

## *Joint Committee on Medical Genetics*

The Royal College of Physicians

The British Society for Human Genetics

The Royal College of Pathologists

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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 Thursday 20<sup>th</sup> September, 2001 at 2.00 pm

### Present

Professor Peter A Farndon	Chairman 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
Dr Paul Brennan	RCP Trainee
Dr Virginia Warren	FPHM
Ms Dianne Kennard	DH Observer
Dr J Tolmie	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
Dr R Zimmern	Observer
Dr F Douglas	Chair Working Party Consent & Confidentiality
Mrs Val Knight	RCP Committee Administrator

### **1 Apologies for absence/Welcome/Introduction**

Apologies for absence were received from Professor I Gilmore, Registrar RCP, Mr J Barber BSHG, Professor JM Connor (Scottish Colleges), Dr H Skirton BSHG, Mr A Kent GIG, Dr J Crow Registrar RCPPath, Professor Mueller who was unwell

Professor Farndon welcomed Dr R Hapgood RCGP, Dr A Green RCPPath, Ms E Woodeson DoH and Dr F Douglas, Chairman Working Party Consent & Confidentiality.

### **2 Minutes**

The minutes of the last meeting of 23<sup>rd</sup> May 2001 were accepted as a true record and signed.

### **3 Matters Arising from the Minutes**

#### **3.1 Patents and genetic testing**

A BSHG proposal re candidate licence holder for BRCA1 testing.

The British Society for Human Genetics (BSHG) had written to the Joint Committee raising the possibility that existing diagnostic laboratories be considered as a candidate licence holder with Myriad Genetics who were still seeking a UK partner. The Joint Committee welcomed this suggestion and would support the BSHG in discussing the proposal further with the Department of Health and Biotechnology Unit.

B Dr Brecker reported that Myriad was still in negotiation with prospective UK partners. The Department of Health and the Biotechnology Unit were aware that several states had voluntarily withdrawn BRCA1 testing in the light of Myriad's enforcement of their patent. A challenge to the patent was being mounted in France, and possibly by genetics colleagues in the Netherlands and Belgium. Dr Brecker and Dr Jecock would keep the issue under review.

#### **3.2. Genetics and Insurance**

The Joint Committee noted the statements from the Association of British Insurers, the BSHG and the United Kingdom Forum on Genetics and Insurance.

The report from the House of Commons Committee is still awaited, as is the appointment of a new chairman for the Genetics and Insurance Committee. GAIC have some applications for the use of genetic tests still "on the table". Ms Woodeson confirmed that Ministers are addressing the issue urgently, with a mandate to the Department of Health to meet with the Association of British Insurers and take the issue forward.

#### **3.3 Patients Panel of the Human Genetics Commission**

Ms E Woodeson advised that a consultative panel of 100 people with genetic disorders and their carers was being set up. The Joint Committee welcomed this initiative.

#### **3.4 Clinical Governance**

Dr Hughes reviewed the work of the Clinical Governance sub-committee of the Clinical Genetics Society, and its links with the RCP London Clinical Governance committee. The sub-committee had produced a paper on follow up and recall in clinical genetic practice, and guidelines on pedigree drawing and management of hypertrophic cardiomyopathy ([www.bshg.org.uk/society/cgs/cgs.htm](http://www.bshg.org.uk/society/cgs/cgs.htm)).

The Joint Committee welcomed these guidelines for clinical practice and discussed how they might be promulgated to other colleges. It was agreed that the route would be through the Joint Committee.

The CGS sub-committee would consider inviting a representative from the other colleges to its Clinical Governance sub-committee when appropriate.

College representatives on the Joint Committee would discuss with their colleges how best to bring these future guidelines on genetic practices for discussion and dissemination. Professor Soothill would discuss this with the RCOG President, Dr Ruth Newbury-Ecob with the RCPCH, and Dr Rhydian Hapgood with the RCGP.

Dr Hapgood reported that a virtual e-mail network for General Practitioners on genetics issues had been set up.

### **3.5 National Electronic Library for Health**

Professor N Haites advised that the BSHG was setting up a small working group to discuss content, which could be placed on the BSHG website if funding for the National Electronic Library for Health was not secure.

### **3.6 Training Needs of Genetic Counsellors**

Dr H Skirton reported that registration had been introduced on 1 July 2001, with the primary aim to ensure the introduction of minimum requirements.

Dr Brecker confirmed that the Department of Health was funding additional places on the Manchester MSc course, and individual modules on the Cardiff course.

Discussions about training for different skill levels – including broader generic skills, and specific skills below those required for registration as a genetic counsellor – were to be held.

Professor Soothill asked whether midwives were to be included in the training initiative. The Joint Committee wished to know if there were links with the Antenatal Quality Programme and the Chair agreed to write to Pat Ward.

### **3.7 Training Posts for Genetic Laboratory Scientists**

a) Mrs Fitchett voiced the concern of the whole committee over the training of sufficient numbers of staff for genetic laboratories. Issues include problems in recruitment to the training posts (which are insufficient in number) and being able to release trainers from clinical service duties. Potential solutions include an increase in training posts and provision of supernumerary trainers, and/or centralisation of training within selected centres within the UK.

The Joint Committee suggested that a small working group be set up to identify the numbers of A-grade trainees, facilities required and whether centralisation is the best option.

It is unclear whether the funding stream should be via service commissioners or educational consortia.

The Chair would forward letters he had received to Ms Woodeson who would take this forward at the Department of Health. He would also write to bring this to the attention of the Chairman of the Green Paper Working Party.

b) Training in laboratory biochemical genetics

Dr Green discussed similar concerns about training; a document was in preparation. The Joint Committee recommended that the document be sent to Mike Hallworth at the Association of Clinical Biochemists and Graham Beastall, Standing Committee for Clinical Science at the Royal College of Pathologists, and that the concerns be discussed with Commissioners at GENCAG.

The Chair also suggested that Mrs Green write to Dr Peter Greenaway, Chief Scientific Officer at the Department of Health. Ms Woodeson confirmed that she would ensure that the need for additional training in biochemical genetics would also inform Department of Health discussions.

### **3.8 Babies conceived by Intracytoplasmic Sperm Injection**

Professor Donnai had brought to the attention of the Chief Medical Officer reports that babies conceived by intracytoplasmic sperm injection were having problems which could be related to abnormal gene expression. The Human Embryology and Fertilisation Authority had convened a large meeting, chaired by Ruth Deech, to discuss the issue. A questionnaire for parents was proposed but the meeting did not address the underlying scientific concerns. It was hoped that Mr Anthony Taylor would be preparing a report for the Department of Health on this.

Although information would become available from the proposed questionnaire for parents, there was concern that as this was relatively new technology, there was no formal mechanism for information about the health and development of children to be obtained. Dr R Zimmern suggested that this could be a public health surveillance issue, and could be considered separately from the scientific issues. He suggested that this be a priority for discussion at the HTA Diagnostic and Screening panel.

The Joint Committee agreed that more research was needed and asked that the Chair write to Dr Naren Patel at the MRC to suggest that this might be a topic for further research.

### **3.9 Standards of Reports from Commercial Laboratories**

Professor Haites had written to Baroness Kennedy, Chair of the Human Genetics Commission about standards in commercial genetics laboratories, especially over requirements for CPA accreditation and mechanisms which might ensure NHS patients whose samples are tested in private laboratories are assured of the same quality assurance as is expected of NHS laboratories. Professor Haites will report back.

### **3.10 UK Population Biomedical Samples Collection**

Carried forward to a future meeting.

## **4 Public Health Genetics in the United States of America**

Dr M Khoury who had been invited to speak on this item was unfortunately unavailable due to the terrorist atrocity in the United States of America.

## **5 Reports of the Joint Committee in Progress**

### **5.1 Genetic history at antenatal booking visits**

The document has been presented to Royal Colleges but responses are yet to be received. It is hoped that the document will be available on the RCP and BSHG web sites. Professor Farndon, on behalf of the Committee, congratulated and thanked all members of the working party for a clear and excellent document.

### **5.2 Consent & Confidentiality Working Party**

Dr Fiona Douglas presented the draft document. Consultation confirmed that consent seemed to be a major issue for genetics units around the country, with a view that many issues of confidentiality had already been addressed in working practices.

It would be especially important that the Joint Committee document be complementary to and able to be read in parallel with the document on consent from the Royal College of Pathologists.

Following a lengthy and helpful discussion, Dr Douglas would prepare the final report to be considered at the next meeting.

### **5.3 Genetics Education**

#### **(a) Undergraduate Medical Education**

The BSHG had established an working group of around 30 individuals, chaired by Professor N Haites, to establish the core educational objectives for a medical undergraduate course. The group is collating teaching resources and examination questions.

#### **(b) Specialist Registrars in clinical genetics**

Meetings with Specialist Registrars in clinical genetics and Consultants had been held to inform discussions about the “basic genetics” course. The Specialist Registrars would prefer the course to be a week long, annual course with ongoing tasks and case interpretations to reinforce learning. They wish the course to follow a definite structure showing how the different disciplines of basic genetics fitted together. They wish the course to have good teachers rather than experts. Opinion was divided between the Specialist Registrars and Consultants over whether formal assessment should be part of the course.

### **5.4 Guidance for Ethical Committees on Genetics**

The working party would present their document at the next meeting.

## **6 National Specialised Service Definition : Genetics**

The Committee endorsed the Medical Genetics definition, no. 20.

## **7 Cardiovascular Genetics**

The President, RCP, had requested the Joint Committee's comments on the report of a workshop organised by the Public Health Genetics Unit on cardiovascular genetics. Dr Zimmern explained that the workshop was designed to explore the issues, with a future workshop planned to discuss the action required. The Joint Committee welcomed the report and supported the proposed workplan.

The Joint Committee was particularly interested in the mechanisms for the planning and implementation of potential services for familial hypercholesterolaemia and other single gene disorders. It is important to include the relevant genetics in each of the National Service Frameworks, as it is unlikely that there will be a NSF for genetics.

Dr A Green suggested that the Chemical Pathology Committee at the Royal College of Pathologists be asked to comment on the report. Dr Hughes confirmed that clinical governance subcommittee of the Clinical Genetics Society would consider the report and prepare a response, especially in view of its own work on guidelines for the management of cardiomyopathies from the genetics perspective.

## 8 Public Health Genetics Unit

Dr Zimmern, reported on several issues:

### 1. **Commissioning screening services**

The National Screening Committee had confirmed that "screening" was to include the diagnostic test undertaken as a result of a screening programme.

### 2. UK education in genetics outside specialist genetics

The Wellcome Trust has asked the PHGU to review education in genetics in the UK for professional groups including nurses, health visitors, midwives, pharmacists and dieticians. Dr Hilary Burton will undertake the work, which will be circulated to the Joint Committee, the Green Paper Working Party, and GenCAG.

3. A workshop on QPCR and FISH in antenatal diagnosis was being organised in February 2002. Names of potential invitees should be forwarded to the PHGU.

4. A project, funded by the DoH, will look at the impact of patent law and the implications for the NHS.

## 9 National Genetics Commissioning Advisory Group

### 9.1 Work in Progress

Dr N Brecker reported that at the GenCAG meeting in June a draft vision document for the UK Genetic Testing Network and the National Reference Laboratories was discussed. After amendment, the document had been distributed to the genetics community via the Joint Committee for consultation. At its next meeting, GenCAG would be considering mechanisms for supporting the Genetic Network.

Regarding the formation of the UK Genetic Testing Network, Professor Farndon reported that he and Dr Elles had been invited to a meeting of Heads of Genetic Laboratories to discuss concerns. That meeting had agreed that the Clinical Molecular Genetics Society would undertake an audit of service provision of molecular genetic testing, and suggest how the UKGTN might be organised, and the distribution of particular molecular tests between centres.

### 9.2 Developing quality standards

Dr N Brecker reported on the meeting of a small working group to consider quality standards. Members were reminded that at a previous meeting of the Joint Committee a request had been made for members to join this working group, and the offer was repeated. The Joint Committee recommended that the clinical genetics quality standards currently under discussion at the Royal College of Physicians be incorporated into the working group's discussions, and Professor Winter agreed to liaise over this.

### 9.3 Concerns from the Genetics community re commissioning arrangements for 2002.

The Joint Committee was extremely concerned about the lack of a commissioning strategy for specialist services in the reorganised health service for the forthcoming

financial year. Dr Brecker confirmed that a decision had not yet been reached by the Department of Health, but that it was hoped that a paper would be ready for the December meeting of GenCAG. The Joint Committee were so concerned that they asked the Chair to bring the concerns to the attention of the NHS Executive.

## **10 Human Genetics Commission/NHS Executive/Department of Health**

- 10.1 The Human Genetics Commission has issued a newsletter.  
[www.hgc.gov.uk/news](http://www.hgc.gov.uk/news)
- 10.2 Human Genetics Commission Working Group on storage, protection and the use of genetic information.  
A response had been made to this consultation document.
- 10.3 National Reference Laboratories and Knowledge Parks  
Invitations to tender had been advertised.
- 10.4 Green Paper on Genetics  
The Green Paper Advisory Group had met twice. The main focus was on NHS services and the impact of genetics on the NHS.

## **11 National Screening Committee**

Professor Haites reported that the National Screening Committee had considered its future role and whether it should be disbanded. It had concluded that there remained a particular remit which it could fulfil, but that it should work closely with other bodies.

A small committee was being set up to oversee the implementation of neonatal screening for cystic fibrosis. Professor Soothill commented on the importance of the first scan in pregnancy in order to confirm dates.

The Joint Committee had been asked to nominate a member to serve on the Ante-Natal Sub-Group. Names should be submitted to Professor Farndon.

## **12 Matters from the Royal College of Physicians**

### **12.1 Standards for Clinical Genetics Practice**

The College had asked that the standards for Clinical Genetics Practice which had been submitted to the College previously be revisited and expanded. As it was understood that the GMC may be asking for similar standards, it was agreed to wait until further information was available.

### **12.2 An organisation with a memory: risk management**

The Chief Medical Officer had requested information about procedures or protocols, which if agreed protocols are not complied with, could potentially pose a serious risk to patient safety. The Joint Committee identified Predictive Testing Protocols

### 12.3 Relationship with RCP London College Committee on Ethical Issues in Medicine

Professor Farndon reported that he had received a letter from the Chairman of the Ethical Issues in Medicine Committee, Sir John Grimley-Evans, requesting an observer from the Ethical Issues in Medicine Committee to attend meetings. It was agreed that an observer would be invited to attend for specific issues with the minutes of Committee meetings being forwarded on a regular basis.

### 12.4 Consultant Physicians Working for Patients

This document was being published in the near future.

### 12.5 Request for proposals for College Conferences/Lectures 2003

Members were requested to advise Professor Farndon of suggestions for possible conferences and lectures for the year 2003.

### 12.6 To note guidance notes have been produced to assist members of Joint Committees in contributing to relevant NICE Health Technology Appraisals

### 12.7 National Institute for Clinical Excellence: Future Work Programme An audit of the outcome of ICSI was suggested.

## 13 Manpower and Training

#### (a) RCPATH SAC

Professor Malcolm reported that the RCPATH had produced documentation to make its examining techniques transparent.

#### (b) JCHMT SAC

Dr Tolmie reported that an extra thirty training posts were being released over the next three years distributed on a weighted population basis. It was noted that there were problems in securing funding for existing training posts, and concern was raised about the source of funding for the new posts.

## 14 Publications Received

- (1) A Commitment to Quality, a Quest for Excellence: a statement on behalf of the Government, the medical profession and NHS. 27<sup>th</sup> June 2001.  
Available : <http://www.doh.gov.uk/cmoh/cmoh.htm>
- (2) Government response to the report from the House of Lords Select Committee on Science and Technology inquiry on human genetic databases: challenges and opportunities July 2001.

**15 Any Other Business**

Dr V Warren informed members that the Wellcome Trust review of its Biomedical Ethics Programme as available on:

<http://www.wellcome.ac.uk/en/1/mismisethrev.html>

**16 Dates of Future Meetings**

**Wednesday 9<sup>th</sup> January 2002 (RCP)**

**Monday 20<sup>th</sup> May 2002 (RCP)**

**Thursday 26<sup>th</sup> September 2002**

Times to be advised.