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Miguel Navascués, Concetta Burgarella. A new composite likelihood method to characterize demographic expansions: preliminary results from Y-chromosome STR data. DNA in Forensics, Università Politecnica delle Marche, May 2008, Ancona, Italy. hal-02755469

**HAL Id: hal-02755469**

**<https://hal.inrae.fr/hal-02755469>**

Submitted on 1 Mar 2021

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# A new composite likelihood method to characterize demographic expansions

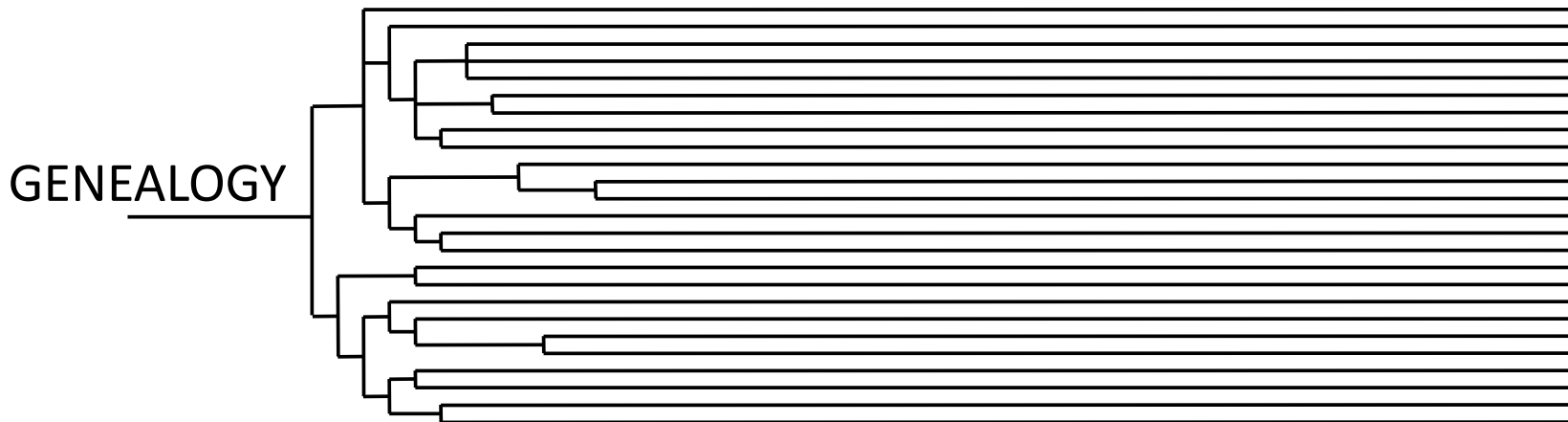
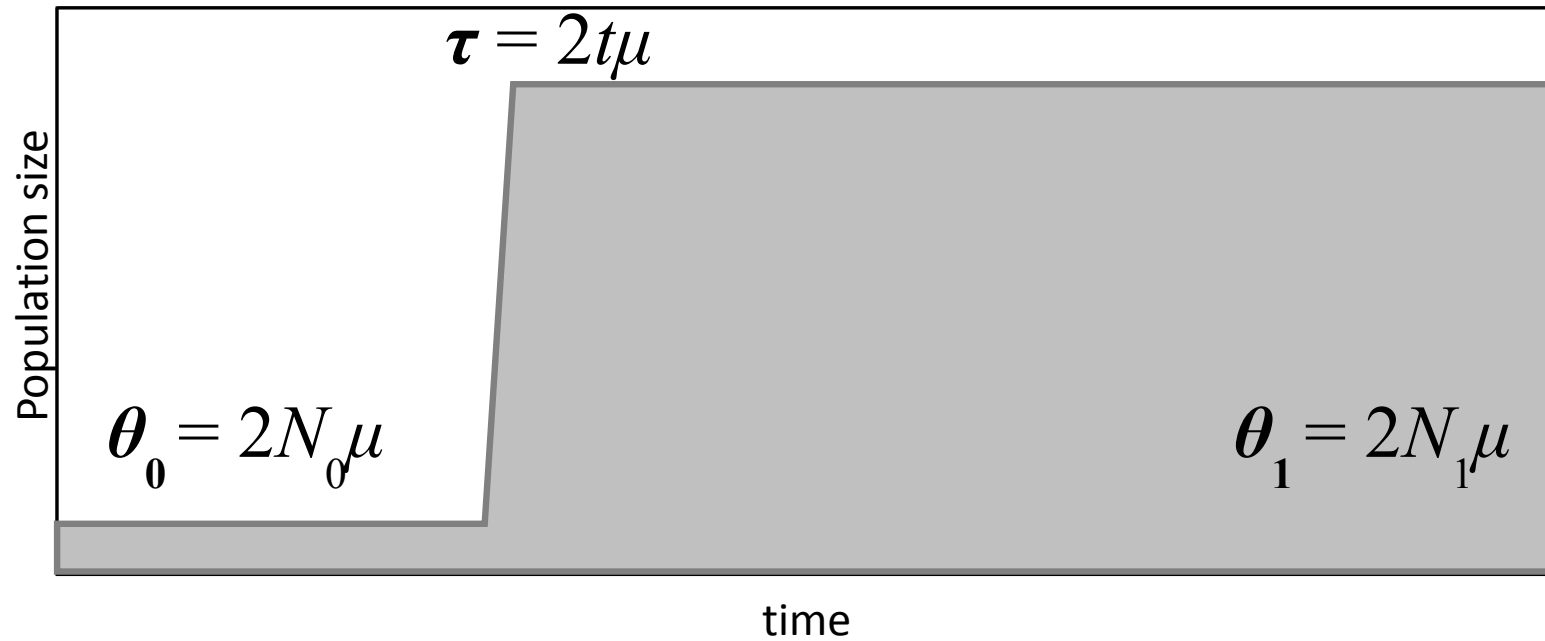
Miguel Navascués & Concetta Burgarella



CNRS UMR 7625 Écologie et Évolution

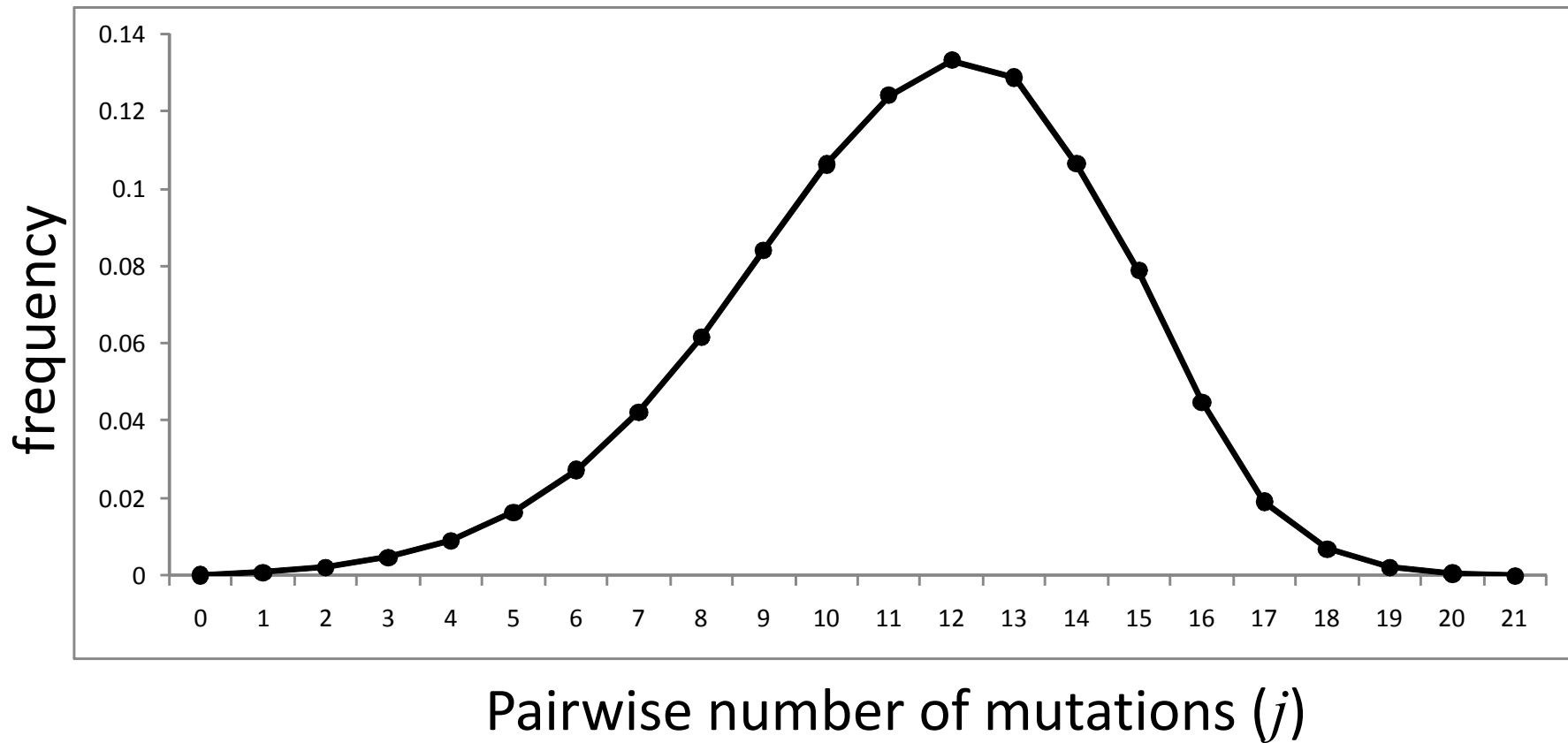
Université Pierre et Marie Curie / École Normale Supérieure

# Demographic expansion



# Mutation distribution

$$P(j; \theta_0, \theta_1, \tau) = P(j; \theta_1) + e^{-\tau \frac{\theta_1 + 1}{\theta_1}} \times \sum_{j'=0}^j \frac{\tau^{j'}}{j'!} [P(j-j'; \theta_0) - P(j-j'; \theta_1)]$$



# Mutations ( $j$ ) $\geq$ Genetic differences ( $i$ )

$$P(i; \theta_0, \theta_1, \tau) = \sum_{j=i}^{\infty} P(j; \theta_0, \theta_1, \tau) \times P(i; j)$$

Mutations ( $j$ )  $\geq$  Genetic differences ( $i$ )

$$P(i; \theta_0, \theta_1, \tau) = \sum_{j=i}^{\infty} P(j; \theta_0, \theta_1, \tau) \times P(i; j)$$

**Distribution of genetic  
differences**

# Mutations ( $j$ ) $\geq$ Genetic differences ( $i$ )

## Distribution of mutations

$$P(i; \theta_0, \theta_1, \tau) = \sum_{j=i}^{\infty} P(j; \theta_0, \theta_1, \tau) \times P(i; j)$$

## Distribution of genetic differences

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Distribution of mutations

$$P(i; \theta_0, \theta_1, \tau) = \sum_{j=i}^{\infty} P(j; \theta_0, \theta_1, \tau) \times P(i; j)$$

Distribution of genetic differences

Mutation model



# Mutational model, $P(i;j)$

## Stepwise Mutation Model

$$P(\delta; j) = \begin{cases} \binom{j}{\frac{j}{2}} \left(\frac{1}{2}\right)^j & \delta=0 \wedge j \text{ even} \\ \binom{j}{\frac{j+2}{2}} \left(\frac{1}{2}\right)^j + \binom{j}{\frac{j-2}{2}} \left(\frac{1}{2}\right)^j & \delta \neq 0 \wedge j \text{ even} \\ 0 & \text{otherwise} \end{cases}$$

## Linked loci with different mutation rates

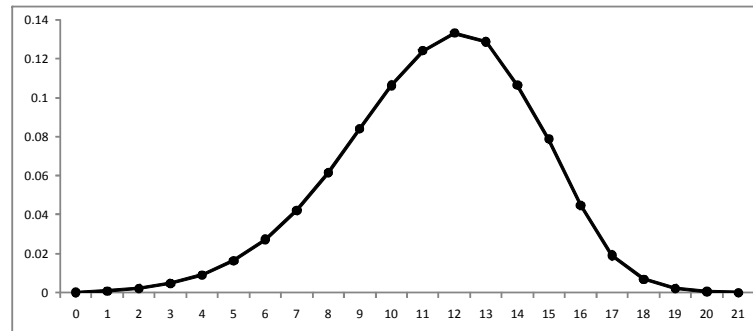
$$P(\{k_1, k_2, \dots, k_L\}; j) = \begin{cases} \frac{j!}{k_1! k_2! \dots k_L!} p_1^2 p_2^2 \dots p_L^2 & \sum_{l=1}^L k_l = j \\ 0 & \text{otherwise} \end{cases}$$

# Estimation of time of expansion

1000 coalescent simulations of demographic expansions

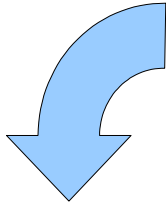
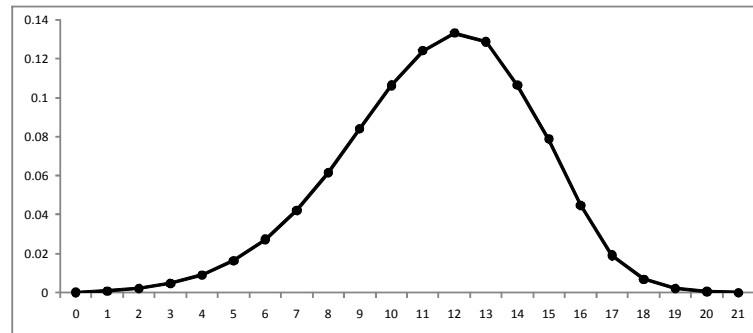
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1000 coalescent simulations of demographic expansions



# Estimation of time of expansion

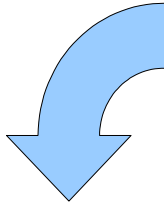
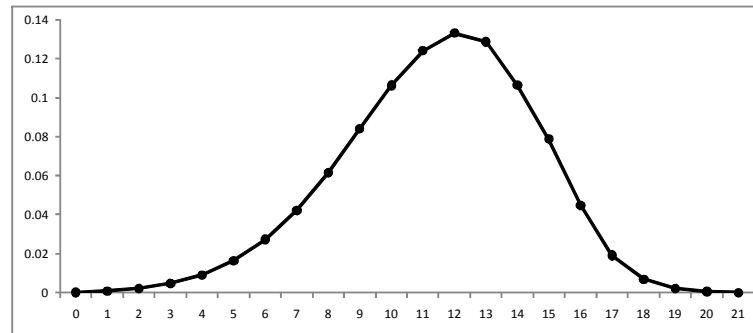
1000 coalescent simulations of demographic expansions



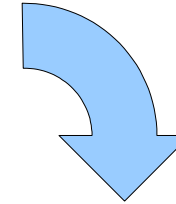
$$P(\textit{mutation}; \theta_0, \theta_1, \tau)$$

# Estimation of time of expansion

1000 coalescent simulations of demographic expansions



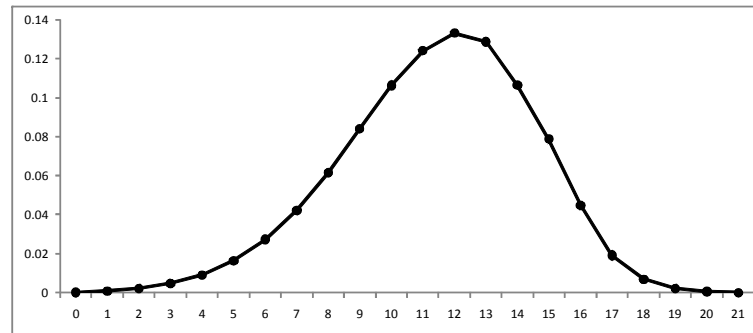
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$$P(\textit{genetic differences}; \theta_0, \theta_1, \tau)$$

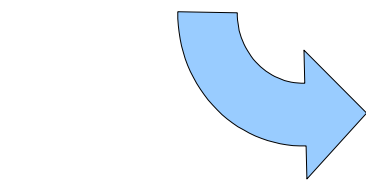
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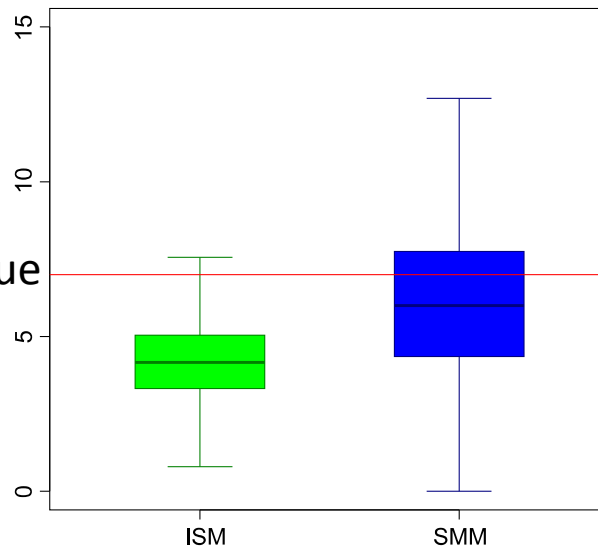


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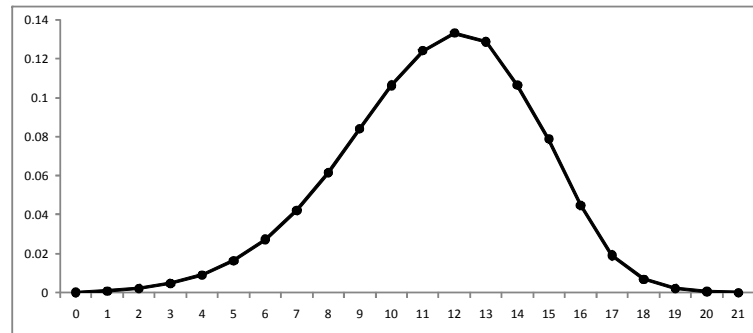
True (simulated) value



1000 estimates of the time of expansion

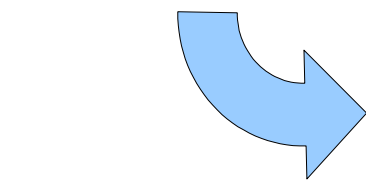
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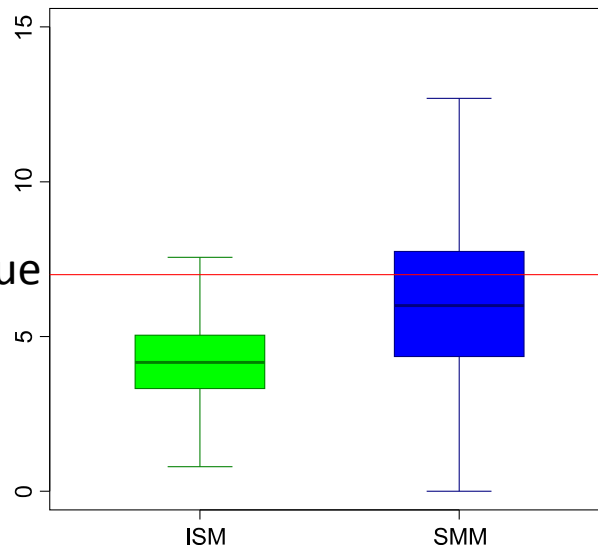


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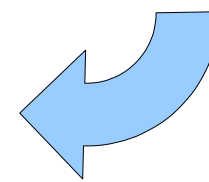
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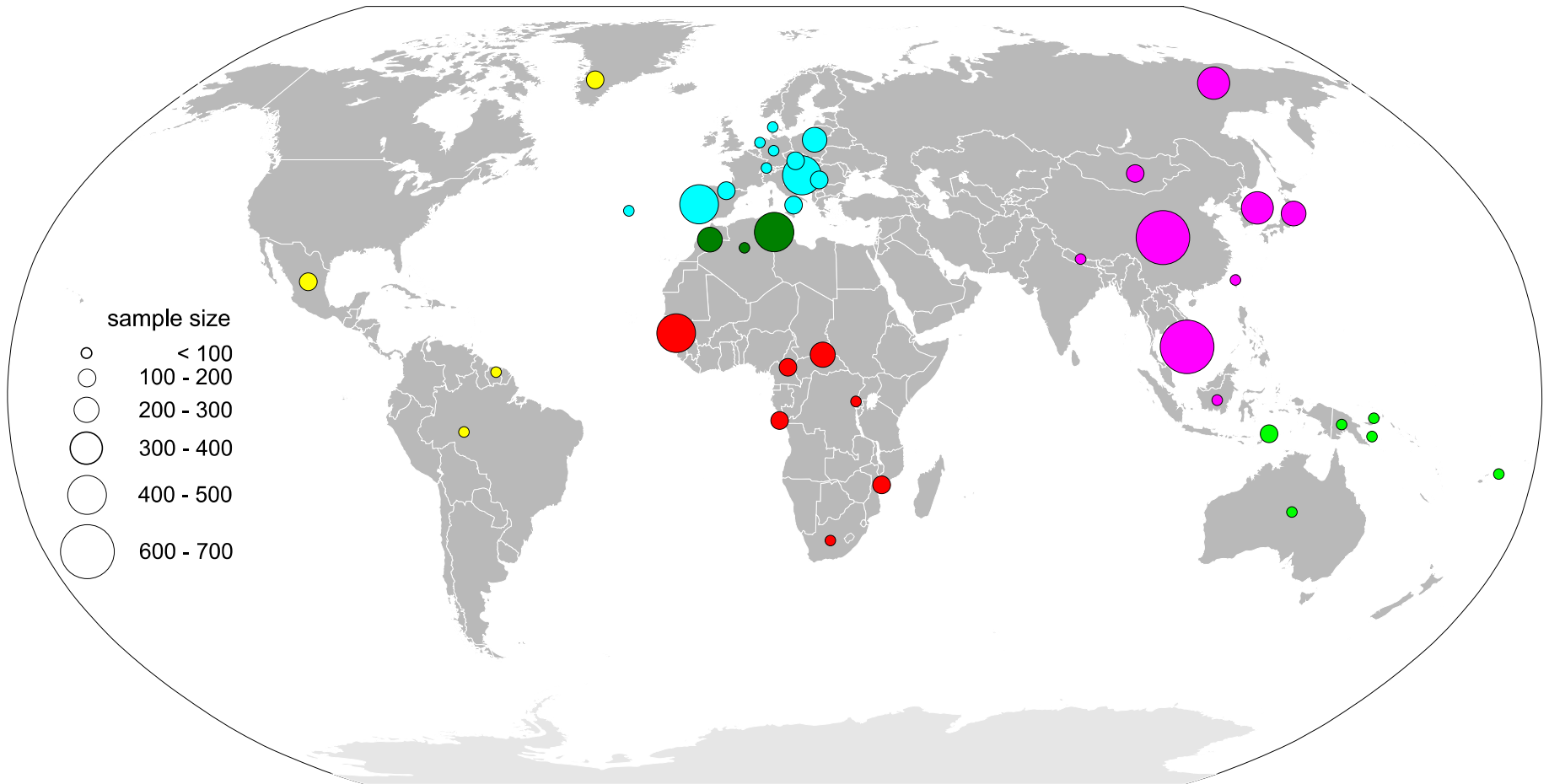
1000 estimates of the time of expansion



Using a mutational model yields less biased estimates

# Y-STRs population samples

DYS19, DYS390, DYS391, DYS392 and DYS393



groups following Li *et al.* (2008) *Science*





# Time of expansion estimates

Group	# individuals	# populations	$\hat{t}$ (95%CI)	$\hat{t}$ years (95%CI) <sup>b</sup>
Sub-Saharan-Africa	1183	23	12.53 (1.29-26.18)	17612 (2672-36811)
N-Africa	694	13	13.69 (0.10-24.51)	19247 (441-34463)
Europe	2150	14	9.40 (5.00-12.81)	13213 (7030-18012)
East-Asia	2621	16	11.51 (8.18-15.75)	16182 (11502-22146)
Oceania	293	7	12.04 (6.76-17.45)	16924 (9505-23552)
America	422	8	8.70 (5.10-15.42)	12231 (7171-21682)
World <sup>a</sup>	9182	94	12.85 (9.25-14.41)	18061 (13006-20262)

<sup>a</sup> Includes samples not classified in previous groups

<sup>b</sup> mutation rate per generation over the five loci,  $\mu = 0.89 \times 10^{-2}$  (www.ystr.org); generation time: 25 years

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Approximate Bayesian Computation:

Pritchard *et al.* (1999) *Mol Biol Evol* (sample size 445)  $t=18,000$  95%HPD (7,000-41,000)

Bayesian Coalescent MCMC:

Wilson *et al.* (2003) *J Roy Stat Soc A* (sample size 115)  $t=14,000$  90%HPD (6,800-28,000)

# Conclusion

- Analytical description of mutational process for linked microsatellites
- This model can be applied in statistical analysis:
  - **Estimation of demographic expansion**
  - *Neutrality test*
  - *Identity by descent/identity in state probabilities*
- Example data set: estimates comparable to those of previous studies (using other statistics and data)

# Thanks

Olivier Hardy (Universite Libre de Bruxelles)  
Frantz Depaulis (CNRS)

Funding:



MAIRIE DE PARIS 

## Data:

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