





# ReporTree: a surveillance-oriented tool to strengthen the linkage between pathogen genetic clusters and epidemiological data

Verónica Mixão

Genomics and Bioinformatics Unit, Department of Infectious Diseases

National Institute of Health Doutor Ricardo Jorge (INSA) – Lisbon, Portugal

"New Tools for Surveillance and Risk Assessment"

March 28<sup>th</sup>, 2023

### **Genomic surveillance**

*"Genomic surveillance is the process of constantly monitoring"* 

pathogens and analyzing their genetic similarities and differences."



A pivotal outcome of genomics surveillance is the identification of pathogen genetic clusters/lineages and their characterization in terms of geotemporal spread or linkage to clinical and demographic data





\*Single-Nucleotide Polymorphisms

Multiple bioinformatics solutions, but similar steps and goals...



\*Single-Nucleotide Polymorphisms

## Identification and characterization of genetic clusters

### The example of *Listeria monocytogenes*...



The detection of genetic clusters with possible public health relevance is still

a challenging step that mostly relies on non-automated and difficult to reproduce approaches

## The routine of genomic surveillance

There is a need for solutions that facilitate, automate and speed-up the

detection of genetic clusters and their linkage to clinical/epidemiological data



### **ReporTree**

- Flexible solution to **automatically identify genetic clusters** at any (or all) distance thresholds (e.g., high resolution thresholds used for <u>outbreak detection</u>)
- Generate surveillance-oriented reports based on the available metadata, such as timespan, geography or vaccination/clinical status
- Allows maintaining cluster nomenclature between runs and generates <u>hierarchical codes for nomenclature</u>

ReporTree: a surveillance-oriented tool to strengthen the linkage between pathogen genetic clusters and epidemiological data

Mixão V, Pinto M, Sobral D, Pasquale AD, Gomes JP, Borges V

Author information **>** 

Preprint from **Research Square**, 30 Sep 2022 DOI: 10.21203/rs.3.rs-1404655/v2 PPR: PPR552299





https://github.com/insapathogenomics/ReporTree





\*Single-Nucleotide Polymorphisms





## Genomic surveillance of L. monocytogenes





#### Main outputs:

- Tree
- Metadata table updated with clusters at any/all resolution levels and nomenclature code
- Summary reports with the statistics/trends for the derived genetic clusters
- Summary reports for the samples of interest (e.g., new samples)

## Identification of potential listeriosis outbreaks

partition	cluster	cluster_ length	samples	source	country	first_seq_ date	last_seq_ date	timespan_ days	ST
MST-4x1.0	cluster_34	2	sample_0419, sample_0464	clinical (50.0%), food (50.0%)	C (50.0%), B (50.0%) (n = 2)	20/06/11	12/03/12	266	87 (100.0%) (n = 2)
MST-7x1.0	cluster_106	33	sample_0017, sample_0037, sample_	clinical (63.6%), food (36.4%)	C (42.4%), B (30.3%), A (27.3%)	30/03/04	14/07/21	6315	87 (100.0%) (n = 33)
MST-14x1.0	cluster_87	46	sample_0017, sample_0037, sample_	clinical (67.4%), food (32.6%)	A (37.0%), C (34.8%), B (28.3%)	30/03/04	14/07/21	6315	87 (100.0%) (n = 46)

## **ReporTree validation and benchmarking**

reporteree TIME-EFFICIENCY

Using diverse datasets with >1,000 isolates of foodborne
 bacterial pathogens, ReporTree identifies genetic clusters
 at potential outbreak level and performs their
 characterization in less than <u>1min</u>



## **ReporTree validation and benchmarking**

report tree

- Using diverse datasets with >1,000 isolates of foodborne bacterial pathogens, ReporTree identifies genetic clusters at potential outbreak level and performs their characterization in less than <u>1min</u>
- For an alignment-based core SNP pipeline, using a diverse dataset with 1788 *Mycobacterium tuberculosis* isolates, the running times is less than <u>6 min</u>



ReporTree can be <u>smoothly implemented in routine surveillance</u>, with negligible computational and time costs, contributing to a <u>sustainable and efficient public health</u> <u>genomics-informed pathogen surveillance</u>

## **Designed for surveillance of multiple pathogens**





- Implemented in the routine surveillance of *L*.
  *monocytogenes* at INSA and has already facilitated the Quality Control Assessments of other <u>foodborne pathogens</u>
- Being tested for application in the genomics surveillance of other important bacterial pathogens, such as <u>Neisseria</u>

#### meningitidis and Mycobacterium spp.

## Take-home message



## ReporTree is an automated and flexible pipeline, applicable to multiple pathogens,

### with a concept aligned with One Health

- ReporTree can be easily implemented in <u>routine surveillance</u> and <u>outbreak investigation</u> workflows, providing solutions to:
  - Obtain genetic clusters at all possible thresholds using multiple clustering methods and maintaining cluster nomenclature between different time-points
  - ✓ Apply a dynamic approach to increase the resolution power in clusters of interest
  - ✓ Obtain summary reports with the statistics/trends for the derived genetic clusters
- Integration in INSaFLU-TELEVIR platform (ongoing)
- Integration in the COHESIVE platform



## **Acknowledgements**

### **Genomics and Bioinformatics Unit**

INSA Alexandra Nunes

Daniel Sobral Joana Isidro João Santos Luís Coelho Miguel Pinto **Rita Ferreira** 

**GENPAT IZSAM** 

Adriano Di Pasquale Nicolas Radomski



Infraestrutura





**Supervisor:** Vítor Borges Team leader: João Paulo Gomes







This work was supported by funding from the European Union's Horizon 2020 Research and Innovation programme under grant agreement No 773830: One Health European Joint Programme.