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

Joint Committee on Genomics in Medicine

A meeting was held on Wednesday 26 October at 14.00pm
hosted by the Royal College of Pathologists via Zoom

Dr Lance Sandle
Registrar

Present:	Prof Anneke Lucassen, Chair & BSGM Representative Prof Louise Jones, RCPath Genomics & Reproductive Science SAC Chair Mr John Davies, RCP Comms and Policy Prof Cathryn Edwards, RCP Registrar Prof Helen Firth, Former JCGM Chair Mr Nick Meade, Director of Policy, Genetic Alliance UK Prof Zosia Miedzybrodzka, Scotland Representative Prof Bill Newman, CRG Chair Prof Eamonn Sheridan, BSGM Representative Ms Roberta Rizzo, AGNC Representative Dr Mark Kroese, PHG Foundation Representative Dr Alex Murray, SAC Chair Prof Sarah Smithson, CGS Chair Prof Mark Kilby, RCOG Representative Ms Sarah Clarke, RCP Representative
In attendance:	Ms Louise Mair, Governance & Committee Services Officer
Apologies:	Prof Diana Eccles, FRCP Representative Prof Kate Tatton-Brown, FRCP Representative Prof Sarah Coupland, Vice President Communications Prof David Rowitch, RCPH Representative Prof Ramesh Arasaradnam, RCP Academic Vice-President Prof Robert Taylor, ACGS Chair
Absent:	Dr Tabib Dabir, Northern Ireland Representative Howard Ellison, Deputy Head of Committees Services (RCP) Prof Andrew Goddard, President, RCP Dr Helena Kemp, Chair of National Metabolic Biochemistry Network

Minutes (Unconfirmed)

JCGM1.23/22 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.

JCGM1.24/22 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 Chair advised she would email Prof Tatton-Brown, as Prof Tatton-Brown agreed to flesh out the differences between GPPG and JCGM.

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 get more insight on expanding trainee numbers and funding and feedback to the committee. This action is ongoing.

JCGM1.16/22(g): The Governance and Committees Services Officer to circulate a table of genomic committees and their roles and overlaps to JCGM for review and comment. This action has been completed.

JCGM1.16/22(h): Mr Meade to contact the Chair and discuss how she could support Mr Meade regarding the call of evidence for the Downs Syndrome Act. Mr Meade is in the process of gathering a position statement and this action will be discussed further during his update later in the meeting.

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.

JCGM1.17/22(a): The Chair to circulate paper on ethical issues in new-born screening by WGS. This action has been completed.

JCGM1.17/22(a): The Governance and Committees Services Officer to add the group chat discussion around new-born testing to this set of minutes. This action has been completed.

JCGM1.21/22(a): The Chair to circulate the guidance on prenatal and pre-implantation for cancer susceptibility syndrome to JCGM for comment. This action has been completed.

JCGM1.21/22(a): Prof Miedzybrodzka to send the Chair information around prenatal familial cancer syndromes, and the Chair to confirm whether this has been incorporated into the guidance on prenatal and pre-implantation for cancer susceptibility syndrome. This action has been completed.

JCGM1.25/22 Matters Arising

a) The Chair summarised the background of the JCGM, noting that the JCGM was first established in 2001 and is a tri-partite committee comprising of the Royal College of Pathologists (RCPath), The Royal College of Physicians (RCP) and the British Society for Genetic Medicine (BSGM).

b) The Chair explained that she is a BSGM representative, and the administrative function currently sits with RCPath, which has raised some queries around the identity of JCGM and whether the administrative aspect should sit within the royal colleges or within BSGM, whilst Prof Lucassen is Chair. The Chair raised the overarching question around the role of this committee and defining its distinction from other similar genomics committees, such as the Clinical Genetics Society.

c) Previous meetings have highlighted that the JCGM provides members with a unique opportunity to have honest and frank discussions around genomics which is not always possible in other settings. However, the Committee agreed that it was important to discuss whether the JCGM has become superfluous, given the sheer number of other genomic committees JCGM members sit on and juggling meetings alongside their increasing workload.

d) The Committee agreed there is a significant value in JCGM and its role in representing all of the royal colleges, which allows a multidisciplinary discussion across genomics and how it impacts patients, such as reproductive medicine, gynaecology etc. The Committee agreed that there is a need for these parties to be represented in this forum.

e) The Chair queried whether the administrative management of the JCGM would be best suited within the same organisation as the Chair, i.e., BSGM? The consensus appeared to be that it stays within the royal colleges.

f) There was consensus from the Committee that JCGM provides an independent and necessary voice from multiple specialities, particularly when contributing to consultations, such as the recent service specification consultation document, as well as critical guidance documents, notably, the recent childhood testing publication from BSGM.

g) The committee reviewed a list of current genomics groups and discussed their distinct remits and overlaps in memberships for JCGM members. Prof Jones clarified that this is not an exhaustive list of genomics groups, but significant ones that she, RCPath President and Prof Sarah Coupland sit on. The list is likely to be expanded, with input from other JCGM members. The Committee agreed the importance of including UK genomics groups, as there are a significant number of English genomics groups, and it is essential that the devolved nations are included. It was agreed that the current list is a good foundation to gain clarity on the different genomics groups remits. The Governance and Committees Services Officer agreed to review the current genomics group list and note which groups are English or UK and report back to the JCGM.

Action: The Governance and Committees Services Officer to review the current genomics groups and distinguish UK and English genomics groups.

h) In summary, the JCGM agreed that this committee provides the opportunity to directly inform the colleges and organisations within it. Some members do not currently attend other genomics groups, therefore the JCGM also provides a space where all voices can be heard and potentially powerful issues can be raised, as well as discussing significant patient care issues.

i) The Chair thanked everyone for their valuable contributions to the discussion and advised she would discuss this further with Dr Louise Jones and Prof Sarah Coupland and then report back at the next meeting.

Action: The Chair to discuss the valuable points raised today around the value / role of JCGM with Prof Louise Jones and Prof Sarah Coupland and then report decision back to the next JCGM meeting.

JCGM1.26/22 Review of the Terms of Reference

a) The Committee reviewed the JCGM terms of reference, which was last updated in 2019, when its administration sat with the RCP.

b) The Committee agreed that the membership list would need updating, as it does not include a representative from the Clinical Genetics Society or the devolved nations, who are members of JCM. Prof Firth referenced a spreadsheet with future JCGM membership which she devised before demitting her post as Chair, the Governance and Committees Services Officer agreed to review it.

Action: 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.

c) The Committee highlighted that the current terms of reference notes the royal colleges statement and agreed that BSGM's mission statement and other colleges statements are valuable and should be included in the terms of reference. Prof Jones recommended the inclusion of the Royal College of Radiologists on JCGM, who have been keen to be involved in these discussions and do not currently have a formal outlet. As discussed earlier in the meeting, the Governance and Committees Services Officer will obtain the contact details for the radiologist recommended by the RCPath President.

d) The Committee agreed the importance of JCGM consisting of a core group of clinicians interfacing with genomics for a living and providing a service delivery who can contribute to this committee, before considering widening JCGM membership.

e) Ms Clarke suggested that an effective way of encouraging other specialities to join the JCGM would be to provide a presentation from JCGM to the Medical Speciality Board. Ms Clarke advised she would be pleased to host a presentation with the support of the Chair.

Action: The Chair and Ms Clarke to discuss the possibility of providing a presentation on the role of JCGM to the Medical Speciality Board.

JCGM1.27/22 Update from Representatives

a) Clinical Genetics Society (Prof Smithson)

Prof Smithson discussed two significant developments for CGS since the last JCGM meeting: providing a critical response to the service specification consultation and conducting a workforce survey for clinicians in the UK and the workforce unit at RCP.

Prof Smithson advised that CGS and Prof Tatton-Brown have worked closely together on increasing training numbers for the Genomics Training Academy. Prof Tatton-Brown has played a key role looking at the clinical genetics workforce and it is likely there will be recruitment drive of more ATN's by March 2023.

Prof Smithson thanked the JCGM for their input into the CGS response to the service specification consultation. There was a discussion around the significant concerns raised from the document and the Committee agreed that whilst the document was aspirational, it needed to manage expectations and provide realistic timescales to deliver effective genomics services to patients.

b) Association of Genetic Nurses and Counsellors (Ms Rizzo)

Ms Rizzo was pleased to highlight the excellent collaboration between AGNC and CGS for the response to the service specification consultation. One area of concern raised from the service specification was around introducing predictive testing and the potential impact this would have on the workloads of genetic counsellors.

The Committee noted that AGNC are working with the Mainstreaming National Task Force and specialist nurses working within genomics and reviewing their training needs and looking at competency framework to ensure they have sufficient support.

A recent survey carried out by AGNC found that 40% of mainstream services are offering predictive testing which JCGM agreed is concerning. AGNC are conducting a report around building a network of specialist nurses, which will be critical due to the link between genetic services and the mainstream.

c) Northern Ireland (Dr Dabir)
There was no update provided in the absence of Dr Dabir.

d) Wales and SAC (Dr Murray)
Dr Murray provided an update as Chair of SAC. The new curriculum is continuing to embed and has taken some time for trainees to transfer to the new curriculum but overall, it has been a successful transition.

Dr Murray reiterated previous discussions around using a European platform to host examinations. Registrars currently take a short knowledge-based assessment hosted by RCPath, a requirement for registrars to pass before they receive their CTC. The SAC have been approached by the European Medical Genomics Body to consider whether the European exam would be a suitable knowledge-based assessment for registrars. It has a two-part multiple-choice format and would pass the responsibility of contributing to the assessment to the European body. There is also a precedent, as Cardiology use this examination as their knowledge-based assessment for their curriculum. There was a discussion whether registrars should continue to take the knowledge-based examination, although it was agreed that the preparation registrars undertake for the examination is beneficial in itself for their career development.

The Committee noted that Dr Murray's term of office as Chair on the SAC ends next spring, with the March SAC meeting being her last meeting.

Dr Murray advised that Wales has recently developed its delivery plan for Genome UK, which will be shared shortly. The Cardiff trust has recently refurbished its building, and will be combining the Genetic Service Clinical labs, Wales Gene Park and Pathogen Genomics unit later this year. There was a discussion around the development for service plans being dependent on space and the acknowledgment that the current facility is unsustainable for staff to work in. These challenges have resulted in staff leaving the trust and taking on remote positions within the GLH's, which the trust is working to mitigate as much as possible.

Prof Miedzybrodzka asked whether moving the knowledge-based examination to the European exam would help recruit clinical geneticists from abroad. Dr Murray explained the current application process for clinical geneticists, which involves joining the specialist register and going through a training program or Caesar process. Dr Murray advised that the Caesar process is in the process of being streamlined, as international geneticists are facing the challenge of carrying out broad based training when they have been in practice for several years, which discourages international geneticists from applying to work in the UK.

e) Scotland (Prof Miedzybrodzka)
Prof Miedzybrodzka advised that the National Strategic Network for Genomics in Scotland has been launched. It is in its early stages and primarily focused on laboratory coordination, running the test directory, and applying for funding from the government to pay for new tests.

Prof Miedzybrodzka highlighted that Scotland has received substantial funding and a business case can now be proposed around the existing service for learning disabilities and offering testing for 12 cancer priorities.

There have been some frustrations around the transformation to systems, as it was felt that the previous system was fit for purpose, however, the transformation has provided an opportunity for additional funding which is extremely positive and much needed.

f) RCPCH (Prof Rowitch)
There was no update in absence of Prof Rowitch.

g) BSGM (Prof Sheridan)
Prof Sheridan advised that BSGM will be holding a face-to-face conference on 8 November, hosted by Prof Firth, subject to train strike action.

BSGM has also responded to the consultation document on clinical genomics specification, which largely echoed the concerns raised by the JCGM. The British Association for Adoption and Fostering has also approached BSGM for input when updating their statement of use of genetic testing on looked after children.

BSGM also plays a role within the Clinical Reference Group of Genomics, which is in the process of being reconstituted, members of the CRG have been invited to maintain their membership until middle of next year. Prof Newman, the Chair, is stepping down as is Prof Smithson. Current incumbents are to stay on for another 9 months, but it is not clear what happens in terms of the future of the group and representation of clinical geneticists.

h) GAUK (Mr Meade)
Mr Meade raised concerns with genetics testing and the quality-of-service patients are receiving, with excessive waiting lists for many patients. GAUK have also responded to the services specification consultation, with an emphasis on outcomes for patients, waiting times and making sure staff have the resources to provide an adequate service.

The Committee were pleased to hear that there has been some positive progress with the Down Syndrome Act. The Department of Social Care have acknowledged that Down Syndrome is one of many genetic conditions that cause learning disabilities and the importance of including other genetic conditions for the whole population. Mr Meade emphasized the need for royal colleges to respond for the call for evidence. The Chair agreed to support with this.

Action: Mr Meade and the Chair to discuss the call for evidence for the Down Syndrome ACT.

Mr Meade advised that progress has been made with the Blood Spot Task Group, there was a recent meeting where the terms of reference were reviewed, and changes were suggested as parts of the terms of reference were unclear to group members.

i) ACGS (Prof Taylor)
There was no update provided in the absence of Prof Taylor.

j) CRG (Prof Newman)
There was no update provided in the absence of Prof Bill Newman.

k) RCOG (Prof Kilby)

During the meeting Prof Kilby provided a written update for RCOG in the group chat. Prof Kilby advised that the genomics taskforce is now an established and funded process within the College, comprising of multidisciplinary members focusing on women's and newborn health. There are several areas of work: educational, focusing on educating specialists and trainees in O&G and emphasising multidisciplinary working with clinical genetics at the heart. There has been an inclusion of genomics in the syllabus of the MRCOG exam and subspecialty training and finally several genomic study days; input into Newborn screening with WGS and highlighting concerns of consent by whom and when (amongst other concerns) and RCOG responded to the NHSE Genomics Strategy. There has also been positive progress in terms of transgenerational genomics, however, RCOG has emphasised the need for funding of the clinical infrastructure to support the clinical introduction of genomics.

l) Registrars (Dr Ross)

Dr Ross advised that there has been a shift in the registrar role, in line with the progress in technology. The Committee noted that some registrars have had to wait for over two years to receive the bulk of their examination results, which the JCGM agreed is frustrating.

Dr Ross highlighted that fellow registrars are feeling some uncertainty around the possibility of the knowledge-based examination being replaced and whether they will have to duplicate their exams. Dr Murray assured the Committee that registrars will not have to take the exam twice if the knowledge-based assessment is replaced.

JCGM1.28/22 New-Born Screening by WGS

a) Prof Firth raised concerns around a clinical service being commissioned by the Genomics Unit, which currently does not include clinical members, whilst other clinical services are commissioned via a different route. The JCGM agreed that this approach could impact patient care, especially as the emphasis on testing potentially excludes patients without a positive diagnosis, who are in as great a need of care as patients with a positive diagnosis. Prof Firth and Prof Sheridan highlighted that the current diagnosis rate for WGS is around 15-20%, the Committee agreed to discuss this item further at the next meeting.

JCGM1.29/22 National Test Directory

a) The Committee agreed to discuss this item in detail at the next JCGM meeting.

JCGM1.30/22 Updating of RCPath Website

a) The Committee agreed to discuss this item in detail at the next JCGM meeting.

JCGM1.31/22 Any Other Business

There being no other business, the Chair thanked the committee members for their valuable contributions and the meeting was concluded.

JCGM1.32/22 Dates of Future Meetings

The Committee agreed that it should aim for three to four meetings a year. Further meeting dates will be confirmed in late 2022 / early 2023.