

For immediate release
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Urgent action needed to make the most of genomics in the fight against infectious disease

A new report from the PHG Foundation reveals the potential for genomics to help the health service in the fight against infectious diseases – but warns of barriers that could delay the health benefits for patients.

Pathogen Genomics Into Practice examines how technological developments in rapid, lower-cost genome sequencing could soon improve hospital infection control, the treatment of patients with infections (including in some cases those resistant to antibiotics), and surveillance and management of new infectious disease threats.

Based on extensive research and stakeholder consultation, supported by funding from the National Institute for Health Research (NIHR) Cambridge Biomedical Research Centre, the report examines both the ground-breaking science of pathogen genomics and the current health policy landscape surrounding use by Public Health England and the NHS. It also highlights case studies where British scientists and doctors are already using genomics to more accurately and rapidly analyse the causes of disease outbreaks and inform responses.

However, potential barriers to the prompt, consistent and efficient use of genomics for infectious disease detection and control at a national level are identified, and we urgently recommend improved data sharing between health service laboratories, and clear strategic leadership and coordinated action from Public Health England.

The report provides a policy roadmap for decision-makers, setting out the necessary actions to ensure that citizens and patients benefit promptly from scientific advances and government investment in infectious disease genomics. This includes a proposed catalyst comprising structures to integrate and amplify current pathogen genomics services, and make them available across the country.

PHG Foundation Director Dr Hilary Burton commented: *“We have heard many examples of how cutting-edge genomics is being used to guide treatment in infectious diseases and control disease outbreaks in hospitals and the community, but we also warn that the full clinical benefits of this excellence will not be achieved without appropriate oversight of the research, implementation and regulatory agenda by the Department of Health. Our roadmap sets out the way ahead to ensure we optimise the use of pathogen genomics for patients and the wider population”.*

Professor Dame Sally Davies, Chief Medical Officer, said: *“I have championed the importance of pathogen sequencing, especially in the battle against Anti-Microbial Resistance. That is why work on pathogens is an integral part of the 100,000*

Genomes Project. This timely report from the PHG Foundation provides an insight into the importance of this work. I welcome it and think it is an excellent contribution to the future debate”.

Professor Tim Peto of the University of Oxford Nuffield School of Medicine, a consultant in infectious diseases and a member of the external expert steering group for the project said: *“Pathogen genomics represents a major advance. This timely and comprehensive review of the evidence and systems shows clearly the need for national leadership, coordination and an effective data sharing strategy to maximise the benefits from sequencing in controlling infection”.*

The PHG Foundation report – now freely available to download - was received in advance by the organisations charged with the national management of infectious diseases (Public Health England, NHS England and the Department of Health).

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Notes for editors

- **The PHG Foundation is an independent, non-profit health policy think-tank based in Cambridge, UK.**
- Our mission is **making science work for health** – promoting the fair and effective translation of the best 21st century scientific innovations into improved medical and public health policy and practice. We have long-standing expertise in the clinical implementation of genomics and other innovative technologies.
- Visit our website at: www.phgfoundation.org
- We are a campus associate of Cambridge University Health Partners (CUHP) www.cuhp.org.uk
- This work was supported by funding from the National Institute for Health Research (NIHR) Cambridge Biomedical Research Centre, a partnership between the University of Cambridge and Cambridge University Hospitals Foundation Trust. They receive substantial levels of funding from the NIHR to translate fundamental biomedical research into clinical research that benefits patients and improves healthcare provision. www.cambridge-brc.org.uk
- The National Institute for Health Research provides the framework through which the research staff and research infrastructure of the NHS in England is positioned, maintained and managed as a national research facility. The NIHR provides the NHS with the support and infrastructure it needs to conduct first-class research funded by the Government and its partners alongside high-quality patient care, education and training. Its aim is to support outstanding individuals (both leaders and collaborators), working in world class facilities (both NHS and university), conducting leading edge research focused on the needs of patients. www.nihr.ac.uk

Press queries

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CAMBRIDGE UNIVERSITY
Health Partners

Knowledge-based healthcare

Genomics in the fight against infectious disease

Selected recent case studies of practical applications of pathogen genomics

Note: these examples are the published work of UK research groups wholly independent from the PHG Foundation

1. *E. coli* food poisoning outbreak from contaminated watercress

Public Health England (PHE), 2015



When food poisoning outbreaks occur, it can be challenging to pinpoint the source as cases can be spread over a wide geographical area. This study used a variety of techniques including whole genome sequencing to investigate a complex outbreak of food poisoning caused by *E. coli* O157. Scientists from PHE found this approach was vital in allowing them to demonstrate that two simultaneous outbreaks took place, rather than one single outbreak. The genome sequence of samples from the first outbreak didn't match any known UK strains, so the PHE team suspected that these cases were imported. The second outbreak was concluded to have been caused by water contamination at a watercress farm, probably from cattle kept in a neighbouring field.

Reference: Jenkins C *et al.* Public Health Investigation of Two Outbreaks of Shiga Toxin-Producing *Escherichia coli* O157 Associated with Consumption of Watercress. *Appl Environ Microbiol.* 2015; 81(12): 3946-52.

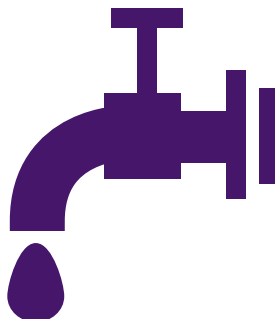
2. *Pseudomonas aeruginosa* outbreak on a burns unit

Queen Elizabeth Hospital Birmingham, 2014

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The bacterium *Pseudomonas aeruginosa* is a common cause of hospital acquired infections, and burns patients are one group that are vulnerable. The bacteria thrive in moist conditions such as water systems. Whole genome sequencing was used to determine the source of *P. aeruginosa* from colonised patients on a burns ward in a Birmingham hospital. The resolution provided by genome sequencing allowed the research team to accurately determine the source of the bacteria. Two cases were found to have been imported into the hospital. The source of the bacteria that originated in the hospital was found to be living in biofilms in sink plumbing. To eradicate this source, parts of the plumbing system were removed and deep cleaned or replaced.

Reference: 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.* 2014; 4(11): e006278.

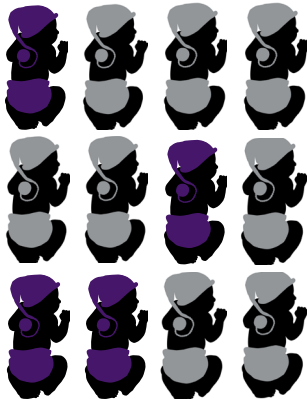
3. MRSA outbreak in a special care baby unit

Cambridge University Hospitals NHS Foundation Trust / Wellcome Trust Sanger Institute, 2013

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Routine surveillance of ill infants on the special care baby unit of a Cambridge hospital led healthcare staff to suspect the presence of an MRSA outbreak. Deep cleaning of the unit was performed alongside genome sequencing and analysis to determine the extent of the outbreak. This revealed that the outbreak was larger than previously thought, including earlier clusters of cases not linked by epidemiological analysis. Further cases after the deep clean prompted screening of staff members; genomic analysis revealed that one was an asymptomatic carrier, unwittingly spreading the infection. They were treated to remove the MRSA, and no further cases occurred in the babies on the unit.

Reference: Harris SR *et al.* Whole-genome sequencing for analysis of an outbreak of methicillin-resistant *Staphylococcus aureus*: a descriptive study. *Lancet Infect Dis.* 2013; 13(2): 130-6.

4. Sequencing used in real-time during a Salmonella outbreak

University of Birmingham, 2015

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This study was the first to use whole genome sequencing to analyse *Salmonella* isolates during an outbreak, rather than afterwards. A hospital outbreak with some cases also present in the community was investigated; such outbreaks require a rapid response to halt the spread of infection but finding the source can be difficult. Genome sequencing helped to distinguish between closely related bacteria (indistinguishable using other techniques) allowing them to monitor the outbreak and how cases related to different sources of *Salmonella*, and to focus infection control measures appropriately.

The researchers developed a new rapid sequencing protocol for a MiSeq sequencer, and also tested a portable handheld sequencer called the MinION. Both gave the team clinically useful information within hours, compared with days for standard microbiological analysis. Infections were shown to have come from many sources: the outbreak strain was circulating in the community and introduced into the hospital from there. This same strain was responsible for a large community outbreak across the UK and Europe, and genome sequencing helped trace its origins back to a single German egg producer.

Reference: Quick J *et al.* Rapid draft sequencing and real-time nanopore sequencing in a hospital outbreak of *Salmonella*. *Genome Biology* 2015; 16: 114.