Association for Clinical Genetic Science
Part of the British Society for Genetic Medicine

Feedback from Joint Committee for Genomic Medicine meeting 20th May 2015

Genomic Medicine Centres (GMC)
Cancer recruitment organisation starting now for those GMCs that are already recruiting for rare diseases aiming for August start. Aim for whole project to be complete in Sep 2017 with final recruitment in July 2017.

Another round of EQA for DNA extraction (not sure whether blood, FFPE or both) had been started and that they were also considering taking into consideration performance on the pilot as a more absolute measure of ability to do DNA extraction to a standard that is suitable for WGS.

Open Clinica for data submission preferred mechanism but not ready yet.

GeCIPs approved are: 9 cancer, 12 rare disease, 7 functional, 1 ethics and will be another call for further GeCIPs.

Cancer GeCIPs include Prostate, lung, breast, ovarian, colorectal, haem malig. Definitive list of rare diseases for volunteers on GEL website and second tier under consideration on NHS networks. Indication of allocation of genotypes for each area and reserve available for further applications. There is discussion regarding exploration of co-funding model to support additional genomes eg LLR. Numbers can be utilised potentially to get further funding. Event on 16th June.

Issue of research agenda vs Clinical agenda (as opposed to clinical need) Pathogen side of 100,000 genomes - no info

Rare diseases register. Working on both GEL register and the Congenital anomalies register - all queries to Tom Fowler. Pick up DDD (eg multisystem disorders not eligible at the moment). Discussions regarding those without an answer from DDD - should be able to go into the dysmorphology workstreams. Difficult sometimes to put a child, particularly in early years, into a defined category and there may be an issue for undefined children getting them into the workstream.

Prenatal microarrays document circulated.

JCGM did not formally respond to consultation re designation as covered by other bodies and organisations

Revised role for Clinical Genetics under discussion/consultation.

RAPID
Reviewed and exec summary to be released. Anne Mackie and National Screening Committee (NSC) have commissioned economic assessment. Will be making a decision in the summer and then will go out to 3 month public consultation. Example of excellent translation.

Lab reconfiguration Colin Pavelin was positive regarding how tender process is going. Colin will investigate what information can be released.

Update on Rare disorders register. 1st meeting of the group. 4 task and finish groups (connectivity and interoperability, data quality, +2 others). Colin to email list. Terms of Reference being agreed. Colin Pavelin and Edmund Jessop co-chairing. Re Con and Interop trying to build into other initiatives. Focussed on worries of Big Data. Rare disorders that are not genetic, the DDD question, etc. National Information Board supposed to oversee.

HEE have given budget to take forward **Genomics in Mainstream Medicine.** HI advisory board supported specialist clinicians become much more aware of importance. 12 clinical champions from wide range of specialties. Template describing how genetics was relevant to own specialty gastro, nephrology, dermatology, respiratory, hearing and vision - up on website (HEE). PHGF reported had discussion with BMJ and BMJ Learning which is a fantastic resource. Want to develop a suite of resources which may be marketable.

Detailed discussion regarding **early diagnosis** from newborn screening through to adulthood but with emphasis on the difference between symptomatic and asymptomatic individuals. Investigating the child with Developmental delay is an example. Hilary Burton very supportive.

Genetic Alliance Report from Alastair that patients wish to be told regarding developing information, not just the information that is definite. Importance emphasised of Genetic counselling either by Clinical Genetics or mainstreamed.

UKGTN 3 exomes are in application for Gene Dossiers. Fiona MacDonald is the new Scientific Lead with Becky Treacy to help.

ACGS working on workforce issues and supporting the labs through designation.