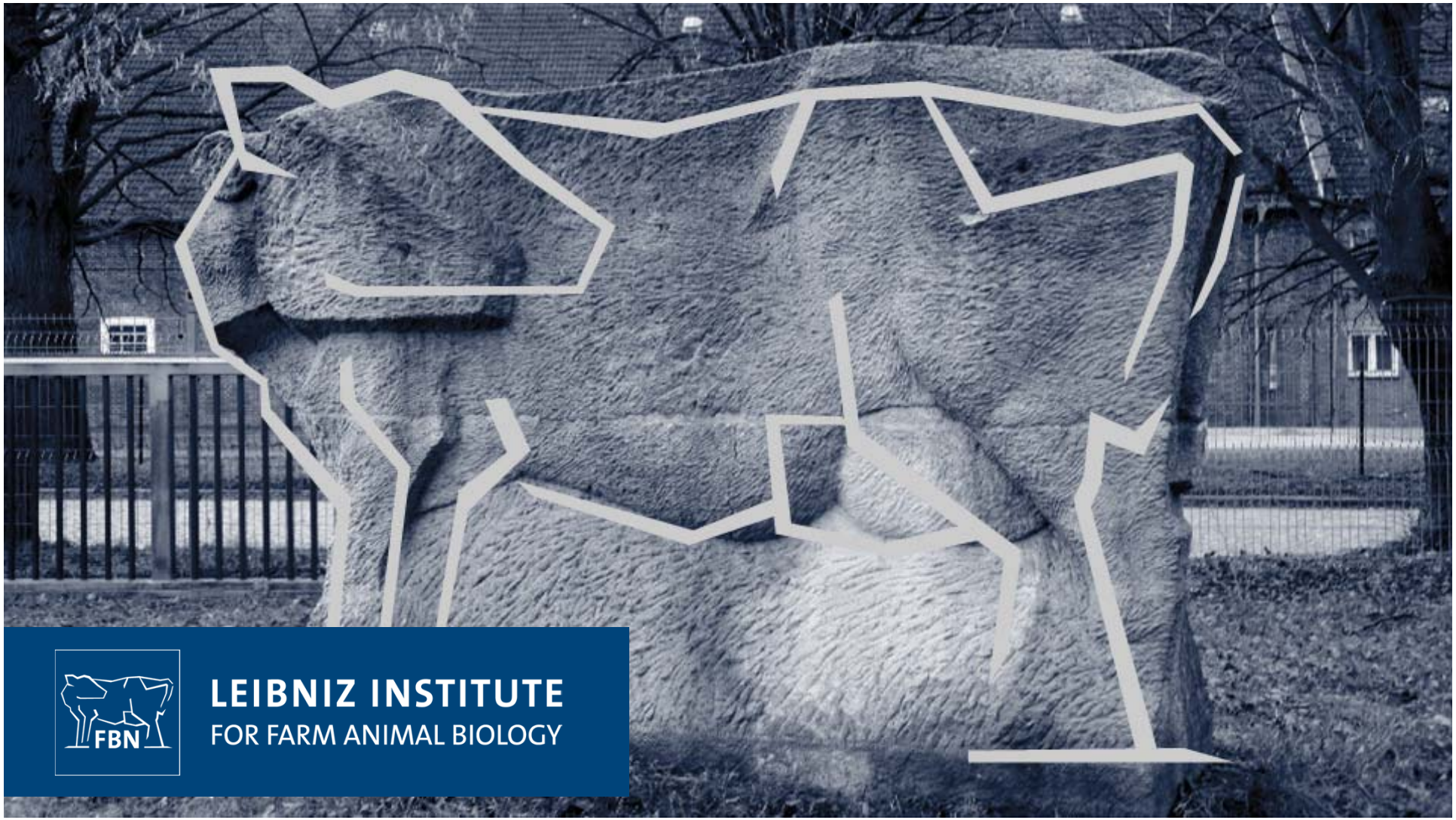
FBN
LEIBNIZ INSTITUTE
FOR FARM ANIMAL BIOLOGY

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B-spline Basis Functions for Modelling Marker Effects in Backcross Experiments

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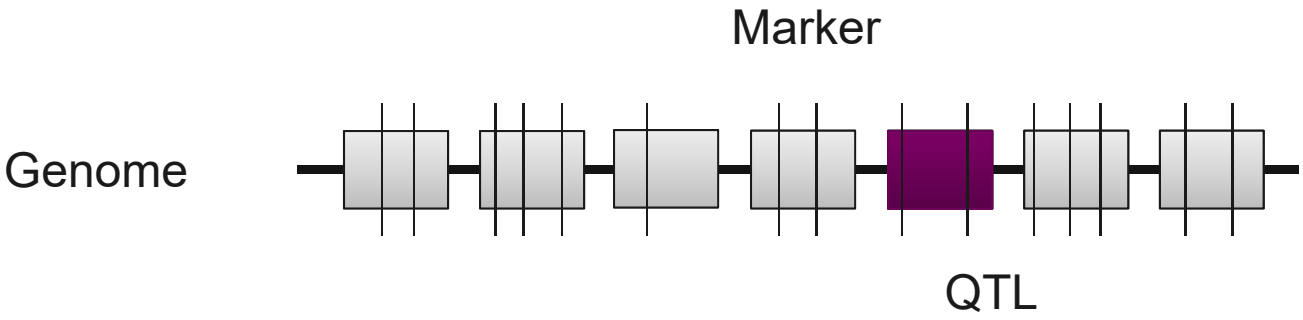
Quantitative Trait Locus

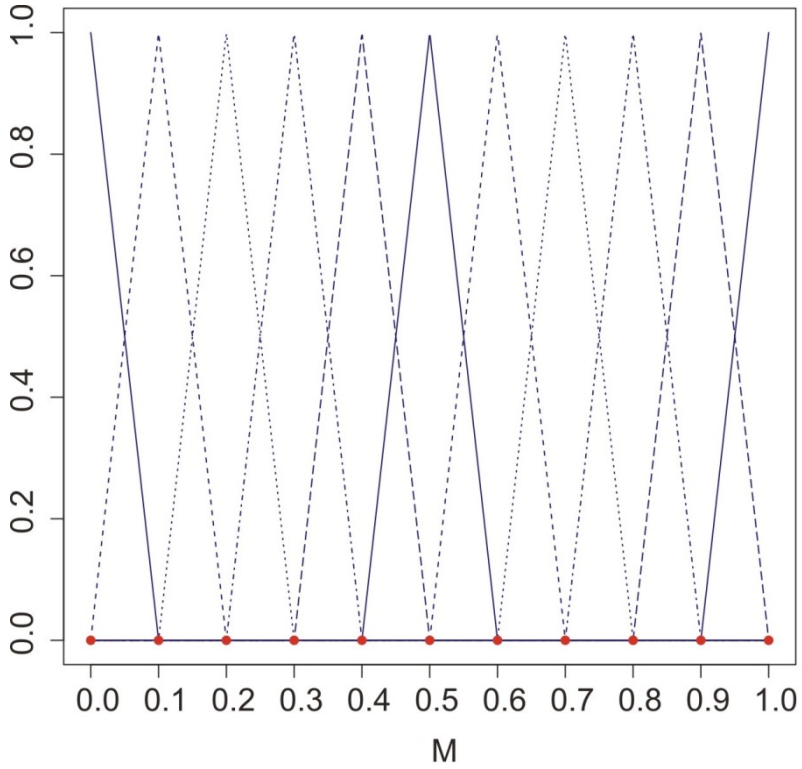
QTL (Quantitative Trait Locus)

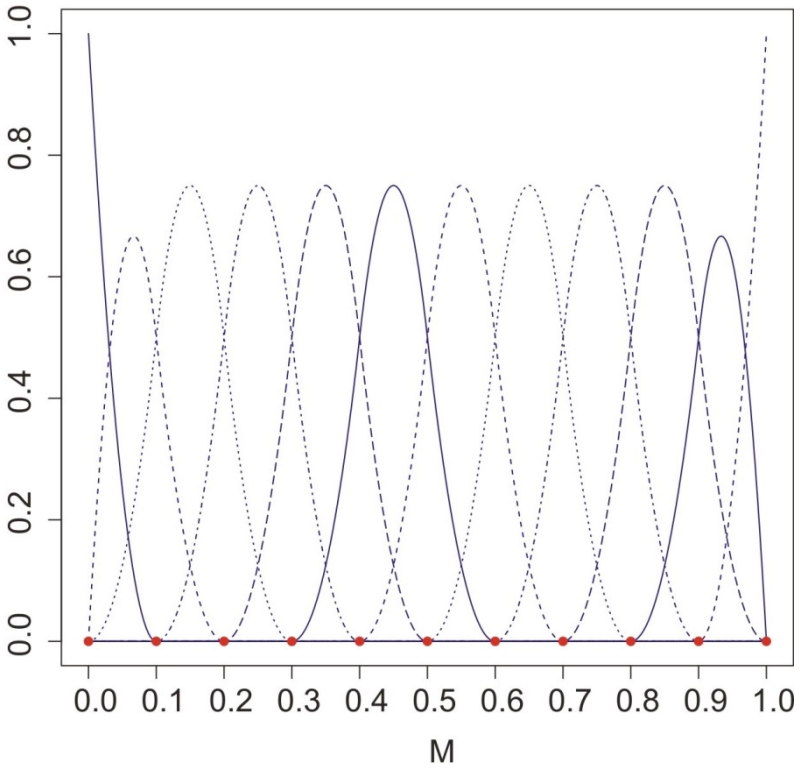
Section of genome which has an impact on quantitative traits



Backcross population

Linear Models





Bayesian Specifications

Prior distributions:

Xu 2003 (model $\boldsymbol{IN}$)

- $p(\mu) \propto 1$
- $p(m_j) = N(0, \sigma_j^2)$
- $p(\sigma_j^2) \propto 1/\sigma_j^2$
- $p(\sigma_e^2) \propto 1/\sigma_e^2$

$j = 1 \ldots \#\text{markereffects}$

B-splines (model $\boldsymbol{IB}_l$)

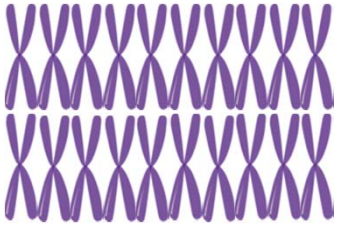
- $p(\mu) \propto 1$
- $p(b_{j'}) = N(0, \sigma_{j'}^2)$
- $p(\sigma_{j'}^2) \propto 1/\sigma_{j'}^2$
- $p(\sigma_e^2) \propto 1/\sigma_e^2$

$j' = 1 \ldots \#\text{basisfunctioneffects}$

Simulation

Mouse genome

20 chromosomes

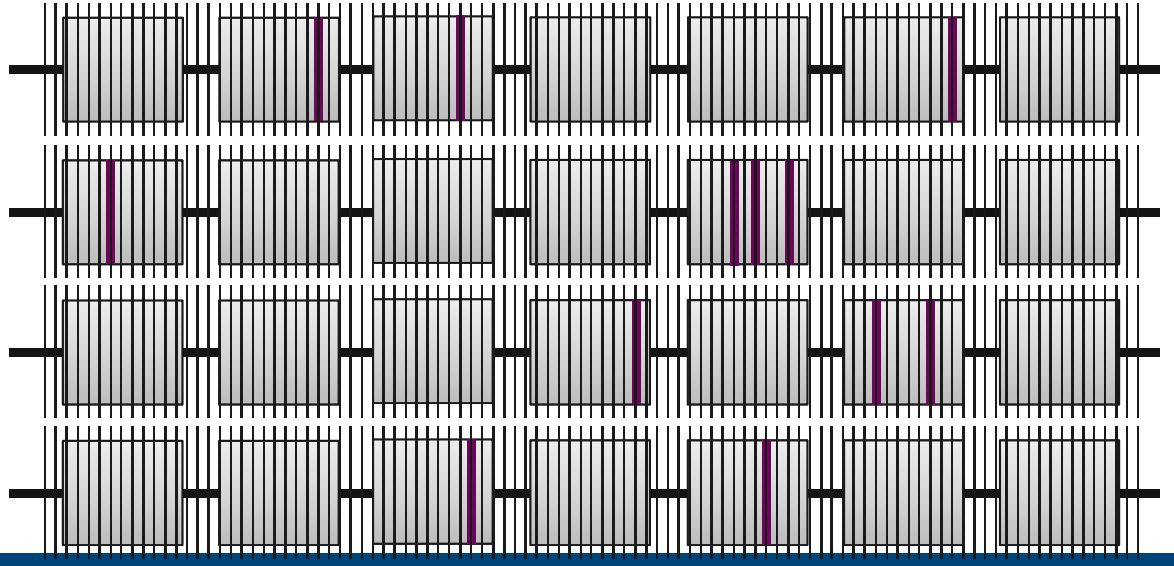


Length of 100 cM

101 equally spaced markers

12 QTLs

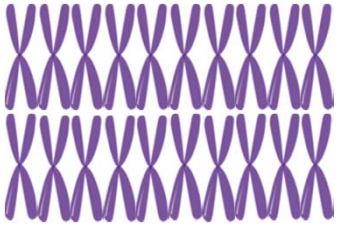
Heritabilities $h^2 \in \{0.17, 0.29\ 0.70\}$



Simulation

Mouse genome

20 chromosomes

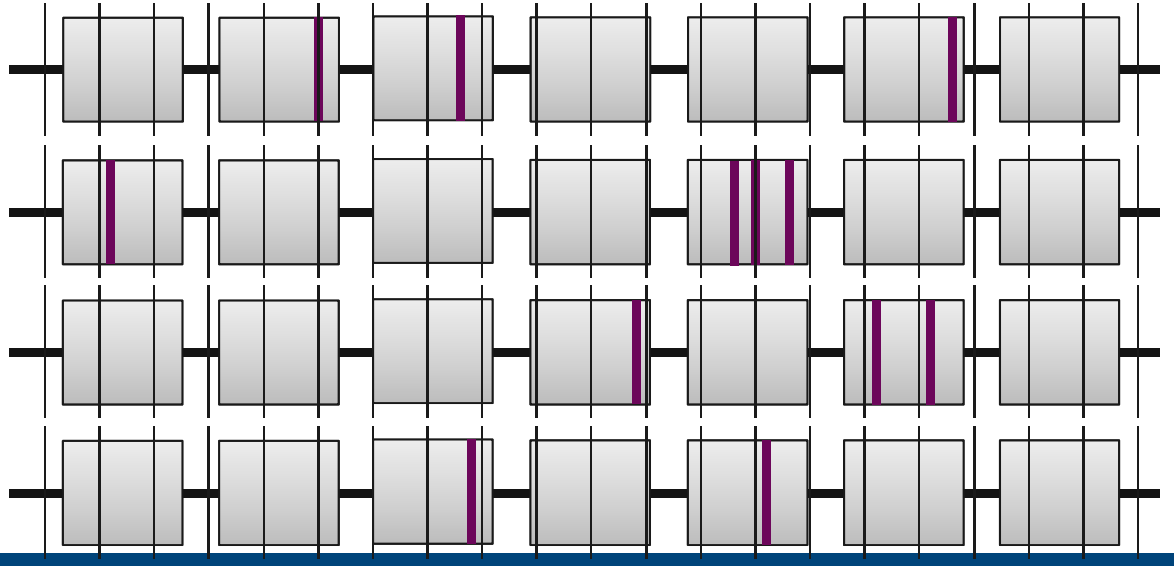


Length of 100 cM

21 equally spaced markers

12 QTLs

Heritabilities $h^2 \in \{0.17, 0.29\ 0.70\}$



Criteria for Comparison

- 200 replications and 500 individuals each
- Two different degrees $l \in \{1,2\}$ and two different number of knots $K \in \{11,21\}$

Estimated marker effects $\hat{\boldsymbol{m}}$ are means of the 200 posterior means

Estimated marker effects $\hat{\boldsymbol{m}} = \boldsymbol{B}\hat{\boldsymbol{b}}$

Genetic variance

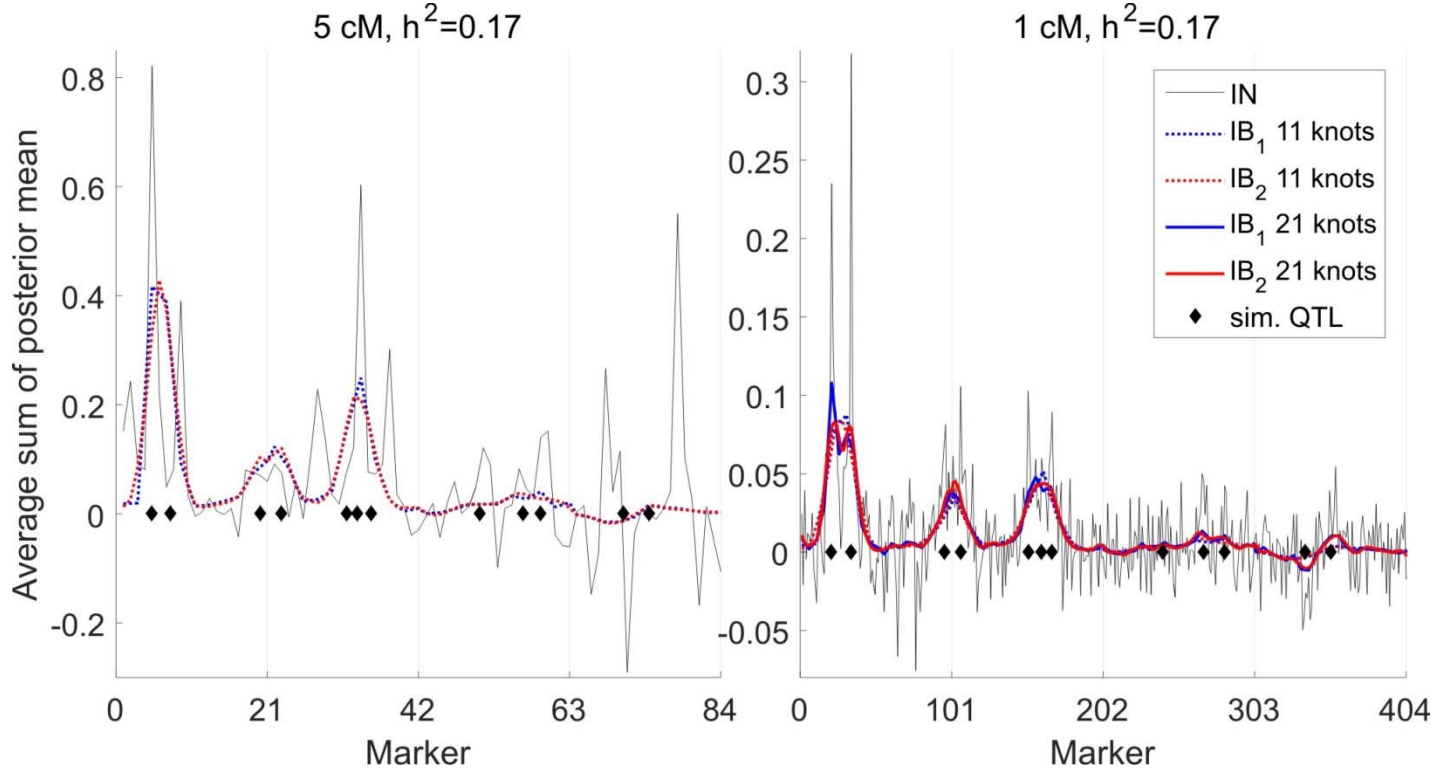
- $\sigma^2_{\hat{g}} = \hat{\boldsymbol{m}}^T \cdot \boldsymbol{R} \cdot \hat{\boldsymbol{m}}$

$\boldsymbol{R}$ Correlation matrix (Bonk et al. 2016)

Genetic value

- $\hat{g}_i = \boldsymbol{z}_i^T \cdot \hat{\boldsymbol{m}}$ $(i = 1, \ldots, n)$
- Each experiment was a training, the remaining were the test sets
- Average true genetic values at a certain proportion of selected individuals were determined

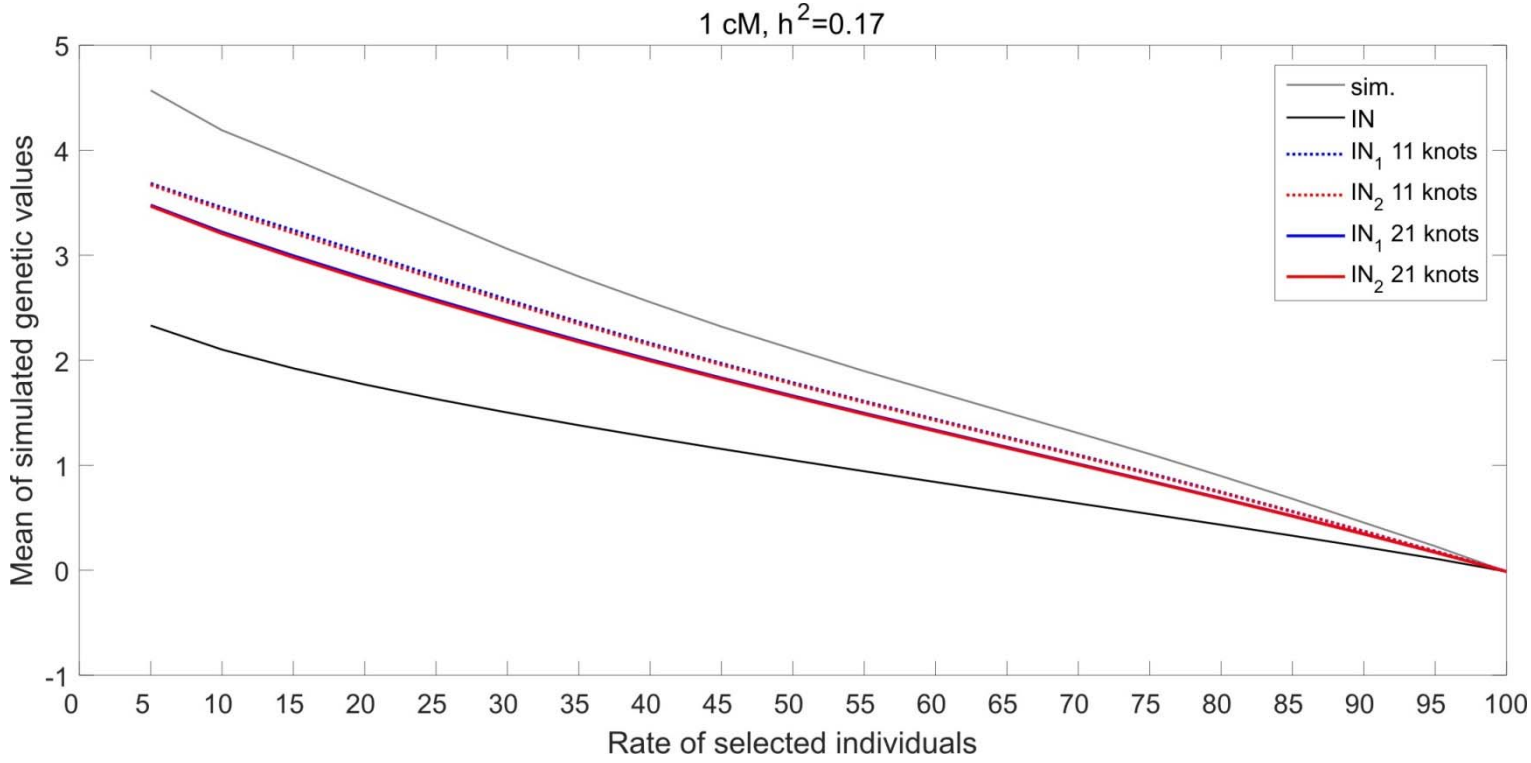
Genetic marker effects



Genetic variance

Scenario						Mean estimate: $\sigma^2_{\hat{g}}$ (rMSE)		
Nr.	h^2	cM	#Knots	σ^2_g	σ^2_e	IN	IB_1	IB_2
1	0,17	5	11	6,54	31,93	8,67 (0,15)	7,33 (0,12)	7,42 (0,12)
2		1	11	6,54	31,93	27,35 (1,18)	7,68 (0,12)	7,77 (0,13)
3		1	21	6,54	31,93	27,35 (1,18)	9,22 (0,16)	9,37 (0,17)
4	0,29	5	11	6,54	16,00	7,50 (0,09)	6,79 (0,08)	6,85 (0,08)
5		1	11	6,54	16,00	17,57 (0,62)	6,99 (0,08)	7,04 (0,08)
6		1	21	6,54	16,00	17,57 (0,62)	7,81 (0,10)	7,89 (0,10)
7	0,7	5	11	6,54	2,80	6,63 (0,03)	6,42 (0,04)	6,42 (0,04)
8		1	11	6,54	2,80	8,73 (0,13)	6,47 (0,04)	6,47 (0,04)
9		1	21	6,54	2,80	8,73 (0,13)	6,68 (0,03)	6,70 (0,03)

Genetic prediction



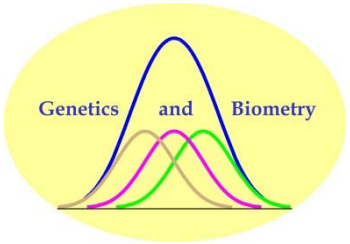
Conclusion

- Improvement of precision of estimated marker effects and genetic variances
- Reduction of number of parameters
- Decreased computational time

Suitable and fast solution to improve a standard Bayesian analysis

Thank you for your attention

Thanks to the
European Federation of Animal Science (EAAP)



Literature:

- Bonk S., Reichelt M., Teuscher F., Segelke D., Reinsch N. (2016): *Mendelian sampling covariability of marker effects and genetic values.* Genet Sel Evol 48: 36
- Xu S. (2003): *Estimating polygenic effects using markers of the entire genome.* Genetics 163: 789-801

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