

100,000 GENOMES PROJECT

Summer 2016

Celebrating the first 100 families recruited to the 100,000 Genomes project in the South West.



On 28 June, patients and their families joined with healthcare professionals from across the region to celebrate the project. Opening the day, Professor Sue Hill, Chief Scientific Officer at NHS England, described the aims of the national project and how it will shape future healthcare. Professor Sian Ellard, Director of the South West NHS Genomic Medicine Centre and Professor of Genomic Medicine at the University of Exeter Medical School, reported local progress before two families talked about their experience of the project.

One of those patients is Aidan Scholey. Seven-year-old Aidan is the first member of the Scholey family to show signs of a rare genetic condition, and his mother Maria hopes the 100,000 Genomes Project will help provide some answers.

Education & Training

The University of Exeter are running a Master's in Genomic Medicine, which is FREE in full or as individual modules for CPPD to all NHS healthcare staff. Click [here](#) for more details on the course and [here](#) for HEE funding information.

We have appointed two Education and Training Leads to help local NHS healthcare professionals to up skill in genomics, Leigh Jackson and Marie Taylor

For assistance with all education and training issues, please contact Leigh or Marie at

rde-tr.SWGMCEducationAndTraining@nhs.net



Marie Taylor and Leigh Jackson

Focus on: Cancer Recruitment

The first patient was recruited in Exeter on 29th January 2016 and as of 30th June, 20 patients with breast, colorectal or bladder cancer have been recruited. We are working with clinical teams at the RD&E to start recruitment of patients with lung, renal and sarcoma cancer next.

A number of additional cancer types have been added to the list of eligible cancers; adult glioma, brain tumours, renal cancer, bladder cancer, endometrial cancer, melanoma, Upper GI tumours and testicular cancer.

Discussions to enable Local Delivery Partners (LDPs) in other Trusts to participate in the cancer arm of the project have now started. Very positive discussions have taken place with the Cancer Nurse Specialists (CNS) in Exeter and Plymouth regarding their involvement in the project by providing information to eligible patients and taking consent from participants who wish to participate. Many thanks to Tina Grose and Sian Dennison for facilitating these extremely important meetings. We are also keen to facilitate the CNS to enrol in the FREE Genomic Medicine modules to support their personal development.



Foundation year doctors hearing about the project in Exeter



Dr Jiong Wu

Focus on: Rare Disease Recruitment

Our original target was to provide 1000 samples from 400 families by the end of 2017 but we have increased our planned recruitment for patients with rare diseases. The first family was recruited in Exeter on April 24th 2015 and 266 family referrals have been received from clinical genetics, renal medicine, paediatrics, endocrinology, diabetes, neurology, ophthalmology, dermatology, immunology and cardiology. As of June 30th 368 participants (149 families) have been recruited.

Work continues to engage clinicians outside of Exeter and the Community Paediatrics team at Truro have already made a significant contribution to the project. Dr Charles Shaw-Smith has also delivered presentations at Grand Rounds in Plymouth and Truro to promote engagement.

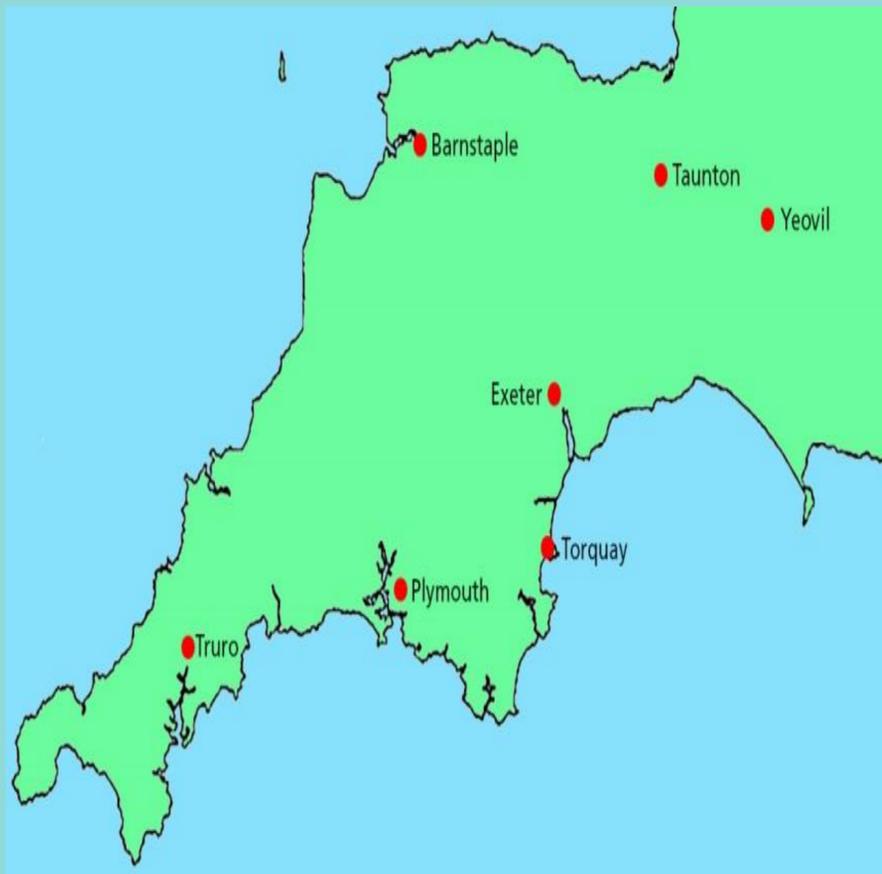


Dr Shaw Smith with Professor Sian Ellard and Rosie Bennyworth

The GMC will be working with clinicians at each of the local delivery partners so that eligible patients can be referred. Interested clinicians in any hospital are encouraged to contact the project office in advance to find out more by emailing rde-tr.gmc@nhs.net. Our regular Rare Disease newsletters and the recruitment 'read me' for recruiting clinicians are available at www.swgmc.org.

Focus on Genomic Champions

We are delighted to welcome our first 3 genomic champions who will be encouraging local healthcare professionals within their specialty to engage with the 100,000 Genomes Project and access the education and training they require.



Dr Kash Patel is based in Exeter and his specialty is Diabetes and Endocrinology



Dr Lucy Leeman is based in Plymouth and her specialty is Immunology



Dr Siying Lin is based in Torbay and her specialty is Ophthalmology

More information on our Champions in the next newsletter!

USEFUL RESOURCES 100,000 GENOMES PROJECT

FOR MORE INFORMATION ABOUT THE PROJECT,
PLEASE CONTACT:

SOUTH WEST NHS GENOMIC MEDICINE CENTRE

EMAIL: RDE-TR.GMC@NHS.NET

TELEPHONE: 01392 408177

Patient and Public Involvement

Sarah Ross, the PPI Lead for the Project left earlier in the summer and we would like to thank Sarah for all of her hard work and dedication in ensuring that patients and the public remain vitally involved in the work moving forward. The South West Academic Health Sciences Network are in the process of recruiting a new lead and updates will continue to be made via this newsletter.