

Ocular disorders known or presumed to be inherited (published)

| | Diagnosis | Description and comments specific to the breed | Inheritance | Gene/ marker test | References |
|---|--|--|--------------------------|----------------------|-------------------------------------|
| A | Entropion | Most cases in puppies 5 months of age or less ; usually lower lateral eyelid | Unknown | NO | 1,2 |
| в | Cataract | Triangular posterior polar subcapsular cataract ; between 6 and 18 months of age ; slowly progressive | Unknown | NO | 1,2 |
| С | Retinal pigment epithelial dystrophy (RPED) | | Unknown | NO | 1,2,29 |
| D | Progressive Retinal Atrophy (PRA) | 1. Rod-cone dysplasia type1 (rcd1):Night blindness by 6 weeks of age; by 1- 2 years of age most affected dogs are completely blind. | 1.Autosomal recessive | 1.PDE6B | 1,2,4-24 2. 2,3,6 3. 1,2,4,25 |
| | | 2. Rod-cone dysplasia type4 (rcd4): Late onset; clinical signs from | 2. Autosomal recessive | 2. c2orf71 | |

| | | 10-12 years of age.3. Clinical signs between 3 and 5 years of age (familial non-rcd1) | 3.Unknown | 3. NO | |
|---|--------------------------------------|--|------------------------|-------|-----------|
| E | Amblyopia with quadriplegia | Puppies blind and not able to stand; often nystagmus is present; they are known as "swimmers" | Unknown | NO | 1,2,26,27 |
| F | Neuronal ceroid lipofuscinosis | Degenerative disease of the retinal visual cells leading to blindness | Autosomal recessive | CLN8 | 28 |

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)

| | Diagnosis | Source | |
|---|---|--|--|
| Α | Microphthalmia | French national panel | |
| в | Distichiasis | French national panel ACVO genetics committee | |
| с | Eversion of the cartilage of the nictitating membrane | ACVO genetics committee | |
| D | Persistent pupillary membranes -iris to iris | ACVO genetics committee | |

| Е | Persistent hyaloid artery | ACVO genetics committee |
|---|---------------------------|-------------------------|
| F | Retinal dysplasia - folds | ACVO genetics committee |

References

- 1. Rubin LF. Inherited eye diseases in purebred dogs. Williams &Wilkins 1989;169-174.
- 2. Chaudieu G. Chahory S. Affections oculaires héréditaires ou à prédisposition raciale chez le chien. 2nd ed. Ed. du Point Vétérinaire 2013;351-355.
- 3. Downs LM, Bell JS, Freeman J, Hartley C, Hayward LJ, Mellersh CS. Late-onset progressive retinal atrophy in the Gordon and Irish Setter breeds is associated with a frameshift mutation in C2orf71. Anim Genet (2013) 44(2):169-77.
- 4. Hodgman SSea. Progressive retinal atrophy in dogs I. The disease of Irish Setters (rcd). Vet Rec. 1949; 61.
- 5. Parry HB. Degenerations of the dog retina. II. Generalized progressive atrophy of hereditary origin. Br J Ophthalmol. 1953; 37: 487-502.
- 6. Aguirre GD, Rubin LF. Rod-cone dysplasia (progressive retinal atrophy) in Irish setters. J Am Vet Med Assoc. 1975; 166.
- 7. Aguirre G, Farber D, Lolley R, et al. Rod-cone dysplasia in Irish setters: a defect in cyclic GMP metabolism in visual cells. Science. 1978; 201: 1133-1134.
- 8. Lewis DG. [Reappearance of PRA in the Irish setter]. Vet Rec. 1977; 101: 122-123.
- Liu YP, Krishna G, Aguirre G, et al. Involvement of cyclic GMP phosphodiesterase activator in an hereditary retinal degeneration. Nature. 1979; 280: 62-64.
- Aguirre G, Farber D, Lolley R, et al. Retinal degeneration in the dog. III. Abnormal cyclic nucleotide metabolism in rod-cone dysplasia. Exp Eye Res. 1982; 35: 625-642.
- Lee RH, Lieberman BS, Hurwitz RL. Phosphodiesterase probes show distinct defects in rd mice and Irish setter dog disorders. Invest Ophthalmol Vis Sci. 19185; 26: 1985.

- 12. Biochemical and immunological characteristics of photoreceptor phosphodiesterase in inherited retinal degeneration of rd mice and affected Irish setter dogs. In LaVail MM, Hollyfield JG, Anderson RE (eds): Retinal Degeneration: Experimental and Clinical Studies. New York, Alan R. Liss, pp133, 1985.
- 13. Schmidt SY, Aguirre GD. Reduction in taurine secondary to photoreceptor loss in Irish setters with rod-cone dysplasia. Invest Ophthalmol Vis Sci. 1985; 1985.
- 14. Fletcher RT, Sanyal S, Krishna G, et al. Genetic expression of cyclic GMP phosphodiesterase activity defines abnormal photoreceptor differentiation in neurological mutants of inherited retinal degeneration. J Neurochem. 1986; 46: 1240-1245.
- 15. Schmidt SY, Andley UP, Heth CA, et al. Deficiency in light-dependent opsin phosphorylation in Irish setters with rod-cone dysplasia. Invest Ophthalmol Vis Sci. 1986; 27: 1551-1559.
- 16. Barbehenn E, Gagnon C, Noelker D, et al. Inherited rod-cone dysplasia: abnormal distribution of cyclic GMP in visual cells of affected Irish setters. Exp Eye Res. 1988; 46: 149-159.
- 17. Cunnick J, Rider M, Takemoto LJ, et al. Rod/cone dysplasia in Irish setters. Presence of an altered rhodopsin. Biochem J. 1988; 250: 335-341.
- Farber DB, Danciger JS, Aguiree GD. Early mRNA defect in Irish setter dog retina. Invest Ophthalmol Vis Sci (Suppl). invest Ophthalmol Vis Sci (Supp). 1990; 31.
- 19. Farber DB, Danciger JS, Aguiree GD. The B subunit of cyclic GMP phosphodiesterase mRNA is deficient in canine rod-cone dysplasia 1. Neuron. 1992; 9.
- 20. Clements PJ, Gregory CY, Peterson-Jones SM, et al. Confirmation of the rod cGMP phosphodiesterase beta subunit (PDE beta) nonsense mutation in affected rcd-1 Irish setters in the UK and development of a diagnostic test. Curr Eye Res. 1993; 12: 861- 866.
- 21. Suber ML, Pittler SJ, Qin N, et al. Irish setter dogs affected with rod/cone dysplasia contain a nonsense mutation in the rod cGMP phosphodiesterase beta-subunit gene. Proc Natl Acad Sci U S A. 1993; 90: 3968-3972.

- 22. Ray K, Baldwin VJ, Acland GM, et al. Cosegregation of codon 807 mutation of the canine rod cGMP phosphodiesterase beta gene and rcd1. Invest Ophthalmol Vis Sci. 1994; 35: 4291-4299.
- 23. Ray K, Baldwin VJ, Acland GM, et al. Molecular diagnostic tests for ascertainment of genotype at the rod cone dysplasia 1 (rcd1) locus in Irish setters. Curr Eye Res. 1995; 14: 243-247.
- 24. Petersen-Jones SM, Clements PJ, Barnett KC, et al. Incidence of the gene mutation causal for rod-cone dysplasia type 1 in Irish setters in the UK. J Small Anim Pract. 1995; 36: 310-314.
- 25. Djajadiningrat-Laanen SC, Boeve MH, Stades FC, et al. Familial non-rcd1 generalised retinal degeneration in Irish setters. J Small Anim Pract. 2003; 44: 113-116.
- 26. Palmer AC, Payne JE, Wallace ME. Hereditary quadriplegia and amblyopia in the Irish Setter. J Small Anim Pract. 1973; 14: 343-352.
- 27. Sakai T, Harashima T, Yamamura H, et al. Two cases of hereditary quadriplegia and amblyopia in a litter of Irish setters. J Small Anim Pract. 1994; 35.
- 28. Katz ML, Khan S, Awano T, Shahid SA, Siakotos AN, Johnson GS. A mutation in the CLN8 gene in English Setter dogs with neuronal ceroid-lipofuscinosis. Biochemical and Biophysical Research Communications (2005) 327,2:541–547.
- 29. Priester WA. Canine progressive retinal atrophy: occurrence by age, breed and sex. Amer J Vet Res 1974 ; 35:571-574.