

## *Joint Committee on Medical Genetics*

The Royal College of Physicians  
College of Pathologists

The British Society for Human Genetics

The Royal

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An unconfirmed summary of the fifth meeting of the Joint Committee on Medical Genetics, held at the Royal College of Physicians on Wednesday 24<sup>th</sup> May 2000

### Present

Professor Peter A Farndon	Chairman RCP
Professor Ian Gilmore	RCP Registrar
Dr Stephen Abbs	RCPPath
Mr John Barber	BSHG
Dr Naomi Brecker	NHSE Observer
Dr Paul Brennan	RCP trainee representative
Ms Caroline Brown	RCPPath trainee representative
Dr Dennis Cox	RCGP
Professor Dian Donnai	CMO Adviser
Dr Rob Elles	BSHG
Mrs Margaret Fitchett	RCPPath
Dr Alan Fryer	RCP
Dr Lorraine Gaunt	BSHG
Mrs Penny Guilbert	BSHG
Dr Helen Hughes	BSHG
Professor Noor Kalsheker	RCPPath
Professor Sue Malcolm	RCPPath SAC Chairman
Professor Robert Mueller	RCP
Professor Peter Soothill	RCOG
Mr Anthony Taylor	DH Observer
Dr Virginia Warren	FPHM
Ms Hilary Irons	RCP Committee Administrator

### 1 **Apologies for absence/Welcome to new members**

Apologies: Dr Julie Crow (RCPPath Registrar), Dr Jill Clayton-Smith (RCPCH), Professor Mike Connor (Scottish Colleges), Professor Neva Haites (BSHG Chairman), Mr Alistair Kent (GIG), Dr John Tolmie (RCP JCHMT SAC).

New members were welcomed: Stephen Abbs (RCPPath) and Dr Paul Brennan (RCP trainee representative). Professor Neva Haites (Chair, BSHG) and Dr John Tolmie (Chair, Clinical Genetics SAC) had become ex officio members.

### 2 **Patents and genetic testing**

Rosgen had signed a contract with Myriad to provide BRCA testing using Myriad's technology. The Department of Health's lawyers were looking at proposals from Rosgen.

Rosgen had presented their position to the Cancer Genetics Group and this was reported. The Joint Committee expressed concern that restrictions over the numbers of tests which would be allowed to be undertaken in NHS laboratories, as proposed by Rosgen, could result in insufficient tests being available in the NHS. Funding for the commercial provision for the difference in numbers of tests would need to be identified as there was no provision in current genetics budgets.

The Joint Committee was also concerned that Myriad's insistence on a certain technology could preclude advances in technology being made available for patients. It was felt that a Health Technology Assessment was needed.

Rosgen had suggested that it could accept samples for testing only through clinical genetics services to ensure adequate counselling, but concern was expressed by the Joint Committee that clinical genetics services would be overwhelmed if confronted with many patients at low risk.

Arrangements for funding the commercial testing through the NHS had not yet been devised but the views of the genetics professions would be sought by DoH.

### **3 Clinical Governance**

The BSHG report "Towards Clinical Governance in Clinical Genetic Practice" was presented. The Clinical Genetics Society were appointing an implementation sub-committee.

### **4 Review of undergraduate medical training in genetics**

The GMC Education Committee had advised that the Joint Committee should approach medical schools individually for syllabuses and curricula. Arrangements for the review would be presented at the next meeting.

### **5 Genetics proforma for antenatal care**

In response to concerns from the Genetic Enquiry Centre, the Joint Committee discussed the possibility of devising a simple proforma for use in antenatal care to detect families at high risk of a genetic disorder.

The view was expressed that it would be difficult to collect sufficient data nationally and that any proforma would need to be at a very basic level. However, others pointed out that a limited proforma could be used as triage. After some discussion members agreed that ideally a national first trimester screening programme was required during which genetics issues were likely to be identified by specialist staff in ultrasound units. Meanwhile members felt that the existing list in the National Pregnancy Record should be reviewed, and the RCOG, RCGP and Royal College of Midwives would be contacted asking for their views on how the situation might be resolved.

### **6 Services for adults with inherited metabolic disorders**

It was noted that a sub-specialty training module for metabolic medicine is being developed by that speciality.

### **7 In Vitro Diagnostic Medical Device (IVD) Directive**

Mr Steven Lee from the Medical Devices Agency gave a presentation on the implications for genetics laboratories of the IVD Directive (98/79/EC).

Members noted that reagents "manufactured" for diagnostic purposes when used by laboratories outside the manufacturing laboratory's Trust would have to be registered, but reagents used for research would not. The MDA would not have to ensure compliance with the Directive until December 2003. Trusts would be held responsible and not the NHS.

It was agreed that work would be undertaken outwith the Joint Committee meeting and advice from the speciality forwarded to the MDA.

## 8 **Nuffield Trust Genetics Scenario Project**

The project report was to be launched the following day, 25<sup>th</sup> May. The Chairman had been able to read an embargoed copy from Dr Zimmern immediately before the Joint Committee meeting, and commended the report to all genetics units and purchasers. There were recommendations in six policy areas, including the provision, organisation and funding of genetics services. (<http://www.official-documents.co.uk/document/nuffield/policyf/genetics.htm>)  
The report would be discussed in detail at the next meeting.

## 9 **Public Health Genetics Unit**

The PHGU had completed a needs assessment of the care of families with colorectal cancer and was hoping that the surgeons would take this forward.

## 10 **DNA Services**

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

This report from Professor Bobrow's group was in the writing stage, and would be used to work with regional commissioning groups, the NHSE and the Human Genetics Commission. The report would be discussed with the Joint Committee to make sure it was an accurate reflection.

### (b) UK specialist genetic testing network for molecular diagnosis

It was agreed to await the report of Professor Bobrow's working group before taking this further.

### (c) DNA Services for rare single gene disorders

The Joint Committee's previous work has been passed to Professor Bobrow's working group on laboratory services.

### (d) Review of operation of OAT system (Document tabled)

The latest survey of genetics laboratories by Dr Elles for the Joint Committee showed that provider to provider systems seemed to be the most reliable. Concern was expressed as to when the mechanism would be clarified because significant debts were being built up. The Joint Committee's continuing concerns were being reported to DoH/NSHE.

## 11 **Human Genetics Commission/Department of Health**

It was noted that the Commission, under the chairmanship of Baroness Kennedy QC, had met twice and was concentrating on wider ethical issues. A public consultation meeting had been held for feedback on its programme of issues to review. A number of sub-groups and working parties had been established, which included storage and protection of genetic information. From 2001 all meetings would be held in public. All activities were being placed on the world wide web.

The Chairman said that he had had an opportunity to inform Baroness Kennedy about the Joint Committee

The Department of Health had established a new genetics strategy group with the NHSE, led by the Planning Division. The Joint Committee would be receiving reports on progress as the strategy group would have an impact on health services.

12 **DH/NHSE Review of Genetic Services**

A meeting was being held on 27<sup>th</sup> June to look at commissioning. Public Health representatives had been asked to nominate suitable participants. The aim was to achieve better mechanisms within existing structures by the Autumn and it was also hoped to reach agreement on longer term needs.

13 **Genetics knowledge/education of non-genetics professionals**

The report of Dr J Clayton-Smith's working group for the Joint Committee was discussed. We were advised that Ministers were keen that the Human Genetics Commission should look at genetics education for non-specialists, particularly at primary care level, and it was requested that the Joint Committee should share the document with the Commission.

14 **Role of the clinical geneticist**

The Clinical Genetics Society was undertaking consultation on this document which should be ready for presentation at the next meeting of the Joint Committee.

15 **Confidentiality and Consent in Medical Genetics Working Party**

It had become apparent that the best way to proceed would be to look at the situation from a practical perspective and to produce a consensus statement or working guidelines.

The working party would have wide representation and would include representatives from the Department of Health, the Vice-chairman of the Human Genetics Commission, and someone with experience of law on databases. Dr Douglas would chair the working party. The working group will identify the main issues and then seek the views of the Joint Committee. Genetics Units will be asked to provide information on their present practices. The Chairman had consulted documents produced by other Colleges and bodies on consent and confidentiality but still felt that the Joint Committee had a special role in offering an overview and practical guidelines as it represented the subspecialties actively providing genetic services.

16 **Royal College of Pathologists Guidelines for retention of tissues at Post Mortem**

Dr John Dean had contacted the Joint Committee to express concerns that genetics had not been mentioned specifically in this document. Mrs Fitchett would report back to the RCPATH Council that the Joint Committee asked that in a future revision of the guidelines consideration should be given to explicit mention of consent for storage of samples for genetic testing where appropriate.

([http://www.rcpath.org/news/tissue\\_retention.pdf](http://www.rcpath.org/news/tissue_retention.pdf);

[http://www.rcpath.org/news/patients\\_leaflet.pdf](http://www.rcpath.org/news/patients_leaflet.pdf))

17. **Matters from the Royal College of Physicians**

The Joint Committee's activities regarding the education of non-genetics professionals had been presented by the Chairman to the RCP Medical Specialties Board. The Board had expressed interest in this initiative and had requested a paper providing more detail

18 **Manpower and Training**

- (a) [A Health Service of all the talents: Developing the NHS workforce. Consultation document on the review of workforce planning](#)

Members discussed the Executive Summary but concluded that they did not wish the Joint Committee to respond to the Consultation document.

(full report available on website address: <http://www.doh.gov.uk/wfprconsult>)

(b) RCPATH SAC

The training programmes for cytogenetics had been revised and would be available on the RCPATH website.

(c) SWAG specialty review: clinical genetics

Professor Mueller advised that he was trying to ascertain the total number of geneticists who were funded by either the NHS or an academic institution, and whether they were full-time or part-time. He was also trying to assess the number of anticipated retirements and hoped that he would have clearer information by the time of the review by SWAG in June. Applying the SWAG formula would result in the loss of eight registrar posts, and some Chairs of Regional Speciality Committees had already been contacted to ask how this could be achieved. The Chairman commented that there was great concern about the proposed reduction in the number of trainees in view of the need to expand the number of consultants. Members agreed that training posts should be located where the best training was available. Professor Mueller commented that he had calculated 1.5 trainees per consultant were needed in view of the academic element.

(d) JCHMT SAC in clinical genetics

The SAC had recently revised the curriculum, but this was to be reviewed in accordance with a common formula from the JCHMT which would be more objective and competence based.

19 **Publications received:**

(a) Therapeutic cloning: submission to the Chief Medical Officer's Expert Group (<http://www.royalsoc.ac.uk/files/statfiles/document-104.pdf>)

(b) Advisory Committee on Genetic Testing Report for Consultation: Human Genetics Commission ([http://www.doh.gov.uk/pub/docs/doh/prenatal\\_gt.pdf](http://www.doh.gov.uk/pub/docs/doh/prenatal_gt.pdf))

(c) Training in Academic Medicine: recommendations from the Academic Medicine Committee of the Royal College of Physicians (March 2000)

20 United Kingdom Haemophilia Centre Directors' Genetics Working Party

Dr Fryer had attended a meeting of the Genetics Working Party as a representative of the Joint Committee, and learnt that many of the concerns were common to both specialties.

21 The Chairman thanked Ms Hilary Irons, the administrator of the Joint Committee, who was retiring. Ms Irons had been the administrator of the Clinical Genetics Committee of the Royal College of Physicians, and had been deeply involved in the arrangements for bringing the Joint Committee into being.

12 **Dates of Future Meetings**

Wednesday 27th September 2000