

What genome sequences can tell us about evolution

Hannes Svardal

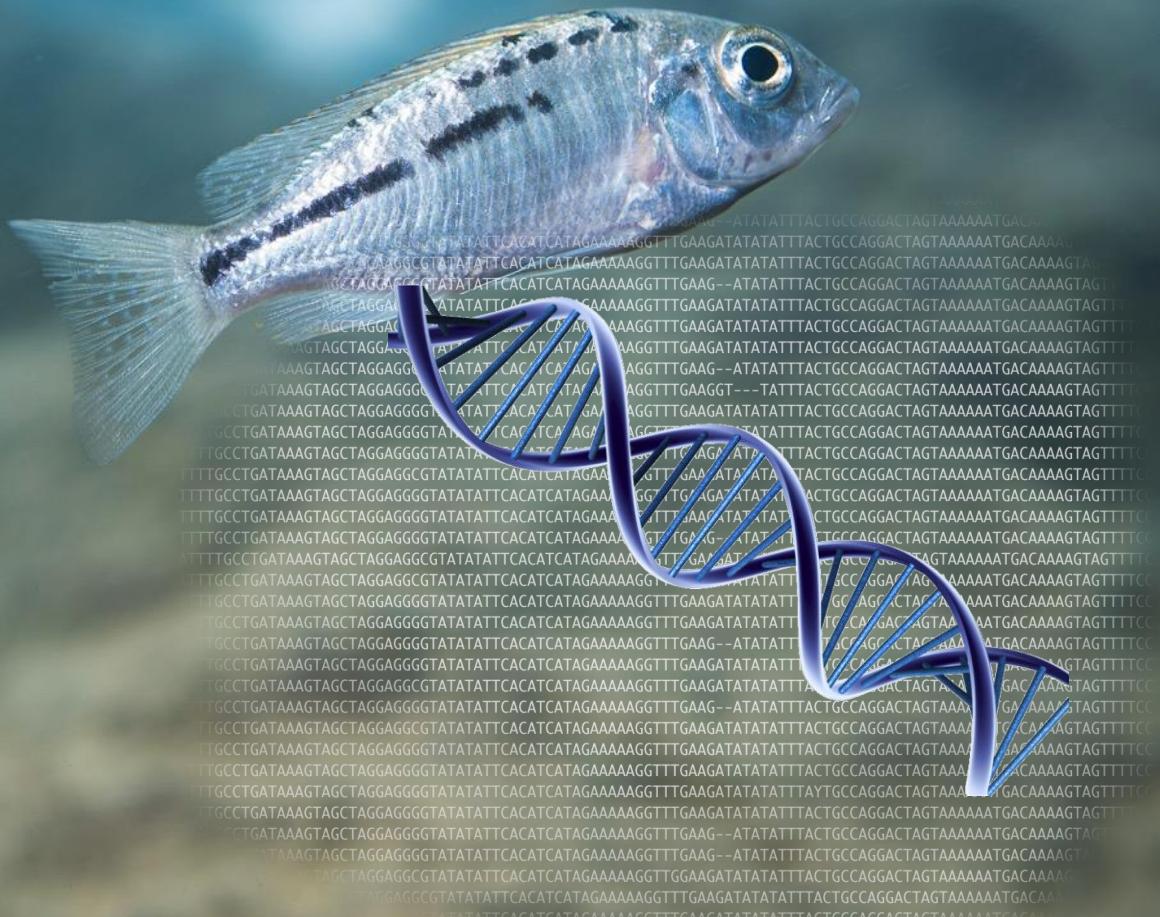
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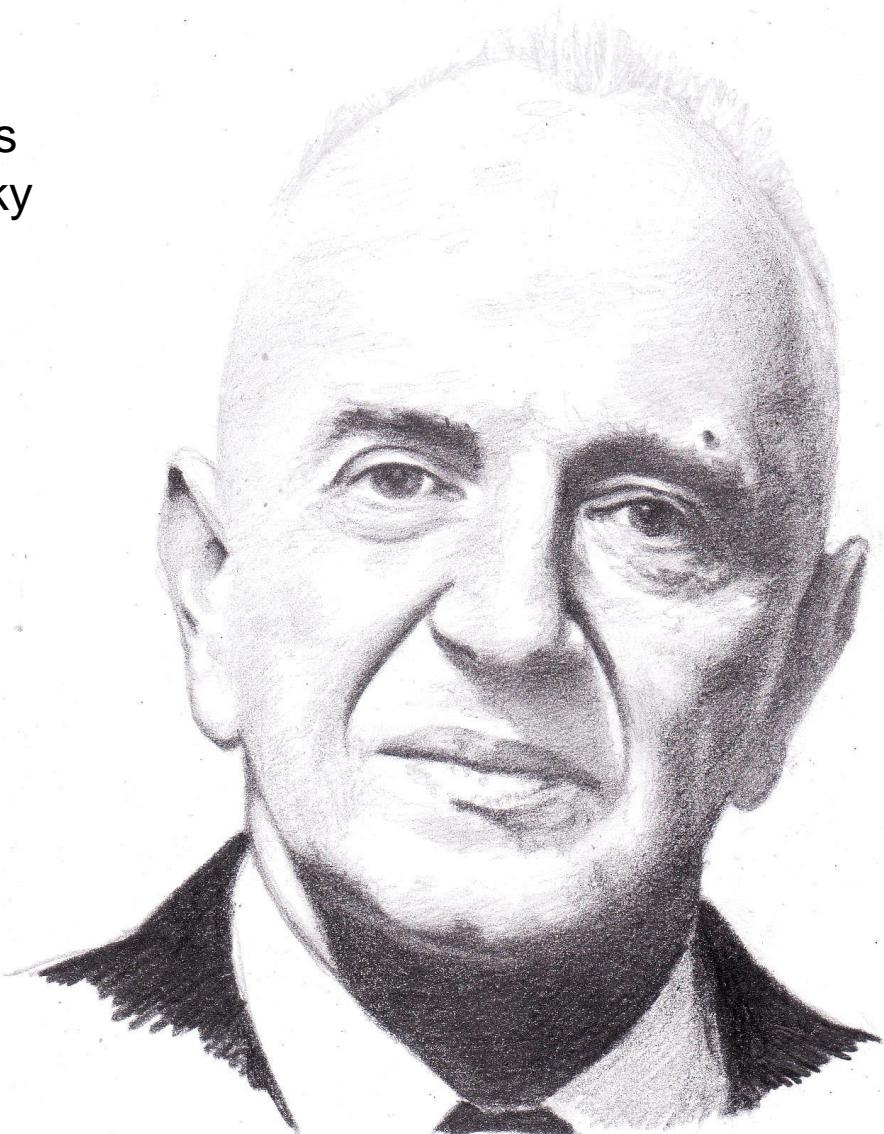
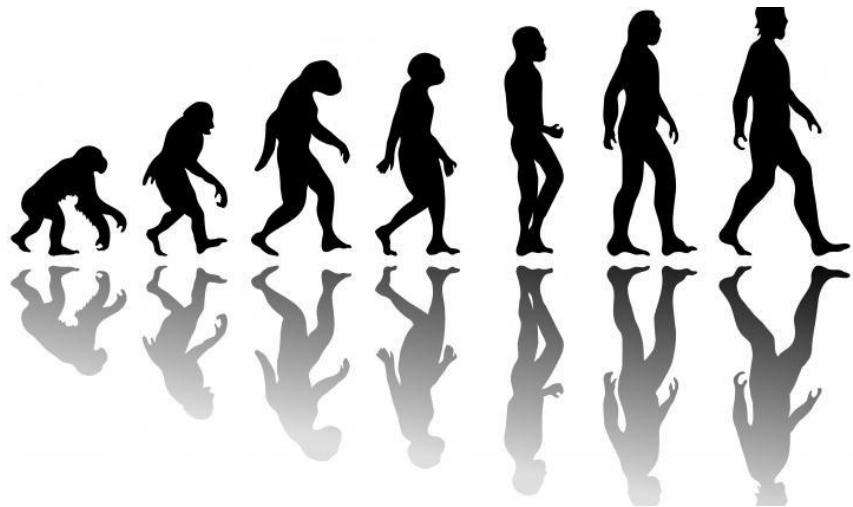


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Nothing in biology makes sense except in the light of evolution.

Theodosius
Dobzhansky



Genomes

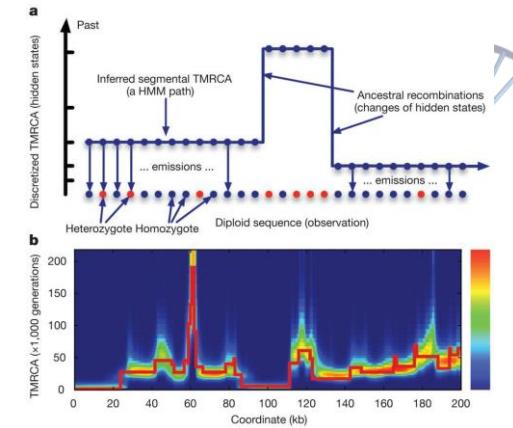
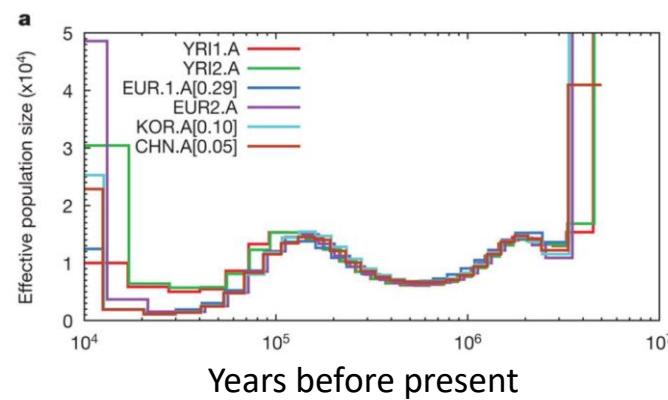
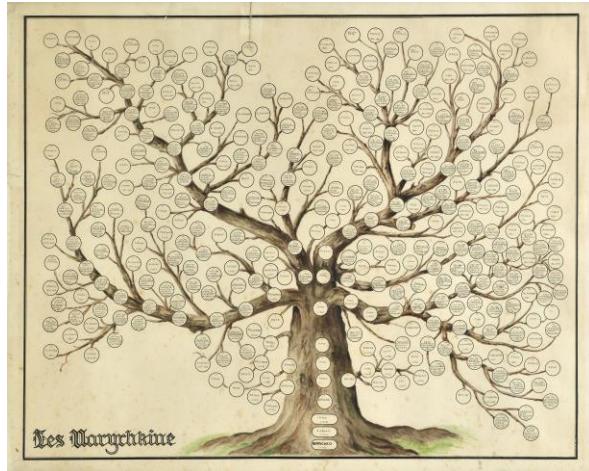


- encode for phenotypes
- are inherited (+ rare mutation)
- are the substrate for the evolutionary process
- provide a record of past evolutionary events

Use genomic information to

- understand the evolutionary history of a population
- understand how genotypes produce phenotypes

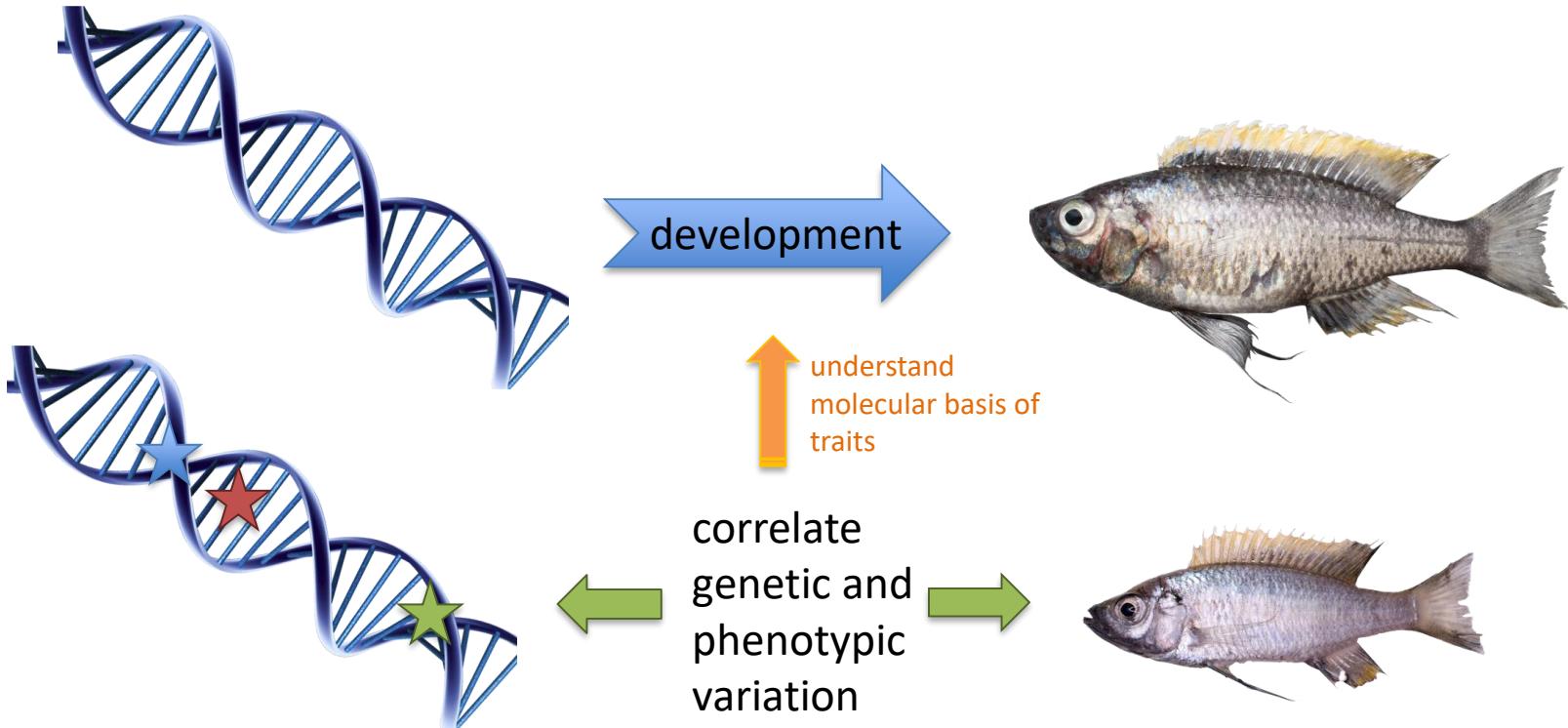
Genetic variants are the fundamental unit of evolution



Li and Durbin 2011

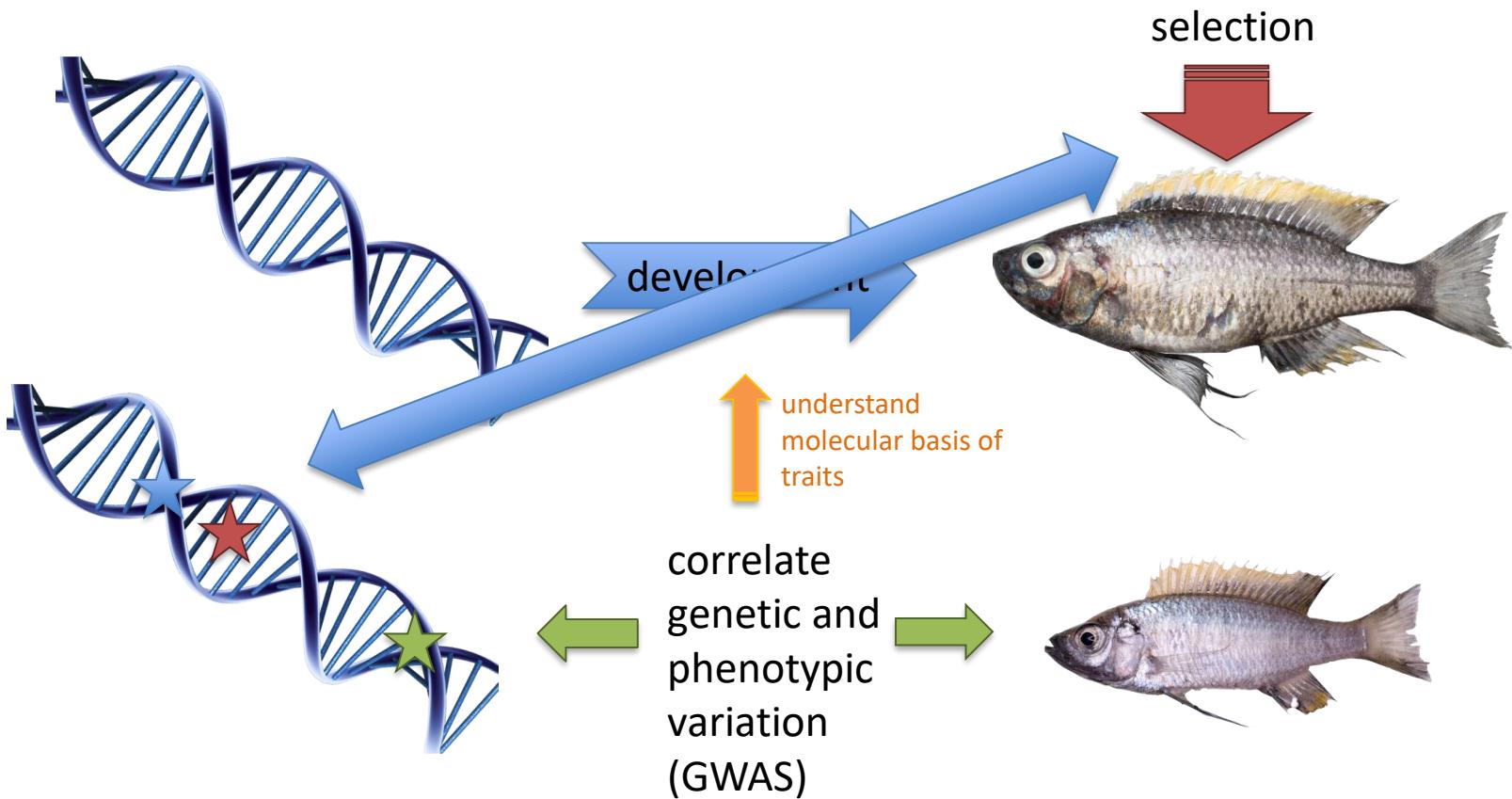
- Most genetic variants are neutral
- Allele frequencies change = random walk
- Genetic variation data allows us to infer
 - relatedness patterns between individuals
 - demographic history of populations
 - past migration and hybridisation events

Genetic variants are the fundamental unit of evolution



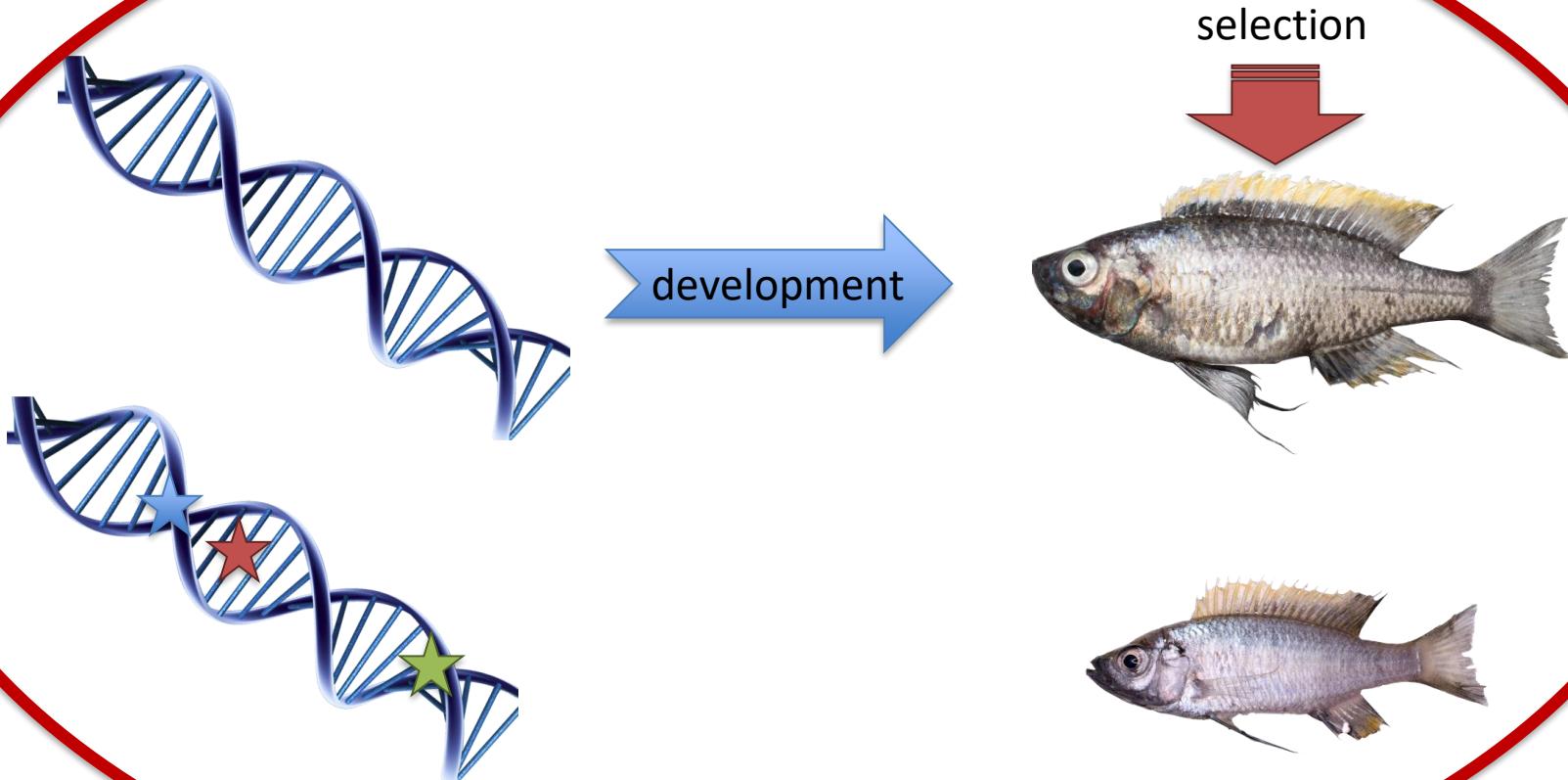
- Genome-wide association (GWA) or quantitative trait locus (QTL) mapping allows us to link genetic variants to phenotypic variation
→ Understanding which genes contribute to traits

Genetic variants are the fundamental unit of evolution



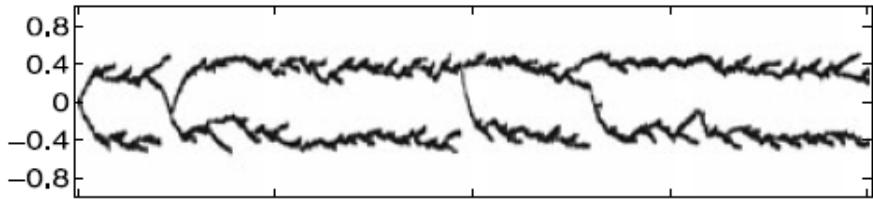
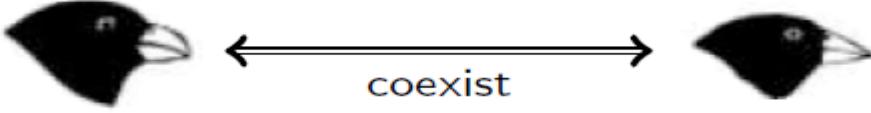
- Alternatively, we can ignore phenotypes and look for signatures of selection in genomic data
- Selection scans use population differentiation or within-population diversity patterns to scan the genome for loci under selection

Genetic variants are the fundamental unit of evolution

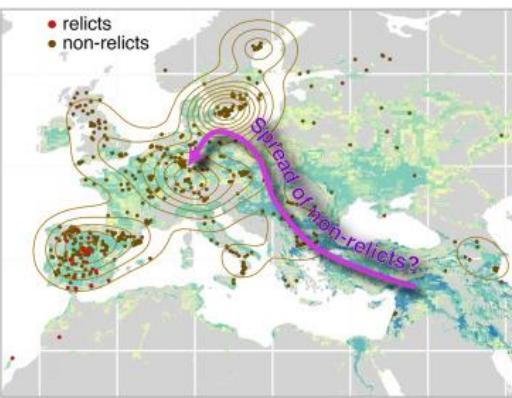
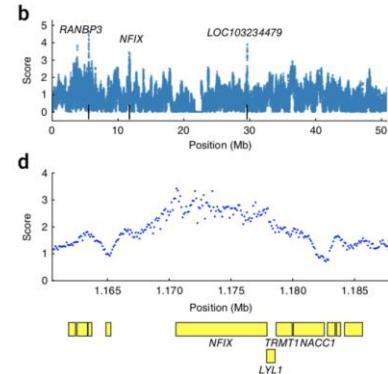
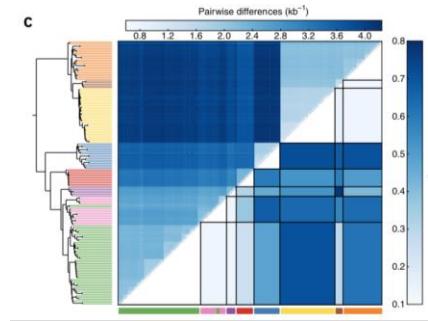
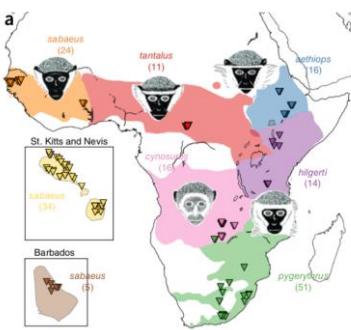


Goal: Link genotypes, phenotypes, and selection pressures

MY BACKGROUND

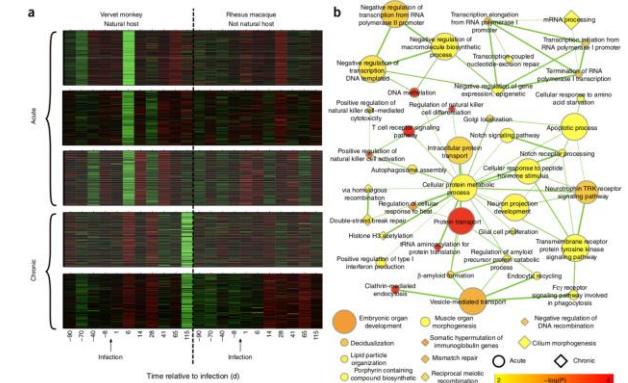


$$\sigma^2 < \text{E}_T [\text{Vars} [\theta]] + \gamma \text{Var}_T [\text{Es} [\theta]] + 2 \frac{1-m}{m} (\text{Vars} [\text{E}_T [\theta]] + \mathcal{C}[\theta])$$



8000 6000 4000 2000 0
Agriculture Established (Years BCE; > 1% land area)

Svardal et al. 2011 Evolution
 Svardal et al. 2014 Evolution
 Svardal et al. 2015 Theoretical Population Biology



Warren et al. 2015 Genome Research
 Svardal et al. 2017 Nature Genetics

The 1001 Genomes Consortium 2016 Cell
 Lee, Svardal et al. 2017 Nature Communications

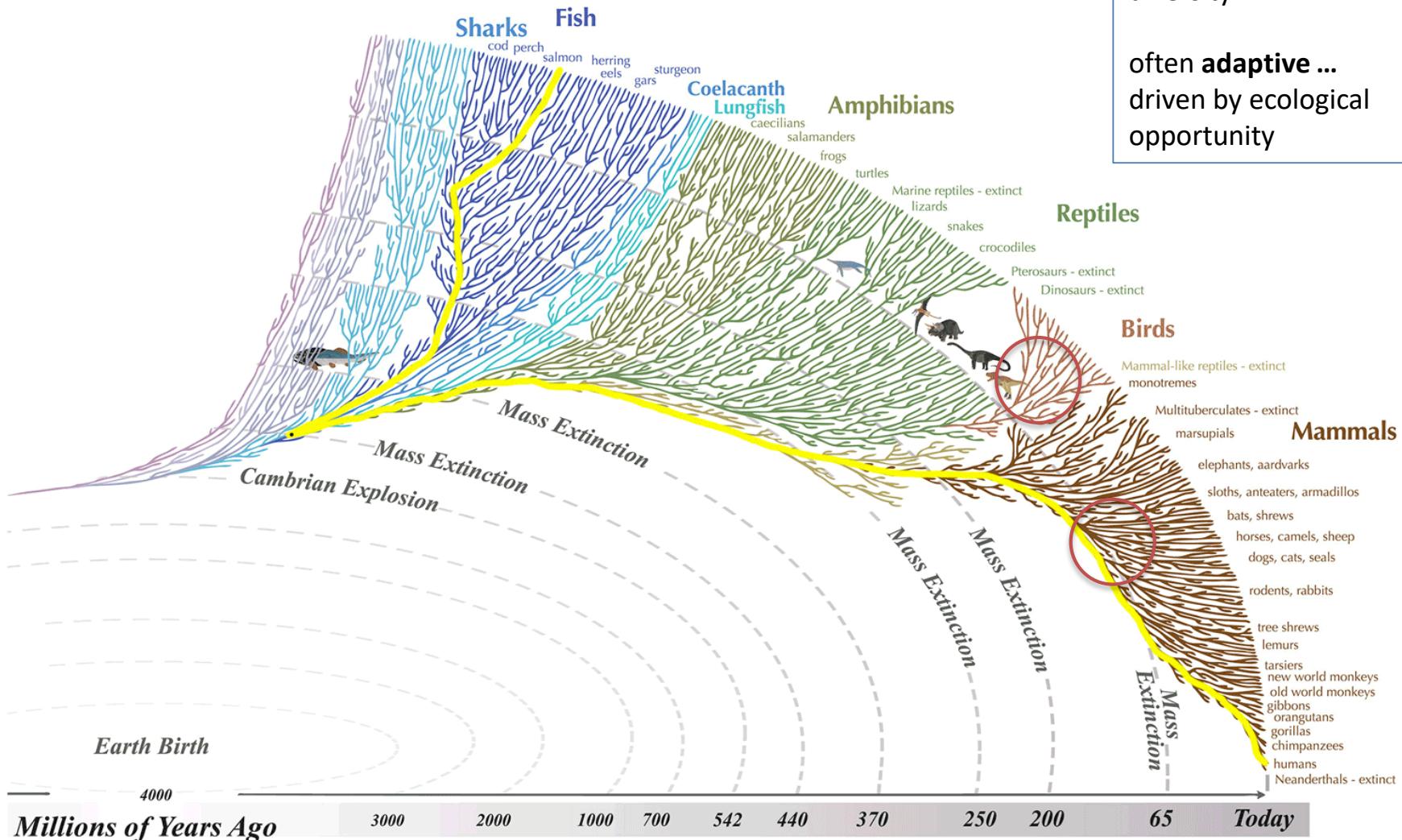


GENOMIC INSIGHTS INTO RAPID ADAPTIVE RADIATION

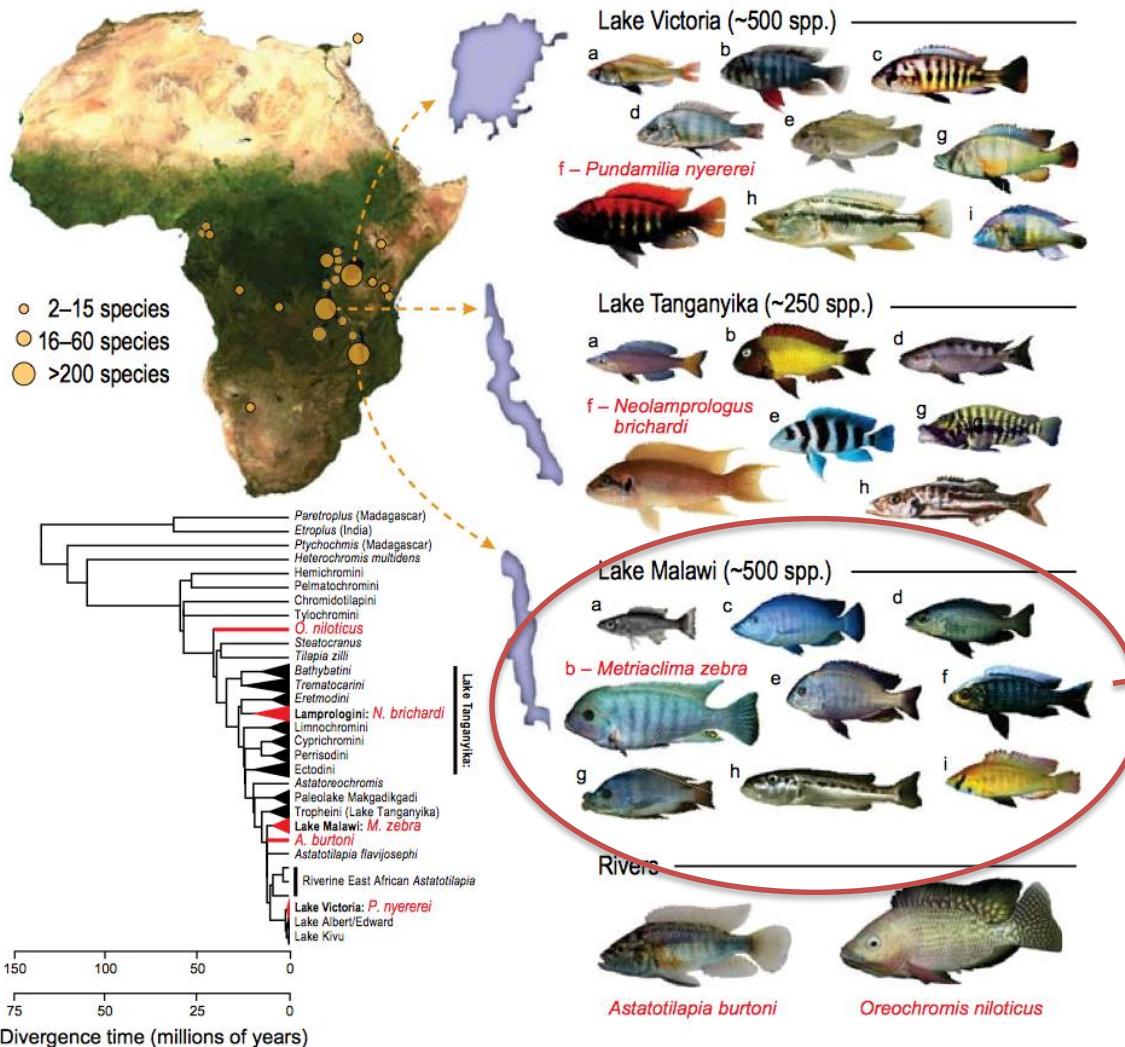
The history of life is a cumulative story of evolutionary radiations*

evolutionary radiation ...
increase in taxonomic diversity

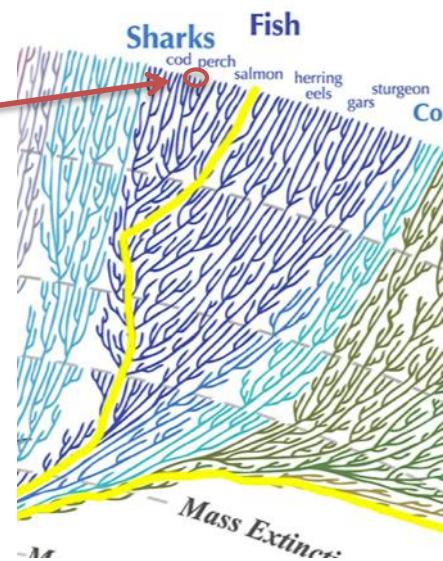
often adaptive ...
driven by ecological opportunity



Cichlids like to radiate



- a – pelagic zooplanktivore;
 - b – rock-dwelling algae scraper;
 - c – paedophage;
 - d – scale eater;
 - e – snail crusher;
 - f – reef-dwelling planktivore;
 - g – fat-lipped insect eater;
 - h – pelagic piscivore;
 - i – ancestral river-dweller;



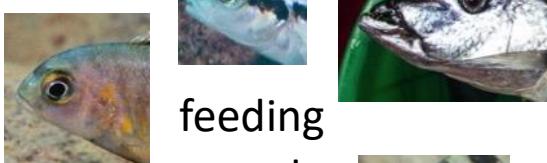
Remarkable phenotypic adaptations allowed Lake Malawi cichlids to conquer ecological niches



head
shape



pigmentation



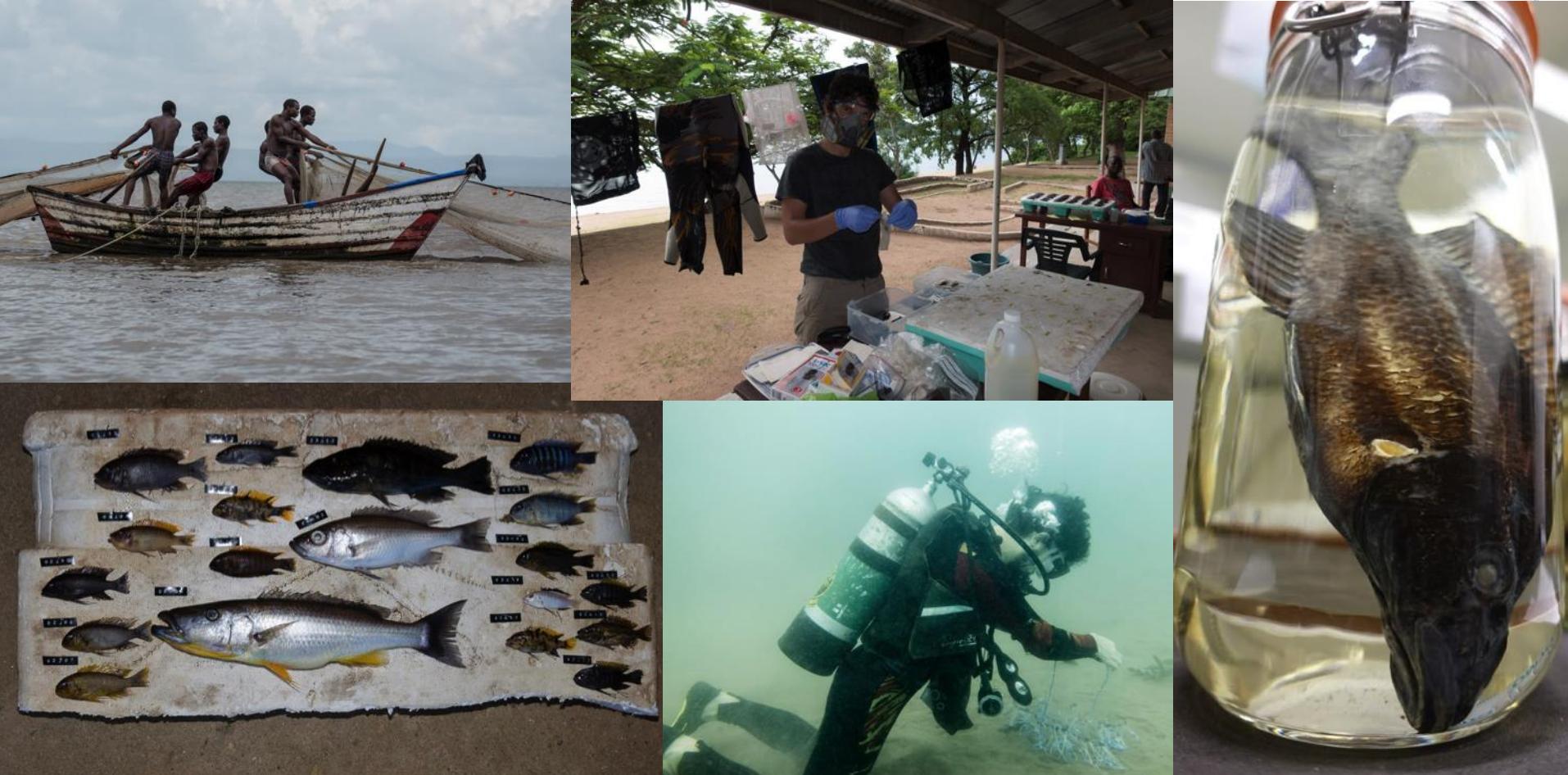
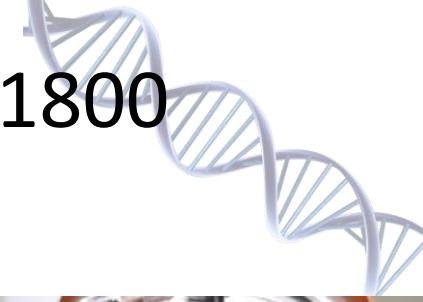
feeding
apparatus



And size, body shape, behaviour...



Sampling: >2500 samples; >250 species; >1800 museum specimen



This talk: ~ 140 samples from 70 species
whole genome sequencing at 15X/5X

Part 1 – Hybridisation within the radiation

- Genomic characterisation of the adaptive radiation
 - genetic relatedness
 - quantifying hybridisation and gene flow
 - genes under selection
 - evidence for adaptive introgression

joint work with



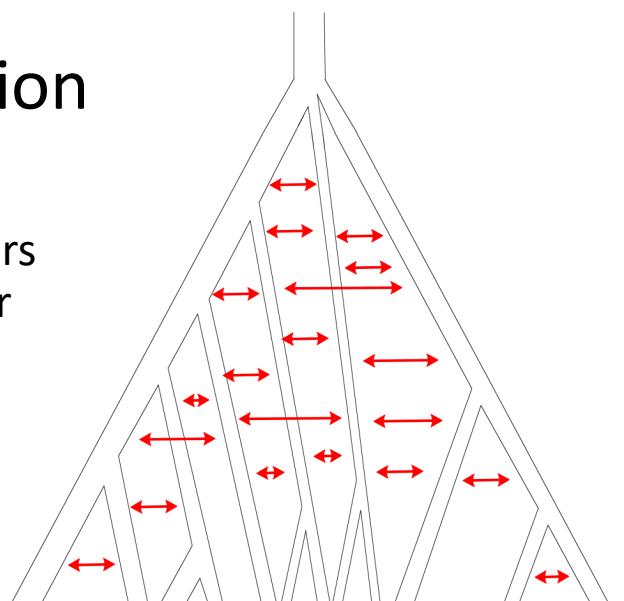
Milan Malinsky (now Uni Basel)
Erik Miska
Richard Durbin



Alexandra Tyers
George Turner



Martin Genner



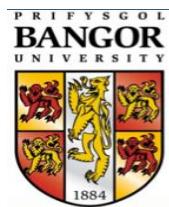
Questions

- Genetic relatedness
- Do species hybridise? Can we quantify gene flow?
- Which genes were under selection?
- Is there any evidence for adaptive introgression?

joint work with



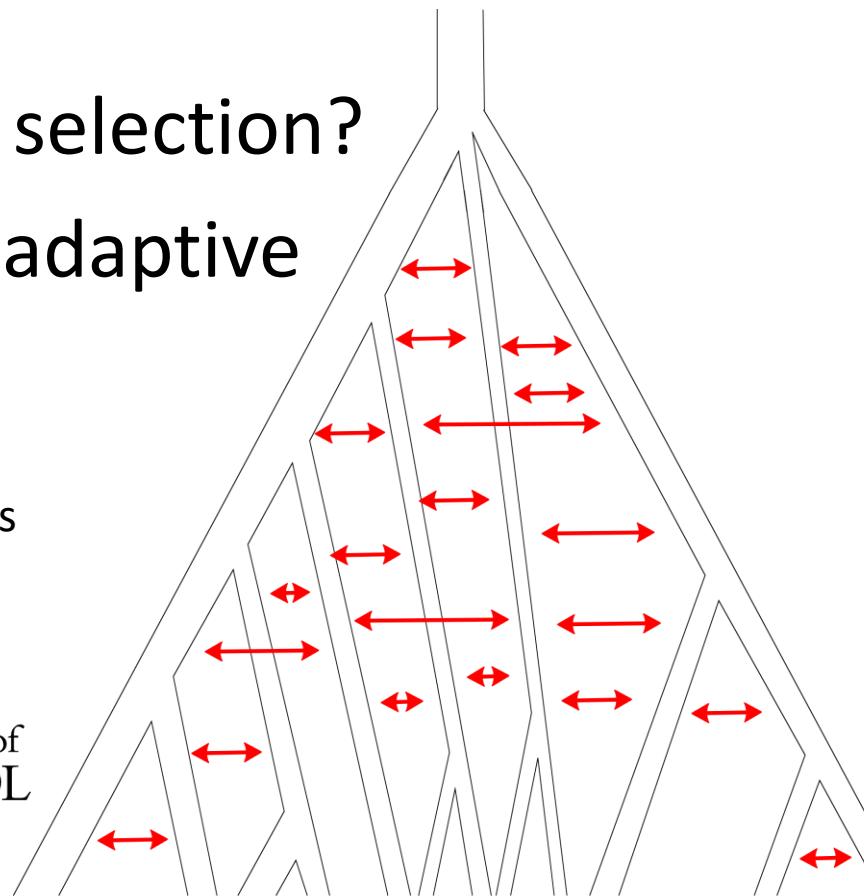
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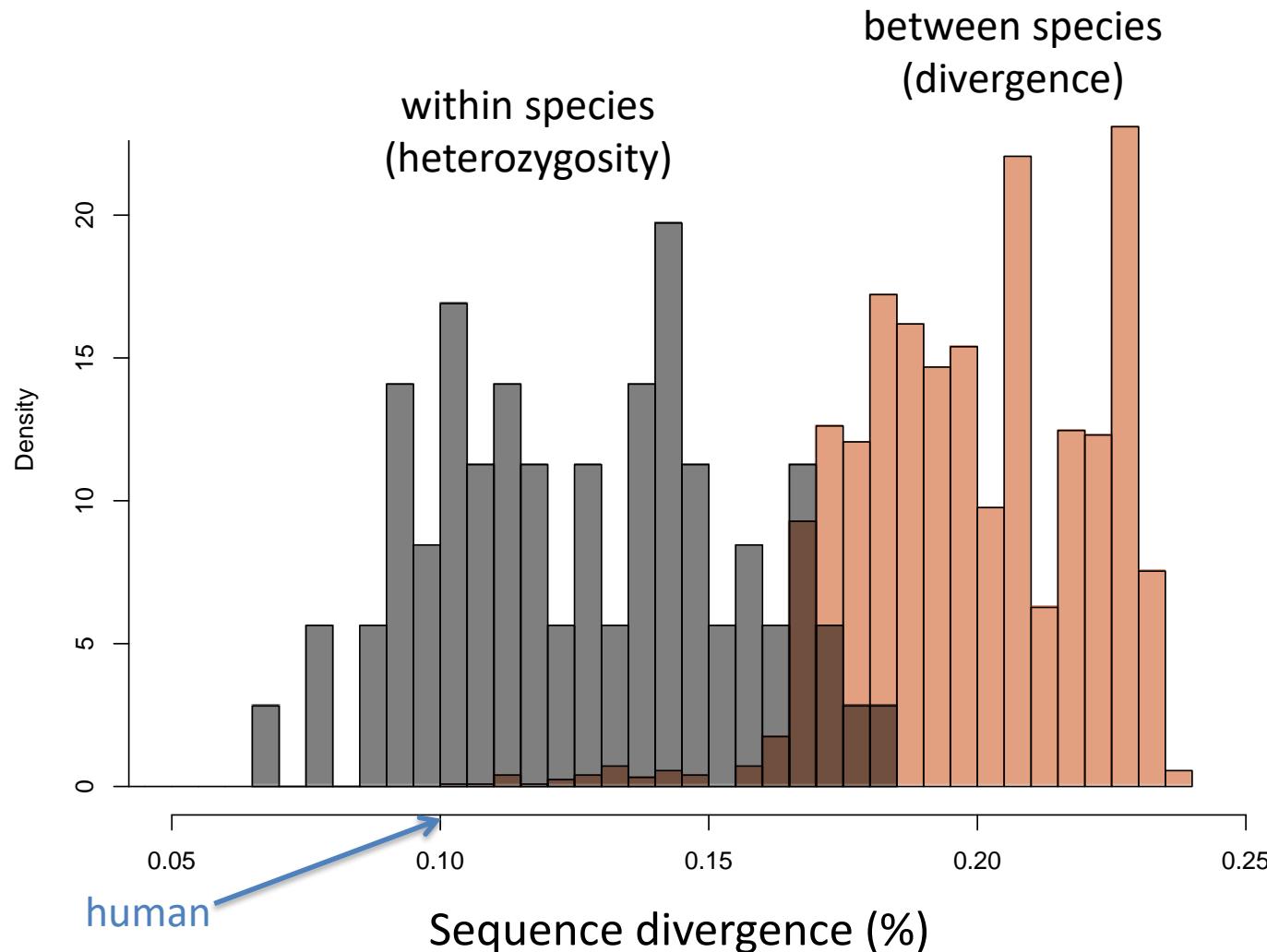
Alexandra Tyers
George Turner



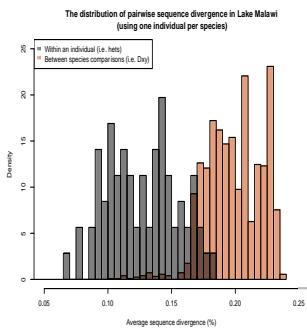
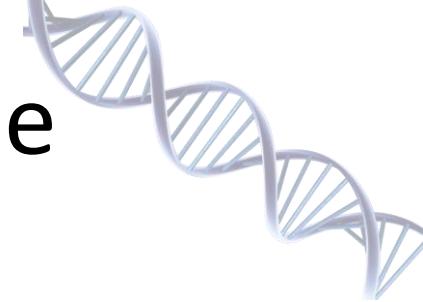
Martin Genner



Species are genetically not very diverse and extremely close



Species are extremely close

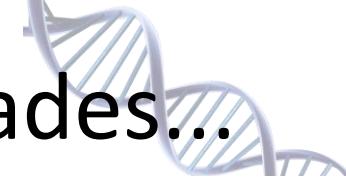


Human-chimp
divergence

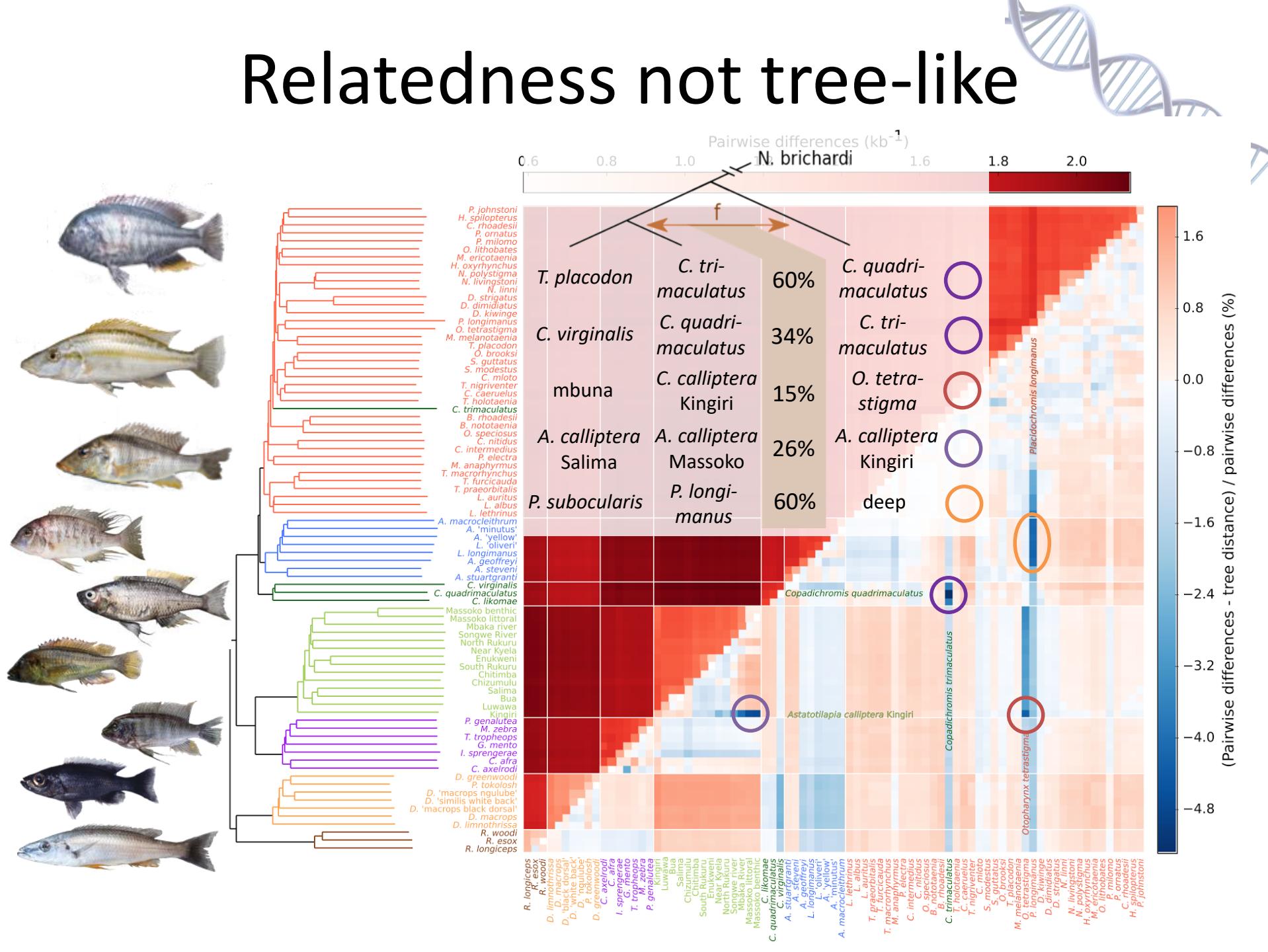


<http://www.spacedaily.com/images-lg/human-child-chimpanzee-baby-lg.jpg>

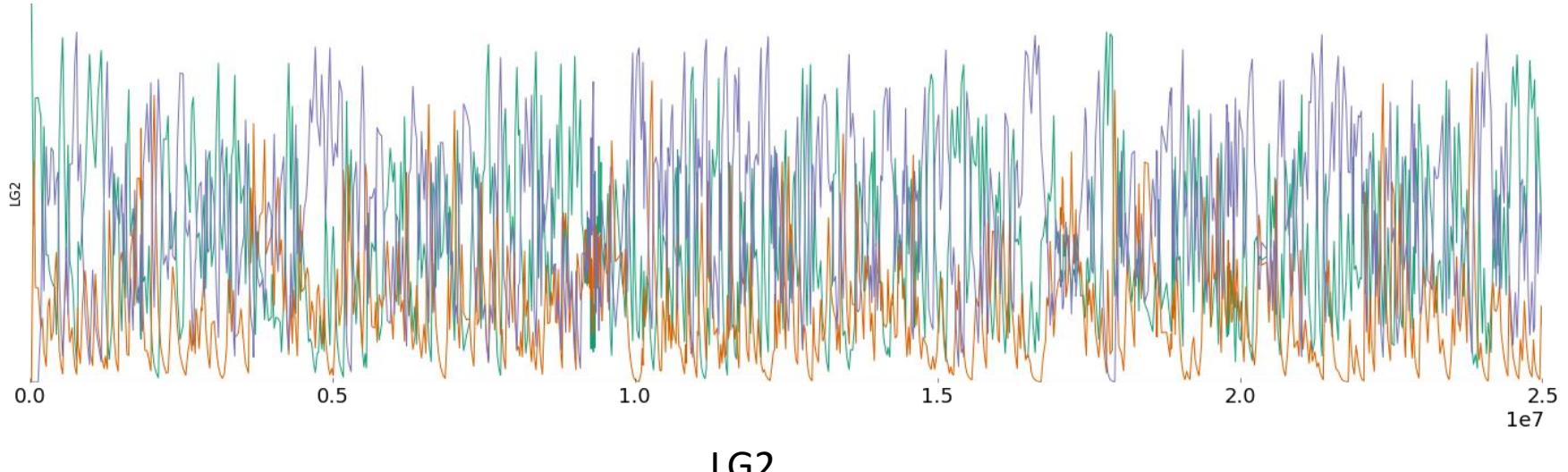
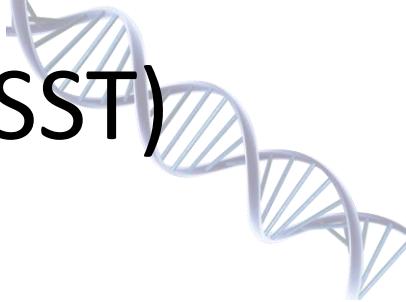
More or less well-defined major clades...



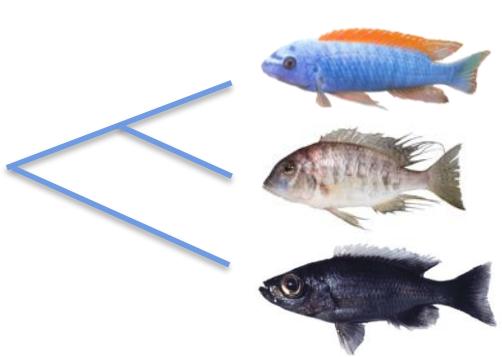
Relatedness not tree-like



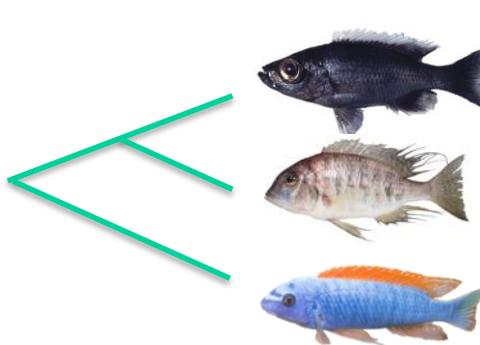
Trees along the genome (TWISST)



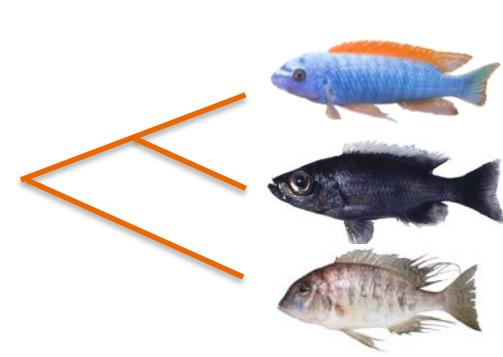
genome



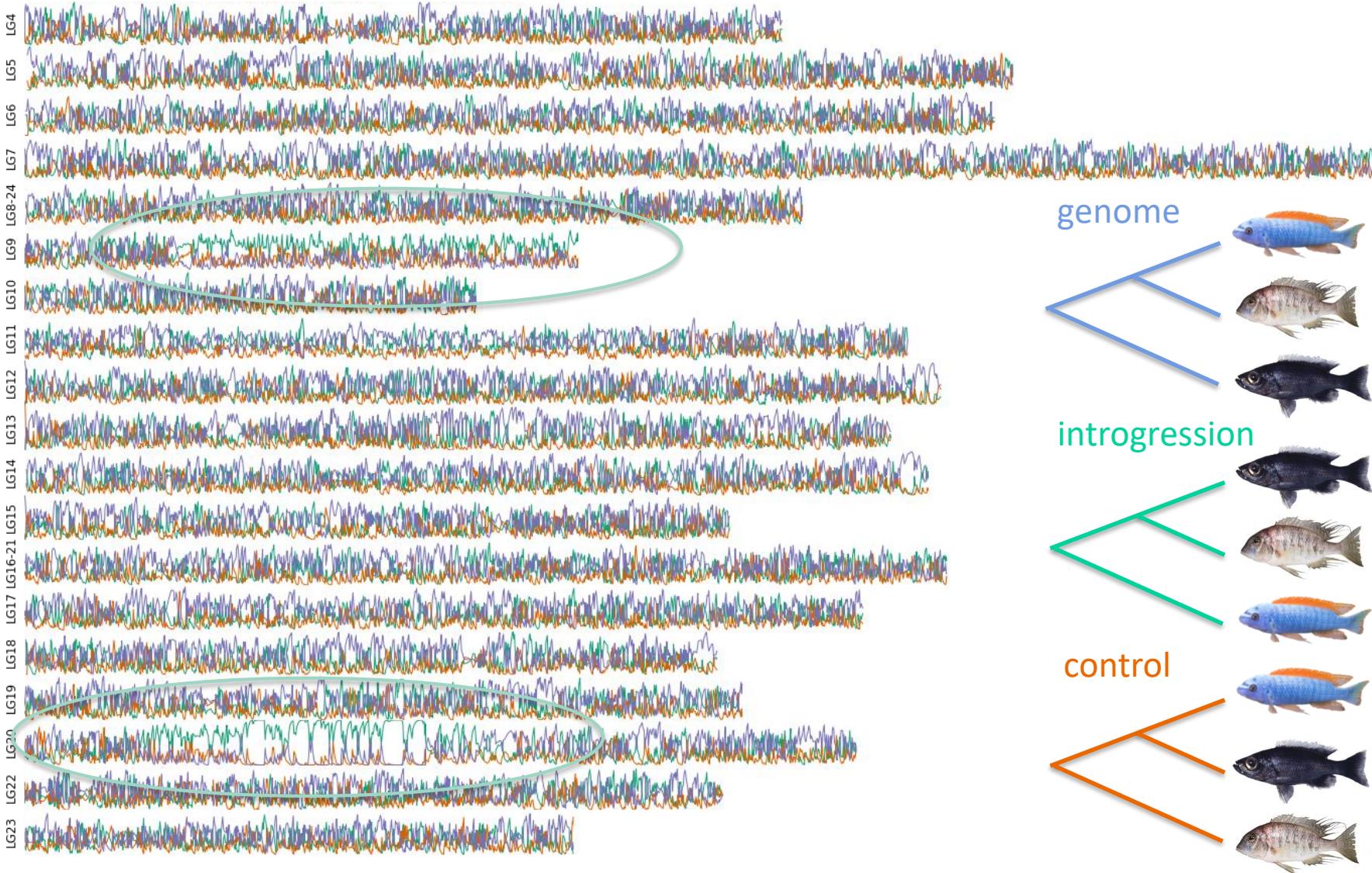
introgression



control



Chromosome-scale differences in relatedness patterns

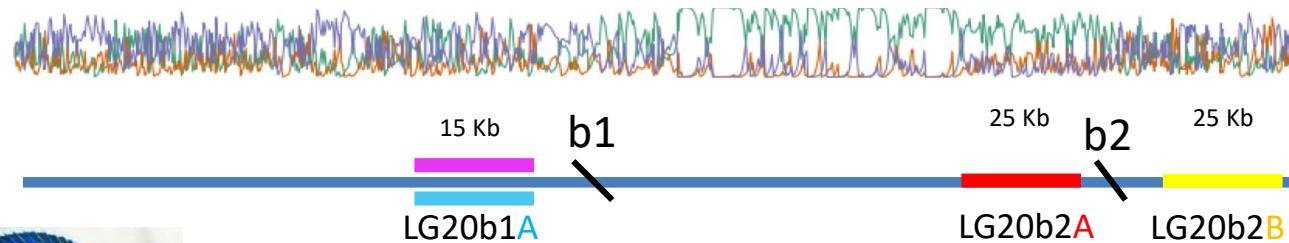


FISH supports inversions

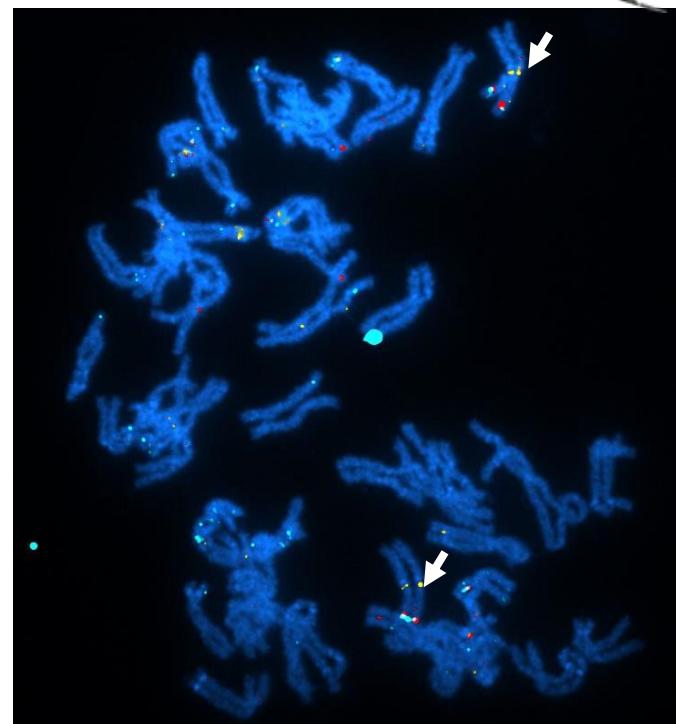
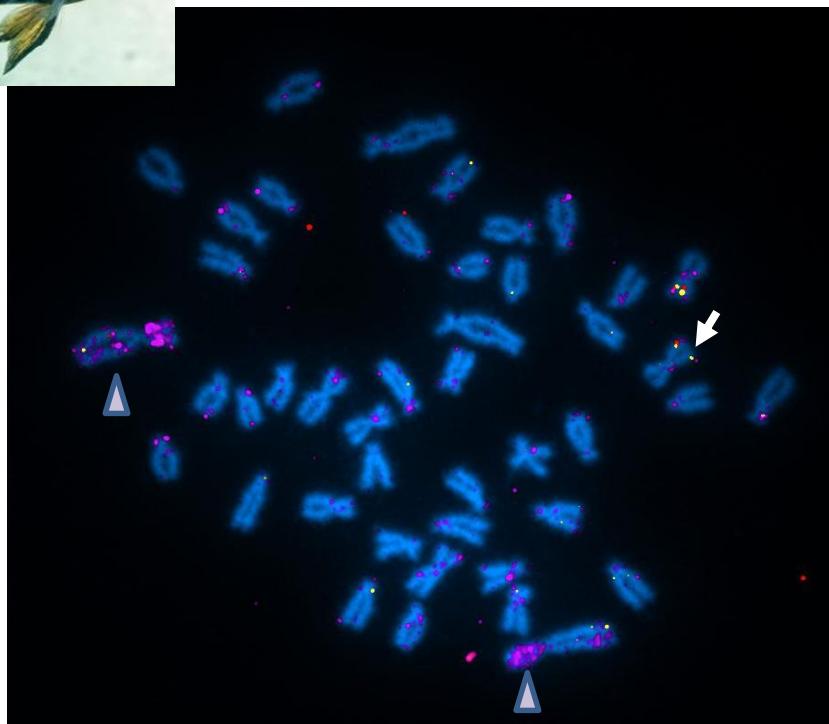
by Cytogenetics @ Sanger

Fluorescent in-situ hybridisation

LG20



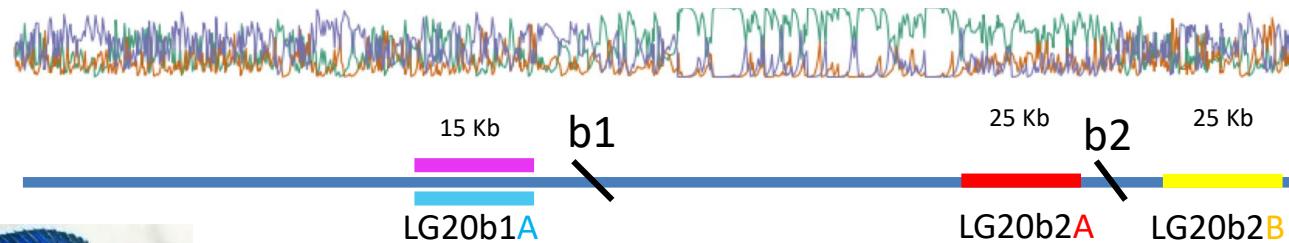
LG20



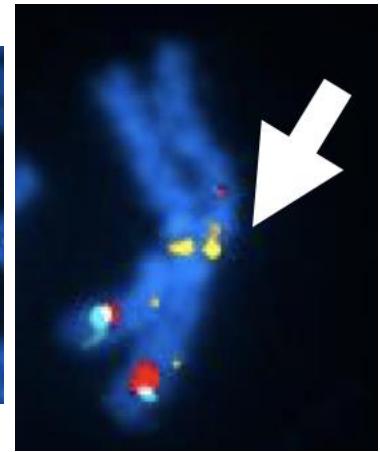
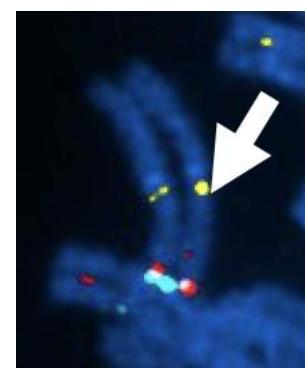
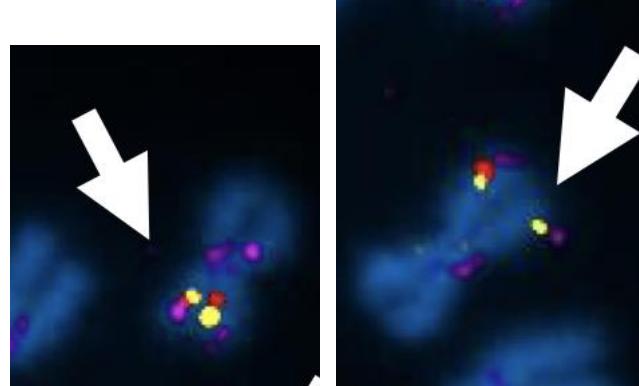
FISH supports inversions

Fluorescent in-situ hybridisation

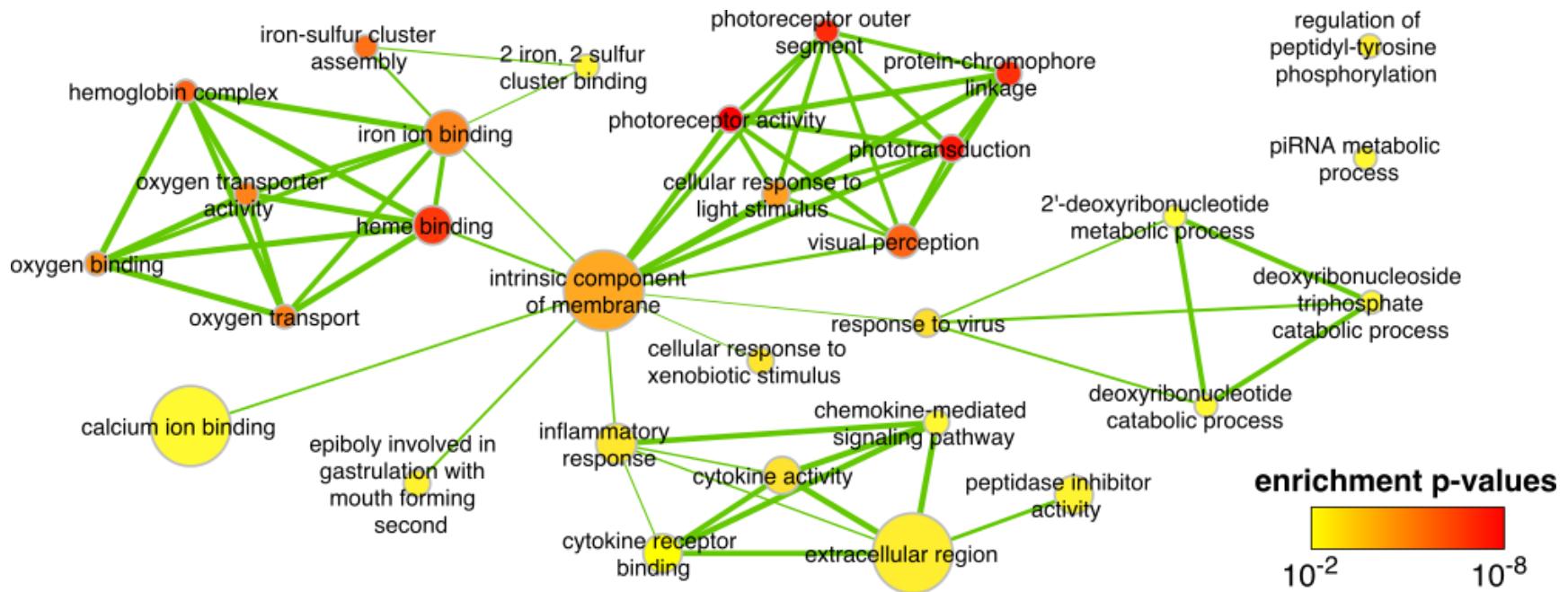
LG20



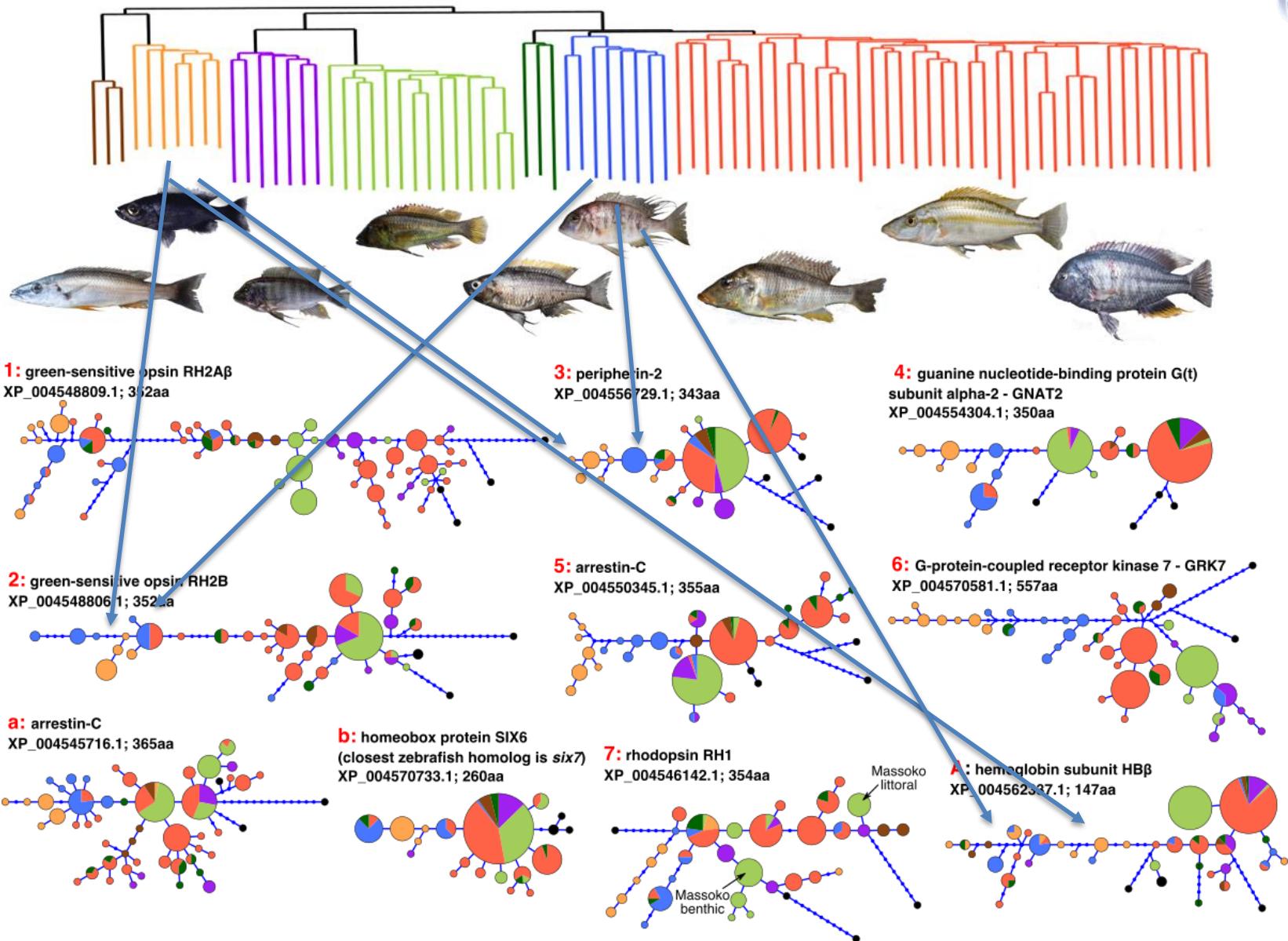
LG20



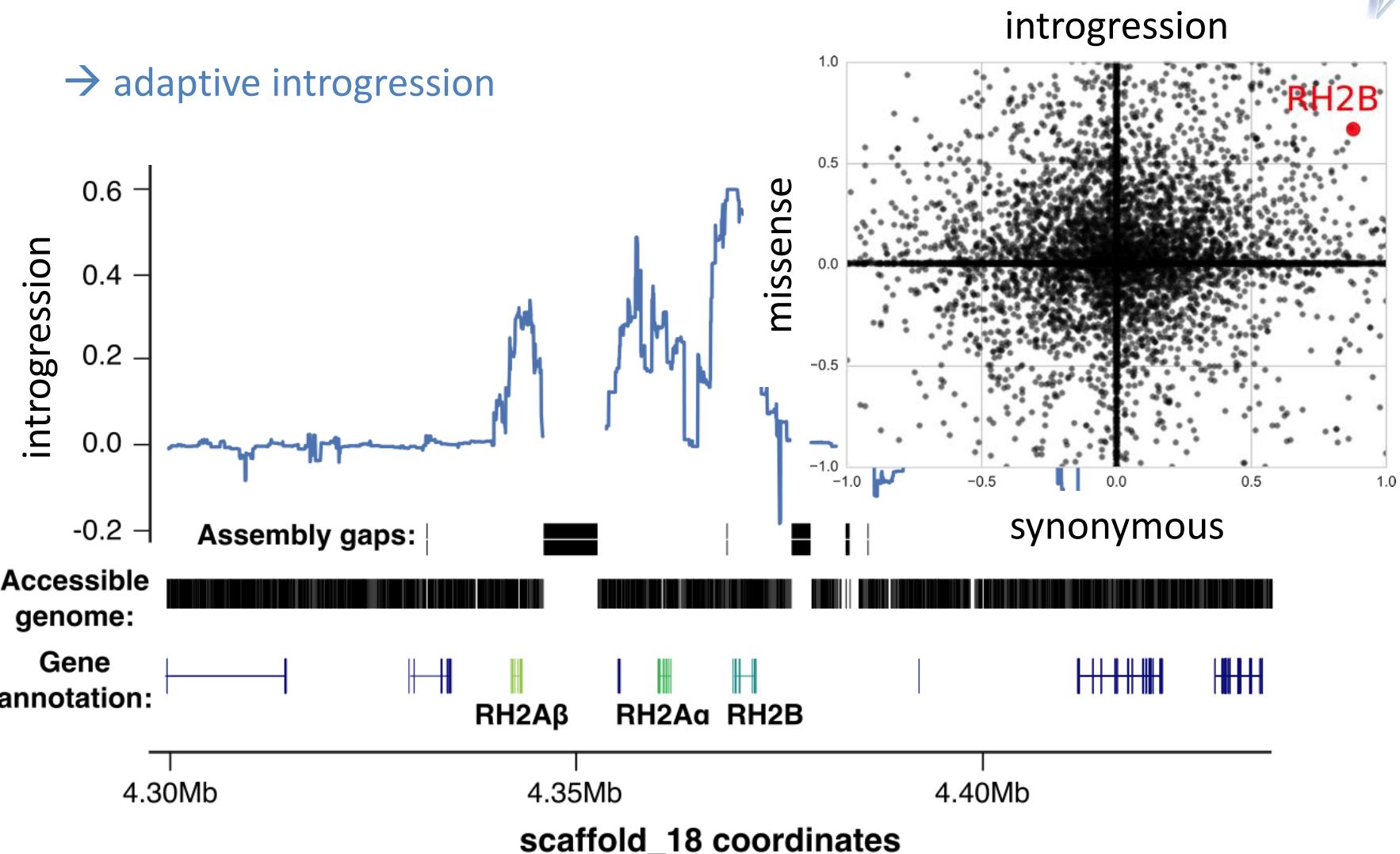
Visual system and oxygen transport pathways enriched for non-synonymous diversity



Haplotype networks of these genes show allele sharing between two genetically distant deep adapted groups

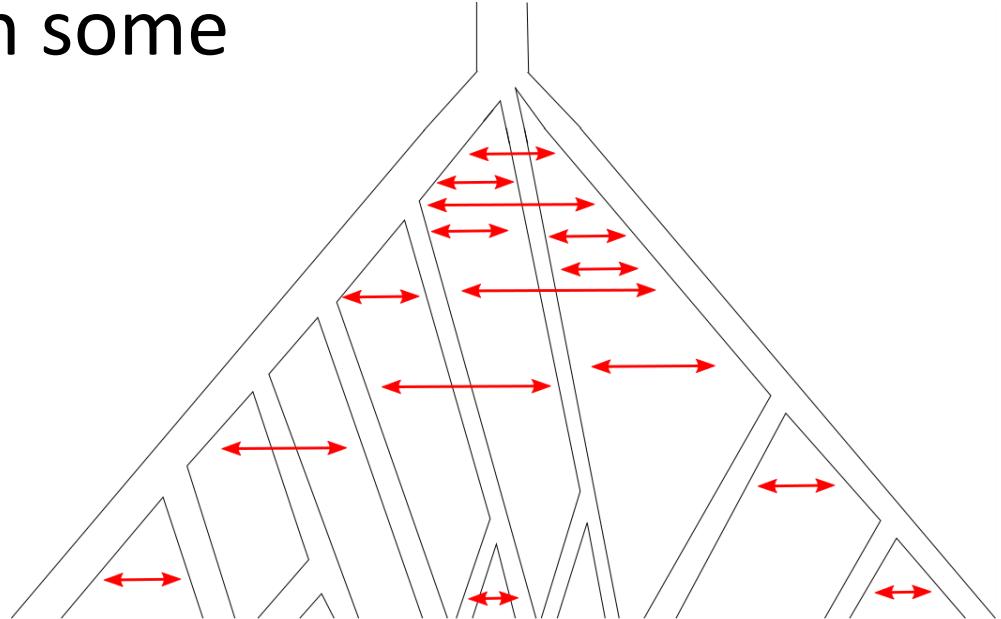


Long introgression haplotypes consistent with adaptive introgression

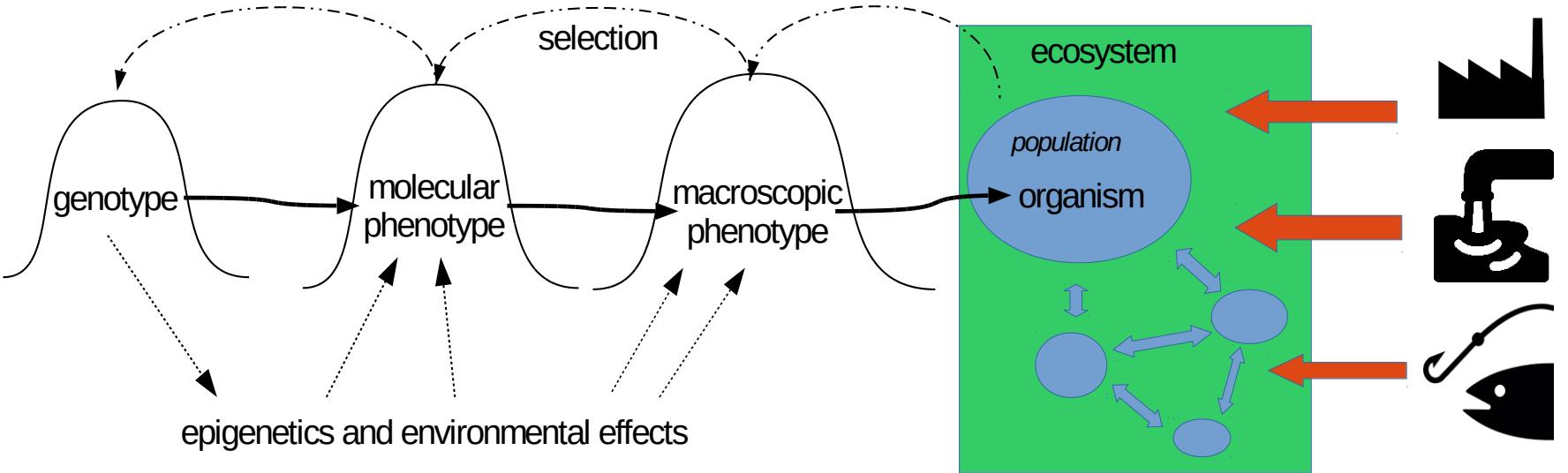


Conclusion

- A tree cannot capture relatedness
- Gene flow within and between major groups (mostly old)
- Long introgressed haplotypes (inversions?)
- Evidence for adaptive introgression of deep-adapted alleles in some opsin genes



Outlook

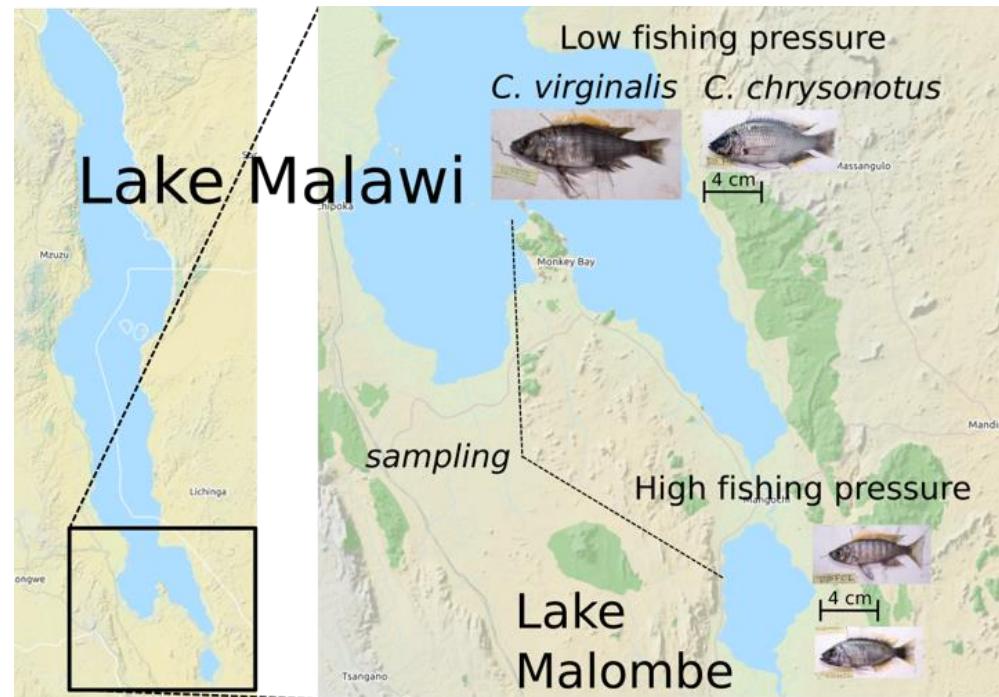


- How do organisms genetically adapt to human-made environmental challenges?
- Can we link adaptive genetic variants to phenotypes and to selective pressures?

The molecular and phenotypic basis of fisheries-induced adaptation in African cichlid fishes

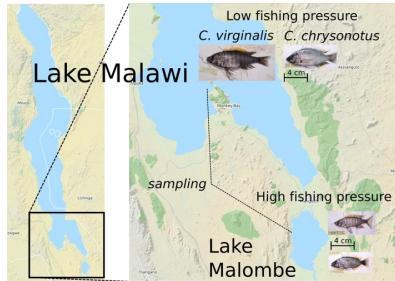
- Independent preliminary funding
- Collected >1000 samples
- Sequenced 144 individuals
- Local collaborators and experimental facilities

Study the genomics of fisheries-induced evolution



Objectives

- Sequence samples from different times and geographic locations



2017 samples

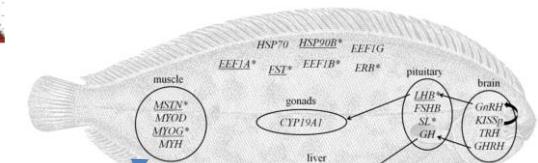
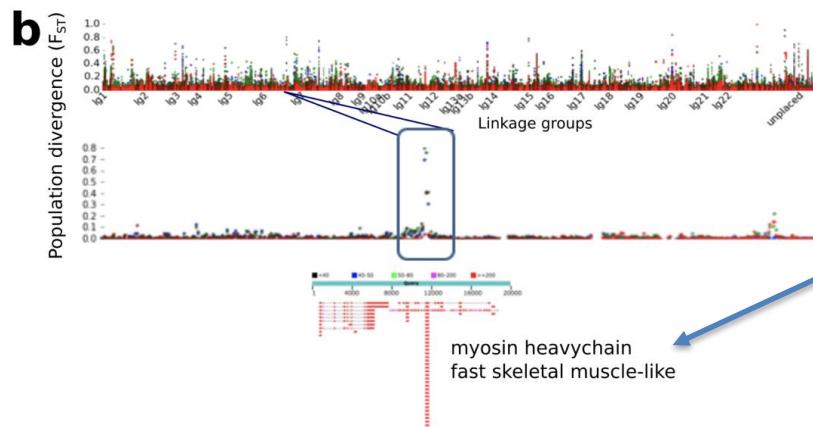
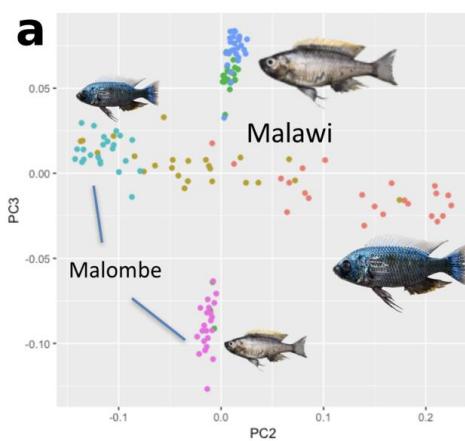


2002 samples



museum samples

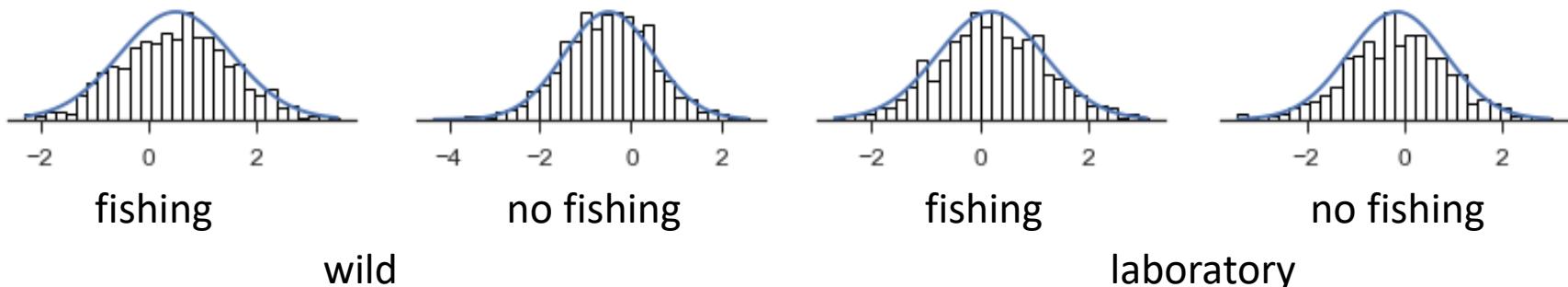
- Understand patterns of genetic variation and population divergence; find candidate loci for fisheries-induced adaptation



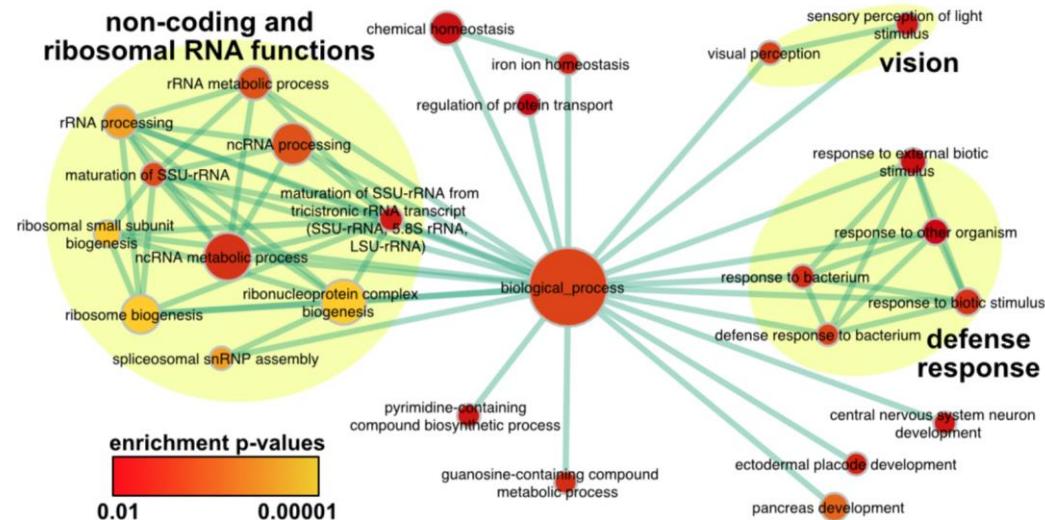
early maturation in European sole fish

Objectives (continued)

- Measure and compare phenotypes in wild/laboratory populations (maturation, metabolic rate, ...)



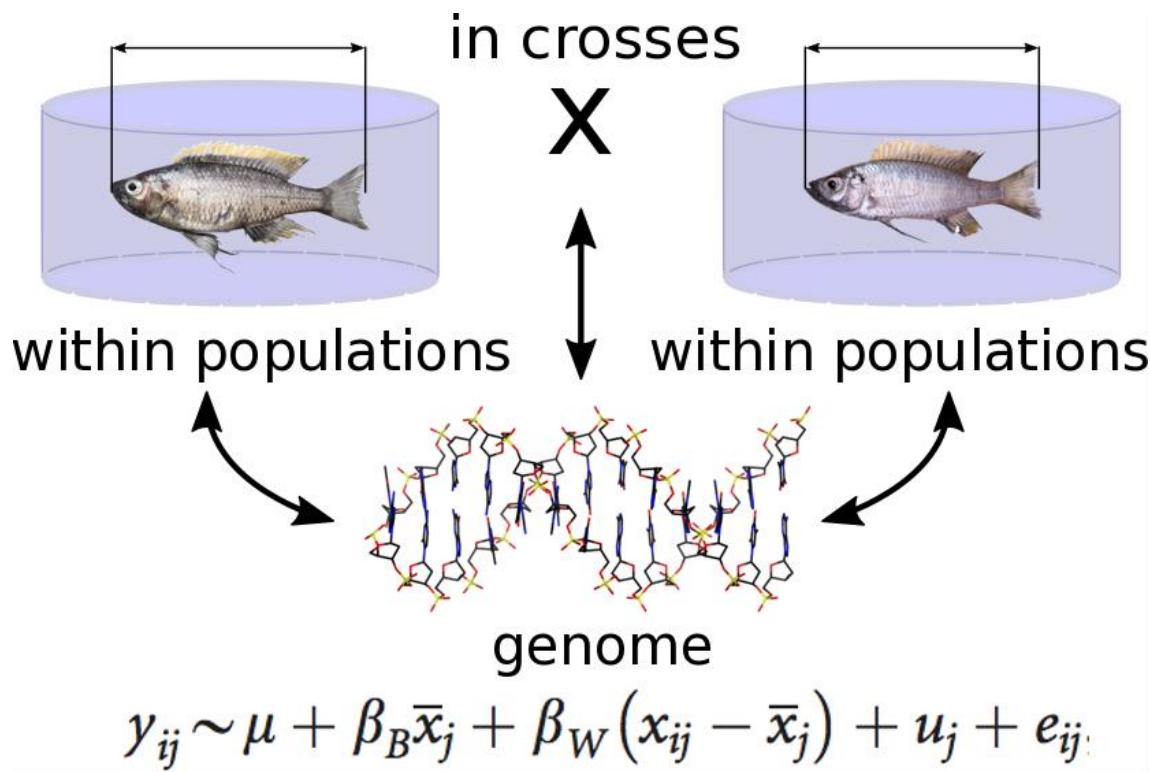
- Differential gene expression of the different populations





Objectives (continued)

- Trait mapping (GWAS, QTL)



Acknowledgements

Thank you!

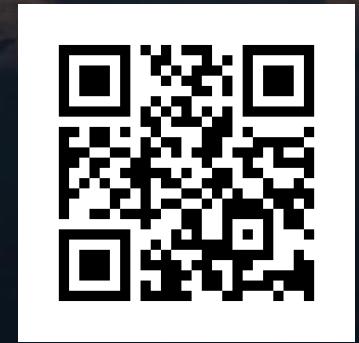


Preprint

<http://www.biorxiv.org/content/early/2017/06/06/143859>

Data and more info

<https://cambridgecichlids.org>



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(Malawi Fisheries)
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(University of Malawi)
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