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 Thursday, 13th January 2005.

Present:

Dr Heather Skirton JCMG Chair, (BSHG)

Dr Hayley Archer	-	RCP, Trainee representative *(part meeting)*
Mrs Sarah Burke	-	In attendance *(part meeting)*
Dr Hilary Burton	-	Observer, PHGU
Dr Trevor Cole	-	RCP
Dr Rob Elles	-	In attendance *(part meeting)*
Professor Peter Farndon	-	BSHG
Dr Anne Green	-	RCPath
Dr Tessa Homfray	-	RCP
Ms Dianne Kennard	-	Observer, DH
Dr Helen Kingston	-	RCP, JCHMT SAC Chair
Professor Christopher Ludlam	-	In attendance *(part meeting)*
Dr Tony Parkin	-	BSHG
Professor Julian Sampson	-	Chairman, BSHG
Ms Su Stenhouse	-	BSHG
Dr Allison Streetly	-	NSC, observer *(part meeting)*
Dr Virginia Warren	-	FPH
Mr Simon Land	-	Committee administrator

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1. Apologies for absence

- Apologies were received from Dr Mark Bale (Observer, DH), Professor Carol Black (RCP, President), Dr Rodney Burnham (RCP, Registrar), Mrs Michelle Collyer (RCP, Patient and Carers Network), Dr John Crolla (RCPath), Dr Sally Davies (Observer, Wales and Workforce), Dr Teresa Davies (RCPath), Dr Hilary Harris (RCGP), Professor Shirley Hodgson (BSHG), Mr Alastair Kent (GIG/Patient and Carers Network), Dr Sian Morgan (RCPath Trainee representative), Dr Ruth Newbury-Ecob (RCPCH), Professor Peter Soothill (RCOG) and Dr Helen Williams (RCPath, Registrar).

2. Membership

- Members noted that Dr Teresa Davies had replaced Dr John Old as RCPath representative.

- The administrator agreed to contact Dr Crolla regarding the progress of replacements for Dr Stephen Abbs and Dr Maggie Williams.

ACTION: ***Committee administrator***

- It was noted that the Scottish Colleges were considering the possibility of nominating an observer to join the committee.

3. To confirm and sign the Minutes for the meeting held on 19 October 2004

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

4. Matters arising on the Minutes

4.1 Length of time for storage of DNA samples

The Chairman agreed to write to Professor Furness reminding him of the offer of JCMG expertise to join the RCPath work on the revision of guidelines for pathology storage.

ACTION: ***Chairman***

4.2 Expensive Drug Therapies

It was agreed to defer this item to the next meeting when Mr Kent could update members. Professor Sampson stated that agreements on the availability of some drugs differed between Wales and England.. Members were concerned by this potential lack of parity and agreed to revisit this at the next meeting.

4.3 RCP Lectureships and Conferences 2006

The Chairman stated that three names and topics had been submitted to the RCP Academic Vice-President for consideration. A reply was awaited.

4.4 NICE guidelines on Familial Breast Cancer

The Committee noted an e-mail that had been sent from Professor Hodgson. It stated that CGC Steering Group was due to discuss the problem of monitoring the effects of the NICE breast cancer guidelines at their next meeting in June. The difficulty was how to monitor activities, which were not directly the remit of genetics centres (i.e. mammograms for moderate risk women). Dr Cole reported that many agencies were involved in this process and that great regional variances currently existed.

4.5 Terms of Reference

The Chairman noted that there was currently no mechanism to provide a deputy to the Chairman in case of unforeseen circumstances. Members believed this would be a useful addition to the Terms of Reference. It was agreed that retiring Chairs would remain on the committee in an ex-officio capacity for half of a new Chairman's term and that new Chairs would be recruited early to cover the latter half of the term. This would provide a deputy at all times and allow a good hand-over period between Chairs. As the RCPath were due to nominate the next Chairman (term beginning 1st January 2006) a letter would be sent detailing the need for an early appointment.

ACTION: ***Committee administrator***

5. Genetics Branch, Department of Health

5.1 White Paper

- Members noted the following e-mail update regarding the progress on implementation of commitments in the genetics White Paper. Ms Kennard added further comments at the meeting.

  Upgrading NHS genetic laboratories: PASA have now completed their procurement process for equipment and laboratories are now making purchases. Progress was good.

  Genetic Counsellor Trainees: 22 trainees are in post and the scheme is running well. Those who started earliest will complete their training during 2005.

  Healthcare scientist training posts:
  30 trainees are now in post (14 cytogenetic, 8 molecular and 8 metabolic biochemistry). The trainer posts, which are mainly part-time, have been appointed to or are being advertised. Each of the three disciplines is aiming to appoint a national-level trainer who can provide more strategic input and coordination, plus several locally based trainers.

  Genetics service development initiatives A list of the 10 funded projects, which covered a range of clinical areas, is on the DH website.

  Evaluation of service developments: An evaluation team, headed by Professor Robert Dingwall of Nottingham University, has been appointed to evaluate the familial hypercholesterolaemia and cancer pilots, service development initiatives and the development of GPs with a special interest in genetics. They are also providing some support to the individual projects and are hosting an event for internal evaluators on 13th January.

Familial cancer services: 4 pilots jointly funded by Macmillan Cancer Relief and DH are already underway. Three further sites that will specifically examine ways of boosting capacity & capability in primary care and will be led by PCTs, were announced over Christmas. These are Oldham, Poole and Somerset Coast.

Cascade screening for familial hypercholesterolaemia: Work is getting underway at the 5 sites selected to pilot cascade screening, and a separate research project to look at the place of DNA testing in FH is also starting. The programme is being managed by the London Genetic Knowledge Park with input from the Cambridge Genetic Knowledge Park.

GPs with a Special Interest in genetics: Final discussions are underway with candidates and host PCTs for 10 GPwSI posts - 8 individuals and two pairs of GPs - and we expect to confirm and announce these very shortly. Most are expected to start between now and April. Raising awareness of genetics and boosting capacity in primary care will be key elements of the role but undertaking their own programme of training and personal development will be a vital early task for these GPs. Some will focus on specific areas such as familial cancers or screening programmes, and others will cover a broader range of genetic topic areas.

Genetics Education and Development Centre: The Centre, led by Professor Peter Farndon at Birmingham Women's Healthcare Trust, has started work and made appointments. www.geneticseducation.nhs.uk .

Visiting Fellowships in genetics: 6 fellowships (4 outgoing and 2 incoming) were approved by the assessment panel in November. Topics covered include screening programmes, cancer, laboratory technologies and genetic test evaluation. The fellows will undertake their study visits at various times during 2005.

Ms Kennard expressed her thanks to JCMG members who had aided this.

Laboratory IT: The National Genetics Reference Laboratory (Manchester) has commissioned a short consultancy study on sample tracking and tracing. This has now been completed and will be considered by the UKGTN Steering Group. This suggests possible options for what might be needed in terms of sample tracking within and between laboratories. It will be used to decide where the £1 million allocated to tracking in the White Paper will be allocated. An Output Based Specification (for genetic services more generally) is also being commissioned by NGRL (Manchester).

Research programmes: Research projects within the pharmacogenetics and gene therapy programmes are now underway, and the recipients of the cystic fibrosis gene therapy research funding were announced in early January. For the health services research programme, an expert group met in November to explore possible topics, and these will be discussed and prioritised by a stakeholder workshop on 12th January. A call for outline proposals to expected to be issued by the end of February 2005.

The health services research programme had a budget of £1.5 million. Topics considered would not cover clinical effectiveness or specific clinical issues. They would more likely be around the following areas: equity of access, barriers to minorities, the role of family history taking and cost-effectiveness of genetic technologies in health services.

6. Appropriate services for adults with inherited metabolic diseases

- Dr Burton stated that the first stakeholder meeting had taken place at the Institute of Child Health in November 2004.
- First steps included a good description of the service need and an in-depth review of the existing service. This would include details of how the current service was commissioned, which would be progressed by Dr Simon Sanderson (SpR). Sensible suggestions would then be made as to how the current service could be improved and the expanded to meet the service need.
- The scope would focus on adult services, but include paediatric ones. It was confirmed that Scotland would be included in the survey and it was hoped that Northern Ireland would also take part. Information was now being collected by means of a questionnaire to existing centres. This would be followed by a further questionnaire to the laboratories dieticians and nurses to give a rounded perspective. It was hoped that responses would be returned by March 2005, therefore the preliminary results could be available to the next JCMG meeting in May 2005.
- Members felt that this work fitted well with previous work on Haemophilia and Neurology and was to be commended.
- Dr Green expressed concern at the current model of training for clinicians. SpR posts in metabolic medicine existed but there was no commissioning link-up for the allocation of consultant posts.
- Members noted that many adult outpatients were still seen in Children's Hospitals. They expected that paediatric teams would continue to act as a bridge until dedicated consultant positions occurred.

7. Genetics Commissioning Advisory Group (GenCAG)

7.1 Update

Ms Kennard reported that the next meeting was due in two weeks time. Therefore the item was deferred to the next meeting.

7.2 UK Genetic Testing Network

- Professor Farndon reported that the Steering Group had not meet since the last JCMG. However, he drew members attention to the UKGTN website which was now live
  http://www.genetictestingnetwork.org.uk/

- At present the content was fairly basic but in the near future it would have a specific ‘testing’ section. This would list all the clinically evaluated tests and locations at which they were available. There would also be a dedicated laboratory section.

- Ms Kennard stated that the remit and future of UKGTN was due to be reviewed and that there was a need to think ‘clinically’ about how to look at issues to aid this. A report from commissioners on the level and location of funding from the previous year was also imminent. It was agreed that Professor Farndon would update the committee on relevant issues after the Steering Group next met via e-mail.

ACTION: ***Professor Farndon***

8. Human Tissue Bill

- Members noted the summary of the Human Tissue Bill prepared by Alison Hall for inclusion in the BSHG newsletter, which had been circulated prior to the meeting. Further to this Professor Farndon had made changes to the Consent and Confidentiality report (see item 12.2).

9. Mental Capacity Bill

- Ms Kennard reported that little activity had occurred since the committee submitted comment and that the second report stage in the House of Lords took place on 4th January 2004.

- Professor Sampson detailed that the specific point raised at the last meeting on a patient exhibiting some objection to having a sample taken for research purposes had been considered by the BSHG. Following this, draft potential amendments had been forwarded to Baroness Finlay, which would be raised at a later date. It was agreed that the administrator would contact Dr Bale for an update, which would be circulated to members by e-mail.

ACTION: ***Committee administrator/ Dr Bale***

10. Electronic Patient Records

- The Chairman welcomed Dr Rob Elles who gave a presentation on the implications of the Electronic Patient Record to genetics. The Record was being implemented as part of the National Programme for I.T. (NPfIT) which was a multi-billion pound information infrastructure investment providing:

  –NHS Care Record Service: centralised medical records
  –Electronic booking and prescribing
  –Picture archiving and communication systems
  –A New NHS Network

- The genetics White Paper 'Our Inheritance, Our Future' was published in June 2003 which stated that 'the Department of Health will invest up to £1 million over three years for IT for genetics laboratories in the Genetic Testing Network. These new systems will be compatible with the Government's wider investment programme to modernise NHS IT systems and introduce electronic patient records'.

- Following the White Paper a review of IT systems in genetic services was commissioned. Its main conclusion was that an Output Based Specification was needed to capture the needs of genetics and NpfIT. This has now started in consultation with the genetics community and NPfIT. Members felt that the I.T. perspective should also include visits to Scotland and Wales. Dr Elles agreed to liaise with Simon McCusker from ITP Ltd who was running the project to progress this.

ACTION: *Dr Elles*

- Members felt that genetics was further advanced than NpfIT but that it would be vital to incorporate a suitable family history tool within the framework at the onset.

- The Chairman thanked Dr Elles for an informative presentation and asked the administrator to circulate copies to the committee by e-mail.

ACTION: Committee administrator

11. Educational issues

11.1 GENE

- The committee received a presentation by Mrs Sarah Burke from the University of Birmingham regarding GENE. The project mapped the current teaching of genetics to non-specialists and set up focus groups for perceived learning needs within 3 specialties (neurology, dermatology and cardiology) with a view to developing a genetics curriculum relevant to other specialties. Mrs Burke confirmed that copies of the final report would be forwarded to JCMG members when published.

- The Chairman thanked Mrs Burke for the presentation and asked the administrator to circulate the presentation to members.

ACTION: ***Committee administrator***

11.2 Multi-disciplinary training opportunities panel

- The Chairman reported that a meeting was due to take place later in the month to progress the issue.

11.3 NHS Genetics Education and Development Centre

- Professor Farndon tabled information, which gave an overview of the Centre, its main strands of work and its core team. For non-attending members information can be found at:
  http://www.geneticseducation.nhs.uk/about_ngedc.htm

12. Reports of the work of the JCMG in progress

12.1 UK Haemophilia Centres Genetics Working Party

- The Chairman welcomed Professor Christopher Ludlam (Working Party Chairman) who gave a presentation on the guidelines produced by the working party. Members were also supplied with a hard copy of the completed UKHCDO report 'Clinical Genetics Services for Haemophilia'. It was agreed that the administrator would circulate the presentation by e-mail and that non-attending members could request a copy of the publication from Simon.Land@rcplondon.ac.uk

ACTION: ***Committee administrator***

- Professor Ludlam thanked JCMG for the immensely valuable input of their members on the working part (Dr Fryer and the Chairman).

- Professor Farndon stated that the UKGTN supported the Guidelines and that they had also been presented at GENCAG.
- The Chairman thanked Professor Ludlam for co-ordinating a valuable piece of work and for presenting it to the committee.

12.2 Consent and Confidentiality Working Party

- Professor Farndon outlined the following points resultant from the HTB, which impacted on the report. He agreed to update the report, gain approval from the DH HTA advisors, convert the following instructions into an easy to use flow-chart and then return the draft to the administrator. This would then be circulated to members for a two-week consultation exercise.

ACTION: ***Professor Farndon/Committee administrator***

New Samples
Requests to take and use a sample for diagnosis or for a scheduled purpose (including for the benefit of another family member) – seek consent (The gaining of such "qualifying consent" is sufficient under the purposes of the Act to prevent an offence under the DNA regulations)
Samples stored after the Act comes into force
To determine scientific or medical information from cellular material from a living or deceased person which may be relevant to any other person (including a future person) – consent required If the person is alive, consent from that person. If the person is deceased, seek consent from the appointed representative or qualifying relatives. If the sample is extracted DNA it does not come under the legal requirements of consent of the Act, but it is good practice to ascertain if the donor left any previous wishes.
Samples held on the day before the Act comes into force
Existing holdings of cellular material are excluded from the provisions of the Act where these are to be analysed for Scheduled purposes (e.g. medical diagnosis) or for any purpose if the material is anonymised. Note that included in the scheduled purposes under this provision is the use for other members of the family.
Existing holdings of DNA (extracted from the human cells from which they derive prior to the Act coming into force) are excluded from the legal requirements of the Act regarding consent, as they do not constitute 'bodily material'. Neither does extracted nucleic acid come within the provisions of the offence of the nonconsensual use of DNA. The storage and use of this material, however, should remain within professional guidelines such as those outlined in this document.
Circumstances where a family member cannot be traced after the Act comes into force
Where it is in the interests of another person (even a future person) that the analysis of DNA (including karyotyping, FISH and PCR) in cellular material be undertaken to provide scientific or medical information about the donor, the Act provides that:
Where a donor cannot be traced and there is no reason to believe that he/she has died, has refused consent or is incompetent, then the analysis can be carried out following application to the Human Tissue Authority which may 'deem consent';
Where the donor can be traced, but consent/lack of consent is not forthcoming despite reasonable efforts to obtain it, and there is no reason to believe that he/she has died, has refused consent or is incompetent, then the analysis of the cellular

material can be carried out where the Human Tissue Authority 'deems consent', provided that the donor has been given notice of the application.
The use of extracted DNA in these circumstances falls under professional guidelines, such as those elsewhere in this report.
If cellular material is to be used in the diagnosis and treatment of its donor, it is exempt from the requirements of the Act.
Specific consent under the Act will be required to use cellular material to assist in the care of other relatives (but not extracted DNA whose use will fall within professional guidelines)
Consent under the Act is not required for material from the living to be analysed for certain purposes including clinical audit, education or training relating to human health, public health monitoring and quality assurance, (the purposes listed in Schedule 4 Para 8); When nucleic acid has been extracted from the cellular material, then the legal requirements of the Act regarding consent, storage and use no longer apply (although professional guidelines for its use should be followed).

The DNA offence

For analysis of DNA in cellular material (includes karyotyping, FISH and PCR on the cellular material) No offence is committed if consent has been obtained or the analysis is for medical diagnosis or treatment of the person whose body manufactured the DNA, or for other purposes (including the functions of a coroner and the prevention or detection of crime)
For analysis of DNA in cellular material in holdings before the Act comes into force
No offence is committed if consent has been obtained or if the analysis is for other scheduled purposes including clinical audit, education or training, obtaining information which may be relevant to any other person (including a future person), quality assurance and research (where the donor of the sample has been anonymised and is unlikely to be identified.)
For analysis of extracted DNA No offence is committed

- Miss Kennard Stated that the Human Tissue Authority was setting up in shadow form from March 2005 and that the report would act as an excellent steer on the issues.

- Members then discussed one of the recommendations of the report. Namely, the formation of a clinical ethics group, to offer advice. Some members felt that this had resulted from anxiety over the Bill, which had now lessened and believed that local workshops would be a less cumbersome way of informing individuals. There were also concerns that the group may be unwieldy and not be able to respond quickly to queries. Professor Sampson agreed to add this item to the BSHG agenda for discussion on what was practicable.

12.3 Training Posts for genetic laboratory scientists

- This item was deferred until a replacement for Dr Abbs could attend.

12.4 Genetic Counsellor Training Post Panel

- The Chairman reported that 22 of the 23 posts were now appointed. Thoughts now focused on the second cohort and a call for interest would be sent to Regional Genetic Centres in early February 2005. It was thought that even if some centres were not in a position to take part that others would replace them.

- An idea to establish other posts from different specialties had also been forwarded to the DH. Discussions were on-going regarding this.

<u>13. Access to records of the deceased for the purposes of genetic counselling</u>

- Members noted the letter received from the Cancer Genetics Group on the access to records of the deceased for the purposes of genetic counselling.

- Members felt that the concerns raised were those of 6 months past and that in practice the answers were tied to the clarification of the HTB and the finalisation of the Consent and Confidentiality report (already discussed). It was agreed that the Chairman would write back attaching the PDF file of the Consent and Confidentiality report, stating that the JCMG felt that the concerns were now answered within it.

ACTION: ***Chairman***

<u>14. Input into planning for antenatal screening programme for cystic fibrosis</u>

- JCMG had received a request for advice on antenatal screening for cystic fibrosis. Dr Streetly stated that lobbying for testing was on going and that comments would be welcomed. The NSC plan was to standardise newborn screening throughout England over the next 3 years. It was felt that ethically this had to be completed first to provide the support to an antenatal programme. The Chairman then asked for interested individuals who would be willing to join the antenatal discussions. The following nominations were noted:

  Dr Homfray
  Dr David Aitken (Scottish perspective)
  A welsh nominee
  An RCGP nominee. The JCMG observer (Dr Harris) would be approached regarding this.

- The Chairman agreed to write back with a list of names once they had been finalized.

ACTION: ***Chairman***

<u>15. National Genetic Reference Laboratories</u>

- The administrator agreed to contact Dr Crolla for an update on the Laboratories.

ACTION: ***Committee administrator***

- Dr Elles spoke as Head of Manchester NGRL on the following issues:

  The NGRLs were now at the mid-stage of the funding round. They were presently looking at programmes in place with a view to developing and shifting these to give the genetics community the best possible service. A stakeholder meeting was planned for June 20/21 2005 to discuss the progress of both the Manchester and Wessex laboratories, which had both been running for two years.

16. Workforce and training

16.1 RCPath SAC

- This item was deferred to the next meeting.

16.2 JCHMT SAC in Clinical Genetics

- Dr Kingston reported that PMETB would become effective on 30th September 2005.

- Knowledge based assessments for trainees were due to be piloted. Dr Archer stated that genetics trainees felt well monitored and that they had a good curriculum to work to and did not want further examinations. Dr Kingston reassured that in practice the amount of work needed would not be dissimilar to that which went before.

- Dr Kingston stated that the CGS agenda included the possibility of sponsoring a basic genetics course for genetic trainees.

16.3 Workforce in Clinical Genetics

- This item was deferred to the next meeting.

17. Involvement in Public Open Day at RCP Saturday 9th July 2005

- The Chairman had received a letter from Nicole Barlow (Manager, RCP Patient Involvement Unit) regarding the Open Day. Specialties were asked to provide representations for the day that should be as ‘hands on’ as possible. Members agreed that this was a good idea and that the BSHG should look to co-ordinate this. Professor Sampson agreed to add this to the BSHG agenda.

ACTION: ***Professor Sampson***

- Members noted the offer of displays from the Cambridge Genetics Park and the CGS.

18. For information

18.1 Pharmacogenetics in the NHS

- The Chairman stated that a briefing pack was available from Roche.

18.2 Specialty pages on RCP website

- Members were asked to visit:
  http://www.rcplondon.ac.uk/specialty/ClinicalGenetics.asp
  and supply any alterations or omissions to the administrator.

ACTION: ***All***

- Dr Streetly asked the administrator to provide a link to the NSC on the webpage.

ACTION: ***Committee administrator***

19. Any other business

19.1 RCP Working Party on Medical Professionalism

Members noted the request for a committee response to the questionnaire at http://www.rcplondon.ac.uk/wp/medprof/

It was agreed that a response from the medically trained members of JCMG would be co-ordinated by Dr Kingston. The administrator would circulate the questionnaire to relevant members asking for a response by Wednesday 16th March 2005.

ACTION: ***Committee administrator/Dr Kingston***

20. Dates of the next meetings

- Tuesday 17th May 2005
- Thursday 20th October