Joint Committee on Medical Genetics

The Royal College of Physicians **The British Society for Human Genetics** **The Royal College of Pathologists**

RCPath, 2 Carlton House Terrace, London, SW1Y 5AF

A meeting of the Joint Committee on Medical Genetics was held at the Royal College of Pathologists on Tuesday, 24th October 2006.

Present:	Dr John Crolla	JCMG Chair, (RCPath)
	Ms Amanda Barry	(BSHG)
	Dr Jim Bonham	RCPath, MetBioNet
	Dr Rodney Burnham	(RCP, Registrar)
	Dr Hilary Burton	(Observer, PHGU)
	Dr Trevor Cole	(RCP)
	Dr Teresa Davies	(RCPath),
	Dr Rob Elles	*deputising for* Prof. Richard Trembath, BSHG, Chairman
	Dr Hilary Harris	RCGP
	Dr Susan Holder	RCP, Workforce representative
	Dr Tessa Homfray	RCP
	Ms Dianne Kennard	Observer, DH
	Mr Alastair Kent	GIG/RCP Patient and Carer Network
	Mrs Gail Norbury	RCPath
	Dr Tony Parkin	BSHG
	Professor Peter Soothill	RCOG
	Dr Fiona Stewart	(RCP)
	Dr Virginia Warren	FPH
In attendance	Dr Raj Lakshmi	(Public Health Genetics Unit)
	Drmelissa Martyn	(NHS National Genetics Education and Development Centre)

1. Apologies for absence

NOTED: apologies were received from Dr Mark Bale (Observer, DoH), Professor Carol Black (RCP, President), Prof. Paola Domizio (RCPath, Registrar), Dr Sally Davies (RCP), Mrs Hilary Grandey (RCP, Patient and Carers Network), Dr Shirley Hodgson (BSHG), Dr Sian Morgan (RCPath Trainee representative), Professor Adrian Newland (RCPath President), Ms Su Stenhouse (BSHG), Dr Allison Streetly (NSC, Observer), Dr Karen Temple (BSHG), Prof. Richard Trembath (BSHG, Chairman),

2. Membership

NOTED: that Dr Geoff Woods had been nominated to act as the Royal College of Paediatrics and Child Health (RCPCH) representative, and Dr Anneke Lucassen and Ms Amanda Barry as the British Society for Human Genetics (BSHG) representatives.

Mrs Kim Smith had succeeded Dr Tony Parkin as Chairman of the Association of Clinical Cytogeneticists (ACC) and therefore would replace Dr Parkin on the JCMG. The Royal College of Physicians (RCP) were in the process of nominating a trainee representative to serve on the JCMG.

3. To confirm and sign the minutes for the meeting held on 19th January 2006

NOTED: a number of amendments that had been suggested by Dr Allison Streetly relating to minute number 18 of the previous minutes, concerning 'Screening for Cystic Fibrosis'.

AGREED: the minutes of the meeting were confirmed and signed as a true record with the following amendments to minute 18:

- the title of that section should read as 'Newborn Screening for Cystic Fibrosis'.
- In paragraph 3, 'Dr Alison Streetly' should read as 'Dr Allison Streetly.
- In paragraph 4, 'screening protocols' should read as ‘screening policy'.
- In paragraph 8, 'Professor Sir Muir Grey' should read as 'Sir J A Muir Grey'.

In addition, a comment to the effect that not all centres offered mutation checks should be included in minute 4 (g) concerning PPUK Duchene Muscular Dystrophy (DMD) Registry.

Action ***Committee Administrator***

4. Matters arising on the Minutes

a) Expensive Drug Therapies

NOTED: a copy of a letter from Mr Andy Burnham MP, Minister of State for Health, in response to a letter from the JCMG about funding enzymatic therapies for ultra-orphan conditions. Dr Bonham noted that there was concern about the cost of such therapies, and that some kind of guidance was required. The number of conditions being covered by this was expanding. Pharmaceutical companies were taking a proactive approach in encouraging change in this area.

Dr Stewart noted that the central funding available for these treatments only applied to England and did not currently extend to other parts of the United Kingdom.

AGREED: that it would be useful to revisit this issue in a few months' time.

Action ***Dr Crolla***

b) Guidelines Clearing House

NOTED: that the Manchester Knowledge Park was awaiting a response from the Department of Health regarding a request submitted to the Department of Health for funding to establish a Genetics Guidelines Clearing House.

Early indications seemed to suggest that the Department of Health would prefer the project to be restricted initially to clinical guidelines before considering whether to extend the project to cover laboratory guidelines. To date, no further comments or response from the Department of Health had been received.

Dr Cole noted that, contrary to the minutes of the previous JCMG minutes, the Guidelines Clearing House proposal had not been referred to the Clinical Genetics Society (CGS) for discussion or approval.

AGREED: that Dr Crolla would circulate to JCMG members any responses that are received regarding the proposal. Dr Cole agreed to inform the CGS Council about the proposal at their forthcoming meeting.

Action ***Dr Crolla, Dr Cole***

c) Higher Specialist Training Toolkit

NOTED : that the "Toolkit" initiative by the Royal College of Pathologists had to a large extent been superseded by Skills for Health's proposed reform of Clinical Scientists' pre-registration and higher specialist training.

AGREED: that discussion of this issue should be deferred till later in the meeting.

d) NSC recommendations on karyotyping for women who have been screened for Down Syndrome

NOTED: a discussion on the JCMG response to recommendations proposed by the NSC on antenatal screening working standards was deferred to item 20.

e) MetBioNet

NOTED: that as agreed at the previous JCMG meeting, Dr Crolla would write to Dr Julia Stallibrass at GENCAG about the importance of continuing the work of the MetBioNet in light of the Specialist Commissioning Review. Dr Crolla would consult Dr Jim Bonham before sending the letter.

Action ***Dr Crolla / Dr Bonham***

g) RCP Payments by Results group

NOTED: that Dr Helen Stewart was acting as the Clinical Genetics Society (CGS) representative on the RCP Payments by Result (PbR) group.

Dr Burnham noted that there had been no significant progress to report although it was recognised that the group was acting as a useful and wide-ranging forum, with representatives from all 26 medical specialties, and a useful means of promoting better understanding with the Department of Health.

Key difficulties seemed to be that the coding was not accurate and that some specialties, such as rehabilitative medicine, had not been included in the initiative. The 'Do Once and Share' programme was looking at how genetic coding could be improved.

The next meeting of the RCP Payments by Results (PbR) group was on 9th November 2006.

AGREED: Dr Crolla would ask Dr Helen Stewart if she would be able to keep the JCMG informed of developments on PbR group.

Action ***Dr Crolla***

i) ***HER2*** **– Implementation of testing across NHS Laboratories – 'Evaluating & introducing new diagnostic tests: The need for a future strategy.**

NOTED: details of an RCPath initiative to highlight the need for a single national body for the evaluation and introduction of new diagnostic tests. Professor Peter Furness, RCPath Vice President, was acting as the lead on the project.

AGREED: that given the increasing importance of clinical and laboratory genetics, it was important for there to be genetics involvement in the project. Dr Crolla agreed to contact Professor Furness and suggest that the UKGTN, MetBioNet, the National Reference Genetics Laboratories, and Genetics Interest Group (GIG) would all be interested in participating in the initiative.

Professor Furness was planning to hold a stakeholder meeting to take the initiative forward.

Dr Crolla would write to Professor Peter Furness to inform him about the information that Dr Bonham had brought to the attention of the JCMG, regarding *HER2*. The arrangements relating to *HER2* appeared to be part of a wider problem that Professor Furness was addressing in the new NHS diagnostic tests initiative.

Action ***Dr Crolla***

i) Medical Specialties Board

NOTED: Dr Tessa Homfray, who had kindly agreed to represent the JCMG on the Board, had attended a recent meeting of the Board but nothing had been discussed with a direct bearing on medical genetics.

AGREED: that it would be useful for Dr Homfray to continue to attend meetings of the Board and keep the JCMG appraised of developments.

Action ***Dr Homfray***

5. Publication of Consent and Confidentiality in Genetic Practice

NOTED: that the report had eventually been published and distributed to key members of the genetics community, including all current members of the British Society for Human Genetics (BSHG), the JCMG and important political and pressure groups such as the Human Genetics Commission. The report had also been posted on the BSHG, RCP, RCPath and RCPCH websites.

The BSHG and the RCP had kindly covered the initial publication costs while the RCPath and the RCP had been asked to cover the distribution costs.

NOTED: that a meeting was due to take place the following week in Manchester to be hosted by NOWGEN, on the topic of the Human Tissue Act in practice.

6. Report from the Genetics Unit, Department of Health

NOTED: a written report from Ms Dianne Kennard regarding NHS Genetics. A 3 year progress review of the genetics white paper was underway. Comments and feedback from a range of stakeholders about forthcoming developments and future priorities had been invited. The outcome was expected to be published in early 2007.

The laboratory equipment purchased with the £18 million White Paper investments to upgrade NHS genetics laboratories had been in operation for a while and the aims of the bids were being realised.

Five famlial hypercholestolaemia, seven famlial cancer pilots, ten service development initiatives and nine GP posts with a special in genetics were all well underway. The planned outgoing fellowships to promote learning exchange in the application of genetics to healthcare had now been completed.

A competitive selection process to select a university to host the white paper funded chair in Pharmacogenetics had been undertaken and a preferred bidder had been selected. A tendering exercise was being initiated for the continued development and delivery of Genepool, the specialist library for genetics within the National Electronic library for Health (NLH).

The results to the fourth survey of regional genetics centres were being analysed before they were to be presented to the Genetics Commissioning Advisory Group.

7. Reports from the National Genetic Reference Laboratories

NOTED: for information, an update report from the National Genetics Reference Laboratories based at Manchester and Wessex. The report from the NGRL Manchester gave details of the progress being made in new diagnostic tests, including the high throughput sequence based mutation screening system and the new Abbott fragile X diagnostic test. Dr Bonham also noted that the Metabolic Biochemistry laboratories were purchasing the STARLIMS laboratory information management system which was also being adapted for use in several regional genetics laboratories.

Professor Soothill asked whether it would be possible to have a more structured way of dealing with rapidly developing technology particularly with respect to tests utilizing free fetal DNA (non-invasive prenatal diagnosis). Dr Elles agreed that the NGRL would be willing to play a role in developing such a process but stressed that it would need guidance from the new steering group, which was responsible for setting priorities for the NGRL. Dr Trevor Cole expressed the view that these emerging tests would be likely to lead to a significant increase in cases. Currently, the National Blood Transfusion Service based in Bristol was offering sexing of free fetal DNA in an antenatal setting.

AGREED: 1: that the JCMG should convene a working party to look at the issue and make recommendations about the how and when these tests could be adopted within test repertoires of Genetics Laboratories. A stakeholders meeting could also be organized on the issue. The recommendations would be submitted to the NGRL Steering Group. Dr Tessa Homfray agreed to chair the working party which would also include Dr Hilary Burton, Mr Alastair Kent, Mrs Gail Norbury and Professor Soothill.

Dr Neil Avent, Head Scientist at the Special Non-Invasive Advances in Fetal and Neonatal Evaluation (SAFE) could be involved in the consultation process, particularly in relation to the input of families.

2. Dr Bonham agreed to forward Dr Elles details of the work being done by STARLIMS across all other pathology disciplines.

Action ***Dr Homfray, Dr Bonham***

8. **Genetics Commissioning Advisory group**

NOTED: an update report from the UK Genetics Testing Network Steering Group, detailing the UKGTN initiatives and the progress achieved between March 2006 and September 2006.

Specialist Cytogenetics laboratories would shortly be receiving letters inviting them to apply to join the UKGTN. Membership would be extended to cytogenetics laboratories that meet the membership criteria and provide testing on a national basis that would benefit from a network approach.

Dr Bonham asked, whether in wake of the Carter Report on Pathology services, the status of genetics as a specialised service was being reconsidered. Ms Kennard confirmed that she was not aware of any changes in this area.

Attention was drawn to a letter from the UKGTN steering group response to the NSC's draft consultation document 2006 on antenatal screening working standards.

The letter stated that the UKGTN was of the view that QF-PCR and karyotyping tests should not <u>both</u> be offered to women deemed to be at high risk following Down Syndrome screening. The UKGTN also expressed concerns about the wording of the NSC's working standards relating to diagnostic testing, which needed to be clarified so it could be understood by women undergoing a test. In particular the term 'higher risk' needed to be defined more clearly.

Action ***Dr Crolla***

9. **Human Tissue Authority**

NOTED: a copy of a document produced to give practical guidance to clinical and laboratory geneticists with regards to the Human Tissue Act when it comes into effect. The document, which had been produced by a JCMG short life working party headed by Alison Hall, contained a number case scenarios relating to how the Act might impact on laboratory and clinical genetics practice.

AGREED: the document should be considered for approval by the RCP Council. Dr Crolla agreed to email a copy of the document to Dr Burnham who would bring it to the attention of Council. The document had already been agreed by the BSHG Council and posted on the BSHG website; the RCPath and RCP Councils would be asked for their approval for inclusion of this paper on their respective websites.

The document was welcomed as an excellent guide to a complex piece of legislation. Dr Alison Hall and Dr Anneke Luccasen were thanked for the lead they had taken in writing the guidance.

Action ***Dr Crolla, Dr Rodney Burnham***

10. Educational Issues

i) JCMG Multidisciplinary Education Group

NOTED: that the multi-discplinary education group work on the web-based education package on ethical, legal, spiritual and community issues was still ongoing. Pamela Black had done considerable work to format the materials. Rajesh Summan of the National Genetics Education and Development Centre was preparing the webpages. The NGEDC would be hosting the package on their website.

ii) NHS Genetics Education and Development Centre

NOTED: a written report from the NHS National Genetics Education and Development Centre, detailing the current activities since May 2006. Professor Soothill drew reference to the work being done by the centre with Obstetrics and Gynaecology SpRs.

iii) Report from Genetics Counsellor Training Panel

NOTED: that Ms Amanda Barry had been appointed to the JCMG and would be providing an update on the Genetics Counsellor Training Panels at future JCMG meetings.

Action ***Ms Barry***

11. National Metabolic Biochemistry Network

NOTED: an update report from Dr Jim Bonham regarding the National Metabolic Biochemistry (Biochemical Genetics) Network. Recent developments included the development of a 'Metabolic Map' by higher specialist trainees, which was due to be produced by Sigma Chemical Co and distributed internationally.

It was noted that the Department of Health had agreed to establish a Strategic Advisory Group to assist with the implementation of the strategy put forward in the 'Metabolic Pathways – Networks of Care' document. The group had been established in response to representations made by the JCMG and others. Two meetings of the group had taken place and another one was scheduled for 13th December 2006.

The group would be helping define standards and suggesting a service delivery model, capable, with commissioner support, of delivering these standards of care on an equitable basis across the UK. The recommendations were expected to be submitted to the national specialist services commissioning group early in 2007.

Professor Soothill noted that the scope of the network guidelines on 'Investigations of hydrops' were too narrow and did not include any fetal aspects. The causes of Hydrops were much wider. Professor Soothill volunteered to assist in improving the guidance to include fetal aspects.

Action **Professor Soothill**

12. UK Haemophilia Centre Doctor's Organisation (UKHCDO)

NOTED: a report from Dr Alan Fryer on the UKHCDO Genetics Working Party meeting held on 11th September 2006. The next meeting of the working party was in February 2007.

13. Manpower and Training

a) RCPath SAC

NOTED: Dr Crolla noted that there was nothing to report, as many of the issues discussed at the previous SAC meeting had been already been covered by JCMG.

b) JCHMT SAC in Clinical Genetics

NOTED: that Dr Davies had not been able to attend the meeting and was not able to give the JCMG a report on the JCHMT SAC in Clinical Genetics.

c) Workforce in Clinical Genetics

NOTED: that Dr Holder**,** the JCMG workforce representative, reported that there was some evidence to suggest a pending shortfall in the number of consultant posts available for the current trainees on completion of their CCT.

Trainees had expressed concerns about the prospect of fewer applicants for training posts in the future and the resulting fluctuations in the numbers of trainees coming through for vacant consultant posts. The current financial crisis in the NHS had seen a relative freeze on consultant expansion in the specialty, limiting plans for further service development.

As this followed a significant expansion in both consultant and SpR numbers, taken up by relatively young consultants, it was likely to lead to fluctuations in the future.

Despite the advances in recent years, Dr Holder noted that it was recognised that there was still a shortfall in WTE consultant numbers when compared to the Royal College Physicians (RCP) recommendations of 3 department specialists per million population. At present there were about 90 WTE consultant posts, whereas the recommendations suggested there ought to be 153.

The NHS Workforce Review Team predicted that about 7 consultant posts would become vacant in 2006 and 8 posts in 2007. On average, between 10 to 14 SpRs would gain their CCT each year. It was identified as a priority that the figures on unfilled consultant posts and time-expired SpRs be collected.

The situation seemed to be similar, if not worse, for trainee genetics counselors. Trained scientists were also experiencing difficulties securing senior posts.

d) Workforce in clinical laboratory scientists

NOTED: a tabled written update on White Paper funded training posts, produced by Dr Su Stenhouse. The report gave details of some of the training initiatives that had been undertaken since the appointment of National / Regional Training Officers financed by the Genetics White Paper.

Dr Teresa Davies expressed concern that the Workforce Development Confederation (WDCs) posts had not been filled, meaning the workforce had not increased as previously expected. This had been further exacerbated by the lack of posts due to the ongoing financial difficulties in the NHS.

JCMG members expressed concerns about plans being developed by the Department of Health to change Higher Specialist Training for Clinical Scientists under the "Skills for Health" initiative. The proposals includes reducing the pre-registration training period from 4 to 3 years, and introducing an MSc or taught PhD qualification to coincide with HPC state registration. The pre-registration training would also include both generic (i.e. across all clinical scientist disciplines) and specialist training, with much of the former sub-contracted to HEIs. The plans also involved the reorganization of clinical scientist Higher Specialist Training and the recruitment of pre-registration trainees is to be linked directly to the number of consultant posts. The stated aim of the proposals was to create a more adaptable and flexible clinical scientist workforce.

JCMG members expressed concern that the proposals would not produce clinical scientists who are fit for purpose especially at the proposed new point of state registration after three years. There was uncertainty about who would be providing the training (HSEs or the Health Service) under the new proposals. The initiative, which was being led by Professor Sue Hill, Chief Scientific Officer, was due to be introduced in 2008 – 2009.

AGREED: that Dr Crolla would write to Professor Sue Hill to express concerns that the proposals for pre registration training were unlikely to produce genetics clinical scientists who could function as independent practitioners and that significant change at this time would be particularly detrimental in the context of rapidly changing technologies.

Action ***Dr Crolla***

14. Presentation on 'Teaching Medical Genetics to Medical Students' by Dr Melissa Martyn, Education Development Officer, National Genetics Education and Development Centre

NOTED: Dr Melissa Martyn, Education Development Officer at the National Genetics Education and Development Centre, presented the revised document on teaching genetics to medical students at the undergraduate level. The document had been last revised in 1996.

Dr Martyn gave details of the learning outcomes of genetics and how they are to be delivered.

It was noted that Professor Peter Farndon would be bringing details regarding learning outcomes for non-genetics healthcare professionals to a future meeting of the JCMG for discussion and approval.

AGREED: the JCMG, on behalf of the medical schools genetics leads, gave its endorsement and support to the revised version of the document. It was hoped the outcomes would gain final agreement in Spring 2007. Dr Martyn was thanked for delivering the presentation.

NOTED: the committee was asked for its views on the suggestion that a National Genetics Education Development (NGEDC) be coopted onto the JCMG in an observer capacity.

AGREED: to give its approval to the proposal that a National Genetics Education Development Centre (NGEDC) representative be invited to serve on the JCMG as an observer.

Action ***Dr Crolla***

15. Newborn Screening for Cystic Fibrosis

NOTED: Mrs Norbury noted that a decision on funding the newborn screening programme in London had been postponed until 2007-2008.

The committee was updated on a recent CMGS best practice meeting on CF which reviewed what was happening around the UK with DNA testing following CF newborn screening. Scotland (Glasgow) was still testing with CF-HT for ~30 mutations. Newcastle would be setting up the 4 mutations plus 2 local mutations. Wales had approval to do a panel of 8 and would only move to the national algorithm if/when shown to be effective. Oxford using CF OLA and masking all but allowed 4 mutations. Only Sheffield and one other region were following the NSC protocol. There was no funding still for London and therefore the NSC's testing algorithm was not being rolled out. The importance of systematic audit against all the agreed standards was emphasised as a means of checking what was happening around the country.

There was additional concern regarding the current lack of a formal EQA programme and availability of suitable control material for the various combinations of mutations. Dr Bonham reported that there were discussions planned to consider EQA arrangements related to all newborn screening programs including sweat test measurement and related CF mutations. These would involve NEQAS, the newborn screening program centre and CMGS representation. A CF advisory group planned for 14.12.06 and the Dried Blood Spot advisory group planned for the 9.01.07 would consider these and related issues. Dr Bonham could update the next JCMG meeting.

There were particular concerns relating to the appropriateness of the selected primary mutation panel in ethnically diverse parts of the country and ethical concerns related to the use of assays which provide information on more than 4 mutations but only go onto report the 4 selected within the National algorithm. Dr Crolla would convey these concerns and those relating to the diversity of practice to Barbara Judge at the Program Centre and to the Chair of the Blood Spot Advisory Group.

Action ***Dr Crolla, Dr Bonham***

16. RCPCH report

NOTED: that Dr Geoff Woods had not been able to attend the meeting and was not able to give the JCMG a report on the RCPCH.

17. NICE Famlial Breast Cancer Update consultation

NOTED: for information, a copy of feedback put together by Dr Diana Eccles and Dr James Mackay in response to the consultation process relating to the Famlial Breast Cancer update. The comments had been submitted to the National Institute for Health and Clinical Excellence (NICE).

18. Research

NOTED: a discussion document produced by Dr Jim Bonham on the subject of 'Best Research for Best Health'. The document noted that the existing arrangements for research funding were due to undergo far-reaching changes and would be entering a three year period of 'transitional funding' during which time the existing funding would be progressively replaced by direct task linked research funding that would be largely obtained following application and peer review.

The new tranche of funding was going to be at or above the previous levels. The research would be of a high quality and geared to the needs of industry.

Dr Bonham also outlined some of the likely implications of the changes, particularly on the training of clinical scientists. Under the new arrangements the successful bids were likely to be large and multi-centre, requiring the specialty to consider mounting joint bids for such funding.

AGREED: that Dr Crolla would be drawing attention to the issue raised in the paper at the forthcoming meeting of the RCPath Council. It was agreed that this was an issue that needed monitoring.

Action ***Dr Crolla***

19. Genetics White Paper Review

NOTED: a survey of the stakeholder responses received so far to the three year review of the Genetics White Paper **'**Our Inheritance Our Future**'.**

AGREED: further suggestions and points raised by JCMG members to be included in the summary which would be submitted to the Department of Health. Dr Crolla agreed to incorporate these comments before submitting the JCMG response to the Department of Health.

Overall**,** the JCMG provided a very positive response to the aims and achievements to date of the Genetics White Paper but recognised that for this to move forward careful consideration needed to be given to how current and future initiatives were to be maintained, disseminated and, most importantly, funded via the commissioning process.

The committee's responses were as follows:

Main achievements of the White Paper.

1. *The White Paper (taken in conjunction with the Milburn initiatives) had positively impacted on raising the profile of Genetics in the U.K.*
2. *There had also been positive responses from many other areas of medicine.*
3. *A positive impact on driving integration of genetics with other disciplines. In this context the integration of IT via the NGRL Manchester and the procurement of the STARLIMS by an increasing number of Laboratories, including those in the MetBioNet network.*
4. *There had been an expansion of the number of disease gene tests.*
5. *The White Paper had also raised expectations but there was a question as to whether these could all be met.*
6. *The White Paper investment had helped to avoid until now possible fragmentation of services during the current rapid period of growth in technology.*

7. *There had been improved benefits to patients (e.g reporting times and test repertoire expansion). There remained a need for a further raising of awareness of genetics in primary and secondary centres.*
8. *GPs are more aware of genetics as are their patients.*
9. *The NGRLs potentially provided an ability to assess the whole life cycle (evaluations and technological) of technologies including their integration into mainline clinical testing and their long term impact(s).*

Current & Forthcoming developments.

1. *The development of parallel sequencing (potential for individualised whole genome sequences to be produced rapidly and cost-effectively).*
2. *The increase in predictive testing – assessments of the true breadth of risks.*
3. *It was important that patients with "traditional" gene disorders were not left behind in the technological revolution.*

Ethical and regulatory issues.

1. *It was important to recognise and address the social and economic differences (inequalities) in current access to genetic services which was particularly true for cancer genetics.*
2. *We need to maintain an European-wide perspective to regulations (e.g. Eurogene Tests, the OECD initiative).*

Priorities for the future.

1. *It was essential to maintain the momentum of the White Paper.*
2. *Target test turnaround times needed to be more focused and appropriate. e.g. Why set a 10 day reporting time target for a patient who might not be seen for several weeks in a genetics clinic?*
3. *Genetics education within primary care was vital but was currently at a very low level of activity.*
4. *Genetics needed to be included in GP's "quality framework outcomes" otherwise it would not be included in this vital audit of GP's workflow and accountability.*

20. NSC Consultation document on 'Antenatal Screening Working Standards

NOTED: the JCMG was asked for their comments on the NSC's consultation on the draft working standards for antenatal screening for Down Syndrome. The deadline for responses had been extended to allow the JCMG the opportunity to consider the standards. Dr Hilary Harris had sent Dr Crolla a number of comments which would be incorporated into the JCMG response. It was agreed to focus the discussion on the section of the standards dealing with 'Diagnostic Testing'.

The following responses to Section 14 "Diagnostic Testing" were approved by the committee:

The JCMG endorsed the recommendation that "A QF-PCR test **and** karyotyping should be offered to all women receiving a higher risk test result." A "higher risk" should be defined as "high enough to warrant an invasive procedure."

The JCMG recommended that following QF-PCR +21 positive test results, follow up cytogenetic studies were essential to assess recurrence risks (Robertsonian translocations).

The clinical and scientific justification for not offering testing for sex chromosome abnormalities was questioned.

The JCMG questioned whether appropriate counselling was in place or was planned which would allow women (couples) to make a fully informed choice for this option.

21. NICE Famlial Hypercholesterolaemia guideline

NOTED: that Professor Tim Altman had been asked by the British Society for Human Genetics (BSHG) and the Clinical Genetics Society (CGS) to act as the clinical genetics representative on the working party considering the NICE Hypercholesterolaemia guideline consultations. Professor Aitman had agreed to keep the JCMG informed of developments.

22. Invitation to bid – Specialist Library for Genetics in Healthcare within National Library for Health

NOTED: Ms Kennard noted that the current contract for Genepool, the specialist library for genetics within the National Electronic Library for Health (NLH) had expired at the end of July 2006. The Department of Health with NLH had undertaken a tendering exercise for the continued development and delivery of the services, which had recently been completed.

23. OECD guidelines for Quality Assurance in Molecular Genetic Testing

NOTED: written comments from Dr Su Stenhouse on behalf of the JCMG regarding the OECD guidelines for Quality Assurance in Molecular Genetics Testing. The document was welcomed as a comprehensive summary covering the major aspects of quality assurance in molecular genetics laboratories.

Dr Elles noted that the period of public consultation on the guidelines had ended recently though, as the next stage for developing the guidelines would be conducted at the level of national governments, any further feedback or comments could be made via the Department of Health.

24. JCMG membership

NOTED: discussed under items 2 and 14.

25. Programme for GTAC / GAIC public meeting Leeds 14 November 2006

NOTED: for information, details of the programme of the GAIC and GTAC public meeting due to be held on 14th November 2006. The meeting would be held on 'Research in Cancer in Genetics Teaching, Gene Therapy and Insurance'.

26. Any other business

i) RCP Project 'Explaining the risks and benefits of treatment options'

NOTED: details of an RCP initiative aimed at improving communication between patients and their hospital doctors in order to suggest ways in which greater understanding of the doctor / patient relationship could produce more effective treatment and care. A key element of the project was explaining the risks and benefits of treatment options. The Genetics Interest Group (GIG) had been involved. The initiative did not directly concern medical genetics.

AGREED: that Dr Crolla would circulate further details to Dr Tessa Homfray.

Action ***Dr Crolla***

ii) RCP new process for establishing College Working Parties

NOTED: for information, details of the new process for establishing College working parties at the Royal College of Physicians (RCP).

iii) RCP Conference Programme 2008

NOTED: that the JCMG had been invited to submit proposals for conferences to be included in the RCP Conferences Programme in 2008.

AGREED: that 'clinical scientists in training' might be a suitable subject for such a conference.

Action ***Dr Crolla***

iv) RCP College Lectureships 2007

NOTED: that the RCP was seeking nominations for five College lectureships for 2008. Prof Veronica Van Heyningen was suggested as someone who could be nominated by the JCMG.

AGREED: that Dr Crolla would nominate Dr Van Heyningen for the 2008 Croonian lectureship.

v) RCPath Annual report & JCMG

NOTED: that a short summary of the work done by the JCMG over the last year had been included in the RCPath Annual Report, helping raise awareness of the committee amongst RCPath members.

vi) RCP Clinical Excellence Awards

NOTED: that Professor Trembath had agreed to take the lead on this for the JCMG. Further details would be sought from Professor Trembath.

vii) RCP Public Open Day

NOTED: that the RCP had been very grateful to the members of the JCMG who had assisted with the clinical genetics stand at the RCP Open Day. The event had been a considerable success.

26. Dates of forthcoming meetings (at Royal College of Pathologists)

NOTED: Thursday, 25th January 2007 at 11:00am
Thursday, 15th May 2007 at 11:00am
Thursday, 23rd October 2007 at 11:00am