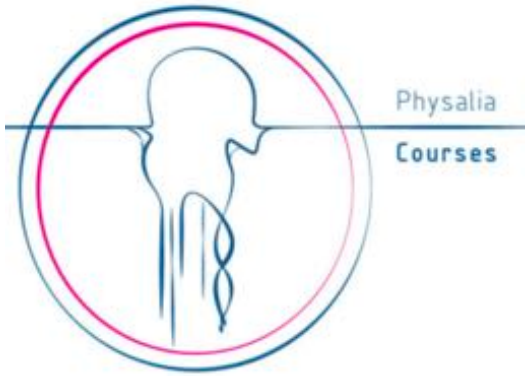
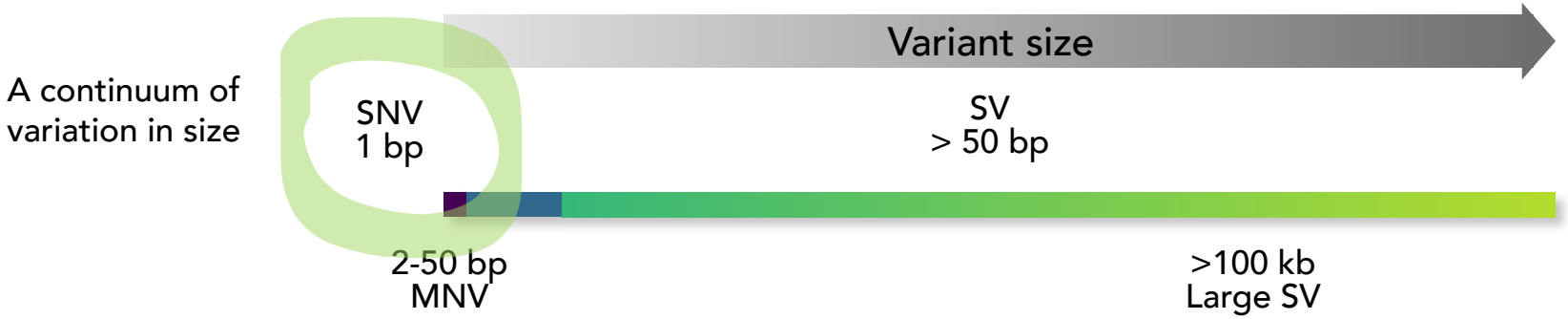


Structural Variants and adaptation

Day 4 - Lecture 1

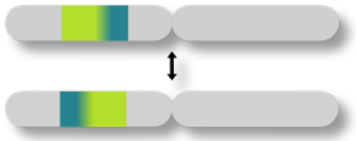


STRUCTURAL VARIANT
all changes in position or direction, as well as gains or losses of sequence

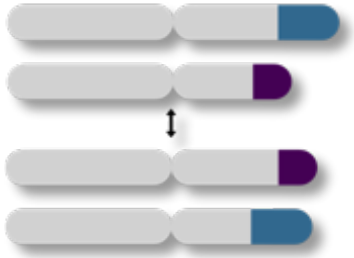
Merot, Oomen, Tigano, Wellenreuther 2020, TREE (in press)

Balanced SVs
(change in position or order)

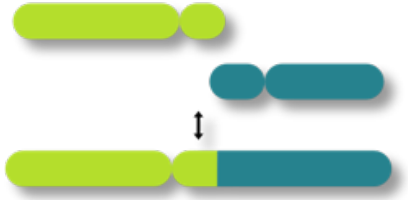
Inversion



Translocation



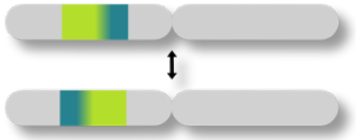
Fusion/fission



Merot, Oomen, Tigano, Wellenreuther 2020, TREE

Balanced SVs
(change in position or order)

Inversion



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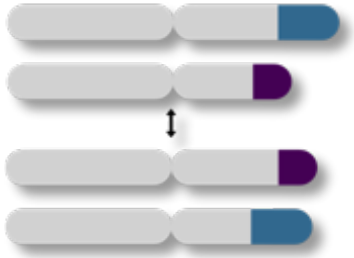

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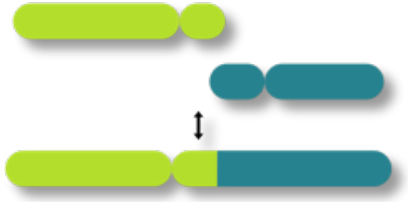
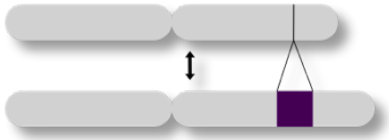
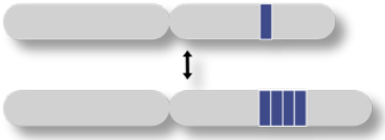


Unbalanced SVs
(gain/loss of DNA)

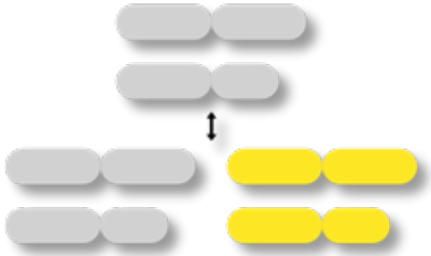
CNV:
Insertion/deletion



CNV:
duplications, tandem repeats



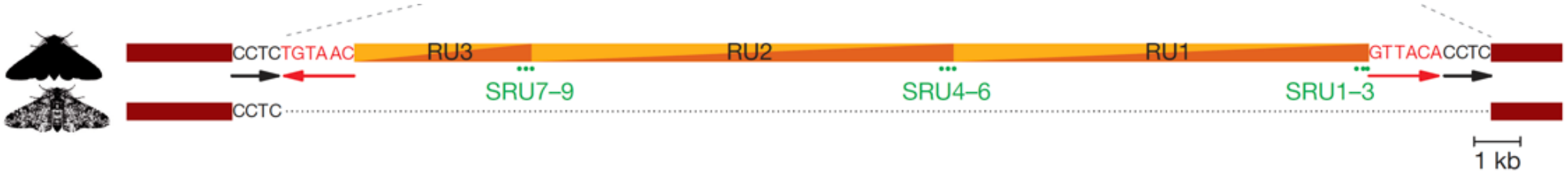
Polyploidy



Merot, Oomen, Tigano, Wellenreuther 2020, TREE

Why do we study them?

SVs are the genetic basis of several traits.

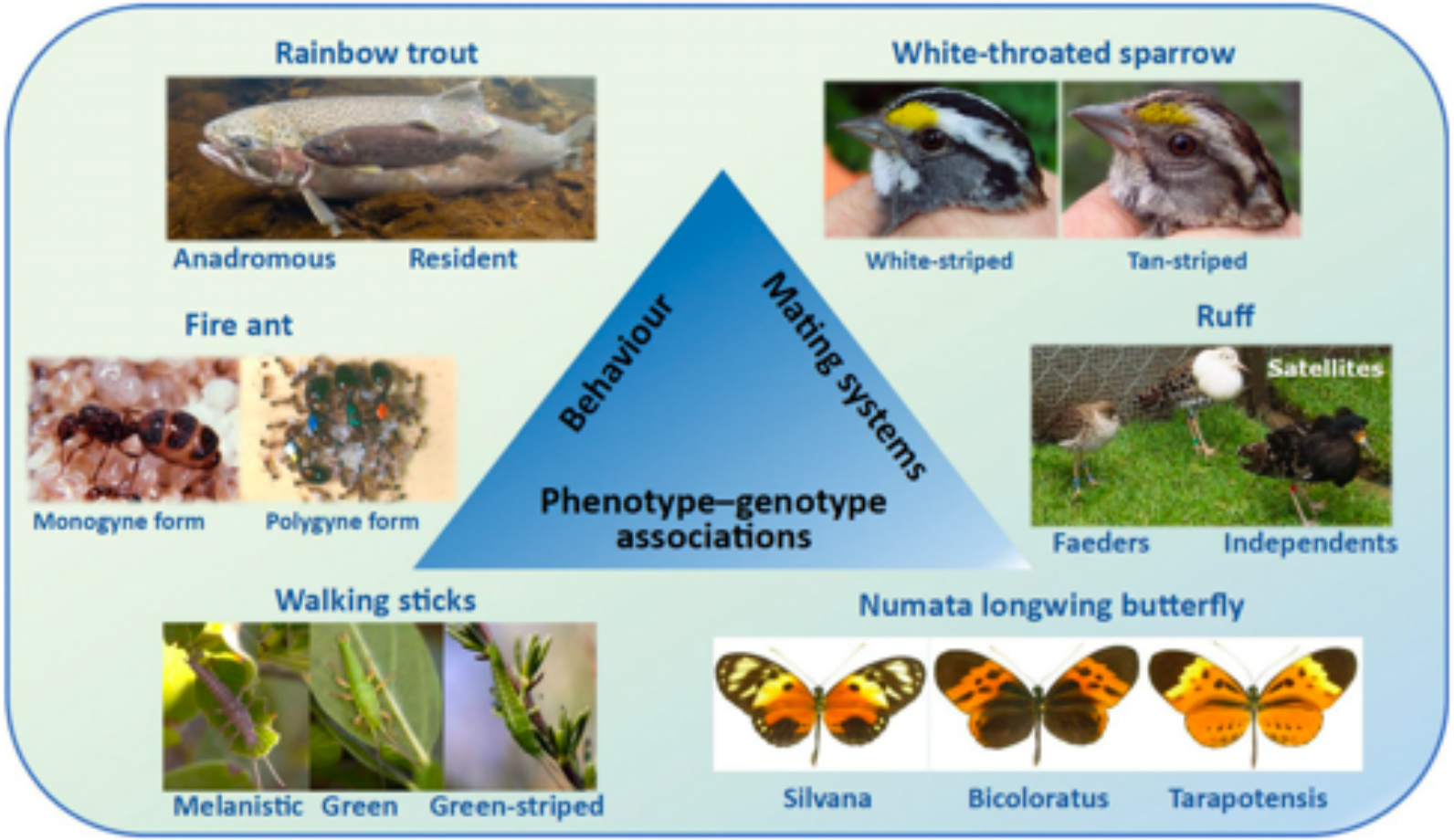


Van't Hof et al. 2016, Nature

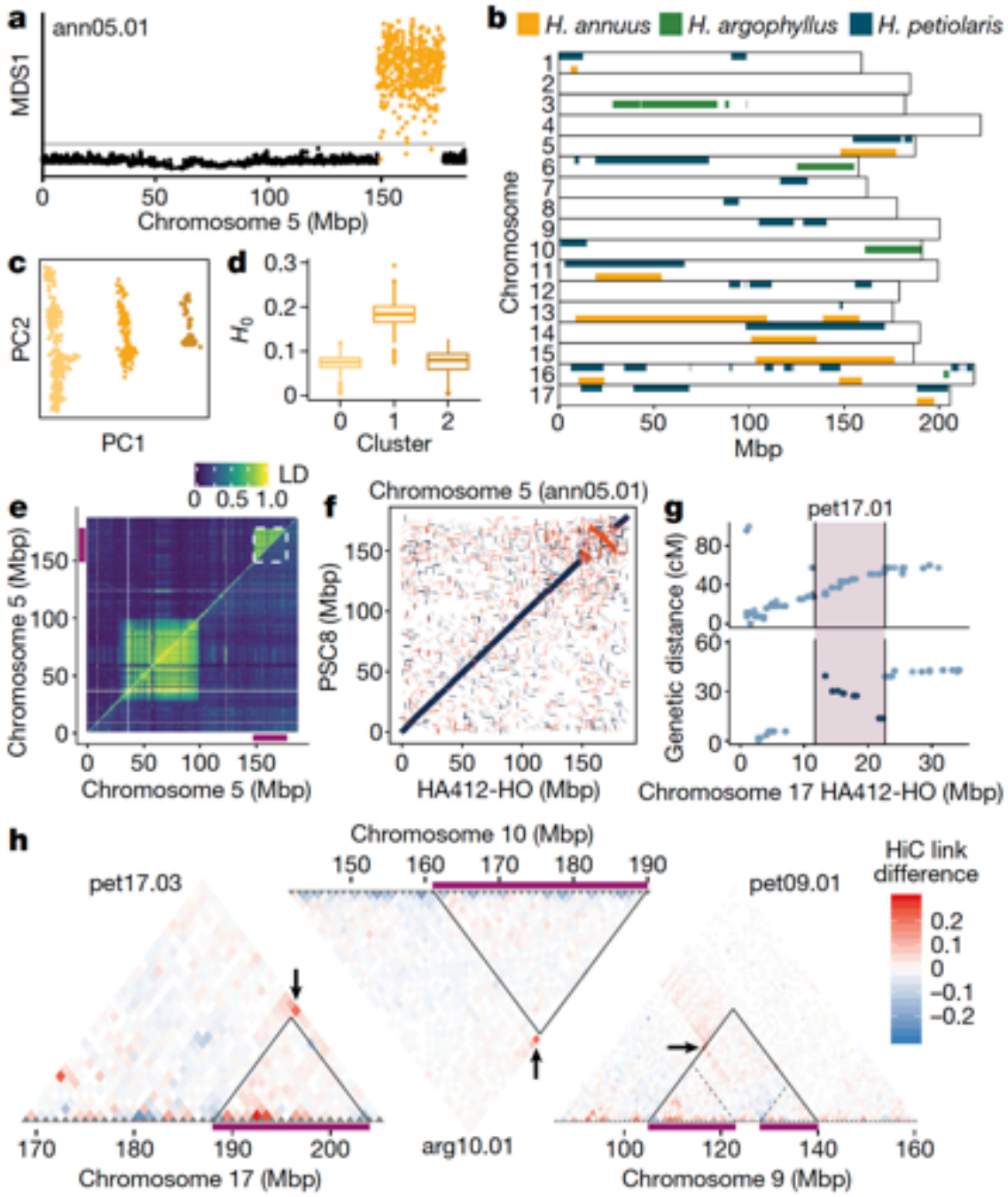
'Missing heritability problem'

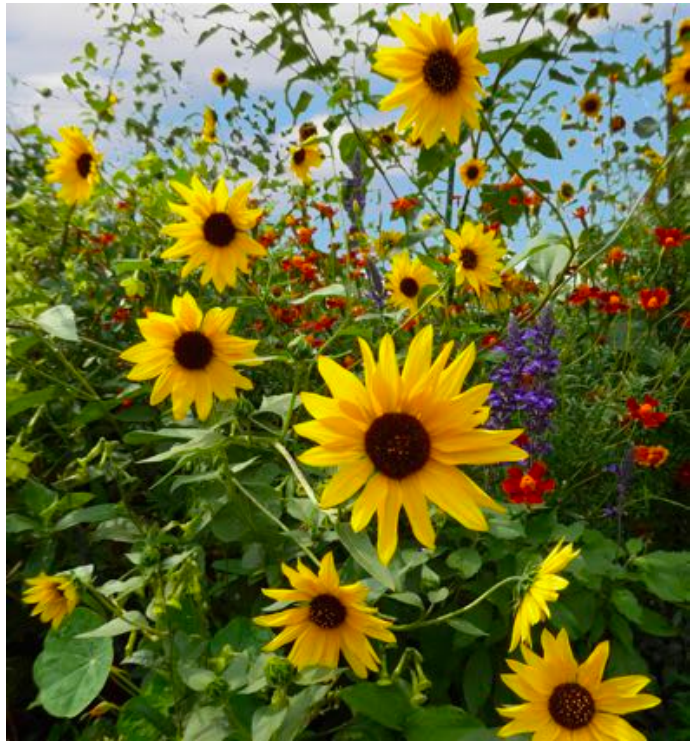
→When sequence variation explains only for a portion of the variation in a given trait (or disease)

Inversions are the SV most commonly associated with adaptive differences



Wellenreuther & Bernatchez 2018, TREE

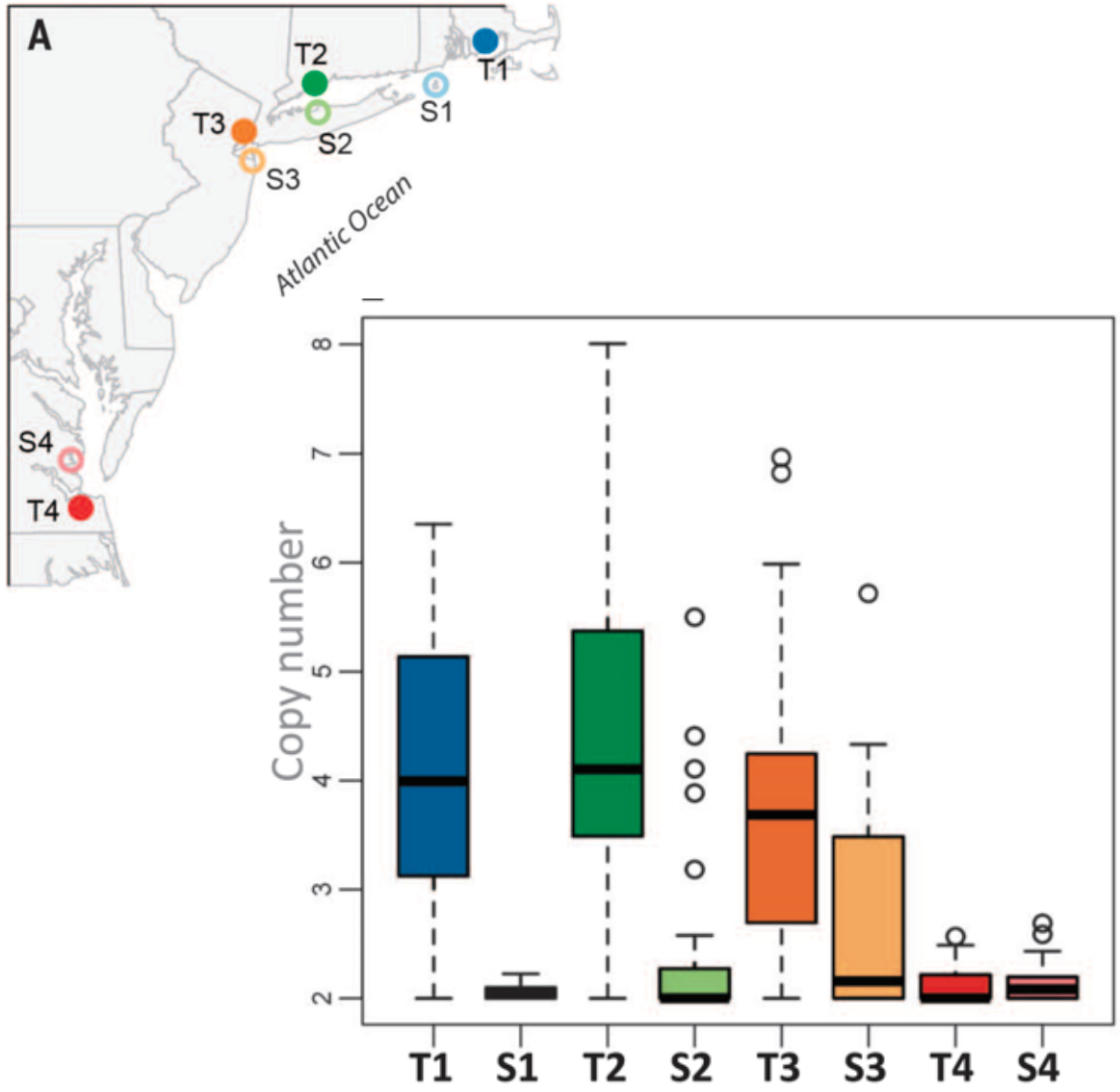


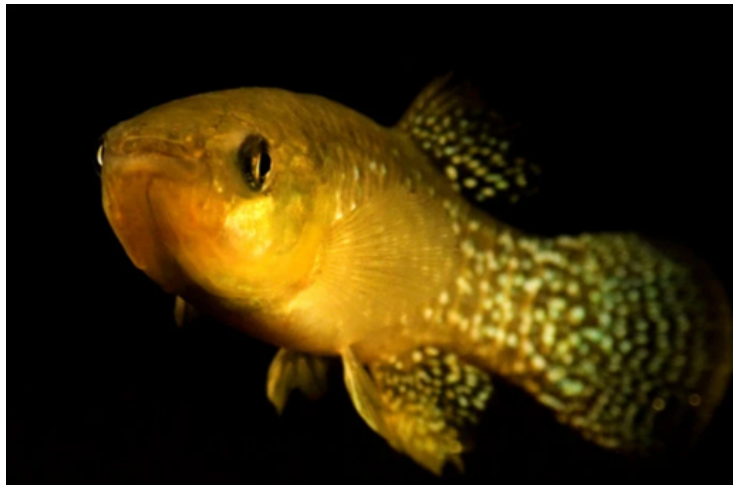


Sunflowers

SVs associated with many ecological relevant traits

Todesco et al. 2020, *Nature*





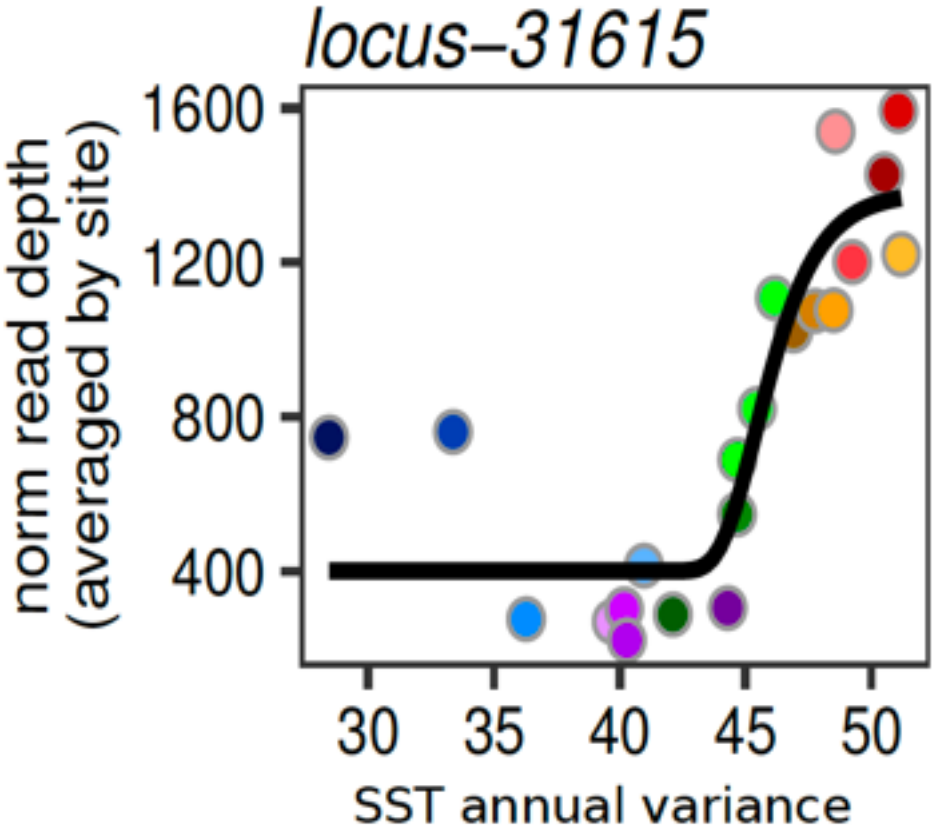
Atlantic killifish

Copy Number Variants (CNVs) associated with rapid adaptation to chemical pollution

Reid et al. 2016, Science

Lobster

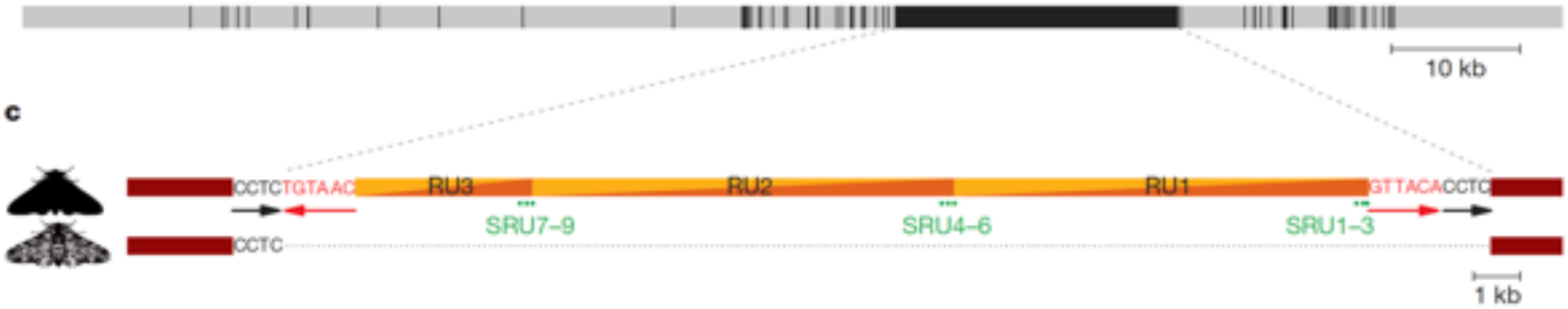
Variation in copy number associated with environmental variance



Dorant et al. 2020, Molecular Ecology

Peppered moth

Insertion of a transposon associated with melanism



Van't Hof et al. 2016, Nature

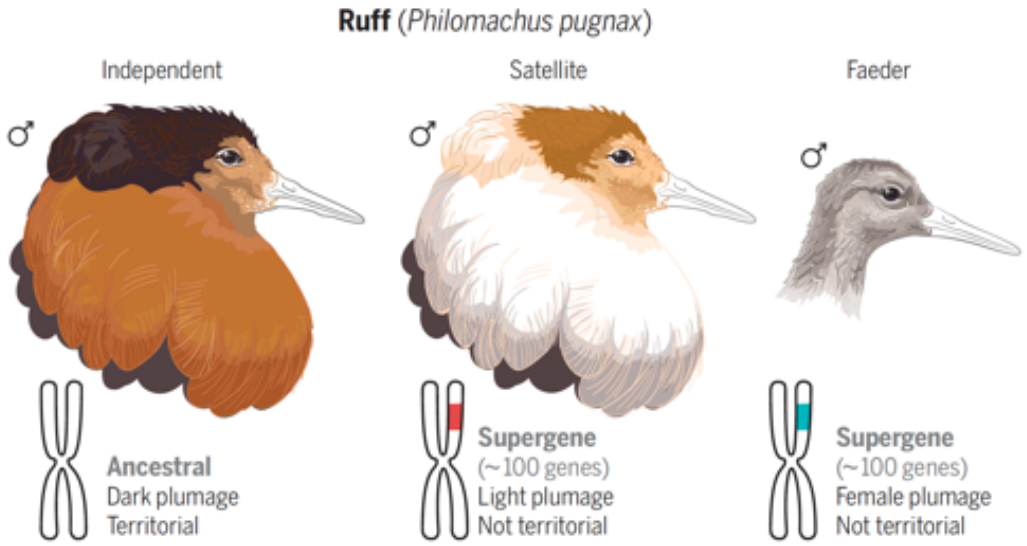
Type of variant	Number of variants	Base pairs affected	% of genome
SNPs	6,547,716	6,547,716	0.93
Indels	1,301,743	6,248,366	0.89
Deletions	17,599	4,666,669	0.66
Duplications	2,427	7,699,239	1.09
Inversions	359	249,945	0.04
Total SVs	1,330,589	18,864,219	2.68

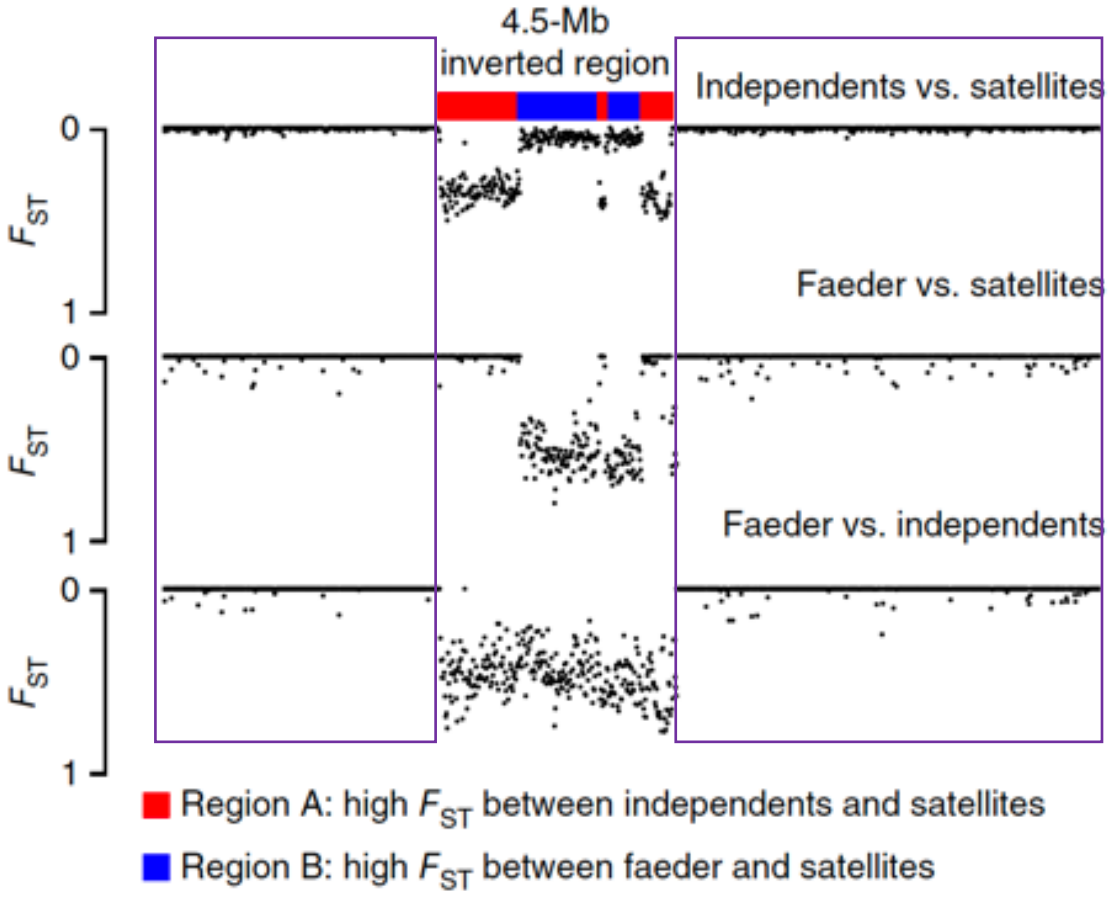
Australasian snapper

SVs cover a higher proportion of the genome than SNPs

Catanach et al. 2019, Molecular Ecology

How do SVs affect phenotypes?

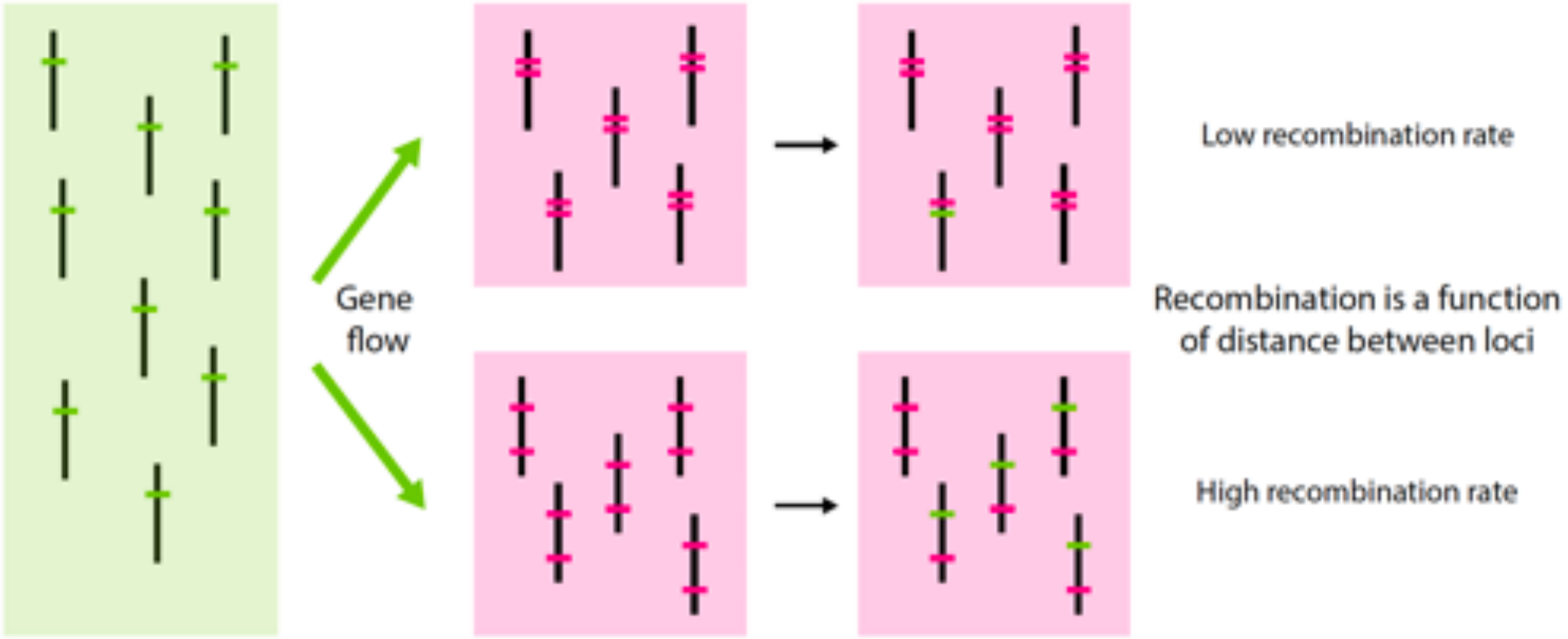




Lamichhaney et al. 2016, Nat. Gen.; Küpper et al. 2016, Nat. Gen; Taylor and Campagna 2016, Science

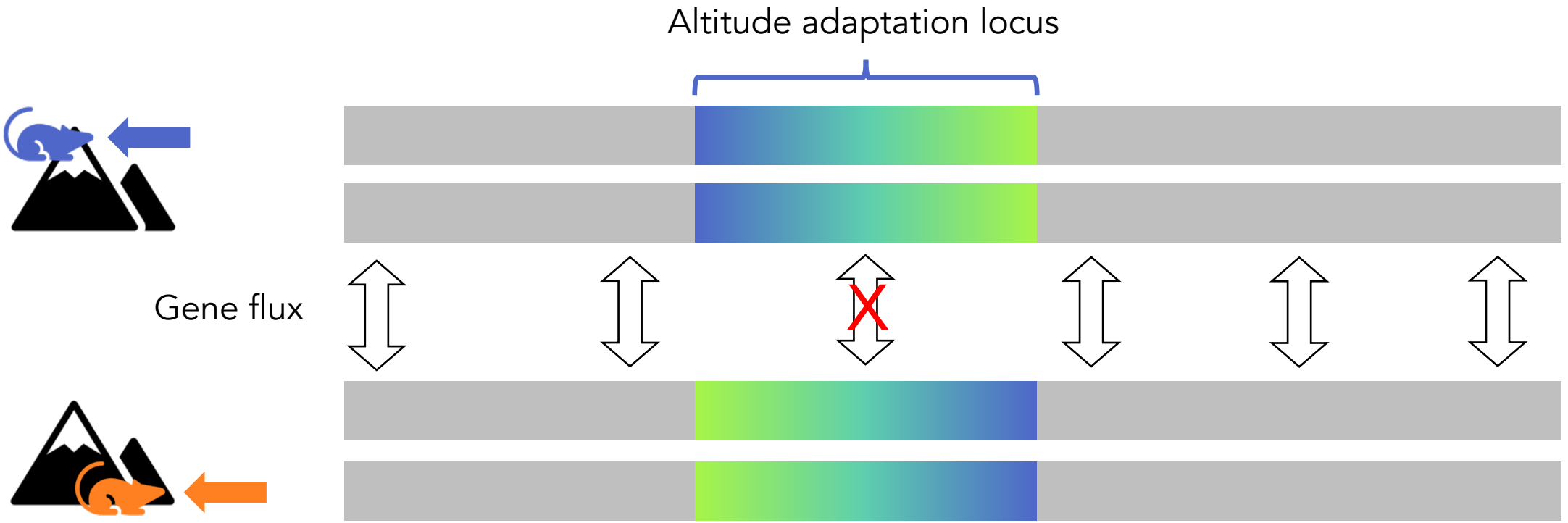
Structural variants that suppress recombination, like inversions, maintain complexes of co-adapted alleles in the face of gene flow.

Tight genetic architectures are more resistant to gene flow



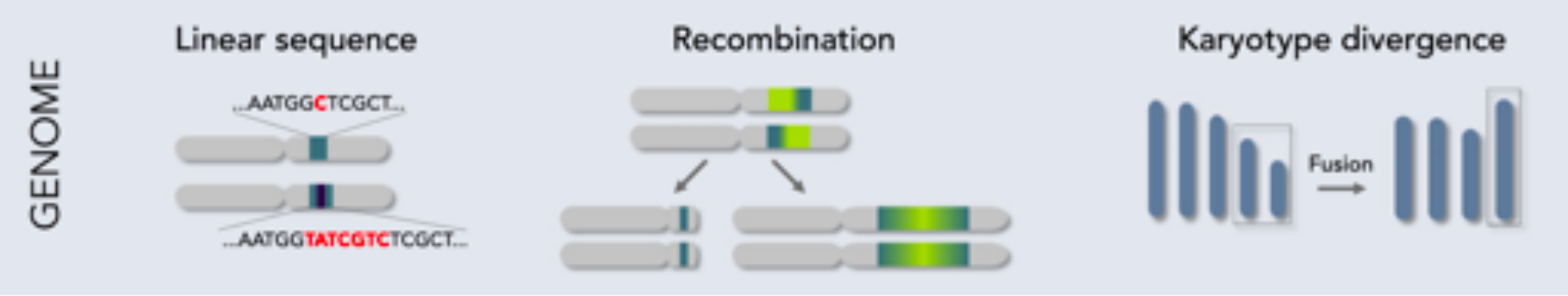
Tigano and Friesen 2016, Molecular Ecology; Yeaman 2013, PNAS

Recombination is further reduced or suppressed if those combinations of alleles are ‘protected’ by inversions or other SVs

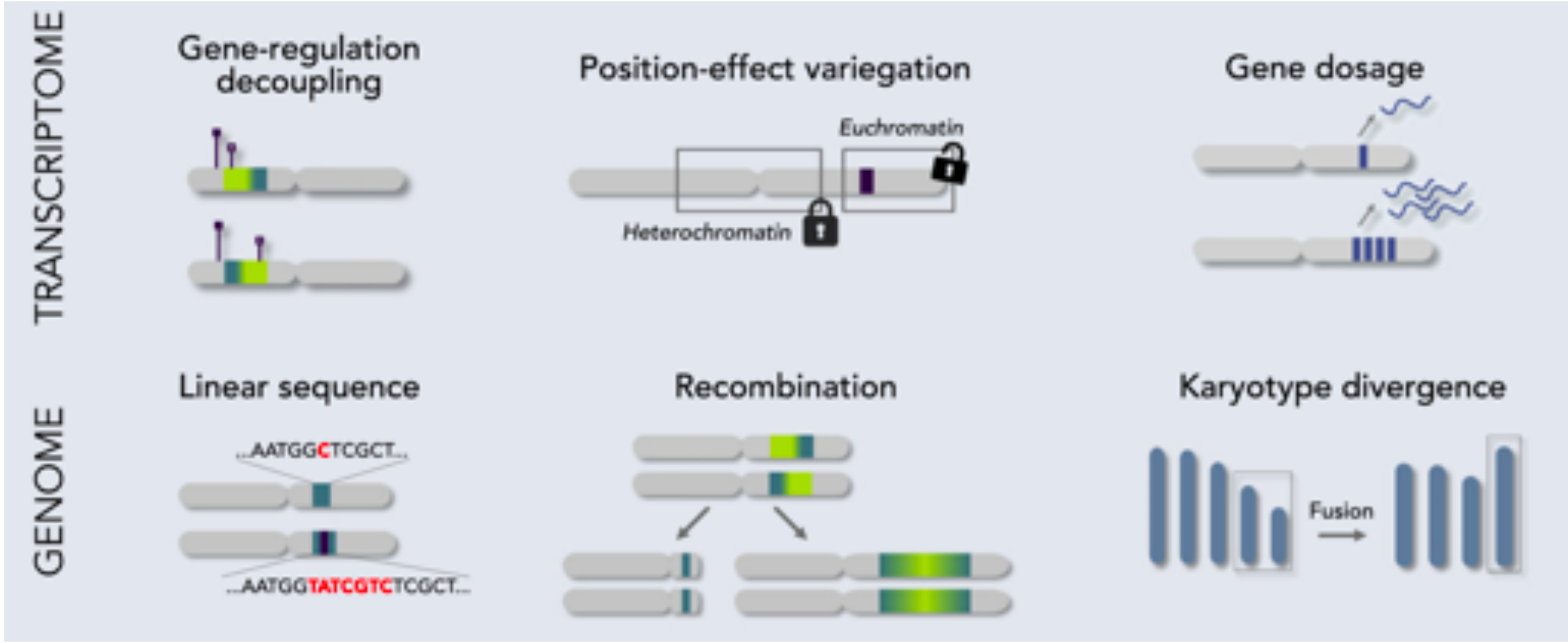


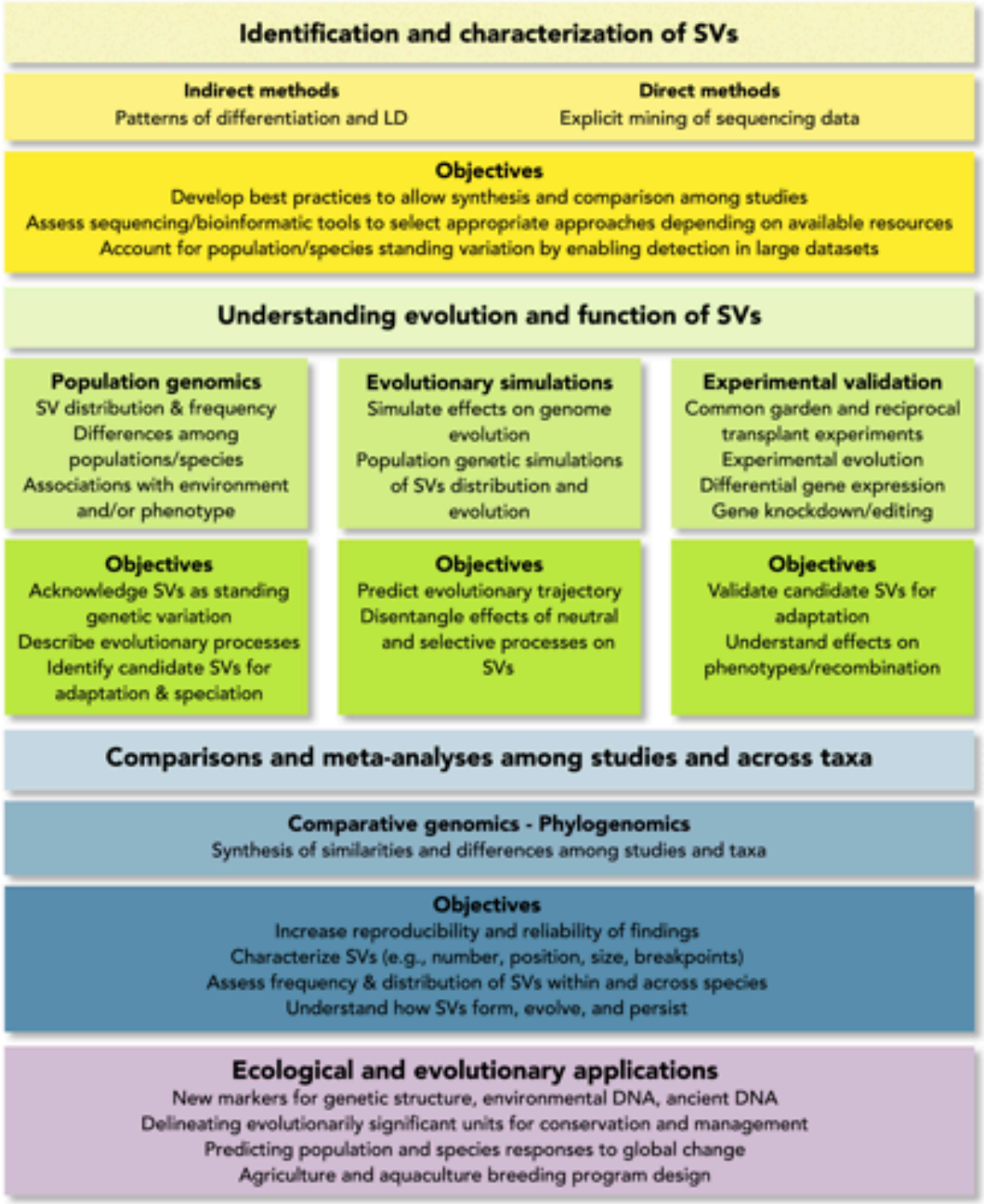
How do SVs affect phenotypes?

Recombination suppression is only one of many ways



How do SVs affect phenotypes?





We still know very little about SVs and lack of standardized protocols hamper progress.

We developed a roadmap to guide future studies focusing on the role of SVs in adaptation and speciation

Identification and characterization of SVs

Indirect methods

Patterns of differentiation and LD

Direct methods

Explicit mining of sequencing data

Objectives

Develop best practices to allow synthesis and comparison among studies

Assess sequencing/bioinformatic tools to select appropriate approaches depending on available resources

Account for population/species standing variation by enabling detection in large datasets

Understanding evolution and function of SVs

Population genomics

SV distribution & frequency
Differences among populations/species
Associations with environment and/or phenotype

Objectives

Acknowledge SVs as standing genetic variation
Describe evolutionary processes
Identify candidate SVs for adaptation & speciation

Evolutionary simulations

Simulate effects on genome evolution
Population genetic simulations of SVs distribution and evolution

Objectives

Predict evolutionary trajectory
Disentangle effects of neutral and selective processes on SVs

Experimental validation

Common garden and reciprocal transplant experiments
Experimental evolution
Differential gene expression
Gene knockdown/editing

Objectives

Validate candidate SVs for adaptation
Understand effects on phenotypes/recombination

Comparisons and meta-analyses among studies and across taxa

Comparative genomics - Phylogenomics

Synthesis of similarities and differences among studies and taxa

Objectives

Increase reproducibility and reliability of findings

Characterize SVs (e.g., number, position, size, breakpoints)

Assess frequency & distribution of SVs within and across species

Understand how SVs form, evolve, and persist

Ecological and evolutionary applications

New markers for genetic structure, environmental DNA, ancient DNA

Delineating evolutionarily significant units for conservation and management

Predicting population and species responses to global change

Agriculture and aquaculture breeding program design

The end!