

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 Wednesday 9 January 2002 at 1 pm

Present

Professor P Farndon	Chairman RCP
Professor I Gilmore	Registrar, RCP
Dr S Abbs	RCPPath
Dr N Brecker	DH Observer
Dr R Elles	BSHG
Dr A Fryer	RCP
Dr H Hughes	Observer (Wales)
Dr V Warren	FPHM
Ms D Kennard	DH Observer
Dr H Kingston	JCHMT SAC
Dr A Green	RCPPath
Professor S Malcolm	RCPPath
Dr L Gaunt	BSHG
Professor R Winter	BSHG
Dr R Newbury-Ecob	RCPCH
Professor P Soothill	RCOG
Dr R Hapgood	RCGP
Mr R Zimmern	PHGU Observer
Dr H Skirton	BSHG
Mr J Barber	BSHG
Dr C Chapman	Chair, Working Party on Guidance for Ethical Committees
Dr F Douglas	Chair Working Party Consent & Confidentiality

In Attendance

Mrs Val Knight

Committee Administrator RCP

1 Apologies for absence/Welcome/Introduction

Apologies for absence were received from Professor N Haites, Dr J Old, Dr P Brennan, Professor M Connor, Professor D Donnai, Professor R Mueller, Mr A Kent, Dr H Williams.

Professor Farndon welcomed Dr H Kingston (who was replacing J Tolmie), Dr C Chapman and Dr F Douglas, Chairman Working Party Consent & Confidentiality.

1b Rotation of Committee Members

Dr Helen Williams has replaced Dr J Crowe as the RcPath Registrar, and Dr J Old has replaced Mrs Maggie Fitchett as one of the RcPath members. Mr John Barber was attending his last meeting as one of the BSHG members and the Chairman thanked him for his contributions, particularly with regard to the Consent and Confidentiality Working Party.

2 Minutes of the last meeting – 20 September 2001

The following amendments were noted;

page 3 – Item 3.7b – Mike Hall changed to Mike Hallworth
page 4 – Item 3.8 - HTA Therapeutics Panel amended to Diagnostic & Screening Panel.

Professor Farndon proposed, due to the late circulation of the minutes, that the minutes were agreed as an accurate record with the proviso that any further amendments to be forwarded to him within the next two/three days.

3 Matters Arising from the Minutes

3.1 Patents & Genetic Testing

a Dr N Brecker, DoH, advised there was little information to report since the last meeting. A detailed response from Myriad was still awaited over the BSHG proposal that existing diagnostic laboratories be considered as a candidate licence holder. Myriad had not yet entered into an agreement with a UK partner.

b Advice was being taken from a patent lawyer on the strength of the BRCA1 patent.

c Challenges to the patent by other countries were being kept under review.

3.2 Clinical Governance

a Dr H Hughes reported that the Clinical Genetics Society had received positive feedback on the documents on follow up and recall in clinical genetic practice, guidelines on pedigree drawing and management of hypertrophic cardiomyopathy. She would report further at the next JCMG meeting.

b At the last JCMG meeting, representatives of other Colleges were asked to take the Genetic Practice Guidelines to their Colleges for discussion and dissemination. Professor Soothill reported that the RCOG had noted their existence and commented that the RCOG was seeking names of Clinical Geneticists for several of its Governance Committees. The Royal College of Paediatricians and the Royal College of General Practitioners had also noted the document; both had genetic representatives on their Governance Committees. JCMG felt it would be helpful to know the names of the Geneticists on these groups.

Action: Dr Newbury-Ecob, Dr Hapgood

c Dr Angela Barnicoat, genetics representative on the Royal College of Physicians London Clinical Governance Committee was reviewing existing Clinical Governance documents in genetics to place those meeting RCP criteria on the RCP website. The relative paucity of Clinical Governance documents in genetics was noted.

d An audit of a document from the Edinburgh genetics unit concerning standards as a patient “journeyed” through a clinical genetics unit was being undertaken.

e The Clinical Genetics Society was preparing a standardised information leaflet on what a patient might expect when referred to the genetics service. It is hoped that this may meet the GenCAG proposal for such a leaflet.

3.3 Training Needs of Genetic Counsellors

Dr H Skirton reported that a Registration Board had been set up. An initial group of Counsellors was to be approved who would act as trainers. About two-thirds of Genetic Counsellors are intending to register.

An analysis of training needs revealed that about one-third of current Genetic Counsellors had unmet training needs and these were starting to be addressed.

A discussion with the Department of Health is to be held in January 2002 about training requirements and the numbers of posts. The Chairman noted it was unclear as to how posts would be funded.

3.4a Training Posts for Genetic Laboratory Scientists

Following Mrs M Fitchett’s letters of concern over the training of sufficient numbers of staff for genetic laboratories, JCMG agreed to set up a Working Group to identify areas of concern. Dr Steven Abbs was invited to be the Chairperson, other members including Lorraine Gaunt, Stuart Imrie, Anne Curtis and a member of the Department of Health Human Resource Directorate (Guy Cross). In addition, representatives from educational consortia and metabolic biochemistry would be sought.

Action: Dr S Abbs

3.4b Specialist Training for Clinical Scientists : Metabolic Biochemistry.

Dr Green reported that the discussion document (ENCLOSURE 1) had met with wide support: manpower requirements for metabolic biochemistry were to be calculated in the near future.

Dr S Abbs was asked to include this within the remit of his working group.

Action: Dr S Abbs

4 Reports of the work of the Joint Committee in progress

4.1 National Pregnancy Record (ENCLOSURE 2)

Professor P Soothill was waiting acceptance of the document by some of the other Colleges. Professor Soothill was requested to consider submitting a commentary to professional journals after the report had been published on the RCP website. Professor Soothill and his working group were thanked for a very clear working document.

4.2 Consent & Confidentiality Working Party (ENCLOSURE 3)

Dr F Douglas thanked all members for their helpful comments. A flow chart for the process of gaining consent had been produced and JCMG found this a very helpful addition. Discussion at JCMG seemed to highlight potential differences between genetic laboratories, clinical genetic services and non genetic clinicians in the interpretation of “consent”. The sequential nature of genetic investigations raised questions about what the original consent covered, and when and how often patients should be asked for further consent before additional tests were performed. A strong case was argued for “mutual confidentiality” within and between genetic departments. It was important to ensure that the JCMG document

contained guidance consistent with that from other sources such as the Royal College of Pathologists, the Department of Health and the Medical Research Council. The Chairman proposed that

- 1 Legislative issues raised by the document be summarised and forwarded urgently to the Human Genetics Commission as part of their work on personal genetic information bodies
- 2 Much of the excellent discussion about current law be presented in a learned journal.
- 3 The guideline section be written in a concise form, and together with suggested consent forms and information leaflets be brought to the next meeting before wider consultation.

Action: Dr F Douglas, Prof P Farndon

4.3 Genetics Education

(a) Medical Undergraduate

Professor Haites is intending to present a document at the next JCMG meeting.

(b) Course in Basic Genetics for Specialist Registrars

A Working Group, under the chairmanship of Professor Farndon, was being formed to devise this course, hopefully to take place in Spring 2003. The Specialist Registrars had been consulted on their learning needs, and these would be taken into account in the design of the course, with further consultation with the Specialist Registrars and Consultants.

(c) Professor Farndon was discussing with the Department of Health the funding of a training needs analysis in genetics of Specialist Registrars in other specialties.

(d) Primary Care

Dr Rhyddian Hapgood reported on an email group considering genetics education for general practitioners. It had been suggested that including genetics questions in the RCGP examination would be a first step. The group was intending to design some sample questions, and to devise a one or two page curriculum for genetics in primary care. The JCMG welcomed this initiative and would be pleased to take part in any joint initiatives.

4.4 Guidance for Ethical Committees in Genetics

Dr C Chapman presented the report 'Ethical Issues in Genetic Research on Human Subjects' (ENCLOSURE 4) which had previously been circulated. He had been invited to produce the document to guide research ethics committees. Dr Chapman and the working party were thanked for an excellent document. Dr Chapman would incorporate the comments made during discussion by JCMG members, and the document would be forwarded to the Central Research Committee office for circulation, as well as to the other Colleges and the British Society for Human Genetics, and be placed on the RCP/BSHG websites.

Action : Dr Chapman & Professor Farndon

4.5 Contract Currencies

15 laboratories were piloting the cytogenetics contract currencies. Dr Abbs reported that the molecular currency data were being collated by the CMGS audit group and he would present further information at the next JCMG meeting.

The CGS had arranged for trials of the clinical contract currencies in six centres, but there were some problems in modifying computer programmes.

Dr N Brecker asked that the JCMG give an interim report to GenCAG at their March meeting to gain involvement of the Commissioners.

Dr A Green reported that currencies in metabolic biochemistry had been aired as an issue, but it would prove difficult to take this further at present.

5 Public Health Genetics Unit

Dr R Zimmern reported:

- (a) A literature review and mapping exercise on the current provision of Genetics Education across all professional groups had been carried out and an interim report would be available shortly. Funding from the Wellcome and Department of Health is being sought to devise and fund an over-arching educational strategy as phase 2 of this project.
- (b) A meeting has been organised on 8 February 2002 on QPCR and FISH for chromosome examination of fetuses identified as being at risk through the National Ante-natal Screening Programme. This will inform the Ante-natal Sub-Group of the National Screening Committee and a Sub-Group of GenCAG. It was noted that an HTA report on this subject was still awaited.
- (c) A project on patents and their implications for the NHS was underway with a report expected in the autumn.
- (d) The Public Health Genetics Unit had secured funding for the next 5 years from the DoH and Wellcome.

6 National Genetics Commissioning Advisory Group

6.1 Summary (ENCLOSURE 5)

The genetics unit had provided an update on their work and plans.

6.2 Cancer Genetics (ENCLOSURE 6)

A draft document from the DoH and the Macmillan Cancer Relief was discussed. Dr Brecker asked for comments to be forwarded by 21 January 2002. The draft report was to be used to inform the Green Paper Advisory Panel and when finalised would be circulated for wide consultation. JCMG supported and endorsed this document.

John Barber agreed to write a paragraph on Laboratory Services for this report.

Action : Mr J Barber, All

6.3 Quality Standards (ENCLOSURE 7)

Dr Brecker, DoH, advised that the quality standards were the result of two workshops and thanked all for their contributions. The standards had been discussed at GenCAG where some amendments had been made.

The standards were discussed in detail by JCMG members and some further amendments were suggested. These would be taken to the GenCAG meeting in March, after which it was expected that Commissioners would be asked to monitor service provision using these standards.

Action: Prof P Farndon

6.4 Commissioning Genetics Services (ENCLOSURE 8)

The NHS Circular 9 (HSC 2001/024) confirmed that in 2002-03 Regional Specialist Commissioning Groups will have a specific role in developing PCT capacity to commission specialised services. PCTs will be financially bound and organisationally committed to the decisions made through PCT consortia to ensure that specialised services continue to be effectively commissioned at StHA and supra StHA levels. PCTs must honour existing agreements (financial and otherwise) negotiated by Regional Specialist Commissioning Groups and current Specialised Service Commissioners.

For screening programmes, the consortia will also be responsible for ensuring that population registers are maintained and developed, that a range of front line staff and others are involved in shaping programme development, and that service continuity with maintenance of the skilled work force is achieved in 2002/3. Each PCT and NHS Trust will be accountable for delivery of the parts of the programme that it directly provides.

Dr Green was extremely concerned that this mechanism would be able to protect and develop the screening services. Dr Brecker advised that she would pass this comment on to the National Screening Committee.

Action: Dr N Brecker

6.5 National Reference Laboratories and Knowledge Parks

Ministers would make decisions following receipt of the advice of the Commissioning Panels and the successful centres would be announced by the Secretary of State at the International Genetics Conference on 16 January 2002.

6.6 Green Paper on Genetics

Lord Turnbull had chaired several meetings of the advisory panel. The Green Paper would be prepared by members of the DH Genetics Unit. Its date of publication had not yet been set.

6.7 Development Bids

Thirty million pounds had been made available by the Secretary of State for developments in genetics and towards preparing the NHS to anticipate and benefit from advances in genetic sciences. Regional Genetics Centres had been invited to prepare bids before Christmas 2001. This development money was for England. The bids were being assessed and the results would be announced by the Secretary of State on 16 January.

7 Human Genetics Commission

7.1 Human Genetics Commission – Workplan (ENCLOSURE 9)

JCMG discussed the Human Genetics Commission work plan as the HGC Chair Baroness Kennedy had asked for comments. The JCMG Chair was asked to write to the HGC to express concerns on the long term outcome of babies born following intra-cytoplasmic sperm injection, how the five years of the insurance moratorium might be used, and that the results of “over the counter tests” might cause undue anxiety and requests for clarification for support from services within the NHS. The JCMG wished to ask the HGC to consider whether a mechanism is required to enforce the code of practice and guidance should the HGC decide a submitted project for tests supplied direct to the public is “undesirable”.

Action : All

7.2 Human Genetics Commission – Pre-implantation Diagnosis

It was noted that the outcome of the public consultation on preimplantation diagnosis was available on www.hgc.gov.uk/business_publications_pgdoutcome.pdf.

8 Disabled Peoples’ International Europe Position Statement on Bioethics & Human Rights

This document (ENCLOSURE 10A) was also available on www.dpieurope.org. A response (ENCLOSURE 10B) prepared by Professor N Haites and Mr A Kent was discussed and endorsed by the Committee with some suggested amendments. Members felt that some of the health statistics needed further clarification and Dr Fryer and Dr Warren were requested to forward their comments in writing to the JCMG Chair.

Action : Dr Fryer, Dr Warren

9 Matters from the Royal College of Physicians

9.1 Good Medical Practice for Clinical Geneticists

Professor I Gilmore advised members that the GMC had requested specialty specific documentation based on its publication ‘Good Medical Practice’. This would form part of the revalidation process.

There was to be a generic document applicable to all Physicians, with specialty specific guidelines. Professor Winter had prepared these for Clinical Genetics on behalf of the JCMG. A response from the GMC is expected early in 2002.

9.2 Consultant Physicians Working for Patients

Professor I Gilmore reported that the document ‘Consultant Physicians Working for Patients’ had now been published. Following a request by Professor Farndon, Professor Gilmore agreed that the section for Genetic Medicine would be available via the Royal College of Physicians’ website.

9.3 College Education Department Proposed Modular Specialty Publications

The RCP London Education Department had been considering developing web based interactive educational material to support SpRs and which would also be suitable for CPD. A meeting had been held with specialist society representatives to assess what material was already available. A decision as to whether the initiative would proceed is awaited.

9.3 Health and Social Care Act 2001

The Royal College of Physicians had been requested to respond to a consultation on the Health and Social Care Act 2001, where regulations under Section 60 are required. This provides the Secretary of State for Health with the power to authorise or require that patient identifiable information is used to support essential NHS activity where there is currently no secure basis in law other than the consent of the patient concerned and it is thought that there are real barriers to seeking or obtaining consent.

As material on this had only just been received, Professor Farndon agreed to review any actions required and respond to the consultation directly.

Action: Prof P Farndon

10 The Establishment of a Genetics Section at the Royal Society of Medicine

The Royal Society of Medicine is establishing a genetics section, chaired by Professor A Emery, which would meet three times a year.

Professor Farndon would write to welcome the Section and ask for further information.

Action : Professor Farndon

11 Manpower and Training

(a) RCPATH SAC

Professor S Malcolm reported that examination procedures were being reviewed, especially in relation to the examinations in Cytogenetics. The funding and resources for training posts, and staff to supervise them, were continuing issues.

(b) JCHMT SAC in Clinical Genetics

Dr Kingston reported:

- (a) the curriculum had been re-written and submitted to the JCHMT. Approval was awaited from the Specialist Training Authority. Entry criteria to an SpR post in genetics had been broadened.
- (b) SHO training was being re-structured, with a broader, longer training period which was thought would be especially helpful to trainees entering Clinical Genetics.
- (c) The appraisal and assessment documents for SpRs in genetics were being reviewed, final versions to be expected in March 2002.
- (d) New SpR (Genetics) Training Posts. Thirty have been allocated. Nine have been released and funded, with twenty one proposed over the next two years. Funding for these posts is currently uncertain.
- (e) The JCHMT had organised two joint visits where every training job in a Deanery was reviewed at the same time. These had been very successful. However single specialty visits would continue.

12 Any Other Business

(a) Chromosome anomaly database

Stop-gap funding for a year had been granted by Sir John Pattison whilst a national review of the Chromosome Anomaly Database was undertaken.

(b) **Direct to the public testing for polymorphisms**

Dr Virginia Warren reported that a commercial company was working with a high street retailer to offer polymorphism testing for life style advice over diet. Their request to provide this testing was currently being reviewed by the Human Genetics Commission. The Chair asked Dr Warren to present more details at the next meeting.

13 Dates of Future Meetings

Monday 20th May 2002 (RCP)
Thursday 26th September 2002