

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 Tuesday, 19 October 2004.

Present:

Dr Heather Skirton JCMG Chair, BSHG

Dr Stephen Abbs	-	RCPPath
Dr Mark Bale	-	Observer, DH
Dr Hilary Burton	-	Observer, PHGU
Dr Trevor Cole	-	RCP
Mrs Michelle Collyer	-	RCP, Patient and Carers Network
Dr John Crolla	-	RCPPath
Professor Peter Farndon	-	BSHG
Dr Alan E Fryer	-	RCP
Dr Anne Green	-	RCPPath
Professor Shirley Hodgson	-	BSHG
Dr Tessa Homfray	-	RCP
Ms Dianne Kennard	-	Observer, DH
Mr Alastair Kent	-	GIG/Patient and Carers Network
Dr Helen Kingston	-	RCP, JCHMT SAC Chair
Dr Ruth Newbury-Ecob	-	RCPCH
Dr Tony Parkin	-	BSHG (<i>part meeting</i>)
Professor Julian Sampson	-	Chairman, BSHG
Ms Su Stenhouse	-	BSHG
Mrs Pat Ward	-	NSC (<i>in attendance, part meeting</i>)
Dr Virginia Warren	-	FPH
Mr Simon Land	-	Committee administrator

1. Apologies for absence

- Apologies were received from Dr Hayley Archer (RCP, Trainee representative), Professor Carol Black (RCP, President), Dr Sally Davies (Observer, Wales and Workforce), Professor Ian Gilmore (RCP, Registrar), Dr Hilary Harris (RCGP), Dr Sian Morgan (RCPPath Trainee representative), Dr Maggie Williams (RCPPath, Trainee representative), Professor Peter Soothill (RCOG) and Dr Allison Streetly (NSC).

2. Membership

- The Chairman formally welcomed Dr Bale and Mrs Collyer to the committee. Members noted that Dr Harris and Dr Streetly were also new representatives but had sent apologies for the meeting.
- The current membership of the committee was noted.
- The Chairman asked whether the composition of the committee was still relevant to the current climate. Members suggested the following representation would be useful:

Professional representation from the devolved countries. It was agreed that where possible the parent bodies would be encouraged to include appropriate representation from devolved countries within their nominations. However, when this was not possible, observers would be invited. It was agreed that the Chairman would write to John Dean of the Scottish Clinical Consortium inviting him to nominate an observer.

ACTION: *Chairman*

Policy makers from the devolved countries. Some members felt that it would be ideal to have policy makers from Wales, Scotland and Northern Ireland as observers to reflect the differences and help aid U.K. cohesion within genetics.

Paediatric representation. Dr Newbury-Ecob suggested that a paediatrician who is not working in specialist genetics would provide a wider perspective on behalf of the members of the RCPCH and that Dr Sian Snelling might be approached to replace herself at a future point.

British Inherited Metabolic Diseases Group (BIMDG). Dr Green felt that a member of this group should be invited to observe the committee.

- It was agreed that Dr Fryer and Dr Abbs would now step-down from the committee after 4 years of service. The Chairman thanked them for their contribution. It was agreed that Dr Fryer would act as the JCMG liaison person at the working party on UK Haemophilia Centres and report back where necessary. The committee administrator would look to find replacements for both members via the RCP and RCPATH.

ACTION: *Dr Fryer/Committee administrator*

- The Chairman recorded that the administration and Chairmanship of the committee would pass to the RCPATH in 2005 and agreed to write to the Registrar, RCPATH to that effect.

ACTION: *Chairman*

3. To confirm and sign the Minutes for the meeting held on 20 May 2004

- After amendments the minutes of the meeting were confirmed and signed as a true record.

4. Matters arising on the Minutes

4.1 Copying of Laboratory Reports

- Dr Crolla tabled a document that had been sent to the BSHG newsletter to request that laboratories forward recent documented examples where copying of laboratory reports had lead to incorrect genetic information being forwarded to a clinician. This was in addition to laboratories amending their Standard Operating Procedures to comply with the new Clinical Pathology Accreditation (UK) Ltd (CPA) Standards.

4.2 Length of time for storage of DNA samples

- Members discussed the amount of time DNA samples should be stored. At present, the practice in U.K. laboratories varies from 'at least 10 years' to indefinitely (under RCPATH regulations this is taken to mean 30 years). It was previously noted that other European countries had set-time limits and Professor Farndon asked members for their thoughts on how/if this should be addressed.
- Members believed this was a professional practice issue and that it warranted action. The reasons for this included the fact that storage space will become increasingly pressured and many of the samples were not utilised in a productive way.
- Dr Crolla indicated that the RCPATH was currently revising its guidelines on pathology storage. This work was being co-ordinated by Professor Peter Furness but was currently on hold to await implications from the Human Tissue Bill.
- It was therefore agreed that the Chairman would write to Professor Furness indicating the committee had discussed this issue. The letter would highlight the need to include representation from both clinical and research fields when new guidelines were written.

ACTION: *Chairman*

- Members of the committee were asked to volunteer to join or nominate others (where appropriate) to the group. The following names were received:

John Crolla – RCPATH (already involved)
Tessa Homfray -RCP
Anne Green – Laboratory Networks
Julian Sampson – BSHG (to nominate an individual)
Hilary Burton – PHGU (to approach Paul Pharoah)

5. Committee Terms of Reference

- Members discussed and revised the Terms of Reference.

Clerks note: *The amended Terms of Reference are appended to these minutes at Appendix 1. Members are asked to review these and return comments via the committee administrator.*

6. Genetics Branch, Department of Health

6.1 White Paper

- Members noted the update on the White Paper commitments. As an amendment to this it was recorded that 21 of the 23 Genetic Counsellor Trainees had now been appointed and that the number of healthcare scientist training posts (currently 25) was changing.
- Ms Kennard stated appointment of the Chair for Pharmacogenetics was in progress and that 6-pharmacogenetic research projects had also been announced.

6.2 GPs with Special Interests (GPwSIs)

- Ms Kennard directed members to the GPwSIs website:
<http://www.gpws.org/subindex.shtml>
Applications for the posts have been received and she thanked the RCGP, JCMG, trainers and other supporters for their help on the issue.

7. Genetics Commissioning Advisory Group (GenCAG)

7.1 Update

- Ms Kennard reported on the following issues:

National Genetic Reference Laboratories and GenCAG had been informed that another 2 years funding had been secured for the reference laboratories following the mid-term review.

A report has been forwarded to the Advisory Group on genetic research regarding accountability of the Knowledge Parks and Reference Laboratories.

GenCAG had issued a series of Quality Markers and was now surveying Genetic Centres. Mr Kent informed members that the equity of access question within the markers had now been expanded to cover ethnicity. GIG would contact all the regional centers to co-ordinate this data.

GenCAG had previously reported to the National Specialist Commissioning Advisory Group (NSCAG) but now was a stand-alone body. It would continue to report informally to the Specialist Commissioners. GenCAGs purpose would be to advise DH, Ministers, PCTs and the CMO.

7.2 UK Genetic Testing Network

- Professor Farndon stated that the UKGTN had been set up in 2002 with the aim of producing funding stability. However, it has not been possible to achieve this aim satisfactorily because currently health service funding is provided by PCTs and therefore every genetic centre has a multitude of different funding strands. The following points were also raised:

UKGTN had invented a system to validate and introduce new tests, which protected against funding vagaries. Other countries had now shown interest in this system.

The steering group had looked at the Haemophilia report and had asked GenCAG to consider the recommendations of the report with regard to funding.

Gene Dossiers had been placed before commissioners for funding. GenCAG had agreed these Dossiers and recommended that individual commissioners and providers negotiate funds locally. Workshops had also been run on the completion of Gene Dossiers

A 2nd cohort of laboratories was to join UKGTN. A non-NHS provider had also made an approach.

UKGTN had discussed the forthcoming National Tariff. The Clinical Molecular Genetics Society had received 6 volunteer labs for the process, which had been chosen by consistent use of Workload Units.

The remit of UKGTN did not include cytogenetics, but this was being revisited as the issues apply equally to new cytogenetic tests.

8. Appropriate Services for adults with inherited metabolic diseases

- Dr Burton thanked the DH for agreeing to fund the meeting scheduled for 10th November 2004. Representatives from 4 centres, laboratories, nurses, dieticians, Commissioners, GIG and the Workforce Numbers Advisory Board had all agreed to attend.

- The plan was to review the current service by looking at the structure, laboratories and voluntary organizations it entailed. A report would then be produced by April 2005.
- The Chairman thanked Dr Burton for organising this group and members were optimistic for progress due to the backing it had received.

9. Human Tissue Bill

- Members noted the letter to Lord Warner from the Chairman on behalf of the committee regarding the “Legal means to allow the use of bodily material (including the extraction and testing of DNA) where consent has been expressly refused”.
- Members noted the letter to Lord Clement Jones from Dr Crolla on behalf of the British Society for Human Genetics (BSHG) regarding “the use of tissue in genetics laboratories within the context of training”.
- The help of Ron Zimmern, Alison Hall and team was acknowledged, as they had been extremely active regarding the Bill and subsequent lobbying.
- Dr Crolla reported that many of the concerns of pathology had now been addressed and that the Bill had been extended to include embryologists. He remained hopeful of good news when the Bill returned from the report stage on 25th October 2004. Mr Kent echoed the comments of Dr Crolla on behalf of GIG.
- Dr Bale stated that consideration of the points made in the Chairman’s letter to Lord Warner had occurred. He believed that the focus should now be aimed at practical issues of implementation by looking at current practice. He stated that this, along with the establishment of Codes of Practice, would make the Bill workable. The genetics community would also need to devise a protocol for dealing with existing samples, which will be exempt from the Bill.
- Dr Bale updated members on the formation of the Human Tissue Authority (which would act as licensing authority for activities involving the removal, storage, use and disposal of human material as well as advising Government on issues related to human tissue). He stated that it would not become fully operational until a merger with the Human Fertilisation and Embryology Authority (HFEA) had occurred, to form the Regulatory Authority for Fertility and Tissue (RAFT). It was envisaged that this would not occur until 2005, but that shadow groups were already meeting to look at implementation. He agreed to forward more details on their progress to members via the committee administrator.

ACTION: *Dr Bale*

- Professor Farndon asked members whether the Consent and Confidentiality report should now be published. Members agreed that this should occur with the proviso that the arrangements under Common Law for samples where no consent was held (pre-dating the HTB) must be revisited by a legal expert. It was agreed that Professor Farndon would contact Dr Graham Laurie (School of Law, University of Edinburgh) to look at this and then circulate the document once more to members for comment. It was hoped that early publication would enable the report to be available to inform future Codes of Practice.

ACTION: Professor Farndon

- After a question on the possible prosecution of health professionals under the Bill, Dr Bale stated that the Bill had been cast in such a way that only those with deliberate knowledge of ‘no consent’ would face prosecution. It was not aimed at those who had received samples in good faith.
- Dr Cole asked whether “the ranking of consent was still in place under the Bill”. Dr Bale stated that it was recognized that exceptions existed within the Bill, of which this was one. This, together with other anomalies, would be covered by a published Code of Practice by the HTA.

10. Mental Incapacity Bill

- The Chairman reported that a synopsis of the Bill had been forwarded to members 5 weeks previously asking for comments. It was noted that only a few responses had been received and the Chairman asked how much concern this Bill caused members.
- Members discussed Section 33 – Safeguards of the Bill which stated that:

Nothing may be done:
 - if P objects (by showing signs of resistance or otherwise)
 - which is contrary to an advance directive
 - if P indicates that he wishes to be withdrawn from the project
- Professor Sampson felt that most severely handicapped people would reject being tested on reflex, but felt that this temporary discomfort was more than balanced by the research into diseases that would have a future benefit for those affected by the condition. Mr Kent stated that GIG had raised this issue previously as it caused concern to family members. GIG believed that incapacitated patients should not be assumed to be unable to act altruistically in the way that other family members often do. It was noted that carers and family have no legal rights on this issue (i.e. cannot gain Power of Attorney over a mentally ill adult). Professor Sampson argued that there would be politicians who have mentally ill relatives who would likely support an amendment and agreed to research this with a view to making contact.

ACTION: Professor Sampson

- Dr Bale agreed to take the issue back to the DH for review of the language used which members felt created a conflict between best interest and research.

ACTION: Dr Bale

- It was also agreed that members would e-mail other points to the Chairman and think of ways to increase communications that research is a good thing (to counteract negative image post Alder Hey etc).

ACTION: All

11. Reports of the work of the JCMG in progress

11.1 Consent and Confidentiality report

- The issue was covered under Item 9.

11.2 Training Posts for genetic laboratory scientists

- Members noted the report prepared by Dr Abbs.
- Feedback from last year was positive overall, barring reports of poor IT access from some. The course was due to begin again in November 2004 and at present 6 trainees had signed up to this.

11.3 UK Haemophilia Centres Genetics Working Party

- A summary paper prepared by Dr Fryer was noted.
- Dr Fryer asked for suggestions on how to push forward the issues raised within the report. It was agreed that members would e-mail Dr Fryer with suggestions.

ACTION: All

11.4 Genetic Counsellor Training Post Panel

- The issue was covered under Item 6.1.

12. Presentation on the Antenatal Screening Programme

- The committee received a presentation from Mrs. Pat Ward, Programme Director for Down's syndrome screening and Fetal Anomaly Ultrasound Screening to the National Screening Committee.

- The Chairman thanked Mrs Ward for a very informative presentation and it was agreed that if absent members wished to see a copy of the slides then they should contact the committee administrator.

ACTION: All

13. Enzyme Replacement Therapy

- Mr Kent reported that he was sitting on the NICE feasibility study on ultra-orphan drugs (Gauchers disease). Chris McKay in Sheffield was leading on the issue and a model was to be constructed by 2nd March 2005. The initial meeting had included commissioners, treatment centres, drug manufacturers and patients. It was agreed that members should e-mail ideas to Chris McKay on factors to be included within the model. It was felt that if this model were correctly constructed then it would be easier to deal with other products that would occur in the future.

ACTION: All

- Some members felt that the model for Gauchers disease would not fit all and that the JCMG should anticipate this by forming a small group to look at the issues of expensive drug therapies. A group to be headed by Mr Kent and to include Dr Burton, Dr Newbury-Ecob and Dr Warren would therefore focus on this.

ACTION: Mr Kent, Dr Burton, Dr Newbury-Ecob and Dr Warren

- Mr Kent also agreed to forward the offer of PHGU and the Scottish Medicine Consortiums help to Chris McKay regarding the model for Gauchers disease.

ACTION: Mr Kent

14. NICE

- Members noted that the Familial Breast Cancer Guidelines had been published in June 2004.
- It was noted that full screening for BRCA1 and BRCA2 was now the Gold Standard and that UKGTN had been given a year to clear the backlog. As steering groups had said this was unlikely to be feasible, tests on individuals who had at least 50% risk of mutation would be undertaken by June 2005. This would be revisited in 6 months time for assessment on progress.
- It was also noted that fewer mammograms for the under 40s for those with moderate risk would be undertaken under the guidelines and that access to psychological support should be universal.

- Dr Cole felt that certain points within the guidelines would cause problems in certain areas. Dr Warren reminded members that NICE review documents on a regular basis and that it would be worth keeping records. It was therefore agreed that Professor Hodgson would forward the issue to the Cancer Genetics Group.

ACTION: Professor Hodgson

15. Educational Issues

15.1 GENE

- Professor Farndon reported that the curriculum for the learning needs of SpRs in non-genetic subjects was being developed to feed into the National Genetics Education and Development Centre <http://www.geneticseducation.nhs.uk/>
- It was agreed that the NHS Genetics Education and Development Centre would become a standing agenda item from the next meeting. Professor Farndon would compile a report on this for that meeting

ACTION: Professor Farndon/Committee administrator

15.2 Multi-disciplinary training opportunities panel

- Members noted the report of the first meeting, which discussed the overlap of educational provision. A list of educational resources to be shared between groups is being compiled. The Chairman stated that the next step would be to develop further resources, predominantly on-line.
- 16. National Genetic Reference Laboratories
- Members noted the tabled reports from the Manchester and Wessex National Genetic Reference Laboratories. If members had questions on this they were invited to contact Nick Cross (Wessex) or Rob Elles (Manchester).

ACTION: All

17. Workforce and Training

17.1 RCPATH SAC

- Members noted the Chairman's report on the progress of JCMG to the RCPATH.
- Dr Crolla reported that the SAC had been extended to incorporate Clinical Embryology. This would be the case until Clinical Embryology set up its own SAC.

- Medical Biochemistry had now been recognized as a specialty and a member was now on the SAC.
- Dr John Old had finished his term as Chief Examiner and would now be replaced by Dr Teresa Davies. Dr Crolla had thanked Dr Old for his dedication and good work.

17.2 JCHMT SAC in Clinical Genetics

- Dr Kingston stated that implementation of the Postgraduate Medical Education and Training Board had been delayed and that the Chairman had recently resigned. The greatest impacts of this were that NCCGs who wished to become consultants under Article 14 (experiential assessment) had been delayed and that JCHMT would continue to advise the Specialist Training Authority with regard to joining the specialist register.
- Dr Kingston also updated members on the progress of Modernising Medical Careers. It was still uncertain how this would impact on training and there were concerns that trainees would be asked choose their specialty at the start of Foundation Year 2.

17.3 Workforce in Clinical Genetics

- 16 trainees would be completing training in 2006.

18. Sudden Unexpected Death in Infancy (SUDI) report

- Members noted the report published jointly between the RCPATH and RCPCH.

19. RCP Business

19.1 Consultant Physicians Working with Patients

- Members noted that this document would be published shortly.

19.2 RCP Lectureships and Conferences 2006

- At the BSHG Strategic Planning meeting, educational initiatives with other professional bodies had been suggested. Following suggestions for conferences and lectures from JCMG members, potential programmes were being informed by the BSHG Scientific program committee and will be forwarded to RCP.

20. Any other business

20.1 Clinical excellence awards 2005

- Professor Farndon had written as Chair of the CGS to one member of each of the genetic units in England and Wales inviting nominations. A full listing had then been compiled and forwarded to the President of RCP.

20.2 Electronic Patient Record

- Concern was expressed as to this would impact on genetic departments. Members were unhappy if genetic records would be accessible by GPs and the general public. It was agreed that the Chairman would write to Rob Elles for a report as to the current status.

ACTION: *Chairman*

21. Dates of the next meetings

- Thursday 13th January 2005
- Tuesday 10th May 2005
- Thursday 20th October 2005

Joint Committee on Medical Genetics - Revised October 2004

Terms of Reference:

- 1 To promote and maintain the highest standards of practice and education in both clinical and laboratory applications of genetics in health.
- 2 To discuss and co-ordinate advice to Government and other bodies on policy and services issues relating to genetics in medicine.
- 3 To report and receive information from the parent bodies and other representative groups.
- 4 To co-ordinate advice on workforce planning.
- 5 To initiate working groups on specific topics of particular importance. This would be subject to agreement by the parent bodies when significant expenditure was envisaged but the Joint Committee would be free to authorise small working groups.
- 6 To assist parent bodies in the development of their academic and educational activities.
- 7 To provide a unified forum for these activities.

Membership of the Committee:

The working group agreed that there should be five nominees from each of the parent bodies to facilitate the inclusion of the interests of chemical pathology and medical oncology, the RCPPath to provide chemical pathology representation and the RCP to provide medical oncology representation.

- 3 President/Registrars of RCP & RCPPath, Chairman/Deputy of BSGH
- 5 nominees of RCP (including chair of JCHMT SAC and one trainee)
- 5 nominees of RCPPath (including chair of SAC and one trainee)
- 5 nominees of BSGH (one from each of the four constituent groups)
- 2 representatives from the Patient and Carer Network (1 to be from the Genetics Interest Group)
- 1 representative from RCPCH
- 1 representative from Scottish Royal Colleges of Physicians
- 1 representative from Faculty of Public Health
- 1 representative from RCGP
- 1 representative from RCOG
- 1 observer from Department of Health
- 1 observer from Wales
- 1 observer from PHGU
- 1 observer from NSC

n.b. 1 of the above members would also be required to cover Workforce issues

Practical Issues:

- 1 Membership: For parent bodies the term of office is to be for 4 years, subject to review.
- 2 Chairmanship: term of office to be for a maximum period of 3 years, to rotate between the three parent bodies.
- 3 Secretarial support and meeting location to be provided by the parent body of the incumbent Chairman.
- 4 Travel costs of meetings to be paid by the parent bodies, including representative bodies, of all members.
- 5 Expenses of working groups to be discussed and authorisation obtained from the parent bodies when significant expenditure involved.