

Seeing the forest past the trees: A holistic approach to utilizing whole-genome sequencing analyses for cluster detection and outbreak response



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South Carolina Department of Health and Environmental Control (SC DHEC) and the Public Health Laboratory (PHL) worked collaboratively to ensure whole genome sequencing data for all pathogens could be exchanged between the two. They also introduced data analysis and visualization tools for which staff are receiving training to ensure optimal use in support of public health response activities.

The “What”

In August 2022, with funding provided by the Centers for Disease Control and Prevention’s (CDC) Epidemiology and Laboratory for the Prevention and Control of Emerging Infectious Diseases (ELC) Cooperative Agreement, the first genomic epidemiologist was hired at the South Carolina Department of Health & Environmental Control (SC DHEC). The priority for this new position was to pursue the development of mechanisms by which



SC’s whole-genome sequencing (WGS) variable outcomes and associated metadata would be transmitted from the Public Health Laboratory (PHL) to their disease surveillance system, the South Carolina Infectious Disease and Outbreak Reporting Network (SCION).

Through initial conversations with other states about their sequencing programs, SC DHEC found that many states focused on one pathogen of interest, or a subset or group of pathogens (such as foodborne pathogens). Given that WGS is an emerging methodology and pathogen sequencing continues to expand, it was important to set up mechanisms that were (at least mostly) pathogen-agnostic and look at the “highway” rather than the individual “cars.” The goal was to have a pathway for information from PHL to SCION for all the pathogens they sequence. To achieve this, collaboration with their PHL was essential.

To initiate and maintain communication and collaboration, monthly workgroup meetings between pertinent parties in the Division of Acute Disease Epidemiology (DADE) and the PHL began. These meetings have sparked discussions and progress that may not have otherwise occurred beforehand and have helped to bridge the communication gap between DHEC epidemiologists and PHL (concerning more than just WGS).



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Given the environment of teamwork and collaboration, the second priority was to work with the PHL to determine bioinformatics analysis and visualization tools that could be used and easily integrated into the department's workflow. These tools, such as GalaxyTrakr, Pathogen Detection, and MicrobeTrace are user-friendly and are now being used regularly. To expand knowledge and access to WGS data, DHEC will also be providing training on these tools for regional and central office epidemiology staff.

The "So What"

First, utilizing a pathogen-independent approach allows a greater capability to detect new pathogens and define protocols. Prioritizing a pathogen-independent approach allowed SC DHEC to efficiently use resources to develop solutions and protocols across all pathogens.

Second, WGS data provides an additional layer of resolution for surveillance and outbreak investigation. For example, cases of a particular disease that appear to be related based on a common long-term care facility, may have distantly related sequences revealed through WGS, indicating different sources of infection. Alternatively, cases not previously indicated as related may share very similar or identical sequences that would spark further investigation into whether there is epi-linkage to uncover. WGS is an additional useful tool for these scenarios and will continue to evolve as DHEC further articulates how best to use it in public health.

Lastly, WGS surveillance will allow DHEC to understand what sequence types or variants are regularly circulating in the South Carolina population, and to detect new sequence types or variants more rapidly as they appear.

The "Now What"

Ongoing training of regional epidemiologists is essential to harness the capabilities of the WGS. Since these epidemiologists are the ones at the front lines of outbreak response, empowering them with the skills to visualize, interpret, and utilize WGS data will ensure the timeliest use of these data for public health action.

Another important aspect is maintaining communication from the PHL, bioinformaticians, and genomic epidemiologist to the regional epidemiologists who will need support while integrating these new data in their investigations. Continually providing expertise and protocols as WGS data is used more and more is crucial.

Next, the epidemiologists will be testing and implementing the newly developed pathway for transmitting WGS data and metadata from the public

health lab to SCION, followed by integrating edits based on assessments of accessibility, functionality, and utility of the solution from subject matter experts. SC DHEC plans to expand the use of their Data Warehouse to incorporate all diseases captured in SCION. This would allow for easier queries of the data, clearer detection of commonalities, and ease of analysis using tools such as SAS, Python or R.

The epidemiologists will also continue to explore the use of additional bioinformatics tools to include exploring Linux-based tools for analyses at DHEC. Alternatively, more and more graphical user interface (GUI) tools are being developed and marketed as open source, so they will continue to test and implement these as well. Lastly, they plan to add to the list of pathogens PHL sequences and develop protocols for use of the data generated by WGS.

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