Coalescent HMMs
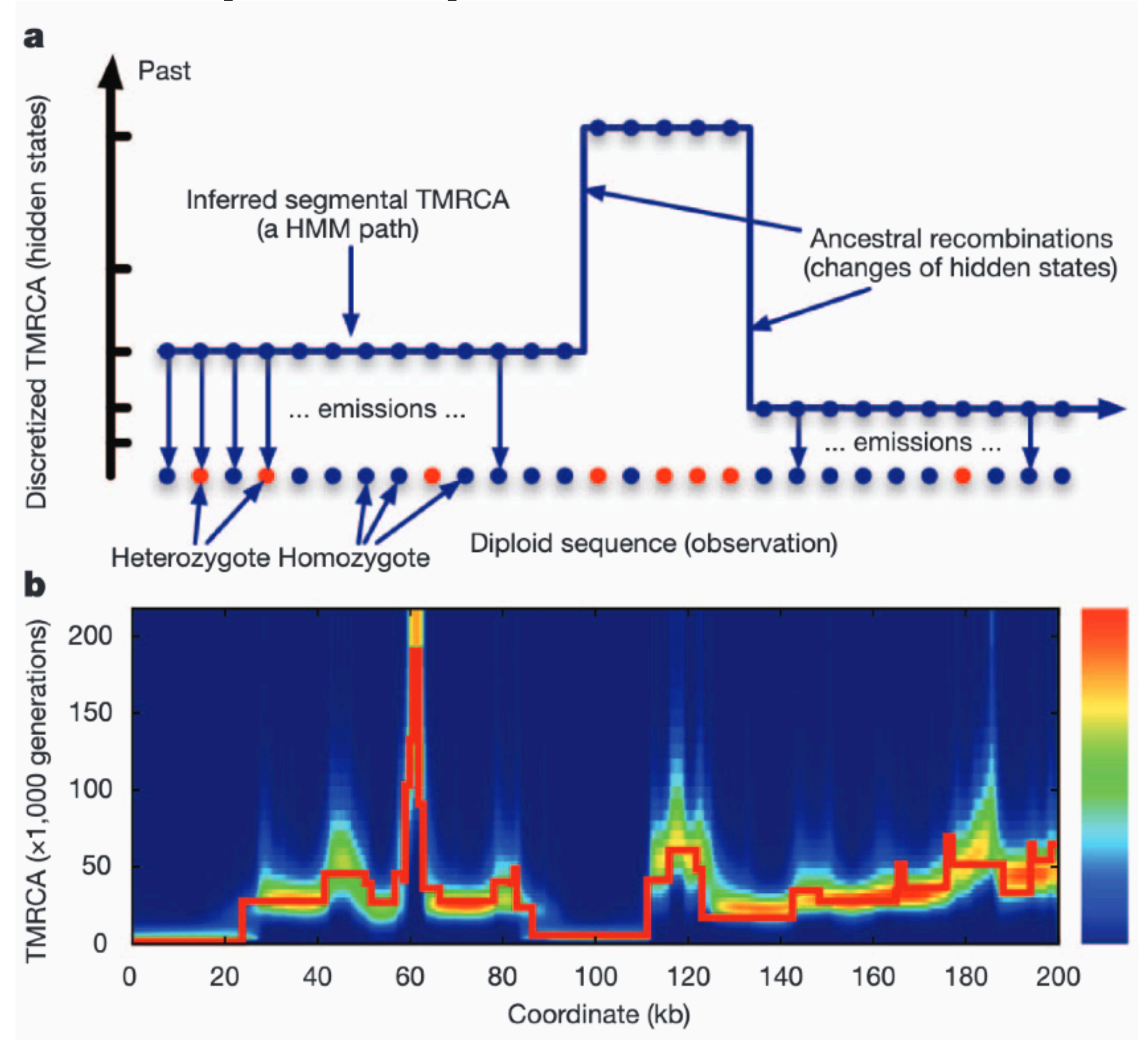## Pairwise sequential Markov coalescent (PSMC)

Hidden state: T_{MRCA}

Emission: heterozygosity $00100110...$

Transitions:

$$p(t|s) = \begin{cases} e^{-\rho s}, & s = t \\ (1-e^{-\rho s})q(t|s), & s \neq t \end{cases}$$

$\overset{4N r}{\smile}$

complicated integral of history $N(t)$

a Past

Discretized TMRCA (hidden states)

Inferred segmental TMRCA (a HMM path)

Ancestral recombinations (changes of hidden states)

... emissions ...

... emissions ...

Heterozygote Homozygote

Diploid sequence (observation)

b

TMRCA (×1,000 generations)

200

150

100

50

0

0   20   40   60   80   100   120   140   160   180   200

Coordinate (kb)

ARTICLE

A global reference for human genetic variation

The 1000 Genomes Project Consortium*

Averaging indv. estimates
to get pop estimate

$N(t)$