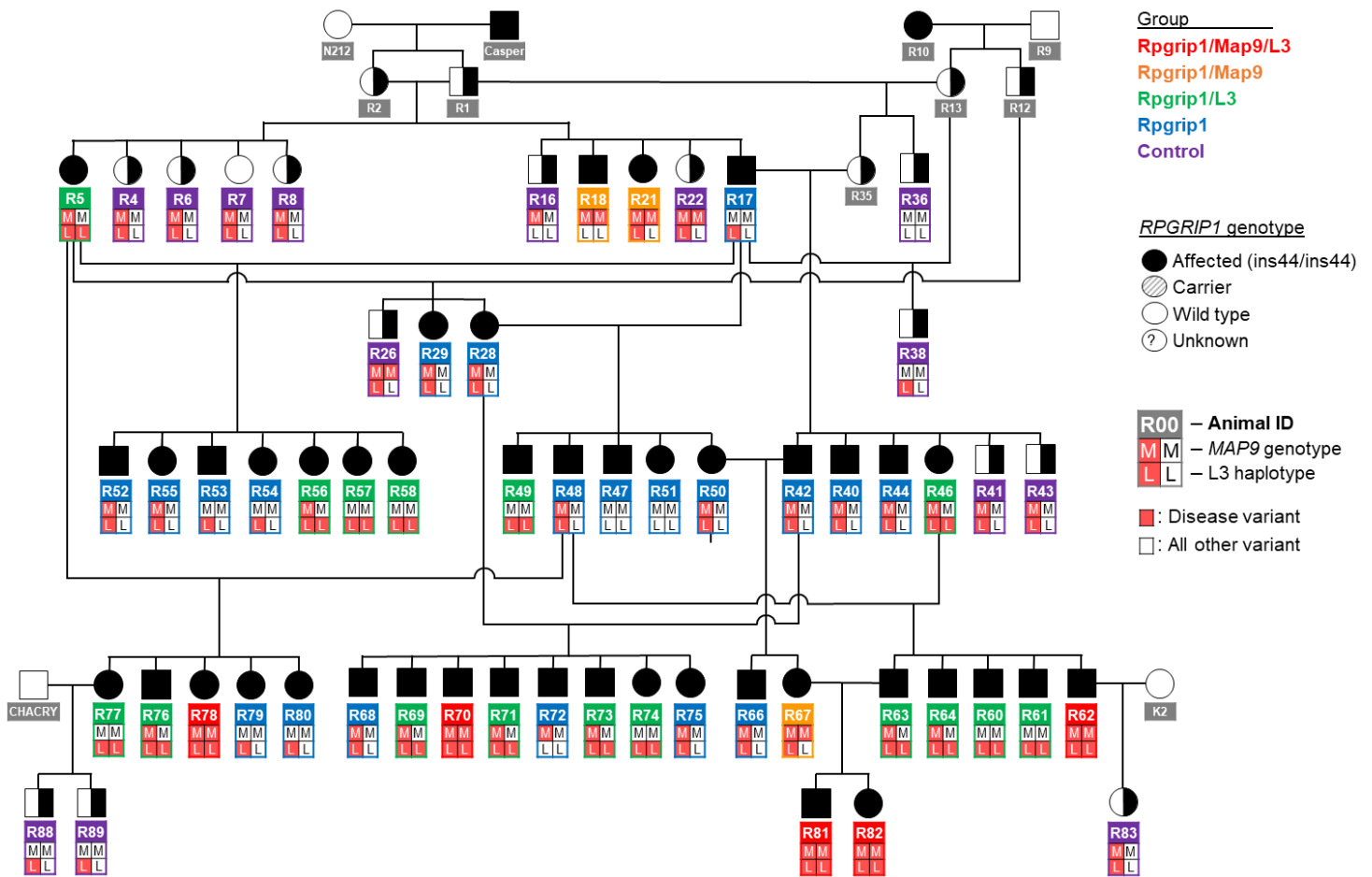
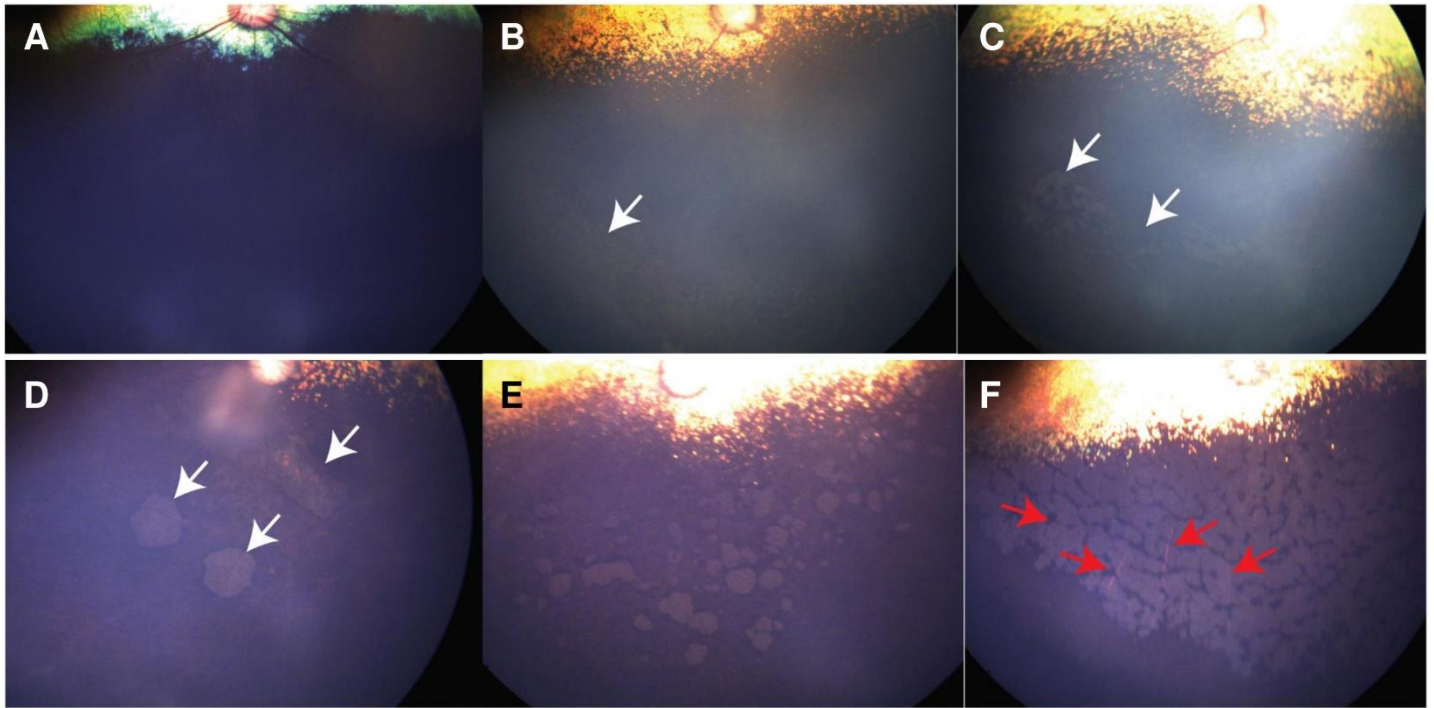


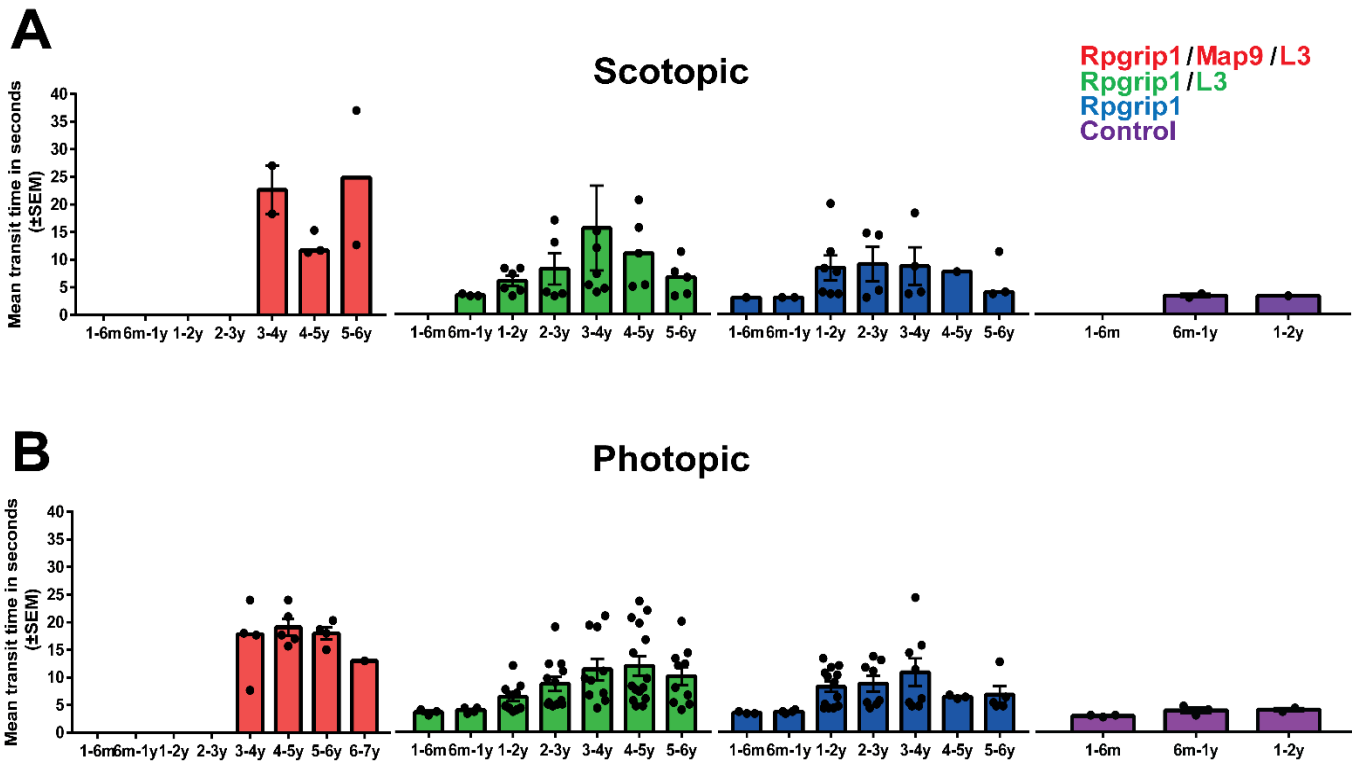
# Supplementary Information



**Supplementary Figure 1. Segregation of the genotype combinations and their phenotypic variability in the canine pedigree studied.** The IDs of the animals enrolled in the current study are color-coded. Of the four original founders, three (Casper, R9, R10) were miniature longhaired dachshunds and one (N212) was a mix breed. The genotypes of the three loci, *RPGRIP1*, *MAP9*, and *L3*, associated with *cord1* are indicated. Circle, female; square, male.



**Supplementary Figure 2. Illustration of the progression of retinal atrophy in the non-tapetal fundus in *RPGRIP1*<sup>ins/ins</sup> dogs.** (A) Normal non-tapetal canine fundus. (B-F) Non-tapetal canine fundus of *RPGRIP1* mutants representing early (B, C), mid (D, E), to advanced (F) disease stage. Initial changes include small areas of depigmentation (white arrows) corresponding to focal retinal atrophy. With progression, these lesions become more numerous and coalesce to encompass a broader area of the fundus (red arrows).



**Supplementary Figure 3. Longitudinal evaluation of scotopic and photopic vision-guided navigation in *RPGRIP1* canine mutants using an obstacle avoidance course.** Vision testing was repeated over time in an obstacle avoidance course under scotopic (A) and photopic (B) conditions in selected dogs from the Rpgrip1/Map9/L3, Rpgrip1/L3 and Rpgrip1 genotype groups as well as the control group. The time spent to complete the obstacle course is displayed (transit time) per age and group. The dashed lines represent the 95% confidence interval obtained from three normal animals unrelated to this study group. Error bars represent SD.

**Supplementary Table 1. Missense variants within L3 found in genes expressed in the retina**

CFA30 (bp)	Gene	Alt (1)	Ref (0)	Alt allele freq. in DBMD	Alt allele freq. in Dog10K	Cases		Controls		cDNA change	Amino acid change	Impact (SnPEff)	Polyphen HumDiv	Polyphen HumVar	PROVEAN	Mut Pred2	SIFT	FATHMM	Vest Score
						R46	R76	R42	R50										
9,162,384	PLA2G4F	G	A	NA*	0.0647	1/1	1/1	0/1	0/1	c.377C>T	p.Ser126Phe	Moderate	Benign (0.009)	Benign (0.018)	Neutral (0.208)	0.251	Tolerated	Tolerated	0.642
9,256,479	GANC	A	G	0.4348	NA†	1/1	1/1	0/1	0/1	c.79G>A	p.Val27Ile	Moderate	Benign (0.000)	Benign (0.001)	Neutral (0.229)	0.191	Tolerated	Tolerated	0.320
<b>9,411,186</b>	<b>ZNF106</b>	<b>G</b>	<b>A</b>	<b>0.0363</b>	0.0337	1/1	1/1	0/1	0/1	c.3626A>G	p.His1209Arg	Moderate	Benign (0.225)	Benign (0.057)	Neutral (-0.898)	0.083	<b>Affects function</b>	Tolerated	0.368
<b>9,626,620</b>	<b>STARD9</b>	<b>T</b>	<b>G</b>	<b>0.0357</b>	0.0354	1/1	1/1	0/1	0/1	c.5821G>T	p.Ala1941Ser	Moderate	Possibly damaging (0.944)	Possibly damaging (0.685)	Neutral (-0.502)	0.047	<b>Affects function</b>	Tolerated	0.103
<b>9,634,432</b>	<b>STARD9</b>	<b>A</b>	<b>G</b>	<b>0.0354</b>	0.0354	1/1	1/1	0/1	0/1	c.13019G>A	p.Arg4340His	Moderate	<b>Probably damaging (1.000)</b>	<b>Probably damaging (1.000)</b>	Neutral (-1.347)	0.444	<b>Affects function</b>	Tolerated	0.713
<b>9,940,463</b>	<b>UBR1</b>	<b>C</b>	<b>A</b>	<b>0.1398</b>	NA†	1/1	1/1	0/1	0/1	c.1694A>C	p.Lys565Thr	Moderate	Possibly damaging (0.810)	Possibly damaging (0.474)	<b>Deleterious (-3.608)</b>	0.339	Tolerated	Tolerated	0.540
10,414,503	PPIP5K1	A	G	0.1281	0.0909	1/1	1/1	0/1	0/1	c.2336G>A	p.Ser779Asn	Moderate	Benign (0.004)	Benign (0.020)	Neutral (-0.705)	0.277	Tolerated	Tolerated	0.285
10,426,046	PPIP5K1	A	G	0.2140	NA†	1/1	1/1	0/1	0/1	c.86G>A	p.Gly29Glu	Moderate	Benign (0.061)	Benign (0.025)	Neutral (-0.752)	0.190	Tolerated	Tolerated	0.077
<b>10,538,649</b>	<b>SERINC4</b>	<b>A</b>	<b>G</b>	<b>0.0639</b>	0.0409	1/1	1/1	0/1	0/1	c.1300G>A	p.Ala434Thr	Moderate	<b>Probably damaging (1.000)</b>	<b>Probably damaging (0.998)</b>	<b>Deleterious (-3.075)</b>	<b>0.877</b>	<b>Affects function</b>	Tolerated	0.302
10,561,536	WDR76	C	A	0.1899	NA†	1/1	1/1	0/1	0/1	c.22A>C	p.Ser8Arg	Moderate	Benign (0.164)	Benign (0.041)	Neutral (-0.314)	0.110	Tolerated	<b>Damaging</b>	0.231
10,603,246	WDR76	T	C	0.1394	NA†	1/1	1/1	0/1	0/1	c.1469C>T	p.Pro490Leu	Moderate	Possibly damaging (0.886)	Possibly damaging (0.764)	Neutral (-1.952)	0.200	<b>Affects function</b>	Tolerated	0.520
11,422,211	C15orf43	T	C	0.4925	NA†	1/1	1/1	0/1	0/1	c.449C>T	p.Ser150Phe	Moderate	Possibly damaging (0.852)	Possibly damaging (0.431)	<b>Deleterious (-2.788)</b>	0.183	Tolerated	Tolerated	0.189

CFA, canis familiaris chromosome; Alt, alternative nucleotide; Ref, reference nucleotide (CanFam3.1). The 9,162,384 position variant has been remapped from canFam4 with the NCBI remapping service.

NA\*, Non-detection of the variant under the CanFam3.1 assembly. NA†, Failure to pass the 0.1 MAF threshold.

**Supplementary Table 2. Fundus score based on ophthalmic changes detected on retinal photographs (refer to Fig. 2).**

Score	Stage of degeneration	Dark discoloration		Hyper-reflectivity
		Severity	Location	
<b>1</b>	<b>Incipient</b>	Incipient	Periphery	Absent
<b>2</b>	<b>Early</b>	Mild	Superior tapetum (1/2)	Absent
<b>3</b>	<b>Mid</b>	Moderate	Superior tapetum (1/2-2/3)	Mild
<b>4</b>	<b>Advanced</b>	Severe	Superior tapetum (>2/3)	Moderate
<b>5</b>	<b>End-stage</b>	Terminal	Generalized	Severe