Association of Clinical Genetic Science (ACGS)

Scientific Sub-Committee Meeting 10/07/13

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

Present:
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Carolyn Tysoe(CT) Exeter carolyn.tysoe@nhs.net

1 Introductions; who we are, what we do, what we bring
Round table introductions

2 Appointment of Secretary
Gordon Hislop

3 Terms of Reference (i); (presented and agreed)
3a) Membership
1) Committee
a) see above
b) further members may be co-opted for specific tasks/ projects
2) Advisors
a) These need not necessarily be actual members of the Scientific SC, or the ACGS. Current co-opted advisors include Carolyn Ogilvie, Matt Hurles, Sian Ellard and Rachel Butler as necessary.
3b) Accountability: membership and report to the Executive Committee of the ACGS

3c) Review;
   - The terms of reference of the Subcommittee and the aims of the group shall be reviewed annually and reported to the ACGS Executive Committee
   - An annual report of the activity of the Subcommittee shall be submitted to the ACGS Chair for the ACGS Annual General Meeting

3d) Working Methods
   1) Quarterly
   2) A full annual meeting will ideally be arranged during a conference, either the BSGM or ACGS Spring Meeting, depending on attendance
   3) Teleconferencing will be used to allow maximum inclusion of SSC members for specific (eg project meetings) where possible

4) Terms of Reference; remit (ii)

Particular discussion was held over future ACGS Conferences

4a) Conferences/meetings
   i. BSGM
   ii. ACGS

   All members contributed to an extensive discussion about whether the ACGS should continue to have a Spring Meeting, or the potential content of this could be rolled fully into extended involvement in the BSGM. Points raised included:

   There was considerable support for retaining a Spring Meeting of some sort. It was felt that, rather than a different host lab each year having to start from scratch, the Meeting would be organised by the Scientific Sub-committee, with the same venue over a period of several years. DM suggested Austin Court, Birmingham, which is frequently used by the NHS as an interim solution for 2014 at least and has a list of available dates in April 2014. MW volunteered lead on content/theming.

   1) Further points:
      a) the BSGM & ESHG meetings could be combined in Edinburgh 2015; there will be no BSGM meeting in 2015
      b) UKGTN are holding a Best Practice/Gene Dossier CNGS study Day on 13th November. This is intended to outline the validation requirements for new NGS gene panels.
      c) UKGTN meeting in London between 29th July and 6th August re: Implementation of NGS and the drive for UKGTN/ACGS initiatives CW to attend on behalf of ACGS if scheduled for 5th or 6th August.  
         AP2
      d) UKCCG has an annual 2-day meeting in Newcastle. NT to discuss closer dialogue between UKCCG and ACGS in the organisation of future meetings.
4b) **Research and Collaboration**

1) DM presented the NCBI gene dosage curation project. This is a working sub-group of ISCA/ ICCG. DM has been a member for one year, and during this time 30% of the original ISCA genes have been removed from the ‘CN significant’ list. Further collaboration with the DDD group is on-going in an attempt to further hone a robust “clinical grade” gene list. DM has suggested to the ISCA group that the UK could increase contribution to this process via the ACGS. KS, IS, GH and AC expressed an interest in involvement in this process. DM to approach ISCA to organise

2) ST suggested the Sub-committee should encourage research within individual labs. This overlaps with remit of the Research & Training Sub-committee. MW suggested a study-day on “de-mystifying new techniques” / RND techniques which may help increase local level research initiatives. KS wondered if there could be an ACGS research fund, similar to that previously held by the ACC

3) **Data-sharing**
   a) There was considerable discussion around the data-sharing by UK labs and the drivers needed to increase contribution. It was felt that a consensus on the consent process was needed as there is a perception that different Trust may be operating differing policies. It was decided that the SSC could have a role in scoping how policies could be employed more consistently

   b) DM discussed a possible model for CNV data-sharing developed by the “Low Land Consortium” using Cartagenia Bench. DECIPHER could also be an alternative vehicle for this is the consent process was relaxed.

   c) The importance of sharing rare findings is exemplified by DMD deletions in non-DMD patients: data from UK labs could be relatively easily be collected to provide strong evidence base for its significance

   d) The ENIGMA Consortium collects BRCA variants from 46 groups in 18 countries, but UK involvement is minimal. ST to determine whether contribution could be encourage.

4) **Incidental findings**
   The recent ACMG statement (Green et al, 2013) was discussed and whether a position statement from ACGS was required. It was felt that this is in the wider remit of the BSGM

5) **Publications**
   Ideas were discussed for ACGS lab data-collection to enable larger series publications. The SSC could provide the network and support for doing this with assistance from expert advisors. Members of the SSC were encouraged to suggest ideas and willing co-opted colleagues prepared to lead on individual projects. IS suggested 16p11.1 microdeletion collection as an example starting point as this is relatively common and represents a reporting challenge.
4d) Special Interest Groups (SIG)
   1) It was considered there that these are expected to emerge organically as the need arises
   2) DM suggested that CT had suggested a networked approach to DDD validation which defines a SIG.
   3) A Molecular Pathology SIG was discussed. This may overlap with efforts of the both Association if Clinical Pathology and UK Cancer Cytogenetic Group (UKCCG). Rachel Butler will be contacted over the ACP and NT will raise this at the next UKCCG meeting in November.

5 Any other business
5a) Matt Hurles had suggested several areas to raise at the SSC with DM
   1) Data-sharing; already discussed above
   2) Sharing detection rate data for diagnostic testing; it was decided that this should be raised with the Quality Subcommittee as data collection is now within their remit.
   3) Increasing phenotypic data to accompany a referral
   4) The potential for assigning GUIDs (Globally Unique IDs to variants to reduce double counting in databases)

5b) ACGS web-site; Scientific Sub-committee portal
   1) Open invitation for ideas from all ACGS members to contribute
   2) Ideas included flagging ACGS lab publications; lists of databases; database synopses/review
   3) MT suggested a forum for exchange of technical & interpretative ideas and practice. DM raised the issue of governance; to be taken further with the ACGS Communication Committee

5c) IS raised the question of how much time a Clinical Scientist should be able to allocate to research. To be considered in consultation with the Workforce & Development Subcommittee (relevant to 4b 2) and AP4 above/below)

5d) Genetic Technologist representation; noted that GT representation was lacking from this Subcommittee and needs to be encouraged.

5e) National FRAX data; can this be collected and be used to illustrate shifting in funding. This has already been employed at Guys and could be used as a national approach; needs to be raised with the Quality Subcommittee.

5g) Genotyping: targeted vs untargeted discussed: DW and KS used the example of Sanger vs pyrosequencing for EGFR – pyro is easier to report, but there is no discovery aspect to the test. How are novel mutations & polymorphisms to be identified?

5h) Collection of haematological malignancy array data: currently no clear pattern of how arrays will be implemented, and a survey of where they have provided useful information may help to guide policy

5i) MW suggested collating multi-centre data on Familial Hypercholesterolemia
   • most common cause of heart disease in the UK
   • around 15% of those tested have a VOUS – some of these could be classified as causal or polymorphic from the results of the study
### Action Points

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<th>Lead</th>
<th>Action Points</th>
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<td>AP1</td>
<td>MW/DM Lead and date for 2014 Spring Meeting to be confirmed</td>
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<td>AP2</td>
<td>NT To discuss enhanced working with UKCCG at their next meeting</td>
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<td>AP3</td>
<td>DJM Organise ACGS contribution to NCBI Gene Dosage project</td>
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<td>AP4</td>
<td>DM Discuss with Workforce SC &amp; Executive idea of R&amp;D Study Day</td>
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<td>AP5</td>
<td>DM Raise idea of Research Awards with ACGS EC</td>
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<td>AP6</td>
<td>DM + HS 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</td>
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<td>AP7</td>
<td>ST To investigate UK participation in ENIGMA</td>
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<td>AP8</td>
<td>IS Explore collecting 16p11.1 microdeletion towards ACGS study</td>
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<td>AP9</td>
<td>DM Contact Rachel Butler about closing working with ACP MolPath group</td>
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<td>AP10</td>
<td>NT Raise ACGS /UKCCG collaboration at UKCCG SG</td>
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<td>AP11</td>
<td>DM + MT Re-visit “Forum” idea for website</td>
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<td>AP12</td>
<td>DM National FRX audit; idea to be raised with ACGS Quality/Audit subcommittee</td>
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<td>AP13</td>
<td>NT?? Survey of labs planning to introduce array in Haem-Onc</td>
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<td>AP14</td>
<td>MW Collection of Multicentre data for FH</td>
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