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South West GMC Team

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

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

Prof Sian Ellard
Programme Lead
sian.ellard@nhs.net
01392 408259

Website

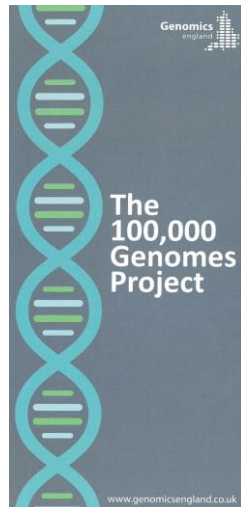
www.swgmc.org

Highlights in this newsletter

Well done! The GMC had its Quarterly Review meeting on Thursday 4th February and we were congratulated on our progress. We would like to pass this message on and to thank everyone for their hard work. Let's keep up the momentum!

- More recruits, please! Recruitment has dipped somewhat over the Christmas/New Year period understandably. This would be an excellent time to contact Steve with some more potential recruits, especially given that it is **Rare Disease Day** at the end of the month!
- Some changes to eligibility criteria for familial colon cancer: see section 3d below.
- Upcoming events including SW GMC launch event- see section 6, below

1. Introductory leaflets



A reminder that Introductory Leaflets for the project are available from the Project Office (Steve Johnson, details opposite).

The leaflets are suitable for having in the outpatient clinic and handing out to patients who express an interest.

They don't say anything about additional findings or about data sharing. Please mention these things to potential recruits who are happy to be contacted by the Project Office.

Timetable for 'onboarding' of Local Delivery Partners (LDPs)

The LDPs are the partner hospital Trusts in the Peninsula and Somerset:

Derriford Hospital, Plymouth

Royal Cornwall Hospital, Truro

North Devon District Hospital, Barnstaple

Torbay Hospital

Musgrove Park Hospital, Taunton

Yeovil District Hospital

LDPs are 'on board' when patients/families can be recruited, and the samples processed, locally in that hospital

Projected timetable for onboarding:

Derriford, RCH Truro: by February 29th, 2016

MPH, Taunton: soon.

NDDH, Yeovil DH, Torbay Hospital by July 31st 2016

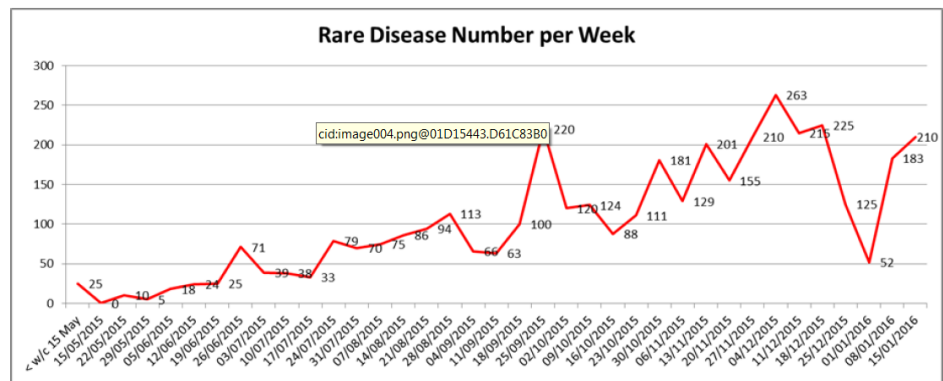
2. Recruitment

a. Recruitment so far

The graph on the next page shows recruitment across the whole country, up to mid-January. Probably the main thing this shows is that the Christmas/New Year period is not a good one for recruitment!

The table shows that the South West is doing well when measured against predicted recruitment.

Rare Diseases: Number of Samples Collected Per Week



GMC	% of Activity YTD against Revised to Q3	% of Activity YTD against Contract to Q3	% of overall GMC Activity (w/e 01/01/16)
West Midlands	86%	37%	12%
West London	53%	24%	2%
Wessex	103%	91%	7%
South West	91%	125%	5%
South London	114%	85%	17%
Oxford	69%	69%	7%
North West Coast	76%	67%	5%
North Thames	71%	57%	22%
North East & North Cumbria	74%	43%	6%
Greater Manchester	91%	91%	10%
East of England	115%	27%	6%

Following the recent dip, this would be a good time to start boosting our numbers again.

If you have any suitable patients sitting in your in-tray, Steve is waiting to hear from you (see below for reminder on how to recruit a patient).

Also worth noting that February 29th, 2016 has been designated Rare Disease Day - this would also be an excellent day to inundate the Project Office!

New disorders open for recruitment

CNS malformations/neurology

Holoprosencephaly
Cerebellar hypoplasia
Joubert syndrome
Rhombencephalosynapsis
Malformations of cortical development
Moyamoya disease
Intracerebral calcification disorders
Inherited white matter disorders

Dermatology

Familial cicatricial alopecia
Peeling skin syndrome

Paediatrics

Cocayne syndrome
Primary microcephaly

Changes to eligibility criteria

Familial colon cancer

A further 9 disorders are awaiting approval.

b. How to recruit a patient

We have tried to make this as simple as possible. An example, the version for paediatricians, is shown on the next page.

We feel that the process is about as straightforward as it could possibly be, and indeed clinicians from different specialties throughout the Peninsula have risen to the occasion and recruited dozens of families so far- see the recruitment figures above.

The easiest way to recruit a patient/family is from clinic, but phoning on receipt of a negative molecular test is also an effective option.

Ask for help if needed!

Please remember to communicate as much detail as possible regarding the degree of intellectual disability where applicable as well as any known issue taking blood, or other relevant history that recruiting nurses should know about before the appointment.



100 000 Genomes Project recruitment for paediatricians

This is for a 'trio' family structure, with affected child and unaffected parents. If family structure differs, please contact for advice.

Step 1: Check eligibility

A comprehensive list of eligibility criteria is available via the link below: <http://www.genomicsengland.co.uk/library-and-resources/> (See under "Eligibility statements"/"Rare disease eligibility statements")

Some examples of eligible disorders:

Intellectual disability; Congenital Heart Disease; Congenital anaemias; Non-syndromic hearing loss; Craniosynostosis syndromes

Step 2: Discuss with the family in clinic or by telephone

1. Purpose of study is to try to obtain a molecular genetic diagnosis [NB there must be a diagnostic question]
2. Blood samples are needed from the affected individual and unaffected parent(s) or other affected family members
3. Family members will be offered opportunity to consent to 'secondary' findings
4. Data will be made available in anonymized form to research/commercial bodies (non-negotiable)

Step 3: Notify GMC office (Steve Johnson, Project Manager)

Please email the following information to: rde-tr.GMC@nhs.net

1. Name and NHS number of affected individual, or name and date of birth
2. Names and dates of birth of other individuals who would be recruited (typically the parents)
3. The diagnostic category (Intellectual disability etc)
4. An email address (preferred but not essential) or postal address for the family

The project office will then contact family re consent/sampling appointment

Step 4: Clinical data entry

Once the consent/sampling process has been completed, the recruiting consultant will be asked to complete an online phenotype data entry form. This will include growth parameters, systems examination and dysmorphology. (Help with this will be available)

Please contact us if you have any questions about any aspect of the project.

charles.shaw-smith@nhs.net

T: 5737

sian.ellard@nhs.net

T: 8259

rde-tr.GMC@nhs.net

T: 8177 (Project office)

Recruitment team

Paediatric Research Nurses, Exeter

Su Wilkins
Caroline Harrill
Sue Ward

Paediatric Research Nurses, Truro

Nina Worrin

Genetic Counsellors, Exeter

Anne Searle
Heather Chalinor
Gemma Corbett

Genetic Counsellors, Plymouth

Matilda Bradford
Nicol Lambord

Specialist Registrars

Lettie Rawlins (Clinical Genetics, Exeter)
Rhian Clissold (Renal Medicine, Exeter)
Harriet Aughey (Paediatrics, Truro)

c. Recruitment outside Exeter

See side panel for definitions of LDPs, onboarding, and projected onboarding timetable. There has been significant progress recently so we are optimistic of achieving these timescales.

There are recruitment appointments for patients and families in Derriford Hospital and RCH Cornwall taking place in February. Significant expressions of interest have also been received from Musgrove Park Hospital, Taunton, and Torbay Hospital.

d. New clinical disorders for recruitment/changes to eligibility criteria

There is a steady trickle of new disorders approved for recruitment- see panel opposite. The entry criteria for familial colon cancer have become more stringent: now 4 affected family members needed rather than 3.

3. First results from the 100 000 Genomes Project

There was significant media coverage in January from London: two families received molecular diagnoses as a result of participation in the project, and this was taken up by print and TV media. See [here](#) if you missed it. Also here for an interview with Mark Caulfield from Genomics England

4. Entry of phenotype data

There is progress to report here. The Genie software is now operational within the SWGMC, and data entry is possible. CSS has entered data for two families, and the process is reasonably user-friendly. We would like help with this from consultants who have entered patients into the project, but we will be offering assistance with the process. Further updates shortly.

5. Recruitment team

Joining the recruitment team

The team is open to new members, especially in Torquay and Barnstaple. Team members should be up to date on the following:

1. Good Clinical Practice training- usually offered as part of Trust mandatory training
2. Completion of online consent training provided by Health Education England- the module can be completed in around an hour
3. Face-to-face consent training specifically for 100 000 Genomes Project, provided to date by Charles Shaw-Smith, Rare Disease Lead.

Thanks to the following for commitment to the project:

All members of Peninsula Regional Clinical Genetics Service

Richard Tomlinson, Eleanor Thomas, and all members of the Community Paediatrics team, RDE, Exeter

Coralie Bingham, Rhian Clissold, Renal Medicine, RDE, Exeter

Claire Bethune, Lucy Leeman, Andrew Whyte, Immunology and Allergy, Derriford Hospital, Plymouth

Bijay Vaidya and Andrew Hattersley, Diabetes and Endocrinology, RDE, Exeter

Nick Gutowski, Neurology, RDE, Exeter

Elizabeth Househam, Neurology, Derriford Hospital, Plymouth

Kayal Vijayakumar, Paediatric Neurology, Bristol Children's Hospital

Tony Quinn, Ophthalmology, RDE, Exeter

Carolyn Charman, Naomi Goldstraw, Dermatology, RDE, Exeter

New members this month:

Heather Chaliner and Gemma Corbett, both Genetic Counsellors based at the Regional Genetics Service in Exeter

New consent material

There is now a 'flipchart' available, designed to be used by healthcare professionals during the consent appointment, should they wish to use it. It is not compulsory and it's not designed to be used by participants alone.

It can be used as a prompt during the consent conversation. It includes additional diagrams which can also support explanations of genetic concepts and the project. This has been approved by the Research Ethics Committee and can now be used in any GMC or LDP.

Please feel free to print or let Steve Johnson in the project office know if you would like a printed copy.

6. Some upcoming events/highlights

We are hosting our first South West GMC event to celebrate recruitment of the first 100 families with a rare disease. It will be held in Exeter on **in April or May (date to be confirmed shortly)**. The meeting will be open to patients and families, clinicians and members of the recruitment team, and other interested parties, although there will be some restrictions on numbers. Representatives from Genomics England will be invited to speak. We hope to stage a panel/audience discussion on a theme relating to whole genome sequencing. Further details shortly.

February 29th, 2016 marks the ninth international [Rare Disease Day](#).

From the website: "On and around this day hundreds of patient organisations from countries and regions all over the world will hold awareness-raising activities based on the slogan Join us in making the voice of rare diseases heard."

Please note the Educational Event below, aimed at non-genetics specialists.

UCL
INSTITUTE OF CHILD HEALTH

Introduction to Applied Genomics in the Clinic

Thursday 7th April 2016
Lockin Price Lecture Theatre,
UCL Institute of Child Health

Are you involved in managing or recruiting patients with genetic conditions? This course is an introduction to how Genomics applies to your clinical practice.

Course Objectives
Prof Maria Striano-Gobatto, Professor of Clinical and Molecular Genetics, UCL Institute of Child Health
Dr Sefik Cenkoyun-Gezici, Senior Research Associate, UCL Institute of Child Health
Dr Mike Hubbard, Director of UCL, Genomics and Senior Lecturer, UCL Institute of Child Health

This one-day course is aimed general and specialist paediatricians and physicians, nurses in adult and paediatric medicine and surgery, and genetic nurses and counsellors. We will introduce participants to the cutting-edge genomic technologies, data analysis methods and resources that are increasingly being adopted by NHS laboratories. The course aims to explain how genomic medicine is becoming part of the one-to-one work of clinical laboratories and is based on real clinical cases. Participants will be directly involved in the analysis of genomic information through interactive learning and discussion. No specialist experience of bioinformatics knowledge is assumed.

Places
Early Bird £175 (before 03/03/2016)
Standard £200 (after 04/03/2016)

UCL Events, UCL Institute of Child Health
10 Bedford Way, London WC1E 6BT
Phone: +44 (0)20 7679 4700
Email: events@ucl.ac.uk
Website: www.ucl.ac.uk

8. Thank you!

Thank you to all clinicians who have recruited to the project, and to all members of the recruitment team for their work.