; Optigen; AHT)
Llewellyn Setter (Optigen)
Polish Lowland Sheepdog (MU; Optigen)
Small Munsterlander (MU; Optigen)
Standard Poodles (vetGen; Optigen)
Tibetan Terrier (MU; vetGen; Optigen; AHT)

CCDC66 [16, 17]***Generalized Progressive Retinal Atrophy (gPRA)***

Schapendoes (Bochum University)

CHST6 [18]***Macular corneal dystrophy (MCD)***

Labrador Retriever (vetGen; AHT)
Labradoodle (vetGen)

CLN5 [19]***Neuronal ceroid lipofuscinosis (CL)***

Border Collie (Optigen)

CLN6 [20]***Neuronal ceroid lipofuscinosis (NCL6)***

Australian Shepherd (vetGen; MU)

CLN8 [21]

Neuronal ceroid lipofuscinosis (NCL8)

English Setter (vetGen; MU)

Gordon Setter (vetGen)

Irish Setter (vetGen)

CNGA1 [22]

CNGA1-Progressive Retinal Atrophy (CNGA1-PRA)

Shetland Sheepdog

CNGA3 [23]

Achromatopsia 2

Australian Cobberdog (Optigen)

Labrador Retriever (Optigen)

Labradoodle (Optigen)

CNGA3 (unpublished)

Achromatopsia 2 German Shepherd (ww.Optigen.com) – different mutation to that in Labrador

CNGB1 [24, 25]

Papillon Progressive Retinal Atrophy (Pap_PRA1)

Papillon (Optigen)

Phalene (Optigen)

CNGB3 [26]

Cone Degeneration (CD achromatopsia)

Alaskan Malamute (Optigen, UCD)

Australian Shepherd (Optigen, UCD)

Australian Shepherd Miniature (Optigen, UCD)

Australian Shepherd Toy (Optigen, UCD)

German Shorthaired Pointer (Optigen, UCD)

Miniature American Sheperd (Optigen, UCD)

Siberian Husky (UCD)

COL9A2 [27]

Ocular Skeletal Dysplasia (RD/OSD dwarfis with retinal dysplasia - Drd2)

Samoyed (Optigen)

Australian Cobberdog (Optigen)

COL9A3 [27, 28]

Ocular Skeletal Dysplasia (RD/OSD dwarfism with retinal dysplasia - Drd1)

Labrador Retriever (Optigen)

Labradoodles (Optigen)

CTSD [29]

Neuronal ceroid lipofuscinosis (NCL10)

American Bulldog (vetGen; MU)

FAM161A [30]

Progressive Retinal Atrophy 3 (PRA3)

Tibetan Spaniel (vetGen; AHT)

Tibetan Terrier (vetGen; AHT)

FAM83H [31-33]***Curly Coat Dry Eye***

Cavalier King Charles Spaniel (vetGen; AHT)

GLB1 [34, 35]***GM1 gangliosidosis***

Shiba Inu (vetGen)

Siberian Husky (vetGen)

Portuguese Water Dog (Optigen)

HEXA [36]***GM2 gangliosidosis***

Japanese Chin (MU)

HSF4-1 [37, 38]***Hereditary Cataract***

Boston Terrier (vetGen; AHT; Optigen)

French Bulldog (vetGen; AHT; Optigen)

Staffordshire Bull Terrier (vetGen; AHT; Optigen)

HSF4-2 [39]***Hereditary Cataract***

Australian Shepherd (vetGen; AHT; Optigen)

Australian Shepherd Miniature (Optigen)

Australian Shepherd Toy (Optigen)

IQCB1 [40]***Cone-Rod Dystrophy 2 (crd2)***

American Pit Bull Terrier (Optigen; UCD)

NHEJ1 [41-43]***Collie Eye Anomaly-Choroidal Hypoplasia (CEA-CH)***

Australian Shepherd (Optigen)

Australian Shepherd Miniature (Optigen)

Australian Shepherd Toy (Optigen)

Bearded Collie (Optigen)

Border Collie (Optigen)

Boykin Spaniel (Optigen)

Hokkaido Dog (Optigen)

Lancashire Heeler (Optigen)

Longhaired Whippet (Optigen)

Miniature American Shepherd (Optigen)

Nova Scotia Duck Tolling Retriever (Optigen)

Rough & Smooth Collie (Optigen)

Shetland Sheepdog (Optigen)

Silken Windhound (Optigen)

Whippet LongHaired (Optigen)

NPHP4 [44]***Cone-Rod Dystrophy (CRD)***

Wire-Haired Dachshund Miniature and Standard (AHT)

Short-haired Dachshund (NSVS)

PDE6A [45]

Rod-Cone Dysplasia 3 (arPRA – rcd3)

Cardigan Welsh Corgi (vetGen; Optigen)

Chinese crested Dog (vetGen; Optigen)

Pembroke Welsh Corgi (vetGen)

PDE6B

Rod-Cone Dysplasia 1 (arPRA – rcd1) [46, 47]

Irish Red and White Setter (vetGen; Optigen, AHT; UCD)

Irish Setter (vetGen; Optigen, AHT; UCD)

Rod-Cone Dysplasia 1a (arPRA – rcd1a) [48]

Sloughi (Optigen; UCD; Bochum University)

PDE6B [40]

Cone-Rod Dystrophy 1 (crd1)

American Staffordshire Terrier (UCD)

PPT1 [49]

Neuronal ceroid lipofuscinosis (NCL1)

Dachshund (vetGen; MU)

PRCD [50-52]

Progressive Rod Cone Degeneration (arPRA - prcd)

American Cocker Spaniel (Optigen)

American Eskimo Dog (Optigen)

Australian Cattle Dog (Optigen)

Australian Cobberdog (Optigen)

Australian Shepherd (Optigen)

Australian Shepherd Miniature (Optigen)

Australian Stumpy Tail Cattle Dog (Optigen)

Bolonka Zwetna (Optigen)

Chesapeake Bay Retriever (Optigen)

Chinese Crested (Optigen)

Cockapoo (Optigen)

Dwarf Poodle (Optigen)

English Cocker Spaniel (Optigen)

English Sheperd (Optigen)

Entlebucher Sennenhund (Optigen)

Finnish Lapphund (Optigen)

German Spitz (Optigen)

Giant Schnauzer (Optigen)

Golden Retriever (Optigen)

Karelina Bear Dog (Optigen)

Kuvasz (Optigen)

Labrador Retriever (Optigen)

Labradoodle (Optigen)

Lapponian Herder (Optigen)

Malipoo (Optigen)

Markiesje (Optigen)

Miniature & Toy Poodle (Optigen)

Moyen Poodle (Optigen)
Norwegian Elkhound (Optigen)
Nova Scotia Duck Tolling Retriever (Optigen)
Portuguese Water Dog (Optigen)
Schipperke (Optigen)
Silky Terrier (Optigen)
Spanish Water Dog (Optigen)
Swedish Lapphund (Optigen)
Yorkshire Terrier (Optigen)

RD3 [53]

Rod Cone Dysplasia (rcd2)

Rough & Smooth Collie (Optigen)

rhodopsin [54]

Dominant Progressive Retinal Atrophy (Dominant-PRA)

Bull Mastiff (vetGen; Optigen)
Old English Mastiff (vetGen; Optigen)

RPE65 [55, 56]

Congenital Stationary Night Blindness (CSNB)

Briard (Optigen, AHT)

RPGR [57-59]

X-linked Progressive Retinal Atrophy (xlPRA1)

Samoyed (Optigen)
Siberian Husky (Optigen)

RPGRIP1 [60-65]

Cone-Rod Dystrophy (CORD1)

Chihuahua (vetGen)
English Springer Spaniel (AHT; MU)
Field Spaniel (vetGen)
Miniature Long-haired & Smooth-haired & Wirehaired Dachshund (vetGen; AHT; MU)
Papillon (vetGen)

Note that the genetic changes underlying *CORD1* are more complex than initially thought. Following identification of the *RPGRIP1* mutation in a closed colony it became apparent that not all dogs in the pet population with this mutation developed clinical signs of PRA. A second modifying locus influencing the expression of the initially described *RPGRIP1* mutation was then identified. Ongoing studies are attempting to fully understand the genotype-phenotype correlation and interaction between the two gene variations. In some breeds the *RPGRIP1* mutation does not segregate with PRA and therefore the *RPGRIP1* genotyping test needs to be interpreted with caution.

SAG mut CFA25 [66]

Basenji Progressive Retinal Atrophy (Bas_PRA1)

Basenji (Optigen; MU)

SLC4A3 [67]

Golden Retriever Progressive Retinal Atrophy 1 (GR_PRA1)

Golden Retriever (vetGen; Optigen; AHT; UCD)

Lhasa Apso (vetGen)
Miniature Schnauzer (vetGen)

STK38L [68]

Early Retinal Degeneration (arPRA- erd)
Norwegian Elkhound (test not available)

TPP1 [69]

Neuronal ceroid lipofuscinosis (NCL2)
Long-haired Dachshund (vetGen; MU)

TTC8 [70]

Golden Retriever Progressive Retinal Atrophy 2 (GR_PRA2)
Golden Retriever (vetGen; Optigen; AHT; UCD)
Labrador Retriever (vetGen; UCD)

Tests for which the gene mutation has not been published:

Day Blindness/Retinal Degeneration (unpublished)

Standard Poodle, Goldendoodle, Labradoodle/Goldendoodle Cross, Labradoodle & Labradoodle (Optigen)

Italian Greyhound Progressive Retinal Atrophy IG_PRA1 in press

Italian Greyhound (Optigen)

Photoreceptor Dysplasia (Type A PRA) unpublished

Miniature Schnauzer (Optigen).

Caution: tests offered by other laboratories using a previously published variant in Phosducin should not be used; the Phosducin variant is a benign sequence change (polymorphism) and is not responsible for the Type A PRA (www.Optigen.com).

Type B PRA unpublished

Miniature Schnauzer (Optigen).

Weimaraner Progressive Retinal Atrophy

Weimaraner_PRA (unpublished)

Weimaraner (Bochum University)

DNA-testing Laboratories

Animal Health Trust (AHT) www.aht.org.uk

Bochum University www.ruhr-uni-bochum.de

Norwegian School of Veterinary Science (NSVS) Division of Genetics, Oslo, Norway

Optigen www.optigen.com

UC Davis School of Veterinary Medicine (UCD) www.vgl.ucdavis.edu/services/dog.php

University of Missouri (MU) www.caninegeneticdiseases.net/DNAtests/TESTmain.htm

vetGen www.vetgen.com

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