

Central and South Genomic Medicine Service Alliance Transformation Projects

The Central and South Genomic Medicine Service Alliance (CAS GMSA) is delivering a range of transformation projects across our geography, aiming to implement new pathways across a range of specialities and disease areas.

The CAS GMSA spans the West Midlands through central England to the south coast with major centres for genomics in Birmingham, Oxford and Southampton.



CaSR

The CaSR project aims to establish a protocol to distinguish pathogenic mutations of the calcium-sensing receptor (CaSR) gene from non-pathogenic variants.

This will be achieved by establishing a reference-database of CaSR mutations, which will help to distinguish between primary hyperparathyroidism (which usually requires surgery to treat) and familial hypocalciuric hypercalcaemia (which is usually a benign disorder).

To date, key progress on the project has included gateway cloning, which in time will help test the function of proteins. Sanger sequencing is underway to assess the success of the cloning. The library is being developed, with the CaSR project expected to continue into 2023 and beyond.

Equipping Cancer Genetics Services to better meet the needs of transgender patients

For Cancer Genetics Services to provide accurate risk assessment, appropriate recommendations and high quality care of transgender (trans) patients, clinicians need to take into account altered risk profiles due to gender affirming treatments and known barriers to healthcare, as well as displaying cultural competence.

Our project firstly evaluates whether UK Cancer Genetics Services are accessible to, and meeting the needs of, trans patients referred with a family history of cancer. Given existing literature, anecdotal reports and personal experience of Genetic Counsellors within Clinical Genetics Departments, we expect to identify opportunities for improvement, and to define clear standards of care.

Ultimately, the project aims to implement positive changes to Cancer Genetics Services, which will improve accessibility for trans patients and the quality of their care.

The estimated proportion of trans people in the UK is 0.5-1%. Among the cancer consultants and genetic counsellors currently working at Wessex Clinical Genetics Service (WCGS), we have only identified 4 trans patients who have been seen in our service in our collective careers at WCGS.

The project has a 2 year life cycle, and we are currently in phase 3.

The project is actively supported by the UK Cancer Genetics group, with strong engagement across genetics services in our geography.

To find out more – please contact Genetic Counsellor and project lead josephine.giblin@nhs.net



Other projects

Other projects the CAS GMSA are working on include:

- Primary care programme:
 - > South Asian Exome panel
 - > Mendlescan
 - > Geneseize
 - > Breast cancer
 - > Calculating familial cancer risks
- Somatic cancer programme
- Diagnosis and management for haem-onc

Contact us

✉ GMSAAdmin@uhb.nhs.uk

🐦 [@CaS_Genomics](https://twitter.com/CaS_Genomics)

🌐 centralsouthgenomics.nhs.uk/