Understanding the evolution of biodiversity

From the field to the genomes

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ECOBIO

Université de Rennes

ecosystems

species

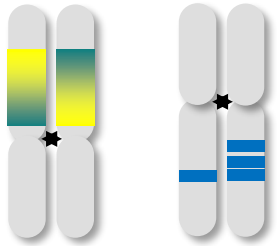
traits

genetic variation

ATGGA

ATTGA

Uni. Laval (Québec)



Structural variants

Structural variants in evolutionary and population genomics

Claire Mérot
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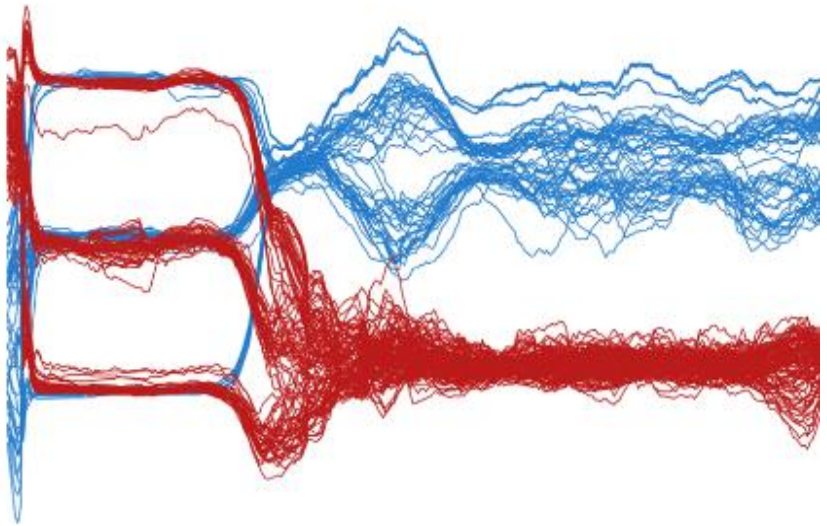
EVOL-SV
(101115983)



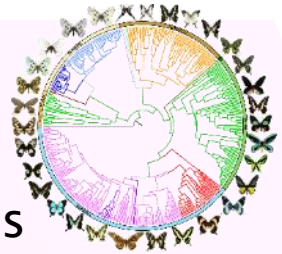
The origin and evolution of biodiversity

Ecology

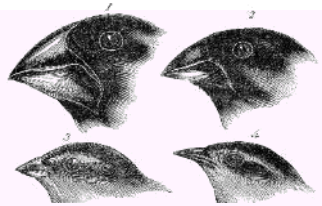
?

Genetics

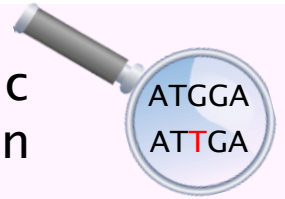
ecosystems



species

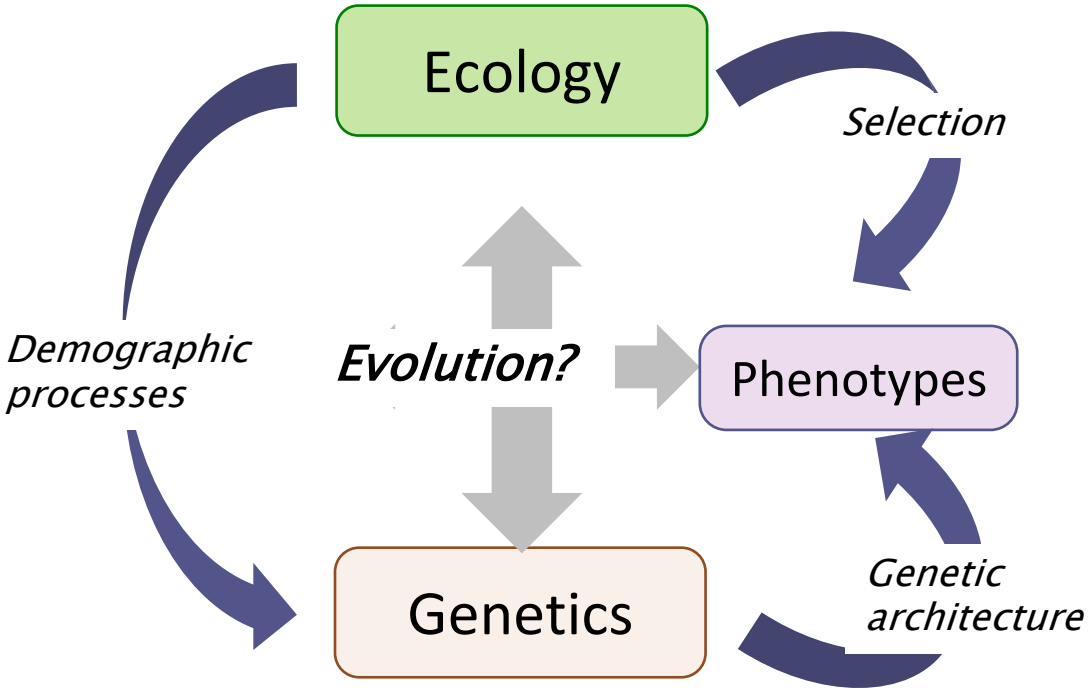


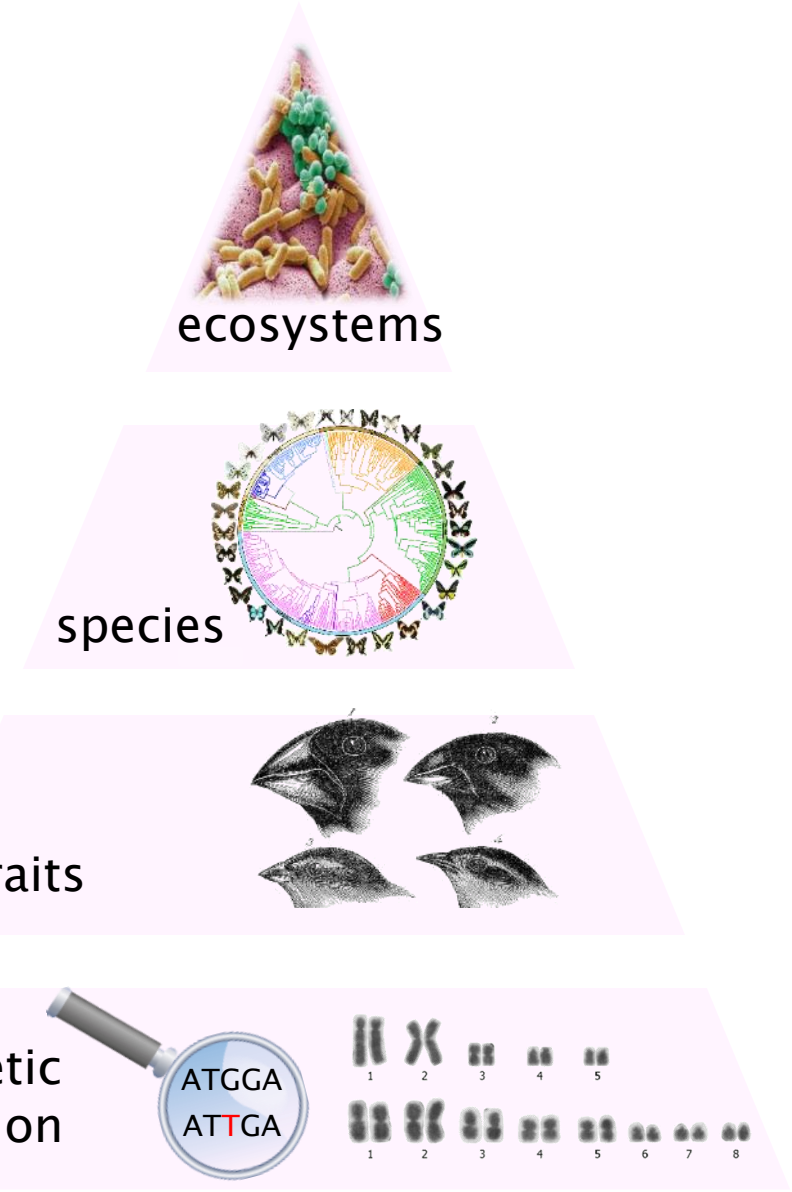
traits



genetic variation

The origin and evolution of biodiversity





⇒ Genomes vary in sequence and structure

⇒ Why and how studying SVs in evolutionary biology and population genomics?

Why and how studying SVs in evolutionary biology and population genomics?

Maren Wellenreuther

A Beginner's Guide to Structural Variants in Eco-Evolutionary Population Genomics

Stuart K,Oomen R, Tigano A,Wellenreuther M, Wold J, Field DL, Mérot C.
*(*accepted in *Molecular Ecology)*
Authorea. Dec 8, 2025.
DOI: 10.22541/au.174853973.36642913/v4

Katarina Stuart

Rebekah Oomen

David Field

Anna Tigano

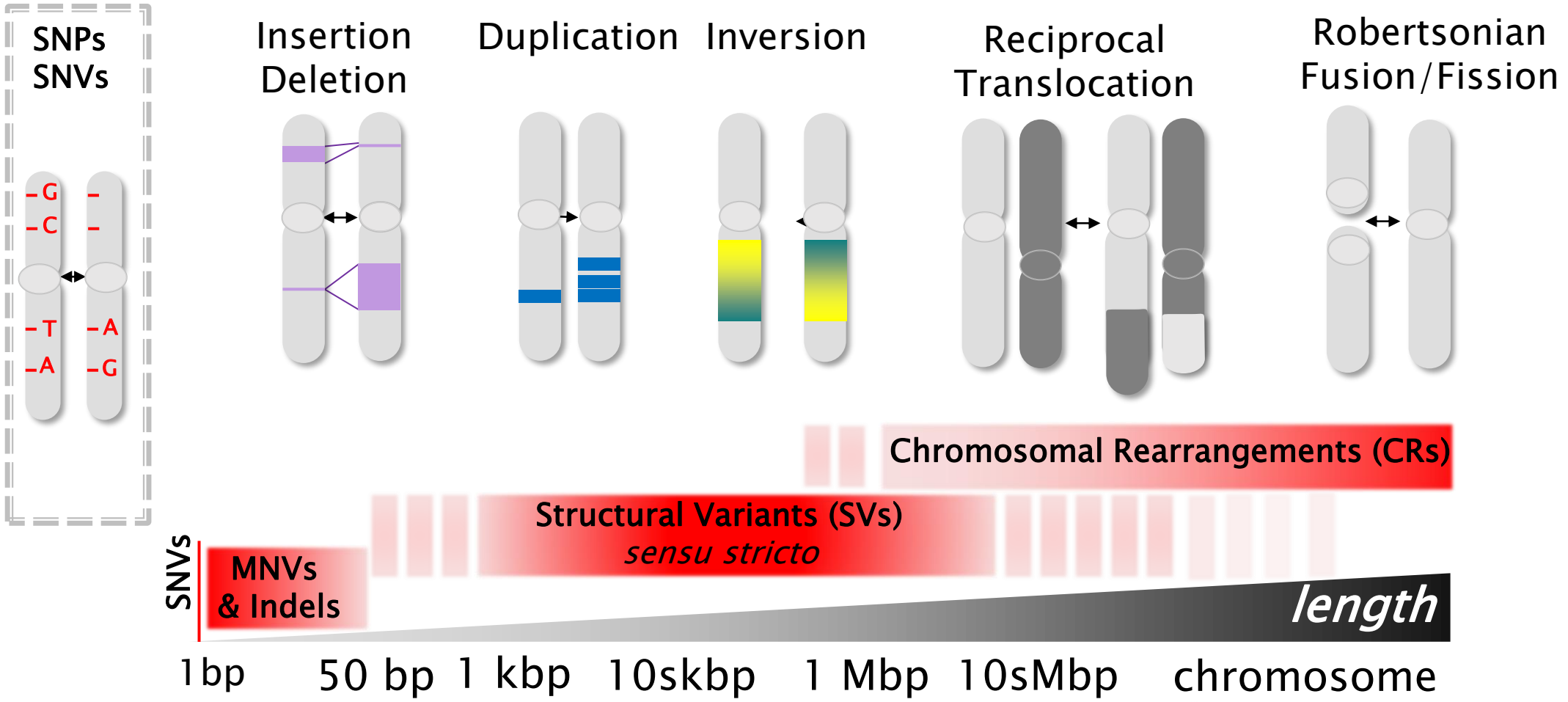
Jana Wold

A beginner's guide to structural variants in eco-evolutionary population genomics

*structural variant**
*CNV**
SV
*Copy number variant**
*chromosomal rearrangement**
*chromosomal inversion**
TE
transposable elements

2020- 2025
-> 1812 studies.
-> About 100 in Ecology & Evolution

What are SVs?



⇒ Challenge:

How to classify and analyse such a diversity of variants…?

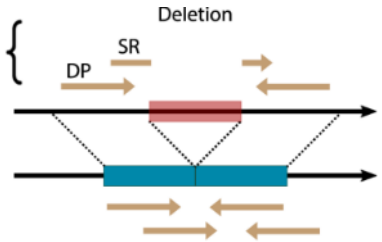
What are SVs?

Bioinformatics

Evolutionary biology

Type ?

(INS/DEL/DUP/INV/TRA/…)



Mutational origin?

eg : TE / non-TE

Properties?

eg : - fitness effect?
- recombination effect?
- dosage effect?

Length?

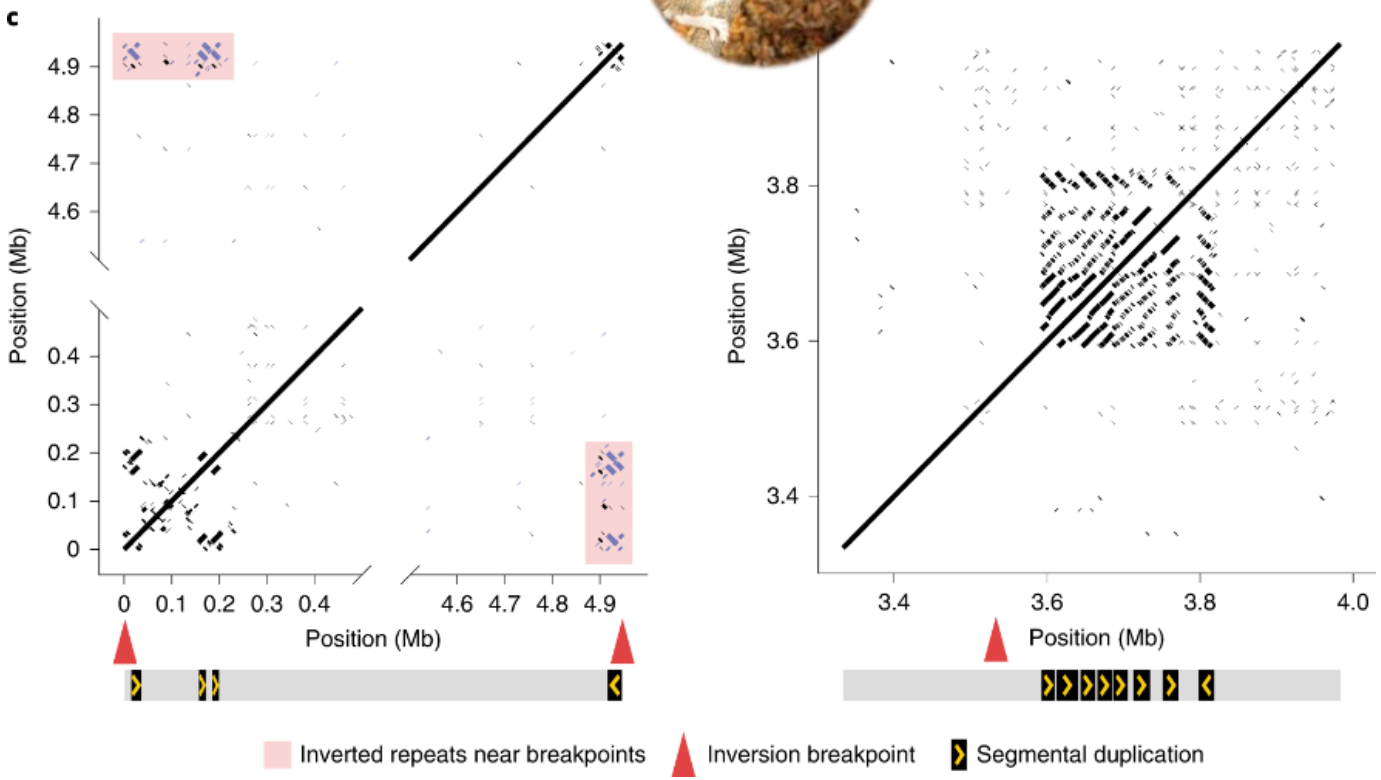
⇒ **Challenge:**

How to classify and analyse such a diversity of variants…?

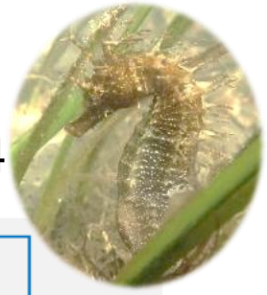
Co-studying large inversions, TEs and duplications provided insights into putative mutational process.

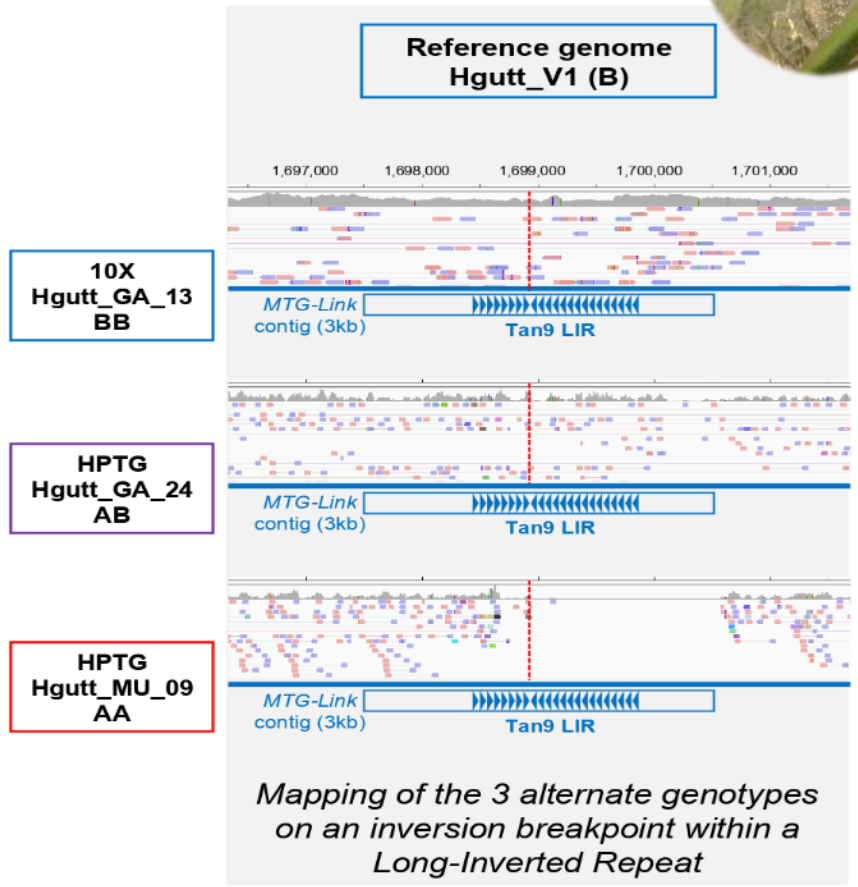
⇒ ectopic recombination driven by inverted repeats is a likely origin for those large inversions

Harringmeyer et al, 2022



Meyer et al, 2024

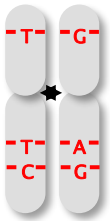




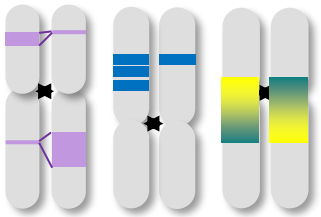
What is the level of genetic diversity attributable to SVs?

The total fraction of the genome varying across individuals because of SVs is 3 to 10 times higher than because of SNPs

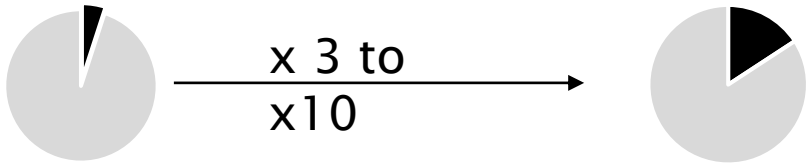
⇒ **Challenge: How to compare SV prevalence across species and datasets?**



SNPs



SVs



Human & Great apes differ by 1.3% (based on SNPs) but more than 3.5% when including SVs

Karageorgiou et al 2024

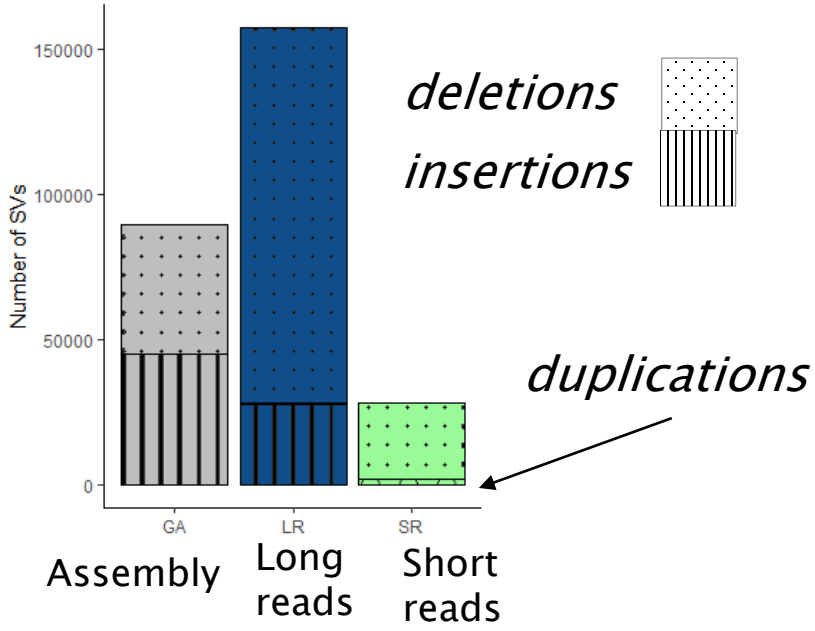
What is the level of genetic diversity attributable to SVs?

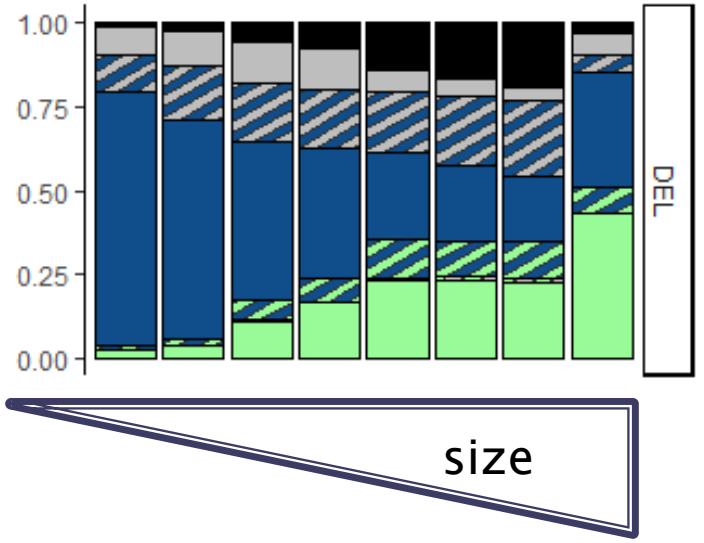
« A mean of 4,442 SVs per human genome (with short-reads) & 27,662 SVs (with long-reads) »

Chaisson et al, 2019; Abel et al 2020

⇒ Challenge:
How to compare SV prevalence across species and datasets?

Coregonus clupeaformis





⇒ Assemblies vs. long-reads vs short-reads: not the same SVs are detected

Structural variants… comparable data matters!

Why studying SVs when we already have SNPs?

Some structural variants have an important functional impact on phenotypes

Guo et al 2025

Wilkinson et al, 2024

Zhou et al 2022

Explained heritability
SNPs: 29%
SNPs+SVs: 41%

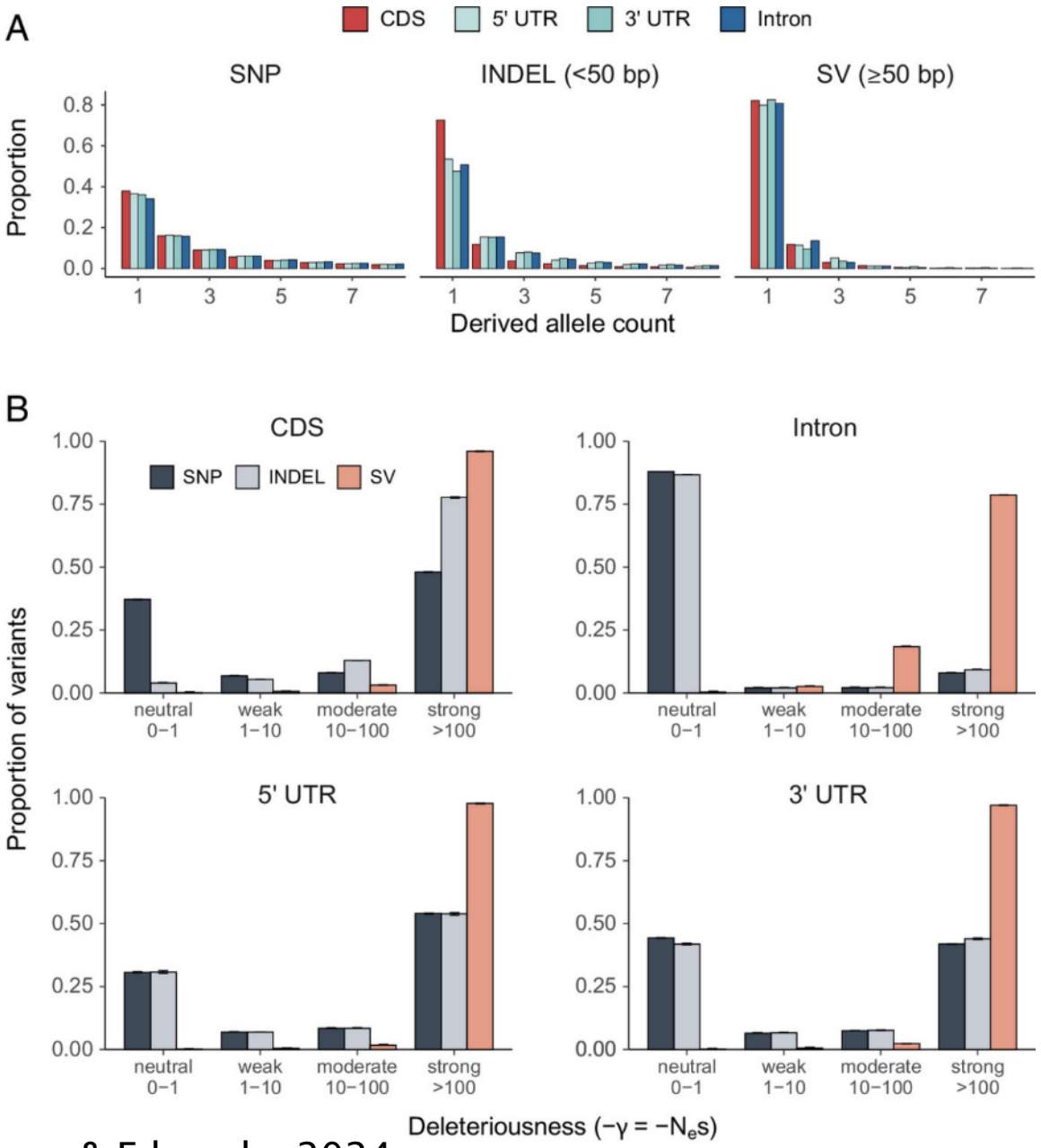
Zhang et al, 2021

Blaj et al, 2022

Jaio et al, 2025

40-50% of SVs are not linked to a SNP marker

Why studying SVs when we already have SNPs?



Fang & Edwards, 2024

The majority of SVs are likely to be deleterious

- Biased towards rare SVs
- Predicted deleterious effects

⇒ Important in conservation genomics!

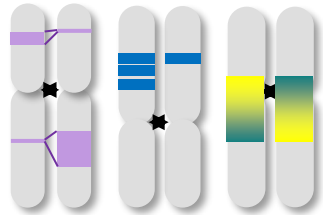
Smeds et al, 2024

Stuart et al, 2024

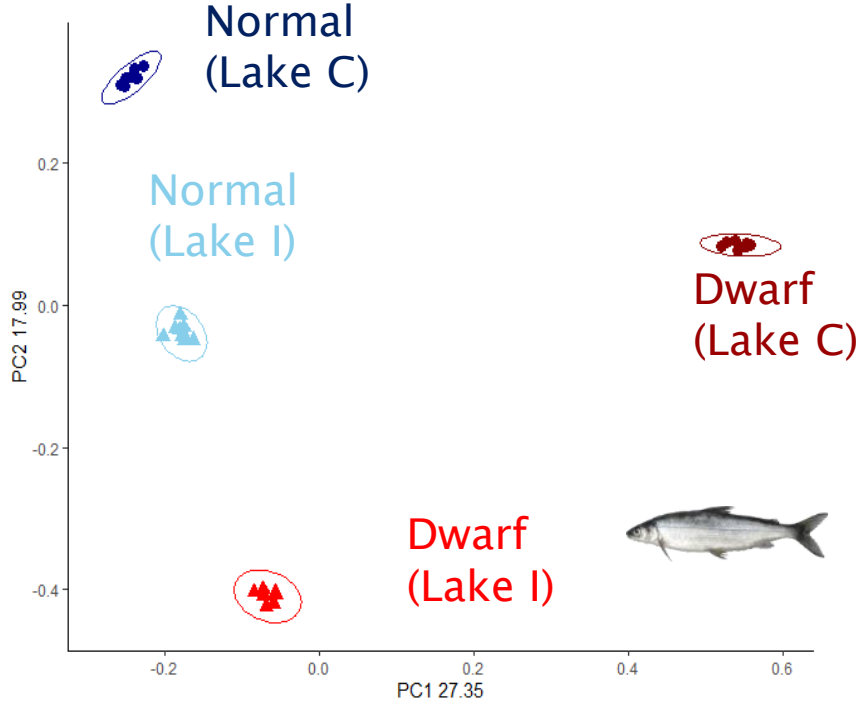
Why studying SVs when we already have SNPs?

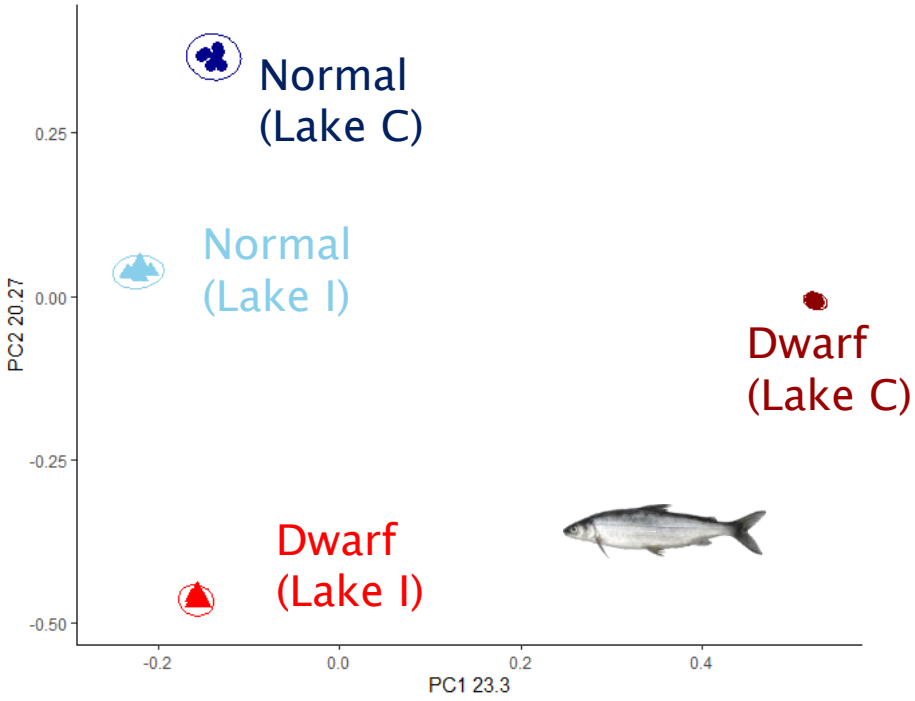
What about neutral markers for population genomics?

SNPs



SVs

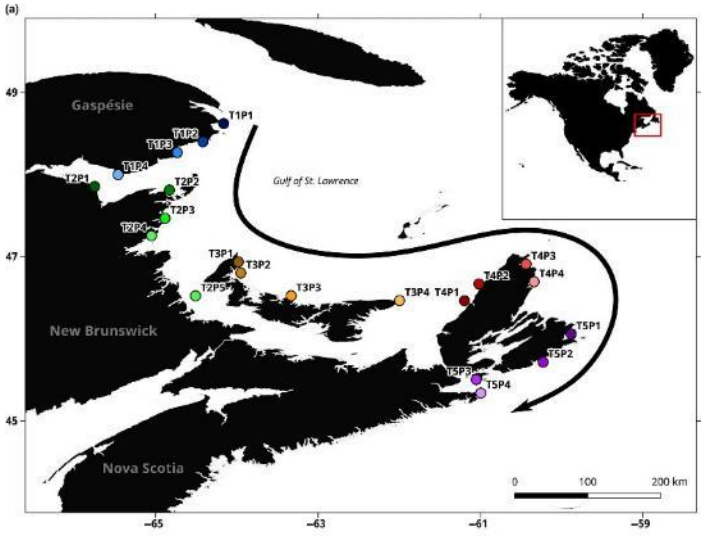




Mérot *et al.* 2023.

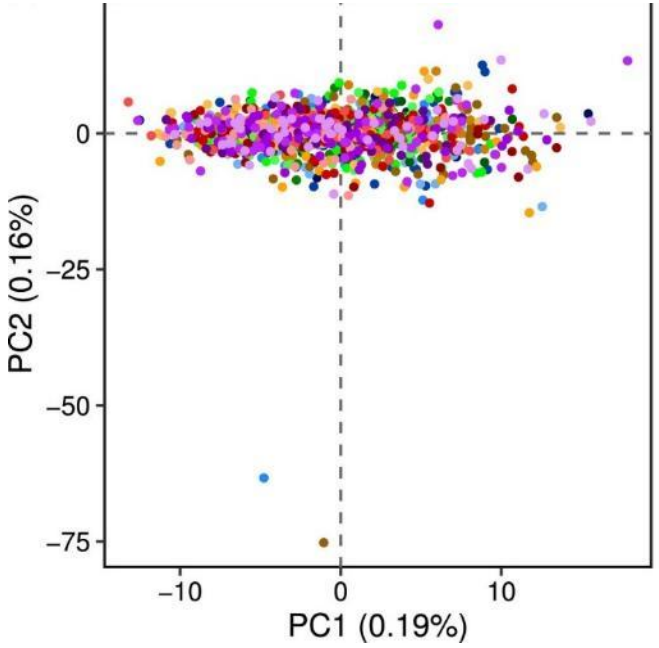
Why studying SVs when we already have SNPs?

What about neutral markers for population genomics?

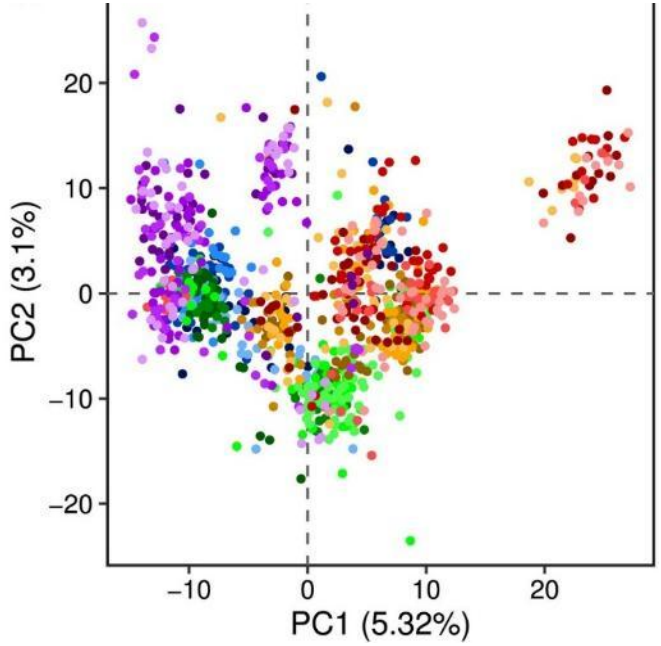


Dorant *et al.* 2020.

SNPs



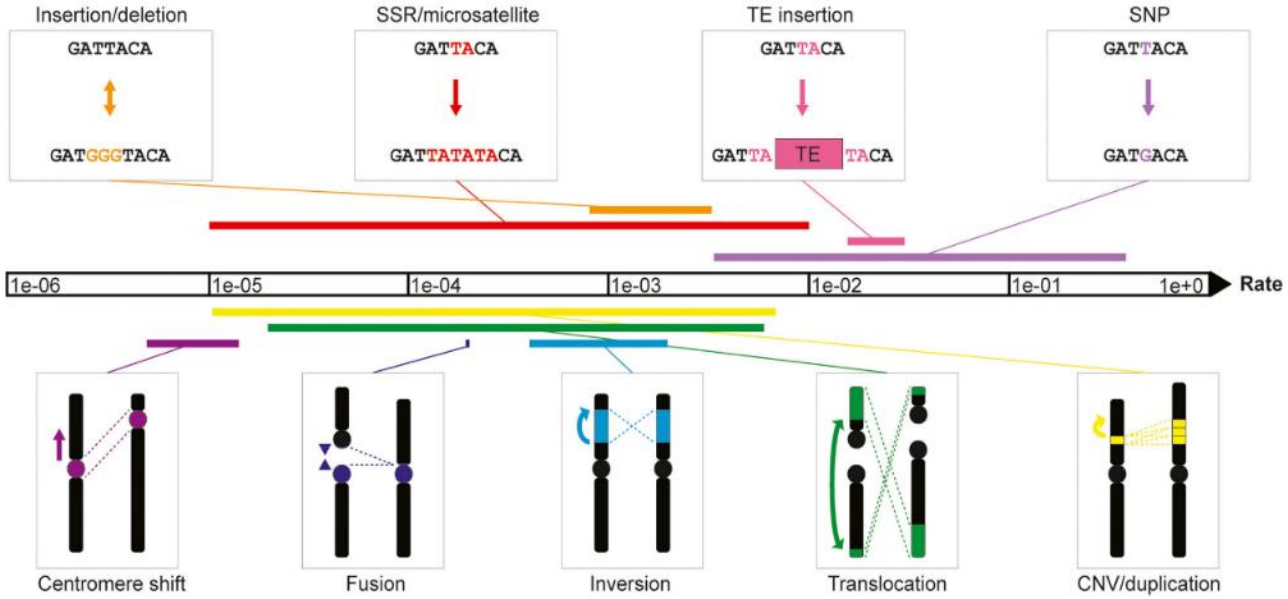
CNVs



Which properties of SVs may matter for evolution?

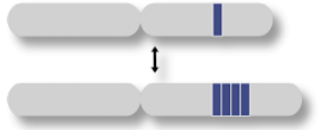
Structural variants contribute to the evolution of genomes, sometimes very quickly…

Transposable elements bursts are important contributor to genome dynamics



Insecticide resistance in *Anopheles* sp.

Weetman et al 2018



CNVs

Rapid adaptation to cave life in *Astyanax mexicanus*

Pokrovac et al, 2024

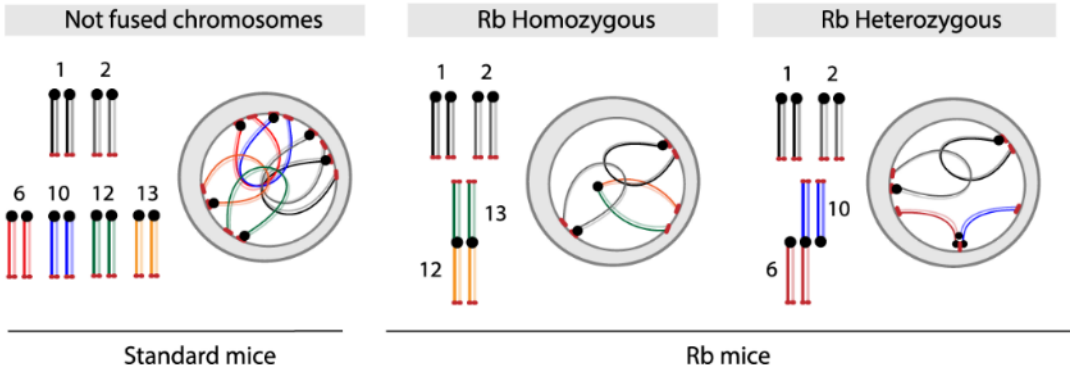
Which properties of SVs may matter for evolution?

Some structural variants have indirect effects,

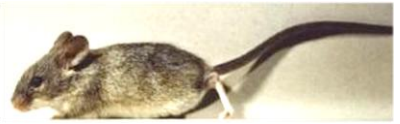
(beyond their sequence change)

- Alter genome organisation and functionality

TAD reorganisation and 3D organisation affected by chromosomal fusions



Vara et al 2021



- Impact methylation & epigenetic marks

TEs affects nearby gene expression & histones

Fablet et al 2021

- Affect pairing and/or cross-over resolution during meiosis

The peculiar case of non-recombining blocks...

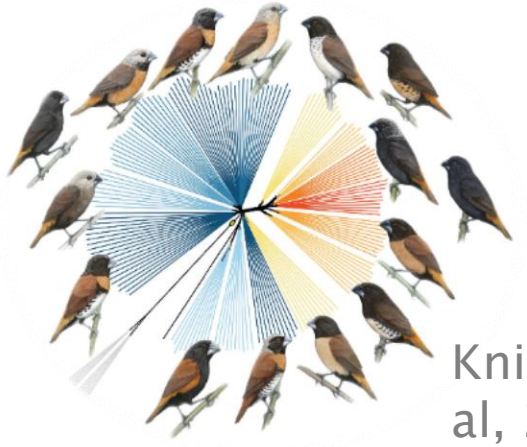
Hoff et al 2024

Salson et al 2025

Reeve et al 2023

Zhang et al 2024

Dallaire et al 2025



Knieff et al, 2024

Huang et al 2020

Todesco et al 2020

Harringmeyer et al, 2022

Wellband et al 2019

Lehnert et al 2025

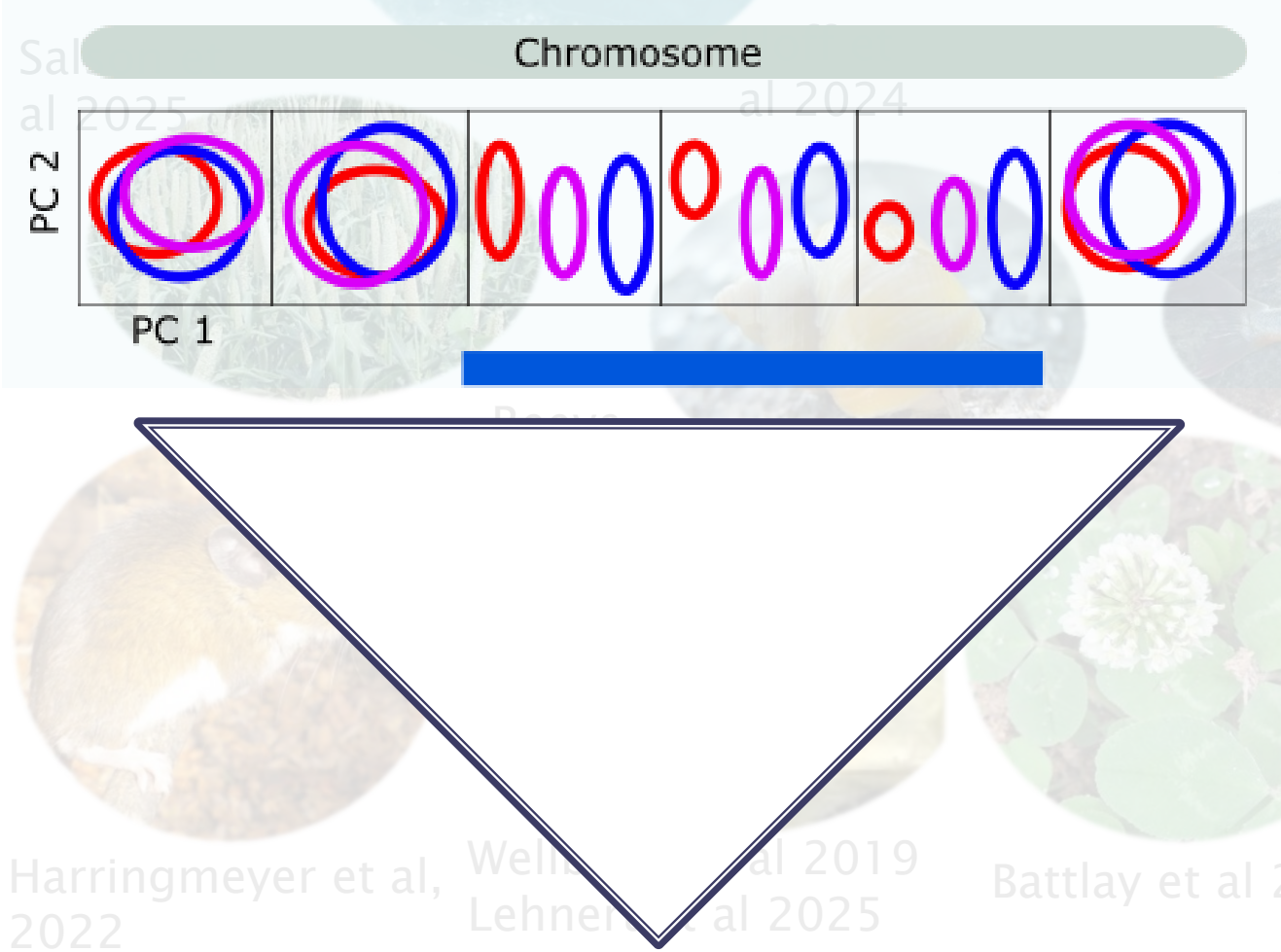
Battlay et al 2025

Mérot et al 2021

Meyer et al, 202

The peculiar case of non-recombining blocks…

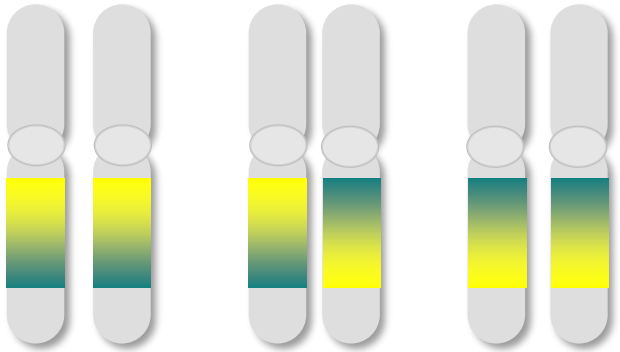
One or several regions of high genomic diversity where variance clusters in differentiated groups



⇒Usually a signature of non-recombining blocks of haplotypes…

…which may (or may not) be large structural rearrangements

The peculiar case of non-recombining blocks...



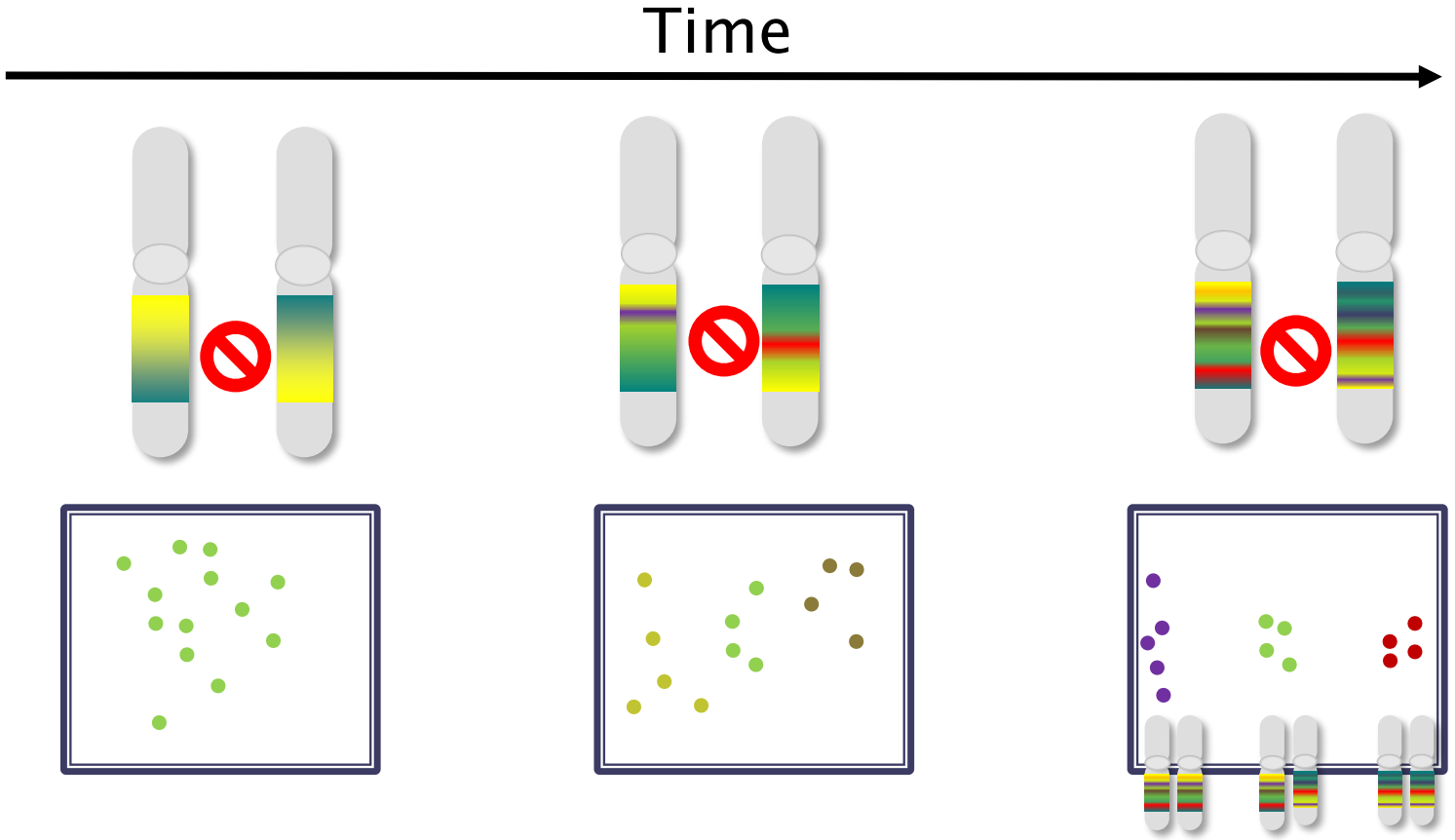
Normal Meiosis

Normal Meiosis

No recombination

Inversions form loops at meiosis

Unbalanced – unviable gametes



⇒ Each « haploblock » evolves separately

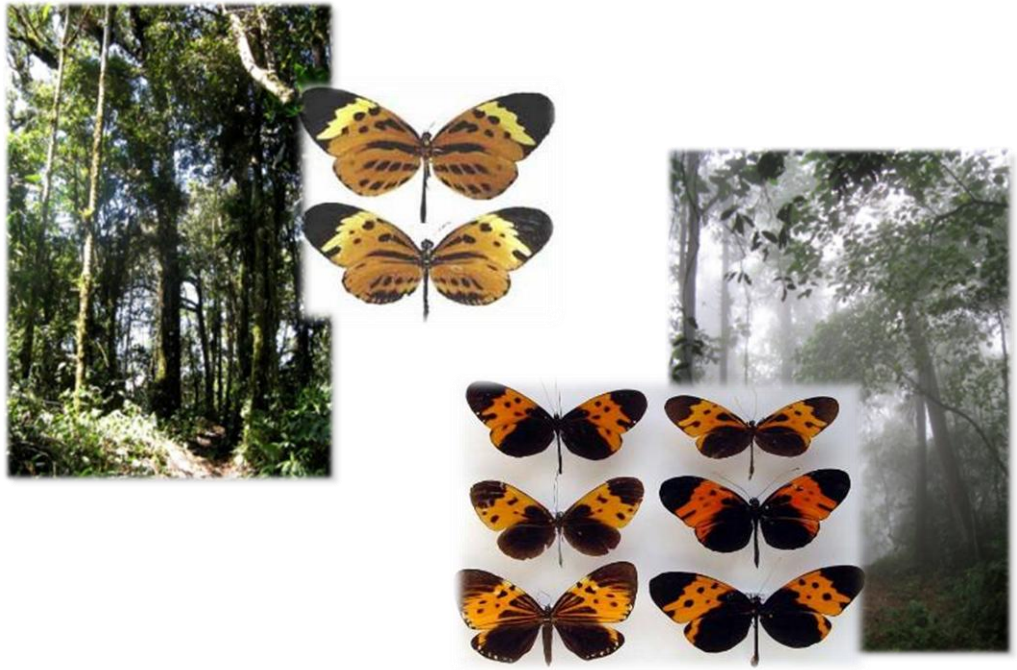
⇒ Chromosomal rearrangements are frequently confirmed by other data

The peculiar case of non-recombining blocks...

Ecology & evolutionary processes

Local adaptation to different ecological niches

Speciation



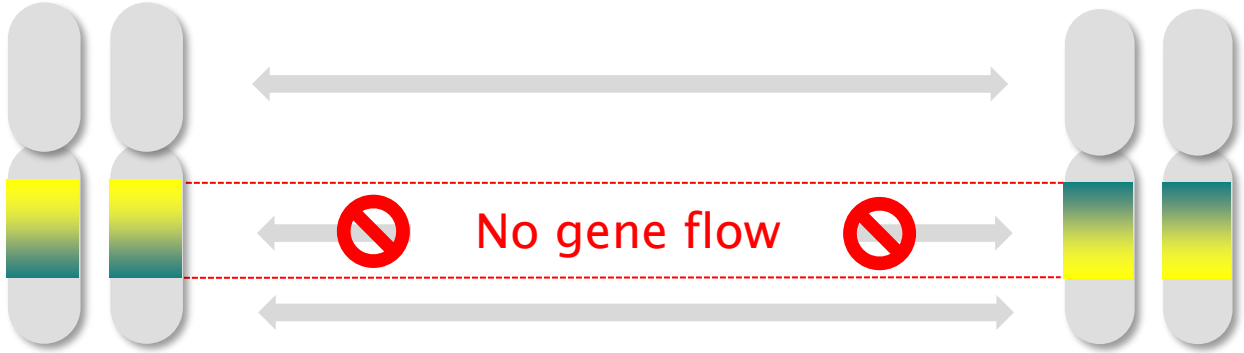
Phenotype

Discrete & complex: “ecotypes” “morphotypes”

Pleiotropy and covariance across traits

Genetics

Linkage across multiple genes and regulatory regions



The peculiar case of non-recombining blocks...

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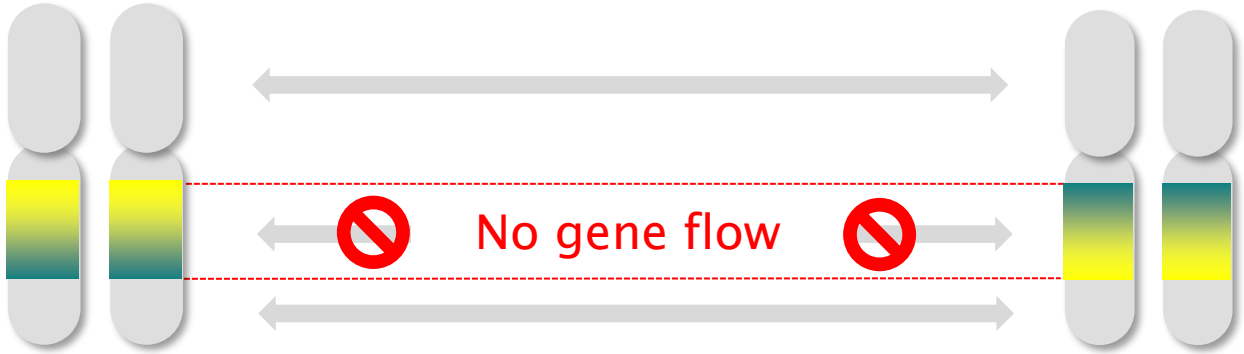
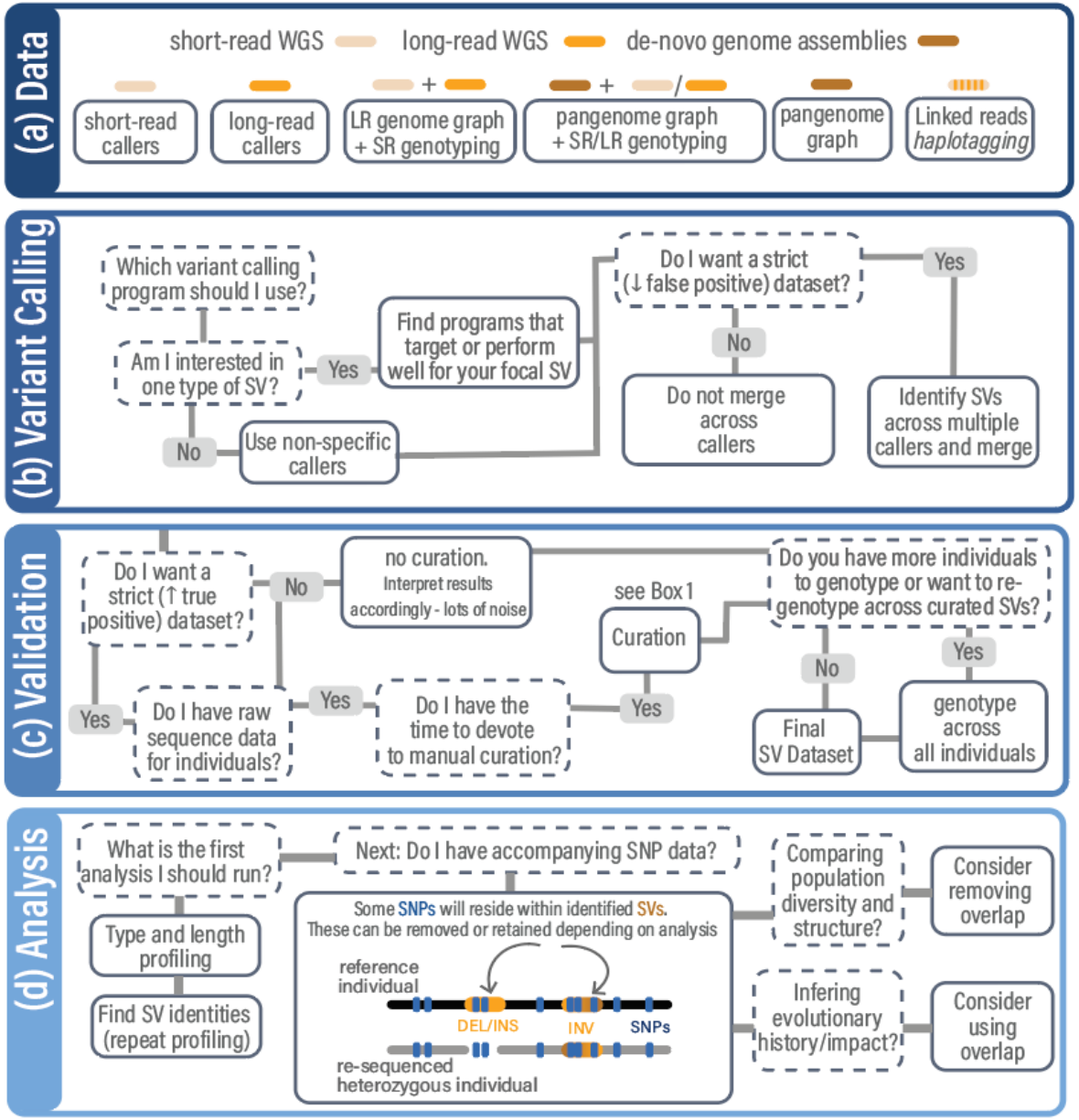


⇒ What are the ecological and demographic conditions favouring such architecture?

⇒ What is the functional effect of haploblocks?

⇒ What is the underlying structure and origin?

How to analyse SVs in population genomics?



⇒ Optimising the ratio information/data

⇒ Validating

⇒ Accounting for SV specificities & diversity

How to analyse SVs in population genomics?

Examples with Lake whitefish and Atlantic salmon

Louis Bernatchez

Laurie Lecomte

Mérot C., Stenløkk KSR., Venney C., Laporte M., Moser M., Normandeau E., Árnyasi M., Kent M., Rougeux C., Flynn JM., Lien S., Bernatchez L. 2023. Mol. Ecol. 32, 1458–1477. https://doi.org/10.1111/mec.16468

Lecomte L., Árnyasi M., Ferchaud A-L., Kent M., Lien S., Stenløkk K., Sylvestre F., Bernatchez L., Mérot C. 2024. Evol. App. 17, e13653. https://doi.org/10.1111/eva.13653

UNIVERSITÉ LAVAL

INSTITUT DE BIOLOGIE INTÉGRATIVE ET DES SYSTÈMES IBIS

Combining short and long-reads to study SVs in population genomics

Louis Bernatchez

Lake Whitefish

Norwegian University of Life Sciences

S. Lien's group

Atlantic Salmon

Laurie Lecomte

Assembly | **Long-reads**

The best dataset

BUT…

ONT N=2

ONT N=4

Only a few samples due to costs & difficulties to get long DNA

Illumina N=32

Illumina N=60

Short-reads

Capture population-level variability

BUT…

Needs high-coverage & has a lot of false positives

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Mérot *et al.* 2023

Lecomte *et al.* 2024

https://github.com/LaurieLecomte

Combining short and long-reads to study SVs in population genomics

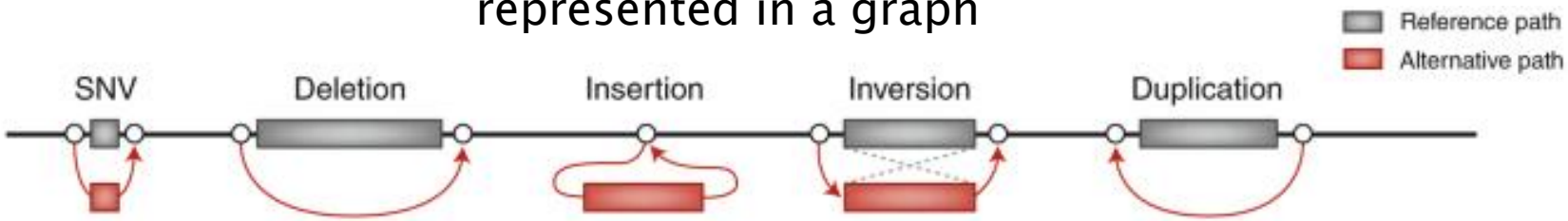
Louis Bernatchez

Lake Whitefish

Assembly | **Long-reads**

High quality short-reads

Catalogue of SVs represented in a graph



Many samples with short-reads

	Ind 1	Ind 2	…	Ind n
DEL – position 125	R/R	R/A	…	A/A
DUP – position 12459	R/A	R/R	…	R/R
INV – position 35894	R/A	A/A	…	A/A

Atlantic Salmon

Laurie Lecomte

⇒Population genotyping of SVs

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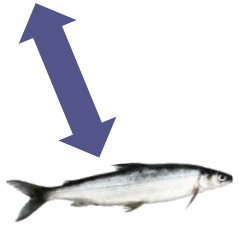
Mérot *et al.* 2023

Lecomte *et al.* 2024
https://github.com/LaurieLecomte

What is the genetic architecture of species differentiation?

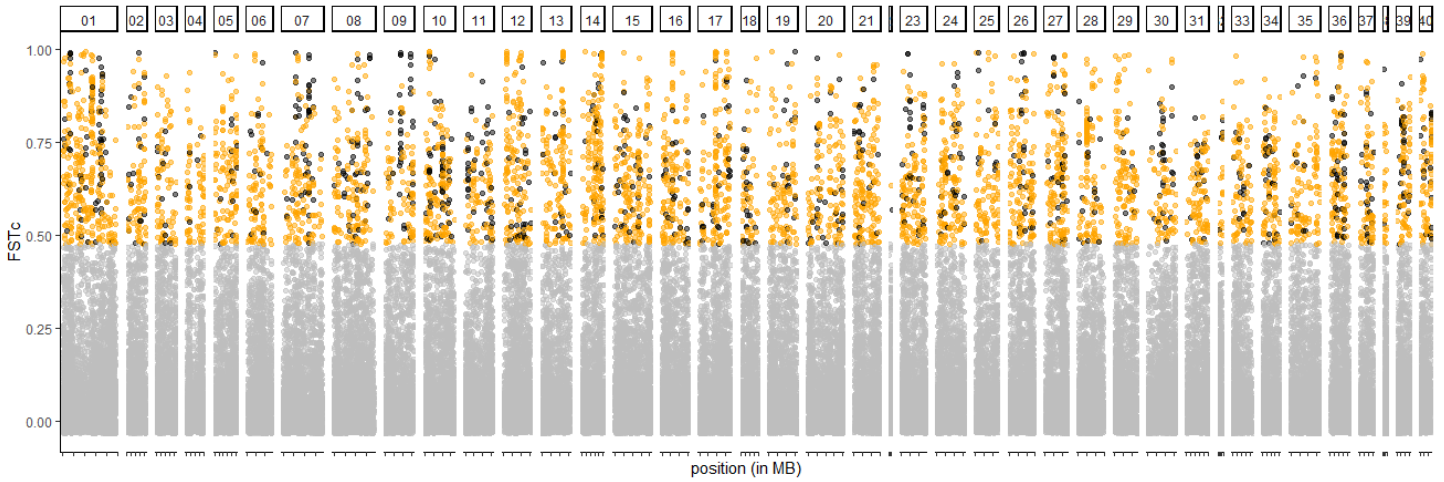
Coregonus clupeaformis

F_{ST}= 0.18



Normal vs. Dwarf
(based on 103,857 SVs)

F_{ST}



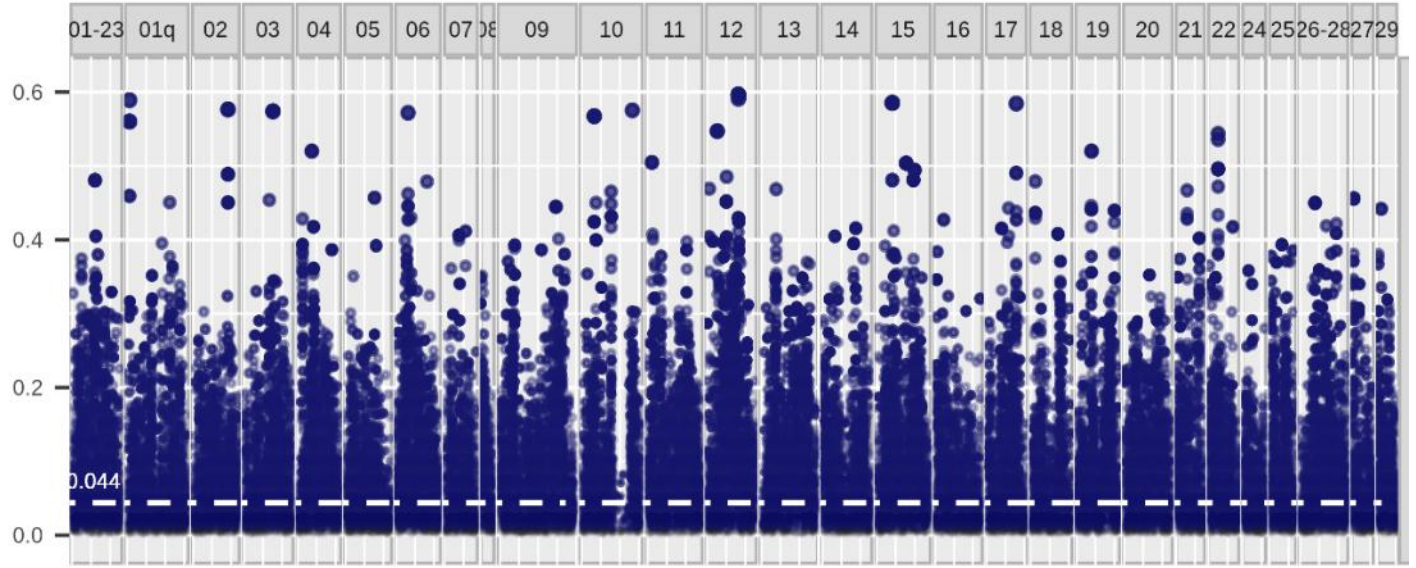
F_{ST}= 0.04

Salmo salar

River Romaine vs. Pujalon

(based on 219,379 SVs)

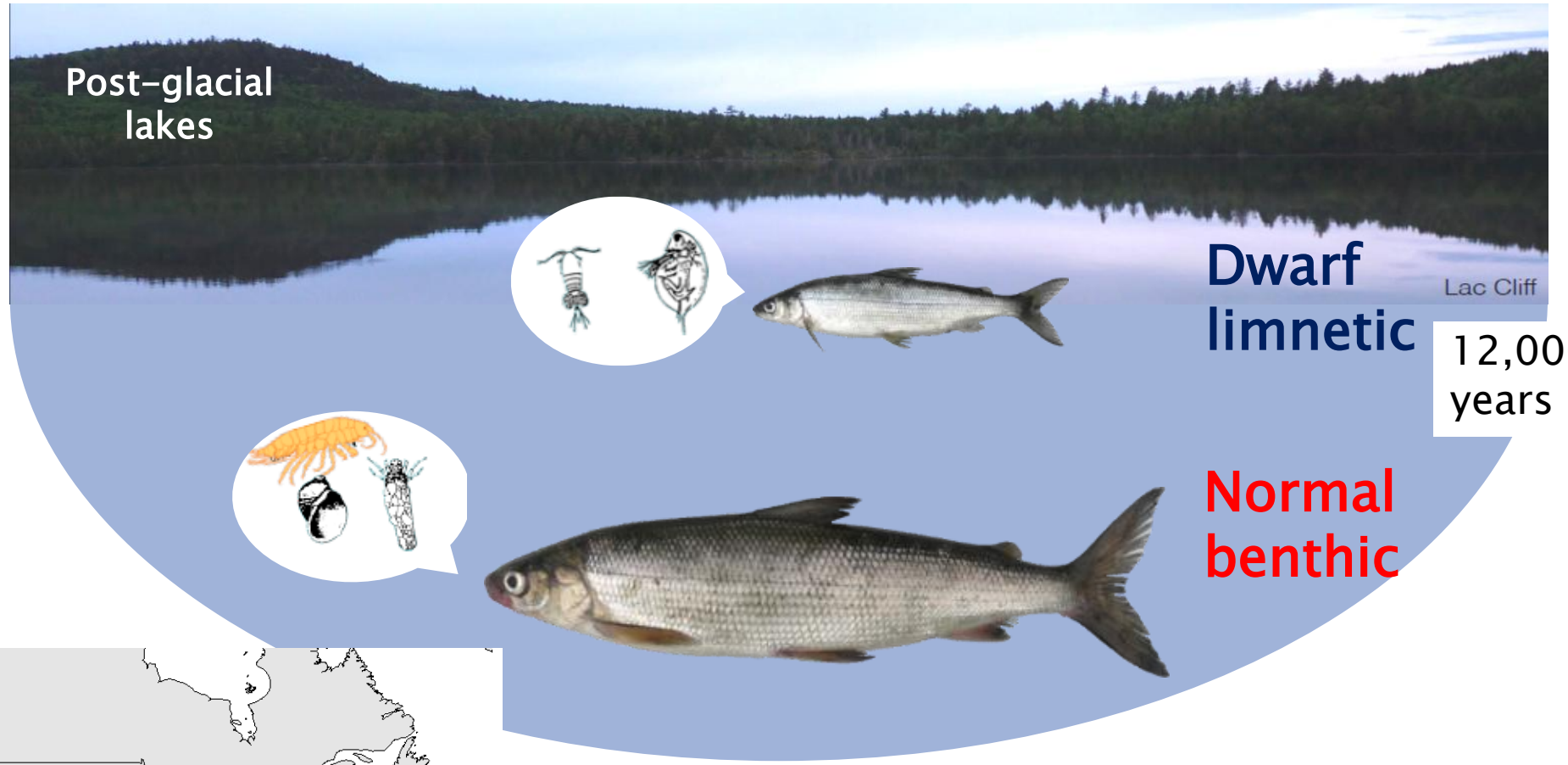
F_{ST}



Species differentiation in Lake Whitefish

Louis Bernatchez

Bernatchez & Dodson, 1990
Bernatchez et al. 2010
Rougeux et al, 2017



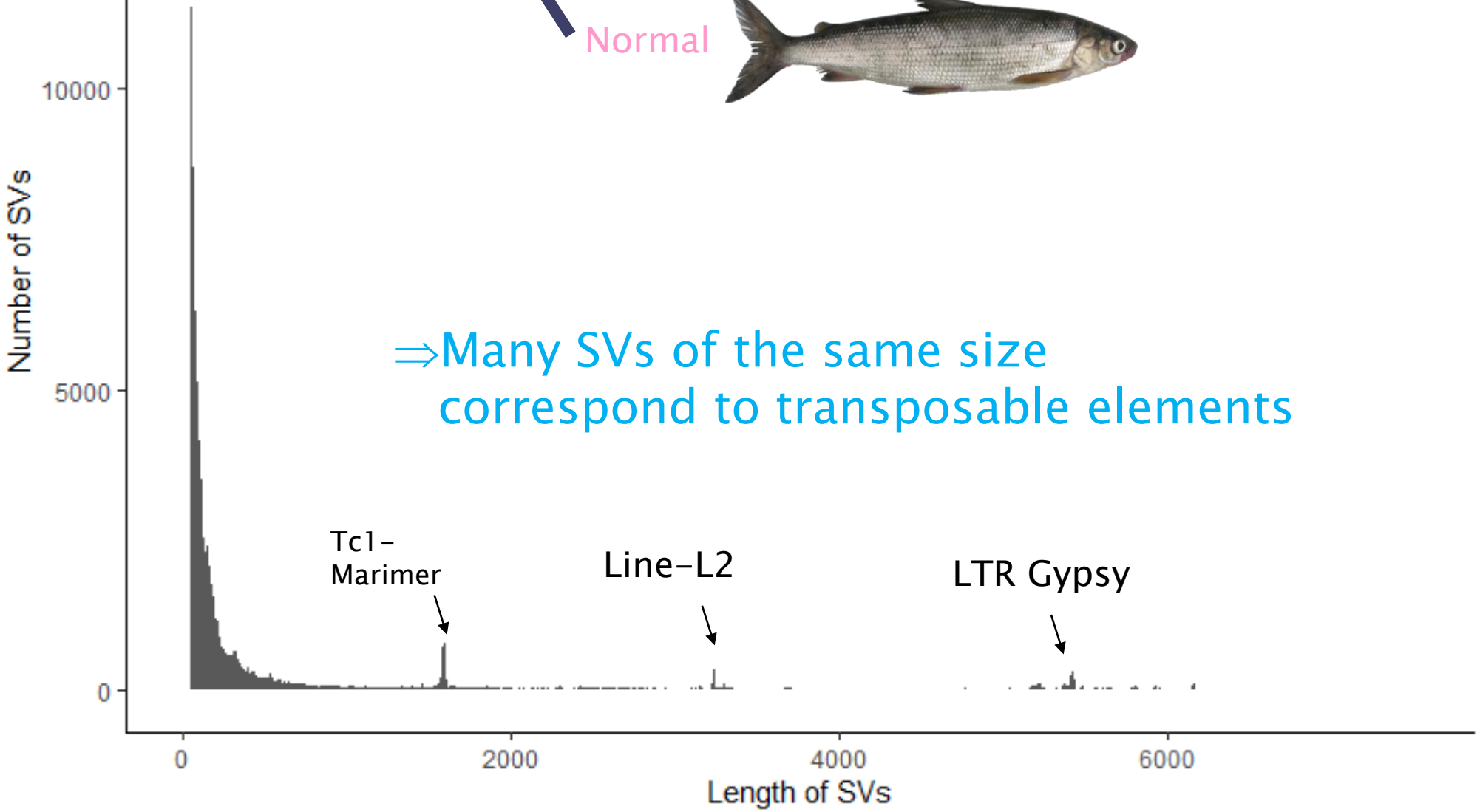
12,000 years ago

Two allopatric glacial lineages

What are SVs? Can we classify them?

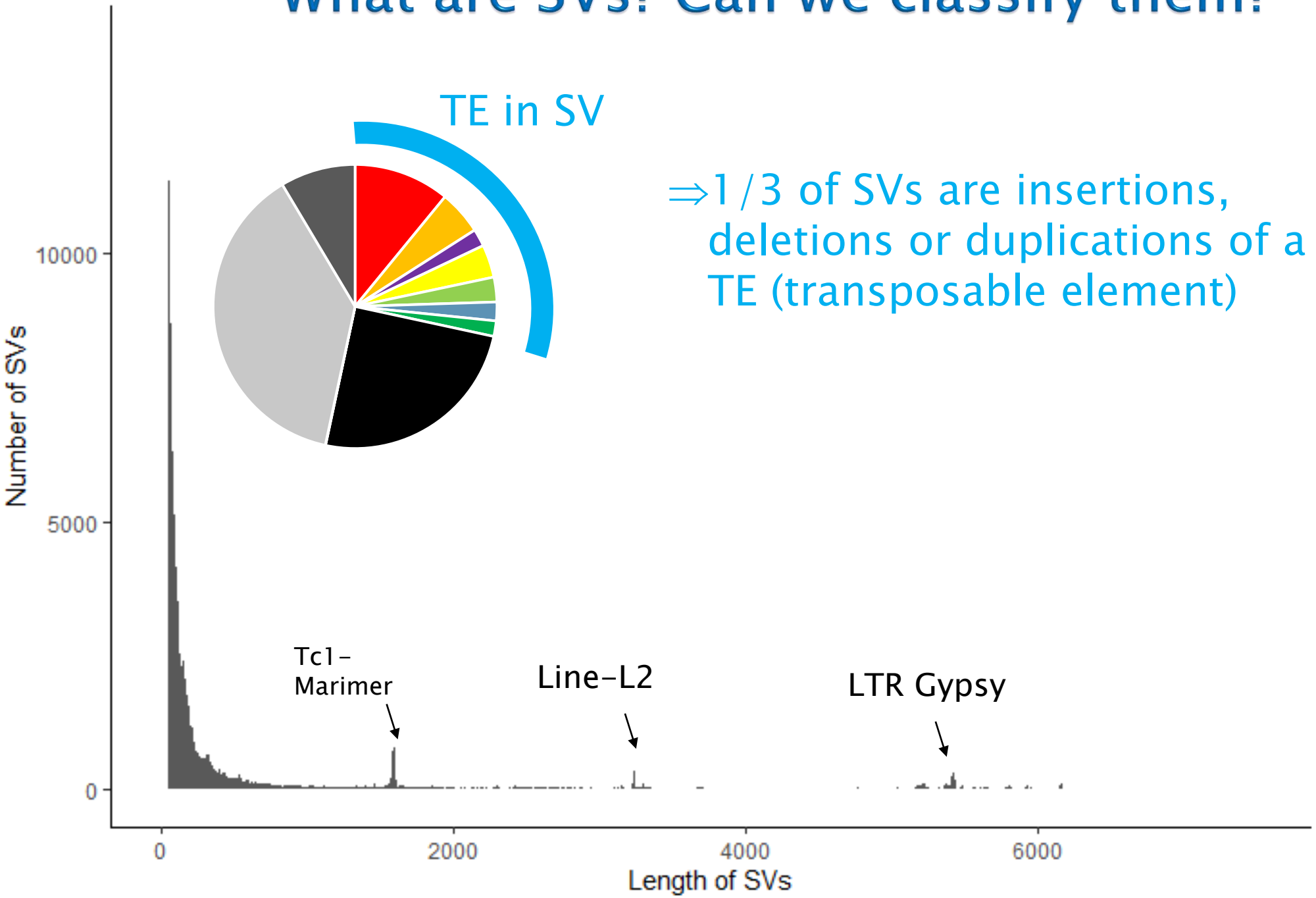
Coregonus clupeaformis

⇒Many SVs of the same size correspond to transposable elements



Mérot et al 2023

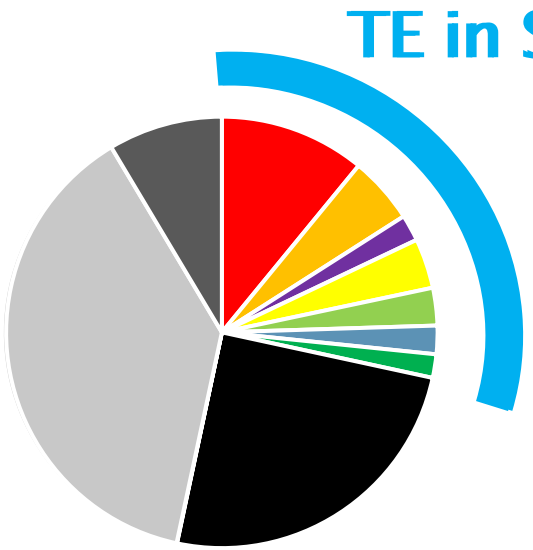
What are SVs? Can we classify them?

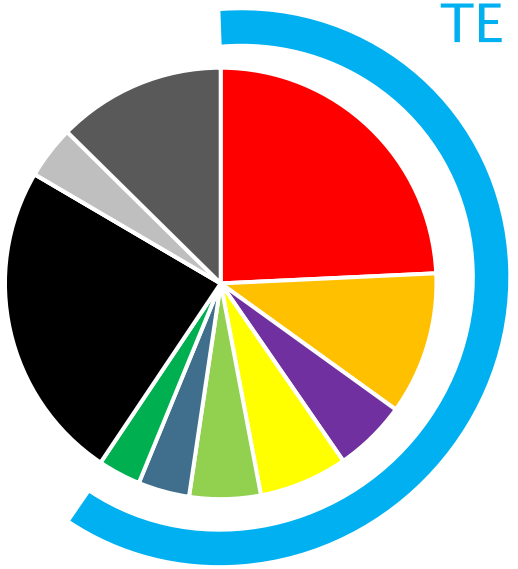


⇒1/3 of SVs are insertions, deletions or duplications of a TE (transposable element)

Mérot et al 2023

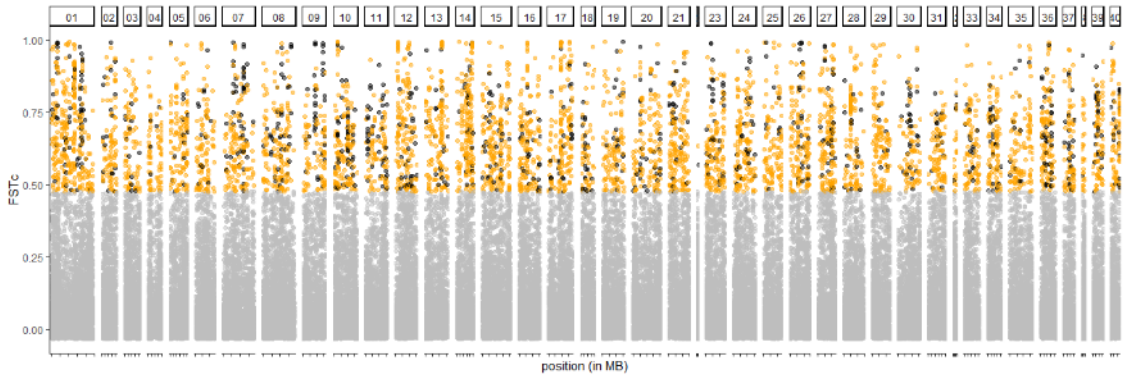
What is the genetic architecture of species differentiation?





Significant enrichment for all TE families

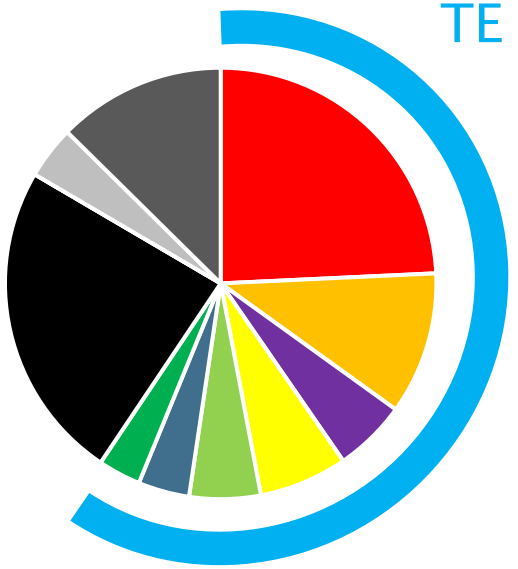
⇒ The most differentiated SV between species are enriched in TE-associated SVs…



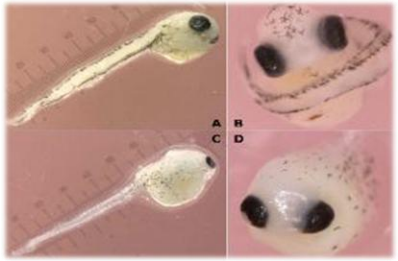
Mérot et al 2023

An importance of TE-associated SVs in species isolation?

⇒ A possible role of Tes in rapid genetic differentiation & post-zygotic breakdown



Significant enrichment for all TE families



Genome-wide misregulation of TEs *(changes in methylation & TE expression)*

Dion-Côté et al, 2014
Laporte et al, 2019

Mérot et al 2023

How do large rearrangements shape populations?

The peculiar case of inversions/non-recombining blocks...

cnrs

erc

Université de Rennes

ECOBIO Rennes

Emma Berdan

Pierre de Wit

Nauras Daraghmeh

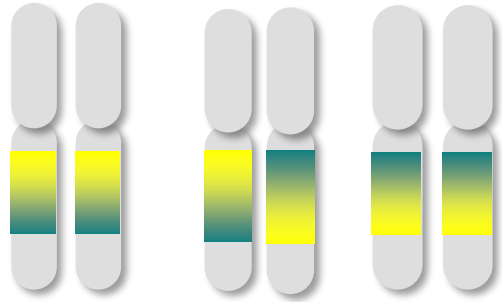
UNIVERSITY OF GOTHENBURG

Multiple polymorphic chromosomal inversions

Ecology ⟷ *Evolution* ⟷ Phenotypes

Genetics

5 polymorphic inversions reported by cytogenetics in a UK population



αα αβ ββ

Cf-Inv(1)

A supergene affecting size & life-history

Aziz, 1971; Butlin et al, 1981,1982,1985; Day et al 1983 Mérot et al 2018

Louis Bernatchez

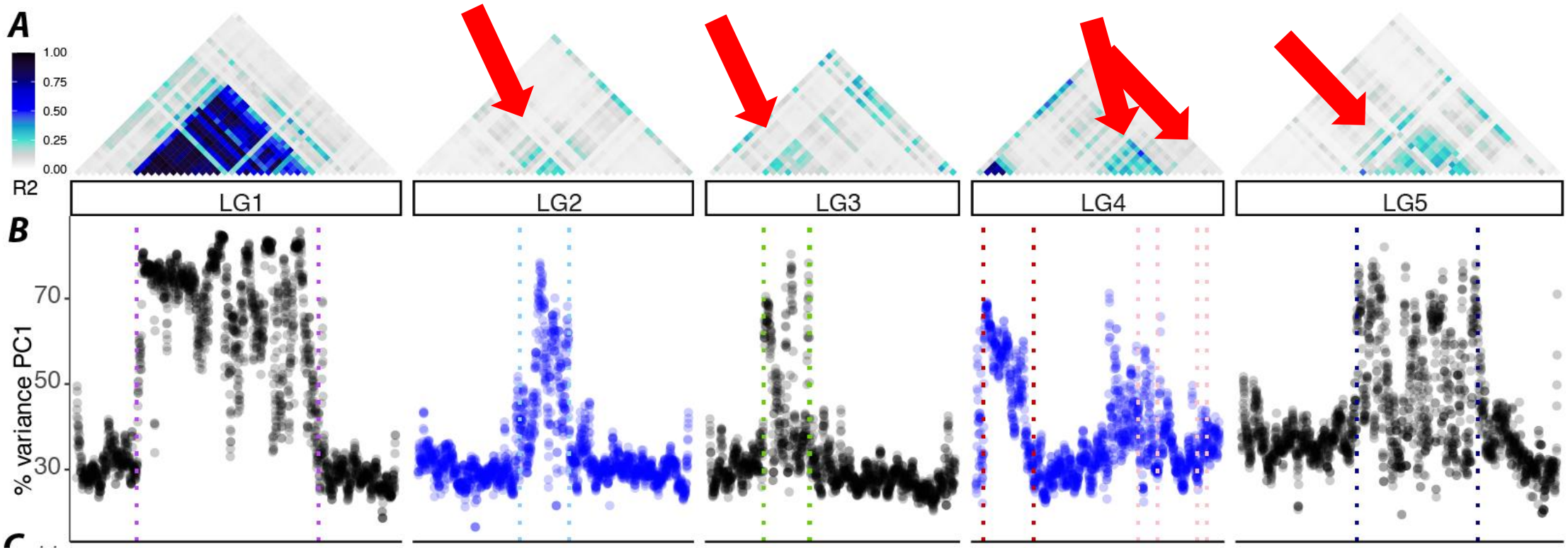
Maren Wellenreuther

⇒ *What do we learn with whole-genome data ?*

Low coverage short-reads
N=1,446 (from America), depth ~1X

Multiple non-recombining blocks in *C. frigida*

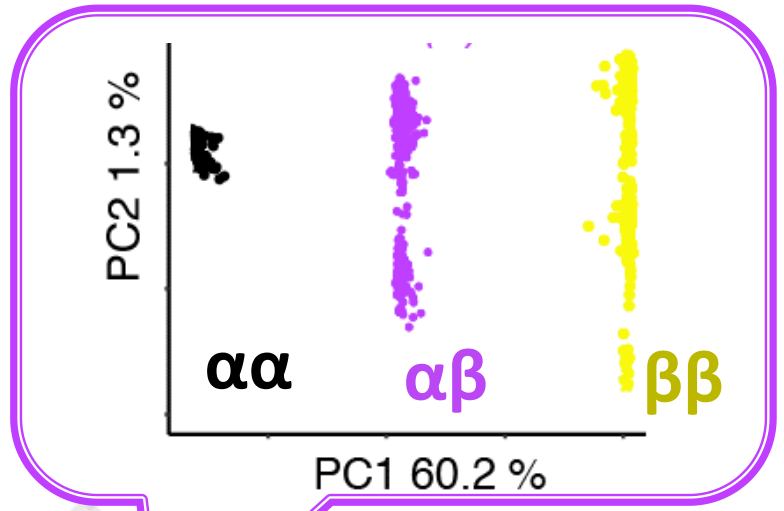
Unresolved yet…

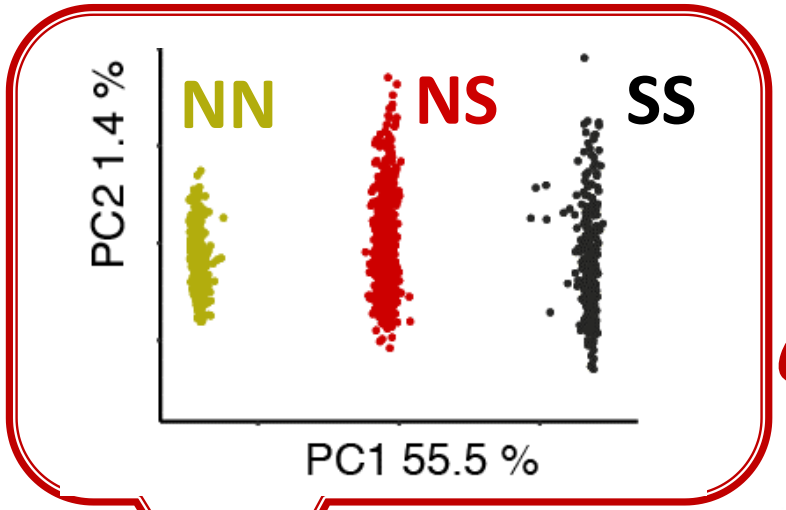


Mérot *et al.* 2021.

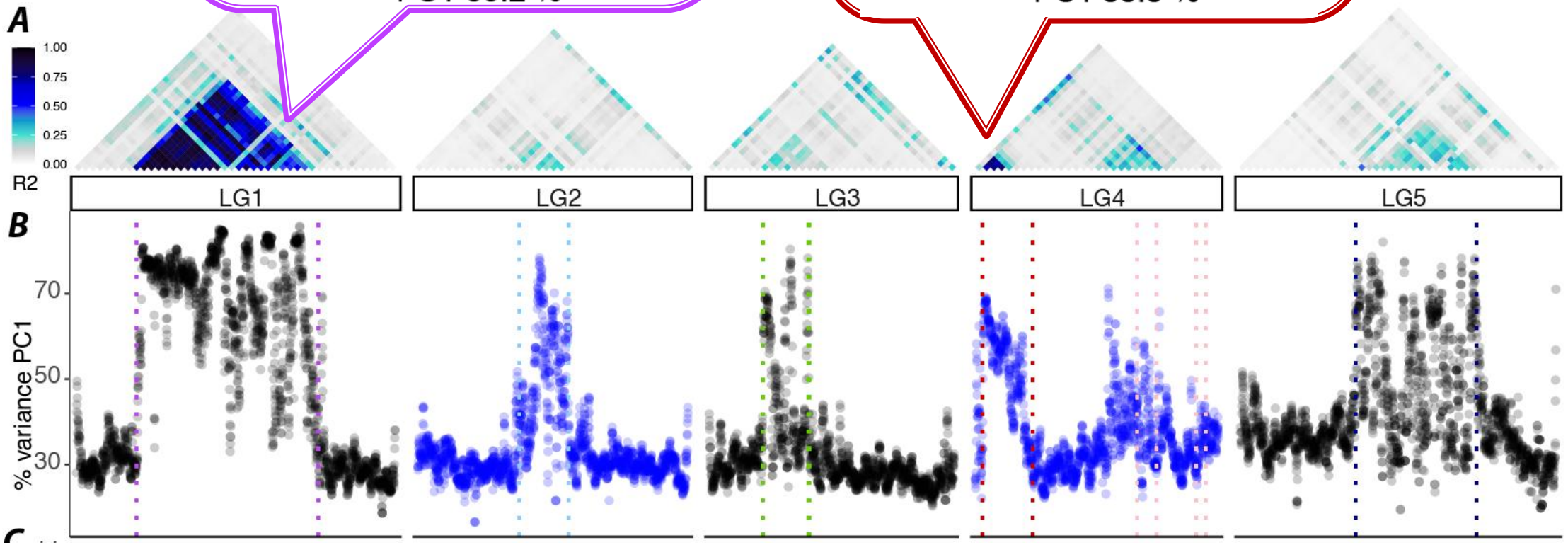
Zoom on 2 inversion polymorphisms in *C. frigida*

Cf-Inv(1)





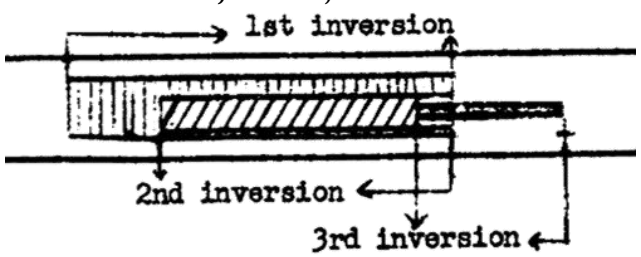
Cf-Inv(4.1)

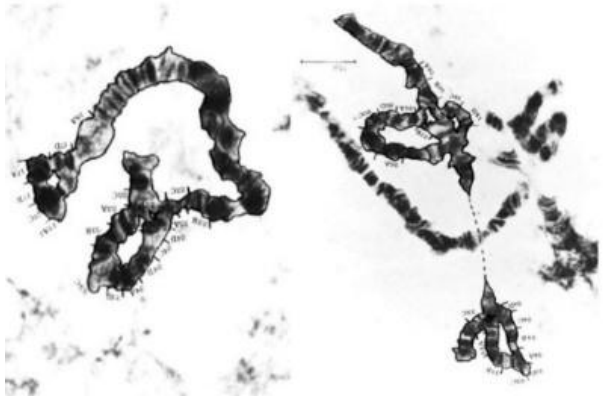


Mérot *et al.* 2021.

Zoom on 2 inversion polymorphisms in *C. frigida*

Aziz, 1971;
Butlin et al,1985;



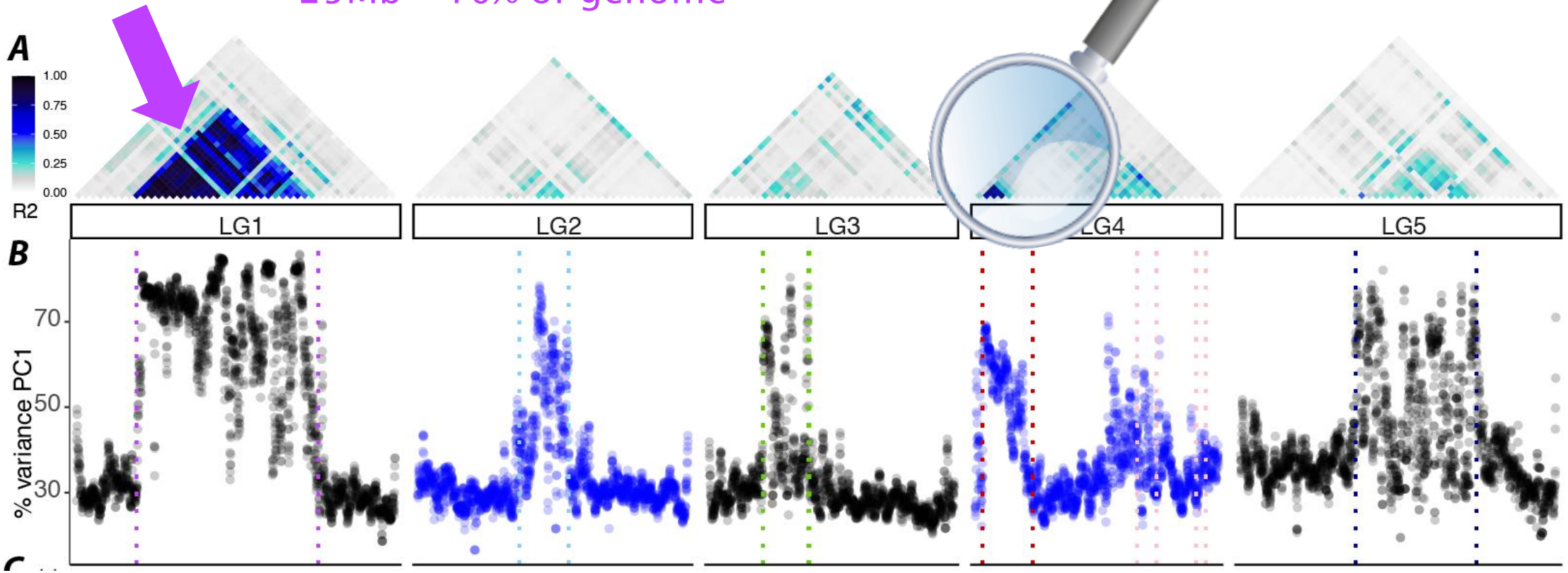


Fantine Benoit

Cf-Inv(1)

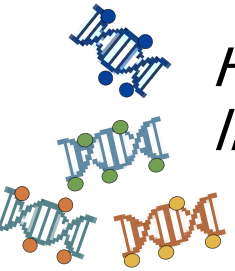
Cf-Inv(4.1)

25Mb – 10% of genome



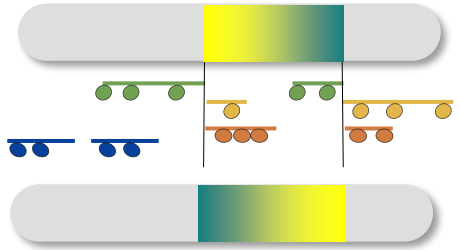
Mérot *et al.* 2021.

How to confirm and precise putative inversions?



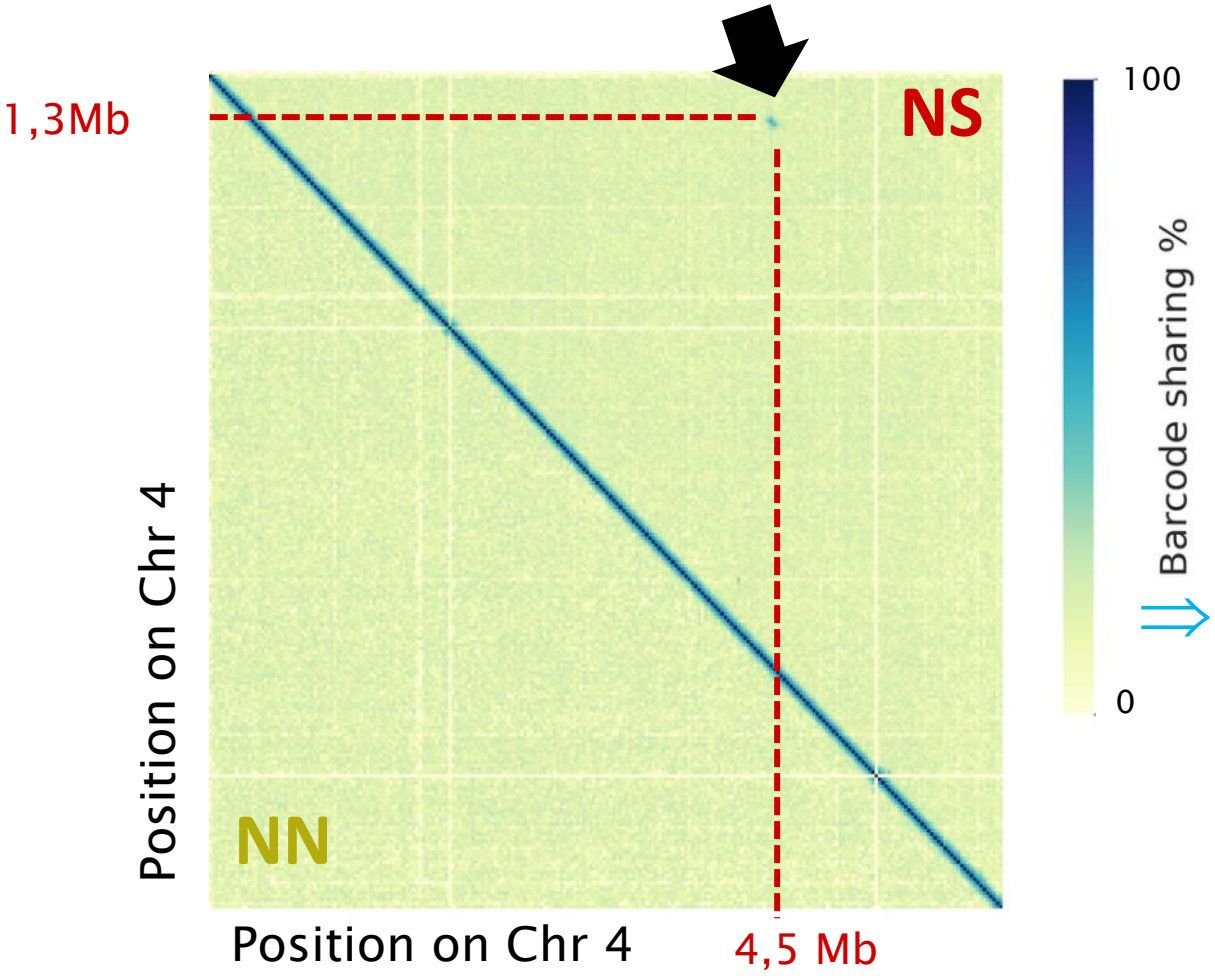
Haplotagging linked-reads

Meier et al 2022



N= 14

Depth = 1-10X

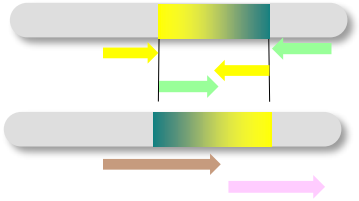


Cf-Inv(4.1)

⇒ Heterogeneity in barcode sharing patterns reveals putative breakpoints of an inversion

How to confirm and precise putative inversions?

Oxford nanopore long reads
N=4, depth ~20X

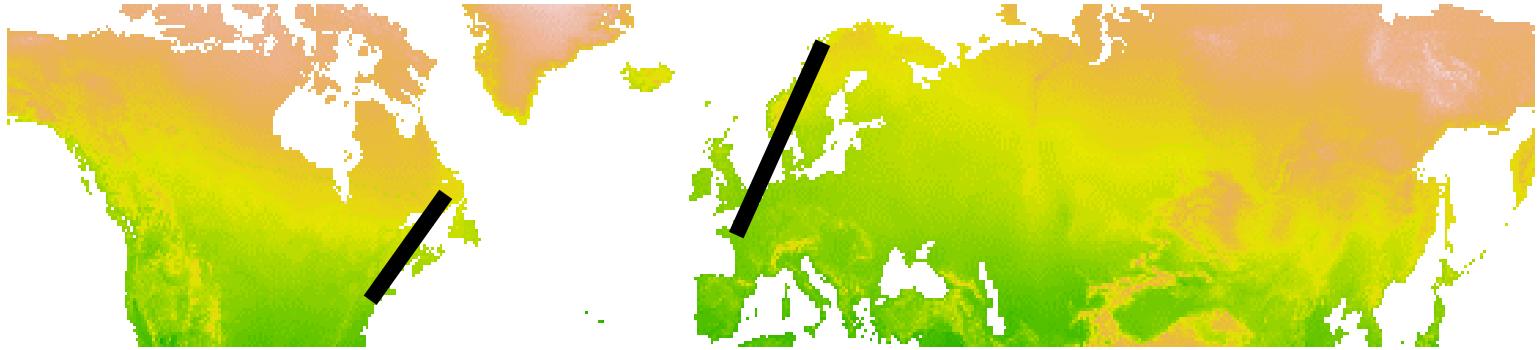


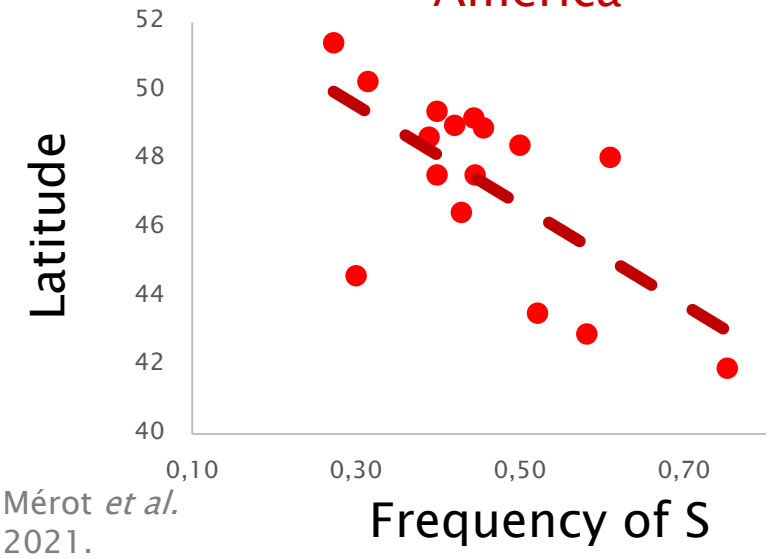
Cf-Inv(4.1)

⇒ Split long-reads confirms the breakpoints of a simple chromosomal inversion

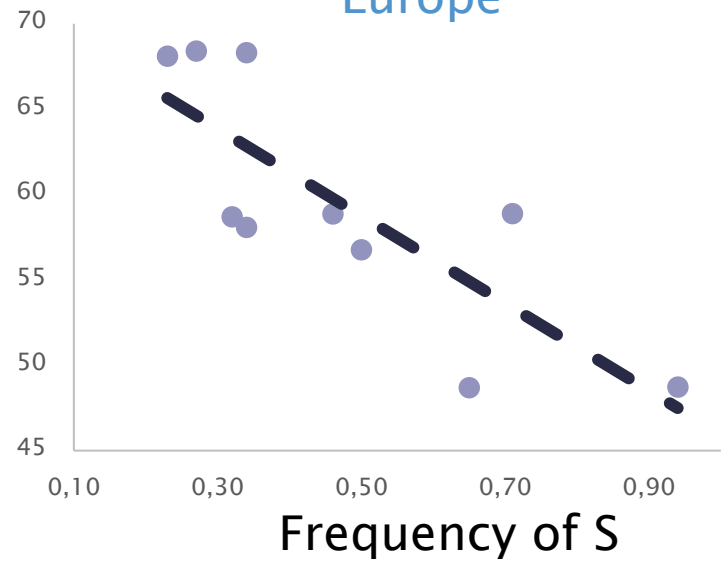
Ecological and geographic distribution of *Cf-Inv(4.1)*

Cf-Inv(4.1)





Mérot *et al.* 2021.

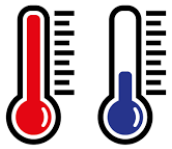


Nicolas *et al.* 2025

⇒ A parallel latitudinal cline in Europe

What is the impact of *Cf-Inv (4.1)* on traits and fitness?

Cf-Inv(4.1)

? 

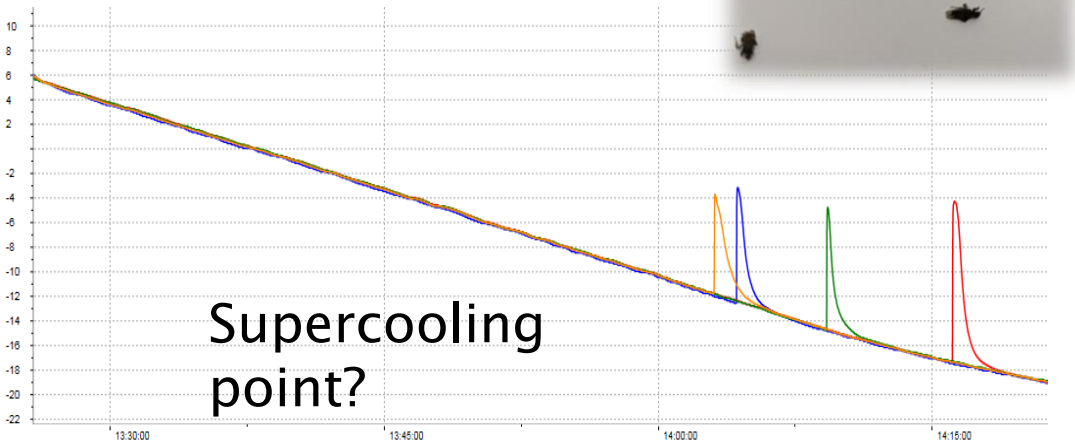
expectation

 **SS > NN**

 **NN > SS**

	30°C	25°C	20°C	15°C	10°C
SS Karyotype	x6	x6	x6	x6	x6
NN Karyotype	x6	x6	x6	x6	x6

Resistance to cold stress?



Supercooling point?

Developmen t time?

Fertility?

Viability?

Longevity?

Nicolas *et al.* 2025

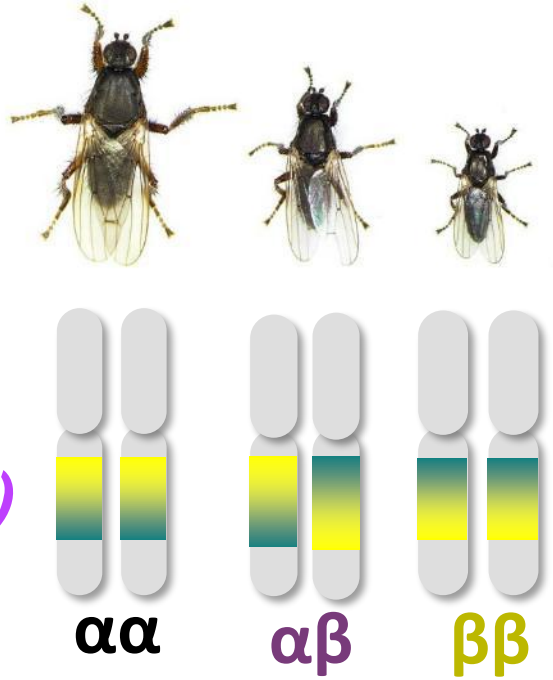
What about inversion 1 – the *supergene*?

Aziz, 1971;
Butlin et al,
1981,1982,1985;
Day et al 1983

Ecology ⟷ *Evolution?* ⟷ Phenotypes

Genetics

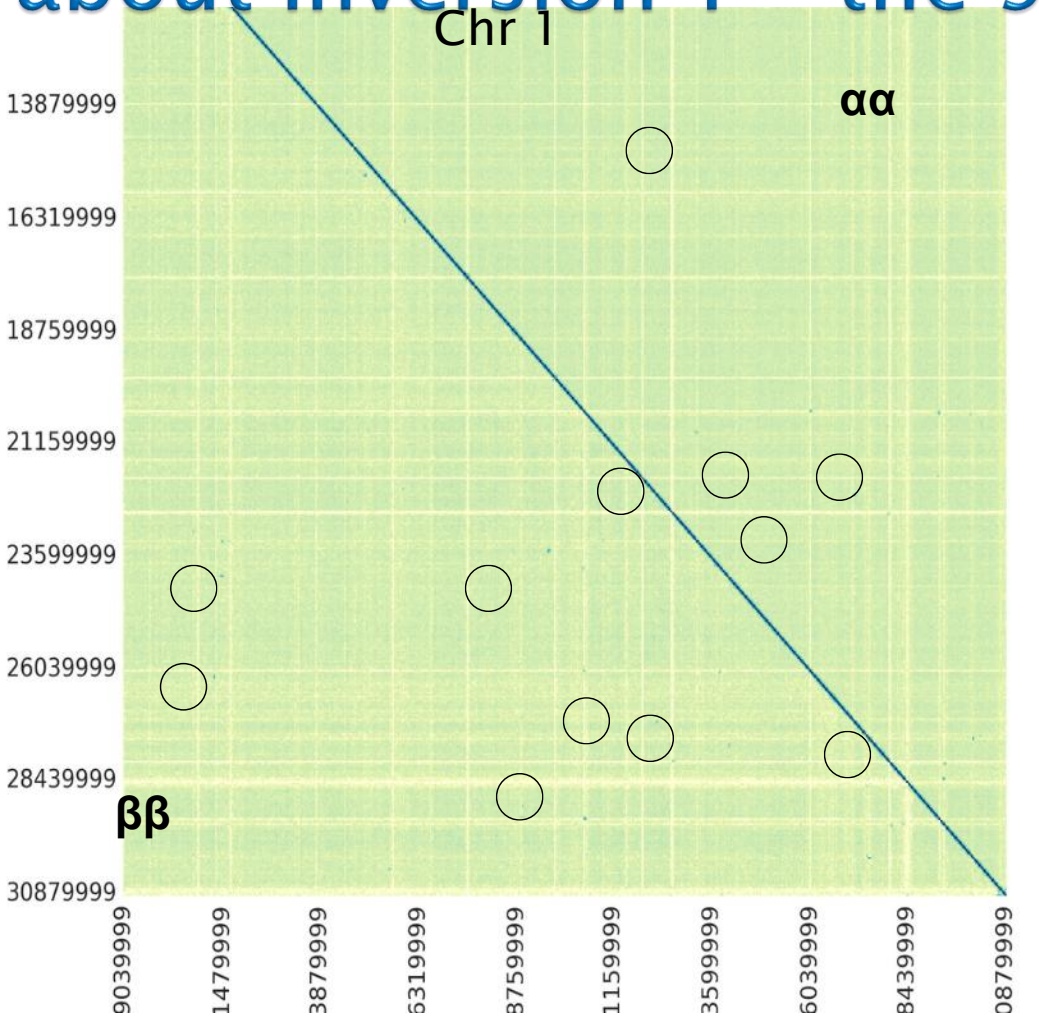
Cf-Inv(1)

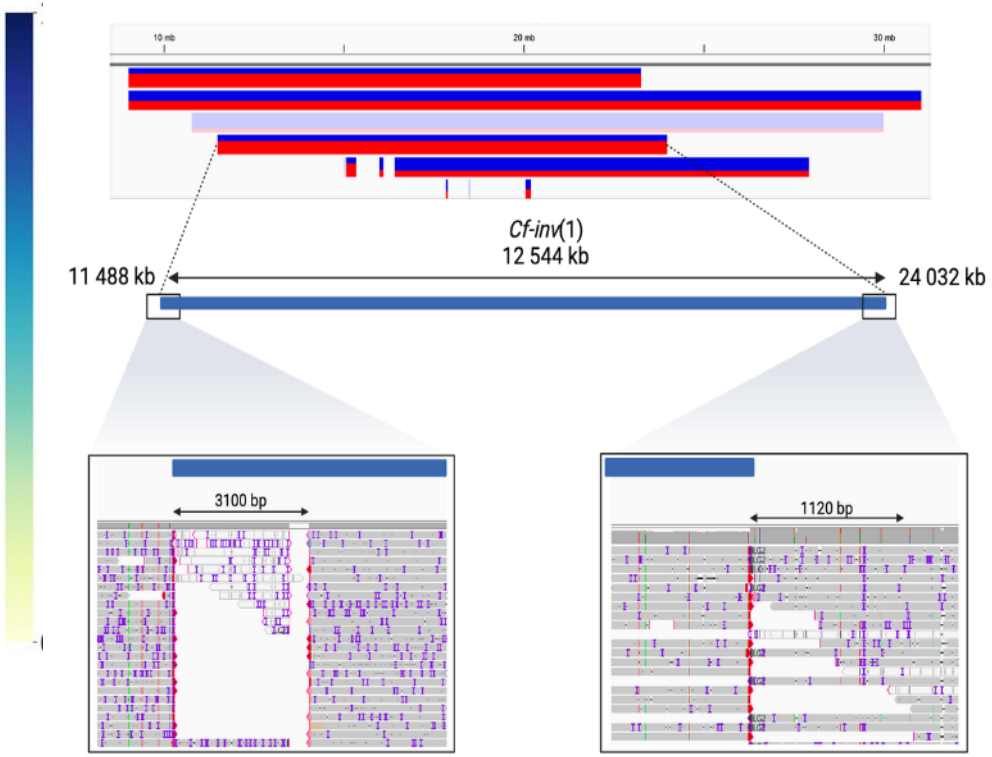


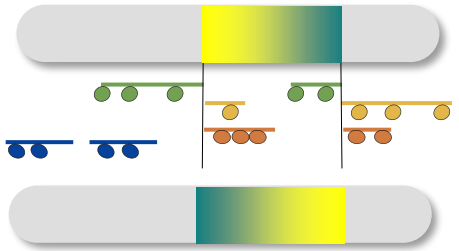
A supergene affecting size & life-history

What about inversion 1 – the *supergene*?

Cf-Inv(1)
When reality challenges bioinformatics







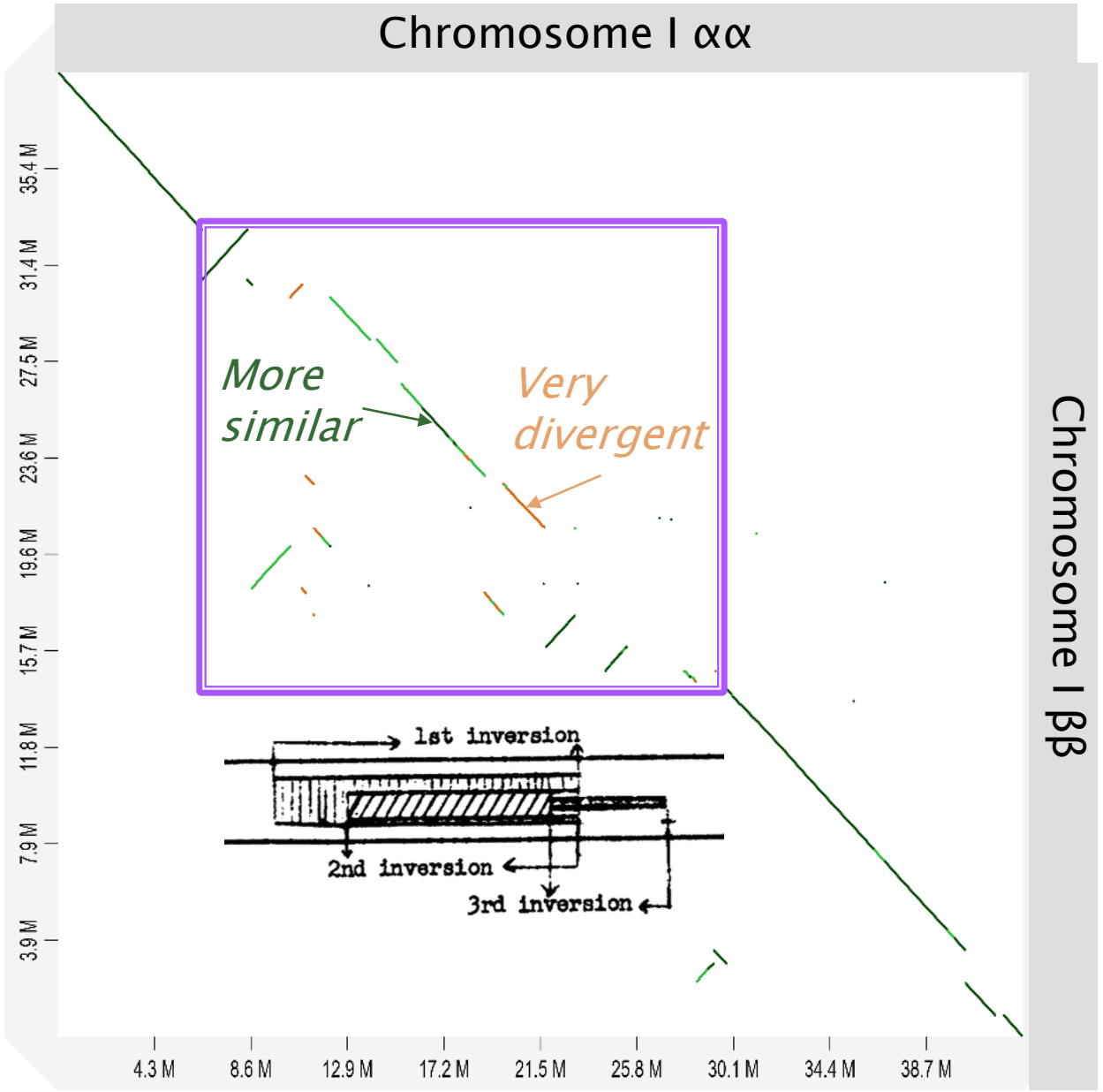
Numerous signals and **unclear** breakpoints

How to confirm and precise chromosomal inversions?

Cf-Inv(1)
When reality challenges bioinformatics

⇒A complex and nested inversion

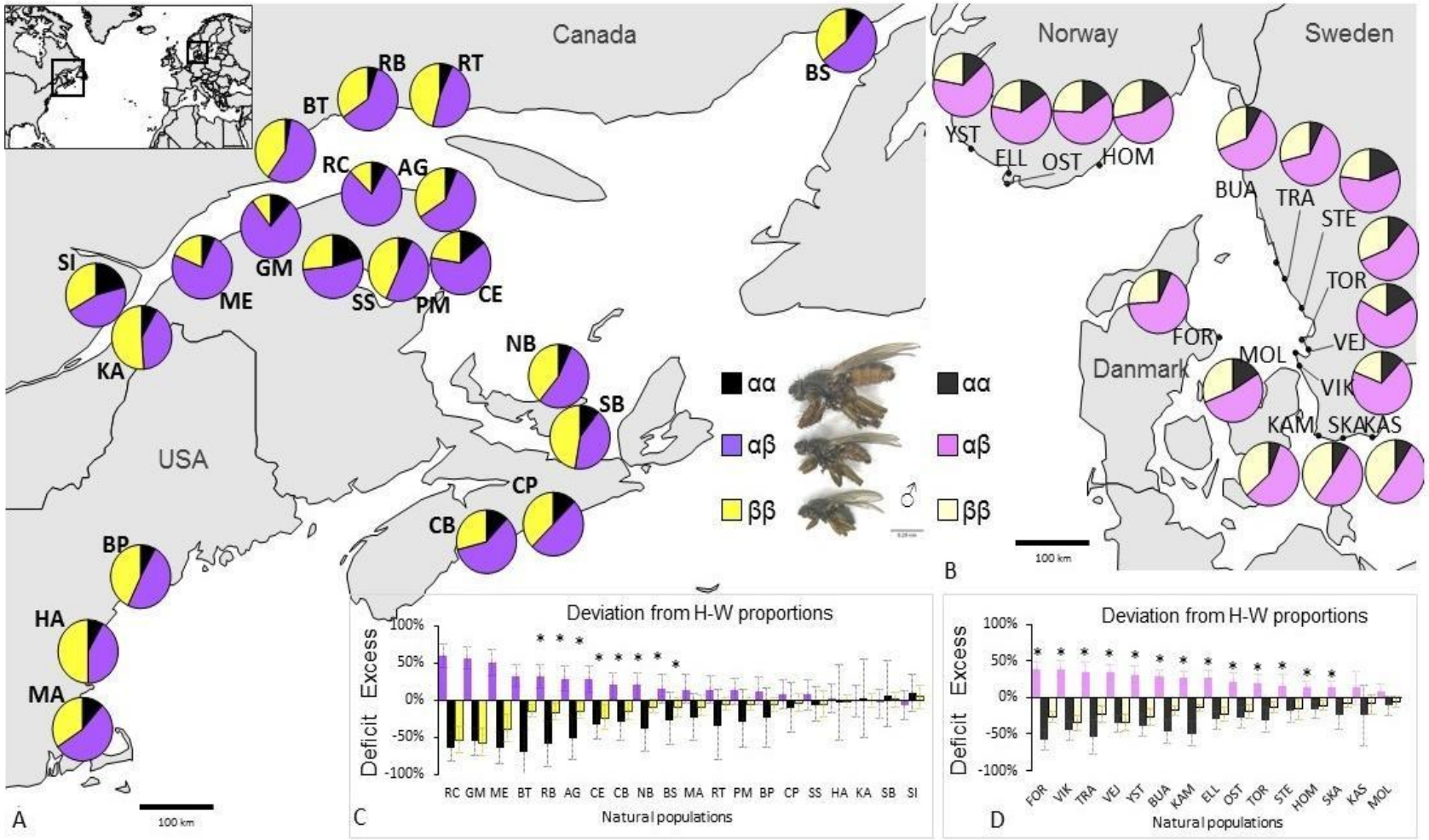
⇒Assemblies to be improved…



What did we learn from the field?

⇒ The inversion is polymorphic in Europe and North-East America

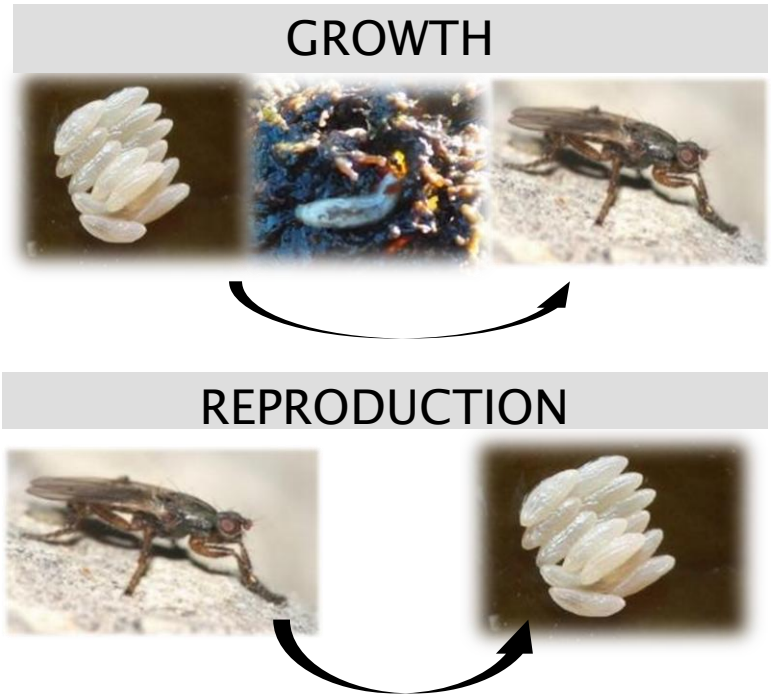
⇒ It co-varies with the composition and depth of the wrackbed habitat

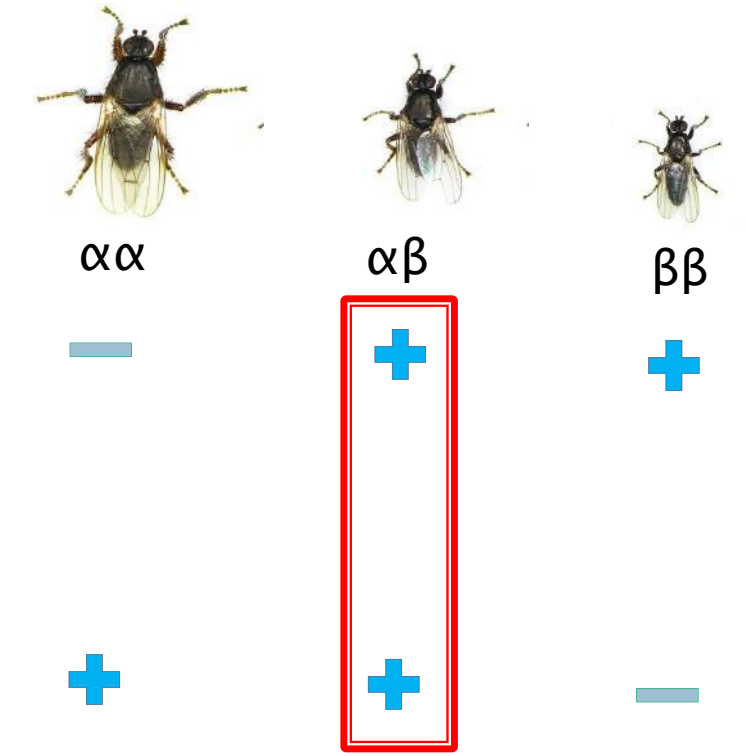


⇒ Strong heterozygote excess

Mérot C, Berdan EL, Babin C, Normandeau E, Wellenreuther M, Bernatchez. 2018. Proc. B 285(1881)

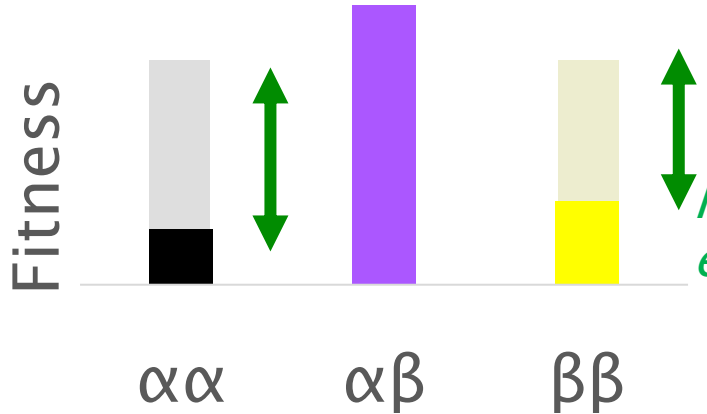
What can we learn from experiments?





⇒A life-history trade-off + overdominance

Balancing selection



Importance of local ecological conditions

Mérot C, Llaurens V, Normandeau E, Bernatchez L, Wellenreuther M. 2020. Nat. Comm.1, 670
https://doi.org/10.1038/s41467-020-14479-7

How is the inversion polymorphism maintained?

Enhance polymorphism maintenance

<u>Life-history traits underlying:</u>

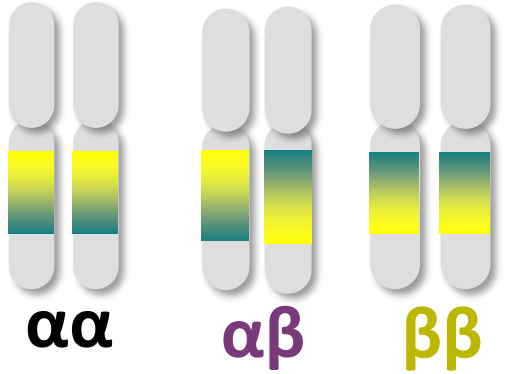
- Trade-off survival/reproduction
- Overdominance
- Non-assortative mating

Temporal and spatial variation in habitat quality and availability

Ecology ⟷ *Evolution?* ⟷ Phenotypes

Genetics

Cf-Inv(1)



Pleiotropic effet
Co-existing "strategies"

Linked haplotypic blocks:

- Highly divergent and rearranged
- Hundreds of genes

Why does haploblock polymorphisms matter for evolution?

A non-exhaustive list…

The more whole-genome data, the more reports of « haploblocks », « superlocus », « putative rearrangements » in evolutionary biology

⇒ **How many more species with haploblock polymorphisms?**

⇒ **How frequent are Mb-scale polymorphic rearrangements ?**

(Or large low-recombining regions?..)

Why is it worth accounting for SVs in population genomics and biogeographic studies?

A cautionary tale in Arctic Char

Xavier Dallaire

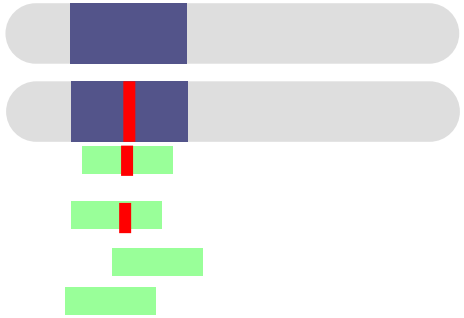
Jean-Sébastien Moore

Dallaire X., Normandeau E., Brazier T., Harris L., Hansen M.M., Mérot C., Moore J-S. 2025. Mol Ecol 34: e17772. https://doi.org/10.1111/mec.17772

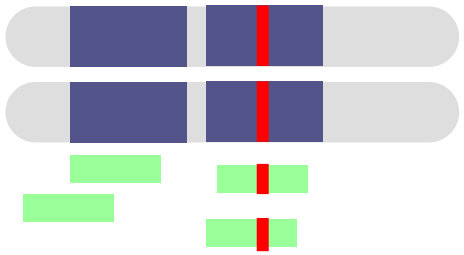
Dallaire X., Bouchard R., Hénault P., Ulmo-Diaz G., Normandeau E., Mérot C., Bernatchez L., Moore J-S. 2023. GBE 15 (12). https://doi.org/10.1093/gbe/evad229

Why accounting for SVs in population genomics?

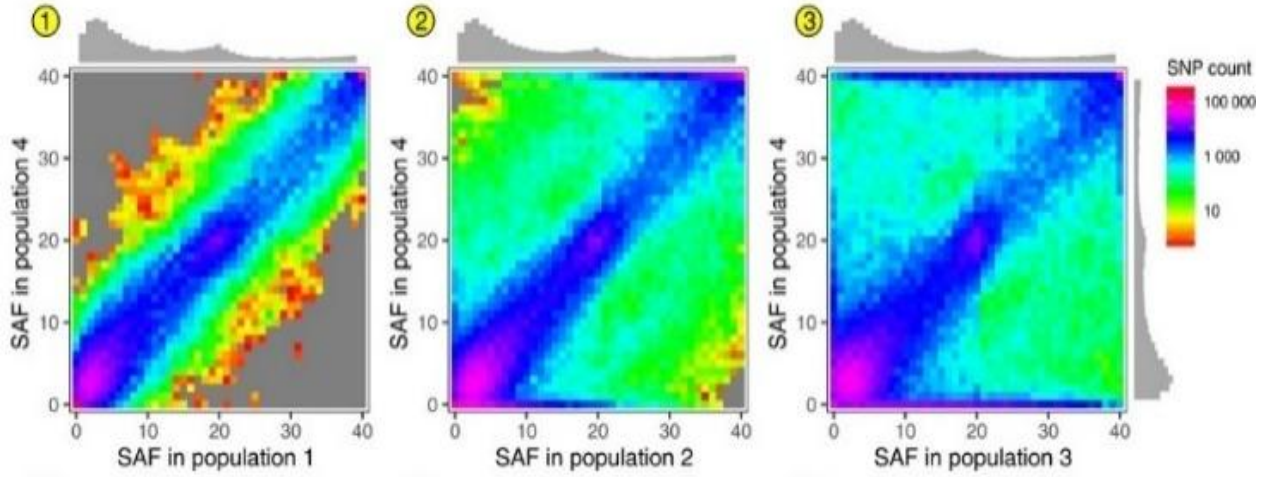
Apparently:
An heterozygote



In reality:
A duplication



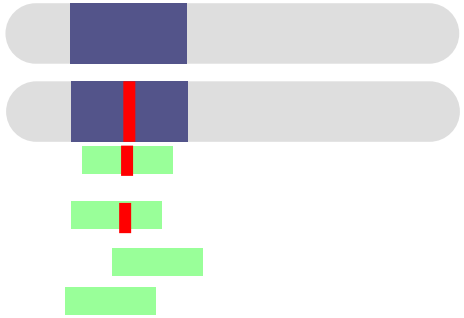
All SNPs



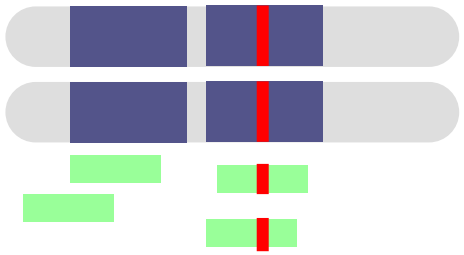
Dallaire et al, 2023

Why accounting for SVs in population genomics?

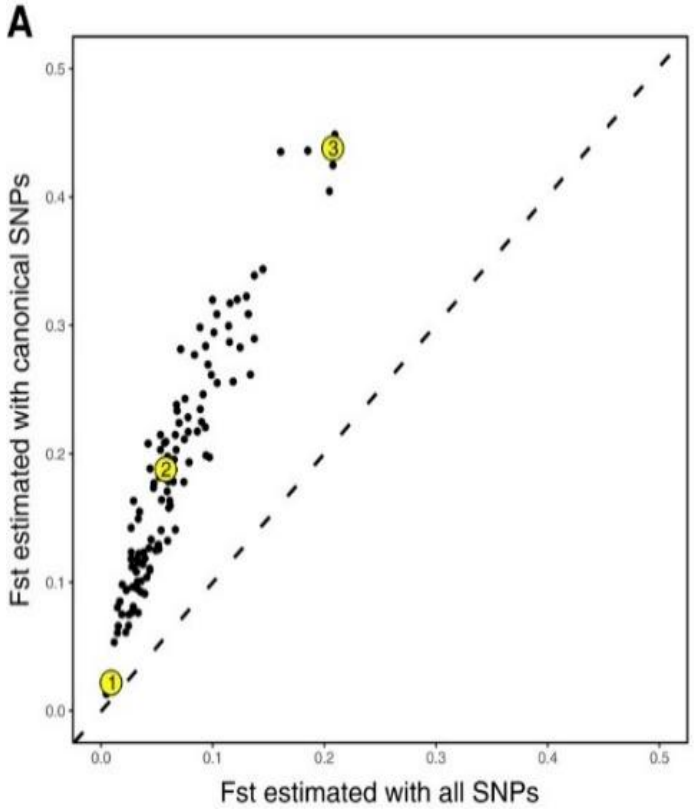
Apparently:
An heterozygote

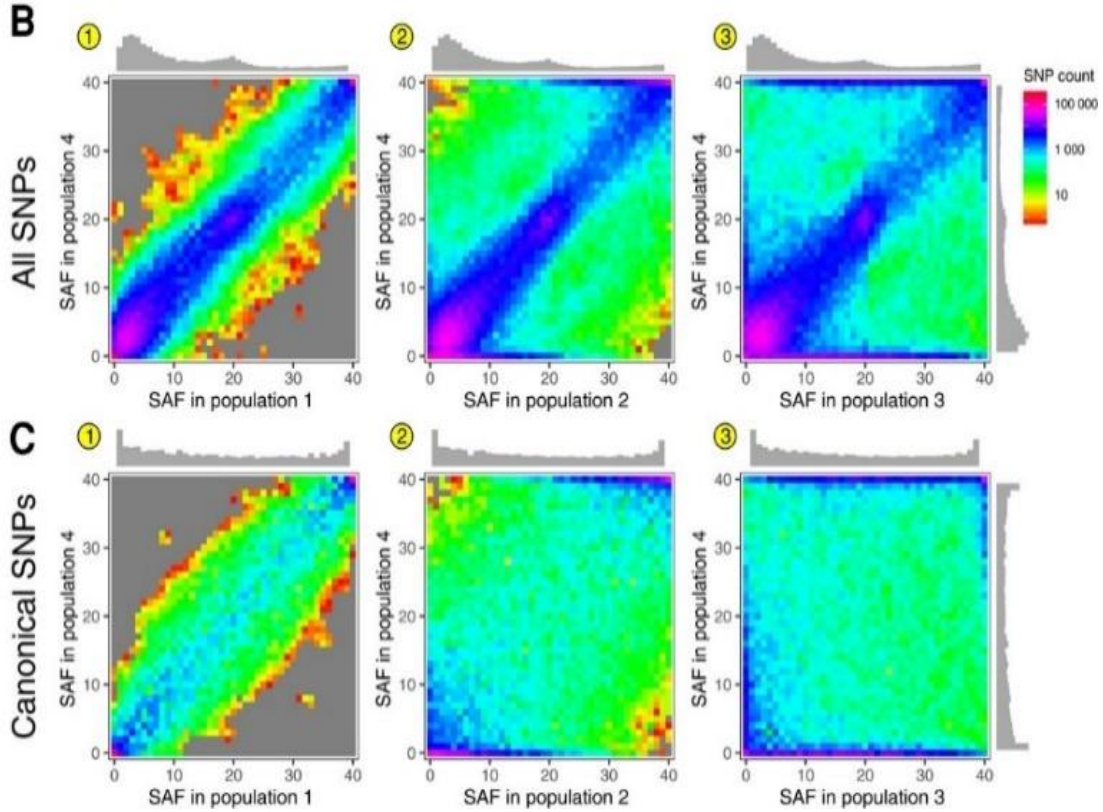


In reality:
A duplication



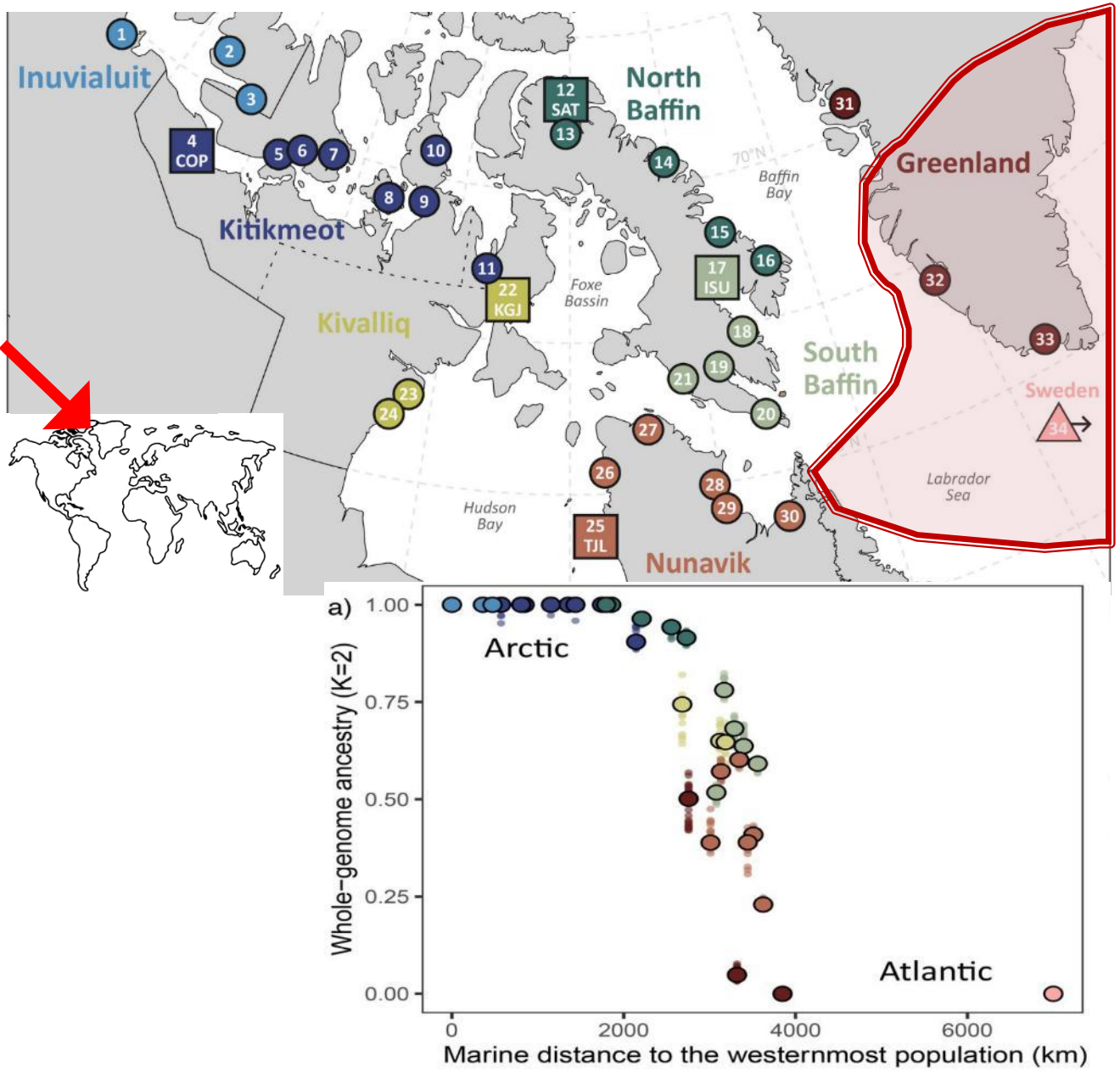
Filtering out duplicated loci remove bias in population genetic analysis

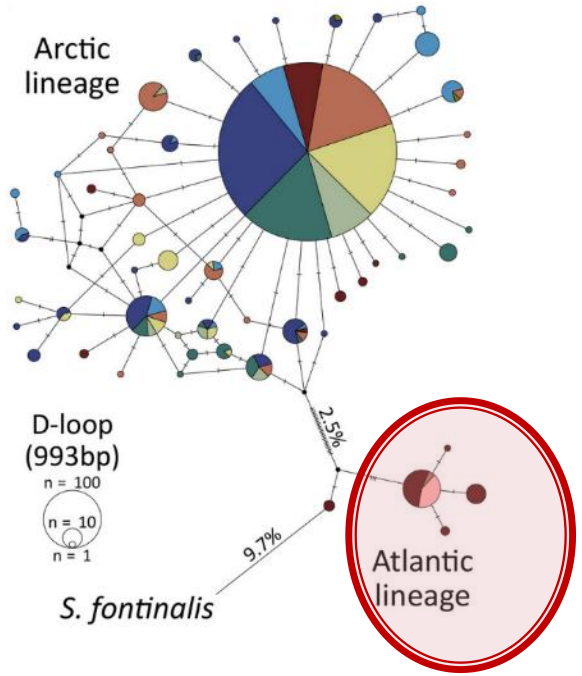




Dallaire et al, 2023

What about SVs in biogeography?

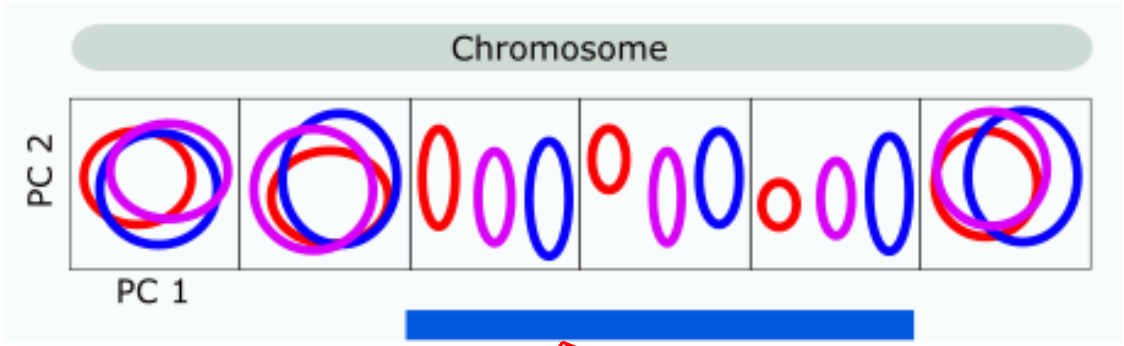


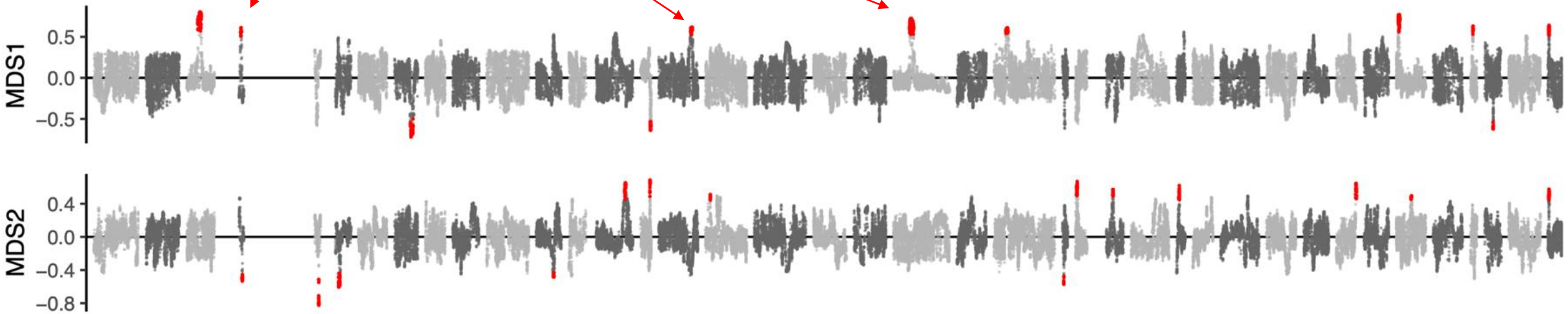


⇒ Contact zone and strong introgression

Dallaire et al, 2025

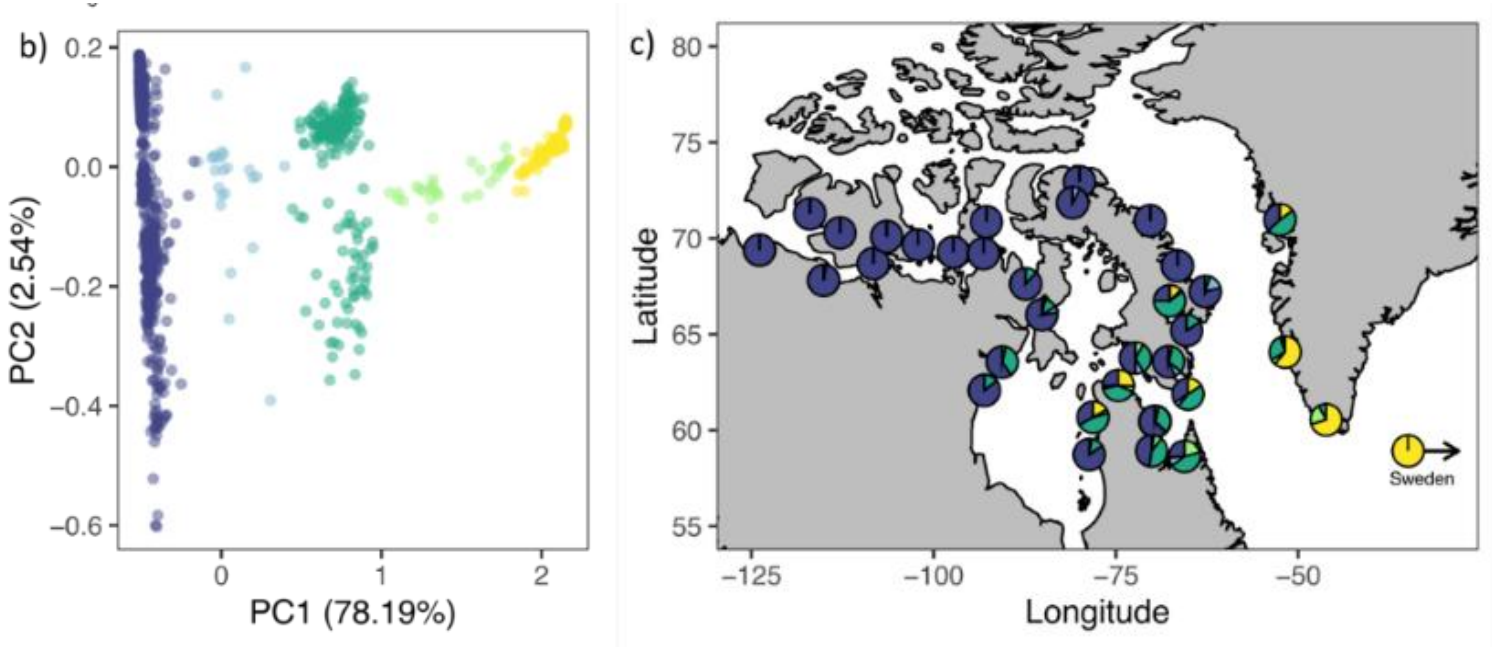
Many haploblocks identified along the genome





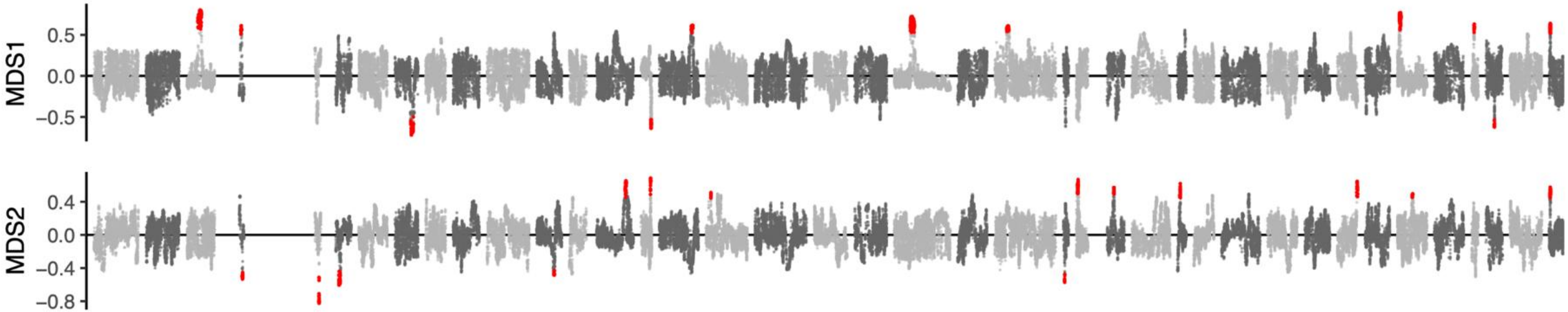
Dallaire et al, 2025

Many haploblocks identified along the genome



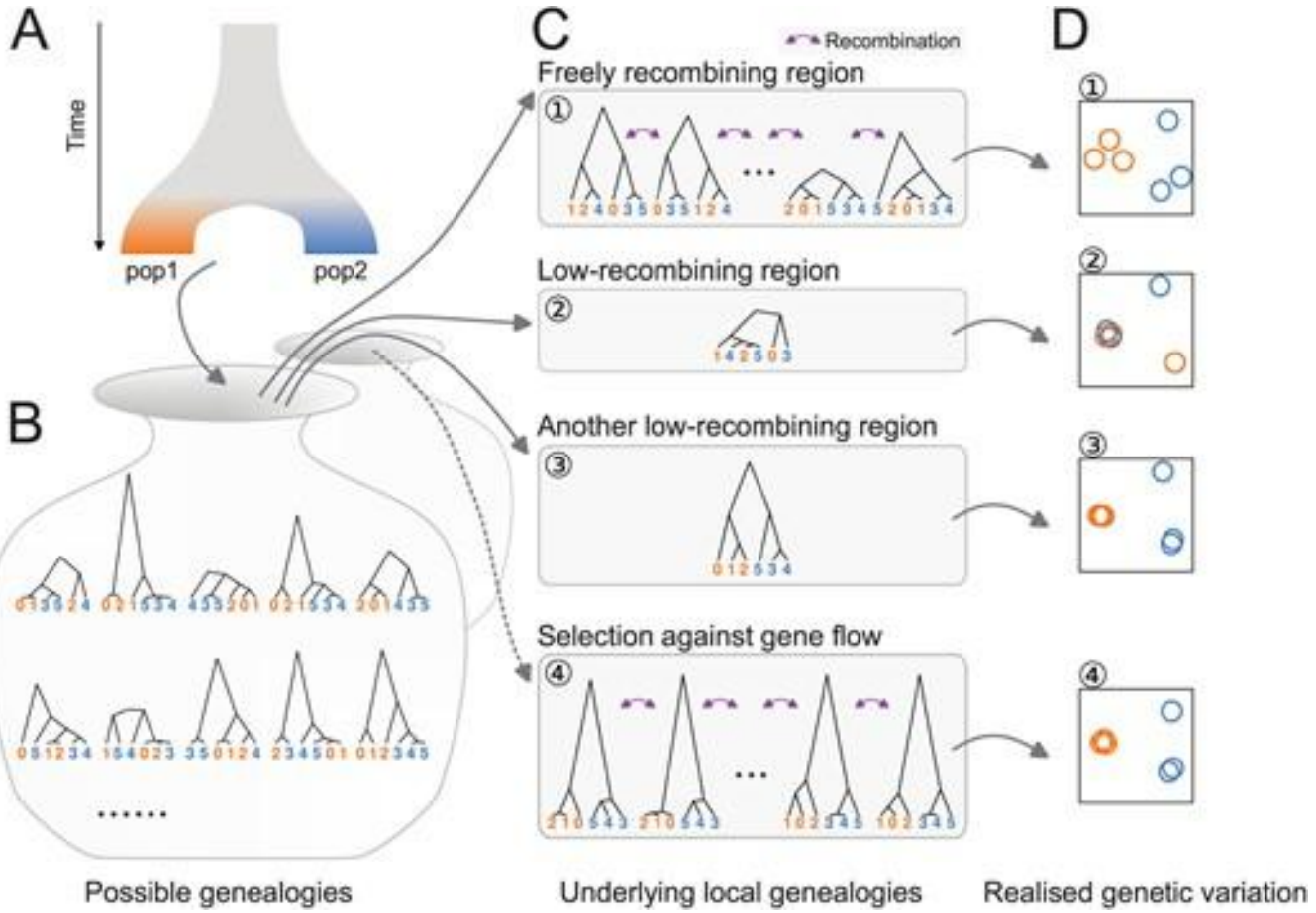
⇒ Most outlier windows have this pattern

⇒ Regions of the genome which resisted gene flow?



Dallaire et al, 2025

Non-recombining regions retain differentiation

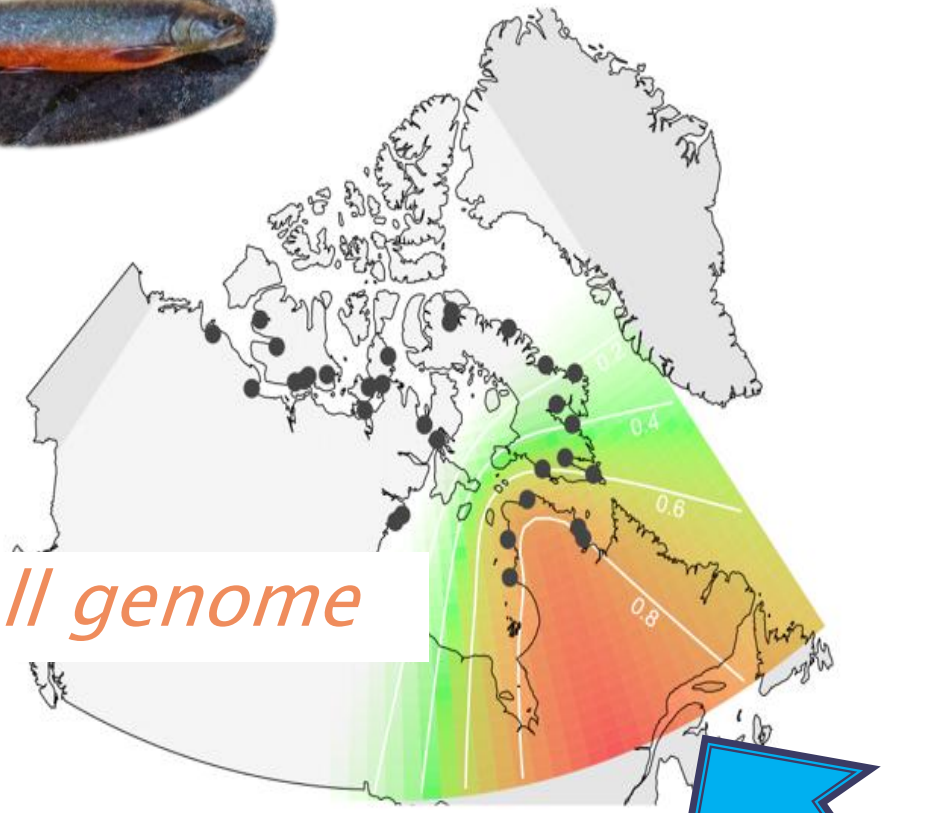


Ishigohoka et al, 2024

⇒ Longer haplotypes with similar genealogy are retained in low-recombination regions.

⇒ To what extent do they represent barriers to reproduction vs. simply extending the time to reshuffle haplotypes?

What about ~~SVs~~ low-recombining blocks in biogeography?

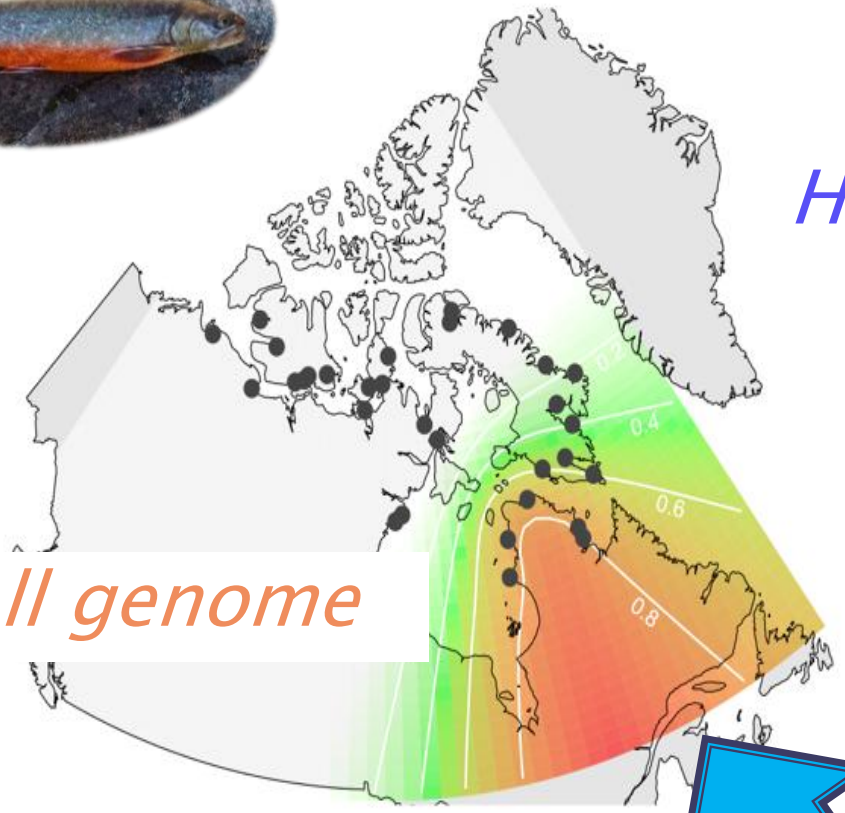


All genome

Colonisation from the South or hybridization?

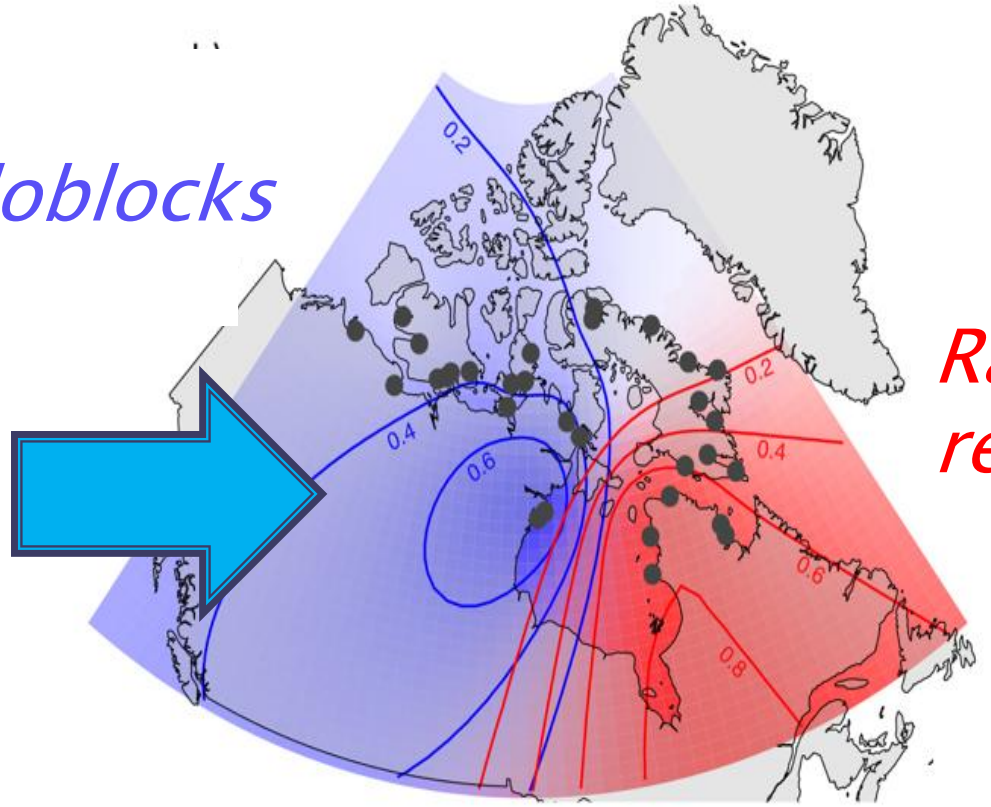
Dallaire et al, 2025

What about ~~SVs~~ low-recombining blocks in biogeography?



All genome

Haploblocks



Random regions

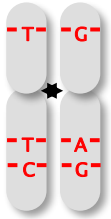
Colonisation from the South or hybridization?

Different insights into the Northern haplotype?

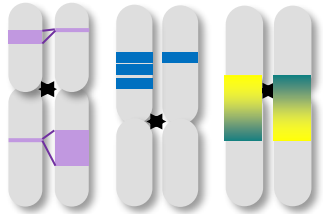
Dallaire et al, 2025

Conclusion – take home message

Several properties make SVs key players of evolution (& variants to consider in population genomics)

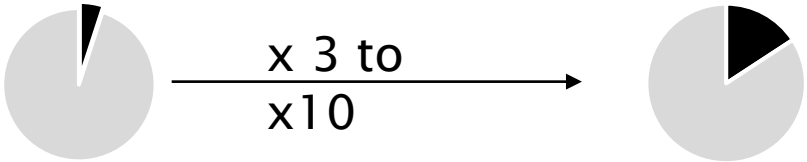


SNPs



SVs

- Fraction of genome covered by variants

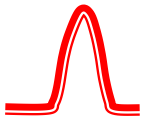


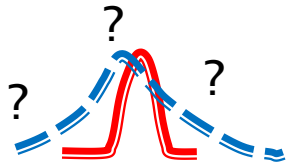
- Length, direct and indirect effects on fitness

1 bp

Up to Mb
⇒ Frequently deleterious

- Speed of evolution





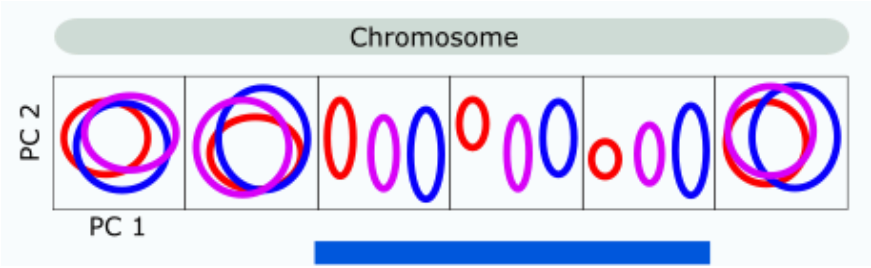
- Recombination impact

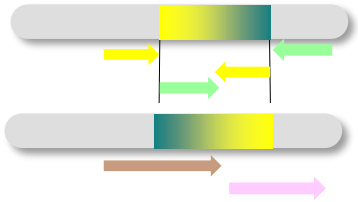
no

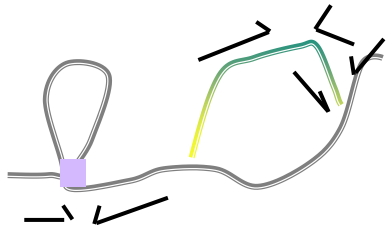
Can be reduced

Conclusion – take home message

Combination of methods to detect/genotype/confirm/analyse SV in pop genomics



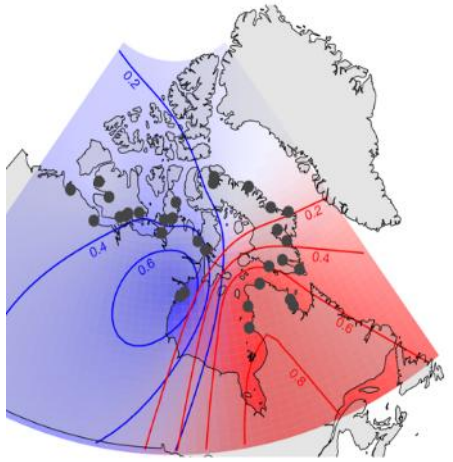




TE in SV

Example of SV implication in speciation, local adaptation and balanced polymorphism

Conclusion – take home message

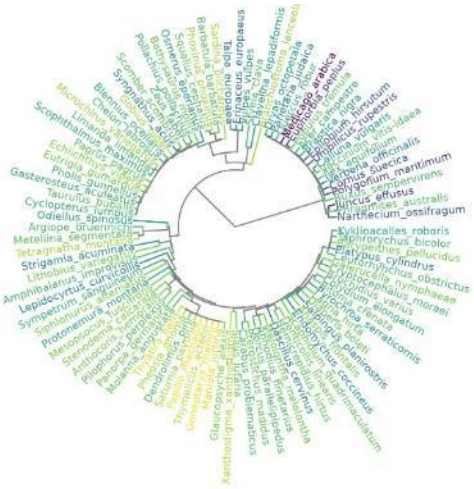


Warnings:

- Comparable data matters
- Not all haploblocks are divergent inversions
- Beware of SVs even in SNPs datasets

Hopes:

- Large-scale comparisons are made possible by new data
- SVs bear extra information about populations



Many open questions…

- *Can we improve SV detection to analyze **the whole spectrum of variants** across large sample datasets?*
- *Which **characteristics** make some SVs particularly involved in adaptation and speciation?*
- *How do **neutral** and **adaptive** processes determine the evolutionary trajectory of SVs?*
- *What are the **abundances**, **diversities**, and **distributions** of SVs in natural populations & across taxa?*
- *What is the **evolutionary rate** of different SVs?*

Structural variants in evolutionary biology…

A special topic network about SVs

-> share ideas and methodologies
-> run comparative/meta analysis
-> build a community

Starting /coming soon …

- A special issue in JEB
- Online seminars
- A summer meeting in Portugal
- Collaborative projects

eseb
European Society for Evolutionary Biology

JOURNAL OF Evolutionary Biology

Adressing:

- *Can we improve SV detection to analyze **the whole spectrum of variants** across large sample datasets?*
- *Which **characteristics** make some SVs particularly involved in adaptation and speciation?*
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- *What are the **abundances**, **diversities**, and **distributions** of SVs in natural populations & across taxa?*
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To contact me:
claire.merot@gmail.com
claire.merot@univ-rennes.fr

EVOL-SV
(101115983)







Many thanks to all colleagues and collaborators

Thank you for your attention

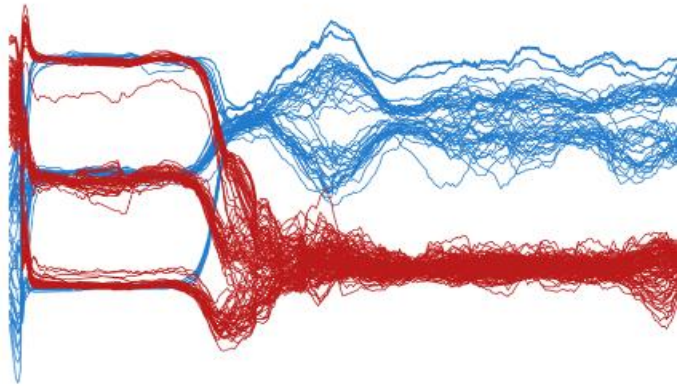


Questions?
Suggestions?

Hands-on lab on structural variants

Claire Mérot
claire.merot@univ-rennes.fr

EVOMICS 2026



Data

Seaweed flies genomes
(Coelopa frigida)

Assemblies
-> 2 genomes
(ref.fasta)
(alt.fasta)

Subset of 10Mb

Long-reads
-> 1 sample (fri59)
ONT – Oxford Nanopore
~20X

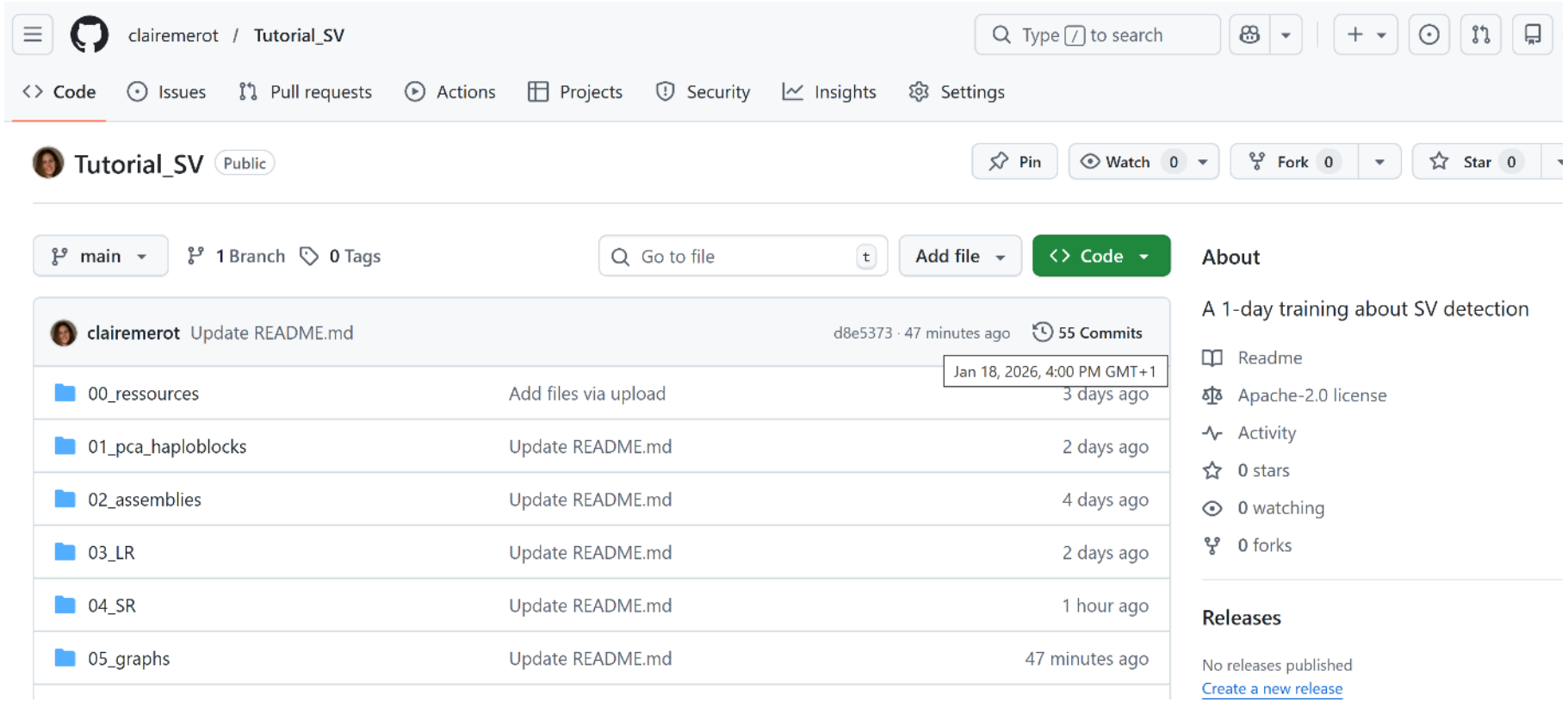
Short-reads
-> 22 samples (illumina PE 150bp)
~10X
1 pair of fastq reads (A_R1, B_R2)
4 bamfiles (A, B, C, D)
1 vcf of SNPs from the 22 samples (A to V)

Plan

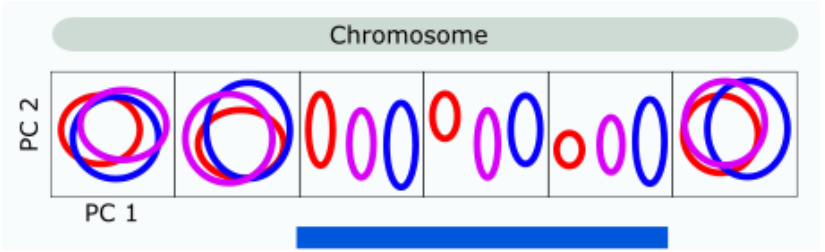
Work at your own pace
Take time to explore and visualise your data
Tutorial wrap-up around 4:30 pm?

Tutorial

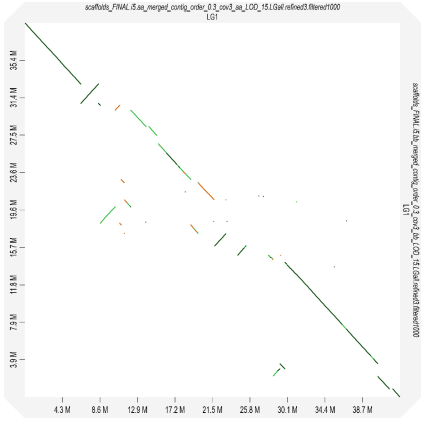
https://github.com/clairemerot/Tutorial_SV



Outline

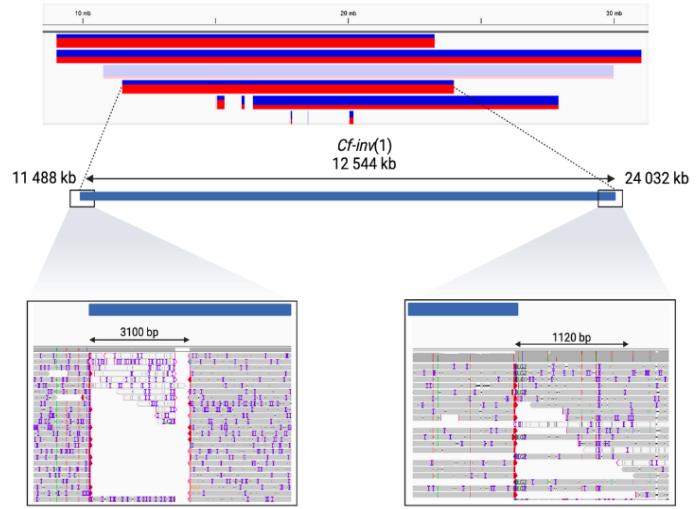


01 - Using SNPs & local PCA to detect haploblocks putatively representing large rearrangements



02 - Comparing assemblies for large rearrangements

03 - Assessing breakpoints and detecting other SVs with long-reads



04 - Using population dataset of short-reads to detect SVs

05 - Towards graph-based analysis