Subject: [Test] GeCIP Newsletter #3 - April 2016
From: Genomics England <chiefscientist@genomicsengland.co.uk>
Date: 29/04/16 16:23
To: Simon Thompson <simon.thompson@genomicsengland.co.uk>

"Hi all - final checks please before we send this out.
Lisa" — Lisa Dinh

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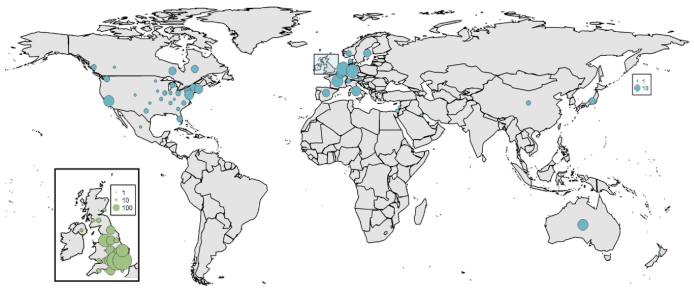
Genomics England Clinical Interpretation Partnership Newsletter - **April 2016**

Clinical Interpretation Partnership Newsletter

View this email in your browser

Dear << Test First Name >>,

Welcome to the April GeCIP newsletter, we hope you'll be interested to read the progress that has been made of the last couple of months. The GeCIP community is now more than 2,300 strong and is spread across 24 countries.



For members receiving the newsletter for the first time, please note that our GeCIP webpages have key information on current activities.

Before going much further we ask that you check the details we have on file for you and update them if they're wrong. We have the following details for you:

Full name: << Test First Name >> << Test Last Name >>
Primary institution:
Email address (must be from your primary institution): simon.thompson@genomicsengland.co.uk
Mobile number (required to register for data access):
Orcid ID number:
If you don't have an Orcid ID, please register for one at www.orcid.org
You can update your details at http://genomicsengland.us3.list-manage2.com/profile?u=75c0225e5d53262524cc4b788&id=7e2419b5c6&e=

We will need **complete details** before we can begin the process of giving you access to the data. Please pay particular attention to your email address, it must be an institutional address (i.e. we cannot accept gmail or hotmail addresses) and the institution with which your email is registered is the one which will be asked to sign the Participation Agreement.

Please note that unless you inform us directly, we will assume that you are happy for us to forward your email address to our partner organisations, for example we may forward your details on to your local Genomic Medicine Centre who may contact you for assistance in driving up recruitment for your domain.

In this newsletter

(Click to jump to each section)

For something different...

'Socialising the Genome' is an engagement project funded by Genomics England, the Wellcome Trust and the Wellcome Trust Sanger Institute. The aim is to help people start a conversation about genomics. Find out more on our site.

Update from the Chief Scientist - Prof Mark Caulfield

Our first GeCIP Steering Committee meeting was successfully held in March and attended by the majority of domain leads. Please refer to the attached minutes and slides for further details. One of the key updates given included the GeCIP publication embargo period which has been extended to 9 months. The prior intent was to offer GeCIP domains 6 months exclusive access to their subset of the data in recognition of their work in developing and supporting their part of the programme. However, for meaningful analysis in many cases each domain may need access to a much broader dataset. Therefore, we proposed to give access across the whole data set to all GeCIP domains and use a publication embargo to protect the primary GeCIP domain who has registered their intent to publish. GeCIP researchers will be asked to register their projects in a 'research registry'. This will give them a 9 month protected period where other domains cannot submit a publication on the same data.

A broad overview of the Fair Usage policy was also given. Many of the GeCIP detailed research plans submitted so far propose activities which are computationally intensive and potentially overlapping with in-house activities and/or those proposed from other GeCIP domains. As a result, we are working on a fair usage policy which will further define resources available to researchers. It is envisaged that each GeCIP domain will be allocated a certain amount of compute power and storage and will have to provide their own funding if research activities exceed this allocation. It is also anticipated that unused computing power may be reallocated within the research community. Over 5,000 participants have been enrolled in the main programme and we will aim to bring the GeCIP domains into the data embassy in a staged way from the end of the second quarter 2016.

Moving forward, it was decided to break into 3 GeCIP steering sub-committees encompassing the Cancer, Cross-Cutting, and Rare Disease arms of the programme. The Cancer and Rare Disease sub-committees will be merged with the existing respective working groups whereas members of the Cross-Cutting sub-committee will retain the option of also attending the above meetings, if relevant to them. Domain representatives who will sit with patients and funders on the GeCIP Board have also now been elected and include the following:

- **Cancer:** Prof Anna Schuh and Prof Charles Swanton (joint membership)
- **Cross-cutting:** Prof Colin Cooper
- **Rare Disease:** Prof Henry Houlden

Professor Mark Caulfield, Chief Scientist

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Current activities - important actions

Rare Disease domains:

- Review Rare Disease gene panels on PanelApp.
- Help with recruitment via engagement with your domain's GMC representative and/or direct contact with relevant GMCs.
- Nominate new disorders

Rare Disease & Cross-cutting domains:

- Submit your detailed research plans.

These are needed before GeCIP domains can access the data. We have now received 14 detailed research plans, 8 from rare disease and 6 from cross-cutting GeCIP domains. We will give detailed feedback to the relevant domains in the next few weeks. Cancer domains are not being asked to submit detailed research plans at the moment, in view of the recent initiation of the cancer main programme.

All domains:

- Tell us if your domain is applying for funding.
- Tell us about your education and training capacity and needs, ahead of the Education and Training domain meeting. Deadline for completely this survey: **Thursday 19th May**.

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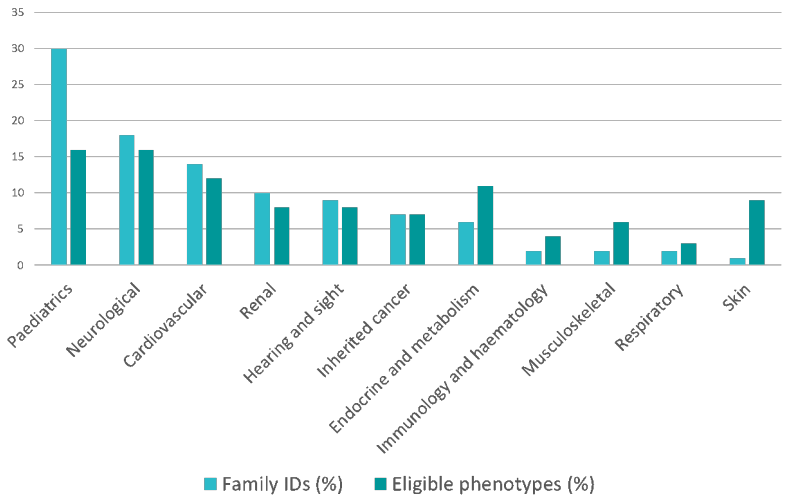
Rare disease update

As a result of your ongoing disease nominations, we now have a total of 180 approved rare diseases. These are mapped onto individual GeCIP domains and can be viewed here.

Following a recent communication with the GMC Clinical Directors, we strongly encourage **GeCIP members to engage with their** domain's GMC representative and help increase recruitment numbers. Please contact Matina Prapa if you would like to discuss this further.

The adjacent graph shows the contribution of rare disease domains to current recruitment numbers (in light green) alongside the proportion of eligible disorders per domain (in dark green).



Over the past few months we have been reviewing the disease-specific data models (HPO terms and clinical tests for each disease) for rare disease. These models define

the data that is requested for registered participants. The overall aim has been to make them as simple, clear and specific as possible, by achieving a balance between too much and too little detail. We started with 26 disorders that account for the most commonly recruited disorders.

We are developing a guidance document for this process with the input of our Rare Disease Clinical Data Working Group and Damian Smedley who has recently joined Genomics England. This will include the numbers of HPO terms in each model (typically 20-40), measures of the specificity and redundancy of the terms and details of preferred terms for phenotypes shared across the programme (for example intellectual disability). A particular focus will be on those models that are very large, for example >100 terms. Over the coming 2-3 months, we will be reviewing the remaining disorders according to this approach. We will be contacting the relevant GeCIP for each disorder for input in the process and look forward to working with you on this.

US Food and Drug Administration – Open Call for Grant Applications
It may be of interest that the US Food and Drug Administration (FDA) has an open grant call for $2m for studies investigating the natural history of rare diseases. The grants are designed to help characterise the natural history of rare diseases, identify subpopulations, and develop and/or validate clinical outcome measures, biomarkers and companion diagnostics.

Grants are available to any foreign or domestic, public or private, for-profit or non-profit entity. For more information see the FDA website.

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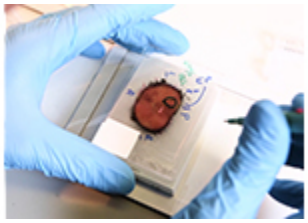
Cancer update



The cancer main programme began in mid-February. A number of GMCs have transitioned from collecting samples under the implementation initiation phase (IIP), and have met the various prerequisites to go 'live' and collect samples under the main programme.

Currently eligible cancers are breast, colorectal, ovarian, lung, prostate, renal cancers and sarcoma. Eligibility for these tumour groups is broad in terms of subtype/grade/stage, although we require that patients have not had chemo-, radio- or hormonal therapy prior to sample acquisition. The Scientific Advisory Committee has agreed to include a number of new tumour types and Genomics England is now working with NHSE to incorporate these into the 100,000 Genomes Project. These include: brain tumours, melanoma, upper GI cancers, bladder cancer, endometrial cancer and testicular cancer. We are also working with NHSE on the recruitment structures by which we can best include childhood cancers and haematological malignancies.

Samples acquired at resection or biopsy are currently eligible. We have developed structures by which material routinely acquired at **diagnostic** biopsy can also be included in the programme: these are under consultation but will be implemented soon. We have also been working with the tumour type leads to

develop some delineated studies involving longitudinal sampling, multi-region biopsying and co-recruitment to research studies/clinical trials, which will be rolled out alongside the more general recruitment.

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Cross-cutting update

Health economics domain
On the 20th January, the Health Economics domain, in collaboration with health economists at the University of Manchester and the PHG Foundation in Cambridge, organised their first workshop. It was held at The Wellcome Trust, London and supported by funding from the National Institute for Health Research. The main aims of the meeting were to provide an overview of the research aims for the domain, to inform non-experts on methods used and finally to allow for exchange of ideas and provide a forum to explore collaboration potential.

Challenges and opportunities for the generation of health economic evidence within the 100,000 Genomes Project were evaluated. The event was well attended by a multidisciplinary group of health economists, scientists, and clinicians representing GeCIP domains and GMCs, as well as representatives from NICE and Genomics England.

Multi -omic samples
Following review of the submitted detailed research plans, a number of domains have been identified which are interested in either using –omics data or developing methods and tools to allow –omics analyses. Please see the website for more details on -omics data.

The best way forward to achieve this is to initiate a working group which will bring together experts from multiple GeCIP domains who can work with Genomics England on design and execution of the **–omics pilot project**, which was put forward at the GeCIP Steering Committee Meeting on the 29th March 2016. For more information, contact Kate Witkowska.

Education and training domain
The education and training domain, led by Health Education England (HEE), is planning a meeting on the 7th June 2016 to establish an operating model for co-ordination of education and training activities. Key staff members of HEE, Genomics England, and NHSE as well as GeCIP domain education and training representatives will be invited to attend. In preparation for this meeting, we are inviting all GeCIP members to complete a survey by the **19th May** and tell us more about their education and training capacity and needs.

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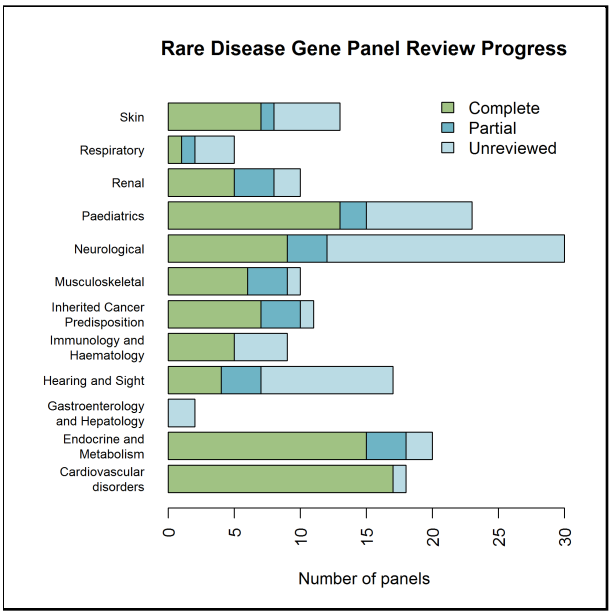
PanelApp - rare disease gene panel review

Thank you to those of you who have already contributed reviews of genes on PanelApp. At present, more than 300 individuals are registered as reviewers out of which around 40% are GeCIP members. Out of a total of 169 rare disease gene panels, 65% has been reviewed so far. Ideally, we would like to have multiple reviewers per panel in order to establish a consensus diagnostic-grade list of genes for each disorder included in the Rare Disease Main Programme.



The adjacent chart shows the progress in PanelApp review across the different rare disease GeCIP domains, and highlights the wide range in the degree of gene panel review completion.

The rare disease interpretation pipeline requires that gene panels have had sufficient expert review before they can be used. **Where panels have had very few (or no) reviews, movement of participants with that disease through the pipeline will be delayed. This will delay availability of the data for your research.** Please therefore review the panels relevant to your domain (summarised in the figure) as soon as possible to avoid delay of data for diagnostic feedback and for your research.

Please see the attached file for user instructions as well as a list of un-reviewed panels requiring your immediate attention.

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Upcoming Events

- HEE Education and Training domain GeCIP event - 7th June
- Curating the Clinical Genome Conference - 22-24th June

If you are planning any meetings within your domain, please contact your GeCIP scientific coordinator:

Rare disease domains - Matina Prapa

Cancer domains - Simon Thompson

Cross-cutting domains - Kate Witkowska

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Next Newsletter

Newseltters are sent every 2-3 months. The next newsletter will include:

- Details on the Publication Committee function following its 1st meeting on the 26th of April
- Updates on data access

Thank you for reading. If you have any questions please contact us.

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