What to do with non-normal data:

Classical test theory versus item response theory in estimating variance components

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BACKGROUND

Observation

Many phenotypes are expressed as sum scores based on a limited number of items

Problems

- Sum scores are ordinal measures
- Sum scores are often not normally distributed
- Items change over time
- Ceiling and floor effects
- Attenuated correlations
- What to do with subjects with missing items?

Consequences

Using common linear genetic models that assume normal distributions, inferences regarding variance components are biased (Derks et al., 2004)*

COMMON SOLUTIONS

- Transformations (e.g., log, √)
  Problems: still ordinal, interpretation more difficult, missing items not resolved
- Threshold modelling:
  Problems: still not using all information, missing items problem not resolved

USING AN IRT MEASUREMENT MODEL

Advantages

- No problems when items are missing
- Avoid bias due to distribution violations: specify your own distribution
- Avoid bias due to limited set of items

Disadvantage

- Model estimation is a computational challenge!

Solution

Bayesian modelling with MCMC estimation

ITEM RESPONSE THEORY

We assume every subject has a score on a latent variable, θ_i

Whether an item is scored 0 or 1, depends on a subject parameter θ and an item parameter β

$$p(Y_{ij} = 1 \mid \theta_i, \beta_j) = \exp(\theta_i - \beta_j) / (1 + \exp(\theta_i - \beta_j))$$

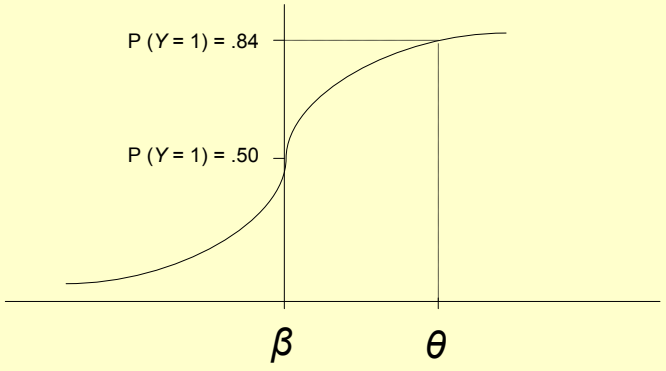
The higher the subject parameter, and the lower the item parameter, the more likely a positive response is to occur.

The *β*s can be conceived of as thresholds in the usual threshold model for nominal or ordinal traits, except that we now have multiple items as indicators for only one latent variable

IRT models can be extended to include polytomous items, factor loadings, covariates, repeated measures, hierarchical structures, modelling of missing data, multidimensionality….

The variance of the subject parameters can be decomposed into genetic and non-genetic parts.

$Var(\theta) = A + D + C + E$



ILLUSTRATION

Attention problems were measured in 3,021 adult twins aged 18-30 using seven polytomous items. Liability *θ* was modelled using a one-parameter logistic model. Variance of *θ* was decomposed into additive genetic variance and non-shared environmental variance. Inference is based on the posterior marginal distributions using an MCMC algorithm implemented in WinBUGS.

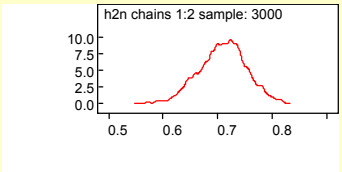
Burn-in: 500 iterations, inference: 2 chains of 1500 iterations

Computation time: +/- 30 minutes

Results heritability for attention problems:

Mean: 0.71 **SD:** 0.04

Quantiles: **2.5%:** 0.62 **50%:** 0.71 **97.5%:** 0.79



* Derks EM, Dolan CV, Boomsma DI (2004). Effects of censoring on parameter estimates and power in genetic modeling. *Twin Res.* 7(6):659-69.