MINUTES

Association of Clinical Genetic Science (ACGS)

Scientific Sub-Committee Meeting 05/11/13

Held on 11.00 to 15:20 in Genetics Meeting Room, Birmingham Women's Hospital

Present:

Dominic	McMullan	Birmingham **(Chair)**	DOMINIC.McMULLAN@bwhct.nhs.uk
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Apologies:

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1 Introductions; who we are, what we do, what we bring

Round table introductions
EV is an academic with a particular interest in molecular cytogenetics
UM is a co-opted member of the Clinical Genetic Speciality Group. This is a panel of Clinical Geneticists with input into major projects, including DDD.
SM is a member of the Fetal Anomaly Screening Programme Advisory Group and a member of the ACGS Quality Sub-committee

2 Minutes of previous meeting

Accepted with following corrections:

1) Point 4d) should read 'Association of Clinical Pathology and UK Cancer Cytogenetics Group'
2) AP9 should read 'closer working'

3 Matters arising

3a) **re: AP1:** MW/DM Lead and date for 2014 Spring Meeting to be confirmed **4a)**

3b) **re: AP2:** NT to discuss enhanced working with UKCCG at their next meeting
AP10: NT Raise ACGS /UKCCG collaboration at UKCCG SG
1) still no date for UKCCG Steering Group meeting
2) topics are mostly progression-related, but there is little molecular content
DM suggested ACGS could provide data to LRF; NT will raise the subject of mutually beneficial collaboration at the next meeting

3c) **re: AP3** DJM Organise ACGS contribution to NCBI Gene Dosage project
1) a Webex tutorial on the use of NCBI's JIRA software was arranged on 12/11/13
2) SSC volunteers have been assigned a gene or region to investigate

3d) **re: AP4** DM has support from the Executive Committee for an R&D Study Day, covering project design and funding streams.
A further study day aimed at Genetic Technologists should be considered in association with the Workforce Sub-committee

3e) **re: AP5** Raise idea of Research Awards with ACGS Executive Committee
DM has established with the EC that Research Awards funded by the ACGS are not feasible at the moment. A discussion followed regarding the possibility of sponsorship
1) SM suggested it is easier to find funding for a project that is in place than to look for funding and then allocate it to a project
2) EV indicated companies need to know what committment scientists can make
3) MW: leadership and R&D are being emphasised by Sue Hill
4) SM: technological advances (eg arrays & NGS) have been major drivers for a greater emphasis on R&D
5) SM: if a research grant could be awarded at a meeting, this would aid recognition of the sub-committee
6) UM suggested asking companies to the R&D Study Day
   a. ACTION POINT: UM - Dom has a list of companies which have supported meetings in the past and could be approached for research funding (£10k)
   b. We need a contact in the Workforce Development SC to act as liaison for the R&D Study Day
7) collaboration & data sharing:
   a. ST: a title could be published on the ACGS website and interested labs can enlist
   b. UM: CRN may offer local backfill to help with data gathering/ writing up
8) the possibility of collaboration outwith the ACGS was raised: **AP1**
   a. KS: many companies are investing in translational technology
   b. EV stressed that academia could act as important partners
   c. KS: CGS could be involved eg an SPR could write-up papers
   d. ACTION POINT: DM to contact Peter Turnpenny, pres CGS, re: collaboration on write-up

AP2

3f) **re: AP6** DM to contact Andrew Devereau to identify problems in data submission/sharing which ACGS could help to address; explore the idea of a survey of consent issues for submission of information to databases across theTrusts. Explore formation of working group to take forward

3g) **re: AP7:** ST to investigate UK participation in ENIGMA
ST gave some background on ENIGMA BRCA1 & BRCA2 database
There are 5 separate working groups
- analysis/ database
- clinical
- functional
- pathology
- splicing

The clinicians he works with are entering missense mutations and want to enter pedigree information, but he is not sure what the consent process is.
There is no official UK involvement. UK VUS's should be entered in DMuDB along with pedigree info.

3h) **re: AP8:** IS to Explore collecting 16p11.1 microdeletion towards ACGS study **7a)**

3i) **re: AP9:** DM to Contact Rachel Butler about closer working with ACP MolPath group **4b)**

3j) **re: AP11**: DM & MT Re-visit "Forum" idea for website
1) DM: governance issues to be discussed with Communication SC
2) EV: sub-categories could be created, with subscription required before contributions can be posted

ACTION POINT: DM to contact Simon McCulloch about communications & about trialling Twitter at the Spring Meeting **AP3**

3k) **re: AP12:** DM National FRX audit; idea to be raised with ACGS Quality/Audit subcommittee
ACTION POINT SM to gather info about triaging & Guys' document (eg they now do not test anyone under 8 years) **AP4**

3l) **re: AP13:** NT?? Survey of labs planning to introduce array in Haem-Onc
1) NT will get a feel for the current use of/ anticipated value of arrays in leukaemias
2) SM: why are Europe & USA using arrays in this field more than the UK
3) UM: cost is an issue, but SNP arrays can be useful in CLL and MDS with normal karyotypes
4) DM: Lisa Shaffer has published guidelines
5) possible topic for Spring Meeting – maybe Gavin Cuthbert

3m) **re: AP14:** MW Collection of Multicentre data for FH **7b)**

4 ACGS meeting 28th-29TH April in Birmingham

4a) ACGS meeting 28th-29TH April at Austin Court in Birmingham
It has a capacity of 150, with room for some sponsors + limited posters

1) sponsors:
   a. rRom for 8 double stands
   b. Should molpath companies be targeted?
   c. but would these companies be interested in both days, if one was constitutional?
2) posters:
   a. considered a helpful way of allowing contributors to attend meetings
   b. it would be particularly valuable for GTs to be able to present a poster – would a single poster category be possible?
3) other
   a. As this is the first ACGS Meeting, the Chair's Welcome is particularly important
   b. Workforce Development SC should be involved to ensure all parts of the profession can participate
   c. Due to changes in the training scheme; no specific trainee session was considered necessary.
   d. ACTION POINT: All to email DM with ideas for invited speakers and session titles

ACTION POINT: DM to arrange site visit
Conference sub-committee volunteers are SA, MW, ST and DM
MW/DM volunteered lead on content/ theming. **AP5**

AP6

4b) 1) Suggestion from Sandi Deans for a joint MolPath meeting
   a. Bill Newman suggested this could take place at the BSGM
   b. An extensive discussion resulted in the suggestion that a whole day as part of ACGS Spring Meeting would be more likely to attract pathologists
   c. DM, IS & MW can feed back to BSGM on behalf of the ACGS
2) There was a discussion about what areas of pathology should be included (ie solid tumour; leukaemia; microbiology)
   a. There has always been a significant cancer component of the Spring Meeting, and so this should not interfere with UKCCG. The focus of the two meetings tend to be slightly different and there is so much going on that overlap of content should not be a problem
   b. Microbiology is not sufficiently relevant to most genetics labs at the moment to be included on this occasion. However, an overview of mol. pathology (other than cancer) would be valuable
3) DM: Liaise with Rachel Butler regarding interest from the ACP
   a. inviting speakers
   b. reviewing abstracts

5 BSGM 2014

ACTION POINT ALL: emails to DM for ideas on ACGS session content **AP7**

1) IS, MW & DM – on the BSGM SPC
2) New Educational session to be included
3) BSGM will be opened by the 100K Genomes Project update
4) A data sharing session could be included
5) UKGTN / new services session could be included

6 Datasharing

6a) CNV datasharing

Andrew Devereau is aware of problems in data submission/sharing and has been in contact with interested bodies such as the Sanger.

1) We should find out how different Trusts are applying the Coldicot Guidelines
   a. SM suggested using Survey Monkey
   b. DM: DMuDB (?Michael Conway) has a talk about problems they have faced, and this background could be useful in designing the ACGS survey
2) DECIPHER is unique among online resources in having its own consent process
3) KS: Sheffield's policy is they consider any unique mutation to be inherently identifiable with the patient. Cafe Variome (Tony Brookes) may offer a solution
4) ACTION POINT: DM to open a diaglogue with The Council of Caldicott Guardians ahead of their next meeting on Tues 14th January **AP8**

(http://systems.hscic.gov.uk/infogov/links/strategy2011.pdf)
Their '5 year strategy 2011-2016' has nine Strategic Aims, including:

4. Skills: "… false barriers to information sharing are overcome/removed…"
7. Innovation: "…information sharing issues are considered at the design phase of development of new processes…"

5) DM: DECIPHER interested in the possibility of a NHS DECIPHER network, eliminating the need for consent to share between NHS labs. Jawahar Swaminathan has suggested a pilot for a few labs could be arranged in the near future.
6) DECIPHER is now accepting sequence data, and Andrew Devereau has indicated DmuDB may merge with DECIPHER
7) National Data Sharing Meeting could be included at the BSGM, as this concerns not just labs but counsellors and consultants.
   a. ACTION POINT: DM to continue conversation with Andrew Devereau in advance of data-sharing symposium. **AP9**
   b. Representation from patient groups at the symposium
   c. Ideally the same day as the Great Debate
8) DM: all the database are linked, and a system of unique identifiers should be implemented
9) In Holland, consent is assumed at sample taking.

6b) Consent issues; Working Group formation

a. Datasharing study-day; public and private solutions

7 Data collection

7a) 16 p11.2 deletions

1) ACTION POINT: IS & SM to create list of regions (eg in Google Spreadsheets) to be included in a survey of labs eg by Survey Monkey **AP10**
    a. 15q bkpts 1-2
    b. 1q21

7b) VOUS in FH

1) MW presented a preview of Familial Hypercolesterolemia data
2) Average cost of cascade testing is £75 (vs >£300 for a full screen)
3) British Heart Foundation has assigned £1.5m for cascade testing by nurses
4) ~10% of referrals have a VUS, but many of these could be re-classified
5) 7 labs carry out testing and 6/7 are collaborating to address the issue of VOUS
6) DMuDB does not currently have the appropriate ontology for the relevant phenotypes

However, Steve Humphries (UCL) has locus specific databases for LDLR, LDLRAP and PCSK9 (http://www.ucl.ac.uk/fh) which could be updated with clinical outcome and VUS follow-up data

7c) DMD deletions

ACTION POINT UM: pick up issue of incidental DMD deletions nationally **AP11**

7d) Cancer risk CNVs

ACTION POINT Approach Morag Collinson & Birmingham Consultant Emma Woodward (CGG) to examine gathering incidental cancer risk findings, esp known dominants vs uncertain risk (eg BRCA deletions) **AP12**

8 AOCB

DM: development of array design / filtering for prenatal diagnosis

1) Joint Committee for Medical Genetics will meet in February and may make a contribution to the discussion

UM: there is a desperate need for PN array BPG (contact Diane Wellesley, JCMG)

CT: DDD gene list

1) CNVs – FISH probe provided to lab for diagnostic confirmation
2) Point mutations will require bespoke tests
3) Google spreadsheet could be created, merging the UKGTN and DDDG2P gene-lists
    a. labs could then populate what amplicons they can carry out as send-away tests for other labs
4) CRLN provides only £130 for follow-up

9 Date of next meeting

In approximately 3 months' time (date TBC)

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7 Action Points

	Lead	Action Points	X-ref
AP1	UM	DM has a list of companies which have supported meetings in the past and could be approached for research funding (£10k)	**3e)**
AP2	DM	Contact Peter Turnpenny, CGS president, re: collaboration on write-up	**3e)**
AP3	DM	Contact Simon McCulloch about communications & about trialling Twitter at the Spring Meeting	**3j)**
AP4	SM	To gather info about triaging & Guys' document (eg they now do not test anyone under 8 years)	**3k)**
AP5	DM	Arrange visit to Austin Court	**4a)**
AP6	All	Email DM with ideas for invited speakers and session titles for Spring Meeting	**4a)**
AP7	All	Emails to DM for ideas on ACGS session content at BSGM	**5**
AP8	DM	Open a diaglogue with The Council of Caldicott Guardians	**6a)**
AP9	DM	Continue conversation with Andrew Devereau in advance of data-sharing symposium.	**6a)**
AP10	IS & SM	Create list of regions (eg in Google Spreadsheets) to be included in a survey of labs eg by Survey Monkey	**7a)**
AP11	UM	pick up DMD deletions	**7c)**
AP12	DM	Approach Morag Collinson Morag + Birmingham Consultant Emma Woodward (CGG) to pick up incidental cancer CNV risk findings, esp known dominants vs uncertain risk (eg BRCA deletions)	**7d)**