# Ancestry Inference

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2022-10-04

Section 1

# Introduction

# Ancestor inference

- inheritable diseases, conservation genetics, the ancestry and migration patterns of natural populations
- 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)
- sNMF (SNMF) (Frichot et al. 2014)
- 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; 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

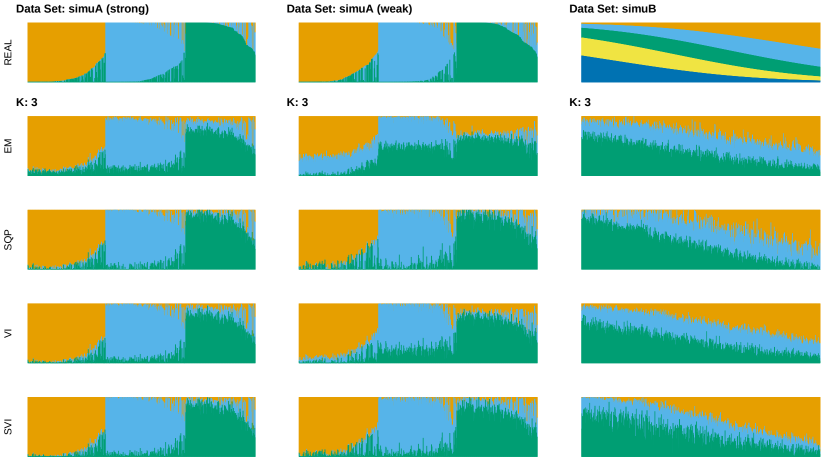
# Relationships with other models

- Poisson NMF model, multinomial topic model (Carbonetto et al. 2021)
- PLSA model (Hofmann 2001)
- LDA model (Blei, Ng, and Jordan 2003)

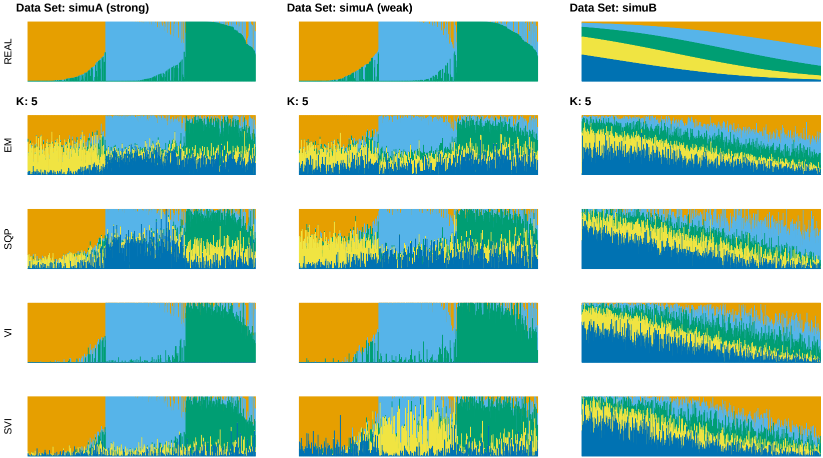
Section 3

# Applications

# Simulated data set

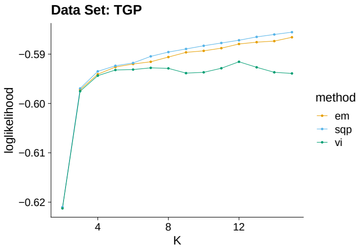


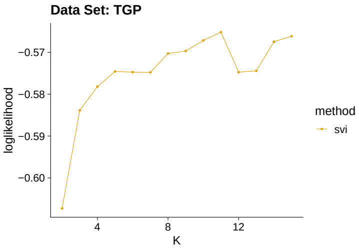
# Simulated data set

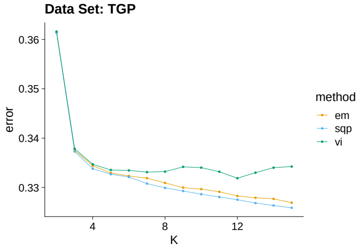


# TGP data set

- choose $K$


Data Set: TGP


Data Set: TGP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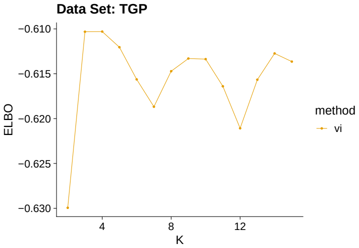
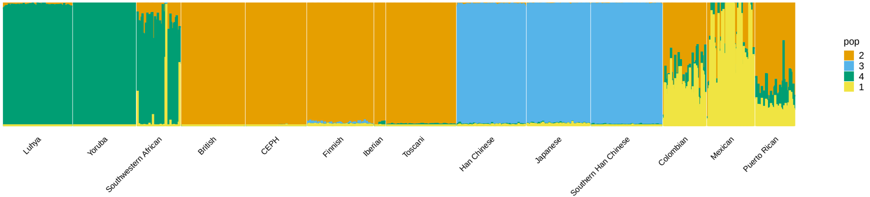

Data Set: TGP

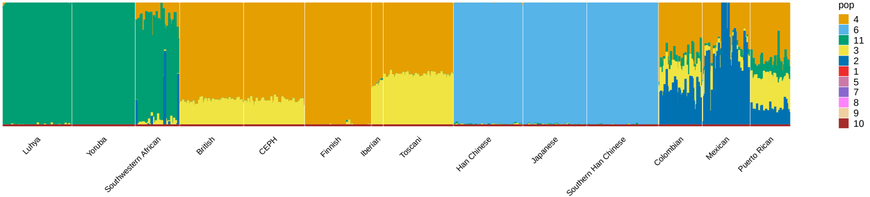
# TGP data set

- structure plot

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

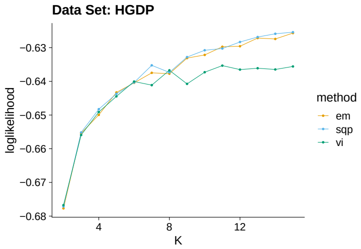


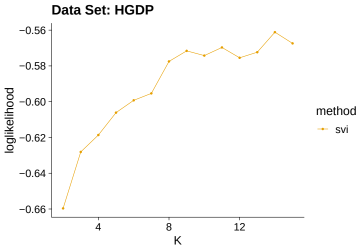
**Data Set: TGP (full) | Method: SVI (1e+6 interations) | K: 11**

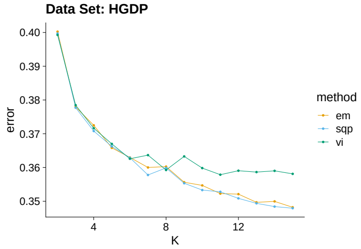


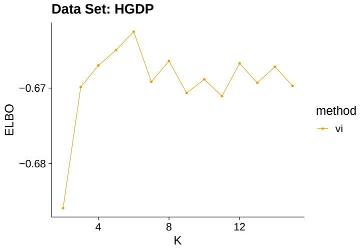
# HGDP data set

- choose $K$





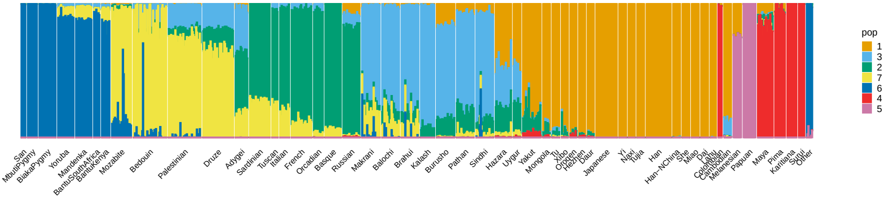




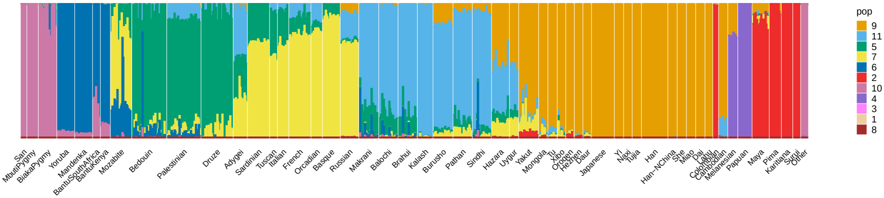
# HGDP data set

- structure plot

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



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

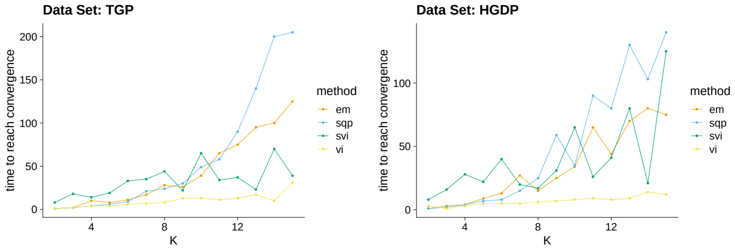


Section 4

# Discussion

# Algorithm evaluation

- convergence accuracy
- convergence efficiency



- algorithm selection criteria

Section 5

# Literature Cited

# Literature Cited I

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