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Leading the way: driving the delivery of pathogen genomics into practice

Pathogen genomics has the potential to enable clinicians and public health practitioners to revolutionise the way in which they treat and prevent the spread of infectious diseases. However, a number of barriers exist to prompt and effective implementation across the health service with its complex landscape of delivery and commissioning organisations. In particular, there is an urgent need for strategic planning and coordination of service development and delivery across organisations.

Current microbiological and molecular methods have limitations which impact on their ability to deliver the timely and detailed information required to effectively manage many cases of infectious disease. Recent advances in the field of genomics offer the opportunity to revolutionise the management of infectious disease by providing an unprecedented level of detailed information about the pathogens responsible.

There are a number of active research projects attempting to increase our understanding of infectious disease pathology with the ultimate aim of improving diagnosis, treatment and infection control practice. In addition, there are major initiatives underway that are aiming to translate pathogen genomics into practice in clinical settings within the UK. Some of the most promising potential applications are in the management of tuberculosis, where sequencing for identification and prediction of drug resistance is being piloted, and tackling MRSA where genomics can be used to identify the source of hospital outbreaks. These collaborative projects aim to provide evidence of the clinical utility of pathogen genomics. However, consideration must be given to how the results of these efforts are implemented in the wider health service for the benefit of patients.

The autonomous and competitive nature of the delivery of microbiology laboratory services to the NHS in England and PHE can undermine the effectiveness of implementation.

The effective configuration of infectious disease clinical and laboratory services will be crucial to realising the anticipated benefits that genomics has to offer.

How can successful implementation be achieved across the health service?

While the sequencing technology and bioinformatic analysis underpinning the use of pathogen genomics in the management of infectious disease are relatively well established, the integration of these new techniques into the delivery of medical and public health microbiology services is only in its infancy.

The effective configuration of infectious disease clinical and laboratory services will be crucial to realising the anticipated benefits that genomics has to offer.

This will require a number of practical steps such as developing the evidence base, workforce and best practice guidelines as well as policies to support effective and complete implementation across the NHS. Underpinning these steps is the need to recognise and tackle some of the systemic barriers to progress described below.

Critical barriers to effective implementation within the health service

Through extensive research and interaction with a wide variety of experts in different organisations working in the field of infectious disease we have identified a number of key barriers to the widespread and effective adoption of pathogen genomics.

Gaps in the knowledge base

Introduction of pathogen genomics will be disruptive in technological terms by enabling replacement of existing tests, and in organisational terms by putting complex molecular analysis within reach of a greater number of laboratories.

Although a number of projects are now generating considerable amounts of pathogen genomic data, the challenge of effectively aggregating and sharing this data at a national level remains to be overcome. This situation, exacerbated by limited co-ordination of activity across service organisations and research groups generating this data, will hinder progress towards effective utilisation of pathogen genomics by preventing the development of consistent and validated genomics services, as well as limiting access by a wider group of clinical services to potentially useful information.

In addition, there is insufficient effort being made to build an evidence base with which to evaluate the clinical utility of pathogen genomics in 'real world' microbiology practice. Both of these practical steps are necessary for facilitating decision making about the introduction of pathogen genomics into routine clinical microbiology services.

Lack of evidence of cost-effectiveness

Deciding how, when and where it is appropriate to introduce pathogen genomics into microbiology services also requires evaluation of the extent of any cost-savings made in the health service through reducing mortality, morbidity and associated costs of managing infectious disease. Currently, there is a lack of evidence in this area.

Some groups have, however, indicated that sufficient cost savings can be made at the level of the laboratory assay itself to justify introduction of genomics in place of more expensive phenotypic typing assays.

Fragmentation of the microbiology network

The autonomous and competitive nature of the delivery of microbiology laboratory services to the NHS in England and PHE can undermine the effectiveness of implementation. Their current configuration does not incentivise the sharing of relevant knowledge and experience developed as new technologies are introduced. This slows progress towards establishing best practice and results in duplication of effort and cost for no additional patient benefit.

The distributed nature of the translational research and service development currently being undertaken in the field of pathogen genomics has led to the proliferation of different approaches to the sequencing and interpretation of genomic data. It has also supported the development of multiple routes to clinical implementation. Whilst such variety enables the competiveness and innovation that can drive development of novel solutions to problems, heterogeneity of approach is unlikely to be conducive to effective, rapid and co-ordinated service implementation.

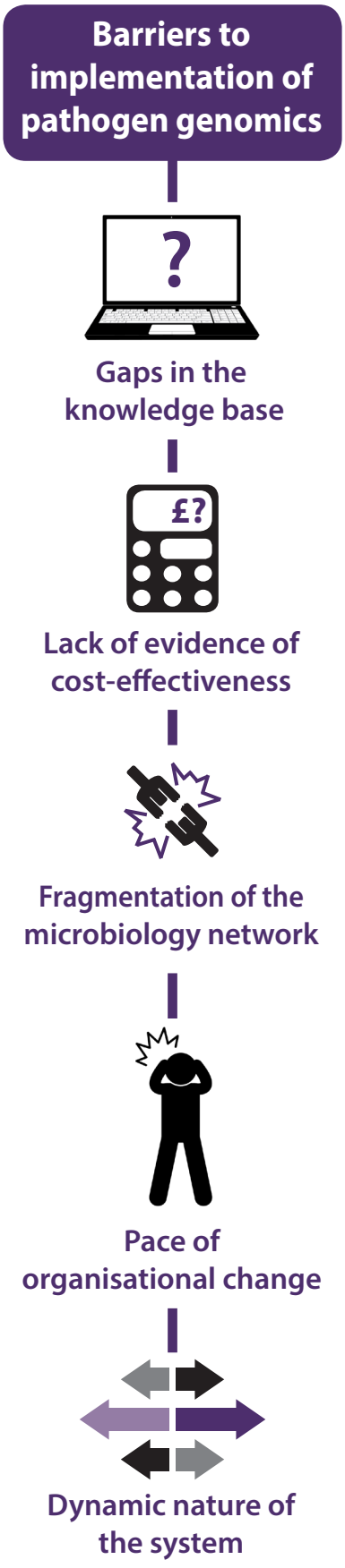
Pace of organisational change

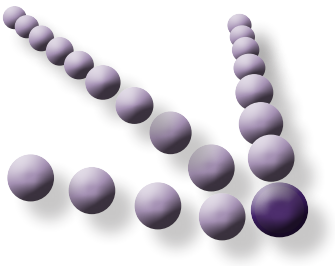
Introduction of pathogen genomics will be disruptive in technological terms by enabling replacement of existing tests and in organisational terms by putting complex molecular analysis within reach of a greater number of laboratories. The perception that this disruption threatens the integrity and continuity of existing service models will need to be addressed, otherwise there may be resistance to the prompt and effective adoption of these new tests.

Dynamic nature of the system

The continually evolving nature of both the underlying genomic science and the system into which it is being implemented creates difficulties for adoption. The field of genomics is developing rapidly both technologically and scientifically. Furthermore, the health system is in constant flux with changes being made to laboratory structures as well as service provision.

The pace of adoption of pathogen genomics will rely on the ability of the health system to adapt and to embrace an evolving field of science in a timely and effective manner.





The need for strategic coordination

The emergence and spread of infectious diseases can often be unpredictable, requiring management that crosses local, national and even international boundaries. It is particularly important, therefore, that measures to control these diseases continue to be undertaken in a flexible, collaborative and cooperative way between the diverse services and individuals involved. This includes clinical scientists, medical and nursing professionals, public health specialists, industry and government.

To support implementation of pathogen genomics there is a pressing need for stronger leadership and coordination to be developed between the centres of excellence currently at the forefront of genomics service development and delivery, both in relation to knowledge development and exchange and in relation to the development of processes that can facilitate decision making and implementation (*e.g.* best practice guidelines, regulation of data, quality assurance systems and so on).

The effective introduction of pathogen genomics will be hampered unless this deficit is rapidly addressed by policy makers and practitioners.

The way forward

None of the barriers described above are insurmountable. but overcoming them will require a greater emphasis on strategic coordination across the complex landscape of organisations involved in delivering the promise of pathogen genomics.

In particular, there is an opportunity for the early-adopters in pathogen genomics, both in academia and the health services, to move towards an explicitly co-operative model in which genomic knowledge, evidence of effectiveness and best practice are developed in a co-ordinated fashion. This would in turn enable the presentation of a unified and consistent view of the practices and policies required for implementation of this exciting new technology, allowing leaders in the health service to ensure that the anticipated benefits, for both infectious disease services and the patients they serve, actually occur.

The PHG Foundation will publish a full report and future policy recommendations for infectious disease genomics in the UK early in 2015.

Widespread adoption of any new technology and scaling up of the programme requires leadership and coordination in order to ensure that best practices are appropriately adopted nationwide.

For more information, visit:

www.phgfoundation.org/project/id