12th Edition

The Blue Book

OCULAR DISORDERS
PRESUMED TO BE INHERITED
IN PUREBRED DOGS

GENETICS COMMITTEE OF THE AMERICAN COLLEGE OF VETERINARY OPHTHALMOLOGISTS

2019



PORTUGUESE PODENGO PEQUENO

	DISORDER	INHERITANCE	REFERENCE	BREEDING ADVICE	GENETIC TESTS AVAILABLE
A.	Distichiasis	Not defined	1	Breeder option	
B.	Persistent pupillary membranes - iris to iris	Not defined	2	Breeder option	
C.	Cataract	Not defined	3	NO	
D.	Vitreous degeneration	Not defined	1	Breeder option	
E.	Retinal atrophy - rod-cone dysplasia type 3 (<i>rcd3</i>)	Autosomal recessive	4	NO	Mutation in the <i>PDE6A</i> gene
F.	Retinal atrophy (prcd)	Autosomal recessive	5	NO	Mutation in the prcd gene

Description and Comments

A. 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.

B. 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.

C. Cataracts

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.

D. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

E. Retinal atrophy - rod-cone dysplasia type 3 (*rcd3*)

PRA in the Protuguese Podengo Pequeno is an autosomal recessive trait caused by a one base pair deletion in the gene encoding the alpha subunit of cyclic GMP phosphodiesterase (rcd3). PRA begins early in life with clinical signs of night blindness and a lack of rod ERG responses is seen at 6-8 weeks of age. Dogs are completely blind by 2-3 years of age when ophthalmoscopic signs are first visible. The mutation is found in the *PDE6A* gene. A DNA test is available.

F. Retinal atrophy - 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.

Other forms of retinal degeneration that are not *prcd* are recognized in the Portuguese Podengo Pequeno. The currently available genetic test will not detect these other forms of PRA.

References

- 1. ACVO Genetics Committee, 2015 and Data from OFA All-Breeds Report, 2014-2015.
- 2. ACVO Genetics Committee, 2013-2014 and Data from OFA All-Breeds Report, 2013-2014.
- 3. ACVO Genetics Committee, 2016 and/or Data from CERF/OFA All-Breeds Report, 2010-2015.
- 4. ACVO Genetics Committee, 2016 and/or Data from OFA All-Breeds Report, 2016.
- 5. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006;88:551-563.

OCULAR DISORDERS REPORT PORTUGUESE PODENGO PEQUENO

	TOTAL DOGS EXAMINED		-2014 64	2015-2019 263	
Diagnostic Name			%	#	%
EYELIDS					
25.110	distichiasis	3	4.7%	11	4.2%
CORNEA					
70.700	corneal dystrophy	0		3	1.1%
UVEA					
93.710	persistent pupillary membranes, iris to iris	3	4.7%	11	4.2%
93.730	persistent pupillary membranes, iris to cornea	0		1	0.4%
93.750	persistent pupillary membranes, lens pigment foci/no strands	0		1	0.4%
LENS					
100.210	cataract. suspect not inherited/significance unknown	1	1.6%	7	2.7%
100.301	punctate cataract, anterior cortex	0		1	0.4%
100.302	punctate cataract, posterior cortex	0		1	0.4%
100.303	punctate cataract, equatorial cortex	0		2	0.8%
100.306	punctate cataract, nucleus	0		1	0.4%
100.311	incipient cataract, anterior cortex	0		5	1.9%
100.312	incipient cataract, posterior cortex	1	1.6%	1	0.4%
100.313	incipient cataract, equatorial cortex	0		1	0.4%
100.315	incipient cataract, posterior sutures	0		2	0.8%
100.316	incipient cataract, nucleus	0		_	0.8%
100.317	incipient cataract, capsular	1	1.6%	0	0.070
100.325	incomplete cataract, posterior sutures	0		1	0.4%
100.328	posterior suture tip opacities	0		1	0.4%
100.330	generalized/complete cataract	0		1	0.4%
100.340	resorbing/hypermature cataract	0		;	0.4%
100.375	subluxation/luxation, unspecified	0		3	1.1%
100.979	significant cataracts (summary)	2	3.1%	20	7.6%
VITREOL 110.120	persistent hyaloid artery/remnant	0		2	0.8%
110.120	vitritis	1	1.6%	5	1.9%
110.200		2	3.1%	9	3.4%
RETINA					
120.310	generalized progressive retinal atrophy (PRA)	1	1.6%	4	1.5%
120.960	retinopathy	2	3.1%	1	0.4%
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OTHER 900.110	other. suspect not inherited/significance unknown	1	1.6%	11	4.2%
NORMAL	-				
0.000	normal globe	52	81.2%	197	74.9%