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

Joint Committee on Genomics in Medicine

A meeting was held on Wednesday 24 August at 14.00pm
hosted by the Royal College of Pathologists via Zoom

Dr Lance Sandle
Registrar

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

Minutes (Unconfirmed)

JCGM1.12/22 Welcome, apologies and Declarations of Interest

The Chair welcomed members to the meeting. Apologies for absence were received and noted above.

There were no new declarations of interest received.

JCGM1.13/22 Minutes of the previous meeting

a) The minutes of the meeting held on 25 May 2022 were approved as a correct record.

2:20pm – Dr Edwards and Prof Coupland joined the meeting

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 is still ongoing and the Chair and Prof Tatton-Brown will liaise for discussions 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.

JCGM1.04/22(e): Prof Taylor to identify and invite a representative from his specialty to join JCGM. This action is ongoing.

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.

JCGM1.07/22(a): The Interim Committees Services Officer to set up a JCGM meeting for July / August. This action is completed and can be closed.

JCGM1.08/22(a): The Committee to review the childhood testing guidelines and Prof Firth's concerns before approval. This action is completed and can be closed.

JCGM1.10/22(a): Chair to review the retention of genetic family records and JCGM comments and feedback to Prof Sheridan. This action is ongoing.

2:40pm – Prof Sheridan joined the meeting

JCGM1.14/22 Matters Arising

a) There was a discussion around the importance of representation from all three organisations at JCGM, further membership recruitment is needed, particularly for RCPath, as it currently has two representatives. The role of the JCGM will also continue to be discussed as the committee develops over the coming months.

JCGM1.15/22 Review of the Terms of Reference

a) The JCGM agreed to review this in more detail at the next meeting scheduled for October where attendance was expected to be higher.

JCGM1.16/22 Update from Representatives

a) Clinical Genetics Society (Prof Smithson)
There was no update provided in the absence of Prof Smithson.

b) Association of Genetic Nurses and Counsellors (Ms Rizzo)
There was no update provided in the absence of Ms Rizzo.

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

d) Wales and SAC (Dr Murray)
There was no update in the absence of Dr Murray.

e) Scotland (Prof Miedzybrodzka)
Prof Miedzybrodzka advised that a new strategic network for genomics is developing, with a focus on clinical voices being heard. Although the network is in its initial stages, core organisers are in place and an interim clinician has recently been appointed.

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

g) BSGM (Prof Sheridan)
Prof Sheridan confirmed that he will be retiring at the end of this year and advised that Dr Gemma Chandratillake, Chair Elect of BSGM, should be invited to join JCGM as the BSGM representative.

The JCGM noted there have been changes to laboratory provision in England, which has also had an impact on other nations. BSGM will continue to lobby and keep JCGM updated with any developments.

Prof Sheridan was pleased to confirm that BSGM are providing monthly learning webinars for members and non-members. BSGM will also be returning to an in-person annual conference this November to be held at RCP.

There was a discussion around the current mechanism for JCGM feeding back to BSGM, especially since the current chair is a BSGM representative. The Chair highlighted the importance of having a mechanism where concerns raised in JCGM can be fed back to relevant parties and actioned. Prof Firth confirmed that historically, JCGM have written letters of concerns to ministers and have responded to relevant consultations.

Prof Coupland advised the JCGM that she and Prof Louise Jones have created a table to summarise all the RCPath related committees involved with genomics. Prof Lucassen attended a meeting to discuss potential overlaps of these committees. It was agreed that the JCGM should review this and provide comment.

Action: The Governance and Committees Services Officer to circulate a table of genomic committees and their roles and overlaps to JCGM for review and comment.

The JCGM was created in the early 2000s to provide a link between rapidly expanding genetic services (both clinical and laboratory) with the Royal Colleges so that workforce planning and NHS service development could be addressed. Given that many other Royal Colleges have been created since then, and that the much newer GPPG has overlapping aims, it is important to consider that the role of JCGM should be in 2022. Discussions at this meeting, and the last one, suggest that there is still a role for a committee exploring the issues facing clinical genomic services today [it was noted that GPPG focusses more on future planning and mainstreaming, leaving a gap for focusing on extant genomic services].

h) GAUK (Dr Meade)

Dr Meade confirmed that GAUK have recently appointed CEO Louise Fish. Lauren Roberts has left the team and has been replaced by Isobel Randall, a key contact for Swann UK.

GAUK recently circulated a survey to its members to review its strategy and also published a survey for family with children with undiagnosed conditions to understand their needs, if any key findings are identified these will be published.

Dr Meade advised that GAUK's key priority is supporting the implementation of the UK rare diseases framework, three actions plans will be published by the end of the year.

JCGM discussed the proposed Downs Syndrome Act and agreed that it is imperative that awareness of the complexities for people accessing learning disability services is raised when providing evidence and that the views of parents of children with other genetic conditions are included in the guidance. The Chair offered to support Dr Meade in the call of evidence and they agreed to speak further offline.

Action: Dr Meade to contact the Chair and discuss how she could support Dr Meade regarding the call of evidence for the Downs Syndrome Act.

There was a discussion around the long waiting lists for patients to see a clinical geneticist and the increasing number of patients deciding to seek private care, which raises concerns on the quality of service and overall health outcomes.

i) ACGS (Prof Taylor)

Prof Taylor confirmed that ACGS held an in-person meeting over the summer which was successful, with representation across the UK and included talks from trainees.

j) CRG Prof Bill Newman

Prof Newman advised that the CRG's primary focus over the past two years has been working on service specification for clinical genomics. The specification has been re-written several times due to changes to the NHS process, but the final specification now addresses the main challenges faced, such as increased amount of cancer testing, separation from Europe etc.

There is also an equality and diversity piece of work being implemented to ensure equity of access and provision, as well as consideration about the workforce, which is recognising there is only a small number of geneticists and genetic counsellors, many of whom will be retiring in the coming years.

The JCGM noted the proposed multi-million-pound investment in genetics in England, with 80 new genetic counsellors and 12 new geneticists. CRG are also looking at clinical outcomes which will replace their current dashboard, ensuring that individuals views have been documented. This will be under public consultation, there will be webinars on 12 and 14 September and if it is approved there will be a launch date of 1 April next year.

Prof Coupland queried whether funding has been secured for diagnostic pathologists. Prof Tatton-Brown advised that this should have been prioritised as this expertise has been recognised in meetings, but she would need to confirm and update JCGM.

There was a discussion around the potential costs and risks of assigning the wrong genetic diagnosis, which can easily happen if a genomic sequence is interpreted without a clinical context. The JCGM acknowledged the enormous emotional cost for the families in such cases as well as for families who receive an uncertain or no diagnosis. The JCGM agreed that the NHS genomics unit often appears to adopt an approach that minimises the role of the clinician in interpreting WGS, and that this needs better highlighting to ensure that patients with serious genetic disorders receive the best quality care. The Chair asked the JCGM for suggestions on how to provide evidence to NHS England, to reflect the pressures the NHS is under and to gain support; one approach may be consulting with the Genomics Focus Group at RCPath, who should be able to provide further insight and evidence to NHS England.

Action: The Chair to contact the Genomics Focus Group for support in providing evidence around the challenges clinicians are facing to NHS England.

j) Registrars (Dr Ross)
There was no update provided in the absence of Dr Ross.

3:05pm – Dr Tatton-Brown left the meeting
3:33pm – Dr Edwards left the meeting

JCGM1.17/22 New-Born Screening by WGS

a) Prof Firth raised concerns about the ethical justifications of implementing new-born screening by whole genome sequencing (WGS). A discussion ensued similar to above, where the risks of interpreting a genotype that is adrift of any phenotype were discussed, and that this seems to be missing in the discourse around new-born screening. There is a political drive to offer new-born screening, and a public consultation appears to be in favour, but the JCGM are concerned that the complexities around this process are not yet well understood (i.e., the risk of false positives or uncertain results which require many years of expensive NHS follow up). The Chair noted that she had just had a paper accepted in a new journal summarising some of these issues. She agreed to circulate this paper to JCGM membership.

Action: The Chair to circulate paper on ethical issues in new-born screening by WGS.

Several discussion points arose in the group chat, so the Chair asked the Governance and Committees Officer to include the group chat when circulating the final minutes to the JCGM.

Action: The Governance and Committees Services Officer to add the group chat discussion around new-born testing to this set of minutes.

JCGM1.18/22 Retention of Genetic Family Records

a) The Committee agreed to discuss this agenda item at the next meeting scheduled for October.

JCGM1.19/22 National Test Directory

a) The Committee agreed to discuss this agenda item at the next meeting scheduled for October.

JCGM1.20/22 Updating of RCPath Website

a) The Committee agreed to discuss this agenda item at the next meeting scheduled for October.

JCGM1.21/22 Any Other Business

a) The Chair thanked the JCGM for commenting on the most recent sets of guidance issued by BSGM and asked the JCGM to provide comments on a fourth set of guidance on prenatal and pre-implantation for cancer susceptibility syndrome.

Action: The Chair to circulate the guidance on prenatal and pre-implantation for cancer susceptibility syndrome to JCGM for comment.

Prof Firth queried whether her concerns raised at the previous JCGM meeting around the use of predictive testing for likely pathogenic variants were considered and incorporated into the final set of childhood testing guidance. The Chair had been reassured by Alison Hall that they have been included.

Prof Miedzybrodzka advised JCGM that she had taken part in a recent debate around prenatal familial cancer syndromes and queried if this was included in the guidance for prenatal and pre-implantation for cancer susceptibility syndrome. The Chair was unsure, and Prof Miedzybrodzka agreed to send across some documentation around this to the Chair.

Action: 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.

JCGM1.22/22 Dates of Future Meetings

Wednesday 26 October 2022, 10:00am

The meeting closed at 4:02pm.