

Labrador Retriever



[Labgenvet](#)
[Donner, 2018](#)
[Donner, 2023](#)

Disease	Gene	Reference	# tests	M/N	
Stargardt Disease (Labrador Retriever)	ABCA4	Donner, 2023	16856	19.8%	
Exercise-Induced Collapse (EIC)	DNM1	Labgenvet, 2021	226	30.1%	
		Donner, 2018	476	25.8%	
		Donner, 2023	16853	18.9%	
Progressive Rod-Cone Degeneration (prcd-PRA)	PRCD	Labgenvet, 2021	66	10.6%	
		Donner, 2018	387	19.6%	
		Donner, 2023	16825	13.3%	
Oculoskeletal dysplasia (Labrador; OSD1)	COL9A3	Labgenvet, 2021	12	15.2%	
Hereditary Nasal Parakeratosis (Labrador Retriever; HNPk)	SUV39H2	Labgenvet, 2021	127	15.0%	
		Donner, 2018	476	12.0%	
		Donner, 2023	16854	3.9%	
Obesity, hereditary (Labrador Retriever)	POMC	Labgenvet, 2021	27	11.1%	DOMINANT
Centronuclear Myopathy (Labrador Retriever; CNM)	HACD1	Donner, 2018	386	4.9%	
		Donner, 2023	16849	2.0%	
Progressive Retinal Atrophy (Golden Retriever; GR_PRA2)	TTC8	Donner, 2018	387	0.8%	
		Donner, 2023	16856	1.8%	
Skeletal Dysplasia 2 (SD2)	COL11A2	Donner, 2018	476	3.4%	
		Donner, 2023	16856	1.7%	
Degenerative Myelopathy (DM)	SOD1	Labgenvet, 2021	49	4.1%	
		Donner, 2018	476	0.6%	
		Donner, 2023	16855	1.2%	
Chondrodystrophy, Intervertebral Disc Disease Risk (CDDY)	FGF4	Donner, 2023	16758	0.82%	DOMINANT
Congenital Myasthenic Syndrome (Labrador Retriever)	COLQ	Donner, 2023	16856	0.43%	
Ichthyosis (Golden Retriever)	PNPLA1	Donner, 2018	476	0.4%	
		Donner, 2023	16856	0.4%	
Cone-Rod Dystrophy (cord1-PRA/crd4)	RPGRIP1	Donner, 2018	476	0.2%	
		Donner, 2023	16829	0.36%	
Collie Eye Anomaly (CEA)	NHEJ1	Donner, 2018	476	0.2%	
		Donner, 2023	16856	0.2%	
Canine Scott Syndrome (CSS)	ANO6	Donner, 2023	16855	0.18%	
Hyperuricosuria (HUU)	SLC2A9	Donner, 2023	16856	0.15%	
Cystinuria Type I-B (SLC7A9 p.A217T)	SLC7A9	Donner, 2023	16853	0.14%	
MDR1 (Multidrug Resistance 1) Medication Sensitivity	ABCB1	Donner, 2023	16853	0.1%	DOMINANT
Dilated Cardiomyopathy risk factor (PDK4-related)	PDK4	Donner, 2023	16856	0.1%	
Von Willebrand's Disease, Type 1 (vWD 1)	VWF	Donner, 2023	16855	0.08%	
Narcolepsy (Labrador Retriever)	HCRTR2	Donner, 2023	16856	0.06%	
Cystinuria Type I-A (SLC3A1 p.I192V)	SLC3A1	Donner, 2023	16855	0.03%	
Factor VII Deficiency	F7	Donner, 2023	16855	0.03%	
Dilated Cardiomyopathy (Schnauzer; DCM)	RBM20	Donner, 2023	16856	0.03%	
Progressive Retinal Atrophy (GR_PRA1)	SLC4A3	Donner, 2023	16855	0.02%	

Neuronal Ceroid Lipofuscinosis 4A (NCL4A)	ARSG	Donner, 2023	16854	0.02%
Cystinuria Type I-A (SLC3A1 p.S698G)	SLC3A1	Donner, 2023	16856	0.02%
Primary Lens Luxation (PLL)	ADAMTS17	Donner, 2023	16856	0.02%
Musladin-Lueke syndrome (MLS), Geleophysic dysplasia	ADAMTS2	Donner, 2023	16843	0.01%
Protein Losing Nephropathy (PLN; NPHS1-related)	NPHS1	Donner, 2023	16855	0.01%
Acral Mutilation Syndrome (AMS)	GDNF	Donner, 2023	16856	0.01%
Familial Nephropathy (English Cocker Spaniel; FN)	COL4A4	Donner, 2023	16856	0.01%
Ichthyosis (American Bulldog; NIPAL4-related)	NIPAL4	Donner, 2023	16856	0.01%

Mutation reported at very low frequency (Donner 2023):

Progressive Retinal Atrophy (Shetland Sheepdog; CNGA1-PRA)	CNGA1	Donner, 2023	16076	>0.01%
Deafness and Vestibular Dysfunction (Doberman Pinscher)	PTPRQ	Donner, 2023	16850	>0.01%
Hypomyelination (Weimaraner)	FNIP2	Donner, 2023	16854	>0.01%
Canine Multifocal Retinopathy 1 (Mastiffs; CMR1)	BEST1	Donner, 2023	16855	>0.01%
Chondrodysplasia, Disproportionate Short-limbed	ITGA10	Donner, 2023	16855	>0.01%
Complement 3 (C3) Deficiency	C3	Donner, 2023	16856	>0.01%
Cone Degeneration (German Shorthaired Pointer)	CNGB3	Donner, 2023	16856	>0.01%
Cystinuria Type II-A (Australian Cattle Dog)	SLC3A1	Donner, 2023	16856	>0.01%
Deafness and Vestibular Dysfunction (Doberman Pinscher)	PTPRQ	Donner, 2023	16856	>0.01%
Hypocatalasia	CAT	Donner, 2023	16856	>0.01%
Macrothrombocytopenia (Norfolk, Cairn Terrier)	TUBB1	Donner, 2023	16856	>0.01%
Neuroaxonal Dystrophy (Rottweiler; VPS11-related)	VPOS11	Donner, 2023	16856	>0.01%
Neuronal Ceroid Lipofuscinosis 5 (Border Collie; NCL5)	CLN5	Donner, 2023	16856	>0.01%
Polycystic Kidney Disease (Bull Terrier)	PKD1	Donner, 2023	16856	>0.01%
Rod-Cone Dysplasia 1 (Irish Setter; rcd1)	PDE6B	Donner, 2023	16856	>0.01%
Shar-Pei Autoinflammatory Disease (SPAID)	MTBP, HAS2	Donner, 2023	16856	>0.01%