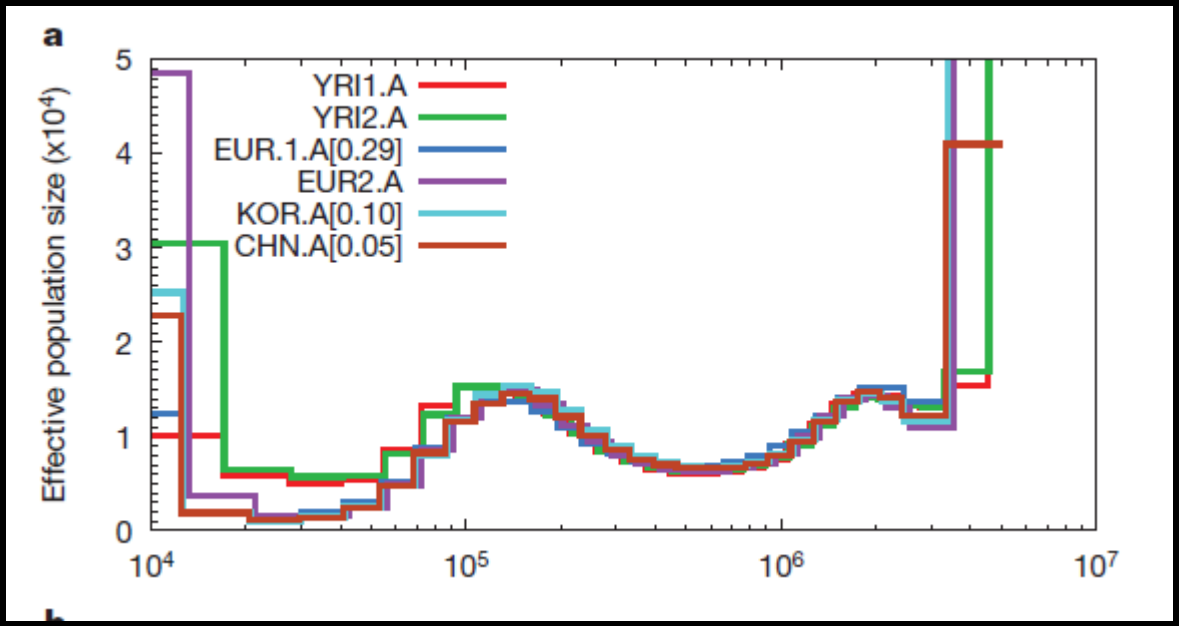
THE PSMC
(Pairwise Sequentially Markovian Coalescent)

LETTER

doi:10.1038/nature10231

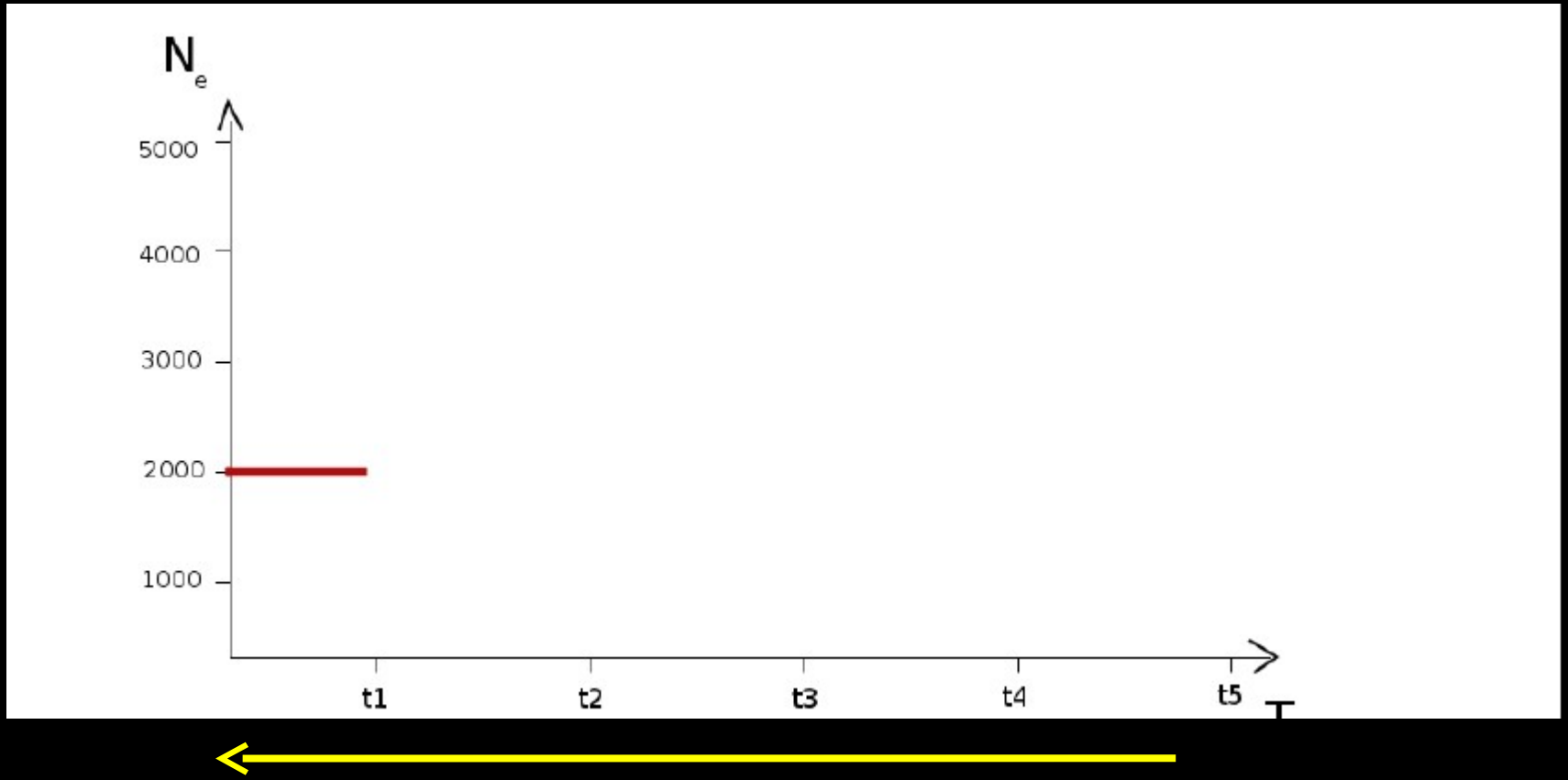
Inference of human population history from individual whole-genome sequences

Heng Li[1,2] & Richard Durbin[1]



THE PSMC

Population size

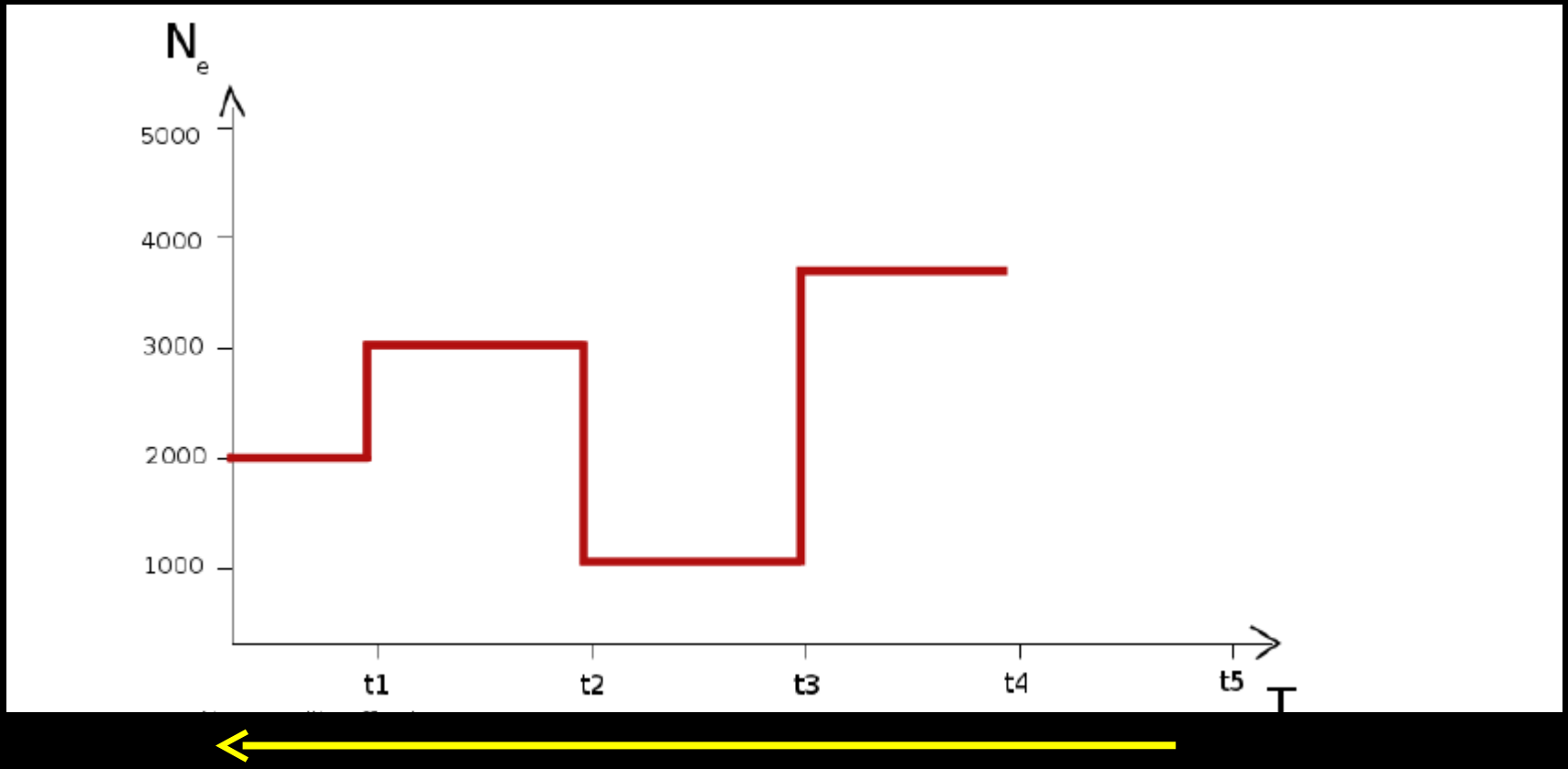


Present

Past

THE PSMC

Population size

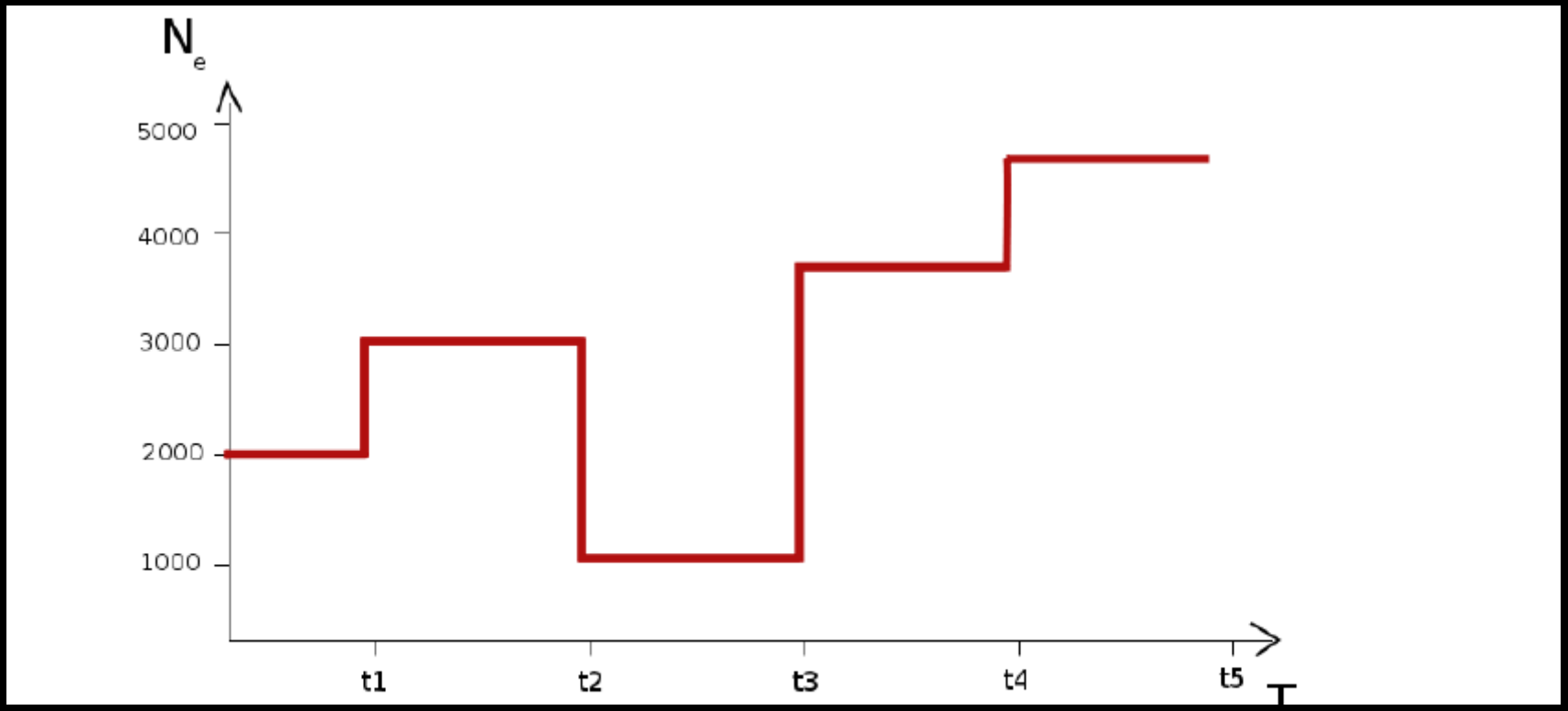


Present

Past

THE PSMC

Population size

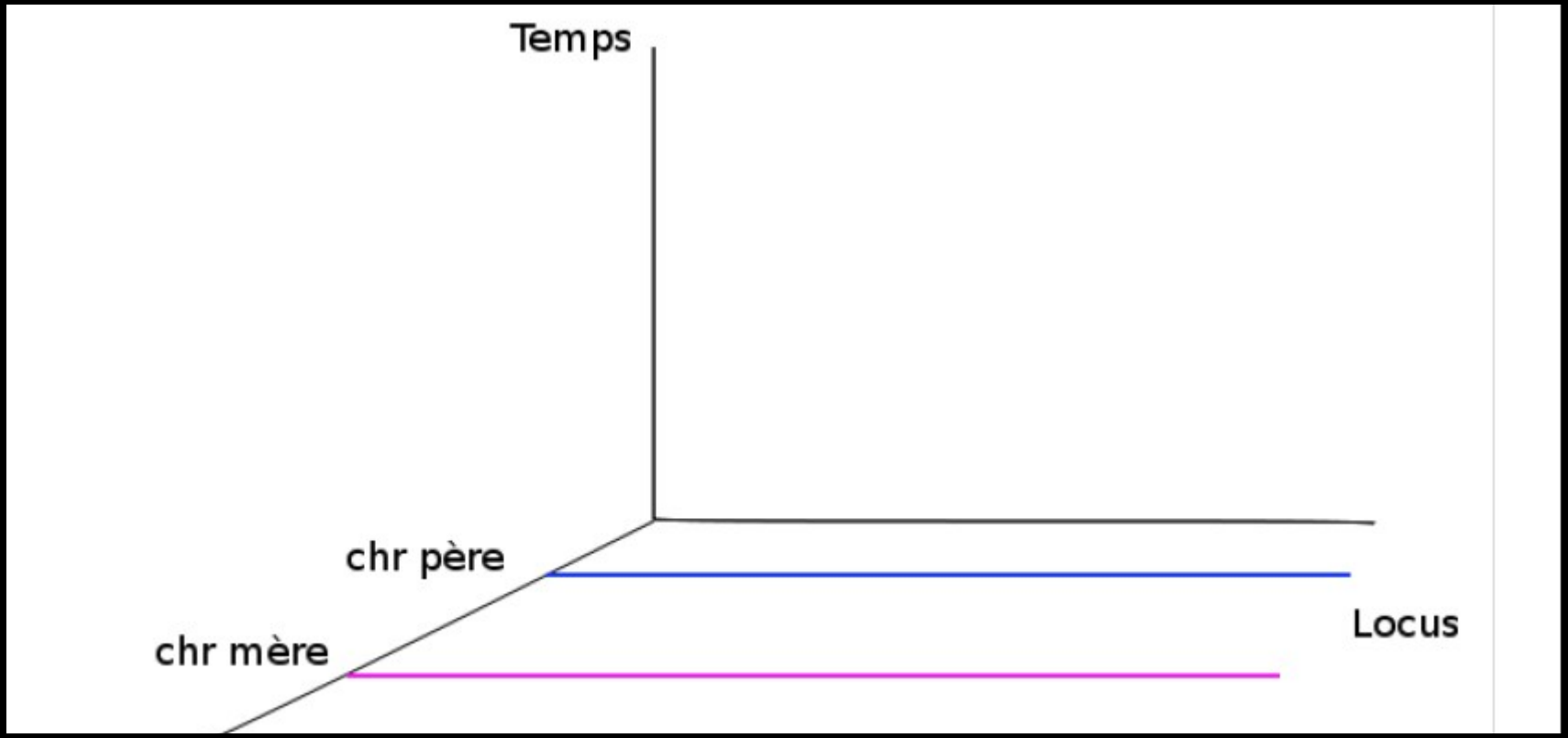


Present

Past

THE PSMC

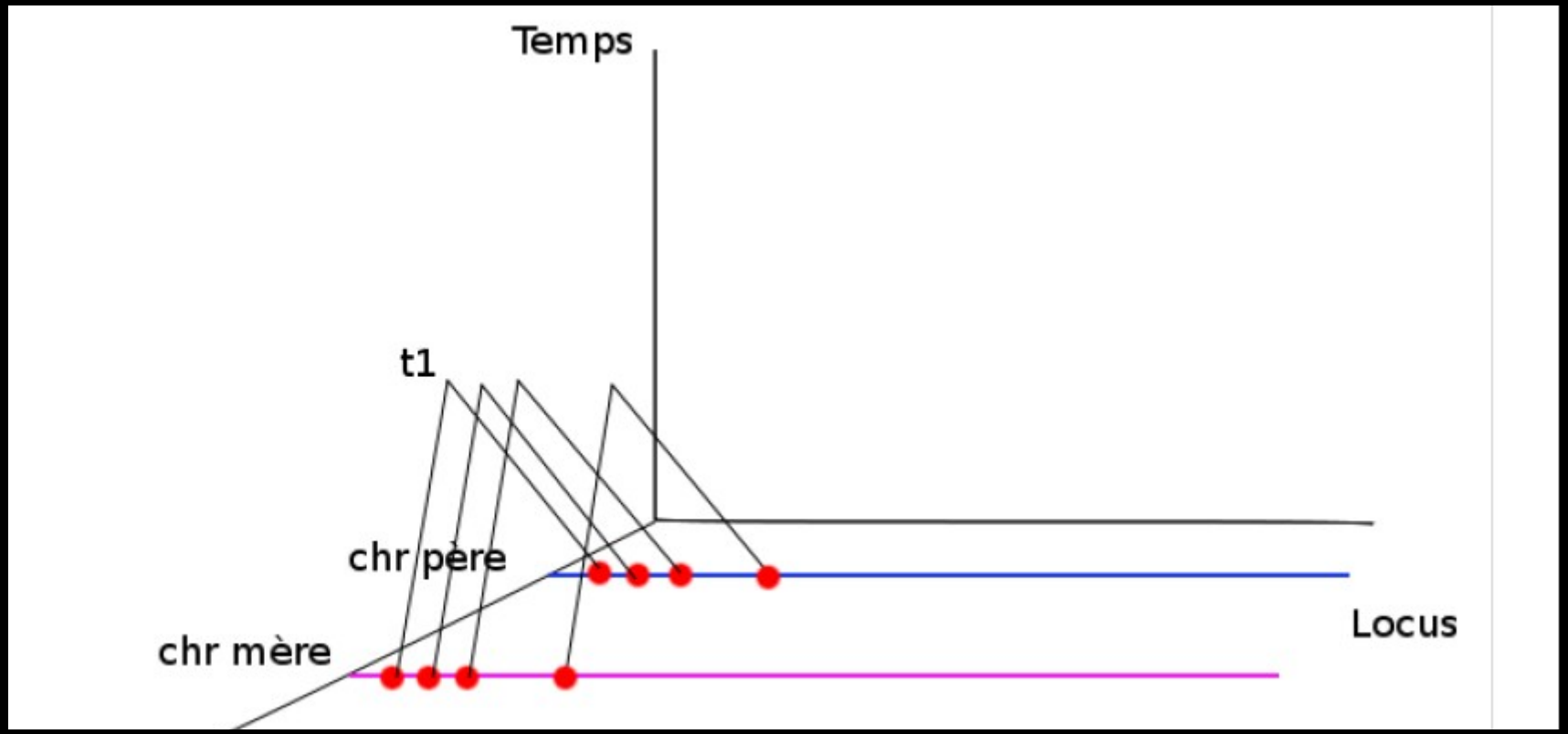
Time



Parental chromosomes

THE PSMC

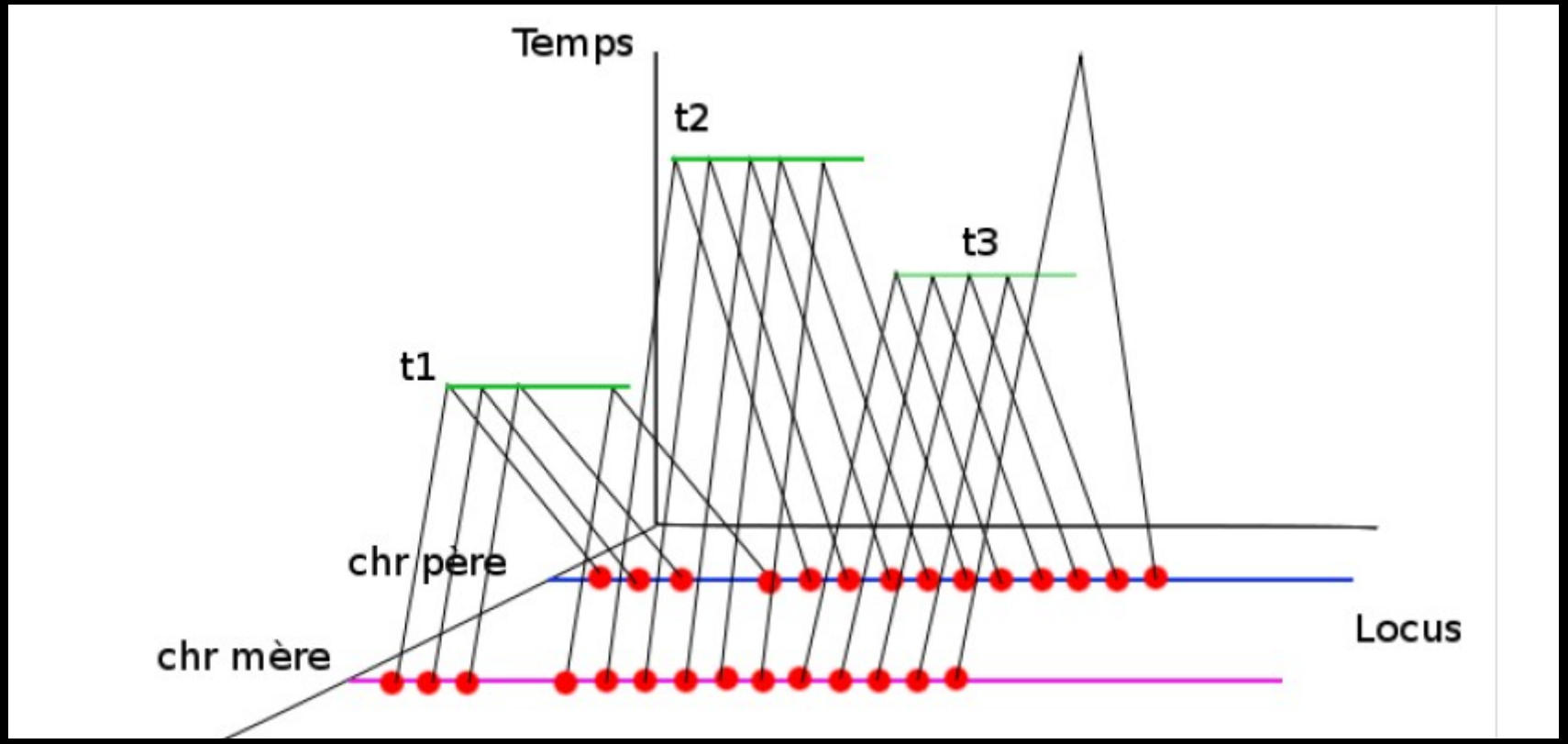
Time



Parental chromosomes

THE PSMC

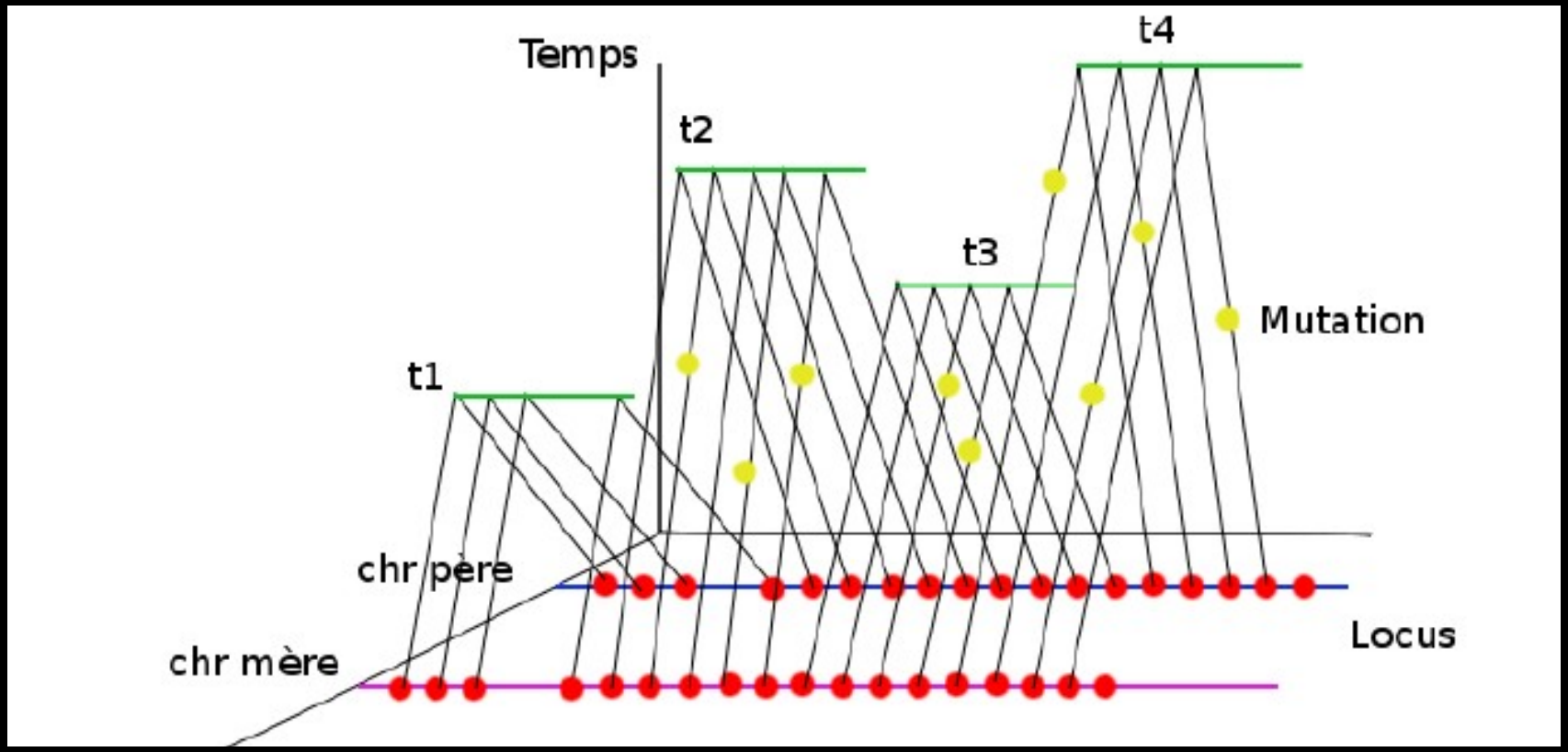
Time



Parental chromosomes

THE PSMC

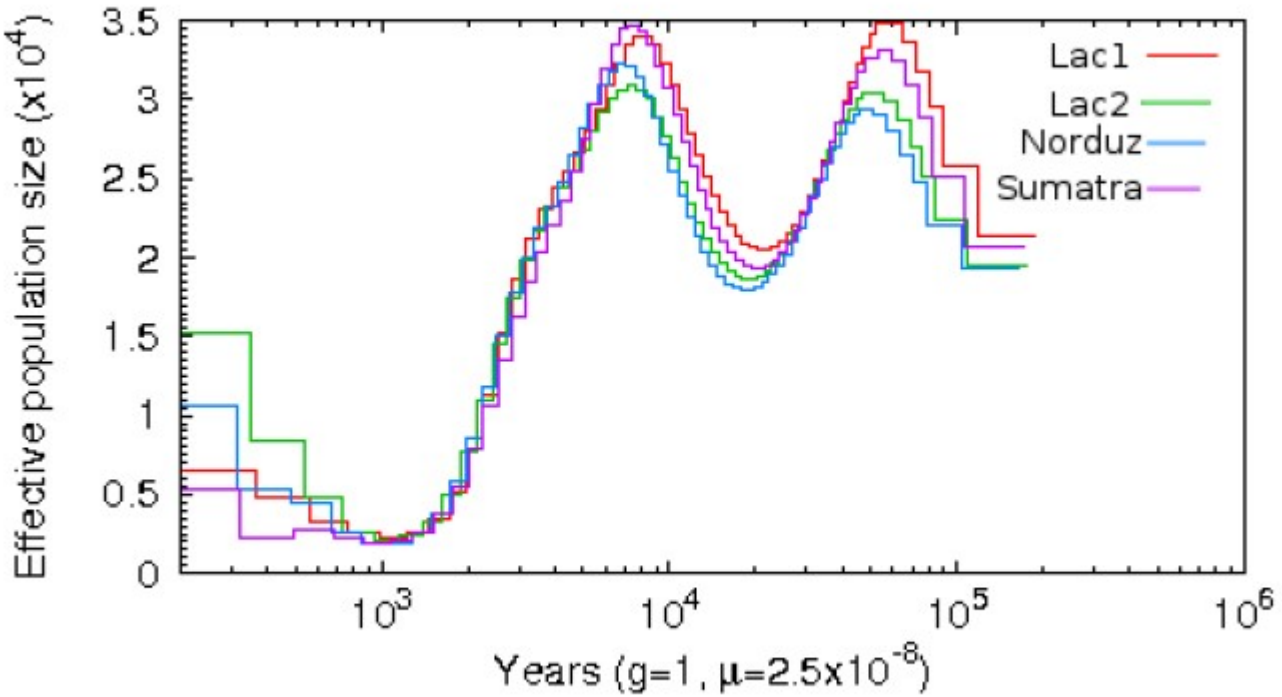
Time



Parental chromosomes

THE PSMC
(applied to three sheep breeds)

Time



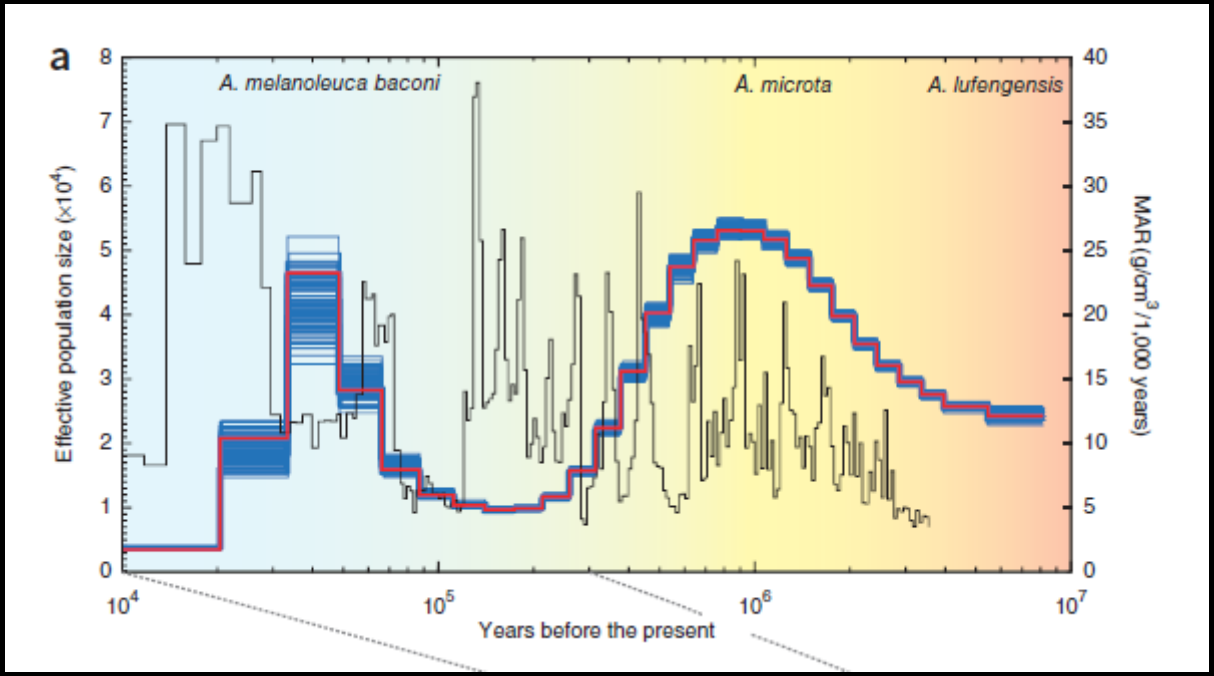
S. Boitard – W. Rodriguez – O. Mazet

THE PSMC

nature genetics

Whole-genome sequencing of giant pandas provides insights into demographic history and local adaptation

Shancen Zhao[1,2,10], Pingping Zheng[1,3,10], Shanshan Dong[2,10], Xiangjiang Zhan[1,10], Qi Wu[1,10], Xiaosen Guo[2], Yibo Hu[1], Weiming He[2], Shanning Zhang[4], Wei Fan[2], Lifeng Zhu[1], Dong Li[2], Xuemei Zhang[2], Quan Chen[2], Hemin Zhang[5], Zhihe Zhang[6], Xuelin Jin[7], Jinguo Zhang[8], Huanming Yang[2], Jian Wang[2], Jun Wang[2,9] & Fuwen Wei[1]



THE PSMC

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PLOS | GENETICS

Demographic Divergence History of Pied Flycatcher and Collared Flycatcher Inferred from Whole-Genome Re-sequencing Data

Krystyna Nadachowska-Brzyska*, Reto Burri, Pall I. Olason, Takeshi Kawakami, Linnéa Smeds, Hans Ellegren

Department of Evolutionary Biology, Evolutionary Biology Centre, Uppsala University, Uppsala, Sweden

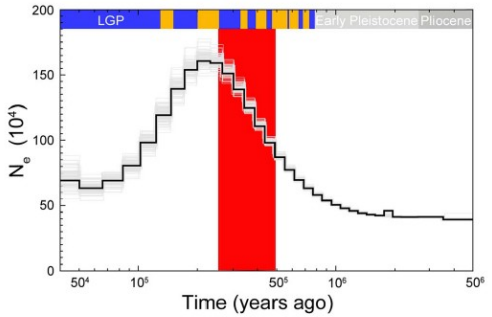
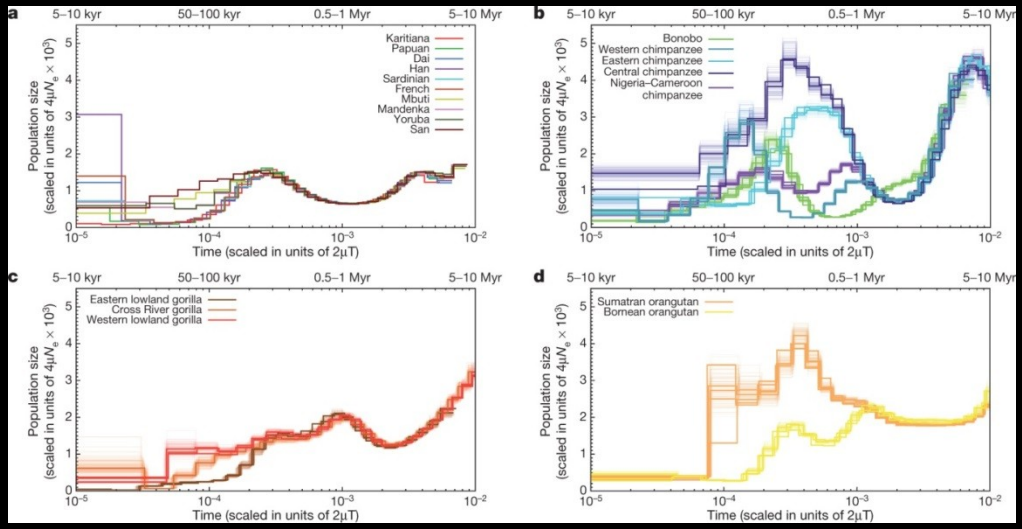


Figure 6. PSMC estimate of the effective population size change over time for collared flycatcher. The black curve is the PSMC estimate for the original data and the grey curves indicate PSMC estimates for 100 bootstrapped sequences. Glacial and interglacial periods of the Late and Middle Pleistocene are indicated by blue and yellow bars, respectively. The interglacial periods corresponds to Marine Isotope Stages: 5e, 7, 9, 11, 13, 15, and 17. The large red-shaded area corresponds to 50% HPDI of the time of divergence (RMASC model). LGP – last glacial period.
doi:10.1371/journal.pgen.1003942.g006

nature

J Prado-Martinez *et al. 2013*



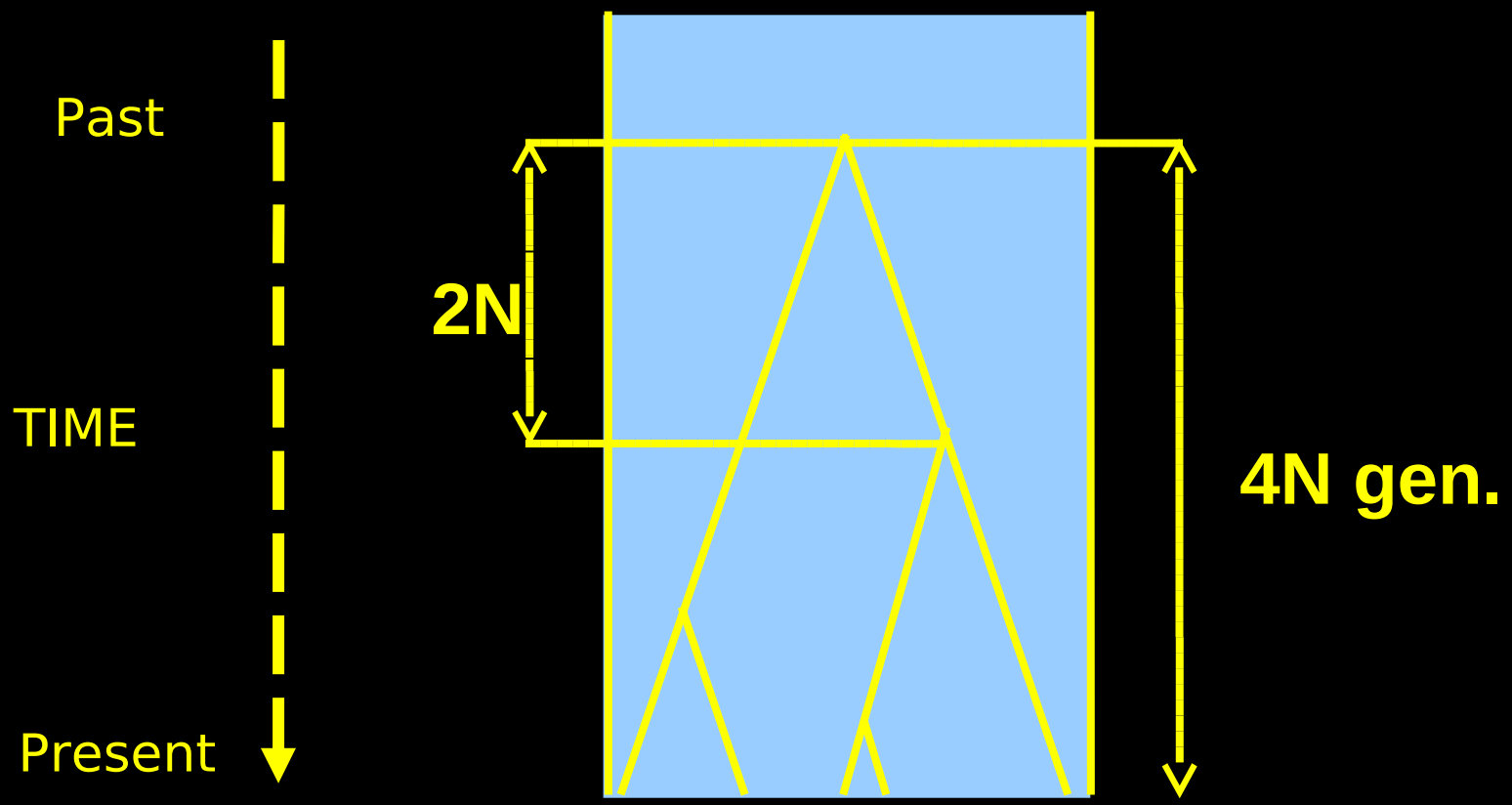
THE PSMC

Now it’s your turn !!!

Run psmc on simulated data

Open the file:
psmc_tutorial_sim.odt

and follow the instructions



N = 1,000 -- gen. = 20 years

4N = 80,000 years

2N = 40,000 years

THE PSMC

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Inference of human population history from individual whole-genome sequences

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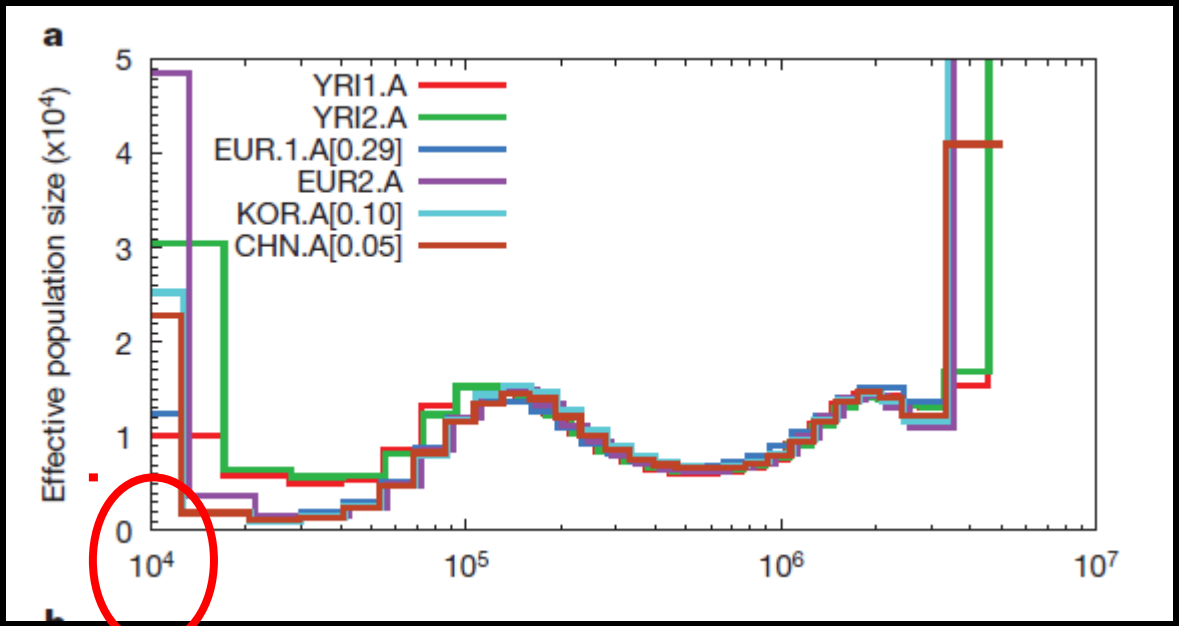


MSMC

Inferring human population size and separation history from multiple genome sequences

Stephan Schiffels & Richard Durbin

nature genetics

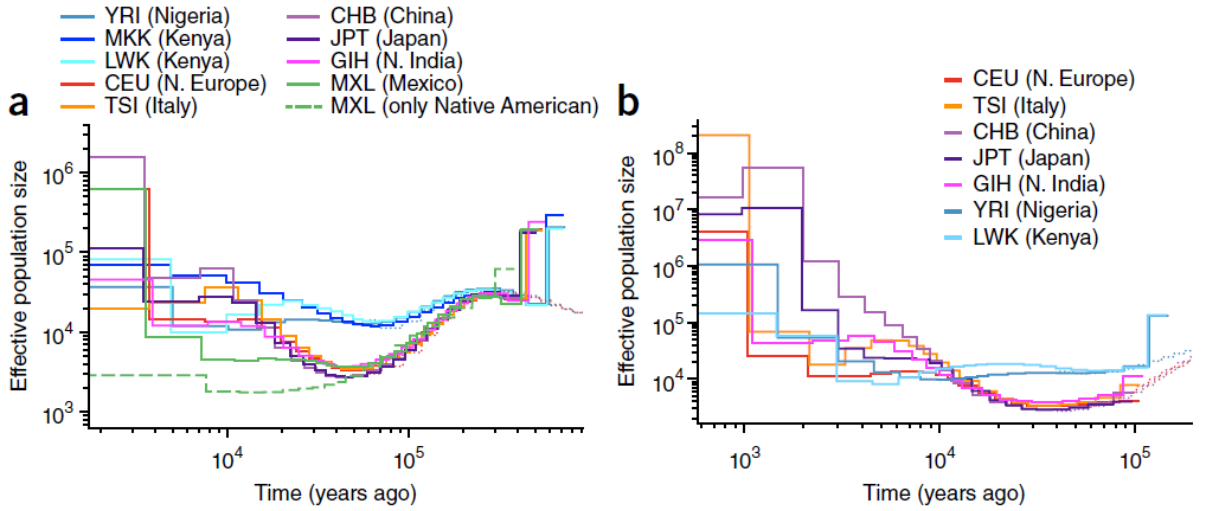
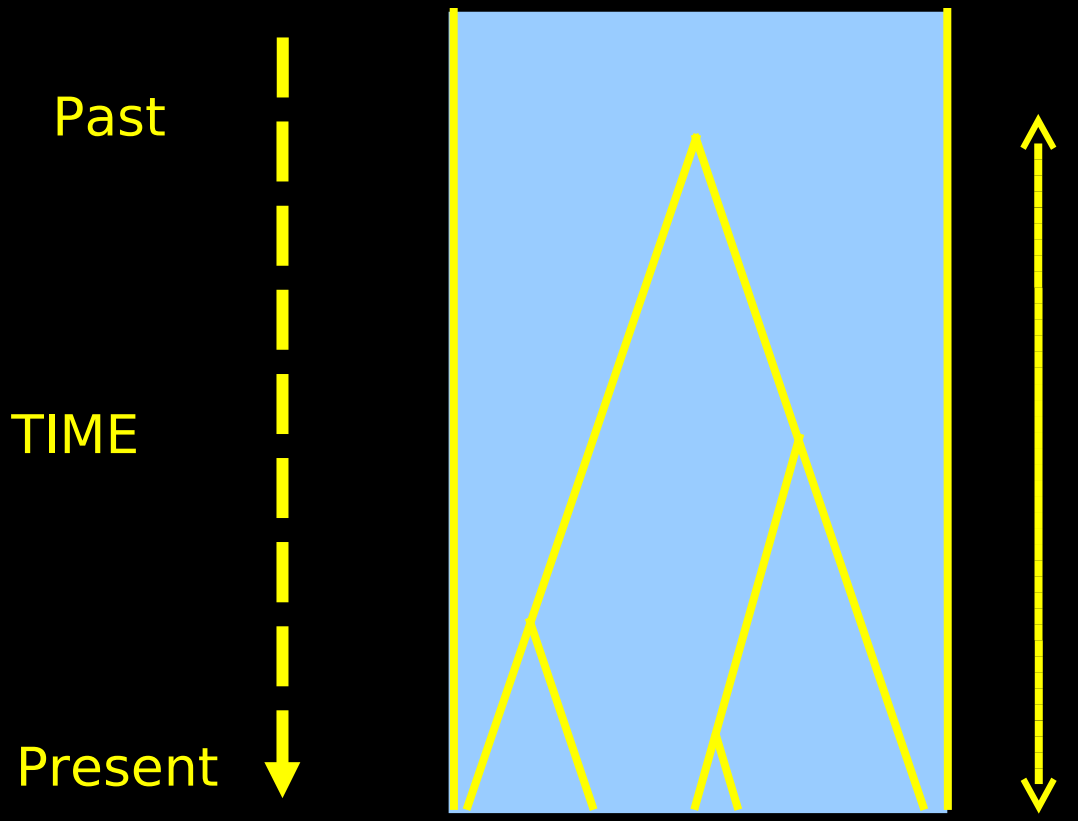


Figure 3 Inference of population size from whole-genome sequences. (**a**) Population size estimates from four haplotypes (two phased individuals) from each of nine populations. The dashed line was generated from a reduced data set of only the Native American components of the MXL genomes. Estimates from two haplotypes for CEU and YRI are shown for comparison as dotted lines. N, Northern. (**b**) Population size estimates from eight haplotypes (four phased individuals) from the same populations as in **a** but excluding MXL and MKK. In contrast to estimates with four haplotypes, estimates are more recent. For comparison, we show the result from four haplotypes for CEU, CHB and YRI as dotted lines.

GENETIC DIVERSITY IN A PANMICTIC POPULATION



Standard Coalescent tree:

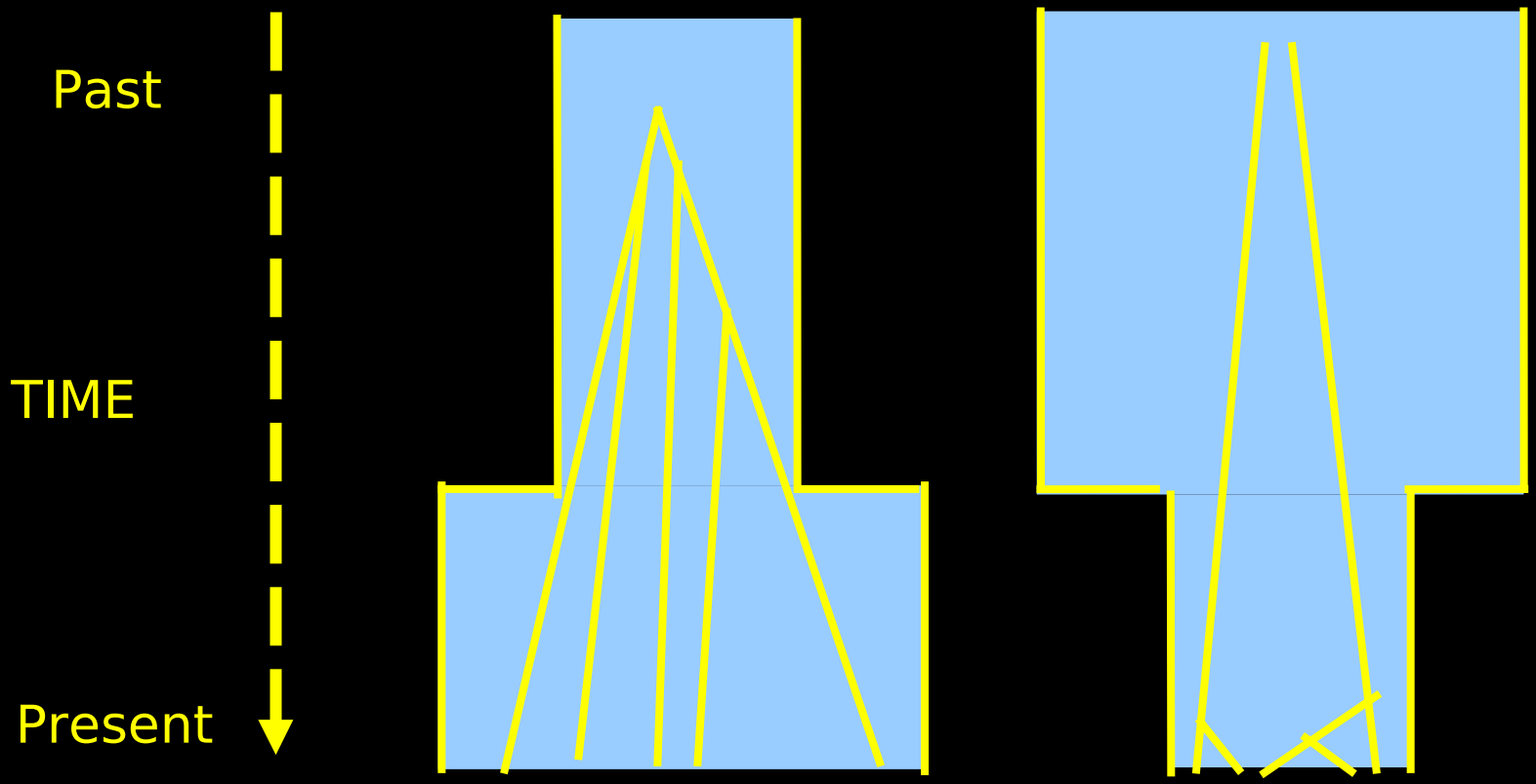
Hight = 4N gen.

STANDARD COALESCENT (STATIONARY POPULATION) PREDICTS

*** SHAPE OF THE GENE TREE / GENEALOGY**

*** PATTERNS OF GENETIC DIVERSITY**

GENETIC DIVERSITY
WHEN POPULATION SIZE CHANGES

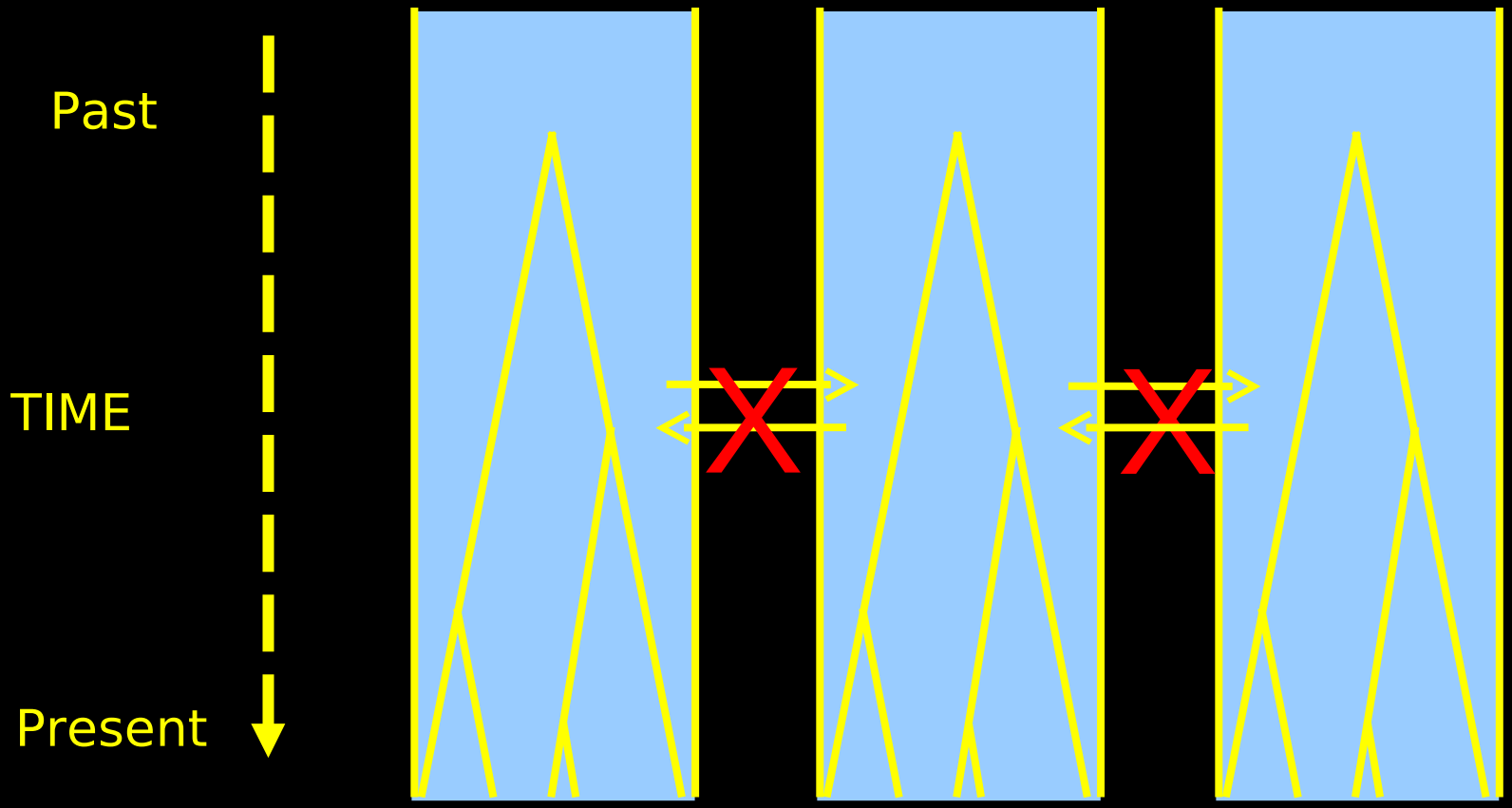


POPULATION SIZE CHANGES / DEMOGRAPHY

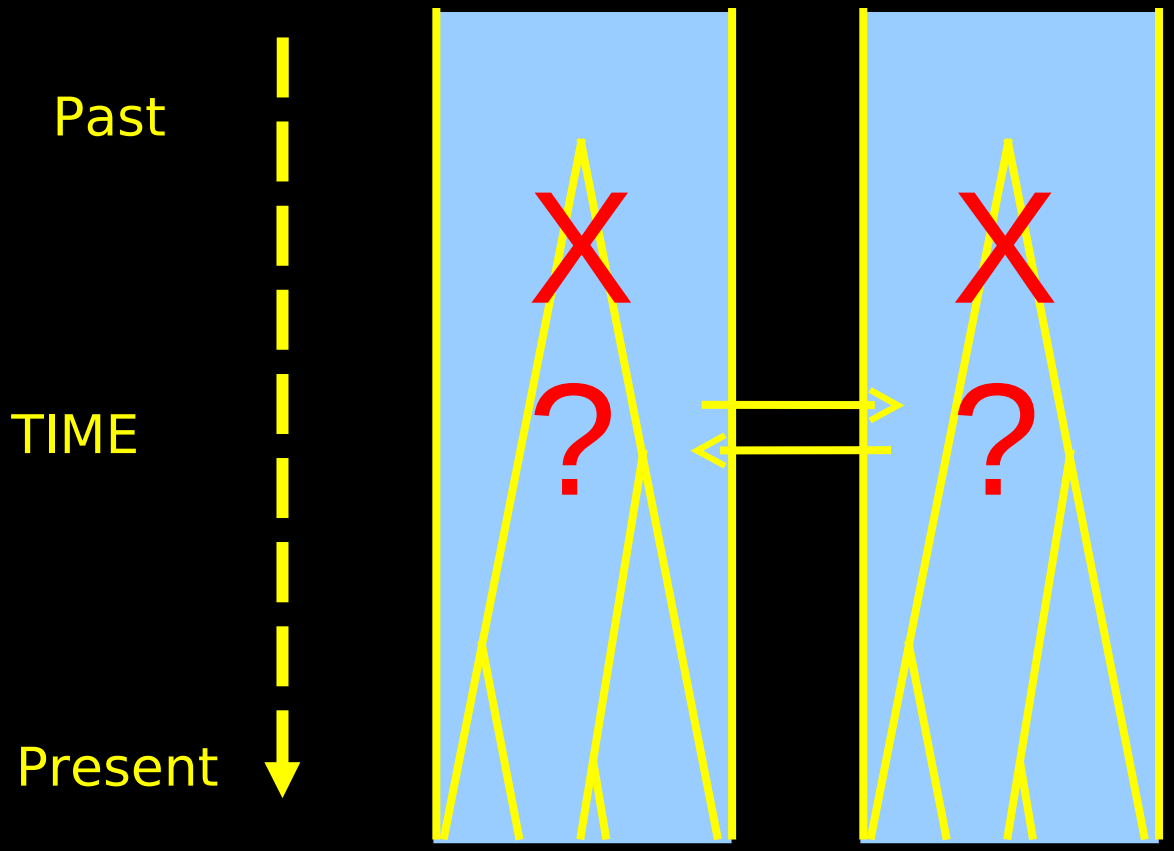
*** SHAPE OF THE GENEALOGY**

*** PATTERNS OF GENETIC DIVERSITY**

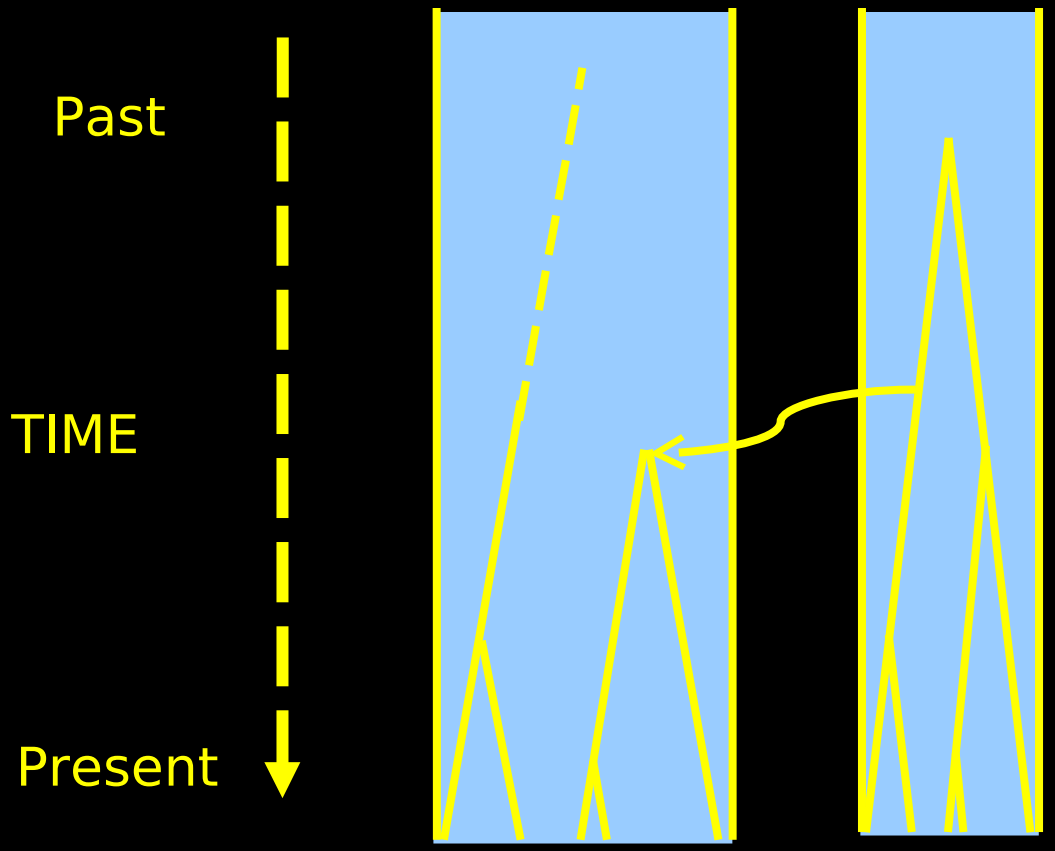
GENETIC DIVERSITY IN ISOLATED PANMICTIC POPULATIONS



GENETIC DIVERSITY
IN THE CASE OF GENE FLOW



GENETIC DIVERSITY IN THE CASE OF GENE FLOW



POPULATION STRUCTURE AND GENE FLOW

* **SHAPE OF THE GENEALOGY**
* **PATTERNS OF GENETIC DIVERSITY**

TO SUMMARISE

- Pop size changes and pop structure modify the shape of gene genealogies
- More specifically population structure often mimics bottlenecks
- (the situation is actually more complex but let us ignore that for a moment)
- (and both can mimic the effect of selection but we will ignore that too, ok?)

The Confounding Effects of Population Structure and the Sampling Scheme on the Detection a of Population Size Changes

Lounès Chikhi,*,†,‡,1 V r C. Sousa,‡,§ Pierre Luisi,**,†† Beno
d Mark A. Beaumont***

Genetics of recent habitat contraction and reduction in population size: does isolation by distance matter?

RAPHAEL LEBLOIS,*†‡ ARNAUD ESTOUP† and REJANE STREIFF†
*Laboratoire Génétique et Environnement, CNRS-UMR 5554, 34095 Montpellier, France, †Centre de Biologie et de Gestion des Populations, INRA, Campus International de Baillarguet, CS 30016, 34988 Montferrier sur Lez cedex, France

Genetic Bottlenecks Driven by Population Disconnection

THOMAS BROQUET,*†† SONIA ANGELONE,†‡ JULIE JAQUIERY,*‡‡ PIERRE JO
JEAN-PAUL LENA,§ THIERRY LENGAGNE,§ SANDRINE PLENET,§ EMILIEN
AND NICOLAS PERRIN*

The Impact of Sampling Schemes on the Site F Nonequilibrium Subdivided Popul

Thomas Städler,*,1 Bernhard Haubold,† Carlos Merino,‡ Wolfg
Peter Pfaffelhuber§
ephan‡ an

Distinguishing between population bottleneck and population subdivision by a Bayesian model choice procedure

BENJAMIN M. PETER,*† DANIEL WEGMANN*1 and LAURENT EXCOFFIER*†
*Computational and Molecular Population Genetics (CMPG), Institute of Ecology and Evolution, University of Bern, Baltzerstrasse 6, CH-3012 Bern, Switzerland, †Swiss Institute of Bioinformatics, 1015 Lausanne, Switzerland

Genetic Bottlenecks Driven by Population Disconnection

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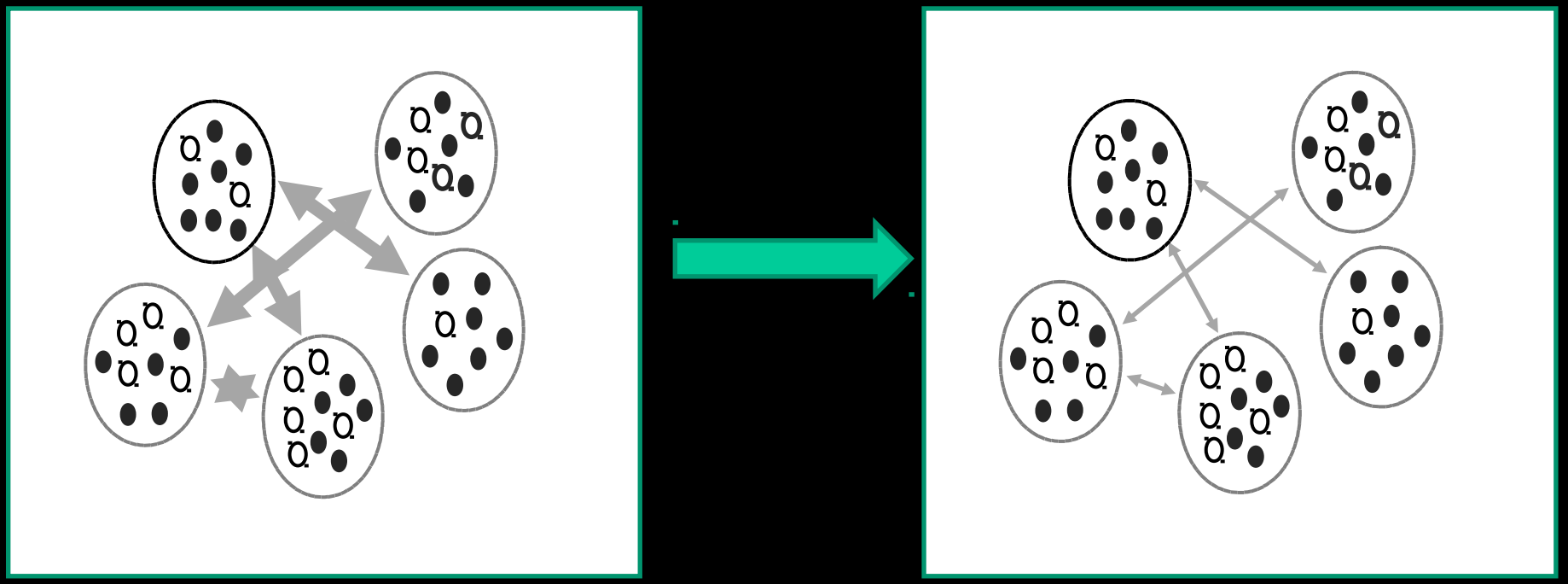

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PLOS | ONE

Signature of a Pre-Human Population Decline in the Critically Endangered Reunion Island Endemic Forest Bird *Coracina newtoni*

Jordi Salmona[1,3]*, Marc Salamolard[1,2], Damien Fouillot[1], Thomas Ghestemme[1], Jerry Larose[1], Jean-François Centon[1], Vitor Sousa[3], Deborah A. Dawson[4], Christophe Thebaud[5], Lounès Chikhi[3,5,6]

OPEN ACCESS Freely available online

PLOS | ONE

The Confounding Effect of Population Structure on Bayesian Skyline Plot Inferences of Demographic History

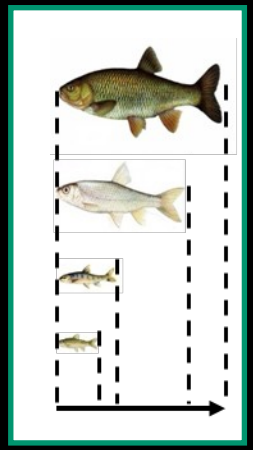
Rasmus Heller[1,2]*, Lounes Chikhi[1], Hans Redlef Siegismund[2]

MOLECULAR ECOLOGY

Molecular Ecology (2013) doi: 10.1111/mec.12321

The demographic history of populations experiencing asymmetric gene flow: combining simulated and empirical data

I. PAZ-VINAS,*†‡ E. QUÉMÉRÉ,§ L.CHIKHI,†‡¶ G. LOOT*‡ and S. BLANCHET*†



Genetic data suggest a natural prehuman origin of open habitats in northern Madagascar and question the deforestation narrative in this region

Erwan Quéméré[a,b,c,1], Xavier Amelot[d], Julie Pierson[d], Brigitte Crouau-Roy[a,b], and Lounès Chikhi[a,b,e,1]

- population size changes: real or artefacts?
- robustness to population structure?
- Can we use genetic information **alone** to identify the best model (structure *versus* pop size change)?
- How much information? (**sample size** *versus* **number of loci** ?)
- Are genomic data the answer?
- Full genomes ? SNPs ?

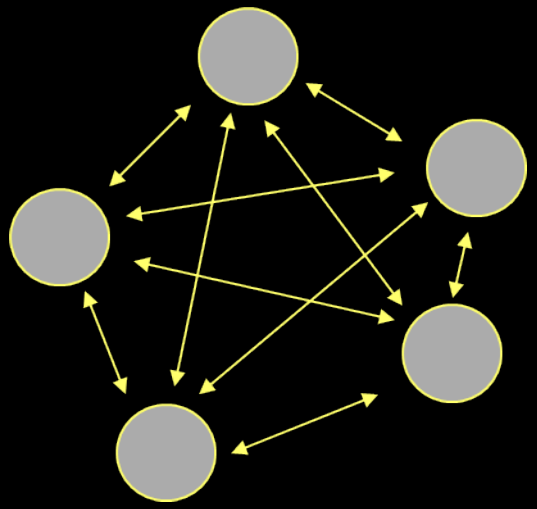
Inferring human population size and separation history from multiple genome sequences

Stephan Schiffels & Richard Durbin

Understand the properties of genetic data under simple structured and population size change models

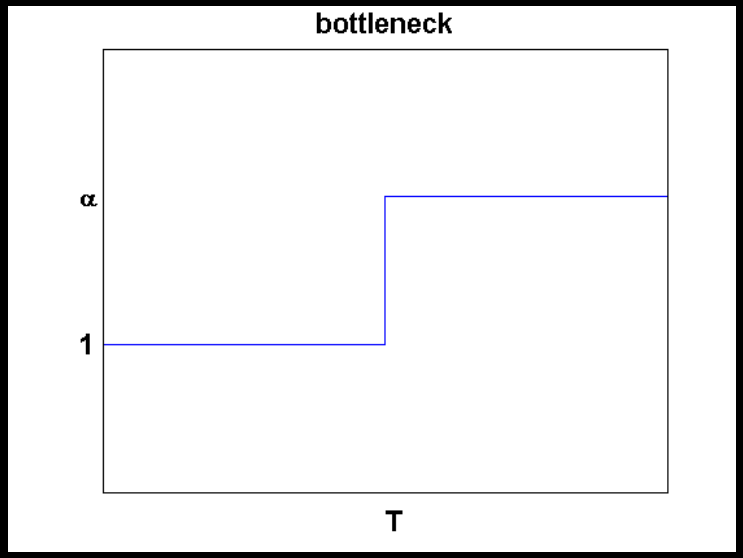
Olivier Mazet, INSA Toulouse
Simona Grusea, INSA Toulouse
Willy Rodriguez, INSA Toulouse
Simon Boitard, INRA Toulouse

n/island *versus* stepwise population size change



alpha*N

N



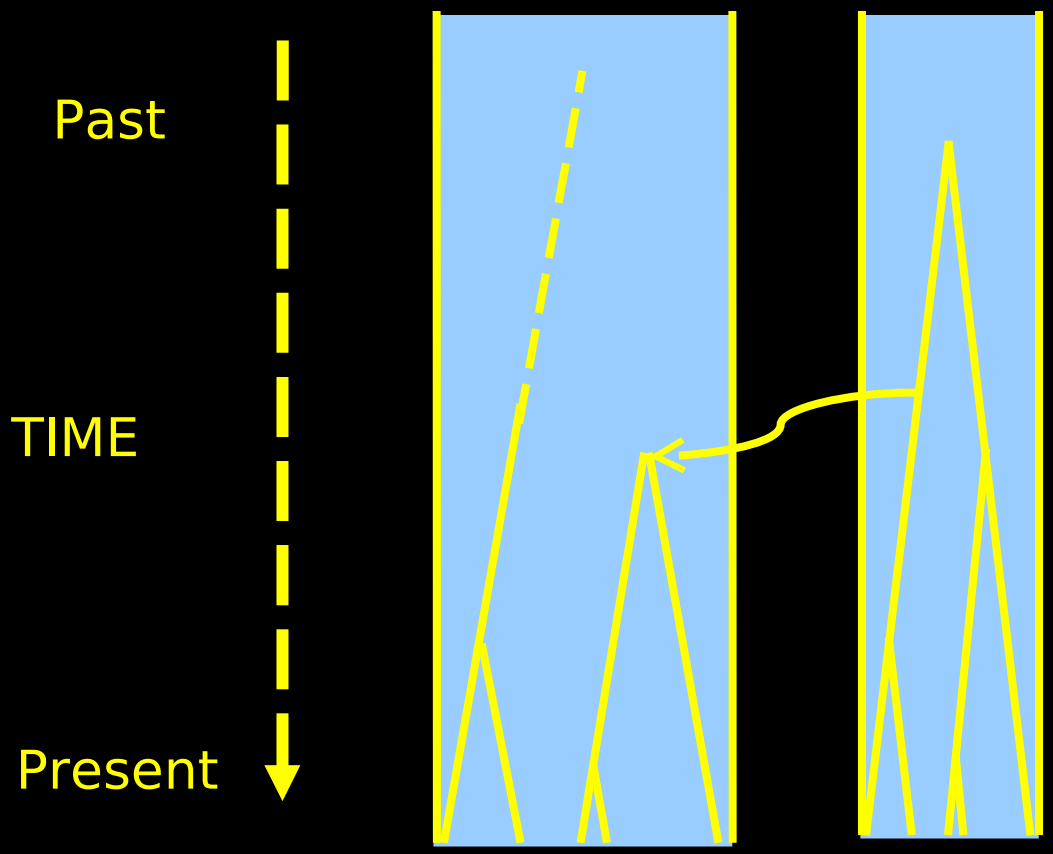
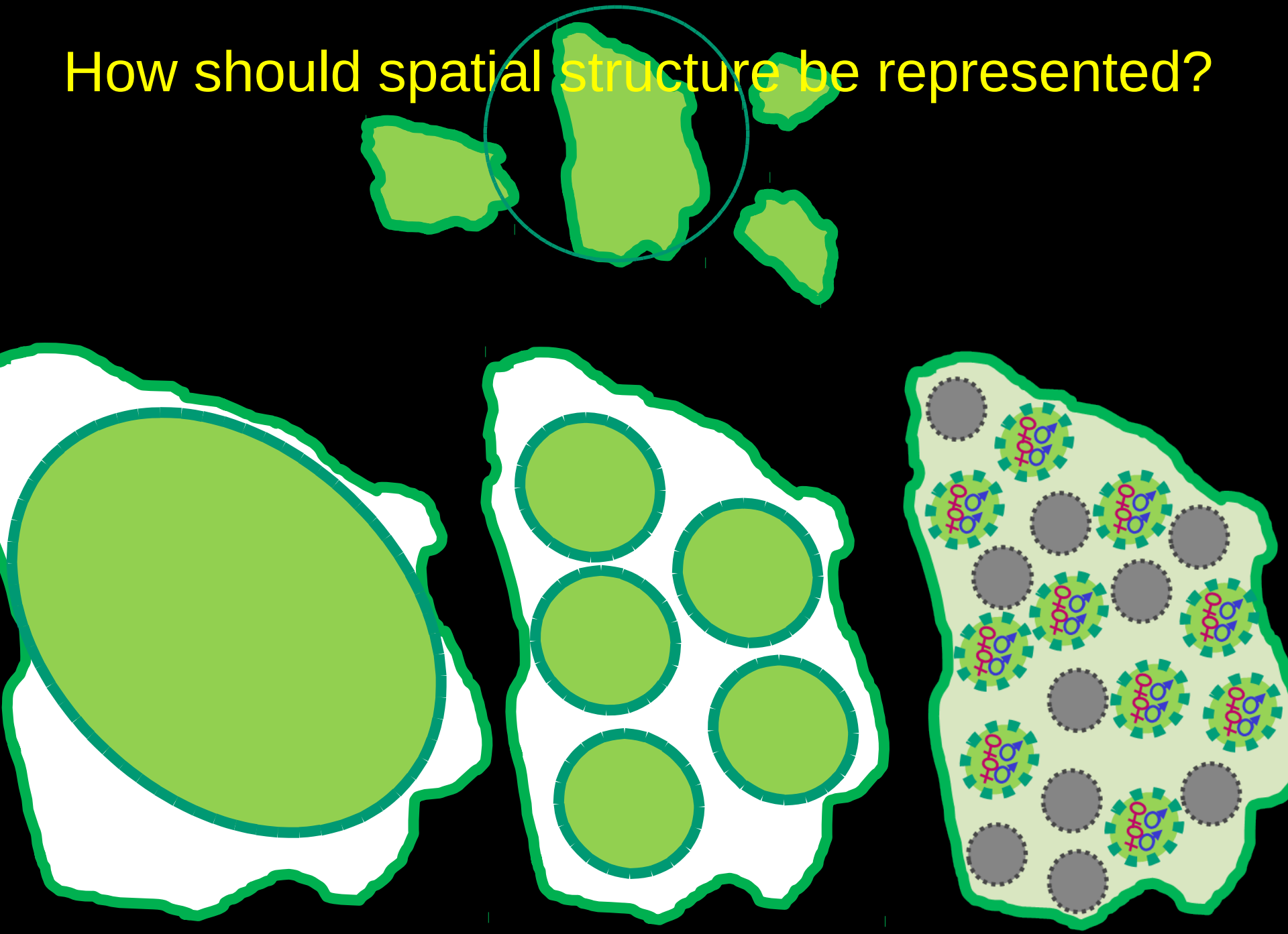
Present

Past

n-island
Wright (1931)

alpha < 1
alpha > 1

Population size change


Past
TIME
Present

How should spatial structure be represented?