Rare Diseases Research Funding and Support

EUROPLAN II

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UK Strategy For Rare Diseases

The UK is an international leader in research into rare diseases and the UK Strategy for Rare Diseases recognises that a culture of innovation will lead to faster access to evidence based care. The UK Strategy for Rare Diseases recognises that research can make an important contribution to its aims to:

- Increase the evidence base
- Engage and involve patients in research
- Speed up research initiation and delivery
- Improve diagnosis
- Encourage partnerships in developing new treatments
- Coordinate rare disease research

In England, the Department has been working with key organisations, including NHS England, the Health Research Authority, Genomics England Ltd and through National Institute for Health Research (NIHR), to ensure that the Strategy’s commitments related to research are taken forward.
Research Commitments

1. Strengthen the mechanisms and opportunities for meaningful and sustained patient involvement in rare disease service provision and research, recognising patient groups as key partners – including in the development of the four country plans to implement the Strategy.

8. Help patients to contribute to research and other activity related to rare diseases.

31. Look at how the 4 UK countries develop, change or expand information systems to capture, connect and analyse data about clinical and social care pathways.

35. Use portals to connect patients and relatives to enhance research participation and, where appropriate, promote self-enrolment to approved research studies with online consenting, self-reporting and use of social media.

36. Encourage patient groups to get involved with regulatory bodies.

37. Help patient organisations and community engagement events develop more formal partnerships with the NHS research-active organisations.

38. Explore the feasibility of the UK Clinical Trials Gateway including experimental medicine trials for rare diseases to provide information for patients and their families about research trials.
Research Commitments

39 Work with the research community, regulators, providers of NHS services and research funders to develop risk-proportional permission systems

40 Encourage researchers to use current guidance to produce generic participant information leaflets and consent forms and participate in future guidance reviews

41 Promote good practice and the use of systems which facilitate a consistent and streamlined process to local NHS permissions of publically, charitably and commercially funded research with an aim to reduce timescales.

46 Work with industry to set priorities and determine how best to support research into rare diseases and promote research collaboration

47 Support initiatives to facilitate engagement between patients, clinical care teams, researchers and industry wherever practical

48 Set out the benefits of collaboration (besides producing specific treatments) for all stakeholders

49 Continue to build a cohesive infrastructure for implementation and coordination of rare disease research in the NHS

50 Encourage major research funders to use current structures to coordinate strategic funding initiatives in rare diseases

51 Improve engagement between key stakeholders, including: patients and relatives, main funding providers, healthcare commissioners, NHS hospitals and specialist care units and industry (pharmaceutical, biotechnology, IT, diagnostics)
Commitment: Patients and public involvement

The NIHR believes that you can’t have high quality research without the involvement of the public. Since 2006, the NIHR has been one of the few Government bodies around the world to fund and support an organisation – INVOLVE – which exists to advance and promote public involvement in research.

On 20 May 2014, the NIHR published a five year strategic plan ‘Promoting a research active nation’ setting out a new programme to encourage public engagement and participation in health, social care and public health research.

The document sets out a range of initiatives including our ‘Ok to ask’ campaign that we will support to grow the number of people taking part in research.

“97% of the public believe it’s important the NHS supports research into new treatments”

Source: Ipsos MORI poll (June 2012) commissioned by the Association of Medical Research Charities, Breast Cancer Campaign and the British Heart Foundation
Commitments: Portals and UK Trials Gateway

Since launch in April 2012, 289,571 unique users have visited UKCTG site and over 12,200 copies of the UKCTG smartphone and tablet app downloaded.
Commitments: Research approvals

- The Government's Plan for Growth announced the launch of the NIHR Research Support Services framework. It is a set of tools and guidelines to support a consistent and streamlined approach to managing health research studies in the NHS.
- Health Research Authority – Assessment and Approval will provide a single approval for research in the NHS incorporating assessments by NHS staff employed by the HRA alongside the independent Research Ethics Committee opinion.
  - The new system will simplify the approvals process for research, making it easier for research studies to be set up.
Commitment 50: Research Coordination

Office for Strategic Coordination of Health Research
National Institute for Health Research

Goals
Develop research in the NHS in order to support:
• outstanding individuals,
• working in world class facilities,
• conducting leading edge research focused on the needs of patients and the public.

Achievements
Since its establishment in April 2006 it has:
• increased the volume of applied health research for the benefit of patients and the public,
• driven faster translation of basic science discoveries into tangible benefits for patients and the economy and,
• developed and supported the people who conduct and contribute to applied health research.
NIHR Health Research System

Faculty
- Investigators & Senior Investigators
- Trainees
- Associates

Infrastructure
- Clinical Research Networks
- Clinical Research Facilities, Centres & Units

Research
- Research Projects & Programmes
- Research Schools

Systems
- Research Management Systems
- Research Information Systems

Universities
- NHS Trusts
- Patients & Public
“...leading-edge research focused on the needs of patients and the public...”
NIHR Very Rare Diseases Themed Call

For diseases affecting less than 1 in 100,000 of the general population.

Six participating NIHR programmes:
- Health Technology Assessment (HTA) Programme
- Efficacy and Mechanism Evaluation (EME) Programme
- Research for Patient Benefit (RfPB) Programme
- Health Services and Delivery Research (HS&DR) Programme
- Invention for Innovation (i4i) Programme
- Programme Grants for Applied Research (PGfAR) Programme

Supported by the National Specialised Commissioning Group and Rare Disease UK.

A total of £5 million has been committed to research through this call, into 7 projects/programmes which cover a wide range of research areas and interventions.
Aim

Strengthen and streamline systems for research management and governance
NIHR Systems

- Integrated Research Application System
- Coordinated system for gaining NHS permission
- UK Regulatory and Governance Advice Service
- Research Passports
- Research governance and management within NIHR Clinical Research Network
- NIHR Research Support Services
- Model Clinical Trials Agreements
- Health Research Authority
- Clinical Practice Research Datalink
Aim

Harness the research potential of the NHS to improve health and deliver competitive advantage for increased economic growth.
We Provide: the support and facilities the NHS needs for first class research that results in high-quality care for patients and the public. All our infrastructure supports collaborations with Industry.

This Allows Us: to harness the research potential of the NHS to improve health and deliver competitive advantage for increased economic growth.

Clinical Research Facilities, Centres & Units

The NIHR funds a range of research centres to provide the facilities the NHS needs to carry out first-class research.

Clinical Research Facilities for Experimental Medicine are purpose-built, cutting-edge facilities, with specialist clinical, research and support staff.

Clinical Research Networks

The NIHR Clinical Research Network makes it possible for all patients and health professionals across England to participate in relevant clinical trials.
Commitment 49 Continue to build a cohesive infrastructure for implementation and coordination of rare disease research in the NHS
Example of NIHR Research
Successful translation for patient benefit:

Duchenne Muscular Dystrophy - a stratified approach

NIHR researchers have collaborated as part of the UK-lead MDEX Consortium to test a novel drug treatment for Duchenne Muscular Dystrophy (DMD).

Results showed that the stratified therapeutic approach developed restored missing dystrophin protein in seven of the 19 children with DMD. In patients given the two highest doses, their levels of functional protein improved to 20 per cent of normal (Cirak et al, 2011). Thirteen per cent of boys with DMD may be suitable for treatment with this novel therapy.

“The support of the NIHR Great Ormond Street Biomedical Research Centre cannot be underestimated. The ability to support this highly intense work in the same location where the therapy is being developed makes the biggest difference to whether a study is successful or not. Staff develop strong relationships with patients, which supports a positive patient experience, and allows us investigators to learn how patients tolerate the experimental therapy.”

Francesco Muntoni, Professor of Paediatric Neurology at University College London Institute of Child Health and chair of the DMD research group
NIHR Rare Diseases Translational Research Collaboration

The NIHR Rare Diseases Translational Research Collaboration (TRC) is an exciting national initiative utilising the unique research infrastructure of the NIHR to study the causes, impacts and treatment of rare diseases.

The programme supports research programmes in a wide range of disease areas and supports work ranging from patient identification and cohort building to “deep phenotyping” and, ultimately, therapeutics development.

With investment of £20 million over four years, the NIHR Rare Diseases TRC will help to increase research collaboration across the NIHR infrastructure.

Provides doctoral and postdoctoral clinical fellowships to those researchers who can demonstrate a role in, or contribution to, improving health, health care or services relating to rare diseases.

Co-chaired by John Bradley (Cambridge BRC) and Patrick Chinnery (Newcastle BRC)
Within the Rare Diseases TRC, there are 14 themes:

<table>
<thead>
<tr>
<th>Rare Disease Theme</th>
<th>Host Organisation</th>
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<tbody>
<tr>
<td>Cancer</td>
<td>NIHR Royal Biomedical Research Centre</td>
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<td>Cardiovascular</td>
<td>NIHR Oxford Biomedical Research Centre</td>
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<tr>
<td>Dementia and Neurodegenerative</td>
<td>NIHR University College London Hospitals Biomedical Centre and Unit</td>
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<td>Eye Disease</td>
<td>NIHR Moorfields Biomedical Research Centre</td>
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<td>Gastrointestinal and hepatology</td>
<td>NIHR Newcastle Biomedical Research Centre</td>
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<td>Immunological Disorders</td>
<td>NIHR Great Ormond Street Biomedical Research Centre</td>
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<td>Metabolism</td>
<td>NIHR Cambridge Biomedical Research Centre</td>
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<tr>
<td>Musculoskeletal</td>
<td>NIHR Oxford Musculoskeletal Biomedical Research Unit</td>
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<tr>
<td>Neuromuscular Disorders</td>
<td>NIHR University College London Hospitals Biomedical Research Centre</td>
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<tr>
<td>Respiratory Disease</td>
<td>NIHR Royal Brompton Respiratory Biomedical Research Unit</td>
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<tr>
<td>Skin</td>
<td>NIHR Guys and St Thomas's Biomedical Research Centre</td>
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<td>Paediatric Cross Cutting Theme</td>
<td>NIHR / Wellcome Trust Birmingham CRF</td>
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<td>Renal</td>
<td>NIHR Cambridge Biomedical Research Centre</td>
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<td>Non-Malignant Haemotology</td>
<td>NIHR Oxford Biomedical Research Centre</td>
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NIHR Rare Diseases Translational Research Collaboration In-Depth Phenotyping Projects

• 22 projects funded
  – total project funding awarded £3,723,171 over 2 years

• 16 projects awarded additional funding for biomarker profiling
  – total funding awarded for biomarker profiling £2,199,248

Commitment 46 Work with industry to set priorities and determine how best to support research into rare diseases and promote research collaboration
The National NIHR Bioresource was launched in March 2014. The NIHR Bioresource is a bank of nearly 75,000 patients, their families and healthy volunteers wishing to take part in early translational research.

Volunteers provide clinical information and a small blood or saliva sample which then allows researchers to contact them for possible research, on the basis of their to be recalled by their genotype and phenotype (their physical characteristics).

Headquartered in Cambridge the NIHR BioResource is being established around six of the NIHR BRCs – Cambridge, Imperial, Guy’s and St Thomas’, South London and Maudsley, Oxford, University College London Hospitals, and the Leicester Cardiovascular Biomedical Research Unit (BRU).
The NIHR BioResource – Rare Diseases has been established to identify genetic causes of rare diseases, improve rates of diagnosis and to enable studies to develop and validate treatments; thus improving care for those with rare diseases and their families.

The NIHR BioResource – Rare Diseases has been awarded funding for the clinical application of Next Generation Sequencing Techniques (NGST) to study the genomes of affected participants and their relatives.

Participants with rare diseases in the following areas are the initial focus of research:

- Infection and Immunity,
- Neuroscience,
- Rare Diseases (including Rare Cancers)
- Cardiovascular Disease.

Commitment 40 Encourage researchers to use current guidance to produce generic participant information leaflets and consent forms and participate in future guidance reviews.
Contacts

Want to find out more about a BioResource centre? We welcome enquiries from volunteers or potential volunteers and the research community. Please contact any of our centre coordinators with your query. Not sure who to contact? Please use the general enquiries email nbr@bioresource.nihr.ac.uk and we shall pass on your query to the appropriate team.

Contact Details

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<tr>
<th>BioResource Centre</th>
<th>Coordinator</th>
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Enrolment office

The NIHR BioResource – Rare Diseases enrolment office provides support to clinical referral centres in the UK to become registered for a Rare Disease study. The enrolment office will ensure that local R&D approval is established for Biomedical Centres/Units and NHS Foundation Trusts, and that a Material Transfer Agreement is put in place if necessary. The enrolment office can also provide financial support for the collection of blood/DNA samples or saliva collection kits.

Please contact Sofie Ashford for information about the services provided by the enrolment office.
MRC-NIHR Phenome Centre

Broad access to a world-class capability in metabolic phenotyping, that will benefit the whole UK translational medicine community

At scale capacity to analyse patient / population-based samples: for biomarker discovery and validation, improved patient stratification through development of robust diagnostic and prognostic markers, early identification of drug efficacy and safety and other responses to treatments
Department of Health capital award of £25m to fund a new robotic bio-sample repository in the south of England. This will enhance England’s capacity to support research into disease mechanisms, diagnosis and treatments, working closely with the NIHR’s experimental medicine infrastructure.
15 Local Clinical Research Networks

North East and North Cumbria
- The Newcastle upon Tyne Hospitals
- NHS Foundation Trust

North West Coast
- Royal Liverpool and Broadgreen University Hospitals
- NHS Foundation Trust

Greater Manchester
- Central Manchester University Hospitals
- NHS Foundation Trust

West Midlands
- The Royal Wolverhampton NHS Trust

West of England
- University Hospitals Bristol
- NHS Foundation Trust

South West Peninsula
- Royal Devon and Exeter
- NHS Foundation Trust

Wessex
- University Hospital
- Southampton NHS Foundation Trust

Yorkshire and Humber
- Sheffield Teaching Hospitals
- NHS Foundation Trust

East Midlands
- University Hospitals of Leicester
- NHS Trust

Eastern
- Norfolk and Norwich University Hospitals
- NHS Foundation Trust

Thames Valley and South Midlands
- Oxford University Hospitals
- NHS Trust

North Thames
- Barts Health NHS Trust

South London
- Guys’ and St Thomas’ NHS Foundation Trust

Kent Surrey and Sussex
- Royal Surrey County Hospital
- NHS Foundation Trust

North West London
- Imperial College Healthcare
- NHS Trust
NIHR Clinical Research Network

Key Achievements:

- **99% of NHS Trusts** participated in CRN Portfolio studies
- **69% of NHS Trusts** participated in CRN commercial Portfolio studies
- **35 Global first patients** and a further 9 European first patients in 2013/14 for multi-centre commercial studies, supported by the NIHR Clinical Research Network.

Additionally, the CRN has a dedicated team working proactively with industry partners to help ensure that the NHS can meet the health research needs of industry.
Genomic technologies: 100,000 whole genomes

• Potential of Genomics, in the form of whole genome and exome sequencing, to transform healthcare

• Life Sciences One Year On announcement

• Unique position of the NHS as a single, national healthcare provider

• Genomics England established

• Wealth creating possibilities
Rare Diseases Research funding and Support

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