

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 Thursday, 20 May 2004 at 11am

Present:

Dr Heather Skirton JCMG Chair, BSHG

Dr Hayley Archer	-	RCP, Trainee representative
Dr Hilary Burton	-	Observer, PHGU
Dr Trevor Cole	-	RCP
Dr John Crolla	-	RCPATH
Dr Sally Davies	-	Observer, Wales
Dr Alan E Fryer	-	RCP
Dr Anne Green	-	RCPATH
Professor Shirley Hodgson	-	BSHG
Dr Tessa Homfray	-	RCP
Ms Dianne Kennard	-	Observer, DH
Mr Alastair Kent	-	Genetics Interest Group
Dr Helen Kingston	-	RCP, JCHMT SAC Chair
Dr Ruth Newbury-Ecob	-	RCPC
Dr Tony Parkin	-	BSHG
Professor Julian Sampson	-	Chairman, BSHG
Professor Peter Soothill	-	RCOG
Ms Su Stenhouse	-	BSHG
Dr Allison Streetly	-	In attendance
Dr Virginia Warren	-	FPH
Mr Simon Land	-	Committee administrator

1. Apologies for absence

- Apologies were received from Dr Stephen Abbs (RCPATH), Professor Carol Black (RCP, President), Professor Peter Farndon (BSHG), Professor Ian Gilmore (RCP, Registrar), Dr Helen Kingston (RCP), Dr Sian Morgan (RCPATH Trainee representative), Dr Maggie Williams (RCPATH, Trainee representative).

2. To confirm and sign the minutes for the meeting held on 13 January 2004

- The following sentence was added to minute 5.2

The National Metabolic Biochemists Network has been funded by the DOH for 3 Trainer posts (or equivalent) and 8 Higher Specialist Training posts (Grade B Clinical Scientists).

- Following this amendment the minutes of the last meeting were confirmed and signed as a correct record.

3. Membership

- The Chairman welcomed Dr Tony Parkin (BSHG) and Ms Su Stenhouse (BSHG) as new members of the committee. Dr Allison Streetly was also welcomed who had been invited to give a presentation on Haemoglobinopathy screening.
- The Chairman reported that a letter had been sent from the Ian Gilmore to the RCGP requesting a replacement for Dr Rydian Hapgood. A reply was awaited.

4. Matters arising on the minutes

4.1 Copying of Laboratory Reports

- Dr Crolla reported that he had prepared and was due to forward a submission to the BSHG Bulletin stating that CPA guidelines cover the issue. It would however invite submissions from individuals where problems occur. It was agreed that he would also forward this to Dr Kingston for information.

ACTION: Dr Crolla

4.2 Contract Currencies

- Dr Parkin asked for clarification on the exact changes (and timeframe) that would be required regarding Cytogenetic currencies. Miss Kennard stated that at present Cytogenetics included other factors (consumables) on top of Workload Units. This issue would be progressed via a different forum, outside of JCMG.

5. Screening

5.1 Haemoglobinopathy screening

- The committee viewed a presentation on antenatal and neonatal screening in Haemoglobinopathy presented by Dr Allison Streetly (Director/Co-ordinator, NHS Sickle Cell & Thalassaemia screening programme).

For the information of the Committee, the presentation has been annexed to these Minutes at Appendix I.

- Members discussed the aims and visions for future Haemoglobinopathy screening as below:
 - Dr Homfray, posed a question about how early in pregnancy health professionals in primary care could interact with couples about Haemoglobinopathy screening. Dr Warren highlighted the work that the NHS HTA programme is commissioning in the area and agreed to forward more information.

ACTION: Dr Warren

- Members noted the lack of Asian counsellors and nurses. There was a need to employ people with the relevant language skills or interpreters. Mr Kent stated that GIG would produce a lexicon of agreed terminology in different languages that would help prevent misunderstandings by those involved. He agreed to forward this to Miss Kennard for information when complete.

ACTION: Mr Kent

- Professor Soothill asked whether the Royal College of Midwives had been engaged, as it was their members who would need to implement any change. Dr Streetly confirmed that a dialogue had been arranged via a conference in June 2004.
- Dr Cole asked whether GPwSIs would be useful in promoting the screening changes. Miss Kennard reported that as their number were likely to be in the region of 10 then they would be more of a strategic post.
- The Chairman asked how the JCMG could help with the process. Dr Streetly and members felt that a member of the National Screening Committee (NSC) would be a useful addition to the committee. It was agreed that the NSC would be approached to nominate a suitable representative to join JCMG to provide an interface.

ACTION: Chairman/Committee administrator

5.2 Prenatal screening

- Dr Parkin reported that the ACC were concerned by the publication of a new standard which appeared in appendix 3 of the Annual Report 2002/03 for the National Down Syndrome Screening Programme for England (completed December 2003). The standard stated that a minimum of 95% of diagnostic (ie. cytogenetic) results should be available by 14 calendar days of receipt of the specimens. This standard was set without reference to actual data; nor with the

knowledge of the Association of Clinical Cytogeneticists as the relevant professional body. This exposed the current lack of linkage between screening and diagnostic programmes. The ACC and other professional bodies would therefore welcome a formal dialogue with the NSC.

6. Appropriate Services for adults with Inherited Metabolic Diseases

- Dr Burton informed members of a meeting held between the PHGU, James Leonard, Phil Lee and Graeme Shortland, where it was agreed that the PHGU would provide operational support for a scoping exercise. The aim would be to present to Commissioners the problems and possible solutions with 'buy in' from the DoH and JCMG.
- It was agreed that a high level key stakeholder meeting would be convened in the first instance to discuss the issue. This would include representatives from the JCMG, its parent bodies, DoH, PHGU, GIG, FPH, Specialised Service Commissioners and the Workforce Numbers Advisory Board. Miss Kennard also recommended that a prescribing expert should also be involved to oversee guidelines by centers on prescribing.

ACTION: Committee administrator, Chairman

- The committee also discussed Enzyme Replacement Therapy and other expensive drug treatments and how they would fit into any framework. Mr Kent stated that there were currently 30 authorised/approved products with high prices and that this number was increasing. It was felt that this trend needed to be addressed by PCTs and that the subject warranted further discussion. An item would be added to the next agenda for this purpose.

ACTION: Committee administrator

7. Human Tissue Bill

- The Chairman introduced the item by stating that a lot of activity had occurred after the last meeting of JCMG. This included the composition of practical scenarios where the Bill (as it stood) would diminish clinical services. Dr Crolla and Ron Zimmern (PHGU) were both thanked for their work on this difficult area.
- The committee received a briefing on the Bill from the RCP. Dr Warren asked whether this was currently within the public domain. The committee administrator agreed to ascertain the position.

ACTION: Committee administrator

- Dr Crolla reported that there was enormous consensus across disciplines on the impracticality of the Bill. However, he was reassured that the Government were

now listening constructively to concerns. The Lords were also being lobbied so there would be opportunities for further amendments at a later date.

- Dr Crolla raised the issue of criminal records being possible under the Bill. These could see people losing their state registration from the Health Professions Council where certain procedures, which were previously considered good practice, could now be considered criminal.
- Mr Kent had attended a meeting of lawyers and social scientists, which highlighted the power of professional bodies over the Courts. The meeting concluded that:
 - Procedures may be stopped under the Bill, which are in effect still legal.
 - Trusts were unlikely to issue legal advice to healthcare workers.

8. Reports of the work of the JCMG in progress

8.1 Consent & Confidentiality Working Party

- The Chairman reported that the report was on hold, awaiting any amendments to the Human Tissue Bill.

8.2 Training posts for genetic laboratory scientists

- The committee noted the report on training posts for genetic laboratory scientists provided by Dr Abbs.
- Ms Stenhouse reported back from the CMGS Head of Laboratories meeting. They favoured “on the ground” trainers rather than the proposed regional trainers and wished to know whether the funding was flexible enough to accommodate this. They also expressed the difficulties incumbent of trying to find a National Training Co-ordinator with the breadth of knowledge to cover both cytogenetics and molecular genetics. A part time post (with time spent in the laboratory) in each discipline was preferred.
- Dr Parkin stated that the ACC would wish for the Trainer roles (3 for each discipline) to be full time posts. Dr Crolla felt that the strategic training posts should reflect the shift from cytogenetics to a molecular/cyto hybrid. Members noted that the Wessex Reference Laboratory had arranged a meeting to look at the issue and Dr Crolla agreed to pull this together with work being progressed by Professor Sampson regarding the delivery of a new service where R&D were funded.

ACTION: Dr Crolla/Professor Sampson

- Dr Green stated that the post for Head of Metabolic Training had now been advertised. The 8 posts for trainers would be advertised in July with a view to appointments being made in October.
- Miss Kennard confirmed that a working group had been convened by Sue Hill to place grade A training within a vocational MSc. She also expressed the DoH thanks to all the professional bodies involved in the above processes.

8.3 UK Haemophilia Centres Genetics Working Party

- Dr Fryer stated that the report had now been disseminated to all genetic, comprehensive care and haemophilia centers. A shortened version of the report would also be sent to the Journal – Haemophilia for consideration and publication. It was noted that the Working Party and report was just the beginning and that the Chairman (Professor Chris Ludlam) would be making recommendations for reform of the Working Party to continue via the Advisory Committee at a meeting in June 2004.

8.4 Genetic Counsellor Training Post Panel

- The committee noted the update provided by the Chairman. It was noted that the last sentence of the first paragraph contained an error. It should read:

“Nineteen trainees have been appointed and more posts are to be filled this year.”

9. NICE

9.1 NICE Familial Breast Cancer Guideline Development

- The committee noted the update provided by Professor Hodgson and that a delay in publication was now expected due to a clash with local and European elections. It was also noted that the draft guidelines would have no funding attached.
- Members questioned whether the information provided was the most recent draft as Miss Kennard stated that GenCAG had recently reviewed the 2nd version, which differed. Dr Cole agreed to look into this and report back.

ACTION: Dr Cole

9.2 Research and Development Strategy Consultation Document

- The committee noted the RCP response to the consultation document.

10. Educational Issues

10.1 Genetic Education: Needs and Evaluation (GENE)

- Members noted a tabled e-mail from Professor Farndon that stated that the final report was being written and should be available at the next JCMG meeting.

10.2 Multi-disciplinary course in formal genetics

- The Chairman reported that the planned meeting had been delayed while awaiting the bids for the Genetics Education Centre. It was hoped that this could now be convened on 16th July 2004. Representatives from the professional bodies agreed to forward the names of attending representatives to the Chairman.

ACTION: Professor Farndon (via Professor Sampson), Dr Parkin, Ms Stenhouse.

11. Genetics Commissioning Advisory Group

11.1 Update

- Miss Kennard updated members on the GenCAG meeting held 18th May 2004:
 - GIG gave a presentation on ethnicity and genetics.
 - GenCAG had seen reviews form all the Home Countries bar Scotland
- Professor Farndon had forwarded the following e-mail regarding a point raised at GenCAG:
 - At GenCAG on Tuesday an interesting point was raised over the time for which DNA samples should be stored. As you know, there seems to be a general assumption that they will be kept indefinitely but this may be untenable as numbers grow. We say this, I believe, because no professional group has ever considered this matter and suggested guidelines. Other European countries have time limits, apparently. Perhaps this is something that the JCMG might like to consider
- Members felt that this discussion should await the publication of the Human Tissue Bill. It was therefore agreed to include this as an item on the next agenda. In the meantime members would research whether other European countries had guidelines on the issue.

ACTION: Committee administrator, All

12. Genetics Branch, Department of Health

12.1 White Paper

- Miss Kennard highlighted that 2 reports had been commissioned by the Manchester Genetics Reference Laboratory, to review the following IT issues:
 - Informatic needs across the service
 - Survey of IT in Genetic Centres

12.2 GPwSIs

- The committee noted that the RCP President and Senior Officers had found the GPwSI in Genetics draft extremely impressive. The Chairman recorded the JCMGs congratulations to Dr Hill who had compiled the draft. Miss Kennard informed members that it would now be forwarded to the Tsar and Ministers for “final sign off” before implementation began.

13. National Genetic Reference Laboratories

- Members noted the reports submitted by the Heads of the Manchester and Salisbury Laboratories. Dr Crolla stated that the strategic mid-term review of the laboratories by the DoH was due to take place on 18th June 2004.

14. Manpower and Training

14.1 RCPATH SAC

- Dr Crolla reported that the SAC was not due to meet until the following week. A full report would be made at the next JCMG
- Dr Crolla stated that he had been asked to speak at the RCPATH annual meeting in September. Members congratulated Dr Crolla on this.

14.2 JCHMT SAC in Clinical Genetics

- Dr Davies reported that the SAC had met the previous week and discussed Clinical Genetics training with regard to Modernising Medical Careers and some minor amendments to the 2003 curriculum.

14.3 Manpower

- Dr Davies reported that there would be no new centrally funded posts for the next year. However, an agreement for 5 conversion posts had been reached which would be made available to centers that had the educational approval and required funding.
- Dr Davies stated that a database of SpRs was to be set and monitored up by Alex Murray (SpR, Cardiff).

- Dr Davies reported that under half of all Clinical Genetic Consultants in England (120 in total) were working without a full commitment to the NHS. This was due to academic posts (34 out of 120) and flexible working patterns.

15. Consultant Physicians Working with Patients 3rd Edition

- The Chairman reported that the document had been discussed at a CGS away-day and was co-ordinated by Professor Farndon and Dr Davies. The committee extended its thanks for their hard work.
- Dr Newbury-Ecob asked if the document could be circulated more widely, as aspects of it seemed outdated. Dr Davies reported that unfortunately it had now reached the printing stage.

16. RCP Media and Public Affairs Expert Database

- Members noted the call for volunteers by the RCP, Press and PR Office.
- Professor Sampson asked whether referrals could go straight to the BSHG Public Affairs Spokesperson. The committee administrator agreed to look into this.

ACTION: *Committee administrator*

17. Conference Programme 2005

- The committee noted the RCP Academic Registrars response to its submission of “the application of genetics to common disease” as a conference in 2005.
- The Chairman asked all members to consider and submit conference suggestions by e-mail for 2006, as these would be required by the time of the next JCMG meeting.

ACTION: *All*

18. Any other business

18.1 Ethical, Legal and Social aspects of Genetic testing

- Mr Kent informed members that a report by D G Research on the subject is available at:

<http://europa.eu.int/rapid/pressReleasesAction.do?reference=IP/04/603&format=HTML&aged=0&language=EN&guiLanguage=en>

18.2 Ethnic monitoring across Clinical Genetic Departments

- Mr Kent informed members that a new post to address the above issue was currently being advertised. The job description can be found at:

http://www.gig.org.uk/gig/docs/jobdescription_PM.pdf

18.3 Myriad test revoked

- Members noted that the European Patent Office had revoked the patent of US firm Myriad Genetics on a genetic test for a predisposition for breast and ovarian cancer on 19th May 2004. Miss Kennard pointed out that Myriad had the right to appeal the decision.

18.4 Patient and Carers Network

- The Chairman informed members that Miss Nicole Barlowe (Manager, Patient and Carers Network, RCP) was to assign a suitable representative to join the committee at its next meeting. It was noted that Mr Kent would also act in this capacity.

18.5 Private Genetic Tests

- Professor Hodgson raised the issue of private firms offering genetic testing to individuals and that the reports issued often had the effect of “terrifying” patients. It was noted that the Human Genetics Commission had previously refused to regulate these services, as they did not want to be part of a “nanny state”. It was agreed that Professor Hodgson would draft a letter to the HGC on behalf of the committee citing examples and stating that regulation should be considered.

ACTION: Professor Hodgson

19. Dates of the next meetings

- Tuesday 19 October 2004