

Effet de la structure spatiale et familiale de la population sur la taille efficace

Ou plutôt

Quelques conséquences de la structure des populations sur la notion de taille efficace

Lounes Chikhi

Lab. Evolution et Diversité Biologique, **CNRS, Toulouse, France**

Population and Conservation Genetics Group, **IGC, Portugal**

Laboratoire International Associé (LIA) BEEG-B

Séminaire Taille Efficace, INRA AgroParisTech, Paris 12 Mai 2016

Outline

1. Context:
 1. habitat loss and fragmentation
 2. endangered species
 3. Social and spatial structure
2. The importance of structure
 1. reminder
 2. there is hope
3. Or maybe there isn't

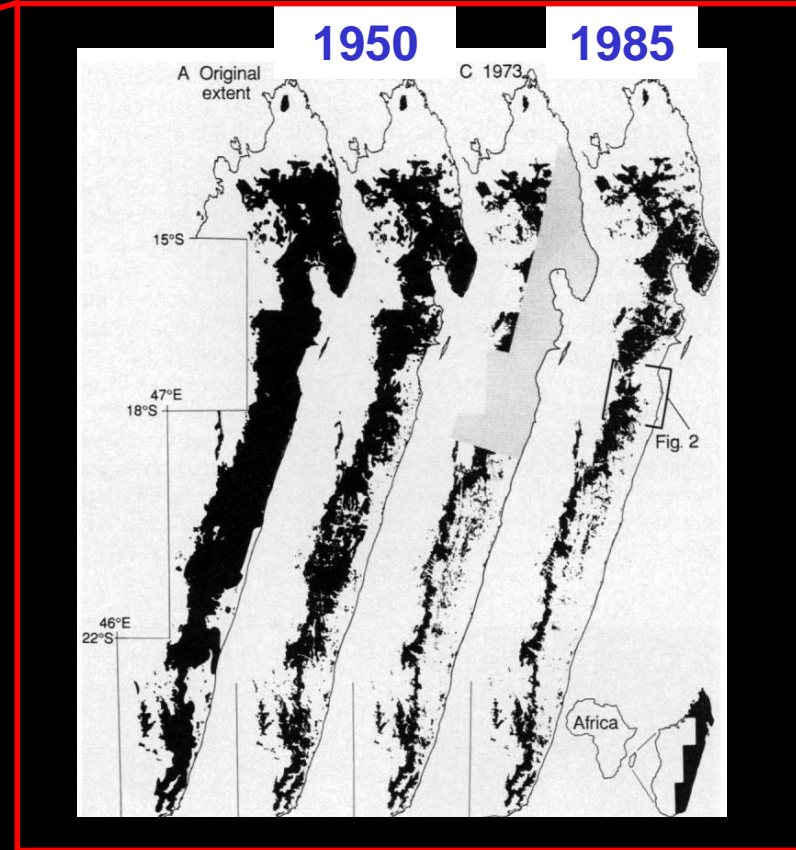
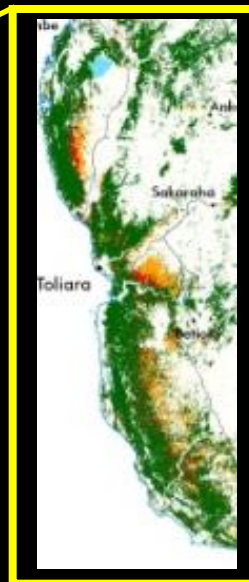
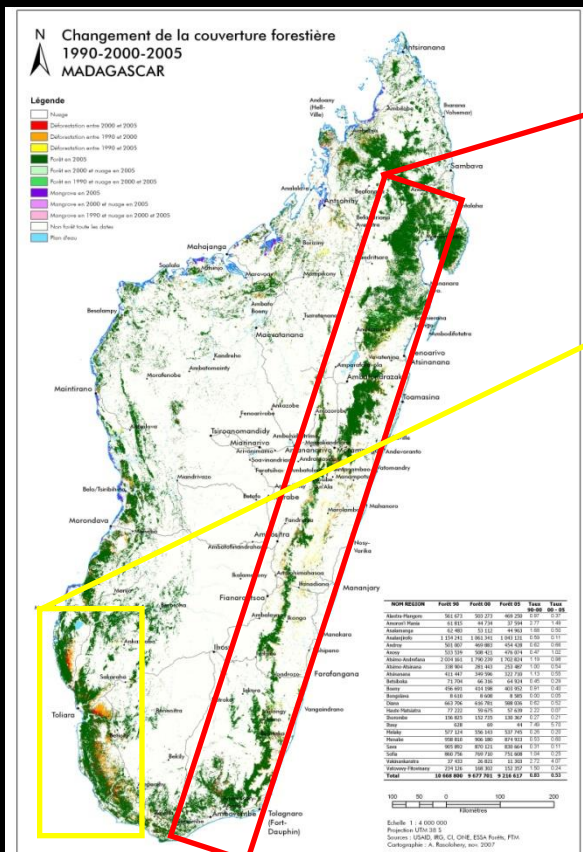
HABITAT LOSS AND FRAGMENTATION IN MADAGASCAR

FOREST HABITATS : 10-20 % TOTAL SURFACE

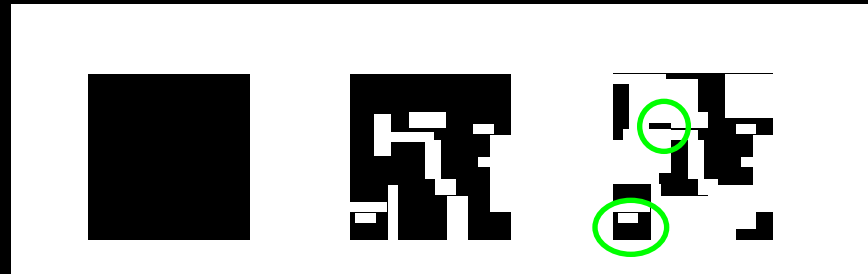
90% of vertebrates depend on forested habitats

Large size lemurs have disappeared

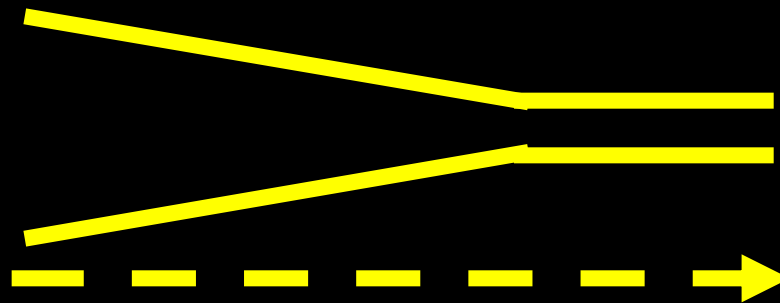
MOST present-day lemurs are threatened in Madagascar



Habitat fragmentation and loss as a temporal process

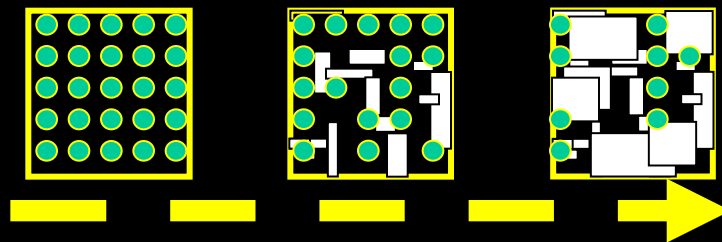


time



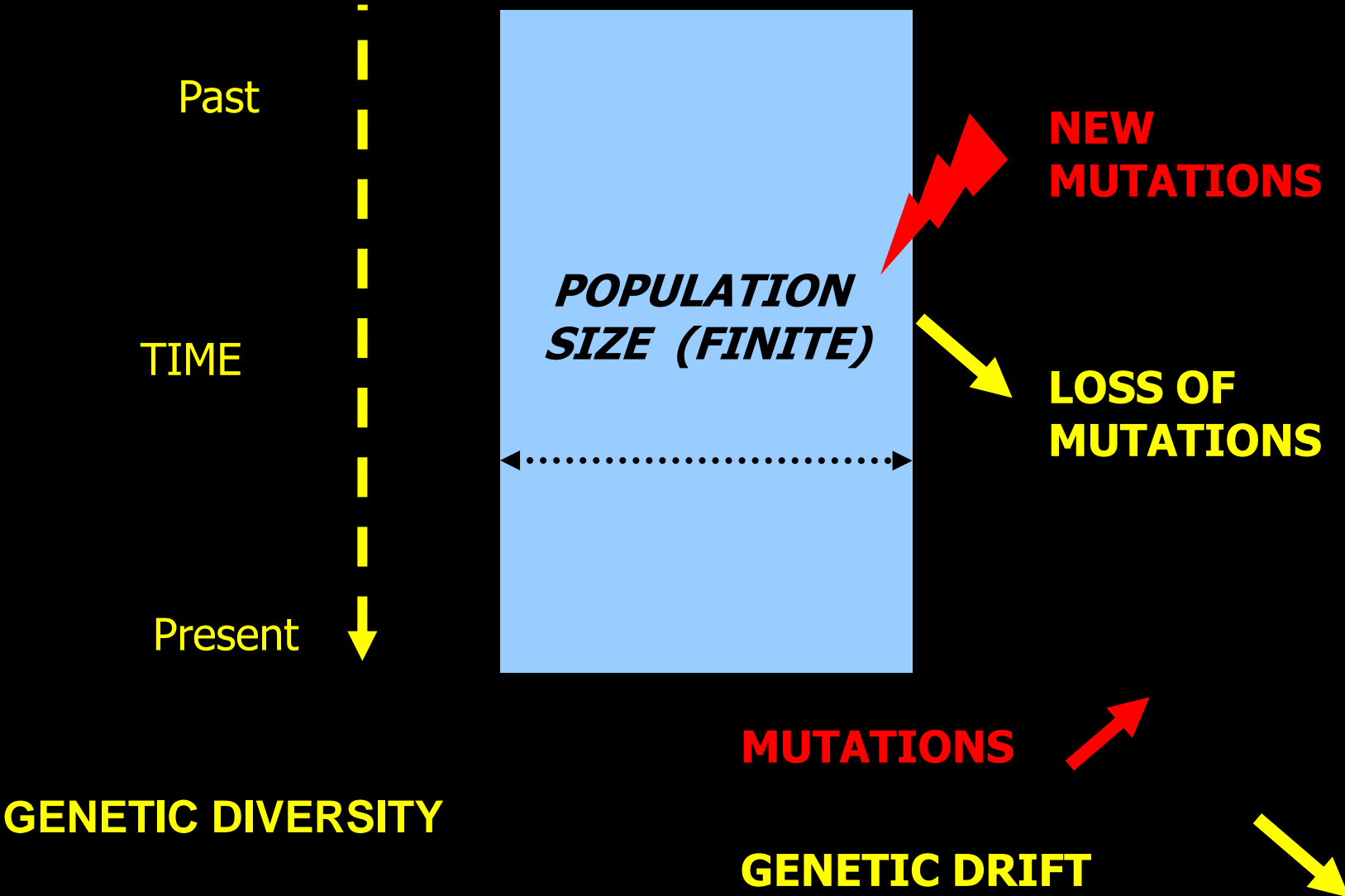
time

ALSO A SPATIAL PROCESS: population structure

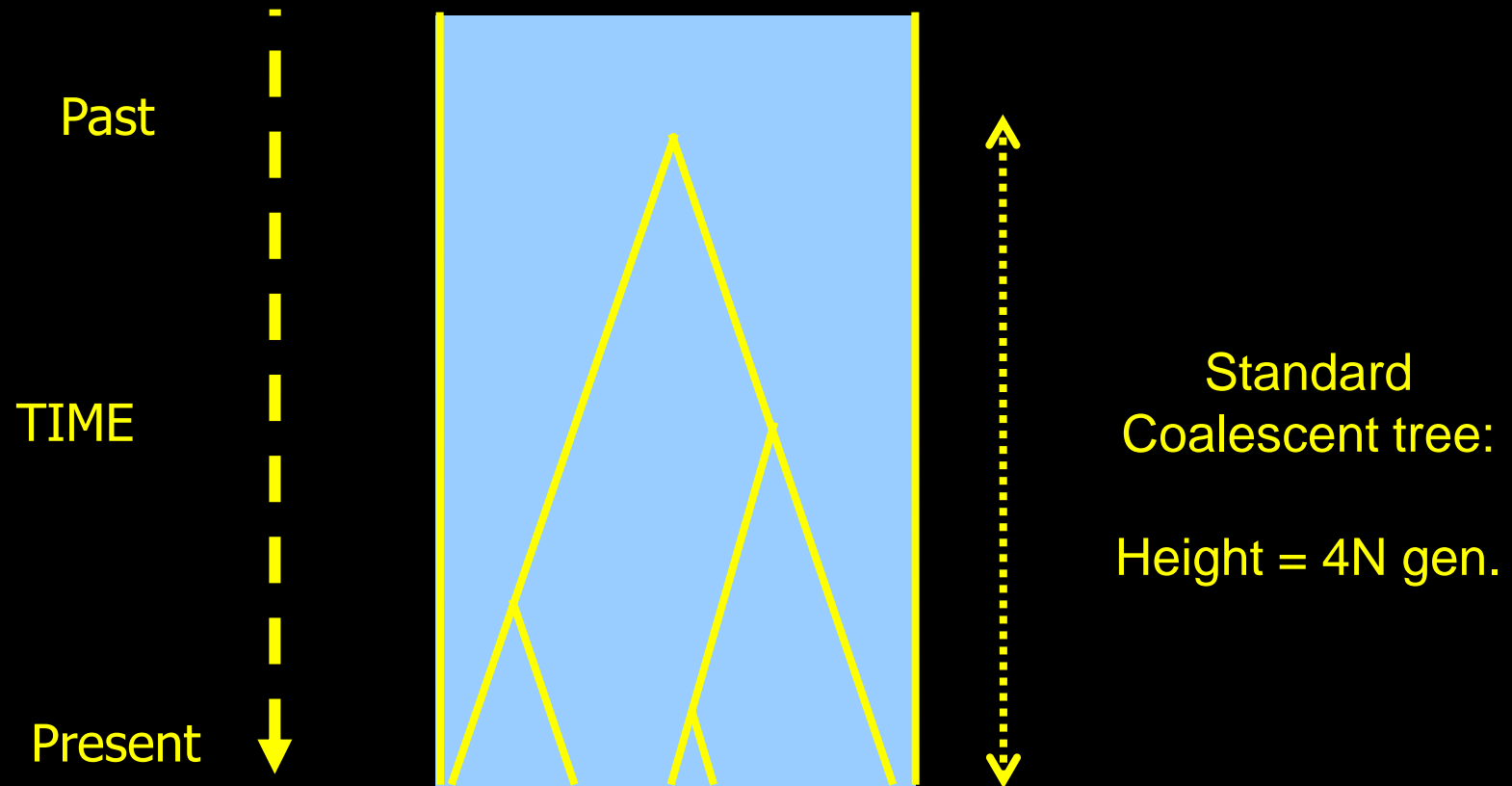


time

GENETIC DIVERSITY IN A PANMICTIC POPULATION



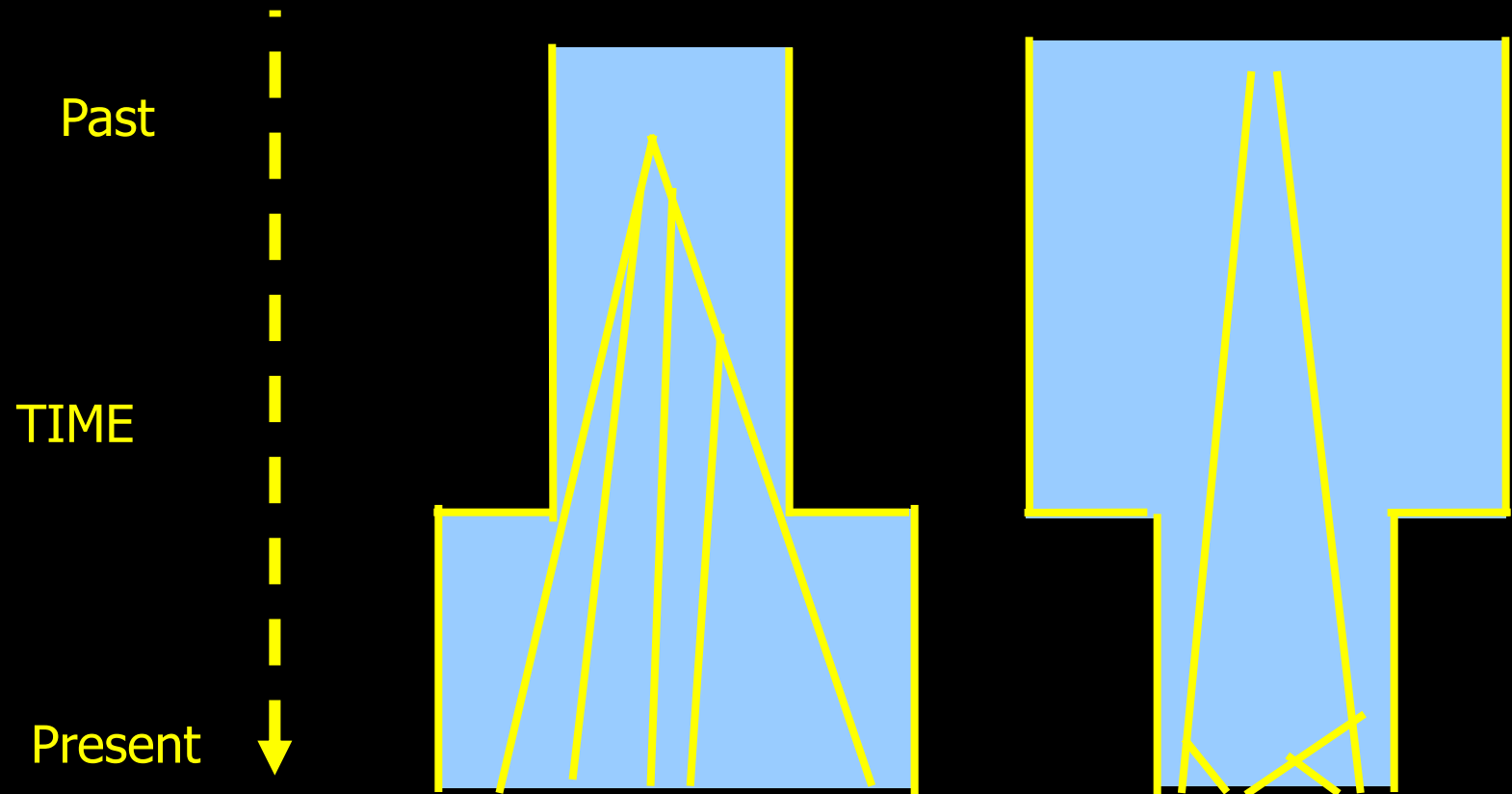
GENETIC DIVERSITY IN A PANMICTIC POPULATION



STANDARD COALESCENT (STATIONARY POPULATION) PREDICTS

- * **SHAPE OF THE GENE TREE / GENEALOGY**
- * **PATTERNS OF GENETIC DIVERSITY**

GENETIC DIVERSITY WHEN POPULATION SIZE CHANGES

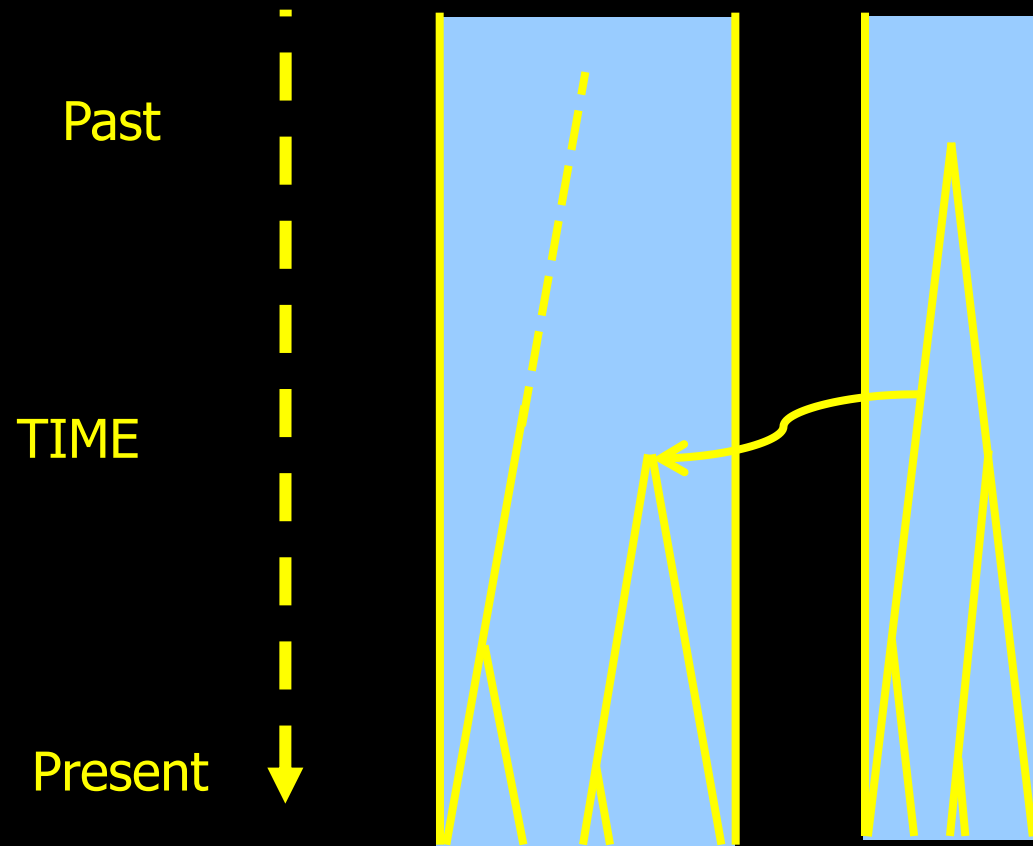


POPULATION SIZE CHANGES / DEMOGRAPHY

*** SHAPE OF THE GENEALOGY**

*** PATTERNS OF GENETIC DIVERSITY**

GENETIC DIVERSITY IN THE CASE OF GENE FLOW



POPULATION STRUCTURE AND GENE FLOW

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

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

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

Loumès Chikhi,^{*,†,‡,1} V. C. Sousa,^{‡,§} Pierre Luisi,^{*,**} Beno. Mark A. Beaumont^{***}

Genetic Bottlenecks Driven by Population Disconnection

THOMAS BROQUET,^{*,††} SONIA ANGELONE,^{†‡} JULIE JAQUIERY,^{*,††} PIERRE JOUVEAU,[†] JEAN-PAUL LENA,[§] THIERRY LENGAGNE,[§] SANDRINE PLENET,[§] EMILIE PERRIN,[†] AND NICOLAS PERRIN^{*}

The Impact of Sampling Schemes on the Site Frequency Spectrum: Distinguishing between population bottleneck and Nonequilibrium Subdivided Population by a Bayesian model choice procedure

Thomas Städler,^{*,1} Bernhard Haubold,[†] Carlos Merino,[‡] Wolfgang Peter Pfaffelhuber[§]

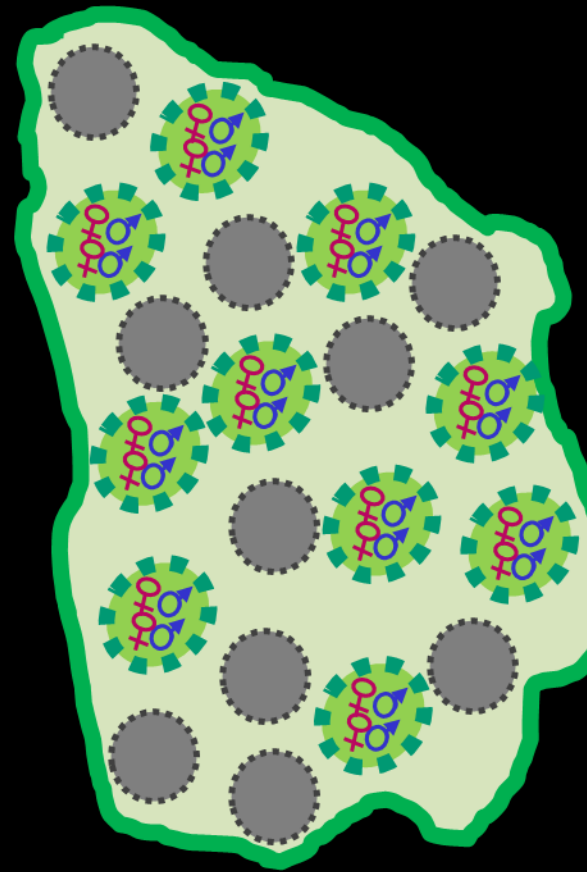
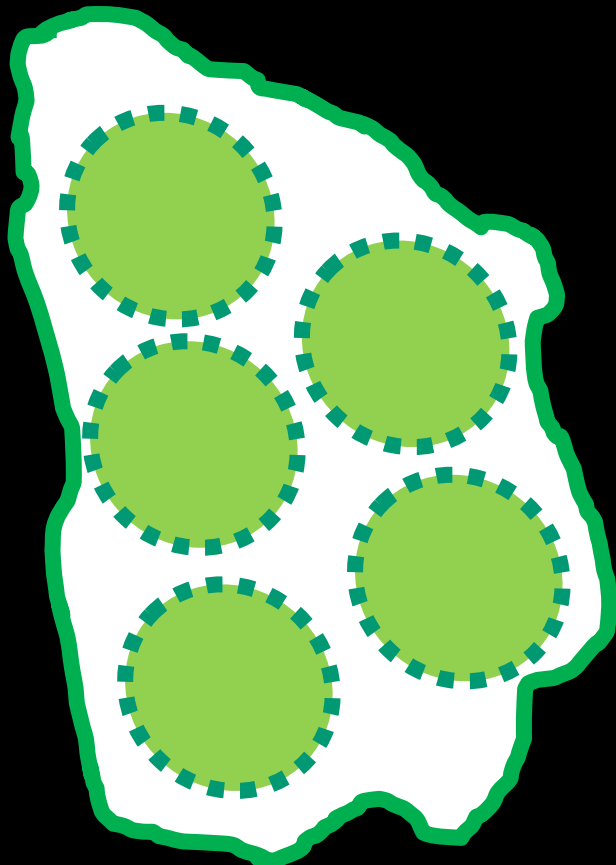
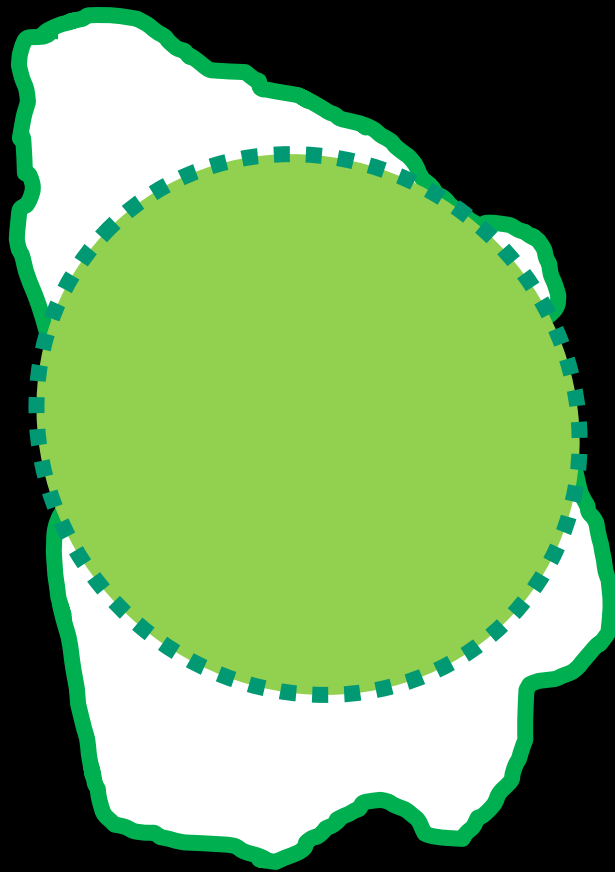
BENJAMIN M. PETER,^{*,†} DANIEL WEGMANN^{*1} and LAURENT EXCOFFIER^{*†}
^{*}Computational and Molecular Population Genetics (CMPG), Institute of Ecology and Evolution, University of Bern, Baltzerstrasse 6, CH-3012 Bern, Switzerland, [†]Swiss Institute of Bioinformatics, 1015 Lausanne, Switzerland

ephan[†] and

Population structure and Population size changes

1. Population structure can mimic bottleneck signals
2. The signal is particularly strong when
 1. Genetic differentiation is high (gene flow is limited)
 2. Genetic diversity is high
 3. The number of loci used is large
3. The effect is less important when more than one population is sampled
4. Asymmetrical gene flow, social structure ?

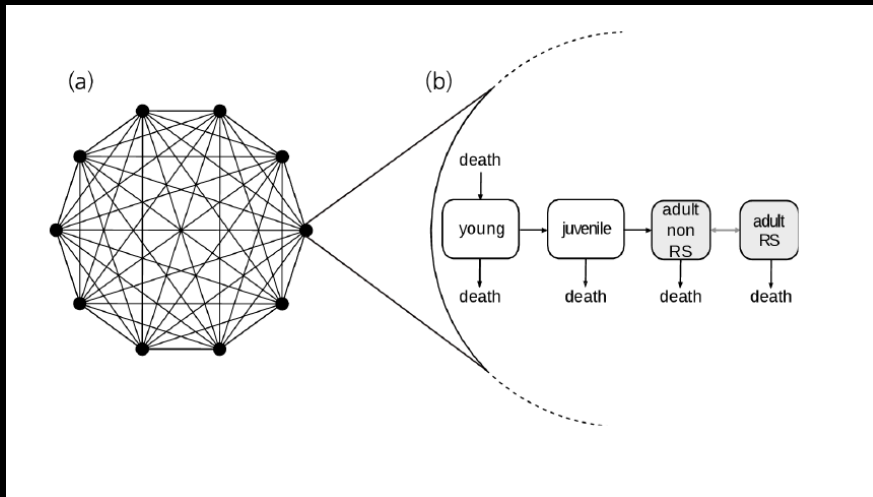
How should populations be modelled?



Integrating social structure in pop gen models

On some genetic consequences of social structure, mating systems, dispersal, and sampling

Bárbara R. Parreira^{a,b,1} and Lounès Chikhi^{a,c}



PhD defence:
26 April 2016
Fac. Ciência Univ. Lisboa

Mating system	# males	# females	Structure	dispersing sex
Monogamy	1	1	island	male male, female
Polygynandry	2	2	island	male male, female
Extreme Polygyny	1	10	island	male male, female
Moderate Polygyny	4	10	island	male male, female

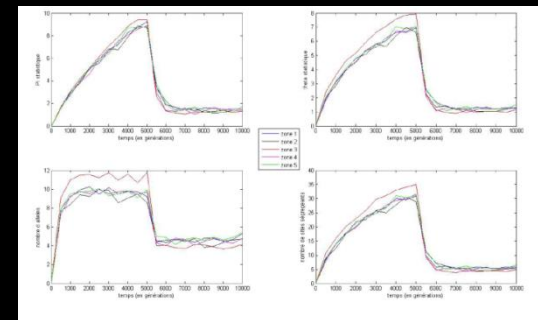
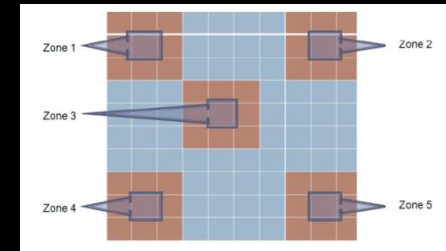
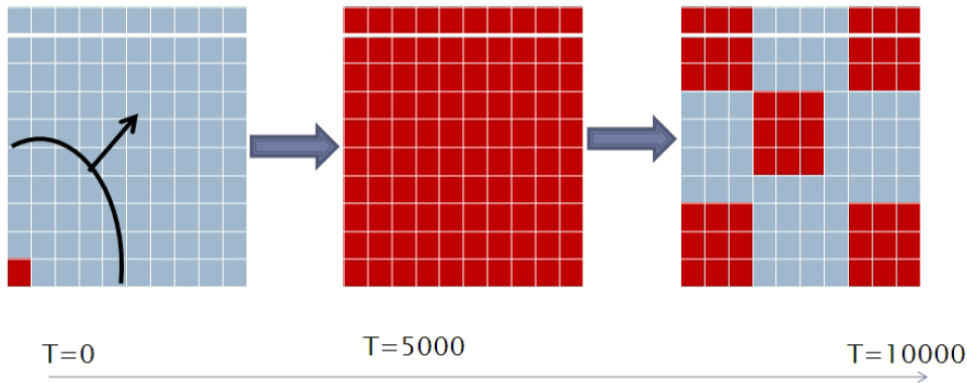


INTEGRATE SPACE AND TIME: SINS: (Simulating INdividuals in Space)



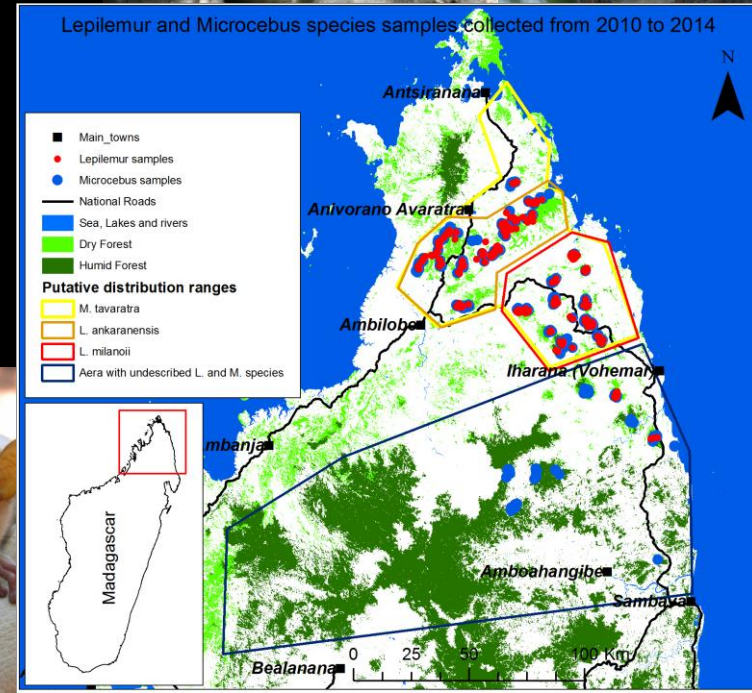
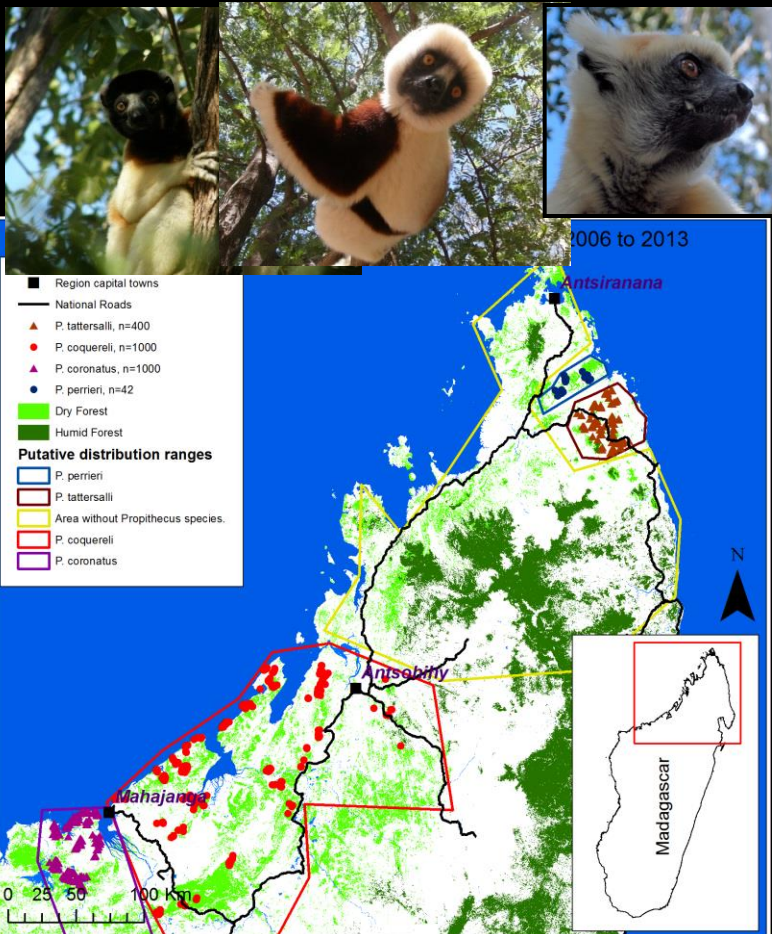
**EXPANSION
(Neolithic)**

FRAGMENTATION



**Rita RASTEIRO, Pierre Antoine
BOUTTIER, Damien MONIER
Vitor SOUSA, Tiago MAIE, Nathalie
CHENIN, Douglas CURBELO**

COMPARATIVE ANALYSES ACROSS SPECIES AND REGIONS

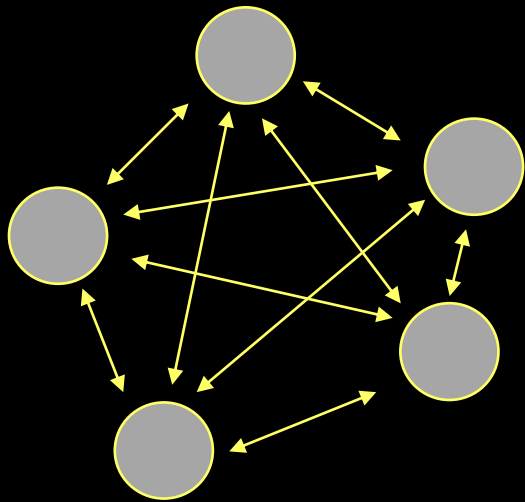


Olivier Mazet, INSA Toulouse
Simona Grusea, INSA Toulouse
Willy Rodriguez, INSA Toulouse
Simon Boitard, INRA Toulouse

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

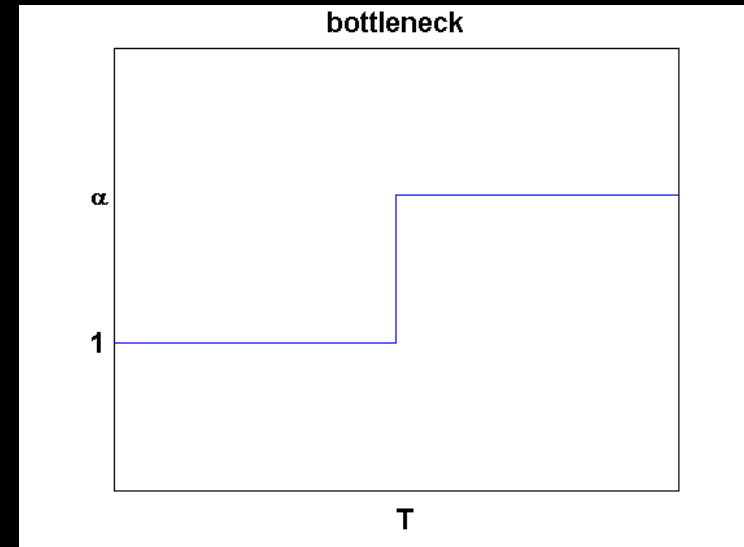


Structured symmetrical island model (StSI) versus Single step population size change (SSPSC)



$\alpha * N$

N



Present

Past

n-island
Wright (1931)

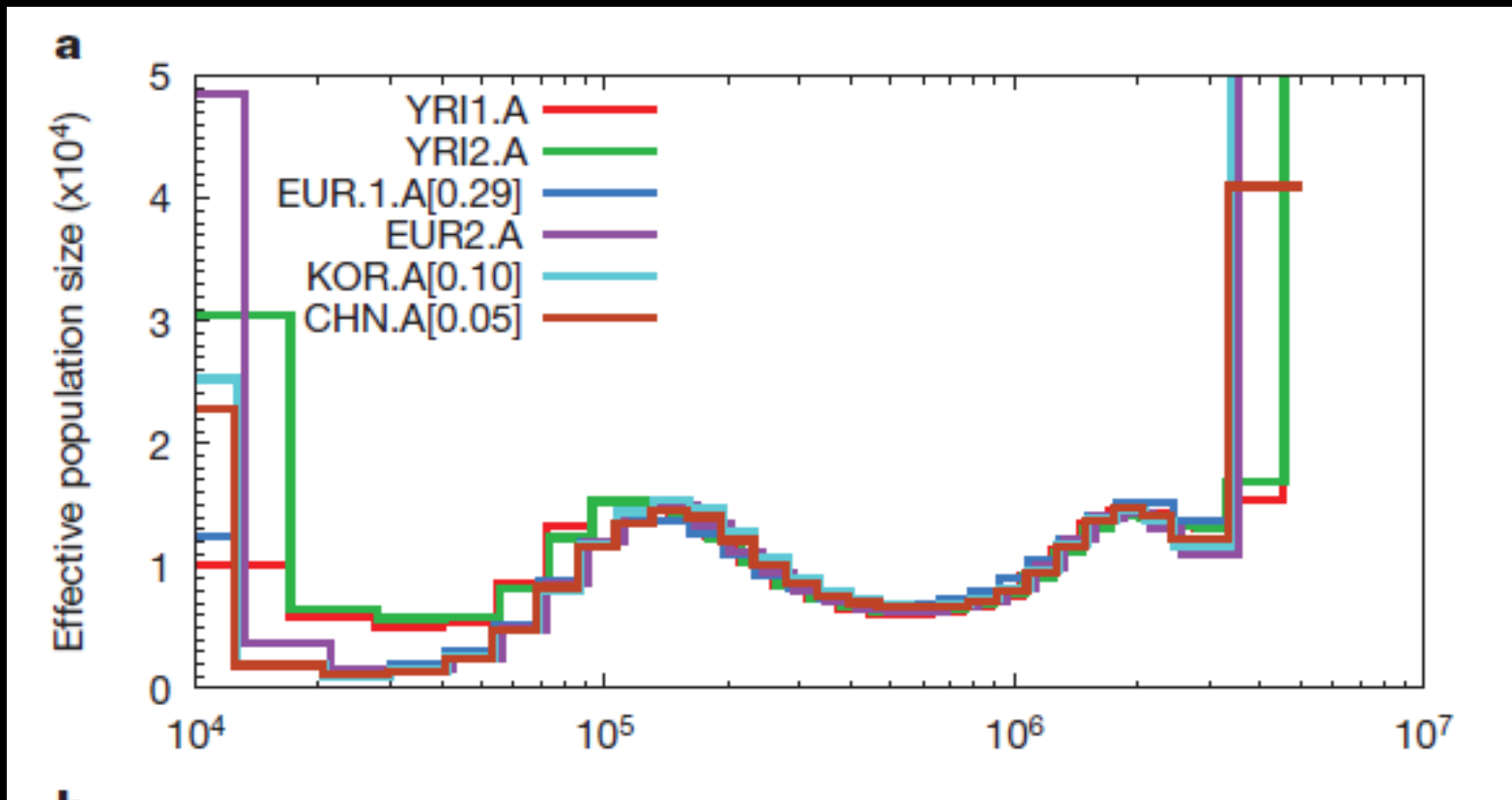
$\alpha < 1$
 $\alpha > 1$

Population size
change

Population structure and Population size changes

1. If population structure mimics bottleneck signals, doesn't that mean that structured populations cannot be summarised by a single effective size ?
2. When is the concept of effective size useful and biologically meaningful ?
3. Gene flow = 0 → Infinite N_e ?
4. What about Asymmetrical gene flow, social structure ?

Li and Durbin (2011): 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 Durbin¹

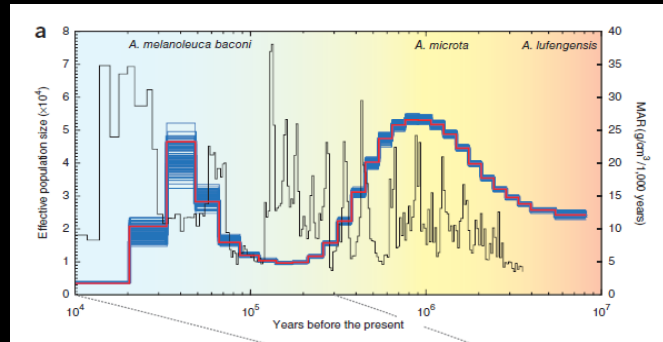
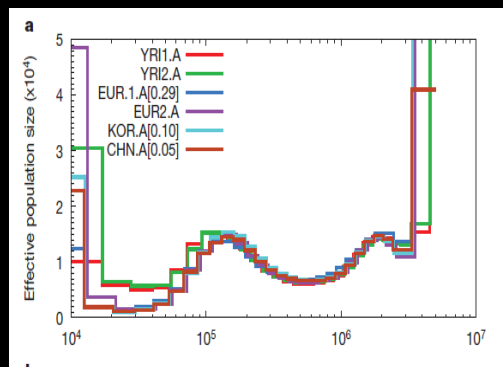
Li and Durbin (2011): THE PSMC (Pairwise Sequentially Markovian Coalescent)

LETTER

doi:10.1038/nature10231

Inference of human population history from individual whole-genome sequences

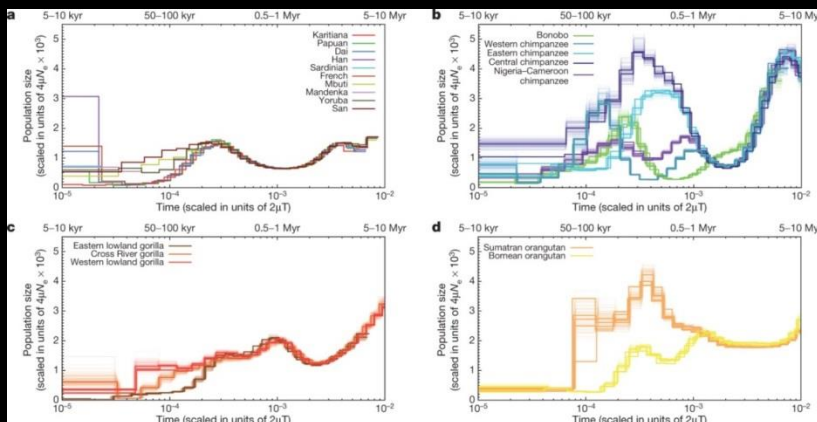
Heng Li^{1,2} & Richard Durbin³



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 Guo², Yibo Hu¹, Weiming He², Shanning Zhang⁴, Wei Fan², Lifeng Zhu¹, Dong Li², Xuemei Zhang², Quan Chen², Hemin Zhang⁵, Zhihe Zhang⁶, Xuelin Jin⁷, Jinguo Zhang⁸, Huanming Yang², Jian Wang², Jun Wang^{2,9} & Fuwen Wei¹



J Prado-Martinez et al. 2013

ORIGINAL ARTICLE

On the importance of being structured: instantaneous coalescence rates and human evolution—lessons for ancestral population size inference?

O Mazet^{1,7}, W Rodríguez^{1,7}, S Grusea¹, S Boitard^{2,3} and L Chikhi^{4,5,6}

What history of population size change would we infer in a n-island model without population size change?

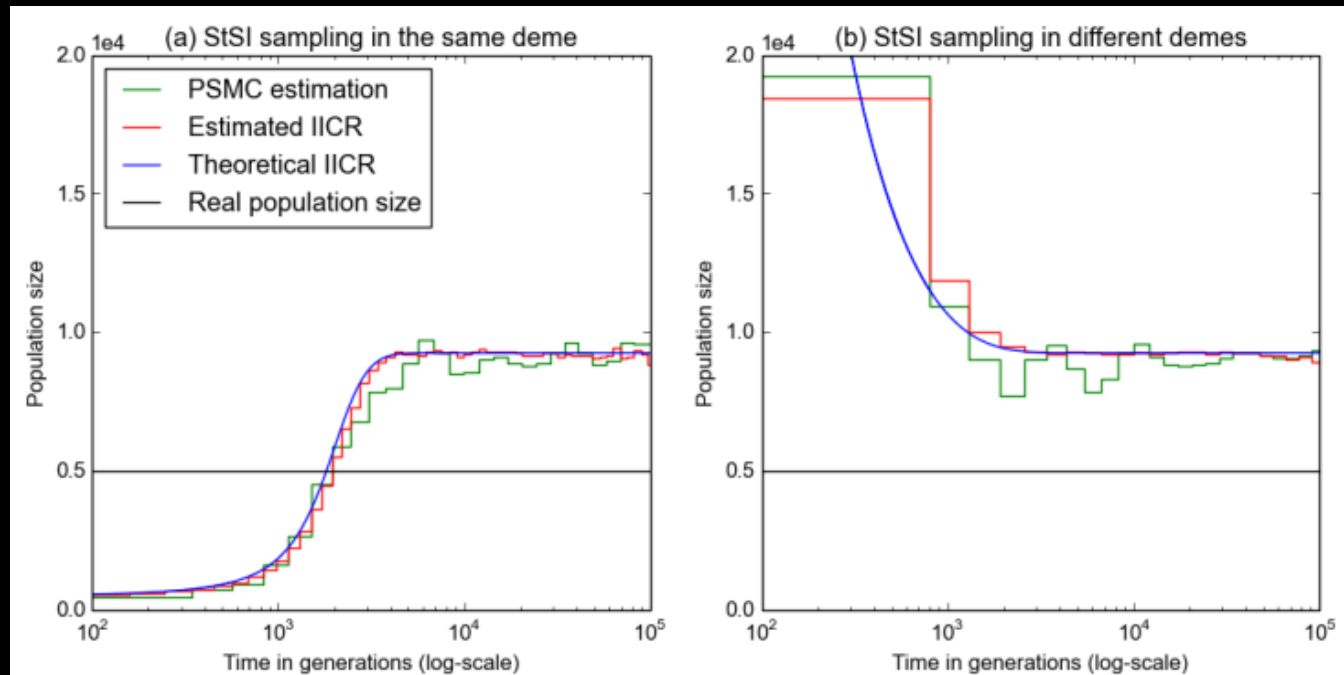
$$\mathbb{P}(T_2 > t) = e^{-\int_0^t \frac{1}{\lambda(x)} dx}$$

$$f_{T_2}^{PSC}(t) = (1 - e^{-\int_0^t \frac{1}{\lambda(x)} dx})' = \frac{1}{\lambda(t)} e^{-\int_0^t \frac{1}{\lambda(x)} dx}$$

$$\lambda(t) = \frac{\mathbb{P}(T_2 > t)}{f_{T_2}^{PSC}(t)}$$

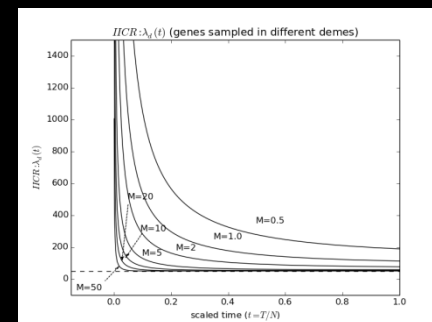
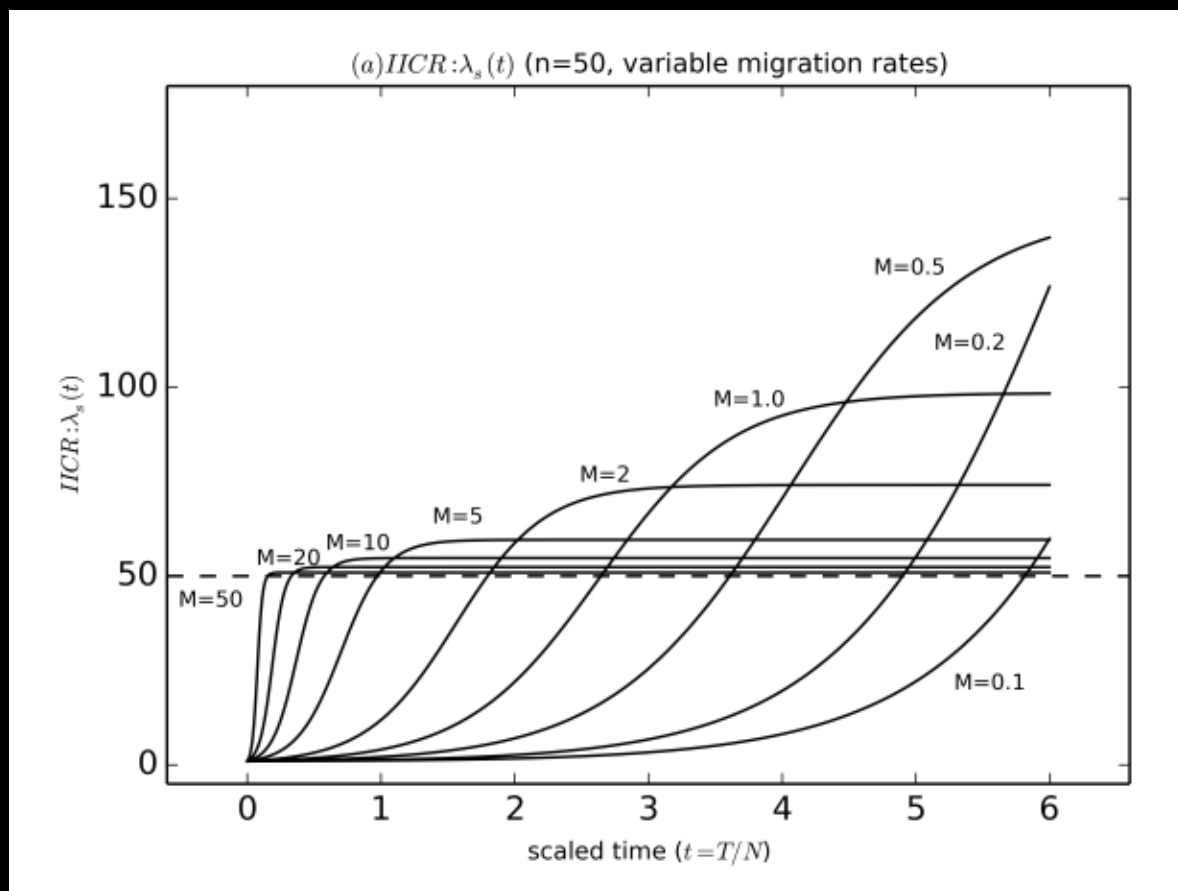
1. Simulate under any model
2. Compute this statistic (IICR: Inverse Instantaneous Coalescence Rate)
3. Plot the IICR

What history of population size change would we infer in a n-island model without population size change?



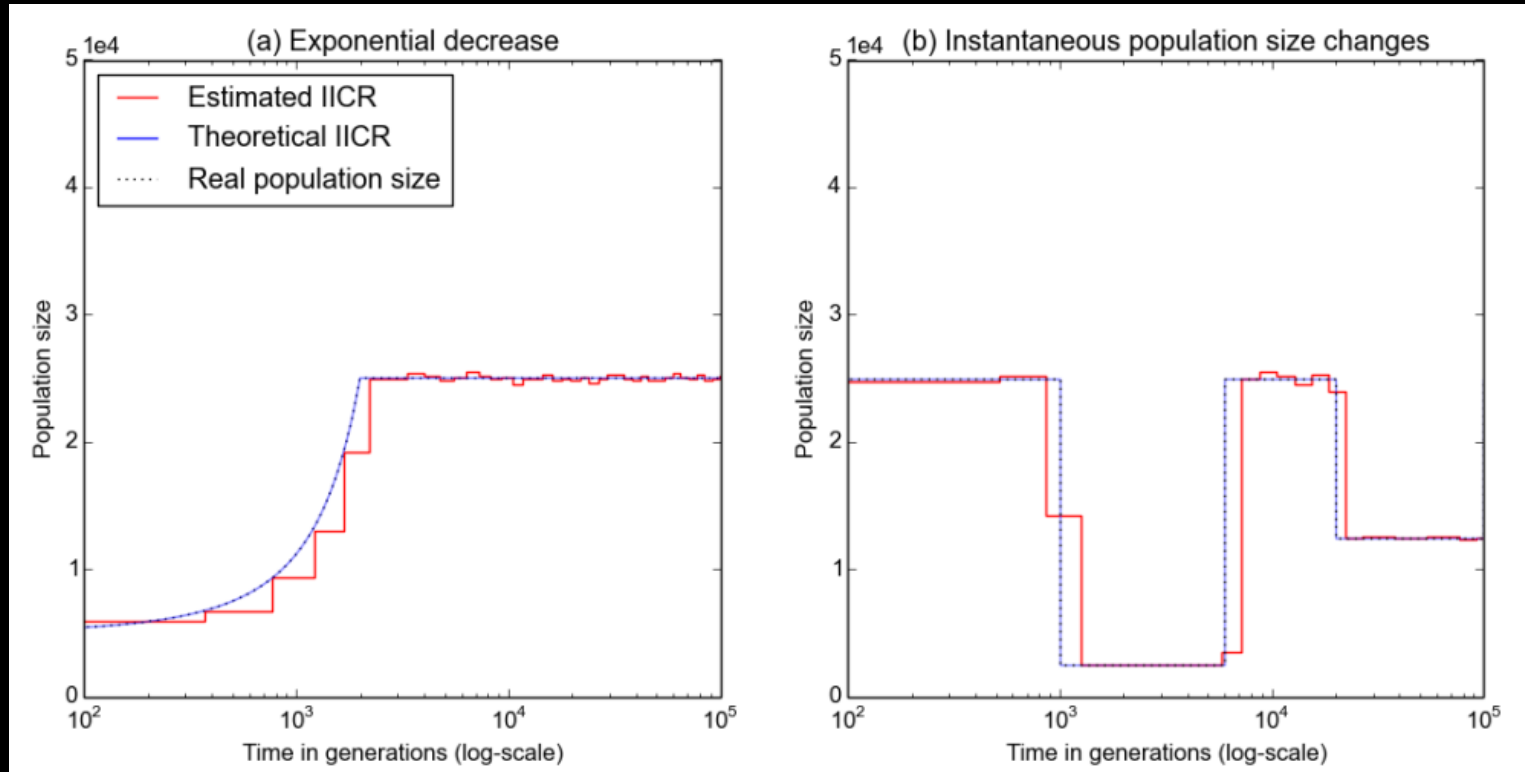
Comparison with the PSMC:
Pop structure
NO population size change

What history of population size change would we infer in a n-island model without population size change?



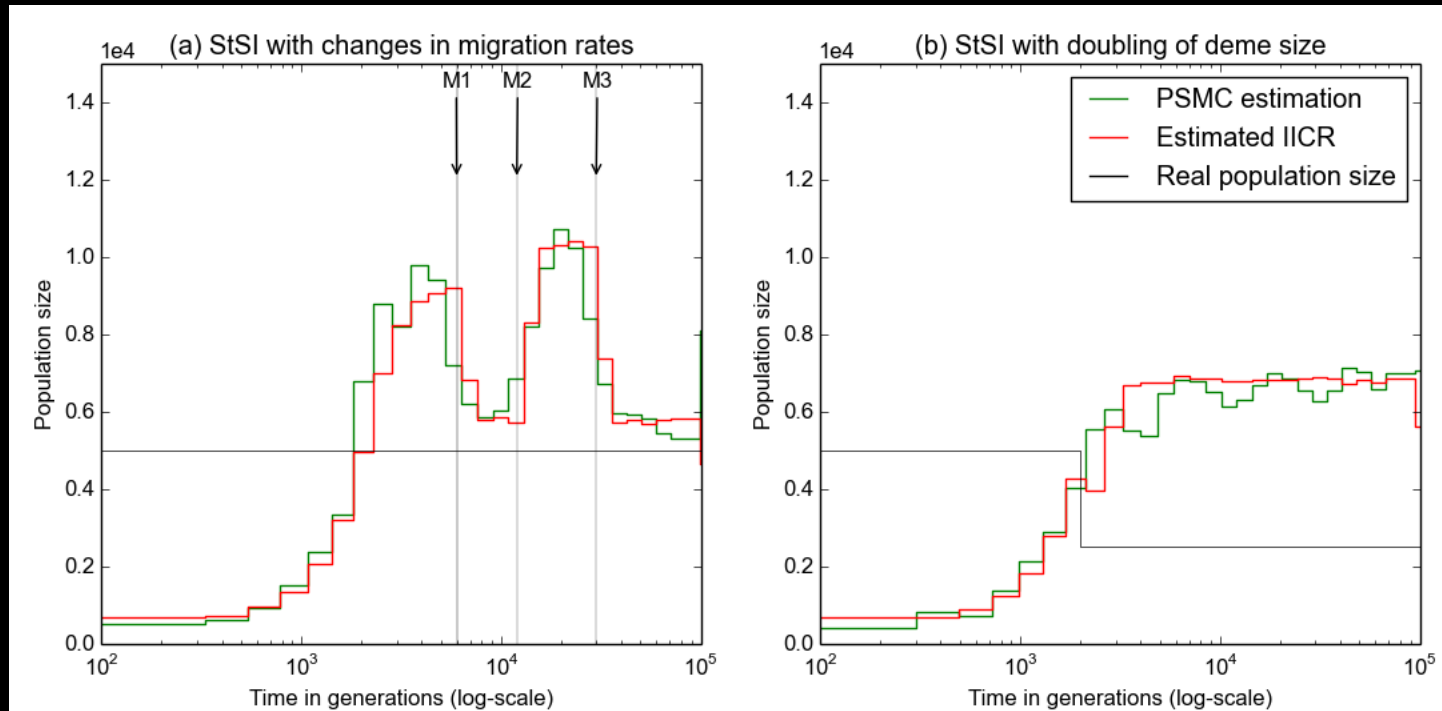
$n = 50$ islands
 M : number of migrants

What history of population size change would we infer?



Simple validation:
No pop structure
Just population size change

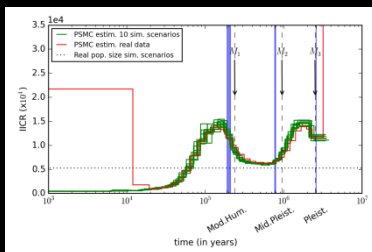
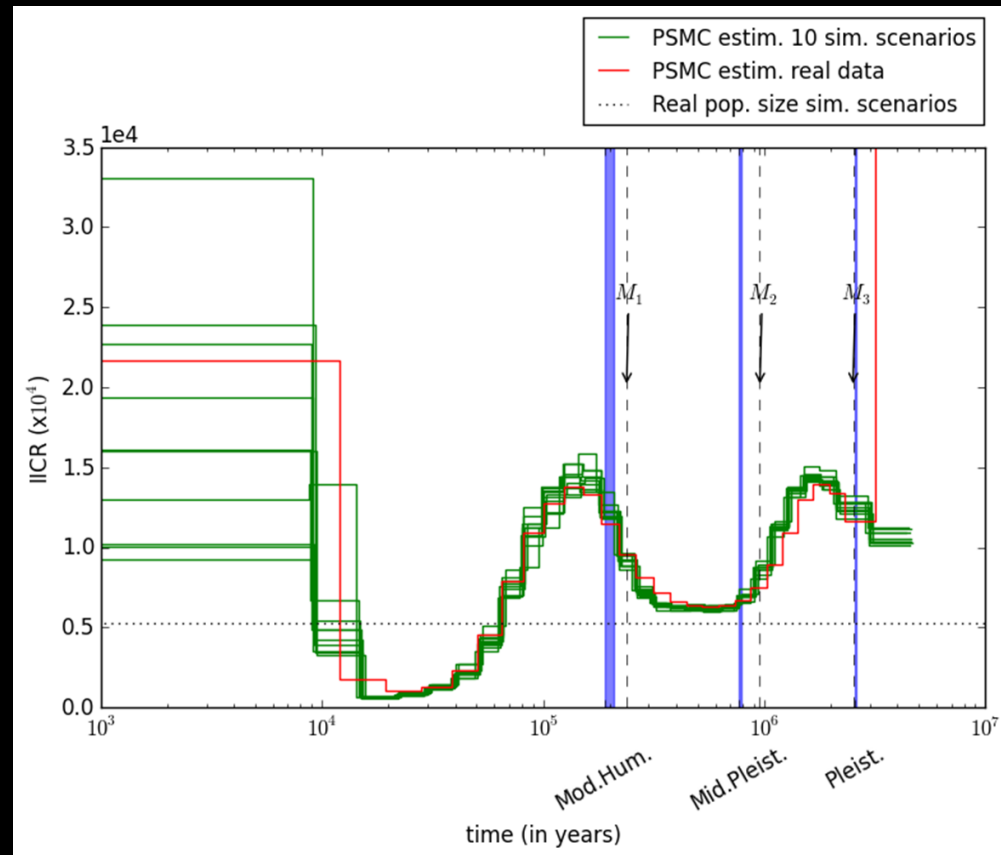
What history would we infer under more complex models?



Pop structure
Change in gene flow
No Pop size change

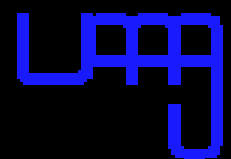
Pop structure
No change in gene flow
Increase in pop size

Human evolutionary history with structure and changes in migration rates?



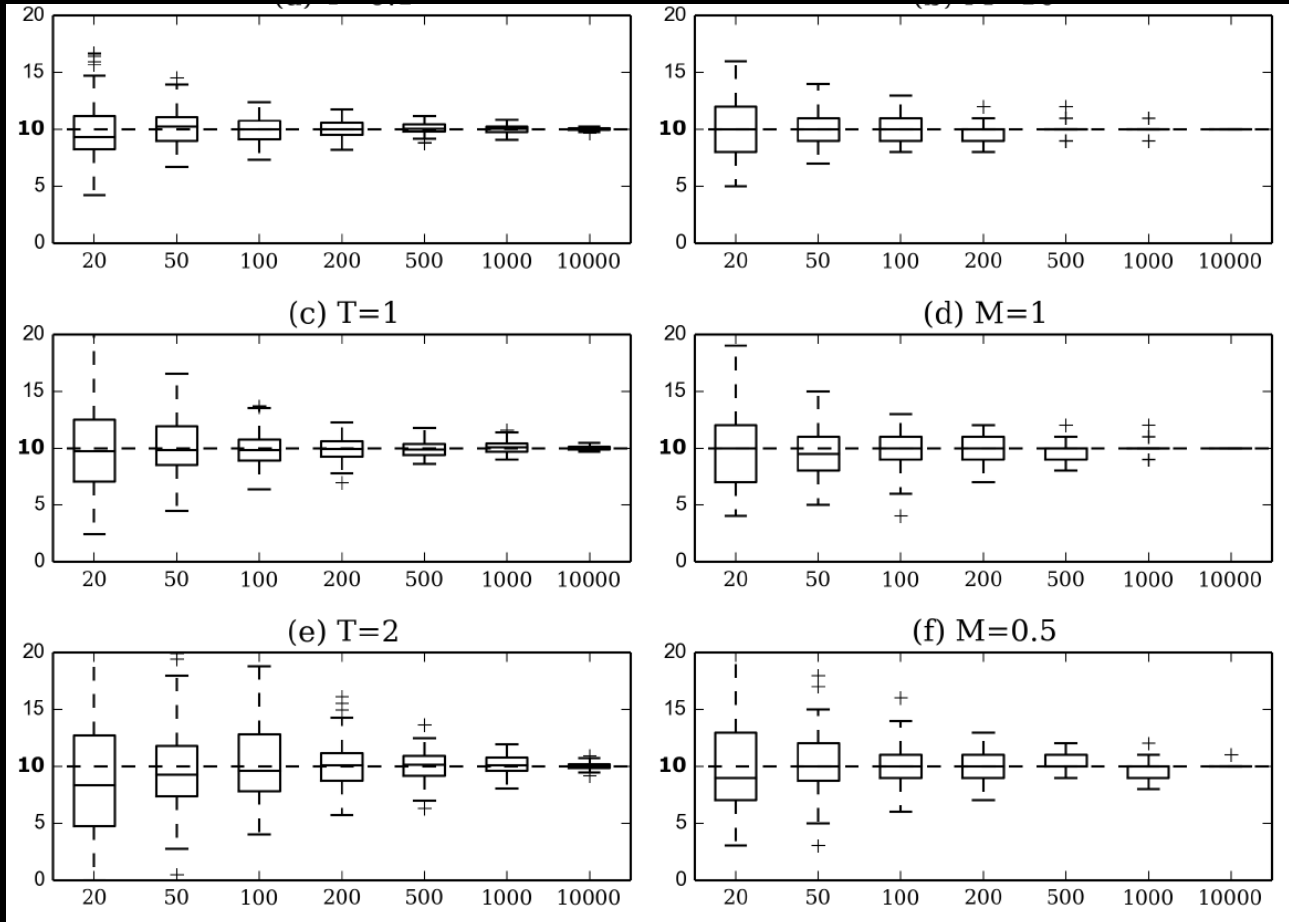
CONCLUSIONS AND PERSPECTIVES

- Genomic data increase our ability to do inference but under the wrong model (wrong family of models) they may be misleading
- Still preliminary: we need to move towards inference of population structure (this is already done to some extent)
- Our models are very simplistic: no change in the number of islands, and pop size.
- How other aspects of population structure (social, spatial, asymmetrical gene flow) should be integrated remains an under-studied area.



Estimation of the number of islands (n) And ratio of population size change (α)

Alpha = 10



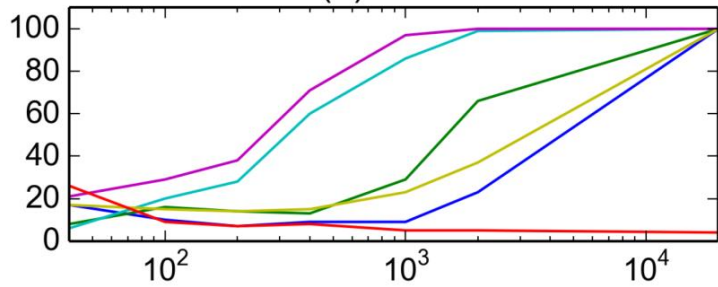
$n = 10$

Demographic inference using genetic data from a single individual:
Separating population size variation from population structure

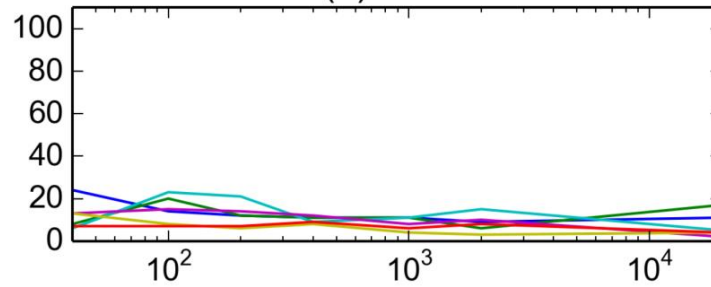
Olivier Mazet^a, Willy Rodríguez^a, Lounès Chikhi^{b,c,d,*}

Data simulated under the SSPSC

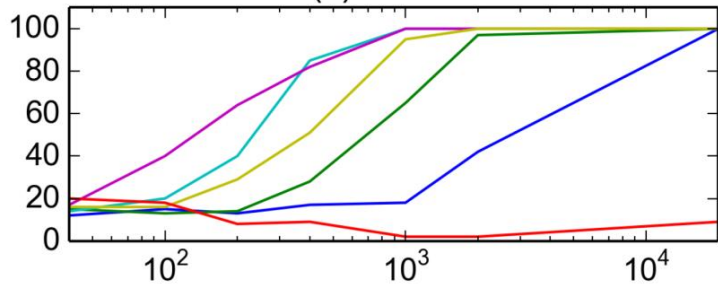
(a) $\alpha = 2$



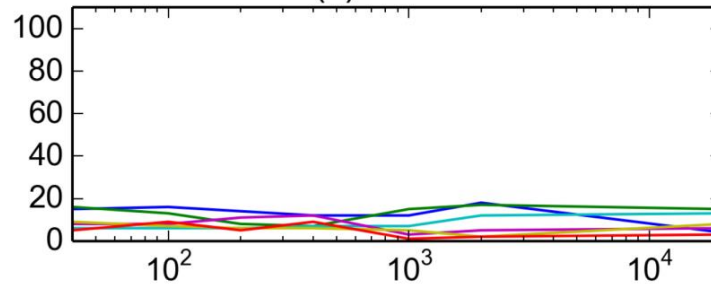
(a) $\alpha = 2$



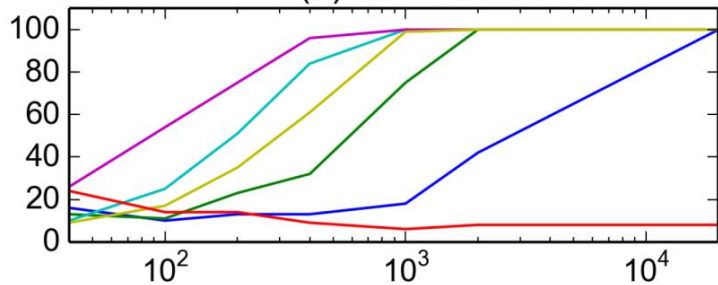
(c) $\alpha = 20$



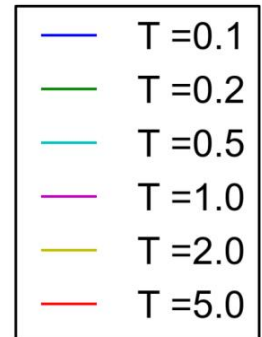
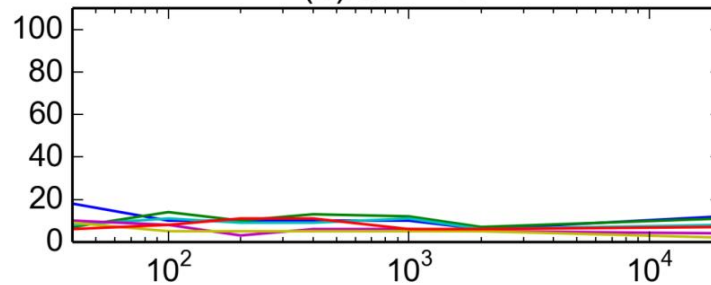
(c) $\alpha = 20$



(e) $\alpha = 100$

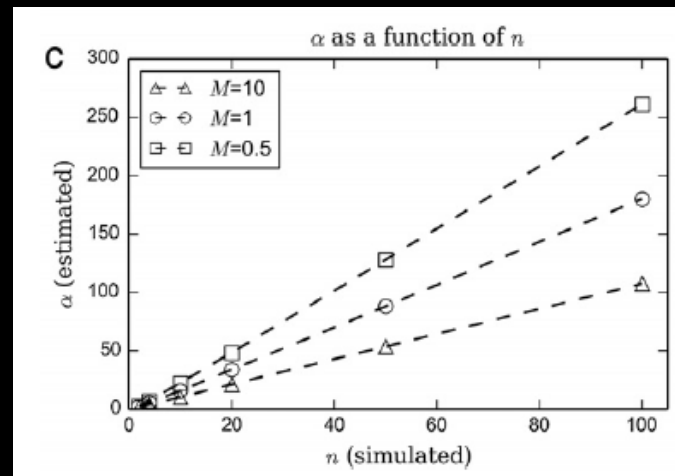


(e) $\alpha = 100$



There is information in genomic data to separate two simple models (pop structure versus pop size change)

But there are “deep” connections between model parameters: the number of islands is “similar” to the population size ratio:



What about more complex models ?

Why Madagascar?



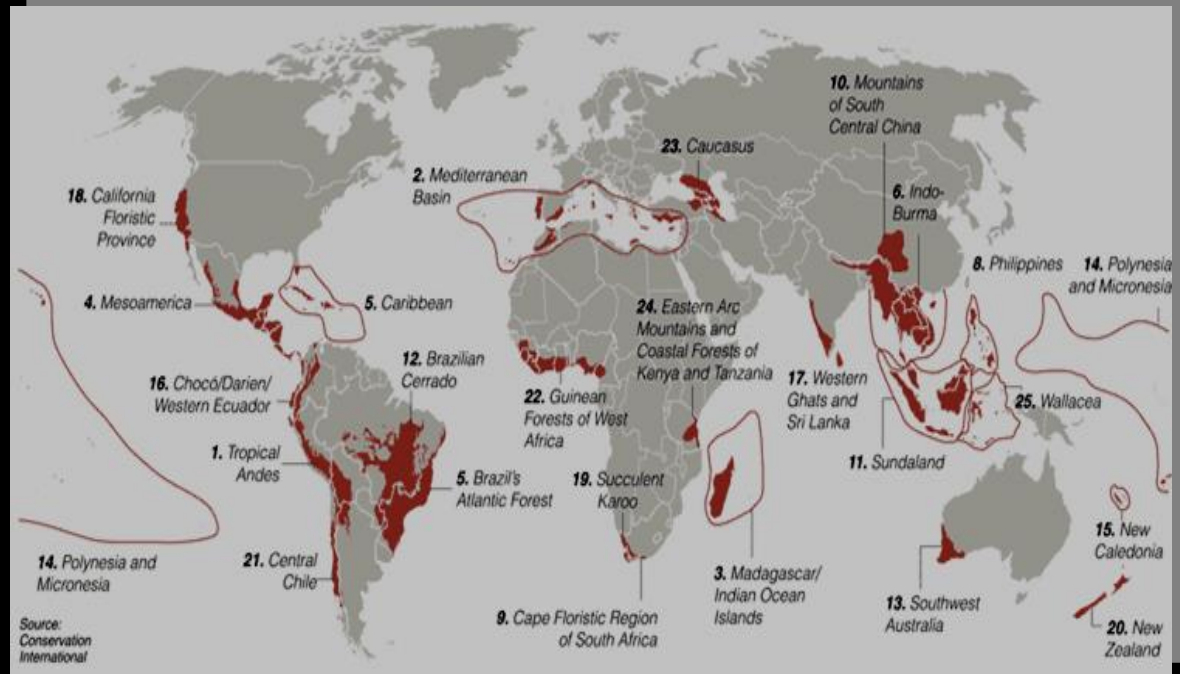
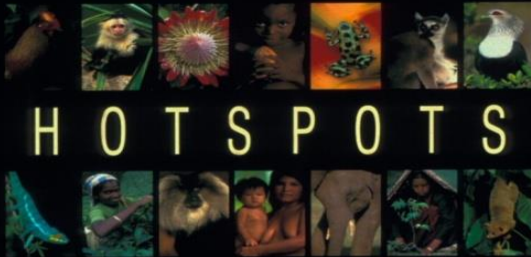
Madagascar and the Indian Ocean Islands Hotspot

HOTSPOTS

Earth's 35

Richest and

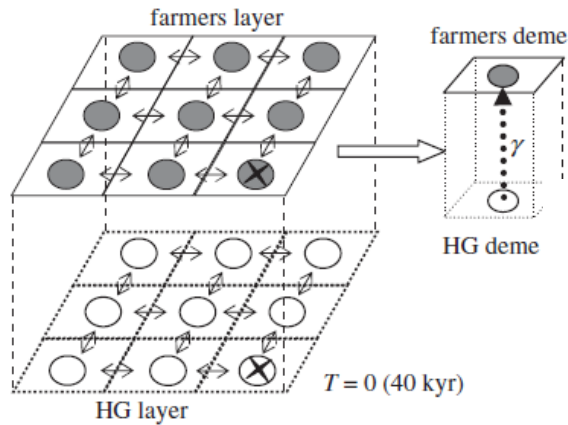
Most Endangered Ecoregions



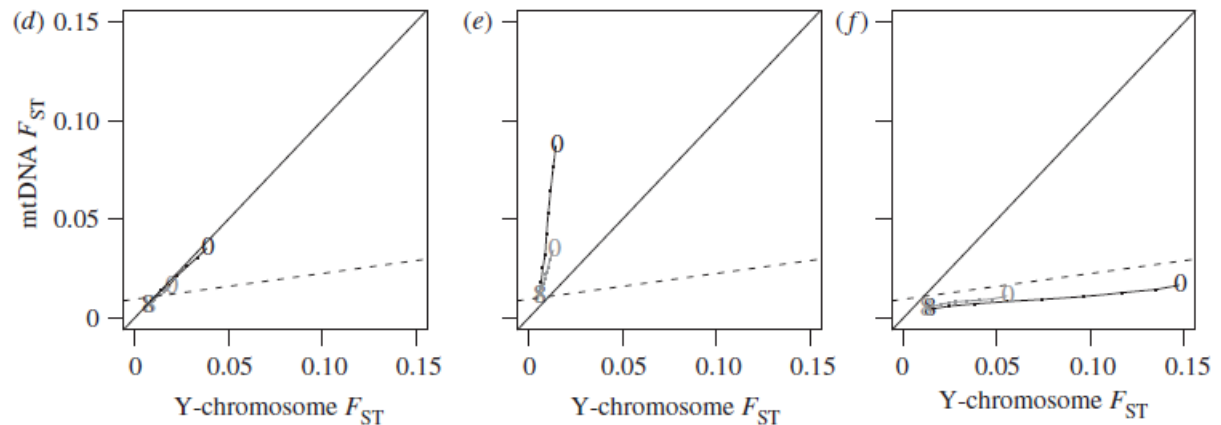
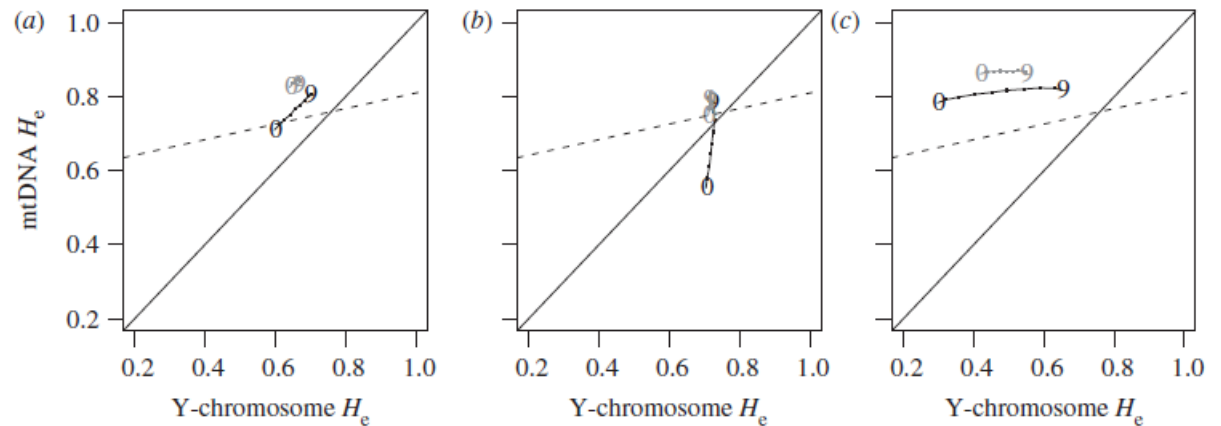
EARTH'S BIOLOGICALLY RICHEST AND MOST ENDANGERED TERRESTRIAL ECOREGIONS
 BURNELL, A. • MOTTUSCHNER, A. • NORDLIE, M. • PEARSON, M. • RYLAND, G. • SODEN, G. • SODEN, G. • SODEN, G.

Investigating sex-biased migration during the Neolithic transition in Europe, using an explicit spatial simulation framework

Rita Rasteiro^{1,*†}, Pierre-Antoine Bouttier^{1,‡}, Vítor C. Sousa^{1,§}
and Lounès Chikhi^{1,2,3,*}



bilocality **matrilocal** **patrilocal**



Future:
Integrating
various mating
systems

Various scenarios of habitat fragmentation

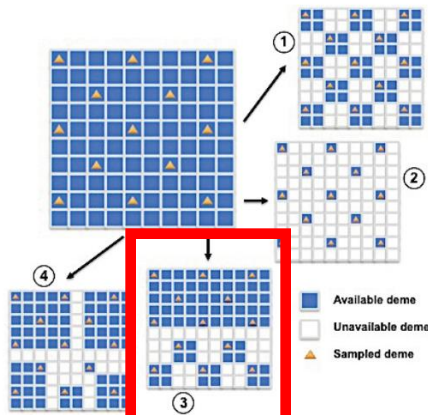


Fig. 2: The different fragmentation scenarios (1 to 4) studied with the 10x10

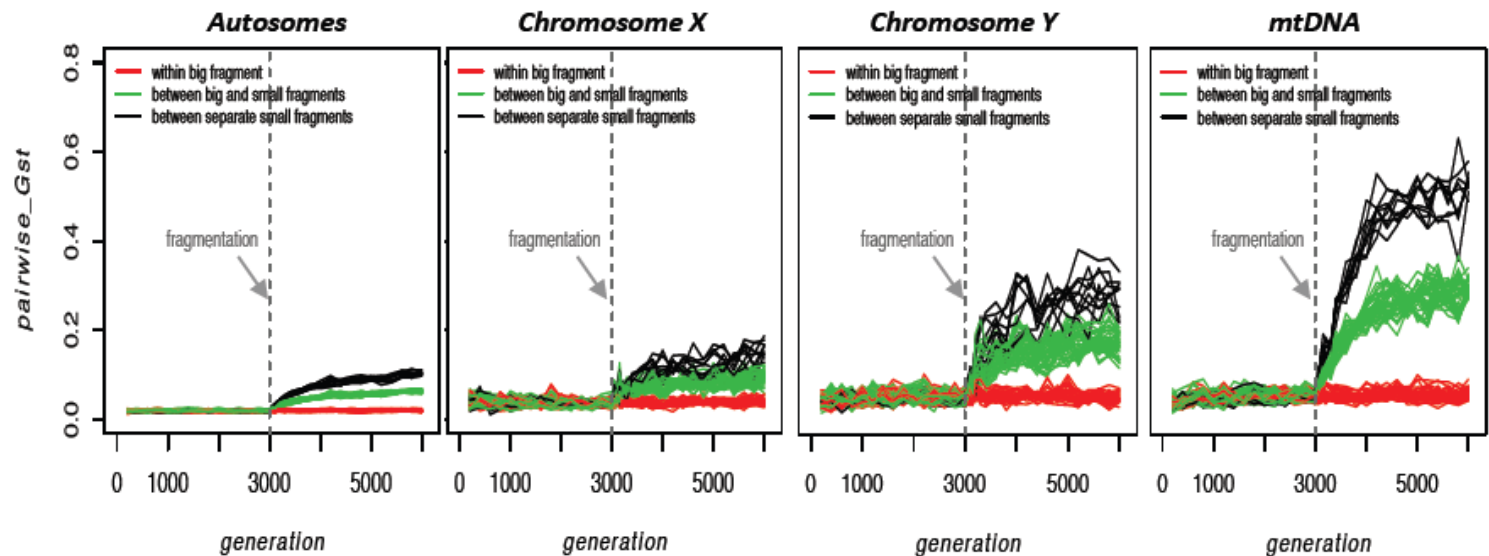
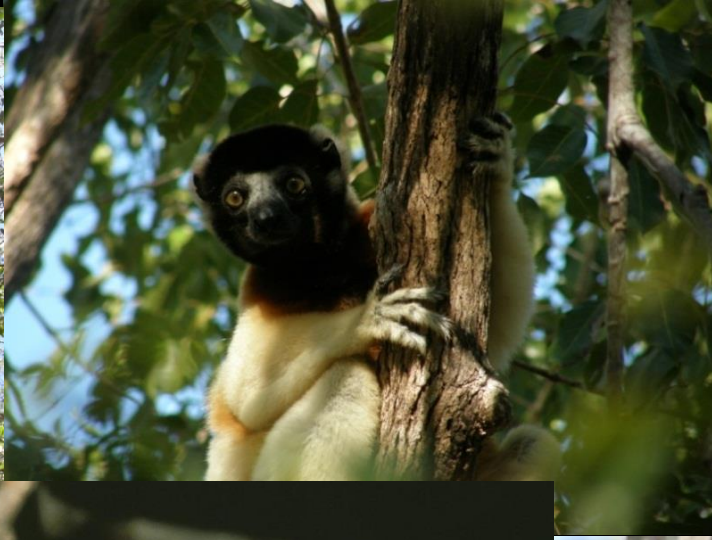
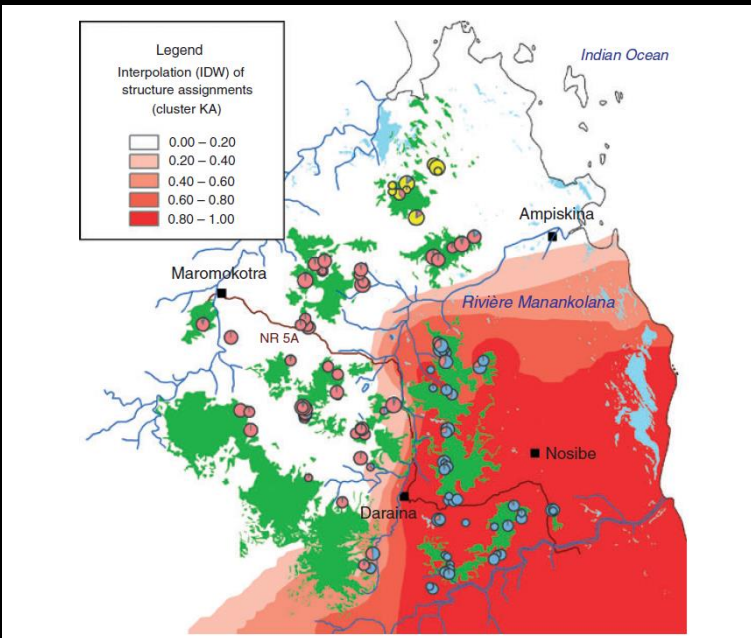
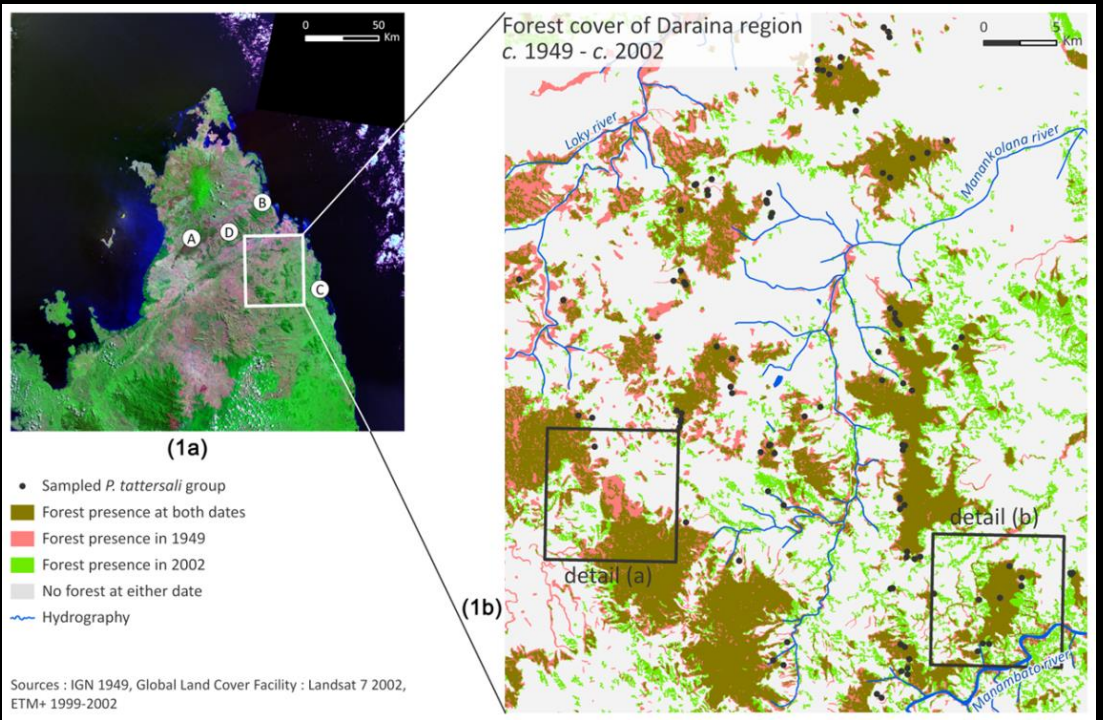


Fig. 10: Evolution of pairwise G_{st} across time with scenario 3 ($m=0.1$, $r=0.4$). Each line corresponds to a couple of demes, and is an average over 10 simulations with the same parameters.



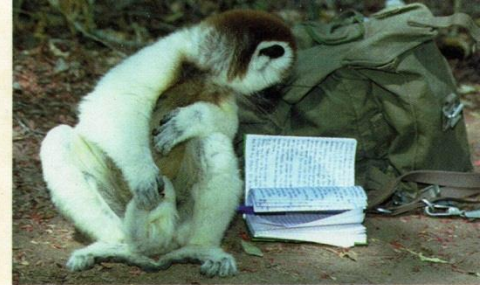
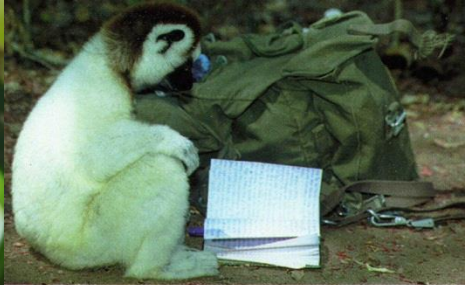


SIFAKAS (Propithecus)





Still much to study



Russ. Mittermeier for these pictures