UK GENETIC TESTING NETWORK CLINICAL & SCIENTIFIC ADVISORY GROUP
THIRD MEETING, 27 SEPTEMBER 2012
FRIENDS HOUSE, EUSTON, LONDON

Present:

Rosalind Skinner (Chair)
Jacquie Westwood, *UKGTN Director*
David Baty, *Clinical Molecular Genetics Society*
Peter Baxter, *Royal College of Paediatrics & Child Health*
Peta Campbell, *UKGTN Project Team*
Jane Deller, *UKGTN Project Team*
Angela Douglas, *Association for Clinical Cytogenetics*
Deirdre Evans, *Devolved Countries - Scotland*
Steve Hannigan, *Patient Representation, CLIMB, National Information Centre for Metabolic Diseases*
Mark Kroese, *UKGTN Public Health Advisor*
Jackie Mcall, *Devolved Countries – Northern Ireland*
Nick Meade, *Patient Representation, UKGTN Genetic Test Evaluation Working Group (attending for Alastair Kent)*
Shehla Mohammed, *UKGTN Clinical Advisor*
Christine Morrell, *Scientific Officer (Pathology and Genetics) - Wales*
Roger Mountford, *Chair, UKGTN Laboratory Membership & Audit Working Group*
Carolyn Owen, *Association of Genetic Nurses and Counsellors*
Cathleen Schulte, *Department of Health (Health Science & Bio Ethics Division)(part meeting)*
Su Stenhouse, *UKGTN Scientific Advisor*
Fiona Stewart, Chair, *UKGTN Genetic Test Evaluation Working Group*
Peter Turnpenny, *Clinical Genetics Society*
Susan Walsh, *Patient Representation, Chronic Granulomatous Disorder Society*
Jonathan Waters, *RCPathologists*
Jo Whittaker, *Chair, UKGTN Development Working Group*

Apologies:

Hilary Burton, *PHG Foundation*
Trevor Cole, *RCPhysicians*
Nick Cross, *National Genetics Reference Laboratory - Wessex*
Val Davison, *National Healthcare Science School of Genetics*
Andrew Devereau, *National Genetics Reference Laboratory – Manchester*
Peter Farndon, *National Genetics Education and Development Centre*
Helen Forgacs, *National Specialised Commissioning Team*
Jacqui Hoyle, *UKGTN Project Team*
Alastair Kent, *Genetic Alliance UK*
Bronwyn Kerr, *Joint Committee for Medical Genetics*
Christine Lavery, *Patient Representation, Mucopolysaccharide Society*
Christine Patch, *British Society of Human Genetics (attending for Sir John Burn)*
Colin Pavelin, *Department of Health*
Dr Imran Rafi, *Primary Care Genetics Society*
Nick Sireau, *Patient Representation, AKU Society*
Karen Temple, *Clinical & Academic Genetics*, *Personal Invitation*

Resignations:

Nick Cross, *National Genetics Reference Laboratory - Wessex*
Christine Patch, *British Society of Human Genetics (attending for Sir John Burn)*
Peter Turnpenny, *Clinical Genetics Society*
Jonathan Waters, *RCPathologists*

01. **Welcome and Apologies**

The Chair welcomed Carolyn Owen and Nick Meade to the meeting. Carolyn has recently been appointed as Chair of the Association of Genetic Nurses and Counsellors. Nick was attending in his capacity as patient representative on the UKGTN Genetic Test and Evaluation Working Group; he was also deputising for Alastair Kent. Jo Whittaker was

welcomed as Chair of the UKGTN Development Working Group.

This meeting would be the last meeting for Peter Turnpenny, Jonathan Waters and Christine Patch as their terms of office have either, or were due to, come to an end. The Chair thanked them for their support and contributions to the work of CSAG during their time as members. It was very much appreciated. Nominations for new members will be requested.

02. **Minutes of last UKGTN CSAG Meeting (22.03.12)**

02.1 **Minutes**

Some corrections to the draft minutes were noted:

- It was noted that Susan Walsh was attending on behalf of the Chronic Granulomatous Disorder Society.
- 3.2, 2nd sentence to read as: The report on the findings of the workshop on how genetics could be managed in mainstream medicine will be available shortly.
- 4.2, 4th para: It was noted that in Scotland work is in hand to put in place a Decision Making Framework to link pathology tests with drug prescribing.
- 6.1, 5th bullet point to read as: Develop engagement with 'Map of Medicine'.
- 11.1: Insert a post meeting to note to reflect that since the meeting the number of Clinical Reference Groups has increased to 66.

With these amendments the minutes were approved as an accurate record of the meeting.

03. **Matters Arising**

03.1 **UK Plan for Rare Diseases**

Nick Meade talked through the update prepared by Alastair Kent who was unable to attend the meeting.

RDUK had recently been in discussions with the DH about progress in developing a UK Rare Diseases Plan following the publication which closed on 25 May. Around 350 responses were submitted to the consultation from organisations and individuals. RDUK suggest that this large number of responses demonstrates that rare diseases should be seen as a major issue in terms of health policy making. The UK's four Health Departments were currently working on their summary of responses which they would make publically available in the autumn. It was hoped that the Rare Disease Plan would be launched early next year, although given the broad scope of the plan and the need to get agreement from four different Health Departments, and all the relevant divisions within those Departments, this may be subject to delay. RDUK is keen to ensure the timely launch of the plan.

03.2 **Progress on development of MolUs into GenUs**

At the last meeting, it was reported that Cytogeneticists had agreed to trial the GenU system to determine whether it met the needs of Cytogenetics and whether any adjustments were needed. The UKGTN Scientific Advisor informed members that a very positive meeting took place yesterday to discuss the outcome of the trial. She reported that a number of minor adjustments were agreed which will result in some adjustment of the overall scheme and that Cytogenetics' colleagues were happy to move forward on that basis. A proposal will now go to the UKGTN Laboratory Membership and Audit Working Group (LMA) for ratification. Following this, it will be passed to the Association for Clinical Genetic Science. The LMA intends to publish the complete scheme before the end of 2012 to give laboratories time to adjust their databases so that they can start collecting GenUs from 1 April 2013. The Chair thanked the UKGTN Scientific Advisor for her hard work and commitment to the project.

04. **Human Genomics Strategy Group**

04.1 **Update on Progress**

The DH representative provided the update.

Following publication of the HGSG report in January 2012, the Secretary of State for Health has asked for a shared strategic framework to be developed for the HGSG's recommendations.

The shared strategic framework will have oversight from the reconstituted HGSG which remains under the Chairmanship of Professor Sir John Bell. Membership has changed and the HGSG Board will be informed by three Working Groups:

- Bioinformatics
- Service Reconfiguration
- Education and Training

The Bioinformatics workstream will take forward the recommendation to establish a Biomedical Informatics Institute within the UK. A workshop is planned for tomorrow to consider amongst other things what form this might take. The group will also look at ethical, legal and social challenges.

Proposals on each of the three work streams will need to be produced. The aim is to take draft versions to the HGSG meeting on 15 October 2012.

The feeling from members of this group is that an important element that is missing is engagement with provider services, particularly laboratories and genetic services. It was discussed that the provider arm is not represented on the reconstituted group. This is an issue that concerns members greatly. Another key concern is the lack of clarity about representation on the Training and Education workstream. The DH representative undertook to feedback comments around engagement to the HGSG and highlighted that there are plans to meet with providers and the professional organisations on service reconfiguration.

05. **UKGTN Review of Population Genetic Testing Rates**

05.1 **2007/08 – 2010/11 Report**

The UKGTN Public Health Advisor introduced the paper on the data collection of genetic test activity by UK Genetic Testing Network member laboratories. The purpose of the data collection is to calculate genetic testing rates by different geographical areas. Laboratories (molecular genetic) were asked to submit resident postcodes for genetic test reports issued for the period 1 April 2007 to 31 March 2011. Certain test reports were not required and the list of exclusions changed over time. In addition, the laboratories were asked to flag tests for Huntington disease and breast cancer in the 2008/09 and later data collections, and Fragile X in the 2009/10 and later data collections.

Twenty eight of the 31 (reduced to 30 from 2008/09 onwards) member laboratories in the constituent countries of the UK submitted data for at least one year, although the maximum number of laboratories that submitted data in any given year was 23. Evidence suggests that there has been a modest improvement in the quality of the reporting of valid resident postcodes over time.

The results show wide variation between Strategic Health Authorities (SHAs) in the rates of genetic test report activity.

It was recommended that the results of this report should not, at this stage, be used for direct commissioning purposes as the quality of the data needs to improve.

The Chair expressed concern that not all centres were able to provide data for the 4 years, due to issues with their IT systems. As a reminder, the service specification for specialised services includes a requirement for the data to be collected. All providers of regional genetic services will be required to submit resident postcode data for genetic tests in 2013/14 using GenUs as the activity measure.

The Clinical Genetics Society member said that there was no mention of variance between patients and those requiring and consenting to testing. The UKGTN Public Health Advisor felt that this would be the type of question that commissioning colleagues would wish to explore.

A query was raised as to whether there were any plans to split the data into other areas. The UKGTN Public Health Advisor answered that to do this; it will first require high quality data. Once this has been achieved, the option to perform a split will be there. It was discussed that splitting data between clinical genetics and non clinical genetics will be requested for the 2011/12 data in line with the Clinical Reference Group's Medical Genetics Service

Specification. Analysing data by specialty will be difficult for laboratories because they tend not to code reports by specialty, e.g. paediatrics and neurology. Some members queried whether this would be one step too far.

Members were asked to i) note the findings of this report and agree that these should not be used for commissioning purposes, and ii) support the continued development of this work programme. Members supported the recommendations proposed.

06. **Genetic Test Evaluations**

06.1 **Testing recommendations for 2012/13**

The Chair of the Genetic Test Evaluation Working Group (GTEWG) thanked all members of the Working Group for their time and expertise, which they provide voluntarily. She especially wanted to thank Nick Meade, patient representative member, who brought an additional perspective to meetings.

Members were provided with the recommendations from the GTEWG for inclusion on the NHS Directory of Genetic Testing for 2013/14. The GTEWG Chair said that one of the most important aspects of the evaluation process is to assess how these tests improve patient care. Tests for conditions that would change the diagnostic pathway were highlighted.

The Working Group evaluated a total of 76 applications between February and August 2012. The recommendations are for 50 new genetic tests for funding by Specialised Commissioning arrangements.

The UKGTN received 10 gene dossiers for panel tests, of which 9 were approved and 8 used Next Generation Sequencing. This was an increase on the previous year. A panel test analyses a number of different genes that are associated with a number of related diseases in a single one off test. The conditions tend to have overlap in clinical presentation and/or overlap in biological pathways affected.

The GTEWG Chair reported that, from a clinical perspective, one of the biggest challenges the Working Group has faced this year has been the introduction of panel tests. This has meant an increased complexity of data requiring evaluation. The Working Group recognised that it has been difficult for laboratories to prepare panel test gene dossiers because the current proposal form was designed for single gene tests; the GTEWG Chair paid tribute to labs for the quality of submissions. The form will be revised to make it easier for laboratories to submit panel tests in the future.

Regarding the proposal for testing of RAD51C/D – this gene dossier was not approved. The GTEWG Chair explained that this was a challenge for the group and they sought expertise from the Cancer Genetics Group. The methodology was good. However, it was felt that the evidence for the clinical utility of the test was unclear as data on penetrance was incomplete. The difficulty was determining where this fits into the investigations of those with inherited cancers.

Overall, the GTEWG Chair said that it was good to see NHS research translated into activity and these were very exciting times.

The CSAG endorsed the recommendations and requested that the UKGTN Director present these recommendations to the Specialised Services commissioners. Presentation to commissioners should include a couple of detailed case studies of the cost savings in the diagnostic pathways due to the introduction of these genetic tests and provide advice on additional funding requirements by SCG in England and separately for each of the devolved countries.

The Chair thanked the GTEWG membership for its excellent work in evaluating these gene dossiers.

07. **Patient Representative Feedback**

07.1 The UKGTN has been looking at ways to develop the existing involvement of patient

representative members on all UKGTN groups. Prior to the meeting, patient representatives were invited to consider a range of questions relating to their experience on the Advisory Group/Working Groups. There were generally positive views on all working groups with nobody saying they had a negative experience. Overall, members found the meetings to be very informative and interesting. 'I feel valued', 'there are adequate opportunities to participate in the discussions' and 'my comments have been taken into account during meetings', were some of the popular comments received. Other comments noted were: 'at first I felt out of place but committee members were helpful and would take time to answer my questions during lunch or after the meeting' and 'I sometimes wonder if the group derives any benefit from my presence and occasional comments'. One member commented that he had been invited to visit labs to help understand more about a subject.

In terms of what UKGTN could do better, some of the key points that came out of the feedback were:

- It was a steep learning curve – acronyms, remit of the group.
- Having a group mentor or buddy could help with new members integrate better
- A clear statement of the role as patient representative would help – a sort of job description with reference to the remit/strategy of the CSAG.
- The group needs to consider how 'we fit in'. Maybe targeted joint projects on certain key issues would help but this must not be too onerous because of other demands on patient representative time.
- Great expectations and the frequency of meetings is an obstacle in bonding with the group.
- Certain topics have been known to cause eye glazing symptoms, important though they may be.

The Chair thanked everyone for their openness and constructive contributions. UKGTN will use these to further improve organisational arrangements concerning patient participation. The Chair will liaise with Alastair Kent and colleagues to produce draft guidelines.

08. **Updates from Professional Organisations**

08.1 **Clinical Genetics Society (CGS)**
The President of the CGS provided a briefing note, Appendix A.

08.2 **Association for Clinical Cytogenetics (ACC)/Clinical Molecular Genetics Society(CMGS)**
The Chair of the ACC provided a briefing note, Appendix B.

08.3 **Association of Genetic Nurses and Counsellors (AGNC)**
The Chair of the AGNC provided a briefing note, Appendix C.

09. **Devolved Countries – verbal updates**

09.1 **Northern Ireland**
The representative from Northern Ireland provided the update. She was pleased to announce that a microarray service has been introduced for patients in Northern Ireland.

It was noted that a number of organisations participated in the Rare Disease Consultation event that took place in the spring with Rare Disease UK. They also submitted a response to the consultation on the UK Plan.

Other highlights included work was underway to develop a pathology network. Any information that Scotland and Wales could share regarding this would be helpful.

09.2 **Scotland**
The representative for Scotland reported that good progress is being made by the Implementation Group established to take forward the development of a National Consortium for molecular pathology in Scotland. The range of molecular pathology tests provided by different laboratories in Scotland have been mapped and work is now focusing on assessing costs to allow funding to be top sliced from individual NHS Boards to fund molecular pathology centrally.

Work is continuing on developing a Decision Making Framework for which cancer molecular pathology tests should be undertaken, and in how many locations in Scotland. There is close collaboration with the Scottish Medicines Consortium which makes recommendations on which medicines are approved for use in Scotland.

09.3 **Wales**

The Scientific Officer (Pathology and Genetics) – Wales representative provided the update. Genetic testing for inherited cardiac conditions for Welsh patients will be available from 1 October 2012 and genetic testing for certain types of cancer is also imminent.

10. **Websites**

10.1 **UKGTN – new website**

The UKGTN Programme Manager guided members through the slides prepared by the UKGTN's Knowledge and Communications Manager who was unable to attend the meeting. The slides covered key activities since the last update and a couple of screen shots of different areas of the website. One reason for revamping the website was to develop an easier admin interface for labs to edit their own data as the current system was too cumbersome. These changes would enable labs to keep their data up to date. The UKGTN Programme Manager reported that labs would be contacted within the next few weeks to organise the pilot of the test service interface. She reported that the plan was to go live in November.

10.2 **National Laboratory Medicine Catalogue – update**

The representative from the RC Pathologists provided the update.

The NLMC is designed to provide accredited information on the scope and clinical value of laboratory tests in Pathology. It will be a national resource accessible from any Trust's information systems. The first draft version of the catalogue (which does not have a Genetics component) is due to be launched on Friday, 28 September.

Gail Norbury has recently been appointed as Genetics lead for this project and will promote harmonisation between the NLMC and UKGTN online database. Gail will be presenting on progress with the NLMC at the UKGTN Conference on November 22 November.

10.3 **Genetic Testing 4 Health**

Members were provided with the latest news on this joint project between NGEDC and the UKGTN. A handout was circulated. In summary:

- The structure and architecture of the website are complete and nearly all content has been uploaded. The aesthetics of the different sections of the site are being harmonised before the site is made live.
- The site was previewed at the Conference of the British Society for Human Genetics, and feedback was very positive – particularly about having brought information about genetic testing together in one place.
- There are sections for patients and the public, and for healthcare professionals.
- Currently there are summaries of 21 conditions most commonly tested for, 18 videos of a consultant explaining what the genetic laboratory findings mean for the patient, 8 interactive learning objects which explain the words and meanings of laboratory reports about specific types of mutations, 20 videos of laboratory techniques, and explanations of the patterns of inheritance.

Members were invited to access the holding page and to send any comments to Peter Farndon, copying in UKGTN. The link is: www.genetictestingforhealth.nhs.uk

11. **UKGTN Business**

11.1 **Review of UKGTN work programme – 6 months progress**

Status of the 2012/13 work programme was noted and endorsed.

11.2 **UKGTN 3rd Report**

The UKGTN Director thanked those members who have contributed to the development of UKGTN's 3rd report. A first draft will be circulated soon. The final report will be launched at the UKGTN conference on 22 November.

11.3 **UKGTN Conference**

UKGTN's 3rd biennial conference, which will take place on 22 November 2012, will focus on transformation – changes to improve patient care. The agenda will explore test evaluation and equity, developments in testing and system enablers in commissioning across clinical genetic services.

12. **Any Other Business**

12.1 **Meeting Dates in 2013**

Noted:

- 21 March
- 26 September

APPENDIX A

CGS Report for UKGTN CSAG Meeting, 27th September 2012

1. BSHG / CGS Constitutional Developments

Since the last UKGTN CSAG meeting the CGS Council met once, on June 13th, a special meeting convened to discuss developments in BSHG and the implications for the CGS constitution. The decisions reached were as follows:

i. The CGS supports the growth of BSHG to embrace the broader constituency of 'genetic medicine' and the restructuring required to achieve this; however, it is not convinced that the BSHG needs to change its name

ii. The CGS welcomes a stronger administrative structure that would offer support to CGS activities

iii. The Council strongly prefers to retain the name of CGS and is unconvinced by the proposed arguments for a change of name

iv. A significant number of minor amendments to the CGS Constitution were drafted, including removal of the category of Corporate Membership and addition of the option to revoke from membership any member whose activities are contrary to the purposes of the Society, or who bring the name of the Society into disrepute; these changes will in due course be brought to the entire membership and AGM

2. Developments in Specialist Commissioning

Prof. Frances Flinter continues as chair of the Medical Genetics Clinical Reference Group (CRG), one of 60 CRGs (to be increased to 65) for specialised services to be commissioned through the NHS Commissioning Board. Medical Genetics will continue to be a specialised service, though the relationship between commissioning at NHSCB level and CCG or regional level remains unclear, as does the role of Clinical Senates. Prof. Flinter and the CGS Chair (PT) attended the Specialised Commissioning Summit in London on 24.09.12. It was strongly emphasised that CRGs are the key to future decision making in relation to specialised services. The Quality Dashboard for Clinical Genetics is complete.

3. Rare Disease Consultation

The government's consultation period closed 25.05.12 and the CGS Chair encouraged individuals and groups to submit responses. The Chair submitted a response on behalf of CGS, which was first circulated to the Council (available on request). The document is available on the CGS website.

4. Integration / 'Mainstreaming' Initiatives

i. Question setting support, MRCPCH examination

The question-setting group for the MRCPCH exam have expressed a keen interest in having clinical genetics input (similar to that which has been in place for the MRCP exam for some years). Alex Henderson (Newcastle) is exploring this further on behalf of CGS.

ii. Map of Medicine

The CGS is endeavouring to engage with the national Map of Medicine (MoM) programme, seeking to establish our own specialty domain on the website. Negotiations are ongoing. MoM is inviting the participation of interested clinical geneticists as 'Fellows' in an advisory capacity.

iii. GP Forum

This educational organisation for primary care physicians has expressed an interest in running medical genetics study days for GPs around the country. The Peninsula Service is beginning to put together a programme for local consumption initially.

5. Collaborative CPD project with Journal of Medical Genetics

Significant discussions have taken place between the Chair, Allison Lang (BMJ/JMG), Constantin Polychronakos (editor, JMG), and Diana Eccles (CGS Academic Vice-President), regarding this project. The aim is to include a CPD section in most issues of JMG eventually, which will take the form of multiple choice questions, and possibly other formats. In response to the Chair's E-Bulletin in July, there were 3 expressions of interest to participate in this project and a further 4 individuals have been for the necessary panel of reviewers/question-setters.

6. International Initiatives

i. CGS International Scholar

There have been 15 applications for 2 scholarships next year – a new 'high' – and a selection process is underway.

ii. Short Overseas Electives for Trainees

At the Clinical Genetics SAC meeting, 21.09.12, approval was expressed for the development of opportunities for interested trainees to spend short elective periods (~ 1 month) in a recognised genetics centre in a developing country, e.g. India. This will be taken forward and appropriately disseminated.

7. Human Variome Project (HVP)

Prof. Richard Cotton (Melbourne), Chair of the HVP, contacted the CGS Chair to seek support and 'join' the HVP initiative. After an exchange of emails it is clear that Prof Cotton would like any interested individual to join and add to the support for the initiative. The CGS will pass on the message to the membership.

8. Medical Student and Junior Doctor Competitions

The CGS is running an essay competition for medical students, for which a suitable title has just been decided. In addition, for junior doctors who spend some 'taster time' in clinical genetics, there is a competition for the best reports on topics of medical genetics. The prizes are monetary.

9. Welcome Letter to New Members

The Chair has initiated a President's welcome letter to new members, with the first batch recently sent out ... and warmly received!

Peter Turnpenny

27.09.12

Appendix B

ACGS (ACC/CMGS) Update Report October 2012

ACC/CMGS

The Professional bodies are in the process of dissolution following an overwhelming endorsement from both memberships. This process is likely to take several months and the Executives (office bearers) of both constitutive bodies continue to meet to take this forward; a meeting is planned for 15th October in Glasgow.

Angela Douglas has been appointed as Chair of the Association for Clinical Genetic Science (ACGS), the new joint professional body for Genetics. The official launch of the ACGS is planned for the new style BSHG meeting in the Autumn of 2013 in Liverpool. There will be a one day Heads of Genetic Laboratory Services meeting organised in Spring 2013 to communicate the proposed framework and constitution of the new Professional body and set a strategic plan of work for the next 1-3 years.

Academy for Healthcare Science (AHCS)

All Healthcare Science Professional Bodies now have a seat on the Council of AHCS. This is welcome news as the Council is the body that discusses and sets the strategic direction of the Academy.

Agreement has been reached between the Association of Clinical Scientists and the AHCS on the assessment of both old style Clinical Scientist Trainees and MSC STP trainees and the route to registration with the Health and Care Professions Council (formerly HPC).

The Academy has created eight Professional Groups, organised along the broad themes of healthcare science. David Baty has been appointed as Chair of the Genetics' Professional Group and has submitted a list of membership nominations to the Academy. The initial responsibility of the Professional Group will be:

- quality assurance of education and training in collaboration with the National School of Healthcare Science
- final professional assessment of individuals
- equivalence judgements of individuals and education and training programmes

Best Practice

The ACC Heads of Service Leads met in Birmingham in September to discuss and agree the Best Practice Guidelines for aCGH technology. This guidance will be published in due course.

Current CMGS Best Practice guidelines are being updated. New guidelines on HD have been produced in collaboration with EMQN and revised guidelines for variants of unknown clinical significance will be issued in due course. A best practice meeting on next generation sequencing was held in Leeds during the summer and draft guidelines have been produced.

Audit

The CMGS is currently collecting annual audit data from member laboratories. In addition, laboratories are also submitting data to UKGTN relating to patient postcode and whether or not the referral came from Clinical Genetics.

Integrated Workload Units.

From the ACC laboratory responses (13 laboratories trialled system), the majority of laboratories are happy with the new iteration of the integrated genetic workload units (suggested GENU) scheme for measuring laboratory activity. The next steps will involve combining the Molecular and Cytogenetic Trials and looking at any areas of concern. A document will be circulated to Members to inform them of the outcome of the work carried out by the working group and an audit will be carried out. A paper will be presented to ACGS at the inaugural meeting on 15th October, update to be provided by Katie Waters and Lara Cresswell. The Laboratory Membership and Audit committee of UKGTN are to publish the complete scheme. GENU's are to be collected for national commissioning purposes from all labs from April 2013.

Human Genomics Strategy Group

This has been re-constituted to implement the recommendations from the Publication of January 2012. Prof Sir John Bell is chairing this group. There are three workstreams working under this Group:

1) Bioinformatics – Met 28th September – David Gokhale and Andrew Devereaux represented Professional Bodies. AD and DB invited at last minute but short notice meant non-attendance. This group is to look at the proposal for a Bioinformatics institute and what form this will take. How we create linkages with research and industry from provider organisations and how we store information to provide patients with better access to this information.

2) Service Reconfiguration – There is a Shared Strategic Framework Workshop hosted by HGSG planned for 7th Nov 2012. Professional body leads have been invited to attend this meeting.

3) Training and Education – Unclear of representation on this meeting clarity has been sought.

A draft proposal will be prepared from the outcomes of these workstreams which are due to go to a HGSG oversight board starting on ?15th October.

Genetics Education and Training

Only 7 STP trainees commissioned in Genetics for 2012 start cohort. Building on the joint workforce reviews performed by the CMGS and ACC, the ACGS will continue to review the laboratory workforce to ensure that there is appropriate and strategic succession planning. The small numbers of trainee scientists being commissioned is a cause for concern.

CSO NIHR Fellowships

Good response from Profession with submissions to the above, we await the outcome of those submissions.

Birmingham Spring Meeting

Thank the Genetics staff of Birmingham Women's Hospital organising and delivering for a fantastic Spring conference. They have set an extremely high standard that the ACGS will need to aspire to replicate at future Conferences

Appendix C

Association of Genetic Nurses and Counsellors

Administrative Office
Clinical Genetics Unit (BSHG)
Birmingham Women's Hospital
Edgbaston, Birmingham
B15 2TG

A constituent group of the British Society of Human Genetics

AGNC Committee Report for the UK Genetic Testing Network Clinical and Scientific Advisory Group (CSAG) Meeting September 2012

Genetic Counsellor Regulation

As we are all aware the work of the Genetic Counsellor Statutory Regulation Steering group (GCSRSG) has been successful with its application to the HPC in December 2009. However given government changes to regulation a new joint working group has been formed with the Genetic Counsellor Registration board (GCRB). This new body is the Joint Committee on Genetic Counsellor Regulation.

This week the Regulation group will be running a workshop at the BSHG meeting in Warwick.

Recently the Joint Committee on Genetic Counselling Regulation requested that the following message was sent with emails as the group are in the process of gathering opinions from other health professionals and the public regarding the need for professional regulation of genetic counsellors.

"As a means of informally collecting views the following statement can be added to an email signature at your discretion.

Genetic counselling plays a vital role in patient care, but the existing voluntary register maintained by the GCRB is not statutory and genetic counsellors in the future may be employed without the necessary education, skills and training. Statutory regulation is important for maintaining the highest levels of patient safety. Support the Joint Committee on Genetic Counsellor Regulation by giving us your opinion on:

http://www.surveymonkey.com/s/68D7BVB "

Regulation work continues to gather the data for risk assessment document. This data will be published in a Q&A document which will have a full summary of the situation. This information will be published soon.

JCGCR members

Georgina Hall, Christine Patch, Mark Longmuir, Jan Walford-Moore, Jan Birch, Chris Barnes, Buddug Cope (Genetic

The Genetic Counsellor Training Post (GCTP) scheme

The GCTP scheme, funded by the Department of Health is nearing an end.

- Of the 9 trainees in the jointly-funded third phase of the scheme, 2 have obtained substantive genetic counselling posts, 4 are due to complete their training during 2012, with the remaining 3 trainees scheduled to finish their post in 2013.

- Of the 43 trainees appointed in the first two (fully DH-funded) rounds of the scheme, 42 went on to work as genetic counsellors, of whom 38 have already gained professional registration with the GCRB.

- Recently, an article about the GCTP scheme, including its historical background and outcome data, was published in the Journal of Community Genetics. The reference is as follows:

  Barnes C, Skirton H, Kerzin-Storrar L, Tocher J (2012) The Department of Health supported Genetic Counsellor Training Post Scheme in England: a unique initiative? J Community Genet. DOI 10.1007/s12687-012-0100-6

  (The article was published online on 31 May 2012, but will appear in the future journal issue, details as yet unknown)

The Panel hopes that the publication may be of help to international colleagues who are in the process of developing genetic counsellor training in their own countries.

Locally-funded training posts and approval of training centres.

The GCTP Panel, the AGNC Committee and the Genetic Counsellor Registration Board (GCRB) remain committed to the continuation of structured training posts and approval of training centres. Although the DH funding for training posts is now at an end, the Panel remains committed to offering structured support to Centres who continue to employ trainee genetic counsellors. **To this end, the Panel is very keen to hear from those centres who fulfill the criteria for an approved GC Training Centre (or are interested in finding out more about such approval), and have a training/Band 6 post in their department.** All enquiries about this can be sent to Chris Barnes.

The future

Not only is the Panel committed to supporting Centres with their efforts to embed recurrent training posts in their department, it is committed to exploring avenues for future structures and funding to ensure the continuation of high-quality genetic counsellor training in the UK. To this end, the Panel will be contacting all genetic centres shortly to canvas information about local situations, views and ideas.

GCTP Membership

Judy Tocher has taken over as Chair of the GCTP Panel, and Chris Barnes will continue to act as Panel Administrator (as well as Panel member until the end of 2012). They are delighted to welcome new Panel members Claire Dolling, Sue Kenwrick and Rhona Macleod.

GCTP Panel

July 2012

Judy Tocher (Sheffield) (Chair) judy.tocher@sch.nhs.uk

Claire Dolling Birmingham claire.dolling@bwhct.nhs.uk

Sue Kenwrick (Cambridge) sue.kenwrick@addenbrookes.nhs.uk

Rhona Macleod (Manchester) rhona.macleod@cmft.nhs.uk

Chris Barnes (London) cabarnes@blueyonder.co.uk

AGNC committee structure

The AGNC Committee has undergone a number changes over the last few months with Mark Longmuir coming to the end of his term as AGNC chair. The AGNC are very grateful to Mark for all his hard work during his six years on the committee and particularly for his unerring commitment during his two years as AGNC chair. We wish him all the best for the future.

With the departure of Mark, Carolyn Owen will be the new AGNC chair. Carolyn has been Vice-Chair of the AGNC for the last two years and is looking forward to the opportunity to leading the AGNC and helping to further develop the genetic counselling profession. Our current Secretary, Oonagh Claber, will assist Carolyn in her new role as Vice-Chair, while Cath King has agreed to continue as AGNC treasurer. Anita Bruce will take over from Oonagh Claber as the new AGNC secretary. Laura Boyes and Liwsi-Kim Protheroe-Davies will continue on the committee, as will our co-opted trainee and new genetic counsellor representative, Sarah Wilcox.

As Mark had served his full six years on the AGNC committee this left a gap for a new member. The AGNC held their first electronic committee election in March / April 2012 and we would like to congratulate Donna McBride on her success and welcome her as our new committee member.

Anita Bruce has been webmaster for the last five years and has decided that it is now time to step down as AGNC webmaster. Liwsi-Kim Protheroe-Davies has kindly agreed to take this role on. The continuing development of the AGNC website as part of the revamp of the wider BSHG website is too big a task for one individual. We have therefore formed new working group to drive this forward. Anita Bruce and Liwsi-Kim Protheroe-Davies have agreed to lead this group with other members from the wider AGNC membership.

AGNC Spring Conference 2013 Durham

The 2013 AGNC spring conference is to be held at Collingwood College on 15th/16th April. As well as presentations from the membership the programme will include presentations the membership from the Newcastle Neuromuscular genetics team on the muscular dystrophies - current research and treatment; an overview of Cleft syndromes including a parent perspective, and 4d prenatal scanning; Mitochondrial disease and family experience of families with unknown syndromes in genetics. We also hope to hold an interactive session about the changing role of genetic counsellor in the light of the new technology in genetics.

Joint meeting of Lead Genetic Counsellors

Following on from the success of last year the AGNC committee has arranged a meeting for November 7th at Great Ormond Street Hospital London. The AGNC has extended the invitation to the Genetic Counsellor Registration Board, the Genetic Counsellor Training Panel, the Genetic Counsellor Statutory Regulation Steering Group and all the lead genetic counsellors from around the UK & EIRE. It is envisaged that this meeting will further develop the cohesiveness of the Genetic Counsellors working together whilst promoting quality benchmarking across the workforce, this year it is hoped that the AGNC Job Plans will be audited as well as exploring current workforce and patient activity across the board.

Further information will hopefully be collected to explore succession planning and measure accurately Registration forecasts.

Other work

The AGNC **travel awards** have been finalised and the website information has been updated.

The updated **'Career Structure'** document for Genetic Counsellors' document has been completed and can be viewed on the website. This important document will require regular

review and updates especially I view of Regulation activity and Registration developments therefore the AGNC plan to form a working group which will include members from the GCRB, Genetic Counsellor Regulation, Training Panel and a member from the original working party.

BSHG Scientific Committee

The AGNC are very excited that Rhona Macleod has agreed to become a member representing the AGNC in preparation for the big launch 2013.

India Collaboration

Following an interesting request from a clinician in India, who wishes to bridge links with UK Genetic Counsellors links are to be explored with a view of interested GC's in the UK.

Carolyn Owen

AGNC Chair