Pathogen Genomics Into Practice

Recommendations

Recommendation no.	Recommendation	Report section
1	PHE will need to work with all microbiology service providers, both public and private sector, to ensure that they participate fully in meeting requirements to contribute to national infectious disease surveillance, through appropriate contributions to the implementation and development of pathogen genomics services.	11.2
2	Agreement needs to be reached between PHE and NHSE with regards to funding for service development and delivery where the pathogen genomics services have a dual clinical and public health benefit.	11.2
3	The initial implementation of pathogen genomics services should be focused in laboratories providing consolidated microbiology services, as these are most likely to be able to realise necessary economies of scale and to achieve the concentrations of expertise and efficient data management required.	11.2
4	A defined pathway, encompassing test referral mechanisms, sequencing, analysis and interpretation must be developed for each pathogen and each application of genomics. Implementation of these pathways will require a coordinated approach.	13.2
5	Robust and effective prioritisation processes will need to be developed for new service developments. These must be informed by consultation including frontline end user groups.	13.2
6	The location of sequencing and analysis services should not be pre-determined, and a mixed model should be allowed to develop which makes optimal use of available resources and takes account of local / national demand for genomics: variables include the cost, throughput achievable at each location, and turnaround time.	13.4
7	All laboratories providing clinical pathogen genomics services need to be accredited to the appropriate national / international standards.	14.2
8	Evaluation and comparison of test performance should span the whole process from sample extraction to clinical report, encompassing assessments of both analytical and clinical validity and clinical utility.	14.2
9	The clinical and public health microbiology 'community' needs to work with UKAS and NEQAS to establish standards that can be used to develop appropriate accreditation processes.	14.2
10	In order to ensure that services are of sufficiently high quality, and delivered in a consistent manner, guidelines (equivalent to SMIs) establishing minimum standards for pathogen genomics services must be developed.	14.2
11	Develop a national collaborative network of pathogen genomic service providers to share knowledge and best practice, collaborate on service and methodology development and agree standards for clinical and public health service delivery.	14.2

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12	Realisation of the strategic public health benefits of the implementation of pathogen genomics services will require coordinated action amongst providers and users to develop underpinning policies and procedures to support co-operation and inter-operation of services. These efforts should be led by Public Health England but be explicitly supported by all relevant health service and policy making organisations.	14.3
13	Criteria must be established to decide under what circumstances sequenced pathogen isolates (or related clinical materials) must be stored for future public health use, timescales for any storage requirements and sources of funding to ensure sustainability of any sample archives created.	14.3
14	Additional investment will be required, above that envisaged for the development of individual pathogen genomics services, to build the infrastructure and capacity required to realise the broader and longer term public health benefits of the implementation of pathogen genomics for disease surveillance, treatment and prevention.	14.3
15	Existing links between the infectious disease aspects of animal and human health services should be exploited and strengthened to ensure that synergies in the developments of their genomics programmes are realised and a 'One Health' approach to managing infectious disease threats can be developed where appropriate.	14.4
16	Organisations leading on the development and delivery of pathogen genomics in the UK should work with and show leadership within transnational organisations and specific international genomics focused initiatives to ensure that best practice is shared and sufficiently standardised, or at least interoperable datasets are developed and regulatory barriers to effective genomic and metadata exchange are addressed.	14.4
17	When considering data release to a publicly accessible database, stakeholders should adopt proportionate safeguards that balance the need to protect the interests of data subjects, particularly relating to privacy and confidentiality, against the likely benefits of proceeding with data sharing.	15.6
18	Raw genomic data and minimal metadata ought to be shared as widely as possible (following appropriate QC and assuming public release is approved) preferably through public data repositories to ensure long term sustainability.	15.6
19	Criteria for defining what minimal data sets are appropriate for release to publicly accessible databases should be developed, with risk assessments being undertaken to identify in particular which elements of metadata can be released publicly for each pathogen. PHE (and their Office of Data Release) would be best placed to deliver on this, along with NHS input.	15.6
20	It must be mandatory for all providers of NHS or PHE pathogen genomic investigations to make sequence data and all other necessary clinical and epidemiological data available for use by legitimate NHS healthcare and public health professionals within agreed timeframes, for the purpose of delivering their stipulated functions. A mandate needs to be implemented urgently to prevent data that is currently being generated from being lost in silos.	15.8

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21	The benefits of data collation and risks of not aggregating data should be articulated to those being mandated to submit data. A feedback or reward strategy should be developed to gain longer term accord with and practical support for a data sharing mandate, and investment made in adequate infrastructure to enable data deposition at the practical level.	15.8
22	All pathogen genomic data and associated metadata required by healthcare and public health professionals to maximise the effectiveness of their management of infectious disease in individual patients and populations should be submitted to the designated database without delay.	15.10
23	Where data release into the public domain is envisaged / considered, a strategy for the timing of genomic data and limited metadata release that takes into account a balance between the need to serve wider public health benefit and the rights of individuals and organisations, should be devised. Provision should be made for access by researchers, companies, and healthcare and public health professionals outside the UK.	15.10
24	A public health authority such as PHE should be responsible for the collation and storage of all genomic data and metadata for the purposes of clinical and public health service delivery, and to support the development of new clinical and public health applications of genomics in the early stages of implementation until solutions can be developed in collaboration with databases such as ENA to provide access to the necessary storage and expertise to build and maintain an optimal sharing system in the longer term.	15.14
25	Accessible interfaces or software tools must be developed that meet the needs of clinical users by enabling straightforward access to the information in genomic and metadata databases and to facilitate the ability of legitimate users to perform analyses on underlying data.	16.2
26	Pathogen genomics service providers will need to invest in developing and maintaining, or procuring remote access to, sufficient computational capacity to enable their data analysis.	16.3
27	PHE and HEE should continue to work together to ensure that education and training are provided to support the development of the bioinformatics workforce and the analytical and interpretive skills of frontline users of pathogen genomics services.	16.3
28	Additional investment to increase the availability of bioinformaticians able to develop and deliver pathogen genome analytical services will be required, at least in the short term, until analytical tools operable by the existing laboratory and clinical workforce are developed.	16.4
29	A PHE led strategy for the organisation of access to computational infrastructure and bioinformatics expertise will be required to ensure access to genome analysis services is not an impediment to the implementation of genomics services.	16.4

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30	Agreement is required on the standards for genomic data quality and format across laboratories undertaking pathogen genomic analysis for clinical and public health investigations. There should also be mechanisms for standardising descriptive clinical and epidemiological information relating to genomic data to maximise the interoperability, and therefore the utility, of data collected across different locations.	17.2
31	Mechanisms need to be developed by relevant professional groups for benchmarking the performance of equivalent genome analysis methods, and for ensuring that methods used in service settings meet minimum standards.	17.3
32	In order to support greater interoperability of data generated across the health system there should be mechanisms (preferably international) established for standardising nomenclature for genomic characteristics of pathogens and their relatedness.	17.4
33	Curation of each organism specific genotype-phenotype database and analytical pipeline, and archiving of isolate / tissue collections must be under the control of a designated responsible authority. Each authority should operate with PHE oversight and funding to support their sustainability.	17.5
34	The challenges of integrating clinical and genomic data, enabling data interoperation and delivering user friendly service requisitioning and reporting interfaces across different LIMS and IT systems need to be addressed. This will require agreement on data management standards between all organisations involved in delivering or using pathogen genomics services.	17.5
35	Evidence for clinical and public health utility and cost effectiveness will need to be clearly demonstrated prior to funding and adoption of pathogen specific genomics services by clinical and public health end-users.	18.4