

Labrador Retriever 98 Gen Breed Formation - SNP chip: last_breed_formation_generation - 50 N_e bottleneck scenario

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0: Preparation

Defining the input and output directories

Loading libraries

```
## Loading required package: knitr

## Loading required package: ggplot2

## Warning: package 'ggplot2' was built under R version 4.3.2

## Loading required package: scatterplot3d

## Warning: package 'scatterplot3d' was built under R version 4.3.1

## Loading required package: RColorBrewer
```

Latex formatting function

Standard Error Confidence interval function

Gene mapping

2: ROH-Frequency

2.1: Autosome ROH-frequencies

2.1.1: Empirical - ROH frequency

2.2: ROH-hotspots - ROH Frequency and Length

5: Summary

5.1: General comparison

5.2: Selection Testing of Empirical ROH-Hotspots

5.2.1: Selection test (Sweep test)

```
## [1] "Selection test results"

## [1] "ROH-hotspot windows with an mean H_e Value lower or equal to the lower confidence interval of t

## [1] "5th percentile of the neutral model is: 0.16359"
```

Name	Under_selection	Window_based_Average_H_e
hotspot_chr28_window_1	Yes	0.118

Name	Under_selection	Window_based_Average_H_e
hotspot_chr1_window_1	Yes	0.144
hotspot_chr24_window_1	No	0.186
hotspot_chr13_window_1	No	0.190
hotspot_chr11_window_2	No	0.205
hotspot_chr15_window_1	No	0.223
hotspot_chr6_window_1	No	0.232
hotspot_chr11_window_1	No	0.233
hotspot_chr8_window_1	No	0.307

```
## [1] "C:/Users/jonat/GitHub/roh-island-simulation/results_chr1_3185_Ne_50TC/Pipeline_results/ROH_hotspots"
```

```
## [1] "ROH-hotspots under selection:"
```

Name	Under_selection	Window_based_Average_H_e
hotspot_chr28_window_1	Yes	0.118
hotspot_chr1_window_1	Yes	0.144

5.2.3: Extracting Hotspots under selection

OMIA Phenotypes

Breed-specific Phenotypes from OMIA

All Breed-specific non-defect phenotypes of the specified breed

Species-specific phenotypes overlapping with the detected empirical ROH hotspot(s)

```
## The following table(s) shows the empirical ROH hotspots overlapping with species-specific phenotypes
## , Hotspot_chr13_window_1, Hotspot_chr15_window_1
```

Non-defect Phenotypes overlapping with candidate regions for selection

Finding if there exists any non-defect phenotypes overlapping with ROH hotspot candidate regions for selection, for the specified breed (Defined from the list of VBO-IDs).

```
## No non-defect phenotypes discovered for the ROH hotspot candidate region(s) for selection
```

Table 3: Table of the non-defect breed-related phenotypes from OMIA

CHR	POS1	POS2	Phene	Phene_category	Considered_defect
20	52963476	52964967	Modifier of copper toxicosis, RETN-related	Homeostasis / metabolism phene	no
CHR	POS1	POS2	Phene	Phene_category	
1	24326030	24331495	Reduced hair shedding, MC5R-related	Integument (skin) phene	
1	111055483	111058288	Hyperlipidaemia/atherosclerosis, APOE-related	Homeostasis / metabolism phene	
3	92125265	92275341	Height, LCORL-associated body-size variation	Growth / size / body region phene	
5	63922271	63923224	Coat colour, extension	Pigmentation phene	
5	63922271	63923224	Coat colour, grizzle	Pigmentation phene	
5	63922271	63923224	Coat colour, melanistic mask	Pigmentation phene	
9	59449060	59512699	Coat colour, harlequin	Pigmentation phene	
10	7971606	8151219	Ears, folded (drop vs prick)	Hearing / vestibular / ear phene	
10	8459176	8604625	Height, HMGA2-associated body-size variation	Growth / size / body region phene	
13	8873544	9022401	Furnishings (moustache and eyebrows)	Integument (skin) phene	
15	30230533	30311189	Coat colour, phaeomelanin dilution, KITLG-related	Pigmentation phene	
16	57081626	57083181	Coat colour, dominant black	Pigmentation phene	
20	21883312	22101930	Coat colour, white spotting	Pigmentation phene	
20	56514447	56522856	Coat colour, phaeomelanin dilution, MFSD12-related	Pigmentation phene	
24	24041356	24084629	Coat colour, agouti	Pigmentation phene	
27	2535235	2543509	Curly coat	Integument (skin) phene	
30	38023809	38029593	Metabolizer of a cognitive enhancer	Homeostasis / metabolism phene	
32	4533042	4556071	Hair, long	Integument (skin) phene	

Table 4: All defect/non-defect phenotypes (for the studied species) overlapping with the empirical ROH hotspots

CHR	POS1	POS2	Phene	Phene_category	Considered_defect	Gene
13	37796064	37804968	Congenital keratoconjunctivitis sicca and ichthyosiform dermatosis	Vision / eye phene		
13	37924192	37980292	Epidermolysis bullosa, simplex, PLEC-related	Integument (skin) phene		
CHR	POS1	POS2	Phene	Phene_category	Considered_defect	Gene
15	18575495	18646528	Retinal atrophy - Cone-rod dystrophy 4	Vision / eye phene	yes	RPG