

Great Pyrenees



[Labgenvet](#)

[Donner, 2018](#)

[Donner, 2023](#)

Disease	Gene	Reference	# tests	M/N	
Canine Multifocal Retinopathy 1 (Mastiffs; CMR1)	BEST1	Donner, 2023	1985	20.8%	
Degenerative Myelopathy (DM)	SOD1	Donner, 2023	1985	15.2%	
Glanzmann Thrombasthenia, Type I (Great Pyrenees)	ITGA2B	Donner, 2023	1985	1.2%	
Collie Eye Anomaly (CEA)	NHEJ1	Donner, 2023	1985	1.0%	
Progressive Rod-Cone Degeneration (prcd-PRA)	PRCD	Donner, 2023	1982	0.80%	
Hyperuricosuria (HUU)	SLC2A9	Donner, 2023	1985	0.80%	
Dilated Cardiomyopathy risk factor (TTN-related)	TTN	Donner, 2023	1985	0.60%	DOMINANT
Dilated Cardiomyopathy risk factor (PDK4-related)	PDK4	Donner, 2023	1985	0.50%	DOMINANT
MDR1 (Multidrug Resistance 1) Medication Sensitivity	ABCB1	Donner, 2023	1985	0.40%	DOMINANT
Cystinuria Type I-B (SLC7A9 p.A217T)	SLC7A9	Donner, 2023	1985	0.25%	
Chondrodystrophy and Intervertebral Disc Disease Risk (CDDY)	FGF4	Donner, 2023	1981	0.20%	DOMINANT
Factor VII Deficiency	F7	Donner, 2023	1985	0.20%	
Ichthyosis (Discovered in the Golden Retriever)	PNPLA1	Donner, 2023	1985	0.20%	
Cystinuria Type I-A (SLC3A1 p.I192V)	SLC3A1	Donner, 2023	1985	0.15%	
Canine Scott Syndrome (CSS)	ANO6	Donner, 2023	1985	0.10%	
Cone-Rod Dystrophy (cord1-PRA/crd4)	RPGRIP1	Donner, 2023	1985	0.10%	
Stargardt Disease (Labrador Retriever)	ABCA4	Donner, 2023	1985	0.10%	
Early-Onset Progressive Polyneuropathy (Greyhound)	NDRG1	Donner, 2023	1984	0.05%	
Dilated Cardiomyopathy (Schnauzer; DCM)	RBM20	Donner, 2023	1985	0.05%	
Exercise-Induced Collapse (EIC)	DMN1	Donner, 2023	1985	0.05%	
Neuronal Ceroid Lipofuscinosis 5 (Border Collie; NCL5)	CLN5	Donner, 2023	1985	0.05%	
Primary Lens Luxation (PLL)	ADAMTS17	Donner, 2023	1985	0.05%	
Progressive Retinal Atrophy (Golden Retriever; GR_PRA1)	SLC4A3	Donner, 2023	1985	0.05%	

Mutation reported at very low frequency (Donner 2023):
