

BSGM Chair's update February 2019

Since my last update, a small lull in Brexit news put a spotlight on the Life Science sector deal 2 plans I highlighted in my previous [update](#). The government has asked Genomics England to consider plans to offer personalised health reports for healthy individuals who pay to have their genome sequenced.

The headlines on the 26th January (e.g. Times, Guardian) suggested that this was a new announcement rather than coverage of a report published almost 2 months earlier (see [here](#)) demonstrating that media attention is still all important in awareness of government reports. The Times headline read: "Gene test for sale on NHS; Healthy patients to learn risk of killer diseases" and it is interesting that much of the reaction to this (if Twitter is anything to go by) was predominantly on how this threatens NHS principles of being free at the point of access. This is of course true, but the "learning about killer diseases" promise also warrants critical attention, even if the proposed test were free of charge. This readership is well aware that predictions of "killer diseases" in healthy people will be vaguer and less reliable than the popular discourse about genomics suggests. What I hope BSGM members can do is engage in this discourse, explain why the healthy adult genomic crystal ball is hazy and help Genomics England in their undertaking to the minister to consider the possibility.

Request from the Gene Curation Coalition (GenCC)

The GenCC represents groups that provide gene-centric resources to the community and include Clinical Genome Resource ([ClinGen](#)), Deciphering Developmental Disorders/Gene2Phenotype ([DDD/G2P](#)), Genetics Home Reference ([GHR](#)), Genomics England PanelApp ([PanelApp](#)), Online Mendelian Inheritance in Man ([OMIM](#)), [Orphanet](#), and the Transforming Genetic Medicine Initiative ([TGMI](#)). The group have designed a short survey to solicit community input on developing a harmonized set of terms for defining the level of evidence for genes implicated in disease. These terms could then be used across all of the GenCC resources to make them more interoperable and consistent for community use. This is outlined in a short video [here](#). Please could BSGM members complete this survey ASAP- ideally this week if possible. Survey [here](#)

Sad news

Many of you will already have heard about the death of Professor Sue Povey. Sue was an early pioneer in human gene mapping and use of human genetic tools to address a variety of medical and other research questions. She contributed to the compilation and annotation of the human genome, with particular involvement in alpha-1-antitrypsin deficiency and tuberous sclerosis. There is a lovely obituary in the guardian [here](#):
Peter harper also interviewed her some years ago available [here](#):

Some dates for your diaries

1. Annual BSGM conference, at RCP again this year 1st October 2019
2. Clinical Genetics Society Spring meeting March 12th with Carter Lecturer Prof I. Karen Temple
3. ACGS Summer Conference 10-11 June 2019 Birmingham
4. Cancer Genetics Group Spring meeting Wednesday 20th of March at the Gillespie Centre, Clare College, Cambridge
5. AGNC Spring meeting 26th April 2019 London

From the Pharmacogenetics and Stratified Medicine Network events that may be of interest to BSGM members:

- (a) an Open Meeting on 6th March at RCP see [here](#) for details and
- (b) Health Education England workshop to inform clinicians how the use of Pharmacogenomics is moving into the NHS on 22nd May 2019 also at the RCP.

Membership of BSGM and constituent societies

Please remember to ask your colleagues whether they have renewed their membership. If they are not receiving these emails, or emails about conferences, chances are they are not! We are

still some 400 members down since the direct debit debacle I outlined previously (another DDD for genetics!)

Chair's updates

As of 2019 we will post chair's updates on the BSGM website so that you can access these summaries without having to trawl through your inboxes.

Please contact me (a.m.lucassen@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.

Anneke Lucassen, Chair BSGM
Feb 2019