

Supplementary Table 1 *RPGRIP1*^{-/-} MLHD samples used in SNP array genotyping

Sample ID	Age	Sex	RD phenotype	Sample source	Notes
AHT_D	<2 (2)	M	EORD	Research colony	-
AHT_E	<2 (2)	F	EORD	Research colony	Excluded from GWAS analysis
MLD002	2.7	F	EORD	Family K	Dam:MLD001
MLD021	<<5 (2)	M	EORD	Family K	Excluded from GWAS analysis, Dam:MLD001
MLD027	1	F	EORD	Japanese pet	WGA_PCR
MLD028	1.5	F	EORD	Japanese pet	-
MLD029	1.3	M	EORD	Japanese pet	-
MLD030	1.8	F	EORD	Japanese pet	-
MLD031	1.8	M	EORD	Japanese pet	-
MLD032	3	F	EORD	Japanese pet	-
MLD033	3	F	EORD	Japanese pet	-
MLD034	<2 (2)	M	EORD	Japanese pet	-
MLD035	1.8	F	EORD	Japanese pet	-
MLD037	1	M	EORD	Japanese pet	-
MLD064	<3.4 (3.4)	M	EORD	Japanese pet	-
MLD066	2	F	EORD	Japanese pet	-
MLD070	3	F	EORD	Japanese pet	WGA_PCR
MLD078	1.5	F	EORD	Japanese pet	-
MLD079	0.7	M	EORD	Japanese pet	-
MLD081	1	M	EORD	Japanese pet	-
MLD082	2	F	EORD	Japanese pet	-
MLD083	2	M	EORD	Japanese pet	WGA_F29
MLD084	<2.5 (2.5)	F	EORD	Japanese pet	-
MLD085	1.4	F	EORD	Japanese pet	-
MLD088	2.5	F	EORD	Japanese pet	WGA_F29
MLD105	0.3	F	EORD	Japanese pet	-
MLD113	<3.3 (3.3)	M	EORD	Japanese pet	-
MLD117	1	F	EORD	Japanese pet	-
MLD124	<<4.5 (2)	F	EORD	Japanese pet	-
MLD142	2.5-3 (3)	M	EORD	Japanese pet	-
MLD222	2.8	F	EORD	Japanese pet	-
MLD234	2.9	M	EORD	Japanese pet	-
MLD455	3	F	EORD	Japanese pet	Dam:MLD453
MLD004	2.4-5.2 (5.2)	F	MORD	Family K	Excluded from analysis, Dam:MLD002
MLD023	5	M	MORD	Japanese pet	-
MLD025	4	M	MORD	Japanese pet	-
MLD112	3.5-4.5 (4.5)	M	MORD	Japanese pet	-
MLD001	9.2-10.6 (10.6)	F	LORD	Family K	Excluded from GWAS analysis
MLD003	7.8-9.3 (9.3)	F	LORD	Family K	Excluded from GWAS analysis, Dam:MLD002
MLD005	>10.8 (10.8)	M	LORD	Family K	Excluded from GWAS analysis, Dam:MLD001
MLD007	9.4	M	LORD	Family K	Excluded from GWAS analysis, Dam:MLD001
MLD009	5.2-6.6 (6.6)	M	LORD	Family K	Excluded from GWAS analysis, Dam:MLD002
MLD022	7	M	LORD	Japanese pet	-
MLD024	9	M	LORD	Japanese pet	-
MLD039	7	F	LORD	Japanese pet	-
MLD061	8	F	LORD	Japanese pet	-
MLD103	15	M	LORD	Japanese pet	-
MLD114	8.5	M	LORD	Japanese pet	-
MLD115	9	F	LORD	Japanese pet	-
MLD151	6	M	LORD	Japanese pet	-
MLD291	5.6	M	LORD	Japanese pet	-
MLD435	10	M	LORD	Japanese pet	-

MLD436	9	F	LORD	Japanese pet	-
MLD438	9.8	M	LORD	Japanese pet	-
MLD443	7.8	F	LORD	Japanese pet	-
MLD445	~16 (16)	M	LORD	Japanese pet	-
MLD446	15	F	LORD	Japanese pet	-
MLD006	8.9	M	Normal	Family K	Excluded from GWAS analysis, Dam:MLD001
MLD008	5.6	M	Normal	Family K	Excluded from GWAS analysis, Dam:MLD004
MLD010	6.1	F	Normal	Family K	Excluded from GWAS analysis, Dam:MLD004
MLD011	5.2	M	Normal	Family K	Excluded from GWAS analysis, Dam:MLD001
MLD012	5.7	F	Normal	Family K	Excluded from GWAS analysis, Dam:MLD003
MLD087	4.8	F	Normal	Japanese pet	-
MLD102	5	F	Normal	Japanese pet	-
MLD104	6.9	M	Normal	Japanese pet	-
MLD106	10.4	M	Normal	Japanese pet	-
MLD127	8.6	M	Normal	Japanese pet	-
MLD130	5.3	F	Normal	Japanese pet	-
MLD132	8.5	M	Normal	Japanese pet	-
MLD134	10	F	Normal	Japanese pet	-
MLD153	10.4	M	Normal	Japanese pet	-
MLD201	9.2	F	Normal	Japanese pet	WGA_PCR
MLD214	9.8	F	Normal	Japanese pet	-
MLD219	7	M	Normal	Japanese pet	WGA_PCR
MLD235	9.9	F	Normal	Japanese pet	-
MLD242	12.4	F	Normal	Japanese pet	-
MLD265	5.4	F	Normal	Japanese pet	-
MLD269	4.9	F	Normal	Japanese pet	-
MLD288	9.4	M	Normal	Japanese pet	-
MLD343	7.2	M	Normal	Japanese pet	-
MLD345	6	M	Normal	Japanese pet	-
MLD354	5.3	F	Normal	Japanese pet	-
MLD366	14	F	Normal	Japanese pet	-
MLD369	8	F	Normal	Japanese pet	-
MLD370	10.4	F	Normal	Japanese pet	-
MLD405	10.7	F	Normal	Japanese pet	-
MLD413	6	M	Normal	Japanese pet	-
MLD415	5.5	F	Normal	Japanese pet	-
MLD424	8.8	M	Normal	Japanese pet	-
MLD453	9.4	F	Normal	Japanese pet	-
AHT11044	6.3	M	Normal	UK pet	-
AHT11056	7.7	F	Normal	UK pet	-
AHT11057	8.8	M	Normal	UK pet	-
AHT11058	6.4	F	Normal	UK pet	-
AHT11062	7.7	F	Normal	UK pet	-
AHT11063	5.4	F	Normal	UK pet	-

The ages denote the onset of RD in affected dogs and the timing of last examination in Normal dogs. Where a definite age was unavailable, the value shown in brackets was used in the PLINK analysis. Some samples were prepared by whole genome amplification either by PCR-based (WGA_PCR, GenomePlex®; Sigma-Aldrich) or Phi29 DNA polymerase-based (WGA_F29, illustra GenomiPhi V2; GE Healthcare) methods.

Supplementary Table 2 Primer sequences for canine *LRAT*

Gene	Featrure	Forward primer sequence (5'>3')	Reverse primer sequence (5'>3')	Nt from <i>LRAT</i>	
				Start	End
<i>LRAT</i>	Exon 1-2	TGCAGGTGAGGTCCCCG	CTGCTCTTTAGTCGTTTCGAC	-63	823
	Exon 3	GGTACGTGTGATCCTTCTCAG	CATGTTTTCATCTGAACGAGGG	4706	5389
	Upstream	ACAGGACAAACGAAACGTC	GGAGTCGCTGGTGTTAATC	-2759	-2037
		CCCCAAAGTTTACAACCGCTCA	GCCCCGATTCGGAGCTAATA	-2091	-1643
		TCTGCTTTTCGATCCGTTTG	CCTGCAAAGGCGGATTAACG	-1960	-1367
		ACCCCGAAGCTAGGGAAAT	GGCGAAACGCACCTGGG	-1412	-721
		CCCAGGTGCGTTTCGCC	CAGCTTCTCCAGCAACAGG	-737	47
	Intron	CGGAGTGTGGCGATTGCAG	TGTAAGGTGACGGATGTTAGC	716	1630
		GCCAAGTCTCTGTTTTATTGAGA	CCAAACTGTTACATCATTCAAAGT	1493	2437
		TGTTGCTGCCTAACACCTCTTGTG	ATCGAGTTTTTCAGGACAATGAGAA	2371	3298
		GCCAAGCACCTTTTCTCATTGTCC	ATGATCCATACCCCTGAGGAAG	3263	4177
		AGAGGGTAGTCTGGACAAGTT	TGCTGAAGCAAGAACAATTCTCTG	4014	4825
	Human 3'-UTR equivalent	CTAAACTAAACGATTTTCCCCTCGT	AAGTCTAGCTTGTGGCCTAGTTTTT	5350	6198
		AGTGAGCGTTCATGTACTTTTGC	GAGATCGTGAAATTACAACAGGTCA	6134	6866
		TGCTAGACACTGACCTGTTGTAAT	TCCACTGCCGTTTCATAAAG	6832	7723
		GCAATGATCAAGTGATGCAAG	GGCTCCCAGATGTAGATGACA	7658	8537

Supplementary Table 3 Primer sequences for the coding regions of canine *MAP9*, *GUCY1A3*, and *GUCY1B3*

Gene	Exon	Forward primer sequence (5'>3')	Reverse primer sequence (5'>3')
<i>MAP9</i>	1	CAGACCCGGTCAGAGTTTGAAT	CGTAGTAAAAACCTAGTACTGAAGC
<i>MAP9</i>	2	TTCTCCTGCCCTAAATACACG	TGACAGGCATCCTTTTACTGC
<i>MAP9</i>	3	TGCCTAAGTTAGCTGTGTTTATAGAGC	TGCAAGACTGACATGCAACTGG
<i>MAP9</i>	4	GCATCCTGGTAGTCGTTGAAG	ACAGTGCCGTGATCATTCAATA
<i>MAP9</i>	5	TTGAGATGCCGGTGAACGTACT	GCTTGGGGTAGGTTGCCCTTAC
<i>MAP9</i>	6	AGATGAATGTAATTGCTCTCCTTG	GGAGTTTTCCACAGACAGTTCAGA
<i>MAP9</i>	7	TTGGCCCTAAGCCTAACTGTGA	GGTGTCCCGTGTCTTATCA
<i>MAP9</i>	8	CCTGCCAACTCCTGAAATAGAGG	CCTTGGTGAATGGGTAACCTTGT
<i>MAP9</i>	9	TTCTTAACTCATGCTCGTCTTTACG	CTAATGGTGCCGACGTGTTGAT
<i>MAP9</i>	10-12	TCGTATCTACCTGTCTCCGTCTGT	ACAGATTCCACAAGGCAGCTCT
<i>MAP9</i>	13	CCCCACATGCAGTCACTTACCA	CAGTCCCACAGAGAAAACACA
<i>MAP9</i>	14-15	TGTGCTAGATCACTTAGGGCAAC	TGACTGTTGTCTGTTGGGGAGTG
<i>MAP9</i>	16	GGAAATTATAACTTGGGTTATCTG	GTTGCTGTGAAAACACGATGG
<i>MAP9</i>	17	CTTCAGCAAGCAACCTGATTTG	GATTGTCACGTTAAGTCTGGGACA
<i>GUCY1A3</i>	1	TTTTCTGGACCAGTGCATAGCGG	CTCGCGTCCACGCTCCACAG
<i>GUCY1A3</i>	2	TGTGTAGCATTGTGCTCCTGCCA	GGCCAGTCAGGAAGACCTGGA
<i>GUCY1A3</i>	3	TTGTGGTAGGGACATGTGGAATGC	GCCTTGAAAGTGAGAAAGCACGC
<i>GUCY1A3</i>	4	GGCTGTCAGCGAGACCACCC	AGCAGCAGTACACGGCGACA
<i>GUCY1A3</i>	5	TTCTTCCCATCCCCCTAGTTGGC	ATGAGTCAGCTTCCCATGCGGC
<i>GUCY1A3</i>	6	TGTTACCAGACGTAAGGGAAC	TTCATTTCTAGTCCTTAACCAAGA
<i>GUCY1A3</i>	7	GCGTCAGCGTCCCCTTAGTGA	CCAGGATGCGGCTTGACAGAGA
<i>GUCY1A3</i>	8	GCCACTCCCTGATGTTGTGCT	ACACAGACGTTTGCCGACTGC
<i>GUCY1A3</i>	9	TGGTGACCGTGCGAGGTGTG	TCAGCGTGGGGGAAGACAGGA
<i>GUCY1A3</i>	10	TCTGAGCACATTAGGGCACGCT	TGTGTTGAATTTGAGTACAGGT
<i>GUCY1B3</i>	1	CCGTTAAACTGGGGGCGGG	CCGCGTGGACAAACACACACTT
<i>GUCY1B3</i>	2	TCAGGCATTGCATTGGGCATTAGA	CCTCCACCATCGTGCAGCGT
<i>GUCY1B3</i>	3	TCCACTTGCACAAAAGTTGGGGAT	GGATGTTCAAGGTGGGCAACCA
<i>GUCY1B3</i>	4	TCAACGCAACCACCACTCTCT	TGCTCGTGCAGCACTGTTTTGGT
<i>GUCY1B3</i>	5	AAGCCTACCCGGACAAGAGGGTA	AGCCAGCCGTCCTAGGGAAAGTT
<i>GUCY1B3</i>	6	TGCATGGAGCCTCTTTCTCCCT	TTCCACCGCCCTCAGGCAAATA
<i>GUCY1B3</i>	7	AGCCCTCGCAAAGGTCTTACCA	ACACTCGCAAAGTATTGAGACAGG
<i>GUCY1B3</i>	8	ACAGACCAGGTTGCACAGGACA	ACAGGCCCGTGCCTTTTTAGT
<i>GUCY1B3</i>	9	ACAGCTTCTACTGTGAGGCCA	AACAGTGACTGCGTGGAGGCTGTA
<i>GUCY1B3</i>	10	GGCAGTGGCTCTTGTGCCGTA	CTTTGCCCTCAAGGTGTCTTTCT
<i>GUCY1B3</i>	11	CAGGTTCAAGTAGATGGCGAATCTGT	TGAGAGTTAAACCACACGTAACGACAC
<i>GUCY1B3</i>	12	AGCAGAACGGAAACCACTGGAGA	TCACAGCCCTCTCGCCTCAGC
<i>GUCY1B3</i>	13	GTGGCCCTGTTTACCCAGAAT	AGAGCGCCTAACATTTATCCCAAGC

Supplementary Fig. 1 SNP genotypes of 83 *RPGRIP1*^{-/-} MLHDs at the CFA15 locus associated with EORD. EORD (red header), MORD (orange header) and LORD (yellow header) samples are shown in the order of the onset age (except individuals with atypical haplotypes), while Normal samples (green header) are shown in the order of the age at examination. SNP alleles in light blue boxes represent the predominant allele in EORD, while those in light yellow boxes represent the alternative allele. The 1.49-Mb region of homozygosity shared by 90% of the EORD cases is outlined in red. The Dog ID of the UK dogs is shown in blue.

Supplementary Fig. 2 Haplotype combinations of the locus associated with EORD. Haplotype blocks were inferred using the controls (M1: 15 LORD + 34 Normal). Haplotype 'a' corresponds to the major haplotype in the EORD cases. Alternative haplotypes (b-g) are indicated in different color blocks where alternative SNP alleles are shown in white letters. Left, four different haplotype pairs in the EORD cases (n=31). Right, haplotype pairs in the controls (n=49) are shown where both haplotype frequencies are ≥ 0.03 . Haplotype frequencies in the controls: a=0.22, b=0.22, c=0.09, d=0.07, e=0.05, f=0.03, g=0.03.

Cases (EORD)				Phenotype	Controls (LORD + Normal)														
28	1	1	1	No. of dogs	4	3	3	2	2	2	2	2	2	2	1	1	1	1	23
a a	b c	a -	- -	Haplotype ID	a b	a a	a d	b b	c c	b c	b d	a f	a e	d d	b g	b f	a g	- -	
A A	A A	A A	A A	55.66	A A	A A	A G	A A	A A	A A	A G	A G	A A	G G	A G	A G	A G	- -	
T T	T T	T T	T T	55.76	T T	T T	T T	T T	T T	T T	T T	T T	T T	T T	T C	T T	T C	- -	
G G	G A	G G	G G	55.86	G G	G G	G A	G G	A A	G A	G A	G A	G A	A A	G G	G A	G G	- -	
G G	G G	G G	G G	55.94	G G	G G	G G	G G	G G	G G	G G	G A	G G	G G	G A	G A	G A	- -	
A A	G G	A G	G G	56.11	A G	A A	A A	G G	G G	G G	G A	A A	A G	A A	G G	G A	A G	- -	
G G	A G	G A	G G	56.28	G A	G G	G G	A A	G G	A G	A G	G G	G G	G G	A A	A G	A G	- -	
G G	G G	G G	G G	56.29	G G	G G	G A	G G	G G	G G	G A	G A	G G	A A	G G	A G	G G	- -	
T T	T C	T T	T C	56.36	T T	T T	T T	T T	C C	T C	T T	T T	T T	T T	T T	T T	T T	- -	
C C	T C	C C	C C	56.38	C T	C C	C C	T T	C C	T C	T C	C T	C T	C C	T T	T T	T T	- -	
A A	G G	A A	A G	56.46	A G	A A	A A	G G	G G	G G	G A	A A	A A	A A	G G	T A	A G	- -	
T T	G G	T G	T G	56.46	T G	T T	T G	G G	G G	G G	G G	T T	T G	G G	G G	G T	T G	- -	
A A	A G	A A	A G	56.51	A A	A A	A A	A A	G G	A G	A A	A A	A G	A A	A G	A A	A G	- -	
C C	G C	C G	C C	56.62	C G	C C	C G	G G	C C	G C	G G	C G	C C	G G	G G	G G	C G	- -	

CFA15 (Mb)

Minor haplotypes

Supplementary Fig. 2