

Binary traits: the speed-accuracy trade-off

Genomic prediction of a binary trait using a threshold model

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Background

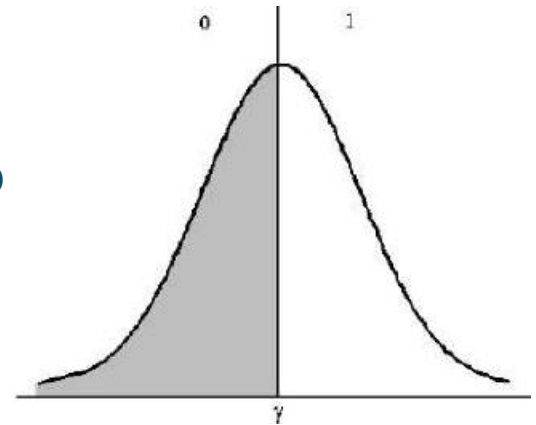
- Many traits have a **continuous** phenotypic distribution
- **Categorical traits** have a **discontinuous** distribution of phenotypes
 - **Binary** traits (0/1, all-or-none, absent-present)
 - **Ordered categorical** traits (e.g. small, medium, large)
 - **Unordered categorical** traits (e.g. red, yellow, blue)
- This has implications for genetic evaluation!

Linear models are incorrect for categorical traits

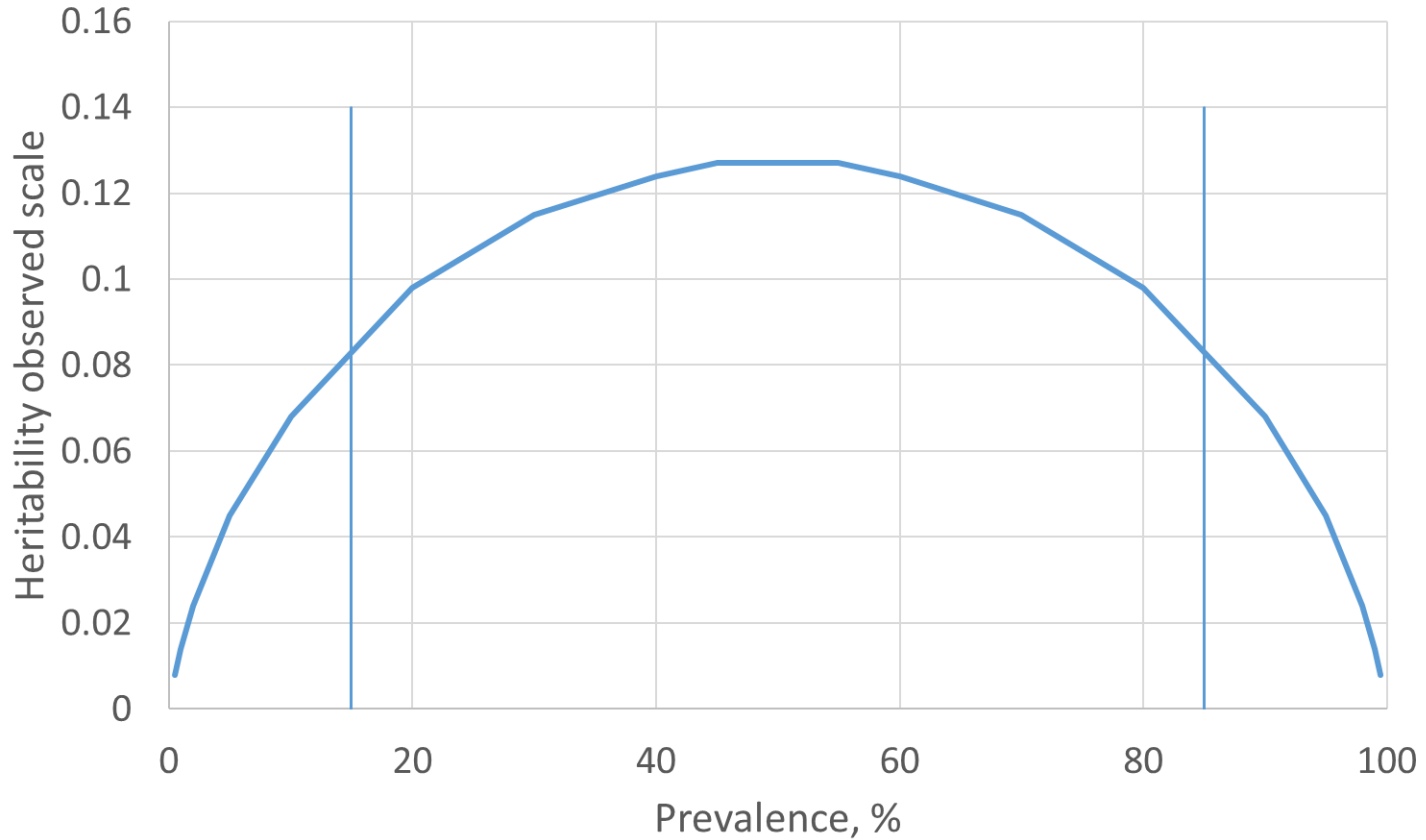
- Common linear methods to estimate genetic merit
 - Assume a **continuous** phenotypic distribution
- Assumptions are **violated** for categorical traits
 - Residual variance can be very heterogeneous
 - Extreme category problem: all observations in a fixed-effect class have the same score
 - Genetic and residual effects not independent
 - Heritability and variance components on observed scale depend on prevalence

Alternative approach: threshold model

- Presume unobserved continuous trait underlying the observed categorical trait: **liability**
- **Threshold** for every change in category
- Liability is **unobserved** so
 - Unknown mean and variance
 - Either mean or a threshold set to zero
 - Residual variance set to 1



Heritability underlying scale is 0.20



Prediction of effects

- Prediction of genetic effects (EBVs)
 - Threshold model on liability scale
 - Linear model on observed scale
- Consensus in literature (analyses based on pedigree)
 - At intermediate range of prevalence (15-85%)
 - Rank correlation of EBV very high
 - Linear model much faster
- Is the benefit of using a threshold model affected by the use of genomic information?

Validation study - aim

- Compare realised reliability for various ways to evaluate a binary trait
- Models
 - Threshold model with Gibbs sampling
 - Threshold model with iterative solving (MiXBLUP)
 - Linear model with iterative solving (MiXBLUP)
- Genetic relationships
 - Pedigree information
 - Pedigree + genomic information

MiXBLUP

Validation study - simulation

■ Population simulated using QMSim

- 10 generations of 1,080 individuals out of 15 sires and 90 dams (N=10,905)
- 19 chromosomes, with 19,000 SNP and 760 QTL

■ Trait

- True breeding value on liability scale ($h^2=0.20$)
- Liability is simulated phenotype, affected by sex and herd
- **Observed phenotype** is derived binary trait with prevalence of 15%

Validation study - design

- Generation 1-9 have phenotypes (training population)
- Generation 10 (validation population)
- Generation 7-10 are genotyped
- **Realised reliability** is squared correlation between EBV and TBV of animals in generation 10

Validation study: realised reliability

Method		Pedigree		Pedigree+genomic	
Threshold – Gibbs		19.1		22.0	
Threshold – solving		19.0		21.8	
Linear - solving	+0.8	18.2	+1.9	20.1	+1.7

- Both threshold models give virtually the same result
- Benefit threshold model increases with use of genomics (+1.7 vs +0.8)
- Benefit threshold model similar to benefit using genomics (+1.7 vs +1.9)

Validation study: run time

■ Run-time

- Substantially longer for Gibbs sampling than iterative solving ($\sim 60x$)
- Only slightly longer for threshold model than linear model ($\sim 1.5-2x$)

Wrap-up

- The benefit of using a threshold model in this example
 - Is comparable to the benefit of using genomic information
 - Is higher when using genomic information than pedigree information only

Genomic breeding values for binary traits?
At least consider a threshold model!

Acknowledgments



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