BSGM Chair's update January 2019

I do hope you have all managed to have a bit of a break over the festive season and that it's not too late for me to wish you all a very happy new year. I had hoped to get this update to you before the holidays, but various technical glitches prevented, and am hoping that your email inboxes are refreshed and better able to receive this update in January. Here is a brief summary of some of the activities that the BSGM has been involved in recently:

Transforming medicine through genomics conference

This BSGM/ Genomics England (GE) collaboration was a one day conference held 15th November at Charterhouse Square Queen Mary University of London. It was generously sponsored by GE and drew high profile speakers from around the UK Nov 15th. The programme is here .Thanks go to (previous BSGM chair) Bill Newman for all his hard work in organizing this meeting which has had excellent feedback. Films of all the talks will be available on GE website in due course. We are in discussion with Genomics England to repeat a similar event next year.

House of Commons Genomics Summit

I was very pleased to represent BSGM at the second 'Genomics Summit' held in Parliament on 31 October 2018, organised jointly by the House of Commons Science & Technology Committee and the Department of Health and Social Care. The event followed on from the Committee's Report on 'genomics and genome-editing in the NHS' and the launch of the Genomic Medicine Service in NHS England. The Rt Hon Norman Lamb MP, Chair of the Science and Technology committee was pleased to have got together "a whole lot of people in the same room who don't talk to each other enough" a sentiment I think we should continue to run with in 2019. Lord O'Shaughnessy then introduced two sessions of diverse short summaries from: Sue Hill, John Mattick, Rebecca Middleton Deborah Alsina, John Bell, myself, Munir Pirmohamed, and Matt Hurles. We discussed the promises of genomic medicine, how to ensure the UK remains at the forefront of developments, how to engage clinicians to use genomics in their practices and at the same time prevent overenthusiastic expectations, or interpretations, of what the technology can deliver.

Response to the launch of the life sciences sector deal- 2.

The government launched the second part of the life sciences sector deal on 5th December 2018. See here for the full details. Much of this focused on developing industry enterprises around genomics. One perhaps surprising element was the promise of direct to consumer (DTC) personalised health reports from genomics england for healthy individuals who pay to have their genome sequenced. On the one hand, this can be seen as an important step to turn some of the burgeoning private DTC market (I wont have been alone in being offered various 'bargains' for ancestry testing in 'black Friday' sales) into benefit for future NHS patients. On the other hand, experience from extant DTC services has already highlighted a tension when such consumers then want to (rapidly) access NHS resources for surveillance, risk reducing surgery etc. for which the evidence basis may be extremely limited. At this stage the undertaking is to look into the possibility, and BSGM is of course keen to liaise with Genomics England in this development work. We have written to the Genomics Board about this and together with JCGM to Matt Hancock (see letter here)

Genomic Medicine Service

Meanwhile it will not have escaped the notice of many that we have a new genomic medicine service, launched on 1st October 2018 to take over from the 100,000 genomes project but not expected to be fully live till early 2019. BSGM has been working with NHSE to develop an approach to consent to WGS (and other forms of testing) that acknowledges the clinical-research hybrid territory required for successful genome interpretation. I am really grateful to Alison Hall of the Ethics and Policy committee for her work on guidance on consent and confidentiality in the practice of genomic medicine. One element that many of you are already

helping to try out -ahead of the GMS being fully active- is the recording of clinical consent. Whilst the 100,000 genome project utilised a REC approved consent form, the NHS service will need to ensure that clinical consent discussions are appropriately informed. We have proposed a "record of discussions form" (RoD form here) to be used as an aide memoire for the consent discussion. We recognise that consent forms are not always deemed necessary, or helpful, when genetic tests are used in clinical practice. Some say, for example, that asking patients to complete a consent form for a genetic test exceptionalizes such tests, especially when done as part of a battery of biochemical and haematological tests for which no such form would be completed. We are working with NHSE and GE to ensure that the RoD form can then be aligned to consent for research on a person's genome, recognizing that although clinical consent and research consent are different (and have different governance mechanisms) they will often be taken at the same time. The new GMS will need to inhabit the hybrid space between research and clinical practice.

NHS long term plan

Then this week saw the long awaited launch of the NHS's long term plan in which genomics features prominently. For example, the plan for the NHS to become 'the first national health care system to offer whole genome sequencing as part of routine care'. The immediate focus of this will be on children with cancer, all of whom will receive whole genome sequencing 'to enable more comprehensive and precise diagnosis, and access to more personalised treatments'. BSGM members will need no convincing that our national health service provides a great vehicle for large-scale introduction of innovative technologies into clinical practice. Indeed since the inception of clinical genetic services we have been at this forefront, introducing new technologies and embracing research findings rapidly into practice. At the same time, through exposure to patients seeking genomic services we are very familiar with some of the social, ethical and legal challenges that are to be overcome, and have much experience at interpreting the complexities and ambiguities that genomic medicine can present.

Membership update

Unfortunately, there have been ongoing problems with the direct debit (DD) system to collect membership fees for BSGM and its constituent societies. This is a long and complicated story (that would be a whole lot worse if it weren't for the efforts of our long suffering treasurer Peter Farndon) but suffice to say, more than a quarter of our membership has not yet paid their fees for 2018. Those who haven't renewed have been sent email reminders, but it is just possible these are emails not accessed regularly. Logging onto the website and being proactive about payment of course requires more effort than many of our busy lives allow for, but could I urge you please to check with your colleagues whether their membership is up-to-date? [one way for them to tell is whether they receive this latest chair update. If they have not, their membership can easily be reinstated by contacting membership@bsgm.org.uk].

Please contact me (annekel@soton.ac.uk, or Anneke.lucassen@nhs.net) if you have any queries about the content of this email or any other suggestions relating to the Society.