

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 23rd May 2001 at 2.00 pm

Present

Professor Peter A Farndon	Chairman RCP
Professor Ian Gilmore	Registrar RCP
Professor Neva Haites	BSHG Chairman
Dr Stephen Abbs	RCPPath
Dr Naomi Brecker	DH Observer
Professor Dian Donnai	CMO Adviser
Dr Rob Elles	BSHG
Mrs Margaret Fitchett	RCPPath
Dr Alan Fryer	RCP
Dr Helen Hughes	BSHG
Professor Noor Kalsheker	RCPPath
Professor Robert Mueller	RCP
Dr Heather Skirton	BSHG
Mrs Katie Waters	BSHG
Dr Paul Brennan	RCP Trainees
Mr Alistair Kent	GIG
Dr Virginia Warren	FPHM
Ms Dianne Kennard	DH Observer

Mrs Val Knight

Temporary RCP Committee Administrator

In attendance

Dr Amanda Collins

1 Apologies for absence/Welcome/Introduction

Apologies for absence were received from Mr John Barber (BSHG), Dr Ruth Newbury-Ecob (RCPCH), Mr Anthony Taylor (DH Observer), Professor Soothill (RCOG), Professor Mike Connor (Scottish Colleges), Dr Ron Zimmern, Dr Julie Crow (RCPPath Registrar), Dr John Tolmie (RCP JCHMT SAC), Professor Robin Winter (BSHG), Professor Sue Malcolm (RCPPath).

Mrs Katie Waters (who was standing in for Dr Lorraine Gaunt), Ms Diane Kennard (who was standing in for Mr Anthony Taylor) and Dr Amanda Collins were welcomed.

The Chairman noted that Dr Newbury-Ecob was joining the Committee as the Royal College of Paediatrics and Child Health representative, and thanked Dr Jill Clayton-Smith for her work for the Committee. He also thanked Dr Dennis Cox who had written to say that he was retiring from the Committee as the Royal College of General Practitioners nominee.

2 Minutes

The minutes of the last meeting of 16th January 2001 were accepted as a true record with the following corrections -

Item 4 – Genetics and Insurance Committee

First paragraph, second sentence. “[Professor Donnai] pointed out that the Government had set up GAIC...”

The second paragraph should be replaced with “the first application had been to use results of tests for Huntington’s disease for life insurance; GAIC had not accepted part of the application which proposed using the number of CAG repeats as a predictive indicator to determine the insurance premium”.

The third paragraph should be replaced: “Professor Donnai gave her personal view that GAIC needed more members and a broader expertise”.

Item 5e) Public Health Genetics Network It was noted that Dr Heather Grimbaldeston’s name was incorrectly given.

Item 9 National Specialised Service Definitions – Genetics A word missing from the first sentence was noticed – “Principally to identify to commissioners what clinical activity within a speciality is regarded as “specialised””.

3 Matters Arising from the Minutes

3.1 Patents and genetic testing

Dr Brecker reported that initial positive contact had been made with Myriad Genetics by Department of Health officials. It was believed that Myriad was trying to identify a commercial partner in the United Kingdom. Dr Rowena Jecock was hoping to present the latest situation to the next meeting of the National Genetics Commissioning Advisory Group in June. Dr Brecker undertook to ensure that any proposals would also be communicated to the Joint Committee.

Action: Dr Brecker

3.2. Genetics and Insurance

The House of Commons Science and Technology Committee had published its report on genetics and insurance. The report confirmed the continuation of the Genetics and Insurance Committee (GAIC) whose composition and remit may be broadened. A new chairman was awaiting approval by Ministers.

It was noted that the Human Genetics Commission had urgently addressed public concerns over genetics and insurance and published interim recommendations, which

included a moratorium on the use of genetic information for insurance. The recommendations are available on the Commission's website – <http://www.hgc.gov.uk>.

3.3 Patients Panel of the Human Genetics Commission

Dr Brecker confirmed that the panel had not yet been set up, but the DH Genetics Unit was reviewing its remit which may entail a role wider than advising solely the Human Genetics Commission, and it may also be asked to address other issues.

Mr Kent asked that this remained on the agenda.

Action: Chairman

4 Progress reports of the work of the Joint Committee

4.1 National Pregnancy Record

Unfortunately Professor Soothill was unavoidably delayed due to travel difficulties, but the Chairman reported that considerable progress was being made in the working group under Professor Soothill's chairmanship and a more detailed report would be available shortly.

4.2 Consent and Confidentiality Working Party

The Group under Dr Fiona Douglas' chairmanship was currently collating the responses to questionnaires completed by all the Regional Genetics Units, and the Chairman thanked the genetics community for their detailed responses. Dr Douglas was preparing the first draft of the report which it was hoped would be available for consultation during the British Society for Human Genetics meeting in September. The Committee was advised that this would be a major document and useful for clinical practice. Professor Farndon explained that he had already had talks with the RCP to discuss having this published.

4.3 Genetics Education

a) Undergraduate medical education

Professor Haites was planning to convene a meeting of representatives from medical schools and representatives from regional genetics centres to discuss the basis of the national undergraduate curriculum in medical genetics in the autumn of this year.

b) Postgraduate training course in genetics

The Chairman confirmed that it was planned to hold a series of meetings with specialist registrars to determine what their educational needs were, and then to hold similar meetings with trainers from the Regional Clinical Genetics Units to determine the form and content of the proposed course. There appeared to be considerable support for a modular course, beginning with "basic genetics" with further specialised modules led by the country's recognised experts on those topics. Whilst it had originally been intended that such a modular course would be for the specialist registrars in clinical genetics, the Joint Committee requested that the modular structure of the course be developed to allow all professionals in genetics to benefit from its opportunities.

The Chairman confirmed that other medical specialties were also interested in modules dealing with the genetics of their specialties.

The Chairman confirmed that it was intended that the structure and contents of the course would be informed by and based upon the identified educational needs.

4.4 **Guidance for Ethical Committees on Genetics**

The Chairman reported that Dr Cyril Chapman's working group expected to have the final draft completed within the next few weeks, and circulated to Joint Committee members. It is hoped that the document will be adopted by the Central Office for Research Ethics Committees and be helpful to local research ethics committees.

5 **Secretary of State's Speech**

The Secretary of State's Speech on Genetics, previously circulated to all members, was discussed in detail.

The Chairman of the Joint Committee had been invited to attend the Secretary of State's Speech and reviewed some of its major points.

The Chairman welcomed the speech on behalf of the genetics community, and commented that he felt there were parts of the speech for which existing mechanisms would ensure that the plans were put into practice. Other parts of the speech needed assistance from the genetics community and commissioners to develop new mechanisms – training of genetic counsellors, for instance. Thirdly, there were aspects of the speech which appeared to be statements of intent and detailed work had not yet been undertaken on them.

Dr Hughes asked about the implementation of the speech in Scotland, Wales and Northern Ireland. Dr Brecker confirmed that the funding announced in the speech was mainly for England, but it was hoped that the overall plan of the speech, suggesting a framework for clinical services, would be adopted by the national assemblies. The Secretary of State had outlined proposed service developments, but the commissioning would be through the existing local systems. The National Genetics Commissioning Advisory Group (GENCAG) would play a key role in co-ordinating developments throughout the United Kingdom.

Mrs Fitchett asked for clarification of the Rare Genetic Testing Network, and Dr Brecker confirmed that Dr Robert Elles and herself had been asked by GENCAG to prepare a document on possible structures for its next meeting.

Dr Brecker confirmed that the mechanisms for bidding for the National Reference Laboratories were still being developed.

Members from all genetics specialties on the Joint Committee were concerned about the mechanism for increasing the number of training posts for genetic laboratory scientists. There was concern that an increased number of training posts would not be available under current planning systems until September 2002, and there was real concern that this would not allow adequate numbers of scientists to be trained ready for the developments envisaged in the Secretary of State's Speech. It was agreed that the Association of Clinical Cytogeneticists and the Clinical Molecular Genetics Society would contact the chief scientist, Dr Peter Greenaway to make proposals for increased numbers of training posts as soon as possible, and the Chairman of the Joint Committee would also write to confirm that this was a concern shared by all professional groups in genetics.

Action: Chairman

Urgent work also needed to be undertaken to ensure that adequate numbers of genetic counsellors could be trained. Dr Brecker confirmed that she was convening a meeting as soon as possible to discuss training requirements and training capacity for genetic counsellors. Dr Brecker confirmed that the DH Genetics Unit was pleased to receive expressions of interest to serve on this group.

Professor Kalsheker raised concerns about the paucity of training posts in biochemical genetics. After discussion, it was agreed that Professor Kalsheker would write to the Royal College of Pathologists Common Pathway Training Group to discuss this matter further.

Action: Prof Kalsheker

6 **National Genetics Commissioning Advisory Group**

Members of the Joint Committee who had been present at the first meeting of the Genetics Commissioning Advisory Group reported that there had been agreement with the commissioners on priority areas, the most urgent of which is the setting up of the genetic testing network. The next meeting is being held on the 25 June.

7 **National Specialised Service Definitions - Genetics**

a) **Definition**

Following extensive consultation, it was likely that the specialised definition for genetics would be finalised at a meeting in early June.

b) **Discussion and implementation of proposed contracting currencies**

The Chairman wished to record his thanks formally to the professional groups who had produced the proposed contracting currencies for molecular genetics, cytogenetics and clinical genetics. The Chairman invited discussion of the documents particularly with regard to the long term feasibility and resources required to provide the information.

After discussion, it was felt it would be helpful if the cytogenetics contracting currencies could be redrafted to remove the capital component, so that they were on the same basis as the molecular genetics currencies. The Joint Committee then identified that capital equipment procurement was a cause of major concern as different policies around the country were leading to inequities between laboratories. The Joint Committee recommended that this should be a topic placed before GENCAG, and the Chair agreed to write to Sir John Pattison to ask that this be placed on the agenda.

Mr Kent pointed out that the contracting currencies applied to only one aspect of regional genetics centres; he was concerned that education and training may not be appreciated to be integral components of the work of the services. The Chair agreed to write to Mrs Barbara Gill, responsible for the specialised services definitions to confirm that the education and training components would be commissioned separately from the workload in the contracted currencies.

Action: Chairman

The Chairman confirmed that he had written to colleagues in biochemical genetics at the request of the specialised service definition group to ask them to prepare a similar document on contracting currencies.

The Committee recognised that the contracting currency documents were draft documents likely to undergo considerable change. It was agreed that the professional groups would each ask several units to undertake a trial of the contracting currencies and report back.

The Chairman confirmed that the specialised service definition group regarded the definitions and contracting currencies to be working documents which would require amendment in the light of experience. He would ensure that the contracting currencies were represented as work in progress rather than schemes for adoption.

Dr Collins tabled a “simplified” scheme for contracting currencies in clinical genetics which was felt by the Committee to be more easily workable.

8 Training needs of Genetic Counsellors

a) Training Posts in Regional Genetic Centres

Dr Heather Skirton reported on the training needs of genetic counsellors, which included –

An increased number of practical experiential placements for MSc students.

Funding for travel to placements.

Bursaries for genetic counselling students (similar to those for nursing students).

Funding for genetic counsellor training posts for those with a nursing background.

Resources to allow senior practitioners to devote time to training.

Dr Brecker commented that as well as the Genetics Unit holding an urgent meeting about training requirements and training capacity for genetic counsellors, it was important to work in parallel with the National Genetics Commissioning Advisory Group.

Professor Mueller spoke for the entire Joint Committee in saying that it was an urgent priority to devise a scheme to ensure the necessary numbers of genetic counsellors to enable the future development of genetic services. The Committee asked the Chairman to write to Professor Sir John Pattison to stress the importance of this work.

Action: Chairman

9 National Electronic Library for Health

Professor Haites reported that it was being proposed that a genetics branch library be added to the National Electronic Library for Health. This was likely to be a simple “front end” for health care professionals, with links to other levels of the library giving information on particular conditions. It was being proposed that the genetics part of the library would concentrate on basic genetics information. The British Society for Human Genetics Council were supportive of the initiative, but, like the Joint Committee were concerned that sufficient funds would be available in the future to complete and maintain the library.

As a first step, Professor Haites was convening a workshop to identify in more detail suitable content.

10 Public Health Genetics Unit

The Public Health Genetics Unit had organised workshops on genetics and health economics and cardiovascular disorders. Reports would be available soon.

Dr Warren raised Dr Zimmern's concern (in his absence) about the future commissioning of genetics screening programmes (including those for Down syndrome, the haemoglobinopathies and neonatal metabolic disorders). The Joint Committee agreed that there could be concern about inequity of access and the relationship of these services with the tertiary metabolic disease laboratories if they were to be commissioned at a local rather than national level. This issue is being addressed as part of the specialised genetics service definition.

11 Human Genetics Commission/Department of Health

- 11.1 Dr Brecker reported on the one day meeting held by the Commission to inform its document on Genetics and Insurance.

Joint work is presently being undertaken with the Human Fertilisation and Embryology Authority on preimplantation genetic diagnosis.

- 11.2 Babies conceived by intracytoplasmic sperm injection.

Professor Donnai reported that she had alerted the Chief Medical Officer to the reports of babies conceived by intra cytoplasmic sperm injection having problems which could be related to abnormal methylation. The Chief Medical Officer asked the HFEA to consider this and they have taken expert advice. It is hoped that a detailed follow up study of children born in this country following ICSI will be instituted. Professor Donnai asked that this be kept on the agenda.

12 National Screening Committee

Professor Haites reported on three workshops held by the National Screening Committee on breast cancer, colon cancer and factor V Leiden deficiency. The conclusion of each was that there is no evidence to support genetic screening for the population, but genetic testing should be available for particular sub groups likely to be at highest risk where surveillance would be helpful.

Professor Haites commented that Health Technology Assessment funding had been requested for assessment.

13 Report from United Kingdom Haemophilia Centres Directors Organisation

Dr Fryer, Joint Committee representative on the UKHCDO Advisory Committee, reported

- 13.1 Following assimilation of responses, the document on gene therapy for haemophilia would be considered by the UKHCDO Advisory Committee early in June.

13.2 A consent form for DNA testing and storage has been drawn up with an information leaflet to accompany it. These will be considered by the UKHCDO Advisory Committee early in June. Joint Committee members asked Dr Fryer to bring to UKHCDO's attention to Dr Fiona Douglas's Working Party on Consent and Confidentiality, and asked that a copy of the UKHCDO document be forwarded to Dr Douglas. **Action: Dr Fryer**

- 13.3 A document on testing in childhood has been produced and is being considered by the UKHCDO Advisory Committee in June.

- 13.4 A consultative letter has been sent by UKHCDO to laboratories providing molecular testing to propose a set of agreed standards so that testing should only be performed in those laboratories. Currently only a few laboratories (6 or 7) across the country would fulfil these requirements.
- 13.5 The role of genetic associates, genetic registers and the relationship with clinical genetic services would be discussed at a future meeting.

14 Royal College of Physicians

a) Continuing Professional Development

The Chairman reported that the Royal College of Physicians Continuing Professional Development Advisory Committee was awaiting the decision of the General Medical Council on the precise form of revalidation. The CPD Advisory Committee had written to the Presidents of the three Medical Royal Colleges to ask that a CPD scheme be made compulsory for physicians.

In the meantime, the CPD Advisory Committee recommended that specialties prepare to develop assessment tools, in a form felt to be most appropriate for the characteristics of the speciality.

The Joint Committee agreed to reconsider this when more information was available.

Several Physician members of the Joint Committee had found it extremely difficult to access and use the on-line version of the Royal College's recording scheme for CPD. Those trying to use the paper based system, had, however, found this even more problematic. The Chairman advised the members to contact the CPD department.

b) Specification of Requirements for clinical information systems in support of secondary care

The Joint Committee discussed and supported this document which aims to ensure that all health care professionals working in hospital practice have access to relevant clinical information necessary to support patients in their care, to have on-line access and to provide a basic uniformity of clinical systems.

There was debate about the confidentiality of some genetic information, as in many units it was stored as family based files. There was special concern about the recording of predictive information on computer systems and the Joint Committee felt that this was not appropriate, especially if information on the system could be accessed, as recommended in the specification of requirements, by other departments within the hospital, and perhaps also transferred to organisations out with the hospital.

Dr Warren wondered whether the specification would also apply to cancer registries. Professor Haites reported that the Scottish Executive Health Department had set up a project (ECCI) to ensure that systems currently in use in different Trusts were able to communicate with each other and provide a core data set. The Chairman would write to the Academy of Colleges Information Group supporting the document but raising the particular features required of a genetics information system.

Action: Chairman

15 Manpower and Training

a) **RCPATH SAC**

The Committee has not met since the last Joint Committee meeting. Professor Neva Haites reported that the Manpower Committee of the Royal College of Pathologists discussed the 3 registrar posts in genetic laboratory medicine as they had been asked whether additional posts were required to complement the increased number of clinical registrar posts. The Manpower Committee believed that 3 training posts for clinical laboratory staff were sufficient.

b) **Numbers of Specialist Registrars**

Professor Mueller confirmed that 9 new specialist registrar posts would be released this year, and that Professor Michael Tunbridge, Lead Dean, had suggested that they are placed in accordance with the weighted capitation index. Regional Specialities advisers would receive details shortly. It is expected that an additional 21 posts will be released over the next two years.

c) **JCHMT SAC in Clinical Genetics**

Dr Tolmie had confirmed that by letter the curriculum for specialist registrar training in clinical genetics had been fully updated. As requested by the JCHMT, methods of assessment had been added to the statements of all training requirements.

The JCHMT has not progressed further with its assessment tools, but the assessment tools developed by the SAC and a group of speciality advisers is currently on trial with specialist registrars. Dr Brennan reported that some trainees could not see the need for such a document whilst others felt the document to offer a reasonable method of assessment. The Chairman regretted that the reasoning which had led to the particular form of the assessment document appears not to have been explained as fully as it might to some groups of registrars. The assessment document would be revised in the light of experience.

16 Any other business

- a) Consultant Clinical Geneticists had asked the British Society for Human Genetics to ask the Joint Committee to write to the Royal College of Obstetricians about their concerns about the standards of reports from some private laboratories. After some discussion it was agreed that the Chairman would consult with Professor Haites Chairman of the British Society for Human Genetics and then discuss the matter with Professor Peter Soothill.

Action: Prof Haites

- b) **Disabled Peoples International Europe Position Statement on Bioethics and Human Rights: Disabled speak on the new genetics.**

It was agreed that the Chairman would try to obtain copies of this document and circulate them to Joint Committee members. It was felt that opening a dialogue with DPIE on some of the perspectives expressed should be considered. The Joint

Committee felt that this may be best undertaken by the Genetic Interest Group and the British Society for Human Genetics, and Mr Kent and Professor Haites would discuss this further.
Action: Mr Kent, Prof Haites

c) UK population Biomedical samples collection (the Wellcome Trust and Medical Research Council).

The Chairman reported that the Wellcome Trust and Medical Research Council are discussing the possible establishment of a DNA collection from 500,000 randomly selected volunteers aged 45-64 and were seeking views from people within the NHS and related fields about the project, its proposed management and the kind of impact it might have on local and regional NHS services. The Chairman proposed that a presentation about the project may be appropriate for a future Joint Committee meeting.

17 Date and Time of Next Meeting

Thursday 20th September 2001 at 2.00 pm, Royal College of Physicians.

Wednesday 9 January 2002 at 2.00 pm