

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 seventh meeting of the Joint Committee on Medical Genetics was held at the Royal College of Physicians on Tuesday 16 January 2001 at 2.00 pm

Present

Professor Peter Farndon	Committee Chairman, RCP
Professor Neva Haites	BSHG Chairman
Dr Stephen Abbs	RCPPath
Mr John Barber	BSHG
Dr Naomi Brecker	NHSE Observer
Dr Paul Brennan	RCP Trainee
Ms Caroline Browne	RCPPath Trainee
Dr Jill Clayton-Smith	RCPCH
Professor Dian Donnai	CMO Adviser
Dr Rob Elles	BSHG
Mrs Margaret Fitchett	RCPPath
Dr Alan Fryer	RCP
Dr Lorraine Gaunt	BSHG
Dr Helen Hughes	BSHG
Mr A Kent	GIG
Professor Sue Malcolm	RCPPath
Professor Robert Mueller	RCP
Dr Heather Skirton	BSHG
Professor Peter Soothill	RCOG
Dr John Tolmie	RCP JCHMT SAC
Dr Virginia Warren	FPHM
Mr I M Andrews	RCP Committee Administrator

In attendance

Dr R Zimmern, Professor R Winter

1 Apologies for absence/Welcome/Introduction

Apologies were received from Professor Mike Connor (Scottish Colleges), Dr Dennis Cox (RCGP), Dr Julie Crow RCPPath (Registrar), Professor Ian Gilmore (RCP Registrar), Professor Noor Kalsheker (RCPPath) and Mr Anthony Taylor (DH Observer).

The Chairman welcomed Dr Heather Skirton who has become one of the BSHG nominees following Ms Penny Guilbert's retirement from the Committee, Professor R Winter and Mr I M Andrews acting as the temporary Committee Administrator.

2 Minutes of the last meeting

The minutes were agreed as a true record of the meeting with the addition of the words "if supported by the Royal Colleges" in the statement about action for item 3.3. Mrs Fitchett asked that the title of number 6 DNA services be changed to laboratory services.

3 Matters Arising from the Minutes

3.1 Patents and genetic testing

a) Report on BRCA testing discussions

Dr Brecker reported that Rosgen had entered voluntary liquidation before the "memorandum of understanding" agreed with the Department of Health over BRCA1 testing could be put into place. The Department of Health was undertaking "fact finding" discussions with Myriad Genetics to understand their position.

3.2 National Pregnancy Record

The chairman reported that the Royal College of General Practitioners and Royal College of Obstetricians and Gynaecologists had responded positively about the possibility of amending the national pregnancy record and in the long term developing a genetics proforma for antenatal care. A response from the Royal College of Midwives is still awaited.

Professor Soothill recalled the previous discussions as to whether the Joint Committee should consider a "best practice summary" or to try directly to influence current practice. The Joint Committee had accepted this latter route.

Professor Soothill suggested that a small group be set up to consider this further. He suggested that the remit should be to consider the recording of genetic and family histories, but to link this with actions to be taken. He suggested that the method of working would be that each working party member would independently devise a suitable form for subsequent discussion by all members. The results would then be reported back to the Joint Committee. The membership would be Professor Peter Soothill, Mr Ralph Settatee (nominated by the Royal College of Obstetricians and Gynaecologists), Dr Lindsay Smith (nominated by the Royal College of General Practitioners), Professor Robert Mueller (Clinical Geneticist) and a midwife. The chairman would write to the Royal College of Midwives.

In the reply from the Royal College of Obstetricians, Professor Rodeck had commented that a review of documents might include documenting whether DNA had been stored on a neonate with a genetic disorder. The Joint Committee discussed this suggestion but felt that the issues surrounding consent and confidentiality and the storage of DNA were out with the remit of this working party.

b) National screening initiative

Dr Warren asked whether the working party's remit was already part of the initiative of the National Screening Committee on antenatal Genetics which was undertaking a systematic quality assurance of antenatal screening. A national co-ordinator and "regional" co-ordinators in England had been appointed. The Joint Committee welcomed this move to better quality in antenatal screening, but several members were concerned that there appears to have been no consideration of the impact on the genetics services through increased numbers of DNA and cytogenetic tests. The Committee asked the chairman to write to the National Screening Committee to ask how links could be strengthened between the two Committees.

Action: Chairman

3.3 **Consent and Confidentiality Working Party**

The working party has held one meeting to design the questionnaires on current practice which will shortly be circulated to clinical and laboratory departments in all regional genetics centres. The chairman, Dr Fiona Douglas, is hoping that preliminary results will be available for the next meeting of the Joint Committee, with a final report in time for the September meeting.

The Clinical Molecular Genetics Society had written to the chairman of the Joint Committee about the storage of DNA from patients in whom there is an uncertain diagnosis or where there is currently no DNA test available. Many molecular genetics laboratories also keep DNA once tests are carried out and reported. The CMGS were concerned about ethical issues in the light of problems of storage of pathological specimens in several centres around the country in recent months. The working party on consent and confidentiality will be asked to consider this also.

3.4 **Genetics Education**

a) Undergraduate medical training

The chairman confirmed that plans were still being discussed with the Wellcome Trust about devising a national undergraduate curriculum in medical genetics.

b) National training course for genetics

The chairman reported that the Department of Health was willing to support the development of a curriculum for post graduate training in medical genetics for specialist registrars.

c) Training in medical genetics for dermatologists

The chairman had received a letter from the Joint Dermatology Committee welcoming the possibility of receiving genetic curriculum guidelines for their specialist registrars. They have nominated Professor Jonathan Reece to liaise with the genetics community.

The chairman will liaise with Dr John Tolmie (Chair Clinical Genetics SAC) as to whether core competencies for our own specialist registrars would be useful for the dermatology SAC until the curriculum for our specialist registrar training course has been devised.

3.5 Guidance for ethical committees on genetics

Dr Cyril Chapman was asked after his presentation at the last meeting to set up a small working group consisting of Dr John Yates, Dr John Dean, Dr Stephen Abbs and himself.

Dr Chapman reported by letter that a recent circular from Professor Stacey confirmed that it was now possible to apply to one multi-centre research ethics committee for a nation-wide study. After MREC approval has been obtained, the local research ethics committee of a patient is to be informed about the research, but it is not necessary to seek consent from the LREC.

4. Genetics Insurance Committee

Professor Donnai, currently acting chairman of this Committee, reported on its recent work. She pointed out that the Government had set up GAIC with a very specific executive function, as a mechanism for the approval of insurance industry proposals to use DNA tests for insurance purposes. The Committee's role was to establish whether a specific test was relevant and accurate, actuarially, clinically and technically.

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.

Professor Donnai gave her personal view that GAIC needed more members and a broader expertise.

Professor Mueller led a discussion on the ethical and social aspects of the work of GAIC and was concerned that some of its work could be contravening the Human Rights Act. Professor Donnai reiterated that GAIC had been charged with an executive role – it was the Human Genetics Commission's role to consider ethical, social and legal aspects, and that indeed, the Commission had planned to do so.

Mr Kent confirmed that the Genetic Interest Group had received funding from the Wellcome Trust to consider the impact of the Human Rights Act in a project beginning later this year. He also commented that as GAIC met in open meetings, its deliberations were transparent, rather than insurance decisions being taken behind "closed doors".

It was also noted that the UK Forum on Genetics and Insurance was keeping this under review.

It was believed that the Human Genetics Commission would be discussing this at their next meeting on the 9 February 2001, which is an open meeting.

The Joint Committee wished to keep this item on future agendas and to maintain an attitude of watchful concern.

5 Public Health Genetics Unit

a) Genetics Scenario Project

Dr Zimmern reported that as many of the issues highlighted in the Nuffield Trust Genetics Scenario Project were now being actioned in the newly formed Department of Health Genetics Unit, no specific action was being planned by the Nuffield Trust. However the Nuffield Trust and the Wellcome Trust were to undertake a study to identify organisations who had set up policies and/or committees on genetics.

The Nuffield Trust had also agreed to continue to host meetings of the parties involved in the scenario project.

b) Health economics

Dr Zimmern reported that with the Department of Health and Dr Brecker a meeting was being organised in London in April/May with health economists to try to determine how work on health economics in genetics can be generated.

c) Cardiology

The Public Health Genetics Unit and Professor Steve Humphries were to hold a workshop to identify developments in science which may lead to policy development for services for familial hypercholesterolemia.

d) Public Health genetics unit was also undertaking some work on patents and their service implications.

e) Public Health genetics network

Dr Zimmern reported that he was standing down as chairman to be replaced by Dr Heather Grimbaldeston.

6 Laboratory Services

a) Working Group on Laboratory Services in Genetics

Dr Brecker confirmed that the report had been considered by the Department of Health Board which agreed that the expert national body should be instituted. This will be a sub-group of NSCAG with powers to designate services and be involved in commissioning. There would be representatives from all United Kingdom countries. It is intended that there be one representative from each Regional Specialist Service Commissioning Group together with representatives of the clinical, laboratory and genetics counselling professions. It is intended that the first meeting will be held in March. The group will be called the National Genetics Commissioning Advisory Group. There will also be patient representation.

Dr Brecker also reported that the “knowledge parks” in the NHS plan would be funded through the strategic research and development programme, and Dr Peter Greenaway would be the lead.

Dr Brecker also reported that the work on intellectual property rights had been taken up by the Biotechnology Unit.

7 Human Genetics Commission

a) Consultation document on “Whose hands on your genes”

The Chairman brought the consultation document “Whose hands on your genes” to the attention of the Joint Committee. There was concern that the document presented some current hypotheses as facts, that there was confusion between high penetrance and multifactorial disorders, and that it was difficult to answer the “yes/no” questions in the summary as considerable qualification was felt necessary in answering some of the questions. There was concern over the short time scale for return of responses and to whom the document had been distributed. GIG had procured and sent a copy to every Genetics Unit, for which they were applauded, but the Joint Committee was surprised that the Human Genetics Commission had not undertaken this directly. Several members reported that their organisations were hoping to submit responses. After discussion it was agreed that Dr Fiona Douglas would be asked to write the response of the Joint Committee.

b) Patients Panel

The Chairman reported that Ms Yvette Cooper, Parliamentary Under Secretary of State at the Department of Health, had replied thanking the Joint Committee for its support of the setting up of a patients panel as a resource for the future work of the Human Genetics Commission.

Mr Kent reported that a meeting had been held with the Department of Health Genetics Unit about the setting up of this patients panel. It was intended that membership would go out to tender in accordance with the Nolan rules. Mr Kent felt that if a patients panel had been consulted during the writing of the consultation document a much clearer document could have been achieved.

8 NHS Executive/Department of Health

a) Genetics strategy project

Dr Brecker reported that a genetics strategy review had been undertaken by the policy division to give a 5-10 year view of likely developments and their impact on the provision of services. Susceptibility testing, pharmacogenetics and the role of industry were some of the topics considered. This review would aid future policy discussions.

b) Secretary of State Seminar and Genetics

The Secretary of State, Mr Alan Milburn, had requested a briefing seminar on genetics, held in December. The Chairman and several members of the Joint Committee had been invited participants. The seminar included presentations on the expectations of the “new” genetics, the implications for the Health Service and the public understanding of the issues. There were representatives of industry as well as clinical services. Dr Brecker reported that the Secretary of State wished to consider follow up action and to be kept actively involved. The Chairman confirmed that he felt that it had been an extremely positive meeting and relevant service and research issues had been clearly placed before Mr Milburn.

c) Establishment of Genetics Unit

Dr Brecker reported that the new Genetics Unit was being set up with Professor Sir John Pattison as sponsor Director. This would achieve one management structure for genetics and would enable close links between health policy, the wider social and ethical debate, and health services delivery. Recruitment was underway to new posts.

The Joint Committee warmly welcomed the setting up of this Unit as it appeared to offer a mechanism for achieving many of the issues which have been, and currently are, of great concern to both clinical providers and researchers in genetics. The Department has confirmed its intention of working with the Joint Committee and the genetics community generally in developing its advice to Ministers on how best to respond to the challenges which developments in genetics present for the NHS and for society.

d) Genetics Commissioning Group

This new national group (recommended in the report of the expert working group on Laboratory Services for Genetics chaired by Professor Martin Bobrow) will provide a strategy and national co-ordination for genetic services. It will function as a sub-group of the National Specialist Commissioning Advisory Group. (see also 6a)

The Department of Health Genetics Unit had asked the Joint Committee to help identify potential clinical, scientific and academic expert members of the new group. It was agreed that the Chairman of the Joint Committee would seek expressions of interest through the existing professional organisations and regional genetics centres, and relay these to the Department of Health.

e) The Joint Committee wished to record its thanks to the observers from the National Health Service Executive and Department of Health for their parts in passing on to appropriate agencies the results of often rigorous discussions in this Committee, particularly regarding the key strategic issues identified by the genetics community.

9 National Specialised Service Definitions - Genetics

a) The Chairman explained that the London Regional Specialist Commissioning Group was undertaking work to draw up national specialised service definitions, principally to identify to commissioners what clinical activity within a speciality is regarded as “specialised”. The definitions are an aid as to what should define the service for commissioning – they are not intended to be service specifications. After considerable debate, there was broad agreement that what was included in the definition was considered “specialist” genetics activity. There was considerable debate about whether screening services should be included within the genetics definition or within some other definition, and this point would be relayed to the London Specialist Commissioning Group by the Chairman.

It was recognised that some genetics activity (for instance perhaps the inherited haematological disorders) would fit better for commissioning purposes within other specialist definitions.

Professor Soothill was concerned whether aspects of fetal medicine should be within the genetics or maternal health definition, and he agreed to speak to the head of the Regional Commissioning Group directly about this.

b) Contracting currency

As part of the national specialised service definitions, contracting currencies needed to be included, again for commissioning purposes.

The Joint Committee Chairman had agreed that he would ask professional groups on the Joint Committee if they would be prepared to undertake work to agree a set of contracting currencies, the collection of data for which would be achievable in daily working practice. The Association of Clinical Cytogeneticists have agreed to set up a working party, and the Clinical Molecular Genetics Society Audit Sub-Committee have also agreed to undertake this work. Professor Winter and Dr Skirton on behalf of the Clinical Genetics Society and Genetic Nurses and Social Workers Association would consider a minimum clinical data set and potential contract currencies. It was agreed that the working deadline would be the end of March, and these would be discussed at the next Joint Committee.

10 Healthcare 2020 - Report of the Foresight Healthcare Panel

The chapter on “Research, development technology” (containing the recommendations on genetics) had been circulated and Professor Donnai spoke to this Report as she had been a member of the Foresight panel. The Joint Committee wondered whether many of the points in the report would now be subsumed in the work of the new genetics unit, and it was agreed that the Chairman would contact the Department of Trade and Industry (who had sponsored the Foresight panel) to clarify the situation.

11 National Screening workshops

Professor Haites reported on the workshops being held by the National Screening Committee, including those on colorectal cancer and breast and ovarian cancer. Draft documents were being produced following the workshops and these would be submitted and considered by the National Screening Committee. Professor Haites would keep the Joint Committee informed.

12 Matters from the Royal College of Physicians

a) Continuing professional development

The Chairman reported that a new scheme for Continuing Professional Development for physicians was being introduced from April 2001.

b) Consultant Physicians working for patients

The College was preparing a new edition of this document to give greater detail about the work of individual specialities including facilities required, workload and manpower and training requirements.

13 Manpower and training

a) RCPATH SAC

Professor Malcolm had nothing to report.

b) Professor Mueller was able to report that it is being proposed by SWAG that the number of Specialist Registrar training posts in Clinical Genetics be increased (possibly by 30 over the next 3 years). This was in response to projected manpower needs for Clinical Geneticists. It is likely that Deaneries would be asked to produce an action plan later this year to implement this increase.

c) JCHMT SAC in Clinical Genetics

Dr Tolmie reported that the curriculum for Specialist Registrars in clinical genetics had been updated in line with current JCHMT requirements and he expected this to be finalised by the end of May.

A group of Regional Specialty Advisors, in conjunction with the SAC, had developed a new appraisal and assessment scheme for clinical genetics which would be discussed at the next meeting of the SAC, and further details brought to the next meeting of the Joint Committee.

14 Publications received

- a) Governance in acute general medicine
- b) Nuffield Council on Bioethics Report 1992-99

15 Any other business

- a) Mr Kent reported that the European Parliament had set up a "European Parliamentary temporary committee on the human genetics and other new technologies of modern medicine". The Chair would be Dr Goebells; the remit of the Committee was awaited.
- b) It was noted that the British Medical Journal were intending to produce a special issue on "Genetics in Practice" – with a likely publication date in March 2001.
- c) Professor Haites reminded the Committee that its constitution said that the Chairmanship rotated every 3 years between the Royal College of Physicians, the British Society for Human Genetics and the Royal College of Pathologists. The current Chairman, Professor Farndon, will complete his 3 year term at the end of 2001. In view of the work currently being undertaken by the Joint Committee, she proposed that Professor Farndon should be asked to remain as Chairman for at least one further year. This seconded by Professor Mueller and unanimously agreed by the Joint Committee.

16 Dates of future meetings

Thursday 20 September 2001 at 2.00 pm at the RCP