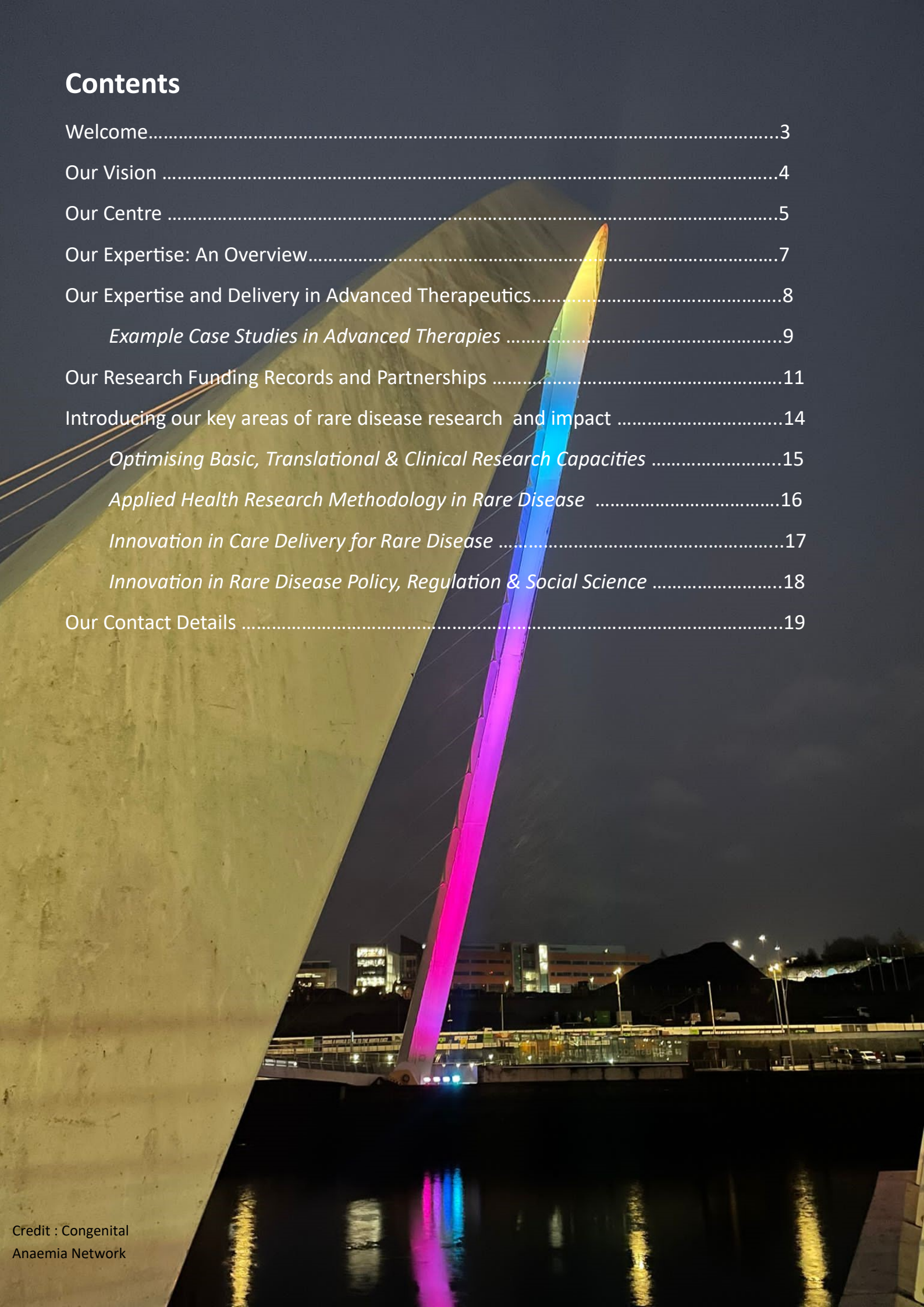


# Newcastle Centre for Rare Disease

A Newcastle University Centre of Research Excellence  
in Collaboration with Newcastle upon Tyne Hospitals &  
Newcastle Health Innovation Partners

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# Welcome to the Newcastle Centre for Rare Disease

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Professor David Jones OBE, Director of NHIP Academy

Director of Newcastle Centre for Rare Disease

Professor of Liver Immunology, Newcastle University

Hon Consultant Hepatologist, Newcastle upon Tyne Hospitals

“Welcome to the Newcastle Centre for Rare Disease. We launched this Centre in 2020, to unite experts, research teams and clinical units working in rare disease (in some cases, for decades). Our philosophy is that by bringing together this diverse expertise and fostering new interdisciplinary collaborations and knowledge-sharing, we ‘make the whole greater than the sum of its parts’.

Why rare disease? Individual rare diseases are, by definition, rare. However, there are an estimated 6-8,000 separate conditions classed as rare, which means that collectively, rare diseases affect a significant proportion of the population, approximately 1 in 18 people. These are typically complex, multisystem conditions, often (though not always) genetically inherited. Only 5-6% have any dedicated treatment. All of this results in multiple challenges for healthcare professionals and health systems at large, but particularly of course for patients and families: very often these conditions have a major impact on multiple aspects of daily life. People living with a rare disease often face inequities in everything from obtaining a diagnosis and accessing the best possible care through to gaining an equal chance in the education, employment and social spheres.

The vision of our Centre, therefore, is to advance research and care for people with a rare disease, going beyond ‘bench-to-bedside’ to support patients and families in living their lives to the fullest. This brochure explains what we do here in Newcastle, what kind of expertise we house, and -we hope!- will encourage you to get in touch with ideas for new collaborations, to help us move forwards together”

Dave Jones, on Behalf of the Newcastle Centre for Rare Disease



# Our Vision

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Newcastle has a long and distinguished history, dating back over 50 years, of both research and clinical care in individual rare diseases. We pioneered clinical care and established world-leading linked research programmes in rare disease areas including neuromuscular disease, immunodeficiency, rare liver disease and mitochondrial disease. Our work has transformed clinical care for patients and is built on a track-record of highly successful collaborations with multiple stakeholder groups.

In 2020 we embarked on the next step in this journey with the launch of the Newcastle Centre for Rare Disease, a Newcastle University Centre of Research Excellence (NUCoRE), bringing together the expertise of researchers in Newcastle University and clinicians in the Newcastle Upon Tyne Hospitals NHS Foundation Trust. Our vision is to unite and share the experience of our 100+ clinicians and researchers across all rare disease areas, providing critical mass for further innovation in our established areas, and to create opportunities for our clinicians, our researchers, our partners and, crucially, our patients. We have a particular focus on productive and highly effective collaborative working with industry. The Centre is a key part of the regional Academic Health Sciences Centre also established in 2020, known as Newcastle Health Innovation Partners.

We welcome the opportunity to work with any and all other universities, hospitals, patient organisations, networks, policymakers, and companies, who share our goal of improving the lives of people living with rare disease.

## Thoughts from the Team



Victoria Hedley, Centre Co-Lead and Rare Disease Policy Manager

*“Each rare disease patient faces their own personal challenges. But there are often commonalities between conditions, both in patient experience and approaches to improving treatment. We established the Newcastle Centre for Rare Disease to give us “beginning to end” capability across multiple diverse disease areas. We focus on innovation, spanning diagnostics to advanced therapies, basic science to clinical delivery, social science to policy.”*

## Our Centre

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The **Newcastle Centre for Rare Disease** was established to strengthen our already-substantial reputation as a seat of multidisciplinary knowledge and expertise in rare diseases. The overarching goal of this Centre is to build synergies between disease areas and cross-cutting areas of methodological expertise, share best practices, and develop new resources to span the discovery and translational pipeline and build on our existing expertise in the discovery, diagnostics, treatment and care of numerous diseases/groups of disorders.

One key milestone in the development of Newcastle as a centre of excellence in rare disease was our unparalleled role in the launch of the European Reference Networks (ERNs: Europe-wide networks of excellence in the clinical management and research of rare disease and highly specialised healthcare).

We coordinated and led 3 ERNs (which was more than any other centre in Europe) for the following:

- Immunodeficiency, Auto-inflammatory & Autoimmune Diseases
- Liver Diseases
- Neuromuscular Diseases

We also participated as full members of an additional 3 ERNs:

- Rare bone
- Rare pulmonary
- Rare renal diseases

Following the UK's exit from the EU, our experts retain these unique insights into building and evolving diverse networks in care and research. In fact, Newcastle was a driving force behind the conceptualisation and implementation of *all* ERNs, which formed a cornerstone of our international rare disease policy portfolio over the past decade. Predating the emergence of ERNs, in 2007 Newcastle University launched the (now-global) TREAT-NMD network, seeking to accelerate translational research in neuromuscular diseases. **Newcastle is thus uniquely placed to inform and lead national and international activities concerning the designation of expert centres and the creation of rare disease networks.**



## Our Centre

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A key factor in Newcastle's achievements in the ERN story, and in our success in the area of rare disease generally, is our **long tradition of translating outstanding underpinning science into novel therapies and integrated care approaches, which are evaluated and then implemented into practice via cutting-edge clinical services**. Comparator centres typically focus on the pure research opportunity and may overlook the application, implementation and clinical care aspects that make Newcastle distinctive.

The launch of the **Newcastle Centre for Rare Disease** is enabling the identification of *new* synergies between groups which may traditionally have worked in silos, and is using these to foster high-impact research. A further strength of the Centre, however, is its ability to provide the **multidisciplinary** focus necessary to improve outcomes and quality of life for people with rare diseases (ca. 3.5 million in the UK alone). We therefore also prioritise themes such as innovation in rare disease policy and regulation (exploiting our solid international policy reputation); applied health research methodologies (including HTA considerations and the incorporation of patient reported outcomes and 'real world data' to drive research and care delivery); integrated and holistic models of care delivery ; and ethical, legal and social issues (ELSI), to support a holistic, person-centred approach. These activities are bolstered through the unique expertise available at Newcastle University in domains such as policy-making, networking, research infrastructures, industry engagement, ELSI, data management, and HTA. For instance, a strategic link to the NIHR is ensured through the former NIHR Academy Dean (for faculty Trainees) and Theme Lead for the former NIHR Translational Research Collaboration in Rare Diseases, and through NIHR Professorships.

Furthermore, Newcastle is one of 3 UK partners in the European Joint Programme for Rare Disease Research and in 2022 launched the UK-International 'Mirror and Action' Group for Rare Disease research, RD-IMAG, to connect UK researchers and research infrastructures with European and International experts, tools and platforms.

The **Centre for Rare Disease** is also developing plans to enhance the broader educational landscape at Newcastle University, promoting an ethos of excellent research-led teaching at Undergraduate and Postgraduate level, whilst fostering a pipeline of independent researchers able to increase research power in future and develop new external collaborations.



### Thoughts from the Team

Professor Brain Walker PVC for Research Strategy and Resources

*"Our Centres of Research Excellence exploit Newcastle University's physical and social cohesion. They make it easy for us to operate as 'One University' to deliver multidisciplinary challenge-led research."*



# Our Expertise: An Overview

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Integral to our model is our capacity to offer expertise both across **rare disease clinical areas** and across the **technologies and methodologies** that are critical to delivering advances in rare disease.

We are one of the leading clinical centres in Europe and offer ‘across the board’ clinical services. We have extensive experience in the development of comprehensive clinical cohorts. **If you are interested in working with us, and the disease area you are interested in is not included in this list, please contact us to discuss your needs.** We are very happy to explore new disease areas and to develop opportunities with existing and potential partners. You can find out more about our collaborations here — <https://www.ncl.ac.uk/rare-diseases/our-collaborations/>



## Our Expertise and Delivery in Advanced Therapeutics

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Advanced Therapeutics represents an area of real strength for the **Newcastle Centre for Rare Diseases** and is one where we are keen to develop further and deeper partnerships. There has been significant investment by the University and Trust in this area, and ambitious plans are being proposed to invest more.

We have substantial experience in all forms of advanced therapeutics including gene therapy, cell-based therapeutics, organ-transplantation, immunotherapy small molecule therapeutics and complex interventions. We have a key partnership with **Therapeutics North-East**, a cross organisation structure focusing on advanced therapies in both common and rare disease. We have particular strength in **gene and cell therapy**.

A particular focus for our **gene therapy** research is rare neuromuscular diseases. For almost a decade, the team at the *John Walton Muscular Dystrophy Research Centre (JWMDRC)* has positioned itself as a global leader by successfully contributing to trials of advanced therapies for several primary muscle diseases and for spinal muscular atrophy (SMA). These trials have led to treatments for conditions previously considered untreatable (Translarna<sup>®</sup>, Eteplirsen<sup>®</sup>, NaMscla<sup>®</sup>), providing the most significant improvement in healthcare for patients and families that the neuromuscular field has ever experienced. FDA and EMA have now approved nusinersin (Spinraza<sup>®</sup>), an intrathecally injected antisense oligonucleotide to treat SMA types 1, 2 and 3, and Onasemnogene abeparvovec (Zolgensma<sup>®</sup>) an adeno-associated virus (AAV) based gene replacement therapy to treat SMA type 1 patients. London and Newcastle are the only clinical trial sites for both of these products.

In the area of **cell therapy** we have a key partnership with The Northern Alliance Advanced Therapies Treatment Centre (Innovate UK) co-led by Newcastle, which has cemented Newcastle as a leading centre for advanced therapy clinical translation, trials delivery and NHS adoption, with a stand-out record in CAR-T trials and as a supra-regional clinical service. Infrastructure is in place for Newcastle to lead in AAV gene therapy delivery for neuromuscular diseases and haemophilia.

Our advanced therapy programme has a substantial portfolio of industry partnerships with 15 industrial partners in the area of cell and gene therapy alone. We had 8 gene and cell therapy trials starting in 2021.

### Thoughts from the Team



Professor Volker Straub, Deputy Dean-NU Translational & Clinical Institute, and Director of the John Walton Muscular Dystrophy Research Centre

*“Our goal is to advance research strategies and infrastructure to boost discovery science. We place an emphasis on supporting the translation of promising research into trials and onwards into clinic”*



## Example Case Studies in Advanced Therapies

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Examples of our delivery in the area of rare diseases include the following (with a particular focus on advanced therapy relevant to rare disease)

**Neuromuscular Disease:** The JWMDRC limb girdle muscular dystrophy (LGMD) natural history study catalysed the first ever phase 3 clinical trial using viral based (AAV) systemic gene replacement therapy for a subset of LGMDs. The Newcastle team is involved in design and implementation, from first in man studies to registration trials and, based on international collaborations, additional compounds that target dystrophin expression by exon skipping have now been approved.

**Haematopoietic stem cell transplantation:** Across both adult and paediatric patients HSCT has increased 3 fold since 2014, treating over 250 patients annually. This work includes sophisticated *ex vivo* manipulation of products, such as T-cell depletion in patients with primary immunodeficiencies. Also the adoptive transfer of multivirus specific T-cells to boost immunity following allogeneic transplant.

**Tolerogenic Immunotherapy:** We have made significant advances in developing our tolerogenic immune therapy, autologous tolerogenic dendritic cells (tolDC). Our phase 1 trial demonstrated safety in patients with inflammatory arthritis. Versus Arthritis have funded a follow-on study comparing different routes of administration, using MRI tracking alongside immune monitoring as a surrogate efficacy marker. Collaborations include the Karolinska Institute and University of Nijmegen. We co-founded the Cooperation in Science & Technology (COST) EU tolerogenic cell therapy network, Action to Foster and Accelerate Cellular Tolerogenic Therapies (AFACTT).

**Lung Transplantation:** We reported the outcome of the NIHR HTA DEVELOP-UK study of donor ex-vivo lung perfusion in UK lung Transplantation. This was world's first non-commercial multi-centre study of EVLP and demonstrated the technique when applied to higher risk marginal donor organs could facilitate more lung transplants reducing waiting time for transplant but at a significant economic cost due to the cost of the EVLP procedure but also longer ITU stays and a higher risk of ECMO use post-transplant.



# Example Case Studies in Advanced Therapies

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**Kidney Transplantation:** First report of the potential use of a cell based therapeutic to treat human kidneys on an *ex-situ* normothermic circuit. This study performed as an industrial collaboration with Athersys showed improvements in function of the discarded kidney and dampened inflammation.

**$\beta$ -cell replacement therapy:** Newcastle led the commissioning of the world's first supra-regional islet transplant service and generated pilot data for subsequent commissioning of a national islet auto-transplant programme. The Institute of Transplantation uniquely delivers all modalities of solid organ transplantation in addition to islet allo/auto-transplantation. We are partnering with industry and academic collaborators to evaluate emergent technologies in  $\beta$ -cell replacement. This includes a commercial research contract with Betalin Therapeutics towards further development and clinical trial of their Engineered Micro-Pancreas technology.

**Biomarker Discovery:** Identification of interleukin-1b as a potential biomarker of donor lung suitability during EVLP which showed excellent sensitivity and specificity to predict 1 year survival after lung transplantation. Worked with MyCartis to develop a rapid point of care test for Interleukin-1b to guide decision making on donor suitability.

**Eye research:** Developed, implemented and validated a novel Limbal Stem Cell therapy from bench to bedside. Funded by an ERC Consolidator Fellowship we have made extensive use of iPSC-disease modelling to identify global spliceosome dysregulation and gelsolin dysfunction in inherited Retinitis Pigmentosa caused by mutations in pre-mRNA splicing factor *PRPF31* and *RPGR*. We were the first to report generation of light sensitive retinal organoids from human pluripotent stem cells.

**Pluripotent stem cells:** We co-lead the IMI1 StemBANCC initiative on derivation and characterisation of iPSC from 500 patients with neurodegenerative disease resulting in successful consortium publications and creation of the European induced pluripotent stem cell bank (EbiSC).

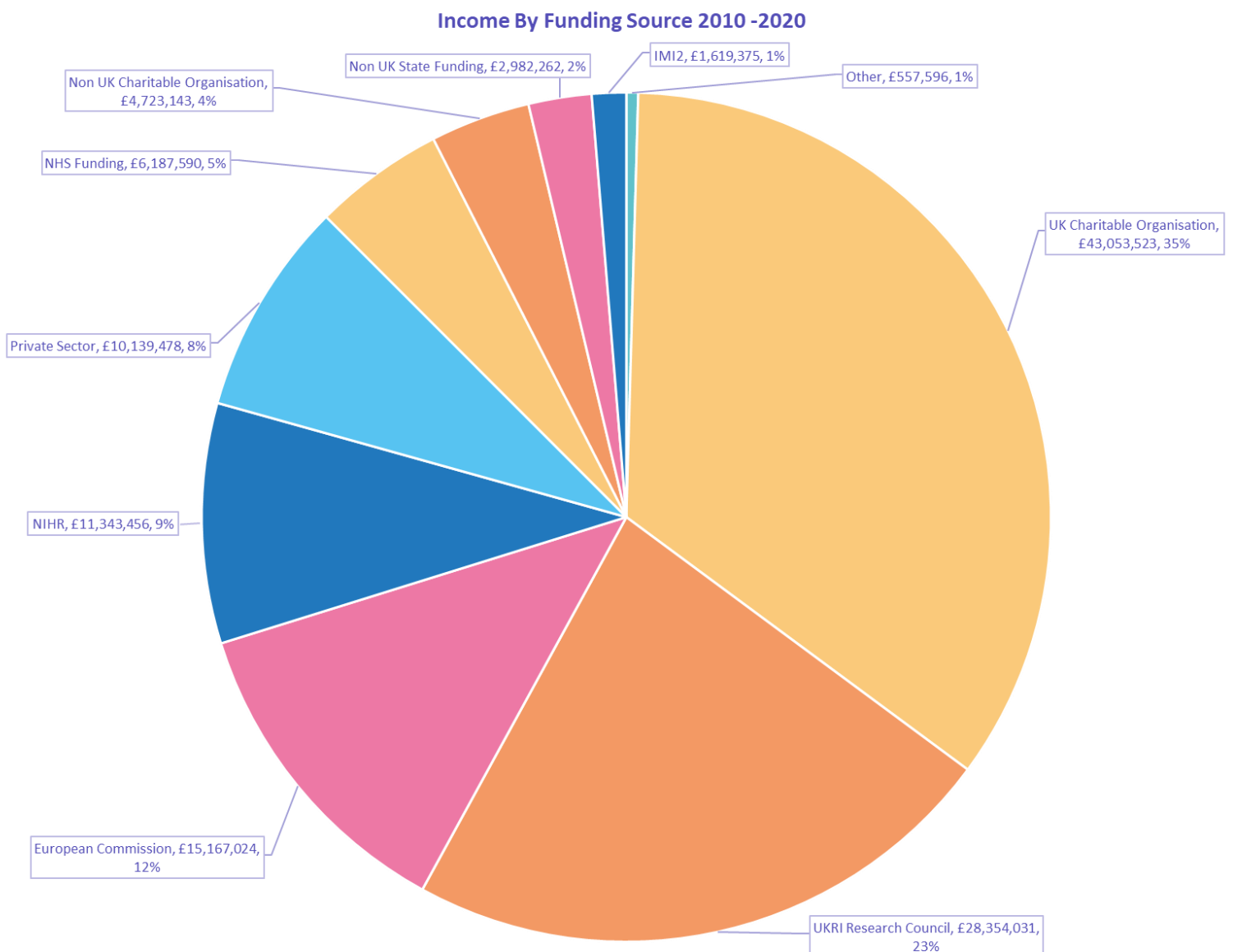


Image courtesy of the Bubble Foundation

# Our Research Funding Records and Partnerships

Cumulatively, the members of our **Newcastle Centre for Rare Disease** secured over £122 Million of rare disease-related funding over the period 2010-2020 (that is, in excess of £122 Million actually coming to Newcastle – the total funds for grants in which Newcastle experts were involved are of course far greater). Grants and funding range from relatively small sums which nonetheless address key topics of relevance to research and patient impact, up to grants of £5+ Million.

The greatest portion of overall funding over the past decade actually came from charitable organisations, mainly UK based. These figures (which actually probably represent a slight underestimation of the full picture) emphasise the **success of this group in developing and delivering patient-centred research**, deemed very valuable to charities and foundations in the rare disease sphere, in particular. Almost a quarter of our combined funding (ca. £28 million) stemmed from UKRI Councils, especially the MRC (which accounts for 92% of that category).





# Our Research Funding Records and Partnerships

A particular hallmark of our research track record is the success in fostering collaborations and partnerships with a wide spectrum of commercial organisations internationally, who share our vision of the delivery of better care for patients with rare diseases. Certain groups within this Centre (including Neuromuscular, Liver, Immunodeficiencies/Immunological and Fibrosis) already have well-established collaborations with the Private Sector, in view of their access to well-stratified patient cohorts and unique knowledge in translating potential therapies for rare diseases into proof of concept and first-in-human trials, and from there to the clinic. In fact, in the period 2019-2020, 91 of 274 active studies (constituting 33%) under the NIHR Newcastle Clinical Research Facility were in rare diseases:

Early Phase Rare Disease Trials in Newcastle	
Rare Disease Area	Number of active studies (2019-2020)
Neuromuscular & Mitochondrial	49
Respiratory	16
Neurodegenerative	10
Liver	8
Musculoskeletal	6
Others	3
Total	91 (33% of 274 active studies)

In 2020 alone, rare diseases accounted for 68% of the Faculty of Medical Science's Investigator Initiated Trials (IITs), attracting a revenue of just over £4.5M from industry. Rare diseases accounted for 50% of the monies awarded for Commercial Research (£500k) and a further 33% of consultancy funds (£495k). We currently work with over 30 industrial partners across the world, covered by confidentiality agreements



# Our Research Funding Records and Partnerships

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To complement the broad array of research funders, we collaborate closely with many key initiatives, both nationally and at international level:

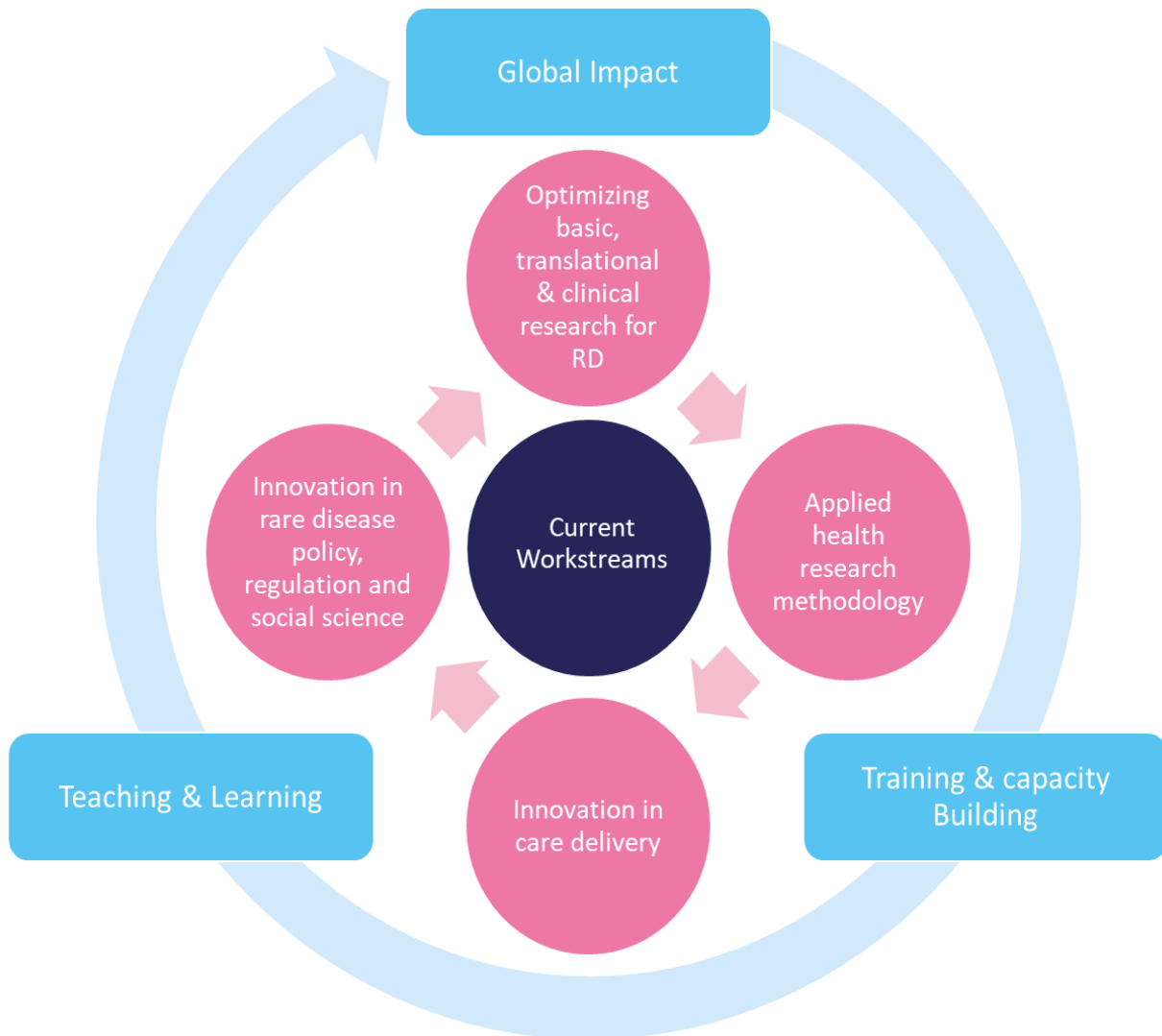
- UK Department of Health and Social Care (Genomics and Rare Diseases team)
- UK Rare Disease Policy Group
- NIHR
- Genomics England
- EURORDIS – Rare Diseases Europe
- IRDiRC – the International Rare Disease Research Consortium
- GA4GH – the Global Alliance 4 Genomics and Health
- ESHG – European Society of Human Genetics
- European Commission Directorates General (especially RTD and SANTE, focusing on the Units for Rare Diseases, Cross-Border Healthcare and eHealth)
- EJP-RD (European Joint Programme for Rare Disease Research)
- Rare Diseases International
- NGO Committee for Rare Diseases at the UN
- NICE
- NHS England
- NHS Blood and Transplant
- ELIXIR UK
- FindacureUK/Beacon
- Genetic Alliance UK
- RDUK – Rare Diseases UK



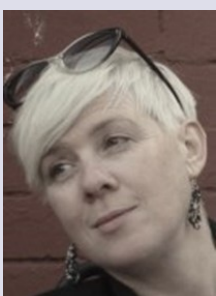
Credit Chris Bishop

# Introducing our key areas of rare disease research and impact

Our structure follows the flow of research from innovation in fundamental research all the way through to the adoption of innovative treatment approaches into practice and national policy change. Our work is loosely organised into 4 inter-linked workstreams with an organisational culture that ensures connectivity. All themes, in addition to progressing their research across all our target diseases, have a major focus on optimising the global impact of the work of the centre, improving education for future clinicians and researchers, and training the next generation.



## Thoughts from the Team



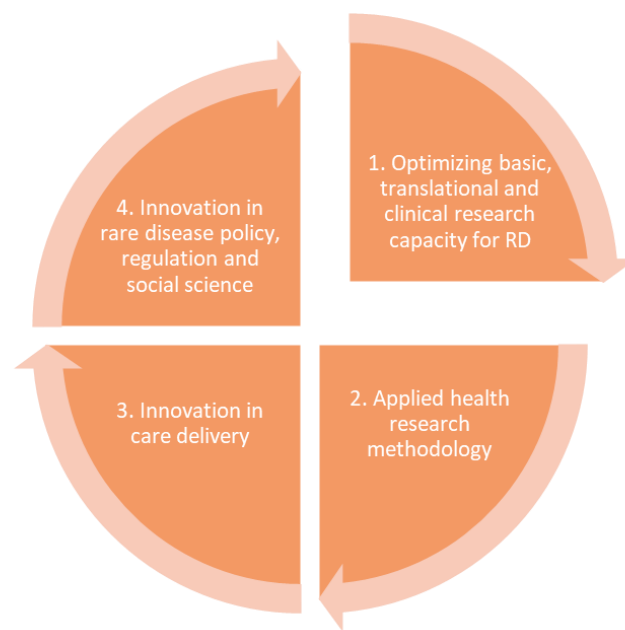
Dr Pauline McCormack Senior Lecturer

*"We foster a dialogue between researchers and patients to encourage trust and confidence. In this way, we can be certain that our research is relevant and needed by the patient community. And that it is carried out with integrity"*



# Optimising Basic, Translational & Clinical Research Capacities

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This workstream is all about creating new opportunities and building capacity around basic, clinical, applied and translational research. One important means of achieving this is exploring approaches or discoveries being advanced by a particular disease/disease area which could be relevant to others. Expertise in areas such as bioengineering, Diagnostics, Geo-Epidemiology & Modelling, and next generation AI, is incorporated. This vast workstream also addresses key questions of how to make rare disease communities 'trial-ready'. Activities include the following:

- Exploring basic and preclinical approaches which could be shared from one disease area to another, and researching pathways or processes common to multiple diseases
- Optimising diagnostics for rare disease, including the pioneering of -omics approaches
- Identifying, optimising and sharing transversal expertise to develop better ways of advancing research, understanding pathogenesis, and identifying therapeutic options for specific genotypic or phenotypic populations
- Generating a shared 'toolkit' of resources which could be applied across domains, by pooling existing (and developing new) assets designed to advance research in cross-disease domains
- Optimising approaches and frameworks for personalised and precision medicine;
- Elucidating the potential for RD to serve as paradigms and provide insights to more common diseases
- Designing more patient-centric research and building effective patient partnerships in areas where these are currently lacking
- Leveraging drug repurposing expertise



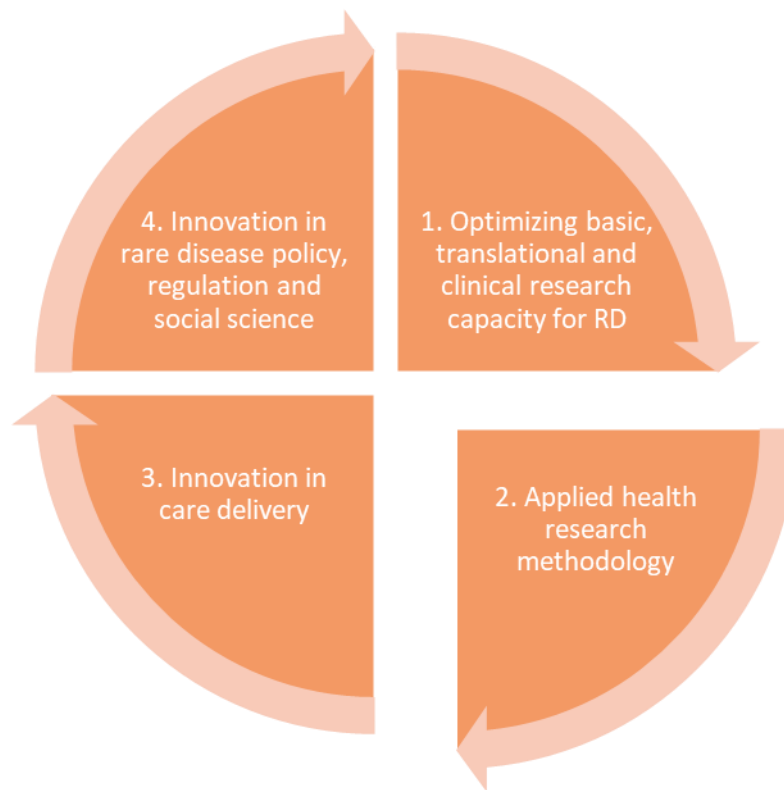
## Thoughts from the Team

Professor Joris Veltman, Dean of Biosciences Institute

*"Gaining insights of the pathogenesis of a rare disease opens up possibilities for actually diagnosing patients with that condition. We have an excellent track-record of using genomics for diagnosis"*

# Applied Health Research Methodology in Rare Disease

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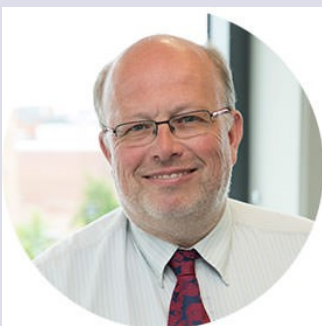


A major part of this area of work concerns the planning and delivery of innovative and more appropriate clinical research for rare diseases, exploring and actively testing new approaches to trial design and the evaluation of specialised interventions. The unique expertise of Professor of Biostatistics James Wason – recipient of a 2021 NIHR Professorship- is central to this research area, which involves colleagues from Newcastle’s Clinical Trials Unit, Biostatistics Research Group, the School of Natural and Environmental Sciences, and more. Our research here includes:

1. Advancing/demonstrating the value of novel trial methodologies and new statistical methods to support innovative trial design in small populations
2. Looking at “real-world” data-driven approaches
3. Incorporation of patient reported outcomes to clinical and health-related research
4. Generating data to demonstrate therapy and treatment regime effectiveness and value, including the post marketing setting.

**One key goal of our activity in this area concerns the development of an early phase clinical trials “hub”, able to support innovative early phase trials to inform larger scale novel methodology trials**

## Thoughts from the Team

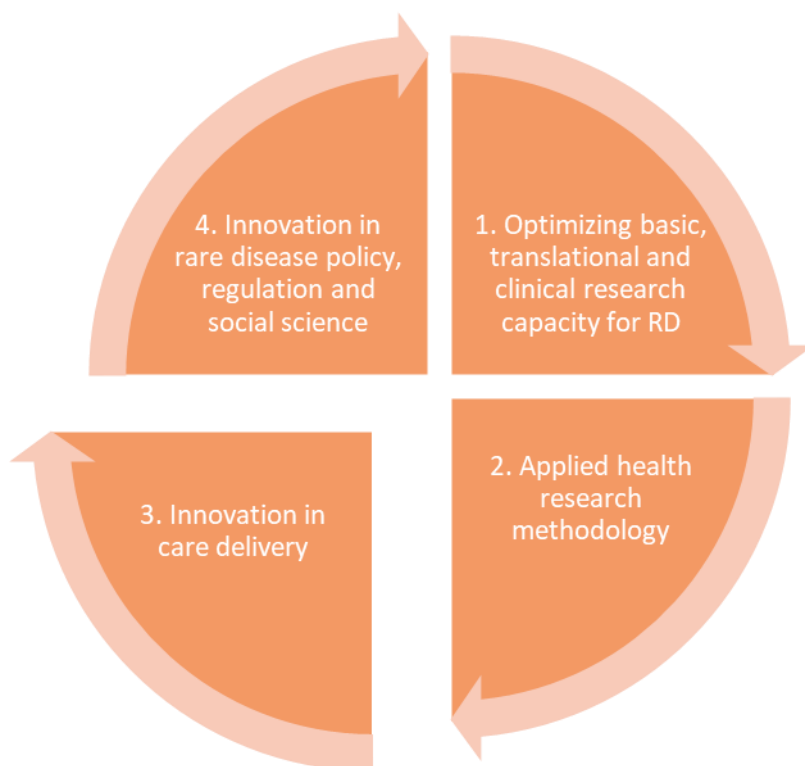


Professor Dave Jones Director of Newcastle Centre for Rare Disease

*“Our experts promote innovative methodological approaches to clinical trial design and delivery. These include basket, platform and umbrella trials. We also go beyond this, to test and refine these methodologies through real-life clinical research.”*

# Innovation in Care Delivery for Rare Disease

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This area of expertise focuses on the particular importance of integrated and multidisciplinary care for people living with a rare disease: we interpret ‘integrated and multidisciplinary’ very broadly, meaning clinics providing access to the appropriate mix of specialists from different disciplines, but also ensuring a more joined-up approach to care incorporating the social sphere. The goal is to establish ‘what works’ for rare diseases, at the cross-disease and disease-specific (and wherever possible also *person*-specific) levels, and to optimise care delivery accordingly. We believe that rare disease treatment and care involves far more than delivering therapies alone (which after all are non-existent for 95% of rare conditions). We are working to identify good practices employed across our most established clinical areas and explore their relevance and expansion to other conditions, optimising care for our diverse rare disease populations. This includes a focus on the following:

- Ensuring a multidisciplinary approach to care and identifying ‘what works’
- Bridging the gap between health and social care, and supporting a holistic approach to patient health and wellbeing
- Generating clinical practice guidelines/standards of care to encapsulate good practices
- Exploring cost-effective approaches to specialised care delivery
- Exploring models of ideal virtual care delivery

## Thoughts from the Team

Dr. Michael Wright, Deputy Medical Director Newcastle Hospitals

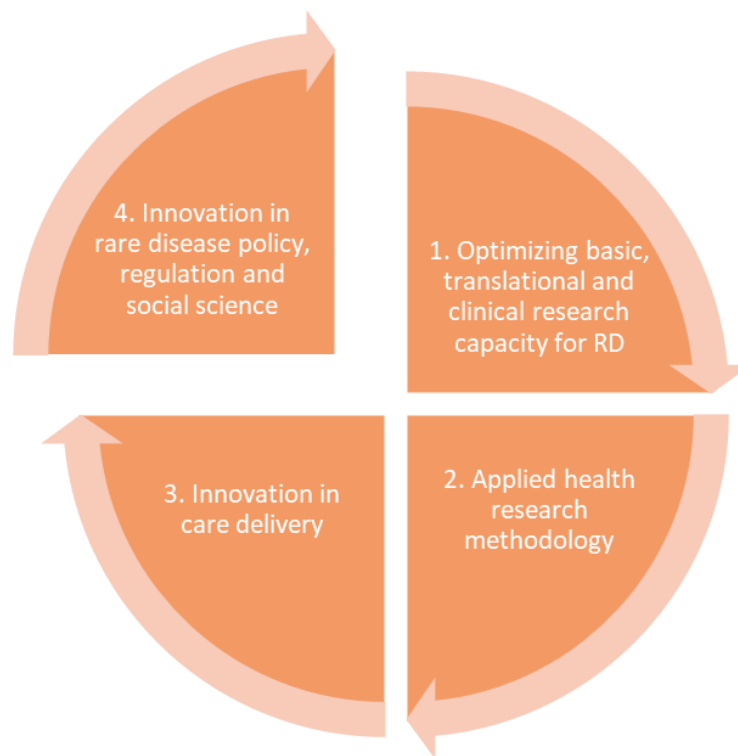
*“Our Centre benefits from the direct membership of over 45 clinical academics and healthcare professionals delivering expert clinical services for rare disease patients at a regional, national and international level. We embrace an innovative and integrated 360° approach to care”*





# Innovation in Rare Disease Policy, Regulation & Social Science

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This area of work sustains and expands our unique role in policy-making for rare disease, at the national, European and global levels. Our particular expertise in rare disease Ethical, Legal and Social Issues (ELSI) will support the development of new sociological research to better understand the day-to-day impact of different rare diseases, exploring the lived experience of patients and families. The particular challenges posed by rare diseases in terms of regulatory science are very much in scope. In addition, this very multidisciplinary workstream is seeking to launch new projects or installations to represent rare disease in the arts and humanities. Specific activities include the following:

- Generating or changing policy, informed by Newcastle research outputs (a huge range of topics are eligible here, as 'policy' in RD covers essentially everything: networking, organisation of highly specialised care, registries, screening and prevention, research, codification data standards and engagement with the regulators and NICE, and much more).
- Continuing to influence European and International policy-making at the pan-rare-diseases level, by supporting the implementation of the Rare 2030 recommendations we co-authored, establishing and co-leading a European Stakeholder Network for Rare Diseases, co-designing a model for a future WHO Global Network for Rare Disease, and much more
- Engaging more proactively with strategic UK RD policy-making; including by launching and coordinating a UK-International Mirror and Action Group for RD Research (RD-IMAG)
- Stimulating new research in the social sphere, including new ELSI research, research to assess the socio-economic and hidden psychosocial burden of rare diseases, research with an emphasis on inequality and the vulnerability of rare disease populations; and more. (And, through the preceding *Innovation in Care* workstream, seeking to influence local, regional, national or international practice or behaviours according to the findings of such research)
- Exploring the portrayal of rare disease in the arts; for instance, how the creative arts are used to express the lived experience of a person with a rare disease; considering the historical perception of rare diseases; and media perceptions and presentations of living with a rare disease.

## Our Contact Details

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To find out more about us please visit our websites:

<https://www.ncl.ac.uk/research/rare-diseases/>

<https://www.newcastlehealthinnovation.org/research-clusters/rare-diseases/>

If you are interested in collaborating with us, please get in touch for a chat through:

[David.Jones@ncl.ac.uk](mailto:David.Jones@ncl.ac.uk) or [Victoria.Hedley@ncl.ac.uk](mailto:Victoria.Hedley@ncl.ac.uk)

You can follow us on Twitter : @NCL\_RareDisease

“Working together for patient innovation: our vision is to replace ‘bench to bedside’ with ‘bench to *outside*, enabling people with a rare disease to live life to the fullest”

