BSGM Chair's update April 2018

This week saw the publication of the House of Commons Science and technology Committee report on Genomics and genome editing in the NHS (third report of session 2017-19). Many BSGM members submitted written and verbal evidence and the report is really worth a read for a concise collation of this evidence and a balanced perspective on the promises of genomic medicine as well as the obstacles to smooth and timely implementation.

Here I've paraphrased some of the key statements and recommendations I think will be of interest to the BSGM membership.

1. The UK is uniquely positioned to benefit from genomics because of our national health service. Nevertheless *much more needs to be done* to achieve the full potential that genomics promises.
2. The government should conduct a detailed evaluation of the 100,000 genomes project – including an assessment of the anticipated clinical and cost effectiveness- to inform the introduction of whole genome sequencing (WGS) into routine NHS care.
3. NHS England should embed processes for evaluating the impact of WGS as it configures the Genome Medicine Service (GMS) in line with recommendations from the CMO report "generation genome"
4. Budgets for required digital infrastructure for GMS should be agreed and confirmed now. Current digital infrastructures do not facilitate the iterative process of WGS interpretation. We need to avoid attempting to roll out a GMS at a speed that cannot be delivered
5. There are widespread concerns about insufficient training and lack of qualified NHS staff. HEE should complete detailed workforce planning and modelling asap.
6. Government should increase efforts to highlight the data sharing (or access) needed to enable a GMS
7. More work is needed on the appropriate consent framework in a GMS where research and clinical practice are inextricably entwined
8. The government should seek to renew the concordat and moratorium with the insurance industry as soon as possible and set up systems to monitor reluctance to undertake testing due to insurance concerns

BSGM members will be vital in helping to realise these recommendations and am sure you will have your own recommendations to add. For many years the discourse around genomics has focused primarily on the technical aspects, getting the sequence delivered cheaply and quickly, the pipelines and bioinformatics sorted. This is of course crucial, without it we would have no imminent GMS. But now we need to also focus on some of the complexities of applying this technology to real people appropriately. In the vast majority of cases, interpretation of a WGS sequence will require skilled clinical examination, family history taking and communication. BSGM wrote to Genomics England and NHSE in February highlighting 4 main areas of concern:

1. Errors arise when a test result at variance with the clinical presentation or family history is given too much weight, and/ or is regarded as a definitive diagnosis rather than one element in the process of diagnosis.
2. Utilising WGS for some diagnoses may mean that fewer WGS tests are available for the conditions where WGS is indicated.
3. More evidence about 'actionability' and clinical pathways are needed before additional 'looked' for findings are offered routinely
4. Premature use of this technology by clinical services unfamiliar with it dramatically increases the chance of litigation

See here for full letter

Professor Sue Hill replied last week acknowledging the need to work more closely with BSGM and its representatives as the GMS rolls out as NHS business as usual, post 100,000 genomes project. I

hope to be able to publish her full reply on the website soon.
Our US colleagues have highlighted some of these issues, albeit without the benefit of a national health service in a Genetics in Medicine paper from 2016:

Workforce planning
As highlighted by the above mentioned report, we are not currently training enough staff in the UK to cover essential posts. We therefore need to ensure there are avenues to attract staff from overseas. However, BSGM has been made aware of recent instances where suitably qualified staff have not been appointable to for example, genetic counsellor posts, because of visa issues. I understand the AGNC has written to the home office asking for genetic counsellors to go on the "Occupations Shortage List" [which could reduce these visa hurdles] but did not receive a reply. If you have further examples of such instances, please let BSGM know so that we can estimate the size of the problem.

Conferences and Meetings
Plans for the 2018 BSGM conference are well under way with an excellent line up of speakers confirmed. Please remember to mark Tuesday 2nd October in your diaries.

Travel Awards
May I encourage members to apply for awards from the BSGM to support attendance at meetings/workshops and training events. We are reorganizing these as we realize how funds are increasingly restricted and wish to use our funds for this activity. More details about how to apply and the expectations on awardees will follow very soon.
If there are issues you would particularly like to see covered in these chair's updates, or if you have relevant news items, please let me know

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