Genetic Inference of Recent Fragmentation

Rémi Tournebize, Simon Boitard, Olivier Mazet, Lounès Chikhi

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genetic inference of recent fragmentation


MidJourney

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Simon Boitard
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05 VI 2024

wild
populations
are
often
structured

MidJourney

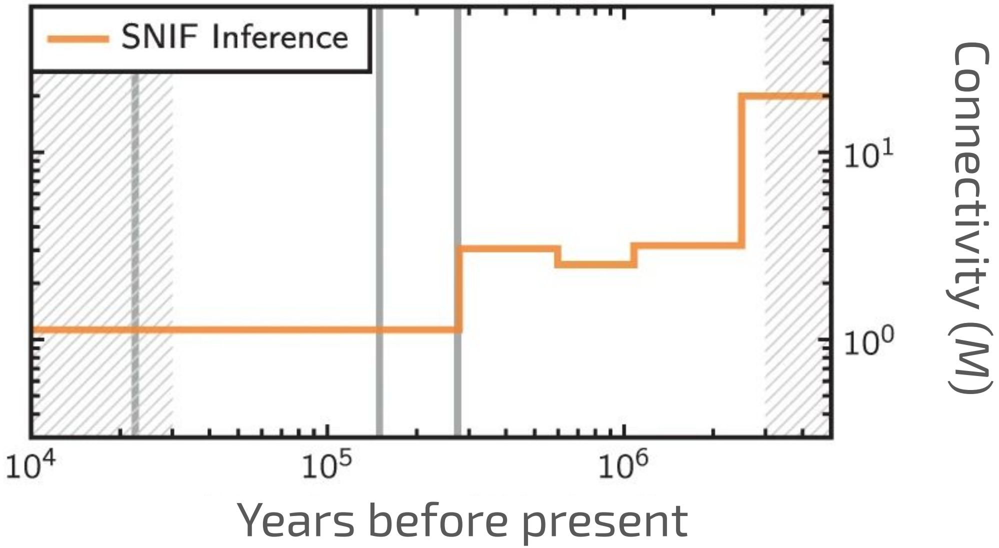
barriers to gene flow can **reduce** or **disrupt** their **connectivity**

MidJourney

in the long run, this can lead to increased **divergence**

MidJourney

Most current genetic methods infer only **ancient** changes in connectivity


Using a French human genome

Arredondo et al. (2021)
Steux et al. (2024) (*in prep.*)

but **human activities** have created **very recent** *possible* barriers to gene flow (< 10 generations ago)

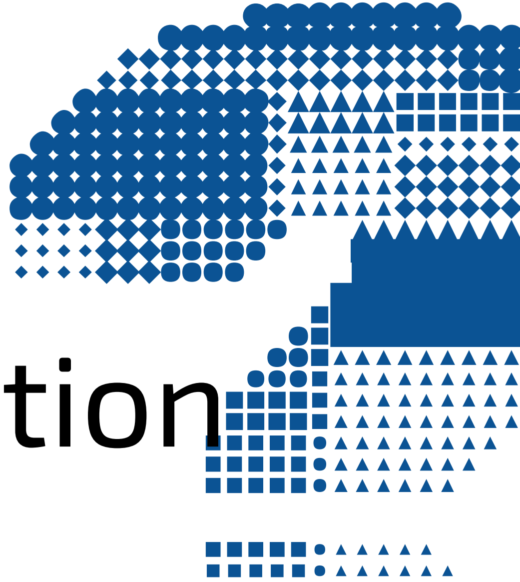
how well DNA

can help us

detect

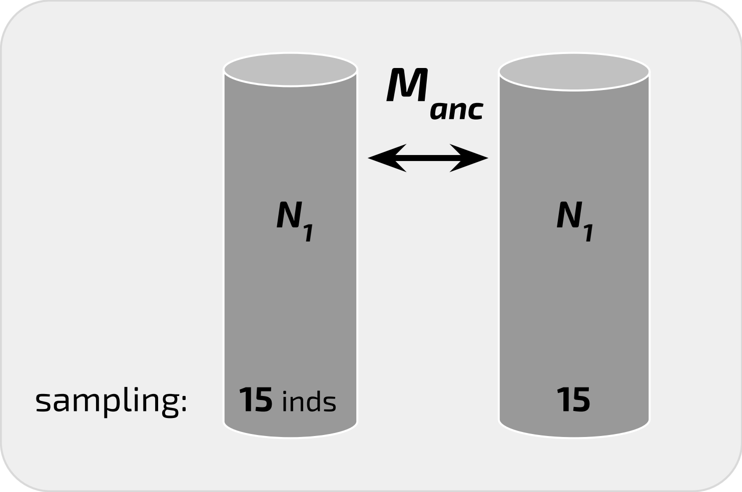
very recent

fragmentation

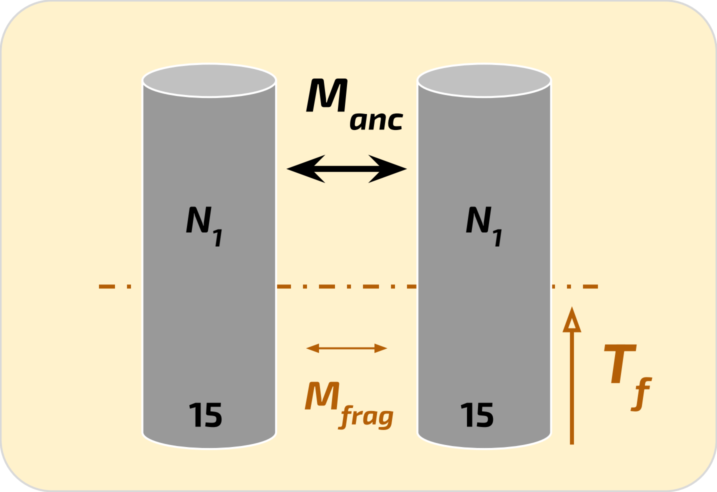


A study using **simulations**

`stationary`
`model`



`fragmentation`
`model`

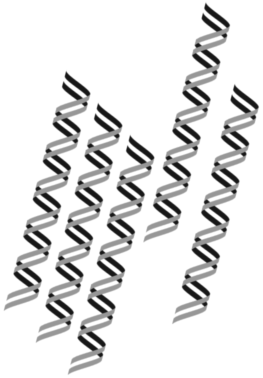


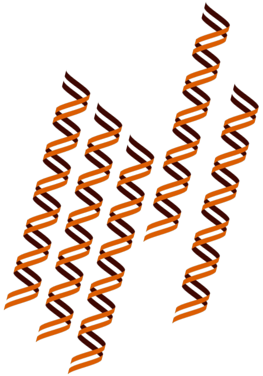
stationary model

fragmented model

2,500 simulated

genetic data per model

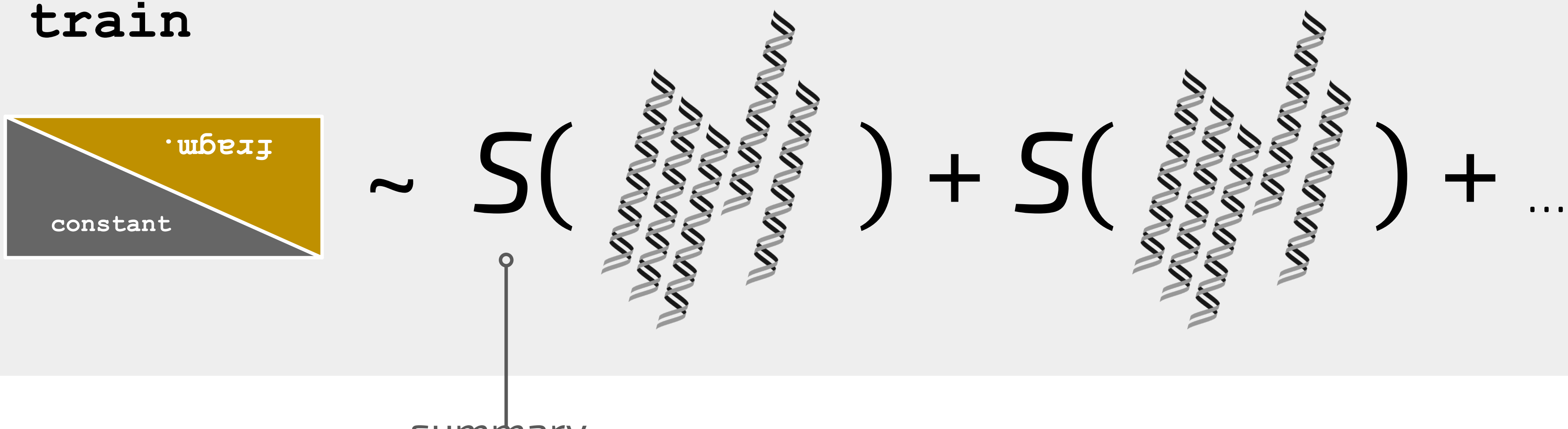




G=5×25 Mbp

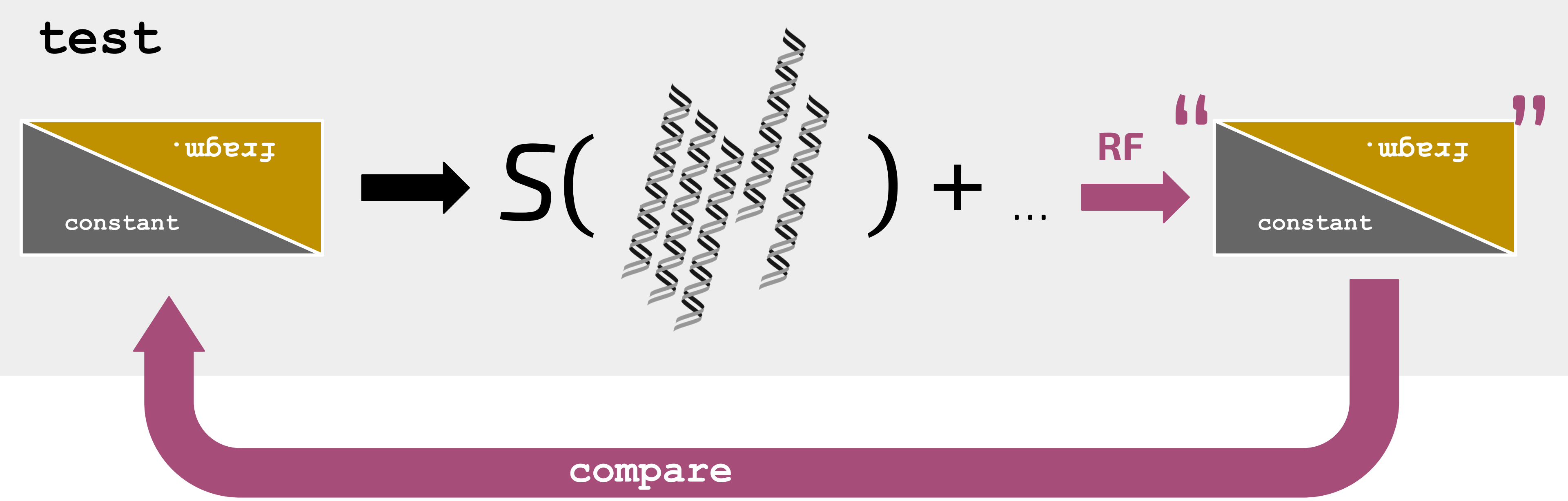
Test the performance to infer underlying models using **ABC** with **random forests**

`train`



summary statistics

Test the performance to infer underlying models using **ABC** with **random forests**

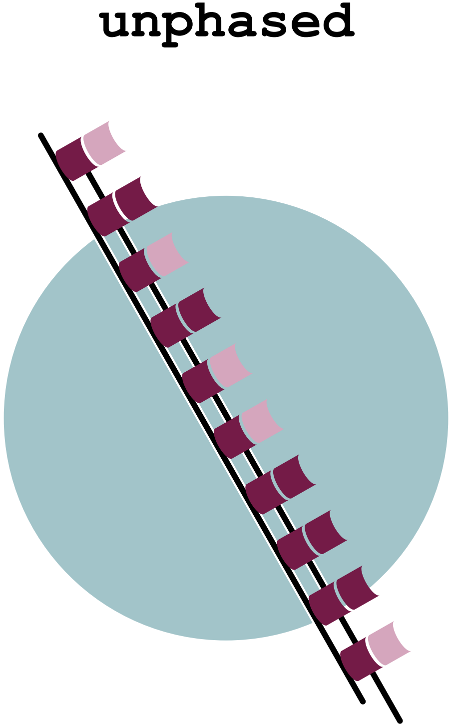


using **different**

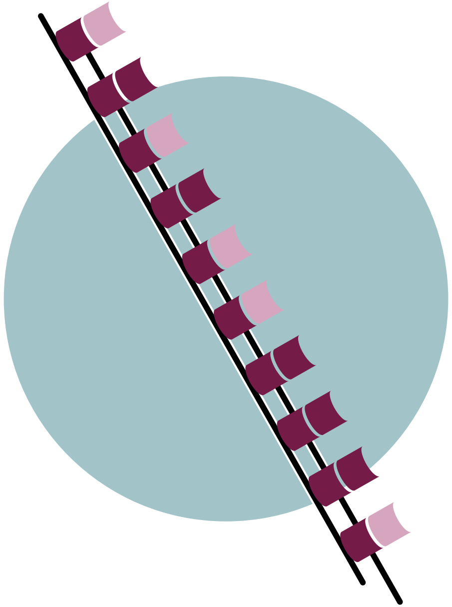
types **of**

genomic **data**

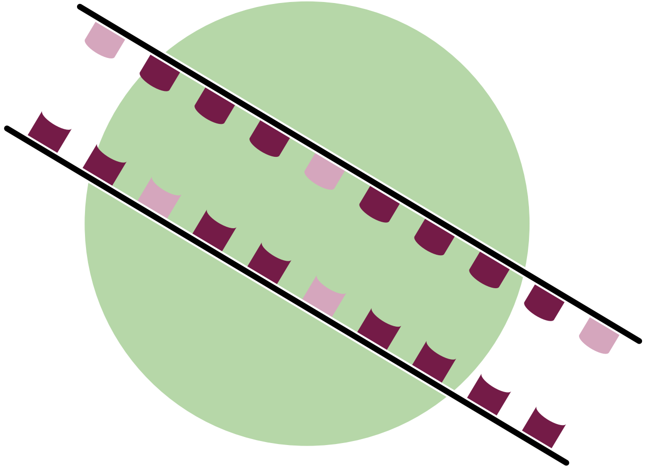
types of genomic **data**



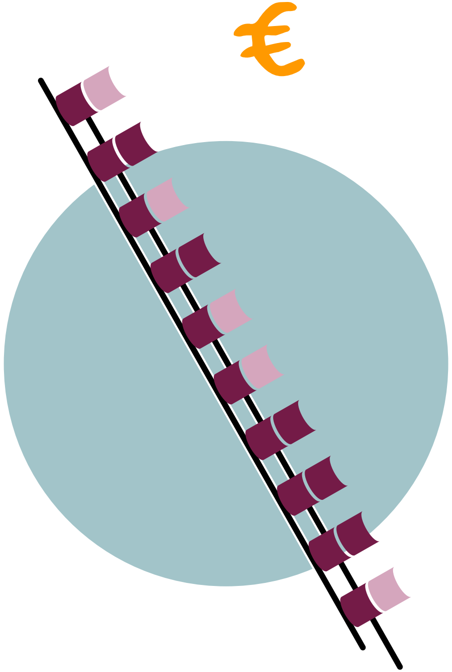
unphased



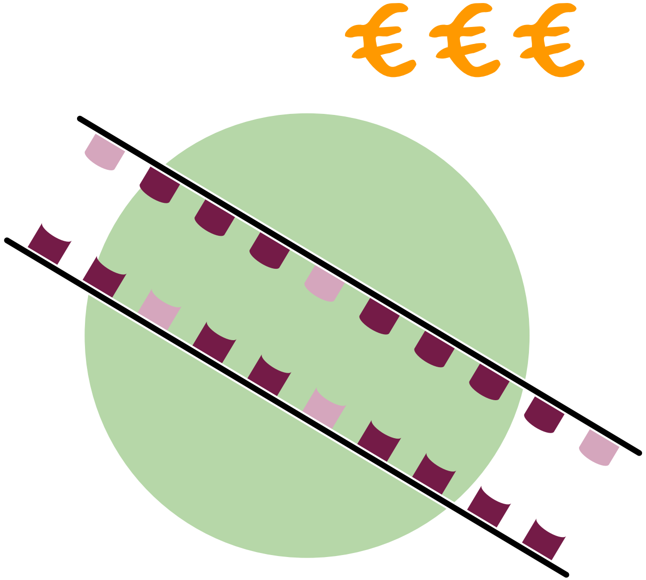
phased



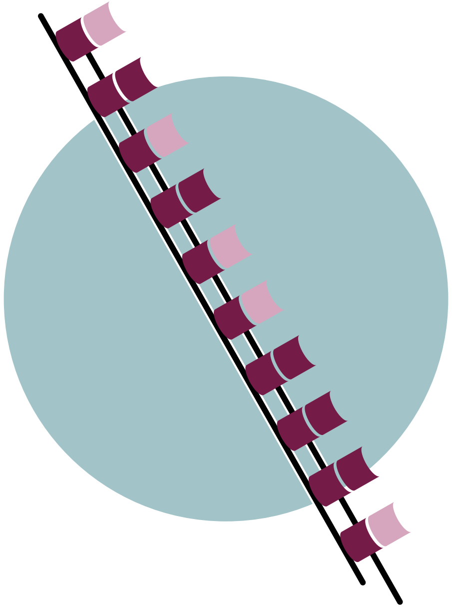
unphased



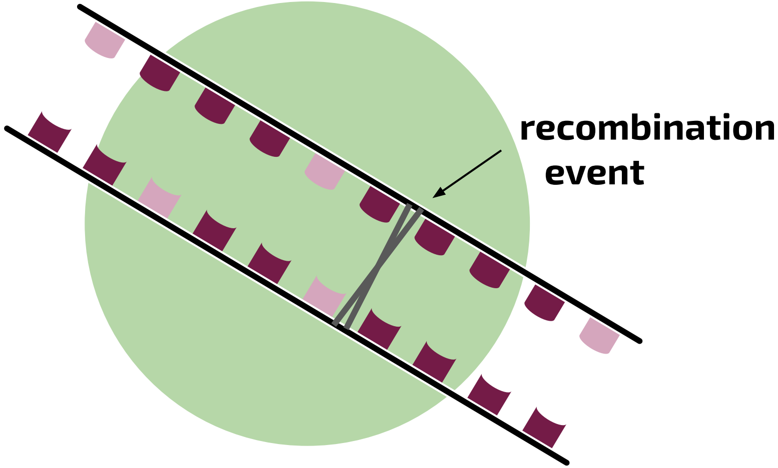
phased



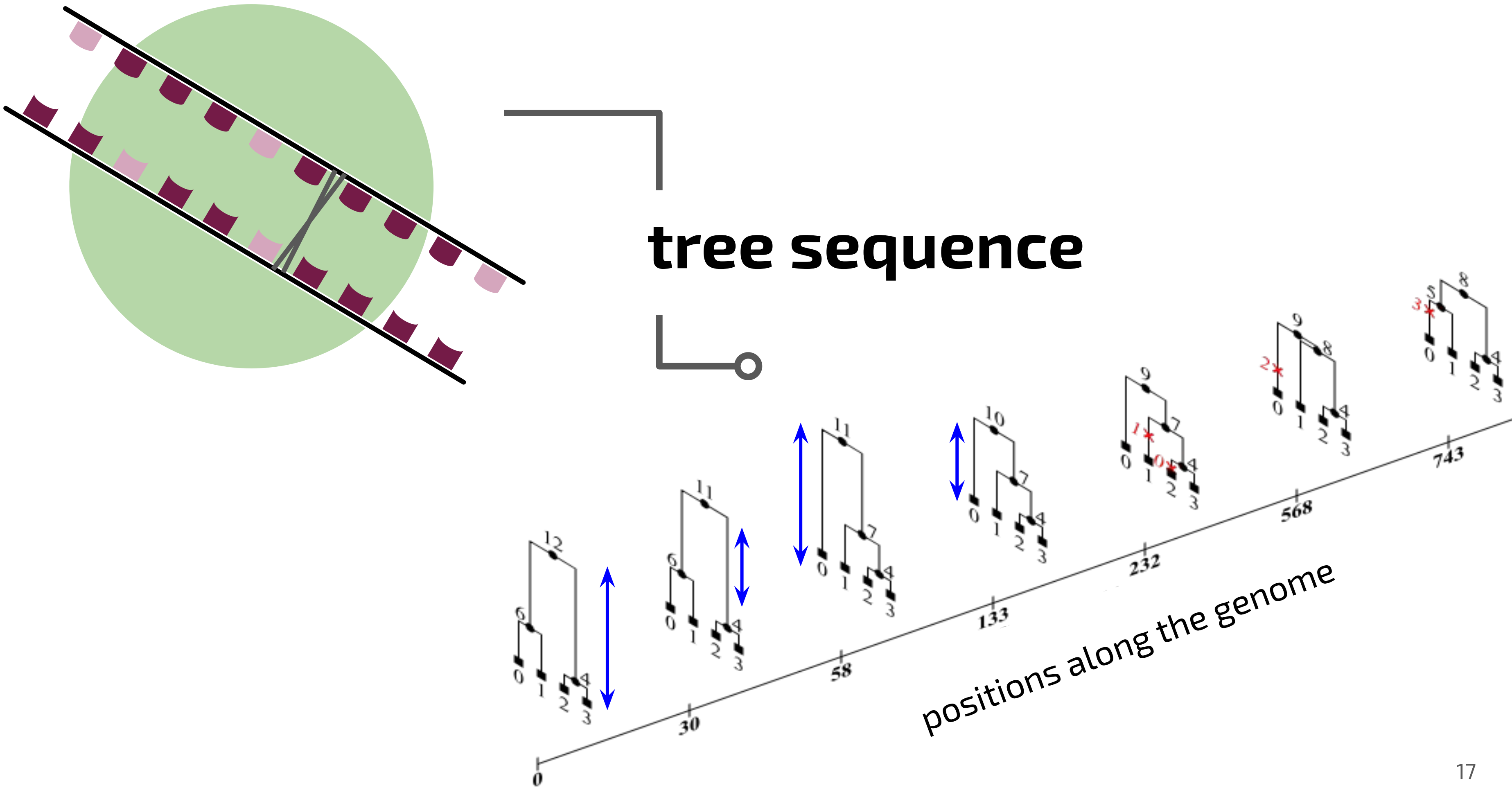
unphased



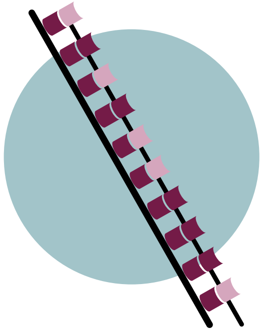
phased



tree sequence



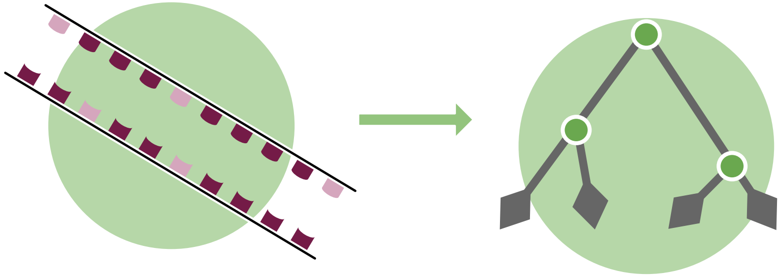
positions along the genome



genotypic statistics

within-pop —— between

- 2D-jAFS (Allele Frequency Spectrum)
- Tajima's *D*
- Nucleotide diversity/divergence
- F_{ST}



genealogical statistics

within-pop —— between

- Distributions of $(T_k)_k$
- Distributions of $(T_c)_k$
- PLFT (Proportion of Lineages as a Function of Time)

Maruvka et al. (2011)

+ subtracted

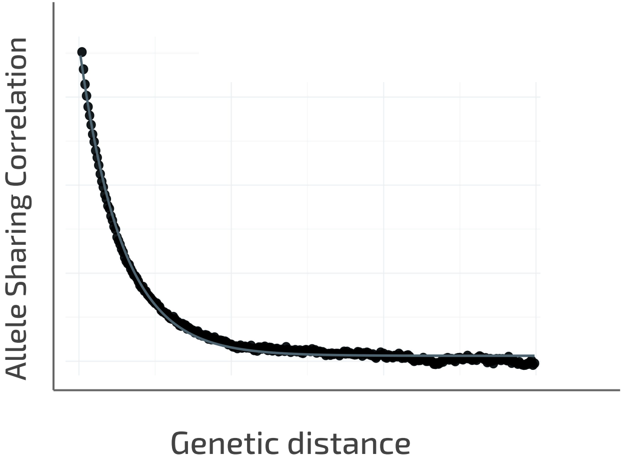
between - E(within1, within2)

genotypic statistics

within-pop ——— between

➔ ***ASCEND***

Tournebize, Chu, Moorjani (2022)

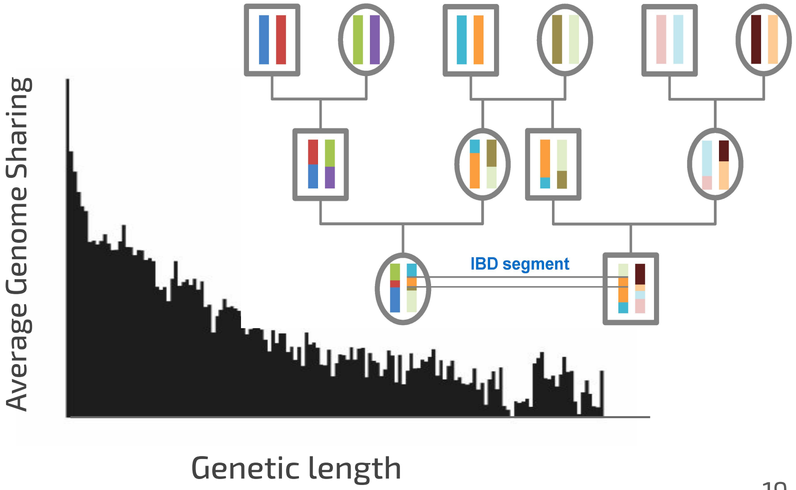


genealogical statistics

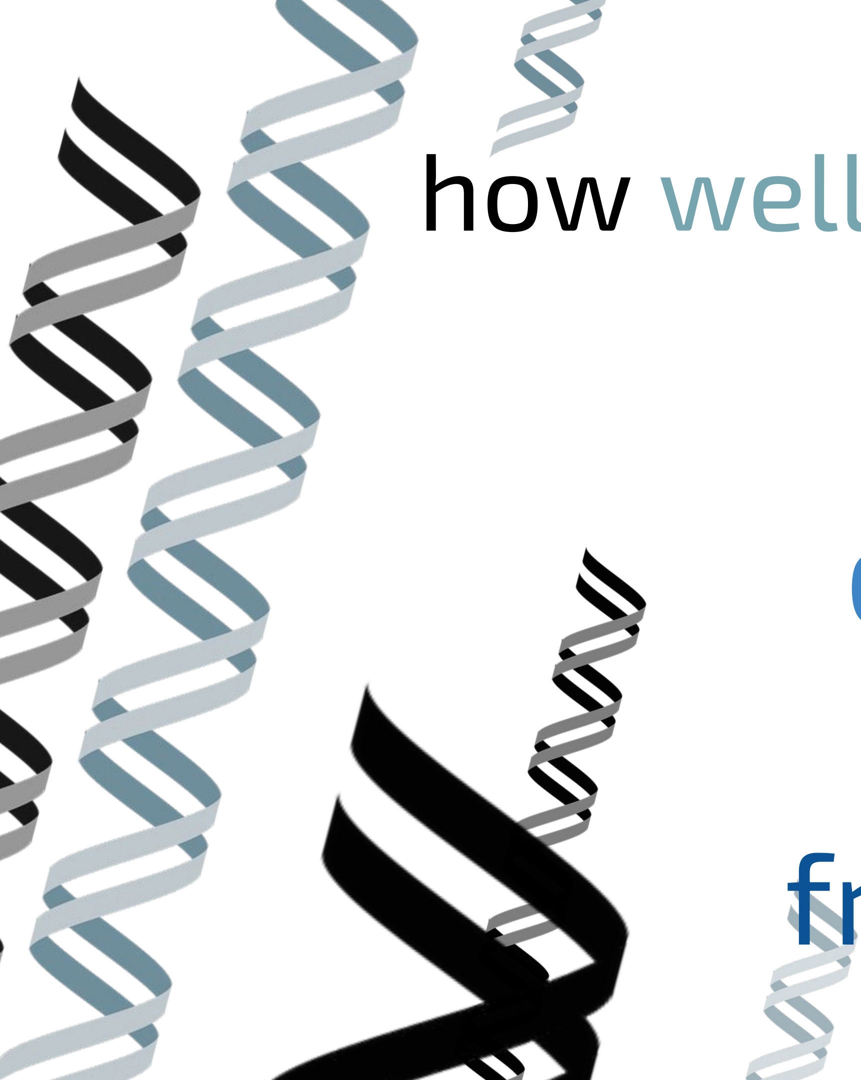
within-pop ——— between

➔ **Genomic correlations in pairwise coalescences + T_k + T_c + PLFT**

➔ **Distribution of IBD lengths**



+ subtracted

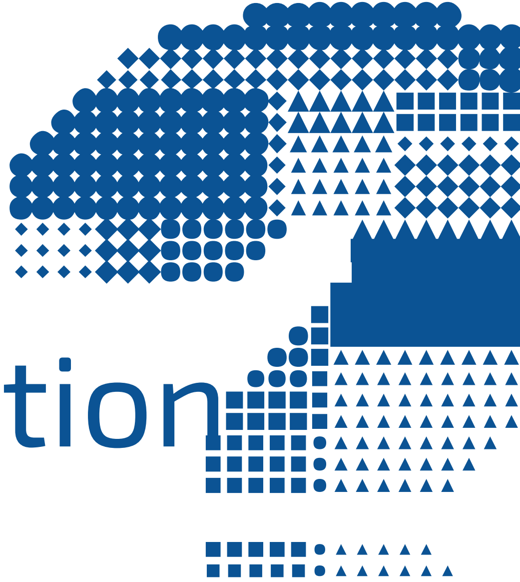


how well ≠ summary statistics

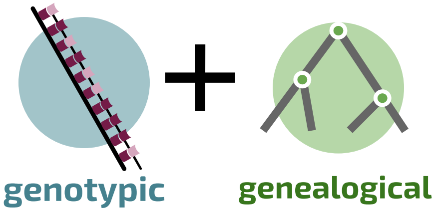
can help us

detect

very recent fragmentation



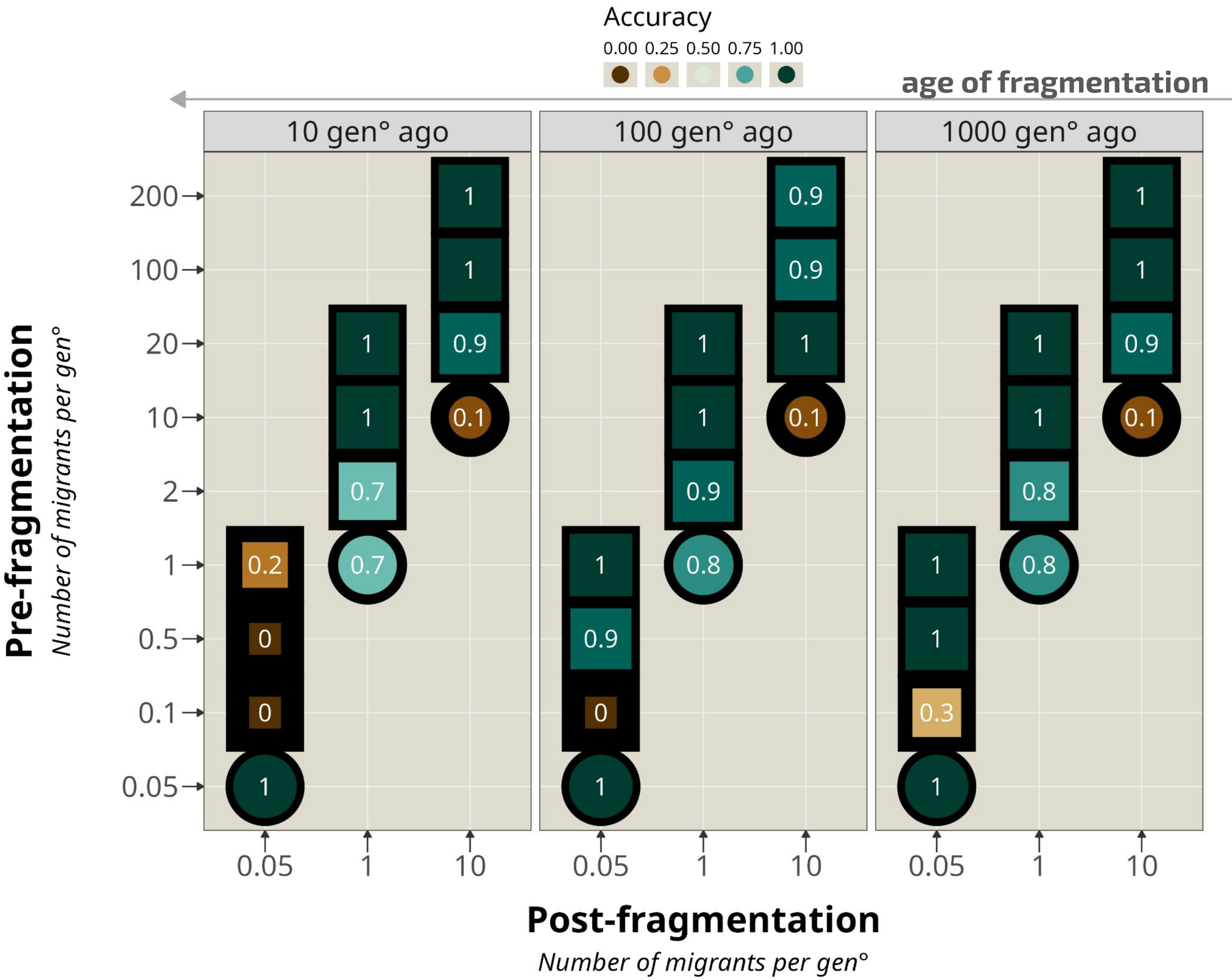
Detection power



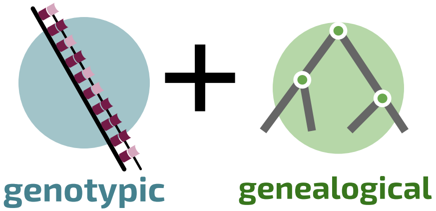
generally a great **power** to detect **fragmentation**

□ fragmentation

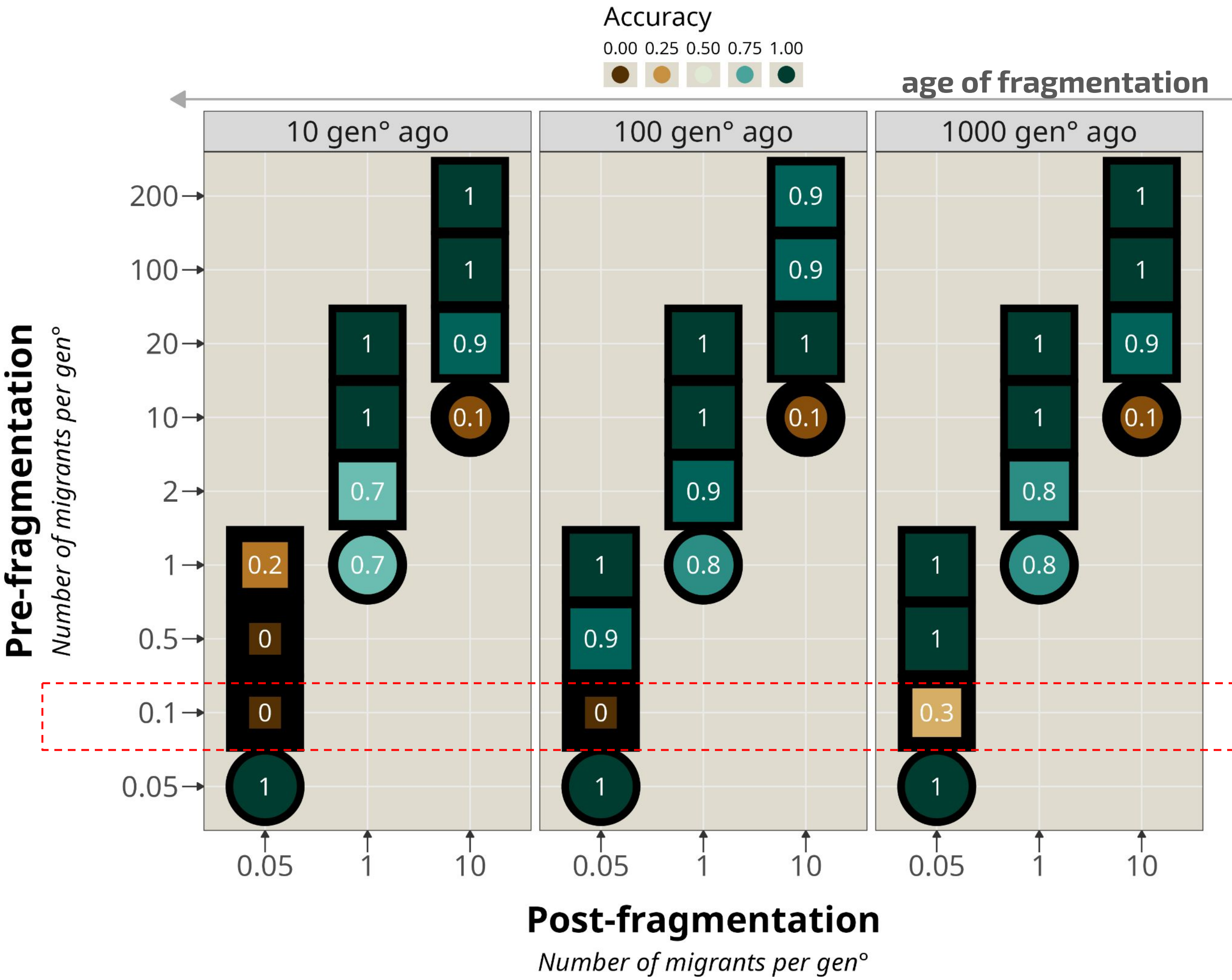
○ stationary



Detection power



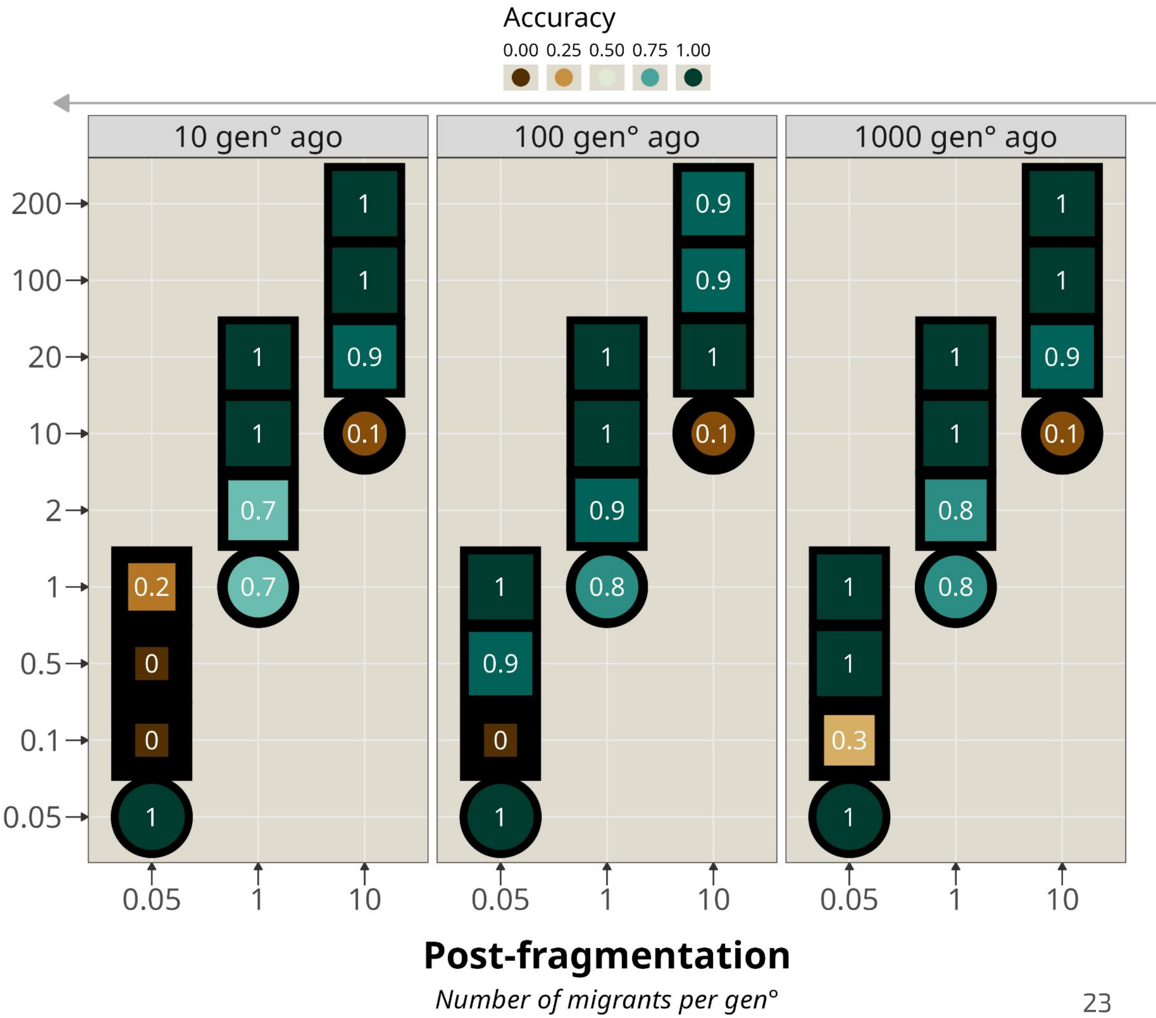
limited power to detect **weak** reduction when pops **already weakly connected**



☐ fragmentation

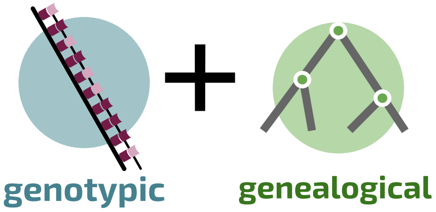
○ stationary

A *conservation genomic* concern



likely to **underdetect** weak fragmentation

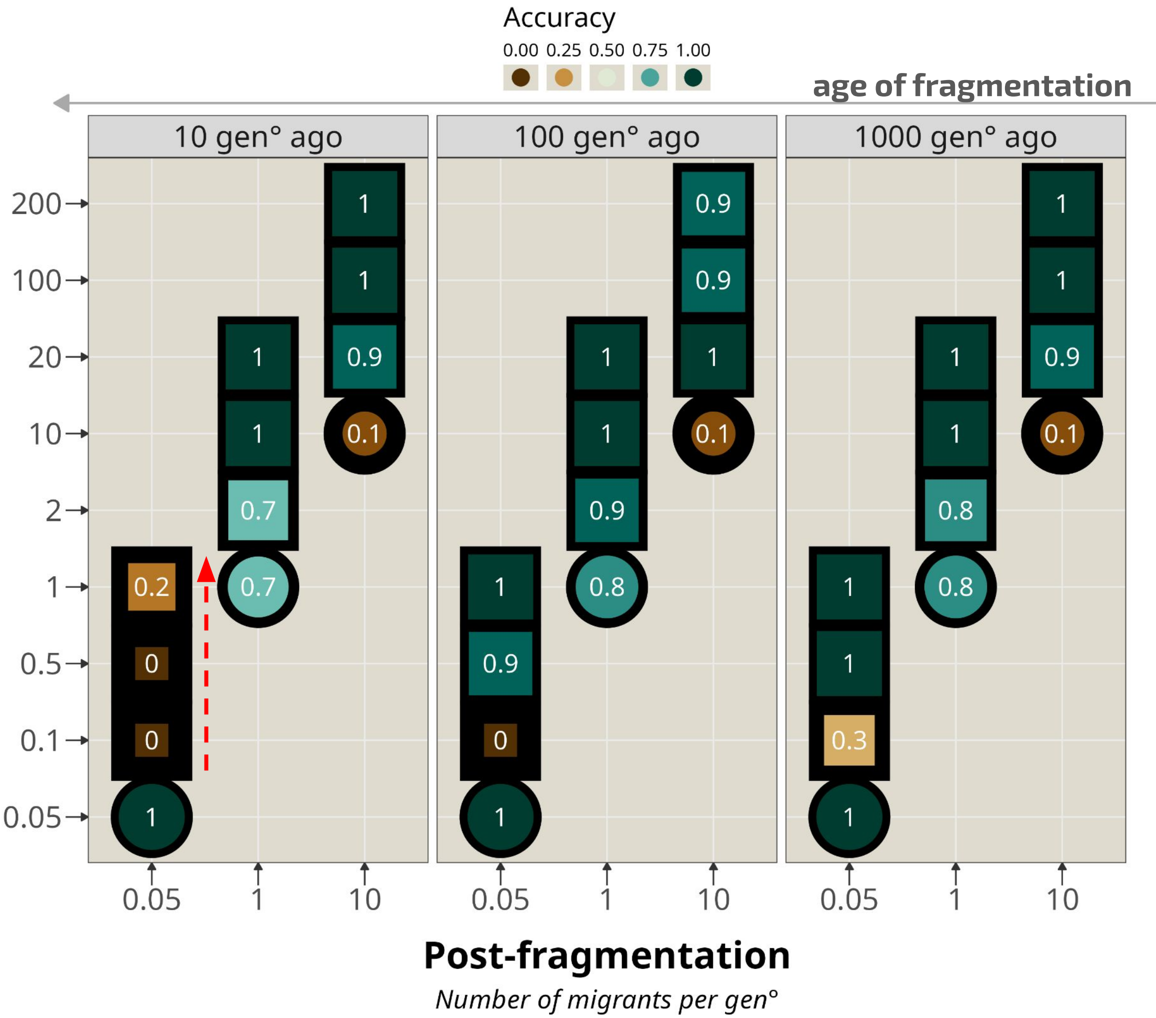
Detection power



yet **power** increases as fragmentation **intensity increases**

☐ fragmentation

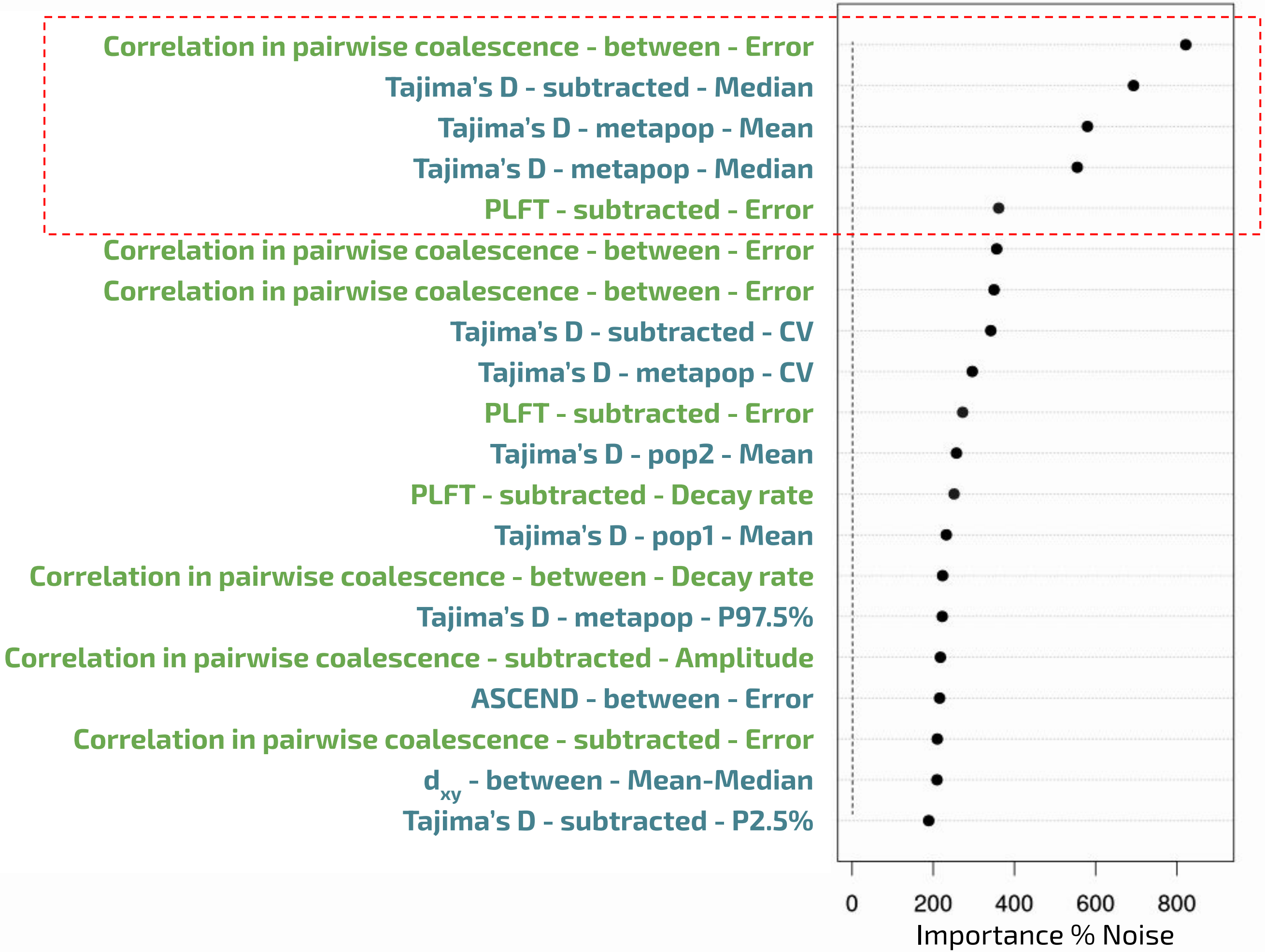
○ stationary



Variable importance

genotypic

genealogical



Conclusions

Genomic data **useful** to detect recent fragm°

Genotypic statistics already **sufficient**

Genealogical data **improve** detection accuracy

Still problematic to identify fragmentation if:
weak &
very recent (< 10 gen°) &
pops were **already poorly connected**

But alternatives possible… i.e. *M* in **other** non-fragmented pops as a proxy to M_{anc}

Thank you!

Simon Boitard
Olivier Mazet
Lounès Chikhi
& the DevOcGen team members

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Biodiversité Occitanie

cnrs
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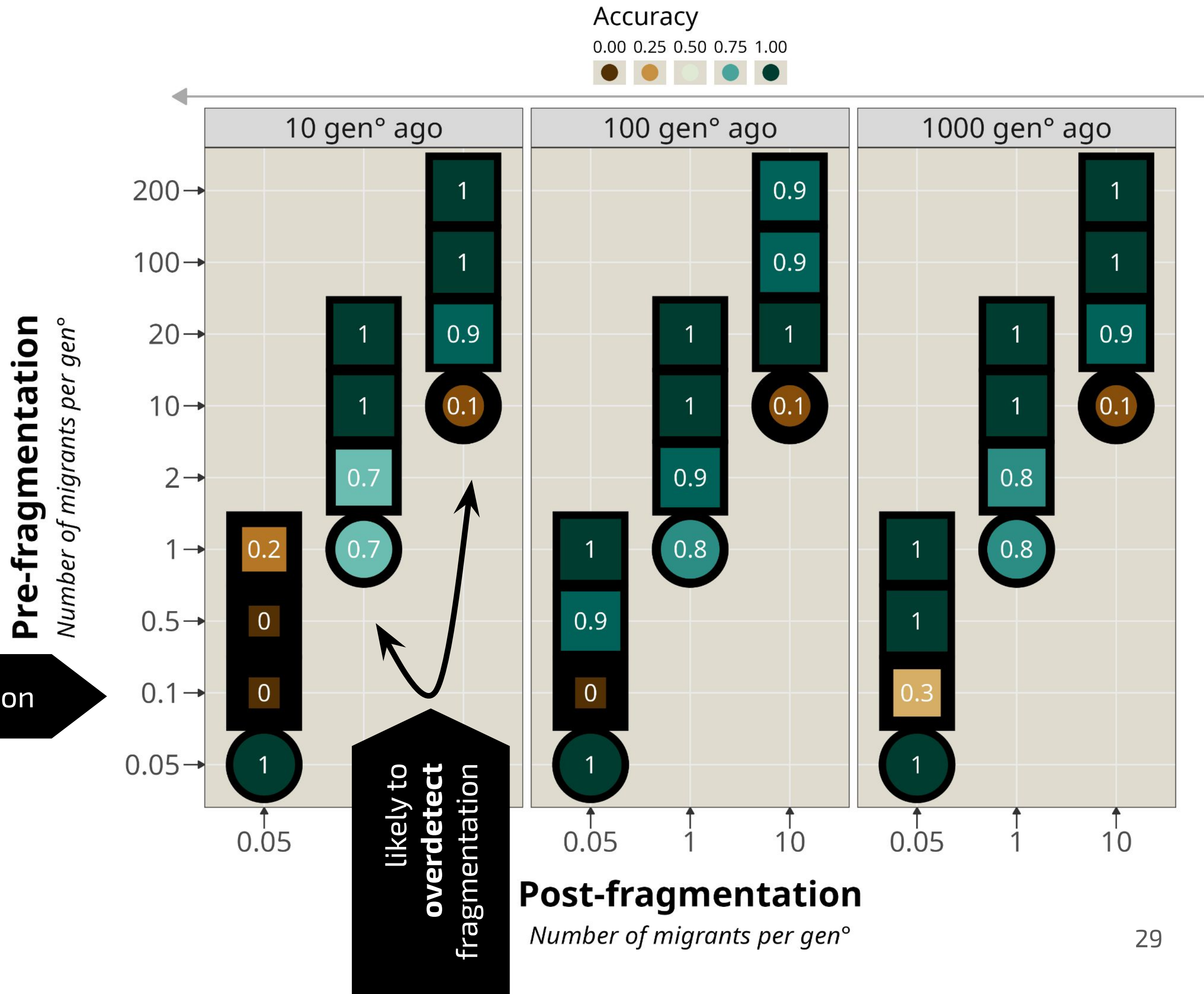
Appendix

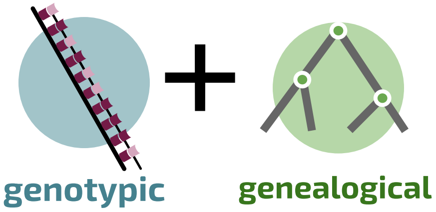
A *conservation genomic* concern



likely to **underdetect** weak fragmentation

likely to **overdetect** fragmentation

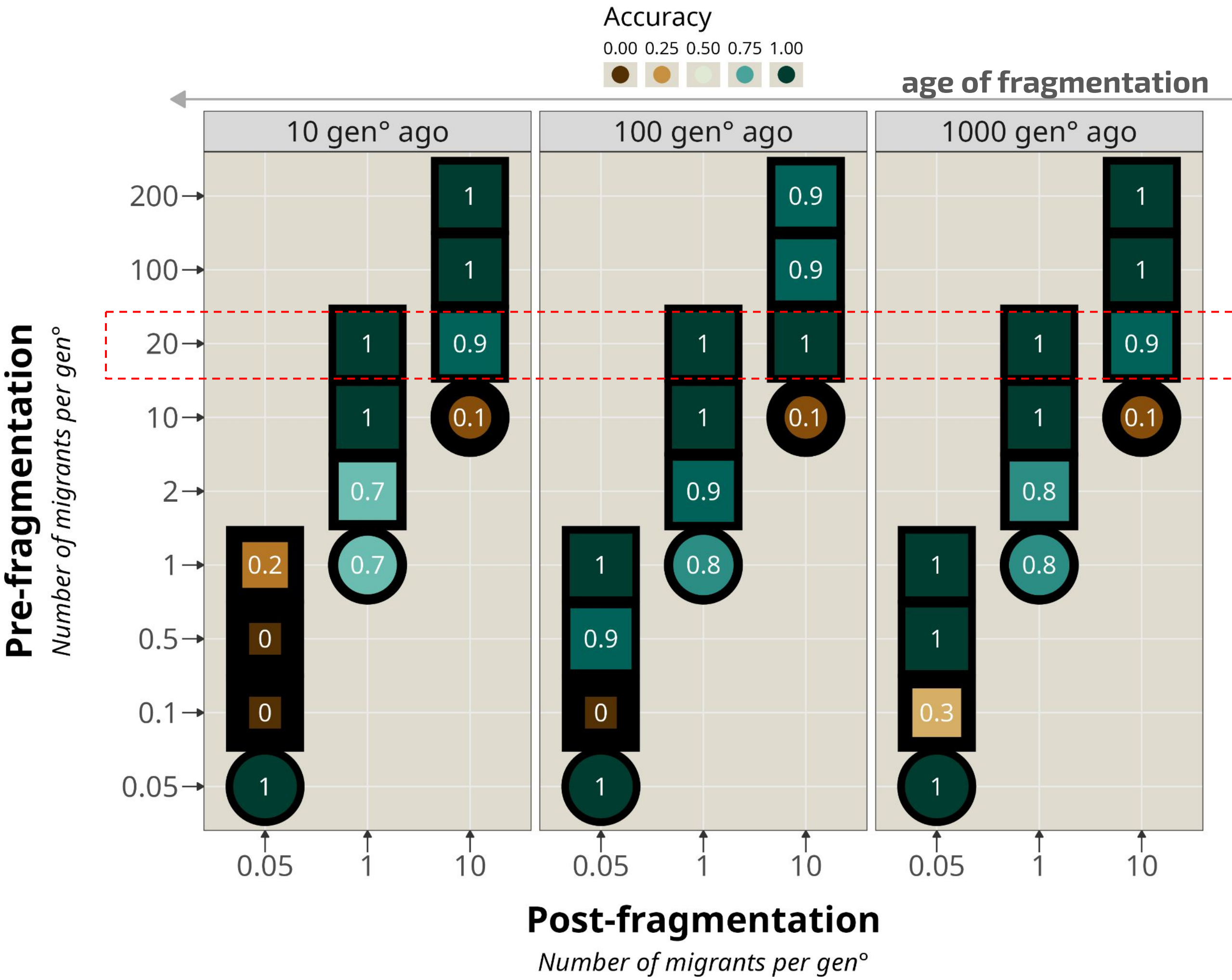
Detection power



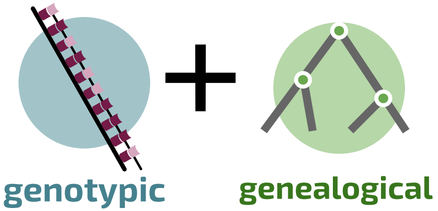
power tends to **increase** with founder **intensity**

☐ fragmentation

○ stationary



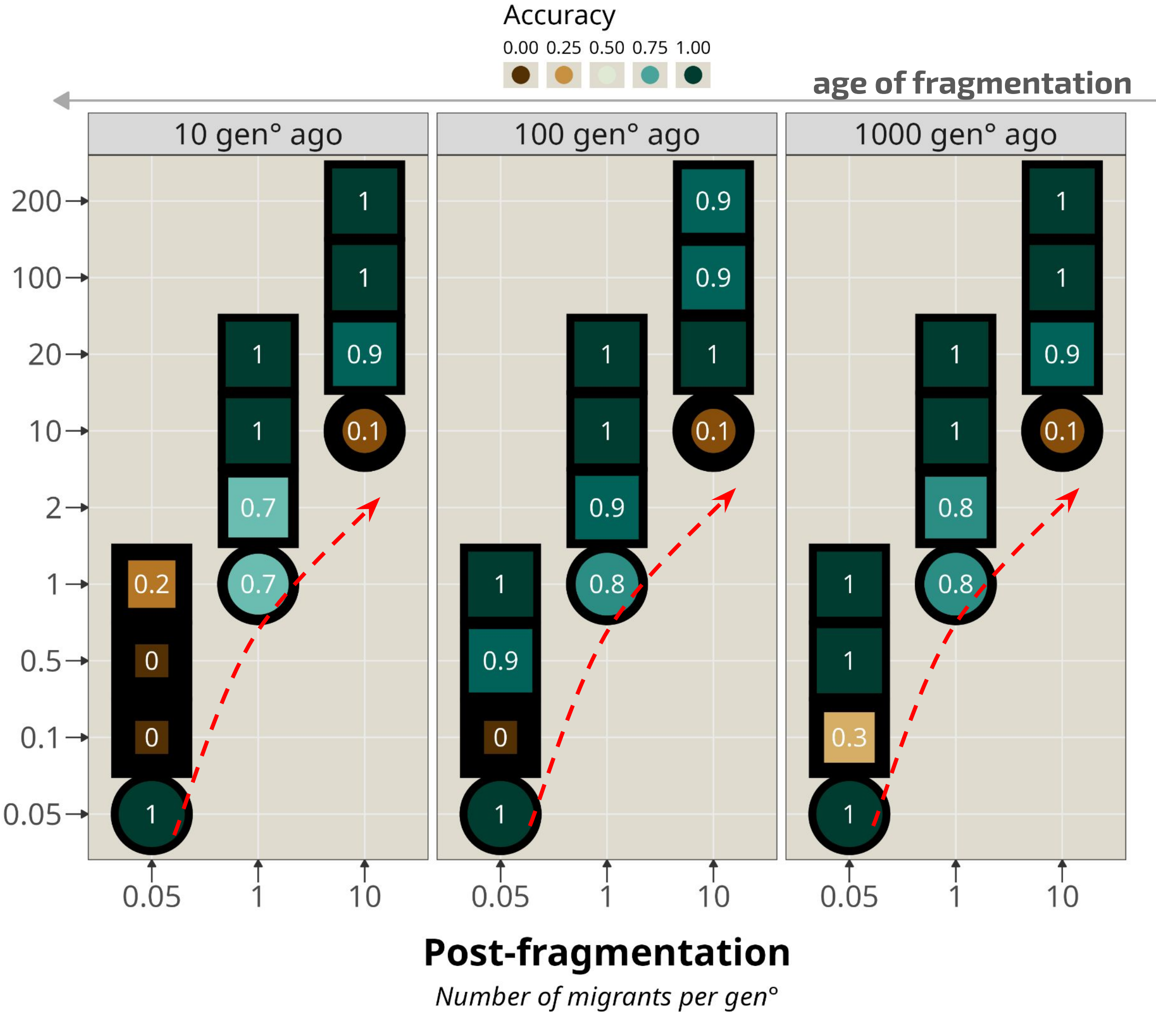
Detection power



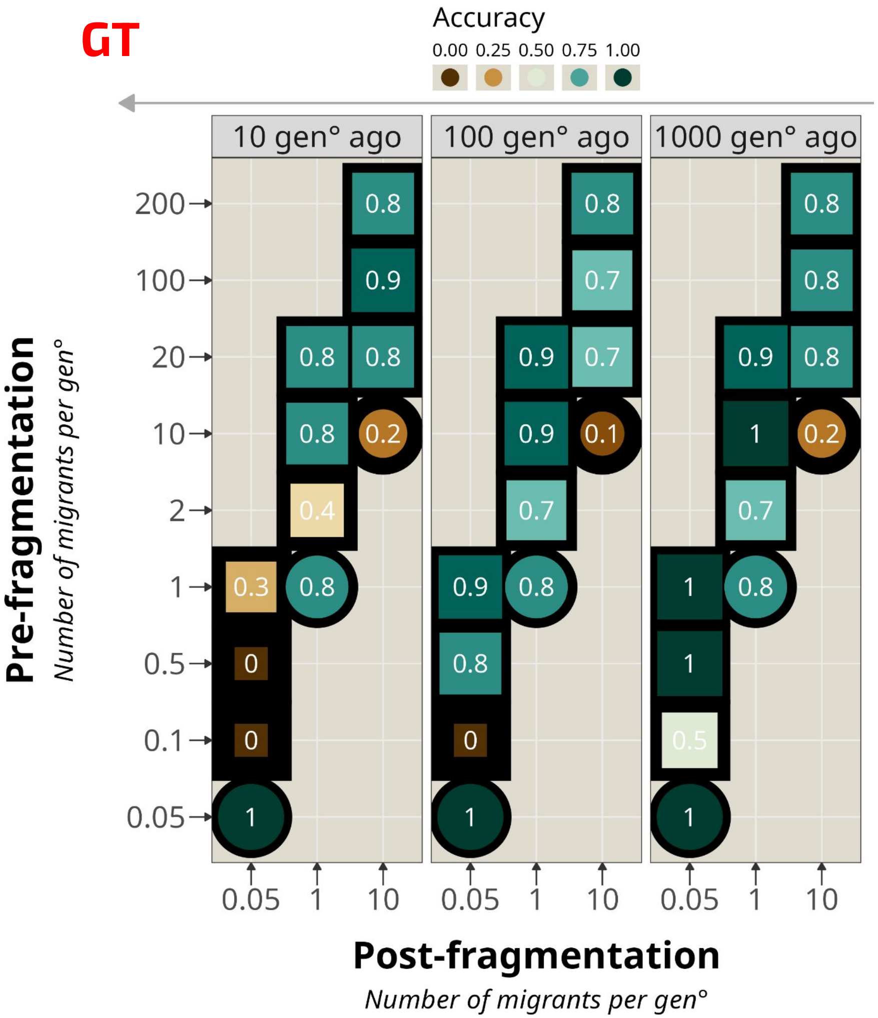
power to detect **stationary** model **decreases** with **migration rate**

☐ fragmentation

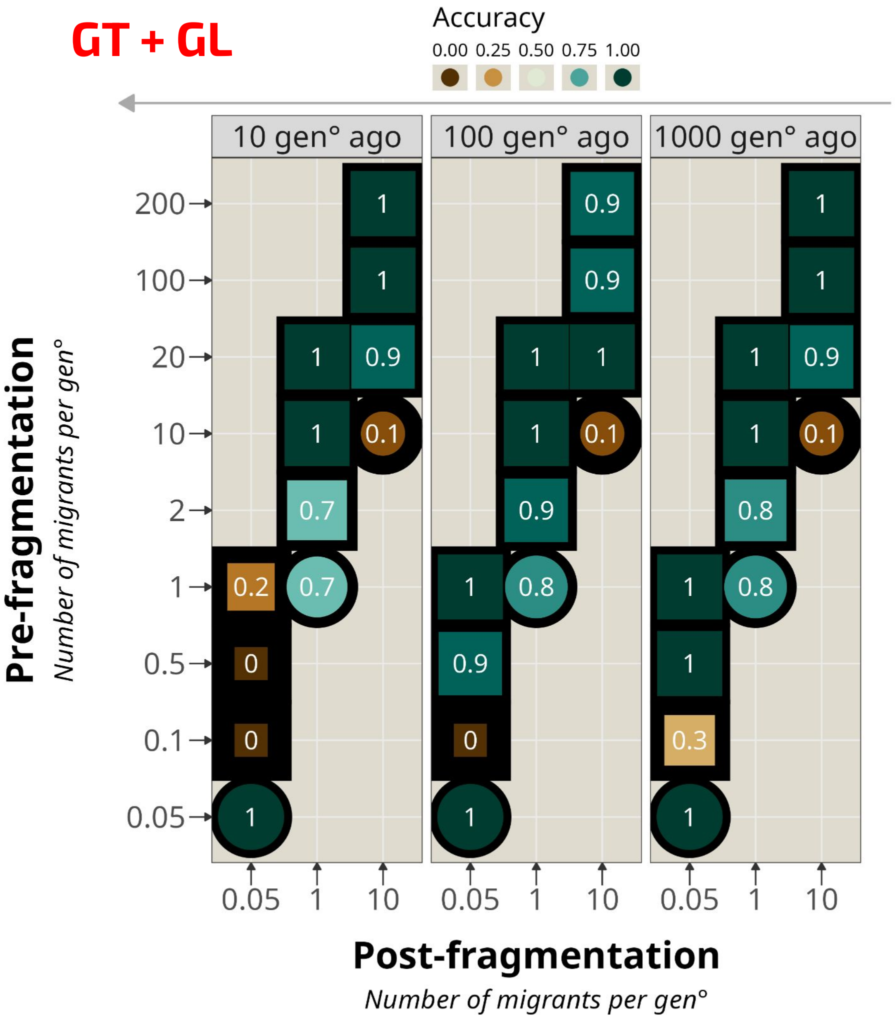
○ stationary



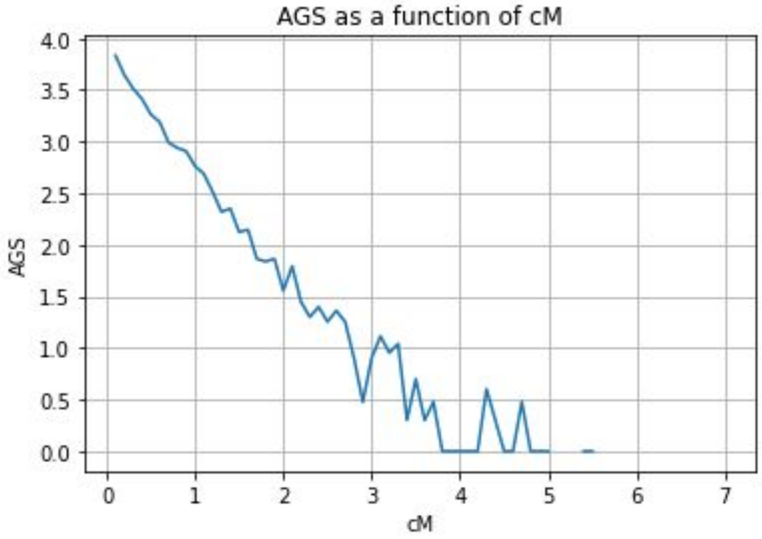
GT


Accuracy
0.00 0.25 0.50 0.75 1.00
10 gen° ago
100 gen° ago
1000 gen° ago
0.8
0.9
0.8 0.8
0.8 0.2
0.4
0.3 0.8
0
0
1
0.8
0.7
0.9 0.7
0.9 0.1
0.7
0.9 0.8
0.8
0
1
0.8
0.8
0.9 0.8
1 0.2
0.7
1 0.8
1
0.5
1
200
100
20
10
2
1
0.5
0.1
0.05
0.05 1 10
Pre-fragmentation
Number of migrants per gen°
Post-fragmentation
Number of migrants per gen°

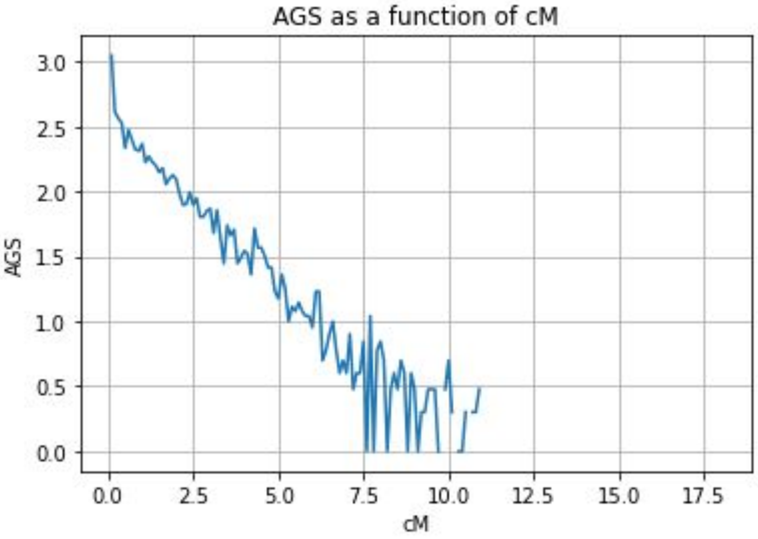
GT + GL


Accuracy
0.00 0.25 0.50 0.75 1.00
10 gen° ago
100 gen° ago
1000 gen° ago
1
1
1 0.9
1 0.1
0.7
0.2 0.7
0
0
1
0.9
0.9
1 1
1 0.1
0.9
1 0.8
0.9
0
1
1
1
1 0.9
1 0.1
0.8
1 0.8
1
0.3
1
200
100
20
10
2
1
0.5
0.1
0.05
0.05 1 10
Pre-fragmentation
Number of migrants per gen°
Post-fragmentation
Number of migrants per gen°

Hudson


AGS as a function of cM
AGS
cM
4.0
3.5
3.0
2.5
2.0
1.5
1.0
0.5
0.0
0
1
2
3
4
5
6
7

DTWF


AGS as a function of cM
AGS
cM
3.0
2.5
2.0
1.5
1.0
0.5
0.0
0.0
2.5
5.0
7.5
10.0
12.5
15.0
17.5