

# Canine inherited retinal diseases

	Gene	Disease designation	Mutation annotation	Mutation type	Inheritance	OMIA reference
<b>Canine multi-focal retinopathy</b>						<b>(n=3)</b>
<b>1</b>	<b>BEST1</b>	CMR1	c.C73T	Substitution	Autosomal Recessive	OMIA 001444-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Donner et al. (2019)	30657768	Donner et al.(2019)	27525650	Gornik et al. (2013)	23998685
	Guziewicz et al. (2007)	17460247	Hoffman et al. (2012)	22432598	Zangerl et al. (2010)	16938425
<u>Breed(s)</u>						
	Bull Mastiff		Mastiff		Pyrenean Mountain dog	
	Cane Corso		Dogue de Bordeaux		Anatolian Shepherd Dog	
	Boerboel		American Bulldog		American Pit Bull Terrier	
	Miniature American Shepherd		Brazilian Terrier		Bulldog	
	French Bull Dog		Perro de Presa Canario			
<b>2</b>	<b>BEST1</b>	CMR2	c.G482A	Substitution	Autosomal Recessive	OMIA 001553-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Miyadera et al. (2012)	22065099				
<u>Breed(s)</u>						
	Coton De Tulear					
<b>3</b>	<b>BEST1</b>	CMR3	c.C1388del	Deletion	Autosomal Recessive	OMIA 001554-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Donner et al. (2019)	30657768	Donner et al.(2019)	27525650	Zangerl et al. (2010)	16938425
<u>Breed(s)</u>						
	Finnish Lapphund		Lapponion Herder			

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<b>Early retinal degeneration</b>					<b>(n=1)</b>
1	<b>STK38L</b>	ERD	SINE ins20447905_20447906_315bp	Sine Insertion	Autosomal Recessive OMIA 001297-9615
<u>Publication(s) - Citation and Pubmed ID</u>					
Goldstein et al. (2010)		20887780			
<u>Breed(s)</u>					
Norwegian Elkhound					
<b>Progressive retinal atrophy</b>					<b>(n=39)</b>
1	<b>BBS4</b>	PRA_BBS4	c.58A>T exon 2	Substitution	Autosomal Recessive OMIA 002045-9615
<u>Publication(s) - Citation and Pubmed ID</u>					
Chew et al. (2017)		28533336			
<u>Breed(s)</u>					
Hungarian Puli					
2	<b>C2ORF71</b>	PRA_RCD4	c.3149_3150insC	Insertion	Autosomal Recessive OMIA 001575-9615
<u>Publication(s) - Citation and Pubmed ID</u>					
Downs et al. (2013)		22686255	Downs et al. (2014)	24255994	Karlskov-Mortenson et al (201
Svensson et al. (2016)		26009980	29624701		
<u>Breed(s)</u>					
Gordon Setter		Irish Setter	Chinese Crested		
Standard Poodle		English Setter	Tibetan Terrier		
Polish Lowland Sheepdog (polski owczarek		Old Danish Pointer	Polish Tatra Sheepdog		
3	<b>CCDC66</b>	Gpra	c.521_522insA	Insertion	Autosomal Recessive OMIA 001521-9615
<u>Publication(s) - Citation and Pubmed ID</u>					
Dekomien (2010)		19777273	Gerding et al. (2011)	21680557	Lippmann et al. (2007)
Schulte-Middelmann (2009)		17327822			
<u>Breed(s)</u>					
Schapendoes (Dutch)					

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<b>4</b>	<b>CNGA1</b>	PRA_CNGA1	c.1752_1755delAACT exon 9	Deletion	Autosomal Recessive	OMIA 001977-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
Wild et al. (2015)						
<u>Breed(s)</u>						
Shetland Sheepdogs						
<b>5</b>	<b>CNGB1</b>	PRA_PAP1	c.2685delA2687_2688insTAGCTA*	Insertion/Deletion	Autosomal Recessive	OMIA 000830-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
Ahonen et al (2013)		24015210	Donner et al. (2019)	30657768	Donner et al.(2019)	27525650
Winkler et al (2013)		23977260				
<u>Breed(s)</u>						
Papillon			Phalene	Griffon bruxellois		
<b>6</b>	<b>FAM161A</b>	PRA3	c.1758-15_1758-16ins238	Insertion	Autosomal Recessive	OMIA 001918-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
Downs et al. (2014)		24705771				
<u>Breed(s)</u>						
Tibetan Spaniel			Tibetan Terrier			
<b>7</b>	<b>IQCB1</b>	PRA_CRD2	c.952-953insC	Insertion	Autosomal Recessive	OMIA 001675-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
Donner et al. (2019)		30657768	Donner et al.(2019)	27525650	Goldstein et al. (2013)	24045995
<u>Breed(s)</u>						
American Staffordshire Bull Terrier			American Pit Bull Terrier			

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<b>8</b>	<b>PDE6B</b>	PRA_RCD1	c.2420G>A, p.W807X	Substitution	Autosomal Recessive	OMIA 000882-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Clements et al. (1993)	8261797	Djajadiningrat-Laanen et al. (20	12653325	Suber et ak. (1993)	8387203
<u>Breed(s)</u>						
	Irish Red and White Setter		Irish Setter			
<b>9</b>	<b>PDE6B</b>	PRA_RCD1	c.2448_2449insTGAAGTCC	Insertion	Autosomal Recessive	OMIA 001669-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Dekomien et al (2000)	11124530				
<u>Breed(s)</u>						
	Sloughie (Arabian Greyhound)					
<b>10</b>	<b>PDE6B</b>	PRA_RCD1	assoc with PDC	Substitution	Autosomal Recessive	
<u>Publication(s) - Citation and Pubmed ID</u>						
	Pandya et al. (2016)					
<u>Breed(s)</u>						
	Spitz					
<b>11</b>	<b>PDE6B</b>	PRA_RCD1	c.2404_2406delAAC	Deletion	Autosomal Recessive	OMIA 001674-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Donner et al. (2019)	30657768	Donner et al.(2019)	27525650	Goldstein et al. (2013)	24045995
<u>Breed(s)</u>						
	American Bulldog		American Staffordshire Bull Terrier			

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<b>12</b>	<b>PDE6A</b>	PRA_RCD3	c.1940delA	Deletion	Autosomal Recessive	OMIA 001314-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Donner et al. (2019)	30657768	Donner et al.(2019)	27525650	Downs et al. (2014)	24255994
	Petersen-Jones et al. (1999)	22550515				
<u>Breed(s)</u>						
	Chinese Crested		Pomeranian		Welsh Corgi (Cardigan)	
	Welsh Corgi (Pembroke)					

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<b>13</b>	<b>PRCD*</b>	PRA_PRC	c.G115A	Substitution	Autosomal Recessive	OMIA 001298-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Acland et al. (1998)	9501213	Aguirre and Acland (1998)	3164273	Dostal et al. (2011)	
	Downs et al. (2014)	24255994	Goldstein et al. (2006)		Miyadera et al. (2012)	22065099
	Moody et al. (2005)	16334947	Palanova et al. (2016)	30050836	Ray et al. (1996)	8603863
	Zangerl et al. (2006)	16938425				
<u>Breed(s)</u>						
	Chinese Crested		Miniature Poodle		Toy Poodle	
	Portuguese Water Dog		Chesapeake Bay Retriever		Labrador Retriever	
	Nova Scotia Duck Tolling Retrievers		Schipperke		American Cocker Spaniel	
	Cocker Spaniel		Australian Cattle Dog		Australian Shepherd	
	Miniature Australian Shepherd		Cockapoo		Entlebucher Sennenhund	
	Finnish Lapphund		Goldendoodle		Kuvasz	
	Labradoodle		Lapponion Herder		Markiesje	
	Norwegian Elkhound		Standard Poodle		Golden Retriever	
	Spanish Water Dog		Swedish Lapphund		Yorkshire Terrier	
	Australian Silky Terrier		Hairless terrier (American)		Barbet	
	Chihuahua		Coton De Tulear		Hungarian Puli	
	Jack Russell Terrier		Japanese Chin		Lagotto Romagnolo	
	Lancashire Heeler		Manchester Terrier		Pomeranian	
	Portuguese Podengo Pequeno (small)		Rat Terrier		Russian Black Terrier	
	Giant Schnauzer		Field Spaniel		Tibetan Terrier	
	Mexican Hairless Dog (Xoloitzcuintli)		American Eskimo Dog		Australian Stumpy Tailed Cattle Dog	
	Karelian Bear Dog		Bolognese		Bolonka Zvetna	
	Mi-Ki		Norrbottenspets		Plott (Hound)	
	Russian-European Laika		Serbian Hound		Swedish Elkhound	

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<b>14</b>	<b>RD3</b>	RCD2	p.Pro139Alafs*	Insertion	Autosomal Recessive	OMIA 001260-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Kukekova et al. (2006)	16505060	Kukekova et al. (2009)	19130129	Miyadera et al. (2012)	22065099
	Molday et al. (2014)	24904271	Perrault et al. (2014)	23308101		
<u>Breed(s)</u>						
	Rough Collie		Smooth Collie			
<b>15</b>	<b>RPGR</b>	XL_PRA2	c.1084-1085delGA*	Deletion	X Linked Recessive	OMIA 001518-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Zhang et al. (2002)	11978759				
<u>Breed(s)</u>						
	Samoyed		Siberian Husky			
<b>16</b>	<b>RPGR</b>	XL_PRA1	c.1028-1032delGAGAA?	Deletion	X Linked Recessive	OMIA 000831-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Andre et al. (2008)		Appelbaum et al. (2020)	33326016	Dave and Dave (2014)	
	Guyon et al. (2007)	17653054	Vilboux et al. (2008)	18315866	Zangerl et al. (2007)	17646274
	Zeiss et al. (1999)	10586956	Zeiss et al. (2000)	10699176	Zhang et al. (2002)	11978759
<u>Breed(s)</u>						
	Cross Breed					
<b>17</b>	<b>RHO</b>	DPRA	c. C11G	Substitution	Autosomal Dominant	OMIA 001346-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Kijas et al. (2002)	11972042	Kijas et al. (2003)	12692159	Miyadera et al. (2012)	22065099
	Palanova et al. (2016)	30050836				
<u>Breed(s)</u>						
	Bull Mastiff		Mastiff			

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18	<b>RPGRIP1</b>	PRA_CRD4	g.8228_8229insA29GGAAGCAACAG GATG	Insertion	Autosomal Recessive	OMIA 001432-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Busse et al. (2011)	21521437	Das et al. (2017)	28993665	Donner et al. (2019)	30657768
	Donner et al.(2019)	27525650	Downs et al. (2014)	24255994	Kuznetsova et al. (2012)	22183349
	Kuznetsova et al. (2012)	22807295	Mellersh et al. (2006)	16806805	Miyadera et al. (2009)	19936303
	Narfstrom et al. (2012)	22550515				
<u>Breed(s)</u>						
	Miniature Long-haired Dachshund		Beagle		French Bull Dog	
	Labrador Retriever		English Springer Spaniel		Cocker Spaniel	
	Chihuahua		German Spitz (Klein)		Field Spaniel	
	Curly Coated Retriever		Long-Haired Dachshund (Standard)		Miniature Smooth Haired Dachshund	
	Miniature Wire Haired Dachshund		Smooth Haired Dachshund (Standard)			
19	<b>SAG</b>	PRA_BAS	c1216T>C	Substitution	Autosomal Recessive	OMIA 001876-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Goldstein et al. (2013)	24019744				
<u>Breed(s)</u>						
	Basenji					
20	<b>SLC4A3</b>	GR-PRA1	c.2601_2602insC	Insertion	Autosomal Recessive	OMIA 001572-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Donner et al. (2019)	30657768	Donner et al.(2019)	27525650	Downs et al. (2011)	21738669
	Downs et al. (2014)	24255994	Miyadera (2014)	26120276		
<u>Breed(s)</u>						
	Golden Retriever		Lhasa Apso		Miniature Schnauzer	
	Labrador Retriever Cross		Goldendoodle			

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<b>21</b>	<b>TTC8</b>	GR-PRA2	c.669delA	Deletion	Autosomal Recessive	OMIA 001984-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Downs et al. (2014)	24255994	Downs et al. (2014)	24255994	Makelainen et al. (2020)	32962042
	Miyadera (2014)	26120276				
<u>Breed(s)</u>						
	Labrador Retriever Cross		Golden Retriever		Goldendoodle	
<b>22</b>	<b>NPHP4</b>	PRA_CRD	180bp del	Deletion	Autosomal Recessive	OMIA 001455-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Donner et al. (2019)	30657768	Donner et al.(2019)	27525650	Palanova et al. (2013)	23998563
	Ropstad et al. (2007)	17324160	Wild et al. (20009)	19392817	Wilk et al. (2008)	26202106
<u>Breed(s)</u>						
	Miniature Wire Haired Dachshund		Wire-haired Dachshund			
<b>23</b>	<b>NECAP1</b>	PRA	c.544G>A	Substitution	Autosomal Recessive	OMIA-002198-9615
<u>Publication(s) - Citation and Pubmed ID</u>						
	Hitti et al. (2019)	31117272				
<u>Breed(s)</u>						
	Giant Schnauzer		German Spitz (Klein)		German Spitz (Mittel)	
	German Spitz (Giant)		Pomeranian		Miniature Long-haired Dachshund	
<b>24</b>	<b>IMPG2</b>	PRA4	15bp ins.	Insertion	Autosomal Recessive	
<u>Publication(s) - Citation and Pubmed ID</u>						
	Hitti et al. (TBP)					
<u>Breed(s)</u>						
	Lhasa Apso					

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<b>25</b>	<b>PPT1</b>	PRA_PPT1	2866454_2877574	Duplication	Autosomal Recessive	
	<u>Publication(s) - Citation and Pubmed ID</u>					
	Murgiano et al (2019)	30541930	Murgiano et al. (2020)			
	<u>Breed(s)</u>					
	Miniature Schnauzer					
<b>26</b>	<b>PPT1</b>	PRA_PPT1	2874661_2875048	Conversion	Autosomal Recessive	
	<u>Publication(s) - Citation and Pubmed ID</u>					
	Murgiano et al (2019)	30541930	Murgiano et al. (2020)			
	<u>Breed(s)</u>					
	Miniature Schnauzer					
<b>27</b>	<b>PPT1</b>	PRA_PPT1	2877563_2877607	Inversion OR Insertion	Autosomal Recessive	
	<u>Publication(s) - Citation and Pubmed ID</u>					
	Murgiano et al (2019)	30541930	Murgiano et al. (2020)			
	<u>Breed(s)</u>					
	Miniature Schnauzer					
<b>28</b>	<b>ADAM9</b>	PRA_CRD3	23221bp del	Deletion	Autosomal Recessive	
	<u>Publication(s) - Citation and Pubmed ID</u>					
	Goldstein et al. (2010)	20806078	Kropatsch et al. (2010)	20691256		
	<u>Breed(s)</u>					
	Glen of Imaal Terrier		Labrador Retriever Cross		Soft-coated Wheaten Terrier	
<b>29</b>	<b>MERTK</b>	PRA_MERTK	6-8k	Insertion	Autosomal Recessive	
	<u>Publication(s) - Citation and Pubmed ID</u>					
	Ahonen et al (2014)	25517981	Everson et al. (2017)	28813472		
	<u>Breed(s)</u>					
	Swedish Vallhund					

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30	<b>BBS2</b>	PRA_BBS2	c.1222G>C p.A408P	Substitution	Autosomal Recessive	
<u>Publication(s) - Citation and Pubmed ID</u>						
<u>Breed(s)</u>						
Shetland Sheepdogs						
31	<b>HIVEP3</b>	PRA Type1	g.1432293G>A	Substitution	Autosomal Recessive	
<u>Publication(s) - Citation and Pubmed ID</u>						
Kaukonen et al. (2020)		32150541	Murgiano et al. (2020)			
<u>Breed(s)</u>						
Miniature Schnauzer						
32	<b>CCDC66</b>	EOPRA	c.2262_c.2263insA	Insertion	Autosomal Recessive	
<u>Publication(s) - Citation and Pubmed ID</u>						
Murgiano et al. (2020)		33273526				
<u>Breed(s)</u>						
Portuguese Water Dog						
33	<b>PDC</b>	Type 1 PRA	p.R82G C/G	Substitution	Autosomal Recessive	
<u>Publication(s) - Citation and Pubmed ID</u>						
Dekomien and Epplen (2002)		12091798	Jeong et al. (2008)	18724412	Zhang et al. (1998)	9714819
<u>Breed(s)</u>						
Miniature Schnauzer						
34	<b>PDC</b>	PRA	1 of 5 PDC SNPs T/A	Substitution	Autosomal Recessive	
<u>Publication(s) - Citation and Pubmed ID</u>						
Pandya et al. (2016)						
<u>Breed(s)</u>						
Spitz						

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<b>35</b>	<b>PDC</b>	PRA	2 of 5 PDC SNPs A/G	Substitution	Autosomal Recessive	
	<u>Publication(s) - Citation and Pubmed ID</u> Pandya et al. (2016)					
	<u>Breed(s)</u> Spitz					
<b>36</b>	<b>PDC</b>	PRA	3 of 5 PDC SNPs C/T	Substitution	Autosomal Recessive	
	<u>Publication(s) - Citation and Pubmed ID</u> Pandya et al. (2016)					
	<u>Breed(s)</u> Spitz					
<b>37</b>	<b>PDC</b>	PRA	4 of 5 PDC SNPs G/A	Substitution	Autosomal Recessive	
	<u>Publication(s) - Citation and Pubmed ID</u> Pandya et al. (2016)					
	<u>Breed(s)</u> Spitz					
<b>38</b>	<b>PDC</b>	PRA	5 of 5 PDC SNPs G/T	Substitution	Autosomal Recessive	
	<u>Publication(s) - Citation and Pubmed ID</u> Pandya et al. (2016)					
	<u>Breed(s)</u> Spitz					
<b>39</b>	<b>IFT122</b>	PRA	c.3176G>A	Substitution	Autosomal Recessive	
	<u>Publication(s) - Citation and Pubmed ID</u> Kaukonen et al. (2021)      33606121					
	<u>Breed(s)</u> Lapponion Herder					

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<b>Retinal degeneration</b>					<b>(n=1)</b>
1	<b>MAP9</b>	Early Onset Retinal Degeneration Modifier ( of CRD4/CORD1)	21961bp	Pseudogene Inversion	Autosomal Recessive
<u>Publication(s) - Citation and Pubmed ID</u>					
Forman et al. (2016)		27017229			
<u>Breed(s)</u>					
Miniature Long-haired Dachshund					
<b>Retinal Dysplasia</b>					<b>(n=1)</b>
1	<b>NDP</b>	RD	c.653_654insC	Insertion	X-Linked
<u>Publication(s) - Citation and Pubmed ID</u>					
Joyce et al. (2021)		33945575			
<u>Breed(s)</u>					
Cocker Spaniel					
<b>Stargardt macular degeneration</b>					<b>(n=1)</b>
1	<b>ABCA4</b>	STGD	g.55146549insC, c.4176insC, p.F1393Lfs*1395	Insertion	Autosomal Recessive OMIA 002179-9615
<u>Publication(s) - Citation and Pubmed ID</u>					
Mäkeläinen et al. (2019)		30889179			
<u>Breed(s)</u>					
Labrador Retriever Cross					

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<b>XL Progressive retinal atrophy</b>					<b>(n=1)</b>
1	<b>RPGR</b>	XLPRA	5kb	Deletion	Autosomal Recessive
<u>Publication(s) - Citation and Pubmed ID</u>					
	Kropatsch et al. (2016)	27398221			
<u>Breed(s)</u>					
	Weimaraner				