

# *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 Tuesday, 8<sup>th</sup> November 2005.

Present:

Dr Heather Skirton JCMG Chair, (BSHG)

Dr Rodney Burnham	-	RCP, Registrar
Dr Hilary Burton	-	Observer, PHGU ( <i>part meeting</i> )
Dr Trevor Cole	-	RCP
Dr John Crolla	-	RCPATH
Dr Teresa Davies	-	RCPATH
Dr Anne Green	-	RCPATH
Dr Hilary Harris	-	RCGP
Dr Shirley Hodgson	-	BSHG
Dr Tessa Homfray	-	RCP
Ms Dianne Kennard	-	Observer, DH ( <i>part meeting</i> )
Mr Alastair Kent	-	GIG/RCP Patient and Carer Network ( <i>part meeting</i> )
Mr Jawade Liquat	-	In attendance
Dr Ruth Newbury-Ecob	-	RCPCH
Dr Gail Norbury	-	RCPATH
Dr Tony	-	BSHG
Professor Peter Soothill	-	RCOG
Ms Su Stenhouse	-	BSHG
Dr Fiona Stewart	-	RCP
Dr Karen Temple	-	BSHG
Professor Richard Trembath	-	BSHG, Chairman
Dr Virginia Warren	-	FPH
Mr Simon Land	-	Committee administrator

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1. Apologies for absence

- Apologies were received from Dr Hayley Archer (RCP, Trainee representative), Dr Mark Bale (Observer, DH), Professor Carol Black (RCP, President), Mrs Michelle Collyer (RCP, Patient and Carers Network), Dr Sally Davies (Observer, Wales and Workforce), Dr Sian Morgan (RCPATH Trainee representative), Dr Allison Streetly (NSC, Observer) and Dr Helen Williams (RCPATH, Registrar).

## 2. Membership

- The Chairman welcomed Dr Gail Norbury (replacing Dr Abbs and representing the RCPATH), Dr Fiona Stewart (replacing Dr Fryer and representing the RCP), and Professor Richard Trembath (replacing Professor Sampson as Chairman of the BSHG) to the committee.
- Members welcomed Mr Jawade Liquat (RCPATH administrator) who would act as secretary to the committee from 1<sup>st</sup> January 2006 when the administration passed to the RCPATH.

## 3. To confirm and sign the Minutes for the meeting held on 17<sup>th</sup> May 2005

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

## 4. Matters arising on the Minutes

- All items were covered by the existing agenda.

## 5. Genetics Branch, Department of Health

### **5.1 White Paper**

- Members noted the pre-circulated and tabled update paper **DOC 05/19**.
- Ms Kennard reported with regard to Workforce issues that Skills for Health had just advertised a genetics post. Dr Elston asked if a job description was available and Ms Kennard agreed to investigate this.

***ACTION: Ms Kennard***

### **5.2 Review of commissioning for specialised services**

- Members noted the Press Release **DOC 05/20** regarding the formation of a Taskforce to undertake a review of commissioning for specialised services.
- Ms Kennard stated that its purpose was to strengthen existing arrangements and that massive change to the system was not envisaged. Dr Burton asked whether there was scope to discuss what was classed as a specialised service. Ms Kennard replied that definitions for specialised services existed on the DH website and that the review was likely to focus on more strategic issues.
- Professor Soothill asked how any new structure might affect tariff settings with reference to Payment by Results. Ms Kennard stated that specialised services were currently being kept outside the Payment by Results system.

## 6. Expensive Drug Therapies

- Mr Kent stated that the NICE feasibility study on ultra-orphan conditions (using Gauchers Disease) had been submitted to Ministers in July 2005. It advocated that existing NICE techniques could be used to assess expensive drug therapies, although the methodology would need to be tested to ensure sensible results. Mr Kent agreed to report back once a Ministerial response had been made.

***ACTION: Mr Kent***

- The committee discussed two recent articles from the BMJ (McCabe and Shah), which members believed would be detrimental to services for patients with rare disorders. It was noted that neither article was based on NICE policy and if allowed to stand unchallenged could be used by commissioners as justification that treatment for rare disorders would be too expensive to fund.
- Mr Kent reported that GIG had compiled a response to the articles (in conjunction with the Gauchers Association and the MSS), which would be forwarded to the BMJ. He hoped that the JCMG would support the letter and it was agreed to circulate this to Dr Stewart, Dr Newbury-Ecob and Dr Green for comment.

***ACTION: Mr Kent/Committee administrator***

- It was agreed that the same individuals would also draft a response on behalf of the JCMG. Members agreed that this was an important topic and that the letter should reference Dr Burton's recent work on IMDs. The RCP Registrar requested that the letter was copied to his Office (which had a lot of contact with NICE). He also recommended that Jim Barnet (Chair of the NICE Approvals Committee) could be approached informally to highlight the matter.

***ACTION: Dr Stewart/Dr Newbury-Ecob/Dr Green***

## 7. National Genetic Reference Laboratories

- Members noted the updates from the Wessex and Manchester Reference Laboratories **DOC 05/21**.
- Dr Crolla reported that the main concern of the laboratories was their future, which was currently being considered by the DH. At present the laboratories were secure until the current funding period ended in March 2007. However, comments from a stakeholder meeting had generally been positive with a consensus that they should continue. Ms Kennard stated that a meeting between the DH and

Reference Laboratories was due to be held the following week. She agreed to feed back any decision on their future in due course.

**ACTION: Ms Kennard**

## 8. Genetics Commissioning Advisory Group (GenCAG)

### **8.1 Update**

- Members noted the update on Quality Markers within **DOC 05/19**.

### **8.2 UK Genetic Testing Network**

- Members noted the update report **DOC 05/22** and a tabled paper on UKGTN.
- Dr Temple stated that UKGTN was now beginning to establish itself with the aid of a useful website and invited questions from members.
- Dr Norbury asked for clarification on the quality standards for testing. Dr Temple responded that not all NSCAG tests were listed on the UKGTN website and that many tests which went through in the first round of Gene Dossiers did so under less strict guidelines. Dr Temple recommended that queries regarding this were directed towards NSCAG while Dr Stewart stated that Maureen Boxer was progressing work looking at the cross over between NSCAG and UKGTN.
- Dr Stewart stated that the submission of Gene Dossiers no longer required the signature of a Trust Chief Executive but could now be replaced by that of a Director of Services. Members agreed that this was a sensible alteration to procedure.

## 9. Human Tissue Authority

- Dr Crolla reported that the RCPATH had formed a Working Party (Chaired by Peter Furness) to collate comments on the HTA consultation. It had collated over 20 pages of comments which highlighted the following problems with the proposals: Inconsistencies in terminology  
Confusion as to how the HTA would interpret the Act.
- It was noted that the HTA Guidelines were due to be published in April 2006 and that the Act would come into force 3 months after this date. The committee resolved to await the publication of the Guidelines and hoped that they would be translatable to practice.
- It was noted that RCPATH Council had discussed options on who would be nominated to hold the licence for activities under the Act. It had been agreed that if this was devolved to the Trust CEO that the Trust could be at risk if a breach occurred. Therefore, the favoured approach would be to devolve the responsibility

to the appropriate professional, which although complicated should still be workable.

- Dr Crolla stated that some Trusts were anticipating the Act and becoming over zealous in refusing to share materials. He urged Trusts to wait and see what the Guidelines would actually recommend.
- Mr Kent stated that GIG had produced a response to the consultation and agreed to forward this to Dr Crolla. It was noted that GIG was concerned that it was not clear whether the HTA would refuse licences on a purely ethical or technical basis.

**ACTION: *Mr Kent***

## 10. Educational issues

### **10.1 Multi-disciplinary training opportunities panel**

- The Chairman stated that an educational CD on ethical issues was being developed. It was hoped that this would be available in January 2006.

### **10.2 NHS Genetics Education and Development Centre**

- Members noted the report from the Centre **DOC 05/23**.
- Dr Temple asked whether the Centre would subsume the work of the multi-disciplinary training opportunities panel. The Chairman stated that this was not the case as the Centre focused on the non-specialist while the Panel focused on the specialist. Dr Temple felt that this was not common knowledge and that both initiatives should be highlighted within the BSHG newsletter. Members agreed that this was a good idea and that Professor Trembath should progress.

**ACTION: *Professor Trembath***

## 11. Reports of the work of the JCMG in progress

### **11.1 UK Haemophilia Centres Genetics Working Party**

- Members noted the update provided by Dr Fryer **Doc 05/24**.
- The Chairman stated that work was ongoing which was dedicated to the electronic record of family trees aimed at expediting family contact.
- Dr Temple reported that a meeting organised by NoWGEN in June 2005 had discussed the use of I.T. in genetics. Concern had been expressed that Pedigree Drawing within the Clinical Genetics Record would be problematic unless the system included the full family tree. It also highlighted that Connecting for Health

had not considered that genetics and haemophilia centres often wanted access to records of individuals who were not under the care of the a different practitioner. Under current proposals only the doctor and patient could access records, as geneticists were not considered primary carers. It was agreed that the Chairman would write to Connecting for Health to flag this issue.

**ACTION: Chairman**

- It was agreed that the issue would be an agenda item at the next meeting and Dr Temple would provide a background paper.

**ACTION: Dr Temple**

- Professor Soothill referred to the following section from the tabled paper stating that it was difficult for Blood Transfusion Centres to offer genetic tests. Dr Newbury-Ecob related that recent evidence suggested that DNA is not reliably present until 10 weeks and that testing.

***Fetal DNA in maternal circulation** – discussion took place about the use of this for fetal sexing prior to CVS. No methods of mutation detection in an X-linked disorder seemed feasible at present. Further discussion to take place about having a survey of what counselling was/had been given to female carriers presenting in pregnancy.*

## **11.2 Consent and Confidentiality Working Party**

- The Chairman stated that it was anticipated that the report would be published in the New Year. Members agreed that early publication was required to set the scene and influence the Human Tissue Authority. It was noted that a revision of the report would be undertaken two years after publication.

## **11.3 Training Posts for genetic laboratory scientists**

- Members noted the report **Doc 05/24**
- Dr Crolla asked whether the ACC or CGS were looking at the funding issues for higher specialist training with regard to clinical scientists who aspired to join the RCPATH. Ms Stenhouse stated that this formed part of the remit for national and regional trainers (who would also look at the training of MTOs). It was agreed to keep this item on future agendas and look to ways of dealing with the problem.

**ACTION: Committee administrator**

- Ms Kennard stated that the DH appreciated the problem and had funded some HST trainees within metabolic biochemistry to try to aid the particular shortage within that discipline. Dr Crolla applauded this but felt that the lack of funding

affected the training of all clinical scientists (not just those within Metabolic Biochemistry).

- Ms Kennard reported that a board meeting regarding the modernisation of pre-registration training within healthcare science disciplines to be taken forward by Skills for Health was due to be held the next day. As yet no details on delivery were available but Ms Kennard agreed to update members when more information was forthcoming.

#### **11.4 Genetic Counsellor Training Post Panel**

- The Chairman stated that 21 posts had been approved in the second round. From the first tranche 11 of the 23 individuals had completed 2 years training and 3 had located full-time posts. A watching brief was being kept on the issue.

#### **11.5 Metabolic Biochemistry (Biochemical Genetics) Network.**

- Members received an update paper **DOC 05/26** and a presentation from Dr Green on the Network.
- It was noted that Metabolic Biochemistry was a sub modality registerable via the Health Professions Council. It had also gained recognition via the CPA as a sub-specialty and therefore was entitled to specialist assessors.
- At the time of the meeting there was lead trainer, 3 local trainers and 11 trainees in within the specialty. Dr Green stated that the next important step was to entice new trainees to the specialty.
- The Chairman thanked Dr Green on behalf of the committee for her hard work. Members were heartened and impressed that the specialty had progressed so far in such a short period of time.
- Dr Green outlined to the committee the various ways in which the project might evolve:

Use as a central hub for dissemination of guidelines

Linkage to Europe – which could help with funding and communication

Exert influence regarding Workforce issues

Review service issues regarding rare tests. It was noted that UKGTN took the view that rare tests need only occur in one laboratory but the committee felt that this service was often dependent on one individual

- It was agreed that a reciprocal link would be added from the BSHG website to the Network webpage.

***ACTION: Professor Trembath***

- The committee recommended that Dr Green should approach GenCAG with a view to presenting the progress at a future meeting. Ms Kennard did not feel that Gen CAG members would be able to help with funding issues but may be able to offer other useful advice.

**ACTION: Dr Green**

12. Appropriate services for adults with inherited metabolic diseases

- Members noted the completed report (tabled): Metabolic Pathways ‘Networks of Care’.
- Dr Burton gave a presentation highlighting the following areas:

Thanks to the JCMG for recommending the formation of a sub-group to progress the work

Thanks to the DH for funding the meetings

Thanks to the PHGU, which had provided the administrative support

The recommendations were agreed at the BIMDG Conference in July 2005

Very few clinical teams (which should include dieticians, nurse, laboratories and clinicians) existed.

The project required further funding. An input of £7 Million would effectively double the existing service.

- The Chairman congratulated Dr Burton and her team on producing a superb report. Members felt that it provided a powerful case for the need to increase funding with regard to IMDs and that it would be interesting to see how commissioners received it. They were especially impressed by the use of case studies within the report to highlight common problems.
- Dr Harris raised the issue of coding used by GPs for each diagnosis. At present this did not include categories for IMDs, which resulted in occurrences being recorded under ‘general paediatrics’ and hence being ‘lost to the system’.
- The Chairman noted that the report recommended the completion of a Masters degree for nurses dealing with IMDs but thought that this would be hard to establish within higher education institutes. Dr Burton stated that she envisaged that it these would form a part of a Masters on ‘paediatrics’ and therefore could be slotted into existing courses. The Chairman recommended that the Royal College of Nursing should be encouraged to lend its support.

**ACTION: Dr Burton/Chairman**

- Members believed that PCTs would be keen to implement the recommendations within the report, as screening would eventually reduce cost to the service. It was therefore agreed that the committee would support the report recommendations in principle. A more detailed discussion and review of the full report would be

scheduled for the next meeting. In the meantime, the Chairman would forward a copy of the report to the Presidents of the Colleges. This would include a covering letter on behalf of the committee commending the recommendations.

**ACTION: *Chairman***

- Dr Burton and Dr Green would also progress a short piece on the project for inclusion within the BSHG newsletter.

**ACTION: *Dr Burton/Dr Green***

### 13. Workforce and training

#### **13.1 RCPATH SAC**

- Dr Crolla stated that the RCPATH was awaiting a recommendation from the NSC on whether FISH should replace karyotyping in pre-natal testing. Professor Soothill spoke as Chair of the Advisory Committee, which had considered this issue and stated that they would deliver a recommendation to the NSC by the end of November 2005.

#### **13.2 JCHMT SAC in Clinical Genetics**

#### **13.3 Workforce in Clinical Genetics**

- Members noted the following e-mail forwarded by Dr Davies with relevance to the above items:

“At the recent Workforce Review in September an overarching approach was taken considering the situation for clinicians, molecular, cytogenetic and biochemical laboratory staff as well as genetic counsellors. It was decided to not increase training numbers in clinical genetics for England. The modelling shows that with current numbers and the part time / research / academic issues of clinical genetics that the desired number of consultants required (4 per million including cancer genetics and new developments such as cardiac) should occur in about 2020.

However there has been much anxiety reflected by the trainees. They are concerned that some of the centres in England are suggesting that they cannot implement any new posts because of funding. We are OK at present as there have been enough jobs including locum consultant posts for those that are able/ willing to move. Jobs in the last 12 months have matched output. In fact there were some jobs not appointed e.g. Cardiff senior lecturer in Neurogenetics (now being advertised for the third time and a Birmingham cancer genetics post).

The main concern is for 2006-2007. There are 15 CCTs due in 2006 and 14 in 2007 (though some of these are currently pregnant / gone flexible / in research / looking for academic- research fellowships).

The aim of workforce planning is to provide enough trainees for the expansion of the specialty predicted even before the White Paper etc. Trainee numbers have been static for several years. We are also aware that recruitment to SpR genetics posts is currently low.

I alerted the Workforce Review Team that there was concern amongst trainees about the expected consultant expansion being held back by Trust issues. The SAC in Genetics are to discuss the issue on Monday 7<sup>th</sup> November 2005. The CGS council will also discuss it in their away day in February 2006.”

- The Registrar stated that it was down to each specialty to highlight problems at the Trust level and to commissioners.
- Dr Cole felt that Clinical Genetics needed to attract more people from non-paediatric backgrounds.
- Dr Newbury-Ecob suggested that statistics on the prevalence/population of specific diseases and the workload of genetic units were powerful tools with commissioners.
- The Registrar stated that Helen Stewart had recently represented the specialty at a workshop on Payment by Results at the College. Professor Trembath reported that the CGS had also discussed the issue at its away day.
- Professor Trembath raised the issue of academic training pathways and concerns that posts would be unable to be filled.

#### 14. Consultations and Correspondence

- The Chairman reported that it had proved difficult to get comments from members on the various consultations, which had been circulated. Members agreed to address the situation.

**ACTION: All**

- Members noted the following consultation responses:

Tissue engineering and beyond: Consultation on a proposal for a Regulation on advanced therapies **DOC 05/28**

Human Tissue Authority - Draft codes of practice for consultation **DOC 05/29**

Consultation paper on the ethics of prolonging life in fetuses and the newborn **DOC 05/30**

- It was noted that the following consultations were due a response and members were highlighted to formulate a response:

Storage of Newborn Babies Blood spot cards

**ACTION: Dr Newbury-Ecob/Dr Homfray/Dr Warren/Ms Stenhouse**

Review of Human Fertilisation and Embryology Act

**ACTION: Dr Newbury-Ecob/Dr Homfray/Professor Soothill**

- Professor Trembath agreed to add this consultation to the BSHG agenda.

**ACTION: Professor Trembath**

#### 15. Clinical Excellence Awards 2006

- Dr Temple reported that the CGS had called for submissions via their website with regard to CEAs. An e-mail to all consultants had also been disseminated via centre representatives. It was noted that the ranking of nominations was completed by 3 top-award winners and the CGS Executive.

#### 16. RCP Lectures and Conferences 2007

- Professor Trembath agreed to progress discussions at the BSHG regarding nominations for the 2007 RCP Lectureships and Conferences.

**ACTION: Professor Trembath**

#### 17. Patient and Carer Network Workshop – December 2005

- The Chairman asked for a volunteer to represent the committee at the workshop. Dr Newbury-Ecob agreed to attend and the administrator would forward the relevant details.

**ACTION: Dr Newbury-Ecob/Committee administrator**

#### 18. Any other business

##### **18.1 NHS Sickle Cell & Thalassaemia Screening Programme**

- Members noted a report **DOC 03/32** submitted by Dr Streetly. The Chairman drew the committee's attention to the Conference planned for 25<sup>th</sup> January 2006 at BMA House.
- Dr Newbury-Ecob stated that she had been co-opted to the Steering Group for the Screening Programme.

- Members noted with concern that certain PCTs were refusing to fund PIGD for sickle cell disorders. Dr Temple stated that PCTs make individual decisions based on individual circumstances and that Dr Streetly should take up the query with the PCTs concerned.

**ACTION: *Dr Streetly***

## **18.2 PPUK DMD Registry**

- Members noted the document **DOC05/33** regarding the set up of the UK Duchenne Muscular Dystrophy Registry. It planned to make a record of every boy with Duchenne in the UK so that when clinical trials became possible then the information to recruit to the trial would be readily available. Dr Stenhouse stated that she had brought this initiative to the attention of the JCMG so that clinicians and others in the field were primed if presented with the forms by patients. Members agreed that it was a sensible resource and that a presentation would be invited at the next meeting. Dr Stenhouse and the administrator were tasked with progressing an invite.

**ACTION: *Dr Stenhouse/Committee administrator***

## **18.3 Guideline Clearing House**

- Members noted a tabled document prepared by Dr Green, which proposed the formation of a national Guideline Clearing House aimed at preventing duplication of effort. Members noted that a similar system existed in the U.S. and agreed that the principle of the idea was sound. Dr Burton stated that a Guideline Library would only be interested in finished Guidelines whereas it would also be useful to have information on works on progress. The main issue was who would fund and handle any future library. It was agreed that the Chairman would write to the BSHG and Knowledge Parks for comment.

**ACTION: *Chairman***

## **18.4 Patient and Carer representation**

- The Chairman stated that Mrs Collyer had stood down from the committee as her family was due to emigrate to New Zealand. Members wished to record their thanks and good wishes for the future to Mrs Collyer and her family.

## **18.5 JCMG Chairmanship**

- The Registrar thanked the Chairman for handling the committee with consummate skill over the preceding 3 years and wished to record that the College appreciated all the hard work.

- Dr Crolla echoed the above stating that he was faced with the unenviable task of following the great achievements of the Chairman's term.
- The Chairman wished to record sincere thanks to all members of the committee and to the administrator for their support and good work during the last 3 years.

19. Dates of meetings in 2006\*

- Thursday, 19<sup>th</sup> January 2006
- Tuesday, 23<sup>rd</sup> May 2006
- Tuesday, 24<sup>th</sup> October 2006

\* It was noted that as of 1<sup>st</sup> January 2006 the administration of JCMG would revert to the RCPATH and that meetings will be held at 2 Carlton House Terrace.