

BOARD PAPER - NHS ENGLAND

Title: NHS England Genomics Strategy Board

Clearance: Sir Bruce Keogh, National Medical Director
Tim Kelsey, National Director for Patients and Information

Purpose of paper:

- Update the NHS England Board on the progress and outcomes from the NHS England Genomics Strategy Board and the genetics lab reconfiguration.

Key issues and recommendations:

- Recommendation: Responsibility for coordination of the NHS England contribution to the 100k genomics programme will reside in the business improvement and research division of the medical directorate.
- Recommendation: Alignment of the genomics agenda with NHS England.
- Recommendation: Given the imperative around data ownership and transparency, the patients and information directorate will represent NHS England on the Board of Genomics England Limited.
- Recommendation: NHS England is responsible for service commissioning elements and will establish a formal service reconfiguration steering group as part of the medical genetics Clinical Reference Group. It will structure to oversee the reconfiguration project using the governance of the clinical priorities advisory group and direct commissioning committee to secure timely delivery. Responsibility for delivering the Human Genome Strategy Group (HGSG) actions of the reconfiguration of existing genetic testing services will reside in the medical directorate of NHS England with clinical specialised services working alongside the operations directorate.

Actions required by Board Members:

- Approve NHS England's work to date supporting the Prime Minister's ambition on genomics to deliver the procurement of 100K genomes.
- Note the progress of outcomes from the NHS England Genomics Strategy Board.
- Approve the establishment of a project to determine the options and deliver rationalisation of the genetic testing facilities in England by the national specialised commissioning team.

NHS England Genomics Strategy Board

Executive Summary

1. The Prime Minister announced on 10 December that the Government was to achieve a paradigm shift in the way that genomics is used in the NHS and thereby contribute to economic growth.
2. The Genomics Strategy Board, hosted by NHS England and chaired by Sir Malcolm Grant, was tasked with providing guidance on the delivery of the Prime Minister's ambition on genomics to deliver the procurement of 100K genomes.
3. The Board oversaw a number of workstreams intended to inform plans to deliver on project:
 - Procurement and intellectual property;
 - Benefits realisation;
 - Clinical, public and media engagement; and
 - NHS staff training and education.
4. This work was also supported by a number of workstreams commissioned by the Department of Health (DH) chief medical officer, which were approved by the Secretary of State for Health in March and reported to the Board in April 2013.
 - Scientific considerations
 - Research data storage
 - Consent and ethics
5. The Board has made good progress since the announcement on 10 December and a range of work has now been completed. Key outcomes are outlined in this paper.
6. Prior to the establishment of the 100k Genome Project, the DH established the Human Genome Strategy Group (HGSG). Over a number of months the HGSG made a series of recommendations, some of which were referred to NHS England to consider. The most pertinent is the rationalisation and reconfiguration of existing genetic testing services to ensure they are fit for purpose in the future.
7. The report by the HGSG, 'Building on our inheritance - genomic technology in healthcare' has challenged the NHS to consider significant modernisation and development of genetic testing services. NHS England specialised services proposes progressing the recommendation of putting in place a network consisting of genomic technology centres, biomedical diagnostic hubs and regional genetics centres. NHS England is the direct commissioner for sedical genetics services and has published the first national specification <http://www.england.nhs.uk/npc-crg/group-e/e01/> .
8. The advancement of laboratory technology means that the service may need to undergo substantial change with a smaller number of testing facilities providing a broader range of genetic analysis. The NHS England Board are asked to approve the launch of a project to examine the issues, establish a case for change and put in place a procurement to deliver.

Background

9. Whole genome sequencing technology has taken massive steps forward in recent years and is relatively straightforward today with an entire sequence of an individual's coding DNA (exome) being possible to produce in 7-10 days (this includes assembling the sequence but not any analysis). A full genome sequence for an individual (coding and non-coding DNA) being possible to sequence and assemble in a matter of weeks.
10. The UK continues to be well placed with its integrated genome project to claim leadership in application of genomic medicine in a health system. However, it should note the substantial infrastructure investments that are being made in the US and China, which might give them an advantage in the longer term if they are not matched.
11. On 10 December, the Prime Minister announced that the government will lead the implementation work to achieve a paradigm shift in the way that genomics is used in the NHS and thereby contribute to economic growth. To provide patients in the NHS with the benefits from the innovation associated with genomics the government proposed that:
 - 100,000 whole genomes of patients in the NHS will be sequenced in the next 5 years to support their clinical care in the next 3-5 years and drive research to support wealth creation.
 - The NHS, working with partners, (such as Health and Social Care Information Centre, Public Health England, National Institute for Health Research and research councils) will ensure that genomic information can be linked to relevant clinical data (emerging initially but not exclusively from our major cancer centres) to support a better understanding of clinical impact of genetic variation and to measure outcomes more precisely.
 - Establish a skills and training programme for the NHS workforce building on plans that the health education England (HEE) and Birmingham are developing on wider genetics education to realise this vision of genomic medicine.
 - The NHS, DH and the Department of Business, Innovations and Skills will develop a shared plan for engagement with clinicians, patients and the public about the value, rationale and benefits of the genomics strategy
12. The Genomics Strategy Board, hosted by NHS England and chaired by Sir Malcolm Grant, was tasked with providing guidance on the delivery of the Prime Minister's ambition on genomics to deliver the procurement of 100K genomes. The Genomics Strategy Board has met six times and concluded its work on 28 June 2013, with recommendations transferred to the newly established Genomics England Limited.
13. Prior to the establishment of the 100k genome project, the DH established the HGSG. Over a number of months the HGSG made a series of recommendations, some of which were referred to NHS England to consider. It is proposed that NHS England specialised services leads the re-design of genetic testing services through its medical genetics Clinical Reference Group. The model was proposed

by the report by the human genomics strategy group, 'Building on our inheritance - Genomic technology in healthcare' (January 2012). The model proposed by the HGSG, and subject to approval by the NHS England Board, has three components:

14. **Genomic technology centres** could operate as specialist centres of excellence with a focus on the interface between translational research and service innovation in genomic services. They could bring together clinical, academic, scientific and bioinformatics specialists to translate cutting-edge research in a collaborative and inclusive manner to ensure the participation of specialist expertise and promote the adoption and spread of research and innovation. They could play a key role in evaluating new markers for cost and clinical effectiveness. A key requirement is that these organisations should be designated as genomic technology centres through open competition against a specification, and commissioning would be through NHS England.
15. **Biomedical diagnostic hubs** with a strong integrated molecular capability could be developed to incorporate *all* current laboratory-based diagnostic services in pathology and genetics (inherited and acquired diseases). They are likely to be regional/network hubs of significant scale, and are emerging from the national pathology transformation programme. These hubs could operate as the essential interface between the clinician and the pathologist for rapid and appropriate testing, particularly where co-ordination of sample processing and analysis is crucial. The exact number of such laboratories and the scope of testing to be undertaken requires further development but is likely to include high throughput analysis, frequently requested biomarkers and, for example, molecular tests for microbiology, virology and haematology.
16. **Regional genetics centres** will continue to have an important role in the diagnosis of inherited disorders and the management of familial aspects of disease. They will continue to provide a key interface with patients with genetic disease. Clinical genetics services could have an expanded role, in partnership with specialist clinicians, to provide genetic expertise as genetic services are expanded and embedded in clinical pathways. As clinicians in other specialties become more proficient and the number and range of specialties involved continues to expand, it is envisaged that the relationship between regional genetics centres and other specialties will evolve to one which provides leadership, expert support and mentoring, and management of particular family issues such as reproductive counselling.

Progress to date

17. DH chief medical officer workstreams
The Chief Medical Officer (CMO), Professor Dame Sally Davies, commissioned three working groups to identify and assess key priorities and potential issues across three workstreams. They are:
 - *Workstream on scientific considerations* - This group was led by David Lomas, and was tasked with determining which genomes should be sequenced and what the principles are for choosing sub sets of patients and

diseases for sequencing. The following priorities for the PM's genomics strategy were recommended: Cancer (lung cancer, paediatric cancers, rare cancers syndrome, cancers of unknown primary origin); rare diseases; infectious diseases (HIV, hepatitis C virus, tuberculosis and potentially antimicrobial resistance);

- *Workstream on data storage* - This group was led by Professor Dame Janet Thornton and was tasked with identifying what parts of the genome should be stored, and how it should be stored and identified for storage purposes. The group looked at 3 areas: data infrastructure and flow; data specification and standards; training and workforce development; and
- *Workstream on ethics and consent* - This group was chaired by Professor Mike Parker, and set out principles for consent and data access for use by researchers and wider commercial platforms, what legally does the system need to assure by way of consent, and how we manage data from sequencing that may not be related to the patients original diagnosis. The following ethical issues were highlighted for consideration: gaining consent; data access; feedback to participants; public confidence and involvement; oversight and governance.

18. All three workstreams submitted reports to the Genomics Strategy Board which had been approved by the Secretary of State for Health in March. The proposals have been referred to Genomics England Limited.

19. Clinical and patient engagement workshop

This workshop was convened at the request of the Genomics Strategy Board (Chair: Malcolm Grant) and was led by Mike Richards, implemented by Dr Nina Wilson. On Friday 26 April 2013, over 70 of these clinicians came together for a seminar to determine the appropriate patient consent pathway for genomic sequencing in the NHS and to identify how clinical advocacy for the genomics strategy across the health system can be developed and improved. The workshop built on the outputs from the Science Working Group established by CMO and chaired by Professor David Lomas. The subsequent report, submitted to the Genomics Strategy Board in June 2013, made recommendations for the engagement of clinicians, patients and the public about the value, rationale and benefits of the genomics strategy. NHS England has engaged with the clinical leaders in the NHS representing those disease areas that the CMO will recommend the genomics strategy focus on. Given Sir Mike's recent appointment as Chief Inspector of Hospitals, NHS England is identifying a replacement for Sir Mike as the National Clinical Advocate for the 100K Genome programme.

20. Informatics and data

Issues of data ownership and transparency are of paramount importance to NHS England as set out in the Mandate and given the hugely positive developments in Care Data. Geraint Lewis is leading this work, and has begun work to consider how the sequencing data might be held, connected to patient records and subsequently be exploited. It will also look at the connections between this work and the establishment of care data in the NHS. The NHS England data and

informatics team will retain oversight of the informatics and data work and discussions continue on how it can best inform and support the implementation of business plan of Genomics England Limited.

21. Genomics England Limited.

The DH has established the company 'Genomics England'. The Secretary of State announced on the 5 July that Sir John Chisholm has been appointed Chair of the company Board. The remainder of the Board will now be appointed with a range of business and clinical expertise, including representative from NHS England. The Board will be responsible for the delivery of the business plan; including managing procurement contracts and taking operational decisions associated with the general running of the business. Genomics England Ltd is intended as a vehicle for delivery and the specific structure of the company and function is yet to be determined. Further work to underpin the business model of Genomics England Ltd. is currently taking place.
22. Genomics programme board.

The DH is also in the process of establishing the Genomics Programme Board. The programme board will report to Richard Douglas, Senior Responsible Officer for genomics at the DH, as part of the Department's contribution to the Growth agenda. The Chief Medical Officer (CMO), Dame Sally Davies, is currently in the process of inviting stakeholders to take part on the Board, which is expected to comprise of lead representatives from:

 - NHS England;
 - Public Health England;
 - Health Education England;
 - Genomics England; and
 - Department of Health.
23. The Programme Board will be chaired by Richard Douglas and Dame Sally Davies and will be tasked with establishing robust governance arrangements for this programme of work, monitoring delivery of the programme of work, and co-ordinating communications strategies and stakeholder engagement on genomics.
24. Benefits realisation

NHS England does recognise that the status quo of existing genetic testing services will be inadequate for the future.
25. NHS England commissioned PA Consulting, on behalf of the Genomics Strategy Board, to embark on a benefits realisation proposition which could underpin a business plan and operating model of the procurement vehicle. With the establishment of Genomics England Limited, this work will be transferred to that forum. In light of the recommendations by the HGSG, rationalisation and reconfiguration of existing genetic testing services will enable optimal benefits realisation
26. Direct commissioning of medical genetics services

NHS England took over the sole commissioning responsibilities for medical genetics from April 2013 through direct commissioning. A medical genetics national Clinical Reference Group (CRG) has been established under the

leadership of Frances Flinter <http://www.england.nhs.uk/npc-crg/group-e/e01/>. A national service specification has been published and the first national policy for pre-implantation genetic diagnosis has been launched in April 2013 giving for the first time equitable national access. The UK genetic testing network (UK GTN) has been preserved in the transition period and is now hosted by NW London commissioning support unit and works closely with the CRG .

27. To deliver the service model change a project brief has been developed by the specialised services national programme of care using the framework established for all direct commissioning specialised services. A revised service specification is planned to be developed for the genomic technology centres, the biomedical diagnostic hubs and the regional genetics centres. A growing suite of national commissioning policies will be developed. The proposed project brief identifies a process that procures the revised service structures for a launch date in January 2015.

Issues to consider

28. NHS England must discharge appropriately its legal duty for research and we will bring the voice of commissioners into sharp focus in the research landscape making an impact on - our legal duty for research, increasing number of NHS organisations involved in research and using research and evidence in commissioning decision making. We need a culture in the NHS where research is everybody's business and one where we encourage patient involvement in priority setting, enable the outcomes in High Quality Care for All, identify research opportunities particularly in anticipatory medicines and science of which genomics is a significant component and seek opportunities for the NHS to play its part in economic growth and increase capacity of NHS staff to effectively use research.

Next steps

29. The DH permanent secretary presented its growth strategy (including genomics) to Cabinet. Those papers included confirmation that the Department held the funding for this programme and as such, the DH will now assume full responsibility and accountability for the 100K genome programme ensuring clarity and coherency for our health care partners, Ministers and the public.
30. NHS England is responsible for service commissioning elements and will establish a formal service reconfiguration steering group as part of the medical genetics CRG. It will structure to oversee the reconfiguration project using the governance of the Clinical Priorities Advisory Group and direct commissioning committee to secure timely delivery.
31. NHS England is committed to ensuring optimum contribution for its part in delivery of patient benefit and patient interest whilst supporting economic growth. Steve Fairman, Director of Business, Improvement & Research, along with Professor *Keith Willett*, National Director for Acute Episodes of Care - *Domain Three*, will be leading NHS England Research Strategy, to which genomics must be aligned.

Sir Bruce Keogh, National Medical Director
Tim Kelsey, National Director for Patients and Information
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