A new composite likelihood method to characterize demographic expansions: preliminary results from Y-chromosome STR data

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A new composite likelihood method to characterize demographic expansions

Miguel Navascués & Concetta Burgarella

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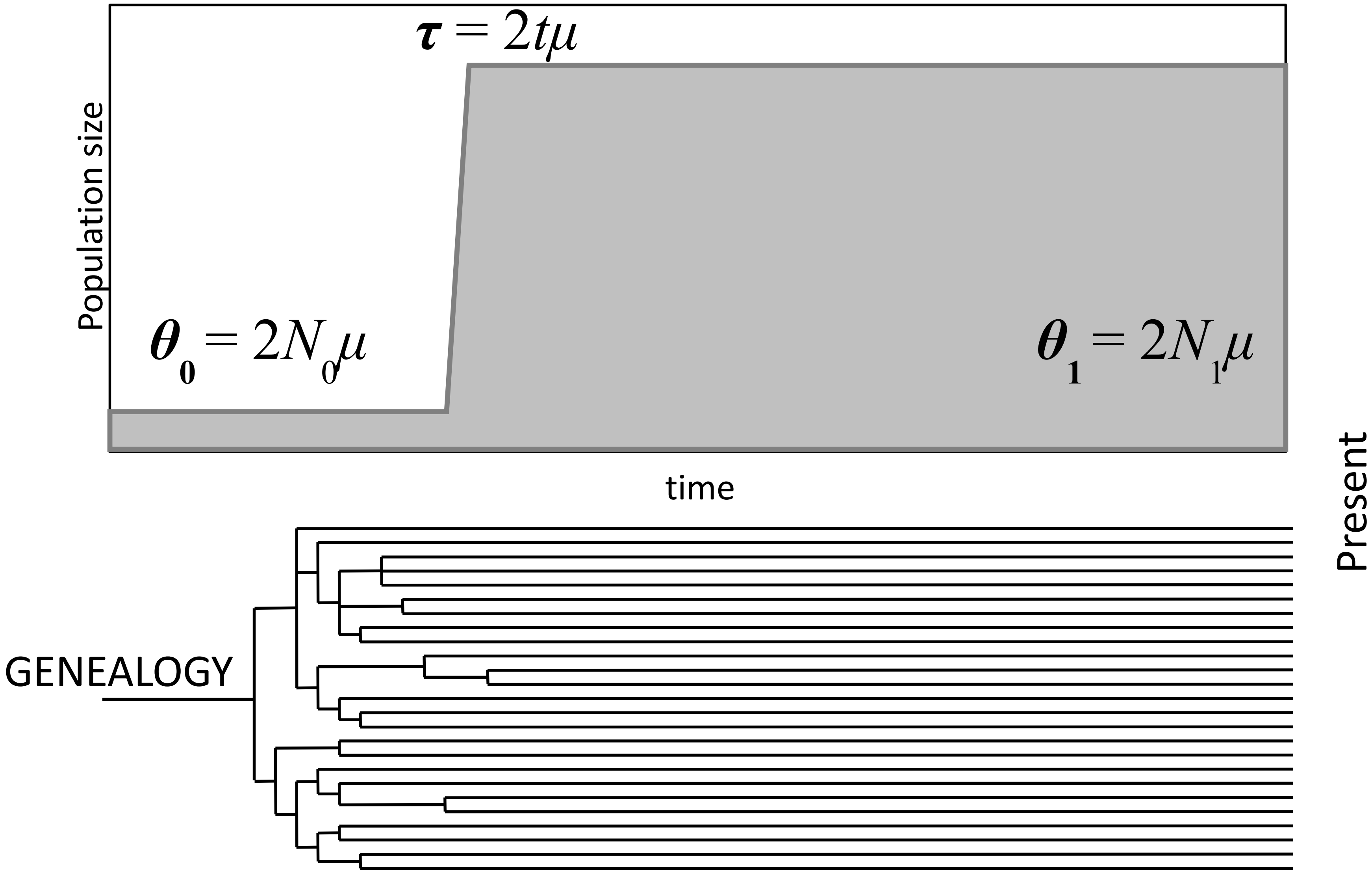
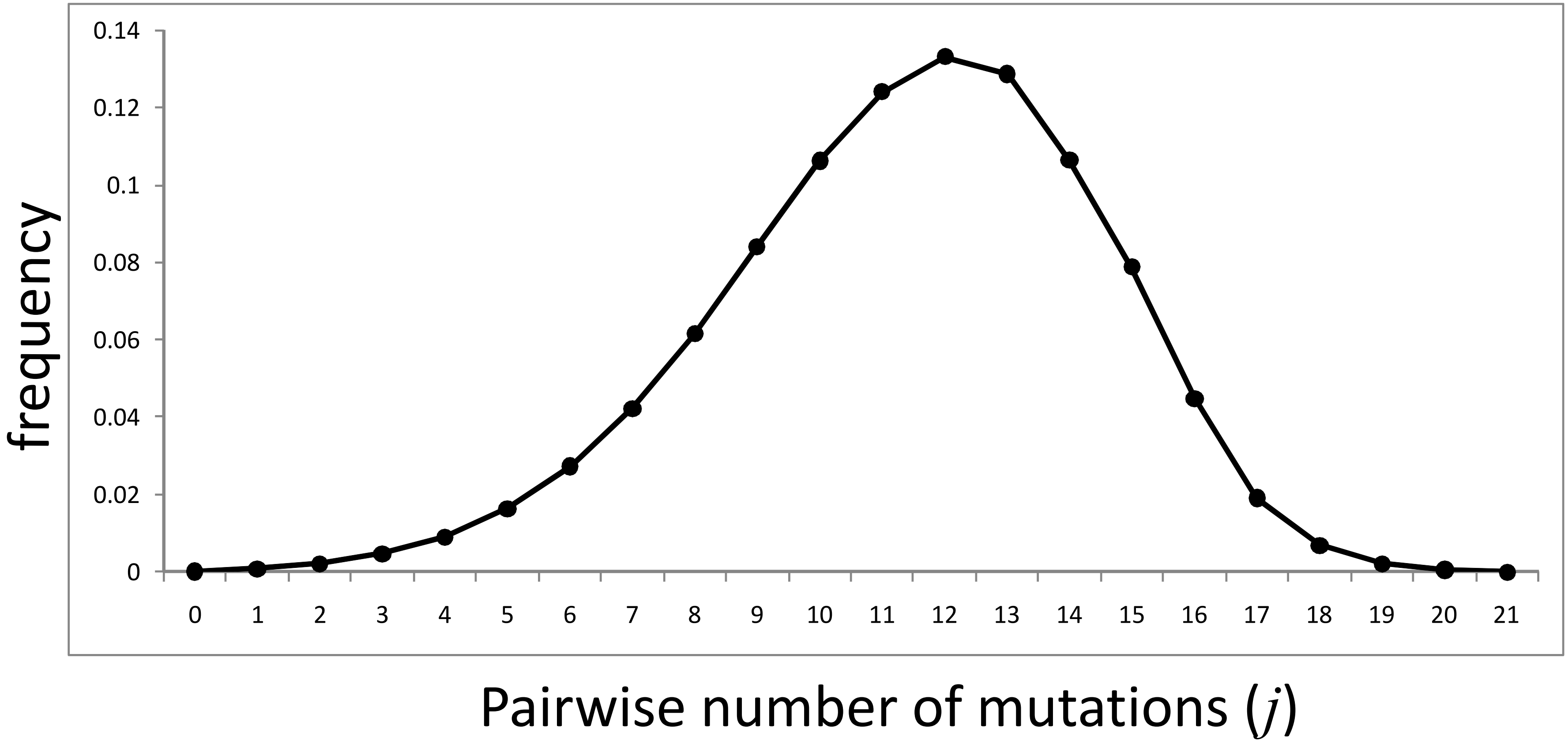
Université Pierre et Marie Curie / École Normale Supérieure

Demographic expansion



Mutation distribution

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



Li (1977) *Genetics // *Slatkin & Hudson (1991) *Genetics*

Mutations (j) ≥ Genetic differences (i)

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

Schneider & Excoffier (1999) *Genetics*

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Distribution of genetic differences

Schneider & Excoffier (1999) *Genetics*

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Distribution of genetic differences

Mutation model

Schneider & Excoffier (1999) *Genetics*

Mutational model, $P(i;j)$

Stepwise Mutation Model

$$P(\delta;j)=\begin{cases}\binom{j}{\frac{j}{2}}\left(\frac{1}{2}\right)^{j} & \delta=0\wedge j\;even\\ \binom{j}{\frac{j+2}{2}}\left(\frac{1}{2}\right)^{j}+\binom{j}{\frac{j-2}{2}}\left(\frac{1}{2}\right)^{j} & \delta\neq0\wedge j\;even\\ 0 & otherwise\end{cases}$$

Linked loci with different mutation rates

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

Estimation of time of expansion

1000 coalescent simulations of demographic expansions

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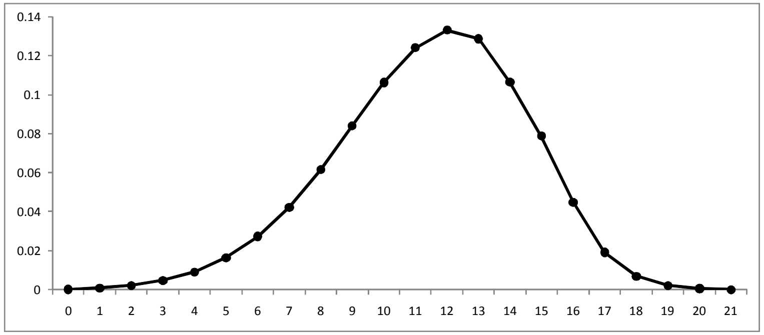
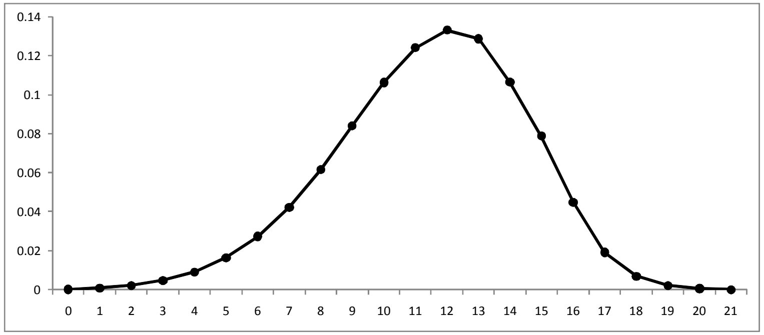
1000 coalescent simulations of demographic expansions



Estimation of time of expansion

1000 coalescent simulations of demographic expansions



$$P(mutation\,;\theta_0,\theta_1,\tau)$$

Estimation of time of expansion

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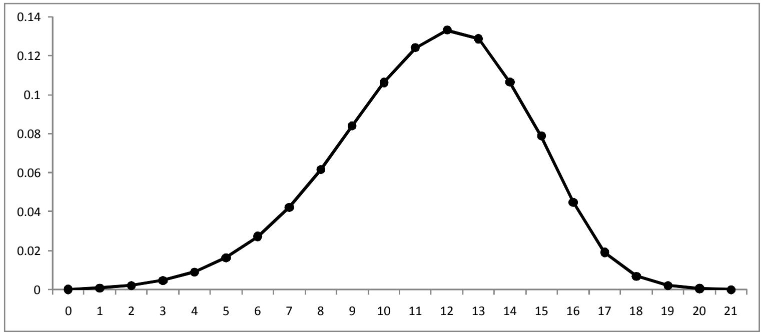
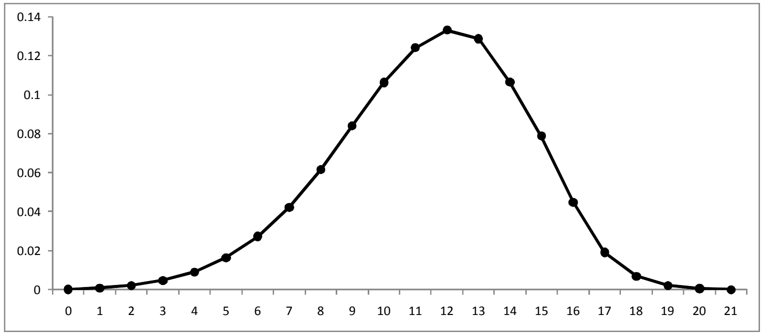


$$\boxed{P(mutation\,;\theta_0,\theta_1,\tau)}$$

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

Estimation of time of expansion

1000 coalescent simulations of demographic expansions



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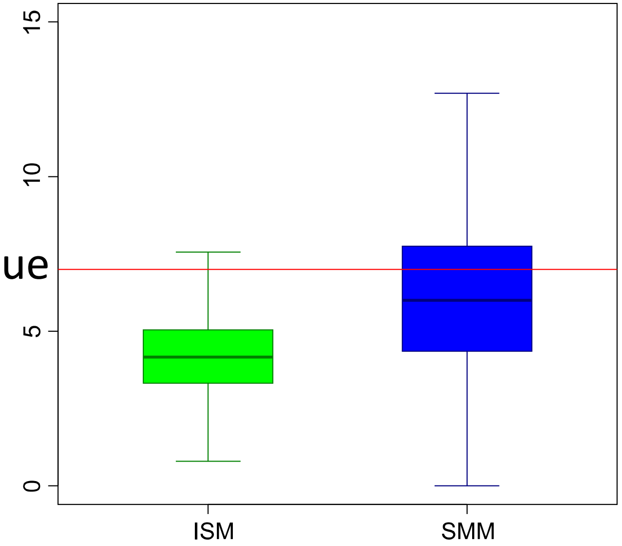
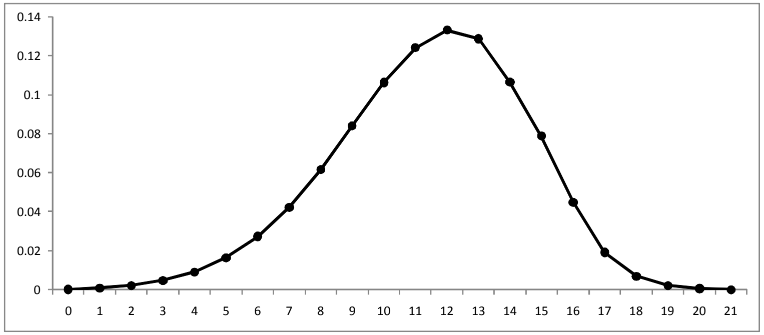


True (simulated) value

1000 estimates of the time of expansion

Estimation of time of expansion

1000 coalescent simulations of demographic expansions



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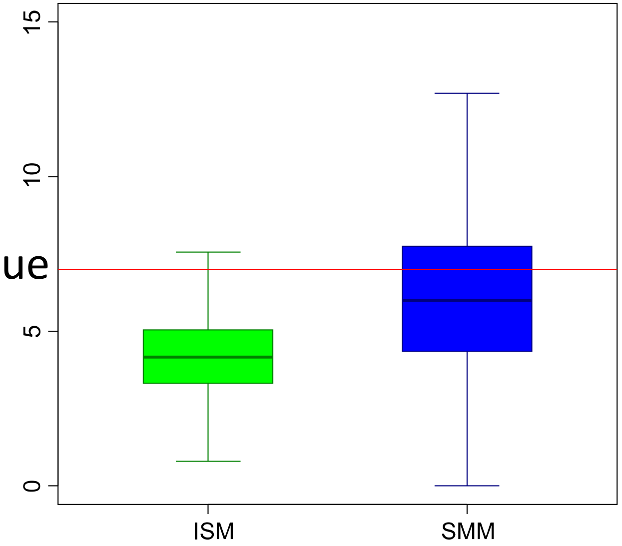


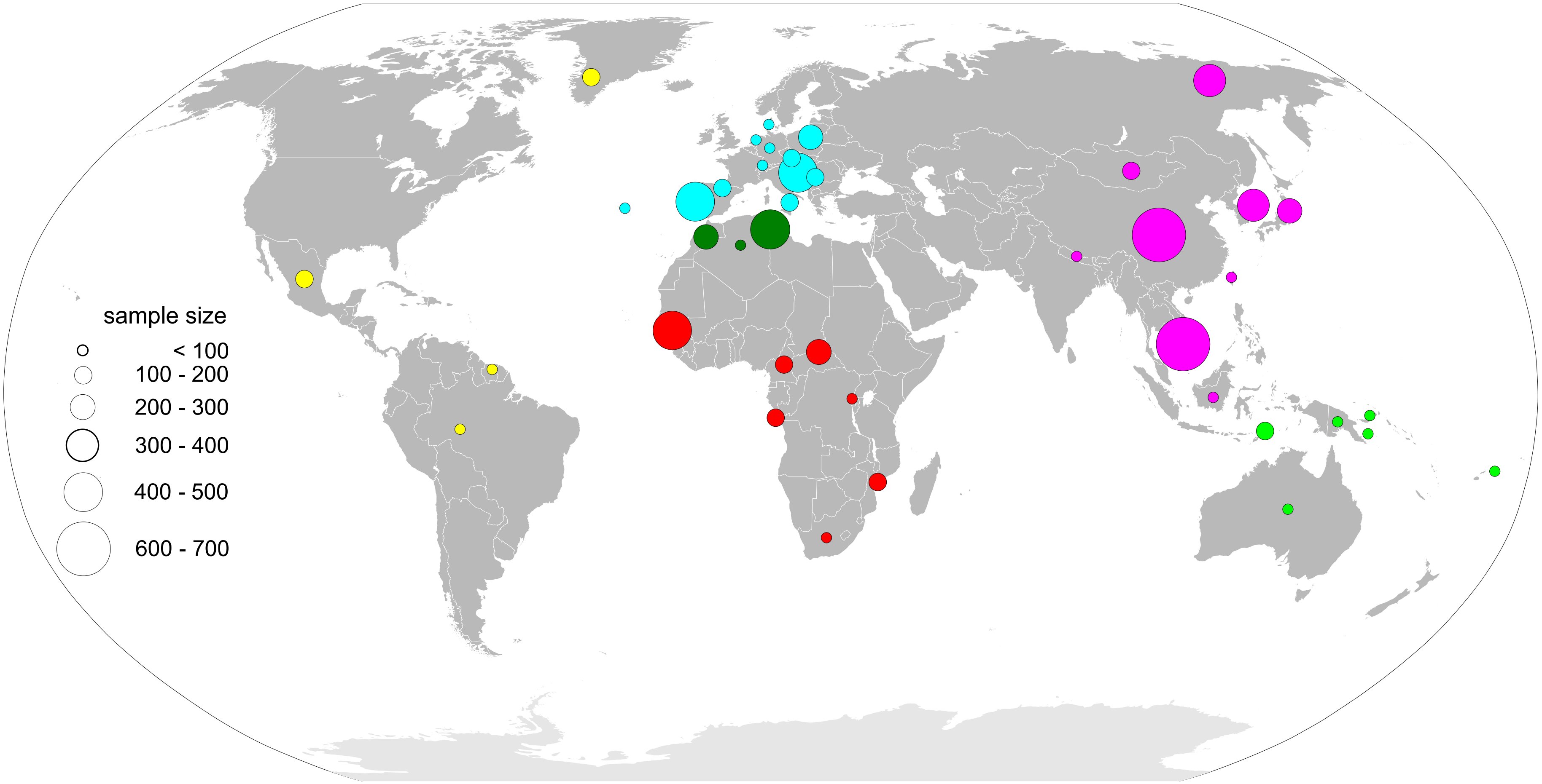
True (simulated) value

1000 estimates of the time of expansion

Using a mutational model yields less biased estimates

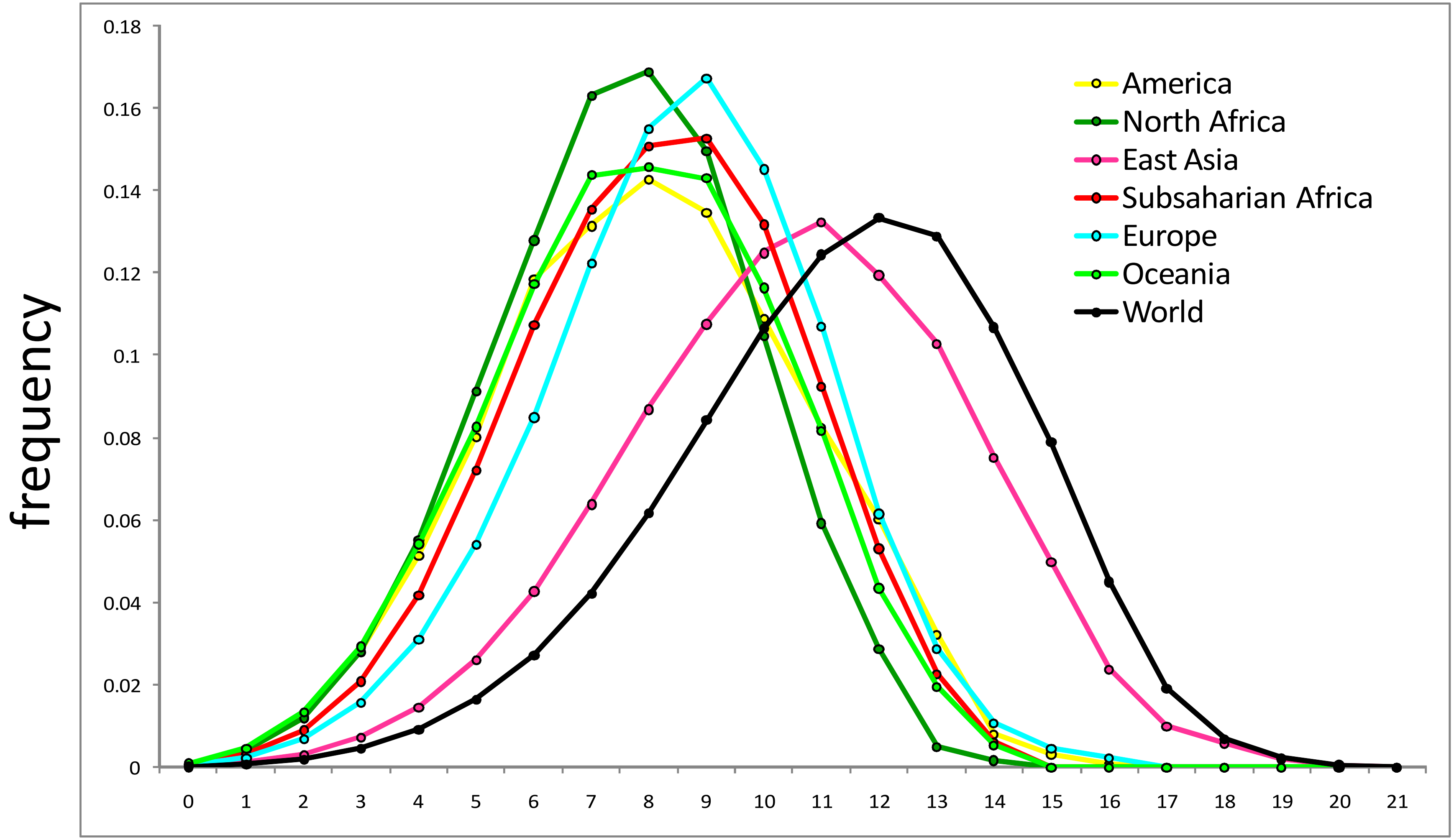
Y-STRs population samples

DYS19, DYS390, DYS391, DYS392 and DYS393



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

Genetic distance distribution



Pairwise difference in number of repeats

Time of expansion estimates

Group	# individuals	# populations	$\hat{\tau}$ (95%CI)	$\hat{t}$ years (95%CI)[b]
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[a]	9182	94	12.85 (9.25-14.41)	18061 (13006-20262)

[a] Includes samples not classified in previous groups

[b] 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 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)

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Data:

Alves et al. 2003. *Forensic Sci. Int.* **134**(2-3): 126-133.
Alves et al. 2007. *Forensic Sci. Int.* **171**(2-3): 250-255.
Ayadi et al. 2006. *Forensic Sci. Int.* **164**(2-3): 249-253.
Barac, et al. 2003. *Forensic Sci. Int.* **138**(1-3): 127-133.
Barrot et al. 2007. *Forensic Sci. Int.* **168**(1), e10-e12.
Berger et al. 2003. *Forensic Sci. Int.* **137**(2-3): 221-230.
Bosch et al. 2000. *Int. J. Legal Med.* **114**(1): 36-40.
Bosch et al. 2003. *Forensic Sci. Int.* **132**(3): 228-232.
Brandt-Casadevall et al. 2003. *Forensic Sci. Int.* **135**(3): 247-250.
Cagliá et al. 2003. *Hum. Biol.* **75**(3): 313-330.
Carvalho et al. 2003. *Forensic Sci. Int.* **134**(1): 29-35.
Chang et al. 2007. *Forensic Sci. Int.* **167**(1): 70-76.
Cherni et al. 2005. *Forensic Sci. Int.* **152**(1): 95-99.
Coia et al. 2004. *Am. J. Hum. Genet.* **16**(1): 57-67.
Destro Bisol et al. 2004. *Mol. Biol. Evol.* **21**(9): 1673-1682.
Frigi et al. 2006. *Forensic Sci. Int.* **160**(1): 80-83.
Foster et al. 1998. *Mol. Biol. Evol.* **15**(9): 1108-1114.
Garcia et al. 2004. *Forensic Sci. Int.* **145**(1): 65-68.
Hu 2006. *Forensic Sci. Int.* **158**(1): 80-85.
Kayser et al. 2001. *Am. J. Hum. Genet.* **68**(4): 990-1018.
Khodjet el Khil et al. 2005. *Forensic Sci. Int.* **148**(2-3): 211-218.
Kumagai et al. 2007. *Forensic Sci. Int.* **172**(1): 72-78.
Leat et al. 2007. *Forensic Sci. Int.* **168**(2-3): 154-161.
Lecerf et al. 2007. *Forensic Sci. Int.* **171**(2-3): 212-215.
Lessig et al. 2006. *Forensic Sci. Int.* **159**(1): 71-76.
Li et al. 2007. *Forensic Sci. Int.* **172**(1): 79-83.
Park et al. 2005. *Forensic Sci. Int.* **152**(2-3): 133-147.
Pereira et al. 2002. *Ann. Hum. Genet*. **66**: 369-378
Quintana-Murci et al. 2004. *Forensic Sci. Int.* **140**(1): 113-115.
Rosa et al. 2007. *BMC Evolutionary Biology* **7**(1): 124.
Soltyszewski et al. 2007. *Forensic Sci. Int.* **168:** 61-67.
Souto et al. 2006. *Forensic Sci. Int.* **156**: 261-265.
Trovoada et al. 2001. *Ann. Hum. Genet.* **65**(3): 271-283.
Veselinovic et al. 2008. *Forensic Sci. Int.* **176**(2-3): 23-28.
Woźniak et al. 2006. *Forensic Sci. Int.* **164**: 271-275.
Xin et al. 2008. *Forensic Sci. Int.* **174**(2-3): 244-248.
Zhang et al. 2008. *Forensic Sci. Int.* **175**(2-3): 244-249.
Zhu et al. 2005. *Forensic Sci. Int.* **153**: 260-263.