NIHR BioResource UNIVERSITY OF CAMBRIDGE

We are the NIHR BioResource, bringing people together and leading research

1

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Background

The NIHR BioResource (https://bioresource.nihr.ac.uk/) is at the heart of efforts to improve healthcare and the long-term prevention and treatment of disease. We achieve this by engaging with participants, and collaborating with leading research groups in academia, NHS and industry to rapidly translate findings in the laboratory into benefits for the clinic.

We also grant access to samples, data and resources to tackle healthcare challenges. The BioResource has a decade of experience and a large team in Recruitment & Recall, Sample Management, Ethics & Governance and Informatics.

To engage with the public, we have an active website, circulate newsletters, attend festivals/fairs, invite lay members to join committees, and hold virtual events.



The BioResource recruits at a national scale to 9 programmes covering common diseases, rare diseases, healthy population and COVID-19. There are 13 NIHR BioResource centres around England.



Participants consent to the NIHR BioResource via a programme (**Stage 1**) either electronically (tablet, website) or using a paper form.



Personal details are kept separate from research data. Consent includes access to medical or other health-related records.



Samples (blood or saliva) are collected allowing DNA, serum and plasma to be banked and accessed on demand.



DNA is genotyped using the UK Biobank v2.1 Axiom Array, and if required, we perform whole genome or whole exome sequencing (WGS/WES).



Depending on the programme, demographic, health & lifestyle (H&S) and clinical data is collected to get phenotypes.



Volunteers may be invited to Stage 2 studies (up to 4 times/year face to face and 4 requests/year for online surveys) based on their genotypes or phenotypes.

Researchers can request to use samples in our biobank, data we have collected or recall participants via a Stage 2 study.

Stage 1 Biobanking										
Participants per programme		DNA extracted from 2 sources		Banked aliquots for further use		Genetic data			Phenotype data	
~ Count (A	ug 2021)	Blood	Saliva	Serum	Plasma	Array	WGS	WES	H&L	Clinical
COV	7,600	1		1	~	~			~	~
CYP	9		~						~	
GEN	40,000	~	√few	~	~	~			~	
IBD	34,000	~		~	✓	✓16.5K	√2K	√12K	~	~
IMID	700	1		~	~				~	~
MH	31,000		~			1			~	~

COV - cutting edge research looking at COVID-19

√few

NAFLD

RDC

STR

250

15,000

52,000

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CYP - recruiting under 25s to the new (Aug 2021) Young People's BioResource

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√8K

GEN - participants recruited from the general population IBD - national platform looking at Inflammatory Bowel Disease

IMID - helping research into Immune-Mediated Inflammatory Disease MH - improving our understanding of different Mental Health illnesses

- NAFLD a partnership to research Non-Alcoholic Fatty Liver Disease
- RDC aiming to identify genetic causes of Rare Diseases

STR - working with NHS Blood and Transplant to recruit donors to STRIDES

Case studies

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We responded quickly to help with the COVID-19 pandemic. Within a month of the first national lockdown, working closely with the Cambridge Institute of Therapeutic Immunology & Infectious Disease, samples taken from patients at screening pods and healthcare worker serology screenings (~6000) were stored. The COVID BioResource has expanded to support Stage 2 studies including vaccination (healthy and disease groups) and long COVID clinics. By rapidly providing banked serum or plasma samples to collaborators, key findings were expedited and published:

- 3% of asymptomatic HCWs tested positive for SARS-• CoV-2 (Rivett et al., eLIFE, 2020).
- Understanding the immune responses following vaccination in elderly participants and younger healthcare workers (Collier et al., Nature, 2021).
- Looking at detailed immune phenotyping at multiple time points up to 90 days from symptom onset in patients with progressive COVID-19 compared to those with mild disease (Bergamaschi et al., Immunity, 2021).

Stage 2 Sample Use

Year	Shipn	nents to res	earchers	Number of aliquots shipped			
	DNA	Serum	Plasma	DNA	Serum	Plasma	
2018	11	4	1	2660	1030	673	
2019	17	1	1	21929	1264	1179	
2020	3	11		4763	10017		
2021	2	13	3	5698	944	860	

Samples are stored at a central biorepository (National Biosample Centre) for future use. The BioResource can help researchers choose samples or data of interest based on genotype and/or phenotype either by participant recall or the provision of data and/or samples. Applications are reviewed by the NIHR BioResource Steering Committee. Work so far has led to:

- > 790.000 stored aliquots
- > 60,000 detailed full blood counts for blood donor studies such as STRIDES
- > 128 Stage 2 studies (including 11 shipments related to COVID-19)
- 234 publications to date since 2009 (https://bioresource.nihr.ac.uk/publications/)



The BioResource has been pivotal in recruiting patients, and in some cases their relatives, with a Rare Disease. We work in more than 50 disease areas, including in immunity, neuroscience, haematology, rheumatology, and cardiovascular disease.

- Contributions have been made to the 100,000 Genomes Project, led by Genomics England in partnership with the NHS, to reach the goal of sequencing 100,000 whole genomes from NHS patients.
- Sequencing samples of rare disease patients has led to recognition of patients' individual genomic variant information in clinical decision-making and a genetic diagnosis to 1,138 patients (Turro et al., Nature, 2020).
- Banked samples (2000 plasma aliquots) provided to a Stage 2 study for von Willebrand factor (VWF) level analysis and comparison to genetic data, this is the most common inherited bleeding disorder in the UK.
- Recently, the BioResource team has expanded to include the Rare Disease RNA Phenotyping project to drive forward knowledge at a cellular level.

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