LD-based estimation of effective population size for two African cattle breeds

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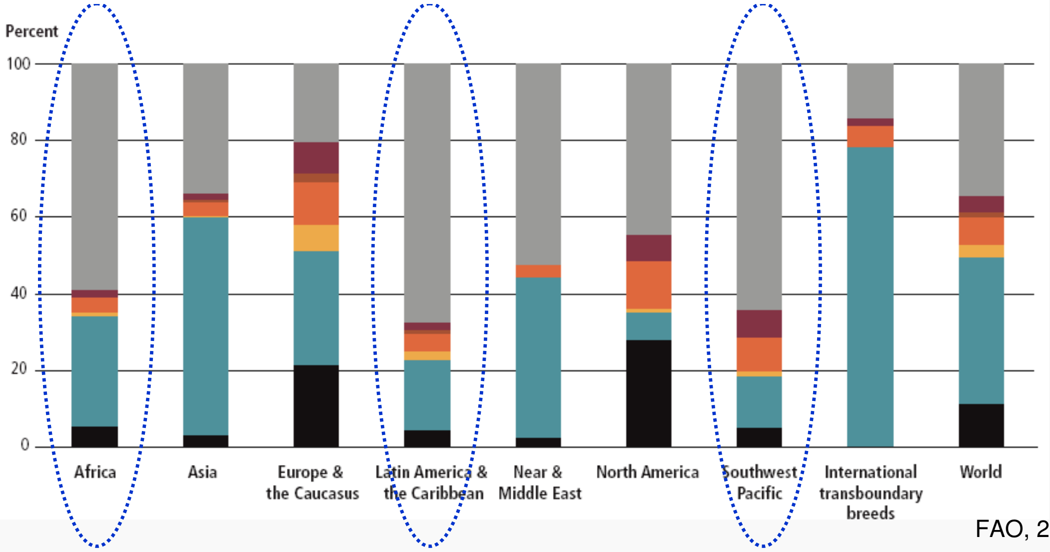
Introduction

> 60% of the breeds have unknown risk status



FAO, 2007

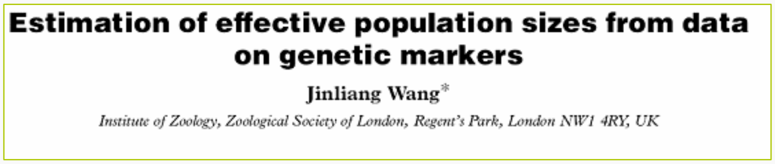
Introduction

$$\boxed{E(r^2) = \frac{1}{(1+4N_e c)}}$$

(Sved, 1971)

$E(r^2)$:	*Expectation of association between two loci*
c:	*Genetic distance between loci in Morgan*
N_e:	*Effective population size.*

Introduction



Estimation of effective population sizes from data on genetic markers

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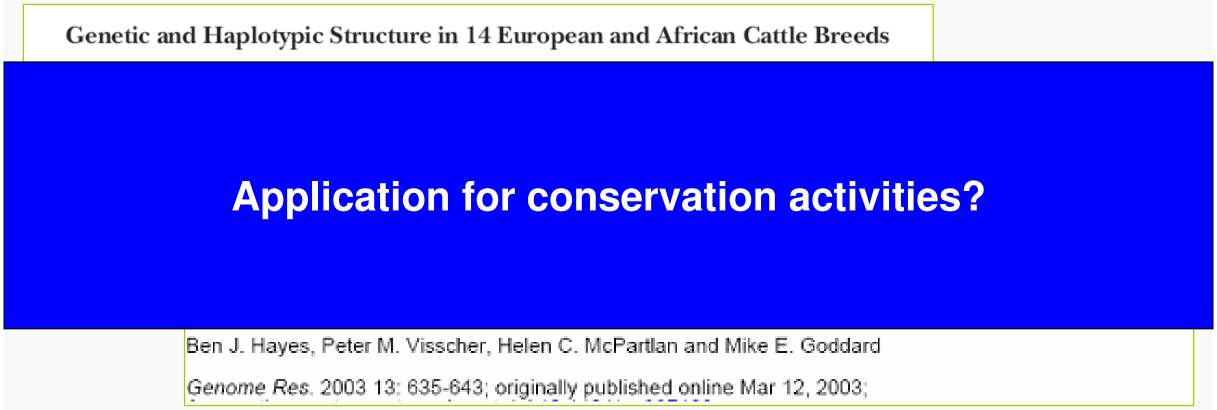
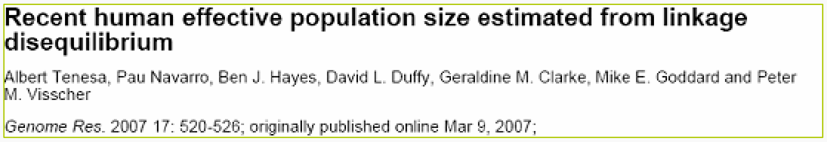


Genetic and Haplotypic Structure in 14 European and African Cattle Breeds

Application for conservation activities?

Ben J. Hayes, Peter M. Visscher, Helen C. McPartlan and Mike E. Goddard

Genome Res. 2003 13: 635-643; originally published online Mar 12, 2003;



Recent human effective population size estimated from linkage disequilibrium

Albert Tenesa, Pau Navarro, Ben J. Hayes, David L. Duffy, Geraldine M. Clarke, Mike E. Goddard and Peter M. Visscher

Genome Res. 2007 17: 520-526; originally published online Mar 9, 2007;

Application to real data

Sheko breed (N=17)
Ethiopia


Photo: A. Workneh


Photo: O. Hanotte

N‘Dama breed (N=22)
Guinea

Application to real data

Samples of the two breeds were genotyped within the bovine HapMap consortium.

8 Chromosomes (2, 4, 6, 7, 9, 12, 20 and 26);
in **total 9140 SNPs**

Exclusion of SNPs:
- non variable
- MAF .lt. 0.1
- not in HWE
- >25% missing genotypes.

Application to real data

1. **Filtering and LD-measures**: Haploview (Barrett et al. 2005)
2. **Recombination rates**: PHASE 2.1 (Li & Stephens 2003; Crawford et al. 2004)
3. **Estimation of effective population size**
   - 1Mb ~ 0.01Morgan vs. genetic distance

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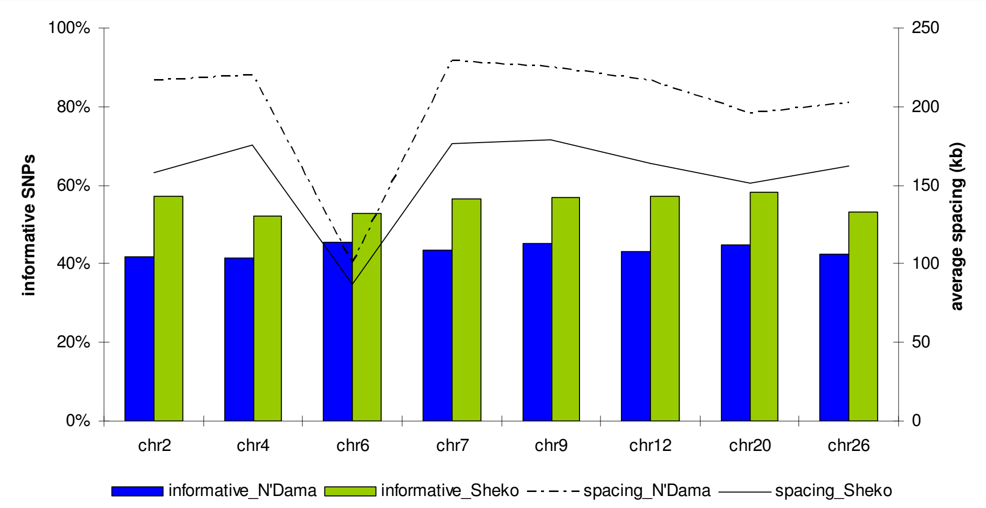
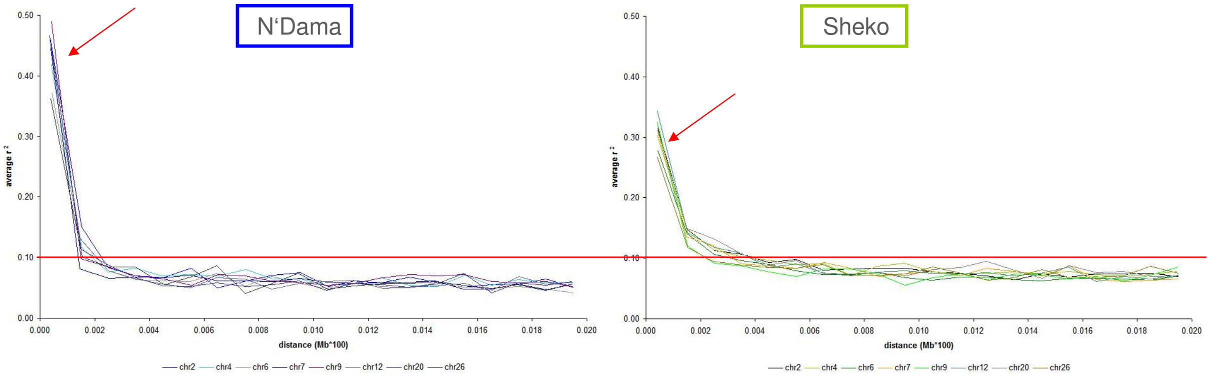
Application to real data

1. Filtering and LD-measures:



Application to real data

1. Filtering and LD-measures: General LD

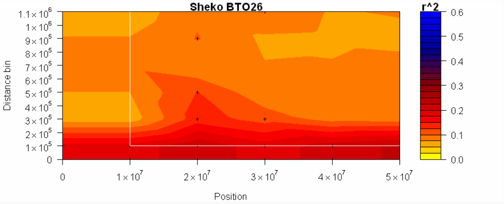


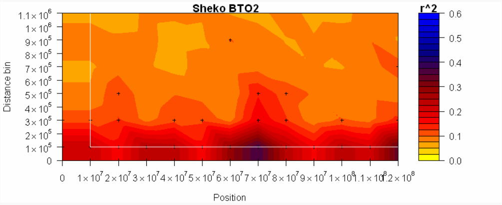
Application to real data

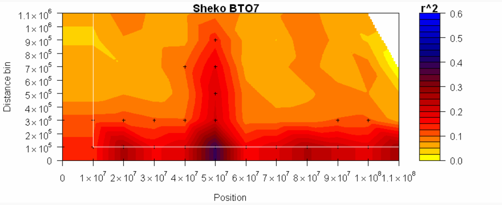
1. Filtering and LD-measures: Regional LD

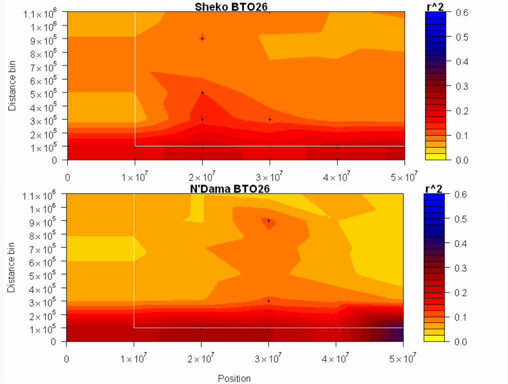
Sliding windows 20Mb (50% overlap)
Bins in 200kb intervals (0-200kb,... 1000-1200kb)

➔ Chromosome-specific patterns within and between breeds?



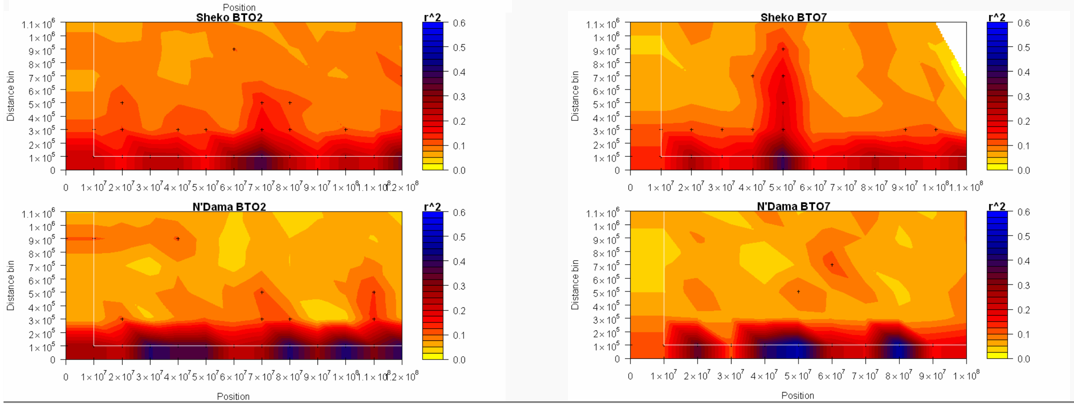






Differences!

Pattern in LD along chromosomes varies between chromosomes and between breeds.



Application to real data

1. **Filtering and LD-measures**: Haploview (Barrett et al. 2005)

2. **Recombination rates**: PHASE 2.1 (Li & Stephens 2003; Crawford et al. 2004)

3. **Estimation of effective population size**
   - 1Mb ~ 0.01Morgan vs. genetic distance

Application to real data

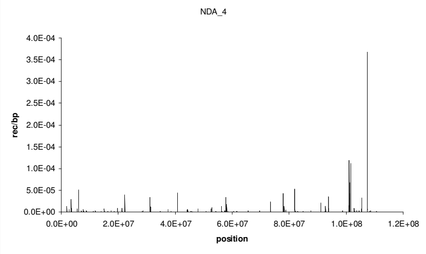
2. Recombination rates

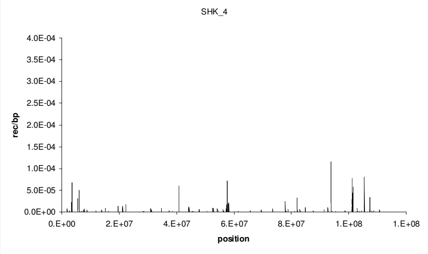
Application to real data

N‘Dama

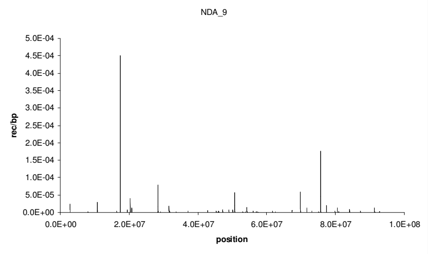
Sheko

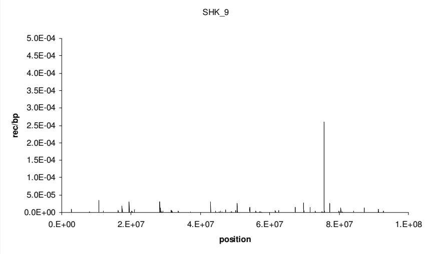
Chr 4





Chr 9





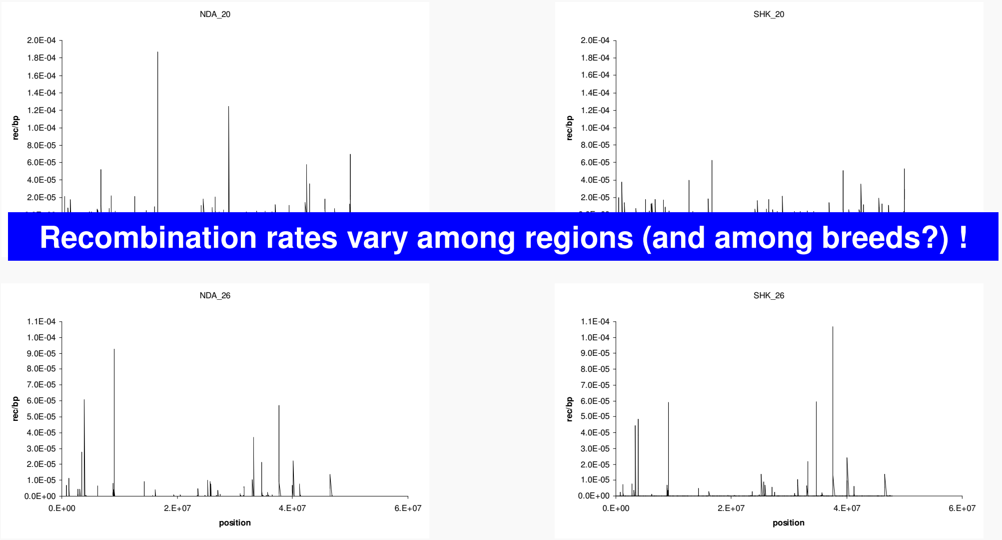
Application to real data

N'Dama

Sheko

Chr 20

Chr 26



Recombination rates vary among regions (and among breeds?) !

Application to real data

1. **Filtering and LD-measures**: Haploview (Barrett et al. 2005)

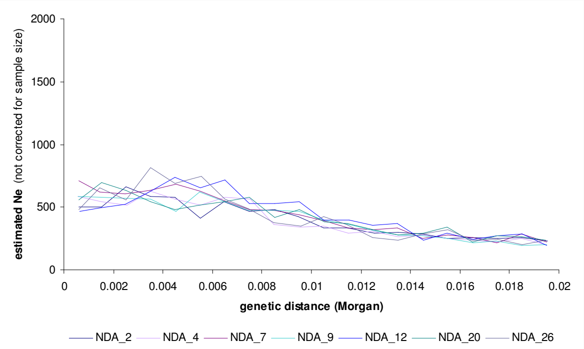
2. **Recombination rates**: PHASE 2.1 (Li & Stephens 2003; Crawford et al. 2004)

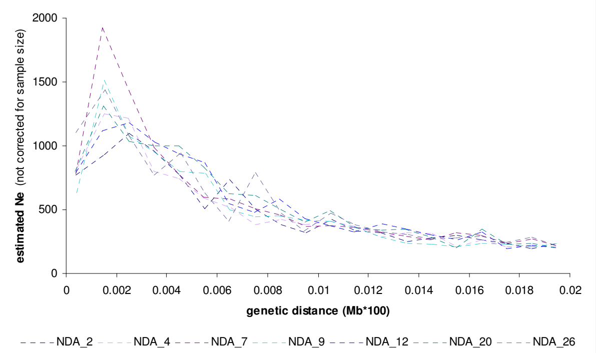
3. **Estimation of effective population size**
   - approximation (1Mb ~ 0.01Morgan) vs. Morgan

Application to real data

3. Effective population size

N‘Dama

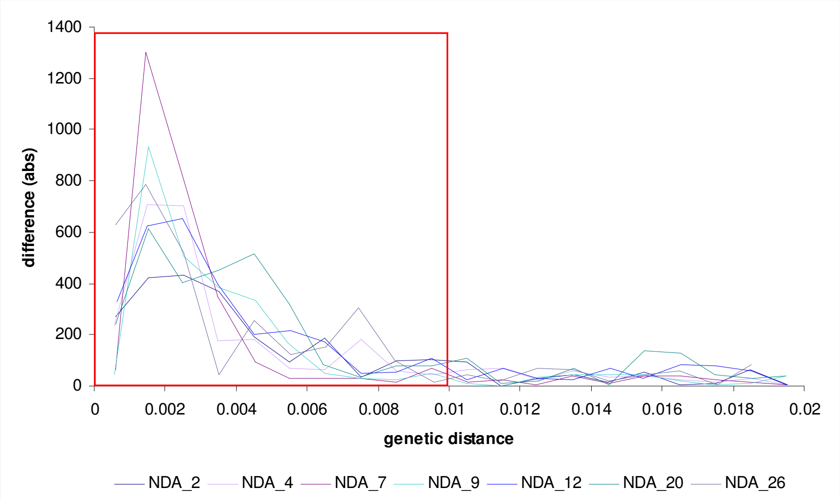




Application to real data

3. Effective population size

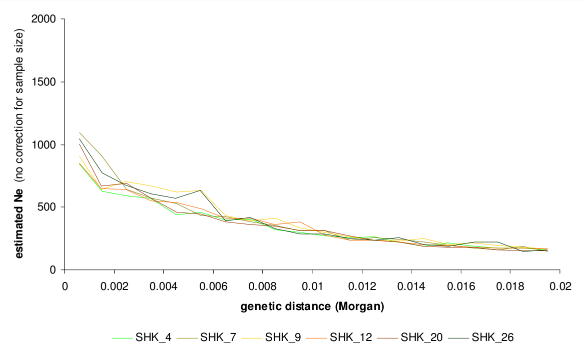
N‘Dama

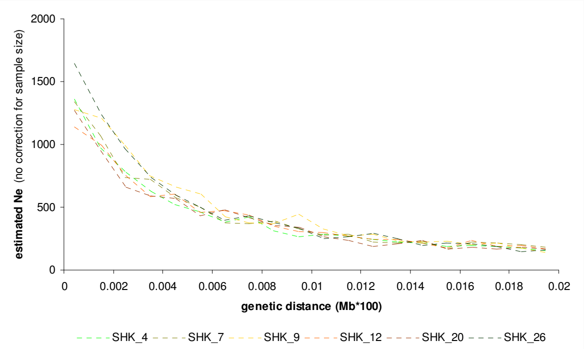


Application to real data

3. Effective population size

Sheko

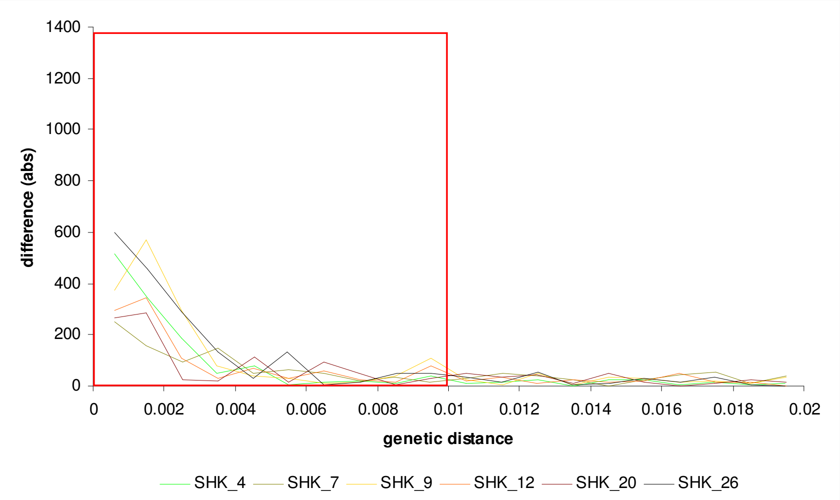




Application to real data

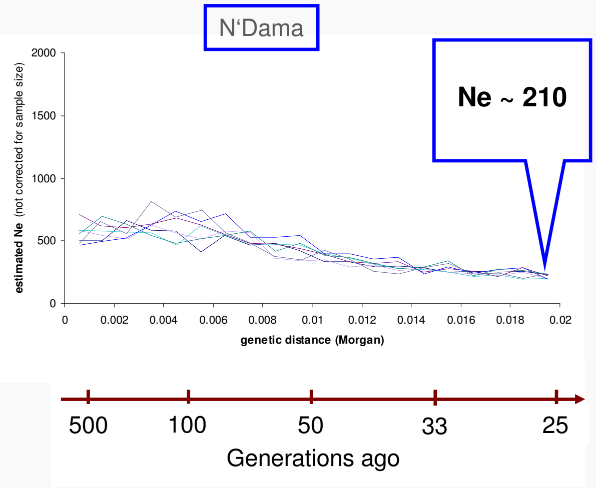
3. Effective population size

Sheko

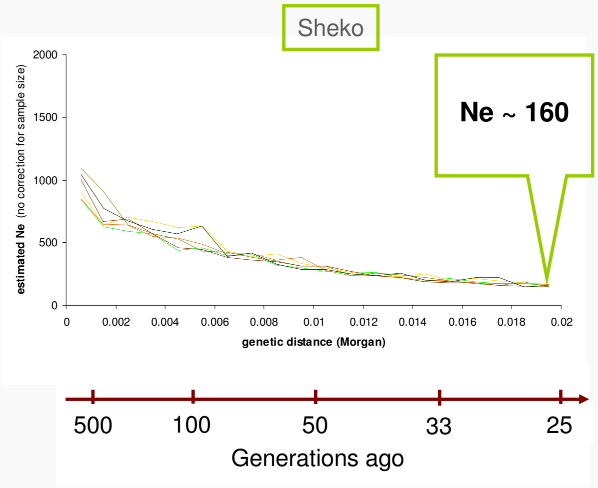


Application to real data

3. Effective population size


N‘Dama

Ne ~ 210


Sheko

Ne ~ 160

Conclusions

Message from LD-measures?

Differences in general and regional LD serve as indicators for differences in population history.

Is the estimation of recombination rates necessary?

Yes, recombination rates are not constant. Working with physical distance is ok for recent history, for past history it might lead to an upward bias (i.e. below 1 Mb).

LD-based estimation of effective population size – useful as background for conservation genetics?

Yes, combined with additional information. However, further investigations on minimum sample size, single chromosome vs. genome-wide data, admixture, etc. required.

Conclusions

Message from LD-measures?

Differences in general and regional LD serve as indicators for differences in population history.

THANK YOU!

Yes, combined with additional information. However, further investigations on minimum sample size, single chromosome vs. genome-wide data, admixture, etc. required.

Acknowledgements

IAEA, Vienna, for the contribution to the Bovine HapMap Project by sponsoring the genotypings of the Sheko breed.

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