Genetic Inference of Recent Fragmentation

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genetic inference of recent fragmentation


MidJourney

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Lounès Chikhi

05 VI 2024

wild
populations
are
often
structured

MidJourney

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

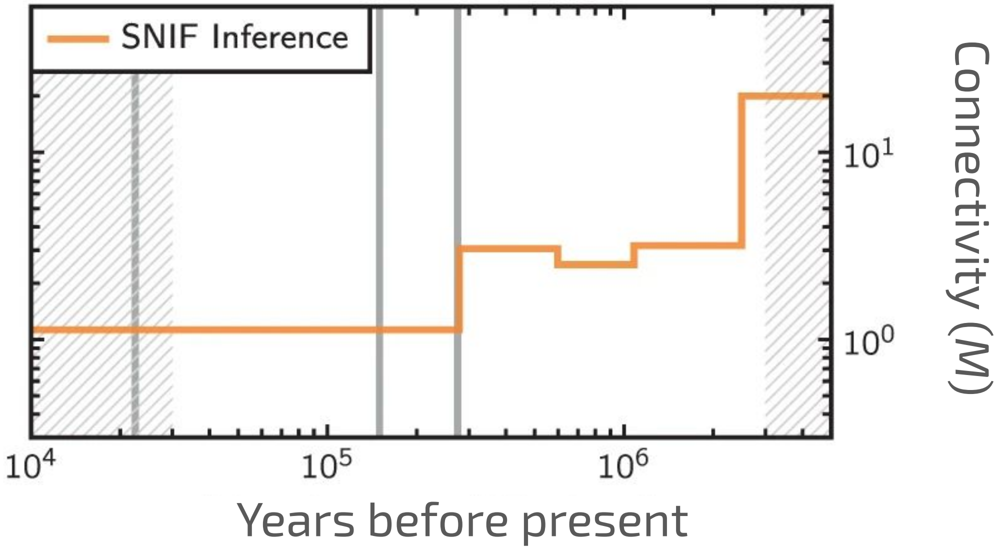
MidJourney

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.*)

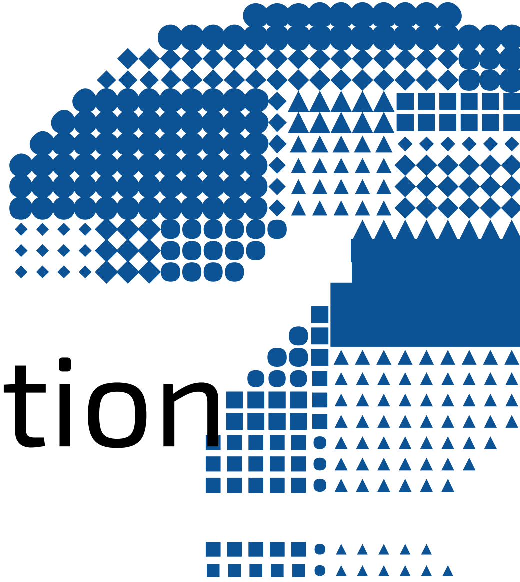
but **human activities** have created **very recent** *possible* barriers to gene flow (< 10 generations ago)

how well DNA

can help us

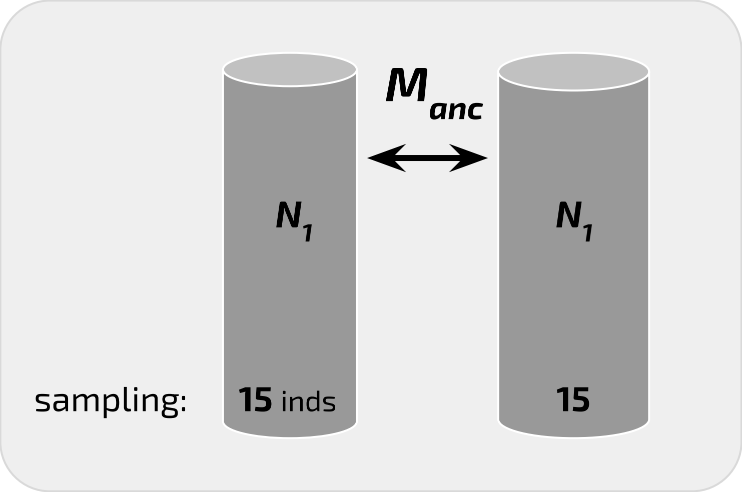
detect

very
recent
fragmentation

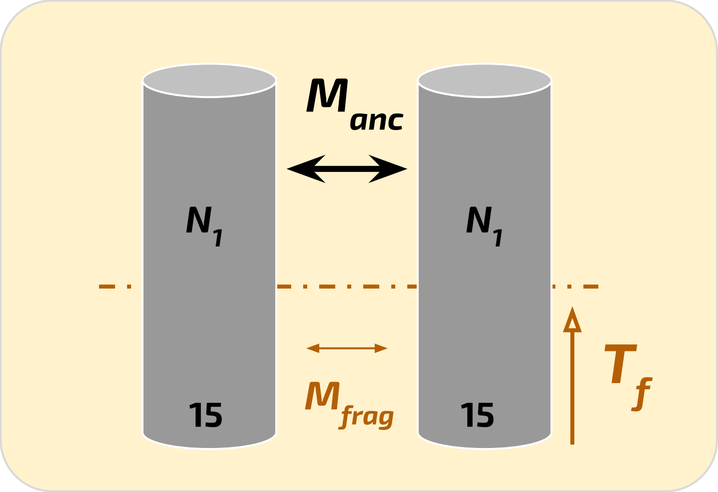


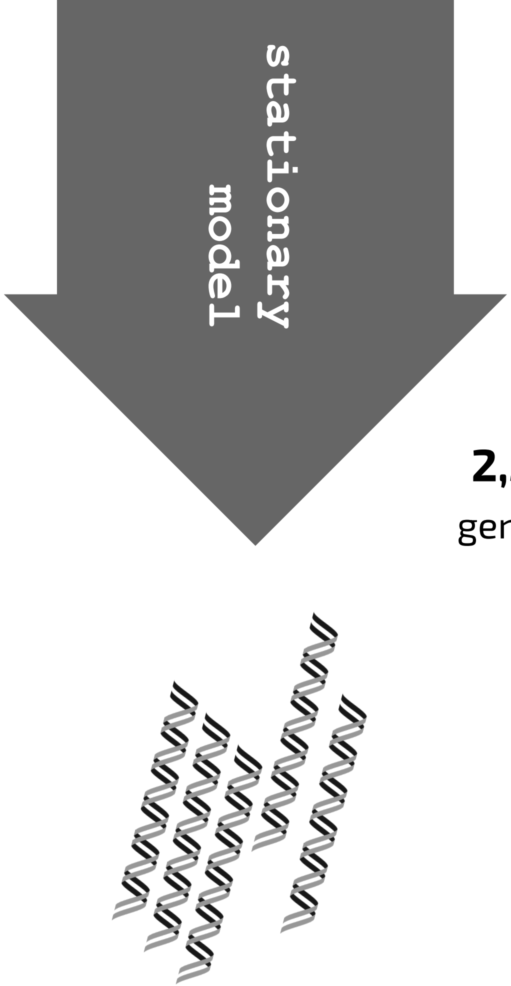
A study using **simulations**

stationary
model

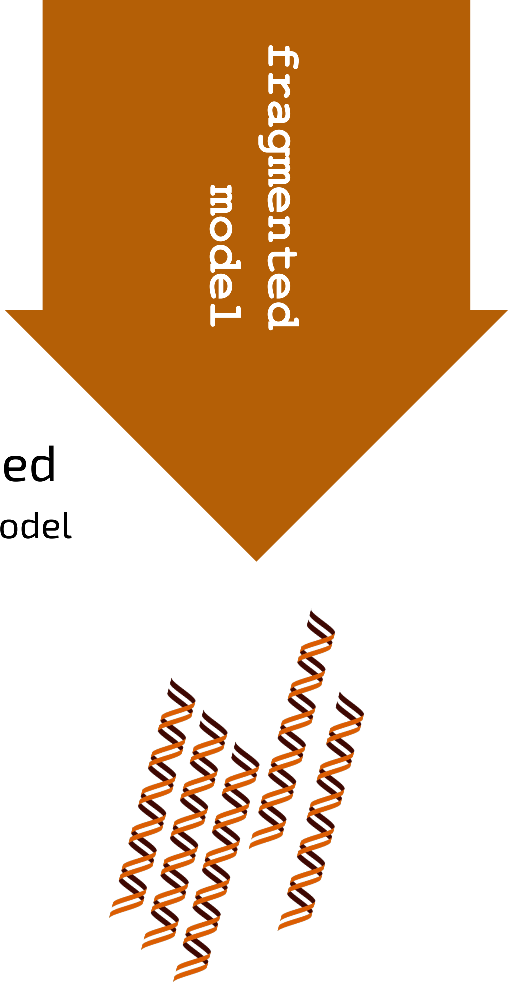


fragmentation
model





`stationary model`



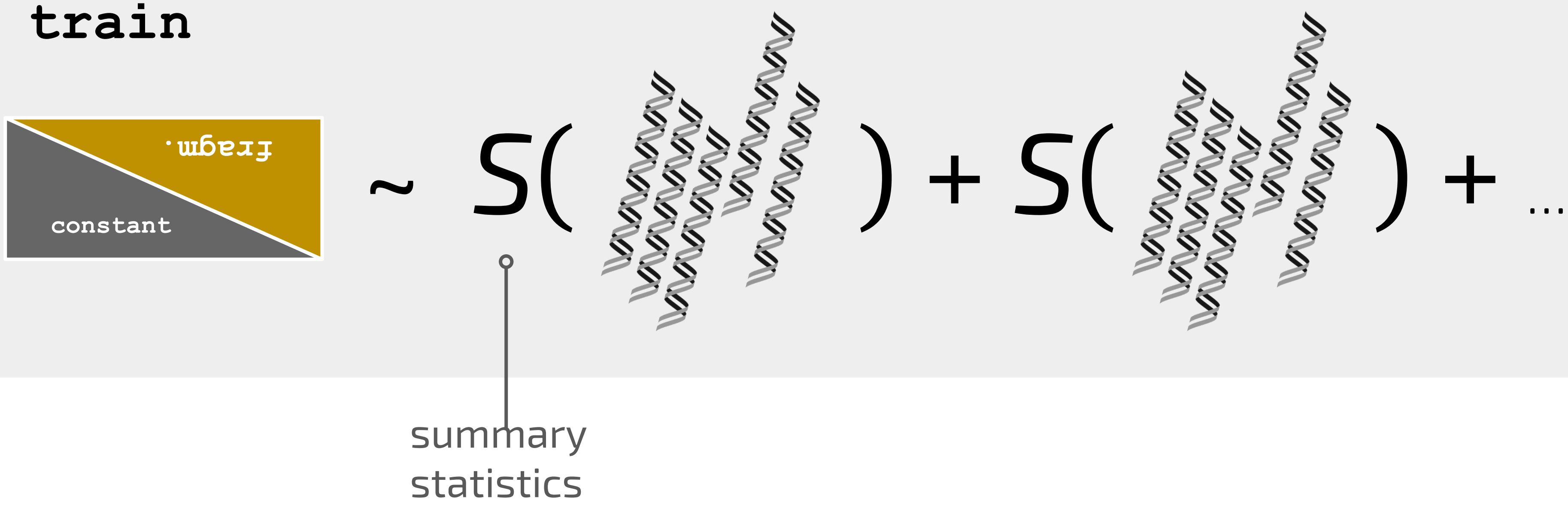
`fragmented model`

2,500 simulated

genetic data per model

G=5×25 Mbp

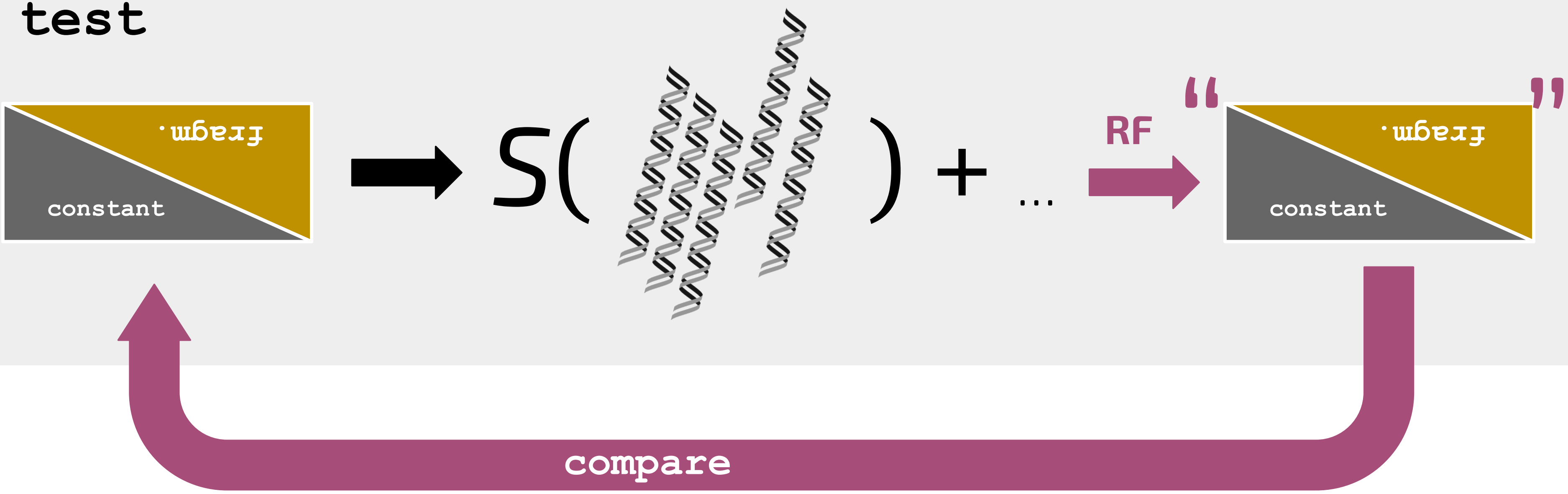
Test the performance to infer underlying models using **ABC** with **random forests**



`train`

summary statistics

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



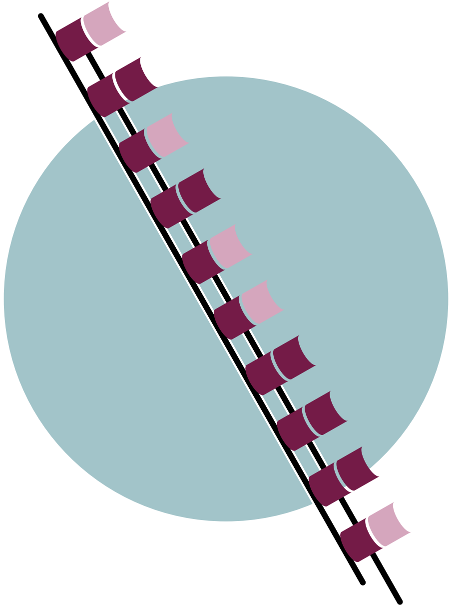
using **different**

types of

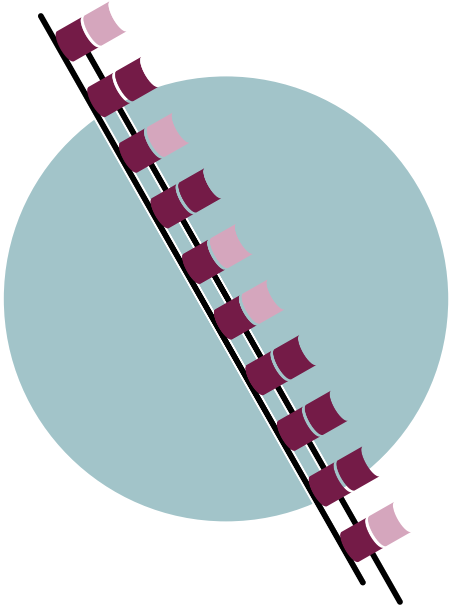
genomic data

types of genomic **data**

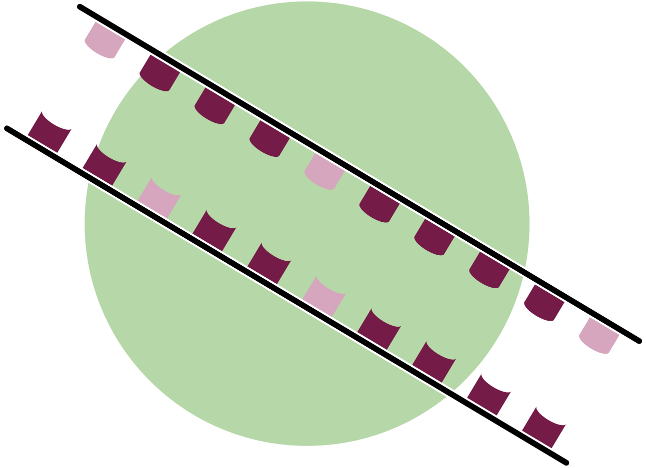
`unphased`



unphased

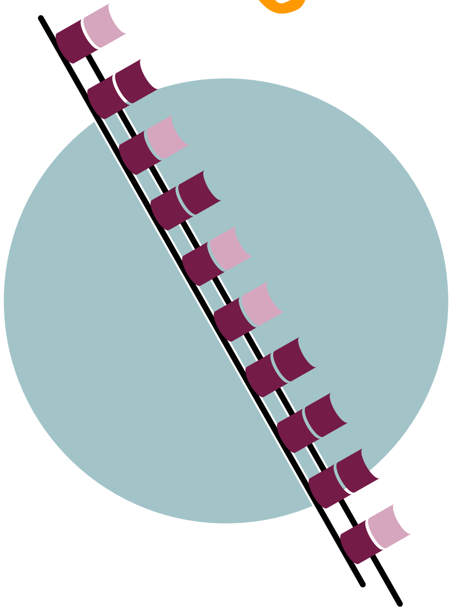


phased



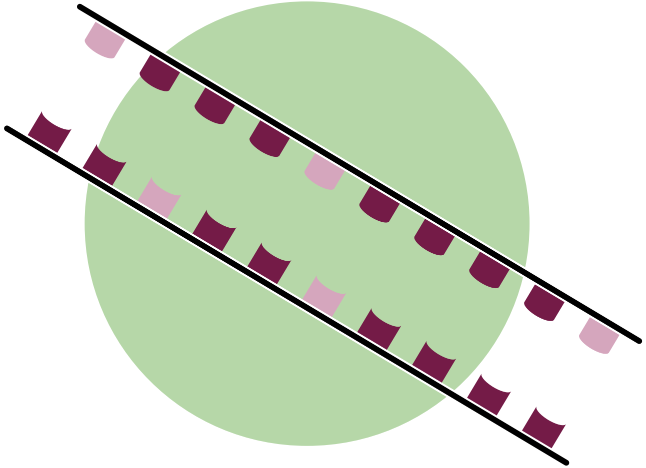
unphased

€

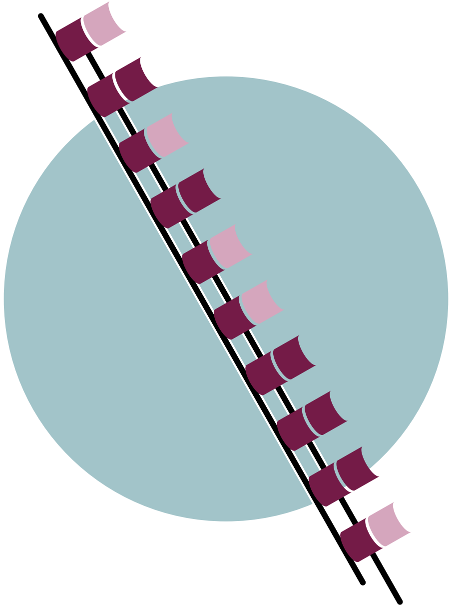


phased

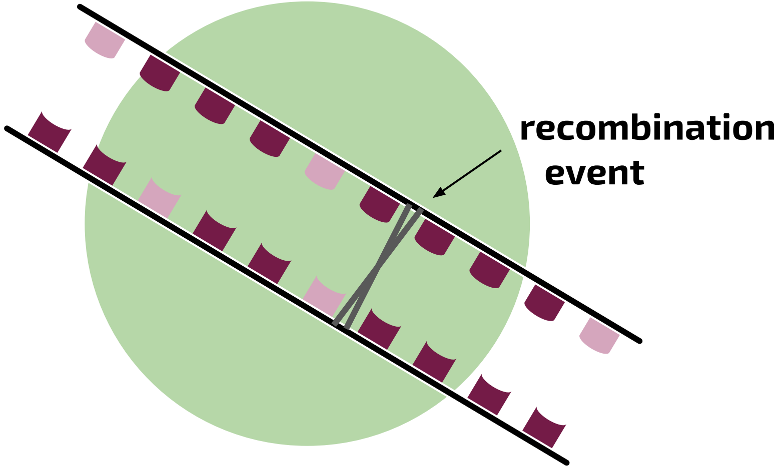
€€€

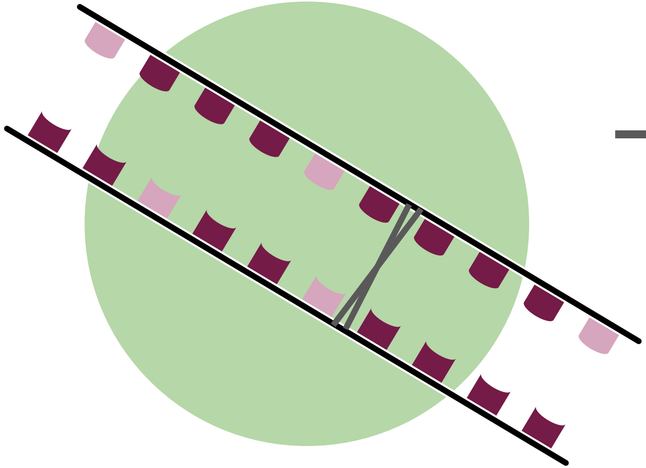


unphased

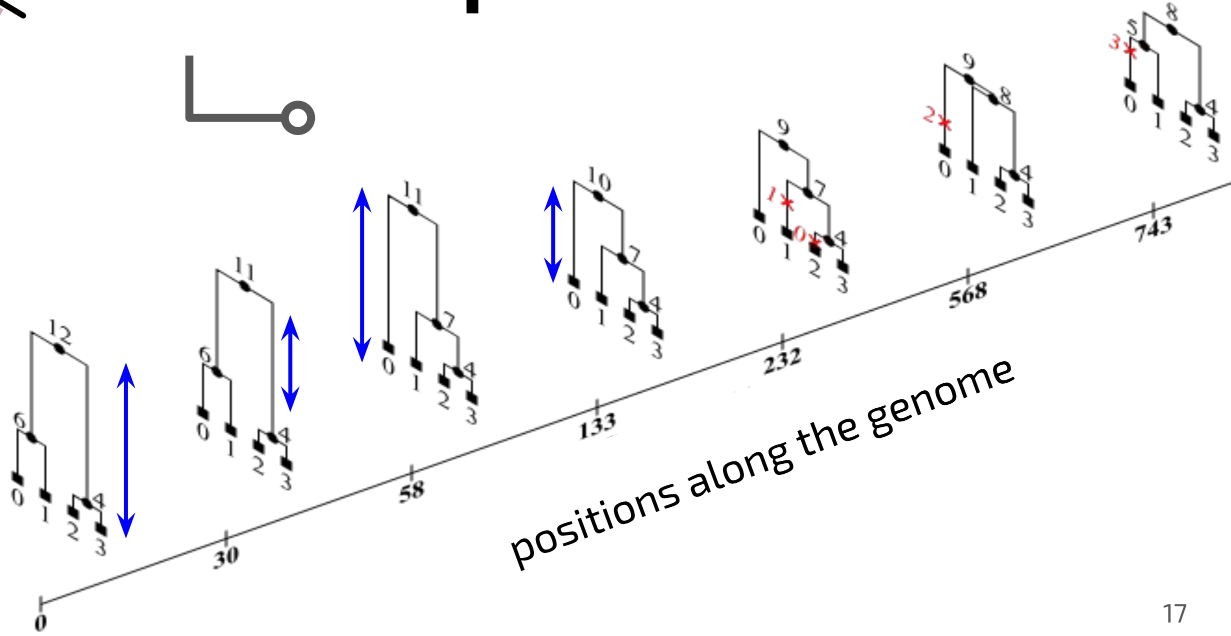


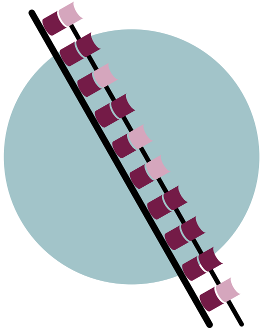
phased





tree sequence

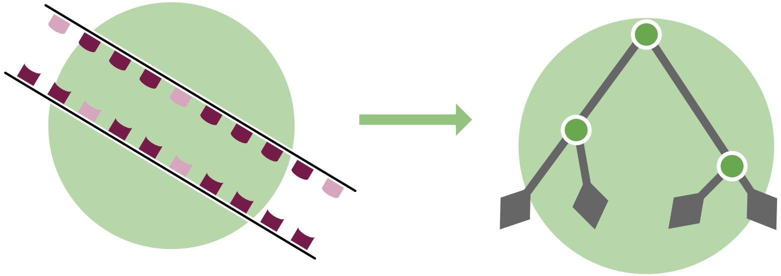




genotypic
statistics

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)

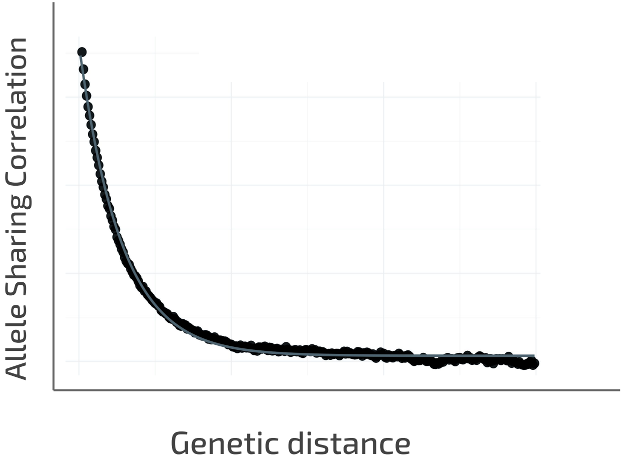
+ subtracted
between - E(within1, within2)

genotypic statistics

within-pop —— between

➔ ***ASCEND***

Tournebize, Chu, Moorjani (2022)

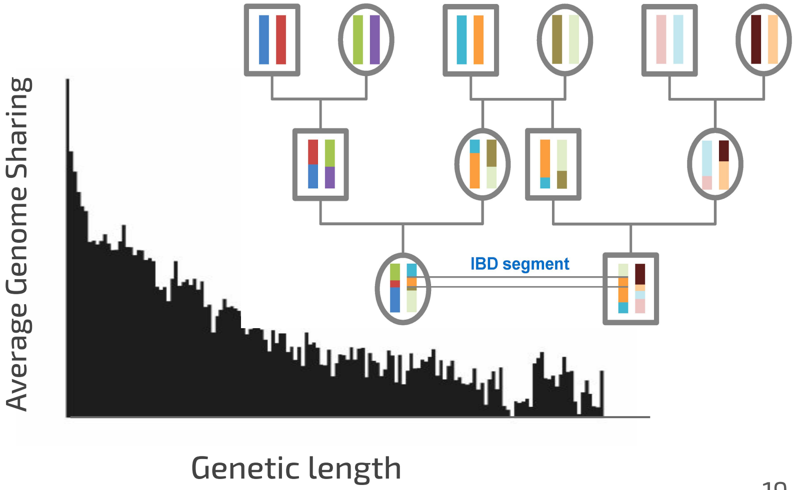


genealogical statistics

within-pop —— between

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

➔ **Distribution of IBD lengths**



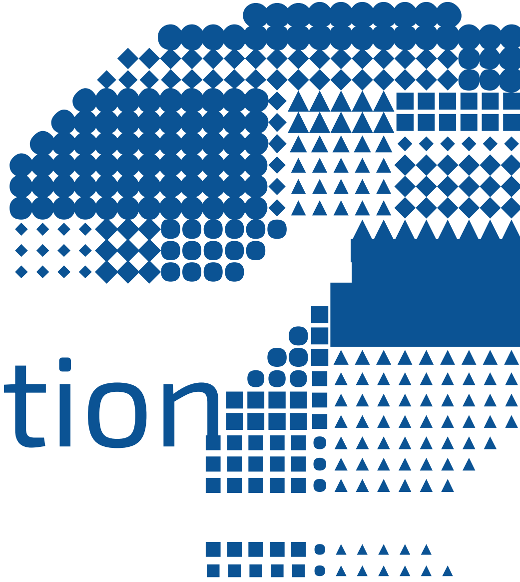
+ subtracted

how well ≠ summary statistics

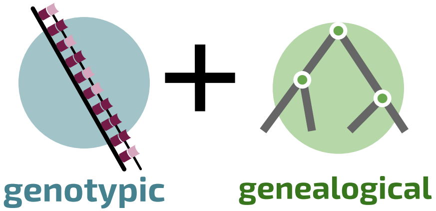
can help us

detect

very recent fragmentation



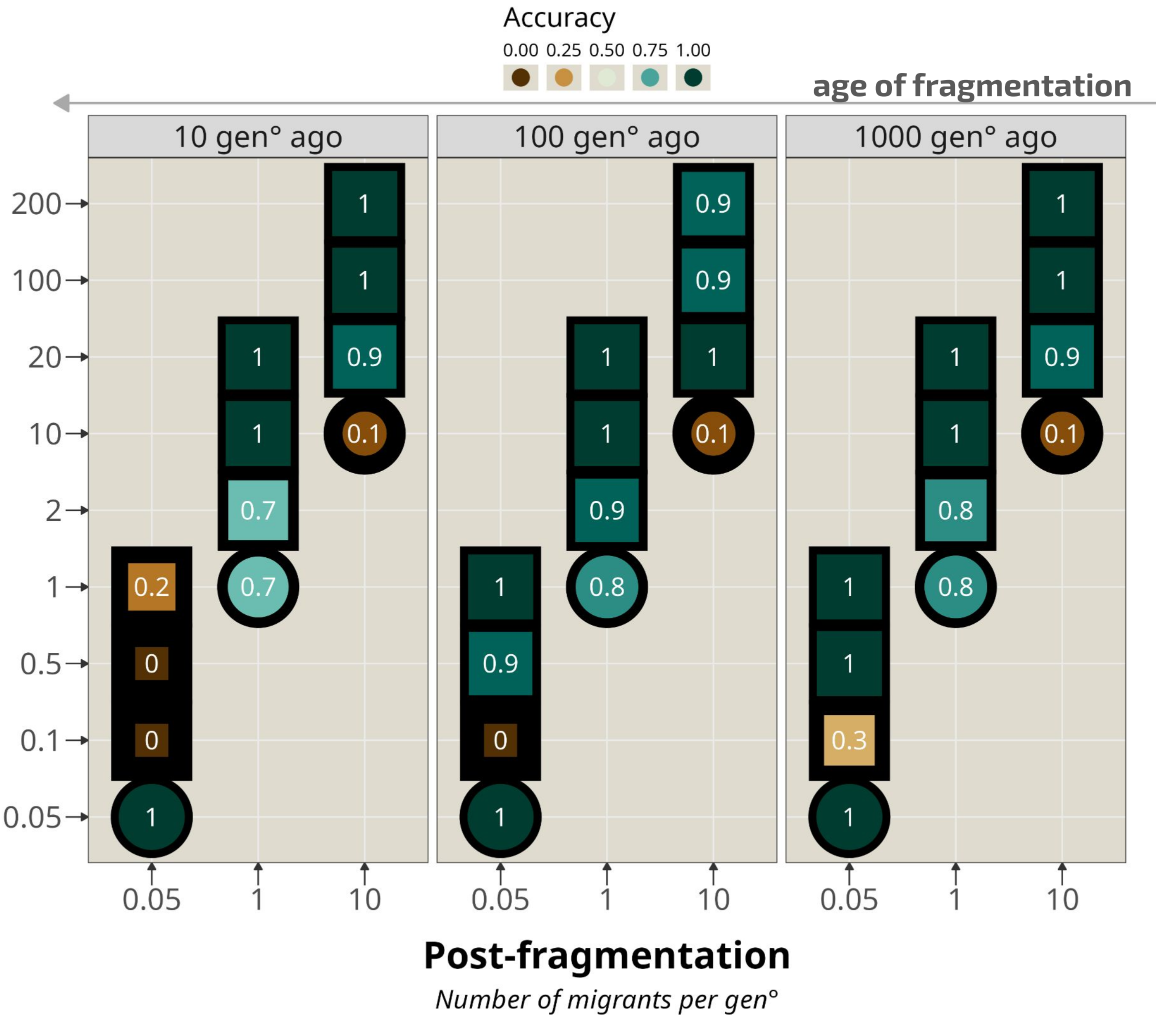
Detection power



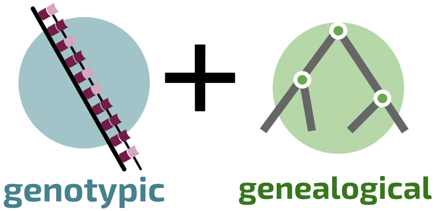
generally a great **power** to detect **fragmentation**

☐ fragmentation

○ stationary



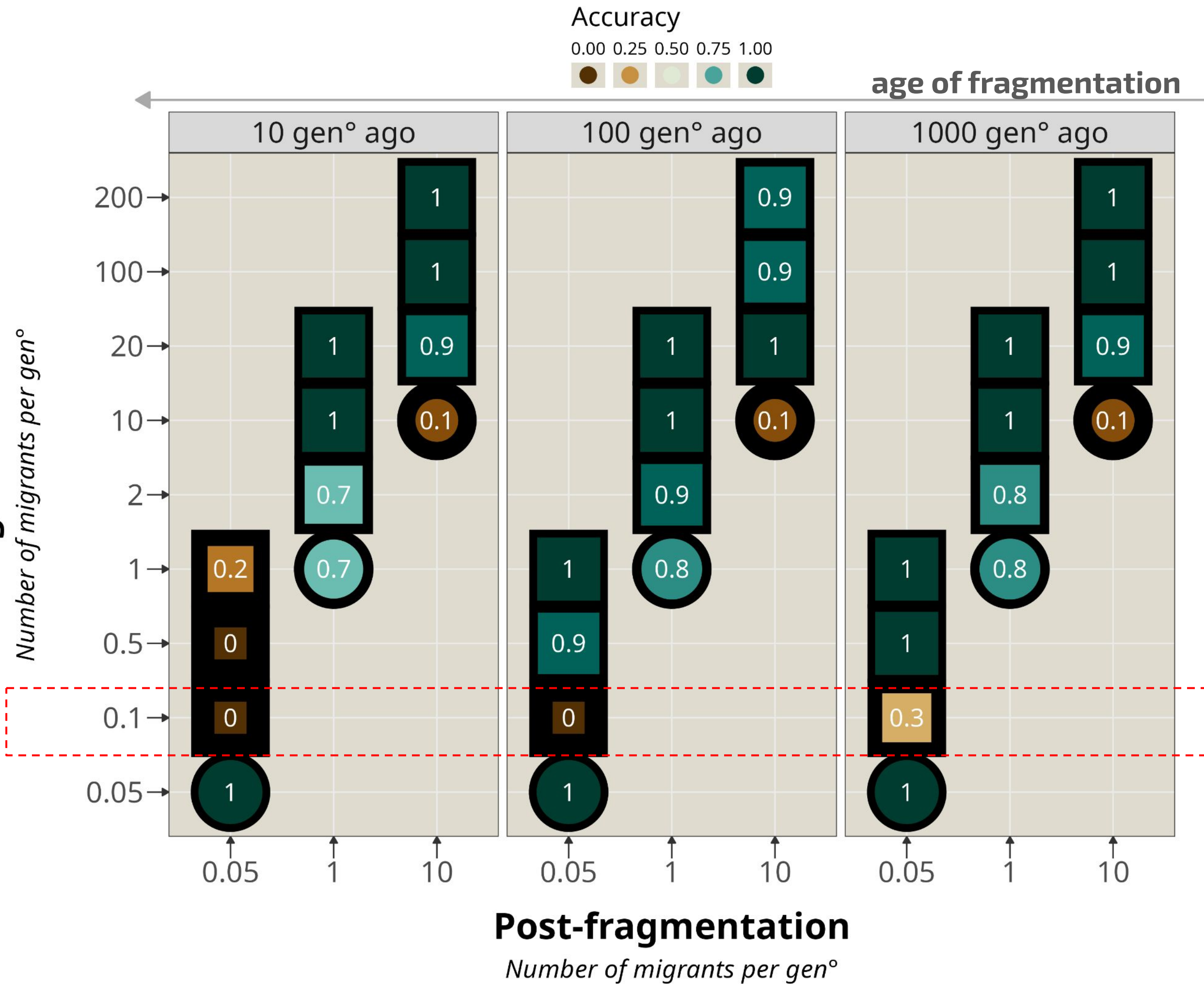
Detection power



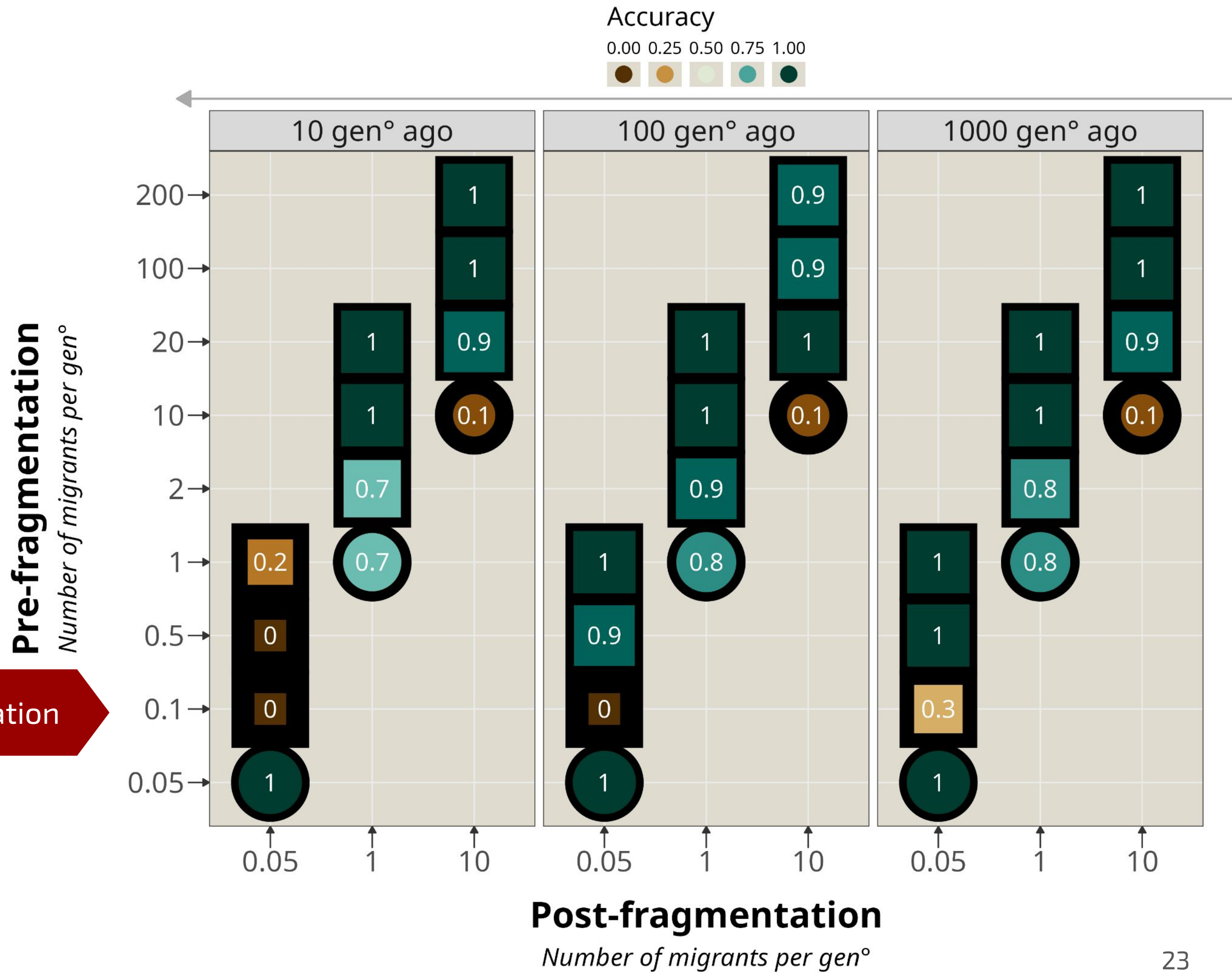
limited power to detect **weak** reduction when pops **already weakly connected**

☐ fragmentation

○ stationary

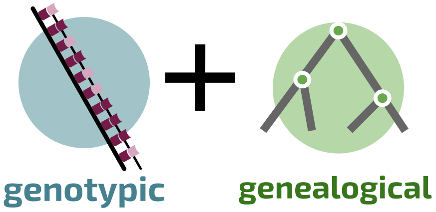


A *conservation genomic* concern

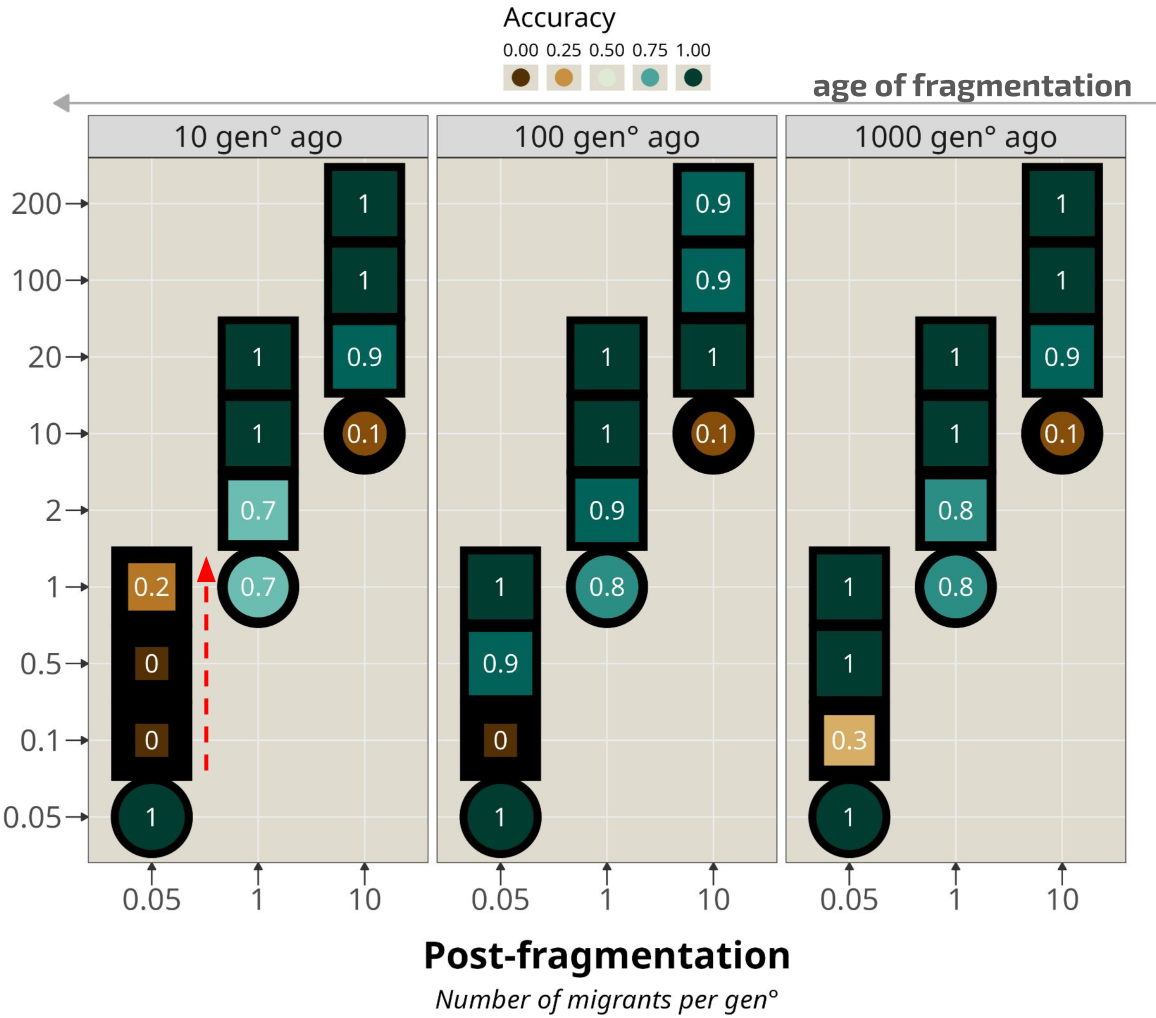


likely to **underdetect** weak fragmentation

Detection power



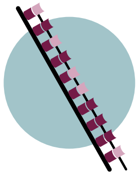
yet **power** increases as fragmentation **intensity increases**



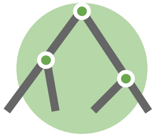
☐ `fragmentation`

○ `stationary`

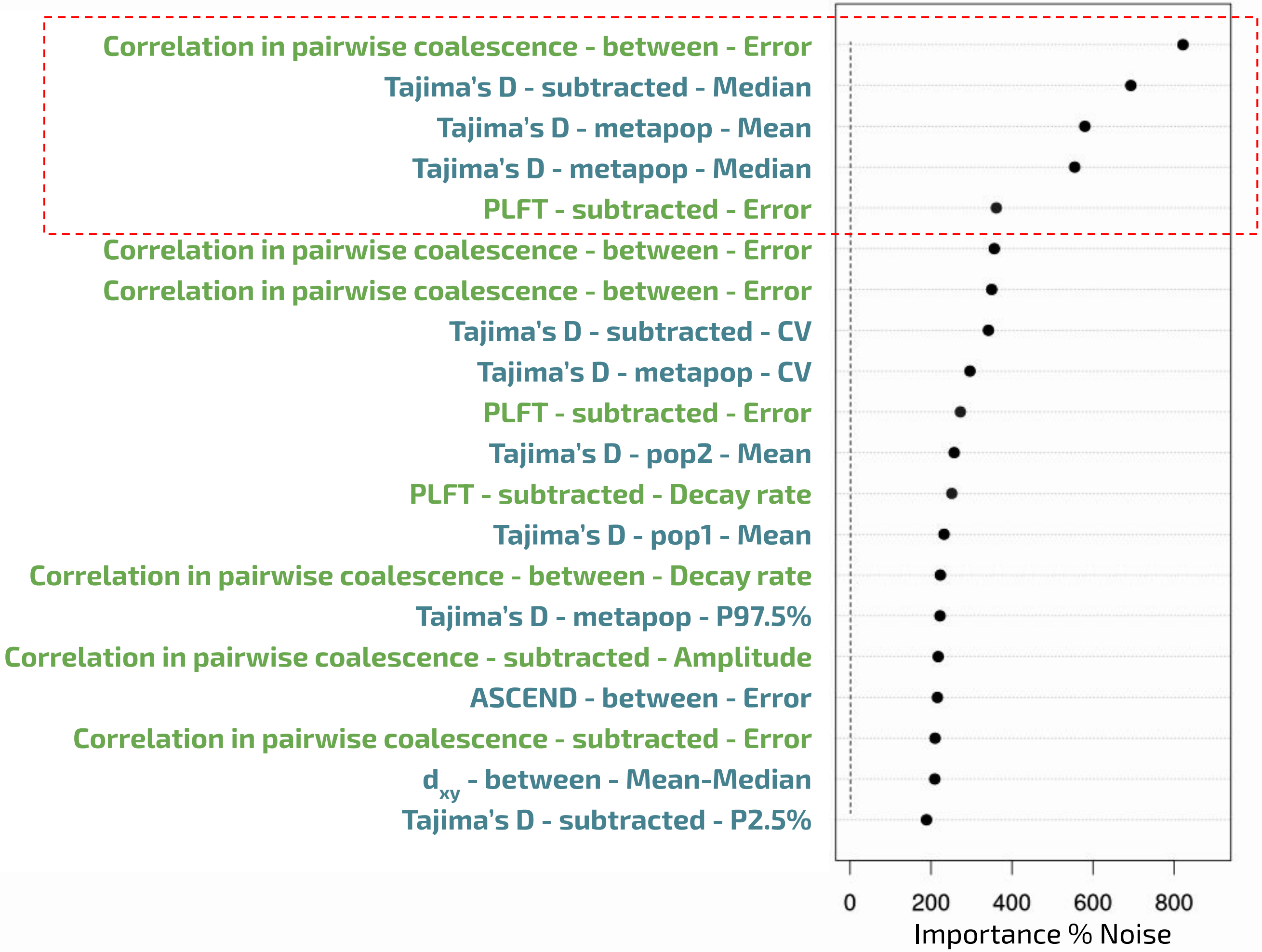
Variable importance



genotypic



genealogical



Conclusions

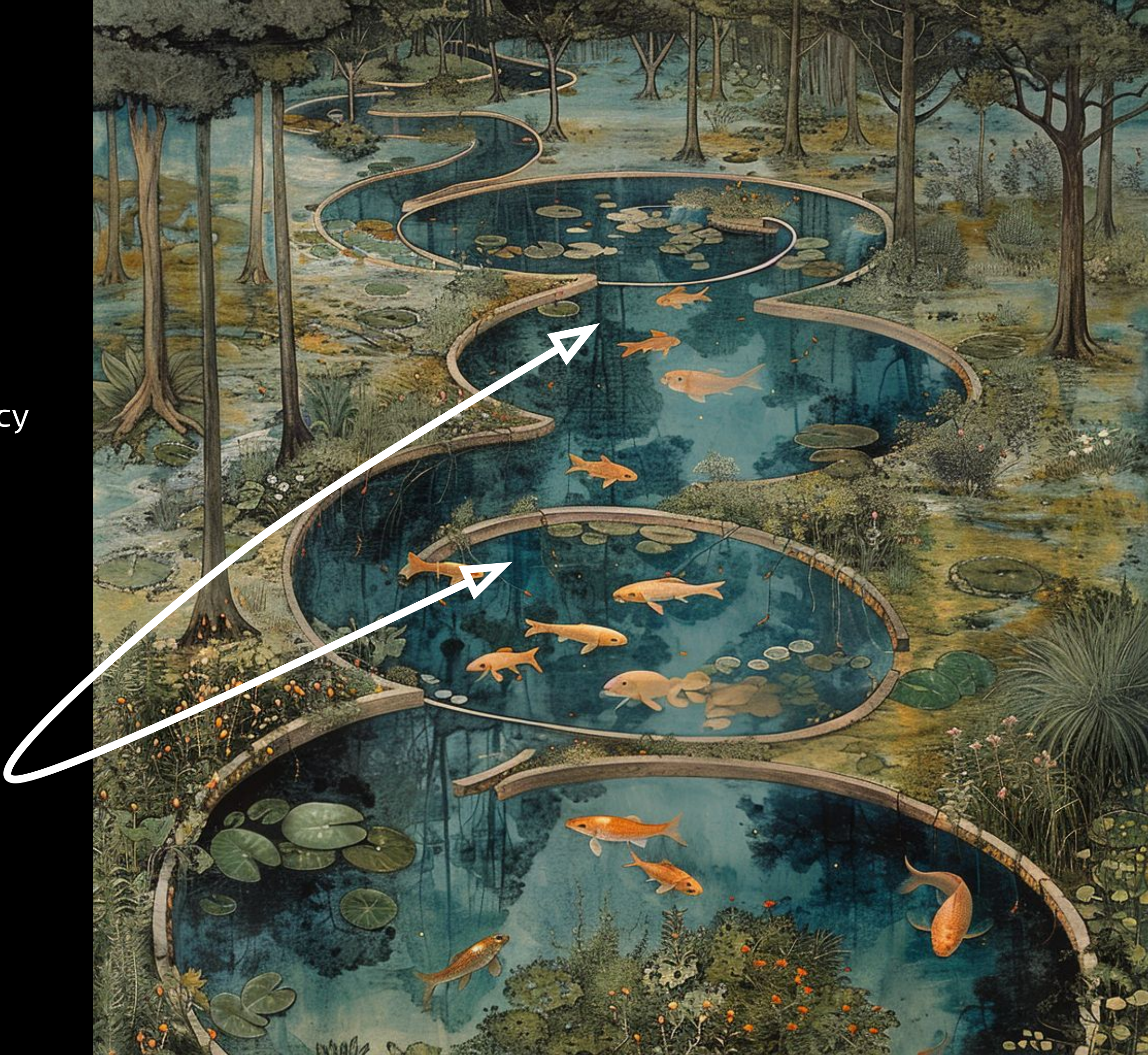
Genomic data **useful** to detect recent fragm°

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

BiodivOc
Biodiversité Occitanie

cnrs
dépasser les frontières

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Université de Toulouse

INSTITUT DE MATHÉMATIQUES DE TOULOUSE

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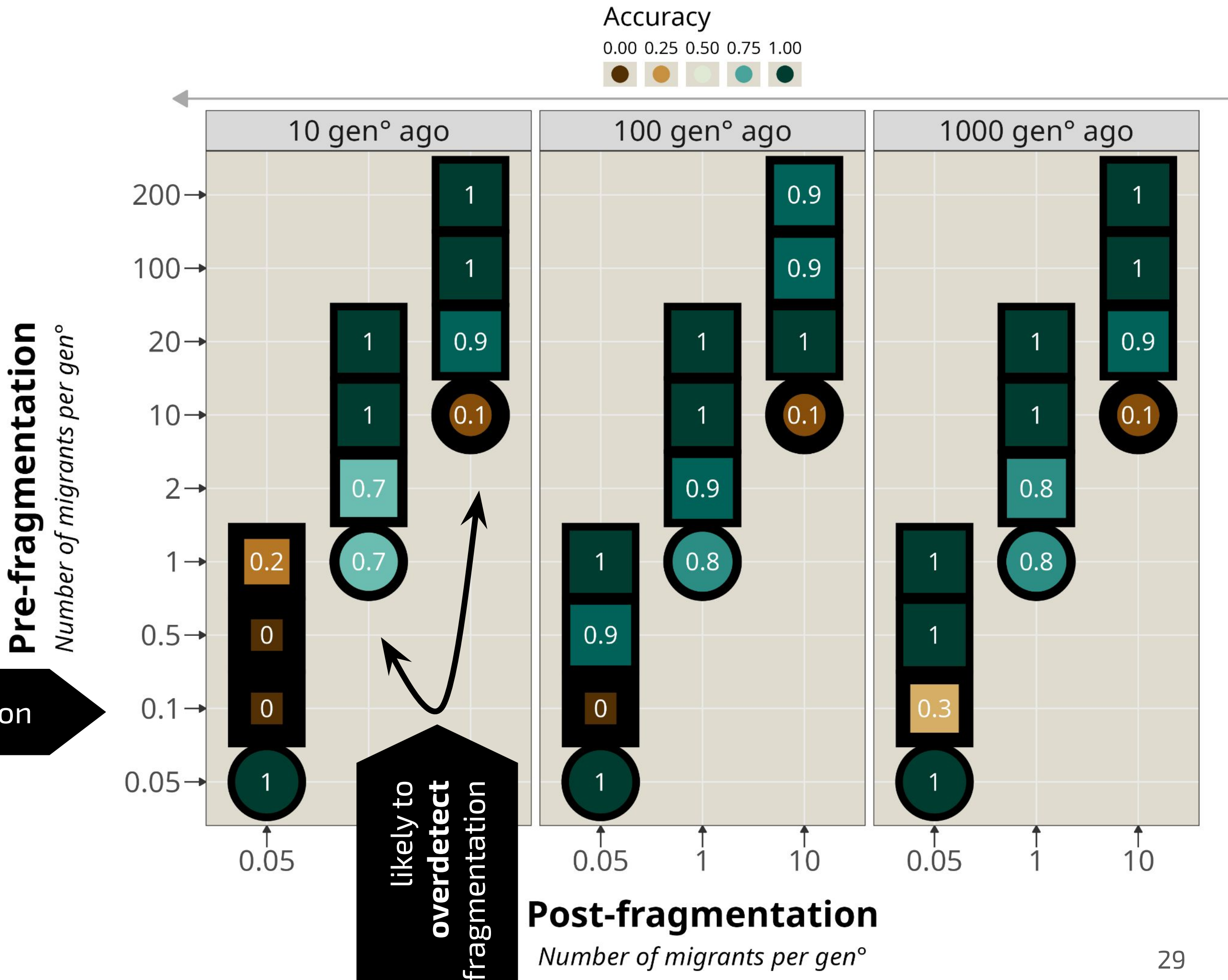
FRANCE

Appendix

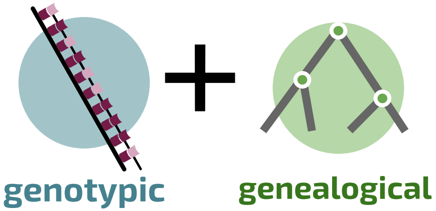
A *conservation genomic* concern



likely to **underdetect** weak fragmentation

likely to **overdetect** fragmentation

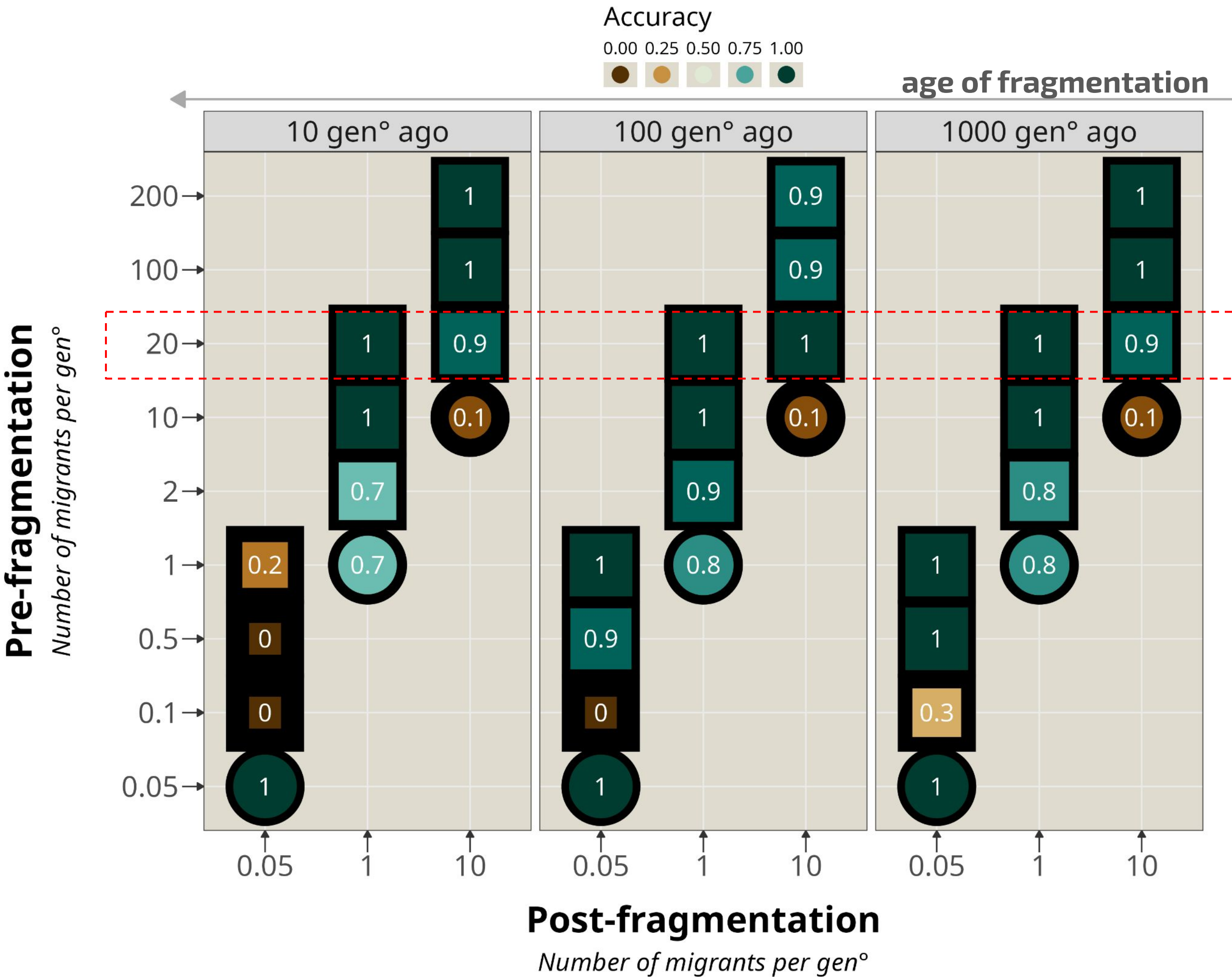
Detection power



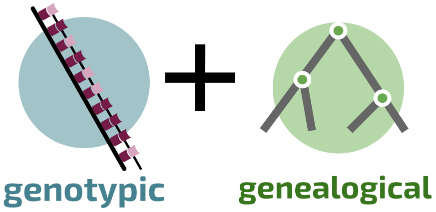
power tends to **increase** with founder **intensity**

☐ `fragmentation`

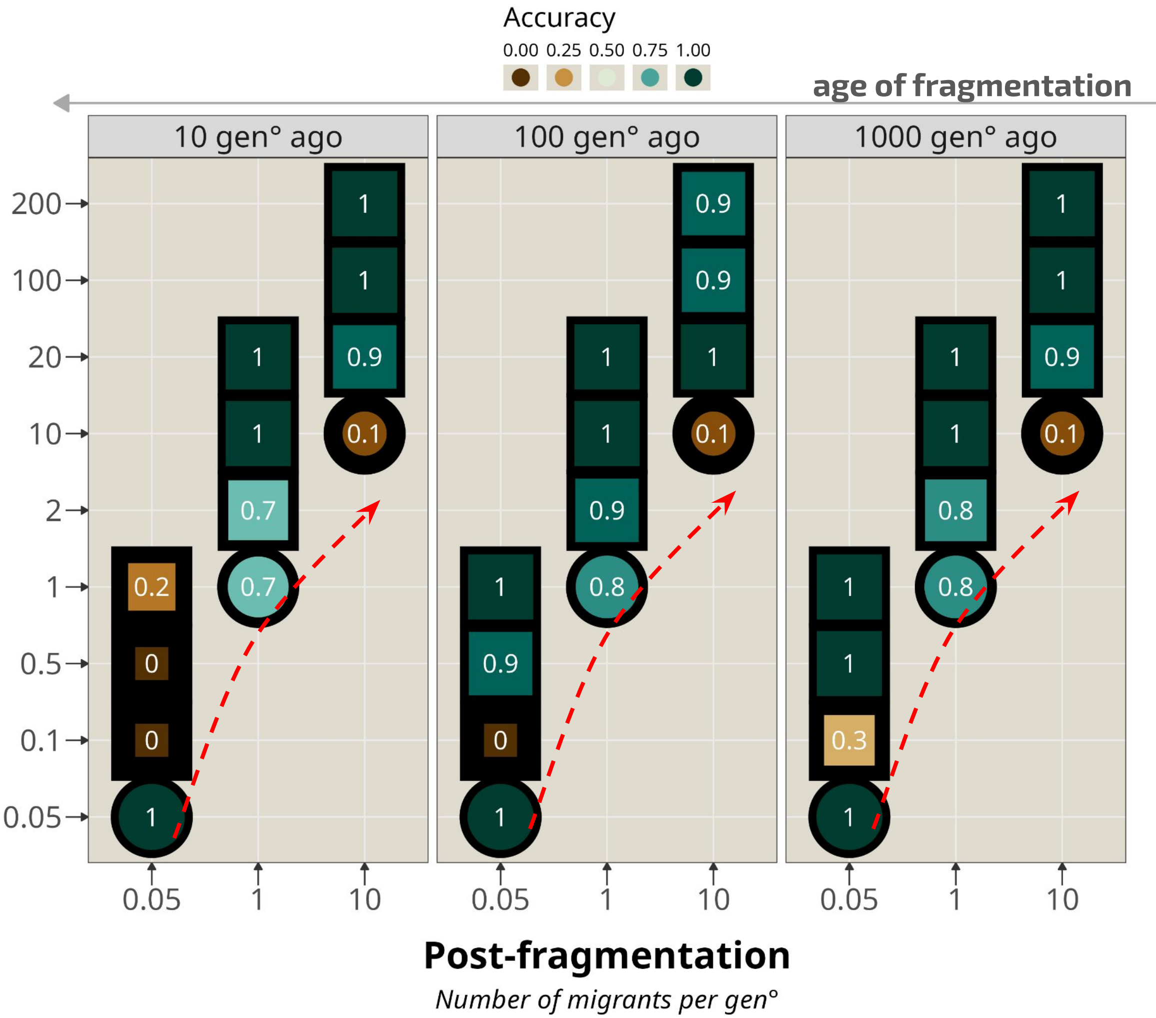
○ `stationary`



Detection power



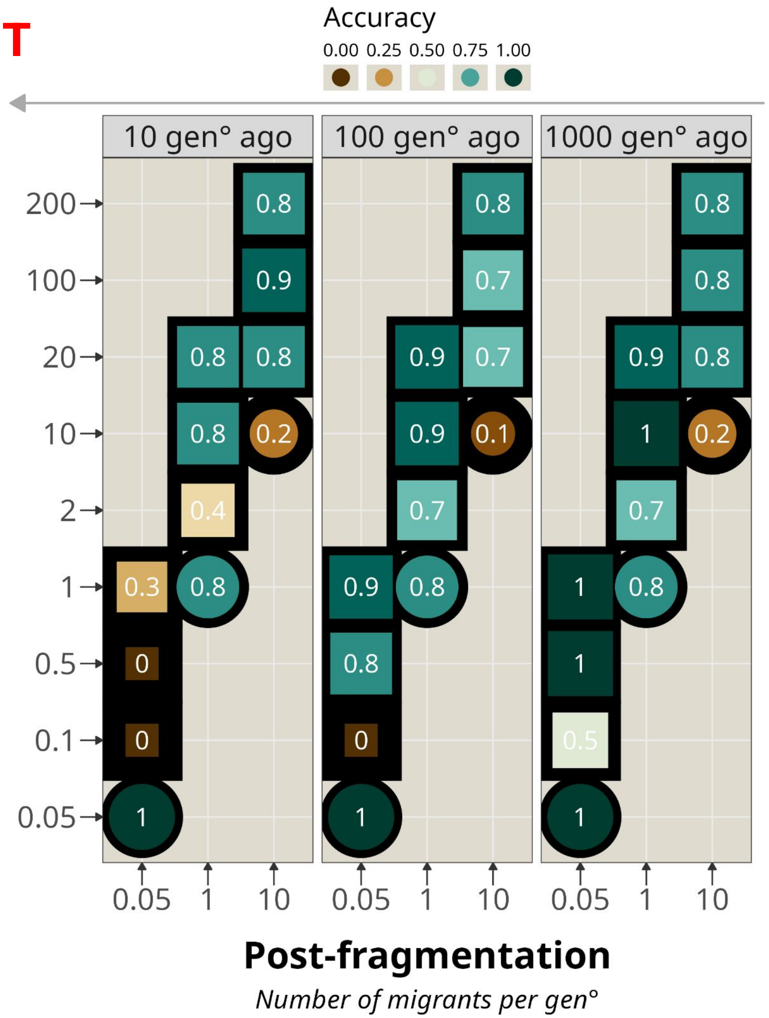
power to detect **stationary** model **decreases** with **migration rate**



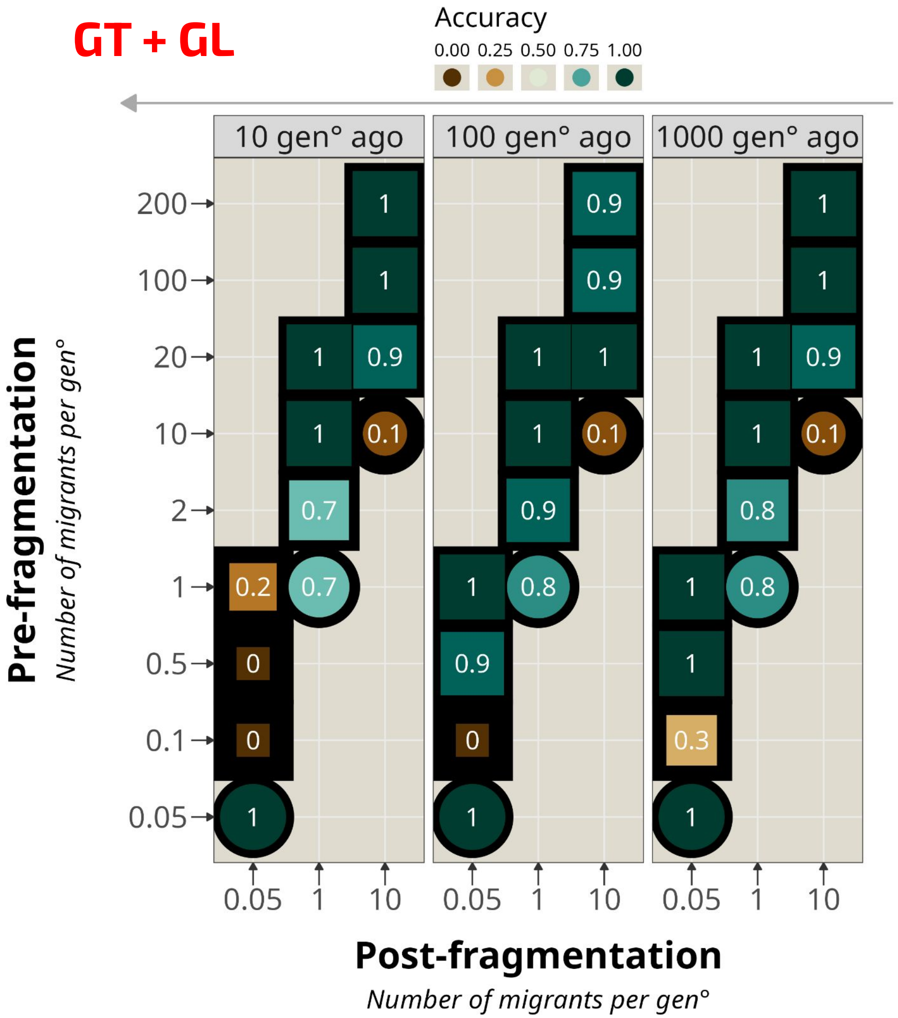
☐ fragmentation

○ stationary

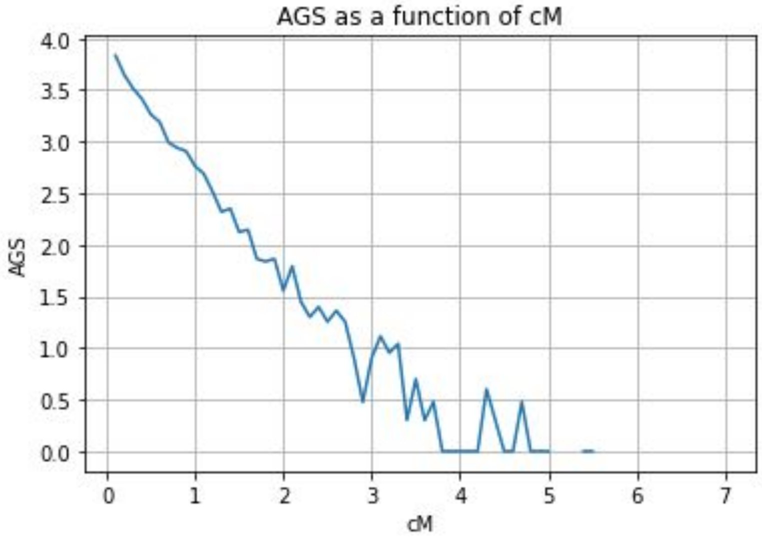
GT


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

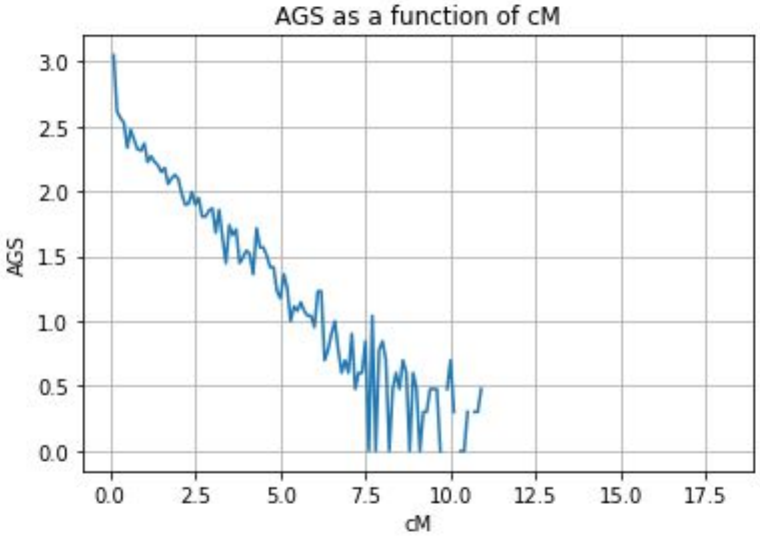
GT + GL


Accuracy
0.00
0.25
0.50
0.75
1.00
10 gen° ago
100 gen° ago
1000 gen° ago
200
100
20
10
2
1
0.5
0.1
0.05
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
0.05
1
10
0.05
1
10
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