

Bayesian Variable Selection in Searching for Additive and Dominant Effects in Genome-wide Data

Supplementary Text S1

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Details of the sampling scheme

The conditional distributions for Gibbs sampling are derived below. Note that ω and ϕ are marginalized out analytically and need not enter the computations. To sample from the posterior

$$p(\gamma, \mathbf{t}, \beta, \alpha, \sigma^2, \tau^2, \mathbf{X} | \mathbf{y}, \mathbf{X}_{obs}) \propto p(\gamma, \mathbf{t}) p(\beta | \sigma^2, \tau^2, \gamma, \mathbf{t}) p(\alpha | \sigma^2) p(\sigma^2) p(\tau^2) p(\mathbf{X} | \mathbf{X}_{obs}) p(\mathbf{y} | \beta, \alpha, \sigma^2, \mathbf{X})$$

the following Gibbs sampling scheme is used (with all steps conditional on observed data \mathbf{y} and \mathbf{X}_{obs}):

1. $x_{ij} | \mathbf{X}_{-ij}, \gamma, \mathbf{t}, \beta, \alpha, \sigma^2, \tau^2$ for all i, j .
2. $\tau^2 | \gamma, \mathbf{t}, \beta, \alpha, \sigma^2, \mathbf{X}$.
3. $\gamma, \mathbf{t} | \tau^2, \mathbf{X}$.
4. $\sigma^2 | \gamma, \mathbf{t}, \tau^2, \mathbf{X}$.
5. $\beta, \alpha | \gamma, \mathbf{t}, \sigma^2, \tau^2, \mathbf{X}$.

Note that $p(\beta | \sigma^2, \tau^2, \gamma, \mathbf{t}) = p(\beta_\gamma | \sigma^2, \tau^2) p(\beta_{-\gamma}) = p(\beta_\gamma | \sigma^2, \tau^2)$ or 0 if some $\beta_j \neq 0$ with $\gamma_j = 0$. Further, only variants with $\gamma_j = 1$ affect the likelihood: $p(\mathbf{y} | \beta, \alpha, \sigma^2, \mathbf{X}) = p(\mathbf{y} | \beta_\gamma, \alpha, \sigma^2, \mathbf{X})$.

Step 1

$$\begin{aligned} p(x_{ij} | \dots) &\propto p(x_{ij}) p(\mathbf{y} | \beta, \alpha, \sigma^2, x_{ij}, \mathbf{X}_{-ij}) \\ &= \text{Categorical}(x_{ij} | \theta_{ij}^0, \theta_{ij}^1, \theta_{ij}^2) p(\mathbf{y} | \beta, \alpha, \sigma^2, x_{ij}, \mathbf{X}_{-ij}) \\ p(x_{ij} | \dots) &= \text{Categorical}(x_{ij} | \hat{\theta}_{ij}^0, \hat{\theta}_{ij}^1, \hat{\theta}_{ij}^2), \end{aligned}$$

where $\hat{\theta}_{ij}^k \propto \theta_{ij}^k p(\mathbf{y} | \beta, \alpha, \sigma^2, x_{ij} = k, \mathbf{X}_{-ij})$ and $\sum_k \hat{\theta}_{ij}^k = 1$.

Step 2

Here, $\boldsymbol{\tau}^2$ has one component in BMA A (τ_A^2) and two in BMA A/AH (τ_A^2, τ_H^2). The components can be sampled independently with only the corresponding part of $\boldsymbol{\beta}$ affecting the sampling.

$$\begin{aligned}
p(\tau_t^2 | \dots) &\propto p(\tau_t^2) p(\boldsymbol{\beta}_t | \sigma^2, \tau_t^2, \boldsymbol{\gamma}, \mathbf{t}) \\
&\propto (\tau_t^2)^{-(\nu_{\tau_t^2}/2+1)} \exp\left(-\frac{1}{2\tau_t^2} \nu_{\tau_t^2} s_{\tau_t^2}^2\right) (2\pi\sigma^2\tau_t^2)^{-q/2} \exp\left(-\frac{1}{2\sigma^2\tau_t^2} \boldsymbol{\beta}_{t,\gamma}^T \boldsymbol{\beta}_{t,\gamma}\right) \\
&\propto (\tau_t^2)^{-((\nu_{\tau_t^2}+q)/2+1)} \exp\left(-\frac{1}{2\tau_t^2} (\nu_{\tau_t^2} s_{\tau_t^2}^2 + \frac{1}{\sigma^2} \boldsymbol{\beta}_{t,\gamma}^T \boldsymbol{\beta}_{t,\gamma})\right) \\
p(\tau_t^2 | \dots) &= \text{Inv-}\chi^2\left(\tau_t^2 | \nu_{\tau_t^2} + q, \frac{\nu_{\tau_t^2} s_{\tau_t^2}^2 + \frac{1}{\sigma^2} \boldsymbol{\beta}_{t,\gamma}^T \boldsymbol{\beta}_{t,\gamma}}{\nu_{\tau_t^2} + q}\right).
\end{aligned}$$

Step 3

A Metropolis-Hastings step is used to update $\boldsymbol{\gamma}$ and \mathbf{t} . Parameters $\boldsymbol{\beta}$, $\boldsymbol{\alpha}$ and σ^2 are integrated out analytically at this step giving the following marginal likelihood:

$$\begin{aligned}
p(\mathbf{y} | \mathbf{X}, \boldsymbol{\tau}^2, \boldsymbol{\gamma}, \mathbf{t}) &= \int p(\boldsymbol{\beta} | \sigma^2, \boldsymbol{\tau}^2, \boldsymbol{\gamma}, \mathbf{t}) p(\boldsymbol{\alpha} | \sigma^2) p(\sigma^2) p(\mathbf{y} | \boldsymbol{\beta}, \boldsymbol{\alpha}, \sigma^2, \mathbf{X}) d\boldsymbol{\beta} d\boldsymbol{\alpha} d\sigma^2 \\
&= \pi^{-N/2} |\boldsymbol{\Sigma}|^{-1/2} |\mathbf{X}_\gamma^T \mathbf{X}_\gamma + \boldsymbol{\Sigma}^{-1}|^{-1/2} \frac{\Gamma((\nu + N)/2)}{\Gamma(\nu/2)} (\nu s^2)^{\nu/2} (\nu s^2 + S_{yy})^{-(\nu+N)/2},
\end{aligned}$$

where $S_{yy} = \mathbf{y}^T \mathbf{y} - \mathbf{y}^T \mathbf{X}_\gamma (\mathbf{X}_\gamma^T \mathbf{X}_\gamma + \boldsymbol{\Sigma}^{-1})^{-1} \mathbf{X}_\gamma^T \mathbf{y}$ and $\boldsymbol{\Sigma}$ is the prior covariance matrix of $\boldsymbol{\alpha}$ and $\boldsymbol{\beta}_\gamma$.

Step 4

Note that $\boldsymbol{\beta}$ and $\boldsymbol{\alpha}$ are integrated out analytically at this step.

$$\begin{aligned}
p(\sigma^2 | \dots) &\propto p(\sigma^2) \int p(\boldsymbol{\alpha} | \sigma^2) p(\boldsymbol{\beta} | \sigma^2, \boldsymbol{\tau}^2, \boldsymbol{\gamma}, \mathbf{t}) p(\mathbf{y} | \boldsymbol{\beta}, \boldsymbol{\alpha}, \sigma^2, \mathbf{X}) d\boldsymbol{\alpha} d\boldsymbol{\beta} \\
&\propto (\sigma^2)^{-(\nu/2+1)} \exp\left(-\frac{1}{2\sigma^2} \nu s^2\right) (\sigma^2)^{-N/2} \exp\left(-\frac{1}{2\sigma^2} S_{yy}\right) \\
&= (\sigma^2)^{-((\nu+N)/2+1)} \exp\left(-\frac{1}{2\sigma^2} (\nu s^2 + S_{yy})\right) \\
p(\sigma^2 | \dots) &= \text{Inv-}\chi^2\left(\sigma^2 | \nu + N, \frac{\nu s^2 + S_{yy}}{\nu + N}\right).
\end{aligned}$$

Step 5

Let $\mathbf{c} = [\boldsymbol{\alpha}^T \boldsymbol{\beta}_\gamma^T]^T$ and $\boldsymbol{\Sigma}$ be the corresponding covariance matrix of the prior.

$$\begin{aligned} p(\mathbf{c}|\dots) &\propto p(\mathbf{c}|\sigma^2, \boldsymbol{\tau}^2, \boldsymbol{\gamma}, \mathbf{t})p(\mathbf{y}|\mathbf{c}, \sigma^2, \mathbf{X}) \\ &= |2\pi\boldsymbol{\Sigma}|^{-1/2} \exp\left(-\frac{1}{2\sigma^2}\mathbf{c}^T\boldsymbol{\Sigma}^{-1}\mathbf{c}\right)(2\pi\sigma^2)^{-N/2} \exp\left(-\frac{1}{2\sigma^2}(\mathbf{y} - \mathbf{X}_c\mathbf{c})^T(\mathbf{y} - \mathbf{X}_c\mathbf{c})\right) \\ &\propto \exp\left(-\frac{1}{2\sigma^2}[(\mathbf{c} - (\mathbf{X}_c^T\mathbf{X}_c + \boldsymbol{\Sigma}^{-1})^{-1}\mathbf{X}_c^T\mathbf{y})^T(\mathbf{X}_c^T\mathbf{X}_c + \boldsymbol{\Sigma}^{-1})(\mathbf{c} - (\mathbf{X}_c^T\mathbf{X}_c + \boldsymbol{\Sigma}^{-1})^{-1}\mathbf{X}_c^T\mathbf{y})]\right) \\ p(\mathbf{c}|\dots) &= \text{N}(\mathbf{c} | (\mathbf{X}_c^T\mathbf{X}_c + \boldsymbol{\Sigma}^{-1})^{-1}\mathbf{X}_c^T\mathbf{y}, \sigma^2(\mathbf{X}_c^T\mathbf{X}_c + \boldsymbol{\Sigma}^{-1})^{-1}). \end{aligned}$$