

Australian Shepherd (Standard, Miniature and Toy)			
--	--	--	--

Ocular disorders known or presumed to be inherited (published)

	Diagnosis	Description and comments specific to the breed	Inheritance	Gene/ marker test	References
A	Microphtalmia with multiple anomalies	Suspected to be associated with merle coat coloration	Unknown	NO	1,2,3,4,5,6,7
B	Cataract	-Posterior polar subcapsular/ Nuclear cataract that progresses to complete cataract	Autosomal dominant, with incomplete penetrance	HSF4-2	1,7,8,9
C	Progressive Retinal Atrophy (PRA)		Autosomal recessive	prcd	1,7
D	Collie Eye Anomaly (CEA)		Autosomal recessive for CH/CRD; for coloboma unknown	NHEJ1	1,7,10,11,12,13
E	Canine multi-focal Retinopathy (CMR1)		Autosomal recessive	BEST1	14,15

F	Cone degeneration (achromatopsia)		Autosomal recessive	CNGB3	16
G	Retinal dysplasia - multifocal		Autosomal recessive	NO	1,7
H	Neuronal ceroid lipofuscinosis			CLN6	17

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
A	Distichiasis	ACVO genetics committee
B	Corneal dystrophy	ACVO genetics committee
C	Iris coloboma/iris hypoplasia	ACVO genetics committee
D	Persistent pupillary membranes	ACVO genetics committee
E	Persistent hyaloid artery	ACVO genetics committee
F	Coloboma/staphyloma without microphthalmia	ACVO genetics committee
G	Micropapilla	ACVO genetics committee
H	Vitreous degeneration	ACVO genetics committee

References

1. Rubin LF: Inherited Eye Diseases in Purebred Dogs, Williams & Wilkins, 1989; 317-321
2. Gelatt KN, McGill LD. Clinical characteristics of microphthalmia with colobomas of the Australian shepherd dog. J Am Vet Med Assoc. 1971; 162.
3. Gelatt KN, Veith LA. Hereditary multiple ocular anomalies in Australian shepherd dogs. Vet Med Small Anim Clin. 1970; 65.
4. Cook CS, Burling K, Nelson EJ. Embryogenesis of posterior segment colobomas in the Australian shepherd dog. Prog in Vet Comp Ophthalmol. 1991; 1.
5. Bertram T, Coignoul F, Cheville N. Ocular dysgenesis in Australian shepherd dogs. J Am Anim Hosp Assoc. 1984; 20: 177.
6. Gelatt KN, Powell NG. Inheritance of microphthalmia with coloboma. Am J Vet Res. 1981; 1.
7. Chaudieu G, Chahory S Affections oculaires héréditaires ou à prédisposition raciale chez le chien. 2nd ed, Ed. du Point Vétérinaire, 2013, pp.105-109.
8. Mellersh CS, Pettitt L, Forman OP, et al. Identification of mutations in HSF4 in dogs of three different breeds with hereditary cataracts. Vet Ophthalmol. 2006; 9: 369-378.
9. Mellersh CS, McLaughlin B, Ahonen S, et al. Mutation in HSF4 is associated with hereditary cataract in the Australian Shepherd. Vet Ophthalmol. 2009; 12: 372-378.
10. Rubin LF, Nelson EJ, Sharp CA. Collie eye anomaly in Australian shepherd dogs. Prog in Vet Comp Ophthalmol. 1991; 1.
11. Lowe JK, Kukekova AV, Kirkness EF, et al. Linkage mapping of the primary disease locus for collie eye anomaly. Genomics. 2003; 82: 86-95.

12. Parker HG, Kukekova AV, Akey DT, et al. Breed relationships facilitate fine-mapping studies: a 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. *Genome Res.* 2007; 17: 1562-1571.
13. Munyard KA, Sherry CR, Sherry L. A retrospective evaluation of congenital ocular defects in Australian Shepherd dogs in Australia. *Vet Ophthalmol.* 2007; 10: 19-22.
14. Guziewicz KE, Zangerl B, Lindauer SJ, et al. Bestrophin gene mutations cause canine multifocal retinopathy: A novel animal model for best disease. *Investigative Ophthal & Visual Science*, 48:1959-1967, 2007.
15. Hoffmann I, Guziewicz KE, Zangerl B, Aguirre GD, Mardin CY. Canine multifocal retinopathy in the Australian Shepherd: a case report. *Vet Ophthalmol* (2012) 15,2:134-8.
16. Sidjanin DJ, Lowe JK, McElwee JL, et al. Canine CNGB3 mutations establish cone degeneration as orthologous to the human achromatopsia locus ACHM3. *Human Molecular Genetics*. 2002; 11: 1823-1833.
17. Katz ML, Farias FH, Sanders DN, Zeng R, Khan S, Johnson GS, O'Brien DP. A Missense Mutation in Canine CLN6 in an Australian Shepherd with Neuronal Ceroid Lipofuscinosis. *Journal of Biomedicine and Biotechnology*. Vol 2011, Article ID 198042, 6 pages doi:10.1155/2011/198042.