# A (short) introduction to phylogenetics

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*Genetic data analysis using* R, University of Leuven
27-10-2014

# Outline

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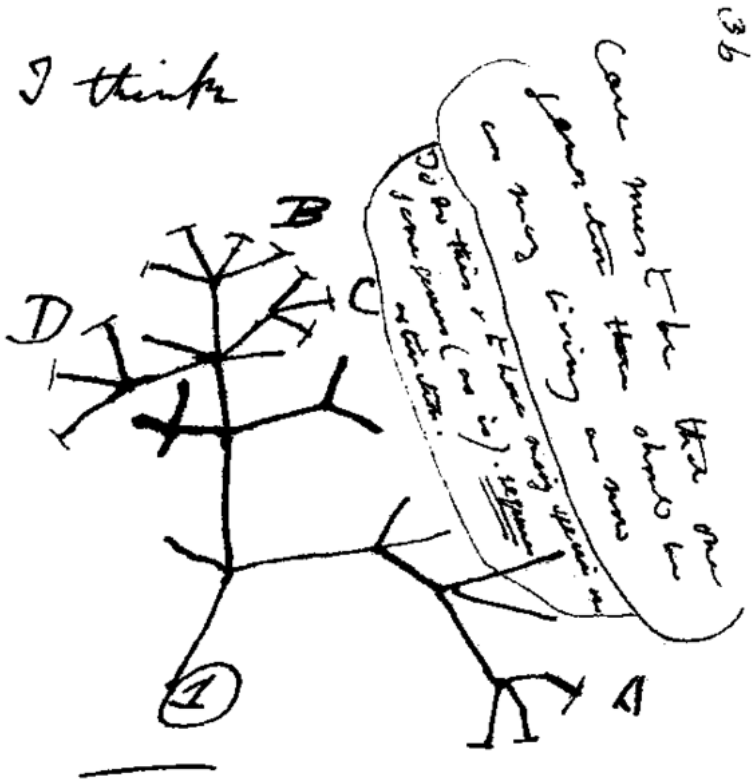
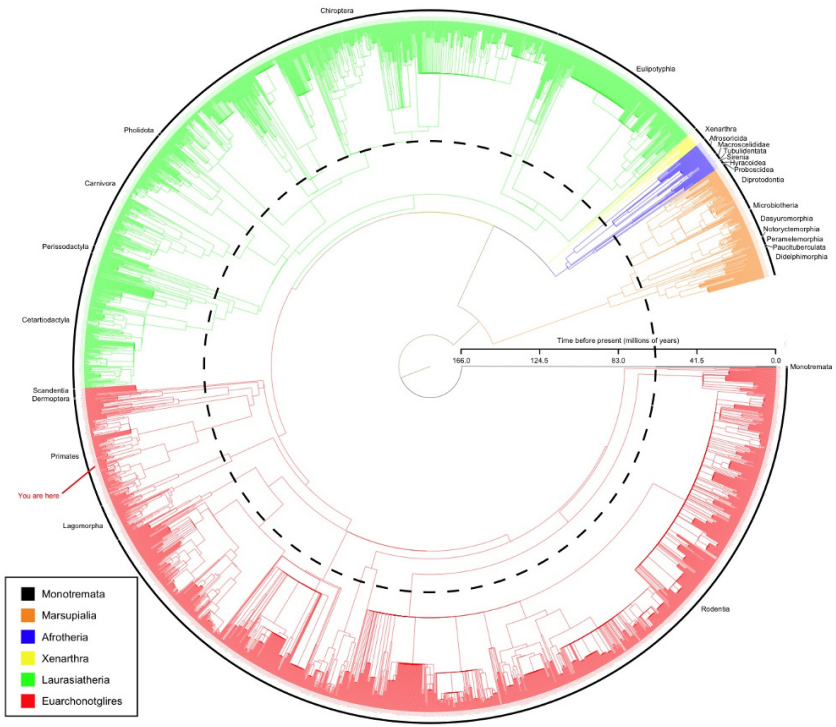
# Phylogenetics: from the origins...




C. Darwin, Notebook, 1837.

*'From the first growth of the tree, many a limb and branch has decayed and dropped off; and these fallen branches of various sizes may represent those whole orders, families, and genera which have now no living representatives, and which are known to us only in a fossil state.'*

# Phylogenetics: ...to the present




Bininda-Emonds *et al.*, 2007, Nature.

- phylogenetic trees are part of the standard toolbox of genetic data analysis
- represent the evolutionary history of a set of (sampled) taxa

# And the main difference is...

**Current trees look better!**

(and some other minor differences)

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- DNA sequencing revolution
- huge data banks freely available (e.g. GenBank)
- easier, cheaper, faster to obtain DNA sequences
- increasing number of full genomes available

**Different ways to exploit this information.**

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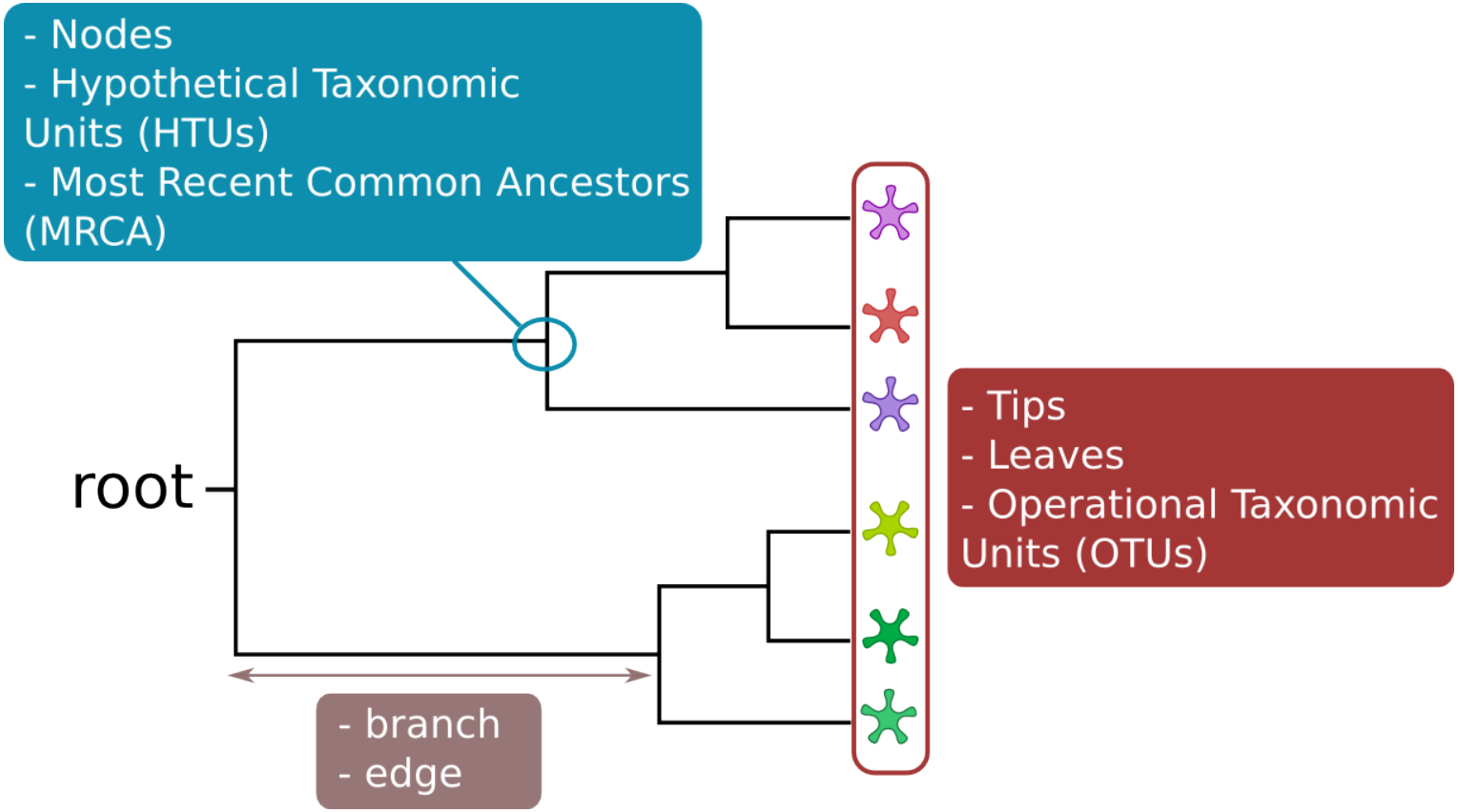
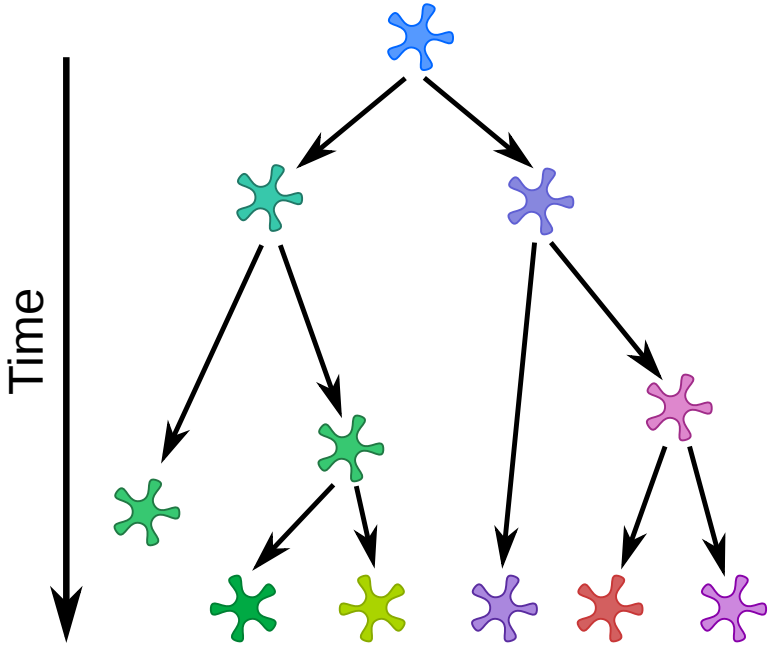
# Phylogenetic trees: some useful terms

**Phylogenetic tree**: representation of evolutionary relationships between a set of taxa.



# Accumulated substitutions tell us about the genealogy

**Substitution**: replacement of a nucleotide (e.g. a $\rightarrow$ t)

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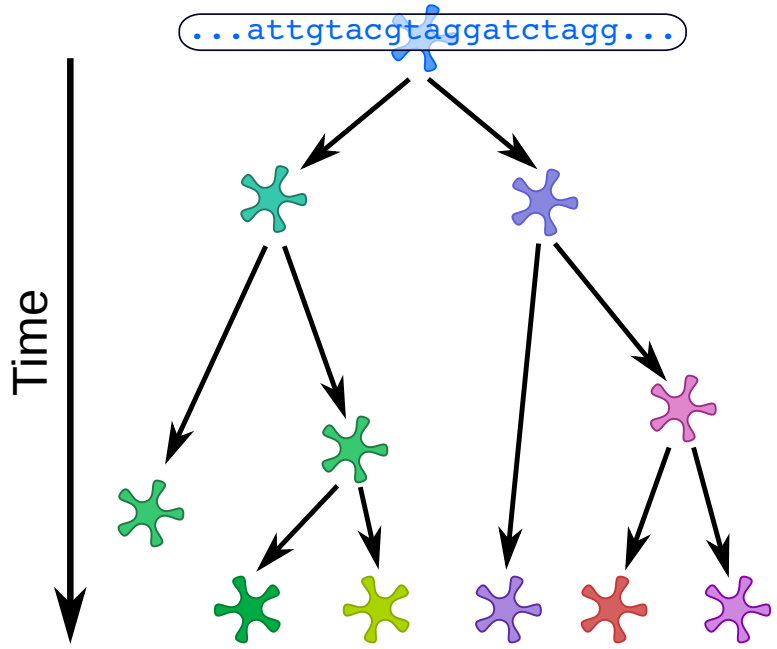


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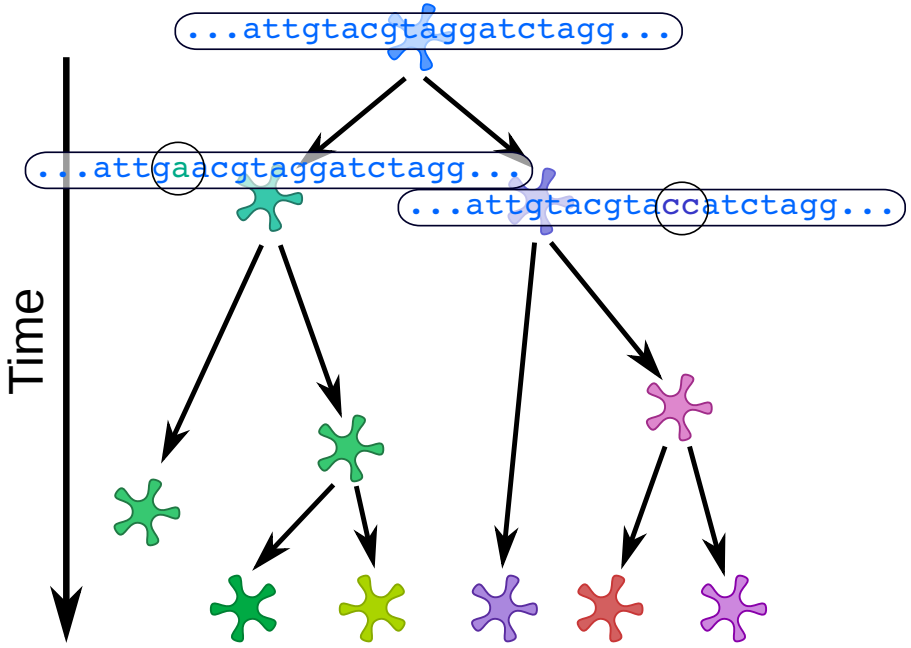


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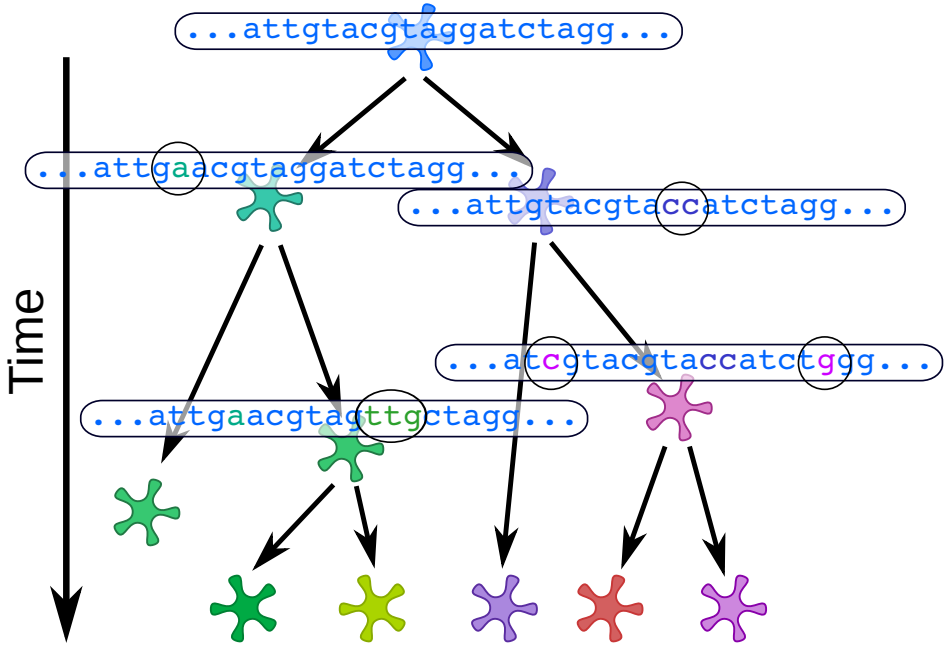


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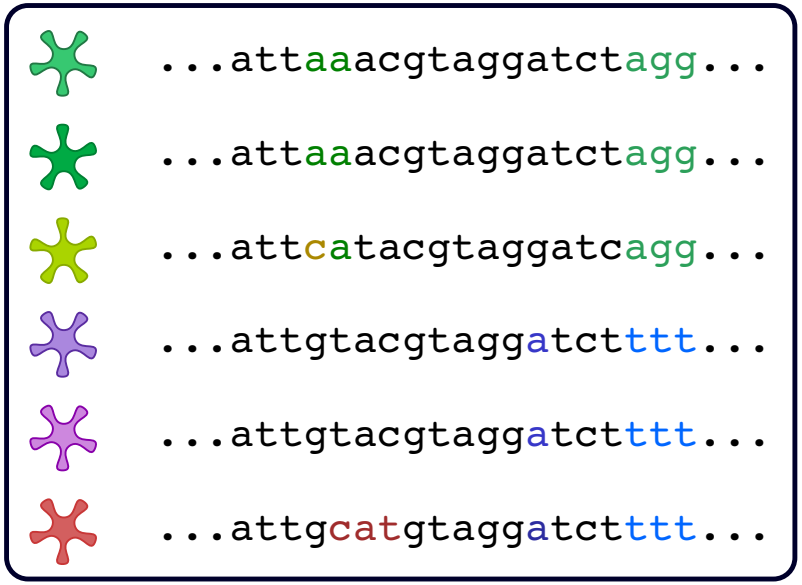
# From alignments to phylogenies



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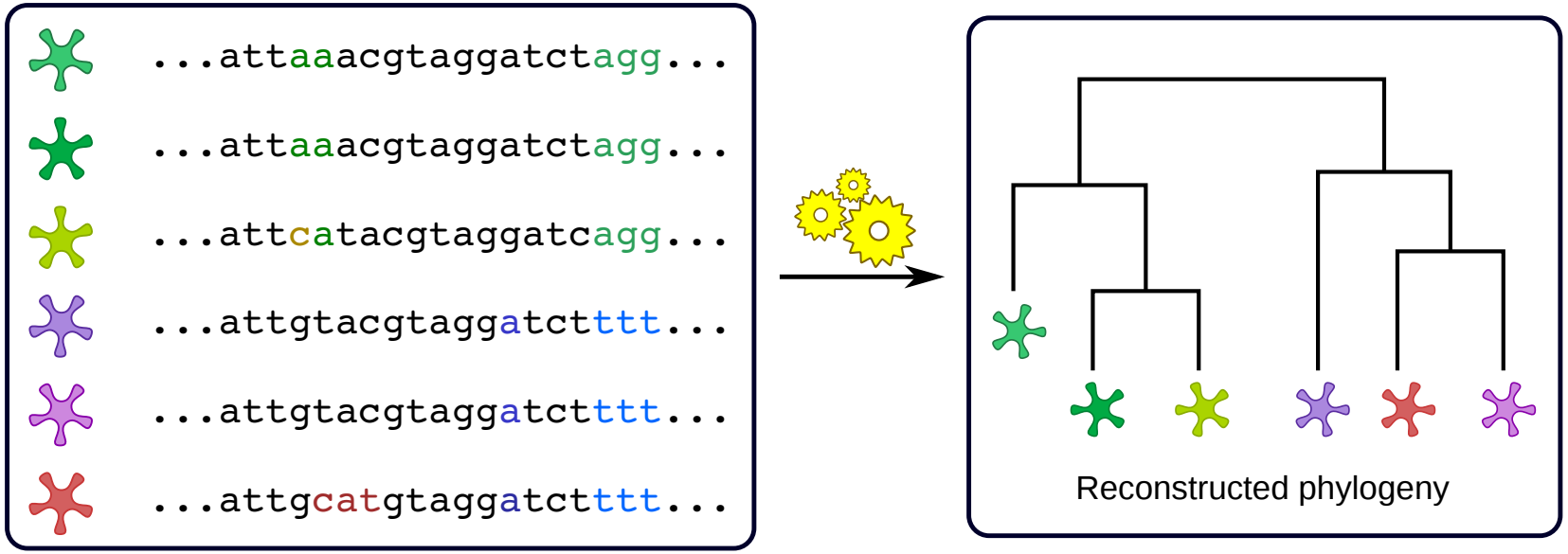

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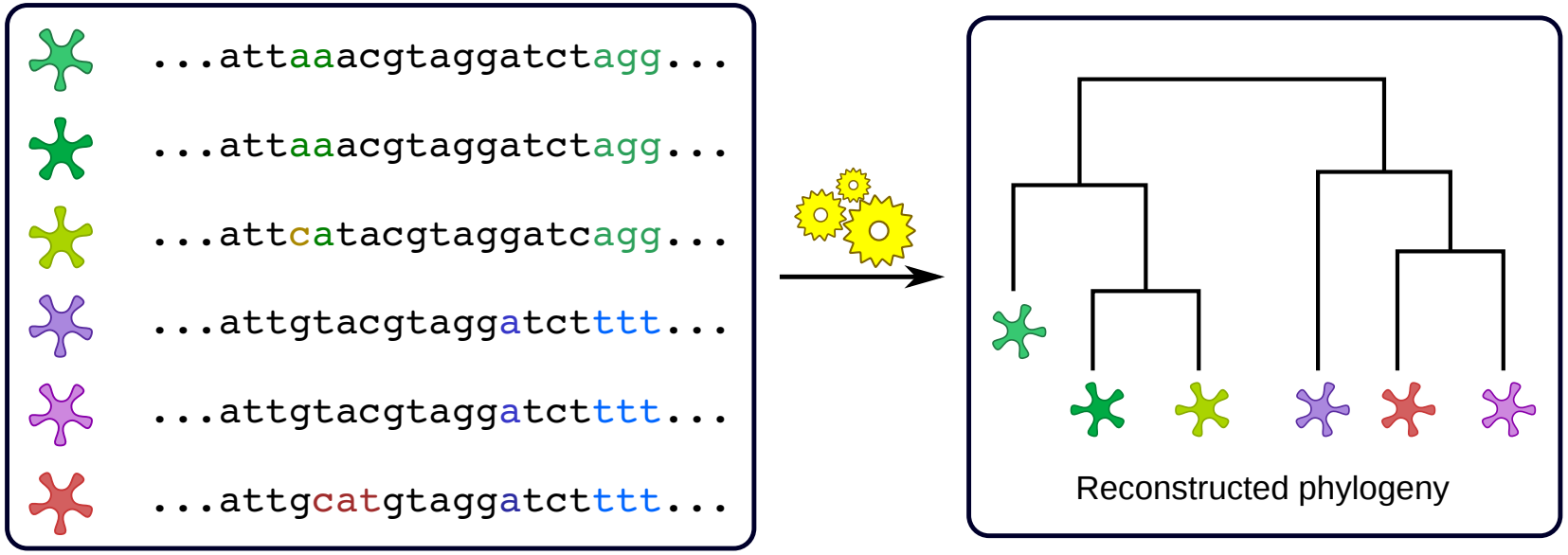


Different methods for achieving phylogenetic reconstruction.

# Workflow

## Prepare data

- align sequences: alignment software + manual refinement

## Build the tree

- distance-based methods
- maximum parsimony
- likelihood-based methods (ML, Bayesian)

## Analyse the tree

- assess uncertainty
- test phylogenetic signal
- model trait evolution
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# Distance-based phylogenetic reconstruction

Approaches relying on **agglomerative clustering** algorithms (e.g. Single linkage, UPGMA, Neighbor-Joining)

## Rationale

1. compute pairwise genetic distances $\mathbf{D}$
2. group closest sequences
3. update $\mathbf{D}$
4. go back to 2) until all sequences are grouped

# Distance-based phylogenetic reconstruction

Alignment

```
...attaaacgtaggatctagg...
...attaaacgtaggatctagg...
...attcatacgtaggatcagg...
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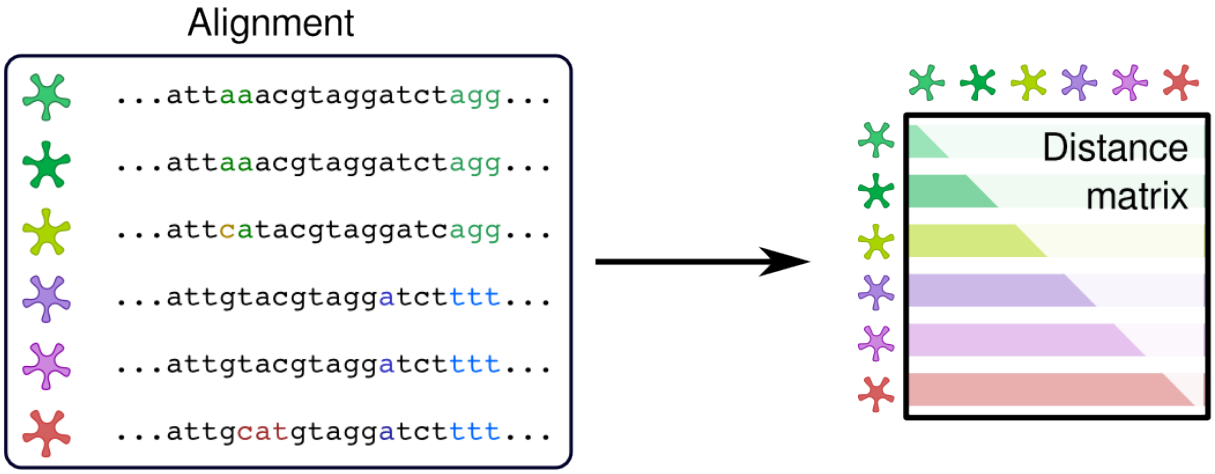

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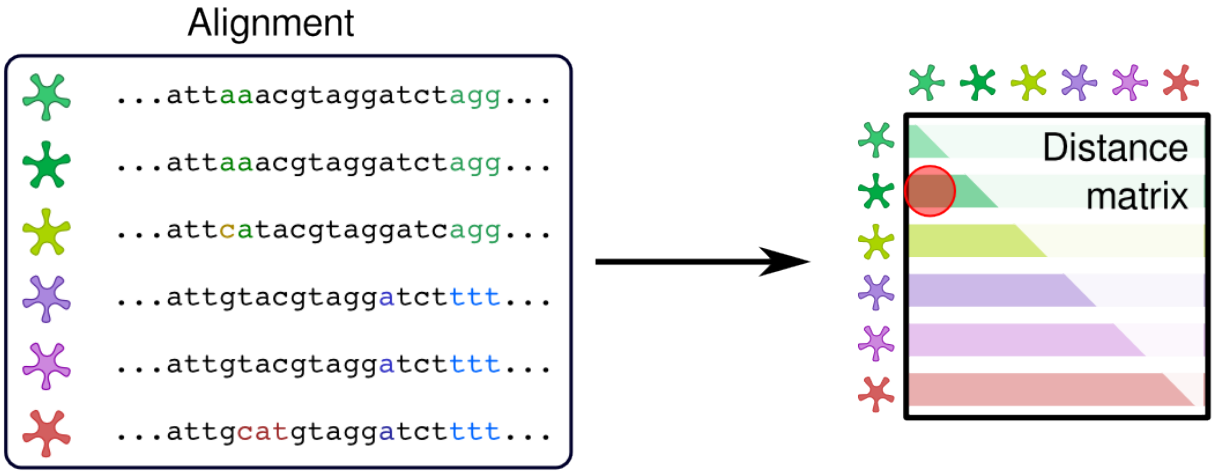


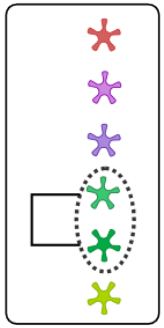

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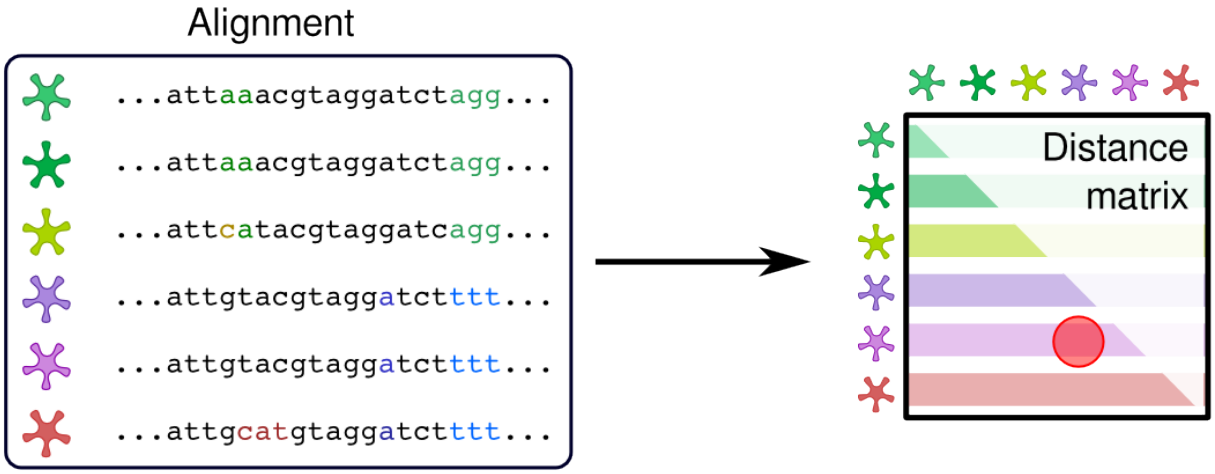


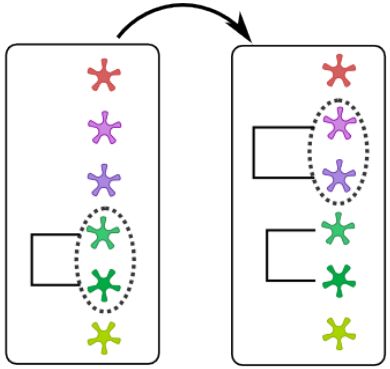

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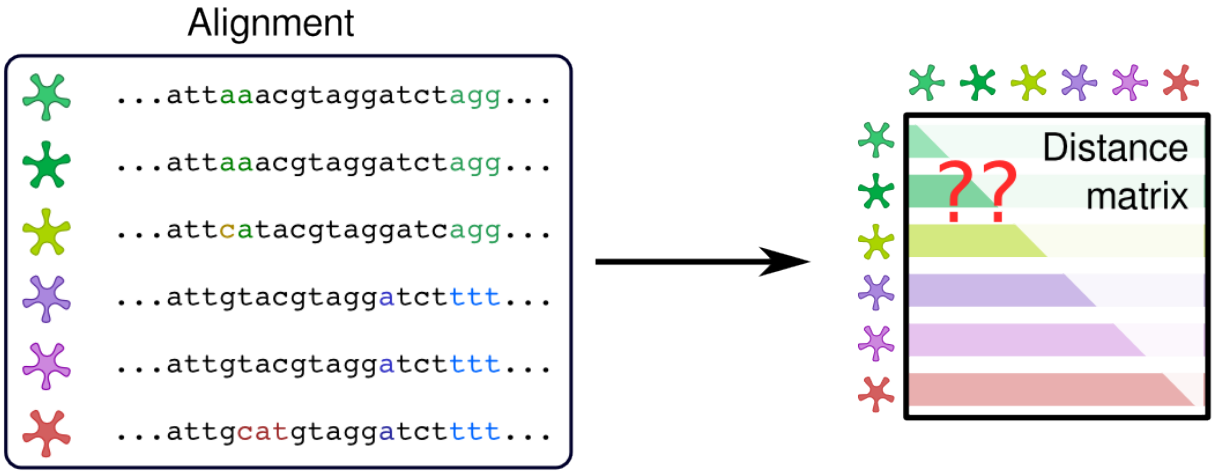


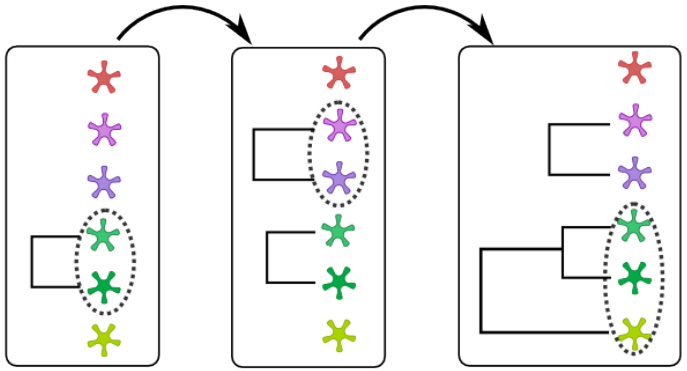

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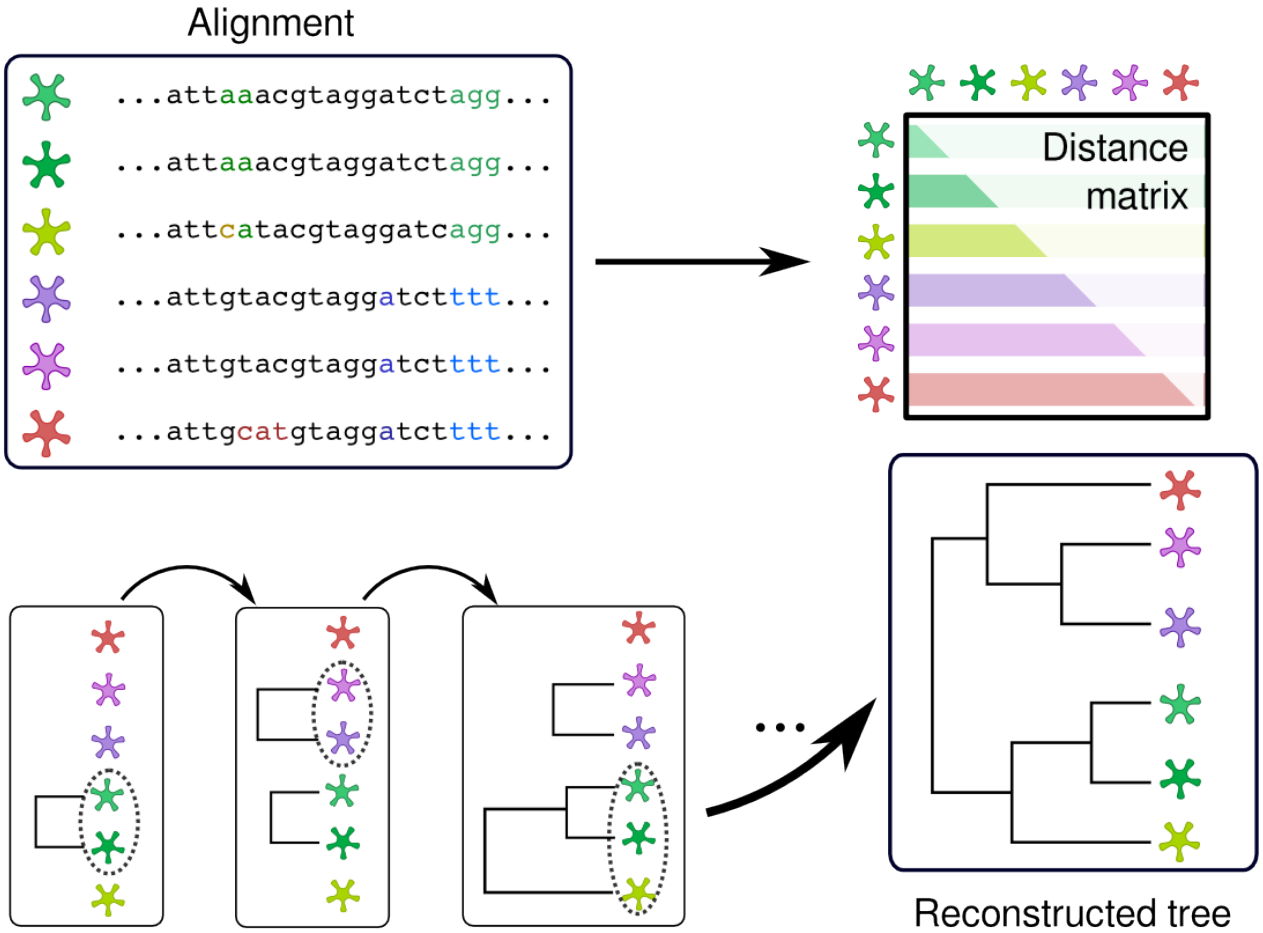
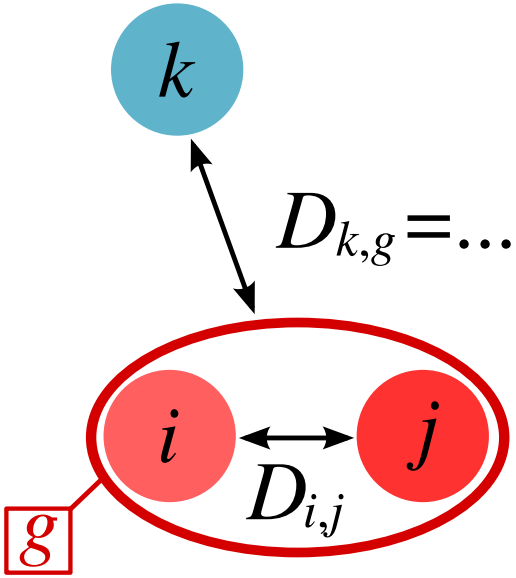


# What is the distance between a node and tips?

**Hierarchical clustering:**




- single linkage: $D_{k,g} = \min(D_{k,i}, D_{k,j})$
- complete linkage: $D_{k,g} = \max(D_{k,i}, D_{k,j})$
- UPGMA: $D_{k,g} = \frac{D_{k,i}+D_{k,j}}{2}$

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Transforms original distances to account for heterogeneous rates of evolution.

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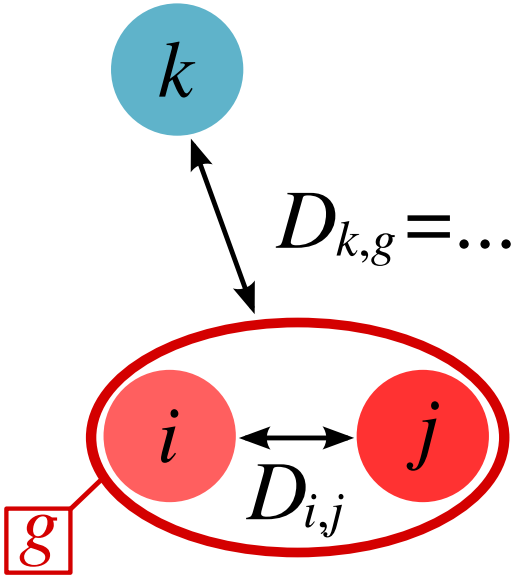


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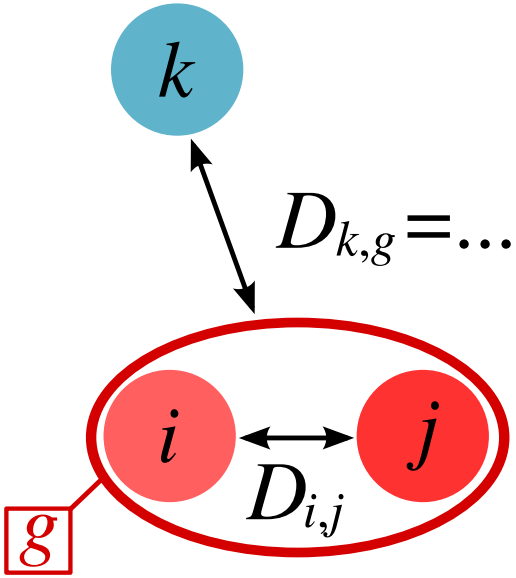


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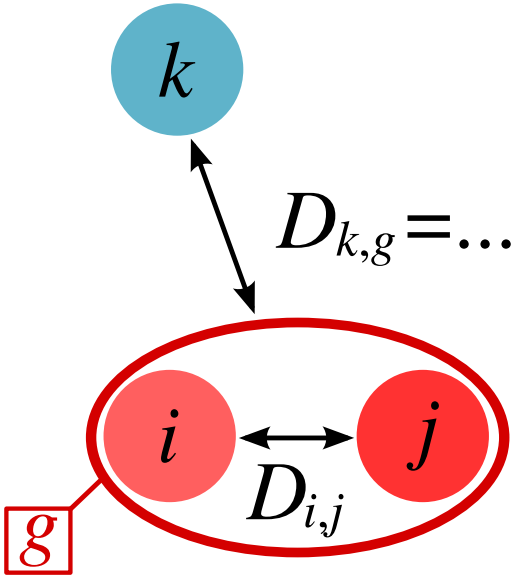


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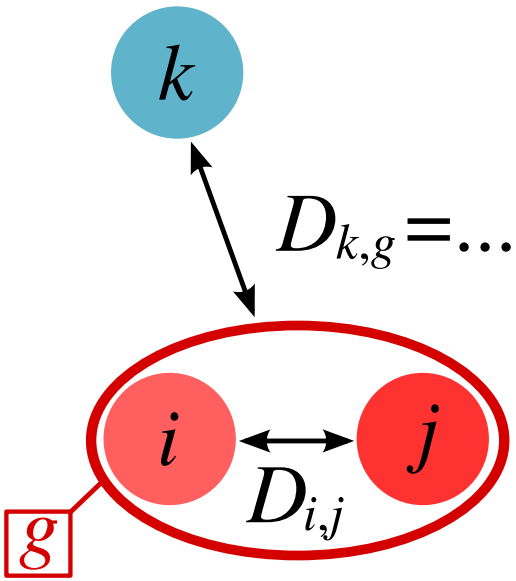


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# Distance-based phylogenetic reconstruction

## Advantages

- simple
- flexible (many distances and clustering algorithms)
- fast and scalable (applicable to large datasets)

## Limitations

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# Maximum parsimony phylogenies

Approaches relying on finding the tree with the **smallest number of character changes (substitutions)**

## Rationale

1. start from a pre-defined tree
2. compute initial parsimony score
3. permute branches and compute parsimony score
4. accept new tree if the parsimony score is improved
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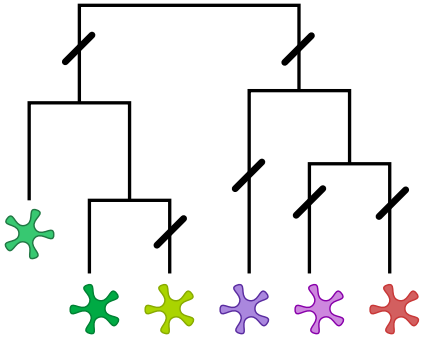
# Maximum parsimony phylogenies



score: 6

Initial tree

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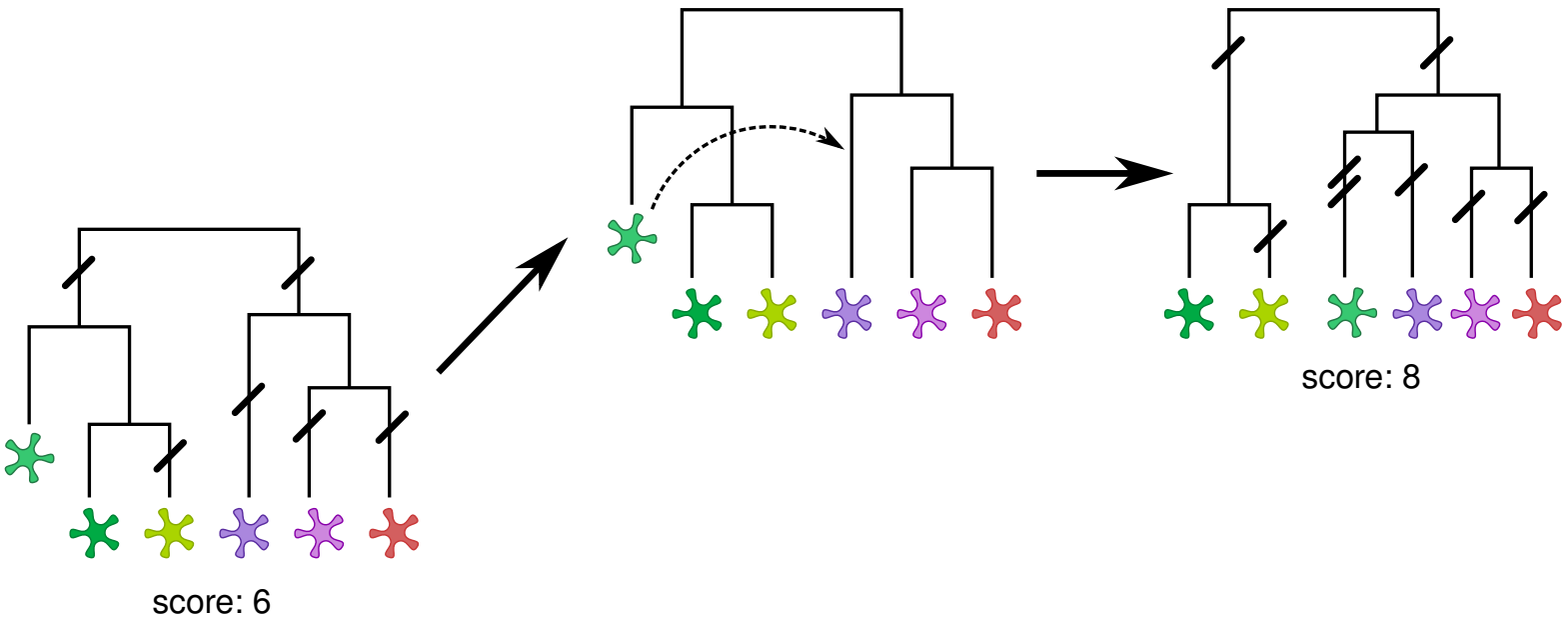


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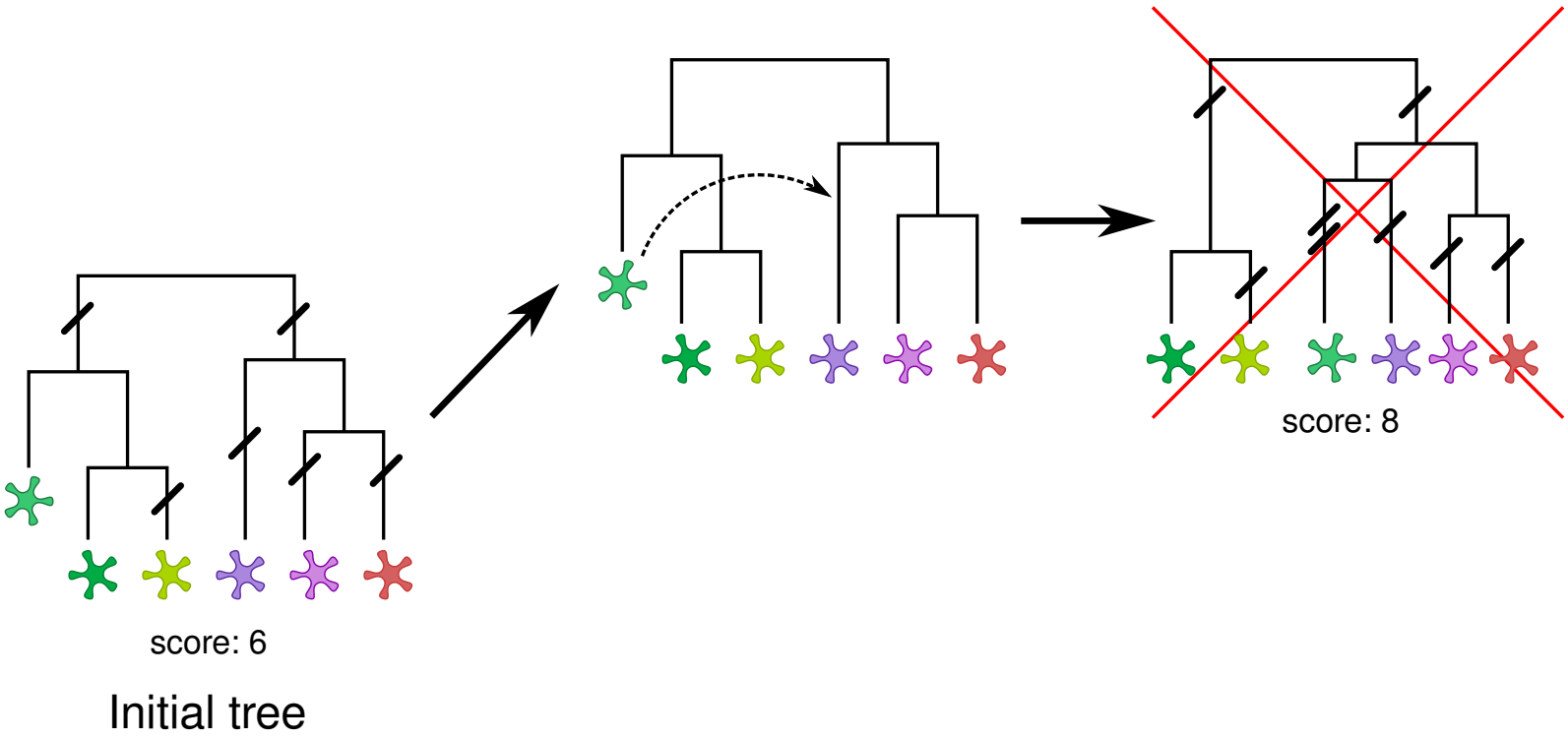


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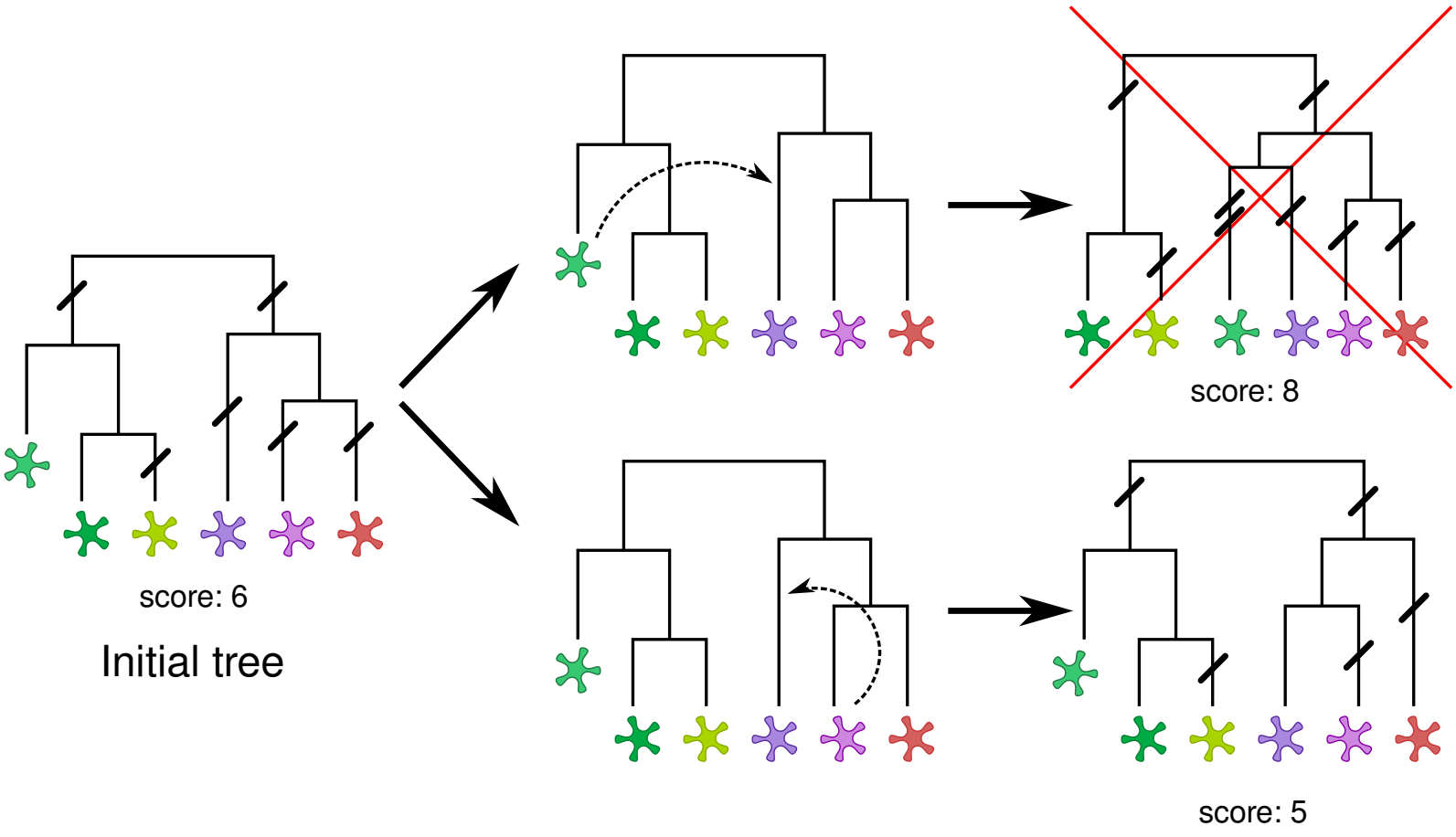

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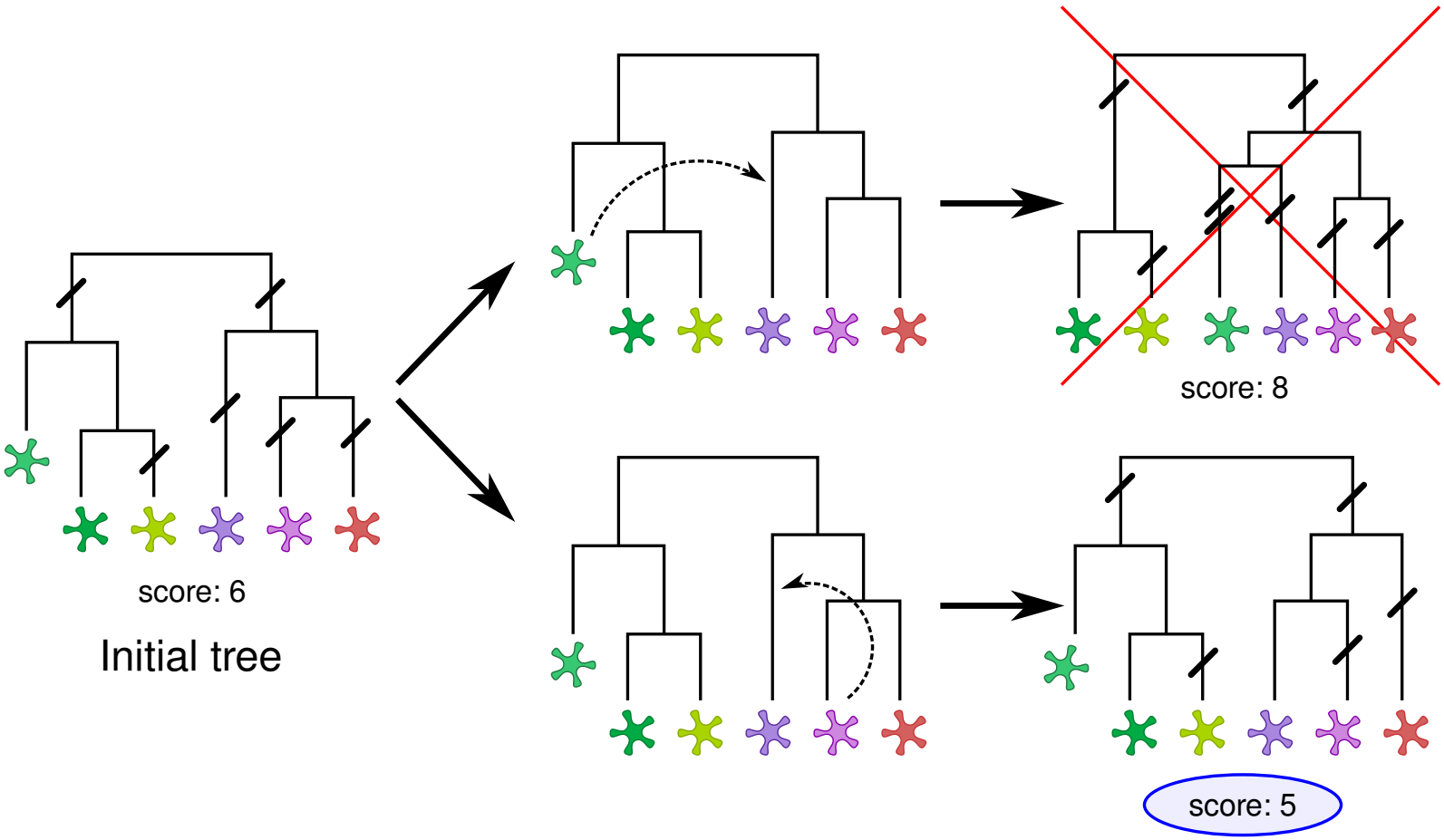

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- applicable to any discontinuous characters (not just DNA)
- intuitive explanation: 'simplest' evolutionary scenario

## Limitations

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- computer-intensive
- different types of substitutions ignored
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Approaches relying on a **model of sequence evolution**:

- **ML**: find tree and evolutionary rates with highest likelihood
- **Bayesian**: find tree and evolutionary rates to posterior probability

## Rationale

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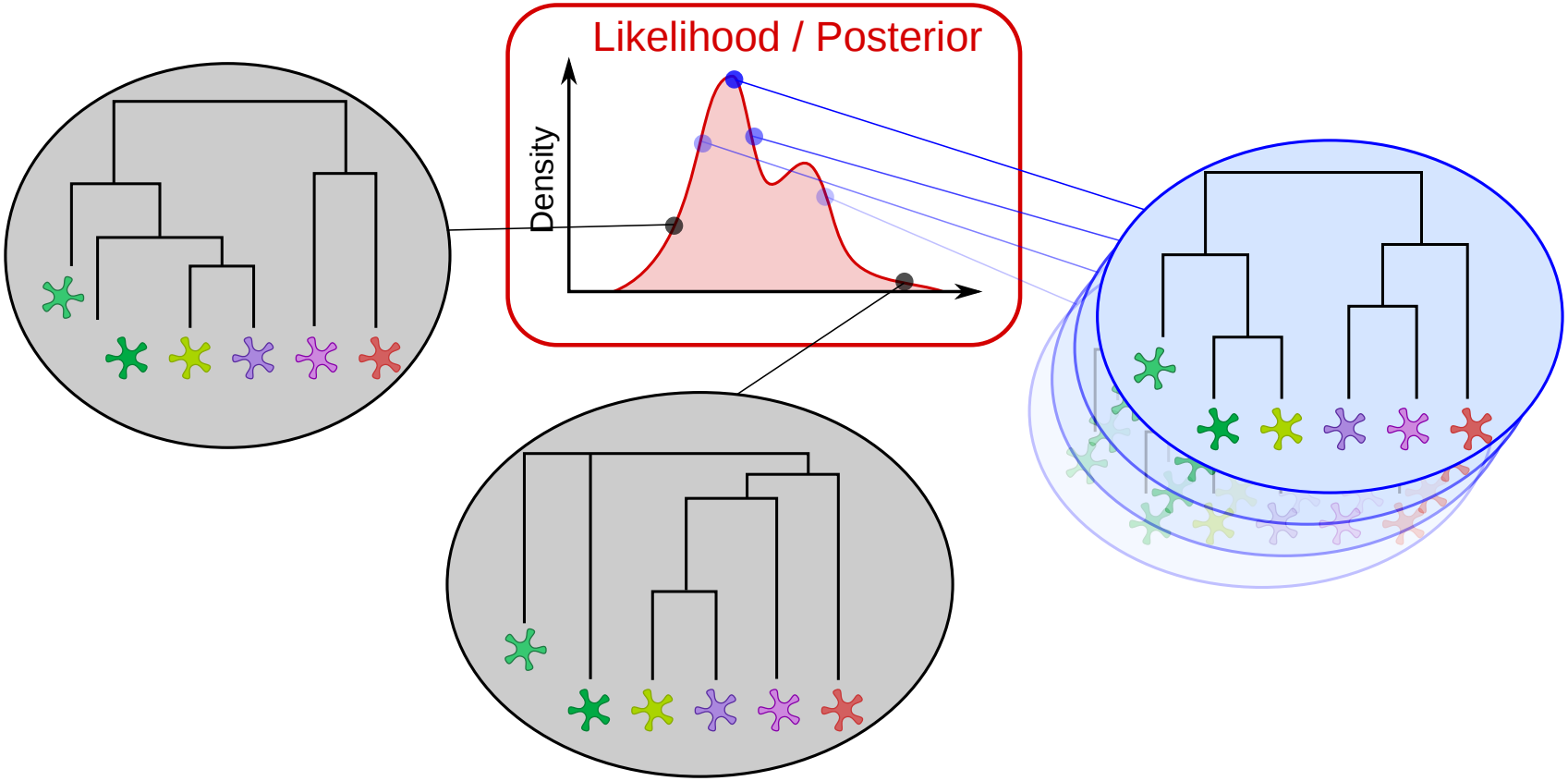

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**Main issue:** assess the uncertainty of the tree topology / individual nodes

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- assess **variability due to sampling the genome** and **conflicting signals**
- relies on analysing **resampled datasets**

## Rationale

1. obtain a reference tree
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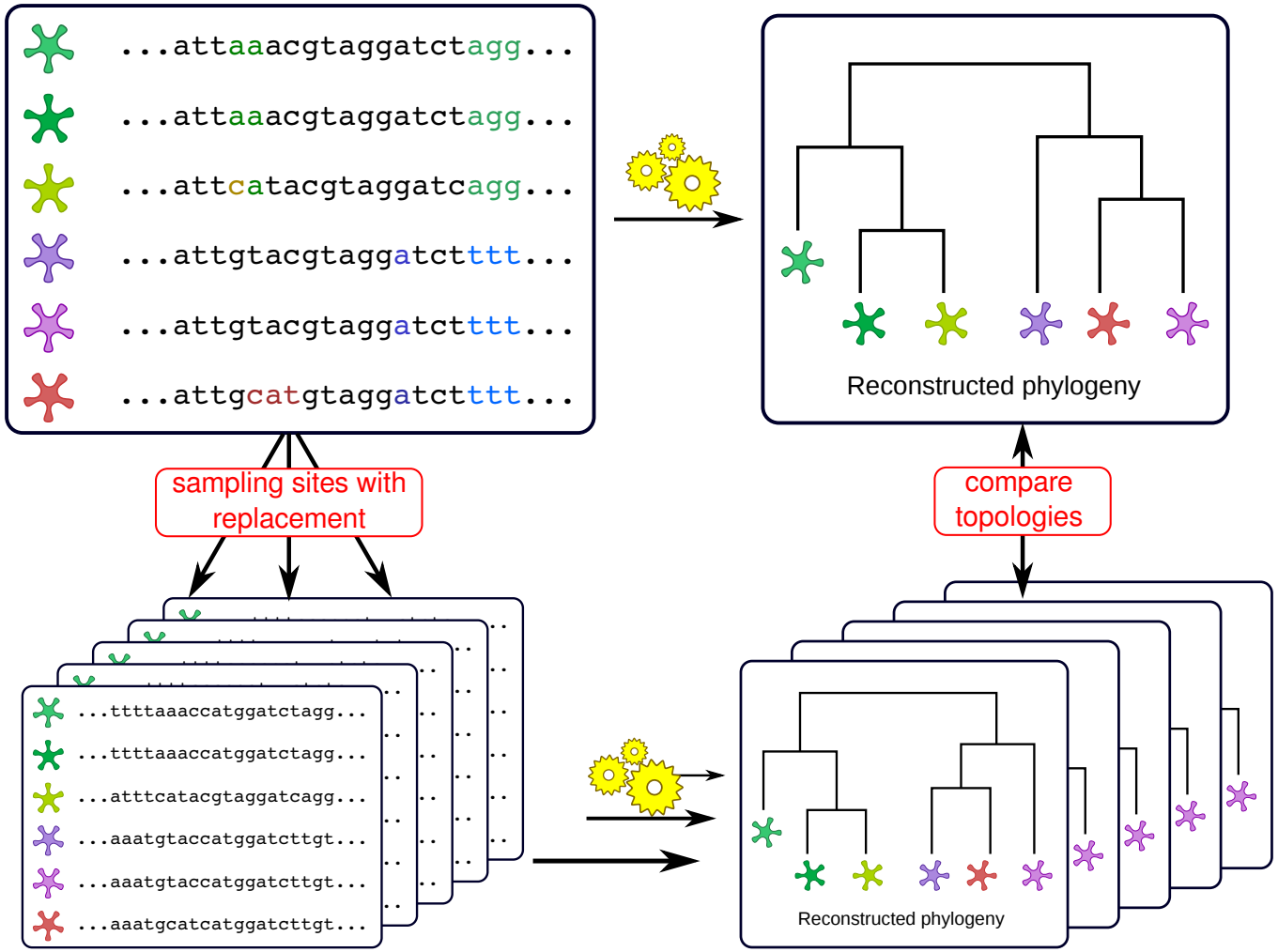

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- model trait evolution (phylogenetic comparative method)
- reconstruct ancestral states
- measure diversity
- infer past demographics/effective population size (coalescence)
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# Time to get your hands dirty!

The pdf of the practical is online:

`http://adegenet.r-forge.r-project.org/`

or

Google → adegenet → documents → "Workshop Leuven, October 2014"