How to design and deliver pathogen genomics training for health and research professionals

Module 3D
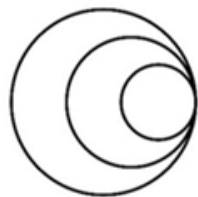
References

1. Bradley, P. *et al.* Rapid antibiotic-resistance predictions from genome sequence data for Staphylococcus aureus and Mycobacterium tuberculosis. *Nature Communications* **6**, 10063 (2015).

2. Köser, C. U. *et al.* Whole-Genome Sequencing for Rapid Susceptibility Testing of M. tuberculosis. *New England Journal of Medicine* **369**, 290–292 (2013).

3. Stoesser, N. *et al.* Predicting antimicrobial susceptibilities for Escherichia coli and Klebsiella pneumoniae isolates using whole genomic sequence data. *Journal of Antimicrobial Chemotherapy* **68**, 2234–2244 (2013).

4. Feldgarden, M. *et al.* AMRFinderPlus and the Reference Gene Catalog facilitate examination of the genomic links among antimicrobial resistance, stress response, and virulence. *Scientific Reports* **11**, 12728 (2021).

wellcome
connecting
science

T3connect

Centre for Genomic
Pathogen Surveillance

References

5. Alcock, B. P. *et al.* CARD 2020: antibiotic resistome surveillance with the comprehensive antibiotic resistance database. *Nucleic Acids Research* **48**, 517–525 (2019).

6. Bortolaia, V. *et al.* ResFinder 4.0 for predictions of phenotypes from genotypes. *Journal of Antimicrobial Chemotherapy* **75**, 3491–3500 (2020).

7. Sherry, N. L. *et al.* An ISO-certified genomics workflow for identification and surveillance of antimicrobial resistance. *Nature Communications* **14**, 60 (2023).

8. Raven, K. E. *et al.* Defining metrics for whole-genome sequence analysis of MRSA in clinical practice. *Microbial Genomics* **6**, (2020).

9. Bogaerts, B. *et al.* Validation strategy of a bioinformatics whole genome sequencing workflow for Shiga toxin-producing Escherichia coli using a reference collection extensively characterized with conventional methods. *Microbial Genomics* **7**, (2021).

References

10. Bogaerts, B. *et al.* A Bioinformatics Whole-Genome Sequencing Workflow for Clinical Mycobacterium tuberculosis Complex Isolate Analysis, Validated Using a Reference Collection Extensively Characterized with Conventional Methods and In Silico Approaches. *Journal of Clinical Microbiology* **59**, (2021).

11. Timme, R. E. *et al.* Optimizing open data to support one health: best practices to ensure interoperability of genomic data from bacterial pathogens. *One Health Outlook* **2**, 20 (2020).

12. Bush, S. J. *et al.* Genomic diversity affects the accuracy of bacterial single-nucleotide polymorphism–calling pipelines. *GigaScience* **9**, 1–21 (2020).

13. Gorrie, C. L. *et al.* Key parameters for genomics-based real-time detection and tracking of multidrug-resistant bacteria: a systematic analysis. *The Lancet Microbe* **2**, e575–e583 (2021).

References

14. Arnold, C. *et al.* Setup, Validation, and Quality Control of a Centralized Whole-Genome-Sequencing Laboratory: Lessons Learned. *Journal of Clinical Microbiology* **56**, 1–8 (2018).

15. Merchant, S., Wood, D. E. & Salzberg, S. L. Unexpected cross-species contamination in genome sequencing projects. *PeerJ* **2**, e675 (2014).

16. Pightling, A. W., Pettengill, J. B., Wang, Y., Rand, H. & Strain, E. Within-species contamination of bacterial whole-genome sequence data has a greater influence on clustering analyses than between-species contamination. *Genome Biology* **20**, 286 (2019).

17. Wood, D. E. & Salzberg, S. L. Kraken: ultrafast metagenomic sequence classification using exact alignments. *Genome Biology* **15**, R46 (2014).

References

18. Low, A. J., Koziol, A. G., Manninger, P. A., Blais, B. & Carrillo, C. D. ConFindr: rapid detection of intraspecies and cross-species contamination in bacterial whole-genome sequence data. *PeerJ* **7**, e6995 (2019).

19. Goig, G. A., Blanco, S., Garcia-Basteiro, A. L. & Comas, I. Contaminant DNA in bacterial sequencing experiments is a major source of false genetic variability. *BMC biology* **18**, 24 (2020).

20. Zhou, P. *et al.* A pneumonia outbreak associated with a new coronavirus of probable bat origin. *Nature* **579**, 270–273 (2020).

21. Charalampous, T. *et al.* Nanopore metagenomics enables rapid clinical diagnosis of bacterial lower respiratory infection. *Nature Biotechnology* **37**, 783–792 (2019).

22. Gauthier, N. P. G. *et al.* Nanopore metagenomic sequencing for detection and characterization of SARS-CoV-2 in clinical samples. *PLOS ONE* **16**, e0259712 (2021).

References

23. Schmidt, K. *et al.* Identification of bacterial pathogens and antimicrobial resistance directly from clinical urines by nanopore-based metagenomic sequencing. *Journal of Antimicrobial Chemotherapy* **72**, 104–114 (2017).

24. Bray, J. E. *et al.* Ribosomal MLST nucleotide identity (rMLST-NI), a rapid bacterial species identification method: application to Klebsiella and Raoultella genomic species validation. *Microbial Genomics* **8**, 1–14 (2022).

25. Lipworth, S. *et al.* SNP-IT tool for identifying subspecies and associated lineages of Mycobacterium tuberculosis complex. *Emerging Infectious Diseases* **25**, 482–488 (2019).

26. Freschi, L. *et al.* Population structure, biogeography and transmissibility of Mycobacterium tuberculosis. *Nature Communications* **12**, 1–11 (2021).

27. Coll, F. *et al.* A robust SNP barcode for typing Mycobacterium tuberculosis complex strains. *Nature Communications* **5**, 4812 (2014).

References

28. Wong, V. K. *et al.* An extended genotyping framework for Salmonella enterica serovar Typhi, the cause of human typhoid. *Nature Communications* **7**, 12827 (2016).

29. Pightling, A. W. *et al.* Interpreting Whole-Genome Sequence Analyses of Foodborne Bacteria for Regulatory Applications and Outbreak Investigations. *Frontiers in Microbiology* **9**, 1–13 (2018).

30. Quick, J. *et al.* Seeking the source of Pseudomonas aeruginosa infections in a recently opened hospital: an observational study using whole-genome sequencing. *BMJ Open* **4**, e006278 (2014).

31. Nikolayevskyy, V. *et al.* Role and value of whole genome sequencing in studying tuberculosis transmission. *Clinical Microbiology and Infection* **25**, 1377–1382 (2019).

32. Hatherell, H.-A. *et al.* Interpreting whole genome sequencing for investigating tuberculosis transmission: a systematic review. *BMC Medicine* **14**, 21 (2016).

wellcome connecting science

T3connect

Centre for Genomic Pathogen Surveillance

References

33. Nikolayevskyy, V., Kranzer, K., Niemann, S. & Drobniewski, F. Whole genome sequencing of Mycobacterium tuberculosis for detection of recent transmission and tracing outbreaks: A systematic review. *Tuberculosis* **98**, 77–85 (2016).

34. Suppli, C. H. *et al.* First outbreak of multidrug-resistant tuberculosis (MDR-TB) in Denmark involving six Danish-born cases. *International Journal of Infectious Diseases* **117**, 258–263 (2022).

35. Coll, F. *et al.* Definition of a genetic relatedness cutoff to exclude recent transmission of meticillin-resistant Staphylococcus aureus: a genomic epidemiology analysis. *The Lancet Microbe* **1**, e328–e335 (2020).

36. Hassoun-Kheir, N. *et al.* Concordance between epidemiological evaluation of probability of transmission and whole genome sequence relatedness among hospitalized patients acquiring Klebsiella pneumoniae carbapenemase-producing Klebsiella pneumoniae. *Clinical Microbiology and Infection* **27**, 468.e1-468.e7 (2021).

References

37. Gröschel, M. I. *et al.* GenTB: A user-friendly genome-based predictor for tuberculosis resistance powered by machine learning. *Genome Medicine* **13**, 138 (2021).

38. Ren, Y. *et al.* Prediction of antimicrobial resistance based on whole-genome sequencing and machine learning. *Bioinformatics* **38**, 325–334 (2022).

39. Břinda, K. *et al.* Rapid inference of antibiotic resistance and susceptibility by genomic neighbour typing. *Nature Microbiology* (2020) doi:10.1038/s41564-019-0656-6.

40. Kohl, T. A. *et al.* MTBseq: A comprehensive pipeline for whole genome sequence analysis of Mycobacterium tuberculosis complex isolates. *PeerJ* **2018**, 1–13 (2018).

41. Hunt, M. *et al.* Antibiotic resistance prediction for Mycobacterium tuberculosis from genome sequence data with Mykrobe. *Wellcome open research* **4**, 191 (2019).

42. Coll, F. *et al.* Rapid determination of anti-tuberculosis drug resistance from whole-genome sequences. *Genome Medicine* **7**, 51 (2015).

References

43. Ellington, M. J. *et al.* The role of whole genome sequencing in antimicrobial susceptibility testing of bacteria: report from the EUCAST Subcommittee. *Clinical Microbiology and Infection* **23**, 2–22 (2017).

44. Pereira, C., Larsson, J., Hjort, K., Elf, J. & Andersson, D. I. The highly dynamic nature of bacterial heteroresistance impairs its clinical detection. *Communications Biology* **4**, 1–12 (2021).

45. Andersson, D. I., Nicoloff, H. & Hjort, K. Mechanisms and clinical relevance of bacterial heteroresistance. *Nature Reviews Microbiology* **17**, 479–496 (2019).

46. Rigouts, L. *et al.* Fluoroquinolone heteroresistance in Mycobacterium tuberculosis: detection by genotypic and phenotypic assays in experimentally mixed populations. *Scientific Reports* **9**, 1–8 (2019).

References

47. Zwe, Y. H. *et al.* Whole genome sequencing (WGS) fails to detect antimicrobial resistance (AMR) from heteroresistant subpopulation of Salmonella enterica. *Food Microbiology* **91**, 103530 (2020).

48. Heyckendorf, J. *et al.* What Is Resistance? Impact of Phenotypic versus Molecular Drug Resistance Testing on Therapy for Multi- and Extensively Drug-Resistant Tuberculosis. *Antimicrobial Agents and Chemotherapy* **62**, e01550-17 (2018).

49. Cooper, A. L. *et al.* Systematic Evaluation of Whole Genome Sequence-Based Predictions of Salmonella Serotype and Antimicrobial Resistance. *Frontiers in Microbiology* **11**, (2020).

50. Crisan, A., McKee, G., Munzner, T. & Gardy, J. L. Evidence-based design and evaluation of a whole genome sequencing clinical report for the reference microbiology laboratory. *PeerJ* **6**, e4218 (2018).

References

51. Stirrup, O. *et al.* Rapid feedback on hospital onset SARS-CoV-2 infections combining epidemiological and sequencing data. *eLife* **10**, (2021).

Resources

- Global Microbial Identifier: https://www.globalmicrobialidentifier.org/about-gmi

- WHO report on "The use of next-generation sequencing technologies for the detection of mutations associated with drug resistance in Mycobacterium tuberculosis complex: technical guide" (https://apps.who.int/iris/handle/10665/274443)

- ISO on Microbiology of the food chain — Whole genome sequencing for typing and genomic characterization of bacteria — General requirements and guidance (https://www.iso.org/standard/75509.html)

- GLASS 2020 report on Whole-genome sequencing for surveillance of antimicrobial resistance (https://apps.who.int/iris/bitstream/handle/10665/334354/9789240011007-eng.pdf)

- Use of Whole Genome Sequencing by the Federal Interagency Collaboration for Genomics for Food and Feed Safety in the United States (https://www.fda.gov/food/science-research-food/whole-genome-sequencing-wgs-program)

Resources

- FIND report on Sequencing for antimicrobial resistance surveillance (https://www.finddx.org/wp-content/uploads/2022/04/20220211_Sequencing-for-AMR-surveillance.pdf)

- PHG foundation report Pathogen Genomics Into Practice (https://www.phgfoundation.org/report/pathogen-genomics-into-practice)

- PHE Expert opinion on whole genome sequencing for public health surveillance (https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/731057/implementing_pathogen_genomics_a_case_study.pdf)

Resources

- UKHSA guidance on "*Mycobacterium tuberculosis* whole-genome sequencing and cluster investigation handbook" (https://www.gov.uk/government/publications/tb-strain-typing-and-cluster-investigation-handbook/mycobacterium-tuberculosis-whole-genome-sequencing-and-cluster-investigation-handbook)

- US CDC "Guide to the Application of Genotyping to Tuberculosis Prevention and Control" (https://www.cdc.gov/tb/programs/genotyping/manual.htm)

Acknowledgements

Creative Commons

This work is licensed under a **Creative Commons Attribution-Share Alike Licence (CC BY-SA 4.0).**

wellcome connecting science

T3connect

Centre for Genomic Pathogen Surveillance

Thank you

T3connect