Genetic Inference of Recent Fragmentation

Rémi Tournebize, Simon Boitard, Olivier Mazet, Lounès Chikhi

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genetic inference of recent fragmentation

Rémi Tournebize
Simon Boitard
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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

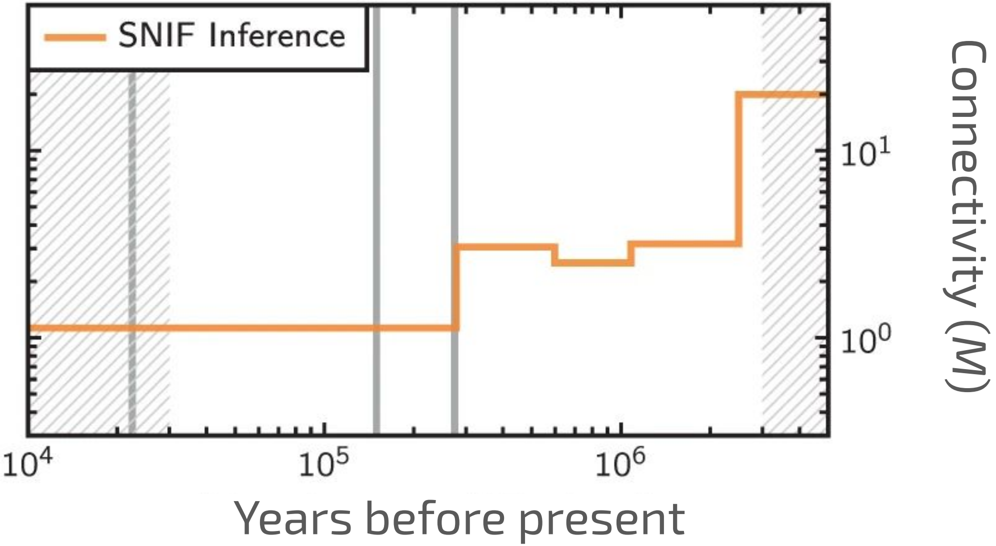
MidJourney

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

MidJourney

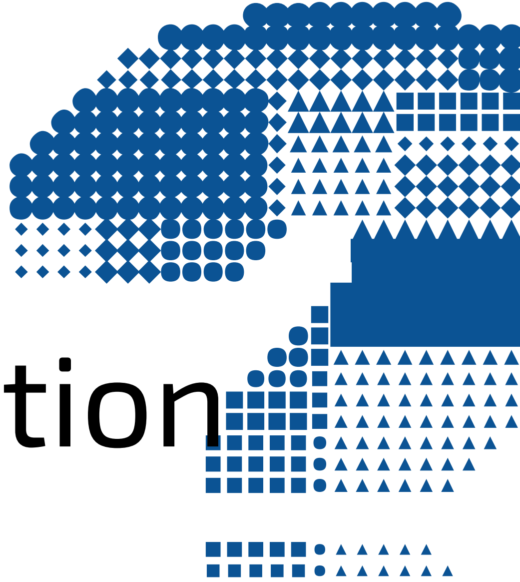
Most current genetic methods infer only **ancient** changes in connectivity

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

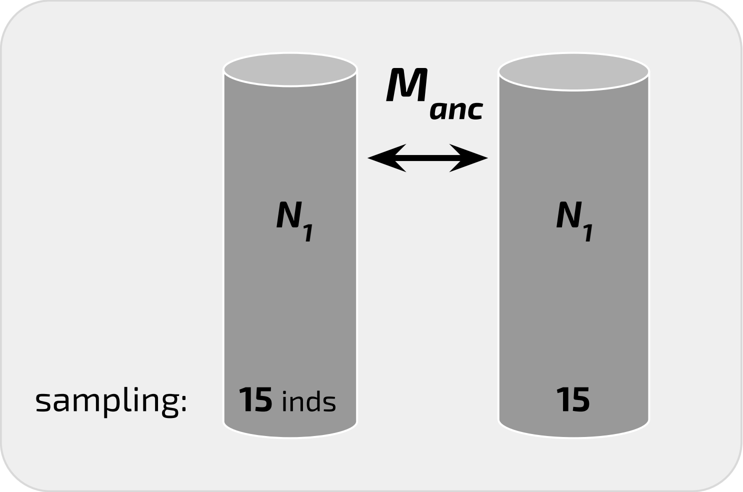
detect

very recent

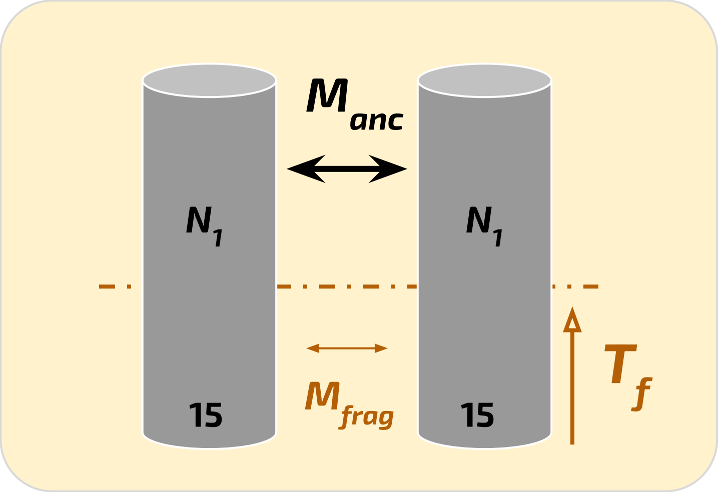
fragmentation

A study using **simulations**

stationary
model



fragmentation
model

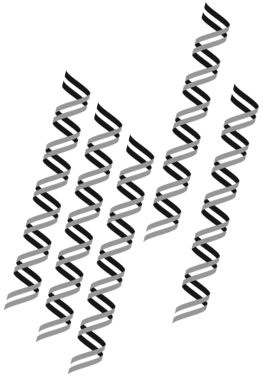


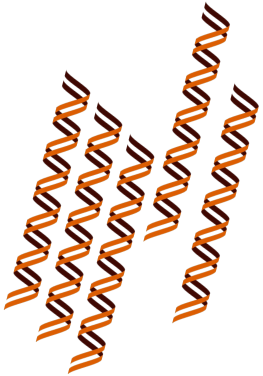
stationary model

fragmented model

2,500 simulated

genetic data per model

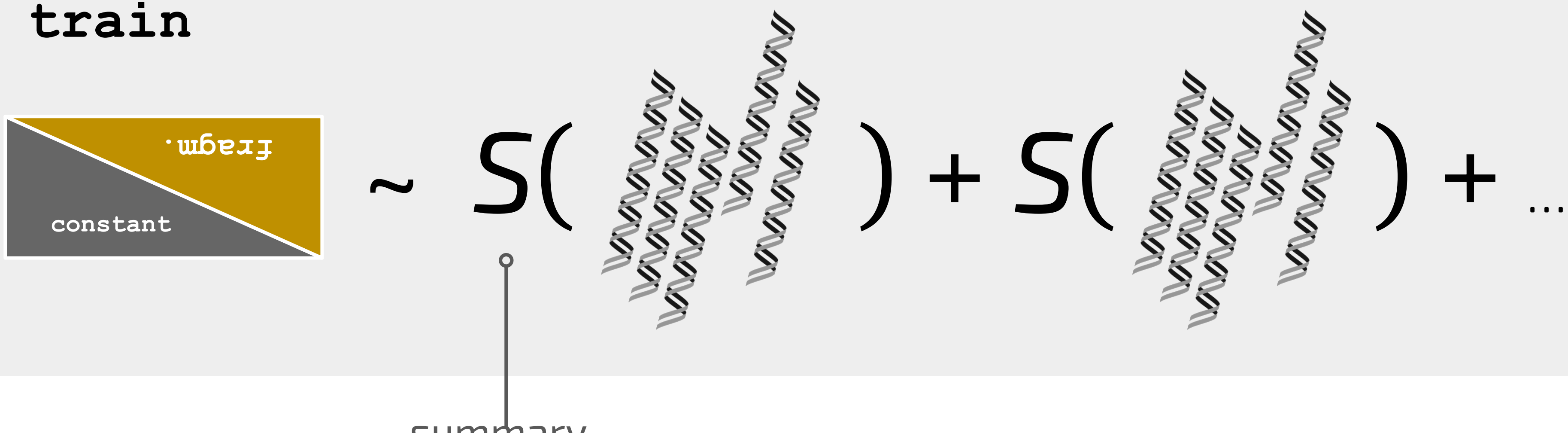




G=5×25 Mbp

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

`train`



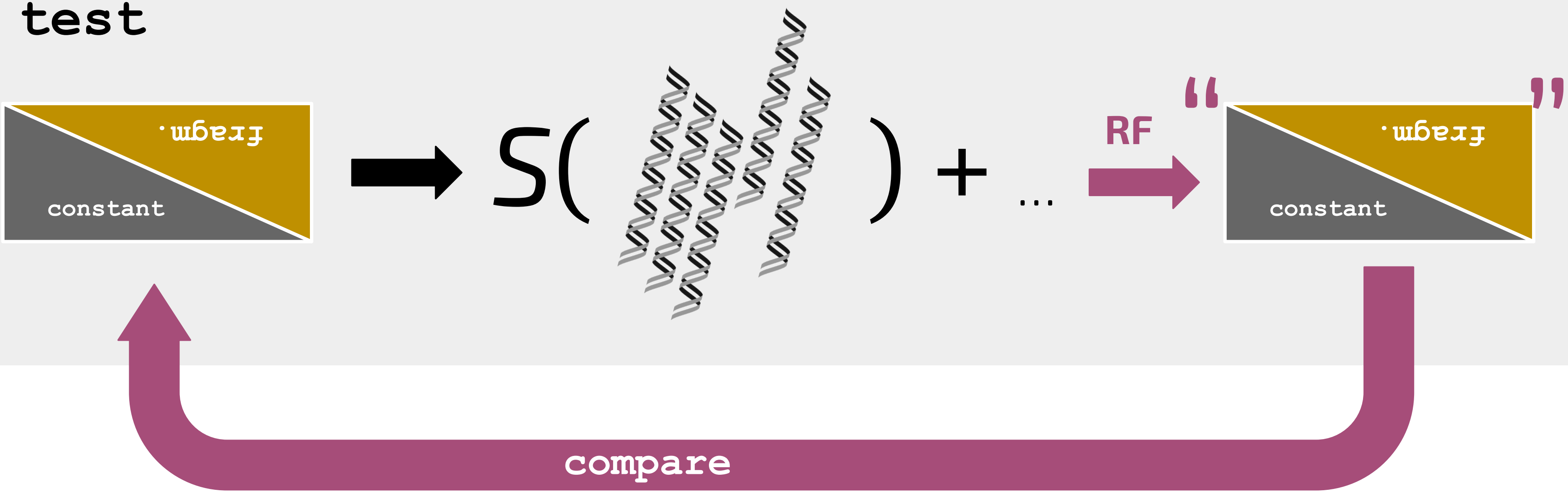
`fragm.`

`constant`

$\sim S(\quad) + S(\quad) + \ldots$

summary statistics

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



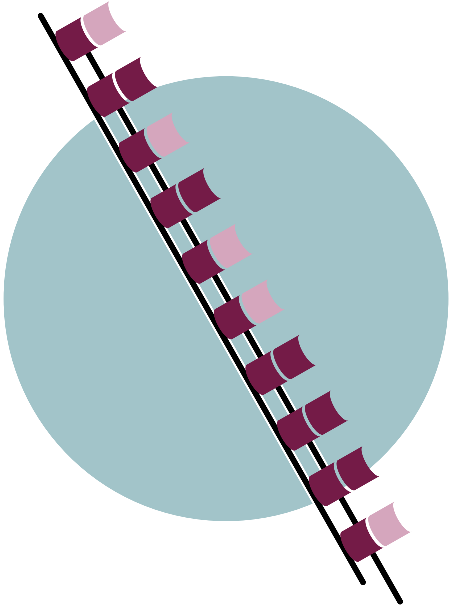
using **different**

types of

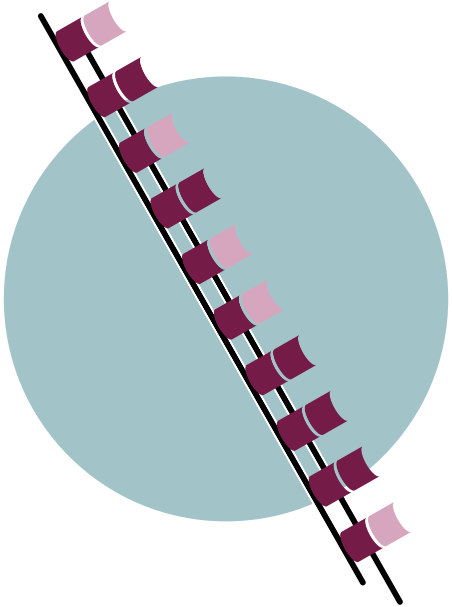
genomic data

types of genomic **data**

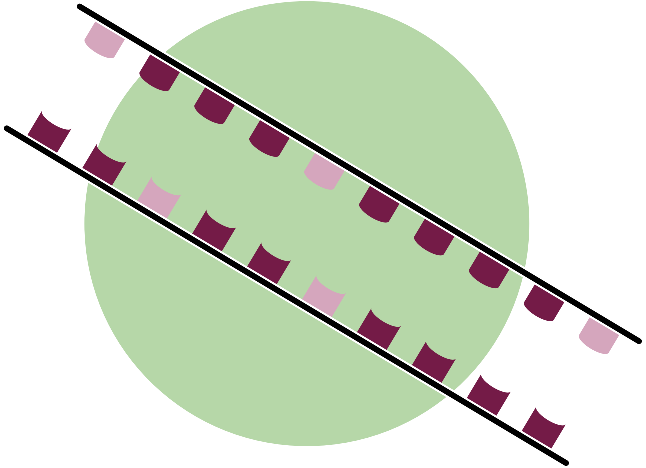
`unphased`



unphased

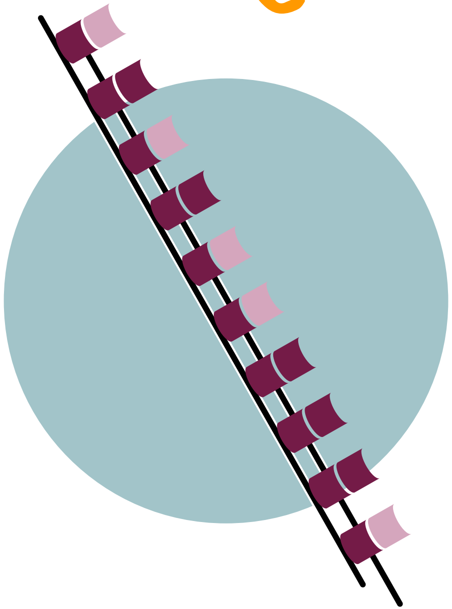


phased



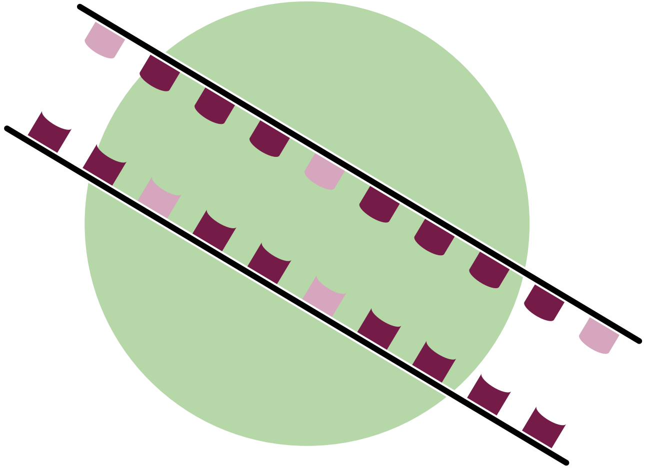
unphased

€

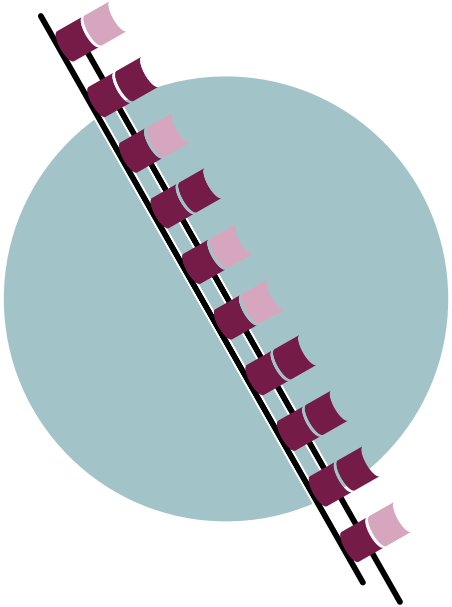


phased

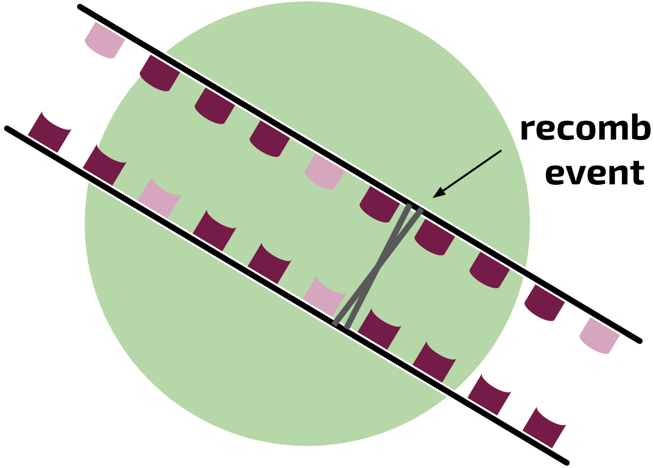
€€€



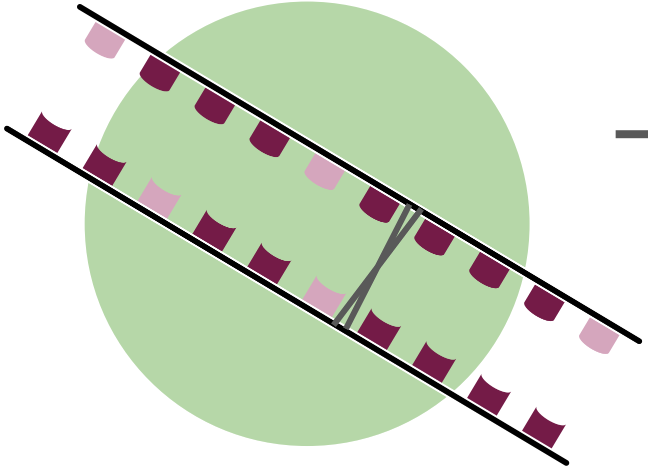
unphased



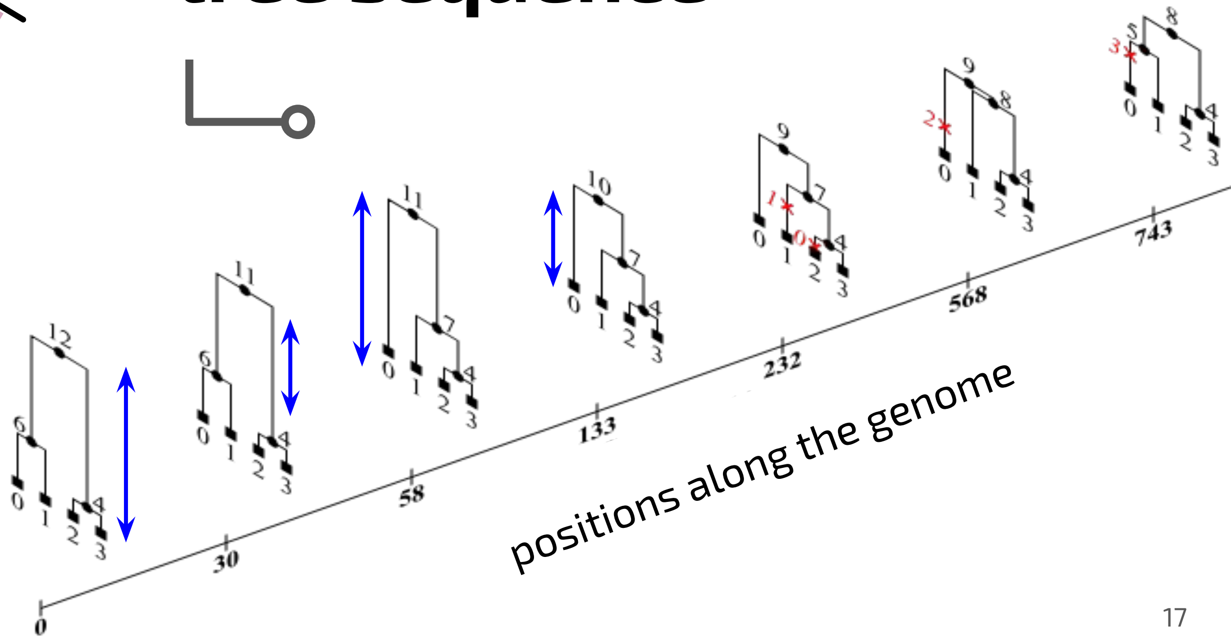
phased

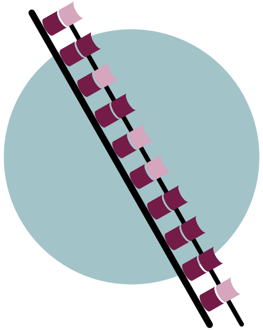


recombination
event



tree sequence

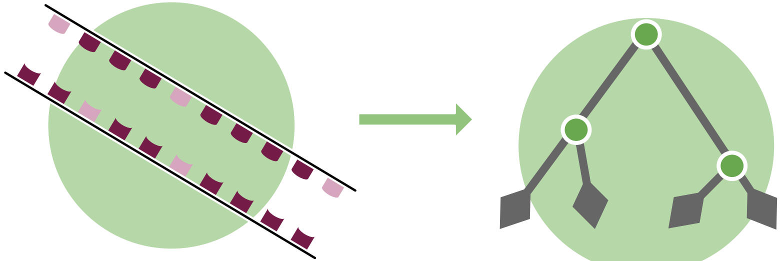




genotypic
statistics

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)

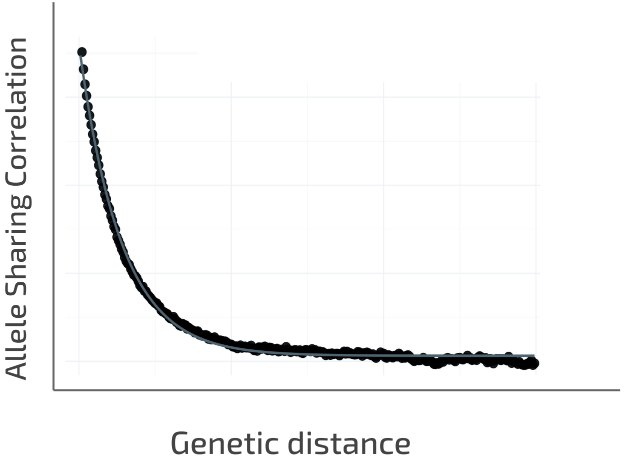
+ **subtracted**
between - E(within1, within2)

genotypic statistics

within-pop —— between

➔ ***ASCEND***

Tournebize, Chu, Moorjani (2022)

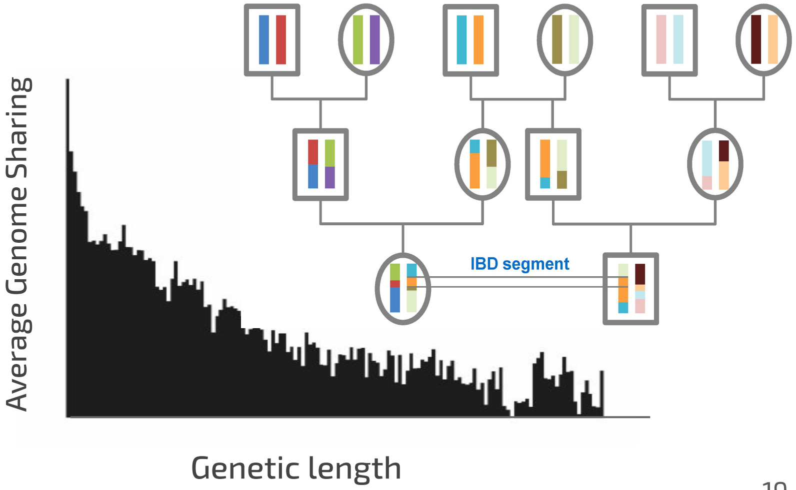


genealogical statistics

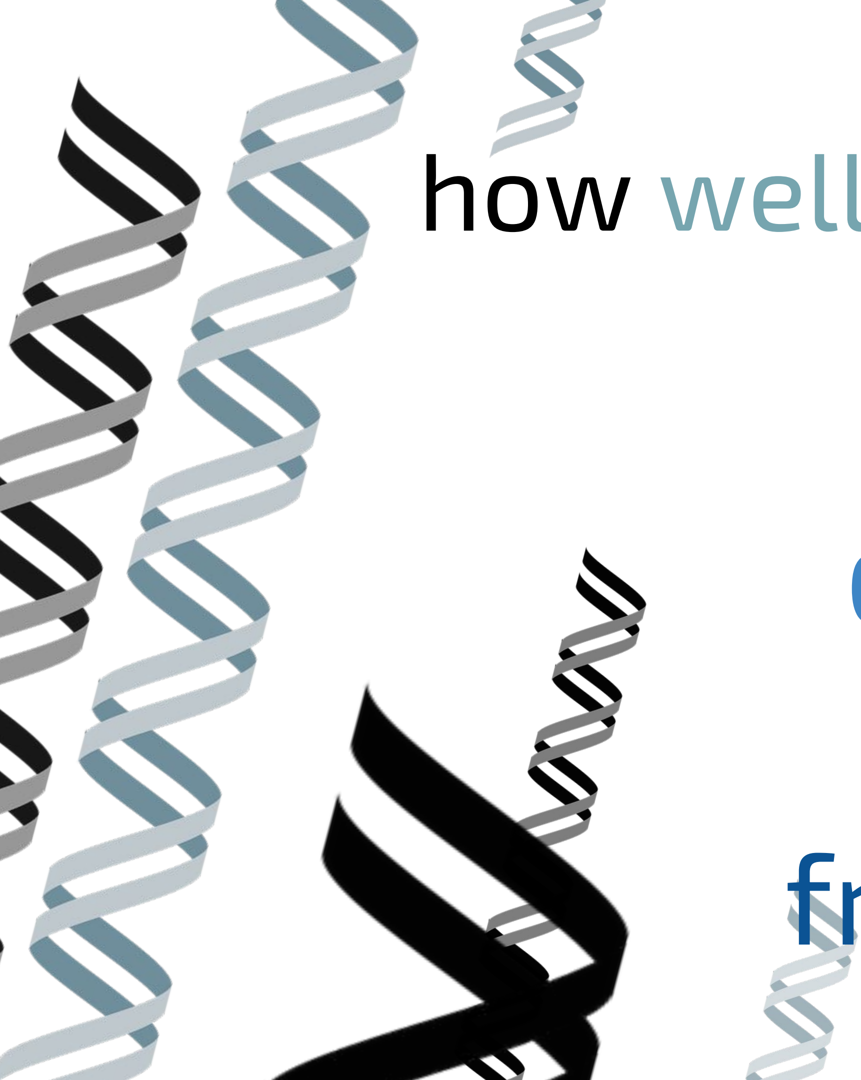
within-pop —— between

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

➔ **Distribution of IBD lengths**



+ subtracted

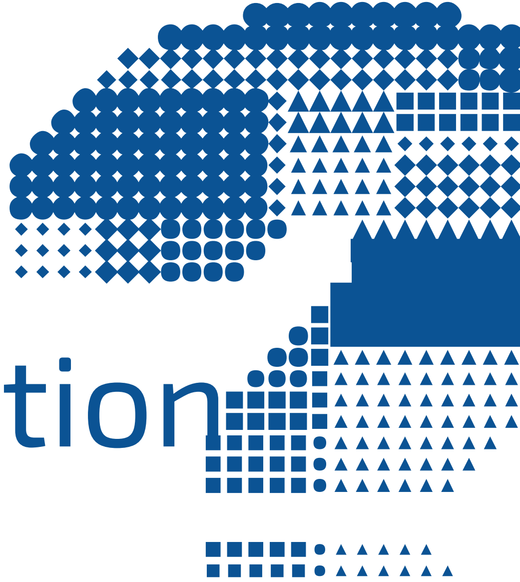


how well ≠ summary statistics

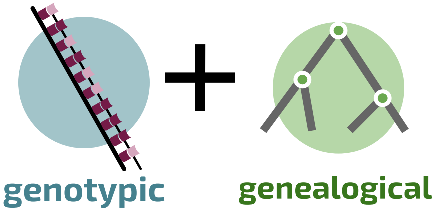
can help us

detect

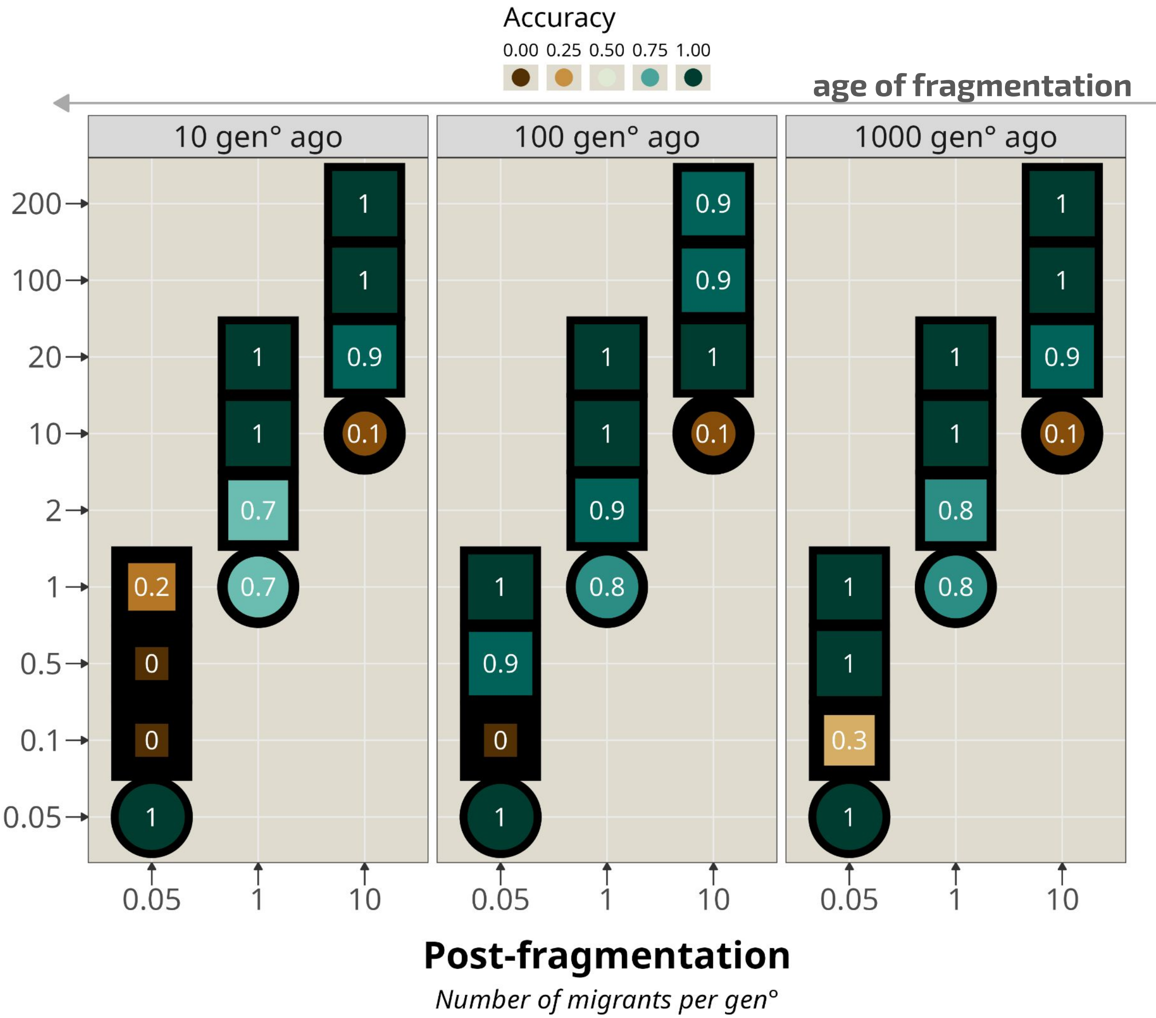
very **recent** fragmentation



Detection power



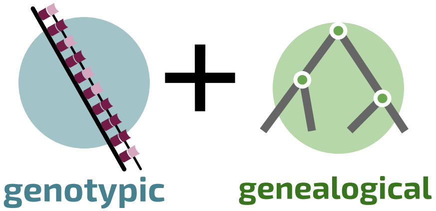
generally a great **power** to detect **fragmentation**



□ fragmentation

○ stationary

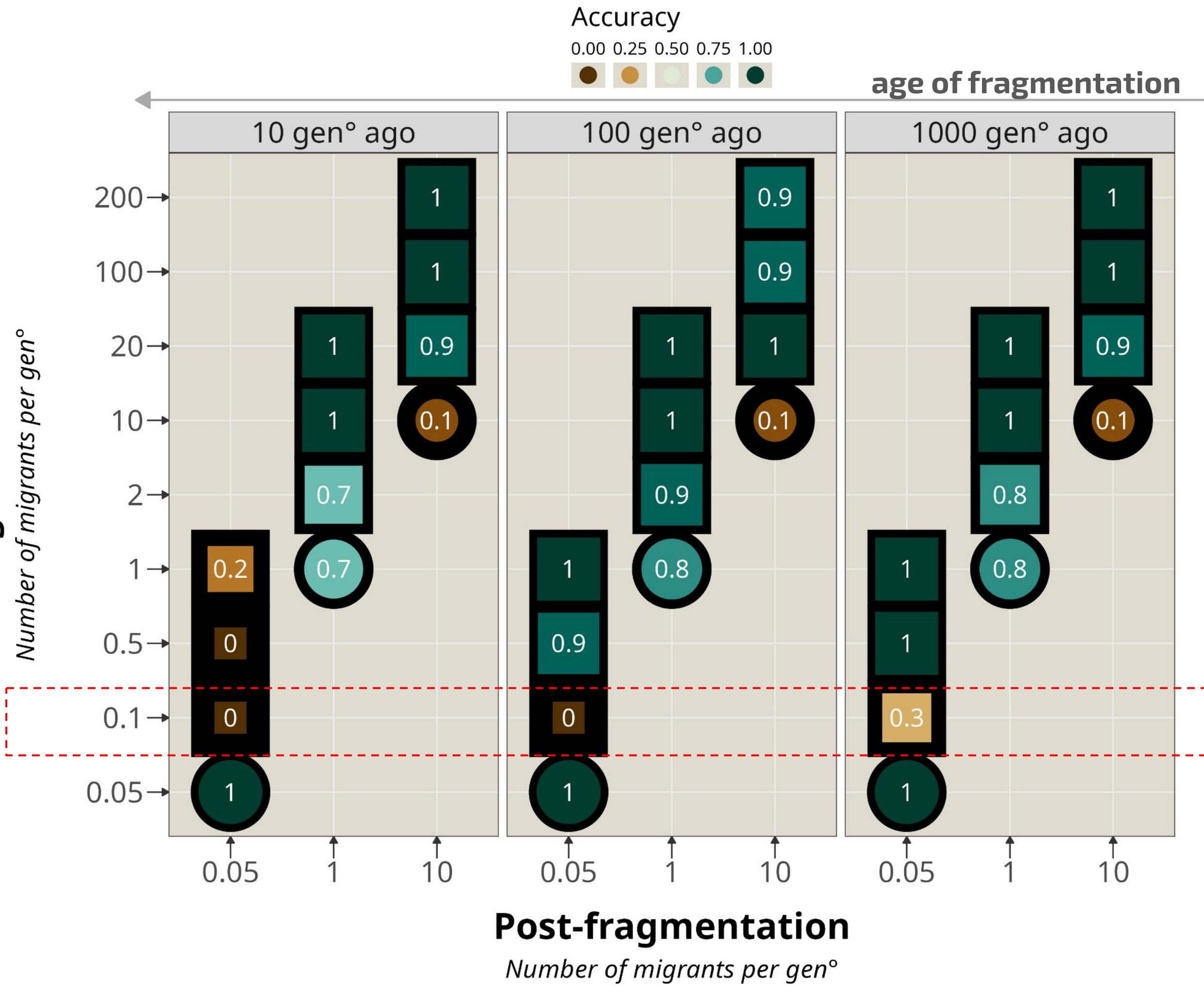
Detection power



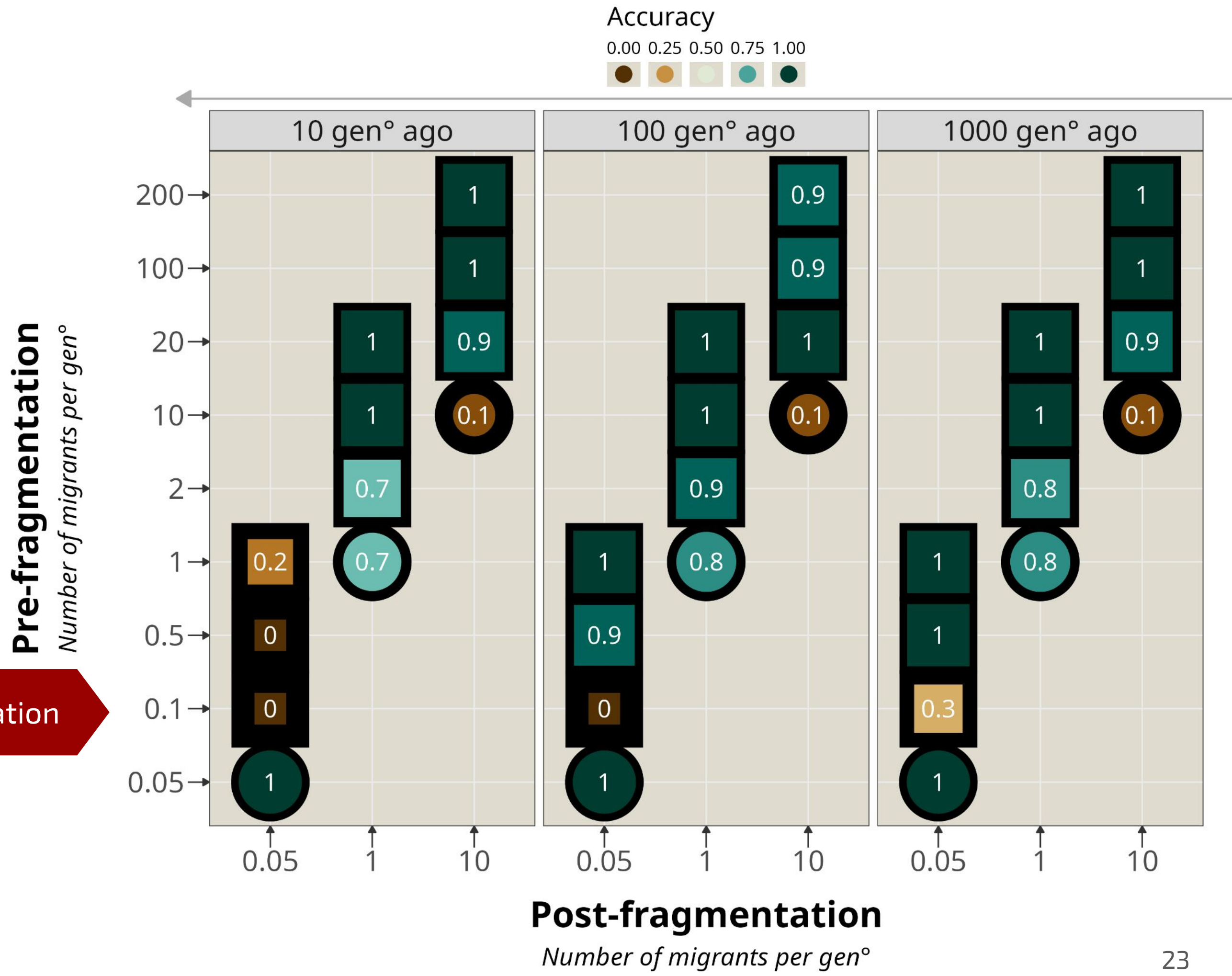
limited power to detect **weak** reduction when pops **already weakly connected**

☐ fragmentation

○ stationary

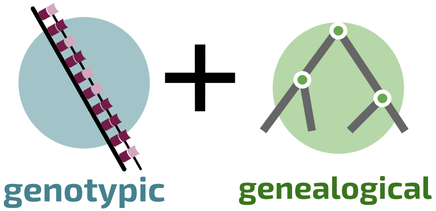


A *conservation genomic* concern



likely to **underdetect** weak fragmentation

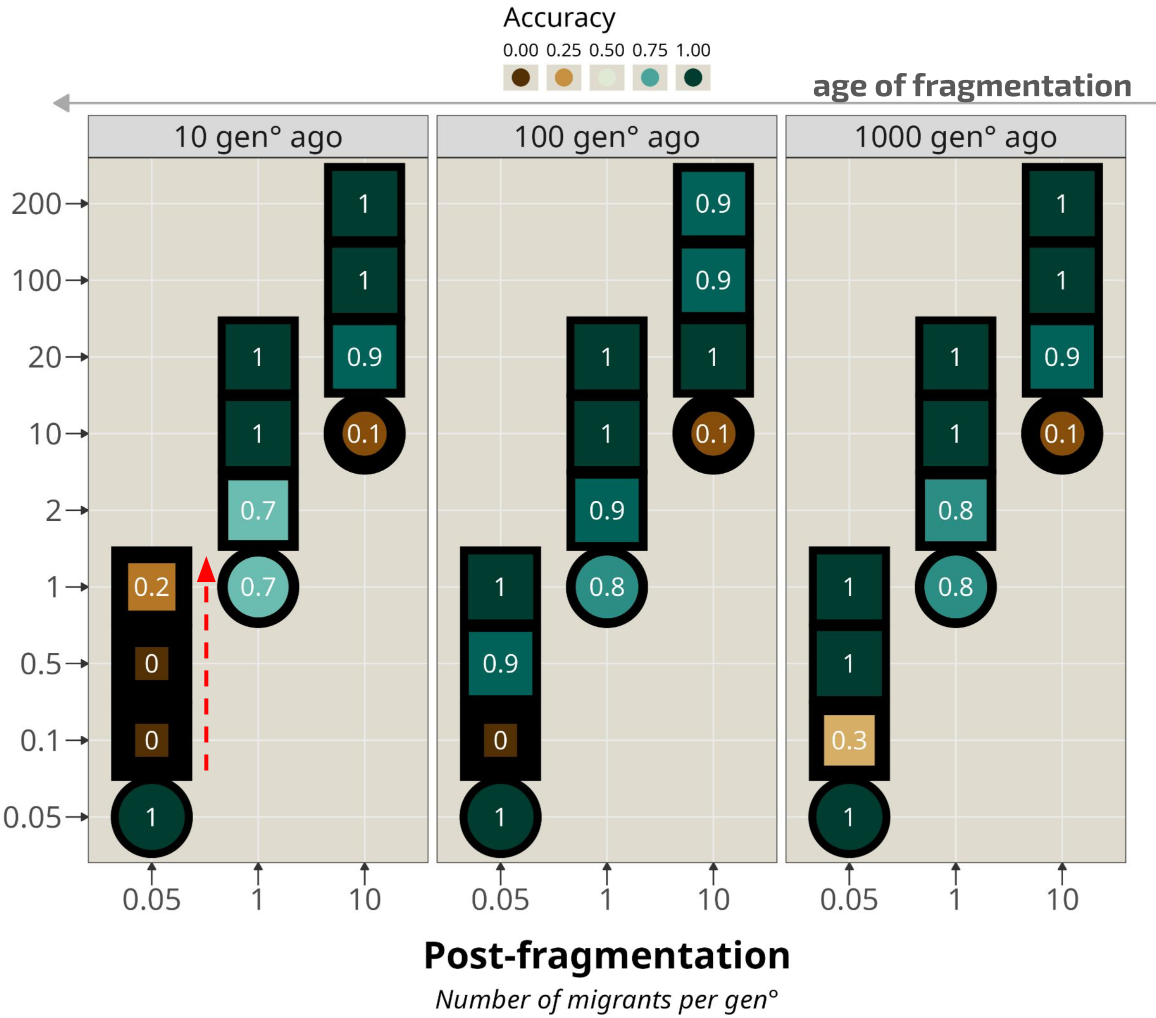
Detection power



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

☐ fragmentation

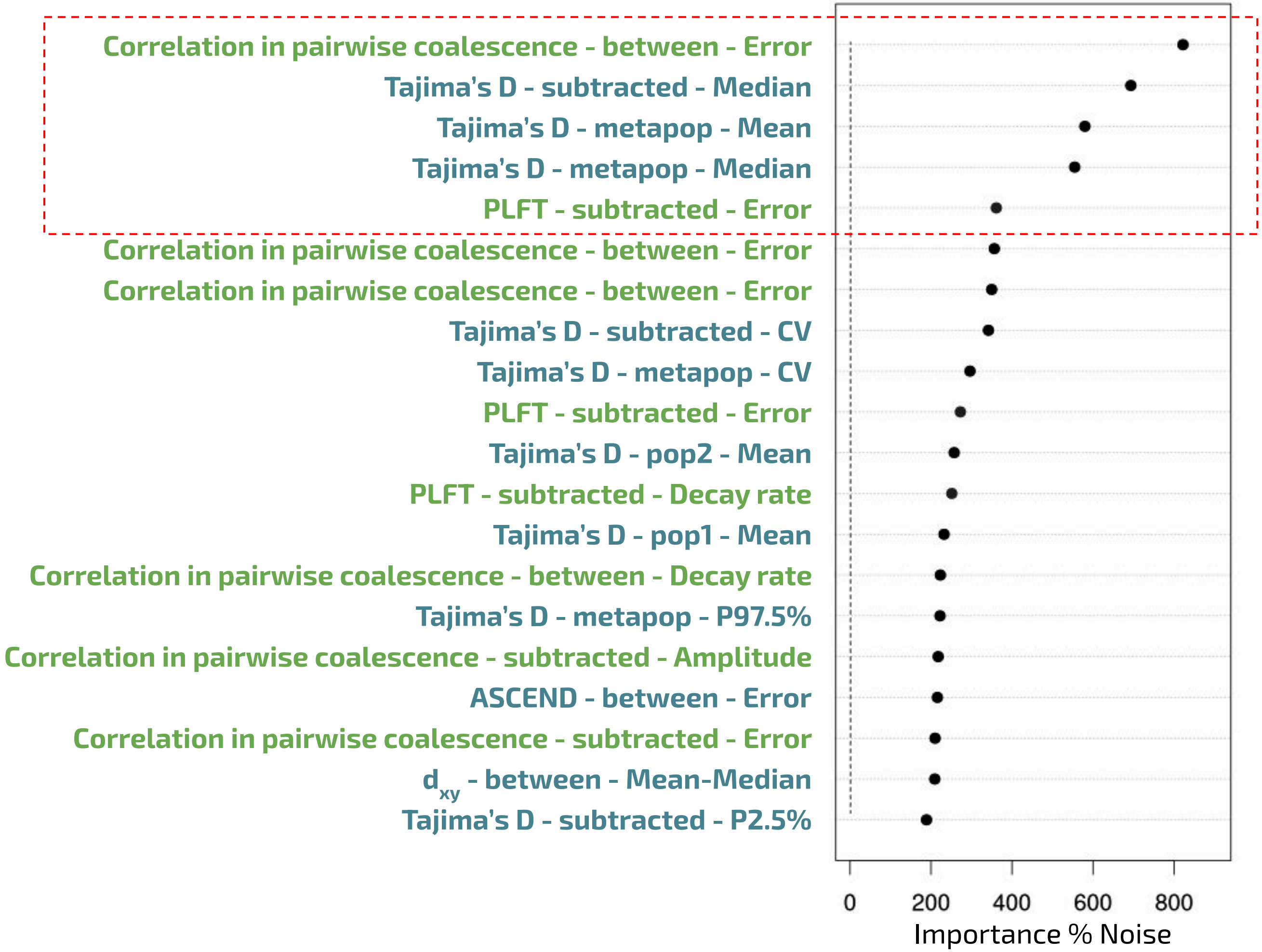
○ stationary



Variable importance

genotypic

genealogical



Conclusions

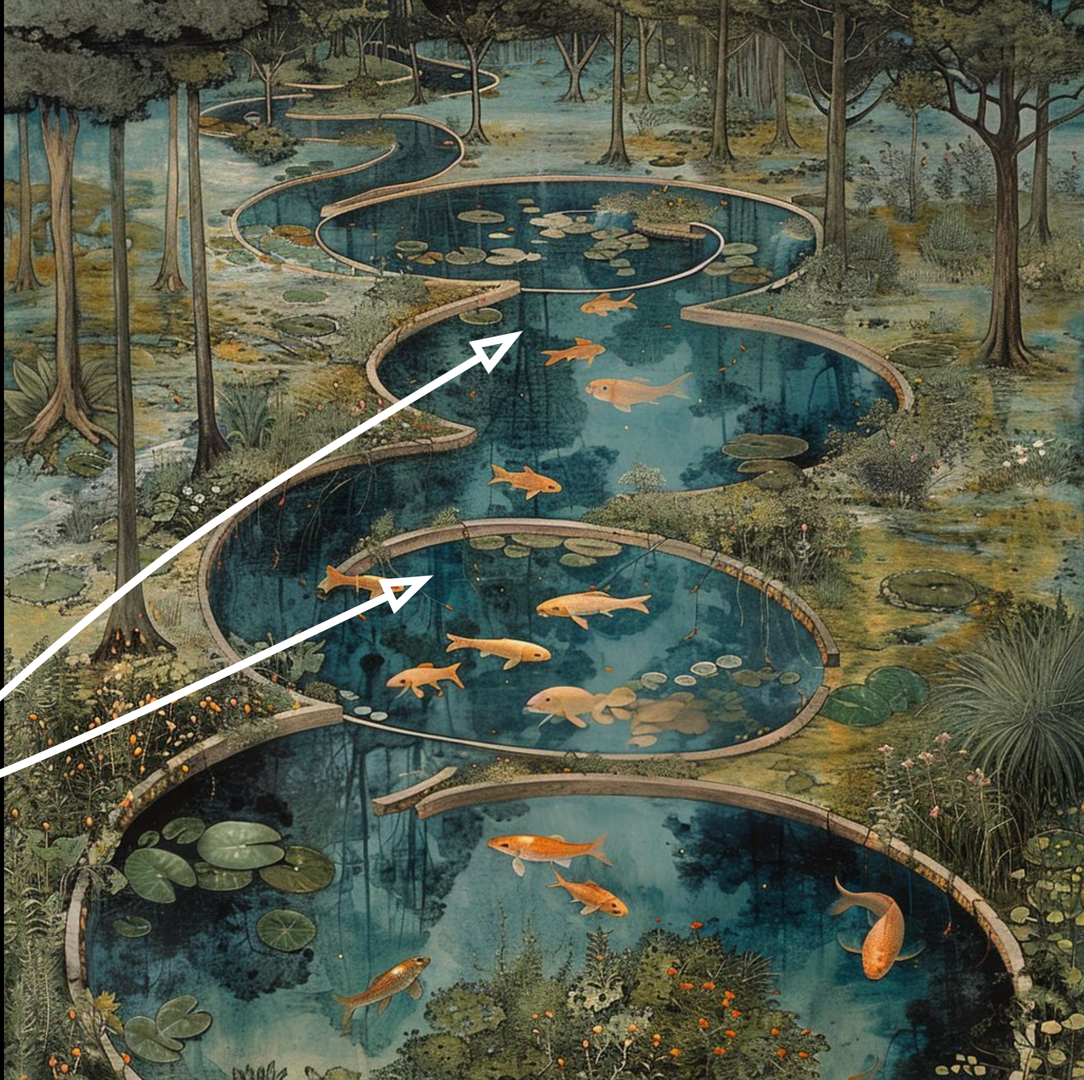
Genomic data **useful** to detect recent fragm°

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

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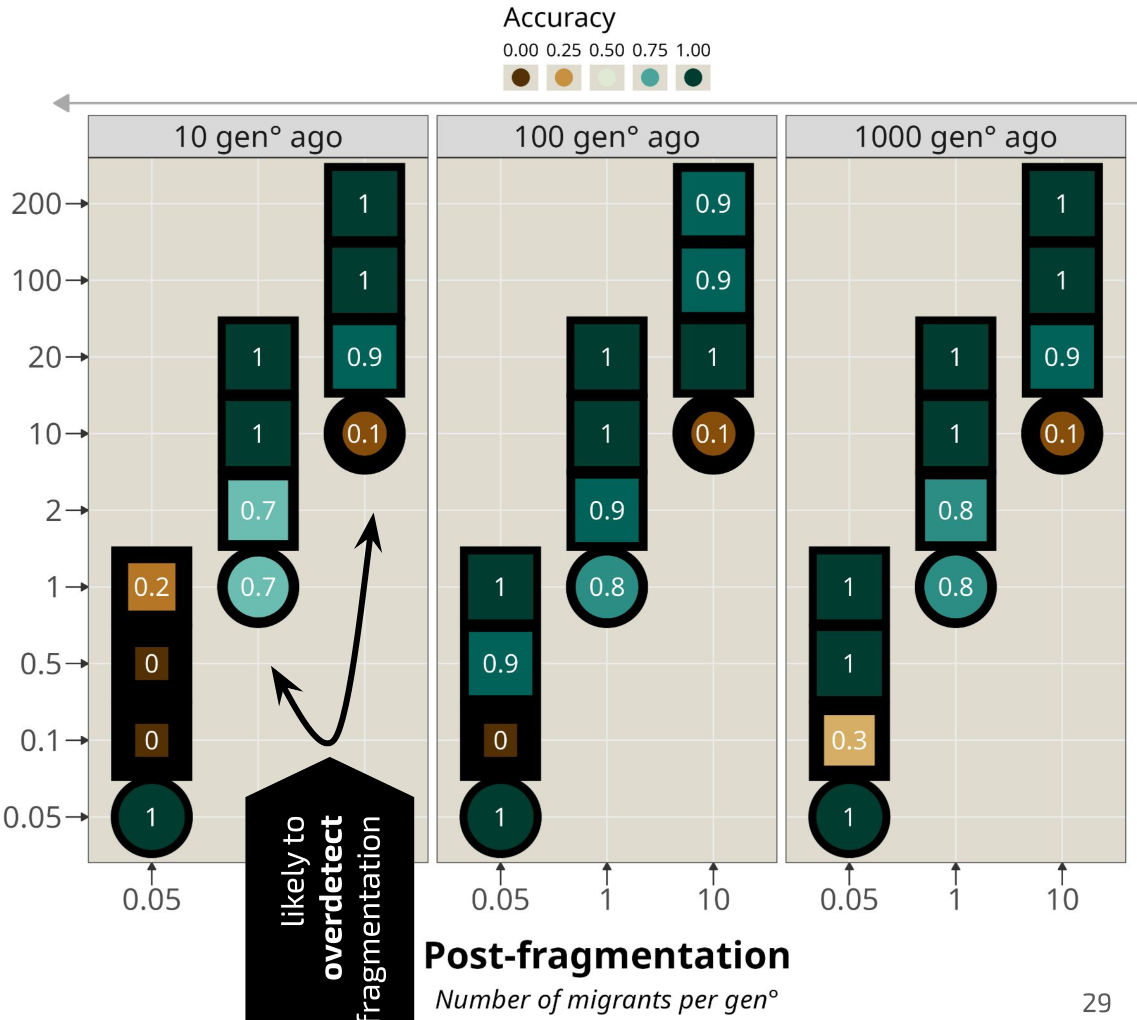
Appendix

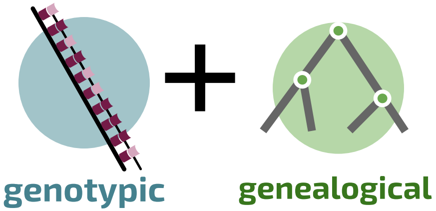
A *conservation genomic* concern



likely to **underdetect** weak fragmentation

likely to **overdetect** fragmentation

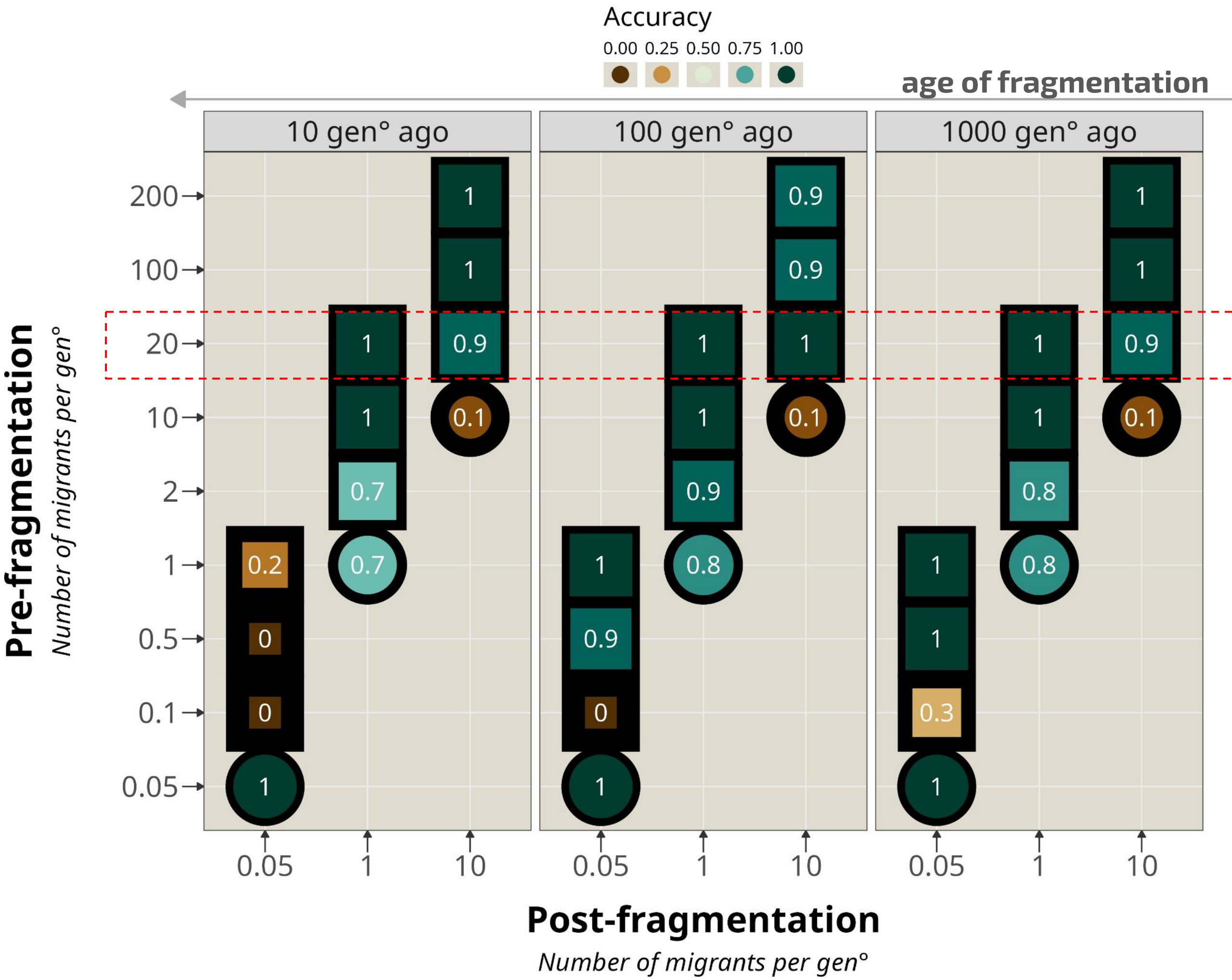
Detection power



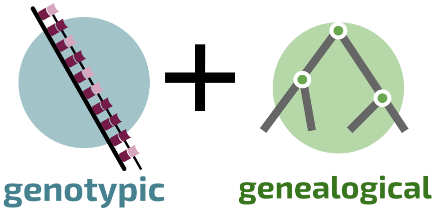
power tends to **increase** with founder **intensity**

□ fragmentation

○ stationary



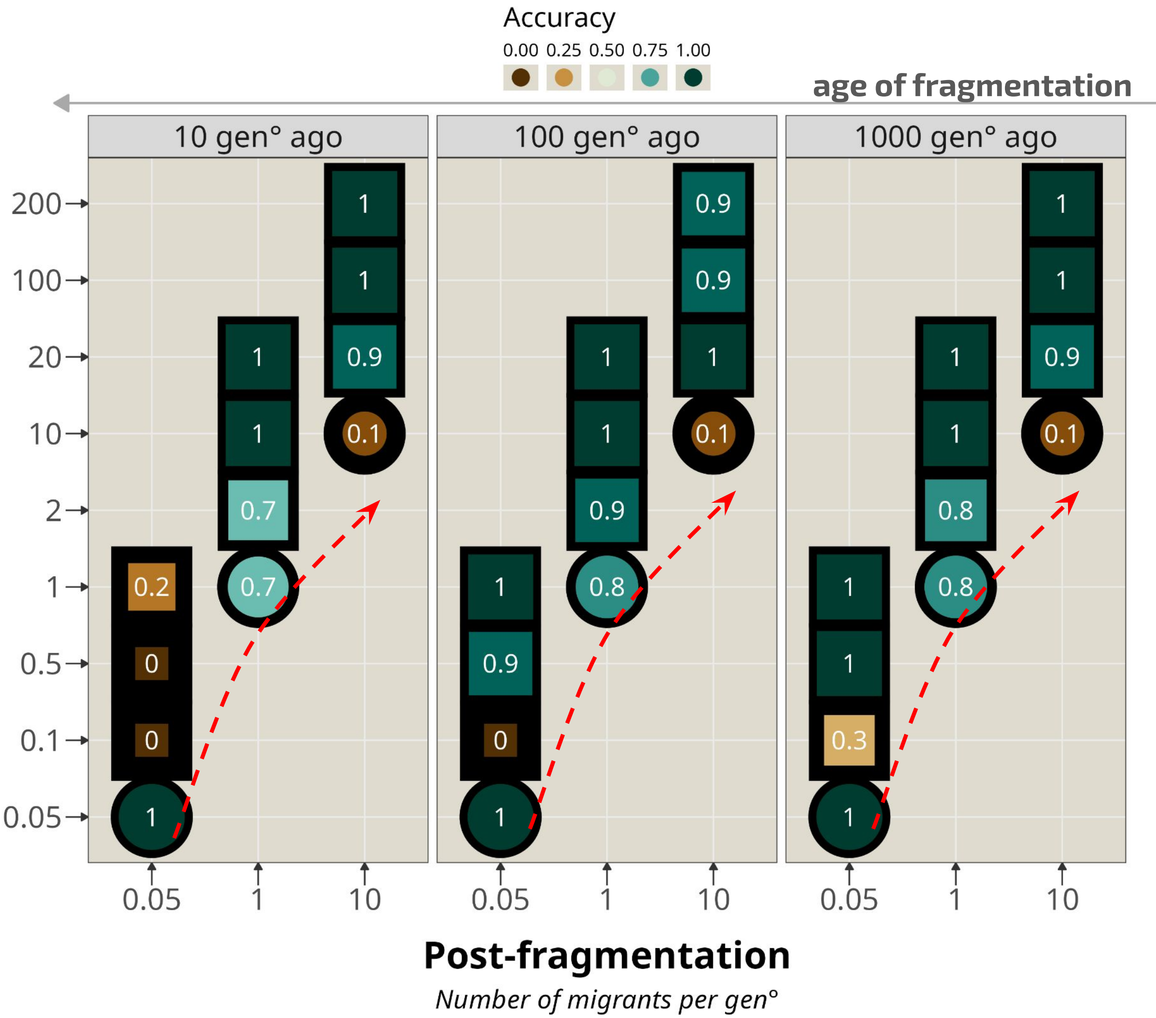
Detection power



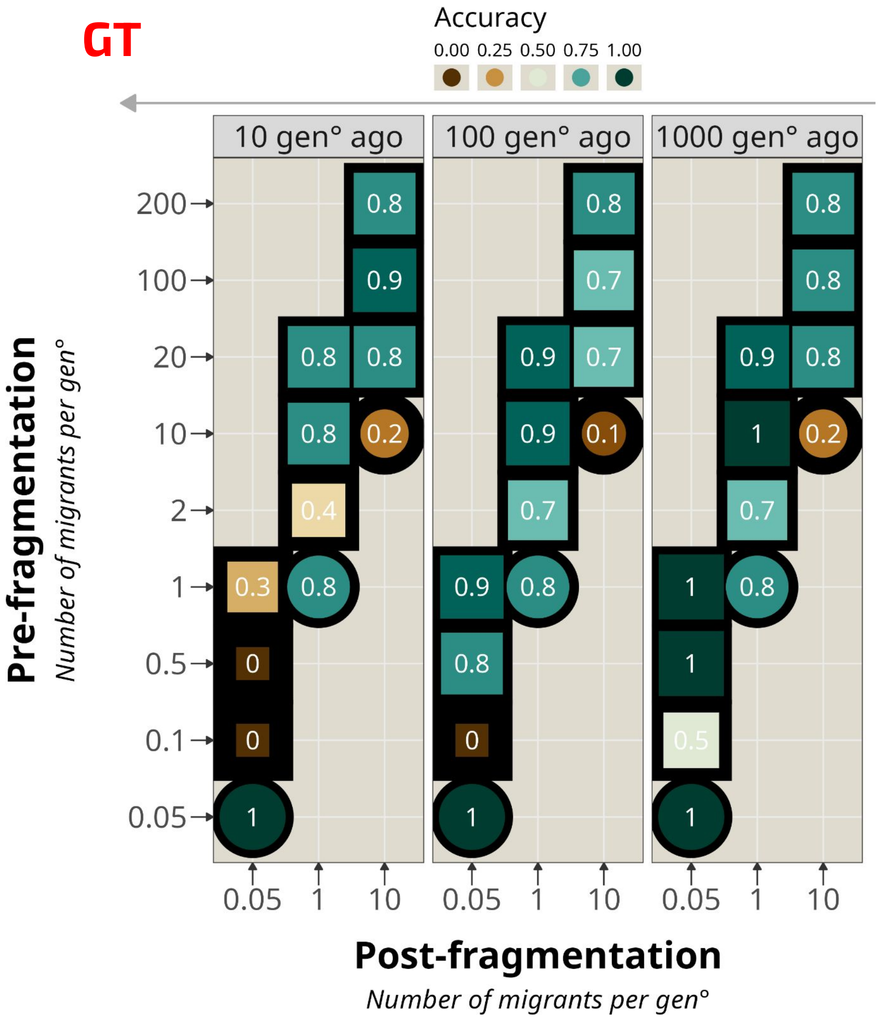
power to detect **stationary** model **decreases** with **migration rate**

☐ fragmentation

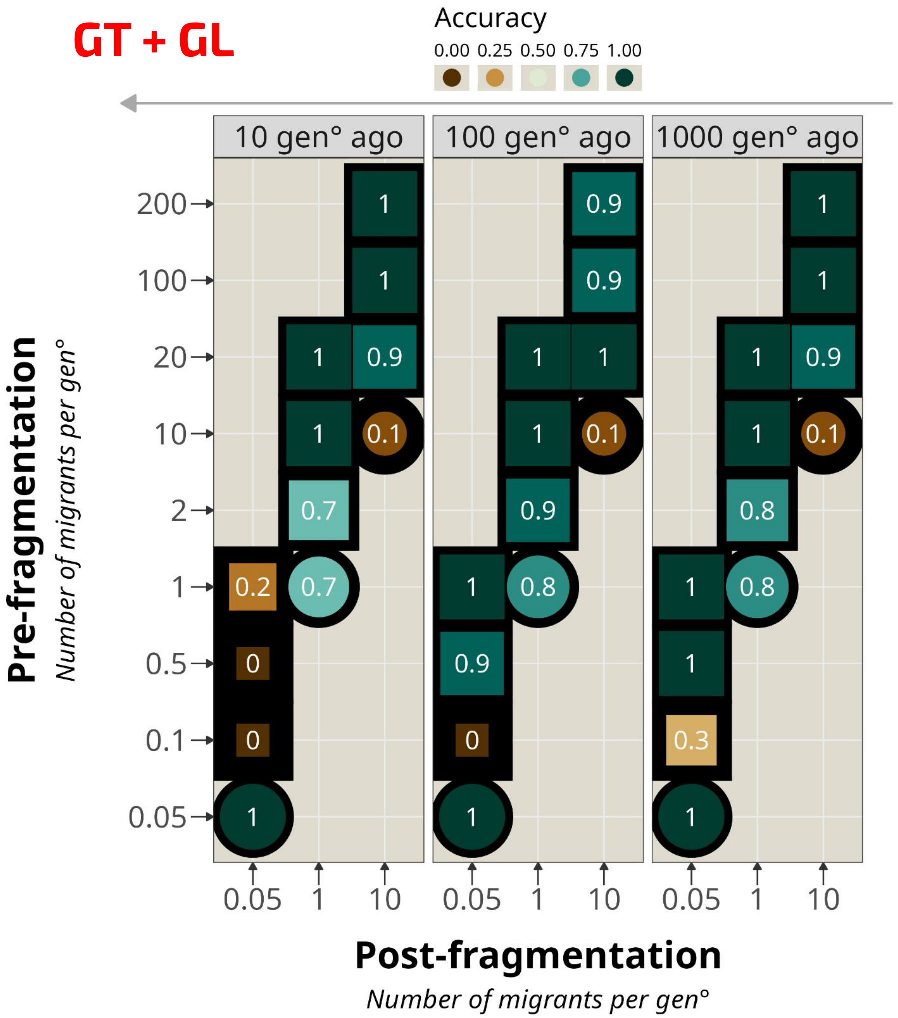
○ stationary



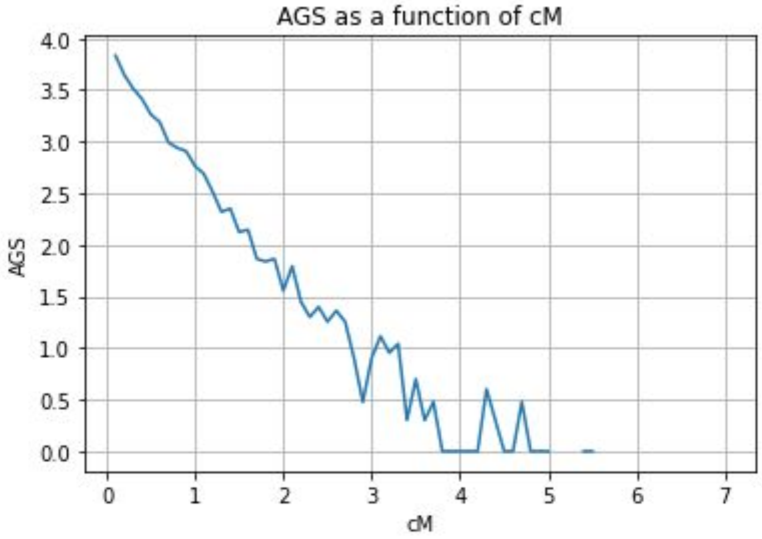
GT


Accuracy
0.00 0.25 0.50 0.75 1.00
10 gen° ago
100 gen° ago
1000 gen° ago
200 0.8 0.8 0.8
100 0.9 0.7 0.8
20 0.8 0.8 0.9 0.7 0.9 0.8
10 0.8 0.2 0.9 0.1 1 0.2
2 0.4 0.7 0.7
1 0.3 0.8 0.9 0.8 1 0.8
0.5 0 0.8 1
0.1 0 0 0.5
0.05 1 1 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°

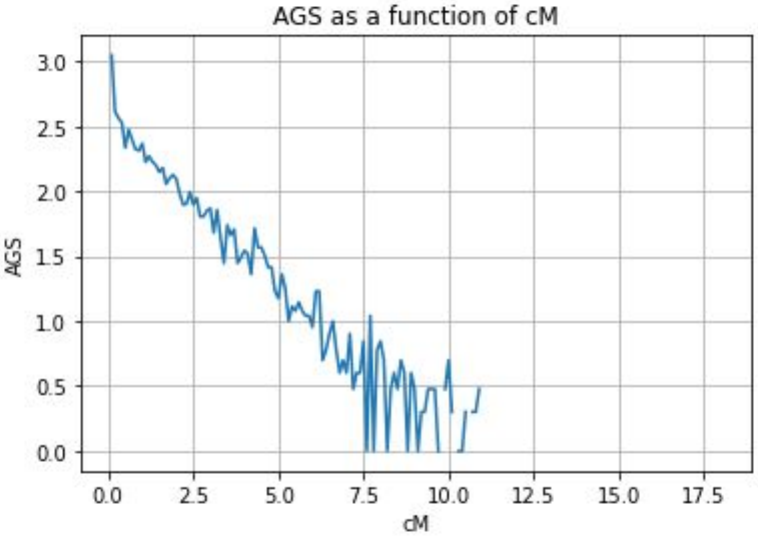
GT + GL


Accuracy
0.00 0.25 0.50 0.75 1.00
10 gen° ago
100 gen° ago
1000 gen° ago
200 1 0.9 1
100 1 0.9 1
20 1 0.9 1 1 1 0.9
10 1 0.1 1 0.1 1 0.1
2 0.7 0.9 0.8
1 0.2 0.7 1 0.8 1 0.8
0.5 0 0.9 1
0.1 0 0 0.3
0.05 1 1 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