

Joint Committee on Medical Genetics

The Royal College of Physicians

The British Society for Human Genetics

The Royal College of Pathologists

RCP, 11 St Andrews Place, Regents Park, London NW1 4LE

A meeting of the Joint Committee on Medical Genetics was held at the Royal College of Physicians on Tuesday, 17th May 2005.

Present:

Dr Heather Skirton JCMG Chair, (BSHG)

Dr Hayley Archer	-	RCP, Trainee representative (<i>part meeting</i>)
Dr Rodney Burnham	-	RCP, Registrar
Dr Hilary Burton	-	Observer, PHGU
Dr Trevor Cole	-	RCP
Mrs Michelle Collyer	-	RCP, Patient and Carers Network
Dr John Crolla	-	RCPPath
Dr Teresa Davies	-	RCPPath
Dr Helen Firth	-	In attendance (<i>part meeting</i>)
Dr Anne Green	-	RCPPath
Dr Hilary Harris	-	RCGP
Dr Edgar Hau	-	In attendance
Dr Shirley Hodgson	-	BSHG
Dr Tessa Homfray	-	RCP
Ms Dianne Kennard	-	Observer, DH
Mr Alastair Kent	-	GIG/RCP Patient and Carer Network (<i>part meeting</i>)
Dr Helen Kingston	-	RCP, JCHMT SAC Chair
Professor Julian Sampson	-	Chairman, BSHG
Professor Peter Soothill	-	RCOG
Ms Su Stenhouse	-	BSHG
Dr Karen Temple	-	BSHG
Mr Simon Land	-	Committee administrator

1. Apologies for absence

- Apologies were received from Dr Mark Bale (Observer, DH), Professor Carol Black (RCP, President), Dr Sally Davies (Observer, Wales and Workforce), Dr Sian Morgan (RCPPath Trainee representative), Dr Ruth Newbury-Ecob (RCPCH), Dr Tony Parkin (BSHG), Dr Allison Streetly (NSC, Observer), Dr Virginia Warren (FPH) and Dr Helen Williams (RCPPath, Registrar).

2. Membership

- The Chairman welcomed Dr Karen Temple as a new BSHG representative, replacing Professor Farndon.
- The Chairman welcomed Dr Edgar Hau a visiting Clinical Geneticist from Hong Kong, as a one off observer.
- Dr Crolla stated that Dr Gail Norbury would be replacing Dr Abbs as an RCPATH representative and that a Trainee representative to replace Dr Williams was being sought.
- The Registrar stated that the RCP Nominations Committee would consider a replacement from Dr Fryer at a meeting in June 2005.

3. To confirm and sign the Minutes for the meeting held on 13th January 2005

- After amendments the minutes of the meeting were confirmed and signed as a true record.

4. Matters arising on the Minutes

4.1 Length of time for storage of DNA samples

- The Chairman reported that a letter on the issue had been sent to Professor Peter Furness. Subsequently, comments from the JCMG had been invited which had been fed back by Dr Crolla.
- Dr Crolla stated that the revised RCPATH Guidelines were now up to date with both the Human Tissue Bill and the Consent and Confidentiality report. The Guidelines were at this stage in the hands of RCPATH lawyers but were due for publication in June 2005 (to be available on the RCPATH website). The committee was pleased that the guidance on the storing of diagnostic (clinical) samples had been clarified. Namely, that they should be stored indefinitely (at least 30yrs by the RCPATH definition).
- It was agreed that Dr Crolla would circulate the Guidance once published.

ACTION: Dr Crolla

4.2 Chairmanship

- The Chairman congratulated Dr Crolla who had been nominated by the RCPATH as the new Chairman (effective 1st January 2006).

4.3 Genetic Community I.T. perspective

- Members noted an e-mail **Doc 05/03** from Andrew Devereau, which stated that representation from the Home Countries was welcome at a workshop to discuss the draft OBS on 9th June 2005. It was agreed that Professor Sampson would seek individuals to attend from Scotland, Wales and Northern Ireland and then communicate this to Andrew Devereau.

ACTION: Professor Sampson

4.4 Input into antenatal screening for cystic fibrosis

- The Chairman reported that there had been no further progress on the issue.

4.5 Involvement in RCP Public Open Day – 9th July 2005

- The Chairman expressed concern that little action had been progressed regarding the specialties representation at the Open Day.
- It was noted that GIG would have stand and Mrs Collyer volunteered to contribute to the genetics stand.
- Dr Burton agreed to raise the issue on the Educational Group agenda of the Knowledge Parks. It was noted that it was an extremely busy time for the Knowledge Parks but it was hoped that a display on their remit could be made available.
- Dr Cole felt that the CGS stand that was used at the BMA could be re-used and that Fiona Hart and Paul Brennan should be contacted regarding this.
- Dr Burton and Professor Sampson agreed to progress the issue and report back to the Chairman and administrator via e-mail. Other comments and suggestion from members were also invited.

ACTION: Dr Burton/Professor Sampson/All

4.6 Specialty Webpage on the RCP Website

- It was noted that the page had now been updated with links suggested by members. The committee was invited to contact the administrator if more suggestions came to mind.

ACTION: All

5. Genetics Branch, Department of Health

5.1 White Paper

- Members noted the pre-circulated and tabled update paper **Doc 05/03** and Ms Kennard invited questions from members.
 - The Registrar asked what training would be available to GPwSI and whether PCTs were signed up to the agreement. Ms Kennard stated that within genetics GPwSI would be designing their own training programmes in collaboration with PCTs. The Genetics Education Centre would also provide some central co-ordination for this. It was noted that GPwSI in genetics were primarily a training/co-ordination role and that the framework had been previously signed off by RCGP and RCP. Without the backing of PCTs, GPwSI would not have been allocated.
 - Dr Crolla asked what the strategic plan was with regard to the OBS Workshop (June 2005) and the distribution of White Paper funding for I.T. Ms Kennard stated that the OBS went beyond the laboratories and will form a backdrop for the spending of the £1 m capital committed under the White Paper for laboratory I.T. Moves were also being made to ally OBS to NPfIT (now renamed NHS Connecting for Health). Consultants writing the OBS were in contact with NPfIT but as yet NPfIT was not addressing issues at the level of detail required in genetics. Dr Crolla felt that delays were negatively affecting laboratories and asked whether the national procurement of I.T. issues for genetic laboratories would come to pass. Ms Kennard appreciated that the community was presently in limbo and agreed to follow up the issues with suppliers and report back.
 - **ACTION: Ms Kennard**
 - Dr Harris asked how far the OBS would link to General Practice. Ms Kennard stated that this was not within its scope but felt that NPfIT should eventually cover this area.
- For information Ms Kennard tabled a report from HM Government “Concordat and Moratorium on Genetics and Insurance”.

6. Expensive Drug Therapies

- Mr Kent stated that NICE had now published its draft report to stakeholders. The consultation period would then close on 27th May 2005 and NICE would publish its advice to DH. The issue had arisen from a request by DH to see whether existing NICE methodologies were applicable to Ultra Orphan therapies (classified as < 1000 patient in England and Wales). As a test, Gaucher Type I had been chosen as its cost per patient per annum was £80,000 and Chris McKay

from Sheffield had undertaken the appraisal. The following problems were discovered in NICE methods when applied to the disease:

- Scales – NICE had 70 scales none of which were adequate enough for the disease and there was a need to formalise existing informal scales used for rare diseases.
 - Evidence base – A crucial finding was that the evidence base required for an effective NICE appraisal was lacking in the case of Gauchers (and therefore many other rare diseases as Gauchers was relatively well researched). The available research evidence would be considered only partial in aspect and variable in quality.
 - Under current NICE guidance, expensive therapies for rare diseases were unlikely to be sanctioned. This will adversely affect future drug development. However, people did not choose their disease and it would therefore fall back to Parliament, DH and NHS to decide on the treatments that would be made available.
 - The scope of NICE appraisals was criticised as it made no recognition of the complexity of disease, its severity or impact on families.
- The Chairman stated that members were concerned by the lack of geographical parity, as Scotland had separate guidance and Wales is not compelled to implement the recommendations.
 - Mr Kent also raised the issue that NSCAG currently made decisions on funding for Gauchers (and would do so for 2 years) but that this did not apply to all rare diseases. Members noted that PCTs funded NSCAG (with monies being reimbursed after 2 years) and they were increasingly alarmed by the prospect of paying for expensive therapies.
 - Professor Sampson was wary of the terminologies used, as not all rare conditions are expensive to treat. There was also disparity between definitions as Europe classified an orphan disease as one affecting 5 in 10,000 and NICE classified ultra orphan as affecting less than 100 people in England and Wales.
 - Professor Soothill expressed concern that assessments for every rare disease would be costly and were likely to indicate that more research was required consequently pushing up drug prices further. Mr Kent indicated that the feasibility study was unlikely to recommend further research.
 - Overall, members felt that this issue would become increasingly important and agreement was required on future research. Discussion was needed with manufacturers at the designation stage of ultra orphan conditions, rather than waiting for the marketing authorisation stage. It was therefore agreed to keep this item on future agendas.
- ACTION: *Committee administrator***

7. Appropriate services for adults with inherited metabolic diseases

- Members noted an update **Doc 05/04** and Dr Burton reported on the following issues:
 - A huge commitment from service providers had been seen in undertaking the review of current services and this exceptional effort required thanks.
 - Workload was substantial with over 1000 new diagnoses per annum.
 - A complex service was required in terms of biochemistry and dietetics and also interactions between genetics and other specialties.
 - Geographically the area was not covered well. Only 5 centres with full team back up (dietetics and specialist nursing support) existed, 3 of these being in London.
- The plan was to finish the assessment of the current service, refer the report to the BIMDG and then to the JCMG and Specialist Commissioners Group.
- Dr Green reported that 2 questionnaires to the biochemistry network were also being processed with the view to a comparison between the existing service and a comprehensive ideal. This had already stimulated activity in regions with a sparse coverage.
- The Chairman congratulated Dr Burton and Dr Green on their efforts and asked the committee what could be done to increase training and consultant posts within IMD. Dr Green stated that a commitment to IMD sessions was the greater problem as trainees were in place but without more consultant posts they would not specialise in the area. The Registrar advocated a two-pronged approach at increasing both consultant and trainee posts via the Workforce Review Team (WRT) led by Judy Curzon and pressure on the DH via the PRCP. It was noted that Debbie Hilder of the WRT sat on the committee looking at the issue and it was agreed that the Chairman would write to the PRCP flagging the issue following the next IMD meeting on 15th July 2005.

ACTION: *Chairman*

8. Genetics Commissioning Advisory Group (GenCAG)

8.1 Update

- Ms Kennard stated that GENCAG had not met since September 2004 and members noted a short update **Doc 05/03**.
- GENCAG had devised set of quality markers, which were due for approval in late May 2005. Ms Kennard agreed to make certain that this information was fed into NPfIT.

ACTION: *Ms Kennard*

8.2 UK Genetic Testing Network

- Members noted a tabled update **Doc 05/03** and it was agreed that Dr Temple would provide reports on the item at future meetings.

ACTION: Dr Temple

9. Human Tissue Bill

- Members noted the updates provided by the DH **Doc 05/05 a,b,c**.
- The committee discussed whether the information should be disseminated (particularly the need for establishments undertaking licenced activities to register their interest with the HTA) via the BSHG. Dr Crolla urged the committee to wait until the HTA issued its guidelines as the law did not formally change until April 2006 and then there followed 3 months grace. It was therefore agreed to defer action on this issue.
- Dr Crolla reported a worrying development in that the revised Coroners Rules would come into effect before the Human Tissue Act. Under Coroners Rules the disposal of patient tissue would be required if specific permission to store tissues had not been obtained by the Coroner, but this would be against good clinical practice and the best interest of the patient. The RCPATH welcomed changes in the Coroners Rules but wished to defer their implementation so as to not create confusion. It was agreed that the JCMG should add its voice to this sensitive issue and therefore the Chairman would write to the Presidents of RCP and RCPATH recommending a private Presidential briefing. The letter would also be copied to the CMO for information.

ACTION: Chairman

10. Mental Capacity Bill

- Members noted the update provided by Dr Bale **Doc 05/06** and the Chairman urged individuals to respond to the consultation due in to be held in the summer of 2005.

11. Educational issues

11.1 Multi-disciplinary training opportunities panel

- The Chairman reported that an effective meeting had taken place of the group and members noted an update report **Doc 05/06**.

11.2 NHS Genetics Education and Development Centre

- The administrator agreed to contact Professor Farndon for an update on the progress of the Centre.

ACTION: *Committee administrator*

12. Reports of the work of the JCMG in progress

12.1 UK Haemophilia Centres Genetics Working Party

- Members noted the update provided by Dr Fryer, which outlined the revised remit and detailed that the guidance had now been published in the journal *Haemophilia* **Doc 05/07**.

12.2 Consent and Confidentiality Working Party

- It was noted that the document had been considered by the RCP and RCPATH Councils in March 2005. Professor Farndon was working on the comments received.

12.3 Training Posts for genetic laboratory scientists

- Members noted the report compiled by Ms Stenhouse **Doc 05/09**.
- Dr Green stated that a lead Biochemistry Trainer was now in place.
- Ms Kennard reported that the vocational MSc was now fixed and this would reduce pre-registration training from 4 to 3 years. Work was being progressed on the competency frameworks by the Skills for Health team.
- Dr Crolla was concerned that not enough support would be available to those trainees coming through the present system. In particular, both molecular geneticists and cytogeneticists are concerned about the slippage in the timeframe for the introduction of the proposed “generic” clinical scientist MSc (Sue Hill, DoH). It is proposed that this MSc, which will form part of the A Grade training will become a statutory requirement for HPC State Registration. However, this plan was first proposed two years ago and to the best of his knowledge, Dr Crolla was aware that project planning will only begin in June this year. Furthermore, the urgent issue of provision of funding for higher specialist training (i.e. post A Grade/MSc) is not being addressed nationally although there have been some local success stories.
- Other members felt that modules of the MSc should be made available in Regional Genetic Centres, not just Higher Education Establishments.

- It was agreed that Ms Kennard would approach Sue Hill to put together a small discussion group to deal with some of these issues.

ACTION: Ms Kennard

12.4 Genetic Counsellor Training Post Panel

- Members noted the update from the Chairman **Doc 05/09**.

13. Learning Disability Guidelines

- Members noted the Guidelines “ A guide to the investigation of children with developmental decay in East Anglia” **Doc 05/10** and the tabled Parents’ guide.
- Following a presentation on the background and formulation of the guidelines by Dr Helen Firth (who was thanked by the Chairman), members were invited to ask questions and suggest ways forward for the work. The following Action Plan was agreed.

Dr Burton/Dr Firth to check with their team that they wished to proceed as outlined below.

ACTION: Dr Burton/Dr Firth

Look for approval and comments from the Genetics Community. Add the guidelines etc to the BSHG website as a formal consultation. Professor Sampson, Dr Burton and Dr Firth would liaise on this.

ACTION: Professor Sampson/Dr Burton/Dr Firth

Dr Temple to take to the documentation to the CGS for endorsement.

ACTION: Dr Temple

Professor Soothill agreed to put the Chairman in contact with individuals at the RCOG (who had put together Greentop Guidelines) with a view to possibly expanding out the work so far and producing a full guideline.

Dr Green alerted members to work she had contributed to on Biochemical testing which had been undertaken with the RCPC. She agreed to share this with the East-Anglian team.

ACTION: Dr Green

The team should consider the consultation of GP’s. Dr Harris felt this would best be done via e-mail.

Overall, members felt the work was very useful and would like to see this expanded out to a full guideline. Eventually it was thought that this would best sit at the RCPCH.

14. National Genetic Reference Laboratories

- Members noted the update reports **Doc 05/10**.
- Dr Crolla stated that a Stakeholder meeting had been called for 21st June 2005 to discuss the past and future of the Reference Laboratories. It was thought particularly important to formulate a strategic long-term view, as the Laboratories would soon be seeking re-financing (presently 5 year funding). The Chairman stated that she would be attending the meeting and asked members to contact her if they wished to express any views.

ACTION: All

15. Workforce and training

15.1 RCPATH SAC

- Dr Crolla reported that the SAC had now been renamed (Genetics and Clinical Embryology SAC) to incorporate the embryologists until they had a large enough membership to form their own SAC.

Numbers taking and passing RCPATH I 2004

Cytogenetics - 18 sat, 10 passed.

Molecular Genetics – 10 sat, 7 passed.

Numbers taking and passing RCPATH II 2004

Cytogenetics – 3 sat, 3 passed.

Molecular Genetics – 4 sat, 4 passed

- It was noted that the SAC had been discussing higher specialist training, RCPATH Guidelines for the storage and retention of tissues and the ACC Prenatal Retrospective Audit: The Clinical Value of Prenatal Karyotyping for 'Down Screen Positive' women, which was due to be published in the Lancet in June 2005. This was tied to the NSC Antenatal sub-group publication on the same subject recommending only QF-PCR detection of trisomies 13, 18 and 21 that had caused much debate.

15.2 JCHMT SAC in Clinical Genetics

- Dr Kingston reported that PMETB was due to 'go live' on 30th September 2005 and that a new Chairman had been appointed. Concern was expressed that

planned JCHMT visits used to quality assure training programmes would not go ahead under PMETB unless there were major problems. JCHMT visits would be replaced by PMETB visits to Deaneries which would assess all specialties at the same time. Negotiations were on going as the SAC felt that this method of assessment would not be sufficient to test training at centres where the last visit took place sometime ago (i.e. trainers and SpRs have since left). The SAC was also concerned that UK trainees would be disgruntled that they would still require MRCP(UK) whereas some overseas graduates (particularly applying under Article 14) may not.

- Dr Kingston stated that MMC would likely make it harder to choose people at an early stage as basic training could be significantly changed and specialties would be allocated before the award of Membership. T
- The Academic Subcommittee of the Modernising Medical Careers and UK Clinical Research Collaboration had produced a welcome report that contained recommendations on increasing academic placements within F2. It can be found at:

http://www.mmc.nhs.uk/academic_medicine.asp?m=10

15.3 Workforce in Clinical Genetics

- Members noted an e-mail update from Dr Davies **Doc 05/12**. Further discussions on Workforce took place under Item 17.

16. Consultations and Correspondence

- Members noted the JCMG nominations **Doc 05/13** to sit on the Payment by Results Panel. The Chairman agreed to pass a further 2 names to the administrator.

ACTION: *Chairman*

- Members noted the JCMG nominations **Doc 05/14** to sit on the RCP Ethical Issues in Medicine Committee.
- It was agreed that Professor Soothill would co-ordinate the JCMG response on the Nuffield Council on Bioethics' consultation. The administrator would e-mail members the link and details of how to respond.

ACTION: *Professor Soothill/Committee administrator*

- Members noted the JCMG comments **Doc 05/15** on Shared Decision Making that had been forwarded to the Patient and Carers Network.
- Members noted the Chairman's response **Doc 05/16** to a letter from the RCP Academic Registrar asking for details of charities that had funded specialty

training in the past. This was with a view to potentially offering joint funded (RCP and charity) Clinical Research Fellowships. Members offered the following:

- Birth Defects Foundation
 - Genetic Research
 - Action Research
 - Wellbeing
 - Breakthrough Breast Cancer
 - Wales Research and Development
- Professor Sampson agreed to further research the issue and make contact with GIG and members were asked to forward other suggestions via the Chairman.

ACTION Professor Sampson/All

17. Funding to Deans for “small specialties”

- Members noted a document “Funding for Foundation Programmes” **Doc 05/17**.
- Dr Kingston spoke to the paper and stated that funding had been allocated for 10% of trainees to spend more time in small specialties (which included genetics). Funding for 5% of trainees to embark on an academic F2 was also available. Members felt that it was for RSAs to discuss this with Deans.
- It was noted that there were geographical problems with trainees generally not wishing to relocate to take up a consultant position and that there were grave concerns over the number of consultant posts that would be available to trainees due to gain CCST in the next 2 years.
- Dr Kingston stated that 20 trainees were due to complete CCST in 2006 and 21 in 2007. Dr Temple agreed to voice the concerns at GENCAG that these individuals may not be able to obtain appropriate posts after completing training.

ACTION: Dr Temple

- Other issues that were noted:
 - Less people were being introduced to academic careers
 - A suggestion that half-day educational slots covering areas outside the curriculum should be formulated.
 - That SpRs with less than 6 months left in post could be utilised and perhaps this period extended.

18. Any other business

18.1 E-mail from Allison Streetly

- Dr Homfray wondered whether the prenatal tests had been refused due to funding or because they were not available. It was agreed that the Chairman would contact Dr Streetly to clarify the position.

ACTION: *Chairman*

18.2 Letter from the RCP Registrar

- The Chairman reported that she had received a letter from Dr Burnham regarding Cardiopulmonary resuscitation and would be responding shortly.

18.3 PHGU - Microarray Review

- Dr Burton reported that the PHGU was to start a one-year review of Microarray in July 2005 and that there would be a call for evidence givers.

18.4 SAC Chairmanship

- Dr Kingston informed members that her term as SAC Chair (and consequently JCMG member) had now expired. It was noted that Dr Sally Davies would replace her at the SAC. The Chairman thanked Dr Kingston for her hard work.

18.5 PRCP briefing report

- Members noted the report from the PRCP **Doc 05/18**.

19. Dates of the next meetings

- It was agreed to change the Autumn 2005 meeting date from:
11.00hrs, Thursday 20th October 2005 to **11.00hrs, Tuesday 8th November 2005**