

Joint Committee on Medical Genetics

The Royal College of Physicians

The British Society for Human Genetics

The Royal College of Pathologists

RCPPath, 2 Carlton House Terrace, London, SW1Y 5AF

A meeting of the Joint Committee on Medical Genetics was held at the Royal College of Pathologists on Thursday, 25th January 2007.

Present:	Dr John Crolla	JCMG Chair, (RCPPath)
	Ms Amanda Barry	BSHG
	Dr Hilary Burton	(Observer, PHGU)
	Dr Teresa Davies	(RCPPath),
	Mrs Hilary Grandey	(RCP, Patient and Carers Network)
	Dr Alison Hill	Observer, DH
	Dr Hilary Harris	RCGP
	Dr Tessa Homfray	RCP
	Professor Adrian Newland	(RCPPath President)
	Mrs Gail Norbury	RCPPath
	Ms Kim Smith	BSHG
	Professor Peter Soothill	RCOG
	Dr Fiona Stewart	(RCP)
	Dr Karen Temple	BSHG
	Professor Richard Thompson	(RCP Treasurer, deputising for Dr Rodney Burnham, RCP Registrar)
	Prof. Richard Trembath	BSHG, Chairman
In attendance	Dr Lyn Chitty	(Safe Network)
	Dr Maggie Ireland	(Public Health Genetics Unit)

1. Apologies for absence

NOTED: apologies were received from Dr Mark Bale (Observer, DoH), Professor Ian Gilmore (RCP, President), Dr Jim Bonham (RCPPath, MetBioNet), Dr Rodney Burnham (RCP, Registrar), Dr Trevor Cole (RCP), Prof. Paola Domizio (RCPPath, Registrar), Dr Sally Davies (RCP), Dr Susan Holder (RCP, workforce representative), Professor Peter Farndon (National Genetics Education and Development Centre), Mr Alastair Kent, (GIG / RCP Patient and Carer Network), Dr Anneke Lucassen (BSHG), Dr Sian Morgan (RCPPath Trainee representative), Dr Allison Streetly (NSC, Observer), Ms Su Stenhouse (BSHG) and Dr Virginia Warren (FPH).

2. Membership

NOTED: that Dr Alison Hill had taken over as the Department of Health (DoH) observer from Ms Dianne Kennard. Dr Geoff Woods had notified the JCMG chair of his resignation from the JCMG as the RCPCH representative, citing other commitments.

3. To confirm and sign the minutes for the meeting held on 24th October 2006

AGREED: the minutes of the meeting were confirmed and signed as a true record with the following amendments:

- 'Dr Amanda Barry' should read as 'Ms Amanda Barry';
- that Ms Amanda Barry's name be included in the list of attendees for the previous meeting;
- that in minute 4 a) 'Guidelines Clearing House'

paragraph 1 should read as follows: '*that the Manchester Knowledge Park was awaiting a response from the Department of Health regarding a request submitted to the Department of Health for funding to establish a Genetics Guidelines Clearing House.*'; and

Paragraph 4 should read as follows: '*that Dr Crolla would circulate to JCMG members any responses that are received regarding the proposal. Dr Cole agreed to inform the CGS Council about the proposal at their forthcoming meeting.*';

- that 'multi-disciplinary' should read as 'Multidisciplinary' in the title of minute 10 i);
- that the title of minute 20 should read as follows: '*NSC consultation document on 'Antenatal Down Screening Working Standards'*'; and
- to delete the following sentence in minute 22: '*The National Genetics Education and Development Centre had conveyed the views of laboratories to the process*'.

Action **Committee Administrator**

4. **Matters Arising on the Minutes**

i) **Guideline Clearing House**

NOTED: that Dr Crolla had written to Professor Dian Donnai regarding the proposal to establish a Genetics Guidance Clearing House but had not received a response. Dr Alison Hill noted that the Department of Health had not received any communications regarding the issue and were not actively pursuing the matter. Dr Hill drew reference to the current financial difficulties being experienced by the NHS which were likely to influence how the Department of Health might view such an initiative.

AGREED: that Dr Crolla would pursue the matter with Professor Dian Donnai.

Action **Dr Crolla**

ii) **NSC's Draft Standards on the Antenatal Screening – Working Standards**

NOTED: (see Agenda Item 14 for discussion).

iii) MetBioNet – letter to Dr Julia Stallibrass

NOTED: that as agreed at the previous JCMG meeting, Dr Crolla would write to Dr Julia Stallibrass at GENCAG about the importance of continuing the work of the MetBioNet in light of the Specialist Commissioning review.

iv) RCP Payment by Results

NOTED: a written report from Dr Helen Stewart, who was representing Clinical Genetics on the RCP / Department of Health Payment by Results liaison group.

Dr Stewart noted that there was a separate process for laboratory molecular genetics within the initiative which was further advanced. In order for clinical genetics to be incorporated into PbR, a complex process of note keeping, coding, HRG (Healthcare resource groups) assignment, within an expert working group (EWG) followed by local calculation of reference costs and the awarding of resources needed to occur. At present there were no tariffs, HRGs or EWGs for clinical genetics.

Dr Stewart was therefore in the process of setting up an EWG for clinical genetics, which would be the first step for clinical genetics to be included in PbR.

JCMG members noted a number of initiatives taking place, which needed to be brought together so they could be fed into Dr Stewart's initiative. These included the contacts between GENCAG and the Department of Health 18 week pathway group, and the work being done by the UKGTN via Jacquie Westwood, relating to molecular genetics. Mrs Norbury noted that molecular laboratory test prices were available on the UKGTN website.

AGREED: that it would be advisable to contact the Department of Health in order to request that a clinical genetics EWG be established. The CMGS and the RCP would also write to the Department of Health to make such a request, copying Dr Alison Hill in on any correspondence. It was important that clinical genetics was not left out of the process, particularly at a time when the process was moving ahead.

It was agreed to invite Dr Jacquie Westwood to deliver a presentation at the next meeting of the JCMG, regarding the work being done by the UKGTN on PbR.

Action Dr Crolla

iv) HER2 – Implementation of testing across NHS laboratories – ‘Evaluating & Introducing new diagnostic tests: The need for a future strategy’.

NOTED: Dr Crolla was pleased to note the positive response from the Department to Health to discuss a proposal from a working party of the Royal College of Pathologists to establish a single national body for the evaluation and introduction of new diagnostic tests.

A meeting had been called by the Department of Health and was taking place at the DoH on 25th January involving Professor Peter Furness, who was leading on this issue for the Royal College of Pathologists, the Department of Health and other invited stakeholders to discuss possible ways of taking the initiative forward. Prof. Martin Bobrow would be attending on behalf of the genetics disciplines.

AGREED: that Dr Crolla would report back to the JCMG the outcome of the meeting.

Action *Dr Crolla*

v) **Medical Specialties Board**

NOTED: Dr Homfray noted that she would be attending the next meeting of the Medical Specialties Board on 13th February 2007, and would report back at the next JCMG meeting.

Action *Dr Homfray*

vi) **Genetics White Paper Review**

NOTED: a copy of the JCMG's response to the three year review of the Genetics White Paper 'Our Inheritance Our Future'. Dr Hill was unable to give a specific timescale for a likely response from the Department of Health to the consultation process.

vii) **RCP College Lectureships 2007**

NOTED: Dr Crolla noted that Professor Veronica Van Heyningen had been willing to be nominated for the 2008 Croonian lectureship but it had not been possible to submit the nomination in time for the deadline for responses. Dr Thompson noted that there were a number of other lectureships for which speakers could be nominated as well as an Advanced Medicines Conference to be held at the RCP in the spring of 2008.

AGREED: Dr Crolla would submit Professor Van Heyningen's name for the other lectureships, as well as for the 2008 RCP Advanced Medicine Conference. Dr Thompson would also inform the RCP.

Action *Dr Crolla*

viii) **RCP Clinical Excellence Awards**

NOTED: Professor Trembath noted that nominations for the RCP Clinical Excellence Awards could be made through the Clinical Genetics Society (CGS).

ix) **NICE committee for hypercholesterolaemia**

NOTED: that Dr Anneke Lucassen had been appointed to act as the British Society for Human Genetics (BSHG) representative on the NICE Committee on hypercholesterolaemia.

5. **Genetics Unit, Department of Health**

NOTED: a written report regarding NHS genetics and white paper commitments. Dr Hill reported that over 50 responses had been received to 3 year progress review of the Genetics White Paper and were being considered by the Department of Health.

There was unlikely to be a radical new approach arising from the consultation or any additional funding but a greater focus on the existing mechanisms. The outcome of the consultation was due to be published sometime in 2007.

The laboratory equipment purchased with the £18 million White Paper investment had now been operational for a while. £2 million capital had also been allocated to genetic laboratories for investment in information technology in cytogenetics and / or molecular genetic laboratories in regional genetics centres in England

Most of the 5 familial hypercholesterolaemia and 7 familial cancer pilots and 10 service development initiatives had been completed or were nearing completion. A second round of Genetic counsellor trainees continued to be appointed in selected centers. The White Paper funded healthcare scientists trainers were working on a range of training issues and many of the agreed third intake of scientist trainees had been appointed.

A competitive selection process to select a University to host the White Paper funded Chair in Pharmacogenetics had been undertaken and a preferred bidder had been selected. Dr Hill was unable to confirm which university had been selected until the university in question had been informed.

6. Reports from the National Genetics Reference Laboratories

NOTED: for information, an update report from the National Genetics Reference Laboratories based at Manchester and Wessex. Dr Crolla asked Dr Hill about the latest position regarding the NGRL laboratories, as both laboratories were still awaiting final decisions regarding future funding. The contracts for the staff were due to expire in a few weeks, and staff at the NGRL (Wessex) had still not been informed in writing about renewal of their employment contracts and consequently some were seeking positions elsewhere. The quinquennial review of both laboratories had been very positive.

AGREED: that Dr Hill would obtain the details about this from the Department of Health and inform Dr Crolla.

Action Dr Crolla

7. Genetics Commissioning Advisory Group

NOTED: a written report regarding UK Genetics Testing Network activities. With regards to the UKGTN dossiers, it was noted that the GenCAG committee had requested that all commissioners should consider funding all the genetics tests on the NHS Directory of Molecular Genetic Testing version 4 which would be available on the UKGTN website shortly.

Coding of genetic diseases was seen as important for Payment by Results and National Tariffs as SNOWMED-CT informed ICD-10 and OPCS coding which in turn informed the Healthcare Resource Groupings which fed into the Payment by Results team for tariff allocation. It was thought it may be possible that the ICD-11 and SNOWMED coding could be considered by the RCPATH.

8. Educational Issues

i) **JCMG Multi-disciplinary Education Group**

NOTED: that new support was being sought to assist with formatting the ELSI material for the website.

ii) NHS Genetics Education and Development Centre (NGEDC)

NOTED: a written report on the NHS National Genetics Education and Development Centre. It was noted that a competence framework for genetics had been prepared by a working party of GPwSIs and NGEDC staff and would be presented for approval by the GPwSIs in January 2007. In addition a UK Workforce Competences for Genetics in Clinical Practice for Non-Genetics Healthcare Staff had been developed by the NGEDC and Skills for Health working together with a wide range of health professionals.

iii) Report from Genetics Counsellor Training Panel

NOTED: written feedback on genetic counsellor training post scheme prepared for the Department of Health 3 year white paper review.

NOTED: that appointment to genetic counsellor posts into the NHS was very competitive. Up to 70 applications had been received after advertisement for individual posts. There were two routes for entry into the posts 1) the scientific route, involving a scientific degree and 2) an experienced based entry and completion of counselling training. The complexity of work done by counsellors, which included deciding whether patients should be referred to a consultant, had increased considerably. Dr Temple gave details of a recent document on the BSHG website:

http://www.clingensoc.org/Docs/WP_AGNC_CGS.pdf

9. National Metabolic Biochemistry Network

NOTED: a written update on current activities of the National Metabolic Biochemistry (Biochemical Genetics) Network. Professor Soothill would be contacting various colleagues who could assist in broadening the scope of the network's guidelines on 'Investigations of hydrops'. Mrs Norbury highlighted the increasing need for greater liaison between genetics and biochemistry, particularly in light of the significant differences in practice between the two disciplines.

It was noted that in response to representations made by amongst others the JCMG, the Department of Health had established a Strategic Advisory Group, chaired by Dr Kieran Morgan. The group was taking forward the planned implementation of strategy relating to Dr Hilary Burton's document 'Metabolic Pathways - Networks of Care'. At present work was concentrating on 1) defining standards of care and 2) describing network functions.

AGREED: that Dr Crolla would contact Dr Jim Bonham about the need for greater liaison between biochemistry and genetics.

Action Dr Crolla, Dr Bonham

10. Manpower and Training

i) RCPATH SAC

NOTED: Dr Crolla noted that there was nothing to report as the issues discussed at the previous SAC meeting were also under discussion by the JCMG.

ii) JCHMT SAC in Clinical Genetics

NOTED: that Dr Davies had been unable to attend the meeting to give the JCMG a report on JCHMT SAC in Clinical Genetics.

iii) Workforce in Clinical Genetics

NOTED: that Dr Holder was unable to attend the meeting but had noted that there had been no change in the situation regarding workforce since the last meeting and would update the JCMG at the May meeting. Dr Holder had noted that the Clinical Genetics Society (CGS) were aware of the current concerns relating to the number of trainees and possible consultant posts which had been discussed at the CGS Council meeting in November 2006. A meeting of the Workforce Review Team was due to be taking place on 26th January 2007.

The shortfall in the number of consultant posts available for the current trainees was seen by JCMG members as a serious problem.

Professor Trembath highlighted the significant number of clinical genetics academic positions that remained unfilled. The Walport initiative had improved the situation somewhat but the issue was still seen as a pressing matter.

iv) Workforce in clinical laboratory scientists

NOTED: a copy of a presentation made by Kim Smith (Chair of the ACC) on training modernisation to the DoH's Skills for Health team. Dr Crolla circulated a copy of a letter sent to Professor Sue Hill, Chief Scientific Officer, to express concerns about the likely impact of proposed re-structuring of pre and post-registration clinical scientists training within the NHS.

Professor Newland had also written to Professor Hill to express similar concerns on behalf of the RCPATH. Further representations could also be made via Dr John Old, RCPATH Vice President, who was acting as the genetics representative on the Life Sciences Task Force which was considering this issue.

11. Presentation by Dr Lyn Chitty on Non-Invasive Advances in Fetal and Neonatal Evaluation Network

NOTED: a presentation by Dr Lyn Chitty giving an overview of the 'Special Non-Invasive Advances in Fetal and Neonatal Evaluation Network' (SAFE) initiative. The aim of the project was to implement routine, cost-effective non-invasive prenatal diagnosis (NIPD) and neonatal screening through the creation of long-term partnerships within and beyond the European Community. The project was funded between March 2004 and February 2009 and involved 50 partners from 19 countries. Key elements of the initiative focused on the isolation and use of free fetal DNA and mRNA. Other initiatives included 'Transcriptomics, proteomics and placental biology'. It was hoped the JCMG could be of assistance in the routine diagnostic implementation of the tests.

NOTED: by JCMG members that the excellent work being done by the SAFE initiative raised the need for a process of dealing with the diagnostic implementation of tests which were currently being offered on a somewhat *ad hoc* basis. It was thought the laboratory and clinical genetics communities needed to look at the process of the implementation of these evolving free fetal nucleic acid technologies into diagnostic genetics laboratories and to liaise with the Department of Health as well as the RCPATH proposals for evaluating new tests.

The composition of a JCMG working group had been agreed at the last meeting (October 2006) to consider the possible integration of these novel technologies into diagnostic Regional Genetics Laboratories and take it forward. The working group was to be chaired by Dr Tessa Homfray who agreed to propose a terms of reference and time frame of the working group for the JCMG's May meeting.

12 **Antenatal screening for cystic fibrosis**

NOTED: that Cystic Fibrosis screening was being rolled out using different DNA testing protocols and an audit was required to monitor performance.

13 **Paper by Dr Stephen Abbs on Disclosure of carrier status in prenatal diagnosis**

NOTED: a document submitted by Dr Stephen Abbs, regarding the different reporting strategies highlighted by a recent UKNEQAS audit of Molecular Genetics Laboratories specifically involving prenatal diagnoses. The document focused on the specific question of whether fetal carrier status should be disclosed at the time of reporting prenatal test results for autosomal recessive (and X-linked) conditions. Dr Abbs reported a marked difference of opinion and reporting strategies between different clinical molecular genetics laboratories concerning whether or not carrier status should be reported on a fetus. Half of the laboratories who responded to the survey did and half did not report the fetal carrier status.

AGREED: that the JCMG were unanimous in the view that there were no grounds on which to withhold reporting the carrier status of the fetus with autosomal and X-linked conditions. Some of the main points raised in support of this conclusion were as follows:

- 1) the patient for whom a report is generated in the context of prenatal diagnosis is the mother and not the fetus in utero.
- 2) The fetal genotype was available under current legislation and could be accessed by the mother at any time.
- 3) Non-disclosure of carrier status may have deleterious consequences, e.g. in populations where consanguineous marriages are common.
- 4) The reporting of the carrier status of the fetus in utero does not guarantee that the information would necessarily be transferred to the child's notes after birth.

Overall, in response to the specific question posed by Dr Abbs, the JCMG recommended that when undertaking molecular genetic prenatal diagnosis the genotype, and hence the carrier status of the fetus in autosomal recessive and X-linked conditions should at all times be reported to the referring clinician and his / her patient.

Dr Crolla would write to Dr Abbs to convey the JCMG's views regarding the paper.

Action *Dr Crolla*

14. NSC consultation document on ‘Antenatal Screening Working Standards’

NOTED: for information, the JCMG’s comments regarding the draft standards on the antenatal screening working standards.

A final statement following this consultation had still not been published by the NSC but it was thought that the recommendation may be that in the absence of a structural ultrasound abnormality, but following an increased screening risk, women should be offered QF-PCR for +13, +18 and +21 rather than karyotyping.

15. Gene patenting and implications for the NHS

NOTED: Dr Temple noted that there were serious concerns amongst genetics laboratories and the wider genetics community regarding gene patenting and its implications for the NHS. Dr Temple gave details of the Medical Innovation Symposium, entitled ‘Enabling Medical innovation through Intellectual Property (IP): from diagnostics to therapy’ due to take place on 1st March 2007.

The meeting was organised by the Patent Office and the NHS National Innovation Centre, and would explore intellectual property issues in regenerative medicine, including genetic diagnostics. Further details regarding the symposium were available from the patent office.

AGREED: that this was an increasingly important issue that the specialist bodies needed to monitor. Dr Crolla agreed to contact Dr Jo Whittaker, who was taking the lead on the issue with regards to molecular genetics, for feedback to the JCMG.

Action: *Dr Crolla*

16. Establishment of Clinical Genetics Society Revalidation Committee

NOTED: Dr Temple noted that as revalidation was becoming a pressing matter, a Clinical Genetics Society Revalidation Committee had been established under the chairmanship of Dr Alan Fryer. The first meeting of the committee was due to be held on 1st March 2007. Dr Fryer would be able to provide reports on the work of the committee if required.

17. Royal Society of Medicine Dinner (December 2006)

NOTED: that due to other commitments neither Dr Crolla nor Professor Trembath had been able to attend the Royal Society of Medicine Dinner in December 2006.

18. Public Health Genetics Unit (PHGU)

NOTED: Dr Burton reported that the existing funding from the knowledge parks for the Public Health Genetics Unit had come to an end. New funding had been obtained from a private endowment which enabled the unit to continue for the next five years albeit on a slightly altered basis. Some cutbacks in staff were expected to take place because of the reduction in overall funding. The unit would continue to report to the JCMG in an observer capacity.

AGREED: Dr Burton would provide a progress report to the next meeting of the JCMG

Action Dr Burton

19. Take-up of NHS funding for Genetics

NOTED: concerns expressed by JCMG members that the NHS funding that had been earmarked for genetics was being diverted to maintain acute services. A significant number of genetics posts were being frozen by Trusts, and consequently, some Genetics Specialist Registrars were finding it difficult to identify vacant consultant posts.

AGREED: that Mrs Norbury would send Dr Temple a copy of letter from Baroness Helena Kennedy (Human Genetics Commission) which had raised similar concerns about this issue.

Action Mrs Norbury

20. Genetics Services for the Deaf

NOTED: a request for assistance from a Department of Health funded project based in Cardiff, in gathering the views of deaf people with respect to their beliefs about genetics counseling and their possible need for their clinical services. The project coordinators had requested the JCMG's assistance in the best possible routes to reaching the genetics community regarding their work.

AGREED: that a representative of the project will be invited to attend the next JCMG meeting to give a presentation on the project's aims and objectives and to receive direct feedback from the committee. It was stressed however, that the JCMG could not provide this or similar project steering groups with representatives.

Action: Dr Crolla

21. Any other business

i) Bearing good witness – proposals for reforming the delivering of medical expert evidence in family law cases

NOTED: a consultation paper produced by the Department of Health entitled 'Bearing Good Witness – Proposals for reforming the delivery of medical expert evidence in family law cases'. The report reviewed the arrangements in place for commissioning medical expert witnesses for the family courts.

It was noted that there was a reluctance to get involved in such work because of the adversary nature of the process and the possibility of complaints being made to the GMC against the expert witnesses.

Professor Newland noted that the RCPATH broadly supported the proposals, seeing the recommendation that such expert witnesses would work in teams as a useful way of avoiding exposing doctors to unnecessary complaints.

AGREED: that Professor Newland would send Dr Crolla a copy of the RCPATH's response to the consultation though it was suggested that it would be useful for the JCMG

to make its own separate response. The need for appropriate training support for such a system and for the costs of such witnesses to be covered by the courts rather than the litigants were points that would need to be highlighted in any such response.

Dr Crolla would draft a response and circulate to the JCMG before forwarding to the RCP for submission to the Department of Health.

Action **Dr Crolla**

ii) Human Fertilisation and Embryology Act HFEA

NOTED: proposals for the revision of the HFEA had been published recently in the form of a White Paper. The main proposal was the establishment of a new regulatory body (RATE) which would be formed from the current HFEA and Human Tissue Authority. Alison Hall at the PHGU had published a helpful commentary on this White Paper which could be viewed at: http://www.phgu.org.uk/ecard?link_ID=2970.

iii) Connecting for Health access to the internet for geneticists

NOTED: information from Dr Crolla about the efforts being made by Connecting for Health to improve internet connection to various genetics internet websites. The recent upgrade from 250 Mb to 1 Gb/sec had significantly improved to genome web-browsers but CfH were still considering the provision of a direct portal from the Ensembl database to the NHS N3 so NHS users would not need to connect to Ensembl via the www. CfH was also looking at providing dedicated bandwidth for specified “business critical” URLs so that connections were not dependent on total NHS internet usage.

iv) Recommendation for honours

NOTED: Professor Thompson noted that specialist societies could submit recommendations for the honours system via the RCP. Any nominations should be sent to the RCP President.

v) Publication

NOTED: Publication (August 2006) and circulated in November 2006 of the PHGU/UKGTN Working Party Report “Evaluation of the use of array comparative genome hybridization in the diagnosis of learning difficulty”. The PDF was downloadable from:

http://www.phgu.org.uk/pages/work/serv_projects.htm#evaluation

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Dates of forthcoming meetings (at Royal College of Pathologists)

NOTED: Thursday, 15th May 2007 at 10:00am (*Please note earlier time which has been arranged because several JCMG members will be traveling to Southampton for a long term commitment to a BSHG “Awayday” meeting later that day*)

Thursday, 23rd October 2007 at 11:00am