AUSTRALIAN LABRADOODLE

(Labradoodle, Australian Cobber Dog)

	DISORDER	INHERITANCE	REFERENCE	BREEDING ADVICE	GENETIC TESTS AVAILABLE
A.	Glaucoma	Not defined	1	NO	
B.	Entropion	Not defined	2-4	Breeder option	
C.	Ectropion	Not defined	2	Breeder option	
D.	Distichiasis	Not defined	2	Breeder option	
E.	Corneal dystrophy - epithelial/stromal	Not defined	2, 5	Breeder option	
F.	Uveal cysts	Not defined	6	Breeder option	
G.	Persistent pupillary membranes				
	- iris to iris	Not defined	2, 6	Breeder option	
	- iris to cornea	Not defined	7	NO	
	- iris sheets	Not defined	6	NO	
	- lens pigment foci no strands	Not defined	8	Passes with no notation	
H.	Cataract				
		Presumed dominant with incomplete penetrance	2-4, 9-11	NO	
		Autosomal recessive	12	NO	
		Not defined	32	NO	
I.	Persistent hyperplastic primary vitreous/ persistent hyperplastic tunica vasculosa lentis (PHPV/PHTVL)	Not defined	2	NO	
J.	Persistent hyaloid artery	Not defined	2	Breeder option	

	DISORDER	INHERITANCE	REFERENCE	BREEDING ADVICE	GENETIC TESTS AVAILABLE
K.	Vitreous degeneration	Not defined	13, 14	Breeder option	
L.	Retinal atrophy - generalized (*prcd*)	Autosomal recessive	2, 15-19	NO	Mutation of the *prcd* gene
M.	Achromatopsia Type 1 (ACHM – Type 1)	Autosomal recessive	20	NO	Deletion in the *CNGA3* gene
N.	Retinal dysplasia - folds	Presumed autosomal recessive	2, 21-29	NO (Breeder option with Normal DNA test)	Mutation of the *COL9A3* gene
O.	Retinal dysplasia - geographic/ detached (without skeletal defects)	Presumed autosomal recessive	2, 21-29	NO	
P.	Retinal dysplasia - folds/geographic/ detached (with skeletal defects)	Autosomal recessive with incomplete dominance for the eyes	2, 21-30	NO	Mutation of the *COL9A3* gene
Q.	Limbal melanoma	Not defined	31	NO	

Description and Comments

A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure which, when sustained even for a brief period of time, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening examination.

B Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. Selection should be directed against entropion and toward a head conformation that reduces or eliminates the likelihood of the defect.

C. Ectropion

A conformational defect resulting in eversion of the eyelid(s), which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

D. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

E. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral. In Labrador Retrievers in Europe, one form of corneal dystrophy has been shown to be caused by accumulations of glycosaminoglycans in the corneal stroma. This form of corneal dystrophy is caused by a mutation in the CHST6 gene.

F. Uveal cysts

Fluid filled sacs arising from the posterior surface of the iris, to which they may remain attached or break free and float into the anterior chamber. Usually occur in mature dogs.

This disorder may be observed in any breed but retriever breeds tend to be predisposed. There is usually no effect on vision unless the cysts are heavily clustered and impinge on the pupillary area. Less frequently, the cysts may rupture and adhere to the cornea or anterior lens capsule. Multiple cysts may occlude the iridocorneal angle and cause glaucoma.

G. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

In the Labrador Retriever, this is a potentially serious problem as many of the PPM’s identified on routine screening examinations bridge from the iris to the cornea and/or from iris sheets bridging the pupils. These forms may cause vision impairment.

H. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts

to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

The most frequently reported cataracts in the Labradoodle (Australian) are bilateral or unilateral, focal, posterior polar (posterior cortical)/subcapsular cataracts, which usually present between 1-3 years of age. These are generally stationary or very slowly progressive and generally do not interfere with vision. It has been suggested that these cataracts are inherited as dominant with incomplete penetrance, but definitive breeding studies are still required to verify this hypothesis.

A second type of cataract is a progressive cortical cataract which may involve the entire lens. It is not clear whether this is a distinct entity, or an aberrant form of the posterior polar cataract.

I. Persistent hyperplastic primary vitreous (PHPV)/persistent hyperplastic tunica vasculosa lentis (PHTVL)

A congenital defect resulting from abnormalities in the development and regression of the hyaloid artery (the primary vitreous) and the interaction of this blood vessel with the posterior lens capsule/cortex during embryogenesis. This condition is often associated with persistent hyperplastic tunica vasculosa lentis (PHTVL) which results from failure of regression of the embryologic vascular network which surrounds the developing lens.

The majority of affected dogs have a retrolental fibrovascular plaque and variable lenticular defects which include posterior lenticonus/globus, colobomata, intralenticular hemorrhage and/or secondary cataracts. Vision impairment may result.

J. Persistent hyaloid artery (PHA)

A congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

K. Vitreous degeneration

Liquefaction of the vitreous gel, which may predispose to retinal detachment.

L. Retinal atrophy - generalized (*prcd*)

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Labradoodle is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day

blindness. A DNA test is available.

M. Achromatopsia Type 2 (ACHM – Type 2)

A congenital form of day blindness. Visual deficits become apparent between 8-10 weeks of age. Normal vision is present in low light conditions. Clinical examination is normal. Cone responses are absent on an electroretinogram. The causative genetic mutation of the *CNGA3* gene (3nt deletion in exon 7). A DNA test is available.

N. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness.

In the Labrador Retriever, the presence of retinal folds may be seen in the heterozygous state described in “R” below, thus the recommendation against breeding.

The breeding advice for Labrador Retrievers and Samoyeds diagnosed with "retinal dysplasia - folds" will be changed from "No" to "Breeder option" if the owner of the dog provides the registering office with results of the DNA test for the affected dog, showing that it is not a carrier of the *COL9A3* mutation.

O. Retinal dysplasia - geographic, detached without skeletal defects

Abnormal development of the retina present at birth; however, in the Golden Retriever, Labrador Retriever, and German Shepherd Dog it has been demonstrated that the geographic form of retinal dysplasia may not be apparent before dogs are 10 weeks of age.

Retinal dysplasia - geographic: Any irregularly shaped area of abnormal retinal development containing both areas of thinning and areas of elevation representing folds and retinal disorganization.

Retinal dysplasia - detached: Severe retinal disorganization associated with separation (detachment) of the retina.

These two forms are associated with vision impairment or blindness. Retinal dysplasia is known to be inherited in many breeds. The genetic relationship between the three forms of retinal dysplasia is not known for all breeds

In Europe, this condition has been documented as an autosomal recessive condition and results in early retinal detachment and blindness. Lens and corneal opacities can also be present, but skeletal abnormalities (see below) are not present. The condition of generalized retinal dysplasia with retinal detachment but without skeletal abnormalities has been reported primarily in Europe, and is rarely if ever seen in the United States.

In the United States, the milder forms of retinal dysplasia (folds/geographic) are seen in Labradors. These may represent the heterozygous form of the condition in which the homozygote also displays skeletal malformations (see “R” below) or it may represent a

genetically distinct entity with an undetermined mode of inheritance. It is not possible clinically to make this distinction. Thus, Labradors with any form of retinal dysplasia should not be used for breeding.

P. Retinal dysplasia - folds or detachment with skeletal defects

This condition is also known as oculo-skeletal dysplasia (OSD) or dwarfism with retinal dysplasia type 1 (DRD1) in the Labrador Retriever. A similar condition, DRD2, occurs in the Samoyed. The condition is autosomal recessive and homozygous affected dogs have shortened forelimbs ("downhill" conformation) with valgus deformity. They have severe ocular defects including cataract, retinal folds/multifocal retinal dysplasia, vitreal degeneration and retinal detachment. The ocular abnormalities result in blindness in most dogs. Heterozygous dogs can have either a normal ocular exam or have multiple retinal folds, vitreal membranes, or vitreal degeneration suggesting a semi-dominant mechanism with respect to the eyes. It is important to note that generally the retinal folds present in heterozygous dogs tend to cluster around the major superior blood vessels of the central tapetal region. The condition is caused by a 1 base pair insertion of *COL9A3.* A DNA test is available.

Q. Limbal melanoma

Most limbal melanomas are really epibulbar melanocytomas, but there is a possibility of an extension of an intraocular melanoma extending outward and presenting as a limbal melanoma. An epibulbar melanocytoma originates from the superficial pigment lining the limbus and the lesion may eventually extend into the eye. Metastasis has not been documented and the mass is characterized by large epithelioid cells. The lesion presents as a subconjunctival smooth mass most commonly in the dorsolateral limbal region and extends later into the cornea and posterior on the sclera. Breed predisposition has been noted in the German Shepherd, Labrador and Golden Retriever.

Historical Note:

Central progressive retinal atrophy was previously a condition listed for this breed. However as the condition is no longer identified in the breed, the condition has been removed. Central progressive retinal atrophy was a progressive retinal degeneration in which photoreceptor death occurred secondary to disease of the underlying pigment epithelium. Progression was slow and some animals never lost vision. CPRA occurred in England, but was uncommon elsewhere.

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