



**HAL**  
open science

## Population genomics reveal PRDM9-dependent recombination landscapes in salmonids

Marie Raynaud, Pierre-Alexandre Gagnaire, Nicolas Galtier

► **To cite this version:**

Marie Raynaud, Pierre-Alexandre Gagnaire, Nicolas Galtier. Population genomics reveal PRDM9-dependent recombination landscapes in salmonids. Congress of the European Society for Evolutionary Biology, Aug 2022, Prague, Czech Republic. hal-04785845

**HAL Id: hal-04785845**

**<https://hal.science/hal-04785845v1>**

Submitted on 15 Nov 2024

**HAL** is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.

# Fast evolving recombination landscapes support a role of PRDM9 in directing hotspot location in Salmon genomes

RAYNAUD M. (1), GAGNAIRE P.-A.(1), GALTIER N.(1)

(1) ISEM, Univ Montpellier, CNRS, EPHE, IRD, Montpellier, France

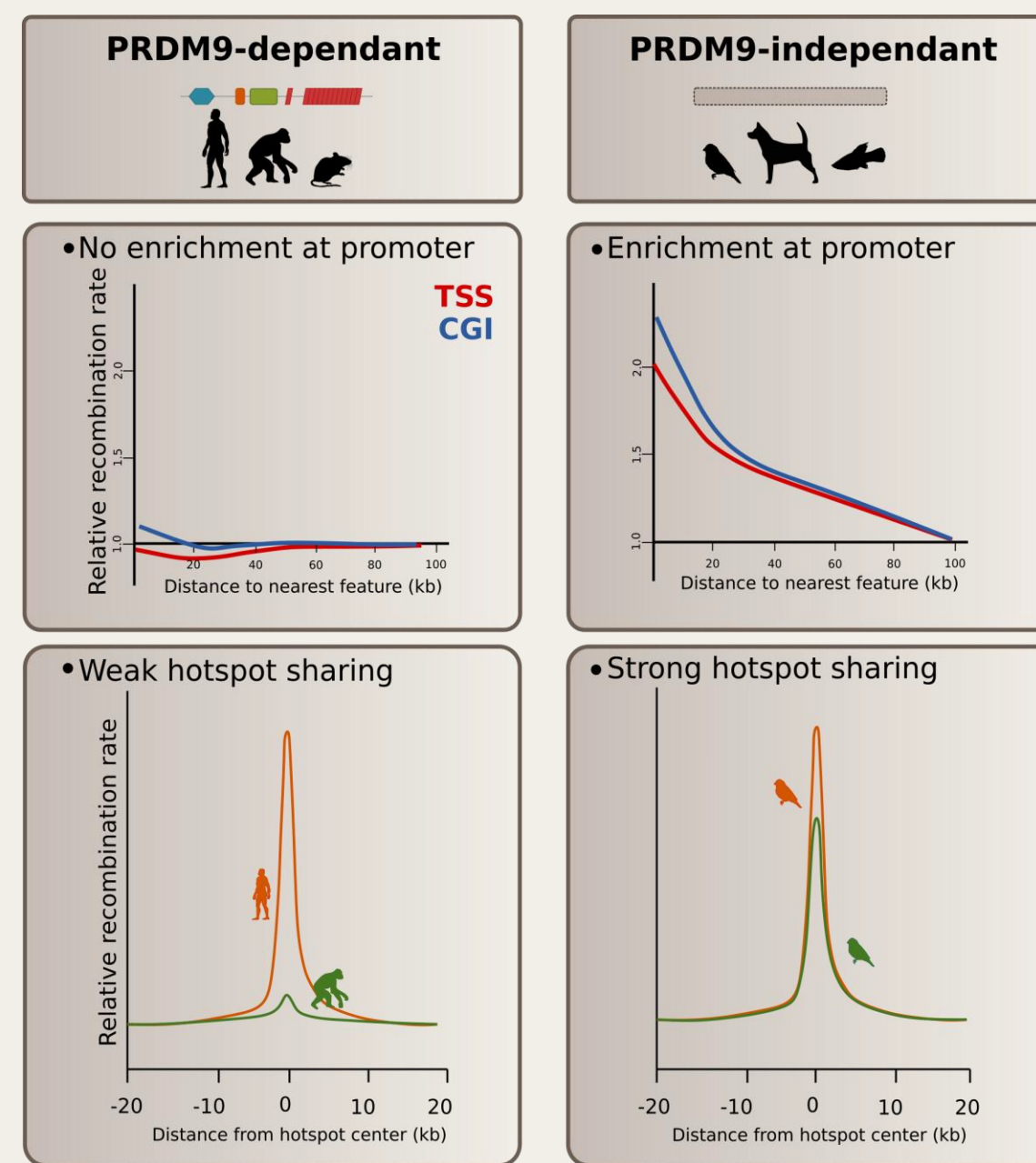
Congress of the European Society for Evolutionary Biology - August 14-19, 2022 – Prague Congress Centre

## Introduction

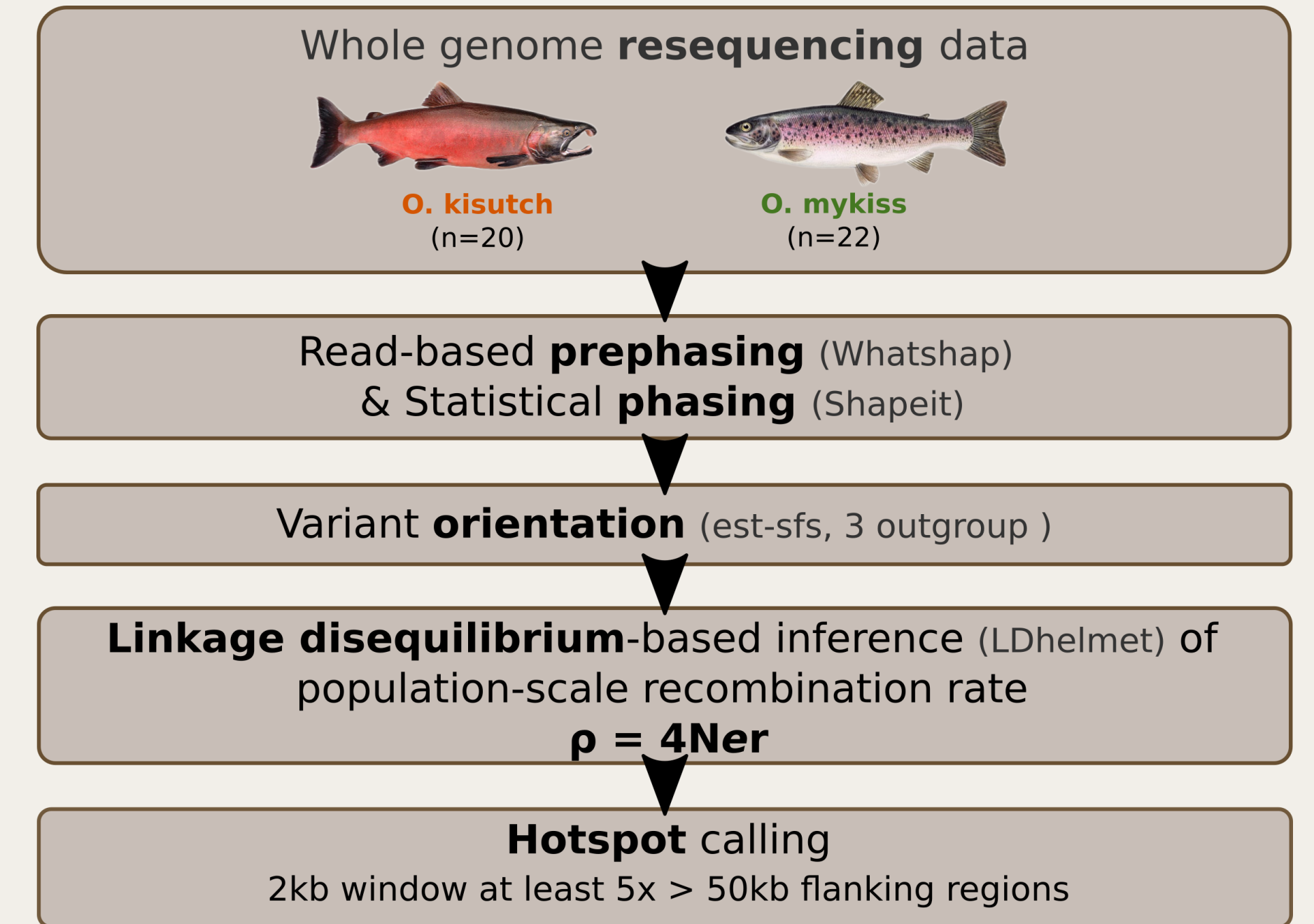
- Recombination has a major role in shaping genome evolution.
- Recombination events tend to concentrate at hotspot positions in a wide range of species.
- PRDM9 protein determines recombination events in mammals and leads to rapid evolution of their recombination landscapes (Myers et al., 2010; Auton et al., 2012). By contrast, it has been lost in other taxa which show conserved hotspots (e.g. birds, dogs, teleosts; Singhal et al., 2015; Auton et al., 2013; Baker et al., 2017).
- Full-length copies of PRDM9 have been found in Salmonids, but their function is still unknown.

Is the regulation of recombination by PRDM9 ancestral in the animal kingdom?

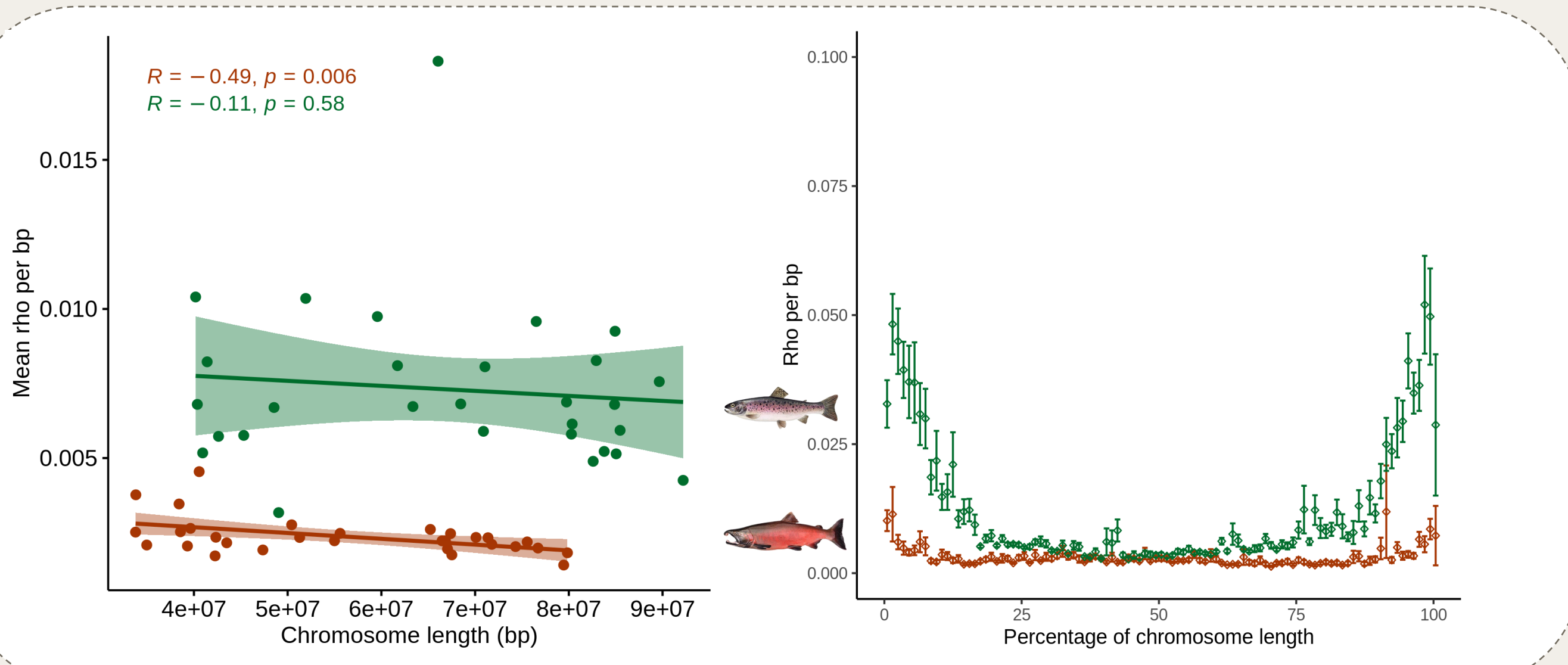
Characterization of the recombination landscapes and their evolution in the coho salmon *Oncorhynchus kisutch* and the rainbow trout *Oncorhynchus mykiss*



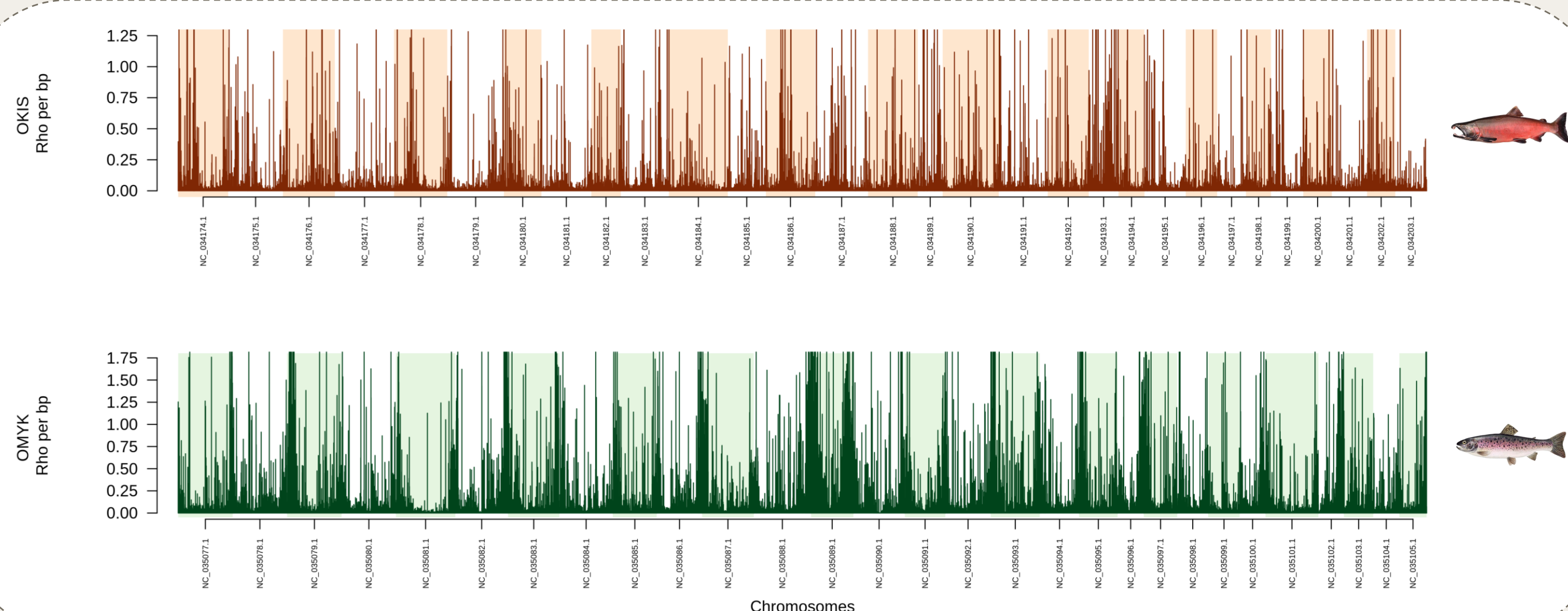
## Materials and Methods



## Inter and intra chromosomal variation in recombination rates

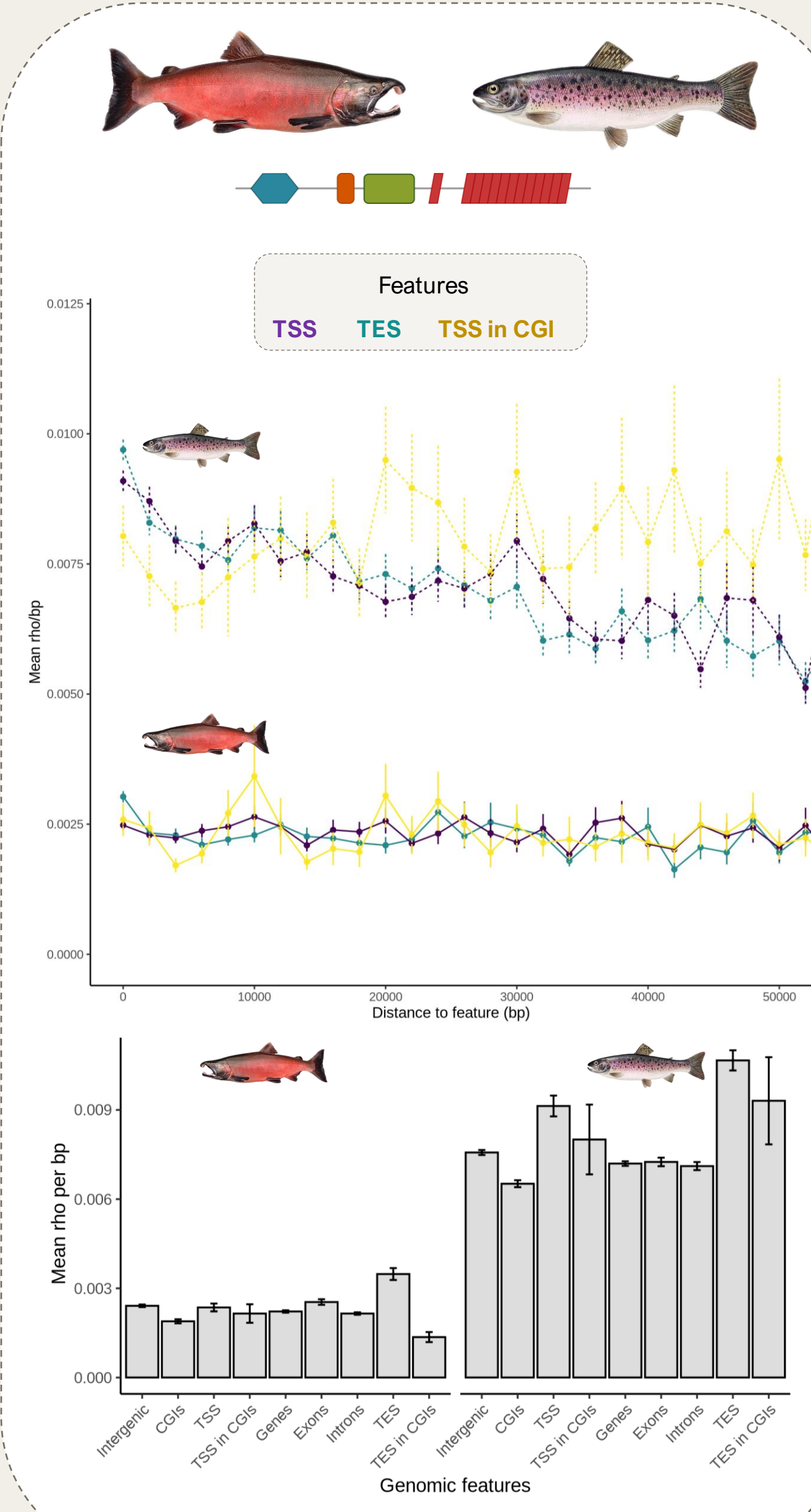


Averaged recombination rates  $\rho$  decrease with chromosome length and with distance to telomeres. One crossing-over (CO) per meiosis per chromosome is obligatory to ensure their proper segregation, but more COs usually prevented by chromosomal interference. Thus small chromosomes display higher recombination rates. Heterochiasmy and interference can explain the deficit of recombination in centro-chromosomal regions as compared to telomeres.



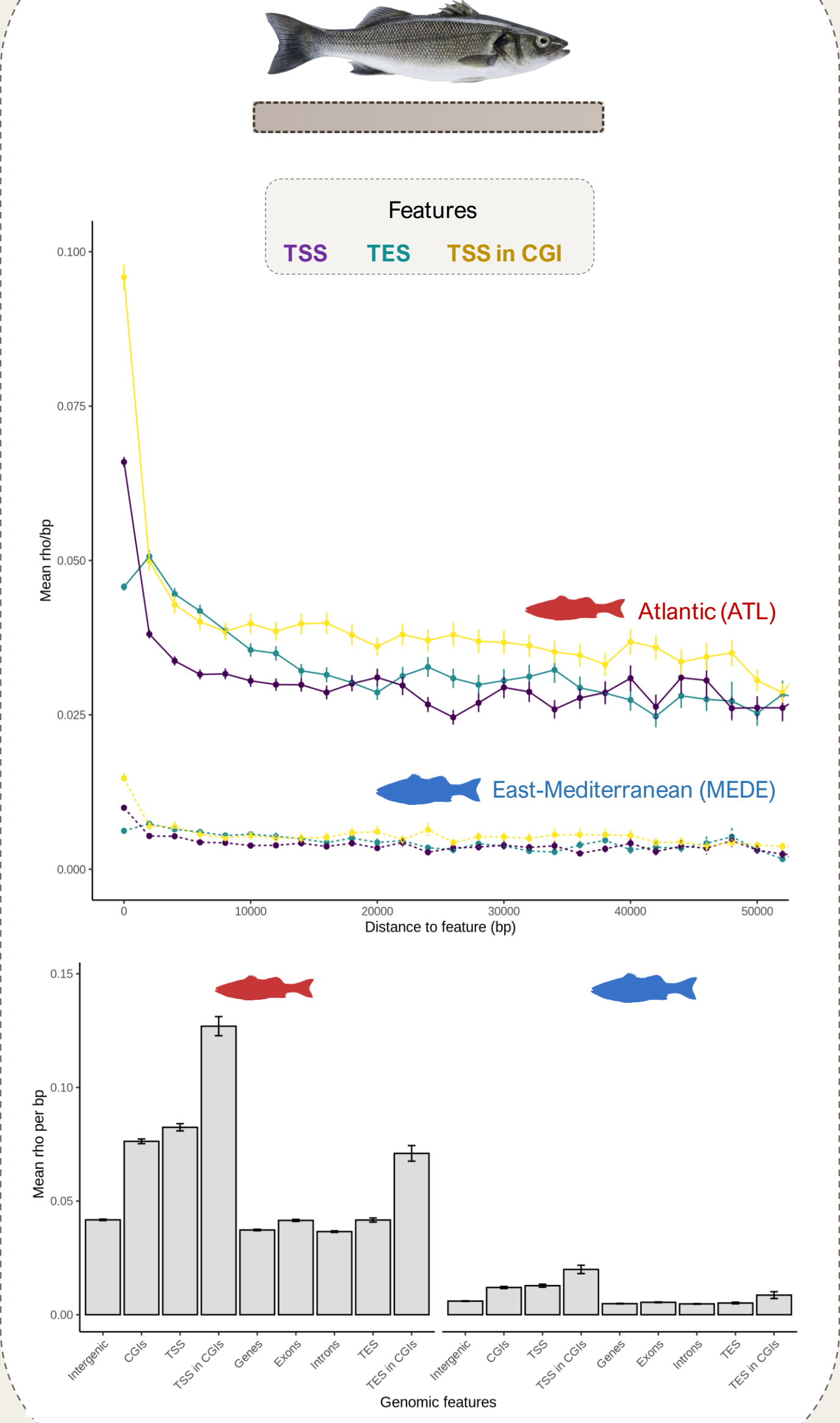
Fine scale recombination landscapes. Recombination rates  $\rho$  are very heterogeneous along chromosomes (~90% of the recombination in 20% of the genome) and concentrate into hotspots.

## Presence / Absence of PRDM9 footprints revealed by recombination rates at promoter-like features

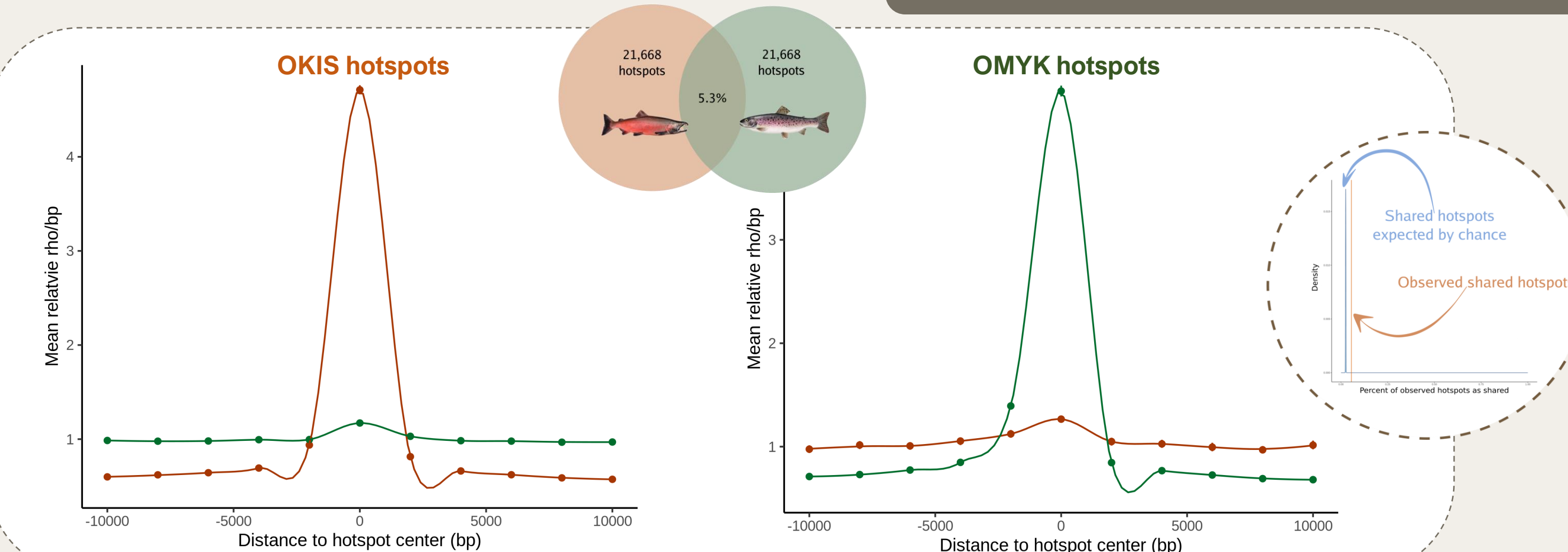


Limited enrichment of recombination rates at promoter-like features in Salmons, compared to the non-PRDM9 teleost *D. labrax*. Recombination does not increase at transcription start and end sites (TSS/TES) and in TSS in CpG islands (CGI), as in human and mice (on the left). On the contrary, the alternative mechanism directing recombination sites in species lacking functional PRDM9 such as *D. labrax* (on the right, two populations) targets these regions (as in birds, dogs, swordtail fish).

## Sea bass (*Dicentrarchus labrax*)

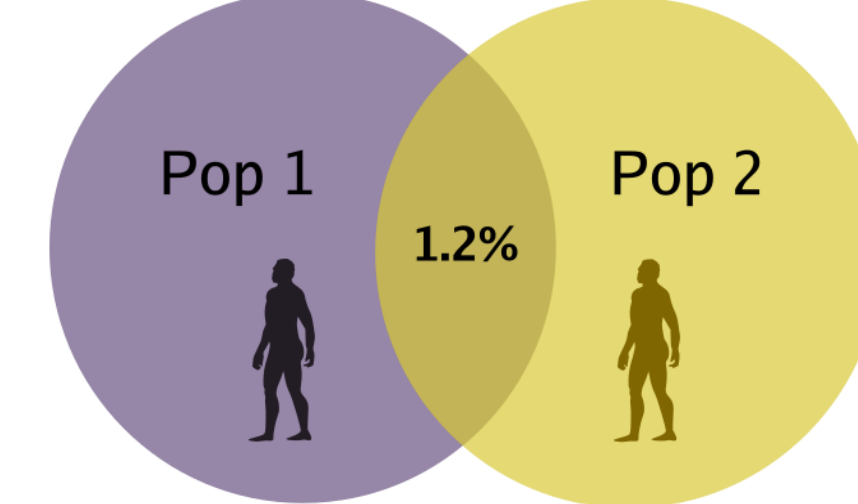


## Rapid evolution of hotspot location

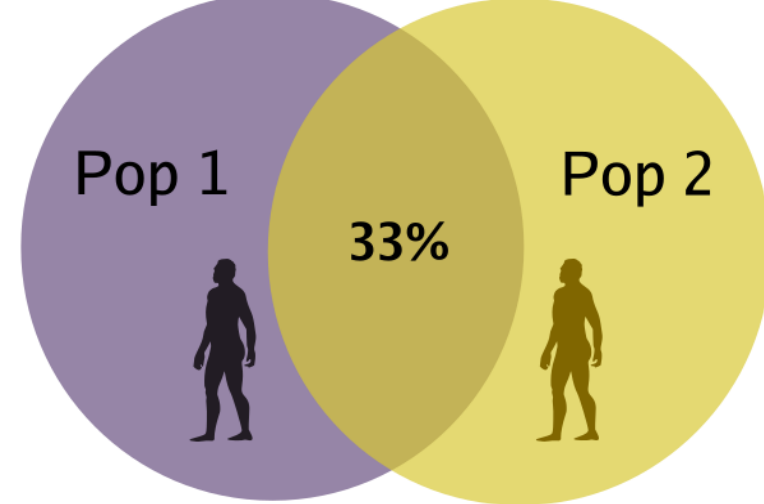


Fast evolving recombination landscapes between *O. kisutch* and *O. mykiss*. Less than 6% of hotspots are shared between the two species, suggesting fast evolving recombination landscapes.

## Different recombination landscapes



## Identical recombination landscapes



Hotspots comparison: how to distinguish between different and identical landscapes? Using simulations to assess the performance of LDhelmet, we found that, depending on the effective population size ( $N_e$ ) and the sample size, ~1-2% hotspots are found shared between two populations with different recombination landscapes, and ~30% between populations sharing the same landscape. The real fraction of shared hotspots is often underestimated due to false positives and false negatives (Raynaud et al., 2022).

## Conclusion

- The low enrichment of recombination events at promoter-like features and the lack of sharing of hotspots between the coho salmon and the rainbow trout are signatures of a PRDM9-dependant system and differ from the patterns in non-PRDM9 organisms (e.g. sea bass).
- The role of PRDM9 in the regulation of recombination in these two *Oncorhynchus* species needs to be validated by analyzing enrichment of PRDM9 binding sites, or by the mapping of double-strand breaks to identify empirically the location of recombination events.
- These PRDM9-like footprints must be determined in other taxa in order to assess its role across Salmonids (work in progress on the Atlantic salmon *Salmo salar*) and more broadly in animals.

## Acknowledgments

We are grateful to Louis Bernatchez, Ben Coop and Quentin Rougemont for providing us the coho salmon resequencing data and genotype information, and to Maud Duranton for the linkage disequilibrium maps of the two populations of sea bass. We also thank Julien Joseph, Laurent Duret, Nicolas Lartillot, Frédéric Baudat and Bernard de Massy for the discussions and feedback.

## References

- Auton et al. (2012). « A Fine-Scale Chimpanzee Genetic Map from Population Sequencing ». *Science* 336 (6078): 193-98.
- Auton et al. (2013). « Genetic Recombination Is Targeted towards Gene Promoter Regions in Dogs ». Édité par Ian Henderson. *PLoS Genetics* 9 (12): e1003984.
- Baker et al. (2017). « Repeated Losses of PRDM9-Directed Recombination despite the Conservation of PRDM9 across Vertebrates ». *ELife* 6.
- Myers et al. (2010). « Drive Against Hotspot Motifs in Primates Implicates the PRDM9 Gene in Meiotic Recombination ». *Science* 327 (5967): 876-79.
- Singhal et al. (2015). « Stable Recombination Hotspots in Birds ». *Science* 350 (6263): 928-32.
- Gao, G., Nome, T., Pearse, D. E., Moen, T., Naish, K. A., Thorgaard, G. H., ... & Palti, Y. (2018). A new single nucleotide polymorphism database for rainbow trout generated through whole genome resequencing. *Frontiers in genetics*, 9, 147.
- Rougemont, Q., Moore, J. S., Leroy, T., Normandeau, E., Rondeau, E. B., Withler, R. E., ... & Bernatchez, L. (2020). Demographic history shaped geographical patterns of deleterious mutation load in a broadly distributed Pacific Salmon. *PLoS genetics*, 16(8), e1008348.
- Raynaud, M., Gagnaire, P. A., & Galtier, N. (2022). Performance and limitations of linkage-disequilibrium-based methods for inferring the genomic landscape of recombination and detecting hotspots: a simulation study. *bioRxiv*.
- Duranton et al., (in prep).