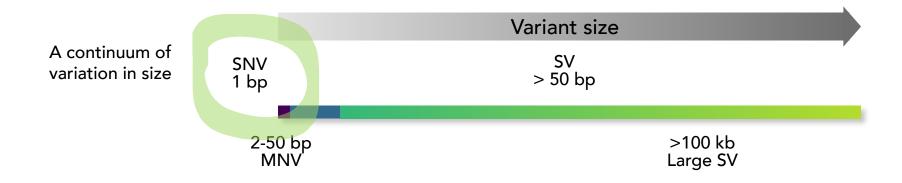


# Structural Variants and adaptation

Day 4 - Lecture 1



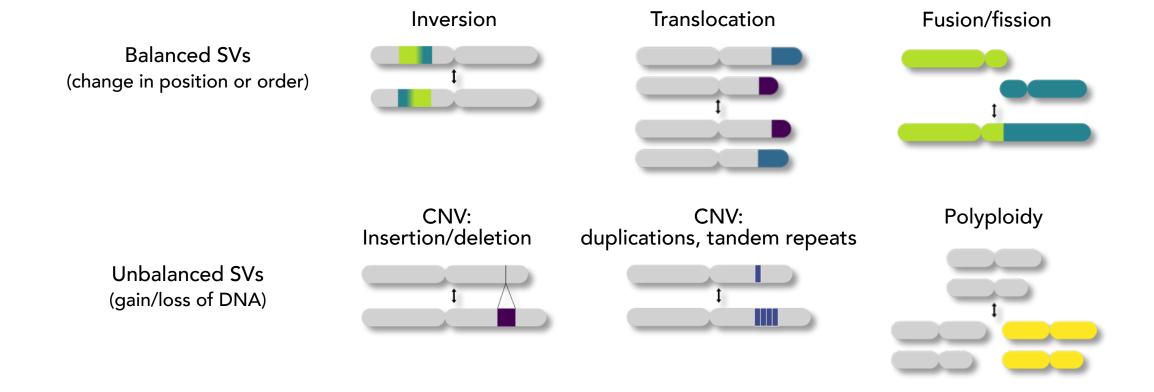
# STRUCTURAL VARIANT all changes in position or direction, as well as gains or losses of sequence

Balanced SVs (change in position or order)

Inversion

Translocation

Fusion/fission



# Why do we study them?

SVs are the genetic basis of several traits.

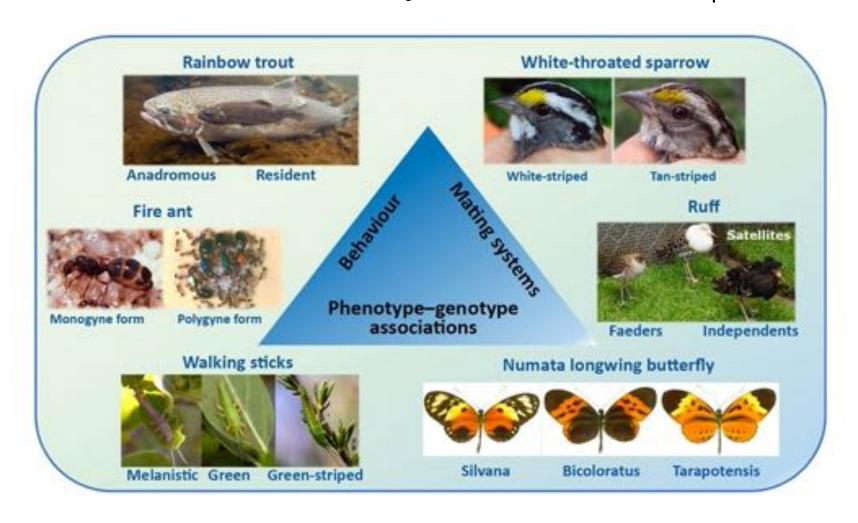


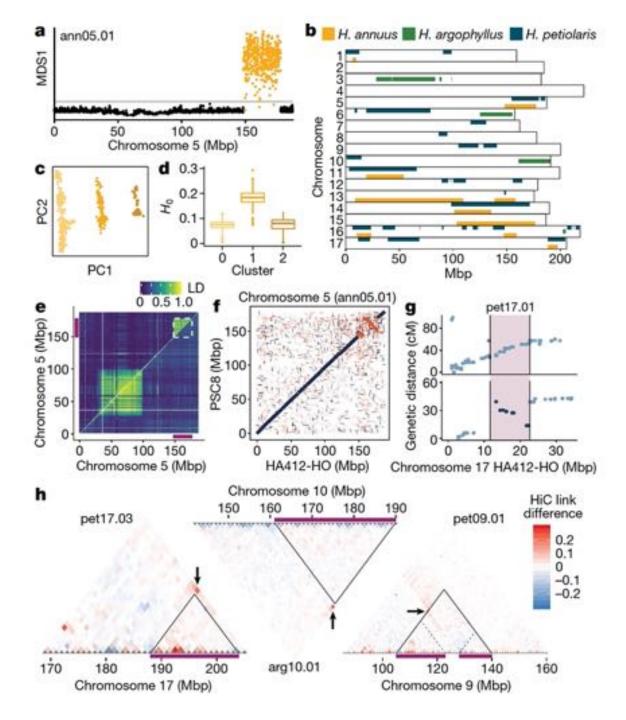
Van't Hof et al. 2016, Nature

# '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

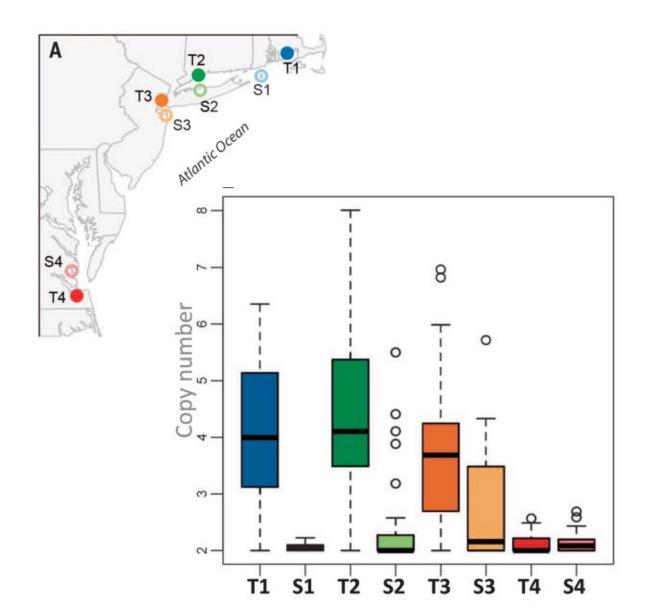






Sunflowers

SVs associated with many ecological relevant traits





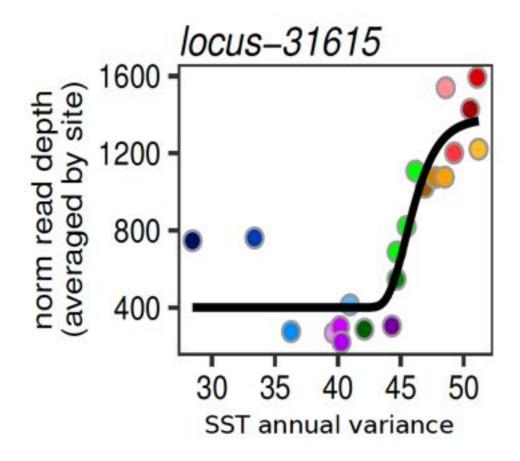
Atlantic killifish

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

Reid et al. 2016, Science

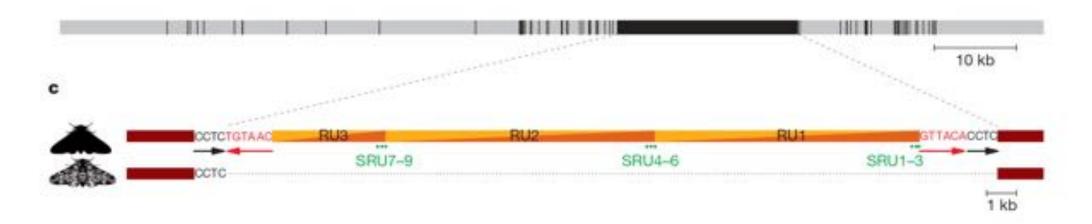


Lobster
Variation in copy number associated with environmental variance



# Peppered moth

Insertion of a transposon associated with melanism

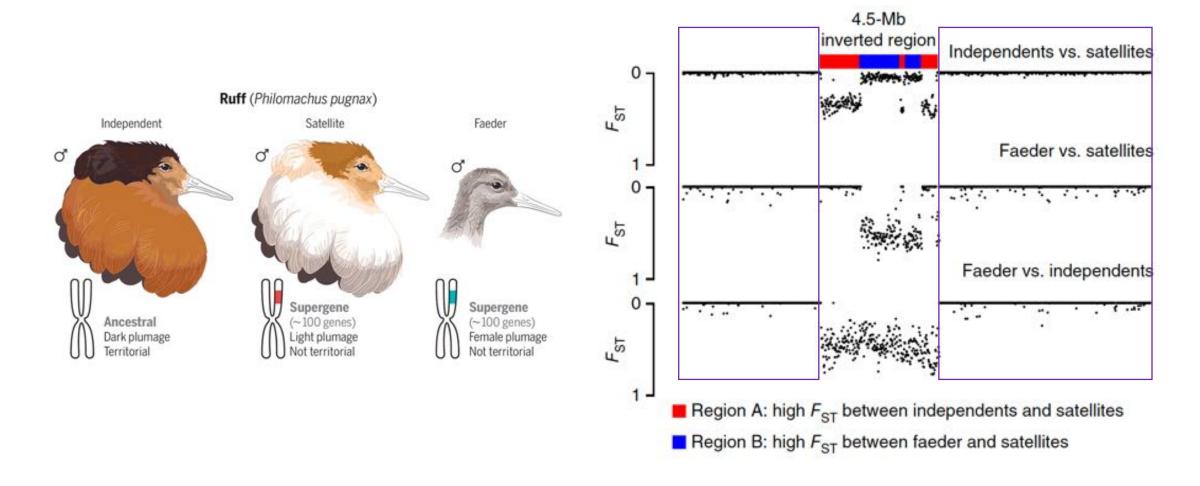


Type of variant	Number of variants	Base pairs affected	% of genome
SNPs	6,547,716	6,547,716	0.93
Indels	1,301,743	6,248,366	0.89
Deletions	17,599	4,666,669	0.66
Duplications	2,427	7,699,239	1.09
Inversions	359	249,945	0.04
Total SVs	1,330,589	18,864,219	2.68



Australasian snapper
SVs cover a higher proportion of the genome than SNPs

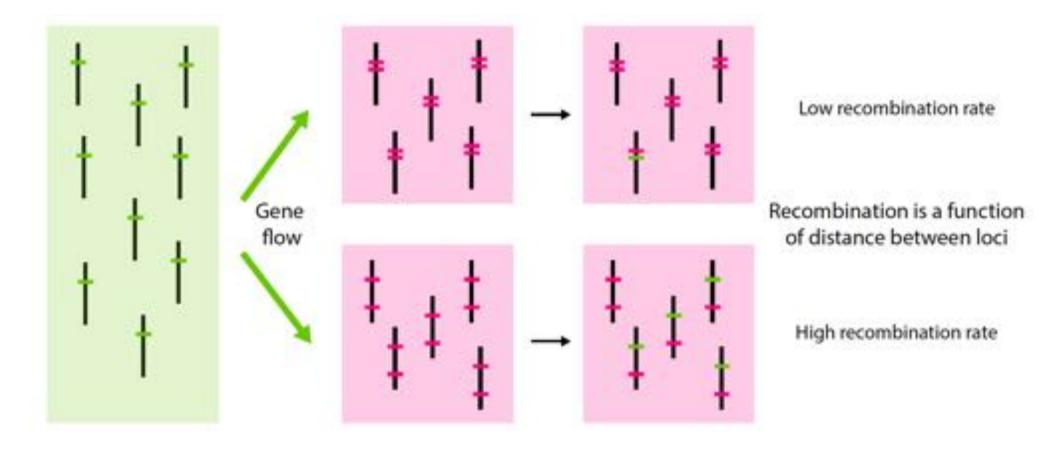
# How do SVs affect phenotypes?



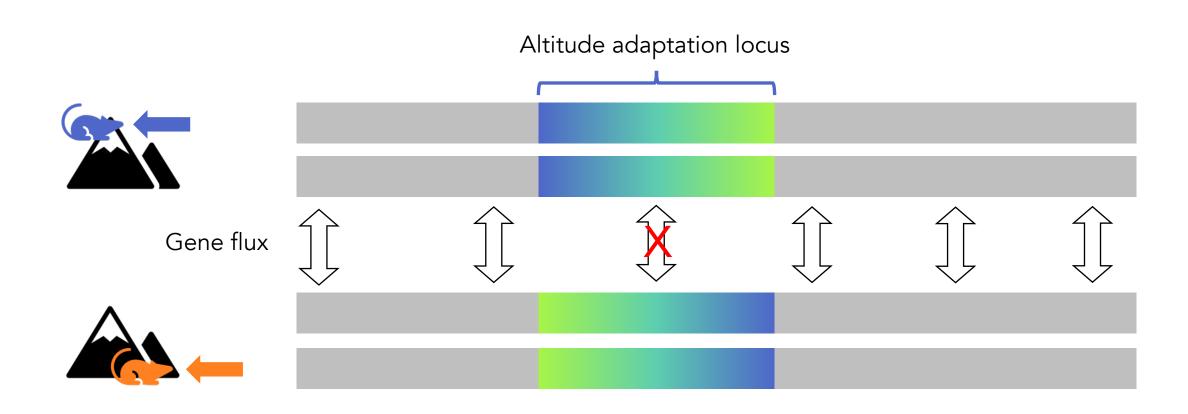
Lamichhaney et al. 2016, Nat. Gen.; Küpper et al. 2016, Nat. Gen; Taylor and Campagna 2016, Science

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

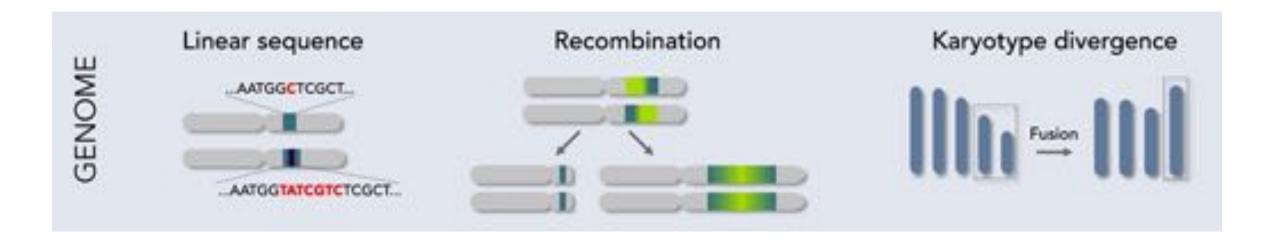


Recombination is further reduced or suppressed if those combinations of alleles are 'protected' by inversions or other SVs

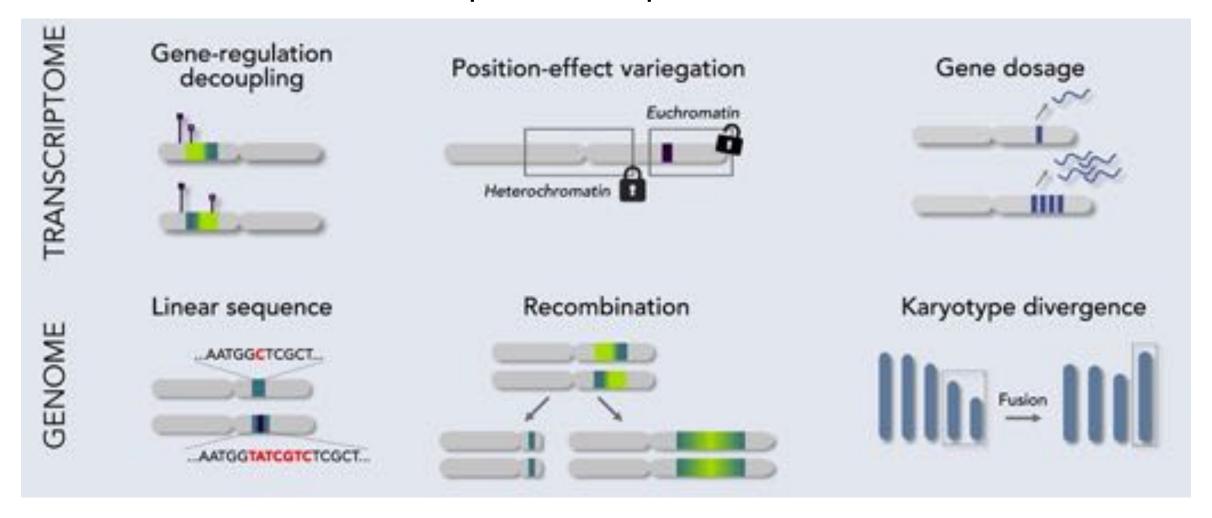


# How do SVs affect phenotypes?

Recombination suppression is only one of many ways



# How do SVs affect phenotypes?



#### 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

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

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Patterns of differentiation and LD

#### Direct methods

Explicit mining of sequencing data

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# The end!