NHS South West NHS Genomic Medicine Centre

100,000 Genomes Project Rare Disease Recruitment

NOV 2017

At a glance

- Welcome
- Testing prior to recruitment
- Highlight on: Cardiology
- Clinical Genetics or 100k?
- Project Update
- Champions
- Patient and Public Involvement
- Education and Training recruiting team

Useful Links:

- Human Phenotype Ontology— www.human-phenotype-ontology.org
- Encephalopathy panel http://www.labs.gosh.nhs.uk/media/529514/eiee_v6.pdf
- Patients can be referred by emailing the project team at rde-tr.SWGMC@nhs.net or send a copy of the clinic letter to the project team at:

The South West NHS Genomic Medicine Centre (GMC) Level 3, RILD Centre, Barrack Road, Exeter EX2 5DW

Welcome

1875 participants from 815 families have been referred into the 100,000 Genomes Project in the Southwest as at the end of November. The project continues for another year before WGS becomes a commissioned test for some diseases. We look forward to extending access to patients at all 7 acute trusts free of charge in the coming months.

Thank you for your continued support.

Investigation of cases prior to recruitment: note in particular to paediatricians

The goal of the project is to achieve a molecular genetic diagnosis. If the patient already has one, then he or she is not eligible! For those patients who have not (yet) received a molecular diagnosis, a possible conflict arises between continuing to investigate by 'traditional' methods (imaging, metabolic screen, neurophysiology), on the one hand, and referring to the 100,000 Genomes Project, on the other. Both approaches contain an element of risk. The 'traditional' approach is the one which the Project is seeking to supersede because it can lead to long delays in diagnosis; nevertheless, it has to be accepted that currently, the 100,000 Genomes Project approach is also currently slow (see Project Results, below). Referral directly into the project contains risk because there is a danger that the clinician has referred in a patient with a condition currently diagnosable by traditional means, where the diagnosis could be obtained sooner. Clearly there is no right or wrong answer here. We wish to highlight that return of results through the Project is not as quick as envisaged at the outset, and there is danger that diagnosable patients are left waiting.

Highlight on: Cardiology

Some of the recruitment categories are more likely to be fruitful than others, reflecting rarity or stringency of eligibility criteria. The following are likely to be 'best bets':

Hypertrophic Cardiomyopathy. A patient with a confirmed diagnosis who is under the age of 40 years is eligible for recruitment on his or her own. Patients over 40 years should have a family history. Peninsula guidelines state that genetic testing in these families should be carried out in the Clinical Genetics service. There will be patients who have been through our service, who have undergone genetic testing and been discharged back to cardiology follow-up. These patients are of course recruitable by cardiologists. Please look out for them!

Dilated thoracic aortic aneurysm disease. Again, patients with a thoracic aortopathy under 50 years of age are recruitable as 'singletons'. Usually though not always they will have been through our service for genetic testing. But again they may have been discharged back to cardiology for follow up. They are eligible.

Congenital heart disease. The type of structural malformation is not specified. Atrial and ventricular septal defects of course qualify, along with other more complex ones. If the CHD is isolated (no additional or syndromic features), then either parental consanguinity or a positive family history is required. If syndromic, 'singleton' recruitment is possible.

Other conditions are rarer and recruitment criteria more stringent: familial dilated cardiomyopathy, Brugada, Long QT, Idiopathic VF, Unexplained sudden death in the young

For full details go to: https://www.genomicsengland.co.uk/information-for-gmc-staff/rare-disease-documents/

Contacts

Genomic Champions:

Dr Lucy Leeman
Immunology
Plymouth
lucy.leeman@nhs.net

Dr Kash Patel
Diabetes and
Endocrinology
Exeter
kashyap.patel1@nhs.net

SWGMC Team:

Dr Charles Shaw-Smith
Rare Disease Lead
charles.shaw-smith@nhs.net
01392 405737

Ana Juett
Project Manager
Rare Disease
ana.juett@nhs.net
01392 408565

Mary Tamplin
Genomic Nurse
mary.tamplin@nhs.net

Amanda Skinner
Genomic Nurse
amanda.skinner@nhs.net

Dr Steven Johnson
Project Manager
steven.johnson@nhs.net
01392 408177

Prof Sian Ellard
Programme Lead
sian.ellard@nhs.net

Phenotyping

Thank you to everyone for responding with phenotyping and phenotyping questions. We have not sent out requests for phenotyping on all your patients in one go but will be sending regular requests. If you would prefer to have a list of all your patients for phenotyping, please contact the project office team. Information on phenotype terms can be found Human Phenotype Ontology . Support can be provided by the project teams.

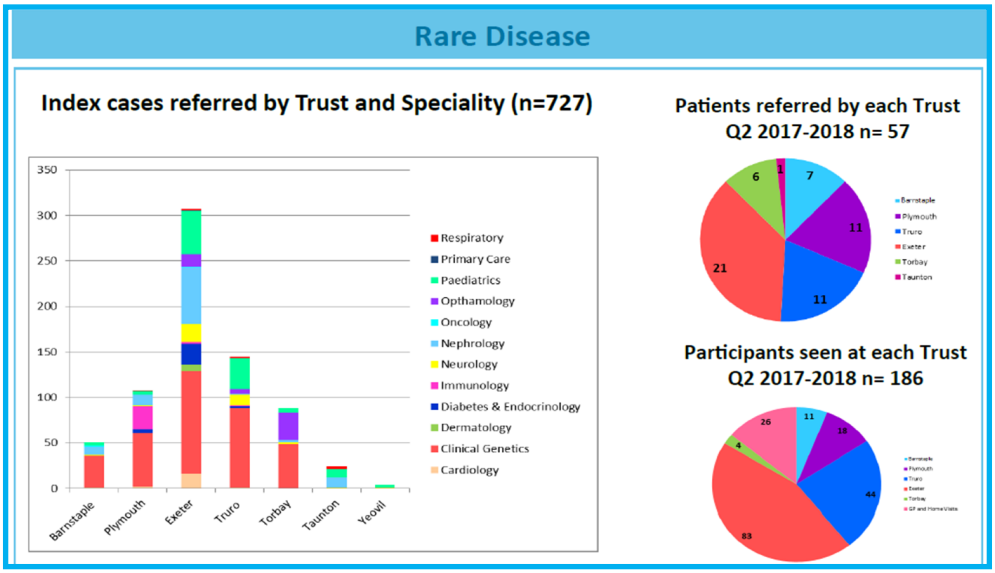
Clinical Genetics opinion or recruitment to the 100,000 Genomes Project?

We receive some referrals from which it is not clear which of the following is required: an opinion from the Clinical Genetics service; or, referral into the 100,000 Genomes Project. Obviously, these are two quite different things. It can be difficult to know which is the right pathway, but these are some pointers towards recruitment to the 100,000 Genomes Project:

1) The patient/family have already been reviewed by a Clinical Geneticist
2) Those genetic investigations which are available within the NHS have already been carried out
3) A diagnosis is not required within an urgent timeframe

Please see also above: Investigation of cases prior to recruitment. If in doubt, a quick email to the project office rde-tr.swgmc@nhs.net) or Rare Disease lead charles.shaw-smith@nhs.net

Quarterly Review: Update referrals and recruitment Q2



If you have any cases suitable for recruitment not yet invited, this would be an excellent moment to take action. It is important that we try to maintain or increase our momentum.

Thank you to our Genomic Champions

Our Genomic Champions have returned to their regular jobs. The work they have undertaken has been vital to share information and knowledge about this new and progressive diagnostic tool. The team are still very much involved and we hope to see more in house champions evolving as we go forward. Please contact the project team if you would like to become more involved with introducing WGS as a tool for your specialty.

Patient and Public Involvement

The patient public forum met in October. The group looked considered how consent might look for genomic testing in the future and supported the development of information to be displayed in GP practice. For full information on outcomes of PPI work please contact Ana Juett 01392 408565 for more information

Training and Education

Any staff wishing to develop consent training skills for free can contact us on rde-tr.SWGMC@nhs.net for more information.

For information about funding for higher education courses: *https://www.genomicseducation.hee.nhs.uk/taught-courses/*

For university applications: *http://www.exeter.ac.uk/postgraduate/taught/medicine/genomicmsc/*