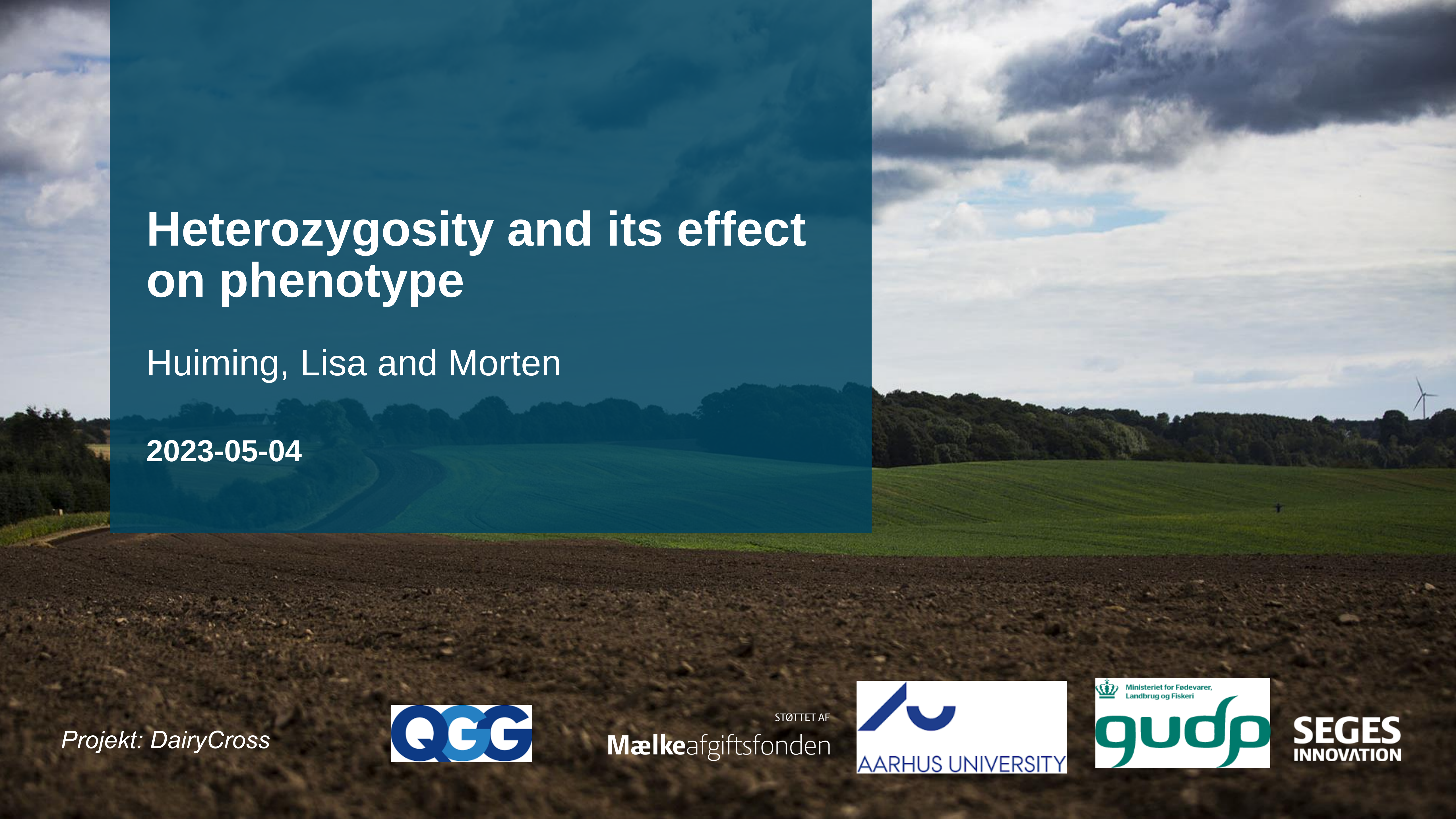

Heterozygosity and its effect on phenotype

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Projekt: DairyCross

QGG

STØTTET AF
Mælkeafgiftsfonden

AARHUS UNIVERSITY

Ministeriet for Fødevarer,
Landbrug og Fiskeri
gudp

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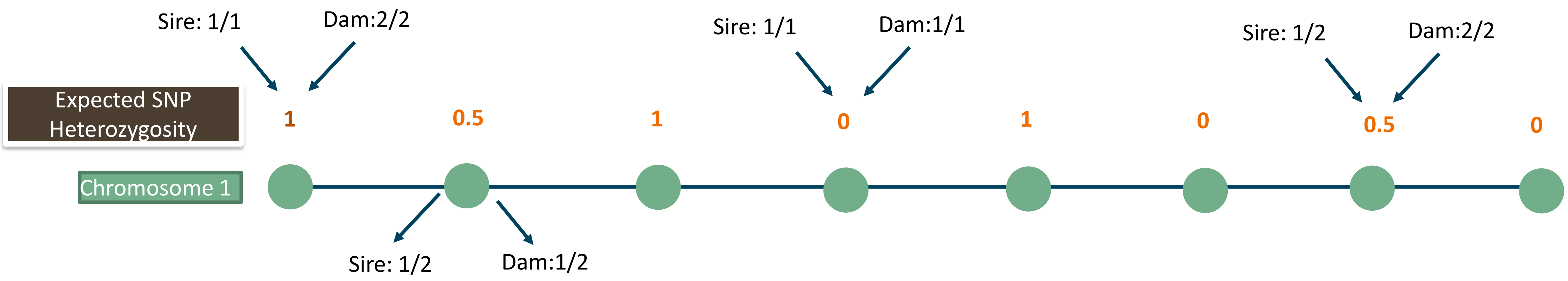
Overview

- Take into account the expected heterozygosity level in mating plan - pipeline
- The gain in heterozygosity by shifting the sire given a specific dam
- The effect of heterozygosity on phenotype

Calculation of the expected heterozygosity

Expected heterozygosity of genotyped parents



Sire: 1/1
Dam:2/2
Sire: 1/1
Dam:1/1
Sire: 1/2
Dam:2/2

Expected SNP Heterozygosity
1
0.5
1
0
1
0
0.5
0

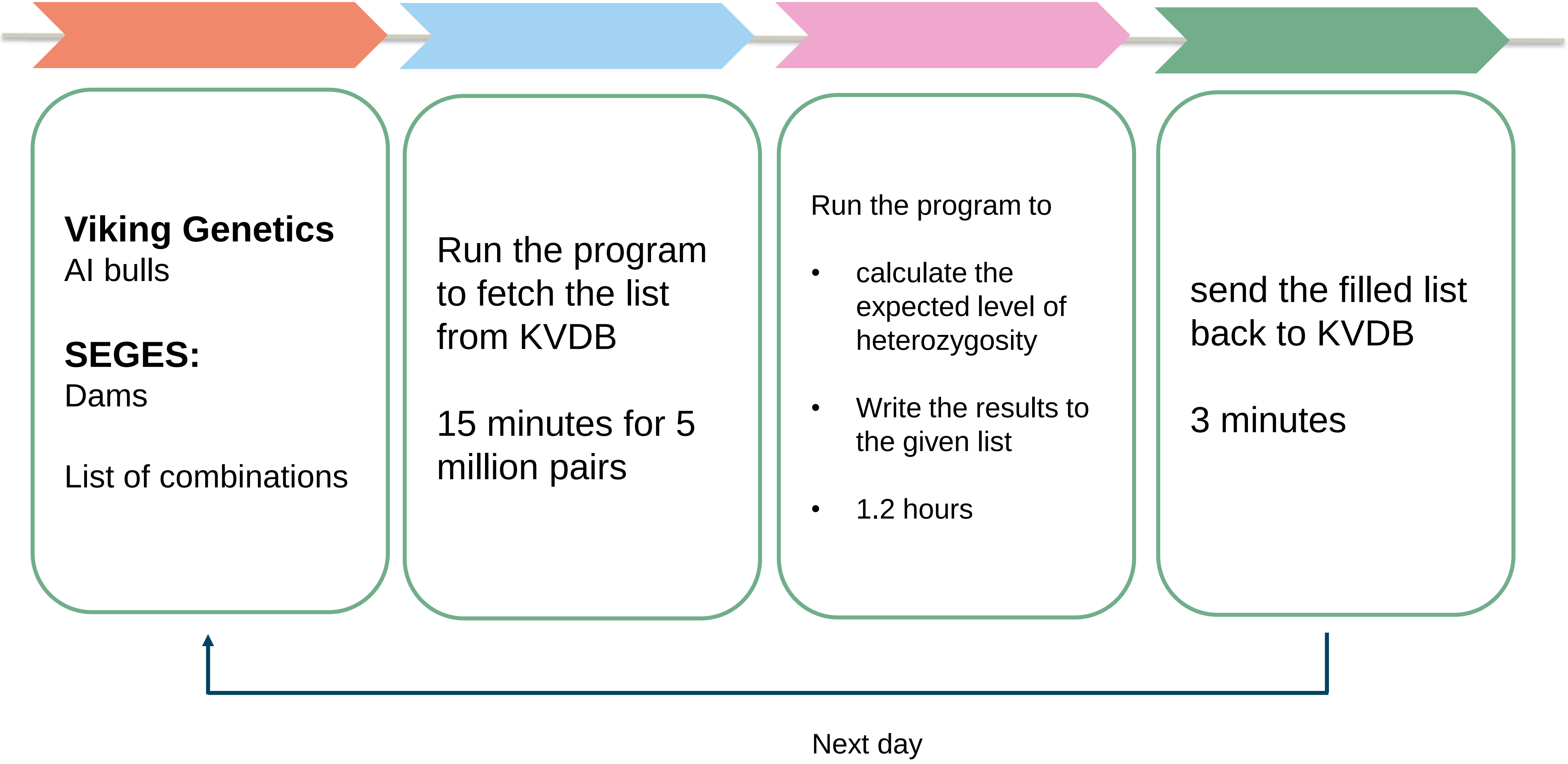
Chromosome 1

Sire: 1/2
Dam:1/2

Expected Het. Level = Nhet/Nsnps =**4/8=0.5**

- The genotypes are the imputed genotypes from the official test for HOL, JER and RDC.
- Map
  - The number of SNPs in the map is 46342 in HOL,41897 in JER,46914 in RDC
  - the maps of the three breeds were merged, and there are 41238 SNPs in common.
- If one of the parents or both parents are heterozygotes for the SNP, then the expected heterozygosity is 0.5.
- If both parents are the alternative homozygotes for the SNP, then the expected heterozygosity is 1.
- The expected level of heterozygosity for a genotyped pair is then calculated as the sum of expected heterozygosity from all the SNPs divided by the total number of SNPs (41238).

Pipeline for integration of KVDB and calc of Het



Viking Genetics
AI bulls

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Dams

List of combinations

Run the program to fetch the list from KVDB

15 minutes for 5 million pairs

Run the program to

- calculate the expected level of heterozygosity
- Write the results to the given list
- 1.2 hours

send the filled list back to KVDB

3 minutes

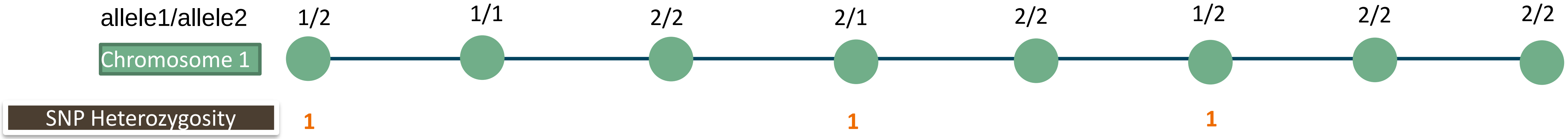
Next day

No genotype

pedigree relationships – expected Het. level
pedigree inbreeding – realized Het. level

Realized heterozygosity of genotyped individuals

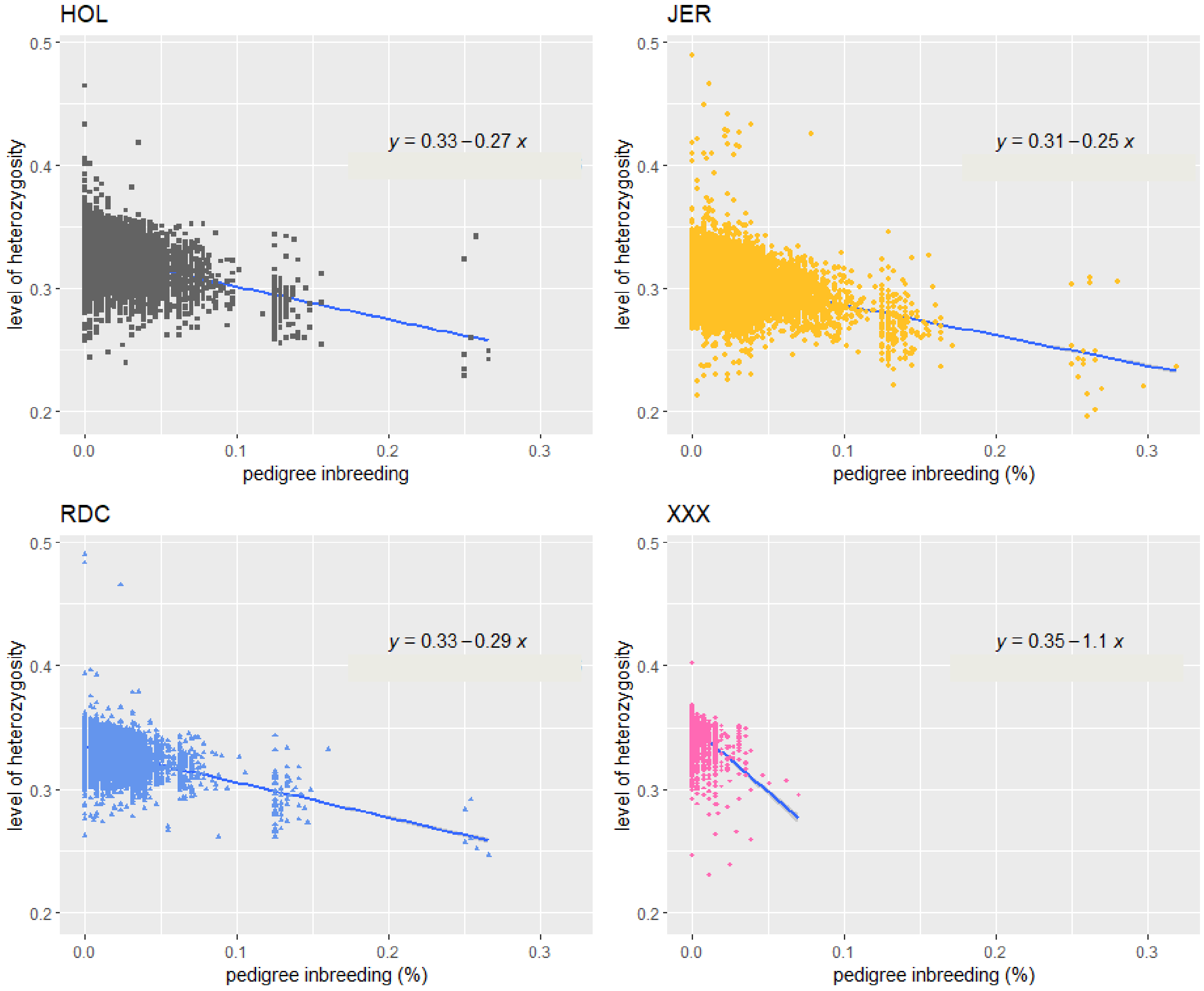
- Calculated based on individual genotypes



allele1/allele2	1/2	1/1	2/2	2/1	2/2	1/2	2/2	2/2
Chromosome 1								
SNP Heterozygosity	1			1		1		

Het. Level = Nhet/Nsnps =**3/8=0.375**

Relationship between Het. level and inbreeding level (per breed)



The gain by shifting the sire given a specific dam

Data

- Animal list – genotyped animals børn in 2014 or 2015 in Lisa’s data

- Make all possible combinations of sires and dams

- 122214 dams, 826 sires
  - 826*122214 -> 10088764 pairs in total

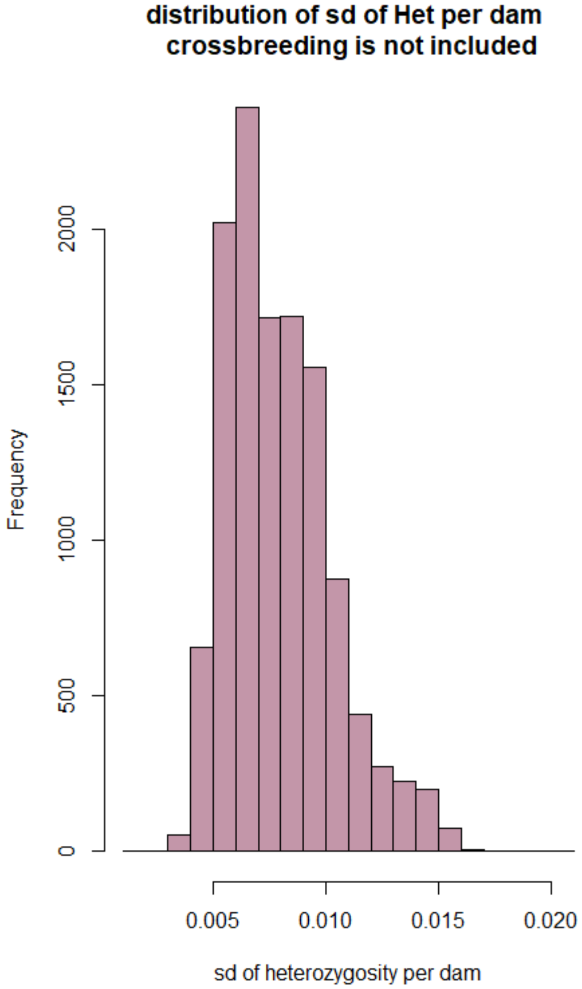
    4190453 pairs where parents are from the same breed

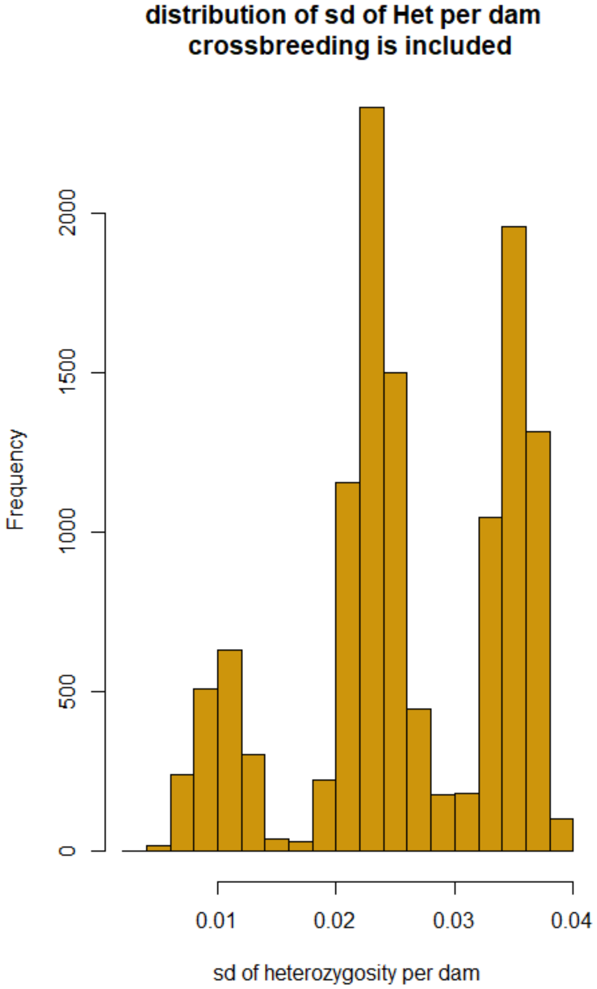
Analyses:

sd of het per dam

Max-Min of het per dam

Standard deviation of het per dam





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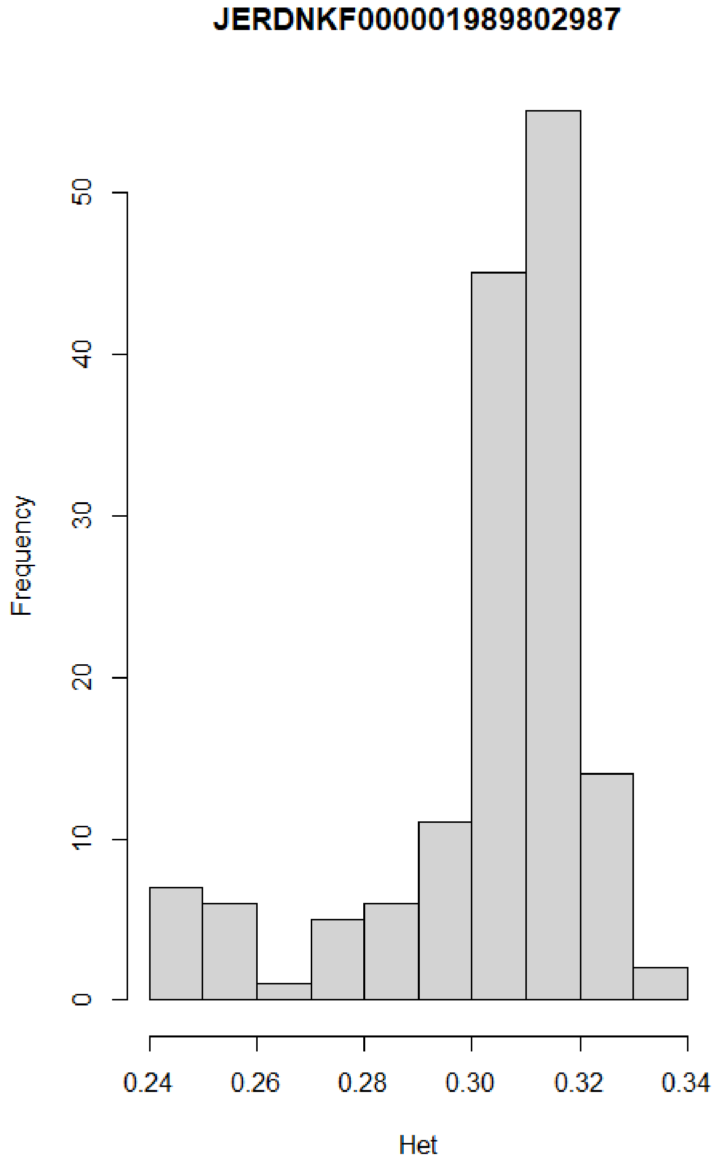
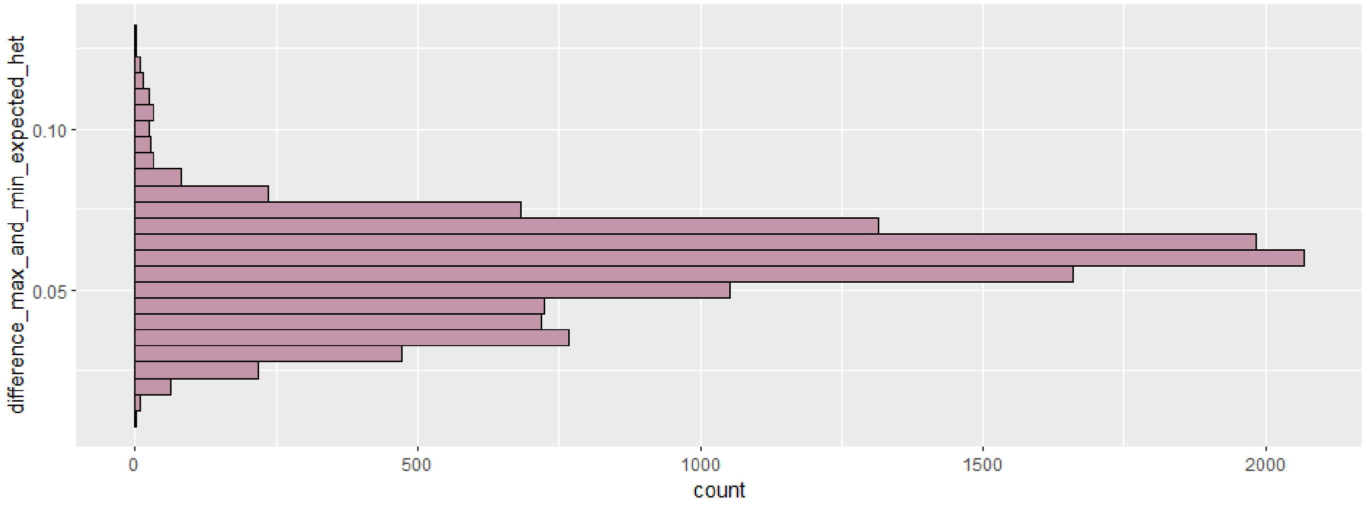
The animal with the highest SD (no crossbreeding considered)

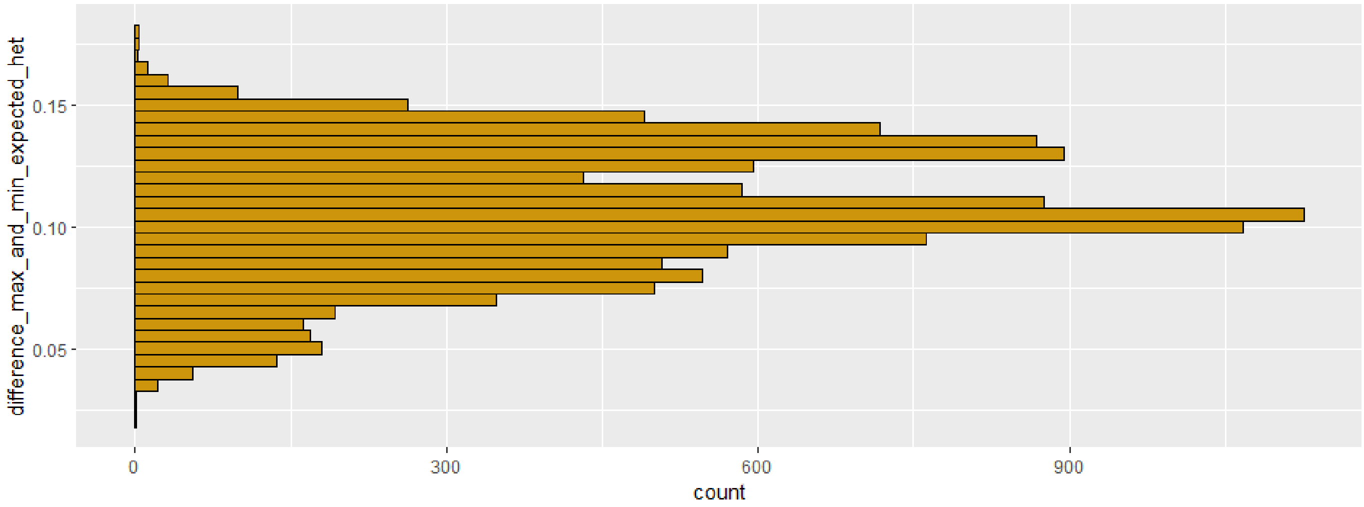


Maxhet – Minhet per dam

A distribution of difference between max and min expected het per dam (same breed)



B distribution of difference between max and min expected het per dam (same + different breeds)



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Lisa’s work
- heterozygosity and phenotype

Data

Breed	N heterozygosity, parents with gebv	+ drp
HOL	7504	5423
RDC	3125	2245
JER	9056	5975

Born 2015-2017
Heterozygosity:

	mean	sd	min	Max
HOL	0.346	0.007	0.257	0.368
RDC	0.362	0.008	0.275	0.383
JER	0.311	0.008	0.231	0.350

Correlations

HOL

	Expected het	Calculated inbreeding
Realized het	0,71	-0.39
Calculated inbreeding	-0,61	

RDC

	Expected het	Calculated inbreeding
Realized het	0,79	-0.43
Calculated inbreeding	-0,64	

JER

	Expected het	Calculated inbreeding
Realized het	0,72	-0.45
Calculated inbreeding	-0,64	

Model

- Proc glm

- DRP = heterozygosity + average parent gebv
  - corrected for the fixed effects

Results, milk- Holstein

Expected het	estimat	standard error	t value	Pr > |t|
Intercept	-61.13	14.32	-4.27	<.0001
heterozygoti	179.95	41.36	4.35	<.0001
forældregebv	1.29	0.039	33.17	<.0001

1% higher heterozygosity gives an increase of 1,8 index units ~ 188 kg milk

Results, milk- Holstein

Expected het	estimat	standard error	t value	Pr > |t|
Intercept	-61.13	14.32	-4.27	<.0001
heterozygoti	179.95	41.36	4.35	<.0001
forældregebv	1.29	0.039	33.17	<.0001

Realized het	estimat	standard error	t value	Pr > |t|
Intercept	-44.83	10.05	-4.46	<.0001
heterozygoti	140.16	30.61	4.58	<.0001
forældregebv	1.30	0.038	34.12	<.0001

Inbreeding	estimat	standard error	t value	Pr > |t|
Intercept	2.13	0.39	5.41	<.0001
heterozygoti	-99.09	26.15	-3.79	0.0002
forældregebv	1.30	0.038	34.04	<.0001

Results

- Effect of 1 % increase in expected heterozygosity: 188 kg milk
- Effect of 1 % increase in realized heterozygosity: 92 kg milk
- Effect of 1% increase in inbreeding: -65 kg milk

Conclusions

- Signficant effects of both heterozygosity (expected/realized) and inbreeding on phenotype

- Greater effect of expected heterozygosity than realized heterozygosity

Thank you very much for your attention ☺

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