The Royal College of Pathologists
Pathology: the science behind the cure

Joint Committee on Genomics in Medicine

A meeting was held on Tuesday 23 May at 10.00am
hosted by the Royal College of Pathologists via Microsoft Teams

Dr Lance Sandle
Registrar

Present:
Prof Anneke Lucassen, Chair & BSGM Representative
Mr John Davies, RCP Comms and Policy
Prof Cathryn Edwards, RCP Registrar
Mr Nick Meade, Director of Policy, Genetic Alliance UK
Prof Zosia Miedzybrodzka, Scotland Representative
Ms Sasha Henriques, AGNC Representative
Dr Mark Kroese, PHG Foundation Representative
Prof Mark Kilby, RCOG Representative
Dr Sarah Clarke, RCP President
Prof Ramesh Arasaradnam, RCP Academic Vice-President
Mr Simon Ramsden, Deputy Chair of ACGS
Mr John Dean, General Secretary of BSGM

In attendance: Louise Mair, Governance & Committee Services Officer

Apologies:
Prof Kate Tatton-Brown, FRCP Representative
Prof Sarah Coupland, Vice President Communications
Prof David Rowitch, RCPH Representative
Dr Sian Morgan, RCPath Genomics & Reproductive Science SAC Chair
Prof Helen Firth, Former JCGM Chair

Absent:
Dr Tabib Dabir, Northern Ireland Representative
Mr Howard Ellison, Deputy Head of Committees Services (RCP)
Dr Helena Kemp, Chair of National Metabolic Biochemistry Network
Dr Alex Murray, SAC Chair

Minutes (Unconfirmed)

JCGM1.01/23 Welcome, apologies and Declarations of Interest

The Chair welcomed members to the meeting and asked for everyone in attendance to introduce themselves. Apologies for absence were received and noted above.

There were no new declarations of interest received.

JCGM1.02/23 Minutes of the previous meeting

a) The minutes of the meeting held on 24 October 2022 were approved as a correct record.

b) The following updates to the action log were noted:

JCGM1.03/22(g): The Chair to contact the Co-Chairs of the Genomics Professional Partnership Group (GPPG) to refresh the relationship between GPPG and JCGM. This action has been discussed to an extent when distinguishing the different remits of GPPG and JCGM in the meeting. The Committee agreed that this action could be closed.

JCGM1.03/22(h): Prof Coupland to look at the recommendation she received from the College of Radiologists on a representative joining the JCGM. This action is ongoing, the Governance and Committees Services Officer will locate an email with a recommendation for a representative from President Mike Osborn following the meeting.

JCGM1.04/22(b): The Chair will add a line around the distinction of JCGM from other genomics groups, for inclusion in the terms of reference. This action is ongoing whilst discussions between various parties are being held.

JCGM1.04/22(c): Prof Coupland to speak to contact from Cancer Research UK about a patient representative for JCGM. This action is ongoing and Prof Coupland to broaden the search by speaking to other cancer organisations. Governance and Committees Services Officer to pick this up further with Prof Coupland.

JCGM1.04/22(e): Prof Taylor to identify and invite a representative from his specialty to join JCGM. This action is ongoing. Governance and Committees Services Officer to reach out to Prof Taylor around recruiting an ACGS representative.

JCGM1.05/22(c): Prof Tatton-Brown to gain more insight on expanding trainee numbers and funding and feedback to the committee. This action can be closed.

JCGM1.16/22(j): The Chair to contact the Genomics Focus Group for support in providing evidence around the challenges clinicians are facing to NHS England. This action is ongoing.

JCGM1.25/22(g): The Governance and Committees Services Officer to review the current genomics groups and distinguish UK and English genomics groups. The Governance and Committees Services Officer advised JCGM that eight out of eleven of the genomics groups are English. The JCGM noted this and agreed this action could be closed.

JCGM1.25/22(i): The Chair to discuss the valuable points raised today around the value / role of JCGM with Prof Jones and Prof Coupland and then report decision back to the next JCGM meeting. The Chair is liaising with Prof Edwards around potentially moving the JCGM to RCP, this action will be discussed further under matters arising.

JCGM1.26/22(b): Governance and Committees Services Officer to review the membership list created by previous Chair Prof Firth and update the terms of reference to reflect the current / required JCGM membership. This action has been completed and can be closed.

JCGM1.26/22(e): The Chair and Dr Clarke to discuss the possibility of providing a presentation on the role of JCGM to the Medical Speciality Board. This action is ongoing.

JCGM1.27/22(h): Nick Meade and the Chair to discuss the call for evidence for the Down Syndrome ACT. This action is ongoing.

JCGM1.03/23 Matters Arising

a) The Committee reiterated previous discussions around the purpose of JCGM, agreeing that other committees tend to be England centric and focus primarily on genomics within laboratory-based services; whilst this multi-disciplinary committee includes the devolved nations and provides a vital opportunity for clinicians to have their voices heard. For some members, this is the only genomics committee they sit on, providing a crucial link between genomics and their specialty.

b) The Chair advised that she and Prof Edwards have discussed whether this committee should move to a Joint Committee within the Royal College of Physicians, so that the Committee administration sits in one place, without needing to rotate every three years between the three organisations. After three years, the Committee could then review and decide whether the Committee is still serving its purpose. The Committee agreed with this approach and Prof Edwards advised she would provide a proposal at the next JCGM meeting once she has completed the review of the current RCP committees structure. The Committee agreed that it will need to redefine its purpose and output if the proposal is approved.

Action: Prof Edwards to review the current RCP committee structure and then provide JCGM with a proposal for this committee to sit within the RCP.

JCGM1.04/23 Review of the Terms of Reference

The Committee agreed to review the terms of reference once the future of the JCGM is confirmed.

JCGM1.05/23 RCPath guidelines for recommendations of the use of chromosome microarray and pregnancy

As Dr Sian Morgan, Chair of the Genomics and Reproductive Science SAC was an apology, she was unable to present the paper at this meeting. Committee members noted that the JCGM released guidance on prenatal genetics in 2022, which highlights the ethical issues in prenatal genetics. As there may be overlap between this guidance and the RCPath guidelines, stakeholders related to pregnancy and perinatal would need to be consulted for input in a review.

JCGM1.06/23 Review retention of family records document

a) Mr Dean provided a short presentation summarising the retention of family records document. The BSGM propose that genetic family records should be retained for 30 years and that clinical genomics laboratories should retain the records of how they did the analysis for at least five years. However, laboratories will also need to retain a copy of the final report, including the analysis and interpretation details that went into that report for 30 years, as this

is equivalent to part of the genetic family record. Therefore, healthcare providers will need to plan for this long-term retention of digital and electronic data.

b) Mr Dean emphasised that as genetic family records contain transgenerational information, which is relevant from one generation to the other, three to eight years retention of records is not long enough to retain that utility.

c) There was a discussion around GDPR and the financial and environmental costs of maintaining large volumes of data over a long period, with Committee members suggesting potentially using specific parts of the genome in genome testing instead of the whole genome to be more environmentally sustainable.

d) Ms Henriques highlighted this proposal would improve issues of equity around how family information is passed through families who are affected by socioeconomics, disabilities, adoption, culture etc. and may not receive information on their family history. Ms Henriques queried whether the document includes patient consent and if patients will be made aware of how long the records are stored. The Chair confirmed that patient consent is included in the consent of confidentiality document and suggested the wording would need to be tweaked around how long the DNA samples are stored, as it is not clear in the document. Mr Dean clarified that the current guidelines state 5 years for raw data and 30 years for filtered data. The Chair asked how long raw data from other specialities are kept, Prof Kilby confirmed that the retention of ultrasound images of a foetus, falls into the perinatal group and is therefore retained for at least 30 years.

e) The JCGM agreed it is important that patients understand why data is stored and as there are no current regulations for data storage, these guidelines will be instrumental in providing best patient care. The JCGM approved the guidance document and thanked Mr Dean for his presentation.

JCGM1.07/23 Update from Representatives

a) Association of Genetic Nurses and Counsellors

The JCGM welcomed Ms Henriques, who is currently providing maternity cover for Ms Roberta Rizzo.

Ms Henrqiues advised that the AGNC have recently convened a group of genetic counsellors to consider how they could map genetic counselling across the UK. The JCGM discussed the current workforce issue impacting genetic nurses and counsellors, as well as considering how to ensure representation for genomics practitioners and the devolved nations.

Ms Henriques informed the JCGM that the Glasgow MSc Genetic Counselling course run by the University of Glasgow has unfortunately closed due to low student numbers, pressures on clinical teams and key training staff being on long-term sick leave. As a result, the AGNC are currently looking to organise a stakeholder genetic counselling group, including key staff involved in teaching and training into genetic counselling, which may then roll out to other stakeholder groups across the UK.

The JCGM noted that there is currently no government funding specifically for genetic counselling training in Scotland. Although there is a route into genetic counselling via the MSc Medical Genetics course, the course is teaching

focused and may be a significant commitment in comparison to the Glasgow MSc Genetic Counseling course.

The JCGM agreed that a strategy to support a UK wide genetic counselling course needs to be implemented, it would need to include key personnel who are instrumental to delivering the course as well as the devolved nations. Ideally, it would be not based in universities and alternative approaches could also involve online learning to provide an accessible route into genetic counselling. Ms Henriques will continue to keep the JCGM informed of any developments with this issue.

b) Royal College of Obstetricians and Gynecologists

Dr Kilby reported that the RCOG Genomics task force is ongoing and has been funded since 2019, but there is discussion within RCOG around the current structure and purpose of the task force. It is felt that genomics should be more embedded in every single committee within the Royal College of Obstetricians and Gynecologists, whilst retaining a slimmed down genomics task force for an out facing discussion.

The JCGM noted that RCOG has delivered several successful educational study days this year. Prof Kilby is also working with subspecialty trainees, particularly in fetal medicine, reproductive medicine and gynecological oncology, where genomics is having the biggest impact, ensuring that it is woven through the curriculum, so that there is an educational assessment on genomics in the RCOG examinations going forward.

Prof Kilby advised that RCOG and the Royal College of Midwives have recently received a grant to facilitate their joint work, which the JCGM agreed was positive news.

Prof Kilby also advised that different educational and podcast discussions will be released in June, involving obstetrics, gynecology and specialties and the medical specialties surrounding that. The JCGM thanked Prof Kilby for his update.

c) Association for Clinical Genomic Science

Mr Ramsden advised that there is new leadership within the ACGS, and they are actively trying to ensure there is collaboration with senior colleagues from the devolved nations, to ensure all voices are heard on current workforce issues.

d) British Society for Genetic Medicine

The Chair advised that the BSGM are looking at workforce, particularly around the technologies and primarily focused on England, but noted that any lessons learned will be extrapolated to the devolved nations.

The Chair highlighted the challenge laboratories are facing despite receiving significant investment, including large backlogs and the difficulties hiring trainees, due to increased workload pressures. However, the JCGM agreed that it is essential that laboratories develop a strategy to overcome these challenges and continue to fund the discipline.

e) Genetic Alliance UK

Mr Meade advised that Genomics England are moving forward with their research study on genome sequencing for screening and are in the process of convening a working group focused on communicating results and providing support to families. Mr Meade is pleased that he and Dr Sarah Bowdin can contribute to this comprehensive consultation and will keep the JCGM informed of any progress made in this area.

Mr Meade highlighted the Down Syndrome Act guidance document, advising that he hopes the guidance will help everyone living with a rare condition and where it overlaps with Down syndrome. The JCGM agreed the guidance document should not be called the Down Syndrome Act guidance, as it excludes other genetic conditions, and noted that Mr Meade is in the process of gathering a position statement and will be asking members for their input before it is sent to the Department of Health and Social Care for consideration.

JCGM discussed concerns around families having to wait for long periods of times for a diagnosis and clinicians feeling unequipped to provide advice on the best possible private healthcare for genetic diagnosis, as their argument has always been to stay within the NHS for the best quality service. The JCGM noted that Mr Meade is having a constructive conversation with Dame Sue Hill around this issue and will continue to keep the committee informed of any developments in the future.

The Chair advised that the BSGM and RCGP have guidance on their website around direct to consumer genetic testing and suggested that Mr Meade and BSGM could work on collaborating on clearer guidance on dealing with large volumes of direct consumer testing results. The Chair added that she would be happy to contribute to the guidelines.

Prof Miedzybrodzka advised that through the lead clinicians group there is an annual audit of direct consumer testing impact on NHS genetics units. Prof Miedzybrodzka is working with a group of sociologists looking at extended carrier testing and added that any contributions from the JCGM would be welcomed.

JCGM agreed that some providers may offer quick and accurate results, but the main concern is the interpretation of the results without the clinical context, which will continue to be discussed.

f) Scotland

Prof Miedzybrodzka reported that the Scottish Government has recently released an intent to publish a strategy on genomic medicine. The JCGM agreed this was fantastic news as it would provide additional funding for cancer testing and retain temporary staff.

Prof Miedzybrodzka highlighted that Scotland are not seeing the same large backlog issues in genetic testing faced in England, JCGM noted that the typical waiting time for results in Scotland is around 3 months, this is despite not receiving the level of funding as England.

Prof Miedzybrodzka advised there is a workforce group looking at staffing and training, but so far there isn't any commitment in terms of increasing numbers of clinical genetics scientists. Prof Miedzybrodzka noted that this may be a result of

the impact of the war in Ukraine and alternative priorities in the Scottish Health Service.

JCGM1.08/23 Any Other Business

a) The Chair highlighted the rising ethical challenge that clinicians are facing when using snip arrays to diagnose development delays in children. As this technique can inadvertently identify an incestuous genetic link, clinicians are having to decide when they should inform authorities about a potential safeguarding issue. Committee members agreed this is an important ethical issue within genomics and the NHS, and that the JCGM should provide a position statement on recommendations for clinicians, in collaboration with the BSGM ethics committee. The Chair will continue to discuss this issue further with the JCGM over the coming months.

b) The Chair noted a slight error in the previous JCGM minutes, on page 7 of 9, the minutes stated that Prof Firth was the host of the BSGM event in November 2022, when she was in fact, a guest speaker. The Governance and Committees Services Officer agreed to amend this.

Action: The Governance and Committees Services Officer to revise the previous minutes to reflect that Prof Firth was a guest speaker and not the host of the BSGM event held in November 2022.

JCGM1.09/23 Dates of Future Meetings

The Committee agreed that it would aim to schedule the next meeting in September / October.

Action: The Governance and Committees Services Officer to circulate potential dates for the next JCGM meeting to the Committee following this meeting.