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

The sixth meeting of the Joint Committee on Medical Genetics was held at the Royal College of Physicians on Tuesday 27th September 2000 at 2.00 pm

Present

Professor Peter A Farndon	Chairman RCP
Dr Julie Crow	RCPath Registrar
Professor Neva Haites	BSHG Chairman
Dr Stephen Abbs	RCPath
Dr Naomi Brecker	NHSE Observer
Dr John Tolmie	RCP JCHMT SAC
Dr Jill Clayton-Smith	RCPCH
Professor Dian Donnai	CMO Adviser
Dr Rob Elles	BSHG
Mrs Margaret Fitchett	RCPath
Dr Alan Fryer	RCP
Dr Helen Hughes	BSHG
Professor Noor Kalsheker	RCPath
Professor Sue Malcolm	RCPath
Professor Robert Mueller	(RCP)
Professor Peter Soothill	RCOG
Dr Virginia Warren	FPHM
Mr Peter Plume	RCP Committee Administrator

In Attendance:

Dr Cyril Chapman and Dr Ron Zimmern

1 Apologies for absence/Welcome/Introduction

Professor Ian Gilmore (RCP Registrar).

Apologies for absence were received from Mr John Barber (BSHG), Dr Paul Brennan (RCP trainee), Ms Caroline Browne (RCPath trainee), Professor Michael Connor (Scottish Colleges), Dr Dennis Cox (RCGP), Dr Lorraine Gaunt (BSHG), Mrs Penny Guilbert (BSHG), Mr Alastair Kent (GIG), and Mr Anthony Taylor (DH Observer).

2 Minutes

The minutes of the meeting held on 24 May 2000 were confirmed and signed.

3 Matters Arising from the Minutes

3.1 **Patents and genetic testing**

a) Report on BRCA testing discussions

Dr Brecker reported that a "memorandum of understanding" was being drawn up with Rosgen, and was in its final stages of preparation. This would be circulated widely, including to the Joint Committee and comments sought.

It was noted that press reports of an agreement having been signed with Rosgen Ltd were untrue.

Action: the Chairman would distribute the memorandum to members for comment (who may consult widely within their professions) after which he would reply to the Department of Health

b) Rosgen (Document)

The Chairman had received a letter from Rosgen stating their interest to work closely with organisation such as the Joint Committee to ensure that their services were offered "in a proper and responsible fashion". They asked whether a representative could attend the next meeting to address the concerns of members. They also asked whether it would be appropriate to grant Rosgen observer status at the Joint Committee meetings.

It was agreed that the chairman would invite representatives from Rosgen to the next meeting to give a presentation of their proposals for providing a service, and to explain parts of their leaflet (such as the term "genetic counsellor".) Professor Donnai had concerns about the provision of counselling as it appeared that women will not be able to undertake the test commercially unless counselling has taken place. Rosgen would be asked to prepare a summary to be distributed before the meeting, in keeping the committee's way of working.

It was agreed that the time was not right to grant a commercial organisation observer status to Joint Committee meetings, but that in future, an industry representative may be appropriate.

Professor Mueller pointed out that it was important that the committee would be willing to receive presentations from other companies in the future when appropriate.

3.2 **Clinical governance**

a) Clinical Genetics Society Implementation Group

Dr Hughes confirmed that the Clinical Genetics Society had set up a working group of 16 (including trainee representatives) to undertake an implementation and development plan.

Four main topics would be considered:

Guidelines re follow-up and recall
Letters to families
The clinical genetic management of cardiomyopathy (as a model)
Presymptomatic testing

Dr Hughes was contacting NICE to inform them of this initiative.

b) National confidential enquiry into genetic counselling by non-geneticists (CEGEN)

The chairman confirmed that he written to the President of the Royal College of Physicians in response to information about CEGEN received by the President, who had forwarded it the Joint Committee.

One of CEGEN's recommendations had been the need for a national policy for improving undergraduate and postgraduate medical, nursing and midwifery education in genetics, and the requirements for clinical governance. The chairman had outlined the initiatives which the joint committee were instituting, which had been forwarded to Professor Harris. Details of these are in other section minutes of this meeting.

Professor Harris has confirmed that a presentation on CEGEN is being given to NICE at their meeting on 29th November.

3.3 **Genetics proforma for antenatal care** (Document)

The chairman confirmed that he had written to the Presidents of the Royal College of Obstetricians and Gynaecologists, the Royal College of General Practitioners, and the Royal College of Midwives with the proposal from this committee that in the long term there should be an attempt to devise a genetics proforma for use in antenatal clinics. In the interim, he asked for their opinions on reviewing the lists of diseases in the National Pregnancy Record about which specific enquiries were made at the antenatal booking clinic. Responses are awaited.

Professor Soothill commented that the existing National Pregnancy Record was likely to form the template for the electronic record, and therefore work on improving the checklist may be beneficial. He felt that, although more difficult, it may be worthwhile compiling a document from this committee on the standard of "genetic care" which should be offered in the antenatal clinic. The committee agreed that there was a need to define what constituted a "genetic disease", that the standard of care proposed should be deliverable, and that such a proforma would need be accompanied by training opportunities (provided by the genetic community) for antenatal clinic and primary care staff.

Action: Professor Soothill would convene a small group to consider this further

3.4 Medical Devices Agency

a) In vitro diagnostic device directive

The chairman has written to the MDA following advice from Dr R Elles. The Joint Committee has welcomed the directive as a protection for patients from sub-standard test kits.

b) CVS transport medium

The Committee noted an MDA alert over the use of CVS transport to flush out CVS cannulae and supported the recommendation that normal saline should be used.

3.5 **United Kingdom Haemophilia Centre Directors' Genetics Working Party**

Dr A Fryer reported that he had attended three meetings. The Working Party had discussed provision of service, gene therapy, and testing of minors for carrier status. They were also concerned with issues of consent and confidentiality, and will be interested in the report of the Joint Committee's Consent and Confidentiality Working Party. Dr Fryer commented on the similarity of issues concerning both the Haemophilia Directors and this Committee.

3.6 **Membership of Consent and Confidentiality Working Party**

The remit is "to identify issues of consent and confidentiality specifically related to genetics and produce guidelines for practice".

The members of this working party are:
Dr Fiona Douglas (chair)
Professor Alexander McCall-Smith (Vice-chairman, Human Genetics Commission)
Professor Neva Haites
Ms Penny Guilbert
Dr Elaine Gadd
Dr Carol Chu
Mr John Barber
Dr Stewart Payne
Mr Alistair Kent
Dr Judith Goodship
Mr Marek Sergot

The chairman of the Joint Committee will be an ex officio member.

The first meeting is being held in Newcastle on November 27th. Questionnaires will be distributed to genetics units about current practice.

4. Nuffield Trust Genetics Scenario Project (Document)

This report was warmly welcomed by the committee, and Dr R Zimmern led a discussion on the main recommendations. The aim had been to assess the likely impact of genetics and so lead to recommendations about services. The two main drivers had been identified as the delivery of the science and public acceptance. Recommendations were in seven main areas: regulatory framework, educational strategies, information and confidentiality, financial framework for health, commercial considerations, investment in the basic science base and health and health service provision. It was noted that there was a need to foster partnerships between academia, the NHS and industry particularly for the last three.

The Chairman was concerned to know if a mechanism existed for implementation of the report's recommendations. Dr Zimmern advised that the Nuffield Trust was organising a Scoping Meeting on October 25th to inform a further meeting in the Spring.

5 Public Health Genetics Unit

Dr Zimmern reported that the first part of the Summer School for Commissioners had been oversubscribed and much appreciated by participants.

He expressed his concern that no formal links existed between genetics committees and the National Screening Committee. The Joint Committee noted that one of our members, Professor Neva Haites, was a member of the National Screening Committee.

Dr Zimmern advised members that he was currently a member of the Health Technology Assessment Screening Committee, and that from 2001, he would take over as Chairman.

He presented two new initiatives of the Public Health Genetics Unit:

a) Setting up a network of health economists to consider issues in the provision of genetic services

The Committee welcomed this initiative to inform the debate about outcomes of genetic services, but Professor Donnai expressed the hope that the group would address and study issues such as health gain rather than using simple measures (for instance, counting numbers of patients seen/samples processed).

b) Applying the "cancer model" to other groups of diseases

Dr Zimmern suggested that the model which has been applied to determine which families are at highest risk of inherited forms of cancer might be applied to cardiac disorders, such as cardiomyopathies. He had been in discussions with Professor Steve Humphries, and a one day meeting was being proposed in the Spring to explore this.

6 DNA Services

a) Working Group on Laboratory Services in Genetics (Document)

A copy of the summary of the report of this Working Group (Chaired by Professor Martin Bobrow) had been circulated to Joint Committee Members. Dr Brecker introduced the report. Although the main focus was on laboratory services, the report emphasised that these were interconnected with the provision of clinical services. The report recommended the need for a national strategy and co-ordination, with the setting up of a national group to oversee this. Dr Brecker confirmed that the Working Group report was to be on several agendas, including that of the NHS Executive Board. Any feedback would be welcomed from the Joint Committee.

The Chairman undertook to distribute the report's appendices by email to the Joint Committee. A printed version of the complete report would be distributed to Joint Committee members immediately on publication which was expected within a few weeks.

Action: Chairman

Professor Kalshaker noted disappointment that the report did not address how the genetic laboratory services would link up with the established network and infrastructure of pathology laboratories in other disciplines. Mrs Fitchett agreed that it was important that

there was co-ordination of services provided by genetic laboratories and those genetic laboratory tests provided by other pathology laboratories.

The Chairman was asked to write to welcome the report as its proposals will strengthen the service to patients, but there was concern that a mechanism be agreed for the recommendations to be instituted. The Chairman thanked the British Society for Human Genetics for agreeing to defer its own proposals and plans for laboratory genetic services until Professor Bobrow's report had been published, but in so doing the British Society had been very mindful of the problems encountered now by the genetic laboratories and how urgent solutions were needed. The Chairman would emphasise to the Department of Health there were problems now which needed to be addressed in the light of Professor Bobrow's report for the future.

Dr Brecker asked that if a national advisory group to implement the report were agreed, the Joint Committee would work with the Department of Health in setting up such a group. The Committee welcomed this.

b) Letter from British Society for Human Genetics (Document)

The President of the Clinical Genetics Society and Chair person of the Association of Genetic Nurses and Counsellors had written to the President of the British Society for Human Genetics with concerns about adequate pre and post test counselling and information with regards to the potential growth of molecular genetic testing in non NHS laboratories. There was concern over potential misinterpretation of molecular tests by untrained professionals. It was important that counselling to the standards found in the NHS genetic services were explicitly funded in service agreements where private laboratories may be asked to provide molecular testing for the National Health Service, and that genetic tests provided privately had a level of counselling and support to an agreed standard.

In discussion, education and training of non genetic professionals were again highlighted and the workforce planning implications these would have on the existing genetic services. Dr Brecker confirmed that the Department of Health was very aware of many of the issues contained in the letter, and that the clinical and laboratory service requirements of genetic testing for predisposition to familial forms of cancer were recognised in the national cancer strategy.

The Chairman undertook to ensure that the letter had been passed on to the Department of Health.

7 **Human Genetics Commission** (Document)

a) The work programme of the Human Genetics Commission was noted. The HGC has decided that its first priority should be to set up a working group relating to storage protection and use of genetic information. The HGC will continue to review (either through its own sub groups or through links with outside organisations) proposals in relation to NHS genetic services (through the genetics strategy project), developments in genetic testing, social and ethical issues in relation to patents, and reproductive choice issues.

b) Patients panel. The Chairman had written to Minister Yvette Cooper welcoming the establishment by the Human Genetics Commission of a patients panel. The Joint Committee had agreed with the Genetic Interest Group that the absence of anyone representing patients with genetic disorders was a major omission on the HGC, although there were representatives from the British Association of Disabled People and the Consumers Association.

8 **Department of Health/NHS Executive**

a) References to genetics in "the NHS plan". (Document) It was noted (paragraph 11.15) that the Government intends to commission NHS research and development in "medical knowledge parks" to "evaluate all aspects of the emerging developments in genetics, from the laboratory testing to the requirement of counselling of patients". As far as could be ascertained, no further information about these is available yet.

b) Revised guidance on laboratory containment measures for work with clinical cytogenetics and tissue samples. (Document)

The Joint Committee supported this guidance.

c) DH/NHSE Review of Genetic Services

Dr Brecker commented on the need to break down administrative boundaries between the Department of Health and the NHS Executive and that a new unit was being established with Sir John Pattison as Sponsor/Director. This would bring together currently disparate groups in Government involved with human genetics. Further details were awaited.

d) Commissioning Workshop, June 2000

A Workshop for Commissioners had taken place in June to encourage sharing of good practice over current commissioning arrangements. In addition, work on future arrangements for commissioning genetic services (as with other regional specialties) was being undertaken with the London Regional Specialised Commissioning Group taking the lead.

Several members of the Joint Committee had attended a meeting earlier in the day where the London Regional Specialised Commissioning Group were considering what should be included in the definition of genetic services as a specialty. A draft definition had been produced and further work on this by members of the Joint Committee would be welcomed.

It had also been identified that different genetics units used different contract currencies. The London Regional Specialised Commissioning Group had requested that the Joint Committee consider work towards the obtaining of consensus of clinical and laboratory contract currencies. Dr Elles commented that the Clinical Molecular Genetics Society already collects standardised data and the format of these may be useful in further discussions. It was noted that several different systems for measuring clinical activity were in use.

It was agreed that the Chairman would consult members outside the meeting and consider forming a working party, involving regional genetics centres.

e) Genetics Strategy project

Dr Brecker reported that the Planning Division of the Department of Health was undertaking a genetics strategy project to identify service models which might be appropriate for 2010. This project should be reporting towards the end of the year.

9 **Genetics Education**

It was noted that several other organisations including the Human Genetics Commission, the Wellcome Trust, the Public Health Genetics Unit and nurses and midwives were also considering this subject.

a) Genetics knowledge/education for non genetics professionals

Dr Clayton-Smith noted from the results of her survey that there are very differing thoughts on what sort of genetics education is needed, and that the content and form need to be tailored to different groups with the support of a more formalised structure.

b) Discussion on genetics education for Physicians; the Royal College of Physicians Medical Specialties Board.

The Chairman reported on the response from other medical specialties to a document he had prepared asking for views on genetics education for physicians. Responses have been received from general internal medicine, genito-urinary medicine, gastroenterology and hepatology, rheumatology, clinical pharmacology and therapeutics and geriatric medicine. They were remarkably similar asking geneticists to suggest the advances in genetics which would be important for the practice of their specialties. They also suggest that objectives and core competencies for specialist registrars in their subjects with regard to genetics be identified. This teaching should be provided by geneticists! The President of the London Royal College has asked the other specialties to reply.

c) Undergraduate medical training in genetics

Professor Haites reported some results from a questionnaire organised by the Wellcome Trust on undergraduate (medical and nursing) training in genetics. Professor Haites felt that there was a willingness to consider a national curriculum for medical schools to ensure that the basic core subjects are covered. The aim would be to develop a consensus view of the curriculum, rather than a specification as to how it should be taught.

The Chairman and Professor Haites would be discussing with the Wellcome Trust a possible source of funding to organise a meeting of medical schools to discuss this further.

d) National training course for genetics

The Chairman commented that setting up a national training course in genetics appeared to be one way forward which would meet many identified needs. In the first instance it would be envisaged as a national modular course for specialist registrars in clinical genetics, but it could be expanded with other modules for registrars in other specialties. The core genetics of such a course would also be appropriate for other professionals in the genetic services.

The Chairman was seeking funding to set up a meeting to consider this further including identifying curriculum content and objectives.

10 **Role of the Clinical Geneticist** (Document)

Dr Hughes presented a report on the role and responsibilities of the clinical geneticist. The Joint Committee felt that this definition would be extremely helpful, especially in manpower strategy discussions.

11 **Genetics Databases: House of Lords Committee on Science and Technology**(Document)

It was noted that many issues raised were within the remit of the Human Genetics Commission, but the House of Lords Committee is investigating current and planned genetic databases. Comments had been received before the meeting (in enclosures) from Dr Fiona Douglas and Dr Mike Creasey and it became apparent that several members of the Joint Committee and its constituent organisations had plans to respond.

12 **Guidance for Ethical Committees on Genetics** (Document)

Dr Cyril Chapman had been invited to discuss the practical problems of submission of genetic projects to ethical committees. Many genetics projects (especially where rare disorders were concerned) involved relatively few patients geographically widely separated. The existing mechanism for multi-centre trials was often inappropriate for genetic projects. Dr Chapman commented that guidance and recommendations for handling projects involving "genetics" would be extremely helpful for medical research ethics committees.

The Chairman believed that national guidance for local research ethics committees and multi-centre research ethics committees was being considered and therefore time was opportune for the genetics community to offer advice and guidance, which he understood would be welcome. It was agreed that Dr Chapman would form a small group to consider this further.

Action: Chairman

13 **Matters from the Royal College of Physicians**

a) Physicians in the pharmaceutical industry. (Document)

This RCP publication was discussed to consider whether it required any amendments specifically relating to genetics. It was felt that the document dealt largely and successfully with the issues of prescribing and drug trials, but not with tests performed in a commercial setting. Do NHS clinicians have responsibility to explain to patients the significance of a genetic test performed through a pharmaceutical company? It was not considered that gene therapy and gene therapy trials posed any matters different from those already in the document.

Pharmaco-genetics was seen as one area where there may be ethical issues related to whom will be performing genotyping so that drug treatment can be tailored. Will the biotechnology/pharmaceutical companies themselves want to supply the tests with their drugs? It is obviously good practice that a physician should be aware of any test that is available that would alter a person's drug response and institute such tests. The same principles apply to consultancy fees as in the existing document.

A Clinician's relationship with a pharmaceutical company who wishes to determine susceptibility to multi-factorial diseases by testing large numbers of patients was also discussed.

These comments will be passed on to the College.

b) Consultant Physicians working for patients; Manpower assessment

Members of the Committee had previously been involved in identifying the components of a consultant clinical geneticist's work, and suggesting the time commitment associated with these components. It had been possible, assuming a full-time consultant providing a clinical service (without clinical director responsibilities, teaching or research) to calculate that a minimum of three whole-time equivalent consultant clinical geneticists were required per million population. This figure was remarkably similar to that estimated, using different methodology, by the Clinical Genetics Society recently.

c) GMC Proposals for revalidation

The Chairman had consulted several members of the Committee to formulate a response to the Royal College of Physicians about the applicability of the General Medical Council's proposals for revalidation to clinical genetics. It had been felt that the GMC proposals for revalidation would apply to the specialty of clinical genetics without amendment.

d) Continued professional development committee

It was noted that the London Royal College had set up a continuing professional development committee.

14 **Manpower and Training**

a) RCPath, SAC Professor S Malcolm

It was noted that there were two unfulfilled NTNs in genetic pathology.

The "genetics curriculum" is available in the "trainees" section of the Royal College of Pathologists website.

b) SWAG specialty review clinical genetics Professor R Mueller.

The Joint Committee was delighted to hear that, after a great deal of pressure over a considerable period of time from the genetics community, it appeared that there would be no further reduction in training numbers for specialist registrars in clinical genetics, and that there was the possibility of 30 new posts being made available over the next 3 years to fill the projected number of consultants. The Joint Committee recommended that consideration be given to placing trainees in centres where all the educational objectives can be met, rather than distributing the trainees on an even geographical basis.

c) JCHMT SAC in clinical genetics Dr J Tolmie

Dr Tolmie commented that JCHMT wishes the curriculum to be re-written in a standard form, and that the first draft should be available by the end of the year. The new form makes competencies clear, but Dr Tolmie warned that increased consultant time would be required to administer and meet the new system.

15 **Publications received**

a) RCOG/RCPath Joint Working Party Report on Fetal and Perinatal Pathology (awaiting publication on RCOG and RCPath websites)

b) Genetics Law Monitor (http://www.geneticslawmonitor.com)

c) Genetic Research and You (leaflet from Consumers for Ethics in Research)
(Document)

d) Genetic screening: technical and ethical issues. Recommendations and background document from the European Society of Human Genetics Professional and Public Policy Committee.

16 **Dates of Future Meetings**

Tuesday 16 January 2001 at 2.00 pm at the RCP
Wednesday 23 May 2001 at 2.00 pm at the RCP