

# How do imputation errors affect genomic breeding values?

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# Motivation / Objective

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- Most studies about the effects of imputation report:
  - overall correlations between GEBV
  - comparisons between software
- Further investigate the causes and patterns underlying the bias in GEBV due to imputation errors

# Material and Methods

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□ DEA-System, December 2013

→ 3494 BSW candidates 50k

masked

□ Data set 1

→ 3494 animals with 6k

Routine

□ Data set 2

→ 3494 animals with 50k

# Material and Methods

□ Data set 1

→ 3494 animals with 6k

findhap / FImpute

□ Data set 1

→ 3494 animals with 50k

□ Data set 2

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→ Prediction of GEBV for 37 traits

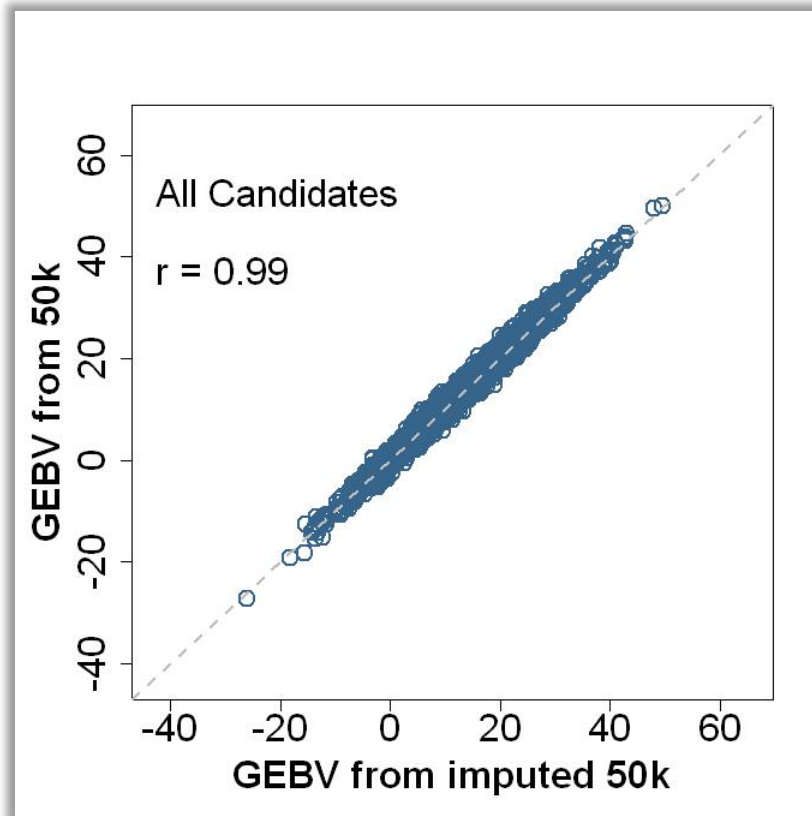
→ Comparison between GEBV from observed 50k with GEBV from imputed 50k

# Changes in ranking within TOP 50 candidates

Trait	Rank correlation		Also top 50 in imputed set	
	findhap	Flmpute	findhap	Flmpute
Milk (kg)	0.82	0.90	42	44
Fat (kg)	0.90	0.91	42	46
Protein (kg)	0.82	0.91	42	43
SCS	0.79	0.87	43	41
Workability	0.71	0.88	40	44
Udder depth	0.89	0.89	42	40
Feet & legs	0.89	0.93	45	44
Udder	0.80	0.84	44	42
Overall score	0.86	0.89	44	43
<b>Average (n=37)</b>	<b>0.84</b>	<b>0.88</b>	<b>44</b>	<b>43</b>

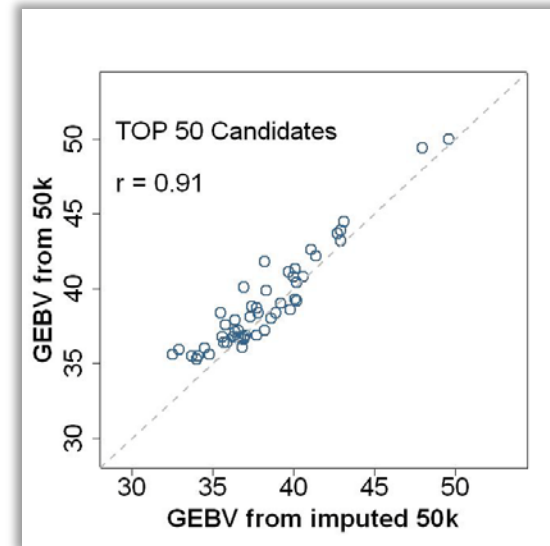
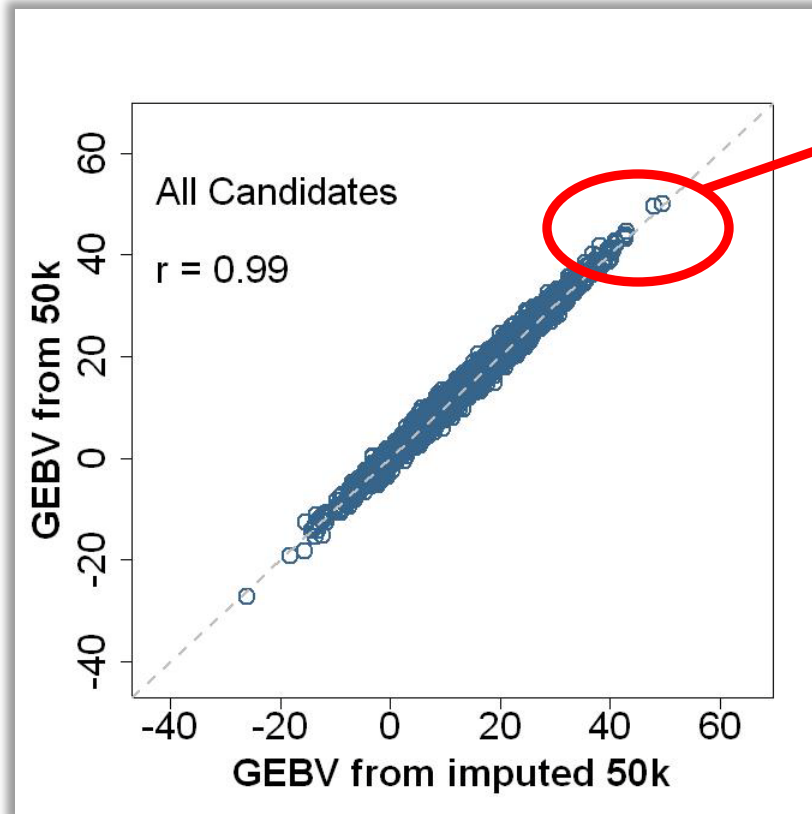
# Impact of Imputation errors on GEBV

Protein (kg) / FImpute



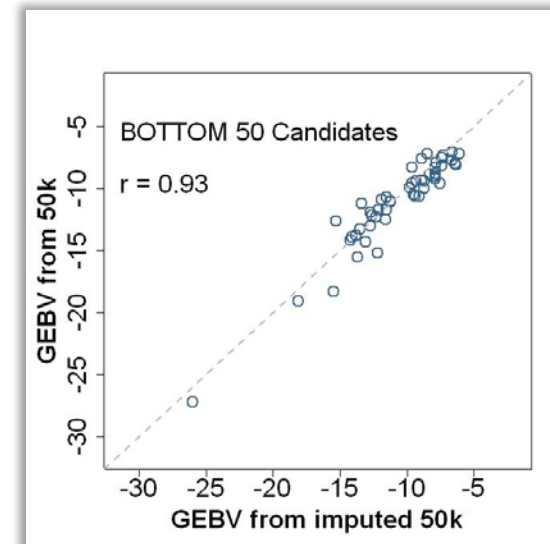
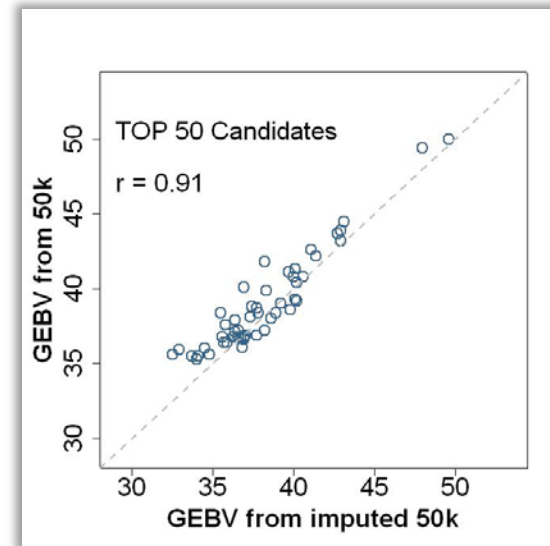
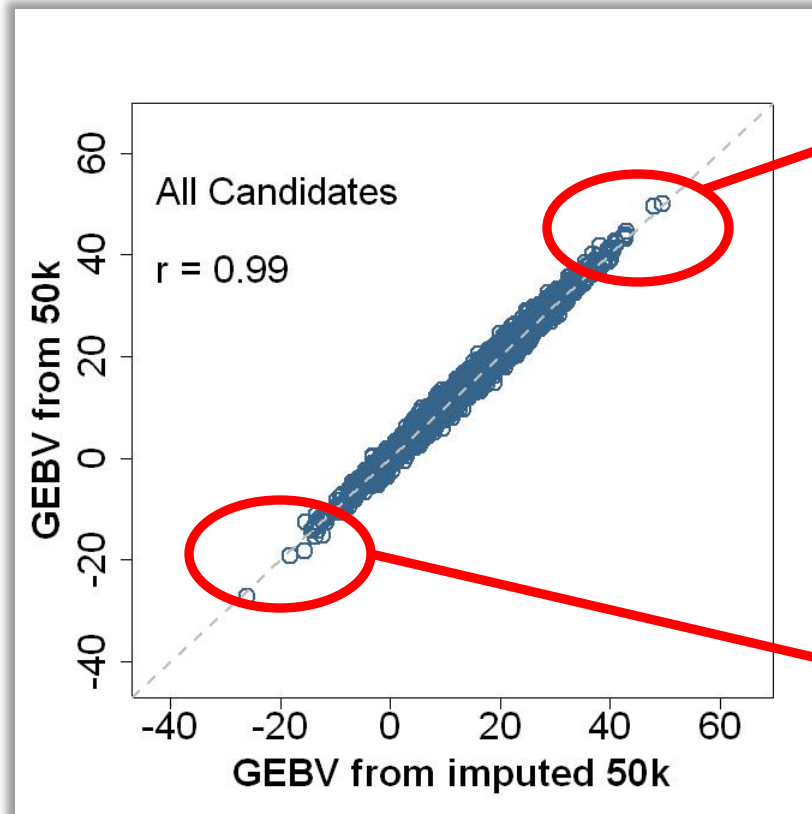
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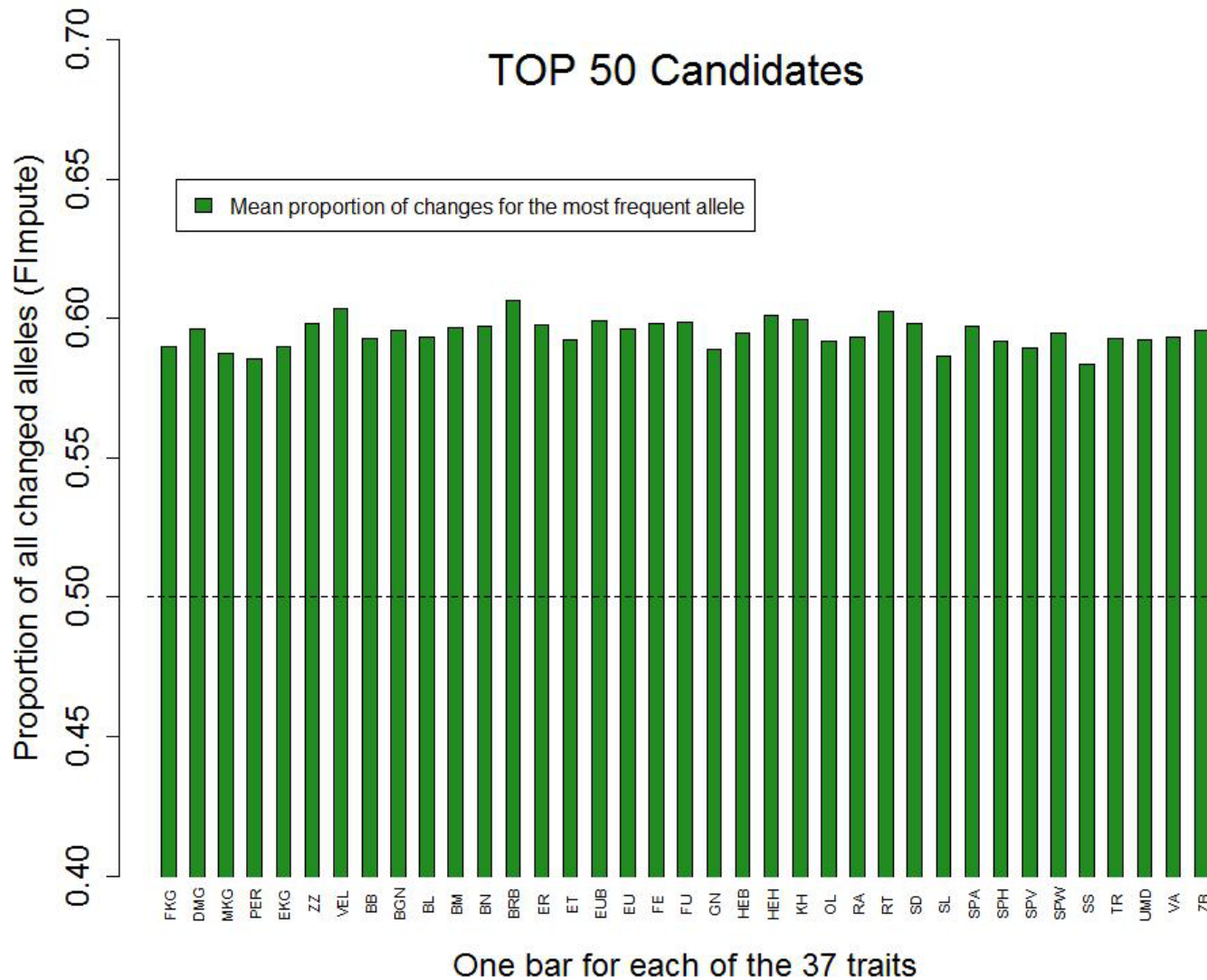
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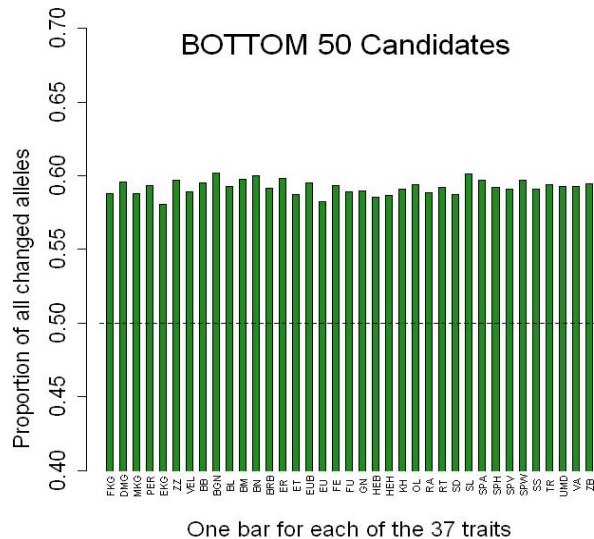
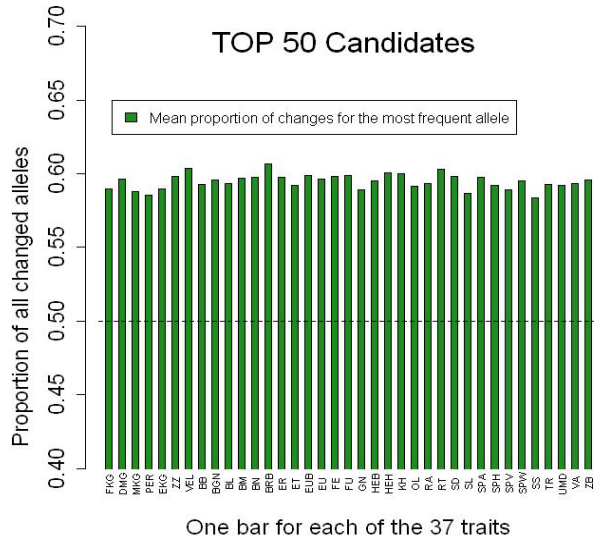
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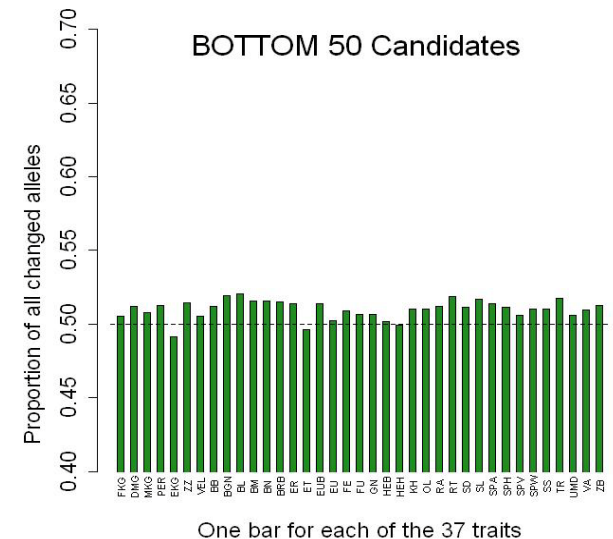
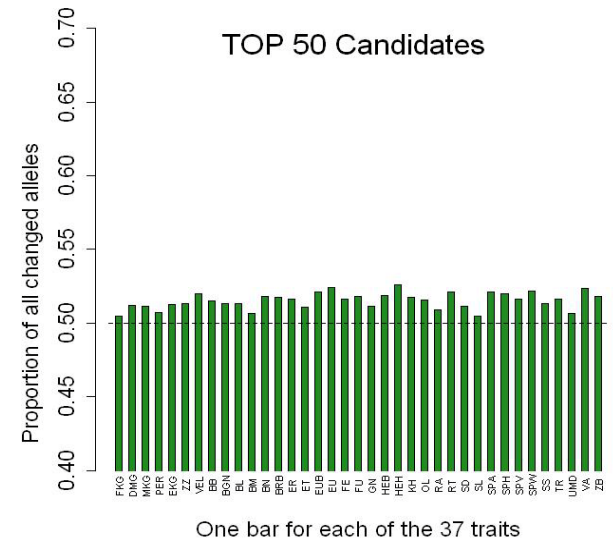


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## FImpute



## findhap



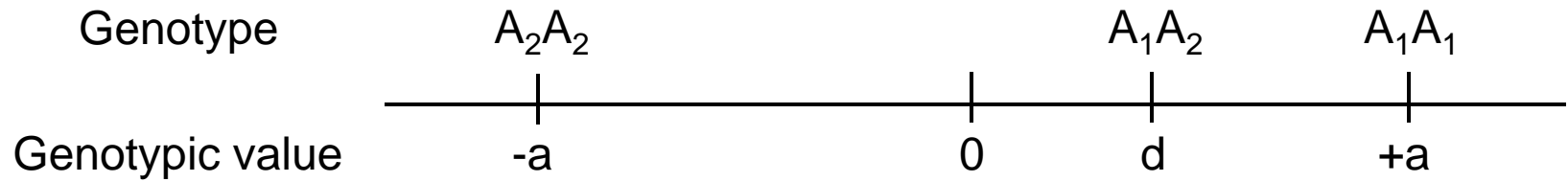
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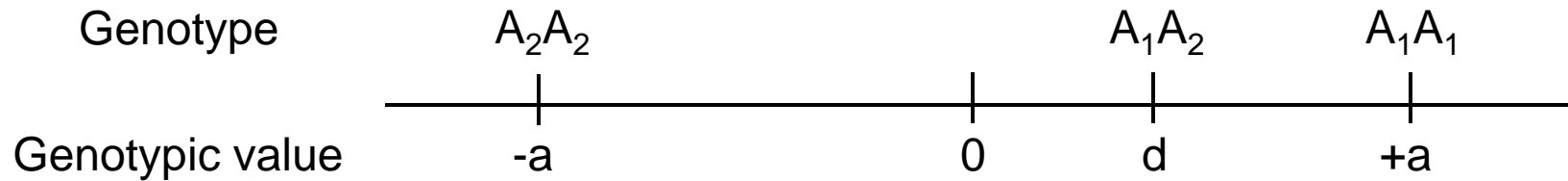
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Assuming Hardy-Weinberg equilibrium:

$$\left\{ \begin{array}{l} f(A_1A_1) = p^2 \\ f(A_1A_2) = 2pq \\ f(A_2A_2) = q^2 \end{array} \right.$$

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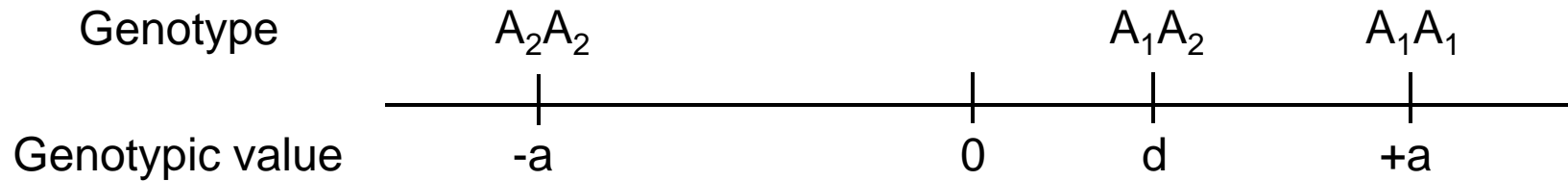
Population mean:  $M = a(p - q) + 2pqd$

\*Average effect of  $A_1$ :  $\alpha_1 = q[a + d(q - p)]$

\*Average effect of  $A_2$ :  $\alpha_2 = -p[a + d(q - p)]$

\*as deviation from the population mean

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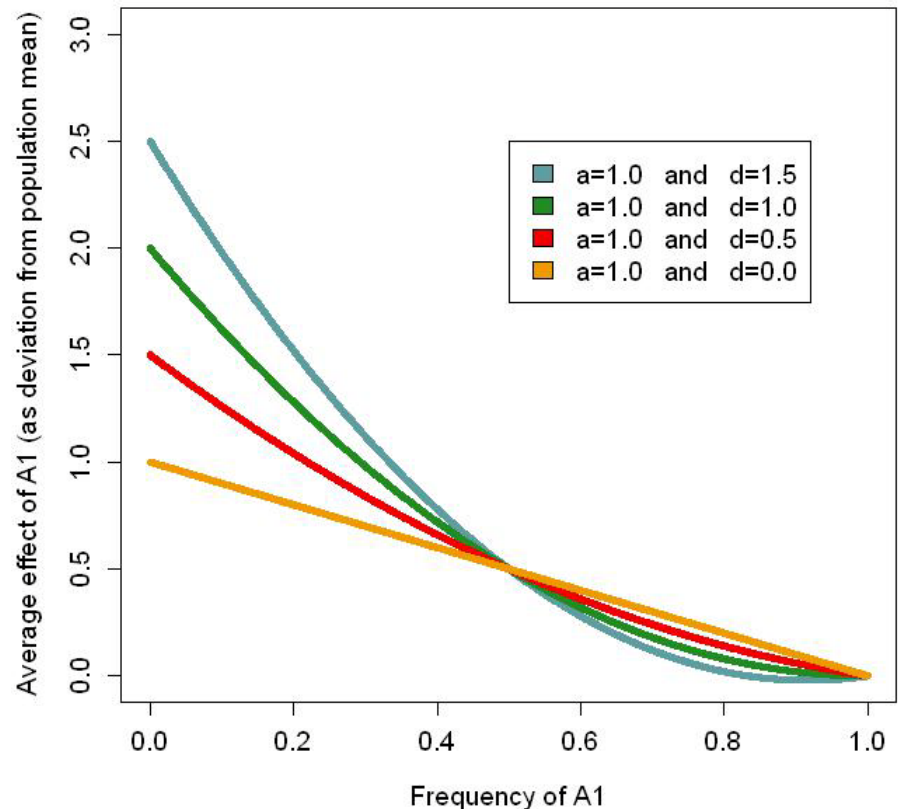
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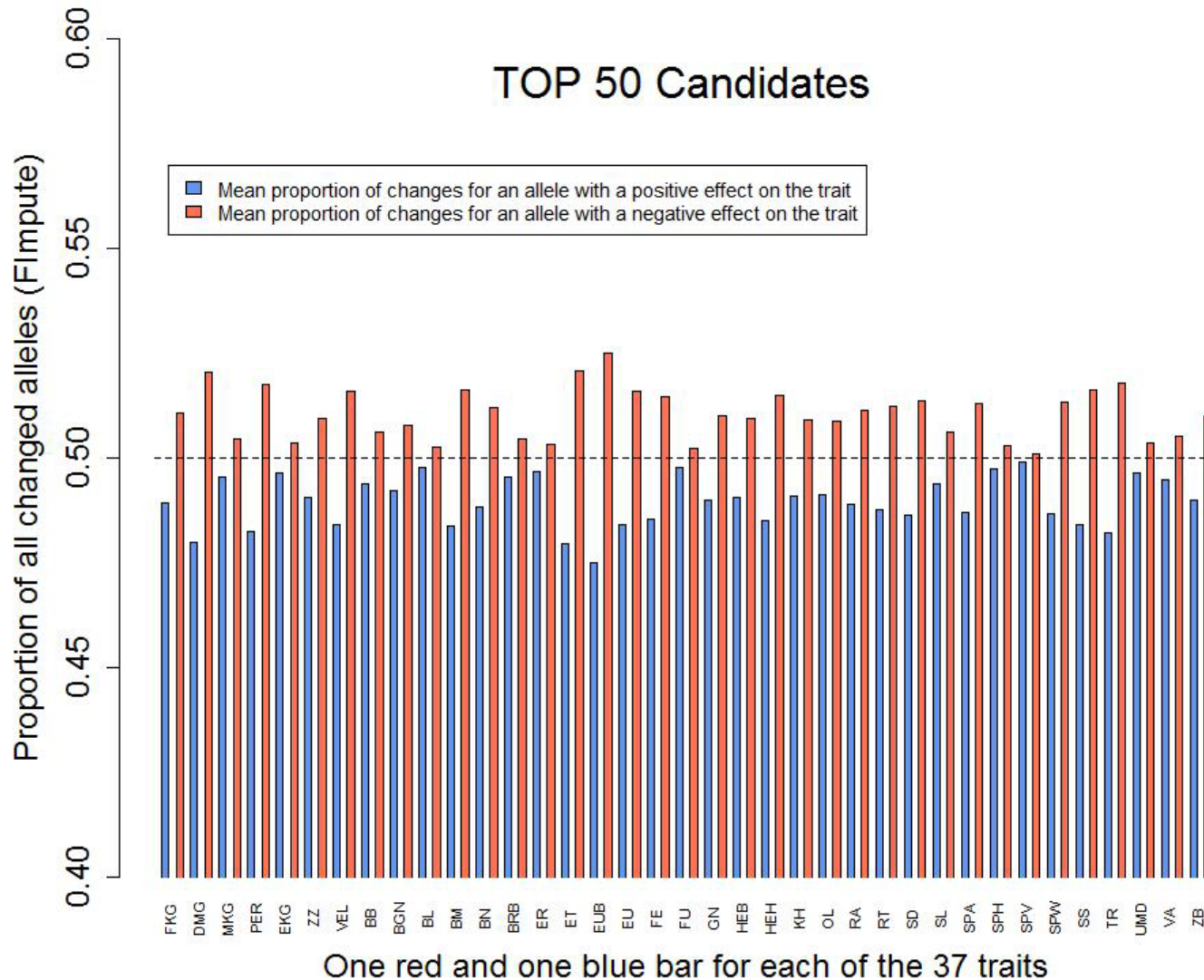


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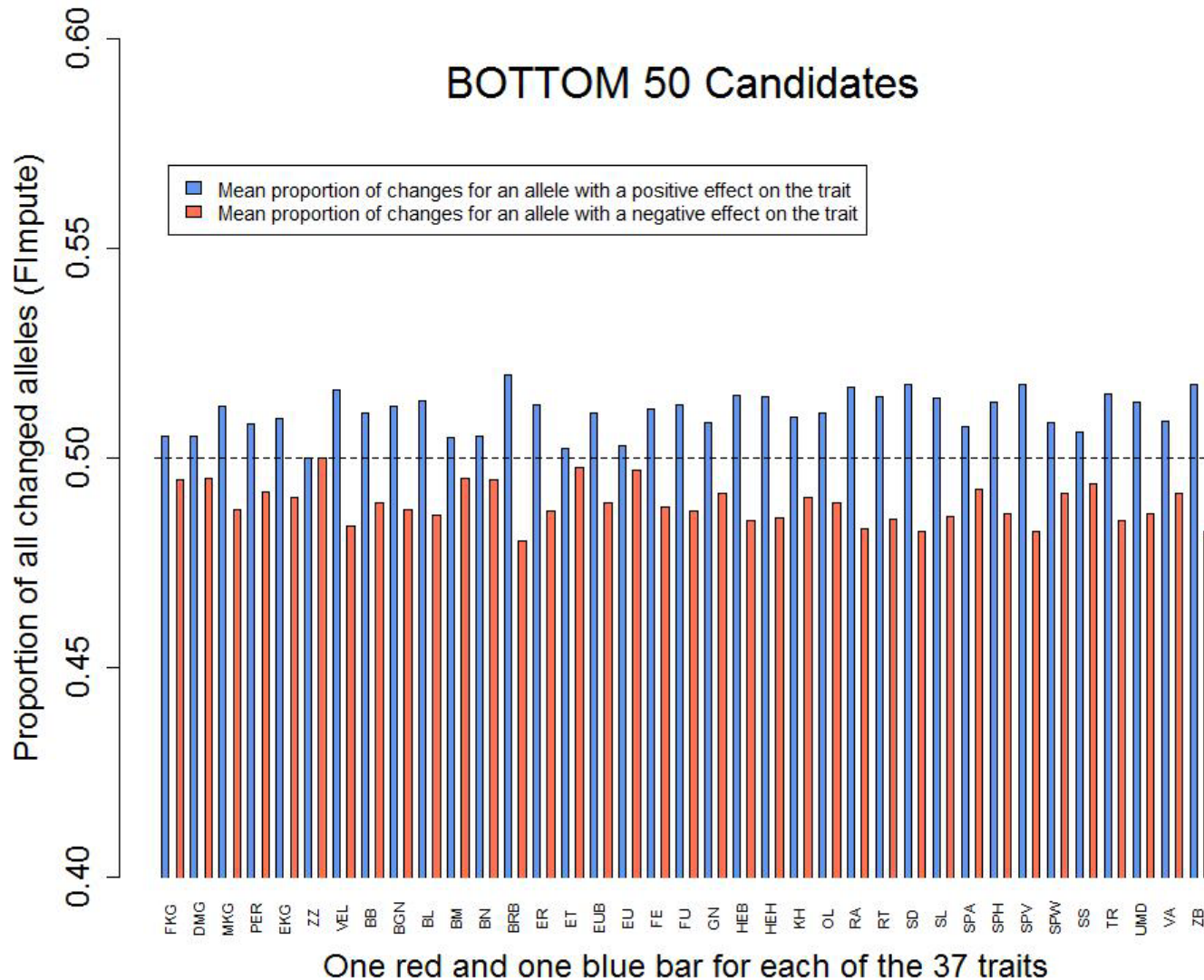
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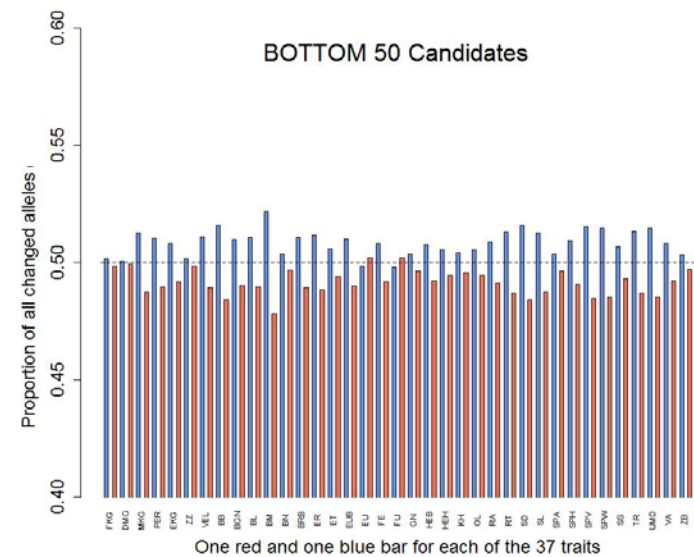
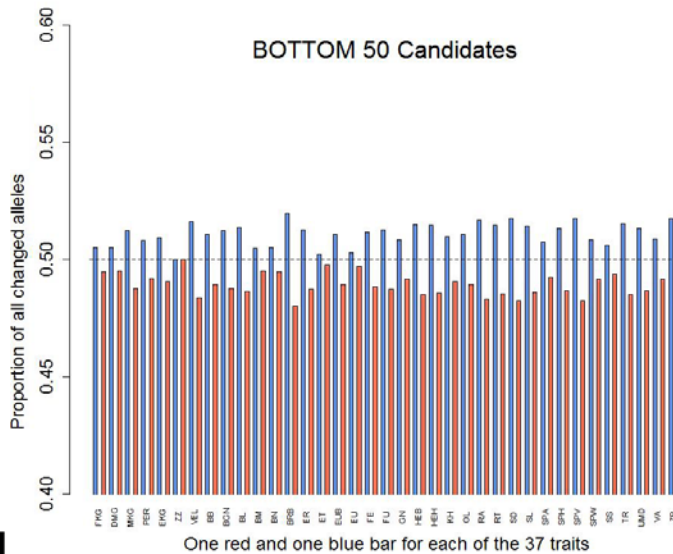
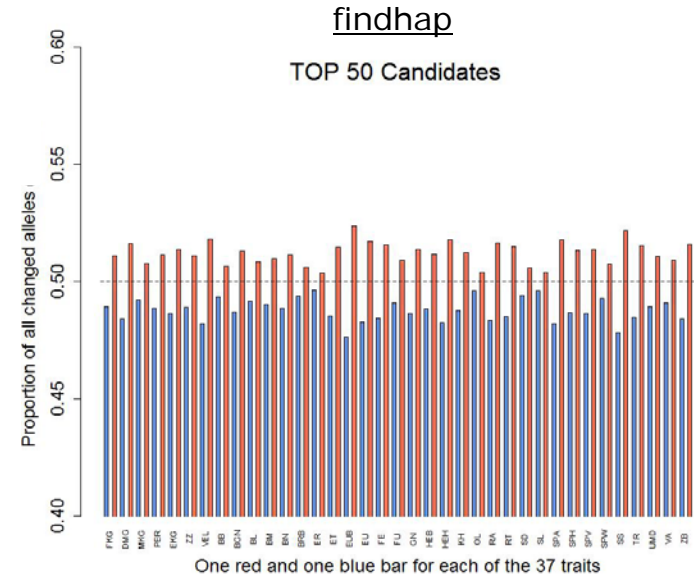
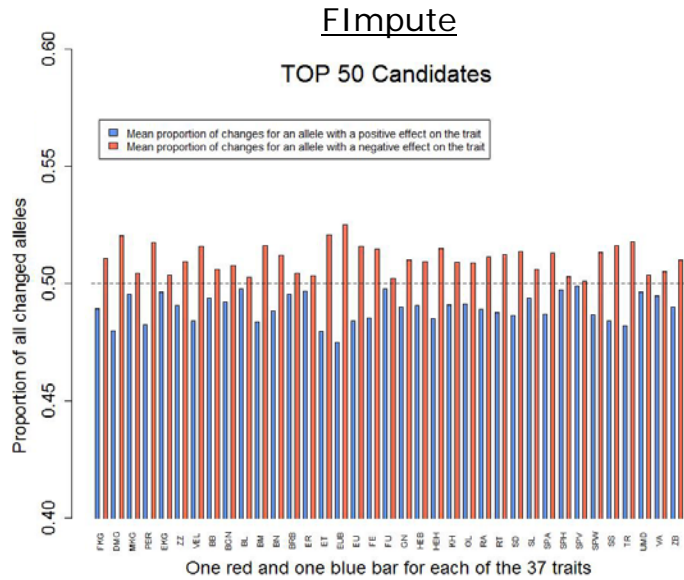
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  - ➔ downwards in top and upwards in bottom segment
- ❑ Imputation algorithms usually suggest haplotypes with higher frequency and more neutral effects
  - ➔ disadvantage for top and advantage for bottom animals
- ❑ Might have implications, especially for mixed pools of candidates genotyped at different densities

# Aknowledgements

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- Paul VanRaden (findhap)
- Mehdi Sargolzaei (FImpute)
- interGenomics

# Thanks!

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