Ancestry Inference

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Section 1

Introduction

Ancestor inference

- PSD model
- PCA-based methods
- model-based approaches

Algorithms

- STRUCTURE (MCMC) (Pritchard, Stephens, and Donnelly 2000)
- FRAPPE (EM) (Tang et al. 2005)
- ADMIXTURE (SQP) (Alexander, Novembre, and Lange 2009)
- fastSTRUCTURE (VI) (Raj, Stephens, and Pritchard 2014)
- TeraStructure (SVI) (Gopalan et al. 2016)

Data

- TGP (Abecasis et al. 2012)
- HGDP (Cann et al. 2002; Cavalli-Sforza 2005; Lu et al. 2011; Li et al. 2008)

Section 2

Models and Methods

PSD model

- observed variable: genotype matrix G
- latent variable: matrix Z of the true origin of genes
- parameters: population scale matrix P, gene scale matrix F
- hyperparameter: population number K

EM algorithm

- E-step: compute the expectation a_{ijk} and b_{ijk}
- M-step: compute the maximization and update the parameters p_{ik} and f_{kj}
- convergence criterion: the log-likelihood of incomplete data $\mathcal{L}(G|P, F)$ converges

SQP algorithm

- update parameters: update P and F block by block alternately
- convergence criterion: the log-likelihood of incomplete data $\mathcal{L}(G|P, F)$ converges

VI algorithm

- update parameters: update variational parameters $\tilde{z}_{ij}^{a}$, $\tilde{p}_i$, $\tilde{f}_{kj}^{1}$, $\tilde{f}_{kj}^{2}$
- convergence criterion: the ELBO converges

SVI algorithm

- sample: sample a SNP
- update parameters: iteratively update local parameter F_j at the SNP until it converges, then update global parameter P
- convergence criterion: the log-likelihood at the validation set converges

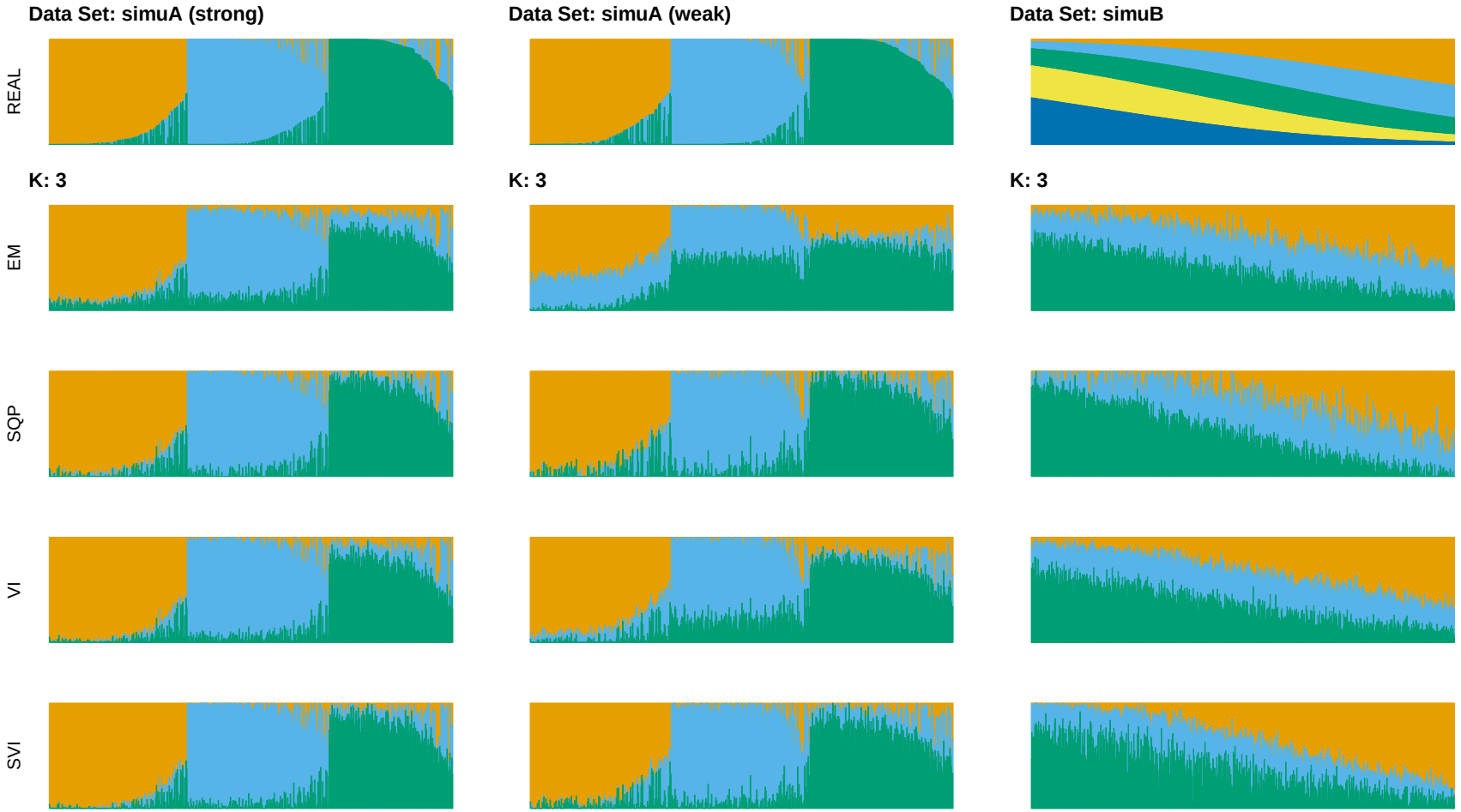
Relationships with other models

- Poisson NMF model (Carbonetto et al. 2021)
- multinomial topic model
- LDA model

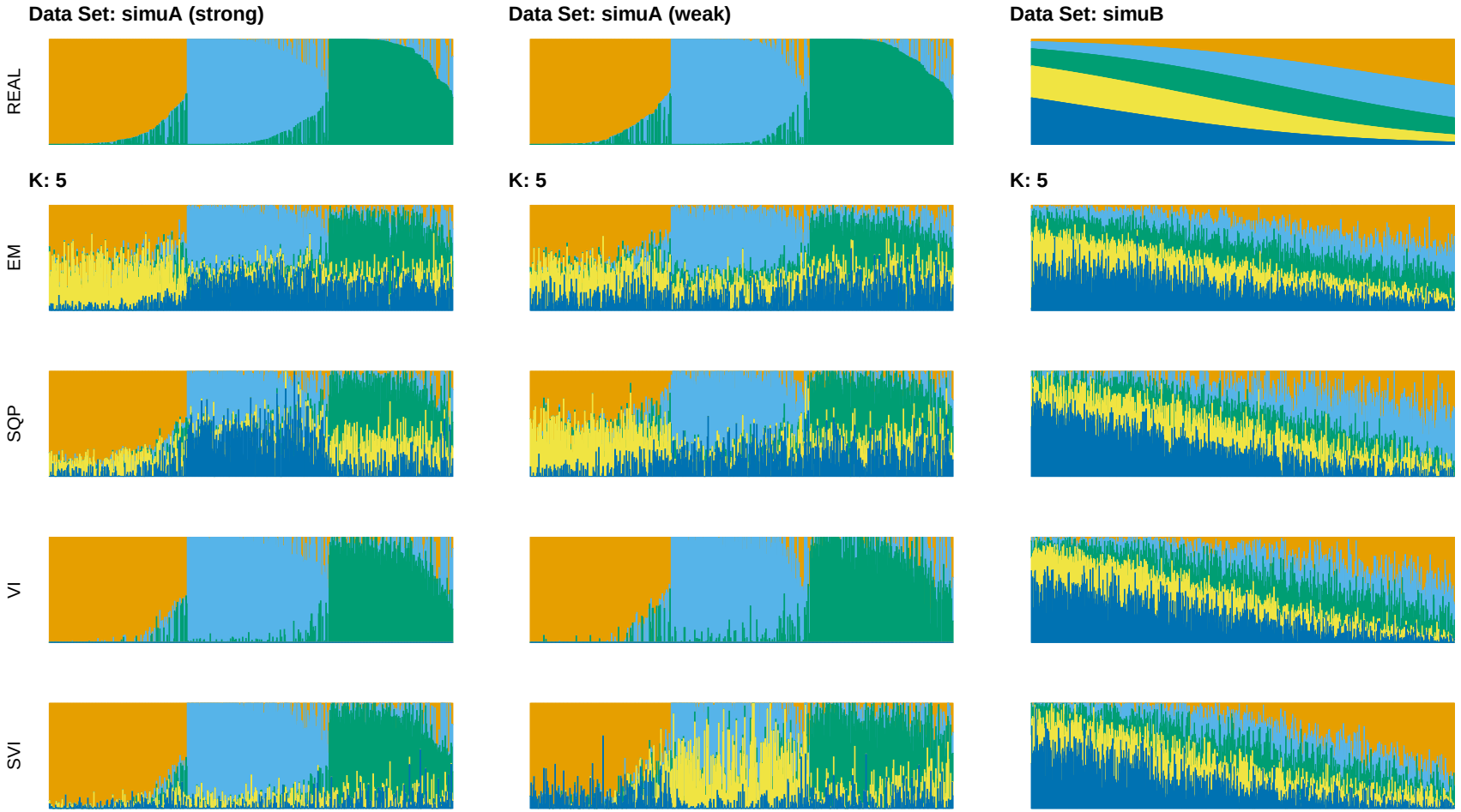
Section 3

Applications

Simulated data set

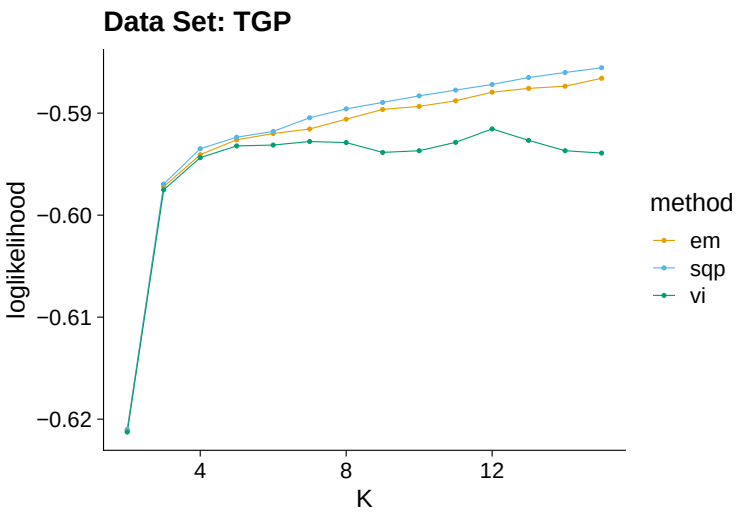


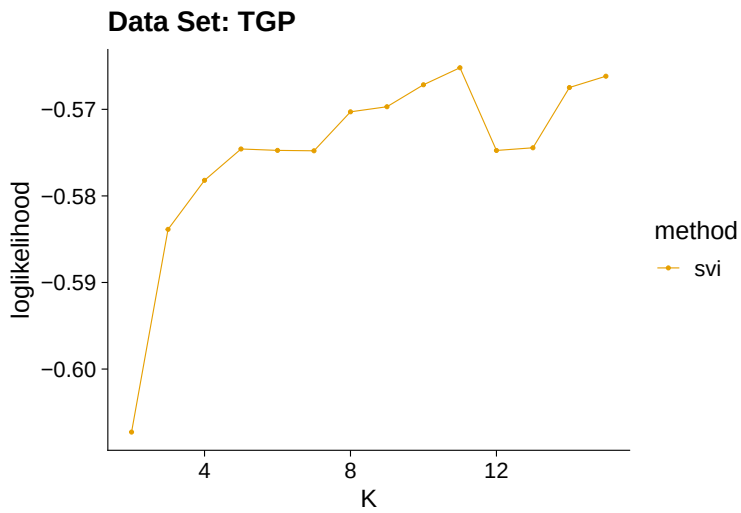
Simulated data set

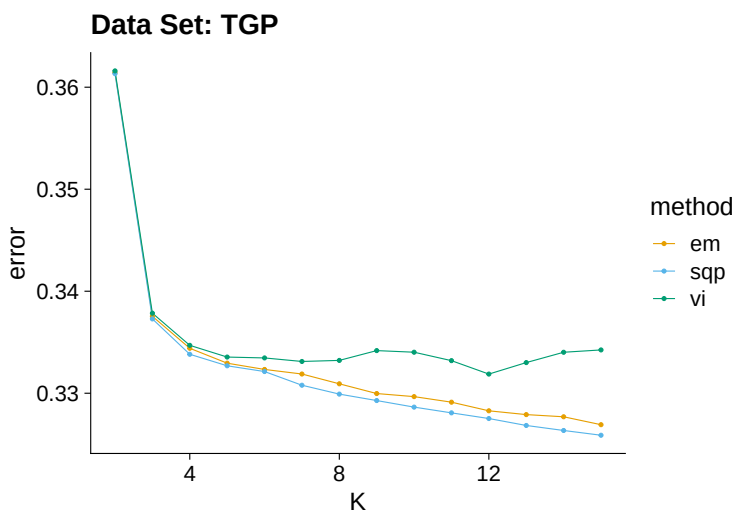


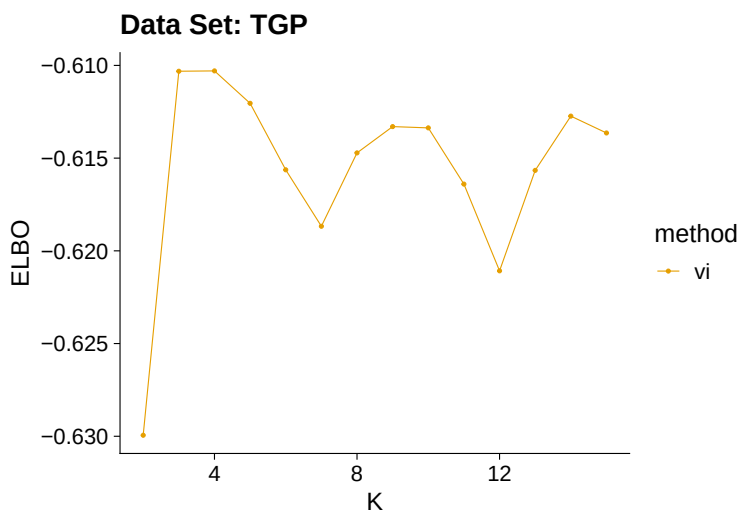
TGP data set

- choose K





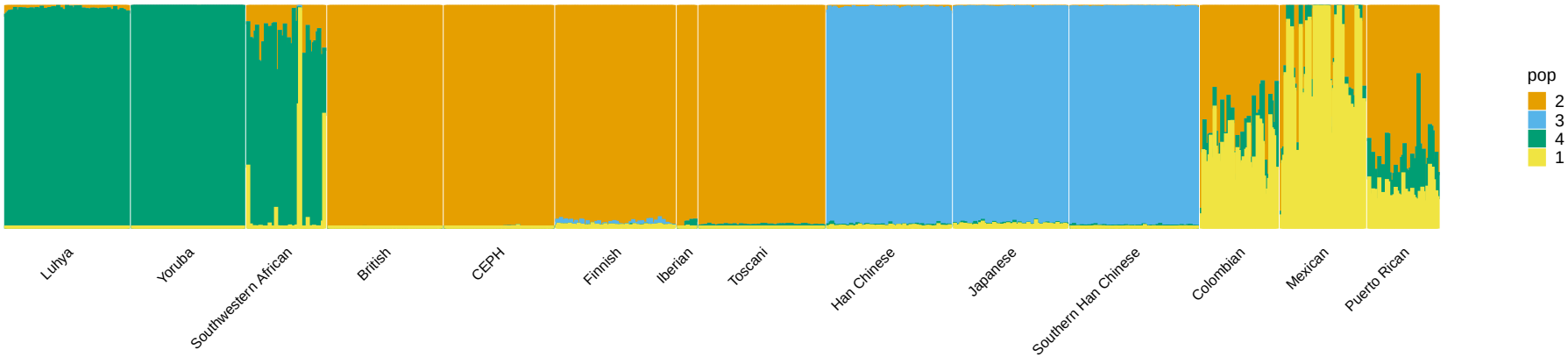




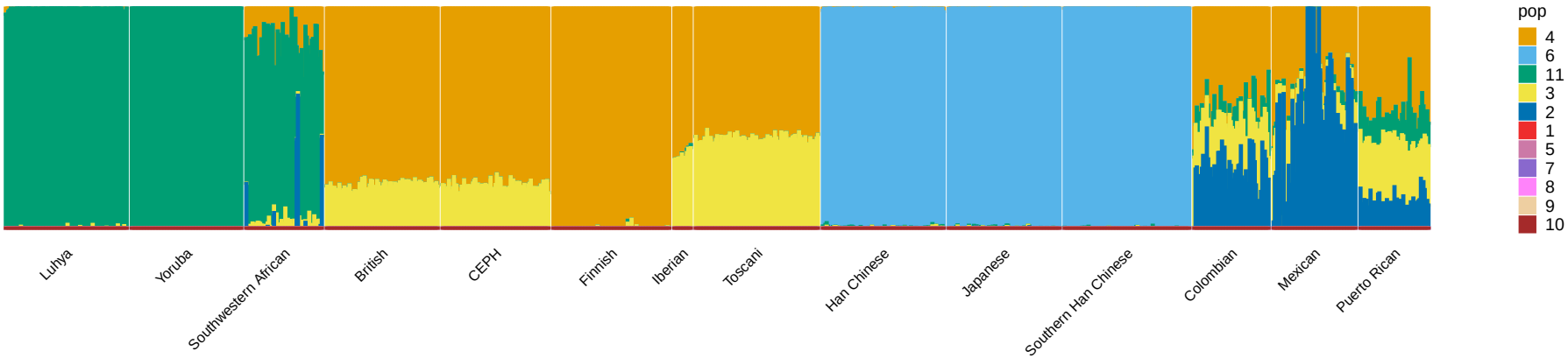
TGP data set

- structure plot

Data Set: TGP (full) | Method: SVI (1e+6 interations) | K: 4

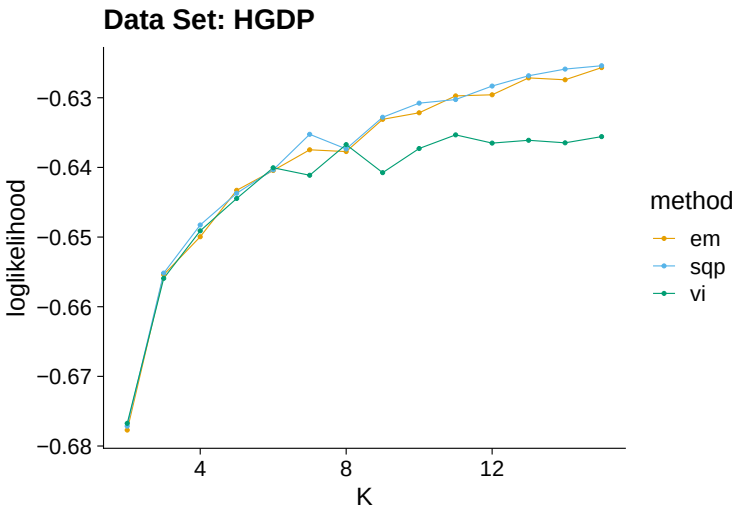


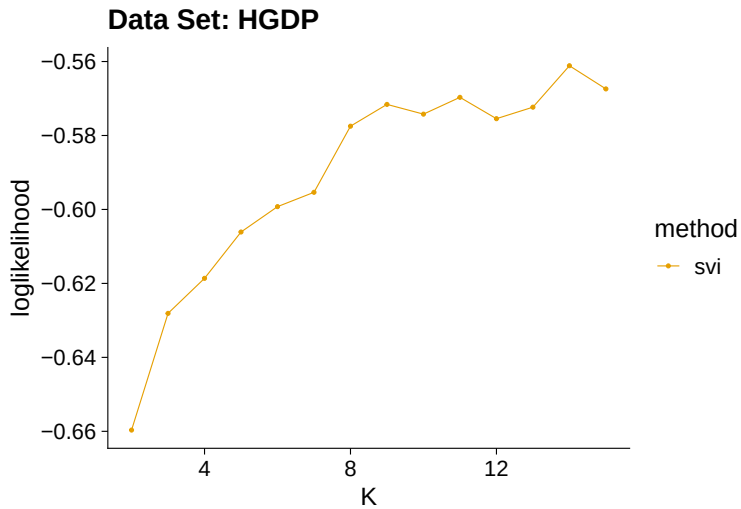
Data Set: TGP (full) | Method: SVI (1e+6 interations) | K: 11

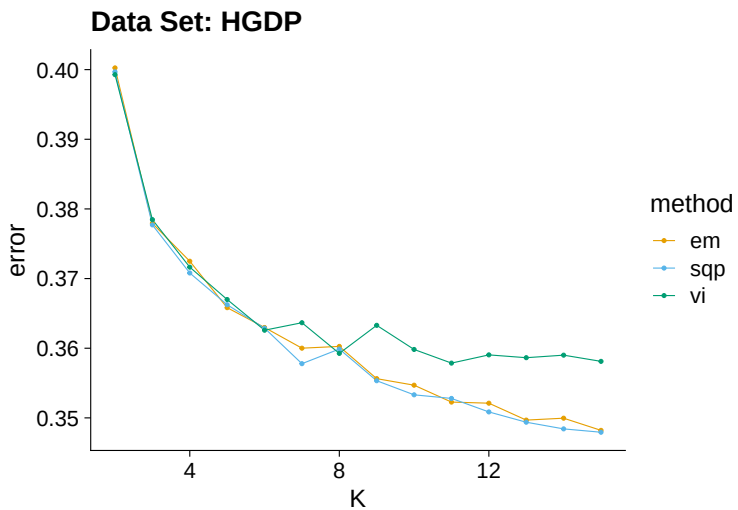


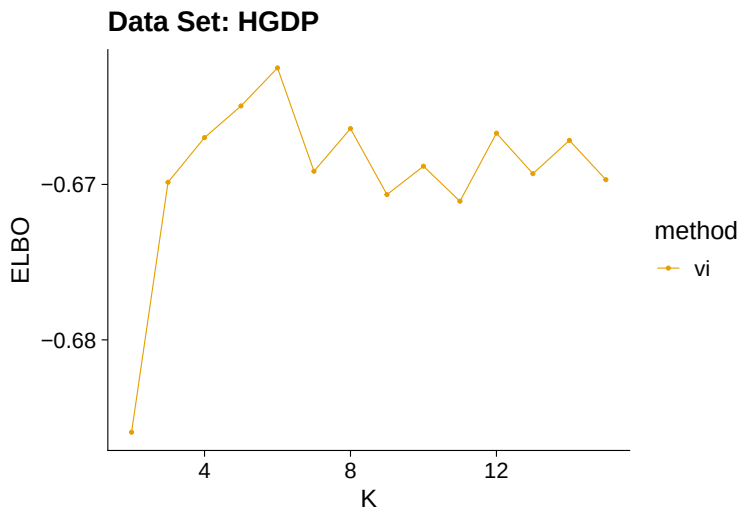
HGDP data set

- choose K





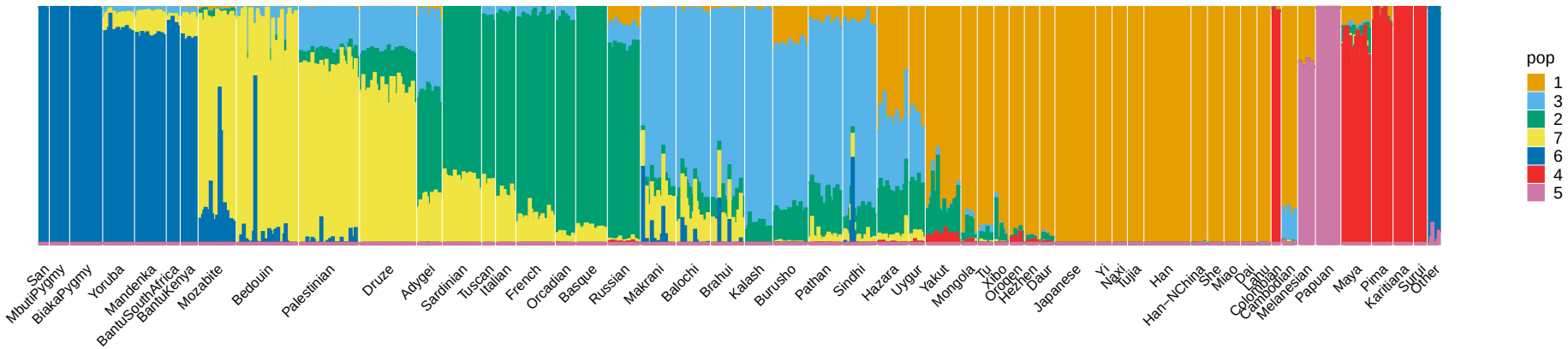




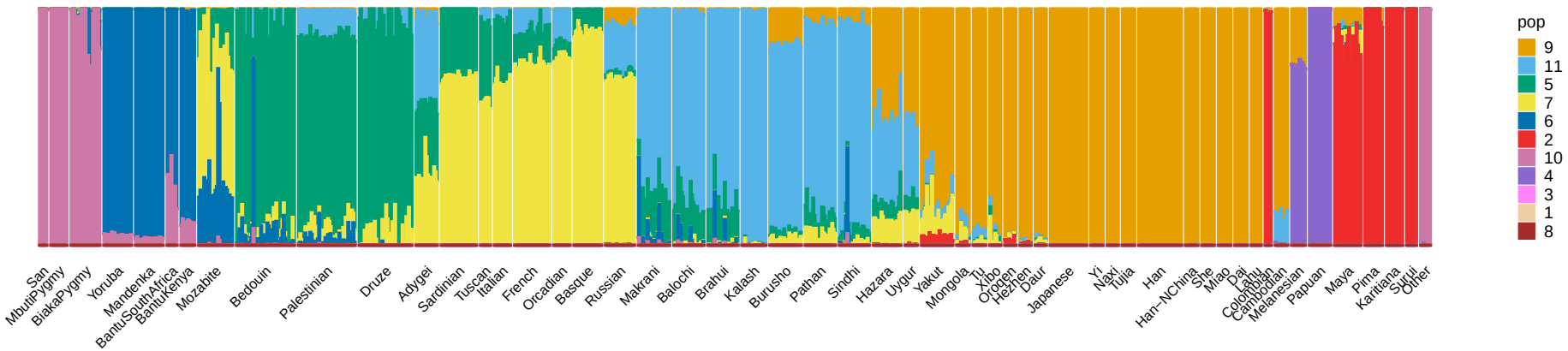
HGDP data set

- structure plot

Data Set: HGDP (full) | Method: SVI (1e+6 interations) | K: 7



Data Set: HGDP (full) | Method: SVI (1e+6 interations) | K: 11

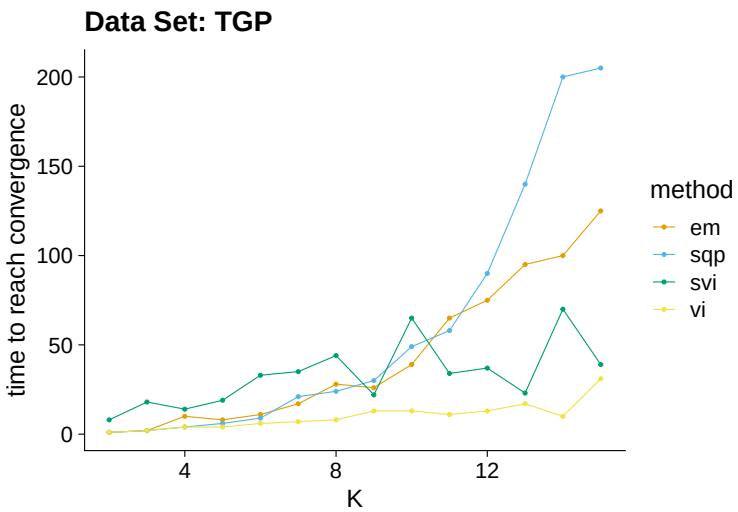


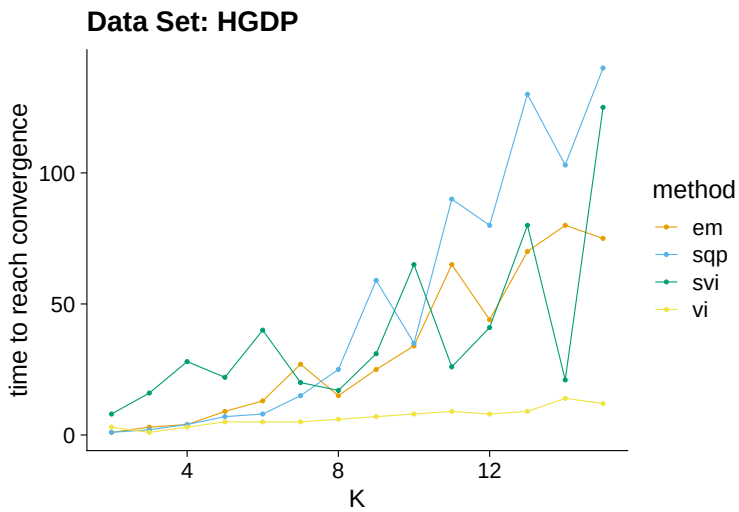
Section 4

Discussion

Algorithm evaluation

- convergence accuracy
- convergence efficiency





- algorithm selection criteria

Section 5

Literature Cited

Literature Cited I

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