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, 21 October at 11am

Present:

Dr Heather Skirton JCMG Chair, BSHG

Dr Stephen Abbs	-	RCPath
Dr Hilary Burton	-	Observer, PHGU
Dr Trevor Cole	-	RCP (Items 1-14)
Dr John Crolla	-	RCPath
Professor Peter Farndon	-	BSHG
Dr Alan E Fryer	-	RCP
Dr Anne Green	-	RCPath (Items 1-8)
Dr Alison Hill	-	Observer, DH (in attendance)
Professor Shirley Hodgson	-	BSHG (Item 5 onwards)
Dr Tessa Homfray	-	RCP
Mr Alastair Kent	-	Genetics Interest Group (Item 5 onwards)
Ms Dianne Kennard	-	Observer, DH
Dr Helen Kingston	-	RCP, JCHMT SAC Chair
Dr Ruth Newbury-Ecob	-	RCPCH
Professor Peter Soothill	-	RCOG
Dr Linda Tyfield	-	BSHG
Dr Virginia Warren	-	FPH
Dr Maggie Williams	-	RCPath, Trainee representative
Mr Philip Masterton-Smith	-	RCP, Chief Executive (Items 1-6)
Mr Simon Land	-	Committee administrator

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1. Apologies for absence and welcome to new members

1.1 Apologies were received from Dr Hayley Archer (RCP, Trainee representative), Professor Carol Black (RCP, President), Dr Sally Davies (Observer, Wales), Professor Dian Donnai (Advisor to the CMO), Professor Ian Gilmore (RCP, Registrar), Professor Neva Haites (BSHG, Chairman), Dr Sian Morgan (RCPath, Trainee representative), Ms Elizabeth Woodeson (Observer, DH).

1.2 Dr Skirton welcomed Dr Hilary Burton as the new PHGU observer and Mr Land as the committee administrator.

2. To confirm and sign the minutes for the meeting held on 1 May 2003

2.1 Following a change to minute 3.6.2, the minutes of the last meeting were confirmed and signed as a true record.

3. Membership of the committee

3.1 The Chairman reported that Dr Hayley Archer had agreed to join the committee as the RCP trainee representative. Due to clinical commitments she was unable to attend the meeting.

3.2 Dr Burton replaced Dr Zimmern as the new PHGU observer.

4. Matters arising on the minutes

4.1 Appropriate services for adults with metabolic diseases

4.1.1 Dr Green reported that the document "Inherited Metabolic Disorders – A Service Vision for Setting Standards of Care and their Provision" was now ready for circulation.

4.1.2 The lack of a co-ordinated approach and appropriate services for patients with inherited metabolic disorders had been discussed at a meeting between the PRCP and the CMO. Dr Hill stated that the CMO would try to help but the situation was difficult as in many cases there are no mechanisms for co-ordinating care of adults with rare diseases. This is clearly an issue for a number of very small specialties.

4.1.3 The committee discussed the problems of training and commissioning as there is no recognition of Adult metabolic conditions as a sub-specialty. The following actions were agreed:

- The Chairman would write to the Presidents of the Colleges and the Medical Director of JCHMT highlighting that it was difficult to train clinicians due to the lack of recognition.

ACTION: ***Chairman***

- Dr Newbury-Ecob would write to Graeme Shortland, on behalf of the JCMG recommending that he looks at the Specialised Services Definitions.

ACTION: ***Dr Newbury-Ecob***

- Members would report on specific cases.

  ACTION: ***All***

- The PRCP would be copied in to all correspondence so the item can be kept on the agenda for future meetings with the CMO.

  ACTION: ***Professor Black***

4.2 "Pharmocogenetics: ethical issues"

4.2.1 Members were informed that the enclosure **Doc OCT 03/1** was wrongly appended and was relevant to item 16.

4.2.2 Dr Warren commented that the report (published 20 September 2003) was sensible and non controversial.
It can be found at:
http://www.nuffieldbioethics.org/publications/pp_0000000018.asp

4.3 Consultation document, "The supply of genetics tests direct to the public".

4.3.1 Ms Kennard stated that the report was available on the website:
http://www.hgc.gov.uk/genesdirect/genesdirect_full.pdf

but as yet no formal reaction had been received.

4.4 Specialty Specific Standards for Good Medical Practice

4.4.1 Professor Farndon reported no further update.

4.5 Copying of Laboratory Reports

4.5.1 Dr Crolla stated that he had been informed by the RCPath that the issue was covered by the CPA standard G2. However, he felt this was not adequate. It was noted that endorsement of the RCPath was required to change CPA standards. It was agreed that Dr Crolla would write to Dr Williams (Registrar, RCPath) to progress this issue.

ACTION: ***Dr Crolla***

4.5.2 Professor Farndon agreed to find out who had initially alerted the JCMG to the problem and report back at the next meeting.

ACTION: ***Professor Farndon***

5. Reports of the work of the JCMG in progress

5.1 Consent & Confidentiality Working Party

5.1.1 The committee had received the latest draft document from Professor Farndon via e-mail. There was substantial discussion of the document. It was noted that the emphasis was on encouraging good practice.

5.1.2 Members agreed that the document was a good, practical and useable guide. The Chairman thanked Professor Farndon and his team for their work.

5.1.3 Members were invited to comment on the content. Professor Fardon agreed to amend the document where necessary.

ACTION: ***Professor Farndon***

5.1.4 It was agreed that the document would now be circulated for consultation purposes. Any subsequent changes would be made via Chairs Action. Following this, the document would be sent to RCP, RCPath and BSHG for final approval.

ACTION: ***Professor Farndon, Chairman***

5.2 Training posts for genetic laboratory scientists

5.2.1 Dr Abbs reported that a meeting of stakeholders with an interest in on-line learning had occurred. They had expressed enthusiasm for progressing the work undertaken by Greenwich University and were keen that new modules should be developed.

5.2.2 Dr Abbs had requested funding from the DH to pilot 10 on-line Grade A training posts. Ms Kennard agreed to follow-up the issue and report back at the next meeting.

ACTION: ***Ms Kennard***

5.2.3 It was noted that at present Grade A training was funded by the Workforce Development Confederations. This funding should be for four years but due to disparities across Federations some are only funded for 2.5 to 3 years. Members therefore wished that a national consensus should take place. Ms Kennard agreed to follow-up the issue and report back at the next meeting.

ACTION: ***Ms Kennard***

5.2.4 It was agreed that the item would be included on the next JCMG agenda for further discussion.

ACTION: ***Committee administrator***

5.3 Genetics Education: Medical Undergraduates

5.3. Professor Haites was not present and the item was not discussed.

5.4 UK Haemophilia Centres Genetics Working Party

5.4.1 The committee noted the RCP response to the draft report UKHCDO: Clinical Genetic Services for Haemaphilia.

5.4.2 Dr Fryer reported that the document had been discussed successfully at the AGM of the UKHCDO in October 2003. He apologised that both the AGNC and BSHG had not been consulted, but stressed that the WP was looking to address this.

5.4.3 The next meeting of the WP would be held in January 2004. It was likely to remain in existence to progress genetic counseling services in Haemaphilia and to act as the body for the laboratory network to report to.

5.4.4 The Chairman stated that if members required a copy of the report that they should contact the committee administrator. It was agreed that a copy would be forwarded to Professor Soothill.

ACTION: ***Committee administrator***

5.5 Genetic counselor training post panel

5.5.1 The Chairman reported that the DH had released funds for the genetic counselor training posts. Currently about 50% of the posts had been filled.

5.5.2 It was noted that a meeting of trainees and mentors would be held in March 2004. This would discuss the progress thus far and establish any further educational needs.

6. DH White Paper: Our Inheritance, Our Future: Realising the potential of genetics in the NHS

6.1 Progress on implementation of commitments in the White Paper

6.1.1 Members noted the tabled report from Ms Kennard.

Clerks note: For the information of the Committee, the report been annexed to these Minutes at Appendix I.

6.1.2 It was noted that a group would be formed (comprising JCMG members, Clinical Scientists and Professional Body members) to cover the training issues resulting from the white paper.

6.2 GPwSIs

6.2.1 Members noted the tabled report from Dr Hill on developing GPwSIs in genetics. It was hoped that GPwSIs would help bridge the gap between primary and secondary care. However, no precedent for training, education and duties currently exists within genetics.

6.2.2 Dr Hill reported that a meeting of key stakeholders would be held in December 2004 and invited the JCMG to lend its specialist knowledge to the programme.

6.2.3 Both the Chairman (on behalf of the committee) and Mr Masterton-Smith (on behalf of the RCP) expressed strong support for the new programme. It was agreed that Dr Cole and Dr Warren would lead on behalf of the committee and liaise with Dr Hill.

ACTION: ***Dr Cole/Dr Hill/Dr Warren***

7. NICE Familial Breast Cancer Guideline Development

7.1 Members were informed that the closing date for comments on the final draft guidelines was 5 November 2003. The document could be found at :

http://www.nice.org.uk/article.asp?a=89142

Members agreed to forward any comments via the Chairman.

ACTION: ***All***

7.2 The Chairman reminded the committee of the huge educational remit in primary and secondary care that the guidelines would require. Ms Kennard stated that the Education and Development Centre (bids being accepted at this time) would eventually deal with this issue, but that the JCMG should begin to think of ways in which it can influence the situation.

ACTION: ***All***

8. Curriculum for non-genetic SpRs

8.1 Genetic Education: Needs and Evaluation (GENE)

8.1.1 The committee noted the summary of the first interim report : Education Needs.

8.1.2 Professor Farndon reported that issue of the educational needs of non-genetic SpRs had arisen from the Medical Specialties Board. It had been been decided to approach SpRs in 3 specialties (Cardiology, Neurology and Dematology) in 2 Deaneries (West Midlands and South Western) to ascertain their views. From these a curriculum would be formulated.

8.2 Multi-disciplinary course in formal genetics

8.2.1 Professor Farndon reported that the CGS was setting up a training committee to review the outcomes of the on-line questionnaire. From this a formal course will be made.

8.2.2 The results of the questionnaire revealed that Specialist registrars in genetics wanted the course to be tailored to medical trainees. There was some concern among committee memebrs that this would work against a multi-disciplinary approach.

8.2.3 The Chairman reported that the AGNC had not progressed the issue of developing a course in formal genetics for genetic counsellors for 2 years, as they believed a multi-disciplinary course was being designed. Therefore it was agreed that the CGS would begin dialogue with the AGNC. Professor Farndon would progress this issue.

ACTION: ***Professor Farndon***

8.2.4 The committee agreed that a sub group of the JCMG should be convened to focus on progressing a shared educational experience. It was hoped that the ACC and CMGS would also attend.

ACTION: ***Chairman***

8.2.5 Dr Abbs stated that an on-line package could be used to provide the course. This would be easily changeable to accommodate other professions.

9. Public Health Genetics Unit

9.1 Launch of report: Addressing Genetics, Delivering Health

9.1.1 Dr Burton expressed thanks to JCMG members who had attended the workshop.

9.1.2 The Chairman commended the report as a good concise document that made sensible recommendations.

9.2 My Very Own Medicine: Report from PHGU on Pharmocogenetics

9.2.1 Members noted the report, which had been previously circulated.

10. National Genetics Commissioning Advisory Group

10.1 UK Genetics Testing Group

10.1.1 Professor Farndon tabled a report on the work of the steering group. The main points of which are listed below.

- The UK Genetic Testing Network Steering Group has met 6 times since being set up as a subgroup of GenCAG. Minutes, terms of reference and other papers have been published on www.doh.gov.uk/genetics/gtn.htm

- The agreed Terms of Reference of the Steering Group were used to scope outputs, which have been used to draw up a project plan.

- The fundamental underlying principle is that of geographical equity.

- In May, 31 laboratories were invited to join the UKGTN in the 1st wave. Following receipt of applications in July, 14 laboratories were offered full membership, 12 laboratories were offered conditional membership and 5 laboratories have been asked to re-apply for membership once they have applied for CPA accreditation.

- A listing of all tests currently available on the UK network has been compiled.

- The DH, UKGTN team and Public Health Genetics Unit collaborated to devise a Genetics Education and Training Programme for commissioners and managers of genetic testing services. The pilot took place on 2nd October for South West England Local Specialist Commissioning Group. Approximately 40 people attended of which a third were commissioners, a third clinicians/laboratory scientists and a third patient representatives. The pilot will be evaluated and rolled out to other areas.

- The 6th meeting of the UKGTN Steering Group took place on 7th October 2003. Members evaluated the proposal forms for new testing provision and agreed to put forward new diseases/tests for consideration for funding by commissioners. These will be presented to GenCAG on 23 October.

- The UKGTN has to work within DH policy; however Steering Group members expressed concern that the commissioning model had been unable to address provider to provider charging and the model conflicts with a network approach.

- BRCA1 and BRCA2 testing requirements are being reviewed for the implementation of NICE guidance that is currently out for consultation. A paper will be produced with recommendations for future BRCA testing.

- A Directory of Diseases for which Diagnostic Molecular Tests are available is being presented to GenCAG on 23rd September, with a request to confirm funding for these tests. Existing tests that have been evaluated as having analytical validity, clinical validity and clinical utility are listed.

- Other issues being considered include the need for a tracking system/sample exchange, data standards and a UKGTN communication strategy.

- The Steering Group is discussing how best to obtain professional opinion to identify diseases for which genetic testing is clinically appropriate and which should be put forward for funding. Two specialist groups (neurogenetics and ophthalmic genetics) have made general presentations to the Steering Group. The Joint Committee's opinion on a mechanism would be greatly valued.

10.1.2 Professor Farndon thanked clinical and laboratory colleagues who had aided the work carried out over the previous 12 months.

10.1.3 Ms Kennard acknowledged the work of Professor Farndon and the group and hoped for benefits in terms of equity access, resulting in a more dependable and secure network.

10.1.4 Ms Kennard stated that the next meeting of GenCAG on 23rd October would be asked to agree to the adoption of the NHS Directory of Molecular Genetic Testing which contains those tests (clinically evaluated) which were available in UK GTN laboratories in May 2003. It is hoped that these tests will then be presented to PCTs as a template for the tests which should be available for their population. Recommendations would then be made for local discussions between commissioners and laboratory heads to discuss the level of funding needed to provide these tests. In addition, proposals for new tests to be added to the portfolio will be presented to GenCAG, following submission by laboratories and evaluation by UK GTN Steering Group. If these are accepted by GenCAG they will be recommended to PCTs for new funding from within the resources available to PCTs.

10.1.5 The Joint Committee expressed concern about the mechanism for the funding of rare tests but noted that recommendations from the UKGTN Steering Group were being presented to the next meeting of GenCAG.

10.2 Cancer Genetics

10.2.1 The topic was covered under item 7.

10.3 Contract Currencies

10.3.1 Members noted a tabled letter from Dr Tyfield regarding Workload Units.

10.3.2 It was noted that WLUs do not take account of education, administration, management or training. Therefore, if WLUs are to be used as a comparison between laboratories they may not provide a true reflection.

10.3.3 Ms Kennard agreed to raise the issue at the next GenCAG meeting.

ACTION: ***Ms Kennard***

11. National Genetic Reference Laboratories

11.1 Dr Crolla informed the committee that the website was now live. It could be found at:
http://www.ngrl.org.uk

12. Human Genetics Commission

12.1 Ms Kennard reported that the HGC met in Cardiff on 25 September 2003. They discussed the following issues:

- Paternity testing and genetics and reproductive decision making.
- UK Biobank Ethics Governance Framework .
- Government response to the report '*Inside Information*' including such issues as DNA theft, genetic discrimination and profiling babies at birth.

12.2 Mr Kent highlighted the following issues that should be considered by the JCMG.

- Progress of the consultation on genetics and reproductive technologies – draft due in the New Year.
- Unauthorised access to DNA – draft has clinical implications.

13. Manpower andTraining

13.1 RCPath SAC

13.1.1 Dr Crolla informed the committee that the Association of Clinical Embryologists had approached the RCPath for membership. This issue was currently being progressed, and if successful they would join via the Genetics SAC.

13.1.2 It was reported that 40 new Part I members had been admitted over the past 3 years. The success rates were 90% for Molecular Genetics and 75% for Cytogenetics. Dr Maggie Williams reported that a good self-help group existed for Part I and that a questionnaire of trainees was currently being undertaken. This would be presented to the SAC and JCMG in the near future. A mentoring scheme for Cytogenetics was also being progressed by Dr Teresa Davis.

13.1.3 The Professional Standards Unit was meeting to look at multi-disciplinary training.

13.1.4 Dr Crolla had spoken at the RCPath Council on the Genetics White Paper. He reported that the main concern was manpower resources.

13.2 JCHMT SAC in Clinical Genetics

13.2.1 Dr Kingston reported on the new legislation governing entry to the specialist register and the impact on genetics.

13.2.2 The Postgraduate Education and Training Board (PMETB) would take over from the STA as the body that recommends entry to the specialist register. The handover would not be for 12 months but would provide an opportunity for NCCGs and those trained overseas to be re-assessed. This assessment would be made via education **and/or** training.

13.2.3 The committee noted that members of the specialist register were at liberty to apply for a consultant post in any specialty.

13.3 Manpower in Clinical Genetics

13.3.1 Attendance prevented the item from being discussed.

14. Fetal Medicine SSM

14.1 Professor Soothill asked members to consider the content of the RCOG, Special Skills Training Module for Fetal Medicine. He highlighted that the genetics component was not aimed at the sub-specialist.

14.2 Members agreed to forward comments by e-mail to Dr Kingston who would co-ordinate the committee's response to RCOG.

ACTION: *Dr Kingston*

15. Antenatal screening – Working standards

15.1 Dr Newbury-Ecob stated that concerns had been raised regarding the level of counselling support for antenatal screening. Professor Farndon had spoken to the Chairman of the ANSG National Screening Committee who confirmed that money was available for screening but not for counselling.

15.2 Members thought that Antenatal Screening Advisors could be helped by examples of good practice from certain areas.

15.3 It was agreed that genetic counsellors should refer matters back to the Antenatal Screening Advisor if they suffered an increased burden.

15.4 It was agreed to keep this item on the agenda for the next meeting.

ACTION: ***Committee administrator***

16. Fit for Practice in the Genetics Era

16.1 The Chairman tabled the executive summary from the document. Members expressed broad support for the competencies detailed in the report. It was felt that knowledge of genetic conditions and their implications (by nurses and midwives) would improve the experience and support structure for families with concerns about genetic conditions. There was a need to train educators and change the pre-registration training curricula to progress this.

16.2 It was noted that the education of nurses and midwives already registered (and potential returnees to practice) would also need addressing.

16.3 It was agreed that the Chairman would write to Dr Kirk with the committee's response to the document.

ACTION: ***Chairman***

17. Intellectual and Property Rights and Genetics

17.1 Members noted the report produced by the University of Sheffield, University of Cambridge and PHGU. Copies had been circulated prior to the meeting.

18. RCP Lectureship and Conference Programme

18.1 Members noted that the college was inviting nominations for lectures and conferences to be held in 2005.

18.2 The committee suggested the following topics:

Lectures

- Adult metabolic disease

Conferences Genetics and common disease
To include:

- Cancer genetics
- Bringing genetics into mainline healthcare
- Genetics and reproductive technologies
- The application of genetics to common diseases
- Pharmocogenetics

18.3 The Chairman asked that members forward other suggestions by e-mail.

ACTION: ***All***

18.4 Members wishing to submit lectures or conferences should contact the administrator for the relevant proposal form.

ACTION: ***All***

18.5 Distinction Awards

As part of the annual JCMG business, the Chair has been asked to forward the names of any medical Consultants who should be considered for Distinction Awards. The Chair encouraged all members of the committee to provide suggestions. These should be forwarded to the Chair or Dr Tessa Homfray as soon as possible.

19. Any other business

19.1 Ms Kennard informed members that copies of the Genetics White Paper were still available. Interested parties should contact Ms Kennard by e-mail.

19.2 Ms Kennard stated that the Sunday Times would include an educational CDROM as part of the DNA 50 year initiative. Dates of publication would be 16 and 23 November 2003.

19.3 Ms Kennard informed the committee that Naomi Brecker had recently given birth. It was agreed that a bouquet of flowers would be forwarded with the JCMGs congratulations and best wishes.

ACTION: ***Committee administrator***

19.4 Ms Kennard was asked the likely timing for the Human Tissue Bill.

ACTION: ***Ms Kennard***

Clerks note: Ms Kennard has since reported that it is hoped it would be introduced in the next session of Parliament. More details will be available after the Queen's Speech (26 November 2003).

20. Dates of the next meetings

20.1 The meeting dates for 2004 were agreed:

- Tuesday 13 January 2004
- Thursday 20 May 2004
- Tuesday 19 October 2004

APPENDIX 1.

Progress on implementation of commitments in the genetics White Paper

New announcements will be publicised via the Department of health website at http://www.doh.gov.uk/genetics/whitepaper.htm

Expanding specialised services (chapter 2):

Application packs for a 2 stage bidding process to access the £18m capital to **upgrade NHS laboratory facilities** were sent out on 29th August. A national workshop for key stakeholders was held on 12th September to explore options for future laboratory configuration and discuss the bidding process for this major investment. Outline bids are required by 31st October. The bidding process should be completed by March to allow allocation of funding from April 2004.

Selected trusts have been recruiting to new **genetic counsellor training posts,** of which DH committed to fund 50.

DH will shortly be setting up a group, to include JCMG representatives, to discuss how best to take forward the commitments around **clinical scientist training.**

The Manchester reference laboratory is commissioning a consultancy study to look at **IT** needs within genetics services. This study and other reference laboratory work will inform decisions on use of the £1m capital intended for use on IT within the UK Genetic Testing Network.

Building genetics into mainstream services (chapter 3):

Macmillan Cancer Relief have invited bids to pilot their model of **familial cancer** services in up to 6 areas, for which DH is contributing funds. Bids are required by 31st October.

DH is also funding the London Knowledge Park to run pilots of cascade screening for **familial hypercholesterolaemia**. The first meeting of the steering group for this development took place on 23rd September and recruitment is underway for project management. Pilot sites are scheduled to start in April 2004.

Bids will be invited with the next few months for a range of **service development initiatives** to look how genetic knowledge might be integrated into mainstream clinical areas within primary, secondary and tertiary care. We envisage a four-month deadline for applications which, following discussions at the Genetics Commissioning Advisory Group, will be co-ordinated via specialised commissioners. An **evaluation** of these initiatives, which will focus on the more qualitative aspects, will be commissioned from a team of academics so that the wider NHS can benefit from the lessons learned and experiences gained. We will tender for the evaluation as a research project within the

next couple of months. We will progress work to develop the role of **GPs with a special interest** in genetics separately and with an expert group (see separate paper).

Spreading knowledge across the NHS (chapter 4):

An invitation for bids was placed on 17th September in the Official Journal of the European Union to establish a **Genetics Education and Development Centre** (http://www.doh.gov.uk/purchasing/tenders.htm)**.** This will catalyse (but not normally provide) education and training in genetics for non-genetic specialist healthcare staff, and support service development. Close links with those involved with education and training and with the genetics community will be vital. It is expected that a contract will be awarded before the end of the financial year.

Contracts have now been signed with the team who will develop 'Genepool', the **genetics branch library of the National electronic Library for Health** which is being developed through the NHS Information Authority.

PHGU and the UKGTN team have put together a **training programme for commissioners;** the first event will take place on 2nd October in the south west of England.

Generating new knowledge and applications (chapter 5):

Bids have now been invited for the gene therapy research programmes announced in the White Paper, plus a research management contract. The call for bids for the pharmacogenetics programme is expected shortly.

Ensuring public confidence (Chapter 6)

The Human Genetic Commission recently discussed the Genetics White Paper and welcomed the Government response to the main recommendations for a new offence of non-consensual DNA testing and a review of the evidence for genetic discrimination. Details of the proposed new legislation on human organs and tissue were published in September.

HGC, working with the National Screening Committee, will review the case for universal DNA profiling at birth to guide treatment and prevention decisions.

HGC and the Genetics and Insurance Committee recently held a joint public meeting to consider the actions that are needed before the moratorium ends in 2006.

NHS Genetics Team, Department of Health

1st October 2003