



**Ocular disorders known or presumed to be inherited (published)**

	<b>Diagnosis</b>	<b>Description and comments specific to the breed</b>	<b>Inheritance</b>	<b>Gene/ marker test</b>	<b>References</b>
<b>A</b>	Microphthalmia	Associated with other ocular anomalies (cataract, retinal dysplasia, neovascularization of the retina, optic nerve hypoplasia)	Autosomal dominant	SIX6	1,2,27,29
<b>B</b>	Uveal cysts	Iris and ciliary body cysts ;2-6 y.o. ; possible relationship with glaucoma	Unknown	NO	1,2,3,5,6
<b>C</b>	Pigmentary uveitis	Both eyes are frequently affected ;4,5-14,5 y.o.; pigmented radial striae on the anterior capsule of the lens ; associated uveal cysts ; fibrin in the anterior chamber, cataract and glaucoma in severe cases	Unknown	NO	2,4,6

<b>D</b>	Glaucoma	Pectinate Ligament Abnormality	Unknown	NO	28
<b>E</b>	Cataract	1.Triangular subcapsular posterior cataract ; between 6 and 18 months of age ; usually slowly or non progressive  2.Anterior cortical striate cataract ; usually 3-6 y.o.  3.Perinuclear posterior cataract ;rarely progressive	1.Autosomal recessive  2.Unknown  3.Unknown	NO	1,2,7,8,9,10,11,12,13,14
<b>F</b>	Retinal dysplasia -multifocal -geographic		Unknown	NO	1,2,15,16
<b>G</b>	Progressive Retinal Atrophy 1.PRA-prcd 2.GR_PRA1 3.GR_PRA2	1.Two different forms : a)within 2 years of age b)5-7 y.o.	Autosomal recessive (all of them)	1.prcd 2.SLC4A3 3.TTC8	1,2,17,18,19,20,21,22
<b>H</b>	Retinal Pigment Epithelial Dystrophy (RPED)		Unknown	NO	1,2,18,19
<b>I</b>	Limbal melanoma	Usually 3-7 y.o.	Unknown	NO	2,25,26

### **The ECVO's advice relating to hereditary eye disease control**

Please see ECVO Manual chapter 8: VET Advice

### **Recommendations regarding age and frequency for eye examinations**

Please see ECVO Manual chapter 7: ECVO Age and Frequency recommendations

### **Other ocular disorders (reported)**

	<b>Diagnosis</b>	<b>Source</b>
<b>A</b>	Distichiasis	ACVO geneticscommittee
<b>B</b>	Entropion	ACVO geneticscommittee
<b>C</b>	Cornealdystrophy -epithelial/stromal	ACVO geneticscommittee
<b>E</b>	Persistent pupillary membranes -iris to iris	ACVO genetics committee
<b>F</b>	Atresia of lacrimal punctum	French National Panel
<b>G</b>	Persistent hyaloid artery	ACVO genetics committee
<b>H</b>	Vitreous degeneration	ACVO genetics committee

### **References**

1. Rubin Lionel F., Inherited Eye Diseases in Purebred Dogs, Baltimore: Williams & Wilkins, 1989;150-159.
2. Chaudieu G. Chahory S. Affections oculaires héréditaires ou à prédisposition raciale chez le chien.2nd ed. Ed. du Point Vétérinaire, 2013;373-380.

3. Townsend WM, Rossman P. Prevalence of pigmentary uveitis and uveal cysts in golden retrievers residing within the midwestern United States. 42nd Annual Meeting of the American College of Veterinary Ophthalmologists 2011 October 26–29, 2011.
4. Sapienza JS, Simo FJ, Prades-Sapienza A. Golden Retriever uveitis: 75 cases (1994-1999). *VetOphthalmol.* 2000; 3: 241-246.
5. Deehr AJ, Dubielzig RR. A histopathological study of iridociliary cysts and glaucoma in Golden Retrievers. *VetOphthalmol.* 1998; 1: 153-158.
6. Holly V, Sandmeyer LS, Bauer BS, Verges L & Grahn BH. Golden Retriever cystic uveal disease: a longitudinal study of iridociliary cysts, pigmentary uveitis and pigmentary/cystic glaucoma over a decade in western Canada. *VetOphthalmol* 2015; published online 29 Jun 2015
7. Bona A. Eine population genetische Untersuchung zur Zuchtsituation und zerblich determinierten Erkrankun- insbesondere Augen- und Gelenkerkrankungen- beim Golden und Labrador Retriever. (A population genetic study of the breeding situation and inherited diseases, particularly eye and joint diseases, in the Golden and Labrador retrievers.). Tierärztliche Hochschule Hannover: Hannover Germany. 1995.
8. Gelatt KN. Cataracts in the Golden Retriever dog. *Vet Med Small AnimClin.* 1972; 67: 1113-1115.
9. Rubin LF. Cataract in Golden Retrievers. *J Am Vet Med Assoc.* 1974; 165: 457-458.
10. Barnett KC. Hereditary cataract in the dog. *J Small AnimPract.* 1978; 19: 109-120.
11. Barnett KC. Cataract in the golden retriever. *VetRec.* 1980; 111: 315.
12. Barnett KC. The diagnosis and differential diagnosis of cataract in the dog. *J Small AnimPract.* 1985; 26: 305.
13. Curtis R. Hereditary cataract in Golden and Labrador retrievers in the United Kingdom. *Trans Am CollVetOphthalmol.* 1986; 17: 23.
14. Aguirre G, Holle D, Yu-Speight A, Sarna C & Hall E. A re-examination of the mode of inheritance of posterior cortical cataracts in Labrador and Golden Retrievers. *Trans Am CollVetOphthalmol.* 2004; 35

15. Long SE, Crispin SM. Inheritance of multifocal retinal dysplasia in the golden retriever in the UK. *VetRec.* 1999; 145: 702-704.
16. Holle DM, Stankovics ME, Sarna CS, et al. The geographic form of retinal dysplasia in dogs is not always a congenital abnormality. *VetOphthalmol.* 1999; 2: 61-66.
17. Gelatt KN. Description and diagnosis of progressive retinal atrophy. *Norden News.* 1974; 24.
18. Acland GM, Ray K, Mellersh CS, Gu W, Langston AA, Rine J, Ostrander EA, Aguirre GD. Linkage analysis and comparative mapping of canine progressive rod-cone degeneration (*prcd*) establishes potential locus homology with retinitis pigmentosa (RP17) in humans. *Proceeding of the National Academy of Sciences of the United States of America* (1998): 95, 3048–3053.
19. Acland GM, Ray K, Mellersh CS, Landston AA, Rine J, Ostrander EA, Aguirre GD. A novel retinal degeneration locus identified by linkage and comparative mapping of canine early retinal degeneration. *Genomics* (1999) 59, 134–142.
20. Zangerl B, Goldstein O, Philp AR, Lindauer SJ, Pearce-Kelling SE, Mullins RF, Graphodatsky AS, Ripoll D, Felix JS, Stone EM, Acland GM, Aguirre GD. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics* (2006) 88(5):551-63.
21. Downs LM, Wallin-Håkansson B, Boursnell M, et al. A frameshift mutation in golden retriever dogs with progressive retinal atrophy endorses *SLC4A3* as a candidate gene for human retinal degenerations. *PLoS One.* 2011; 6: e21452.
22. Downs LM, Wallin-Håkansson B, Boursnell M, Bergström T, Mellersh CS. A novel mutation in *TTC8* is associated with progressive retinal atrophy in the golden retriever. *Canine Genetics and Epidemiology* (2014) 1,4:1-12.
23. Barnett KC. Canine retinopathies III. The other breeds. *J Small AnimPract.* 1965; 6: 185.
24. Parry HB. Degenerations of the dog retina VI. CPRA with pigment epithelial dystrophy. *Br J Ophthalmol.* 1954; 38.
25. Donaldson D, Sansom J, Scase T, Adams V & Mellersh. Canine limbal melanoma: 30 cases (1992-2004). Part 1. Signalment, clinical and histological features and pedigree analysis. *Vet Ophthalmol.* 2006; 9:115-119.

26. Donaldson D, Sansom J, Adams V. Canine limbal melanoma: 30 cases (1992-2004). Part 2. Treatment with lamellar resection and adjunctive strontium-90beta plesiotherapy- efficacy and morbidity. *VetOphthalmol.* 2006; 9: 179-185.
27. Williams LW, Peiffer RL, Gelatt KN et al. Multiple ocular defects in a Golden Retriever puppy. *Vet Med Small AnimClin.* 1977;72:1463-1465.
28. Oliver JAC, Ekiri AB, Mellersh CS. Pectinate ligament dysplasia in the Border Collie , Hungarian Vizsla and Golden Retriever. *Vet Record* 180 (11): 279, 2017.
29. Hug P, AndereggL, Durig N, Lepori V et al. A SIX6 Nonsense variant in Golden Retrievers with Congenital Eye Malformations. *Genes* 2019, 10(6), 454.