



HAL
open science

Orphan genes: Their identification and evolution in plant-parasitic nematodes

Ercan Seçkin, Dominique Colinet, Marc Bailly-Bechet, Edoardo Sarti, Etienne
Danchin

► **To cite this version:**

Ercan Seçkin, Dominique Colinet, Marc Bailly-Bechet, Edoardo Sarti, Etienne Danchin. Orphan genes: Their identification and evolution in plant-parasitic nematodes. JOBIM 2024 - Journées ouvertes en biologie, informatique, et mathématiques 2024, Jun 2024, Toulouse, France. hal-04615706v2

HAL Id: hal-04615706

<https://hal.science/hal-04615706v2>

Submitted on 19 Jun 2024

HAL is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.



Distributed under a Creative Commons Attribution 4.0 International License

ORPHAN GENES: THEIR IDENTIFICATION AND EVOLUTION IN PLANT PARASITIC NEMATODES



Ercan SEÇKIN*^{1,2}, Dominique COLINET¹, Marc BAILLY-BECHET¹, Edoardo SARTI², Etienne GJ DANCHIN¹

¹ ISA - INRAE PACA, Université Côte d'Azur, CNRS, 400 Route des Chappes, 06903, Sophia Antipolis, France.

² Centre Inria d'Université Côte d'Azur, 2004 Route des Lucioles, 06902, Sophia Antipolis, France.



INTRODUCTION

- **Orphan genes** are genes that have no identifiable homologs. They are systematically found in eukaryotic genomes, and represent up to 30% of the genes [1].
- The **emergence** of such genes is an important mechanism of functional acquisition during evolution. They may originate from duplication or horizontal transfer events followed by **rapid divergence**. Another possibility is that they may emerge from non-genic regions, which is known as **de novo gene birth** [2]. Hence, there is a debate on the real origin of orphan genes [3].
- The most common method for **identifying** orphan genes involves the search for homologues (identification of their absence). Then, their origin can be studied by syntenic approaches. However, these approaches cannot always distinguish between *de novo* genes and the ones that diverged significantly from their ancestor.

OBJECTIVES

- We developed a bioinformatics pipeline to **identify orphan genes** and investigate their **emergence dynamics** and possible **de novo origin**.
- The biological purpose is to apply the built pipeline to the genus **Meloidogyne** of **root-knot nematodes**, which cause a huge global crop loss. The species of this genus secrete parasitism effector, proteins, many of which are encoded by genes having no identifiable homologues [4].

METHOD & RESULTS

PIPELINE

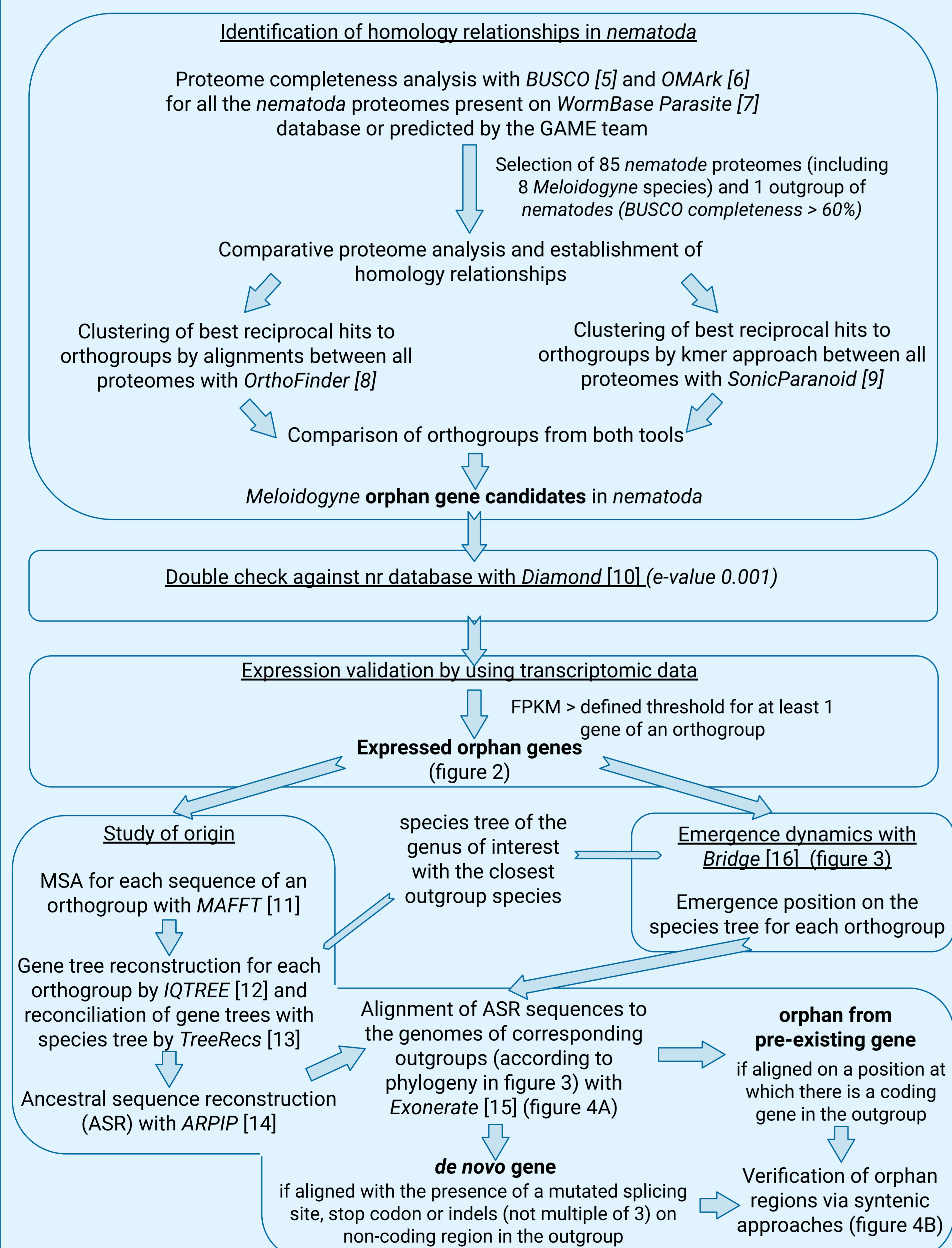


Figure 1: Developed bioinformatics pipeline to identify orphan genes and to study their evolution in Meloidogyne.

RESULTS

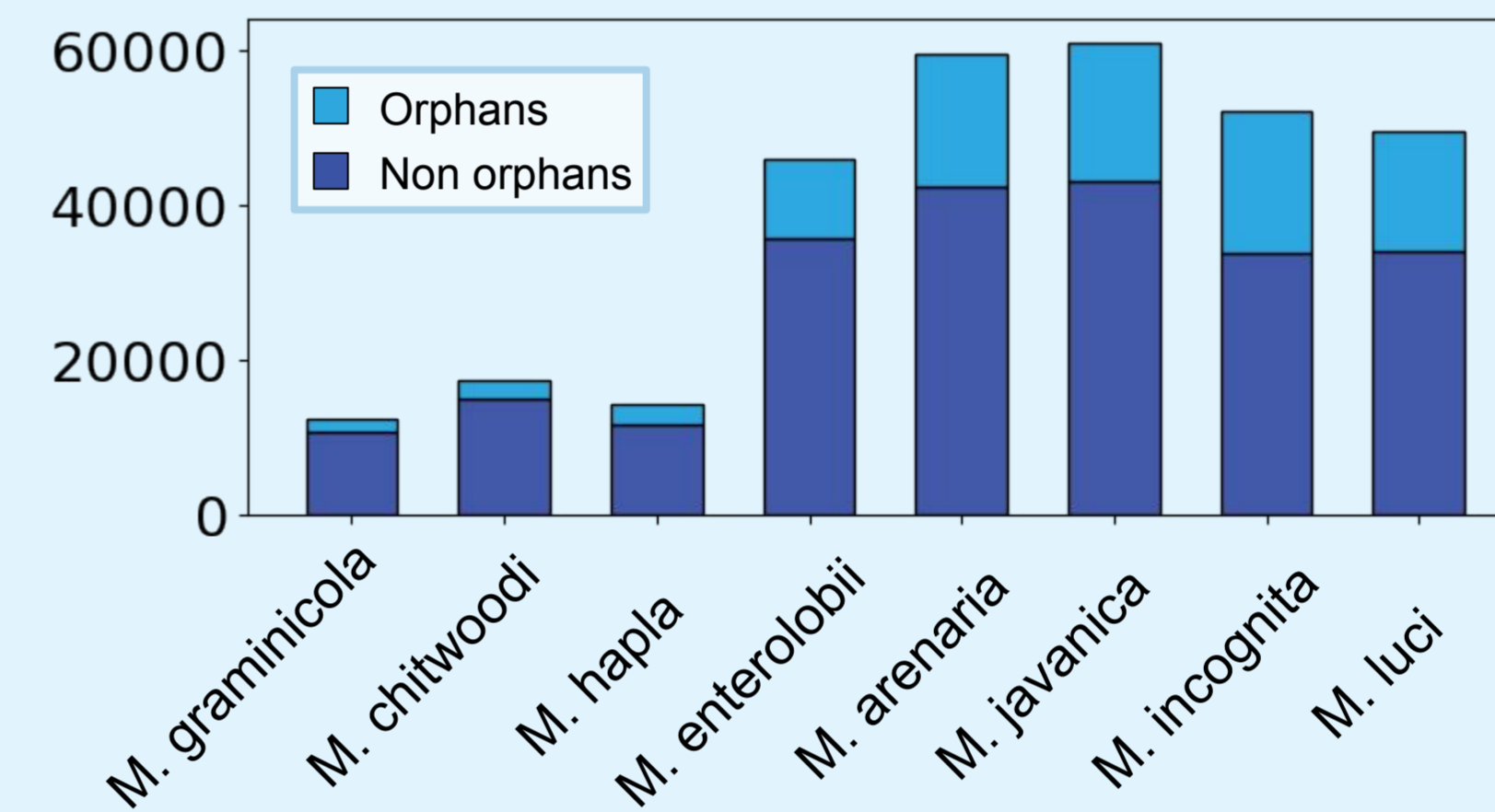


Figure 2: Number of orphan and non-orphan genes for 8 Meloidogyne species studied. Barplot representing the number of orphan genes identified for each of the 8 Meloidogyne species. Each bar corresponds to the total number of genes of each species. Light blue represents orphans and dark blue represents non-orphan genes.



Figure 3: Species tree of Meloidogyne species with their closest outgroup species P. penetrans. Species tree with 8 Meloidogyne species corresponding to three different clades (represented at the right of the species) of Meloidogyne genus along with P. penetrans, which is the closest outgroup to this genus. Each ancestral node is represented by a dot. Number of orphan orthogroups emerged in each node is given by a value next to it.

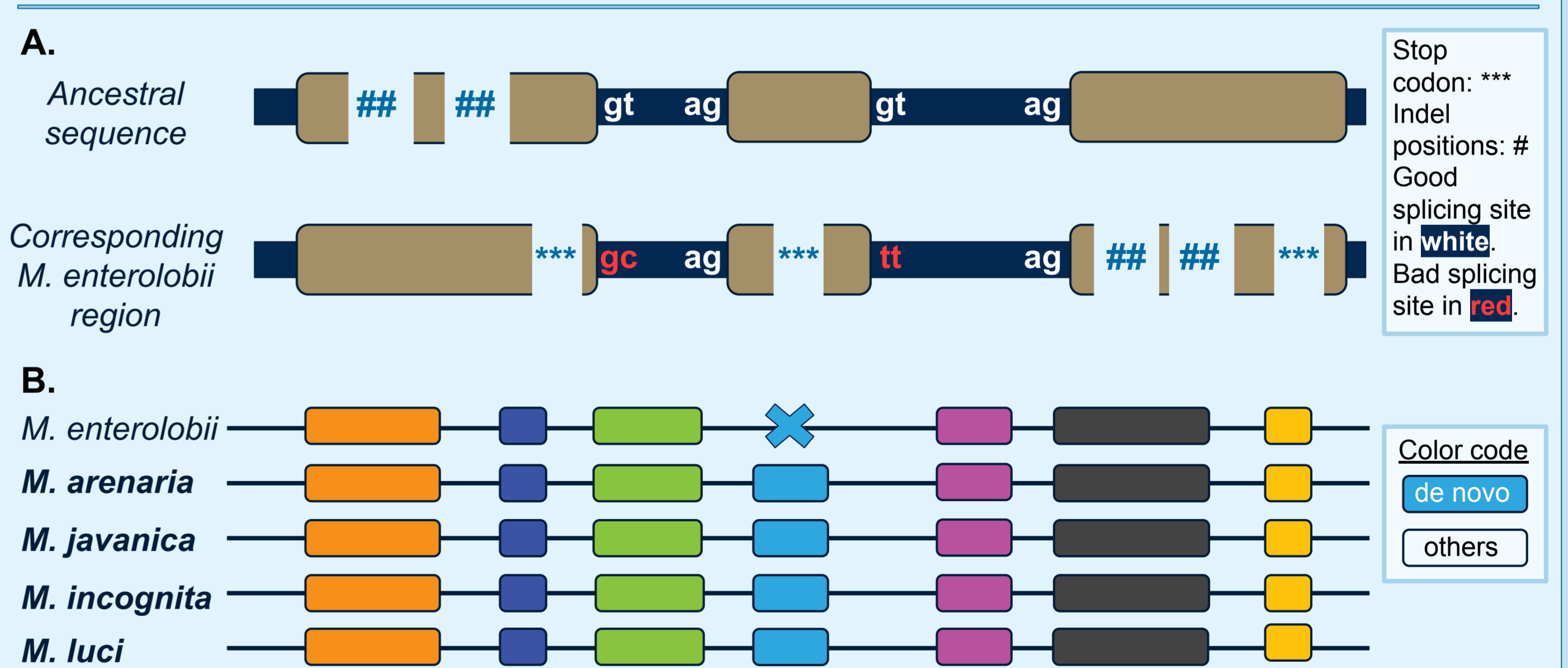


Figure 4: Example of a de novo gene birth that emerged inside the clade I. A. Alignment of the reconstructed ancestral sequence of an example orthogroup common to all members of clade I except for M. enterolobii to the genome of M. enterolobii by Exonerate [15]. Brown represents exons and dark blue represents introns. Stop codons are represented by ***. Insertion-deletion positions (not a multiple of 3) are represented by #. Good splicing sites are in white, mutated ones in red. B. Verification of the syntenic conservation of the identified de novo gene. For the position where ancestral sequence aligns to M. enterolobii, closest 3 genes in both directions are studied to determine the conservation of the region. Lines corresponds to introns and boxes corresponds to genes. Blue boxes corresponds to the de novo gene and blue cross is where it aligns on M. enterolobii. The color code indicates where the same genes align on different species.

CONCLUSION & DISCUSSION

- This study introduces a pipeline that combines homology, transcriptomics, and ancestral sequence reconstruction to identify and investigate the origin of orphan genes. This makes it possible to correctly distinguish between orphan genes that have diverged from pre-existing genes and those that have emerged *de novo*. To date, results indicate that **24%** of *Meloidogyne* genomes consist of **orphan genes**. At least **7%** of these are the result of **de novo gene birth**. Yet, it should be kept in mind that the results depend highly on the quality of genomes in disposition.
- Despite the fact that some effectors in *Meloidogyne* are coded by non-homologous genes, there is still no connection between the orphan genes and their functions in *Meloidogyne*. Thus, we are exploring structural approaches to better characterize these gene-structure-functions associations.

References

1. Tautz D, Domazet-Lošo T. The evolutionary origin of orphan genes. Nat Rev Genet (2011).
2. Weisman CM. The Origins and Functions of De Novo Genes: Against All Odds? J Mol Evol. (2022).
3. Vakirlis, N. et al. Synteny-based analyses indicate that sequence divergence is not the main source of orphan genes. eLife (2020).
4. Mitchum et al., Nematode effector proteins: an emerging paradigm of parasitism, New Phytologist (2013).
5. Simão, F.A. et al. BUSCO: assessing genome assembly and annotation completeness with single-copy orthologs. Bioinformatics (2015).
6. Nevers Y et al. Multifaceted quality assessment of gene repertoire annotation with OMArk. Bioinformatics (2022).
7. https://parasite.wormbase.org/index.html
8. Emms, D.M. et al. OrthoFinder: phylogenetic orthology inference for comparative genomics. Genome Biol. (2019).
9. Cosentino S, Iwasaki W. SonicParanoid2: fast, accurate, and comprehensive orthology inference with machine learning and language models. Bioinformatics (2023).
10. Buchfink et al., Fast and sensitive protein alignment using DIAMOND. Nat. Methods (2015).
11. Katoh, K., and Standley, D.M. MAFFT Multiple Sequence Alignment Software Version 7: Improvements in Performance and Usability. Mol. Biol. Evol. (2017).
12. Minh BQ et al. IQ-TREE 2: New Models and Efficient Methods for Phylogenetic Inference in the Genomic Era. Teeling E, editor. Mol Biol Evol. (2020).
13. Comte N et al. TreeRecs: an integrated phylogenetic tool, from sequences to reconciliations. Bioinformatics (2020).
14. Jowkar G. et al. ARPIP: Ancestral Sequence Reconstruction with Insertions and Deletions under the Poisson Indel Process. Syst Biol. (2023).
15. Slater, G., and Birney, E. Automated generation of heuristics for biological sequence comparison. BMC Bioinformatics (2005).