

Golden Retriever



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

Disease	Gene	Reference	# tests	M/N	
Ichthyosis (Discovered in the Golden Retriever)	PNPLA1	Labgenvet, 2021	180	41.7%	
		Donner, 2018	331	35.0%	
		Donner, 2023	12881	39.9%	
Cystinuria Type I-B (SLC7A9 p.A217T)	SLC3A1	Donner, 2023	12861	36.2%	
Progressive Retinal Atrophy (GR PRA2)	TTC8	Donner, 2018	175	2.3%	
		Donner, 2023	12881	5.5%	
Progressive Rod-Cone Degeneration (prcd-PRA)	PRCD	Donner, 2018	175	0.6%	
		Donner, 2023	12860	5.2%	
Progressive Retinal Atrophy (GR PRA1)	SLC4A3	Labgenvet, 2021	46	19.6%	
		Donner, 2018	328	8.5%	
		Donner, 2023	12860	3.1%	
Congenital Myasthenic Syndrome (CHRNE-related; CMS)	CHRNE	Donner, 2023	436	0.46%	DOMINANT
Chondrodystrophy, Intervertebral Disc Disease Risk (CDDY)	FGF4	Donner, 2023	12842	0.36%	
Degenerative Myelopathy (DM)	SOD1	Labgenvet, 2021	33	0.06%	
		Donner, 2023	12881	0.35%	
Acral Mutilation Syndrome (AMS)	GDNF	Donner, 2023	12881	0.14%	
Congenital Myasthenic Syndrome (COLQ-related)	COLQ	Donner, 2023	12881	0.14%	
Exercise-Induced Collapse (EIC)	DNM1	Donner, 2023	12879	0.12%	
Von Willebrand's Disease, Type 1 (vWD 1)	VWF	Donner, 2023	12881	0.1%	
Progressive Retinal Atrophy (MERTK-related)	MERTK	Donner, 2023	12881	0.07%	
MDR1 (Multidrug Resistance 1) Medication Sensitivity	ABCB1	Donner, 2023	12880	0.05%	
Early-Onset Progressive Polyneuropathy (Greyhound)	NDRG1	Donner, 2023	12877	0.04%	
Collie Eye Anomaly (CEA)	NHEJ1	Donner, 2023	12881	0.04%	
Stargardt Disease (Labrador Retriever)	ABCA4	Donner, 2023	12881	0.03%	
Factor VII Deficiency	F7	Donner, 2023	12881	0.02%	
Cone-Rod Dystrophy (cord1-PRA/crd4)	RPGRIP1	Donner, 2018	330	0.3%	
		Donner, 2023	12853	0.06%	
Rod-Cone Dysplasia 1a (Sloughi; rcd1a)	PDE6B	Donner, 2023	12875	0.016%	
Hyperuricosuria (HUU)	SLC2A9	Donner, 2023	12879	0.016%	
Congenital Myasthenic Syndrome (CHRNE-related; CMS)	CHRNE	Donner, 2023	12880	0.016%	
Canine Multifocal Retinopathy 1 (CMR1)	CMR1	Donner, 2023	12881	0.016%	DOMINANT
Dilated Cardiomyopathy risk factor (PDK4-related)	PDK4	Donner, 2023	12881	0.016%	
Neuronal Ceroid Lipofuscinosis 8 (Saluki; NCL8)	CLN8	Donner, 2023	12881	0.016%	
Mutations reported at very low frequency (Donner, 2023):					
Van den Ende-Gupta Syndrome (VDEGS)	SCARF2	Donner, 2023	12875	>0.01%	X-LINKED
Musladin-Lueke syndrome (MLS)	ADAMTSL2	Donner, 2023	12876	>0.01%	
Dental Hypomineralisation (Border Collie)	FAM20C	Donner, 2023	12877	>0.01%	
Dystrophic Epidermolysis Bullosa (Golden Retriever)	COL7A1	Donner, 2023	12878	>0.01%	
Muscular Dystrophy (Golden Retriever)	DMD	Donner, 2023	12878	>0.01%	

Acute Respiratory Distress Syndrome (Dalmatian; ARDS)	ANLN	Donner, 2023	12879	>0.01%	DOMINANT
Dilated Cardiomyopathy risk factor (TTN-related)	TTN	Donner, 2023	12881	>0.01%	
Early-Onset Progressive Polyneuropathy (Alaskan Malamute)	NDRG1	Donner, 2023	12881	>0.01%	
Episodic Falling (EF)	BCAN	Donner, 2023	12881	>0.01%	DOMINANT
Hereditary Elliptocytosis	SPTB	Donner, 2023	12881	>0.01%	
Macrothrombocytopenia (Norfolk, Cairn Terrier)	TUBB1	Donner, 2023	12881	>0.01%	