## Biodiversity Bioinformatics – Case study

## Moulting genes across arthropod diversity



SIB course 31-08-2023









Giulia Campli, PhD student Prof. Robert Waterhouse group University of Lausanne, Department of Ecology and Evolution

#### The question: why moulting?

Arthropods represent the richest and most diverse Metazoan phylum (reported close to 1 million species); it comprehends:

- Chelicerata (spiders, scorpion, mites...)
  Myriapoda (millipedes, centipedes)
  Crustacea (crabs, water fleas, barnacles ...)
  Hexapoda (insects and non-insects)

They share a segmented, modular body plan, jointed appendages and an outer exoskeleton.

The exoskeleton has to be periodically shed, in a process defined as Moulting (ecdysis), which represents a key event in all arthropods life cycle.

→ Understanding the mechanisms of moulting and the diversity of moulting patterns is a key to understand arthropod evolutionary history and success.

BBC

https://www.youtube.com/watch?v=C0D1cR5hPN8 above: Spider crab (BBC, Blue Planet II) https://www.youtube.com/watch?v=yi8G17dessg&t=28s bottom: Cicada (tdtangents)

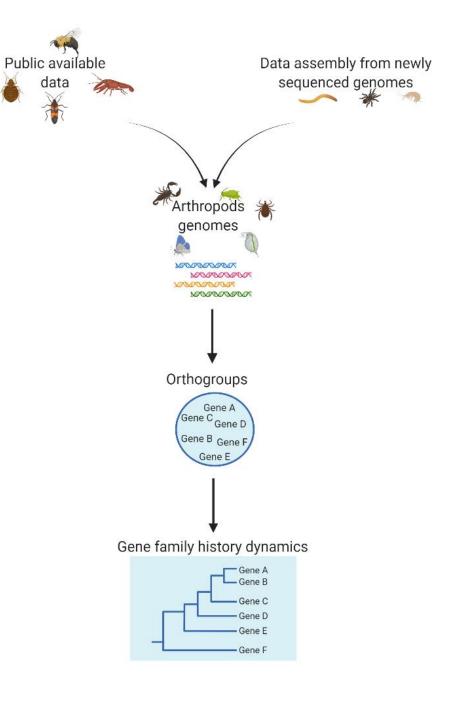
Giulia Campli

#### **Outline**

Genomics to investigate changes in moulting gene repertories and their functional role

- 1) Which genomic resources can be used to study the moulting gene repertories?
- 2) Is it possible to identify genes coding for moulting pathway components across Arthropoda evolution?
  - → OrthoGroup\* inference
- 3) Which are the evolutionary events occurred in their gene family histories?
  - → Ancestral state reconstruction and gene-tree-species-tree reconciliation

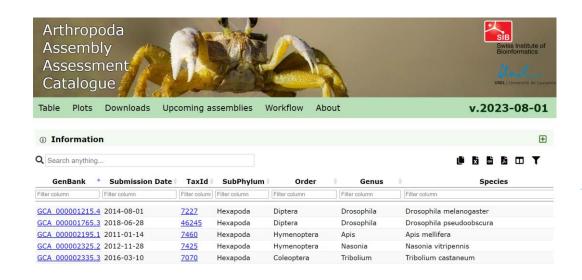
OrthoGroup\*: set of genes descended from a single gene in the last common ancestor of the compared genomes



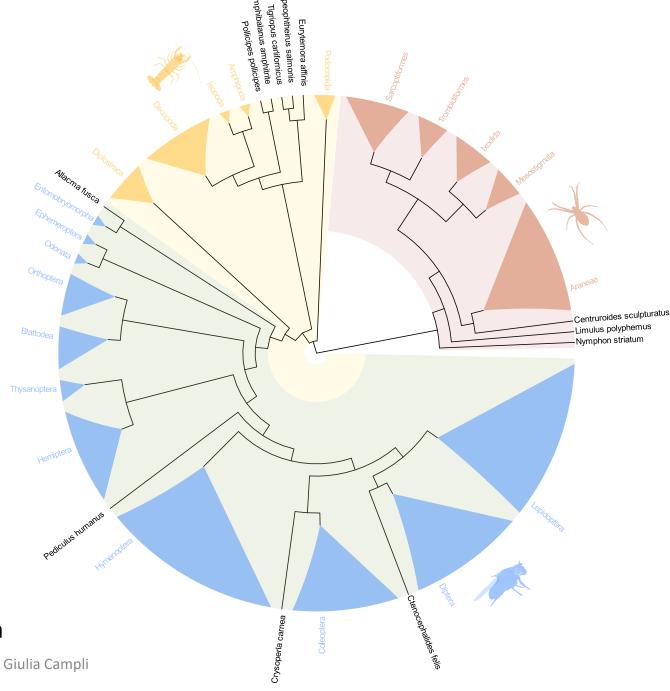
3 Giulia Campli

#### 1) Input data selection

a) Arthropoda Assembly Assessment Catalogue (https://evofunvm.dcsr.unil.ch/table.html)

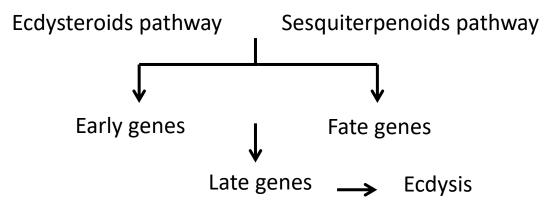


- b) Phylogeny from single-copy orthologs (BUSCOphile)
- c) Rooting
- d) Calibration
- → 150 species spanning almost 600 mya evolution

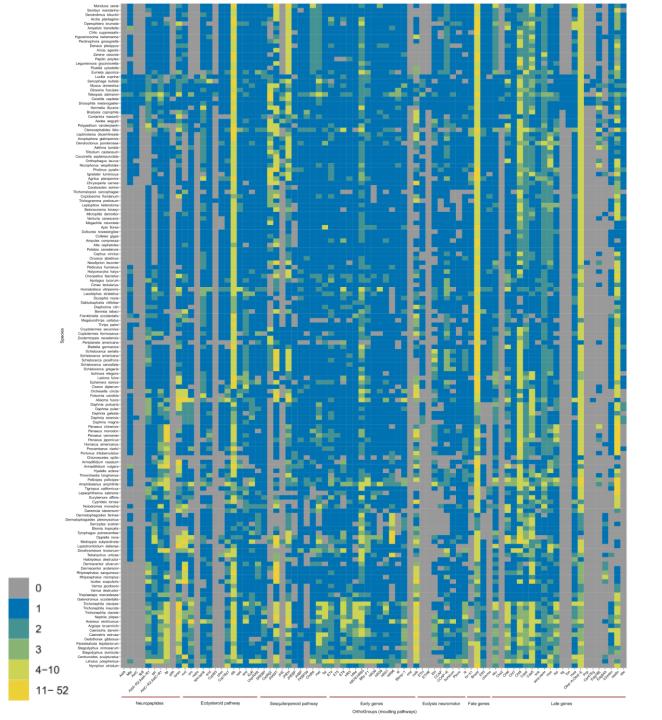


## 2) Identification of «moulting genes» across arthropods

Orthology delineation (OrthoLoger) and extrapolation of 65 gene families for downstream analysis:

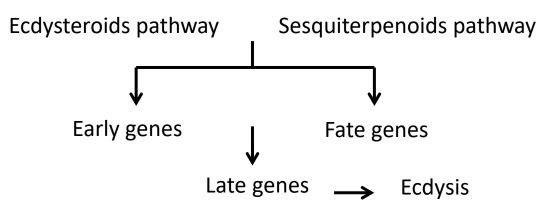


- Mostly single-copy genes across all the species
- Multicopy genes in nearly all the species
- Mostly restricted to few species

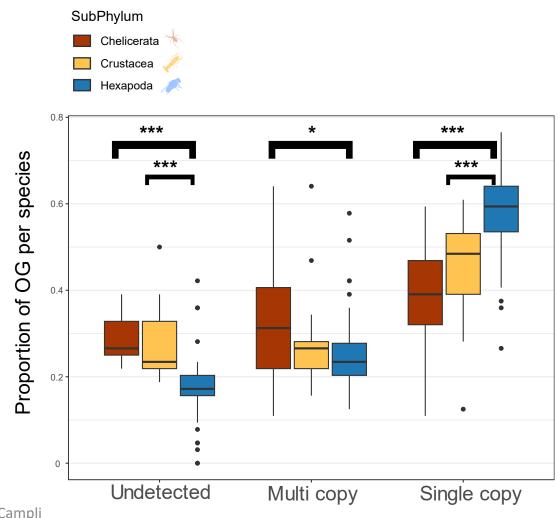


# 2) Identification of «moulting genes» across arthropods: are some lineages more dynamic than others?

Orthology delineation (OrthoLoger) and extrapolation of 65 gene families for downstream analysis:



- Mostly single-copy genes across all the species
- Multicopy genes in nearly all the species
- Mostly restricted to few species
- → Orthology delineation suggest gene history in Chelicerata are highly dynamic



#### Ancestral count reconstruction of gene copy number

Given orthology data and a species phylogeny, the ancestral gene copy number in ancestral species is estimated from gene copy number in extant species (CAFÉ software)

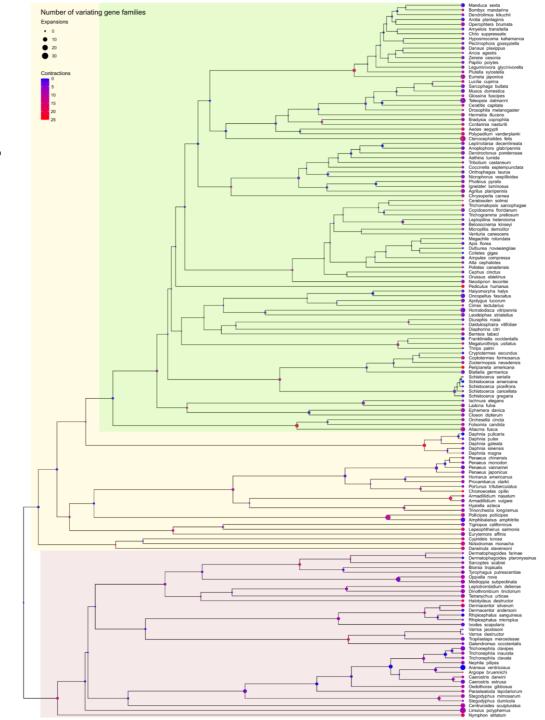
→ It allows quantification of potential gene gains and gene losses in the framework of the species evolution

1449 total expansions and 1401 total contractions occurred across all the families

Point big and red = many families expanded and contracted, Point small and blue = no events

Considering size variation from all the families:

- A. Which are the nodes having the most contractions/expansions?
- B. Which dynamics do order LCAs show?
- C. How many, on average, contractions/expansions do orders have?
- D. How many expanding/contracting families are found along the species evolutionary path?
- E. Which is the contraction/expansion family rate?



#### Ancestral count reconstruction of gene copy number

Given orthology data and a species phylogeny, the ancestral gene copy number in ancestral species is estimated from gene copy number in extant species

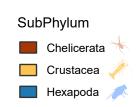
→ It allows quantification of potential gene gains and gene losses in the framework of the species evolution

1449 total expansions and 1401 total contractions occurred across all the families

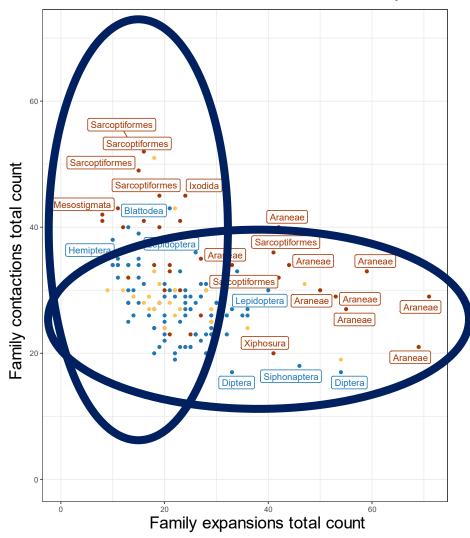
Point big and red = many families expanded and contracted, Point small and blue = no events)

## D. How many expanding/contracting families are found along the species evolutionary path?

→ Two main evolutionary trajectories



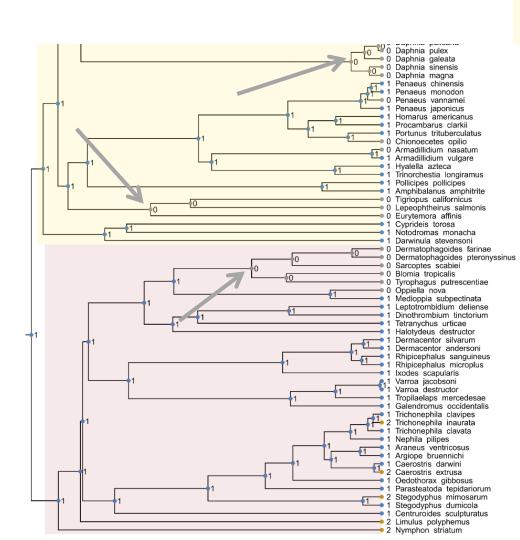
#### Ancestral state reconstruction analysis

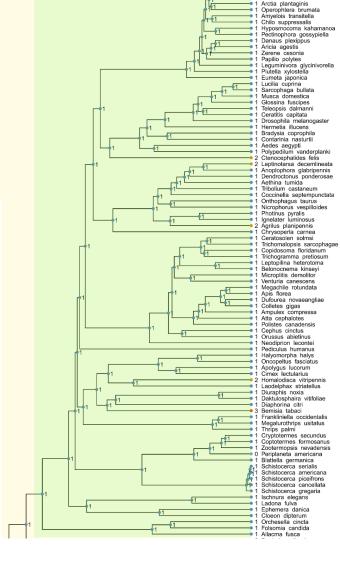


#### Ancestral count reconstruction of gene copy number

A closer look to individual families: E93 gene family

- •11 total gains and 8 total losses
- Single copy in Hexapoda
- Present beyond Hexapoda
- Lost in sublineages: Sarcoptiformes,
   Daphnids, Copepoda
- Duplicated in 5 chelicerate and 5 insect species





Bombyx mandarina

Gene copy number  $\bullet$  0  $\bullet$  1  $\bullet$  2  $\bullet$  3

# Gene-tree-species-tree reconciliation and results comparison

- A. Reconciliation (NOTUNG): 3 total duplications and 20 total losses
- B. Ancestral reconstruction: 11 total gains and 8 total losses
- → candidate gained copies are fragmented sequences, which are removed from the alignment and counted as losses (MAFFT-TrimAl-RAxML)
- → Duplications in *L. polyphemus*, *N. striatum* and *A. planipennis* confirmed
- → Ancestral losses Sarcoptiformes, Copepoda and Daphnida confirmed

THANKS FOR YOUR ATTENTION!

